

GENE LIST DG-4.2.0 (5396 GENES)

Releasedate: 24-07-2025

Gene	TWIST X2 covered >10x	TWIST X2 covered >20x	srWGS GRCh38 covered >10x	srWGS GRCh38 covered >20x	Associated Phenotype description and OMIM disease ID	Genepanel
A2M	100%	100%	100%	99.6%		HEMOSTATIC/THROMBOTIC DISORDERS PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS
A4GALT	100%	100%	100%	98.4%	[Blood group, P1Pk system, P(2) phenotype], 111400;NOR polyagglutination syndrome, 111400;[Blood group, P1Pk system, p phenotype], 111400	MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS

AAAS	100%	100%	100%	99.1%	Achalasia-addisonianism-alacrimia syndrome, 231550	SKIN DISORDERS PANEL ¹ DISORDERS/DIFFERENCES OF SEX DEVELOPMENT (DSD) / PRIMARY ADRENAL INSUFFICIENCY PANEL POLYNEUROPATHIES PANEL ¹ INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹
AAGAB	100%	100%	100%	99.2%	Keratoderma, palmoplantar, punctate type IA, 148600	SKIN DISORDERS PANEL ¹ MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS

AARS1	100%	100%	100%	99.2%	Developmental and epileptic encephalopathy 29, 616339;Charcot-Marie-Tooth disease, axonal, type 2N, 613287;?Leukoencephalopathy, hereditary diffuse, with spheroids 2, 619661;Trichothiodystrophy 8, nonphotosensitive, 619691	SKIN DISORDERS PANEL ¹ COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹ EPILEPSY PANEL POLYNEUROPATHIES PANEL ¹ INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS
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AARS2	100%	100%	100%	98.6%	Leukoencephalopathy, progressive, with ovarian failure, 615889;Combined oxidative phosphorylation deficiency 8, 614096	MOVEMENT DISORDERS PANEL DISORDERS/DIFFERENCES OF SEX DEVELOPMENT (DSD) / PRIMARY ADRENAL INSUFFICIENCY PANEL HEART DISORDERS PANEL ¹ MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS MITOCHONDRIAL DISORDERS PANEL COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹ PREMATURE OVARIAN INSUFFICIENCY PANEL
AASS	100%	100%	100%	99.5%	Hyperlysinemia, 238700	METABOLIC DISORDERS PANEL INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹

ABAT	100%	100%	100%	99.2%	GABA-transaminase deficiency, 613163	EPILEPSY PANEL METABOLIC DISORDERS PANEL INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS MITOCHONDRIAL DISORDERS PANEL COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹
ABCA1	100%	100%	100%	99.4%	Tangier disease, 205400;HDL deficiency, familial, 1, 604091	POLYNEUROPATHIES PANEL ¹ MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹

ABCA12	100%	100%	99.9%	98.8%	Ichthyosis, congenital, autosomal recessive 4B (harlequin), 242500;Ichthyosis, congenital, autosomal recessive 4A, 601277	SKIN DISORDERS PANEL ¹ MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹
ABCA2	100%	99.7%	99.9%	96.6%	Intellectual developmental disorder with poor growth and with or without seizures or ataxia, 618808	INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS
ABCA3	100%	100%	100%	99.3%	Surfactant metabolism dysfunction, pulmonary, 3, 610921	MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹

ABCA4	100%	100%	100%	99.1%	Retinal dystrophy, early-onset severe, 248200;Retinitis pigmentosa 19, 601718;{Macular degeneration, age-related, 2}, 153800;Cone-rod dystrophy 3, 604116;Fundus flavimaculatus, 248200;Stargardt disease 1, 248200	VISION DISORDERS PANEL COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹ MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS
ABCA5	100%	100%	100%	99.5%	?Hypertrichosis, congenital generalized, with gingival hyperplasia, 135400	MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS
ABCB1	100%	100%	100%	99.7%	Encephalopathy, acute transient, 620950;{Inflammatory bowel disease 13}, 612244;{Colchicine resistance}, 120080	MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹
ABCB11	100%	100%	100%	99.5%	Cholestasis, benign recurrent intrahepatic, 2, 605479;Cholestasis, progressive familial intrahepatic 2, 601847	LIVER DISORDERS PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹

ABCB4	100%	100%	100%	99.5%	Gallbladder disease 1, 600803;Cholestasis, intrahepatic, of pregnancy, 3, 614972;Cholestasis, progressive familial intrahepatic 3, 602347	LIVER DISORDERS PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹
ABCB6	100%	100%	100%	98.9%	Dyschromatosis universalis hereditaria 3, 615402;[Blood group, Langereis system], 111600;Pseudohyperkalemia, familial, 2, due to red cell leak, 609153;Microphthalmia/coloboma 7, 614497	VISION DISORDERS PANEL SKIN DISORDERS PANEL ¹ MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS

ABCB7	100%	99.9%	99%	69.4%	Anemia, sideroblastic, with ataxia, 301310	MOVEMENT DISORDERS PANEL INHERITED BONE MARROW FAILURE AND/OR PREDISPOSITION PANEL TO HEMATOLOGICAL MALIGNANCIES IRON DISORDERS PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS MITOCHONDRIAL DISORDERS PANEL
ABCC1	100%	100%	100%	98.6%	?Deafness, autosomal dominant 77, 618915	HEARING IMPAIRMENT PANEL (INCLUDING GJB2) MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS

ABCC2	100%	100%	100%	99.2%	Dubin-Johnson syndrome, 237500	LIVER DISORDERS PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹
ABCC6	98.4%	98.4%	100%	98.9%	Pseudoxanthoma elasticum, 264800;Arterial calcification, generalized, of infancy, 2, 614473;Pseudoxanthoma elasticum, forme fruste, 177850	VISION DISORDERS PANEL SKIN DISORDERS PANEL ¹ HEART DISORDERS PANEL ¹ MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹

ABCC8	100%	100%	100%	99%	Diabetes mellitus, permanent neonatal 3, with or without neurologic features, 618857;Diabetes mellitus, transient neonatal 2, 610374;Diabetes mellitus, noninsulin-dependent, 125853;Hypoglycemia of infancy, leucine-sensitive, 240800;Hyperinsulinemic hypoglycemia, familial, 1, 256450	METABOLIC DISORDERS PANEL INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS EPILEPSY PANEL COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL¹
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ABCC9	96%	96%	100%	99.3%	Cardiomyopathy, dilated, 1O, 608569;Hypertrichotic osteochondrodysplasia (Cantu syndrome), 239850;?Atrial fibrillation, familial, 12, 614050;Intellectual disability and myopathy syndrome, 619719	SKIN DISORDERS PANEL ¹ TALL STATURE PANEL WITH GENOME WIDE CNV ANALYSIS LYMPHATIC ANOMALIES PANEL WITH GENOME WIDE CNV ANALYSIS INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS HEART DISORDERS PANEL ¹ SHORT STATURE/SKELETAL DYSPLASIA PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹
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ABCD1	100%	99.1%	98.8%	68.9%	Adrenoleukodystrophy, 300100;Adrenomyeloneuropathy, adult, 300100	MOVEMENT DISORDERS PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS METABOLIC DISORDERS PANEL INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS VISION DISORDERS PANEL DISORDERS/DIFFEREN CES OF SEX DEVELOPMENT (DSD) / PRIMARY ADRENAL INSUFFICIENCY PANEL POLYNEUROPATHIES PANEL ¹
ABCD2	100%	100%	100%	99.9%		METABOLIC DISORDERS PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS

ABCD3	100%	100%	100%	99.7%	?Bile acid synthesis defect, congenital, 5, 616278	MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS LIVER DISORDERS PANEL METABOLIC DISORDERS PANEL
ABCD4	100%	100%	100%	98.9%	Methylmalonic aciduria and homocystinuria, cblJ type, 614857	INHERITED BONE MARROW FAILURE AND/OR PREDISPOSITION PANEL TO HEMATOLOGICAL MALIGNANCIES METABOLIC DISORDERS PANEL INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹

ABCG5	100%	100%	100%	99.3%	Sitosterolemia 2, 618666	HEMOSTATIC/THROMBOTIC DISORDERS PANEL METABOLIC DISORDERS PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹
ABCG8	100%	100%	100%	99.3%	Sitosterolemia 1, 210250;{Gallbladder disease 4}, 611465	HEMOSTATIC/THROMBOTIC DISORDERS PANEL METABOLIC DISORDERS PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹

ABHD12	100%	100%	100%	98.8%	Polyneuropathy, hearing loss, ataxia, retinitis pigmentosa, and cataract, 612674	MOVEMENT DISORDERS PANEL VISION DISORDERS PANEL POLYNEUROPATHIES PANEL ¹ METABOLIC DISORDERS PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹ HEARING IMPAIRMENT PANEL (INCLUDING GJB2)
ABHD16A	100%	100%	100%	98.7%	Spastic paraplegia 86, autosomal recessive, 619735	MOVEMENT DISORDERS PANEL INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹

ABHD5	100%	100%	100%	99.7%	Chanarin-Dorfman syndrome, 275630	SKIN DISORDERS PANEL ¹ METABOLIC DISORDERS PANEL INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹
ABL1	100%	100%	100%	99.2%	Leukemia, Philadelphia chromosome-positive, resistant to imatinib, 608232;Congenital heart defects and skeletal malformations syndrome, 617602	THORACIC AORTIC ANEURYSM AND/OR DISSECTION PANEL CONGENITAL HEARTDISEASE PANEL ¹ HEART DISORDERS PANEL ¹ MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS

ACACA	100%	100%	100%	99.3%	Acetyl-CoA carboxylase deficiency, 613933	METABOLIC DISORDERS PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹
ACAD8	100%	100%	100%	99%	Isobutyryl-CoA dehydrogenase deficiency, 611283	HEART DISORDERS PANEL ¹ METABOLIC DISORDERS PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹

ACAD9	100%	100%	100%	99.2%	Mitochondrial complex I deficiency, nuclear type 20, 611126	HEART DISORDERS PANEL ¹ METABOLIC DISORDERS PANEL INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS MITOCHONDRIAL DISORDERS PANEL COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹
ACADM	97.1%	94.2%	100%	99.4%	Acyl-CoA dehydrogenase, medium chain, deficiency of, 201450	METABOLIC DISORDERS PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹

ACADS	100%	100%	100%	99.2%	Acyl-CoA dehydrogenase, short-chain, deficiency of, 201470	METABOLIC DISORDERS PANEL INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL¹
ACADSB	100%	100%	100%	99.1%	2-methylbutyrylglucosuria, 610006	METABOLIC DISORDERS PANEL INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL¹

ACADVL	100%	100%	100%	98.1%	VLCAD deficiency, 201475	HEART DISORDERS PANEL ¹ METABOLIC DISORDERS PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹ MUSCLE DISORDERS PANEL
ACAN	99%	98.8%	97.5%	94.4%	?Spondyloepiphyseal dysplasia, Kimberley type, 608361;Short stature and advanced bone age, with or without early-onset osteoarthritis and/or osteochondritis dissecans, 165800;Spondyloepimetaphyseal dysplasia, aggrecan type, 612813	SHORT STATURE/SKELETAL DYSPLASIA PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹ MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS

ACAT1	100%	100%	100%	99.6%	Alpha-methylacetoacetic aciduria, 203750	METABOLIC DISORDERS PANEL INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹
ACAT2	100%	100%	100%	99%	?ACAT2 deficiency, 614055	METABOLIC DISORDERS PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS

ACBD5	85.6%	85.6%	100%	99%	Retinal dystrophy with leukodystrophy, 618863	VISION DISORDERS PANEL INHERITED BONE MARROW FAILURE AND/OR PREDISPOSITION PANEL TO HEMATOLOGICAL MALIGNANCIES HEMOSTATIC/THROMB OTIC DISORDERS PANEL METABOLIC DISORDERS PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹
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ACBD6	100%	100%	100%	99.6%	Neurodevelopmental disorder with progressive movement abnormalities, 620785	METABOLIC DISORDERS PANEL INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹
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ACD	100%	100%	100%	99.4%	?Dyskeratosis congenita, autosomal recessive 7, 616553;?Dyskeratosis congenita, autosomal dominant 6, 616553	INHERITED BONE MARROW FAILURE AND/OR PREDISPOSITION PANEL TO HEMATOLOGICAL MALIGNANCIES SKIN DISORDERS PANEL ¹ MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS PRIMARY IMMUNODEFICIENCIES PANEL DYSKERATOSIS CONGENITA AND APLASTIC ANEMIA PANEL HEREDITARY CANCER PANEL
ACE	100%	100%	99.9%	97.1%	{Stroke, hemorrhagic}, 614519;Renal tubular dysgenesis, 267430;{Myocardial infarction, susceptibility to};{Microvascular complications of diabetes 3}, 612624;[Angiotensin I-converting enzyme, benign serum increase];{SARS, progression of}	RENAL DISORDERS PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹

ACER3	96.3%	96.3%	100%	99.7%	?Leukodystrophy, progressive, early childhood-onset, 617762	INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹
ACKR3	100%	100%	100%	99.5%	?Oculomotor-abducens synkinesis, 619215	MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS

ACO2	93.2%	90.8%	100%	99.1%	Optic atrophy 9, 616289;Infantile cerebellar-retinal degeneration, 614559	MOVEMENT DISORDERS PANEL VISION DISORDERS PANEL EPILEPSY PANEL METABOLIC DISORDERS PANEL INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS MITOCHONDRIAL DISORDERS PANEL COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL¹
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ACOX1	100%	100%	100%	99.1%	Mitchell syndrome, 618960;Peroxisomal acyl-CoA oxidase deficiency, 264470	POLYNEUROPATHIES PANEL ¹ METABOLIC DISORDERS PANEL INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹
ACOX2	100%	100%	100%	99.4%	Bile acid synthesis defect, congenital, 6, 617308	LIVER DISORDERS PANEL METABOLIC DISORDERS PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹

ACP4	100%	100%	100%	99.1%	Amelogenesis imperfecta, type IJ, 617297	CRANIOFACIAL ANOMALIES PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS
ACP5	100%	100%	100%	97.9%	Spondyloenchondrodysplasia with immune dysregulation, 607944	PRIMARY IMMUNODEFICIENCIES PANEL SHORT STATURE/SKELETAL DYSPLASIA PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹
ACR	99.9%	98%	94.1%	81%	?Spermatogenic failure 87, 620500	MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS

ACSF3	100%	100%	100%	99.3%	Combined malonic and methylmalonic aciduria, 614265	HEART DISORDERS PANEL ¹ METABOLIC DISORDERS PANEL INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹
ACSL4	100%	100%	99%	69.8%	Intellectual developmental disorder, X-linked 63, 300387	METABOLIC DISORDERS PANEL INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS
ACSL5	100%	100%	100%	99.3%	?Diarrhea 13, 620357	MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS

ACSL6	100%	99.9%	100%	99.4%		MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS
ACTA1	100%	100%	100%	97.2%	Congenital myopathy 2B, severe infantile, autosomal recessive, 620265;?Myopathy, scapulohumeroperoneal, 616852;Congenital myopathy 2C, severe infantile, autosomal dominant, 620278;Congenital myopathy 2A, typical, autosomal dominant, 161800	FETAL AKINESIA PANEL HEART DISORDERS PANEL ¹ MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS MITOCHONDRIAL DISORDERS PANEL COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹ MUSCLE DISORDERS PANEL

ACTA2	100%	100%	100%	99.5%	Smooth muscle dysfunction syndrome, 613834;Aortic aneurysm, familial thoracic 6, 611788;Moyamoya disease 5, 614042	THORACIC AORTIC ANEURYSM AND/OR DISSECTION PANEL CONGENITAL HEARTDISEASE PANEL ¹ SKIN DISORDERS PANEL ¹ HEART DISORDERS PANEL ¹ LIVER DISORDERS PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS
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ACTB	100%	100%	100%	99.5%	Baraitser-Winter syndrome 1, 243310;Becker nevus, syndromic or isolated, somatic mosaic, 604919;Thrombocytopenia 8, with dysmorphic features and developmental delay, 620475;Dystonia-deafness syndrome 1, 607371;Congenital smooth muscle hamartoma with or without hemihypertrophy, somatic mosaic, 620470	MOVEMENT DISORDERS PANEL INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS OROFACIAL CLEFTING PANEL WITH GENOME WIDE CNV ANALYSIS HEMOSTATIC/THROMB OTIC DISORDERS PANEL PRIMARY IMMUNODEFICIENCIES PANEL SHORT STATURE/SKELETAL DYSPLASIA PANEL WITH GENOME WIDE CNV ANALYSIS HEARING IMPAIRMENT PANEL (INCLUDING GJB2) DISORDERS/DIFFEREN CES OF SEX DEVELOPMENT (DSD) / PRIMARY ADRENAL INSUFFICIENCY PANEL EPILEPSY PANEL SKIN DISORDERS PANEL ¹
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ACTC1	100%	100%	100%	99.3%	Left ventricular noncompaction 4, 613424;Cardiomyopathy, hypertrophic, 11, 612098;Atrial septal defect 5, 612794;Cardiomyopathy, dilated, 1R, 613424	CONGENITAL HEARTDISEASE PANEL¹ MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS DILATED CARDIOMYOPATHY PANEL¹ HEART DISORDERS PANEL¹ HYPERTROPHIC CARDIOMYOPATHY PANEL¹
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ACTG1	100%	100%	100%	98.5%	Deafness, autosomal dominant 20/26, 604717;Baraitser-Winter syndrome 2, 614583	CRANIOFACIAL ANOMALIES PANEL WITH GENOME WIDE CNV ANALYSIS HEARING IMPAIRMENT PANEL (INCLUDING GJB2) MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS OROFACIAL CLEFTING PANEL WITH GENOME WIDE CNV ANALYSIS SHORT STATURE/SKELETAL DYSPLASIA PANEL WITH GENOME WIDE CNV ANALYSIS INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS
ACTG2	100%	100%	100%	99.1%	Megacystis-microcolon-intestinal hypoperistalsis syndrome 5, 619431;Visceral myopathy 1, 155310	LIVER DISORDERS PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS

ACTL6A	100%	100%	100%	99.7%		INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS
ACTL6B	100%	100%	100%	98.3%	Developmental and epileptic encephalopathy 76, 618468;Intellectual developmental disorder with severe speech and ambulation defects, 618470	EPILEPSY PANEL INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹
ACTL7A	100%	100%	100%	98.9%	Spermatogenic failure 86, 620499	MALE INFERTILITY PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS

ACTL9	100%	100%	100%	99.2%	Spermatogenic failure 53, 619258	MALE INFERTILITY PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS
ACTN1	100%	100%	100%	98.8%	Bleeding disorder, platelet-type, 15, 615193	HEMOSTATIC/THROMB OTIC DISORDERS PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS
ACTN2	100%	100%	99.9%	97.1%	Myopathy, distal, 6, adult onset, 618655;Cardiomyopathy, hypertrophic, 23, with or without LVNC, 612158;Congenital myopathy 8, 618654;Cardiomyopathy, dilated, 1AA, with or without LVNC, 612158	DILATED CARDIOMYOPATHY PANEL ¹ HEART DISORDERS PANEL ¹ HYPERTROPHIC CARDIOMYOPATHY PANEL ¹ MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS MUSCLE DISORDERS PANEL

ACTN4	98.9%	98.9%	100%	98.4%	Glomerulosclerosis, focal segmental, 1, 603278	MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS RENAL DISORDERS PANEL
ACTRT1	100%	100%	98.6%	68.3%		MALE INFERTILITY PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS
ACVR1	100%	100%	100%	99.6%	Fibrodysplasia ossificans progressiva, 135100	SHORT STATURE/SKELETAL DYSPLASIA PANEL WITH GENOME WIDE CNV ANALYSIS INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS MUSCLE DISORDERS PANEL

ACVR1B	100%	100%	100%	98.7%	Pancreatic cancer, somatic, 260350	MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS
ACVR2B	100%	100%	100%	98.5%	Heterotaxy, visceral, 4, autosomal, 613751	CONGENITAL HEARTDISEASE PANEL¹ CILIOPATHIES PANEL HEART DISORDERS PANEL¹ MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS
ACVRL1	100%	100%	100%	99%	Telangiectasia, hereditary hemorrhagic, type 2, 600376	SKIN DISORDERS PANEL¹ HEMOSTATIC/THROMB OTIC DISORDERS PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS

ACY1	100%	100%	100%	98.9%	Aminoacylase 1 deficiency, 609924	EPILEPSY PANEL METABOLIC DISORDERS PANEL INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹
ADA	86.1%	84.5%	100%	98.7%	Adenosine deaminase deficiency, partial, 102700;Severe combined immunodeficiency due to ADA deficiency, 102700	PRIMARY IMMUNODEFICIENCIES PANEL METABOLIC DISORDERS PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹ SEVERE COMBINED IMMUNODEFICIENCY (SCID) PANEL

ADA2	95.1%	93.3%	100%	98.9%	Sneddon syndrome, 182410;Vasculitis, autoinflammation, immunodeficiency, and hematologic defects syndrome, 615688	SKIN DISORDERS PANEL ¹ PRIMARY IMMUNODEFICIENCIES PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹
ADAD2	100%	100%	100%	98.6%		MALE INFERTILITY PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS
ADAM10	100%	100%	100%	99.2%	{Alzheimer disease 18, susceptibility to}, 615590;Reticulate acropigmentation of Kitamura, 615537	SKIN DISORDERS PANEL ¹ MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS

ADAM17	99.2%	99.2%	100%	98.8%	?Inflammatory skin and bowel disease, neonatal, 1, 614328	SKIN DISORDERS PANEL ¹ PRIMARY IMMUNODEFICIENCIES PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹
ADAM22	100%	100%	100%	99.5%	Developmental and epileptic encephalopathy 61, 617933	EPILEPSY PANEL INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹

ADAM9	95%	95%	100%	99.3%	Cone-rod dystrophy 9, 612775	VISION DISORDERS PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹
ADAMTS1	100%	100%	100%	99.2%		HEARING IMPAIRMENT PANEL (INCLUDING GJB2) MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS

ADAMTS10	100%	100%	100%	98.7%	Weill-Marchesani syndrome 1, recessive, 277600	VISION DISORDERS PANEL SKIN DISORDERS PANEL ¹ SHORT STATURE/SKELETAL DYSPLASIA PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS MITOCHONDRIAL DISORDERS PANEL COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹
ADAMTS13	100%	100%	100%	98.5%	Thrombotic thrombocytopenic purpura, hereditary, 274150	HEMOSTATIC/THROMB OTIC DISORDERS PANEL RENAL DISORDERS PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹

ADAMTS15	100%	100%	100%	99.1%	Arthrogryposis, distal, type 12, 620545	FETAL AKINESIA PANEL SHORT STATURE/SKELETAL DYSPLASIA PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹
ADAMTS17	100%	99.5%	100%	98.6%	Weill-Marchesani 4 syndrome, recessive, 613195	VISION DISORDERS PANEL SKIN DISORDERS PANEL ¹ SHORT STATURE/SKELETAL DYSPLASIA PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹

ADAMTS18	100%	100%	100%	99.7%	Microcornea, myopic chorioretinal atrophy, and telecanthus, 615458	VISION DISORDERS PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹
ADAMTS19	100%	100%	100%	98.4%	Cardiac valvular dysplasia 2, 620067	CONGENITAL HEARTDISEASE PANEL ¹ HEART DISORDERS PANEL ¹ MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹
ADAMTS2	97.9%	97.8%	100%	99.3%	Ehlers-Danlos syndrome, dermatosparaxis type, 225410	SKIN DISORDERS PANEL ¹ MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹

ADAMTS3	100%	100%	100%	99.4%	Hennekam lymphangiectasia-lymphedema syndrome 3, 618154	SKIN DISORDERS PANEL ¹ LYMPHATIC ANOMALIES PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹
ADAMTS9	100%	100%	100%	98.1%		CILIOPATHIES PANEL RENAL DISORDERS PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS

ADAMTSL2	100%	99.7%	100%	99.1%	Geleophysic dysplasia 1, 231050	SKIN DISORDERS PANEL ¹ SHORT STATURE/SKELETAL DYSPLASIA PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹
ADAMTSL4	100%	100%	100%	98.8%	Ectopia lentis et pupillae, 225200;Ectopia lentis, isolated, autosomal recessive, 225100	VISION DISORDERS PANEL CRANIOFACIAL ANOMALIES PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹

ADAR	100%	100%	100%	98.8%	Dyschromatosis symmetrica hereditaria, 127400;Aicardi-Goutieres syndrome 6, 615010	MOVEMENT DISORDERS PANEL MITOCHONDRIAL DISORDERS PANEL COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹ SKIN DISORDERS PANEL ¹ PRIMARY IMMUNODEFICIENCIES PANEL INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS
ADARB1	94.8%	94.7%	100%	99.2%	Neurodevelopmental disorder with hypotonia, microcephaly, and seizures, 618862	EPILEPSY PANEL INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹

ADAT3	100%	100%	100%	99.7%	Neurodevelopmental disorder with brain abnormalities, poor growth, and dysmorphic facies, 615286	INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹
ADCK2	100%	99.9%	100%	98.3%		MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS MITOCHONDRIAL DISORDERS PANEL
ADCK5	100%	100%	100%	98.2%		METABOLIC DISORDERS PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS
ADCY1	98.9%	98.2%	100%	98.2%	?Deafness, autosomal recessive 44, 610154	HEARING IMPAIRMENT PANEL (INCLUDING GJB2) MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS

ADCY10	100%	100%	100%	99.5%	{Hypercalciuria, absorptive, susceptibility to}, 143870	MALE INFERTILITY PANEL RENAL DISORDERS PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS
ADCY3	100%	100%	100%	98.8%	{Obesity, susceptibility to, BMIQ19}, 617885	DISORDERS/DIFFEREN CES OF SEX DEVELOPMENT (DSD) / PRIMARY ADRENAL INSUFFICIENCY PANEL HYPOGONADOTROPIC HYPOGONADISM (KALLMANN) PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS

ADCY5	97.3%	96.7%	100%	98.6%	Dyskinesia with orofacial involvement, autosomal dominant, 606703;Neurodevelopmental disorder with hyperkinetic movements and dyskinesia, 619651;Dyskinesia with orofacial involvement, autosomal recessive, 619647	MOVEMENT DISORDERS PANEL HEART DISORDERS PANEL ¹ METABOLIC DISORDERS PANEL INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹
ADCY6	100%	100%	100%	99.4%	Lethal congenital contracture syndrome 8, 616287	FETAL AKINESIA PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹

ADD1	100%	100%	100%	99.1%	{Hypertension, essential, salt-sensitive}, 145500	INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS
ADD3	100%	100%	100%	99.2%	Cerebral palsy, spastic quadriplegic, 3, 617008	INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹
ADGB	100%	100%	100%	98.5%		MALE INFERTILITY PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS
ADGRE2	99.8%	99%	99.1%	96.2%	Vibratory urticaria, 125630	MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS

ADGRG1	100%	100%	100%	98.7%	Cortical dysplasia, complex, with other brain malformations 14B, (bilateral perisylvian), 615752;Cortical dysplasia, complex, with other brain malformations 14A, (bilateral frontoparietal), 606854	MOVEMENT DISORDERS PANEL INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹
ADGRG2	100%	99.8%	99.2%	70.7%	Congenital bilateral absence of vas deferens, X-linked, 300985	MALE INFERTILITY PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS
ADGRG6	100%	100%	100%	99.6%	Lethal congenital contracture syndrome 9, 616503	FETAL AKINESIA PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹

ADGRL1	100%	100%	100%	98.6%	Developmental delay, behavioral abnormalities, and neuropsychiatric disorders, 620065	INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS
ADGRV1	100%	100%	100%	99.4%	Usher syndrome, type 2C, 605472;Usher syndrome, type 2C, GPR98/PDZD7 digenic, 605472;?Febrile seizures, familial, 4, 604352	VISION DISORDERS PANEL HEARING IMPAIRMENT PANEL (INCLUDING GJB2) MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹
ADH5	100%	100%	100%	99.6%	AMED syndrome, digenic, 619151	MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS
ADIPOQ	100%	100%	100%	99.3%	Adiponectin deficiency, 612556	MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS

ADIPOR1	100%	100%	100%	99.5%		VISION DISORDERS PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS
ADK	90.9%	90.9%	100%	99.6%	Hypermethioninemia due to adenosine kinase deficiency, 614300	LIVER DISORDERS PANEL METABOLIC DISORDERS PANEL INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹

ADNP	100%	100%	100%	99.6%	Helsmoortel-van der Aa syndrome, 615873	CONGENITAL HEARTDISEASE PANEL¹ HEART DISORDERS PANEL¹ INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS
ADPRS	100%	100%	100%	99.4%	Neurodegeneration, childhood-onset, stress-induced, with variable ataxia and seizures, 618170	MOVEMENT DISORDERS PANEL POLYNEUROPATHIES PANEL¹ INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS MITOCHONDRIAL DISORDERS PANEL COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL¹

ADRA2A	100%	100%	99.9%	94.2%	?Lipodystrophy, familial partial, type 8, 620679	MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS
ADRB1	99.8%	96.4%	100%	96.7%	?[Short sleep, familial natural, 2], 618591;[Resting heart rate], 607276	MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS
ADRB2	100%	100%	100%	99.3%	Beta-2-adrenoreceptor agonist, reduced response to	MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS
ADSL	100%	100%	100%	99.4%	Adenylosuccinase deficiency, 103050	EPILEPSY PANEL METABOLIC DISORDERS PANEL INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹

ADSS1	100%	100%	99.9%	96.8%	Myopathy, distal, 5, 617030	MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹ MUSCLE DISORDERS PANEL
AEBP1	100%	100%	100%	97.6%	Ehlers-Danlos syndrome, classic-like, 2, 618000	MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹
AFF2	100%	99.8%	99.3%	70.2%	Intellectual developmental disorder, X-linked 109, 309548	INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS

AFF3	100%	100%	100%	98.8%	KINSSHIP syndrome, 619297	SHORT STATURE/SKELETAL DYSPLASIA PANEL WITH GENOME WIDE CNV ANALYSIS INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS
AFF4	100%	100%	100%	99.5%	CHOPS syndrome, 616368	INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS

AFG2A	100%	100%	100%	99.4%	Neurodevelopmental disorder with hearing loss, seizures, and brain abnormalities, 616577	HEARING IMPAIRMENT PANEL (INCLUDING GJB2) EPILEPSY PANEL INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS MITOCHONDRIAL DISORDERS PANEL COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹
AFG2B	100%	100%	100%	99.6%	Deafness, autosomal recessive 119, 619615;Neurodevelopmental disorder with hearing loss and spasticity, 619616	HEARING IMPAIRMENT PANEL (INCLUDING GJB2) INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹

AFG3L2	100%	100%	100%	99%	Spastic ataxia 5, autosomal recessive, 614487;Optic atrophy 12, 618977;Spinocerebellar ataxia 28, 610246	MOVEMENT DISORDERS PANEL VISION DISORDERS PANEL POLYNEUROPATHIES PANEL ¹ INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS MITOCHONDRIAL DISORDERS PANEL COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹
AFP	100%	100%	100%	99.6%	[Hereditary persistence of alpha-fetoprotein], 615970;Alpha-fetoprotein deficiency, 615969	MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS

AGA	100%	100%	100%	99.2%	Aspartylglucosaminuria, 208400	MOVEMENT DISORDERS PANEL SKIN DISORDERS PANEL ¹ EPILEPSY PANEL PRIMARY IMMUNODEFICIENCIES PANEL SHORT STATURE/SKELETAL DYSPLASIA PANEL WITH GENOME WIDE CNV ANALYSIS METABOLIC DISORDERS PANEL INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹
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AGAP1	100%	99.9%	100%	97.4%		INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS
AGBL1	100%	100%	100%	99.4%	Corneal dystrophy, Fuchs endothelial, 8, 615523	VISION DISORDERS PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS
AGBL5	100%	100%	100%	98.8%	Retinitis pigmentosa 75, 617023	VISION DISORDERS PANEL HEARING IMPAIRMENT PANEL (INCLUDING GJB2) MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹

AGK	91.7%	91.7%	100%	99.6%	Cataract 38, autosomal recessive, 614691;Sengers syndrome, 212350	VISION DISORDERS PANEL HEART DISORDERS PANEL ¹ METABOLIC DISORDERS PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS MITOCHONDRIAL DISORDERS PANEL COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹
AGL	100%	100%	100%	99.6%	Glycogen storage disease IIIa, 232400;Glycogen storage disease IIIb, 232400	HEART DISORDERS PANEL ¹ METABOLIC DISORDERS PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹ MUSCLE DISORDERS PANEL

AGMO	100%	100%	100%	99.4%		INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS
AGO1	100%	100%	100%	99.2%	Neurodevelopmental disorder with language delay and behavioral abnormalities, with or without seizures, 620292	INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS
AGO2	99.8%	99.3%	100%	98.7%	Lessel-Kreienkamp syndrome, 619149	INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS

AGPAT2	100%	100%	100%	98.8%	Lipodystrophy, congenital generalized, type 1, 608594	SKIN DISORDERS PANEL ¹ HEART DISORDERS PANEL ¹ METABOLIC DISORDERS PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹
AGPS	97.3%	97.3%	100%	99.1%	Rhizomelic chondrodysplasia punctata, type 3, 600121	SHORT STATURE/SKELETAL DYSPLASIA PANEL WITH GENOME WIDE CNV ANALYSIS METABOLIC DISORDERS PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹

AGR2	100%	100%	100%	99.1%	Respiratory infections, recurrent, and failure to thrive with or without diarrhea, 620233	MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹
AGRN	100%	100%	100%	97.5%	Myasthenic syndrome, congenital, 8, with pre- and postsynaptic defects, 615120	POLYNEUROPATHIES PANEL ¹ MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹ MUSCLE DISORDERS PANEL
AGT	96.1%	95.3%	100%	99.5%	Renal tubular dysgenesis, 267430	RENAL DISORDERS PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹

AGTPBP1	100%	100%	100%	99.4%	Neurodegeneration, childhood-onset, with cerebellar atrophy, 618276	MOVEMENT DISORDERS PANEL POLYNEUROPATHIES PANEL ¹ INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹
AGTR1	100%	100%	100%	99.4%	{Hypertension, essential}, 145500;Renal tubular dysgenesis, 267430	RENAL DISORDERS PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹

AGXT	100%	100%	100%	98.8%	Hyperoxaluria, primary, type 1, 259900	METABOLIC DISORDERS PANEL RENAL DISORDERS PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL¹
AHCY	100%	100%	100%	99.4%	Hypermethioninemia with deficiency of S-adenosylhomocysteine hydrolase, 613752	LIVER DISORDERS PANEL METABOLIC DISORDERS PANEL INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL¹

AHDC1	100%	100%	100%	96.7%	Xia-Gibbs syndrome, 615829	INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS
AHI1	98.7%	98.7%	100%	98.8%	Joubert syndrome 3, 608629	VISION DISORDERS PANEL CILIOPATHIES PANEL INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS RENAL DISORDERS PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹
AHNAK2	97.9%	97.8%	96.8%	91.2%		POLYNEUROPATHIES PANEL ¹ MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS

AHR	100%	100%	100%	99.7%	Foveal hypoplasia 3, 620958;?Retinitis pigmentosa 85, 618345	VISION DISORDERS PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹
AHSG	100%	100%	100%	99.1%	?Alopecia-intellectual disability syndrome 1, 203650	INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS
AICDA	92.1%	92.1%	100%	98.5%	Immunodeficiency with hyper-IgM, type 2, 605258	PRIMARY IMMUNODEFICIENCIES PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹

AIFM1	100%	99.8%	99.3%	70.3%	Combined oxidative phosphorylation deficiency 6, 300816;Cowchock syndrome, 310490;Spondyloepimetaphyseal dysplasia, X-linked, with hypomyelinating leukodystrophy, 300232;Deafness, X-linked 5, 300614	HEARING IMPAIRMENT PANEL (INCLUDING GJB2) POLYNEUROPATHIES PANEL ¹ SHORT STATURE/SKELETAL DYSPLASIA PANEL WITH GENOME WIDE CNV ANALYSIS INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS MITOCHONDRIAL DISORDERS PANEL
AIMP1	100%	100%	100%	99.2%	Leukodystrophy, hypomyelinating, 3, 260600	MOVEMENT DISORDERS PANEL INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹

AIMP2	100%	100%	100%	98.7%	Leukodystrophy, hypomyelinating, 17, 618006	INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹
AIP	100%	100%	100%	99.2%	Pituitary adenoma 1, multiple types, 102200;Pituitary adenoma predisposition, 102200	MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS HEREDITARY CANCER PANEL
AIPL1	100%	99.9%	100%	99.1%	Leber congenital amaurosis 4, 604393;Retinitis pigmentosa, juvenile, 604393;Cone-rod dystrophy, 604393	VISION DISORDERS PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹

AIRE	100%	100%	100%	98.3%	Autoimmune polyendocrinopathy syndrome , type I, with or without reversible metaphyseal dysplasia, 240300	SKIN DISORDERS PANEL ¹ DISORDERS/DIFFERENCES OF SEX DEVELOPMENT (DSD) / PRIMARY ADRENAL INSUFFICIENCY PANEL PRIMARY IMMUNODEFICIENCIES PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹
AJAP1	100%	100%	100%	97.9%		INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS

AK1	100%	100%	100%	98.6%	Anemia, congenital, nonspherocytic hemolytic, 3, adenylate kinase deficient, 612631	COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹ METABOLIC DISORDERS PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS
AK2	100%	100%	100%	99.3%	Reticular dysgenesis, 267500	PRIMARY IMMUNODEFICIENCIES PANEL SEVERE COMBINED IMMUNODEFICIENCY (SCID) PANEL METABOLIC DISORDERS PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹
AK3	100%	100%	100%	99%		MITOCHONDRIAL DISORDERS PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS

AK7	100%	100%	100%	99.2%	?Spermatogenic failure 27, 617965	MALE INFERTILITY PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS
AK9	100%	100%	100%	99.6%	Spermatogenic failure 89, 620705	MALE INFERTILITY PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS
AKAP3	100%	100%	100%	99.6%	Spermatogenic failure 82, 620353	MALE INFERTILITY PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS
AKAP9	100%	100%	100%	99.1%	?Long QT syndrome 11, 611820	HEART DISORDERS PANEL ¹ MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS

AKR1C1	100%	100%	100%	99.5%		METABOLIC DISORDERS PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS
AKR1C2	100%	100%	99.9%	97.3%	46XY sex reversal 8, 614279	DISORDERS/DIFFEREN CES OF SEX DEVELOPMENT (DSD) / PRIMARY ADRENAL INSUFFICIENCY PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL¹
AKR1D1	100%	100%	100%	99.5%	Bile acid synthesis defect, congenital, 2, 235555	LIVER DISORDERS PANEL METABOLIC DISORDERS PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL¹

AKT1	100%	99.9%	100%	99.6%	Breast cancer, somatic, 114480;Cowden syndrome 6, 615109;Colorectal cancer, somatic, 114500;Proteus syndrome, somatic, 176920;Ovarian cancer, somatic, 167000	SKIN DISORDERS PANEL ¹ MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS HEREDITARY CANCER PANEL
AKT2	100%	99.9%	100%	98.9%	Diabetes mellitus, type II, 125853;Hypoinsulinemic hypoglycemia with hemihypertrophy, 240900	TALL STATURE PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS
AKT3	94.6%	94.5%	100%	99.4%	Megalencephaly-polymicrogyria-polydactyly-hydrocephalus syndrome 2, 615937	SKIN DISORDERS PANEL ¹ INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS

ALAD	100%	100%	100%	98.7%	Porphyria, acute hepatic, 612740;{Lead poisoning, susceptibility to}, 612740	SKIN DISORDERS PANEL ¹ METABOLIC DISORDERS PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹
ALAS2	100%	99.1%	98.6%	66.6%	Anemia, sideroblastic, 1, 300751;Protoporphyria, erythropoietic, X-linked, 300752	INHERITED BONE MARROW FAILURE AND/OR PREDISPOSITION PANEL TO HEMATOLOGICAL MALIGNANCIES SKIN DISORDERS PANEL ¹ IRON DISORDERS PANEL METABOLIC DISORDERS PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS

ALB	100%	100%	100%	99.5%	?[Dysalbuminemic hypertriiodothyroninemia], 615999;Analbuminemia, 616000;[Dysalbuminemic hyperthyroxinemia], 615999	MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹
ALDH18A1	100%	100%	100%	99.1%	Spastic paraplegia 9A, autosomal dominant, 601162;Cutis laxa, autosomal recessive, type IIIA, 219150;Spastic paraplegia 9B, autosomal recessive, 616586;Cutis laxa, autosomal dominant 3, 616603	MOVEMENT DISORDERS PANEL INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹ VISION DISORDERS PANEL SKIN DISORDERS PANEL ¹ METABOLIC DISORDERS PANEL

ALDH1A2	100%	100%	100%	99.3%	Diaphragmatic hernia 4, with cardiovascular defects, 620025	CONGENITAL HEARTDISEASE PANEL¹ HEART DISORDERS PANEL¹ MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL¹
ALDH1A3	100%	100%	100%	99.4%	Microphthalmia, isolated 8, 615113	VISION DISORDERS PANEL METABOLIC DISORDERS PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL¹
ALDH1B1	100%	100%	100%	98.8%		MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS MITOCHONDRIAL DISORDERS PANEL

ALDH2	100%	100%	100%	98.8%	{Esophageal cancer, alcohol-related, susceptibility to};{Sublingual nitroglycerin, susceptibility to poor response to};Alcohol sensitivity, acute, 610251;{Hangover, susceptibility to}, 610251	METABOLIC DISORDERS PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS
ALDH3A2	93.5%	93.5%	100%	99.7%	Sjogren-Larsson syndrome, 270200	MOVEMENT DISORDERS PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹ VISION DISORDERS PANEL SKIN DISORDERS PANEL ¹ METABOLIC DISORDERS PANEL INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS

ALDH4A1	100%	100%	100%	98.5%	Hyperprolinemia, type II, 239510	EPILEPSY PANEL METABOLIC DISORDERS PANEL INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL¹
ALDH5A1	100%	100%	100%	98.7%	Succinic semialdehyde dehydrogenase deficiency, 271980	MOVEMENT DISORDERS PANEL EPILEPSY PANEL METABOLIC DISORDERS PANEL INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL¹

ALDH6A1	100%	100%	100%	99.6%	Methylmalonate semialdehyde dehydrogenase deficiency, 614105	METABOLIC DISORDERS PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹
ALDH7A1	100%	100%	100%	99.1%	Epilepsy, early-onset, 4, vitamin B6-dependent, 266100	EPILEPSY PANEL METABOLIC DISORDERS PANEL INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹
ALDOA	100%	100%	100%	98.7%	Glycogen storage disease XII, 611881	METABOLIC DISORDERS PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹

ALDOB	100%	100%	100%	99.4%	Fructose intolerance, hereditary, 229600	SKIN DISORDERS PANEL ¹ LIVER DISORDERS PANEL METABOLIC DISORDERS PANEL RENAL DISORDERS PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹
ALG1	100%	100%	100%	99.1%	Congenital disorder of glycosylation, type Ik, 608540	EPILEPSY PANEL METABOLIC DISORDERS PANEL INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS RENAL DISORDERS PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹

ALG10	100%	100%	100%	99.6%		METABOLIC DISORDERS PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS
ALG11	91%	91%	100%	99.7%	Congenital disorder of glycosylation, type Ip, 613661	EPILEPSY PANEL METABOLIC DISORDERS PANEL INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL¹

ALG12	100%	100%	100%	98.7%	Congenital disorder of glycosylation, type Ig, 607143	SHORT STATURE/SKELETAL DYSPLASIA PANEL WITH GENOME WIDE CNV ANALYSIS METABOLIC DISORDERS PANEL INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL¹
ALG13	100%	100%	98.7%	69.3%	Developmental and epileptic encephalopathy 36, 300884	EPILEPSY PANEL PRIMARY IMMUNODEFICIENCIES PANEL METABOLIC DISORDERS PANEL INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS

ALG14	100%	100%	100%	99.6%	Intellectual developmental disorder with epilepsy, behavioral abnormalities, and coarse facies, 619031;Myopathy, epilepsy, and progressive cerebral atrophy, 619036;?Myasthenic syndrome, congenital, 15, without tubular aggregates, 616227	METABOLIC DISORDERS PANEL INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹ EPILEPSY PANEL
ALG2	100%	100%	100%	99.1%	Congenital disorder of glycosylation, type li, 607906;Myasthenic syndrome, congenital, 14, with tubular aggregates, 616228	METABOLIC DISORDERS PANEL INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹ MUSCLE DISORDERS PANEL

ALG3	100%	100%	100%	98.9%	Congenital disorder of glycosylation, type Id, 601110	FETAL AKINESIA PANEL EPILEPSY PANEL SHORT STATURE/SKELETAL DYSPLASIA PANEL WITH GENOME WIDE CNV ANALYSIS METABOLIC DISORDERS PANEL INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹
ALG5	100%	100%	100%	99.5%	Polycystic kidney disease 7, 620056	RENAL DISORDERS PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS

ALG6	100%	100%	100%	99.8%	Congenital disorder of glycosylation, type Ic, 603147	EPILEPSY PANEL METABOLIC DISORDERS PANEL INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL¹
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ALG8	78.5%	77.5%	100%	98.9%	Congenital disorder of glycosylation, type Ih, 608104;Polycystic liver disease 3 with or without kidney cysts, 617874	LIVER DISORDERS PANEL LYMPHATIC ANOMALIES PANEL WITH GENOME WIDE CNV ANALYSIS METABOLIC DISORDERS PANEL INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS RENAL DISORDERS PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹
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ALG9	100%	100%	100%	99.2%	Gillesen-Kaesbach-Nishimura syndrome, 263210;Congenital disorder of glycosylation, type II, 608776	SHORT STATURE/SKELETAL DYSPLASIA PANEL WITH GENOME WIDE CNV ANALYSIS LIVER DISORDERS PANEL METABOLIC DISORDERS PANEL INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS RENAL DISORDERS PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL¹
ALK	100%	100%	100%	99.1%	{Neuroblastoma, susceptibility to, 3}, 613014	MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS HEREDITARY CANCER PANEL

ALKBH1	100%	100%	100%	99.6%		MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS MITOCHONDRIAL DISORDERS PANEL
ALKBH8	100%	100%	100%	99.4%	Intellectual developmental disorder, autosomal recessive 71, 618504	INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹

ALMS1	100%	100%	100%	99.6%	Alstrom syndrome, 203800	VISION DISORDERS PANEL CILIOPATHIES PANEL HEARING IMPAIRMENT PANEL (INCLUDING GJB2) HEART DISORDERS PANEL ¹ SHORT STATURE/SKELETAL DYSPLASIA PANEL WITH GENOME WIDE CNV ANALYSIS INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS RENAL DISORDERS PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹
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ALOX12B	100%	100%	100%	99%	Ichthyosis, congenital, autosomal recessive 2, 242100	SKIN DISORDERS PANEL ¹ METABOLIC DISORDERS PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹
ALOXE3	100%	100%	100%	98.9%	Ichthyosis, congenital, autosomal recessive 3, 606545	SKIN DISORDERS PANEL ¹ MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹
ALPI	100%	100%	100%	99.2%		PRIMARY IMMUNODEFICIENCIES PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS

ALPK1	100%	100%	100%	99.5%	ROSAH syndrome, 614979	VISION DISORDERS PANEL PRIMARY IMMUNODEFICIENCIES PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS
ALPK3	100%	100%	100%	99%	Cardiomyopathy, familial hypertrophic 27, 618052	HEART DISORDERS PANEL ¹ HYPERTROPHIC CARDIOMYOPATHY PANEL ¹ MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹

ALPL	100%	100%	100%	97.8%	Odontohypophosphatasia, 146300;Hypophosphatasia, infantile, 241500;Hypophosphatasia, childhood, 241510;Hypophosphatasia, adult, 146300	SKIN DISORDERS PANEL ¹ SHORT STATURE/SKELETAL DYSPLASIA PANEL WITH GENOME WIDE CNV ANALYSIS METABOLIC DISORDERS PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹
ALS2	97.1%	97.1%	100%	99.3%	Primary lateral sclerosis, juvenile, 606353;Spastic paralysis, infantile onset ascending, 607225;Amyotrophic lateral sclerosis 2, juvenile, 205100	AMYOTROPHIC LATERAL SCLEROSIS, ALS PANEL MOVEMENT DISORDERS PANEL COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹ MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS

ALX1	100%	100%	100%	99.4%	Frontonasal dysplasia 3, 613456	CRANIOFACIAL ANOMALIES PANEL WITH GENOME WIDE CNV ANALYSIS SHORT STATURE/SKELETAL DYSPLASIA PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL¹ OROFACIAL CLEFTING PANEL WITH GENOME WIDE CNV ANALYSIS
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ALX3	100%	100%	100%	97.8%	Frontonasal dysplasia 1, 136760	CRANIOFACIAL ANOMALIES PANEL WITH GENOME WIDE CNV ANALYSIS SHORT STATURE/SKELETAL DYSPLASIA PANEL WITH GENOME WIDE CNV ANALYSIS INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL¹ OROFACIAL CLEFTING PANEL WITH GENOME WIDE CNV ANALYSIS
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ALX4	100%	100%	100%	99.4%	Parietal foramina 2, 609597;{Craniosynostosis 5, susceptibility to}, 615529;Frontonasal dysplasia 2, 613451	CRANIOFACIAL ANOMALIES PANEL WITH GENOME WIDE CNV ANALYSIS SKIN DISORDERS PANEL ¹ SHORT STATURE/SKELETAL DYSPLASIA PANEL WITH GENOME WIDE CNV ANALYSIS INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹
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AMACR	100%	100%	100%	99.4%	Alpha-methylacyl-CoA racemase deficiency, 614307;Bile acid synthesis defect, congenital, 4, 214950	VISION DISORDERS PANEL EPILEPSY PANEL POLYNEUROPATHIES PANEL ¹ LIVER DISORDERS PANEL METABOLIC DISORDERS PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹
AMBN	100%	100%	100%	99.5%	Amelogenesis imperfecta, type IF, 616270	CRANIOFACIAL ANOMALIES PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹

AMELX	100%	100%	99%	67.2%	Amelogenesis imperfecta, type 1E, 301200	CRANIOFACIAL ANOMALIES PANEL WITH GENOME WIDE CNV ANALYSIS SKIN DISORDERS PANEL ¹ MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS
AMER1	100%	99.9%	98.5%	65%	Osteopathia striata with cranial sclerosis, 300373	CRANIOFACIAL ANOMALIES PANEL WITH GENOME WIDE CNV ANALYSIS SHORT STATURE/SKELETAL DYSPLASIA PANEL WITH GENOME WIDE CNV ANALYSIS INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS OROFACIAL CLEFTING PANEL WITH GENOME WIDE CNV ANALYSIS

AMFR	100%	99.9%	100%	99%	Spastic paraplegia 89, autosomal recessive, 620379	MOVEMENT DISORDERS PANEL INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL¹
AMH	100%	100%	100%	98.7%	Persistent Mullerian duct syndrome, type I, 261550	DISORDERS/DIFFEREN CES OF SEX DEVELOPMENT (DSD) / PRIMARY ADRENAL INSUFFICIENCY PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS HEREDITARY CANCER PANEL

AMHR2	100%	100%	100%	99.2%	Persistent Mullerian duct syndrome, type II, 261550	DISORDERS/DIFFERENCES OF SEX DEVELOPMENT (DSD) / PRIMARY ADRENAL INSUFFICIENCY PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS HEREDITARY CANCER PANEL
AMMECR1	99.2%	96.1%	98.6%	68.9%	Midface hypoplasia, hearing impairment, elliptocytosis, and nephrocalcinosis, 300990	HEARING IMPAIRMENT PANEL (INCLUDING GJB2) SHORT STATURE/SKELETAL DYSPLASIA PANEL WITH GENOME WIDE CNV ANALYSIS INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS OROFACIAL CLEFTING PANEL WITH GENOME WIDE CNV ANALYSIS

AMN	100%	100%	100%	98.9%	Imerslund-Grasbeck syndrome 2, 618882	INHERITED BONE MARROW FAILURE AND/OR PREDISPOSITION PANEL TO HEMATOLOGICAL MALIGNANCIES METABOLIC DISORDERS PANEL RENAL DISORDERS PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL¹
AMOTL1	100%	100%	100%	99%		INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS OROFACIAL CLEFTING PANEL WITH GENOME WIDE CNV ANALYSIS

AMPD1	100%	100%	100%	99.2%	Myopathy due to myoadenylate deaminase deficiency, 615511	METABOLIC DISORDERS PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹ MUSCLE DISORDERS PANEL
AMPD2	100%	100%	100%	98.7%	Pontocerebellar hypoplasia, type 9, 615809;?Spastic paraplegia 63, autosomal recessive, 615686	MOVEMENT DISORDERS PANEL EPILEPSY PANEL INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹
AMPD3	100%	100%	100%	98.8%	[AMP deaminase deficiency, erythrocytic], 612874	METABOLIC DISORDERS PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS

AMT	100%	100%	100%	98.4%	Glycine encephalopathy 2, 620398	EPILEPSY PANEL METABOLIC DISORDERS PANEL INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹
AMTN	100%	100%	100%	99.7%	?Amelogenesis imperfecta, type IIIB, 617607	CRANIOFACIAL ANOMALIES PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS

ANAPC1	100%	100%	100%	99.1%	Rothmund-Thomson syndrome, type 1, 618625	SHORT STATURE/SKELETAL DYSPLASIA PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹
ANAPC7	100%	100%	100%	98.6%	Ferguson-Bonni neurodevelopmental syndrome, 619699	MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹
ANG	100%	100%	100%	98.8%	Amyotrophic lateral sclerosis 9, 611895	AMYOTROPHIC LATERAL SCLEROSIS, ALS PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS

ANGPT1	100%	100%	100%	99.5%	?Angioedema, hereditary, 5, 619361	PRIMARY IMMUNODEFICIENCIES PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS
ANGPT2	100%	100%	100%	99.3%	Lymphatic malformation 10, 619369	SKIN DISORDERS PANEL ¹ LYMPHATIC ANOMALIES PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS
ANGPTL3	100%	100%	100%	99.5%	Hypobetalipoproteinemia, familial, 2, 605019	MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹
ANGPTL4	100%	100%	100%	98.3%	Plasma triglyceride level QTL, low, 615881	MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS

ANK1	100%	100%	100%	99.2%	Spherocytosis, type 1, 182900	MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹
ANK2	100%	100%	100%	99.5%	Long QT syndrome 4, 600919;Cardiac arrhythmia, ankyrin-B-related, 600919	HEART DISORDERS PANEL ¹ INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS
ANK3	99.7%	99.7%	100%	99.5%	Intellectual developmental disorder, autosomal recessive 37, 615493	VISION DISORDERS PANEL INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹

ANKFY1	100%	100%	100%	99.1%		RENAL DISORDERS PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS
ANKH	100%	100%	100%	98.8%	Chondrocalcinosis 2, 118600;Craniometaphyseal dysplasia, 123000	SHORT STATURE/SKELETAL DYSPLASIA PANEL WITH GENOME WIDE CNV ANALYSIS INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL¹

ANKLE2	100%	100%	100%	97.3%	Microcephaly 16, primary, autosomal recessive, 616681	INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹
ANKRD1	100%	100%	99.8%	97.2%		CONGENITAL HEARTDISEASE PANEL ¹ HEART DISORDERS PANEL ¹ MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS

ANKRD11	100%	100%	100%	98.7%	KBG syndrome, 148050	CRANIOFACIAL ANOMALIES PANEL WITH GENOME WIDE CNV ANALYSIS CONGENITAL HEARTDISEASE PANEL¹ SKIN DISORDERS PANEL¹ EPILEPSY PANEL HEART DISORDERS PANEL¹ SHORT STATURE/SKELETAL DYSPLASIA PANEL WITH GENOME WIDE CNV ANALYSIS INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS OROFACIAL CLEFTING PANEL WITH GENOME WIDE CNV ANALYSIS
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ANKRD17	100%	100%	100%	99.4%	Chopra-Amiel-Gordon syndrome, 619504	INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS
ANKRD26	100%	100%	100%	99.5%	Thrombocytopenia 2, 188000	INHERITED BONE MARROW FAILURE AND/OR PREDISPOSITION PANEL TO HEMATOLOGICAL MALIGNANCIES HEMOSTATIC/THROMB OTIC DISORDERS PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS HEREDITARY CANCER PANEL
ANKS1B	100%	100%	100%	99.3%		INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS

ANKS6	99.3%	97.2%	100%	99.3%	Nephronophthisis 16, 615382	CILIOPATHIES PANEL LIVER DISORDERS PANEL RENAL DISORDERS PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹
ANLN	100%	100%	100%	99.4%	Focal segmental glomerulosclerosis 8, 616032	HEARING IMPAIRMENT PANEL (INCLUDING GJB2) RENAL DISORDERS PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS
ANO1	100%	100%	99.9%	97.3%	Moyamoya disease 7, 620687;?Intestinal dysmotility syndrome, 620045	MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS

ANO10	100%	100%	100%	99.5%	Spinocerebellar ataxia, autosomal recessive 10, 613728	MOVEMENT DISORDERS PANEL INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS MITOCHONDRIAL DISORDERS PANEL COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹
ANO3	100%	100%	100%	99.5%	Dystonia 24, 615034	MOVEMENT DISORDERS PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS
ANO4	100%	100%	100%	99.5%		INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS

ANO5	100%	100%	100%	99.6%	Muscular dystrophy, limb-girdle, autosomal recessive 12, 611307;Miyoshi muscular dystrophy 3, 613319;Gnathodiaphyseal dysplasia, 166260	SHORT STATURE/SKELETAL DYSPLASIA PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS MUSCLE DISORDERS PANEL COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL¹
ANO6	98.4%	98.4%	100%	99.1%	Scott syndrome, 262890	HEMOSTATIC/THROMBOTIC DISORDERS PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL¹

ANOS1	100%	99.8%	99.2%	66.3%	Hypogonadotropic hypogonadism 1 with or without anosmia (Kallmann syndrome 1), 308700	SKIN DISORDERS PANEL ¹ DISORDERS/DIFFERENCES OF SEX DEVELOPMENT (DSD) / PRIMARY ADRENAL INSUFFICIENCY PANEL HYPOGONADOTROPIC HYPOGONADISM (KALLMANN) PANEL RENAL DISORDERS PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS
ANTXR1	100%	99.9%	100%	97.3%	GAP0 syndrome, 230740;{?Hemangioma, capillary infantile, susceptibility to}, 602089	SKIN DISORDERS PANEL ¹ INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹

ANTXR2	96.3%	96.3%	100%	98.9%	Hyaline fibromatosis syndrome, 228600	SKIN DISORDERS PANEL ¹ SHORT STATURE/SKELETAL DYSPLASIA PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹
ANXA11	100%	100%	100%	99%	Amyotrophic lateral sclerosis 23, 617839;Inclusion body myopathy and brain white matter abnormalities, 619733	AMYOTROPHIC LATERAL SCLEROSIS, ALS PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS
AOPEP	100%	100%	100%	99.6%	Dystonia 31, 619565	MOVEMENT DISORDERS PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹

AP1B1	100%	100%	100%	99.4%	Keratitis-ichthyosis-deafness syndrome, autosomal recessive, 242150	SKIN DISORDERS PANEL ¹ HEARING IMPAIRMENT PANEL (INCLUDING GJB2) METABOLIC DISORDERS PANEL INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹
AP1G1	100%	100%	100%	99.6%	Usmani-Riazuddin syndrome, autosomal recessive, 619548;Usmani-Riazuddin syndrome, autosomal dominant, 619467	EPILEPSY PANEL INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹ MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS

AP1S1	100%	100%	100%	98.8%	MEDNIK syndrome, 609313	LIVER DISORDERS PANEL METABOLIC DISORDERS PANEL INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹
AP1S2	100%	99.9%	99.4%	75.7%	Pettigrew syndrome, 304340	INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS
AP1S3	90.6%	90.6%	100%	99%	{Psoriasis 15, pustular, susceptibility to}, 616106	SKIN DISORDERS PANEL ¹ PRIMARY IMMUNODEFICIENCIES PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS

AP2M1	100%	100%	100%	99.4%	Intellectual developmental disorder 60 with seizures, 618587	EPILEPSY PANEL INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS
AP2S1	100%	100%	100%	97.1%	Hypocalciuric hypercalcemia, type III, 600740	INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS RENAL DISORDERS PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS

AP3B1	100%	100%	100%	99.5%	Hermansky-Pudlak syndrome 2, 608233	INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹ VISION DISORDERS PANEL INHERITED BONE MARROW FAILURE AND/OR PREDISPOSITION PANEL TO HEMATOLOGICAL MALIGNANCIES SKIN DISORDERS PANEL ¹ HEMOSTATIC/THROMB OTIC DISORDERS PANEL PRIMARY IMMUNODEFICIENCIES PANEL
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AP3B2	100%	100%	100%	98.9%	Developmental and epileptic encephalopathy 48, 617276	EPILEPSY PANEL METABOLIC DISORDERS PANEL INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹
AP3D1	100%	100%	100%	99.1%	?Hermansky-Pudlak syndrome 10, 617050	VISION DISORDERS PANEL HEMOSTATIC/THROMB OTIC DISORDERS PANEL PRIMARY IMMUNODEFICIENCIES PANEL INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹

AP4B1	100%	100%	100%	99.4%	Spastic paraplegia 47, autosomal recessive, 614066	MOVEMENT DISORDERS PANEL INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL¹
AP4E1	100%	100%	100%	99.4%	Stuttering, familial persistent, 1, 184450;Spastic paraplegia 51, autosomal recessive, 613744	MOVEMENT DISORDERS PANEL INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL¹

AP4M1	100%	100%	100%	99%	Spastic paraplegia 50, autosomal recessive, 612936	MOVEMENT DISORDERS PANEL INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL¹
AP4S1	87.8%	87.3%	100%	99.4%	Spastic paraplegia 52, autosomal recessive, 614067	MOVEMENT DISORDERS PANEL INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL¹
AP5B1	100%	100%	100%	99.4%		VISION DISORDERS PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS

AP5M1	100%	100%	100%	99.6%		VISION DISORDERS PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS
AP5Z1	100%	100%	100%	99%	Spastic paraplegia 48, autosomal recessive, 613647	MOVEMENT DISORDERS PANEL VISION DISORDERS PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹
APC	100%	100%	100%	99.4%	Colorectal cancer, somatic, 114500;Brain tumor-polypoid syndrome 2, 175100;Desmoid disease, hereditary, 135290;Adenoma, periampullary, somatic, 175100;Hepatoblastoma, somatic, 114550;Gastric cancer, somatic, 613659;Gastric adenocarcinoma and proximal polyposis of the stomach, 619182;Gardner syndrome, 175100;Adenomatous polyposis coli, 175100	PANEL HEREDITARY COLORECTAL AND POLYPOSIS SKIN DISORDERS PANEL ¹ MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS HEREDITARY CANCER PANEL

APC2	100%	99.9%	100%	98.6%	Cortical dysplasia, complex, with other brain malformations 10, 618677;Intellectual developmental disorder, autosomal recessive 74, 617169	TALL STATURE PANEL WITH GENOME WIDE CNV ANALYSIS SHORT STATURE/SKELETAL DYSPLASIA PANEL WITH GENOME WIDE CNV ANALYSIS INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹
APCDD1	100%	100%	100%	99.1%	Hypotrichosis 1, 605389	SKIN DISORDERS PANEL ¹ MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS
APOA1	100%	100%	100%	99.4%	Hypoalphalipoproteinemia, primary, 2, 618463;Amyloidosis, hereditary systemic 3, 620657;Hypoalphalipoproteinemia, primary, 2, intermediate, 619836	MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS

APOA2	100%	100%	100%	98.5%	Apolipoprotein A-II deficiency;{Hypercholesterolemia, familial, modifier of}, 143890	MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS
APOA5	100%	100%	100%	99.4%	Hyperchylomicronemia, late-onset, 144650;{Hypertriglyceridemia, susceptibility to}, 145750	METABOLIC DISORDERS PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS
APOB	100%	100%	100%	99.5%	Hypercholesterolemia, familial, 2, 144010;Hypobetalipoproteinemia, 615558	MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS
APOC2	100%	100%	100%	97.1%	Hyperlipoproteinemia, type Ib, 207750	METABOLIC DISORDERS PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹
APOC3	100%	100%	100%	99.5%	Apolipoprotein C-III deficiency, 614028	MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS

APOE	100%	100%	100%	98.6%	Alzheimer disease 2, 104310;Sea-blue histiocyte disease, 269600;{?Alzheimer disease, protection against, due to APOE3-Christchurch}, 607822;{Coronary artery disease, severe, susceptibility to}, 617347;Lipoprotein glomerulopathy, 611771;{?Macular degeneration, age-related}, 603075;Hyperlipoproteinemia, type III, 617347	MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹
APOL1	100%	100%	100%	99.2%	{Glomerulosclerosis, focal segmental, 4, susceptibility to}, 612551	PRIMARY IMMUNODEFICIENCIES PANEL RENAL DISORDERS PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS
APOLD1	100%	100%	100%	96.8%	?Bleeding disorder, vascular-type, 620715	HEMOSTATIC/THROMB OTIC DISORDERS PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS
APOO	100%	99.9%	99%	67.6%		MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS MITOCHONDRIAL DISORDERS PANEL

APP	100%	100%	100%	98.9%	Cerebral amyloid angiopathy, Dutch, Italian, Iowa, Flemish, Arctic variants, 605714;Alzheimer disease 1, familial, 104300	MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS
APRT	100%	100%	100%	99.5%	Adenine phosphoribosyltransferase deficiency, 614723	METABOLIC DISORDERS PANEL RENAL DISORDERS PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹
APTX	100%	100%	100%	99.2%	Ataxia, early-onset, with oculomotor apraxia and hypoalbuminemia, 208920	MOVEMENT DISORDERS PANEL POLYNEUROPATHIES PANEL ¹ INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS MITOCHONDRIAL DISORDERS PANEL COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹

AQP2	100%	100%	100%	99.7%	Diabetes insipidus, nephrogenic, 2, 125800	RENAL DISORDERS PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹
AQP4	100%	100%	100%	99.9%	?Megalencephalic leukoencephalopathy with subcortical cysts 4, remitting, 620448	MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS
AQP5	100%	100%	100%	97.5%	Palmoplantar keratoderma, Bothnian type, 600231	SKIN DISORDERS PANEL ¹ MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS
AR	100%	99.5%	97.9%	64.8%	Androgen insensitivity, partial, with or without breast cancer, 312300;Spinal and bulbar muscular atrophy, X-linked 1, 313200;{Prostate cancer, susceptibility to}, 301120;Androgen insensitivity, 300068;Hypospadias 1, X-linked, 300633	DISORDERS/DIFFEREN CES OF SEX DEVELOPMENT (DSD) / PRIMARY ADRENAL INSUFFICIENCY PANEL MALE INFERTILITY PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS

ARAF	100%	99.3%	98.4%	65.5%		LYMPHATIC ANOMALIES PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS
ARCN1	100%	100%	100%	99.4%	Short stature-micrognathia syndrome, 617164	SHORT STATURE/SKELETAL DYSPLASIA PANEL WITH GENOME WIDE CNV ANALYSIS LIVER DISORDERS PANEL INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS
ARF1	100%	100%	100%	99.2%	Periventricular nodular heterotopia 8, 618185	INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS

ARF3	100%	100%	100%	99%		INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS
ARFGEF1	100%	100%	100%	99.3%	Developmental delay, impaired speech, and behavioral abnormalities, with or without seizures, 619964	INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS
ARFGEF2	100%	100%	100%	99.2%	Periventricular heterotopia with microcephaly, 608097	INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹

ARG1	93%	93%	100%	99.6%	Argininemia, 207800	MOVEMENT DISORDERS PANEL METABOLIC DISORDERS PANEL INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹
ARHGAP24	100%	100%	100%	99%		RENAL DISORDERS PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS
ARHGAP26	100%	100%	100%	99%	Leukemia, juvenile myelomonocytic, somatic, 607785	MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS

ARHGAP29	100%	100%	100%	99.6%		CRANIOFACIAL ANOMALIES PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS OROFACIAL CLEFTING PANEL WITH GENOME WIDE CNV ANALYSIS
ARHGAP31	100%	100%	100%	99.3%	Adams-Oliver syndrome 1, 100300	SKIN DISORDERS PANEL ¹ SHORT STATURE/SKELETAL DYSPLASIA PANEL WITH GENOME WIDE CNV ANALYSIS INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS OROFACIAL CLEFTING PANEL WITH GENOME WIDE CNV ANALYSIS

ARHGAP35	100%	100%	100%	99.2%		DISORDERS/DIFFERENCES OF SEX DEVELOPMENT (DSD) / PRIMARY ADRENAL INSUFFICIENCY PANEL HYPOGONADOTROPIC HYPOGONADISM (KALLMANN) PANEL INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS
ARHGDIA	100%	100%	100%	99.3%	Nephrotic syndrome, type 8, 615244	RENAL DISORDERS PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹
ARHGEF1	100%	99.6%	100%	97.5%	?Immunodeficiency 62, 618459	PRIMARY IMMUNODEFICIENCIES PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS

ARHGEF10	100%	100%	100%	99.4%	?Slowed nerve conduction velocity, AD, 608236	POLYNEUROPATHIES PANEL ¹ MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS
ARHGEF18	100%	99.9%	100%	98.4%	Retinitis pigmentosa 78, 617433	VISION DISORDERS PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹
ARHGEF2	97.9%	97.9%	100%	98.7%	?Neurodevelopmental disorder with midbrain and hindbrain malformations, 617523	MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹
ARHGEF28	100%	100%	100%	99.2%		POLYNEUROPATHIES PANEL ¹ MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS

ARHGEF6	100%	100%	99%	70.6%		INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS RENAL DISORDERS PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS
ARHGEF9	96.9%	96.7%	98.5%	67.9%	Developmental and epileptic encephalopathy 8, 300607	EPILEPSY PANEL INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS

ARID1A	100%	100%	100%	97.6%	Coffin-Siris syndrome 2, 614607	MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS SKIN DISORDERS PANEL ¹ SHORT STATURE/SKELETAL DYSPLASIA PANEL WITH GENOME WIDE CNV ANALYSIS INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS
ARID1B	98.4%	98.1%	99.8%	93%	Coffin-Siris syndrome 1, 135900	SHORT STATURE/SKELETAL DYSPLASIA PANEL WITH GENOME WIDE CNV ANALYSIS SKIN DISORDERS PANEL ¹ EPILEPSY PANEL INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS

ARID2	100%	100%	100%	99.3%	Coffin-Siris syndrome 6, 617808	INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS
ARIH1	100%	100%	100%	99.5%		THORACIC AORTIC ANEURYSM AND/OR DISSECTION PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS
ARL13B	93.4%	93%	100%	98.9%	Joubert syndrome 8, 612291	VISION DISORDERS PANEL CILIOPATHIES PANEL INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS RENAL DISORDERS PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹

ARL2	100%	100%	100%	99.4%	?Microcornea, rod-cone dystrophy, cataract, and posterior staphyloma 1, 619082	VISION DISORDERS PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS MITOCHONDRIAL DISORDERS PANEL
ARL2BP	100%	100%	100%	99.6%	Retinitis pigmentosa 82 with or without situs inversus, 615434	VISION DISORDERS PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹
ARL3	100%	100%	100%	99.2%	Retinitis pigmentosa 83, 618173;Joubert syndrome 35, 618161	VISION DISORDERS PANEL CILIOPATHIES PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹

ARL6	100%	100%	100%	99.7%	Retinitis pigmentosa 55, 613575;{Bardet-Biedl syndrome 1, modifier of}, 209900;Bardet-Biedl syndrome 3, 600151	VISION DISORDERS PANEL COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹ CILIOPATHIES PANEL HYPOGONADOTROPIC HYPOGONADISM (KALLMANN) PANEL INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS RENAL DISORDERS PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS
ARL6IP1	100%	100%	100%	99.7%	Spastic paraplegia 61, autosomal recessive, 615685	MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹
ARMC12	100%	100%	100%	99.5%	Spermatogenic failure 90, 620744	MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS

ARMC2	100%	100%	100%	99.3%	Spermatogenic failure 38, 618433	MALE INFERTILITY PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS
ARMC5	100%	100%	100%	98.9%	{ACTH-independent macronodular adrenal hyperplasia 2}, 615954	DISORDERS/DIFFEREN CES OF SEX DEVELOPMENT (DSD) / PRIMARY ADRENAL INSUFFICIENCY PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS HEREDITARY CANCER PANEL
ARMC9	96.2%	93.8%	100%	99.4%	Joubert syndrome 30, 617622	CILIOPATHIES PANEL INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹

ARNT2	100%	99.9%	100%	99.2%	?Webb-Dattani syndrome, 615926	MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS MITOCHONDRIAL DISORDERS PANEL COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL¹
ARPC1B	100%	100%	100%	98.7%	Immunodeficiency 71 with inflammatory disease and congenital thrombocytopenia, 617718	HEMOSTATIC/THROMB OTIC DISORDERS PANEL PRIMARY IMMUNODEFICIENCIES PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL¹
ARPC4	100%	100%	100%	99.7%	Developmental delay, language impairment, and ocular abnormalities, 620141	INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS

ARPC5	100%	100%	100%	99%	Immunodeficiency 133 with autoimmunity and autoinflammation, 620565	PRIMARY IMMUNODEFICIENCIES PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL¹
ARR3	100%	100%	98.1%	65.9%	Myopia 26, X-linked, female-limited, 301010	VISION DISORDERS PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS

ARSA	100%	100%	100%	99.1%	Metachromatic leukodystrophy, 250100	MOVEMENT DISORDERS PANEL POLYNEUROPATHIES PANEL ¹ METABOLIC DISORDERS PANEL INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹
ARSB	100%	100%	100%	99.3%	Mucopolysaccharidosis type VI (Maroteaux-Lamy), 253200	SHORT STATURE/SKELETAL DYSPLASIA PANEL WITH GENOME WIDE CNV ANALYSIS METABOLIC DISORDERS PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹

ARSG	100%	100%	100%	99.1%	Usher syndrome, type IV, 618144	VISION DISORDERS PANEL HEARING IMPAIRMENT PANEL (INCLUDING GJB2) MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹
ARSK	100%	100%	100%	99.4%	Mucopolysaccharidosis, type X, 619698	SHORT STATURE/SKELETAL DYSPLASIA PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹

ARSL	100%	99.7%	98.8%	65.8%	Chondrodysplasia punctata, X-linked recessive, 302950	SHORT STATURE/SKELETAL DYSPLASIA PANEL WITH GENOME WIDE CNV ANALYSIS INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS
ARV1	100%	100%	100%	99.4%	Developmental and epileptic encephalopathy 38, 617020	EPILEPSY PANEL INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹

ARX	96.9%	92.6%	92.9%	54%	Proud syndrome, 300004;Hydranencephaly with abnormal genitalia, 300215;Partington syndrome, 309510;Developmental and epileptic encephalopathy 1, 308350;Lissencephaly, X-linked 2, 300215;Intellectual developmental disorder, X-linked 29, 300419	MOVEMENT DISORDERS PANEL DISORDERS/DIFFERENCES OF SEX DEVELOPMENT (DSD) / PRIMARY ADRENAL INSUFFICIENCY PANEL EPILEPSY PANEL INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS
ASAH1	100%	100%	100%	99%	Spinal muscular atrophy with progressive myoclonic epilepsy, 159950;Farber lipogranulomatosis, 228000	EPILEPSY PANEL METABOLIC DISORDERS PANEL INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹

ASB10	100%	100%	100%	99.1%	Glaucoma 1, open angle, F, 603383	VISION DISORDERS PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS
ASCC1	86.6%	86.6%	100%	99.3%	Spinal muscular atrophy with congenital bone fractures 2, 616867;Barrett esophagus/esophageal adenocarcinoma, 614266	FETAL AKINESIA PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹ MUSCLE DISORDERS PANEL
ASCC3	100%	100%	100%	99.6%	Intellectual developmental disorder, autosomal recessive 81, 620700	MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹
ASCL1	100%	100%	100%	93.7%		MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS

ASH1L	100%	100%	100%	99.2%	Intellectual developmental disorder, autosomal dominant 52, 617796	INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS
ASIP	100%	100%	100%	98%	[Skin/hair/eye pigmentation 9, brown/nonbrown eyes], 611742;[Skin/hair/eye pigmentation 9, dark/light hair], 611742	SKIN DISORDERS PANEL ¹ MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS
ASL	100%	100%	100%	98.8%	Argininosuccinic aciduria, 207900	SKIN DISORDERS PANEL ¹ EPILEPSY PANEL METABOLIC DISORDERS PANEL INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹

ASNS	100%	100%	100%	99.7%	Asparagine synthetase deficiency, 615574	EPILEPSY PANEL METABOLIC DISORDERS PANEL INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹
ASPA	100%	100%	100%	99.6%	Canavan disease, 271900	MOVEMENT DISORDERS PANEL METABOLIC DISORDERS PANEL INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹

ASPH	100%	100%	100%	99.3%	Traboulsi syndrome, 601552	THORACIC AORTIC ANEURYSM AND/OR DISSECTION PANEL VISION DISORDERS PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹
ASPM	97.9%	97.9%	100%	99.5%	Microcephaly 5, primary, autosomal recessive, 608716	INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹
ASPRV1	100%	100%	100%	99.4%	Ichthyosis, lamellar, autosomal dominant, 146750	SKIN DISORDERS PANEL ¹ MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS

ASPSR1	100%	100%	100%	98.7%	Alveolar soft-part sarcoma, 606243	MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS
ASRGL1	100%	100%	100%	99.5%		VISION DISORDERS PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS
ASS1	100%	100%	100%	98.9%	Citrullinemia, 215700	METABOLIC DISORDERS PANEL INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹

ASTL	100%	100%	100%	99.5%	?Oocyte/zygote/embryo maturation arrest 11, 619643	UNSUCCESSFUL IVF TREATMENTS, RECURRENT MOLAR PREGNANCIES AND/OR RECURRENT PREGNANCY LOSS PANEL ¹ MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS
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ASXL1	100%	100%	100%	99.2%	Myelodysplastic syndrome, somatic, 614286;Bohring-Opitz syndrome, 605039	INHERITED BONE MARROW FAILURE AND/OR PREDISPOSITION PANEL TO HEMATOLOGICAL MALIGNANCIES SKIN DISORDERS PANEL ¹ PRIMARY IMMUNODEFICIENCIES PANEL SHORT STATURE/SKELETAL DYSPLASIA PANEL WITH GENOME WIDE CNV ANALYSIS INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS OROFACIAL CLEFTING PANEL WITH GENOME WIDE CNV ANALYSIS HEREDITARY CANCER PANEL
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ASXL2	100%	100%	100%	99.5%	Shashi-Pena syndrome, 617190	INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS
ASXL3	100%	100%	100%	99.2%	Bainbridge-Ropers syndrome, 615485	SKIN DISORDERS PANEL ¹ EPILEPSY PANEL INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS
ATAD1	100%	100%	100%	99.7%	Hyperekplexia 4, 618011	INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS MITOCHONDRIAL DISORDERS PANEL COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹

ATAD3A	100%	100%	100%	98.6%	Harel-Yoon syndrome, 617183;Pontocerebellar hypoplasia, hypotonia, and respiratory insufficiency syndrome, neonatal lethal, 618810	POLYNEUROPATHIES PANEL ¹ PRIMARY IMMUNODEFICIENCIES PANEL INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS MITOCHONDRIAL DISORDERS PANEL COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹
ATAD3B	100%	100%	100%	97.6%		MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS MITOCHONDRIAL DISORDERS PANEL

ATCAY	100%	100%	100%	98.4%	Ataxia, cerebellar, Cayman type, 601238	MOVEMENT DISORDERS PANEL INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹
ATF3	100%	100%	100%	99.6%		DISORDERS/DIFFEREN CES OF SEX DEVELOPMENT (DSD) / PRIMARY ADRENAL INSUFFICIENCY PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS
ATF6	90.9%	90.9%	100%	99%	Achromatopsia 7, 616517	VISION DISORDERS PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹

ATG4A	100%	100%	99.4%	71.1%		PRIMARY IMMUNODEFICIENCIES PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS
ATG4D	100%	100%	100%	98%		INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS
ATG5	100%	100%	100%	99.9%	?Spinocerebellar ataxia, autosomal recessive 25, 617584	MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹

ATG7	100%	100%	100%	99.5%	Spinocerebellar ataxia, autosomal recessive 31, 619422	MOVEMENT DISORDERS PANEL INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹
ATIC	100%	100%	100%	99.5%	AICA-ribosiduria due to ATIC deficiency, 608688	SKIN DISORDERS PANEL ¹ METABOLIC DISORDERS PANEL INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹

ATL1	100%	100%	100%	99.5%	Spastic paraplegia 3A, autosomal dominant, 182600;Neuropathy, hereditary sensory, type ID, 613708	MOVEMENT DISORDERS PANEL INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS POLYNEUROPATHIES PANEL ¹ NEUROLOGICAL PAIN DISORDERS PANEL ¹
ATL3	100%	100%	100%	99.3%	Neuropathy, hereditary sensory, type IF, 615632	POLYNEUROPATHIES PANEL ¹ NEUROLOGICAL PAIN DISORDERS PANEL ¹ MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS

ATM	100%	100%	100%	99.4%	Lymphoma, B-cell non-Hodgkin, somatic;Ataxia-telangiectasia, 208900;{Breast cancer, susceptibility to}, 114480;T-cell prolymphocytic leukemia, somatic;Lymphoma, mantle cell, somatic	MOVEMENT DISORDERS PANEL PRIMARY IMMUNODEFICIENCIES PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS PANEL HEREDITARY PANCREATIC CANCER (BRCA1, BRCA2, PALB2, ATM, CDKN2A, MLH1, MSH2, MSH6, PMS2, STK11) COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹ HEREDITARY CANCER PANEL
ATN1	100%	100%	100%	96.4%	Dentatorubral-pallidoluysian atrophy, 125370;Congenital hypotonia, epilepsy, developmental delay, and digital anomalies, 618494	MOVEMENT DISORDERS PANEL EPILEPSY PANEL INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS

ATOH1	100%	100%	100%	99.4%	?Deafness, autosomal dominant 89, 620284	HEARING IMPAIRMENT PANEL (INCLUDING GJB2) MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS
ATOH7	100%	96.3%	100%	99.2%	Persistent hyperplastic primary vitreous, autosomal recessive, 221900	VISION DISORDERS PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹
ATP11A	100%	100%	100%	99.4%	?Auditory neuropathy, autosomal dominant 2, 620384;?Leukodystrophy, hypomyelinating, 24, 619851;Deafness, autosomal dominant 84, 619810	HEARING IMPAIRMENT PANEL (INCLUDING GJB2) MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS
ATP11C	100%	100%	98.9%	70%	?Hemolytic anemia, congenital, X-linked, 301015	MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS

ATP13A2	100%	100%	100%	98.9%	Spastic paraplegia 78, autosomal recessive, 617225;Kufor-Rakeb syndrome, 606693	MOVEMENT DISORDERS PANEL POLYNEUROPATHIES PANEL ¹ INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS MITOCHONDRIAL DISORDERS PANEL PARKINSON DISEASE PANEL COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹
ATP13A3	100%	100%	100%	99.6%	Pulmonary hypertension, primary, 5, 265400	MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹

ATP1A1	100%	100%	100%	99.4%	Hypomagnesemia, seizures, and impaired intellectual development 2, 618314;Charcot-Marie-Tooth disease, axonal, type 2DD, 618036	HEARING IMPAIRMENT PANEL (INCLUDING GJB2) EPILEPSY PANEL POLYNEUROPATHIES PANEL ¹ METABOLIC DISORDERS PANEL INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS RENAL DISORDERS PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS
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ATP1A2	100%	100%	100%	98.9%	Developmental and epileptic encephalopathy 98, 619605;Fetal akinesia, respiratory insufficiency, microcephaly, polymicrogyria, and dysmorphic facies, 619602;Alternating hemiplegia of childhood 1, 104290;Migraine, familial basilar, 602481;Migraine, familial hemiplegic, 2, 602481	FETAL AKINESIA PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹ EPILEPSY PANEL INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MOVEMENT DISORDERS PANEL
ATP1A3	100%	100%	100%	98.3%	Alternating hemiplegia of childhood 2, 614820;Dystonia-12, 128235;CAPOS syndrome, 601338;Developmental and epileptic encephalopathy 99, 619606	MOVEMENT DISORDERS PANEL PARKINSON DISEASE PANEL INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS HEARING IMPAIRMENT PANEL (INCLUDING GJB2) EPILEPSY PANEL

ATP2A1	100%	100%	100%	98.2%	Brody myopathy, 601003	MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹ MUSCLE DISORDERS PANEL
ATP2A2	100%	100%	100%	99.3%	Acrokeratosis verruciformis, 101900;Darier disease, 124200	SKIN DISORDERS PANEL ¹ INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS MUSCLE DISORDERS PANEL
ATP2B1	100%	100%	100%	99.3%	Intellectual developmental disorder, autosomal dominant 66, 619910	INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS

ATP2B2	100%	100%	100%	98.8%	Deafness, autosomal dominant 82, 619804;{Deafness, autosomal recessive 12, modifier of}, 601386	HEARING IMPAIRMENT PANEL (INCLUDING GJB2) INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS
ATP2B3	100%	99.5%	98.2%	65.8%	?Spinocerebellar ataxia, X-linked 1, 302500	MOVEMENT DISORDERS PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS
ATP2C1	100%	100%	100%	99.4%	Hailey-Hailey disease, 169600	SKIN DISORDERS PANEL ¹ MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS

ATP5F1A	100%	100%	100%	99.6%	Mitochondrial complex V (ATP synthase) deficiency, nuclear type 4A, 620358;?Combined oxidative phosphorylation deficiency 22, 616045;?Mitochondrial complex V (ATP synthase) deficiency, nuclear type 4B, encephalopathic type, 615228	INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS MITOCHONDRIAL DISORDERS PANEL COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹
ATP5F1B	100%	100%	100%	99.2%	?Hypermetabolism due to uncoupled mitochondrial oxidative phosphorylation 2, 620085	MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS MITOCHONDRIAL DISORDERS PANEL
ATP5F1C	100%	100%	100%	99.8%		MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS MITOCHONDRIAL DISORDERS PANEL

ATP5F1D	100%	100%	100%	97.8%	Mitochondrial complex V (ATP synthase) deficiency, 618120	MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS MITOCHONDRIAL DISORDERS PANEL COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹
ATP5F1E	100%	100%	100%	99%	Mitochondrial complex V (ATP synthase) deficiency, nuclear type 3, 614053	MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS MITOCHONDRIAL DISORDERS PANEL COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹
ATP5IF1	100%	100%	100%	99.2%		MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS MITOCHONDRIAL DISORDERS PANEL
ATP5MC1	100%	100%	100%	98.1%		MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS MITOCHONDRIAL DISORDERS PANEL

ATP5MC2	100%	100%	100%	99.6%		MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS MITOCHONDRIAL DISORDERS PANEL
ATP5MC3	100%	100%	100%	99.5%	Dystonia, early-onset, and/or spastic paraplegia, 619681	MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS MITOCHONDRIAL DISORDERS PANEL
ATP5ME	100%	100%	100%	98.8%		MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS MITOCHONDRIAL DISORDERS PANEL
ATP5MF	100%	100%	100%	98.8%		MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS MITOCHONDRIAL DISORDERS PANEL

ATP5MG	95.4%	95.3%	100%	98.7%		MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS MITOCHONDRIAL DISORDERS PANEL
ATP5MGL	100%	100%	100%	98.3%		MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS MITOCHONDRIAL DISORDERS PANEL
ATP5MK	100%	100%	100%	99.6%	Mitochondrial complex V (ATP synthase) deficiency, nuclear type 6, 618683	MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS MITOCHONDRIAL DISORDERS PANEL
ATP5PB	100%	100%	100%	99.5%		MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS MITOCHONDRIAL DISORDERS PANEL

ATP5PD	100%	100%	100%	99%		MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS MITOCHONDRIAL DISORDERS PANEL
ATP5PF	100%	100%	100%	99.8%		MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS MITOCHONDRIAL DISORDERS PANEL
ATP5PO	100%	100%	100%	99.6%	Mitochondrial complex V (ATP synthase) deficiency, nuclear type 7, 620359	INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS MITOCHONDRIAL DISORDERS PANEL COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹

ATP6AP1	100%	99.8%	99%	67.3%	Immunodeficiency 47, 300972	PRIMARY IMMUNODEFICIENCIES PANEL METABOLIC DISORDERS PANEL INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS
ATP6AP2	100%	99.9%	99.2%	71.8%	Intellectual developmental disorder, X-linked syndromic, Hedera type, 300423;?Parkinsonism with spasticity, X-linked, 300911;Congenital disorder of glycosylation, type IIr, 301045	MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS EPILEPSY PANEL METABOLIC DISORDERS PANEL INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS

ATP6V0A1	92.9%	92.8%	100%	99.3%	Neurodevelopmental disorder with epilepsy and brain atrophy, 619971;Developmental and epileptic encephalopathy 104, 619970	EPILEPSY PANEL INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS
ATP6V0A2	100%	100%	100%	99.6%	Wrinkly skin syndrome, 278250;Cutis laxa, autosomal recessive, type IIA, 219200	SKIN DISORDERS PANEL ¹ MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹ SHORT STATURE/SKELETAL DYSPLASIA PANEL WITH GENOME WIDE CNV ANALYSIS METABOLIC DISORDERS PANEL INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS

ATP6V0A4	100%	100%	100%	98.4%	Distal renal tubular acidosis 3, with or without sensorineural hearing loss, 602722	HEARING IMPAIRMENT PANEL (INCLUDING GJB2) RENAL DISORDERS PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹
ATP6V0C	100%	100%	100%	98.5%	Epilepsy, early-onset, 3, with or without developmental delay, 620465	EPILEPSY PANEL INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS

ATP6V1A	100%	100%	100%	99.6%	Cutis laxa, autosomal recessive, type IID, 617403;Developmental and epileptic encephalopathy 93, 618012	EPILEPSY PANEL METABOLIC DISORDERS PANEL INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹
ATP6V1B1	100%	100%	100%	99.3%	Distal renal tubular acidosis 2 with progressive sensorineural hearing loss, 267300	HEARING IMPAIRMENT PANEL (INCLUDING GJB2) RENAL DISORDERS PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹

ATP6V1B2	100%	100%	100%	99.6%	Zimmermann-Laband syndrome 2, 616455;Deafness, congenital, with onychodystrophy, autosomal dominant, 124480	HEARING IMPAIRMENT PANEL (INCLUDING GJB2) INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS
ATP6V1E1	100%	100%	100%	99.5%	Cutis laxa, autosomal recessive, type IIC, 617402	METABOLIC DISORDERS PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL¹

ATP7A	94.9%	94.5%	99.3%	69.7%	Occipital horn syndrome, 304150;Neuronopathy, distal hereditary motor, X-linked, 300489;Menkes disease, 309400	SKIN DISORDERS PANEL ¹ INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS MUSCLE DISORDERS PANEL EPILEPSY PANEL POLYNEUROPATHIES PANEL ¹ METABOLIC DISORDERS PANEL
ATP7B	100%	100%	100%	99.4%	Wilson disease, 277900	MOVEMENT DISORDERS PANEL LIVER DISORDERS PANEL METABOLIC DISORDERS PANEL RENAL DISORDERS PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹

ATP8A2	100%	100%	100%	99.4%	Cerebellar ataxia, impaired intellectual development, and dysequilibrium syndrome 4, 615268	INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹
ATP8B1	100%	100%	100%	99.2%	Cholestasis, progressive familial intrahepatic 1, 211600;Cholestasis, intrahepatic, of pregnancy, 1, 147480;Cholestasis, benign recurrent intrahepatic, 243300	LIVER DISORDERS PANEL METABOLIC DISORDERS PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹

ATP9A	100%	100%	100%	98.8%	Neurodevelopmental disorder with poor growth and behavioral abnormalities, 620242	INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹
ATPAF1	100%	100%	100%	96.6%		MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS MITOCHONDRIAL DISORDERS PANEL
ATPAF2	100%	100%	100%	99.6%	?Mitochondrial complex V (ATP synthase) deficiency, nuclear type 1, 604273	HEART DISORDERS PANEL ¹ MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS MITOCHONDRIAL DISORDERS PANEL COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹

ATR	100%	100%	99.7%	98.3%	Seckel syndrome 1, 210600;?Cutaneous telangiectasia and cancer syndrome, familial, 614564	INHERITED BONE MARROW FAILURE AND/OR PREDISPOSITION PANEL TO HEMATOLOGICAL MALIGNANCIES SKIN DISORDERS PANEL ¹ COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹ HEREDITARY CANCER PANEL SHORT STATURE/SKELETAL DYSPLASIA PANEL WITH GENOME WIDE CNV ANALYSIS INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS
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ATRX	100%	99.9%	98.9%	69.8%	Alpha-thalassemia myelodysplasia syndrome, somatic, 300448;Intellectual disability-hypotonic facies syndrome, X-linked, 309580;Alpha-thalassemia/impaired intellectual development syndrome, 301040	DISORDERS/DIFFERENCES OF SEX DEVELOPMENT (DSD) / PRIMARY ADRENAL INSUFFICIENCY PANEL EPILEPSY PANEL INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS
ATXN1	100%	100%	100%	98.8%	Spinocerebellar ataxia 1, 164400	MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS
ATXN10	100%	100%	100%	99.3%	Spinocerebellar ataxia 10, 603516	MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS
ATXN2	100%	100%	100%	98.7%	{Amyotrophic lateral sclerosis, susceptibility to, 13}, 183090;Spinocerebellar ataxia 2, 183090;{Parkinson disease, late-onset, susceptibility to}, 168600	MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS

ATXN2L	100%	100%	100%	97.3%		INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS
ATXN3	96.9%	96.9%	100%	99.2%	{Parkinson disease, late-onset, susceptibility to}, 168600;Machado-Joseph disease, 109150	MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS
ATXN7	100%	99.5%	100%	97.6%	Spinocerebellar ataxia 7, 164500	MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS
ATXN7L3	100%	100%	100%	99.3%		INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS
ATXN8OS					{Parkinson disease, susceptibility to}, 168600;Spinocerebellar ataxia 8, 608768	MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS

AUH	100%	100%	100%	99.7%	3-methylglutaconic aciduria, type I, 250950	VISION DISORDERS PANEL COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL¹ METABOLIC DISORDERS PANEL INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS
AURKC	100%	100%	100%	98.4%	Spermatogenic failure 5, 243060	MALE INFERTILITY PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL¹

AUTS2	100%	100%	100%	98.4%	Intellectual developmental disorder, autosomal dominant 26, 615834	EPILEPSY PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS
AVIL	100%	100%	100%	99.6%	Nephrotic syndrome, type 21, 618594	RENAL DISORDERS PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹
AVP	100%	100%	100%	99.6%	Diabetes insipidus, neurohypophyseal, 125700	RENAL DISORDERS PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS

AVPR2	100%	100%	99.2%	71.2%	Diabetes insipidus, nephrogenic, 1, 304800;Nephrogenic syndrome of inappropriate antidiuresis, 300539	INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS RENAL DISORDERS PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS
AXIN1	100%	100%	100%	99.1%	Hepatocellular carcinoma, somatic, 114550;Craniometadiaphyseal osteosclerosis with hip dysplasia, 620558;?Caudal duplication anomaly, 607864	SHORT STATURE/SKELETAL DYSPLASIA PANEL WITH GENOME WIDE CNV ANALYSIS INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL¹

AXIN2	100%	100%	100%	98.4%	Colorectal cancer, somatic, 114500;Oligodontia-colorectal cancer syndrome, 608615	CRANIOFACIAL ANOMALIES PANEL WITH GENOME WIDE CNV ANALYSIS PANEL HEREDITARY COLORECTAL AND POLYPOSIS SKIN DISORDERS PANEL ¹ MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS HEREDITARY CANCER PANEL
AXL	100%	100%	100%	98.1%		DISORDERS/DIFFEREN CES OF SEX DEVELOPMENT (DSD) / PRIMARY ADRENAL INSUFFICIENCY PANEL HYPOGONADOTROPIC HYPOGONADISM (KALLMANN) PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS

B2M	100%	100%	100%	99.7%	Amyloidosis, hereditary systemic 6, 620659;Immunodeficiency 43, 241600	POLYNEUROPATHIES PANEL ¹ PRIMARY IMMUNODEFICIENCIES PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹ SEVERE COMBINED IMMUNODEFICIENCY (SCID) PANEL
B3GALNT1	100%	100%	100%	99.8%	[Blood group, P1PK system, P(k) phenotype], 111400;[Blood group, globoside system], 615021	METABOLIC DISORDERS PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS

B3GALNT2	92.6%	92.6%	100%	99%	Muscular dystrophy-dystroglycanopathy (congenital with brain and eye anomalies), type A, 11, 615181	METABOLIC DISORDERS PANEL INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹ MUSCLE DISORDERS PANEL
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B3GALT6	94%	84.7%	100%	97%	Ehlers-Danlos syndrome, spondylodysplastic type, 2, 615349;Spondyloepimetaphyseal dysplasia with joint laxity, type 1, with or without fractures, 271640;Al-Gazali syndrome, 609465	SKIN DISORDERS PANEL ¹ SHORT STATURE/SKELETAL DYSPLASIA PANEL WITH GENOME WIDE CNV ANALYSIS METABOLIC DISORDERS PANEL OROFACIAL CLEFTING PANEL WITH GENOME WIDE CNV ANALYSIS INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹
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B3GAT3	95.8%	93.8%	100%	99.2%	Multiple joint dislocations, short stature, craniofacial dysmorphism, with or without congenital heart defects, 245600	SHORT STATURE/SKELETAL DYSPLASIA PANEL WITH GENOME WIDE CNV ANALYSIS METABOLIC DISORDERS PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹
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B3GLCT	100%	99.9%	100%	99.4%	Peters-plus syndrome, 261540	VISION DISORDERS PANEL SHORT STATURE/SKELETAL DYSPLASIA PANEL WITH GENOME WIDE CNV ANALYSIS OROFACIAL CLEFTING PANEL WITH GENOME WIDE CNV ANALYSIS METABOLIC DISORDERS PANEL INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹
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B4GALNT1	100%	100%	100%	99.1%	Spastic paraplegia 26, autosomal recessive, 609195	MOVEMENT DISORDERS PANEL POLYNEUROPATHIES PANEL ¹ INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹
B4GALNT2	100%	100%	100%	98.8%	[Blood group, Sid system], 615018;Sd(a) polyagglutination syndrome, 615018	MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS

B4GALT1	100%	100%	100%	99.4%	Combined low LDL and fibrinogen, 620364;Congenital disorder of glycosylation, type IId, 607091	MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹ LIVER DISORDERS PANEL METABOLIC DISORDERS PANEL INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS
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B4GALT7	100%	100%	100%	99%	Ehlers-Danlos syndrome, spondylodysplastic type, 1, 130070	SKIN DISORDERS PANEL ¹ SHORT STATURE/SKELETAL DYSPLASIA PANEL WITH GENOME WIDE CNV ANALYSIS OROFACIAL CLEFTING PANEL WITH GENOME WIDE CNV ANALYSIS METABOLIC DISORDERS PANEL INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹
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B4GAT1	100%	100%	100%	98.2%	Muscular dystrophy-dystroglycanopathy (congenital with brain and eye anomalies), type A, 13, 615287	METABOLIC DISORDERS PANEL INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹ MUSCLE DISORDERS PANEL
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B9D1	100%	99.9%	100%	99.3%	?Meckel syndrome 9, 614209;Joubert syndrome 27, 617120	CILIOPATHIES PANEL DISORDERS/DIFFERENCES OF SEX DEVELOPMENT (DSD) / PRIMARY ADRENAL INSUFFICIENCY PANEL INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS RENAL DISORDERS PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL¹
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B9D2	100%	100%	100%	99.9%	?Meckel syndrome 10, 614175;Joubert syndrome 34, 614175	CILIOPATHIES PANEL INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS RENAL DISORDERS PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹ OROFACIAL CLEFTING PANEL WITH GENOME WIDE CNV ANALYSIS
BAAT	100%	100%	100%	99.5%	Bile acid conjugation defect 1, 619232	LIVER DISORDERS PANEL METABOLIC DISORDERS PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹

BACH2	100%	100%	100%	98.5%	Immunodeficiency 60 and autoimmunity, 618394	PRIMARY IMMUNODEFICIENCIES PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS
BAG3	100%	100%	100%	99.2%	Cardiomyopathy, dilated, 1HH, 613881;Myopathy, myofibrillar, 6, 612954	DILATED CARDIOMYOPATHY PANEL ¹ MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS MUSCLE DISORDERS PANEL POLYNEUROPATHIES PANEL ¹ HEART DISORDERS PANEL ¹
BAG5	100%	100%	100%	97.4%	Cardiomyopathy, dilated, 2F, 619747	MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS

BANF1	100%	100%	100%	97.5%	Nestor-Guillermo progeria syndrome, 614008	SKIN DISORDERS PANEL ¹ HEART DISORDERS PANEL ¹ SHORT STATURE/SKELETAL DYSPLASIA PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹
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BAP1	100%	100%	100%	99%	Kury-Isidor syndrome, 619762;Tumor predisposition syndrome 1, 614327;{Uveal melanoma, susceptibility to, 2}, 606661	SKIN DISORDERS PANEL ¹ MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS PANEL HEREDITARY RENALCANCER HEREDITARY CANCER PANEL PANEL MELANOMA, EXTENSIVE (CDKN2A, CDK4, MITF P.(GLU318LYS), BAP1, POT1, TERT PROMOTER) ¹ INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS
BARD1	100%	100%	100%	99.6%	{Breast cancer, susceptibility to}, 114480	MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS HEREDITARY CANCER PANEL
BAX	100%	100%	100%	94.6%	Colorectal cancer, somatic, 114500;T-cell acute lymphoblastic leukemia, somatic, 613065	MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS

BAZ2B	100%	100%	99.9%	98.2%		INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS
BBIP1	100%	100%	100%	99.6%	Bardet-Biedl syndrome 18, 615995	VISION DISORDERS PANEL CILIOPATHIES PANEL RENAL DISORDERS PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL¹

BBS1	100%	100%	100%	98.4%	Bardet-Biedl syndrome 1, 209900	VISION DISORDERS PANEL CILIOPATHIES PANEL HYPOGONADOTROPIC HYPOGONADISM (KALLMANN) PANEL INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS RENAL DISORDERS PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL¹
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BBS10	100%	100%	100%	99.5%	Bardet-Biedl syndrome 10, 615987	VISION DISORDERS PANEL CILIOPATHIES PANEL HYPOGONADOTROPIC HYPOGONADISM (KALLMANN) PANEL INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS RENAL DISORDERS PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL¹
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BBS12	100%	100%	100%	99.7%	Bardet-Biedl syndrome 12, 615989	VISION DISORDERS PANEL CILIOPATHIES PANEL HYPOGONADOTROPIC HYPOGONADISM (KALLMANN) PANEL INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS RENAL DISORDERS PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹
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BBS2	98%	98%	100%	99.6%	Retinitis pigmentosa 74, 616562;Bardet-Biedl syndrome 2, 615981	VISION DISORDERS PANEL RENAL DISORDERS PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹ CILIOPATHIES PANEL HYPOGONADOTROPIC HYPOGONADISM (KALLMANN) PANEL INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS
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BBS4	100%	100%	100%	99.7%	Bardet-Biedl syndrome 4, 615982	VISION DISORDERS PANEL CILIOPATHIES PANEL HYPOGONADOTROPIC HYPOGONADISM (KALLMANN) PANEL INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS RENAL DISORDERS PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹
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BBS5	100%	100%	100%	99.5%	Bardet-Biedl syndrome 5, 615983	VISION DISORDERS PANEL CILIOPATHIES PANEL HYPOGONADOTROPIC HYPOGONADISM (KALLMANN) PANEL INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS RENAL DISORDERS PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹
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BBS7	100%	100%	100%	99.7%	Bardet-Biedl syndrome 7, 615984	VISION DISORDERS PANEL CILIOPATHIES PANEL HYPOGONADOTROPIC HYPOGONADISM (KALLMANN) PANEL INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS RENAL DISORDERS PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL¹
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BBS9	95.8%	95.8%	100%	99.6%	Bardet-Biedl syndrome 9, 615986	COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹ VISION DISORDERS PANEL CILIOPATHIES PANEL INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS RENAL DISORDERS PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS
BCAP31	98.2%	87%	98.8%	68.1%	Deafness, dystonia, and cerebral hypomyelination, 300475	MITOCHONDRIAL DISORDERS PANEL MOVEMENT DISORDERS PANEL HEARING IMPAIRMENT PANEL (INCLUDING GJB2) INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS

BCAS3	100%	100%	100%	99.4%	Hengel-Marooofian-Schols syndrome, 619641	INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹
BCAT1	100%	100%	100%	99.4%		METABOLIC DISORDERS PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS
BCAT2	100%	100%	100%	98.6%	Hypervalinemia and hyperleucine-isoleucinemia, 618850	METABOLIC DISORDERS PANEL INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹

BCHE	100%	100%	100%	99.6%	Butyrylcholinesterase deficiency, 617936;{Apnea, postanesthetic, susceptibility to, due to BCHE deficiency}, 617936	MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS
BCKDHA	100%	100%	100%	98.9%	Maple syrup urine disease, type Ia, 248600	MOVEMENT DISORDERS PANEL METABOLIC DISORDERS PANEL INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹

BCKDHB	100%	100%	100%	99.1%	Maple syrup urine disease, type Ib, 620698	MOVEMENT DISORDERS PANEL METABOLIC DISORDERS PANEL INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹
BCKDK	100%	100%	100%	98.7%	Branched-chain keto acid dehydrogenase kinase deficiency, 614923	INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹ METABOLIC DISORDERS PANEL

BCL10	100%	100%	100%	99.5%	{Lymphoma, follicular, somatic}, 605027;?Immunodeficiency 37, 616098;{Sezary syndrome, somatic};{Male germ cell tumor, somatic}, 273300;Lymphoma, MALT, somatic, 137245;{Mesothelioma, somatic}, 156240	PRIMARY IMMUNODEFICIENCIES PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹
BCL11A	100%	100%	100%	98.1%	Dias-Logan syndrome, 617101	INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS
BCL11B	99.9%	98.8%	100%	94.9%	Immunodeficiency 49, severe combined, 617237;Intellectual developmental disorder with dysmorphic facies, speech delay, and T-cell abnormalities, 618092	MOVEMENT DISORDERS PANEL PRIMARY IMMUNODEFICIENCIES PANEL INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS

BCL2	92.5%	92.5%	100%	99.2%	Leukemia/lymphoma, B-cell, 2	MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS
BCL7A	100%	99.8%	100%	97.9%		MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS
BCO1	100%	100%	100%	99.2%	?Hypercarotenemia and vitamin A deficiency, autosomal dominant, 115300	METABOLIC DISORDERS PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS

BCOR	100%	99.4%	98.6%	66.6%	Microphthalmia, syndromic 2, 300166	OROFACIAL CLEFTING PANEL WITH GENOME WIDE CNV ANALYSIS VISION DISORDERS PANEL CRANIOFACIAL ANOMALIES PANEL WITH GENOME WIDE CNV ANALYSIS SKIN DISORDERS PANEL ¹ INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS
BCORL1	100%	99.4%	97.9%	63.4%	Shukla-Vernon syndrome, 301029	MALE INFERTILITY PANEL INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS

BCS1L	100%	100%	100%	99.8%	GRACILE syndrome, 603358;Mitochondrial complex III deficiency, nuclear type 1, 124000;Bjornstad syndrome, 262000	SKIN DISORDERS PANEL ¹ COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹ HEARING IMPAIRMENT PANEL (INCLUDING GJB2) LIVER DISORDERS PANEL INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS RENAL DISORDERS PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS MITOCHONDRIAL DISORDERS PANEL
BDP1	100%	100%	100%	99.4%	?Deafness, autosomal recessive 112, 618257	HEARING IMPAIRMENT PANEL (INCLUDING GJB2) MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS

BEAN1	100%	100%	100%	99.2%	Spinocerebellar ataxia 31, 117210	MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS
BEST1	100%	99.9%	100%	98.2%	Macular dystrophy, vitelliform, 2, 153700;?Microcornea, rod-cone dystrophy, cataract, and posterior staphyloma 2, 193220;Retinitis pigmentosa-50, 613194;Retinitis pigmentosa, concentric, 613194;Vitreoretinchoroidopathy, 193220;Bestrophinopathy, autosomal recessive, 611809	VISION DISORDERS PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS
BET1	100%	100%	100%	99.7%	Muscular dystrophy, congenital, with rapid progression, 254100	MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹
BFSP1	100%	100%	100%	99.2%	Cataract 33, multiple types, 611391	VISION DISORDERS PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹

BFSP2	100%	100%	100%	98.6%	Cataract 12, multiple types, 611597	VISION DISORDERS PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL¹
BGN	100%	99.7%	98%	67%	Meester-Loeys syndrome, 300989;Spondyloepimetaphyseal dysplasia, X-linked, 300106	THORACIC AORTIC ANEURYSM AND/OR DISSECTION PANEL SHORT STATURE/SKELETAL DYSPLASIA PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS

BHLHA9	100%	100%	100%	98.1%	?Camptosynpolydactyly, complex, 607539;Syndactyly, mesoaxial synostotic, with phalangeal reduction, 609432	SHORT STATURE/SKELETAL DYSPLASIA PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹
BHLHE22	100%	99.9%	100%	96.4%		INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS
BICC1	100%	100%	100%	99.3%	{Renal dysplasia, cystic, susceptibility to}, 601331	RENAL DISORDERS PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS

BICD1	100%	100%	100%	99.1%		HEARING IMPAIRMENT PANEL (INCLUDING GJB2) MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS
BICD2	100%	100%	100%	99.5%	Spinal muscular atrophy, lower extremity-predominant, 2B, autosomal dominant, 618291;Spinal muscular atrophy, lower extremity-predominant, 2A, autosomal dominant, 615290	FETAL AKINESIA PANEL HEART DISORDERS PANEL ¹ POLYNEUROPATHIES PANEL ¹ INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS MUSCLE DISORDERS PANEL
BICRA	100%	100%	100%	96.4%	Coffin-Siris syndrome 12, 619325	INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS

BIN1	100%	100%	100%	98.8%	Centronuclear myopathy 2, 255200	FETAL AKINESIA PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL¹ MUSCLE DISORDERS PANEL
BLK	100%	100%	100%	99.2%	Maturity-onset diabetes of the young, type 11, 613375	PRIMARY IMMUNODEFICIENCIES PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS

BLM	96.8%	96.6%	100%	99.5%	Bloom syndrome, 210900	INHERITED BONE MARROW FAILURE AND/OR PREDISPOSITION PANEL TO HEMATOLOGICAL MALIGNANCIES SKIN DISORDERS PANEL ¹ PRIMARY IMMUNODEFICIENCIES PANEL SHORT STATURE/SKELETAL DYSPLASIA PANEL WITH GENOME WIDE CNV ANALYSIS INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹ HEREDITARY CANCER PANEL
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BLNK	94.4%	94.4%	100%	99.7%	?Agammaglobulinemia 4, 613502	PRIMARY IMMUNODEFICIENCIES PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS
BLOC1S1	100%	100%	100%	99.2%		INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS
BLOC1S3	100%	100%	100%	98.7%	Hermansky-Pudlak syndrome 8, 614077	VISION DISORDERS PANEL SKIN DISORDERS PANEL ¹ HEMOSTATIC/THROMB OTIC DISORDERS PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹

BLOC1S5	100%	100%	100%	99.3%	Hermansky-Pudlak syndrome 11, 619172	VISION DISORDERS PANEL HEMOSTATIC/THROMB OTIC DISORDERS PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹
BLOC1S6	100%	100%	100%	99.5%	Hermansky-Pudlak syndrome 9, 614171	VISION DISORDERS PANEL SKIN DISORDERS PANEL ¹ HEMOSTATIC/THROMB OTIC DISORDERS PANEL PRIMARY IMMUNODEFICIENCIES PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹

BLTP1	100%	100%	100%	99.4%	Alkuraya-Kucinskas syndrome, 617822	FETAL AKINESIA PANEL INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL¹
BLVRA	100%	100%	100%	99.6%	Hyperbiliverdinemia, 614156	LIVER DISORDERS PANEL METABOLIC DISORDERS PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL¹

BMP1	100%	100%	100%	99%	Osteogenesis imperfecta, type XIII, 614856	SHORT STATURE/SKELETAL DYSPLASIA PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹
BMP15	100%	99.9%	99%	69.5%	Premature ovarian failure 4, 300510;Ovarian dysgenesis 2, 300510	DISORDERS/DIFFEREN CES OF SEX DEVELOPMENT (DSD) / PRIMARY ADRENAL INSUFFICIENCY PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS PREMATURE OVARIAN INSUFFICIENCY PANEL

BMP2	100%	100%	100%	98.3%	Short stature, facial dysmorphism, and skeletal anomalies with or without cardiac anomalies 1, 617877;Brachydactyly, type A2, 112600;{HFE hemochromatosis, modifier of}, 235200	CRANIOFACIAL ANOMALIES PANEL WITH GENOME WIDE CNV ANALYSIS SHORT STATURE/SKELETAL DYSPLASIA PANEL WITH GENOME WIDE CNV ANALYSIS METABOLIC DISORDERS PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS OROFACIAL CLEFTING PANEL WITH GENOME WIDE CNV ANALYSIS
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BMP4	100%	100%	100%	99.3%	Orofacial cleft 11, 600625;Microphthalmia, syndromic 6, 607932	VISION DISORDERS PANEL CRANIOFACIAL ANOMALIES PANEL WITH GENOME WIDE CNV ANALYSIS HEARING IMPAIRMENT PANEL (INCLUDING GJB2) DISORDERS/DIFFEREN CES OF SEX DEVELOPMENT (DSD) / PRIMARY ADRENAL INSUFFICIENCY PANEL INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS
BMP5	100%	100%	100%	99%		SHORT STATURE/SKELETAL DYSPLASIA PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS

BMP6	100%	99.8%	100%	99.5%	{Iron overload, susceptibility to}, 620121	IRON DISORDERS PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS
BMP7	100%	100%	100%	98.3%		DISORDERS/DIFFEREN CES OF SEX DEVELOPMENT (DSD) / PRIMARY ADRENAL INSUFFICIENCY PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS
BMPER	100%	100%	100%	98.8%	Diaphanospondylodysostosis, 608022	SHORT STATURE/SKELETAL DYSPLASIA PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹ OROFACIAL CLEFTING PANEL WITH GENOME WIDE CNV ANALYSIS

BMPR1A	100%	100%	100%	99.5%	Polyposis syndrome, hereditary mixed, 2, 610069;Polyposis, juvenile intestinal, 174900	PANEL HEREDITARY COLORECTAL AND POLYPOSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS HEREDITARY CANCER PANEL
BMPR1B	100%	100%	100%	99.6%	Acromesomelic dysplasia 3, 609441;Brachydactyly, type A2, 112600;Brachydactyly, type A1, D, 616849	VISION DISORDERS PANEL SHORT STATURE/SKELETAL DYSPLASIA PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹
BMPR2	100%	100%	100%	99.6%	Pulmonary hypertension, familial primary, 1, with or without HHT, 178600;Pulmonary hypertension, primary, fenfluramine or dexfenfluramine-associated, 178600;Pulmonary venoocclusive disease 1, 265450	HEART DISORDERS PANEL ¹ MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS

BMS1	100%	99.8%	100%	99.5%	?Aplasia cutis congenita, nonsyndromic, 107600	SKIN DISORDERS PANEL ¹ MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS
BNC1	99.8%	99.3%	100%	99.5%	?Premature ovarian failure 16, 618723	DISORDERS/DIFFEREN CES OF SEX DEVELOPMENT (DSD) / PRIMARY ADRENAL INSUFFICIENCY PANEL MALE INFERTILITY PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS PREMATURE OVARIAN INSUFFICIENCY PANEL
BNC2	100%	100%	100%	99.3%	Lower urinary tract obstruction, congenital, 618612	MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS
BOLA1	100%	100%	100%	99.7%		MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS MITOCHONDRIAL DISORDERS PANEL

BOLA2	100%	100%	100%	100%		MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS MITOCHONDRIAL DISORDERS PANEL
BOLA3	100%	100%	100%	98.9%	Multiple mitochondrial dysfunctions syndrome 2 with hyperglycinemia, 614299	EPILEPSY PANEL INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS MITOCHONDRIAL DISORDERS PANEL COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹
BORCS8	82.8%	82.8%	100%	98.4%	Neurodegeneration, infantile-onset, with optic atrophy and brain abnormalities, 620987	INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹

BPGM	100%	100%	100%	99.4%	Erythrocytosis, familial, 8, 222800	METABOLIC DISORDERS PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹
BPNT2	100%	100%	100%	99.6%	Chondrodysplasia with joint dislocations, GPAPP type, 614078	CRANIOFACIAL ANOMALIES PANEL WITH GENOME WIDE CNV ANALYSIS SHORT STATURE/SKELETAL DYSPLASIA PANEL WITH GENOME WIDE CNV ANALYSIS METABOLIC DISORDERS PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹ OROFACIAL CLEFTING PANEL WITH GENOME WIDE CNV ANALYSIS

BPTF	100%	99.9%	100%	98.9%	Neurodevelopmental disorder with dysmorphic facies and distal limb anomalies, 617755;{Kaposi sarcoma, susceptibility to}, 148000	INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS
BPY2	50%	48.6%	49.7%	27.8%		MALE INFERTILITY PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS
BPY2B	49.9%	48.1%	49.7%	26.4%		MALE INFERTILITY PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS
BPY2C	49.7%	48%	49.5%	26.3%		MALE INFERTILITY PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS

BRAF	100%	100%	99.9%	97.1%	Melanoma, malignant, somatic, 155600;LEOPARD syndrome 3, 613707;Cardiofaciocutaneous syndrome, 115150;Adenocarcinoma of lung, somatic, 211980;Noonan syndrome 7, 613706;Colorectal cancer, somatic, 114500;Nonsmall cell lung cancer, somatic, 211980	MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS NOONAN SYNDROME / RASOPATHY PANEL HEREDITARY CANCER PANEL PANEL LYNCH SYNDROME (MLH1, PMS2, MSH2, MSH6) INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS LYMPHATIC ANOMALIES PANEL WITH GENOME WIDE CNV ANALYSIS HEMOSTATIC/THROMBOTIC DISORDERS PANEL SHORT STATURE/SKELETAL DYSPLASIA PANEL WITH GENOME WIDE CNV ANALYSIS SKIN DISORDERS PANEL ¹ HEART DISORDERS PANEL ¹ CONGENITAL HEARTDISEASE PANEL ¹ INHERITED BONE MARROW FAILURE
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						AND/OR PREDISPOSITION PANEL TO HEMATOLOGICAL MALIGNANCIES
BRAT1	100%	100%	100%	98.9%	Neurodevelopmental disorder with cerebellar atrophy and with or without seizures, 618056;Rigidity and multifocal seizure syndrome, lethal neonatal, 614498	MOVEMENT DISORDERS PANEL EPILEPSY PANEL INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL¹

BRCA1	100%	100%	100%	99.3%	Fanconi anemia, complementation group S, 617883;{Breast-ovarian cancer, familial, 1}, 604370;{Pancreatic cancer, susceptibility to, 4}, 614320	INHERITED BONE MARROW FAILURE AND/OR PREDISPOSITION PANEL TO HEMATOLOGICAL MALIGNANCIES MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS PANEL HEREDITARY PANCREATIC CANCER (BRCA1, BRCA2, PALB2, ATM, CDKN2A, MLH1, MSH2, MSH6, PMS2, STK11) COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹ HEREDITARY CANCER PANEL
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BRCA2	100%	100%	100%	99.6%	Fanconi anemia, complementation group D1, 605724;{Glioblastoma 3}, 613029;{Medulloblastoma}, 155255;{Prostate cancer}, 176807;{Breast-ovarian cancer, familial, 2}, 612555;{Breast cancer, male, susceptibility to}, 114480;{Pancreatic cancer 2}, 613347;Wilms tumor, 194070	INHERITED BONE MARROW FAILURE AND/OR PREDISPOSITION PANEL TO HEMATOLOGICAL MALIGNANCIES MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS PANEL HEREDITARY PANCREATIC CANCER (BRCA1, BRCA2, PALB2, ATM, CDKN2A, MLH1, MSH2, MSH6, PMS2, STK11) COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹ SONIC HEDGEHOG MEDULLOBLASTOMA PANEL HEREDITARY CANCER PANEL
BRD4	100%	100%	100%	96.1%	Cornelia de Lange syndrome 6, 620568	INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS

BRDT	100%	100%	100%	99.5%	?Spermatogenic failure 21, 617644	MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS
BRF1	100%	99.9%	100%	99%	Cerebellofaciodental syndrome, 616202	SHORT STATURE/SKELETAL DYSPLASIA PANEL WITH GENOME WIDE CNV ANALYSIS INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL¹

BRIP1	96%	96%	100%	99.5%	Fanconi anemia, complementation group J, 609054;{Breast cancer, early-onset, susceptibility to}, 114480	INHERITED BONE MARROW FAILURE AND/OR PREDISPOSITION PANEL TO HEMATOLOGICAL MALIGNANCIES SKIN DISORDERS PANEL ¹ MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹ HEREDITARY CANCER PANEL
BRPF1	100%	99.9%	100%	99.1%	Intellectual developmental disorder with dysmorphic facies and ptosis, 617333	INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS

BRSK2	100%	99.9%	100%	97.6%		INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS
BRWD1	100%	100%	100%	99.3%	Ciliary dyskinesia, primary, 51, 620438	MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS
BRWD3	100%	100%	98.7%	68%	Intellectual developmental disorder, X-linked 93, 300659	TALL STATURE PANEL WITH GENOME WIDE CNV ANALYSIS INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS

BSCL2	100%	100%	100%	99.2%	Lipodystrophy, congenital generalized, type 2, 269700;Neuronopathy, distal hereditary motor, autosomal dominant 13, 619112;Silver spastic paraplegia syndrome, 270685;Encephalopathy, progressive, with or without lipodystrophy, 615924	MOVEMENT DISORDERS PANEL SKIN DISORDERS PANEL ¹ HEART DISORDERS PANEL ¹ POLYNEUROPATHIES PANEL ¹ METABOLIC DISORDERS PANEL INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹
BSN	99.7%	99.5%	100%	98.8%		INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS

BSND	100%	99.8%	100%	99.3%	Sensorineural deafness with mild renal dysfunction, 602522;Bartter syndrome, type 4a, 602522	MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹ HEARING IMPAIRMENT PANEL (INCLUDING GJB2) RENAL DISORDERS PANEL
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BTD	94.2%	94.2%	100%	99.4%	Biotinidase deficiency, 253260	EPILEPSY PANEL METABOLIC DISORDERS PANEL INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS MITOCHONDRIAL DISORDERS PANEL MOVEMENT DISORDERS PANEL VISION DISORDERS PANEL SKIN DISORDERS PANEL ¹ HEARING IMPAIRMENT PANEL (INCLUDING GJB2) COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹
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BTG4	100%	100%	100%	99.5%	Oocyte/zygote/embryo maturation arrest 8, 619009	UNSUCCESSFUL IVF TREATMENTS, RECURRENT MOLAR PREGNANCIES AND/OR RECURRENT PREGNANCY LOSS PANEL ¹ MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS
BTK	100%	99.5%	98.6%	69.6%	Agammaglobulinemia, X-linked 1, 300755;Isolated growth hormone deficiency, type III, with agammaglobulinemia, 307200	PRIMARY IMMUNODEFICIENCIES PANEL SHORT STATURE/SKELETAL DYSPLASIA PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS
BTRC	100%	100%	100%	99.1%		SHORT STATURE/SKELETAL DYSPLASIA PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS

BUB1	100%	100%	100%	99.6%	Colorectal cancer with chromosomal instability, somatic, 114500;Microcephaly 30, primary, autosomal recessive, 620183	MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹ HEREDITARY CANCER PANEL INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS
BUB1B	100%	100%	100%	99.4%	Colorectal cancer, somatic, 114500;[Premature chromatid separation trait], 176430;Mosaic variegated aneuploidy syndrome 1, 257300	SHORT STATURE/SKELETAL DYSPLASIA PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹ HEREDITARY CANCER PANEL INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS

BUB3	100%	100%	100%	99.5%		MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS HEREDITARY CANCER PANEL
BVES	100%	100%	100%	99.8%		HEART DISORDERS PANEL ¹ MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹ MUSCLE DISORDERS PANEL
C11orf80	92.2%	92.2%	100%	99.2%		UNSUCCESSFUL IVF TREATMENTS, RECURRENT MOLAR PREGNANCIES AND/OR RECURRENT PREGNANCY LOSS PANEL ¹ MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS

C12orf4	100%	100%	100%	99.1%		INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹
C12orf57	100%	100%	100%	99.1%	Temtamy syndrome, 218340	INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹

C14orf39	100%	100%	100%	99.5%	Spermatogenic failure 52, 619202;?Premature ovarian failure 18, 619203	DISORDERS/DIFFERENCES OF SEX DEVELOPMENT (DSD) / PRIMARY ADRENAL INSUFFICIENCY PANEL MALE INFERTILITY PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS PREMATURE OVARIAN INSUFFICIENCY PANEL
C18orf32	100%	100%	100%	99.2%	?Glycosylphosphatidylinositol biosynthesis defect 25, 619985	MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS

C19orf12	100%	100%	100%	97.5%	Neurodegeneration with brain iron accumulation 4, 614298;?Spastic paraplegia 43, autosomal recessive, 615043	MOVEMENT DISORDERS PANEL VISION DISORDERS PANEL POLYNEUROPATHIES PANEL ¹ MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS MITOCHONDRIAL DISORDERS PANEL PARKINSON DISEASE PANEL COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹
C19orf44	100%	100%	100%	99.2%		VISION DISORDERS PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS
C1GALT1C1	100%	99.9%	98.8%	67%	Hemolytic uremic syndrome, atypical, 8, with rhizomelic short stature, 301110;Tn polyagglutination syndrome, somatic, 300622	METABOLIC DISORDERS PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS

C1QA	76.7%	74.3%	100%	99.7%	C1q deficiency 1, 613652	SKIN DISORDERS PANEL ¹ PRIMARY IMMUNODEFICIENCIES PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹
C1QB	77.2%	76.9%	100%	99.1%	C1q deficiency 2, 620321	SKIN DISORDERS PANEL ¹ PRIMARY IMMUNODEFICIENCIES PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹

C1QBP	100%	100%	100%	99.1%	Combined oxidative phosphorylation deficiency 33, 617713	MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS MITOCHONDRIAL DISORDERS PANEL COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹
C1QC	99.8%	97.7%	100%	97.9%	C1q deficiency 3, 620322	SKIN DISORDERS PANEL ¹ PRIMARY IMMUNODEFICIENCIES PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹
C1QTNF5	100%	100%	100%	98.8%	Retinal degeneration, late-onset, autosomal dominant, 605670	VISION DISORDERS PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS

C1R	100%	100%	100%	99.1%	Ehlers-Danlos syndrome, periodontal type, 1, 130080	PRIMARY IMMUNODEFICIENCIES PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS
C1S	100%	100%	100%	99.3%	C1s deficiency, 613783;Ehlers-Danlos syndrome, periodontal type, 2, 617174	PRIMARY IMMUNODEFICIENCIES PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹
C2	100%	100%	100%	99.3%	C2 deficiency, 217000;{Macular degeneration, age-related, 14, reduced risk of}, 615489	PRIMARY IMMUNODEFICIENCIES PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS

C2CD3	96%	96%	100%	98.8%	Orofaciodigital syndrome XIV, 615948	CILIOPATHIES PANEL SKIN DISORDERS PANEL ¹ INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹ OROFACIAL CLEFTING PANEL WITH GENOME WIDE CNV ANALYSIS
C2CD6	100%	100%	100%	99.6%	?Spermatogenic failure 68, 619805	MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS

C2orf69	100%	100%	100%	99.2%	Combined oxidative phosphorylation deficiency 53, 619423	PRIMARY IMMUNODEFICIENCIES PANEL METABOLIC DISORDERS PANEL INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS MITOCHONDRIAL DISORDERS PANEL COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹
C3	97.5%	97.5%	100%	97.5%	C3 deficiency, 613779;{Hemolytic uremic syndrome, atypical, susceptibility to, 5}, 612925;{Macular degeneration, age-related, 9}, 611378	HEMOSTATIC/THROMB OTIC DISORDERS PANEL COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹ MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS RENAL DISORDERS PANEL PRIMARY IMMUNODEFICIENCIES PANEL

C3orf52	100%	100%	100%	99.8%	Hypotrichosis 15, 620177	MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS
C4A	99.8%	99.3%	99.7%	96.1%	[Blood group, Rodgers], 614374;C4a deficiency, 614380	MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹
C4B	99.9%	99.6%	99.4%	96%	C4B deficiency, 614379	MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹
C5	100%	100%	100%	99.7%	C5 deficiency, 609536;[Eculizumab, poor response to], 615749	PRIMARY IMMUNODEFICIENCIES PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹

C6	100%	100%	100%	99.6%	C6 deficiency, 612446	PRIMARY IMMUNODEFICIENCIES PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹
C7	98.3%	97.2%	100%	99.5%	C7 deficiency, 610102	PRIMARY IMMUNODEFICIENCIES PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS
C8A	100%	100%	100%	99.6%	C8 deficiency, type I, 613790	PRIMARY IMMUNODEFICIENCIES PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹

C8B	100%	100%	100%	99.4%	C8 deficiency, type II, 613789	PRIMARY IMMUNODEFICIENCIES PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹
C8G	100%	100%	100%	98.6%		PRIMARY IMMUNODEFICIENCIES PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS
C9	99.3%	99.3%	100%	99.5%	C9 deficiency, 613825;{Macular degeneration, age-related, 15, susceptibility to}, 615591	PRIMARY IMMUNODEFICIENCIES PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹

C9orf72	100%	100%	100%	99.2%	Frontotemporal dementia and/or amyotrophic lateral sclerosis 1, 105550	MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS
CA12	100%	100%	100%	99.5%	Hyperchlorhidrosis, isolated, 143860	MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹

CA2	100%	100%	100%	99.6%	Osteopetrosis, autosomal recessive 3, with renal tubular acidosis, 259730	INHERITED BONE MARROW FAILURE AND/OR PREDISPOSITION PANEL TO HEMATOLOGICAL MALIGNANCIES SKIN DISORDERS PANEL ¹ PRIMARY IMMUNODEFICIENCIES PANEL SHORT STATURE/SKELETAL DYSPLASIA PANEL WITH GENOME WIDE CNV ANALYSIS INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS RENAL DISORDERS PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹
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CA4	100%	100%	100%	98.8%		MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS
CA5A	100%	100%	100%	99%	Hyperammonemia due to carbonic anhydrase VA deficiency, 615751	METABOLIC DISORDERS PANEL INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS MITOCHONDRIAL DISORDERS PANEL COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹
CA8	100%	100%	100%	99.7%	Spinocerebellar ataxia, autosomal recessive 34, 613227	MOVEMENT DISORDERS PANEL INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹

CABIN1	100%	99.9%	100%	99%		NEUROLOGICAL PAIN DISORDERS PANEL¹ MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS
CABP2	100%	100%	100%	95.4%	Deafness, autosomal recessive 93, 614899	HEARING IMPAIRMENT PANEL (INCLUDING GJB2) MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL¹
CABP4	100%	100%	100%	98.7%	Cone-rod synaptic disorder, congenital nonprogressive, 610427	VISION DISORDERS PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL¹

CACHD1	100%	100%	100%	99.1%		INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS
CACNA1A	100%	100%	100%	97.6%	Spinocerebellar ataxia 6, 183086;Episodic ataxia, type 2, 108500;Developmental and epileptic encephalopathy 42, 617106;Migraine, familial hemiplegic, 1, with progressive cerebellar ataxia, 141500;Migraine, familial hemiplegic, 1, 141500	MOVEMENT DISORDERS PANEL EPILEPSY PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS NEUROLOGICAL PAIN DISORDERS PANEL¹
CACNA1B	100%	99.8%	100%	98.4%	Neurodevelopmental disorder with seizures and nonepileptic hyperkinetic movements, 618497	INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL¹

CACNA1C	100%	100%	100%	99.2%	Timothy syndrome, 601005;Long QT syndrome 8, 618447;Neurodevelopmental disorder with hypotonia, language delay, and skeletal defects with or without seizures, 620029;Brugada syndrome 3, 611875	CONGENITAL HEARTDISEASE PANEL¹ ARRHYTHMIA AND CARDIAC CONDUCTION DISORDERS PANEL¹ HEART DISORDERS PANEL¹ HYPERTROPHIC CARDIOMYOPATHY PANEL¹ LONG QT SYNDROME PANEL¹ INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS
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CACNA1D	100%	100%	100%	99.1%	Primary aldosteronism, seizures, and neurologic abnormalities, 615474;Sinoatrial node dysfunction and deafness, 614896	HEARING IMPAIRMENT PANEL (INCLUDING GJB2) HEART DISORDERS PANEL ¹ INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹
CACNA1E	100%	100%	100%	99.2%	Developmental and epileptic encephalopathy 69, 618285	MOVEMENT DISORDERS PANEL EPILEPSY PANEL INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS

CACNA1F	100%	99.9%	98.1%	64.2%	Cone-rod dystrophy, X-linked, 3, 300476;Night blindness, congenital stationary (incomplete), 2A, X-linked, 300071;Aland Island eye disease, 300600	VISION DISORDERS PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS
CACNA1G	100%	100%	100%	98.5%	Spinocerebellar ataxia 42, 616795;Spinocerebellar ataxia 42, early-onset, severe, with neurodevelopmental deficits, 618087	MOVEMENT DISORDERS PANEL INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS
CACNA1H	99.5%	98.9%	100%	97.5%	{Epilepsy, childhood absence, susceptibility to, 6}, 611942;Hyperaldosteronism, familial, type IV, 617027;{Epilepsy, idiopathic generalized, susceptibility to, 6}, 611942	NEUROLOGICAL PAIN DISORDERS PANEL' RENAL DISORDERS PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS

CACNA1I	100%	100%	100%	97.6%	Neurodevelopmental disorder with speech impairment and with or without seizures, 620114	INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS
CACNA1S	100%	100%	100%	99.1%	{Thyrotoxic periodic paralysis, susceptibility to, 1}, 188580;Congenital myopathy 18 due to dihydropyridine receptor defect, 620246;Hypokalemic periodic paralysis, type 1, 170400;{Malignant hyperthermia susceptibility 5}, 601887	MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹ MUSCLE DISORDERS PANEL FETAL AKINESIA PANEL

CACNA2D1	100%	100%	100%	98.5%	Developmental and epileptic encephalopathy 110, 620149	HEART DISORDERS PANEL ¹ INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹ EPILEPSY PANEL
CACNA2D2	100%	100%	100%	98.5%	Cerebellar atrophy with seizures and variable developmental delay, 618501	COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹ EPILEPSY PANEL INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS
CACNA2D4	100%	100%	100%	98.9%	Retinal cone dystrophy 4, 610478	VISION DISORDERS PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS

CACNB2	100%	100%	100%	99.3%	Brugada syndrome 4, 611876	HEART DISORDERS PANEL ¹ MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS
CACNB4	100%	100%	100%	99.3%	{Epilepsy, juvenile myoclonic, susceptibility to, 6}, 607682;?Episodic ataxia, type 5, 613855;{Epilepsy, idiopathic generalized, susceptibility to, 9}, 607682	MOVEMENT DISORDERS PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS EPILEPSY PANEL
CACNG2	100%	100%	100%	97.5%	?Intellectual developmental disorder, autosomal dominant 10, 614256	MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS

CAD	100%	100%	100%	98.8%	Developmental and epileptic encephalopathy 50, 616457	COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹ METABOLIC DISORDERS PANEL INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS INHERITED BONE MARROW FAILURE AND/OR PREDISPOSITION PANEL TO HEMATOLOGICAL MALIGNANCIES EPILEPSY PANEL
CADM3	100%	100%	100%	98.9%	Charcot-Marie-Tooth disease, axonal, type 2FF, 619519	POLYNEUROPATHIES PANEL ¹ MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS
CALCRL	100%	100%	100%	99.8%	?Lymphatic malformation 8, 618773	MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS

CALM1	100%	100%	100%	99.2%	Ventricular tachycardia, catecholaminergic polymorphic, 4, 614916;Long QT syndrome 14, 616247	HEART DISORDERS PANEL ¹ LONG QT SYNDROME PANEL ¹ MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS ARRHYTHMIA AND CARDIAC CONDUCTION DISORDERS PANEL ¹
CALM2	73.5%	73.5%	100%	99.3%	Long QT syndrome 15, 616249	HEART DISORDERS PANEL ¹ LONG QT SYNDROME PANEL ¹ MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS ARRHYTHMIA AND CARDIAC CONDUCTION DISORDERS PANEL ¹

CALM3	100%	100%	100%	99.6%	Long QT syndrome 16, 618782;?Ventricular tachycardia, catecholaminergic polymorphic 6, 618782	HEART DISORDERS PANEL ¹ LONG QT SYNDROME PANEL ¹ MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS ARRHYTHMIA AND CARDIAC CONDUCTION DISORDERS PANEL ¹
CALR	100%	100%	100%	98.9%	Myelofibrosis, somatic, 254450;Thrombocythemia, somatic, 187950	INHERITED BONE MARROW FAILURE AND/OR PREDISPOSITION PANEL TO HEMATOLOGICAL MALIGNANCIES HEMOSTATIC/THROMB OTIC DISORDERS PANEL IRON DISORDERS PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS

CALR3	100%	100%	100%	99.3%		MALE INFERTILITY PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS
CAMK2A	100%	100%	100%	98.5%	Intellectual developmental disorder, autosomal dominant 53, 617798;?Intellectual developmental disorder, autosomal recessive 63, 618095	INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹
CAMK2B	100%	99.8%	100%	98.3%	Intellectual developmental disorder, autosomal dominant 54, 617799	INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS

CAMK2D	100%	100%	100%	99.6%		INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS
CAMK2G	100%	100%	100%	99.3%	Intellectual developmental disorder, autosomal dominant 59, 618522	INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS
CAMK4	100%	100%	100%	99.8%		INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS
CAMLG	100%	100%	100%	99.3%	?Congenital disorder of glycosylation, type IIz, 620201	MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS

CAMSAP1	100%	100%	100%	99%	Cortical dysplasia, complex, with other brain malformations 12, 620316	INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹
CAMTA1	100%	99.9%	100%	99%	Cerebellar dysfunction with variable cognitive and behavioral abnormalities, 614756	MOVEMENT DISORDERS PANEL INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS

CANT1	100%	99.9%	100%	98.1%	Desbuquois dysplasia 1, 251450;Epiphyseal dysplasia, multiple, 7, 617719	SHORT STATURE/SKELETAL DYSPLASIA PANEL WITH GENOME WIDE CNV ANALYSIS METABOLIC DISORDERS PANEL INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL¹
CAP2	100%	100%	100%	99.1%	Cardiomyopathy, dilated, 2l, 620462	MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL¹

CAPN1	100%	100%	100%	97.8%	Spastic paraplegia 76, autosomal recessive, 616907	MOVEMENT DISORDERS PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹
CAPN10	100%	100%	100%	99%	{Diabetes mellitus, noninsulin-dependent 1}, 601283	MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹
CAPN12	100%	100%	100%	95.9%		SKIN DISORDERS PANEL ¹ MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS

CAPN15	100%	100%	100%	97.6%	Oculogastrointestinal neurodevelopmental syndrome, 619318	INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹
CAPN3	100%	100%	100%	99.2%	Muscular dystrophy, limb-girdle, autosomal recessive 1, 253600;Muscular dystrophy, limb-girdle, autosomal dominant 4, 618129	MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹ MUSCLE DISORDERS PANEL
CAPN5	100%	100%	100%	99.2%	Vitreoretinopathy, neovascular inflammatory, 193235	VISION DISORDERS PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS
CAPNS1	100%	100%	100%	98.5%	Pulmonary hypertension, primary, 6, 620777	MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS

CAPRIN1	100%	100%	100%	98.9%	Neurodevelopmental disorder with language impairment, autism, and attention deficit-hyperactivity disorder, 620782;Neurodegeneration, childhood-onset, with cerebellar ataxia and cognitive decline, 620636	INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS
CAPZA2	100%	100%	100%	99.1%		INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS
CARD10	100%	100%	100%	97.4%	?Immunodeficiency 89 and autoimmunity, 619632	PRIMARY IMMUNODEFICIENCIES PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS

CARD11	100%	100%	100%	98.8%	B-cell expansion with NFkB and T-cell anergy, 616452;Immunodeficiency 11B with atopic dermatitis, 617638;Immunodeficiency 11A, 615206	SKIN DISORDERS PANEL ¹ PRIMARY IMMUNODEFICIENCIES PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹ HEREDITARY CANCER PANEL
CARD14	100%	100%	100%	99%	Psoriasis 2, 602723;Pityriasis rubra pilaris, 173200	SKIN DISORDERS PANEL ¹ PRIMARY IMMUNODEFICIENCIES PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS
CARD8	100%	100%	100%	99.4%	?Inflammatory bowel disease (Crohn disease) 30, 619079	MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS

CARD9	100%	100%	100%	98.9%	Immunodeficiency 103, susceptibility to fungal infection, 212050	SKIN DISORDERS PANEL ¹ PRIMARY IMMUNODEFICIENCIES PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹
CARMIL2	100%	99.4%	100%	98.4%	Immunodeficiency 58, 618131	SKIN DISORDERS PANEL ¹ PRIMARY IMMUNODEFICIENCIES PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹

CARS1	100%	99.9%	100%	99.4%	Microcephaly, developmental delay, and brittle hair syndrome, 618891	SKIN DISORDERS PANEL ¹ LIVER DISORDERS PANEL INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹
CARS2	100%	100%	100%	99.2%	Combined oxidative phosphorylation deficiency 27, 616672	INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹ MITOCHONDRIAL DISORDERS PANEL

CASK	100%	99.9%	99.1%	70.3%	Intellectual developmental disorder, with or without nystagmus, 300422;Intellectual developmental disorder and microcephaly with pontine and cerebellar hypoplasia, 300749;FG syndrome 4, 300422	EPILEPSY PANEL INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS
CASP10	100%	99.7%	100%	99.5%	Autoimmune lymphoproliferative syndrome, type II, 603909;Gastric cancer, somatic, 613659;Lymphoma, non-Hodgkin, somatic, 605027	INHERITED BONE MARROW FAILURE AND/OR PREDISPOSITION PANEL TO HEMATOLOGICAL MALIGNANCIES PRIMARY IMMUNODEFICIENCIES PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS
CASP14	100%	100%	100%	98.3%	Ichthyosis, congenital, autosomal recessive 12, 617320	SKIN DISORDERS PANEL ¹ MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹

CASP2	100%	100%	100%	99.2%	Intellectual developmental disorder, autosomal recessive 80, with variant lissencephaly, 620653	INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS
CASP8	97%	97%	100%	99.5%	{Breast cancer, protection against}, 114480;?Caspase 8 lymphadenopathy syndrome, 607271;Hepatocellular carcinoma, somatic, 114550;{Lung cancer, protection against}, 211980	PRIMARY IMMUNODEFICIENCIES PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹
CASQ1	100%	100%	100%	98.2%	Myopathy, vacuolar, with CASQ1 aggregates, 616231	MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS MUSCLE DISORDERS PANEL

CASQ2	100%	100%	100%	99.7%	Ventricular tachycardia, catecholaminergic polymorphic, 2, 611938	EPILEPSY PANEL HEART DISORDERS PANEL ¹ MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹ ARRHYTHMIA AND CARDIAC CONDUCTION DISORDERS PANEL ¹
CASR	100%	100%	100%	99.5%	Hypocalcemia, autosomal dominant, with Bartter syndrome, 601198;Hyperparathyroidism, neonatal, 239200;Hypocalcemia, autosomal dominant, 601198;Hypocalciuric hypercalcemia, type I, 145980;{?Epilepsy idiopathic generalized, susceptibility to, 8}, 612899	SHORT STATURE/SKELETAL DYSPLASIA PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹ RENAL DISORDERS PANEL

CAST	100%	100%	100%	99.4%	Peeling skin with leukonychia, acral punctate keratoses, cheilitis, and knuckle pads, 616295	SKIN DISORDERS PANEL ¹ MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹
CASZ1	99.1%	96.7%	100%	98.5%		HEART DISORDERS PANEL ¹ MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS
CAT	100%	100%	100%	99.5%	Acatlasemia, 614097	METABOLIC DISORDERS PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹
CATIP	100%	100%	100%	98.4%	?Spermatogenic failure 54, 619379	MALE INFERTILITY PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS

CATSPER1	100%	100%	100%	97.8%	Spermatogenic failure 7, 612997	MALE INFERTILITY PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹
CATSPER2	100%	100%	100%	96.9%		MALE INFERTILITY PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS
CAV1	74.6%	74.6%	100%	98.7%	Lipodystrophy, congenital generalized, type 3, 612526;Pulmonary hypertension, primary, 3, 615343;Lipodystrophy, familial partial, type 7, 606721	SKIN DISORDERS PANEL ¹ HEART DISORDERS PANEL ¹ METABOLIC DISORDERS PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹

CAV3	100%	100%	100%	99%	Myopathy, distal, Tateyama type, 614321;Creatine phosphokinase, elevated serum, 123320;Cardiomyopathy, familial hypertrophic, 192600;Rippling muscle disease 2, 606072;Long QT syndrome 9, 611818	HEART DISORDERS PANEL ¹ MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS MUSCLE DISORDERS PANEL
CAVIN1	100%	100%	100%	97.9%	Lipodystrophy, congenital generalized, type 4, 613327	SKIN DISORDERS PANEL ¹ PRIMARY IMMUNODEFICIENCIES PANEL METABOLIC DISORDERS PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹ MUSCLE DISORDERS PANEL
CAVIN2	100%	100%	100%	99.2%		SKIN DISORDERS PANEL ¹ MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS

CBFB	100%	100%	100%	99.1%	Cleidocranial dysplasia 2, 620099	SHORT STATURE/SKELETAL DYSPLASIA PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS
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CBL	100%	100%	100%	98.9%	Noonan syndrome-like disorder with or without juvenile myelomonocytic leukemia, 613563;?Juvenile myelomonocytic leukemia, 607785	INHERITED BONE MARROW FAILURE AND/OR PREDISPOSITION PANEL TO HEMATOLOGICAL MALIGNANCIES NOONAN SYNDROME / RASOPATHY PANEL HEREDITARY CANCER PANEL LYMPHATIC ANOMALIES PANEL WITH GENOME WIDE CNV ANALYSIS INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS SKIN DISORDERS PANEL ¹ HEMOSTATIC/THROMBOTIC DISORDERS PANEL SHORT STATURE/SKELETAL DYSPLASIA PANEL WITH GENOME WIDE CNV ANALYSIS
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CBLB	100%	100%	100%	99.4%	Autoimmune disease, multisystem, infantile-onset, 3, 620430	PRIMARY IMMUNODEFICIENCIES PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹
CBLIF	100%	100%	100%	99.1%	Intrinsic factor deficiency, 261000	METABOLIC DISORDERS PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹

CBS	100%	100%	100%	98.6%	Thrombosis, hyperhomocysteinemic, 236200;Homocystinuria, B6-responsive and nonresponsive types, 236200	THORACIC AORTIC ANEURYSM AND/OR DISSECTION PANEL VISION DISORDERS PANEL SKIN DISORDERS PANEL ¹ TALL STATURE PANEL WITH GENOME WIDE CNV ANALYSIS METABOLIC DISORDERS PANEL INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹
CBX1	100%	100%	100%	99.5%		INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS

CBX2	100%	100%	100%	98.9%	?46XY sex reversal 5, 613080	DISORDERS/DIFFERENCES OF SEX DEVELOPMENT (DSD) / PRIMARY ADRENAL INSUFFICIENCY PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS
CBY1	100%	100%	100%	98.8%		CILIOPATHIES PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS
CC2D1A	100%	100%	100%	98%	Intellectual developmental disorder, autosomal recessive 3, 608443	INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹

CC2D2A	98.2%	98.2%	100%	99.3%	COACH syndrome 2, 619111;Retinitis pigmentosa 93, 619845;Meckel syndrome 6, 612284;Joubert syndrome 9, 612285	MOVEMENT DISORDERS PANEL VISION DISORDERS PANEL CILIOPATHIES PANEL SHORT STATURE/SKELETAL DYSPLASIA PANEL WITH GENOME WIDE CNV ANALYSIS LIVER DISORDERS PANEL INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS RENAL DISORDERS PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹ OROFACIAL CLEFTING PANEL WITH GENOME WIDE CNV ANALYSIS
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CCBE1	100%	100%	100%	99.3%	Hennekam lymphangiectasia-lymphedema syndrome 1, 235510	CRANIOFACIAL ANOMALIES PANEL WITH GENOME WIDE CNV ANALYSIS SKIN DISORDERS PANEL ¹ PRIMARY IMMUNODEFICIENCIES PANEL LYMPHATIC ANOMALIES PANEL WITH GENOME WIDE CNV ANALYSIS INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹
CCDC103	100%	100%	100%	99.2%		CILIOPATHIES PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹

CCDC115	100%	100%	100%	96.8%	Congenital disorder of glycosylation, type Ilo, 616828	METABOLIC DISORDERS PANEL INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL¹
CCDC134	100%	100%	100%	98.9%	Osteogenesis imperfecta, type XXII, 619795	SHORT STATURE/SKELETAL DYSPLASIA PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS

CCDC141	100%	100%	100%	99.6%		DISORDERS/DIFFERENCES OF SEX DEVELOPMENT (DSD) / PRIMARY ADRENAL INSUFFICIENCY PANEL HYPOGONADOTROPIC HYPOGONADISM (KALLMANN) PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS
CCDC146	100%	100%	100%	99.3%	Spermatogenic failure 94, 620850	MALE INFERTILITY PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS
CCDC174	100%	100%	100%	99.3%	Hypotonia, infantile, with psychomotor retardation, 616816	INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹

CCDC186	100%	100%	100%	99.8%		INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS
CCDC22	100%	99.5%	98.3%	65%	Ritscher-Schinzel syndrome 2, 300963	INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS
CCDC28B	100%	100%	100%	98.3%	{Bardet-Biedl syndrome 1, modifier of}, 209900	CILIOPATHIES PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS

CCDC32	100%	100%	100%	98.5%	Cardiofacioneurodevelopmental syndrome, 619123	INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹ OROFACIAL CLEFTING PANEL WITH GENOME WIDE CNV ANALYSIS
CCDC34	100%	100%	100%	99.5%	Spermatogenic failure 76, 620084	MALE INFERTILITY PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS
CCDC39	100%	100%	100%	99.4%	Ciliary dyskinesia, primary, 14, 613807	CILIOPATHIES PANEL MALE INFERTILITY PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹

CCDC40	100%	100%	100%	98.4%	Ciliary dyskinesia, primary, 15, 613808	CILIOPATHIES PANEL MALE INFERTILITY PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹
CCDC47	100%	100%	100%	98.9%	Trichohepatoneurodevelopmental syndrome, 618268	INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹
CCDC50	100%	100%	100%	99.3%	?Deafness, autosomal dominant 44, 607453	HEARING IMPAIRMENT PANEL (INCLUDING GJB2) MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS

CCDC62	100%	100%	100%	99.4%	?Spermatogenic failure 67, 619803	MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS
CCDC65	100%	100%	100%	99.4%	Ciliary dyskinesia, primary, 27, 615504	CILIOPATHIES PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹
CCDC78	100%	100%	100%	99%	?Centronuclear myopathy 4, 614807	MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS MUSCLE DISORDERS PANEL
CCDC8	99.6%	97.1%	100%	98.3%	3-M syndrome 3, 614205	SHORT STATURE/SKELETAL DYSPLASIA PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹

CCDC88A	97.4%	97.4%	100%	98.8%	PEHO syndrome-like, 617507	INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹
CCDC88C	100%	100%	100%	98.7%	?Spinocerebellar ataxia 40, 616053;Hydrocephalus, congenital, 1, 236600	INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹
CCIN	100%	100%	100%	99.6%	Spermatogenic failure 91, 620838	MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS

CCM2	100%	99.9%	100%	98.8%	Cerebral cavernous malformations-2, 603284	EPILEPSY PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS
CCN2	100%	100%	100%	99.4%		SHORT STATURE/SKELETAL DYSPLASIA PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS
CCN6	100%	100%	100%	99.1%	Progressive pseudorheumatoid dysplasia, 208230	SHORT STATURE/SKELETAL DYSPLASIA PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹

CCNB3	100%	100%	99.2%	71.5%		UNSUCCESSFUL IVF TREATMENTS, RECURRENT MOLAR PREGNANCIES AND/OR RECURRENT PREGNANCY LOSS PANEL ¹ MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS
CCND2	100%	100%	100%	98.7%	Megalencephaly-polymicrogyria-polydactyly-hydrocephalus syndrome 3, 615938	INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS
CCNF	100%	100%	100%	98.5%	Frontotemporal dementia and/or amyotrophic lateral sclerosis 5, 619141	MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS

CCNK	99.9%	97.6%	95.8%	85.6%	?Intellectual developmental disorder with hypertelorism and distinctive facies, 618147	INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS
CCNO	100%	100%	100%	99.3%	Ciliary dyskinesia, primary, 29, 615872	CILIOPATHIES PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹
CCNQ	100%	99.9%	99.3%	67.8%	STAR syndrome, 300707	DISORDERS/DIFFEREN CES OF SEX DEVELOPMENT (DSD) / PRIMARY ADRENAL INSUFFICIENCY PANEL SHORT STATURE/SKELETAL DYSPLASIA PANEL WITH GENOME WIDE CNV ANALYSIS RENAL DISORDERS PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS

CCR2	100%	100%	100%	99.3%	{HIV infection, susceptibility/resistance to}, 609423;Polycystic lung disease, 219600	PRIMARY IMMUNODEFICIENCIES PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹
CCT2	100%	100%	100%	99.9%		VISION DISORDERS PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS
CCT3	100%	100%	100%	99.4%		INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS

CCT5	100%	100%	100%	99.4%	?Neuropathy, hereditary sensory, with spastic paraplegia, 256840	MOVEMENT DISORDERS PANEL POLYNEUROPATHIES PANEL ¹ MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹
CD151	100%	100%	100%	99.1%	[Blood group, Raph], 179620;Epidermolysis bullosa simplex 7, with nephropathy and deafness, 609057	SKIN DISORDERS PANEL ¹ HEARING IMPAIRMENT PANEL (INCLUDING GJB2) RENAL DISORDERS PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹
CD164	100%	100%	100%	99.5%	?Deafness, autosomal dominant 66, 616969	HEARING IMPAIRMENT PANEL (INCLUDING GJB2) MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS

CD19	100%	100%	100%	98.5%	Immunodeficiency, common variable, 3, 613493	PRIMARY IMMUNODEFICIENCIES PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL¹
CD247	77.1%	71.6%	100%	99.2%	?Immunodeficiency 25, 610163	PRIMARY IMMUNODEFICIENCIES PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL¹ SEVERE COMBINED IMMUNODEFICIENCY (SCID) PANEL

CD27	100%	100%	100%	98.6%	Lymphoproliferative syndrome 2, 615122	PRIMARY IMMUNODEFICIENCIES PANEL HEREDITARY CANCER PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹
CD28	100%	100%	100%	99.1%	?Immunodeficiency 123 with HPV-related verrucosis, 620901	PRIMARY IMMUNODEFICIENCIES PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS
CD2AP	100%	100%	100%	99.4%	Glomerulosclerosis, focal segmental, 3, 607832	RENAL DISORDERS PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹

CD320	100%	100%	100%	99%	Methylmalonic aciduria, transient, due to transcobalamin receptor defect, 613646	METABOLIC DISORDERS PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹
CD36	100%	100%	100%	99.5%	Platelet glycoprotein IV deficiency, 608404;{Coronary heart disease, susceptibility to, 7}, 610938;{Malaria, cerebral, susceptibility to}, 611162;{Malaria, cerebral, reduced risk of}, 611162	HEMOSTATIC/THROMB OTIC DISORDERS PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS
CD3D	100%	100%	100%	99%	Immunodeficiency 19, severe combined, 615617	PRIMARY IMMUNODEFICIENCIES PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹ SEVERE COMBINED IMMUNODEFICIENCY (SCID) PANEL

CD3E	100%	100%	100%	99.4%	Immunodeficiency 18, 615615;Immunodeficiency 18, SCID variant, 615615	PRIMARY IMMUNODEFICIENCIES PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL¹ SEVERE COMBINED IMMUNODEFICIENCY (SCID) PANEL
CD3G	100%	100%	100%	99.7%	Immunodeficiency 17, CD3 gamma deficient, 615607	PRIMARY IMMUNODEFICIENCIES PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL¹ SEVERE COMBINED IMMUNODEFICIENCY (SCID) PANEL

CD4	100%	100%	100%	98.8%	Immunodeficiency 79, 619238;OKT4 epitope deficiency, 613949	PRIMARY IMMUNODEFICIENCIES PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS
CD40	100%	100%	100%	99.4%	Immunodeficiency with hyper-IgM, type 3, 606843	PRIMARY IMMUNODEFICIENCIES PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹
CD40LG	100%	100%	99.4%	69.2%	Immunodeficiency, X-linked, with hyper-IgM, 308230	PRIMARY IMMUNODEFICIENCIES PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS

CD46	100%	100%	100%	99.6%	{Hemolytic uremic syndrome, atypical, susceptibility to, 2}, 612922	RENAL DISORDERS PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS HEMOSTATIC/THROMB OTIC DISORDERS PANEL PRIMARY IMMUNODEFICIENCIES PANEL
CD48	100%	99.9%	100%	99.6%		PRIMARY IMMUNODEFICIENCIES PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS

CD55	96.1%	92.5%	100%	99.3%	[Blood group Cromer], 613793;Complement hyperactivation, angiopathic thrombosis, and protein-losing enteropathy, 226300	PRIMARY IMMUNODEFICIENCIES PANEL LYMPHATIC ANOMALIES PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL¹
CD59	100%	99.9%	100%	99.9%	Hemolytic anemia, CD59-mediated, with or without immune-mediated polyneuropathy, 612300	COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL¹ POLYNEUROPATHIES PANEL¹ PRIMARY IMMUNODEFICIENCIES PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS

CD70	100%	100%	100%	98%	Lymphoproliferative syndrome 3, 618261	PRIMARY IMMUNODEFICIENCIES PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS HEREDITARY CANCER PANEL
CD79A	100%	99.5%	100%	97.5%	Agammaglobulinemia 3, 613501	PRIMARY IMMUNODEFICIENCIES PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹
CD79B	100%	100%	100%	98.5%	Agammaglobulinemia 6, 612692	PRIMARY IMMUNODEFICIENCIES PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹

CD81	99.9%	98.7%	100%	97.7%	Immunodeficiency, common variable, 6, 613496	PRIMARY IMMUNODEFICIENCIES PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹
CD8A	100%	100%	100%	98.7%	Immunodeficiency 116, 608957	SEVERE COMBINED IMMUNODEFICIENCY (SCID) PANEL PRIMARY IMMUNODEFICIENCIES PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹
CD96	100%	100%	100%	99.8%	C syndrome, 211750	MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS

CDAN1	100%	100%	100%	98.3%	Dyserythropoietic anemia, congenital, type Ia, 224120	INHERITED BONE MARROW FAILURE AND/OR PREDISPOSITION PANEL TO HEMATOLOGICAL MALIGNANCIES SKIN DISORDERS PANEL ¹ IRON DISORDERS PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹
CDC14A	100%	100%	100%	99.5%	Deafness, autosomal recessive 32, with or without immotile sperm, 608653	HEARING IMPAIRMENT PANEL (INCLUDING GJB2) MALE INFERTILITY PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹

CDC20	100%	100%	100%	98.8%	Oocyte/zygote/embryo maturation arrest 14, 620276	UNSUCCESSFUL IVF TREATMENTS, RECURRENT MOLAR PREGNANCIES AND/OR RECURRENT PREGNANCY LOSS PANEL ¹ MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS
CDC40	100%	100%	100%	99.4%	?Pontocerebellar hypoplasia, type 15, 619302	MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS

CDC42	100%	100%	100%	99.5%	Takenouchi-Kosaki syndrome, 616737	HEARING IMPAIRMENT PANEL (INCLUDING GJB2) HEMOSTATIC/THROMB OTIC DISORDERS PANEL PRIMARY IMMUNODEFICIENCIES PANEL SHORT STATURE/SKELETAL DYSPLASIA PANEL WITH GENOME WIDE CNV ANALYSIS LYMPHATIC ANOMALIES PANEL WITH GENOME WIDE CNV ANALYSIS INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS NOONAN SYNDROME / RASOPATHY PANEL
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CDC42BPB	100%	100%	100%	98.7%	Chilton-Okur-Chung neurodevelopmental syndrome, 619841	INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS
CDC45	100%	100%	100%	99.3%	Meier-Gorlin syndrome 7, 617063	CRANIOFACIAL ANOMALIES PANEL WITH GENOME WIDE CNV ANALYSIS SHORT STATURE/SKELETAL DYSPLASIA PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL¹ OROFACIAL CLEFTING PANEL WITH GENOME WIDE CNV ANALYSIS

CDC6	100%	100%	100%	99%	?Meier-Gorlin syndrome 5, 613805	SHORT STATURE/SKELETAL DYSPLASIA PANEL WITH GENOME WIDE CNV ANALYSIS INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹
CDC73	100%	100%	100%	99.5%	Hyperparathyroidism, familial primary, 145000;Parathyroid adenoma with cystic changes, 145001;Parathyroid carcinoma, 608266;Hyperparathyroidism-jaw tumor syndrome, 145001	SHORT STATURE/SKELETAL DYSPLASIA PANEL WITH GENOME WIDE CNV ANALYSIS RENAL DISORDERS PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS HEREDITARY CANCER PANEL

CDCA7	100%	100%	100%	99.1%	Immunodeficiency-centromeric instability-facial anomalies syndrome 3, 616910	PRIMARY IMMUNODEFICIENCIES PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹
CDH1	100%	100%	100%	98.9%	Ovarian cancer, somatic, 167000;Blepharocheilodontic syndrome 1, 119580;Diffuse gastric and lobular breast cancer syndrome with or without cleft lip and/or palate, 137215;Endometrial carcinoma, somatic, 608089;Breast cancer, lobular, somatic, 114480	PANEL HEREDITARY CANCER (CDH1, CTNNA1, MLH1, MSH2, MSH6, PMS2) MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS OROFACIAL CLEFTING PANEL WITH GENOME WIDE CNV ANALYSIS HEREDITARY CANCER PANEL

CDH11	100%	100%	100%	99.7%	Teebi hypertelorism syndrome 2, 619736;Elsahy-Waters syndrome, 211380	INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹
CDH15	100%	100%	100%	97.6%	Intellectual developmental disorder, autosomal dominant 3, 612580	INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS

CDH2	100%	100%	100%	99.3%	Arrhythmogenic right ventricular dysplasia 14, 618920;?Attention deficit-hyperactivity disorder 8, 619957;Agenesis of corpus callosum, cardiac, ocular, and genital syndrome, 618929	VISION DISORDERS PANEL DISORDERS/DIFFERENCES OF SEX DEVELOPMENT (DSD) / PRIMARY ADRENAL INSUFFICIENCY PANEL HEART DISORDERS PANEL ¹ INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS
CDH23	100%	100%	100%	98.9%	Usher syndrome, type 1D, 601067;{Pituitary adenoma 5, multiple types}, 617540;Usher syndrome, type 1D/F digenic, 601067;Deafness, autosomal recessive 12, 601386	VISION DISORDERS PANEL HEARING IMPAIRMENT PANEL (INCLUDING GJB2) MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹ HEREDITARY CANCER PANEL

CDH3	100%	100%	100%	99.2%	Hypotrichosis, congenital, with juvenile macular dystrophy, 601553;Ectodermal dysplasia, ectrodactyly, and macular dystrophy, 225280	VISION DISORDERS PANEL SKIN DISORDERS PANEL ¹ SHORT STATURE/SKELETAL DYSPLASIA PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹
CDH4	100%	100%	100%	98.3%		VISION DISORDERS PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS
CDHR1	100%	100%	100%	99.1%	Macular dystrophy, retinal, 613660;Cone-rod dystrophy 15, 613660;Retinitis pigmentosa 65, 613660	VISION DISORDERS PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹

CDIN1	100%	100%	100%	99.5%	Dyserythropoietic anemia, congenital, type Ib, 615631	INHERITED BONE MARROW FAILURE AND/OR PREDISPOSITION PANEL TO HEMATOLOGICAL MALIGNANCIES IRON DISORDERS PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹
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CDK10	100%	100%	100%	99.3%	Al Kaissi syndrome, 617694	VISION DISORDERS PANEL SHORT STATURE/SKELETAL DYSPLASIA PANEL WITH GENOME WIDE CNV ANALYSIS INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL¹
CDK13	100%	100%	100%	98.9%	Congenital heart defects, dysmorphic facial features, and intellectual developmental disorder, 617360	CONGENITAL HEARTDISEASE PANEL¹ HEART DISORDERS PANEL¹ INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS

CDK16	100%	99.7%	98.5%	65.6%		INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS
CDK19	100%	100%	100%	99%	Developmental and epileptic encephalopathy 87, 618916	EPILEPSY PANEL INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS
CDK4	100%	100%	100%	98.9%	{Melanoma, cutaneous malignant, 3}, 609048	PANEL MELANOMA, BASIC (CDKN2A, CDK4, MITF P.(GLU318LYS) ¹ PANEL MELANOMA, EXTENSIVE (CDKN2A, CDK4, MITF P.(GLU318LYS), BAP1, POT1, TERT PROMOTER) ¹ MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS HEREDITARY CANCER PANEL

CDK5	100%	100%	100%	98.5%	?Lissencephaly 7 with cerebellar hypoplasia, 616342	MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹
CDK5RAP2	100%	100%	100%	99.5%	Microcephaly 3, primary, autosomal recessive, 604804	INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹
CDK6	100%	100%	100%	99.5%	?Microcephaly 12, primary, autosomal recessive, 616080	MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹

CDK8	100%	100%	100%	99.5%	Intellectual developmental disorder with hypotonia and behavioral abnormalities, 618748	INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS
CDKL5	95.7%	95.5%	97.9%	66.9%	Developmental and epileptic encephalopathy 2, 300672	EPILEPSY PANEL INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS MITOCHONDRIAL DISORDERS PANEL
CDKN1A	100%	100%	100%	99.6%		MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS HEREDITARY CANCER PANEL

CDKN1B	100%	100%	100%	99.4%	Multiple endocrine neoplasia, type IV, 610755	MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS HEREDITARY CANCER PANEL
CDKN1C	99.9%	99.6%	99.9%	94.5%	IMAGE syndrome, 614732;Beckwith-Wiedemann syndrome, 130650	MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS OROFACIAL CLEFTING PANEL WITH GENOME WIDE CNV ANALYSIS HEREDITARY CANCER PANEL DISORDERS/DIFFEREN CES OF SEX DEVELOPMENT (DSD) / PRIMARY ADRENAL INSUFFICIENCY PANEL TALL STATURE PANEL WITH GENOME WIDE CNV ANALYSIS SHORT STATURE/SKELETAL DYSPLASIA PANEL WITH GENOME WIDE CNV ANALYSIS INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS

CDKN2A	100%	100%	100%	97.6%	{Melanoma and neural system tumor syndrome}, 155755;{Melanoma, cutaneous malignant, 2}, 155601;{Melanoma-pancreatic cancer syndrome}, 606719	PANEL MELANOMA, BASIC (CDKN2A, CDK4, MITF P.(GLU318LYS) ¹ PANEL MELANOMA, EXTENSIVE (CDKN2A, CDK4, MITF P.(GLU318LYS), BAP1, POT1, TERT PROMOTER) ¹ MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS PANEL HEREDITARY PANCREATIC CANCER (BRCA1, BRCA2, PALB2, ATM, CDKN2A, MLH1, MSH2, MSH6, PMS2, STK11) HEREDITARY CANCER PANEL
CDKN2B	100%	100%	100%	98.4%		MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS HEREDITARY CANCER PANEL

CDKN2C	100%	100%	100%	98.4%		MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS HEREDITARY CANCER PANEL
CDON	100%	100%	100%	99.6%	Holoprosencephaly 11, 614226	VISION DISORDERS PANEL CRANIOFACIAL ANOMALIES PANEL WITH GENOME WIDE CNV ANALYSIS INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS

CDSN	100%	100%	100%	99.4%	Hypotrichosis 2, 146520;Peeling skin syndrome 1, 270300	CRANIOFACIAL ANOMALIES PANEL WITH GENOME WIDE CNV ANALYSIS SKIN DISORDERS PANEL ¹ MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹
CDT1	100%	99.7%	100%	98.9%	Meier-Gorlin syndrome 4, 613804	SHORT STATURE/SKELETAL DYSPLASIA PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹
CDY1	50%	50%	48.5%	26.8%		MALE INFERTILITY PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS

CDY1B	50%	50%	48.3%	29.1%		MALE INFERTILITY PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS
CDY2A	50%	50%	49.3%	28.2%		MALE INFERTILITY PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS
CDY2B	50%	50%	49.4%	27.7%		MALE INFERTILITY PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS
CEACAM16	100%	100%	100%	98.5%	Deafness, autosomal dominant 4B, 614614;Deafness, autosomal recessive 113, 618410	HEARING IMPAIRMENT PANEL (INCLUDING GJB2) MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹

CEBPA	100%	99.7%	99.7%	91.4%	Leukemia, acute myeloid, somatic, 601626;?Leukemia, acute myeloid, 601626	INHERITED BONE MARROW FAILURE AND/OR PREDISPOSITION PANEL TO HEMATOLOGICAL MALIGNANCIES MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS HEREDITARY CANCER PANEL
CEBPE	100%	100%	100%	97.8%	?Immunodeficiency 108 with autoinflammation, 260570;Specific granule deficiency, 245480	PRIMARY IMMUNODEFICIENCIES PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹
CEL	100%	99.8%	99.2%	90.4%	Maturity-onset diabetes of the young, type VIII, 609812	METABOLIC DISORDERS PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS

CELA2A	100%	100%	100%	98.7%	Abdominal obesity-metabolic syndrome 4, 618620	MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS
CELF2	100%	100%	100%	99.2%	Developmental and epileptic encephalopathy 97, 619561	EPILEPSY PANEL INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS
CELSR1	100%	99.7%	100%	98.3%	Lymphatic malformation 9, 619319	SKIN DISORDERS PANEL ¹ LYMPHATIC ANOMALIES PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS

CELSR3	100%	100%	100%	98.9%		INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS
CENATAC	100%	100%	100%	99.5%	?Mosaic variegated aneuploidy syndrome 4, 620153	MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS
CENPE	100%	100%	100%	99.1%	?Microcephaly 13, primary, autosomal recessive, 616051	SHORT STATURE/SKELETAL DYSPLASIA PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹

CENPF	100%	100%	100%	99.5%	Stromme syndrome, 243605	CILIOPATHIES PANEL INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹
CENPJ	100%	100%	100%	99.7%		SHORT STATURE/SKELETAL DYSPLASIA PANEL WITH GENOME WIDE CNV ANALYSIS INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹
CENPT	100%	100%	100%	99.5%	?Short stature and microcephaly with genital anomalies, 618702	MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS

CEP104	100%	100%	100%	99%	Joubert syndrome 25, 616781;Intellectual developmental disorder, autosomal recessive 77, 619988	CILIOPATHIES PANEL INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹
CEP112	100%	100%	100%	99.2%	Spermatogenic failure 44, 619044	MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS

CEP120	100%	100%	100%	99.2%	Short-rib thoracic dysplasia 13 with or without polydactyly, 616300;Joubert syndrome 31, 617761	VISION DISORDERS PANEL CILIOPATHIES PANEL SHORT STATURE/SKELETAL DYSPLASIA PANEL WITH GENOME WIDE CNV ANALYSIS INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS RENAL DISORDERS PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹
CEP135	100%	100%	100%	99.5%	Microcephaly 8, primary, autosomal recessive, 614673	INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹

CEP152	100%	100%	100%	99.6%	Microcephaly 9, primary, autosomal recessive, 614852;Seckel syndrome 5, 613823	SHORT STATURE/SKELETAL DYSPLASIA PANEL WITH GENOME WIDE CNV ANALYSIS INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹
CEP162	100%	100%	100%	99.2%		VISION DISORDERS PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS

CEP164	100%	100%	100%	99.3%	Nephronophthisis 15, 614845	VISION DISORDERS PANEL CILIOPATHIES PANEL RENAL DISORDERS PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹
CEP19	100%	100%	100%	99%	Morbid obesity and spermatogenic failure, 615703	MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹
CEP250	100%	99.9%	100%	99.1%	Cone-rod dystrophy and hearing loss 2, 618358	VISION DISORDERS PANEL HEARING IMPAIRMENT PANEL (INCLUDING GJB2) MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS

CEP290	100%	100%	100%	99.5%	Leber congenital amaurosis 10, 611755;Joubert syndrome 5, 610188;Senior-Loken syndrome 6, 610189;?Bardet-Biedl syndrome 14, 615991;Meckel syndrome 4, 611134	VISION DISORDERS PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹ RENAL DISORDERS PANEL INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS CILIOPATHIES PANEL SHORT STATURE/SKELETAL DYSPLASIA PANEL WITH GENOME WIDE CNV ANALYSIS
CEP295	100%	100%	100%	99.5%	Seckel syndrome 11, 620767	INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS

CEP41	100%	100%	100%	99.3%	Joubert syndrome 15, 614464	VISION DISORDERS PANEL CILIOPATHIES PANEL DISORDERS/DIFFEREN CES OF SEX DEVELOPMENT (DSD) / PRIMARY ADRENAL INSUFFICIENCY PANEL INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS RENAL DISORDERS PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL¹
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CEP55	100%	100%	100%	99.4%	Multinucleated neurons, anhydramnios, renal dysplasia, cerebellar hypoplasia, and hydranencephaly, 236500	CILIOPATHIES PANEL INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS RENAL DISORDERS PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹
CEP57	100%	100%	100%	99.6%	Mosaic variegated aneuploidy syndrome 2, 614114	SHORT STATURE/SKELETAL DYSPLASIA PANEL WITH GENOME WIDE CNV ANALYSIS INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹

CEP63	92.8%	92.8%	100%	99.6%	?Seckel syndrome 6, 614728	SHORT STATURE/SKELETAL DYSPLASIA PANEL WITH GENOME WIDE CNV ANALYSIS INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹
CEP78	100%	100%	100%	99.6%	Cone-rod dystrophy and hearing loss, 617236	VISION DISORDERS PANEL HEARING IMPAIRMENT PANEL (INCLUDING GJB2) MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹

CEP83	100%	100%	100%	99.5%	Nephronophthisis 18, 615862	VISION DISORDERS PANEL CILIOPATHIES PANEL LIVER DISORDERS PANEL INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS RENAL DISORDERS PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL¹
CEP85L	100%	100%	100%	99.7%	Lissencephaly 10, 618873	EPILEPSY PANEL INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS

CEP89	100%	100%	100%	99.1%		INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS MITOCHONDRIAL DISORDERS PANEL
CERKL	100%	100%	100%	99.5%	Retinitis pigmentosa 26, 608380	VISION DISORDERS PANEL METABOLIC DISORDERS PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹
CERS1	98.4%	94.4%	100%	95.8%	Epilepsy, progressive myoclonic, 8, 616230	MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹

CERS3	100%	100%	100%	99.5%	Ichthyosis, congenital, autosomal recessive 9, 615023	SKIN DISORDERS PANEL ¹ METABOLIC DISORDERS PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹
CERT1	100%	100%	100%	99.3%	Intellectual developmental disorder, autosomal dominant 34, 616351	EPILEPSY PANEL INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS
CES1	99.8%	99.5%	99.8%	97.3%	Drug metabolism, altered, CES1-related, 618057	MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS
CETP	100%	100%	100%	99%	[High density lipoprotein cholesterol level QTL 10], 143470;Hyperalphalipoproteinemia, 143470	MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS

CFAP251	100%	100%	100%	99.2%	Spermatogenic failure 33, 618152	CILIOPATHIES PANEL MALE INFERTILITY PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS
CFAP276	100%	100%	100%	99.1%		POLYNEUROPATHIES PANEL ¹ MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS
CFAP298	100%	100%	100%	99.5%	Ciliary dyskinesia, primary, 26, 615500	CILIOPATHIES PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹
CFAP300	100%	100%	100%	99.1%	Ciliary dyskinesia, primary, 38, 618063	CILIOPATHIES PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹

CFAP410	100%	100%	100%	97.4%	Retinal dystrophy with macular staphyloma, 617547;Spondylometaphyseal dysplasia, axial, 602271	AMYOTROPHIC LATERAL SCLEROSIS, ALS PANEL COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL¹ SHORT STATURE/SKELETAL DYSPLASIA PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS VISION DISORDERS PANEL CILIOPATHIES PANEL
CFAP418	100%	100%	100%	99.4%	Retinitis pigmentosa 64, 614500;Cone-rod dystrophy 16, 614500;Bardet-Biedl syndrome 21, 617406	VISION DISORDERS PANEL CILIOPATHIES PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL¹

CFAP43	100%	100%	100%	99.2%	Hydrocephalus, normal pressure, 1, 236690;Spermatogenic failure 19, 617592	MALE INFERTILITY PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS
CFAP44	100%	100%	100%	99.5%	Spermatogenic failure 20, 617593	CILIOPATHIES PANEL MALE INFERTILITY PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS
CFAP45	100%	100%	100%	98.5%	Heterotaxy, visceral, 11, autosomal, with male infertility, 619608	CONGENITAL HEARTDISEASE PANEL ¹ CILIOPATHIES PANEL HEART DISORDERS PANEL ¹ MALE INFERTILITY PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS
CFAP47	100%	99.9%	99.1%	69.9%	Spermatogenic failure, X-linked 3, 301059	MALE INFERTILITY PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS

CFAP52	100%	100%	100%	99.4%	Heterotaxy, visceral, 10, autosomal, with male infertility, 619607	CONGENITAL HEARTDISEASE PANEL¹ CILIOPATHIES PANEL HEART DISORDERS PANEL¹ MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS
CFAP53	100%	100%	100%	99.4%	Heterotaxy, visceral, 6, autosomal recessive, 614779	CONGENITAL HEARTDISEASE PANEL¹ CILIOPATHIES PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL¹ HEART DISORDERS PANEL¹
CFAP54	100%	100%	100%	99.4%		MALE INFERTILITY PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS

CFAP57	100%	100%	100%	99.1%	Spermatogenic failure 95, 620917	MALE INFERTILITY PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS
CFAP58	100%	100%	100%	99.3%	Spermatogenic failure 49, 619144	MALE INFERTILITY PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS MITOCHONDRIAL DISORDERS PANEL
CFAP61	100%	100%	100%	99.6%	Spermatogenic failure 84, 620409	MALE INFERTILITY PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS
CFAP65	100%	100%	100%	98.6%	Spermatogenic failure 40, 618664	MALE INFERTILITY PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS

CFAP69	100%	100%	100%	99.6%	Spermatogenic failure 24, 617959	CILIOPATHIES PANEL MALE INFERTILITY PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS
CFAP70	100%	100%	100%	99.5%	?Spermatogenic failure 41, 618670	MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS MALE INFERTILITY PANEL
CFAP74	100%	99.9%	99.7%	96.7%	Ciliary dyskinesia, primary, 49, without situs inversus, 620197	MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS
CFAP91	100%	100%	100%	98.8%	Spermatogenic failure 51, 619177	MALE INFERTILITY PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS

CFB	100%	100%	100%	99.4%	?Complement factor B deficiency, 615561;{Hemolytic uremic syndrome, atypical, susceptibility to, 4}, 612924;{Macular degeneration, age-related, 14, reduced risk of}, 615489	HEMOSTATIC/THROMBOTIC DISORDERS PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS RENAL DISORDERS PANEL PRIMARY IMMUNODEFICIENCIES PANEL
CFC1	100%	100%	100%	99.9%	Heterotaxy, visceral, 2, autosomal, 605376	CONGENITAL HEARTDISEASE PANEL ¹ CILIOPATHIES PANEL HEART DISORDERS PANEL ¹ LIVER DISORDERS PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS

CFD	100%	100%	99.9%	94.8%	Complement factor D deficiency, 613912	PRIMARY IMMUNODEFICIENCIES PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹
CFH	97.5%	97.5%	100%	99.6%	{Macular degeneration, age-related, 4}, 610698;Basal laminar drusen, 126700;Complement factor H deficiency, 609814;{Hemolytic uremic syndrome, atypical, susceptibility to, 1}, 235400	VISION DISORDERS PANEL COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹ MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS RENAL DISORDERS PANEL PRIMARY IMMUNODEFICIENCIES PANEL HEMOSTATIC/THROMB OTIC DISORDERS PANEL

CFHR1	94.6%	92.9%	90.9%	74.7%	{Macular degeneration, age-related, reduced risk of}, 603075;{Hemolytic uremic syndrome, atypical, susceptibility to}, 235400	RENAL DISORDERS PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS
CFHR2	76.4%	76.4%	100%	99.1%		MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS
CFHR3	94.3%	93.6%	91.3%	80.7%	{Macular degeneration, age-related, reduced risk of}, 603075;{Hemolytic uremic syndrome, atypical, susceptibility to}, 235400	RENAL DISORDERS PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS
CFHR4	100%	100%	99.8%	97.5%		MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS
CFHR5	100%	100%	100%	99.5%	Nephropathy due to CFHR5 deficiency, 614809	RENAL DISORDERS PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS

CFI	100%	100%	100%	99.7%	{Hemolytic uremic syndrome, atypical, susceptibility to, 3}, 612923;{Macular degeneration, age-related, 13, susceptibility to}, 615439;Complement factor I deficiency, 610984	HEMOSTATIC/THROMBOTIC DISORDERS PANEL PRIMARY IMMUNODEFICIENCIES PANEL COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹ MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS RENAL DISORDERS PANEL
CFL2	100%	100%	100%	99.9%	Nemaline myopathy 7, autosomal recessive, 610687	MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹ MUSCLE DISORDERS PANEL
CFP	100%	99.6%	98.7%	64.5%	Properdin deficiency, X-linked, 312060	PRIMARY IMMUNODEFICIENCIES PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS

CFTR	100%	100%	100%	98.7%	Cystic fibrosis, 219700;Sweat chloride elevation without CF;Congenital bilateral absence of vas deferens, 277180;{Pancreatitis, hereditary}, 167800;{Bronchiectasis with or without elevated sweat chloride 1, modifier of}, 211400;{Hypertrypsinemia, neonatal}	MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹ SKIN DISORDERS PANEL ¹ PRIMARY IMMUNODEFICIENCIES PANEL LIVER DISORDERS PANEL MALE INFERTILITY PANEL METABOLIC DISORDERS PANEL
CGN	100%	100%	100%	98.9%		HEARING IMPAIRMENT PANEL (INCLUDING GJB2) MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS

CHAMP1	100%	100%	100%	99.4%	Neurodevelopmental disorder with hypotonia, impaired language, and dysmorphic features, 616579	INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS
CHASERR					Neurodevelopmental disorder with dysmorphic facies, absent speech and ambulation, and brain abnormalities, 621012	INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS
CHAT	100%	99.9%	100%	99.3%	Myasthenic syndrome, congenital, 6, presynaptic, 254210	FETAL AKINESIA PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹ MUSCLE DISORDERS PANEL

CHCHD10	100%	100%	99.8%	97.2%	?Myopathy, isolated mitochondrial, autosomal dominant, 616209;Spinal muscular atrophy, Jokela type, 615048;Frontotemporal dementia and/or amyotrophic lateral sclerosis 2, 615911	AMYOTROPHIC LATERAL SCLEROSIS, ALS PANEL POLYNEUROPATHIES PANEL ¹ MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS MITOCHONDRIAL DISORDERS PANEL MUSCLE DISORDERS PANEL
CHCHD2	100%	100%	100%	99.6%	Parkinson disease 22, autosomal dominant, 616710	MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS MITOCHONDRIAL DISORDERS PANEL PARKINSON DISEASE PANEL
CHD1	100%	100%	100%	99.5%	Pilarowski-Bjornsson syndrome, 617682	INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS

CHD2	100%	100%	100%	99.3%	Developmental and epileptic encephalopathy 94, 615369	EPILEPSY PANEL INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS
CHD3	99.9%	99.6%	100%	98%	Snijders Blok-Campeau syndrome, 618205	INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS
CHD4	100%	100%	100%	98.8%	Sifrim-Hitz-Weiss syndrome, 617159	CONGENITAL HEARTDISEASE PANEL ¹ HEART DISORDERS PANEL ¹ INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS

CHD5	100%	100%	100%	98.3%	Parenti-Mignot neurodevelopmental syndrome, 619873	EPILEPSY PANEL INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS
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CHD7	100%	100%	100%	99.4%	Hypogonadotropic hypogonadism 5 with or without anosmia, 612370;CHARGE syndrome, 214800	VISION DISORDERS PANEL OROFACIAL CLEFTING PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS PRIMARY IMMUNODEFICIENCIES PANEL INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS HEART DISORDERS PANEL ¹ HYPOGONADOTROPIC HYPOGONADISM (KALLMANN) PANEL DISORDERS/DIFFEREN CES OF SEX DEVELOPMENT (DSD) / PRIMARY ADRENAL INSUFFICIENCY PANEL HEARING IMPAIRMENT PANEL (INCLUDING GJB2) CONGENITAL HEARTDISEASE PANEL ¹ CRANIOFACIAL ANOMALIES PANEL WITH GENOME WIDE CNV ANALYSIS
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CHD8	100%	100%	100%	99.3%	Intellectual developmental disorder with autism and macrocephaly, 615032	TALL STATURE PANEL WITH GENOME WIDE CNV ANALYSIS LIVER DISORDERS PANEL INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS
CHEK1	100%	100%	100%	99.4%	Oocyte/zygote/embryo maturation arrest 21, 620610	MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS
CHEK2	100%	99.8%	100%	98.7%	Prostate cancer, somatic, 176807;Osteosarcoma, somatic, 259500;Tumor predisposition syndrome 4, breast/prostate/colorectal, 609265	MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS HEREDITARY CANCER PANEL
CHIT1	100%	100%	100%	99.2%	[Chitotriosidase deficiency], 614122	METABOLIC DISORDERS PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS

CHKA	100%	97.9%	100%	98%	Neurodevelopmental disorder with microcephaly, movement abnormalities, and seizures, 620023	INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹
CHKB	100%	100%	100%	99.1%	Muscular dystrophy, congenital, megaconial type, 602541	MITOCHONDRIAL DISORDERS PANEL COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹ METABOLIC DISORDERS PANEL INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS SKIN DISORDERS PANEL ¹ HEART DISORDERS PANEL ¹ MUSCLE DISORDERS PANEL

CHM	99.1%	97.5%	97.8%	67.7%	Choroideremia, 303100	VISION DISORDERS PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS
CHMP1A	100%	100%	100%	98.2%	Pontocerebellar hypoplasia, type 8, 614961	MOVEMENT DISORDERS PANEL INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹
CHMP2B	92.7%	88.2%	100%	99%	Frontotemporal dementia and/or amyotrophic lateral sclerosis 7, 600795	AMYOTROPHIC LATERAL SCLEROSIS, ALS PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS PARKINSON DISEASE PANEL

CHMP4B	100%	100%	100%	97.3%	Cataract 31, multiple types, 605387	VISION DISORDERS PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS
CHN1	96.5%	96.5%	100%	99.3%	Duane retraction syndrome 2, 604356	VISION DISORDERS PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS
CHP1	100%	100%	100%	99.4%	?Spastic ataxia 9, autosomal recessive, 618438	MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹
CHRD1	100%	99.8%	98.5%	67.9%	Megalocornea 1, X-linked, 309300	VISION DISORDERS PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS

CHRM1	100%	100%	100%	99.2%		INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS
CHRM2	100%	100%	100%	99.4%		HEART DISORDERS PANEL ¹ MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS
CHRM3	100%	100%	100%	99.4%	Prune belly syndrome, 100100	LIVER DISORDERS PANEL RENAL DISORDERS PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹

CHRNA1	100%	100%	100%	99.2%	Myasthenic syndrome, congenital, 1B, fast-channel, 608930;Myasthenic syndrome, congenital, 1A, slow-channel, 601462;Multiple pterygium syndrome, lethal type, 253290	FETAL AKINESIA PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹ MUSCLE DISORDERS PANEL
CHRNA2	100%	100%	100%	97.8%	Epilepsy, nocturnal frontal lobe, type 4, 610353	EPILEPSY PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS
CHRNA3	100%	100%	100%	99.1%	{Lung cancer susceptibility 2}, 612052;Bladder dysfunction, autonomic, with impaired pupillary reflex and secondary CAKUT, 191800	LIVER DISORDERS PANEL RENAL DISORDERS PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹

CHRNA4	100%	100%	100%	93.4%	{Nicotine addiction, susceptibility to}, 188890;Epilepsy, nocturnal frontal lobe, 1, 600513	EPILEPSY PANEL INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS
CHRNA1	100%	100%	100%	98%	Myasthenic syndrome, congenital, 2C, associated with acetylcholine receptor deficiency, 616314;Myasthenic syndrome, congenital, 2A, slow-channel, 616313	FETAL AKINESIA PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹ MUSCLE DISORDERS PANEL
CHRNA2	100%	100%	100%	98.8%	Epilepsy, nocturnal frontal lobe, 3, 605375	EPILEPSY PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS

CHRND	100%	100%	100%	98.9%	?Myasthenic syndrome, congenital, 3C, associated with acetylcholine receptor deficiency, 616323;Multiple pterygium syndrome, lethal type, 253290;Myasthenic syndrome, congenital, 3B, fast-channel, 616322;?Myasthenic syndrome, congenital, 3A, slow-channel, 616321	FETAL AKINESIA PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹ MUSCLE DISORDERS PANEL
CHRNE	100%	100%	100%	98.9%	Myasthenic syndrome, congenital, 4A, slow-channel, 605809;Myasthenic syndrome, congenital, 4C, associated with acetylcholine receptor deficiency, 608931;Myasthenic syndrome, congenital, 4B, fast-channel, 616324	FETAL AKINESIA PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹ MUSCLE DISORDERS PANEL

CHRNA	100%	100%	100%	99%	Multiple pterygium syndrome, lethal type, 253290;Escobar syndrome, 265000	FETAL AKINESIA PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹ OROFACIAL CLEFTING PANEL WITH GENOME WIDE CNV ANALYSIS MUSCLE DISORDERS PANEL
CHST11	100%	100%	100%	98.5%	?Osteochondrodysplasia, brachydactyly, and overlapping malformed digits, 618167	SHORT STATURE/SKELETAL DYSPLASIA PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹

CHST14	100%	100%	100%	97.5%	Ehlers-Danlos syndrome, musculocontractural type 1, 601776	FETAL AKINESIA PANEL SKIN DISORDERS PANEL ¹ HEMOSTATIC/THROMB OTIC DISORDERS PANEL SHORT STATURE/SKELETAL DYSPLASIA PANEL WITH GENOME WIDE CNV ANALYSIS METABOLIC DISORDERS PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹ OROFACIAL CLEFTING PANEL WITH GENOME WIDE CNV ANALYSIS
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CHST3	100%	100%	100%	98.8%	Spondyloepiphyseal dysplasia with congenital joint dislocations, 143095	SHORT STATURE/SKELETAL DYSPLASIA PANEL WITH GENOME WIDE CNV ANALYSIS METABOLIC DISORDERS PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹
CHST6	100%	100%	100%	99.9%	Macular corneal dystrophy, 217800	VISION DISORDERS PANEL METABOLIC DISORDERS PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹

CHST8	100%	100%	100%	98.5%		SKIN DISORDERS PANEL ¹ MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹
CHSY1	99.8%	97.9%	100%	98.7%	Temtamy preaxial brachydactyly syndrome, 605282	SKIN DISORDERS PANEL ¹ HEARING IMPAIRMENT PANEL (INCLUDING GJB2) SHORT STATURE/SKELETAL DYSPLASIA PANEL WITH GENOME WIDE CNV ANALYSIS METABOLIC DISORDERS PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹

CHUK	100%	100%	100%	99.2%	?Popliteal pterygium syndrome, Bartsocas-Papas type 2, 619339;?Cocoon syndrome, 613630	SKIN DISORDERS PANEL ¹ PRIMARY IMMUNODEFICIENCIES PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS
CIAO1	100%	100%	100%	99%	Multiple mitochondrial dysfunctions syndrome 10, 620960	METABOLIC DISORDERS PANEL INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹
CIB1	100%	100%	100%	98.3%	{Epidermodysplasia verruciformis, susceptibility to, 3}, 618267	SKIN DISORDERS PANEL ¹ PRIMARY IMMUNODEFICIENCIES PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS

CIB2	100%	100%	100%	99.4%	Deafness, autosomal recessive 48, 609439;Usher syndrome, type IJ, 614869	VISION DISORDERS PANEL HEARING IMPAIRMENT PANEL (INCLUDING GJB2) MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹
CIBAR1	100%	100%	100%	99.7%	?Polydactyly, postaxial, type A9, 618219	MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS
CIC	100%	100%	100%	98%	Intellectual developmental disorder, autosomal dominant 45, 617600	EPILEPSY PANEL INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS

CIDEC	100%	100%	100%	99.5%	?Lipodystrophy, familial partial, type 5, 615238	METABOLIC DISORDERS PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹
CIITA	100%	100%	100%	98.1%	{Rheumatoid arthritis, susceptibility to}, 180300;MHC class II deficiency 1, 209920	PRIMARY IMMUNODEFICIENCIES PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹ SEVERE COMBINED IMMUNODEFICIENCY (SCID) PANEL

CILK1	100%	100%	100%	99.5%	{Epilepsy, juvenile myoclonic, susceptibility to, 10}, 617924;Endocrine-cerebroosteodysplasia, 612651	SHORT STATURE/SKELETAL DYSPLASIA PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL¹ OROFACIAL CLEFTING PANEL WITH GENOME WIDE CNV ANALYSIS
CIROP	98.3%	94.6%	100%	99.1%	Heterotaxy, visceral, 12, autosomal, 619702	CONGENITAL HEARTDISEASE PANEL¹ HEART DISORDERS PANEL¹ MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL¹

CISD2	100%	100%	100%	99.1%	Wolfram syndrome 2, 604928	VISION DISORDERS PANEL HEARING IMPAIRMENT PANEL (INCLUDING GJB2) MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS MITOCHONDRIAL DISORDERS PANEL COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL¹
CIT	95.8%	95.8%	100%	98.8%	Microcephaly 17, primary, autosomal recessive, 617090	INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL¹

CITED2	100%	100%	100%	99.5%	Atrial septal defect 8, 614433;Ventricular septal defect 2, 614431	CONGENITAL HEARTDISEASE PANEL¹ HEART DISORDERS PANEL¹ MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS
CKAP2L	100%	100%	100%	99.5%	Filippi syndrome, 272440	SKIN DISORDERS PANEL¹ SHORT STATURE/SKELETAL DYSPLASIA PANEL WITH GENOME WIDE CNV ANALYSIS INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL¹
CLCC1	98.6%	98.6%	100%	99.4%	Retinitis pigmentosa 32, 609913	VISION DISORDERS PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS

CLCF1	100%	100%	100%	97.7%	Cold-induced sweating syndrome 2, 610313	MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹
CLCN1	100%	100%	100%	98.6%	Myotonia congenita, recessive, 255700;Myotonia congenita, dominant, 160800;Myotonia levior, 160800	MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹ MUSCLE DISORDERS PANEL
CLCN2	100%	100%	100%	99.3%	Leukoencephalopathy with ataxia, 615651;Hyperaldosteronism, familial, type II, 605635;{Epilepsy, juvenile myoclonic, susceptibility to, 8}, 607628;{Epilepsy, juvenile absence, susceptibility to, 2}, 607628;{Epilepsy, idiopathic generalized, susceptibility to, 11}, 607628	MOVEMENT DISORDERS PANEL RENAL DISORDERS PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹

CLCN3	100%	100%	100%	99.4%	Neurodevelopmental disorder with seizures and brain abnormalities, 619517;Neurodevelopmental disorder with hypotonia and brain abnormalities, 619512	INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹
CLCN4	100%	99.8%	98.2%	65.4%	Raynaud-Claes syndrome, 300114	MOVEMENT DISORDERS PANEL EPILEPSY PANEL INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS

CLCN5	100%	100%	99.2%	70.1%	Proteinuria, low molecular weight, with hypercalciuric nephrocalcinosis, 308990;Hypophosphatemic rickets, 300554;Dent disease 1, 300009;Nephrolithiasis, type I, 310468	SHORT STATURE/SKELETAL DYSPLASIA PANEL WITH GENOME WIDE CNV ANALYSIS RENAL DISORDERS PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS
CLCN6	100%	100%	100%	98.8%	Ceroid lipofuscinosis, neuronal, 15, 619173	MOVEMENT DISORDERS PANEL INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS

CLCN7	100%	100%	100%	98.9%	Hypopigmentation, organomegaly, and delayed myelination and development, 618541;Osteopetrosis, autosomal recessive 4, 611490;Osteopetrosis, autosomal dominant 2, 166600	SHORT STATURE/SKELETAL DYSPLASIA PANEL WITH GENOME WIDE CNV ANALYSIS METABOLIC DISORDERS PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS INHERITED BONE MARROW FAILURE AND/OR PREDISPOSITION PANEL TO HEMATOLOGICAL MALIGNANCIES PRIMARY IMMUNODEFICIENCIES PANEL COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹
CLCNKA	100%	100%	100%	99.2%	Bartter syndrome, type 4b, digenic, 613090	MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS

CLCNKB	100%	100%	100%	98.6%	Bartter syndrome, type 3, 607364;Bartter syndrome, type 4b, digenic, 613090	RENAL DISORDERS PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹
CLDN1	100%	100%	100%	99.4%	Ichthyosis, leukocyte vacuoles, alopecia, and sclerosing cholangitis, 607626	SKIN DISORDERS PANEL ¹ LIVER DISORDERS PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹
CLDN10	100%	100%	100%	99.5%	HELIX syndrome, 617671	SKIN DISORDERS PANEL ¹ RENAL DISORDERS PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹

CLDN11	100%	100%	100%	98.6%	Leukodystrophy, hypomyelinating, 22, 619328	INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS
CLDN14	100%	100%	100%	99.4%	Deafness, autosomal recessive 29, 614035	HEARING IMPAIRMENT PANEL (INCLUDING GJB2) MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹
CLDN16	100%	100%	100%	99%	Hypomagnesemia 3, renal, 248250	RENAL DISORDERS PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹ EPILEPSY PANEL

CLDN19	100%	100%	100%	98.5%	Hypomagnesemia 5, renal, with ocular involvement, 248190	VISION DISORDERS PANEL EPILEPSY PANEL RENAL DISORDERS PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹
CLDN2	100%	99.9%	98.9%	69.8%	?Azoospermia, obstructive, with nephrolithiasis, 301060	MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS
CLDN5	100%	100%	100%	99.2%		INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS

CLDN9	100%	100%	100%	99.2%	Deafness, autosomal recessive 116, 619093	HEARING IMPAIRMENT PANEL (INCLUDING GJB2) MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹
CLEC3B	100%	100%	100%	99.7%	Macular dystrophy, retinal, 4, 619977	VISION DISORDERS PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS
CLEC4D	100%	100%	100%	98.9%		PRIMARY IMMUNODEFICIENCIES PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS
CLEC7A	100%	100%	100%	99.1%	Candidiasis, familial, 4, autosomal recessive, 613108;{Aspergillosis, susceptibility to}, 614079	PRIMARY IMMUNODEFICIENCIES PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS

CLIC2	100%	100%	99.5%	71.9%		INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS
CLIC5	100%	100%	100%	99.4%	?Deafness, autosomal recessive 103, 616042	HEARING IMPAIRMENT PANEL (INCLUDING GJB2) MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS
CLIP1	100%	100%	100%	99.4%		INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹

CLMP	100%	100%	100%	99.6%	Congenital short bowel syndrome, 615237	LIVER DISORDERS PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL¹
CLN3	93.1%	93.1%	100%	98.8%	Ceroid lipofuscinosis, neuronal, 3, 204200	VISION DISORDERS PANEL INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL¹ EPILEPSY PANEL METABOLIC DISORDERS PANEL

CLN5	83%	83%	100%	99.4%	Ceroid lipofuscinosis, neuronal, 5, 256731	MOVEMENT DISORDERS PANEL VISION DISORDERS PANEL EPILEPSY PANEL METABOLIC DISORDERS PANEL INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL¹
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CLN6	100%	100%	100%	99%	Ceroid lipofuscinosis, neuronal, 6B (Kufs type), 204300;Ceroid lipofuscinosis, neuronal, 6A, 601780	MOVEMENT DISORDERS PANEL VISION DISORDERS PANEL EPILEPSY PANEL METABOLIC DISORDERS PANEL INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL¹
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CLN8	100%	100%	100%	99%	Ceroid lipofuscinosis, neuronal, 8, Northern epilepsy variant, 610003;Ceroid lipofuscinosis, neuronal, 8, 600143	VISION DISORDERS PANEL COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹ EPILEPSY PANEL METABOLIC DISORDERS PANEL INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS
CLP1	100%	100%	100%	98.5%	Pontocerebellar hypoplasia, type 10, 615803	MOVEMENT DISORDERS PANEL INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹

CLPB	100%	100%	100%	98.5%	Neutropenia, severe congenital, 9, autosomal dominant, 619813;3-methylglutaconic aciduria, type VIIB, autosomal recessive, 616271;3-methylglutaconic aciduria, type VIIA, autosomal dominant, 619835	MOVEMENT DISORDERS PANEL INHERITED BONE MARROW FAILURE AND/OR PREDISPOSITION PANEL TO HEMATOLOGICAL MALIGNANCIES PRIMARY IMMUNODEFICIENCIES PANEL METABOLIC DISORDERS PANEL INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS MITOCHONDRIAL DISORDERS PANEL COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹
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CLPP	100%	100%	100%	97.4%	Perrault syndrome 3, 614129	HEARING IMPAIRMENT PANEL (INCLUDING GJB2) DISORDERS/DIFFEREN CES OF SEX DEVELOPMENT (DSD) / PRIMARY ADRENAL INSUFFICIENCY PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS MITOCHONDRIAL DISORDERS PANEL COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL¹ PREMATURE OVARIAN INSUFFICIENCY PANEL
CLPX	100%	100%	100%	99.8%	?Protoporphyria, erythropoietic, 2, 618015	MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS

CLRN1	100%	100%	100%	99.2%	Usher syndrome, type 3A, 276902;Retinitis pigmentosa 61, 614180	VISION DISORDERS PANEL HEARING IMPAIRMENT PANEL (INCLUDING GJB2) MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹
CLRN2	100%	100%	100%	99%	Deafness, autosomal recessive 117, 619174	HEARING IMPAIRMENT PANEL (INCLUDING GJB2) MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS
CLTC	99.2%	99.2%	100%	99.6%	Intellectual developmental disorder, autosomal dominant 56, 617854	EPILEPSY PANEL INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS

CLTCL1	100%	100%	100%	99.3%		NEUROLOGICAL PAIN DISORDERS PANEL' MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS
CLUAP1	100%	100%	100%	99.5%		VISION DISORDERS PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS
CLXN	100%	100%	100%	99.4%	Ciliary dyskinesia, primary, 53, 620642	MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL¹
CMAS	100%	100%	100%	99.1%		METABOLIC DISORDERS PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS

CMPK2	100%	100%	100%	99.2%	Basal ganglia calcification, idiopathic, 10, autosomal recessive, 621018	MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹
CNBP	100%	100%	100%	99.7%	Myotonic dystrophy 2, 602668	MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS
CNGA1	100%	100%	100%	99.6%	Retinitis pigmentosa 49, 613756	VISION DISORDERS PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹

CNGA2	99.9%	98.9%	96.4%	60.4%		DISORDERS/DIFFERENCES OF SEX DEVELOPMENT (DSD) / PRIMARY ADRENAL INSUFFICIENCY PANEL HYPOGONADOTROPIC HYPOGONADISM (KALLMANN) PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS
CNGA3	100%	100%	100%	99.3%	Achromatopsia 2, 216900	VISION DISORDERS PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹
CNGB1	100%	100%	100%	98.3%	Retinitis pigmentosa 45, 613767	VISION DISORDERS PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹

CNGB3	100%	100%	100%	99.1%	Achromatopsia 3, 262300	VISION DISORDERS PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹
CNKS2	100%	99.8%	99.1%	69.6%	Intellectual developmental disorder, X-linked syndromic, Houge type, 301008	INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS
CNNM2	100%	100%	100%	98.9%	Hypomagnesemia 6, renal, 613882;Hypomagnesemia, seizures, and impaired intellectual development 1, 616418	EPILEPSY PANEL INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS RENAL DISORDERS PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹

CNNM4	100%	100%	100%	98.7%	Jalili syndrome, 217080	VISION DISORDERS PANEL SKIN DISORDERS PANEL ¹ MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹
CNOT1	100%	100%	100%	99.2%	Vissers-Bodmer syndrome, 619033;Holoprosencephaly 12, with or without pancreatic agenesis, 618500	INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS
CNOT2	100%	100%	100%	99.4%	Intellectual developmental disorder with nasal speech, dysmorphic facies, and variable skeletal anomalies, 618608	INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS

CNOT3	100%	100%	100%	98%	Intellectual developmental disorder with speech delay, autism, and dysmorphic facies, 618672	INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS
CNOT9	97.9%	93.2%	100%	99.8%		INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS
CNP	100%	100%	100%	98.9%	?Leukodystrophy, hypomyelinating, 20, 619071	MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS

CNPY3	100%	100%	100%	98.5%	Developmental and epileptic encephalopathy 60, 617929	INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹
CNTN1	100%	100%	100%	99.5%	Congenital myopathy 12, 612540	MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹ MUSCLE DISORDERS PANEL
CNTN2	100%	100%	100%	99%	Epilepsy, early-onset, 5, with or without developmental delay, 615400	EPILEPSY PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹

CNTNAP1	100%	100%	100%	98.7%	Lethal congenital contracture syndrome 7, 616286;Hypomyelinating neuropathy, congenital, 3, 618186	FETAL AKINESIA PANEL POLYNEUROPATHIES PANEL ¹ INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹
CNTNAP2	100%	100%	100%	99.4%	Pitt-Hopkins like syndrome 1, 610042;{Autism susceptibility 15}, 612100	EPILEPSY PANEL INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹

COA1	100%	100%	100%	99.4%		MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS MITOCHONDRIAL DISORDERS PANEL
COA3	100%	100%	100%	97.7%	?Mitochondrial complex IV deficiency, nuclear type 14, 619058	POLYNEUROPATHIES PANEL ¹ MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS MITOCHONDRIAL DISORDERS PANEL
COA5	82.4%	82.4%	100%	99.3%	?Mitochondrial complex IV, deficiency, nuclear type 9, 616500	MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS MITOCHONDRIAL DISORDERS PANEL COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹

COA6	100%	100%	100%	99.5%	Mitochondrial complex IV deficiency, nuclear type 13, 616501	MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS MITOCHONDRIAL DISORDERS PANEL COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹
COA7	100%	100%	100%	99.5%	Spinocerebellar ataxia, autosomal recessive, with axonal neuropathy 3, 618387	POLYNEUROPATHIES PANEL ¹ MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS MITOCHONDRIAL DISORDERS PANEL COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹

COA8	100%	99.8%	100%	98.9%	Mitochondrial complex IV deficiency, nuclear type 17, 619061	MITOCHONDRIAL DISORDERS PANEL COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL¹ EPILEPSY PANEL INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS VISION DISORDERS PANEL HEARING IMPAIRMENT PANEL (INCLUDING GJB2)
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COASY	100%	100%	100%	99.2%	Pontocerebellar hypoplasia, type 12, 618266;Neurodegeneration with brain iron accumulation 6, 615643	MOVEMENT DISORDERS PANEL INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS MITOCHONDRIAL DISORDERS PANEL COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹
COCH	100%	100%	100%	99.5%	Deafness, autosomal dominant 9, 601369;?Deafness, autosomal recessive 110, 618094	HEARING IMPAIRMENT PANEL (INCLUDING GJB2) MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹

COG1	100%	100%	100%	98.2%	Congenital disorder of glycosylation, type IIg, 611209	SHORT STATURE/SKELETAL DYSPLASIA PANEL WITH GENOME WIDE CNV ANALYSIS METABOLIC DISORDERS PANEL INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL¹
COG2	100%	100%	100%	99.3%	?Congenital disorder of glycosylation, type IIq, 617395	METABOLIC DISORDERS PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS

COG3	100%	100%	100%	99.5%	Congenital disorder of glycosylation, type IIbb, 620546	METABOLIC DISORDERS PANEL INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹
COG4	100%	100%	100%	99.1%	Congenital disorder of glycosylation, type IIj, 613489;Saul-Wilson syndrome, 618150	SHORT STATURE/SKELETAL DYSPLASIA PANEL WITH GENOME WIDE CNV ANALYSIS METABOLIC DISORDERS PANEL INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹

COG5	100%	100%	100%	99.6%	Congenital disorder of glycosylation, type Ili, 613612	METABOLIC DISORDERS PANEL INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹
COG6	100%	100%	100%	99.6%	Shaheen syndrome, 615328;Congenital disorder of glycosylation, type III, 614576	COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹ SKIN DISORDERS PANEL ¹ LIVER DISORDERS PANEL METABOLIC DISORDERS PANEL INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS

COG7	100%	100%	100%	98.9%	Congenital disorder of glycosylation, type IIe, 608779	LIVER DISORDERS PANEL METABOLIC DISORDERS PANEL INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹
COG8	100%	100%	100%	99%	Congenital disorder of glycosylation, type IIh, 611182	METABOLIC DISORDERS PANEL INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹

COL10A1	100%	100%	100%	99.5%	Metaphyseal chondrodysplasia, Schmid type, 156500	SHORT STATURE/SKELETAL DYSPLASIA PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS
COL11A1	100%	100%	100%	99.3%	Fibrochondrogenesis 1, 228520;Stickler syndrome, type II, 604841;Marshall syndrome, 154780;Deafness, autosomal dominant 37, 618533;{Lumbar disc herniation, susceptibility to}, 603932	VISION DISORDERS PANEL CRANIOFACIAL ANOMALIES PANEL WITH GENOME WIDE CNV ANALYSIS OROFACIAL CLEFTING PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹ SHORT STATURE/SKELETAL DYSPLASIA PANEL WITH GENOME WIDE CNV ANALYSIS HEARING IMPAIRMENT PANEL (INCLUDING GJB2)

COL11A2	100%	100%	100%	98.7%	Deafness, autosomal dominant 13, 601868;Otospondylomegaepiphyseal dysplasia, autosomal recessive, 215150;Fibrochondrogenesis 2, 614524;Deafness, autosomal recessive 53, 609706;Otospondylomegaepiphyseal dysplasia, autosomal dominant, 184840	MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹ OROFACIAL CLEFTING PANEL WITH GENOME WIDE CNV ANALYSIS HEARING IMPAIRMENT PANEL (INCLUDING GJB2) SHORT STATURE/SKELETAL DYSPLASIA PANEL WITH GENOME WIDE CNV ANALYSIS CRANIOFACIAL ANOMALIES PANEL WITH GENOME WIDE CNV ANALYSIS
COL12A1	100%	100%	100%	99.6%	Bethlem myopathy 2, 616471;?Ullrich congenital muscular dystrophy 2, 616470	FETAL AKINESIA PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹ MUSCLE DISORDERS PANEL

COL13A1	100%	100%	100%	99%	Myasthenic syndrome, congenital, 19, 616720	MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹ MUSCLE DISORDERS PANEL
COL14A1	100%	100%	100%	99.5%		SKIN DISORDERS PANEL ¹ MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS
COL17A1	100%	100%	100%	99%	Epithelial recurrent erosion dystrophy, 122400;Epidermolysis bullosa, junctional 4, intermediate, 619787	VISION DISORDERS PANEL COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹ SKIN DISORDERS PANEL ¹ MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS

COL18A1	100%	100%	100%	98.7%	Knobloch syndrome, type 1, 267750;Glaucoma, primary closed-angle, 618880	VISION DISORDERS PANEL INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹
COL1A1	100%	100%	100%	98.7%	Osteogenesis imperfecta, type II, 166210;Caffey disease, 114000;Ehlers-Danlos syndrome, arthrochalasia type, 1, 130060;Osteogenesis imperfecta, type I, 166200;{Bone mineral density variation QTL, osteoporosis}, 166710;Combined osteogenesis imperfecta and Ehlers-Danlos syndrome 1, 619115;Osteogenesis imperfecta, type IV, 166220;Osteogenesis imperfecta, type III, 259420	THORACIC AORTIC ANEURYSM AND/OR DISSECTION PANEL HEMOSTATIC/THROMB OTIC DISORDERS PANEL SHORT STATURE/SKELETAL DYSPLASIA PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS

COL1A2	100%	100%	100%	99.3%	Osteogenesis imperfecta, type III, 259420;{Osteoporosis, postmenopausal}, 166710;Ehlers-Danlos syndrome, arthrochalasia type, 2, 617821;Combined osteogenesis imperfecta and Ehlers-Danlos syndrome 2, 619120;Ehlers-Danlos syndrome, cardiac valvular type, 225320;Osteogenesis imperfecta, type IV, 166220;Osteogenesis imperfecta, type II, 166210	THORACIC AORTIC ANEURYSM AND/OR DISSECTION PANEL SKIN DISORDERS PANEL ¹ SHORT STATURE/SKELETAL DYSPLASIA PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹
COL25A1	100%	100%	100%	99.3%	Fibrosis of extraocular muscles, congenital, 5, 616219	VISION DISORDERS PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹

COL27A1	100%	100%	100%	98.9%	Steel syndrome, 615155	SHORT STATURE/SKELETAL DYSPLASIA PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹
COL2A1	100%	100%	100%	99.2%	?Vitreoretinopathy with phalangeal epiphyseal dysplasia, 619248;Czech dysplasia, 609162;Achondrogenesis, type II or hypochondrogenesis, 200610;Spondyloperipheral dysplasia, 271700;SMED Strudwick type, 184250;?Epiphyseal dysplasia, multiple, with myopia and deafness, 132450;SED congenita, 183900;Kniest dysplasia, 156550;Stickler syndrome, type I, nonsyndromic ocular, 609508;Osteoarthritis with mild chondrodysplasia, 604864;Stickler syndrome, type I, 108300;Platyspondylic skeletal dysplasia, Torrance type, 151210;Spondyloepiphyseal dysplasia, Stanescu type, 616583;Avascular necrosis of the femoral head, 608805;Legg-Calve-Perthes disease, 150600	VISION DISORDERS PANEL OROFACIAL CLEFTING PANEL WITH GENOME WIDE CNV ANALYSIS SHORT STATURE/SKELETAL DYSPLASIA PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS HEARING IMPAIRMENT PANEL (INCLUDING GJB2) CRANIOFACIAL ANOMALIES PANEL WITH GENOME WIDE CNV ANALYSIS

COL3A1	100%	100%	100%	99.2%	Ehlers-Danlos syndrome, vascular type, 130050;Polymicrogyria with or without vascular-type EDS, 618343	HEART DISORDERS PANEL ¹ HEMOSTATIC/THROMB OTIC DISORDERS PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹ THORACIC AORTIC ANEURYSM AND/OR DISSECTION PANEL VISION DISORDERS PANEL CONGENITAL HEARTDISEASE PANEL ¹ SKIN DISORDERS PANEL ¹
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COL4A1	100%	100%	100%	99.1%	?Retinal arteries, tortuosity of, 180000;{Hemorrhage, intracerebral, susceptibility to}, 614519;Angiopathy, hereditary, with nephropathy, aneurysms, and muscle cramps, 611773;Microangiopathy and leukoencephalopathy, pontine, autosomal dominant, 618564;Brain small vessel disease with or without ocular anomalies, 175780	MOVEMENT DISORDERS PANEL VISION DISORDERS PANEL EPILEPSY PANEL INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS RENAL DISORDERS PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS
COL4A2	100%	100%	100%	98.6%	Brain small vessel disease 2, 614483;{Hemorrhage, intracerebral, susceptibility to}, 614519	MOVEMENT DISORDERS PANEL INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS

COL4A3	100%	100%	100%	99.1%	Alport syndrome 3A, autosomal dominant, 104200;Hematuria, benign familial, 2, 620320;Alport syndrome 3B, autosomal recessive, 620536	HEARING IMPAIRMENT PANEL (INCLUDING GJB2) RENAL DISORDERS PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹
COL4A4	99.7%	98.9%	100%	99%	Hematuria, familial benign, 1, 141200;Alport syndrome 2, autosomal recessive, 203780	COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹ MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS HEARING IMPAIRMENT PANEL (INCLUDING GJB2) RENAL DISORDERS PANEL

COL4A5	100%	99.8%	98.9%	67.4%	Alport syndrome 1, X-linked, 301050	HEARING IMPAIRMENT PANEL (INCLUDING GJB2) RENAL DISORDERS PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS
COL4A6	100%	99.4%	98.5%	68.3%	?Deafness, X-linked 6, 300914	HEARING IMPAIRMENT PANEL (INCLUDING GJB2) MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS
COL5A1	100%	100%	100%	99%	Ehlers-Danlos syndrome, classic type, 1, 130000;Fibromuscular dysplasia, multifocal, 619329	THORACIC AORTIC ANEURYSM AND/OR DISSECTION PANEL VISION DISORDERS PANEL SKIN DISORDERS PANEL ¹ HEMOSTATIC/THROMB OTIC DISORDERS PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS

COL5A2	100%	100%	100%	99.4%	Ehlers-Danlos syndrome, classic type, 2, 130010	THORACIC AORTIC ANEURYSM AND/OR DISSECTION PANEL SKIN DISORDERS PANEL ¹ HEMOSTATIC/THROMBOTIC DISORDERS PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS
COL6A1	100%	100%	100%	98.8%	Ullrich congenital muscular dystrophy 1A, 254090;Bethlem myopathy 1A, 158810	FETAL AKINESIA PANEL MOVEMENT DISORDERS PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹ MUSCLE DISORDERS PANEL

COL6A2	100%	100%	100%	98.9%	?Myosclerosis, congenital, 255600;Ullrich congenital muscular dystrophy 1B, 620727;Bethlem myopathy 1B, 620725	FETAL AKINESIA PANEL MOVEMENT DISORDERS PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹ MUSCLE DISORDERS PANEL
COL6A3	100%	100%	100%	99.4%	Bethlem myopathy 1C, 620726;Ullrich congenital muscular dystrophy 1C, 620728;Dystonia 27, 616411	FETAL AKINESIA PANEL MOVEMENT DISORDERS PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹ MUSCLE DISORDERS PANEL

COL6A5	100%	100%	100%	99.6%		POLYNEUROPATHIES PANEL ¹ NEUROLOGICAL PAIN DISORDERS PANEL ¹ MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS
COL7A1	100%	100%	100%	98.7%	Nail disorder, nonsyndromic congenital, 8, 607523;Epidermolysis bullosa dystrophica, Bart type, 132000;Epidermolysis bullosa dystrophica inversa, 226600;Epidermolysis bullosa dystrophica, autosomal recessive, 226600;Epidermolysis bullosa, pretibial, 131850;Epidermolysis bullosa dystrophica, autosomal dominant, 131750;Transient bullous of the newborn, 131705;Epidermolysis bullosa pruriginosa, 604129;Epidermolysis bullosa dystrophica, localisata variant, 226600	SKIN DISORDERS PANEL ¹ MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹
COL8A2	100%	100%	100%	95.4%	Corneal dystrophy, posterior polymorphous 2, 609140;Corneal dystrophy, Fuchs endothelial, 1, 136800	VISION DISORDERS PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS

COL9A1	100%	100%	100%	99.6%	Stickler syndrome, type IV, 614134;?Epiphyseal dysplasia, multiple, 6, 614135	VISION DISORDERS PANEL CRANIOFACIAL ANOMALIES PANEL WITH GENOME WIDE CNV ANALYSIS HEARING IMPAIRMENT PANEL (INCLUDING GJB2) SHORT STATURE/SKELETAL DYSPLASIA PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹ OROFACIAL CLEFTING PANEL WITH GENOME WIDE CNV ANALYSIS
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COL9A2	100%	100%	100%	98.8%	Epiphyseal dysplasia, multiple, 2, 600204;?Stickler syndrome, type V, 614284	MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹ HEARING IMPAIRMENT PANEL (INCLUDING GJB2) SHORT STATURE/SKELETAL DYSPLASIA PANEL WITH GENOME WIDE CNV ANALYSIS VISION DISORDERS PANEL CRANIOFACIAL ANOMALIES PANEL WITH GENOME WIDE CNV ANALYSIS
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COL9A3	100%	99.9%	100%	98.2%	{Intervertebral disc disease, susceptibility to}, 603932;Epiphyseal dysplasia, multiple, 3, with or without myopathy, 600969;Stickler syndrome, type VI, 620022	MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹ HEARING IMPAIRMENT PANEL (INCLUDING GJB2) SHORT STATURE/SKELETAL DYSPLASIA PANEL WITH GENOME WIDE CNV ANALYSIS CRANIOFACIAL ANOMALIES PANEL WITH GENOME WIDE CNV ANALYSIS VISION DISORDERS PANEL
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COLEC10	100%	100%	100%	99.7%	3MC syndrome 3, 248340	OROFACIAL CLEFTING PANEL WITH GENOME WIDE CNV ANALYSIS SHORT STATURE/SKELETAL DYSPLASIA PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹
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COLEC11	100%	100%	100%	99%	3MC syndrome 2, 265050	CRANIOFACIAL ANOMALIES PANEL WITH GENOME WIDE CNV ANALYSIS PRIMARY IMMUNODEFICIENCIES PANEL SHORT STATURE/SKELETAL DYSPLASIA PANEL WITH GENOME WIDE CNV ANALYSIS INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹ OROFACIAL CLEFTING PANEL WITH GENOME WIDE CNV ANALYSIS
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COLGALT1	100%	99.9%	100%	97.8%	Brain small vessel disease 3, 618360	EPILEPSY PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹
COLQ	100%	100%	100%	99.3%	Myasthenic syndrome, congenital, 5, 603034	MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹ MUSCLE DISORDERS PANEL
COMP	100%	100%	100%	97.4%	Pseudoachondroplasia, 177170;Carpal tunnel syndrome 2, 619161;Epiphyseal dysplasia, multiple, 1, 132400	POLYNEUROPATHIES PANEL ¹ NEUROLOGICAL PAIN DISORDERS PANEL ¹ SHORT STATURE/SKELETAL DYSPLASIA PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS

COMT	92.7%	91.9%	100%	99.1%	{Schizophrenia, susceptibility to}, 181500;{Panic disorder, susceptibility to}, 167870	METABOLIC DISORDERS PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS
COPA	100%	100%	100%	99.2%	{Autoinflammation and autoimmunity, systemic, with immune dysregulation}, 616414	PRIMARY IMMUNODEFICIENCIES PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS
COPB1	100%	100%	100%	99.5%	Baralle-Macken syndrome, 619255	INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS

COPB2	100%	100%	100%	99.4%	Osteoporosis, childhood- or juvenile-onset, with developmental delay, 619884;?Microcephaly 19, primary, autosomal recessive, 617800	INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL¹
COPG1	100%	100%	100%	99.1%	?Immunodeficiency 128, 620983	PRIMARY IMMUNODEFICIENCIES PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS
COPZ1	100%	100%	100%	99.6%		INHERITED BONE MARROW FAILURE AND/OR PREDISPOSITION PANEL TO HEMATOLOGICAL MALIGNANCIES PRIMARY IMMUNODEFICIENCIES PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS

COQ2	96.3%	96.3%	100%	99.4%	{Multiple system atrophy, susceptibility to}, 146500;Coenzyme Q10 deficiency, primary, 1, 607426	MOVEMENT DISORDERS PANEL VISION DISORDERS PANEL EPILEPSY PANEL HEART DISORDERS PANEL ¹ METABOLIC DISORDERS PANEL INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS RENAL DISORDERS PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS MITOCHONDRIAL DISORDERS PANEL COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹
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COQ4	100%	100%	100%	97.8%	Coenzyme Q10 deficiency, primary, 7, 616276;Spastic ataxia 10, autosomal recessive, 620666	MOVEMENT DISORDERS PANEL EPILEPSY PANEL METABOLIC DISORDERS PANEL INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS MITOCHONDRIAL DISORDERS PANEL COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL¹
COQ5	100%	100%	100%	99.5%	?Coenzyme Q10 deficiency, primary, 9, 619028	METABOLIC DISORDERS PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS MITOCHONDRIAL DISORDERS PANEL

COQ6	100%	100%	100%	99.7%	Coenzyme Q10 deficiency, primary, 6, 614650	NEUROLOGICAL PAIN DISORDERS PANEL¹ METABOLIC DISORDERS PANEL RENAL DISORDERS PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS MITOCHONDRIAL DISORDERS PANEL COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL¹
COQ7	100%	100%	100%	99%	Coenzyme Q10 deficiency, primary, 8, 616733;Neuronopathy, distal hereditary motor, autosomal recessive 9, 620402	HEART DISORDERS PANEL¹ METABOLIC DISORDERS PANEL RENAL DISORDERS PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS MITOCHONDRIAL DISORDERS PANEL COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL¹

COQ8A	100%	100%	100%	98.8%	Coenzyme Q10 deficiency, primary, 4, 612016	MOVEMENT DISORDERS PANEL EPILEPSY PANEL METABOLIC DISORDERS PANEL INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS MITOCHONDRIAL DISORDERS PANEL COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹
COQ8B	100%	100%	100%	98.9%	Nephrotic syndrome, type 9, 615573	VISION DISORDERS PANEL METABOLIC DISORDERS PANEL RENAL DISORDERS PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS MITOCHONDRIAL DISORDERS PANEL COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹

COQ9	100%	100%	100%	98.9%	Coenzyme Q10 deficiency, primary, 5, 614654	MOVEMENT DISORDERS PANEL METABOLIC DISORDERS PANEL INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS RENAL DISORDERS PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS MITOCHONDRIAL DISORDERS PANEL COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹
CORIN	100%	100%	100%	99.5%	?Cardiomyopathy, familial hypertrophic, 30, atrial, 620734;Preeclampsia/eclampsia 5, 614595	MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS

CORO1A	100%	100%	100%	99.4%	Immunodeficiency 8, 615401	PRIMARY IMMUNODEFICIENCIES PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL¹ SEVERE COMBINED IMMUNODEFICIENCY (SCID) PANEL
COX10	100%	100%	100%	99.5%	Mitochondrial complex IV deficiency, nuclear type 3, 619046	INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS MITOCHONDRIAL DISORDERS PANEL COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL¹

COX11	100%	100%	100%	97.2%	Mitochondrial complex IV deficiency, nuclear type 23, 620275	INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS MITOCHONDRIAL DISORDERS PANEL COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL¹
COX14	100%	100%	100%	98.1%	?Mitochondrial complex IV deficiency, nuclear type 10, 619053	MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS MITOCHONDRIAL DISORDERS PANEL COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL¹

COX15	100%	100%	100%	99.1%	Mitochondrial complex IV deficiency, nuclear type 6, 615119	FETAL AKINESIA PANEL HEART DISORDERS PANEL ¹ INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS MITOCHONDRIAL DISORDERS PANEL COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹
COX16	100%	100%	100%	99.4%	Mitochondrial complex IV deficiency, nuclear type 22, 619355	INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS MITOCHONDRIAL DISORDERS PANEL COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹

COX18	100%	100%	100%	98.1%		POLYNEUROPATHIES PANEL ¹ MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS MITOCHONDRIAL DISORDERS PANEL
COX20	100%	100%	100%	99.7%	Mitochondrial complex IV deficiency, nuclear type 11, 619054	MOVEMENT DISORDERS PANEL POLYNEUROPATHIES PANEL ¹ MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS MITOCHONDRIAL DISORDERS PANEL COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹
COX411	100%	100%	100%	98.9%	Mitochondrial complex IV deficiency, nuclear type 16, 619060	MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS MITOCHONDRIAL DISORDERS PANEL

COX4I2	100%	100%	100%	99%	Exocrine pancreatic insufficiency, dyserythropoietic anemia, and calvarial hyperostosis, 612714	INHERITED BONE MARROW FAILURE AND/OR PREDISPOSITION PANEL TO HEMATOLOGICAL MALIGNANCIES SKIN DISORDERS PANEL ¹ MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS MITOCHONDRIAL DISORDERS PANEL COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹
COX5A	100%	100%	100%	99.5%	Mitochondrial complex IV deficiency, nuclear type 20, 619064	MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS MITOCHONDRIAL DISORDERS PANEL COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹

COX5B	100%	100%	100%	98.7%		MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS MITOCHONDRIAL DISORDERS PANEL
COX6A1	100%	100%	100%	99.9%	Charcot-Marie-Tooth disease, recessive intermediate D, 616039	POLYNEUROPATHIES PANEL ¹ MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS MITOCHONDRIAL DISORDERS PANEL COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹
COX6A2	100%	99.7%	100%	88%	Mitochondrial complex IV deficiency, nuclear type 18, 619062	MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS MITOCHONDRIAL DISORDERS PANEL COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹ MUSCLE DISORDERS PANEL

COX6B1	100%	100%	100%	99.8%	Mitochondrial complex IV deficiency, nuclear type 7, 619051	INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS MITOCHONDRIAL DISORDERS PANEL COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹
COX6B2	100%	100%	100%	99.3%		MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS MITOCHONDRIAL DISORDERS PANEL
COX6C	100%	100%	100%	99.5%		MITOCHONDRIAL DISORDERS PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS

COX7A1	100%	100%	100%	99.7%		MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS MITOCHONDRIAL DISORDERS PANEL
COX7A2	100%	100%	100%	99.2%		MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS MITOCHONDRIAL DISORDERS PANEL
COX7B	100%	100%	98.4%	62.3%	Linear skin defects with multiple congenital anomalies 2, 300887	VISION DISORDERS PANEL SKIN DISORDERS PANEL ¹ MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS MITOCHONDRIAL DISORDERS PANEL
COX7B2	100%	100%	100%	100%		MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS MITOCHONDRIAL DISORDERS PANEL

COX7C	100%	100%	100%	99.5%		MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS MITOCHONDRIAL DISORDERS PANEL
COX8A	100%	100%	100%	98.9%	?Mitochondrial complex IV deficiency, nuclear type 15, 619059	MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS MITOCHONDRIAL DISORDERS PANEL COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹
COX8C	100%	100%	100%	99.8%		MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS MITOCHONDRIAL DISORDERS PANEL

CP	100%	100%	100%	99.5%	Aceruloplasminemia, 604290	IRON DISORDERS PANEL MOVEMENT DISORDERS PANEL METABOLIC DISORDERS PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS MITOCHONDRIAL DISORDERS PANEL COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹
CPA6	100%	100%	100%	99.3%	Febrile seizures, familial, 11, 614418;Epilepsy, familial temporal lobe, 5, 614417	EPILEPSY PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹
CPAMD8	100%	100%	100%	98.3%	Anterior segment dysgenesis 8, 617319	VISION DISORDERS PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹

CPE	100%	100%	100%	99.1%	BDV syndrome, 619326	INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹
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CPLANE1	100%	100%	100%	99%	Orofaciodigital syndrome VI, 277170;Joubert syndrome 17, 614615	VISION DISORDERS PANEL CILIOPATHIES PANEL RENAL DISORDERS PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹ OROFACIAL CLEFTING PANEL WITH GENOME WIDE CNV ANALYSIS INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS SHORT STATURE/SKELETAL DYSPLASIA PANEL WITH GENOME WIDE CNV ANALYSIS
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CPLX1	100%	100%	100%	99%	Developmental and epileptic encephalopathy 63, 617976	EPILEPSY PANEL INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹
CPN1	100%	100%	100%	98.7%	Carboxypeptidase N deficiency, 212070	MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹
CPOX	100%	100%	100%	98.6%	Coproporphyrria, 121300;Harderoporphyria, 618892	MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹ SKIN DISORDERS PANEL ¹ METABOLIC DISORDERS PANEL

CPS1	100%	100%	100%	99.6%	Carbamoylphosphate synthetase I deficiency, 237300;{Pulmonary hypertension, neonatal, susceptibility to}, 615371	EPILEPSY PANEL METABOLIC DISORDERS PANEL INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹
CPSF1	100%	100%	100%	97.9%	Myopia 27, 618827	VISION DISORDERS PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS
CPSF3	100%	100%	100%	99.4%	Neurodevelopmental disorder with microcephaly, hypotonia, nystagmus, and seizures, 619876	INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS

CPT1A	100%	100%	100%	99.4%	CPT deficiency, hepatic, type IA, 255120	HEART DISORDERS PANEL ¹ METABOLIC DISORDERS PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹
CPT1C	100%	99.9%	100%	97.1%	?Spastic paraplegia 73, autosomal dominant, 616282	MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS
CPT2	100%	100%	100%	98.8%	{Encephalopathy, acute, infection-induced, 4, susceptibility to}, 614212;CPT II deficiency, infantile, 600649;CPT II deficiency, lethal neonatal, 608836;CPT II deficiency, myopathic, stress-induced, 255110	EPILEPSY PANEL HEART DISORDERS PANEL ¹ METABOLIC DISORDERS PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹ MUSCLE DISORDERS PANEL

CR2	100%	100%	100%	99.4%	{Systemic lupus erythematosus, susceptibility to, 9}, 610927;?Immunodeficiency, common variable, 7, 614699	PRIMARY IMMUNODEFICIENCIES PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹
CRACR2A	100%	100%	100%	99.1%		PRIMARY IMMUNODEFICIENCIES PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS
CRADD	100%	100%	100%	99.4%	Intellectual developmental disorder, autosomal recessive 34, with variant lissencephaly, 614499	INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹

CRAT	100%	100%	100%	99.3%	?Neurodegeneration with brain iron accumulation 8, 617917	METABOLIC DISORDERS PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS MITOCHONDRIAL DISORDERS PANEL COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹
CRB1	98.6%	98.6%	100%	99.7%	Leber congenital amaurosis 8, 613835;Retinitis pigmentosa-12, 600105;Pigmented paravenous chorioretinal atrophy, 172870	VISION DISORDERS PANEL COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹ MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS
CRB2	100%	100%	100%	98.7%	Focal segmental glomerulosclerosis 9, 616220;Ventriculomegaly with cystic kidney disease, 219730	RENAL DISORDERS PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹

CRBN	100%	99.2%	100%	99.3%	Intellectual developmental disorder, autosomal recessive 2, 607417	INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹
CREB1	100%	100%	100%	99.5%	Histiocytoma, angiomatoid fibrous, somatic, 612160	MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS
CREB3L1	100%	99.9%	100%	98.6%	Osteogenesis imperfecta, type XVI, 616229	SHORT STATURE/SKELETAL DYSPLASIA PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹

CREB3L3	100%	100%	100%	99%	Hypertriglyceridemia 2, 619324	MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS
CREBBP	100%	100%	100%	98.6%	Menke-Hennekam syndrome 1, 618332;Rubinstein-Taybi syndrome 1, 180849	VISION DISORDERS PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS HEREDITARY CANCER PANEL DISORDERS/DIFFEREN CES OF SEX DEVELOPMENT (DSD) / PRIMARY ADRENAL INSUFFICIENCY PANEL PRIMARY IMMUNODEFICIENCIES PANEL SHORT STATURE/SKELETAL DYSPLASIA PANEL WITH GENOME WIDE CNV ANALYSIS INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS

CRELD1	100%	100%	100%	98.8%	Atrioventricular septal defect, partial, with heterotaxy syndrome, 606217;Jeffries-Lakhani neurodevelopmental syndrome, 620771;{Atrioventricular septal defect, susceptibility to, 2}, 606217	MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS CONGENITAL HEARTDISEASE PANEL ¹ HEART DISORDERS PANEL ¹ INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS
CRIP1	100%	100%	100%	99.6%	Rothmund-Thomson syndrome, type 3, 615789	SKIN DISORDERS PANEL ¹ SHORT STATURE/SKELETAL DYSPLASIA PANEL WITH GENOME WIDE CNV ANALYSIS INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹

CRLF1	99.3%	97.4%	99.1%	89.7%	Cold-induced sweating syndrome 1, 272430	COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹ INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS
CRLS1	100%	100%	100%	99%	Combined oxidative phosphorylation deficiency 57, 620167	COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹ MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS MITOCHONDRIAL DISORDERS PANEL INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS METABOLIC DISORDERS PANEL

CRPPA	100%	100%	100%	99.6%	Muscular dystrophy-dystroglycanopathy (limb-girdle), type C, 7, 616052; Muscular dystrophy-dystroglycanopathy (congenital with brain and eye anomalies), type A, 7, 614643	FETAL AKINESIA PANEL METABOLIC DISORDERS PANEL INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MUSCLE DISORDERS PANEL COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹ MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS
CRTAP	100%	100%	100%	98.8%	Osteogenesis imperfecta, type VII, 610682	SHORT STATURE/SKELETAL DYSPLASIA PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹ MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS

CRTC1	100%	99.9%	100%	97.7%	Mucoepidermoid salivary gland carcinoma	MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS
CRX	100%	100%	100%	97.6%	Leber congenital amaurosis 7, 613829;Cone-rod retinal dystrophy-2, 120970	VISION DISORDERS PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS
CRYAA	100%	100%	100%	98.7%	Cataract 9, multiple types, 604219	VISION DISORDERS PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹

CRYAB	100%	100%	100%	99.6%	Myopathy, myofibrillar, fatal infantile hypertonic, alpha-B crystallin-related, 613869;Myopathy, myofibrillar, 2, 608810;Cataract 16, multiple types, 613763;Cardiomyopathy, dilated, 1II, 615184	VISION DISORDERS PANEL HEART DISORDERS PANEL ¹ MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹ MUSCLE DISORDERS PANEL
CRYBA1	100%	100%	100%	99%	Cataract 10, multiple types, 600881	VISION DISORDERS PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS
CRYBA2	100%	100%	100%	97.2%	?Cataract 42, 115900	VISION DISORDERS PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS

CRYBA4	100%	100%	100%	99.2%	Cataract 23, 610425	VISION DISORDERS PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS
CRYBB1	100%	100%	100%	98.6%	Cataract 17, multiple types, 611544	VISION DISORDERS PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹
CRYBB2	100%	100%	100%	99.3%	Cataract 3, multiple types, 601547	VISION DISORDERS PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS
CRYBB3	100%	100%	100%	98.4%	Cataract 22, 609741	VISION DISORDERS PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹

CRYGB	100%	100%	100%	98.1%	Cataract 39, multiple types, autosomal dominant, 615188	VISION DISORDERS PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS
CRYGC	100%	100%	100%	99%	Cataract 2, multiple types, 604307	VISION DISORDERS PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS
CRYGD	100%	100%	100%	99.6%	Cataract 4, multiple types, 115700	VISION DISORDERS PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS
CRYGS	100%	100%	100%	99.6%	Cataract 20, multiple types, 116100	VISION DISORDERS PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS

CRYL1	100%	100%	99.9%	95.7%		HEARING IMPAIRMENT PANEL (INCLUDING GJB2) MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS
CRYM	100%	100%	100%	98%	Deafness, autosomal dominant 40, 616357	HEARING IMPAIRMENT PANEL (INCLUDING GJB2) MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS
CSDE1	98%	97.8%	100%	99.4%		INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS

CSF1R	100%	100%	100%	98.9%	Brain abnormalities, neurodegeneration, and dysosteosclerosis, 618476;Leukoencephalopathy, diffuse hereditary, with spheroids 1, 221820	MOVEMENT DISORDERS PANEL SHORT STATURE/SKELETAL DYSPLASIA PANEL WITH GENOME WIDE CNV ANALYSIS INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS PARKINSON DISEASE PANEL COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹
CSF2RA	48.9%	48.3%	50%	49.5%	Surfactant metabolism dysfunction, pulmonary, 4, 300770	PRIMARY IMMUNODEFICIENCIES PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS

CSF2RB	100%	100%	100%	99%	Surfactant metabolism dysfunction, pulmonary, 5, 614370	PRIMARY IMMUNODEFICIENCIES PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹
CSF3R	100%	100%	100%	99%	Neutropenia, severe congenital, 7, autosomal recessive, 617014;?Neutrophilia, hereditary, 162830	INHERITED BONE MARROW FAILURE AND/OR PREDISPOSITION PANEL TO HEMATOLOGICAL MALIGNANCIES PRIMARY IMMUNODEFICIENCIES PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹

CSGALNAC T1	100%	100%	100%	99.2%	Skeletal dysplasia, mild, with joint laxity and advanced bone age, 618870	SHORT STATURE/SKELETAL DYSPLASIA PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹
CSMD1	100%	100%	100%	99.1%		INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS
CSNK1D	100%	100%	100%	98.8%	Advanced sleep-phase syndrome, familial, 2, 615224	MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS

CSNK1G1	100%	100%	100%	99.2%		INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS
CSNK2A1	94.2%	94.2%	100%	99.7%	Okur-Chung neurodevelopmental syndrome, 617062	INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS
CSNK2B	100%	100%	100%	99.1%	Poirier-Bienvenu neurodevelopmental syndrome, 618732	EPILEPSY PANEL INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS

CSPP1	96.9%	96.9%	100%	99%	Joubert syndrome 21, 615636	VISION DISORDERS PANEL CILIOPATHIES PANEL SHORT STATURE/SKELETAL DYSPLASIA PANEL WITH GENOME WIDE CNV ANALYSIS INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS RENAL DISORDERS PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹
CSR3	100%	100%	100%	99.8%	?Cardiomyopathy, dilated, 1M, 607482;Cardiomyopathy, hypertrophic, 12, 612124	HEART DISORDERS PANEL ¹ HYPERTROPHIC CARDIOMYOPATHY PANEL ¹ MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS

CST3	100%	100%	100%	97.1%	{Macular degeneration, age-related, 11}, 611953;Cerebral amyloid angiopathy, 105150	MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS
CST6	100%	100%	100%	97%	?Ectodermal dysplasia 15, hypohidrotic/hair type, 618535	SKIN DISORDERS PANEL ¹ MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS
CSTA	100%	100%	100%	99.4%	Peeling skin syndrome 4, 607936	SKIN DISORDERS PANEL ¹ MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹

CSTB	100%	100%	100%	98.6%	Epilepsy, progressive myoclonic 1A (Unverricht and Lundborg), 254800	MOVEMENT DISORDERS PANEL EPILEPSY PANEL INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹
CSTF2	100%	99.7%	99%	70%	?Intellectual developmental disorder, X-linked 113, 301116	MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS
CT55	100%	99.8%	99%	73.7%	?Spermatogenic failure, X-linked, 7, 301106	MALE INFERTILITY PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS

CTBP1	99.3%	97.8%	99.9%	97.4%	Hypotonia, ataxia, developmental delay, and tooth enamel defect syndrome, 617915	MOVEMENT DISORDERS PANEL INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS MITOCHONDRIAL DISORDERS PANEL
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CTC1	100%	100%	100%	98.8%	Cerebroretinal microangiopathy with calcifications and cysts, 612199	MOVEMENT DISORDERS PANEL VISION DISORDERS PANEL INHERITED BONE MARROW FAILURE AND/OR PREDISPOSITION PANEL TO HEMATOLOGICAL MALIGNANCIES SKIN DISORDERS PANEL ¹ DYSKERATOSIS CONGENITA AND APLASTIC ANEMIA PANEL PRIMARY IMMUNODEFICIENCIES PANEL INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹ HEREDITARY CANCER PANEL
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CTCF	100%	100%	100%	98.5%	Intellectual developmental disorder, autosomal dominant 21, 615502	INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS OROFACIAL CLEFTING PANEL WITH GENOME WIDE CNV ANALYSIS
CTDP1	100%	100%	100%	99.3%	Congenital cataracts, facial dysmorphism, and neuropathy, 604168	VISION DISORDERS PANEL POLYNEUROPATHIES PANEL ¹ INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹
CTH	100%	100%	100%	99.6%	Cystathioninuria, 219500	METABOLIC DISORDERS PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS

CTHRC1	100%	100%	100%	99.3%	Barrett esophagus/esophageal adenocarcinoma, 614266	MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS
CTLA4	93.2%	93.2%	100%	99.3%	Immune dysregulation with autoimmunity, immunodeficiency, and lymphoproliferation, 616100;{Diabetes mellitus, insulin-dependent, 12}, 601388;{Celiac disease, susceptibility to, 3}, 609755;{Hashimoto thyroiditis}, 140300;{Systemic lupus erythematosus, susceptibility to}, 152700	INHERITED BONE MARROW FAILURE AND/OR PREDISPOSITION PANEL TO HEMATOLOGICAL MALIGNANCIES HEMOSTATIC/THROMB OTIC DISORDERS PANEL PRIMARY IMMUNODEFICIENCIES PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS HEREDITARY CANCER PANEL

CTNNA1	100%	100%	100%	99.3%	Macular dystrophy, patterned, 2, 608970	VISION DISORDERS PANEL PANEL HEREDITARY CANCER (CDH1, CTNNA1, MLH1, MSH2, MSH6, PMS2) MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS HEREDITARY CANCER PANEL
CTNNA2	100%	100%	100%	99%	Cortical dysplasia, complex, with other brain malformations 9, 618174	INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹
CTNNA3	100%	100%	100%	99.5%	Arrhythmogenic right ventricular dysplasia 13, 615616	HEART DISORDERS PANEL ¹ MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS

CTNNB1	100%	100%	100%	99.6%	Exudative vitreoretinopathy 7, 617572;Pilomatricoma, somatic, 132600;Colorectal cancer, somatic, 114500;Neurodevelopmental disorder with spastic diplegia and visual defects, 615075;Medulloblastoma, somatic, 155255;Ovarian cancer, somatic, 167000;Hepatocellular carcinoma, somatic, 114550	VISION DISORDERS PANEL INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS
CTNNBL1	100%	100%	100%	99.6%	?Immunodeficiency 99 with hypogammaglobulinemia and autoimmune cytopenias, 619846	PRIMARY IMMUNODEFICIENCIES PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS
CTNND1	100%	100%	100%	99.3%	Blepharocheilodontic syndrome 2, 617681	CONGENITAL HEARTDISEASE PANEL ¹ HEART DISORDERS PANEL ¹ INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS OROFACIAL CLEFTING PANEL WITH GENOME WIDE CNV ANALYSIS

CTNND2	99.8%	98.6%	100%	97.7%		INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS
CTNS	100%	100%	100%	96.1%	Cystinosis, nephropathic, 219800;Cystinosis, ocular nonnephropathic, 219750;Cystinosis, late-onset juvenile or adolescent nephropathic, 219900;Cystinosis, atypical nephropathic, 219800	METABOLIC DISORDERS PANEL RENAL DISORDERS PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹
CTPS1	100%	100%	100%	99.5%	Immunodeficiency 24, 615897	PRIMARY IMMUNODEFICIENCIES PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹

CTR9	100%	100%	100%	99.6%		INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS HEREDITARY CANCER PANEL
CTSA	100%	99.9%	100%	99.5%	Galactosialidosis, 256540	SKIN DISORDERS PANEL ¹ SHORT STATURE/SKELETAL DYSPLASIA PANEL WITH GENOME WIDE CNV ANALYSIS METABOLIC DISORDERS PANEL INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹

CTSB	87.1%	83.9%	100%	99%	Keratolytic winter erythema, 148370	SKIN DISORDERS PANEL ¹ MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS
CTSC	94.8%	94.5%	100%	99.3%	Periodontitis 1, juvenile, 170650;Haim-Munk syndrome, 245010;Papillon-Lefevre syndrome, 245000	SKIN DISORDERS PANEL ¹ PRIMARY IMMUNODEFICIENCIES PANEL METABOLIC DISORDERS PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹

CTSD	100%	100%	100%	98.5%	Ceroid lipofuscinosis, neuronal, 10, 610127	MOVEMENT DISORDERS PANEL VISION DISORDERS PANEL EPILEPSY PANEL METABOLIC DISORDERS PANEL INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL¹
CTSF	100%	100%	100%	99.1%	Ceroid lipofuscinosis, neuronal, 13 (Kufs type), 615362	MOVEMENT DISORDERS PANEL EPILEPSY PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL¹

CTSH	96.9%	94.5%	100%	99.1%		VISION DISORDERS PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS
CTSK	100%	100%	100%	99.6%	Pycnodysostosis, 265800	CRANIOFACIAL ANOMALIES PANEL WITH GENOME WIDE CNV ANALYSIS SHORT STATURE/SKELETAL DYSPLASIA PANEL WITH GENOME WIDE CNV ANALYSIS METABOLIC DISORDERS PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL¹
CTSZ	81.1%	72.9%	100%	99.1%		SKIN DISORDERS PANEL¹ MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS

CTTNBP2	100%	100%	100%	99.3%		INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS
CTU2	100%	100%	100%	97.9%	Microcephaly, facial dysmorphism, renal agenesis, and ambiguous genitalia syndrome, 618142	DISORDERS/DIFFEREN CES OF SEX DEVELOPMENT (DSD) / PRIMARY ADRENAL INSUFFICIENCY PANEL INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL¹

CUBN	100%	100%	100%	99.2%	[Proteinuria, chronic benign], 618884;Imerslund-Grasbeck syndrome 1, 261100	INHERITED BONE MARROW FAILURE AND/OR PREDISPOSITION PANEL TO HEMATOLOGICAL MALIGNANCIES METABOLIC DISORDERS PANEL RENAL DISORDERS PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹
CUL3	100%	100%	100%	99.8%	Neurodevelopmental disorder with or without autism or seizures, 619239;Pseudohypoaldosteronism, type IIE, 614496	INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS RENAL DISORDERS PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS

CUL4B	96.7%	96.7%	99.1%	69.8%	Intellectual developmental disorder, X-linked syndromic, Cabezas type, 300354	DISORDERS/DIFFERENCES OF SEX DEVELOPMENT (DSD) / PRIMARY ADRENAL INSUFFICIENCY PANEL EPILEPSY PANEL HYPOGONADOTROPIC HYPOGONADISM (KALLMANN) PANEL INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS
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CUL7	100%	100%	100%	98.8%	3-M syndrome 1, 273750	DISORDERS/DIFFERENCES OF SEX DEVELOPMENT (DSD) / PRIMARY ADRENAL INSUFFICIENCY PANEL SHORT STATURE/SKELETAL DYSPLASIA PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹
CUX1	100%	99.8%	100%	97.8%	Global developmental delay with or without impaired intellectual development, 618330	INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS

CUX2	100%	99.9%	100%	98.7%	Developmental and epileptic encephalopathy 67, 618141	MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS EPILEPSY PANEL INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS
CWC27	82.6%	82.6%	100%	99.7%	Retinitis pigmentosa with or without skeletal anomalies, 250410	VISION DISORDERS PANEL INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL¹

CWF19L1	100%	100%	100%	99.6%	Spinocerebellar ataxia, autosomal recessive 17, 616127	MOVEMENT DISORDERS PANEL INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL¹
CXCR2	100%	100%	100%	97.8%	?WHIM syndrome 2, 619407	INHERITED BONE MARROW FAILURE AND/OR PREDISPOSITION PANEL TO HEMATOLOGICAL MALIGNANCIES PRIMARY IMMUNODEFICIENCIES PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS

CXCR4	99%	99%	100%	99.3%	WHIM syndrome 1, 193670;Myelokathexis, isolated, 193670	INHERITED BONE MARROW FAILURE AND/OR PREDISPOSITION PANEL TO HEMATOLOGICAL MALIGNANCIES SKIN DISORDERS PANEL ¹ PRIMARY IMMUNODEFICIENCIES PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS
CYB561	100%	100%	100%	98%	Orthostatic hypotension 2, 618182	METABOLIC DISORDERS PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹

CYB5A	100%	100%	100%	99.8%	Methemoglobinemia and ambiguous genitalia, 250790	DISORDERS/DIFFERENCES OF SEX DEVELOPMENT (DSD) / PRIMARY ADRENAL INSUFFICIENCY PANEL METABOLIC DISORDERS PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹
CYB5R3	95.5%	93.6%	100%	99%	Methemoglobinemia, type I, 250800;Methemoglobinemia, type II, 250800	METABOLIC DISORDERS PANEL INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹

CYBA	70.8%	69.7%	100%	98.2%	Chronic granulomatous disease 4, autosomal recessive, 233690	PRIMARY IMMUNODEFICIENCIES PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹
CYBB	100%	100%	99%	68.7%	Immunodeficiency 34, mycobacteriosis, X-linked, 300645;Chronic granulomatous disease, X-linked, 306400	PRIMARY IMMUNODEFICIENCIES PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS
CYBC1	100%	100%	100%	99.3%	Chronic granulomatous disease 5, autosomal recessive, 618935	PRIMARY IMMUNODEFICIENCIES PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹

CYBRD1	100%	100%	100%	99.5%		IRON DISORDERS PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS
CYC1	100%	100%	100%	97.4%	Mitochondrial complex III deficiency, nuclear type 6, 615453	MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS MITOCHONDRIAL DISORDERS PANEL COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹
CYCS	100%	100%	100%	100%	Thrombocytopenia 4, 612004	INHERITED BONE MARROW FAILURE AND/OR PREDISPOSITION PANEL TO HEMATOLOGICAL MALIGNANCIES HEMOSTATIC/THROMB OTIC DISORDERS PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS MITOCHONDRIAL DISORDERS PANEL

CYFIP2	98.1%	98.1%	100%	99.3%	Developmental and epileptic encephalopathy 65, 618008	EPILEPSY PANEL INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS
CYLC1	100%	100%	98.4%	68.8%	{Spermatogenic failure, X-linked, 8, susceptibility to}, 301119	MALE INFERTILITY PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS
CYLD	100%	100%	100%	99.1%	Brooke-Spiegler syndrome, 605041;Cylindromatosis, familial, 132700;Trichoepithelioma, multiple familial, 1, 601606;?Frontotemporal dementia and/or amyotrophic lateral sclerosis 8, 619132	SKIN DISORDERS PANEL ¹ MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS HEREDITARY CANCER PANEL

CYP11A1	100%	100%	100%	99.3%	Adrenal insufficiency, congenital, with 46XY sex reversal, partial or complete, 613743	DISORDERS/DIFFERENCES OF SEX DEVELOPMENT (DSD) / PRIMARY ADRENAL INSUFFICIENCY PANEL METABOLIC DISORDERS PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL¹
CYP11B1	100%	100%	100%	99.6%	Aldosteronism, glucocorticoid-remediable, 103900;Adrenal hyperplasia, congenital, due to 11-beta-hydroxylase deficiency, 202010	DISORDERS/DIFFERENCES OF SEX DEVELOPMENT (DSD) / PRIMARY ADRENAL INSUFFICIENCY PANEL METABOLIC DISORDERS PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL¹

CYP11B2	100%	100%	100%	99.3%	Hypoaldosteronism, congenital, due to CMO I deficiency, 203400;Aldosterone to renin ratio raised;{Low renin hypertension, susceptibility to};Hypoaldosteronism, congenital, due to CMO II deficiency, 610600	DISORDERS/DIFFERENCES OF SEX DEVELOPMENT (DSD) / PRIMARY ADRENAL INSUFFICIENCY PANEL METABOLIC DISORDERS PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL¹
CYP17A1	100%	100%	100%	98.8%	17,20-lyase deficiency, isolated, 202110;17-alpha-hydroxylase/17,20-lyase deficiency, 202110	DISORDERS/DIFFERENCES OF SEX DEVELOPMENT (DSD) / PRIMARY ADRENAL INSUFFICIENCY PANEL METABOLIC DISORDERS PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL¹ PREMATURE OVARIAN INSUFFICIENCY PANEL

CYP19A1	100%	100%	100%	99.3%	Aromatase deficiency, 613546;Aromatase excess syndrome, 139300	DISORDERS/DIFFERENCES OF SEX DEVELOPMENT (DSD) / PRIMARY ADRENAL INSUFFICIENCY PANEL TALL STATURE PANEL WITH GENOME WIDE CNV ANALYSIS METABOLIC DISORDERS PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹ PREMATURE OVARIAN INSUFFICIENCY PANEL
CYP1B1	100%	100%	100%	99.3%	Glaucoma 3A, primary open angle, congenital, juvenile, or adult onset, 231300;Anterior segment dysgenesis 6, multiple subtypes, 617315	VISION DISORDERS PANEL METABOLIC DISORDERS PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹

CYP21A2	100%	100%	100%	99.7%	Hyperandrogenism, nonclassic type, due to 21-hydroxylase deficiency, 201910;Adrenal hyperplasia, congenital, due to 21-hydroxylase deficiency, 201910	DISORDERS/DIFFERENCES OF SEX DEVELOPMENT (DSD) / PRIMARY ADRENAL INSUFFICIENCY PANEL METABOLIC DISORDERS PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS
CYP24A1	100%	100%	100%	99.5%	Hypercalcemia, infantile, 1, 143880	RENAL DISORDERS PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL¹

CYP26B1	100%	100%	100%	98.9%	Craniosynostosis with radiohumeral fusions and other skeletal and craniofacial anomalies, 614416	CRANIOFACIAL ANOMALIES PANEL WITH GENOME WIDE CNV ANALYSIS SHORT STATURE/SKELETAL DYSPLASIA PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹
CYP26C1	100%	100%	100%	99%	Focal facial dermal dysplasia 4, 614974	SKIN DISORDERS PANEL ¹ MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹

CYP27A1	100%	100%	100%	98.6%	Cerebrotendinous xanthomatosis, 213700	MOVEMENT DISORDERS PANEL VISION DISORDERS PANEL POLYNEUROPATHIES PANEL ¹ LIVER DISORDERS PANEL METABOLIC DISORDERS PANEL INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹
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CYP27B1	100%	100%	100%	99.5%	Vitamin D-dependent rickets, type I, 264700	SHORT STATURE/SKELETAL DYSPLASIA PANEL WITH GENOME WIDE CNV ANALYSIS METABOLIC DISORDERS PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹
CYP2A6	99.8%	99.7%	100%	97.8%	{Lung cancer, resistance to}, 211980;Coumarin resistance, 122700;{Nicotine addiction, protection from}, 188890	MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS
CYP2B6	100%	100%	100%	97.9%	{Efavirenz central nervous system toxicity, susceptibility to}, 614546;Efavirenz, poor metabolism of, 614546	MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS
CYP2C19	100%	100%	100%	99.5%	Proguanil poor metabolizer, 609535;Mephenytoin poor metabolizer, 609535;Clopidogrel, impaired responsiveness to, 609535;Omeprazole poor metabolizer, 609535	MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS

CYP2C8	100%	100%	100%	99.6%	{Drug metabolism, altered, CYP2C8-related}, 618018	MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹
CYP2C9	100%	100%	100%	99.5%	Warfarin sensitivity, 122700;Tolbutamide poor metabolizer	MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS
CYP2R1	100%	100%	100%	99.5%	Rickets due to defect in vitamin D 25-hydroxylation deficiency, 600081	SHORT STATURE/SKELETAL DYSPLASIA PANEL WITH GENOME WIDE CNV ANALYSIS METABOLIC DISORDERS PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹

CYP2U1	100%	100%	100%	96.6%	Spastic paraplegia 56, autosomal recessive, 615030	MOVEMENT DISORDERS PANEL POLYNEUROPATHIES PANEL ¹ METABOLIC DISORDERS PANEL INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹
CYP3A4	100%	100%	100%	99.6%	Vitamin D-dependent rickets, type 3, 619073	MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS
CYP4F22	100%	100%	100%	98.7%	Ichthyosis, congenital, autosomal recessive 5, 604777	SKIN DISORDERS PANEL ¹ MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹

CYP4V2	100%	100%	100%	99.3%	Bietti crystalline corneoretinal dystrophy, 210370	VISION DISORDERS PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹
CYP7B1	100%	100%	100%	99.6%	Spastic paraplegia 5A, autosomal recessive, 270800;Bile acid synthesis defect, congenital, 3, 613812	MOVEMENT DISORDERS PANEL POLYNEUROPATHIES PANEL ¹ LIVER DISORDERS PANEL METABOLIC DISORDERS PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹

D2HGDH	100%	100%	100%	98.6%	D-2-hydroxyglutaric aciduria, 600721	EPILEPSY PANEL METABOLIC DISORDERS PANEL INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS MITOCHONDRIAL DISORDERS PANEL COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹
DAAM2	100%	99.9%	100%	99.2%	Nephrotic syndrome, type 24, 619263	RENAL DISORDERS PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS
DAB1	100%	100%	100%	99.1%	Spinocerebellar ataxia 37, 615945	MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS
DACT1	100%	100%	100%	98.4%	Townes-Brocks syndrome 2, 617466	MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS

DAG1	100%	100%	100%	99%	Muscular dystrophy-dystroglycanopathy (congenital with brain and eye anomalies), type A, 9, 616538;Muscular dystrophy-dystroglycanopathy (limb-girdle), type C, 9, 613818	INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹ MUSCLE DISORDERS PANEL
DAGLA	100%	100%	100%	99.2%	Neuroocular syndrome 2, paroxysmal type, 168885	INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS
DALRD3	100%	100%	100%	99.2%	?Developmental and epileptic encephalopathy 86, 618910	MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS

DAO	100%	100%	100%	98.5%		METABOLIC DISORDERS PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS
DAP3	100%	100%	100%	99.3%		HEARING IMPAIRMENT PANEL (INCLUDING GJB2) DISORDERS/DIFFEREN CES OF SEX DEVELOPMENT (DSD) / PRIMARY ADRENAL INSUFFICIENCY PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS MITOCHONDRIAL DISORDERS PANEL COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL¹ PREMATURE OVARIAN INSUFFICIENCY PANEL

DARS1	100%	100%	100%	99.7%	Hypomyelination with brainstem and spinal cord involvement and leg spasticity, 615281	MOVEMENT DISORDERS PANEL EPILEPSY PANEL INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL¹
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DARS2	100%	100%	100%	99.7%	Leukoencephalopathy with brain stem and spinal cord involvement and lactate elevation, 611105	MOVEMENT DISORDERS PANEL EPILEPSY PANEL POLYNEUROPATHIES PANEL ¹ INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS MITOCHONDRIAL DISORDERS PANEL COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹ MUSCLE DISORDERS PANEL
DAW1	100%	100%	100%	99.3%	Ciliary dyskinesia, primary, 52, 620570	MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹

DAZ1	50%	49.5%	48.2%	32.3%		MALE INFERTILITY PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS
DAZ2	49.8%	48.9%	47.2%	29.8%		MALE INFERTILITY PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS
DAZ3	49.8%	48.1%	45.1%	26.6%		MALE INFERTILITY PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS
DAZ4	49.5%	47.8%	46.1%	25.6%		MALE INFERTILITY PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS

DBF4	100%	100%	100%	98.5%		INHERITED BONE MARROW FAILURE AND/OR PREDISPOSITION PANEL TO HEMATOLOGICAL MALIGNANCIES PRIMARY IMMUNODEFICIENCIES PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS
DBH	100%	100%	100%	99.3%	Orthostatic hypotension 1, due to DBH deficiency, 223360	METABOLIC DISORDERS PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹
DBR1	100%	100%	100%	99.2%	Xerosis and growth failure with immune and pulmonary dysfunction syndrome, 620510;{Encephalitis, acute, infection (viral)-induced, susceptibility to, 11}, 619441	PRIMARY IMMUNODEFICIENCIES PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS

DBT	100%	100%	100%	99.7%	Maple syrup urine disease, type II, 620699	MOVEMENT DISORDERS PANEL METABOLIC DISORDERS PANEL INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹
DCAF12L1	100%	100%	98.1%	67%		MALE INFERTILITY PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS

DCAF17	100%	100%	100%	99.5%	Woodhouse-Sakati syndrome, 241080	MOVEMENT DISORDERS PANEL SKIN DISORDERS PANEL ¹ HEARING IMPAIRMENT PANEL (INCLUDING GJB2) DISORDERS/DIFFEREN CES OF SEX DEVELOPMENT (DSD) / PRIMARY ADRENAL INSUFFICIENCY PANEL HYPOGONADOTROPIC HYPOGONADISM (KALLMANN) PANEL INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS MITOCHONDRIAL DISORDERS PANEL COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹ PREMATURE OVARIAN INSUFFICIENCY PANEL
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DCAF8	100%	100%	100%	99.4%	?Giant axonal neuropathy 2, autosomal dominant, 610100	POLYNEUROPATHIES PANEL ¹ MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS
DCC	100%	100%	100%	98.7%	Mirror movements 1 and/or agenesis of the corpus callosum, 157600;Esophageal carcinoma, somatic, 133239;Colorectal cancer, somatic, 114500;Gaze palsy, familial horizontal, with progressive scoliosis, 2, 617542	MOVEMENT DISORDERS PANEL DISORDERS/DIFFEREN CES OF SEX DEVELOPMENT (DSD) / PRIMARY ADRENAL INSUFFICIENCY PANEL HYPOGONADOTROPIC HYPOGONADISM (KALLMANN) PANEL INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹

DCDC1	100%	100%	100%	99.4%		VISION DISORDERS PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS
DCDC2	100%	100%	100%	99.2%	Nephronophthisis 19, 616217;?Deafness, autosomal recessive 66, 610212;Sclerosing cholangitis, neonatal, 617394	CILIOPATHIES PANEL HEARING IMPAIRMENT PANEL (INCLUDING GJB2) LIVER DISORDERS PANEL RENAL DISORDERS PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL¹

DCHS1	100%	100%	100%	99.3%	Mitral valve prolapse 2, 607829;Van Maldergem syndrome 1, 601390	HEART DISORDERS PANEL ¹ INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹
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DCLRE1B	100%	100%	100%	99%	Dyskeratosis congenita, autosomal recessive 8, 620133	INHERITED BONE MARROW FAILURE AND/OR PREDISPOSITION PANEL TO HEMATOLOGICAL MALIGNANCIES DYSKERATOSIS CONGENITA AND APLASTIC ANEMIA PANEL PRIMARY IMMUNODEFICIENCIES PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹ HEREDITARY CANCER PANEL
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DCLRE1C	97.1%	97.1%	100%	99.4%	Severe combined immunodeficiency, Athabaskan type, 602450;Omenn syndrome, 603554	SKIN DISORDERS PANEL ¹ PRIMARY IMMUNODEFICIENCIES PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹ SEVERE COMBINED IMMUNODEFICIENCY (SCID) PANEL
DCN	95.1%	95.1%	100%	99.4%	Corneal dystrophy, congenital stromal, 610048	VISION DISORDERS PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS
DCPS	100%	100%	100%	99.3%	Al-Raqad syndrome, 616459	INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹

DCT	100%	100%	99.9%	97.4%	Oculocutaneous albinism, type VIII, 619165	VISION DISORDERS PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS
DCTN1	100%	100%	100%	98.7%	Perry syndrome, 168605;{Amyotrophic lateral sclerosis, susceptibility to}, 105400;Neuronopathy, distal hereditary motor, autosomal dominant 14, 607641	MOVEMENT DISORDERS PANEL POLYNEUROPATHIES PANEL ¹ MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS PARKINSON DISEASE PANEL
DCTN2	100%	100%	100%	99.6%		POLYNEUROPATHIES PANEL ¹ MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS

DCX	100%	100%	99.1%	71.6%	Subcortical laminar heterotopia, X-linked, 300067;Lissencephaly, X-linked, 300067	EPILEPSY PANEL INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS
DCXR	100%	100%	100%	99.7%	[Pentosuria], 260800	METABOLIC DISORDERS PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS
DDB1	100%	100%	100%	99.1%	White-Kernohan syndrome, 619426	INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS

DDB2	100%	100%	100%	98.8%	Xeroderma pigmentosum, group E, DDB-negative subtype, 278740	SKIN DISORDERS PANEL ¹ MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹ HEREDITARY CANCER PANEL
DDC	100%	100%	100%	99.4%	Aromatic L-amino acid decarboxylase deficiency, 608643	MOVEMENT DISORDERS PANEL METABOLIC DISORDERS PANEL INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹

DDHD1	100%	100%	100%	99%	Spastic paraplegia 28, autosomal recessive, 609340	MOVEMENT DISORDERS PANEL VISION DISORDERS PANEL METABOLIC DISORDERS PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS MITOCHONDRIAL DISORDERS PANEL COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹
DDHD2	100%	100%	100%	99.4%	Spastic paraplegia 54, autosomal recessive, 615033	MOVEMENT DISORDERS PANEL INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹

DDOST	100%	100%	100%	99%	Congenital disorder of glycosylation, type I _r , 614507	METABOLIC DISORDERS PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹
DDR1	100%	99.9%	100%	98.8%		SHORT STATURE/SKELETAL DYSPLASIA PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS
DDR2	100%	100%	100%	99.5%	Warburg-Cinotti syndrome, 618175;Spondylometaepiphyseal dysplasia, short limb-hand type, 271665	SHORT STATURE/SKELETAL DYSPLASIA PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹

DDRGK1	100%	100%	100%	98.6%	Spondyloepimetaphyseal dysplasia, Shohat type, 602557	SHORT STATURE/SKELETAL DYSPLASIA PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL¹
DDX11	100%	100%	100%	99.4%	Warsaw breakage syndrome, 613398	INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL¹ HEREDITARY CANCER PANEL

DDX17	100%	100%	100%	98.6%		INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS
DDX23	100%	100%	100%	99.4%		INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS
DDX39B	100%	100%	100%	98.1%		INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS

DDX3X	99.4%	98.6%	98.7%	68.6%	Intellectual developmental disorder, X-linked syndromic, Snijders Blok type, 300958	EPILEPSY PANEL INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS OROFACIAL CLEFTING PANEL WITH GENOME WIDE CNV ANALYSIS
DDX3Y	50%	49.9%	49%	20.1%		MALE INFERTILITY PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS

DDX41	100%	100%	100%	98.8%	{Myeloproliferative/lymphoproliferative neoplasms, familial (multiple types), susceptibility to}, 616871	INHERITED BONE MARROW FAILURE AND/OR PREDISPOSITION PANEL TO HEMATOLOGICAL MALIGNANCIES PRIMARY IMMUNODEFICIENCIES PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS HEREDITARY CANCER PANEL
DDX59	100%	100%	100%	99.3%	Orofaciodigital syndrome V, 174300	OROFACIAL CLEFTING PANEL WITH GENOME WIDE CNV ANALYSIS CILIOPATHIES PANEL INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹

DDX6	100%	100%	100%	99.4%	Intellectual developmental disorder with impaired language and dysmorphic facies, 618653	INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS
DEAF1	93.7%	91.9%	100%	96.5%	Vulto-van Silfout-de Vries syndrome, 615828;Neurodevelopmental disorder with hypotonia, impaired expressive language, and with or without seizures, 617171	INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹
DEF6	100%	100%	100%	98.1%	Immunodeficiency 87 and autoimmunity, 619573	PRIMARY IMMUNODEFICIENCIES PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS

DEGS1	100%	100%	100%	100%	Leukodystrophy, hypomyelinating, 18, 618404	MOVEMENT DISORDERS PANEL METABOLIC DISORDERS PANEL INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹
DENND5A	100%	100%	100%	99.4%	Developmental and epileptic encephalopathy 49, 617281	EPILEPSY PANEL INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹

DENND5B	95.9%	95.9%	100%	99.2%		INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS
DEPDC5	100%	100%	100%	99.2%	Epilepsy, familial focal, with variable foci 1, 604364;Developmental and epileptic encephalopathy 111, 620504	EPILEPSY PANEL INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS

DES	100%	100%	100%	97.2%	Scapuloperoneal syndrome, neurogenic, Kaeser type, 181400;Cardiomyopathy, dilated, 1l, 604765;Myopathy, myofibrillar, 1, 601419	ARITMOGENE CARDIOMYOPATHY PANEL ¹ COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹ ARRHYTHMIA AND CARDIAC CONDUCTION DISORDERS PANEL ¹ MUSCLE DISORDERS PANEL HYPERTROPHIC CARDIOMYOPATHY PANEL ¹ MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS MITOCHONDRIAL DISORDERS PANEL DILATED CARDIOMYOPATHY PANEL ¹ HEART DISORDERS PANEL ¹
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DGAT1	100%	100%	100%	99%	Diarrhea 7, protein-losing enteropathy type, 615863	METABOLIC DISORDERS PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹ LYMPHATIC ANOMALIES PANEL WITH GENOME WIDE CNV ANALYSIS
DGAT2	100%	100%	100%	99.3%		POLYNEUROPATHIES PANEL ¹ MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS
DGCR8	100%	100%	100%	99%		MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS HEREDITARY CANCER PANEL

DGKE	100%	100%	100%	99.8%	{Hemolytic uremic syndrome, atypical, susceptibility to, 7}, 615008;Nephrotic syndrome, type 7, 615008	HEMOSTATIC/THROMBOTIC DISORDERS PANEL COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹ MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS RENAL DISORDERS PANEL METABOLIC DISORDERS PANEL
DGUOK	100%	100%	100%	98.9%	Portal hypertension, noncirrhotic, 1, 617068;Progressive external ophthalmoplegia with mitochondrial DNA deletions, autosomal recessive 4, 617070;Mitochondrial DNA depletion syndrome 3 (hepatocerebral type), 251880	COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹ MUSCLE DISORDERS PANEL LIVER DISORDERS PANEL METABOLIC DISORDERS PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS MITOCHONDRIAL DISORDERS PANEL

DHCR24	100%	100%	100%	98.7%	Desmosterolosis, 602398	FETAL AKINESIA PANEL SHORT STATURE/SKELETAL DYSPLASIA PANEL WITH GENOME WIDE CNV ANALYSIS METABOLIC DISORDERS PANEL INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL¹
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DHCR7	96.2%	96.2%	100%	99.2%	Smith-Lemli-Opitz syndrome, 270400	FETAL AKINESIA PANEL SKIN DISORDERS PANEL ¹ DISORDERS/DIFFEREN CES OF SEX DEVELOPMENT (DSD) / PRIMARY ADRENAL INSUFFICIENCY PANEL LIVER DISORDERS PANEL METABOLIC DISORDERS PANEL INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹ OROFACIAL CLEFTING PANEL WITH GENOME WIDE CNV ANALYSIS
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DHDDS	73.7%	73.7%	100%	98.3%	Developmental delay and seizures with or without movement abnormalities, 617836;?Congenital disorder of glycosylation, type 1bb, 613861;Retinitis pigmentosa 59, 613861	MOVEMENT DISORDERS PANEL VISION DISORDERS PANEL INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹ METABOLIC DISORDERS PANEL EPILEPSY PANEL
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DHFR	100%	100%	100%	99.1%	Megaloblastic anemia due to dihydrofolate reductase deficiency, 613839	INHERITED BONE MARROW FAILURE AND/OR PREDISPOSITION PANEL TO HEMATOLOGICAL MALIGNANCIES PRIMARY IMMUNODEFICIENCIES PANEL METABOLIC DISORDERS PANEL INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹
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DHH	100%	100%	100%	99.5%	46XY gonadal dysgenesis with minifascicular neuropathy, 607080;46XY sex reversal 7, 233420	DISORDERS/DIFFERENCES OF SEX DEVELOPMENT (DSD) / PRIMARY ADRENAL INSUFFICIENCY PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹
DHODH	100%	100%	100%	98.6%	Miller syndrome, 263750	CRANIOFACIAL ANOMALIES PANEL WITH GENOME WIDE CNV ANALYSIS SHORT STATURE/SKELETAL DYSPLASIA PANEL WITH GENOME WIDE CNV ANALYSIS METABOLIC DISORDERS PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹ OROFACIAL CLEFTING PANEL WITH GENOME WIDE CNV ANALYSIS

DHPS	97.8%	93.3%	100%	97.4%	Neurodevelopmental disorder with seizures and speech and walking impairment, 618480	INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL¹
DHRX	50%	49.9%	50%	49.4%	Congenital disorder of glycosylation, type 1DD, 301133	METABOLIC DISORDERS PANEL INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS

DHTKD1	100%	100%	100%	99.3%	?Charcot-Marie-Tooth disease, axonal, type 2Q, 615025;Alpha-aminoadipic and alpha-ketoadipic aciduria, 204750	POLYNEUROPATHIES PANEL ¹ METABOLIC DISORDERS PANEL INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS MITOCHONDRIAL DISORDERS PANEL COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹
DHX16	100%	100%	100%	99.1%	Neuromuscular disease and ocular or auditory anomalies with or without seizures, 618733	HEARING IMPAIRMENT PANEL (INCLUDING GJB2) INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS

DHX30	100%	100%	100%	98.7%	Neurodevelopmental disorder with variable motor and speech impairment, 617804	INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS
DHX37	100%	100%	100%	99.1%	Neurodevelopmental disorder with brain anomalies and with or without vertebral or cardiac anomalies, 618731;46XY sex reversal 11, 273250	DISORDERS/DIFFEREN CES OF SEX DEVELOPMENT (DSD) / PRIMARY ADRENAL INSUFFICIENCY PANEL INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS
DHX38	100%	100%	100%	99.2%	Retinitis pigmentosa 84, 618220	VISION DISORDERS PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL¹

DHX9	100%	100%	100%	99.6%	Intellectual developmental disorder, autosomal dominant 75, 620988	INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS
DIABLO	100%	100%	100%	99.3%	Deafness, autosomal dominant 64, 614152	HEARING IMPAIRMENT PANEL (INCLUDING GJB2) MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS

DIAPH1	100%	100%	100%	98.8%	Deafness, autosomal dominant 1, with or without thrombocytopenia, 124900;Seizures, cortical blindness, microcephaly syndrome, 616632	HEARING IMPAIRMENT PANEL (INCLUDING GJB2) EPILEPSY PANEL HEMOSTATIC/THROMBOTIC DISORDERS PANEL PRIMARY IMMUNODEFICIENCIES PANEL INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹
DIAPH2	100%	99.8%	98.6%	70.9%	?Premature ovarian failure 2A, 300511	MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS
DIAPH3	100%	100%	100%	99.4%	Auditory neuropathy, autosomal dominant 1, 609129	HEARING IMPAIRMENT PANEL (INCLUDING GJB2) MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS

DICER1	100%	100%	100%	99.3%	Pleuropulmonary blastoma, 601200;Goiter, multinodular 1, with or without Sertoli-Leydig cell tumors, 138800;GLOW syndrome, somatic mosaic, 618272;Rhabdomyosarcoma, embryonal, 2, 180295	INHERITED BONE MARROW FAILURE AND/OR PREDISPOSITION PANEL TO HEMATOLOGICAL MALIGNANCIES MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS HEREDITARY CANCER PANEL
DIO1	100%	100%	100%	99.3%	Thyroid hormone metabolism, abnormal, 2, 619855	MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS
DIP2B	100%	100%	100%	99.5%	Intellectual developmental disorder, autosomal dominant, FRA12A type, 136630	INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS

DIP2C	100%	100%	100%	98.7%		INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS
DIS3	100%	100%	100%	99.2%		INHERITED BONE MARROW FAILURE AND/OR PREDISPOSITION PANEL TO HEMATOLOGICAL MALIGNANCIES MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS

DIS3L2	100%	99.9%	100%	99.4%	Perlman syndrome, 267000	TALL STATURE PANEL WITH GENOME WIDE CNV ANALYSIS INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL¹ HEREDITARY CANCER PANEL
DISP1	100%	100%	100%	99.2%		CRANIOFACIAL ANOMALIES PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL¹

DKC1	100%	99.7%	98.6%	67.8%	?Cataracts, hearing impairment, nephrotic syndrome, and enterocolitis 1, 301108;Dyskeratosis congenita, X-linked, 305000	VISION DISORDERS PANEL INHERITED BONE MARROW FAILURE AND/OR PREDISPOSITION PANEL TO HEMATOLOGICAL MALIGNANCIES SKIN DISORDERS PANEL ¹ DYSKERATOSIS CONGENITA AND APLASTIC ANEMIA PANEL PRIMARY IMMUNODEFICIENCIES PANEL LIVER DISORDERS PANEL INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS HEREDITARY CANCER PANEL
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DLAT	100%	100%	100%	99.5%	Pyruvate dehydrogenase E2 deficiency, 245348	MOVEMENT DISORDERS PANEL EPILEPSY PANEL INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS MITOCHONDRIAL DISORDERS PANEL COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL¹
DLC1	100%	100%	100%	98.7%	Colorectal cancer, somatic, 114500	RENAL DISORDERS PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS

DLD	100%	100%	100%	99.4%	Dihydrolipoamide dehydrogenase deficiency, 246900	MOVEMENT DISORDERS PANEL METABOLIC DISORDERS PANEL INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS MITOCHONDRIAL DISORDERS PANEL COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL¹
DLG3	100%	99.6%	98.2%	66.1%	Intellectual developmental disorder, X-linked 90, 300850	INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS

DLG4	100%	100%	100%	98.3%	Intellectual developmental disorder, autosomal dominant 62, 618793	THORACIC AORTIC ANEURYSM AND/OR DISSECTION PANEL INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS
DLG5	100%	100%	100%	98.8%	Yuksel-Vogel-Bauser syndrome, 620703	MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹
DLK1	100%	100%	100%	98.3%		DISORDERS/DIFFERENCES OF SEX DEVELOPMENT (DSD) / PRIMARY ADRENAL INSUFFICIENCY PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS

DLL1	100%	100%	100%	99.4%	Neurodevelopmental disorder with nonspecific brain abnormalities and with or without seizures, 618709	HEARING IMPAIRMENT PANEL (INCLUDING GJB2) INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS
DLL3	100%	100%	100%	97.7%	Spondylocostal dysostosis 1, autosomal recessive, 277300	SHORT STATURE/SKELETAL DYSPLASIA PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹

DLL4	100%	100%	100%	99.2%	Adams-Oliver syndrome 6, 616589	SHORT STATURE/SKELETAL DYSPLASIA PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS OROFACIAL CLEFTING PANEL WITH GENOME WIDE CNV ANALYSIS
DLST	100%	100%	100%	98.8%	Pheochromocytoma/paraganglioma syndrome 7, 618475	MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS MITOCHONDRIAL DISORDERS PANEL HEREDITARY CANCER PANEL

DLX3	100%	100%	100%	98.8%	Trichodontoosseous syndrome, 190320;Amelogenesis imperfecta, type IV, 104510	CRANIOFACIAL ANOMALIES PANEL WITH GENOME WIDE CNV ANALYSIS SKIN DISORDERS PANEL ¹ SHORT STATURE/SKELETAL DYSPLASIA PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS
DLX4	100%	100%	100%	98.9%	?Orofacial cleft 15, 616788	CRANIOFACIAL ANOMALIES PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS

DLX5	100%	100%	100%	98.4%	Split-hand/foot malformation 1, 183600;?Split-hand/foot malformation 1 with sensorineural hearing loss, 220600	SKIN DISORDERS PANEL ¹ HEARING IMPAIRMENT PANEL (INCLUDING GJB2) SHORT STATURE/SKELETAL DYSPLASIA PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹
DLX6	100%	100%	100%	95.4%		SHORT STATURE/SKELETAL DYSPLASIA PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS
DMAC1	100%	100%	100%	97.9%		MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS MITOCHONDRIAL DISORDERS PANEL

DMAC2	100%	100%	100%	99.3%		MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS MITOCHONDRIAL DISORDERS PANEL
DMAC2L	100%	100%	100%	99.2%		MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS MITOCHONDRIAL DISORDERS PANEL
DMC1	100%	100%	100%	99.4%		MALE INFERTILITY PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS

DMD	99.6%	99.5%	98.8%	69.2%	Becker muscular dystrophy, 300376;Cardiomyopathy, dilated, 3B, 302045;Duchenne muscular dystrophy, 310200	DILATED CARDIOMYOPATHY PANEL ¹ MUSCLE DISORDERS PANEL HEART DISORDERS PANEL ¹ INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS
DMGDH	100%	100%	100%	99.2%	Dimethylglycine dehydrogenase deficiency, 605850	METABOLIC DISORDERS PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹

DMP1	100%	100%	100%	99%	Hypophosphatemic rickets, AR, 241520	SHORT STATURE/SKELETAL DYSPLASIA PANEL WITH GENOME WIDE CNV ANALYSIS RENAL DISORDERS PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL¹
DMPK	100%	100%	100%	98.7%	Myotonic dystrophy 1, 160900	INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS

DMRT1	100%	100%	100%	99.1%		DISORDERS/DIFFERENCES OF SEX DEVELOPMENT (DSD) / PRIMARY ADRENAL INSUFFICIENCY PANEL MALE INFERTILITY PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS
DMRT2	100%	100%	100%	99.1%		DISORDERS/DIFFERENCES OF SEX DEVELOPMENT (DSD) / PRIMARY ADRENAL INSUFFICIENCY PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS

DMXL2	100%	100%	100%	99.6%	Developmental and epileptic encephalopathy 81, 618663;?Deafness, autosomal dominant 71, 617605;?Polyendocrine-polyneuropathy syndrome, 616113	HEARING IMPAIRMENT PANEL (INCLUDING GJB2) EPILEPSY PANEL INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹
DNA2	100%	100%	100%	99.5%	Progressive external ophthalmoplegia with mitochondrial DNA deletions, autosomal dominant 6, 615156;Rothmund-Thomson syndrome, type 4, 620819;Seckel syndrome 8, 615807	SHORT STATURE/SKELETAL DYSPLASIA PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS MITOCHONDRIAL DISORDERS PANEL COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹ MUSCLE DISORDERS PANEL

DNAAF1	100%	100%	100%	99%	Ciliary dyskinesia, primary, 13, 613193	CILIOPATHIES PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹
DNAAF11	100%	100%	100%	99.2%	Ciliary dyskinesia, primary, 19, 614935	CILIOPATHIES PANEL MALE INFERTILITY PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹
DNAAF2	100%	100%	100%	99.1%	Ciliary dyskinesia, primary, 10, 612518	CILIOPATHIES PANEL MALE INFERTILITY PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹

DNAAF3	100%	99.9%	100%	99%	Ciliary dyskinesia, primary, 2, 606763	CILIOPATHIES PANEL MALE INFERTILITY PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹
DNAAF4	100%	100%	100%	99.7%	{Dyslexia, susceptibility to, 1}, 127700;Ciliary dyskinesia, primary, 25, 615482	CILIOPATHIES PANEL MALE INFERTILITY PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹
DNAAF5	99.9%	99.1%	99.9%	97.8%	Ciliary dyskinesia, primary, 18, 614874	CILIOPATHIES PANEL MALE INFERTILITY PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹

DNAAF6	100%	100%	99%	68.9%	Ciliary dyskinesia, primary, 36, X-linked, 300991	CILIOPATHIES PANEL MALE INFERTILITY PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS
DNAH1	100%	100%	100%	98.9%	Spermatogenic failure 18, 617576;Ciliary dyskinesia, primary, 37, 617577	CILIOPATHIES PANEL MALE INFERTILITY PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹
DNAH10	100%	100%	100%	99.3%	Spermatogenic failure 56, 619515	MALE INFERTILITY PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS

DNAH11	100%	100%	100%	99.4%	Ciliary dyskinesia, primary, 7, with or without situs inversus, 611884	CILIOPATHIES PANEL MALE INFERTILITY PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹
DNAH12	100%	100%	100%	99.4%		MALE INFERTILITY PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS
DNAH17	100%	100%	100%	98.8%	Spermatogenic failure 39, 618643	CILIOPATHIES PANEL MALE INFERTILITY PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS
DNAH2	100%	99.7%	100%	98.5%	Spermatogenic failure 45, 619094	MALE INFERTILITY PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS

DNAH3	100%	100%	100%	99.2%		MALE INFERTILITY PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS
DNAH5	100%	100%	100%	99.4%	Ciliary dyskinesia, primary, 3, with or without situs inversus, 608644	CILIOPATHIES PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹
DNAH6	100%	100%	100%	99.4%		MALE INFERTILITY PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS
DNAH7	100%	100%	100%	99.5%	Ciliary dyskinesia, primary, 50, 620356	MALE INFERTILITY PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS

DNAH8	100%	100%	100%	99.5%	Spermatogenic failure 46, 619095	CILIOPATHIES PANEL MALE INFERTILITY PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS
DNAH9	100%	100%	100%	99.1%	Ciliary dyskinesia, primary, 40, 618300	CONGENITAL HEARTDISEASE PANEL ¹ CILIOPATHIES PANEL HEART DISORDERS PANEL ¹ MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹
DNAI1	100%	100%	100%	99.5%	Ciliary dyskinesia, primary, 1, with or without situs inversus, 244400	CILIOPATHIES PANEL MALE INFERTILITY PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹

DNAI2	100%	99.9%	100%	99%	Ciliary dyskinesia, primary, 9, with or without situs inversus, 612444	CILIOPATHIES PANEL MALE INFERTILITY PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹
DNAJA3	100%	100%	100%	99%		MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS MITOCHONDRIAL DISORDERS PANEL
DNAJB11	100%	100%	100%	99.2%	Polycystic kidney disease 6 with or without polycystic liver disease, 618061	LIVER DISORDERS PANEL RENAL DISORDERS PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS

DNAJB13	100%	100%	100%	98.9%	Ciliary dyskinesia, primary, 34, 617091	CILIOPATHIES PANEL MALE INFERTILITY PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS
DNAJB2	100%	100%	100%	99.6%	Neuronopathy, distal hereditary motor, autosomal recessive 5, 614881	POLYNEUROPATHIES PANEL ¹ MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹
DNAJB4	100%	100%	100%	99.4%	Congenital myopathy 21 with early respiratory failure, 620326	MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹ MUSCLE DISORDERS PANEL

DNAJB5	100%	100%	100%	98.8%		POLYNEUROPATHIES PANEL ¹ MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS
DNAJB6	100%	100%	100%	99.4%	Muscular dystrophy, limb-girdle, autosomal dominant 1, 603511	MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS MUSCLE DISORDERS PANEL
DNAJC12	100%	100%	100%	99.2%	Hyperphenylalaninemia, mild, non-BH4-deficient, 617384	MOVEMENT DISORDERS PANEL METABOLIC DISORDERS PANEL INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹

DNAJC19	100%	100%	100%	99.7%	3-methylglutaconic aciduria, type V, 610198	HEART DISORDERS PANEL ¹ METABOLIC DISORDERS PANEL INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS MITOCHONDRIAL DISORDERS PANEL COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹
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DNAJC21	100%	100%	100%	99%	Bone marrow failure syndrome 3, 617052	INHERITED BONE MARROW FAILURE AND/OR PREDISPOSITION PANEL TO HEMATOLOGICAL MALIGNANCIES SHORT STATURE/SKELETAL DYSPLASIA PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹ HEREDITARY CANCER PANEL
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DNAJC3	100%	100%	100%	99.3%	Ataxia, combined cerebellar and peripheral, with hearing loss and diabetes mellitus, 616192	MOVEMENT DISORDERS PANEL POLYNEUROPATHIES PANEL ¹ MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS MITOCHONDRIAL DISORDERS PANEL COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹
DNAJC30	100%	100%	100%	98.9%	Leber-like hereditary optic neuropathy, autosomal recessive 1, 619382	VISION DISORDERS PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS MITOCHONDRIAL DISORDERS PANEL COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹
DNAJC5	100%	100%	99.9%	98.1%	Ceroid lipofuscinosis, neuronal, 4 (Kufs type), autosomal dominant, 162350	EPILEPSY PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS

DNAJC6	100%	100%	100%	99.4%	Parkinson disease 19a, juvenile-onset, 615528;Parkinson disease 19b, early-onset, 615528	MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS PARKINSON DISEASE PANEL COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹
DNAL1	100%	100%	100%	99.2%	Ciliary dyskinesia, primary, 16, 614017	CILIOPATHIES PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹
DNAL4	100%	100%	100%	98.4%	?Mirror movements 3, 616059	MOVEMENT DISORDERS PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS
DNALI1	100%	100%	100%	99.1%	Spermatogenic failure 83, 620354	MALE INFERTILITY PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS

DNASE1	100%	100%	100%	99.2%	{Systemic lupus erythematosus, susceptibility to}, 152700	HEMOSTATIC/THROMBOTIC DISORDERS PANEL PRIMARY IMMUNODEFICIENCIES PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS
DNASE1L3	100%	100%	100%	99.7%	Systemic lupus erythematosus 16, 614420	PRIMARY IMMUNODEFICIENCIES PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹
DNASE2	100%	100%	100%	99.5%	Autoinflammatory-pancytopenia syndrome, 619858	PRIMARY IMMUNODEFICIENCIES PANEL LIVER DISORDERS PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS

DNHD1	100%	100%	100%	99.3%	Spermatogenic failure 65, 619712	MALE INFERTILITY PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS
DNM1	100%	99.9%	100%	97.6%	Developmental and epileptic encephalopathy 31B, autosomal recessive, 620352;Developmental and epileptic encephalopathy 31A, autosomal dominant, 616346	EPILEPSY PANEL INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹

DNM1L	100%	100%	100%	99.5%	Optic atrophy 5, 610708;Encephalopathy, lethal, due to defective mitochondrial peroxisomal fission 1, 614388	MOVEMENT DISORDERS PANEL VISION DISORDERS PANEL EPILEPSY PANEL POLYNEUROPATHIES PANEL ¹ NEUROLOGICAL PAIN DISORDERS PANEL ¹ METABOLIC DISORDERS PANEL INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS MITOCHONDRIAL DISORDERS PANEL COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹
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DNM2	100%	100%	100%	99.2%	Centronuclear myopathy 1, 160150;Charcot-Marie-Tooth disease, axonal type 2M, 606482;Charcot-Marie-Tooth disease, dominant intermediate B, 606482;Lethal congenital contracture syndrome 5, 615368	FETAL AKINESIA PANEL POLYNEUROPATHIES PANEL ¹ METABOLIC DISORDERS PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS MITOCHONDRIAL DISORDERS PANEL COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹ MUSCLE DISORDERS PANEL
DNMBP	100%	100%	100%	99.3%	Cataract 48, 618415	VISION DISORDERS PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹

DNMT1	99.9%	99%	100%	99%	Neuropathy, hereditary sensory, type IE, 614116;Cerebellar ataxia, deafness, and narcolepsy, autosomal dominant, 604121	MOVEMENT DISORDERS PANEL POLYNEUROPATHIES PANEL ¹ NEUROLOGICAL PAIN DISORDERS PANEL ¹ METABOLIC DISORDERS PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS
DNMT3A	100%	100%	100%	99%	Tatton-Brown-Rahman syndrome, 615879;Acute myeloid leukemia, somatic, 601626;Heyn-Sproul-Jackson syndrome, 618724	TALL STATURE PANEL WITH GENOME WIDE CNV ANALYSIS SHORT STATURE/SKELETAL DYSPLASIA PANEL WITH GENOME WIDE CNV ANALYSIS INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS

DNMT3B	100%	100%	100%	99.3%	Immunodeficiency-centromeric instability-facial anomalies syndrome 1, 242860;Facioscapulohumeral muscular dystrophy 4, digenic, 619478	PRIMARY IMMUNODEFICIENCIES PANEL METABOLIC DISORDERS PANEL INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL¹
DOCK11	100%	99.9%	99%	70.3%	Autoinflammatory disease, multisystem, with immune dysregulation, X-linked, 301109	PRIMARY IMMUNODEFICIENCIES PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS

DOCK2	100%	100%	100%	99.4%	Immunodeficiency 40, 616433	PRIMARY IMMUNODEFICIENCIES PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹ SEVERE COMBINED IMMUNODEFICIENCY (SCID) PANEL
DOCK3	100%	100%	100%	99.5%	Neurodevelopmental disorder with impaired intellectual development, hypotonia, and ataxia, 618292	INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹
DOCK4	100%	100%	100%	99.4%		INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS

DOCK6	100%	100%	100%	98.5%	Adams-Oliver syndrome 2, 614219	SKIN DISORDERS PANEL ¹ SHORT STATURE/SKELETAL DYSPLASIA PANEL WITH GENOME WIDE CNV ANALYSIS INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹ OROFACIAL CLEFTING PANEL WITH GENOME WIDE CNV ANALYSIS
DOCK7	100%	100%	100%	99.5%	Developmental and epileptic encephalopathy 23, 615859	EPILEPSY PANEL INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹

DOCK8	98.6%	98.6%	100%	99.4%	Hyper-IgE syndrome 2, autosomal recessive, with recurrent infections, 243700	SKIN DISORDERS PANEL ¹ PRIMARY IMMUNODEFICIENCIES PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹ SEVERE COMBINED IMMUNODEFICIENCY (SCID) PANEL
DOHH	100%	100%	100%	98.5%	Neurodevelopmental disorder with microcephaly, cerebral atrophy, and visual impairment, 620066	INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹

DOK7	100%	100%	100%	98.5%	Fetal akinesia deformation sequence 3, 618389;Myasthenic syndrome, congenital, 10, 254300	FETAL AKINESIA PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹ MUSCLE DISORDERS PANEL
DOLK	100%	100%	100%	99.2%	Congenital disorder of glycosylation, type Im, 610768	COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹ SKIN DISORDERS PANEL ¹ HEART DISORDERS PANEL ¹ METABOLIC DISORDERS PANEL INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS

DONSON	100%	100%	100%	99%	Microcephaly, short stature, and limb abnormalities, 617604;Microcephaly-micromelia syndrome, 251230	SHORT STATURE/SKELETAL DYSPLASIA PANEL WITH GENOME WIDE CNV ANALYSIS INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹
DOT1L	100%	100%	100%	98.6%		HEART DISORDERS PANEL ¹ INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS

DPAGT1	100%	100%	100%	99.4%	Myasthenic syndrome, congenital, 13, with tubular aggregates, 614750;Congenital disorder of glycosylation, type Ij, 608093	EPILEPSY PANEL METABOLIC DISORDERS PANEL INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL¹ MUSCLE DISORDERS PANEL
DPCD	100%	100%	100%	97.9%		SHORT STATURE/SKELETAL DYSPLASIA PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS

DPF2	95.7%	95.7%	100%	98.5%	Coffin-Siris syndrome 7, 618027	SHORT STATURE/SKELETAL DYSPLASIA PANEL WITH GENOME WIDE CNV ANALYSIS INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS
DPH1	100%	100%	100%	98.5%	Developmental delay with short stature, dysmorphic facial features, and sparse hair, 616901	INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹
DPH2	100%	100%	100%	99.6%	Developmental delay with short stature, dysmorphic facial features, and sparse hair 2, 620062	MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹

DPH5	100%	100%	100%	99.3%	Neurodevelopmental disorder with short stature, prominent forehead, and feeding difficulties, 620070	INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹
DPM1	99.3%	95.5%	96.9%	89%	Congenital disorder of glycosylation, type 1e, 608799	EPILEPSY PANEL SHORT STATURE/SKELETAL DYSPLASIA PANEL WITH GENOME WIDE CNV ANALYSIS METABOLIC DISORDERS PANEL INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹ MUSCLE DISORDERS PANEL

DPM2	100%	99.9%	100%	99.4%	Congenital disorder of glycosylation, type lu, 615042	EPILEPSY PANEL METABOLIC DISORDERS PANEL INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹ MUSCLE DISORDERS PANEL
DPM3	100%	100%	100%	98.1%	?Muscular dystrophy-dystroglycanopathy (congenital with impaired intellectual development), type B, 15, 618992;Muscular dystrophy-dystroglycanopathy (limb-girdle), type C, 15, 612937	HEART DISORDERS PANEL ¹ METABOLIC DISORDERS PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹ MUSCLE DISORDERS PANEL

DPP6	100%	99.7%	100%	99.2%	Intellectual developmental disorder, autosomal dominant 33, 616311;{Ventricular fibrillation, paroxysmal familial, 2}, 612956	ARRHYTHMIA AND CARDIAC CONDUCTION DISORDERS PANEL ¹ HEART DISORDERS PANEL ¹ INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS
DPP9	100%	100%	100%	98.5%	Hatipoglu immunodeficiency syndrome, 620331	PRIMARY IMMUNODEFICIENCIES PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹

DPY19L2	100%	100%	100%	99.2%	Spermatogenic failure 9, 613958	MALE INFERTILITY PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL¹
DPYD	100%	100%	100%	99.6%	Dihydropyrimidine dehydrogenase deficiency, 274270;5-fluorouracil toxicity, 274270	EPILEPSY PANEL METABOLIC DISORDERS PANEL INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL¹

DPYS	100%	100%	100%	99%	Dihydropyrimidinuria, 222748	MOVEMENT DISORDERS PANEL EPILEPSY PANEL METABOLIC DISORDERS PANEL INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹
DPYSL2	100%	100%	100%	99.2%		INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS
DPYSL5	100%	100%	100%	98.7%	Ritscher-Schinzel syndrome 4, 619435	INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS

DRAM2	100%	100%	100%	98.9%	Cone-rod dystrophy 21, 616502	VISION DISORDERS PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹
DRC1	100%	100%	100%	98.8%	Spermatogenic failure 80, 620222;Ciliary dyskinesia, primary, 21, 615294	CILIOPATHIES PANEL MALE INFERTILITY PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹
DRD4	100%	100%	100%	97.1%	{Attention deficit-hyperactivity disorder}, 143465;Autonomic nervous system dysfunction	MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS

DRG1	100%	100%	100%	99.2%	Tan-Almurshedi syndrome, 620641	INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹
DRP2	100%	99.4%	98.5%	65.5%		MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS POLYNEUROPATHIES PANEL ¹

DSC2	100%	100%	100%	99.1%	Arrhythmogenic right ventricular dysplasia 11 with mild palmoplantar keratoderma and woolly hair, 610476;Arrhythmogenic right ventricular dysplasia 11, 610476	ARITMOGENE CARDIOMYOPATHY PANEL ¹ SKIN DISORDERS PANEL ¹ COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹ ARRHYTHMIA AND CARDIAC CONDUCTION DISORDERS PANEL ¹ HEART DISORDERS PANEL ¹ MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS
DSC3	100%	100%	100%	99.2%	Hypotrichosis and recurrent skin vesicles, 613102	COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹ SKIN DISORDERS PANEL ¹ MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS

DSE	100%	100%	100%	99.7%	Ehlers-Danlos syndrome, musculocontractural type 2, 615539	SKIN DISORDERS PANEL ¹ SHORT STATURE/SKELETAL DYSPLASIA PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹
DSG1	100%	100%	100%	99.2%	Keratosis palmoplantaris striata I, AD, 148700;Erythroderma, congenital, with palmoplantar keratoderma, hypotrichosis, and hyper IgE, 615508	SKIN DISORDERS PANEL ¹ MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹

DSG2	100%	100%	100%	99.4%	Cardiomyopathy, dilated, 1BB, 612877;Arrhythmogenic right ventricular dysplasia 10, 610193	ARITMOGENE CARDIOMYOPATHY PANEL ¹ HEART DISORDERS PANEL ¹ MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹ ARRHYTHMIA AND CARDIAC CONDUCTION DISORDERS PANEL ¹
DSG3	100%	100%	100%	99.7%	Blistering, acantholytic, of oral and laryngeal mucosa, 619226	SKIN DISORDERS PANEL ¹ MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS
DSG4	100%	100%	100%	99.4%	Hypotrichosis 6, 607903	SKIN DISORDERS PANEL ¹ MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹

DSP	100%	100%	100%	99.1%	Arrhythmogenic right ventricular dysplasia 8, 607450;Epidermolysis bullosa, lethal acantholytic, 609638;Keratosis palmoplantaris striata II, 612908;Dilated cardiomyopathy with woolly hair, keratoderma, and tooth agenesis, 615821;Cardiomyopathy, dilated, with woolly hair and keratoderma, 605676	ARITMOGENE CARDIOMYOPATHY PANEL ¹ DILATED CARDIOMYOPATHY PANEL ¹ ARRHYTHMIA AND CARDIAC CONDUCTION DISORDERS PANEL ¹ MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹ HEART DISORDERS PANEL ¹ SKIN DISORDERS PANEL ¹
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DSPP	100%	100%	99%	96.1%	Dentinogenesis imperfecta, Shields type III, 125500;Dentinogenesis imperfecta, Shields type II, 125490;Dentin dysplasia, type II, 125420;Deafness, autosomal dominant 39, with dentinogenesis, 605594	MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS HEARING IMPAIRMENT PANEL (INCLUDING GJB2) CRANIOFACIAL ANOMALIES PANEL WITH GENOME WIDE CNV ANALYSIS SKIN DISORDERS PANEL ¹
DST	100%	100%	100%	99.5%	Neuropathy, hereditary sensory and autonomic, type VI, 614653;Epidermolysis bullosa simplex 3, localized or generalized intermediate, with bp230 deficiency, 615425	MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹ SKIN DISORDERS PANEL ¹ POLYNEUROPATHIES PANEL ¹

DSTYK	100%	100%	100%	99.3%	Spastic paraplegia 23, autosomal recessive, 270750;Congenital anomalies of kidney and urinary tract 1, 610805	MOVEMENT DISORDERS PANEL RENAL DISORDERS PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹
DTNA	98.6%	97.2%	100%	99.7%	Left ventricular noncompaction 1, with or without congenital heart defects, 604169;Myopathy with myalgia, increased serum creatine kinase, and with or without episodic rhabdomyolysis 2, 620971	HEART DISORDERS PANEL ¹ MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS
DTNBP1	100%	100%	100%	99.1%	Hermansky-Pudlak syndrome 7, 614076	VISION DISORDERS PANEL SKIN DISORDERS PANEL ¹ HEMOSTATIC/THROMBOTIC DISORDERS PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹

DTYMK	100%	100%	100%	98.7%	Neurodegeneration, childhood-onset, with progressive microcephaly, 619847	MOVEMENT DISORDERS PANEL EPILEPSY PANEL METABOLIC DISORDERS PANEL INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹
DUOX2	100%	100%	100%	98.7%	Thyroid dyshormonogenesis 6, 607200	MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹
DUOXA2	100%	100%	100%	99.3%	Thyroid dyshormonogenesis 5, 274900	MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹

DUSP6	100%	100%	100%	99.3%	Hypogonadotropic hypogonadism 19 with or without anosmia, 615269	SKIN DISORDERS PANEL ¹ DISORDERS/DIFFERENCES OF SEX DEVELOPMENT (DSD) / PRIMARY ADRENAL INSUFFICIENCY PANEL HYPOGONADOTROPIC HYPOGONADISM (KALLMANN) PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS
DUT	100%	100%	100%	99.2%	Bone marrow failure and diabetes mellitus syndrome, 620044	INHERITED BONE MARROW FAILURE AND/OR PREDISPOSITION PANEL TO HEMATOLOGICAL MALIGNANCIES MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹ HEREDITARY CANCER PANEL

DVL1	100%	99.9%	100%	98.9%	Robinow syndrome, autosomal dominant 2, 616331	SHORT STATURE/SKELETAL DYSPLASIA PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS OROFACIAL CLEFTING PANEL WITH GENOME WIDE CNV ANALYSIS
DVL3	100%	100%	100%	98.9%	Robinow syndrome, autosomal dominant 3, 616894	SHORT STATURE/SKELETAL DYSPLASIA PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS OROFACIAL CLEFTING PANEL WITH GENOME WIDE CNV ANALYSIS

DYM	100%	100%	100%	99.8%	Smith-McCort dysplasia, 607326;Dyggve-Melchior-Clausen disease, 223800	SHORT STATURE/SKELETAL DYSPLASIA PANEL WITH GENOME WIDE CNV ANALYSIS INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹
DYNC1H1	99.3%	99.3%	100%	99.2%	Charcot-Marie-Tooth disease, axonal, type 2O, 614228;Spinal muscular atrophy, lower extremity-predominant 1, AD, 158600;Cortical dysplasia, complex, with other brain malformations 13, 614563	EPILEPSY PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS MUSCLE DISORDERS PANEL NEUROLOGICAL PAIN DISORDERS PANEL ¹ INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS POLYNEUROPATHIES PANEL ¹

DYNC112	100%	100%	100%	99.5%	Neurodevelopmental disorder with microcephaly and structural brain anomalies, 618492	MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹ INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS
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DYNC2H1	100%	100%	100%	99.4%	Short-rib thoracic dysplasia 3 with or without polydactyly, 613091	VISION DISORDERS PANEL CILIOPATHIES PANEL DISORDERS/DIFFEREN CES OF SEX DEVELOPMENT (DSD) / PRIMARY ADRENAL INSUFFICIENCY PANEL SHORT STATURE/SKELETAL DYSPLASIA PANEL WITH GENOME WIDE CNV ANALYSIS RENAL DISORDERS PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL¹ OROFACIAL CLEFTING PANEL WITH GENOME WIDE CNV ANALYSIS
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DYNC2I1	100%	100%	100%	99.2%	Short-rib thoracic dysplasia 8 with or without polydactyly, 615503	CILIOPATHIES PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹ RENAL DISORDERS PANEL DISORDERS/DIFFEREN CES OF SEX DEVELOPMENT (DSD) / PRIMARY ADRENAL INSUFFICIENCY PANEL SHORT STATURE/SKELETAL DYSPLASIA PANEL WITH GENOME WIDE CNV ANALYSIS
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DYNC2I2	100%	100%	100%	98.9%	Short-rib thoracic dysplasia 11 with or without polydactyly, 615633	VISION DISORDERS PANEL CILIOPATHIES PANEL SHORT STATURE/SKELETAL DYSPLASIA PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹
DYNC2LI1	100%	100%	100%	99.5%	Short-rib thoracic dysplasia 15 with polydactyly, 617088	CILIOPATHIES PANEL SHORT STATURE/SKELETAL DYSPLASIA PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹ OROFACIAL CLEFTING PANEL WITH GENOME WIDE CNV ANALYSIS

DYNLT2B	100%	100%	100%	99.1%	Short-rib thoracic dysplasia 17 with or without polydactyly, 617405	CILIOPATHIES PANEL SHORT STATURE/SKELETAL DYSPLASIA PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹
DYRK1A	100%	100%	100%	99.2%	Intellectual developmental disorder, autosomal dominant 7, 614104	CONGENITAL HEARTDISEASE PANEL ¹ EPILEPSY PANEL HEART DISORDERS PANEL ¹ INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS
DYRK1B	96.2%	96.1%	100%	98.3%	Abdominal obesity-metabolic syndrome 3, 615812	MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS

DYSF	96.9%	96.8%	100%	99.3%	Muscular dystrophy, limb-girdle, autosomal recessive 2, 253601;Miyoshi muscular dystrophy 1, 254130;Myopathy, distal, with anterior tibial onset, 606768	MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹ MUSCLE DISORDERS PANEL
DZIP1	100%	100%	100%	99.3%	Spermatogenic failure 47, 619102;?Mitral valve prolapse 3, 610840	CONGENITAL HEARTDISEASE PANEL ¹ HEART DISORDERS PANEL ¹ MALE INFERTILITY PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS
DZIP1L	100%	100%	100%	98.9%	Polycystic kidney disease 5, 617610	RENAL DISORDERS PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹

E2F1	99.8%	98.4%	100%	99.2%		MALE INFERTILITY PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS
EARS2	100%	100%	100%	99.2%	Combined oxidative phosphorylation deficiency 12, 614924	INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS MITOCHONDRIAL DISORDERS PANEL COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹
EBF3	100%	100%	100%	98.3%	Hypotonia, ataxia, and delayed development syndrome, 617330	MOVEMENT DISORDERS PANEL INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS

EBP	100%	99.1%	97.8%	68.4%	MEND syndrome, 300960;Chondrodysplasia punctata, X-linked dominant, 302960	SKIN DISORDERS PANEL ¹ EPILEPSY PANEL SHORT STATURE/SKELETAL DYSPLASIA PANEL WITH GENOME WIDE CNV ANALYSIS METABOLIC DISORDERS PANEL INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS OROFACIAL CLEFTING PANEL WITH GENOME WIDE CNV ANALYSIS
ECE1	100%	100%	100%	99%	{Hypertension, essential, susceptibility to}, 145500;?Hirschsprung disease, cardiac defects, and autonomic dysfunction, 613870	MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS

ECEL1	100%	100%	100%	98.8%	Arthrogryposis, distal, type 5D, 615065	FETAL AKINESIA PANEL SHORT STATURE/SKELETAL DYSPLASIA PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹ MUSCLE DISORDERS PANEL
ECHS1	100%	100%	100%	99.4%	Mitochondrial short-chain enoyl-CoA hydratase 1 deficiency, 616277	MOVEMENT DISORDERS PANEL METABOLIC DISORDERS PANEL INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS MITOCHONDRIAL DISORDERS PANEL COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹

ECM1	100%	100%	100%	98.5%	Urbach-Wiethe disease, 247100	SKIN DISORDERS PANEL ¹ MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹
ECSIT	100%	100%	100%	97%		MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS MITOCHONDRIAL DISORDERS PANEL
EDA	100%	99.7%	98.3%	64.2%	Tooth agenesis, selective, X-linked 1, 313500;Ectodermal dysplasia 1, hypohidrotic, X-linked, 305100	CRANIOFACIAL ANOMALIES PANEL WITH GENOME WIDE CNV ANALYSIS SKIN DISORDERS PANEL ¹ MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS

EDAR	100%	100%	100%	99.1%	[Hair morphology 1, hair thickness], 612630;Ectodermal dysplasia 10A, hypohidrotic/hair/nail type, autosomal dominant, 129490;Ectodermal dysplasia 10B, hypohidrotic/hair/tooth type, autosomal recessive, 224900	CRANIOFACIAL ANOMALIES PANEL WITH GENOME WIDE CNV ANALYSIS SKIN DISORDERS PANEL ¹ MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹
EDARADD	100%	100%	100%	98.2%	Ectodermal dysplasia 11B, hypohidrotic/hair/tooth type, autosomal recessive, 614941;Ectodermal dysplasia 11A, hypohidrotic/hair/tooth type, autosomal dominant, 614940	CRANIOFACIAL ANOMALIES PANEL WITH GENOME WIDE CNV ANALYSIS SKIN DISORDERS PANEL ¹ MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹

EDC3	100%	100%	100%	99.2%	?Intellectual developmental disorder, autosomal recessive 50, 616460	INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL¹
EDEM3	100%	100%	100%	99.3%	Congenital disorder of glycosylation, type IIv, 619493	METABOLIC DISORDERS PANEL INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL¹

EDN1	100%	100%	100%	98.9%	Question mark ears, isolated, 612798;Auriculocondylar syndrome 3, 615706	CRANIOFACIAL ANOMALIES PANEL WITH GENOME WIDE CNV ANALYSIS SHORT STATURE/SKELETAL DYSPLASIA PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹ OROFACIAL CLEFTING PANEL WITH GENOME WIDE CNV ANALYSIS
EDN3	100%	100%	100%	99.4%	Waardenburg syndrome, type 4B, 613265;{Hirschsprung disease, susceptibility to, 4}, 613712	SKIN DISORDERS PANEL ¹ HEARING IMPAIRMENT PANEL (INCLUDING GJB2) MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹

EDNRA	100%	100%	100%	99.5%	{Migraine, resistance to}, 157300;Mandibulofacial dysostosis with alopecia, 616367	CRANIOFACIAL ANOMALIES PANEL WITH GENOME WIDE CNV ANALYSIS SKIN DISORDERS PANEL ¹ SHORT STATURE/SKELETAL DYSPLASIA PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS OROFACIAL CLEFTING PANEL WITH GENOME WIDE CNV ANALYSIS
EDNRB	100%	100%	100%	99.5%	{Hirschsprung disease, susceptibility to, 2}, 600155;?ABCD syndrome, 600501;Waardenburg syndrome, type 4A, 277580	SKIN DISORDERS PANEL ¹ HEARING IMPAIRMENT PANEL (INCLUDING GJB2) LIVER DISORDERS PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹

EED	99.1%	99.1%	99.9%	94.2%	Cohen-Gibson syndrome, 617561	TALL STATURE PANEL WITH GENOME WIDE CNV ANALYSIS INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS
EEF1A2	99.2%	96.8%	100%	98.7%	Developmental and epileptic encephalopathy 33, 616409;Intellectual developmental disorder, autosomal dominant 38, 616393	EPILEPSY PANEL HEART DISORDERS PANEL ¹ INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS
EEF1D	100%	100%	100%	99.1%		INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS

EEF2	100%	100%	100%	98.9%	?Spinocerebellar ataxia 26, 609306	MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS
EEFSEC	100%	100%	100%	98.9%		METABOLIC DISORDERS PANEL INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹
EFEMP1	100%	100%	100%	99.3%	Doyne honeycomb degeneration of retina, 126600;Cutis laxa, autosomal recessive, type ID, 620780;Glaucoma 1, open angle, H, 611276	THORACIC AORTIC ANEURYSM AND/OR DISSECTION PANEL VISION DISORDERS PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹

EFEMP2	100%	100%	100%	99.1%	Cutis laxa, autosomal recessive, type IB, 614437	THORACIC AORTIC ANEURYSM AND/OR DISSECTION PANEL SKIN DISORDERS PANEL ¹ MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹
EFHC1	97.8%	97.5%	100%	99.6%	{Epilepsy, juvenile absence, susceptibility to, 1}, 607631;{Myoclonic epilepsy, juvenile, susceptibility to, 1}, 254770	EPILEPSY PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS

EFL1	99.2%	99.2%	100%	99.6%	Shwachman-Diamond syndrome 2, 617941	INHERITED BONE MARROW FAILURE AND/OR PREDISPOSITION PANEL TO HEMATOLOGICAL MALIGNANCIES SHORT STATURE/SKELETAL DYSPLASIA PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹
EFNA4	100%	100%	100%	99.1%		CRANIOFACIAL ANOMALIES PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS

EFNB1	100%	100%	98.7%	67.5%	Craniofrontonasal dysplasia, 304110	CRANIOFACIAL ANOMALIES PANEL WITH GENOME WIDE CNV ANALYSIS SKIN DISORDERS PANEL ¹ SHORT STATURE/SKELETAL DYSPLASIA PANEL WITH GENOME WIDE CNV ANALYSIS INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS OROFACIAL CLEFTING PANEL WITH GENOME WIDE CNV ANALYSIS
EFNB2	100%	100%	100%	99.3%		HEARING IMPAIRMENT PANEL (INCLUDING GJB2) INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS

EFTUD2	100%	100%	100%	99%	Mandibulofacial dysostosis, Guion-Almeida type, 610536	CRANIOFACIAL ANOMALIES PANEL WITH GENOME WIDE CNV ANALYSIS SHORT STATURE/SKELETAL DYSPLASIA PANEL WITH GENOME WIDE CNV ANALYSIS INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS OROFACIAL CLEFTING PANEL WITH GENOME WIDE CNV ANALYSIS
EGF	100%	100%	100%	99.5%	?Hypomagnesemia 4, renal, 611718	EPILEPSY PANEL RENAL DISORDERS PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹

EGFLAM	100%	100%	100%	99.4%		VISION DISORDERS PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS
EGFR	100%	100%	100%	99%	Neonatal nephrocutaneous inflammatory syndrome, 616069;Nonsmall cell lung cancer, response to tyrosine kinase inhibitor in, 211980;Adenocarcinoma of lung, response to tyrosine kinase inhibitor in, 211980;{Nonsmall cell lung cancer, susceptibility to}, 211980	MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹ HEREDITARY CANCER PANEL
EGLN1	100%	100%	100%	97.5%	Erythrocytosis, familial, 3, 609820;[Hemoglobin, high altitude adaptation], 609070	MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS HEREDITARY CANCER PANEL
EGLN2	100%	100%	100%	98.8%		MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS HEREDITARY CANCER PANEL

EGR2	100%	100%	100%	98.8%	Dejerine-Sottas disease, 145900;Charcot-Marie-Tooth disease, type 1D, 607678;Hypomyelinating neuropathy, congenital, 1, 605253	FETAL AKINESIA PANEL POLYNEUROPATHIES PANEL ¹ MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹
EHD1	100%	100%	100%	98.6%		HEARING IMPAIRMENT PANEL (INCLUDING GJB2) MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS
EHHADH	100%	100%	100%	99.4%	?Fanconi renotubular syndrome 3, 615605	RENAL DISORDERS PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS MITOCHONDRIAL DISORDERS PANEL

EHMT1	99.8%	99.5%	100%	98.5%	Kleefstra syndrome 1, 610253	CONGENITAL HEARTDISEASE PANEL¹ EPILEPSY PANEL HEART DISORDERS PANEL¹ INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS
EIF1AY	50%	50%	49.1%	22.2%		MALE INFERTILITY PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS
EIF2AK1	100%	100%	100%	99.5%	?Leukoencephalopathy, motor delay, spasticity, and dysarthria syndrome, 618878	MOVEMENT DISORDERS PANEL INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS

EIF2AK2	100%	100%	100%	99.8%	Leukoencephalopathy, developmental delay, and episodic neurologic regression syndrome, 618877;Dystonia 33, 619687	MOVEMENT DISORDERS PANEL INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹
EIF2AK3	100%	100%	100%	99.4%	Wolcott-Rallison syndrome, 226980	SKIN DISORDERS PANEL ¹ SHORT STATURE/SKELETAL DYSPLASIA PANEL WITH GENOME WIDE CNV ANALYSIS INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹

EIF2AK4	100%	100%	100%	99.2%	Pulmonary venoocclusive disease 2, 234810	HEART DISORDERS PANEL ¹ MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹
EIF2B1	100%	100%	100%	99.4%	Leukoencephalopathy with vanishing white matter 1, with or without ovarian failure, 603896	MOVEMENT DISORDERS PANEL EPILEPSY PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹
EIF2B2	100%	100%	100%	97.9%	Leukoencephalopathy with vanishing white matter 2, with or without ovarian failure, 620312	MOVEMENT DISORDERS PANEL EPILEPSY PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹

EIF2B3	100%	100%	100%	99.3%	Leukoencephalopathy with vanishing white matter 3, with or without ovarian failure, 620313	MOVEMENT DISORDERS PANEL EPILEPSY PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL¹
EIF2B4	100%	100%	100%	99.2%	Leukoencephalopathy with vanishing white matter 4, with or without ovarian failure, 620314	MOVEMENT DISORDERS PANEL EPILEPSY PANEL INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL¹

EIF2B5	100%	100%	100%	99.3%	Leukoencephalopathy with vanishing white matter 5, with or without ovarian failure, 620315	MOVEMENT DISORDERS PANEL DISORDERS/DIFFERENCES OF SEX DEVELOPMENT (DSD) / PRIMARY ADRENAL INSUFFICIENCY PANEL EPILEPSY PANEL INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹ PREMATURE OVARIAN INSUFFICIENCY PANEL
EIF2S3	100%	100%	98.8%	70%	MEHMO syndrome, 300148	INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS OROFACIAL CLEFTING PANEL WITH GENOME WIDE CNV ANALYSIS

EIF3F	100%	100%	100%	98.6%	Intellectual developmental disorder, autosomal recessive 67, 618295	INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹
EIF4A2	100%	100%	100%	99.8%	Neurodevelopmental disorder with hypotonia and speech delay, with or without seizures, 620455	INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹

EIF4A3	100%	100%	100%	99.2%	Robin sequence with cleft mandible and limb anomalies, 268305	CRANIOFACIAL ANOMALIES PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL¹ OROFACIAL CLEFTING PANEL WITH GENOME WIDE CNV ANALYSIS INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS SHORT STATURE/SKELETAL DYSPLASIA PANEL WITH GENOME WIDE CNV ANALYSIS
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EIF4ENIF1	100%	100%	100%	99.2%		DISORDERS/DIFFERENCES OF SEX DEVELOPMENT (DSD) / PRIMARY ADRENAL INSUFFICIENCY PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS PREMATURE OVARIAN INSUFFICIENCY PANEL
EIF5A	100%	100%	100%	98.6%	Faundes-Banka syndrome, 619376	INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS

ELAC2	100%	100%	100%	99.4%	{Prostate cancer, hereditary, 2, susceptibility to}, 614731;Combined oxidative phosphorylation deficiency 17, 615440	MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS MITOCHONDRIAL DISORDERS PANEL COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹ HEART DISORDERS PANEL ¹ INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS
ELANE	100%	100%	100%	99.2%	Neutropenia, cyclic, 162800;Neutropenia, severe congenital 1, autosomal dominant, 202700	HEREDITARY CANCER PANEL INHERITED BONE MARROW FAILURE AND/OR PREDISPOSITION PANEL TO HEMATOLOGICAL MALIGNANCIES PRIMARY IMMUNODEFICIENCIES PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS

ELF2	100%	100%	100%	99.5%		POLYNEUROPATHIES PANEL ¹ MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS
ELF4	100%	99.4%	98.3%	65.5%	Autoinflammatory syndrome, familial, X-linked, Behcet-like 2, 301074	PRIMARY IMMUNODEFICIENCIES PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS
ELMO2	100%	100%	100%	99.7%	Vascular malformation, primary intraosseous, 606893	SHORT STATURE/SKELETAL DYSPLASIA PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹

ELMOD3	100%	100%	100%	99.7%	?Deafness, autosomal recessive 88, 615429;?Deafness, autosomal dominant 81, 619500	HEARING IMPAIRMENT PANEL (INCLUDING GJB2) MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS
ELN	100%	100%	100%	98.7%	Cutis laxa, autosomal dominant, 123700;Supravalvar aortic stenosis, 185500	THORACIC AORTIC ANEURYSM AND/OR DISSECTION PANEL CONGENITAL HEARTDISEASE PANEL¹ SKIN DISORDERS PANEL¹ HEART DISORDERS PANEL¹ MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS

ELOVL1	100%	100%	100%	99.4%	Ichthyotic keratoderma, spasticity, hypomyelination, and dysmorphic facies, 618527	MOVEMENT DISORDERS PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS VISION DISORDERS PANEL SKIN DISORDERS PANEL ¹ HEARING IMPAIRMENT PANEL (INCLUDING GJB2) METABOLIC DISORDERS PANEL
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ELOVL4	100%	100%	100%	99.4%	Spinocerebellar ataxia 34, 133190;Stargardt disease 3, 600110;Ichthyosis, spastic quadriplegia, and impaired intellectual development, 614457	MOVEMENT DISORDERS PANEL VISION DISORDERS PANEL SKIN DISORDERS PANEL ¹ METABOLIC DISORDERS PANEL INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹
ELOVL5	100%	100%	100%	99.6%	Spinocerebellar ataxia 38, 615957	MOVEMENT DISORDERS PANEL POLYNEUROPATHIES PANEL ¹ MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS

ELP1	100%	100%	100%	99.6%	{Medulloblastoma}, 155255;Dysautonomia, familial, 223900	POLYNEUROPATHIES PANEL ¹ HEREDITARY CANCER PANEL NEUROLOGICAL PAIN DISORDERS PANEL ¹ MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹ SONIC HEDGEHOG MEDULLOBLASTOMA PANEL
ELP2	100%	100%	100%	99.6%	Intellectual developmental disorder, autosomal recessive 58, 617270	INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹

ELP4	87.8%	87.8%	100%	99.4%	?Aniridia 2, 617141	VISION DISORDERS PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS
EMC1	100%	100%	100%	98.9%	Cerebellar atrophy, visual impairment, and psychomotor retardation, 616875	VISION DISORDERS PANEL INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹
EMC10	100%	100%	100%	98.4%	Neurodevelopmental disorder with dysmorphic facies and variable seizures, 619264	INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹

EMD	93.1%	89.5%	98.5%	62.9%	Emery-Dreifuss muscular dystrophy 1, X-linked, 310300	HEART DISORDERS PANEL ¹ MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS MITOCHONDRIAL DISORDERS PANEL MUSCLE DISORDERS PANEL
EMG1	100%	100%	100%	99.1%	Bowen-Conradi syndrome, 211180	MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹
EMILIN1	100%	100%	100%	98.9%	Neuronopathy, distal hereditary motor, autosomal dominant 10, 620080;Arterial tortuosity-bone fragility syndrome, 620908	THORACIC AORTIC ANEURYSM AND/OR DISSECTION PANEL POLYNEUROPATHIES PANEL ¹ MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹

EML1	100%	100%	100%	98.8%	Band heterotopia, 600348	INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹
EMP2	100%	99.9%	100%	97.7%	Nephrotic syndrome, type 10, 615861	RENAL DISORDERS PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹
EMX2	100%	100%	100%	93.6%	Schizencephaly, 269160	INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS

EN1	100%	99.5%	100%	94.5%	?ENDOVE syndrome, limb-brain type, 619218	SHORT STATURE/SKELETAL DYSPLASIA PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS
ENAM	100%	100%	100%	99.7%	Amelogenesis imperfecta, type IC, 204650;Amelogenesis imperfecta, type IB, 104500	CRANIOFACIAL ANOMALIES PANEL WITH GENOME WIDE CNV ANALYSIS SKIN DISORDERS PANEL ¹ MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹
ENG	100%	99.9%	100%	95.5%	Telangiectasia, hereditary hemorrhagic, type 1, 187300	SKIN DISORDERS PANEL ¹ HEMOSTATIC/THROMB OTIC DISORDERS PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS

ENO3	100%	100%	100%	96.5%	Glycogen storage disease XIII, 612932	METABOLIC DISORDERS PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹ MUSCLE DISORDERS PANEL
ENPP1	99.7%	98.8%	100%	98.9%	{Obesity, susceptibility to}, 601665;Hypophosphatemic rickets, autosomal recessive, 2, 613312;{Diabetes mellitus, non-insulin-dependent, susceptibility to}, 125853;Arterial calcification, generalized, of infancy, 1, 208000;Cole disease, 615522	COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹ SKIN DISORDERS PANEL ¹ HEART DISORDERS PANEL ¹ SHORT STATURE/SKELETAL DYSPLASIA PANEL WITH GENOME WIDE CNV ANALYSIS RENAL DISORDERS PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS

ENTPD1	100%	100%	100%	99.5%	Spastic paraplegia 64, autosomal recessive, 615683	MOVEMENT DISORDERS PANEL INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹
EOGT	98.6%	95%	100%	99.6%	Adams-Oliver syndrome 4, 615297	SHORT STATURE/SKELETAL DYSPLASIA PANEL WITH GENOME WIDE CNV ANALYSIS METABOLIC DISORDERS PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹ OROFACIAL CLEFTING PANEL WITH GENOME WIDE CNV ANALYSIS

EP300	100%	100%	100%	99.1%	Menke-Hennekam syndrome 2, 618333;Colorectal cancer, somatic, 114500;Rubinstein-Taybi syndrome 2, 613684	SHORT STATURE/SKELETAL DYSPLASIA PANEL WITH GENOME WIDE CNV ANALYSIS INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS
EP400	100%	100%	100%	98.7%		INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS
EPAS1	100%	100%	100%	99%	Erythrocytosis, familial, 4, 611783	MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS HEREDITARY CANCER PANEL

EPB41	100%	100%	100%	99.1%	Elliptocytosis-1, 611804	MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹
EPB41L1	100%	100%	100%	99.1%	?Intellectual developmental disorder, autosomal dominant 11, 614257	MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS
EPB41L3	100%	100%	100%	99%		INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS
EPB42	100%	100%	100%	98.5%	Spherocytosis, type 5, 612690	MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹

EPCAM	100%	100%	100%	99.3%	Diarrhea 5, with tufting enteropathy, congenital, 613217;Lynch syndrome 8, 613244	PANEL HEREDITARY COLORECTAL AND POLYPOSIS PANEL HEREDITARY CANCER (CDH1, CTNNA1, MLH1, MSH2, MSH6, PMS2) PANEL LYNCH SYNDROME (MLH1, PMS2, MSH2, MSH6) MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS PANEL HEREDITARY PANCREATIC CANCER (BRCA1, BRCA2, PALB2, ATM, CDKN2A, MLH1, MSH2, MSH6, PMS2, STK11) COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹ HEREDITARY CANCER PANEL
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EPG5	100%	100%	100%	99.4%	Vici syndrome, 242840	VISION DISORDERS PANEL SKIN DISORDERS PANEL ¹ PRIMARY IMMUNODEFICIENCIES PANEL METABOLIC DISORDERS PANEL INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS MITOCHONDRIAL DISORDERS PANEL COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹ OROFACIAL CLEFTING PANEL WITH GENOME WIDE CNV ANALYSIS
EPHA10	100%	100%	100%	97.7%	?Deafness, autosomal dominant 88, 620283	MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS

EPHA2	100%	100%	100%	98.5%	Cataract 6, multiple types, 116600	VISION DISORDERS PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS
EPHA7	100%	100%	100%	99.3%		INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS
EPHB2	99.7%	98.9%	100%	98.5%	?Bleeding disorder, platelet-type, 22, 618462;{Prostate cancer/brain cancer susceptibility, somatic}, 603688	HEMOSTATIC/THROMB OTIC DISORDERS PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS

EPHB4	100%	100%	100%	98.9%	Capillary malformation-arteriovenous malformation 2, 618196;Lymphatic malformation 7, 617300	SKIN DISORDERS PANEL ¹ LYMPHATIC ANOMALIES PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS
EPHX1	100%	100%	100%	98.6%		LIVER DISORDERS PANEL METABOLIC DISORDERS PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹
EPHX2	100%	100%	100%	98.4%	{Hypercholesterolemia, familial, due to LDLR defect, modifier of}, 143890	METABOLIC DISORDERS PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS

EPM2A	100%	100%	100%	98.9%	Myoclonic epilepsy of Lafora 1, 254780	EPILEPSY PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL¹
EPO	100%	100%	100%	98.4%	{Microvascular complications of diabetes 2}, 612623;Erythrocytosis, familial, 5, 617907;?Diamond-Blackfan anemia-like, 617911	INHERITED BONE MARROW FAILURE AND/OR PREDISPOSITION PANEL TO HEMATOLOGICAL MALIGNANCIES MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL¹

EPRS1	100%	100%	100%	99%	Leukodystrophy, hypomyelinating, 15, 617951	INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹
EPS8	100%	100%	100%	99.4%	?Deafness, autosomal recessive 102, 615974	HEARING IMPAIRMENT PANEL (INCLUDING GJB2) MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS
EPS8L2	100%	100%	100%	97.6%	Deafness autosomal recessive 106, 617637	HEARING IMPAIRMENT PANEL (INCLUDING GJB2) MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹

EPS8L3	100%	100%	100%	98.8%	?Hypotrichosis 5, 612841	SKIN DISORDERS PANEL ¹ MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS
ERAL1	100%	100%	100%	99.2%	Perrault syndrome 6, 617565	HEARING IMPAIRMENT PANEL (INCLUDING GJB2) DISORDERS/DIFFEREN CES OF SEX DEVELOPMENT (DSD) / PRIMARY ADRENAL INSUFFICIENCY PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS MITOCHONDRIAL DISORDERS PANEL COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹ PREMATURE OVARIAN INSUFFICIENCY PANEL
ERBB2	100%	100%	100%	98.2%	Gastric cancer, somatic, 613659;Adenocarcinoma of lung, somatic, 211980;Ovarian cancer, somatic, 167000;?Visceral neuropathy, familial, 2, autosomal recessive, 619465;Glioblastoma, somatic, 137800	POLYNEUROPATHIES PANEL ¹ MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS

ERBB3	100%	100%	100%	98.3%	?Lethal congenital contractural syndrome 2, 607598;{?Erythroleukemia, familial, susceptibility to}, 133180;Visceral neuropathy, familial, 1, autosomal recessive, 243180	FETAL AKINESIA PANEL POLYNEUROPATHIES PANEL ¹ LIVER DISORDERS PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹ MUSCLE DISORDERS PANEL
ERBB4	100%	100%	100%	99.3%	Amyotrophic lateral sclerosis 19, 615515	AMYOTROPHIC LATERAL SCLEROSIS, ALS PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS
ERBIN	100%	100%	100%	99.5%		PRIMARY IMMUNODEFICIENCIES PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS

ERCC1	100%	100%	100%	98.5%	Cerebrooculofacioskeletal syndrome 4, 610758	INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹ HEREDITARY CANCER PANEL
ERCC2	99.9%	98.3%	100%	98.7%	Xeroderma pigmentosum, group D, 278730;Trichothiodystrophy 1, photosensitive, 601675;?Cerebrooculofacioskeletal syndrome 2, 610756	MOVEMENT DISORDERS PANEL VISION DISORDERS PANEL SKIN DISORDERS PANEL ¹ INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹ HEREDITARY CANCER PANEL

ERCC3	100%	100%	100%	99.6%	Trichothiodystrophy 2, photosensitive, 616390;Xeroderma pigmentosum, group B, 610651	VISION DISORDERS PANEL SKIN DISORDERS PANEL ¹ INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹ HEREDITARY CANCER PANEL
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ERCC4	100%	100%	100%	99.1%	Xeroderma pigmentosum, type F/Cockayne syndrome, 278760;XFE progeroid syndrome, 610965;Xeroderma pigmentosum, group F, 278760;Fanconi anemia, complementation group Q, 615272	MOVEMENT DISORDERS PANEL INHERITED BONE MARROW FAILURE AND/OR PREDISPOSITION PANEL TO HEMATOLOGICAL MALIGNANCIES SKIN DISORDERS PANEL ¹ SHORT STATURE/SKELETAL DYSPLASIA PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹ HEREDITARY CANCER PANEL
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ERCC5	100%	100%	100%	99.3%	Xeroderma pigmentosum, group G, 278780;Cerebrooculofacioskeletal syndrome 3, 616570;Xeroderma pigmentosum, group G/Cockayne syndrome, 278780	FETAL AKINESIA PANEL SKIN DISORDERS PANEL ¹ INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹ HEREDITARY CANCER PANEL
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ERCC6	100%	100%	100%	99.3%	UV-sensitive syndrome 1, 600630;Cerebrooculofacioskeletal syndrome 1, 214150;?De Sanctis-Cacchione syndrome, 278800;Cockayne syndrome, type B, 133540;{Macular degeneration, age-related, susceptibility to, 5}, 613761;Premature ovarian failure 11, 616946;{Lung cancer, susceptibility to}, 211980	FETAL AKINESIA PANEL PREMATURE OVARIAN INSUFFICIENCY PANEL HEREDITARY CANCER PANEL INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹ VISION DISORDERS PANEL SKIN DISORDERS PANEL ¹ DISORDERS/DIFFEREN CES OF SEX DEVELOPMENT (DSD) / PRIMARY ADRENAL INSUFFICIENCY PANEL
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ERCC6L2	100%	100%	100%	99.7%	Bone marrow failure syndrome 2, 615715	INHERITED BONE MARROW FAILURE AND/OR PREDISPOSITION PANEL TO HEMATOLOGICAL MALIGNANCIES MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹
ERCC8	95.2%	95.2%	99.7%	95.5%	UV-sensitive syndrome 2, 614621; Cockayne syndrome, type A, 216400	VISION DISORDERS PANEL SKIN DISORDERS PANEL ¹ INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹

ERF	100%	100%	100%	96%	Craniosynostosis 4, 600775;Chitayat syndrome, 617180	CRANIOFACIAL ANOMALIES PANEL WITH GENOME WIDE CNV ANALYSIS SHORT STATURE/SKELETAL DYSPLASIA PANEL WITH GENOME WIDE CNV ANALYSIS LYMPHATIC ANOMALIES PANEL WITH GENOME WIDE CNV ANALYSIS INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS NOONAN SYNDROME / RASOPATHY PANEL
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ERG	100%	100%	100%	99.4%	Lymphatic malformation 14, 620602	INHERITED BONE MARROW FAILURE AND/OR PREDISPOSITION PANEL TO HEMATOLOGICAL MALIGNANCIES HEMOSTATIC/THROMBOTIC DISORDERS PANEL LYMPHATIC ANOMALIES PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS
ERGIC1	100%	100%	100%	99.1%	?Arthrogryposis multiplex congenita 2, neurogenic type, 208100	MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS

ERI1	100%	100%	100%	99.4%	Hoxha-Aliu syndrome, 620662;Spondyloepimetaphyseal dysplasia, Guo-Campeau type, 620663	SHORT STATURE/SKELETAL DYSPLASIA PANEL WITH GENOME WIDE CNV ANALYSIS INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL¹
ERLIN1	100%	100%	100%	99.7%	Spastic paraplegia 62, autosomal recessive, 615681	MOVEMENT DISORDERS PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL¹

ERLIN2	100%	100%	100%	99.3%	Spastic paraplegia 18A, autosomal dominant, 620512;Spastic paraplegia 18B, autosomal recessive, 611225	MOVEMENT DISORDERS PANEL INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹
ERMARD	100%	100%	100%	99.7%	?Periventricular nodular heterotopia 6, 615544	MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS
ESAM	100%	100%	100%	98.7%	Neurodevelopmental disorder with intracranial hemorrhage, seizures, and spasticity, 620371	INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹

ESCO2	100%	100%	100%	99.3%	Juberg-Hayward syndrome, 216100;Roberts-SC phocomelia syndrome, 268300	CRANIOFACIAL ANOMALIES PANEL WITH GENOME WIDE CNV ANALYSIS SHORT STATURE/SKELETAL DYSPLASIA PANEL WITH GENOME WIDE CNV ANALYSIS INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹ OROFACIAL CLEFTING PANEL WITH GENOME WIDE CNV ANALYSIS
ESPN	100%	100%	100%	97.3%	Deafness, neurosensory, without vestibular involvement, autosomal dominant, 609006;Deafness, autosomal recessive 36, 609006;?Usher syndrome, type 1M, 618632	HEARING IMPAIRMENT PANEL (INCLUDING GJB2) MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹

ESR1	100%	100%	100%	98.9%	Breast cancer, somatic, 114480;{Migraine, susceptibility to}, 157300;Estrogen resistance, 615363;{Myocardial infarction, susceptibility to}, 608446	DISORDERS/DIFFERENCES OF SEX DEVELOPMENT (DSD) / PRIMARY ADRENAL INSUFFICIENCY PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹
ESR2	100%	100%	100%	99.3%	?Ovarian dysgenesis 8, 618187	DISORDERS/DIFFERENCES OF SEX DEVELOPMENT (DSD) / PRIMARY ADRENAL INSUFFICIENCY PANEL PREMATURE OVARIAN INSUFFICIENCY PANEL HEREDITARY CANCER PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS
ESRP1	100%	100%	100%	99.2%	?Deafness, autosomal recessive 109, 618013	HEARING IMPAIRMENT PANEL (INCLUDING GJB2) MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS

ESRRB	100%	100%	100%	99.3%	Deafness, autosomal recessive 35, 608565	HEARING IMPAIRMENT PANEL (INCLUDING GJB2) MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹
ETFA	82.4%	82.4%	100%	99.2%	Glutaric acidemia IIA, 231680	METABOLIC DISORDERS PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹
ETFB	100%	100%	100%	99.6%	Glutaric acidemia IIB, 231680	METABOLIC DISORDERS PANEL INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹

ETFDH	95.4%	92.6%	100%	99.5%	Glutaric acidemia IIC, 231680	POLYNEUROPATHIES PANEL ¹ LIVER DISORDERS PANEL METABOLIC DISORDERS PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS MITOCHONDRIAL DISORDERS PANEL COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹
ETHE1	100%	100%	100%	98.8%	Ethylmalonic encephalopathy, 602473	MOVEMENT DISORDERS PANEL EPILEPSY PANEL METABOLIC DISORDERS PANEL INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS MITOCHONDRIAL DISORDERS PANEL COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹

ETS1	100%	100%	100%	99.2%		CONGENITAL HEARTDISEASE PANEL¹ HEART DISORDERS PANEL¹ MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS
ETV6	100%	100%	100%	99%	Thrombocytopenia 5, 616216;Leukemia, acute myeloid, somatic, 601626	INHERITED BONE MARROW FAILURE AND/OR PREDISPOSITION PANEL TO HEMATOLOGICAL MALIGNANCIES HEMOSTATIC/THROMB OTIC DISORDERS PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS HEREDITARY CANCER PANEL

EVC	99.8%	98.5%	100%	98.8%	Ellis-van Creveld syndrome, 225500;?Weyers acrofacial dysostosis, 193530	CILIOPATHIES PANEL SKIN DISORDERS PANEL ¹ SHORT STATURE/SKELETAL DYSPLASIA PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹
EVC2	100%	100%	100%	99.1%	Ellis-van Creveld syndrome, 225500;Weyers acrofacial dysostosis, 193530	CILIOPATHIES PANEL SKIN DISORDERS PANEL ¹ SHORT STATURE/SKELETAL DYSPLASIA PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹

EWSR1	100%	100%	100%	96.6%	Neuroepithelioma, 612219;Ewing sarcoma, 612219	MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS
EXOC2	100%	100%	100%	99.7%	Neurodevelopmental disorder with dysmorphic facies and cerebellar hypoplasia, 619306	INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹
EXOC3L2	100%	99.9%	100%	97.5%	Brain malformation renal syndrome, 620943	MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹

EXOC6B	100%	100%	100%	98.7%	Spondyloepimetaphyseal dysplasia with joint laxity, type 3, 618395	SHORT STATURE/SKELETAL DYSPLASIA PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹
EXOC7	100%	100%	100%	98.9%	Neurodevelopmental disorder with seizures and brain atrophy, 619072	EPILEPSY PANEL INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹

EXOC8	100%	100%	100%	99.1%	?Neurodevelopmental disorder with microcephaly, seizures, and brain atrophy, 619076	CILIOPATHIES PANEL INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS
EXOSC1	100%	100%	100%	99.4%	?Pontocerebellar hypoplasia, type 1F, 619304	MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS
EXOSC2	100%	99.1%	100%	99%	Short stature, hearing loss, retinitis pigmentosa, and distinctive facies, 617763	VISION DISORDERS PANEL HEARING IMPAIRMENT PANEL (INCLUDING GJB2) INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹

EXOSC3	100%	100%	100%	99.4%	Pontocerebellar hypoplasia, type 1B, 614678	MOVEMENT DISORDERS PANEL EPILEPSY PANEL INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹
EXOSC5	100%	100%	100%	97.7%	Cerebellar ataxia, brain abnormalities, and cardiac conduction defects, 619576	MOVEMENT DISORDERS PANEL SHORT STATURE/SKELETAL DYSPLASIA PANEL WITH GENOME WIDE CNV ANALYSIS INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹

EXOSC8	100%	100%	100%	99.3%	Pontocerebellar hypoplasia, type 1C, 616081	MOVEMENT DISORDERS PANEL POLYNEUROPATHIES PANEL ¹ INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS MITOCHONDRIAL DISORDERS PANEL COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹ MUSCLE DISORDERS PANEL
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EXOSC9	100%	100%	100%	99.5%	Pontocerebellar hypoplasia, type 1D, 618065	MOVEMENT DISORDERS PANEL POLYNEUROPATHIES PANEL ¹ INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹
EXPH5	100%	100%	100%	99.4%	Epidermolysis bullosa simplex 4, localized or generalized intermediate, autosomal recessive, 615028	SKIN DISORDERS PANEL ¹ MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹

EXT1	100%	100%	100%	99.5%	Exostoses, multiple, type 1, 133700;Chondrosarcoma, 215300	SHORT STATURE/SKELETAL DYSPLASIA PANEL WITH GENOME WIDE CNV ANALYSIS METABOLIC DISORDERS PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS HEREDITARY CANCER PANEL
EXT2	100%	100%	100%	99.1%	Seizures, scoliosis, and macrocephaly syndrome, 616682;Exostoses, multiple, type 2, 133701	SHORT STATURE/SKELETAL DYSPLASIA PANEL WITH GENOME WIDE CNV ANALYSIS METABOLIC DISORDERS PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹ HEREDITARY CANCER PANEL

EXTL3	100%	99.7%	100%	99.6%	Immunoskeletal dysplasia with neurodevelopmental abnormalities, 617425	CILIOPATHIES PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL¹ PRIMARY IMMUNODEFICIENCIES PANEL SHORT STATURE/SKELETAL DYSPLASIA PANEL WITH GENOME WIDE CNV ANALYSIS INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS
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EYA1	100%	100%	100%	99.2%	Branchiootic syndrome 1, 602588;Branchiootorenal syndrome 1, with or without cataracts, 113650;Anterior segment anomalies with or without cataract, 602588;?Otofaciocervical syndrome, 166780	VISION DISORDERS PANEL CRANIOFACIAL ANOMALIES PANEL WITH GENOME WIDE CNV ANALYSIS RENAL DISORDERS PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS OROFACIAL CLEFTING PANEL WITH GENOME WIDE CNV ANALYSIS HEARING IMPAIRMENT PANEL (INCLUDING GJB2) METABOLIC DISORDERS PANEL
EYA4	100%	100%	100%	99.6%	?Cardiomyopathy, dilated, 1J, 605362;Deafness, autosomal dominant 10, 601316	MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS HEARING IMPAIRMENT PANEL (INCLUDING GJB2)

EYS	100%	100%	100%	99.7%	Retinitis pigmentosa 25, 602772	VISION DISORDERS PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL¹
EZH1	100%	100%	100%	98.8%		INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS

EZH2	100%	100%	100%	99.5%	Weaver syndrome, 277590	INHERITED BONE MARROW FAILURE AND/OR PREDISPOSITION PANEL TO HEMATOLOGICAL MALIGNANCIES MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS HEREDITARY CANCER PANEL CRANIOFACIAL ANOMALIES PANEL WITH GENOME WIDE CNV ANALYSIS TALL STATURE PANEL WITH GENOME WIDE CNV ANALYSIS SHORT STATURE/SKELETAL DYSPLASIA PANEL WITH GENOME WIDE CNV ANALYSIS INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS
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F10	100%	100%	100%	98.9%	Factor X deficiency, 227600	HEMOSTATIC/THROMBOTIC DISORDERS PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL¹
F11	100%	100%	100%	99.1%	Factor XI deficiency, autosomal dominant, 612416;Factor XI deficiency, autosomal recessive, 612416	HEMOSTATIC/THROMBOTIC DISORDERS PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL¹

F12	100%	100%	100%	99.2%	Angioedema, hereditary, 3, 610618;Factor XII deficiency, 234000	HEMOSTATIC/THROMBOTIC DISORDERS PANEL COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹ PRIMARY IMMUNODEFICIENCIES PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS
F13A1	100%	100%	100%	99.4%	Factor XIII A deficiency, 613225;{Myocardial infarction, protection against}, 608446;{Venous thrombosis, protection against}, 188050	SKIN DISORDERS PANEL ¹ HEMOSTATIC/THROMBOTIC DISORDERS PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹

F13B	100%	100%	100%	99.2%	Factor XIIIIB deficiency, 613235	HEMOSTATIC/THROMBOTIC DISORDERS PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹
F2	100%	100%	100%	98.8%	Hypoprothrombinemia, 613679;{Pregnancy loss, recurrent, susceptibility to, 2}, 614390;Dysprothrombinemia, 613679;Thrombophilia 1 due to thrombin defect, 188050;{Stroke, ischemic, susceptibility to}, 601367	HEMOSTATIC/THROMBOTIC DISORDERS PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹
F2RL3	100%	100%	100%	98.9%		HEMOSTATIC/THROMBOTIC DISORDERS PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS

F5	100%	100%	100%	99.3%	Thrombophilia 2 due to activated protein C resistance, 188055;{Pregnancy loss, recurrent, susceptibility to, 1}, 614389;{Thrombophilia, susceptibility to, due to factor V Leiden}, 188055;{Budd-Chiari syndrome}, 600880;{Stroke, ischemic, susceptibility to}, 601367;Factor V deficiency, 227400	HEMOSTATIC/THROMBOTIC DISORDERS PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹
F7	100%	100%	100%	98.8%	{Myocardial infarction, decreased susceptibility to}, 608446;Factor VII deficiency, 227500	HEMOSTATIC/THROMBOTIC DISORDERS PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹
F8	100%	100%	99.2%	72.1%	Thrombophilia 13, X-linked, due to factor VIII defect, 301071;Hemophilia A, 306700	HEMOSTATIC/THROMBOTIC DISORDERS PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS

F9	100%	100%	99.2%	71.3%	{Deep venous thrombosis, protection against}, 300807;Hemophilia B, 306900;Thrombophilia 8, X-linked, due to factor IX defect, 300807;{Warfarin sensitivity}, 301052	HEMOSTATIC/THROMBOTIC DISORDERS PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS
FA2H	100%	100%	100%	98.8%	Spastic paraplegia 35, autosomal recessive, 612319	MOVEMENT DISORDERS PANEL VISION DISORDERS PANEL EPILEPSY PANEL METABOLIC DISORDERS PANEL INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹ MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS MITOCHONDRIAL DISORDERS PANEL

FAAH	100%	100%	100%	99.2%	{Drug addiction, susceptibility to}, 606581	NEUROLOGICAL PAIN DISORDERS PANEL' MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS
FAAP100	100%	100%	100%	99.3%		INHERITED BONE MARROW FAILURE AND/OR PREDISPOSITION PANEL TO HEMATOLOGICAL MALIGNANCIES MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS HEREDITARY CANCER PANEL
FAAP24	100%	100%	100%	99.7%		PRIMARY IMMUNODEFICIENCIES PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS

FADD	100%	100%	100%	98.7%	Immunodeficiency 90 with encephalopathy, functional hyposplenia, and hepatic dysfunction, 613759	PRIMARY IMMUNODEFICIENCIES PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹
FAH	100%	100%	100%	99.7%	Tyrosinemia, type I, 276700	HEART DISORDERS PANEL ¹ LIVER DISORDERS PANEL METABOLIC DISORDERS PANEL RENAL DISORDERS PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹

FAM111A	100%	100%	100%	99.8%	Kenny-Caffey syndrome, type 2, 127000;Gracile bone dysplasia, 602361	SHORT STATURE/SKELETAL DYSPLASIA PANEL WITH GENOME WIDE CNV ANALYSIS RENAL DISORDERS PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS
FAM111B	100%	100%	100%	99%	Poikiloderma, hereditary fibrosing, with tendon contractures, myopathy, and pulmonary fibrosis, 615704	SKIN DISORDERS PANEL ¹ MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS MUSCLE DISORDERS PANEL
FAM149B1	100%	100%	100%	99.4%	Joubert syndrome 36, 618763	CILIOPATHIES PANEL INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹

FAM161A	100%	100%	100%	99.3%	Retinitis pigmentosa 28, 606068	VISION DISORDERS PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹
FAM177A1	86.5%	86.5%	100%	99.1%		INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS
FAM20A	100%	100%	100%	99%	Amelogenesis imperfecta, type IG (enamel-renal syndrome), 204690	CRANIOFACIAL ANOMALIES PANEL WITH GENOME WIDE CNV ANALYSIS SKIN DISORDERS PANEL ¹ RENAL DISORDERS PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹

FAM20B	100%	100%	100%	99.4%		SHORT STATURE/SKELETAL DYSPLASIA PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS
FAM20C	100%	100%	100%	98.4%	Raine syndrome, 259775	SKIN DISORDERS PANEL ¹ SHORT STATURE/SKELETAL DYSPLASIA PANEL WITH GENOME WIDE CNV ANALYSIS INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹ OROFACIAL CLEFTING PANEL WITH GENOME WIDE CNV ANALYSIS

FAM50A	100%	99.3%	97.8%	65.4%	Intellectual developmental disorder, X-linked syndromic, Armfield type, 300261	INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS
FAM83G	100%	100%	100%	98.3%		SKIN DISORDERS PANEL ¹ MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS
FAM83H	100%	100%	100%	99.2%	Amelogenesis imperfecta, type IIIA, 130900	CRANIOFACIAL ANOMALIES PANEL WITH GENOME WIDE CNV ANALYSIS SKIN DISORDERS PANEL ¹ MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS

FAN1	100%	100%	100%	98.9%	Interstitial nephritis, karyomegalic, 614817	RENAL DISORDERS PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹
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FANCA	100%	100%	100%	99%	Fanconi anemia, complementation group A, 227650	INHERITED BONE MARROW FAILURE AND/OR PREDISPOSITION PANEL TO HEMATOLOGICAL MALIGNANCIES SKIN DISORDERS PANEL ¹ SHORT STATURE/SKELETAL DYSPLASIA PANEL WITH GENOME WIDE CNV ANALYSIS MALE INFERTILITY PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹ HEREDITARY CANCER PANEL
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FANCB	96.2%	96.1%	98.7%	69.8%	Fanconi anemia, complementation group B, 300514	INHERITED BONE MARROW FAILURE AND/OR PREDISPOSITION PANEL TO HEMATOLOGICAL MALIGNANCIES SKIN DISORDERS PANEL ¹ SHORT STATURE/SKELETAL DYSPLASIA PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS HEREDITARY CANCER PANEL
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FANCC	100%	100%	100%	99.3%	Fanconi anemia, complementation group C, 227645	INHERITED BONE MARROW FAILURE AND/OR PREDISPOSITION PANEL TO HEMATOLOGICAL MALIGNANCIES SKIN DISORDERS PANEL ¹ SHORT STATURE/SKELETAL DYSPLASIA PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹ HEREDITARY CANCER PANEL
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FANCD2	100%	100%	100%	99.3%	Fanconi anemia, complementation group D2, 227646	INHERITED BONE MARROW FAILURE AND/OR PREDISPOSITION PANEL TO HEMATOLOGICAL MALIGNANCIES SKIN DISORDERS PANEL ¹ SHORT STATURE/SKELETAL DYSPLASIA PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹ HEREDITARY CANCER PANEL
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FANCE	100%	100%	100%	98.9%	Fanconi anemia, complementation group E, 600901	INHERITED BONE MARROW FAILURE AND/OR PREDISPOSITION PANEL TO HEMATOLOGICAL MALIGNANCIES SKIN DISORDERS PANEL ¹ SHORT STATURE/SKELETAL DYSPLASIA PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹ HEREDITARY CANCER PANEL
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FANCF	100%	100%	100%	99.6%	Fanconi anemia, complementation group F, 603467	INHERITED BONE MARROW FAILURE AND/OR PREDISPOSITION PANEL TO HEMATOLOGICAL MALIGNANCIES SKIN DISORDERS PANEL ¹ SHORT STATURE/SKELETAL DYSPLASIA PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹ HEREDITARY CANCER PANEL
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FANCG	100%	100%	100%	98.9%	Fanconi anemia, complementation group G, 614082	INHERITED BONE MARROW FAILURE AND/OR PREDISPOSITION PANEL TO HEMATOLOGICAL MALIGNANCIES SKIN DISORDERS PANEL ¹ SHORT STATURE/SKELETAL DYSPLASIA PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹ HEREDITARY CANCER PANEL
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FANCI	100%	100%	100%	99.5%	Fanconi anemia, complementation group I, 609053	INHERITED BONE MARROW FAILURE AND/OR PREDISPOSITION PANEL TO HEMATOLOGICAL MALIGNANCIES SKIN DISORDERS PANEL ¹ SHORT STATURE/SKELETAL DYSPLASIA PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹ HEREDITARY CANCER PANEL
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FANCL	91.1%	88.6%	99.8%	96.6%	Fanconi anemia, complementation group L, 614083	INHERITED BONE MARROW FAILURE AND/OR PREDISPOSITION PANEL TO HEMATOLOGICAL MALIGNANCIES SKIN DISORDERS PANEL ¹ SHORT STATURE/SKELETAL DYSPLASIA PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹ HEREDITARY CANCER PANEL
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FANCM	100%	100%	100%	99.6%	Premature ovarian failure 15, 618096;Spermatogenic failure 28, 618086	INHERITED BONE MARROW FAILURE AND/OR PREDISPOSITION PANEL TO HEMATOLOGICAL MALIGNANCIES SKIN DISORDERS PANEL ¹ DISORDERS/DIFFERENCES OF SEX DEVELOPMENT (DSD) / PRIMARY ADRENAL INSUFFICIENCY PANEL MALE INFERTILITY PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS PREMATURE OVARIAN INSUFFICIENCY PANEL HEREDITARY CANCER PANEL
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FAR1	100%	100%	100%	99.4%	Peroxisomal fatty acyl-CoA reductase 1 disorder, 616154;Cataracts, spastic paraparesis, and speech delay, 619338	MOVEMENT DISORDERS PANEL SHORT STATURE/SKELETAL DYSPLASIA PANEL WITH GENOME WIDE CNV ANALYSIS METABOLIC DISORDERS PANEL INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL¹
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FARS2	100%	100%	100%	99.1%	Combined oxidative phosphorylation deficiency 14, 614946;Spastic paraplegia 77, autosomal recessive, 617046	MOVEMENT DISORDERS PANEL EPILEPSY PANEL INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS MITOCHONDRIAL DISORDERS PANEL COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL¹
FARSA	100%	100%	100%	99.1%	?Rajab interstitial lung disease with brain calcifications 2, 619013	INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS

FARSB	100%	100%	100%	99.5%	Rajab interstitial lung disease with brain calcifications 1, 613658	LIVER DISORDERS PANEL INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS MITOCHONDRIAL DISORDERS PANEL COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹
FAS	100%	99.9%	100%	99.5%	Squamous cell carcinoma, burn scar-related, somatic;Autoimmune lymphoproliferative syndrome, type IA, 601859;{Autoimmune lymphoproliferative syndrome}, 601859	INHERITED BONE MARROW FAILURE AND/OR PREDISPOSITION PANEL TO HEMATOLOGICAL MALIGNANCIES PRIMARY IMMUNODEFICIENCIES PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS HEREDITARY CANCER PANEL

FASLG	100%	100%	100%	99.2%	Autoimmune lymphoproliferative syndrome, type IB, 601859;{Lung cancer, susceptibility to}, 211980	INHERITED BONE MARROW FAILURE AND/OR PREDISPOSITION PANEL TO HEMATOLOGICAL MALIGNANCIES PRIMARY IMMUNODEFICIENCIES PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS HEREDITARY CANCER PANEL
FASTKD2	100%	100%	100%	99.5%	Combined oxidative phosphorylation deficiency 44, 618855	INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS MITOCHONDRIAL DISORDERS PANEL COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹

FASTKD5	100%	100%	100%	99.7%		MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS MITOCHONDRIAL DISORDERS PANEL
FAT1	100%	100%	100%	99.4%		RENAL DISORDERS PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS
FAT2	100%	100%	100%	99.2%	Spinocerebellar ataxia 45, 617769	MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS

FAT4	100%	100%	100%	99.2%	Van Maldergem syndrome 2, 615546;Hennekam lymphangiectasia-lymphedema syndrome 2, 616006	SKIN DISORDERS PANEL ¹ PRIMARY IMMUNODEFICIENCIES PANEL LYMPHATIC ANOMALIES PANEL WITH GENOME WIDE CNV ANALYSIS INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹
FBLN1	100%	100%	100%	98.4%	Synpolydactyly, 3/3'4, associated with metacarpal and metatarsal synostoses, 608180	SHORT STATURE/SKELETAL DYSPLASIA PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS

FBLN5	92.8%	92.8%	100%	99.3%	Cutis laxa, autosomal recessive, type 1A, 219100;Charcot-Marie-Tooth disease, demyelinating, type 1H, 619764;Macular degeneration, age-related, 3, 608895;?Cutis laxa, autosomal dominant 2, 614434	SKIN DISORDERS PANEL ¹ POLYNEUROPATHIES PANEL ¹ NEUROLOGICAL PAIN DISORDERS PANEL ¹ MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹
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FBN1	100%	100%	100%	99.5%	Geleophysic dysplasia 2, 614185;Weill-Marchesani syndrome 2, dominant, 608328;Ectopia lentis, familial, 129600;MASS syndrome, 604308;Marfan lipodystrophy syndrome, 616914;Acromicric dysplasia, 102370;Marfan syndrome, 154700;Stiff skin syndrome, 184900	THORACIC AORTIC ANEURYSM AND/OR DISSECTION PANEL VISION DISORDERS PANEL CONGENITAL HEARTDISEASE PANEL ¹ TALL STATURE PANEL WITH GENOME WIDE CNV ANALYSIS HEART DISORDERS PANEL ¹ HEMOSTATIC/THROMBOTIC DISORDERS PANEL SHORT STATURE/SKELETAL DYSPLASIA PANEL WITH GENOME WIDE CNV ANALYSIS METABOLIC DISORDERS PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS
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FBN2	99.2%	99.2%	100%	99.4%	Macular degeneration, early-onset, 616118;Contractural arachnodactyly, congenital, 121050	FETAL AKINESIA PANEL THORACIC AORTIC ANEURYSM AND/OR DISSECTION PANEL VISION DISORDERS PANEL CONGENITAL HEARTDISEASE PANEL¹ TALL STATURE PANEL WITH GENOME WIDE CNV ANALYSIS HEART DISORDERS PANEL¹ POLYNEUROPATHIES PANEL¹ NEUROLOGICAL PAIN DISORDERS PANEL¹ SHORT STATURE/SKELETAL DYSPLASIA PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS
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FBP1	100%	100%	100%	99.1%	Fructose-1,6-bisphosphatase deficiency, 229700	METABOLIC DISORDERS PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹
FBP2	100%	100%	100%	99.1%	?Leukodystrophy, childhood-onset, remitting, 619864	METABOLIC DISORDERS PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS
FBRSL1	97.8%	94.5%	100%	90.7%		INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS

FBXL3	100%	100%	100%	99.9%	Intellectual developmental disorder with short stature, facial anomalies, and speech defects, 606220	INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹
FBXL4	100%	100%	100%	99.5%	Mitochondrial DNA depletion syndrome 13 (encephalomyopathic type), 615471	INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS MITOCHONDRIAL DISORDERS PANEL COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹
FBXO11	100%	100%	100%	98.9%	Intellectual developmental disorder with dysmorphic facies and behavioral abnormalities, 618089	INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS

FBXO28	100%	100%	100%	99.7%	Developmental and epileptic encephalopathy 100, 619777	EPILEPSY PANEL INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS
FBXO31	100%	100%	100%	98%	?Intellectual developmental disorder, autosomal recessive 45, 615979	INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹
FBXO32	100%	100%	100%	99.7%		HEART DISORDERS PANEL ¹ MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS

FBXO38	100%	100%	100%	99.7%	Neuronopathy, distal hereditary motor, autosomal dominant 6, 615575	POLYNEUROPATHIES PANEL ¹ MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS
FBXO43	100%	100%	100%	99.6%	Spermatogenic failure 64, 619696;Oocyte/zygote/embryo maturation arrest 12, 619697	UNSUCCESSFUL IVF TREATMENTS, RECURRENT MOLAR PREGNANCIES AND/OR RECURRENT PREGNANCY LOSS PANEL ¹ MALE INFERTILITY PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS
FBXO7	100%	100%	100%	99.5%	Parkinson disease 15, autosomal recessive, 260300	MOVEMENT DISORDERS PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS MITOCHONDRIAL DISORDERS PANEL PARKINSON DISEASE PANEL COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹

FBXW11	100%	100%	100%	99%	Neurodevelopmental, jaw, eye, and digital syndrome, 618914	VISION DISORDERS PANEL PRIMARY IMMUNODEFICIENCIES PANEL INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS
FBXW4	100%	100%	100%	98.7%		SHORT STATURE/SKELETAL DYSPLASIA PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS
FBXW7	100%	100%	100%	99.5%	Developmental delay, hypotonia, and impaired language, 620012	INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS HEREDITARY CANCER PANEL

FCGR3A	99.6%	96.1%	99.5%	95.4%	Immunodeficiency 20, 615707	MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL¹ PRIMARY IMMUNODEFICIENCIES PANEL
FCGR3B	99.4%	97.7%	95.8%	82.6%		MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS
FCHO1	98.5%	96.1%	100%	98.3%	Immunodeficiency 76, 619164	PRIMARY IMMUNODEFICIENCIES PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL¹ SEVERE COMBINED IMMUNODEFICIENCY (SCID) PANEL

FCN3	100%	99.9%	100%	99.2%	Immunodeficiency due to ficolin 3 deficiency, 613860	PRIMARY IMMUNODEFICIENCIES PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL¹
FCSK	100%	100%	100%	99.2%	Congenital disorder of glycosylation with defective fucosylation 2, 618324	EPILEPSY PANEL METABOLIC DISORDERS PANEL INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL¹

FDFT1	100%	100%	100%	98.6%	Squalene synthase deficiency, 618156	SKIN DISORDERS PANEL ¹ METABOLIC DISORDERS PANEL INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹
FDPS	100%	100%	100%	98.9%	Porokeratosis 9, multiple types, 616631	SKIN DISORDERS PANEL ¹ MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS
FDX2	99.9%	99.7%	100%	96.9%	Mitochondrial myopathy, episodic, with optic atrophy and reversible leukoencephalopathy, 251900	MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS MITOCHONDRIAL DISORDERS PANEL COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹ MUSCLE DISORDERS PANEL

FDXR	100%	100%	100%	99%	Multiple mitochondrial dysfunctions syndrome 9B, 620887;Auditory neuropathy and optic atrophy, 617717	VISION DISORDERS PANEL HEARING IMPAIRMENT PANEL (INCLUDING GJB2) MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS MITOCHONDRIAL DISORDERS PANEL COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹
FECH	100%	100%	100%	99.6%	Protoporphyrria, erythropoietic, 1, 177000	MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS LIVER DISORDERS PANEL METABOLIC DISORDERS PANEL SKIN DISORDERS PANEL ¹ IRON DISORDERS PANEL COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹

FEM1B	100%	100%	100%	99.8%		INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS
FERMT1	100%	100%	100%	99.4%	Kindler syndrome, 173650	PRIMARY IMMUNODEFICIENCIES PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL¹ SKIN DISORDERS PANEL¹

FERMT3	100%	100%	100%	99.1%	Leukocyte adhesion deficiency, type III, 612840	MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS HEMOSTATIC/THROMB OTIC DISORDERS PANEL PRIMARY IMMUNODEFICIENCIES PANEL SHORT STATURE/SKELETAL DYSPLASIA PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL¹
FEZF1	100%	100%	100%	98.7%	Hypogonadotropic hypogonadism 22, with or without anosmia, 616030	DISORDERS/DIFFEREN CES OF SEX DEVELOPMENT (DSD) / PRIMARY ADRENAL INSUFFICIENCY PANEL HYPOGONADOTROPIC HYPOGONADISM (KALLMANN) PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL¹

FEZF2	100%	100%	100%	97.7%		INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS
FGA	100%	100%	100%	99.4%	Amyloidosis, hereditary systemic 2, 105200;Hypodysfibrinogenemia, congenital, 616004;Dysfibrinogenemia, congenital, 616004;Afibrinogenemia, congenital, 202400	HEMOSTATIC/THROMB OTIC DISORDERS PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹
FGB	100%	100%	100%	99.6%	Hypofibrinogenemia, congenital, 202400;Dysfibrinogenemia, congenital, 616004;Afibrinogenemia, congenital, 202400	HEMOSTATIC/THROMB OTIC DISORDERS PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹

FGD1	100%	99%	98.8%	67.6%	Intellectual developmental disorder, X-linked syndromic 16, 305400;Aarskog-Scott syndrome, 305400	CRANIOFACIAL ANOMALIES PANEL WITH GENOME WIDE CNV ANALYSIS EPILEPSY PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS OROFACIAL CLEFTING PANEL WITH GENOME WIDE CNV ANALYSIS SHORT STATURE/SKELETAL DYSPLASIA PANEL WITH GENOME WIDE CNV ANALYSIS INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS
FGD4	100%	100%	100%	99.2%	Charcot-Marie-Tooth disease, type 4H, 609311	POLYNEUROPATHIES PANEL ¹ MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹

FGF10	100%	100%	100%	99.8%	LADD syndrome 3, 620193;Aplasia of lacrimal and salivary glands, 180920	CRANIOFACIAL ANOMALIES PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS SKIN DISORDERS PANEL ¹ SHORT STATURE/SKELETAL DYSPLASIA PANEL WITH GENOME WIDE CNV ANALYSIS
FGF12	100%	100%	100%	98.9%	Developmental and epileptic encephalopathy 47, 617166	EPILEPSY PANEL HEART DISORDERS PANEL ¹ INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS

FGF13	100%	99.9%	99%	72.3%	Developmental and epileptic encephalopathy 90, 301058;Intellectual developmental disorder, X-linked 110, 301095	EPILEPSY PANEL INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS
FGF14	100%	100%	100%	99.4%	Spinocerebellar ataxia 27A, 193003;Spinocerebellar ataxia 27B, late-onset, 620174	MOVEMENT DISORDERS PANEL INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS
FGF16	100%	99.8%	98.6%	66.1%	Metacarpal 4-5 fusion, 309630	SHORT STATURE/SKELETAL DYSPLASIA PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS

FGF17	100%	100%	100%	98.9%	Hypogonadotropic hypogonadism 20 with or without anosmia, 615270	DISORDERS/DIFFERENCES OF SEX DEVELOPMENT (DSD) / PRIMARY ADRENAL INSUFFICIENCY PANEL HYPOGONADOTROPIC HYPOGONADISM (KALLMANN) PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS
FGF20	100%	100%	100%	98.7%	?Renal hypodysplasia/aplasia 2, 615721	MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹

FGF23	100%	100%	100%	97.5%	Tumoral calcinosis, hyperphosphatemic, familial, 2, 617993;Hypophosphatemic rickets, autosomal dominant, 193100	COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹ MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS RENAL DISORDERS PANEL SHORT STATURE/SKELETAL DYSPLASIA PANEL WITH GENOME WIDE CNV ANALYSIS SKIN DISORDERS PANEL ¹
FGF3	100%	100%	100%	95.9%	Deafness, congenital with inner ear agenesis, microtia, and microdontia, 610706	MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹ HEARING IMPAIRMENT PANEL (INCLUDING GJB2) CRANIOFACIAL ANOMALIES PANEL WITH GENOME WIDE CNV ANALYSIS SKIN DISORDERS PANEL ¹

FGF5	100%	100%	100%	99.7%	Trichomegaly, 190330	SKIN DISORDERS PANEL ¹ MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS
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FGF8	100%	100%	100%	97.5%	Hypogonadotropic hypogonadism 6 with or without anosmia, 612702	OROFACIAL CLEFTING PANEL WITH GENOME WIDE CNV ANALYSIS DISORDERS/DIFFEREN CES OF SEX DEVELOPMENT (DSD) / PRIMARY ADRENAL INSUFFICIENCY PANEL HEART DISORDERS PANEL ¹ HYPOGONADOTROPIC HYPOGONADISM (KALLMANN) PANEL SHORT STATURE/SKELETAL DYSPLASIA PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS CRANIOFACIAL ANOMALIES PANEL WITH GENOME WIDE CNV ANALYSIS CONGENITAL HEARTDISEASE PANEL ¹ SKIN DISORDERS PANEL ¹
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FGF9	100%	100%	100%	99.5%	Multiple synostoses syndrome 3, 612961	CRANIOFACIAL ANOMALIES PANEL WITH GENOME WIDE CNV ANALYSIS SHORT STATURE/SKELETAL DYSPLASIA PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS
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FGFR1	99.9%	98.9%	100%	99.2%	Pfeiffer syndrome, 101600;Hypogonadotropic hypogonadism 2 with or without anosmia, 147950;Jackson-Weiss syndrome, 123150;Hartsfield syndrome, 615465;Trigonocephaly 1, 190440;Osteoglophonic dysplasia, 166250;Encephalocraniocutaneous lipomatosis, somatic mosaic, 613001	CRANIOFACIAL ANOMALIES PANEL WITH GENOME WIDE CNV ANALYSIS INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS SHORT STATURE/SKELETAL DYSPLASIA PANEL WITH GENOME WIDE CNV ANALYSIS HYPOGONADOTROPIC HYPOGONADISM (KALLMANN) PANEL SKIN DISORDERS PANEL ¹ DISORDERS/DIFFERENCES OF SEX DEVELOPMENT (DSD) / PRIMARY ADRENAL INSUFFICIENCY PANEL OROFACIAL CLEFTING PANEL WITH GENOME WIDE CNV ANALYSIS
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FGFR2	100%	100%	100%	99%	<p>Bent bone dysplasia syndrome, 614592;LADD syndrome 1, 149730;Antley-Bixler syndrome without genital anomalies or disordered steroidogenesis, 207410;Scaphocephaly and Axenfeld-Rieger anomaly;Jackson-Weiss syndrome, 123150;Gastric cancer, somatic, 613659;Craniofacial-skeletal-dermatologic dysplasia, 101600;Apert syndrome, 101200;Pfeiffer syndrome, 101600;Craniosynostosis, nonspecific;?Scaphocephaly, maxillary retrusion, and impaired intellectual development, 609579;Beare-Stevenson cutis gyrata syndrome, 123790;Crouzon syndrome, 123500;Saethre-Chotzen syndrome, 101400</p>	<p>CRANIOFACIAL ANOMALIES PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS OROFACIAL CLEFTING PANEL WITH GENOME WIDE CNV ANALYSIS SKIN DISORDERS PANEL¹ DISORDERS/DIFFERENCES OF SEX DEVELOPMENT (DSD) / PRIMARY ADRENAL INSUFFICIENCY PANEL SHORT STATURE/SKELETAL DYSPLASIA PANEL WITH GENOME WIDE CNV ANALYSIS INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS</p>
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FGFR3	100%	100%	100%	98.4%	Muenke syndrome, 602849;SADDAN, 616482;Hypochondroplasia, 146000;Thanatophoric dysplasia, type II, 187601;Nevus, epidermal, somatic, 162900;CATSHL syndrome, 610474;Thanatophoric dysplasia, type I, 187600;Spermatocytic seminoma, somatic, 273300;Bladder cancer, somatic, 109800;LADD syndrome 2, 620192;Achondroplasia, 100800;Cervical cancer, somatic, 603956;Colorectal cancer, somatic, 114500;Crouzon syndrome with acanthosis nigricans, 612247	CRANIOFACIAL ANOMALIES PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS SHORT STATURE/SKELETAL DYSPLASIA PANEL WITH GENOME WIDE CNV ANALYSIS INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS HEARING IMPAIRMENT PANEL (INCLUDING GJB2) TALL STATURE PANEL WITH GENOME WIDE CNV ANALYSIS SKIN DISORDERS PANEL ¹
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FGG	100%	100%	100%	99.8%	Dysfibrinogenemia, congenital, 616004;Hypodysfibrinogenemia, 616004;Hypofibrinogenemia, congenital, 202400;Afibrinogenemia, congenital, 202400	HEMOSTATIC/THROMBOTIC DISORDERS PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹
FH	100%	100%	100%	99.3%	Leiomyomatosis and renal cell cancer, 150800;Fumarase deficiency, 606812	SKIN DISORDERS PANEL ¹ PANEL HEREDITARY RENALCANCER HEREDITARY CANCER PANEL MITOCHONDRIAL DISORDERS PANEL COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹ METABOLIC DISORDERS PANEL INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS LIVER DISORDERS PANEL

FHL1	100%	99.7%	99.2%	67.2%	Myopathy, X-linked, with postural muscle atrophy, 300696;Emery-Dreifuss muscular dystrophy 6, X-linked, 300696;?Uruguay faciocardiomusculoskeletal syndrome, 300280;Scapuloperoneal myopathy, X-linked dominant, 300695;Reducing body myopathy, X-linked 1b, with late childhood or adult onset, 300718;Reducing body myopathy, X-linked 1a, severe, infantile or early childhood onset, 300717	MUSCLE DISORDERS PANEL HEART DISORDERS PANEL ¹ HYPERTROPHIC CARDIOMYOPATHY PANEL ¹ MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS
FHL2	100%	100%	100%	99.6%		HEART DISORDERS PANEL ¹ MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS
FHOD3	100%	100%	100%	98.7%	Cardiomyopathy, familial hypertrophic, 28, 619402	HEART DISORDERS PANEL ¹ HYPERTROPHIC CARDIOMYOPATHY PANEL ¹ MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS

FIBP	100%	100%	100%	98.5%	Thauvin-Robinet-Faivre syndrome, 617107	TALL STATURE PANEL WITH GENOME WIDE CNV ANALYSIS INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL¹
FICD	100%	100%	100%	99.2%	Spastic paraplegia 92, autosomal recessive, 620911	MOVEMENT DISORDERS PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS

FIG4	98.4%	98.4%	100%	99.7%	Yunis-Varon syndrome, 216340;?Polymicrogyria, bilateral temporooccipital, 612691;Amyotrophic lateral sclerosis 11, 612577;Charcot-Marie-Tooth disease, type 4J, 611228	AMYOTROPHIC LATERAL SCLEROSIS, ALS PANEL POLYNEUROPATHIES PANEL ¹ SHORT STATURE/SKELETAL DYSPLASIA PANEL WITH GENOME WIDE CNV ANALYSIS INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹
FIGLA	100%	100%	100%	99.5%	Premature ovarian failure 6, 612310	DISORDERS/DIFFEREN CES OF SEX DEVELOPMENT (DSD) / PRIMARY ADRENAL INSUFFICIENCY PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS PREMATURE OVARIAN INSUFFICIENCY PANEL

FIGN	100%	100%	100%	99%		INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS
FIGNL1	100%	100%	100%	99.8%		DISORDERS/DIFFEREN CES OF SEX DEVELOPMENT (DSD) / PRIMARY ADRENAL INSUFFICIENCY PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS PREMATURE OVARIAN INSUFFICIENCY PANEL
FILIP1	100%	100%	100%	99.5%	Neuromuscular disorder, congenital, with dysmorphic facies, 620775	INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL¹

FITM2	100%	100%	100%	98.8%	Siddiqi syndrome, 618635	HEARING IMPAIRMENT PANEL (INCLUDING GJB2) INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹
FKBP10	100%	100%	100%	98.3%	Osteogenesis imperfecta, type XI, 610968;Bruck syndrome 1, 259450	SKIN DISORDERS PANEL ¹ SHORT STATURE/SKELETAL DYSPLASIA PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹

FKBP14	100%	100%	100%	99.7%	Ehlers-Danlos syndrome, kyphoscoliotic type, 2, 614557	THORACIC AORTIC ANEURYSM AND/OR DISSECTION PANEL SKIN DISORDERS PANEL ¹ SHORT STATURE/SKELETAL DYSPLASIA PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹ MUSCLE DISORDERS PANEL
FKBP6	100%	100%	100%	99.4%	Spermatogenic failure 77, 620103	MALE INFERTILITY PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS

FKRP	100%	100%	100%	98.6%	Muscular dystrophy-dystroglycanopathy (congenital with or without impaired intellectual development), type B, 5, 606612;Muscular dystrophy-dystroglycanopathy (limb-girdle), type C, 5, 607155;Muscular dystrophy-dystroglycanopathy (congenital with brain and eye anomalies), type A, 5, 613153	FETAL AKINESIA PANEL HEART DISORDERS PANEL ¹ METABOLIC DISORDERS PANEL INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹ MUSCLE DISORDERS PANEL
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FKTN	100%	100%	99.9%	99.5%	Muscular dystrophy-dystroglycanopathy (limb-girdle), type C, 4, 611588;Muscular dystrophy-dystroglycanopathy (congenital with brain and eye anomalies), type A, 4, 253800;Cardiomyopathy, dilated, 1X, 611615;Muscular dystrophy-dystroglycanopathy (congenital without impaired intellectual development), type B, 4, 613152	HEART DISORDERS PANEL ¹ METABOLIC DISORDERS PANEL INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹ MUSCLE DISORDERS PANEL
FLAD1	100%	100%	100%	99%	Lipid storage myopathy due to flavin adenine dinucleotide synthetase deficiency, 255100	COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹ MUSCLE DISORDERS PANEL METABOLIC DISORDERS PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS MITOCHONDRIAL DISORDERS PANEL

FLCN	100%	100%	100%	98.9%	Birt-Hogg-Dube syndrome, 135150;Colorectal cancer, somatic, 114500;Pneumothorax, primary spontaneous, 173600;Renal carcinoma, chromophobe, somatic, 144700	SKIN DISORDERS PANEL ¹ MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS PANEL HEREDITARY RENALCANCER HEREDITARY CANCER PANEL
FLG	100%	100%	100%	98.9%	Ichthyosis vulgaris, 146700;{Dermatitis, atopic, susceptibility to, 2}, 605803	SKIN DISORDERS PANEL ¹ MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS
FLG2	100%	100%	99.9%	98.8%	Peeling skin syndrome 6, 618084	SKIN DISORDERS PANEL ¹ MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS
FLI1	97%	97%	100%	99.3%	Bleeding disorder, platelet-type, 21, 617443	HEMOSTATIC/THROMB OTIC DISORDERS PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS

FLII	100%	100%	99.9%	97.6%	Cardiomyopathy, dilated, 2J, 620635	HEART DISORDERS PANEL ¹ MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS
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FLNA	100%	99.7%	98.4%	66.4%	Otopalatodigital syndrome, type II, 304120;Intestinal pseudoobstruction, neuronal, 300048;Cardiac valvular dysplasia, X-linked, 314400;?FG syndrome 2, 300321;Melnick-Needles syndrome, 309350;Terminal osseous dysplasia, 300244;Congenital short bowel syndrome, 300048;Otopalatodigital syndrome, type I, 311300;Heterotopia, periventricular, 1, 300049;Frontometaphyseal dysplasia 1, 305620	THORACIC AORTIC ANEURYSM AND/OR DISSECTION PANEL CRANIOFACIAL ANOMALIES PANEL WITH GENOME WIDE CNV ANALYSIS CONGENITAL HEARTDISEASE PANEL ¹ EPILEPSY PANEL OROFACIAL CLEFTING PANEL WITH GENOME WIDE CNV ANALYSIS INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS HEART DISORDERS PANEL ¹ HEMOSTATIC/THROMBOTIC DISORDERS PANEL SHORT STATURE/SKELETAL DYSPLASIA PANEL WITH GENOME WIDE CNV ANALYSIS LIVER DISORDERS PANEL
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FLNB	100%	100%	100%	99.2%	Larsen syndrome, 150250;Atelosteogenesis, type I, 108720;Atelosteogenesis, type III, 108721;Spondylocarpotarsal synostosis syndrome, 272460;Boomerang dysplasia, 112310	CRANIOFACIAL ANOMALIES PANEL WITH GENOME WIDE CNV ANALYSIS SHORT STATURE/SKELETAL DYSPLASIA PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL¹ OROFACIAL CLEFTING PANEL WITH GENOME WIDE CNV ANALYSIS
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FLNC	100%	100%	100%	99%	Cardiomyopathy, familial hypertrophic, 26, 617047;Arrhythmogenic right ventricular dysplasia, familial, 617047;Cardiomyopathy, familial restrictive 5, 617047;Myopathy, distal, 4, 614065;Myopathy, myofibrillar, 5, 609524	ARITMOGENE CARDIOMYOPATHY PANEL ¹ FETAL AKINESIA PANEL DILATED CARDIOMYOPATHY PANEL ¹ HEART DISORDERS PANEL ¹ HYPERTROPHIC CARDIOMYOPATHY PANEL ¹ MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS ARRHYTHMIA AND CARDIAC CONDUCTION DISORDERS PANEL ¹ MUSCLE DISORDERS PANEL
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FLRT3	100%	100%	100%	99.7%	Hypogonadotropic hypogonadism 21 with anosmia, 615271	DISORDERS/DIFFERENCES OF SEX DEVELOPMENT (DSD) / PRIMARY ADRENAL INSUFFICIENCY PANEL HYPOGONADOTROPIC HYPOGONADISM (KALLMANN) PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS
FLT3	100%	100%	100%	99.4%	Leukemia, acute lymphoblastic, somatic, 613065;Leukemia, acute myeloid, reduced survival in, somatic, 601626;Leukemia, acute myeloid, somatic, 601626	MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS
FLT3LG	100%	100%	100%	98%	?Immunodeficiency 125, 620926	PRIMARY IMMUNODEFICIENCIES PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS

FLT4	100%	100%	100%	98.8%	Hemangioma, capillary infantile, somatic, 602089;Lymphatic malformation 1, 153100;Congenital heart defects, multiple types, 7, 618780	CONGENITAL HEARTDISEASE PANEL¹ SKIN DISORDERS PANEL¹ HEART DISORDERS PANEL¹ LYMPHATIC ANOMALIES PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS
FLVCR1	100%	100%	100%	99.4%	Ataxia, posterior column, with retinitis pigmentosa, 609033	MOVEMENT DISORDERS PANEL COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL¹ VISION DISORDERS PANEL POLYNEUROPATHIES PANEL¹ NEUROLOGICAL PAIN DISORDERS PANEL¹ INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS

FLVCR2	100%	100%	100%	99.4%	Proliferative vasculopathy and hydranencephaly-hydrocephaly syndrome, 225790	FETAL AKINESIA PANEL INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹
FMN1	100%	100%	100%	98.4%		SHORT STATURE/SKELETAL DYSPLASIA PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS

FMN2	100%	100%	99.8%	94.1%	Intellectual developmental disorder, autosomal recessive 47, 616193	INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹
FMO3	100%	100%	100%	99.3%	Trimethylaminuria, 602079	METABOLIC DISORDERS PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹
FMR1	100%	99.8%	98.7%	70%	Fragile X tremor/ataxia syndrome, 300623;Fragile X syndrome, 300624;Premature ovarian failure 1, 311360	INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS

FN1	100%	100%	100%	99.2%	Spondylometaphyseal dysplasia, corner fracture type, 184255;Glomerulopathy with fibronectin deposits 2, 601894	SHORT STATURE/SKELETAL DYSPLASIA PANEL WITH GENOME WIDE CNV ANALYSIS RENAL DISORDERS PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS
FNIP1	100%	100%	100%	99.3%	Immunodeficiency 93 and hypertrophic cardiomyopathy, 619705	SKIN DISORDERS PANEL ¹ HEART DISORDERS PANEL ¹ PRIMARY IMMUNODEFICIENCIES PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹

FOCAD	100%	100%	100%	99.3%	Liver disease, severe congenital, 619991	LIVER DISORDERS PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹
FOLR1	100%	100%	100%	99%	Neurodegeneration due to cerebral folate transport deficiency, 613068	MOVEMENT DISORDERS PANEL EPILEPSY PANEL METABOLIC DISORDERS PANEL INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹

FOSL2	100%	100%	100%	98.2%	Aplasia cutis-enamel dysplasia syndrome, 620789	INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS
FOXC1	100%	99.7%	100%	92.7%	Axenfeld-Rieger syndrome, type 3, 602482;Anterior segment dysgenesis 3, multiple subtypes, 601631	VISION DISORDERS PANEL CRANIOFACIAL ANOMALIES PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS

FOXC2	100%	100%	100%	96.7%	Lymphedema-distichiasis syndrome, 153400;Lymphedema-distichiasis syndrome with renal disease and diabetes mellitus, 153400	SKIN DISORDERS PANEL ¹ HEART DISORDERS PANEL ¹ LYMPHATIC ANOMALIES PANEL WITH GENOME WIDE CNV ANALYSIS RENAL DISORDERS PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS OROFACIAL CLEFTING PANEL WITH GENOME WIDE CNV ANALYSIS
FOXE1	100%	100%	100%	95%	Bamforth-Lazarus syndrome, 241850;{Thyroid cancer, nonmedullary, 4}, 616534	CRANIOFACIAL ANOMALIES PANEL WITH GENOME WIDE CNV ANALYSIS SKIN DISORDERS PANEL ¹ MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹ OROFACIAL CLEFTING PANEL WITH GENOME WIDE CNV ANALYSIS

FOXE3	99.7%	96.8%	100%	93%	Anterior segment dysgenesis 2, multiple subtypes, 610256;{Aortic aneurysm, familial thoracic 11, susceptibility to}, 617349;Cataract 34, multiple types, 612968	THORACIC AORTIC ANEURYSM AND/OR DISSECTION PANEL VISION DISORDERS PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹
FOXF1	100%	100%	100%	97.1%	Alveolar capillary dysplasia with misalignment of pulmonary veins, 265380	CILIOPATHIES PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS
FOXF2	98.7%	96.5%	100%	94.7%		HEARING IMPAIRMENT PANEL (INCLUDING GJB2) MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS

FOXG1	100%	99.5%	100%	95.7%	Rett syndrome, congenital variant, 613454	EPILEPSY PANEL INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS
FOXH1	100%	100%	100%	99.2%		CONGENITAL HEARTDISEASE PANEL ¹ HEART DISORDERS PANEL ¹ MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS
FOXI1	100%	100%	100%	99.1%	Enlarged vestibular aqueduct, 600791	HEARING IMPAIRMENT PANEL (INCLUDING GJB2) RENAL DISORDERS PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹

FOXI3	97%	92.7%	99.8%	94.4%	Craniofacial microsomia 2, 620444	PRIMARY IMMUNODEFICIENCIES PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹ SEVERE COMBINED IMMUNODEFICIENCY (SCID) PANEL
FOXJ1	100%	100%	100%	98.2%	Ciliary dyskinesia, primary, 43, 618699	CONGENITAL HEARTDISEASE PANEL ¹ CILIOPATHIES PANEL HEART DISORDERS PANEL ¹ INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS
FOXL1	100%	100%	100%	96.8%	Otosclerosis 11, 620576	MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS

FOXL2	100%	99.9%	100%	94.9%	Blepharophimosis, epicanthus inversus, and ptosis, type 2, 110100;Blepharophimosis, epicanthus inversus, and ptosis, type 1, 110100;Premature ovarian failure 3, 608996	DISORDERS/DIFFERENCES OF SEX DEVELOPMENT (DSD) / PRIMARY ADRENAL INSUFFICIENCY PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS PREMATURE OVARIAN INSUFFICIENCY PANEL
FOXN1	100%	100%	100%	99.2%	T-cell lymphopenia, infantile, with or without nail dystrophy, autosomal dominant, 618806;T-cell immunodeficiency, congenital alopecia, and nail dystrophy, 601705	SKIN DISORDERS PANEL ¹ PRIMARY IMMUNODEFICIENCIES PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹ SEVERE COMBINED IMMUNODEFICIENCY (SCID) PANEL
FOXO1	100%	99.8%	100%	96.5%	Rhabdomyosarcoma, alveolar, 268220	MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS

FOXP1	100%	100%	100%	99.2%	Intellectual developmental disorder with language impairment with or without autistic features, 613670	INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS
FOXP2	100%	100%	100%	98.1%	Speech-language disorder-1, 602081	INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS
FOXP3	100%	99.6%	98.5%	65.6%	Immunodysregulation, polyendocrinopathy, and enteropathy, X-linked, 304790	SKIN DISORDERS PANEL ¹ PRIMARY IMMUNODEFICIENCIES PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS

FOXP4	100%	100%	100%	98.4%		INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS
FOXRED1	100%	100%	100%	98.3%	Mitochondrial complex I deficiency, nuclear type 19, 618241	EPILEPSY PANEL INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS MITOCHONDRIAL DISORDERS PANEL COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL¹
FPR1	100%	100%	100%	99.4%		PRIMARY IMMUNODEFICIENCIES PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS

FRA10AC1	100%	100%	100%	99.8%	Neurodevelopmental disorder with growth retardation, dysmorphic facies, and corpus callosum abnormalities, 620113	INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL¹
FRAS1	100%	100%	100%	99.4%	Fraser syndrome 1, 219000	DISORDERS/DIFFEREN CES OF SEX DEVELOPMENT (DSD) / PRIMARY ADRENAL INSUFFICIENCY PANEL INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS RENAL DISORDERS PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL¹ OROFACIAL CLEFTING PANEL WITH GENOME WIDE CNV ANALYSIS

FREM1	100%	100%	100%	99.1%	Manitoba oculotrichoanal syndrome, 248450;Bifid nose with or without anorectal and renal anomalies, 608980;Trigonocephaly 2, 614485	VISION DISORDERS PANEL SKIN DISORDERS PANEL ¹ RENAL DISORDERS PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹
FREM2	100%	100%	100%	99.5%	Fraser syndrome 2, 617666;Cryptophthalmos, unilateral or bilateral, isolated, 123570	DISORDERS/DIFFEREN CES OF SEX DEVELOPMENT (DSD) / PRIMARY ADRENAL INSUFFICIENCY PANEL RENAL DISORDERS PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹

FRMD4A	96.6%	96.6%	100%	99%	?Corpus callosum, agenesis of, with facial anomalies and cerebellar ataxia, 616819	INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS
FRMD5	100%	100%	100%	99.4%	Neurodevelopmental disorder with eye movement abnormalities and ataxia, 620094	MOVEMENT DISORDERS PANEL INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS
FRMD7	100%	99.9%	99.4%	71.9%	Nystagmus, infantile periodic alternating, X-linked, 310700;Nystagmus 1, congenital, X-linked, 310700	MOVEMENT DISORDERS PANEL VISION DISORDERS PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS

FRMPD4	100%	99.7%	98.1%	66.5%	Intellectual developmental disorder, X-linked 104, 300983	EPILEPSY PANEL INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS
FRRS1L	100%	100%	99.8%	93.6%	Developmental and epileptic encephalopathy 37, 616981	EPILEPSY PANEL INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹
FRYL	100%	100%	100%	99.4%		INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS

FSCN2	100%	100%	100%	98.3%	Retinitis pigmentosa 30, 607921	MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS
FSHB	100%	100%	100%	99.8%	Hypogonadotropic hypogonadism 24 without anosmia, 229070	DISORDERS/DIFFEREN CES OF SEX DEVELOPMENT (DSD) / PRIMARY ADRENAL INSUFFICIENCY PANEL HYPOGONADOTROPIC HYPOGONADISM (KALLMANN) PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL¹ PREMATURE OVARIAN INSUFFICIENCY PANEL

FSHR	100%	100%	100%	99.3%	Ovarian hyperstimulation syndrome, 608115;Ovarian dysgenesis 1, 233300	PREMATURE OVARIAN INSUFFICIENCY PANEL DISORDERS/DIFFEREN CES OF SEX DEVELOPMENT (DSD) / PRIMARY ADRENAL INSUFFICIENCY PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL¹
FSIP2	100%	100%	100%	99.7%	Spermatogenic failure 34, 618153	MALE INFERTILITY PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS

FTCD	100%	100%	100%	95.4%	Glutamate formiminotransferase deficiency, 229100	METABOLIC DISORDERS PANEL INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL¹
FTH1	100%	100%	100%	99.4%	Neurodegeneration with brain iron accumulation 9, 620669;?Hemochromatosis, type 5, 615517	MOVEMENT DISORDERS PANEL IRON DISORDERS PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS

FTL	100%	100%	100%	98.8%	Hyperferritinemia-cataract syndrome, 600886;L-ferritin deficiency, dominant and recessive, 615604;Neurodegeneration with brain iron accumulation 3, 606159	MOVEMENT DISORDERS PANEL VISION DISORDERS PANEL IRON DISORDERS PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS MITOCHONDRIAL DISORDERS PANEL PARKINSON DISEASE PANEL
FTO	94.5%	94.5%	100%	99.6%	Growth retardation, developmental delay, facial dysmorphism, 612938;{Obesity, susceptibility to, BMIQ14}, 612460	INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹ OROFACIAL CLEFTING PANEL WITH GENOME WIDE CNV ANALYSIS

FTSJ1	100%	99.9%	98.8%	64%	Intellectual developmental disorder, X-linked 9, 309549	INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS
FUCA1	100%	100%	100%	99.4%	Fucosidosis, 230000	SKIN DISORDERS PANEL ¹ SHORT STATURE/SKELETAL DYSPLASIA PANEL WITH GENOME WIDE CNV ANALYSIS METABOLIC DISORDERS PANEL INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹

FURIN	100%	100%	100%	99.1%		THORACIC AORTIC ANEURYSM AND/OR DISSECTION PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS
FUS	100%	100%	100%	97.5%	Amyotrophic lateral sclerosis 6, with or without frontotemporal dementia, 608030;Essential tremor, hereditary, 4, 614782	AMYOTROPHIC LATERAL SCLEROSIS, ALS PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS
FUT2	100%	100%	100%	97.8%	{Norwalk virus infection, resistance to};{Vitamin B12 plasma level QTL1}, 612542;[Bombay phenotype, digenic], 616754	METABOLIC DISORDERS PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS
FUT6	100%	100%	100%	98.2%	[Fucosyltransferase 6 deficiency], 613852	METABOLIC DISORDERS PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS

FUT8	100%	100%	100%	99.4%	Congenital disorder of glycosylation with defective fucosylation 1, 618005	METABOLIC DISORDERS PANEL INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹
FUZ	100%	100%	100%	98.4%	{Neural tube defects, susceptibility to}, 182940	CILIOPATHIES PANEL SHORT STATURE/SKELETAL DYSPLASIA PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS

FXN	100%	100%	100%	98.6%	Friedreich ataxia with retained reflexes, 229300;Friedreich ataxia, 229300	COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹ MITOCHONDRIAL DISORDERS PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS POLYNEUROPATHIES PANEL ¹
FXR1	100%	100%	99.7%	95.5%	Congenital myopathy 9B, proximal, with minicore lesions, 618823;?Congenital myopathy 9A with respiratory insufficiency and bone fractures, 618822	MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS MUSCLE DISORDERS PANEL
FXD2	100%	100%	100%	99.2%	Hypomagnesemia 2, renal, 154020	EPILEPSY PANEL RENAL DISORDERS PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS

FYB1	100%	100%	100%	99.6%	Thrombocytopenia 3, 273900	HEMOSTATIC/THROMBOTIC DISORDERS PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS
FYCO1	100%	100%	100%	99.1%	Cataract 18, autosomal recessive, 610019	VISION DISORDERS PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹
FZD2	100%	100%	99.9%	97.2%	Omodysplasia 2, 164745	DISORDERS/DIFFERENCES OF SEX DEVELOPMENT (DSD) / PRIMARY ADRENAL INSUFFICIENCY PANEL SHORT STATURE/SKELETAL DYSPLASIA PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS

FZD4	100%	100%	100%	99%	Retinopathy of prematurity, 133780;Exudative vitreoretinopathy 1, 133780	VISION DISORDERS PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS
FZD5	100%	100%	100%	99.3%	Microphthalmia/coloboma 11, 620731	VISION DISORDERS PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS
FZD6	100%	100%	100%	99.2%	Nail disorder, nonsyndromic congenital, 1, 161050	SKIN DISORDERS PANEL ¹ COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹ MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS
FZR1	100%	100%	100%	99.1%	Developmental and epileptic encephalopathy 109, 620145	EPILEPSY PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS

G6PC1	100%	100%	100%	99.1%	Glycogen storage disease Ia, 232200	COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL¹ MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS RENAL DISORDERS PANEL METABOLIC DISORDERS PANEL PRIMARY IMMUNODEFICIENCIES PANEL
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G6PC3	96.7%	96.7%	100%	99.3%	Dursun syndrome, 612541;Neutropenia, severe congenital 4, autosomal recessive, 612541	COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹ MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS METABOLIC DISORDERS PANEL PRIMARY IMMUNODEFICIENCIES PANEL INHERITED BONE MARROW FAILURE AND/OR PREDISPOSITION PANEL TO HEMATOLOGICAL MALIGNANCIES HEREDITARY CANCER PANEL
G6PD	86.3%	85.9%	98.7%	69.1%	Anemia, congenital, nonspherocytic hemolytic, 1, G6PD deficient, 300908;{Resistance to malaria due to G6PD deficiency}, 611162	PRIMARY IMMUNODEFICIENCIES PANEL METABOLIC DISORDERS PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS

GAA	100%	100%	100%	99.3%	Glycogen storage disease II, 232300	HEART DISORDERS PANEL ¹ COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹ MUSCLE DISORDERS PANEL METABOLIC DISORDERS PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS
GAB1	100%	100%	100%	99.6%	?Deafness, autosomal recessive 26, 605428	HEARING IMPAIRMENT PANEL (INCLUDING GJB2) MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS
GABBR1	100%	100%	100%	97.4%	Neurodevelopmental disorder with language delay and variable cognitive abnormalities, 620502	INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS

GABBR2	99.6%	99.1%	100%	98.7%	{Nicotine dependence, protection against}, 188890;{Nicotine dependence, susceptibility to}, 188890;Developmental and epileptic encephalopathy 59, 617904;Neurodevelopmental disorder with poor language and loss of hand skills, 617903	EPILEPSY PANEL INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS
GABRA1	100%	100%	100%	99%	{Epilepsy, juvenile myoclonic, susceptibility to, 5}, 611136;Developmental and epileptic encephalopathy 19, 615744;{Epilepsy, childhood absence, susceptibility to, 4}, 611136	EPILEPSY PANEL INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS
GABRA2	100%	100%	100%	99.1%	Developmental and epileptic encephalopathy 78, 618557;{Alcohol dependence, susceptibility to}, 103780	EPILEPSY PANEL INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS

GABRA3	100%	100%	98.7%	68%	Epilepsy, X-linked 2, with or without impaired intellectual development and dysmorphic features, 301091	EPILEPSY PANEL INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS
GABRA4	100%	100%	100%	99.6%		INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS
GABRA5	100%	100%	100%	98.8%	Developmental and epileptic encephalopathy 79, 618559	INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS

GABRB1	100%	100%	100%	99.3%	Developmental and epileptic encephalopathy 45, 617153	EPILEPSY PANEL INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS
GABRB2	100%	100%	100%	99.3%	Developmental and epileptic encephalopathy 92, 617829	EPILEPSY PANEL INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS
GABRB3	100%	100%	100%	99.1%	{Epilepsy, childhood absence, susceptibility to, 5}, 612269;Developmental and epileptic encephalopathy 43, 617113	EPILEPSY PANEL INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS

GABRD	100%	99.7%	100%	97.3%	{?Generalized epilepsy with febrile seizures plus, type 5, susceptibility to}, 613060	INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS
GABRG2	92.9%	92.9%	100%	99.1%	Developmental and epileptic encephalopathy 74, 618396;Febrile seizures, familial, 8, 607681;Generalized epilepsy with febrile seizures plus, type 3, 607681	EPILEPSY PANEL INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS
GAD1	100%	100%	100%	99.2%	Developmental and epileptic encephalopathy 89, 619124	EPILEPSY PANEL METABOLIC DISORDERS PANEL INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹

GAL	100%	100%	100%	99.1%	?Epilepsy, familial temporal lobe, 8, 616461	MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS
GALC	100%	100%	100%	99.2%	Krabbe disease, 245200	MOVEMENT DISORDERS PANEL POLYNEUROPATHIES PANEL ¹ METABOLIC DISORDERS PANEL INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹

GALE	100%	100%	100%	99.4%	Thrombocytopenia 13, syndromic, 620776;Galactose epimerase deficiency, 230350	INHERITED BONE MARROW FAILURE AND/OR PREDISPOSITION PANEL TO HEMATOLOGICAL MALIGNANCIES HEMOSTATIC/THROMBOTIC DISORDERS PANEL METABOLIC DISORDERS PANEL INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹
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GALK1	100%	100%	100%	99.2%	Galactokinase deficiency with cataracts, 230200	VISION DISORDERS PANEL METABOLIC DISORDERS PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹
GALM	100%	100%	100%	99.2%	Galactosemia IV, 618881	VISION DISORDERS PANEL LIVER DISORDERS PANEL METABOLIC DISORDERS PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS

GALNS	100%	100%	100%	98.6%	Mucopolysaccharidosis IVA, 253000	SKIN DISORDERS PANEL ¹ SHORT STATURE/SKELETAL DYSPLASIA PANEL WITH GENOME WIDE CNV ANALYSIS METABOLIC DISORDERS PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹
GALNT12	99.9%	98.9%	100%	99.3%	{Colorectal cancer, susceptibility to, 1}, 608812	MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS HEREDITARY CANCER PANEL

GALNT2	100%	100%	100%	99.2%	Congenital disorder of glycosylation, type II, 618885	SHORT STATURE/SKELETAL DYSPLASIA PANEL WITH GENOME WIDE CNV ANALYSIS METABOLIC DISORDERS PANEL INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL¹
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GALNT3	100%	100%	100%	99.6%	Tumoral calcinosis, hyperphosphatemic, familial, 1, 211900	SKIN DISORDERS PANEL ¹ SHORT STATURE/SKELETAL DYSPLASIA PANEL WITH GENOME WIDE CNV ANALYSIS METABOLIC DISORDERS PANEL RENAL DISORDERS PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹
GALNTL5	100%	100%	100%	99.4%		MALE INFERTILITY PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS

GALT	100%	100%	100%	99.1%	Galactosemia, 230400	VISION DISORDERS PANEL PREMATURE OVARIAN INSUFFICIENCY PANEL DISORDERS/DIFFEREN CES OF SEX DEVELOPMENT (DSD) / PRIMARY ADRENAL INSUFFICIENCY PANEL LIVER DISORDERS PANEL METABOLIC DISORDERS PANEL INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL¹
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GAMT	100%	100%	100%	99.1%	Cerebral creatine deficiency syndrome 2, 612736	MOVEMENT DISORDERS PANEL EPILEPSY PANEL METABOLIC DISORDERS PANEL INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹
GAN	100%	100%	100%	99.3%	Giant axonal neuropathy-1, 256850	MOVEMENT DISORDERS PANEL SKIN DISORDERS PANEL ¹ POLYNEUROPATHIES PANEL ¹ MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹

GANAB	100%	100%	100%	99.2%	Polycystic kidney disease 3, 600666	LIVER DISORDERS PANEL METABOLIC DISORDERS PANEL RENAL DISORDERS PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS
GAPVD1	100%	100%	100%	99.6%		RENAL DISORDERS PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS
GARS1	98.9%	98.9%	100%	99.4%	Spinal muscular atrophy, infantile, James type, 619042;Neuronopathy, distal hereditary motor, autosomal dominant 5, 600794;Charcot-Marie-Tooth disease, type 2D, 601472	POLYNEUROPATHIES PANEL ¹ MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS MITOCHONDRIAL DISORDERS PANEL

GAS2	100%	100%	100%	99.2%	?Deafness, autosomal recessive 125, 620877	HEARING IMPAIRMENT PANEL (INCLUDING GJB2) MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS
GAS2L2	100%	100%	100%	98.7%	?Ciliary dyskinesia, primary, 41, 618449	MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS
GAS8	100%	100%	100%	98.5%	Ciliary dyskinesia, primary, 33, 616726	CILIOPATHIES PANEL MALE INFERTILITY PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹

GATA1	100%	99.8%	97%	64.8%	Anemia, congenital, nonspherocytic hemolytic, 9, 301083;Leukemia, megakaryoblastic, with or without Down syndrome, somatic, 159595;Thrombocytopenia, X-linked, with or without dyserythropoietic anemia, 300367;Anemia, X-linked, with/without neutropenia and/or platelet abnormalities, 300835;Thrombocytopenia with beta-thalassemia, X-linked, 314050	INHERITED BONE MARROW FAILURE AND/OR PREDISPOSITION PANEL TO HEMATOLOGICAL MALIGNANCIES HEMOSTATIC/THROMBOTIC DISORDERS PANEL IRON DISORDERS PANEL PRIMARY IMMUNODEFICIENCIES PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS
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GATA2	85.7%	85.7%	100%	99.2%	{Leukemia, acute myeloid, susceptibility to}, 601626;Emberger syndrome, 614038;Immunodeficiency 21, 614172;{Myelodysplastic syndrome, susceptibility to}, 614286	INHERITED BONE MARROW FAILURE AND/OR PREDISPOSITION PANEL TO HEMATOLOGICAL MALIGNANCIES SKIN DISORDERS PANEL ¹ HEMOSTATIC/THROMBOTIC DISORDERS PANEL PRIMARY IMMUNODEFICIENCIES PANEL LYMPHATIC ANOMALIES PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS HEREDITARY CANCER PANEL
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GATA3	100%	100%	100%	97.1%	Hypoparathyroidism, sensorineural deafness, and renal dysplasia, 146255	HEARING IMPAIRMENT PANEL (INCLUDING GJB2) RENAL DISORDERS PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS
GATA4	100%	100%	100%	98.4%	Tetralogy of Fallot, 187500;Atrial septal defect 2, 607941;Ventricular septal defect 1, 614429;Atrioventricular septal defect 4, 614430;?Testicular anomalies with or without congenital heart disease, 615542	CONGENITAL HEARTDISEASE PANEL ¹ DISORDERS/DIFFEREN CES OF SEX DEVELOPMENT (DSD) / PRIMARY ADRENAL INSUFFICIENCY PANEL HEART DISORDERS PANEL ¹ MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS
GATA5	100%	100%	100%	98.5%	Congenital heart defects, multiple types, 5, 617912	CONGENITAL HEARTDISEASE PANEL ¹ HEART DISORDERS PANEL ¹ MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS

GATA6	100%	100%	100%	96.4%	Atrial septal defect 9, 614475;Persistent truncus arteriosus, 217095;Pancreatic agenesis and congenital heart defects, 600001;Atrioventricular septal defect 5, 614474;Tetralogy of Fallot, 187500	CONGENITAL HEARTDISEASE PANEL ¹ HEART DISORDERS PANEL ¹ MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS
GATAD1	100%	100%	100%	99.3%	?Cardiomyopathy, dilated, 2B, 614672	HEART DISORDERS PANEL ¹ MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS
GATAD2A	100%	99.8%	100%	99.2%		INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS
GATAD2B	100%	100%	100%	99%	GAND syndrome, 615074	INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS

GATB	100%	100%	100%	99.3%	?Combined oxidative phosphorylation deficiency 41, 618838	HEART DISORDERS PANEL ¹ MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS MITOCHONDRIAL DISORDERS PANEL
GATC	100%	100%	100%	98.7%	Combined oxidative phosphorylation deficiency 42, 618839	HEART DISORDERS PANEL ¹ MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS MITOCHONDRIAL DISORDERS PANEL

GATM	100%	100%	100%	99.4%	Cerebral creatine deficiency syndrome 3, 612718;Fanconi renotubular syndrome 1, 134600	MUSCLE DISORDERS PANEL METABOLIC DISORDERS PANEL INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS MITOCHONDRIAL DISORDERS PANEL COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹
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GBA1	100%	100%	100%	99.3%	{Lewy body dementia, susceptibility to}, 127750;Gaucher disease, type II, 230900;Gaucher disease, type IIIC, 231005;Gaucher disease, type III, 231000;Gaucher disease, type I, 230800;Gaucher disease, perinatal lethal, 608013;{Parkinson disease, late-onset, susceptibility to}, 168600	FETAL AKINESIA PANEL MOVEMENT DISORDERS PANEL INHERITED BONE MARROW FAILURE AND/OR PREDISPOSITION PANEL TO HEMATOLOGICAL MALIGNANCIES HEMOSTATIC/THROMB OTIC DISORDERS PANEL COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL¹ PARKINSON DISEASE PANEL SHORT STATURE/SKELETAL DYSPLASIA PANEL WITH GENOME WIDE CNV ANALYSIS LIVER DISORDERS PANEL METABOLIC DISORDERS PANEL INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE
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						CNV ANALYSIS
GBA2	100%	100%	100%	99.1%	Spastic paraplegia 46, autosomal recessive, 614409	MOVEMENT DISORDERS PANEL POLYNEUROPATHIES PANEL ¹ METABOLIC DISORDERS PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹

GBE1	100%	100%	100%	99.5%	Glycogen storage disease IV, 232500;Polyglucosan body disease, adult form, 263570	FETAL AKINESIA PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹ MUSCLE DISORDERS PANEL METABOLIC DISORDERS PANEL HEART DISORDERS PANEL ¹ POLYNEUROPATHIES PANEL ¹ LIVER DISORDERS PANEL MOVEMENT DISORDERS PANEL
GBF1	100%	100%	100%	98.9%	Charcot-Marie-Tooth disease, axonal, type 2GG, 606483	POLYNEUROPATHIES PANEL ¹ MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS MITOCHONDRIAL DISORDERS PANEL

GCDH	100%	100%	100%	99.1%	Glutaricaciduria, type I, 231670	MOVEMENT DISORDERS PANEL METABOLIC DISORDERS PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL¹
GCGR	100%	100%	100%	99.1%	Mahvash disease, 619290	MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS
GCH1	100%	100%	100%	99.8%	Dystonia, DOPA-responsive, 128230;Hyperphenylalaninemia, BH4-deficient, B, 233910	MOVEMENT DISORDERS PANEL METABOLIC DISORDERS PANEL INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS PARKINSON DISEASE PANEL COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL¹

GCK	100%	100%	100%	99.1%	MODY, type II, 125851;Diabetes mellitus, permanent neonatal 1, 606176;Hyperinsulinemic hypoglycemia, familial, 3, 602485;Diabetes mellitus, noninsulin-dependent, late onset, 125853	EPILEPSY PANEL METABOLIC DISORDERS PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹
GCLC	100%	100%	100%	99.5%	{Myocardial infarction, susceptibility to}, 608446;Anemia, congenital, nonspherocytic hemolytic, 7, 230450	METABOLIC DISORDERS PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹
GCLM	100%	100%	100%	99.3%	{Myocardial infarction, susceptibility to}, 608446	METABOLIC DISORDERS PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS

GCM2	100%	100%	100%	99.2%	Hypoparathyroidism, familial isolated 2, 618883;Hyperparathyroidism 4, 617343	SHORT STATURE/SKELETAL DYSPLASIA PANEL WITH GENOME WIDE CNV ANALYSIS RENAL DISORDERS PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS
GCNA	100%	100%	98.2%	70.2%	Spermatogenic failure, X-linked, 4, 301077	MALE INFERTILITY PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS
GCNT2	100%	100%	100%	98.7%	[Blood group, li], 110800;Adult i phenotype without cataract, 110800;Cataract 13 with adult i phenotype, 116700	VISION DISORDERS PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹

GCSH	100%	100%	100%	99.6%	Multiple mitochondrial dysfunctions syndrome 7, 620423	EPILEPSY PANEL METABOLIC DISORDERS PANEL INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS MITOCHONDRIAL DISORDERS PANEL COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹
GDAP1	86.7%	86.7%	98.6%	97.3%	Charcot-Marie-Tooth disease, axonal, with vocal cord paresis, 607706;Charcot-Marie-Tooth disease, recessive intermediate, A, 608340;Charcot-Marie-Tooth disease, axonal, type 2K, 607831;Charcot-Marie-Tooth disease, type 4A, 214400	POLYNEUROPATHIES PANEL ¹ MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS MITOCHONDRIAL DISORDERS PANEL COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹

GDAP2	100%	100%	100%	99.6%	Spinocerebellar ataxia, autosomal recessive 27, 618369	MOVEMENT DISORDERS PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹
GDF1	99.6%	98.2%	100%	96.7%	Congenital heart defects, multiple types, 6, 613854;Right atrial isomerism (Ivemark), 208530	CONGENITAL HEARTDISEASE PANEL ¹ CILIOPATHIES PANEL HEART DISORDERS PANEL ¹ MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹
GDF11	100%	99%	100%	92.5%	?Vertebral hypersegmentation and orofacial anomalies, 619122	MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS

GDF2	100%	100%	100%	99.2%	Telangiectasia, hereditary hemorrhagic, type 5, 615506	SKIN DISORDERS PANEL ¹ HEART DISORDERS PANEL ¹ HEMOSTATIC/THROMB OTIC DISORDERS PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS
GDF3	100%	100%	100%	99.4%	Klippel-Feil syndrome 3, autosomal dominant, 613702;Microphthalmia, isolated, with coloboma 6, 613703;Microphthalmia, isolated 7, 613704	VISION DISORDERS PANEL CRANIOFACIAL ANOMALIES PANEL WITH GENOME WIDE CNV ANALYSIS SHORT STATURE/SKELETAL DYSPLASIA PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS

GDF5	100%	100%	100%	98.2%	Acromesomelic dysplasia 2A, 200700;Acromesomelic dysplasia 2B, 228900;Multiple synostoses syndrome 2, 610017;Symphalangism, proximal, 1B, 615298;Brachydactyly, type A2, 112600;?Acromesomelic dysplasia 2C, Hunter-Thompson type, 201250;Brachydactyly, type C, 113100;{Osteoarthritis-5}, 612400;Brachydactyly, type A1, C, 615072	SKIN DISORDERS PANEL ¹ SHORT STATURE/SKELETAL DYSPLASIA PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹
GDF6	100%	100%	100%	97.7%	Microphthalmia with coloboma 6, digenic, 613703;Microphthalmia, isolated 4, 613094;Leber congenital amaurosis 17, 615360;Multiple synostoses syndrome 4, 617898;Klippel-Feil syndrome 1, autosomal dominant, 118100	SHORT STATURE/SKELETAL DYSPLASIA PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS OROFACIAL CLEFTING PANEL WITH GENOME WIDE CNV ANALYSIS CRANIOFACIAL ANOMALIES PANEL WITH GENOME WIDE CNV ANALYSIS VISION DISORDERS PANEL

GDF9	100%	100%	100%	99.7%	Premature ovarian failure 14, 618014	MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS PREMATURE OVARIAN INSUFFICIENCY PANEL DISORDERS/DIFFEREN CES OF SEX DEVELOPMENT (DSD) / PRIMARY ADRENAL INSUFFICIENCY PANEL
GDI1	100%	100%	98.3%	67.2%	Intellectual developmental disorder, X-linked 41, 300849	INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS
GDNF	100%	100%	100%	98.8%	{Hirschsprung disease, susceptibility to, 3}, 613711	LIVER DISORDERS PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS HEREDITARY CANCER PANEL

GDPD1	100%	100%	100%	99.3%		VISION DISORDERS PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS
GEMIN4	100%	100%	100%	99.6%	Neurodevelopmental disorder with microcephaly, cataracts, and renal abnormalities, 617913	INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL¹
GEMIN5	100%	100%	100%	99.4%	Neurodevelopmental disorder with cerebellar atrophy and motor dysfunction, 619333	MOVEMENT DISORDERS PANEL INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL¹

GET3	100%	100%	100%	97.5%	?Cardiomyopathy, dilated, 2H, 620203	MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS
GET4	100%	100%	100%	97.9%	?Congenital disorder of glycosylation, type Ily, 620200	MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS
GFAP	100%	100%	100%	99.5%	Alexander disease, 203450	MOVEMENT DISORDERS PANEL INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS

GFER	100%	100%	100%	98%	Myopathy, mitochondrial progressive, with congenital cataract and developmental delay, 613076	VISION DISORDERS PANEL INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS MITOCHONDRIAL DISORDERS PANEL COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹
GFI1	100%	100%	100%	98.8%	?Neutropenia, nonimmune chronic idiopathic, of adults, 607847;Neutropenia, severe congenital 2, autosomal dominant, 613107	INHERITED BONE MARROW FAILURE AND/OR PREDISPOSITION PANEL TO HEMATOLOGICAL MALIGNANCIES PRIMARY IMMUNODEFICIENCIES PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS HEREDITARY CANCER PANEL

GFI1B	100%	100%	100%	98.6%	Bleeding disorder, platelet-type, 17, 187900	HEMOSTATIC/THROMBOTIC DISORDERS PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS
GFM1	100%	100%	100%	99.6%	Combined oxidative phosphorylation deficiency 1, 609060	LIVER DISORDERS PANEL INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS MITOCHONDRIAL DISORDERS PANEL COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL¹

GFM2	100%	100%	100%	99.6%	Combined oxidative phosphorylation deficiency 39, 618397	INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS MITOCHONDRIAL DISORDERS PANEL COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹
GFPT1	100%	100%	100%	99.6%	Myasthenia, congenital, 12, with tubular aggregates, 610542	METABOLIC DISORDERS PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹ MUSCLE DISORDERS PANEL

GFRA1	94%	94%	100%	99.4%	Renal hypodysplasia/aplasia 4, 619887	RENAL DISORDERS PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹
GFUS	100%	100%	100%	99.5%		METABOLIC DISORDERS PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS
GGCX	100%	100%	100%	98.9%	Vitamin K-dependent clotting factors, combined deficiency of, 1, 277450;Pseudoxanthoma elasticum-like disorder with multiple coagulation factor deficiency, 610842	SKIN DISORDERS PANEL ¹ HEMOSTATIC/THROMB OTIC DISORDERS PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹

GGN	100%	100%	100%	99.5%	Spermatogenic failure 69, 619826	MALE INFERTILITY PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS
GGPS1	100%	100%	100%	99.9%	Muscular dystrophy, congenital hearing loss, and ovarian insufficiency syndrome, 619518	DISORDERS/DIFFEREN CES OF SEX DEVELOPMENT (DSD) / PRIMARY ADRENAL INSUFFICIENCY PANEL METABOLIC DISORDERS PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL¹ PREMATURE OVARIAN INSUFFICIENCY PANEL
GGT1	100%	100%	100%	99.2%	?Glutathioninuria, 231950	MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL¹

GH1	100%	100%	100%	98.9%	Kowarski syndrome, 262650;Growth hormone deficiency, isolated, type II, 173100;Growth hormone deficiency, isolated, type IB, 612781;Growth hormone deficiency, isolated, type IA, 262400	SHORT STATURE/SKELETAL DYSPLASIA PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹
GHR	99.4%	99.4%	99.8%	98.4%	Laron dwarfism, 262500;Increased responsiveness to growth hormone, 604271;Growth hormone insensitivity, partial, 604271;{Hypercholesterolemia, familial, modifier of}, 143890	SHORT STATURE/SKELETAL DYSPLASIA PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹

GHRHR	100%	100%	100%	98.4%	Growth hormone deficiency, isolated, type IV, 618157	SHORT STATURE/SKELETAL DYSPLASIA PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹
GHSR	100%	100%	100%	99.1%	Growth hormone deficiency, isolated partial, 615925	SHORT STATURE/SKELETAL DYSPLASIA PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹
GIGYF1	100%	100%	100%	98.8%		INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS

GIMAP5	100%	100%	100%	99.5%	Portal hypertension, noncirrhotic, 2, 619463	PRIMARY IMMUNODEFICIENCIES PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹
GIMAP6	100%	100%	100%	99%		PRIMARY IMMUNODEFICIENCIES PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS
GINS1	81%	81%	100%	99.6%	Immunodeficiency 55, 617827	PRIMARY IMMUNODEFICIENCIES PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹

GINS2	100%	100%	100%	98.9%		SHORT STATURE/SKELETAL DYSPLASIA PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS
GINS4	100%	100%	100%	99.1%		INHERITED BONE MARROW FAILURE AND/OR PREDISPOSITION PANEL TO HEMATOLOGICAL MALIGNANCIES PRIMARY IMMUNODEFICIENCIES PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS
GIPC1	100%	100%	100%	99.1%	Oculopharyngodistal myopathy 2, 618940	MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS

GIPC3	100%	100%	100%	98.8%	Deafness, autosomal recessive 15, 601869	HEARING IMPAIRMENT PANEL (INCLUDING GJB2) MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL¹
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GJA1	100%	100%	100%	98.9%	Erythrokeratoderma variabilis et progressiva 3, 617525;Craniometaphyseal dysplasia, autosomal recessive, 218400;Oculodentodigital dysplasia, 164200;Palmoplantar keratoderma with congenital alopecia, 104100;Syndactyly, type III, 186100;Oculodentodigital dysplasia, autosomal recessive, 257850	VISION DISORDERS PANEL CRANIOFACIAL ANOMALIES PANEL WITH GENOME WIDE CNV ANALYSIS SKIN DISORDERS PANEL ¹ SHORT STATURE/SKELETAL DYSPLASIA PANEL WITH GENOME WIDE CNV ANALYSIS LYMPHATIC ANOMALIES PANEL WITH GENOME WIDE CNV ANALYSIS INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹ OROFACIAL CLEFTING PANEL WITH GENOME WIDE CNV ANALYSIS
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GJA3	100%	100%	100%	98.1%	Cataract 14, multiple types, 601885	VISION DISORDERS PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS
GJA5	100%	100%	100%	99.9%	Atrial fibrillation, familial, 11, 614049;Atrial standstill, digenic (GJA5/SCN5A), 108770	CONGENITAL HEARTDISEASE PANEL' HEART DISORDERS PANEL' MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS
GJA8	100%	100%	100%	98.8%	Cataract 1, multiple types, 116200	VISION DISORDERS PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS

GJB1	100%	99.9%	94.5%	56.4%	Charcot-Marie-Tooth neuropathy, X-linked dominant, 1, 302800	MOVEMENT DISORDERS PANEL POLYNEUROPATHIES PANEL ¹ INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS
GJB2	100%	100%	100%	99.5%	Keratoderma, palmoplantar, with deafness, 148350;Deafness, autosomal recessive 1A, 220290;Deafness, autosomal dominant 3A, 601544;Hystrix-like ichthyosis with deafness, 602540;Bart-Pumphrey syndrome, 149200;Keratitis-ichthyosis-deafness syndrome, 148210;Vohwinkel syndrome, 124500	SKIN DISORDERS PANEL ¹ HEARING IMPAIRMENT PANEL (INCLUDING GJB2) MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹

GJB3	100%	100%	100%	98.9%	Deafness, digenic, GJB2/GJB3, 220290;Erythrokeratoderma variabilis et progressiva 1, 133200;Deafness, autosomal dominant 2B, with or without peripheral neuropathy, 612644	SKIN DISORDERS PANEL ¹ MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹ HEARING IMPAIRMENT PANEL (INCLUDING GJB2) POLYNEUROPATHIES PANEL ¹
GJB4	100%	100%	100%	99.1%	Erythrokeratoderma variabilis et progressiva 2, 617524	SKIN DISORDERS PANEL ¹ MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS

GJB6	100%	100%	100%	97.5%	Ectodermal dysplasia 2, Clouston type, 129500;Deafness, autosomal dominant 3B, 612643;Deafness, autosomal recessive 1B, 612645;Deafness, digenic GJB2/GJB6, 220290	CRANIOFACIAL ANOMALIES PANEL WITH GENOME WIDE CNV ANALYSIS SKIN DISORDERS PANEL ¹ HEARING IMPAIRMENT PANEL (INCLUDING GJB2) MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹
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GJC2	99.7%	95.8%	100%	98%	Lymphatic malformation 3, 613480;?Spastic paraplegia 44, autosomal recessive, 613206;Leukodystrophy, hypomyelinating, 2, 608804	INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS PRIMARY IMMUNODEFICIENCIES PANEL LYMPHATIC ANOMALIES PANEL WITH GENOME WIDE CNV ANALYSIS MOVEMENT DISORDERS PANEL SKIN DISORDERS PANEL ¹ MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹
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GK	100%	99.8%	99%	72.3%	Glycerol kinase deficiency, 307030	DISORDERS/DIFFERENCES OF SEX DEVELOPMENT (DSD) / PRIMARY ADRENAL INSUFFICIENCY PANEL METABOLIC DISORDERS PANEL INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS
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GLA	91.4%	91.3%	98.9%	67.5%	Fabry disease, cardiac variant, 301500;Fabry disease, 301500	SKIN DISORDERS PANEL ¹ SMALL FIBRE NEUROPATHY PANEL ¹ METABOLIC DISORDERS PANEL RENAL DISORDERS PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS NEUROLOGICAL PAIN DISORDERS PANEL ¹ LYMPHATIC ANOMALIES PANEL WITH GENOME WIDE CNV ANALYSIS HYPERTROPHIC CARDIOMYOPATHY PANEL ¹ POLYNEUROPATHIES PANEL ¹ HEARING IMPAIRMENT PANEL (INCLUDING GJB2) HEART DISORDERS PANEL ¹
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GLB1	100%	100%	100%	99.2%	GM1-gangliosidosis, type I, 230500;GM1-gangliosidosis, type III, 230650;Mucopolysaccharidosis type IVB (Morquio), 253010;GM1-gangliosidosis, type II, 230600	MOVEMENT DISORDERS PANEL SKIN DISORDERS PANEL ¹ HEART DISORDERS PANEL ¹ SHORT STATURE/SKELETAL DYSPLASIA PANEL WITH GENOME WIDE CNV ANALYSIS METABOLIC DISORDERS PANEL INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹
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GLDC	100%	99.9%	100%	99.3%	Glycine encephalopathy1, 605899	EPILEPSY PANEL METABOLIC DISORDERS PANEL INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL¹
GLDN	100%	100%	100%	99.4%	Lethal congenital contracture syndrome 11, 617194	FETAL AKINESIA PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL¹

GLE1	100%	100%	100%	99.1%	Lethal congenital contracture syndrome 1, 253310;Congenital arthrogryposis with anterior horn cell disease, 611890	FETAL AKINESIA PANEL POLYNEUROPATHIES PANEL ¹ MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹
GLI1	100%	100%	100%	98.8%	Polydactyly, preaxial I, 174400;Polydactyly, postaxial, type A8, 618123	SHORT STATURE/SKELETAL DYSPLASIA PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹

GLI2	100%	100%	100%	98.7%	Culler-Jones syndrome, 615849;Holoprosencephaly 9, 610829	CRANIOFACIAL ANOMALIES PANEL WITH GENOME WIDE CNV ANALYSIS DISORDERS/DIFFEREN CES OF SEX DEVELOPMENT (DSD) / PRIMARY ADRENAL INSUFFICIENCY PANEL HYPOGONADOTROPIC HYPOGONADISM (KALLMANN) PANEL SHORT STATURE/SKELETAL DYSPLASIA PANEL WITH GENOME WIDE CNV ANALYSIS INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS OROFACIAL CLEFTING PANEL WITH GENOME WIDE CNV ANALYSIS
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GLI3	99.3%	99.3%	100%	99.2%	Greig cephalopolysyndactyly syndrome, 175700;Polydactyly, postaxial, types A1 and B, 174200;Pallister-Hall syndrome, 146510;Polydactyly, preaxial, type IV, 174700	CRANIOFACIAL ANOMALIES PANEL WITH GENOME WIDE CNV ANALYSIS SHORT STATURE/SKELETAL DYSPLASIA PANEL WITH GENOME WIDE CNV ANALYSIS LIVER DISORDERS PANEL INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS RENAL DISORDERS PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS OROFACIAL CLEFTING PANEL WITH GENOME WIDE CNV ANALYSIS
GLIS1	100%	100%	100%	98.3%		CONGENITAL HEARTDISEASE PANEL ¹ HEART DISORDERS PANEL ¹ MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS

GLIS2	100%	100%	100%	98.7%	Nephronophthisis 7, 611498	CILIOPATHIES PANEL RENAL DISORDERS PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹
GLIS3	100%	100%	100%	99.2%	Diabetes mellitus, neonatal, with congenital hypothyroidism, 610199	LIVER DISORDERS PANEL INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS RENAL DISORDERS PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹
GLMN	100%	100%	100%	99.6%	Glomuvenous malformations, 138000	SKIN DISORDERS PANEL ¹ MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS

GLRA1	100%	100%	100%	99.5%	Hyperekplexia 1, 149400	EPILEPSY PANEL METABOLIC DISORDERS PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹
GLRA2	100%	99.9%	99.2%	68.9%	Intellectual developmental disorder, X-linked syndromic, Pilorge type, 301076	INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS
GLRB	100%	100%	100%	99.3%	Hyperekplexia 2, 614619	EPILEPSY PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹

GLRX5	100%	99.7%	100%	99.6%	Anemia, sideroblastic, 3, pyridoxine-refractory, 616860;Spasticity, childhood-onset, with hyperglycinemia, 616859	IRON DISORDERS PANEL METABOLIC DISORDERS PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS MITOCHONDRIAL DISORDERS PANEL COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹
GLS	100%	100%	100%	99.3%	Global developmental delay, progressive ataxia, and elevated glutamine, 618412;?Infantile cataract, skin abnormalities, glutamate excess, and impaired intellectual development, 618339;Developmental and epileptic encephalopathy 71, 618328	MOVEMENT DISORDERS PANEL EPILEPSY PANEL METABOLIC DISORDERS PANEL INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹

GLUD1	100%	100%	100%	99.2%	Hyperinsulinism-hyperammonemia syndrome, 606762	EPILEPSY PANEL METABOLIC DISORDERS PANEL INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS MITOCHONDRIAL DISORDERS PANEL
GLUL	100%	100%	100%	99.2%	Glutamine deficiency, congenital, 610015;Developmental and epileptic encephalopathy 116, 620806	EPILEPSY PANEL METABOLIC DISORDERS PANEL INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹

GLYCTK	100%	100%	100%	99.1%	D-glyceric aciduria, 220120	METABOLIC DISORDERS PANEL INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹
GLYR1	100%	100%	100%	98.8%		CONGENITAL HEARTDISEASE PANEL ¹ HEART DISORDERS PANEL ¹ MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS

GM2A	100%	100%	100%	98.8%	GM2-gangliosidosis, AB variant, 272750	METABOLIC DISORDERS PANEL INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹
GMNN	100%	100%	100%	99.3%	Meier-Gorlin syndrome 6, 616835	SHORT STATURE/SKELETAL DYSPLASIA PANEL WITH GENOME WIDE CNV ANALYSIS INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS

GMPPA	100%	100%	100%	98.8%	Alacrima, achalasia, and impaired intellectual development syndrome, 615510	SKIN DISORDERS PANEL ¹ METABOLIC DISORDERS PANEL INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹
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GMPPB	100%	100%	100%	99.2%	Muscular dystrophy-dystroglycanopathy (limb-girdle), type C, 14, 615352; Muscular dystrophy-dystroglycanopathy (congenital with impaired intellectual development), type B, 14, 615351; Muscular dystrophy-dystroglycanopathy (congenital with brain and eye anomalies), type A, 14, 615350	FETAL AKINESIA PANEL HEART DISORDERS PANEL ¹ METABOLIC DISORDERS PANEL INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹ MUSCLE DISORDERS PANEL
GMPR	100%	100%	100%	99.1%		MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS MITOCHONDRIAL DISORDERS PANEL
GMPS	100%	100%	100%	99.1%		METABOLIC DISORDERS PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS

GNA11	100%	100%	100%	98.5%	Hypocalciuric hypercalcemia, type II, 145981;Hypocalcemia, autosomal dominant 2, 615361	SKIN DISORDERS PANEL ¹ RENAL DISORDERS PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS
GNA14	100%	100%	100%	99.7%		SKIN DISORDERS PANEL ¹ MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS
GNAI1	100%	100%	100%	98.9%	Neurodevelopmental disorder with hypotonia, impaired speech, and behavioral abnormalities, 619854	INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS
GNAI2	100%	100%	100%	98.2%	Ventricular tachycardia, idiopathic, 192605;Pituitary adenoma, ACTH-secreting, somatic	PRIMARY IMMUNODEFICIENCIES PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS

GNAI3	100%	100%	100%	98.6%	Auriculocondylar syndrome 1, 602483	CRANIOFACIAL ANOMALIES PANEL WITH GENOME WIDE CNV ANALYSIS SHORT STATURE/SKELETAL DYSPLASIA PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS OROFACIAL CLEFTING PANEL WITH GENOME WIDE CNV ANALYSIS
GNAL	100%	100%	100%	98.7%	Dystonia 25, 615073	MOVEMENT DISORDERS PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS

GNAO1	100%	100%	100%	99.1%	Developmental and epileptic encephalopathy 17, 615473;Neurodevelopmental disorder with involuntary movements, 617493	MOVEMENT DISORDERS PANEL EPILEPSY PANEL INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS
GNAQ	100%	100%	100%	99.2%	Capillary malformations, congenital, 1, somatic, mosaic, 163000;Sturge-Weber syndrome, somatic, mosaic, 185300	SKIN DISORDERS PANEL ¹ MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS
GNAS	99.9%	98.1%	100%	97.1%	Pituitary adenoma 3, multiple types, somatic, 617686;Pseudohypoparathyroidism Ic, 612462;Pseudohypoparathyroidism Ia, 103580;Osseous heteroplasia, progressive, 166350;McCune-Albright syndrome, somatic mosaic, 174800;Pseudohypoparathyroidism Ib, 603233;Pseudopseudohypoparathyroidism, 612463;ACTH-independent macronodular adrenal hyperplasia 1, somatic, 219080	SKIN DISORDERS PANEL ¹ SHORT STATURE/SKELETAL DYSPLASIA PANEL WITH GENOME WIDE CNV ANALYSIS INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS

GNAS-AS1					Pseudohypoparathyroidism Ib, 603233	MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS
GNAT1	100%	100%	100%	98.9%	Night blindness, congenital stationary, autosomal dominant 3, 610444;Night blindness, congenital stationary, type 1G, 616389	VISION DISORDERS PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS
GNAT2	100%	100%	100%	99.5%	Achromatopsia 4, 613856	VISION DISORDERS PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹
GNB1	100%	100%	100%	99.1%	Myelodysplastic syndrome, somatic, 614286;Leukemia, acute lymphoblastic, somatic, 613065;Intellectual developmental disorder, autosomal dominant 42, 616973	INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS OROFACIAL CLEFTING PANEL WITH GENOME WIDE CNV ANALYSIS

GNB2	100%	100%	100%	99%	Neurodevelopmental disorder with hypotonia and dysmorphic facies, 619503;?Sick sinus syndrome 4, 619464	HEART DISORDERS PANEL ¹ INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS ARRHYTHMIA AND CARDIAC CONDUCTION DISORDERS PANEL ¹
GNB3	100%	100%	100%	98.3%	Night blindness, congenital stationary, type 1H, 617024;{Hypertension, essential, susceptibility to}, 145500	VISION DISORDERS PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹
GNB4	100%	100%	100%	99.2%	Charcot-Marie-Tooth disease, dominant intermediate F, 615185	POLYNEUROPATHIES PANEL ¹ MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS

GNB5	100%	100%	100%	99.2%	Lodder-Merla syndrome, type 2, with developmental delay and with or without cardiac arrhythmia, 617182;Lodder-Merla syndrome, type 1, with impaired intellectual development and cardiac arrhythmia, 617173	INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹
GNE	100%	100%	100%	99.3%	Sialuria, 269921;Thrombocytopenia 12 with or without myopathy, 620757;Nonaka myopathy, 605820	HEMOSTATIC/THROMB OTIC DISORDERS PANEL POLYNEUROPATHIES PANEL ¹ METABOLIC DISORDERS PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹ MUSCLE DISORDERS PANEL

GNMT	100%	100%	100%	94%	Glycine N-methyltransferase deficiency, 606664	METABOLIC DISORDERS PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹
GNPAT	100%	100%	100%	99.6%	Rhizomelic chondrodysplasia punctata, type 2, 222765	SHORT STATURE/SKELETAL DYSPLASIA PANEL WITH GENOME WIDE CNV ANALYSIS METABOLIC DISORDERS PANEL INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹

GNPNAT1	100%	100%	100%	99.2%	?Rhizomelic dysplasia, Ain-Naz type, 619598	SHORT STATURE/SKELETAL DYSPLASIA PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS
GNPTAB	100%	100%	100%	99.7%	Mucopolidosis III alpha/beta, 252600;Mucopolidosis II alpha/beta, 252500	CRANIOFACIAL ANOMALIES PANEL WITH GENOME WIDE CNV ANALYSIS HEART DISORDERS PANEL ¹ SHORT STATURE/SKELETAL DYSPLASIA PANEL WITH GENOME WIDE CNV ANALYSIS METABOLIC DISORDERS PANEL INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹

GNPTG	100%	100%	100%	98.3%	Mucopolidosis III gamma, 252605	VISION DISORDERS PANEL SHORT STATURE/SKELETAL DYSPLASIA PANEL WITH GENOME WIDE CNV ANALYSIS METABOLIC DISORDERS PANEL INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL¹
GNRH1	100%	100%	100%	99.6%	?Hypogonadotropic hypogonadism 12 with or without anosmia, 614841	DISORDERS/DIFFEREN CES OF SEX DEVELOPMENT (DSD) / PRIMARY ADRENAL INSUFFICIENCY PANEL HYPOGONADOTROPIC HYPOGONADISM (KALLMANN) PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS

GNRHR	100%	100%	100%	99.6%	Hypogonadotropic hypogonadism 7 without anosmia, 146110	DISORDERS/DIFFERENCES OF SEX DEVELOPMENT (DSD) / PRIMARY ADRENAL INSUFFICIENCY PANEL HYPOGONADOTROPIC HYPOGONADISM (KALLMANN) PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹
GNS	100%	100%	100%	99.3%	Mucopolysaccharidosis type IIID, 252940	SHORT STATURE/SKELETAL DYSPLASIA PANEL WITH GENOME WIDE CNV ANALYSIS METABOLIC DISORDERS PANEL INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹

GOLGA2	100%	99.9%	100%	99.2%	Developmental delay with hypotonia, myopathy, and brain abnormalities, 620240	INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹
GON7	100%	100%	100%	99.6%	Galloway-Mowat syndrome 9, 619603	MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹
GORAB	100%	100%	100%	99.3%	Geroderma osteodysplasticum, 231070	SKIN DISORDERS PANEL ¹ SHORT STATURE/SKELETAL DYSPLASIA PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹

GOSR2	100%	100%	100%	98.9%	Epilepsy, progressive myoclonic 6, 614018;Muscular dystrophy, congenital, with or without seizures, 620166	MOVEMENT DISORDERS PANEL HEARING IMPAIRMENT PANEL (INCLUDING GJB2) EPILEPSY PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL¹
GOT1	100%	100%	100%	99.4%	Aspartate aminotransferase, serum level of, QTL1, 614419	METABOLIC DISORDERS PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS

GOT2	100%	100%	100%	99.1%	Developmental and epileptic encephalopathy 82, 618721	EPILEPSY PANEL METABOLIC DISORDERS PANEL INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS MITOCHONDRIAL DISORDERS PANEL COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹
GP1BA	100%	100%	100%	96.3%	Bernard-Soulier syndrome, type A1 (recessive), 231200;Bernard-Soulier syndrome, type A2 (dominant), 153670;von Willebrand disease, platelet-type, 177820;{Nonarteritic anterior ischemic optic neuropathy, susceptibility to}, 258660	INHERITED BONE MARROW FAILURE AND/OR PREDISPOSITION PANEL TO HEMATOLOGICAL MALIGNANCIES HEMOSTATIC/THROMB OTIC DISORDERS PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹

GP1BB	100%	100%	100%	98.8%	Giant platelet disorder, isolated, 231200;Bernard-Soulier syndrome, type B, 231200	INHERITED BONE MARROW FAILURE AND/OR PREDISPOSITION PANEL TO HEMATOLOGICAL MALIGNANCIES HEMOSTATIC/THROMBOTIC DISORDERS PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹
GP6	99.8%	97.7%	100%	99.3%	Bleeding disorder, platelet-type, 11, 614201	HEMOSTATIC/THROMBOTIC DISORDERS PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹

GP9	100%	100%	100%	98.8%	Bernard-Soulier syndrome, type C, 231200	HEMOSTATIC/THROMBOTIC DISORDERS PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹
GPAA1	88.5%	88.5%	100%	99.1%	Glycosylphosphatidylinositol biosynthesis defect 15, 617810	INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹
GPAT2	99.9%	99.8%	99.5%	98.2%		MALE INFERTILITY PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS

GPATCH11	100%	100%	100%	97.6%		VISION DISORDERS PANEL INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS
GPC3	100%	99.9%	99%	69.4%	Wilms tumor, somatic, 194070;Simpson-Golabi-Behmel syndrome, type 1, 312870	EPILEPSY PANEL HEREDITARY CANCER PANEL TALL STATURE PANEL WITH GENOME WIDE CNV ANALYSIS SHORT STATURE/SKELETAL DYSPLASIA PANEL WITH GENOME WIDE CNV ANALYSIS INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS OROFACIAL CLEFTING PANEL WITH GENOME WIDE CNV ANALYSIS

GPC4	100%	99.9%	98.7%	67.7%	Keipert syndrome, 301026	SHORT STATURE/SKELETAL DYSPLASIA PANEL WITH GENOME WIDE CNV ANALYSIS INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS
GPC6	100%	100%	100%	99.4%	Omodysplasia 1, 258315	SHORT STATURE/SKELETAL DYSPLASIA PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹

GPD1	100%	100%	100%	99%	Hypertriglyceridemia, transient infantile, 614480	METABOLIC DISORDERS PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹
GPD1L	100%	100%	100%	99.4%	Brugada syndrome 2, 611777	HEART DISORDERS PANEL ¹ METABOLIC DISORDERS PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS
GPHN	100%	100%	100%	99.4%	Molybdenum cofactor deficiency C, 615501	EPILEPSY PANEL METABOLIC DISORDERS PANEL INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹

GPI	100%	100%	100%	98.1%	Anemia, congenital, nonspherocytic hemolytic, 4, glucose phosphate isomerase deficient, 613470	METABOLIC DISORDERS PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹
GPIHBP1	100%	100%	100%	97.9%	Hyperlipoproteinemia, type 1D, 615947	MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹ METABOLIC DISORDERS PANEL
GPNMB	95.1%	95.1%	100%	99.2%	Amyloidosis, primary localized cutaneous, 3, 617920	SKIN DISORDERS PANEL ¹ MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹

GPR101	100%	99.8%	97.5%	64.5%	Pituitary adenoma 2, GH-secreting, 300943	TALL STATURE PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS
GPR143	100%	99.8%	98.8%	68.6%	Ocular albinism, type I, Nettleship-Falls type, 300500;Nystagmus 6, congenital, X-linked, 300814	MOVEMENT DISORDERS PANEL VISION DISORDERS PANEL SKIN DISORDERS PANEL ¹ MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS
GPR156	100%	100%	100%	99.5%	Deafness, autosomal recessive 121, 620551	HEARING IMPAIRMENT PANEL (INCLUDING GJB2) MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹

GPR161	100%	100%	100%	99.1%	{Medulloblastoma predisposition syndrome}, 155255	SHORT STATURE/SKELETAL DYSPLASIA PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS SONIC HEDGEHOG MEDULLOBLASTOMA PANEL HEREDITARY CANCER PANEL
GPR179	100%	100%	100%	99.2%	Night blindness, congenital stationary (complete), 1E, autosomal recessive, 614565	VISION DISORDERS PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹
GPR68	100%	100%	100%	99.3%	Amelogenesis imperfecta, hypomaturation type, IIA6, 617217	CRANIOFACIAL ANOMALIES PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS

GPR88	99.8%	98.9%	100%	98.2%	?Chorea, childhood-onset, with psychomotor retardation, 616939	MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS
GPRASP2	100%	100%	99%	68.6%	?Deafness, X-linked 7, 301018	HEARING IMPAIRMENT PANEL (INCLUDING GJB2) MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS
GPRC5B	100%	100%	100%	98.9%	Megalencephalic leukoencephalopathy with subcortical cysts 3, 620447	INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS

GPSM2	95.5%	95.5%	100%	99.6%	Chudley-McCullough syndrome, 604213	HEARING IMPAIRMENT PANEL (INCLUDING GJB2) INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹
GPT2	100%	100%	100%	99.1%	Neurodevelopmental disorder with microcephaly and spastic paraplegia, 616281	METABOLIC DISORDERS PANEL INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS MITOCHONDRIAL DISORDERS PANEL COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹

GPX1	100%	100%	100%	98.5%	Hemolytic anemia due to glutathione peroxidase deficiency, 614164	METABOLIC DISORDERS PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS
GPX4	100%	99.7%	100%	98.5%	Spondylometaphyseal dysplasia, Sedaghatian type, 250220	SHORT STATURE/SKELETAL DYSPLASIA PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹
GRAP	100%	100%	100%	99.3%	Deafness, autosomal recessive 114, 618456	HEARING IMPAIRMENT PANEL (INCLUDING GJB2) MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹

GREB1L	100%	100%	100%	99.4%	Deafness, autosomal dominant 80, 619274;Renal hypodysplasia/aplasia 3, 617805	HEARING IMPAIRMENT PANEL (INCLUDING GJB2) DISORDERS/DIFFEREN CES OF SEX DEVELOPMENT (DSD) / PRIMARY ADRENAL INSUFFICIENCY PANEL RENAL DISORDERS PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS
GREM1	100%	100%	100%	99.3%		MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS HEREDITARY CANCER PANEL
GREM2	100%	100%	100%	98.2%	Tooth agenesis, selective, 9, 617275	MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS

GRHL2	100%	100%	100%	99.1%	Deafness, autosomal dominant 28, 608641;Ectodermal dysplasia/short stature syndrome, 616029;Corneal dystrophy, posterior polymorphous, 4, 618031	VISION DISORDERS PANEL INHERITED BONE MARROW FAILURE AND/OR PREDISPOSITION PANEL TO HEMATOLOGICAL MALIGNANCIES SKIN DISORDERS PANEL ¹ DYSKERATOSIS CONGENITA AND APLASTIC ANEMIA PANEL HEARING IMPAIRMENT PANEL (INCLUDING GJB2) PRIMARY IMMUNODEFICIENCIES PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹ HEREDITARY CANCER PANEL
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GRHL3	100%	99.9%	100%	98.8%	van der Woude syndrome 2, 606713	CRANIOFACIAL ANOMALIES PANEL WITH GENOME WIDE CNV ANALYSIS SKIN DISORDERS PANEL ¹ MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS OROFACIAL CLEFTING PANEL WITH GENOME WIDE CNV ANALYSIS
GRHPR	100%	100%	100%	98.5%	Hyperoxaluria, primary, type II, 260000	METABOLIC DISORDERS PANEL RENAL DISORDERS PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹
GRIA1	100%	100%	100%	98.9%	?Intellectual developmental disorder, autosomal recessive 76, 619931;Intellectual developmental disorder, autosomal dominant 67, 619927	INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS

GRIA2	100%	100%	100%	99.6%	Neurodevelopmental disorder with language impairment and behavioral abnormalities, 618917	INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS
GRIA3	100%	99.9%	98.8%	67.8%	Intellectual developmental disorder, X-linked syndromic, Wu type, 300699	EPILEPSY PANEL INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS
GRIA4	100%	100%	99.9%	98.7%	Neurodevelopmental disorder with or without seizures and gait abnormalities, 617864	INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS

GRID2	100%	100%	100%	99.5%	Spinocerebellar ataxia, autosomal recessive 18, 616204	MOVEMENT DISORDERS PANEL INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹
GRIK2	95.3%	95.3%	99.9%	97.8%	Neurodevelopmental disorder with impaired language and ataxia and with or without seizures, 619580;Intellectual developmental disorder, autosomal recessive 6, 611092	INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹

GRIN1	100%	100%	100%	98.1%	Neurodevelopmental disorder with or without hyperkinetic movements and seizures, autosomal recessive, 617820;Developmental and epileptic encephalopathy 101, 619814;Neurodevelopmental disorder with or without hyperkinetic movements and seizures, autosomal dominant, 614254	MOVEMENT DISORDERS PANEL EPILEPSY PANEL INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL¹ MUSCLE DISORDERS PANEL
GRIN2A	100%	100%	100%	98.9%	Epilepsy, focal, with speech disorder and with or without impaired intellectual development, 245570	EPILEPSY PANEL INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS

GRIN2B	100%	100%	100%	99.3%	Developmental and epileptic encephalopathy 27, 616139;Intellectual developmental disorder, autosomal dominant 6, with or without seizures, 613970	MOVEMENT DISORDERS PANEL EPILEPSY PANEL INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS
GRIN2D	98.7%	95.3%	99.9%	94.7%	Developmental and epileptic encephalopathy 46, 617162	EPILEPSY PANEL INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS

GRIP1	100%	100%	100%	99.3%	Fraser syndrome 3, 617667	COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹ DISORDERS/DIFFEREN CES OF SEX DEVELOPMENT (DSD) / PRIMARY ADRENAL INSUFFICIENCY PANEL INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS RENAL DISORDERS PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS
GRK1	100%	100%	100%	99%	Oguchi disease-2, 613411	MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS VISION DISORDERS PANEL COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹

GRM1	100%	100%	100%	99.5%	Spinocerebellar ataxia, autosomal recessive 13, 614831;Spinocerebellar ataxia 44, 617691	MOVEMENT DISORDERS PANEL INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL¹
GRM6	100%	99.7%	100%	98.8%	Night blindness, congenital stationary (complete), 1B, autosomal recessive, 257270	VISION DISORDERS PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL¹

GRM7	100%	100%	100%	98.3%	Neurodevelopmental disorder with seizures, hypotonia, and brain abnormalities, 618922	INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹
GRN	100%	100%	100%	99.6%	Frontotemporal dementia 2, 607485;Aphasia, primary progressive, 607485;Ceroid lipofuscinosis, neuronal, 11, 614706	AMYOTROPHIC LATERAL SCLEROSIS, ALS PANEL MOVEMENT DISORDERS PANEL EPILEPSY PANEL INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS PARKINSON DISEASE PANEL COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹

GRXCR1	100%	100%	100%	99.6%	Deafness, autosomal recessive 25, 613285	HEARING IMPAIRMENT PANEL (INCLUDING GJB2) MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL¹
GRXCR2	100%	100%	100%	99.3%	?Deafness, autosomal recessive 101, 615837	HEARING IMPAIRMENT PANEL (INCLUDING GJB2) MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS

GSC	100%	100%	100%	97%	Short stature, auditory canal atresia, mandibular hypoplasia, skeletal abnormalities, 602471	CRANIOFACIAL ANOMALIES PANEL WITH GENOME WIDE CNV ANALYSIS SHORT STATURE/SKELETAL DYSPLASIA PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹
GSDME	100%	100%	100%	99%	Deafness, autosomal dominant 5, 600994	HEARING IMPAIRMENT PANEL (INCLUDING GJB2) MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS
GSE1	100%	100%	100%	98.7%		INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS

GSN	100%	100%	100%	98.7%	Amyloidosis, Finnish type, 105120	VISION DISORDERS PANEL SKIN DISORDERS PANEL ¹ POLYNEUROPATHIES PANEL ¹ RENAL DISORDERS PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS
GSR	100%	100%	100%	98.8%	Anemia, congenital, nonspherocytic hemolytic, 10, glutathione reductase deficient, 618660	MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS
GSS	100%	100%	100%	98.7%	Anemia, congenital, nonspherocytic hemolytic, 6, glutathione synthetase deficient, 266130;Glutathione synthetase deficiency, 266130	METABOLIC DISORDERS PANEL INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹

GSX2	100%	100%	100%	98.8%	Diencephalic-mesencephalic junction dysplasia syndrome 2, 618646	MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹
GTF2E2	100%	100%	100%	99.7%	Trichothiodystrophy 6, nonphotosensitive, 616943	SKIN DISORDERS PANEL ¹ INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹

GTF2H5	59.2%	59.2%	100%	99.3%	Trichothiodystrophy 3, photosensitive, 616395	SKIN DISORDERS PANEL ¹ PRIMARY IMMUNODEFICIENCIES PANEL INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹
GTF3C3	100%	100%	100%	99.3%		INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹

GTF3C5	100%	99.8%	100%	99%		INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS
GTPBP1	100%	100%	100%	98.2%	Neurodevelopmental disorder with characteristic facial and ectodermal features and tetraparesis 1, 620888	INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹
GTPBP2	100%	100%	100%	99.1%	Jaberi-Elahi syndrome, 617988	INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS MITOCHONDRIAL DISORDERS PANEL COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹

GTPBP3	100%	100%	100%	97.6%	Combined oxidative phosphorylation deficiency 23, 616198	INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS MITOCHONDRIAL DISORDERS PANEL COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹
GUCA1A	100%	100%	100%	98.8%	Cone-rod dystrophy 14, 602093;Cone dystrophy-3, 602093	VISION DISORDERS PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS
GUCA1B	100%	100%	100%	98.3%	Retinitis pigmentosa 48, 613827	VISION DISORDERS PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS

GUCY1A1	100%	100%	100%	99.7%	Moyamoya 6 with achalasia, 615750	MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹
GUCY2C	100%	100%	100%	99.4%	Diarrhea 6, 614616;Meconium ileus, 614665	MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹
GUCY2D	100%	100%	100%	98.8%	Cone-rod dystrophy 6, 601777;?Choroidal dystrophy, central areolar 1, 215500;Leber congenital amaurosis 1, 204000;Night blindness, congenital stationary, type 1I, 618555	VISION DISORDERS PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹
GUF1	100%	100%	100%	99.3%	?Developmental and epileptic encephalopathy 40, 617065	MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS MITOCHONDRIAL DISORDERS PANEL

GUK1	100%	100%	100%	98.9%		MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS MITOCHONDRIAL DISORDERS PANEL COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹
GULOP					Scurvy	MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS
GUSB	100%	100%	100%	98.8%	Mucopolysaccharidosis VII, 253220	SHORT STATURE/SKELETAL DYSPLASIA PANEL WITH GENOME WIDE CNV ANALYSIS METABOLIC DISORDERS PANEL INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹

GYG1	100%	100%	100%	99.5%	?Glycogen storage disease XV, 613507;Polyglucosan body myopathy 2, 616199	METABOLIC DISORDERS PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹ MUSCLE DISORDERS PANEL
GYS1	100%	100%	100%	99%	Glycogen storage disease 0, muscle, 611556	METABOLIC DISORDERS PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹ MUSCLE DISORDERS PANEL
GYS2	100%	100%	100%	99.6%	Glycogen storage disease 0, liver, 240600	METABOLIC DISORDERS PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹

GZF1	100%	100%	100%	99.5%	Joint laxity, short stature, and myopia, 617662	VISION DISORDERS PANEL SHORT STATURE/SKELETAL DYSPLASIA PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹
H1-4	100%	100%	100%	98.9%	Rahman syndrome, 617537	INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS

H19						TALL STATURE PANEL WITH GENOME WIDE CNV ANALYSIS SHORT STATURE/SKELETAL DYSPLASIA PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS
H3-3A	100%	100%	100%	99.6%	Bryant-Li-Bhoj neurodevelopmental syndrome 1, 619720	INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS
H3-3B	100%	100%	100%	99.1%	Bryant-Li-Bhoj neurodevelopmental syndrome 2, 619721	INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS

H4C11	100%	100%	100%	97.8%	?Tessadori-Bicknell-van Haaften neurodevelopmental syndrome 2, 619759	MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS
H4C3	100%	100%	100%	97.1%	Tessadori-Bicknell-van Haaften neurodevelopmental syndrome 1, 619758	INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS
H4C5	100%	100%	100%	99.5%	Tessadori-Bicknell-van Haaften neurodevelopmental syndrome 3, 619950	INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS
H4C9	100%	100%	100%	99.2%	Tessadori-Bicknell-van Haaften neurodevelopmental syndrome 4, 619951	INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS

H6PD	100%	100%	100%	99.5%	Cortisone reductase deficiency 1, 604931	METABOLIC DISORDERS PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹
HAAO	100%	100%	100%	99.7%	Vertebral, cardiac, renal, and limb defects syndrome 1, 617660	SHORT STATURE/SKELETAL DYSPLASIA PANEL WITH GENOME WIDE CNV ANALYSIS INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹
HABP2	100%	100%	100%	99.4%	{?Thyroid cancer, nonmedullary, 5}, 616535;{Venous thromboembolism, susceptibility to}, 188050	HEMOSTATIC/THROMB OTIC DISORDERS PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS

HACD1	80.3%	80.3%	100%	99%	Congenital myopathy 11, 619967	MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹ MUSCLE DISORDERS PANEL
HACE1	100%	100%	100%	98.6%	Spastic paraplegia and psychomotor retardation with or without seizures, 616756	MOVEMENT DISORDERS PANEL EPILEPSY PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS MITOCHONDRIAL DISORDERS PANEL INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹

HADH	100%	100%	100%	99.3%	Hyperinsulinemic hypoglycemia, familial, 4, 609975;3-hydroxyacyl-CoA dehydrogenase deficiency, 231530	EPILEPSY PANEL COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL¹ MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS METABOLIC DISORDERS PANEL INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS
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HADHA	100%	100%	100%	99.6%	HELLP syndrome, maternal, of pregnancy, 609016;LCHAD deficiency, 609016;Mitochondrial trifunctional protein deficiency 1, 609015;Fatty liver, acute, of pregnancy, 609016	COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹ MUSCLE DISORDERS PANEL MITOCHONDRIAL DISORDERS PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS METABOLIC DISORDERS PANEL LIVER DISORDERS PANEL POLYNEUROPATHIES PANEL ¹ HEART DISORDERS PANEL ¹
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HADHB	100%	100%	100%	98.9%	Mitochondrial trifunctional protein deficiency 2, 620300	MITOCHONDRIAL DISORDERS PANEL COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹ POLYNEUROPATHIES PANEL ¹ METABOLIC DISORDERS PANEL INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS HEART DISORDERS PANEL ¹ MUSCLE DISORDERS PANEL
HAGH	100%	99.3%	100%	98.4%	[Glyoxalase II deficiency], 614033	METABOLIC DISORDERS PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS

HAMP	100%	99.9%	100%	99.1%	Hemochromatosis, type 2B, 613313	IRON DISORDERS PANEL LIVER DISORDERS PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹
HAND1	100%	100%	100%	98.6%		CONGENITAL HEARTDISEASE PANEL ¹ HEART DISORDERS PANEL ¹ MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS
HAND2	100%	100%	99.7%	90.4%		CONGENITAL HEARTDISEASE PANEL ¹ HEART DISORDERS PANEL ¹ MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS

HARS1	100%	100%	100%	99.4%	Charcot-Marie-Tooth disease, axonal, type 2W, 616625;Usher syndrome type 3B, 614504	VISION DISORDERS PANEL HEARING IMPAIRMENT PANEL (INCLUDING GJB2) POLYNEUROPATHIES PANEL ¹ MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹
HARS2	100%	100%	100%	99%	Perrault syndrome 2, 614926	HEARING IMPAIRMENT PANEL (INCLUDING GJB2) PREMATURE OVARIAN INSUFFICIENCY PANEL DISORDERS/DIFFEREN CES OF SEX DEVELOPMENT (DSD) / PRIMARY ADRENAL INSUFFICIENCY PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS MITOCHONDRIAL DISORDERS PANEL COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹

HAVCR2	100%	100%	100%	99.5%	T-cell lymphoma, subcutaneous panniculitis-like, 618398	INHERITED BONE MARROW FAILURE AND/OR PREDISPOSITION PANEL TO HEMATOLOGICAL MALIGNANCIES PRIMARY IMMUNODEFICIENCIES PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹ HEREDITARY CANCER PANEL
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HAX1	100%	100%	100%	98.7%	Neutropenia, severe congenital 3, autosomal recessive, 610738	INHERITED BONE MARROW FAILURE AND/OR PREDISPOSITION PANEL TO HEMATOLOGICAL MALIGNANCIES PRIMARY IMMUNODEFICIENCIES PANEL INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹ HEREDITARY CANCER PANEL
HBA1	100%	100%	100%	99.9%	Hemoglobin H disease, nondeletional, 613978;Thalassemias, alpha-, 604131;Heinz body anemias, alpha-, 140700;Methemoglobinemia, alpha type, 617973;Erythrocytosis, familial, 7, 617981	MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS
HBA2	100%	100%	97.5%	87.7%	Heinz body anemia, 140700;Thalassemia, alpha-, 604131;Erythrocytosis, familial, 7, 617981;Hemoglobin H disease, deletional and nondeletional, 613978	MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS

HBB	100%	100%	100%	99.5%	Methemoglobinemia, beta type, 617971;Thalassemia-beta, dominant inclusion-body, 603902;Sickle cell disease, 603903;Thalassemia, beta, 613985;Delta-beta thalassemia, 141749;{Malaria, resistance to}, 611162;Hereditary persistence of fetal hemoglobin, 141749;Erythrocytosis, familial, 6, 617980;Heinz body anemia, 140700	MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹
HBD	100%	100%	100%	99.8%	Thalassemia due to Hb Lepore;Thalassemia, delta-	MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS
HBG1	96.3%	92.1%	91.5%	73.1%	Fetal hemoglobin quantitative trait locus 1, 141749	MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS
HBG2	100%	100%	100%	99.4%	Fetal hemoglobin quantitative trait locus 1, 141749;Cyanosis, transient neonatal, 613977	MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS

HCCS	100%	99.9%	99.1%	69.7%	Linear skin defects with multiple congenital anomalies 1, 309801	VISION DISORDERS PANEL SKIN DISORDERS PANEL ¹ INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS MITOCHONDRIAL DISORDERS PANEL
HCFC1	100%	99.7%	98%	66.2%	Methylmalonic aciduria and homocysteinemia, cblX type, 309541	EPILEPSY PANEL METABOLIC DISORDERS PANEL INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS
HCK	100%	100%	100%	98.7%	Autoinflammation with pulmonary and cutaneous vasculitis, 620296	PRIMARY IMMUNODEFICIENCIES PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS

HCN1	100%	100%	100%	98.9%	Developmental and epileptic encephalopathy 24, 615871;Generalized epilepsy with febrile seizures plus, type 10, 618482	EPILEPSY PANEL NEUROLOGICAL PAIN DISORDERS PANEL¹ INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS
HCN2	90.7%	85.3%	97.7%	83.3%	Febrile seizures, familial, 2, 602477;{Epilepsy, idiopathic generalized, susceptibility to, 17}, 602477;Generalized epilepsy with febrile seizures plus, type 11, 602477	EPILEPSY PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS HEART DISORDERS PANEL¹ NEUROLOGICAL PAIN DISORDERS PANEL¹ INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS
HCN3	100%	99.9%	100%	98.8%		MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS HEART DISORDERS PANEL¹ NEUROLOGICAL PAIN DISORDERS PANEL¹

HCN4	100%	100%	100%	98.2%	Sick sinus syndrome 2, 163800;{Epilepsy, idiopathic generalized, susceptibility to, 18}, 619521;Brugada syndrome 8, 613123	THORACIC AORTIC ANEURYSM AND/OR DISSECTION PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS ARRHYTHMIA AND CARDIAC CONDUCTION DISORDERS PANEL ¹ DILATED CARDIOMYOPATHY PANEL ¹ HEART DISORDERS PANEL ¹
HCRT	100%	100%	100%	95.3%	?Narcolepsy 1, 161400	MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS
HDAC3	100%	100%	100%	99.1%		INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS

HDAC4	100%	100%	100%	98.6%	Neurodevelopmental disorder with central hypotonia and dysmorphic facies, 619797	SHORT STATURE/SKELETAL DYSPLASIA PANEL WITH GENOME WIDE CNV ANALYSIS INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS
HDAC6	100%	99.8%	98.8%	67.8%	?Chondrodysplasia with platyspondyly, distinctive brachydactyly, hydrocephaly, and microphthalmia, 300863	INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS

HDAC8	97.6%	97.2%	99%	68.6%	Cornelia de Lange syndrome 5, 300882	SKIN DISORDERS PANEL ¹ SHORT STATURE/SKELETAL DYSPLASIA PANEL WITH GENOME WIDE CNV ANALYSIS INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS OROFACIAL CLEFTING PANEL WITH GENOME WIDE CNV ANALYSIS
HDAC9	100%	100%	100%	99.2%	?Auriculocondylar syndrome 4, 620457	MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS

HEATR3	100%	100%	100%	99.6%	Diamond-Blackfan anemia 21, 620072	INHERITED BONE MARROW FAILURE AND/OR PREDISPOSITION PANEL TO HEMATOLOGICAL MALIGNANCIES INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹
HEATR5B	100%	100%	100%	99.3%		INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS

HECTD1	100%	100%	100%	99.6%		CONGENITAL HEARTDISEASE PANEL¹ HEART DISORDERS PANEL¹ INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS
HECTD4	100%	100%	100%	99.2%	Neurodevelopmental disorder with seizures, spasticity, and complete or partial agenesis of the corpus callosum, 620250	INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL¹
HECW2	100%	100%	100%	99.1%	Neurodevelopmental disorder with hypotonia, seizures, and absent language, 617268	EPILEPSY PANEL INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS

HELLS	100%	100%	100%	99.6%	Immunodeficiency-centromeric instability-facial anomalies syndrome 4, 616911	PRIMARY IMMUNODEFICIENCIES PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹
HENMT1	92.1%	92.1%	100%	99.5%		MALE INFERTILITY PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS
HEPACAM	100%	100%	100%	99.1%	Megalencephalic leukoencephalopathy with subcortical cysts 2A, 613925;Megalencephalic leukoencephalopathy with subcortical cysts 2B, remitting, with or without impaired intellectual development, 613926	INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹

HEPHL1	100%	100%	100%	98.9%	?Abnormal hair, joint laxity, and developmental delay, 261990	MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS
HERC1	100%	100%	100%	99.4%	Macrocephaly, dysmorphic facies, and psychomotor retardation, 617011	TALL STATURE PANEL WITH GENOME WIDE CNV ANALYSIS INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹
HERC2	100%	99.9%	100%	99.1%	Intellectual developmental disorder, autosomal recessive 38, 615516;[Skin/hair/eye pigmentation 1, blond/brown hair], 227220;[Skin/hair/eye pigmentation 1, blue/nonblue eyes], 227220	SKIN DISORDERS PANEL ¹ INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹

HES7	100%	100%	100%	93.9%	Spondylocostal dysostosis 4, autosomal recessive, 613686	SHORT STATURE/SKELETAL DYSPLASIA PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹
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HESX1	100%	100%	100%	99.5%	Pituitary hormone deficiency, combined, 5, 182230;Septo-optic dysplasia, 182230;Growth hormone deficiency with pituitary anomalies, 182230	DISORDERS/DIFFERENCES OF SEX DEVELOPMENT (DSD) / PRIMARY ADRENAL INSUFFICIENCY PANEL HYPOGONADOTROPIC HYPOGONADISM (KALLMANN) PANEL SHORT STATURE/SKELETAL DYSPLASIA PANEL WITH GENOME WIDE CNV ANALYSIS INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL¹
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HEXA	100%	100%	100%	99.4%	[Hex A pseudodeficiency], 272800;GM2-gangliosidosis, several forms, 272800;Tay-Sachs disease, 272800	METABOLIC DISORDERS PANEL INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹
HEXB	100%	100%	100%	98.5%	Sandhoff disease, infantile, juvenile, and adult forms, 268800	MOVEMENT DISORDERS PANEL METABOLIC DISORDERS PANEL INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹

HEY2	100%	100%	100%	97%		CONGENITAL HEARTDISEASE PANEL¹ HEART DISORDERS PANEL¹ MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS
HFE	100%	100%	100%	99%	Hemochromatosis, type 1, 235200	HEART DISORDERS PANEL¹ IRON DISORDERS PANEL LIVER DISORDERS PANEL METABOLIC DISORDERS PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS

HFM1	100%	100%	100%	99.7%	Premature ovarian failure 9, 615724	DISORDERS/DIFFERENCES OF SEX DEVELOPMENT (DSD) / PRIMARY ADRENAL INSUFFICIENCY PANEL MALE INFERTILITY PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL¹ PREMATURE OVARIAN INSUFFICIENCY PANEL
HGD	100%	100%	100%	99.5%	Alkaptonuria, 203500	METABOLIC DISORDERS PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL¹

HGF	100%	100%	100%	99.6%	Deafness, autosomal recessive 39, 608265	HEARING IMPAIRMENT PANEL (INCLUDING GJB2) MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹
HGSNAT	92.4%	92.3%	100%	99.4%	Mucopolysaccharidosis type IIIC (Sanfilippo C), 252930;Retinitis pigmentosa 73, 616544	VISION DISORDERS PANEL SHORT STATURE/SKELETAL DYSPLASIA PANEL WITH GENOME WIDE CNV ANALYSIS METABOLIC DISORDERS PANEL INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹

HHAT	100%	100%	100%	99.2%	Nivelon-Nivelon-Mabille syndrome, 600092	DISORDERS/DIFFERENCES OF SEX DEVELOPMENT (DSD) / PRIMARY ADRENAL INSUFFICIENCY PANEL SHORT STATURE/SKELETAL DYSPLASIA PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS
HIBADH	100%	100%	100%	99%		METABOLIC DISORDERS PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS

HIBCH	100%	100%	100%	99.8%	3-hydroxyisobutryl-CoA hydrolase deficiency, 250620	METABOLIC DISORDERS PANEL INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL¹ MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS MITOCHONDRIAL DISORDERS PANEL
HID1	100%	100%	100%	99%	Developmental and epileptic encephalopathy 105 with hypopituitarism, 619983	INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL¹

HIKESHI	100%	100%	100%	98.7%	Leukodystrophy, hypomyelinating, 13, 616881	INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹
HINT1	100%	100%	100%	99.4%	Neuromyotonia and axonal neuropathy, autosomal recessive, 137200	POLYNEUROPATHIES PANEL ¹ MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹
HIVEP2	100%	100%	100%	99.3%	Intellectual developmental disorder, autosomal dominant 43, 616977	INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS

HJV	100%	100%	100%	98.9%	Hemochromatosis, type 2A, 602390	HEART DISORDERS PANEL ¹ IRON DISORDERS PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹
HK1	100%	100%	100%	99.1%	Anemia, congenital, nonspherocytic hemolytic, 5, hexokinase deficient, 235700;Retinitis pigmentosa 79, 617460;Neuropathy, hereditary motor and sensory, Russe type, 605285;Neurodevelopmental disorder with visual defects and brain anomalies, 618547	MOVEMENT DISORDERS PANEL VISION DISORDERS PANEL POLYNEUROPATHIES PANEL ¹ METABOLIC DISORDERS PANEL INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹

HKDC1	100%	100%	100%	98.9%	Retinitis pigmentosa 92, 619614	VISION DISORDERS PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS
HLCS	100%	99.9%	100%	99.3%	Holocarboxylase synthetase deficiency, 253270	SKIN DISORDERS PANEL ¹ EPILEPSY PANEL METABOLIC DISORDERS PANEL INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS MITOCHONDRIAL DISORDERS PANEL COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹
HLX	100%	100%	100%	98.9%		MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹

HMBS	100%	100%	100%	99.3%	Leukoencephalopathy, porphyria-related, 620711;Encephalopathy, porphyria-related, 620704;Porphyria, acute intermittent, nonerythroid variant, 176000;Porphyria, acute intermittent, 176000	SKIN DISORDERS PANEL ¹ POLYNEUROPATHIES PANEL ¹ METABOLIC DISORDERS PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹
HMCN1	100%	100%	100%	99.3%	{Macular degeneration, age-related, 1}, 603075	SKIN DISORDERS PANEL ¹ MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS
HMGA2	94.5%	83.9%	100%	99.5%	Silver-Russell syndrome 5, 618908	SHORT STATURE/SKELETAL DYSPLASIA PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS

HMGB1	100%	100%	100%	98.7%		INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS
HMGB3	100%	99.8%	99%	67.9%	?Microphthalmia, syndromic 13, 300915	VISION DISORDERS PANEL SKIN DISORDERS PANEL ¹ MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS
HMGCL	100%	100%	100%	99.3%	HMG-CoA lyase deficiency, 246450	METABOLIC DISORDERS PANEL INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹

HMGCRCR	100%	100%	100%	99.3%	Muscular dystrophy, limb-girdle, autosomal recessive 28, 620375;[Statins, response to], 620410;[Low density lipoprotein cholesterol level QTL 3], 620410	METABOLIC DISORDERS PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS MUSCLE DISORDERS PANEL COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹
HMGCS2	100%	100%	100%	99.4%	HMG-CoA synthase-2 deficiency, 605911	COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹ LIVER DISORDERS PANEL METABOLIC DISORDERS PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS

HMOX1	100%	100%	100%	99.4%	Heme oxygenase-1 deficiency, 614034;{Pulmonary disease, chronic obstructive, susceptibility to}, 606963	IRON DISORDERS PANEL PRIMARY IMMUNODEFICIENCIES PANEL METABOLIC DISORDERS PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹
HMX1	100%	99.8%	100%	97.1%	Oculoauricular syndrome, 612109	VISION DISORDERS PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹
HNF1A	100%	100%	100%	98.8%	Hepatic adenoma, somatic, 142330;Diabetes mellitus, insulin-dependent, 20, 612520;{Diabetes mellitus, noninsulin-dependent, 2}, 125853;MODY, type III, 600496;{Diabetes mellitus, insulin-dependent}, 222100;Renal cell carcinoma, 144700	METABOLIC DISORDERS PANEL RENAL DISORDERS PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS

HNFB	100%	100%	100%	99.2%	Type 2 diabetes mellitus, 125853;Renal cysts and diabetes syndrome, 137920;{Renal cell carcinoma}, 144700	LIVER DISORDERS PANEL RENAL DISORDERS PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS
HNFB	100%	100%	100%	98.8%	Fanconi renotubular syndrome 4, with maturity-onset diabetes of the young, 616026;{Diabetes mellitus, noninsulin-dependent}, 125853;MODY, type I, 125850	METABOLIC DISORDERS PANEL RENAL DISORDERS PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS
HNMT	100%	100%	100%	99.6%	Intellectual developmental disorder, autosomal recessive 51, 616739;{Asthma, susceptibility to}, 600807	INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹

HNRNPA1	100%	100%	100%	99.3%	?Inclusion body myopathy with early-onset Paget disease without frontotemporal dementia 3, 615424;?Myopathy, distal, 3, 610099;Amyotrophic lateral sclerosis 20, 615426	POLYNEUROPATHIES PANEL ¹ MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS MUSCLE DISORDERS PANEL
HNRNPA2B1	100%	100%	100%	99.3%	Oculopharyngeal muscular dystrophy 2, 620460;?Inclusion body myopathy with early-onset Paget disease with or without frontotemporal dementia 2, 615422	MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS MUSCLE DISORDERS PANEL
HNRNPC	100%	100%	100%	99.3%	Intellectual developmental disorder, autosomal dominant 74, 620688	INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS
HNRNPD	100%	100%	100%	99.3%		INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS

HNRNPDL	100%	100%	99.7%	94.4%	Muscular dystrophy, limb-girdle, autosomal dominant 3, 609115	MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS MUSCLE DISORDERS PANEL
HNRNPH1	100%	100%	100%	98.8%	Neurodevelopmental disorder with craniofacial dysmorphism and skeletal defects, 620083	INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS
HNRNPH2	100%	100%	98.3%	68%	Intellectual developmental disorder, X-linked syndromic, Bain type, 300986	INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS
HNRNPK	100%	100%	100%	99.7%	Au-Kline syndrome, 616580	INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS

HNRNPR	100%	100%	100%	99.3%	Neurodevelopmental disorder with dysmorphic facies and skeletal and brain abnormalities, 620073	INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS
HNRNPU	100%	100%	100%	97.6%	Developmental and epileptic encephalopathy 54, 617391	EPILEPSY PANEL INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS
HOGA1	100%	100%	100%	99.1%	Hyperoxaluria, primary, type III, 613616	METABOLIC DISORDERS PANEL RENAL DISORDERS PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL¹

HOMER2	100%	100%	100%	99.3%	?Deafness, autosomal dominant 68, 616707	HEARING IMPAIRMENT PANEL (INCLUDING GJB2) MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS
HOXA1	100%	100%	100%	99.2%	Bosley-Salih-Alorainy syndrome, 601536;Athabaskan brainstem dysgenesis syndrome, 601536	INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹

HOXA11	100%	100%	100%	98.2%	Radioulnar synostosis with amegakaryocytic thrombocytopenia 1, 605432	INHERITED BONE MARROW FAILURE AND/OR PREDISPOSITION PANEL TO HEMATOLOGICAL MALIGNANCIES HEMOSTATIC/THROMBOTIC DISORDERS PANEL SHORT STATURE/SKELETAL DYSPLASIA PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS
HOXA13	97.4%	89.4%	99%	82.7%	Hand-foot-genital syndrome, 140000;?Guttmacher syndrome, 176305	MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS DISORDERS/DIFFERENCES OF SEX DEVELOPMENT (DSD) / PRIMARY ADRENAL INSUFFICIENCY PANEL SHORT STATURE/SKELETAL DYSPLASIA PANEL WITH GENOME WIDE CNV ANALYSIS

HOXA2	100%	100%	100%	98.5%	Microtia with or without hearing impairment (AD), 612290;?Microtia, hearing impairment, and cleft palate (AR), 612290	CRANIOFACIAL ANOMALIES PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹
HOXB1	100%	100%	100%	99.4%	Facial paresis, hereditary congenital, 3, 614744	MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹
HOXB13	100%	100%	100%	99.6%	{Prostate cancer, hereditary, 9}, 610997	MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS HEREDITARY CANCER PANEL

HOXC13	100%	100%	100%	97.6%	Ectodermal dysplasia 9, hair/nail type, 614931	SKIN DISORDERS PANEL ¹ MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹
HOXD10	100%	100%	99.8%	96.1%	Vertical talus, congenital, 192950;Charcot-Marie-Tooth disease, foot deformity of, 192950	POLYNEUROPATHIES PANEL ¹ MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS
HOXD13	100%	100%	100%	98.2%	Syndactyly, type V, 186300;Synpolydactyly 1, 186000;Brachydactyly, type E, 113300;Brachydactyly, type D, 113200;?Brachydactyly-syndactyly syndrome, 610713	SHORT STATURE/SKELETAL DYSPLASIA PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS

HPCA	100%	100%	100%	98.5%	Dystonia 2, torsion, autosomal recessive, 224500	MOVEMENT DISORDERS PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL¹
HPD	100%	100%	100%	97.3%	Hawkinsinuria, 140350;Tyrosinemia, type III, 276710	METABOLIC DISORDERS PANEL INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL¹

HPDL	100%	100%	100%	99.8%	Neurodevelopmental disorder with progressive spasticity and brain white matter abnormalities, 619026;Spastic paraplegia 83, autosomal recessive, 619027	MOVEMENT DISORDERS PANEL METABOLIC DISORDERS PANEL INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS MITOCHONDRIAL DISORDERS PANEL COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹
HPGD	100%	100%	100%	99.4%	?Digital clubbing, isolated congenital, 119900;Hypertrophic osteoarthropathy, primary, autosomal recessive 1, 259100;Cranioosteoarthropathy, 259100	SKIN DISORDERS PANEL ¹ SHORT STATURE/SKELETAL DYSPLASIA PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹

HPRT1	100%	99.8%	99%	69.4%	Hyperuricemia, HRPT-related, 300323;Lesch-Nyhan syndrome, 300322	MOVEMENT DISORDERS PANEL METABOLIC DISORDERS PANEL INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS RENAL DISORDERS PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS
HPS1	100%	100%	100%	99.1%	Hermansky-Pudlak syndrome 1, 203300	VISION DISORDERS PANEL SKIN DISORDERS PANEL ¹ HEMOSTATIC/THROMB OTIC DISORDERS PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹

HPS3	100%	100%	100%	99.5%	Hermansky-Pudlak syndrome 3, 614072	VISION DISORDERS PANEL SKIN DISORDERS PANEL ¹ HEMOSTATIC/THROMB OTIC DISORDERS PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹
HPS4	100%	100%	100%	99.5%	Hermansky-Pudlak syndrome 4, 614073	VISION DISORDERS PANEL SKIN DISORDERS PANEL ¹ HEMOSTATIC/THROMB OTIC DISORDERS PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹

HPS5	100%	100%	100%	99.5%	Hermansky-Pudlak syndrome 5, 614074	VISION DISORDERS PANEL SKIN DISORDERS PANEL ¹ HEMOSTATIC/THROMB OTIC DISORDERS PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹
HPS6	100%	100%	100%	99.5%	Hermansky-Pudlak syndrome 6, 614075	VISION DISORDERS PANEL SKIN DISORDERS PANEL ¹ HEMOSTATIC/THROMB OTIC DISORDERS PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹

HPSE2	100%	100%	100%	99%	Urofacial syndrome 1, 236730	RENAL DISORDERS PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹
HR	100%	100%	100%	99.4%	Atrichia with papular lesions, 209500;Alopecia universalis, 203655	SKIN DISORDERS PANEL ¹ MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹

HRAS	100%	100%	100%	99.3%	Bladder cancer, somatic, 109800;Thyroid carcinoma, follicular, somatic, 188470;Congenital myopathy with excess of muscle spindles, 218040;Nevus sebaceous or woolly hair nevus, somatic, 162900;Schimmelpenning-Feuerstein-Mims syndrome, somatic mosaic, 163200;Spitz nevus or nevus spilus, somatic, 137550;Costello syndrome, 218040	VISION DISORDERS PANEL SKIN DISORDERS PANEL ¹ SHORT STATURE/SKELETAL DYSPLASIA PANEL WITH GENOME WIDE CNV ANALYSIS INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS NOONAN SYNDROME / RASOPATHY PANEL LYMPHATIC ANOMALIES PANEL WITH GENOME WIDE CNV ANALYSIS
HRG	100%	100%	100%	99.2%	Thrombophilia 11 due to HRG deficiency, 613116	HEMOSTATIC/THROMB OTIC DISORDERS PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS

HROB	100%	100%	100%	98.9%	Ovarian dysgenesis 11, 620897	DISORDERS/DIFFERENCES OF SEX DEVELOPMENT (DSD) / PRIMARY ADRENAL INSUFFICIENCY PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS PREMATURE OVARIAN INSUFFICIENCY PANEL
HRURF	100%	100%	100%	99.8%	Hypotrichosis 4, 146550	MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS
HS2ST1	100%	100%	100%	99.4%	Neurofacioskeletal syndrome with or without renal agenesis, 619194	SHORT STATURE/SKELETAL DYSPLASIA PANEL WITH GENOME WIDE CNV ANALYSIS INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹

HS3ST6	99.3%	96.5%	100%	96.7%	?Angioedema, hereditary, 8, 619367	PRIMARY IMMUNODEFICIENCIES PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS
HS6ST1	100%	100%	100%	97.9%	{Hypogonadotropic hypogonadism 15 with or without anosmia}, 614880	MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS DISORDERS/DIFFEREN CES OF SEX DEVELOPMENT (DSD) / PRIMARY ADRENAL INSUFFICIENCY PANEL HYPOGONADOTROPIC HYPOGONADISM (KALLMANN) PANEL METABOLIC DISORDERS PANEL
HS6ST2	100%	100%	98.8%	66.3%	?Paganini-Miozzo syndrome, 301025	MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS

HSCB	100%	100%	100%	99.3%	?Anemia, sideroblastic, 5, 619523	IRON DISORDERS PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS
HSD11B1	100%	100%	100%	99.4%	Cortisone reductase deficiency 2, 614662	METABOLIC DISORDERS PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS
HSD11B2	100%	99.7%	100%	98.3%	Apparent mineralocorticoid excess, 218030	METABOLIC DISORDERS PANEL RENAL DISORDERS PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹

HSD17B10	100%	99.9%	99.3%	71.3%	HSD10 mitochondrial disease, 300438	EPILEPSY PANEL METABOLIC DISORDERS PANEL INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS MITOCHONDRIAL DISORDERS PANEL
HSD17B3	100%	100%	100%	99.2%	Pseudohermaphroditism, male, with gynecomastia, 264300	DISORDERS/DIFFEREN CES OF SEX DEVELOPMENT (DSD) / PRIMARY ADRENAL INSUFFICIENCY PANEL METABOLIC DISORDERS PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹

HSD17B4	100%	100%	100%	99.3%	D-bifunctional protein deficiency, 261515;Perrault syndrome 1, 233400	MOVEMENT DISORDERS PANEL HEARING IMPAIRMENT PANEL (INCLUDING GJB2) LIVER DISORDERS PANEL METABOLIC DISORDERS PANEL INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS DISORDERS/DIFFEREN CES OF SEX DEVELOPMENT (DSD) / PRIMARY ADRENAL INSUFFICIENCY PANEL EPILEPSY PANEL POLYNEUROPATHIES PANEL ¹ MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹ PREMATURE OVARIAN INSUFFICIENCY PANEL
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HSD3B2	100%	100%	100%	98.4%	Adrenal hyperplasia, congenital, due to 3-beta-hydroxysteroid dehydrogenase 2 deficiency, 201810	DISORDERS/DIFFERENCES OF SEX DEVELOPMENT (DSD) / PRIMARY ADRENAL INSUFFICIENCY PANEL METABOLIC DISORDERS PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹
HSD3B7	100%	100%	100%	99%	Bile acid synthesis defect, congenital, 1, 607765	LIVER DISORDERS PANEL METABOLIC DISORDERS PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹
HSF2	100%	100%	100%	99.6%		MALE INFERTILITY PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS

HSF2BP	100%	100%	100%	99.4%	Premature ovarian failure 19, 619245	DISORDERS/DIFFERENCES OF SEX DEVELOPMENT (DSD) / PRIMARY ADRENAL INSUFFICIENCY PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS PREMATURE OVARIAN INSUFFICIENCY PANEL
HSF4	100%	100%	100%	98.8%	Cataract 5, multiple types, 116800	VISION DISORDERS PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS
HSFY1	50%	50%	49.7%	30.8%		MALE INFERTILITY PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS
HSFY2	50%	50%	49.7%	32.2%		MALE INFERTILITY PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS

HSPA9	100%	100%	100%	98.8%	Even-plus syndrome, 616854;Anemia, sideroblastic, 4, 182170	IRON DISORDERS PANEL SHORT STATURE/SKELETAL DYSPLASIA PANEL WITH GENOME WIDE CNV ANALYSIS INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS MITOCHONDRIAL DISORDERS PANEL COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹
HSPB1	100%	100%	100%	97.9%	Charcot-Marie-Tooth disease, axonal, type 2F, 606595;Neuronopathy, distal hereditary motor, autosomal dominant 3, 608634	POLYNEUROPATHIES PANEL ¹ NEUROLOGICAL PAIN DISORDERS PANEL ¹ MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS

HSPB3	100%	100%	100%	99.2%	?Neuronopathy, distal hereditary motor, autosomal dominant 4, 613376	POLYNEUROPATHIES PANEL ¹ MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS
HSPB6	100%	100%	99.8%	95.5%		HEART DISORDERS PANEL ¹ MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS
HSPB8	100%	100%	100%	99.3%	Neuronopathy, distal hereditary motor, autosomal dominant 2, 158590;Charcot-Marie-Tooth disease, axonal, type 2L, 608673	POLYNEUROPATHIES PANEL ¹ MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS

HSPD1	99.9%	98.2%	100%	99.6%	Spastic paraplegia 13, autosomal dominant, 605280;Leukodystrophy, hypomyelinating, 4, 612233	MOVEMENT DISORDERS PANEL HEART DISORDERS PANEL ¹ INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS MITOCHONDRIAL DISORDERS PANEL COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹
HSPG2	100%	100%	100%	99.1%	Dyssegmental dysplasia, Silverman-Handmaker type, 224410;Schwartz-Jampel syndrome, type 1, 255800	VISION DISORDERS PANEL SHORT STATURE/SKELETAL DYSPLASIA PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹ MUSCLE DISORDERS PANEL

HTR1A	100%	100%	100%	98.5%	?Periodic fever, menstrual cycle dependent, 614674	MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS
HTRA1	99.8%	98.7%	100%	98.5%	{Macular degeneration, age-related, neovascular type}, 610149;{Macular degeneration, age-related, 7}, 610149;CARASIL syndrome, 600142;Cerebral arteriopathy, autosomal dominant, with subcortical infarcts and leukoencephalopathy, type 2, 616779	SKIN DISORDERS PANEL ¹ MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹
HTRA2	100%	100%	100%	99.2%	{Parkinson disease 13}, 610297;3-methylglutaconic aciduria, type VIII, 617248	METABOLIC DISORDERS PANEL INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS MITOCHONDRIAL DISORDERS PANEL COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹

HTT	99.9%	99.9%	100%	99.3%	Lopes-Maciel-Rodan syndrome, 617435;Huntington disease, 143100	MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS
HUWE1	100%	99.6%	98.6%	67.5%	Intellectual developmental disorder, X-linked syndromic, Turner type, 309590	CRANIOFACIAL ANOMALIES PANEL WITH GENOME WIDE CNV ANALYSIS INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS
HYAL1	100%	100%	100%	97.9%	Mucopolysaccharidosis type IX, 601492	SKIN DISORDERS PANEL ¹ METABOLIC DISORDERS PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹

HYAL2	100%	100%	100%	99.3%		CRANIOFACIAL ANOMALIES PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹
HYCC1	100%	100%	100%	99.6%	Leukodystrophy, hypomyelinating, 5, 610532	MOVEMENT DISORDERS PANEL INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹ VISION DISORDERS PANEL POLYNEUROPATHIES PANEL ¹

HYDIN	99.9%	99.9%	100%	99.3%	Ciliary dyskinesia, primary, 5, 608647	CILIOPATHIES PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹
HYLS1	100%	100%	100%	99.3%	Hydrolethalus syndrome, 236680	CILIOPATHIES PANEL SHORT STATURE/SKELETAL DYSPLASIA PANEL WITH GENOME WIDE CNV ANALYSIS INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹ OROFACIAL CLEFTING PANEL WITH GENOME WIDE CNV ANALYSIS

HYOU1	100%	100%	100%	98.9%	?Immunodeficiency 59 and hypoglycemia, 233600	PRIMARY IMMUNODEFICIENCIES PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS
IARS1	100%	100%	100%	99.5%	Growth retardation, impaired intellectual development, hypotonia, and hepatopathy, 617093	LIVER DISORDERS PANEL INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL¹

IARS2	100%	100%	100%	99.4%	Cataracts, growth hormone deficiency, sensory neuropathy, sensorineural hearing loss, and skeletal dysplasia, 616007	POLYNEUROPATHIES PANEL ¹ MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS MITOCHONDRIAL DISORDERS PANEL SHORT STATURE/SKELETAL DYSPLASIA PANEL WITH GENOME WIDE CNV ANALYSIS INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹
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IBA57	100%	100%	100%	98.4%	Multiple mitochondrial dysfunctions syndrome 3, 615330;?Spastic paraplegia 74, autosomal recessive, 616451	MOVEMENT DISORDERS PANEL VISION DISORDERS PANEL INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS MITOCHONDRIAL DISORDERS PANEL COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹
ICOS	100%	100%	100%	99%	Immunodeficiency, common variable, 1, 607594	PRIMARY IMMUNODEFICIENCIES PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹

ICOSLG	100%	100%	100%	98.9%	?Immunodeficiency 119, 620825	PRIMARY IMMUNODEFICIENCIES PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS
ID4	100%	99.5%	100%	95.9%		SHORT STATURE/SKELETAL DYSPLASIA PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS
IDH1	100%	100%	100%	99.2%	{Glioma, susceptibility to, somatic}, 137800	SHORT STATURE/SKELETAL DYSPLASIA PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS HEREDITARY CANCER PANEL

IDH2	100%	100%	100%	98.6%	D-2-hydroxyglutaric aciduria 2, 613657	MITOCHONDRIAL DISORDERS PANEL HEREDITARY CANCER PANEL EPILEPSY PANEL SHORT STATURE/SKELETAL DYSPLASIA PANEL WITH GENOME WIDE CNV ANALYSIS METABOLIC DISORDERS PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS
IDH3A	100%	100%	100%	99.5%	Retinitis pigmentosa 90, 619007	VISION DISORDERS PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹

IDH3B	100%	100%	100%	99%	Retinitis pigmentosa 46, 612572	VISION DISORDERS PANEL METABOLIC DISORDERS PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL¹
IDH3G	100%	100%	98.3%	65.3%		VISION DISORDERS PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS
IDI1	100%	100%	100%	99.8%		METABOLIC DISORDERS PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS

IDS	100%	99.5%	98.8%	72.9%	Mucopolysaccharidosis II, 309900	SHORT STATURE/SKELETAL DYSPLASIA PANEL WITH GENOME WIDE CNV ANALYSIS LIVER DISORDERS PANEL METABOLIC DISORDERS PANEL INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS
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IDUA	100%	100%	100%	98.4%	Mucopolysaccharidosis Is, 607016;Mucopolysaccharidosis Ih/s, 607015;Mucopolysaccharidosis Ih, 607014	METABOLIC DISORDERS PANEL INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹ VISION DISORDERS PANEL SKIN DISORDERS PANEL ¹ HEART DISORDERS PANEL ¹ SHORT STATURE/SKELETAL DYSPLASIA PANEL WITH GENOME WIDE CNV ANALYSIS
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IER3IP1	100%	100%	100%	99.6%	Microcephaly, epilepsy, and diabetes syndrome, 614231	INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL¹ EPILEPSY PANEL
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IFIH1	100%	100%	100%	99.5%	Immunodeficiency 95, 619773;Aicardi-Goutieres syndrome 7, 615846;Singleton-Merten syndrome 1, 182250	INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹ VISION DISORDERS PANEL EPILEPSY PANEL PRIMARY IMMUNODEFICIENCIES PANEL SHORT STATURE/SKELETAL DYSPLASIA PANEL WITH GENOME WIDE CNV ANALYSIS
IFITM5	100%	99.9%	100%	98.8%	Osteogenesis imperfecta, type V, 610967	SHORT STATURE/SKELETAL DYSPLASIA PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS

IFNAR1	94.5%	94.5%	100%	99.1%	Immunodeficiency 106, susceptibility to viral infections, 619935	PRIMARY IMMUNODEFICIENCIES PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL¹
IFNAR2	100%	100%	100%	99.4%	{Hepatitis B virus, susceptibility to}, 610424;Immunodeficiency 45, 616669	PRIMARY IMMUNODEFICIENCIES PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL¹
IFNG	100%	100%	100%	99.2%	{Hepatitis C virus, response to therapy of}, 609532;{TSC2 angiomyolipomas, renal, modifier of}, 613254;{Aplastic anemia}, 609135;?Immunodeficiency 69, mycobacteriosis, 618963;{Tuberculosis, protection against}, 607948;{AIDS, rapid progression to}, 609423	PRIMARY IMMUNODEFICIENCIES PANEL RENAL DISORDERS PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS

IFNGR1	100%	100%	100%	99.6%	{H. pylori infection, susceptibility to}, 600263;Immunodeficiency 27A, mycobacteriosis, AR, 209950;Immunodeficiency 27B, mycobacteriosis, AD, 615978;{Tuberculosis infection, protection against}, 607948;{Tuberculosis, susceptibility to}, 607948;{Hepatitis B virus infection, susceptibility to}, 610424	PRIMARY IMMUNODEFICIENCIES PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹
IFNGR2	100%	100%	100%	98.9%	Immunodeficiency 28, mycobacteriosis, 614889	PRIMARY IMMUNODEFICIENCIES PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹
IFNLR1	100%	100%	100%	98.4%		HEARING IMPAIRMENT PANEL (INCLUDING GJB2) MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS

IFRD1	100%	100%	100%	99.4%		POLYNEUROPATHIES PANEL ¹ MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS
IFT122	100%	100%	100%	99.3%	Cranioectodermal dysplasia 1, 218330	CRANIOFACIAL ANOMALIES PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹ CILIOPATHIES PANEL SKIN DISORDERS PANEL ¹ SHORT STATURE/SKELETAL DYSPLASIA PANEL WITH GENOME WIDE CNV ANALYSIS RENAL DISORDERS PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS

IFT140	100%	100%	100%	99.1%	Short-rib thoracic dysplasia 9 with or without polydactyly, 266920;Retinitis pigmentosa 80, 617781	VISION DISORDERS PANEL CILIOPATHIES PANEL SHORT STATURE/SKELETAL DYSPLASIA PANEL WITH GENOME WIDE CNV ANALYSIS LIVER DISORDERS PANEL INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS RENAL DISORDERS PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹ OROFACIAL CLEFTING PANEL WITH GENOME WIDE CNV ANALYSIS
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IFT172	100%	100%	100%	98.9%	Retinitis pigmentosa 71, 616394;Bardet-Biedl syndrome 20, 619471;Short-rib thoracic dysplasia 10 with or without polydactyly, 615630	<p>OROFACIAL CLEFTING PANEL WITH GENOME WIDE CNV ANALYSIS LIVER DISORDERS PANEL INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS RENAL DISORDERS PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL¹ VISION DISORDERS PANEL CILIOPATHIES PANEL DISORDERS/DIFFEREN CES OF SEX DEVELOPMENT (DSD) / PRIMARY ADRENAL INSUFFICIENCY PANEL HYPOGONADOTROPIC HYPOGONADISM (KALLMANN) PANEL SHORT STATURE/SKELETAL DYSPLASIA PANEL WITH GENOME WIDE CNV ANALYSIS</p>
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IFT27	100%	100%	100%	99.9%	Bardet-Biedl syndrome 19, 615996	RENAL DISORDERS PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL¹ VISION DISORDERS PANEL CILIOPATHIES PANEL HYPOGONADOTROPIC HYPOGONADISM (KALLMANN) PANEL INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS
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IFT43	100%	100%	100%	99.5%	?Cranioectodermal dysplasia 3, 614099;?Retinitis pigmentosa 81, 617871;Short-rib thoracic dysplasia 18 with polydactyly, 617866	VISION DISORDERS PANEL CRANIOFACIAL ANOMALIES PANEL WITH GENOME WIDE CNV ANALYSIS CILIOPATHIES PANEL SKIN DISORDERS PANEL ¹ SHORT STATURE/SKELETAL DYSPLASIA PANEL WITH GENOME WIDE CNV ANALYSIS LIVER DISORDERS PANEL RENAL DISORDERS PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹
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IFT52	100%	100%	100%	99.3%	Short-rib thoracic dysplasia 16 with or without polydactyly, 617102	VISION DISORDERS PANEL CILIOPATHIES PANEL SHORT STATURE/SKELETAL DYSPLASIA PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹
IFT56	100%	100%	100%	99.5%	Biliary, renal, neurologic, and skeletal syndrome, 619534	CILIOPATHIES PANEL LIVER DISORDERS PANEL RENAL DISORDERS PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS

IFT57	100%	100%	100%	99.7%	?Orofaciodigital syndrome XVIII, 617927	VISION DISORDERS PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹ OROFACIAL CLEFTING PANEL WITH GENOME WIDE CNV ANALYSIS
IFT74	100%	100%	100%	99.4%	Bardet-Biedl syndrome 22, 617119;Spermatogenic failure 58, 619585;Joubert syndrome 40, 619582	VISION DISORDERS PANEL CILIOPATHIES PANEL HYPOGONADOTROPIC HYPOGONADISM (KALLMANN) PANEL MALE INFERTILITY PANEL INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹

IFT80	100%	100%	100%	99.1%	Short-rib thoracic dysplasia 2 with or without polydactyly, 611263	CILIOPATHIES PANEL SHORT STATURE/SKELETAL DYSPLASIA PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹ OROFACIAL CLEFTING PANEL WITH GENOME WIDE CNV ANALYSIS
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IFT81	94.9%	94.9%	100%	99.4%	Short-rib thoracic dysplasia 19 with or without polydactyly, 617895	VISION DISORDERS PANEL CILIOPATHIES PANEL SHORT STATURE/SKELETAL DYSPLASIA PANEL WITH GENOME WIDE CNV ANALYSIS INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹
IFT88	100%	100%	100%	99.4%		CRANIOFACIAL ANOMALIES PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS

IGBP1	100%	99.6%	99.2%	70.2%	?Corpus callosum, agenesis of, with impaired intellectual development, ocular coloboma and micrognathia, 300472	VISION DISORDERS PANEL INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS
IGF1	100%	100%	100%	98.9%	Insulin-like growth factor I deficiency, 608747	SHORT STATURE/SKELETAL DYSPLASIA PANEL WITH GENOME WIDE CNV ANALYSIS INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL¹

IGF1R	100%	100%	100%	99.4%	Insulin-like growth factor I, resistance to, 270450	TALL STATURE PANEL WITH GENOME WIDE CNV ANALYSIS SHORT STATURE/SKELETAL DYSPLASIA PANEL WITH GENOME WIDE CNV ANALYSIS INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹
IGF2	100%	100%	100%	98.6%	Silver-Russell syndrome 3, 616489	SHORT STATURE/SKELETAL DYSPLASIA PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS
IGF2R	100%	99.9%	100%	99.1%	Hepatocellular carcinoma, somatic, 114550	MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS

IGFALS	100%	100%	100%	99.1%	Acid-labile subunit, deficiency of, 615961	SHORT STATURE/SKELETAL DYSPLASIA PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹
IGFBP7	100%	100%	100%	99.1%	Retinal arterial macroaneurysm with supravalvular pulmonic stenosis, 614224	MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹
IGHG2	100%	100%	99.6%	94.6%	IgG2 deficiency, selective	MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS

IGHM	100%	100%	100%	99.1%	Agammaglobulinemia 1, 601495	PRIMARY IMMUNODEFICIENCIES PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹
IGHMBP2	100%	100%	100%	99.6%	Charcot-Marie-Tooth disease, axonal, type 2S, 616155;Neuronopathy, distal hereditary motor, autosomal recessive 1, 604320	MUSCLE DISORDERS PANEL FETAL AKINESIA PANEL POLYNEUROPATHIES PANEL ¹ MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹
IGKC	100%	100%	100%	99%	Kappa light chain deficiency, 614102	MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹

IGLL1	100%	100%	100%	97.9%	Agammaglobulinemia 2, 613500	PRIMARY IMMUNODEFICIENCIES PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹
IGSF1	100%	99.8%	99.1%	69.7%	Hypothyroidism, central, and testicular enlargement, 300888	SHORT STATURE/SKELETAL DYSPLASIA PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS
IGSF10	100%	100%	100%	99.6%		DISORDERS/DIFFEREN CES OF SEX DEVELOPMENT (DSD) / PRIMARY ADRENAL INSUFFICIENCY PANEL HYPOGONADOTROPIC HYPOGONADISM (KALLMANN) PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS

IGSF3	100%	100%	100%	99.6%	?Lacrimal duct defect, 149700	VISION DISORDERS PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS
IHH	100%	100%	100%	99.5%	Acrocapitofemoral dysplasia, 607778;Brachydactyly, type A1, 112500	SHORT STATURE/SKELETAL DYSPLASIA PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL¹

IKBKB	100%	100%	100%	98.9%	Immunodeficiency 15B, 615592;Immunodeficiency 15A, 618204	SKIN DISORDERS PANEL ¹ PRIMARY IMMUNODEFICIENCIES PANEL SHORT STATURE/SKELETAL DYSPLASIA PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹
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IKBKG	96.4%	94.2%	98.8%	72.8%	Incontinentia pigmenti, 308300;Ectodermal dysplasia and immunodeficiency 1, 300291;Immunodeficiency 33, 300636;Autoinflammatory disease, systemic, X-linked, 301081	VISION DISORDERS PANEL CRANIOFACIAL ANOMALIES PANEL WITH GENOME WIDE CNV ANALYSIS SKIN DISORDERS PANEL ¹ PRIMARY IMMUNODEFICIENCIES PANEL SHORT STATURE/SKELETAL DYSPLASIA PANEL WITH GENOME WIDE CNV ANALYSIS INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS
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IKZF1	100%	100%	100%	98.7%	Immunodeficiency, common variable, 13, 616873	INHERITED BONE MARROW FAILURE AND/OR PREDISPOSITION PANEL TO HEMATOLOGICAL MALIGNANCIES PRIMARY IMMUNODEFICIENCIES PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS HEREDITARY CANCER PANEL
IKZF2	100%	100%	100%	99.5%		INHERITED BONE MARROW FAILURE AND/OR PREDISPOSITION PANEL TO HEMATOLOGICAL MALIGNANCIES HEARING IMPAIRMENT PANEL (INCLUDING GJB2) PRIMARY IMMUNODEFICIENCIES PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS

IKZF3	100%	100%	100%	99.3%	?Immunodeficiency 84, 619437	PRIMARY IMMUNODEFICIENCIES PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS
IKZF5	100%	100%	100%	99.1%	Thrombocytopenia, autosomal dominant, 7, 619130	INHERITED BONE MARROW FAILURE AND/OR PREDISPOSITION PANEL TO HEMATOLOGICAL MALIGNANCIES HEMOSTATIC/THROMB OTIC DISORDERS PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS
IL10	100%	100%	100%	99.3%	{Rheumatoid arthritis, progression of}, 180300;{Graft-versus-host disease, protection against}, 614395;{HIV-1, susceptibility to}, 609423	PRIMARY IMMUNODEFICIENCIES PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS

IL10RA	100%	100%	100%	99.2%	Inflammatory bowel disease 28, early onset, autosomal recessive, 613148	PRIMARY IMMUNODEFICIENCIES PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹
IL10RB	100%	100%	100%	99.2%	{Hepatitis B virus, susceptibility to}, 610424;Inflammatory bowel disease 25, early onset, autosomal recessive, 612567	PRIMARY IMMUNODEFICIENCIES PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹
IL11RA	100%	100%	100%	98.3%	Craniosynostosis and dental anomalies, 614188	CRANIOFACIAL ANOMALIES PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹

IL12B	100%	100%	100%	99.8%	Immunodeficiency 29, mycobacteriosis, 614890	PRIMARY IMMUNODEFICIENCIES PANEL COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL¹ MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS
IL12RB1	94.1%	94.1%	100%	98.3%	Immunodeficiency 30, 614891	PRIMARY IMMUNODEFICIENCIES PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL¹
IL17F	100%	100%	100%	99.6%	?Candidiasis, familial, 6, autosomal dominant, 613956	PRIMARY IMMUNODEFICIENCIES PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS

IL17RA	100%	100%	100%	98.9%	Immunodeficiency 51, 613953	SKIN DISORDERS PANEL ¹ PRIMARY IMMUNODEFICIENCIES PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹
IL17RC	100%	100%	100%	99.2%	Candidiasis, familial, 9, 616445	PRIMARY IMMUNODEFICIENCIES PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹

IL17RD	100%	100%	100%	98.6%	Hypogonadotropic hypogonadism 18 with or without anosmia, 615267	SKIN DISORDERS PANEL ¹ DISORDERS/DIFFERENCES OF SEX DEVELOPMENT (DSD) / PRIMARY ADRENAL INSUFFICIENCY PANEL HYPOGONADOTROPIC HYPOGONADISM (KALLMANN) PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS
IL18BP	100%	100%	100%	98.8%	{?Hepatitis, fulminant viral, susceptibility to}, 618549	PRIMARY IMMUNODEFICIENCIES PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS
IL1R1	97.7%	97.7%	100%	99.2%	?Chronic recurrent multifocal osteomyelitis 3, 259680	PRIMARY IMMUNODEFICIENCIES PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS

IL1RAPL1	100%	100%	98.5%	66.1%	Intellectual developmental disorder, X-linked 21, 300143	INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS
IL1RN	100%	100%	100%	99.2%	Chronic recurrent multifocal osteomyelitis 2, with periostitis and pustulosis, 612852;{Gastric cancer risk after H. pylori infection}, 613659;{Microvascular complications of diabetes 4}, 612628;Interleukin 1 receptor antagonist deficiency, 612852	SKIN DISORDERS PANEL ¹ PRIMARY IMMUNODEFICIENCIES PANEL SHORT STATURE/SKELETAL DYSPLASIA PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹
IL2	100%	100%	100%	99.7%		PRIMARY IMMUNODEFICIENCIES PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS

IL21	100%	100%	100%	99.6%	?Immunodeficiency, common variable, 11, 615767	PRIMARY IMMUNODEFICIENCIES PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS
IL21R	100%	100%	100%	99.4%	Immunodeficiency 56, 615207	PRIMARY IMMUNODEFICIENCIES PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL¹
IL27RA	100%	100%	100%	97.7%		PRIMARY IMMUNODEFICIENCIES PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS

IL2RA	100%	100%	100%	98.6%	Immunodeficiency 41 with lymphoproliferation and autoimmunity, 606367;{Diabetes, mellitus, insulin-dependent, susceptibility to, 10}, 601942	PRIMARY IMMUNODEFICIENCIES PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL¹
IL2RB	96.1%	96.1%	100%	98.2%	Immunodeficiency 63 with lymphoproliferation and autoimmunity, 618495	PRIMARY IMMUNODEFICIENCIES PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL¹

IL2RG	100%	99.9%	98.4%	66%	Combined immunodeficiency, X-linked, moderate, 312863;Severe combined immunodeficiency, X-linked, 300400	PRIMARY IMMUNODEFICIENCIES PANEL SHORT STATURE/SKELETAL DYSPLASIA PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS SEVERE COMBINED IMMUNODEFICIENCY (SCID) PANEL
IL31RA	100%	100%	100%	99.4%	?Amyloidosis, primary localized cutaneous, 2, 613955	SKIN DISORDERS PANEL ¹ MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS

IL36RN	100%	100%	100%	99.7%	Psoriasis 14, pustular, 614204	SKIN DISORDERS PANEL ¹ PRIMARY IMMUNODEFICIENCIES PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹
IL37	100%	100%	100%	99.7%	?Inflammatory bowel disease (infantile ulcerative colitis) 31, 619398	MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS
IL6R	92.5%	92.5%	100%	98.9%	[Interleukin 6, serum level of, QTL], 614752;Hyper-IgE syndrome 5, autosomal recessive, with recurrent infections, 618944;[Interleukin-6 receptor, soluble, serum level of, QTL], 614689	PRIMARY IMMUNODEFICIENCIES PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹

IL6ST	100%	100%	100%	99.5%	Hyper-IgE syndrome 4A, autosomal dominant, with recurrent infections, 619752;Stuve-Wiedemann syndrome 2, 619751;Hyper-IgE syndrome 4B, autosomal recessive, with recurrent infections, 618523;?Immunodeficiency 94 with autoinflammation and dysmorphic facies, 619750	CRANIOFACIAL ANOMALIES PANEL WITH GENOME WIDE CNV ANALYSIS PRIMARY IMMUNODEFICIENCIES PANEL SHORT STATURE/SKELETAL DYSPLASIA PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹
IL7	100%	100%	100%	98.6%	{?Epidermodysplasia verruciformis, susceptibility to, 5}, 618309	PRIMARY IMMUNODEFICIENCIES PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS

IL7R	100%	100%	100%	99.6%	Immunodeficiency 104, severe combined, 608971	PRIMARY IMMUNODEFICIENCIES PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹ SEVERE COMBINED IMMUNODEFICIENCY (SCID) PANEL
ILDR1	100%	100%	100%	99.4%	Deafness, autosomal recessive 42, 609646	HEARING IMPAIRMENT PANEL (INCLUDING GJB2) MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹
ILK	100%	100%	100%	99.4%		HEART DISORDERS PANEL ¹ MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS

IMPA1	100%	100%	100%	99%	Intellectual developmental disorder, autosomal recessive 59, 617323	INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹
IMPDH1	100%	100%	100%	99%	Retinitis pigmentosa 10, 180105;Leber congenital amaurosis 11, 613837	VISION DISORDERS PANEL METABOLIC DISORDERS PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS
IMPG1	100%	100%	100%	99%	Macular dystrophy, vitelliform, 4, 616151;Retinitis pigmentosa 91, 153870	VISION DISORDERS PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS

IMPG2	100%	100%	100%	99.3%	Retinitis pigmentosa 56, 613581;Macular dystrophy, vitelliform, 5, 616152	VISION DISORDERS PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹
INF2	100%	99.9%	99.4%	94.7%	Glomerulosclerosis, focal segmental, 5, 613237;Charcot-Marie-Tooth disease, dominant intermediate E, 614455	POLYNEUROPATHIES PANEL ¹ RENAL DISORDERS PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS
ING1	100%	100%	100%	98.3%	Squamous cell carcinoma, head and neck, somatic, 275355	MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS
INO80	100%	100%	100%	99.3%		PRIMARY IMMUNODEFICIENCIES PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS

INPP4A	100%	100%	100%	99.2%		INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS
INPP5E	100%	100%	100%	96.9%	Impaired intellectual development, truncal obesity, retinal dystrophy, and micropenis syndrome, 610156;Joubert syndrome 1, 213300	VISION DISORDERS PANEL CILIOPATHIES PANEL INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS RENAL DISORDERS PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL' METABOLIC DISORDERS PANEL

INPP5K	100%	100%	100%	98.9%	Muscular dystrophy, congenital, with cataracts and intellectual disability, 617404	INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹ MUSCLE DISORDERS PANEL
INPPL1	100%	100%	100%	98.7%	Opsismodysplasia, 258480	SHORT STATURE/SKELETAL DYSPLASIA PANEL WITH GENOME WIDE CNV ANALYSIS METABOLIC DISORDERS PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹
INS	100%	100%	100%	99.8%	Diabetes mellitus, insulin-dependent, 2, 125852;Maturity-onset diabetes of the young, type 10, 613370;Hyperproinsulinemia, 616214;Diabetes mellitus, permanent neonatal 4, 618858	MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS

INSC	94.4%	94.4%	100%	99%		POLYNEUROPATHIES PANEL ¹ MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS
INSL3	78.8%	78.8%	100%	97.9%	Cryptorchidism, 219050	DISORDERS/DIFFEREN CES OF SEX DEVELOPMENT (DSD) / PRIMARY ADRENAL INSUFFICIENCY PANEL MALE INFERTILITY PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS

INSR	100%	100%	100%	98.5%	Rabson-Mendenhall syndrome, 262190;Diabetes mellitus, insulin-resistant, with acanthosis nigricans, 610549;Donohue syndrome, 246200;Hyperinsulinemic hypoglycemia, familial, 5, 609968	SKIN DISORDERS PANEL ¹ PRIMARY IMMUNODEFICIENCIES PANEL LIVER DISORDERS PANEL METABOLIC DISORDERS PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹
INTS1	100%	100%	100%	98.9%	Neurodevelopmental disorder with cataracts, poor growth, and dysmorphic facies, 618571	INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹

INTS11	100%	100%	100%	97.4%	Neurodevelopmental disorder with motor and language delay, ocular defects, and brain abnormalities, 620428	INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹
INTS13	100%	100%	100%	99.6%		INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS
INTS8	100%	100%	100%	99%	?Neurodevelopmental disorder with cerebellar hypoplasia and spasticity, 618572	MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹

INTU	100%	100%	100%	99.3%	?Orofaciodigital syndrome XVII, 617926;?Short-rib thoracic dysplasia 20 with polydactyly, 617925	CRANIOFACIAL ANOMALIES PANEL WITH GENOME WIDE CNV ANALYSIS CILIOPATHIES PANEL SHORT STATURE/SKELETAL DYSPLASIA PANEL WITH GENOME WIDE CNV ANALYSIS RENAL DISORDERS PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL¹ OROFACIAL CLEFTING PANEL WITH GENOME WIDE CNV ANALYSIS
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INVS	100%	100%	100%	99.5%	Nephronophthisis 2, infantile, 602088	VISION DISORDERS PANEL CILIOPATHIES PANEL LIVER DISORDERS PANEL RENAL DISORDERS PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL¹
IPMK	100%	100%	100%	99.4%		MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS HEREDITARY CANCER PANEL

IPO8	100%	100%	100%	99.6%	VISS syndrome, 619472	THORACIC AORTIC ANEURYSM AND/OR DISSECTION PANEL PRIMARY IMMUNODEFICIENCIES PANEL INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹
IQCB1	100%	100%	100%	99.5%	Senior-Loken syndrome 5, 609254	VISION DISORDERS PANEL CILIOPATHIES PANEL RENAL DISORDERS PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹

IQCE	100%	100%	100%	97.9%	Polydactyly, postaxial, type A7, 617642	MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹
IQCN	100%	100%	100%	99.2%	Spermatogenic failure 78, 620170	MALE INFERTILITY PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS
IQGAP1	100%	100%	100%	98.8%		MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS
IQSEC1	100%	99.2%	100%	97.3%	Intellectual developmental disorder with short stature and behavioral abnormalities, 618687	INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹

IQSEC2	99.3%	96.4%	96%	61.7%	Intellectual developmental disorder, X-linked 1, 309530	EPILEPSY PANEL INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS
IQUB	100%	100%	100%	99.6%		MALE INFERTILITY PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS
IRAK1	100%	99.5%	97.2%	65.4%		PRIMARY IMMUNODEFICIENCIES PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS

IRAK4	100%	100%	100%	99.8%	Immunodeficiency 67, 607676	PRIMARY IMMUNODEFICIENCIES PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹
IREB2	100%	100%	100%	99.6%	Neurodegeneration, early-onset, with choreoathetoid movements and microcytic anemia, 618451	METABOLIC DISORDERS PANEL INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹
IRF1	100%	100%	100%	99.7%	Nonsmall cell lung cancer, somatic, 211980;Gastric cancer, somatic, 613659;Immunodeficiency 117, mycobacteriosis, autosomal recessive, 620668	PRIMARY IMMUNODEFICIENCIES PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS

IRF2BP2	100%	100%	100%	97.4%	?Immunodeficiency, common variable, 14, 617765	PRIMARY IMMUNODEFICIENCIES PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS
IRF2BPL	100%	100%	99.8%	91.8%	Neurodevelopmental disorder with regression, abnormal movements, loss of speech, and seizures, 618088	MOVEMENT DISORDERS PANEL EPILEPSY PANEL INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS
IRF3	100%	100%	100%	98.8%	{Encephalopathy, acute, infection-induced (herpes-specific), susceptibility to, 7}, 616532	PRIMARY IMMUNODEFICIENCIES PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS

IRF4	100%	100%	100%	98.8%	[Skin/hair/eye pigmentation, variation in, 8], 611724	SKIN DISORDERS PANEL ¹ PRIMARY IMMUNODEFICIENCIES PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹
IRF6	100%	100%	100%	99%	{Orofacial cleft 6}, 608864;Popliteal pterygium syndrome 1, 119500;van der Woude syndrome 1, 119300	CRANIOFACIAL ANOMALIES PANEL WITH GENOME WIDE CNV ANALYSIS SKIN DISORDERS PANEL ¹ DISORDERS/DIFFEREN CES OF SEX DEVELOPMENT (DSD) / PRIMARY ADRENAL INSUFFICIENCY PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS OROFACIAL CLEFTING PANEL WITH GENOME WIDE CNV ANALYSIS

IRF7	100%	100%	100%	98.8%	?Immunodeficiency 39, 616345	PRIMARY IMMUNODEFICIENCIES PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹
IRF8	100%	100%	100%	98.6%	Immunodeficiency 32A, mycobacteriosis, autosomal dominant, 614893;Immunodeficiency 32B, monocyte and dendritic cell deficiency, autosomal recessive, 226990	PRIMARY IMMUNODEFICIENCIES PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹
IRF9	100%	100%	100%	99.1%	Immunodeficiency 65, susceptibility to viral infections, 618648	PRIMARY IMMUNODEFICIENCIES PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹

IRGM	100%	100%	100%	100%	{Mycobacterium tuberculosis, protection against}, 607948;{Inflammatory bowel disease (Crohn disease) 19}, 612278	PRIMARY IMMUNODEFICIENCIES PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS
IRS4	100%	99.7%	97.8%	63.2%	Hypothyroidism, congenital, nongoitrous, 9, 301035	MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS
IRX1	99.8%	96%	100%	97.9%		VISION DISORDERS PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS
IRX5	100%	100%	100%	96.4%	Hamamy syndrome, 611174	INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹

ISCA1	92.4%	92.4%	100%	98.8%	Multiple mitochondrial dysfunctions syndrome 5, 617613	MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS MITOCHONDRIAL DISORDERS PANEL COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹
ISCA2	100%	100%	100%	99.7%	Multiple mitochondrial dysfunctions syndrome 4, 616370	MOVEMENT DISORDERS PANEL INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS MITOCHONDRIAL DISORDERS PANEL COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹

ISCU	100%	100%	100%	99.7%	Myopathy with lactic acidosis, hereditary, 255125	MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS MITOCHONDRIAL DISORDERS PANEL COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹ MUSCLE DISORDERS PANEL
ISG15	100%	100%	100%	99%	Immunodeficiency 38, 616126	SKIN DISORDERS PANEL ¹ PRIMARY IMMUNODEFICIENCIES PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹
ISL1	100%	100%	100%	99.3%		CONGENITAL HEARTDISEASE PANEL ¹ HEART DISORDERS PANEL ¹ MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS

ITCH	92.5%	92.5%	100%	99.2%	Autoimmune disease, multisystem, with facial dysmorphism, 613385	PRIMARY IMMUNODEFICIENCIES PANEL METABOLIC DISORDERS PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹
ITGA2	100%	100%	100%	99.6%		HEMOSTATIC/THROMB OTIC DISORDERS PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS
ITGA2B	100%	100%	100%	98.8%	Thrombocytopenia, neonatal alloimmune, BAK antigen related;Glanzmann thrombasthenia 1, 273800;Bleeding disorder, platelet-type, 16, autosomal dominant, 187800	HEMOSTATIC/THROMB OTIC DISORDERS PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹

ITGA3	100%	100%	100%	99.1%	Epidermolysis bullosa, junctional 7, with interstitial lung disease and nephrotic syndrome, 614748	COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹ SKIN DISORDERS PANEL ¹ RENAL DISORDERS PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS
ITGA6	100%	100%	100%	99.5%	Epidermolysis bullosa, junctional 6, with pyloric atresia, 619817	SKIN DISORDERS PANEL ¹ MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹

ITGA7	100%	100%	100%	99.2%	Muscular dystrophy, congenital, due to ITGA7 deficiency, 613204	HEART DISORDERS PANEL ¹ INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹ MUSCLE DISORDERS PANEL
ITGA8	100%	100%	100%	99.1%	Renal hypodysplasia/aplasia 1, 191830	RENAL DISORDERS PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹

ITGB2	100%	99.9%	100%	99.3%	Leukocyte adhesion deficiency, 116920	PRIMARY IMMUNODEFICIENCIES PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹
ITGB3	100%	100%	100%	99.4%	Bleeding disorder, platelet-type, 24, autosomal dominant, 619271;Thrombocytopenia, neonatal alloimmune;Purpura, posttransfusion;{Myocardial infarction, susceptibility to}, 608446;Glanzmann thrombasthenia 2, 619267	HEMOSTATIC/THROMB OTIC DISORDERS PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹
ITGB4	100%	100%	100%	98.7%	Epidermolysis bullosa, junctional 5B, with pyloric atresia, 226730;Epidermolysis bullosa, junctional 5A, intermediate, 619816	SKIN DISORDERS PANEL ¹ MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹

ITGB6	100%	100%	100%	99.4%	Amelogenesis imperfecta, type IH, 616221	CRANIOFACIAL ANOMALIES PANEL WITH GENOME WIDE CNV ANALYSIS SKIN DISORDERS PANEL ¹ MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹
ITK	100%	100%	100%	99.6%	Lymphoproliferative syndrome 1, 613011	PRIMARY IMMUNODEFICIENCIES PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹ HEREDITARY CANCER PANEL
ITM2B	100%	100%	100%	99.4%	?Retinal dystrophy with inner retinal dysfunction and ganglion cell abnormalities, 616079;Dementia, familial British, 176500;Dementia, familial Danish, 117300	VISION DISORDERS PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS

ITPA	100%	100%	100%	99%	[Inosine triphosphatase deficiency], 613850;Developmental and epileptic encephalopathy 35, 616647	EPILEPSY PANEL HEART DISORDERS PANEL ¹ METABOLIC DISORDERS PANEL INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹
ITPKB	100%	100%	100%	98.4%		PRIMARY IMMUNODEFICIENCIES PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS SEVERE COMBINED IMMUNODEFICIENCY (SCID) PANEL

ITPR1	100%	100%	100%	99.3%	Gillespie syndrome, 206700;Spinocerebellar ataxia 29, congenital nonprogressive, 117360;Spinocerebellar ataxia 15, 606658	MOVEMENT DISORDERS PANEL VISION DISORDERS PANEL INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹
ITPR2	100%	100%	100%	99.6%	?Anhidrosis, isolated, with normal sweat glands, 106190	MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS
ITPR3	100%	100%	100%	98.9%	Charcot-Marie-Tooth disease, demyelinating, type 1J, 620111;{Diabetes, type 1, susceptibility to}, 222100	POLYNEUROPATHIES PANEL ¹ PRIMARY IMMUNODEFICIENCIES PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS

ITSN1	100%	100%	100%	99.3%		INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS RENAL DISORDERS PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS
ITSN2	100%	100%	100%	99.3%		RENAL DISORDERS PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS

IVD	100%	100%	100%	99.5%	Isovaleric acidemia, 243500	INHERITED BONE MARROW FAILURE AND/OR PREDISPOSITION PANEL TO HEMATOLOGICAL MALIGNANCIES METABOLIC DISORDERS PANEL INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL¹
IVNS1ABP	100%	100%	100%	99.4%	Immunodeficiency 70, 618969	PRIMARY IMMUNODEFICIENCIES PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS

IYD	100%	100%	100%	99.3%	Thyroid dyshormonogenesis 4, 274800	MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹
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JAG1	100%	100%	100%	99.2%	?Deafness, congenital heart defects, and posterior embryotoxon, 617992;Charcot-Marie-Tooth disease, axonal, type 2HH, 619574;Alagille syndrome 1, 118450;Tetralogy of Fallot, 187500	THORACIC AORTIC ANEURYSM AND/OR DISSECTION PANEL VISION DISORDERS PANEL SHORT STATURE/SKELETAL DYSPLASIA PANEL WITH GENOME WIDE CNV ANALYSIS LIVER DISORDERS PANEL INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS RENAL DISORDERS PANEL CONGENITAL HEARTDISEASE PANEL ¹ HEART DISORDERS PANEL ¹ POLYNEUROPATHIES PANEL ¹ MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS
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JAG2	99.8%	99.1%	100%	98.7%	Muscular dystrophy, limb-girdle, autosomal recessive 27, 619566	INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹ MUSCLE DISORDERS PANEL
JAGN1	100%	100%	100%	98.6%	Neutropenia, severe congenital, 6, autosomal recessive, 616022	INHERITED BONE MARROW FAILURE AND/OR PREDISPOSITION PANEL TO HEMATOLOGICAL MALIGNANCIES PRIMARY IMMUNODEFICIENCIES PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹

JAK1	100%	100%	100%	99.2%	Autoinflammation, immune dysregulation, and eosinophilia, 618999	PRIMARY IMMUNODEFICIENCIES PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS
JAK2	100%	100%	100%	99.2%	{Budd-Chiari syndrome, somatic}, 600880;Myelofibrosis, somatic, 254450;Erythrocytosis, somatic, 133100;Leukemia, acute myeloid, somatic, 601626;Thrombocythemia 3, 614521;Polycythemia vera, somatic, 263300	INHERITED BONE MARROW FAILURE AND/OR PREDISPOSITION PANEL TO HEMATOLOGICAL MALIGNANCIES PRIMARY IMMUNODEFICIENCIES PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS IRON DISORDERS PANEL HEMOSTATIC/THROMB OTIC DISORDERS PANEL

JAK3	100%	100%	100%	97.5%	Severe combined immunodeficiency, autosomal recessive, T-negative/B-positive type, 600802	PRIMARY IMMUNODEFICIENCIES PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL¹ SEVERE COMBINED IMMUNODEFICIENCY (SCID) PANEL
JAM2	92.8%	92%	100%	99.4%	Basal ganglia calcification, idiopathic, 8, autosomal recessive, 618824	MOVEMENT DISORDERS PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL¹

JAM3	100%	100%	100%	99.6%	Hemorrhagic destruction of the brain, subependymal calcification, and cataracts, 613730	MOVEMENT DISORDERS PANEL VISION DISORDERS PANEL EPILEPSY PANEL INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL¹
JARID2	100%	100%	100%	98.8%	Developmental delay with variable intellectual disability and dysmorphic facies, 620098	INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS
JMJD1C	100%	100%	100%	99.5%		INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS

JPH1	100%	100%	100%	99.1%	Congenital myopathy 25, 620964;{?Charcot-Marie-Tooth disease, axonal, autosomal dominant, type 2K, modifier of}, 607831	MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS
JPH2	100%	99.9%	100%	98.4%	Cardiomyopathy, dilated, 2E, 619492;Cardiomyopathy, hypertrophic, 17, 613873	DILATED CARDIOMYOPATHY PANEL ¹ HEART DISORDERS PANEL ¹ HYPERTROPHIC CARDIOMYOPATHY PANEL ¹ MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹
JPH3	100%	100%	100%	97%	Huntington disease-like 2, 606438	MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS

JUP	100%	99.9%	100%	98.8%	Naxos disease, 601214;?Arrhythmogenic right ventricular dysplasia 12, 611528	ARITMOGENE CARDIOMYOPATHY PANEL ¹ SKIN DISORDERS PANEL ¹ HEART DISORDERS PANEL ¹ MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹ ARRHYTHMIA AND CARDIAC CONDUCTION DISORDERS PANEL ¹
KALRN	98.3%	98.3%	100%	99.1%		MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹

KANK1	98.1%	98.1%	100%	99.6%	Cerebral palsy, spastic quadriplegic, 2, 612900	INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS RENAL DISORDERS PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS
KANK2	100%	100%	100%	98.9%	Nephrotic syndrome, type 16, 617783;Palmoplantar keratoderma and woolly hair, 616099	SKIN DISORDERS PANEL ¹ RENAL DISORDERS PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹

KANSL1	100%	100%	100%	99.4%	Koolen-De Vries syndrome, 610443	THORACIC AORTIC ANEURYSM AND/OR DISSECTION PANEL EPILEPSY PANEL HEART DISORDERS PANEL ¹ INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS OROFACIAL CLEFTING PANEL WITH GENOME WIDE CNV ANALYSIS
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KARS1	100%	100%	100%	99.5%	Deafness, autosomal recessive 89, 613916;Leukoencephalopathy, progressive, infantile-onset, with or without deafness, 619147;?Charcot-Marie-Tooth disease, recessive intermediate, B, 613641;Deafness, congenital, and adult-onset progressive leukoencephalopathy, 619196	HEARING IMPAIRMENT PANEL (INCLUDING GJB2) POLYNEUROPATHIES PANEL ¹ PRIMARY IMMUNODEFICIENCIES PANEL INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS MITOCHONDRIAL DISORDERS PANEL COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹
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KASH5	100%	100%	100%	98.8%	Spermatogenic failure 88, 620547;Premature ovarian failure 22, 620548	DISORDERS/DIFFERENCES OF SEX DEVELOPMENT (DSD) / PRIMARY ADRENAL INSUFFICIENCY PANEL MALE INFERTILITY PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS PREMATURE OVARIAN INSUFFICIENCY PANEL
KAT5	100%	100%	100%	98.9%	Neurodevelopmental disorder with dysmorphic facies, sleep disturbance, and brain abnormalities, 619103	INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS
KAT6A	100%	100%	100%	98.9%	Arboleda-Tham syndrome, 616268	INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS OROFACIAL CLEFTING PANEL WITH GENOME WIDE CNV ANALYSIS

KAT6B	100%	100%	100%	99.2%	SBYSS syndrome, 603736;Genitopatellar syndrome, 606170	CRANIOFACIAL ANOMALIES PANEL WITH GENOME WIDE CNV ANALYSIS CONGENITAL HEARTDISEASE PANEL¹ SKIN DISORDERS PANEL¹ DISORDERS/DIFFEREN CES OF SEX DEVELOPMENT (DSD) / PRIMARY ADRENAL INSUFFICIENCY PANEL HEART DISORDERS PANEL¹ SHORT STATURE/SKELETAL DYSPLASIA PANEL WITH GENOME WIDE CNV ANALYSIS INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS
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KAT8	100%	100%	100%	98.5%	Li-Ghorgani-Weisz-Hubshman syndrome, 618974	INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS
KATNB1	100%	100%	100%	99.1%	Lissencephaly 6, with microcephaly, 616212	MOVEMENT DISORDERS PANEL EPILEPSY PANEL INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL¹

KATNIP	100%	100%	100%	99.3%	Joubert syndrome 26, 616784	CILIOPATHIES PANEL INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS RENAL DISORDERS PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹
KBTBD13	100%	100%	100%	99%	Nemaline myopathy 6, autosomal dominant, 609273	HEART DISORDERS PANEL ¹ POLYNEUROPATHIES PANEL ¹ MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS MUSCLE DISORDERS PANEL
KBTBD2	100%	100%	100%	99.7%		INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS

KCNA1	100%	100%	100%	98.5%	Episodic ataxia/myokymia syndrome, 160120	MOVEMENT DISORDERS PANEL EPILEPSY PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS
KCNA2	100%	100%	100%	99.2%	Developmental and epileptic encephalopathy 32, 616366	MOVEMENT DISORDERS PANEL EPILEPSY PANEL METABOLIC DISORDERS PANEL INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS
KCNA3	100%	100%	100%	98.7%		EPILEPSY PANEL INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS

KCNA4	100%	100%	100%	99%	Microcephaly, cataracts, impaired intellectual development, and dystonia with abnormal striatum, 618284	INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS
KCNA5	100%	100%	100%	98.6%	Atrial fibrillation, familial, 7, 612240	HEART DISORDERS PANEL ¹ MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS
KCNB1	100%	100%	100%	98.8%	Developmental and epileptic encephalopathy 26, 616056	EPILEPSY PANEL INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS

KCNB2	100%	100%	100%	99.3%		INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS
KCNC1	100%	100%	100%	98.5%	Epilepsy, progressive myoclonic 7, 616187	MOVEMENT DISORDERS PANEL EPILEPSY PANEL INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS
KCNC2	100%	100%	100%	99%	Developmental and epileptic encephalopathy 103, 619913	INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS

KCNC3	98.8%	95.8%	98.9%	87.2%	Spinocerebellar ataxia 13, 605259	MOVEMENT DISORDERS PANEL INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS
KCND1	100%	99.8%	98.2%	61.9%		EPILEPSY PANEL INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS
KCND2	100%	100%	100%	98.7%		HEART DISORDERS PANEL ¹ INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS

KCND3	100%	100%	100%	98.2%	Spinocerebellar ataxia 19, 607346;Brugada syndrome 9, 616399	MOVEMENT DISORDERS PANEL EPILEPSY PANEL HEART DISORDERS PANEL ¹ MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS
KCNE1	100%	100%	100%	99.5%	Jervell and Lange-Nielsen syndrome 2, 612347;Long QT syndrome 5, 613695	HEARING IMPAIRMENT PANEL (INCLUDING GJB2) HEART DISORDERS PANEL ¹ LONG QT SYNDROME PANEL ¹ MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹ ARRHYTHMIA AND CARDIAC CONDUCTION DISORDERS PANEL ¹

KCNE2	100%	100%	100%	99.4%	Long QT syndrome 6, 613693;Atrial fibrillation, familial, 4, 611493	HEART DISORDERS PANEL ¹ LONG QT SYNDROME PANEL ¹ MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS ARRHYTHMIA AND CARDIAC CONDUCTION DISORDERS PANEL ¹
KCNE3	100%	100%	100%	98%	?Brugada syndrome 6, 613119	HEART DISORDERS PANEL ¹ MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS
KCNE4	100%	100%	100%	98.5%		HEART DISORDERS PANEL ¹ MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS
KCNE5	100%	99.3%	97.1%	63.4%		HEART DISORDERS PANEL ¹ MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS

KCNH1	98.6%	98.6%	100%	99.6%	Zimmermann-Laband syndrome 1, 135500; Temple-Baraitser syndrome, 611816	SKIN DISORDERS PANEL ¹ EPILEPSY PANEL INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS
KCNH2	100%	100%	100%	97.8%	Short QT syndrome 1, 609620; Long QT syndrome 2, 613688	HEART DISORDERS PANEL ¹ LONG QT SYNDROME PANEL ¹ MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS ARRHYTHMIA AND CARDIAC CONDUCTION DISORDERS PANEL ¹
KCNH5	100%	100%	100%	99.7%	Developmental and epileptic encephalopathy 112, 620537	INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS

KCNJ1	100%	100%	100%	99.8%	Bartter syndrome, type 2, 241200	RENAL DISORDERS PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹
KCNJ10	100%	100%	100%	99.7%	Enlarged vestibular aqueduct, digenic, 600791;SESAME syndrome, 612780	MOVEMENT DISORDERS PANEL HEARING IMPAIRMENT PANEL (INCLUDING GJB2) EPILEPSY PANEL INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS RENAL DISORDERS PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹

KCNJ11	100%	100%	100%	98.9%	Diabetes, permanent neonatal 2, with or without neurologic features, 618856;{Diabetes mellitus, type 2, susceptibility to}, 125853;Maturity-onset diabetes of the young, type 13, 616329;Diabetes mellitus, transient neonatal 3, 610582;Hyperinsulinemic hypoglycemia, familial, 2, 601820	EPILEPSY PANEL HEART DISORDERS PANEL ¹ METABOLIC DISORDERS PANEL INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹
KCNJ13	100%	100%	100%	99.2%	Snowflake vitreoretinal degeneration, 193230;Leber congenital amaurosis 16, 614186	VISION DISORDERS PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹

KCNJ16	100%	100%	100%	99.9%	Hypokalemic tubulopathy and deafness, 619406	RENAL DISORDERS PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹
KCNJ2	100%	100%	100%	99.3%	Atrial fibrillation, familial, 9, 613980;Andersen syndrome, 170390;Short QT syndrome 3, 609622	HEART DISORDERS PANEL ¹ ARRHYTHMIA AND CARDIAC CONDUCTION DISORDERS PANEL ¹ OROFACIAL CLEFTING PANEL WITH GENOME WIDE CNV ANALYSIS LONG QT SYNDROME PANEL ¹ MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS SHORT STATURE/SKELETAL DYSPLASIA PANEL WITH GENOME WIDE CNV ANALYSIS MUSCLE DISORDERS PANEL

KCNJ5	100%	100%	100%	98%	Long QT syndrome 13, 613485;Hyperaldosteronism, familial, type III, 613677	HEART DISORDERS PANEL ¹ RENAL DISORDERS PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS
KCNJ6	100%	100%	100%	99.6%	Keppen-Lubinsky syndrome, 614098	MOVEMENT DISORDERS PANEL INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS
KCNJ8	100%	100%	100%	99.6%		HEART DISORDERS PANEL ¹ MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS

KCNK3	100%	99.7%	100%	98.1%	Pulmonary hypertension, primary, 4, 615344	HEART DISORDERS PANEL ¹ INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS
KCNK4	99.4%	99.4%	100%	98.2%	Facial dysmorphism, hypertrichosis, epilepsy, intellectual/developmental delay, and gingival overgrowth syndrome, 618381	INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS
KCNK9	100%	100%	100%	98.5%	Birk-Barel syndrome, 612292	SKIN DISORDERS PANEL ¹ INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS OROFACIAL CLEFTING PANEL WITH GENOME WIDE CNV ANALYSIS

KCNMA1	100%	100%	100%	98.9%	{Epilepsy, idiopathic generalized, susceptibility to, 16}, 618596;Paroxysmal nonkinesigenic dyskinesia, 3, with or without generalized epilepsy, 609446;Cerebellar atrophy, developmental delay, and seizures, 617643;Liang-Wang syndrome, 618729	MOVEMENT DISORDERS PANEL EPILEPSY PANEL LIVER DISORDERS PANEL INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹
KCNN2	100%	100%	100%	99%	?Dystonia 34, myoclonic, 619724;Neurodevelopmental disorder with or without variable movement or behavioral abnormalities, 619725	INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS

KCNN3	100%	100%	100%	98.1%	Zimmermann-Laband syndrome 3, 618658	HEART DISORDERS PANEL ¹ INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS
KCNN4	100%	100%	100%	98.6%	Dehydrated hereditary stomatocytosis 2, 616689	MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS
KCNQ1	100%	99.6%	100%	98.4%	Short QT syndrome 2, 609621;Atrial fibrillation, familial, 3, 607554;Long QT syndrome 1, 192500;{Long QT syndrome 1, acquired, susceptibility to}, 192500;Jervell and Lange-Nielsen syndrome, 220400	ARRHYTHMIA AND CARDIAC CONDUCTION DISORDERS PANEL ¹ COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹ MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS LONG QT SYNDROME PANEL ¹ HEART DISORDERS PANEL ¹ HEARING IMPAIRMENT PANEL (INCLUDING GJB2)

KCNQ1OT1					Beckwith-Wiedemann syndrome, 130650	TALL STATURE PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS
KCNQ2	100%	100%	100%	98.8%	Developmental and epileptic encephalopathy 7, 613720;Seizures, benign neonatal, 1, 121200;Myokymia, 121200	INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS EPILEPSY PANEL
KCNQ3	100%	100%	100%	99%	Seizures, benign neonatal, 2, 121201	EPILEPSY PANEL NEUROLOGICAL PAIN DISORDERS PANEL ¹ INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS

KCNQ4	99.5%	98.6%	100%	97.9%	Deafness, autosomal dominant 2A, 600101	HEARING IMPAIRMENT PANEL (INCLUDING GJB2) MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS
KCNQ5	100%	100%	100%	99.4%	Intellectual developmental disorder, autosomal dominant 46, 617601	INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS
KCNT1	100%	99.8%	100%	98.4%	Developmental and epileptic encephalopathy 14, 614959;Epilepsy nocturnal frontal lobe, 5, 615005	EPILEPSY PANEL INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS

KCNT2	100%	100%	100%	99.4%	Developmental and epileptic encephalopathy 57, 617771	MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS EPILEPSY PANEL INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS
KCNU1	100%	100%	100%	99.5%	Spermatogenic failure 79, 620196	MALE INFERTILITY PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS
KCNV2	100%	100%	100%	99.4%	Retinal cone dystrophy 3B, 610356	VISION DISORDERS PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹
KCTD1	100%	100%	100%	96.1%	Scalp-ear-nipple syndrome, 181270	SKIN DISORDERS PANEL ¹ MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS

KCTD17	100%	100%	100%	96.9%	Dystonia 26, myoclonic, 616398	MOVEMENT DISORDERS PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS
KCTD19	100%	100%	100%	99.5%		MALE INFERTILITY PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS
KCTD3	100%	100%	100%	99.5%		CILIOPATHIES PANEL INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS

KCTD7	100%	100%	100%	99.3%	Epilepsy, progressive myoclonic 3, with or without intracellular inclusions, 611726	MOVEMENT DISORDERS PANEL EPILEPSY PANEL INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL¹
KDELR2	100%	100%	100%	98%	Osteogenesis imperfecta, type XXI, 619131	SHORT STATURE/SKELETAL DYSPLASIA PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL¹

KDF1	100%	100%	100%	98.6%	?Ectodermal dysplasia 12, hypohidrotic/hair/tooth/nail type, 617337	CRANIOFACIAL ANOMALIES PANEL WITH GENOME WIDE CNV ANALYSIS SKIN DISORDERS PANEL ¹ MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS
KDM1A	96.9%	96.9%	100%	99.2%	Cleft palate, psychomotor retardation, and distinctive facial features, 616728;{ACTH-independent macronodular adrenal hyperplasia 3}, 620990	INHERITED BONE MARROW FAILURE AND/OR PREDISPOSITION PANEL TO HEMATOLOGICAL MALIGNANCIES CRANIOFACIAL ANOMALIES PANEL WITH GENOME WIDE CNV ANALYSIS INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS

KDM2A	100%	100%	100%	99.1%		INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS
KDM2B	100%	100%	100%	98.3%		INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS
KDM3B	100%	100%	100%	99.2%	Diets-Jongmans syndrome, 618846	INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS

KDM4B	100%	99.9%	100%	98%	Intellectual developmental disorder, autosomal dominant 65, 619320	INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS
KDM5A	100%	100%	100%	99.3%	El Hayek-Chahrour neurodevelopmental syndrome, 620820	INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹
KDM5B	97.7%	96.8%	100%	99.4%	Intellectual developmental disorder, autosomal recessive 65, 618109	INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹

KDM5C	97.9%	97.5%	98%	65.7%	Intellectual developmental disorder, X-linked syndromic, Claes-Jensen type, 300534	EPILEPSY PANEL INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS
KDM5D	48.9%	48.8%	49.2%	18.3%		MALE INFERTILITY PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS

KDM6A	100%	100%	99.1%	70.8%	Kabuki syndrome 2, 300867	CRANIOFACIAL ANOMALIES PANEL WITH GENOME WIDE CNV ANALYSIS PRIMARY IMMUNODEFICIENCIES PANEL INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS OROFACIAL CLEFTING PANEL WITH GENOME WIDE CNV ANALYSIS
KDM6B	100%	99.9%	100%	97.5%	Stolerman neurodevelopmental syndrome, 618505	EPILEPSY PANEL INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS

KDR	100%	100%	100%	99.5%	{Hemangioma, capillary infantile, susceptibility to}, 602089;Hemangioma, capillary infantile, somatic, 602089	CONGENITAL HEARTDISEASE PANEL¹ HEART DISORDERS PANEL¹ MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS
KDSR	100%	100%	100%	98.6%	Erythrokeratoderma variabilis et progressiva 4, 617526	SKIN DISORDERS PANEL¹ HEMOSTATIC/THROMB OTIC DISORDERS PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL¹
KERA	100%	100%	100%	99.6%	Cornea plana 2, autosomal recessive, 217300	VISION DISORDERS PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL¹

KHDC3L	100%	100%	100%	99.2%	Hydatidiform mole, recurrent, 2, 614293	UNSUCCESSFUL IVF TREATMENTS, RECURRENT MOLAR PREGNANCIES AND/OR RECURRENT PREGNANCY LOSS PANEL ¹ MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹
KHK	100%	100%	100%	99.1%	?[Fructosuria, essential], 229800	MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS

KIAA0586	95.6%	95.6%	100%	99.5%	Short-rib thoracic dysplasia 14 with polydactyly, 616546;Joubert syndrome 23, 616490	COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹ OROFACIAL CLEFTING PANEL WITH GENOME WIDE CNV ANALYSIS VISION DISORDERS PANEL CILIOPATHIES PANEL SHORT STATURE/SKELETAL DYSPLASIA PANEL WITH GENOME WIDE CNV ANALYSIS INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS
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KIAA0753	100%	100%	100%	99.5%	?Orofaciodigital syndrome XV, 617127;?Joubert syndrome 38, 619476;Short-rib thoracic dysplasia 21 without polydactyly, 619479	VISION DISORDERS PANEL CILIOPATHIES PANEL SHORT STATURE/SKELETAL DYSPLASIA PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹
KIAA0825	100%	100%	100%	99.5%	Polydactyly, postaxial, type A10, 618498	SHORT STATURE/SKELETAL DYSPLASIA PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹

KIAA1549	99.6%	99.3%	100%	98.6%	Retinitis pigmentosa 86, 618613	VISION DISORDERS PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL¹
KICS2	100%	100%	100%	99.6%		INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS
KIDINS220	100%	100%	100%	99.5%	Spastic paraplegia, intellectual disability, nystagmus, and obesity, 617296;Ventriculomegaly and arthrogryposis, 619501	FETAL AKINESIA PANEL MOVEMENT DISORDERS PANEL INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL¹

KIF11	100%	100%	100%	99.7%	Microcephaly with or without chorioretinopathy, lymphedema, or impaired intellectual development, 152950	VISION DISORDERS PANEL SKIN DISORDERS PANEL ¹ LYMPHATIC ANOMALIES PANEL WITH GENOME WIDE CNV ANALYSIS INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS
KIF12	100%	100%	100%	98.9%	Cholestasis, progressive familial intrahepatic, 8, 619662	LIVER DISORDERS PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹

KIF14	100%	100%	100%	99.6%	Microcephaly 20, primary, autosomal recessive, 617914;?Meckel syndrome 12, 616258	CILIOPATHIES PANEL INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS RENAL DISORDERS PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL¹
KIF15	100%	100%	100%	99.2%	?Braddock-Carey syndrome 2, 619981	MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS

KIF1A	100%	100%	100%	99.3%	NESCAV syndrome, 614255;Neuropathy, hereditary sensory, type IIC, 614213;Spastic paraplegia 30, autosomal dominant, 610357;Spastic paraplegia 30, autosomal recessive, 620607	MOVEMENT DISORDERS PANEL POLYNEUROPATHIES PANEL ¹ INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS MITOCHONDRIAL DISORDERS PANEL NEUROLOGICAL PAIN DISORDERS PANEL ¹ COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹
KIF1B	94.9%	94.9%	100%	99.5%	{Neuroblastoma, susceptibility to, 1}, 256700;Charcot-Marie-Tooth disease, type 2A1, 118210	POLYNEUROPATHIES PANEL ¹ MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS HEREDITARY CANCER PANEL

KIF1C	100%	100%	100%	98.5%	Spastic ataxia 2, autosomal recessive, 611302	MOVEMENT DISORDERS PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹
KIF20A	100%	100%	100%	99.6%	?Cardiomyopathy, familial restrictive, 6, 619433	CONGENITAL HEARTDISEASE PANEL ¹ HEART DISORDERS PANEL ¹ MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS
KIF21A	100%	100%	100%	99.3%	Fibrosis of extraocular muscles, congenital, 3B, 135700;Fibrosis of extraocular muscles, congenital, 1, 135700	VISION DISORDERS PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS MUSCLE DISORDERS PANEL

KIF21B	100%	100%	100%	99.3%		INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS
KIF22	100%	100%	100%	99.1%	Spondyloepimetaphyseal dysplasia with joint laxity, type 2, 603546	SHORT STATURE/SKELETAL DYSPLASIA PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS
KIF23	100%	100%	100%	99.4%	Anemia, congenital dyserythropoietic, type IIIA, 105600	INHERITED BONE MARROW FAILURE AND/OR PREDISPOSITION PANEL TO HEMATOLOGICAL MALIGNANCIES IRON DISORDERS PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS

KIF24	100%	100%	100%	99.6%		SHORT STATURE/SKELETAL DYSPLASIA PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS
KIF26A	97.7%	97.6%	100%	99.5%	Cortical dysplasia, complex, with other brain malformations 11, 620156	INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹
KIF2A	100%	100%	100%	99.3%	Cortical dysplasia, complex, with other brain malformations 3, 615411	INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS

KIF3B	100%	100%	100%	99.1%	Retinitis pigmentosa 89, 618955	VISION DISORDERS PANEL INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS
KIF4A	100%	99.8%	99%	68.6%	Taurodontism, microdontia, and dens invaginatus, 313490;Intellectual developmental disorder, X-linked 100, 300923	INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS

KIF5A	100%	100%	100%	98.7%	Myoclonus, intractable, neonatal, 617235;{Amyotrophic lateral sclerosis, susceptibility to, 25}, 617921;Spastic paraplegia 10, autosomal dominant, 604187	AMYOTROPHIC LATERAL SCLEROSIS, ALS PANEL MOVEMENT DISORDERS PANEL EPILEPSY PANEL POLYNEUROPATHIES PANEL ¹ INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS
KIF5B	100%	100%	100%	99.5%		SHORT STATURE/SKELETAL DYSPLASIA PANEL WITH GENOME WIDE CNV ANALYSIS INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS

KIF5C	100%	100%	100%	99.4%	Cortical dysplasia, complex, with other brain malformations 2, 615282	FETAL AKINESIA PANEL INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS
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KIF7	99.9%	99.3%	100%	98%	Joubert syndrome 12, 200990;Acrocallosal syndrome, 200990;?Hydrolethalus syndrome 2, 614120;?Al-Gazali-Bakalinova syndrome, 607131	VISION DISORDERS PANEL CILIOPATHIES PANEL OROFACIAL CLEFTING PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹ INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS RENAL DISORDERS PANEL SHORT STATURE/SKELETAL DYSPLASIA PANEL WITH GENOME WIDE CNV ANALYSIS
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KIFBP	95.9%	95.8%	100%	99.3%	Goldberg-Shprintzen megacolon syndrome, 609460	INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹ OROFACIAL CLEFTING PANEL WITH GENOME WIDE CNV ANALYSIS
KIRREL1	100%	100%	100%	98.5%	Nephrotic syndrome, type 23, 619201	RENAL DISORDERS PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹
KIRREL3	100%	100%	100%	97.4%		INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS

KISS1	100%	100%	100%	97.5%	?Hypogonadotropic hypogonadism 13 with or without anosmia, 614842	DISORDERS/DIFFERENCES OF SEX DEVELOPMENT (DSD) / PRIMARY ADRENAL INSUFFICIENCY PANEL HYPOGONADOTROPIC HYPOGONADISM (KALLMANN) PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS
KISS1R	100%	100%	100%	98.6%	Hypogonadotropic hypogonadism 8 with or without anosmia, 614837;?Precocious puberty, central, 1, 176400	DISORDERS/DIFFERENCES OF SEX DEVELOPMENT (DSD) / PRIMARY ADRENAL INSUFFICIENCY PANEL HYPOGONADOTROPIC HYPOGONADISM (KALLMANN) PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹

KIT	100%	100%	100%	99.5%	Gastrointestinal stromal tumor, familial, 606764;Mastocytosis, cutaneous, 154800;Piebaldism, 172800;Germ cell tumors, somatic, 273300;Mastocytosis, systemic, somatic, 154800;Leukemia, acute myeloid, somatic, 601626	INHERITED BONE MARROW FAILURE AND/OR PREDISPOSITION PANEL TO HEMATOLOGICAL MALIGNANCIES SKIN DISORDERS PANEL ¹ MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS HEREDITARY CANCER PANEL
KITLG	100%	100%	100%	99.4%	Hyperpigmentation with or without hypopigmentation, 145250;Waardenburg syndrome, type 2F, 619947;Deafness, autosomal dominant 69, unilateral or asymmetric, 616697;[Skin/hair/eye pigmentation 7, blond/brown hair], 611664	SKIN DISORDERS PANEL ¹ HEARING IMPAIRMENT PANEL (INCLUDING GJB2) MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹

KIZ	100%	100%	100%	99.4%	Retinitis pigmentosa 69, 615780	VISION DISORDERS PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹
KL	99.5%	98.7%	99.9%	98.4%	?Tumoral calcinosis, hyperphosphatemic, familial, 3, 617994	SHORT STATURE/SKELETAL DYSPLASIA PANEL WITH GENOME WIDE CNV ANALYSIS RENAL DISORDERS PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹

KLB	100%	100%	100%	99.5%		DISORDERS/DIFFERENCES OF SEX DEVELOPMENT (DSD) / PRIMARY ADRENAL INSUFFICIENCY PANEL HYPOGONADOTROPIC HYPOGONADISM (KALLMANN) PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS
KLC2	100%	100%	100%	99.4%	Spastic paraplegia, optic atrophy, and neuropathy, 609541	POLYNEUROPATHIES PANEL ¹ MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹

KLF1	100%	100%	100%	99.1%	Blood group--Lutheran inhibitor, 111150;[Hereditary persistence of fetal hemoglobin], 613566;Anemia, dyserythropoietic congenital, type IVa, 613673;Anemia, congenital dyserythropoietic, type IVb, 620969	INHERITED BONE MARROW FAILURE AND/OR PREDISPOSITION PANEL TO HEMATOLOGICAL MALIGNANCIES MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹ IRON DISORDERS PANEL
KLF11	100%	100%	100%	99%	Maturity-onset diabetes of the young, type VII, 610508	MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS
KLF13	100%	99.7%	99.9%	91.2%		CONGENITAL HEARTDISEASE PANEL ¹ HEART DISORDERS PANEL ¹ MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS

KLF4	100%	100%	100%	99.2%		SKIN DISORDERS PANEL ¹ MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS
KLF6	100%	100%	100%	98.9%	Gastric cancer, somatic, 613659;Prostate cancer, somatic, 176807	MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS
KLF7	100%	100%	100%	99%		INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS
KLHL10	100%	100%	100%	99.2%	Spermatogenic failure 11, 615081	MALE INFERTILITY PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS

KLHL15	89.1%	89.1%	99.2%	69.9%	Intellectual developmental disorder, X-linked 103, 300982	INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS
KLHL20	100%	100%	100%	99.6%		INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS
KLHL24	100%	100%	100%	99.7%	Cardiomyopathy, familial hypertrophic, 29, with polyglucosan bodies, 620236;Epidermolysis bullosa simplex 6, generalized intermediate, with or without cardiomyopathy, 617294	SKIN DISORDERS PANEL ¹ HEART DISORDERS PANEL ¹ HYPERTROPHIC CARDIOMYOPATHY PANEL ¹ MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹

KLHL3	100%	100%	100%	99.2%	Pseudohypoaldosteronism, type IID, 614495	RENAL DISORDERS PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹
KLHL40	100%	100%	100%	98.8%	Nemaline myopathy 8, autosomal recessive, 615348	FETAL AKINESIA PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹ MUSCLE DISORDERS PANEL
KLHL41	100%	100%	100%	99.8%	Nemaline myopathy 9, 615731	FETAL AKINESIA PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹ MUSCLE DISORDERS PANEL

KLHL7	100%	100%	100%	99.5%	Retinitis pigmentosa 42, 612943;PERCHING syndrome, 617055	VISION DISORDERS PANEL INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹
KLHL9	100%	100%	100%	99.5%		MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS MUSCLE DISORDERS PANEL
KLK11	100%	100%	100%	98.3%	Ichthyosis with erythrokeratoderma, 620507	SKIN DISORDERS PANEL ¹ MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS

KLK4	100%	100%	100%	98%	Amelogenesis imperfecta, type IIA1, 204700	CRANIOFACIAL ANOMALIES PANEL WITH GENOME WIDE CNV ANALYSIS SKIN DISORDERS PANEL ¹ MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹
KLKB1	100%	100%	100%	99.8%	Fletcher factor (prekallikrein) deficiency, 612423	HEMOSTATIC/THROMB OTIC DISORDERS PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹
KLLN	100%	100%	100%	99.7%	Cowden syndrome 4, 615107	SKIN DISORDERS PANEL ¹ MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS

KMT2A	99.2%	99.2%	100%	99.3%	Wiedemann-Steiner syndrome, 605130	CONGENITAL HEARTDISEASE PANEL¹ HEART DISORDERS PANEL¹ PRIMARY IMMUNODEFICIENCIES PANEL SHORT STATURE/SKELETAL DYSPLASIA PANEL WITH GENOME WIDE CNV ANALYSIS METABOLIC DISORDERS PANEL INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS
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KMT2B	99.5%	99.1%	100%	97.5%	Intellectual developmental disorder, autosomal dominant 68, 619934;Dystonia 28, childhood-onset, 617284	MOVEMENT DISORDERS PANEL INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹
KMT2C	100%	100%	100%	99.3%	Kleefstra syndrome 2, 617768	INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS

KMT2D	100%	100%	100%	98.3%	Branchial arch abnormalities, choanal atresia, athelia, hearing loss, and hypothyroidism syndrome, 620186;Kabuki syndrome 1, 147920	CRANIOFACIAL ANOMALIES PANEL WITH GENOME WIDE CNV ANALYSIS CONGENITAL HEARTDISEASE PANEL ¹ SKIN DISORDERS PANEL ¹ HEART DISORDERS PANEL ¹ PRIMARY IMMUNODEFICIENCIES PANEL METABOLIC DISORDERS PANEL INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS OROFACIAL CLEFTING PANEL WITH GENOME WIDE CNV ANALYSIS
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KMT2E	100%	100%	100%	98.8%	O'Donnell-Luria-Rodan syndrome, 618512	INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS
KMT5B	100%	100%	100%	99.2%	Intellectual developmental disorder, autosomal dominant 51, 617788	EPILEPSY PANEL INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS
KNG1	100%	100%	100%	99.3%	[Kininogen deficiency], 228960;Angioedema, hereditary, 6, 619363;[High molecular weight kininogen deficiency], 228960	HEMOSTATIC/THROMB OTIC DISORDERS PANEL PRIMARY IMMUNODEFICIENCIES PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS

KNL1	98.7%	98.7%	100%	99.6%	Microcephaly 4, primary, autosomal recessive, 604321	INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹
KNSTRN	100%	100%	100%	99.5%	?Roifman-Chitayat syndrome, digenic, 613328	MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS
KPNA3	100%	100%	100%	99.5%	Spastic paraplegia 88, autosomal dominant, 620106	MOVEMENT DISORDERS PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS

KPNA7	100%	100%	100%	98.9%	Oocyte/zygote/embryo maturation arrest 17, 620319	UNSUCCESSFUL IVF TREATMENTS, RECURRENT MOLAR PREGNANCIES AND/OR RECURRENT PREGNANCY LOSS PANEL ¹ MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS
KPTN	100%	100%	100%	97%	Intellectual developmental disorder, autosomal recessive 41, 615637	EPILEPSY PANEL INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹

KRAS	100%	100%	100%	99.6%	Gastric cancer, somatic, 613659;Oculoectodermal syndrome, somatic, 600268;Breast cancer, somatic, 114480;Noonan syndrome 3, 609942;RAS-associated autoimmune leukoproliferative disorder, 614470;Arteriovenous malformation of the brain, somatic, 108010;Lung cancer, somatic, 211980;Pancreatic carcinoma, somatic, 260350;Leukemia, acute myeloid, somatic, 601626;Schimmelpenning-Feuerstein-Mims syndrome, somatic mosaic, 163200;Cardiofaciocutaneous syndrome 2, 615278;Bladder cancer, somatic, 109800	INHERITED BONE MARROW FAILURE AND/OR PREDISPOSITION PANEL TO HEMATOLOGICAL MALIGNANCIES CONGENITAL HEARTDISEASE PANEL ¹ INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS NOONAN SYNDROME / RASOPATHY PANEL HEREDITARY CANCER PANEL HEMOSTATIC/THROMBOTIC DISORDERS PANEL PRIMARY IMMUNODEFICIENCIES PANEL SHORT STATURE/SKELETAL DYSPLASIA PANEL WITH GENOME WIDE CNV ANALYSIS LYMPHATIC ANOMALIES PANEL WITH GENOME WIDE
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						CNV ANALYSIS SKIN DISORDERS PANEL ¹ HEART DISORDERS PANEL ¹
KREMEN1	100%	99.7%	100%	99.1%	Ectodermal dysplasia 13, hair/tooth type, 617392	CRANIOFACIAL ANOMALIES PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹
KRIT1	100%	100%	100%	99.8%	Hyperkeratotic cutaneous capillary-venous malformations associated with cerebral capillary malformations, 116860;Cerebral cavernous malformations-1, 116860;Cavernous malformations of CNS and retina, 116860	EPILEPSY PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS
KRT1	100%	100%	100%	98.8%	Ichthyosis, annular epidermolytic 2, 620148;Palmoplantar keratoderma, nonepidermolytic, 600962;Epidermolytic hyperkeratosis 1, 113800;Palmoplantar keratoderma, epidermolytic, 2, 620411;Keratosis palmoplantaris striata III, 607654;Ichthyosis hystrix, Curth-Macklin type, 146590	SKIN DISORDERS PANEL ¹ MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS

KRT10	100%	100%	100%	98.3%	Ichthyosis, annular epidermolytic 1, 607602;Epidermolytic hyperkeratosis 2B, autosomal recessive, 620707;Epidermolytic hyperkeratosis 2A, autosomal dominant, 620150;?Ichthyosis histrix, Lambert type, 146600;Ichthyosis with confetti, 609165	SKIN DISORDERS PANEL ¹ MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹
KRT12	100%	100%	100%	99%	Meesmann corneal dystrophy 1, 122100	VISION DISORDERS PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS
KRT13	100%	100%	100%	98.9%	White sponge nevus 2, 615785	SKIN DISORDERS PANEL ¹ MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS
KRT14	100%	100%	100%	99%	Epidermolysis bullosa simplex 1D, generalized, intermediate or severe, autosomal recessive, 601001;Epidermolysis bullosa simplex 1C, localized, 131800;Dermatopathia pigmentosa reticularis, 125595;Epidermolysis bullosa simplex 1A, generalized severe, 131760;Naegeli-Franceschetti-Jadassohn syndrome, 161000;Epidermolysis bullosa simplex 1B, generalized intermediate, 131900	SKIN DISORDERS PANEL ¹ MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹

KRT16	100%	100%	100%	98.8%	Palmoplantar keratoderma, nonepidermolytic, focal, 613000;Pachyonychia congenita 1, 167200	SKIN DISORDERS PANEL ¹ MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS
KRT17	100%	100%	100%	98.9%	Steatocystoma multiplex, 184500;Pachyonychia congenita 2, 167210	SKIN DISORDERS PANEL ¹ MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS
KRT18	100%	100%	100%	98.8%	Cirrhosis, cryptogenic, 215600;{Cirrhosis, noncryptogenic, susceptibility to}, 215600	MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹
KRT2	100%	100%	100%	98.7%	Ichthyosis bullosa of Siemens, 146800	SKIN DISORDERS PANEL ¹ MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS
KRT25	100%	100%	100%	99.4%	Woolly hair, autosomal recessive 3, 616760	MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS

KRT3	100%	100%	100%	99.3%	Meesmann corneal dystrophy 2, 618767	VISION DISORDERS PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS
KRT4	100%	100%	100%	98.3%	White sponge nevus 1, 193900	SKIN DISORDERS PANEL ¹ MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS
KRT5	100%	100%	100%	98.8%	Epidermolysis bullosa simplex 2A, generalized severe, 619555;Dowling-Degos disease 1, 179850;Epidermolysis bullosa simplex 2F, with mottled pigmentation, 131960;Epidermolysis bullosa simplex 2D, generalized, intermediate or severe, autosomal recessive, 619599;Epidermolysis bullosa simplex 2B, generalized intermediate, 619588;Epidermolysis bullosa simplex 2C, localized, 619594;Epidermolysis bullosa simplex 2E, with migratory circinate erythema, 609352	SKIN DISORDERS PANEL ¹ MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹
KRT6A	100%	100%	100%	99.2%	Pachyonychia congenita 3, 615726	SKIN DISORDERS PANEL ¹ MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS

KRT6B	100%	100%	100%	99.6%	Pachyonychia congenita 4, 615728	SKIN DISORDERS PANEL ¹ MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS
KRT6C	99.7%	99.4%	98.7%	92%	Palmoplantar keratoderma, nonepidermolytic, focal or diffuse, 615735	SKIN DISORDERS PANEL ¹ MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS
KRT71	100%	100%	100%	99.3%	?Hypotrichosis 13, 615896	SKIN DISORDERS PANEL ¹ MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS
KRT74	100%	100%	100%	99%	Woolly hair, autosomal dominant, 194300;?Hypotrichosis 3, 613981;?Ectodermal dysplasia 7, hair/nail type, 614929	SKIN DISORDERS PANEL ¹ MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS

KRT75	100%	100%	100%	99.7%	{Pseudofolliculitis barbae, susceptibility to}, 612318	SKIN DISORDERS PANEL ¹ MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS
KRT8	100%	100%	100%	98.7%		MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹
KRT81	100%	100%	100%	98.6%	Monilethrix, 158000	SKIN DISORDERS PANEL ¹ MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS
KRT82	100%	100%	100%	99.1%		SKIN DISORDERS PANEL ¹ MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS

KRT83	100%	100%	100%	99.5%	Monilethrix, 158000;Erythrokeratoderma variabilis et progressiva 5, 617756	SKIN DISORDERS PANEL ¹ MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS
KRT85	100%	100%	100%	99%	Ectodermal dysplasia 4, hair/nail type, 602032	SKIN DISORDERS PANEL ¹ MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹
KRT86	100%	100%	100%	99.2%	Monilethrix, 158000	SKIN DISORDERS PANEL ¹ MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS
KRT9	100%	100%	100%	97.7%	Palmoplantar keratoderma, epidermolytic, 1, 144200	SKIN DISORDERS PANEL ¹ MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS

KY	100%	100%	100%	99.4%	Myopathy, myofibrillar, 7, 617114	MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹
KYNU	100%	100%	100%	99.3%	?Hydroxykynureninuria, 236800;Vertebral, cardiac, renal, and limb defects syndrome 2, 617661	SHORT STATURE/SKELETAL DYSPLASIA PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹
L1CAM	100%	99.7%	98.2%	66.3%	MASA syndrome, 303350;Hydrocephalus, congenital, X-linked, 307000;?Corpus callosum, partial agenesis of, 304100	MOVEMENT DISORDERS PANEL LIVER DISORDERS PANEL INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS

L2HGDH	100%	100%	100%	99.2%	L-2-hydroxyglutaric aciduria, 236792	METABOLIC DISORDERS PANEL INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹
LACC1	100%	100%	100%	99.5%	Juvenile arthritis, 618795	PRIMARY IMMUNODEFICIENCIES PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹
LACTB	100%	100%	100%	99%		MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS MITOCHONDRIAL DISORDERS PANEL

LAGE3	100%	100%	98.9%	68.4%	Galloway-Mowat syndrome 2, X-linked, 301006	INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS RENAL DISORDERS PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS
LAMA1	100%	100%	100%	99.1%	Poretti-Boltshauser syndrome, 615960	MOVEMENT DISORDERS PANEL VISION DISORDERS PANEL INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹

LAMA2	100%	100%	100%	99.5%	Muscular dystrophy, limb-girdle, autosomal recessive 23, 618138;Muscular dystrophy, congenital, merosin deficient or partially deficient, 607855	HEART DISORDERS PANEL ¹ POLYNEUROPATHIES PANEL ¹ INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹ MUSCLE DISORDERS PANEL
LAMA3	100%	100%	100%	99.4%	Epidermolysis bullosa, junctional 2A, intermediate, 619783;Epidermolysis bullosa, junctional 2C, laryngoonychocutaneous, 245660;Epidermolysis bullosa, junctional 2B, severe, 619784	SKIN DISORDERS PANEL ¹ MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹
LAMA4	100%	100%	100%	99.6%	Cardiomyopathy, dilated, 1JJ, 615235	HEART DISORDERS PANEL ¹ MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS

LAMA5	100%	100%	100%	98.2%	Nephrotic syndrome, type 26, 620049;?Bent bone dysplasia syndrome 2, 620076	SHORT STATURE/SKELETAL DYSPLASIA PANEL WITH GENOME WIDE CNV ANALYSIS RENAL DISORDERS PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL¹
LAMB1	100%	99.9%	100%	99.2%	Lissencephaly 5, 615191	MOVEMENT DISORDERS PANEL EPILEPSY PANEL INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL¹

LAMB2	100%	100%	100%	99.5%	Nephrotic syndrome, type 5, with or without ocular abnormalities, 614199;Pierson syndrome, 609049	VISION DISORDERS PANEL INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS RENAL DISORDERS PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹
LAMB3	100%	100%	100%	98.6%	Epidermolysis bullosa, junctional 1B, severe, 226700;Epidermolysis bullosa, junctional 1A, intermediate, 226650;Amelogenesis imperfecta, type IA, 104530	CRANIOFACIAL ANOMALIES PANEL WITH GENOME WIDE CNV ANALYSIS SKIN DISORDERS PANEL ¹ MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹

LAMC2	100%	100%	100%	99.3%	Epidermolysis bullosa, junctional 3B, severe, 619786;Epidermolysis bullosa, junctional 3A, intermediate, 619785	SKIN DISORDERS PANEL ¹ MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹
LAMC3	100%	100%	100%	99.1%	Cortical malformations, occipital, 614115	INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹

LAMP2	85.3%	85.3%	98.6%	67.8%	Danon disease, 300257	VISION DISORDERS PANEL HEART DISORDERS PANEL ¹ HYPERTROPHIC CARDIOMYOPATHY PANEL ¹ METABOLIC DISORDERS PANEL INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS MUSCLE DISORDERS PANEL
LAMTOR2	100%	100%	100%	99.8%	Immunodeficiency due to defect in MAPBP-interacting protein, 610798	SKIN DISORDERS PANEL ¹ PRIMARY IMMUNODEFICIENCIES PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹

LAPTM5	100%	99.8%	100%	99.2%		INHERITED BONE MARROW FAILURE AND/OR PREDISPOSITION PANEL TO HEMATOLOGICAL MALIGNANCIES MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS
LARGE1	100%	100%	100%	99.3%	Muscular dystrophy-dystroglycanopathy (congenital with impaired intellectual development), type B, 6, 608840;Muscular dystrophy-dystroglycanopathy (congenital with brain and eye anomalies), type A, 6, 613154	METABOLIC DISORDERS PANEL INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹ MUSCLE DISORDERS PANEL

LARP1	100%	100%	100%	98.9%		MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS
LARP7	100%	100%	100%	99.4%	Alazami syndrome, 615071	COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹ INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS
LARS1	100%	100%	100%	99.3%	?Infantile liver failure syndrome 1, 615438	LIVER DISORDERS PANEL INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹

LARS2	100%	100%	100%	99.5%	Perrault syndrome 4, 615300;Hydrops, lactic acidosis, and sideroblastic anemia, 617021	HEARING IMPAIRMENT PANEL (INCLUDING GJB2) DISORDERS/DIFFEREN CES OF SEX DEVELOPMENT (DSD) / PRIMARY ADRENAL INSUFFICIENCY PANEL IRON DISORDERS PANEL LIVER DISORDERS PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS MITOCHONDRIAL DISORDERS PANEL COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹ PREMATURE OVARIAN INSUFFICIENCY PANEL
LAS1L	95.8%	95.5%	97.8%	65.3%	Wilson-Turner syndrome, 309585	INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS

LAT	100%	100%	100%	99.6%	Immunodeficiency 52, 617514	PRIMARY IMMUNODEFICIENCIES PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL¹ SEVERE COMBINED IMMUNODEFICIENCY (SCID) PANEL
LBR	100%	100%	100%	99.5%	Pelger-Huet anomaly, 169400;?Reynolds syndrome, 613471;Rhizomelic skeletal dysplasia with or without Pelger-Huet anomaly, 618019;Greenberg skeletal dysplasia, 215140	CILIOPATHIES PANEL SHORT STATURE/SKELETAL DYSPLASIA PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL¹

LBX1	100%	100%	100%	98%	?Central hypoventilation syndrome, congenital, 3, 619483	CONGENITAL HEARTDISEASE PANEL¹ HEART DISORDERS PANEL¹ SHORT STATURE/SKELETAL DYSPLASIA PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS
LCA5	100%	100%	100%	99.1%	Leber congenital amaurosis 5, 604537	VISION DISORDERS PANEL CILIOPATHIES PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL¹

LCAT	100%	100%	100%	98.7%	Fish-eye disease, 136120;Norum disease, 245900	VISION DISORDERS PANEL METABOLIC DISORDERS PANEL RENAL DISORDERS PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL¹
LCK	100%	100%	100%	98.7%	Immunodeficiency 22, 615758	PRIMARY IMMUNODEFICIENCIES PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL¹ SEVERE COMBINED IMMUNODEFICIENCY (SCID) PANEL

LCP1	100%	100%	100%	99.5%		INHERITED BONE MARROW FAILURE AND/OR PREDISPOSITION PANEL TO HEMATOLOGICAL MALIGNANCIES PRIMARY IMMUNODEFICIENCIES PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS
LCP2	100%	100%	100%	99.1%	Immunodeficiency 81, 619374	PRIMARY IMMUNODEFICIENCIES PANEL COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹ SEVERE COMBINED IMMUNODEFICIENCY (SCID) PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS

LCT	100%	100%	100%	99.2%	Lactase deficiency, congenital, 223000	METABOLIC DISORDERS PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹
LDB3	100%	100%	100%	98%	Left ventricular noncompaction 3, 601493;Cardiomyopathy, hypertrophic, 24, 601493;Myopathy, myofibrillar, 4, 609452;Cardiomyopathy, dilated, 1C, with or without LVNC, 601493	HEART DISORDERS PANEL ¹ MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS MUSCLE DISORDERS PANEL
LDHA	100%	100%	100%	99.6%	Glycogen storage disease XI, 612933	MUSCLE DISORDERS PANEL SKIN DISORDERS PANEL ¹ METABOLIC DISORDERS PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹

LDHB	100%	100%	100%	98.7%	[Lactate dehydrogenase-B deficiency], 614128	METABOLIC DISORDERS PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS
LDHD	100%	100%	100%	99.6%	D-lactic aciduria with susceptibility to gout, 245450	MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS MITOCHONDRIAL DISORDERS PANEL COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹
LDLR	100%	100%	100%	98.4%	LDL cholesterol level QTL2, 143890;Hypercholesterolemia, familial, 1, 143890	MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹
LDLRAP1	100%	100%	100%	98.2%	Hypercholesterolemia, familial, 4, 603813	SKIN DISORDERS PANEL ¹ MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS

LEF1	100%	100%	100%	99.6%		MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS
LEMD2	100%	100%	100%	98.8%	Marbach-Rustad progeroid syndrome, 619322;Cataract 46, juvenile-onset, 212500	VISION DISORDERS PANEL HEART DISORDERS PANEL ¹ MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹
LEMD3	100%	100%	100%	98.7%	Buschke-Ollendorff syndrome, 166700;Osteopoikilosis with or without melorheostosis, 166700	SKIN DISORDERS PANEL ¹ SHORT STATURE/SKELETAL DYSPLASIA PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS

LEP	100%	100%	100%	98.5%	Obesity, morbid, due to leptin deficiency, 614962	DISORDERS/DIFFERENCES OF SEX DEVELOPMENT (DSD) / PRIMARY ADRENAL INSUFFICIENCY PANEL HYPOGONADOTROPIC HYPOGONADISM (KALLMANN) PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹
LEPR	94.6%	94.6%	100%	99.6%	Obesity, morbid, due to leptin receptor deficiency, 614963	DISORDERS/DIFFERENCES OF SEX DEVELOPMENT (DSD) / PRIMARY ADRENAL INSUFFICIENCY PANEL HYPOGONADOTROPIC HYPOGONADISM (KALLMANN) PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹

LETM1	100%	100%	100%	98.7%	Neurodegeneration, childhood-onset, with multisystem involvement due to mitochondrial dysfunction, 620089	INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS MITOCHONDRIAL DISORDERS PANEL COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹
LFNG	96.1%	90.5%	99.9%	96.7%	Spondylocostal dysostosis 3, autosomal recessive, 609813	SHORT STATURE/SKELETAL DYSPLASIA PANEL WITH GENOME WIDE CNV ANALYSIS METABOLIC DISORDERS PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹
LGI1	100%	100%	100%	99.7%	Epilepsy, familial temporal lobe, 1, 600512	EPILEPSY PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS

LGI3	100%	100%	100%	99.4%	Intellectual developmental disorder with muscle tone abnormalities and distal skeletal defects, 620007	INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹
LGI4	100%	100%	100%	98.4%	Arthrogryposis multiplex congenita 1, neurogenic, with myelin defect, 617468	FETAL AKINESIA PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹
LGR4	100%	100%	100%	98.7%	Delayed puberty, self-limited, 619613;{Bone mineral density, low, susceptibility to}, 615311	MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS

LHB	100%	100%	100%	98.4%	Hypogonadotropic hypogonadism 23 with or without anosmia, 228300	DISORDERS/DIFFERENCES OF SEX DEVELOPMENT (DSD) / PRIMARY ADRENAL INSUFFICIENCY PANEL HYPOGONADOTROPIC HYPOGONADISM (KALLMANN) PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹
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LHCGR	100%	100%	100%	99.1%	Leydig cell adenoma, somatic, with precocious puberty, 176410;Leydig cell hypoplasia with pseudohermaphroditism, 238320;Leydig cell hypoplasia with hypergonadotropic hypogonadism, 238320;Luteinizing hormone resistance, female, 238320;Precocious puberty, male, 176410	DISORDERS/DIFFERENCES OF SEX DEVELOPMENT (DSD) / PRIMARY ADRENAL INSUFFICIENCY PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹ HEREDITARY CANCER PANEL UNSUCCESSFUL IVF TREATMENTS, RECURRENT MOLAR PREGNANCIES AND/OR RECURRENT PREGNANCY LOSS PANEL ¹
LHFPL5	100%	100%	100%	99%	Deafness, autosomal recessive 67, 610265	HEARING IMPAIRMENT PANEL (INCLUDING GJB2) MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹

LHX1	100%	100%	99.8%	94.8%		DISORDERS/DIFFERENCES OF SEX DEVELOPMENT (DSD) / PRIMARY ADRENAL INSUFFICIENCY PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS
LHX2	100%	100%	100%	97.4%		INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS

LHX3	100%	100%	100%	99.3%	Pituitary hormone deficiency, combined, 3, 221750	DISORDERS/DIFFERENCES OF SEX DEVELOPMENT (DSD) / PRIMARY ADRENAL INSUFFICIENCY PANEL HYPOGONADOTROPIC HYPOGONADISM (KALLMANN) PANEL SHORT STATURE/SKELETAL DYSPLASIA PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹
LHX4	100%	100%	100%	99.5%	Pituitary hormone deficiency, combined, 4, 262700	SHORT STATURE/SKELETAL DYSPLASIA PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS

LIAS	100%	100%	100%	99.2%	Hyperglycinemia, lactic acidosis, and seizures, 614462	EPILEPSY PANEL METABOLIC DISORDERS PANEL INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS MITOCHONDRIAL DISORDERS PANEL COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹
LIFR	100%	100%	100%	99.5%	Stuve-Wiedemann syndrome/Schwartz-Jampel type 2 syndrome, 601559	NEUROLOGICAL PAIN DISORDERS PANEL ¹ SHORT STATURE/SKELETAL DYSPLASIA PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹

LIG1	100%	100%	100%	97.8%	Immunodeficiency 96, 619774	PRIMARY IMMUNODEFICIENCIES PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL¹
LIG3	100%	100%	100%	99.1%	Mitochondrial DNA depletion syndrome 20 (MNGIE type), 619780	LIVER DISORDERS PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS MITOCHONDRIAL DISORDERS PANEL COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL¹

LIG4	100%	100%	100%	99.8%	LIG4 syndrome, 606593;{Multiple myeloma, resistance to}, 254500	MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹ SEVERE COMBINED IMMUNODEFICIENCY (SCID) PANEL HEREDITARY CANCER PANEL INHERITED BONE MARROW FAILURE AND/OR PREDISPOSITION PANEL TO HEMATOLOGICAL MALIGNANCIES DYSKERATOSIS CONGENITA AND APLASTIC ANEMIA PANEL PRIMARY IMMUNODEFICIENCIES PANEL INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS
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LIM2	100%	100%	100%	96.2%	Cataract 19, multiple types, 615277	VISION DISORDERS PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹
LIMS2	100%	100%	100%	98.5%	?Muscular dystrophy, autosomal recessive, with cardiomyopathy and triangular tongue, 616827	HEART DISORDERS PANEL ¹ MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹
LINGO1	100%	100%	100%	96.6%	Intellectual developmental disorder, autosomal recessive 64, 618103	INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹

LINS1	100%	100%	100%	99.9%	Intellectual developmental disorder, autosomal recessive 27, 614340	INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹
LIPA	95.1%	95.1%	100%	99.7%	Wolman disease, 620151;Cholesteryl ester storage disease, 278000	COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹ DISORDERS/DIFFEREN CES OF SEX DEVELOPMENT (DSD) / PRIMARY ADRENAL INSUFFICIENCY PANEL METABOLIC DISORDERS PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS
LIPC	100%	100%	100%	99.2%	{Diabetes mellitus, noninsulin-dependent}, 125853;Hepatic lipase deficiency, 614025;[High density lipoprotein cholesterol level QTL 12], 612797	METABOLIC DISORDERS PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS

LIPE	100%	100%	100%	98.8%	Lipodystrophy, familial partial, type 6, 615980	METABOLIC DISORDERS PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹
LIPH	100%	100%	100%	99.2%	Hypotrichosis 7, 604379;Woolly hair, autosomal recessive 2 with or without hypotrichosis, 604379	SKIN DISORDERS PANEL ¹ MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹
LIPN	100%	100%	100%	99.6%	Ichthyosis, congenital, autosomal recessive 8, 613943	SKIN DISORDERS PANEL ¹ MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹

LIPT1	100%	100%	100%	99.8%	Lipoyltransferase 1 deficiency, 616299	METABOLIC DISORDERS PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS MITOCHONDRIAL DISORDERS PANEL COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹
LIPT2	100%	100%	100%	98.5%	Encephalopathy, neonatal severe, with lactic acidosis and brain abnormalities, 617668	EPILEPSY PANEL METABOLIC DISORDERS PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS MITOCHONDRIAL DISORDERS PANEL COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹
LITAF	100%	100%	100%	98.5%	Charcot-Marie-Tooth disease, type 1C, 601098	POLYNEUROPATHIES PANEL ¹ MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS

LMAN1	100%	100%	100%	99.1%	Combined factor V and VIII deficiency, 227300	HEMOSTATIC/THROMBOTIC DISORDERS PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹
LMAN2L	100%	100%	100%	98.5%	?Intellectual developmental disorder, autosomal dominant 69, 617863;?Intellectual developmental disorder, autosomal recessive 52, 616887	COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹ MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS

LMBR1	100%	100%	100%	99.4%	Syndactyly, type IV, 186200;Laurin-Sandrow syndrome, 135750;Acheiropody, 200500;Triphalangeal thumb-polysyndactyly syndrome, 190605	SHORT STATURE/SKELETAL DYSPLASIA PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹
LMBRD1	100%	100%	100%	99.6%	Methylmalonic aciduria and homocystinuria, cblF type, 277380	COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹ SKIN DISORDERS PANEL ¹ METABOLIC DISORDERS PANEL INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS

LMBRD2	100%	100%	100%	99.7%	Developmental delay with variable neurologic and brain abnormalities, 619694	INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS
LMCD1	100%	100%	100%	98.4%		CONGENITAL HEARTDISEASE PANEL¹ HEART DISORDERS PANEL¹ MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS
LMF1	100%	100%	100%	99.4%	Lipase deficiency, combined, 246650	METABOLIC DISORDERS PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL¹

LMNA	100%	100%	100%	99.1%	Mandibuloacral dysplasia, 248370;Heart-hand syndrome, Slovenian type, 610140;Cardiomyopathy, dilated, 1A, 115200;Emery-Dreifuss muscular dystrophy 3, autosomal recessive, 616516;Restrictive dermopathy 2, 619793;Charcot-Marie-Tooth disease, type 2B1, 605588;Emery-Dreifuss muscular dystrophy 2, autosomal dominant, 181350;Hutchinson-Gilford progeria, 176670;Lipodystrophy, familial partial, type 2, 151660;Muscular dystrophy, congenital, 613205;Malouf syndrome, 212112	MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS SHORT STATURE/SKELETAL DYSPLASIA PANEL WITH GENOME WIDE CNV ANALYSIS METABOLIC DISORDERS PANEL HEART DISORDERS PANEL ¹ POLYNEUROPATHIES PANEL ¹ SKIN DISORDERS PANEL ¹ DILATED CARDIOMYOPATHY PANEL ¹ FETAL AKINESIA PANEL COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹ MUSCLE DISORDERS PANEL ARRHYTHMIA AND CARDIAC CONDUCTION DISORDERS PANEL ¹
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LMNB1	100%	100%	100%	99%	Leukodystrophy, adult-onset, autosomal dominant, 169500;Microcephaly 26, primary, autosomal dominant, 619179	MOVEMENT DISORDERS PANEL INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS
LMNB2	99.6%	98.8%	100%	96.6%	Microcephaly 27, primary, autosomal dominant, 619180;?Epilepsy, progressive myoclonic, 9, 616540;{Lipodystrophy, partial, acquired, susceptibility to}, 608709	METABOLIC DISORDERS PANEL INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹
LMOD1	100%	100%	100%	98.8%	?Megacystis-microcolon-intestinal hypoperistalsis syndrome 3, 619362	THORACIC AORTIC ANEURYSM AND/OR DISSECTION PANEL LIVER DISORDERS PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS

LMOD2	100%	100%	100%	98.7%	Cardiomyopathy, dilated, 2G, 619897	HEART DISORDERS PANEL ¹ MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹
LMOD3	100%	100%	99.9%	97.4%	Nemaline myopathy 10, 616165	FETAL AKINESIA PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹ MUSCLE DISORDERS PANEL
LMX1A	100%	100%	100%	99.4%	Deafness, autosomal dominant 7, 601412	HEARING IMPAIRMENT PANEL (INCLUDING GJB2) MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS

LMX1B	100%	100%	100%	97.1%	Focal segmental glomerulosclerosis 10, 256020;Nail-patella syndrome, 161200	VISION DISORDERS PANEL SKIN DISORDERS PANEL ¹ SHORT STATURE/SKELETAL DYSPLASIA PANEL WITH GENOME WIDE CNV ANALYSIS RENAL DISORDERS PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS
LNPK	93.1%	93.1%	100%	99.3%	Neurodevelopmental disorder with epilepsy and hypoplasia of the corpus callosum, 618090	INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹

LONP1	100%	100%	100%	99%	CODAS syndrome, 600373	VISION DISORDERS PANEL SKIN DISORDERS PANEL ¹ SHORT STATURE/SKELETAL DYSPLASIA PANEL WITH GENOME WIDE CNV ANALYSIS INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS MITOCHONDRIAL DISORDERS PANEL COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹
LORICRIN	100%	100%	100%	97.8%	Vohwinkel syndrome with ichthyosis, 604117	SKIN DISORDERS PANEL ¹ MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS

LOX	100%	100%	100%	98.9%	Aortic aneurysm, familial thoracic 10, 617168	THORACIC AORTIC ANEURYSM AND/OR DISSECTION PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS
LOXHD1	100%	100%	100%	98.9%	Deafness, autosomal recessive 77, 613079	HEARING IMPAIRMENT PANEL (INCLUDING GJB2) MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹
LOXL3	100%	100%	100%	99.3%	Myopia 28, autosomal recessive, 619781	VISION DISORDERS PANEL HEARING IMPAIRMENT PANEL (INCLUDING GJB2) MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS

LPAR6	100%	100%	100%	99.5%	Hypotrichosis 8, 278150;Woolly hair, autosomal recessive 1, with or without hypotrichosis, 278150	SKIN DISORDERS PANEL ¹ MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹
LPIN1	100%	100%	100%	99.4%	Myoglobinuria, acute recurrent, autosomal recessive, 268200	METABOLIC DISORDERS PANEL MUSCLE DISORDERS PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹

LPIN2	99.4%	99.2%	100%	99.3%	Majeed syndrome, 609628	INHERITED BONE MARROW FAILURE AND/OR PREDISPOSITION PANEL TO HEMATOLOGICAL MALIGNANCIES SHORT STATURE/SKELETAL DYSPLASIA PANEL WITH GENOME WIDE CNV ANALYSIS METABOLIC DISORDERS PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS SKIN DISORDERS PANEL ¹ IRON DISORDERS PANEL PRIMARY IMMUNODEFICIENCIES PANEL COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹
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LPL	100%	100%	100%	99.3%	Lipoprotein lipase deficiency, 238600;[High density lipoprotein cholesterol level QTL 11], 238600;Combined hyperlipidemia, familial, 144250	METABOLIC DISORDERS PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹
LPP	100%	100%	100%	99.1%	Lipoma;Leukemia, acute myeloid, 601626	MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS
LRAT	100%	100%	100%	98.2%	Leber congenital amaurosis 14, 613341;Retinal dystrophy, early-onset severe, 613341;Retinitis pigmentosa, juvenile, 613341	VISION DISORDERS PANEL METABOLIC DISORDERS PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹

LRBA	99.8%	99.8%	100%	99.5%	Immunodeficiency, common variable, 8, with autoimmunity, 614700	PRIMARY IMMUNODEFICIENCIES PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹
LRIF1	100%	100%	100%	99.6%	?Facioscapulohumeral muscular dystrophy 3, digenic, 619477	MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS
LRIG2	100%	100%	100%	99.3%	Urofacial syndrome 2, 615112	RENAL DISORDERS PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹

LRIT3	100%	100%	100%	99.7%	Night blindness, congenital stationary (complete), 1F, autosomal recessive, 615058	VISION DISORDERS PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹
LRMDA	97.8%	97.8%	100%	98.6%	Albinism, oculocutaneous, type VII, 615179	VISION DISORDERS PANEL SKIN DISORDERS PANEL ¹ MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹
LRP1	100%	100%	100%	99.2%	?Keratosis pilaris atrophicans, 604093;Developmental dysplasia of the hip 3, 620690	MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹

LRP12	100%	100%	100%	99.5%	Oculopharyngodistal myopathy 1, 164310;Amyotrophic lateral sclerosis 28, 620452	MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS
LRP2	100%	100%	100%	99.4%	Donnai-Barrow syndrome, 222448	VISION DISORDERS PANEL CRANIOFACIAL ANOMALIES PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹ RENAL DISORDERS PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS HEARING IMPAIRMENT PANEL (INCLUDING GJB2) INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS

LRP4	100%	100%	100%	99.1%	?Myasthenic syndrome, congenital, 17, 616304;Sclerosteosis 2, 614305;Cenani-Lenz syndactyly syndrome, 212780	SHORT STATURE/SKELETAL DYSPLASIA PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL¹ RENAL DISORDERS PANEL
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LRP5	100%	99.7%	100%	98.1%	Osteopetrosis, autosomal dominant 1, 607634;[Bone mineral density variability 1], 601884;Polycystic liver disease 4 with or without kidney cysts, 617875;Endosteal hyperostosis, 144750;Osteoporosis-pseudoglioma syndrome, 259770;Exudative vitreoretinopathy 4, 601813	VISION DISORDERS PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹ RENAL DISORDERS PANEL LIVER DISORDERS PANEL SHORT STATURE/SKELETAL DYSPLASIA PANEL WITH GENOME WIDE CNV ANALYSIS HEARING IMPAIRMENT PANEL (INCLUDING GJB2)
LRP6	100%	100%	100%	99.2%	{Coronary artery disease, autosomal dominant, 2}, 610947;Tooth agenesis, selective, 7, 616724	CRANIOFACIAL ANOMALIES PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS

LRPAP1	100%	100%	100%	98.7%	Myopia 23, autosomal recessive, 615431	VISION DISORDERS PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹
LRPPRC	96.8%	96.6%	100%	99.6%	Mitochondrial complex IV deficiency, nuclear type 5, (French-Canadian), 220111	INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS MITOCHONDRIAL DISORDERS PANEL COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹
LRRC10	100%	100%	100%	99.2%		HEART DISORDERS PANEL ¹ MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS

LRRC23	100%	100%	100%	98.1%	Spermatogenic failure 92, 620848	MALE INFERTILITY PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS
LRRC32	100%	100%	100%	98.9%	Cleft palate, proliferative retinopathy, and developmental delay, 619074	PRIMARY IMMUNODEFICIENCIES PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS
LRRC56	100%	100%	100%	99.4%	Ciliary dyskinesia, primary, 39, 618254	CILIOPATHIES PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹
LRRC7	100%	100%	100%	99.6%		INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS

LRRC8A	100%	100%	100%	98.7%	?Agammaglobulinemia 5, 613506	PRIMARY IMMUNODEFICIENCIES PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS
LRRK1	100%	100%	100%	99.4%	Osteosclerotic metaphyseal dysplasia, 615198	SHORT STATURE/SKELETAL DYSPLASIA PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹
LRRK2	100%	100%	100%	99%	{Parkinson disease 8}, 607060	MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS PARKINSON DISEASE PANEL

LRSAM1	100%	100%	100%	98.8%	Charcot-Marie-Tooth disease, axonal, type 2P, 614436	POLYNEUROPATHIES PANEL ¹ MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹
LRTOMT	100%	99.9%	100%	99.3%	Deafness, autosomal recessive 63, 611451	HEARING IMPAIRMENT PANEL (INCLUDING GJB2) MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹
LSM1	100%	100%	100%	99.2%		INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹

LSM11	100%	100%	100%	98.1%	?Aicardi-Goutieres syndrome 8, 619486	PRIMARY IMMUNODEFICIENCIES PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS
LSS	100%	100%	100%	99.2%	Hypotrichosis 14, 618275;Cataract 44, 616509;Alopecia-intellectual disability syndrome 4, 618840	VISION DISORDERS PANEL SKIN DISORDERS PANEL ¹ INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹

LTBP1	100%	100%	100%	99.2%	Cutis laxa, autosomal recessive, type IIE, 619451	SHORT STATURE/SKELETAL DYSPLASIA PANEL WITH GENOME WIDE CNV ANALYSIS INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹
LTBP2	100%	100%	100%	99.4%	Glaucoma 3, primary congenital, D, 613086;Microspherophakia and/or megalocornea, with ectopia lentis and with or without secondary glaucoma, 251750;?Weill-Marchesani syndrome 3, recessive, 614819	VISION DISORDERS PANEL SHORT STATURE/SKELETAL DYSPLASIA PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹

LTBP3	100%	100%	100%	97.5%	Dental anomalies and short stature, 601216;Geleophysic dysplasia 3, 617809	THORACIC AORTIC ANEURYSM AND/OR DISSECTION PANEL CRANIOFACIAL ANOMALIES PANEL WITH GENOME WIDE CNV ANALYSIS SKIN DISORDERS PANEL ¹ SHORT STATURE/SKELETAL DYSPLASIA PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹
LTBP4	100%	100%	100%	98.2%	Cutis laxa, autosomal recessive, type IC, 613177	SKIN DISORDERS PANEL ¹ MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹

LTC4S	100%	100%	100%	99.3%	Leukotriene C4 synthase deficiency, 614037	METABOLIC DISORDERS PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹
LTV1	100%	100%	100%	99.6%	Inflammatory poikiloderma with hair abnormalities and acral keratoses, 620199	SKIN DISORDERS PANEL ¹ MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS
LYN	100%	100%	100%	99.2%	Autoinflammatory disease, systemic, with vasculitis, 620376	PRIMARY IMMUNODEFICIENCIES PANEL LIVER DISORDERS PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS
LYRM4	68%	68%	100%	99.3%	?Combined oxidative phosphorylation deficiency 19, 615595	MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS MITOCHONDRIAL DISORDERS PANEL

LYRM7	100%	100%	100%	99.8%	Mitochondrial complex III deficiency, nuclear type 8, 615838	INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS MITOCHONDRIAL DISORDERS PANEL COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹
LYSET	100%	100%	100%	99.9%	Dysostosis multiplex, Ain-Naz type, 619345	SHORT STATURE/SKELETAL DYSPLASIA PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS

LYST	99.5%	99.5%	100%	99.5%	Chediak-Higashi syndrome, 214500	VISION DISORDERS PANEL SKIN DISORDERS PANEL ¹ HEMOSTATIC/THROMB OTIC DISORDERS PANEL PRIMARY IMMUNODEFICIENCIES PANEL METABOLIC DISORDERS PANEL INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹
LYZ	100%	100%	100%	99.7%	Amyloidosis, hereditary systemic 5, 620658	SKIN DISORDERS PANEL ¹ RENAL DISORDERS PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS

LZTFL1	100%	100%	100%	99.7%	Bardet-Biedl syndrome 17, 615994	VISION DISORDERS PANEL CILIOPATHIES PANEL HYPOGONADOTROPIC HYPOGONADISM (KALLMANN) PANEL INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS RENAL DISORDERS PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL¹
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LZTR1	100%	100%	100%	99.2%	Noonan syndrome 2, 605275;Noonan syndrome 10, 616564;{Schwannomatosis-2, susceptibility to}, 615670	CONGENITAL HEARTDISEASE PANEL¹ HEART DISORDERS PANEL¹ HEMOSTATIC/THROMB OTIC DISORDERS PANEL NOONAN SYNDROME / RASOPATHY PANEL HEREDITARY CANCER PANEL INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL¹ NEUROLOGICAL PAIN DISORDERS PANEL¹ SHORT STATURE/SKELETAL DYSPLASIA PANEL WITH GENOME WIDE CNV ANALYSIS LYMPHATIC ANOMALIES PANEL WITH GENOME WIDE CNV ANALYSIS
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LZTS1	100%	100%	100%	99.1%	Esophageal squamous cell carcinoma, somatic, 133239	MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS
M1AP	100%	100%	100%	99.1%	Spermatogenic failure 48, 619108	MALE INFERTILITY PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS
MAB21L1	100%	100%	100%	99.2%	Cerebellar, ocular, craniofacial, and genital syndrome, 618479	COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹ INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS

MAB21L2	100%	100%	100%	98.8%	Microphthalmia/coloboma and skeletal dysplasia syndrome, 615877	VISION DISORDERS PANEL SHORT STATURE/SKELETAL DYSPLASIA PANEL WITH GENOME WIDE CNV ANALYSIS INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL¹
MACF1	100%	100%	100%	99.4%	Lissencephaly 9 with complex brainstem malformation, 618325	INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS
MAD1L1	100%	100%	100%	98.4%	Prostate cancer, somatic, 176807;Mosaic variegated aneuploidy syndrome 7 with inflammation and tumor predisposition, 620189;Lymphoma, B-cell, somatic	MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS

MAD2L2	100%	100%	100%	99.2%	?Fanconi anemia, complementation group V, 617243	MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS HEREDITARY CANCER PANEL INHERITED BONE MARROW FAILURE AND/OR PREDISPOSITION PANEL TO HEMATOLOGICAL MALIGNANCIES SHORT STATURE/SKELETAL DYSPLASIA PANEL WITH GENOME WIDE CNV ANALYSIS
MADD	100%	100%	100%	99.3%	Neurodevelopmental disorder with dysmorphic facies, impaired speech and hypotonia, 619005;DEEAH syndrome, 619004	INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹

MAF	90.6%	85.7%	100%	94.6%	Cataract 21, multiple types, 610202;Ayme-Gripp syndrome, 601088	MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS VISION DISORDERS PANEL INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS
MAFA	99.8%	98.3%	99.8%	88.4%	Insulinomatosis and diabetes mellitus, 147630	MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS
MAFB	100%	100%	100%	98.5%	Duane retraction syndrome 3, 617041;Multicentric carpotarsal osteolysis syndrome, 166300	VISION DISORDERS PANEL SHORT STATURE/SKELETAL DYSPLASIA PANEL WITH GENOME WIDE CNV ANALYSIS RENAL DISORDERS PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS

MAG	100%	100%	100%	98.3%	Spastic paraplegia 75, autosomal recessive, 616680	MOVEMENT DISORDERS PANEL POLYNEUROPATHIES PANEL ¹ INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹
MAGED2	100%	99.4%	99%	67.6%	Bartter syndrome, type 5, antenatal, transient, 300971	RENAL DISORDERS PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS

MAGEL2	100%	100%	100%	98.7%	Schaaf-Yang syndrome, 615547	FETAL AKINESIA PANEL LIVER DISORDERS PANEL INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS
MAGI2	97.4%	96.2%	100%	97.1%	Nephrotic syndrome, type 15, 617609	RENAL DISORDERS PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹
MAGT1	93.8%	93.7%	98.1%	67.6%	Immunodeficiency, X-linked, with magnesium defect, Epstein-Barr virus infection and neoplasia, 300853;Congenital disorder of glycosylation, type Icc, 301031	PRIMARY IMMUNODEFICIENCIES PANEL METABOLIC DISORDERS PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS

MAK	100%	100%	100%	99.6%	Retinitis pigmentosa 62, 614181	VISION DISORDERS PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹
MAL	100%	100%	100%	97.9%	?Leukodystrophy, hypomyelinating, 28, 620978	MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS
MALT1	100%	100%	100%	99.3%	Immunodeficiency 12, 615468	PRIMARY IMMUNODEFICIENCIES PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹
MAML2	100%	100%	100%	99.5%	Mucoepidermoid salivary gland carcinoma	MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS

MAMLD1	100%	99.9%	98.8%	67.5%	Hypospadias 2, X-linked, 300758	DISORDERS/DIFFERENCES OF SEX DEVELOPMENT (DSD) / PRIMARY ADRENAL INSUFFICIENCY PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS
MAN1B1	100%	99.6%	100%	99.2%	Rafiq syndrome, 614202	METABOLIC DISORDERS PANEL INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL¹

MAN2B1	100%	100%	100%	97.6%	Mannosidosis, alpha-, types I and II, 248500	HEARING IMPAIRMENT PANEL (INCLUDING GJB2) COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹ PRIMARY IMMUNODEFICIENCIES PANEL SHORT STATURE/SKELETAL DYSPLASIA PANEL WITH GENOME WIDE CNV ANALYSIS METABOLIC DISORDERS PANEL INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS
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MAN2B2	100%	100%	100%	99.3%		PRIMARY IMMUNODEFICIENCIES PANEL METABOLIC DISORDERS PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹
MAN2C1	100%	100%	100%	99.3%	Congenital disorder of deglycosylation 2, 619775	METABOLIC DISORDERS PANEL INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹

MANBA	100%	100%	100%	99.2%	Mannosidosis, beta, 248510	PRIMARY IMMUNODEFICIENCIES PANEL SHORT STATURE/SKELETAL DYSPLASIA PANEL WITH GENOME WIDE CNV ANALYSIS METABOLIC DISORDERS PANEL INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹
MANF	100%	100%	100%	98.7%	Diabetes, deafness, developmental delay, and short stature syndrome, 620651	MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS

MAOA	100%	99.7%	98.9%	69.7%	Brunner syndrome, 300615	METABOLIC DISORDERS PANEL INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS
MAP1B	100%	100%	100%	99.4%	?Deafness, autosomal dominant 83, 619808;Periventricular nodular heterotopia 9, 618918	HEARING IMPAIRMENT PANEL (INCLUDING GJB2) INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS
MAP1LC3B2	100%	100%	100%	98.6%		PRIMARY IMMUNODEFICIENCIES PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS

MAP2K1	95.8%	95.8%	100%	99.2%	Cardiofaciocutaneous syndrome 3, 615279;Melorheostosis, isolated, somatic mosaic, 155950	SKIN DISORDERS PANEL ¹ SHORT STATURE/SKELETAL DYSPLASIA PANEL WITH GENOME WIDE CNV ANALYSIS LYMPHATIC ANOMALIES PANEL WITH GENOME WIDE CNV ANALYSIS INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS NOONAN SYNDROME / RASOPATHY PANEL HEREDITARY CANCER PANEL
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MAP2K2	100%	100%	100%	98.4%	Cardiofaciocutaneous syndrome 4, 615280	NOONAN SYNDROME / RASOPATHY PANEL HEREDITARY CANCER PANEL SKIN DISORDERS PANEL ¹ SHORT STATURE/SKELETAL DYSPLASIA PANEL WITH GENOME WIDE CNV ANALYSIS LYMPHATIC ANOMALIES PANEL WITH GENOME WIDE CNV ANALYSIS INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS
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MAP3K1	100%	100%	100%	99.1%	46XY sex reversal 6, 613762	HEARING IMPAIRMENT PANEL (INCLUDING GJB2) DISORDERS/DIFFEREN CES OF SEX DEVELOPMENT (DSD) / PRIMARY ADRENAL INSUFFICIENCY PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS
MAP3K14	100%	100%	100%	99.1%	Immunodeficiency 112, 620449	PRIMARY IMMUNODEFICIENCIES PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS

MAP3K20	100%	100%	100%	99.4%	Centronuclear myopathy 6 with fiber-type disproportion, 617760;Split-foot malformation with mesoaxial polydactyly, 616890	SHORT STATURE/SKELETAL DYSPLASIA PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹ MUSCLE DISORDERS PANEL
MAP3K7	100%	100%	100%	99.4%	Frontometaphyseal dysplasia 2, 617137;Cardiospondylocarpofacial syndrome, 157800	SHORT STATURE/SKELETAL DYSPLASIA PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS OROFACIAL CLEFTING PANEL WITH GENOME WIDE CNV ANALYSIS
MAP3K8	100%	100%	100%	99.5%	Lung cancer, somatic, 211980	MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS

MAP4K4	100%	99.8%	100%	99.2%		INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS
MAPK1	100%	100%	100%	99.4%	Noonan syndrome 13, 619087	SHORT STATURE/SKELETAL DYSPLASIA PANEL WITH GENOME WIDE CNV ANALYSIS LYMPHATIC ANOMALIES PANEL WITH GENOME WIDE CNV ANALYSIS INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS NOONAN SYNDROME / RASOPATHY PANEL

MAPK8	100%	100%	100%	99.4%		MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS PRIMARY IMMUNODEFICIENCIES PANEL
MAPK8IP3	100%	100%	100%	99.2%	Neurodevelopmental disorder with or without variable brain abnormalities, 618443	MOVEMENT DISORDERS PANEL EPILEPSY PANEL INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS
MAPKAPK3	100%	100%	100%	98.8%	?Macular dystrophy, patterned, 3, 617111	VISION DISORDERS PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS

MAPKAPK5	100%	100%	100%	99.3%	Neurocardiofaciodigital syndrome, 619869	SHORT STATURE/SKELETAL DYSPLASIA PANEL WITH GENOME WIDE CNV ANALYSIS INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL¹
MAPKBP1	100%	100%	100%	99%	Nephronophthisis 20, 617271	CILIOPATHIES PANEL RENAL DISORDERS PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL¹

MAPRE2	100%	100%	100%	99%	Symmetric circumferential skin creases, congenital, 2, 616734	MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS OROFACIAL CLEFTING PANEL WITH GENOME WIDE CNV ANALYSIS
MAPT	95.4%	95.3%	100%	98.5%	Supranuclear palsy, progressive, 601104;Frontotemporal dementia 1, with or without parkinsonism, 600274;Supranuclear palsy, progressive atypical, 260540;{Parkinson disease, susceptibility to}, 168600;Pick disease, 172700	AMYOTROPHIC LATERAL SCLEROSIS, ALS PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS MITOCHONDRIAL DISORDERS PANEL PARKINSON DISEASE PANEL COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹
MARCHF6	100%	100%	100%	99.3%	Epilepsy, familial adult myoclonic, 3, 613608	MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS

MARK2	100%	100%	100%	98.6%		INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS
MARK3	100%	99.9%	100%	99.3%	?Visual impairment and progressive phthisis bulbi, 618283	MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS
MARS1	100%	100%	100%	99.1%	Spastic paraplegia 70, autosomal recessive, 620323;Interstitial lung and liver disease, 615486;?Trichothiodystrophy 9, nonphotosensitive, 619692;Charcot-Marie-Tooth disease, axonal, type 2U, 616280	SKIN DISORDERS PANEL ¹ POLYNEUROPATHIES PANEL ¹ LIVER DISORDERS PANEL INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹

MARS2	100%	100%	100%	99.8%	?Combined oxidative phosphorylation deficiency 25, 616430;Spastic ataxia 3, autosomal recessive, 611390	MOVEMENT DISORDERS PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS MITOCHONDRIAL DISORDERS PANEL COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹
MARVELD2	100%	100%	100%	99.6%	Deafness, autosomal recessive 49, 610153	HEARING IMPAIRMENT PANEL (INCLUDING GJB2) MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹

MASP1	100%	100%	100%	99.1%	3MC syndrome 1, 257920	CRANIOFACIAL ANOMALIES PANEL WITH GENOME WIDE CNV ANALYSIS SHORT STATURE/SKELETAL DYSPLASIA PANEL WITH GENOME WIDE CNV ANALYSIS INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL¹ OROFACIAL CLEFTING PANEL WITH GENOME WIDE CNV ANALYSIS
MASP2	100%	100%	100%	99.4%	MASP2 deficiency, 613791	PRIMARY IMMUNODEFICIENCIES PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS

MAST1	100%	100%	100%	98.8%	Mega-corpus-callosum syndrome with cerebellar hypoplasia and cortical malformations, 618273	INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS
MAST3	100%	100%	100%	97.6%	Developmental and epileptic encephalopathy 108, 620115	EPILEPSY PANEL INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS
MAST4	100%	100%	100%	99.2%		INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS

MASTL	100%	100%	100%	99.6%		HEMOSTATIC/THROMBOTIC DISORDERS PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS
MAT1A	100%	100%	100%	99.4%	Hypermethioninemia, persistent, autosomal dominant, due to methionine adenosyltransferase I/III deficiency, 250850;Methionine adenosyltransferase deficiency, autosomal recessive, 250850	METABOLIC DISORDERS PANEL INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹
MAT2A	100%	100%	100%	99.6%		THORACIC AORTIC ANEURYSM AND/OR DISSECTION PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS

MATN3	100%	100%	100%	99.2%	{Osteoarthritis susceptibility 2}, 140600;Spondyloepimetaphyseal dysplasia, Borochowitz-Cormier-Daire type, 608728;Epiphyseal dysplasia, multiple, 5, 607078	SHORT STATURE/SKELETAL DYSPLASIA PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹
MATR3	100%	100%	100%	99.6%	Amyotrophic lateral sclerosis 21, 606070	AMYOTROPHIC LATERAL SCLEROSIS, ALS PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS
MAX	100%	100%	100%	98.8%	Polydactyly-macrocephaly syndrome, 620712;{Pheochromocytoma, susceptibility to}, 171300	INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS HEREDITARY CANCER PANEL

MB	100%	100%	100%	97.3%	Myopathy, sarcoplasmic body, 620286	MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS MUSCLE DISORDERS PANEL
MBD4	100%	100%	100%	99.2%	{Uveal melanoma, susceptibility to, 1}, 606660;Tumor predisposition syndrome 2, 619975	INHERITED BONE MARROW FAILURE AND/OR PREDISPOSITION PANEL TO HEMATOLOGICAL MALIGNANCIES PANEL HEREDITARY COLORECTAL AND POLYPOSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS HEREDITARY CANCER PANEL
MBD5	100%	100%	100%	99.4%	Intellectual developmental disorder, autosomal dominant 1, 156200	EPILEPSY PANEL INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS

MBIP	100%	100%	100%	99.9%		MOVEMENT DISORDERS PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS
MBOAT7	100%	100%	100%	98.6%	Intellectual developmental disorder, autosomal recessive 57, 617188	EPILEPSY PANEL METABOLIC DISORDERS PANEL INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL¹

MBTPS1	99.8%	99.3%	100%	99.4%	?Spondyloepiphyseal dysplasia, Kondo-Fu type, 618392	SHORT STATURE/SKELETAL DYSPLASIA PANEL WITH GENOME WIDE CNV ANALYSIS METABOLIC DISORDERS PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹
MBTPS2	100%	99.9%	99%	69.7%	Keratosis follicularis spinulosa decalvans, X-linked, 308800;Osteogenesis imperfecta, type XIX, 301014;IFAP syndrome with or without BRESHECK syndrome, 308205;?Olmsted syndrome, X-linked, 300918	SKIN DISORDERS PANEL ¹ SHORT STATURE/SKELETAL DYSPLASIA PANEL WITH GENOME WIDE CNV ANALYSIS INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS OROFACIAL CLEFTING PANEL WITH GENOME WIDE CNV ANALYSIS

MC2R	100%	100%	100%	99.3%	Glucocorticoid deficiency, due to ACTH unresponsiveness, 202200	DISORDERS/DIFFERENCES OF SEX DEVELOPMENT (DSD) / PRIMARY ADRENAL INSUFFICIENCY PANEL PRIMARY IMMUNODEFICIENCIES PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹
MC4R	100%	100%	100%	99.4%	Obesity (BMIQ20), 618406;{Obesity, resistance to (BMIQ20)}, 618406	MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS
MCAT	100%	100%	100%	99.5%	Optic atrophy 15, 620583	VISION DISORDERS PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS MITOCHONDRIAL DISORDERS PANEL COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹

MCC	100%	100%	100%	99%	Colorectal cancer, somatic, 114500	MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS
MCCC1	100%	100%	100%	99.7%	3-Methylcrotonyl-CoA carboxylase 1 deficiency, 210200	METABOLIC DISORDERS PANEL INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹
MCCC2	93.4%	93.4%	100%	99.4%	3-Methylcrotonyl-CoA carboxylase 2 deficiency, 210210	METABOLIC DISORDERS PANEL INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹

MCEE	100%	100%	100%	99.1%	Methylmalonyl-CoA epimerase deficiency, 251120	METABOLIC DISORDERS PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹
MCFD2	100%	100%	100%	99.7%	Factor V and factor VIII, combined deficiency of, 613625	HEMOSTATIC/THROMB OTIC DISORDERS PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹
MCIDAS	100%	100%	100%	99.1%	Ciliary dyskinesia, primary, 42, 618695	CILIOPATHIES PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹

MCM10	100%	100%	100%	99.5%	Immunodeficiency 80 with or without cardiomyopathy, 619313	PRIMARY IMMUNODEFICIENCIES PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹
MCM2	100%	100%	100%	99.1%	?Deafness, autosomal dominant 70, 616968	HEARING IMPAIRMENT PANEL (INCLUDING GJB2) MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS
MCM3AP	100%	100%	100%	99.3%	Peripheral neuropathy, autosomal recessive, with or without impaired intellectual development, 618124	POLYNEUROPATHIES PANEL ¹ INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹

MCM4	95.3%	95.3%	100%	99.3%	Immunodeficiency 54, 609981	INHERITED BONE MARROW FAILURE AND/OR PREDISPOSITION PANEL TO HEMATOLOGICAL MALIGNANCIES PRIMARY IMMUNODEFICIENCIES PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹
MCM5	100%	100%	100%	99%	?Meier-Gorlin syndrome 8, 617564	SHORT STATURE/SKELETAL DYSPLASIA PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹

MCM6	100%	100%	100%	99.5%	Lactase persistence/nonpersistence, 223100	INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS
MCM8	94.4%	94.4%	100%	99.5%	?Premature ovarian failure 10, 612885	PANEL HEREDITARY COLORECTAL AND POLYPOSIS DISORDERS/DIFFEREN CES OF SEX DEVELOPMENT (DSD) / PRIMARY ADRENAL INSUFFICIENCY PANEL MALE INFERTILITY PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS PREMATURE OVARIAN INSUFFICIENCY PANEL HEREDITARY CANCER PANEL

MCM9	100%	100%	100%	99.2%	Ovarian dysgenesis 4, 616185	PANEL HEREDITARY COLORECTAL AND POLYPOSIS DISORDERS/DIFFEREN CES OF SEX DEVELOPMENT (DSD) / PRIMARY ADRENAL INSUFFICIENCY PANEL MALE INFERTILITY PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL¹ PREMATURE OVARIAN INSUFFICIENCY PANEL HEREDITARY CANCER PANEL
MCMD2	100%	100%	100%	99.5%		MALE INFERTILITY PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS

MCOLN1	100%	99.9%	100%	99%	Lisch epithelial corneal dystrophy, 620763;Mucopolidosis IV, 252650	VISION DISORDERS PANEL METABOLIC DISORDERS PANEL INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL¹
MCPH1	94.6%	92.7%	100%	99.5%	Microcephaly 1, primary, autosomal recessive, 251200	INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL¹

MCTS1	100%	100%	99%	71.2%	Immunodeficiency 118, mycobacteriosis, 301115	PRIMARY IMMUNODEFICIENCIES PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS
MCUR1	100%	100%	100%	99.3%		MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS MITOCHONDRIAL DISORDERS PANEL
MDFIC	100%	100%	100%	98.9%	Lymphatic malformation 12, 620014	SKIN DISORDERS PANEL ¹ LYMPHATIC ANOMALIES PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹

MDH1	100%	100%	100%	99.8%	?Developmental and epileptic encephalopathy 88, 618959	METABOLIC DISORDERS PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS MITOCHONDRIAL DISORDERS PANEL
MDH2	100%	100%	100%	98.6%	Developmental and epileptic encephalopathy 51, 617339	EPILEPSY PANEL INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MITOCHONDRIAL DISORDERS PANEL COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹ HEREDITARY CANCER PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS
MDM2	94%	94%	100%	99.3%	{Accelerated tumor formation, susceptibility to}, 614401;?Lessel-Kubisch syndrome, 618681	MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS

MDM4	100%	100%	100%	99.1%	?Bone marrow failure syndrome 6, 618849	INHERITED BONE MARROW FAILURE AND/OR PREDISPOSITION PANEL TO HEMATOLOGICAL MALIGNANCIES MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS
ME2	94.1%	94.1%	100%	99.5%		METABOLIC DISORDERS PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS

MECOM	100%	100%	100%	99.4%	Radioulnar synostosis with amegakaryocytic thrombocytopenia 2, 616738	INHERITED BONE MARROW FAILURE AND/OR PREDISPOSITION PANEL TO HEMATOLOGICAL MALIGNANCIES HEMOSTATIC/THROMBOTIC DISORDERS PANEL SHORT STATURE/SKELETAL DYSPLASIA PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS
MECP2	100%	99.3%	97.7%	64.9%	Rett syndrome, atypical, 312750;Encephalopathy, neonatal severe, 300673;Intellectual developmental disorder, X-linked syndromic, Lubs type, 300260;{Autism susceptibility, X-linked 3}, 300496;Intellectual developmental disorder, X-linked syndromic 13, 300055;Rett syndrome, 312750;Rett syndrome, preserved speech variant, 312750	MOVEMENT DISORDERS PANEL EPILEPSY PANEL INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS

MECR	100%	100%	100%	99.2%	Dystonia, childhood-onset, with optic atrophy and basal ganglia abnormalities, 617282;Optic atrophy 16, 620629	MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS MITOCHONDRIAL DISORDERS PANEL COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹ MOVEMENT DISORDERS PANEL VISION DISORDERS PANEL INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS
MED11	100%	100%	100%	98.8%	Neurodegeneration with developmental delay, early respiratory failure, myoclonic seizures, and brain abnormalities, 620327	INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS

MED12	100%	99.7%	98.4%	64.8%	Lujan-Fryns syndrome, 309520;Ohdo syndrome, X-linked, 300895;Hardikar syndrome, 301068;Opitz-Kaveggia syndrome, 305450	THORACIC AORTIC ANEURYSM AND/OR DISSECTION PANEL INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS LIVER DISORDERS PANEL EPILEPSY PANEL TALL STATURE PANEL WITH GENOME WIDE CNV ANALYSIS SKIN DISORDERS PANEL ¹ CRANIOFACIAL ANOMALIES PANEL WITH GENOME WIDE CNV ANALYSIS
MED12L	100%	100%	100%	99.6%	Nizon-Isidor syndrome, 618872	INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS

MED13	100%	100%	100%	99.4%	Intellectual developmental disorder, autosomal dominant 61, 618009	INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS
MED13L	100%	100%	100%	99.4%	Impaired intellectual development and distinctive facial features with or without cardiac defects, 616789	CONGENITAL HEARTDISEASE PANEL¹ HEART DISORDERS PANEL¹ INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS
MED16	100%	100%	99.9%	94.7%		INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS

MED17	100%	100%	100%	99.4%	Microcephaly, postnatal progressive, with seizures and brain atrophy, 613668	EPILEPSY PANEL INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹
MED23	100%	100%	100%	99.4%	Intellectual developmental disorder, autosomal recessive 18, with or without epilepsy, 614249	INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹

MED25	100%	100%	100%	98.4%	Basel-Vanagait-Smirin-Yosef syndrome, 616449	INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹ OROFACIAL CLEFTING PANEL WITH GENOME WIDE CNV ANALYSIS
MED27	100%	100%	100%	99.5%	Neurodevelopmental disorder with spasticity, cataracts, and cerebellar hypoplasia, 619286	INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹

MEF2C	100%	100%	100%	99.7%	Chromosome 5q14.3 deletion syndrome, 613443;Neurodevelopmental disorder with hypotonia, stereotypic hand movements, and impaired language, 613443	EPILEPSY PANEL INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS
MEFV	96.1%	96.1%	100%	99.4%	Neutrophilic dermatosis, acute febrile, 608068;Familial Mediterranean fever, AR, 249100;Familial Mediterranean fever, AD, 134610	SKIN DISORDERS PANEL ¹ PRIMARY IMMUNODEFICIENCIES PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹
MEGF10	100%	100%	100%	99.2%	Congenital myopathy 10A, severe variant, 614399;Congenital myopathy 10B, mild variant, 620249	FETAL AKINESIA PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹ MUSCLE DISORDERS PANEL

MEGF8	100%	100%	99.9%	97.6%	Carpenter syndrome 2, 614976	CRANIOFACIAL ANOMALIES PANEL WITH GENOME WIDE CNV ANALYSIS SHORT STATURE/SKELETAL DYSPLASIA PANEL WITH GENOME WIDE CNV ANALYSIS INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹
MEI1	100%	100%	100%	99.3%	Hydatidiform mole, recurrent, 3, 618431	UNSUCCESSFUL IVF TREATMENTS, RECURRENT MOLAR PREGNANCIES AND/OR RECURRENT PREGNANCY LOSS PANEL ¹ MALE INFERTILITY PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS

MEIOB	100%	100%	100%	99.8%	Premature ovarian failure 23, 620686;Spermatogenic failure 22, 617706	MALE INFERTILITY PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS
MEIS2	91.5%	91.5%	100%	98.6%	Cleft palate, cardiac defects, and impaired intellectual development, 600987	CRANIOFACIAL ANOMALIES PANEL WITH GENOME WIDE CNV ANALYSIS CONGENITAL HEARTDISEASE PANEL¹ HEART DISORDERS PANEL¹ INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS OROFACIAL CLEFTING PANEL WITH GENOME WIDE CNV ANALYSIS
MEN1	100%	100%	100%	98.5%	Lipoma, somatic;Angiofibroma, somatic;Multiple endocrine neoplasia 1, 131100;Carcinoid tumor of lung;Adrenal adenoma, somatic;Parathyroid adenoma, somatic	MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS HEREDITARY CANCER PANEL

MEOX1	100%	99.9%	100%	98.6%	Klippel-Feil syndrome 2, 214300	CRANIOFACIAL ANOMALIES PANEL WITH GENOME WIDE CNV ANALYSIS SHORT STATURE/SKELETAL DYSPLASIA PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL¹
MERTK	100%	100%	100%	99.3%	Retinitis pigmentosa 38, 613862	VISION DISORDERS PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL¹

MESD	100%	100%	100%	97.3%	Osteogenesis imperfecta, type XX, 618644	SHORT STATURE/SKELETAL DYSPLASIA PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹
MESP1	100%	100%	100%	99.4%		CONGENITAL HEARTDISEASE PANEL ¹ HEART DISORDERS PANEL ¹ MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS
MESP2	100%	99.1%	100%	99.1%	Spondylocostal dysostosis 2, autosomal recessive, 608681	SHORT STATURE/SKELETAL DYSPLASIA PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹

MET	100%	100%	100%	99.5%	Renal cell carcinoma, papillary, 1, familial and somatic, 605074;?Arthrogryposis, distal, type 11, 620019;Hepatocellular carcinoma, childhood type, somatic, 114550;{Osteofibrous dysplasia, susceptibility to}, 607278;?Deafness, autosomal recessive 97, 616705	HEARING IMPAIRMENT PANEL (INCLUDING GJB2) SHORT STATURE/SKELETAL DYSPLASIA PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹ PANEL HEREDITARY RENALCANCER HEREDITARY CANCER PANEL
METTL13	100%	100%	100%	99.3%	{?Deafness, autosomal recessive 26, modifier of}, 605429	MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS

METTL23	100%	100%	100%	99.6%	Intellectual developmental disorder, autosomal recessive 44, 615942	INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹
METTL5	100%	100%	100%	99.9%	Intellectual developmental disorder, autosomal recessive 72, 618665	INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹
MFAP5	100%	100%	100%	98.9%	Aortic aneurysm, familial thoracic 9, 616166	THORACIC AORTIC ANEURYSM AND/OR DISSECTION PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS

MFF	95.9%	95.9%	100%	99.6%	Encephalopathy due to defective mitochondrial and peroxisomal fission 2, 617086	MOVEMENT DISORDERS PANEL EPILEPSY PANEL INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS MITOCHONDRIAL DISORDERS PANEL COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹
MFN2	100%	100%	100%	99.3%	Lipomatosis, multiple symmetric, with or without peripheral neuropathy, 151800;Charcot-Marie-Tooth disease, axonal, type 2A2A, 609260;Charcot-Marie-Tooth disease, axonal, type 2A2B, 617087;Hereditary motor and sensory neuropathy VIA, 601152	VISION DISORDERS PANEL POLYNEUROPATHIES PANEL ¹ MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS MITOCHONDRIAL DISORDERS PANEL COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹

MFRP	100%	99.9%	100%	98.9%	Microphthalmia, isolated 5, 611040;Nanophthalmos 2, 609549	VISION DISORDERS PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL¹
MFSD2A	100%	100%	100%	98.8%	Neurodevelopmental disorder with progressive microcephaly, spasticity, and brain abnormalities, 616486	METABOLIC DISORDERS PANEL INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL¹

MFSD8	100%	100%	100%	99.6%	Macular dystrophy with central cone involvement, 616170;Ceroid lipofuscinosis, neuronal, 7, 610951	MOVEMENT DISORDERS PANEL VISION DISORDERS PANEL EPILEPSY PANEL METABOLIC DISORDERS PANEL INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹
MGAT2	100%	100%	100%	99%	Congenital disorder of glycosylation, type IIa, 212066	METABOLIC DISORDERS PANEL INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹

MGME1	100%	100%	100%	99.4%	Mitochondrial DNA depletion syndrome 11, 615084	MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS MITOCHONDRIAL DISORDERS PANEL COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹
MGP	100%	100%	100%	99.3%	Keutel syndrome, 245150	SKIN DISORDERS PANEL ¹ HEARING IMPAIRMENT PANEL (INCLUDING GJB2) SHORT STATURE/SKELETAL DYSPLASIA PANEL WITH GENOME WIDE CNV ANALYSIS INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹

MIA3	100%	100%	100%	99.3%	?Ondotochondrodysplasia 2 with hearing loss and diabetes, 619269	HEARING IMPAIRMENT PANEL (INCLUDING GJB2) INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS
MIB1	100%	100%	100%	99.4%	Left ventricular noncompaction 7, 615092	HEART DISORDERS PANEL ¹ MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS
MICOS13	100%	100%	99.9%	97.9%	Combined oxidative phosphorylation deficiency 37, 618329	INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS MITOCHONDRIAL DISORDERS PANEL COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹

MICU1	100%	100%	100%	99.3%	Myopathy with extrapyramidal signs, 615673	MOVEMENT DISORDERS PANEL INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS MITOCHONDRIAL DISORDERS PANEL COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹ MUSCLE DISORDERS PANEL
MICU2	100%	100%	100%	99.6%		MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS MITOCHONDRIAL DISORDERS PANEL

MID1	100%	99.9%	98.7%	67.1%	Opitz GBBB syndrome, 300000	CRANIOFACIAL ANOMALIES PANEL WITH GENOME WIDE CNV ANALYSIS INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS OROFACIAL CLEFTING PANEL WITH GENOME WIDE CNV ANALYSIS
MID2	100%	99.9%	99.2%	69.1%	?Intellectual developmental disorder, X-linked 101, 300928	INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS
MIEF1	100%	100%	100%	99.5%	Optic atrophy 14, 620550	VISION DISORDERS PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS MITOCHONDRIAL DISORDERS PANEL

MIEF2	100%	100%	100%	98.8%	?Combined oxidative phosphorylation deficiency 49, 619024	MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS MITOCHONDRIAL DISORDERS PANEL
MINAR2	100%	100%	100%	99.3%	Deafness, autosomal recessive 120, 620238	HEARING IMPAIRMENT PANEL (INCLUDING GJB2) MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹
MINPP1	100%	100%	100%	99.3%	{Thyroid carcinoma, follicular}, 188470;Pontocerebellar hypoplasia, type 16, 619527	METABOLIC DISORDERS PANEL INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹

MIP	100%	100%	100%	97.7%	Cataract 15, multiple types, 615274	VISION DISORDERS PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS
MIPEP	100%	100%	100%	99.5%	Combined oxidative phosphorylation deficiency 31, 617228	HEART DISORDERS PANEL ¹ MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS MITOCHONDRIAL DISORDERS PANEL COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹
MIR140					Spondyloepiphyseal dysplasia, Nishimura type, 618618	SHORT STATURE/SKELETAL DYSPLASIA PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS

MIR17HG						INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS
MIR184					EDICT syndrome, 614303	VISION DISORDERS PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS
MIR204					Retinal dystrophy and iris coloboma with or without cataract, 616722	VISION DISORDERS PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS
MIR96					Deafness, autosomal dominant 50, 613074	HEARING IMPAIRMENT PANEL (INCLUDING GJB2) MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS

MITF	100%	100%	100%	99.3%	Waardenburg syndrome, type 2A, 193510;{Melanoma, cutaneous malignant, susceptibility to, 8}, 614456;Tietz albinism-deafness syndrome, 103500;COMMAD syndrome, 617306	VISION DISORDERS PANEL CRANIOFACIAL ANOMALIES PANEL WITH GENOME WIDE CNV ANALYSIS SKIN DISORDERS PANEL ¹ HEARING IMPAIRMENT PANEL (INCLUDING GJB2) PANEL MELANOMA, BASIC (CDKN2A, CDK4, MITF P.(GLU318LYS) ¹ PANEL MELANOMA, EXTENSIVE (CDKN2A, CDK4, MITF P.(GLU318LYS), BAP1, POT1, TERT PROMOTER) ¹ MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹ HEREDITARY CANCER PANEL
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MKKS	100%	100%	100%	99.6%	McKusick-Kaufman syndrome, 236700;Bardet-Biedl syndrome 6, 605231	VISION DISORDERS PANEL CILIOPATHIES PANEL COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL¹ INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS RENAL DISORDERS PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS DISORDERS/DIFFEREN CES OF SEX DEVELOPMENT (DSD) / PRIMARY ADRENAL INSUFFICIENCY PANEL HYPOGONADOTROPIC HYPOGONADISM (KALLMANN) PANEL
MKRN3	100%	100%	100%	99.6%	Precocious puberty, central, 2, 615346	DISORDERS/DIFFEREN CES OF SEX DEVELOPMENT (DSD) / PRIMARY ADRENAL INSUFFICIENCY PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS

MKS1	99.2%	99%	100%	99.3%	Bardet-Biedl syndrome 13, 615990;Meckel syndrome 1, 249000;Joubert syndrome 28, 617121	VISION DISORDERS PANEL RENAL DISORDERS PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹ OROFACIAL CLEFTING PANEL WITH GENOME WIDE CNV ANALYSIS CILIOPATHIES PANEL SHORT STATURE/SKELETAL DYSPLASIA PANEL WITH GENOME WIDE CNV ANALYSIS INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS
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MLC1	100%	100%	100%	99.1%	Megalencephalic leukoencephalopathy with subcortical cysts 1, 604004	MOVEMENT DISORDERS PANEL EPILEPSY PANEL INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL¹
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MLH1	100%	100%	100%	99.4%	Lynch syndrome 2, 609310;Muir-Torre syndrome, 158320;Mismatch repair cancer syndrome 1, 276300	INHERITED BONE MARROW FAILURE AND/OR PREDISPOSITION PANEL TO HEMATOLOGICAL MALIGNANCIES PANEL HEREDITARY COLORECTAL AND POLYPOSIS SKIN DISORDERS PANEL ¹ HEREDITARY CANCER PANEL PANEL HEREDITARY PANCREATIC CANCER (BRCA1, BRCA2, PALB2, ATM, CDKN2A, MLH1, MSH2, MSH6, PMS2, STK11) COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹ MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS PANEL LYNCH SYNDROME (MLH1, PMS2, MSH2, MSH6) PANEL HEREDITARY CANCER (CDH1, CTNNA1, MLH1, MSH2, MSH6, PMS2)
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MLH3	100%	100%	99.8%	98.5%	{Endometrial cancer, susceptibility to}, 608089;Colorectal cancer, somatic, 114500;Colorectal cancer, hereditary nonpolyposis, type 7, 614385	PANEL HEREDITARY COLORECTAL AND POLYPOSIS MALE INFERTILITY PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS HEREDITARY CANCER PANEL
MLIP	100%	100%	100%	99%	Myopathy with myalgia, increased serum creatine kinase, and with or without episodic rhabdomyolysis, 620138	MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹ MUSCLE DISORDERS PANEL
MLLT10	97%	97%	100%	98.6%	Leukemia, acute myeloid, 601626	MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS

MLPH	100%	100%	99.9%	98.5%	Griscelli syndrome, type 3, 609227	SKIN DISORDERS PANEL ¹ HEMOSTATIC/THROMB OTIC DISORDERS PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹
MLYCD	100%	100%	100%	98.7%	Malonyl-CoA decarboxylase deficiency, 248360	HEART DISORDERS PANEL ¹ METABOLIC DISORDERS PANEL INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹

MMAA	100%	100%	100%	99.3%	Methylmalonic aciduria, vitamin B12-responsive, cblA type, 251100	METABOLIC DISORDERS PANEL INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹
MMAB	100%	100%	100%	99.1%	Methylmalonic aciduria, vitamin B12-responsive, cblB type, 251110	METABOLIC DISORDERS PANEL INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹

MMACHC	100%	100%	100%	99%	Methylmalonic aciduria and homocystinuria, cbIC type, 277400	SKIN DISORDERS PANEL ¹ METABOLIC DISORDERS PANEL INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS RENAL DISORDERS PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹
MMADHC	89.3%	89.3%	100%	99.3%	Methylmalonic aciduria and homocystinuria, cbID type, 277410;Methylmalonic aciduria, cbID type, 620953;Homocystinuria-megaloblastic anemia, cbID type, 620952	MOVEMENT DISORDERS PANEL METABOLIC DISORDERS PANEL INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹

MME	97.7%	97.7%	100%	99.6%	?Spinocerebellar ataxia 43, 617018;Charcot-Marie-Tooth disease, axonal, type 2T, 617017	POLYNEUROPATHIES PANEL ¹ NEUROLOGICAL PAIN DISORDERS PANEL ¹ MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹
MMGT1	100%	100%	98.3%	70.3%		INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS
MMP1	100%	100%	100%	99.8%		SKIN DISORDERS PANEL ¹ MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS

MMP13	92.1%	92.1%	100%	98.9%	?Spondyloepimetaphyseal dysplasia, Missouri type, 602111;Metaphyseal anadysplasia 1, 602111;Metaphyseal dysplasia, Spahr type, 250400	SHORT STATURE/SKELETAL DYSPLASIA PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹
MMP14	94.9%	94.9%	100%	99%	Winchester syndrome, 277950	SKIN DISORDERS PANEL ¹ SHORT STATURE/SKELETAL DYSPLASIA PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹
MMP19	100%	100%	100%	99.1%	Cavitary optic disc anomalies, 611543	MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS

MMP2	100%	100%	100%	99%	Multicentric osteolysis, nodulosis, and arthropathy, 259600	SKIN DISORDERS PANEL ¹ SHORT STATURE/SKELETAL DYSPLASIA PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹
MMP20	100%	100%	100%	99.7%	Amelogenesis imperfecta, type IIA2, 612529	CRANIOFACIAL ANOMALIES PANEL WITH GENOME WIDE CNV ANALYSIS SKIN DISORDERS PANEL ¹ MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹

MMP21	100%	100%	100%	99.5%	Heterotaxy, visceral, 7, autosomal, 616749	CONGENITAL HEARTDISEASE PANEL¹ CILIOPATHIES PANEL HEART DISORDERS PANEL¹ MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL¹
MMP9	100%	100%	100%	98.8%	Metaphyseal anadysplasia 2, 613073	SHORT STATURE/SKELETAL DYSPLASIA PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL¹
MMS19	100%	100%	100%	99.3%		METABOLIC DISORDERS PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS

MMUT	100%	100%	100%	99.6%	Methylmalonic aciduria, mut(0) type, 251000	METABOLIC DISORDERS PANEL INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹
MN1	100%	100%	100%	98.8%	CEBALID syndrome, 618774;Meningioma, 607174	CRANIOFACIAL ANOMALIES PANEL WITH GENOME WIDE CNV ANALYSIS INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS

MNS1	100%	100%	100%	99.5%	Heterotaxy, visceral, 9, autosomal, with male infertility, 618948	CONGENITAL HEARTDISEASE PANEL¹ CILIOPATHIES PANEL HEART DISORDERS PANEL¹ MALE INFERTILITY PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS
MNX1	90.2%	84.1%	99.9%	92.2%	Currarino syndrome, 176450	SHORT STATURE/SKELETAL DYSPLASIA PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS
MOCOS	100%	100%	100%	98.9%	Xanthinuria, type II, 603592	METABOLIC DISORDERS PANEL RENAL DISORDERS PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL¹

MOCS1	100%	100%	100%	98.2%	Molybdenum cofactor deficiency A, 252150	EPILEPSY PANEL METABOLIC DISORDERS PANEL INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹
MOCS2	100%	100%	100%	99.3%	Molybdenum cofactor deficiency B, 252160	EPILEPSY PANEL METABOLIC DISORDERS PANEL INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹
MOG	100%	100%	100%	99.4%	?Narcolepsy 7, 614250	MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS

MOGS	100%	100%	100%	99.1%	Congenital disorder of glycosylation, type IIb, 606056	PRIMARY IMMUNODEFICIENCIES PANEL METABOLIC DISORDERS PANEL INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹
MORC2	100%	100%	100%	99.2%	Charcot-Marie-Tooth disease, axonal, type 2Z, 616688;Developmental delay, impaired growth, dysmorphic facies, and axonal neuropathy, 619090	POLYNEUROPATHIES PANEL ¹ METABOLIC DISORDERS PANEL INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS MITOCHONDRIAL DISORDERS PANEL

MOS	100%	100%	100%	97.1%	Oocyte/zygote/embryo maturation arrest 20, 620383	UNSUCCESSFUL IVF TREATMENTS, RECURRENT MOLAR PREGNANCIES AND/OR RECURRENT PREGNANCY LOSS PANEL ¹ MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS
MOV10L1	100%	100%	100%	99.3%	?Spermatogenic failure 73, 619878	MALE INFERTILITY PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS
MPC1	100%	100%	100%	99.8%	Mitochondrial pyruvate carrier deficiency, 614741	MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS MITOCHONDRIAL DISORDERS PANEL COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹

MPC2	100%	100%	100%	99.5%		METABOLIC DISORDERS PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS MITOCHONDRIAL DISORDERS PANEL
MPDU1	100%	100%	100%	98.4%	Congenital disorder of glycosylation, type If, 609180	SKIN DISORDERS PANEL ¹ EPILEPSY PANEL METABOLIC DISORDERS PANEL INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹

MPDZ	100%	100%	100%	99.2%	Hydrocephalus, congenital, 2, with or without brain or eye anomalies, 615219	VISION DISORDERS PANEL HEARING IMPAIRMENT PANEL (INCLUDING GJB2) EPILEPSY PANEL INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL¹
MPEG1	100%	100%	100%	99.1%	Immunodeficiency 77, 619223	PRIMARY IMMUNODEFICIENCIES PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS

MPI	100%	100%	100%	99.8%	Congenital disorder of glycosylation, type Ib, 602579	LYMPHATIC ANOMALIES PANEL WITH GENOME WIDE CNV ANALYSIS METABOLIC DISORDERS PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹
MPIG6B	100%	100%	100%	98.3%	?Thrombocytopenia, anemia, and myelofibrosis, 617441	HEMOSTATIC/THROMB OTIC DISORDERS PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS

MPL	100%	100%	100%	99.1%	Myelofibrosis with myeloid metaplasia, somatic, 254450;Amegakaryocytic thrombocytopenia, congenital, 1, 604498;Thrombocythemia 2, 601977	INHERITED BONE MARROW FAILURE AND/OR PREDISPOSITION PANEL TO HEMATOLOGICAL MALIGNANCIES HEMOSTATIC/THROMBOTIC DISORDERS PANEL IRON DISORDERS PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹ HEREDITARY CANCER PANEL
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MPLKIP	100%	100%	100%	97.7%	Trichothiodystrophy 4, nonphotosensitive, 234050	SKIN DISORDERS PANEL ¹ INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹
MPO	100%	100%	100%	99.2%	{Alzheimer disease, susceptibility to}, 104300;Myeloperoxidase deficiency, 254600;{Lung cancer, protection against, in smokers}	MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹

MPV17	100%	100%	100%	99.4%	Charcot-Marie-Tooth disease, axonal, type 2EE, 618400;Mitochondrial DNA depletion syndrome 6 (hepatocerebral type), 256810	POLYNEUROPATHIES PANEL ¹ LIVER DISORDERS PANEL INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS MITOCHONDRIAL DISORDERS PANEL COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹
MPZ	100%	100%	100%	98.6%	Charcot-Marie-Tooth disease, type 2I, 607677;Dejerine-Sottas disease, 145900;Charcot-Marie-Tooth disease, type 1B, 118200;Roussy-Levy syndrome, 180800;Charcot-Marie-Tooth disease, dominant intermediate D, 607791;Hypomyelinating neuropathy, congenital, 2, 618184;Charcot-Marie-Tooth disease, type 2J, 607736	FETAL AKINESIA PANEL HEARING IMPAIRMENT PANEL (INCLUDING GJB2) COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹ MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS POLYNEUROPATHIES PANEL ¹ NEUROLOGICAL PAIN DISORDERS PANEL ¹

MPZL2	100%	100%	100%	99.8%	Deafness, autosomal recessive 111, 618145	HEARING IMPAIRMENT PANEL (INCLUDING GJB2) MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL¹
MRAP	100%	100%	100%	99.8%	Glucocorticoid deficiency 2, 607398	DISORDERS/DIFFEREN CES OF SEX DEVELOPMENT (DSD) / PRIMARY ADRENAL INSUFFICIENCY PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL¹

MRAS	100%	100%	100%	99.5%	Noonan syndrome 11, 618499	MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS NOONAN SYNDROME / RASOPATHY PANEL SHORT STATURE/SKELETAL DYSPLASIA PANEL WITH GENOME WIDE CNV ANALYSIS LYMPHATIC ANOMALIES PANEL WITH GENOME WIDE CNV ANALYSIS INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS
MRE11	100%	100%	100%	99.4%	Ataxia-telangiectasia-like disorder 1, 604391	MOVEMENT DISORDERS PANEL HEREDITARY CANCER PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹ SKIN DISORDERS PANEL ¹

MRM2	97%	97%	100%	98.3%	Mitochondrial DNA depletion syndrome 17, 618567	COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹ MITOCHONDRIAL DISORDERS PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS
MRPL12	100%	100%	100%	99.1%	?Combined oxidative phosphorylation deficiency 45, 618951	MITOCHONDRIAL DISORDERS PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS
MRPL24	100%	100%	100%	96.8%		MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS MITOCHONDRIAL DISORDERS PANEL
MRPL3	100%	100%	100%	99.7%	Combined oxidative phosphorylation deficiency 9, 614582	MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS MITOCHONDRIAL DISORDERS PANEL COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹

MRPL39	100%	100%	100%	99.5%	Combined oxidative phosphorylation deficiency 59, 620646	MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS MITOCHONDRIAL DISORDERS PANEL COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹
MRPL40	100%	100%	100%	99.8%		MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS MITOCHONDRIAL DISORDERS PANEL
MRPL42	100%	100%	100%	99.8%		MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS MITOCHONDRIAL DISORDERS PANEL

MRPL44	100%	100%	100%	99.6%	Combined oxidative phosphorylation deficiency 16, 615395	HEART DISORDERS PANEL ¹ METABOLIC DISORDERS PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS MITOCHONDRIAL DISORDERS PANEL COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹
MRPL49	100%	100%	100%	98.9%		INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS MITOCHONDRIAL DISORDERS PANEL
MRPL50	100%	100%	100%	98.8%		HEARING IMPAIRMENT PANEL (INCLUDING GJB2) MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS MITOCHONDRIAL DISORDERS PANEL

MRPL57	100%	100%	100%	100%		MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS MITOCHONDRIAL DISORDERS PANEL
MRPS14	100%	100%	100%	99.2%	?Combined oxidative phosphorylation deficiency 38, 618378	MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS MITOCHONDRIAL DISORDERS PANEL
MRPS16	100%	100%	100%	99.6%	Combined oxidative phosphorylation deficiency 2, 610498	MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS MITOCHONDRIAL DISORDERS PANEL COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹
MRPS2	100%	100%	100%	99%	Combined oxidative phosphorylation deficiency 36, 617950	MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS MITOCHONDRIAL DISORDERS PANEL COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹

MRPS22	100%	100%	100%	99.1%	Ovarian dysgenesis 7, 618117;Combined oxidative phosphorylation deficiency 5, 611719	INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS MITOCHONDRIAL DISORDERS PANEL COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹
MRPS23	100%	100%	100%	99%	?Combined oxidative phosphorylation deficiency 46, 618952	MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS MITOCHONDRIAL DISORDERS PANEL
MRPS25	74.2%	74.2%	100%	99.4%	?Combined oxidative phosphorylation deficiency 50, 619025	MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS MITOCHONDRIAL DISORDERS PANEL

MRPS28	85.3%	85.3%	99.6%	91.1%	?Combined oxidative phosphorylation deficiency 47, 618958	MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS MITOCHONDRIAL DISORDERS PANEL
MRPS34	100%	100%	100%	99.6%	Combined oxidative phosphorylation deficiency 32, 617664	INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS MITOCHONDRIAL DISORDERS PANEL COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹
MRPS36	100%	100%	100%	98.8%		METABOLIC DISORDERS PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS MITOCHONDRIAL DISORDERS PANEL

MRPS7	100%	100%	100%	98.9%	?Combined oxidative phosphorylation deficiency 34, 617872	MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS MITOCHONDRIAL DISORDERS PANEL
MRRF	100%	100%	100%	99.4%		MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS MITOCHONDRIAL DISORDERS PANEL
MRTFA	100%	100%	100%	98.4%	?Immunodeficiency 66, 618847	PRIMARY IMMUNODEFICIENCIES PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS
MRTFB	100%	100%	100%	99.3%		INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS

MS4A1	100%	100%	100%	99.8%	?Immunodeficiency, common variable, 5, 613495	PRIMARY IMMUNODEFICIENCIES PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL¹
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MSH2	100%	100%	100%	99.4%	Lynch syndrome 1, 120435;Muir-Torre syndrome, 158320;Mismatch repair cancer syndrome 2, 619096	COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹ HEREDITARY CANCER PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS PANEL HEREDITARY PANCREATIC CANCER (BRCA1, BRCA2, PALB2, ATM, CDKN2A, MLH1, MSH2, MSH6, PMS2, STK11) SKIN DISORDERS PANEL ¹ PANEL HEREDITARY CANCER (CDH1, CTNNA1, MLH1, MSH2, MSH6, PMS2) PANEL LYNCH SYNDROME (MLH1, PMS2, MSH2, MSH6) INHERITED BONE MARROW FAILURE AND/OR PREDISPOSITION PANEL TO HEMATOLOGICAL MALIGNANCIES PANEL HEREDITARY COLORECTAL AND POLYPOSIS
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MSH3	100%	100%	100%	98.7%	Familial adenomatous polyposis 4, 617100;Endometrial carcinoma, somatic, 608089	PANEL HEREDITARY COLORECTAL AND POLYPOSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹ HEREDITARY CANCER PANEL
MSH4	100%	100%	100%	99.7%	Premature ovarian failure 20, 619938;Spermatogenic failure 2, 108420	DISORDERS/DIFFERENCES OF SEX DEVELOPMENT (DSD) / PRIMARY ADRENAL INSUFFICIENCY PANEL MALE INFERTILITY PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS PREMATURE OVARIAN INSUFFICIENCY PANEL
MSH5	100%	100%	100%	98.9%	?Premature ovarian failure 13, 617442;Spermatogenic failure 74, 619937	MALE INFERTILITY PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS

MSH6	100%	100%	100%	98.6%	Lynch syndrome 5, 614350;Mismatch repair cancer syndrome 3, 619097;{Endometrial cancer, familial}, 608089	PANEL HEREDITARY PANCREATIC CANCER (BRCA1, BRCA2, PALB2, ATM, CDKN2A, MLH1, MSH2, MSH6, PMS2, STK11) COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹ HEREDITARY CANCER PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS PANEL HEREDITARY COLORECTAL AND POLYPOSIS PANEL HEREDITARY CANCER (CDH1, CTNNA1, MLH1, MSH2, MSH6, PMS2) PANEL LYNCH SYNDROME (MLH1, PMS2, MSH2, MSH6) INHERITED BONE MARROW FAILURE AND/OR PREDISPOSITION PANEL TO HEMATOLOGICAL MALIGNANCIES
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MSL2	92.7%	92.7%	100%	99.8%	Karayol-Borroto-Haghshenas neurodevelopmental syndrome, 620985	INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS
MSL3	100%	99.8%	99%	68.9%	Basilicata-Akhtar syndrome, 301032	INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS
MSMO1	100%	100%	100%	99.3%	Microcephaly, congenital cataract, and psoriasiform dermatitis, 616834	METABOLIC DISORDERS PANEL INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹

MSN	100%	98.6%	99.2%	68.4%	Immunodeficiency 50, 300988	PRIMARY IMMUNODEFICIENCIES PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS
MSR1	100%	100%	100%	98.3%	Barrett esophagus/esophageal adenocarcinoma, 614266	MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS
MSRB3	100%	100%	100%	98.9%	Deafness, autosomal recessive 74, 613718	HEARING IMPAIRMENT PANEL (INCLUDING GJB2) MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹
MST1R	100%	100%	100%	99.3%	{Nasopharyngeal carcinoma, susceptibility to, 3}, 617075	CONGENITAL HEARTDISEASE PANEL ¹ HEART DISORDERS PANEL ¹ MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS

MSTN	100%	100%	100%	99.9%	?Muscle hypertrophy, 614160	MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS MUSCLE DISORDERS PANEL
MSTO1	100%	100%	100%	97.7%	Myopathy, mitochondrial, and ataxia, 617675	MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS MITOCHONDRIAL DISORDERS PANEL COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹ MUSCLE DISORDERS PANEL
MSX1	100%	100%	100%	99.6%	Tooth agenesis, selective, 1, with or without orofacial cleft, 106600;Ectodermal dysplasia 3, Witkop type, 189500;Orofacial cleft 5, 608874	CRANIOFACIAL ANOMALIES PANEL WITH GENOME WIDE CNV ANALYSIS SKIN DISORDERS PANEL ¹ MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS OROFACIAL CLEFTING PANEL WITH GENOME WIDE CNV ANALYSIS

MSX2	100%	100%	100%	98.8%	Parietal foramina with cleidocranial dysplasia, 168550;Craniosynostosis 2, 604757;Parietal foramina 1, 168500	CRANIOFACIAL ANOMALIES PANEL WITH GENOME WIDE CNV ANALYSIS SHORT STATURE/SKELETAL DYSPLASIA PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS
MTAP	100%	100%	100%	98.3%	Diaphyseal medullary stenosis with malignant fibrous histiocyoma, 112250	SHORT STATURE/SKELETAL DYSPLASIA PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS HEREDITARY CANCER PANEL

MTFMT	100%	100%	100%	97.9%	Combined oxidative phosphorylation deficiency 15, 614947;Mitochondrial complex I deficiency, nuclear type 27, 618248	VISION DISORDERS PANEL EPILEPSY PANEL INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS MITOCHONDRIAL DISORDERS PANEL COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹
MTHFD1	100%	100%	100%	99.5%	{Neural tube defects, folate-sensitive, susceptibility to}, 601634;Combined immunodeficiency and megaloblastic anemia with or without hyperhomocysteinemia, 617780	PRIMARY IMMUNODEFICIENCIES PANEL METABOLIC DISORDERS PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹

MTHFR	100%	100%	100%	99.2%	{Vascular disease, susceptibility to};Homocystinuria due to MTHFR deficiency, 236250;{Thromboembolism, susceptibility to}, 188050;{Schizophrenia, susceptibility to}, 181500;{Neural tube defects, susceptibility to}, 601634	MOVEMENT DISORDERS PANEL EPILEPSY PANEL HEMOSTATIC/THROMB OTIC DISORDERS PANEL METABOLIC DISORDERS PANEL INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹
MTHFS	100%	100%	100%	98.4%	Neurodevelopmental disorder with microcephaly, epilepsy, and hypomyelination, 618367	MOVEMENT DISORDERS PANEL INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹

MTM1	100%	100%	99.4%	69.2%	Myopathy, centronuclear, X-linked, 310400	FETAL AKINESIA PANEL LIVER DISORDERS PANEL METABOLIC DISORDERS PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS MUSCLE DISORDERS PANEL
MTMR2	100%	100%	100%	99.4%	Charcot-Marie-Tooth disease, type 4B1, 601382	POLYNEUROPATHIES PANEL ¹ METABOLIC DISORDERS PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹

MTO1	95%	91.8%	100%	99%	Combined oxidative phosphorylation deficiency 10, 614702	HEART DISORDERS PANEL ¹ INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS MITOCHONDRIAL DISORDERS PANEL COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹
MTOR	100%	100%	100%	99.2%	Focal cortical dysplasia, type II, somatic, 607341;Smith-Kingsmore syndrome, 616638	SKIN DISORDERS PANEL ¹ EPILEPSY PANEL TALL STATURE PANEL WITH GENOME WIDE CNV ANALYSIS INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS

MTPAP	100%	100%	100%	98.4%	?Spastic ataxia 4, autosomal recessive, 613672	MOVEMENT DISORDERS PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS MITOCHONDRIAL DISORDERS PANEL COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹
MTR	100%	100%	100%	99.5%	{Neural tube defects, folate-sensitive, susceptibility to}, 601634;Homocystinuria-megaloblastic anemia, cblG complementation type, 250940	METABOLIC DISORDERS PANEL INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹

MTRFR	100%	99.8%	100%	99%	Spastic paraplegia 55, autosomal recessive, 615035;Combined oxidative phosphorylation deficiency 7, 613559	MOVEMENT DISORDERS PANEL VISION DISORDERS PANEL POLYNEUROPATHIES PANEL ¹ INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS MITOCHONDRIAL DISORDERS PANEL COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹
MTRR	100%	100%	100%	99.4%	Homocystinuria-megaloblastic anemia, cbl E type, 236270;{Neural tube defects, folate-sensitive, susceptibility to}, 601634	EPILEPSY PANEL METABOLIC DISORDERS PANEL INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹

MTSS2	100%	100%	100%	98.6%	Intellectual developmental disorder with ocular anomalies and distinctive facial features, 620086	INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS
MT-TI	99.6%	95.9%				MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS HEART DISORDERS PANEL ¹ HYPERTROPHIC CARDIOMYOPATHY PANEL ¹
MTTP	100%	100%	100%	99.5%	Abetalipoproteinemia, 200100	MOVEMENT DISORDERS PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹

MTX2	100%	100%	100%	99%	Mandibuloacral dysplasia progeroid syndrome, 619127	SHORT STATURE/SKELETAL DYSPLASIA PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS MITOCHONDRIAL DISORDERS PANEL COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹
MUC1	100%	100%	100%	98%	Tubulointerstitial kidney disease, autosomal dominant, 2, 174000	MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS
MUC16	100%	100%	100%	99.2%		CONGENITAL HEARTDISEASE PANEL ¹ HEART DISORDERS PANEL ¹ MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS

MUSK	100%	100%	100%	98.9%	Fetal akinesia deformation sequence 1, 208150;Myasthenic syndrome, congenital, 9, associated with acetylcholine receptor deficiency, 616325	FETAL AKINESIA PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹ MUSCLE DISORDERS PANEL
MUTYH	100%	100%	100%	99.5%	Adenomas, multiple colorectal, 608456;Gastric cancer, somatic, 613659	PANEL HEREDITARY COLORECTAL AND POLYPOSIS SKIN DISORDERS PANEL ¹ MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹ HEREDITARY CANCER PANEL
MVD	100%	100%	100%	99%	Porokeratosis 7, multiple types, 614714	SKIN DISORDERS PANEL ¹ MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS

MVK	100%	100%	100%	99.2%	Hyper-IgD syndrome, 260920;Porokeratosis 3, multiple types, 175900;Mevalonic aciduria, 610377	VISION DISORDERS PANEL INHERITED BONE MARROW FAILURE AND/OR PREDISPOSITION PANEL TO HEMATOLOGICAL MALIGNANCIES SKIN DISORDERS PANEL ¹ PRIMARY IMMUNODEFICIENCIES PANEL METABOLIC DISORDERS PANEL INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹
MXI1	100%	99.2%	99.9%	96.6%	Prostate cancer, somatic, 176807;Neurofibrosarcoma, somatic	MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS

MYBPC1	100%	100%	100%	99.5%	Congenital myopathy 16, 618524;Lethal congenital contracture syndrome 4, 614915;Arthrogryposis, distal, type 1B, 614335	FETAL AKINESIA PANEL MOVEMENT DISORDERS PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹ MUSCLE DISORDERS PANEL
MYBPC3	100%	100%	100%	98.5%	Cardiomyopathy, hypertrophic, 4, 115197;Cardiomyopathy, dilated, 1MM, 615396;Left ventricular noncompaction 10, 615396	HEART DISORDERS PANEL ¹ HYPERTROPHIC CARDIOMYOPATHY PANEL ¹ MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS
MYBPHL	100%	100%	100%	99.2%		HEART DISORDERS PANEL ¹ MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS

MYC	100%	100%	100%	99%	Burkitt lymphoma, somatic, 113970	MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS
MYCBP2	100%	99.9%	100%	99.4%		INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS
MYCN	100%	99.4%	100%	98.6%	Feingold syndrome 1, 164280;Megalencephaly-polydactyly syndrome, 620748	FETAL AKINESIA PANEL SHORT STATURE/SKELETAL DYSPLASIA PANEL WITH GENOME WIDE CNV ANALYSIS INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS

MYD88	100%	100%	100%	99.4%	Macroglobulinemia, Waldenstrom, somatic, 153600;Immunodeficiency 68, 612260	PRIMARY IMMUNODEFICIENCIES PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL¹
MYF5	100%	100%	100%	99.4%	Ophthalmoplegia, external, with rib and vertebral anomalies, 618155	MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL¹
MYH10	100%	100%	100%	99%		INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS

MYH11	100%	100%	100%	98.8%	Megacystis-microcolon-intestinal hypoperistalsis syndrome 2, 619351;Aortic aneurysm, familial thoracic 4, 132900;Visceral myopathy 2, 619350	THORACIC AORTIC ANEURYSM AND/OR DISSECTION PANEL CONGENITAL HEARTDISEASE PANEL ¹ HEART DISORDERS PANEL ¹ LIVER DISORDERS PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹
MYH14	100%	100%	100%	97.1%	?Peripheral neuropathy, myopathy, hoarseness, and hearing loss, 614369;Deafness, autosomal dominant 4A, 600652	HEARING IMPAIRMENT PANEL (INCLUDING GJB2) POLYNEUROPATHIES PANEL ¹ MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS

MYH2	100%	100%	100%	99.6%	Congenital myopathy 6 with ophthalmoplegia, 605637	MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹ MUSCLE DISORDERS PANEL
MYH3	100%	100%	100%	99.3%	Contractures, pterygia, and spondylocarpostarsal fusion syndrome 1A, 178110;Contractures, pterygia, and spondylocarpotarsal fusion syndrome 1B, 618469;Arthrogryposis, distal, type 2B3 (Sheldon-Hall), 618436;Arthrogryposis, distal, type 2A (Freeman-Sheldon), 193700	FETAL AKINESIA PANEL SHORT STATURE/SKELETAL DYSPLASIA PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹ MUSCLE DISORDERS PANEL

MYH6	100%	100%	100%	99%	?Atrial septal defect 3, 614089;{Sick sinus syndrome 3}, 614090;Cardiomyopathy, dilated, 1EE, 613252;Cardiomyopathy, hypertrophic, 14, 613251	CONGENITAL HEARTDISEASE PANEL ¹ HEART DISORDERS PANEL ¹ MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS
MYH7	100%	100%	100%	99%	Laing distal myopathy, 160500;Cardiomyopathy, hypertrophic, 1, 192600;Left ventricular noncompaction 5, 613426;Cardiomyopathy, dilated, 1S, 613426;Congenital myopathy 7B, myosin storage, autosomal recessive, 255160;Congenital myopathy 7A, myosin storage, autosomal dominant, 608358	CONGENITAL HEARTDISEASE PANEL ¹ DILATED CARDIOMYOPATHY PANEL ¹ HEART DISORDERS PANEL ¹ HYPERTROPHIC CARDIOMYOPATHY PANEL ¹ MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS MUSCLE DISORDERS PANEL
MYH7B	100%	100%	100%	99.1%		HEART DISORDERS PANEL ¹ MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS

MYH8	100%	100%	100%	99.5%	Carney complex variant, 608837;Trismus-pseudocamptodactyly syndrome, 158300	FETAL AKINESIA PANEL SKIN DISORDERS PANEL ¹ MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS
MYH9	97.2%	97.2%	100%	99%	Macrothrombocytopenia and granulocyte inclusions with or without nephritis or sensorineural hearing loss, 155100;Deafness, autosomal dominant 17, 603622	VISION DISORDERS PANEL INHERITED BONE MARROW FAILURE AND/OR PREDISPOSITION PANEL TO HEMATOLOGICAL MALIGNANCIES RENAL DISORDERS PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS HEARING IMPAIRMENT PANEL (INCLUDING GJB2) HEMOSTATIC/THROMB OTIC DISORDERS PANEL

MYL1	100%	100%	100%	99.2%	Congenital myopathy 14, 618414	MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹ MUSCLE DISORDERS PANEL
MYL11	100%	100%	100%	98.3%	Arthrogryposis, distal, type 1C, 619110	FETAL AKINESIA PANEL SHORT STATURE/SKELETAL DYSPLASIA PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS
MYL2	100%	100%	100%	98.1%	Cardiomyopathy, hypertrophic, 10, 608758;Myopathy, myofibrillar, 12, infantile-onset, with cardiomyopathy, 619424	HEART DISORDERS PANEL ¹ HYPERTROPHIC CARDIOMYOPATHY PANEL ¹ MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹

MYL3	100%	100%	100%	98.6%	Cardiomyopathy, hypertrophic, 8, 608751	HEART DISORDERS PANEL ¹ HYPERTROPHIC CARDIOMYOPATHY PANEL ¹ MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹
MYL4	100%	100%	100%	99.7%	?Atrial fibrillation, familial, 18, 617280	HEART DISORDERS PANEL ¹ MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS
MYL7	100%	99.9%	100%	98.8%		HEART DISORDERS PANEL ¹ MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS
MYL9	100%	100%	100%	99.5%	?Megacystis-microcolon-intestinal hypoperistalsis syndrome 4, 619365	LIVER DISORDERS PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS

MYLK	99.2%	99.2%	100%	98.8%	Megacystis-microcolon-intestinal hypoperistalsis syndrome 1, 249210;Aortic aneurysm, familial thoracic 7, 613780	THORACIC AORTIC ANEURYSM AND/OR DISSECTION PANEL LIVER DISORDERS PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹
MYLK2	100%	100%	100%	99%	Cardiomyopathy, hypertrophic, 1, digenic, 192600	MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS
MYLK3	100%	100%	100%	99%		HEART DISORDERS PANEL ¹ MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS

MYMK	100%	100%	100%	98.6%	Carey-Fineman-Ziter syndrome, 254940	FETAL AKINESIA PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹ OROFACIAL CLEFTING PANEL WITH GENOME WIDE CNV ANALYSIS
MYMX	100%	100%	100%	98.7%	?Carey-Fineman-Ziter syndrome 2, 619941	MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS OROFACIAL CLEFTING PANEL WITH GENOME WIDE CNV ANALYSIS
MYO15A	100%	100%	100%	98.6%	Deafness, autosomal recessive 3, 600316	HEARING IMPAIRMENT PANEL (INCLUDING GJB2) MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹

MYO18B	100%	100%	100%	98.5%	Klippel-Feil syndrome 4, autosomal recessive, with myopathy and facial dysmorphism, 616549	MUSCLE DISORDERS PANEL SHORT STATURE/SKELETAL DYSPLASIA PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹
MYO1E	100%	100%	100%	99.4%	Glomerulosclerosis, focal segmental, 6, 614131	RENAL DISORDERS PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹
MYO1H	100%	100%	100%	99.3%	?Central hypoventilation syndrome, congenital, 2, and autonomic dysfunction, 619482	MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS

MYO3A	100%	100%	100%	99.5%	Deafness, autosomal recessive 30, 607101;Deafness, autosomal dominant 90, 620722	HEARING IMPAIRMENT PANEL (INCLUDING GJB2) MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹
MYO5A	99.1%	99%	100%	99.3%	Griscelli syndrome, type 1, 214450	VISION DISORDERS PANEL SKIN DISORDERS PANEL ¹ HEMOSTATIC/THROMB OTIC DISORDERS PANEL INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹

MYO5B	100%	100%	100%	99.1%	Diarrhea 2, with microvillus atrophy, with or without cholestasis, 251850;Cholestasis, progressive familial intrahepatic, 10, 619868	LIVER DISORDERS PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹
MYO6	100%	100%	100%	99.5%	Deafness, autosomal dominant 22, with hypertrophic cardiomyopathy, 606346;Deafness, autosomal dominant 22, 606346;Deafness, autosomal recessive 37, 607821	HEARING IMPAIRMENT PANEL (INCLUDING GJB2) HEART DISORDERS PANEL ¹ MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹

MYO7A	100%	100%	100%	99.1%	Deafness, autosomal recessive 2, 600060;Usher syndrome, type 1B, 276900;Deafness, autosomal dominant 11, 601317	VISION DISORDERS PANEL HEARING IMPAIRMENT PANEL (INCLUDING GJB2) MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹
MYO9A	100%	100%	100%	99.6%	Myasthenic syndrome, congenital, 24, presynaptic, 618198	INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹
MYOC	100%	100%	100%	99.1%	Glaucoma 1A, primary open angle, 137750	VISION DISORDERS PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS

MYOCD	100%	100%	100%	99.2%	Megabladder, congenital, 618719	MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS
MYOD1	100%	100%	100%	98.5%	Congenital myopathy 17, 618975	FETAL AKINESIA PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS MUSCLE DISORDERS PANEL
MYOF	100%	100%	100%	99.4%	?Angioedema, hereditary, 7, 619366	PRIMARY IMMUNODEFICIENCIES PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS
MYOM1	100%	100%	100%	99.3%		HEART DISORDERS PANEL ¹ MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS

MYORG	100%	100%	100%	99.2%	Basal ganglia calcification, idiopathic, 7, autosomal recessive, 618317	MOVEMENT DISORDERS PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS PARKINSON DISEASE PANEL COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹
MYOT	100%	100%	100%	99%	Myopathy, myofibrillar, 3, 609200	HEART DISORDERS PANEL ¹ POLYNEUROPATHIES PANEL ¹ MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS MUSCLE DISORDERS PANEL
MYOZ2	100%	100%	100%	99.7%	Cardiomyopathy, hypertrophic, 16, 613838	HEART DISORDERS PANEL ¹ MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS

MYPN	98.4%	98.4%	100%	99.3%	Cardiomyopathy, hypertrophic, 22, 615248;Congenital myopathy 24, 617336;Cardiomyopathy, familial restrictive, 4, 615248;Cardiomyopathy, dilated, 1KK, 615248	HEART DISORDERS PANEL ¹ MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹ MUSCLE DISORDERS PANEL
MYRF	100%	100%	100%	98.8%	Encephalitis/encephalopathy, mild, with reversible myelin vacuolization, 618113;Cardiac-urogenital syndrome, 618280	VISION DISORDERS PANEL CONGENITAL HEARTDISEASE PANEL ¹ DISORDERS/DIFFEREN CES OF SEX DEVELOPMENT (DSD) / PRIMARY ADRENAL INSUFFICIENCY PANEL HEART DISORDERS PANEL ¹ MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS

MYSM1	100%	100%	100%	99.6%	Bone marrow failure syndrome 4, 618116	INHERITED BONE MARROW FAILURE AND/OR PREDISPOSITION PANEL TO HEMATOLOGICAL MALIGNANCIES PRIMARY IMMUNODEFICIENCIES PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL¹
MYT1L	100%	100%	100%	99%	Intellectual developmental disorder, autosomal dominant 39, 616521	INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS

MYZAP	100%	100%	100%	99.3%	Cardiomyopathy, dilated, 2K, 620894	HEART DISORDERS PANEL ¹ MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹
NAA10	100%	99.7%	98.4%	66.6%	Microphthalmia, syndromic 1, 309800;Ogden syndrome, 300855	VISION DISORDERS PANEL CRANIOFACIAL ANOMALIES PANEL WITH GENOME WIDE CNV ANALYSIS SKIN DISORDERS PANEL ¹ INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS

NAA15	100%	100%	100%	99.3%	Intellectual developmental disorder, autosomal dominant 50, with behavioral abnormalities, 617787	INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS CONGENITAL HEARTDISEASE PANEL ¹ HEART DISORDERS PANEL ¹
NAA20	100%	100%	100%	99.8%	Intellectual developmental disorder, autosomal recessive 73, 619717	INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹

NAA60	100%	99.9%	100%	98.5%	Basal ganglia calcification, idiopathic, 9, autosomal recessive, 620786	INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹
NAA80	100%	100%	100%	98.7%	?Auroneurodental syndrome, 620830	MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS
NACC1	100%	100%	100%	99.3%	Neurodevelopmental disorder with epilepsy, cataracts, feeding difficulties, and delayed brain myelination, 617393	EPILEPSY PANEL INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS

NADK2	100%	99.6%	100%	99.2%	2,4-dienoyl-CoA reductase deficiency, 616034	METABOLIC DISORDERS PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS MITOCHONDRIAL DISORDERS PANEL COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹
NADSYN1	100%	100%	100%	99.1%	Vertebral, cardiac, renal, and limb defects syndrome 3, 618845	SHORT STATURE/SKELETAL DYSPLASIA PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹

NAE1	100%	100%	100%	99.5%	Neurodevelopmental disorder with dysmorphic facies and ischiopubic hypoplasia, 620210	INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹
NAF1	100%	100%	100%	98.6%	Pulmonary fibrosis and/or bone marrow failure syndrome, telomere-related, 7, 620365	INHERITED BONE MARROW FAILURE AND/OR PREDISPOSITION PANEL TO HEMATOLOGICAL MALIGNANCIES DYSKERATOSIS CONGENITA AND APLASTIC ANEMIA PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS HEREDITARY CANCER PANEL

NAGA	100%	100%	100%	98.9%	Schindler disease, type I, 609241;Kanzaki disease, 609242;Schindler disease, type III, 609241	SKIN DISORDERS PANEL ¹ METABOLIC DISORDERS PANEL INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹
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NAGLU	100%	100%	100%	99.1%	?Charcot-Marie-Tooth disease, axonal, type 2V, 616491;Mucopolysaccharidosis type IIIB (Sanfilippo B), 252920	POLYNEUROPATHIES PANEL ¹ NEUROLOGICAL PAIN DISORDERS PANEL ¹ SHORT STATURE/SKELETAL DYSPLASIA PANEL WITH GENOME WIDE CNV ANALYSIS METABOLIC DISORDERS PANEL INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹
NAGS	100%	100%	100%	99.1%	N-acetylglutamate synthase deficiency, 237310	METABOLIC DISORDERS PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹

NALCN	100%	100%	100%	99.3%	Congenital contractures of the limbs and face, hypotonia, and developmental delay, 616266;Hypotonia, infantile, with psychomotor retardation and characteristic facies 1, 615419	INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹
NANOS1	99.8%	97.8%	100%	96.3%	Spermatogenic failure 12, 615413	MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS

NANS	100%	100%	100%	99.4%	Spondyloepimetaphyseal dysplasia, Genevieve type, 610442	MOVEMENT DISORDERS PANEL EPILEPSY PANEL SHORT STATURE/SKELETAL DYSPLASIA PANEL WITH GENOME WIDE CNV ANALYSIS METABOLIC DISORDERS PANEL INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹
NAPB	100%	100%	100%	99.7%	Developmental and epileptic encephalopathy 107, 620033	INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹

NARS1	100%	100%	100%	99.4%	Neurodevelopmental disorder with microcephaly, impaired language, epilepsy, and gait abnormalities, autosomal dominant, 619092;Neurodevelopmental disorder with microcephaly, impaired language, and gait abnormalities, autosomal recessive, 619091	POLYNEUROPATHIES PANEL ¹ INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹
NARS2	92.3%	92.3%	100%	99.5%	Combined oxidative phosphorylation deficiency 24, 616239;?Deafness, autosomal recessive 94, 618434	MOVEMENT DISORDERS PANEL HEARING IMPAIRMENT PANEL (INCLUDING GJB2) EPILEPSY PANEL INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS MITOCHONDRIAL DISORDERS PANEL COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹

NAT8L	92.4%	82.1%	100%	95.8%	?N-acetylaspartate deficiency, 614063	MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹
NAV3	100%	100%	100%	99.3%		INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS
NAXD	99.6%	96.6%	100%	99.1%	Encephalopathy, progressive, early-onset, with brain edema and/or leukoencephalopathy, 2, 618321	METABOLIC DISORDERS PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS MITOCHONDRIAL DISORDERS PANEL COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹

NAXE	97.2%	91.7%	100%	98.5%	Encephalopathy, progressive, early-onset, with brain edema and/or leukoencephalopathy, 617186	METABOLIC DISORDERS PANEL INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS MITOCHONDRIAL DISORDERS PANEL COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹
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NBAS	100%	100%	100%	99.2%	Short stature, optic nerve atrophy, and Pelger-Huet anomaly, 614800;Infantile liver failure syndrome 2, 616483	VISION DISORDERS PANEL SKIN DISORDERS PANEL ¹ PRIMARY IMMUNODEFICIENCIES PANEL SHORT STATURE/SKELETAL DYSPLASIA PANEL WITH GENOME WIDE CNV ANALYSIS LIVER DISORDERS PANEL METABOLIC DISORDERS PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹
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NBEA	97.6%	97.6%	100%	99.3%	Neurodevelopmental disorder with or without early-onset generalized epilepsy, 619157	EPILEPSY PANEL HEMOSTATIC/THROMBOTIC DISORDERS PANEL INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS
NBEAL2	100%	100%	100%	99.3%	Gray platelet syndrome, 139090	INHERITED BONE MARROW FAILURE AND/OR PREDISPOSITION PANEL TO HEMATOLOGICAL MALIGNANCIES HEMOSTATIC/THROMBOTIC DISORDERS PANEL PRIMARY IMMUNODEFICIENCIES PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹

NBN	97.5%	97.5%	100%	99.7%	Leukemia, acute lymphoblastic, 613065;Aplastic anemia, 609135;Nijmegen breakage syndrome, 251260	INHERITED BONE MARROW FAILURE AND/OR PREDISPOSITION PANEL TO HEMATOLOGICAL MALIGNANCIES PRIMARY IMMUNODEFICIENCIES PANEL INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹ HEREDITARY CANCER PANEL
NCAPD2	100%	100%	100%	99.3%	Microcephaly 21, primary, autosomal recessive, 617983	MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹

NCAPD3	100%	100%	100%	99.6%	Microcephaly 22, primary, autosomal recessive, 617984	MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹
NCAPG2	100%	100%	100%	99.6%	Khan-Khan-Katsanis syndrome, 618460	CILIOPATHIES PANEL INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS RENAL DISORDERS PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹
NCAPH	100%	100%	100%	99.6%	?Microcephaly 23, primary, autosomal recessive, 617985	MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹

NCDN	100%	100%	100%	99.1%	Neurodevelopmental disorder with infantile epileptic spasms, 619373	EPILEPSY PANEL INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS
NCF1	100%	99.6%	99.9%	96.3%	Chronic granulomatous disease 1, autosomal recessive, 233700	SKIN DISORDERS PANEL ¹ PRIMARY IMMUNODEFICIENCIES PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹
NCF2	100%	100%	100%	99.3%	Chronic granulomatous disease 2, autosomal recessive, 233710	PRIMARY IMMUNODEFICIENCIES PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹

NCF4	100%	100%	100%	97.9%	Chronic granulomatous disease 3, autosomal recessive, 613960	PRIMARY IMMUNODEFICIENCIES PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL¹
NCKAP1	100%	100%	100%	99.4%		INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS
NCKAP1L	100%	100%	100%	99.5%	Immunodeficiency 72 with autoinflammation, 618982	PRIMARY IMMUNODEFICIENCIES PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL¹

NCOA3	100%	100%	100%	99.5%		HEARING IMPAIRMENT PANEL (INCLUDING GJB2) MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS
NCOR2	100%	100%	100%	98.4%		INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS
NCSTN	100%	100%	100%	99.2%	Acne inversa, familial, 1, 142690	SKIN DISORDERS PANEL ¹ PRIMARY IMMUNODEFICIENCIES PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS

NDC1	100%	100%	100%	99.3%		INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS
NDE1	100%	100%	100%	98.4%	Microhydranencephaly, 605013;Lissencephaly 4 (with microcephaly), 614019	INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹
NDN	100%	100%	100%	98.7%		MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS

NDNF	100%	100%	100%	99.4%	Hypogonadotropic hypogonadism 25 with anosmia, 618841	DISORDERS/DIFFERENCES OF SEX DEVELOPMENT (DSD) / PRIMARY ADRENAL INSUFFICIENCY PANEL HYPOGONADOTROPIC HYPOGONADISM (KALLMANN) PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS
NDP	100%	99.7%	99.3%	69.9%	Exudative vitreoretinopathy 2, X-linked, 305390;Norrie disease, 310600	VISION DISORDERS PANEL HEARING IMPAIRMENT PANEL (INCLUDING GJB2) INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS

NDRG1	100%	100%	100%	99.6%	Charcot-Marie-Tooth disease, type 4D, 601455	POLYNEUROPATHIES PANEL ¹ MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹
NDST1	100%	100%	100%	99.1%	Intellectual developmental disorder, autosomal recessive 46, 616116	INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹
NDUFA1	100%	100%	98.7%	62.6%	Mitochondrial complex I deficiency, nuclear type 12, 301020	EPILEPSY PANEL INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS MITOCHONDRIAL DISORDERS PANEL

NDUFA10	84%	81.6%	100%	99.2%	Mitochondrial complex I deficiency, nuclear type 22, 618243	MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS MITOCHONDRIAL DISORDERS PANEL COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹
NDUFA11	100%	98.9%	100%	99%	Mitochondrial complex I deficiency, nuclear type 14, 618236	EPILEPSY PANEL INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS MITOCHONDRIAL DISORDERS PANEL COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹

NDUFA12	79.4%	79.4%	100%	99.5%	Mitochondrial complex I deficiency, nuclear type 23, 618244	VISION DISORDERS PANEL INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS MITOCHONDRIAL DISORDERS PANEL COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL¹
NDUFA13	100%	100%	100%	99.5%	{Thyroid carcinoma, Hurthle cell}, 607464;Mitochondrial complex I deficiency, nuclear type 28, 618249	MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS MITOCHONDRIAL DISORDERS PANEL COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL¹

NDUFA2	100%	100%	100%	99%	Mitochondrial complex I deficiency, nuclear type 13, 618235	INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS MITOCHONDRIAL DISORDERS PANEL COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹
NDUFA3	94%	87.4%	100%	95.1%		MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS MITOCHONDRIAL DISORDERS PANEL
NDUFA4	100%	100%	100%	99.3%	?Mitochondrial complex IV deficiency, nuclear type 21, 619065	MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS MITOCHONDRIAL DISORDERS PANEL

NDUFA5	75%	75%	100%	99.4%		MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS MITOCHONDRIAL DISORDERS PANEL
NDUFA6	100%	100%	100%	99.2%	Mitochondrial complex I deficiency, nuclear type 33, 618253	MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS MITOCHONDRIAL DISORDERS PANEL COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹
NDUFA7	100%	100%	100%	98.9%		VISION DISORDERS PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS MITOCHONDRIAL DISORDERS PANEL

NDUFA8	100%	100%	100%	99.2%	Mitochondrial complex I deficiency, nuclear type 37, 619272	INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS MITOCHONDRIAL DISORDERS PANEL COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹
NDUFA9	100%	100%	100%	99.5%	Mitochondrial complex I deficiency, nuclear type 26, 618247	POLYNEUROPATHIES PANEL ¹ MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS MITOCHONDRIAL DISORDERS PANEL COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹
NDUFAB1	100%	100%	100%	99.5%		MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS MITOCHONDRIAL DISORDERS PANEL

NDUFAF1	100%	100%	100%	99.6%	Mitochondrial complex I deficiency, nuclear type 11, 618234	CONGENITAL HEARTDISEASE PANEL¹ EPILEPSY PANEL HEART DISORDERS PANEL¹ MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS MITOCHONDRIAL DISORDERS PANEL COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL¹
NDUFAF2	67.4%	67.4%	100%	99.3%	Mitochondrial complex I deficiency, nuclear type 10, 618233	VISION DISORDERS PANEL EPILEPSY PANEL INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS MITOCHONDRIAL DISORDERS PANEL COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL¹

NDUFAF3	100%	100%	100%	99.2%	Mitochondrial complex I deficiency, nuclear type 18, 618240	EPILEPSY PANEL INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS MITOCHONDRIAL DISORDERS PANEL COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL¹
NDUFAF4	100%	100%	100%	99.9%	Mitochondrial complex I deficiency, nuclear type 15, 618237	EPILEPSY PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS MITOCHONDRIAL DISORDERS PANEL COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL¹

NDUFAF5	100%	100%	100%	99.6%	Mitochondrial complex I deficiency, nuclear type 16, 618238	VISION DISORDERS PANEL EPILEPSY PANEL INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS MITOCHONDRIAL DISORDERS PANEL COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹
NDUFAF6	100%	100%	100%	99.3%	Mitochondrial complex I deficiency, nuclear type 17, 618239;Fanconi renotubular syndrome 5, 618913	MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS MITOCHONDRIAL DISORDERS PANEL COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹
NDUFAF7	100%	100%	100%	99%		MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS MITOCHONDRIAL DISORDERS PANEL

NDUFAF8	100%	100%	100%	99.1%	Mitochondrial complex I deficiency, nuclear type 34, 618776	INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS MITOCHONDRIAL DISORDERS PANEL COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹
NDUFB1	100%	100%	100%	97.7%		MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS MITOCHONDRIAL DISORDERS PANEL
NDUFB10	100%	100%	100%	99.6%	?Mitochondrial complex I deficiency, nuclear type 35, 619003	MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS MITOCHONDRIAL DISORDERS PANEL

NDUFB11	99.6%	96.9%	89.4%	53.4%	Linear skin defects with multiple congenital anomalies 3, 300952;?Mitochondrial complex I deficiency, nuclear type 30, 301021	VISION DISORDERS PANEL SKIN DISORDERS PANEL ¹ HEART DISORDERS PANEL ¹ IRON DISORDERS PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS MITOCHONDRIAL DISORDERS PANEL COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹
NDUFB2	100%	100%	100%	98.5%		MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS MITOCHONDRIAL DISORDERS PANEL

NDUFB3	100%	100%	100%	99.8%	Mitochondrial complex I deficiency, nuclear type 25, 618246	EPILEPSY PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS MITOCHONDRIAL DISORDERS PANEL COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹
NDUFB4	100%	100%	100%	98.7%		MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS MITOCHONDRIAL DISORDERS PANEL
NDUFB5	100%	100%	100%	99.3%		MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS MITOCHONDRIAL DISORDERS PANEL
NDUFB6	100%	100%	100%	99.7%		MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS MITOCHONDRIAL DISORDERS PANEL

NDUFB7	100%	100%	100%	93.8%	?Mitochondrial complex I deficiency, nuclear type 39, 620135	MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS MITOCHONDRIAL DISORDERS PANEL
NDUFB8	100%	100%	100%	99.3%	Mitochondrial complex I deficiency, nuclear type 32, 618252	MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS MITOCHONDRIAL DISORDERS PANEL COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹
NDUFB9	100%	100%	100%	99.6%	?Mitochondrial complex I deficiency, nuclear type 24, 618245	EPILEPSY PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS MITOCHONDRIAL DISORDERS PANEL COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹

NDUFC1	100%	100%	100%	99.4%		MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS MITOCHONDRIAL DISORDERS PANEL
NDUFC2	100%	100%	100%	99.4%	Mitochondrial complex I deficiency, nuclear type 36, 619170	MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS MITOCHONDRIAL DISORDERS PANEL
NDUFS1	100%	100%	100%	99.4%	Mitochondrial complex I deficiency, nuclear type 5, 618226	VISION DISORDERS PANEL EPILEPSY PANEL INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS MITOCHONDRIAL DISORDERS PANEL COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹

NDUFS2	99.8%	97.4%	100%	98.9%	?Leber-like hereditary optic neuropathy, autosomal recessive 2, 620569;Mitochondrial complex I deficiency, nuclear type 6, 618228	VISION DISORDERS PANEL EPILEPSY PANEL INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS MITOCHONDRIAL DISORDERS PANEL COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹
NDUFS3	98.6%	93%	100%	99.3%	Mitochondrial complex I deficiency, nuclear type 8, 618230	EPILEPSY PANEL INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS MITOCHONDRIAL DISORDERS PANEL COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹

NDUFS4	100%	100%	100%	99.2%	Mitochondrial complex I deficiency, nuclear type 1, 252010	EPILEPSY PANEL INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS MITOCHONDRIAL DISORDERS PANEL COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹
NDUFS5	100%	100%	100%	99.1%		MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS MITOCHONDRIAL DISORDERS PANEL

NDUFS6	100%	100%	100%	99.5%	Mitochondrial complex I deficiency, nuclear type 9, 618232	EPILEPSY PANEL POLYNEUROPATHIES PANEL ¹ INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS MITOCHONDRIAL DISORDERS PANEL COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹
NDUFS7	100%	99.9%	100%	96.4%	Mitochondrial complex I deficiency, nuclear type 3, 618224	MOVEMENT DISORDERS PANEL INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS MITOCHONDRIAL DISORDERS PANEL COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹

NDUFS8	100%	100%	100%	99.3%	Mitochondrial complex I deficiency, nuclear type 2, 618222	INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS MITOCHONDRIAL DISORDERS PANEL COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹
NDUFV1	100%	100%	100%	96.7%	Mitochondrial complex I deficiency, nuclear type 4, 618225	EPILEPSY PANEL INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS MITOCHONDRIAL DISORDERS PANEL COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹

NDUFV2	100%	100%	100%	99.5%	Mitochondrial complex I deficiency, nuclear type 7, 618229	EPILEPSY PANEL INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS MITOCHONDRIAL DISORDERS PANEL COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹
NDUFV3	100%	100%	100%	99%		MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS MITOCHONDRIAL DISORDERS PANEL
NEB	99.9%	99.8%	99.7%	98%	Nemaline myopathy 2, autosomal recessive, 256030;Arthrogryposis multiplex congenita 6, 619334	FETAL AKINESIA PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹ MUSCLE DISORDERS PANEL

NEBL	100%	100%	100%	98.9%		HEART DISORDERS PANEL ¹ MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS
NECAP1	100%	100%	100%	99.7%	Developmental and epileptic encephalopathy 21, 615833	EPILEPSY PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹
NECTIN1	93.4%	93.4%	100%	98%	Cleft lip/palate-ectodermal dysplasia syndrome, 225060;Orofacial cleft 7, 225060	CRANIOFACIAL ANOMALIES PANEL WITH GENOME WIDE CNV ANALYSIS SKIN DISORDERS PANEL ¹ MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹ OROFACIAL CLEFTING PANEL WITH GENOME WIDE CNV ANALYSIS

NECTIN4	100%	100%	100%	99%	Ectodermal dysplasia-syndactyly syndrome 1, 613573	SKIN DISORDERS PANEL ¹ MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹
NEDD4L	100%	100%	100%	99.1%	Periventricular nodular heterotopia 7, 617201	EPILEPSY PANEL INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS OROFACIAL CLEFTING PANEL WITH GENOME WIDE CNV ANALYSIS
NEFH	100%	100%	100%	98.5%	Charcot-Marie-Tooth disease, axonal, type 2CC, 616924;{?Amyotrophic lateral sclerosis, susceptibility to}, 105400	POLYNEUROPATHIES PANEL ¹ MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS MUSCLE DISORDERS PANEL

NEFL	100%	100%	100%	98.3%	Charcot-Marie-Tooth disease, type 1F, 607734;Charcot-Marie-Tooth disease, dominant intermediate G, 617882;Charcot-Marie-Tooth disease, type 2E, 607684	MOVEMENT DISORDERS PANEL POLYNEUROPATHIES PANEL ¹ MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS
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NEK1	100%	100%	100%	99.4%	Short-rib thoracic dysplasia 6 with or without polydactyly, 263520;?Orofaciodigital syndrome II, 252100;{Amyotrophic lateral sclerosis, susceptibility to, 24}, 617892	AMYOTROPHIC LATERAL SCLEROSIS, ALS PANEL VISION DISORDERS PANEL COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹ OROFACIAL CLEFTING PANEL WITH GENOME WIDE CNV ANALYSIS CILIOPATHIES PANEL DISORDERS/DIFFEREN CES OF SEX DEVELOPMENT (DSD) / PRIMARY ADRENAL INSUFFICIENCY PANEL SHORT STATURE/SKELETAL DYSPLASIA PANEL WITH GENOME WIDE CNV ANALYSIS RENAL DISORDERS PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS
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NEK10	100%	100%	100%	99.5%	Ciliary dyskinesia, primary, 44, 618781	CILIOPATHIES PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹
NEK11	100%	100%	100%	99.4%		SKIN DISORDERS PANEL ¹ MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS
NEK2	95.9%	95.9%	100%	99.3%	?Retinitis pigmentosa 67, 615565	VISION DISORDERS PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹

NEK8	100%	100%	100%	99.2%	Renal-hepatic-pancreatic dysplasia 2, 615415;Polycystic kidney disease 8, 620903;?Nephronophthisis 9, 613824	CILIOPATHIES PANEL RENAL DISORDERS PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹
NEK9	100%	100%	100%	99.5%	?Arthrogryposis, Perthes disease, and upward gaze palsy, 614262;Nevus comedonicus, somatic, 617025;Lethal congenital contracture syndrome 10, 617022	FETAL AKINESIA PANEL SKIN DISORDERS PANEL ¹ SHORT STATURE/SKELETAL DYSPLASIA PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹

NEMF	100%	100%	99.9%	98.1%	Intellectual developmental disorder with speech delay and axonal peripheral neuropathy, 619099	POLYNEUROPATHIES PANEL ¹ INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹
NEPRO	100%	100%	100%	99.3%	Anauxetic dysplasia 3, 618853	SHORT STATURE/SKELETAL DYSPLASIA PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹

NEU1	100%	100%	100%	99.3%	Sialidosis, type II, 256550;Sialidosis, type I, 256550	MOVEMENT DISORDERS PANEL EPILEPSY PANEL SHORT STATURE/SKELETAL DYSPLASIA PANEL WITH GENOME WIDE CNV ANALYSIS METABOLIC DISORDERS PANEL INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS RENAL DISORDERS PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹
NEUROD1	100%	100%	100%	99.1%	{Type 2 diabetes mellitus, susceptibility to}, 125853;Maturity-onset diabetes of the young 6, 606394	VISION DISORDERS PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS

NEUROD2	100%	100%	99.9%	94.9%	Developmental and epileptic encephalopathy 72, 618374	EPILEPSY PANEL INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS
NEUROG1	100%	100%	100%	98.8%	Cranial dysinnervation disorder, congenital, with absent corneal reflex and developmental delay, 620469	INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹
NEUROG3	100%	100%	100%	97.5%	Diarrhea 4, malabsorptive, congenital, 610370	MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹

NEXMIF	100%	100%	99.2%	70.2%	Intellectual developmental disorder, X-linked 98, 300912	MOVEMENT DISORDERS PANEL EPILEPSY PANEL INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS
NEXN	100%	100%	100%	99.5%	Cardiomyopathy, dilated, 1CC, 613122;Cardiomyopathy, hypertrophic, 20, 613876	DILATED CARDIOMYOPATHY PANEL ¹ HEART DISORDERS PANEL ¹ MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS

NF1	99.4%	99.4%	100%	99.3%	Watson syndrome, 193520;Leukemia, juvenile myelomonocytic, 607785;Neurofibromatosis, familial spinal, 162210;Neurofibromatosis, type 1, 162200;Neurofibromatosis-Noonan syndrome, 601321	INHERITED BONE MARROW FAILURE AND/OR PREDISPOSITION PANEL TO HEMATOLOGICAL MALIGNANCIES CONGENITAL HEARTDISEASE PANEL ¹ SKIN DISORDERS PANEL ¹ HEREDITARY CANCER PANEL HEART DISORDERS PANEL ¹ SHORT STATURE/SKELETAL DYSPLASIA PANEL WITH GENOME WIDE CNV ANALYSIS LYMPHATIC ANOMALIES PANEL WITH GENOME WIDE CNV ANALYSIS INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS NOONAN SYNDROME / RASOPATHY PANEL
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NF2	100%	100%	100%	99.2%	Meningioma, NF2-related, somatic, 607174;Schwannomatosis, vestibular, 101000;Schwannomatosis, somatic, 101000	MOVEMENT DISORDERS PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS HEREDITARY CANCER PANEL
NFASC	100%	100%	100%	99.1%	Neurodevelopmental disorder with central and peripheral motor dysfunction, 618356	POLYNEUROPATHIES PANEL ¹ INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹
NFAT5	100%	100%	100%	99.2%		PRIMARY IMMUNODEFICIENCIES PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS

NFATC1	100%	99.9%	100%	98.1%		PRIMARY IMMUNODEFICIENCIES PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS
NFATC2	100%	100%	100%	98.3%	?Joint contracture, osteochondromas, and B-cell lymphoma, 620232	MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS
NFE2	100%	100%	100%	98.8%		INHERITED BONE MARROW FAILURE AND/OR PREDISPOSITION PANEL TO HEMATOLOGICAL MALIGNANCIES HEMOSTATIC/THROMB OTIC DISORDERS PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS

NFE2L2	81.2%	81.2%	100%	99.2%	Immunodeficiency, developmental delay, and hypohomocysteinemia, 617744	PRIMARY IMMUNODEFICIENCIES PANEL INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS
NFIA	92.2%	92.2%	100%	97.7%	Brain malformations with or without urinary tract defects, 613735	INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS
NFIB	100%	100%	100%	99.5%	Macrocephaly, acquired, with impaired intellectual development, 618286	INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS

NFIX	99.3%	98.5%	100%	98.1%	Marshall-Smith syndrome, 602535;Malan syndrome, 614753	TALL STATURE PANEL WITH GENOME WIDE CNV ANALYSIS SHORT STATURE/SKELETAL DYSPLASIA PANEL WITH GENOME WIDE CNV ANALYSIS INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS
NFKB1	100%	100%	100%	99.2%	Immunodeficiency, common variable, 12, 616576	PRIMARY IMMUNODEFICIENCIES PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS
NFKB2	100%	100%	100%	98.7%	Immunodeficiency, common variable, 10, 615577	PRIMARY IMMUNODEFICIENCIES PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS

NFKBIA	100%	100%	100%	99.1%	Ectodermal dysplasia and immunodeficiency 2, 612132	CRANIOFACIAL ANOMALIES PANEL WITH GENOME WIDE CNV ANALYSIS SKIN DISORDERS PANEL ¹ PRIMARY IMMUNODEFICIENCIES PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS
NFS1	89.8%	89.8%	100%	98.5%	Combined oxidative phosphorylation deficiency 52, 619386	MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS MITOCHONDRIAL DISORDERS PANEL COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹

NFU1	100%	100%	100%	99.7%	Spastic paraplegia 93, autosomal recessive, 620938;Multiple mitochondrial dysfunctions syndrome 1, 605711	MOVEMENT DISORDERS PANEL INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS MITOCHONDRIAL DISORDERS PANEL COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹
NGF	100%	100%	100%	98.6%	Neuropathy, hereditary sensory and autonomic, type V, 608654	POLYNEUROPATHIES PANEL ¹ NEUROLOGICAL PAIN DISORDERS PANEL ¹ MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹

NGLY1	100%	100%	100%	99.4%	Congenital disorder of deglycosylation 1, 615273	MOVEMENT DISORDERS PANEL EPILEPSY PANEL METABOLIC DISORDERS PANEL INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS MITOCHONDRIAL DISORDERS PANEL COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL¹
NHEJ1	100%	100%	100%	98.8%	Microphthalmia/coloboma 13, 620968;Immunodeficiency 124, severe combined, 611291	VISION DISORDERS PANEL PRIMARY IMMUNODEFICIENCIES PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL¹ SEVERE COMBINED IMMUNODEFICIENCY (SCID) PANEL

NHERF1	100%	100%	100%	98.6%	Nephrolithiasis/osteoporosis, hypophosphatemic, 2, 612287	POLYNEUROPATHIES PANEL ¹ RENAL DISORDERS PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS HEARING IMPAIRMENT PANEL (INCLUDING GJB2)
NHLH2	100%	100%	100%	99.4%	?Hypogonadotropic hypogonadism 27 without anosmia, 619755	MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS
NHLRC1	100%	100%	100%	99.4%	Myoclonic epilepsy of Lafora 2, 620681	EPILEPSY PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹

NHLRC2	100%	100%	100%	99.8%	FINCA syndrome, 618278	INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹
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NHP2	100%	100%	100%	98.9%	Dyskeratosis congenita, autosomal recessive 2, 613987	INHERITED BONE MARROW FAILURE AND/OR PREDISPOSITION PANEL TO HEMATOLOGICAL MALIGNANCIES SKIN DISORDERS PANEL ¹ DYSKERATOSIS CONGENITA AND APLASTIC ANEMIA PANEL PRIMARY IMMUNODEFICIENCIES PANEL LIVER DISORDERS PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹ HEREDITARY CANCER PANEL
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NHS	100%	100%	98.8%	68.3%	Cataract 40, X-linked, 302200;Nance-Horan syndrome, 302350	VISION DISORDERS PANEL INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS
NIN	100%	100%	100%	99.3%	?Seckel syndrome 7, 614851	SHORT STATURE/SKELETAL DYSPLASIA PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹
NIPA1	100%	99.9%	100%	97.7%	Spastic paraplegia 6, autosomal dominant, 600363	MOVEMENT DISORDERS PANEL POLYNEUROPATHIES PANEL ¹ MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS

NIPAL4	100%	100%	100%	98.4%	Ichthyosis, congenital, autosomal recessive 6, 612281	SKIN DISORDERS PANEL ¹ MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹
NIPBL	100%	100%	100%	99.3%	Cornelia de Lange syndrome 1, 122470	CRANIOFACIAL ANOMALIES PANEL WITH GENOME WIDE CNV ANALYSIS SKIN DISORDERS PANEL ¹ SHORT STATURE/SKELETAL DYSPLASIA PANEL WITH GENOME WIDE CNV ANALYSIS INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS OROFACIAL CLEFTING PANEL WITH GENOME WIDE CNV ANALYSIS

NKAP	100%	99.8%	98.4%	71.2%	Intellectual developmental disorder, X-linked syndromic, Hackman-Di Donato type, 301039	TALL STATURE PANEL WITH GENOME WIDE CNV ANALYSIS INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS
NKPD1	100%	100%	100%	98.3%		SKIN DISORDERS PANEL ¹ MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS
NKX2-1	100%	100%	100%	97.8%	Chorea, hereditary benign, 118700;{Thyroid cancer, nonmedullary, 1}, 188550;Choreoathetosis, hypothyroidism, and neonatal respiratory distress, 610978	MOVEMENT DISORDERS PANEL INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS

NKX2-5	100%	100%	100%	98.9%	Hypoplastic left heart syndrome 2, 614435;Tetralogy of Fallot, 187500;Hypothyroidism, congenital nongoitrous, 5, 225250;Conotruncal heart malformations, variable, 217095;Ventricular septal defect 3, 614432;Atrial septal defect 7, with or without AV conduction defects, 108900	CONGENITAL HEARTDISEASE PANEL ¹ HEART DISORDERS PANEL ¹ MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS
NKX2-6	100%	99.9%	100%	99.1%	Persistent truncus arteriosus, 217095;Conotruncal heart malformations, 217095	CONGENITAL HEARTDISEASE PANEL ¹ HEART DISORDERS PANEL ¹ MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹
NKX3-2	100%	100%	100%	99.8%	Spondylo-megaepiphyseal-metaphyseal dysplasia, 613330	SHORT STATURE/SKELETAL DYSPLASIA PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹

NKX6-2	100%	100%	100%	95.1%	Spastic ataxia 8, autosomal recessive, with hypomyelinating leukodystrophy, 617560	MOVEMENT DISORDERS PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹
NLGN2	100%	99.9%	100%	95.9%		INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS
NLGN3	99%	97.3%	97.8%	64.7%	{Autism susceptibility, X-linked 1}, 300425	INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS

NLGN4X	100%	100%	98.5%	65.1%	Intellectual developmental disorder, X-linked, 300495;{Autism susceptibility, X-linked 2}, 300495	INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS
NLRC4	100%	100%	100%	99.6%	?Familial cold autoinflammatory syndrome 4, 616115;Autoinflammation with infantile enterocolitis, 616050	PRIMARY IMMUNODEFICIENCIES PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS
NLRP1	98.1%	98.1%	100%	99.1%	{Vitiligo-associated multiple autoimmune disease susceptibility 1}, 606579;?Respiratory papillomatosis, juvenile recurrent, congenital, 618803;Autoinflammation with arthritis and dyskeratosis, 617388;Palmoplantar carcinoma, multiple self-healing, 615225	SKIN DISORDERS PANEL ¹ PRIMARY IMMUNODEFICIENCIES PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹

NLRP12	100%	100%	100%	98.8%	Familial cold autoinflammatory syndrome 2, 611762	SKIN DISORDERS PANEL ¹ PRIMARY IMMUNODEFICIENCIES PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS
NLRP2	100%	100%	100%	99.1%	Oocyte/zygote/embryo maturation arrest 18, 620332	UNSUCCESSFUL IVF TREATMENTS, RECURRENT MOLAR PREGNANCIES AND/OR RECURRENT PREGNANCY LOSS PANEL ¹ MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS

NLRP3	100%	100%	100%	99%	CINCA syndrome, 607115;Familial cold inflammatory syndrome 1, 120100;Keratoendothelitis fugax hereditaria, 148200;Deafness, autosomal dominant 34, with or without inflammation, 617772;Muckle-Wells syndrome, 191900	SKIN DISORDERS PANEL ¹ HEARING IMPAIRMENT PANEL (INCLUDING GJB2) PRIMARY IMMUNODEFICIENCIES PANEL SHORT STATURE/SKELETAL DYSPLASIA PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS
NLRP5	100%	100%	100%	99.1%	Oocyte/zygote/embryo maturation arrest 19, 620333	UNSUCCESSFUL IVF TREATMENTS, RECURRENT MOLAR PREGNANCIES AND/OR RECURRENT PREGNANCY LOSS PANEL ¹ MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS

NLRP6	100%	100%	100%	98.9%		PRIMARY IMMUNODEFICIENCIES PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS
NLRP7	100%	100%	100%	99.2%	Hydatidiform mole, recurrent, 1, 231090	UNSUCCESSFUL IVF TREATMENTS, RECURRENT MOLAR PREGNANCIES AND/OR RECURRENT PREGNANCY LOSS PANEL ¹ MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹
NME1	100%	100%	100%	99.4%		SKIN DISORDERS PANEL ¹ MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS

NME3	100%	100%	100%	98.7%		MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS MITOCHONDRIAL DISORDERS PANEL
NME5	100%	100%	100%	99.8%	Ciliary dyskinesia, primary, 48, without situs inversus, 620032	CILIOPATHIES PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹
NME8	100%	100%	100%	99.7%	?Ciliary dyskinesia, primary, 6, 610852	CILIOPATHIES PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹

NMNAT1	99.9%	98.3%	100%	99.9%	Spondyloepiphyseal dysplasia, sensorineural hearing loss, intellectual developmental disorder, and Leber congenital amaurosis, 619260;Leber congenital amaurosis 9, 608553	VISION DISORDERS PANEL SHORT STATURE/SKELETAL DYSPLASIA PANEL WITH GENOME WIDE CNV ANALYSIS METABOLIC DISORDERS PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹
NMNAT2	100%	100%	100%	99.4%		POLYNEUROPATHIES PANEL ¹ NEUROLOGICAL PAIN DISORDERS PANEL ¹ MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS

NNT	96.3%	96.3%	100%	99.3%	Glucocorticoid deficiency 4, with or without mineralocorticoid deficiency, 614736	DISORDERS/DIFFERENCES OF SEX DEVELOPMENT (DSD) / PRIMARY ADRENAL INSUFFICIENCY PANEL METABOLIC DISORDERS PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹
NOBOX	100%	100%	100%	98.8%	Premature ovarian failure 5, 611548	DISORDERS/DIFFERENCES OF SEX DEVELOPMENT (DSD) / PRIMARY ADRENAL INSUFFICIENCY PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS PREMATURE OVARIAN INSUFFICIENCY PANEL

NOD2	100%	100%	100%	99.6%	Blau syndrome, 186580;{Yao syndrome}, 617321;{Inflammatory bowel disease 1, Crohn disease}, 266600	SKIN DISORDERS PANEL ¹ PRIMARY IMMUNODEFICIENCIES PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS
NODAL	100%	99.9%	100%	98.9%	Heterotaxy, visceral, 5, 270100	CONGENITAL HEARTDISEASE PANEL ¹ CILIOPATHIES PANEL HEART DISORDERS PANEL ¹ MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS

NOG	100%	99.9%	100%	98.6%	Symphalangism, proximal, 1A, 185800;Brachydactyly, type B2, 611377;Stapes ankylosis with broad thumbs and toes, 184460;Tarsal-carpal coalition syndrome, 186570;Multiple synostoses syndrome 1, 186500	CRANIOFACIAL ANOMALIES PANEL WITH GENOME WIDE CNV ANALYSIS HEARING IMPAIRMENT PANEL (INCLUDING GJB2) SHORT STATURE/SKELETAL DYSPLASIA PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS
NOL3	100%	100%	100%	99.6%	?Myoclonus, familial, 1, 614937	MOVEMENT DISORDERS PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS

NONO	96.3%	90.9%	99.3%	70.9%	Intellectual developmental disorder, X-linked syndromic 34, 300967	CONGENITAL HEARTDISEASE PANEL¹ HEART DISORDERS PANEL¹ INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS
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NOP10	92.5%	92.5%	100%	99.3%	?Pulmonary fibrosis and/or bone marrow failure syndrome, telomere-related, 9, 620400;?Cataracts, hearing impairment, nephrotic syndrome, and enterocolitis 2, 620425;?Dyskeratosis congenita, autosomal recessive 1, 224230	LIVER DISORDERS PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹ HEREDITARY CANCER PANEL VISION DISORDERS PANEL INHERITED BONE MARROW FAILURE AND/OR PREDISPOSITION PANEL TO HEMATOLOGICAL MALIGNANCIES SKIN DISORDERS PANEL ¹ DYSKERATOSIS CONGENITA AND APLASTIC ANEMIA PANEL PRIMARY IMMUNODEFICIENCIES PANEL
NOP56	100%	100%	100%	99.5%	Spinocerebellar ataxia 36, 614153	MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS

NOS1	100%	100%	100%	98.6%		DISORDERS/DIFFERENCES OF SEX DEVELOPMENT (DSD) / PRIMARY ADRENAL INSUFFICIENCY PANEL HYPOGONADOTROPIC HYPOGONADISM (KALLMANN) PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS
NOS1AP	100%	100%	100%	99.4%	Nephrotic syndrome, type 22, 619155	HEART DISORDERS PANEL ¹ RENAL DISORDERS PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS
NOS2	100%	99.9%	100%	99%	{Malaria, resistance to}, 611162	PRIMARY IMMUNODEFICIENCIES PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS

NOTCH1	99.2%	99%	100%	99.2%	Adams-Oliver syndrome 5, 616028;Aortic valve disease 1, 109730	THORACIC AORTIC ANEURYSM AND/OR DISSECTION PANEL CONGENITAL HEARTDISEASE PANEL ¹ SHORT STATURE/SKELETAL DYSPLASIA PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS OROFACIAL CLEFTING PANEL WITH GENOME WIDE CNV ANALYSIS SKIN DISORDERS PANEL ¹ HEART DISORDERS PANEL ¹
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NOTCH2	100%	100%	100%	99.4%	Alagille syndrome 2, 610205;Hajdu-Cheney syndrome, 102500	LIVER DISORDERS PANEL RENAL DISORDERS PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS CONGENITAL HEARTDISEASE PANEL ¹ HEART DISORDERS PANEL ¹ SHORT STATURE/SKELETAL DYSPLASIA PANEL WITH GENOME WIDE CNV ANALYSIS
NOTCH2NL C	100%	100%	100%	98.7%	Tremor, hereditary essential, 6, 618866;Oculopharyngodistal myopathy 3, 619473;Neuronal intranuclear inclusion disease, 603472	MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS
NOTCH3	100%	100%	100%	98.3%	Lateral meningocele syndrome, 130720;?Myofibromatosis, infantile 2, 615293;Cerebral arteriopathy with subcortical infarcts and leukoencephalopathy 1, 125310	MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS

NOVA2	99.5%	96.9%	98.9%	88%	Neurodevelopmental disorder with or without autistic features and/or structural brain abnormalities, 618859	INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS
NPAT	100%	100%	100%	99.7%		INHERITED BONE MARROW FAILURE AND/OR PREDISPOSITION PANEL TO HEMATOLOGICAL MALIGNANCIES MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS

NPC1	100%	100%	100%	99.2%	Niemann-Pick disease, type C1, 257220;Niemann-Pick disease, type D, 257220	MOVEMENT DISORDERS PANEL LIVER DISORDERS PANEL METABOLIC DISORDERS PANEL INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL¹
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NPC2	100%	100%	100%	99%	Niemann-pick disease, type C2, 607625	MOVEMENT DISORDERS PANEL COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL¹ LIVER DISORDERS PANEL METABOLIC DISORDERS PANEL INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS
NPHP1	100%	100%	100%	99.5%	Joubert syndrome 4, 609583;Nephronophthisis 1, juvenile, 256100;Senior-Loken syndrome-1, 266900	VISION DISORDERS PANEL COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL¹ MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS RENAL DISORDERS PANEL CILIOPATHIES PANEL

NPHP3	100%	100%	100%	99.5%	Nephronophthisis 3, 604387;Renal-hepatic-pancreatic dysplasia 1, 208540;Meckel syndrome 7, 267010	VISION DISORDERS PANEL CILIOPATHIES PANEL LIVER DISORDERS PANEL RENAL DISORDERS PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL¹
NPHP4	100%	100%	100%	99%	Senior-Loken syndrome 4, 606996;Nephronophthisis 4, 606966	VISION DISORDERS PANEL CILIOPATHIES PANEL RENAL DISORDERS PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL¹

NPHS1	100%	100%	100%	99%	Nephrotic syndrome, type 1, 256300	RENAL DISORDERS PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹
NPHS2	100%	100%	100%	99.4%	Nephrotic syndrome, type 2, 600995	RENAL DISORDERS PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹
NPL	100%	100%	100%	99.7%		METABOLIC DISORDERS PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS

NPM1	87.6%	87.6%	100%	98.8%	Leukemia, acute myeloid, somatic, 601626	INHERITED BONE MARROW FAILURE AND/OR PREDISPOSITION PANEL TO HEMATOLOGICAL MALIGNANCIES DYSKERATOSIS CONGENITA AND APLASTIC ANEMIA PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS HEREDITARY CANCER PANEL
NPPA	100%	100%	100%	98.6%	Atrial standstill 2, 615745;Atrial fibrillation, familial, 6, 612201	HEART DISORDERS PANEL ¹ MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹
NPPB	100%	100%	100%	99.7%		HEART DISORDERS PANEL ¹ MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS

NPPC	100%	100%	100%	99.7%		SHORT STATURE/SKELETAL DYSPLASIA PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS
NPR2	100%	100%	100%	99.2%	Epiphyseal chondrodysplasia, Miura type, 615923;Short stature with nonspecific skeletal abnormalities, 616255;Acromesomelic dysplasia 1, Maroteaux type, 602875	THORACIC AORTIC ANEURYSM AND/OR DISSECTION PANEL TALL STATURE PANEL WITH GENOME WIDE CNV ANALYSIS SHORT STATURE/SKELETAL DYSPLASIA PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹

NPR3	100%	100%	100%	99%	Boudin-Mortier syndrome, 619543	THORACIC AORTIC ANEURYSM AND/OR DISSECTION PANEL TALL STATURE PANEL WITH GENOME WIDE CNV ANALYSIS SHORT STATURE/SKELETAL DYSPLASIA PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹
NPRL2	100%	100%	100%	98.6%	Epilepsy, familial focal, with variable foci 2, 617116	EPILEPSY PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS
NPRL3	100%	100%	100%	98.2%	Epilepsy, familial focal, with variable foci 3, 617118	EPILEPSY PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS

NPTX1	100%	100%	100%	98.3%	Spinocerebellar ataxia 50, 620158	MOVEMENT DISORDERS PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS
NR0B1	100%	100%	98.6%	67.7%	Adrenal hypoplasia, congenital, 300200;46XY sex reversal 2, dosage-sensitive, 300018	DISORDERS/DIFFEREN CES OF SEX DEVELOPMENT (DSD) / PRIMARY ADRENAL INSUFFICIENCY PANEL HYPOGONADOTROPIC HYPOGONADISM (KALLMANN) PANEL MALE INFERTILITY PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS
NR0B2	100%	100%	100%	99.5%	Obesity, mild, early-onset, 601665	MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹

NR1H4	100%	100%	100%	98.8%	Cholestasis, progressive familial intrahepatic, 5, 617049	LIVER DISORDERS PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹
NR2E3	100%	100%	100%	98.8%	Retinitis pigmentosa 37, 611131;Enhanced S-cone syndrome, 268100	VISION DISORDERS PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹
NR2F1	100%	99.2%	100%	95.1%	Bosch-Boonstra-Schaaf optic atrophy syndrome, 615722	VISION DISORDERS PANEL EPILEPSY PANEL INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS MITOCHONDRIAL DISORDERS PANEL

NR2F2	100%	100%	100%	98.2%	46XX sex reversal 5, 618901;Congenital heart defects, multiple types, 4, 615779	CONGENITAL HEARTDISEASE PANEL ¹ DISORDERS/DIFFEREN CES OF SEX DEVELOPMENT (DSD) / PRIMARY ADRENAL INSUFFICIENCY PANEL HEART DISORDERS PANEL ¹ INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS
NR3C1	100%	100%	100%	99.7%	Glucocorticoid resistance, 615962	DISORDERS/DIFFEREN CES OF SEX DEVELOPMENT (DSD) / PRIMARY ADRENAL INSUFFICIENCY PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS

NR3C2	100%	100%	100%	98.3%	Pseudohypoaldosteronism type I, autosomal dominant, 177735;Hypertension, early-onset, autosomal dominant, with exacerbation in pregnancy, 605115	DISORDERS/DIFFERENCES OF SEX DEVELOPMENT (DSD) / PRIMARY ADRENAL INSUFFICIENCY PANEL RENAL DISORDERS PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS
NR4A2	100%	100%	100%	99.5%	Intellectual developmental disorder with language impairment and early-onset DOPA-responsive dystonia-parkinsonism, 619911	MOVEMENT DISORDERS PANEL EPILEPSY PANEL INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS
NR4A3	100%	100%	100%	97.6%	Chondrosarcoma, extraskeletal myxoid, 612237	MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS

NR5A1	100%	99.9%	100%	99.2%	46XX sex reversal 4, 617480;Premature ovarian failure 7, 612964;46XY sex reversal 3, 612965;Adrenocortical insufficiency, 612964;Spermatogenic failure 8, 613957	PREMATURE OVARIAN INSUFFICIENCY PANEL MALE INFERTILITY PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS DISORDERS/DIFFERENCES OF SEX DEVELOPMENT (DSD) / PRIMARY ADRENAL INSUFFICIENCY PANEL
NRAP	100%	100%	100%	99.4%		HEART DISORDERS PANEL ¹ MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS

NRAS	100%	100%	100%	99.2%	Noonan syndrome 6, 613224;?RAS-associated autoimmune lymphoproliferative syndrome type IV, somatic, 614470;Melanocytic nevus syndrome, congenital, somatic, 137550;Epidermal nevus, somatic, 162900;Schimmelpenning-Feuerstein-Mims syndrome, somatic mosaic, 163200;Thyroid carcinoma, follicular, somatic, 188470;Neurocutaneous melanosis, somatic, 249400;Colorectal cancer, somatic, 114500	INHERITED BONE MARROW FAILURE AND/OR PREDISPOSITION PANEL TO HEMATOLOGICAL MALIGNANCIES SKIN DISORDERS PANEL ¹ HEMOSTATIC/THROMBOTIC DISORDERS PANEL PRIMARY IMMUNODEFICIENCIES PANEL HEREDITARY CANCER PANEL SHORT STATURE/SKELETAL DYSPLASIA PANEL WITH GENOME WIDE CNV ANALYSIS LYMPHATIC ANOMALIES PANEL WITH GENOME WIDE CNV ANALYSIS INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS NOONAN SYNDROME /
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						RASOPATHY PANEL
NRCAM	100%	100%	100%	99.4%	Neurodevelopmental disorder with neuromuscular and skeletal abnormalities, 619833	INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹
NRG1	100%	99.7%	100%	99.3%	{?Schizophrenia, susceptibility to}, 603013	POLYNEUROPATHIES PANEL ¹ MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS
NRIP1	100%	100%	100%	99.6%	?Congenital anomalies of kidney and urinary tract 3, 618270	MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS
NRL	100%	100%	100%	99%	Retinitis pigmentosa 27, 613750;Retinal degeneration, autosomal recessive, clumped pigment type	VISION DISORDERS PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS

NRROS	100%	100%	100%	99.1%	Seizures, early-onset, with neurodegeneration and brain calcification, 618875	EPILEPSY PANEL INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS MITOCHONDRIAL DISORDERS PANEL COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL¹
NRXN1	100%	100%	100%	99%	Pitt-Hopkins-like syndrome 2, 614325;{Schizophrenia, susceptibility to, 17}, 614332	EPILEPSY PANEL INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL¹

NSD1	100%	100%	100%	99.5%	Sotos syndrome, 117550	METABOLIC DISORDERS PANEL INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS HEREDITARY CANCER PANEL CRANIOFACIAL ANOMALIES PANEL WITH GENOME WIDE CNV ANALYSIS CONGENITAL HEARTDISEASE PANEL¹ SKIN DISORDERS PANEL¹ TALL STATURE PANEL WITH GENOME WIDE CNV ANALYSIS HEART DISORDERS PANEL¹ SHORT STATURE/SKELETAL DYSPLASIA PANEL WITH GENOME WIDE CNV ANALYSIS
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NSD2	99.5%	99.4%	100%	99%	Rauch-Steindl syndrome, 619695	SHORT STATURE/SKELETAL DYSPLASIA PANEL WITH GENOME WIDE CNV ANALYSIS INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS
NSDHL	100%	99.6%	99.5%	70.1%	CK syndrome, 300831;CHILD syndrome, 308050	SKIN DISORDERS PANEL ¹ SHORT STATURE/SKELETAL DYSPLASIA PANEL WITH GENOME WIDE CNV ANALYSIS METABOLIC DISORDERS PANEL INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS

NSF	100%	100%	99.9%	95.2%	Developmental and epileptic encephalopathy 96, 619340	INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS
NSMCE2	100%	100%	100%	99.6%	Seckel syndrome 10, 617253	SHORT STATURE/SKELETAL DYSPLASIA PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹
NSMCE3	100%	100%	100%	98.3%	Lung disease, immunodeficiency, and chromosome breakage syndrome, 617241	PRIMARY IMMUNODEFICIENCIES PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹

NSMF	100%	100%	100%	98.5%	Hypogonadotropic hypogonadism 9 with or without anosmia, 614838	DISORDERS/DIFFERENCES OF SEX DEVELOPMENT (DSD) / PRIMARY ADRENAL INSUFFICIENCY PANEL HYPOGONADOTROPIC HYPOGONADISM (KALLMANN) PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS
NSRP1	91%	91%	100%	98.8%	Neurodevelopmental disorder with spasticity, seizures, and brain abnormalities, 620001	INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹

NSUN2	100%	100%	100%	99.1%	Intellectual developmental disorder, autosomal recessive 5, 611091	INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹
NSUN3	100%	100%	100%	99.5%	Combined oxidative phosphorylation deficiency 48, 619012	VISION DISORDERS PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS MITOCHONDRIAL DISORDERS PANEL COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹

NSUN6	100%	100%	100%	99.4%	Intellectual developmental disorder, autosomal recessive 82, 620779	INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL¹
NT5C2	100%	100%	100%	99.9%	Spastic paraplegia 45, autosomal recessive, 613162	MOVEMENT DISORDERS PANEL INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL¹

NT5C3A	100%	100%	100%	99.3%	Anemia, congenital, nonspherocytic hemolytic, 8, 266120	METABOLIC DISORDERS PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹
NT5E	100%	100%	100%	99.2%	Calcification of joints and arteries, 211800	METABOLIC DISORDERS PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹
NTF4	100%	100%	100%	99%	Glaucoma 1, open angle, 1O, 613100	MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS

NTHL1	100%	99.9%	100%	99.1%	Familial adenomatous polyposis 3, 616415	PANEL HEREDITARY COLORECTAL AND POLYPOSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹ HEREDITARY CANCER PANEL
NTN1	100%	99.9%	100%	99.4%	Mirror movements 4, 618264	DISORDERS/DIFFERENCES OF SEX DEVELOPMENT (DSD) / PRIMARY ADRENAL INSUFFICIENCY PANEL HYPOGONADOTROPIC HYPOGONADISM (KALLMANN) PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS

NTNG2	100%	100%	100%	98.2%	Neurodevelopmental disorder with behavioral abnormalities, absent speech, and hypotonia, 618718	INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹
NTRK1	100%	99.9%	100%	98.6%	Insensitivity to pain, congenital, with anhidrosis, 256800	POLYNEUROPATHIES PANEL ¹ NEUROLOGICAL PAIN DISORDERS PANEL ¹ INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹

NTRK2	89.5%	89.5%	100%	99.1%	Developmental and epileptic encephalopathy 58, 617830;Obesity, hyperphagia, and developmental delay, 613886	INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS
NUAK2	100%	100%	100%	98.6%	?Anencephaly 2, 619452	MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS
NUBPL	100%	100%	100%	99.7%	Mitochondrial complex I deficiency, nuclear type 21, 618242	EPILEPSY PANEL INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS MITOCHONDRIAL DISORDERS PANEL COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹

NUDCD3	100%	100%	100%	99.1%		PRIMARY IMMUNODEFICIENCIES PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS SEVERE COMBINED IMMUNODEFICIENCY (SCID) PANEL
NUDT2	100%	100%	100%	99.8%	Intellectual developmental disorder with or without peripheral neuropathy, 619844	INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL¹
NUMA1	100%	100%	100%	99.2%		MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS

NUP107	100%	100%	100%	99.5%	?Ovarian dysgenesis 6, 618078;Galloway-Mowat syndrome 7, 618348;Nephrotic syndrome, type 11, 616730	INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS RENAL DISORDERS PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹
NUP133	100%	100%	100%	99.4%	?Galloway-Mowat syndrome 8, 618349;Nephrotic syndrome, type 18, 618177	INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS RENAL DISORDERS PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹
NUP155	100%	100%	100%	99.5%	?Atrial fibrillation 15, 615770	HEART DISORDERS PANEL ¹ MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS

NUP160	100%	100%	100%	99.5%	?Nephrotic syndrome, type 19, 618178	RENAL DISORDERS PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹
NUP188	100%	100%	100%	99.3%	Sandestig-Stefanova syndrome, 618804	INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹
NUP205	100%	100%	100%	99.4%	?Nephrotic syndrome, type 13, 616893	RENAL DISORDERS PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹

NUP214	100%	100%	100%	99.3%	Leukemia, T-cell acute lymphoblastic, somatic, 613065;Leukemia, acute myeloid, somatic, 601626;{Encephalopathy, acute, infection-induced, susceptibility to, 9}, 618426	INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹
NUP37	100%	100%	100%	99.7%	?Microcephaly 24, primary, autosomal recessive, 618179	MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹
NUP54	100%	100%	100%	99.7%	Dystonia 37, early-onset, with striatal lesions, 620427	INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS MITOCHONDRIAL DISORDERS PANEL COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹

NUP62	100%	100%	100%	98.5%	Striatonigral degeneration, infantile, 271930	MOVEMENT DISORDERS PANEL INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL¹
NUP85	100%	100%	100%	98.9%	Nephrotic syndrome, type 17, 618176	INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS RENAL DISORDERS PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL¹

NUP88	93.4%	93.4%	100%	99.5%	Fetal akinesia deformation sequence 4, 618393	FETAL AKINESIA PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹
NUP93	95.5%	95.5%	100%	99.4%	Nephrotic syndrome, type 12, 616892	RENAL DISORDERS PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹

NUS1	100%	100%	100%	99.5%	Intellectual developmental disorder, autosomal dominant 55, with seizures, 617831;?Congenital disorder of glycosylation, type 1aa, 617082	MOVEMENT DISORDERS PANEL EPILEPSY PANEL METABOLIC DISORDERS PANEL INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL¹
NUTF2	97.7%	96.8%	100%	98.5%		MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS MITOCHONDRIAL DISORDERS PANEL
NUTM2B- AS1					?Oculopharyngeal myopathy with leukoencephalopathy 1, 618637	MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS

NXF5						RENAL DISORDERS PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS
NXN	100%	99.3%	100%	98%	Robinow syndrome, autosomal recessive 2, 618529	SHORT STATURE/SKELETAL DYSPLASIA PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹
NYNRIN	100%	100%	100%	99%		MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS HEREDITARY CANCER PANEL
NYX	100%	100%	97.8%	63.1%	Night blindness, congenital stationary (complete), 1A, X-linked, 310500	VISION DISORDERS PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS

OAS1	100%	100%	100%	98.9%	Immunodeficiency 100 with pulmonary alveolar proteinosis and hypogammaglobulinemia, 618042	PRIMARY IMMUNODEFICIENCIES PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS
OAT	100%	100%	100%	98.7%	Gyrate atrophy of choroid and retina with or without ornithinemia, 258870	VISION DISORDERS PANEL METABOLIC DISORDERS PANEL INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL¹

OBSL1	100%	100%	100%	98.7%	3-M syndrome 2, 612921	DISORDERS/DIFFERENCES OF SEX DEVELOPMENT (DSD) / PRIMARY ADRENAL INSUFFICIENCY PANEL SHORT STATURE/SKELETAL DYSPLASIA PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹
OCA2	100%	100%	100%	99.3%	[Skin/hair/eye pigmentation 1, blue/nonblue eyes], 227220;[Skin/hair/eye pigmentation 1, blond/brown hair], 227220;Albinism, brown oculocutaneous, 203200;Albinism, oculocutaneous, type II, 203200	VISION DISORDERS PANEL SKIN DISORDERS PANEL ¹ MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹

OCLN	94.5%	94.5%	100%	97.6%	Pseudo-TORCH syndrome 1, 251290	MOVEMENT DISORDERS PANEL COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹ EPILEPSY PANEL INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS
OCRL	100%	99.9%	98.7%	70.5%	Dent disease 2, 300555;Lowe syndrome, 309000	VISION DISORDERS PANEL INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS RENAL DISORDERS PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS CILIOPATHIES PANEL HEMOSTATIC/THROMB OTIC DISORDERS PANEL METABOLIC DISORDERS PANEL

ODAD1	100%	100%	100%	98.7%	Ciliary dyskinesia, primary, 20, 615067	COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹ CONGENITAL HEARTDISEASE PANEL ¹ CILIOPATHIES PANEL HEART DISORDERS PANEL ¹ MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS
ODAD2	96.1%	96.1%	100%	99.5%	Ciliary dyskinesia, primary, 23, 615451	CILIOPATHIES PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹
ODAD3	100%	100%	100%	98.3%	Ciliary dyskinesia, primary, 30, 616037	COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹ CILIOPATHIES PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS

ODAD4	100%	100%	100%	98.7%	Ciliary dyskinesia, primary, 35, 617092	CILIOPATHIES PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹
ODAM	100%	100%	100%	99.6%		SKIN DISORDERS PANEL ¹ MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS
ODAPH	100%	100%	100%	99.8%	Amelogenesis imperfecta, type IIA4, 614832	CRANIOFACIAL ANOMALIES PANEL WITH GENOME WIDE CNV ANALYSIS SKIN DISORDERS PANEL ¹ MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹

ODC1	100%	100%	100%	99%	Bachmann-Bupp syndrome, 619075	METABOLIC DISORDERS PANEL INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS
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OFD1	100%	99.9%	98.8%	70.7%	Simpson-Golabi-Behmel syndrome, type 2, 300209;?Retinitis pigmentosa 23, 300424;Orofaciodigital syndrome I, 311200;Joubert syndrome 10, 300804	OROFACIAL CLEFTING PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS RENAL DISORDERS PANEL INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS SHORT STATURE/SKELETAL DYSPLASIA PANEL WITH GENOME WIDE CNV ANALYSIS EPILEPSY PANEL CILIOPATHIES PANEL SKIN DISORDERS PANEL ¹ VISION DISORDERS PANEL CRANIOFACIAL ANOMALIES PANEL WITH GENOME WIDE CNV ANALYSIS
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OGDH	100%	100%	100%	99.1%	Oxoglutarate dehydrogenase deficiency, 203740	METABOLIC DISORDERS PANEL INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS MITOCHONDRIAL DISORDERS PANEL COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹
OGDHL	100%	100%	100%	99.2%	Yoon-Bellen neurodevelopmental syndrome, 619701	MOVEMENT DISORDERS PANEL EPILEPSY PANEL METABOLIC DISORDERS PANEL INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS MITOCHONDRIAL DISORDERS PANEL COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹

OGG1	100%	100%	100%	98.9%	Renal cell carcinoma, clear cell, somatic, 144700	MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS
OGT	100%	99.8%	99.1%	72.6%	Intellectual developmental disorder, X-linked 106, 300997	INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS
OOEP	100%	100%	100%	99%		UNSUCCESSFUL IVF TREATMENTS, RECURRENT MOLAR PREGNANCIES AND/OR RECURRENT PREGNANCY LOSS PANEL ¹ MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS

OPA1	100%	100%	100%	99.8%	Optic atrophy plus syndrome, 125250;{Glaucoma, normal tension, susceptibility to}, 606657;Optic atrophy 1, 165500;Behr syndrome, 210000;?Mitochondrial DNA depletion syndrome 14 (encephalocardiomyopathic type), 616896	MOVEMENT DISORDERS PANEL VISION DISORDERS PANEL HEARING IMPAIRMENT PANEL (INCLUDING GJB2) MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS MITOCHONDRIAL DISORDERS PANEL COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL¹ MUSCLE DISORDERS PANEL
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OPA3	100%	100%	100%	99.4%	3-methylglutaconic aciduria, type III, 258501;Optic atrophy 3 with cataract, 165300	VISION DISORDERS PANEL METABOLIC DISORDERS PANEL COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹ INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS MITOCHONDRIAL DISORDERS PANEL
OPCML	100%	100%	100%	99.5%	Ovarian cancer, somatic, 167000	MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS
OPHN1	94%	93.9%	99%	69.4%	Intellectual developmental disorder, X-linked syndromic, Billuart type, 300486	MOVEMENT DISORDERS PANEL EPILEPSY PANEL INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS

OPLAH	100%	100%	100%	98.6%	5-oxoprolinase deficiency, 260005	METABOLIC DISORDERS PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹
OPN1LW	97.4%	95.3%	96.9%	66.2%	Blue cone monochromacy, 303700;Colorblindness, protan, 303900	VISION DISORDERS PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS
OPN1MW	95.9%	88.6%	87.7%	53.1%	Colorblindness, deutan, 303800;Blue cone monochromacy, 303700	VISION DISORDERS PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS
OPN1SW	100%	100%	100%	99%	Colorblindness, tritan, 190900	MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS

OPTN	100%	100%	100%	99.1%	Glaucoma 1, open angle, E, 137760;Amyotrophic lateral sclerosis 12 with or without frontotemporal dementia, 613435;{Glaucoma, normal tension, susceptibility to}, 606657	AMYOTROPHIC LATERAL SCLEROSIS, ALS PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS
ORAI1	100%	100%	100%	97.4%	Immunodeficiency 9, 612782;Myopathy, tubular aggregate, 2, 615883	HEMOSTATIC/THROMB OTIC DISORDERS PANEL PRIMARY IMMUNODEFICIENCIES PANEL MUSCLE DISORDERS PANEL COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL¹ MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS

ORC1	100%	100%	100%	99%	Meier-Gorlin syndrome 1, 224690	SHORT STATURE/SKELETAL DYSPLASIA PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹ INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS OROFACIAL CLEFTING PANEL WITH GENOME WIDE CNV ANALYSIS
ORC4	100%	100%	100%	99.6%	Meier-Gorlin syndrome 2, 613800	SHORT STATURE/SKELETAL DYSPLASIA PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹

ORC6	100%	100%	100%	99.7%	Meier-Gorlin syndrome 3, 613803	COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹ SHORT STATURE/SKELETAL DYSPLASIA PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS
OSBPL2	100%	100%	100%	99.3%	Deafness, autosomal dominant 67, 616340	HEARING IMPAIRMENT PANEL (INCLUDING GJB2) MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS
OSGEP	100%	100%	100%	99.5%	Galloway-Mowat syndrome 3, 617729	INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS RENAL DISORDERS PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹

OSMR	100%	100%	100%	99.7%	Amyloidosis, primary localized cutaneous, 1, 105250	SKIN DISORDERS PANEL ¹ MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS
OSTC	84.7%	84.7%	100%	98.1%		METABOLIC DISORDERS PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS

OSTM1	99.9%	98.9%	100%	99.7%	Osteopetrosis, autosomal recessive 5, 259720	INHERITED BONE MARROW FAILURE AND/OR PREDISPOSITION PANEL TO HEMATOLOGICAL MALIGNANCIES PRIMARY IMMUNODEFICIENCIES PANEL SHORT STATURE/SKELETAL DYSPLASIA PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹
OTC	100%	99.9%	98.9%	71%	Ornithine transcarbamylase deficiency, 311250	METABOLIC DISORDERS PANEL INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS

OTOA	100%	100%	100%	98.3%	Deafness, autosomal recessive 22, 607039	HEARING IMPAIRMENT PANEL (INCLUDING GJB2) MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹
OTOF	100%	100%	100%	98.7%	Auditory neuropathy, autosomal recessive, 1, 601071;Deafness, autosomal recessive 9, 601071	HEARING IMPAIRMENT PANEL (INCLUDING GJB2) MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹
OTOG	100%	100%	100%	98.8%	Deafness, autosomal recessive 18B, 614945	HEARING IMPAIRMENT PANEL (INCLUDING GJB2) MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹

OTOGL	100%	100%	100%	99.1%	Deafness, autosomal recessive 84B, 614944	HEARING IMPAIRMENT PANEL (INCLUDING GJB2) MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹
OTUD5	100%	98.8%	97.4%	64.7%	Multiple congenital anomalies-neurodevelopmental syndrome, X-linked, 301056	INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS
OTUD6B	100%	100%	100%	99.6%	Intellectual developmental disorder with dysmorphic facies, seizures, and distal limb anomalies, 617452	INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹

OTUD7A	97.9%	94.6%	100%	96.9%	Neurodevelopmental disorder with hypotonia and seizures, 620790	INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL¹
OTULIN	100%	99.9%	100%	99.2%	Autoinflammation, panniculitis, and dermatosis syndrome, 617099;{Immunodeficiency 107, susceptibility to invasive staphylococcus aureus infection}, 619986	PRIMARY IMMUNODEFICIENCIES PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL¹

OTX2	100%	100%	100%	99.3%	Retinal dystrophy, early-onset, with or without pituitary dysfunction, 610125;Pituitary hormone deficiency, combined, 6, 613986;Microphthalmia, syndromic 5, 610125	VISION DISORDERS PANEL CRANIOFACIAL ANOMALIES PANEL WITH GENOME WIDE CNV ANALYSIS SHORT STATURE/SKELETAL DYSPLASIA PANEL WITH GENOME WIDE CNV ANALYSIS INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS MITOCHONDRIAL DISORDERS PANEL
OVOL2	100%	99.8%	100%	98.7%	Corneal dystrophy, posterior polymorphous, 1, 122000	VISION DISORDERS PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS

OXA1L	100%	100%	99.9%	99.7%		MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS MITOCHONDRIAL DISORDERS PANEL
OXCT1	100%	100%	100%	99.4%	Succinyl CoA:3-oxoacid CoA transferase deficiency, 245050	METABOLIC DISORDERS PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹
OXGR1	100%	100%	100%	99.7%	Nephrolithiasis, calcium oxalate, 2, with nephrocalcinosis, 620374	RENAL DISORDERS PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS
OXR1	100%	100%	100%	99.6%	Cerebellar hypoplasia/atrophy, epilepsy, and global developmental delay, 213000	INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS

P2RX2	100%	100%	99.9%	97%	Deafness, autosomal dominant 41, 608224	HEARING IMPAIRMENT PANEL (INCLUDING GJB2) MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS
P2RY12	100%	100%	100%	98.5%	Bleeding disorder, platelet-type, 8, 609821	HEMOSTATIC/THROMB OTIC DISORDERS PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹
P3H1	100%	100%	100%	99.2%	Osteogenesis imperfecta, type VIII, 610915	MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹ VISION DISORDERS PANEL SHORT STATURE/SKELETAL DYSPLASIA PANEL WITH GENOME WIDE CNV ANALYSIS

P3H2	100%	100%	100%	99.1%	Myopia, high, with cataract and vitreoretinal degeneration, 614292	VISION DISORDERS PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹
P4HA2	100%	100%	100%	99.5%	Myopia 25, autosomal dominant, 617238	VISION DISORDERS PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS
P4HB	96%	94.7%	100%	99.2%	Cole-Carpenter syndrome 1, 112240	CRANIOFACIAL ANOMALIES PANEL WITH GENOME WIDE CNV ANALYSIS SHORT STATURE/SKELETAL DYSPLASIA PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS

P4HTM	100%	100%	100%	98.8%	Hypotonia, hypoventilation, impaired intellectual development, dysautonomia, epilepsy, and eye abnormalities, 618493	INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS MITOCHONDRIAL DISORDERS PANEL COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹
PABPC1	98.7%	98.6%	100%	99.6%		INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS
PABPC1L	100%	100%	100%	98.4%		UNSUCCESSFUL IVF TREATMENTS, RECURRENT MOLAR PREGNANCIES AND/OR RECURRENT PREGNANCY LOSS PANEL ¹ MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS

PABPN1	100%	100%	100%	97.1%	Oculopharyngeal muscular dystrophy, 164300	MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS MUSCLE DISORDERS PANEL
PACS1	100%	100%	100%	98.4%	Schuurs-Hoeijmakers syndrome, 615009	EPILEPSY PANEL INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS
PACS2	100%	99.8%	100%	98.8%	Developmental and epileptic encephalopathy 66, 618067	MOVEMENT DISORDERS PANEL EPILEPSY PANEL INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS

PADI3	100%	100%	100%	98.7%	Uncombable hair syndrome, 191480	SKIN DISORDERS PANEL ¹ MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS
PADI6	100%	99.9%	99.4%	95.7%	Oocyte/zygote/embryo maturation arrest 16, 617234	UNSUCCESSFUL IVF TREATMENTS, RECURRENT MOLAR PREGNANCIES AND/OR RECURRENT PREGNANCY LOSS PANEL ¹ MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS
PAFAH1B1	100%	100%	100%	99.5%	Subcortical laminar heterotopia, 607432;Lissencephaly 1, 607432	EPILEPSY PANEL INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS

PAH	100%	100%	100%	99.1%	[Hyperphenylalaninemia, non-PKU mild], 261600;Phenylketonuria, 261600	SKIN DISORDERS PANEL ¹ METABOLIC DISORDERS PANEL INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹
PAICS	100%	100%	100%	99.2%	?Phosphoribosylaminoimidazole carboxylase deficiency, 619859	MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS
PAK1	100%	100%	100%	99.6%	Intellectual developmental disorder with macrocephaly, seizures, and speech delay, 618158	INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS

PAK2	100%	100%	100%	99.5%	?Knobloch syndrome 2, 618458	VISION DISORDERS PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS
PAK3	100%	100%	99%	70.5%	Intellectual developmental disorder, X-linked 30, 300558	EPILEPSY PANEL INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS

PALB2	100%	100%	100%	99.4%	{Breast-ovarian cancer, familial, susceptibility to, 5}, 620442;{Pancreatic cancer, susceptibility to, 3}, 613348;Fanconi anemia, complementation group N, 610832	INHERITED BONE MARROW FAILURE AND/OR PREDISPOSITION PANEL TO HEMATOLOGICAL MALIGNANCIES SKIN DISORDERS PANEL ¹ MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS PANEL HEREDITARY PANCREATIC CANCER (BRCA1, BRCA2, PALB2, ATM, CDKN2A, MLH1, MSH2, MSH6, PMS2, STK11) SONIC HEDGEHOG MEDULLOBLASTOMA PANEL HEREDITARY CANCER PANEL
PALS1	100%	100%	100%	99.4%		INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS

PAM16	84.8%	84.5%	100%	98.4%	Spondylometaphyseal dysplasia, Megarbane-Dagher-Melike type, 613320	SHORT STATURE/SKELETAL DYSPLASIA PANEL WITH GENOME WIDE CNV ANALYSIS INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹
PAN2	100%	100%	100%	99.4%		INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS

PANK2	100%	100%	100%	99.2%	Neurodegeneration with brain iron accumulation 1, 234200	MOVEMENT DISORDERS PANEL IRON DISORDERS PANEL METABOLIC DISORDERS PANEL INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS VISION DISORDERS PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS MITOCHONDRIAL DISORDERS PANEL COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹
PANK4	100%	99.9%	100%	98.1%	?Cataract 49, 619593	VISION DISORDERS PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS

PANX1	100%	100%	100%	99.3%	Oocyte/zygote/embryo maturation arrest 7, 618550	UNSUCCESSFUL IVF TREATMENTS, RECURRENT MOLAR PREGNANCIES AND/OR RECURRENT PREGNANCY LOSS PANEL ¹ INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS
PAPPA2	100%	100%	100%	99.3%	Short stature, Dauber-Argente type, 619489	SHORT STATURE/SKELETAL DYSPLASIA PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS

PAPSS2	100%	100%	100%	99%	Brachyolmia 4 with mild epiphyseal and metaphyseal changes, 612847	SHORT STATURE/SKELETAL DYSPLASIA PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹
PARK7	100%	100%	100%	99.7%	Parkinson disease 7, autosomal recessive early-onset, 606324	MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS PARKINSON DISEASE PANEL COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹

PARN	97%	95.3%	100%	99.5%	Dyskeratosis congenita, autosomal recessive 6, 616353;Pulmonary fibrosis and/or bone marrow failure syndrome, telomere-related, 4, 616371	INHERITED BONE MARROW FAILURE AND/OR PREDISPOSITION PANEL TO HEMATOLOGICAL MALIGNANCIES DYSKERATOSIS CONGENITA AND APLASTIC ANEMIA PANEL PRIMARY IMMUNODEFICIENCIES PANEL INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹ HEREDITARY CANCER PANEL
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PARP4	100%	100%	100%	99.4%		INHERITED BONE MARROW FAILURE AND/OR PREDISPOSITION PANEL TO HEMATOLOGICAL MALIGNANCIES MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS
PARP6	100%	100%	100%	99.4%		INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS

PARS2	100%	100%	100%	99.6%	Developmental and epileptic encephalopathy 75, 618437	EPILEPSY PANEL INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS MITOCHONDRIAL DISORDERS PANEL COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹
PATL2	100%	100%	100%	99.3%	Oocyte/zygote/embryo maturation arrest 4, 617743	UNSUCCESSFUL IVF TREATMENTS, RECURRENT MOLAR PREGNANCIES AND/OR RECURRENT PREGNANCY LOSS PANEL ¹ MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹

PAX1	100%	100%	100%	96.9%	Otofaciocervical syndrome 2 with T-cell deficiency, 615560	PRIMARY IMMUNODEFICIENCIES PANEL COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL¹ INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS SEVERE COMBINED IMMUNODEFICIENCY (SCID) PANEL
PAX2	100%	100%	100%	98.9%	Glomerulosclerosis, focal segmental, 7, 616002;Papillorenal syndrome, 120330	VISION DISORDERS PANEL RENAL DISORDERS PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS

PAX3	100%	100%	100%	98.5%	Craniofacial-deafness-hand syndrome, 122880;Waardenburg syndrome, type 3, 148820;Waardenburg syndrome, type 1, 193500;Rhabdomyosarcoma 2, alveolar, 268220	CRANIOFACIAL ANOMALIES PANEL WITH GENOME WIDE CNV ANALYSIS SKIN DISORDERS PANEL ¹ HEARING IMPAIRMENT PANEL (INCLUDING GJB2) SHORT STATURE/SKELETAL DYSPLASIA PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹ OROFACIAL CLEFTING PANEL WITH GENOME WIDE CNV ANALYSIS
PAX4	100%	100%	100%	99.7%	{Diabetes mellitus, ketosis-prone, susceptibility to}, 612227;Maturity-onset diabetes of the young, type IX, 612225;Diabetes mellitus, type 2, 125853	MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS

PAX5	100%	100%	100%	99.1%	{Leukemia, acute lymphoblastic, susceptibility to, 3}, 615545	INHERITED BONE MARROW FAILURE AND/OR PREDISPOSITION PANEL TO HEMATOLOGICAL MALIGNANCIES HEREDITARY CANCER PANEL PRIMARY IMMUNODEFICIENCIES PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS
PAX6	100%	100%	100%	99%	Optic nerve hypoplasia, 165550;Cataract with late-onset corneal dystrophy, 106210;Microphthalmia/coloboma 12, 120200;?Coloboma of optic nerve, 120430;Aniridia, 106210;Anterior segment dysgenesis 5, multiple subtypes, 604229;?Morning glory disc anomaly, 120430;Foveal hypoplasia 1, 136520;Keratitis, 148190	MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS MOVEMENT DISORDERS PANEL VISION DISORDERS PANEL CRANIOFACIAL ANOMALIES PANEL WITH GENOME WIDE CNV ANALYSIS

PAX7	100%	100%	100%	98.4%	Congenital myopathy 19, 618578;Rhabdomyosarcoma 2, alveolar, 268220	CRANIOFACIAL ANOMALIES PANEL WITH GENOME WIDE CNV ANALYSIS INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹ MUSCLE DISORDERS PANEL
PAX8	100%	100%	100%	98.6%	Hypothyroidism, congenital, due to thyroid dysgenesis or hypoplasia, 218700	INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS

PAX9	100%	100%	100%	98.9%	Tooth agenesis, selective, 3, 604625	CRANIOFACIAL ANOMALIES PANEL WITH GENOME WIDE CNV ANALYSIS SKIN DISORDERS PANEL ¹ MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS
PBRM1	100%	100%	100%	99.3%	?Renal cell carcinoma, clear cell, 144700	MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS

PBX1	100%	100%	100%	99%	Congenital anomalies of kidney and urinary tract syndrome with or without hearing loss, abnormal ears, or developmental delay, 617641	HEARING IMPAIRMENT PANEL (INCLUDING GJB2) DISORDERS/DIFFEREN CES OF SEX DEVELOPMENT (DSD) / PRIMARY ADRENAL INSUFFICIENCY PANEL PRIMARY IMMUNODEFICIENCIES PANEL INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS RENAL DISORDERS PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS
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PC	100%	100%	100%	99%	Pyruvate carboxylase deficiency, 266150	EPILEPSY PANEL METABOLIC DISORDERS PANEL INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS MITOCHONDRIAL DISORDERS PANEL COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹
PCARE	100%	99.9%	100%	99.4%	Retinitis pigmentosa 54, 613428	VISION DISORDERS PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹

PCBD1	100%	100%	100%	99.4%	Hyperphenylalaninemia, BH4-deficient, D, 264070	METABOLIC DISORDERS PANEL RENAL DISORDERS PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹
PCCA	100%	100%	100%	99.4%	Propionicacidemia, 606054	HEART DISORDERS PANEL ¹ PRIMARY IMMUNODEFICIENCIES PANEL METABOLIC DISORDERS PANEL INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹

PCCB	99.6%	96.7%	100%	99.4%	Propionicacidemia, 606054	HEART DISORDERS PANEL ¹ PRIMARY IMMUNODEFICIENCIES PANEL METABOLIC DISORDERS PANEL INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹
PCDH12	100%	100%	100%	99.2%	Diencephalic-mesencephalic junction dysplasia syndrome 1, 251280	INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹

PCDH15	100%	100%	100%	99.4%	Usher syndrome, type 1D/F digenic, 601067;Deafness, autosomal recessive 23, 609533;Usher syndrome, type 1F, 602083	VISION DISORDERS PANEL HEARING IMPAIRMENT PANEL (INCLUDING GJB2) MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹
PCDH19	100%	99.9%	98.1%	66.6%	Developmental and epileptic encephalopathy 9, 300088	EPILEPSY PANEL INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS
PCDHGC4	100%	100%	100%	99.1%	Neurodevelopmental disorder with poor growth and skeletal anomalies, 619880	INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹

PCGF2	100%	100%	100%	97.9%	Turnpenny-Fry syndrome, 618371	INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS
PCK1	100%	100%	100%	99%	Phosphoenolpyruvate carboxykinase deficiency, cytosolic, 261680	METABOLIC DISORDERS PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹
PCK2	100%	100%	100%	99%	PEPCK deficiency, mitochondrial, 261650	METABOLIC DISORDERS PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹

PCLO	100%	100%	100%	98.2%	?Pontocerebellar hypoplasia, type 3, 608027	INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹
PCNA	100%	100%	100%	99.7%	?Ataxia-telangiectasia-like disorder 2, 615919	SKIN DISORDERS PANEL ¹ MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS

PCNT	100%	100%	100%	99.1%	Microcephalic osteodysplastic primordial dwarfism, type II, 210720	SHORT STATURE/SKELETAL DYSPLASIA PANEL WITH GENOME WIDE CNV ANALYSIS INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹
PCSK1	100%	100%	100%	99.4%	{Obesity, susceptibility to, BMIQ12}, 612362;Endocrinopathy due to proprotein convertase 1/3 deficiency, 600955	DISORDERS/DIFFEREN CES OF SEX DEVELOPMENT (DSD) / PRIMARY ADRENAL INSUFFICIENCY PANEL HYPOGONADOTROPIC HYPOGONADISM (KALLMANN) PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹

PCSK9	100%	100%	100%	98.8%	{Low density lipoprotein cholesterol level QTL 1}, 603776;Hypercholesterolemia, familial, 3, 603776	MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS
PCYT1A	100%	100%	100%	99.7%	Spondylometaphyseal dysplasia with cone-rod dystrophy, 608940;Lipodystrophy, congenital generalized, type 5, 620680	VISION DISORDERS PANEL SHORT STATURE/SKELETAL DYSPLASIA PANEL WITH GENOME WIDE CNV ANALYSIS METABOLIC DISORDERS PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL¹

PCYT2	100%	99.5%	100%	98.8%	Spastic paraplegia 82, autosomal recessive, 618770	MOVEMENT DISORDERS PANEL METABOLIC DISORDERS PANEL INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹
PDCD1	100%	100%	100%	98.9%	?Autoimmune disease with susceptibility to mycobacterium tuberculosis, 621004	PRIMARY IMMUNODEFICIENCIES PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS
PDCD10	100%	100%	100%	99.4%	Cerebral cavernous malformations-3, 603285	EPILEPSY PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS

PDCD6IP	100%	100%	100%	99.5%	?Microcephaly 29, primary, autosomal recessive, 620047	MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS
PDE10A	99.1%	97%	99.8%	94.6%	Striatal degeneration, autosomal dominant, 616922;Dyskinesia, limb and orofacial, infantile-onset, 616921	MOVEMENT DISORDERS PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹
PDE11A	100%	100%	100%	99.5%	Pigmented nodular adrenocortical disease, primary, 2, 610475	MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS
PDE12	100%	100%	100%	99.4%		MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS MITOCHONDRIAL DISORDERS PANEL

PDE1C	100%	100%	100%	99.4%	?Deafness, autosomal dominant 74, 618140	HEARING IMPAIRMENT PANEL (INCLUDING GJB2) MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS
PDE2A	100%	99.9%	100%	99.2%	Intellectual developmental disorder with paroxysmal dyskinesia or seizures, 619150	MOVEMENT DISORDERS PANEL INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS MITOCHONDRIAL DISORDERS PANEL COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹
PDE3A	100%	100%	100%	98.6%	Hypertension and brachydactyly syndrome, 112410	SHORT STATURE/SKELETAL DYSPLASIA PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS

PDE4D	100%	100%	100%	98.7%	Acrodysostosis 2, with or without hormone resistance, 614613	SHORT STATURE/SKELETAL DYSPLASIA PANEL WITH GENOME WIDE CNV ANALYSIS INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS
PDE6A	100%	100%	100%	99.3%	Retinitis pigmentosa 43, 613810	VISION DISORDERS PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹
PDE6B	100%	100%	100%	98.8%	Retinitis pigmentosa-40, 613801;Night blindness, congenital stationary, autosomal dominant 2, 163500	VISION DISORDERS PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹

PDE6C	100%	100%	100%	99.6%	Cone dystrophy 4, 613093	VISION DISORDERS PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL¹
PDE6D	100%	100%	100%	99.3%	Joubert syndrome 22, 615665	VISION DISORDERS PANEL CILIOPATHIES PANEL RENAL DISORDERS PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL¹
PDE6G	100%	100%	100%	99.5%	Retinitis pigmentosa 57, 613582	VISION DISORDERS PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL¹

PDE6H	100%	100%	100%	99.7%	Retinal cone dystrophy 3, 610024;Achromatopsia 6, 610024	VISION DISORDERS PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹
PDE8B	100%	100%	100%	99.1%	Pigmented nodular adrenocortical disease, primary, 3, 614190;Striatal degeneration, autosomal dominant, 609161	MOVEMENT DISORDERS PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS
PDGFB	100%	100%	100%	99.4%	Meningioma, SIS-related, 607174;Basal ganglia calcification, idiopathic, 5, 615483;Dermatofibrosarcoma protuberans, 607907	MOVEMENT DISORDERS PANEL SKIN DISORDERS PANEL ¹ MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS PARKINSON DISEASE PANEL HEREDITARY CANCER PANEL

PDGFRA	100%	100%	100%	99.5%	Gastrointestinal stromal tumor/GIST-plus syndrome, somatic or familial, 175510;Hypereosinophilic syndrome, idiopathic, resistant to imatinib, 607685	VISION DISORDERS PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS HEREDITARY CANCER PANEL
PDGFRB	100%	100%	100%	98.5%	Premature aging syndrome, Penttinen type, 601812;Kosaki overgrowth syndrome, 616592;Myofibromatosis, infantile, 1, 228550;Basal ganglia calcification, idiopathic, 4, 615007;Myeloproliferative disorder with eosinophilia, 131440	MOVEMENT DISORDERS PANEL INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS SKIN DISORDERS PANEL ¹ TALL STATURE PANEL WITH GENOME WIDE CNV ANALYSIS SHORT STATURE/SKELETAL DYSPLASIA PANEL WITH GENOME WIDE CNV ANALYSIS PARKINSON DISEASE PANEL

PDGFRL	100%	100%	100%	98.1%	Hepatocellular cancer, somatic, 114550;Colorectal cancer, somatic, 114500	MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS
PDHA1	99%	94.5%	98.9%	68.2%	Pyruvate dehydrogenase E1-alpha deficiency, 312170	MOVEMENT DISORDERS PANEL EPILEPSY PANEL POLYNEUROPATHIES PANEL ¹ INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS MITOCHONDRIAL DISORDERS PANEL
PDHA2	100%	100%	100%	99.3%	Spermatogenic failure 70, 619828	MALE INFERTILITY PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS

PDHB	100%	100%	100%	99.4%	Pyruvate dehydrogenase E1-beta deficiency, 614111	EPILEPSY PANEL INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS MITOCHONDRIAL DISORDERS PANEL COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL¹
PDHX	100%	100%	100%	99.4%	Lacticacidemia due to PDX1 deficiency, 245349	MOVEMENT DISORDERS PANEL EPILEPSY PANEL INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS MITOCHONDRIAL DISORDERS PANEL COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL¹

PDIA6	100%	100%	100%	99.5%		MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹
PDK1	100%	100%	100%	99.3%		MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS MITOCHONDRIAL DISORDERS PANEL
PDK2	100%	99.9%	100%	98.6%		MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS MITOCHONDRIAL DISORDERS PANEL
PDK3	100%	100%	99.1%	70.7%	?Charcot-Marie-Tooth disease, X-linked dominant, 6, 300905	POLYNEUROPATHIES PANEL ¹ MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS MITOCHONDRIAL DISORDERS PANEL

PDK4	100%	100%	100%	99.5%		MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS MITOCHONDRIAL DISORDERS PANEL
PDLIM3	100%	100%	100%	99.6%		HEART DISORDERS PANEL ¹ MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS
PDLIM5	99.8%	98.2%	100%	98.7%		HEART DISORDERS PANEL ¹ MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS

PDP1	100%	100%	100%	99.7%	Pyruvate dehydrogenase phosphatase deficiency, 608782	EPILEPSY PANEL INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS MITOCHONDRIAL DISORDERS PANEL COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL¹
PDSS1	100%	100%	100%	99.4%	Coenzyme Q10 deficiency, primary, 2, 614651	MITOCHONDRIAL DISORDERS PANEL COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL¹ INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS MOVEMENT DISORDERS PANEL VISION DISORDERS PANEL METABOLIC DISORDERS PANEL

PDSS2	100%	100%	100%	99.2%	Coenzyme Q10 deficiency, primary, 3, 614652	MOVEMENT DISORDERS PANEL MITOCHONDRIAL DISORDERS PANEL COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL¹ METABOLIC DISORDERS PANEL INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS RENAL DISORDERS PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS
PDX1	100%	100%	100%	99%	{Diabetes mellitus, type II, susceptibility to}, 125853;Pancreatic agenesis 1, 260370;MODY, type IV, 606392	MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL¹ EPILEPSY PANEL

PDXK	99.8%	97.7%	100%	98.7%	Neuropathy, hereditary motor and sensory, type VIC, with optic atrophy, 618511	POLYNEUROPATHIES PANEL ¹ MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹
PDYN	100%	100%	100%	98.8%	Spinocerebellar ataxia 23, 610245	MOVEMENT DISORDERS PANEL POLYNEUROPATHIES PANEL ¹ MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS
PDZD7	100%	99.5%	100%	98.9%	Deafness, autosomal recessive 57, 618003;{Retinal disease in Usher syndrome type IIA, modifier of}, 276901;Usher syndrome, type IIC, GPR98/PDZD7 digenic, 605472	VISION DISORDERS PANEL HEARING IMPAIRMENT PANEL (INCLUDING GJB2) MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹

PDZD8	100%	100%	100%	97.9%	Intellectual developmental disorder with autism and dysmorphic facies, 620021	INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹
PEPD	93.9%	93.9%	100%	99.6%	Prolidase deficiency, 170100	SKIN DISORDERS PANEL ¹ MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹ PRIMARY IMMUNODEFICIENCIES PANEL METABOLIC DISORDERS PANEL INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS

PER2	100%	100%	100%	99.3%	?Advanced sleep phase syndrome, familial, 1, 604348	MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS
PER3	100%	100%	100%	98.5%	?Advanced sleep phase syndrome, familial, 3, 616882	MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS
PERCC1	100%	100%	100%	99.5%	Diarrhea 11, malabsorptive, congenital, 618662	MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹
PERP	100%	100%	100%	98.1%	Erythrokeratoderma variabilis et progressiva 7, 619209;Olmsted syndrome 2, 619208	SKIN DISORDERS PANEL ¹ MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹

PET100	100%	100%	100%	98.4%	Mitochondrial complex IV deficiency, nuclear type 12, 619055	VISION DISORDERS PANEL HEARING IMPAIRMENT PANEL (INCLUDING GJB2) EPILEPSY PANEL INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS MITOCHONDRIAL DISORDERS PANEL COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹
PET117	100%	100%	100%	99%	?Mitochondrial complex IV deficiency, nuclear type 19, 619063	MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS MITOCHONDRIAL DISORDERS PANEL

PEX1	100%	100%	100%	99.3%	Heimler syndrome 1, 234580;Peroxisome biogenesis disorder 1B (NALD/IRD), 601539;Peroxisome biogenesis disorder 1A (Zellweger), 214100	MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹ LIVER DISORDERS PANEL METABOLIC DISORDERS PANEL INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS VISION DISORDERS PANEL HEARING IMPAIRMENT PANEL (INCLUDING GJB2) EPILEPSY PANEL POLYNEUROPATHIES PANEL ¹
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PEX10	100%	99.9%	100%	99.3%	Peroxisome biogenesis disorder 6A (Zellweger), 614870;Peroxisome biogenesis disorder 6B, 614871	COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹ METABOLIC DISORDERS PANEL INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS MOVEMENT DISORDERS PANEL EPILEPSY PANEL POLYNEUROPATHIES PANEL ¹ LIVER DISORDERS PANEL
PEX11B	100%	100%	100%	98.8%	Peroxisome biogenesis disorder 14B, 614920	METABOLIC DISORDERS PANEL INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹

PEX12	100%	100%	100%	98.7%	Peroxisome biogenesis disorder 3B, 266510;Peroxisome biogenesis disorder 3A (Zellweger), 614859	EPILEPSY PANEL LIVER DISORDERS PANEL METABOLIC DISORDERS PANEL INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹
PEX13	100%	100%	100%	99.6%	Peroxisome biogenesis disorder 11A (Zellweger), 614883;Peroxisome biogenesis disorder 11B, 614885	EPILEPSY PANEL LIVER DISORDERS PANEL METABOLIC DISORDERS PANEL INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹

PEX14	100%	100%	100%	99.2%	Peroxisome biogenesis disorder 13A (Zellweger), 614887	EPILEPSY PANEL LIVER DISORDERS PANEL METABOLIC DISORDERS PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹
PEX16	100%	100%	100%	99.2%	Peroxisome biogenesis disorder 8B, 614877;Peroxisome biogenesis disorder 8A (Zellweger), 614876	EPILEPSY PANEL POLYNEUROPATHIES PANEL ¹ PRIMARY IMMUNODEFICIENCIES PANEL LIVER DISORDERS PANEL METABOLIC DISORDERS PANEL INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹

PEX19	100%	100%	100%	99.3%	Peroxisome biogenesis disorder 12A (Zellweger), 614886	EPILEPSY PANEL LIVER DISORDERS PANEL METABOLIC DISORDERS PANEL INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL¹
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PEX2	100%	100%	100%	99.1%	Peroxisome biogenesis disorder 5A (Zellweger), 614866;Peroxisome biogenesis disorder 5B, 614867	MOVEMENT DISORDERS PANEL VISION DISORDERS PANEL LIVER DISORDERS PANEL METABOLIC DISORDERS PANEL INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL¹
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PEX26	100%	100%	100%	98.3%	Peroxisome biogenesis disorder 7B, 614873;Peroxisome biogenesis disorder 7A (Zellweger), 614872	VISION DISORDERS PANEL HEARING IMPAIRMENT PANEL (INCLUDING GJB2) EPILEPSY PANEL LIVER DISORDERS PANEL METABOLIC DISORDERS PANEL INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹
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PEX3	100%	100%	100%	99.1%	Peroxisome biogenesis disorder 10A (Zellweger), 614882;?Peroxisome biogenesis disorder 10B, 617370	EPILEPSY PANEL LIVER DISORDERS PANEL METABOLIC DISORDERS PANEL INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL¹
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PEX5	100%	100%	100%	98.4%	Peroxisome biogenesis disorder 2B, 202370;Peroxisome biogenesis disorder 2A (Zellweger), 214110;Rhizomelic chondrodysplasia punctata, type 5, 616716	EPILEPSY PANEL HEART DISORDERS PANEL ¹ SHORT STATURE/SKELETAL DYSPLASIA PANEL WITH GENOME WIDE CNV ANALYSIS LIVER DISORDERS PANEL METABOLIC DISORDERS PANEL INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹
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PEX6	100%	100%	100%	98.5%	Peroxisome biogenesis disorder 4B, 614863;Peroxisome biogenesis disorder 4A (Zellweger), 614862;Heimler syndrome 2, 616617	VISION DISORDERS PANEL HEARING IMPAIRMENT PANEL (INCLUDING GJB2) EPILEPSY PANEL SHORT STATURE/SKELETAL DYSPLASIA PANEL WITH GENOME WIDE CNV ANALYSIS LIVER DISORDERS PANEL METABOLIC DISORDERS PANEL INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹
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PEX7	97.9%	97.9%	100%	99.4%	Rhizomelic chondrodysplasia punctata, type 1, 215100;Peroxisome biogenesis disorder 9B, 614879	MOVEMENT DISORDERS PANEL VISION DISORDERS PANEL SKIN DISORDERS PANEL ¹ HEART DISORDERS PANEL ¹ POLYNEUROPATHIES PANEL ¹ SHORT STATURE/SKELETAL DYSPLASIA PANEL WITH GENOME WIDE CNV ANALYSIS LIVER DISORDERS PANEL METABOLIC DISORDERS PANEL INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹
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PFAS	100%	100%	100%	98.5%		METABOLIC DISORDERS PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS
PFKM	100%	100%	100%	99.2%	Glycogen storage disease VII, 232800	METABOLIC DISORDERS PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹ MUSCLE DISORDERS PANEL
PFN1	100%	100%	100%	98.1%	Amyotrophic lateral sclerosis 18, 614808	AMYOTROPHIC LATERAL SCLEROSIS, ALS PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS

PGAM2	100%	100%	100%	99.8%	Glycogen storage disease X, 261670	METABOLIC DISORDERS PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL¹ MUSCLE DISORDERS PANEL
PGAP1	100%	100%	100%	99.2%	Neurodevelopmental disorder with dysmorphic features, spasticity, and brain abnormalities, 615802	METABOLIC DISORDERS PANEL INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL¹

PGAP2	100%	100%	100%	99%	Hyperphosphatasia with impaired intellectual development syndrome 3, 614207	METABOLIC DISORDERS PANEL INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹
PGAP3	100%	100%	100%	99.1%	Hyperphosphatasia with impaired intellectual development syndrome 4, 615716	EPILEPSY PANEL METABOLIC DISORDERS PANEL INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹

PGK1	100%	99.2%	98.8%	70.3%	Phosphoglycerate kinase 1 deficiency, 300653	VISION DISORDERS PANEL METABOLIC DISORDERS PANEL INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS MUSCLE DISORDERS PANEL
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PGM1	94%	94%	100%	99.2%	Congenital disorder of glycosylation, type It, 614921	CRANIOFACIAL ANOMALIES PANEL WITH GENOME WIDE CNV ANALYSIS HEART DISORDERS PANEL ¹ METABOLIC DISORDERS PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹ OROFACIAL CLEFTING PANEL WITH GENOME WIDE CNV ANALYSIS MUSCLE DISORDERS PANEL
PGM2L1	100%	100%	100%	99.7%	Neurodevelopmental disorder with hypotonia, dysmorphic facies, and skin abnormalities, 620191	METABOLIC DISORDERS PANEL INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹

PGM3	100%	100%	100%	99.6%	Immunodeficiency 23, 615816	PRIMARY IMMUNODEFICIENCIES PANEL METABOLIC DISORDERS PANEL INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹
PHACTR1	100%	99.9%	100%	97.5%	Developmental and epileptic encephalopathy 70, 618298	EPILEPSY PANEL INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS
PHC1	100%	99.9%	100%	98.9%	?Microcephaly 11, primary, autosomal recessive, 615414	MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS

PHEX	100%	100%	99.3%	70.3%	Hypophosphatemic rickets, X-linked dominant, 307800	SKIN DISORDERS PANEL ¹ SHORT STATURE/SKELETAL DYSPLASIA PANEL WITH GENOME WIDE CNV ANALYSIS RENAL DISORDERS PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS
PHF21A	100%	100%	100%	99.1%	Intellectual developmental disorder with behavioral abnormalities and craniofacial dysmorphism with or without seizures, 618725	EPILEPSY PANEL INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS
PHF5A	100%	100%	100%	99.3%		INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS

PHF6	100%	100%	99.1%	71.4%	Borjeson-Forssman-Lehmann syndrome, 301900	DISORDERS/DIFFERENCES OF SEX DEVELOPMENT (DSD) / PRIMARY ADRENAL INSUFFICIENCY PANEL EPILEPSY PANEL HYPOGONADOTROPIC HYPOGONADISM (KALLMANN) PANEL INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS
PHF8	100%	99.7%	98.6%	68%	Intellectual developmental disorder, X-linked syndromic, Siderius type, 300263	INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS OROFACIAL CLEFTING PANEL WITH GENOME WIDE CNV ANALYSIS

PHGDH	100%	100%	100%	99.4%	Neu-Laxova syndrome 1, 256520;Phosphoglycerate dehydrogenase deficiency, 601815	FETAL AKINESIA PANEL SKIN DISORDERS PANEL ¹ EPILEPSY PANEL SHORT STATURE/SKELETAL DYSPLASIA PANEL WITH GENOME WIDE CNV ANALYSIS METABOLIC DISORDERS PANEL INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹ OROFACIAL CLEFTING PANEL WITH GENOME WIDE CNV ANALYSIS
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PHIP	99.3%	98.8%	100%	98.9%	Chung-Jansen syndrome, 617991	INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS
PHKA1	100%	100%	99%	69.7%	Muscle glycogenosis, 300559	HEART DISORDERS PANEL ¹ METABOLIC DISORDERS PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS MUSCLE DISORDERS PANEL
PHKA2	100%	99.7%	99%	68.5%	Glycogen storage disease, type IXa2, 306000;Glycogen storage disease, type IXa1, 306000	METABOLIC DISORDERS PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS

PHKB	100%	100%	100%	99.7%	Phosphorylase kinase deficiency of liver and muscle, autosomal recessive, 261750	METABOLIC DISORDERS PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹
PHKG1	100%	100%	100%	99.1%		METABOLIC DISORDERS PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS
PHKG2	100%	100%	100%	99%	Glycogen storage disease IXc, 613027	LIVER DISORDERS PANEL METABOLIC DISORDERS PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹
PHLDB1	100%	100%	100%	98.8%	Osteogenesis imperfecta, type XXIII, 620639	MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS

PHOX2A	100%	99.7%	100%	99%	Fibrosis of extraocular muscles, congenital, 2, 602078	VISION DISORDERS PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹ MUSCLE DISORDERS PANEL
PHOX2B	100%	99.8%	100%	97.5%	{Neuroblastoma, susceptibility to, 2}, 613013;Neuroblastoma with Hirschsprung disease, 613013;Central hypoventilation syndrome, congenital, 1, with or without Hirschsprung disease, 209880	MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS HEREDITARY CANCER PANEL

PHYH	100%	100%	100%	99.2%	Refsum disease, 266500	MOVEMENT DISORDERS PANEL VISION DISORDERS PANEL SKIN DISORDERS PANEL ¹ HEART DISORDERS PANEL ¹ POLYNEUROPATHIES PANEL ¹ METABOLIC DISORDERS PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹
PI4K2A	100%	100%	100%	98.4%	Neurodevelopmental disorder with hyperkinetic movements, seizures and structural brain abnormalities, 620732	EPILEPSY PANEL METABOLIC DISORDERS PANEL INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹

PI4KA	100%	99.8%	100%	98.7%	Spastic paraplegia 84, autosomal recessive, 619621;Gastrointestinal defects and immunodeficiency syndrome 2, 619708;Polymicrogyria, perisylvian, with cerebellar hypoplasia and arthrogryposis, 616531	PRIMARY IMMUNODEFICIENCIES PANEL METABOLIC DISORDERS PANEL INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹
PI4KB	100%	100%	100%	98.8%	Deafness, autosomal dominant 87, 620281	HEARING IMPAIRMENT PANEL (INCLUDING GJB2) MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS

PIBF1	100%	100%	100%	99.6%	Joubert syndrome 33, 617767	CILIOPATHIES PANEL INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹
PICALM	100%	100%	100%	99.5%	Leukemia, acute myeloid, somatic, 601626	MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS
PICK1	100%	100%	100%	98.6%		MALE INFERTILITY PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS

PIDD1	100%	100%	100%	99.2%	Intellectual developmental disorder, autosomal recessive 75, with neuropsychiatric features and variant lissencephaly, 619827	INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹
PIEZO1	100%	100%	100%	98.7%	[ER blood group system], 620207;Lymphatic malformation 6, 616843;Dehydrated hereditary stomatocytosis with or without pseudohyperkalemia and/or perinatal edema, 194380	SKIN DISORDERS PANEL ¹ LYMPHATIC ANOMALIES PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹

PIEZO2	100%	100%	100%	99.1%	Arthrogryposis, distal, type 5, 108145;Arthrogryposis, distal, with impaired proprioception and touch, 617146;Arthrogryposis, distal, type 3, 114300;?Marden-Walker syndrome, 248700	FETAL AKINESIA PANEL POLYNEUROPATHIES PANEL ¹ NEUROLOGICAL PAIN DISORDERS PANEL ¹ MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹ OROFACIAL CLEFTING PANEL WITH GENOME WIDE CNV ANALYSIS MUSCLE DISORDERS PANEL
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PIGA	100%	100%	99.3%	71.1%	Paroxysmal nocturnal hemoglobinuria, somatic, 300818;Multiple congenital anomalies-hypotonia-seizures syndrome 2, 300868;Neurodevelopmental disorder with epilepsy and hemochromatosis, 301072	SKIN DISORDERS PANEL ¹ EPILEPSY PANEL HEMOSTATIC/THROMB OTIC DISORDERS PANEL PRIMARY IMMUNODEFICIENCIES PANEL METABOLIC DISORDERS PANEL INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS MITOCHONDRIAL DISORDERS PANEL
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PIGB	100%	100%	100%	99.3%	Developmental and epileptic encephalopathy 80, 618580	EPILEPSY PANEL POLYNEUROPATHIES PANEL ¹ METABOLIC DISORDERS PANEL INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹
PIGC	100%	100%	100%	99.4%	Glycosylphosphatidylinositol biosynthesis defect 16, 617816	METABOLIC DISORDERS PANEL INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹

PIGF	100%	100%	100%	99.6%	Onychodystrophy, osteodystrophy, impaired intellectual development, and seizures syndrome, 619356	INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS
PIGG	100%	100%	100%	98.9%	[Blood group, EMM system], 619812;Neurodevelopmental disorder with or without hypotonia, seizures, and cerebellar atrophy, 616917	MOVEMENT DISORDERS PANEL EPILEPSY PANEL POLYNEUROPATHIES PANEL ¹ INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹

PIGH	84.3%	76.2%	100%	99.4%	Glycosylphosphatidylinositol biosynthesis defect 17, 618010	INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹
PIGK	100%	100%	100%	99.6%	Neurodevelopmental disorder with hypotonia and cerebellar atrophy, with or without seizures, 618879	EPILEPSY PANEL INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹

PIGL	100%	100%	100%	99%	CHIME syndrome, 280000	SKIN DISORDERS PANEL ¹ METABOLIC DISORDERS PANEL INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹
PIGM	100%	100%	100%	98.5%	Glycosylphosphatidylinositol deficiency, 610293	METABOLIC DISORDERS PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹

PIGN	100%	100%	100%	99.7%	Multiple congenital anomalies-hypotonia-seizures syndrome 1, 614080	SKIN DISORDERS PANEL ¹ OROFACIAL CLEFTING PANEL WITH GENOME WIDE CNV ANALYSIS EPILEPSY PANEL METABOLIC DISORDERS PANEL INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹
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PIGO	100%	100%	100%	99%	Hyperphosphatasia with impaired intellectual development syndrome 2, 614749	EPILEPSY PANEL METABOLIC DISORDERS PANEL INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL¹ OROFACIAL CLEFTING PANEL WITH GENOME WIDE CNV ANALYSIS
PIGP	100%	100%	100%	99.6%	Developmental and epileptic encephalopathy 55, 617599	EPILEPSY PANEL METABOLIC DISORDERS PANEL COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL¹ INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS

PIGQ	100%	100%	100%	99.4%	Multiple congenital anomalies-hypotonia-seizures syndrome 4, 618548	EPILEPSY PANEL METABOLIC DISORDERS PANEL INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL¹
PIGS	98.7%	95.2%	100%	98.9%	Developmental and epileptic encephalopathy 95, 618143	EPILEPSY PANEL INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL¹

PIGT	100%	99.9%	100%	98.9%	?Paroxysmal nocturnal hemoglobinuria 2, 615399;Multiple congenital anomalies-hypotonia-seizures syndrome 3, 615398	EPILEPSY PANEL METABOLIC DISORDERS PANEL INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹
PIGU	100%	100%	100%	99.5%	Neurodevelopmental disorder with brain anomalies, seizures, and scoliosis, 618590	EPILEPSY PANEL INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹

PIGV	100%	99.8%	100%	99.6%	Hyperphosphatasia with impaired intellectual development syndrome 1, 239300	SKIN DISORDERS PANEL ¹ SHORT STATURE/SKELETAL DYSPLASIA PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹ OROFACIAL CLEFTING PANEL WITH GENOME WIDE CNV ANALYSIS METABOLIC DISORDERS PANEL INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS
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PIGW	100%	100%	100%	99.2%	Glycosylphosphatidylinositol biosynthesis defect 11, 616025	METABOLIC DISORDERS PANEL INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL¹
PIGY	100%	100%	100%	99.9%	Hyperphosphatasia with impaired intellectual development syndrome 6, 616809	METABOLIC DISORDERS PANEL INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL¹

PIK3C2A	100%	100%	100%	99.5%	Oculoskeletodental syndrome, 618440	CILIOPATHIES PANEL INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹
PIK3CA	100%	100%	100%	99.5%	Hemifacial myohyperplasia, somatic, 606773;CLOVE syndrome, somatic, 612918;Hepatocellular carcinoma, somatic, 114550;Breast cancer, somatic, 114480;Cerebral cavernous malformations 4, somatic, 619538;Ovarian cancer, somatic, 167000;Colorectal cancer, somatic, 114500;Macrodactyly, somatic, 155500;CLAPO syndrome, somatic, 613089;Keratosis, seborrheic, somatic, 182000;Nevus, epidermal, somatic, 162900;Gastric cancer, somatic, 613659;Non-small cell lung cancer, somatic, 211980;Megalencephaly-capillary malformation-polymicrogyria syndrome, somatic, 602501;Cowden syndrome 5, 615108	SKIN DISORDERS PANEL ¹ LYMPHATIC ANOMALIES PANEL WITH GENOME WIDE CNV ANALYSIS METABOLIC DISORDERS PANEL INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS HEREDITARY CANCER PANEL

PIK3CD	100%	100%	100%	98.8%	Immunodeficiency 14A, autosomal dominant, 615513;Immunodeficiency 14B, autosomal recessive, 619281;?Roifman-Chitayat syndrome, digenic, 613328	PRIMARY IMMUNODEFICIENCIES PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL¹
PIK3CG	100%	100%	100%	99.7%	Immunodeficiency 97 with autoinflammation, 619802	PRIMARY IMMUNODEFICIENCIES PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS

PIK3R1	100%	100%	100%	99.6%	Immunodeficiency 36, 616005;?Agammaglobulinemia 7, autosomal recessive, 615214;SHORT syndrome, 269880	SKIN DISORDERS PANEL ¹ PRIMARY IMMUNODEFICIENCIES PANEL SHORT STATURE/SKELETAL DYSPLASIA PANEL WITH GENOME WIDE CNV ANALYSIS METABOLIC DISORDERS PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹
PIK3R2	100%	99.9%	100%	97.9%	Megalencephaly-polymicrogyria-polydactyly-hydrocephalus syndrome 1, 603387	SKIN DISORDERS PANEL ¹ METABOLIC DISORDERS PANEL INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS

PIK3R5	100%	99.9%	100%	98.1%	Ataxia-oculomotor apraxia 3, 615217	MOVEMENT DISORDERS PANEL METABOLIC DISORDERS PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL¹
PIKFYVE	100%	100%	100%	99.4%	Corneal fleck dystrophy, 121850	VISION DISORDERS PANEL METABOLIC DISORDERS PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS
PINK1	100%	100%	100%	98.2%	Parkinson disease 6, early onset, 605909	MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS PARKINSON DISEASE PANEL COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL¹

PIP5K1C	100%	100%	100%	98.8%	Lethal congenital contractural syndrome 3, 611369	FETAL AKINESIA PANEL METABOLIC DISORDERS PANEL INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹ MUSCLE DISORDERS PANEL
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PISD	100%	100%	100%	99.5%	Liberfarb syndrome, 618889	HEARING IMPAIRMENT PANEL (INCLUDING GJB2) SHORT STATURE/SKELETAL DYSPLASIA PANEL WITH GENOME WIDE CNV ANALYSIS INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS MITOCHONDRIAL DISORDERS PANEL COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL¹
PITPNM3	100%	100%	99.9%	97.5%	Cone-rod dystrophy 5, 600977	MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS

PITRM1	100%	100%	100%	99.1%	Spinocerebellar ataxia, autosomal recessive 30, 619405	INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS MITOCHONDRIAL DISORDERS PANEL
PITX1	100%	100%	100%	97.3%	Clubfoot, congenital, with or without deficiency of long bones and/or mirror-image polydactyly, 119800	SHORT STATURE/SKELETAL DYSPLASIA PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS

PITX2	100%	99.8%	100%	98.7%	Ring dermoid of cornea, 180550;Axenfeld-Rieger syndrome, type 1, 180500;Anterior segment dysgenesis 4, 137600	VISION DISORDERS PANEL CRANIOFACIAL ANOMALIES PANEL WITH GENOME WIDE CNV ANALYSIS CONGENITAL HEARTDISEASE PANEL ¹ SKIN DISORDERS PANEL ¹ HEART DISORDERS PANEL ¹ SHORT STATURE/SKELETAL DYSPLASIA PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS
PITX3	100%	100%	100%	97.4%	Cataract 11, multiple types, 610623;Anterior segment dysgenesis 1, multiple subtypes, 107250;Cataract 11, syndromic, autosomal recessive, 610623	VISION DISORDERS PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS

PIWIL2	100%	100%	99.9%	99.4%		MALE INFERTILITY PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS
PJA1	100%	100%	96.7%	62.1%		INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS
PJVK	100%	100%	100%	99.5%	Deafness, autosomal recessive 59, 610220	HEARING IMPAIRMENT PANEL (INCLUDING GJB2) MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹

PKD1	99.7%	99.5%	100%	97.8%	Polycystic kidney disease 1, 173900	CILIOPATHIES PANEL LIVER DISORDERS PANEL RENAL DISORDERS PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS
PKD1L1	100%	100%	100%	99.2%	Heterotaxy, visceral, 8, autosomal, 617205	CONGENITAL HEARTDISEASE PANEL ¹ CILIOPATHIES PANEL HEART DISORDERS PANEL ¹ MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹
PKD2	100%	100%	100%	98.1%	Polycystic kidney disease 2, 613095	CILIOPATHIES PANEL LIVER DISORDERS PANEL RENAL DISORDERS PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS

PKDCC	100%	99.2%	99.7%	89.6%	Rhizomelic limb shortening with dysmorphic features, 618821	SHORT STATURE/SKELETAL DYSPLASIA PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹
PKHD1	100%	100%	100%	99%	Polycystic kidney disease 4, with or without hepatic disease, 263200	CILIOPATHIES PANEL LIVER DISORDERS PANEL RENAL DISORDERS PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹
PKHD1L1	100%	100%	100%	99.6%	Deafness, autosomal recessive 124, 620794	HEARING IMPAIRMENT PANEL (INCLUDING GJB2) MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS

PKLR	100%	100%	100%	98.8%	Anemia, congenital, nonspherocytic hemolytic, 2, pyruvate kinase deficient, 266200;[Adenosine triphosphate, elevated, of erythrocytes], 102900	METABOLIC DISORDERS PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹
PKP1	100%	100%	100%	99.3%	Ectodermal dysplasia/skin fragility syndrome, 604536	SKIN DISORDERS PANEL ¹ MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹

PKP2	99%	97.7%	100%	99.2%	Arrhythmogenic right ventricular dysplasia 9, 609040	ARITMOGENE CARDIOMYOPATHY PANEL ¹ CONGENITAL HEARTDISEASE PANEL ¹ HEART DISORDERS PANEL ¹ MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS ARRHYTHMIA AND CARDIAC CONDUCTION DISORDERS PANEL ¹
PLA2G4A	100%	100%	100%	99.3%	Gastrointestinal ulceration, recurrent, with dysfunctional platelets, 618372	HEMOSTATIC/THROMB OTIC DISORDERS PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹

PLA2G5	100%	100%	100%	99.5%	[Fleck retina, familial benign], 228980	VISION DISORDERS PANEL METABOLIC DISORDERS PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS
PLA2G6	100%	99.9%	100%	98.9%	Parkinson disease 14, autosomal recessive, 612953;Neurodegeneration with brain iron accumulation 2B, 610217;Infantile neuroaxonal dystrophy 1, 256600	MOVEMENT DISORDERS PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS MITOCHONDRIAL DISORDERS PANEL EPILEPSY PANEL POLYNEUROPATHIES PANEL ¹ METABOLIC DISORDERS PANEL INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS PARKINSON DISEASE PANEL COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹

PLA2G7	100%	100%	100%	99.6%	Platelet-activating factor acetylhydrolase deficiency, 614278	HEMOSTATIC/THROMBOTIC DISORDERS PANEL METABOLIC DISORDERS PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹
PLAA	100%	100%	100%	99.6%	Neurodevelopmental disorder with progressive microcephaly, spasticity, and brain anomalies, 617527	INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹

PLAAT3	100%	100%	100%	99.4%	Lipodystrophy, familial partial, type 9, 620683	METABOLIC DISORDERS PANEL INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹
PLAG1	100%	100%	100%	99.4%	Adenomas, salivary gland pleomorphic, somatic, 181030;Silver-Russell syndrome 4, 618907	SHORT STATURE/SKELETAL DYSPLASIA PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS
PLAT	96%	96%	100%	98.8%		HEMOSTATIC/THROMB OTIC DISORDERS PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS

PLAU	100%	100%	100%	99.3%	Quebec platelet disorder, 601709;{Alzheimer disease, late-onset, susceptibility to}, 104300	HEMOSTATIC/THROMBOTIC DISORDERS PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS
PLCB1	100%	100%	100%	99.4%	Developmental and epileptic encephalopathy 12, 613722	EPILEPSY PANEL METABOLIC DISORDERS PANEL INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹
PLCB3	100%	100%	100%	98.4%	Spondylometaphyseal dysplasia with corneal dystrophy, 618961	SHORT STATURE/SKELETAL DYSPLASIA PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS

PLCB4	99%	99%	100%	99.6%	Auriculocondylar syndrome 2B, 620458;Auriculocondylar syndrome 2A, 614669	CRANIOFACIAL ANOMALIES PANEL WITH GENOME WIDE CNV ANALYSIS SHORT STATURE/SKELETAL DYSPLASIA PANEL WITH GENOME WIDE CNV ANALYSIS METABOLIC DISORDERS PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹ OROFACIAL CLEFTING PANEL WITH GENOME WIDE CNV ANALYSIS
PLCD1	100%	100%	100%	99%	Nail disorder, nonsyndromic congenital, 3, (leukonychia), 151600	SKIN DISORDERS PANEL ¹ METABOLIC DISORDERS PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹

PLCE1	100%	100%	100%	99.4%	Nephrotic syndrome, type 3, 610725	METABOLIC DISORDERS PANEL RENAL DISORDERS PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹
PLCG1	100%	100%	100%	99.3%	?Immune dysregulation, autoimmunity, and autoinflammation, 620514	HEARING IMPAIRMENT PANEL (INCLUDING GJB2) MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS
PLCG2	100%	100%	100%	99.1%	Autoinflammation, antibody deficiency, and immune dysregulation syndrome, 614878;Familial cold autoinflammatory syndrome 3, 614468	SKIN DISORDERS PANEL ¹ PRIMARY IMMUNODEFICIENCIES PANEL METABOLIC DISORDERS PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS

PLCH1	100%	100%	100%	99.5%	Holoprosencephaly 14, 619895	MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS
PLCZ1	100%	100%	99.6%	96.9%	Spermatogenic failure 17, 617214	MALE INFERTILITY PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS
PLD1	100%	100%	100%	99.6%	Cardiac valvular dysplasia 1, 212093	CONGENITAL HEARTDISEASE PANEL¹ HEART DISORDERS PANEL¹ MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL¹
PLD3	100%	100%	100%	97.9%	?Spinocerebellar ataxia 46, 617770	POLYNEUROPATHIES PANEL¹ MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS

PLD6	100%	100%	100%	98.8%		MALE INFERTILITY PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS
PLEC	100%	100%	100%	98.6%	?Epidermolysis bullosa simplex 5D, generalized intermediate, autosomal recessive, 616487;Epidermolysis bullosa simplex 5B, with muscular dystrophy, 226670;Epidermolysis bullosa simplex 5C, with pyloric atresia, 612138;Epidermolysis bullosa simplex 5A, Ogna type, 131950;Muscular dystrophy, limb-girdle, autosomal recessive 17, 613723	SKIN DISORDERS PANEL ¹ MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹ MUSCLE DISORDERS PANEL
PLEKHG2	100%	100%	100%	98.7%	Leukodystrophy and acquired microcephaly with or without dystonia, 616763	INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹

PLEKHG5	100%	100%	100%	99.1%	Neuronopathy, distal hereditary motor, autosomal recessive 4, 611067;Charcot-Marie-Tooth disease, recessive intermediate C, 615376	POLYNEUROPATHIES PANEL ¹ MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹
PLEKHM1	100%	100%	100%	98.5%	?Osteopetrosis, autosomal recessive 6, 611497;Osteopetrosis, autosomal dominant 3, 618107	INHERITED BONE MARROW FAILURE AND/OR PREDISPOSITION PANEL TO HEMATOLOGICAL MALIGNANCIES PRIMARY IMMUNODEFICIENCIES PANEL SHORT STATURE/SKELETAL DYSPLASIA PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹

PLEKHM2	100%	100%	100%	98.5%		HEART DISORDERS PANEL ¹ MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS
PLG	100%	100%	100%	99.4%	Dysplasminogenemia, 217090;Angioedema, hereditary, 4, 619360;Plasminogen deficiency, type I, 217090	SKIN DISORDERS PANEL ¹ HEMOSTATIC/THROMB OTIC DISORDERS PANEL PRIMARY IMMUNODEFICIENCIES PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹
PLIN1	100%	100%	100%	98.6%	Lipodystrophy, familial partial, type 4, 613877	SKIN DISORDERS PANEL ¹ METABOLIC DISORDERS PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS

PLIN4	99.9%	99.9%	99.8%	97.2%	Myopathy with rimmed ubiquitin-positive autophagic vacuolation, autosomal dominant, 601846	MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS
PLK1	100%	100%	100%	98.8%		INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS
PLK4	100%	100%	100%	99.5%	Microcephaly and chorioretinopathy, autosomal recessive, 2, 616171	VISION DISORDERS PANEL SHORT STATURE/SKELETAL DYSPLASIA PANEL WITH GENOME WIDE CNV ANALYSIS MALE INFERTILITY PANEL INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹

PLN	100%	100%	100%	99.4%	Cardiomyopathy, dilated, 1P, 609909;Cardiomyopathy, hypertrophic, 18, 613874	ARITMOGENE CARDIOMYOPATHY PANEL ¹ DILATED CARDIOMYOPATHY PANEL ¹ MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS ARRHYTHMIA AND CARDIAC CONDUCTION DISORDERS PANEL ¹ HEART DISORDERS PANEL ¹ HYPERTROPHIC CARDIOMYOPATHY PANEL ¹
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PLOD1	100%	100%	100%	97.7%	Ehlers-Danlos syndrome, kyphoscoliotic type, 1, 225400	MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹ FETAL AKINESIA PANEL THORACIC AORTIC ANEURYSM AND/OR DISSECTION PANEL SKIN DISORDERS PANEL ¹ SHORT STATURE/SKELETAL DYSPLASIA PANEL WITH GENOME WIDE CNV ANALYSIS METABOLIC DISORDERS PANEL
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PLOD2	100%	100%	100%	99.4%	Bruck syndrome 2, 609220	SHORT STATURE/SKELETAL DYSPLASIA PANEL WITH GENOME WIDE CNV ANALYSIS METABOLIC DISORDERS PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL¹
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PLOD3	100%	100%	100%	98.3%	BCARD syndrome (lysyl hydroxylase 3 deficiency), 612394	VISION DISORDERS PANEL SKIN DISORDERS PANEL ¹ HEARING IMPAIRMENT PANEL (INCLUDING GJB2) SHORT STATURE/SKELETAL DYSPLASIA PANEL WITH GENOME WIDE CNV ANALYSIS METABOLIC DISORDERS PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹
PLP1	100%	99.5%	99%	69.2%	Pelizaeus-Merzbacher disease, 312080;Spastic paraplegia 2, X-linked, 312920	MOVEMENT DISORDERS PANEL EPILEPSY PANEL INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS

PLPBP	100%	100%	100%	99%	Epilepsy, early-onset, 1, vitamin B6-dependent, 617290	EPILEPSY PANEL METABOLIC DISORDERS PANEL INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS MITOCHONDRIAL DISORDERS PANEL COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹
PLPP6	100%	100%	100%	98%		MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS
PLS1	100%	100%	100%	99.2%	Deafness, autosomal dominant 76, 618787	HEARING IMPAIRMENT PANEL (INCLUDING GJB2) MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS

PLS3	96.8%	96.7%	98.7%	68.3%	Bone mineral density QTL18, osteoporosis, 300910;Diaphragmatic hernia 5, X-linked, 306950	SHORT STATURE/SKELETAL DYSPLASIA PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS
PLVAP	100%	100%	100%	98.5%	Diarrhea 10, protein-losing enteropathy type, 618183	LYMPHATIC ANOMALIES PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL¹

PLXNA1	100%	100%	100%	99.4%	Dworschak-Punetha neurodevelopmental syndrome, 619955	DISORDERS/DIFFERENCES OF SEX DEVELOPMENT (DSD) / PRIMARY ADRENAL INSUFFICIENCY PANEL HYPOGONADOTROPIC HYPOGONADISM (KALLMANN) PANEL INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹
PLXNA2	100%	100%	100%	99%		INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS

PLXNB2	100%	100%	100%	97.9%		HEARING IMPAIRMENT PANEL (INCLUDING GJB2) INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS
PLXND1	100%	99.8%	100%	99%	Congenital heart defects, multiple types, 9, 620294	CONGENITAL HEARTDISEASE PANEL ¹ HEART DISORDERS PANEL ¹ INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS
PMEPA1	99.8%	95.2%	100%	97.2%		THORACIC AORTIC ANEURYSM AND/OR DISSECTION PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS

PMFBP1	100%	100%	100%	99.1%	Spermatogenic failure 31, 618112	CILIOPATHIES PANEL MALE INFERTILITY PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS
PML	100%	100%	100%	99.2%		MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS

PMM2	100%	100%	100%	99.1%	Congenital disorder of glycosylation, type Ia, 212065	MOVEMENT DISORDERS PANEL DISORDERS/DIFFERENCES OF SEX DEVELOPMENT (DSD) / PRIMARY ADRENAL INSUFFICIENCY PANEL EPILEPSY PANEL HEART DISORDERS PANEL ¹ POLYNEUROPATHIES PANEL ¹ PREMATURE OVARIAN INSUFFICIENCY PANEL PRIMARY IMMUNODEFICIENCIES PANEL LYMPHATIC ANOMALIES PANEL WITH GENOME WIDE CNV ANALYSIS METABOLIC DISORDERS PANEL INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS RENAL DISORDERS PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION
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						CARRIER TEST PANEL ¹
PMP2	100%	100%	100%	99.8%	Charcot-Marie-Tooth disease, demyelinating, type 1G, 618279	POLYNEUROPATHIES PANEL ¹ MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS
PMP22	100%	99.9%	100%	99.2%	Charcot-Marie-Tooth disease, type 1A, 118220;Roussy-Levy syndrome, 180800;Charcot-Marie-Tooth disease, type 1E, 118300;?Neuropathy, inflammatory demyelinating, 139393;Neuropathy, recurrent, with pressure palsies, 162500;Dejerine-Sottas disease, 145900	MOVEMENT DISORDERS PANEL NEUROLOGICAL PAIN DISORDERS PANEL ¹ MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS POLYNEUROPATHIES PANEL ¹

PMPCA	96%	96%	100%	99.1%	Spinocerebellar ataxia, autosomal recessive 2, 213200	MOVEMENT DISORDERS PANEL VISION DISORDERS PANEL INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS MITOCHONDRIAL DISORDERS PANEL COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹
PMPCB	91.4%	91.4%	100%	99.4%	Multiple mitochondrial dysfunctions syndrome 6, 617954	INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS MITOCHONDRIAL DISORDERS PANEL COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹

PMS2	93.7%	93.4%	100%	99.2%	Lynch syndrome 4, 614337;Mismatch repair cancer syndrome 4, 619101	INHERITED BONE MARROW FAILURE AND/OR PREDISPOSITION PANEL TO HEMATOLOGICAL MALIGNANCIES COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹ HEREDITARY CANCER PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS PANEL HEREDITARY PANCREATIC CANCER (BRCA1, BRCA2, PALB2, ATM, CDKN2A, MLH1, MSH2, MSH6, PMS2, STK11) PANEL HEREDITARY CANCER (CDH1, CTNNA1, MLH1, MSH2, MSH6, PMS2) PANEL LYNCH SYNDROME (MLH1, PMS2, MSH2, MSH6) PANEL HEREDITARY COLORECTAL AND POLYPOSIS SKIN DISORDERS PANEL ¹
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PMS2CL						MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS HEREDITARY CANCER PANEL
PMVK	100%	100%	100%	99.5%	Porokeratosis 1, multiple types, 175800	SKIN DISORDERS PANEL ¹ PRIMARY IMMUNODEFICIENCIES PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS
PNKD	100%	100%	100%	99.2%	Paroxysmal nonkinesigenic dyskinesia 1, 118800	MOVEMENT DISORDERS PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS

PNKP	100%	100%	100%	98.2%	?Charcot-Marie-Tooth disease, type 2B2, 605589;Ataxia-oculomotor apraxia 4, 616267;Microcephaly, seizures, and developmental delay, 613402	MOVEMENT DISORDERS PANEL EPILEPSY PANEL POLYNEUROPATHIES PANEL ¹ INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹
PNLDC1	100%	100%	100%	99%	Spermatogenic failure 57, 619528	MALE INFERTILITY PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS
PNLIP	100%	100%	100%	99.5%	?Pancreatic lipase deficiency, 614338	METABOLIC DISORDERS PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹

PNMT	100%	100%	100%	98.4%		METABOLIC DISORDERS PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS
PNP	100%	100%	100%	99.9%	Immunodeficiency due to purine nucleoside phosphorylase deficiency, 613179	PRIMARY IMMUNODEFICIENCIES PANEL METABOLIC DISORDERS PANEL INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL¹ SEVERE COMBINED IMMUNODEFICIENCY (SCID) PANEL

PNPLA1	100%	99.9%	100%	99.4%	Ichthyosis, congenital, autosomal recessive 10, 615024	SKIN DISORDERS PANEL ¹ MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹
PNPLA2	100%	100%	100%	99%	Neutral lipid storage disease with myopathy, 610717	SKIN DISORDERS PANEL ¹ HEART DISORDERS PANEL ¹ METABOLIC DISORDERS PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹ MUSCLE DISORDERS PANEL

PNPLA6	100%	100%	100%	99%	Spastic paraplegia 39, autosomal recessive, 612020;Oliver-McFarlane syndrome, 275400;?Laurence-Moon syndrome, 245800;Boucher-Neuhauser syndrome, 215470	MOVEMENT DISORDERS PANEL HYPOGONADOTROPIC HYPOGONADISM (KALLMANN) PANEL SHORT STATURE/SKELETAL DYSPLASIA PANEL WITH GENOME WIDE CNV ANALYSIS METABOLIC DISORDERS PANEL VISION DISORDERS PANEL DISORDERS/DIFFEREN CES OF SEX DEVELOPMENT (DSD) / PRIMARY ADRENAL INSUFFICIENCY PANEL INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹
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PNPLA8	100%	100%	100%	99.7%	?Mitochondrial myopathy with lactic acidosis, 251950	INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS MITOCHONDRIAL DISORDERS PANEL COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹
PNPO	100%	100%	100%	99.5%	Pyridoxamine 5'-phosphate oxidase deficiency, 610090	EPILEPSY PANEL METABOLIC DISORDERS PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹

PNPT1	100%	100%	100%	99.7%	Spinocerebellar ataxia 25, 608703;Deafness, autosomal recessive 70, with or without adult-onset neurodegeneration, 614934;Combined oxidative phosphorylation deficiency 13, 614932	MOVEMENT DISORDERS PANEL HEARING IMPAIRMENT PANEL (INCLUDING GJB2) POLYNEUROPATHIES PANEL ¹ INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS MITOCHONDRIAL DISORDERS PANEL COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹
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POC1A	100%	100%	100%	99.5%	Short stature, onychodysplasia, facial dysmorphism, and hypotrichosis, 614813	CILIOPATHIES PANEL SKIN DISORDERS PANEL ¹ SHORT STATURE/SKELETAL DYSPLASIA PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹
POC1B	100%	100%	100%	99.3%	Cone-rod dystrophy 20, 615973	VISION DISORDERS PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹
POC5	100%	100%	100%	99.7%		VISION DISORDERS PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS

PODXL	93.8%	93.8%	100%	98.6%		RENAL DISORDERS PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS
POF1B	100%	99.9%	99.1%	68.9%	?Premature ovarian failure 2B, 300604	MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS
POFUT1	100%	100%	100%	99%	Dowling-Degos disease 2, 615327	SKIN DISORDERS PANEL ¹ METABOLIC DISORDERS PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS

POGLUT1	100%	100%	100%	99.4%	Dowling-Degos disease 4, 615696;Muscular dystrophy, limb-girdle, autosomal recessive 21, 617232	SKIN DISORDERS PANEL ¹ METABOLIC DISORDERS PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹ MUSCLE DISORDERS PANEL
POGZ	100%	99.7%	100%	98.9%	White-Sutton syndrome, 616364	INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS
POLA1	100%	99.9%	99%	69.9%	Pigmentary disorder, reticulate, with systemic manifestations, X-linked, 301220;Van Esch-O'Driscoll syndrome, 301030	PRIMARY IMMUNODEFICIENCIES PANEL INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS

POLD1	100%	100%	100%	98%	Mandibular hypoplasia, deafness, progeroid features, and lipodystrophy syndrome, 615381;Immunodeficiency 120, 620836;{Colorectal cancer, susceptibility to, 10}, 612591	PANEL HEREDITARY COLORECTAL AND POLYPOSIS SKIN DISORDERS PANEL ¹ HEARING IMPAIRMENT PANEL (INCLUDING GJB2) PRIMARY IMMUNODEFICIENCIES PANEL METABOLIC DISORDERS PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS HEREDITARY CANCER PANEL
POLD3	100%	100%	100%	99.5%	Immunodeficiency 122, 620869	PRIMARY IMMUNODEFICIENCIES PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS SEVERE COMBINED IMMUNODEFICIENCY (SCID) PANEL

POLE	100%	100%	100%	99%	{Colorectal cancer, susceptibility to, 12}, 615083;FILS syndrome, 615139;IMAGE-I syndrome, 618336	PANEL HEREDITARY COLORECTAL AND POLYPOSIS DISORDERS/DIFFERENCES OF SEX DEVELOPMENT (DSD) / PRIMARY ADRENAL INSUFFICIENCY PANEL SHORT STATURE/SKELETAL DYSPLASIA PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹ HEREDITARY CANCER PANEL
POLE2	100%	100%	100%	99.7%		PRIMARY IMMUNODEFICIENCIES PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS

POLG	100%	100%	100%	99.2%	Mitochondrial recessive ataxia syndrome (includes SANDO and SCAE), 607459;Mitochondrial DNA depletion syndrome 4B (MNGIE type), 613662;Mitochondrial DNA depletion syndrome 4A (Alpers type), 203700;Progressive external ophthalmoplegia, autosomal dominant 1, 157640;Progressive external ophthalmoplegia, autosomal recessive 1, 258450	MOVEMENT DISORDERS PANEL PARKINSON DISEASE PANEL COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹ PREMATURE OVARIAN INSUFFICIENCY PANEL MUSCLE DISORDERS PANEL INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS MITOCHONDRIAL DISORDERS PANEL HYPOGONADOTROPIC HYPOGONADISM (KALLMANN) PANEL POLYNEUROPATHIES PANEL ¹ LIVER DISORDERS PANEL VISION DISORDERS PANEL DISORDERS/DIFFEREN CES OF SEX DEVELOPMENT (DSD) / PRIMARY ADRENAL INSUFFICIENCY PANEL
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						EPILEPSY PANEL
POLG2	100%	100%	100%	99.1%	Progressive external ophthalmoplegia with mitochondrial DNA deletions, autosomal dominant 4, 610131;?Mitochondrial DNA depletion syndrome 16 (hepatic type), 618528;?Mitochondrial DNA depletion syndrome 16B (neuroophthalmic type), 619425	VISION DISORDERS PANEL POLYNEUROPATHIES PANEL ¹ MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS MITOCHONDRIAL DISORDERS PANEL MUSCLE DISORDERS PANEL
POLH	100%	100%	100%	99.5%	Xeroderma pigmentosum, variant type, 278750	SKIN DISORDERS PANEL ¹ MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹ HEREDITARY CANCER PANEL

POLL	100%	100%	100%	99.5%		SHORT STATURE/SKELETAL DYSPLASIA PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS
POLR1A	100%	100%	100%	99.4%	Leukodystrophy, hypomyelinating, 27, 620675;Acrofacial dysostosis, Cincinnati type, 616462	SHORT STATURE/SKELETAL DYSPLASIA PANEL WITH GENOME WIDE CNV ANALYSIS INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS OROFACIAL CLEFTING PANEL WITH GENOME WIDE CNV ANALYSIS

POLR1B	100%	100%	100%	99.4%	Treacher-Collins syndrome 4, 618939	CRANIOFACIAL ANOMALIES PANEL WITH GENOME WIDE CNV ANALYSIS SHORT STATURE/SKELETAL DYSPLASIA PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS
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POLR1C	83.4%	83.2%	100%	98.6%	Leukodystrophy, hypomyelinating, 11, 616494;Treacher Collins syndrome 3, 248390	MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹ OROFACIAL CLEFTING PANEL WITH GENOME WIDE CNV ANALYSIS MOVEMENT DISORDERS PANEL CRANIOFACIAL ANOMALIES PANEL WITH GENOME WIDE CNV ANALYSIS SKIN DISORDERS PANEL ¹ HEARING IMPAIRMENT PANEL (INCLUDING GJB2) SHORT STATURE/SKELETAL DYSPLASIA PANEL WITH GENOME WIDE CNV ANALYSIS INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS
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POLR1D	100%	100%	100%	99.6%	Treacher Collins syndrome 2, 613717	CRANIOFACIAL ANOMALIES PANEL WITH GENOME WIDE CNV ANALYSIS SKIN DISORDERS PANEL ¹ HEARING IMPAIRMENT PANEL (INCLUDING GJB2) SHORT STATURE/SKELETAL DYSPLASIA PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹ OROFACIAL CLEFTING PANEL WITH GENOME WIDE CNV ANALYSIS
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POLR2A	100%	100%	100%	98.1%	Neurodevelopmental disorder with hypotonia and variable intellectual and behavioral abnormalities, 618603	MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MITOCHONDRIAL DISORDERS PANEL
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POLR3A	100%	100%	100%	99.1%	Wiedemann-Rautenstrauch syndrome, 264090;Leukodystrophy, hypomyelinating, 7, with or without oligodontia and/or hypogonadotropic hypogonadism, 607694	MOVEMENT DISORDERS PANEL SKIN DISORDERS PANEL ¹ DISORDERS/DIFFEREN CES OF SEX DEVELOPMENT (DSD) / PRIMARY ADRENAL INSUFFICIENCY PANEL INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹ HYPOGONADOTROPIC HYPOGONADISM (KALLMANN) PANEL SHORT STATURE/SKELETAL DYSPLASIA PANEL WITH GENOME WIDE CNV ANALYSIS METABOLIC DISORDERS PANEL
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POLR3B	100%	100%	100%	99.3%	Leukodystrophy, hypomyelinating, 8, with or without oligodontia and/or hypogonadotropic hypogonadism, 614381;Charcot-Marie-Tooth disease, demyelinating, type 1I, 619742	MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹ HYPOGONADOTROPIC HYPOGONADISM (KALLMANN) PANEL POLYNEUROPATHIES PANEL ¹ SHORT STATURE/SKELETAL DYSPLASIA PANEL WITH GENOME WIDE CNV ANALYSIS METABOLIC DISORDERS PANEL INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MOVEMENT DISORDERS PANEL SKIN DISORDERS PANEL ¹ DISORDERS/DIFFEREN CES OF SEX DEVELOPMENT (DSD) / PRIMARY ADRENAL INSUFFICIENCY PANEL
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POLR3F	92.6%	90.7%	100%	99.3%	?Immunodeficiency 101 (varicella zoster virus-specific), 619872	PRIMARY IMMUNODEFICIENCIES PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS
POLR3GL	100%	100%	100%	99.6%	Short stature, oligodontia, dysmorphic facies, and motor delay, 619234	DISORDERS/DIFFEREN CES OF SEX DEVELOPMENT (DSD) / PRIMARY ADRENAL INSUFFICIENCY PANEL HYPOGONADOTROPIC HYPOGONADISM (KALLMANN) PANEL SHORT STATURE/SKELETAL DYSPLASIA PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹

POLR3K	100%	100%	100%	99.4%	Leukodystrophy, hypomyelinating, 21, 619310	INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS
POLRMT	100%	100%	100%	98.2%	Combined oxidative phosphorylation deficiency 55, 619743	INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS MITOCHONDRIAL DISORDERS PANEL

POMC	100%	100%	100%	98.4%	{Obesity, early-onset, susceptibility to}, 601665;Obesity, adrenal insufficiency, and red hair due to POMC deficiency, 609734	SKIN DISORDERS PANEL ¹ DISORDERS/DIFFERENCES OF SEX DEVELOPMENT (DSD) / PRIMARY ADRENAL INSUFFICIENCY PANEL COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹ MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS LIVER DISORDERS PANEL
POMGNT1	100%	100%	100%	99.1%	Muscular dystrophy-dystroglycanopathy (limb-girdle), type C, 3, 613157;Muscular dystrophy-dystroglycanopathy (congenital with impaired intellectual development), type B, 3, 613151;Retinitis pigmentosa 76, 617123;Muscular dystrophy-dystroglycanopathy (congenital with brain and eye anomalies), type A, 3, 253280	VISION DISORDERS PANEL METABOLIC DISORDERS PANEL INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹ MUSCLE DISORDERS PANEL

POMGNT2	100%	100%	100%	99%	Muscular dystrophy-dystroglycanopathy (congenital with brain and eye anomalies), type A, 8, 614830;Muscular dystrophy-dystroglycanopathy (limb-girdle) type C, 8, 618135	METABOLIC DISORDERS PANEL INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹ MUSCLE DISORDERS PANEL
POMK	100%	100%	100%	98.8%	?Muscular dystrophy-dystroglycanopathy (limb-girdle), type C, 12, 616094;Muscular dystrophy-dystroglycanopathy (congenital with brain and eye anomalies), type A, 12, 615249	METABOLIC DISORDERS PANEL INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹ MUSCLE DISORDERS PANEL

POMP	83.2%	83.2%	100%	99.7%	Proteasome-associated autoinflammatory syndrome 2, 618048;Keratosis linearis with ichthyosis congenita and sclerosing keratoderma, 601952	SKIN DISORDERS PANEL ¹ PRIMARY IMMUNODEFICIENCIES PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹
POMT1	100%	100%	100%	98.9%	Muscular dystrophy-dystroglycanopathy (congenital with brain and eye anomalies), type A, 1, 236670;Muscular dystrophy-dystroglycanopathy (limb-girdle), type C, 1, 609308;Muscular dystrophy-dystroglycanopathy (congenital with impaired intellectual development), type B, 1, 613155	HEART DISORDERS PANEL ¹ METABOLIC DISORDERS PANEL INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹ OROFACIAL CLEFTING PANEL WITH GENOME WIDE CNV ANALYSIS MUSCLE DISORDERS PANEL

POMT2	100%	100%	99.9%	98.3%	Muscular dystrophy-dystroglycanopathy (limb-girdle), type C, 2, 613158;Muscular dystrophy-dystroglycanopathy (congenital with brain and eye anomalies), type A, 2, 613150;Muscular dystrophy-dystroglycanopathy (congenital with impaired intellectual development), type B, 2, 613156	HEART DISORDERS PANEL ¹ METABOLIC DISORDERS PANEL INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹ MUSCLE DISORDERS PANEL
POP1	100%	100%	100%	99.2%	Anauxetic dysplasia 2, 617396	SHORT STATURE/SKELETAL DYSPLASIA PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹

POPDC2	100%	100%	100%	99%		CONGENITAL HEARTDISEASE PANEL¹ HEART DISORDERS PANEL¹ MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS
POPDC3	100%	100%	100%	99.4%	Muscular dystrophy, limb-girdle, autosomal recessive 26, 618848	MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL¹ MUSCLE DISORDERS PANEL

POR	100%	100%	100%	99.2%	Antley-Bixler syndrome with genital anomalies and disordered steroidogenesis, 201750;Disordered steroidogenesis due to cytochrome P450 oxidoreductase, 613571	CRANIOFACIAL ANOMALIES PANEL WITH GENOME WIDE CNV ANALYSIS DISORDERS/DIFFERENCES OF SEX DEVELOPMENT (DSD) / PRIMARY ADRENAL INSUFFICIENCY PANEL SHORT STATURE/SKELETAL DYSPLASIA PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹
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PORCN	100%	99.7%	98.5%	66%	Focal dermal hypoplasia, 305600	CRANIOFACIAL ANOMALIES PANEL WITH GENOME WIDE CNV ANALYSIS SKIN DISORDERS PANEL ¹ SHORT STATURE/SKELETAL DYSPLASIA PANEL WITH GENOME WIDE CNV ANALYSIS INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS OROFACIAL CLEFTING PANEL WITH GENOME WIDE CNV ANALYSIS
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POT1	100%	100%	100%	99.5%	Tumor predisposition syndrome 3, 615848;?Cerebroretinal microangiopathy with calcifications and cysts 3, 620368;?Pulmonary fibrosis and/or bone marrow failure syndrome, telomere-related, 8, 620367	INHERITED BONE MARROW FAILURE AND/OR PREDISPOSITION PANEL TO HEMATOLOGICAL MALIGNANCIES SKIN DISORDERS PANEL ¹ PRIMARY IMMUNODEFICIENCIES PANEL PANEL MELANOMA, EXTENSIVE (CDKN2A, CDK4, MITF P.(GLU318LYS), BAP1, POT1, TERT PROMOTER) ¹ MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS HEREDITARY CANCER PANEL DYSKERATOSIS CONGENITA AND APLASTIC ANEMIA PANEL
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POU1F1	100%	100%	100%	99.6%	Pituitary hormone deficiency, combined or isolated, 1, 613038	SHORT STATURE/SKELETAL DYSPLASIA PANEL WITH GENOME WIDE CNV ANALYSIS INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL¹
POU2AF1	100%	100%	100%	98.8%		PRIMARY IMMUNODEFICIENCIES PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS
POU3F2	100%	100%	100%	96%		INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS

POU3F3	96.9%	85.7%	99.6%	80%	Snijders Blok-Fisher syndrome, 618604	INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS
POU3F4	100%	100%	97.5%	62%	Deafness, X-linked 2, 304400	HEARING IMPAIRMENT PANEL (INCLUDING GJB2) MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS
POU4F1	90.8%	82.1%	99.9%	93%	Ataxia, intention tremor, and hypotonia syndrome, childhood-onset, 619352	MOVEMENT DISORDERS PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS
POU4F3	100%	100%	100%	99.3%	Deafness, autosomal dominant 15/52, 602459	HEARING IMPAIRMENT PANEL (INCLUDING GJB2) MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS

POU6F2	100%	100%	100%	98.9%	{Wilms tumor susceptibility-5}, 601583	MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS HEREDITARY CANCER PANEL
PPA2	100%	100%	100%	99.6%	?Sudden cardiac failure, alcohol-induced, 617223;Sudden cardiac failure, infantile, 617222	HEART DISORDERS PANEL ¹ MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS MITOCHONDRIAL DISORDERS PANEL COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹
PPARG	100%	100%	100%	99.2%	{Diabetes, type 2}, 125853;Insulin resistance, severe, digenic, 125853;Lipodystrophy, familial partial, type 3, 604367;Obesity, severe, 601665	METABOLIC DISORDERS PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS

PPCDC	100%	100%	100%	99.5%		HEART DISORDERS PANEL ¹ METABOLIC DISORDERS PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS
PPCS	100%	100%	100%	99.3%	Cardiomyopathy, dilated, 2C, 618189	HEART DISORDERS PANEL ¹ METABOLIC DISORDERS PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS MITOCHONDRIAL DISORDERS PANEL COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹
PPFIA3	100%	100%	100%	97.4%		METABOLIC DISORDERS PANEL INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS

PPFIBP1	100%	100%	100%	99.6%	Neurodevelopmental disorder with seizures, microcephaly, and brain abnormalities, 620024	EPILEPSY PANEL INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹
PPIB	100%	100%	100%	99.4%	Osteogenesis imperfecta, type IX, 259440	SHORT STATURE/SKELETAL DYSPLASIA PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹

PPIL1	100%	100%	100%	99.6%	Pontocerebellar hypoplasia, type 14, 619301	INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹
PPIP5K2	100%	100%	100%	99.2%	Deafness, autosomal recessive 100, 618422	HEARING IMPAIRMENT PANEL (INCLUDING GJB2) MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹

PPM1D	100%	100%	100%	99.3%	Breast cancer, somatic, 114480;Jansen-de Vries syndrome, 617450	SHORT STATURE/SKELETAL DYSPLASIA PANEL WITH GENOME WIDE CNV ANALYSIS INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS
PPM1K	100%	100%	100%	99.4%	Maple syrup urine disease, mild variant, 615135	METABOLIC DISORDERS PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL¹

PPOX	100%	100%	100%	98.6%	Variegate porphyria, childhood-onset, 620483;Variegate porphyria, 176200	SKIN DISORDERS PANEL ¹ METABOLIC DISORDERS PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹
PPP1CB	87.9%	87.3%	100%	99.2%	Noonan syndrome-like disorder with loose anagen hair 2, 617506	SHORT STATURE/SKELETAL DYSPLASIA PANEL WITH GENOME WIDE CNV ANALYSIS LYMPHATIC ANOMALIES PANEL WITH GENOME WIDE CNV ANALYSIS INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS NOONAN SYNDROME / RASOPATHY PANEL

PPP1R12A	100%	100%	100%	99.5%	Genitourinary and/or/brain malformation syndrome, 618820	DISORDERS/DIFFERENCES OF SEX DEVELOPMENT (DSD) / PRIMARY ADRENAL INSUFFICIENCY PANEL INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS
PPP1R13L	100%	99.9%	100%	95%	Arrhythmogenic cardiomyopathy with variable ectodermal abnormalities, 620519	CONGENITAL HEARTDISEASE PANEL ¹ HEART DISORDERS PANEL ¹ MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS
PPP1R15B	100%	100%	100%	99.6%	Microcephaly, short stature, and impaired glucose metabolism 2, 616817	INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹

PPP1R21	100%	100%	100%	99.2%	Neurodevelopmental disorder with hypotonia, facial dysmorphism, and brain abnormalities, 619383	SHORT STATURE/SKELETAL DYSPLASIA PANEL WITH GENOME WIDE CNV ANALYSIS INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹
PPP1R3A	100%	100%	100%	99.2%	Insulin resistance, severe, digenic, 125853	MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS
PPP1R3F	100%	99.9%	98.2%	66.9%		MOVEMENT DISORDERS PANEL INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS

PPP2CA	100%	100%	100%	99.3%	Houge-Janssens syndrome 3, 618354	INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS
PPP2R1A	93.9%	93.9%	100%	99.2%	Houge-Janssens syndrome 2, 616362	EPILEPSY PANEL INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS
PPP2R1B	100%	100%	100%	99.4%	Lung cancer, somatic, 211980	MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS
PPP2R2B	100%	100%	100%	98.9%	Spinocerebellar ataxia 12, 604326	INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS

PPP2R3C	100%	100%	100%	99.8%	Spermatogenic failure 36, 618420;Myoectodermal gonadal dysgenesis syndrome, 618419	DISORDERS/DIFFERENCES OF SEX DEVELOPMENT (DSD) / PRIMARY ADRENAL INSUFFICIENCY PANEL COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹ INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS
PPP2R5B	100%	100%	100%	99.3%		INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS
PPP2R5C	100%	99.9%	100%	98.9%		INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS

PPP2R5D	100%	100%	100%	98.7%	Houge-Janssens syndrome 1, 616355	EPILEPSY PANEL INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS
PPP3CA	100%	100%	100%	99.5%	Arthrogryposis, cleft palate, craniosynostosis, and impaired intellectual development, 618265;Developmental and epileptic encephalopathy 91, 617711	EPILEPSY PANEL INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS

PPT1	90.3%	90.3%	100%	99.4%	Ceroid lipofuscinosis, neuronal, 1, 256730	MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹ MOVEMENT DISORDERS PANEL VISION DISORDERS PANEL EPILEPSY PANEL METABOLIC DISORDERS PANEL INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS
PQBP1	100%	100%	98.1%	65.5%	Renpenning syndrome, 309500	SKIN DISORDERS PANEL ¹ EPILEPSY PANEL INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS OROFACIAL CLEFTING PANEL WITH GENOME WIDE CNV ANALYSIS

PRCC	100%	100%	100%	98.9%	Renal cell carcinoma, papillary, 605074	MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS
PRCD	100%	100%	100%	99.1%	Retinitis pigmentosa 36, 610599	VISION DISORDERS PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹
PRDM10	100%	100%	100%	99.2%	?Birt-Hogg-Dube syndrome 2, 620459	SKIN DISORDERS PANEL ¹ MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS PANEL HEREDITARY RENALCANCER HEREDITARY CANCER PANEL

PRDM12	93.6%	91.1%	100%	96.8%	Neuropathy, hereditary sensory and autonomic, type VIII, 616488	SKIN DISORDERS PANEL ¹ POLYNEUROPATHIES PANEL ¹ NEUROLOGICAL PAIN DISORDERS PANEL ¹ MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹
PRDM13	100%	100%	100%	99.1%	Pontocerebellar hypoplasia, type 17, 619909;Cerebellar dysfunction, impaired intellectual development, and hypogonadotropic hypogonadism, 619761	VISION DISORDERS PANEL INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹

PRDM15	100%	100%	100%	98.8%		INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS
PRDM16	100%	100%	100%	97.9%	Left ventricular noncompaction 8, 615373;Cardiomyopathy, dilated, 1LL, 615373	HEART DISORDERS PANEL ¹ MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS
PRDM5	100%	100%	100%	99.3%	Brittle cornea syndrome 2, 614170	VISION DISORDERS PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹
PRDM6	100%	100%	100%	97.4%	Patent ductus arteriosus 3, 617039	HEART DISORDERS PANEL ¹ MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS

PRDM8	100%	100%	100%	96.4%	?Epilepsy, progressive myoclonic, 10, 616640	MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS
PRDX1	100%	100%	100%	99.3%	Methylmalonic aciduria and homocystinuria, cblC type, digenic, 277400	MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹
PRDX2	100%	99.7%	100%	97.7%		INHERITED BONE MARROW FAILURE AND/OR PREDISPOSITION PANEL TO HEMATOLOGICAL MALIGNANCIES MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS

PRDX3	100%	100%	100%	99.7%	Spinocerebellar ataxia, autosomal recessive 32, 619862;Corneal dystrophy, punctiform and polychromatic pre-Descemet, 619871	MOVEMENT DISORDERS PANEL VISION DISORDERS PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS MITOCHONDRIAL DISORDERS PANEL COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹
PREPL	100%	100%	100%	99.5%	Myasthenic syndrome, congenital, 22, 616224	MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹ MUSCLE DISORDERS PANEL

PRF1	100%	100%	100%	99.6%	Hemophagocytic lymphohistiocytosis, familial, 2, 603553;Aplastic anemia, 609135;Lymphoma, non-Hodgkin, 605027	COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹ HEREDITARY CANCER PANEL MOVEMENT DISORDERS PANEL INHERITED BONE MARROW FAILURE AND/OR PREDISPOSITION PANEL TO HEMATOLOGICAL MALIGNANCIES EPILEPSY PANEL PRIMARY IMMUNODEFICIENCIES PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS
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PRG4	100%	100%	100%	93.9%	Camptodactyly-arthropathy-coxa vara-pericarditis syndrome, 208250	SHORT STATURE/SKELETAL DYSPLASIA PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹
PRICKLE1	100%	100%	100%	99.3%	Epilepsy, progressive myoclonic 1B, 612437	MOVEMENT DISORDERS PANEL EPILEPSY PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹
PRICKLE2	100%	100%	100%	99.1%		INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS

PRIM1	91.8%	91.8%	100%	99.3%	Primordial dwarfism-immunodeficiency-lipodystrophy syndrome, 620005	PRIMARY IMMUNODEFICIENCIES PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS
PRIMPOL	100%	100%	100%	99.6%	Myopia 22, autosomal dominant, 615420	VISION DISORDERS PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS
PRKAA1	100%	100%	100%	99.7%		MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS MITOCHONDRIAL DISORDERS PANEL
PRKACA	100%	100%	100%	98.3%	Cushing syndrome, ACTH-independent adrenal, somatic, 615830;Cardioacrofacial dysplasia 1, 619142	SHORT STATURE/SKELETAL DYSPLASIA PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS

PRKACB	100%	100%	100%	99.3%	Cardioacrofacial dysplasia 2, 619143	SHORT STATURE/SKELETAL DYSPLASIA PANEL WITH GENOME WIDE CNV ANALYSIS INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS
PRKACG	100%	100%	100%	96.7%	?Bleeding disorder, platelet-type, 19, 616176	HEMOSTATIC/THROMB OTIC DISORDERS PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS
PRKAG2	100%	100%	100%	98.9%	Glycogen storage disease of heart, lethal congenital, 261740;Wolff-Parkinson- White syndrome, 194200;Cardiomyopathy, hypertrophic 6, 600858	HEART DISORDERS PANEL ¹ HYPERTROPHIC CARDIOMYOPATHY PANEL ¹ METABOLIC DISORDERS PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS

PRKAR1A	100%	100%	100%	99.1%	Pigmented nodular adrenocortical disease, primary, 1, 610489;Acrodysostosis 1, with or without hormone resistance, 101800;Adrenocortical tumor, somatic;Carney complex, type 1, 160980;Myxoma, intracardiac, 255960	SKIN DISORDERS PANEL ¹ SHORT STATURE/SKELETAL DYSPLASIA PANEL WITH GENOME WIDE CNV ANALYSIS INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS HEREDITARY CANCER PANEL
PRKAR1B	100%	100%	100%	97.7%	Marbach-Schaaf neurodevelopmental syndrome, 619680	INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS
PRKCA	100%	100%	100%	99.2%	Pituitary tumor, invasive	MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS

PRKCB	100%	100%	100%	99.1%		HEARING IMPAIRMENT PANEL (INCLUDING GJB2) MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS
PRKCD	100%	100%	100%	99.3%	Autoimmune lymphoproliferative syndrome, type III, 615559	PRIMARY IMMUNODEFICIENCIES PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹
PRKCG	100%	100%	100%	96.2%	Spinocerebellar ataxia 14, 605361	MOVEMENT DISORDERS PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS

PRKCSH	100%	100%	100%	98.7%	Polycystic liver disease 1, 174050	LIVER DISORDERS PANEL METABOLIC DISORDERS PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS
PRKD1	100%	100%	100%	97.8%	Congenital heart defects and ectodermal dysplasia, 617364	CONGENITAL HEARTDISEASE PANEL¹ HEART DISORDERS PANEL¹ MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS
PRKDC	100%	100%	100%	99.5%	Immunodeficiency 26, with or without neurologic abnormalities, 615966	PRIMARY IMMUNODEFICIENCIES PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL¹ SEVERE COMBINED IMMUNODEFICIENCY (SCID) PANEL

PRKG1	95.9%	95.9%	100%	99.2%	Aortic aneurysm, familial thoracic 8, 615436	THORACIC AORTIC ANEURYSM AND/OR DISSECTION PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS
PRKG2	100%	100%	100%	99.5%	Spondylometaphyseal dysplasia, Pagnamenta type, 619638;Acromesomelic dysplasia 4, 619636	SHORT STATURE/SKELETAL DYSPLASIA PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹
PRKN	100%	100%	100%	98.5%	Adenocarcinoma of lung, somatic, 211980;Parkinson disease, juvenile, type 2, 600116;Ovarian cancer, somatic, 167000	MOVEMENT DISORDERS PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS PARKINSON DISEASE PANEL COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹ HEREDITARY CANCER PANEL

PRKRA	100%	100%	100%	99.6%	Dystonia 16, 612067	MOVEMENT DISORDERS PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS PARKINSON DISEASE PANEL COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹
PRLR	100%	100%	100%	99.6%	Multiple fibroadenomas of the breast, 615554;Hyperprolactinemia, 615555	MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS
PRMT7	100%	100%	100%	98.9%	Short stature, brachydactyly, intellectual developmental disability, and seizures, 617157	SHORT STATURE/SKELETAL DYSPLASIA PANEL WITH GENOME WIDE CNV ANALYSIS INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹

PRNP	100%	100%	100%	99%	Spongiform encephalopathy with neuropsychiatric features, 606688;Gerstmann-Straussler disease, 137440;Huntington disease-like 1, 603218;Insomnia, fatal familial, 600072;{Kuru, susceptibility to}, 245300;Cerebral amyloid angiopathy, PRNP-related, 137440;Creutzfeldt-Jakob disease, 123400	POLYNEUROPATHIES PANEL ¹ MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS
PROC	100%	100%	100%	99.5%	Thrombophilia 3 due to protein C deficiency, autosomal dominant, 176860;Thrombophilia 3 due to protein C deficiency, autosomal recessive, 612304	HEMOSTATIC/THROMB OTIC DISORDERS PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹
PRODH	100%	100%	100%	98.6%	{Schizophrenia, susceptibility to, 4}, 600850;Hyperprolinemia, type I, 239500	EPILEPSY PANEL METABOLIC DISORDERS PANEL INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹

PROK2	100%	100%	100%	99.1%	Hypogonadotropic hypogonadism 4 with or without anosmia, 610628	DISORDERS/DIFFERENCES OF SEX DEVELOPMENT (DSD) / PRIMARY ADRENAL INSUFFICIENCY PANEL HYPOGONADOTROPIC HYPOGONADISM (KALLMANN) PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS
PROKR2	100%	100%	100%	99.8%	Hypogonadotropic hypogonadism 3 with or without anosmia, 244200	DISORDERS/DIFFERENCES OF SEX DEVELOPMENT (DSD) / PRIMARY ADRENAL INSUFFICIENCY PANEL HYPOGONADOTROPIC HYPOGONADISM (KALLMANN) PANEL SHORT STATURE/SKELETAL DYSPLASIA PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS OROFACIAL CLEFTING PANEL WITH GENOME WIDE CNV ANALYSIS

PROM1	100%	100%	100%	99.6%	Macular dystrophy, retinal, 2, 608051;Retinitis pigmentosa 41, 612095;Stargardt disease 4, 603786;Cone-rod dystrophy 12, 612657	VISION DISORDERS PANEL COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹ MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS
PROP1	100%	100%	100%	97.8%	Pituitary hormone deficiency, combined, 2, 262600	DISORDERS/DIFFEREN CES OF SEX DEVELOPMENT (DSD) / PRIMARY ADRENAL INSUFFICIENCY PANEL HYPOGONADOTROPIC HYPOGONADISM (KALLMANN) PANEL SHORT STATURE/SKELETAL DYSPLASIA PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹

PRORP	100%	100%	100%	99.5%	Combined oxidative phosphorylation deficiency 54, 619737	HEARING IMPAIRMENT PANEL (INCLUDING GJB2) MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS MITOCHONDRIAL DISORDERS PANEL COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹
PROS1	100%	100%	100%	99.3%	Thrombophilia 5 due to protein S deficiency, autosomal recessive, 614514;Thrombophilia 5 due to protein S deficiency, autosomal dominant, 612336	HEMOSTATIC/THROMB OTIC DISORDERS PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹
PROZ	100%	100%	100%	98.7%	[Protein Z deficiency], 614024	HEMOSTATIC/THROMB OTIC DISORDERS PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS

PRPF19	100%	100%	100%	97.3%		INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS
PRPF3	100%	100%	100%	99.2%	Retinitis pigmentosa 18, 601414	VISION DISORDERS PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS
PRPF31	100%	100%	100%	98.8%	Retinitis pigmentosa 11, 600138	VISION DISORDERS PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS
PRPF4	100%	100%	100%	99.6%	Retinitis pigmentosa 70, 615922	VISION DISORDERS PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS

PRPF6	100%	100%	100%	99%	Retinitis pigmentosa 60, 613983	VISION DISORDERS PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS
PRPF8	100%	100%	100%	99.2%	Retinitis pigmentosa 13, 600059	VISION DISORDERS PANEL INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS
PRPH2	100%	100%	100%	98.1%	Macular dystrophy, patterned, 1, 169150;Choroidal dystrophy, central areolar 2, 613105;Retinitis punctata albescens, 136880;Leber congenital amaurosis 18, 608133;Macular dystrophy, vitelliform, 3, 608161;Retinitis pigmentosa 7 and digenic form, 608133	MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS VISION DISORDERS PANEL

PRPS1	100%	99.9%	98.5%	67.1%	Arts syndrome, 301835;Phosphoribosylpyrophosphate synthetase superactivity, 300661;Charcot-Marie-Tooth disease, X-linked recessive, 5, 311070;Deafness, X-linked 1, 304500;Gout, PRPS-related, 300661	VISION DISORDERS PANEL HEARING IMPAIRMENT PANEL (INCLUDING GJB2) POLYNEUROPATHIES PANEL ¹ PRIMARY IMMUNODEFICIENCIES PANEL MUSCLE DISORDERS PANEL METABOLIC DISORDERS PANEL INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS MITOCHONDRIAL DISORDERS PANEL
PRR11	100%	100%	100%	98.9%		VISION DISORDERS PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS

PRR12	100%	100%	100%	97.1%	Neuroocular syndrome, 619539	VISION DISORDERS PANEL INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS
PRRT2	100%	100%	100%	98%	Convulsions, familial infantile, with paroxysmal choreoathetosis, 602066;Seizures, benign familial infantile, 2, 605751;Episodic kinesigenic dyskinesia 1, 128200	MOVEMENT DISORDERS PANEL EPILEPSY PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS
PRRX1	100%	100%	100%	99%	Agnathia-otocephaly complex, 202650	CRANIOFACIAL ANOMALIES PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS OROFACIAL CLEFTING PANEL WITH GENOME WIDE CNV ANALYSIS

PRSS1	100%	100%	100%	96.9%	Pancreatitis, hereditary, 167800	MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS HEREDITARY CANCER PANEL
PRSS12	100%	100%	100%	98.9%	Intellectual developmental disorder, autosomal recessive 1, 249500	INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹
PRSS56	100%	100%	100%	98.9%	Microphthalmia, isolated 6, 613517	VISION DISORDERS PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹

PRUNE1	93.1%	93.1%	100%	99.3%	Neurodevelopmental disorder with microcephaly, hypotonia, and variable brain anomalies, 617481	INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹
PRX	100%	100%	100%	98.8%	Charcot-Marie-Tooth disease, type 4F, 614895;Dejerine-Sottas disease, 145900	POLYNEUROPATHIES PANEL ¹ MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹
PRY	50%	50%	49.1%	28.7%		MALE INFERTILITY PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS

PRY2	50%	50%	49.9%	32.2%		MALE INFERTILITY PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS
PSAP	100%	100%	100%	99.2%	Combined SAP deficiency, 611721;Krabbe disease, atypical, 611722;Metachromatic leukodystrophy due to SAP-b deficiency, 249900;Gaucher disease, atypical, 610539;{Parkinson disease 24, autosomal dominant, susceptibility to}, 619491	MOVEMENT DISORDERS PANEL EPILEPSY PANEL POLYNEUROPATHIES PANEL ¹ METABOLIC DISORDERS PANEL INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹

PSAT1	100%	100%	100%	99.8%	Neu-Laxova syndrome 2, 616038;Phosphoserine aminotransferase deficiency, 610992	FETAL AKINESIA PANEL SKIN DISORDERS PANEL ¹ SHORT STATURE/SKELETAL DYSPLASIA PANEL WITH GENOME WIDE CNV ANALYSIS METABOLIC DISORDERS PANEL INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹
PSEN1	100%	100%	100%	99.6%	Pick disease, 172700;Dementia, frontotemporal, 600274;?Acne inversa, familial, 3, 613737;?Cardiomyopathy, dilated, 1U, 613694;Alzheimer disease, type 3, with or without spastic paraparesis, 607822	SKIN DISORDERS PANEL ¹ MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS PARKINSON DISEASE PANEL

PSEN2	73.5%	73.5%	100%	98.9%	Alzheimer disease-4, 606889;Cardiomyopathy, dilated, 1V, 613697	MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS
PSENEN	100%	100%	100%	94.8%	Acne inversa, familial, 2, with or without Dowling-Degos disease, 613736	SKIN DISORDERS PANEL ¹ PRIMARY IMMUNODEFICIENCIES PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS
PSIP1	100%	100%	100%	99.2%		HEARING IMPAIRMENT PANEL (INCLUDING GJB2) MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS
PSKH1	100%	100%	100%	99.6%	Cholestasis, progressive familial intrahepatic, 13, 620962	MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS

PSMA3	100%	100%	100%	99.2%		PRIMARY IMMUNODEFICIENCIES PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS
PSMB1	100%	100%	100%	99.7%	?Neurodevelopmental disorder with microcephaly, hypotonia, and absent language, 620038	SHORT STATURE/SKELETAL DYSPLASIA PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS
PSMB10	100%	100%	100%	99.4%	Immunodeficiency 121 with autoinflammation, 620807;Proteasome-associated autoinflammatory syndrome 5, 619175	PRIMARY IMMUNODEFICIENCIES PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹ SEVERE COMBINED IMMUNODEFICIENCY (SCID) PANEL

PSMB4	100%	100%	100%	99.7%	?Proteasome-associated autoinflammatory syndrome 3 and digenic forms, 617591	PRIMARY IMMUNODEFICIENCIES PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹
PSMB8	100%	100%	100%	98.1%	Proteasome-associated autoinflammatory syndrome 1 and digenic forms, 256040	SKIN DISORDERS PANEL ¹ PRIMARY IMMUNODEFICIENCIES PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹
PSMB9	100%	100%	100%	99%	Proteasome-associated autoinflammatory syndrome 6, 620796	PRIMARY IMMUNODEFICIENCIES PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹

PSMC1	100%	100%	100%	99.6%	?Birk-Aharoni syndrome, 620071	MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS
PSMC3	100%	100%	100%	98.8%	?Deafness, cataract, impaired intellectual development, and polyneuropathy, 619354	INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS
PSMC3IP	100%	100%	100%	99.8%	Ovarian dysgenesis 3, 614324	DISORDERS/DIFFEREN CES OF SEX DEVELOPMENT (DSD) / PRIMARY ADRENAL INSUFFICIENCY PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹ PREMATURE OVARIAN INSUFFICIENCY PANEL

PSMC5	100%	100%	100%	99.4%		INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS
PSMD11	100%	100%	100%	99.2%		INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS
PSMD12	100%	100%	100%	99.4%	Stankiewicz-Isidor syndrome, 617516	MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS

PSMG2	88.4%	88.4%	100%	99.9%	?Proteasome-associated autoinflammatory syndrome 4, 619183	PRIMARY IMMUNODEFICIENCIES PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS
PSPH	100%	100%	100%	99.1%	Phosphoserine phosphatase deficiency, 614023	SKIN DISORDERS PANEL ¹ METABOLIC DISORDERS PANEL INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹
PSTPIP1	100%	100%	100%	99.2%	Autoinflammatory syndrome with cytopenia, hyperzincemia, and hypercalprotectinemia, 601979;Pyogenic sterile arthritis, pyoderma gangrenosum, and acne, 604416	SKIN DISORDERS PANEL ¹ PRIMARY IMMUNODEFICIENCIES PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS

PTBP1	100%	100%	100%	98.5%		SHORT STATURE/SKELETAL DYSPLASIA PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS
PTCD3	100%	100%	100%	99.2%	Combined oxidative phosphorylation deficiency 51, 619057	INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS MITOCHONDRIAL DISORDERS PANEL COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL¹

PTCH1	100%	100%	100%	98.7%	Basal cell nevus syndrome 1, 109400;Basal cell carcinoma, somatic, 605462;Holoprosencephaly 7, 610828	CRANIOFACIAL ANOMALIES PANEL WITH GENOME WIDE CNV ANALYSIS SKIN DISORDERS PANEL ¹ INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS HEREDITARY CANCER PANEL OROFACIAL CLEFTING PANEL WITH GENOME WIDE CNV ANALYSIS SONIC HEDGEHOG MEDULLOBLASTOMA PANEL
PTCH2	100%	100%	100%	99.1%	Medulloblastoma, somatic, 155255;Basal cell carcinoma, somatic, 605462	SKIN DISORDERS PANEL ¹ MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS OROFACIAL CLEFTING PANEL WITH GENOME WIDE CNV ANALYSIS

PTCHD1	100%	99.8%	99.2%	68.7%	{Autism, susceptibility to, X-linked 4}, 300830	VISION DISORDERS PANEL INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS
PTCRA	100%	100%	100%	99.4%	Immunodeficiency 126, 620931	PRIMARY IMMUNODEFICIENCIES PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS
PTDSS1	100%	100%	100%	99.4%	Lenz-Majewski hyperostotic dwarfism, 151050	SKIN DISORDERS PANEL ¹ SHORT STATURE/SKELETAL DYSPLASIA PANEL WITH GENOME WIDE CNV ANALYSIS INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS

PTEN	94.5%	94.5%	100%	99.2%	{Glioma susceptibility 2}, 613028;{Meningioma}, 607174;Cowden syndrome 1, 158350;Lhermitte-Duclos disease, 158350;Prostate cancer, somatic, 176807;Macrocephaly/autism syndrome, 605309	PANEL HEREDITARY COLORECTAL AND POLYPOSIS HEREDITARY CANCER PANEL INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS PANEL HEREDITARY RENALCANCER PRIMARY IMMUNODEFICIENCIES PANEL METABOLIC DISORDERS PANEL SKIN DISORDERS PANEL ¹
PTF1A	100%	99.7%	100%	98.3%	Pancreatic and cerebellar agenesis, 609069;Pancreatic agenesis 2, 615935	INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹

PTGIS	100%	100%	100%	99%	Hypertension, essential, 145500	METABOLIC DISORDERS PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS
PTGS1	100%	100%	100%	98.9%		HEMOSTATIC/THROMB OTIC DISORDERS PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS
PTH	100%	100%	100%	99.5%	Hypoparathyroidism, familial isolated 1, 146200	MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS

PTH1R	100%	100%	100%	98.7%	Metaphyseal chondrodysplasia, Murk Jansen type, 156400;Eiken syndrome, 600002;Failure of tooth eruption, primary, 125350;Chondrodysplasia, Blomstrand type, 215045	CRANIOFACIAL ANOMALIES PANEL WITH GENOME WIDE CNV ANALYSIS SHORT STATURE/SKELETAL DYSPLASIA PANEL WITH GENOME WIDE CNV ANALYSIS RENAL DISORDERS PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹
PTHLH	100%	100%	100%	99.7%	Brachydactyly, type E2, 613382	SKIN DISORDERS PANEL ¹ SHORT STATURE/SKELETAL DYSPLASIA PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS

PTPA	100%	100%	100%	98.2%	Parkinson disease 25, autosomal recessive early-onset, with impaired intellectual development, 620482	INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS
PTPMT1	100%	100%	100%	98.7%		INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS MITOCHONDRIAL DISORDERS PANEL

PTPN11	89.4%	89.2%	100%	99.5%	Noonan syndrome 1, 163950;LEOPARD syndrome 1, 151100;Metachondromatosis, 156250;Leukemia, juvenile myelomonocytic, somatic, 607785	HEREDITARY CANCER PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS NOONAN SYNDROME / RASOPATHY PANEL METABOLIC DISORDERS PANEL INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS SHORT STATURE/SKELETAL DYSPLASIA PANEL WITH GENOME WIDE CNV ANALYSIS LYMPHATIC ANOMALIES PANEL WITH GENOME WIDE CNV ANALYSIS HYPERTROPHIC CARDIOMYOPATHY PANEL ¹ HEMOSTATIC/THROMB OTIC DISORDERS PANEL SKIN DISORDERS PANEL ¹ HEART DISORDERS PANEL ¹ INHERITED BONE MARROW FAILURE
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						AND/OR PREDISPOSITION PANEL TO HEMATOLOGICAL MALIGNANCIES CONGENITAL HEARTDISEASE PANEL ¹
PTPN12	100%	100%	100%	99.5%	Colon cancer, somatic, 114500	MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS
PTPN14	100%	100%	100%	99.2%	Choanal atresia and lymphedema, 613611	SKIN DISORDERS PANEL ¹ LYMPHATIC ANOMALIES PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹

PTPN22	100%	100%	100%	99.8%	{Rheumatoid arthritis, susceptibility to}, 180300;{Systemic lupus erythematosus susceptibility to}, 152700;{Diabetes, type 1, susceptibility to}, 222100	HEMOSTATIC/THROMBOTIC DISORDERS PANEL PRIMARY IMMUNODEFICIENCIES PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS
PTPN23	100%	99.9%	100%	98%	Neurodevelopmental disorder and structural brain anomalies with or without seizures and spasticity, 618890	INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹
PTPN4	100%	100%	100%	99.7%		INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS

PTPRC	100%	100%	100%	99.6%	Immunodeficiency 105, severe combined, 619924	PRIMARY IMMUNODEFICIENCIES PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹ SEVERE COMBINED IMMUNODEFICIENCY (SCID) PANEL
PTPRF	100%	100%	100%	99.2%	?Breasts and/or nipples, aplasia or hypoplasia of, 2, 616001	SKIN DISORDERS PANEL ¹ MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS
PTPRJ	100%	99.9%	100%	98.7%	Colon cancer, somatic, 114500;Thrombocytopenia 10, 620484	HEMOSTATIC/THROMB OTIC DISORDERS PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS

PTPRO	100%	100%	100%	99.5%	Nephrotic syndrome, type 6, 614196	RENAL DISORDERS PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹
PTPRQ	100%	100%	100%	99.6%	Deafness, autosomal dominant 73, 617663;Deafness, autosomal recessive 84A, 613391	HEARING IMPAIRMENT PANEL (INCLUDING GJB2) MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹

PTRH2	100%	100%	100%	99.6%	Infantile-onset multisystem neurologic, endocrine, and pancreatic disease, 616263	FETAL AKINESIA PANEL MOVEMENT DISORDERS PANEL HEARING IMPAIRMENT PANEL (INCLUDING GJB2) EPILEPSY PANEL INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS MITOCHONDRIAL DISORDERS PANEL COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹ MUSCLE DISORDERS PANEL
PTRHD1	100%	100%	100%	99.9%	Neurodevelopmental disorder with early-onset parkinsonism and behavioral abnormalities, 620747	INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS

PTS	100%	100%	100%	98.8%	Hyperphenylalaninemia, BH4-deficient, A, 261640	MOVEMENT DISORDERS PANEL EPILEPSY PANEL METABOLIC DISORDERS PANEL INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL¹
PUF60	100%	100%	100%	99.3%	Verheij syndrome, 615583	SHORT STATURE/SKELETAL DYSPLASIA PANEL WITH GENOME WIDE CNV ANALYSIS INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS

PUM1	100%	100%	99.9%	97.6%	Spinocerebellar ataxia 47, 617931;Neurodevelopmental disorder with motor abnormalities, seizures, and facial dysmorphism, 620719	MOVEMENT DISORDERS PANEL EPILEPSY PANEL INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS
PURA	100%	100%	100%	98.9%	Neurodevelopmental disorder with neonatal respiratory insufficiency, hypotonia, and feeding difficulties, 616158	EPILEPSY PANEL INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS

PUS1	100%	100%	100%	99%	Myopathy, lactic acidosis, and sideroblastic anemia 1, 600462	IRON DISORDERS PANEL INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS MITOCHONDRIAL DISORDERS PANEL COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹ MUSCLE DISORDERS PANEL
PUS3	100%	100%	100%	99.5%	Neurodevelopmental disorder with microcephaly and gray sclerae, 617051	METABOLIC DISORDERS PANEL INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹

PUS7	100%	100%	100%	99.4%	Intellectual developmental disorder with abnormal behavior, microcephaly, and short stature, 618342	INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL¹
PXDN	100%	99.5%	100%	99%	Anterior segment dysgenesis 7, with sclerocornea, 269400	VISION DISORDERS PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL¹

PYCR1	100%	100%	100%	99.6%	Cutis laxa, autosomal recessive, type IIIB, 614438;Cutis laxa, autosomal recessive, type IIB, 612940	SKIN DISORDERS PANEL ¹ SHORT STATURE/SKELETAL DYSPLASIA PANEL WITH GENOME WIDE CNV ANALYSIS METABOLIC DISORDERS PANEL INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS MITOCHONDRIAL DISORDERS PANEL COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹
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PYCR2	100%	100%	100%	99%	Leukodystrophy, hypomyelinating, 10, 616420	COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹ MOVEMENT DISORDERS PANEL EPILEPSY PANEL METABOLIC DISORDERS PANEL INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS MITOCHONDRIAL DISORDERS PANEL
PYGL	100%	100%	100%	99.2%	Glycogen storage disease VI, 232700	METABOLIC DISORDERS PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹

PYGM	100%	100%	100%	98.8%	McArdle disease, 232600	METABOLIC DISORDERS PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL¹ MUSCLE DISORDERS PANEL
PYROXD1	100%	100%	100%	99.4%	Myopathy, myofibrillar, 8, 617258	MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS MITOCHONDRIAL DISORDERS PANEL COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL¹ MUSCLE DISORDERS PANEL
PYROXD2	90.9%	88.6%	100%	98.8%		MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS MITOCHONDRIAL DISORDERS PANEL

QARS1	100%	100%	100%	99.2%	Microcephaly, progressive, seizures, and cerebral and cerebellar atrophy, 615760	EPILEPSY PANEL INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹
QDPR	100%	100%	100%	99.3%	Hyperphenylalaninemia, BH4-deficient, C, 261630	MOVEMENT DISORDERS PANEL EPILEPSY PANEL METABOLIC DISORDERS PANEL INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹

QRICH1	100%	100%	100%	99.3%	Ververi-Brady syndrome, 617982	INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS
QRICH2	100%	100%	100%	99%	Spermatogenic failure 35, 618341	MALE INFERTILITY PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS
QRSL1	100%	100%	100%	99.7%	Combined oxidative phosphorylation deficiency 40, 618835	HEART DISORDERS PANEL ¹ MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS MITOCHONDRIAL DISORDERS PANEL
RAB11A	100%	100%	100%	99.6%		INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS

RAB11B	100%	100%	100%	98.8%	Neurodevelopmental disorder with ataxic gait, absent speech, and decreased cortical white matter, 617807	INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS
RAB14	100%	100%	100%	99.2%		INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS
RAB18	100%	100%	100%	99.8%	Warburg micro syndrome 3, 614222	MOVEMENT DISORDERS PANEL INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹

RAB23	100%	100%	100%	99.3%	Carpenter syndrome, 201000	CRANIOFACIAL ANOMALIES PANEL WITH GENOME WIDE CNV ANALYSIS SKIN DISORDERS PANEL ¹ SHORT STATURE/SKELETAL DYSPLASIA PANEL WITH GENOME WIDE CNV ANALYSIS INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹
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RAB27A	100%	100%	100%	99.1%	Griscelli syndrome, type 2, 607624	SKIN DISORDERS PANEL ¹ HEMOSTATIC/THROMB OTIC DISORDERS PANEL PRIMARY IMMUNODEFICIENCIES PANEL INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹
RAB28	100%	100%	100%	99.1%	Cone-rod dystrophy 18, 615374	VISION DISORDERS PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹

RAB33B	100%	100%	100%	99.2%	Smith-McCort dysplasia 2, 615222	SHORT STATURE/SKELETAL DYSPLASIA PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹
RAB34	100%	99.9%	100%	99.2%	Orofaciodigital syndrome XX, 620718	INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹
RAB39B	100%	100%	98.6%	74.7%	Intellectual developmental disorder, X-linked 72, 300271;Waisman syndrome, 311510	EPILEPSY PANEL INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS

RAB3GAP1	100%	100%	100%	99.6%	Martsolf syndrome 2, 619420;Warburg micro syndrome 1, 600118	MOVEMENT DISORDERS PANEL INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL¹
RAB3GAP2	94.4%	94.4%	100%	99.3%	Martsolf syndrome 1, 212720;Warburg micro syndrome 2, 614225	MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL¹ MOVEMENT DISORDERS PANEL VISION DISORDERS PANEL INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS

RAB5C	100%	100%	100%	98.5%		INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS
RAB5IF	100%	100%	100%	98.8%	?Craniofacial dysmorphism, skeletal anomalies, and impaired intellectual development syndrome 2, 616994	MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS
RAB7A	100%	99.5%	100%	99.2%	Charcot-Marie-Tooth disease, type 2B, 600882	POLYNEUROPATHIES PANEL ¹ NEUROLOGICAL PAIN DISORDERS PANEL ¹ MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS
RABGAP1	100%	100%	100%	99.3%		INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS

RAC1	86.4%	86.4%	100%	99%	Intellectual developmental disorder, autosomal dominant 48, 617751	LYMPHATIC ANOMALIES PANEL WITH GENOME WIDE CNV ANALYSIS INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS NOONAN SYNDROME / RASOPATHY PANEL
RAC2	100%	100%	100%	98.8%	Immunodeficiency 73A with defective neutrophil chemotaxis and leukocytosis, 608203;?Immunodeficiency 73C with defective neutrophil chemotaxis and hypogammaglobulinemia, 618987;Immunodeficiency 73B with defective neutrophil chemotaxis and lymphopenia, 618986	PRIMARY IMMUNODEFICIENCIES PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS SEVERE COMBINED IMMUNODEFICIENCY (SCID) PANEL

RAC3	100%	99.7%	100%	97.3%	Neurodevelopmental disorder with structural brain anomalies and dysmorphic facies, 618577	SHORT STATURE/SKELETAL DYSPLASIA PANEL WITH GENOME WIDE CNV ANALYSIS INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS
RACGAP1	100%	100%	100%	99.3%	Anemia, congenital dyserythropoietic, type IIIb, autosomal recessive, 619789	MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS

RAD21	100%	100%	100%	99.6%	Cornelia de Lange syndrome 4, 614701;?Mungan syndrome, 611376	CRANIOFACIAL ANOMALIES PANEL WITH GENOME WIDE CNV ANALYSIS SKIN DISORDERS PANEL ¹ SHORT STATURE/SKELETAL DYSPLASIA PANEL WITH GENOME WIDE CNV ANALYSIS LIVER DISORDERS PANEL INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS RENAL DISORDERS PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS
RAD21L1	100%	100%	100%	99.5%		MALE INFERTILITY PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS

RAD50	100%	100%	100%	99.7%	Nijmegen breakage syndrome-like disorder, 613078	SKIN DISORDERS PANEL ¹ MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹ HEREDITARY CANCER PANEL
RAD51	89.3%	89.3%	100%	99.1%	Mirror movements 2, 614508;{Breast cancer, susceptibility to}, 114480;Fanconi anemia, complementation group R, 617244	MOVEMENT DISORDERS PANEL INHERITED BONE MARROW FAILURE AND/OR PREDISPOSITION PANEL TO HEMATOLOGICAL MALIGNANCIES SHORT STATURE/SKELETAL DYSPLASIA PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS

RAD51C	90.3%	90.3%	100%	98.7%	{Breast-ovarian cancer, familial, susceptibility to, 3}, 613399;Fanconi anemia, complementation group O, 613390	INHERITED BONE MARROW FAILURE AND/OR PREDISPOSITION PANEL TO HEMATOLOGICAL MALIGNANCIES MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹ HEREDITARY CANCER PANEL
RAD51D	100%	100%	100%	99.1%	{Breast-ovarian cancer, familial, susceptibility to, 4}, 614291	MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS HEREDITARY CANCER PANEL
RAD54B	100%	100%	100%	99.4%	Colon cancer, somatic, 114500;Lymphoma, non-Hodgkin, somatic, 605027	MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS
RAD54L	100%	100%	100%	99.5%	{Breast cancer, invasive ductal}, 114480;Adenocarcinoma, colonic, somatic;Lymphoma, non-Hodgkin, somatic, 605027	MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS

RAF1	96.6%	93.7%	100%	99.3%	Cardiomyopathy, dilated, 1NN, 615916;Noonan syndrome 5, 611553;LEOPARD syndrome 2, 611554	MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS NOONAN SYNDROME / RASOPATHY PANEL HEREDITARY CANCER PANEL SHORT STATURE/SKELETAL DYSPLASIA PANEL WITH GENOME WIDE CNV ANALYSIS LYMPHATIC ANOMALIES PANEL WITH GENOME WIDE CNV ANALYSIS INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS HYPERTROPHIC CARDIOMYOPATHY PANEL ¹ HEMOSTATIC/THROMB OTIC DISORDERS PANEL CONGENITAL HEARTDISEASE PANEL ¹ SKIN DISORDERS PANEL ¹ HEART DISORDERS PANEL ¹
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RAG1	100%	100%	100%	99.4%	Omenn syndrome, 603554;Severe combined immunodeficiency, B cell-negative, 601457;Combined cellular and humoral immune defects with granulomas, 233650;Alpha/beta T-cell lymphopenia with gamma/delta T-cell expansion, severe cytomegalovirus infection, and autoimmunity, 609889	SKIN DISORDERS PANEL ¹ PRIMARY IMMUNODEFICIENCIES PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹ SEVERE COMBINED IMMUNODEFICIENCY (SCID) PANEL
RAG2	100%	100%	100%	99.6%	Severe combined immunodeficiency, B cell-negative, 601457;Combined cellular and humoral immune defects with granulomas, 233650;Omenn syndrome, 603554	SKIN DISORDERS PANEL ¹ PRIMARY IMMUNODEFICIENCIES PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹ SEVERE COMBINED IMMUNODEFICIENCY (SCID) PANEL

RAI1	100%	100%	100%	98.7%	Smith-Magenis syndrome, 182290	SKIN DISORDERS PANEL ¹ HEARING IMPAIRMENT PANEL (INCLUDING GJB2) INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS
RALA	100%	100%	100%	99.9%	Hiatt-Neu-Cooper neurodevelopmental syndrome, 619311	SHORT STATURE/SKELETAL DYSPLASIA PANEL WITH GENOME WIDE CNV ANALYSIS INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS

RALGAPA1	100%	100%	100%	99.6%	Neurodevelopmental disorder with hypotonia, neonatal respiratory insufficiency, and thermodysregulation, 618797	INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹
RANBP2	100%	100%	100%	99.7%	{Encephalopathy, acute, infection-induced, 3, susceptibility to}, 608033	PRIMARY IMMUNODEFICIENCIES PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS MITOCHONDRIAL DISORDERS PANEL
RANGRF	100%	100%	100%	98.9%		HEART DISORDERS PANEL ¹ MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS

RAP1B	100%	100%	100%	99.7%	Thrombocytopenia 11 with multiple congenital anomalies and dysmorphic facies, 620654	INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS
RAP1GDS1	100%	100%	100%	99.5%	Alfadhel syndrome, 620655	MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS
RAPGEF2	100%	100%	100%	99.3%	?Epilepsy, familial adult myoclonic, 7, 618075	MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS
RAPSN	100%	100%	100%	98.3%	Fetal akinesia deformation sequence 2, 618388;Myasthenic syndrome, congenital, 11, associated with acetylcholine receptor deficiency, 616326	FETAL AKINESIA PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹ MUSCLE DISORDERS PANEL

RARB	98%	98%	100%	99.5%	Microphthalmia, syndromic 12, 615524	VISION DISORDERS PANEL INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL¹
RARS1	94.6%	94.3%	100%	99.2%	Leukodystrophy, hypomyelinating, 9, 616140	MOVEMENT DISORDERS PANEL INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL¹

RARS2	94.1%	93.4%	100%	99.4%	Pontocerebellar hypoplasia, type 6, 611523	MOVEMENT DISORDERS PANEL EPILEPSY PANEL INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS MITOCHONDRIAL DISORDERS PANEL COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL¹
RASA1	100%	100%	100%	99.5%	Capillary malformation-arteriovenous malformation 1, 608354;Basal cell carcinoma, somatic, 605462	LYMPHATIC ANOMALIES PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS

RASGRP1	95%	95%	100%	99.3%	Immunodeficiency 64, 618534	PRIMARY IMMUNODEFICIENCIES PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL¹
RASGRP2	100%	100%	100%	98.1%	?Bleeding disorder, platelet-type, 18, 615888	HEMOSTATIC/THROMB OTIC DISORDERS PANEL PRIMARY IMMUNODEFICIENCIES PANEL SHORT STATURE/SKELETAL DYSPLASIA PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS

RAX	100%	100%	100%	98.8%	Microphthalmia, syndromic 16, 611038	VISION DISORDERS PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹
RAX2	100%	100%	100%	97.8%	Retinitis pigmentosa 95, 620102;Cone-rod dystrophy 11, 610381;?Macular degeneration, age-related, 6, 613757	MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS VISION DISORDERS PANEL
RB1	100%	100%	100%	99.4%	Small cell cancer of the lung, somatic, 182280;Bladder cancer, somatic, 109800;Retinoblastoma, trilateral, 180200;Osteosarcoma, somatic, 259500;Retinoblastoma, 180200	MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS HEREDITARY CANCER PANEL
RB1CC1	100%	100%	100%	99.4%	Breast cancer, somatic, 114480	MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS

RBBP5	100%	100%	100%	99.7%		INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS
RBBP6	100%	100%	100%	99.2%		INHERITED BONE MARROW FAILURE AND/OR PREDISPOSITION PANEL TO HEMATOLOGICAL MALIGNANCIES MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS
RBBP7	100%	99.3%	98.1%	67.4%		MALE INFERTILITY PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS

RBBP8	100%	100%	100%	99.4%	Seckel syndrome 2, 606744;Jawad syndrome, 251255;Pancreatic carcinoma, somatic	SKIN DISORDERS PANEL ¹ SHORT STATURE/SKELETAL DYSPLASIA PANEL WITH GENOME WIDE CNV ANALYSIS INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹
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RBCK1	100%	100%	99.9%	95.7%	Polyglucosan body myopathy 1 with or without immunodeficiency, 615895	HEART DISORDERS PANEL ¹ PRIMARY IMMUNODEFICIENCIES PANEL METABOLIC DISORDERS PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹ MUSCLE DISORDERS PANEL
RBFOX1	99.9%	99.4%	100%	99%		INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS
RBFOX2	92.7%	91.2%	100%	99.1%		CONGENITAL HEARTDISEASE PANEL ¹ HEART DISORDERS PANEL ¹ MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS

RBL2	100%	100%	100%	99.6%	Brunet-Wagner neurodevelopmental syndrome, 619690	INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹
RBM10	100%	99.6%	98%	66.1%	TARP syndrome, 311900	CRANIOFACIAL ANOMALIES PANEL WITH GENOME WIDE CNV ANALYSIS INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS OROFACIAL CLEFTING PANEL WITH GENOME WIDE CNV ANALYSIS

RBM20	100%	100%	100%	99.1%	Cardiomyopathy, dilated, 1DD, 613172	DILATED CARDIOMYOPATHY PANEL ¹ HEART DISORDERS PANEL ¹ MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS
RBM28	100%	100%	100%	99.2%	?Alopecia, neurologic defects, and endocrinopathy syndrome, 612079	SKIN DISORDERS PANEL ¹ INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS

RBM8A	100%	100%	100%	99%	Thrombocytopenia-absent radius syndrome, 274000	INHERITED BONE MARROW FAILURE AND/OR PREDISPOSITION PANEL TO HEMATOLOGICAL MALIGNANCIES HEMOSTATIC/THROMBOTIC DISORDERS PANEL SHORT STATURE/SKELETAL DYSPLASIA PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹
RBMX	100%	99.7%	99%	76.4%	?Intellectual developmental disorder, X-linked syndromic, Gustavson type, 309555;?Intellectual developmental disorder, X-linked syndromic, Shashi type, 300238	MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS
RBM1A1	50%	50%	50%	48.5%		MALE INFERTILITY PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS

RBM1A	50%	49.8%	49.3%	42.7%		MALE INFERTILITY PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS
RBM1B	49.5%	48.1%	47.6%	42.7%		MALE INFERTILITY PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS
RBM1C	49.9%	49.6%	49.5%	45.9%		MALE INFERTILITY PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS
RBM1D	49.3%	48.8%	48.8%	34.8%		MALE INFERTILITY PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS

RBM1J	49.9%	49.8%	49.8%	35%		MALE INFERTILITY PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS
RBP3	100%	100%	100%	99.5%	?Retinitis pigmentosa 66, 615233	VISION DISORDERS PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹
RBP4	100%	100%	100%	99.7%	Microphthalmia/coloboma 10, 616428;Retinal dystrophy, iris coloboma, and comedogenic acne syndrome, 615147	VISION DISORDERS PANEL SKIN DISORDERS PANEL ¹ MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹

RBPJ	100%	100%	100%	99.1%	Adams-Oliver syndrome 3, 614814	SKIN DISORDERS PANEL ¹ SHORT STATURE/SKELETAL DYSPLASIA PANEL WITH GENOME WIDE CNV ANALYSIS INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS
RBSN	100%	100%	100%	99.4%	Myelofibrosis, congenital, with anemia, neutropenia, developmental delay, and ocular abnormalities, 620939;Kariminejad-Reversade neurodevelopmental syndrome, 620937	INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹

RC3H1	100%	100%	100%	99.3%	?Immune dysregulation and systemic hyperinflammation syndrome, 618998	PRIMARY IMMUNODEFICIENCIES PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS
RCBTB1	100%	100%	100%	99.4%	Retinal dystrophy with or without extraocular anomalies, 617175	VISION DISORDERS PANEL INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹
RD3	100%	100%	100%	99.3%	Leber congenital amaurosis 12, 610612	VISION DISORDERS PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹

RDH11	100%	100%	100%	99.6%	?Retinal dystrophy, juvenile cataracts, and short stature syndrome, 616108	VISION DISORDERS PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹
RDH12	100%	100%	100%	99.2%	Leber congenital amaurosis 13, 612712	VISION DISORDERS PANEL METABOLIC DISORDERS PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹
RDH5	100%	100%	100%	99.1%	Fundus albipunctatus, 136880	VISION DISORDERS PANEL METABOLIC DISORDERS PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹

RDX	100%	100%	100%	99.5%	Deafness, autosomal recessive 24, 611022	HEARING IMPAIRMENT PANEL (INCLUDING GJB2) MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹
REC114	100%	100%	100%	99.1%	Oocyte/zygote/embryo maturation arrest 10, 619176	UNSUCCESSFUL IVF TREATMENTS, RECURRENT MOLAR PREGNANCIES AND/OR RECURRENT PREGNANCY LOSS PANEL ¹ MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS
RECQL	100%	100%	100%	99.8%	RECON progeroid syndrome, 620370	MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS

RECQL4	100%	100%	100%	99.1%	Baller-Gerold syndrome, 218600;Rothmund-Thomson syndrome, type 2, 268400;RAPADILINO syndrome, 266280	CRANIOFACIAL ANOMALIES PANEL WITH GENOME WIDE CNV ANALYSIS SKIN DISORDERS PANEL ¹ PRIMARY IMMUNODEFICIENCIES PANEL COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹ HEREDITARY CANCER PANEL SHORT STATURE/SKELETAL DYSPLASIA PANEL WITH GENOME WIDE CNV ANALYSIS INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS
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REEP1	85.8%	85.8%	100%	99.6%	Neuronopathy, distal hereditary motor, autosomal recessive 6, 620011;Spastic paraplegia 31, autosomal dominant, 610250;?Neuronopathy, distal hereditary motor, autosomal dominant 12, 614751	MOVEMENT DISORDERS PANEL POLYNEUROPATHIES PANEL ¹ MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS
REEP2	100%	100%	100%	98.9%	Spastic paraplegia 72A, autosomal dominant, 615625;?Spastic paraplegia 72B, autosomal recessive, 620606	MOVEMENT DISORDERS PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹
REEP6	100%	100%	100%	98.7%	Retinitis pigmentosa 77, 617304	VISION DISORDERS PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹

REL	100%	99.9%	100%	99.5%	Immunodeficiency 92, 619652	PRIMARY IMMUNODEFICIENCIES PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹
RELA	100%	100%	100%	98.6%	Autoinflammatory disease, familial, Behcet-like-3, 618287	PRIMARY IMMUNODEFICIENCIES PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS
RELB	100%	99.8%	100%	96.6%	?Immunodeficiency 53, 617585	PRIMARY IMMUNODEFICIENCIES PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹

RELN	100%	100%	100%	99.4%	{Epilepsy, familial temporal lobe, 7}, 616436;Lissencephaly 2 (Norman-Roberts type), 257320	INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹
RELT	100%	100%	100%	98.9%	Amelogenesis imperfecta, type IIIC, 618386	MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS
REN	100%	100%	100%	99.2%	Renal tubular dysgenesis, 267430;[Hyperproreninemia];Tubulointerstitial kidney disease, autosomal dominant, 4, 613092	RENAL DISORDERS PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹

REPS1	100%	100%	100%	99.5%	?Neurodegeneration with brain iron accumulation 7, 617916	MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹
RERE	99%	98.9%	100%	96.3%	Neurodevelopmental disorder with or without anomalies of the brain, eye, or heart, 616975	INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS
REST	100%	100%	100%	99.6%	Deafness, autosomal dominant 27, 612431;{Wilms tumor 6, susceptibility to}, 616806;Fibromatosis, gingival, 5, 617626	HEARING IMPAIRMENT PANEL (INCLUDING GJB2) MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS HEREDITARY CANCER PANEL
RET	100%	100%	100%	99.1%	{Hirschsprung disease, susceptibility to, 1}, 142623;Multiple endocrine neoplasia IIA, 171400;{Hirschsprung disease, protection against}, 142623;Medullary thyroid carcinoma, 155240;Pheochromocytoma, 171300;Multiple endocrine neoplasia IIB, 162300	MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS HEREDITARY CANCER PANEL

RETREG1	91.3%	91.3%	100%	99%	Neuropathy, hereditary sensory and autonomic, type IIB, 613115	POLYNEUROPATHIES PANEL ¹ NEUROLOGICAL PAIN DISORDERS PANEL ¹ MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹
REV3L	100%	100%	100%	99.5%		INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS
RFC1	100%	100%	100%	99.3%	Cerebellar ataxia, neuropathy, and vestibular areflexia syndrome, 614575	MOVEMENT DISORDERS PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹

RFC4	100%	100%	100%	99.6%	Morimoto-Ryu-Malicdan neuromuscular syndrome, 621010	INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL¹
RFT1	100%	100%	100%	99.3%	Congenital disorder of glycosylation, type In, 612015	METABOLIC DISORDERS PANEL INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL¹

RFWD3	100%	100%	100%	99.2%	?Fanconi anemia, complementation group W, 617784	INHERITED BONE MARROW FAILURE AND/OR PREDISPOSITION PANEL TO HEMATOLOGICAL MALIGNANCIES SHORT STATURE/SKELETAL DYSPLASIA PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹
RFX3	100%	100%	100%	99.5%		INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS

RFX4	100%	100%	100%	98.3%		INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS
RFX5	100%	100%	100%	99.5%	?MHC class II deficiency 5, 620818;MHC class II deficiency 3, 620816	PRIMARY IMMUNODEFICIENCIES PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹ SEVERE COMBINED IMMUNODEFICIENCY (SCID) PANEL
RFX6	100%	100%	100%	99.6%	Mitchell-Riley syndrome, 615710	LIVER DISORDERS PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹

RFX7	100%	100%	100%	99.1%	Intellectual developmental disorder, autosomal dominant 71, with behavioral abnormalities, 620330	INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS
RFXANK	100%	100%	100%	97.6%	MHC class II deficiency 2, 620815	PRIMARY IMMUNODEFICIENCIES PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL¹ SEVERE COMBINED IMMUNODEFICIENCY (SCID) PANEL

RFXAP	100%	100%	100%	96.9%	MHC class II deficiency 4, 620817	PRIMARY IMMUNODEFICIENCIES PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL¹ SEVERE COMBINED IMMUNODEFICIENCY (SCID) PANEL
RGR	100%	100%	100%	98.8%	Retinitis pigmentosa 44, 613769	MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL¹
RGS10	100%	100%	100%	99.4%		PRIMARY IMMUNODEFICIENCIES PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS

RGS9	100%	100%	100%	99.3%	Prolonged electroretinal response suppression 1, 608415	VISION DISORDERS PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹
RGS9BP	100%	100%	100%	98.4%	Prolonged electroretinal response suppression 2, 620344	VISION DISORDERS PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS
RHAG	100%	100%	100%	99.5%	Overhydrated hereditary stomatocytosis, 185000;Anemia, hemolytic, Rh-null, regulator type, 268150	MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹

RHBDF2	100%	99.9%	100%	97.3%	Tylosis with esophageal cancer, 148500	SKIN DISORDERS PANEL ¹ PRIMARY IMMUNODEFICIENCIES PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS HEREDITARY CANCER PANEL
RHCE	96.8%	96.8%	98.2%	91.3%	Rh-null disease, amorph type, 617970	MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS
RHEB	100%	100%	100%	98.9%		INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS

RHO	100%	100%	100%	99.3%	Night blindness, congenital stationary, autosomal dominant 1, 610445;Retinitis pigmentosa 4, autosomal dominant or recessive, 613731;Retinitis punctata albescens, 136880	VISION DISORDERS PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹
RHOA	80.4%	80.4%	100%	99.2%	Ectodermal dysplasia with facial dysmorphism and acral, ocular, and brain anomalies, somatic mosaic, 618727	SKIN DISORDERS PANEL ¹ MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS
RHOBTB2	98.7%	98.7%	100%	98.8%	Developmental and epileptic encephalopathy 64, 618004	MOVEMENT DISORDERS PANEL EPILEPSY PANEL INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS

RHOG	100%	100%	100%	99.5%		PRIMARY IMMUNODEFICIENCIES PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS
RHOH	100%	100%	100%	98.7%	{?Epidermodysplasia verruciformis, susceptibility to, 4}, 618307	PRIMARY IMMUNODEFICIENCIES PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS
RIC1	100%	100%	100%	99.4%	CATIFA syndrome, 618761	INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS
RICTOR	100%	100%	100%	99.5%		INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS

RIGI	98.6%	98.6%	100%	99.4%	Singleton-Merten syndrome 2, 616298	VISION DISORDERS PANEL PRIMARY IMMUNODEFICIENCIES PANEL SHORT STATURE/SKELETAL DYSPLASIA PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS
RILPL1	100%	100%	100%	98.9%	Oculopharyngodistal myopathy 4, 619790	MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS
RIMS1	100%	100%	100%	99.5%		MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS

RIMS2	99.2%	99.2%	100%	99.2%	Cone-rod synaptic disorder syndrome, congenital nonprogressive, 618970	VISION DISORDERS PANEL INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹
RIN2	100%	100%	100%	99.2%	Macrocephaly, alopecia, cutis laxa, and scoliosis, 613075	SKIN DISORDERS PANEL ¹ MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹

RINT1	100%	100%	100%	99.7%	Infantile liver failure syndrome 3, 618641	LIVER DISORDERS PANEL METABOLIC DISORDERS PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹
RIPK1	100%	100%	100%	98.6%	Immunodeficiency 57 with autoinflammation, 618108;Autoinflammation with episodic fever and lymphadenopathy, 618852	PRIMARY IMMUNODEFICIENCIES PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹

RIPK4	100%	99.9%	100%	99.3%	CHAND syndrome, 214350;Popliteal pterygium syndrome, Bartsocas-Papas type 1, 263650	FETAL AKINESIA PANEL CRANIOFACIAL ANOMALIES PANEL WITH GENOME WIDE CNV ANALYSIS SKIN DISORDERS PANEL ¹ DISORDERS/DIFFEREN CES OF SEX DEVELOPMENT (DSD) / PRIMARY ADRENAL INSUFFICIENCY PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹ OROFACIAL CLEFTING PANEL WITH GENOME WIDE CNV ANALYSIS
RIPOR2	100%	100%	100%	99.1%	Deafness, autosomal dominant 21, 607017;?Deafness, autosomal recessive 104, 616515	HEARING IMPAIRMENT PANEL (INCLUDING GJB2) MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹

RIPPLY2	100%	100%	100%	99.2%	?Spondylocostal dysostosis 6, 616566	SHORT STATURE/SKELETAL DYSPLASIA PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹
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RIT1	100%	100%	100%	99.3%	Noonan syndrome 8, 615355	CONGENITAL HEARTDISEASE PANEL¹ INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS NOONAN SYNDROME / RASOPATHY PANEL HEREDITARY CANCER PANEL HEART DISORDERS PANEL¹ HYPERTROPHIC CARDIOMYOPATHY PANEL¹ HEMOSTATIC/THROMB OTIC DISORDERS PANEL SHORT STATURE/SKELETAL DYSPLASIA PANEL WITH GENOME WIDE CNV ANALYSIS LYMPHATIC ANOMALIES PANEL WITH GENOME WIDE CNV ANALYSIS
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RLBP1	100%	100%	100%	99.4%	Bothnia retinal dystrophy, 607475;Newfoundland rod-cone dystrophy, 607476;Retinitis punctata albescens, 136880;Fundus albipunctatus, 136880	VISION DISORDERS PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹
RLF	100%	100%	100%	99.6%		INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS
RLIM	100%	100%	98.8%	67.4%	Tonne-Kalscheuer syndrome, 300978	INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS

RMND1	85.6%	85.6%	100%	98.7%	Combined oxidative phosphorylation deficiency 11, 614922	HEARING IMPAIRMENT PANEL (INCLUDING GJB2) INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS RENAL DISORDERS PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS MITOCHONDRIAL DISORDERS PANEL COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL¹
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RMRP					Anauxetic dysplasia 1, 607095;Metaphyseal dysplasia without hypotrichosis, 250460;Cartilage-hair hypoplasia, 250250	INHERITED BONE MARROW FAILURE AND/OR PREDISPOSITION PANEL TO HEMATOLOGICAL MALIGNANCIES SKIN DISORDERS PANEL ¹ PRIMARY IMMUNODEFICIENCIES PANEL SHORT STATURE/SKELETAL DYSPLASIA PANEL WITH GENOME WIDE CNV ANALYSIS INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹ SEVERE COMBINED IMMUNODEFICIENCY (SCID) PANEL
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RNASEH1	100%	99.6%	100%	98.9%	Progressive external ophthalmoplegia with mitochondrial DNA deletions, autosomal recessive 2, 616479	MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS MITOCHONDRIAL DISORDERS PANEL COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹ MUSCLE DISORDERS PANEL
RNASEH2A	100%	100%	100%	98.1%	Aicardi-Goutieres syndrome 4, 610333	MOVEMENT DISORDERS PANEL SKIN DISORDERS PANEL ¹ EPILEPSY PANEL PRIMARY IMMUNODEFICIENCIES PANEL INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹

RNASEH2B	91.4%	91.4%	100%	99.7%	Aicardi-Goutieres syndrome 2, 610181	MOVEMENT DISORDERS PANEL SKIN DISORDERS PANEL ¹ EPILEPSY PANEL PRIMARY IMMUNODEFICIENCIES PANEL INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹
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RNASEH2C	100%	100%	99.9%	95.9%	Aicardi-Goutieres syndrome 3, 610329	MOVEMENT DISORDERS PANEL SKIN DISORDERS PANEL ¹ EPILEPSY PANEL PRIMARY IMMUNODEFICIENCIES PANEL INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹
RNASEL	100%	100%	100%	99.5%	Prostate cancer 1, 601518	MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS HEREDITARY CANCER PANEL

RNASET2	100%	100%	100%	99.4%	Leukoencephalopathy, cystic, without megalencephaly, 612951	INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL¹
RNF111	100%	100%	100%	99.4%		DISORDERS/DIFFEREN CES OF SEX DEVELOPMENT (DSD) / PRIMARY ADRENAL INSUFFICIENCY PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS PREMATURE OVARIAN INSUFFICIENCY PANEL
RNF113A	100%	99.8%	99%	65.8%	Trichothiodystrophy 5, nonphotosensitive, 300953	SKIN DISORDERS PANEL¹ INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS

RNF125	100%	100%	100%	99.2%	Tenorio syndrome, 616260	TALL STATURE PANEL WITH GENOME WIDE CNV ANALYSIS INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS
RNF13	100%	100%	100%	99.5%	Developmental and epileptic encephalopathy 73, 618379	EPILEPSY PANEL INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS
RNF135	100%	100%	100%	98.9%		TALL STATURE PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS

RNF139	100%	100%	100%	99.7%	Renal cell carcinoma, 144700	MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS
RNF168	100%	100%	100%	99%	RIDDLE syndrome, 611943	PRIMARY IMMUNODEFICIENCIES PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹
RNF170	100%	100%	100%	99.2%	Ataxia, sensory, 1, autosomal dominant, 608984;Spastic paraplegia 85, autosomal recessive, 619686	MOVEMENT DISORDERS PANEL POLYNEUROPATHIES PANEL ¹ MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹

RNF2	100%	100%	100%	99.1%	Luo-Schoch-Yamamoto syndrome, 619460	INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS
RNF212	100%	100%	100%	97.6%	?Spermatogenic failure 62, 619673;Recombination rate QTL 1, 612042	MALE INFERTILITY PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS
RNF213	100%	100%	100%	99.2%	{Moyamoya disease 2, susceptibility to}, 607151	MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS MITOCHONDRIAL DISORDERS PANEL

RNF216	100%	100%	100%	99.5%	Cerebellar ataxia and hypogonadotropic hypogonadism, 212840	MOVEMENT DISORDERS PANEL HYPOGONADOTROPIC HYPOGONADISM (KALLMANN) PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹
RNF220	100%	100%	100%	99.5%	Leukodystrophy, hypomyelinating, 23, with ataxia, deafness, liver dysfunction, and dilated cardiomyopathy, 619688	INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹

RNF31	100%	100%	100%	98.7%	Immunodeficiency 115 with autoinflammation, 620632	PRIMARY IMMUNODEFICIENCIES PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹
RNF43	100%	100%	100%	99.1%	Sessile serrated polyposis cancer syndrome, 617108	PANEL HEREDITARY COLORECTAL AND POLYPOSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS HEREDITARY CANCER PANEL
RNF6	100%	100%	100%	99.5%	Esophageal carcinoma, somatic, 133239	MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS

RNPC3	100%	100%	100%	99.4%	Pituitary hormone deficiency, combined or isolated, 7, 618160	SHORT STATURE/SKELETAL DYSPLASIA PANEL WITH GENOME WIDE CNV ANALYSIS INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL¹
RNU12					CDAGS syndrome, 603116;?Spinocerebellar ataxia, autosomal recessive 33, 620208	CRANIOFACIAL ANOMALIES PANEL WITH GENOME WIDE CNV ANALYSIS INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS

RNU2-2P						INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS
RNU4-2					ReNU syndrome, 620851	VISION DISORDERS PANEL EPILEPSY PANEL INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS

RNU4ATAC					Roifman syndrome, 616651;Lowry-Wood syndrome, 226960;Microcephalic osteodysplastic primordial dwarfism, type I, 210710	VISION DISORDERS PANEL LIVER DISORDERS PANEL INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS RENAL DISORDERS PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹ SKIN DISORDERS PANEL ¹ HEMOSTATIC/THROMB OTIC DISORDERS PANEL PRIMARY IMMUNODEFICIENCIES PANEL SHORT STATURE/SKELETAL DYSPLASIA PANEL WITH GENOME WIDE CNV ANALYSIS
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RNU5B-1						INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS
RNU6-1						VISION DISORDERS PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS
RNU6-2						VISION DISORDERS PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS
RNU6-8						VISION DISORDERS PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS

RNU6-9						VISION DISORDERS PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS
RNU7-1					Aicardi-Goutieres syndrome 9, 619487	PRIMARY IMMUNODEFICIENCIES PANEL INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL¹

ROBO1	100%	100%	100%	99.3%	Pituitary hormone deficiency, combined or isolated, 8, 620303;Neurooculorenal syndrome, 620305;?Nystagmus 8, congenital, autosomal recessive, 257400	HEARING IMPAIRMENT PANEL (INCLUDING GJB2) INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹
ROBO2	100%	100%	100%	98.9%	Vesicoureteral reflux 2, 610878	RENAL DISORDERS PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS
ROBO3	100%	100%	100%	99.1%	Gaze palsy, familial horizontal, with progressive scoliosis, 1, 607313	MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹

ROBO4	100%	100%	100%	98.9%	Aortic valve disease 3, 618496	THORACIC AORTIC ANEURYSM AND/OR DISSECTION PANEL CONGENITAL HEARTDISEASE PANEL ¹ HEART DISORDERS PANEL ¹ MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS
ROGDI	100%	99.9%	100%	98.4%	Kohlschutter-Tonz syndrome, 226750	SKIN DISORDERS PANEL ¹ EPILEPSY PANEL INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹
ROM1	100%	99.9%	100%	99.1%	Retinitis pigmentosa 7, digenic form, 608133	VISION DISORDERS PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS

ROR1	100%	99.9%	100%	99.4%	?Deafness, autosomal recessive 108, 617654	HEARING IMPAIRMENT PANEL (INCLUDING GJB2) MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS
ROR2	100%	100%	100%	99.2%	Brachydactyly, type B1, 113000;Robinow syndrome, autosomal recessive, 268310	DISORDERS/DIFFEREN CES OF SEX DEVELOPMENT (DSD) / PRIMARY ADRENAL INSUFFICIENCY PANEL SHORT STATURE/SKELETAL DYSPLASIA PANEL WITH GENOME WIDE CNV ANALYSIS OROFACIAL CLEFTING PANEL WITH GENOME WIDE CNV ANALYSIS INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹

RORA	100%	100%	100%	98.6%	Intellectual developmental disorder with or without epilepsy or cerebellar ataxia, 618060	MOVEMENT DISORDERS PANEL EPILEPSY PANEL INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS
RORB	100%	100%	100%	99.8%	{Epilepsy, idiopathic generalized, susceptibility to, 15}, 618357	INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS
RORC	100%	100%	100%	98.3%	Immunodeficiency 42, 616622	PRIMARY IMMUNODEFICIENCIES PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹

RP1	100%	100%	100%	99.6%	Retinitis pigmentosa 1, 180100	VISION DISORDERS PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹
RP1L1	100%	100%	100%	98.1%	Occult macular dystrophy, 613587;Retinitis pigmentosa 88, 618826	VISION DISORDERS PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹
RP2	100%	100%	98.8%	68.8%	Retinitis pigmentosa 2, 312600	VISION DISORDERS PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS
RP9	100%	100%	100%	95.8%	?Retinitis pigmentosa 9, 180104	VISION DISORDERS PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS

RPA1	100%	100%	100%	99.3%	Pulmonary fibrosis and/or bone marrow failure syndrome, telomere-related, 6, 619767	INHERITED BONE MARROW FAILURE AND/OR PREDISPOSITION PANEL TO HEMATOLOGICAL MALIGNANCIES DYSKERATOSIS CONGENITA AND APLASTIC ANEMIA PANEL PRIMARY IMMUNODEFICIENCIES PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS HEREDITARY CANCER PANEL
RPE65	100%	100%	100%	99.5%	Retinitis pigmentosa 20, 613794;Retinitis pigmentosa 87 with choroidal involvement, 618697;Leber congenital amaurosis 2, 204100	MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹ VISION DISORDERS PANEL METABOLIC DISORDERS PANEL

RPGR	99.8%	97.1%	88.4%	53.4%	Retinitis pigmentosa, X-linked, and sinorespiratory infections, with or without deafness, 300455;Cone-rod dystrophy, X-linked, 1, 304020;Retinitis pigmentosa 3, 300029;Macular degeneration, X-linked atrophic, 300834	VISION DISORDERS PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS
RPGRIP1	100%	100%	100%	98.5%	Cone-rod dystrophy 13, 608194;Leber congenital amaurosis 6, 613826	VISION DISORDERS PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹

RPGRIP1L	100%	100%	100%	99.5%	Joubert syndrome 7, 611560;Meckel syndrome 5, 611561;?COACH syndrome 3, 619113	INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS RENAL DISORDERS PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS VISION DISORDERS PANEL CILIOPATHIES PANEL SHORT STATURE/SKELETAL DYSPLASIA PANEL WITH GENOME WIDE CNV ANALYSIS LIVER DISORDERS PANEL COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹ OROFACIAL CLEFTING PANEL WITH GENOME WIDE CNV ANALYSIS
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RPH3A	100%	100%	100%	99.1%		INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS
RPIA	100%	100%	100%	99.5%	Ribose 5-phosphate isomerase deficiency, 608611	METABOLIC DISORDERS PANEL INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹

RPL10	100%	99.6%	98.8%	69.8%	{Autism, susceptibility to, X-linked 5}, 300847;Intellectual developmental disorder, X-linked syndromic 35, 300998	DISORDERS/DIFFERENCES OF SEX DEVELOPMENT (DSD) / PRIMARY ADRENAL INSUFFICIENCY PANEL SHORT STATURE/SKELETAL DYSPLASIA PANEL WITH GENOME WIDE CNV ANALYSIS INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS
RPL10L	100%	100%	100%	99%	?Spermatogenic failure 63, 619689	MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS

RPL11	100%	100%	100%	99.1%	Diamond-Blackfan anemia 7, 612562	INHERITED BONE MARROW FAILURE AND/OR PREDISPOSITION PANEL TO HEMATOLOGICAL MALIGNANCIES MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS HEREDITARY CANCER PANEL OROFACIAL CLEFTING PANEL WITH GENOME WIDE CNV ANALYSIS
RPL13	100%	99.9%	100%	98.2%	Spondyloepimetaphyseal dysplasia, Isidor-Toutain type, 618728	SHORT STATURE/SKELETAL DYSPLASIA PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS

RPL15	99.2%	93.4%	100%	99.4%	Diamond-Blackfan anemia 12, 615550	INHERITED BONE MARROW FAILURE AND/OR PREDISPOSITION PANEL TO HEMATOLOGICAL MALIGNANCIES MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS HEREDITARY CANCER PANEL
RPL18	100%	100%	100%	99.5%	?Diamond-Blackfan anemia 18, 618310	HEREDITARY CANCER PANEL INHERITED BONE MARROW FAILURE AND/OR PREDISPOSITION PANEL TO HEMATOLOGICAL MALIGNANCIES MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS
RPL21	100%	99.9%	100%	99.4%	Hypotrichosis 12, 615885	SKIN DISORDERS PANEL ¹ MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS

RPL26	100%	100%	100%	99.6%	?Diamond-Blackfan anemia 11, 614900	INHERITED BONE MARROW FAILURE AND/OR PREDISPOSITION PANEL TO HEMATOLOGICAL MALIGNANCIES MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS OROFACIAL CLEFTING PANEL WITH GENOME WIDE CNV ANALYSIS
RPL27	100%	99.9%	100%	99.7%	?Diamond-Blackfan anemia 16, 617408	INHERITED BONE MARROW FAILURE AND/OR PREDISPOSITION PANEL TO HEMATOLOGICAL MALIGNANCIES MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS HEREDITARY CANCER PANEL

RPL31	100%	99.9%	100%	99%		INHERITED BONE MARROW FAILURE AND/OR PREDISPOSITION PANEL TO HEMATOLOGICAL MALIGNANCIES MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS
RPL35	100%	100%	100%	99.6%	?Diamond-Blackfan anemia 19, 618312	INHERITED BONE MARROW FAILURE AND/OR PREDISPOSITION PANEL TO HEMATOLOGICAL MALIGNANCIES MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS

RPL35A	100%	100%	100%	99.7%	Diamond-Blackfan anemia 5, 612528	INHERITED BONE MARROW FAILURE AND/OR PREDISPOSITION PANEL TO HEMATOLOGICAL MALIGNANCIES MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS HEREDITARY CANCER PANEL
RPL3L	100%	100%	100%	98.3%	Cardiomyopathy, dilated, 2D, 619371	HEART DISORDERS PANEL ¹ MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹

RPL4	100%	100%	100%	99.4%		INHERITED BONE MARROW FAILURE AND/OR PREDISPOSITION PANEL TO HEMATOLOGICAL MALIGNANCIES MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS
RPL5	100%	100%	100%	99.8%	Diamond-Blackfan anemia 6, 612561	INHERITED BONE MARROW FAILURE AND/OR PREDISPOSITION PANEL TO HEMATOLOGICAL MALIGNANCIES MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS OROFACIAL CLEFTING PANEL WITH GENOME WIDE CNV ANALYSIS HEREDITARY CANCER PANEL

RPL9	100%	100%	100%	99.6%		INHERITED BONE MARROW FAILURE AND/OR PREDISPOSITION PANEL TO HEMATOLOGICAL MALIGNANCIES MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS
RPN2	100%	100%	100%	99.2%		METABOLIC DISORDERS PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS
RPS10	100%	100%	100%	99.4%	Diamond-Blackfan anemia 9, 613308	INHERITED BONE MARROW FAILURE AND/OR PREDISPOSITION PANEL TO HEMATOLOGICAL MALIGNANCIES MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS HEREDITARY CANCER PANEL

RPS14	100%	100%	100%	99.5%	Macrocytic anemia, refractory, due to 5q deletion, somatic, 153550	MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS
RPS15A	79.7%	79.7%	100%	99.5%	?Diamond-Blackfan anemia 20, 618313	INHERITED BONE MARROW FAILURE AND/OR PREDISPOSITION PANEL TO HEMATOLOGICAL MALIGNANCIES MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS HEREDITARY CANCER PANEL
RPS17	100%	100%	100%	99.4%	Diamond-Blackfan anemia 4, 612527	INHERITED BONE MARROW FAILURE AND/OR PREDISPOSITION PANEL TO HEMATOLOGICAL MALIGNANCIES MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS HEREDITARY CANCER PANEL

RPS19	100%	99.8%	100%	99.1%	Diamond-Blackfan anemia 1, 105650	INHERITED BONE MARROW FAILURE AND/OR PREDISPOSITION PANEL TO HEMATOLOGICAL MALIGNANCIES INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS OROFACIAL CLEFTING PANEL WITH GENOME WIDE CNV ANALYSIS HEREDITARY CANCER PANEL
RPS20	100%	100%	100%	99%		PANEL HEREDITARY COLORECTAL AND POLYPOSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS HEREDITARY CANCER PANEL

RPS23	100%	100%	100%	99.6%	Brachycephaly, trichomegaly, and developmental delay, 617412	MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS
RPS24	100%	100%	100%	99.6%	Diamond-blackfan anemia 3, 610629	INHERITED BONE MARROW FAILURE AND/OR PREDISPOSITION PANEL TO HEMATOLOGICAL MALIGNANCIES MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS HEREDITARY CANCER PANEL

RPS26	100%	97.7%	100%	99.2%	Diamond-Blackfan anemia 10, 613309	INHERITED BONE MARROW FAILURE AND/OR PREDISPOSITION PANEL TO HEMATOLOGICAL MALIGNANCIES MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS OROFACIAL CLEFTING PANEL WITH GENOME WIDE CNV ANALYSIS HEREDITARY CANCER PANEL
RPS27	100%	100%	100%	99.5%	?Diamond-Blackfan anemia 17, 617409	INHERITED BONE MARROW FAILURE AND/OR PREDISPOSITION PANEL TO HEMATOLOGICAL MALIGNANCIES MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS HEREDITARY CANCER PANEL

RPS28	100%	100%	100%	97.9%	Diamond Blackfan anemia 15 with mandibulofacial dysostosis, 606164	INHERITED BONE MARROW FAILURE AND/OR PREDISPOSITION PANEL TO HEMATOLOGICAL MALIGNANCIES MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS OROFACIAL CLEFTING PANEL WITH GENOME WIDE CNV ANALYSIS HEREDITARY CANCER PANEL
RPS29	100%	100%	100%	99.8%	Diamond-Blackfan anemia 13, 615909	INHERITED BONE MARROW FAILURE AND/OR PREDISPOSITION PANEL TO HEMATOLOGICAL MALIGNANCIES MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS HEREDITARY CANCER PANEL

RPS4Y2	50%	49.9%	49.3%	19.8%		MALE INFERTILITY PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS
RPS6KA3	100%	99.9%	99.2%	71.2%	Intellectual developmental disorder, X-linked 19, 300844;Coffin-Lowry syndrome, 303600	EPILEPSY PANEL INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS
RPS6KB1	100%	100%	100%	99.3%		HEART DISORDERS PANEL ¹ MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS

RPS7	100%	100%	100%	99.5%	Diamond-Blackfan anemia 8, 612563	INHERITED BONE MARROW FAILURE AND/OR PREDISPOSITION PANEL TO HEMATOLOGICAL MALIGNANCIES MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS HEREDITARY CANCER PANEL
RPSA	100%	99.8%	100%	99.7%	Asplenia, isolated congenital, 271400	PRIMARY IMMUNODEFICIENCIES PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS
RRAD	100%	100%	100%	98.1%		HEART DISORDERS PANEL ¹ MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS

RRAGC	100%	100%	100%	99.2%	Long-Olsen-Distelmaier syndrome, 620609	HEART DISORDERS PANEL ¹ MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS
RRAGD	100%	100%	100%	99.2%	Hypomagnesemia 7, renal, with or without dilated cardiomyopathy, 620152	RENAL DISORDERS PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS
RRAS	100%	99.7%	100%	96.9%		SHORT STATURE/SKELETAL DYSPLASIA PANEL WITH GENOME WIDE CNV ANALYSIS LYMPHATIC ANOMALIES PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS NOONAN SYNDROME / RASOPATHY PANEL

RRAS2	100%	100%	100%	99.5%	Ovarian carcinoma;Noonan syndrome 12, 618624	SHORT STATURE/SKELETAL DYSPLASIA PANEL WITH GENOME WIDE CNV ANALYSIS LYMPHATIC ANOMALIES PANEL WITH GENOME WIDE CNV ANALYSIS INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS NOONAN SYNDROME / RASOPATHY PANEL
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RREB1	100%	100%	100%	98.4%		SHORT STATURE/SKELETAL DYSPLASIA PANEL WITH GENOME WIDE CNV ANALYSIS LYMPHATIC ANOMALIES PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS NOONAN SYNDROME / RASOPATHY PANEL
RRM1	100%	100%	100%	99.1%	Progressive external ophthalmoplegia with mitochondrial DNA deletions, autosomal recessive 6, 620647	MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS MITOCHONDRIAL DISORDERS PANEL COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL¹ MUSCLE DISORDERS PANEL

RRM2B	100%	100%	100%	99.2%	Mitochondrial DNA depletion syndrome 8B (MNGIE type), 612075;Mitochondrial DNA depletion syndrome 8A (encephalomyopathic type with renal tubulopathy), 612075;Rod-cone dystrophy, sensorineural deafness, and Fanconi-type renal dysfunction, 268315;Progressive external ophthalmoplegia with mitochondrial DNA deletions, autosomal dominant 5, 613077	HEARING IMPAIRMENT PANEL (INCLUDING GJB2) COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹ MUSCLE DISORDERS PANEL RENAL DISORDERS PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS MITOCHONDRIAL DISORDERS PANEL EPILEPSY PANEL INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS
RRP7A	100%	99.9%	100%	97.4%	?Microcephaly 28, primary, autosomal recessive, 619453	INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS

RS1	100%	99.8%	98.5%	65.7%	Retinoschisis, 312700	VISION DISORDERS PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS
RSPH1	100%	100%	100%	99.2%	Ciliary dyskinesia, primary, 24, 615481	CILIOPATHIES PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹
RSPH3	100%	100%	100%	99%	Ciliary dyskinesia, primary, 32, 616481	CILIOPATHIES PANEL MALE INFERTILITY PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹

RSPH4A	100%	100%	100%	99.1%	Ciliary dyskinesia, primary, 11, 612649	CILIOPATHIES PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹
RSPH9	100%	100%	100%	99.3%	Ciliary dyskinesia, primary, 12, 612650	CILIOPATHIES PANEL PRIMARY IMMUNODEFICIENCIES PANEL MALE INFERTILITY PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹

RSPO1	100%	100%	100%	99.4%	Palmoplantar hyperkeratosis and true hermaphroditism, 610644;Palmoplantar hyperkeratosis with squamous cell carcinoma of skin and sex reversal, 610644	SKIN DISORDERS PANEL ¹ DISORDERS/DIFFERENCES OF SEX DEVELOPMENT (DSD) / PRIMARY ADRENAL INSUFFICIENCY PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹
RSPO2	100%	100%	100%	99.3%	?Humero femoral hypoplasia with radiotibial ray deficiency, 618022;Tetraamelia syndrome 2, 618021	SHORT STATURE/SKELETAL DYSPLASIA PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹

RSPO4	100%	100%	100%	98.5%	Anonychia congenita, 206800	SKIN DISORDERS PANEL ¹ MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹
RSPRY1	100%	100%	100%	99.7%	Spondyloepimetaphyseal dysplasia, Faden-Alkuraya type, 616723	SHORT STATURE/SKELETAL DYSPLASIA PANEL WITH GENOME WIDE CNV ANALYSIS INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹

RSRC1	100%	100%	100%	99.4%	Intellectual developmental disorder, autosomal recessive 70, 618402	INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹
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RTEL1	100%	100%	100%	99%	Dyskeratosis congenita, autosomal dominant 4, 615190;Dyskeratosis congenita, autosomal recessive 5, 615190;Pulmonary fibrosis and/or bone marrow failure syndrome, telomere-related, 3, 616373	INHERITED BONE MARROW FAILURE AND/OR PREDISPOSITION PANEL TO HEMATOLOGICAL MALIGNANCIES HEREDITARY CANCER PANEL PRIMARY IMMUNODEFICIENCIES PANEL INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹ SKIN DISORDERS PANEL ¹ DYSKERATOSIS CONGENITA AND APLASTIC ANEMIA PANEL
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RTN2	100%	100%	100%	98.9%	Neuronopathy, distal hereditary motor, autosomal recessive 11, with spasticity, 620854;Spastic paraplegia 12, autosomal dominant, 604805	MOVEMENT DISORDERS PANEL POLYNEUROPATHIES PANEL ¹ MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹
RTN4IP1	100%	100%	100%	99.3%	Optic atrophy 10 with or without ataxia, impaired intellectual development and seizures, 616732	VISION DISORDERS PANEL INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS MITOCHONDRIAL DISORDERS PANEL COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹

RTTN	100%	100%	100%	99.7%	Microcephaly, short stature, and polymicrogyria with seizures, 614833	INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹
RUBCN	100%	100%	100%	98.9%	Spinocerebellar ataxia, autosomal recessive 15, 615705	MOVEMENT DISORDERS PANEL INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹

RUNX1	100%	100%	100%	95.9%	Platelet disorder, familial, with associated myeloid malignancy, 601399;Leukemia, acute myeloid, 601626	INHERITED BONE MARROW FAILURE AND/OR PREDISPOSITION PANEL TO HEMATOLOGICAL MALIGNANCIES HEMOSTATIC/THROMBOTIC DISORDERS PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS HEREDITARY CANCER PANEL
RUNX1T1	100%	100%	100%	99.1%		INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS

RUNX2	100%	100%	100%	98.8%	Metaphyseal dysplasia with maxillary hypoplasia with or without brachydactyly, 156510;Cleidocranial dysplasia, forme fruste, with brachydactyly, 119600;Cleidocranial dysplasia, forme fruste, dental anomalies only, 119600;Cleidocranial dysplasia, 119600	CRANIOFACIAL ANOMALIES PANEL WITH GENOME WIDE CNV ANALYSIS SKIN DISORDERS PANEL ¹ SHORT STATURE/SKELETAL DYSPLASIA PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS OROFACIAL CLEFTING PANEL WITH GENOME WIDE CNV ANALYSIS
RUSC2	100%	100%	100%	99.1%	Intellectual developmental disorder, autosomal recessive 61, 617773	INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹

RXFP2	100%	100%	100%	99.9%		DISORDERS/DIFFERENCES OF SEX DEVELOPMENT (DSD) / PRIMARY ADRENAL INSUFFICIENCY PANEL MALE INFERTILITY PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS
RXYLT1	100%	100%	100%	99.2%	Muscular dystrophy-dystroglycanopathy (congenital with brain and eye anomalies), type A, 10, 615041	METABOLIC DISORDERS PANEL INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹ MUSCLE DISORDERS PANEL

RYBP	100%	100%	100%	98.8%		INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS
RYR1	99.9%	99.6%	100%	98%	Congenital myopathy 1B, autosomal recessive, 255320;Congenital myopathy 1A, autosomal dominant, with susceptibility to malignant hyperthermia, 117000;King-Denborough syndrome, 619542;{Malignant hyperthermia susceptibility 1}, 145600	FETAL AKINESIA PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS MITOCHONDRIAL DISORDERS PANEL COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹ MUSCLE DISORDERS PANEL
RYR2	100%	100%	100%	99.4%	Ventricular tachycardia, catecholaminergic polymorphic, 1, 604772;Ventricular arrhythmias due to cardiac ryanodine receptor calcium release deficiency syndrome, 115000	HEART DISORDERS PANEL ¹ MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS ARRHYTHMIA AND CARDIAC CONDUCTION DISORDERS PANEL ¹

RYR3	100%	100%	100%	99.5%	Congenital myopathy 20, 620310	MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS
S100A3	100%	100%	100%	98%		SKIN DISORDERS PANEL ¹ MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS
S1PR2	100%	100%	100%	98.9%	Deafness, autosomal recessive 68, 610419	HEARING IMPAIRMENT PANEL (INCLUDING GJB2) MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹

SACS	99%	99%	100%	99.6%	Spastic ataxia, Charlevoix-Saguenay type, 270550	MOVEMENT DISORDERS PANEL POLYNEUROPATHIES PANEL ¹ MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS MITOCHONDRIAL DISORDERS PANEL COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹
SAG	100%	100%	100%	99.6%	Retinitis pigmentosa 47, autosomal recessive, 613758;Retinitis pigmentosa 96, autosomal dominant, 620228;Oguchi disease-1, 258100	VISION DISORDERS PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹

SALL1	100%	100%	100%	99.3%	Townes-Brocks syndrome 1, 107480;Townes-Brocks branchiootorenal-like syndrome, 107480	CRANIOFACIAL ANOMALIES PANEL WITH GENOME WIDE CNV ANALYSIS SHORT STATURE/SKELETAL DYSPLASIA PANEL WITH GENOME WIDE CNV ANALYSIS INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS RENAL DISORDERS PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS
SALL2	100%	100%	100%	99.4%	?Coloboma, ocular, autosomal recessive, 216820	VISION DISORDERS PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS

SALL4	100%	100%	100%	99.3%	?IVIC syndrome, 147750;Duane-radial ray syndrome, 607323	CRANIOFACIAL ANOMALIES PANEL WITH GENOME WIDE CNV ANALYSIS SHORT STATURE/SKELETAL DYSPLASIA PANEL WITH GENOME WIDE CNV ANALYSIS RENAL DISORDERS PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS OROFACIAL CLEFTING PANEL WITH GENOME WIDE CNV ANALYSIS
SAMD11	100%	100%	100%	98.2%		VISION DISORDERS PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS
SAMD12	100%	100%	100%	98.7%	Epilepsy, familial adult myoclonic, 1, 601068	MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS

SAMD7	100%	100%	100%	98.2%	Macular dystrophy with or without cone dysfunction, 620762	VISION DISORDERS PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹
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SAMD9	100%	100%	100%	99.5%	Tumoral calcinosis, familial, normophosphatemic, 610455;Monosomy 7 myelodysplasia and leukemia syndrome 2, 619041;MIRAGE syndrome, 617053	INHERITED BONE MARROW FAILURE AND/OR PREDISPOSITION PANEL TO HEMATOLOGICAL MALIGNANCIES SKIN DISORDERS PANEL ¹ DISORDERS/DIFFERENCES OF SEX DEVELOPMENT (DSD) / PRIMARY ADRENAL INSUFFICIENCY PANEL PRIMARY IMMUNODEFICIENCIES PANEL HEREDITARY CANCER PANEL METABOLIC DISORDERS PANEL INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS MITOCHONDRIAL DISORDERS PANEL COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹
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SAMD9L	100%	100%	100%	99.8%	Ataxia-pancytopenia syndrome, 159550;?Spinocerebellar ataxia 49, 619806;Monosomy 7 myelodysplasia and leukemia syndrome 1, 252270	MOVEMENT DISORDERS PANEL INHERITED BONE MARROW FAILURE AND/OR PREDISPOSITION PANEL TO HEMATOLOGICAL MALIGNANCIES POLYNEUROPATHIES PANEL ¹ PRIMARY IMMUNODEFICIENCIES PANEL METABOLIC DISORDERS PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS MITOCHONDRIAL DISORDERS PANEL HEREDITARY CANCER PANEL
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SAMHD1	100%	100%	100%	99.2%	?Chilblain lupus 2, 614415;Aicardi-Goutieres syndrome 5, 612952	MOVEMENT DISORDERS PANEL SKIN DISORDERS PANEL ¹ EPILEPSY PANEL PRIMARY IMMUNODEFICIENCIES PANEL INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS MITOCHONDRIAL DISORDERS PANEL COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹
SAR1B	100%	100%	100%	98.9%	Chylomicron retention disease, 246700	MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹

SARDH	91.7%	91.7%	100%	98.8%	[Sarcosinemia], 268900	METABOLIC DISORDERS PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS
SARS1	100%	100%	100%	99.1%	Neurodevelopmental disorder with microcephaly, ataxia, and seizures, 617709	INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹
SARS2	100%	100%	100%	99.1%	Hyperuricemia, pulmonary hypertension, renal failure, and alkalosis, 613845	RENAL DISORDERS PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS MITOCHONDRIAL DISORDERS PANEL COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹

SART3	100%	100%	100%	99.7%		SKIN DISORDERS PANEL ¹ INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS
SASH1	100%	100%	100%	98.7%	Dyschromatosis universalis hereditaria 1, 127500;?Cancer, alopecia, pigment dyscrasia, onychodystrophy, and keratoderma, 618373	SKIN DISORDERS PANEL ¹ MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹
SASH3	100%	99.5%	97.6%	65.1%	Immunodeficiency 102, 301082	PRIMARY IMMUNODEFICIENCIES PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS

SASS6	100%	100%	100%	99.5%	Microcephaly 14, primary, autosomal recessive, 616402	MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹
SAT1	100%	100%	99.2%	69.6%		SKIN DISORDERS PANEL ¹ PRIMARY IMMUNODEFICIENCIES PANEL METABOLIC DISORDERS PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS
SATB1	100%	100%	100%	99.6%	den Hoed-de Boer-Voisin syndrome, 619229;Developmental delay with dysmorphic facies and dental anomalies, 619228	INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS

SATB2	100%	100%	100%	99%	Glass syndrome, 612313	CRANIOFACIAL ANOMALIES PANEL WITH GENOME WIDE CNV ANALYSIS SKIN DISORDERS PANEL ¹ SHORT STATURE/SKELETAL DYSPLASIA PANEL WITH GENOME WIDE CNV ANALYSIS INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS MITOCHONDRIAL DISORDERS PANEL OROFACIAL CLEFTING PANEL WITH GENOME WIDE CNV ANALYSIS
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SBDS	100%	100%	100%	99.7%	{Aplastic anemia, susceptibility to}, 609135;Shwachman-Diamond syndrome 1, 260400	INHERITED BONE MARROW FAILURE AND/OR PREDISPOSITION PANEL TO HEMATOLOGICAL MALIGNANCIES PRIMARY IMMUNODEFICIENCIES PANEL SHORT STATURE/SKELETAL DYSPLASIA PANEL WITH GENOME WIDE CNV ANALYSIS INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹ HEREDITARY CANCER PANEL
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SBF1	100%	100%	100%	99%	Charcot-Marie-Tooth disease, type 4B3, 615284	POLYNEUROPATHIES PANEL ¹ MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹
SBF2	93.7%	93.7%	100%	99.5%	Charcot-Marie-Tooth disease, type 4B2, 604563	VISION DISORDERS PANEL POLYNEUROPATHIES PANEL ¹ MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹

SC5D	100%	100%	100%	99.4%	Lathosterolosis, 607330	VISION DISORDERS PANEL LIVER DISORDERS PANEL METABOLIC DISORDERS PANEL INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL¹
SCAF4	100%	100%	100%	99.2%	Fliedner-Zweier syndrome, 620511	EPILEPSY PANEL INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS

SCAMP5	100%	100%	100%	98.3%		INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS
SCAPER	100%	100%	100%	99.5%	Intellectual developmental disorder and retinitis pigmentosa, 618195	VISION DISORDERS PANEL INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹

SCARB2	100%	100%	100%	99.3%	Epilepsy, progressive myoclonic 4, with or without renal failure, 254900	EPILEPSY PANEL POLYNEUROPATHIES PANEL ¹ METABOLIC DISORDERS PANEL RENAL DISORDERS PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹
SCARF2	100%	99.8%	100%	95.9%	Van den Ende-Gupta syndrome, 600920	CRANIOFACIAL ANOMALIES PANEL WITH GENOME WIDE CNV ANALYSIS SHORT STATURE/SKELETAL DYSPLASIA PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹ OROFACIAL CLEFTING PANEL WITH GENOME WIDE CNV ANALYSIS

SCD5	100%	100%	100%	98.8%	?Deafness, autosomal dominant 79, 619086	HEARING IMPAIRMENT PANEL (INCLUDING GJB2) MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS
SCLT1	95.2%	95.2%	100%	99.4%		VISION DISORDERS PANEL CILIOPATHIES PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS
SCN10A	100%	100%	100%	99.5%	Episodic pain syndrome, familial, 2, 615551	SKIN DISORDERS PANEL ¹ HEART DISORDERS PANEL ¹ POLYNEUROPATHIES PANEL ¹ NEUROLOGICAL PAIN DISORDERS PANEL ¹ MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS SMALL FIBRE NEUROPATHY PANEL ¹

SCN11A	100%	99.9%	100%	98.7%	Episodic pain syndrome, familial, 3, 615552;Neuropathy, hereditary sensory and autonomic, type VII, 615548	MOVEMENT DISORDERS PANEL SKIN DISORDERS PANEL ¹ POLYNEUROPATHIES PANEL ¹ NEUROLOGICAL PAIN DISORDERS PANEL ¹ MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS SMALL FIBRE NEUROPATHY PANEL ¹
SCN1A	100%	100%	100%	99.5%	Developmental and epileptic encephalopathy 6B, non-Dravet, 619317;Migraine, familial hemiplegic, 3, 609634;Dravet syndrome, 607208;Febrile seizures, familial, 3A, 604403;Generalized epilepsy with febrile seizures plus, type 2, 604403	MOVEMENT DISORDERS PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS EPILEPSY PANEL

SCN1B	99.9%	99%	100%	98.7%	Generalized epilepsy with febrile seizures plus, type 1, 604233;Developmental and epileptic encephalopathy 52, 617350;Cardiac conduction defect, nonspecific, 612838;Atrial fibrillation, familial, 13, 615377;Brugada syndrome 5, 612838	NEUROLOGICAL PAIN DISORDERS PANEL ¹ INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS HEART DISORDERS PANEL ¹ COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹ SMALL FIBRE NEUROPATHY PANEL ¹ EPILEPSY PANEL
SCN2A	100%	100%	100%	99.6%	Seizures, benign familial infantile, 3, 607745;Developmental and epileptic encephalopathy 11, 613721;Episodic ataxia, type 9, 618924	MOVEMENT DISORDERS PANEL INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS EPILEPSY PANEL

SCN2B	100%	100%	100%	99.3%	Atrial fibrillation, familial, 14, 615378	HEART DISORDERS PANEL ¹ NEUROLOGICAL PAIN DISORDERS PANEL ¹ MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS SMALL FIBRE NEUROPATHY PANEL ¹
SCN3A	100%	100%	100%	99.5%	Epilepsy, familial focal, with variable foci 4, 617935;Developmental and epileptic encephalopathy 62, 617938	EPILEPSY PANEL NEUROLOGICAL PAIN DISORDERS PANEL ¹ INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS SMALL FIBRE NEUROPATHY PANEL ¹

SCN3B	100%	100%	100%	98.8%	Atrial fibrillation, familial, 16, 613120;Brugada syndrome 7, 613120	HEART DISORDERS PANEL ¹ NEUROLOGICAL PAIN DISORDERS PANEL ¹ MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS SMALL FIBRE NEUROPATHY PANEL ¹
SCN4A	100%	100%	100%	98.9%	Paramyotonia congenita, 168300;Hyperkalemic periodic paralysis, 170500;Congenital myopathy 22B, severe fetal, 620369;Hypokalemic periodic paralysis, type 2, 613345;Myotonia congenita, atypical, acetazolamide-responsive, 608390;Myasthenic syndrome, congenital, 16, 614198;Congenital myopathy 22A, classic, 620351	FETAL AKINESIA PANEL MUSCLE DISORDERS PANEL COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹ MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS
SCN4B	100%	100%	100%	98.2%	Atrial fibrillation, familial, 17, 611819;Long QT syndrome 10, 611819	HEART DISORDERS PANEL ¹ NEUROLOGICAL PAIN DISORDERS PANEL ¹ MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS SMALL FIBRE NEUROPATHY PANEL ¹

SCN5A	100%	100%	100%	99.3%	Ventricular fibrillation, familial, 1, 603829;Heart block, progressive, type IA, 113900;Cardiomyopathy, dilated, 1E, 601154;Heart block, nonprogressive, 113900;Long QT syndrome 3, 603830;Sick sinus syndrome 1, 608567;Brugada syndrome 1, 601144;Atrial fibrillation, familial, 10, 614022;{Sudden infant death syndrome, susceptibility to}, 272120	DILATED CARDIOMYOPATHY PANEL ¹ MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS ARRHYTHMIA AND CARDIAC CONDUCTION DISORDERS PANEL ¹ HEART DISORDERS PANEL ¹ LONG QT SYNDROME PANEL ¹
SCN7A	100%	100%	100%	99.5%		NEUROLOGICAL PAIN DISORDERS PANEL ¹ MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS SMALL FIBRE NEUROPATHY PANEL ¹

SCN8A	100%	100%	100%	98.7%	?Myoclonus, familial, 2, 618364;Seizures, benign familial infantile, 5, 617080;Cognitive impairment with or without cerebellar ataxia, 614306;Developmental and epileptic encephalopathy 13, 614558	MOVEMENT DISORDERS PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS SMALL FIBRE NEUROPATHY PANEL ¹ NEUROLOGICAL PAIN DISORDERS PANEL ¹ INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS EPILEPSY PANEL
SCN9A	100%	100%	100%	98.1%	Erythralgia, primary, 133020;Insensitivity to pain, congenital, 243000;Small fiber neuropathy, 133020;Paroxysmal extreme pain disorder, 167400;Neuropathy, hereditary sensory and autonomic, type IID, 243000	SMALL FIBRE NEUROPATHY PANEL ¹ MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹ POLYNEUROPATHIES PANEL ¹ NEUROLOGICAL PAIN DISORDERS PANEL ¹ SKIN DISORDERS PANEL ¹

SCNM1	100%	100%	100%	98.8%	Orofaciodigital syndrome XIX, 620107	MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹
SCNN1A	100%	100%	100%	98.2%	Pseudohypoaldosteronism, type IB1, autosomal recessive, 264350;?Liddle syndrome 3, 618126;Bronchiectasis with or without elevated sweat chloride 2, 613021	RENAL DISORDERS PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹
SCNN1B	100%	100%	100%	99.2%	Bronchiectasis with or without elevated sweat chloride 1, 211400;Pseudohypoaldosteronism, type IB2, autosomal recessive, 620125;Liddle syndrome 1, 177200	RENAL DISORDERS PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹

SCNN1G	100%	100%	100%	99.1%	Bronchiectasis with or without elevated sweat chloride 3, 613071;Pseudohypoaldosteronism, type IB3, autosomal recessive, 620126;Liddle syndrome 2, 618114	RENAL DISORDERS PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL¹
SCO1	100%	100%	100%	99.7%	Mitochondrial complex IV deficiency, nuclear type 4, 619048	LIVER DISORDERS PANEL INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS MITOCHONDRIAL DISORDERS PANEL COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL¹

SCO2	100%	100%	100%	98.8%	Myopia 6, 608908;Mitochondrial complex IV deficiency, nuclear type 2, 604377	VISION DISORDERS PANEL HEART DISORDERS PANEL ¹ POLYNEUROPATHIES PANEL ¹ INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS MITOCHONDRIAL DISORDERS PANEL COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹
SCP2	100%	100%	100%	99.4%	?Leukoencephalopathy with dystonia and motor neuropathy, 613724	POLYNEUROPATHIES PANEL ¹ METABOLIC DISORDERS PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS MITOCHONDRIAL DISORDERS PANEL COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹

SCUBE3	100%	100%	100%	98.9%	Short stature, facial dysmorphism, and skeletal anomalies with or without cardiac anomalies, 619184	SHORT STATURE/SKELETAL DYSPLASIA PANEL WITH GENOME WIDE CNV ANALYSIS INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹
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SCYL1	100%	100%	100%	98.2%	Spinocerebellar ataxia, autosomal recessive 21, 616719	MOVEMENT DISORDERS PANEL POLYNEUROPATHIES PANEL ¹ LIVER DISORDERS PANEL METABOLIC DISORDERS PANEL INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹
SCYL2	100%	100%	100%	99.5%	Arthrogryposis multiplex congenita 4, neurogenic, with agenesis of the corpus callosum, 618766	FETAL AKINESIA PANEL INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹

SDCCAG8	100%	100%	100%	99.3%	Senior-Loken syndrome 7, 613615;Bardet-Biedl syndrome 16, 615993	INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS RENAL DISORDERS PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL¹ VISION DISORDERS PANEL CILIOPATHIES PANEL HYPOGONADOTROPIC HYPOGONADISM (KALLMANN) PANEL
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SDHA	100%	100%	100%	99%	Cardiomyopathy, dilated, 1GG, 613642;Mitochondrial complex II deficiency, nuclear type 1, 252011;Neurodegeneration with ataxia and late-onset optic atrophy, 619259;Pheochromocytoma/paraganglioma syndrome 5, 614165	HEREDITARY CANCER PANEL COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹ PANEL HEREDITARY RENALCANCER MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS MITOCHONDRIAL DISORDERS PANEL HEART DISORDERS PANEL ¹ INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS
SDHAF1	100%	100%	100%	98.7%	Mitochondrial complex II deficiency, nuclear type 2, 619166	INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS MITOCHONDRIAL DISORDERS PANEL COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹

SDHAF2	100%	99.2%	100%	99.5%	Pheochromocytoma/paraganglioma syndrome 2, 601650	MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS PANEL HEREDITARY RENALCANCER HEREDITARY CANCER PANEL
SDHB	100%	100%	100%	98.9%	Pheochromocytoma/paraganglioma syndrome 4, 115310;Mitochondrial complex II deficiency, nuclear type 4, 619224;Gastrointestinal stromal tumor, 606764;Paraganglioma and gastric stromal sarcoma, 606864	MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹ PANEL HEREDITARY RENALCANCER MITOCHONDRIAL DISORDERS PANEL HEREDITARY CANCER PANEL
SDHC	100%	100%	100%	99.2%	Pheochromocytoma/paraganglioma syndrome 3, 605373;Paraganglioma and gastric stromal sarcoma, 606864;Gastrointestinal stromal tumor, 606764	MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS PANEL HEREDITARY RENALCANCER HEREDITARY CANCER PANEL

SDHD	78.9%	78.9%	100%	98.1%	Pheochromocytoma/paraganglioma syndrome 1, 168000;Paraganglioma and gastric stromal sarcoma, 606864;Mitochondrial complex II deficiency, nuclear type 3, 619167	PANEL HEREDITARY RENALCANCER HEREDITARY CANCER PANEL MITOCHONDRIAL DISORDERS PANEL COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹ MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS
SDR9C7	100%	100%	100%	99.8%	Ichthyosis, congenital, autosomal recessive 13, 617574	SKIN DISORDERS PANEL ¹ MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹
SEC23A	100%	100%	100%	99.5%	Craniolenticulosutural dysplasia, 607812	MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹ OROFACIAL CLEFTING PANEL WITH GENOME WIDE CNV ANALYSIS

SEC23B	100%	100%	100%	99.6%	?Cowden syndrome 7, 616858;Dyserythropoietic anemia, congenital, type II, 224100	METABOLIC DISORDERS PANEL INHERITED BONE MARROW FAILURE AND/OR PREDISPOSITION PANEL TO HEMATOLOGICAL MALIGNANCIES SKIN DISORDERS PANEL ¹ IRON DISORDERS PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹
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SEC24D	100%	100%	100%	99.4%	Cole-Carpenter syndrome 2, 616294	COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹ CRANIOFACIAL ANOMALIES PANEL WITH GENOME WIDE CNV ANALYSIS SHORT STATURE/SKELETAL DYSPLASIA PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS
SEC31A	100%	100%	100%	99.4%	?Halperin-Birk syndrome, 618651	INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹

SEC61A1	100%	100%	100%	99.2%	Immunodeficiency, common variable, 15, 620670;?Neutropenia, severe congenital, 11, autosomal dominant, 620674;Tubulointerstitial kidney disease, autosomal dominant, 5, 617056	PRIMARY IMMUNODEFICIENCIES PANEL RENAL DISORDERS PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS
SEC61B	100%	100%	100%	98.9%		LIVER DISORDERS PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS
SEC63	100%	100%	100%	99.4%	Polycystic liver disease 2, 617004	LIVER DISORDERS PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS

SECISBP2	100%	100%	100%	99.3%	Thyroid hormone metabolism, abnormal, 1, 609698	INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹ MUSCLE DISORDERS PANEL
SELENBP1	100%	99.9%	100%	99.1%	Extraoral halitosis due to MTO deficiency, 618148	METABOLIC DISORDERS PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS
SELENOI	100%	100%	100%	99.8%	Spastic paraplegia 81, autosomal recessive, 618768	MOVEMENT DISORDERS PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹

SELENON	90.4%	86.7%	100%	98.3%	Congenital myopathy 3 with rigid spine, 602771	FETAL AKINESIA PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹ MUSCLE DISORDERS PANEL
SEMA3A	100%	100%	100%	99.3%	{Hypogonadotropic hypogonadism 16 with or without anosmia}, 614897	DISORDERS/DIFFEREN CES OF SEX DEVELOPMENT (DSD) / PRIMARY ADRENAL INSUFFICIENCY PANEL HYPOGONADOTROPIC HYPOGONADISM (KALLMANN) PANEL SHORT STATURE/SKELETAL DYSPLASIA PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS

SEMA3E	100%	100%	100%	99.3%		CRANIOFACIAL ANOMALIES PANEL WITH GENOME WIDE CNV ANALYSIS DISORDERS/DIFFEREN CES OF SEX DEVELOPMENT (DSD) / PRIMARY ADRENAL INSUFFICIENCY PANEL HYPOGONADOTROPIC HYPOGONADISM (KALLMANN) PANEL PRIMARY IMMUNODEFICIENCIES PANEL INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS OROFACIAL CLEFTING PANEL WITH GENOME WIDE CNV ANALYSIS
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SEMA4A	100%	100%	100%	98.5%	Retinitis pigmentosa 35, 610282;Cone-rod dystrophy 10, 610283	VISION DISORDERS PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹ HEREDITARY CANCER PANEL
SEMA6B	100%	100%	100%	96.9%	Epilepsy, progressive myoclonic, 11, 618876	EPILEPSY PANEL INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS
SEMA7A	100%	100%	100%	99.3%	?Cholestasis, progressive familial intrahepatic, 11, 619874;[Blood group, John-Milton-Hagen system], 614745	LIVER DISORDERS PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS

SEPHS1	100%	100%	100%	99.7%		METABOLIC DISORDERS PANEL INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS
SEPSECS	99.5%	96.4%	100%	99.2%	Pontocerebellar hypoplasia type 2D, 613811	MOVEMENT DISORDERS PANEL EPILEPSY PANEL METABOLIC DISORDERS PANEL INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL¹
SEPTIN12	100%	100%	100%	98%	Spermatogenic failure 10, 614822	MALE INFERTILITY PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS

SEPTIN4	100%	100%	100%	98.8%		MALE INFERTILITY PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS
SEPTIN9	100%	100%	100%	97.4%	Amyotrophy, hereditary neuralgic, 162100	POLYNEUROPATHIES PANEL ¹ NEUROLOGICAL PAIN DISORDERS PANEL ¹ MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS OROFACIAL CLEFTING PANEL WITH GENOME WIDE CNV ANALYSIS

SERAC1	100%	100%	100%	99.6%	3-methylglutaconic aciduria with deafness, encephalopathy, and Leigh-like syndrome, 614739	MOVEMENT DISORDERS PANEL HEARING IMPAIRMENT PANEL (INCLUDING GJB2) COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹ INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS MITOCHONDRIAL DISORDERS PANEL PRIMARY IMMUNODEFICIENCIES PANEL METABOLIC DISORDERS PANEL
SERPINA1	100%	100%	100%	98.5%	Hemorrhagic diathesis due to antithrombin Pittsburgh, 613490;Emphysema due to AAT deficiency, 613490;Emphysema-cirrhosis, due to AAT deficiency, 613490	LIVER DISORDERS PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹

SERPINA12	100%	100%	100%	99.7%		SKIN DISORDERS PANEL ¹ MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS
SERPINA3	100%	100%	100%	99.3%	Alpha-1-antichymotrypsin deficiency;Cerebrovascular disease, occlusive	SKIN DISORDERS PANEL ¹ MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS
SERPINA6	100%	100%	100%	99.7%	Corticosteroid-binding globulin deficiency, 611489	MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹
SERPINB6	100%	100%	100%	99.3%	?Deafness, autosomal recessive 91, 613453	HEARING IMPAIRMENT PANEL (INCLUDING GJB2) MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS

SERPINB7	100%	100%	100%	99.8%	Palmoplantar keratoderma, Nagashima type, 615598	SKIN DISORDERS PANEL ¹ MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹
SERPINB8	100%	100%	100%	99.7%	Peeling skin syndrome 5, 617115	SKIN DISORDERS PANEL ¹ MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹
SERPINC1	100%	100%	100%	99.8%	Thrombophilia 7 due to antithrombin III deficiency, 613118	HEMOSTATIC/THROMB OTIC DISORDERS PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹

SERPIND1	100%	100%	100%	99.4%	Thrombophilia 10 due to heparin cofactor II deficiency, 612356	HEMOSTATIC/THROMBOTIC DISORDERS PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS
SERPINE1	100%	100%	100%	99.1%	Plasminogen activator inhibitor-1 deficiency, 613329;{Transcription of plasminogen activator inhibitor, modulator of}	HEMOSTATIC/THROMBOTIC DISORDERS PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹
SERPINF1	100%	100%	100%	98.8%	Osteogenesis imperfecta, type VI, 613982	SHORT STATURE/SKELETAL DYSPLASIA PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹

SERPINF2	100%	99.9%	100%	99.4%	Alpha-2-plasmin inhibitor deficiency, 262850	HEMOSTATIC/THROMBOTIC DISORDERS PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹
SERPING1	100%	100%	100%	98.7%	Angioedema, hereditary, 1 and 2, 106100;Complement component 4, partial deficiency of, 120790	SKIN DISORDERS PANEL ¹ PRIMARY IMMUNODEFICIENCIES PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹

SERPINH1	100%	100%	100%	99.4%	{Preterm premature rupture of the membranes, susceptibility to}, 610504;Osteogenesis imperfecta, type X, 613848	SKIN DISORDERS PANEL ¹ SHORT STATURE/SKELETAL DYSPLASIA PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹
SERPINI1	100%	100%	100%	99.2%	Encephalopathy, familial, with neuroserpin inclusion bodies, 604218	EPILEPSY PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS
SET	99.9%	99.2%	100%	98.2%	Intellectual developmental disorder, autosomal dominant 58, 618106	INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS

SETBP1	100%	100%	100%	99%	Schinzel-Giedion midface retraction syndrome, 269150;Intellectual developmental disorder, autosomal dominant 29, 616078	INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS
SETD1A	100%	100%	100%	98.4%	Epilepsy, early-onset, 2, with or without developmental delay, 618832;Neurodevelopmental disorder with speech impairment and dysmorphic facies, 619056	EPILEPSY PANEL INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS
SETD1B	100%	99.9%	99.7%	94.7%	Intellectual developmental disorder with seizures and language delay, 619000	EPILEPSY PANEL INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS

SETD2	100%	100%	100%	99.4%	Luscan-Lumish syndrome, 616831;Intellectual developmental disorder, autosomal dominant 70, 620157;Rabin-Pappas syndrome, 620155	TALL STATURE PANEL WITH GENOME WIDE CNV ANALYSIS SHORT STATURE/SKELETAL DYSPLASIA PANEL WITH GENOME WIDE CNV ANALYSIS INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS
SETD5	99.5%	98.5%	100%	99.3%	Intellectual developmental disorder, autosomal dominant 23, 615761	INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS

SETX	100%	100%	100%	99.4%	Spinocerebellar ataxia, autosomal recessive, with axonal neuropathy 2, 606002;Amyotrophic lateral sclerosis 4, juvenile, 602433	AMYOTROPHIC LATERAL SCLEROSIS, ALS PANEL POLYNEUROPATHIES PANEL ¹ MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹ MOVEMENT DISORDERS PANEL
SEZ6	100%	100%	100%	98.6%		HEARING IMPAIRMENT PANEL (INCLUDING GJB2) MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS
SF3B1	100%	100%	100%	99.6%	Myelodysplastic syndrome, somatic, 614286	IRON DISORDERS PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS

SF3B2	100%	100%	100%	98.9%	Craniofacial microsomia, 164210	CRANIOFACIAL ANOMALIES PANEL WITH GENOME WIDE CNV ANALYSIS CONGENITAL HEARTDISEASE PANEL¹ HEART DISORDERS PANEL¹ MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS
SF3B4	100%	100%	100%	97.8%	Acrofacial dysostosis 1, Nager type, 154400	CRANIOFACIAL ANOMALIES PANEL WITH GENOME WIDE CNV ANALYSIS SHORT STATURE/SKELETAL DYSPLASIA PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS OROFACIAL CLEFTING PANEL WITH GENOME WIDE CNV ANALYSIS

SFRP4	100%	100%	100%	99.7%	Pyle disease, 265900	SHORT STATURE/SKELETAL DYSPLASIA PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹
SFTPA1	100%	100%	100%	98.8%	Interstitial lung disease 1, 619611	MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS HEREDITARY CANCER PANEL
SFTPA2	100%	100%	100%	98.8%	Interstitial lung disease 2, 178500	MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS HEREDITARY CANCER PANEL

SFTPB	100%	99.8%	100%	99.4%	Surfactant metabolism dysfunction, pulmonary, 1, 265120	MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹
SFTPC	100%	100%	100%	99.5%	Surfactant metabolism dysfunction, pulmonary, 2, 610913	MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS
SFXN4	100%	100%	100%	99.6%	Combined oxidative phosphorylation deficiency 18, 615578	IRON DISORDERS PANEL INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS MITOCHONDRIAL DISORDERS PANEL COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹

SGCA	100%	100%	100%	99%	Muscular dystrophy, limb-girdle, autosomal recessive 3, 608099	HEART DISORDERS PANEL ¹ MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹ MUSCLE DISORDERS PANEL
SGCB	100%	100%	100%	99.2%	Muscular dystrophy, limb-girdle, autosomal recessive 4, 604286	HEART DISORDERS PANEL ¹ MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹ MUSCLE DISORDERS PANEL

SGCD	100%	100%	100%	99.6%	Cardiomyopathy, dilated, 1L, 606685;Muscular dystrophy, limb-girdle, autosomal recessive 6, 601287	HEART DISORDERS PANEL ¹ MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹ MUSCLE DISORDERS PANEL
SGCE	90.8%	90.8%	100%	99.4%	Dystonia-11, myoclonic, 159900	MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS MOVEMENT DISORDERS PANEL
SGCG	100%	100%	100%	99.8%	Muscular dystrophy, limb-girdle, autosomal recessive 5, 253700	HEART DISORDERS PANEL ¹ MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹ MUSCLE DISORDERS PANEL

SGMS1	100%	100%	100%	99.6%		METABOLIC DISORDERS PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS
SGMS2	100%	100%	100%	99.5%	Calvarial doughnut lesions with bone fragility with or without spondylometaphyseal dysplasia, 126550	SHORT STATURE/SKELETAL DYSPLASIA PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS
SGO1	100%	100%	100%	99.5%	Chronic atrial and intestinal dysrhythmia, 616201	LIVER DISORDERS PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹

SGPL1	96.6%	96.6%	100%	99.6%	RENI syndrome, 617575	SKIN DISORDERS PANEL ¹ DISORDERS/DIFFERENCES OF SEX DEVELOPMENT (DSD) / PRIMARY ADRENAL INSUFFICIENCY PANEL POLYNEUROPATHIES PANEL ¹ INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS RENAL DISORDERS PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹
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SGSH	100%	100%	100%	98.7%	Mucopolysaccharidosis type IIIA (Sanfilippo A), 252900	VISION DISORDERS PANEL SHORT STATURE/SKELETAL DYSPLASIA PANEL WITH GENOME WIDE CNV ANALYSIS METABOLIC DISORDERS PANEL INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL¹
SGSM3	100%	100%	100%	99%		INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS

SH2B3	100%	100%	100%	99.3%	Thrombocythemia, somatic, 187950;Myelofibrosis, somatic, 254450;Erythrocytosis, somatic, 133100	INHERITED BONE MARROW FAILURE AND/OR PREDISPOSITION PANEL TO HEMATOLOGICAL MALIGNANCIES HEMOSTATIC/THROMBOTIC DISORDERS PANEL PRIMARY IMMUNODEFICIENCIES PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS HEREDITARY CANCER PANEL
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SH2D1A	100%	100%	98.9%	68.7%	Lymphoproliferative syndrome, X-linked, 1, 308240	INHERITED BONE MARROW FAILURE AND/OR PREDISPOSITION PANEL TO HEMATOLOGICAL MALIGNANCIES PRIMARY IMMUNODEFICIENCIES PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS HEREDITARY CANCER PANEL
SH3BP2	99%	97.6%	100%	98.7%	Cherubism, 118400	CRANIOFACIAL ANOMALIES PANEL WITH GENOME WIDE CNV ANALYSIS PRIMARY IMMUNODEFICIENCIES PANEL SHORT STATURE/SKELETAL DYSPLASIA PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS

SH3KBP1	98.8%	98.5%	98.9%	70.1%	?Immunodeficiency 61, 300310	PRIMARY IMMUNODEFICIENCIES PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS
SH3PXD2B	100%	100%	100%	99.2%	Frank-ter Haar syndrome, 249420	VISION DISORDERS PANEL SHORT STATURE/SKELETAL DYSPLASIA PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹
SH3TC2	100%	100%	100%	99.3%	Charcot-Marie-Tooth disease, type 4C, 601596;Mononeuropathy of the median nerve, mild, 613353	POLYNEUROPATHIES PANEL ¹ MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹

SHANK1	100%	100%	100%	94.1%		INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS
SHANK2	100%	100%	100%	98.4%	{Autism susceptibility 17}, 613436	INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS
SHANK3	99.1%	96.3%	99.8%	96.7%	Phelan-McDermid syndrome, 606232;{Schizophrenia 15}, 613950	EPILEPSY PANEL INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS

SHARPIN	100%	100%	100%	99.1%	Autoinflammation with episodic fever and immune dysregulation, 620795	PRIMARY IMMUNODEFICIENCIES PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL¹
SHH	100%	100%	100%	97.8%	Single median maxillary central incisor, 147250;Holoprosencephaly 3, 142945;Microphthalmia/coloboma 5, 611638	VISION DISORDERS PANEL CRANIOFACIAL ANOMALIES PANEL WITH GENOME WIDE CNV ANALYSIS SHORT STATURE/SKELETAL DYSPLASIA PANEL WITH GENOME WIDE CNV ANALYSIS INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS OROFACIAL CLEFTING PANEL WITH GENOME WIDE CNV ANALYSIS

SHMT2	100%	100%	100%	99.7%	Neurodevelopmental disorder with cardiomyopathy, spasticity, and brain abnormalities, 619121	HEART DISORDERS PANEL ¹ METABOLIC DISORDERS PANEL INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS MITOCHONDRIAL DISORDERS PANEL COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹
SHOC1	100%	100%	100%	99.7%	Spermatogenic failure 75, 619949	MALE INFERTILITY PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS

SHOC2	100%	100%	100%	99.4%	Noonan syndrome-like with loose anagen hair 1, 607721	SKIN DISORDERS PANEL ¹ HEART DISORDERS PANEL ¹ SHORT STATURE/SKELETAL DYSPLASIA PANEL WITH GENOME WIDE CNV ANALYSIS LYMPHATIC ANOMALIES PANEL WITH GENOME WIDE CNV ANALYSIS INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS NOONAN SYNDROME / RASOPATHY PANEL HEREDITARY CANCER PANEL
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SHOX	47.4%	47.4%	50%	49.3%	Short stature, idiopathic familial, 300582;Leri-Weill dyschondrosteosis, 127300;Langer mesomelic dysplasia, 249700;Short stature, idiopathic familial, 300582;Langer mesomelic dysplasia, 249700;Leri-Weill dyschondrosteosis, 127300	SHORT STATURE/SKELETAL DYSPLASIA PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS
SHOX2	100%	99.8%	99.4%	92.7%		HEART DISORDERS PANEL ¹ MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS
SHQ1	100%	100%	100%	99.6%	Neurodevelopmental disorder with dystonia and seizures, 619922;?Dystonia 35, childhood-onset, 619921	INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹

SHROOM3	100%	100%	100%	99.2%		CONGENITAL HEARTDISEASE PANEL¹ HEART DISORDERS PANEL¹ MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS
SHROOM4	100%	99.8%	98.9%	70%		INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS
SI	100%	100%	99.8%	98.3%	Sucrase-isomaltase deficiency, congenital, 222900	METABOLIC DISORDERS PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL¹

SIAH1	100%	100%	100%	98.9%	Buratti-Harel syndrome, 619314	INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS
SIGMAR1	100%	100%	100%	99.5%	?Neuronopathy, distal hereditary motor, autosomal recessive 2, 605726;?Amyotrophic lateral sclerosis 16, juvenile, 614373	AMYOTROPHIC LATERAL SCLEROSIS, ALS PANEL POLYNEUROPATHIES PANEL ¹ MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹
SIK1	100%	100%	100%	98.6%	Developmental and epileptic encephalopathy 30, 616341	EPILEPSY PANEL INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS

SIK3	100%	99.8%	100%	98.9%	?Spondyloepimetaphyseal dysplasia, Krakow type, 618162	MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹
SIL1	100%	100%	100%	99.2%	Marinesco-Sjogren syndrome, 248800	MOVEMENT DISORDERS PANEL VISION DISORDERS PANEL INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹ MUSCLE DISORDERS PANEL
SIN3A	100%	100%	100%	99.3%	Witteveen-Kolk syndrome, 613406	INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS

SIN3B	100%	100%	100%	98.7%		INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS
SIPA1L3	100%	100%	100%	99%	?Cataract 45, 616851	VISION DISORDERS PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS
SIRT5	100%	100%	100%	99.4%		MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS MITOCHONDRIAL DISORDERS PANEL

SIX1	100%	100%	100%	99.4%	Deafness, autosomal dominant 23, 605192;Branchiootoc syndrome 3, 608389	CRANIOFACIAL ANOMALIES PANEL WITH GENOME WIDE CNV ANALYSIS HEARING IMPAIRMENT PANEL (INCLUDING GJB2) MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS OROFACIAL CLEFTING PANEL WITH GENOME WIDE CNV ANALYSIS
SIX3	100%	100%	100%	96.5%	Schizencephaly, 269160;Holoprosencephaly 2, 157170	CRANIOFACIAL ANOMALIES PANEL WITH GENOME WIDE CNV ANALYSIS INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS OROFACIAL CLEFTING PANEL WITH GENOME WIDE CNV ANALYSIS

SIX5	100%	100%	100%	97.9%	Branchiootorenal syndrome 2, 610896	CRANIOFACIAL ANOMALIES PANEL WITH GENOME WIDE CNV ANALYSIS HEARING IMPAIRMENT PANEL (INCLUDING GJB2) RENAL DISORDERS PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS OROFACIAL CLEFTING PANEL WITH GENOME WIDE CNV ANALYSIS
SIX6	100%	100%	100%	99.3%	Optic disc anomalies with retinal and/or macular dystrophy, 212550	VISION DISORDERS PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹

SKI	100%	99.7%	100%	97.8%	Shprintzen-Goldberg syndrome, 182212	THORACIC AORTIC ANEURYSM AND/OR DISSECTION PANEL CRANIOFACIAL ANOMALIES PANEL WITH GENOME WIDE CNV ANALYSIS SKIN DISORDERS PANEL ¹ SHORT STATURE/SKELETAL DYSPLASIA PANEL WITH GENOME WIDE CNV ANALYSIS INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS OROFACIAL CLEFTING PANEL WITH GENOME WIDE CNV ANALYSIS
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SKIC2	100%	100%	100%	99.2%	Trichohepatoenteric syndrome 2, 614602	SKIN DISORDERS PANEL ¹ PRIMARY IMMUNODEFICIENCIES PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹
SKIC3	98.9%	98.9%	100%	99.5%	Trichohepatoenteric syndrome 1, 222470	SKIN DISORDERS PANEL ¹ PRIMARY IMMUNODEFICIENCIES PANEL LIVER DISORDERS PANEL INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹

SLC10A1	100%	100%	100%	98.6%	Hypercholanemia, familial 2, 619256	LIVER DISORDERS PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS
SLC10A2	100%	100%	100%	99.5%	?Bile acid malabsorption, primary, 1, 613291	MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS
SLC10A7	92.8%	92.8%	100%	99.6%	Short stature, amelogenesis imperfecta, and skeletal dysplasia with scoliosis, 618363	SHORT STATURE/SKELETAL DYSPLASIA PANEL WITH GENOME WIDE CNV ANALYSIS METABOLIC DISORDERS PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹ OROFACIAL CLEFTING PANEL WITH GENOME WIDE CNV ANALYSIS

SLC11A2	100%	100%	100%	99.5%	Anemia, hypochromic microcytic, with iron overload 1, 206100	IRON DISORDERS PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹
SLC12A1	96.3%	96.3%	100%	99.7%	Bartter syndrome, type 1, 601678	HEARING IMPAIRMENT PANEL (INCLUDING GJB2) METABOLIC DISORDERS PANEL RENAL DISORDERS PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹

SLC12A2	100%	100%	100%	99.4%	Kilquist syndrome, 619080;Delpire-McNeill syndrome, 619083;Deafness, autosomal dominant 78, 619081	HEARING IMPAIRMENT PANEL (INCLUDING GJB2) INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL¹
SLC12A3	100%	100%	100%	98.5%	Gitelman syndrome, 263800	RENAL DISORDERS PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL¹

SLC12A5	100%	99.9%	100%	98.5%	{Epilepsy, idiopathic generalized, susceptibility to, 14}, 616685;Developmental and epileptic encephalopathy 34, 616645	EPILEPSY PANEL INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹
SLC12A6	100%	100%	100%	99.3%	Agenesis of the corpus callosum with peripheral neuropathy, 218000;Charcot-Marie-Tooth disease, axonal, type 2II, 620068	MOVEMENT DISORDERS PANEL POLYNEUROPATHIES PANEL ¹ INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹

SLC12A9	100%	100%	100%	97.8%		INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS
SLC13A3	100%	100%	100%	98.8%	Leukoencephalopathy, acute reversible, with increased urinary alpha-ketoglutarate, 618384	METABOLIC DISORDERS PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹
SLC13A5	100%	100%	100%	99.3%	Developmental and epileptic encephalopathy 25, with amelogenesis imperfecta, 615905	EPILEPSY PANEL INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹

SLC16A1	100%	100%	100%	99.3%	Hyperinsulinemic hypoglycemia, familial, 7, 610021;Erythrocyte lactate transporter defect, 245340;Monocarboxylate transporter 1 deficiency, 616095	EPILEPSY PANEL METABOLIC DISORDERS PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹
SLC16A12	100%	100%	100%	99.5%	Cataract 47, juvenile, with microcornea, 612018	VISION DISORDERS PANEL RENAL DISORDERS PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS
SLC16A2	100%	99.9%	99.1%	69.1%	Allan-Herndon-Dudley syndrome, 300523	MOVEMENT DISORDERS PANEL INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS

SLC17A5	100%	100%	100%	99.4%	Salla disease, 604369;Sialic acid storage disorder, infantile, 269920	SHORT STATURE/SKELETAL DYSPLASIA PANEL WITH GENOME WIDE CNV ANALYSIS METABOLIC DISORDERS PANEL INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹
SLC17A8	100%	100%	100%	99.8%	Deafness, autosomal dominant 25, 605583	HEARING IMPAIRMENT PANEL (INCLUDING GJB2) MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS
SLC17A9	100%	100%	100%	98.8%	Porokeratosis 8, disseminated superficial actinic type, 616063	SKIN DISORDERS PANEL ¹ MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS

SLC18A2	100%	100%	100%	99.2%	Parkinsonism-dystonia, infantile, 2, 618049	MOVEMENT DISORDERS PANEL METABOLIC DISORDERS PANEL INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹
SLC18A3	100%	100%	100%	99.3%	Myasthenic syndrome, congenital, 21, presynaptic, 617239	MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹ MUSCLE DISORDERS PANEL
SLC19A1	100%	100%	100%	98%	Immunodeficiency 114, folate-responsive, 620603;?Megaloblastic anemia, folate-responsive, 601775	PRIMARY IMMUNODEFICIENCIES PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS

SLC19A2	100%	100%	100%	99.6%	Thiamine-responsive megaloblastic anemia syndrome, 249270	VISION DISORDERS PANEL INHERITED BONE MARROW FAILURE AND/OR PREDISPOSITION PANEL TO HEMATOLOGICAL MALIGNANCIES HEARING IMPAIRMENT PANEL (INCLUDING GJB2) IRON DISORDERS PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS MITOCHONDRIAL DISORDERS PANEL COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹
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SLC19A3	99.7%	98.6%	100%	99.4%	Thiamine metabolism dysfunction syndrome 2 (biotin/thiamine-responsive basal ganglia disease type), 607483	COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹ MOVEMENT DISORDERS PANEL EPILEPSY PANEL INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS MITOCHONDRIAL DISORDERS PANEL
SLC1A1	100%	100%	100%	99.5%	Dicarboxylic aminoaciduria, 222730;{?Schizophrenia susceptibility 18}, 615232	METABOLIC DISORDERS PANEL INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹

SLC1A2	100%	100%	100%	99.4%	Developmental and epileptic encephalopathy 41, 617105	EPILEPSY PANEL INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS
SLC1A3	97%	94.1%	100%	99.2%	Episodic ataxia, type 6, 612656	MOVEMENT DISORDERS PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS
SLC1A4	100%	100%	100%	99.1%	Spastic tetraplegia, thin corpus callosum, and progressive microcephaly, 616657	METABOLIC DISORDERS PANEL INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹

SLC20A2	100%	100%	100%	98.8%	Basal ganglia calcification, idiopathic, 1, 213600	MOVEMENT DISORDERS PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS PARKINSON DISEASE PANEL
SLC22A12	100%	99.7%	99.7%	95.8%	Hypouricemia, renal, 220150	METABOLIC DISORDERS PANEL RENAL DISORDERS PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹
SLC22A18	100%	100%	100%	98.9%	Breast cancer, somatic, 114480;Lung cancer, somatic, 211980;Rhabdomyosarcoma, somatic, 268210	MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS
SLC22A4	100%	100%	100%	99.6%	{Rheumatoid arthritis, susceptibility to}, 180300	HEARING IMPAIRMENT PANEL (INCLUDING GJB2) MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS

SLC22A5	100%	100%	100%	99.2%	Carnitine deficiency, systemic primary, 212140	HEART DISORDERS PANEL ¹ METABOLIC DISORDERS PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹
SLC24A1	100%	100%	100%	99%	Night blindness, congenital stationary (complete), 1D, autosomal recessive, 613830	VISION DISORDERS PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹

SLC24A4	100%	100%	100%	99.5%	[Skin/hair/eye pigmentation 6, blond/brown hair], 210750;Amelogenesis imperfecta, type IIA5, 615887;[Skin/hair/eye pigmentation 6, blue/green eyes], 210750	CRANIOFACIAL ANOMALIES PANEL WITH GENOME WIDE CNV ANALYSIS SKIN DISORDERS PANEL ¹ MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹
SLC24A5	100%	100%	100%	99.5%	[Skin/hair/eye pigmentation 4, fair/dark skin], 113750;Albinism, oculocutaneous, type VI, 113750	VISION DISORDERS PANEL SKIN DISORDERS PANEL ¹ MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹

SLC25A1	100%	100%	100%	98.1%	Combined D-2- and L-2-hydroxyglutaric aciduria, 615182;Myasthenic syndrome, congenital, 23, presynaptic, 618197	EPILEPSY PANEL MUSCLE DISORDERS PANEL METABOLIC DISORDERS PANEL INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS MITOCHONDRIAL DISORDERS PANEL COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹
SLC25A10	100%	100%	100%	99.7%	?Mitochondrial DNA depletion syndrome 19, 618972	MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS MITOCHONDRIAL DISORDERS PANEL
SLC25A11	100%	100%	100%	99%	Pheochromocytoma/paraganglioma syndrome 6, 618464	MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS HEREDITARY CANCER PANEL

SLC25A12	100%	100%	100%	99.5%	Developmental and epileptic encephalopathy 39, 612949	INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS MITOCHONDRIAL DISORDERS PANEL COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL¹
SLC25A13	100%	100%	100%	99.7%	Citrullinemia, type II, neonatal-onset, 605814;Citrullinemia, adult-onset type II, 603471	LIVER DISORDERS PANEL METABOLIC DISORDERS PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS MITOCHONDRIAL DISORDERS PANEL COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL¹

SLC25A15	100%	100%	100%	99.6%	Hyperornithinemia-hyperammonemia-homocitrullinemia syndrome, 238970	MOVEMENT DISORDERS PANEL EPILEPSY PANEL METABOLIC DISORDERS PANEL INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹
SLC25A19	100%	100%	100%	99%	Microcephaly, Amish type, 607196;Thiamine metabolism dysfunction syndrome 4 (progressive polyneuropathy type), 613710	POLYNEUROPATHIES PANEL ¹ METABOLIC DISORDERS PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS MITOCHONDRIAL DISORDERS PANEL COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹

SLC25A20	100%	100%	100%	99.2%	Carnitine-acylcarnitine translocase deficiency, 212138	HEART DISORDERS PANEL ¹ METABOLIC DISORDERS PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹
SLC25A21	100%	100%	100%	99.6%	?Mitochondrial DNA depletion syndrome 18, 618811	METABOLIC DISORDERS PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS MITOCHONDRIAL DISORDERS PANEL

SLC25A22	100%	100%	100%	99.5%	Developmental and epileptic encephalopathy 3, 609304	EPILEPSY PANEL INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS MITOCHONDRIAL DISORDERS PANEL COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL¹
SLC25A24	99%	99%	99.5%	96.8%	Fontaine progeroid syndrome, 612289	SHORT STATURE/SKELETAL DYSPLASIA PANEL WITH GENOME WIDE CNV ANALYSIS INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS MITOCHONDRIAL DISORDERS PANEL

SLC25A26	100%	100%	100%	99.4%	Combined oxidative phosphorylation deficiency 28, 616794	MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS MITOCHONDRIAL DISORDERS PANEL COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹
SLC25A3	100%	100%	100%	99.5%	Mitochondrial phosphate carrier deficiency, 610773	MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS MITOCHONDRIAL DISORDERS PANEL COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹
SLC25A32	100%	100%	100%	99.1%	?Exercise intolerance, riboflavin-responsive, 616839	METABOLIC DISORDERS PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS MITOCHONDRIAL DISORDERS PANEL

SLC25A36	100%	100%	100%	97.7%	Hyperinsulinemic hypoglycemia, familial, 8, 620211	METABOLIC DISORDERS PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS MITOCHONDRIAL DISORDERS PANEL COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹
SLC25A38	100%	100%	100%	99.2%	Anemia, sideroblastic, 2, pyridoxine-refractory, 205950	INHERITED BONE MARROW FAILURE AND/OR PREDISPOSITION PANEL TO HEMATOLOGICAL MALIGNANCIES IRON DISORDERS PANEL METABOLIC DISORDERS PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS MITOCHONDRIAL DISORDERS PANEL COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹

SLC25A4	100%	100%	100%	99.5%	Mitochondrial DNA depletion syndrome 12B (cardiomyopathic type) AR, 615418;Progressive external ophthalmoplegia with mitochondrial DNA deletions, autosomal dominant 2, 609283;Mitochondrial DNA depletion syndrome 12A (cardiomyopathic type) AD, 617184	HEART DISORDERS PANEL ¹ MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS MITOCHONDRIAL DISORDERS PANEL COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹ MUSCLE DISORDERS PANEL
SLC25A42	100%	100%	100%	98.4%	Metabolic crises, recurrent, with variable encephalomyopathic features and neurologic regression, 618416	METABOLIC DISORDERS PANEL INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS MITOCHONDRIAL DISORDERS PANEL COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹

SLC25A46	100%	100%	100%	99.7%	Neuropathy, hereditary motor and sensory, type VIB, 616505;Pontocerebellar hypoplasia, type 1E, 619303	MOVEMENT DISORDERS PANEL VISION DISORDERS PANEL POLYNEUROPATHIES PANEL ¹ MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS MITOCHONDRIAL DISORDERS PANEL COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹
SLC26A1	100%	100%	100%	99.3%	?Hypersulfaturia, 620372;?Nephrolithiasis, calcium oxalate, 1, 167030	RENAL DISORDERS PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹

SLC26A2	100%	100%	100%	99.8%	Epiphyseal dysplasia, multiple, 4, 226900;De la Chapelle dysplasia, 256050;Diastrophic dysplasia, 222600;Diastrophic dysplasia, broad bone-platyspondylic variant, 222600;Achondrogenesis Ib, 600972;Atelosteogenesis, type II, 256050	CRANIOFACIAL ANOMALIES PANEL WITH GENOME WIDE CNV ANALYSIS OROFACIAL CLEFTING PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹ MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS SHORT STATURE/SKELETAL DYSPLASIA PANEL WITH GENOME WIDE CNV ANALYSIS SKIN DISORDERS PANEL ¹
SLC26A3	100%	100%	100%	99.7%	Diarrhea 1, secretory chloride, congenital, 214700	RENAL DISORDERS PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹

SLC26A4	100%	100%	100%	99.3%	Deafness, autosomal recessive 4, with enlarged vestibular aqueduct, 600791;Pendred syndrome, 274600	MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹ HEARING IMPAIRMENT PANEL (INCLUDING GJB2)
SLC26A5	100%	100%	100%	99.6%	?Deafness, autosomal recessive 61, 613865	HEARING IMPAIRMENT PANEL (INCLUDING GJB2) MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS
SLC26A8	100%	100%	99.9%	97.5%	Spermatogenic failure 3, 606766	MALE INFERTILITY PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS

SLC27A4	100%	100%	100%	98.5%	Ichthyosis prematurity syndrome, 608649	SKIN DISORDERS PANEL ¹ MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹
SLC28A1	100%	100%	100%	97.9%	[Uridine-cytidineuria], 618477	METABOLIC DISORDERS PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS

SLC29A3	100%	100%	100%	99%	Histiocytosis-lymphadenopathy plus syndrome, 602782	SKIN DISORDERS PANEL ¹ HEARING IMPAIRMENT PANEL (INCLUDING GJB2) PRIMARY IMMUNODEFICIENCIES PANEL SHORT STATURE/SKELETAL DYSPLASIA PANEL WITH GENOME WIDE CNV ANALYSIS METABOLIC DISORDERS PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹
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SLC2A1	100%	100%	100%	99.2%	Dystonia 9, 601042;GLUT1 deficiency syndrome 1, infantile onset, severe, 606777;Stomatin-deficient cryohydrocytosis with neurologic defects, 608885;{Epilepsy, idiopathic generalized, susceptibility to, 12}, 614847;GLUT1 deficiency syndrome 2, childhood onset, 612126	MOVEMENT DISORDERS PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹ METABOLIC DISORDERS PANEL INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS EPILEPSY PANEL
SLC2A10	100%	100%	100%	99.3%	Arterial tortuosity syndrome, 208050	THORACIC AORTIC ANEURYSM AND/OR DISSECTION PANEL SKIN DISORDERS PANEL ¹ MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹

SLC2A2	100%	100%	100%	99.5%	Fanconi-Bickel syndrome, 227810;{Diabetes mellitus, noninsulin-dependent}, 125853	METABOLIC DISORDERS PANEL RENAL DISORDERS PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹
SLC2A9	100%	100%	100%	98.5%	{Uric acid concentration, serum, QTL 2}, 612076;Hypouricemia, renal, 2, 612076	METABOLIC DISORDERS PANEL RENAL DISORDERS PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹

SLC30A10	100%	100%	100%	98.6%	Hypermanganesemia with dystonia 1, 613280	MOVEMENT DISORDERS PANEL METABOLIC DISORDERS PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS PARKINSON DISEASE PANEL COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹
SLC30A2	100%	100%	100%	98.8%	Zinc deficiency, transient neonatal, 608118	METABOLIC DISORDERS PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS
SLC30A5	100%	100%	100%	98.9%		HEART DISORDERS PANEL ¹ MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS

SLC30A7	100%	100%	100%	99.8%	Ziegler-Huang syndrome, 620501	MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹
SLC30A9	100%	100%	100%	99.4%	Birk-Landau-Perez syndrome, 617595	HEARING IMPAIRMENT PANEL (INCLUDING GJB2) METABOLIC DISORDERS PANEL INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹

SLC31A1	100%	100%	100%	99.3%	Neurodegeneration and seizures due to copper transport defect, 620306	INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹
SLC32A1	100%	100%	100%	99.3%	Generalized epilepsy with febrile seizures plus, type 12, 620755;Developmental and epileptic encephalopathy 114, 620774	EPILEPSY PANEL INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS

SLC33A1	100%	100%	100%	98.8%	Spastic paraplegia 42, autosomal dominant, 612539;Huppke-Brendel syndrome, 614482	MOVEMENT DISORDERS PANEL VISION DISORDERS PANEL HEARING IMPAIRMENT PANEL (INCLUDING GJB2) METABOLIC DISORDERS PANEL INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹
SLC34A1	100%	100%	100%	98.8%	?Fanconi renotubular syndrome 2, 613388;Hypercalcemia, infantile, 2, 616963;Nephrolithiasis/osteoporosis, hypophosphatemic, 1, 612286	RENAL DISORDERS PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹

SLC34A2	100%	100%	100%	98.9%	Pulmonary alveolar microlithiasis, 265100	MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹
SLC34A3	100%	100%	100%	97.2%	Hypophosphatemic rickets with hypercalciuria, 241530	SHORT STATURE/SKELETAL DYSPLASIA PANEL WITH GENOME WIDE CNV ANALYSIS RENAL DISORDERS PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹

SLC35A1	100%	100%	100%	99.3%	Congenital disorder of glycosylation, type II _f , 603585	PRIMARY IMMUNODEFICIENCIES PANEL METABOLIC DISORDERS PANEL INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹
SLC35A2	100%	99.9%	98.5%	67.2%	Congenital disorder of glycosylation, type II _m , 300896	EPILEPSY PANEL METABOLIC DISORDERS PANEL INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS

SLC35A3	97.6%	90%	100%	99.4%	Arthrogryposis, impaired intellectual development, and seizures, 615553	FETAL AKINESIA PANEL METABOLIC DISORDERS PANEL INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹
SLC35B2	100%	100%	100%	99.2%	Leukodystrophy, hypomyelinating, 26, with chondrodysplasia, 620269	INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹

SLC35C1	100%	100%	100%	99.1%	Congenital disorder of glycosylation, type IIc, 266265	PRIMARY IMMUNODEFICIENCIES PANEL SHORT STATURE/SKELETAL DYSPLASIA PANEL WITH GENOME WIDE CNV ANALYSIS METABOLIC DISORDERS PANEL INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL¹
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SLC35D1	100%	100%	100%	99.7%	Schneckenbecken dysplasia, 269250	SHORT STATURE/SKELETAL DYSPLASIA PANEL WITH GENOME WIDE CNV ANALYSIS METABOLIC DISORDERS PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL¹
SLC36A2	100%	100%	100%	98.9%	[Iminoglycinuria], 242600;[Hyperglycinuria], 138500	METABOLIC DISORDERS PANEL RENAL DISORDERS PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS
SLC37A3	100%	100%	100%	99.6%		VISION DISORDERS PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS

SLC37A4	100%	99.9%	100%	99.4%	Glycogen storage disease Ib, 232220;Congenital disorder of glycosylation, type IIw, 619525;Glycogen storage disease Ic, 232240	RENAL DISORDERS PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹ INHERITED BONE MARROW FAILURE AND/OR PREDISPOSITION PANEL TO HEMATOLOGICAL MALIGNANCIES PRIMARY IMMUNODEFICIENCIES PANEL METABOLIC DISORDERS PANEL
SLC38A3	100%	100%	100%	98.6%	Developmental and epileptic encephalopathy 102, 619881	INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS EPILEPSY PANEL METABOLIC DISORDERS PANEL

SLC38A8	100%	100%	100%	98.5%	Foveal hypoplasia 2, with or without optic nerve misrouting and/or anterior segment dysgenesis, 609218	VISION DISORDERS PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹
SLC39A12	100%	100%	100%	99.3%		VISION DISORDERS PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS
SLC39A13	100%	100%	100%	98.5%	Ehlers-Danlos syndrome, spondylodysplastic type, 3, 612350	SKIN DISORDERS PANEL ¹ SHORT STATURE/SKELETAL DYSPLASIA PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹

SLC39A14	93.6%	93.6%	100%	98.9%	?Hyperostosis cranalis interna, 144755;Hypermanganesemia with dystonia 2, 617013	MOVEMENT DISORDERS PANEL METABOLIC DISORDERS PANEL INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS PARKINSON DISEASE PANEL COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL¹
SLC39A4	100%	100%	100%	99.2%	Acrodermatitis enteropathica, 201100	MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL¹ SKIN DISORDERS PANEL¹ PRIMARY IMMUNODEFICIENCIES PANEL METABOLIC DISORDERS PANEL

SLC39A5	100%	100%	100%	98.5%	Myopia 24, autosomal dominant, 615946	VISION DISORDERS PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS
SLC39A7	100%	100%	100%	99.2%	Agammaglobulinemia 9, autosomal recessive, 619693	PRIMARY IMMUNODEFICIENCIES PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹
SLC39A8	100%	99.6%	100%	99.7%	Congenital disorder of glycosylation, type IIa, 616721	METABOLIC DISORDERS PANEL INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS MITOCHONDRIAL DISORDERS PANEL COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹

SLC3A1	96.2%	96.2%	100%	99.5%	Cystinuria, 220100	METABOLIC DISORDERS PANEL COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹ RENAL DISORDERS PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS
SLC40A1	100%	100%	100%	99.5%	Hemochromatosis, type 4, 606069	IRON DISORDERS PANEL LIVER DISORDERS PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS
SLC41A1	100%	100%	100%	99.3%	?Nephronophthisis-like nephropathy 2, 619468	RENAL DISORDERS PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS

SLC44A1	100%	100%	100%	99.2%	Neurodegeneration, childhood-onset, with ataxia, tremor, optic atrophy, and cognitive decline, 618868	METABOLIC DISORDERS PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹
SLC44A4	100%	100%	100%	98.6%	?Deafness, autosomal dominant 72, 617606	HEARING IMPAIRMENT PANEL (INCLUDING GJB2) MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS
SLC45A1	100%	100%	100%	98.9%	Intellectual developmental disorder with neuropsychiatric features, 617532	INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹

SLC45A2	100%	100%	100%	99.5%	[Skin/hair/eye pigmentation 5, dark/light eyes], 227240;[Skin/hair/eye pigmentation 5, black/nonblack hair], 227240;Albinism, oculocutaneous, type IV, 606574;[Skin/hair/eye pigmentation 5, dark/fair skin], 227240	VISION DISORDERS PANEL SKIN DISORDERS PANEL ¹ MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹
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SLC46A1	100%	100%	100%	98.6%	Folate malabsorption, hereditary, 229050	INHERITED BONE MARROW FAILURE AND/OR PREDISPOSITION PANEL TO HEMATOLOGICAL MALIGNANCIES IRON DISORDERS PANEL PRIMARY IMMUNODEFICIENCIES PANEL METABOLIC DISORDERS PANEL INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹
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SLC4A1	100%	100%	100%	98.5%	[Blood group, Swann], 601550;[Blood group, Wright], 112050;Distal renal tubular acidosis 1, 179800;[Blood group, Waldner], 112010;Spherocytosis, type 4, 612653;[Blood group, Froese], 601551;Distal renal tubular acidosis 4 with hemolytic anemia, 611590;{Malaria, resistance to}, 611162;Cryohydrocytosis, 185020;Ovalocytosis, SA type, 166900;[Blood group, Diego], 110500	RENAL DISORDERS PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹
SLC4A10	100%	100%	100%	99.2%	Neurodevelopmental disorder with hypotonia and characteristic brain abnormalities, 620746	INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹
SLC4A11	100%	100%	100%	99.1%	Corneal endothelial dystrophy, autosomal recessive, 217700;Corneal dystrophy, Fuchs endothelial, 4, 613268;Corneal endothelial dystrophy and perceptive deafness, 217400	VISION DISORDERS PANEL HEARING IMPAIRMENT PANEL (INCLUDING GJB2) MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹

SLC4A2	100%	100%	100%	98.8%	?Osteopetrosis, autosomal recessive 9, 620366	INHERITED BONE MARROW FAILURE AND/OR PREDISPOSITION PANEL TO HEMATOLOGICAL MALIGNANCIES SHORT STATURE/SKELETAL DYSPLASIA PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS
SLC4A3	100%	100%	100%	98.2%	Short QT syndrome 7, 620231	HEART DISORDERS PANEL ¹ MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS ARRHYTHMIA AND CARDIAC CONDUCTION DISORDERS PANEL ¹

SLC4A4	97.3%	97.3%	100%	99.3%	Proximal renal tubular acidosis-ocular anomaly syndrome, 604278	SKIN DISORDERS PANEL ¹ INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS RENAL DISORDERS PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹ VISION DISORDERS PANEL
SLC4A7	100%	100%	100%	99.5%		VISION DISORDERS PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS
SLC51A	100%	100%	100%	98.3%	?Cholestasis, progressive familial intrahepatic, 6, 619484	LIVER DISORDERS PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS

SLC51B	100%	100%	100%	99.5%	?Bile acid malabsorption, primary, 2, 619481	MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS
SLC52A1	100%	100%	100%	99.4%	Riboflavin deficiency, 615026	METABOLIC DISORDERS PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS
SLC52A2	100%	100%	100%	99.9%	Brown-Vialetto-Van Laere syndrome 2, 614707	MOVEMENT DISORDERS PANEL VISION DISORDERS PANEL HEARING IMPAIRMENT PANEL (INCLUDING GJB2) POLYNEUROPATHIES PANEL ¹ METABOLIC DISORDERS PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS MITOCHONDRIAL DISORDERS PANEL COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹ MUSCLE DISORDERS PANEL

SLC52A3	100%	100%	100%	99.3%	?Fazio-Londe disease, 211500;Brown-Vialetto-Van Laere syndrome 1, 211530	MOVEMENT DISORDERS PANEL HEARING IMPAIRMENT PANEL (INCLUDING GJB2) POLYNEUROPATHIES PANEL ¹ METABOLIC DISORDERS PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS MITOCHONDRIAL DISORDERS PANEL COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹ MUSCLE DISORDERS PANEL
SLC5A1	100%	99.9%	100%	98.9%	Glucose/galactose malabsorption, 606824	METABOLIC DISORDERS PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹

SLC5A2	100%	100%	100%	99.2%	Renal glucosuria, 233100	METABOLIC DISORDERS PANEL RENAL DISORDERS PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹
SLC5A5	100%	100%	100%	97.9%	Thyroid dyshormonogenesis 1, 274400	MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹
SLC5A6	100%	100%	100%	98.9%	Sodium-dependent multivitamin transporter deficiency, 618973;Peripheral motor neuropathy, childhood-onset, biotin-responsive, 619903	POLYNEUROPATHIES PANEL ¹ INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹

SLC5A7	100%	100%	100%	98.9%	Neuronopathy, distal hereditary motor, autosomal dominant 7, 158580;Myasthenic syndrome, congenital, 20, presynaptic, 617143	FETAL AKINESIA PANEL POLYNEUROPATHIES PANEL ¹ INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹ MUSCLE DISORDERS PANEL
SLC66A1	100%	100%	100%	99.2%		VISION DISORDERS PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS
SLC6A1	100%	99.9%	100%	98.3%	Myoclonic-atonic epilepsy, 616421	EPILEPSY PANEL INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS

SLC6A17	100%	100%	100%	99%	Intellectual developmental disorder, autosomal recessive 48, 616269	INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹
SLC6A19	100%	99.9%	100%	98.4%	Hartnup disorder, 234500	SKIN DISORDERS PANEL ¹ METABOLIC DISORDERS PANEL INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS RENAL DISORDERS PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹
SLC6A2	100%	100%	100%	99.1%	?Orthostatic intolerance, 604715	MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS

SLC6A20	100%	100%	100%	98.8%		RENAL DISORDERS PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS
SLC6A3	100%	100%	100%	98.9%	Parkinsonism-dystonia, infantile, 1, 613135;{Nicotine dependence, protection against}, 188890	MOVEMENT DISORDERS PANEL INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS PARKINSON DISEASE PANEL COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL¹
SLC6A5	100%	100%	100%	99.1%	Hyperekplexia 3, 614618	MOVEMENT DISORDERS PANEL METABOLIC DISORDERS PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL¹

SLC6A6	100%	100%	100%	98.4%	Hypotaurinemic retinal degeneration and cardiomyopathy, 145350	HEART DISORDERS PANEL ¹ METABOLIC DISORDERS PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹
SLC6A8	100%	99.2%	98.7%	65.3%	Cerebral creatine deficiency syndrome 1, 300352	EPILEPSY PANEL METABOLIC DISORDERS PANEL INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS

SLC6A9	100%	100%	100%	98.6%	Glycine encephalopathy with normal serum glycine, 617301	FETAL AKINESIA PANEL METABOLIC DISORDERS PANEL INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹
SLC7A14	100%	100%	100%	99.4%	Retinitis pigmentosa 68, 615725	COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹ VISION DISORDERS PANEL HEARING IMPAIRMENT PANEL (INCLUDING GJB2) MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS
SLC7A6OS	100%	100%	100%	99.4%	Epilepsy, progressive myoclonic, 12, 619191	MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS

SLC7A7	100%	100%	100%	97.1%	Lysinuric protein intolerance, 222700	SKIN DISORDERS PANEL ¹ COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹ PRIMARY IMMUNODEFICIENCIES PANEL METABOLIC DISORDERS PANEL INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS RENAL DISORDERS PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS
SLC7A9	100%	100%	100%	98.7%	Cystinuria, 220100	METABOLIC DISORDERS PANEL RENAL DISORDERS PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹

SLC8B1	100%	100%	100%	99.1%		MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS MITOCHONDRIAL DISORDERS PANEL
SLC9A1	100%	100%	100%	98.4%	Lichtenstein-Knorr syndrome, 616291	MOVEMENT DISORDERS PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹
SLC9A3	99.7%	98.6%	100%	97.1%	Diarrhea 8, secretory sodium, congenital, 616868	RENAL DISORDERS PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹

SLC9A6	100%	99.8%	98.9%	68.2%	Intellectual developmental disorder, X-linked syndromic, Christianson type, 300243	EPILEPSY PANEL INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS
SLC9A7	100%	99.5%	98.8%	70.1%	Intellectual developmental disorder, X-linked 108, 301024	INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS
SLCO1B1	100%	99.9%	99.9%	98.6%	Hyperbilirubinemia, Rotor type, digenic, 237450	METABOLIC DISORDERS PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS
SLCO1B3	100%	100%	100%	99.5%	Hyperbilirubinemia, Rotor type, digenic, 237450	METABOLIC DISORDERS PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS

SLCO2A1	100%	100%	100%	98.9%	Hypertrophic osteoarthropathy, primary, autosomal dominant, 167100;PHOAR2-enteropathy syndrome, 614441	SKIN DISORDERS PANEL ¹ SHORT STATURE/SKELETAL DYSPLASIA PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹
SLCO5A1	100%	100%	100%	99.2%		SHORT STATURE/SKELETAL DYSPLASIA PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS

SLF2	100%	100%	100%	99.5%	Atelis syndrome 1, 620184	INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹
SLFN14	100%	100%	100%	99.4%	Bleeding disorder, platelet-type, 20, 616913	HEMOSTATIC/THROMB OTIC DISORDERS PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS
SLIRP	100%	100%	100%	99.5%		MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS MITOCHONDRIAL DISORDERS PANEL
SLIT3	100%	100%	100%	99%		RENAL DISORDERS PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS

SLITRK1	100%	100%	100%	99.4%	Tourette syndrome, 137580;?Trichotillomania, 613229	MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS
SLITRK2	100%	100%	98.9%	69.2%	Intellectual developmental disorder, X-linked 111, 301107	INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS
SLITRK6	100%	100%	100%	99.5%	Deafness and myopia, 221200	VISION DISORDERS PANEL HEARING IMPAIRMENT PANEL (INCLUDING GJB2) MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹
SLMAP	100%	100%	100%	99.6%		HEART DISORDERS PANEL ¹ MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS

SLURP1	100%	100%	100%	99.3%	Meleda disease, 248300	SKIN DISORDERS PANEL ¹ MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹
SLX4	100%	100%	100%	98.9%	Fanconi anemia, complementation group P, 613951	INHERITED BONE MARROW FAILURE AND/OR PREDISPOSITION PANEL TO HEMATOLOGICAL MALIGNANCIES SKIN DISORDERS PANEL ¹ SHORT STATURE/SKELETAL DYSPLASIA PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹ HEREDITARY CANCER PANEL

SMAD1	100%	100%	100%	99.3%		HEART DISORDERS PANEL ¹ MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS
SMAD2	100%	100%	100%	99.1%	Loeys-Dietz syndrome 6, 619656;Congenital heart defects, multiple types, 8, with or without heterotaxy, 619657	THORACIC AORTIC ANEURYSM AND/OR DISSECTION PANEL CONGENITAL HEARTDISEASE PANEL ¹ TALL STATURE PANEL WITH GENOME WIDE CNV ANALYSIS HEART DISORDERS PANEL ¹ SHORT STATURE/SKELETAL DYSPLASIA PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS

SMAD3	100%	100%	100%	98.9%	Loeys-Dietz syndrome 3, 613795	THORACIC AORTIC ANEURYSM AND/OR DISSECTION PANEL SKIN DISORDERS PANEL ¹ TALL STATURE PANEL WITH GENOME WIDE CNV ANALYSIS SHORT STATURE/SKELETAL DYSPLASIA PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS OROFACIAL CLEFTING PANEL WITH GENOME WIDE CNV ANALYSIS
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SMAD4	100%	100%	100%	99.5%	Pancreatic cancer, somatic, 260350;Myhre syndrome, 139210;Polyposis, juvenile intestinal, 174900;Juvenile polyposis/hereditary hemorrhagic telangiectasia syndrome, 175050	THORACIC AORTIC ANEURYSM AND/OR DISSECTION PANEL PANEL HEREDITARY COLORECTAL AND POLYPOSIS OROFACIAL CLEFTING PANEL WITH GENOME WIDE CNV ANALYSIS HEREDITARY CANCER PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS HEMOSTATIC/THROMBOTIC DISORDERS PANEL SHORT STATURE/SKELETAL DYSPLASIA PANEL WITH GENOME WIDE CNV ANALYSIS
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SMAD5	100%	100%	100%	99.7%		CONGENITAL HEARTDISEASE PANEL¹ HEART DISORDERS PANEL¹ MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS
SMAD6	100%	100%	100%	96.7%	Aortic valve disease 2, 614823;{Radioulnar synostosis, nonsyndromic}, 179300;{Craniosynostosis 7, susceptibility to}, 617439	MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS SHORT STATURE/SKELETAL DYSPLASIA PANEL WITH GENOME WIDE CNV ANALYSIS CONGENITAL HEARTDISEASE PANEL¹ HEART DISORDERS PANEL¹ THORACIC AORTIC ANEURYSM AND/OR DISSECTION PANEL CRANIOFACIAL ANOMALIES PANEL WITH GENOME WIDE CNV ANALYSIS

SMAD9	100%	100%	100%	98.4%	Pulmonary hypertension, primary, 2, 615342	MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS HEREDITARY CANCER PANEL
SMARCA1	100%	100%	98.7%	69.1%		INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS
SMARCA2	97.9%	97.1%	100%	99.3%	Nicolaides-Baraitser syndrome, 601358;Blepharophimosis-impaired intellectual development syndrome, 619293	SKIN DISORDERS PANEL ¹ EPILEPSY PANEL INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS

SMARCA4	100%	100%	100%	99%	Coffin-Siris syndrome 4, 614609;{Rhabdoid tumor predisposition syndrome 2}, 613325;?Otosclerosis 12, 620792	CONGENITAL HEARTDISEASE PANEL¹ SKIN DISORDERS PANEL¹ HEART DISORDERS PANEL¹ SHORT STATURE/SKELETAL DYSPLASIA PANEL WITH GENOME WIDE CNV ANALYSIS INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS HEREDITARY CANCER PANEL
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SMARCA5	100%	100%	100%	99.2%		SHORT STATURE/SKELETAL DYSPLASIA PANEL WITH GENOME WIDE CNV ANALYSIS INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS
SMARCA1	100%	100%	100%	99.5%	Basan syndrome, 129200;Huriez syndrome, 181600;Adermatoglyphia, 136000	SKIN DISORDERS PANEL ¹ MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS

SMARCAL1	100%	100%	100%	99.1%	Schimke immunoosseous dysplasia, 242900	SKIN DISORDERS PANEL ¹ PRIMARY IMMUNODEFICIENCIES PANEL SHORT STATURE/SKELETAL DYSPLASIA PANEL WITH GENOME WIDE CNV ANALYSIS RENAL DISORDERS PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹
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SMARCB1	100%	99.9%	100%	98.4%	Rhabdoid tumors, somatic, 609322;{Schwannomatosis-1, susceptibility to}, 162091;Coffin-Siris syndrome 3, 614608;{Rhabdoid tumor predisposition syndrome 1}, 609322	SKIN DISORDERS PANEL ¹ INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS SONIC HEDGEHOG MEDULLOBLASTOMA PANEL HEREDITARY CANCER PANEL NEUROLOGICAL PAIN DISORDERS PANEL ¹ SHORT STATURE/SKELETAL DYSPLASIA PANEL WITH GENOME WIDE CNV ANALYSIS
SMARCC2	100%	100%	100%	98.3%	Coffin-Siris syndrome 8, 618362	INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS

SMARCD1	100%	100%	100%	98.7%	Coffin-Siris syndrome 11, 618779	INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS
SMARCD2	100%	100%	100%	98%	Specific granule deficiency 2, 617475	PRIMARY IMMUNODEFICIENCIES PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹

SMARCE1	100%	100%	100%	98.9%	{Meningioma, familial, susceptibility to}, 607174;Coffin-Siris syndrome 5, 616938	SHORT STATURE/SKELETAL DYSPLASIA PANEL WITH GENOME WIDE CNV ANALYSIS INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS HEREDITARY CANCER PANEL
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SMC1A	100%	99.7%	98.5%	65%	Cornelia de Lange syndrome 2, 300590;Developmental and epileptic encephalopathy 85, with or without midline brain defects, 301044	CRANIOFACIAL ANOMALIES PANEL WITH GENOME WIDE CNV ANALYSIS EPILEPSY PANEL SHORT STATURE/SKELETAL DYSPLASIA PANEL WITH GENOME WIDE CNV ANALYSIS INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS OROFACIAL CLEFTING PANEL WITH GENOME WIDE CNV ANALYSIS
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SMC3	100%	100%	100%	99.3%	Cornelia de Lange syndrome 3, 610759	CRANIOFACIAL ANOMALIES PANEL WITH GENOME WIDE CNV ANALYSIS SHORT STATURE/SKELETAL DYSPLASIA PANEL WITH GENOME WIDE CNV ANALYSIS INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS OROFACIAL CLEFTING PANEL WITH GENOME WIDE CNV ANALYSIS
SMC5	100%	100%	100%	99.5%	Atelis syndrome 2, 620185	INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹

SMCHD1	100%	100%	100%	99.6%	Facioscapulohumeral muscular dystrophy 2, digenic, 158901;Bosma arhinia microphthalmia syndrome, 603457	MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS OROFACIAL CLEFTING PANEL WITH GENOME WIDE CNV ANALYSIS MUSCLE DISORDERS PANEL
SMDT1	100%	100%	100%	99.5%		MOVEMENT DISORDERS PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS MITOCHONDRIAL DISORDERS PANEL MUSCLE DISORDERS PANEL
SMG8	100%	100%	100%	99.2%	Alzahrani-Kuwahara syndrome, 619268	VISION DISORDERS PANEL INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹

SMG9	100%	100%	100%	98.5%	Heart and brain malformation syndrome, 616920;Neurodevelopmental disorder with intention tremor, pyramidal signs, dyspraxia, and ocular anomalies, 619995	INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹
SMN1	93.9%	93.9%	99.8%	94.5%	Spinal muscular atrophy-2, 253550;Spinal muscular atrophy-4, 271150;Spinal muscular atrophy-3, 253400;Spinal muscular atrophy-1, 253300	MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹

SMO	100%	100%	100%	99.1%	Pallister-Hall-like syndrome, 241800;Basal cell carcinoma, somatic, 605462;Curry-Jones syndrome, somatic mosaic, 601707	CRANIOFACIAL ANOMALIES PANEL WITH GENOME WIDE CNV ANALYSIS SHORT STATURE/SKELETAL DYSPLASIA PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹ SKIN DISORDERS PANEL ¹
SMOC1	100%	100%	100%	99.2%	Microphthalmia with limb anomalies, 206920	VISION DISORDERS PANEL INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹

SMOC2	100%	100%	100%	98.9%	Dentin dysplasia, type I, with microdontia and misshapen teeth, 125400	CRANIOFACIAL ANOMALIES PANEL WITH GENOME WIDE CNV ANALYSIS SKIN DISORDERS PANEL ¹ SHORT STATURE/SKELETAL DYSPLASIA PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹
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SMPD1	100%	100%	100%	98.5%	Niemann-Pick disease, type B, 607616;Niemann-Pick disease, type A, 257200	MOVEMENT DISORDERS PANEL LIVER DISORDERS PANEL METABOLIC DISORDERS PANEL INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹
SMPD4	100%	100%	100%	99.2%	Neurodevelopmental disorder with microcephaly, arthrogryposis, and structural brain anomalies, 618622	FETAL AKINESIA PANEL EPILEPSY PANEL INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹

SMPX	100%	100%	98.2%	65.3%	Myopathy, distal, 7, adult-onset, X-linked, 301075;Deafness, X-linked 4, 300066	HEARING IMPAIRMENT PANEL (INCLUDING GJB2) MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS
SMS	100%	97.8%	99%	68.9%	Intellectual developmental disorder, X-linked syndromic, Snyder-Robinson type, 309583	EPILEPSY PANEL METABOLIC DISORDERS PANEL INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS OROFACIAL CLEFTING PANEL WITH GENOME WIDE CNV ANALYSIS

SNAI2	100%	100%	100%	99.6%		SKIN DISORDERS PANEL ¹ HEARING IMPAIRMENT PANEL (INCLUDING GJB2) MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹ CRANIOFACIAL ANOMALIES PANEL WITH GENOME WIDE CNV ANALYSIS
SNAP25	100%	100%	100%	99.8%	?Myasthenic syndrome, congenital, 18, 616330	EPILEPSY PANEL INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS

SNAP29	100%	100%	100%	99.1%	Cerebral dysgenesis, neuropathy, ichthyosis, and palmoplantar keratoderma syndrome, 609528	SKIN DISORDERS PANEL ¹ INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹
SNAPC4	100%	100%	100%	99.1%	Neurodevelopmental disorder with motor regression, progressive spastic paraplegia, and oromotor dysfunction, 620515	INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹
SNCA	100%	100%	100%	99.4%	Dementia, Lewy body, 127750;Parkinson disease 1, 168601;Parkinson disease 4, 605543	MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS PARKINSON DISEASE PANEL

SNCB	100%	100%	100%	99%	Dementia, Lewy body, 127750	MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS
SNF8	100%	100%	100%	99%	Developmental and epileptic encephalopathy 115, 620783;Neurodevelopmental disorder plus optic atrophy, 620784	VISION DISORDERS PANEL INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹
SNIP1	100%	100%	100%	99.4%	Neurodevelopmental disorder with hypotonia, craniofacial abnormalities, and seizures, 614501	INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹

SNORA31					{Encephalopathy, acute, infection-induced (herpes-specific), susceptibility to, 10}, 619396	PRIMARY IMMUNODEFICIENCIES PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS
SNORD118					Leukoencephalopathy, brain calcifications, and cysts, 614561	MOVEMENT DISORDERS PANEL INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹
SNRNP200	100%	100%	100%	99.2%	Retinitis pigmentosa 33, 610359	VISION DISORDERS PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS

SNRPB	100%	100%	100%	99.1%	Cerebrocostomandibular syndrome, 117650	SHORT STATURE/SKELETAL DYSPLASIA PANEL WITH GENOME WIDE CNV ANALYSIS INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS OROFACIAL CLEFTING PANEL WITH GENOME WIDE CNV ANALYSIS
SNRPE	100%	100%	100%	99.9%	Hypotrichosis 11, 615059	SKIN DISORDERS PANEL ¹ MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS
SNRPN	100%	100%	100%	99.5%		INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS

SNTA1	100%	100%	100%	97.8%	Long QT syndrome 12, 612955	HEART DISORDERS PANEL ¹ MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS
SNUPN	100%	100%	100%	99.4%	Muscular dystrophy, limb-girdle, autosomal recessive 29, 620793	MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹ MUSCLE DISORDERS PANEL

SNX10	89.3%	89.3%	100%	99.7%	Osteopetrosis, autosomal recessive 8, 615085	VISION DISORDERS PANEL INHERITED BONE MARROW FAILURE AND/OR PREDISPOSITION PANEL TO HEMATOLOGICAL MALIGNANCIES SKIN DISORDERS PANEL ¹ PRIMARY IMMUNODEFICIENCIES PANEL SHORT STATURE/SKELETAL DYSPLASIA PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹
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SNX14	95%	95%	100%	99.7%	Spinocerebellar ataxia, autosomal recessive 20, 616354	MOVEMENT DISORDERS PANEL METABOLIC DISORDERS PANEL INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹
SNX27	100%	100%	100%	99.4%		INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS

SOBP	99.5%	98%	99.9%	94.3%	?Impaired intellectual development, anterior maxillary protrusion, and strabismus, 613671	INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹
SOCS1	100%	100%	100%	97.2%	Autoinflammatory syndrome, familial, with or without immunodeficiency, 619375	PRIMARY IMMUNODEFICIENCIES PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS
SOCS4	100%	100%	100%	99.7%		PRIMARY IMMUNODEFICIENCIES PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS

SOD1	100%	100%	100%	99.7%	Spastic tetraplegia and axial hypotonia, progressive, 618598;Amyotrophic lateral sclerosis 1, 105400	AMYOTROPHIC LATERAL SCLEROSIS, ALS PANEL METABOLIC DISORDERS PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹
SOD2	100%	100%	100%	98.5%	{Microvascular complications of diabetes 6}, 612634	HEART DISORDERS PANEL ¹ METABOLIC DISORDERS PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS MITOCHONDRIAL DISORDERS PANEL

SOHLH1	100%	100%	100%	98.6%	Ovarian dysgenesis 5, 617690;Spermatogenic failure 32, 618115	DISORDERS/DIFFERENCES OF SEX DEVELOPMENT (DSD) / PRIMARY ADRENAL INSUFFICIENCY PANEL MALE INFERTILITY PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS PREMATURE OVARIAN INSUFFICIENCY PANEL
SON	100%	100%	100%	99.4%	ZTTK syndrome, 617140	INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS OROFACIAL CLEFTING PANEL WITH GENOME WIDE CNV ANALYSIS

SORD	94.5%	89.7%	98.7%	90.1%	Neuronopathy, distal hereditary motor, autosomal recessive 8, 618912	POLYNEUROPATHIES PANEL ¹ MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹
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SOS1	98.8%	98.5%	100%	99.4%	Noonan syndrome 4, 610733;Fibromatosis, gingival, 1, 135300	INHERITED BONE MARROW FAILURE AND/OR PREDISPOSITION PANEL TO HEMATOLOGICAL MALIGNANCIES CONGENITAL HEARTDISEASE PANEL ¹ SKIN DISORDERS PANEL ¹ HEART DISORDERS PANEL ¹ HEMOSTATIC/THROMBOTIC DISORDERS PANEL SHORT STATURE/SKELETAL DYSPLASIA PANEL WITH GENOME WIDE CNV ANALYSIS LYMPHATIC ANOMALIES PANEL WITH GENOME WIDE CNV ANALYSIS INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS NOONAN SYNDROME / RASOPATHY PANEL
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						HEREDITARY CANCER PANEL
SOS2	100%	100%	100%	99%	Noonan syndrome 9, 616559	HEMOSTATIC/THROMB OTIC DISORDERS PANEL SHORT STATURE/SKELETAL DYSPLASIA PANEL WITH GENOME WIDE CNV ANALYSIS LYMPHATIC ANOMALIES PANEL WITH GENOME WIDE CNV ANALYSIS INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS NOONAN SYNDROME / RASOPATHY PANEL

SOST	100%	100%	100%	99.4%	Sclerosteosis 1, 269500;Craniodiaphyseal dysplasia, autosomal dominant, 122860	SHORT STATURE/SKELETAL DYSPLASIA PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹
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SOX10	97.8%	97.8%	100%	98.2%	Waardenburg syndrome, type 4C, 613266;PCWH syndrome, 609136;Waardenburg syndrome, type 2E, with or without neurologic involvement, 611584	MOVEMENT DISORDERS PANEL LIVER DISORDERS PANEL INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS DISORDERS/DIFFEREN CES OF SEX DEVELOPMENT (DSD) / PRIMARY ADRENAL INSUFFICIENCY PANEL HYPOGONADOTROPIC HYPOGONADISM (KALLMANN) PANEL POLYNEUROPATHIES PANEL ¹ CRANIOFACIAL ANOMALIES PANEL WITH GENOME WIDE CNV ANALYSIS SKIN DISORDERS PANEL ¹ HEARING IMPAIRMENT PANEL (INCLUDING GJB2)
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SOX11	100%	100%	100%	93.6%	Intellectual developmental disorder with microcephaly and with or without ocular malformations or hypogonadotropic hypogonadism, 615866	DISORDERS/DIFFERENCES OF SEX DEVELOPMENT (DSD) / PRIMARY ADRENAL INSUFFICIENCY PANEL HYPOGONADOTROPIC HYPOGONADISM (KALLMANN) PANEL INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS PREMATURE OVARIAN INSUFFICIENCY PANEL
SOX17	100%	100%	100%	98.9%	Vesicoureteral reflux 3, 613674	RENAL DISORDERS PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS

SOX18	99.2%	96.3%	100%	94.9%	Hypotrichosis-lymphedema-telangiectasia syndrome, 607823;Hypotrichosis-lymphedema-telangiectasia-renal defect syndrome, 137940	SKIN DISORDERS PANEL ¹ LYMPHATIC ANOMALIES PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹
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SOX2	100%	100%	99.9%	95.5%	Optic nerve hypoplasia and abnormalities of the central nervous system, 206900;Microphthalmia, syndromic 3, 206900	VISION DISORDERS PANEL SKIN DISORDERS PANEL ¹ DISORDERS/DIFFERENCES OF SEX DEVELOPMENT (DSD) / PRIMARY ADRENAL INSUFFICIENCY PANEL HYPOGONADOTROPIC HYPOGONADISM (KALLMANN) PANEL SHORT STATURE/SKELETAL DYSPLASIA PANEL WITH GENOME WIDE CNV ANALYSIS INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS
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SOX3	100%	99.8%	96.7%	57.3%	Intellectual developmental disorder, X-linked, with isolated growth hormone deficiency, 300123;Panhypopituitarism, X-linked, 312000	DISORDERS/DIFFERENCES OF SEX DEVELOPMENT (DSD) / PRIMARY ADRENAL INSUFFICIENCY PANEL SHORT STATURE/SKELETAL DYSPLASIA PANEL WITH GENOME WIDE CNV ANALYSIS INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS
SOX4	100%	100%	100%	97.2%	Coffin-Siris syndrome 10, 618506	INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS

SOX5	100%	100%	100%	99.1%	Lamb-Shaffer syndrome, 616803	VISION DISORDERS PANEL INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS
SOX6	100%	100%	100%	99.4%	Tolchin-Le Caignec syndrome, 618971	CRANIOFACIAL ANOMALIES PANEL WITH GENOME WIDE CNV ANALYSIS INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS
SOX7	100%	100%	100%	97.8%		CONGENITAL HEARTDISEASE PANEL ¹ HEART DISORDERS PANEL ¹ MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS

SOX9	100%	100%	100%	95.8%	Campomelic dysplasia with autosomal sex reversal, 114290;Acampomelic campomelic dysplasia, 114290;Campomelic dysplasia, 114290	CRANIOFACIAL ANOMALIES PANEL WITH GENOME WIDE CNV ANALYSIS DISORDERS/DIFFERENCES OF SEX DEVELOPMENT (DSD) / PRIMARY ADRENAL INSUFFICIENCY PANEL SHORT STATURE/SKELETAL DYSPLASIA PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS OROFACIAL CLEFTING PANEL WITH GENOME WIDE CNV ANALYSIS
SP110	100%	100%	100%	99.2%	{Mycobacterium tuberculosis, susceptibility to}, 607948;Hepatic venoocclusive disease with immunodeficiency, 235550	PRIMARY IMMUNODEFICIENCIES PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹

SP6	100%	100%	100%	99.2%	Amelogenesis imperfecta, type IK, 620104	MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS
SP7	100%	100%	100%	99%	Osteogenesis imperfecta, type XII, 613849	SKIN DISORDERS PANEL ¹ SHORT STATURE/SKELETAL DYSPLASIA PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹
SP9	100%	100%	100%	97%		INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS

SPACA1	100%	100%	100%	99.6%	?Spermatogenic failure 85, 620490	MALE INFERTILITY PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS
SPAG1	100%	99.9%	100%	99.5%	Ciliary dyskinesia, primary, 28, 615505	CILIOPATHIES PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹
SPAG17	100%	100%	100%	99.5%	?Spermatogenic failure 55, 619380	MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS
SPAG6	100%	100%	100%	99.4%		MALE INFERTILITY PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS

SPARC	100%	100%	100%	99.5%	Osteogenesis imperfecta, type XVII, 616507	SHORT STATURE/SKELETAL DYSPLASIA PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹
SPARCL1	100%	100%	100%	99.4%		VISION DISORDERS PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS

SPART	100%	100%	100%	99.4%	Troyer syndrome, 275900	MOVEMENT DISORDERS PANEL INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS MITOCHONDRIAL DISORDERS PANEL COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹
SPAST	100%	100%	100%	99.1%	Spastic paraplegia 4, autosomal dominant, 182601	MOVEMENT DISORDERS PANEL POLYNEUROPATHIES PANEL ¹ INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS

SPATA16	100%	100%	100%	99.7%	?Spermatogenic failure 6, 102530	MALE INFERTILITY PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS
SPATA22	100%	100%	100%	99.7%	Premature ovarian failure 25, 621002;Spermatogenic failure 96, 621001	DISORDERS/DIFFEREN CES OF SEX DEVELOPMENT (DSD) / PRIMARY ADRENAL INSUFFICIENCY PANEL MALE INFERTILITY PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS PREMATURE OVARIAN INSUFFICIENCY PANEL
SPATA7	100%	100%	100%	99.8%	Leber congenital amaurosis 3, 604232;Retinitis pigmentosa 94, variable age at onset, autosomal recessive, 604232	VISION DISORDERS PANEL CILIOPATHIES PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹

SPECC1L	100%	99.9%	100%	99.5%	Teebi hypertelorism syndrome 1, 145420;?Facial clefting, oblique, 1, 600251	CRANIOFACIAL ANOMALIES PANEL WITH GENOME WIDE CNV ANALYSIS SHORT STATURE/SKELETAL DYSPLASIA PANEL WITH GENOME WIDE CNV ANALYSIS INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS OROFACIAL CLEFTING PANEL WITH GENOME WIDE CNV ANALYSIS
SPEF2	100%	100%	100%	99.6%	Spermatogenic failure 43, 618751	MALE INFERTILITY PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS

SPEG	100%	100%	100%	98.3%	Centronuclear myopathy 5, 615959	MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹ MUSCLE DISORDERS PANEL
SPEN	100%	100%	100%	98.8%	Radio-Tartaglia syndrome, 619312	INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS

SPG11	99.6%	99.6%	100%	99.2%	Amyotrophic lateral sclerosis 5, juvenile, 602099;Charcot-Marie-Tooth disease, axonal, type 2X, 616668;Spastic paraplegia 11, autosomal recessive, 604360	AMYOTROPHIC LATERAL SCLEROSIS, ALS PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL¹ POLYNEUROPATHIES PANEL¹ INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MOVEMENT DISORDERS PANEL
SPG21	100%	100%	100%	99.8%	Mast syndrome, 248900	COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL¹ MOVEMENT DISORDERS PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS

SPG7	100%	100%	100%	98.7%	Spastic paraplegia 7, autosomal recessive, 607259	MOVEMENT DISORDERS PANEL VISION DISORDERS PANEL POLYNEUROPATHIES PANEL ¹ MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS MITOCHONDRIAL DISORDERS PANEL COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹
SPI1	100%	100%	100%	98.7%	Agammaglobulinemia 10, autosomal dominant, 619707	PRIMARY IMMUNODEFICIENCIES PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS

SPIDR	100%	100%	100%	99.2%	Ovarian dysgenesis 9, 619665	DISORDERS/DIFFERENCES OF SEX DEVELOPMENT (DSD) / PRIMARY ADRENAL INSUFFICIENCY PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS PREMATURE OVARIAN INSUFFICIENCY PANEL
SPIN4	100%	100%	98.8%	65.6%	?Lui-Jee-Baron syndrome, 301114	MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS
SPINK1	100%	100%	100%	99.3%	Tropical calcific pancreatitis, 608189;Pancreatitis, hereditary, 167800;{Fibrocalculous pancreatic diabetes, susceptibility to}, 608189	MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS HEREDITARY CANCER PANEL
SPINK2	100%	100%	100%	99.2%	?Spermatogenic failure 29, 618091	MALE INFERTILITY PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS

SPINK5	100%	100%	100%	99.5%	Netherton syndrome, 256500	SKIN DISORDERS PANEL ¹ PRIMARY IMMUNODEFICIENCIES PANEL SHORT STATURE/SKELETAL DYSPLASIA PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹
SPINT2	100%	100%	100%	99.1%	Diarrhea 3, secretory sodium, congenital, syndromic, 270420	SKIN DISORDERS PANEL ¹ LIVER DISORDERS PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹

SPNS2	99.4%	98.6%	100%	97.8%	?Deafness, autosomal recessive 115, 618457	HEARING IMPAIRMENT PANEL (INCLUDING GJB2) MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS
SPO11	100%	100%	100%	99.6%		MALE INFERTILITY PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS
SPOCK1	100%	100%	100%	98.5%		INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS
SPOP	100%	100%	100%	99.7%	Nabais Sa-de Vries syndrome, type 1, 618828;Nabais Sa-de Vries syndrome, type 2, 618829	INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS

SPOUT1	100%	100%	100%	99.4%		INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS
SPP2	100%	100%	100%	99.6%		VISION DISORDERS PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS
SPPL2A	100%	100%	100%	99.5%	Immunodeficiency 86, mycobacteriosis, 619549	PRIMARY IMMUNODEFICIENCIES PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS

SPR	100%	100%	100%	99.5%	Dystonia, dopa-responsive, due to sepiapterin reductase deficiency, 612716	MOVEMENT DISORDERS PANEL SHORT STATURE/SKELETAL DYSPLASIA PANEL WITH GENOME WIDE CNV ANALYSIS METABOLIC DISORDERS PANEL INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL¹
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SPRED1	100%	100%	100%	99.7%	Legius syndrome, 611431	SKIN DISORDERS PANEL ¹ SHORT STATURE/SKELETAL DYSPLASIA PANEL WITH GENOME WIDE CNV ANALYSIS LYMPHATIC ANOMALIES PANEL WITH GENOME WIDE CNV ANALYSIS INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS NOONAN SYNDROME / RASOPATHY PANEL HEREDITARY CANCER PANEL
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SPRED2	100%	100%	100%	98.9%	Noonan syndrome 14, 619745	SHORT STATURE/SKELETAL DYSPLASIA PANEL WITH GENOME WIDE CNV ANALYSIS LYMPHATIC ANOMALIES PANEL WITH GENOME WIDE CNV ANALYSIS INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL¹ NOONAN SYNDROME / RASOPATHY PANEL
SPRTN	100%	100%	100%	99.5%	Ruijs-Aalfs syndrome, 616200	MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL¹

SPRY4	100%	100%	100%	99%	Hypogonadotropic hypogonadism 17 with or without anosmia, 615266	SKIN DISORDERS PANEL ¹ DISORDERS/DIFFERENCES OF SEX DEVELOPMENT (DSD) / PRIMARY ADRENAL INSUFFICIENCY PANEL HYPOGONADOTROPIC HYPOGONADISM (KALLMANN) PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS
SPTA1	100%	100%	100%	99.4%	Spherocytosis, type 3, 270970;Elliptocytosis-2, 130600;Pyropoikilocytosis, 266140	MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹

SPTAN1	99.4%	98.8%	100%	99%	Developmental delay with or without epilepsy, 620540;Developmental and epileptic encephalopathy 5, 613477;Spastic paraplegia 91, autosomal dominant, with or without cerebellar ataxia, 620538;Neuronopathy, distal hereditary motor, autosomal dominant 11, 620528	MOVEMENT DISORDERS PANEL EPILEPSY PANEL POLYNEUROPATHIES PANEL ¹ INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS
SPTB	100%	100%	100%	99%	Anemia, neonatal hemolytic, fatal or near-fatal, 617948;Elliptocytosis-3, 617948;Spherocytosis, type 2, 616649	MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹
SPTBN1	100%	100%	100%	99.3%	Developmental delay, impaired speech, and behavioral abnormalities, 619475	INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS

SPTBN2	100%	100%	100%	98.8%	Spinocerebellar ataxia 5, 600224;Spinocerebellar ataxia, autosomal recessive 14, 615386	MOVEMENT DISORDERS PANEL INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹
SPTBN4	100%	100%	100%	97.7%	Neurodevelopmental disorder with hypotonia, neuropathy, and deafness, 617519	POLYNEUROPATHIES PANEL ¹ INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS MITOCHONDRIAL DISORDERS PANEL COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹

SPTLC1	88.7%	88.7%	100%	99.4%	Amyotrophic lateral sclerosis 27, juvenile, 620285;Neuropathy, hereditary sensory and autonomic, type IA, 162400	POLYNEUROPATHIES PANEL ¹ NEUROLOGICAL PAIN DISORDERS PANEL ¹ METABOLIC DISORDERS PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS
SPTLC2	100%	100%	100%	99.2%	Neuropathy, hereditary sensory and autonomic, type IC, 613640	POLYNEUROPATHIES PANEL ¹ NEUROLOGICAL PAIN DISORDERS PANEL ¹ METABOLIC DISORDERS PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS
SPTLC3	100%	100%	100%	99.6%		POLYNEUROPATHIES PANEL ¹ MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS

SPTSSA	100%	100%	100%	96.4%	Spastic paraplegia 90A, autosomal dominant, 620416;?Spastic paraplegia 90B, autosomal recessive, 620417	METABOLIC DISORDERS PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS
SQOR	100%	100%	100%	99.1%	Sulfide:quinone oxidoreductase deficiency, 619221	METABOLIC DISORDERS PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS MITOCHONDRIAL DISORDERS PANEL
SQSTM1	100%	100%	100%	98.9%	Neurodegeneration with ataxia, dystonia, and gaze palsy, childhood-onset, 617145;Frontotemporal dementia and/or amyotrophic lateral sclerosis 3, 616437;Myopathy, distal, with rimmed vacuoles, 617158;Paget disease of bone 3, 167250	AMYOTROPHIC LATERAL SCLEROSIS, ALS PANEL MOVEMENT DISORDERS PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS MITOCHONDRIAL DISORDERS PANEL COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹ HEREDITARY CANCER PANEL

SRC	96.3%	96.3%	100%	98.8%	?Thrombocytopenia 6, 616937;Colon cancer, advanced, somatic, 114500	HEMOSTATIC/THROMBOTIC DISORDERS PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS
SRCAP	100%	100%	100%	98.2%	Developmental delay, hypotonia, musculoskeletal defects, and behavioral abnormalities, 619595;Floating-Harbor syndrome, 136140	DISORDERS/DIFFERENCES OF SEX DEVELOPMENT (DSD) / PRIMARY ADRENAL INSUFFICIENCY PANEL SHORT STATURE/SKELETAL DYSPLASIA PANEL WITH GENOME WIDE CNV ANALYSIS INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS

SRD5A2	100%	100%	100%	99.6%	Pseudovaginal perineoscrotal hypospadias, 264600	DISORDERS/DIFFERENCES OF SEX DEVELOPMENT (DSD) / PRIMARY ADRENAL INSUFFICIENCY PANEL METABOLIC DISORDERS PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹
SRD5A3	100%	100%	100%	99.4%	Kahrizi syndrome, 612713;Congenital disorder of glycosylation, type Iq, 612379	SKIN DISORDERS PANEL ¹ METABOLIC DISORDERS PANEL INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹

SREBF1	100%	100%	99.9%	96.4%	Ichthyosis, follicular, with atrichia and photophobia syndrome 2, 619016;Mucoepithelial dysplasia, hereditary, 158310	SKIN DISORDERS PANEL ¹ MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS
SRF	100%	99.9%	100%	97.2%		CONGENITAL HEARTDISEASE PANEL ¹ HEART DISORDERS PANEL ¹ MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS
SRI	100%	100%	100%	99.7%		HEART DISORDERS PANEL ¹ MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS

SRP54	100%	100%	100%	99.8%	Neutropenia, severe congenital, 8, autosomal dominant, 618752	INHERITED BONE MARROW FAILURE AND/OR PREDISPOSITION PANEL TO HEMATOLOGICAL MALIGNANCIES PRIMARY IMMUNODEFICIENCIES PANEL SHORT STATURE/SKELETAL DYSPLASIA PANEL WITH GENOME WIDE CNV ANALYSIS INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS
SRP68	100%	99.8%	100%	99.5%	?Neutropenia, severe congenital, 10, autosomal recessive, 620534	MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS

SRP72	100%	100%	100%	99.6%	Bone marrow failure syndrome 1, 614675	INHERITED BONE MARROW FAILURE AND/OR PREDISPOSITION PANEL TO HEMATOLOGICAL MALIGNANCIES PRIMARY IMMUNODEFICIENCIES PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS
SRPK3	100%	99.3%	98.4%	66%	Intellectual developmental disorder, X-linked 114, 301134	FETAL AKINESIA PANEL INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS MUSCLE DISORDERS PANEL

SRPX2	100%	99.6%	97.5%	67.1%	?Rolandic epilepsy, impaired intellectual development, and speech dyspraxia, 300643	INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS
SRRM2	100%	100%	100%	98.5%	Intellectual developmental disorder, autosomal dominant 72, 620439	INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS
SRSF1	100%	100%	100%	99.4%	Neurodevelopmental disorder with dysmorphic facies and behavioral abnormalities, 620489	INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS

SRY	50%	49.9%	49.3%	19.3%	46XY sex reversal 1, 400044;46XX sex reversal 1, 400045	DISORDERS/DIFFERENCES OF SEX DEVELOPMENT (DSD) / PRIMARY ADRENAL INSUFFICIENCY PANEL MALE INFERTILITY PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS
SSBP1	100%	100%	100%	99%	Optic atrophy 13 with retinal and foveal abnormalities, 165510	VISION DISORDERS PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS MITOCHONDRIAL DISORDERS PANEL
SSR3	100%	100%	100%	99.5%		METABOLIC DISORDERS PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS

SSR4	100%	99.4%	98.5%	65.5%	Congenital disorder of glycosylation, type ly, 300934	METABOLIC DISORDERS PANEL INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS
SSTR5	100%	100%	99.9%	96.1%		MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS
SSX1	100%	100%	99.3%	71.9%	Spermatogenic failure, X-linked, 5, 301099	MALE INFERTILITY PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS
SSX2	100%	100%	99.9%	80.5%	?Sarcoma, synovial, 300813	MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS

ST14	100%	100%	100%	98.9%	Ichthyosis, congenital, autosomal recessive 11, 602400	SKIN DISORDERS PANEL ¹ MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹
ST3GAL3	95.9%	95%	100%	98.6%	Developmental and epileptic encephalopathy 15, 615006;Intellectual developmental disorder, autosomal recessive 12, 611090	EPILEPSY PANEL COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹ METABOLIC DISORDERS PANEL INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS

ST3GAL5	98.3%	98.3%	100%	99.4%	Salt and pepper developmental regression syndrome, 609056	SKIN DISORDERS PANEL ¹ EPILEPSY PANEL COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹ METABOLIC DISORDERS PANEL INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS
STAB2	100%	100%	100%	99.4%		HEMOSTATIC/THROMB OTIC DISORDERS PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS

STAC3	100%	100%	100%	99.2%	Congenital myopathy 13, 255995	FETAL AKINESIA PANEL MUSCLE DISORDERS PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS MITOCHONDRIAL DISORDERS PANEL COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL¹ OROFACIAL CLEFTING PANEL WITH GENOME WIDE CNV ANALYSIS
STAG1	100%	100%	100%	99.5%	Intellectual developmental disorder, autosomal dominant 47, 617635	INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS

STAG2	100%	99.9%	99%	70.6%	Holoprosencephaly 13, X-linked, 301043;Mullegama-Klein-Martinez syndrome, 301022	HEARING IMPAIRMENT PANEL (INCLUDING GJB2) INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS
STAG3	100%	100%	100%	98.7%	Spermatogenic failure 61, 619672;Premature ovarian failure 8, 615723	MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS PREMATURE OVARIAN INSUFFICIENCY PANEL DISORDERS/DIFFERENCES OF SEX DEVELOPMENT (DSD) / PRIMARY ADRENAL INSUFFICIENCY PANEL MALE INFERTILITY PANEL

STAMBP	96.3%	96.2%	100%	99.4%	Microcephaly-capillary malformation syndrome, 614261	MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹ SKIN DISORDERS PANEL ¹ INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS OROFACIAL CLEFTING PANEL WITH GENOME WIDE CNV ANALYSIS
STAR	100%	100%	100%	99.3%	Lipoid adrenal hyperplasia, 201710	DISORDERS/DIFFEREN CES OF SEX DEVELOPMENT (DSD) / PRIMARY ADRENAL INSUFFICIENCY PANEL METABOLIC DISORDERS PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹

STARD7	100%	100%	100%	99%	Epilepsy, familial adult myoclonic, 2, 607876	MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS
STAT1	95.9%	95.9%	100%	99.5%	Immunodeficiency 31C, chronic mucocutaneous candidiasis, autosomal dominant, 614162;Immunodeficiency 31A, mycobacteriosis, autosomal dominant, 614892;Immunodeficiency 31B, mycobacterial and viral infections, autosomal recessive, 613796	PRIMARY IMMUNODEFICIENCIES PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹
STAT2	100%	100%	100%	99.2%	Pseudo-TORCH syndrome 3, 618886;Immunodeficiency 44, 616636	PRIMARY IMMUNODEFICIENCIES PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS MITOCHONDRIAL DISORDERS PANEL COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹

STAT3	100%	100%	100%	99.3%	Hyper-IgE syndrome 1, autosomal dominant, with recurrent infections, 147060;Autoimmune disease, multisystem, infantile-onset, 1, 615952	SKIN DISORDERS PANEL ¹ PRIMARY IMMUNODEFICIENCIES PANEL SHORT STATURE/SKELETAL DYSPLASIA PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS
STAT4	100%	100%	100%	99.8%	Disabling pansclerotic morphea of childhood, 620443;{Systemic lupus erythematosus, susceptibility to, 11}, 612253	PRIMARY IMMUNODEFICIENCIES PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS

STAT5B	100%	100%	100%	98.9%	Growth hormone insensitivity with immune dysregulation 1, autosomal recessive, 245590;Growth hormone insensitivity with immune dysregulation 2, autosomal dominant, 618985	SKIN DISORDERS PANEL ¹ PRIMARY IMMUNODEFICIENCIES PANEL SHORT STATURE/SKELETAL DYSPLASIA PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹
STAT6	100%	100%	100%	98.6%	Hyper-IgE syndrome 6, autosomal dominant, with recurrent infections, 620532	PRIMARY IMMUNODEFICIENCIES PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS
STEAP3	100%	99.9%	100%	99.6%	?Anemia, hypochromic microcytic, with iron overload 2, 615234	IRON DISORDERS PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS

STEEP1	100%	98.3%	98.9%	65.9%	?Intellectual developmental disorder, X-linked 107, 301013	INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS
STIL	100%	100%	100%	99.5%	Microcephaly 7, primary, autosomal recessive, 612703	INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹

STIM1	100%	99.7%	100%	99.5%	Myopathy, tubular aggregate, 1, 160565;Stormorken syndrome, 185070;Immunodeficiency 10, 612783	INHERITED BONE MARROW FAILURE AND/OR PREDISPOSITION PANEL TO HEMATOLOGICAL MALIGNANCIES SKIN DISORDERS PANEL ¹ HEMOSTATIC/THROMBOTIC DISORDERS PANEL PRIMARY IMMUNODEFICIENCIES PANEL SHORT STATURE/SKELETAL DYSPLASIA PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹ MUSCLE DISORDERS PANEL
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STING1	100%	100%	100%	99.4%	STING-associated vasculopathy, infantile-onset, 615934	SKIN DISORDERS PANEL ¹ PRIMARY IMMUNODEFICIENCIES PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS
STK11	100%	100%	100%	99.2%	Melanoma, malignant, somatic, 155600;Pancreatic cancer, somatic, 260350;Peutz-Jeghers syndrome, 175200;Testicular tumor, somatic, 273300	SKIN DISORDERS PANEL ¹ MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS PANEL HEREDITARY PANCREATIC CANCER (BRCA1, BRCA2, PALB2, ATM, CDKN2A, MLH1, MSH2, MSH6, PMS2, STK11) HEREDITARY CANCER PANEL PANEL HEREDITARY COLORECTAL AND POLYPOSIS
STK33	100%	100%	100%	99.5%	?Spermatogenic failure 93, 620849	MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS

STK36	100%	100%	100%	99.3%	?Ciliary dyskinesia, primary, 46, 619436	CILIOPATHIES PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS
STK4	100%	100%	100%	99.6%	T-cell immunodeficiency, recurrent infections, autoimmunity, and cardiac malformations, 614868	SKIN DISORDERS PANEL ¹ PRIMARY IMMUNODEFICIENCIES PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹ SEVERE COMBINED IMMUNODEFICIENCY (SCID) PANEL

STN1	87.1%	87%	100%	99.4%	Cerebroretinal microangiopathy with calcifications and cysts 2, 617341	INHERITED BONE MARROW FAILURE AND/OR PREDISPOSITION PANEL TO HEMATOLOGICAL MALIGNANCIES LIVER DISORDERS PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹
STOX1	98.2%	96%	93.7%	86%	Preeclampsia/eclampsia 4, 609404	MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS

STRA6	100%	100%	100%	99%	Microphthalmia, syndromic 9, 601186;Microphthalmia, isolated, with coloboma 8, 601186	VISION DISORDERS PANEL INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS RENAL DISORDERS PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹
STRA8	100%	100%	100%	99.1%		MALE INFERTILITY PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS

STRADA	100%	99.8%	100%	98.5%	Polyhydramnios, megalencephaly, and symptomatic epilepsy, 611087	EPILEPSY PANEL INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹
STRC	100%	100%	100%	99.4%	Deafness, autosomal recessive 16, 603720	HEARING IMPAIRMENT PANEL (INCLUDING GJB2) MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹
STS	97.1%	97%	98%	67.9%	Ichthyosis, X-linked, 308100	VISION DISORDERS PANEL SKIN DISORDERS PANEL ¹ METABOLIC DISORDERS PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS

STT3A	100%	100%	100%	99.6%	Congenital disorder of glycosylation, type lw, autosomal dominant, 619714;Congenital disorder of glycosylation, type lw, autosomal recessive, 615596	METABOLIC DISORDERS PANEL INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹
STT3B	100%	100%	100%	98.8%	Congenital disorder of glycosylation, type lx, 615597	METABOLIC DISORDERS PANEL INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹

STUB1	100%	100%	100%	98.7%	Spinocerebellar ataxia 48, 618093;Spinocerebellar ataxia, autosomal recessive 16, 615768	MOVEMENT DISORDERS PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹
STX11	100%	100%	100%	98.3%	Hemophagocytic lymphohistiocytosis, familial, 4, 603552	PRIMARY IMMUNODEFICIENCIES PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹
STX16	100%	100%	100%	99.2%	Pseudohypoparathyroidism Ib, 603233	RENAL DISORDERS PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS

STX1A	100%	100%	100%	98.6%		INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS
STX1B	100%	100%	100%	94.9%	Generalized epilepsy with febrile seizures plus, type 9, 616172	EPILEPSY PANEL INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS
STX2	100%	100%	100%	99.4%		MALE INFERTILITY PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS

STX3	100%	100%	100%	99.5%	Retinal dystrophy and microvillus inclusion disease, 619446;Diarrhea 12, with microvillus atrophy, 619445	VISION DISORDERS PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹
STX4	100%	100%	100%	99.4%	?Deafness, autosomal recessive 123, 620745	HEARING IMPAIRMENT PANEL (INCLUDING GJB2) MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS
STX5	100%	100%	100%	99.5%	?Congenital disorder of glycosylation, type Ilaa, 620454	METABOLIC DISORDERS PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS

STXBP1	100%	100%	100%	98.8%	Developmental and epileptic encephalopathy 4, 612164	EPILEPSY PANEL INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS MITOCHONDRIAL DISORDERS PANEL COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹
STXBP2	100%	99.9%	100%	98.2%	Hemophagocytic lymphohistiocytosis, familial, 5, with or without microvillus inclusion disease, 613101	HEMOSTATIC/THROMB OTIC DISORDERS PANEL PRIMARY IMMUNODEFICIENCIES PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹

SUCLA2	100%	99.7%	100%	99.1%	Mitochondrial DNA depletion syndrome 5 (encephalomyopathic with or without methylmalonic aciduria), 612073	HEARING IMPAIRMENT PANEL (INCLUDING GJB2) METABOLIC DISORDERS PANEL INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS MITOCHONDRIAL DISORDERS PANEL COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL¹ MUSCLE DISORDERS PANEL
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SUCLG1	100%	100%	100%	98.9%	Mitochondrial DNA depletion syndrome 9 (encephalomyopathic type with methylmalonic aciduria), 245400	METABOLIC DISORDERS PANEL INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS MITOCHONDRIAL DISORDERS PANEL COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹
SUCLG2	100%	99.9%	100%	99.9%		METABOLIC DISORDERS PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS MITOCHONDRIAL DISORDERS PANEL HEREDITARY CANCER PANEL

SUFU	100%	100%	100%	99.2%	{Meningioma, familial, susceptibility to}, 607174;Joubert syndrome 32, 617757;Basal cell nevus syndrome 2, 620343;{Medulloblastoma}, 155255	SKIN DISORDERS PANEL ¹ INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹ SONIC HEDGEHOG MEDULLOBLASTOMA PANEL HEREDITARY CANCER PANEL
SUGCT	100%	100%	100%	99.6%	Glutaric aciduria III, 231690	METABOLIC DISORDERS PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS

SULF1	100%	100%	100%	99.6%		SHORT STATURE/SKELETAL DYSPLASIA PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS
SULT2B1	100%	100%	100%	97.9%	Ichthyosis, congenital, autosomal recessive 14, 617571	SKIN DISORDERS PANEL ¹ MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹

SUMF1	100%	100%	100%	99.1%	Multiple sulfatase deficiency, 272200	MOVEMENT DISORDERS PANEL VISION DISORDERS PANEL SKIN DISORDERS PANEL ¹ SHORT STATURE/SKELETAL DYSPLASIA PANEL WITH GENOME WIDE CNV ANALYSIS METABOLIC DISORDERS PANEL INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹
SUMO1	71%	71%	100%	99.9%	?Orofacial cleft 10, 613705	CRANIOFACIAL ANOMALIES PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS

SUN5	100%	100%	100%	99%	Spermatogenic failure 16, 617187	MALE INFERTILITY PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS
SUOX	100%	100%	100%	98.9%	Sulfite oxidase deficiency, 272300	MOVEMENT DISORDERS PANEL EPILEPSY PANEL METABOLIC DISORDERS PANEL INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹
SUPT16H	100%	100%	100%	99.4%	Neurodevelopmental disorder with dysmorphic facies and thin corpus callosum, 619480	INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS

SUPT7L	100%	100%	100%	99.6%		MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹
SUPV3L1	100%	100%	100%	99.5%		MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS MITOCHONDRIAL DISORDERS PANEL
SURF1	100%	100%	100%	98%	Charcot-Marie-Tooth disease, type 4K, 616684;Mitochondrial complex IV deficiency, nuclear type 1, 220110	HEART DISORDERS PANEL ¹ POLYNEUROPATHIES PANEL ¹ INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS MITOCHONDRIAL DISORDERS PANEL COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹

SUZ12	100%	100%	100%	99.1%	Imagawa-Matsumoto syndrome, 618786	TALL STATURE PANEL WITH GENOME WIDE CNV ANALYSIS INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS
SV2A	100%	100%	100%	98.3%	Developmental and epileptic encephalopathy 113, 620772	MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹
SVBP	100%	100%	100%	99.2%	Neurodevelopmental disorder with ataxia, hypotonia, and microcephaly, 618569	INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹

SVIL	100%	100%	100%	99.3%	Myofibrillar myopathy 10, 619040	HEART DISORDERS PANEL ¹ MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS
SYCE1	100%	100%	100%	98.6%	?Spermatogenic failure 15, 616950;?Premature ovarian failure 12, 616947	DISORDERS/DIFFEREN CES OF SEX DEVELOPMENT (DSD) / PRIMARY ADRENAL INSUFFICIENCY PANEL MALE INFERTILITY PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS PREMATURE OVARIAN INSUFFICIENCY PANEL
SYCP2	100%	100%	100%	99.6%	Spermatogenic failure 1, 258150	MALE INFERTILITY PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS

SYCP2L	100%	100%	100%	99.4%	Premature ovarian failure 24, 620840	PREMATURE OVARIAN INSUFFICIENCY PANEL DISORDERS/DIFFEREN CES OF SEX DEVELOPMENT (DSD) / PRIMARY ADRENAL INSUFFICIENCY PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS
SYCP3	100%	100%	100%	99%	Pregnancy loss, recurrent, 4, 270960;Spermatogenic failure 4, 270960	MALE INFERTILITY PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS
SYK	100%	100%	100%	99.7%	Immunodeficiency 82 with systemic inflammation, 619381	PRIMARY IMMUNODEFICIENCIES PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS

SYN1	100%	99.9%	97.6%	64.4%	Epilepsy, X-linked 1, with variable learning disabilities and behavior disorders, 300491;Intellectual developmental disorder, X-linked 50, 300115	EPILEPSY PANEL INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS
SYNCRIP	92.9%	92.9%	100%	99.2%		INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS
SYNE1	100%	100%	100%	99.5%	Arthrogryposis multiplex congenita 3, myogenic type, 618484;Emery-Dreifuss muscular dystrophy 4, autosomal dominant, 612998;Spinocerebellar ataxia, autosomal recessive 8, 610743	FETAL AKINESIA PANEL MOVEMENT DISORDERS PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL¹

SYNE2	100%	100%	100%	99.5%	Emery-Dreifuss muscular dystrophy 5, autosomal dominant, 612999	MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS
SYNE4	100%	99.9%	100%	99.3%	Deafness, autosomal recessive 76, 615540	HEARING IMPAIRMENT PANEL (INCLUDING GJB2) MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹
SYNGAP1	100%	100%	100%	97.4%	Intellectual developmental disorder, autosomal dominant 5, 612621	EPILEPSY PANEL INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS

SYNJ1	100%	100%	100%	99.5%	Parkinson disease 20, early-onset, 615530;Developmental and epileptic encephalopathy 53, 617389	EPILEPSY PANEL INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹
SYP	100%	99.7%	97.5%	64.1%	Intellectual developmental disorder, X-linked 96, 300802	EPILEPSY PANEL INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS
SYT1	96%	96%	100%	99.5%	Baker-Gordon syndrome, 618218	INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS

SYT14	100%	100%	100%	99.6%	?Spinocerebellar ataxia, autosomal recessive 11, 614229	MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹
SYT2	100%	100%	100%	99%	Myasthenic syndrome, congenital, 7A, presynaptic, and distal motor neuropathy, autosomal dominant, 616040;Myasthenic syndrome, congenital, 7B, presynaptic, autosomal recessive, 619461	POLYNEUROPATHIES PANEL ¹ MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹ MUSCLE DISORDERS PANEL
SZT2	100%	100%	100%	99.2%	Developmental and epileptic encephalopathy 18, 615476	EPILEPSY PANEL INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS MITOCHONDRIAL DISORDERS PANEL COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹

TAB2	100%	100%	100%	98.4%	Congenital heart defects, nonsyndromic, 2, 614980	CONGENITAL HEARTDISEASE PANEL¹ HEART DISORDERS PANEL¹ SHORT STATURE/SKELETAL DYSPLASIA PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS
TAC3	100%	100%	100%	98.5%	Hypogonadotropic hypogonadism 10 with or without anosmia, 614839	DISORDERS/DIFFEREN CES OF SEX DEVELOPMENT (DSD) / PRIMARY ADRENAL INSUFFICIENCY PANEL HYPOGONADOTROPIC HYPOGONADISM (KALLMANN) PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL¹

TACO1	100%	100%	100%	99.2%	Mitochondrial complex IV deficiency, nuclear type 8, 619052	INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS MITOCHONDRIAL DISORDERS PANEL COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL¹
TACR3	100%	100%	100%	99.4%	Hypogonadotropic hypogonadism 11 with or without anosmia, 614840	DISORDERS/DIFFEREN CES OF SEX DEVELOPMENT (DSD) / PRIMARY ADRENAL INSUFFICIENCY PANEL HYPOGONADOTROPIC HYPOGONADISM (KALLMANN) PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL¹

TACSTD2	100%	100%	100%	98.9%	Corneal dystrophy, gelatinous drop-like, 204870	VISION DISORDERS PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹
TAF1	98.7%	98.5%	98.4%	67.2%	Intellectual developmental disorder, X-linked syndromic 33, 300966;Dystonia-Parkinsonism, X-linked, 314250	CONGENITAL HEARTDISEASE PANEL ¹ HEART DISORDERS PANEL ¹ INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS PARKINSON DISEASE PANEL

TAF13	89.5%	78.1%	100%	98.6%	Intellectual developmental disorder, autosomal recessive 60, 617432	INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹
TAF1A	100%	100%	100%	99.7%		HEART DISORDERS PANEL ¹ MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS
TAF1C	100%	100%	100%	99%		INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹

TAF2	96.3%	96.3%	100%	99.5%	Intellectual developmental disorder, autosomal recessive 40, 615599	INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹
TAF4	85%	79.9%	99.2%	83.4%	Intellectual developmental disorder, autosomal dominant 73, 620450	INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS
TAF4B	100%	100%	100%	99.4%	?Spermatogenic failure 13, 615841	MALE INFERTILITY PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS

TAF6	100%	100%	100%	98.4%	Alazami-Yuan syndrome, 617126	INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹
TAF8	85.3%	85.3%	100%	98.6%	Neurodevelopmental disorder with severe motor impairment, absent language, cerebral hypomyelination, and brain atrophy, 619972	INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹

TAFAZZIN	100%	99.8%	98.3%	64.5%	Barth syndrome, 302060	INHERITED BONE MARROW FAILURE AND/OR PREDISPOSITION PANEL TO HEMATOLOGICAL MALIGNANCIES HEART DISORDERS PANEL ¹ PRIMARY IMMUNODEFICIENCIES PANEL METABOLIC DISORDERS PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS MITOCHONDRIAL DISORDERS PANEL
TAL1	100%	100%	100%	97.7%	Leukemia, T-cell acute lymphocytic, somatic, 613065	MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS
TAL2	100%	100%	100%	99.6%	Leukemia, T-cell acute lymphocytic, somatic, 613065	MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS

TALDO1	100%	100%	100%	98.8%	Transaldolase deficiency, 606003	SKIN DISORDERS PANEL ¹ HEMOSTATIC/THROMB OTIC DISORDERS PANEL LIVER DISORDERS PANEL METABOLIC DISORDERS PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹
TAMM41	100%	100%	100%	99.2%	Combined oxidative phosphorylation deficiency 56, 620139	MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS MITOCHONDRIAL DISORDERS PANEL COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹

TANC2	100%	100%	100%	99.3%	Intellectual developmental disorder with autistic features and language delay, with or without seizures, 618906	INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS
TANGO2	100%	100%	100%	99.6%	Metabolic encephalomyopathic crises, recurrent, with rhabdomyolysis, cardiac arrhythmias, and neurodegeneration, 616878	MOVEMENT DISORDERS PANEL EPILEPSY PANEL MUSCLE DISORDERS PANEL METABOLIC DISORDERS PANEL INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS MITOCHONDRIAL DISORDERS PANEL COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹

TAOK1	100%	100%	100%	99.3%	Developmental delay with or without intellectual impairment or behavioral abnormalities, 619575	INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS MITOCHONDRIAL DISORDERS PANEL
TAOK2	100%	100%	100%	98.9%		INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS

TAP1	99.5%	97.2%	100%	98.9%	MHC class I deficiency 1, 604571	SKIN DISORDERS PANEL ¹ PRIMARY IMMUNODEFICIENCIES PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹ SEVERE COMBINED IMMUNODEFICIENCY (SCID) PANEL
TAP2	98%	97.9%	100%	98.2%	MHC class I deficiency 2, 620813	SKIN DISORDERS PANEL ¹ PRIMARY IMMUNODEFICIENCIES PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹ SEVERE COMBINED IMMUNODEFICIENCY (SCID) PANEL

TAPBP	88.8%	88.8%	100%	96.5%	?MHC class I deficiency 3, 620814	SKIN DISORDERS PANEL ¹ PRIMARY IMMUNODEFICIENCIES PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹ SEVERE COMBINED IMMUNODEFICIENCY (SCID) PANEL
TAPT1	100%	99.6%	100%	99.6%	Osteochondrodysplasia, complex lethal, Symoens-Barnes-Gistelinck type, 616897	SHORT STATURE/SKELETAL DYSPLASIA PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹ OROFACIAL CLEFTING PANEL WITH GENOME WIDE CNV ANALYSIS

TARDBP	100%	99.7%	100%	99.2%	Frontotemporal lobar degeneration, TARDBP-related, 612069;Amyotrophic lateral sclerosis 10, with or without FTD, 612069	AMYOTROPHIC LATERAL SCLEROSIS, ALS PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS
TARS1	100%	100%	100%	99.4%	Trichothiodystrophy 7, nonphotosensitive, 618546	SKIN DISORDERS PANEL ¹ MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS
TARS2	100%	100%	100%	99.2%	Combined oxidative phosphorylation deficiency 21, 615918	INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS MITOCHONDRIAL DISORDERS PANEL COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹

TASP1	100%	100%	100%	99.3%	Suleiman-El-Hattab syndrome, 618950	INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹
TAT	100%	100%	100%	99.5%	Tyrosinemia, type II, 276600	SKIN DISORDERS PANEL ¹ METABOLIC DISORDERS PANEL INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹
TAX1BP3	100%	100%	100%	100%		HEART DISORDERS PANEL ¹ MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS

TBC1D20	92.2%	92.2%	100%	98.9%	Warburg micro syndrome 4, 615663	MOVEMENT DISORDERS PANEL INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹
TBC1D23	100%	100%	100%	99.6%	Pontocerebellar hypoplasia, type 11, 617695	MOVEMENT DISORDERS PANEL EPILEPSY PANEL INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹

TBC1D24	100%	100%	100%	98.6%	Deafness, autosomal recessive 86, 614617;Epilepsy, rolandic, with paroxysmal exercise-induce dystonia and writer's cramp, 608105;Myoclonic epilepsy, infantile, familial, 605021;Deafness, autosomal dominant 65, 616044;Developmental and epileptic encephalopathy 16, 615338;DOORS syndrome, 220500	SKIN DISORDERS PANEL ¹ HEARING IMPAIRMENT PANEL (INCLUDING GJB2) EPILEPSY PANEL INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹
TBC1D2B	99.5%	98.8%	100%	99.2%	Neurodevelopmental disorder with seizures and gingival overgrowth, 619323	VISION DISORDERS PANEL INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS

TBC1D32	100%	100%	100%	99.6%		VISION DISORDERS PANEL CILIOPATHIES PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS
TBC1D7	100%	100%	100%	99.6%	Macrocephaly/megalencephaly syndrome, autosomal recessive, 248000	INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹
TBC1D8B	100%	99.9%	99.1%	70.4%	Nephrotic syndrome, type 20, 301028	RENAL DISORDERS PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS

TBCD	91.2%	90.6%	100%	98.5%	Encephalopathy, progressive, early-onset, with brain atrophy and thin corpus callosum, 617193	FETAL AKINESIA PANEL MOVEMENT DISORDERS PANEL EPILEPSY PANEL INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL¹
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TBCE	100%	100%	100%	99.1%	Kenny-Caffey syndrome, type 1, 244460;Hypoparathyroidism-retardation-dysmorphism syndrome, 241410;Encephalopathy, progressive, with amyotrophy and optic atrophy, 617207	EPILEPSY PANEL POLYNEUROPATHIES PANEL ¹ SHORT STATURE/SKELETAL DYSPLASIA PANEL WITH GENOME WIDE CNV ANALYSIS INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹
TBCK	100%	100%	100%	99.2%	Hypotonia, infantile, with psychomotor retardation and characteristic facies 3, 616900	INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS MITOCHONDRIAL DISORDERS PANEL COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹

TBK1	100%	100%	100%	99.6%	{Encephalopathy, acute, infection-induced (herpes-specific), susceptibility to, 8}, 617900;Frontotemporal dementia and/or amyotrophic lateral sclerosis 4, 616439;Autoinflammation with arthritis and vasculitis, 620880	AMYOTROPHIC LATERAL SCLEROSIS, ALS PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹
TBL1X	100%	99.9%	98%	66.3%	Hypothyroidism, congenital, nongoitrous, 8, 301033	HEARING IMPAIRMENT PANEL (INCLUDING GJB2) MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS
TBL1XR1	100%	100%	100%	99.6%	Intellectual developmental disorder, autosomal dominant 41, 616944;Pierpont syndrome, 602342	INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS

TBL1Y	50%	49.4%	48%	15.7%	?Deafness, Y-linked 2, 400047	HEARING IMPAIRMENT PANEL (INCLUDING GJB2) MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS
TBP	100%	100%	100%	98.8%	Spinocerebellar ataxia 17, 607136;{Parkinson disease, susceptibility to}, 168600	INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS
TBPL2	100%	100%	100%	99.3%		UNSUCCESSFUL IVF TREATMENTS, RECURRENT MOLAR PREGNANCIES AND/OR RECURRENT PREGNANCY LOSS PANEL ¹ MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS

TBR1	100%	100%	100%	98.2%	Intellectual developmental disorder with autism and speech delay, 606053	INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS
TBX1	96%	91.6%	99.8%	93.2%	Tetralogy of Fallot, 187500;DiGeorge syndrome, 188400;Conotruncal anomaly face syndrome, 217095;Velocardiofacial syndrome, 192430	CRANIOFACIAL ANOMALIES PANEL WITH GENOME WIDE CNV ANALYSIS CONGENITAL HEARTDISEASE PANEL ¹ HEART DISORDERS PANEL ¹ HEMOSTATIC/THROMB OTIC DISORDERS PANEL PRIMARY IMMUNODEFICIENCIES PANEL INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS OROFACIAL CLEFTING PANEL WITH GENOME WIDE CNV ANALYSIS

TBX15	100%	100%	100%	98.9%	Cousin syndrome, 260660	SHORT STATURE/SKELETAL DYSPLASIA PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹ OROFACIAL CLEFTING PANEL WITH GENOME WIDE CNV ANALYSIS
TBX18	100%	100%	100%	98.4%	Congenital anomalies of kidney and urinary tract 2, 143400	RENAL DISORDERS PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS

TBX19	100%	100%	100%	99.1%	Adrenocorticotrophic hormone deficiency, 201400	DISORDERS/DIFFERENCES OF SEX DEVELOPMENT (DSD) / PRIMARY ADRENAL INSUFFICIENCY PANEL LIVER DISORDERS PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹
TBX2	99.6%	95.8%	100%	98%	Vertebral anomalies and variable endocrine and T-cell dysfunction, 618223	SHORT STATURE/SKELETAL DYSPLASIA PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS OROFACIAL CLEFTING PANEL WITH GENOME WIDE CNV ANALYSIS

TBX20	100%	100%	100%	99.2%	Atrial septal defect 4, 611363	THORACIC AORTIC ANEURYSM AND/OR DISSECTION PANEL CONGENITAL HEARTDISEASE PANEL ¹ HEART DISORDERS PANEL ¹ MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS
TBX21	100%	100%	100%	98.1%	Asthma and nasal polyps, 208550;?Immunodeficiency 88, 619630;{Asthma, aspirin-induced, susceptibility to}, 208550	PRIMARY IMMUNODEFICIENCIES PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS
TBX22	100%	99.9%	99%	68.1%	Cleft palate with ankyloglossia, 303400;?Abruzzo-Erickson syndrome, 302905	CRANIOFACIAL ANOMALIES PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS OROFACIAL CLEFTING PANEL WITH GENOME WIDE CNV ANALYSIS

TBX3	100%	100%	100%	98.1%	Ulnar-mammary syndrome, 181450	SKIN DISORDERS PANEL ¹ DISORDERS/DIFFERENCES OF SEX DEVELOPMENT (DSD) / PRIMARY ADRENAL INSUFFICIENCY PANEL SHORT STATURE/SKELETAL DYSPLASIA PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS
TBX4	100%	100%	100%	98.9%	Ischiocoxopodopatellar syndrome with or without pulmonary arterial hypertension, 147891;Amelia, posterior, with pelvic and pulmonary hypoplasia syndrome, 601360	SHORT STATURE/SKELETAL DYSPLASIA PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS OROFACIAL CLEFTING PANEL WITH GENOME WIDE CNV ANALYSIS

TBX5	100%	100%	100%	99.1%	Holt-Oram syndrome, 142900	CONGENITAL HEARTDISEASE PANEL¹ HEART DISORDERS PANEL¹ SHORT STATURE/SKELETAL DYSPLASIA PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS
TBX6	100%	99.9%	100%	98.7%	Spondylocostal dysostosis 5, 122600	SHORT STATURE/SKELETAL DYSPLASIA PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL¹
TBXA2R	100%	98.9%	100%	98.2%	{Bleeding disorder, platelet-type, 13, susceptibility to}, 614009	HEMOSTATIC/THROMB OTIC DISORDERS PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS

TBXAS1	100%	100%	100%	99.6%	Ghosal hematodiaphyseal syndrome, 231095	INHERITED BONE MARROW FAILURE AND/OR PREDISPOSITION PANEL TO HEMATOLOGICAL MALIGNANCIES HEMOSTATIC/THROMBOTIC DISORDERS PANEL SHORT STATURE/SKELETAL DYSPLASIA PANEL WITH GENOME WIDE CNV ANALYSIS METABOLIC DISORDERS PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹
TBXT	100%	99.9%	100%	99.1%	Sacral agenesis with vertebral anomalies, 615709;{Neural tube defects, susceptibility to}, 182940	MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹

TCAP	100%	100%	100%	100%	Cardiomyopathy, hypertrophic, 25, 607487;Muscular dystrophy, limb-girdle, autosomal recessive 7, 601954	HEART DISORDERS PANEL ¹ MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS MUSCLE DISORDERS PANEL COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹
TCEAL1	100%	100%	98.9%	68%	Hijazi-Reis syndrome, 301094	INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS

TCF12	100%	100%	100%	99.4%	Craniosynostosis 3, 615314;Hypogonadotropic hypogonadism 26 with or without anosmia, 619718	CRANIOFACIAL ANOMALIES PANEL WITH GENOME WIDE CNV ANALYSIS DISORDERS/DIFFERENCES OF SEX DEVELOPMENT (DSD) / PRIMARY ADRENAL INSUFFICIENCY PANEL HYPOGONADOTROPIC HYPOGONADISM (KALLMANN) PANEL SHORT STATURE/SKELETAL DYSPLASIA PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹
TCF20	100%	100%	100%	99.1%	Developmental delay with variable intellectual impairment and behavioral abnormalities, 618430	INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS

TCF3	100%	100%	100%	97.9%	Agammaglobulinemia 8B, autosomal recessive, 619824;Agammaglobulinemia 8A, autosomal dominant, 616941	PRIMARY IMMUNODEFICIENCIES PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹
TCF4	100%	100%	100%	99.6%	Pitt-Hopkins syndrome, 610954;Corneal dystrophy, Fuchs endothelial, 3, 613267	VISION DISORDERS PANEL EPILEPSY PANEL INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS
TCF7L2	100%	99.8%	100%	96.4%	{Diabetes mellitus, type 2, susceptibility to}, 125853	INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS

TCHH	100%	100%	99.9%	95.8%	?Uncombable hair syndrome 3, 617252	SKIN DISORDERS PANEL ¹ MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS
TCIRG1	100%	100%	100%	99%	Osteopetrosis, autosomal recessive 1, 259700	INHERITED BONE MARROW FAILURE AND/OR PREDISPOSITION PANEL TO HEMATOLOGICAL MALIGNANCIES COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹ SHORT STATURE/SKELETAL DYSPLASIA PANEL WITH GENOME WIDE CNV ANALYSIS METABOLIC DISORDERS PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS SKIN DISORDERS PANEL ¹ PRIMARY IMMUNODEFICIENCIES PANEL

TCN2	94.2%	94.2%	100%	99.2%	Transcobalamin II deficiency, 275350	INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS PRIMARY IMMUNODEFICIENCIES PANEL METABOLIC DISORDERS PANEL COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL¹
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TCOF1	100%	100%	100%	98.9%	Treacher Collins syndrome 1, 154500	CRANIOFACIAL ANOMALIES PANEL WITH GENOME WIDE CNV ANALYSIS OROFACIAL CLEFTING PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS SHORT STATURE/SKELETAL DYSPLASIA PANEL WITH GENOME WIDE CNV ANALYSIS HEARING IMPAIRMENT PANEL (INCLUDING GJB2)
TCP1	100%	100%	100%	99.6%		INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS

TCTN1	98.5%	96.9%	100%	99.3%	Joubert syndrome 13, 614173	VISION DISORDERS PANEL CILIOPATHIES PANEL RENAL DISORDERS PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹
TCTN2	98.5%	98.5%	100%	99.3%	Joubert syndrome 24, 616654;?Meckel syndrome 8, 613885	VISION DISORDERS PANEL CILIOPATHIES PANEL SHORT STATURE/SKELETAL DYSPLASIA PANEL WITH GENOME WIDE CNV ANALYSIS INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS RENAL DISORDERS PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹

TCTN3	100%	100%	100%	99.5%	Joubert syndrome 18, 614815;Orofaciodigital syndrome IV, 258860	COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹ OROFACIAL CLEFTING PANEL WITH GENOME WIDE CNV ANALYSIS VISION DISORDERS PANEL CILIOPATHIES PANEL DISORDERS/DIFFEREN CES OF SEX DEVELOPMENT (DSD) / PRIMARY ADRENAL INSUFFICIENCY PANEL SHORT STATURE/SKELETAL DYSPLASIA PANEL WITH GENOME WIDE CNV ANALYSIS INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS RENAL DISORDERS PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS
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TDGF1	100%	100%	100%	99.6%		CONGENITAL HEARTDISEASE PANEL¹ HEART DISORDERS PANEL¹ MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS
TDP1	100%	100%	100%	99.5%	?Spinocerebellar ataxia, autosomal recessive, with axonal neuropathy 1, 607250	MOVEMENT DISORDERS PANEL POLYNEUROPATHIES PANEL¹ MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL¹

TDP2	100%	100%	100%	99.2%	Spinocerebellar ataxia, autosomal recessive 23, 616949	MOVEMENT DISORDERS PANEL EPILEPSY PANEL INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS MITOCHONDRIAL DISORDERS PANEL COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹
TDRD12	100%	100%	100%	99.4%		MALE INFERTILITY PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS
TDRD7	100%	100%	100%	99.5%	Cataract 36, 613887	VISION DISORDERS PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹

TDRD9	100%	100%	100%	99.1%	?Spermatogenic failure 30, 618110	MALE INFERTILITY PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS
TDRKH	100%	100%	100%	97.6%		POLYNEUROPATHIES PANEL ¹ MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS
TEAD1	100%	100%	100%	99.2%	Sveinsson chorioretinal atrophy, 108985	VISION DISORDERS PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS

TECPR2	100%	100%	100%	99.1%	Neuropathy, hereditary sensory and autonomic, type IX, with developmental delay, 615031	MOVEMENT DISORDERS PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL¹ POLYNEUROPATHIES PANEL¹ NEUROLOGICAL PAIN DISORDERS PANEL¹ INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS
TECR	100%	100%	100%	97.5%	Intellectual developmental disorder, autosomal recessive 14, 614020	METABOLIC DISORDERS PANEL INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL¹

TECRL	100%	100%	100%	99.7%	Ventricular tachycardia, catecholaminergic polymorphic, 3, 614021	HEART DISORDERS PANEL ¹ LONG QT SYNDROME PANEL ¹ MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹ ARRHYTHMIA AND CARDIAC CONDUCTION DISORDERS PANEL ¹
TECTA	100%	100%	100%	99.4%	Deafness, autosomal dominant 8/12, 601543;Deafness, autosomal recessive 21, 603629	HEARING IMPAIRMENT PANEL (INCLUDING GJB2) MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹

TEFM	100%	100%	100%	98.8%	Combined oxidative phosphorylation deficiency 58, 620451	INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS MITOCHONDRIAL DISORDERS PANEL COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹
TEK	100%	100%	100%	99.3%	Venous malformations, multiple cutaneous and mucosal, 600195;Glaucoma 3, primary congenital, E, 617272	VISION DISORDERS PANEL SKIN DISORDERS PANEL ¹ MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS
TEKT3	100%	100%	100%	99.1%	Spermatogenic failure 81, 620277	MALE INFERTILITY PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS

TELO2	100%	99.9%	100%	99.4%	You-Hoover-Fong syndrome, 616954	INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL¹
TENM1	100%	99.8%	98.9%	68.9%		DISORDERS/DIFFEREN CES OF SEX DEVELOPMENT (DSD) / PRIMARY ADRENAL INSUFFICIENCY PANEL HYPOGONADOTROPIC HYPOGONADISM (KALLMANN) PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS

TENM3	100%	100%	100%	99.2%	Microphthalmia, syndromic 15, 615145;?Microphthalmia/coloboma 9, 615145	VISION DISORDERS PANEL INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹
TENM4	100%	100%	100%	99%	Essential tremor, hereditary, 5, 616736	MOVEMENT DISORDERS PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS
TENT5A	100%	100%	100%	98.7%	Osteogenesis imperfecta, type XVIII, 617952	SHORT STATURE/SKELETAL DYSPLASIA PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹

TENT5D	100%	100%	99.2%	70%		MALE INFERTILITY PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS
TERB1	100%	100%	100%	99.3%	Spermatogenic failure 60, 619646	MALE INFERTILITY PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS
TERB2	100%	100%	100%	99.3%	?Spermatogenic failure 59, 619645	MALE INFERTILITY PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS

TERC					Pulmonary fibrosis and/or bone marrow failure syndrome, telomere-related, 2, 614743;Dyskeratosis congenita, autosomal dominant 1, 127550	INHERITED BONE MARROW FAILURE AND/OR PREDISPOSITION PANEL TO HEMATOLOGICAL MALIGNANCIES SKIN DISORDERS PANEL ¹ DYSKERATOSIS CONGENITA AND APLASTIC ANEMIA PANEL PRIMARY IMMUNODEFICIENCIES PANEL LIVER DISORDERS PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS HEREDITARY CANCER PANEL
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TERF2IP	99.7%	94.2%	100%	99.5%		INHERITED BONE MARROW FAILURE AND/OR PREDISPOSITION PANEL TO HEMATOLOGICAL MALIGNANCIES SKIN DISORDERS PANEL ¹ DYSKERATOSIS CONGENITA AND APLASTIC ANEMIA PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS HEREDITARY CANCER PANEL
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TERT	100%	100%	100%	99%	Dyskeratosis congenita, autosomal dominant 2, 613989;Dyskeratosis congenita, autosomal recessive 4, 613989;Pulmonary fibrosis and/or bone marrow failure syndrome, telomere-related, 1, 614742;{Melanoma, cutaneous malignant, 9}, 615134;{Leukemia, acute myeloid}, 601626	INHERITED BONE MARROW FAILURE AND/OR PREDISPOSITION PANEL TO HEMATOLOGICAL MALIGNANCIES SKIN DISORDERS PANEL ¹ DYSKERATOSIS CONGENITA AND APLASTIC ANEMIA PANEL PRIMARY IMMUNODEFICIENCIES PANEL LIVER DISORDERS PANEL PANEL MELANOMA, EXTENSIVE (CDKN2A, CDK4, MITF P.(GLU318LYS), BAP1, POT1, TERT PROMOTER) ¹ MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS HEREDITARY CANCER PANEL
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TES	100%	100%	100%	99.7%		THORACIC AORTIC ANEURYSM AND/OR DISSECTION PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS
TET2	100%	100%	100%	99.5%	Myelodysplastic syndrome, somatic, 614286;Immunodeficiency 75, 619126	INHERITED BONE MARROW FAILURE AND/OR PREDISPOSITION PANEL TO HEMATOLOGICAL MALIGNANCIES PRIMARY IMMUNODEFICIENCIES PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL¹

TET3	100%	100%	100%	98.5%	Beck-Fahrner syndrome, 618798	INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹
TEX11	97.1%	97%	98.9%	68.5%	Spermatogenic failure, X-linked 2, 309120	MALE INFERTILITY PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS
TEX14	100%	100%	100%	99.5%	Spermatogenic failure 23, 617707	MALE INFERTILITY PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS
TEX15	100%	100%	100%	99.5%	Spermatogenic failure 25, 617960	MALE INFERTILITY PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS

TF	100%	100%	100%	99.3%	Atransferrinemia, 209300	IRON DISORDERS PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹
TFAM	100%	100%	100%	99.6%	?Mitochondrial DNA depletion syndrome 15 (hepatocerebral type), 617156	MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS MITOCHONDRIAL DISORDERS PANEL COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹

TFAP2A	100%	100%	100%	97.7%	Branchiooculofacial syndrome, 113620	VISION DISORDERS PANEL CRANIOFACIAL ANOMALIES PANEL WITH GENOME WIDE CNV ANALYSIS SKIN DISORDERS PANEL ¹ OROFACIAL CLEFTING PANEL WITH GENOME WIDE CNV ANALYSIS HEARING IMPAIRMENT PANEL (INCLUDING GJB2) INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS
TFAP2B	100%	100%	100%	98.6%	Patent ductus arteriosus 2, 617035;Char syndrome, 169100	CONGENITAL HEARTDISEASE PANEL ¹ HEART DISORDERS PANEL ¹ MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS

TFB2M	100%	100%	100%	99.9%		MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS MITOCHONDRIAL DISORDERS PANEL
TFE3	100%	99.4%	97.4%	63.3%	Intellectual developmental disorder, X-linked syndromic, with pigmentary mosaicism and coarse facies, 301066;Renal cell carcinoma, papillary, 1, 300854	INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS
TFG	100%	100%	100%	99.5%	?Spastic paraplegia 57, autosomal recessive, 615658;Hereditary motor and sensory neuropathy, Okinawa type, 604484	POLYNEUROPATHIES PANEL ¹ MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹
TFPT	100%	100%	100%	98.4%		VISION DISORDERS PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS

TFR2	100%	99.9%	100%	97.5%	Hemochromatosis, type 3, 604250	IRON DISORDERS PANEL LIVER DISORDERS PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹
TFRC	95.5%	95.5%	99.9%	98.7%	Immunodeficiency 46, 616740	COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹ IRON DISORDERS PANEL PRIMARY IMMUNODEFICIENCIES PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS
TG	100%	100%	100%	98.8%	{Autoimmune thyroid disease, susceptibility to, 3}, 608175;Thyroid dyshormonogenesis 3, 274700	MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹ HEREDITARY CANCER PANEL

TGDS	100%	100%	100%	99.5%	Catel-Manzke syndrome, 616145	SHORT STATURE/SKELETAL DYSPLASIA PANEL WITH GENOME WIDE CNV ANALYSIS INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL¹ OROFACIAL CLEFTING PANEL WITH GENOME WIDE CNV ANALYSIS
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TGFB1	100%	99.8%	100%	96%	Inflammatory bowel disease, immunodeficiency, and encephalopathy, 618213;Camurati-Engelmann disease, 131300;{Cystic fibrosis lung disease, modifier of}, 219700	PRIMARY IMMUNODEFICIENCIES PANEL SHORT STATURE/SKELETAL DYSPLASIA PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹
TGFB2	100%	100%	100%	99.1%	Loeys-Dietz syndrome 4, 614816	THORACIC AORTIC ANEURYSM AND/OR DISSECTION PANEL SKIN DISORDERS PANEL ¹ TALL STATURE PANEL WITH GENOME WIDE CNV ANALYSIS SHORT STATURE/SKELETAL DYSPLASIA PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS

TGFB3	100%	100%	100%	99.1%	Arrhythmogenic right ventricular dysplasia 1, 107970;Loeys-Dietz syndrome 5, 615582	OROFACIAL CLEFTING PANEL WITH GENOME WIDE CNV ANALYSIS THORACIC AORTIC ANEURYSM AND/OR DISSECTION PANEL TALL STATURE PANEL WITH GENOME WIDE CNV ANALYSIS HEART DISORDERS PANEL ¹ SHORT STATURE/SKELETAL DYSPLASIA PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS
TGFBI	100%	100%	100%	99.3%	Corneal dystrophy, Avellino type, 607541;Corneal dystrophy, Reis-Bucklers type, 608470;Corneal dystrophy, Thiel-Behnke type, 602082;Corneal dystrophy, Groenouw type I, 121900;Corneal dystrophy, epithelial basement membrane, 121820;Corneal dystrophy, lattice type I, 122200;Corneal dystrophy, lattice type IIIA, 608471	VISION DISORDERS PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS

TGFBR1	99.8%	99.3%	100%	99.4%	{Multiple self-healing squamous epithelioma, susceptibility to}, 132800;Loeys-Dietz syndrome 1, 609192	THORACIC AORTIC ANEURYSM AND/OR DISSECTION PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS OROFACIAL CLEFTING PANEL WITH GENOME WIDE CNV ANALYSIS HEREDITARY CANCER PANEL SHORT STATURE/SKELETAL DYSPLASIA PANEL WITH GENOME WIDE CNV ANALYSIS INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS CRANIOFACIAL ANOMALIES PANEL WITH GENOME WIDE CNV ANALYSIS SKIN DISORDERS PANEL ¹ TALL STATURE PANEL WITH GENOME WIDE CNV ANALYSIS
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TGFBR2	100%	100%	100%	98.8%	Loeys-Dietz syndrome 2, 610168;Colorectal cancer, hereditary nonpolyposis, type 6, 614331;Esophageal cancer, somatic, 133239	THORACIC AORTIC ANEURYSM AND/OR DISSECTION PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS TALL STATURE PANEL WITH GENOME WIDE CNV ANALYSIS SHORT STATURE/SKELETAL DYSPLASIA PANEL WITH GENOME WIDE CNV ANALYSIS CRANIOFACIAL ANOMALIES PANEL WITH GENOME WIDE CNV ANALYSIS SKIN DISORDERS PANEL ¹ OROFACIAL CLEFTING PANEL WITH GENOME WIDE CNV ANALYSIS
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TGIF1	100%	100%	100%	99.5%	Holoprosencephaly 4, 142946	MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS OROFACIAL CLEFTING PANEL WITH GENOME WIDE CNV ANALYSIS CRANIOFACIAL ANOMALIES PANEL WITH GENOME WIDE CNV ANALYSIS INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS
TGM1	100%	100%	100%	98.9%	Ichthyosis, congenital, autosomal recessive 1, 242300	SKIN DISORDERS PANEL ¹ MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹
TGM3	100%	100%	100%	99.2%	?Uncombable hair syndrome 2, 617251	SKIN DISORDERS PANEL ¹ MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS

TGM5	100%	100%	100%	99.2%	Peeling skin syndrome 2, 609796	SKIN DISORDERS PANEL ¹ MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹
TGM6	100%	100%	100%	99.4%	Spinocerebellar ataxia 35, 613908	MOVEMENT DISORDERS PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS
TH	100%	100%	100%	98.4%	Segawa syndrome, recessive, 605407	MOVEMENT DISORDERS PANEL METABOLIC DISORDERS PANEL INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS PARKINSON DISEASE PANEL COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹

THAP1	100%	100%	100%	99.7%	Dystonia 6, torsion, 602629	MOVEMENT DISORDERS PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS
THAP11	100%	100%	100%	98%	?Methylmalonic aciduria and homocystinuria, cblL type, 620940;Spinocerebellar ataxia 51, 620947	MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS
THBD	100%	100%	100%	98.4%	Thrombophilia 12 due to thrombomodulin defect, 614486;{Hemolytic uremic syndrome, atypical, susceptibility to, 6}, 612926	HEMOSTATIC/THROMB OTIC DISORDERS PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS RENAL DISORDERS PANEL PRIMARY IMMUNODEFICIENCIES PANEL
THBS1	100%	100%	100%	99.4%		HEARING IMPAIRMENT PANEL (INCLUDING GJB2) MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS

THBS2	100%	100%	100%	98.8%	?Ehlers-Danlos syndrome, classic-like, 3, 620865;{Lumbar disc herniation, susceptibility to}, 603932	THORACIC AORTIC ANEURYSM AND/OR DISSECTION PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS
THBS4	100%	100%	100%	99.6%		HEART DISORDERS PANEL ¹ MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS
THG1L	100%	100%	100%	99.5%	Spinocerebellar ataxia, autosomal recessive 28, 618800	INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS MITOCHONDRIAL DISORDERS PANEL COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹

THOC1	100%	100%	100%	99.6%	?Deafness, autosomal dominant 86, 620280	HEARING IMPAIRMENT PANEL (INCLUDING GJB2) MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS
THOC2	100%	99.9%	99.2%	71.3%	Arthrogryposis multiplex congenita 7, X-linked, 301127;Intellectual developmental disorder, X-linked syndromic, Kumar type, 300957	INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS
THOC6	100%	100%	100%	98.6%	Beaulieu-Boycott-Innes syndrome, 613680	INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹

THPO	100%	100%	100%	99.6%	Thrombocythemia 1, 187950;Thrombocytopenia 9, 620478;Amegakaryocytic thrombocytopenia, congenital, 2, 620481	INHERITED BONE MARROW FAILURE AND/OR PREDISPOSITION PANEL TO HEMATOLOGICAL MALIGNANCIES HEMOSTATIC/THROMBOTIC DISORDERS PANEL SHORT STATURE/SKELETAL DYSPLASIA PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹ HEREDITARY CANCER PANEL
THRA	100%	100%	100%	99%	Hypothyroidism, congenital, nongoitrous, 6, 614450	MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS

THRB	100%	100%	100%	99.3%	Thyroid hormone resistance, autosomal recessive, 274300;Thyroid hormone resistance, 188570;Thyroid hormone resistance, selective pituitary, 145650	VISION DISORDERS PANEL INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹
THSD1	100%	100%	100%	99.7%	?Aneurysm, intracranial berry, 12, 618734;Lymphatic malformation 13, 620244	THORACIC AORTIC ANEURYSM AND/OR DISSECTION PANEL LYMPHATIC ANOMALIES PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹

THSD4	100%	100%	100%	99.3%	Aortic aneurysm, familial thoracic 12, 619825	THORACIC AORTIC ANEURYSM AND/OR DISSECTION PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS
THUMPD1	100%	99.8%	100%	99%	Neurodevelopmental disorder with speech delay and variable ocular anomalies, 619989	INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹
TIA1	100%	100%	100%	99.6%	Welander distal myopathy, 604454;Amyotrophic lateral sclerosis 26 with or without frontotemporal dementia, 619133	MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS MUSCLE DISORDERS PANEL

TIAM1	100%	100%	100%	99.1%	Neurodevelopmental disorder with language delay and seizures, 619908	INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹
TICAM1	100%	100%	100%	98.9%	{Encephalopathy, acute, infection-induced (herpes-specific), susceptibility to, 6}, 614850	PRIMARY IMMUNODEFICIENCIES PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS
TIE1	100%	100%	100%	99%	Lymphatic malformation 11, 619401	LYMPHATIC ANOMALIES PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS
TIMELESS	100%	100%	100%	99.2%	?Advance sleep phase syndrome, familial, 4, 620015	MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS

TIMM22	100%	100%	99.8%	97.3%	?Combined oxidative phosphorylation deficiency 43, 618851	MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS MITOCHONDRIAL DISORDERS PANEL
TIMM44	100%	100%	100%	98.4%		MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS MITOCHONDRIAL DISORDERS PANEL
TIMM50	100%	100%	100%	99%	3-methylglutaconic aciduria, type IX, 617698	EPILEPSY PANEL METABOLIC DISORDERS PANEL INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS MITOCHONDRIAL DISORDERS PANEL COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹

TIMM8A	100%	98.6%	99.4%	65%	Mohr-Tranebjaerg syndrome, 304700	MOVEMENT DISORDERS PANEL VISION DISORDERS PANEL HEARING IMPAIRMENT PANEL (INCLUDING GJB2) INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS MITOCHONDRIAL DISORDERS PANEL
TIMMDC1	100%	100%	100%	99.6%	Mitochondrial complex I deficiency, nuclear type 31, 618251	MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS MITOCHONDRIAL DISORDERS PANEL COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹
TIMP3	100%	100%	100%	99%	Sorsby fundus dystrophy, 136900	VISION DISORDERS PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS

TINF2	100%	100%	100%	98.7%	Dyskeratosis congenita, autosomal dominant 3, 613990;Revesz syndrome, 268130	VISION DISORDERS PANEL INHERITED BONE MARROW FAILURE AND/OR PREDISPOSITION PANEL TO HEMATOLOGICAL MALIGNANCIES SKIN DISORDERS PANEL ¹ DYSKERATOSIS CONGENITA AND APLASTIC ANEMIA PANEL PRIMARY IMMUNODEFICIENCIES PANEL PANEL MELANOMA, EXTENSIVE (CDKN2A, CDK4, MITF P.(GLU318LYS), BAP1, POT1, TERT PROMOTER) ¹ INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS HEREDITARY CANCER PANEL
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TIRAP	100%	100%	100%	99.3%	{Malaria, protection against}, 611162;{Tuberculosis, protection against}, 607948;{Bacteremia, protection against}, 614382	PRIMARY IMMUNODEFICIENCIES PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS
TJP1	100%	100%	100%	99.4%		HEART DISORDERS PANEL ¹ MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS
TJP2	100%	100%	100%	99.3%	Hypercholanemia, familial 1, 607748;Cholestasis, progressive familial intrahepatic 4, 615878	HEARING IMPAIRMENT PANEL (INCLUDING GJB2) LIVER DISORDERS PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹

TK2	100%	100%	100%	99.4%	Mitochondrial DNA depletion syndrome 2 (myopathic type), 609560;?Progressive external ophthalmoplegia with mitochondrial DNA deletions, autosomal recessive 3, 617069	METABOLIC DISORDERS PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS MITOCHONDRIAL DISORDERS PANEL COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹ MUSCLE DISORDERS PANEL
TKFC	100%	100%	100%	99%	Triokinase and FMN cyclase deficiency syndrome, 618805	METABOLIC DISORDERS PANEL INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS

TKT	98.1%	98.1%	100%	99%	Short stature, developmental delay, and congenital heart defects, 617044	METABOLIC DISORDERS PANEL INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹
TKTL1	100%	98.8%	98.7%	68.5%		MALE INFERTILITY PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS
TLCD3B	100%	100%	100%	97.8%	Cone-rod dystrophy 22, 619531	VISION DISORDERS PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS

TLE6	100%	100%	100%	98.3%	Oocyte/zygote/embryo maturation arrest 15, 616814	UNSUCCESSFUL IVF TREATMENTS, RECURRENT MOLAR PREGNANCIES AND/OR RECURRENT PREGNANCY LOSS PANEL ¹ MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹
TLK2	100%	100%	100%	99.4%	Intellectual developmental disorder, autosomal dominant 57, 618050	CRANIOFACIAL ANOMALIES PANEL WITH GENOME WIDE CNV ANALYSIS INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS

TLL1	100%	100%	100%	99.2%	Atrial septal defect 6, 613087	CONGENITAL HEARTDISEASE PANEL¹ HEART DISORDERS PANEL¹ MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS
TLR3	100%	100%	100%	99.3%	{HIV1 infection, resistance to}, 609423;{Immunodeficiency 83, susceptibility to viral infections}, 613002	PRIMARY IMMUNODEFICIENCIES PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS
TLR4	100%	100%	100%	99.7%		PRIMARY IMMUNODEFICIENCIES PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS
TLR5	100%	100%	100%	99.4%	{Melioidosis, susceptibility to}, 615557;{Systemic lupus erythematosus, susceptibility to, 1}, 601744;{Systemic lupus erythematosus, resistance to}, 601744;{Legionnaire disease, susceptibility to}, 608556	PRIMARY IMMUNODEFICIENCIES PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS

TLR7	100%	100%	99.4%	74.5%	Immunodeficiency 74, COVID19-related, X-linked, 301051;Systemic lupus erythematosus 17, 301080	PRIMARY IMMUNODEFICIENCIES PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS
TLR8	100%	100%	99.3%	73.8%	Immunodeficiency 98 with autoinflammation, X-linked, 301078	INHERITED BONE MARROW FAILURE AND/OR PREDISPOSITION PANEL TO HEMATOLOGICAL MALIGNANCIES MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS PRIMARY IMMUNODEFICIENCIES PANEL
TMC1	100%	100%	100%	99%	Deafness, autosomal dominant 36, 606705;Deafness, autosomal recessive 7, 600974	COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹ MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS HEARING IMPAIRMENT PANEL (INCLUDING GJB2)

TMC6	100%	100%	100%	98.9%	{Epidermodysplasia verruciformis, susceptibility to, 1}, 226400	MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹ PRIMARY IMMUNODEFICIENCIES PANEL SKIN DISORDERS PANEL ¹
TMC8	100%	100%	100%	99.4%	{Epidermodysplasia verruciformis, susceptibility to, 2}, 618231	MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹ SKIN DISORDERS PANEL ¹ PRIMARY IMMUNODEFICIENCIES PANEL

TMCO1	87.7%	87.7%	100%	98.7%	Craniofacial dysmorphism, skeletal anomalies, and impaired intellectual development 1, 213980	SHORT STATURE/SKELETAL DYSPLASIA PANEL WITH GENOME WIDE CNV ANALYSIS INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹ OROFACIAL CLEFTING PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS
TMCO3	100%	100%	100%	99.2%		VISION DISORDERS PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS

TMEM106B	100%	100%	100%	99.5%	Leukodystrophy, hypomyelinating, 16, 617964	MOVEMENT DISORDERS PANEL METABOLIC DISORDERS PANEL INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS
TMEM107	100%	100%	100%	98.3%	Orofaciodigital syndrome XVI, 617563;Meckel syndrome 13, 617562;?Joubert syndrome 29, 617562	CILIOPATHIES PANEL INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS RENAL DISORDERS PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL¹

TMEM126A	100%	100%	100%	99.4%	Optic atrophy 7, 612989	VISION DISORDERS PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS MITOCHONDRIAL DISORDERS PANEL COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹
TMEM126B	100%	100%	100%	99.6%	Mitochondrial complex I deficiency, nuclear type 29, 618250	MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS MITOCHONDRIAL DISORDERS PANEL COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹
TMEM127	100%	100%	100%	98.5%	{Pheochromocytoma, susceptibility to}, 171300	MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS HEREDITARY CANCER PANEL

TMEM132E	100%	100%	100%	98.8%	Deafness, autosomal recessive 99, 618481	HEARING IMPAIRMENT PANEL (INCLUDING GJB2) MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL¹
TMEM138	99.9%	97.6%	100%	99%	Joubert syndrome 16, 614465	VISION DISORDERS PANEL CILIOPATHIES PANEL RENAL DISORDERS PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL¹

TMEM147	100%	100%	100%	99.2%	Neurodevelopmental disorder with facial dysmorphism, absent language, and pseudo-Pelger-Huet anomaly, 620075	INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹
TMEM14C	100%	100%	100%	99.3%		IRON DISORDERS PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS
TMEM151A	100%	100%	100%	97.8%	Episodic kinesigenic dyskinesia 3, 620245	MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS
TMEM163	100%	99.9%	100%	99.2%	Leukodystrophy, hypomyelinating, 25, 620243	INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS

TMEM165	100%	100%	100%	99.6%	Congenital disorder of glycosylation, type IIk, 614727	SKIN DISORDERS PANEL ¹ SHORT STATURE/SKELETAL DYSPLASIA PANEL WITH GENOME WIDE CNV ANALYSIS METABOLIC DISORDERS PANEL INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹
TMEM184B	100%	100%	100%	99%		INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS

TMEM186	100%	100%	100%	99.4%		MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS MITOCHONDRIAL DISORDERS PANEL
TMEM199	100%	100%	100%	98.8%	Congenital disorder of glycosylation, type IIp, 616829	METABOLIC DISORDERS PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹

TMEM216	100%	100%	100%	99.8%	Joubert syndrome 2, 608091;Retinitis pigmentosa 98, 620996;Meckel syndrome 2, 603194	VISION DISORDERS PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹ OROFACIAL CLEFTING PANEL WITH GENOME WIDE CNV ANALYSIS CILIOPATHIES PANEL SHORT STATURE/SKELETAL DYSPLASIA PANEL WITH GENOME WIDE CNV ANALYSIS INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS RENAL DISORDERS PANEL
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TMEM218	100%	100%	100%	99%	Joubert syndrome 39, 619562	VISION DISORDERS PANEL COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹ CILIOPATHIES PANEL INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS
TMEM222	100%	100%	100%	99.7%	Neurodevelopmental disorder with motor and speech delay and behavioral abnormalities, 619470	INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹

TMEM231	93.2%	93.2%	100%	98.7%	Joubert syndrome 20, 614970;Meckel syndrome 11, 615397	VISION DISORDERS PANEL CILIOPATHIES PANEL COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL¹ SHORT STATURE/SKELETAL DYSPLASIA PANEL WITH GENOME WIDE CNV ANALYSIS INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS RENAL DISORDERS PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS
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TMEM237	98.2%	98.2%	100%	99.2%	Joubert syndrome 14, 614424	VISION DISORDERS PANEL CILIOPATHIES PANEL INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS RENAL DISORDERS PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL¹
TMEM240	100%	100%	100%	93.7%	Spinocerebellar ataxia 21, 607454	MOVEMENT DISORDERS PANEL INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS

TMEM260	100%	100%	100%	99.5%	Structural heart defects and renal anomalies syndrome, 617478	CONGENITAL HEARTDISEASE PANEL¹ CILIOPATHIES PANEL HEART DISORDERS PANEL¹ RENAL DISORDERS PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL¹
TMEM38B	100%	100%	100%	99.7%	Osteogenesis imperfecta, type XIV, 615066	SHORT STATURE/SKELETAL DYSPLASIA PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL¹

TMEM43	100%	100%	100%	99.3%	Arrhythmogenic right ventricular dysplasia 5, 604400;Auditory neuropathy, autosomal dominant 3, 619832;Emery-Dreifuss muscular dystrophy 7, AD, 614302	ARITMOGENE CARDIOMYOPATHY PANEL ¹ HEARING IMPAIRMENT PANEL (INCLUDING GJB2) HEART DISORDERS PANEL ¹ MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS ARRHYTHMIA AND CARDIAC CONDUCTION DISORDERS PANEL ¹
TMEM53	100%	100%	100%	99.2%	Craniotubular dysplasia, Ikegawa type, 619727	SHORT STATURE/SKELETAL DYSPLASIA PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹

TMEM63A	100%	100%	100%	99.5%	Leukodystrophy, hypomyelinating, 19, transient infantile, 618688	INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS
TMEM63B	100%	100%	100%	98.9%		INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS
TMEM63C	100%	100%	100%	98.4%	Spastic paraplegia 87, autosomal recessive, 619966	INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS MITOCHONDRIAL DISORDERS PANEL COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹

TMEM65	99.7%	95.3%	100%	97.8%		MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS MITOCHONDRIAL DISORDERS PANEL
TMEM67	96.1%	96.1%	100%	99.5%	Nephronophthisis 11, 613550;{Bardet-Biedl syndrome 14, modifier of}, 615991;Joubert syndrome 6, 610688;Meckel syndrome 3, 607361;?RHYNS syndrome, 602152;COACH syndrome 1, 216360	MOVEMENT DISORDERS PANEL VISION DISORDERS PANEL CILIOPATHIES PANEL SHORT STATURE/SKELETAL DYSPLASIA PANEL WITH GENOME WIDE CNV ANALYSIS LIVER DISORDERS PANEL INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS RENAL DISORDERS PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹

TMEM70	100%	100%	100%	99.6%	Mitochondrial complex V (ATP synthase) deficiency, nuclear type 2, 614052	LIVER DISORDERS PANEL METABOLIC DISORDERS PANEL MITOCHONDRIAL DISORDERS PANEL COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL¹ INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS
TMEM94	100%	100%	100%	99.1%	Intellectual developmental disorder with cardiac defects and dysmorphic facies, 618316	INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL¹

TMEM98	100%	100%	100%	99.4%	Nanophthalmos 4, 615972	VISION DISORDERS PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS
TMIE	100%	100%	100%	99.3%	Deafness, autosomal recessive 6, 600971	HEARING IMPAIRMENT PANEL (INCLUDING GJB2) MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹
TMLHE	100%	99.9%	99.2%	74%	{Autism, susceptibility to, X-linked 6}, 300872	METABOLIC DISORDERS PANEL INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS

TMPO	100%	100%	100%	99.6%		HEART DISORDERS PANEL ¹ MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS
TMPRSS15	100%	100%	100%	99.5%	Enterokinase deficiency, 226200	MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹
TMPRSS3	100%	100%	100%	99%	Deafness, autosomal recessive 8/10, 601072	HEARING IMPAIRMENT PANEL (INCLUDING GJB2) MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹

TMPRSS6	100%	99.9%	100%	98.6%	Iron-refractory iron deficiency anemia, 206200	IRON DISORDERS PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL¹
TMTC2	97.5%	97.1%	100%	99.6%		HEARING IMPAIRMENT PANEL (INCLUDING GJB2) MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS
TMTC3	100%	100%	100%	99.3%	Lissencephaly 8, 617255	EPILEPSY PANEL INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL¹

TMTC4	100%	100%	100%	99.4%	?Deafness, autosomal recessive 122, 620714	HEARING IMPAIRMENT PANEL (INCLUDING GJB2) MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS
TMX2	100%	100%	100%	99.1%	Neurodevelopmental disorder with microcephaly, cortical malformations, and spasticity, 618730	INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS MITOCHONDRIAL DISORDERS PANEL COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹
TNC	100%	100%	100%	99.1%	Deafness, autosomal dominant 56, 615629	HEARING IMPAIRMENT PANEL (INCLUDING GJB2) MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS

TNF	100%	100%	100%	98.3%	{Migraine without aura, susceptibility to}, 157300;{Dementia, vascular, susceptibility to};?Immunodeficiency 127, 620977;{Asthma, susceptibility to}, 600807;{Septic shock, susceptibility to};{Malaria, cerebral, susceptibility to}, 611162	MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS
TNFAIP3	100%	100%	100%	99.4%	Autoinflammatory syndrome, familial, Behcet-like 1, 616744	MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS PRIMARY IMMUNODEFICIENCIES PANEL
TNFRSF10B	100%	100%	100%	99.1%	Squamous cell carcinoma, head and neck, 275355	MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS

TNFRSF11A	99.6%	98.6%	100%	99%	Osteopetrosis, autosomal recessive 7, 612301;{Paget disease of bone 2, early-onset}, 602080;Osteolysis, familial expansile, 174810	INHERITED BONE MARROW FAILURE AND/OR PREDISPOSITION PANEL TO HEMATOLOGICAL MALIGNANCIES SKIN DISORDERS PANEL ¹ PRIMARY IMMUNODEFICIENCIES PANEL SHORT STATURE/SKELETAL DYSPLASIA PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹ HEREDITARY CANCER PANEL
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TNFRSF11B	100%	100%	100%	99.4%	Paget disease of bone 5, juvenile-onset, 239000	SKIN DISORDERS PANEL ¹ SHORT STATURE/SKELETAL DYSPLASIA PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹
TNFRSF13B	100%	100%	100%	98.8%	Immunodeficiency, common variable, 2, 240500;Immunoglobulin A deficiency 2, 609529	PRIMARY IMMUNODEFICIENCIES PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹

TNFRSF13C	100%	100%	100%	95.3%	Immunodeficiency, common variable, 4, 613494	PRIMARY IMMUNODEFICIENCIES PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹
TNFRSF1A	100%	100%	100%	99%	{Multiple sclerosis, susceptibility to, 5}, 614810;Periodic fever, familial, 142680	SKIN DISORDERS PANEL ¹ PRIMARY IMMUNODEFICIENCIES PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS
TNFRSF4	100%	99.9%	100%	98.3%	?Immunodeficiency 16, 615593	PRIMARY IMMUNODEFICIENCIES PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹

TNFRSF9	100%	100%	100%	99.8%	Immunodeficiency 109 with lymphoproliferation, 620282	PRIMARY IMMUNODEFICIENCIES PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL¹
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TNFSF11	100%	100%	100%	99.4%	Osteopetrosis, autosomal recessive 2, 259710	INHERITED BONE MARROW FAILURE AND/OR PREDISPOSITION PANEL TO HEMATOLOGICAL MALIGNANCIES SKIN DISORDERS PANEL ¹ PRIMARY IMMUNODEFICIENCIES PANEL SHORT STATURE/SKELETAL DYSPLASIA PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹
TNFSF12	100%	100%	100%	97.8%		PRIMARY IMMUNODEFICIENCIES PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS

TNFSF13	100%	99.5%	100%	96.8%		PRIMARY IMMUNODEFICIENCIES PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS
TNIK	100%	100%	100%	98.9%	Intellectual developmental disorder, autosomal recessive 54, 617028	METABOLIC DISORDERS PANEL INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹
TNNC1	100%	100%	100%	98.9%	Cardiomyopathy, dilated, 1Z, 611879;Cardiomyopathy, hypertrophic, 13, 613243	DILATED CARDIOMYOPATHY PANEL ¹ HEART DISORDERS PANEL ¹ HYPERTROPHIC CARDIOMYOPATHY PANEL ¹ MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS

TNNC2	100%	100%	100%	98.4%	Congenital myopathy 15, 620161	MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS MUSCLE DISORDERS PANEL
TNNI2	100%	100%	100%	99.6%	Arthrogryposis, distal, type 2B1, 601680	FETAL AKINESIA PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS MUSCLE DISORDERS PANEL
TNNI3	100%	100%	100%	97.2%	?Cardiomyopathy, dilated, 2A, 611880;Cardiomyopathy, hypertrophic, 7, 613690;Cardiomyopathy, familial restrictive, 1, 115210;Cardiomyopathy, dilated, 1FF, 613286	DILATED CARDIOMYOPATHY PANEL ¹ HEART DISORDERS PANEL ¹ HYPERTROPHIC CARDIOMYOPATHY PANEL ¹ MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹

TNNI3K	100%	100%	100%	99.8%	Cardiac conduction disease with or without dilated cardiomyopathy, 616117	HEART DISORDERS PANEL ¹ MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS
TNNT1	100%	100%	100%	97%	Nemaline myopathy 5C, autosomal dominant, 620389;Nemaline myopathy 5A, autosomal recessive, severe infantile, 605355;Nemaline myopathy 5B, autosomal recessive, childhood-onset, 620386	MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹ MUSCLE DISORDERS PANEL
TNNT2	100%	100%	100%	98.9%	Cardiomyopathy, dilated, 1D, 601494;Cardiomyopathy, hypertrophic, 2, 115195;Cardiomyopathy, familial restrictive, 3, 612422;Left ventricular noncompaction 6, 601494	DILATED CARDIOMYOPATHY PANEL ¹ HEART DISORDERS PANEL ¹ HYPERTROPHIC CARDIOMYOPATHY PANEL ¹ MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS ARRHYTHMIA AND CARDIAC CONDUCTION DISORDERS PANEL ¹

TNNT3	100%	100%	100%	99%	Arthrogryposis, distal, type 2B2, 618435	FETAL AKINESIA PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS
TNPO2	100%	100%	100%	98.3%	Intellectual developmental disorder with hypotonia, impaired speech, and dysmorphic facies, 619556	INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS
TNPO3	100%	100%	100%	99.7%	Muscular dystrophy, limb-girdle, autosomal dominant 2, 608423	MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS MUSCLE DISORDERS PANEL

TNR	100%	100%	100%	99.2%	Neurodevelopmental disorder, nonprogressive, with spasticity and transient opisthotonus, 619653	INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹
TNRC6A	100%	100%	100%	99.4%	?Epilepsy, familial adult myoclonic, 6, 618074	MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS
TNRC6B	100%	100%	100%	99.1%	Global developmental delay with speech and behavioral abnormalities, 619243	INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS MUSCLE DISORDERS PANEL

TNS1	100%	100%	100%	99.2%		CONGENITAL HEARTDISEASE PANEL¹ HEART DISORDERS PANEL¹ MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS
TNS2	100%	100%	100%	98.7%		RENAL DISORDERS PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS
TNXB	100%	100%	100%	98.5%	Ehlers-Danlos syndrome, classic-like, 1, 606408;Vesicoureteral reflux 8, 615963	SKIN DISORDERS PANEL¹ HEMOSTATIC/THROMB OTIC DISORDERS PANEL RENAL DISORDERS PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL¹

TOE1	100%	100%	100%	98.9%	Pontocerebellar hypoplasia, type 7, 614969	MOVEMENT DISORDERS PANEL DISORDERS/DIFFEREN CES OF SEX DEVELOPMENT (DSD) / PRIMARY ADRENAL INSUFFICIENCY PANEL EPILEPSY PANEL INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL¹
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TOGARAM1	100%	100%	100%	99.3%	Joubert syndrome 37, 619185	VISION DISORDERS PANEL CILIOPATHIES PANEL LIVER DISORDERS PANEL INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL¹
TOM1	100%	100%	100%	98.6%	?Immunodeficiency 85 and autoimmunity, 619510	PRIMARY IMMUNODEFICIENCIES PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS
TOMM40L	100%	100%	100%	99.2%		MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS MITOCHONDRIAL DISORDERS PANEL

TOMM7	100%	100%	100%	99.1%	Garg-Mishra progeroid syndrome, 620601	MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS MITOCHONDRIAL DISORDERS PANEL COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹
TOMM70	100%	100%	100%	99.6%		INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS MITOCHONDRIAL DISORDERS PANEL
TONSL	100%	100%	100%	99.4%	Spondyloepimetaphyseal dysplasia, sponastrime type, 271510	SHORT STATURE/SKELETAL DYSPLASIA PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹

TOP1	100%	100%	100%	99.5%	DNA topoisomerase I, camptothecin-resistant	MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS
TOP2A	100%	100%	100%	99.5%	DNA topoisomerase II, resistance to inhibition of, by amsacrine	MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS
TOP2B	100%	100%	100%	99.4%	B-cell immunodeficiency, distal limb anomalies, and urogenital malformations, 609296	PRIMARY IMMUNODEFICIENCIES PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS
TOP3A	100%	100%	100%	99.3%	Microcephaly, growth restriction, and increased sister chromatid exchange 2, 618097;Progressive external ophthalmoplegia with mitochondrial DNA deletions, autosomal recessive 5, 618098	MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS MITOCHONDRIAL DISORDERS PANEL COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹

TOPORS	100%	100%	100%	99.6%	Retinitis pigmentosa 31, 609923	VISION DISORDERS PANEL CILIOPATHIES PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS
TOR1A	93.1%	90.8%	100%	99.2%	{Dystonia-1, modifier of};Arthrogryposis multiplex congenita 5, 618947;Dystonia-1, torsion, 128100	FETAL AKINESIA PANEL MOVEMENT DISORDERS PANEL NEUROLOGICAL PAIN DISORDERS PANEL¹ INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL¹

TOR1AIP1	100%	100%	100%	98.9%	?Muscular dystrophy, autosomal recessive, with rigid spine and distal joint contractures, 617072	HEART DISORDERS PANEL ¹ MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹ MUSCLE DISORDERS PANEL
TP53	94.7%	94.7%	100%	99%	{Basal cell carcinoma 7}, 614740;{Adrenocortical carcinoma, pediatric}, 202300;Hepatocellular carcinoma, somatic, 114550;Breast cancer, somatic, 114480;Li-Fraumeni syndrome, 151623;Pancreatic cancer, somatic, 260350;Nasopharyngeal carcinoma, somatic, 607107;{Osteosarcoma}, 259500;{Choroid plexus papilloma}, 260500;{Colorectal cancer}, 114500;{Glioma susceptibility 1}, 137800;Bone marrow failure syndrome 5, 618165	INHERITED BONE MARROW FAILURE AND/OR PREDISPOSITION PANEL TO HEMATOLOGICAL MALIGNANCIES MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS SONIC HEDGEHOG MEDULLOBLASTOMA PANEL HEREDITARY CANCER PANEL

TP53RK	100%	100%	100%	99.1%	Galloway-Mowat syndrome 4, 617730	INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS RENAL DISORDERS PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹
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TP63	100%	100%	100%	99%	Premature ovarian failure 21, 620311;Ectrodactyly, ectodermal dysplasia, and cleft lip/palate syndrome 3, 604292;Hay-Wells syndrome, 106260;Split-hand/foot malformation 4, 605289;Orofacial cleft 8, 618149;Rapp-Hodgkin syndrome, 129400;ADULT syndrome, 103285;Limb-mammary syndrome, 603543	CRANIOFACIAL ANOMALIES PANEL WITH GENOME WIDE CNV ANALYSIS OROFACIAL CLEFTING PANEL WITH GENOME WIDE CNV ANALYSIS PREMATURE OVARIAN INSUFFICIENCY PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS SHORT STATURE/SKELETAL DYSPLASIA PANEL WITH GENOME WIDE CNV ANALYSIS DISORDERS/DIFFERENCES OF SEX DEVELOPMENT (DSD) / PRIMARY ADRENAL INSUFFICIENCY PANEL SKIN DISORDERS PANEL ¹
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TP73	100%	100%	100%	99.2%	Ciliary dyskinesia, primary, 47, and lissencephaly, 619466	CILIOPATHIES PANEL INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹
TPCN2	100%	100%	100%	98.9%	[Skin/hair/eye pigmentation 10, blond/brown hair], 612267	SKIN DISORDERS PANEL ¹ MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS
TPI1	100%	100%	100%	98.9%	Hemolytic anemia due to triosephosphate isomerase deficiency, 615512	METABOLIC DISORDERS PANEL INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹

TPK1	100%	100%	100%	99.5%	Thiamine metabolism dysfunction syndrome 5 (episodic encephalopathy type), 614458	METABOLIC DISORDERS PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS MITOCHONDRIAL DISORDERS PANEL COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹
TPM1	100%	100%	100%	98.6%	Left ventricular noncompaction 9, 611878;Cardiomyopathy, hypertrophic, 3, 115196;Cardiomyopathy, dilated, 1Y, 611878	DILATED CARDIOMYOPATHY PANEL ¹ HEART DISORDERS PANEL ¹ HYPERTROPHIC CARDIOMYOPATHY PANEL ¹ MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS
TPM2	100%	100%	100%	98.8%	Arthrogryposis, distal, type 2B4, 108120;Arthrogryposis, distal, type 1A, 108120;Congenital myopathy 23, 609285	FETAL AKINESIA PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS MUSCLE DISORDERS PANEL

TPM3	100%	100%	100%	99%	Congenital myopathy 4A, autosomal dominant, 255310;Congenital myopathy 4B, autosomal recessive, 609284	FETAL AKINESIA PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS MUSCLE DISORDERS PANEL COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹
TPM4	100%	99.8%	100%	98.5%	Bleeding disorder, platelet-type, 25, 620486	INHERITED BONE MARROW FAILURE AND/OR PREDISPOSITION PANEL TO HEMATOLOGICAL MALIGNANCIES HEMOSTATIC/THROMB OTIC DISORDERS PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS
TPMT	100%	100%	100%	99.8%	{Thiopurines, poor metabolism of, 1}, 610460	METABOLIC DISORDERS PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS

TPO	100%	100%	100%	99.6%	Thyroid dyshormonogenesis 2A, 274500	INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹
TPP1	100%	100%	100%	99.4%	Ceroid lipofuscinosis, neuronal, 2, 204500;Spinocerebellar ataxia, autosomal recessive 7, 609270	METABOLIC DISORDERS PANEL INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS MOVEMENT DISORDERS PANEL VISION DISORDERS PANEL EPILEPSY PANEL COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹

TPP2	100%	100%	100%	99.6%	Immunodeficiency 78 with autoimmunity and developmental delay, 619220	PRIMARY IMMUNODEFICIENCIES PANEL INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS
TPR	100%	100%	100%	98.7%	?Intellectual developmental disorder, autosomal recessive 79, 620393	MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS
TPRKB	84.1%	81.2%	100%	99.4%	Galloway-Mowat syndrome 5, 617731	INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS RENAL DISORDERS PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹

TPRN	96.2%	92.6%	99.9%	92.7%	Deafness, autosomal recessive 79, 613307	HEARING IMPAIRMENT PANEL (INCLUDING GJB2) MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹
TRA2B	100%	100%	100%	99.4%		INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS
TRAC	100%	100%	100%	99.8%	Immunodeficiency 7, TCR-alpha/beta deficient, 615387	PRIMARY IMMUNODEFICIENCIES PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹

TRAF3	100%	100%	100%	98.9%	{?Encephalopathy, acute, infection-induced (herpes-specific), susceptibility to, 5}, 614849	PRIMARY IMMUNODEFICIENCIES PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS
TRAF3IP1	100%	100%	100%	98.7%	Senior-Loken syndrome 9, 616629	VISION DISORDERS PANEL CILIOPATHIES PANEL SHORT STATURE/SKELETAL DYSPLASIA PANEL WITH GENOME WIDE CNV ANALYSIS LIVER DISORDERS PANEL RENAL DISORDERS PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL¹

TRAF3IP2	99.9%	98.6%	100%	99.6%	?Candidiasis, familial, 8, 615527;{Psoriasis susceptibility 13}, 614070	PRIMARY IMMUNODEFICIENCIES PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS
TRAF6	100%	100%	100%	99.5%		CRANIOFACIAL ANOMALIES PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS
TRAF7	100%	100%	100%	98.8%	Cardiac, facial, and digital anomalies with developmental delay, 618164	SHORT STATURE/SKELETAL DYSPLASIA PANEL WITH GENOME WIDE CNV ANALYSIS INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS

TRAIP	100%	100%	100%	99.1%	Seckel syndrome 9, 616777	SHORT STATURE/SKELETAL DYSPLASIA PANEL WITH GENOME WIDE CNV ANALYSIS INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹
TRAK1	100%	100%	100%	99.3%	Developmental and epileptic encephalopathy 68, 618201	EPILEPSY PANEL METABOLIC DISORDERS PANEL INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹

TRAPPC10	100%	100%	100%	99%	Neurodevelopmental disorder with microcephaly, short stature, and speech delay, 620027	MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹
TRAPPC11	100%	100%	100%	99.6%	Muscular dystrophy, limb-girdle, autosomal recessive 18, 615356	METABOLIC DISORDERS PANEL INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹ MUSCLE DISORDERS PANEL

TRAPPC12	100%	100%	100%	98.4%	Encephalopathy, progressive, early-onset, with brain atrophy and spasticity, 617669	INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹
TRAPPC14	100%	100%	100%	99.1%	?Microcephaly 25, primary, autosomal recessive, 618351	MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹
TRAPPC2	100%	100%	99.6%	71.3%	Spondyloepiphyseal dysplasia tarda, 313400	SHORT STATURE/SKELETAL DYSPLASIA PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS

TRAPPC2L	100%	100%	100%	99%	Encephalopathy, progressive, early-onset, with episodic rhabdomyolysis, 618331	METABOLIC DISORDERS PANEL INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS MITOCHONDRIAL DISORDERS PANEL COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL¹
TRAPPC4	100%	100%	100%	98.7%	Neurodevelopmental disorder with epilepsy, spasticity, and brain atrophy, 618741	INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL¹

TRAPPC6B	100%	100%	100%	99.7%	Neurodevelopmental disorder with microcephaly, epilepsy, and brain atrophy, 617862	INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL¹
TRAPPC9	100%	100%	100%	99.4%	Intellectual developmental disorder, autosomal recessive 13, 613192	METABOLIC DISORDERS PANEL INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL¹

TRDN	100%	100%	100%	99.5%	Cardiac arrhythmia syndrome, with or without skeletal muscle weakness, 615441	HEART DISORDERS PANEL ¹ LONG QT SYNDROME PANEL ¹ MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹ ARRHYTHMIA AND CARDIAC CONDUCTION DISORDERS PANEL ¹ MUSCLE DISORDERS PANEL
TREH	100%	100%	100%	99.7%	Trehalase deficiency, 612119	METABOLIC DISORDERS PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS

TREM2	100%	100%	100%	99.6%	{Alzhieimer disease 17, susceptibility to}, 615080;Polycystic lipomembranous osteodysplasia with sclerosing leukoencephalopathy 2, 618193	MOVEMENT DISORDERS PANEL SHORT STATURE/SKELETAL DYSPLASIA PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL¹
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TREX1	100%	100%	100%	99.8%	Vasculopathy, retinal, with cerebral leukoencephalopathy and systemic manifestations, 192315;Aicardi-Goutieres syndrome 1, dominant and recessive, 225750;{Systemic lupus erythematosus, susceptibility to}, 152700;Chilblain lupus, 610448	MOVEMENT DISORDERS PANEL VISION DISORDERS PANEL SKIN DISORDERS PANEL ¹ EPILEPSY PANEL HEMOSTATIC/THROMB OTIC DISORDERS PANEL PRIMARY IMMUNODEFICIENCIES PANEL INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹
TRH	100%	100%	100%	99.1%	Thyrotropin-releasing hormone deficiency, 275120	MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹

TRHR	100%	100%	100%	99.5%	Hypothyroidism, congenital, nongoitrous, 7, 618573	MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹
TRIM2	96.1%	96.1%	100%	99.3%	Charcot-Marie-Tooth disease, type 2R, 615490	POLYNEUROPATHIES PANEL ¹ MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹
TRIM22	100%	100%	100%	99.6%		PRIMARY IMMUNODEFICIENCIES PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS
TRIM28	100%	100%	100%	99.1%		MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS HEREDITARY CANCER PANEL

TRIM32	100%	100%	100%	99.1%	?Bardet-Biedl syndrome 11, 615988;Muscular dystrophy, limb-girdle, autosomal recessive 8, 254110	VISION DISORDERS PANEL COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹ MUSCLE DISORDERS PANEL CILIOPATHIES PANEL SKIN DISORDERS PANEL ¹ HYPOGONADOTROPIC HYPOGONADISM (KALLMANN) PANEL INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS RENAL DISORDERS PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS
TRIM36	100%	100%	100%	99.4%	?Anencephaly 1, 206500	MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS

TRIM37	98.3%	98.3%	100%	98.9%	Mulibrey nanism, 253250	SKIN DISORDERS PANEL ¹ SHORT STATURE/SKELETAL DYSPLASIA PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹ OROFACIAL CLEFTING PANEL WITH GENOME WIDE CNV ANALYSIS HEREDITARY CANCER PANEL
TRIM44	100%	100%	100%	99.1%	?Aniridia 3, 617142	MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS
TRIM63	100%	100%	100%	99.5%		HEART DISORDERS PANEL ¹ HYPERTROPHIC CARDIOMYOPATHY PANEL ¹ MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS

TRIM71	100%	100%	100%	98.2%	Hydrocephalus, congenital, 4, 618667	MALE INFERTILITY PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS
TRIM8	100%	100%	100%	97.8%	Focal segmental glomerulosclerosis and neurodevelopmental syndrome, 619428	INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS
TRIO	99%	98.5%	100%	99%	Intellectual developmental disorder, autosomal dominant 44, with microcephaly, 617061;Intellectual developmental disorder, autosomal dominant 63, with macrocephaly, 618825	INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS

TRIOBP	100%	100%	100%	98.1%	Deafness, autosomal recessive 28, 609823	HEARING IMPAIRMENT PANEL (INCLUDING GJB2) MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹
TRIP11	100%	100%	100%	99.5%	Odontochondrodysplasia 1, 184260; Achondrogenesis, type IA, 200600	SHORT STATURE/SKELETAL DYSPLASIA PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹
TRIP12	100%	100%	100%	99.5%	Intellectual developmental disorder, autosomal dominant 49, 617752	INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS

TRIP13	100%	100%	100%	99.1%	Oocyte/zygote/embryo maturation arrest 9, 619011;Mosaic variegated aneuploidy syndrome 3, 617598	COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹ HEREDITARY CANCER PANEL UNSUCCESSFUL IVF TREATMENTS, RECURRENT MOLAR PREGNANCIES AND/OR RECURRENT PREGNANCY LOSS PANEL ¹ SHORT STATURE/SKELETAL DYSPLASIA PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS
TRIP4	100%	100%	100%	99.2%	?Muscular dystrophy, congenital, Davignon-Chauveau type, 617066;Spinal muscular atrophy with congenital bone fractures 1, 616866	FETAL AKINESIA PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹ MUSCLE DISORDERS PANEL

TRIT1	100%	100%	100%	99.5%	Combined oxidative phosphorylation deficiency 35, 617873	MOVEMENT DISORDERS PANEL EPILEPSY PANEL COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹ INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS MITOCHONDRIAL DISORDERS PANEL
TRMT1	100%	100%	100%	98.6%	Intellectual developmental disorder, autosomal recessive 68, 618302	INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹

TRMT10A	100%	100%	100%	99.5%	Microcephaly, short stature, and impaired glucose metabolism 1, 616033	INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹
TRMT10C	100%	100%	100%	99.6%	Combined oxidative phosphorylation deficiency 30, 616974	MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS MITOCHONDRIAL DISORDERS PANEL COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹

TRMT5	100%	100%	100%	99.8%	Peripheral neuropathy with variable spasticity, exercise intolerance, and developmental delay, 616539	INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS MITOCHONDRIAL DISORDERS PANEL COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹
TRMU	100%	100%	100%	99.5%	{Deafness, mitochondrial, modifier of}, 580000;Liver failure, transient infantile, 613070	LIVER DISORDERS PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS MITOCHONDRIAL DISORDERS PANEL COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹

TRNT1	91.9%	91.8%	100%	99.5%	Sideroblastic anemia with B-cell immunodeficiency, periodic fevers, and developmental delay, 616084;Retinitis pigmentosa and erythrocytic microcytosis, 616959	VISION DISORDERS PANEL IRON DISORDERS PANEL PRIMARY IMMUNODEFICIENCIES PANEL INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS MITOCHONDRIAL DISORDERS PANEL COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹
TRPA1	100%	100%	100%	99.5%	?Episodic pain syndrome, familial, 1, 615040	NEUROLOGICAL PAIN DISORDERS PANEL ¹ MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS
TRPC3	100%	100%	100%	99.5%	?Spinocerebellar ataxia 41, 616410	MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS

TRPC6	100%	100%	100%	99%	Glomerulosclerosis, focal segmental, 2, 603965	RENAL DISORDERS PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS
TRPM1	100%	100%	100%	99.4%	Night blindness, congenital stationary (complete), 1C, autosomal recessive, 613216	VISION DISORDERS PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹
TRPM3	97.8%	97.8%	100%	99.2%	?Cataract 50 with or without glaucoma, 620253;Neurodevelopmental disorder with hypotonia, dysmorphic facies, and skeletal anomalies, with or without seizures, 620224	MOVEMENT DISORDERS PANEL VISION DISORDERS PANEL EPILEPSY PANEL INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS

TRPM4	100%	100%	100%	98.5%	Progressive familial heart block, type IB, 604559;Erythrokeratoderma variabilis et progressiva 6, 618531	SKIN DISORDERS PANEL ¹ HEART DISORDERS PANEL ¹ MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS
TRPM6	100%	100%	100%	99.4%	Hypomagnesemia 1, intestinal, 602014	EPILEPSY PANEL RENAL DISORDERS PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹
TRPM7	100%	100%	100%	99.6%	{Amyotrophic lateral sclerosis-parkinsonism/dementia complex, susceptibility to}, 105500	NEUROLOGICAL PAIN DISORDERS PANEL ¹ INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS

TRPM8	100%	100%	100%	98.9%		NEUROLOGICAL PAIN DISORDERS PANEL¹ MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS
TRPS1	100%	100%	100%	99.5%	Trichorhinophalangeal syndrome, type III, 190351;Trichorhinophalangeal syndrome, type I, 190350	SKIN DISORDERS PANEL¹ SHORT STATURE/SKELETAL DYSPLASIA PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS
TRPV1	100%	100%	100%	98.1%		NEUROLOGICAL PAIN DISORDERS PANEL¹ MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS

TRPV3	100%	100%	100%	98.7%	?Palmoplantar keratoderma, nonepidermolytic, focal 2, 616400;Olmsted syndrome 1, 614594	SKIN DISORDERS PANEL ¹ NEUROLOGICAL PAIN DISORDERS PANEL ¹ MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS
TRPV4	100%	100%	100%	98.5%	Neuronopathy, distal hereditary motor, autosomal dominant 8, 600175;Spondylometaphyseal dysplasia, Kozlowski type, 184252;Digital arthropathy-brachydactyly, familial, 606835;[Sodium serum level QTL 1], 613508;SED, Maroteaux type, 184095;Metatropic dysplasia, 156530;Scapuloperoneal spinal muscular atrophy, 181405;Hereditary motor and sensory neuropathy, type IIc, 606071;?Avascular necrosis of femoral head, primary, 2, 617383;Parastremmatic dwarfism, 168400;Brachyolmia type 3, 113500	MUSCLE DISORDERS PANEL SHORT STATURE/SKELETAL DYSPLASIA PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS NEUROLOGICAL PAIN DISORDERS PANEL ¹ POLYNEUROPATHIES PANEL ¹ FETAL AKINESIA PANEL

TRPV6	100%	100%	100%	98.9%	Hyperparathyroidism, transient neonatal, 618188	SHORT STATURE/SKELETAL DYSPLASIA PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹
TRRAP	100%	100%	100%	98.8%	?Deafness, autosomal dominant 75, 618778;Developmental delay with or without dysmorphic facies and autism, 618454	HEARING IMPAIRMENT PANEL (INCLUDING GJB2) INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS

TSC1	100%	100%	100%	99.1%	Focal cortical dysplasia, type II, somatic, 607341;Tuberous sclerosis-1, 191100;Lymphangi leiomyomatosis, 606690	CONGENITAL HEARTDISEASE PANEL¹ SKIN DISORDERS PANEL¹ EPILEPSY PANEL HEART DISORDERS PANEL¹ INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS RENAL DISORDERS PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS HEREDITARY CANCER PANEL
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TSC2	100%	100%	100%	99.2%	Lymphangiomyomatosis, somatic, 606690;?Focal cortical dysplasia, type II, somatic, 607341;Tuberous sclerosis-2, 613254	SKIN DISORDERS PANEL ¹ EPILEPSY PANEL INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS RENAL DISORDERS PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS HEREDITARY CANCER PANEL
TSEN15	100%	100%	100%	99.3%	Pontocerebellar hypoplasia, type 2F, 617026	MOVEMENT DISORDERS PANEL EPILEPSY PANEL INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹

TSEN2	88.4%	88.4%	100%	99.3%	Pontocerebellar hypoplasia type 2B, 612389	MOVEMENT DISORDERS PANEL EPILEPSY PANEL INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL¹
TSEN34	100%	100%	100%	97.9%	?Pontocerebellar hypoplasia type 2C, 612390	MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL¹

TSEN54	100%	100%	100%	99%	Pontocerebellar hypoplasia type 2A, 277470;Pontocerebellar hypoplasia type 4, 225753;?Pontocerebellar hypoplasia type 5, 610204	COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹ MOVEMENT DISORDERS PANEL EPILEPSY PANEL INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS
TSFM	94.3%	94.3%	100%	98.7%	Combined oxidative phosphorylation deficiency 3, 610505	VISION DISORDERS PANEL HEART DISORDERS PANEL ¹ INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS MITOCHONDRIAL DISORDERS PANEL COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹

TSGA10	100%	100%	100%	99.6%	?Spermatogenic failure 26, 617961	MALE INFERTILITY PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS
TSHB	100%	100%	100%	99.7%	Hypothyroidism, congenital, nongoitrous 4, 275100	INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹
TSHR	100%	100%	100%	99.3%	Hyperthyroidism, familial gestational, 603373;Hyperthyroidism, nonautoimmune, 609152;Thyroid adenoma, hyperfunctioning, somatic, 609152;Hypothyroidism, congenital, nongoitrous, 1, 275200;Thyroid carcinoma with thyrotoxicosis, somatic, 609152	MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹

TSHZ1	100%	100%	100%	98.8%	Aural atresia, congenital, 607842	CRANIOFACIAL ANOMALIES PANEL WITH GENOME WIDE CNV ANALYSIS HEARING IMPAIRMENT PANEL (INCLUDING GJB2) MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS
TSPAN12	100%	100%	100%	99.3%	Exudative vitreoretinopathy 5, 613310	VISION DISORDERS PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹
TSPAN7	100%	99.6%	98.6%	65.8%	Intellectual developmental disorder, X-linked 58, 300210	INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS

TSPEAR	100%	100%	100%	98.4%	Tooth agenesis, selective, 10, 620173;?Deafness, autosomal recessive 98, 614861;Ectodermal dysplasia 14, hair/tooth type with or without hypohidrosis, 618180	CRANIOFACIAL ANOMALIES PANEL WITH GENOME WIDE CNV ANALYSIS SKIN DISORDERS PANEL ¹ MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹
TSPOAP1	100%	100%	100%	98.9%	Dystonia 22, juvenile-onset, 620453;?Dystonia 22, adult-onset, 620456	MOVEMENT DISORDERS PANEL INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS

TSPYL1	100%	100%	100%	99.2%	Sudden infant death with dysgenesis of the testes syndrome, 608800	DISORDERS/DIFFERENCES OF SEX DEVELOPMENT (DSD) / PRIMARY ADRENAL INSUFFICIENCY PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹
TSR2	100%	99.9%	98.4%	69.2%	?Diamond-Blackfan anemia 14 with mandibulofacial dysostosis, 300946	INHERITED BONE MARROW FAILURE AND/OR PREDISPOSITION PANEL TO HEMATOLOGICAL MALIGNANCIES MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS
TTBK2	100%	100%	100%	99.4%	Spinocerebellar ataxia 11, 604432	MOVEMENT DISORDERS PANEL CILIOPATHIES PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS

TTC12	100%	100%	100%	99.6%	Ciliary dyskinesia, primary, 45, 618801	CILIOPATHIES PANEL MALE INFERTILITY PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹
TTC19	100%	100%	100%	99.4%	Mitochondrial complex III deficiency, nuclear type 2, 615157	MOVEMENT DISORDERS PANEL INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS MITOCHONDRIAL DISORDERS PANEL COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹ MUSCLE DISORDERS PANEL

TTC21A	100%	100%	100%	99.5%	Spermatogenic failure 37, 618429	MALE INFERTILITY PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS
TTC21B	98.3%	98.1%	100%	99.5%	Short-rib thoracic dysplasia 4 with or without polydactyly, 613819;Nephronophthisis 12, 613820	CILIOPATHIES PANEL SHORT STATURE/SKELETAL DYSPLASIA PANEL WITH GENOME WIDE CNV ANALYSIS RENAL DISORDERS PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹
TTC29	100%	100%	100%	99.5%	Spermatogenic failure 42, 618745	MALE INFERTILITY PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS

TTC5	100%	100%	100%	99.6%	Neurodevelopmental disorder with cerebral atrophy and variable facial dysmorphism, 619244	INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹
TTC7A	100%	100%	100%	99.3%	Gastrointestinal defects and immunodeficiency syndrome, 243150	PRIMARY IMMUNODEFICIENCIES PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹ SEVERE COMBINED IMMUNODEFICIENCY (SCID) PANEL

TTC8	100%	100%	100%	99.6%	Bardet-Biedl syndrome 8, 615985;?Retinitis pigmentosa 51, 613464	VISION DISORDERS PANEL CILIOPATHIES PANEL HYPOGONADOTROPIC HYPOGONADISM (KALLMANN) PANEL INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS RENAL DISORDERS PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹
TTI1	100%	100%	100%	99%	Neurodevelopmental disorder with microcephaly and movement abnormalities, 620445	INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹

TTI2	100%	100%	100%	98.9%	Intellectual developmental disorder, autosomal recessive 39, 615541	SKIN DISORDERS PANEL ¹ SHORT STATURE/SKELETAL DYSPLASIA PANEL WITH GENOME WIDE CNV ANALYSIS INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹
TTLL5	100%	100%	100%	99.3%	Cone-rod dystrophy 19, 615860	VISION DISORDERS PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹

TTN	100%	100%	100%	99.5%	Muscular dystrophy, limb-girdle, autosomal recessive 10, 608807;Cardiomyopathy, familial hypertrophic, 9, 613765;Congenital myopathy 5 with cardiomyopathy, 611705;Tibial muscular dystrophy, tardive, 600334;Cardiomyopathy, dilated, 1G, 604145;Myopathy, myofibrillar, 9, with early respiratory failure, 603689	FETAL AKINESIA PANEL DILATED CARDIOMYOPATHY PANEL ¹ MUSCLE DISORDERS PANEL HEART DISORDERS PANEL ¹ MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS
TTPA	100%	100%	100%	99.2%	Ataxia with isolated vitamin E deficiency, 277460	MOVEMENT DISORDERS PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹

TTR	100%	100%	100%	99.4%	Amyloidosis, hereditary, transthyretin-related, 105210;Carpal tunnel syndrome, familial, 115430;[Dystransthyretinemic hyperthyroxinemia], 145680	HEART DISORDERS PANEL ¹ MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS SMALL FIBRE NEUROPATHY PANEL ¹ HYPERTROPHIC CARDIOMYOPATHY PANEL ¹ POLYNEUROPATHIES PANEL ¹ NEUROLOGICAL PAIN DISORDERS PANEL ¹
TUB	100%	100%	100%	99.1%	?Retinal dystrophy and obesity, 616188	VISION DISORDERS PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹

TUBA1A	100%	100%	100%	99.2%	Lissencephaly 3, 611603	FETAL AKINESIA PANEL MOVEMENT DISORDERS PANEL VISION DISORDERS PANEL EPILEPSY PANEL INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS
TUBA3D	100%	99.8%	99.9%	98.5%	Keratoconus 9, 617928	VISION DISORDERS PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS
TUBA4A	100%	100%	100%	99%	Amyotrophic lateral sclerosis 22 with or without frontotemporal dementia, 616208	AMYOTROPHIC LATERAL SCLEROSIS, ALS PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS

TUBA8	95.2%	95.1%	99.9%	99.3%	Macrothrombocytopenia, isolated, 2, autosomal dominant, 619840	INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹
TUBB	99.9%	99%	100%	99.5%	Symmetric circumferential skin creases, congenital, 1, 156610;Cortical dysplasia, complex, with other brain malformations 6, 615771	MOVEMENT DISORDERS PANEL INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS OROFACIAL CLEFTING PANEL WITH GENOME WIDE CNV ANALYSIS

TUBB1	100%	100%	100%	99.5%	Macrothrombocytopenia, isolated, 1, autosomal dominant, 613112	INHERITED BONE MARROW FAILURE AND/OR PREDISPOSITION PANEL TO HEMATOLOGICAL MALIGNANCIES HEMOSTATIC/THROMBOTIC DISORDERS PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS
TUBB2A	100%	100%	100%	97.9%	Cortical dysplasia, complex, with other brain malformations 5, 615763	EPILEPSY PANEL POLYNEUROPATHIES PANEL ¹ INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS

TUBB2B	100%	100%	100%	99.2%	Cortical dysplasia, complex, with other brain malformations 7, 610031	FETAL AKINESIA PANEL EPILEPSY PANEL INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS
TUBB3	100%	100%	100%	99.3%	Fibrosis of extraocular muscles, congenital, 3A, 600638;Cortical dysplasia, complex, with other brain malformations 1, 614039	VISION DISORDERS PANEL POLYNEUROPATHIES PANEL ¹ INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS MUSCLE DISORDERS PANEL

TUBB4A	99.3%	96.6%	100%	98.3%	Dystonia 4, torsion, autosomal dominant, 128101;Leukodystrophy, hypomyelinating, 6, 612438	MOVEMENT DISORDERS PANEL EPILEPSY PANEL INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS
TUBB4B	100%	100%	100%	98.8%	Leber congenital amaurosis with early-onset deafness, 617879	VISION DISORDERS PANEL HEARING IMPAIRMENT PANEL (INCLUDING GJB2) MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS
TUBB6	100%	100%	100%	98.7%	?Facial palsy, congenital, with ptosis and velopharyngeal dysfunction, 617732	MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS

TUBB8	100%	100%	100%	99.7%	Oocyte/zygote/embryo maturation arrest 2, 616780	UNSUCCESSFUL IVF TREATMENTS, RECURRENT MOLAR PREGNANCIES AND/OR RECURRENT PREGNANCY LOSS PANEL ¹ MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS
TUBG1	100%	100%	100%	99.1%	Cortical dysplasia, complex, with other brain malformations 4, 615412	MOVEMENT DISORDERS PANEL EPILEPSY PANEL INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS

TUBGCP2	97%	97%	100%	99.1%	Pachygyria, microcephaly, developmental delay, and dysmorphic facies, with or without seizures, 618737	INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹
TUBGCP4	100%	99.8%	99.5%	96.3%	Microcephaly and chorioretinopathy, autosomal recessive, 3, 616335	VISION DISORDERS PANEL INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹

TUBGCP6	100%	100%	100%	99%	Microcephaly and chorioretinopathy, autosomal recessive, 1, 251270	VISION DISORDERS PANEL INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹
TUFM	100%	100%	100%	98.8%	Combined oxidative phosphorylation deficiency 4, 610678	MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS MITOCHONDRIAL DISORDERS PANEL COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹
TUFT1	100%	100%	100%	98.7%	Woolly hair-skin fragility syndrome, 620415	SKIN DISORDERS PANEL ¹ MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS

TULP1	100%	100%	100%	98%	Leber congenital amaurosis 15, 613843;Retinitis pigmentosa 14, 600132	VISION DISORDERS PANEL CILIOPATHIES PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹
TULP3	100%	100%	100%	98.9%	Hepatorenocardiac degenerative fibrosis, 619902	HEART DISORDERS PANEL ¹ LIVER DISORDERS PANEL RENAL DISORDERS PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹

TUSC3	100%	100%	100%	99.4%	Intellectual developmental disorder, autosomal recessive 7, 611093	METABOLIC DISORDERS PANEL INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹
TWIST1	99.9%	98.9%	100%	98.8%	Craniosynostosis 1, 123100;Robinow-Sorauf syndrome, 180750;Sweeney-Cox syndrome, 617746;Saethre-Chotzen syndrome with or without eyelid anomalies, 101400	CRANIOFACIAL ANOMALIES PANEL WITH GENOME WIDE CNV ANALYSIS OROFACIAL CLEFTING PANEL WITH GENOME WIDE CNV ANALYSIS SHORT STATURE/SKELETAL DYSPLASIA PANEL WITH GENOME WIDE CNV ANALYSIS INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS

TWIST2	100%	100%	100%	94.2%	Ablepharon-macrostomia syndrome, 200110;Barber-Say syndrome, 209885;Focal facial dermal dysplasia 3, Setleis type, 227260	SKIN DISORDERS PANEL ¹ MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹
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TWNK	100%	100%	100%	99.6%	Mitochondrial DNA depletion syndrome 7 (hepatocerebral type), 271245;Progressive external ophthalmoplegia with mitochondrial DNA deletions, autosomal dominant 3, 609286;Perrault syndrome 5, 616138	MOVEMENT DISORDERS PANEL COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹ PREMATURE OVARIAN INSUFFICIENCY PANEL MUSCLE DISORDERS PANEL POLYNEUROPATHIES PANEL ¹ LIVER DISORDERS PANEL INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS MITOCHONDRIAL DISORDERS PANEL VISION DISORDERS PANEL HEARING IMPAIRMENT PANEL (INCLUDING GJB2) DISORDERS/DIFFEREN CES OF SEX DEVELOPMENT (DSD) / PRIMARY ADRENAL INSUFFICIENCY PANEL
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TXN2	100%	100%	100%	99.2%	?Combined oxidative phosphorylation deficiency 29, 616811	MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS MITOCHONDRIAL DISORDERS PANEL
TXNDC15	100%	100%	100%	98.7%	Meckel syndrome 14, 619879	CILIOPATHIES PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹
TXNL4A	100%	100%	100%	99.6%	Burn-McKeown syndrome, 608572	MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹ OROFACIAL CLEFTING PANEL WITH GENOME WIDE CNV ANALYSIS

TXNRD2	100%	100%	100%	98.8%	?Glucocorticoid deficiency 5, 617825	DISORDERS/DIFFERENCES OF SEX DEVELOPMENT (DSD) / PRIMARY ADRENAL INSUFFICIENCY PANEL HEART DISORDERS PANEL ¹ MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS
TYK2	100%	100%	100%	98.4%	Immunodeficiency 35, 611521	INHERITED BONE MARROW FAILURE AND/OR PREDISPOSITION PANEL TO HEMATOLOGICAL MALIGNANCIES PRIMARY IMMUNODEFICIENCIES PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹

TYMP	100%	100%	100%	99%	Mitochondrial DNA depletion syndrome 1 (MNGIE type), 603041	POLYNEUROPATHIES PANEL ¹ LIVER DISORDERS PANEL METABOLIC DISORDERS PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS MITOCHONDRIAL DISORDERS PANEL COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹
TYMS	100%	100%	100%	99.5%	Dyskeratosis congenita, digenic, 620040	METABOLIC DISORDERS PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS

TYR	100%	100%	100%	99.4%	[Skin/hair/eye pigmentation 3, light/dark/freckling skin], 601800;[Skin/hair/eye pigmentation 3, blue/green eyes], 601800;{Melanoma, cutaneous malignant, susceptibility to, 8}, 601800;Albinism, oculocutaneous, type IB, 606952;Albinism, oculocutaneous, type IA, 203100	VISION DISORDERS PANEL SKIN DISORDERS PANEL ¹ HEARING IMPAIRMENT PANEL (INCLUDING GJB2) METABOLIC DISORDERS PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹
TYROBP	100%	100%	100%	99%	Polycystic lipomembranous osteodysplasia with sclerosing leukoencephalopathy 1, 221770	MOVEMENT DISORDERS PANEL SHORT STATURE/SKELETAL DYSPLASIA PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹

TYRP1	100%	100%	100%	99.6%	[Skin/hair/eye pigmentation, variation in, 11 (Melanesian blond hair)], 612271;Albinism, oculocutaneous, type III, 203290	VISION DISORDERS PANEL COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹ MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS SKIN DISORDERS PANEL ¹ METABOLIC DISORDERS PANEL
U2AF2	100%	99.9%	99.9%	97.6%	Developmental delay, dysmorphic facies, and brain anomalies, 620535	INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS

UBA1	100%	99.7%	98.9%	68%	Spinal muscular atrophy, X-linked 2, infantile, 301830;VEXAS syndrome, somatic, 301054	FETAL AKINESIA PANEL INHERITED BONE MARROW FAILURE AND/OR PREDISPOSITION PANEL TO HEMATOLOGICAL MALIGNANCIES PRIMARY IMMUNODEFICIENCIES PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS MUSCLE DISORDERS PANEL
UBA2	100%	100%	100%	99.3%	ACCES syndrome, 619959	SHORT STATURE/SKELETAL DYSPLASIA PANEL WITH GENOME WIDE CNV ANALYSIS INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS

UBA5	99.6%	97.7%	100%	99.2%	?Spinocerebellar ataxia, autosomal recessive 24, 617133;Developmental and epileptic encephalopathy 44, 617132	MOVEMENT DISORDERS PANEL EPILEPSY PANEL POLYNEUROPATHIES PANEL ¹ INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹
UBAP1	100%	100%	100%	98.5%	Spastic paraplegia 80, autosomal dominant, 618418	MOVEMENT DISORDERS PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS
UBAP1L	100%	100%	100%	99.1%		VISION DISORDERS PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS

UBAP2L	100%	100%	100%	99.1%	Neurodevelopmental disorder with impaired language, behavioral abnormalities, and dysmorphic facies, 620494	INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS
UBB	100%	100%	100%	99.3%		CRANIOFACIAL ANOMALIES PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS
UBE2A	95.1%	89.6%	99.1%	69.4%	Intellectual developmental disorder, X-linked syndromic, Nascimento type, 300860	SKIN DISORDERS PANEL ¹ INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS

UBE2T	100%	100%	100%	98.7%	Fanconi anemia, complementation group T, 616435	INHERITED BONE MARROW FAILURE AND/OR PREDISPOSITION PANEL TO HEMATOLOGICAL MALIGNANCIES SHORT STATURE/SKELETAL DYSPLASIA PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹
UBE3A	100%	100%	100%	99.7%	Angelman syndrome, 105830	EPILEPSY PANEL INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS

UBE3B	100%	100%	100%	99.3%	Kaufman oculocerebrofacial syndrome, 244450	INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹
UBE3C	100%	100%	100%	99.2%	Neurodevelopmental disorder with absent speech and movement and behavioral abnormalities, 620270	INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹
UBE4A	100%	100%	100%	98.8%	Neurodevelopmental disorder with hypotonia and gross motor and speech delay, 619639	INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS

UBIAD1	100%	100%	100%	99.9%	Corneal dystrophy, Schnyder type, 121800	VISION DISORDERS PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS
UBQLN2	100%	100%	99.4%	73%	Amyotrophic lateral sclerosis 15, with or without frontotemporal dementia, 300857	AMYOTROPHIC LATERAL SCLEROSIS, ALS PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS
UBR1	98%	98%	100%	99.3%	Johanson-Blizzard syndrome, 243800	COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹ INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS SKIN DISORDERS PANEL ¹ LIVER DISORDERS PANEL

UBR2	100%	99.8%	99.7%	98.8%		MALE INFERTILITY PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS
UBR5	100%	100%	100%	99.6%		INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS
UBR7	100%	100%	100%	99.5%	Li-Campeau syndrome, 619189	INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹

UBTF	100%	100%	100%	98.6%	Neurodegeneration, childhood-onset, with brain atrophy, 617672	MOVEMENT DISORDERS PANEL EPILEPSY PANEL INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS
UCHL1	100%	100%	100%	99.7%	{?Parkinson disease 5, susceptibility to}, 613643;Spastic paraplegia 79A, autosomal dominant, 620221;Spastic paraplegia 79B, autosomal recessive, 615491	MOVEMENT DISORDERS PANEL VISION DISORDERS PANEL POLYNEUROPATHIES PANEL ¹ MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS MITOCHONDRIAL DISORDERS PANEL COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹

UFC1	100%	100%	100%	99%	Neurodevelopmental disorder with spasticity and poor growth, 618076	INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL¹
UFM1	100%	100%	100%	99.2%	Leukodystrophy, hypomyelinating, 14, 617899	METABOLIC DISORDERS PANEL INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS MITOCHONDRIAL DISORDERS PANEL COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL¹

UFSP2	100%	100%	100%	99.6%	?Hip dysplasia, Beukes type, 142669;Spondyloepimetaphyseal dysplasia, Di Rocco type, 617974;Developmental and epileptic encephalopathy 106, 620028	SHORT STATURE/SKELETAL DYSPLASIA PANEL WITH GENOME WIDE CNV ANALYSIS INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS
UGCG	100%	100%	100%	99.2%		SKIN DISORDERS PANEL ¹ MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS
UGDH	100%	100%	100%	99.4%	Developmental and epileptic encephalopathy 84, 618792	EPILEPSY PANEL INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹

UGGT1	100%	100%	100%	99.4%		METABOLIC DISORDERS PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS
UGP2	98.8%	96.8%	100%	99.4%	Developmental and epileptic encephalopathy 83, 618744	EPILEPSY PANEL INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS
UGT1A1	100%	100%	100%	98.8%	Crigler-Najjar syndrome, type I, 218800;[Bilirubin, serum level of, QTL1], 601816;Hyperbilirubinemia, familial transient neonatal, 237900;Crigler-Najjar syndrome, type II, 606785;[Gilbert syndrome], 143500	MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹ LIVER DISORDERS PANEL METABOLIC DISORDERS PANEL

UMOD	100%	100%	100%	99.3%	Tubulointerstitial kidney disease, autosomal dominant, 1, 162000	RENAL DISORDERS PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS
UMPS	100%	100%	100%	99.6%	Orotic aciduria, 258900	METABOLIC DISORDERS PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹
UNC119	100%	100%	100%	97.4%	Cone-rod dystrophy 24, 620342;?Immunodeficiency 13, 615518	VISION DISORDERS PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS
UNC13A	100%	100%	100%	98.6%		INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS

UNC13D	100%	100%	100%	99.5%	Hemophagocytic lymphohistiocytosis, familial, 3, 608898	PRIMARY IMMUNODEFICIENCIES PANEL LIVER DISORDERS PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹
UNC45A	100%	100%	100%	99.3%	Osteotohepatoenteric syndrome, 619377	LIVER DISORDERS PANEL INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS
UNC45B	100%	100%	100%	98.8%	?Cataract 43, 616279;Myofibrillar myopathy 11, 619178	VISION DISORDERS PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS MUSCLE DISORDERS PANEL

UNC79	100%	100%	100%	99.4%		INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS
UNC80	100%	100%	100%	99.4%	Hypotonia, infantile, with psychomotor retardation and characteristic facies 2, 616801	INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹
UNC93B1	99.9%	99.6%	99.9%	94.7%	{Encephalopathy, acute, infection-induced (herpes-specific), susceptibility to, 1}, 610551	PRIMARY IMMUNODEFICIENCIES PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS

UNG	96.5%	96.5%	100%	99.1%	Immunodeficiency with hyper IgM, type 5, 608106	PRIMARY IMMUNODEFICIENCIES PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹
UPB1	100%	100%	100%	99%	Beta-ureidopropionase deficiency, 613161	METABOLIC DISORDERS PANEL INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹
UPF1	99.2%	98.1%	100%	99%		INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS

UPF3B	100%	99.8%	98.6%	66.9%	Intellectual developmental disorder, X-linked syndromic 14, 300676	INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS
UPK3A	100%	100%	100%	98.6%		RENAL DISORDERS PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS
UQCC1	100%	100%	100%	99.5%		MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS MITOCHONDRIAL DISORDERS PANEL

UQCC2	100%	100%	100%	99.9%	Mitochondrial complex III deficiency, nuclear type 7, 615824	RENAL DISORDERS PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS MITOCHONDRIAL DISORDERS PANEL COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹
UQCC3	100%	100%	100%	99.5%	?Mitochondrial complex III deficiency, nuclear type 9, 616111	MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS MITOCHONDRIAL DISORDERS PANEL COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹
UQCR10	100%	100%	100%	97.8%		MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS MITOCHONDRIAL DISORDERS PANEL

UQCR11	100%	100%	100%	98.4%		MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS MITOCHONDRIAL DISORDERS PANEL
UQCRB	100%	100%	100%	99.3%	Mitochondrial complex III deficiency, nuclear type 3, 615158	MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS MITOCHONDRIAL DISORDERS PANEL COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹
UQCRC1	100%	100%	100%	99.1%	Parkinsonism with polyneuropathy, 619279	POLYNEUROPATHIES PANEL ¹ MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS MITOCHONDRIAL DISORDERS PANEL

UQCRC2	100%	100%	100%	99.6%	Mitochondrial complex III deficiency, nuclear type 5, 615160	MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS MITOCHONDRIAL DISORDERS PANEL COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹
UQCRFS1	100%	100%	100%	99.5%	Mitochondrial complex III deficiency, nuclear type 10, 618775	MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS MITOCHONDRIAL DISORDERS PANEL COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹
UQCRH	100%	100%	100%	99.7%	?Mitochondrial complex III deficiency, nuclear type 11, 620137	MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS MITOCHONDRIAL DISORDERS PANEL

UQCRQ	100%	100%	100%	99.8%	Mitochondrial complex III deficiency, nuclear type 4, 615159	MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS MITOCHONDRIAL DISORDERS PANEL COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL¹
UROCI	100%	100%	100%	99.4%	?Urocanase deficiency, 276880	METABOLIC DISORDERS PANEL INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL¹

UROD	100%	100%	100%	99.5%	Porphyria, hepatoerythropoietic, 176100;Porphyria cutanea tarda, 176100	SKIN DISORDERS PANEL ¹ METABOLIC DISORDERS PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹
UROS	100%	100%	100%	99.9%	Porphyria, congenital erythropoietic, 263700	SKIN DISORDERS PANEL ¹ IRON DISORDERS PANEL METABOLIC DISORDERS PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹

USB1	95%	93.2%	100%	99%	Poikiloderma with neutropenia, 604173	INHERITED BONE MARROW FAILURE AND/OR PREDISPOSITION PANEL TO HEMATOLOGICAL MALIGNANCIES SKIN DISORDERS PANEL ¹ DYSKERATOSIS CONGENITA AND APLASTIC ANEMIA PANEL PRIMARY IMMUNODEFICIENCIES PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹ HEREDITARY CANCER PANEL
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USH1C	100%	100%	100%	98.8%	Usher syndrome, type 1C, 276904;Deafness, autosomal recessive 18A, 602092	VISION DISORDERS PANEL COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL¹ MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS HEARING IMPAIRMENT PANEL (INCLUDING GJB2)
USH1G	100%	100%	100%	98.8%	Usher syndrome, type 1G, 606943	VISION DISORDERS PANEL HEARING IMPAIRMENT PANEL (INCLUDING GJB2) MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL¹

USH2A	100%	100%	100%	99.2%	Usher syndrome, type 2A, 276901;Retinitis pigmentosa 39, 613809	VISION DISORDERS PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹ HEARING IMPAIRMENT PANEL (INCLUDING GJB2)
USP14	100%	100%	100%	99.6%		INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS
USP18	100%	100%	100%	98.8%	Pseudo-TORCH syndrome 2, 617397	PRIMARY IMMUNODEFICIENCIES PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹

USP26	100%	100%	99.5%	72.3%	Spermatogenic failure, X-linked, 6, 301101	MALE INFERTILITY PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS
USP27X	100%	100%	98.3%	68.2%	Intellectual developmental disorder, X-linked 105, 300984	INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS
USP45	100%	100%	100%	99.5%	?Leber congenital amaurosis 19, 618513	VISION DISORDERS PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹
USP48	100%	100%	100%	99.4%	Deafness, autosomal dominant 85, 620227	HEARING IMPAIRMENT PANEL (INCLUDING GJB2) MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS

USP53	100%	100%	100%	99.3%	Cholestasis, progressive familial intrahepatic, 7, with or without hearing loss, 619658	LIVER DISORDERS PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹
USP7	100%	99.7%	100%	98.7%	Hao-Fountain syndrome, 616863	INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS
USP8	100%	100%	100%	99.3%	Pituitary adenoma 4, ACTH-secreting, somatic, 219090	MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS

USP9X	100%	100%	99.1%	70.4%	Intellectual developmental disorder, X-linked 99, 300919;Intellectual developmental disorder, X-linked 99, syndromic, female-restricted, 300968	INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS OROFACIAL CLEFTING PANEL WITH GENOME WIDE CNV ANALYSIS
USP9Y	50%	49.8%	49%	21.2%	Spermatogenic failure, Y-linked, 2, 415000	MALE INFERTILITY PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS
UVSSA	100%	100%	100%	99.1%	UV-sensitive syndrome 3, 614640	SKIN DISORDERS PANEL ¹ MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹

VAC14	100%	100%	100%	99.6%	Striatonigral degeneration, childhood-onset, 617054	MOVEMENT DISORDERS PANEL SHORT STATURE/SKELETAL DYSPLASIA PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹
VAMP1	100%	100%	100%	98.6%	Myasthenic syndrome, congenital, 25, 618323;Spastic ataxia 1, autosomal dominant, 108600	MOVEMENT DISORDERS PANEL INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹ MUSCLE DISORDERS PANEL FETAL AKINESIA PANEL

VAMP2	100%	100%	100%	98.1%	Neurodevelopmental disorder with hypotonia and autistic features with or without hyperkinetic movements, 618760	INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS
VANGL1	100%	100%	100%	99.2%	{Neural tube defects, susceptibility to}, 182940;Caudal regression syndrome, 600145	MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS
VANGL2	100%	99.9%	100%	98.1%	Neural tube defects, 182940	SKIN DISORDERS PANEL ¹ MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS
VAPB	100%	100%	100%	99.6%	Spinal muscular atrophy, late-onset, Finkel type, 182980;Amyotrophic lateral sclerosis 8, 608627	AMYOTROPHIC LATERAL SCLEROSIS, ALS PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS

VAR1	100%	100%	100%	98.6%	Neurodevelopmental disorder with microcephaly, seizures, and cortical atrophy, 617802	INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL¹
VAR2	100%	100%	100%	98.7%	Combined oxidative phosphorylation deficiency 20, 615917	MOVEMENT DISORDERS PANEL INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS MITOCHONDRIAL DISORDERS PANEL COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL¹

VAV1	98.3%	98.3%	100%	98%		PRIMARY IMMUNODEFICIENCIES PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS
VAX1	99.4%	96.4%	100%	98.5%	?Microphthalmia, syndromic 11, 614402	VISION DISORDERS PANEL CRANIOFACIAL ANOMALIES PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹
VCAN	100%	100%	100%	99.4%	Wagner syndrome 1, 143200	VISION DISORDERS PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS

VCL	100%	100%	100%	99.3%	Cardiomyopathy, dilated, 1W, 611407;Cardiomyopathy, hypertrophic, 15, 613255	DILATED CARDIOMYOPATHY PANEL ¹ HEART DISORDERS PANEL ¹ MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS
VCP	100%	100%	100%	99.4%	Frontotemporal dementia and/or amyotrophic lateral sclerosis 6, 613954;Charcot-Marie-Tooth disease, type 2Y, 616687;Inclusion body myopathy with early-onset Paget disease and frontotemporal dementia 1, 167320	INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS MUSCLE DISORDERS PANEL POLYNEUROPATHIES PANEL ¹ AMYOTROPHIC LATERAL SCLEROSIS, ALS PANEL MOVEMENT DISORDERS PANEL

VDR	100%	100%	100%	98.9%	Rickets, vitamin D-resistant, type IIA, 277440	SKIN DISORDERS PANEL ¹ SHORT STATURE/SKELETAL DYSPLASIA PANEL WITH GENOME WIDE CNV ANALYSIS RENAL DISORDERS PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹
VEGFC	100%	100%	100%	98.7%	Lymphatic malformation 4, 615907	SKIN DISORDERS PANEL ¹ LYMPHATIC ANOMALIES PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS
VEZF1	100%	100%	100%	98.5%	?Cardiomyopathy, dilated, 100, 620247	HEART DISORDERS PANEL ¹ MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS

VHL	88%	87.7%	100%	98.3%	Hemangioblastoma, cerebellar, somatic;Erythrocytosis, familial, 2, 263400;von Hippel-Lindau syndrome, 193300;Renal cell carcinoma, somatic, 144700;Pheochromocytoma, 171300	VISION DISORDERS PANEL CILIOPATHIES PANEL SKIN DISORDERS PANEL ¹ MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹ PANEL HEREDITARY RENALCANCER HEREDITARY CANCER PANEL
VIM	100%	100%	100%	99%	Cataract 30, pulverulent, 116300	VISION DISORDERS PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS

VIPAS39	100%	100%	100%	99.4%	Arthrogryposis, renal dysfunction, and cholestasis 2, 613404	FETAL AKINESIA PANEL SKIN DISORDERS PANEL ¹ HEMOSTATIC/THROMB OTIC DISORDERS PANEL LIVER DISORDERS PANEL INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS RENAL DISORDERS PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹ MUSCLE DISORDERS PANEL
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VKORC1	98.7%	94.5%	100%	98.2%	Vitamin K-dependent clotting factors, combined deficiency of, 2, 607473;Warfarin resistance, 122700	HEMOSTATIC/THROMBOTIC DISORDERS PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹
VLDLR	100%	100%	100%	99.3%	Cerebellar hypoplasia, impaired intellectual development, and dysequilibrium syndrome 1, 224050	MOVEMENT DISORDERS PANEL INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹
VMA21	100%	100%	98.7%	66.1%	Myopathy, X-linked, with excessive autophagy, 310440	METABOLIC DISORDERS PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS MUSCLE DISORDERS PANEL

VPS11	100%	100%	100%	98.7%	?Dystonia 32, 619637;Leukodystrophy, hypomyelinating, 12, 616683	MOVEMENT DISORDERS PANEL EPILEPSY PANEL INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL¹
VPS13A	100%	100%	100%	99.6%	Choreoacanthocytosis, 200150	MOVEMENT DISORDERS PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL¹

VPS13B	100%	100%	100%	99.5%	Cohen syndrome, 216550	METABOLIC DISORDERS PANEL INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL¹ VISION DISORDERS PANEL INHERITED BONE MARROW FAILURE AND/OR PREDISPOSITION PANEL TO HEMATOLOGICAL MALIGNANCIES SKIN DISORDERS PANEL¹ PRIMARY IMMUNODEFICIENCIES PANEL
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VPS13C	100%	100%	100%	99.5%	Parkinson disease 23, autosomal recessive, early onset, 616840	MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS PARKINSON DISEASE PANEL COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹
VPS13D	100%	100%	100%	99.4%	Spinocerebellar ataxia, autosomal recessive 4, 607317	MOVEMENT DISORDERS PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS MITOCHONDRIAL DISORDERS PANEL COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹
VPS16	100%	100%	100%	99.3%	Dystonia 30, 619291	MOVEMENT DISORDERS PANEL INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS

VPS33A	89.5%	89.5%	100%	98.9%	Mucopolysaccharidosis-plus syndrome, 617303	SHORT STATURE/SKELETAL DYSPLASIA PANEL WITH GENOME WIDE CNV ANALYSIS METABOLIC DISORDERS PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹
VPS33B	100%	100%	100%	98.6%	Keratoderma-ichthyosis-deafness syndrome, autosomal recessive, 620009;Cholestasis, progressive familial intrahepatic, 12, 620010;Arthrogryposis, renal dysfunction, and cholestasis 1, 208085	FETAL AKINESIA PANEL SKIN DISORDERS PANEL ¹ HEMOSTATIC/THROMB OTIC DISORDERS PANEL LIVER DISORDERS PANEL RENAL DISORDERS PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹

VPS35	100%	100%	100%	99.7%	{Parkinson disease 17}, 614203	MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS PARKINSON DISEASE PANEL
VPS35L	100%	100%	100%	99.4%	Ritscher-Schinzel syndrome 3, 619135	SHORT STATURE/SKELETAL DYSPLASIA PANEL WITH GENOME WIDE CNV ANALYSIS INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS
VPS37A	100%	100%	100%	99.4%	Spastic paraplegia 53, autosomal recessive, 614898	MOVEMENT DISORDERS PANEL INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹

VPS41	100%	100%	100%	99.5%	Spinocerebellar ataxia, autosomal recessive 29, 619389	MOVEMENT DISORDERS PANEL INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹
VPS45	87.7%	86.8%	100%	99.2%	Neutropenia, severe congenital, 5, autosomal recessive, 615285	INHERITED BONE MARROW FAILURE AND/OR PREDISPOSITION PANEL TO HEMATOLOGICAL MALIGNANCIES PRIMARY IMMUNODEFICIENCIES PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹

VPS4A	100%	100%	100%	97.6%	CIMDAG syndrome, 619273	INHERITED BONE MARROW FAILURE AND/OR PREDISPOSITION PANEL TO HEMATOLOGICAL MALIGNANCIES INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS
VPS50	100%	100%	100%	99.6%	Neurodevelopmental disorder with microcephaly, seizures, and neonatal cholestasis, 619685	LIVER DISORDERS PANEL INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹

VPS51	100%	100%	100%	98.7%	Pontocerebellar hypoplasia, type 13, 618606	MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹
VPS53	83.1%	81.4%	100%	99.2%	Pontocerebellar hypoplasia, type 2E, 615851	MOVEMENT DISORDERS PANEL EPILEPSY PANEL INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹

VRK1	99.5%	98.1%	100%	99.6%	Pontocerebellar hypoplasia type 1A, 607596;Neuronopathy, distal hereditary motor, autosomal recessive 10, 620542	MOVEMENT DISORDERS PANEL POLYNEUROPATHIES PANEL ¹ INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹ MUSCLE DISORDERS PANEL
VSX1	100%	100%	100%	98.8%	?Craniofacial anomalies and anterior segment dysgenesis syndrome, 614195;Keratoconus 1, 148300	VISION DISORDERS PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS
VSX2	100%	99.9%	100%	99.6%	Microphthalmia, isolated 2, 610093;Microphthalmia/coloboma 3, 610092	VISION DISORDERS PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹

VTN	100%	100%	100%	99.2%		MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS
VWA1	99.8%	99.1%	100%	99.1%	Neuronopathy, distal hereditary motor, autosomal recessive 7, 619216	POLYNEUROPATHIES PANEL ¹ MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹ MUSCLE DISORDERS PANEL
VWA3B	100%	100%	100%	98.5%	?Spinocerebellar ataxia, autosomal recessive 22, 616948	INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹

VWA8	100%	100%	100%	99.4%	?Retinitis pigmentosa 97, 620422	VISION DISORDERS PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS
VWF	100%	100%	100%	99.4%	von Willebrand disease, type 1, 193400;von Willebrand disease, types 2A, 2B, 2M, and 2N, 613554;von Willebrand disease, type 3, 277480	HEMOSTATIC/THROMB OTIC DISORDERS PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹
WAC	94%	93.9%	100%	99.2%	Desanto-Shinawi syndrome, 616708	INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS

WARS1	100%	100%	100%	99%	Neuronopathy, distal hereditary motor, autosomal dominant 9, 617721;Neurodevelopmental disorder with microcephaly and speech delay, with or without brain abnormalities, 620317	POLYNEUROPATHIES PANEL ¹ INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹
WARS2	100%	100%	100%	99.5%	Parkinsonism-dystonia 3, childhood-onset, 619738;Neurodevelopmental disorder, mitochondrial, with abnormal movements and lactic acidosis, with or without seizures, 617710	INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS MITOCHONDRIAL DISORDERS PANEL COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹

WAS	98.7%	91.4%	97.2%	63.5%	Wiskott-Aldrich syndrome, 301000;Neutropenia, severe congenital, X-linked, 300299;Thrombocytopenia, X-linked, intermittent, 313900;Thrombocytopenia, X-linked, 313900	INHERITED BONE MARROW FAILURE AND/OR PREDISPOSITION PANEL TO HEMATOLOGICAL MALIGNANCIES SKIN DISORDERS PANEL ¹ HEMOSTATIC/THROMBOTIC DISORDERS PANEL PRIMARY IMMUNODEFICIENCIES PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS HEREDITARY CANCER PANEL
WASF1	100%	100%	100%	98.7%	Neurodevelopmental disorder with absent language and variable seizures, 618707	INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS

WASHC4	100%	100%	100%	99.6%	Intellectual developmental disorder, autosomal recessive 43, 615817	INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹
WASHC5	100%	100%	100%	99.2%	Ritscher-Schinzel syndrome 1, 220210;Spastic paraplegia 8, autosomal dominant, 603563	MOVEMENT DISORDERS PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹ OROFACIAL CLEFTING PANEL WITH GENOME WIDE CNV ANALYSIS
WBP11	100%	100%	100%	98.9%	Vertebral, cardiac, tracheoesophageal, renal, and limb defects, 619227	MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS

WBP2	100%	100%	100%	98.7%	Deafness, autosomal recessive 107, 617639	HEARING IMPAIRMENT PANEL (INCLUDING GJB2) MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹
WBP4	100%	100%	100%	99.2%	Neurodevelopmental disorder with hypotonia, feeding difficulties, facial dysmorphism, and brain abnormalities, 620852	INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹
WDFY3	100%	100%	100%	99.3%	?Microcephaly 18, primary, autosomal dominant, 617520	INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS

WDPCP	97.7%	97.7%	100%	99.7%	Bardet-Biedl syndrome 15, 615992;Congenital heart defects, hamartomas of tongue, and polysyndactyly, 217085	VISION DISORDERS PANEL CILIOPATHIES PANEL DISORDERS/DIFFERENCES OF SEX DEVELOPMENT (DSD) / PRIMARY ADRENAL INSUFFICIENCY PANEL COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹ HYPOGONADOTROPIC HYPOGONADISM (KALLMANN) PANEL INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS
WDR1	100%	100%	100%	99%	Periodic fever, immunodeficiency, and thrombocytopenia syndrome, 150550	PRIMARY IMMUNODEFICIENCIES PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹

WDR11	100%	100%	100%	99.6%	Intellectual developmental disorder, autosomal recessive 78, 620237;Hypogonadotropic hypogonadism 14 with or without anosmia, 614858	DISORDERS/DIFFERENCES OF SEX DEVELOPMENT (DSD) / PRIMARY ADRENAL INSUFFICIENCY PANEL HYPOGONADOTROPIC HYPOGONADISM (KALLMANN) PANEL INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹
WDR13	100%	99.3%	97.9%	66.5%		INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS

WDR19	100%	100%	100%	99.6%	Nephronophthisis 13, 614377;Cranioectodermal dysplasia 4, 614378;Senior-Loken syndrome 8, 616307;Short-rib thoracic dysplasia 5 with or without polydactyly, 614376;?Spermatogenic failure 72, 619867	VISION DISORDERS PANEL CRANIOFACIAL ANOMALIES PANEL WITH GENOME WIDE CNV ANALYSIS CILIOPATHIES PANEL SKIN DISORDERS PANEL ¹ SHORT STATURE/SKELETAL DYSPLASIA PANEL WITH GENOME WIDE CNV ANALYSIS RENAL DISORDERS PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹
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WDR26	93.8%	93.8%	100%	98.3%	Skraban-Deardorff syndrome, 617616	MOVEMENT DISORDERS PANEL EPILEPSY PANEL INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS
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WDR35	100%	100%	100%	99.4%	Short-rib thoracic dysplasia 7 with or without polydactyly, 614091;Cranioectodermal dysplasia 2, 613610	MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹ SHORT STATURE/SKELETAL DYSPLASIA PANEL WITH GENOME WIDE CNV ANALYSIS LIVER DISORDERS PANEL RENAL DISORDERS PANEL CRANIOFACIAL ANOMALIES PANEL WITH GENOME WIDE CNV ANALYSIS CILIOPATHIES PANEL SKIN DISORDERS PANEL ¹ OROFACIAL CLEFTING PANEL WITH GENOME WIDE CNV ANALYSIS
WDR36	100%	100%	100%	99.5%	Glaucoma 1, open angle, G, 609887	VISION DISORDERS PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS

WDR37	90.3%	89.8%	100%	99.3%	Neurooculocardiogenitourinary syndrome, 618652	INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS
WDR4	100%	99.9%	100%	98.8%	Galloway-Mowat syndrome 6, 618347;Microcephaly, growth deficiency, seizures, and brain malformations, 618346	INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹
WDR44	100%	99.9%	99.4%	71.3%		INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS

WDR45	100%	99.9%	98.6%	66.5%	Neurodegeneration with brain iron accumulation 5, 300894	MOVEMENT DISORDERS PANEL MITOCHONDRIAL DISORDERS PANEL PARKINSON DISEASE PANEL EPILEPSY PANEL INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS
WDR45B	100%	100%	100%	99%	Neurodevelopmental disorder with spastic quadriplegia and brain abnormalities with or without seizures, 617977	INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL¹

WDR47	100%	100%	100%	99.5%		INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS
WDR5	100%	100%	100%	99.1%		INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS
WDR62	98.8%	98.7%	100%	99.3%	Microcephaly 2, primary, autosomal recessive, with or without cortical malformations, 604317	FETAL AKINESIA PANEL INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹

WDR72	96.8%	96.8%	100%	99.5%	Amelogenesis imperfecta, type IIA3, 613211	CRANIOFACIAL ANOMALIES PANEL WITH GENOME WIDE CNV ANALYSIS SKIN DISORDERS PANEL ¹ MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹
WDR73	100%	100%	100%	99.5%	Galloway-Mowat syndrome 1, 251300	MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹ MOVEMENT DISORDERS PANEL VISION DISORDERS PANEL INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS RENAL DISORDERS PANEL

WDR81	100%	100%	100%	99.3%	Cerebellar ataxia, impaired intellectual development, and dysquilibrium syndrome 2, 610185;Hydrocephalus, congenital, 3, with brain anomalies, 617967	MOVEMENT DISORDERS PANEL INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹
WDR83OS	100%	100%	100%	98.7%		INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS

WEE2	100%	100%	100%	99.5%	Oocyte/zygote/embryo maturation arrest 5, 617996	UNSUCCESSFUL IVF TREATMENTS, RECURRENT MOLAR PREGNANCIES AND/OR RECURRENT PREGNANCY LOSS PANEL ¹ MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹
WFDC2	100%	100%	100%	98.7%	Bronchiectasis and nasal polyposis, 620984	MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹

WFS1	91.2%	91.2%	100%	98.9%	Deafness, autosomal dominant 6/14/38, 600965;?Cataract 41, 116400;Wolfram-like syndrome, autosomal dominant, 614296;{Diabetes mellitus, noninsulin-dependent, association with}, 125853;Wolfram syndrome 1, 222300	MOVEMENT DISORDERS PANEL VISION DISORDERS PANEL HEARING IMPAIRMENT PANEL (INCLUDING GJB2) MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹ EPILEPSY PANEL INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS
WHRN	100%	100%	100%	98.9%	Deafness, autosomal recessive 31, 607084;Usher syndrome, type 2D, 611383	VISION DISORDERS PANEL HEARING IMPAIRMENT PANEL (INCLUDING GJB2) MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹

WIPF1	100%	100%	100%	99.1%	Wiskott-Aldrich syndrome 2, 614493	SKIN DISORDERS PANEL ¹ HEMOSTATIC/THROMB OTIC DISORDERS PANEL PRIMARY IMMUNODEFICIENCIES PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹
WIP12	100%	100%	100%	99%	?Intellectual developmental disorder with short stature and variable skeletal anomalies, 618453	INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹

WLS	100%	100%	100%	99.5%	Zaki syndrome, 619648	MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹
WNK1	100%	100%	100%	98.5%	Neuropathy, hereditary sensory and autonomic, type II, 201300;Pseudohypoaldosteronism, type IIC, 614492	POLYNEUROPATHIES PANEL ¹ NEUROLOGICAL PAIN DISORDERS PANEL ¹ RENAL DISORDERS PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹
WNK3	100%	99.9%	98.9%	69.2%	Prieto syndrome, 309610	INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS

WNK4	100%	99.9%	100%	98%	Pseudohypoaldosteronism, type IIB, 614491	RENAL DISORDERS PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS
WNT1	100%	100%	100%	99.3%	{Osteoporosis, early-onset, susceptibility to, autosomal dominant}, 615221;Osteogenesis imperfecta, type XV, 615220	SHORT STATURE/SKELETAL DYSPLASIA PANEL WITH GENOME WIDE CNV ANALYSIS INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL¹

WNT10A	100%	100%	100%	98.8%	Schopf-Schulz-Passarge syndrome, 224750;Tooth agenesis, selective, 4, 150400;Ectodermal dysplasia 16 (odontoonychodermal dysplasia), 257980	CRANIOFACIAL ANOMALIES PANEL WITH GENOME WIDE CNV ANALYSIS SKIN DISORDERS PANEL ¹ MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹
WNT10B	100%	100%	100%	99.2%	Tooth agenesis, selective, 8, 617073;Split-hand/foot malformation 6, 225300	CRANIOFACIAL ANOMALIES PANEL WITH GENOME WIDE CNV ANALYSIS SKIN DISORDERS PANEL ¹ SHORT STATURE/SKELETAL DYSPLASIA PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹

WNT2B	100%	100%	100%	99.2%	Diarrhea 9, 618168	MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹
WNT3	100%	99.8%	100%	99.5%	?Tetra-amelia syndrome 1, 273395	SHORT STATURE/SKELETAL DYSPLASIA PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹

WNT4	99.7%	96.5%	99.9%	98%	?SERKAL syndrome, 611812;Mullerian aplasia and hyperandrogenism, 158330	DISORDERS/DIFFERENCES OF SEX DEVELOPMENT (DSD) / PRIMARY ADRENAL INSUFFICIENCY PANEL RENAL DISORDERS PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹ OROFACIAL CLEFTING PANEL WITH GENOME WIDE CNV ANALYSIS
WNT5A	100%	100%	99.9%	95.1%	Robinow syndrome, autosomal dominant 1, 180700	SKIN DISORDERS PANEL ¹ SHORT STATURE/SKELETAL DYSPLASIA PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS OROFACIAL CLEFTING PANEL WITH GENOME WIDE CNV ANALYSIS

WNT6	100%	100%	100%	98.3%		SHORT STATURE/SKELETAL DYSPLASIA PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS
WNT7A	100%	100%	100%	99.2%	Fuhrmann syndrome, 228930;Ulna and fibula, absence of, with severe limb deficiency, 276820	SKIN DISORDERS PANEL ¹ SHORT STATURE/SKELETAL DYSPLASIA PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹

WRAP53	100%	100%	100%	98.7%	Dyskeratosis congenita, autosomal recessive 3, 613988	INHERITED BONE MARROW FAILURE AND/OR PREDISPOSITION PANEL TO HEMATOLOGICAL MALIGNANCIES SKIN DISORDERS PANEL ¹ DYSKERATOSIS CONGENITA AND APLASTIC ANEMIA PANEL PRIMARY IMMUNODEFICIENCIES PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹ HEREDITARY CANCER PANEL
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WRN	100%	100%	100%	99.5%	Werner syndrome, 277700	VISION DISORDERS PANEL SKIN DISORDERS PANEL ¹ MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹ HEREDITARY CANCER PANEL
WSB2	100%	99.5%	100%	98.5%		INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS

WT1	100%	100%	100%	98.8%	Mesothelioma, somatic, 156240;Meacham syndrome, 608978;Frasier syndrome, 136680;Nephrotic syndrome, type 4, 256370;Denys-Drash syndrome, 194080;Wilms tumor, type 1, 194070	DISORDERS/DIFFERENCES OF SEX DEVELOPMENT (DSD) / PRIMARY ADRENAL INSUFFICIENCY PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS HEREDITARY CANCER PANEL RENAL DISORDERS PANEL
WWOX	100%	100%	100%	99.4%	Esophageal squamous cell carcinoma, somatic, 133239;Developmental and epileptic encephalopathy 28, 616211;Spinocerebellar ataxia, autosomal recessive 12, 614322	MOVEMENT DISORDERS PANEL EPILEPSY PANEL INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹

XDH	100%	100%	100%	99.2%	Xanthinuria, type I, 278300	METABOLIC DISORDERS PANEL RENAL DISORDERS PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹
XIAP	100%	100%	98.8%	72.2%	Lymphoproliferative syndrome, X-linked, 2, 300635	MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS PRIMARY IMMUNODEFICIENCIES PANEL
XIRP2	100%	100%	100%	99.6%		HEART DISORDERS PANEL ¹ MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS
XIST					X-inactivation, familial skewed, 300087	MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS

XK	100%	99.1%	98.9%	68.3%	McLeod syndrome, 300842	MOVEMENT DISORDERS PANEL EPILEPSY PANEL HEART DISORDERS PANEL ¹ MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS MUSCLE DISORDERS PANEL
XKR8	100%	100%	100%	98.7%		HEARING IMPAIRMENT PANEL (INCLUDING GJB2) MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS
XKRY						MALE INFERTILITY PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS
XKRYP7						MALE INFERTILITY PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS

XPA	100%	100%	100%	99.9%	Xeroderma pigmentosum, group A, 278700	MOVEMENT DISORDERS PANEL SKIN DISORDERS PANEL ¹ INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹ HEREDITARY CANCER PANEL
XPC	100%	100%	100%	99.4%	Xeroderma pigmentosum, group C, 278720	SKIN DISORDERS PANEL ¹ MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹ HEREDITARY CANCER PANEL

XPNPEP3	100%	100%	100%	99.4%	Nephronophthisis-like nephropathy 1, 613159	CILIOPATHIES PANEL RENAL DISORDERS PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS MITOCHONDRIAL DISORDERS PANEL COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹
XPO5	100%	100%	100%	99.5%		RENAL DISORDERS PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS
XPR1	100%	100%	100%	99.4%	Basal ganglia calcification, idiopathic, 6, 616413	MOVEMENT DISORDERS PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS PARKINSON DISEASE PANEL

XRCC1	100%	100%	100%	98.2%	?Spinocerebellar ataxia, autosomal recessive 26, 617633	MOVEMENT DISORDERS PANEL POLYNEUROPATHIES PANEL ¹ MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹
XRCC2	100%	100%	100%	99.6%	Spermatogenic failure 50, 619145;?Premature ovarian failure 17, 619146;?Fanconi anemia, complementation group U, 617247	INHERITED BONE MARROW FAILURE AND/OR PREDISPOSITION PANEL TO HEMATOLOGICAL MALIGNANCIES MALE INFERTILITY PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹

XRCC4	100%	100%	100%	99.7%	Short stature, microcephaly, and endocrine dysfunction, 616541	SHORT STATURE/SKELETAL DYSPLASIA PANEL WITH GENOME WIDE CNV ANALYSIS INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL¹
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XYLT1	99.8%	98.1%	100%	98.5%	Desbuquois dysplasia 2, 615777;{Pseudoxanthoma elasticum, modifier of severity of}, 264800	SKIN DISORDERS PANEL ¹ SHORT STATURE/SKELETAL DYSPLASIA PANEL WITH GENOME WIDE CNV ANALYSIS METABOLIC DISORDERS PANEL INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹ OROFACIAL CLEFTING PANEL WITH GENOME WIDE CNV ANALYSIS
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XYLT2	99.6%	97.3%	100%	99.2%	{Pseudoxanthoma elasticum, modifier of severity of}, 264800;Spondyloocular syndrome, 605822	MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹ SKIN DISORDERS PANEL ¹ HEARING IMPAIRMENT PANEL (INCLUDING GJB2) SHORT STATURE/SKELETAL DYSPLASIA PANEL WITH GENOME WIDE CNV ANALYSIS METABOLIC DISORDERS PANEL
YAP1	100%	100%	100%	99.4%	Coloboma, ocular, with or without hearing impairment, cleft lip/palate, and/or impaired intellectual development, 120433	VISION DISORDERS PANEL HEARING IMPAIRMENT PANEL (INCLUDING GJB2) MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS

YARS1	100%	100%	100%	99.3%	Infantile-onset multisystem neurologic, endocrine, and pancreatic disease 2, 619418;Charcot-Marie-Tooth disease, dominant intermediate C, 608323	VISION DISORDERS PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹ HEARING IMPAIRMENT PANEL (INCLUDING GJB2) POLYNEUROPATHIES PANEL ¹ LIVER DISORDERS PANEL INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS
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YARS2	100%	100%	100%	99.5%	Myopathy, lactic acidosis, and sideroblastic anemia 2, 613561	INHERITED BONE MARROW FAILURE AND/OR PREDISPOSITION PANEL TO HEMATOLOGICAL MALIGNANCIES IRON DISORDERS PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS MITOCHONDRIAL DISORDERS PANEL COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹ MUSCLE DISORDERS PANEL
YEATS2	100%	100%	100%	99.5%	?Epilepsy, myoclonic, familial adult, 4, 615127	MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS

YIF1B	90%	90%	100%	99.2%	Kaya-Barakat-Masson syndrome, 619125	INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹
YIPF5	100%	100%	100%	99%	Microcephaly, epilepsy, and diabetes syndrome 2, 619278	INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹

YME1L1	100%	100%	100%	99.7%	?Optic atrophy 11, 617302	VISION DISORDERS PANEL METABOLIC DISORDERS PANEL INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS MITOCHONDRIAL DISORDERS PANEL COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹
YPEL2	100%	100%	100%	99.9%		VISION DISORDERS PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS
YRDC	100%	100%	100%	98.7%	Galloway-Mowat syndrome 10, 619609	MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹

YWHAE	100%	100%	100%	99%		INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS
YWHAG	100%	100%	100%	99.4%	Developmental and epileptic encephalopathy 56, 617665	EPILEPSY PANEL INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS
YWHAZ	100%	100%	100%	98.3%		SKIN DISORDERS PANEL ¹ INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS

YY1	100%	99.9%	99.9%	93.2%	Gabriele-de Vries syndrome, 617557	INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS
YY1AP1	100%	100%	100%	99.2%	Grange syndrome, 602531	INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹
ZAP70	100%	100%	100%	99.5%	Immunodeficiency 48, 269840;Autoimmune disease, multisystem, infantile-onset, 2, 617006	PRIMARY IMMUNODEFICIENCIES PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹ SEVERE COMBINED IMMUNODEFICIENCY (SCID) PANEL

ZBTB11	100%	100%	100%	99.5%	Intellectual developmental disorder, autosomal recessive 69, 618383	METABOLIC DISORDERS PANEL INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹
ZBTB16	96%	96%	100%	98.7%	Leukemia, acute promyelocytic, PL2F/RARA type	SHORT STATURE/SKELETAL DYSPLASIA PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹ INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS

ZBTB17	100%	100%	100%	99.2%		HEART DISORDERS PANEL ¹ MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS
ZBTB18	93.2%	93.2%	100%	98.7%	Intellectual developmental disorder, autosomal dominant 22, 612337	INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS
ZBTB20	100%	100%	100%	99.1%	Primrose syndrome, 259050	SKIN DISORDERS PANEL ¹ INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS

ZBTB24	100%	100%	100%	99.2%	Immunodeficiency-centromeric instability-facial anomalies syndrome 2, 614069	PRIMARY IMMUNODEFICIENCIES PANEL INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL¹
ZBTB42	100%	100%	100%	99.6%	?Lethal congenital contracture syndrome 6, 616248	MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL¹
ZBTB47	99.5%	99.3%	100%	97.7%		INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS

ZBTB7A	100%	99.9%	100%	95.3%	Macrocephaly, neurodevelopmental delay, lymphoid hyperplasia, and persistent fetal hemoglobin, 619769	INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS
ZC3H14	100%	100%	100%	99.7%	Intellectual developmental disorder, autosomal recessive 56, 617125	INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹

ZC4H2	100%	99.7%	98.9%	69.6%	Wieacker-Wolff syndrome, 314580;Wieacker-Wolff syndrome, female-restricted, 301041	FETAL AKINESIA PANEL MOVEMENT DISORDERS PANEL SHORT STATURE/SKELETAL DYSPLASIA PANEL WITH GENOME WIDE CNV ANALYSIS INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS MUSCLE DISORDERS PANEL
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ZCCHC8	100%	100%	100%	99.4%	?Pulmonary fibrosis and/or bone marrow failure syndrome, telomere-related, 5, 618674	INHERITED BONE MARROW FAILURE AND/OR PREDISPOSITION PANEL TO HEMATOLOGICAL MALIGNANCIES DYSKERATOSIS CONGENITA AND APLASTIC ANEMIA PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS HEREDITARY CANCER PANEL
ZDHHC9	100%	99.7%	98.5%	69.4%	Intellectual developmental disorder, X-linked syndromic, Raymond type, 300799	INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS
ZEB1	100%	100%	100%	99.5%	Corneal dystrophy, posterior polymorphous, 3, 609141;Corneal dystrophy, Fuchs endothelial, 6, 613270	VISION DISORDERS PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS

ZEB2	100%	100%	100%	98.5%	Mowat-Wilson syndrome, 235730	CRANIOFACIAL ANOMALIES PANEL WITH GENOME WIDE CNV ANALYSIS EPILEPSY PANEL INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS OROFACIAL CLEFTING PANEL WITH GENOME WIDE CNV ANALYSIS
ZFHX2	100%	100%	100%	98.9%	?Marsili syndrome, 147430	NEUROLOGICAL PAIN DISORDERS PANEL¹ MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS
ZFHX3	100%	100%	100%	98.9%	Prostate cancer, somatic, 176807;{Atrial fibrillation 8, susceptibility to}, 613055;Spinocerebellar ataxia 4, 600223	INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS

ZFHx4	100%	100%	100%	99.2%	?Ptosis, congenital, 178300	INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS OROFACIAL CLEFTING PANEL WITH GENOME WIDE CNV ANALYSIS
ZFP36L2	100%	100%	100%	98.3%	Oocyte/zygote/embryo maturation arrest 13, 620154	UNSUCCESSFUL IVF TREATMENTS, RECURRENT MOLAR PREGNANCIES AND/OR RECURRENT PREGNANCY LOSS PANEL ¹ MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS
ZFP57	100%	100%	100%	98.8%	Diabetes mellitus, transient neonatal 1, 601410	MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS

ZFPM2	100%	100%	100%	99.6%	Diaphragmatic hernia 3, 610187;46XY sex reversal 9, 616067;Tetralogy of Fallot, 187500	CONGENITAL HEARTDISEASE PANEL ¹ DISORDERS/DIFFEREN CES OF SEX DEVELOPMENT (DSD) / PRIMARY ADRENAL INSUFFICIENCY PANEL HEART DISORDERS PANEL ¹ MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS
ZFTRAF1	92.1%	87.8%	100%	94.3%		INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS
ZFX	100%	100%	99.3%	69.8%	Intellectual developmental disorder, X-linked syndromic 37, 301118	MALE INFERTILITY PANEL INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS

ZFYVE19	100%	100%	100%	99.2%	Cholestasis, progressive familial intrahepatic, 9, 619849	LIVER DISORDERS PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹
ZFYVE26	100%	100%	100%	99.4%	Spastic paraplegia 15, autosomal recessive, 270700	MOVEMENT DISORDERS PANEL POLYNEUROPATHIES PANEL ¹ INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹
ZFYVE27	100%	100%	100%	98.8%		MOVEMENT DISORDERS PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS

ZIC1	100%	100%	100%	98.9%	?Craniosynostosis 6, 616602;Structural brain anomalies with impaired intellectual development and craniosynostosis, 618736	CRANIOFACIAL ANOMALIES PANEL WITH GENOME WIDE CNV ANALYSIS INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS
ZIC2	100%	98.1%	100%	95.3%	Holoprosencephaly 5, 609637	CRANIOFACIAL ANOMALIES PANEL WITH GENOME WIDE CNV ANALYSIS INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS OROFACIAL CLEFTING PANEL WITH GENOME WIDE CNV ANALYSIS

ZIC3	100%	100%	98.6%	66.3%	Congenital heart defects, nonsyndromic, 1, X-linked, 306955;Heterotaxy, visceral, 1, X-linked, 306955;VACTERL association, X-linked, 314390	CONGENITAL HEARTDISEASE PANEL¹ CILIOPATHIES PANEL HEART DISORDERS PANEL¹ MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS OROFACIAL CLEFTING PANEL WITH GENOME WIDE CNV ANALYSIS
ZMIZ1	99.9%	99.6%	100%	98.3%	Neurodevelopmental disorder with dysmorphic facies and distal skeletal anomalies, 618659	INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS

ZMPSTE24	100%	100%	100%	99.5%	Mandibuloacral dysplasia with type B lipodystrophy, 608612;Restrictive dermopathy 1, 275210	FETAL AKINESIA PANEL SKIN DISORDERS PANEL ¹ RENAL DISORDERS PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹ OROFACIAL CLEFTING PANEL WITH GENOME WIDE CNV ANALYSIS SHORT STATURE/SKELETAL DYSPLASIA PANEL WITH GENOME WIDE CNV ANALYSIS METABOLIC DISORDERS PANEL
ZMYM2	100%	100%	100%	99.4%	Neurodevelopmental-craniofacial syndrome with variable renal and cardiac abnormalities, 619522	INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS

ZMYM3	100%	99.5%	98.7%	66.9%	Intellectual developmental disorder, X-linked 112, 301111	INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS
ZMYND10	100%	100%	100%	99.5%	Ciliary dyskinesia, primary, 22, 615444	CILIOPATHIES PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹
ZMYND11	100%	100%	100%	99.2%	Intellectual developmental disorder, autosomal dominant 30, 616083	INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS
ZMYND15	100%	100%	100%	97.9%	?Spermatogenic failure 14, 615842	MALE INFERTILITY PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS

ZMYND8	100%	100%	100%	99.2%		INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS
ZNF141	100%	100%	100%	99.6%	?Polydactyly, postaxial, type A6, 615226	MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS
ZNF142	100%	100%	100%	99.3%	Neurodevelopmental disorder with impaired speech and hyperkinetic movements, 618425	INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹
ZNF143	100%	100%	100%	99.3%		METABOLIC DISORDERS PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS

ZNF148	100%	100%	100%	99.5%	Global developmental delay, absent or hypoplastic corpus callosum, and dysmorphic facies, 617260	INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS
ZNF292	99.4%	99.4%	100%	99.6%	Intellectual developmental disorder, autosomal dominant 64, 619188	INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS
ZNF335	100%	100%	100%	99.4%	Microcephaly 10, primary, autosomal recessive, 615095	INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹

ZNF341	100%	100%	100%	98.7%	Hyper-IgE syndrome 3, autosomal recessive, with recurrent infections, 618282	PRIMARY IMMUNODEFICIENCIES PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹
ZNF407	100%	100%	100%	99.7%	SIMHA syndrome, 619557	INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹
ZNF408	100%	100%	100%	99.2%	Retinitis pigmentosa 72, 616469;?Exudative vitreoretinopathy 6, 616468	VISION DISORDERS PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹

ZNF41	100%	100%	99.2%	72.7%		INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS
ZNF423	100%	100%	100%	99.5%	Nephronophthisis 14, 614844;Joubert syndrome 19, 614844	VISION DISORDERS PANEL CILIOPATHIES PANEL RENAL DISORDERS PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹
ZNF462	100%	100%	100%	99.3%	Weiss-Kruszka syndrome, 618619	INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS

ZNF469	100%	100%	100%	98.6%	Brittle cornea syndrome 1, 229200	VISION DISORDERS PANEL SKIN DISORDERS PANEL ¹ MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹
ZNF513	100%	100%	100%	98.5%	?Retinitis pigmentosa 58, 613617	VISION DISORDERS PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹
ZNF526	100%	100%	100%	98.9%	Dentici-Novelli neurodevelopmental syndrome, 619877	INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹

ZNF541	100%	100%	100%	99%		DISORDERS/DIFFERENCES OF SEX DEVELOPMENT (DSD) / PRIMARY ADRENAL INSUFFICIENCY PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS PREMATURE OVARIAN INSUFFICIENCY PANEL
ZNF592	100%	100%	100%	99.5%		SKIN DISORDERS PANEL ¹ MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS
ZNF644	100%	100%	100%	99.5%	Myopia 21, autosomal dominant, 614167	VISION DISORDERS PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS

ZNF668	100%	100%	100%	99.2%	Neurodevelopmental disorder with poor growth, large ears, and dysmorphic facies, 620194	INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹
ZNF687	100%	100%	100%	99.3%	Paget disease of bone 6, 616833	MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS
ZNF699	100%	100%	100%	99.5%	DEGCAGS syndrome, 619488	INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹

ZNF711	100%	100%	98.7%	67.6%	Intellectual developmental disorder, X-linked 97, 300803	INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS
ZNF750	100%	100%	100%	99.4%	?Seborrhea-like dermatitis with psoriasiform elements, 610227	SKIN DISORDERS PANEL ¹ MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS
ZNF808	100%	100%	100%	99.8%	Pancreatic agenesis 3, 620991	MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹
ZNF862	100%	100%	100%	99.2%	?Fibromatosis, gingival 6, 620999	MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS

ZNFX1	100%	100%	100%	99.5%	Immunodeficiency 91 and hyperinflammation, 619644	PRIMARY IMMUNODEFICIENCIES PANEL INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL¹
ZNG1A	98%	96.8%	96.6%	94.3%		RENAL DISORDERS PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS
ZNHIT3	76.8%	76.2%	100%	99.1%	PEHO syndrome, 260565	INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL¹

ZNRF3	96.8%	95.3%	100%	99%		INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS
ZP1	100%	100%	100%	98.9%	Oocyte/zygote/embryo maturation arrest 1, 615774	UNSUCCESSFUL IVF TREATMENTS, RECURRENT MOLAR PREGNANCIES AND/OR RECURRENT PREGNANCY LOSS PANEL ¹ MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹

ZP2	100%	100%	100%	99.2%	Oocyte/zygote/embryo maturation arrest 6, 618353	UNSUCCESSFUL IVF TREATMENTS, RECURRENT MOLAR PREGNANCIES AND/OR RECURRENT PREGNANCY LOSS PANEL ¹ MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS
ZP3	100%	100%	100%	99.2%	Oocyte/zygote/embryo maturation arrest 3, 617712	UNSUCCESSFUL IVF TREATMENTS, RECURRENT MOLAR PREGNANCIES AND/OR RECURRENT PREGNANCY LOSS PANEL ¹ MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS
ZBPB	100%	100%	100%	99.5%	?Spermatogenic failure 66, 619799	MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS
ZPR1	100%	100%	100%	98.9%	?Growth restriction, hypoplastic kidneys, alopecia, and distinctive facies, 619321	MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS

ZRSR2	84.1%	84.1%	98.9%	68.6%	Orofaciodigital syndrome XXI, 301132	INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS
ZSCAN10	100%	100%	100%	99.3%	Otofacial neurodevelopmental syndrome, 620910	INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL ¹

ZSWIM6	96.1%	92.8%	99.2%	94.5%	Neurodevelopmental disorder with movement abnormalities, abnormal gait, and autistic features, 617865;Acromelic frontonasal dysostosis, 603671	OROFACIAL CLEFTING PANEL WITH GENOME WIDE CNV ANALYSIS SHORT STATURE/SKELETAL DYSPLASIA PANEL WITH GENOME WIDE CNV ANALYSIS INTELLECTUAL DISABILITY PANEL WITH GENOME WIDE CNV ANALYSIS MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS
ZSWIM7	89.7%	88.9%	100%	99.7%	Spermatogenic failure 71, 619831;?Ovarian dysgenesis 10, 619834	MALE INFERTILITY PANEL MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS

Gene symbols used follow HGCN guidelines: Gray KA, Yates B, Seal RL, Wright MW, Bruford EA. Nucleic Acids Res. 2015 Jan 43(Database issue):D1079-85.

TWIST X2 covered 10x describes the percentage of a gene's coding sequence that is covered at least 10x when analyzed by WES using TWIST X2 chemistry mapped against GRCh38.

TWIST X2 covered 20x describes the percentage of a gene's coding sequence that is covered at least 20x when analyzed by WES using TWIST X2 chemistry mapped against GRCh38.

srWGS covered 10x describes the percentage of a gene's coding sequence that is covered at least 10x when analyzed by WGS mapped against GRCh38.

srWGS covered 15x describes the percentage of a gene's coding sequence that is covered at least 15x when analyzed by WGS mapped against GRCh38.

srWGS covered 20x describes the percentage of a gene's coding sequence that is covered at least 20x when analyzed by WGS mapped against GRCh38.

non-protein coding genes are covered, but as coverage statistics are based on protein coding regions, statistics could not be generated.

OMIM release used for OMIM disease identifiers and descriptions : November 25th, 2024.

This list is accurate for panel version DG 4.2.0

Ad 1. Blank field signifies a gene without a current OMIM association Ad 2. OMIM phenotype descriptions between {} signify risk factors