MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS DG-4.1.0 (5369 GENES)

Gene	*Twist X2 covered >10x*	*Twist X2 covered >20x*	*WGS covered >10x*	*WGS covered >20x*	*Associated Phenotype description and OMIM disease ID*
A2M	100%	100%	100%	99.7%	
A4GALT	100%	100%	100%	97.2%	[Blood group, P1Pk system, P(2) phenotype], 111400;NOR polyagglutination syndrome, 111400;[Blood group, P1Pk system, p phenotype], 111400
AAAS	100%	100%	100%	99%	Achalasia-addisonianism-alacrimia syndrome, 231550
AAGAB	100%	100%	100%	99.5%	Keratoderma, palmoplantar, punctate type IA, 148600
AARS1	100%	100%	100%	99.1%	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

AARS2	100%	100%	100%	99.3%	Leukoencephalopathy, progressive, with ovarian failure, 615889;Combined oxidative phosphorylation deficiency 8, 614096
AASS	100%	100%	100%	99.5%	Hyperlysinemia, 238700
ABAT	100%	100%	100%	98.9%	GABA-transaminase deficiency, 613163
ABCA1	100%	100%	100%	99.6%	Tangier disease, 205400;HDL deficiency, familial, 1, 604091
ABCA12	100%	100%	100%	99.3%	Ichthyosis, congenital, autosomal recessive 4B (harlequin), 242500;Ichthyosis, congenital, autosomal recessive 4A, 601277
ABCA2	100%	99.7%	100%	97.8%	Intellectual developmental disorder with poor growth and with or without seizures or ataxia, 618808
ABCA3	100%	100%	100%	99.1%	Surfactant metabolism dysfunction, pulmonary, 3, 610921

ABCA4	100%	100%	100%	99.4%	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
ABCA5	100%	100%	100%	99.8%	?Hypertrichosis, congenital generalized, with gingival hyperplasia, 135400
ABCB1	100%	100%	100%	99.5%	Encephalopathy, acute transient, 620950;{Inflammatory bowel disease 13}, 612244;{Colchicine resistance}, 120080
ABCB11	100%	100%	100%	99.6%	Cholestasis, benign recurrent intrahepatic, 2, 605479;Cholestasis, progressive familial intrahepatic 2, 601847
ABCB4	100%	100%	100%	99.7%	Gallbladder disease 1, 600803;Cholestasis, intrahepatic, of pregnancy, 3, 614972;Cholestasis, progressive familial intrahepatic 3, 602347

ABCB6	100%	100%	100%	99.2%	Dyschromatosis universalis hereditaria 3, 615402;[Blood group, Langereis system], 111600;Pseudohyperkalemia, familial, 2, due to red cell leak, 609153;Microphthalmia/coloboma 7, 614497
ABCB7	100%	99.7%	99.1%	74.9%	Anemia, sideroblastic, with ataxia, 301310
ABCC1	100%	100%	100%	99%	?Deafness, autosomal dominant 77, 618915
ABCC2	100%	100%	100%	99.6%	Dubin-Johnson syndrome, 237500
ABCC6	98.4%	98.4%	100%	99.2%	Pseudoxanthoma elasticum, 264800;Arterial calcification, generalized, of infancy, 2, 614473;Pseudoxanthoma elasticum, forme fruste, 177850
ABCC8	100%	100%	100%	99.2%	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

ABCC9	96%	96%	100%	99.6%	Cardiomyopathy, dilated, 1O, 608569;Hypertrichotic osteochondrodysplasia (Cantu syndrome), 239850;?Atrial fibrillation, familial, 12, 614050;Intellectual disability and myopathy syndrome, 619719
ABCD1	100%	98.5%	98.7%	69.5%	Adrenoleukodystrophy, 300100;Adrenomyeloneuropathy, adult, 300100
ABCD2	100%	100%	100%	99.7%	
ABCD3	100%	100%	100%	99.8%	?Bile acid synthesis defect, congenital, 5, 616278
ABCD4	100%	100%	100%	99.2%	Methylmalonic aciduria and homocystinuria, cblJ type, 614857
ABCG5	100%	100%	100%	99.4%	Sitosterolemia 2, 618666
ABCG8	100%	100%	100%	99.2%	Sitosterolemia 1, 210250;{Gallbladder disease 4}, 611465
ABHD12	100%	100%	100%	99.6%	Polyneuropathy, hearing loss, ataxia, retinitis pigmentosa, and cataract, 612674
ABHD16A	100%	100%	100%	99%	Spastic paraplegia 86, autosomal recessive, 619735
ABHD5	100%	100%	100%	99.2%	Chanarin-Dorfman syndrome, 275630

ABL1	100%	100%	100%	98.8%	Leukemia, Philadelphia chromosome-positive, resistant to imatinib, 608232;Congenital heart defects and skeletal malformations syndrome, 617602
ACACA	100%	100%	100%	99.7%	Acetyl-CoA carboxylase deficiency, 613933
ACAD8	100%	100%	100%	98.9%	Isobutyryl-CoA dehydrogenase deficiency, 611283
ACAD9	100%	100%	100%	99.7%	Mitochondrial complex I deficiency, nuclear type 20, 611126
ACADM	96.6%	94.2%	100%	99.8%	Acyl-CoA dehydrogenase, medium chain, deficiency of, 201450
ACADS	100%	100%	100%	99.2%	Acyl-CoA dehydrogenase, short-chain, deficiency of, 201470
ACADSB	100%	100%	100%	99.9%	2-methylbutyrylglycinuria, 610006
ACADVL	100%	100%	100%	98.3%	VLCAD deficiency, 201475

ACAN	99.1%	98.9%	98.4%	95.4%	?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
ACAT1	100%	100%	100%	98.9%	Alpha-methylacetoacetic aciduria, 203750
ACAT2	100%	100%	100%	99.6%	?ACAT2 deficiency, 614055
ACBD5	85.6%	85.6%	100%	99.4%	Retinal dystrophy with leukodystrophy, 618863
ACBD6	100%	100%	100%	99.6%	Neurodevelopmental disorder with progressive movement abnormalities, 620785
ACD	100%	100%	100%	98.9%	?Dyskeratosis congenita, autosomal recessive 7, 616553;?Dyskeratosis congenita, autosomal dominant 6, 616553

ACE	100%	100%	99.9%	96.7%	{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}
ACER3	96.3%	96.3%	100%	99.7%	?Leukodystrophy, progressive, early childhood-onset, 617762
ACKR3	100%	100%	100%	99%	?Oculomotor-abducens synkinesis, 619215
ACO2	93.1%	90.3%	100%	99.3%	Optic atrophy 9, 616289;Infantile cerebellar-retinal degeneration, 614559
ACOX1	100%	100%	100%	99.8%	Mitchell syndrome, 618960;Peroxisomal acyl-CoA oxidase deficiency, 264470
ACOX2	100%	100%	100%	99.3%	Bile acid synthesis defect, congenital, 6, 617308
ACP4	100%	100%	100%	99.2%	Amelogenesis imperfecta, type IJ, 617297
ACP5	100%	100%	100%	98.7%	Spondyloenchondrodysplasia with immune dysregulation, 607944

ACR	100%	98.8%	97.5%	85.4%	?Spermatogenic failure 87, 620500
ACSF3	100%	100%	100%	99.4%	Combined malonic and methylmalonic aciduria, 614265
ACSL4	100%	99.9%	99.4%	74.8%	Intellectual developmental disorder, X-linked 63, 300387
ACSL5	100%	100%	100%	99.6%	?Diarrhea 13, 620357
ACSL6	100%	100%	100%	99.3%	
ACTA1	100%	100%	100%	98.5%	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
ACTA2	100%	100%	100%	99.5%	Smooth muscle dysfunction syndrome, 613834;Aortic aneurysm, familial thoracic 6, 611788;Moyamoya disease 5, 614042

ACTB	100%	100%	100%	99.3%	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
ACTC1	100%	100%	100%	99.4%	Left ventricular noncompaction 4, 613424;Cardiomyopathy, hypertrophic, 11, 612098;Atrial septal defect 5, 612794;Cardiomyopathy, dilated, 1R, 613424
ACTG1	100%	100%	100%	99.3%	Deafness, autosomal dominant 20/26, 604717;Baraitser-Winter syndrome 2, 614583
ACTG2	100%	100%	100%	99%	Megacystis-microcolon-intestinal hypoperistalsis syndrome 5, 619431;Visceral myopathy 1, 155310
ACTL6A	100%	100%	100%	99.9%	

ACTL6B	100%	100%	100%	99.1%	Developmental and epileptic encephalopathy 76, 618468;Intellectual developmental disorder with severe speech and ambulation defects, 618470
ACTL7A	100%	100%	100%	99.1%	Spermatogenic failure 86, 620499
ACTL9	100%	100%	100%	99.4%	Spermatogenic failure 53, 619258
ACTN1	100%	100%	100%	99.2%	Bleeding disorder, platelet-type, 15, 615193
ACTN2	100%	100%	99.9%	97.4%	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
ACTN4	98.9%	98.9%	100%	98.6%	Glomerulosclerosis, focal segmental, 1, 603278
ACTRT1	100%	100%	98.6%	71.3%	
ACVR1	100%	100%	100%	99.6%	Fibrodysplasia ossificans progressiva, 135100
ACVR1B	100%	100%	100%	97.8%	Pancreatic cancer, somatic, 260350
ACVR2B	100%	100%	100%	99.1%	Heterotaxy, visceral, 4, autosomal, 613751

ACVRL1	100%	100%	100%	98.9%	Telangiectasia, hereditary hemorrhagic, type 2, 600376
ACY1	100%	100%	100%	99.4%	Aminoacylase 1 deficiency, 609924
ADA	87.2%	84.6%	100%	99.5%	Adenosine deaminase deficiency, partial, 102700;Severe combined immunodeficiency due to ADA deficiency, 102700
ADA2	95.2%	93.2%	100%	99.3%	Sneddon syndrome, 182410;Vasculitis, autoinflammation, immunodeficiency, and hematologic defects syndrome, 615688
ADAD2	100%	100%	100%	99.1%	
ADAM10	100%	100%	100%	99.8%	{Alzheimer disease 18, susceptibility to}, 615590;Reticulate acropigmentation of Kitamura, 615537
ADAM17	99.2%	99.2%	100%	99.4%	?Inflammatory skin and bowel disease, neonatal, 1, 614328
ADAM22	100%	100%	100%	99.6%	Developmental and epileptic encephalopathy 61, 617933
ADAM9	95%	95%	100%	99.4%	Cone-rod dystrophy 9, 612775
ADAMTS1	100%	100%	100%	99.6%	

ADAMTS10	100%	100%	100%	98.7%	Weill-Marchesani syndrome 1, recessive, 277600
ADAMTS13	100%	100%	100%	98.7%	Thrombotic thrombocytopenic purpura, hereditary, 274150
ADAMTS15	100%	100%	100%	98.4%	Arthrogryposis, distal, type 12, 620545
ADAMTS17	100%	99.9%	100%	97.2%	Weill-Marchesani 4 syndrome, recessive, 613195
ADAMTS18	100%	100%	100%	99.7%	Microcornea, myopic chorioretinal atrophy, and telecanthus, 615458
ADAMTS19	100%	100%	100%	99.7%	Cardiac valvular dysplasia 2, 620067
ADAMTS2	97.9%	97.9%	100%	99.2%	Ehlers-Danlos syndrome, dermatosparaxis type, 225410
ADAMTS3	100%	100%	100%	99.5%	Hennekam lymphangiectasia-lymphedema syndrome 3, 618154
ADAMTS9	100%	100%	100%	98.6%	
ADAMTSL2	100%	99.8%	100%	99.2%	Geleophysic dysplasia 1, 231050
ADAMTSL4	100%	100%	100%	99%	Ectopia lentis et pupillae, 225200;Ectopia lentis, isolated, autosomal recessive, 225100

ADAR	100%	100%	100%	99.2%	Dyschromatosis symmetrica hereditaria, 127400;Aicardi-Goutieres syndrome 6, 615010
ADARB1	94.7%	94.7%	100%	99.5%	Neurodevelopmental disorder with hypotonia, microcephaly, and seizures, 618862
ADAT3	100%	100%	100%	98.2%	Neurodevelopmental disorder with brain abnormalities, poor growth, and dysmorphic facies, 615286
ADCK2	100%	100%	100%	98.5%	
ADCK5	100%	100%	100%	99.1%	
ADCY1	99.2%	98.6%	99.9%	96.4%	?Deafness, autosomal recessive 44, 610154
ADCY10	100%	100%	100%	99.6%	{Hypercalciuria, absorptive, susceptibility to}, 143870
ADCY3	100%	100%	100%	99.2%	{Obesity, susceptibility to, BMIQ19}, 617885
ADCY5	97.4%	97.1%	100%	98.2%	Dyskinesia with orofacial involvement, autosomal dominant, 606703;Neurodevelopmental disorder with hyperkinetic movements and dyskinesia, 619651;Dyskinesia with orofacial involvement, autosomal recessive, 619647

ADCY6	100%	100%	100%	99.4%	Lethal congenital contracture syndrome 8, 616287
ADD1	100%	100%	100%	99.4%	{Hypertension, essential, salt-sensitive}, 145500
ADD3	100%	100%	100%	99.7%	Cerebral palsy, spastic quadriplegic, 3, 617008
ADGB	100%	100%	100%	99.3%	
ADGRE2	99.6%	99%	99.8%	97.3%	Vibratory urticaria, 125630
ADGRG1	100%	100%	100%	98.8%	Cortical dysplasia, complex, with other brain malformations 14B, (bilateral perisylvian), 615752;Cortical dysplasia, complex, with other brain malformations 14A, (bilateral frontoparietal), 606854
ADGRG2	100%	99.7%	99%	74.2%	Congenital bilateral absence of vas deferens, X-linked, 300985
ADGRG6	100%	100%	100%	99.8%	Lethal congenital contracture syndrome 9, 616503
ADGRL1	100%	100%	100%	98.6%	Developmental delay, behavioral abnormalities, and neuropsychiatric disorders, 620065

ADGRV1	100%	100%	100%	99.7%	Usher syndrome, type 2C, 605472;Usher syndrome, type 2C, GPR98/PDZD7 digenic, 605472;?Febrile seizures, familial, 4, 604352
ADH5	100%	99.9%	100%	99.2%	AMED syndrome, digenic, 619151
ADIPOQ	100%	100%	100%	99.5%	Adiponectin deficiency, 612556
ADIPOR1	100%	100%	100%	99.7%	
ADK	90.9%	90.9%	100%	99.8%	Hypermethioninemia due to adenosine kinase deficiency, 614300
ADNP	100%	100%	100%	99.6%	Helsmoortel-van der Aa syndrome, 615873
ADPRS	100%	100%	100%	98.6%	Neurodegeneration, childhood-onset, stress-induced, with variable ataxia and seizures, 618170
ADRA2A	100%	100%	100%	93.2%	?Lipodystrophy, familial partial, type 8, 620679
ADRB1	99.9%	98.4%	100%	97.1%	?[Short sleep, familial natural, 2], 618591;[Resting heart rate], 607276
ADRB2	100%	100%	100%	99.4%	Beta-2-adrenoreceptor agonist, reduced response to
ADSL	100%	100%	100%	99.7%	Adenylosuccinase deficiency, 103050
ADSS1	100%	100%	100%	98.5%	Myopathy, distal, 5, 617030

AEBP1	100%	100%	100%	98.8%	Ehlers-Danlos syndrome, classic-like, 2, 618000
AFF2	100%	99.9%	99.3%	74.3%	Intellectual developmental disorder, X-linked 109, 309548
AFF3	100%	100%	100%	98.7%	KINSSHIP syndrome, 619297
AFF4	100%	100%	100%	99.7%	CHOPS syndrome, 616368
AFG2A	100%	100%	100%	99.6%	Neurodevelopmental disorder with hearing loss, seizures, and brain abnormalities, 616577
AFG2B	100%	100%	100%	99.6%	Deafness, autosomal recessive 119, 619615;Neurodevelopmental disorder with hearing loss and spasticity, 619616
AFG3L2	100%	100%	100%	99.4%	Spastic ataxia 5, autosomal recessive, 614487;Optic atrophy 12, 618977;Spinocerebellar ataxia 28, 610246
AFP	100%	100%	100%	99.9%	[Hereditary persistence of alpha-fetoprotein], 615970;Alpha-fetoprotein deficiency, 615969
AGA	100%	100%	100%	99.4%	Aspartylglucosaminuria, 208400
AGAP1	100%	99.9%	100%	96.8%	
AGBL1	100%	100%	100%	99.6%	Corneal dystrophy, Fuchs endothelial, 8, 615523

AGBL5	100%	100%	100%	99.3%	Retinitis pigmentosa 75, 617023
AGK	91.7%	91.7%	100%	99.7%	Cataract 38, autosomal recessive, 614691;Sengers syndrome, 212350
AGL	100%	100%	100%	99.7%	Glycogen storage disease IIIa, 232400;Glycogen storage disease IIIb, 232400
AGMO	100%	100%	100%	99.7%	
AGO1	100%	100%	100%	99.3%	Neurodevelopmental disorder with language delay and behavioral abnormalities, with or without seizures, 620292
AGO2	99.9%	99.4%	100%	98.9%	Lessel-Kreienkamp syndrome, 619149
AGPAT2	100%	100%	100%	98%	Lipodystrophy, congenital generalized, type 1, 608594
AGPS	97.3%	97.3%	100%	99.4%	Rhizomelic chondrodysplasia punctata, type 3, 600121
AGR2	100%	100%	100%	99.4%	Respiratory infections, recurrent, and failure to thrive with or without diarrhea, 620233
AGRN	100%	100%	100%	98%	Myasthenic syndrome, congenital, 8, with pre- and postsynaptic defects, 615120

AGT	96.4%	95.3%	100%	99.6%	Renal tubular dysgenesis, 267430
AGTPBP1	100%	100%	100%	99.7%	Neurodegeneration, childhood-onset, with cerebellar atrophy, 618276
AGTR1	100%	100%	100%	99.9%	{Hypertension, essential}, 145500;Renal tubular dysgenesis, 267430
AGXT	100%	100%	100%	99.4%	Hyperoxaluria, primary, type 1, 259900
AHCY	100%	100%	100%	99.3%	Hypermethioninemia with deficiency of S-adenosylhomocysteine hydrolase, 613752
AHDC1	100%	100%	100%	98.2%	Xia-Gibbs syndrome, 615829
AHI1	98.7%	98.7%	100%	99.2%	Joubert syndrome 3, 608629
AHNAK2	97.4%	97.3%	97.4%	92.5%	
AHR	100%	100%	100%	99.9%	Foveal hypoplasia 3, 620958;?Retinitis pigmentosa 85, 618345
AHSG	100%	100%	100%	99.4%	?Alopecia-intellectual disability syndrome 1, 203650
AICDA	92.1%	92%	100%	98.8%	Immunodeficiency with hyper-IgM, type 2, 605258

AIFM1	100%	99.7%	99%	73.5%	Combined oxidative phosphorylation deficiency 6, 300816;Cowchock syndrome, 310490;Spondyloepimetaphyseal dysplasia, X-linked, with hypomyelinating leukodystrophy, 300232;Deafness, X-linked 5, 300614
AIMP1	100%	100%	100%	99.6%	Leukodystrophy, hypomyelinating, 3, 260600
AIMP2	100%	100%	100%	98.7%	Leukodystrophy, hypomyelinating, 17, 618006
AIP	100%	100%	100%	99.4%	Pituitary adenoma 1, multiple types, 102200;Pituitary adenoma predisposition, 102200
AIPL1	100%	100%	100%	98.8%	Leber congenital amaurosis 4, 604393;Retinitis pigmentosa, juvenile, 604393;Cone-rod dystrophy, 604393
AIRE	100%	100%	100%	99%	Autoimmune polyendocrinopathy syndrome , type I, with or without reversible metaphyseal dysplasia, 240300
AJAP1	100%	100%	100%	98.6%	

AK1	100%	100%	100%	98.3%	Anemia, congenital, nonspherocytic hemolytic, 3, adenylate kinase deficient, 612631
AK2	100%	100%	100%	99.7%	Reticular dysgenesis, 267500
AK3	100%	100%	100%	99.8%	
AK7	100%	100%	100%	99.6%	?Spermatogenic failure 27, 617965
AK9	100%	100%	100%	99.8%	Spermatogenic failure 89, 620705
AKAP3	100%	100%	100%	99.6%	Spermatogenic failure 82, 620353
AKAP9	100%	100%	100%	99.5%	?Long QT syndrome 11, 611820
AKR1C1	100%	100%	100%	99.7%	
AKR1C2	100%	100%	99.9%	99.1%	46XY sex reversal 8, 614279
AKR1D1	100%	100%	100%	99.8%	Bile acid synthesis defect, congenital, 2, 235555
AKT1	100%	100%	100%	99.2%	Breast cancer, somatic, 114480;Cowden syndrome 6, 615109;Colorectal cancer, somatic, 114500;Proteus syndrome, somatic, 176920;Ovarian cancer, somatic, 167000

AKT2	100%	100%	100%	99.1%	Diabetes mellitus, type II, 125853;Hypoinsulinemic hypoglycemia with hemihypertrophy, 240900
AKT3	94.6%	94.2%	100%	99.7%	Megalencephaly-polymicrogyria-polydactyly-hydrocephalus syndrome 2, 615937
ALAD	100%	100%	100%	99%	Porphyria, acute hepatic, 612740;{Lead poisoning, susceptibility to}, 612740
ALAS2	100%	99.3%	99%	71.3%	Anemia, sideroblastic, 1, 300751;Protoporphyria, erythropoietic, X-linked, 300752
ALB	100%	100%	100%	99.9%	?[Dysalbuminemic hypertriiodothyroninemia], 615999;Analbuminemia, 616000;[Dysalbuminemic hyperthyroxinemia], 615999
ALDH18A1	100%	100%	100%	99.6%	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
ALDH1A2	100%	100%	100%	99.6%	Diaphragmatic hernia 4, with cardiovascular defects, 620025

ALDH1A3	100%	100%	100%	99.5%	Microphthalmia, isolated 8, 615113
ALDH1B1	100%	100%	100%	99.3%	
ALDH2	100%	100%	100%	99.1%	{Esophageal cancer, alcohol-related, susceptibility to};{Sublingual nitroglycerin, susceptibility to poor response to};Alcohol sensitivity, acute, 610251;{Hangover, susceptibility to}, 610251
ALDH3A2	93.5%	93.5%	100%	99.6%	Sjogren-Larsson syndrome, 270200
ALDH4A1	100%	100%	100%	99.1%	Hyperprolinemia, type II, 239510
ALDH5A1	100%	100%	100%	98.9%	Succinic semialdehyde dehydrogenase deficiency, 271980
ALDH6A1	100%	100%	100%	99.8%	Methylmalonate semialdehyde dehydrogenase deficiency, 614105
ALDH7A1	100%	100%	100%	99.4%	Epilepsy, early-onset, 4, vitamin B6-dependent, 266100
ALDOA	100%	100%	100%	99.5%	Glycogen storage disease XII, 611881
ALDOB	100%	100%	100%	99.6%	Fructose intolerance, hereditary, 229600

ALG1	100%	100%	100%	99.1%	Congenital disorder of glycosylation, type Ik, 608540
ALG10	100%	100%	100%	99.2%	
ALG11	91%	91%	100%	99.7%	Congenital disorder of glycosylation, type Ip, 613661
ALG12	100%	100%	100%	98.6%	Congenital disorder of glycosylation, type Ig, 607143
ALG13	100%	99.8%	99.1%	72.6%	Developmental and epileptic encephalopathy 36, 300884
ALG14	100%	100%	100%	99.4%	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
ALG2	100%	100%	100%	99.4%	Congenital disorder of glycosylation, type Ii, 607906;Myasthenic syndrome, congenital, 14, with tubular aggregates, 616228
ALG3	100%	100%	100%	99.4%	Congenital disorder of glycosylation, type Id, 601110

ALG5	100%	100%	100%	99.6%	Polycystic kidney disease 7, 620056
ALG6	100%	100%	100%	99.8%	Congenital disorder of glycosylation, type Ic, 603147
ALG8	77.9%	77.5%	100%	99.5%	Congenital disorder of glycosylation, type Ih, 608104;Polycystic liver disease 3 with or without kidney cysts, 617874
ALG9	100%	100%	100%	99.6%	Gillessen-Kaesbach-Nishimura syndrome, 263210;Congenital disorder of glycosylation, type Il, 608776
ALK	100%	100%	100%	99%	{Neuroblastoma, susceptibility to, 3}, 613014
ALKBH1	100%	100%	100%	99.8%	
ALKBH8	100%	100%	100%	99.8%	Intellectual developmental disorder, autosomal recessive 71, 618504
ALMS1	100%	100%	100%	99.7%	Alstrom syndrome, 203800
ALOX12B	100%	100%	100%	99.1%	Ichthyosis, congenital, autosomal recessive 2, 242100
ALOXE3	100%	100%	100%	98.8%	Ichthyosis, congenital, autosomal recessive 3, 606545
ALPI	100%	100%	100%	98.4%	
ALPK1	100%	100%	100%	99.6%	ROSAH syndrome, 614979

ALPK3	100%	100%	100%	98.9%	Cardiomyopathy, familial hypertrophic 27, 618052
ALPL	100%	100%	100%	98.8%	Odontohypophosphatasia, 146300;Hypophosphatasia, infantile, 241500;Hypophosphatasia, childhood, 241510;Hypophosphatasia, adult, 146300
ALS2	97.1%	97%	100%	99.6%	Primary lateral sclerosis, juvenile, 606353;Spastic paralysis, infantile onset ascending, 607225;Amyotrophic lateral sclerosis 2, juvenile, 205100
ALX1	100%	100%	100%	99.5%	Frontonasal dysplasia 3, 613456
ALX3	100%	99.9%	100%	97.6%	Frontonasal dysplasia 1, 136760
ALX4	100%	100%	100%	98.6%	Parietal foramina 2, 609597;{Craniosynostosis 5, susceptibility to}, 615529;Frontonasal dysplasia 2, 613451
AMACR	100%	100%	100%	99.5%	Alpha-methylacyl-CoA racemase deficiency, 614307;Bile acid synthesis defect, congenital, 4, 214950
AMBN	100%	100%	100%	99.7%	Amelogenesis imperfecta, type IF, 616270

AMELX	100%	100%	98.7%	69.9%	Amelogenesis imperfecta, type 1E, 301200
AMER1	100%	99.9%	98.9%	69.7%	Osteopathia striata with cranial sclerosis, 300373
AMFR	100%	100%	100%	98.4%	Spastic paraplegia 89, autosomal recessive, 620379
AMH	100%	100%	100%	98.6%	Persistent Mullerian duct syndrome, type I, 261550
AMHR2	100%	100%	100%	98.4%	Persistent Mullerian duct syndrome, type II, 261550
AMMECR1	99.4%	96.6%	98.7%	70.5%	Midface hypoplasia, hearing impairment, elliptocytosis, and nephrocalcinosis, 300990
AMN	100%	100%	100%	97.2%	Imerslund-Grasbeck syndrome 2, 618882
AMOTL1	100%	100%	100%	99.3%	
AMPD1	100%	100%	100%	99.5%	Myopathy due to myoadenylate deaminase deficiency, 615511
AMPD2	100%	100%	100%	99.1%	Pontocerebellar hypoplasia, type 9, 615809;?Spastic paraplegia 63, autosomal recessive, 615686
AMPD3	100%	100%	100%	99.1%	[AMP deaminase deficiency, erythrocytic], 612874
AMT	100%	100%	100%	98.8%	Glycine encephalopathy 2, 620398

AMTN	100%	100%	100%	99.7%	?Amelogenesis imperfecta, type IIIB, 617607
ANAPC1	100%	100%	100%	99.4%	Rothmund-Thomson syndrome, type 1, 618625
ANAPC7	100%	100%	100%	98.9%	Ferguson-Bonni neurodevelopmental syndrome, 619699
ANG	100%	100%	100%	99.2%	Amyotrophic lateral sclerosis 9, 611895
ANGPT1	100%	100%	100%	99.7%	?Angioedema, hereditary, 5, 619361
ANGPT2	100%	100%	100%	99.6%	Lymphatic malformation 10, 619369
ANGPTL3	100%	100%	100%	99.6%	Hypobetalipoproteinemia, familial, 2, 605019
ANGPTL4	100%	100%	100%	99.1%	Plasma triglyceride level QTL, low, 615881
ANK1	100%	100%	100%	99.2%	Spherocytosis, type 1, 182900
ANK2	100%	100%	100%	99.6%	Long QT syndrome 4, 600919;Cardiac arrhythmia, ankyrin-B-related, 600919
ANK3	99.7%	99.7%	100%	99.6%	Intellectual developmental disorder, autosomal recessive 37, 615493
ANKFY1	100%	100%	100%	99.3%	
ANKH	100%	100%	100%	99%	Chondrocalcinosis 2, 118600;Craniometaphyseal dysplasia, 123000

ANKLE2	100%	100%	100%	98.6%	Microcephaly 16, primary, autosomal recessive, 616681
ANKRD1	100%	100%	100%	97.6%	
ANKRD11	100%	100%	100%	98.7%	KBG syndrome, 148050
ANKRD17	100%	100%	100%	99.7%	Chopra-Amiel-Gordon syndrome, 619504
ANKRD26	100%	100%	100%	99.7%	Thrombocytopenia 2, 188000
ANKS1B	100%	100%	100%	99.8%	
ANKS6	99.7%	98.6%	100%	99.3%	Nephronophthisis 16, 615382
ANLN	100%	100%	100%	99.6%	Focal segmental glomerulosclerosis 8, 616032
ANO1	100%	100%	100%	98.1%	Moyamoya disease 7, 620687;?Intestinal dysmotility syndrome, 620045
ANO10	100%	100%	100%	99.7%	Spinocerebellar ataxia, autosomal recessive 10, 613728
ANO3	100%	100%	100%	99.7%	Dystonia 24, 615034
ANO4	100%	100%	100%	99.7%	
ANO5	100%	100%	100%	99.7%	Muscular dystrophy, limb-girdle, autosomal recessive 12, 611307;Miyoshi muscular dystrophy 3, 613319;Gnathodiaphyseal dysplasia, 166260

ANO6	98.4%	98.4%	100%	99.6%	Scott syndrome, 262890
ANOS1	100%	99.7%	98.5%	71.5%	Hypogonadotropic hypogonadism 1 with or without anosmia (Kallmann syndrome 1), 308700
ANTXR1	100%	100%	100%	98.5%	GAPO syndrome, 230740;{?Hemangioma, capillary infantile, susceptibility to}, 602089
ANTXR2	96.3%	96.3%	100%	99.4%	Hyaline fibromatosis syndrome, 228600
ANXA11	100%	100%	100%	99.2%	Amyotrophic lateral sclerosis 23, 617839;Inclusion body myopathy and brain white matter abnormalities, 619733
AOPEP	100%	100%	100%	99.3%	Dystonia 31, 619565
AP1B1	100%	100%	100%	99.3%	Keratitis-ichthyosis-deafness syndrome, autosomal recessive, 242150
AP1G1	100%	100%	100%	99.7%	Usmani-Riazuddin syndrome, autosomal recessive, 619548;Usmani-Riazuddin syndrome, autosomal dominant, 619467
AP1S1	100%	100%	100%	99.1%	MEDNIK syndrome, 609313
AP1S2	100%	98.7%	99.2%	76.5%	Pettigrew syndrome, 304340

AP1S3	90.6%	90.6%	100%	98.9%	{Psoriasis 15, pustular, susceptibility to}, 616106
AP2M1	100%	100%	100%	99.4%	Intellectual developmental disorder 60 with seizures, 618587
AP2S1	100%	100%	100%	98.6%	Hypocalciuric hypercalcemia, type III, 600740
AP3B1	100%	100%	100%	99.7%	Hermansky-Pudlak syndrome 2, 608233
AP3B2	100%	100%	100%	98.9%	Developmental and epileptic encephalopathy 48, 617276
AP3D1	100%	100%	100%	99%	?Hermansky-Pudlak syndrome 10, 617050
AP4B1	100%	100%	100%	99.5%	Spastic paraplegia 47, autosomal recessive, 614066
AP4E1	100%	100%	100%	99.7%	Stuttering, familial persistent, 1, 184450;Spastic paraplegia 51, autosomal recessive, 613744
AP4M1	100%	100%	100%	98.7%	Spastic paraplegia 50, autosomal recessive, 612936
AP4S1	87.8%	87.5%	100%	99.6%	Spastic paraplegia 52, autosomal recessive, 614067
AP5Z1	100%	100%	100%	98.7%	Spastic paraplegia 48, autosomal recessive, 613647

APC	100%	100%	100%	99.8%	Colorectal cancer, somatic, 114500;Brain tumor-polyposis 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
APC2	100%	100%	100%	98.1%	Cortical dysplasia, complex, with other brain malformations 10, 618677;Intellectual developmental disorder, autosomal recessive 74, 617169
APCDD1	100%	100%	100%	99.4%	Hypotrichosis 1, 605389
APOA1	100%	100%	100%	99.3%	Hypoalphalipoproteinemia, primary, 2, 618463;Amyloidosis, hereditary systemic 3, 620657;Hypoalphalipoproteinemia, primary, 2, intermediate, 619836

APOA2	100%	100%	100%	98.9%	Apolipoprotein A-II deficiency;{Hypercholesterolemia, familial, modifier of}, 143890
APOA5	100%	100%	100%	99.4%	Hyperchylomicronemia, late-onset, 144650;{Hypertriglyceridemia, susceptibility to}, 145750
APOB	100%	100%	100%	99.6%	Hypercholesterolemia, familial, 2, 144010;Hypobetalipoproteinemia, 615558
APOC2	100%	100%	100%	99.6%	Hyperlipoproteinemia, type Ib, 207750
APOC3	100%	100%	100%	100%	Apolipoprotein C-III deficiency, 614028
APOE	100%	100%	100%	97.9%	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

APOL1	100%	100%	100%	99.4%	{Glomerulosclerosis, focal segmental, 4, susceptibility to}, 612551
APOLD1	100%	100%	99.9%	94.5%	?Bleeding disorder, vascular-type, 620715
APOO	100%	99.4%	99.3%	73.5%	
APP	100%	100%	100%	99.5%	Cerebral amyloid angiopathy, Dutch, Italian, Iowa, Flemish, Arctic variants, 605714;Alzheimer disease 1, familial, 104300
APRT	100%	100%	100%	98.6%	Adenine phosphoribosyltransferase deficiency, 614723
APTX	100%	100%	100%	99.3%	Ataxia, early-onset, with oculomotor apraxia and hypoalbuminemia, 208920
AQP2	100%	100%	100%	99.6%	Diabetes insipidus, nephrogenic, 2, 125800
AQP4	100%	100%	100%	99.8%	?Megalencephalic leukoencephalopathy with subcortical cysts 4, remitting, 620448
AQP5	100%	100%	100%	98.9%	Palmoplantar keratoderma, Bothnian type, 600231

AR	100%	99.6%	97.5%	67.6%	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
ARAF	100%	99.1%	98.4%	67.1%	
ARCN1	100%	100%	100%	99.7%	Short stature-micrognathia syndrome, 617164
ARF1	100%	100%	100%	99.8%	Periventricular nodular heterotopia 8, 618185
ARF3	100%	100%	100%	99.1%	
ARFGEF1	100%	100%	100%	99.6%	Developmental delay, impaired speech, and behavioral abnormalities, with or without seizures, 619964
ARFGEF2	100%	100%	100%	99.6%	Periventricular heterotopia with microcephaly, 608097
ARG1	93%	93%	100%	99.8%	Argininemia, 207800
ARHGAP24	100%	100%	100%	99%	
ARHGAP26	100%	100%	100%	98.8%	Leukemia, juvenile myelomonocytic, somatic, 607785
ARHGAP29	100%	100%	100%	99.6%	

ARHGAP31	100%	100%	100%	99.3%	Adams-Oliver syndrome 1, 100300
ARHGAP35	100%	100%	100%	99.1%	
ARHGDIA	100%	100%	100%	98.8%	Nephrotic syndrome, type 8, 615244
ARHGEF1	100%	99.5%	100%	98.3%	?Immunodeficiency 62, 618459
ARHGEF10	100%	100%	100%	99.5%	?Slowed nerve conduction velocity, AD, 608236
ARHGEF18	100%	100%	100%	98.6%	Retinitis pigmentosa 78, 617433
ARHGEF2	97.9%	97.9%	100%	99.1%	?Neurodevelopmental disorder with midbrain and hindbrain malformations, 617523
ARHGEF28	100%	100%	100%	99.6%	
ARHGEF6	100%	99.9%	99.3%	75.2%	
ARHGEF9	96.9%	96.4%	98.5%	73%	Developmental and epileptic encephalopathy 8, 300607
ARID1A	100%	100%	99.9%	95.6%	Coffin-Siris syndrome 2, 614607
ARID1B	98.5%	98.2%	99.5%	90.8%	Coffin-Siris syndrome 1, 135900
ARID2	100%	100%	100%	99.3%	Coffin-Siris syndrome 6, 617808
ARIH1	100%	100%	100%	99.2%	
ARL13B	93.4%	92.8%	100%	99.7%	Joubert syndrome 8, 612291

ARL2	100%	100%	100%	98.7%	?Microcornea, rod-cone dystrophy, cataract, and posterior staphyloma 1, 619082
ARL2BP	100%	100%	100%	99.3%	Retinitis pigmentosa 82 with or without situs inversus, 615434
ARL3	100%	100%	100%	99.8%	Retinitis pigmentosa 83, 618173;Joubert syndrome 35, 618161
ARL6	100%	100%	100%	99.8%	Retinitis pigmentosa 55, 613575;{Bardet-Biedl syndrome 1, modifier of}, 209900;Bardet-Biedl syndrome 3, 600151
ARL6IP1	100%	100%	100%	99.4%	Spastic paraplegia 61, autosomal recessive, 615685
ARMC12	100%	100%	100%	99.3%	Spermatogenic failure 90, 620744
ARMC2	100%	100%	100%	99.8%	Spermatogenic failure 38, 618433
ARMC5	100%	100%	100%	98.4%	{ACTH-independent macronodular adrenal hyperplasia 2}, 615954
ARMC9	96.2%	93.7%	100%	99.4%	Joubert syndrome 30, 617622
ARNT2	100%	100%	100%	99.2%	?Webb-Dattani syndrome, 615926

ARPC1B	100%	100%	100%	99.2%	Immunodeficiency 71 with inflammatory disease and congenital thrombocytopenia, 617718
ARPC4	100%	100%	100%	99.3%	Developmental delay, language impairment, and ocular abnormalities, 620141
ARPC5	100%	100%	100%	99.1%	Immunodeficiency 133 with autoimmunity and autoinflammation, 620565
ARR3	100%	99.9%	98.5%	70.4%	Myopia 26, X-linked, female-limited, 301010
ARSA	100%	100%	100%	98.7%	Metachromatic leukodystrophy, 250100
ARSB	100%	100%	100%	99.2%	Mucopolysaccharidosis type VI (Maroteaux-Lamy), 253200
ARSG	100%	100%	100%	99.3%	Usher syndrome, type IV, 618144
ARSK	100%	100%	100%	99.5%	Mucopolysaccharidosis, type X, 619698
ARSL	100%	99.8%	98.5%	69.9%	Chondrodysplasia punctata, X-linked recessive, 302950
ARV1	100%	100%	100%	99.8%	Developmental and epileptic encephalopathy 38, 617020

ARX	98.5%	94.3%	91.9%	52.1%	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
ASAH1	100%	100%	100%	99.6%	Spinal muscular atrophy with progressive myoclonic epilepsy, 159950;Farber lipogranulomatosis, 228000
ASB10	100%	100%	100%	99.2%	Glaucoma 1, open angle, F, 603383
ASCC1	86.6%	86.6%	100%	99.7%	Spinal muscular atrophy with congenital bone fractures 2, 616867;Barrett esophagus/esophageal adenocarcinoma, 614266
ASCC3	100%	100%	100%	99.8%	Intellectual developmental disorder, autosomal recessive 81, 620700
ASCL1	100%	100%	100%	95.1%	
ASH1L	100%	100%	100%	99.6%	Intellectual developmental disorder, autosomal dominant 52, 617796

ASIP	100%	100%	100%	99.4%	[Skin/hair/eye pigmentation 9, brown/nonbrown eyes], 611742;[Skin/hair/eye pigmentation 9, dark/light hair], 611742
ASL	100%	100%	100%	98.7%	Argininosuccinic aciduria, 207900
ASNS	100%	100%	100%	99.8%	Asparagine synthetase deficiency, 615574
ASPA	100%	100%	100%	99.8%	Canavan disease, 271900
ASPH	100%	100%	100%	99.7%	Traboulsi syndrome, 601552
ASPM	97.9%	97.9%	100%	99.6%	Microcephaly 5, primary, autosomal recessive, 608716
ASPRV1	100%	100%	100%	99.1%	Ichthyosis, lamellar, autosomal dominant, 146750
ASPSCR1	100%	100%	100%	98.8%	Alveolar soft-part sarcoma, 606243
ASRGL1	100%	100%	100%	99.6%	
ASS1	100%	100%	100%	99.1%	Citrullinemia, 215700
ASTL	100%	100%	100%	99%	?Oocyte/zygote/embryo maturation arrest 11, 619643
ASXL1	100%	100%	100%	99.5%	Myelodysplastic syndrome, somatic, 614286;Bohring-Opitz syndrome, 605039
ASXL2	100%	100%	100%	99.6%	Shashi-Pena syndrome, 617190

ASXL3	100%	100%	100%	99.6%	Bainbridge-Ropers syndrome, 615485
ATAD1	100%	100%	100%	99.7%	Hyperekplexia 4, 618011
ATAD3A	100%	100%	99.9%	97.4%	Harel-Yoon syndrome, 617183;Pontocerebellar hypoplasia, hypotonia, and respiratory insufficiency syndrome, neonatal lethal, 618810
ATAD3B	100%	100%	99.7%	94.9%	
ATCAY	100%	100%	100%	98.9%	Ataxia, cerebellar, Cayman type, 601238
ATF3	100%	100%	100%	99.5%	
ATF6	90.9%	90.9%	100%	99.8%	Achromatopsia 7, 616517
ATG4A	100%	99.9%	99.1%	75.6%	
ATG4D	100%	100%	100%	98.9%	
ATG5	100%	100%	100%	99.9%	?Spinocerebellar ataxia, autosomal recessive 25, 617584
ATG7	100%	100%	100%	99.8%	Spinocerebellar ataxia, autosomal recessive 31, 619422
ATIC	100%	100%	100%	99.5%	AICA-ribosiduria due to ATIC deficiency, 608688
ATL1	100%	100%	100%	99.7%	Spastic paraplegia 3A, autosomal dominant, 182600;Neuropathy, hereditary sensory, type ID, 613708

ATL3	100%	100%	100%	99.5%	Neuropathy, hereditary sensory, type IF, 615632
ATM	100%	100%	100%	99.5%	Lymphoma, B-cell non-Hodgkin, somatic;Ataxia-telangiectasia, 208900;{Breast cancer, susceptibility to}, 114480;T-cell prolymphocytic leukemia, somatic;Lymphoma, mantle cell, somatic
ATN1	100%	100%	99.9%	95.7%	Dentatorubral-pallidoluysian atrophy, 125370;Congenital hypotonia, epilepsy, developmental delay, and digital anomalies, 618494
ATOH1	100%	100%	100%	98.6%	?Deafness, autosomal dominant 89, 620284
ATOH7	100%	99.9%	100%	97%	Persistent hyperplastic primary vitreous, autosomal recessive, 221900
ATP11A	100%	100%	100%	99.4%	?Auditory neuropathy, autosomal dominant 2, 620384;?Leukodystrophy, hypomyelinating, 24, 619851;Deafness, autosomal dominant 84, 619810
ATP11C	100%	99.8%	99.2%	74.5%	?Hemolytic anemia, congenital, X-linked, 301015

ATP13A2	100%	100%	100%	99.2%	Spastic paraplegia 78, autosomal recessive, 617225;Kufor-Rakeb syndrome, 606693
ATP13A3	100%	100%	100%	99.7%	Pulmonary hypertension, primary, 5, 265400
ATP1A1	100%	100%	100%	99.3%	Hypomagnesemia, seizures, and impaired intellectual development 2, 618314;Charcot-Marie-Tooth disease, axonal, type 2DD, 618036
ATP1A2	100%	100%	100%	99.1%	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
ATP1A3	100%	100%	100%	98.5%	Alternating hemiplegia of childhood 2, 614820;Dystonia-12, 128235;CAPOS syndrome, 601338;Developmental and epileptic encephalopathy 99, 619606
ATP2A1	100%	100%	100%	98.7%	Brody myopathy, 601003

ATP2A2	100%	100%	100%	99.2%	Acrokeratosis verruciformis, 101900;Darier disease, 124200
ATP2B1	100%	100%	100%	99.4%	Intellectual developmental disorder, autosomal dominant 66, 619910
ATP2B2	100%	100%	100%	99%	Deafness, autosomal dominant 82, 619804;{Deafness, autosomal recessive 12, modifier of}, 601386
ATP2B3	100%	99.3%	97.9%	67.7%	?Spinocerebellar ataxia, X-linked 1, 302500
ATP2C1	100%	100%	100%	99.7%	Hailey-Hailey disease, 169600
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
ATP5F1B	100%	100%	100%	99.1%	?Hypermetabolism due to uncoupled mitochondrial oxidative phosphorylation 2, 620085
ATP5F1C	100%	100%	100%	99.8%	

ATP5F1D	100%	100%	100%	97.8%	Mitochondrial complex V (ATP synthase) deficiency, 618120
ATP5F1E	100%	100%	100%	99.4%	Mitochondrial complex V (ATP synthase) deficiency, nuclear type 3, 614053
ATP5IF1	100%	100%	100%	99.9%	
ATP5MC1	100%	100%	100%	98.3%	
ATP5MC2	100%	100%	100%	99.7%	
ATP5MC3	100%	100%	100%	99.7%	Dystonia, early-onset, and/or spastic paraplegia, 619681
ATP5ME	100%	100%	100%	98.7%	
ATP5MF	100%	100%	100%	100%	
ATP5MG	95.4%	95.4%	100%	99%	
ATP5MGL	100%	100%	100%	98.5%	
ATP5MK	100%	100%	100%	100%	Mitochondrial complex V (ATP synthase) deficiency, nuclear type 6, 618683
ATP5PB	100%	100%	100%	99.7%	
ATP5PD	100%	100%	100%	99.2%	
ATP5PF	100%	100%	100%	99.9%	
ATP5PO	100%	100%	100%	99.8%	Mitochondrial complex V (ATP synthase) deficiency, nuclear type 7, 620359
ATP6AP1	100%	99.2%	98.2%	70.9%	Immunodeficiency 47, 300972

ATP6AP2	100%	99.7%	99.1%	74.7%	Intellectual developmental disorder, X-linked syndromic, Hedera type, 300423;?Parkinsonism with spasticity, X-linked, 300911;Congenital disorder of glycosylation, type IIr, 301045
ATP6V0A1	92.9%	92.9%	100%	99.5%	Neurodevelopmental disorder with epilepsy and brain atrophy, 619971;Developmental and epileptic encephalopathy 104, 619970
ATP6V0A2	100%	100%	100%	99.5%	Wrinkly skin syndrome, 278250;Cutis laxa, autosomal recessive, type IIA, 219200
ATP6V0A4	100%	100%	100%	99%	Distal renal tubular acidosis 3, with or without sensorineural hearing loss, 602722
ATP6V0C	100%	100%	100%	98.4%	Epilepsy, early-onset, 3, with or without developmental delay, 620465
ATP6V1A	100%	100%	100%	99.9%	Cutis laxa, autosomal recessive, type IID, 617403;Developmental and epileptic encephalopathy 93, 618012

ATP6V1B1	100%	100%	100%	99.3%	Distal renal tubular acidosis 2 with progressive sensorineural hearing loss, 267300
ATP6V1B2	100%	100%	100%	99.6%	Zimmermann-Laband syndrome 2, 616455;Deafness, congenital, with onychodystrophy, autosomal dominant, 124480
ATP6V1E1	100%	100%	100%	99.9%	Cutis laxa, autosomal recessive, type IIC, 617402
ATP7A	94.9%	94.3%	99.4%	73.1%	Occipital horn syndrome, 304150;Neuronopathy, distal hereditary motor, X-linked, 300489;Menkes disease, 309400
ATP7B	100%	100%	100%	99.7%	Wilson disease, 277900
ATP8A2	100%	100%	100%	99.5%	Cerebellar ataxia, impaired intellectual development, and dysequilibrium syndrome 4, 615268
ATP8B1	100%	100%	100%	99.5%	Cholestasis, progressive familial intrahepatic 1, 211600;Cholestasis, intrahepatic, of pregnancy, 1, 147480;Cholestasis, benign recurrent intrahepatic, 243300

ATP9A	100%	100%	100%	99.1%	Neurodevelopmental disorder with poor growth and behavioral abnormalities, 620242
ATPAF1	100%	100%	99.8%	92.2%	
ATPAF2	100%	100%	100%	98.9%	?Mitochondrial complex V (ATP synthase) deficiency, nuclear type 1, 604273
ATR	100%	100%	99.7%	98.7%	Seckel syndrome 1, 210600;?Cutaneous telangiectasia and cancer syndrome, familial, 614564
ATRX	100%	99.9%	99.3%	74.5%	Alpha-thalassemia myelodysplasia syndrome, somatic, 300448;Intellectual disability-hypotonic facies syndrome, X-linked, 309580;Alpha-thalassemia/impaired intellectual development syndrome, 301040
ATXN1	100%	100%	100%	98.5%	Spinocerebellar ataxia 1, 164400
ATXN10	100%	100%	100%	99.6%	Spinocerebellar ataxia 10, 603516
ATXN2	100%	100%	100%	98.7%	{Amyotrophic lateral sclerosis, susceptibility to, 13}, 183090;Spinocerebellar ataxia 2, 183090;{Parkinson disease, late-onset, susceptibility to}, 168600
ATXN2L	100%	100%	100%	97.8%	

ATXN3	96.9%	96.9%	100%	99.5%	{Parkinson disease, late-onset, susceptibility to}, 168600;Machado-Joseph disease, 109150
ATXN7	100%	99.9%	100%	97.9%	Spinocerebellar ataxia 7, 164500
ATXN7L3	100%	100%	100%	98.6%	
ATXN8OS					{Parkinson disease, susceptibility to}, 168600;Spinocerebellar ataxia 8, 608768
AUH	100%	100%	100%	99.9%	3-methylglutaconic aciduria, type I, 250950
AURKC	100%	100%	100%	98.8%	Spermatogenic failure 5, 243060
AUTS2	100%	100%	100%	98.4%	Intellectual developmental disorder, autosomal dominant 26, 615834
AVIL	100%	100%	100%	99.7%	Nephrotic syndrome, type 21, 618594
AVP	100%	100%	100%	99.3%	Diabetes insipidus, neurohypophyseal, 125700
AVPR2	100%	100%	97.8%	67.3%	Diabetes insipidus, nephrogenic, 1, 304800;Nephrogenic syndrome of inappropriate antidiuresis, 300539

AXIN1	100%	100%	100%	99.5%	Hepatocellular carcinoma, somatic, 114550;Craniometadiaphyseal osteosclerosis with hip dysplasia, 620558;?Caudal duplication anomaly, 607864
AXIN2	100%	100%	100%	98.7%	Colorectal cancer, somatic, 114500;Oligodontia-colorectal cancer syndrome, 608615
AXL	100%	100%	100%	98.7%	
B2M	100%	100%	100%	99.8%	Amyloidosis, hereditary systemic 6, 620659;Immunodeficiency 43, 241600
B3GALNT1	100%	100%	100%	99.9%	[Blood group, P1PK system, P(k) phenotype], 111400;[Blood group, globoside system], 615021
B3GALNT2	92.6%	92.6%	100%	99.3%	Muscular dystrophy-dystroglycanopathy (congenital with brain and eye anomalies), type A, 11, 615181
B3GALT6	96.8%	88.3%	100%	95.4%	Ehlers-Danlos syndrome, spondylodysplastic type, 2, 615349;Spondyloepimetaphyseal dysplasia with joint laxity, type 1, with or without fractures, 271640;Al-Gazali syndrome, 609465

B3GAT3	96.6%	93.8%	100%	99.2%	Multiple joint dislocations, short stature, craniofacial dysmorphism, with or without congenital heart defects, 245600
B3GLCT	100%	99.9%	100%	99.6%	Peters-plus syndrome, 261540
B4GALNT1	100%	100%	100%	99%	Spastic paraplegia 26, autosomal recessive, 609195
B4GALNT2	100%	100%	100%	99%	[Blood group, Sid system], 615018;Sd(a) polyagglutination syndrome, 615018
B4GALT1	100%	100%	100%	98.7%	Combined low LDL and fibrinogen, 620364;Congenital disorder of glycosylation, type IId, 607091
B4GALT7	100%	100%	100%	99%	Ehlers-Danlos syndrome, spondylodysplastic type, 1, 130070
B4GAT1	100%	100%	100%	98.4%	Muscular dystrophy-dystroglycanopathy (congenital with brain and eye anomalies), type A, 13, 615287
B9D1	100%	100%	100%	99.5%	?Meckel syndrome 9, 614209;Joubert syndrome 27, 617120

B9D2	100%	100%	100%	99.6%	?Meckel syndrome 10, 614175;Joubert syndrome 34, 614175
BAAT	100%	100%	100%	99.5%	Bile acid conjugation defect 1, 619232
BACH2	100%	100%	100%	98.8%	Immunodeficiency 60 and autoimmunity, 618394
BAG3	100%	100%	100%	99.3%	Cardiomyopathy, dilated, 1HH, 613881;Myopathy, myofibrillar, 6, 612954
BAG5	100%	100%	100%	98.7%	Cardiomyopathy, dilated, 2F, 619747
BANF1	100%	100%	100%	97.6%	Nestor-Guillermo progeria syndrome, 614008
BAP1	100%	100%	100%	99.2%	Kury-Isidor syndrome, 619762;Tumor predisposition syndrome 1, 614327;{Uveal melanoma, susceptibility to, 2}, 606661
BARD1	100%	100%	100%	99.7%	{Breast cancer, susceptibility to}, 114480
BAX	100%	100%	100%	97.1%	Colorectal cancer, somatic, 114500;T-cell acute lymphoblastic leukemia, somatic, 613065
BAZ2B	100%	100%	100%	98.8%	
BBIP1	100%	100%	100%	99.8%	Bardet-Biedl syndrome 18, 615995
BBS1	100%	100%	100%	98.9%	Bardet-Biedl syndrome 1, 209900

BBS10	100%	100%	100%	99.8%	Bardet-Biedl syndrome 10, 615987
BBS12	100%	100%	100%	99.9%	Bardet-Biedl syndrome 12, 615989
BBS2	98%	98%	100%	99.5%	Retinitis pigmentosa 74, 616562;Bardet-Biedl syndrome 2, 615981
BBS4	100%	100%	100%	99.6%	Bardet-Biedl syndrome 4, 615982
BBS5	100%	100%	100%	99.4%	Bardet-Biedl syndrome 5, 615983
BBS7	100%	100%	100%	99.8%	Bardet-Biedl syndrome 7, 615984
BBS9	95.8%	95.8%	100%	99.7%	Bardet-Biedl syndrome 9, 615986
BCAP31	98.9%	86.5%	98.5%	69.8%	Deafness, dystonia, and cerebral hypomyelination, 300475
BCAS3	100%	100%	100%	99.6%	Hengel-Maroofian-Schols syndrome, 619641
BCAT1	100%	100%	100%	99.1%	
BCAT2	100%	100%	100%	98.5%	Hypervalinemia and hyperleucine-isoleucinemia, 618850
BCHE	100%	100%	100%	99.7%	Butyrylcholinesterase deficiency, 617936;{Apnea, postanesthetic, susceptibility to, due to BCHE deficiency}, 617936

BCKDHA	100%	100%	100%	98.4%	Maple syrup urine disease, type Ia, 248600
BCKDHB	100%	100%	100%	99.2%	Maple syrup urine disease, type Ib, 620698
BCKDK	100%	100%	100%	98.8%	Branched-chain keto acid dehydrogenase kinase deficiency, 614923
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
BCL11A	100%	100%	100%	98.6%	Dias-Logan syndrome, 617101
BCL11B	99.9%	99.3%	100%	94.8%	Immunodeficiency 49, severe combined, 617237;Intellectual developmental disorder with dysmorphic facies, speech delay, and T-cell abnormalities, 618092
BCL2	92.5%	92.5%	100%	97.6%	Leukemia/lymphoma, B-cell, 2
BCL7A	100%	99.9%	100%	96.2%	

BCO1	100%	100%	100%	99.2%	?Hypercarotenemia and vitamin A deficiency, autosomal dominant, 115300
BCOR	100%	99.6%	98.6%	69.2%	Microphthalmia, syndromic 2, 300166
BCORL1	100%	99%	97.4%	65.1%	Shukla-Vernon syndrome, 301029
BCS1L	100%	100%	100%	99.4%	GRACILE syndrome, 603358;Mitochondrial complex III deficiency, nuclear type 1, 124000;Bjornstad syndrome, 262000
BDP1	100%	100%	100%	99.7%	?Deafness, autosomal recessive 112, 618257
BEAN1	100%	100%	100%	99%	Spinocerebellar ataxia 31, 117210
BEST1	100%	99.9%	100%	98.5%	Macular dystrophy, vitelliform, 2, 153700;?Microcornea, rod-cone dystrophy, cataract, and posterior staphyloma 2, 193220;Retinitis pigmentosa-50, 613194;Retinitis pigmentosa, concentric, 613194;Vitreoretinochoroidopathy, 193220;Bestrophinopathy, autosomal recessive, 611809

BET1	100%	100%	100%	99.8%	Muscular dystrophy, congenital, with rapid progression, 254100
BFSP1	100%	100%	100%	99%	Cataract 33, multiple types, 611391
BFSP2	100%	100%	100%	98.7%	Cataract 12, multiple types, 611597
BGN	100%	99.7%	98.9%	68.6%	Meester-Loeys syndrome, 300989;Spondyloepimetaphyseal dysplasia, X-linked, 300106
BHLHA9	100%	100%	100%	98.5%	?Camptosynpolydactyly, complex, 607539;Syndactyly, mesoaxial synostotic, with phalangeal reduction, 609432
BHLHE22	100%	100%	100%	92%	
BICC1	100%	100%	100%	99.6%	{Renal dysplasia, cystic, susceptibility to}, 601331
BICD1	100%	100%	100%	99.5%	
BICD2	100%	100%	100%	99.4%	Spinal muscular atrophy, lower extremity-predominant, 2B, autosomal dominant, 618291;Spinal muscular atrophy, lower extremity-predominant, 2A, autosomal dominant, 615290
BICRA	100%	100%	100%	97.3%	Coffin-Siris syndrome 12, 619325

BIN1	100%	100%	100%	98.6%	Centronuclear myopathy 2, 255200
BLK	100%	100%	100%	99.2%	Maturity-onset diabetes of the young, type 11, 613375
BLM	96.8%	96.6%	100%	99.7%	Bloom syndrome, 210900
BLNK	94.4%	94.4%	100%	99.5%	?Agammaglobulinemia 4, 613502
BLOC1S1	100%	100%	100%	99.7%	
BLOC1S3	100%	100%	100%	99.7%	Hermansky-Pudlak syndrome 8, 614077
BLOC1S5	100%	100%	100%	98.9%	Hermansky-Pudlak syndrome 11, 619172
BLOC1S6	100%	100%	100%	99.6%	Hermansky-Pudlak syndrome 9, 614171
BLTP1	100%	100%	100%	99.7%	Alkuraya-Kucinskas syndrome, 617822
BLVRA	100%	100%	100%	99.9%	Hyperbiliverdinemia, 614156
BMP1	100%	100%	100%	99%	Osteogenesis imperfecta, type XIII, 614856
BMP15	100%	99.7%	98.8%	72.8%	Premature ovarian failure 4, 300510;Ovarian dysgenesis 2, 300510

BMP2	100%	100%	100%	98.1%	Short stature, facial dysmorphism, and skeletal anomalies with or without cardiac anomalies 1, 617877;Brachydactyly, type A2, 112600;{HFE hemochromatosis, modifier of}, 235200
BMP4	100%	100%	100%	98.8%	Orofacial cleft 11, 600625;Microphthalmia, syndromic 6, 607932
BMP5	100%	99.9%	100%	99.5%	
BMP6	100%	99.9%	100%	99.1%	{Iron overload, susceptibility to}, 620121
BMP7	100%	100%	100%	98.8%	
BMPER	100%	100%	100%	99.7%	Diaphanospondylodysostosis, 608022
BMPR1A	100%	100%	100%	99.5%	Polyposis syndrome, hereditary mixed, 2, 610069;Polyposis, juvenile intestinal, 174900
BMPR1B	100%	100%	100%	99.9%	Acromesomelic dysplasia 3, 609441;Brachydactyly, type A2, 112600;Brachydactyly, type A1, D, 616849

BMPR2	100%	100%	100%	99.7%	Pulmonary hypertension, familial primary, 1, with or without HHT, 178600;Pulmonary hypertension, primary, fenfluramine or dexfenfluramine-associated, 178600;Pulmonary venoocclusive disease 1, 265450
BMS1	100%	99.9%	100%	99.4%	?Aplasia cutis congenita, nonsyndromic, 107600
BNC1	100%	99.7%	100%	99.6%	?Premature ovarian failure 16, 618723
BNC2	100%	100%	100%	99.3%	Lower urinary tract obstruction, congenital, 618612
BOLA1	100%	100%	100%	99.6%	
BOLA2	100%	100%	100%	100%	
BOLA3	100%	100%	100%	98.7%	Multiple mitochondrial dysfunctions syndrome 2 with hyperglycinemia, 614299
BORCS8	82.8%	82.8%	100%	99%	Neurodegeneration, infantile-onset, with optic atrophy and brain abnormalities, 620987
BPGM	100%	100%	100%	99.7%	Erythrocytosis, familial, 8, 222800

BPNT2	100%	100%	100%	98.8%	Chondrodysplasia with joint dislocations, GPAPP type, 614078
BPTF	100%	100%	100%	98.8%	Neurodevelopmental disorder with dysmorphic facies and distal limb anomalies, 617755;{Kaposi sarcoma, susceptibility to}, 148000
BPY2	50%	50%	49.2%	24.8%	
BPY2B	50%	50%	48.1%	27.8%	
BPY2C	50%	50%	48%	26.1%	
BRAF	100%	100%	99.1%	96.2%	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
BRAT1	100%	100%	100%	99.2%	Neurodevelopmental disorder with cerebellar atrophy and with or without seizures, 618056;Rigidity and multifocal seizure syndrome, lethal neonatal, 614498

BRCA1	100%	100%	100%	99.7%	Fanconi anemia, complementation group S, 617883;{Breast-ovarian cancer, familial, 1}, 604370;{Pancreatic cancer, susceptibility to, 4}, 614320
BRCA2	100%	100%	100%	99.7%	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
BRD4	100%	100%	100%	96.3%	Cornelia de Lange syndrome 6, 620568
BRDT	100%	100%	100%	99.8%	?Spermatogenic failure 21, 617644
BRF1	100%	100%	100%	99.1%	Cerebellofaciodental syndrome, 616202
BRIP1	96%	96%	100%	99.7%	Fanconi anemia, complementation group J, 609054;{Breast cancer, early-onset, susceptibility to}, 114480
BRPF1	100%	99.9%	100%	99.1%	Intellectual developmental disorder with dysmorphic facies and ptosis, 617333

BRSK2	100%	99.9%	100%	98%	
BRWD1	100%	100%	100%	99.6%	Ciliary dyskinesia, primary, 51, 620438
BRWD3	100%	99.9%	99%	73.4%	Intellectual developmental disorder, X-linked 93, 300659
BSCL2	100%	100%	100%	98.9%	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
BSND	100%	99.9%	100%	99.2%	Sensorineural deafness with mild renal dysfunction, 602522;Bartter syndrome, type 4a, 602522
BTD	94.2%	94.2%	100%	99.7%	Biotinidase deficiency, 253260
BTG4	100%	100%	100%	99.7%	Oocyte/zygote/embryo maturation arrest 8, 619009
BTK	100%	99.7%	99%	71.9%	Agammaglobulinemia, X-linked 1, 300755;Isolated growth hormone deficiency, type III, with agammaglobulinemia, 307200
BTRC	100%	100%	100%	99.4%	

BUB1	100%	100%	100%	99.7%	Colorectal cancer with chromosomal instability, somatic, 114500;Microcephaly 30, primary, autosomal recessive, 620183
BUB1B	100%	100%	100%	99.9%	Colorectal cancer, somatic, 114500;[Premature chromatid separation trait], 176430;Mosaic variegated aneuploidy syndrome 1, 257300
BUB3	100%	100%	100%	99.6%	
BVES	100%	100%	100%	99.5%	
C11orf80	92.2%	92.2%	100%	99.5%	
C12orf4	100%	100%	100%	99.8%	
C12orf57	100%	100%	100%	98.1%	Temtamy syndrome, 218340
C14orf39	100%	100%	100%	99.8%	Spermatogenic failure 52, 619202;?Premature ovarian failure 18, 619203
C18orf32	100%	100%	100%	99.4%	?Glycosylphosphatidylinositol biosynthesis defect 25, 619985
C19orf12	100%	100%	100%	98.5%	Neurodegeneration with brain iron accumulation 4, 614298;?Spastic paraplegia 43, autosomal recessive, 615043

C1GALT1C1	100%	99.9%	98.8%	75%	Hemolytic uremic syndrome, atypical, 8, with rhizomelic short stature, 301110;Tn polyagglutination syndrome, somatic, 300622
C1QA	77%	74%	100%	99.6%	C1q deficiency 1, 613652
C1QB	77.3%	76.6%	100%	98.2%	C1q deficiency 2, 620321
C1QBP	100%	100%	100%	99.3%	Combined oxidative phosphorylation deficiency 33, 617713
C1QC	99.7%	97.8%	100%	99.3%	C1q deficiency 3, 620322
C1QTNF5	100%	100%	100%	98.2%	Retinal degeneration, late-onset, autosomal dominant, 605670
C1R	100%	100%	100%	99.3%	Ehlers-Danlos syndrome, periodontal type, 1, 130080
C1S	100%	100%	100%	99.5%	C1s deficiency, 613783;Ehlers-Danlos syndrome, periodontal type, 2, 617174
C2	100%	100%	100%	99.6%	C2 deficiency, 217000;{Macular degeneration, age-related, 14, reduced risk of}, 615489
C2CD3	96%	96%	100%	99.3%	Orofaciodigital syndrome XIV, 615948
C2CD6	100%	100%	100%	99.7%	?Spermatogenic failure 68, 619805

C2orf69	100%	100%	100%	99.5%	Combined oxidative phosphorylation deficiency 53, 619423
C3	97.5%	97.5%	100%	98.1%	C3 deficiency, 613779;{Hemolytic uremic syndrome, atypical, susceptibility to, 5}, 612925;{Macular degeneration, age-related, 9}, 611378
C3orf52	100%	100%	100%	99.5%	Hypotrichosis 15, 620177
C4A	99.9%	99.6%	98.7%	87.5%	[Blood group, Rodgers], 614374;C4a deficiency, 614380
C4B	99.8%	99.5%	98.8%	89.4%	C4B deficiency, 614379
C5	100%	100%	100%	99.8%	C5 deficiency, 609536;[Eculizumab, poor response to], 615749
C6	100%	100%	100%	99.8%	C6 deficiency, 612446
C7	98.2%	97.2%	100%	99.6%	C7 deficiency, 610102
C8A	100%	100%	100%	99.7%	C8 deficiency, type I, 613790
C8B	100%	100%	100%	99.9%	C8 deficiency, type II, 613789
C8G	100%	100%	100%	98.3%	
C9	99.3%	99.3%	100%	99.8%	C9 deficiency, 613825;{Macular degeneration, age-related, 15, susceptibility to}, 615591

C9orf72	100%	100%	100%	99.7%	Frontotemporal dementia and/or amyotrophic lateral sclerosis 1, 105550
CA12	100%	100%	100%	99.1%	Hyperchlorhidrosis, isolated, 143860
CA2	100%	100%	100%	99.8%	Osteopetrosis, autosomal recessive 3, with renal tubular acidosis, 259730
CA4	100%	100%	100%	99%	
CA5A	100%	100%	100%	98.9%	Hyperammonemia due to carbonic anhydrase VA deficiency, 615751
CA8	100%	100%	100%	99.8%	Spinocerebellar ataxia, autosomal recessive 34, 613227
CABIN1	100%	100%	100%	99.2%	
CABP2	100%	100%	100%	97%	Deafness, autosomal recessive 93, 614899
CABP4	100%	100%	100%	98.2%	Cone-rod synaptic disorder, congenital nonprogressive, 610427
CACHD1	100%	100%	100%	99.6%	

CACNA1A	100%	100%	100%	98%	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
CACNA1B	100%	100%	100%	98%	Neurodevelopmental disorder with seizures and nonepileptic hyperkinetic movements, 618497
CACNA1C	100%	100%	100%	99.3%	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
CACNA1D	100%	100%	100%	99.4%	Primary aldosteronism, seizures, and neurologic abnormalities, 615474;Sinoatrial node dysfunction and deafness, 614896
CACNA1E	100%	100%	100%	99.2%	Developmental and epileptic encephalopathy 69, 618285

CACNA1F	100%	99.6%	98.5%	67.5%	Cone-rod dystrophy, X-linked, 3, 300476;Night blindness, congenital stationary (incomplete), 2A, X-linked, 300071;Aland Island eye disease, 300600
CACNA1G	100%	100%	100%	98.8%	Spinocerebellar ataxia 42, 616795;Spinocerebellar ataxia 42, early-onset, severe, with neurodevelopmental deficits, 618087
CACNA1H	99.6%	99%	100%	97.9%	{Epilepsy, childhood absence, susceptibility to, 6}, 611942;Hyperaldosteronism, familial, type IV, 617027;{Epilepsy, idiopathic generalized, susceptibility to, 6}, 611942
CACNA1I	100%	100%	99.8%	96.9%	Neurodevelopmental disorder with speech impairment and with or without seizures, 620114

CACNA1S	100%	100%	100%	99%	{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
CACNA2D1	100%	100%	100%	99.3%	Developmental and epileptic encephalopathy 110, 620149
CACNA2D2	100%	100%	100%	98.9%	Cerebellar atrophy with seizures and variable developmental delay, 618501
CACNA2D4	100%	100%	100%	99.4%	Retinal cone dystrophy 4, 610478
CACNB2	100%	100%	100%	99%	Brugada syndrome 4, 611876
CACNB4	100%	100%	100%	99.6%	{Epilepsy, juvenile myoclonic, susceptibility to, 6}, 607682;?Episodic ataxia, type 5, 613855;{Epilepsy, idiopathic generalized, susceptibility to, 9}, 607682
CACNG2	100%	100%	100%	98.4%	?Intellectual developmental disorder, autosomal dominant 10, 614256

CAD	100%	100%	100%	98.8%	Developmental and epileptic encephalopathy 50, 616457
CADM3	100%	100%	100%	99.4%	Charcot-Marie-Tooth disease, axonal, type 2FF, 619519
CALCRL	100%	100%	100%	99.8%	?Lymphatic malformation 8, 618773
CALM1	100%	100%	100%	99.8%	Ventricular tachycardia, catecholaminergic polymorphic, 4, 614916;Long QT syndrome 14, 616247
CALM2	73.5%	73.5%	100%	98.8%	Long QT syndrome 15, 616249
CALM3	100%	100%	100%	99.4%	Long QT syndrome 16, 618782;?Ventricular tachycardia, catecholaminergic polymorphic 6, 618782
CALR	100%	100%	100%	99.3%	Myelofibrosis, somatic, 254450;Thrombocythemia, somatic, 187950
CALR3	100%	100%	100%	99.3%	
CAMK2A	100%	100%	100%	98.9%	Intellectual developmental disorder, autosomal dominant 53, 617798;?Intellectual developmental disorder, autosomal recessive 63, 618095

CAMK2B	100%	100%	100%	98.5%	Intellectual developmental disorder, autosomal dominant 54, 617799
CAMK2D	100%	100%	100%	99.9%	
CAMK2G	100%	100%	100%	99.3%	Intellectual developmental disorder, autosomal dominant 59, 618522
CAMK4	100%	100%	100%	99.4%	
CAMLG	100%	100%	100%	99.1%	?Congenital disorder of glycosylation, type IIz, 620201
CAMSAP1	100%	100%	100%	98.7%	Cortical dysplasia, complex, with other brain malformations 12, 620316
CAMTA1	100%	100%	100%	99.2%	Cerebellar dysfunction with variable cognitive and behavioral abnormalities, 614756
CANT1	100%	100%	100%	99.2%	Desbuquois dysplasia 1, 251450;Epiphyseal dysplasia, multiple, 7, 617719
CAP2	100%	100%	100%	99.5%	Cardiomyopathy, dilated, 2I, 620462
CAPN1	100%	100%	100%	99.1%	Spastic paraplegia 76, autosomal recessive, 616907
CAPN10	100%	100%	100%	98.9%	{Diabetes mellitus, noninsulin-dependent 1}, 601283

CAPN12	100%	100%	100%	97.8%	
CAPN15	100%	100%	100%	98.2%	Oculogastrointestinal neurodevelopmental syndrome, 619318
CAPN3	100%	100%	100%	99.4%	Muscular dystrophy, limb-girdle, autosomal recessive 1, 253600;Muscular dystrophy, limb-girdle, autosomal dominant 4, 618129
CAPN5	100%	100%	100%	99.1%	Vitreoretinopathy, neovascular inflammatory, 193235
CAPNS1	100%	100%	100%	97.9%	Pulmonary hypertension, primary, 6, 620777
CAPRIN1	100%	100%	100%	97.7%	Neurodevelopmental disorder with language impairment, autism, and attention deficit-hyperactivity disorder, 620782;Neurodegeneration, childhood-onset, with cerebellar ataxia and cognitive decline, 620636
CAPZA2	100%	100%	100%	99.5%	
CARD10	100%	100%	100%	98.7%	?Immunodeficiency 89 and autoimmunity, 619632

CARD11	100%	100%	100%	99.1%	B-cell expansion with NFKB and T-cell anergy, 616452;Immunodeficiency 11B with atopic dermatitis, 617638;Immunodeficiency 11A, 615206
CARD14	100%	100%	100%	99.4%	Psoriasis 2, 602723;Pityriasis rubra pilaris, 173200
CARD8	100%	100%	100%	99.8%	?Inflammatory bowel disease (Crohn disease) 30, 619079
CARD9	100%	100%	100%	99.2%	Immunodeficiency 103, susceptibility to fungal infection, 212050
CARMIL2	100%	99.9%	100%	98.3%	Immunodeficiency 58, 618131
CARS1	100%	99.9%	100%	99.2%	Microcephaly, developmental delay, and brittle hair syndrome, 618891
CARS2	100%	100%	100%	99.2%	Combined oxidative phosphorylation deficiency 27, 616672

CASK	100%	99.8%	99.2%	75.1%	Intellectual developmental disorder, with or without nystagmus, 300422;Intellectual developmental disorder and microcephaly with pontine and cerebellar hypoplasia, 300749;FG syndrome 4, 300422
CASP10	100%	99.5%	100%	99.6%	Autoimmune lymphoproliferative syndrome, type II, 603909;Gastric cancer, somatic, 613659;Lymphoma, non-Hodgkin, somatic, 605027
CASP14	100%	100%	100%	99.3%	Ichthyosis, congenital, autosomal recessive 12, 617320
CASP2	100%	100%	100%	99.6%	Intellectual developmental disorder, autosomal recessive 80, with variant lissencephaly, 620653
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
CASQ1	100%	100%	100%	98.9%	Myopathy, vacuolar, with CASQ1 aggregates, 616231

CASQ2	100%	100%	100%	99.7%	Ventricular tachycardia, catecholaminergic polymorphic, 2, 611938
CASR	100%	100%	100%	99.4%	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
CAST	100%	100%	100%	99.6%	Peeling skin with leukonychia, acral punctate keratoses, cheilitis, and knuckle pads, 616295
CASZ1	99.6%	97.3%	100%	96.7%	
CAT	100%	100%	100%	99.9%	Acatalasemia, 614097
CATIP	100%	100%	100%	99.4%	?Spermatogenic failure 54, 619379
CATSPER1	100%	100%	100%	98.4%	Spermatogenic failure 7, 612997
CATSPER2	100%	100%	99.9%	96.6%	

CAV1	74.7%	74.6%	100%	99%	Lipodystrophy, congenital generalized, type 3, 612526;Pulmonary hypertension, primary, 3, 615343;Lipodystrophy, familial partial, type 7, 606721
CAV3	100%	100%	100%	99.3%	Myopathy, distal, Tateyama type, 614321;Creatine phosphokinase, elevated serum, 123320;Cardiomyopathy, familial hypertrophic, 192600;Rippling muscle disease 2, 606072;Long QT syndrome 9, 611818
CAVIN1	100%	100%	100%	97.2%	Lipodystrophy, congenital generalized, type 4, 613327
CAVIN2	100%	100%	100%	99.4%	
CBFB	100%	100%	100%	99%	Cleidocranial dysplasia 2, 620099
CBL	100%	100%	100%	99%	Noonan syndrome-like disorder with or without juvenile myelomonocytic leukemia, 613563;?Juvenile myelomonocytic leukemia, 607785
CBLB	100%	100%	100%	99.5%	Autoimmune disease, multisystem, infantile-onset, 3, 620430
CBLIF	100%	100%	100%	99.5%	Intrinsic factor deficiency, 261000

CBS	100%	100%	100%	99.2%	Thrombosis, hyperhomocysteinemic, 236200;Homocystinuria, B6-responsive and nonresponsive types, 236200
CBX1	100%	100%	100%	99.9%	
CBX2	100%	100%	100%	98.7%	?46XY sex reversal 5, 613080
CBY1	100%	100%	100%	99.1%	
CC2D1A	100%	100%	100%	98.4%	Intellectual developmental disorder, autosomal recessive 3, 608443
CC2D2A	98.2%	98.2%	100%	99.6%	COACH syndrome 2, 619111;Retinitis pigmentosa 93, 619845;Meckel syndrome 6, 612284;Joubert syndrome 9, 612285
CCBE1	100%	100%	100%	99.5%	Hennekam lymphangiectasia-lymphedema syndrome 1, 235510
CCDC103	100%	100%	100%	98.8%	
CCDC115	100%	100%	100%	98.6%	Congenital disorder of glycosylation, type IIo, 616828
CCDC134	100%	100%	100%	99.5%	Osteogenesis imperfecta, type XXII, 619795
CCDC141	100%	100%	100%	99.8%	

CCDC146	100%	100%	100%	99.6%	Spermatogenic failure 94, 620850
CCDC174	100%	100%	100%	99.6%	Hypotonia, infantile, with psychomotor retardation, 616816
CCDC186	100%	100%	100%	99.8%	
CCDC22	100%	99.4%	97.7%	68.3%	Ritscher-Schinzel syndrome 2, 300963
CCDC28B	100%	100%	100%	97.7%	{Bardet-Biedl syndrome 1, modifier of}, 209900
CCDC32	100%	100%	100%	97.8%	Cardiofacioneurodevelopmental syndrome, 619123
CCDC34	100%	100%	100%	99.7%	Spermatogenic failure 76, 620084
CCDC39	100%	100%	100%	99.7%	Ciliary dyskinesia, primary, 14, 613807
CCDC40	100%	100%	100%	97.8%	Ciliary dyskinesia, primary, 15, 613808
CCDC47	100%	100%	100%	99.4%	Trichohepatoneurodevelopmental syndrome, 618268
CCDC50	100%	100%	100%	99.4%	?Deafness, autosomal dominant 44, 607453
CCDC62	100%	100%	100%	99.3%	?Spermatogenic failure 67, 619803
CCDC65	100%	100%	100%	99.7%	Ciliary dyskinesia, primary, 27, 615504
CCDC78	100%	100%	100%	99%	?Centronuclear myopathy 4, 614807
CCDC8	99.8%	97.2%	100%	98.6%	3-M syndrome 3, 614205

CCDC88A	97.4%	97.4%	100%	99.4%	PEHO syndrome-like, 617507
CCDC88C	100%	100%	100%	98.9%	?Spinocerebellar ataxia 40, 616053;Hydrocephalus, congenital, 1, 236600
CCIN	100%	100%	100%	99.7%	Spermatogenic failure 91, 620838
CCM2	100%	100%	100%	98.8%	Cerebral cavernous malformations-2, 603284
CCN2	100%	100%	100%	98.8%	
CCN6	100%	100%	100%	99.6%	Progressive pseudorheumatoid dysplasia, 208230
CCNB3	100%	99.8%	99.5%	76.1%	
CCND2	100%	100%	100%	99.4%	Megalencephaly-polymicrogyria-polydactyly-hydrocephalus syndrome 3, 615938
CCNF	100%	100%	100%	98%	Frontotemporal dementia and/or amyotrophic lateral sclerosis 5, 619141
CCNK	100%	98.3%	97.3%	86.6%	?Intellectual developmental disorder with hypertelorism and distinctive facies, 618147
CCNO	100%	100%	100%	99.7%	Ciliary dyskinesia, primary, 29, 615872
CCNQ	100%	99.7%	98.4%	70.2%	STAR syndrome, 300707

CCR2	100%	100%	100%	99.7%	{HIV infection, susceptibility/resistance to}, 609423;Polycystic lung disease, 219600
CCT2	100%	100%	100%	99.8%	
CCT3	100%	100%	100%	99.5%	
CCT5	100%	100%	100%	99.5%	?Neuropathy, hereditary sensory, with spastic paraplegia, 256840
CD151	100%	100%	100%	98.4%	[Blood group, Raph], 179620;Epidermolysis bullosa simplex 7, with nephropathy and deafness, 609057
CD164	100%	100%	100%	99.5%	?Deafness, autosomal dominant 66, 616969
CD19	100%	100%	100%	98.4%	Immunodeficiency, common variable, 3, 613493
CD247	77.5%	71.7%	100%	99.5%	?Immunodeficiency 25, 610163
CD27	100%	100%	100%	99%	Lymphoproliferative syndrome 2, 615122
CD28	100%	100%	100%	99.6%	?Immunodeficiency 123 with HPV-related verrucosis, 620901
CD2AP	100%	100%	100%	99.7%	Glomerulosclerosis, focal segmental, 3, 607832

CD320	100%	100%	100%	99.3%	Methylmalonic aciduria, transient, due to transcobalamin receptor defect, 613646
CD36	100%	100%	100%	99.8%	Platelet glycoprotein IV deficiency, 608404;{Coronary heart disease, susceptibility to, 7}, 610938;{Malaria, cerebral, susceptibility to}, 611162;{Malaria, cerebral, reduced risk of}, 611162
CD3D	100%	100%	100%	99.2%	Immunodeficiency 19, severe combined, 615617
CD3E	100%	100%	100%	99.2%	Immunodeficiency 18, 615615;Immunodeficiency 18, SCID variant, 615615
CD3G	100%	100%	100%	98.7%	Immunodeficiency 17, CD3 gamma deficient, 615607
CD4	100%	100%	100%	97.6%	Immunodeficiency 79, 619238;OKT4 epitope deficiency, 613949
CD40	100%	100%	100%	99.1%	Immunodeficiency with hyper-IgM, type 3, 606843
CD40LG	100%	99.9%	99.5%	76%	Immunodeficiency, X-linked, with hyper-IgM, 308230
CD46	100%	100%	100%	99.9%	{Hemolytic uremic syndrome, atypical, susceptibility to, 2}, 612922
CD48	100%	100%	100%	99.3%	

CD55	96.4%	91.2%	100%	99.3%	[Blood group Cromer], 613793;Complement hyperactivation, angiopathic thrombosis, and protein-losing enteropathy, 226300
CD59	100%	100%	100%	99.9%	Hemolytic anemia, CD59-mediated, with or without immune-mediated polyneuropathy, 612300
CD70	100%	100%	99.9%	97.5%	Lymphoproliferative syndrome 3, 618261
CD79A	100%	99.4%	100%	98.4%	Agammaglobulinemia 3, 613501
CD79B	100%	100%	100%	99.5%	Agammaglobulinemia 6, 612692
CD81	100%	99.1%	100%	98%	Immunodeficiency, common variable, 6, 613496
CD8A	100%	100%	100%	98.4%	Immunodeficiency 116, 608957
CD96	100%	100%	100%	99.8%	C syndrome, 211750
CDAN1	100%	100%	100%	98.4%	Dyserythropoietic anemia, congenital, type Ia, 224120
CDC14A	100%	100%	100%	99.7%	Deafness, autosomal recessive 32, with or without immotile sperm, 608653
CDC20	100%	100%	100%	99%	Oocyte/zygote/embryo maturation arrest 14, 620276
CDC40	100%	100%	100%	99.8%	?Pontocerebellar hypoplasia, type 15, 619302

CDC42	100%	100%	100%	99.8%	Takenouchi-Kosaki syndrome, 616737
CDC42BPB	100%	100%	100%	99%	Chilton-Okur-Chung neurodevelopmental syndrome, 619841
CDC45	100%	100%	100%	99.2%	Meier-Gorlin syndrome 7, 617063
CDC6	100%	100%	100%	99.1%	?Meier-Gorlin syndrome 5, 613805
CDC73	100%	100%	100%	99.5%	Hyperparathyroidism, familial primary, 145000;Parathyroid adenoma with cystic changes, 145001;Parathyroid carcinoma, 608266;Hyperparathyroidism-jaw tumor syndrome, 145001
CDCA7	100%	100%	100%	99.5%	Immunodeficiency-centromeric instability-facial anomalies syndrome 3, 616910

CDH1	100%	100%	100%	99.3%	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
CDH11	100%	100%	100%	99.5%	Teebi hypertelorism syndrome 2, 619736;Elsahy-Waters syndrome, 211380
CDH15	100%	100%	100%	98.3%	Intellectual developmental disorder, autosomal dominant 3, 612580
CDH2	100%	100%	100%	99.4%	Arrhythmogenic right ventricular dysplasia 14, 618920;?Attention deficit-hyperactivity disorder 8, 619957;Agenesis of corpus callosum, cardiac, ocular, and genital syndrome, 618929
CDH23	100%	100%	100%	99.2%	Usher syndrome, type 1D, 601067;{Pituitary adenoma 5, multiple types}, 617540;Usher syndrome, type 1D/F digenic, 601067;Deafness, autosomal recessive 12, 601386

CDH3	100%	100%	100%	99.2%	Hypotrichosis, congenital, with juvenile macular dystrophy, 601553;Ectodermal dysplasia, ectrodactyly, and macular dystrophy, 225280
CDH4	100%	100%	100%	98.8%	
CDHR1	100%	100%	100%	99.5%	Macular dystrophy, retinal, 613660;Cone-rod dystrophy 15, 613660;Retinitis pigmentosa 65, 613660
CDIN1	100%	100%	100%	99.6%	Dyserythropoietic anemia, congenital, type Ib, 615631
CDK10	100%	100%	100%	98.9%	Al Kaissi syndrome, 617694
CDK13	100%	100%	100%	99.2%	Congenital heart defects, dysmorphic facial features, and intellectual developmental disorder, 617360
CDK19	100%	100%	100%	98.9%	Developmental and epileptic encephalopathy 87, 618916
CDK4	100%	100%	100%	98.9%	{Melanoma, cutaneous malignant, 3}, 609048
CDK5	100%	100%	100%	97.8%	?Lissencephaly 7 with cerebellar hypoplasia, 616342
CDK5RAP2	100%	100%	100%	99.3%	Microcephaly 3, primary, autosomal recessive, 604804

CDK6	100%	100%	100%	98.5%	?Microcephaly 12, primary, autosomal recessive, 616080
CDK8	100%	100%	100%	99.6%	Intellectual developmental disorder with hypotonia and behavioral abnormalities, 618748
CDKL5	95.7%	95.2%	98.6%	70%	Developmental and epileptic encephalopathy 2, 300672
CDKN1A	100%	100%	100%	99.7%	
CDKN1B	100%	100%	100%	99.7%	Multiple endocrine neoplasia, type IV, 610755
CDKN1C	100%	100%	100%	97%	IMAGE syndrome, 614732;Beckwith-Wiedemann syndrome, 130650
CDKN2A	100%	100%	100%	97.5%	{Melanoma and neural system tumor syndrome}, 155755;{Melanoma, cutaneous malignant, 2}, 155601;{Melanoma-pancreatic cancer syndrome}, 606719
CDKN2B	100%	100%	100%	98.6%	
CDKN2C	100%	100%	100%	98.6%	
CDON	100%	100%	100%	99.8%	Holoprosencephaly 11, 614226
CDSN	100%	100%	100%	99.8%	Hypotrichosis 2, 146520;Peeling skin syndrome 1, 270300

CDT1	100%	99.9%	100%	98.9%	Meier-Gorlin syndrome 4, 613804
CDY1	50%	50%	47.1%	29.3%	
CDY1B	50%	50%	47.1%	27.1%	
CDY2A	50%	50%	49.6%	27%	
CDY2B	50%	50%	49.1%	25.7%	
CEACAM16	100%	100%	100%	98.7%	Deafness, autosomal dominant 4B, 614614;Deafness, autosomal recessive 113, 618410
CEBPA	100%	100%	99.6%	80.9%	Leukemia, acute myeloid, somatic, 601626;?Leukemia, acute myeloid, 601626
CEBPE	100%	100%	100%	98%	?Immunodeficiency 108 with autoinflammation, 260570;Specific granule deficiency, 245480
CEL	100%	100%	99.5%	92.8%	Maturity-onset diabetes of the young, type VIII, 609812
CELA2A	100%	100%	100%	98.3%	Abdominal obesity-metabolic syndrome 4, 618620
CELF2	100%	100%	100%	99.5%	Developmental and epileptic encephalopathy 97, 619561
CELSR1	100%	99.9%	100%	97.8%	Lymphatic malformation 9, 619319
CELSR3	100%	100%	100%	98.9%	

CENATAC	100%	100%	100%	99.6%	?Mosaic variegated aneuploidy syndrome 4, 620153
CENPE	100%	100%	100%	99.4%	?Microcephaly 13, primary, autosomal recessive, 616051
CENPF	100%	100%	100%	99.7%	Stromme syndrome, 243605
CENPJ	100%	100%	100%	99.5%	
CENPT	100%	100%	100%	99.3%	?Short stature and microcephaly with genital anomalies, 618702
CEP104	100%	100%	100%	99.3%	Joubert syndrome 25, 616781;Intellectual developmental disorder, autosomal recessive 77, 619988
CEP112	100%	100%	100%	99.6%	Spermatogenic failure 44, 619044
CEP120	100%	100%	100%	99.6%	Short-rib thoracic dysplasia 13 with or without polydactyly, 616300;Joubert syndrome 31, 617761
CEP135	100%	100%	100%	99.7%	Microcephaly 8, primary, autosomal recessive, 614673
CEP152	100%	100%	100%	99.9%	Microcephaly 9, primary, autosomal recessive, 614852;Seckel syndrome 5, 613823
CEP162	100%	100%	100%	99.7%	

CEP164	100%	100%	100%	99.1%	Nephronophthisis 15, 614845
CEP19	100%	100%	100%	99.7%	Morbid obesity and spermatogenic failure, 615703
CEP250	100%	99.9%	100%	99%	Cone-rod dystrophy and hearing loss 2, 618358
CEP290	100%	100%	100%	99.6%	Leber congenital amaurosis 10, 611755;Joubert syndrome 5, 610188;Senior-Loken syndrome 6, 610189;?Bardet-Biedl syndrome 14, 615991;Meckel syndrome 4, 611134
CEP295	100%	100%	100%	99.7%	Seckel syndrome 11, 620767
CEP41	100%	100%	100%	99.5%	Joubert syndrome 15, 614464
CEP55	100%	100%	100%	99.8%	Multinucleated neurons, anhydramnios, renal dysplasia, cerebellar hypoplasia, and hydranencephaly, 236500
CEP57	100%	100%	100%	99.7%	Mosaic variegated aneuploidy syndrome 2, 614114
CEP63	92.8%	92.8%	100%	99.7%	?Seckel syndrome 6, 614728

CEP78	100%	100%	100%	99.8%	Cone-rod dystrophy and hearing loss, 617236
CEP83	100%	100%	100%	99.7%	Nephronophthisis 18, 615862
CEP85L	100%	100%	100%	99.8%	Lissencephaly 10, 618873
CEP89	100%	100%	100%	99.3%	
CERKL	100%	100%	100%	99.8%	Retinitis pigmentosa 26, 608380
CERS1	99.3%	97.1%	100%	97.9%	Epilepsy, progressive myoclonic, 8, 616230
CERS3	100%	100%	100%	99.7%	Ichthyosis, congenital, autosomal recessive 9, 615023
CERT1	100%	100%	100%	99.7%	Intellectual developmental disorder, autosomal dominant 34, 616351
CES1	99.9%	99.7%	99.7%	96.5%	Drug metabolism, altered, CES1-related, 618057
CETP	100%	100%	100%	99.5%	[High density lipoprotein cholesterol level QTL 10], 143470;Hyperalphalipoproteinemia, 143470
CFAP251	100%	100%	100%	99.3%	Spermatogenic failure 33, 618152
CFAP276	100%	100%	100%	99.1%	
CFAP298	100%	100%	100%	99.7%	Ciliary dyskinesia, primary, 26, 615500
CFAP300	100%	100%	100%	99.4%	Ciliary dyskinesia, primary, 38, 618063

CFAP410	100%	100%	100%	98.5%	Retinal dystrophy with macular staphyloma, 617547;Spondylometaphyseal dysplasia, axial, 602271
CFAP418	100%	100%	100%	99.7%	Retinitis pigmentosa 64, 614500;Cone-rod dystrophy 16, 614500;Bardet-Biedl syndrome 21, 617406
CFAP43	100%	100%	100%	99.8%	Hydrocephalus, normal pressure, 1, 236690;Spermatogenic failure 19, 617592
CFAP44	100%	100%	100%	99.6%	Spermatogenic failure 20, 617593
CFAP45	100%	100%	100%	99.5%	Heterotaxy, visceral, 11, autosomal, with male infertility, 619608
CFAP47	100%	99.8%	99.3%	75.1%	Spermatogenic failure, X-linked 3, 301059
CFAP52	100%	100%	100%	99.7%	Heterotaxy, visceral, 10, autosomal, with male infertility, 619607
CFAP53	100%	100%	100%	99.6%	Heterotaxy, visceral, 6, autosomal recessive, 614779
CFAP54	100%	100%	100%	99.7%	
CFAP57	100%	100%	100%	99.3%	Spermatogenic failure 95, 620917
CFAP58	100%	100%	100%	99.5%	Spermatogenic failure 49, 619144

CFAP61	100%	100%	100%	99.4%	Spermatogenic failure 84, 620409
CFAP65	100%	100%	100%	99.1%	Spermatogenic failure 40, 618664
CFAP69	100%	100%	100%	99.7%	Spermatogenic failure 24, 617959
CFAP70	100%	100%	100%	99.7%	?Spermatogenic failure 41, 618670
CFAP74	100%	100%	99.8%	97.5%	Ciliary dyskinesia, primary, 49, without situs inversus, 620197
CFAP91	100%	100%	100%	99.6%	Spermatogenic failure 51, 619177
CFB	100%	100%	100%	99%	?Complement factor B deficiency, 615561;{Hemolytic uremic syndrome, atypical, susceptibility to, 4}, 612924;{Macular degeneration, age-related, 14, reduced risk of}, 615489
CFC1	100%	100%	100%	99.8%	Heterotaxy, visceral, 2, autosomal, 605376
CFD	100%	100%	100%	95.8%	Complement factor D deficiency, 613912

CFH	97.5%	97.5%	100%	99.8%	{Macular degeneration, age-related, 4}, 610698;Basal laminar drusen, 126700;Complement factor H deficiency, 609814;{Hemolytic uremic syndrome, atypical, susceptibility to, 1}, 235400
CFHR1	94.2%	92.3%	93.4%	77.5%	{Macular degeneration, age-related, reduced risk of}, 603075;{Hemolytic uremic syndrome, atypical, susceptibility to}, 235400
CFHR2	76.4%	76.4%	100%	99.4%	
CFHR3	96%	95%	95.6%	83.4%	{Macular degeneration, age-related, reduced risk of}, 603075;{Hemolytic uremic syndrome, atypical, susceptibility to}, 235400
CFHR4	100%	100%	100%	98.8%	
CFHR5	100%	100%	100%	99.8%	Nephropathy due to CFHR5 deficiency, 614809
CFI	100%	100%	100%	99.9%	{Hemolytic uremic syndrome, atypical, susceptibility to, 3}, 612923;{Macular degeneration, age-related, 13, susceptibility to}, 615439;Complement factor I deficiency, 610984

CFL2	100%	100%	100%	99.9%	Nemaline myopathy 7, autosomal recessive, 610687
CFP	100%	98.8%	98.9%	67.2%	Properdin deficiency, X-linked, 312060
CFTR	100%	100%	100%	99.5%	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}
CGN	100%	100%	100%	98.8%	
CHAMP1	100%	100%	100%	99.2%	Neurodevelopmental disorder with hypotonia, impaired language, and dysmorphic features, 616579
CHASERR					Neurodevelopmental disorder with dysmorphic facies, absent speech and ambulation, and brain abnormalities, 621012
CHAT	100%	100%	100%	98.7%	Myasthenic syndrome, congenital, 6, presynaptic, 254210

CHCHD10	100%	100%	100%	99.5%	?Myopathy, isolated mitochondrial, autosomal dominant, 616209;Spinal muscular atrophy, Jokela type, 615048;Frontotemporal dementia and/or amyotrophic lateral sclerosis 2, 615911
CHCHD2	100%	100%	100%	99.9%	Parkinson disease 22, autosomal dominant, 616710
CHD1	100%	100%	100%	99.7%	Pilarowski-Bjornsson syndrome, 617682
CHD2	100%	100%	100%	99.5%	Developmental and epileptic encephalopathy 94, 615369
CHD3	100%	99.8%	100%	98%	Snijders Blok-Campeau syndrome, 618205
CHD4	100%	100%	100%	99.3%	Sifrim-Hitz-Weiss syndrome, 617159
CHD5	100%	100%	100%	98.5%	Parenti-Mignot neurodevelopmental syndrome, 619873
CHD7	100%	100%	100%	99.4%	Hypogonadotropic hypogonadism 5 with or without anosmia, 612370;CHARGE syndrome, 214800
CHD8	100%	100%	100%	99.4%	Intellectual developmental disorder with autism and macrocephaly, 615032

CHEK1	100%	100%	100%	99.6%	Oocyte/zygote/embryo maturation arrest 21, 620610
CHEK2	100%	99.9%	100%	99.2%	Prostate cancer, somatic, 176807;Osteosarcoma, somatic, 259500;Tumor predisposition syndrome 4, breast/prostate/colorectal, 609265
CHIT1	100%	100%	100%	99.7%	[Chitotriosidase deficiency], 614122
CHKA	100%	99.5%	100%	97.3%	Neurodevelopmental disorder with microcephaly, movement abnormalities, and seizures, 620023
CHKB	100%	100%	100%	98.9%	Muscular dystrophy, congenital, megaconial type, 602541
CHM	98.8%	97.1%	98.6%	75.3%	Choroideremia, 303100
CHMP1A	100%	100%	100%	98.8%	Pontocerebellar hypoplasia, type 8, 614961
CHMP2B	91.5%	88.2%	100%	99.5%	Frontotemporal dementia and/or amyotrophic lateral sclerosis 7, 600795
CHMP4B	100%	100%	100%	98.8%	Cataract 31, multiple types, 605387
CHN1	96.5%	96.5%	100%	99.3%	Duane retraction syndrome 2, 604356
CHP1	100%	100%	100%	99.4%	?Spastic ataxia 9, autosomal recessive, 618438

CHRDL1	100%	99.9%	99.5%	74.1%	Megalocornea 1, X-linked, 309300
CHRM1	100%	100%	100%	99.6%	
CHRM2	100%	100%	100%	99.6%	
CHRM3	100%	100%	100%	98.4%	Prune belly syndrome, 100100
CHRNA1	100%	100%	100%	99.3%	Myasthenic syndrome, congenital, 1B, fast-channel, 608930;Myasthenic syndrome, congenital, 1A, slow-channel, 601462;Multiple pterygium syndrome, lethal type, 253290
CHRNA2	100%	100%	100%	98.7%	Epilepsy, nocturnal frontal lobe, type 4, 610353
CHRNA3	100%	100%	100%	99%	{Lung cancer susceptibility 2}, 612052;Bladder dysfunction, autonomic, with impaired pupillary reflex and secondary CAKUT, 191800
CHRNA4	100%	100%	100%	95.4%	{Nicotine addiction, susceptibility to}, 188890;Epilepsy, nocturnal frontal lobe, 1, 600513

CHRNB1	100%	100%	100%	98.5%	?Myasthenic syndrome, congenital, 2C, associated with acetylcholine receptor deficiency, 616314;Myasthenic syndrome, congenital, 2A, slow-channel, 616313
CHRNB2	100%	100%	100%	98.8%	Epilepsy, nocturnal frontal lobe, 3, 605375
CHRND	100%	100%	100%	99%	?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
CHRNE	100%	100%	100%	98.7%	Myasthenic syndrome, congenital, 4A, slow-channel, 605809;Myasthenic syndrome, congenital, 4C, associated with acetylcholine receptor deficiency, 608931;Myasthenic syndrome, congenital, 4B, fast-channel, 616324

CHRNG	100%	100%	100%	98.7%	Multiple pterygium syndrome, lethal type, 253290;Escobar syndrome, 265000
CHST11	100%	100%	100%	99.2%	?Osteochondrodysplasia, brachydactyly, and overlapping malformed digits, 618167
CHST14	100%	100%	100%	97.1%	Ehlers-Danlos syndrome, musculocontractural type 1, 601776
CHST3	100%	100%	99.9%	97.8%	Spondyloepiphyseal dysplasia with congenital joint dislocations, 143095
CHST6	100%	100%	100%	99.2%	Macular corneal dystrophy, 217800
CHST8	100%	100%	100%	98.6%	
CHSY1	99.8%	99%	100%	97.9%	Temtamy preaxial brachydactyly syndrome, 605282
CHUK	100%	100%	100%	99.7%	?Popliteal pterygium syndrome, Bartsocas-Papas type 2, 619339;?Cocoon syndrome, 613630
CIAO1	100%	100%	100%	99.2%	Multiple mitochondrial dysfunctions syndrome 10, 620960
CIB1	100%	100%	100%	99.4%	{Epidermodysplasia verruciformis, susceptibility to, 3}, 618267

CIB2	100%	99.9%	100%	98.8%	Deafness, autosomal recessive 48, 609439;Usher syndrome, type IJ, 614869
CIBAR1	100%	100%	100%	99.5%	?Polydactyly, postaxial, type A9, 618219
CIC	100%	100%	100%	97.9%	Intellectual developmental disorder, autosomal dominant 45, 617600
CIDEC	100%	100%	100%	99.6%	?Lipodystrophy, familial partial, type 5, 615238
CIITA	100%	100%	100%	98.5%	{Rheumatoid arthritis, susceptibility to}, 180300;MHC class II deficiency 1, 209920
CILK1	100%	100%	100%	99.7%	{Epilepsy, juvenile myoclonic, susceptibility to, 10}, 617924;Endocrine-cerebroosteodysplasia, 612651
CIROP	98.4%	93.9%	100%	99.5%	Heterotaxy, visceral, 12, autosomal, 619702
CISD2	100%	100%	100%	99.5%	Wolfram syndrome 2, 604928
CIT	95.8%	95.8%	100%	99.1%	Microcephaly 17, primary, autosomal recessive, 617090
CITED2	100%	100%	100%	98.8%	Atrial septal defect 8, 614433;Ventricular septal defect 2, 614431
CKAP2L	100%	100%	100%	99.7%	Filippi syndrome, 272440

CLCC1	98.6%	98.6%	100%	99.7%	Retinitis pigmentosa 32, 609913
CLCF1	100%	100%	100%	99.4%	Cold-induced sweating syndrome 2, 610313
CLCN1	100%	100%	100%	99.3%	Myotonia congenita, recessive, 255700;Myotonia congenita, dominant, 160800;Myotonia levior, 160800
CLCN2	100%	100%	100%	99.4%	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
CLCN3	100%	100%	100%	99.7%	Neurodevelopmental disorder with seizures and brain abnormalities, 619517;Neurodevelopmental disorder with hypotonia and brain abnormalities, 619512
CLCN4	100%	99.6%	98.8%	69%	Raynaud-Claes syndrome, 300114

CLCN5	100%	99.7%	99.1%	73.4%	Proteinuria, low molecular weight, with hypercalciuric nephrocalcinosis, 308990;Hypophosphatemic rickets, 300554;Dent disease 1, 300009;Nephrolithiasis, type I, 310468
CLCN6	100%	100%	100%	99.1%	Ceroid lipofuscinosis, neuronal, 15, 619173
CLCN7	100%	100%	100%	99%	Hypopigmentation, organomegaly, and delayed myelination and development, 618541;Osteopetrosis, autosomal recessive 4, 611490;Osteopetrosis, autosomal dominant 2, 166600
CLCNKA	100%	100%	100%	99.3%	Bartter syndrome, type 4b, digenic, 613090
CLCNKB	100%	100%	100%	99%	Bartter syndrome, type 3, 607364;Bartter syndrome, type 4b, digenic, 613090
CLDN1	100%	100%	100%	99.5%	Ichthyosis, leukocyte vacuoles, alopecia, and sclerosing cholangitis, 607626
CLDN10	100%	100%	100%	99.6%	HELIX syndrome, 617671
CLDN11	100%	100%	100%	98.8%	Leukodystrophy, hypomyelinating, 22, 619328

CLDN14	100%	100%	100%	98.9%	Deafness, autosomal recessive 29, 614035
CLDN16	100%	100%	100%	99.6%	Hypomagnesemia 3, renal, 248250
CLDN19	100%	99.9%	100%	99.5%	Hypomagnesemia 5, renal, with ocular involvement, 248190
CLDN2	100%	99.9%	99.7%	77.2%	?Azoospermia, obstructive, with nephrolithiasis, 301060
CLDN5	100%	100%	100%	98.7%	
CLDN9	100%	100%	100%	99.2%	Deafness, autosomal recessive 116, 619093
CLEC3B	100%	100%	100%	99.4%	Macular dystrophy, retinal, 4, 619977
CLEC4D	100%	100%	100%	99.8%	
CLEC7A	100%	100%	100%	99.8%	Candidiasis, familial, 4, autosomal recessive, 613108;{Aspergillosis, susceptibility to}, 614079
CLIC2	100%	99.9%	99%	75.4%	
CLIC5	100%	100%	100%	99.5%	?Deafness, autosomal recessive 103, 616042
CLIP1	100%	100%	100%	99.5%	
CLMP	100%	100%	100%	99.4%	Congenital short bowel syndrome, 615237
CLN3	93.1%	93.1%	100%	99%	Ceroid lipofuscinosis, neuronal, 3, 204200
CLN5	83%	83%	100%	99.7%	Ceroid lipofuscinosis, neuronal, 5, 256731

CLN6	100%	100%	100%	98.8%	Ceroid lipofuscinosis, neuronal, 6B (Kufs type), 204300;Ceroid lipofuscinosis, neuronal, 6A, 601780
CLN8	100%	100%	100%	99.7%	Ceroid lipofuscinosis, neuronal, 8, Northern epilepsy variant, 610003;Ceroid lipofuscinosis, neuronal, 8, 600143
CLP1	100%	100%	100%	98.8%	Pontocerebellar hypoplasia, type 10, 615803
CLPB	100%	100%	100%	98.9%	Neutropenia, severe congenital, 9, autosomal dominant, 619813;3-methylglutaconic aciduria, type VIIB, autosomal recessive, 616271;3-methylglutaconic aciduria, type VIIA, autosomal dominant, 619835
CLPP	100%	100%	100%	96%	Perrault syndrome 3, 614129
CLPX	100%	100%	100%	99.8%	?Protoporphyria, erythropoietic, 2, 618015
CLRN1	100%	100%	100%	99.7%	Usher syndrome, type 3A, 276902;Retinitis pigmentosa 61, 614180
CLRN2	100%	100%	100%	99.8%	Deafness, autosomal recessive 117, 619174

CLTC	99.2%	99.2%	100%	99.7%	Intellectual developmental disorder, autosomal dominant 56, 617854
CLTCL1	100%	100%	100%	99.4%	
CLUAP1	100%	100%	100%	99.7%	
CLXN	100%	100%	100%	99.4%	Ciliary dyskinesia, primary, 53, 620642
CMAS	100%	100%	100%	99.5%	
CMPK2	100%	100%	100%	99.1%	Basal ganglia calcification, idiopathic, 10, autosomal recessive, 621018
CNBP	100%	100%	100%	99.8%	Myotonic dystrophy 2, 602668
CNGA1	100%	100%	100%	99.8%	Retinitis pigmentosa 49, 613756
CNGA2	99.9%	98.7%	97.5%	62.9%	
CNGA3	100%	100%	100%	99.1%	Achromatopsia 2, 216900
CNGB1	100%	100%	100%	98.3%	Retinitis pigmentosa 45, 613767
CNGB3	100%	100%	100%	99.7%	Achromatopsia 3, 262300
CNKSR2	100%	99.8%	99.3%	74.9%	Intellectual developmental disorder, X-linked syndromic, Houge type, 301008
CNNM2	100%	100%	100%	98.6%	Hypomagnesemia 6, renal, 613882;Hypomagnesemia, seizures, and impaired intellectual development 1, 616418

CNNM4	100%	100%	100%	98.9%	Jalili syndrome, 217080
CNOT1	100%	100%	100%	99.5%	Vissers-Bodmer syndrome, 619033;Holoprosencephaly 12, with or without pancreatic agenesis, 618500
CNOT2	100%	100%	100%	99.3%	Intellectual developmental disorder with nasal speech, dysmorphic facies, and variable skeletal anomalies, 618608
CNOT3	100%	100%	100%	98.3%	Intellectual developmental disorder with speech delay, autism, and dysmorphic facies, 618672
CNOT9	96.8%	92.4%	100%	99.9%	
CNP	100%	100%	100%	99.6%	?Leukodystrophy, hypomyelinating, 20, 619071
CNPY3	100%	100%	100%	98.5%	Developmental and epileptic encephalopathy 60, 617929
CNTN1	100%	100%	100%	99.5%	Congenital myopathy 12, 612540
CNTN2	100%	100%	100%	99.2%	Epilepsy, early-onset, 5, with or without developmental delay, 615400

CNTNAP1	100%	100%	100%	98.8%	Lethal congenital contracture syndrome 7, 616286;Hypomyelinating neuropathy, congenital, 3, 618186
CNTNAP2	100%	100%	100%	99%	Pitt-Hopkins like syndrome 1, 610042;{Autism susceptibility 15}, 612100
COA1	100%	100%	100%	99.6%	
COA3	100%	100%	100%	98.6%	?Mitochondrial complex IV deficiency, nuclear type 14, 619058
COA5	82.4%	82.4%	100%	97.4%	?Mitochondrial complex IV, deficiency, nuclear type 9, 616500
COA6	100%	100%	100%	99.8%	Mitochondrial complex IV deficiency, nuclear type 13, 616501
COA7	100%	100%	100%	99.2%	Spinocerebellar ataxia, autosomal recessive, with axonal neuropathy 3, 618387
COA8	100%	99.9%	100%	98.2%	Mitochondrial complex IV deficiency, nuclear type 17, 619061
COASY	100%	100%	100%	98.9%	Pontocerebellar hypoplasia, type 12, 618266;Neurodegeneration with brain iron accumulation 6, 615643

COCH	100%	100%	100%	99.9%	Deafness, autosomal dominant 9, 601369;?Deafness, autosomal recessive 110, 618094
COG1	100%	100%	100%	99%	Congenital disorder of glycosylation, type IIg, 611209
COG2	100%	100%	100%	99.5%	?Congenital disorder of glycosylation, type IIq, 617395
COG3	100%	100%	100%	99.7%	Congenital disorder of glycosylation, type IIbb, 620546
COG4	100%	100%	100%	99.1%	Congenital disorder of glycosylation, type IIj, 613489;Saul-Wilson syndrome, 618150
COG5	100%	100%	100%	99.8%	Congenital disorder of glycosylation, type IIi, 613612
COG6	100%	100%	100%	99.7%	Shaheen syndrome, 615328;Congenital disorder of glycosylation, type III, 614576
COG7	100%	100%	100%	99.2%	Congenital disorder of glycosylation, type IIe, 608779
COG8	100%	100%	100%	98.4%	Congenital disorder of glycosylation, type IIh, 611182

COL10A1	100%	100%	100%	99.3%	Metaphyseal chondrodysplasia, Schmid type, 156500
COL11A1	100%	100%	100%	99.6%	Fibrochondrogenesis 1, 228520;Stickler syndrome, type II, 604841;Marshall syndrome, 154780;Deafness, autosomal dominant 37, 618533;{Lumbar disc herniation, susceptibility to}, 603932
COL11A2	100%	100%	100%	98.2%	Deafness, autosomal dominant 13, 601868;Otospondylomegaepiphyseal dysplasia, autosomal recessive, 215150;Fibrochondrogenesis 2, 614524;Deafness, autosomal recessive 53, 609706;Otospondylomegaepiphyseal dysplasia, autosomal dominant, 184840
COL12A1	100%	100%	100%	99.7%	Bethlem myopathy 2, 616471;?Ullrich congenital muscular dystrophy 2, 616470
COL13A1	100%	100%	100%	98.9%	Myasthenic syndrome, congenital, 19, 616720
COL14A1	100%	100%	100%	99.6%	

COL17A1	100%	100%	100%	99.2%	Epithelial recurrent erosion dystrophy, 122400;Epidermolysis bullosa, junctional 4, intermediate, 619787
COL18A1	100%	100%	100%	98.9%	Knobloch syndrome, type 1, 267750;Glaucoma, primary closed-angle, 618880
COL1A1	100%	100%	100%	98.8%	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

COL1A2	100%	100%	100%	99.8%	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
COL25A1	100%	100%	100%	99.5%	Fibrosis of extraocular muscles, congenital, 5, 616219
COL27A1	100%	100%	100%	98.9%	Steel syndrome, 615155

COL2A1	100%	100%	100%	99.5%	?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
COL3A1	100%	100%	100%	99.2%	Ehlers-Danlos syndrome, vascular type, 130050;Polymicrogyria with or without vascular-type EDS, 618343

COL4A1	100%	100%	100%	99.3%	?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
COL4A2	100%	100%	100%	98.7%	Brain small vessel disease 2, 614483;{Hemorrhage, intracerebral, susceptibility to}, 614519
COL4A3	100%	100%	100%	99.5%	Alport syndrome 3A, autosomal dominant, 104200;Hematuria, benign familial, 2, 620320;Alport syndrome 3B, autosomal recessive, 620536
COL4A4	99.7%	99%	100%	99.4%	Hematuria, familial benign, 1, 141200;Alport syndrome 2, autosomal recessive, 203780
COL4A5	100%	99.4%	99.1%	71.2%	Alport syndrome 1, X-linked, 301050
COL4A6	100%	99%	99%	71.9%	?Deafness, X-linked 6, 300914

COL5A1	100%	100%	100%	98.4%	Ehlers-Danlos syndrome, classic type, 1, 130000;Fibromuscular dysplasia, multifocal, 619329
COL5A2	100%	100%	100%	99.6%	Ehlers-Danlos syndrome, classic type, 2, 130010
COL6A1	100%	100%	100%	98.7%	Ullrich congenital muscular dystrophy 1A, 254090;Bethlem myopathy 1A, 158810
COL6A2	100%	100%	100%	99%	?Myosclerosis, congenital, 255600;Ullrich congenital muscular dystrophy 1B, 620727;Bethlem myopathy 1B, 620725
COL6A3	100%	100%	100%	99.6%	Bethlem myopathy 1C, 620726;Ullrich congenital muscular dystrophy 1C, 620728;Dystonia 27, 616411
COL6A5	100%	100%	100%	99.6%	

COL7A1	100%	100%	100%	98.9%	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
COL8A2	100%	100%	100%	96.2%	Corneal dystrophy, posterior polymorphous 2, 609140;Corneal dystrophy, Fuchs endothelial, 1, 136800
COL9A1	100%	100%	100%	99.4%	Stickler syndrome, type IV, 614134;?Epiphyseal dysplasia, multiple, 6, 614135
COL9A2	100%	100%	100%	98.4%	Epiphyseal dysplasia, multiple, 2, 600204;?Stickler syndrome, type V, 614284

COL9A3	100%	100%	100%	98.4%	{Intervertebral disc disease, susceptibility to}, 603932;Epiphyseal dysplasia, multiple, 3, with or without myopathy, 600969;Stickler syndrome, type VI, 620022
COLEC10	100%	100%	100%	99.7%	3MC syndrome 3, 248340
COLEC11	100%	100%	100%	99%	3MC syndrome 2, 265050
COLGALT1	100%	100%	100%	98.1%	Brain small vessel disease 3, 618360
COLQ	100%	100%	100%	99.7%	Myasthenic syndrome, congenital, 5, 603034
COMP	100%	100%	100%	98.1%	Pseudoachondroplasia, 177170;Carpal tunnel syndrome 2, 619161;Epiphyseal dysplasia, multiple, 1, 132400
COMT	93.3%	91.9%	100%	99%	{Schizophrenia, susceptibility to}, 181500;{Panic disorder, susceptibility to}, 167870
COPA	100%	100%	100%	99.4%	{Autoinflammation and autoimmunity, systemic, with immune dysregulation}, 616414
COPB1	100%	100%	100%	99.6%	Baralle-Macken syndrome, 619255

COPB2	100%	100%	100%	99.6%	Osteoporosis, childhood- or juvenile-onset, with developmental delay, 619884;?Microcephaly 19, primary, autosomal recessive, 617800
COPG1	100%	100%	100%	99.3%	?Immunodeficiency 128, 620983
COQ2	96.3%	96.3%	100%	98.7%	{Multiple system atrophy, susceptibility to}, 146500;Coenzyme Q10 deficiency, primary, 1, 607426
COQ4	100%	100%	100%	98.7%	Coenzyme Q10 deficiency, primary, 7, 616276;Spastic ataxia 10, autosomal recessive, 620666
COQ5	100%	100%	100%	98.9%	?Coenzyme Q10 deficiency, primary, 9, 619028
COQ6	100%	100%	100%	99.6%	Coenzyme Q10 deficiency, primary, 6, 614650
COQ7	100%	100%	100%	98.5%	Coenzyme Q10 deficiency, primary, 8, 616733;Neuronopathy, distal hereditary motor, autosomal recessive 9, 620402
COQ8A	100%	100%	100%	98.7%	Coenzyme Q10 deficiency, primary, 4, 612016
COQ8B	100%	100%	100%	99.3%	Nephrotic syndrome, type 9, 615573

COQ9	100%	100%	100%	98.6%	Coenzyme Q10 deficiency, primary, 5, 614654
CORIN	100%	100%	100%	99.6%	?Cardiomyopathy, familial hypertrophic, 30, atrial, 620734;Preeclampsia/eclampsia 5, 614595
CORO1A	100%	100%	100%	99.2%	Immunodeficiency 8, 615401
COX10	100%	100%	100%	99.5%	Mitochondrial complex IV deficiency, nuclear type 3, 619046
COX11	100%	100%	100%	97.1%	Mitochondrial complex IV deficiency, nuclear type 23, 620275
COX14	100%	100%	100%	98.3%	?Mitochondrial complex IV deficiency, nuclear type 10, 619053
COX15	100%	100%	100%	99.7%	Mitochondrial complex IV deficiency, nuclear type 6, 615119
COX16	100%	100%	100%	99.8%	Mitochondrial complex IV deficiency, nuclear type 22, 619355
COX18	100%	100%	100%	98%	
COX20	100%	100%	100%	99.6%	Mitochondrial complex IV deficiency, nuclear type 11, 619054
COX4I1	100%	100%	100%	99.9%	Mitochondrial complex IV deficiency, nuclear type 16, 619060

COX4I2	100%	100%	100%	98.7%	Exocrine pancreatic insufficiency, dyserythropoietic anemia, and calvarial hyperostosis, 612714
COX5A	100%	100%	100%	99.9%	Mitochondrial complex IV deficiency, nuclear type 20, 619064
COX5B	100%	100%	100%	100%	
COX6A1	100%	100%	100%	100%	Charcot-Marie-Tooth disease, recessive intermediate D, 616039
COX6A2	100%	99.9%	100%	93.9%	Mitochondrial complex IV deficiency, nuclear type 18, 619062
COX6B1	100%	100%	100%	98.7%	Mitochondrial complex IV deficiency, nuclear type 7, 619051
COX6B2	100%	100%	100%	98.7%	
COX6C	100%	100%	100%	99.7%	
COX7A1	100%	100%	100%	98.4%	
COX7A2	100%	100%	100%	99.7%	
COX7B	100%	99.8%	99.4%	70.3%	Linear skin defects with multiple congenital anomalies 2, 300887
COX7B2	100%	100%	100%	100%	
COX7C	100%	100%	100%	99.6%	

COX8A	100%	100%	100%	98.8%	?Mitochondrial complex IV deficiency, nuclear type 15, 619059
COX8C	100%	100%	100%	97.9%	
CP	100%	100%	100%	99.2%	Aceruloplasminemia, 604290
CPA6	100%	100%	100%	99.8%	Febrile seizures, familial, 11, 614418;Epilepsy, familial temporal lobe, 5, 614417
CPAMD8	100%	100%	100%	98.9%	Anterior segment dysgenesis 8, 617319
CPE	100%	100%	100%	99.1%	BDV syndrome, 619326
CPLANE1	100%	100%	100%	99.5%	Orofaciodigital syndrome VI, 277170;Joubert syndrome 17, 614615
CPLX1	100%	100%	100%	98.5%	Developmental and epileptic encephalopathy 63, 617976
CPN1	100%	100%	100%	99.1%	Carboxypeptidase N deficiency, 212070
CPOX	100%	100%	100%	98.3%	Coproporphyria, 121300;Harderoporphyria, 618892
CPS1	100%	100%	100%	99.6%	Carbamoylphosphate synthetase I deficiency, 237300;{Pulmonary hypertension, neonatal, susceptibility to}, 615371
CPSF1	100%	100%	100%	98.5%	Myopia 27, 618827

CPSF3	100%	100%	100%	99.6%	Neurodevelopmental disorder with microcephaly, hypotonia, nystagmus, and seizures, 619876
CPT1A	100%	100%	100%	99.3%	CPT deficiency, hepatic, type IA, 255120
CPT1C	100%	100%	100%	98.5%	?Spastic paraplegia 73, autosomal dominant, 616282
CPT2	100%	100%	100%	99.6%	{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
CR2	100%	100%	100%	99.7%	{Systemic lupus erythematosus, susceptibility to, 9}, 610927;?Immunodeficiency, common variable, 7, 614699
CRACR2A	100%	100%	100%	99.6%	
CRADD	100%	100%	100%	99.5%	Intellectual developmental disorder, autosomal recessive 34, with variant lissencephaly, 614499
CRAT	100%	100%	100%	98.9%	?Neurodegeneration with brain iron accumulation 8, 617917

CRB1	98.6%	98.6%	100%	99.8%	Leber congenital amaurosis 8, 613835;Retinitis pigmentosa-12, 600105;Pigmented paravenous chorioretinal atrophy, 172870
CRB2	100%	100%	100%	99.2%	Focal segmental glomerulosclerosis 9, 616220;Ventriculomegaly with cystic kidney disease, 219730
CRBN	99.8%	98.5%	100%	99.4%	Intellectual developmental disorder, autosomal recessive 2, 607417
CREB1	100%	100%	100%	99.9%	Histiocytoma, angiomatoid fibrous, somatic, 612160
CREB3L1	100%	100%	100%	97.1%	Osteogenesis imperfecta, type XVI, 616229
CREB3L3	100%	100%	100%	98.7%	Hypertriglyceridemia 2, 619324
CREBBP	100%	100%	100%	99%	Menke-Hennekam syndrome 1, 618332;Rubinstein-Taybi syndrome 1, 180849

CRELD1	100%	100%	100%	99%	Atrioventricular septal defect, partial, with heterotaxy syndrome, 606217;Jeffries-Lakhani neurodevelopmental syndrome, 620771;{Atrioventricular septal defect, susceptibility to, 2}, 606217
CRIPT	100%	100%	100%	99.4%	Rothmund-Thomson syndrome, type 3, 615789
CRLF1	99.4%	97.5%	99.2%	88.9%	Cold-induced sweating syndrome 1, 272430
CRLS1	100%	100%	100%	99.2%	Combined oxidative phosphorylation deficiency 57, 620167
CRPPA	100%	100%	100%	99.4%	Muscular dystrophy-dystroglycanopathy (limb-girdle), type C, 7, 616052;Muscular dystrophy-dystroglycanopathy (congenital with brain and eye anomalies), type A, 7, 614643
CRTAP	100%	100%	100%	99%	Osteogenesis imperfecta, type VII, 610682
CRTC1	100%	100%	100%	97.9%	Mucoepidermoid salivary gland carcinoma
CRX	100%	100%	100%	98.2%	Leber congenital amaurosis 7, 613829;Cone-rod retinal dystrophy-2, 120970

CRYAA	100%	100%	100%	97.9%	Cataract 9, multiple types, 604219
CRYAB	100%	100%	100%	99.7%	Myopathy, myofibrillar, fatal infantile hypertonic, alpha-B crystallin-related, 613869;Myopathy, myofibrillar, 2, 608810;Cataract 16, multiple types, 613763;Cardiomyopathy, dilated, 1II, 615184
CRYBA1	100%	100%	100%	99.5%	Cataract 10, multiple types, 600881
CRYBA2	100%	100%	98.2%	90.5%	?Cataract 42, 115900
CRYBA4	100%	100%	100%	99.5%	Cataract 23, 610425
CRYBB1	100%	100%	100%	98.6%	Cataract 17, multiple types, 611544
CRYBB2	100%	100%	100%	99.4%	Cataract 3, multiple types, 601547
CRYBB3	100%	100%	100%	99.2%	Cataract 22, 609741
CRYGB	100%	100%	100%	98.4%	Cataract 39, multiple types, autosomal dominant, 615188
CRYGC	100%	100%	100%	99.4%	Cataract 2, multiple types, 604307
CRYGD	100%	100%	100%	99.6%	Cataract 4, multiple types, 115700
CRYGS	100%	100%	100%	99.4%	Cataract 20, multiple types, 116100
CRYL1	100%	100%	100%	98.9%	

CRYM	100%	100%	100%	99.3%	Deafness, autosomal dominant 40, 616357
CSDE1	98.2%	97.8%	100%	99.7%	
CSF1R	100%	100%	100%	99.2%	Brain abnormalities, neurodegeneration, and dysosteosclerosis, 618476;Leukoencephalopathy, diffuse hereditary, with spheroids 1, 221820
CSF2RA	48.8%	48.2%	50%	49.5%	Surfactant metabolism dysfunction, pulmonary, 4, 300770
CSF2RB	100%	100%	100%	98.7%	Surfactant metabolism dysfunction, pulmonary, 5, 614370
CSF3R	100%	100%	100%	99.3%	Neutropenia, severe congenital, 7, autosomal recessive, 617014;?Neutrophilia, hereditary, 162830
CSGALNACT1	100%	100%	100%	99.2%	Skeletal dysplasia, mild, with joint laxity and advanced bone age, 618870
CSMD1	100%	100%	100%	99.5%	
CSNK1D	100%	100%	100%	98.6%	Advanced sleep-phase syndrome, familial, 2, 615224
CSNK1G1	100%	100%	100%	99.8%	

CSNK2A1	94.2%	94.2%	100%	99.4%	Okur-Chung neurodevelopmental syndrome, 617062
CSNK2B	100%	100%	100%	99.1%	Poirier-Bienvenu neurodevelopmental syndrome, 618732
CSPP1	96.9%	96.9%	100%	99.4%	Joubert syndrome 21, 615636
CSRP3	100%	100%	100%	99.7%	?Cardiomyopathy, dilated, 1M, 607482;Cardiomyopathy, hypertrophic, 12, 612124
CST3	100%	100%	99.8%	93.3%	{Macular degeneration, age-related, 11}, 611953;Cerebral amyloid angiopathy, 105150
CST6	100%	100%	100%	99.4%	?Ectodermal dysplasia 15, hypohidrotic/hair type, 618535
CSTA	100%	100%	100%	100%	Peeling skin syndrome 4, 607936
CSTB	100%	100%	100%	99.5%	Epilepsy, progressive myoclonic 1A (Unverricht and Lundborg), 254800
CSTF2	100%	99.8%	99.1%	71.1%	?Intellectual developmental disorder, X-linked 113, 301116
CT55	100%	98.8%	98.8%	75.7%	?Spermatogenic failure, X-linked, 7, 301106

CTBP1	99.3%	97.9%	99.9%	97.1%	Hypotonia, ataxia, developmental delay, and tooth enamel defect syndrome, 617915
CTC1	100%	100%	100%	99.3%	Cerebroretinal microangiopathy with calcifications and cysts, 612199
CTCF	100%	100%	100%	99.2%	Intellectual developmental disorder, autosomal dominant 21, 615502
CTDP1	100%	100%	100%	99%	Congenital cataracts, facial dysmorphism, and neuropathy, 604168
CTH	100%	100%	100%	99.8%	Cystathioninuria, 219500
CTHRC1	100%	100%	100%	99.5%	Barrett esophagus/esophageal adenocarcinoma, 614266
CTLA4	93.2%	93.2%	100%	99.4%	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

CTNNA1	100%	100%	100%	99.5%	Macular dystrophy, patterned, 2, 608970
CTNNA2	100%	100%	100%	99.1%	Cortical dysplasia, complex, with other brain malformations 9, 618174
CTNNA3	100%	100%	100%	99.8%	Arrhythmogenic right ventricular dysplasia 13, 615616
CTNNB1	100%	100%	100%	99.8%	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
CTNNBL1	100%	100%	100%	99.7%	?Immunodeficiency 99 with hypogammaglobulinemia and autoimmune cytopenias, 619846
CTNND1	100%	100%	100%	99.6%	Blepharocheilodontic syndrome 2, 617681
CTNND2	100%	99.5%	99.9%	97%	

CTNS	100%	100%	100%	98.1%	Cystinosis, nephropathic, 219800;Cystinosis, ocular nonnephropathic, 219750;Cystinosis, late-onset juvenile or adolescent nephropathic, 219900;Cystinosis, atypical nephropathic, 219800
CTPS1	100%	100%	100%	99.8%	Immunodeficiency 24, 615897
CTR9	100%	100%	100%	99.8%	
CTSA	100%	99.9%	100%	99.2%	Galactosialidosis, 256540
CTSB	87.8%	84.4%	100%	99.2%	Keratolytic winter erythema, 148370
CTSC	94.8%	94.3%	100%	99.5%	Periodontitis 1, juvenile, 170650;Haim-Munk syndrome, 245010;Papillon-Lefevre syndrome, 245000
CTSD	100%	100%	100%	99.4%	Ceroid lipofuscinosis, neuronal, 10, 610127
CTSF	100%	100%	100%	99.1%	Ceroid lipofuscinosis, neuronal, 13 (Kufs type), 615362
CTSH	97.2%	94.7%	100%	99.3%	
CTSK	100%	100%	100%	99.2%	Pycnodysostosis, 265800
CTSZ	81.6%	72.7%	100%	99.3%	
CTTNBP2	100%	100%	100%	99.2%	

CTU2	100%	100%	100%	98.9%	Microcephaly, facial dysmorphism, renal agenesis, and ambiguous genitalia syndrome, 618142
CUBN	100%	100%	100%	99.6%	[Proteinuria, chronic benign], 618884;Imerslund-Grasbeck syndrome 1, 261100
CUL3	100%	100%	100%	99.5%	Neurodevelopmental disorder with or without autism or seizures, 619239;Pseudohypoaldosteronism, type IIE, 614496
CUL4B	96.7%	96.5%	99.2%	72.6%	Intellectual developmental disorder, X-linked syndromic, Cabezas type, 300354
CUL7	100%	100%	100%	98.6%	3-M syndrome 1, 273750
CUX1	100%	99.9%	100%	97.4%	Global developmental delay with or without impaired intellectual development, 618330
CUX2	100%	100%	100%	98.9%	Developmental and epileptic encephalopathy 67, 618141
CWC27	82.6%	82.6%	100%	99.6%	Retinitis pigmentosa with or without skeletal anomalies, 250410
CWF19L1	100%	100%	100%	99.7%	Spinocerebellar ataxia, autosomal recessive 17, 616127

CXCR2	100%	100%	100%	99.6%	?WHIM syndrome 2, 619407
CXCR4	99%	99%	100%	99.2%	WHIM syndrome 1, 193670;Myelokathexis, isolated, 193670
CYB561	100%	100%	100%	97.9%	Orthostatic hypotension 2, 618182
CYB5A	100%	100%	100%	99.3%	Methemoglobinemia and ambiguous genitalia, 250790
CYB5R3	95.5%	93.5%	100%	98.4%	Methemoglobinemia, type I, 250800;Methemoglobinemia, type II, 250800
CYBA	71.5%	69.6%	100%	99.3%	Chronic granulomatous disease 4, autosomal recessive, 233690
CYBB	100%	99.9%	98.9%	74.5%	Immunodeficiency 34, mycobacteriosis, X-linked, 300645;Chronic granulomatous disease, X-linked, 306400
CYBC1	100%	100%	100%	99.1%	Chronic granulomatous disease 5, autosomal recessive, 618935
CYBRD1	100%	100%	100%	99.3%	
CYC1	100%	100%	100%	96.8%	Mitochondrial complex III deficiency, nuclear type 6, 615453
CYCS	100%	100%	100%	99.9%	Thrombocytopenia 4, 612004

CYFIP2	98.1%	98.1%	100%	99.4%	Developmental and epileptic encephalopathy 65, 618008
CYLC1	100%	99.8%	99.4%	73%	{Spermatogenic failure, X-linked, 8, susceptibility to}, 301119
CYLD	100%	100%	100%	99.6%	Brooke-Spiegler syndrome, 605041;Cylindromatosis, familial, 132700;Trichoepithelioma, multiple familial, 1, 601606;?Frontotemporal dementia and/or amyotrophic lateral sclerosis 8, 619132
CYP11A1	100%	100%	100%	99.3%	Adrenal insufficiency, congenital, with 46XY sex reversal, partial or complete, 613743
CYP11B1	100%	100%	100%	99.6%	Aldosteronism, glucocorticoid-remediable, 103900;Adrenal hyperplasia, congenital, due to 11-beta-hydroxylase deficiency, 202010
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

CYP17A1	100%	100%	100%	98.7%	17,20-lyase deficiency, isolated, 202110;17-alpha-hydroxylase/17,20-lyase deficiency, 202110
CYP19A1	100%	100%	100%	99.8%	Aromatase deficiency, 613546;Aromatase excess syndrome, 139300
CYP1B1	100%	100%	100%	99.2%	Glaucoma 3A, primary open angle, congenital, juvenile, or adult onset, 231300;Anterior segment dysgenesis 6, multiple subtypes, 617315
CYP21A2	100%	99.9%	100%	98.5%	Hyperandrogenism, nonclassic type, due to 21-hydroxylase deficiency, 201910;Adrenal hyperplasia, congenital, due to 21-hydroxylase deficiency, 201910
CYP24A1	100%	100%	100%	99.8%	Hypercalcemia, infantile, 1, 143880
CYP26B1	100%	100%	100%	98.7%	Craniosynostosis with radiohumeral fusions and other skeletal and craniofacial anomalies, 614416
CYP26C1	100%	100%	100%	97.6%	Focal facial dermal dysplasia 4, 614974
CYP27A1	100%	100%	100%	99%	Cerebrotendinous xanthomatosis, 213700

CYP27B1	100%	100%	100%	99.4%	Vitamin D-dependent rickets, type I, 264700
CYP2A6	100%	100%	100%	97.8%	{Lung cancer, resistance to}, 211980;Coumarin resistance, 122700;{Nicotine addiction, protection from}, 188890
CYP2B6	100%	100%	100%	98.5%	{Efavirenz central nervous system toxicity, susceptibility to}, 614546;Efavirenz, poor metabolism of, 614546
CYP2C19	100%	100%	100%	99.6%	Proguanil poor metabolizer, 609535;Mephenytoin poor metabolizer, 609535;Clopidogrel, impaired responsiveness to, 609535;Omeprazole poor metabolizer, 609535
CYP2C8	100%	100%	100%	99.8%	{Drug metabolism, altered, CYP2C8-related}, 618018
CYP2C9	100%	100%	100%	99.8%	Warfarin sensitivity, 122700;Tolbutamide poor metabolizer
CYP2R1	100%	100%	100%	99.5%	Rickets due to defect in vitamin D 25-hydroxylation deficiency, 600081
CYP2U1	100%	100%	100%	97%	Spastic paraplegia 56, autosomal recessive, 615030
CYP3A4	100%	100%	100%	99.3%	Vitamin D-dependent rickets, type 3, 619073

CYP4F22	100%	100%	100%	99.4%	Ichthyosis, congenital, autosomal recessive 5, 604777
CYP4V2	100%	100%	100%	99.4%	Bietti crystalline corneoretinal dystrophy, 210370
CYP7B1	100%	100%	100%	99.7%	Spastic paraplegia 5A, autosomal recessive, 270800;Bile acid synthesis defect, congenital, 3, 613812
D2HGDH	100%	100%	100%	99.4%	D-2-hydroxyglutaric aciduria, 600721
DAAM2	100%	100%	100%	99.2%	Nephrotic syndrome, type 24, 619263
DAB1	100%	100%	100%	99.6%	Spinocerebellar ataxia 37, 615945
DACT1	100%	100%	100%	98.6%	Townes-Brocks syndrome 2, 617466
DAG1	100%	100%	100%	98.8%	Muscular dystrophy-dystroglycanopathy (congenital with brain and eye anomalies), type A, 9, 616538;Muscular dystrophy-dystroglycanopathy (limb-girdle), type C, 9, 613818
DAGLA	100%	100%	100%	99%	Neuroocular syndrome 2, paroxysmal type, 168885

DALRD3	100%	100%	100%	98.4%	?Developmental and epileptic encephalopathy 86, 618910
DAND5	87.8%	87.8%	100%	99.4%	
DAO	100%	100%	100%	98.9%	
DAP3	100%	100%	100%	99.6%	
DARS1	100%	100%	100%	99.9%	Hypomyelination with brainstem and spinal cord involvement and leg spasticity, 615281
DARS2	100%	100%	100%	99.5%	Leukoencephalopathy with brain stem and spinal cord involvement and lactate elevation, 611105
DAW1	100%	100%	100%	99.5%	Ciliary dyskinesia, primary, 52, 620570
DAZ1	49.9%	49.5%	46.5%	32.5%	
DAZ2	49.7%	48.6%	46%	29%	
DAZ3	49.9%	48.7%	45.2%	28.4%	
DAZ4	49.5%	48%	45.6%	26.5%	
DBF4	100%	100%	100%	99.5%	
DBH	100%	100%	100%	99.1%	Orthostatic hypotension 1, due to DBH deficiency, 223360

DBR1	100%	100%	100%	99.4%	Xerosis and growth failure with immune and pulmonary dysfunction syndrome, 620510;{Encephalitis, acute, infection (viral)-induced, susceptibility to, 11}, 619441
DBT	100%	100%	100%	99.5%	Maple syrup urine disease, type II, 620699
DCAF12L1	100%	100%	99.1%	70.4%	
DCAF17	100%	100%	100%	99.4%	Woodhouse-Sakati syndrome, 241080
DCAF8	100%	100%	100%	99.4%	?Giant axonal neuropathy 2, autosomal dominant, 610100
DCC	100%	100%	100%	99.2%	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
DCDC1	100%	100%	100%	99.8%	
DCDC2	100%	100%	100%	99.4%	Nephronophthisis 19, 616217;?Deafness, autosomal recessive 66, 610212;Sclerosing cholangitis, neonatal, 617394

DCHS1	100%	100%	100%	99.2%	Mitral valve prolapse 2, 607829;Van Maldergem syndrome 1, 601390
DCLRE1B	100%	100%	100%	99%	Dyskeratosis congenita, autosomal recessive 8, 620133
DCLRE1C	97.1%	97.1%	100%	99.6%	Severe combined immunodeficiency, Athabascan type, 602450;Omenn syndrome, 603554
DCN	95.1%	95.1%	100%	99.6%	Corneal dystrophy, congenital stromal, 610048
DCPS	100%	100%	100%	99.1%	Al-Raqad syndrome, 616459
DCT	100%	99.9%	100%	97.9%	Oculocutaneous albinism, type VIII, 619165
DCTN1	100%	100%	100%	98.9%	Perry syndrome, 168605;{Amyotrophic lateral sclerosis, susceptibility to}, 105400;Neuronopathy, distal hereditary motor, autosomal dominant 14, 607641
DCTN2	100%	100%	100%	99.5%	
DCX	100%	100%	99.2%	70.7%	Subcortical laminal heterotopia, X-linked, 300067;Lissencephaly, X-linked, 300067
DCXR	100%	100%	100%	99.1%	[Pentosuria], 260800

DDB1	100%	100%	100%	99.2%	White-Kernohan syndrome, 619426
DDB2	100%	100%	100%	98.8%	Xeroderma pigmentosum, group E, DDB-negative subtype, 278740
DDC	100%	100%	100%	99.2%	Aromatic L-amino acid decarboxylase deficiency, 608643
DDHD1	100%	100%	100%	98.7%	Spastic paraplegia 28, autosomal recessive, 609340
DDHD2	100%	100%	100%	99.8%	Spastic paraplegia 54, autosomal recessive, 615033
DDOST	100%	100%	100%	99.1%	Congenital disorder of glycosylation, type Ir, 614507
DDR1	100%	100%	100%	99.2%	
DDR2	100%	100%	100%	99.6%	Warburg-Cinotti syndrome, 618175;Spondylometaepiphyseal dysplasia, short limb-hand type, 271665
DDRGK1	100%	100%	100%	98.8%	Spondyloepimetaphyseal dysplasia, Shohat type, 602557
DDX11	100%	100%	100%	99.7%	Warsaw breakage syndrome, 613398
DDX17	100%	100%	100%	99.3%	
DDX23	100%	100%	100%	99.4%	

DDX3X	99.4%	98.5%	99.1%	71%	Intellectual developmental disorder, X-linked syndromic, Snijders Blok type, 300958
DDX3Y	50%	49.8%	48.8%	22.1%	
DDX41	100%	100%	100%	98.7%	{Myeloproliferative/lymphoproliferative neoplasms, familial (multiple types), susceptibility to}, 616871
DDX59	100%	100%	100%	99.8%	Orofaciodigital syndrome V, 174300
DDX6	100%	100%	100%	99.7%	Intellectual developmental disorder with impaired language and dysmorphic facies, 618653
DEAF1	93.6%	91.3%	98.6%	94%	Vulto-van Silfout-de Vries syndrome, 615828;Neurodevelopmental disorder with hypotonia, impaired expressive language, and with or without seizures, 617171
DEF6	100%	100%	100%	97.5%	Immunodeficiency 87 and autoimmunity, 619573
DEGS1	100%	100%	100%	99.6%	Leukodystrophy, hypomyelinating, 18, 618404
DENND5A	100%	100%	100%	99.3%	Developmental and epileptic encephalopathy 49, 617281
DENND5B	95.9%	95.9%	100%	99.5%	

DEPDC5	100%	100%	100%	99.4%	Epilepsy, familial focal, with variable foci 1, 604364;Developmental and epileptic encephalopathy 111, 620504
DES	100%	100%	100%	98.2%	Scapuloperoneal syndrome, neurogenic, Kaeser type, 181400;Cardiomyopathy, dilated, 1I, 604765;Myopathy, myofibrillar, 1, 601419
DGAT1	100%	100%	100%	99.4%	Diarrhea 7, protein-losing enteropathy type, 615863
DGAT2	100%	100%	100%	99.4%	
DGCR8	100%	100%	100%	99.1%	
DGKE	100%	100%	100%	99.5%	{Hemolytic uremic syndrome, atypical, susceptibility to, 7}, 615008;Nephrotic syndrome, type 7, 615008
DGUOK	100%	100%	100%	99.8%	Portal hypertension, noncirrhotic, 1, 617068;Progressive external ophthalmoplegia with mitochondrial DNA deletions, autosomal recessive 4, 617070;Mitochondrial DNA depletion syndrome 3 (hepatocerebral type), 251880
DHCR24	100%	100%	100%	98.8%	Desmosterolosis, 602398

DHCR7	96.2%	96.1%	100%	99.4%	Smith-Lemli-Opitz syndrome, 270400
DHDDS	73.7%	73.7%	100%	99%	Developmental delay and seizures with or without movement abnormalities, 617836;?Congenital disorder of glycosylation, type 1bb, 613861;Retinitis pigmentosa 59, 613861
DHFR	100%	100%	100%	99.5%	Megaloblastic anemia due to dihydrofolate reductase deficiency, 613839
DHH	100%	100%	100%	99.3%	46XY gonadal dysgenesis with minifascicular neuropathy, 607080;46XY sex reversal 7, 233420
DHODH	100%	100%	100%	98.9%	Miller syndrome, 263750
DHPS	97%	93.3%	100%	98.4%	Neurodevelopmental disorder with seizures and speech and walking impairment, 618480
DHRSX	50%	50%	50%	49.7%	Congenital disorder of glycosylation, type 1DD, 301133
DHTKD1	100%	100%	100%	99.4%	?Charcot-Marie-Tooth disease, axonal, type 2Q, 615025;Alpha-aminoadipic and alpha-ketoadipic aciduria, 204750

DHX16	100%	100%	100%	99.2%	Neuromuscular disease and ocular or auditory anomalies with or without seizures, 618733
DHX30	100%	100%	100%	98.8%	Neurodevelopmental disorder with variable motor and speech impairment, 617804
DHX37	100%	100%	100%	98.5%	Neurodevelopmental disorder with brain anomalies and with or without vertebral or cardiac anomalies, 618731;46XY sex reversal 11, 273250
DHX38	100%	100%	100%	99.2%	Retinitis pigmentosa 84, 618220
DHX9	100%	100%	100%	99.8%	Intellectual developmental disorder, autosomal dominant 75, 620988
DIABLO	100%	100%	100%	99.6%	Deafness, autosomal dominant 64, 614152
DIAPH1	100%	100%	100%	99.2%	Deafness, autosomal dominant 1, with or without thrombocytopenia, 124900;Seizures, cortical blindness, microcephaly syndrome, 616632
DIAPH2	100%	99.6%	99%	74%	?Premature ovarian failure 2A, 300511
DIAPH3	100%	100%	100%	99.8%	Auditory neuropathy, autosomal dominant 1, 609129

DICER1	100%	100%	100%	99.6%	Pleuropulmonary blastoma, 601200;Goiter, multinodular 1, with or without Sertoli-Leydig cell tumors, 138800;GLOW syndrome, somatic mosaic, 618272;Rhabdomyosarcoma, embryonal, 2, 180295
DIO1	100%	100%	100%	99.2%	Thyroid hormone metabolism, abnormal, 2, 619855
DIP2B	100%	100%	100%	99.6%	Intellectual developmental disorder, autosomal dominant, FRA12A type, 136630
DIP2C	100%	100%	100%	99%	
DIS3	100%	100%	100%	99.7%	
DIS3L2	100%	99.9%	100%	99.3%	Perlman syndrome, 267000
DISP1	100%	100%	100%	99.4%	
DKC1	100%	99.4%	99%	70.2%	?Cataracts, hearing impairment, nephrotic syndrome, and enterocolitis 1, 301108;Dyskeratosis congenita, X-linked, 305000
DLAT	100%	100%	100%	99.7%	Pyruvate dehydrogenase E2 deficiency, 245348
DLC1	100%	100%	100%	99.4%	Colorectal cancer, somatic, 114500
DLD	100%	100%	100%	99.7%	Dihydrolipoamide dehydrogenase deficiency, 246900

DLG3	100%	99.6%	98.8%	69.5%	Intellectual developmental disorder, X-linked 90, 300850
DLG4	100%	100%	100%	98.7%	Intellectual developmental disorder, autosomal dominant 62, 618793
DLG5	100%	100%	100%	99.2%	Yuksel-Vogel-Bauser syndrome, 620703
DLK1	100%	100%	100%	97.9%	
DLL1	100%	100%	100%	98.8%	Neurodevelopmental disorder with nonspecific brain abnormalities and with or without seizures, 618709
DLL3	100%	100%	100%	97.8%	Spondylocostal dysostosis 1, autosomal recessive, 277300
DLL4	100%	100%	100%	98.6%	Adams-Oliver syndrome 6, 616589
DLST	100%	100%	100%	99.3%	Pheochromocytoma/paraganglioma syndrome 7, 618475
DLX3	100%	100%	100%	97.7%	Trichodontoosseous syndrome, 190320;Amelogenesis imperfecta, type IV, 104510
DLX4	100%	100%	100%	99.1%	?Orofacial cleft 15, 616788
DLX5	100%	100%	100%	98.8%	Split-hand/foot malformation 1, 183600;?Split-hand/foot malformation 1 with sensorineural hearing loss, 220600

DLX6	100%	100%	98.2%	87%	
DMAC1	100%	100%	100%	97.8%	
DMAC2	100%	100%	100%	98.8%	
DMAC2L	100%	100%	100%	98.7%	
DMC1	100%	100%	100%	99.7%	
DMD	99.6%	99.3%	99.3%	74.1%	Becker muscular dystrophy, 300376;Cardiomyopathy, dilated, 3B, 302045;Duchenne muscular dystrophy, 310200
DMGDH	100%	100%	100%	99.4%	Dimethylglycine dehydrogenase deficiency, 605850
DMP1	100%	100%	100%	98.8%	Hypophosphatemic rickets, AR, 241520
DMPK	100%	100%	100%	98.6%	Myotonic dystrophy 1, 160900
DMRT1	100%	100%	100%	99.4%	
DMRT2	100%	100%	100%	99.4%	
DMXL2	100%	100%	100%	99.7%	Developmental and epileptic encephalopathy 81, 618663;?Deafness, autosomal dominant 71, 617605;?Polyendocrine-polyneuropathy syndrome, 616113

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
DNAAF1	100%	100%	100%	99.1%	Ciliary dyskinesia, primary, 13, 613193
DNAAF11	100%	100%	100%	99.7%	Ciliary dyskinesia, primary, 19, 614935
DNAAF2	100%	100%	100%	99.2%	Ciliary dyskinesia, primary, 10, 612518
DNAAF3	100%	100%	100%	98.8%	Ciliary dyskinesia, primary, 2, 606763
DNAAF4	100%	100%	100%	99.8%	{Dyslexia, susceptibility to, 1}, 127700;Ciliary dyskinesia, primary, 25, 615482
DNAAF5	100%	99.8%	100%	97.5%	Ciliary dyskinesia, primary, 18, 614874
DNAAF6	100%	99.9%	99.1%	75.6%	Ciliary dyskinesia, primary, 36, X-linked, 300991
DNAH1	100%	100%	100%	99%	Spermatogenic failure 18, 617576;Ciliary dyskinesia, primary, 37, 617577
DNAH10	100%	100%	100%	99.4%	Spermatogenic failure 56, 619515

DNAH11	100%	100%	100%	99.7%	Ciliary dyskinesia, primary, 7, with or without situs inversus, 611884
DNAH17	100%	100%	100%	98.8%	Spermatogenic failure 39, 618643
DNAH2	99.9%	99.7%	100%	98.9%	Spermatogenic failure 45, 619094
DNAH3	100%	100%	100%	99.3%	
DNAH5	100%	100%	100%	99.7%	Ciliary dyskinesia, primary, 3, with or without situs inversus, 608644
DNAH6	100%	100%	100%	99.8%	
DNAH7	100%	100%	100%	99.8%	Ciliary dyskinesia, primary, 50, 620356
DNAH8	100%	100%	100%	99.8%	Spermatogenic failure 46, 619095
DNAH9	100%	100%	100%	99.3%	Ciliary dyskinesia, primary, 40, 618300
DNAI1	100%	100%	100%	99.5%	Ciliary dyskinesia, primary, 1, with or without situs inversus, 244400
DNAI2	100%	100%	100%	99.1%	Ciliary dyskinesia, primary, 9, with or without situs inversus, 612444
DNAJA3	100%	100%	100%	99.2%	
DNAJB11	100%	100%	100%	99.4%	Polycystic kidney disease 6 with or without polycystic liver disease, 618061

DNAJB13	100%	100%	100%	98.9%	Ciliary dyskinesia, primary, 34, 617091
DNAJB2	100%	100%	100%	99.5%	Neuronopathy, distal hereditary motor, autosomal recessive 5, 614881
DNAJB4	100%	100%	100%	99.8%	Congenital myopathy 21 with early respiratory failure, 620326
DNAJB5	100%	100%	100%	99%	
DNAJB6	100%	100%	100%	99.7%	Muscular dystrophy, limb-girdle, autosomal dominant 1, 603511
DNAJC12	100%	100%	100%	99.6%	Hyperphenylalaninemia, mild, non-BH4-deficient, 617384
DNAJC19	100%	100%	100%	99.9%	3-methylglutaconic aciduria, type V, 610198
DNAJC21	100%	100%	100%	98.4%	Bone marrow failure syndrome 3, 617052
DNAJC3	100%	100%	100%	99.9%	Ataxia, combined cerebellar and peripheral, with hearing loss and diabetes mellitus, 616192
DNAJC30	100%	100%	100%	99%	Leber-like hereditary optic neuropathy, autosomal recessive 1, 619382
DNAJC5	100%	100%	100%	98.6%	Ceroid lipofuscinosis, neuronal, 4 (Kufs type), autosomal dominant, 162350

DNAJC6	100%	100%	100%	99.5%	Parkinson disease 19a, juvenile-onset, 615528;Parkinson disease 19b, early-onset, 615528
DNAL1	100%	100%	100%	99.6%	Ciliary dyskinesia, primary, 16, 614017
DNAL4	100%	100%	100%	97.5%	?Mirror movements 3, 616059
DNALI1	100%	100%	100%	98.7%	Spermatogenic failure 83, 620354
DNASE1	100%	100%	100%	99.4%	{Systemic lupus erythematosus, susceptibility to}, 152700
DNASE1L3	100%	100%	100%	99.7%	Systemic lupus erythematosus 16, 614420
DNASE2	100%	100%	100%	99%	Autoinflammatory-pancytopenia syndrome, 619858
DNHD1	100%	100%	100%	99.4%	Spermatogenic failure 65, 619712
DNM1	100%	100%	100%	98.4%	Developmental and epileptic encephalopathy 31B, autosomal recessive, 620352;Developmental and epileptic encephalopathy 31A, autosomal dominant, 616346

DNM1L	100%	100%	100%	99.6%	Optic atrophy 5, 610708;Encephalopathy, lethal, due to defective mitochondrial peroxisomal fission 1, 614388
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
DNMBP	100%	100%	100%	99.4%	Cataract 48, 618415
DNMT1	99.9%	99.1%	100%	99.4%	Neuropathy, hereditary sensory, type IE, 614116;Cerebellar ataxia, deafness, and narcolepsy, autosomal dominant, 604121
DNMT3A	100%	100%	100%	98.9%	Tatton-Brown-Rahman syndrome, 615879;Acute myeloid leukemia, somatic, 601626;Heyn-Sproul-Jackson syndrome, 618724
DNMT3B	100%	100%	100%	99.1%	Immunodeficiency-centromeric instability-facial anomalies syndrome 1, 242860;Facioscapulohumeral muscular dystrophy 4, digenic, 619478

DOCK11	100%	99.8%	99.2%	74.8%	Autoinflammatory disease, multisystem, with immune dysregulation, X-linked, 301109
DOCK2	100%	100%	100%	99.7%	Immunodeficiency 40, 616433
DOCK3	100%	100%	100%	99.6%	Neurodevelopmental disorder with impaired intellectual development, hypotonia, and ataxia, 618292
DOCK4	100%	100%	100%	99.6%	
DOCK6	100%	100%	100%	98.8%	Adams-Oliver syndrome 2, 614219
DOCK7	100%	100%	100%	99.8%	Developmental and epileptic encephalopathy 23, 615859
DOCK8	98.6%	98.6%	100%	99.6%	Hyper-IgE syndrome 2, autosomal recessive, with recurrent infections, 243700
DOHH	100%	100%	100%	99.2%	Neurodevelopmental disorder with microcephaly, cerebral atrophy, and visual impairment, 620066
DOK7	100%	100%	100%	98.7%	Fetal akinesia deformation sequence 3, 618389;Myasthenic syndrome, congenital, 10, 254300
DOLK	100%	100%	100%	98.9%	Congenital disorder of glycosylation, type Im, 610768

DONSON	100%	100%	100%	99.8%	Microcephaly, short stature, and limb abnormalities, 617604;Microcephaly-micromelia syndrome, 251230
DOT1L	100%	100%	100%	98.3%	
DPAGT1	100%	100%	100%	99.5%	Myasthenic syndrome, congenital, 13, with tubular aggregates, 614750;Congenital disorder of glycosylation, type Ij, 608093
DPCD	100%	100%	100%	98.3%	
DPF2	95.7%	95.7%	100%	99.3%	Coffin-Siris syndrome 7, 618027
DPH1	100%	100%	100%	99%	Developmental delay with short stature, dysmorphic facial features, and sparse hair, 616901
DPH2	100%	100%	100%	99.3%	Developmental delay with short stature, dysmorphic facial features, and sparse hair 2, 620062
DPH5	100%	100%	100%	99.9%	Neurodevelopmental disorder with short stature, prominent forehead, and feeding difficulties, 620070
DPM1	98.3%	93.5%	97.2%	90.4%	Congenital disorder of glycosylation, type Ie, 608799

DPM2	100%	100%	100%	99.5%	Congenital disorder of glycosylation, type Iu, 615042
DPM3	100%	100%	100%	98.3%	?Muscular dystrophy-dystroglycanopathy (congenital with impaired intellectual development), type B, 15, 618992;Muscular dystrophy-dystroglycanopathy (limb-girdle), type C, 15, 612937
DPP6	100%	100%	100%	98.4%	Intellectual developmental disorder, autosomal dominant 33, 616311;{Ventricular fibrillation, paroxysmal familial, 2}, 612956
DPP9	100%	100%	100%	98.6%	Hatipoglu immunodeficiency syndrome, 620331
DPY19L2	100%	100%	100%	99.6%	Spermatogenic failure 9, 613958
DPYD	100%	100%	100%	99.8%	Dihydropyrimidine dehydrogenase deficiency, 274270;5-fluorouracil toxicity, 274270
DPYS	100%	100%	100%	99.4%	Dihydropyrimidinuria, 222748
DPYSL2	100%	100%	100%	99.3%	
DPYSL5	100%	100%	100%	99.2%	Ritscher-Schinzel syndrome 4, 619435

DRAM2	100%	100%	100%	99.8%	Cone-rod dystrophy 21, 616502
DRC1	100%	100%	100%	99.1%	Spermatogenic failure 80, 620222;Ciliary dyskinesia, primary, 21, 615294
DRD4	100%	100%	100%	96.8%	{Attention deficit-hyperactivity disorder}, 143465;Autonomic nervous system dysfunction
DRG1	100%	100%	100%	99.7%	Tan-Almurshedi syndrome, 620641
DRP2	100%	99.4%	98.5%	71.5%	
DSC2	100%	100%	100%	99.4%	Arrhythmogenic right ventricular dysplasia 11 with mild palmoplantar keratoderma and woolly hair, 610476;Arrhythmogenic right ventricular dysplasia 11, 610476
DSC3	100%	100%	100%	99.6%	Hypotrichosis and recurrent skin vesicles, 613102
DSE	100%	100%	100%	99.8%	Ehlers-Danlos syndrome, musculocontractural type 2, 615539
DSG1	100%	100%	100%	99.6%	Keratosis palmoplantaris striata I, AD, 148700;Erythroderma, congenital, with palmoplantar keratoderma, hypotrichosis, and hyper IgE, 615508

DSG2	100%	99.9%	100%	99.6%	Cardiomyopathy, dilated, 1BB, 612877;Arrhythmogenic right ventricular dysplasia 10, 610193
DSG3	100%	100%	100%	99.8%	Blistering, acantholytic, of oral and laryngeal mucosa, 619226
DSG4	100%	100%	100%	99.7%	Hypotrichosis 6, 607903
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
DSPP	100%	100%	98.7%	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

DST	100%	100%	100%	99.7%	Neuropathy, hereditary sensory and autonomic, type VI, 614653;Epidermolysis bullosa simplex 3, localized or generalized intermediate, with bp230 deficiency, 615425
DSTYK	100%	100%	100%	99.3%	Spastic paraplegia 23, autosomal recessive, 270750;Congenital anomalies of kidney and urinary tract 1, 610805
DTNA	98.5%	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
DTNBP1	100%	100%	100%	99.5%	Hermansky-Pudlak syndrome 7, 614076
DTYMK	100%	100%	100%	99.1%	Neurodegeneration, childhood-onset, with progressive microcephaly, 619847
DUOX2	100%	100%	100%	98.8%	Thyroid dyshormonogenesis 6, 607200
DUOXA2	100%	100%	100%	98.4%	Thyroid dyshormonogenesis 5, 274900

DUSP6	100%	100%	100%	99.5%	Hypogonadotropic hypogonadism 19 with or without anosmia, 615269
DUT	100%	100%	100%	98.9%	Bone marrow failure and diabetes mellitus syndrome, 620044
DVL1	100%	100%	100%	99.2%	Robinow syndrome, autosomal dominant 2, 616331
DVL3	100%	100%	100%	98.3%	Robinow syndrome, autosomal dominant 3, 616894
DYM	100%	100%	100%	99.8%	Smith-McCort dysplasia, 607326;Dyggve-Melchior-Clausen disease, 223800
DYNC1H1	99.3%	99.3%	100%	99.3%	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
DYNC1I2	100%	100%	100%	99.3%	Neurodevelopmental disorder with microcephaly and structural brain anomalies, 618492
DYNC2H1	100%	100%	100%	99.7%	Short-rib thoracic dysplasia 3 with or without polydactyly, 613091

DYNC2I1	100%	100%	100%	99.5%	Short-rib thoracic dysplasia 8 with or without polydactyly, 615503
DYNC2I2	100%	100%	100%	98.5%	Short-rib thoracic dysplasia 11 with or without polydactyly, 615633
DYNC2LI1	100%	100%	100%	99.8%	Short-rib thoracic dysplasia 15 with polydactyly, 617088
DYNLT2B	100%	100%	100%	98.8%	Short-rib thoracic dysplasia 17 with or without polydactyly, 617405
DYRK1A	100%	100%	100%	99.7%	Intellectual developmental disorder, autosomal dominant 7, 614104
DYRK1B	96.2%	96.1%	100%	98.6%	Abdominal obesity-metabolic syndrome 3, 615812
DYSF	96.9%	96.9%	100%	99.2%	Muscular dystrophy, limb-girdle, autosomal recessive 2, 253601;Miyoshi muscular dystrophy 1, 254130;Myopathy, distal, with anterior tibial onset, 606768
DZIP1	100%	100%	100%	99.7%	Spermatogenic failure 47, 619102;?Mitral valve prolapse 3, 610840
DZIP1L	100%	100%	100%	99.1%	Polycystic kidney disease 5, 617610
E2F1	100%	99.3%	99.8%	96.1%	

EARS2	100%	100%	100%	99.4%	Combined oxidative phosphorylation deficiency 12, 614924
EBF3	100%	100%	100%	98.1%	Hypotonia, ataxia, and delayed development syndrome, 617330
EBP	100%	99.9%	98.7%	68.3%	MEND syndrome, 300960;Chondrodysplasia punctata, X-linked dominant, 302960
ECE1	100%	100%	100%	98.9%	{Hypertension, essential, susceptibility to}, 145500;?Hirschsprung disease, cardiac defects, and autonomic dysfunction, 613870
ECEL1	100%	100%	100%	97.4%	Arthrogryposis, distal, type 5D, 615065
ECHS1	100%	100%	100%	99.4%	Mitochondrial short-chain enoyl-CoA hydratase 1 deficiency, 616277
ECM1	100%	100%	100%	98.5%	Urbach-Wiethe disease, 247100
ECSIT	100%	100%	100%	98.9%	
EDA	100%	99.6%	98%	65.3%	Tooth agenesis, selective, X-linked 1, 313500;Ectodermal dysplasia 1, hypohidrotic, X-linked, 305100

EDAR	100%	100%	100%	99.3%	[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
EDARADD	100%	100%	100%	99.7%	Ectodermal dysplasia 11B, hypohidrotic/hair/tooth type, autosomal recessive, 614941;Ectodermal dysplasia 11A, hypohidrotic/hair/tooth type, autosomal dominant, 614940
EDC3	100%	100%	100%	99.2%	?Intellectual developmental disorder, autosomal recessive 50, 616460
EDEM3	100%	100%	100%	99.7%	Congenital disorder of glycosylation, type IIv, 619493
EDN1	100%	100%	100%	99.8%	Question mark ears, isolated, 612798;Auriculocondylar syndrome 3, 615706

EDN3	100%	100%	100%	99.8%	Waardenburg syndrome, type 4B, 613265;{Hirschsprung disease, susceptibility to, 4}, 613712
EDNRA	100%	100%	100%	99.6%	{Migraine, resistance to}, 157300;Mandibulofacial dysostosis with alopecia, 616367
EDNRB	100%	100%	100%	99.4%	{Hirschsprung disease, susceptibility to, 2}, 600155;?ABCD syndrome, 600501;Waardenburg syndrome, type 4A, 277580
EED	99.1%	99.1%	99.6%	93.8%	Cohen-Gibson syndrome, 617561
EEF1A2	99.4%	96.8%	100%	97.6%	Developmental and epileptic encephalopathy 33, 616409;Intellectual developmental disorder, autosomal dominant 38, 616393
EEF1D	100%	100%	100%	99.2%	
EEF2	100%	100%	100%	99%	?Spinocerebellar ataxia 26, 609306
EEFSEC	100%	100%	100%	99.4%	
EFEMP1	100%	100%	100%	99%	Doyne honeycomb degeneration of retina, 126600;Cutis laxa, autosomal recessive, type ID, 620780;Glaucoma 1, open angle, H, 611276

EFEMP2	100%	100%	100%	99%	Cutis laxa, autosomal recessive, type IB, 614437
EFHC1	97.9%	97.5%	100%	99.7%	{Epilepsy, juvenile absence, susceptibility to, 1}, 607631;{Myoclonic epilepsy, juvenile, susceptibility to, 1}, 254770
EFL1	99.2%	99.2%	100%	99.8%	Shwachman-Diamond syndrome 2, 617941
EFNA4	100%	100%	100%	98.6%	
EFNB1	100%	99.8%	97.5%	67.7%	Craniofrontonasal dysplasia, 304110
EFNB2	100%	100%	100%	99.7%	
EFTUD2	100%	100%	100%	99.1%	Mandibulofacial dysostosis, Guion-Almeida type, 610536
EGF	100%	100%	100%	99.6%	?Hypomagnesemia 4, renal, 611718
EGFR	100%	100%	100%	99.4%	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

EGLN1	100%	100%	100%	95.4%	Erythrocytosis, familial, 3, 609820;[Hemoglobin, high altitude adaptation], 609070
EGLN2	100%	100%	100%	99.4%	
EGR2	100%	100%	100%	98.2%	Dejerine-Sottas disease, 145900;Charcot-Marie-Tooth disease, type 1D, 607678;Hypomyelinating neuropathy, congenital, 1, 605253
EHD1	100%	100%	100%	98.4%	
EHHADH	100%	100%	100%	99.2%	?Fanconi renotubular syndrome 3, 615605
EHMT1	99.9%	99.7%	100%	98.9%	Kleefstra syndrome 1, 610253
EIF1AY	50%	49.9%	49.4%	21.9%	
EIF2AK1	100%	100%	100%	99.7%	?Leukoencephalopathy, motor delay, spasticity, and dysarthria syndrome, 618878
EIF2AK2	100%	100%	100%	99.6%	Leukoencephalopathy, developmental delay, and episodic neurologic regression syndrome, 618877;Dystonia 33, 619687
EIF2AK3	100%	100%	100%	99.7%	Wolcott-Rallison syndrome, 226980
EIF2AK4	100%	100%	100%	99.6%	Pulmonary venoocclusive disease 2, 234810

EIF2B1	100%	100%	100%	99.3%	Leukoencephalopathy with vanishing white matter 1, with or without ovarian failure, 603896
EIF2B2	100%	100%	100%	97.8%	Leukoencephalopathy with vanishing white matter 2, with or without ovarian failure, 620312
EIF2B3	100%	100%	100%	99.3%	Leukoencephalopathy with vanishing white matter 3, with or without ovarian failure, 620313
EIF2B4	100%	100%	100%	99.6%	Leukoencephalopathy with vanishing white matter 4, with or without ovarian failure, 620314
EIF2B5	100%	100%	100%	99.4%	Leukoencephalopathy with vanishing white matter 5, with or without ovarian failure, 620315
EIF2S3	100%	99.9%	99.2%	72.1%	MEHMO syndrome, 300148
EIF3F	100%	100%	100%	99.5%	Intellectual developmental disorder, autosomal recessive 67, 618295
EIF4A2	100%	100%	100%	99.7%	Neurodevelopmental disorder with hypotonia and speech delay, with or without seizures, 620455
EIF4A3	100%	100%	100%	99.1%	Robin sequence with cleft mandible and limb anomalies, 268305

EIF4ENIF1	100%	100%	100%	99.4%	
EIF5A	100%	100%	100%	99.3%	Faundes-Banka syndrome, 619376
ELAC2	100%	100%	100%	99.4%	{Prostate cancer, hereditary, 2, susceptibility to}, 614731;Combined oxidative phosphorylation deficiency 17, 615440
ELANE	100%	100%	100%	98.6%	Neutropenia, cyclic, 162800;Neutropenia, severe congenital 1, autosomal dominant, 202700
ELF2	100%	100%	100%	99.8%	
ELF4	100%	99.3%	98.7%	66.8%	Autoinflammatory syndrome, familial, X-linked, Behcet-like 2, 301074
ELMO2	100%	100%	100%	99.8%	Vascular malformation, primary intraosseous, 606893
ELMOD3	100%	100%	100%	99.7%	?Deafness, autosomal recessive 88, 615429;?Deafness, autosomal dominant 81, 619500
ELN	100%	100%	100%	99.1%	Cutis laxa, autosomal dominant, 123700;Supravalvar aortic stenosis, 185500

ELOVL1	100%	100%	100%	99.1%	Ichthyotic keratoderma, spasticity, hypomyelination, and dysmorphic facies, 618527
ELOVL4	100%	100%	100%	99%	Spinocerebellar ataxia 34, 133190;Stargardt disease 3, 600110;Ichthyosis, spastic quadriplegia, and impaired intellectual development, 614457
ELOVL5	100%	100%	100%	99.4%	Spinocerebellar ataxia 38, 615957
ELP1	100%	100%	100%	99.8%	{Medulloblastoma}, 155255;Dysautonomia, familial, 223900
ELP2	100%	100%	100%	99.7%	Intellectual developmental disorder, autosomal recessive 58, 617270
ELP4	87.8%	87.8%	100%	99.8%	?Aniridia 2, 617141
EMC1	100%	100%	100%	99.2%	Cerebellar atrophy, visual impairment, and psychomotor retardation, 616875
EMC10	100%	100%	100%	98.5%	Neurodevelopmental disorder with dysmorphic facies and variable seizures, 619264
EMD	94.2%	89.4%	96.7%	63.6%	Emery-Dreifuss muscular dystrophy 1, X-linked, 310300

EMG1	100%	100%	100%	98.4%	Bowen-Conradi syndrome, 211180
EMILIN1	100%	100%	100%	98.6%	Neuronopathy, distal hereditary motor, autosomal dominant 10, 620080;Arterial tortuosity-bone fragility syndrome, 620908
EML1	100%	100%	100%	98.2%	Band heterotopia, 600348
EMP2	100%	100%	100%	98.9%	Nephrotic syndrome, type 10, 615861
EMX2	100%	100%	99.9%	91.2%	Schizencephaly, 269160
EN1	100%	100%	100%	95.8%	?ENDOVE syndrome, limb-brain type, 619218
ENAM	100%	100%	100%	99.7%	Amelogenesis imperfecta, type IC, 204650;Amelogenesis imperfecta, type IB, 104500
ENG	100%	100%	100%	96.9%	Telangiectasia, hereditary hemorrhagic, type 1, 187300
ENO3	100%	100%	100%	98.4%	Glycogen storage disease XIII, 612932

ENPP1	99.9%	99.3%	100%	99.6%	{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
ENTPD1	100%	100%	100%	99.7%	Spastic paraplegia 64, autosomal recessive, 615683
EOGT	97.8%	93.7%	100%	99.8%	Adams-Oliver syndrome 4, 615297
EP300	100%	100%	100%	99.2%	Menke-Hennekam syndrome 2, 618333;Colorectal cancer, somatic, 114500;Rubinstein-Taybi syndrome 2, 613684
EP400	100%	100%	100%	98.9%	
EPAS1	100%	100%	100%	99.4%	Erythrocytosis, familial, 4, 611783
EPB41	100%	100%	100%	99.5%	Elliptocytosis-1, 611804
EPB41L1	100%	100%	100%	99.4%	?Intellectual developmental disorder, autosomal dominant 11, 614257
EPB41L3	100%	100%	100%	99.4%	

EPB42	100%	100%	100%	98.6%	Spherocytosis, type 5, 612690
EPCAM	100%	100%	100%	99.3%	Diarrhea 5, with tufting enteropathy, congenital, 613217;Lynch syndrome 8, 613244
EPG5	100%	100%	100%	99.6%	Vici syndrome, 242840
EPHA10	100%	100%	100%	98.2%	?Deafness, autosomal dominant 88, 620283
EPHA2	100%	100%	100%	98.8%	Cataract 6, multiple types, 116600
EPHA7	100%	100%	100%	99.8%	
EPHB2	99.9%	99.2%	100%	98.6%	?Bleeding disorder, platelet-type, 22, 618462;{Prostate cancer/brain cancer susceptibility, somatic}, 603688
EPHB4	100%	100%	100%	99.1%	Capillary malformation-arteriovenous malformation 2, 618196;Lymphatic malformation 7, 617300
EPHX1	100%	100%	100%	98.7%	
EPHX2	100%	100%	100%	99.3%	{Hypercholesterolemia, familial, due to LDLR defect, modifier of}, 143890
EPM2A	100%	100%	99.3%	93.4%	Myoclonic epilepsy of Lafora 1, 254780

EPO	100%	100%	100%	98.8%	{Microvascular complications of diabetes 2}, 612623;Erythrocytosis, familial, 5, 617907;?Diamond-Blackfan anemia-like, 617911
EPRS1	100%	100%	100%	99.2%	Leukodystrophy, hypomyelinating, 15, 617951
EPS8	100%	100%	100%	99.7%	?Deafness, autosomal recessive 102, 615974
EPS8L2	100%	100%	100%	98.1%	Deafness autosomal recessive 106, 617637
EPS8L3	100%	100%	100%	98.6%	?Hypotrichosis 5, 612841
ERAL1	100%	99.9%	100%	99.6%	Perrault syndrome 6, 617565
ERBB2	100%	100%	100%	98.4%	Gastric cancer, somatic, 613659;Adenocarcinoma of lung, somatic, 211980;Ovarian cancer, somatic, 167000;?Visceral neuropathy, familial, 2, autosomal recessive, 619465;Glioblastoma, somatic, 137800

ERBB3	100%	100%	100%	98.8%	?Lethal congenital contractural syndrome 2, 607598;{?Erythroleukemia, familial, susceptibility to}, 133180;Visceral neuropathy, familial, 1, autosomal recessive, 243180
ERBB4	100%	100%	100%	99.6%	Amyotrophic lateral sclerosis 19, 615515
ERBIN	100%	100%	100%	99.8%	
ERCC1	100%	100%	100%	98.6%	Cerebrooculofacioskeletal syndrome 4, 610758
ERCC2	99.9%	98.8%	100%	98.9%	Xeroderma pigmentosum, group D, 278730;Trichothiodystrophy 1, photosensitive, 601675;?Cerebrooculofacioskeletal syndrome 2, 610756
ERCC3	100%	100%	100%	99.6%	Trichothiodystrophy 2, photosensitive, 616390;Xeroderma pigmentosum, group B, 610651

ERCC4	100%	100%	100%	99.5%	Xeroderma pigmentosum, type F/Cockayne syndrome, 278760;XFE progeroid syndrome, 610965;Xeroderma pigmentosum, group F, 278760;Fanconi anemia, complementation group Q, 615272
ERCC5	100%	100%	100%	99.5%	Xeroderma pigmentosum, group G, 278780;Cerebrooculofacioskeletal syndrome 3, 616570;Xeroderma pigmentosum, group G/Cockayne syndrome, 278780
ERCC6	100%	100%	100%	99.5%	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
ERCC6L2	100%	100%	100%	99.8%	Bone marrow failure syndrome 2, 615715

ERCC8	95.2%	95.2%	99.9%	96.3%	UV-sensitive syndrome 2, 614621;Cockayne syndrome, type A, 216400
ERF	100%	100%	99.9%	94.5%	Craniosynostosis 4, 600775;Chitayat syndrome, 617180
ERG	100%	100%	100%	99.6%	Lymphatic malformation 14, 620602
ERGIC1	100%	100%	100%	98.9%	?Arthrogryposis multiplex congenita 2, neurogenic type, 208100
ERI1	100%	100%	100%	99.7%	Hoxha-Aliu syndrome, 620662;Spondyloepimetaphyseal dysplasia, Guo-Campeau type, 620663
ERLIN1	100%	100%	100%	99%	Spastic paraplegia 62, autosomal recessive, 615681
ERLIN2	100%	100%	100%	99.2%	Spastic paraplegia 18A, autosomal dominant, 620512;Spastic paraplegia 18B, autosomal recessive, 611225
ERMARD	100%	100%	100%	99.7%	?Periventricular nodular heterotopia 6, 615544
ESAM	100%	100%	100%	99.1%	Neurodevelopmental disorder with intracranial hemorrhage, seizures, and spasticity, 620371

ESCO2	100%	100%	100%	99.7%	Juberg-Hayward syndrome, 216100;Roberts-SC phocomelia syndrome, 268300
ESPN	100%	100%	100%	97.7%	Deafness, neurosensory, without vestibular involvement, autosomal dominant, 609006;Deafness, autosomal recessive 36, 609006;?Usher syndrome, type 1M, 618632
ESR1	100%	100%	100%	99.2%	Breast cancer, somatic, 114480;{Migraine, susceptibility to}, 157300;Estrogen resistance, 615363;{Myocardial infarction, susceptibility to}, 608446
ESR2	100%	100%	100%	99.5%	?Ovarian dysgenesis 8, 618187
ESRP1	100%	100%	100%	99.5%	?Deafness, autosomal recessive 109, 618013
ESRRB	100%	100%	100%	99.3%	Deafness, autosomal recessive 35, 608565
ETFA	82.4%	82.4%	100%	99.6%	Glutaric acidemia IIA, 231680
ETFB	100%	100%	100%	99.6%	Glutaric acidemia IIB, 231680
ETFDH	95.3%	92.7%	100%	99.5%	Glutaric acidemia IIC, 231680

ETHE1	100%	100%	100%	99.3%	Ethylmalonic encephalopathy, 602473
ETV6	100%	100%	100%	99.5%	Thrombocytopenia 5, 616216;Leukemia, acute myeloid, somatic, 601626
EVC	100%	99.8%	100%	99.3%	Ellis-van Creveld syndrome, 225500;?Weyers acrofacial dysostosis, 193530
EVC2	100%	100%	100%	99.1%	Ellis-van Creveld syndrome, 225500;Weyers acrofacial dysostosis, 193530
EWSR1	100%	100%	100%	98.1%	Neuroepithelioma, 612219;Ewing sarcoma, 612219
EXOC2	100%	100%	100%	99.7%	Neurodevelopmental disorder with dysmorphic facies and cerebellar hypoplasia, 619306
EXOC3L2	100%	100%	100%	98.1%	Brain malformation renal syndrome, 620943
EXOC6B	100%	100%	100%	99.3%	Spondyloepimetaphyseal dysplasia with joint laxity, type 3, 618395
EXOC7	100%	100%	100%	99%	Neurodevelopmental disorder with seizures and brain atrophy, 619072
EXOC8	100%	100%	100%	98.9%	?Neurodevelopmental disorder with microcephaly, seizures, and brain atrophy, 619076

EXOSC1	100%	100%	100%	98.7%	?Pontocerebellar hypoplasia, type 1F, 619304
EXOSC2	100%	99.4%	100%	98.7%	Short stature, hearing loss, retinitis pigmentosa, and distinctive facies, 617763
EXOSC3	100%	100%	100%	99.6%	Pontocerebellar hypoplasia, type 1B, 614678
EXOSC5	100%	100%	100%	97%	Cerebellar ataxia, brain abnormalities, and cardiac conduction defects, 619576
EXOSC8	100%	100%	100%	99.9%	Pontocerebellar hypoplasia, type 1C, 616081
EXOSC9	100%	99.9%	100%	99.3%	Pontocerebellar hypoplasia, type 1D, 618065
EXPH5	100%	100%	100%	99.5%	Epidermolysis bullosa simplex 4, localized or generalized intermediate, autosomal recessive, 615028
EXT1	100%	100%	100%	99.5%	Exostoses, multiple, type 1, 133700;Chondrosarcoma, 215300
EXT2	100%	100%	100%	99.7%	Seizures, scoliosis, and macrocephaly syndrome, 616682;Exostoses, multiple, type 2, 133701
EXTL3	100%	99.5%	100%	99.7%	Immunoskeletal dysplasia with neurodevelopmental abnormalities, 617425

EYA1	100%	100%	100%	99.7%	Branchiootic syndrome 1, 602588;Branchiootorenal syndrome 1, with or without cataracts, 113650;Anterior segment anomalies with or without cataract, 602588;?Otofaciocervical syndrome, 166780
EYA4	100%	100%	100%	99.7%	?Cardiomyopathy, dilated, 1J, 605362;Deafness, autosomal dominant 10, 601316
EYS	100%	100%	100%	99.8%	Retinitis pigmentosa 25, 602772
EZH1	100%	100%	100%	99.2%	
EZH2	100%	100%	100%	99.5%	Weaver syndrome, 277590
F10	100%	100%	100%	99.4%	Factor X deficiency, 227600
F11	100%	100%	100%	99.5%	Factor XI deficiency, autosomal dominant, 612416;Factor XI deficiency, autosomal recessive, 612416
F12	100%	100%	100%	99.4%	Angioedema, hereditary, 3, 610618;Factor XII deficiency, 234000
F13A1	100%	100%	100%	99.7%	Factor XIIIA deficiency, 613225;{Myocardial infarction, protection against}, 608446;{Venous thrombosis, protection against}, 188050

F13B	100%	100%	100%	99.9%	Factor XIIIB deficiency, 613235
F2	100%	100%	100%	99.1%	Hypoprothrombinemia, 613679;{Pregnancy loss, recurrent, susceptibility to, 2}, 614390;Dysprothrombinemia, 613679;Thrombophilia 1 due to thrombin defect, 188050;{Stroke, ischemic, susceptibility to}, 601367
F2RL3	100%	100%	100%	99.3%	
F5	100%	100%	100%	99.6%	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
F7	100%	100%	100%	99.1%	{Myocardial infarction, decreased susceptibility to}, 608446;Factor VII deficiency, 227500
F8	100%	99.7%	99.2%	74.9%	Thrombophilia 13, X-linked, due to factor VIII defect, 301071;Hemophilia A, 306700

F9	100%	99.9%	99.7%	76.7%	{Deep venous thrombosis, protection against}, 300807;Hemophilia B, 306900;Thrombophilia 8, X-linked, due to factor IX defect, 300807;{Warfarin sensitivity}, 301052
FA2H	100%	100%	100%	99.1%	Spastic paraplegia 35, autosomal recessive, 612319
FAAH	100%	100%	100%	98.5%	{Drug addiction, susceptibility to}, 606581
FAAP24	100%	100%	100%	99.9%	
FADD	100%	99.9%	100%	97%	Immunodeficiency 90 with encephalopathy, functional hyposplenia, and hepatic dysfunction, 613759
FAH	100%	100%	100%	99.4%	Tyrosinemia, type I, 276700
FAM111A	100%	100%	100%	99.9%	Kenny-Caffey syndrome, type 2, 127000;Gracile bone dysplasia, 602361
FAM111B	100%	100%	100%	99.9%	Poikiloderma, hereditary fibrosing, with tendon contractures, myopathy, and pulmonary fibrosis, 615704
FAM149B1	100%	100%	100%	99.7%	Joubert syndrome 36, 618763
FAM161A	100%	100%	100%	99.7%	Retinitis pigmentosa 28, 606068
FAM177A1	86.5%	86.5%	100%	99.1%	

FAM20A	100%	100%	100%	98.5%	Amelogenesis imperfecta, type IG (enamel-renal syndrome), 204690
FAM20B	100%	100%	100%	99.9%	
FAM20C	100%	100%	100%	97.4%	Raine syndrome, 259775
FAM50A	100%	99.2%	98.1%	65.5%	Intellectual developmental disorder, X-linked syndromic, Armfield type, 300261
FAM83G	100%	100%	100%	98.2%	
FAM83H	100%	100%	100%	98.8%	Amelogenesis imperfecta, type IIIA, 130900
FAN1	100%	100%	100%	99.3%	Interstitial nephritis, karyomegalic, 614817
FANCA	100%	100%	100%	99.4%	Fanconi anemia, complementation group A, 227650
FANCB	96.2%	95.9%	99.1%	73.4%	Fanconi anemia, complementation group B, 300514
FANCC	100%	100%	100%	99.6%	Fanconi anemia, complementation group C, 227645
FANCD2	100%	100%	100%	99.6%	Fanconi anemia, complementation group D2, 227646
FANCE	100%	100%	100%	98.3%	Fanconi anemia, complementation group E, 600901

FANCF	100%	100%	100%	99.4%	Fanconi anemia, complementation group F, 603467
FANCG	100%	100%	100%	98.9%	Fanconi anemia, complementation group G, 614082
FANCI	100%	100%	100%	99.6%	Fanconi anemia, complementation group I, 609053
FANCL	91%	88.1%	99.8%	97.1%	Fanconi anemia, complementation group L, 614083
FANCM	100%	100%	100%	99.8%	Premature ovarian failure 15, 618096;Spermatogenic failure 28, 618086
FAR1	100%	100%	100%	99.9%	Peroxisomal fatty acyl-CoA reductase 1 disorder, 616154;Cataracts, spastic paraparesis, and speech delay, 619338
FARS2	100%	100%	100%	99%	Combined oxidative phosphorylation deficiency 14, 614946;Spastic paraplegia 77, autosomal recessive, 617046
FARSA	100%	100%	100%	99.3%	?Rajab interstitial lung disease with brain calcifications 2, 619013
FARSB	100%	100%	100%	99.4%	Rajab interstitial lung disease with brain calcifications 1, 613658

FAS	100%	99.8%	100%	99.3%	Squamous cell carcinoma, burn scar-related, somatic;Autoimmune lymphoproliferative syndrome, type IA, 601859;{Autoimmune lymphoproliferative syndrome}, 601859
FASLG	100%	100%	100%	99.3%	Autoimmune lymphoproliferative syndrome, type IB, 601859;{Lung cancer, susceptibility to}, 211980
FASTKD2	100%	100%	100%	99.6%	Combined oxidative phosphorylation deficiency 44, 618855
FAT1	100%	100%	100%	99.6%	
FAT2	100%	100%	100%	99.6%	Spinocerebellar ataxia 45, 617769
FAT4	100%	100%	100%	99.6%	Van Maldergem syndrome 2, 615546;Hennekam lymphangiectasia-lymphedema syndrome 2, 616006
FBLN1	100%	100%	100%	98.6%	Synpolydactyly, 3/3'4, associated with metacarpal and metatarsal synostoses, 608180

FBLN5	92.8%	92.8%	100%	99.5%	Cutis laxa, autosomal recessive, type IA, 219100;Charcot-Marie-Tooth disease, demyelinating, type 1H, 619764;Macular degeneration, age-related, 3, 608895;?Cutis laxa, autosomal dominant 2, 614434
FBN1	100%	100%	100%	99.8%	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
FBN2	99.2%	99.2%	100%	99.6%	Macular degeneration, early-onset, 616118;Contractural arachnodactyly, congenital, 121050
FBP1	100%	100%	100%	99.2%	Fructose-1,6-bisphosphatase deficiency, 229700
FBP2	100%	100%	100%	99.7%	?Leukodystrophy, childhood-onset, remitting, 619864
FBRSL1	99%	96.3%	99.5%	87.4%	

FBXL3	100%	100%	100%	99.8%	Intellectual developmental disorder with short stature, facial anomalies, and speech defects, 606220
FBXL4	100%	100%	100%	99.8%	Mitochondrial DNA depletion syndrome 13 (encephalomyopathic type), 615471
FBXO11	100%	100%	100%	99.2%	Intellectual developmental disorder with dysmorphic facies and behavioral abnormalities, 618089
FBXO28	100%	100%	100%	99.8%	Developmental and epileptic encephalopathy 100, 619777
FBXO31	100%	100%	100%	98.3%	?Intellectual developmental disorder, autosomal recessive 45, 615979
FBXO32	100%	100%	100%	99.1%	
FBXO38	100%	100%	100%	99.6%	Neuronopathy, distal hereditary motor, autosomal dominant 6, 615575
FBXO43	100%	100%	100%	99.8%	Spermatogenic failure 64, 619696;Oocyte/zygote/embryo maturation arrest 12, 619697
FBXO7	100%	100%	100%	99.6%	Parkinson disease 15, autosomal recessive, 260300

FBXW11	100%	100%	100%	99.5%	Neurodevelopmental, jaw, eye, and digital syndrome, 618914
FBXW4	100%	100%	100%	98%	
FBXW7	100%	100%	100%	99.6%	Developmental delay, hypotonia, and impaired language, 620012
FCGR3A	99.7%	96.8%	100%	99.3%	Immunodeficiency 20, 615707
FCGR3B	99.3%	97%	95.6%	81.2%	
FCHO1	98%	96.1%	100%	99.1%	Immunodeficiency 76, 619164
FCN3	100%	100%	100%	99%	Immunodeficiency due to ficolin 3 deficiency, 613860
FCSK	100%	100%	100%	99.1%	Congenital disorder of glycosylation with defective fucosylation 2, 618324
FDFT1	100%	100%	100%	98.5%	Squalene synthase deficiency, 618156
FDPS	100%	100%	100%	99%	Porokeratosis 9, multiple types, 616631
FDX2	100%	99.8%	100%	96.9%	Mitochondrial myopathy, episodic, with optic atrophy and reversible leukoencephalopathy, 251900
FDXR	100%	100%	100%	99.4%	Multiple mitochondrial dysfunctions syndrome 9B, 620887;Auditory neuropathy and optic atrophy, 617717

FECH	100%	100%	100%	99.8%	Protoporphyria, erythropoietic, 1, 177000
FEM1B	100%	100%	100%	99.7%	
FERMT1	100%	100%	100%	99.4%	Kindler syndrome, 173650
FERMT3	100%	100%	100%	99.3%	Leukocyte adhesion deficiency, type III, 612840
FEZF1	100%	100%	100%	99.2%	Hypogonadotropic hypogonadism 22, with or without anosmia, 616030
FEZF2	100%	100%	100%	96.3%	
FGA	100%	100%	100%	99.6%	Amyloidosis, hereditary systemic 2, 105200;Hypodysfibrinogenemia, congenital, 616004;Dysfibrinogenemia, congenital, 616004;Afibrinogenemia, congenital, 202400
FGB	100%	100%	100%	99.9%	Hypofibrinogenemia, congenital, 202400;Dysfibrinogenemia, congenital, 616004;Afibrinogenemia, congenital, 202400
FGD1	100%	98.9%	98.7%	67.8%	Intellectual developmental disorder, X-linked syndromic 16, 305400;Aarskog-Scott syndrome, 305400
FGD4	100%	100%	100%	99.8%	Charcot-Marie-Tooth disease, type 4H, 609311

FGF10	100%	100%	100%	100%	LADD syndrome 3, 620193;Aplasia of lacrimal and salivary glands, 180920
FGF12	100%	100%	100%	99.5%	Developmental and epileptic encephalopathy 47, 617166
FGF13	100%	99.3%	98.7%	73.7%	Developmental and epileptic encephalopathy 90, 301058;Intellectual developmental disorder, X-linked 110, 301095
FGF14	100%	100%	100%	99.3%	Spinocerebellar ataxia 27A, 193003;Spinocerebellar ataxia 27B, late-onset, 620174
FGF16	100%	99.3%	97.8%	69.3%	Metacarpal 4-5 fusion, 309630
FGF17	100%	100%	100%	99.2%	Hypogonadotropic hypogonadism 20 with or without anosmia, 615270
FGF20	100%	100%	100%	98.4%	?Renal hypodysplasia/aplasia 2, 615721
FGF23	100%	100%	100%	98.6%	Tumoral calcinosis, hyperphosphatemic, familial, 2, 617993;Hypophosphatemic rickets, autosomal dominant, 193100
FGF3	100%	100%	100%	95.9%	Deafness, congenital with inner ear agenesis, microtia, and microdontia, 610706

FGF5	100%	100%	100%	99.4%	Trichomegaly, 190330
FGF8	100%	100%	100%	97.3%	Hypogonadotropic hypogonadism 6 with or without anosmia, 612702
FGF9	100%	100%	100%	99.9%	Multiple synostoses syndrome 3, 612961
FGFR1	99.8%	98.9%	100%	99.1%	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

FGFR2	100%	100%	100%	99.7%	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

FGFR3	100%	100%	100%	99.2%	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
FGG	100%	100%	100%	100%	Dysfibrinogenemia, congenital, 616004;Hypodysfibrinogenemia, 616004;Hypofibrinogenemia, congenital, 202400;Afibrinogenemia, congenital, 202400
FH	100%	100%	100%	99.7%	Leiomyomatosis and renal cell cancer, 150800;Fumarase deficiency, 606812

FHL1	100%	99.4%	98.4%	71.2%	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
FHL2	100%	100%	100%	99.4%	
FHOD3	100%	100%	100%	99.1%	Cardiomyopathy, familial hypertrophic, 28, 619402
FIBP	100%	100%	100%	99.1%	Thauvin-Robinet-Faivre syndrome, 617107
FICD	100%	100%	100%	99.4%	Spastic paraplegia 92, autosomal recessive, 620911
FIG4	98.4%	98.4%	100%	99.6%	Yunis-Varon syndrome, 216340;?Polymicrogyria, bilateral temporooccipital, 612691;Amyotrophic lateral sclerosis 11, 612577;Charcot-Marie-Tooth disease, type 4J, 611228

FIGLA	100%	100%	100%	99.6%	Premature ovarian failure 6, 612310
FIGN	100%	100%	100%	99.5%	
FIGNL1	100%	100%	100%	99.8%	
FILIP1	100%	100%	100%	99.5%	Neuromuscular disorder, congenital, with dysmorphic facies, 620775
FITM2	100%	100%	100%	99.5%	Siddiqi syndrome, 618635
FKBP10	100%	100%	100%	98.9%	Osteogenesis imperfecta, type XI, 610968;Bruck syndrome 1, 259450
FKBP14	100%	100%	100%	99.7%	Ehlers-Danlos syndrome, kyphoscoliotic type, 2, 614557
FKBP6	100%	100%	100%	99.2%	Spermatogenic failure 77, 620103
FKRP	100%	100%	100%	96.9%	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

FKTN	100%	100%	100%	99.9%	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
FLAD1	100%	100%	100%	99.4%	Lipid storage myopathy due to flavin adenine dinucleotide synthetase deficiency, 255100
FLCN	100%	100%	100%	99.4%	Birt-Hogg-Dube syndrome, 135150;Colorectal cancer, somatic, 114500;Pneumothorax, primary spontaneous, 173600;Renal carcinoma, chromophobe, somatic, 144700
FLG	100%	100%	100%	98.9%	Ichthyosis vulgaris, 146700;{Dermatitis, atopic, susceptibility to, 2}, 605803
FLG2	99.9%	99.9%	99.9%	99.2%	Peeling skin syndrome 6, 618084

FLI1	97%	97%	100%	99.2%	Bleeding disorder, platelet-type, 21, 617443
FLII	100%	100%	100%	98.9%	Cardiomyopathy, dilated, 2J, 620635
FLNA	100%	99.8%	98.2%	67.3%	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
FLNB	100%	100%	100%	99.2%	Larsen syndrome, 150250;Atelosteogenesis, type I, 108720;Atelosteogenesis, type III, 108721;Spondylocarpotarsal synostosis syndrome, 272460;Boomerang dysplasia, 112310

FLNC	100%	100%	100%	99.1%	Cardiomyopathy, familial hypertrophic, 26, 617047;Arrhythmogenic right ventricular dysplasia, familial, 617047;Cardiomyopathy, familial restrictive 5, 617047;Myopathy, distal, 4, 614065;Myopathy, myofibrillar, 5, 609524
FLRT3	100%	100%	100%	99.8%	Hypogonadotropic hypogonadism 21 with anosmia, 615271
FLT3	100%	100%	100%	99.6%	Leukemia, acute lymphoblastic, somatic, 613065;Leukemia, acute myeloid, reduced survival in, somatic, 601626;Leukemia, acute myeloid, somatic, 601626
FLT3LG	100%	100%	100%	99.1%	?Immunodeficiency 125, 620926
FLT4	100%	100%	100%	99%	Hemangioma, capillary infantile, somatic, 602089;Lymphatic malformation 1, 153100;Congenital heart defects, multiple types, 7, 618780
FLVCR1	100%	100%	100%	99.2%	Ataxia, posterior column, with retinitis pigmentosa, 609033

FLVCR2	100%	100%	100%	99.5%	Proliferative vasculopathy and hydranencephaly-hydrocephaly syndrome, 225790
FMN1	100%	100%	100%	98.5%	
FMN2	100%	100%	99.8%	95.2%	Intellectual developmental disorder, autosomal recessive 47, 616193
FMO3	100%	100%	100%	99.7%	Trimethylaminuria, 602079
FMR1	100%	99.5%	99.1%	75.8%	Fragile X tremor/ataxia syndrome, 300623;Fragile X syndrome, 300624;Premature ovarian failure 1, 311360
FN1	100%	100%	100%	99.5%	Spondylometaphyseal dysplasia, corner fracture type, 184255;Glomerulopathy with fibronectin deposits 2, 601894
FNIP1	100%	100%	100%	99.7%	Immunodeficiency 93 and hypertrophic cardiomyopathy, 619705
FOCAD	100%	100%	100%	99.4%	Liver disease, severe congenital, 619991
FOLR1	100%	100%	100%	98.9%	Neurodegeneration due to cerebral folate transport deficiency, 613068
FOSL2	100%	100%	100%	98.3%	Aplasia cutis-enamel dysplasia syndrome, 620789

FOXC1	100%	100%	99.7%	84.6%	Axenfeld-Rieger syndrome, type 3, 602482;Anterior segment dysgenesis 3, multiple subtypes, 601631
FOXC2	100%	100%	100%	94.5%	Lymphedema-distichiasis syndrome, 153400;Lymphedema-distichiasis syndrome with renal disease and diabetes mellitus, 153400
FOXE1	100%	100%	100%	89%	Bamforth-Lazarus syndrome, 241850;{Thyroid cancer, nonmedullary, 4}, 616534
FOXE3	100%	98.5%	98.7%	82.9%	Anterior segment dysgenesis 2, multiple subtypes, 610256;{Aortic aneurysm, familial thoracic 11, susceptibility to}, 617349;Cataract 34, multiple types, 612968
FOXF1	100%	100%	100%	94.7%	Alveolar capillary dysplasia with misalignment of pulmonary veins, 265380
FOXF2	99.7%	98.2%	100%	87%	
FOXG1	100%	99.9%	99.9%	94.7%	Rett syndrome, congenital variant, 613454
FOXH1	100%	100%	100%	99.1%	
FOXI1	100%	100%	100%	98.8%	Enlarged vestibular aqueduct, 600791

FOXI3	99.2%	95.9%	100%	94%	Craniofacial microsomia 2, 620444
FOXJ1	100%	100%	100%	98.2%	Ciliary dyskinesia, primary, 43, 618699
FOXL1	100%	100%	99.6%	92.6%	Otosclerosis 11, 620576
FOXL2	100%	100%	99.9%	91.8%	Blepharophimosis, epicanthus inversus, and ptosis, type 2, 110100;Blepharophimosis, epicanthus inversus, and ptosis, type 1, 110100;Premature ovarian failure 3, 608996
FOXN1	100%	100%	100%	99.3%	T-cell lymphopenia, infantile, with or without nail dystrophy, autosomal dominant, 618806;T-cell immunodeficiency, congenital alopecia, and nail dystrophy, 601705
FOXO1	100%	100%	100%	94.9%	Rhabdomyosarcoma, alveolar, 268220
FOXP1	100%	100%	100%	99.5%	Intellectual developmental disorder with language impairment with or without autistic features, 613670
FOXP2	100%	100%	100%	99%	Speech-language disorder-1, 602081
FOXP3	100%	99.6%	98.5%	67.1%	Immunodysregulation, polyendocrinopathy, and enteropathy, X-linked, 304790

FOXP4	100%	100%	100%	98.9%	
FOXRED1	100%	100%	100%	99.2%	Mitochondrial complex I deficiency, nuclear type 19, 618241
FPR1	100%	100%	100%	99.7%	
FRA10AC1	100%	100%	100%	99.9%	Neurodevelopmental disorder with growth retardation, dysmorphic facies, and corpus callosum abnormalities, 620113
FRAS1	100%	100%	100%	99.6%	Fraser syndrome 1, 219000
FREM1	100%	100%	100%	99.7%	Manitoba oculotrichoanal syndrome, 248450;Bifid nose with or without anorectal and renal anomalies, 608980;Trigonocephaly 2, 614485
FREM2	100%	100%	100%	99.7%	Fraser syndrome 2, 617666;Cryptophthalmos, unilateral or bilateral, isolated, 123570
FRMD4A	96.6%	96.6%	100%	98.7%	?Corpus callosum, agenesis of, with facial anomalies and cerebellar ataxia, 616819
FRMD5	100%	100%	100%	99.2%	Neurodevelopmental disorder with eye movement abnormalities and ataxia, 620094

FRMD7	100%	99.8%	98.6%	73.9%	Nystagmus, infantile periodic alternating, X-linked, 310700;Nystagmus 1, congenital, X-linked, 310700
FRMPD4	100%	99.6%	98.1%	69.2%	Intellectual developmental disorder, X-linked 104, 300983
FRRS1L	100%	100%	100%	96.4%	Developmental and epileptic encephalopathy 37, 616981
FRYL	100%	100%	100%	99.6%	
FSCN2	100%	100%	100%	98.4%	Retinitis pigmentosa 30, 607921
FSHB	100%	100%	100%	99.9%	Hypogonadotropic hypogonadism 24 without anosmia, 229070
FSHR	100%	100%	100%	99.7%	Ovarian hyperstimulation syndrome, 608115;Ovarian dysgenesis 1, 233300
FSIP2	100%	100%	100%	99.8%	Spermatogenic failure 34, 618153
FTCD	100%	100%	100%	97.2%	Glutamate formiminotransferase deficiency, 229100
FTH1	100%	100%	100%	98.5%	Neurodegeneration with brain iron accumulation 9, 620669;?Hemochromatosis, type 5, 615517

FTL	100%	99.9%	100%	99%	Hyperferritinemia-cataract syndrome, 600886;L-ferritin deficiency, dominant and recessive, 615604;Neurodegeneration with brain iron accumulation 3, 606159
FTO	94.5%	94.5%	100%	99.6%	Growth retardation, developmental delay, facial dysmorphism, 612938;{Obesity, susceptibility to, BMIQ14}, 612460
FTSJ1	100%	99.7%	98.8%	67.6%	Intellectual developmental disorder, X-linked 9, 309549
FUCA1	100%	100%	100%	99%	Fucosidosis, 230000
FURIN	100%	100%	100%	98.7%	
FUS	100%	100%	100%	98.8%	Amyotrophic lateral sclerosis 6, with or without frontotemporal dementia, 608030;Essential tremor, hereditary, 4, 614782
FUT2	100%	100%	100%	97.2%	{Norwalk virus infection, resistance to};{Vitamin B12 plasma level QTL1}, 612542;[Bombay phenotype, digenic], 616754
FUT6	100%	100%	100%	99%	[Fucosyltransferase 6 deficiency], 613852
FUT8	100%	100%	100%	99.8%	Congenital disorder of glycosylation with defective fucosylation 1, 618005

FUZ	100%	100%	100%	99.1%	{Neural tube defects, susceptibility to}, 182940
FXN	100%	100%	100%	95.1%	Friedreich ataxia with retained reflexes, 229300;Friedreich ataxia, 229300
FXR1	100%	100%	100%	97.1%	Congenital myopathy 9B, proximal, with minicore lesions, 618823;?Congenital myopathy 9A with respiratory insufficiency and bone fractures, 618822
FXYD2	100%	100%	100%	97.8%	Hypomagnesemia 2, renal, 154020
FYB1	100%	100%	100%	99.7%	Thrombocytopenia 3, 273900
FYCO1	100%	100%	100%	99.2%	Cataract 18, autosomal recessive, 610019
FZD2	100%	100%	99.6%	94%	Omodysplasia 2, 164745
FZD4	100%	100%	100%	99.1%	Retinopathy of prematurity, 133780;Exudative vitreoretinopathy 1, 133780
FZD5	100%	100%	100%	99.1%	Microphthalmia/coloboma 11, 620731
FZD6	100%	100%	100%	99.4%	Nail disorder, nonsyndromic congenital, 1, 161050
FZR1	100%	100%	100%	98.8%	Developmental and epileptic encephalopathy 109, 620145

G6PC1	100%	100%	100%	99.6%	Glycogen storage disease Ia, 232200
G6PC3	96.7%	96.7%	100%	99.7%	Dursun syndrome, 612541;Neutropenia, severe congenital 4, autosomal recessive, 612541
G6PD	86.3%	85.6%	98.6%	69%	Anemia, congenital, nonspherocytic hemolytic, 1, G6PD deficient, 300908;{Resistance to malaria due to G6PD deficiency}, 611162
GAA	100%	100%	100%	99.3%	Glycogen storage disease II, 232300
GAB1	100%	100%	100%	99.7%	?Deafness, autosomal recessive 26, 605428
GABBR1	100%	100%	99.9%	97.5%	Neurodevelopmental disorder with language delay and variable cognitive abnormalities, 620502
GABBR2	99.9%	99.6%	100%	98.2%	{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

GABRA1	100%	100%	100%	99.3%	{Epilepsy, juvenile myoclonic, susceptibility to, 5}, 611136;Developmental and epileptic encephalopathy 19, 615744;{Epilepsy, childhood absence, susceptibility to, 4}, 611136
GABRA2	100%	100%	100%	99.9%	Developmental and epileptic encephalopathy 78, 618557;{Alcohol dependence, susceptibility to}, 103780
GABRA3	100%	99.9%	98.5%	70.8%	Epilepsy, X-linked 2, with or without impaired intellectual development and dysmorphic features, 301091
GABRA4	100%	100%	100%	99.4%	
GABRA5	100%	100%	100%	99.3%	Developmental and epileptic encephalopathy 79, 618559
GABRB1	100%	100%	100%	99.6%	Developmental and epileptic encephalopathy 45, 617153
GABRB2	100%	100%	100%	99.6%	Developmental and epileptic encephalopathy 92, 617829
GABRB3	100%	100%	100%	98.9%	{Epilepsy, childhood absence, susceptibility to, 5}, 612269;Developmental and epileptic encephalopathy 43, 617113

GABRD	100%	100%	99.8%	97.1%	{?Generalized epilepsy with febrile seizures plus, type 5, susceptibility to}, 613060
GABRG2	92.9%	92.9%	100%	99.3%	Developmental and epileptic encephalopathy 74, 618396;Febrile seizures, familial, 8, 607681;Generalized epilepsy with febrile seizures plus, type 3, 607681
GAD1	100%	100%	100%	99.6%	Developmental and epileptic encephalopathy 89, 619124
GAL	100%	100%	100%	98.5%	?Epilepsy, familial temporal lobe, 8, 616461
GALC	100%	100%	100%	99.1%	Krabbe disease, 245200
GALE	100%	100%	100%	99.2%	Thrombocytopenia 13, syndromic, 620776;Galactose epimerase deficiency, 230350
GALK1	100%	100%	100%	99.4%	Galactokinase deficiency with cataracts, 230200
GALM	100%	100%	100%	99.3%	Galactosemia IV, 618881
GALNS	100%	100%	100%	98.3%	Mucopolysaccharidosis IVA, 253000
GALNT12	100%	99.6%	100%	99%	{Colorectal cancer, susceptibility to, 1}, 608812
GALNT2	100%	100%	100%	98.7%	Congenital disorder of glycosylation, type IIt, 618885

GALNT3	100%	100%	100%	99.9%	Tumoral calcinosis, hyperphosphatemic, familial, 1, 211900
GALNTL5	100%	100%	100%	99.8%	
GALT	100%	100%	100%	99%	Galactosemia, 230400
GAMT	100%	100%	100%	98.2%	Cerebral creatine deficiency syndrome 2, 612736
GAN	100%	100%	100%	99.7%	Giant axonal neuropathy-1, 256850
GANAB	100%	100%	100%	99.5%	Polycystic kidney disease 3, 600666
GAPVD1	100%	100%	100%	99.8%	
GARS1	98.9%	98.9%	100%	99.6%	Spinal muscular atrophy, infantile, James type, 619042;Neuronopathy, distal hereditary motor, autosomal dominant 5, 600794;Charcot-Marie-Tooth disease, type 2D, 601472
GAS2	100%	100%	100%	99.8%	?Deafness, autosomal recessive 125, 620877
GAS2L2	100%	100%	100%	99.2%	?Ciliary dyskinesia, primary, 41, 618449
GAS8	100%	100%	100%	99.2%	Ciliary dyskinesia, primary, 33, 616726

GATA1	100%	99.7%	98%	66.6%	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
GATA2	85.7%	85.7%	100%	99.3%	{Leukemia, acute myeloid, susceptibility to}, 601626;Emberger syndrome, 614038;Immunodeficiency 21, 614172;{Myelodysplastic syndrome, susceptibility to}, 614286
GATA3	100%	100%	100%	98.4%	Hypoparathyroidism, sensorineural deafness, and renal dysplasia, 146255

GATA4	100%	100%	100%	97.7%	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
GATA5	100%	99.9%	100%	98.4%	Congenital heart defects, multiple types, 5, 617912
GATA6	100%	100%	100%	95.2%	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
GATAD1	100%	100%	100%	99.1%	?Cardiomyopathy, dilated, 2B, 614672
GATAD2A	100%	100%	100%	99.1%	
GATAD2B	100%	100%	100%	99.2%	GAND syndrome, 615074
GATB	100%	100%	100%	99.5%	?Combined oxidative phosphorylation deficiency 41, 618838
GATC	100%	100%	100%	98.9%	Combined oxidative phosphorylation deficiency 42, 618839

GATM	100%	100%	100%	99.3%	Cerebral creatine deficiency syndrome 3, 612718;Fanconi renotubular syndrome 1, 134600
GBA1	100%	100%	100%	99.1%	{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
GBA2	100%	100%	100%	99.3%	Spastic paraplegia 46, autosomal recessive, 614409
GBE1	100%	100%	100%	99.8%	Glycogen storage disease IV, 232500;Polyglucosan body disease, adult form, 263570
GBF1	100%	100%	100%	99.2%	Charcot-Marie-Tooth disease, axonal, type 2GG, 606483
GCDH	100%	100%	100%	99.2%	Glutaricaciduria, type I, 231670
GCGR	100%	100%	100%	99.3%	Mahvash disease, 619290

GCH1	100%	100%	100%	99.4%	Dystonia, DOPA-responsive, 128230;Hyperphenylalaninemia, BH4-deficient, B, 233910
GCK	100%	100%	100%	99.4%	MODY, type II, 125851;Diabetes mellitus, permanent neonatal 1, 606176;Hyperinsulinemic hypoglycemia, familial, 3, 602485;Diabetes mellitus, noninsulin-dependent, late onset, 125853
GCLC	100%	100%	100%	99.3%	{Myocardial infarction, susceptibility to}, 608446;Anemia, congenital, nonspherocytic hemolytic, 7, 230450
GCLM	100%	100%	100%	99.2%	{Myocardial infarction, susceptibility to}, 608446
GCM2	100%	100%	100%	99.6%	Hypoparathyroidism, familial isolated 2, 618883;Hyperparathyroidism 4, 617343
GCNA	100%	99.9%	99.1%	71.6%	Spermatogenic failure, X-linked, 4, 301077
GCNT2	100%	100%	100%	99.8%	[Blood group, Ii], 110800;Adult i phenotype without cataract, 110800;Cataract 13 with adult i phenotype, 116700

GCSH	100%	100%	100%	99.7%	Multiple mitochondrial dysfunctions syndrome 7, 620423
GDAP1	86.5%	86.5%	98.7%	96.6%	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
GDAP2	100%	100%	100%	99.6%	Spinocerebellar ataxia, autosomal recessive 27, 618369
GDF1	100%	99.7%	100%	97.8%	Congenital heart defects, multiple types, 6, 613854;Right atrial isomerism (Ivemark), 208530
GDF11	100%	99.8%	100%	94.3%	?Vertebral hypersegmentation and orofacial anomalies, 619122
GDF2	100%	100%	100%	99.5%	Telangiectasia, hereditary hemorrhagic, type 5, 615506

GDF3	100%	100%	100%	99.6%	Klippel-Feil syndrome 3, autosomal dominant, 613702;Microphthalmia, isolated, with coloboma 6, 613703;Microphthalmia, isolated 7, 613704
GDF5	100%	100%	100%	99%	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
GDF6	100%	100%	100%	98.1%	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
GDF9	100%	100%	100%	99.6%	Premature ovarian failure 14, 618014

GDI1	100%	99.9%	98%	67.5%	Intellectual developmental disorder, X-linked 41, 300849
GDNF	100%	100%	100%	99%	{Hirschsprung disease, susceptibility to, 3}, 613711
GDPD1	100%	100%	100%	99.5%	
GEMIN4	100%	100%	100%	99.5%	Neurodevelopmental disorder with microcephaly, cataracts, and renal abnormalities, 617913
GEMIN5	100%	100%	100%	99.6%	Neurodevelopmental disorder with cerebellar atrophy and motor dysfunction, 619333
GET3	100%	100%	100%	98.2%	?Cardiomyopathy, dilated, 2H, 620203
GET4	100%	100%	100%	98.6%	?Congenital disorder of glycosylation, type IIy, 620200
GFAP	100%	100%	100%	99.6%	Alexander disease, 203450
GFER	100%	100%	100%	98.3%	Myopathy, mitochondrial progressive, with congenital cataract and developmental delay, 613076
GFI1	100%	100%	100%	99.2%	?Neutropenia, nonimmune chronic idiopathic, of adults, 607847;Neutropenia, severe congenital 2, autosomal dominant, 613107

GFI1B	100%	100%	100%	99%	Bleeding disorder, platelet-type, 17, 187900
GFM1	100%	100%	100%	99.5%	Combined oxidative phosphorylation deficiency 1, 609060
GFM2	100%	100%	100%	99.9%	Combined oxidative phosphorylation deficiency 39, 618397
GFPT1	100%	100%	100%	99.6%	Myasthenia, congenital, 12, with tubular aggregates, 610542
GFRA1	94%	93.9%	100%	99.3%	Renal hypodysplasia/aplasia 4, 619887
GFUS	100%	100%	100%	99%	
GGCX	100%	100%	100%	99.2%	Vitamin K-dependent clotting factors, combined deficiency of, 1, 277450;Pseudoxanthoma elasticum-like disorder with multiple coagulation factor deficiency, 610842
GGN	100%	100%	100%	98.9%	Spermatogenic failure 69, 619826
GGPS1	100%	100%	100%	99.7%	Muscular dystrophy, congenital hearing loss, and ovarian insufficiency syndrome, 619518
GGT1	100%	100%	100%	99.2%	?Glutathioninuria, 231950

GH1	100%	100%	100%	99%	Kowarski syndrome, 262650;Growth hormone deficiency, isolated, type II, 173100;Growth hormone deficiency, isolated, type IB, 612781;Growth hormone deficiency, isolated, type IA, 262400
GHR	99.8%	99.8%	99.8%	98.2%	Laron dwarfism, 262500;Increased responsiveness to growth hormone, 604271;Growth hormone insensitivity, partial, 604271;{Hypercholesterolemia, familial, modifier of}, 143890
GHRHR	100%	100%	100%	99.3%	Growth hormone deficiency, isolated, type IV, 618157
GHSR	100%	100%	100%	98.7%	Growth hormone deficiency, isolated partial, 615925
GIGYF1	100%	100%	100%	98.3%	
GIMAP5	100%	100%	100%	99.5%	Portal hypertension, noncirrhotic, 2, 619463
GINS1	81%	81%	100%	99.9%	Immunodeficiency 55, 617827
GINS2	100%	100%	100%	99.1%	
GINS4	100%	100%	100%	99.8%	
GIPC1	100%	100%	100%	99.1%	Oculopharyngodistal myopathy 2, 618940

GIPC3	100%	100%	100%	99.4%	Deafness, autosomal recessive 15, 601869
GJA1	100%	100%	100%	99.1%	Erythrokeratodermia 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
GJA3	100%	100%	100%	98.8%	Cataract 14, multiple types, 601885
GJA5	100%	100%	100%	99.6%	Atrial fibrillation, familial, 11, 614049;Atrial standstill, digenic (GJA5/SCN5A), 108770
GJA8	100%	100%	100%	99%	Cataract 1, multiple types, 116200
GJB1	100%	100%	95.7%	60.5%	Charcot-Marie-Tooth neuropathy, X-linked dominant, 1, 302800

GJB2	100%	100%	100%	99.9%	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
GJB3	100%	100%	100%	99.4%	Deafness, digenic, GJB2/GJB3, 220290;Erythrokeratodermia variabilis et progressiva 1, 133200;Deafness, autosomal dominant 2B, with or without peripheral neuropathy, 612644
GJB4	100%	100%	100%	99.2%	Erythrokeratodermia variabilis et progressiva 2, 617524
GJB6	100%	100%	100%	99.9%	Ectodermal dysplasia 2, Clouston type, 129500;Deafness, autosomal dominant 3B, 612643;Deafness, autosomal recessive 1B, 612645;Deafness, digenic GJB2/GJB6, 220290

GJC2	99.9%	97.5%	100%	97.2%	Lymphatic malformation 3, 613480;?Spastic paraplegia 44, autosomal recessive, 613206;Leukodystrophy, hypomyelinating, 2, 608804
GK	100%	99.5%	99.1%	75.4%	Glycerol kinase deficiency, 307030
GLA	91.4%	91.2%	99.2%	73.3%	Fabry disease, cardiac variant, 301500;Fabry disease, 301500
GLB1	100%	100%	100%	99.5%	GM1-gangliosidosis, type I, 230500;GM1-gangliosidosis, type III, 230650;Mucopolysaccharidosis type IVB (Morquio), 253010;GM1-gangliosidosis, type II, 230600
GLDC	100%	100%	100%	99.6%	Glycine encephalopathy1, 605899
GLDN	100%	100%	100%	99.3%	Lethal congenital contracture syndrome 11, 617194
GLE1	100%	100%	100%	99.6%	Lethal congenital contracture syndrome 1, 253310;Congenital arthrogryposis with anterior horn cell disease, 611890
GLI1	100%	100%	100%	98.9%	Polydactyly, preaxial I, 174400;Polydactyly, postaxial, type A8, 618123

GLI2	100%	100%	100%	98.7%	Culler-Jones syndrome, 615849;Holoprosencephaly 9, 610829
GLI3	99.3%	99.3%	100%	99.1%	Greig cephalopolysyndactyly syndrome, 175700;Polydactyly, postaxial, types A1 and B, 174200;Pallister-Hall syndrome, 146510;Polydactyly, preaxial, type IV, 174700
GLIS1	100%	100%	100%	98.9%	
GLIS2	100%	100%	100%	99%	Nephronophthisis 7, 611498
GLIS3	100%	100%	100%	99.1%	Diabetes mellitus, neonatal, with congenital hypothyroidism, 610199
GLMN	100%	100%	100%	99.8%	Glomuvenous malformations, 138000
GLRA1	100%	100%	100%	99.4%	Hyperekplexia 1, 149400
GLRA2	100%	99.4%	98.9%	71.1%	Intellectual developmental disorder, X-linked syndromic, Pilorge type, 301076
GLRB	100%	100%	100%	99.7%	Hyperekplexia 2, 614619
GLRX5	100%	100%	100%	99.8%	Anemia, sideroblastic, 3, pyridoxine-refractory, 616860;Spasticity, childhood-onset, with hyperglycinemia, 616859

GLS	100%	100%	100%	99.5%	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
GLUD1	100%	100%	100%	99.2%	Hyperinsulinism-hyperammonemia syndrome, 606762
GLUL	100%	100%	100%	99.5%	Glutamine deficiency, congenital, 610015;Developmental and epileptic encephalopathy 116, 620806
GLYCTK	100%	100%	100%	99.2%	D-glyceric aciduria, 220120
GLYR1	100%	100%	100%	99.3%	
GM2A	100%	100%	100%	99%	GM2-gangliosidosis, AB variant, 272750
GMNN	100%	100%	100%	99.9%	Meier-Gorlin syndrome 6, 616835
GMPPA	100%	100%	100%	98.2%	Alacrima, achalasia, and impaired intellectual development syndrome, 615510

GMPPB	100%	100%	100%	99.4%	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
GMPR	100%	100%	100%	99.3%	
GMPS	100%	100%	100%	99.6%	
GNA11	100%	100%	100%	97.9%	Hypocalciuric hypercalcemia, type II, 145981;Hypocalcemia, autosomal dominant 2, 615361
GNA14	100%	100%	100%	99.6%	
GNAI1	100%	100%	100%	99.8%	Neurodevelopmental disorder with hypotonia, impaired speech, and behavioral abnormalities, 619854
GNAI2	100%	100%	100%	98.3%	Ventricular tachycardia, idiopathic, 192605;Pituitary adenoma, ACTH-secreting, somatic

GNAI3	100%	100%	100%	99.4%	Auriculocondylar syndrome 1, 602483
GNAL	100%	100%	100%	99.4%	Dystonia 25, 615073
GNAO1	100%	100%	100%	99.4%	Developmental and epileptic encephalopathy 17, 615473;Neurodevelopmental disorder with involuntary movements, 617493
GNAQ	100%	100%	100%	98.5%	Capillary malformations, congenital, 1, somatic, mosaic, 163000;Sturge-Weber syndrome, somatic, mosaic, 185300
GNAS	99.9%	98.3%	99.9%	95.6%	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
GNAS-AS1					Pseudohypoparathyroidism Ib, 603233

GNAT1	100%	100%	100%	99.2%	Night blindness, congenital stationary, autosomal dominant 3, 610444;Night blindness, congenital stationary, type 1G, 616389
GNAT2	100%	100%	100%	99.7%	Achromatopsia 4, 613856
GNB1	100%	100%	100%	99.2%	Myelodysplastic syndrome, somatic, 614286;Leukemia, acute lymphoblastic, somatic, 613065;Intellectual developmental disorder, autosomal dominant 42, 616973
GNB2	100%	100%	100%	98.7%	Neurodevelopmental disorder with hypotonia and dysmorphic facies, 619503;?Sick sinus syndrome 4, 619464
GNB3	100%	100%	100%	98%	Night blindness, congenital stationary, type 1H, 617024;{Hypertension, essential, susceptibility to}, 145500
GNB4	100%	100%	100%	99.8%	Charcot-Marie-Tooth disease, dominant intermediate F, 615185

GNB5	100%	100%	100%	99.6%	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
GNE	100%	100%	100%	99.8%	Sialuria, 269921;Thrombocytopenia 12 with or without myopathy, 620757;Nonaka myopathy, 605820
GNMT	100%	100%	100%	97.5%	Glycine N-methyltransferase deficiency, 606664
GNPAT	100%	100%	100%	99.7%	Rhizomelic chondrodysplasia punctata, type 2, 222765
GNPNAT1	100%	100%	100%	99.6%	?Rhizomelic dysplasia, Ain-Naz type, 619598
GNPTAB	100%	100%	100%	99.5%	Mucolipidosis III alpha/beta, 252600;Mucolipidosis II alpha/beta, 252500
GNPTG	100%	100%	100%	99.2%	Mucolipidosis III gamma, 252605
GNRH1	100%	100%	100%	100%	?Hypogonadotropic hypogonadism 12 with or without anosmia, 614841

GNRHR	100%	100%	100%	99.7%	Hypogonadotropic hypogonadism 7 without anosmia, 146110
GNS	100%	100%	100%	99.6%	Mucopolysaccharidosis type IIID, 252940
GOLGA2	100%	100%	100%	99.1%	Developmental delay with hypotonia, myopathy, and brain abnormalities, 620240
GON7	100%	100%	100%	99.2%	Galloway-Mowat syndrome 9, 619603
GORAB	100%	100%	100%	99%	Geroderma osteodysplasticum, 231070
GOSR2	100%	100%	100%	99.2%	Epilepsy, progressive myoclonic 6, 614018;Muscular dystrophy, congenital, with or without seizures, 620166
GOT1	100%	100%	100%	99.6%	Aspartate aminotransferase, serum level of, QTL1, 614419
GOT2	100%	100%	100%	99.5%	Developmental and epileptic encephalopathy 82, 618721
GP1BA	100%	100%	99.9%	96.9%	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

GP1BB	100%	100%	100%	96.1%	Giant platelet disorder, isolated, 231200;Bernard-Soulier syndrome, type B, 231200
GP6	99.7%	97.3%	100%	99.2%	Bleeding disorder, platelet-type, 11, 614201
GP9	100%	100%	100%	99.3%	Bernard-Soulier syndrome, type C, 231200
GPAA1	88.5%	88.5%	100%	99.3%	Glycosylphosphatidylinositol biosynthesis defect 15, 617810
GPAT2	99.5%	99.5%	99.6%	97.6%	
GPATCH11	100%	100%	100%	99.6%	
GPC3	100%	99.5%	98.9%	71.1%	Wilms tumor, somatic, 194070;Simpson-Golabi-Behmel syndrome, type 1, 312870
GPC4	100%	99.8%	98.9%	72.4%	Keipert syndrome, 301026
GPC6	100%	100%	100%	99.4%	Omodysplasia 1, 258315
GPD1	100%	100%	100%	99.3%	Hypertriglyceridemia, transient infantile, 614480
GPD1L	100%	100%	100%	99.7%	Brugada syndrome 2, 611777
GPHN	100%	100%	100%	99.7%	Molybdenum cofactor deficiency C, 615501
GPI	100%	100%	100%	98.4%	Anemia, congenital, nonspherocytic hemolytic, 4, glucose phosphate isomerase deficient, 613470

GPIHBP1	100%	100%	100%	99.3%	Hyperlipoproteinemia, type 1D, 615947
GPNMB	95.1%	95.1%	100%	99.4%	Amyloidosis, primary localized cutaneous, 3, 617920
GPR101	100%	99.7%	96.5%	62.2%	Pituitary adenoma 2, GH-secreting, 300943
GPR143	100%	99.8%	98.9%	72.4%	Ocular albinism, type I, Nettleship-Falls type, 300500;Nystagmus 6, congenital, X-linked, 300814
GPR156	100%	100%	100%	99.3%	Deafness, autosomal recessive 121, 620551
GPR161	100%	100%	100%	99.3%	{Medulloblastoma predisposition syndrome}, 155255
GPR179	100%	100%	100%	99.4%	Night blindness, congenital stationary (complete), 1E, autosomal recessive, 614565
GPR68	100%	100%	100%	98.4%	Amelogenesis imperfecta, hypomaturation type, IIA6, 617217
GPR88	100%	99.7%	100%	97.4%	?Chorea, childhood-onset, with psychomotor retardation, 616939
GPRASP2	100%	100%	99%	70.8%	?Deafness, X-linked 7, 301018

GPRC5B	100%	100%	100%	99.4%	Megalencephalic leukoencephalopathy with subcortical cysts 3, 620447
GPSM2	95.5%	95.5%	100%	99.8%	Chudley-McCullough syndrome, 604213
GPT2	100%	100%	100%	98.2%	Neurodevelopmental disorder with microcephaly and spastic paraplegia, 616281
GPX1	100%	100%	100%	99.4%	Hemolytic anemia due to glutathione peroxidase deficiency, 614164
GPX4	100%	99.8%	100%	99.3%	Spondylometaphyseal dysplasia, Sedaghatian type, 250220
GRAP	100%	100%	99.9%	98.9%	Deafness, autosomal recessive 114, 618456
GREB1L	100%	100%	100%	99.7%	Deafness, autosomal dominant 80, 619274;Renal hypodysplasia/aplasia 3, 617805
GREM1	100%	100%	100%	99.3%	
GREM2	100%	100%	100%	98.5%	Tooth agenesis, selective, 9, 617275
GRHL2	100%	100%	100%	99.5%	Deafness, autosomal dominant 28, 608641;Ectodermal dysplasia/short stature syndrome, 616029;Corneal dystrophy, posterior polymorphous, 4, 618031

GRHL3	100%	99.9%	100%	99.3%	van der Woude syndrome 2, 606713
GRHPR	100%	100%	100%	99.2%	Hyperoxaluria, primary, type II, 260000
GRIA1	100%	100%	100%	99.6%	?Intellectual developmental disorder, autosomal recessive 76, 619931;Intellectual developmental disorder, autosomal dominant 67, 619927
GRIA2	100%	100%	100%	99.6%	Neurodevelopmental disorder with language impairment and behavioral abnormalities, 618917
GRIA3	100%	99.6%	98.5%	73%	Intellectual developmental disorder, X-linked syndromic, Wu type, 300699
GRIA4	100%	100%	100%	99%	Neurodevelopmental disorder with or without seizures and gait abnormalities, 617864
GRID2	100%	100%	100%	99.5%	Spinocerebellar ataxia, autosomal recessive 18, 616204

GRIK2	95.3%	95.3%	99.8%	97.9%	Neurodevelopmental disorder with impaired language and ataxia and with or without seizures, 619580;Intellectual developmental disorder, autosomal recessive 6, 611092
GRIN1	100%	100%	100%	98.7%	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
GRIN2A	100%	100%	100%	99%	Epilepsy, focal, with speech disorder and with or without impaired intellectual development, 245570
GRIN2B	100%	100%	100%	99.4%	Developmental and epileptic encephalopathy 27, 616139;Intellectual developmental disorder, autosomal dominant 6, with or without seizures, 613970
GRIN2D	99.7%	97.6%	99.8%	92.5%	Developmental and epileptic encephalopathy 46, 617162

GRIP1	100%	100%	100%	99.4%	Fraser syndrome 3, 617667
GRK1	100%	100%	100%	98.9%	Oguchi disease-2, 613411
GRM1	100%	100%	100%	99.4%	Spinocerebellar ataxia, autosomal recessive 13, 614831;Spinocerebellar ataxia 44, 617691
GRM6	100%	100%	100%	98.7%	Night blindness, congenital stationary (complete), 1B, autosomal recessive, 257270
GRM7	100%	100%	99.8%	97%	Neurodevelopmental disorder with seizures, hypotonia, and brain abnormalities, 618922
GRN	100%	100%	100%	99.8%	Frontotemporal dementia 2, 607485;Aphasia, primary progressive, 607485;Ceroid lipofuscinosis, neuronal, 11, 614706
GRXCR1	100%	100%	100%	99.6%	Deafness, autosomal recessive 25, 613285
GRXCR2	100%	100%	100%	99.4%	?Deafness, autosomal recessive 101, 615837
GSC	100%	100%	100%	97.3%	Short stature, auditory canal atresia, mandibular hypoplasia, skeletal abnormalities, 602471
GSDME	100%	100%	100%	99.7%	Deafness, autosomal dominant 5, 600994
GSE1	100%	100%	100%	98.1%	

GSN	100%	100%	100%	97.6%	Amyloidosis, Finnish type, 105120
GSR	100%	100%	100%	99.3%	Anemia, congenital, nonspherocytic hemolytic, 10, glutathione reductase deficient, 618660
GSS	100%	100%	100%	99.6%	Anemia, congenital, nonspherocytic hemolytic, 6, glutatione synthetase deficient, 266130;Glutathione synthetase deficiency, 266130
GSX2	100%	100%	100%	98.4%	Diencephalic-mesencephalic junction dysplasia syndrome 2, 618646
GTF2E2	100%	100%	100%	99.8%	Trichothiodystrophy 6, nonphotosensitive, 616943
GTF2H5	59.2%	59.2%	100%	99.6%	Trichothiodystrophy 3, photosensitive, 616395
GTF3C3	100%	100%	100%	99.8%	
GTF3C5	100%	99.9%	100%	99.2%	
GTPBP1	100%	100%	100%	97.6%	Neurodevelopmental disorder with characteristic facial and ectodermal features and tetraparesis 1, 620888
GTPBP2	100%	100%	100%	99.1%	Jaberi-Elahi syndrome, 617988

GTPBP3	100%	100%	100%	99%	Combined oxidative phosphorylation deficiency 23, 616198
GUCA1A	100%	100%	100%	100%	Cone-rod dystrophy 14, 602093;Cone dystrophy-3, 602093
GUCA1B	100%	100%	100%	98.6%	Retinitis pigmentosa 48, 613827
GUCY1A1	100%	100%	100%	99.4%	Moyamoya 6 with achalasia, 615750
GUCY2C	100%	100%	100%	99.6%	Diarrhea 6, 614616;Meconium ileus, 614665
GUCY2D	100%	100%	100%	99.2%	Cone-rod dystrophy 6, 601777;?Choroidal dystrophy, central areolar 1, 215500;Leber congenital amaurosis 1, 204000;Night blindness, congenital stationary, type 1I, 618555
GUF1	100%	100%	100%	99.6%	?Developmental and epileptic encephalopathy 40, 617065
GUK1	100%	100%	100%	99.2%	
GULOP					Scurvy
GUSB	100%	100%	100%	98.7%	Mucopolysaccharidosis VII, 253220
GYG1	100%	100%	100%	99.7%	?Glycogen storage disease XV, 613507;Polyglucosan body myopathy 2, 616199

GYS1	100%	100%	100%	99%	Glycogen storage disease 0, muscle, 611556
GYS2	100%	100%	100%	99.6%	Glycogen storage disease 0, liver, 240600
GZF1	100%	100%	100%	99.4%	Joint laxity, short stature, and myopia, 617662
H1-4	100%	100%	100%	99.2%	Rahman syndrome, 617537
H19					
H3-3A	100%	100%	100%	99.4%	Bryant-Li-Bhoj neurodevelopmental syndrome 1, 619720
H3-3B	100%	100%	100%	98.7%	Bryant-Li-Bhoj neurodevelopmental syndrome 2, 619721
H4C11	100%	100%	100%	99.7%	?Tessadori-Bicknell-van Haaften neurodevelopmental syndrome 2, 619759
H4C3	100%	100%	100%	97.7%	Tessadori-Bicknell-van Haaften neurodevelopmental syndrome 1, 619758
H4C5	100%	100%	100%	99.8%	Tessadori-Bicknell-van Haaften neurodevelopmental syndrome 3, 619950
H4C9	100%	100%	99.9%	98%	Tessadori-Bicknell-van Haaften neurodevelopmental syndrome 4, 619951

H6PD	100%	100%	100%	99.3%	Cortisone reductase deficiency 1, 604931
HAAO	100%	100%	100%	99.6%	Vertebral, cardiac, renal, and limb defects syndrome 1, 617660
HABP2	100%	100%	100%	99.7%	{?Thyroid cancer, nonmedullary, 5}, 616535;{Venous thromboembolism, susceptibility to}, 188050
HACD1	80.3%	80.3%	100%	98.9%	Congenital myopathy 11, 619967
HACE1	100%	100%	100%	99.3%	Spastic paraplegia and psychomotor retardation with or without seizures, 616756
HADH	100%	100%	100%	99.7%	Hyperinsulinemic hypoglycemia, familial, 4, 609975;3-hydroxyacyl-CoA dehydrogenase deficiency, 231530
HADHA	100%	100%	100%	99.5%	HELLP syndrome, maternal, of pregnancy, 609016;LCHAD deficiency, 609016;Mitochondrial trifunctional protein deficiency 1, 609015;Fatty liver, acute, of pregnancy, 609016
HADHB	100%	100%	100%	99.7%	Mitochondrial trifunctional protein deficiency 2, 620300

HAGH	100%	99.8%	100%	97.4%	[Glyoxalase II deficiency], 614033
HAMP	100%	100%	100%	99.2%	Hemochromatosis, type 2B, 613313
HAND1	100%	100%	100%	97.5%	
HAND2	100%	100%	99.9%	89%	
HARS1	100%	100%	100%	99.2%	Charcot-Marie-Tooth disease, axonal, type 2W, 616625;Usher syndrome type 3B, 614504
HARS2	100%	100%	100%	99.2%	Perrault syndrome 2, 614926
HAVCR2	100%	100%	100%	99.6%	T-cell lymphoma, subcutaneous panniculitis-like, 618398
HAX1	100%	100%	100%	99.4%	Neutropenia, severe congenital 3, autosomal recessive, 610738
HBA1	100%	100%	100%	99.4%	Hemoglobin H disease, nondeletional, 613978;Thalassemias, alpha-, 604131;Heinz body anemias, alpha-, 140700;Methemoglobinemia, alpha type, 617973;Erythrocytosis, familial, 7, 617981

HBA2	100%	100%	98.7%	82.9%	Heinz body anemia, 140700;Thalassemia, alpha-, 604131;Erythrocytosis, familial, 7, 617981;Hemoglobin H disease, deletional and nondeletional, 613978
HBB	100%	100%	100%	99.4%	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
HBD	100%	100%	100%	99.3%	Thalassemia due to Hb Lepore;Thalassemia, delta-
HBG1	97.7%	96.9%	96%	73.8%	Fetal hemoglobin quantitative trait locus 1, 141749
HBG2	100%	100%	100%	99.3%	Fetal hemoglobin quantitative trait locus 1, 141749;Cyanosis, transient neonatal, 613977

HCCS	100%	99.8%	99.5%	75%	Linear skin defects with multiple congenital anomalies 1, 309801
HCFC1	100%	99.5%	98.4%	68.2%	Methylmalonic aciduria and homocysteinemia, cblX type, 309541
HCK	100%	100%	100%	99.1%	Autoinflammation with pulmonary and cutaneous vasculitis, 620296
HCN1	100%	100%	100%	97.9%	Developmental and epileptic encephalopathy 24, 615871;Generalized epilepsy with febrile seizures plus, type 10, 618482
HCN2	92.9%	88.3%	96.5%	86.4%	Febrile seizures, familial, 2, 602477;{Epilepsy, idiopathic generalized, susceptibility to, 17}, 602477;Generalized epilepsy with febrile seizures plus, type 11, 602477
HCN3	100%	100%	100%	98.8%	
HCN4	100%	100%	100%	98.3%	Sick sinus syndrome 2, 163800;{Epilepsy, idiopathic generalized, susceptibility to, 18}, 619521;Brugada syndrome 8, 613123
HCRT	100%	100%	100%	97.1%	?Narcolepsy 1, 161400
HDAC3	100%	100%	100%	99.2%	

HDAC4	100%	100%	100%	98.9%	Neurodevelopmental disorder with central hypotonia and dysmorphic facies, 619797
HDAC6	100%	99.7%	98.7%	70.4%	?Chondrodysplasia with platyspondyly, distinctive brachydactyly, hydrocephaly, and microphthalmia, 300863
HDAC8	97.6%	96.8%	98.6%	71.6%	Cornelia de Lange syndrome 5, 300882
HDAC9	100%	100%	100%	99.6%	?Auriculocondylar syndrome 4, 620457
HEATR3	100%	100%	100%	99.3%	Diamond-Blackfan anemia 21, 620072
HEATR5B	100%	100%	100%	99.6%	
HECTD1	100%	100%	100%	99.6%	
HECTD4	100%	100%	100%	99.4%	Neurodevelopmental disorder with seizures, spasticity, and complete or partial agenesis of the corpus callosum, 620250
HECW2	100%	100%	100%	99.4%	Neurodevelopmental disorder with hypotonia, seizures, and absent language, 617268
HELLS	100%	100%	100%	99.9%	Immunodeficiency-centromeric instability-facial anomalies syndrome 4, 616911
HENMT1	92.1%	92.1%	100%	99.9%	

HEPACAM	100%	100%	100%	98.8%	Megalencephalic leukoencephalopathy with subcortical cysts 2A, 613925;Megalencephalic leukoencephalopathy with subcortical cysts 2B, remitting, with or without impaired intellectual development, 613926
HEPHL1	100%	100%	100%	99.6%	?Abnormal hair, joint laxity, and developmental delay, 261990
HERC1	100%	100%	100%	99.7%	Macrocephaly, dysmorphic facies, and psychomotor retardation, 617011
HERC2	100%	99.7%	100%	99.3%	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
HES7	100%	100%	100%	97.2%	Spondylocostal dysostosis 4, autosomal recessive, 613686
HESX1	100%	100%	100%	99.5%	Pituitary hormone deficiency, combined, 5, 182230;Septooptic dysplasia, 182230;Growth hormone deficiency with pituitary anomalies, 182230

HEXA	100%	100%	100%	99.7%	[Hex A pseudodeficiency], 272800;GM2-gangliosidosis, several forms, 272800;Tay-Sachs disease, 272800
HEXB	100%	100%	100%	98.8%	Sandhoff disease, infantile, juvenile, and adult forms, 268800
HEY2	100%	100%	100%	96.9%	
HFE	100%	100%	100%	98.7%	Hemochromatosis, type 1, 235200
HFM1	100%	100%	100%	99.7%	Premature ovarian failure 9, 615724
HGD	100%	100%	100%	99.8%	Alkaptonuria, 203500
HGF	100%	100%	100%	99.6%	Deafness, autosomal recessive 39, 608265
HGSNAT	92.4%	92.4%	100%	99.3%	Mucopolysaccharidosis type IIIC (Sanfilippo C), 252930;Retinitis pigmentosa 73, 616544
HHAT	100%	100%	100%	99.3%	Nivelon-Nivelon-Mabille syndrome, 600092
HIBADH	100%	100%	100%	99.4%	
HIBCH	100%	100%	100%	99.9%	3-hydroxyisobutryl-CoA hydrolase deficiency, 250620
HID1	100%	100%	100%	99%	Developmental and epileptic encephalopathy 105 with hypopituitarism, 619983

HIKESHI	100%	100%	100%	99.1%	Leukodystrophy, hypomyelinating, 13, 616881
HINT1	100%	100%	100%	98.9%	Neuromyotonia and axonal neuropathy, autosomal recessive, 137200
HIVEP2	100%	100%	100%	99.6%	Intellectual developmental disorder, autosomal dominant 43, 616977
HJV	100%	100%	100%	99.5%	Hemochromatosis, type 2A, 602390
HK1	100%	100%	100%	99.4%	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
HKDC1	100%	100%	100%	99.5%	Retinitis pigmentosa 92, 619614
HLCS	100%	100%	100%	98.6%	Holocarboxylase synthetase deficiency, 253270
HLX	100%	100%	100%	99.2%	

HMBS	100%	100%	100%	99.5%	Leukoencephalopathy, porphyria-related, 620711;Encephalopathy, porphyria-related, 620704;Porphyria, acute intermittent, nonerythroid variant, 176000;Porphyria, acute intermittent, 176000
HMCN1	100%	100%	100%	99.8%	{Macular degeneration, age-related, 1}, 603075
HMGA2	94.4%	83.5%	100%	99.3%	Silver-Russell syndrome 5, 618908
HMGB1	100%	100%	100%	99.9%	
HMGB3	100%	99.4%	99.8%	74.1%	?Microphthalmia, syndromic 13, 300915
HMGCL	100%	100%	100%	99.5%	HMG-CoA lyase deficiency, 246450
HMGCR	100%	100%	100%	99.8%	Muscular dystrophy, limb-girdle, autosomal recessive 28, 620375;[Statins, response to], 620410;[Low density lipoprotein cholesterol level QTL 3], 620410
HMGCS2	100%	100%	100%	99.6%	HMG-CoA synthase-2 deficiency, 605911
HMOX1	100%	100%	100%	99.4%	Heme oxygenase-1 deficiency, 614034;{Pulmonary disease, chronic obstructive, susceptibility to}, 606963

HMX1	100%	99.9%	100%	97.3%	Oculoauricular syndrome, 612109
HNF1A	100%	100%	100%	99.2%	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
HNF1B	100%	100%	100%	99.4%	Type 2 diabetes mellitus, 125853;Renal cysts and diabetes syndrome, 137920;{Renal cell carcinoma}, 144700
HNF4A	100%	100%	100%	99.1%	Fanconi renotubular syndrome 4, with maturity-onset diabetes of the young, 616026;{Diabetes mellitus, noninsulin-dependent}, 125853;MODY, type I, 125850
HNMT	100%	100%	100%	99.9%	Intellectual developmental disorder, autosomal recessive 51, 616739;{Asthma, susceptibility to}, 600807

HNRNPA1	100%	100%	100%	99.6%	?Inclusion body myopathy with early-onset Paget disease without frontotemporal dementia 3, 615424;?Myopathy, distal, 3, 610099;Amyotrophic lateral sclerosis 20, 615426
HNRNPA2B1	100%	100%	100%	99.6%	Oculopharyngeal muscular dystrophy 2, 620460;?Inclusion body myopathy with early-onset Paget disease with or without frontotemporal dementia 2, 615422
HNRNPC	100%	100%	100%	99.8%	Intellectual developmental disorder, autosomal dominant 74, 620688
HNRNPD	100%	100%	100%	99%	
HNRNPDL	100%	100%	99.8%	92.5%	Muscular dystrophy, limb-girdle, autosomal dominant 3, 609115
HNRNPH1	100%	100%	100%	99.8%	Neurodevelopmental disorder with craniofacial dysmorphism and skeletal defects, 620083
HNRNPH2	100%	100%	99.1%	72.7%	Intellectual developmental disorder, X-linked syndromic, Bain type, 300986
HNRNPK	100%	100%	100%	99.8%	Au-Kline syndrome, 616580

HNRNPR	100%	100%	100%	99.8%	Neurodevelopmental disorder with dysmorphic facies and skeletal and brain abnormalities, 620073
HNRNPU	100%	100%	100%	97.3%	Developmental and epileptic encephalopathy 54, 617391
HOGA1	100%	100%	100%	99%	Hyperoxaluria, primary, type III, 613616
HOMER2	100%	100%	100%	99.1%	?Deafness, autosomal dominant 68, 616707
HOXA1	100%	100%	100%	99.2%	Bosley-Salih-Alorainy syndrome, 601536;Athabaskan brainstem dysgenesis syndrome, 601536
HOXA11	100%	100%	100%	97.5%	Radioulnar synostosis with amegakaryocytic thrombocytopenia 1, 605432
HOXA13	99%	95.2%	95.3%	70.6%	Hand-foot-genital syndrome, 140000;?Guttmacher syndrome, 176305
HOXA2	100%	100%	100%	99%	Microtia with or without hearing impairment (AD), 612290;?Microtia, hearing impairment, and cleft palate (AR), 612290
HOXB1	100%	100%	100%	99.3%	Facial paresis, hereditary congenital, 3, 614744

HOXB13	100%	100%	100%	98.6%	{Prostate cancer, hereditary, 9}, 610997
HOXC13	100%	100%	100%	98.1%	Ectodermal dysplasia 9, hair/nail type, 614931
HOXD10	100%	100%	100%	98.2%	Vertical talus, congenital, 192950;Charcot-Marie-Tooth disease, foot deformity of, 192950
HOXD13	100%	100%	100%	96.1%	Syndactyly, type V, 186300;Synpolydactyly 1, 186000;Brachydactyly, type E, 113300;Brachydactyly, type D, 113200;?Brachydactyly-syndactyly syndrome, 610713
HPCA	100%	100%	100%	98.3%	Dystonia 2, torsion, autosomal recessive, 224500
HPD	100%	100%	100%	97.9%	Hawkinsinuria, 140350;Tyrosinemia, type III, 276710
HPDL	100%	100%	100%	98.9%	Neurodevelopmental disorder with progressive spasticity and brain white matter abnormalities, 619026;Spastic paraplegia 83, autosomal recessive, 619027

HPGD	100%	100%	100%	99.1%	?Digital clubbing, isolated congenital, 119900;Hypertrophic osteoarthropathy, primary, autosomal recessive 1, 259100;Cranioosteoarthropathy, 259100
HPRT1	100%	99.8%	99.1%	73.6%	Hyperuricemia, HRPT-related, 300323;Lesch-Nyhan syndrome, 300322
HPS1	100%	100%	100%	99%	Hermansky-Pudlak syndrome 1, 203300
HPS3	100%	100%	100%	99.6%	Hermansky-Pudlak syndrome 3, 614072
HPS4	100%	100%	100%	99.7%	Hermansky-Pudlak syndrome 4, 614073
HPS5	100%	100%	100%	99.7%	Hermansky-Pudlak syndrome 5, 614074
HPS6	100%	100%	100%	98.4%	Hermansky-Pudlak syndrome 6, 614075
HPSE2	100%	100%	100%	99.3%	Urofacial syndrome 1, 236730
HR	100%	100%	100%	99.3%	Atrichia with papular lesions, 209500;Alopecia universalis, 203655

HRAS	100%	100%	100%	98.6%	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
HRG	100%	100%	100%	99.6%	Thrombophilia 11 due to HRG deficiency, 613116
HROB	100%	100%	100%	99%	Ovarian dysgenesis 11, 620897
HRURF	100%	100%	100%	99.4%	Hypotrichosis 4, 146550
HS2ST1	100%	100%	100%	99.6%	Neurofacioskeletal syndrome with or without renal agenesis, 619194
HS3ST6	100%	98.6%	99.9%	92.9%	?Angioedema, hereditary, 8, 619367
HS6ST1	100%	100%	100%	97.2%	{Hypogonadotropic hypogonadism 15 with or without anosmia}, 614880
HS6ST2	100%	99.9%	98.3%	69.3%	?Paganini-Miozzo syndrome, 301025

HSCB	100%	100%	100%	99.9%	?Anemia, sideroblastic, 5, 619523
HSD11B1	100%	100%	100%	99.3%	Cortisone reductase deficiency 2, 614662
HSD11B2	100%	100%	100%	95.8%	Apparent mineralocorticoid excess, 218030
HSD17B10	100%	99.7%	99.4%	73.5%	HSD10 mitochondrial disease, 300438
HSD17B3	100%	100%	100%	99.4%	Pseudohermaphroditism, male, with gynecomastia, 264300
HSD17B4	100%	100%	100%	99.6%	D-bifunctional protein deficiency, 261515;Perrault syndrome 1, 233400
HSD3B2	100%	100%	100%	99.2%	Adrenal hyperplasia, congenital, due to 3-beta-hydroxysteroid dehydrogenase 2 deficiency, 201810
HSD3B7	100%	100%	100%	99.6%	Bile acid synthesis defect, congenital, 1, 607765
HSF2	100%	100%	100%	99.6%	
HSF2BP	100%	100%	100%	99.5%	Premature ovarian failure 19, 619245
HSF4	100%	100%	100%	98.7%	Cataract 5, multiple types, 116800
HSFY1	50%	50%	49.6%	28.6%	
HSFY2	50%	50%	49.5%	29.9%	

HSPA9	100%	100%	100%	99.6%	Even-plus syndrome, 616854;Anemia, sideroblastic, 4, 182170
HSPB1	100%	100%	100%	97.8%	Charcot-Marie-Tooth disease, axonal, type 2F, 606595;Neuronopathy, distal hereditary motor, autosomal dominant 3, 608634
HSPB3	100%	100%	100%	100%	?Neuronopathy, distal hereditary motor, autosomal dominant 4, 613376
HSPB6	100%	100%	99.7%	95%	
HSPB8	100%	100%	100%	98.6%	Neuronopathy, distal hereditary motor, autosomal dominant 2, 158590;Charcot-Marie-Tooth disease, axonal, type 2L, 608673
HSPD1	99.6%	97.6%	100%	99.8%	Spastic paraplegia 13, autosomal dominant, 605280;Leukodystrophy, hypomyelinating, 4, 612233
HSPG2	100%	100%	100%	99.1%	Dyssegmental dysplasia, Silverman-Handmaker type, 224410;Schwartz-Jampel syndrome, type 1, 255800
HTR1A	100%	100%	100%	98.4%	?Periodic fever, menstrual cycle dependent, 614674

HTRA1	100%	99.8%	100%	99.1%	{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
HTRA2	100%	100%	100%	99.1%	{Parkinson disease 13}, 610297;3-methylglutaconic aciduria, type VIII, 617248
HTT	99.9%	99.9%	100%	99.4%	Lopes-Maciel-Rodan syndrome, 617435;Huntington disease, 143100
HUWE1	100%	99.4%	98.7%	70.9%	Intellectual developmental disorder, X-linked syndromic, Turner type, 309590
HYAL1	100%	100%	100%	98.2%	Mucopolysaccharidosis type IX, 601492
HYAL2	100%	100%	100%	99.5%	
HYCC1	100%	100%	100%	99.5%	Leukodystrophy, hypomyelinating, 5, 610532
HYDIN	99.9%	99.8%	100%	99.4%	Ciliary dyskinesia, primary, 5, 608647
HYLS1	100%	100%	100%	99.6%	Hydrolethalus syndrome, 236680

HYOU1	100%	100%	100%	99.4%	?Immunodeficiency 59 and hypoglycemia, 233600
IARS1	100%	100%	100%	99.6%	Growth retardation, impaired intellectual development, hypotonia, and hepatopathy, 617093
IARS2	100%	100%	100%	99.5%	Cataracts, growth hormone deficiency, sensory neuropathy, sensorineural hearing loss, and skeletal dysplasia, 616007
IBA57	100%	100%	100%	99.1%	Multiple mitochondrial dysfunctions syndrome 3, 615330;?Spastic paraplegia 74, autosomal recessive, 616451
ICOS	100%	100%	100%	99.6%	Immunodeficiency, common variable, 1, 607594
ICOSLG	100%	100%	100%	99.1%	?Immunodeficiency 119, 620825
ID4	100%	99.9%	99.9%	92.4%	
IDH1	100%	100%	100%	99.9%	{Glioma, susceptibility to, somatic}, 137800
IDH2	100%	100%	100%	98.6%	D-2-hydroxyglutaric aciduria 2, 613657
IDH3A	100%	100%	100%	99.4%	Retinitis pigmentosa 90, 619007
IDH3B	100%	100%	100%	99.5%	Retinitis pigmentosa 46, 612572
IDI1	100%	100%	100%	100%	

IDS	100%	99.3%	99.2%	72.1%	Mucopolysaccharidosis II, 309900
IDUA	100%	100%	100%	97.7%	Mucopolysaccharidosis Is, 607016;Mucopolysaccharidosis Ih/s, 607015;Mucopolysaccharidosis Ih, 607014
IER3IP1	100%	100%	100%	99.7%	Microcephaly, epilepsy, and diabetes syndrome, 614231
IFIH1	100%	100%	100%	99.8%	Immunodeficiency 95, 619773;Aicardi-Goutieres syndrome 7, 615846;Singleton-Merten syndrome 1, 182250
IFITM5	100%	100%	100%	99.4%	Osteogenesis imperfecta, type V, 610967
IFNAR1	94.5%	94.5%	100%	99.6%	Immunodeficiency 106, susceptibility to viral infections, 619935
IFNAR2	100%	100%	100%	99.7%	{Hepatitis B virus, susceptibility to}, 610424;Immunodeficiency 45, 616669

IFNG	100%	100%	100%	99.9%	{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
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
IFNGR2	100%	100%	100%	99.3%	Immunodeficiency 28, mycobacteriosis, 614889
IFNLR1	100%	100%	100%	98.5%	
IFRD1	100%	100%	100%	99.9%	
IFT122	100%	100%	100%	99.3%	Cranioectodermal dysplasia 1, 218330

IFT140	100%	100%	100%	99%	Short-rib thoracic dysplasia 9 with or without polydactyly, 266920;Retinitis pigmentosa 80, 617781
IFT172	100%	100%	100%	99.3%	Retinitis pigmentosa 71, 616394;Bardet-Biedl syndrome 20, 619471;Short-rib thoracic dysplasia 10 with or without polydactyly, 615630
IFT27	100%	100%	100%	99.2%	Bardet-Biedl syndrome 19, 615996
IFT43	100%	100%	100%	99.4%	?Cranioectodermal dysplasia 3, 614099;?Retinitis pigmentosa 81, 617871;Short-rib thoracic dysplasia 18 with polydactyly, 617866
IFT52	100%	100%	100%	99.6%	Short-rib thoracic dysplasia 16 with or without polydactyly, 617102
IFT56	100%	100%	100%	99.7%	Biliary, renal, neurologic, and skeletal syndrome, 619534
IFT57	100%	100%	100%	99.7%	?Orofaciodigital syndrome XVIII, 617927
IFT74	100%	100%	100%	99.7%	Bardet-Biedl syndrome 22, 617119;Spermatogenic failure 58, 619585;Joubert syndrome 40, 619582

IFT80	100%	100%	100%	99.4%	Short-rib thoracic dysplasia 2 with or without polydactyly, 611263
IFT81	94.9%	94.9%	100%	99.9%	Short-rib thoracic dysplasia 19 with or without polydactyly, 617895
IFT88	100%	100%	100%	99.7%	
IGBP1	100%	99.2%	99.3%	72.4%	?Corpus callosum, agenesis of, with impaired intellectual development, ocular coloboma and micrognathia, 300472
IGF1	100%	100%	100%	99.6%	Insulin-like growth factor I deficiency, 608747
IGF1R	100%	100%	100%	99.3%	Insulin-like growth factor I, resistance to, 270450
IGF2	100%	100%	100%	99.1%	Silver-Russell syndrome 3, 616489
IGF2R	100%	100%	100%	99.4%	Hepatocellular carcinoma, somatic, 114550
IGFALS	100%	100%	100%	99.1%	Acid-labile subunit, deficiency of, 615961
IGFBP7	100%	100%	100%	98.5%	Retinal arterial macroaneurysm with supravalvular pulmonic stenosis, 614224
IGHG2	100%	99.9%	99.7%	92.2%	IgG2 deficiency, selective
IGHM	100%	100%	100%	99%	Agammaglobulinemia 1, 601495

IGHMBP2	100%	100%	100%	99.2%	Charcot-Marie-Tooth disease, axonal, type 2S, 616155;Neuronopathy, distal hereditary motor, autosomal recessive 1, 604320
IGKC	100%	100%	100%	99.3%	Kappa light chain deficiency, 614102
IGLL1	100%	100%	100%	99.1%	Agammaglobulinemia 2, 613500
IGSF1	100%	99.6%	99.2%	72.2%	Hypothyroidism, central, and testicular enlargement, 300888
IGSF10	100%	100%	100%	99.7%	
IGSF3	100%	100%	100%	99.5%	?Lacrimal duct defect, 149700
IHH	100%	100%	100%	98.7%	Acrocapitofemoral dysplasia, 607778;Brachydactyly, type A1, 112500
IKBKB	100%	100%	100%	99.3%	Immunodeficiency 15B, 615592;Immunodeficiency 15A, 618204
IKBKG	96.4%	94.1%	99.3%	75%	Incontinentia pigmenti, 308300;Ectodermal dysplasia and immunodeficiency 1, 300291;Immunodeficiency 33, 300636;Autoinflammatory disease, systemic, X-linked, 301081

IKZF1	100%	100%	100%	98.6%	Immunodeficiency, common variable, 13, 616873
IKZF2	100%	100%	100%	99.6%	
IKZF3	100%	100%	100%	99.4%	?Immunodeficiency 84, 619437
IKZF5	100%	100%	100%	99.6%	Thrombocytopenia, autosomal dominant, 7, 619130
IL10	100%	100%	100%	100%	{Rheumatoid arthritis, progression of}, 180300;{Graft-versus-host disease, protection against}, 614395;{HIV-1, susceptibility to}, 609423
IL10RA	100%	100%	100%	99.3%	Inflammatory bowel disease 28, early onset, autosomal recessive, 613148
IL10RB	100%	100%	100%	99.4%	{Hepatitis B virus, susceptibility to}, 610424;Inflammatory bowel disease 25, early onset, autosomal recessive, 612567
IL11RA	100%	100%	100%	98%	Craniosynostosis and dental anomalies, 614188
IL12B	100%	100%	100%	99.9%	Immunodeficiency 29, mycobacteriosis, 614890
IL12RB1	94.1%	94.1%	100%	98.9%	Immunodeficiency 30, 614891

IL17F	100%	100%	100%	99.3%	?Candidiasis, familial, 6, autosomal dominant, 613956
IL17RA	100%	100%	100%	99.4%	Immunodeficiency 51, 613953
IL17RC	100%	100%	100%	98.9%	Candidiasis, familial, 9, 616445
IL17RD	100%	100%	100%	99.3%	Hypogonadotropic hypogonadism 18 with or without anosmia, 615267
IL18BP	100%	100%	100%	99.4%	{?Hepatitis, fulminant viral, susceptibility to}, 618549
IL1R1	97.7%	97.7%	100%	99.6%	?Chronic recurrent multifocal osteomyelitis 3, 259680
IL1RAPL1	100%	99.9%	98.9%	70.5%	Intellectual developmental disorder, X-linked 21, 300143
IL1RN	100%	100%	100%	99.8%	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
IL2	100%	100%	100%	99.9%	

IL21	100%	100%	100%	100%	?Immunodeficiency, common variable, 11, 615767
IL21R	100%	100%	100%	99.3%	Immunodeficiency 56, 615207
IL2RA	100%	100%	100%	98.5%	Immunodeficiency 41 with lymphoproliferation and autoimmunity, 606367;{Diabetes, mellitus, insulin-dependent, susceptibility to, 10}, 601942
IL2RB	96.1%	96.1%	100%	98.5%	Immunodeficiency 63 with lymphoproliferation and autoimmunity, 618495
IL2RG	100%	99.8%	98.2%	68.6%	Combined immunodeficiency, X-linked, moderate, 312863;Severe combined immunodeficiency, X-linked, 300400
IL31RA	100%	100%	100%	99.2%	?Amyloidosis, primary localized cutaneous, 2, 613955
IL36RN	100%	100%	100%	99.5%	Psoriasis 14, pustular, 614204
IL37	100%	100%	100%	99.5%	?Inflammatory bowel disease (infantile ulcerative colitis) 31, 619398

IL6R	92.5%	92.5%	100%	99.2%	[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
IL6ST	100%	100%	100%	99.6%	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
IL7	100%	100%	100%	99.8%	{?Epidermodysplasia verruciformis, susceptibility to, 5}, 618309
IL7R	100%	100%	100%	99.6%	Immunodeficiency 104, severe combined, 608971
ILDR1	100%	100%	100%	99.3%	Deafness, autosomal recessive 42, 609646
ILK	100%	100%	100%	99.1%	
IMPA1	100%	100%	100%	99.7%	Intellectual developmental disorder, autosomal recessive 59, 617323

IMPDH1	100%	100%	100%	99.2%	Retinitis pigmentosa 10, 180105;Leber congenital amaurosis 11, 613837
IMPG1	100%	100%	100%	99.3%	Macular dystrophy, vitelliform, 4, 616151;Retinitis pigmentosa 91, 153870
IMPG2	100%	100%	100%	99.8%	Retinitis pigmentosa 56, 613581;Macular dystrophy, vitelliform, 5, 616152
INF2	100%	100%	99.9%	94.6%	Glomerulosclerosis, focal segmental, 5, 613237;Charcot-Marie-Tooth disease, dominant intermediate E, 614455
ING1	100%	100%	99.9%	96.4%	Squamous cell carcinoma, head and neck, somatic, 275355
INO80	100%	100%	100%	99.4%	
INPP4A	100%	100%	100%	99.4%	
INPP5E	100%	100%	100%	96.7%	Impaired intellectual development, truncal obesity, retinal dystrophy, and micropenis syndrome, 610156;Joubert syndrome 1, 213300
INPP5K	100%	100%	100%	99%	Muscular dystrophy, congenital, with cataracts and intellectual disability, 617404
INPPL1	100%	100%	100%	98.8%	Opsismodysplasia, 258480

INS	100%	100%	100%	98%	Diabetes mellitus, insulin-dependent, 2, 125852;Maturity-onset diabetes of the young, type 10, 613370;Hyperproinsulinemia, 616214;Diabetes mellitus, permanent neonatal 4, 618858
INSC	94.4%	94.4%	100%	99.1%	
INSL3	78.8%	78.8%	100%	99.1%	Cryptorchidism, 219050
INSR	100%	100%	100%	98.9%	Rabson-Mendenhall syndrome, 262190;Diabetes mellitus, insulin-resistant, with acanthosis nigricans, 610549;Donohue syndrome, 246200;Hyperinsulinemic hypoglycemia, familial, 5, 609968
INTS1	100%	100%	100%	98.9%	Neurodevelopmental disorder with cataracts, poor growth, and dysmorphic facies, 618571
INTS11	100%	100%	100%	98.4%	Neurodevelopmental disorder with motor and language delay, ocular defects, and brain abnormalities, 620428
INTS13	100%	100%	100%	99.8%	

INTS8	100%	100%	100%	99.5%	?Neurodevelopmental disorder with cerebellar hypoplasia and spasticity, 618572
INTU	100%	100%	100%	99.7%	?Orofaciodigital syndrome XVII, 617926;?Short-rib thoracic dysplasia 20 with polydactyly, 617925
INVS	100%	100%	100%	99.6%	Nephronophthisis 2, infantile, 602088
IPMK	100%	100%	100%	99.7%	
IPO8	100%	100%	100%	99.8%	VISS syndrome, 619472
IQCB1	100%	100%	100%	99.8%	Senior-Loken syndrome 5, 609254
IQCE	100%	100%	100%	98.1%	Polydactyly, postaxial, type A7, 617642
IQCN	100%	100%	100%	99.1%	Spermatogenic failure 78, 620170
IQGAP1	100%	100%	99.9%	98.8%	
IQSEC1	100%	99.7%	100%	97.1%	Intellectual developmental disorder with short stature and behavioral abnormalities, 618687
IQSEC2	99.2%	96%	97.1%	63.9%	Intellectual developmental disorder, X-linked 1, 309530
IQUB	100%	100%	100%	99.8%	
IRAK1	100%	99.5%	98.2%	64.9%	
IRAK4	100%	100%	100%	99.9%	Immunodeficiency 67, 607676

IREB2	100%	100%	100%	99.8%	Neurodegeneration, early-onset, with choreoathetoid movements and microcytic anemia, 618451
IRF1	100%	100%	100%	99.3%	Nonsmall cell lung cancer, somatic, 211980;Gastric cancer, somatic, 613659;Immunodeficiency 117, mycobacteriosis, autosomal recessive, 620668
IRF2BP2	100%	100%	100%	94.9%	?Immunodeficiency, common variable, 14, 617765
IRF2BPL	100%	100%	98.2%	89.6%	Neurodevelopmental disorder with regression, abnormal movements, loss of speech, and seizures, 618088
IRF3	100%	100%	100%	99.1%	{Encephalopathy, acute, infection-induced (herpes-specific), susceptibility to, 7}, 616532
IRF4	100%	100%	100%	98.9%	[Skin/hair/eye pigmentation, variation in, 8], 611724
IRF6	100%	100%	100%	99%	{Orofacial cleft 6}, 608864;Popliteal pterygium syndrome 1, 119500;van der Woude syndrome 1, 119300
IRF7	100%	100%	100%	98.9%	?Immunodeficiency 39, 616345

IRF8	100%	100%	100%	99.1%	Immunodeficiency 32A, mycobacteriosis, autosomal dominant, 614893;Immunodeficiency 32B, monocyte and dendritic cell deficiency, autosomal recessive, 226990
IRF9	100%	100%	100%	99%	Immunodeficiency 65, susceptibility to viral infections, 618648
IRGM	100%	100%	100%	99.8%	{Mycobacterium tuberculosis, protection against}, 607948;{Inflammatory bowel disease (Crohn disease) 19}, 612278
IRS4	100%	99.9%	97.1%	62.3%	Hypothyroidism, congenital, nongoitrous, 9, 301035
IRX1	100%	97.7%	100%	94.7%	
IRX5	100%	100%	100%	95.3%	Hamamy syndrome, 611174
ISCA1	92.4%	92.4%	100%	99.7%	Multiple mitochondrial dysfunctions syndrome 5, 617613
ISCA2	100%	100%	100%	99.5%	Multiple mitochondrial dysfunctions syndrome 4, 616370
ISCU	100%	100%	100%	99.7%	Myopathy with lactic acidosis, hereditary, 255125
ISG15	100%	100%	100%	99.5%	Immunodeficiency 38, 616126

ITCH	92.5%	92.4%	100%	99.8%	Autoimmune disease, multisystem, with facial dysmorphism, 613385
ITGA2	100%	100%	100%	99.8%	
ITGA2B	100%	100%	100%	99.1%	Thrombocytopenia, neonatal alloimmune, BAK antigen related;Glanzmann thrombasthenia 1, 273800;Bleeding disorder, platelet-type, 16, autosomal dominant, 187800
ITGA3	100%	100%	100%	98.9%	Epidermolysis bullosa, junctional 7, with interstitial lung disease and nephrotic syndrome, 614748
ITGA6	100%	100%	100%	99.7%	Epidermolysis bullosa, junctional 6, with pyloric atresia, 619817
ITGA7	100%	100%	100%	99.3%	Muscular dystrophy, congenital, due to ITGA7 deficiency, 613204
ITGA8	100%	100%	100%	99.2%	Renal hypodysplasia/aplasia 1, 191830
ITGB2	100%	100%	100%	99.2%	Leukocyte adhesion deficiency, 116920

ITGB3	100%	100%	100%	99.3%	Bleeding disorder, platelet-type, 24, autosomal dominant, 619271;Thrombocytopenia, neonatal alloimmune;Purpura, posttransfusion;{Myocardial infarction, susceptibility to}, 608446;Glanzmann thrombasthenia 2, 619267
ITGB4	100%	100%	100%	98.9%	Epidermolysis bullosa, junctional 5B, with pyloric atresia, 226730;Epidermolysis bullosa, junctional 5A, intermediate, 619816
ITGB6	100%	100%	100%	99.8%	Amelogenesis imperfecta, type IH, 616221
ITK	100%	100%	100%	99.5%	Lymphoproliferative syndrome 1, 613011
ITM2B	100%	100%	100%	99.1%	?Retinal dystrophy with inner retinal dysfunction and ganglion cell abnormalities, 616079;Dementia, familial British, 176500;Dementia, familial Danish, 117300
ITPA	100%	100%	100%	99.5%	[Inosine triphosphatase deficiency], 613850;Developmental and epileptic encephalopathy 35, 616647
ITPKB	100%	100%	100%	98.6%	

ITPR1	100%	100%	100%	99.5%	Gillespie syndrome, 206700;Spinocerebellar ataxia 29, congenital nonprogressive, 117360;Spinocerebellar ataxia 15, 606658
ITPR2	100%	100%	100%	99.8%	?Anhidrosis, isolated, with normal sweat glands, 106190
ITPR3	100%	100%	100%	98.8%	Charcot-Marie-Tooth disease, demyelinating, type 1J, 620111;{Diabetes, type 1, susceptibility to}, 222100
ITSN1	100%	100%	100%	99.5%	
ITSN2	100%	100%	100%	99.6%	
IVD	100%	100%	100%	99.4%	Isovaleric acidemia, 243500
IVNS1ABP	100%	100%	100%	99.8%	Immunodeficiency 70, 618969
IYD	100%	100%	100%	99.6%	Thyroid dyshormonogenesis 4, 274800
JAG1	100%	100%	100%	99.4%	?Deafness, congenital heart defects, and posterior embryotoxon, 617992;Charcot-Marie-Tooth disease, axonal, type 2HH, 619574;Alagille syndrome 1, 118450;Tetralogy of Fallot, 187500

JAG2	99.9%	99.4%	100%	98%	Muscular dystrophy, limb-girdle, autosomal recessive 27, 619566
JAGN1	100%	100%	100%	99.7%	Neutropenia, severe congenital, 6, autosomal recessive, 616022
JAK1	100%	100%	100%	99.5%	Autoinflammation, immune dysregulation, and eosinophilia, 618999
JAK2	100%	100%	100%	99.5%	{Budd-Chiari syndrome, somatic}, 600880;Myelofibrosis, somatic, 254450;Erythrocytosis, somatic, 133100;Leukemia, acute myeloid, somatic, 601626;Thrombocythemia 3, 614521;Polycythemia vera, somatic, 263300
JAK3	100%	100%	100%	97.9%	Severe combined immunodeficiency, autosomal recessive, T-negative/B-positive type, 600802
JAM2	92.5%	92%	100%	99.6%	Basal ganglia calcification, idiopathic, 8, autosomal recessive, 618824
JAM3	100%	100%	100%	99.8%	Hemorrhagic destruction of the brain, subependymal calcification, and cataracts, 613730

JARID2	100%	100%	100%	99.2%	Developmental delay with variable intellectual disability and dysmorphic facies, 620098
JMJD1C	100%	100%	100%	99.6%	
JPH1	100%	100%	100%	99%	Congenital myopathy 25, 620964;{?Charcot-Marie-Tooth disease, axonal, autosomal dominant, type 2K, modifier of}, 607831
JPH2	100%	100%	100%	97.8%	Cardiomyopathy, dilated, 2E, 619492;Cardiomyopathy, hypertrophic, 17, 613873
JPH3	100%	100%	100%	97.5%	Huntington disease-like 2, 606438
JUP	100%	100%	100%	98.4%	Naxos disease, 601214;?Arrhythmogenic right ventricular dysplasia 12, 611528
KALRN	98.4%	98.3%	100%	99.2%	
KANK1	98.1%	98.1%	100%	99.5%	Cerebral palsy, spastic quadriplegic, 2, 612900
KANK2	100%	100%	100%	99.7%	Nephrotic syndrome, type 16, 617783;Palmoplantar keratoderma and woolly hair, 616099
KANSL1	100%	100%	100%	99.7%	Koolen-De Vries syndrome, 610443

KARS1	100%	100%	100%	99.8%	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
KASH5	100%	100%	100%	98.8%	Spermatogenic failure 88, 620547;Premature ovarian failure 22, 620548
KAT5	100%	100%	100%	98.2%	Neurodevelopmental disorder with dysmorphic facies, sleep disturbance, and brain abnormalities, 619103
KAT6A	100%	100%	100%	99.3%	Arboleda-Tham syndrome, 616268
KAT6B	100%	100%	100%	99.4%	SBBYSS syndrome, 603736;Genitopatellar syndrome, 606170
KAT8	100%	100%	100%	99.3%	Li-Ghorgani-Weisz-Hubshman syndrome, 618974
KATNB1	100%	100%	100%	98.8%	Lissencephaly 6, with microcephaly, 616212

KATNIP	100%	100%	100%	99.2%	Joubert syndrome 26, 616784
KBTBD13	100%	100%	100%	96.8%	Nemaline myopathy 6, autosomal dominant, 609273
KBTBD2	100%	100%	100%	99.9%	
KCNA1	100%	100%	100%	99.1%	Episodic ataxia/myokymia syndrome, 160120
KCNA2	100%	100%	100%	99.3%	Developmental and epileptic encephalopathy 32, 616366
KCNA3	100%	100%	100%	98.8%	
KCNA4	100%	100%	100%	98.2%	Microcephaly, cataracts, impaired intellectual development, and dystonia with abnormal striatum, 618284
KCNA5	100%	100%	100%	98.8%	Atrial fibrillation, familial, 7, 612240
KCNB1	100%	100%	100%	98.7%	Developmental and epileptic encephalopathy 26, 616056
KCNB2	100%	100%	100%	98.3%	
KCNC1	100%	100%	100%	98%	Epilepsy, progressive myoclonic 7, 616187
KCNC2	100%	100%	100%	99.2%	Developmental and epileptic encephalopathy 103, 619913
KCNC3	99.6%	97.6%	98.9%	87.9%	Spinocerebellar ataxia 13, 605259
KCND1	100%	99.6%	98%	67.3%	

KCND2	100%	100%	100%	98.5%	
KCND3	100%	100%	100%	97.8%	Spinocerebellar ataxia 19, 607346;Brugada syndrome 9, 616399
KCNE1	100%	100%	100%	99.8%	Jervell and Lange-Nielsen syndrome 2, 612347;Long QT syndrome 5, 613695
KCNE2	100%	100%	100%	99.5%	Long QT syndrome 6, 613693;Atrial fibrillation, familial, 4, 611493
KCNE3	100%	100%	100%	99.8%	?Brugada syndrome 6, 613119
KCNE4	100%	100%	100%	98.4%	
KCNE5	100%	99.5%	97.5%	64.3%	
KCNH1	98.6%	98.6%	100%	99.5%	Zimmermann-Laband syndrome 1, 135500;Temple-Baraitser syndrome, 611816
KCNH2	100%	100%	100%	98.1%	Short QT syndrome 1, 609620;Long QT syndrome 2, 613688
KCNH5	100%	100%	100%	99.7%	Developmental and epileptic encephalopathy 112, 620537
KCNJ1	100%	100%	100%	99.9%	Bartter syndrome, type 2, 241200
KCNJ10	100%	100%	100%	99.3%	Enlarged vestibular aqueduct, digenic, 600791;SESAME syndrome, 612780

KCNJ11	100%	100%	100%	99.2%	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
KCNJ13	100%	100%	100%	99.6%	Snowflake vitreoretinal degeneration, 193230;Leber congenital amaurosis 16, 614186
KCNJ16	100%	100%	100%	99.8%	Hypokalemic tubulopathy and deafness, 619406
KCNJ2	100%	100%	100%	99.2%	Atrial fibrillation, familial, 9, 613980;Andersen syndrome, 170390;Short QT syndrome 3, 609622
KCNJ5	100%	100%	100%	98.6%	Long QT syndrome 13, 613485;Hyperaldosteronism, familial, type III, 613677
KCNJ6	100%	100%	100%	99.6%	Keppen-Lubinsky syndrome, 614098
KCNJ8	100%	100%	100%	99.7%	
KCNK3	100%	99.9%	100%	98.9%	Pulmonary hypertension, primary, 4, 615344

KCNK4	99.4%	99.3%	100%	98.6%	Facial dysmorphism, hypertrichosis, epilepsy, intellectual/developmental delay, and gingival overgrowth syndrome, 618381
KCNK9	100%	100%	100%	98.6%	Birk-Barel syndrome, 612292
KCNMA1	100%	100%	100%	99%	{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
KCNN2	100%	100%	100%	99%	?Dystonia 34, myoclonic, 619724;Neurodevelopmental disorder with or without variable movement or behavioral abnormalities, 619725
KCNN3	100%	100%	100%	98.5%	Zimmermann-Laband syndrome 3, 618658
KCNN4	100%	100%	100%	98.2%	Dehydrated hereditary stomatocytosis 2, 616689

KCNQ1	100%	99.7%	100%	98.1%	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
KCNQ1OT1					Beckwith-Wiedemann syndrome, 130650
KCNQ2	100%	100%	100%	98.9%	Developmental and epileptic encephalopathy 7, 613720;Seizures, benign neonatal, 1, 121200;Myokymia, 121200
KCNQ3	100%	100%	100%	98.7%	Seizures, benign neonatal, 2, 121201
KCNQ4	99.8%	99.3%	100%	98.6%	Deafness, autosomal dominant 2A, 600101
KCNQ5	100%	100%	100%	99.1%	Intellectual developmental disorder, autosomal dominant 46, 617601
KCNT1	100%	99.9%	100%	98.2%	Developmental and epileptic encephalopathy 14, 614959;Epilepsy nocturnal frontal lobe, 5, 615005
KCNT2	100%	100%	100%	99.8%	Developmental and epileptic encephalopathy 57, 617771
KCNU1	100%	100%	100%	99.6%	Spermatogenic failure 79, 620196

KCNV2	100%	100%	100%	99.6%	Retinal cone dystrophy 3B, 610356
KCTD1	100%	100%	99.9%	95.5%	Scalp-ear-nipple syndrome, 181270
KCTD17	100%	100%	100%	97.2%	Dystonia 26, myoclonic, 616398
KCTD19	100%	100%	100%	99%	
KCTD3	100%	100%	100%	99.3%	
KCTD7	100%	100%	100%	99.1%	Epilepsy, progressive myoclonic 3, with or without intracellular inclusions, 611726
KDELR2	100%	100%	100%	99%	Osteogenesis imperfecta, type XXI, 619131
KDF1	100%	100%	100%	99.2%	?Ectodermal dysplasia 12, hypohidrotic/hair/tooth/nail type, 617337
KDM1A	96.9%	96.9%	100%	99.3%	Cleft palate, psychomotor retardation, and distinctive facial features, 616728;{ACTH-independent macronodular adrenal hyperplasia 3}, 620990
KDM2B	100%	100%	100%	98.9%	
KDM3B	100%	100%	100%	99.4%	Diets-Jongmans syndrome, 618846
KDM4B	100%	100%	100%	98.7%	Intellectual developmental disorder, autosomal dominant 65, 619320

KDM5A	100%	100%	100%	99.7%	El Hayek-Chahrour neurodevelopmental syndrome, 620820
KDM5B	97.6%	96.5%	100%	99.4%	Intellectual developmental disorder, autosomal recessive 65, 618109
KDM5C	98%	97.4%	98.5%	68%	Intellectual developmental disorder, X-linked syndromic, Claes-Jensen type, 300534
KDM5D	48.9%	48.7%	48.8%	20.3%	
KDM6A	100%	99.8%	99.2%	74.3%	Kabuki syndrome 2, 300867
KDM6B	100%	100%	100%	97.9%	Stolerman neurodevelopmental syndrome, 618505
KDR	100%	100%	100%	99.8%	{Hemangioma, capillary infantile, susceptibility to}, 602089;Hemangioma, capillary infantile, somatic, 602089
KDSR	100%	100%	100%	98.9%	Erythrokeratodermia variabilis et progressiva 4, 617526
KERA	100%	100%	100%	99.5%	Cornea plana 2, autosomal recessive, 217300
KHDC3L	100%	100%	100%	98.6%	Hydatidiform mole, recurrent, 2, 614293
KHK	100%	100%	100%	99%	?[Fructosuria, essential], 229800

KIAA0586	95.6%	95.6%	100%	99.8%	Short-rib thoracic dysplasia 14 with polydactyly, 616546;Joubert syndrome 23, 616490
KIAA0753	100%	100%	100%	99.6%	?Orofaciodigital syndrome XV, 617127;?Joubert syndrome 38, 619476;Short-rib thoracic dysplasia 21 without polydactyly, 619479
KIAA0825	100%	100%	100%	99.8%	Polydactyly, postaxial, type A10, 618498
KIAA1549	99.8%	99.5%	100%	98.5%	Retinitis pigmentosa 86, 618613
KICS2	100%	100%	100%	99.4%	
KIDINS220	100%	100%	100%	99.6%	Spastic paraplegia, intellectual disability, nystagmus, and obesity, 617296;Ventriculomegaly and arthrogryposis, 619501
KIF11	100%	100%	100%	99.6%	Microcephaly with or without chorioretinopathy, lymphedema, or impaired intellectual development, 152950
KIF12	100%	100%	100%	98.9%	Cholestasis, progressive familial intrahepatic, 8, 619662
KIF14	100%	100%	100%	99.8%	Microcephaly 20, primary, autosomal recessive, 617914;?Meckel syndrome 12, 616258

KIF15	100%	100%	100%	99.6%	?Braddock-Carey syndrome 2, 619981
KIF1A	100%	100%	100%	98.9%	NESCAV syndrome, 614255;Neuropathy, hereditary sensory, type IIC, 614213;Spastic paraplegia 30, autosomal dominant, 610357;Spastic paraplegia 30, autosomal recessive, 620607
KIF1B	94.9%	94.9%	100%	99.6%	{Neuroblastoma, susceptibility to, 1}, 256700;Charcot-Marie-Tooth disease, type 2A1, 118210
KIF1C	100%	100%	100%	98.8%	Spastic ataxia 2, autosomal recessive, 611302
KIF20A	100%	100%	100%	99.6%	?Cardiomyopathy, familial restrictive, 6, 619433
KIF21A	100%	100%	100%	99.7%	Fibrosis of extraocular muscles, congenital, 3B, 135700;Fibrosis of extraocular muscles, congenital, 1, 135700
KIF21B	100%	100%	100%	99.2%	
KIF22	100%	100%	100%	98.9%	Spondyloepimetaphyseal dysplasia with joint laxity, type 2, 603546
KIF23	100%	100%	100%	99.7%	Anemia, congenital dyserythropoietic, type IIIA, 105600

KIF24	100%	100%	100%	99.6%	
KIF26A	97.6%	97.6%	100%	99.1%	Cortical dysplasia, complex, with other brain malformations 11, 620156
KIF2A	100%	100%	100%	99.7%	Cortical dysplasia, complex, with other brain malformations 3, 615411
KIF3B	100%	100%	100%	99.2%	Retinitis pigmentosa 89, 618955
KIF4A	100%	99.8%	99.1%	74.4%	Taurodontism, microdontia, and dens invaginatus, 313490;Intellectual developmental disorder, X-linked 100, 300923
KIF5A	100%	100%	100%	99.1%	Myoclonus, intractable, neonatal, 617235;{Amyotrophic lateral sclerosis, susceptibility to, 25}, 617921;Spastic paraplegia 10, autosomal dominant, 604187
KIF5B	100%	100%	100%	99.8%	
KIF5C	100%	100%	100%	99.5%	Cortical dysplasia, complex, with other brain malformations 2, 615282
KIF7	100%	99.8%	100%	98.4%	Joubert syndrome 12, 200990;Acrocallosal syndrome, 200990;?Hydrolethalus syndrome 2, 614120;?Al-Gazali-Bakalinova syndrome, 607131

KIFBP	95.9%	95.9%	100%	99.8%	Goldberg-Shprintzen megacolon syndrome, 609460
KIRREL1	100%	100%	100%	98.9%	Nephrotic syndrome, type 23, 619201
KIRREL3	100%	100%	100%	97.9%	
KISS1	100%	100%	100%	98.5%	?Hypogonadotropic hypogonadism 13 with or without anosmia, 614842
KISS1R	100%	100%	100%	99.5%	Hypogonadotropic hypogonadism 8 with or without anosmia, 614837;?Precocious puberty, central, 1, 176400
KIT	100%	100%	100%	99.8%	Gastrointestinal stromal tumor, familial, 606764;Mastocytosis, cutaneous, 154800;Piebaldism, 172800;Germ cell tumors, somatic, 273300;Mastocytosis, systemic, somatic, 154800;Leukemia, acute myeloid, somatic, 601626

KITLG	100%	100%	100%	99.9%	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
KIZ	100%	100%	100%	99.9%	Retinitis pigmentosa 69, 615780
KL	99.7%	99.1%	99.6%	97.4%	?Tumoral calcinosis, hyperphosphatemic, familial, 3, 617994
KLB	100%	100%	100%	99.7%	
KLC2	100%	100%	100%	99.2%	Spastic paraplegia, optic atrophy, and neuropathy, 609541
KLF1	100%	100%	100%	98.6%	Blood group--Lutheran inhibitor, 111150;[Hereditary persistence of fetal hemoglobin], 613566;Anemia, dyserythropoietic congenital, type IVa, 613673;Anemia, congenital dyserythropoietic, type IVb, 620969
KLF11	100%	100%	100%	98.6%	Maturity-onset diabetes of the young, type VII, 610508
KLF4	100%	100%	100%	99.4%	

KLF6	100%	100%	100%	99%	Gastric cancer, somatic, 613659;Prostate cancer, somatic, 176807
KLF7	100%	100%	100%	99%	
KLHL10	100%	100%	100%	99.3%	Spermatogenic failure 11, 615081
KLHL15	89.1%	89.1%	99.5%	73.1%	Intellectual developmental disorder, X-linked 103, 300982
KLHL20	100%	100%	100%	99.7%	
KLHL24	100%	100%	100%	99.9%	Cardiomyopathy, familial hypertrophic, 29, with polyglucosan bodies, 620236;Epidermolysis bullosa simplex 6, generalized intermediate, with or without cardiomyopathy, 617294
KLHL3	100%	100%	100%	99.6%	Pseudohypoaldosteronism, type IID, 614495
KLHL40	100%	100%	100%	99.3%	Nemaline myopathy 8, autosomal recessive, 615348
KLHL41	100%	100%	100%	99.7%	Nemaline myopathy 9, 615731
KLHL7	100%	100%	100%	99.3%	Retinitis pigmentosa 42, 612943;PERCHING syndrome, 617055
KLHL9	100%	100%	100%	99.7%	

KLK11	100%	100%	100%	99.2%	Ichthyosis with erythrokeratoderma, 620507
KLK4	100%	100%	100%	98.3%	Amelogenesis imperfecta, type IIA1, 204700
KLKB1	100%	100%	100%	99.9%	Fletcher factor (prekallikrein) deficiency, 612423
KLLN	100%	100%	100%	98.8%	Cowden syndrome 4, 615107
KMT2A	99.2%	99.2%	100%	99.2%	Wiedemann-Steiner syndrome, 605130
KMT2B	99.8%	99.4%	100%	98.2%	Intellectual developmental disorder, autosomal dominant 68, 619934;Dystonia 28, childhood-onset, 617284
KMT2C	100%	100%	100%	99.5%	Kleefstra syndrome 2, 617768
KMT2D	100%	100%	100%	98.6%	Branchial arch abnormalities, choanal atresia, athelia, hearing loss, and hypothyroidism syndrome, 620186;Kabuki syndrome 1, 147920
KMT2E	100%	100%	100%	98.5%	O'Donnell-Luria-Rodan syndrome, 618512
KMT5B	100%	100%	100%	99.1%	Intellectual developmental disorder, autosomal dominant 51, 617788

KNG1	100%	100%	100%	99.6%	[Kininogen deficiency], 228960;Angioedema, hereditary, 6, 619363;[High molecular weight kininogen deficiency], 228960
KNL1	98.7%	98.7%	100%	99.7%	Microcephaly 4, primary, autosomal recessive, 604321
KNSTRN	100%	100%	100%	99.5%	?Roifman-Chitayat syndrome, digenic, 613328
KPNA3	100%	100%	100%	99.6%	Spastic paraplegia 88, autosomal dominant, 620106
KPNA7	100%	100%	100%	99.3%	Oocyte/zygote/embryo maturation arrest 17, 620319
KPTN	100%	100%	100%	98.4%	Intellectual developmental disorder, autosomal recessive 41, 615637

KRAS	100%	100%	100%	99.8%	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
KREMEN1	100%	99.9%	100%	99.1%	Ectodermal dysplasia 13, hair/tooth type, 617392
KRIT1	100%	100%	100%	99.9%	Hyperkeratotic cutaneous capillary-venous malformations associated with cerebral capillary malformations, 116860;Cerebral cavernous malformations-1, 116860;Cavernous malformations of CNS and retina, 116860

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 histrix, Curth-Macklin type, 146590
KRT10	100%	100%	100%	98.9%	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
KRT12	100%	100%	100%	99.5%	Meesmann corneal dystrophy 1, 122100
KRT13	100%	100%	100%	98.6%	White sponge nevus 2, 615785

KRT14	100%	100%	100%	98.3%	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
KRT16	100%	100%	100%	98.3%	Palmoplantar keratoderma, nonepidermolytic, focal, 613000;Pachyonychia congenita 1, 167200
KRT17	100%	100%	100%	98.4%	Steatocystoma multiplex, 184500;Pachyonychia congenita 2, 167210
KRT18	100%	100%	100%	99.3%	Cirrhosis, cryptogenic, 215600;{Cirrhosis, noncryptogenic, susceptibility to}, 215600
KRT2	100%	100%	100%	98.8%	Ichthyosis bullosa of Siemens, 146800

KRT25	100%	100%	100%	99.8%	Woolly hair, autosomal recessive 3, 616760
KRT3	100%	100%	100%	99.3%	Meesmann corneal dystrophy 2, 618767
KRT4	100%	100%	100%	98.9%	White sponge nevus 1, 193900
KRT5	100%	100%	100%	99.4%	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
KRT6A	100%	100%	100%	99.3%	Pachyonychia congenita 3, 615726
KRT6B	100%	100%	100%	99.1%	Pachyonychia congenita 4, 615728

KRT6C	99.8%	99.6%	98.7%	92.5%	Palmoplantar keratoderma, nonepidermolytic, focal or diffuse, 615735
KRT71	100%	100%	100%	99.2%	?Hypotrichosis 13, 615896
KRT74	100%	100%	100%	98.9%	Woolly hair, autosomal dominant, 194300;?Hypotrichosis 3, 613981;?Ectodermal dysplasia 7, hair/nail type, 614929
KRT75	100%	100%	100%	99.4%	{Pseudofolliculitis barbae, susceptibility to}, 612318
KRT8	100%	100%	100%	98.9%	
KRT81	100%	100%	100%	98.3%	Monilethrix, 158000
KRT82	100%	100%	100%	99.3%	
KRT83	100%	100%	100%	99.5%	Monilethrix, 158000;Erythrokeratodermia variabilis et progressiva 5, 617756
KRT85	100%	100%	100%	99%	Ectodermal dysplasia 4, hair/nail type, 602032
KRT86	100%	100%	100%	99.2%	Monilethrix, 158000
KRT9	100%	100%	100%	97.8%	Palmoplantar keratoderma, epidermolytic, 1, 144200
KY	100%	100%	100%	98.9%	Myopathy, myofibrillar, 7, 617114
KYNU	100%	100%	100%	99.6%	?Hydroxykynureninuria, 236800;Vertebral, cardiac, renal, and limb defects syndrome 2, 617661

L1CAM	100%	99.7%	98.4%	68.6%	MASA syndrome, 303350;Hydrocephalus, congenital, X-linked, 307000;?Corpus callosum, partial agenesis of, 304100
L2HGDH	100%	100%	100%	99.7%	L-2-hydroxyglutaric aciduria, 236792
LACC1	100%	100%	100%	99.7%	Juvenile arthritis, 618795
LACTB	100%	100%	100%	99.9%	
LAGE3	100%	100%	98.4%	71%	Galloway-Mowat syndrome 2, X-linked, 301006
LAMA1	100%	100%	100%	99.5%	Poretti-Boltshauser syndrome, 615960
LAMA2	100%	100%	100%	99.8%	Muscular dystrophy, limb-girdle, autosomal recessive 23, 618138;Muscular dystrophy, congenital, merosin deficient or partially deficient, 607855
LAMA3	100%	100%	100%	99.6%	Epidermolysis bullosa, junctional 2A, intermediate, 619783;Epidermolysis bullosa, junctional 2C, laryngoonychocutaneous, 245660;Epidermolysis bullosa, junctional 2B, severe, 619784
LAMA4	100%	100%	100%	99.7%	Cardiomyopathy, dilated, 1JJ, 615235

LAMA5	100%	100%	100%	98.7%	Nephrotic syndrome, type 26, 620049;?Bent bone dysplasia syndrome 2, 620076
LAMB1	100%	99.8%	100%	99.7%	Lissencephaly 5, 615191
LAMB2	100%	100%	100%	99.3%	Nephrotic syndrome, type 5, with or without ocular abnormalities, 614199;Pierson syndrome, 609049
LAMB3	100%	100%	100%	99.3%	Epidermolysis bullosa, junctional 1B, severe, 226700;Epidermolysis bullosa, junctional 1A, intermediate, 226650;Amelogenesis imperfecta, type IA, 104530
LAMC2	100%	100%	100%	99.4%	Epidermolysis bullosa, junctional 3B, severe, 619786;Epidermolysis bullosa, junctional 3A, intermediate, 619785
LAMC3	100%	100%	100%	98.7%	Cortical malformations, occipital, 614115
LAMP2	85.3%	85.1%	98.7%	73.3%	Danon disease, 300257
LAMTOR2	100%	100%	100%	99.2%	Immunodeficiency due to defect in MAPBP-interacting protein, 610798
LAPTM5	100%	99.8%	100%	99.7%	

LARGE1	100%	100%	100%	99.5%	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
LARP1	100%	100%	100%	98.4%	
LARP7	100%	100%	100%	99.7%	Alazami syndrome, 615071
LARS1	100%	100%	100%	99.3%	?Infantile liver failure syndrome 1, 615438
LARS2	100%	100%	100%	99.6%	Perrault syndrome 4, 615300;Hydrops, lactic acidosis, and sideroblastic anemia, 617021
LAS1L	95.8%	95.4%	98.8%	68.6%	Wilson-Turner syndrome, 309585
LAT	100%	100%	100%	99.3%	Immunodeficiency 52, 617514
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

LBX1	100%	100%	100%	98.4%	?Central hypoventilation syndrome, congenital, 3, 619483
LCA5	100%	100%	100%	99.5%	Leber congenital amaurosis 5, 604537
LCAT	100%	100%	100%	97.5%	Fish-eye disease, 136120;Norum disease, 245900
LCK	100%	100%	100%	99.2%	Immunodeficiency 22, 615758
LCP1	100%	100%	100%	99.8%	
LCP2	100%	100%	100%	99.3%	Immunodeficiency 81, 619374
LCT	100%	100%	100%	99.2%	Lactase deficiency, congenital, 223000
LDB3	100%	100%	100%	98.7%	Left ventricular noncompaction 3, 601493;Cardiomyopathy, hypertrophic, 24, 601493;Myopathy, myofibrillar, 4, 609452;Cardiomyopathy, dilated, 1C, with or without LVNC, 601493
LDHA	100%	100%	100%	99.7%	Glycogen storage disease XI, 612933
LDHB	100%	100%	100%	99.6%	[Lactate dehydrogenase-B deficiency], 614128
LDHD	100%	100%	100%	99.4%	D-lactic aciduria with susceptibility to gout, 245450

LDLR	100%	100%	100%	98.8%	LDL cholesterol level QTL2, 143890;Hypercholesterolemia, familial, 1, 143890
LDLRAP1	100%	100%	100%	99%	Hypercholesterolemia, familial, 4, 603813
LEF1	100%	100%	100%	99.3%	
LEFTY2	100%	100%	100%	98.8%	
LEMD2	100%	100%	100%	99%	Marbach-Rustad progeroid syndrome, 619322;Cataract 46, juvenile-onset, 212500
LEMD3	100%	100%	100%	98.7%	Buschke-Ollendorff syndrome, 166700;Osteopoikilosis with or without melorheostosis, 166700
LEP	100%	100%	100%	98.3%	Obesity, morbid, due to leptin deficiency, 614962
LEPR	94.6%	94.6%	100%	99.8%	Obesity, morbid, due to leptin receptor deficiency, 614963
LETM1	100%	100%	100%	99.2%	Neurodegeneration, childhood-onset, with multisystem involvement due to mitochondrial dysfunction, 620089
LFNG	97.8%	92.4%	99.8%	94.9%	Spondylocostal dysostosis 3, autosomal recessive, 609813
LGI1	100%	100%	100%	99.7%	Epilepsy, familial temporal lobe, 1, 600512

LGI3	100%	100%	100%	98.9%	Intellectual developmental disorder with muscle tone abnormalities and distal skeletal defects, 620007
LGI4	100%	100%	100%	98.8%	Arthrogryposis multiplex congenita 1, neurogenic, with myelin defect, 617468
LGR4	100%	100%	100%	98.4%	Delayed puberty, self-limited, 619613;{Bone mineral density, low, susceptibility to}, 615311
LHB	100%	100%	100%	99.4%	Hypogonadotropic hypogonadism 23 with or without anosmia, 228300
LHCGR	100%	100%	100%	99.7%	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
LHFPL5	100%	100%	100%	99.3%	Deafness, autosomal recessive 67, 610265
LHX1	100%	100%	99.9%	96.4%	
LHX2	100%	100%	100%	98.4%	

LHX3	100%	100%	100%	99.1%	Pituitary hormone deficiency, combined, 3, 221750
LHX4	100%	100%	100%	99.1%	Pituitary hormone deficiency, combined, 4, 262700
LIAS	100%	100%	100%	99.8%	Hyperglycinemia, lactic acidosis, and seizures, 614462
LIFR	100%	100%	100%	99.9%	Stuve-Wiedemann syndrome/Schwartz-Jampel type 2 syndrome, 601559
LIG1	100%	100%	100%	98.4%	Immunodeficiency 96, 619774
LIG3	100%	100%	100%	99.4%	Mitochondrial DNA depletion syndrome 20 (MNGIE type), 619780
LIG4	100%	100%	100%	99.8%	LIG4 syndrome, 606593;{Multiple myeloma, resistance to}, 254500
LIM2	100%	100%	100%	97.3%	Cataract 19, multiple types, 615277
LIMS2	100%	100%	100%	99.3%	?Muscular dystrophy, autosomal recessive, with cardiomyopathy and triangular tongue, 616827
LINGO1	100%	100%	100%	97.2%	Intellectual developmental disorder, autosomal recessive 64, 618103

LINS1	100%	100%	100%	99.8%	Intellectual developmental disorder, autosomal recessive 27, 614340
LIPA	95.1%	95.1%	100%	99.6%	Wolman disease, 620151;Cholesteryl ester storage disease, 278000
LIPC	100%	100%	100%	99.2%	{Diabetes mellitus, noninsulin-dependent}, 125853;Hepatic lipase deficiency, 614025;[High density lipoprotein cholesterol level QTL 12], 612797
LIPE	100%	100%	100%	99.3%	Lipodystrophy, familial partial, type 6, 615980
LIPH	100%	100%	100%	99.4%	Hypotrichosis 7, 604379;Woolly hair, autosomal recessive 2 with or without hypotrichosis, 604379
LIPN	100%	100%	100%	99.5%	Ichthyosis, congenital, autosomal recessive 8, 613943
LIPT1	100%	100%	100%	99.9%	Lipoyltransferase 1 deficiency, 616299
LIPT2	100%	100%	100%	98.7%	Encephalopathy, neonatal severe, with lactic acidosis and brain abnormalities, 617668
LITAF	100%	100%	100%	97.8%	Charcot-Marie-Tooth disease, type 1C, 601098

LMAN1	100%	100%	100%	99.7%	Combined factor V and VIII deficiency, 227300
LMAN2L	100%	100%	100%	99.5%	?Intellectual developmental disorder, autosomal dominant 69, 617863;?Intellectual developmental disorder, autosomal recessive 52, 616887
LMBR1	100%	100%	100%	99.8%	Syndactyly, type IV, 186200;Laurin-Sandrow syndrome, 135750;Acheiropody, 200500;Triphalangeal thumb-polysyndactyly syndrome, 190605
LMBRD1	100%	100%	100%	99.7%	Methylmalonic aciduria and homocystinuria, cblF type, 277380
LMBRD2	100%	100%	100%	99.8%	Developmental delay with variable neurologic and brain abnormalities, 619694
LMCD1	100%	100%	100%	99.2%	
LMF1	100%	100%	100%	99.3%	Lipase deficiency, combined, 246650

LMNA	100%	100%	100%	98.7%	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
LMNB1	100%	100%	100%	99.4%	Leukodystrophy, adult-onset, autosomal dominant, 169500;Microcephaly 26, primary, autosomal dominant, 619179
LMNB2	99.9%	99.4%	99.9%	95.1%	Microcephaly 27, primary, autosomal dominant, 619180;?Epilepsy, progressive myoclonic, 9, 616540;{Lipodystrophy, partial, acquired, susceptibility to}, 608709

LMOD1	100%	100%	100%	98.2%	?Megacystis-microcolon-intestinal hypoperistalsis syndrome 3, 619362
LMOD2	100%	100%	100%	98.8%	Cardiomyopathy, dilated, 2G, 619897
LMOD3	100%	100%	100%	99.4%	Nemaline myopathy 10, 616165
LMX1A	100%	100%	100%	99.4%	Deafness, autosomal dominant 7, 601412
LMX1B	100%	100%	100%	97.8%	Focal segmental glomerulosclerosis 10, 256020;Nail-patella syndrome, 161200
LNPK	93.1%	93.1%	100%	99.8%	Neurodevelopmental disorder with epilepsy and hypoplasia of the corpus callosum, 618090
LONP1	100%	100%	100%	98.9%	CODAS syndrome, 600373
LORICRIN	100%	100%	100%	98.2%	Vohwinkel syndrome with ichthyosis, 604117
LOX	100%	100%	100%	98%	Aortic aneurysm, familial thoracic 10, 617168
LOXHD1	100%	100%	100%	99.2%	Deafness, autosomal recessive 77, 613079
LOXL3	100%	100%	100%	99.2%	Myopia 28, autosomal recessive, 619781

LPAR6	100%	100%	100%	99.8%	Hypotrichosis 8, 278150;Woolly hair, autosomal recessive 1, with or without hypotrichosis, 278150
LPIN1	100%	100%	100%	99.5%	Myoglobinuria, acute recurrent, autosomal recessive, 268200
LPIN2	99.5%	99.2%	100%	99.5%	Majeed syndrome, 609628
LPL	100%	100%	100%	99.4%	Lipoprotein lipase deficiency, 238600;[High density lipoprotein cholesterol level QTL 11], 238600;Combined hyperlipidemia, familial, 144250
LPP	100%	100%	100%	99.1%	Lipoma;Leukemia, acute myeloid, 601626
LRAT	100%	100%	100%	98.4%	Leber congenital amaurosis 14, 613341;Retinal dystrophy, early-onset severe, 613341;Retinitis pigmentosa, juvenile, 613341
LRBA	99.8%	99.8%	100%	99.7%	Immunodeficiency, common variable, 8, with autoimmunity, 614700
LRIF1	100%	100%	100%	99.6%	?Facioscapulohumeral muscular dystrophy 3, digenic, 619477
LRIG2	100%	100%	100%	99.8%	Urofacial syndrome 2, 615112

LRIT3	100%	100%	100%	99.9%	Night blindness, congenital stationary (complete), 1F, autosomal recessive, 615058
LRMDA	97.8%	97.8%	100%	99.2%	Albinism, oculocutaneous, type VII, 615179
LRP1	100%	100%	100%	99.1%	?Keratosis pilaris atrophicans, 604093;Developmental dysplasia of the hip 3, 620690
LRP12	100%	100%	100%	99.7%	Oculopharyngodistal myopathy 1, 164310;Amyotrophic lateral sclerosis 28, 620452
LRP2	100%	100%	100%	99.6%	Donnai-Barrow syndrome, 222448
LRP4	100%	100%	100%	99.3%	?Myasthenic syndrome, congenital, 17, 616304;Sclerosteosis 2, 614305;Cenani-Lenz syndactyly syndrome, 212780

LRP5	100%	100%	100%	98.3%	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
LRP6	100%	100%	100%	99.4%	{Coronary artery disease, autosomal dominant, 2}, 610947;Tooth agenesis, selective, 7, 616724
LRPAP1	100%	100%	100%	99%	Myopia 23, autosomal recessive, 615431
LRPPRC	96.8%	96.6%	100%	99.8%	Mitochondrial complex IV deficiency, nuclear type 5, (French-Canadian), 220111
LRRC10	100%	100%	100%	98.4%	
LRRC23	100%	100%	100%	98.9%	Spermatogenic failure 92, 620848
LRRC32	100%	100%	100%	99.1%	Cleft palate, proliferative retinopathy, and developmental delay, 619074
LRRC56	100%	100%	100%	99.5%	Ciliary dyskinesia, primary, 39, 618254
LRRC7	100%	100%	100%	99.7%	

LRRC8A	100%	100%	100%	98.6%	?Agammaglobulinemia 5, 613506
LRRK1	100%	100%	100%	99.5%	Osteosclerotic metaphyseal dysplasia, 615198
LRRK2	100%	100%	100%	99.6%	{Parkinson disease 8}, 607060
LRSAM1	100%	100%	100%	98.8%	Charcot-Marie-Tooth disease, axonal, type 2P, 614436
LRTOMT	100%	100%	100%	99.4%	Deafness, autosomal recessive 63, 611451
LSM1	100%	100%	100%	99.6%	
LSM11	100%	100%	100%	96.7%	?Aicardi-Goutieres syndrome 8, 619486
LSS	100%	100%	100%	99.1%	Hypotrichosis 14, 618275;Cataract 44, 616509;Alopecia-intellectual disability syndrome 4, 618840
LTBP1	100%	100%	100%	99.1%	Cutis laxa, autosomal recessive, type IIE, 619451
LTBP2	100%	100%	100%	99%	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

LTBP3	100%	100%	100%	98.1%	Dental anomalies and short stature, 601216;Geleophysic dysplasia 3, 617809
LTBP4	100%	100%	100%	98.2%	Cutis laxa, autosomal recessive, type IC, 613177
LTC4S	100%	100%	100%	99%	Leukotriene C4 synthase deficiency, 614037
LTV1	100%	100%	100%	99.7%	Inflammatory poikiloderma with hair abnormalities and acral keratoses, 620199
LYN	100%	100%	100%	99.6%	Autoinflammatory disease, systemic, with vasculitis, 620376
LYRM4	68%	68%	100%	99.2%	?Combined oxidative phosphorylation deficiency 19, 615595
LYRM7	100%	100%	100%	99.7%	Mitochondrial complex III deficiency, nuclear type 8, 615838
LYSET	100%	100%	100%	99.8%	Dysostosis multiplex, Ain-Naz type, 619345
LYST	99.5%	99.4%	100%	99.8%	Chediak-Higashi syndrome, 214500
LYZ	100%	100%	100%	99.9%	Amyloidosis, hereditary systemic 5, 620658
LZTFL1	100%	100%	100%	99.8%	Bardet-Biedl syndrome 17, 615994

LZTR1	100%	100%	100%	99.1%	Noonan syndrome 2, 605275;Noonan syndrome 10, 616564;{Schwannomatosis-2, susceptibility to}, 615670
LZTS1	100%	100%	100%	99.4%	Esophageal squamous cell carcinoma, somatic, 133239
M1AP	100%	100%	100%	99.6%	Spermatogenic failure 48, 619108
MAB21L1	100%	100%	100%	99.6%	Cerebellar, ocular, craniofacial, and genital syndrome, 618479
MAB21L2	100%	100%	100%	98.8%	Microphthalmia/coloboma and skeletal dysplasia syndrome, 615877
MACF1	100%	100%	100%	99.7%	Lissencephaly 9 with complex brainstem malformation, 618325
MAD1L1	100%	100%	100%	99.3%	Prostate cancer, somatic, 176807;Mosaic variegated aneuploidy syndrome 7 with inflammation and tumor predisposition, 620189;Lymphoma, B-cell, somatic
MAD2L2	100%	100%	100%	99.1%	?Fanconi anemia, complementation group V, 617243

MADD	100%	100%	100%	99.1%	Neurodevelopmental disorder with dysmorphic facies, impaired speech and hypotonia, 619005;DEEAH syndrome, 619004
MAF	92.2%	87.3%	99.9%	88.8%	Cataract 21, multiple types, 610202;Ayme-Gripp syndrome, 601088
MAFA	100%	98.7%	99.3%	80.9%	Insulinomatosis and diabetes mellitus, 147630
MAFB	100%	100%	100%	98.5%	Duane retraction syndrome 3, 617041;Multicentric carpotarsal osteolysis syndrome, 166300
MAG	100%	100%	100%	99%	Spastic paraplegia 75, autosomal recessive, 616680
MAGED2	100%	99.5%	98.7%	70%	Bartter syndrome, type 5, antenatal, transient, 300971
MAGEL2	100%	100%	100%	99.4%	Schaaf-Yang syndrome, 615547
MAGI2	98.4%	96.6%	99.9%	95.5%	Nephrotic syndrome, type 15, 617609
MAGT1	93.8%	93.5%	98.8%	72.1%	Immunodeficiency, X-linked, with magnesium defect, Epstein-Barr virus infection and neoplasia, 300853;Congenital disorder of glycosylation, type Icc, 301031

MAK	100%	100%	100%	99.8%	Retinitis pigmentosa 62, 614181
MAL	100%	100%	100%	97.6%	?Leukodystrophy, hypomyelinating, 28, 620978
MALT1	100%	100%	100%	99.6%	Immunodeficiency 12, 615468
MAML2	100%	100%	100%	99.1%	Mucoepidermoid salivary gland carcinoma
MAMLD1	100%	99.9%	99%	70.7%	Hypospadias 2, X-linked, 300758
MAN1B1	100%	99.5%	100%	99.2%	Rafiq syndrome, 614202
MAN2B1	100%	100%	100%	98.3%	Mannosidosis, alpha-, types I and II, 248500
MAN2B2	100%	100%	100%	99%	
MAN2C1	100%	100%	100%	99.3%	Congenital disorder of deglycosylation 2, 619775
MANBA	100%	100%	100%	99.7%	Mannosidosis, beta, 248510
MANF	100%	100%	100%	99.1%	Diabetes, deafness, developmental delay, and short stature syndrome, 620651
MAOA	100%	99.3%	99.2%	74%	Brunner syndrome, 300615
MAP1B	100%	100%	100%	99.2%	?Deafness, autosomal dominant 83, 619808;Periventricular nodular heterotopia 9, 618918
MAP1LC3B2	100%	100%	100%	99.2%	

MAP2K1	95.8%	95.8%	100%	99.5%	Cardiofaciocutaneous syndrome 3, 615279;Melorheostosis, isolated, somatic mosaic, 155950
MAP2K2	100%	100%	100%	98.2%	Cardiofaciocutaneous syndrome 4, 615280
MAP3K1	100%	100%	100%	99.1%	46XY sex reversal 6, 613762
MAP3K14	100%	100%	100%	99%	Immunodeficiency 112, 620449
MAP3K20	100%	100%	100%	99.4%	Centronuclear myopathy 6 with fiber-type disproportion, 617760;Split-foot malformation with mesoaxial polydactyly, 616890
MAP3K7	100%	99.9%	100%	99.5%	Frontometaphyseal dysplasia 2, 617137;Cardiospondylocarpofacial syndrome, 157800
MAP3K8	100%	100%	100%	99.2%	Lung cancer, somatic, 211980
MAP4K4	100%	99.9%	100%	99.3%	
MAPK1	100%	100%	100%	99.1%	Noonan syndrome 13, 619087
MAPK8	100%	100%	100%	99.8%	
MAPK8IP3	100%	100%	100%	99.2%	Neurodevelopmental disorder with or without variable brain abnormalities, 618443

MAPKAPK3	100%	100%	100%	99.1%	?Macular dystrophy, patterned, 3, 617111
MAPKAPK5	100%	100%	100%	99.6%	Neurocardiofaciodigital syndrome, 619869
MAPKBP1	100%	100%	100%	99.2%	Nephronophthisis 20, 617271
MAPRE2	100%	100%	100%	99.4%	Symmetric circumferential skin creases, congenital, 2, 616734
MAPT	95.4%	95.4%	100%	98.7%	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
MARCHF6	100%	100%	100%	99.3%	Epilepsy, familial adult myoclonic, 3, 613608
MARK2	100%	100%	100%	98.9%	
MARK3	100%	99.8%	100%	99.6%	?Visual impairment and progressive phthisis bulbi, 618283

MARS1	100%	100%	100%	99.2%	Spastic paraplegia 70, autosomal recessive, 620323;Interstitial lung and liver disease, 615486;?Trichothiodystrophy 9, nonphotosensitive, 619692;Charcot-Marie-Tooth disease, axonal, type 2U, 616280
MARS2	100%	100%	100%	99.8%	?Combined oxidative phosphorylation deficiency 25, 616430;Spastic ataxia 3, autosomal recessive, 611390
MARVELD2	100%	100%	100%	99.5%	Deafness, autosomal recessive 49, 610153
MASP1	100%	100%	100%	99.4%	3MC syndrome 1, 257920
MASP2	100%	100%	100%	99.5%	MASP2 deficiency, 613791
MAST1	100%	100%	100%	99%	Mega-corpus-callosum syndrome with cerebellar hypoplasia and cortical malformations, 618273
MAST3	100%	100%	100%	97.9%	Developmental and epileptic encephalopathy 108, 620115
MAST4	100%	100%	100%	98.7%	
MASTL	100%	100%	100%	99.6%	

MAT1A	100%	100%	100%	99%	Hypermethioninemia, persistent, autosomal dominant, due to methionine adenosyltransferase I/III deficiency, 250850;Methionine adenosyltransferase deficiency, autosomal recessive, 250850
MAT2A	100%	100%	100%	99.7%	
MATN3	100%	100%	100%	99.1%	{Osteoarthritis susceptibility 2}, 140600;Spondyloepimetaphyseal dysplasia, Borochowitz-Cormier-Daire type, 608728;Epiphyseal dysplasia, multiple, 5, 607078
MATR3	100%	100%	100%	99.4%	Amyotrophic lateral sclerosis 21, 606070
MAX	100%	100%	100%	99.5%	Polydactyly-macrocephaly syndrome, 620712;{Pheochromocytoma, susceptibility to}, 171300
MB	100%	100%	100%	98.4%	Myopathy, sarcoplasmic body, 620286
MBD4	100%	100%	100%	99.8%	{Uveal melanoma, susceptibility to, 1}, 606660;Tumor predisposition syndrome 2, 619975

MBD5	100%	100%	100%	99.6%	Intellectual developmental disorder, autosomal dominant 1, 156200
MBIP	100%	100%	100%	99.7%	
MBOAT7	100%	100%	100%	99.2%	Intellectual developmental disorder, autosomal recessive 57, 617188
MBTPS1	99.8%	99.2%	100%	99.3%	?Spondyloepiphyseal dysplasia, Kondo-Fu type, 618392
MBTPS2	100%	99.7%	98.9%	75.5%	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
MC2R	100%	100%	100%	99.8%	Glucocorticoid deficiency, due to ACTH unresponsiveness, 202200
MC4R	100%	100%	100%	99.8%	Obesity (BMIQ20), 618406;{Obesity, resistance to (BMIQ20)}, 618406
MCAT	100%	100%	100%	99.8%	Optic atrophy 15, 620583
MCC	100%	100%	100%	99.6%	Colorectal cancer, somatic, 114500

MCCC1	100%	100%	100%	99.4%	3-Methylcrotonyl-CoA carboxylase 1 deficiency, 210200
MCCC2	93.4%	93.4%	100%	99.7%	3-Methylcrotonyl-CoA carboxylase 2 deficiency, 210210
MCEE	100%	100%	100%	99.8%	Methylmalonyl-CoA epimerase deficiency, 251120
MCFD2	100%	100%	100%	98.9%	Factor V and factor VIII, combined deficiency of, 613625
MCIDAS	100%	100%	100%	99.2%	Ciliary dyskinesia, primary, 42, 618695
MCM10	100%	100%	100%	99.5%	Immunodeficiency 80 with or without cardiomyopathy, 619313
MCM2	100%	100%	100%	99.5%	?Deafness, autosomal dominant 70, 616968
MCM3AP	100%	100%	100%	99.3%	Peripheral neuropathy, autosomal recessive, with or without impaired intellectual development, 618124
MCM4	95.3%	95.3%	100%	99.6%	Immunodeficiency 54, 609981
MCM5	100%	100%	100%	98.7%	?Meier-Gorlin syndrome 8, 617564
MCM6	100%	100%	100%	99.6%	Lactase persistence/nonpersistence, 223100

MCM8	94.4%	94.4%	100%	99.8%	?Premature ovarian failure 10, 612885
MCM9	100%	100%	100%	99.5%	Ovarian dysgenesis 4, 616185
MCMDC2	100%	100%	100%	99.6%	
MCOLN1	100%	100%	100%	99.6%	Lisch epithelial corneal dystrophy, 620763;Mucolipidosis IV, 252650
MCPH1	94.8%	93.1%	100%	99.8%	Microcephaly 1, primary, autosomal recessive, 251200
MCTP2	100%	100%	100%	99.6%	
MCTS1	100%	99.9%	99.2%	76.4%	Immunodeficiency 118, mycobacteriosis, 301115
MCUR1	100%	100%	100%	98.9%	
MDFIC	100%	100%	100%	99.1%	Lymphatic malformation 12, 620014
MDH1	100%	100%	100%	99.3%	?Developmental and epileptic encephalopathy 88, 618959
MDH2	100%	100%	99.9%	98.1%	Developmental and epileptic encephalopathy 51, 617339
MDM2	94%	94%	100%	99.6%	{Accelerated tumor formation, susceptibility to}, 614401;?Lessel-Kubisch syndrome, 618681
MDM4	100%	100%	100%	99.6%	?Bone marrow failure syndrome 6, 618849

ME2	94.1%	94.1%	100%	99.8%	
MECOM	100%	100%	100%	99.6%	Radioulnar synostosis with amegakaryocytic thrombocytopenia 2, 616738
MECP2	100%	99.3%	97.1%	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
MECR	100%	100%	100%	99.2%	Dystonia, childhood-onset, with optic atrophy and basal ganglia abnormalities, 617282;Optic atrophy 16, 620629
MED11	100%	100%	100%	100%	Neurodegeneration with developmental delay, early respiratory failure, myoclonic seizures, and brain abnormalities, 620327

MED12	100%	99.6%	98.6%	67.4%	Lujan-Fryns syndrome, 309520;Ohdo syndrome, X-linked, 300895;Hardikar syndrome, 301068;Opitz-Kaveggia syndrome, 305450
MED12L	100%	100%	100%	99.7%	Nizon-Isidor syndrome, 618872
MED13	100%	100%	100%	99.7%	Intellectual developmental disorder, autosomal dominant 61, 618009
MED13L	100%	100%	100%	99.4%	Impaired intellectual development and distinctive facial features with or without cardiac defects, 616789
MED17	100%	100%	100%	99.7%	Microcephaly, postnatal progressive, with seizures and brain atrophy, 613668
MED23	100%	100%	100%	99.8%	Intellectual developmental disorder, autosomal recessive 18, with or without epilepsy, 614249
MED25	100%	100%	100%	98.2%	Basel-Vanagait-Smirin-Yosef syndrome, 616449
MED27	100%	100%	100%	98.9%	Neurodevelopmental disorder with spasticity, cataracts, and cerebellar hypoplasia, 619286

MEF2C	100%	100%	100%	99.4%	Chromosome 5q14.3 deletion syndrome, 613443;Neurodevelopmental disorder with hypotonia, stereotypic hand movements, and impaired language, 613443
MEFV	96.1%	96.1%	100%	99.3%	Neutrophilic dermatosis, acute febrile, 608068;Familial Mediterranean fever, AR, 249100;Familial Mediterranean fever, AD, 134610
MEGF10	100%	100%	100%	99.7%	Congenital myopathy 10A, severe variant, 614399;Congenital myopathy 10B, mild variant, 620249
MEGF8	100%	100%	100%	98.5%	Carpenter syndrome 2, 614976
MEI1	100%	100%	100%	99.4%	Hydatidiform mole, recurrent, 3, 618431
MEIOB	100%	100%	100%	99.8%	Premature ovarian failure 23, 620686;Spermatogenic failure 22, 617706
MEIS2	91.5%	91.5%	100%	98.8%	Cleft palate, cardiac defects, and impaired intellectual development, 600987

MEN1	100%	100%	100%	98.3%	Lipoma, somatic;Angiofibroma, somatic;Multiple endocrine neoplasia 1, 131100;Carcinoid tumor of lung;Adrenal adenoma, somatic;Parathyroid adenoma, somatic
MEOX1	100%	100%	100%	98.6%	Klippel-Feil syndrome 2, 214300
MERTK	100%	100%	100%	99.6%	Retinitis pigmentosa 38, 613862
MESD	100%	100%	100%	98.4%	Osteogenesis imperfecta, type XX, 618644
MESP2	99.9%	98.1%	100%	98.7%	Spondylocostal dysostosis 2, autosomal recessive, 608681
MET	100%	100%	100%	99.8%	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
METTL13	100%	100%	100%	99.4%	{?Deafness, autosomal recessive 26, modifier of}, 605429

METTL23	100%	100%	100%	99.9%	Intellectual developmental disorder, autosomal recessive 44, 615942
METTL5	100%	100%	100%	99.6%	Intellectual developmental disorder, autosomal recessive 72, 618665
MFAP5	100%	100%	100%	99.8%	Aortic aneurysm, familial thoracic 9, 616166
MFF	95.9%	95.9%	100%	99.7%	Encephalopathy due to defective mitochondrial and peroxisomal fission 2, 617086
MFN2	100%	100%	100%	99.1%	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
MFRP	100%	100%	100%	99.3%	Microphthalmia, isolated 5, 611040;Nanophthalmos 2, 609549
MFSD2A	100%	100%	100%	99%	Neurodevelopmental disorder with progressive microcephaly, spasticity, and brain abnormalities, 616486

MFSD8	100%	100%	100%	99.9%	Macular dystrophy with central cone involvement, 616170;Ceroid lipofuscinosis, neuronal, 7, 610951
MGAT2	100%	100%	100%	99%	Congenital disorder of glycosylation, type IIa, 212066
MGME1	100%	100%	100%	99.9%	Mitochondrial DNA depletion syndrome 11, 615084
MGP	100%	100%	100%	99.1%	Keutel syndrome, 245150
MIA3	100%	100%	100%	99.6%	?Ondontochondrodysplasia 2 with hearing loss and diabetes, 619269
MIB1	100%	100%	100%	99.6%	Left ventricular noncompaction 7, 615092
MICOS13	100%	100%	100%	98.4%	Combined oxidative phosphorylation deficiency 37, 618329
MICU1	100%	100%	100%	99.8%	Myopathy with extrapyramidal signs, 615673
MICU2	100%	100%	100%	99.9%	
MID1	100%	99.9%	98.6%	70.3%	Opitz GBBB syndrome, 300000
MID2	100%	99.8%	99.4%	75.4%	?Intellectual developmental disorder, X-linked 101, 300928
MIEF1	100%	100%	100%	99.2%	Optic atrophy 14, 620550

MIEF2	100%	100%	100%	98.4%	?Combined oxidative phosphorylation deficiency 49, 619024
MINAR2	100%	100%	100%	99.9%	Deafness, autosomal recessive 120, 620238
MINPP1	100%	100%	100%	99.7%	{Thyroid carcinoma, follicular}, 188470;Pontocerebellar hypoplasia, type 16, 619527
MIP	100%	100%	100%	99.3%	Cataract 15, multiple types, 615274
MIPEP	100%	100%	100%	99.6%	Combined oxidative phosphorylation deficiency 31, 617228
MIR140					Spondyloepiphyseal dysplasia, Nishimura type, 618618
MIR17HG					
MIR184					EDICT syndrome, 614303
MIR204					Retinal dystrophy and iris coloboma with or without cataract, 616722
MIR96					Deafness, autosomal dominant 50, 613074

MITF	100%	100%	100%	99.4%	Waardenburg syndrome, type 2A, 193510;{Melanoma, cutaneous malignant, susceptibility to, 8}, 614456;Tietz albinism-deafness syndrome, 103500;COMMAD syndrome, 617306
MKKS	100%	100%	100%	99.9%	McKusick-Kaufman syndrome, 236700;Bardet-Biedl syndrome 6, 605231
MKRN3	100%	100%	100%	99.7%	Precocious puberty, central, 2, 615346
MKS1	99.1%	99%	100%	99.2%	Bardet-Biedl syndrome 13, 615990;Meckel syndrome 1, 249000;Joubert syndrome 28, 617121
MLC1	100%	100%	100%	98.7%	Megalencephalic leukoencephalopathy with subcortical cysts 1, 604004
MLH1	100%	100%	100%	99.1%	Lynch syndrome 2, 609310;Muir-Torre syndrome, 158320;Mismatch repair cancer syndrome 1, 276300
MLH3	100%	100%	99.9%	98.9%	{Endometrial cancer, susceptibility to}, 608089;Colorectal cancer, somatic, 114500;Colorectal cancer, hereditary nonpolyposis, type 7, 614385

MLIP	100%	100%	100%	99.5%	Myopathy with myalgia, increased serum creatine kinase, and with or without episodic rhabdomyolysis, 620138
MLLT10	97%	97%	100%	99.2%	Leukemia, acute myeloid, 601626
MLPH	100%	100%	100%	98.6%	Griscelli syndrome, type 3, 609227
MLYCD	100%	100%	100%	98.9%	Malonyl-CoA decarboxylase deficiency, 248360
MMAA	100%	100%	100%	99.8%	Methylmalonic aciduria, vitamin B12-responsive, cblA type, 251100
MMAB	100%	100%	100%	99.1%	Methylmalonic aciduria, vitamin B12-responsive, cblB type, 251110
MMACHC	100%	100%	100%	99.6%	Methylmalonic aciduria and homocystinuria, cblC type, 277400
MMADHC	89.3%	89.3%	100%	99.3%	Methylmalonic aciduria and homocystinuria, cblD type, 277410;Methylmalonic aciduria, cblD type, 620953;Homocystinuria-megaloblastic anemia, cblD type, 620952
MME	97.7%	97.7%	100%	99.6%	?Spinocerebellar ataxia 43, 617018;Charcot-Marie-Tooth disease, axonal, type 2T, 617017

MMGT1	100%	100%	99.7%	73.7%	
MMP1	100%	100%	100%	99.9%	
MMP13	92.1%	92.1%	100%	99.5%	?Spondyloepimetaphyseal dysplasia, Missouri type, 602111;Metaphyseal anadysplasia 1, 602111;Metaphyseal dysplasia, Spahr type, 250400
MMP14	94.9%	94.9%	100%	98.4%	Winchester syndrome, 277950
MMP19	100%	100%	100%	99.4%	Cavitary optic disc anomalies, 611543
MMP2	100%	100%	100%	98.6%	Multicentric osteolysis, nodulosis, and arthropathy, 259600
MMP20	100%	100%	100%	99.6%	Amelogenesis imperfecta, type IIA2, 612529
MMP21	100%	100%	100%	99.3%	Heterotaxy, visceral, 7, autosomal, 616749
MMP9	100%	100%	100%	98.1%	Metaphyseal anadysplasia 2, 613073
MMS19	100%	100%	100%	99.7%	
MMUT	100%	100%	100%	99.7%	Methylmalonic aciduria, mut(0) type, 251000
MN1	100%	100%	100%	98.7%	CEBALID syndrome, 618774;Meningioma, 607174

MNS1	100%	100%	100%	99.7%	Heterotaxy, visceral, 9, autosomal, with male infertility, 618948
MNX1	92.5%	86.3%	96.9%	79.1%	Currarino syndrome, 176450
MOCOS	100%	100%	100%	99.1%	Xanthinuria, type II, 603592
MOCS1	100%	100%	100%	97.9%	Molybdenum cofactor deficiency A, 252150
MOCS2	100%	100%	100%	99.8%	Molybdenum cofactor deficiency B, 252160
MOG	100%	100%	100%	99.5%	?Narcolepsy 7, 614250
MOGS	100%	100%	100%	99.3%	Congenital disorder of glycosylation, type IIb, 606056
MORC2	100%	100%	100%	99%	Charcot-Marie-Tooth disease, axonal, type 2Z, 616688;Developmental delay, impaired growth, dysmorphic facies, and axonal neuropathy, 619090
MOS	100%	100%	99.9%	94.7%	Oocyte/zygote/embryo maturation arrest 20, 620383
MOV10L1	100%	100%	100%	99.2%	?Spermatogenic failure 73, 619878
MPC1	100%	100%	100%	99.9%	Mitochondrial pyruvate carrier deficiency, 614741
MPC2	100%	100%	100%	99.6%	

MPDU1	100%	100%	100%	98.5%	Congenital disorder of glycosylation, type If, 609180
MPDZ	100%	100%	100%	99.5%	Hydrocephalus, congenital, 2, with or without brain or eye anomalies, 615219
MPEG1	100%	100%	100%	99%	Immunodeficiency 77, 619223
MPI	100%	100%	100%	99.7%	Congenital disorder of glycosylation, type Ib, 602579
MPIG6B	100%	100%	100%	98.7%	?Thrombocytopenia, anemia, and myelofibrosis, 617441
MPL	100%	100%	100%	98.8%	Myelofibrosis with myeloid metaplasia, somatic, 254450;Amegakaryocytic thrombocytopenia, congenital, 1, 604498;Thrombocythemia 2, 601977
MPLKIP	100%	100%	100%	98.6%	Trichothiodystrophy 4, nonphotosensitive, 234050
MPO	100%	100%	100%	99.5%	{Alzheimer disease, susceptibility to}, 104300;Myeloperoxidase deficiency, 254600;{Lung cancer, protection against, in smokers}

MPV17	100%	100%	100%	99.6%	Charcot-Marie-Tooth disease, axonal, type 2EE, 618400;Mitochondrial DNA depletion syndrome 6 (hepatocerebral type), 256810
MPZ	100%	100%	100%	98.4%	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
MPZL2	100%	100%	100%	99.5%	Deafness, autosomal recessive 111, 618145
MRAP	100%	100%	100%	99.6%	Glucocorticoid deficiency 2, 607398
MRAS	100%	100%	100%	99.6%	Noonan syndrome 11, 618499
MRE11	100%	100%	100%	99.6%	Ataxia-telangiectasia-like disorder 1, 604391
MRM2	97%	97%	100%	98.6%	Mitochondrial DNA depletion syndrome 17, 618567

MRPL12	100%	100%	100%	99.7%	?Combined oxidative phosphorylation deficiency 45, 618951
MRPL24	100%	100%	100%	99.3%	
MRPL3	100%	100%	100%	99.8%	Combined oxidative phosphorylation deficiency 9, 614582
MRPL39	100%	100%	100%	99.7%	Combined oxidative phosphorylation deficiency 59, 620646
MRPL40	100%	100%	100%	99.4%	
MRPL42	100%	100%	100%	99.6%	
MRPL44	100%	100%	100%	99.7%	Combined oxidative phosphorylation deficiency 16, 615395
MRPL49	100%	100%	100%	99.3%	
MRPL50	100%	100%	100%	99.8%	
MRPL57	100%	100%	100%	98.4%	
MRPS14	100%	100%	100%	99.5%	?Combined oxidative phosphorylation deficiency 38, 618378
MRPS16	100%	100%	100%	98.9%	Combined oxidative phosphorylation deficiency 2, 610498
MRPS2	100%	100%	100%	99.4%	Combined oxidative phosphorylation deficiency 36, 617950

MRPS22	100%	100%	100%	99.7%	Ovarian dysgenesis 7, 618117;Combined oxidative phosphorylation deficiency 5, 611719
MRPS23	100%	100%	100%	99.5%	?Combined oxidative phosphorylation deficiency 46, 618952
MRPS25	74.2%	74.2%	100%	99.8%	?Combined oxidative phosphorylation deficiency 50, 619025
MRPS28	85.3%	85.3%	100%	96.4%	?Combined oxidative phosphorylation deficiency 47, 618958
MRPS34	100%	100%	100%	99.3%	Combined oxidative phosphorylation deficiency 32, 617664
MRPS36	100%	100%	100%	99.6%	
MRPS7	100%	100%	100%	99.3%	?Combined oxidative phosphorylation deficiency 34, 617872
MRRF	100%	100%	100%	99.9%	
MRTFA	100%	100%	100%	98.7%	?Immunodeficiency 66, 618847
MRTFB	100%	100%	100%	99.7%	
MS4A1	100%	100%	100%	99.7%	?Immunodeficiency, common variable, 5, 613495

MSH2	100%	100%	100%	99.3%	Lynch syndrome 1, 120435;Muir-Torre syndrome, 158320;Mismatch repair cancer syndrome 2, 619096
MSH3	100%	100%	100%	98.5%	Familial adenomatous polyposis 4, 617100;Endometrial carcinoma, somatic, 608089
MSH4	100%	100%	100%	99.7%	Premature ovarian failure 20, 619938;Spermatogenic failure 2, 108420
MSH5	100%	100%	100%	98.8%	?Premature ovarian failure 13, 617442;Spermatogenic failure 74, 619937
MSH6	100%	100%	100%	98.4%	Lynch syndrome 5, 614350;Mismatch repair cancer syndrome 3, 619097;{Endometrial cancer, familial}, 608089
MSL2	92.7%	92.7%	100%	99.5%	Karayol-Borroto-Haghshenas neurodevelopmental syndrome, 620985
MSL3	100%	99.7%	99.1%	71.3%	Basilicata-Akhtar syndrome, 301032
MSMO1	100%	100%	100%	100%	Microcephaly, congenital cataract, and psoriasiform dermatitis, 616834
MSN	100%	98.3%	99.1%	74.4%	Immunodeficiency 50, 300988

MSR1	100%	100%	100%	98.7%	Barrett esophagus/esophageal adenocarcinoma, 614266
MSRB3	100%	100%	100%	99.6%	Deafness, autosomal recessive 74, 613718
MSTN	100%	100%	100%	99.6%	?Muscle hypertrophy, 614160
MSTO1	100%	100%	99.9%	96.4%	Myopathy, mitochondrial, and ataxia, 617675
MSX1	100%	100%	100%	97.7%	Tooth agenesis, selective, 1, with or without orofacial cleft, 106600;Ectodermal dysplasia 3, Witkop type, 189500;Orofacial cleft 5, 608874
MSX2	100%	100%	100%	99%	Parietal foramina with cleidocranial dysplasia, 168550;Craniosynostosis 2, 604757;Parietal foramina 1, 168500
MTAP	100%	100%	100%	98.8%	Diaphyseal medullary stenosis with malignant fibrous histiocytoma, 112250
MTFMT	100%	100%	100%	97.6%	Combined oxidative phosphorylation deficiency 15, 614947;Mitochondrial complex I deficiency, nuclear type 27, 618248

MTHFD1	100%	100%	100%	99.4%	{Neural tube defects, folate-sensitive, susceptibility to}, 601634;Combined immunodeficiency and megaloblastic anemia with or without hyperhomocysteinemia, 617780
MTHFR	100%	100%	100%	99.1%	{Vascular disease, susceptibility to};Homocystinuria due to MTHFR deficiency, 236250;{Thromboembolism, susceptibility to}, 188050;{Schizophrenia, susceptibility to}, 181500;{Neural tube defects, susceptibility to}, 601634
MTHFS	100%	100%	100%	98%	Neurodevelopmental disorder with microcephaly, epilepsy, and hypomyelination, 618367
MTM1	100%	99.9%	99.3%	74.8%	Myopathy, centronuclear, X-linked, 310400
MTMR2	100%	100%	100%	99.9%	Charcot-Marie-Tooth disease, type 4B1, 601382
MTO1	94.7%	92.3%	100%	99.7%	Combined oxidative phosphorylation deficiency 10, 614702

MTOR	100%	100%	100%	99.4%	Focal cortical dysplasia, type II, somatic, 607341;Smith-Kingsmore syndrome, 616638
MTPAP	100%	100%	100%	99.4%	?Spastic ataxia 4, autosomal recessive, 613672
MTR	100%	100%	100%	99.5%	{Neural tube defects, folate-sensitive, susceptibility to}, 601634;Homocystinuria-megaloblastic anemia, cblG complementation type, 250940
MTRFR	100%	99.8%	100%	98.8%	Spastic paraplegia 55, autosomal recessive, 615035;Combined oxidative phosphorylation deficiency 7, 613559
MTRR	100%	100%	100%	99.2%	Homocystinuria-megaloblastic anemia, cbl E type, 236270;{Neural tube defects, folate-sensitive, susceptibility to}, 601634
MTSS2	100%	100%	100%	98.4%	Intellectual developmental disorder with ocular anomalies and distinctive facial features, 620086
MT-TI	99.6%	95.9%			
MTTP	100%	100%	100%	99.9%	Abetalipoproteinemia, 200100

MTX2	100%	100%	100%	99.5%	Mandibuloacral dysplasia progeroid syndrome, 619127
MUC1	100%	100%	100%	98.5%	Tubulointerstitial kidney disease, autosomal dominant, 2, 174000
MUC16	100%	100%	100%	99.5%	
MUSK	100%	100%	99.8%	98.2%	Fetal akinesia deformation sequence 1, 208150;Myasthenic syndrome, congenital, 9, associated with acetylcholine receptor deficiency, 616325
MUTYH	100%	100%	100%	99.5%	Adenomas, multiple colorectal, 608456;Gastric cancer, somatic, 613659
MVD	100%	100%	100%	99%	Porokeratosis 7, multiple types, 614714
MVK	100%	100%	100%	99.2%	Hyper-IgD syndrome, 260920;Porokeratosis 3, multiple types, 175900;Mevalonic aciduria, 610377
MXI1	100%	100%	100%	97.1%	Prostate cancer, somatic, 176807;Neurofibrosarcoma, somatic
MYBPC1	100%	100%	100%	99.7%	Congenital myopathy 16, 618524;Lethal congenital contracture syndrome 4, 614915;Arthrogryposis, distal, type 1B, 614335

MYBPC3	100%	100%	100%	99%	Cardiomyopathy, hypertrophic, 4, 115197;Cardiomyopathy, dilated, 1MM, 615396;Left ventricular noncompaction 10, 615396
MYBPHL	100%	100%	100%	98.9%	
MYC	100%	100%	100%	99.3%	Burkitt lymphoma, somatic, 113970
MYCBP2	100%	99.8%	100%	99.5%	
MYCN	100%	100%	100%	97.1%	Feingold syndrome 1, 164280;Megalencephaly-polydactyly syndrome, 620748
MYD88	100%	100%	100%	99.7%	Macroglobulinemia, Waldenstrom, somatic, 153600;Immunodeficiency 68, 612260
MYF5	100%	100%	100%	99.5%	Ophthalmoplegia, external, with rib and vertebral anomalies, 618155
MYH10	100%	100%	100%	99.4%	
MYH11	100%	100%	100%	98.7%	Megacystis-microcolon-intestinal hypoperistalsis syndrome 2, 619351;Aortic aneurysm, familial thoracic 4, 132900;Visceral myopathy 2, 619350

MYH14	100%	100%	100%	98.1%	?Peripheral neuropathy, myopathy, hoarseness, and hearing loss, 614369;Deafness, autosomal dominant 4A, 600652
MYH2	100%	100%	100%	99.6%	Congenital myopathy 6 with ophthalmoplegia, 605637
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
MYH6	100%	100%	100%	99.1%	?Atrial septal defect 3, 614089;{Sick sinus syndrome 3}, 614090;Cardiomyopathy, dilated, 1EE, 613252;Cardiomyopathy, hypertrophic, 14, 613251

MYH7	100%	100%	100%	99.2%	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
MYH7B	100%	100%	100%	98.8%	
MYH8	100%	100%	100%	99.6%	Carney complex variant, 608837;Trismus-pseudocamptodactyly syndrome, 158300
MYH9	97.3%	97.2%	100%	99.1%	Macrothrombocytopenia and granulocyte inclusions with or without nephritis or sensorineural hearing loss, 155100;Deafness, autosomal dominant 17, 603622
MYL1	100%	100%	100%	99.7%	Congenital myopathy 14, 618414
MYL11	100%	100%	100%	98.9%	Arthrogryposis, distal, type 1C, 619110

MYL2	100%	100%	100%	99.2%	Cardiomyopathy, hypertrophic, 10, 608758;Myopathy, myofibrillar, 12, infantile-onset, with cardiomyopathy, 619424
MYL3	100%	100%	100%	99.4%	Cardiomyopathy, hypertrophic, 8, 608751
MYL4	100%	100%	100%	98.7%	?Atrial fibrillation, familial, 18, 617280
MYL7	100%	100%	100%	99%	
MYL9	100%	100%	100%	99.1%	?Megacystis-microcolon-intestinal hypoperistalsis syndrome 4, 619365
MYLK	99.2%	99.2%	100%	99.2%	Megacystis-microcolon-intestinal hypoperistalsis syndrome 1, 249210;Aortic aneurysm, familial thoracic 7, 613780
MYLK2	100%	100%	100%	99.1%	Cardiomyopathy, hypertrophic, 1, digenic, 192600
MYLK3	100%	100%	100%	99.4%	
MYMK	100%	100%	100%	99.1%	Carey-Fineman-Ziter syndrome, 254940
MYMX	100%	100%	100%	99.9%	?Carey-Fineman-Ziter syndrome 2, 619941
MYO15A	100%	100%	100%	98.6%	Deafness, autosomal recessive 3, 600316

MYO18B	100%	100%	100%	98.8%	Klippel-Feil syndrome 4, autosomal recessive, with myopathy and facial dysmorphism, 616549
MYO1E	100%	100%	100%	99.6%	Glomerulosclerosis, focal segmental, 6, 614131
MYO1H	100%	100%	100%	99.4%	?Central hypoventilation syndrome, congenital, 2, and autonomic dysfunction, 619482
MYO3A	100%	100%	100%	99.6%	Deafness, autosomal recessive 30, 607101;Deafness, autosomal dominant 90, 620722
MYO5A	99%	99%	100%	99.7%	Griscelli syndrome, type 1, 214450
MYO5B	100%	100%	100%	99.4%	Diarrhea 2, with microvillus atrophy, with or without cholestasis, 251850;Cholestasis, progressive familial intrahepatic, 10, 619868
MYO6	100%	100%	100%	99.8%	Deafness, autosomal dominant 22, with hypertrophic cardiomyopathy, 606346;Deafness, autosomal dominant 22, 606346;Deafness, autosomal recessive 37, 607821

MYO7A	100%	100%	100%	99.2%	Deafness, autosomal recessive 2, 600060;Usher syndrome, type 1B, 276900;Deafness, autosomal dominant 11, 601317
MYO9A	100%	100%	100%	99.7%	Myasthenic syndrome, congenital, 24, presynaptic, 618198
MYOC	100%	100%	100%	98.5%	Glaucoma 1A, primary open angle, 137750
MYOCD	100%	100%	100%	99.4%	Megabladder, congenital, 618719
MYOD1	100%	100%	100%	97%	Congenital myopathy 17, 618975
MYOF	100%	100%	100%	99.4%	?Angioedema, hereditary, 7, 619366
MYOM1	100%	100%	100%	99.3%	
MYORG	100%	100%	100%	98.8%	Basal ganglia calcification, idiopathic, 7, autosomal recessive, 618317
MYOT	100%	100%	100%	99.7%	Myopathy, myofibrillar, 3, 609200
MYOZ2	100%	100%	100%	99.6%	Cardiomyopathy, hypertrophic, 16, 613838

MYPN	98.4%	98.4%	100%	99.5%	Cardiomyopathy, hypertrophic, 22, 615248;Congenital myopathy 24, 617336;Cardiomyopathy, familial restrictive, 4, 615248;Cardiomyopathy, dilated, 1KK, 615248
MYRF	100%	100%	100%	98.5%	Encephalitis/encephalopathy, mild, with reversible myelin vacuolization, 618113;Cardiac-urogenital syndrome, 618280
MYSM1	100%	100%	100%	99.8%	Bone marrow failure syndrome 4, 618116
MYT1L	100%	100%	100%	99.2%	Intellectual developmental disorder, autosomal dominant 39, 616521
MYZAP	100%	100%	100%	99.3%	Cardiomyopathy, dilated, 2K, 620894
NAA10	100%	99.8%	98.9%	68.4%	Microphthalmia, syndromic 1, 309800;Ogden syndrome, 300855
NAA15	100%	100%	100%	99.6%	Intellectual developmental disorder, autosomal dominant 50, with behavioral abnormalities, 617787
NAA20	100%	100%	100%	100%	Intellectual developmental disorder, autosomal recessive 73, 619717

NAA60	100%	100%	100%	98.7%	Basal ganglia calcification, idiopathic, 9, autosomal recessive, 620786
NAA80	100%	100%	100%	98.5%	?Auroneurodental syndrome, 620830
NACC1	100%	100%	100%	99%	Neurodevelopmental disorder with epilepsy, cataracts, feeding difficulties, and delayed brain myelination, 617393
NADK2	100%	100%	100%	99.4%	2,4-dienoyl-CoA reductase deficiency, 616034
NADSYN1	100%	100%	100%	99.2%	Vertebral, cardiac, renal, and limb defects syndrome 3, 618845
NAE1	100%	100%	100%	99.8%	Neurodevelopmental disorder with dysmorphic facies and ischiopubic hypoplasia, 620210
NAF1	100%	100%	100%	99%	Pulmonary fibrosis and/or bone marrow failure syndrome, telomere-related, 7, 620365
NAGA	100%	100%	100%	99.1%	Schindler disease, type I, 609241;Kanzaki disease, 609242;Schindler disease, type III, 609241
NAGLU	100%	100%	100%	99.4%	?Charcot-Marie-Tooth disease, axonal, type 2V, 616491;Mucopolysaccharidosis type IIIB (Sanfilippo B), 252920

NAGS	100%	100%	100%	99.5%	N-acetylglutamate synthase deficiency, 237310
NALCN	100%	100%	100%	99.7%	Congenital contractures of the limbs and face, hypotonia, and developmental delay, 616266;Hypotonia, infantile, with psychomotor retardation and characteristic facies 1, 615419
NANOS1	100%	99.1%	99.5%	85.6%	Spermatogenic failure 12, 615413
NANS	100%	100%	100%	99.1%	Spondyloepimetaphyseal dysplasia, Genevieve type, 610442
NAPB	100%	100%	100%	99.8%	Developmental and epileptic encephalopathy 107, 620033
NARS1	100%	100%	100%	99.8%	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

NARS2	92.3%	92.3%	100%	99.7%	Combined oxidative phosphorylation deficiency 24, 616239;?Deafness, autosomal recessive 94, 618434
NAT8L	96.4%	85.8%	98.8%	90.1%	?N-acetylaspartate deficiency, 614063
NAV3	100%	100%	100%	99.5%	
NAXD	98.7%	94.8%	100%	99%	Encephalopathy, progressive, early-onset, with brain edema and/or leukoencephalopathy, 2, 618321
NAXE	97.7%	92.7%	100%	99.4%	Encephalopathy, progressive, early-onset, with brain edema and/or leukoencephalopathy, 617186
NBAS	100%	99.9%	100%	99.7%	Short stature, optic nerve atrophy, and Pelger-Huet anomaly, 614800;Infantile liver failure syndrome 2, 616483
NBEA	97.6%	97.6%	100%	99.3%	Neurodevelopmental disorder with or without early-onset generalized epilepsy, 619157
NBEAL2	100%	100%	100%	99.1%	Gray platelet syndrome, 139090

NBN	97.5%	97.5%	100%	99.9%	Leukemia, acute lymphoblastic, 613065;Aplastic anemia, 609135;Nijmegen breakage syndrome, 251260
NCAPD2	100%	100%	100%	99.5%	Microcephaly 21, primary, autosomal recessive, 617983
NCAPD3	100%	100%	100%	99.7%	Microcephaly 22, primary, autosomal recessive, 617984
NCAPG2	100%	100%	100%	99.6%	Khan-Khan-Katsanis syndrome, 618460
NCAPH	100%	100%	100%	99.5%	?Microcephaly 23, primary, autosomal recessive, 617985
NCDN	100%	100%	100%	98.8%	Neurodevelopmental disorder with infantile epileptic spasms, 619373
NCF1	100%	99.7%	100%	97.3%	Chronic granulomatous disease 1, autosomal recessive, 233700
NCF2	100%	100%	100%	99.5%	Chronic granulomatous disease 2, autosomal recessive, 233710
NCF4	100%	100%	100%	99%	Chronic granulomatous disease 3, autosomal recessive, 613960
NCKAP1	100%	100%	100%	99.8%	
NCKAP1L	100%	100%	100%	99.5%	Immunodeficiency 72 with autoinflammation, 618982

NCOA3	100%	100%	100%	99.7%	
NCOR2	100%	100%	100%	98.2%	
NCSTN	100%	100%	100%	98.8%	Acne inversa, familial, 1, 142690
NDC1	100%	100%	100%	99.7%	
NDE1	100%	100%	100%	97.8%	Microhydranencephaly, 605013;Lissencephaly 4 (with microcephaly), 614019
NDN	100%	100%	100%	97.7%	
NDNF	100%	100%	100%	99.9%	Hypogonadotropic hypogonadism 25 with anosmia, 618841
NDP	100%	99.7%	99%	72.6%	Exudative vitreoretinopathy 2, X-linked, 305390;Norrie disease, 310600
NDRG1	100%	100%	100%	99.4%	Charcot-Marie-Tooth disease, type 4D, 601455
NDST1	100%	100%	100%	99%	Intellectual developmental disorder, autosomal recessive 46, 616116
NDUFA1	100%	99.4%	98.5%	71.2%	Mitochondrial complex I deficiency, nuclear type 12, 301020
NDUFA10	84.3%	81.5%	100%	99.5%	Mitochondrial complex I deficiency, nuclear type 22, 618243
NDUFA11	99.9%	98.4%	100%	98.8%	Mitochondrial complex I deficiency, nuclear type 14, 618236

NDUFA12	79.4%	79.4%	100%	99.8%	Mitochondrial complex I deficiency, nuclear type 23, 618244
NDUFA13	100%	100%	100%	99.3%	{Thyroid carcinoma, Hurthle cell}, 607464;Mitochondrial complex I deficiency, nuclear type 28, 618249
NDUFA2	100%	100%	100%	98.8%	Mitochondrial complex I deficiency, nuclear type 13, 618235
NDUFA3	94.1%	86.9%	100%	97.7%	
NDUFA4	100%	100%	100%	100%	?Mitochondrial complex IV deficiency, nuclear type 21, 619065
NDUFA5	75%	75%	100%	99.4%	
NDUFA6	100%	100%	100%	99.8%	Mitochondrial complex I deficiency, nuclear type 33, 618253
NDUFA7	100%	100%	100%	98.3%	
NDUFA8	100%	100%	100%	99.8%	Mitochondrial complex I deficiency, nuclear type 37, 619272
NDUFA9	100%	100%	100%	99.8%	Mitochondrial complex I deficiency, nuclear type 26, 618247
NDUFAB1	100%	100%	100%	98.8%	
NDUFAF1	100%	100%	100%	99.5%	Mitochondrial complex I deficiency, nuclear type 11, 618234

NDUFAF2	67.4%	67.4%	100%	99.6%	Mitochondrial complex I deficiency, nuclear type 10, 618233
NDUFAF3	100%	100%	100%	99.8%	Mitochondrial complex I deficiency, nuclear type 18, 618240
NDUFAF4	100%	100%	100%	99.8%	Mitochondrial complex I deficiency, nuclear type 15, 618237
NDUFAF5	100%	100%	100%	99.5%	Mitochondrial complex I deficiency, nuclear type 16, 618238
NDUFAF6	100%	100%	100%	99.8%	Mitochondrial complex I deficiency, nuclear type 17, 618239;Fanconi renotubular syndrome 5, 618913
NDUFAF7	100%	100%	100%	99.7%	
NDUFAF8	100%	100%	100%	99.4%	Mitochondrial complex I deficiency, nuclear type 34, 618776
NDUFB1	100%	100%	100%	98.8%	
NDUFB10	100%	100%	100%	99.1%	?Mitochondrial complex I deficiency, nuclear type 35, 619003
NDUFB11	99%	93.8%	92%	56%	Linear skin defects with multiple congenital anomalies 3, 300952;?Mitochondrial complex I deficiency, nuclear type 30, 301021
NDUFB2	100%	100%	100%	99.5%	

NDUFB3	100%	100%	100%	100%	Mitochondrial complex I deficiency, nuclear type 25, 618246
NDUFB4	100%	100%	100%	98.2%	
NDUFB5	100%	100%	100%	99.8%	
NDUFB6	100%	100%	100%	99.5%	
NDUFB7	100%	100%	100%	99.2%	?Mitochondrial complex I deficiency, nuclear type 39, 620135
NDUFB8	100%	100%	100%	99.3%	Mitochondrial complex I deficiency, nuclear type 32, 618252
NDUFB9	100%	100%	100%	99.6%	?Mitochondrial complex I deficiency, nuclear type 24, 618245
NDUFC1	100%	100%	100%	97.9%	
NDUFC2	100%	100%	100%	99.2%	Mitochondrial complex I deficiency, nuclear type 36, 619170
NDUFS1	100%	100%	100%	99.7%	Mitochondrial complex I deficiency, nuclear type 5, 618226
NDUFS2	99.7%	97.2%	100%	99.3%	?Leber-like hereditary optic neuropathy, autosomal recessive 2, 620569;Mitochondrial complex I deficiency, nuclear type 6, 618228

NDUFS3	96.7%	91.9%	100%	99.1%	Mitochondrial complex I deficiency, nuclear type 8, 618230
NDUFS4	100%	100%	100%	99.8%	Mitochondrial complex I deficiency, nuclear type 1, 252010
NDUFS5	100%	100%	100%	99.6%	
NDUFS6	100%	100%	100%	99%	Mitochondrial complex I deficiency, nuclear type 9, 618232
NDUFS7	100%	100%	100%	98.3%	Mitochondrial complex I deficiency, nuclear type 3, 618224
NDUFS8	100%	100%	100%	98.8%	Mitochondrial complex I deficiency, nuclear type 2, 618222
NDUFV1	100%	100%	100%	97%	Mitochondrial complex I deficiency, nuclear type 4, 618225
NDUFV2	100%	100%	100%	99.8%	Mitochondrial complex I deficiency, nuclear type 7, 618229
NDUFV3	100%	100%	100%	98.6%	
NEB	99.9%	99.8%	99.7%	98.1%	Nemaline myopathy 2, autosomal recessive, 256030;Arthrogryposis multiplex congenita 6, 619334
NEBL	100%	100%	100%	99.2%	

NECAP1	100%	100%	100%	99.5%	Developmental and epileptic encephalopathy 21, 615833
NECTIN1	93.4%	93.4%	100%	98%	Cleft lip/palate-ectodermal dysplasia syndrome, 225060;Orofacial cleft 7, 225060
NECTIN4	100%	100%	100%	98.9%	Ectodermal dysplasia-syndactyly syndrome 1, 613573
NEDD4L	100%	100%	100%	98.9%	Periventricular nodular heterotopia 7, 617201
NEFH	100%	100%	100%	97.9%	Charcot-Marie-Tooth disease, axonal, type 2CC, 616924;{?Amyotrophic lateral sclerosis, susceptibility to}, 105400
NEFL	100%	100%	100%	98.1%	Charcot-Marie-Tooth disease, type 1F, 607734;Charcot-Marie-Tooth disease, dominant intermediate G, 617882;Charcot-Marie-Tooth disease, type 2E, 607684
NEK1	100%	100%	100%	99.8%	Short-rib thoracic dysplasia 6 with or without polydactyly, 263520;?Orofaciodigital syndrome II, 252100;{Amyotrophic lateral sclerosis, susceptibility to, 24}, 617892

NEK10	100%	100%	100%	99.8%	Ciliary dyskinesia, primary, 44, 618781
NEK11	100%	100%	100%	99.6%	
NEK2	95.9%	95.9%	100%	99.6%	?Retinitis pigmentosa 67, 615565
NEK8	100%	100%	100%	99.3%	Renal-hepatic-pancreatic dysplasia 2, 615415;Polycystic kidney disease 8, 620903;?Nephronophthisis 9, 613824
NEK9	100%	100%	100%	99.5%	?Arthrogryposis, Perthes disease, and upward gaze palsy, 614262;Nevus comedonicus, somatic, 617025;Lethal congenital contracture syndrome 10, 617022
NEMF	100%	100%	100%	98.5%	Intellectual developmental disorder with speech delay and axonal peripheral neuropathy, 619099
NEPRO	100%	100%	100%	99.8%	Anauxetic dysplasia 3, 618853
NEU1	100%	100%	100%	99.3%	Sialidosis, type II, 256550;Sialidosis, type I, 256550
NEUROD1	100%	100%	100%	99.4%	{Type 2 diabetes mellitus, susceptibility to}, 125853;Maturity-onset diabetes of the young 6, 606394

NEUROD2	100%	100%	100%	93.8%	Developmental and epileptic encephalopathy 72, 618374
NEUROG1	100%	100%	100%	97.5%	Cranial dysinnervation disorder, congenital, with absent corneal reflex and developmental delay, 620469
NEUROG3	100%	100%	100%	97.2%	Diarrhea 4, malabsorptive, congenital, 610370
NEXMIF	100%	100%	99%	72.1%	Intellectual developmental disorder, X-linked 98, 300912
NEXN	100%	100%	100%	99.7%	Cardiomyopathy, dilated, 1CC, 613122;Cardiomyopathy, hypertrophic, 20, 613876
NF1	99.4%	99.4%	100%	99.7%	Watson syndrome, 193520;Leukemia, juvenile myelomonocytic, 607785;Neurofibromatosis, familial spinal, 162210;Neurofibromatosis, type 1, 162200;Neurofibromatosis-Noonan syndrome, 601321
NF2	100%	100%	100%	99.3%	Meningioma, NF2-related, somatic, 607174;Schwannomatosis, vestibular, 101000;Schwannomatosis, somatic, 101000

NFASC	100%	100%	100%	99.2%	Neurodevelopmental disorder with central and peripheral motor dysfunction, 618356
NFAT5	100%	100%	100%	99.8%	
NFATC1	100%	100%	100%	96.8%	
NFATC2	100%	100%	100%	98.2%	?Joint contracture, osteochondromas, and B-cell lymphoma, 620232
NFE2	100%	100%	100%	99.2%	
NFE2L2	81.2%	81.2%	100%	99.4%	Immunodeficiency, developmental delay, and hypohomocysteinemia, 617744
NFIA	92.2%	92.2%	100%	98.1%	Brain malformations with or without urinary tract defects, 613735
NFIB	100%	100%	100%	99.9%	Macrocephaly, acquired, with impaired intellectual development, 618286
NFIX	99.8%	98.8%	99.9%	96.8%	Marshall-Smith syndrome, 602535;Malan syndrome, 614753
NFKB1	100%	100%	100%	99.5%	Immunodeficiency, common variable, 12, 616576
NFKB2	100%	100%	100%	98.7%	Immunodeficiency, common variable, 10, 615577
NFKBIA	100%	100%	100%	99.4%	Ectodermal dysplasia and immunodeficiency 2, 612132

NFS1	89.8%	89.8%	100%	99.4%	Combined oxidative phosphorylation deficiency 52, 619386
NFU1	100%	100%	100%	99.8%	Spastic paraplegia 93, autosomal recessive, 620938;Multiple mitochondrial dysfunctions syndrome 1, 605711
NGF	100%	100%	100%	99.3%	Neuropathy, hereditary sensory and autonomic, type V, 608654
NGLY1	100%	100%	100%	99.3%	Congenital disorder of deglycosylation 1, 615273
NHEJ1	100%	100%	100%	99.4%	Microphthalmia/coloboma 13, 620968;Immunodeficiency 124, severe combined, 611291
NHERF1	100%	100%	100%	98.2%	Nephrolithiasis/osteoporosis, hypophosphatemic, 2, 612287
NHLH2	100%	100%	100%	97.6%	?Hypogonadotropic hypogonadism 27 without anosmia, 619755
NHLRC1	100%	100%	100%	99.6%	Myoclonic epilepsy of Lafora 2, 620681
NHLRC2	100%	100%	100%	99.6%	FINCA syndrome, 618278
NHP2	100%	100%	100%	99%	Dyskeratosis congenita, autosomal recessive 2, 613987

NHS	100%	99.9%	98.9%	71.9%	Cataract 40, X-linked, 302200;Nance-Horan syndrome, 302350
NIN	100%	100%	100%	99.5%	?Seckel syndrome 7, 614851
NIPA1	100%	100%	100%	96.7%	Spastic paraplegia 6, autosomal dominant, 600363
NIPAL4	100%	100%	100%	97.7%	Ichthyosis, congenital, autosomal recessive 6, 612281
NIPBL	100%	100%	100%	99.7%	Cornelia de Lange syndrome 1, 122470
NKAP	100%	99.5%	99.2%	72.7%	Intellectual developmental disorder, X-linked syndromic, Hackman-Di Donato type, 301039
NKPD1	100%	100%	100%	98.9%	
NKX2-1	100%	100%	100%	98.7%	Chorea, hereditary benign, 118700;{Thyroid cancer, nonmedullary, 1}, 188550;Choreoathetosis, hypothyroidism, and neonatal respiratory distress, 610978

NKX2-5	100%	100%	100%	99%	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
NKX2-6	100%	100%	100%	99.2%	Persistent truncus arteriosus, 217095;Conotruncal heart malformations, 217095
NKX3-2	100%	100%	100%	99.1%	Spondylo-megaepiphyseal-metaphyseal dysplasia, 613330
NKX6-2	100%	100%	100%	96.4%	Spastic ataxia 8, autosomal recessive, with hypomyelinating leukodystrophy, 617560
NLGN2	100%	100%	100%	97.9%	
NLGN3	99.1%	97.2%	98%	68.8%	{Autism susceptibility, X-linked 1}, 300425
NLGN4X	100%	100%	98.7%	69.2%	Intellectual developmental disorder, X-linked, 300495;{Autism susceptibility, X-linked 2}, 300495

NLRC4	100%	100%	100%	99.8%	?Familial cold autoinflammatory syndrome 4, 616115;Autoinflammation with infantile enterocolitis, 616050
NLRP1	98.1%	98.1%	100%	99.4%	{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
NLRP12	100%	100%	100%	99.1%	Familial cold autoinflammatory syndrome 2, 611762
NLRP2	100%	100%	100%	99.5%	Oocyte/zygote/embryo maturation arrest 18, 620332
NLRP3	100%	100%	100%	99.3%	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

NLRP5	100%	100%	100%	99.4%	Oocyte/zygote/embryo maturation arrest 19, 620333
NLRP6	100%	100%	100%	99.6%	
NLRP7	100%	100%	100%	99.3%	Hydatidiform mole, recurrent, 1, 231090
NME1	100%	100%	100%	99.7%	
NME3	100%	100%	100%	98.8%	
NME5	100%	100%	100%	99.2%	Ciliary dyskinesia, primary, 48, without situs inversus, 620032
NME8	100%	100%	100%	99.8%	?Ciliary dyskinesia, primary, 6, 610852
NMNAT1	100%	97.8%	100%	99.8%	Spondyloepiphyseal dysplasia, sensorineural hearing loss, intellectual developmental disorder, and Leber congenital amaurosis, 619260;Leber congenital amaurosis 9, 608553
NMNAT2	100%	100%	100%	99.2%	
NNT	96.3%	96.3%	100%	99.5%	Glucocorticoid deficiency 4, with or without mineralocorticoid deficiency, 614736
NOBOX	100%	100%	100%	98.6%	Premature ovarian failure 5, 611548

NOD2	100%	100%	100%	99.6%	Blau syndrome, 186580;{Yao syndrome}, 617321;{Inflammatory bowel disease 1, Crohn disease}, 266600
NODAL	100%	100%	100%	98.9%	Heterotaxy, visceral, 5, 270100
NOG	100%	100%	100%	97.5%	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
NOL3	100%	100%	100%	99.6%	?Myoclonus, familial, 1, 614937
NONO	96.2%	90.7%	99.1%	71.5%	Intellectual developmental disorder, X-linked syndromic 34, 300967
NOP10	92.5%	92.5%	100%	98.8%	?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
NOP56	100%	100%	100%	99.5%	Spinocerebellar ataxia 36, 614153

NOS1	100%	100%	100%	98.6%	
NOS1AP	100%	100%	100%	98.8%	Nephrotic syndrome, type 22, 619155
NOS2	100%	100%	100%	99.2%	{Malaria, resistance to}, 611162
NOTCH1	99.4%	99.1%	100%	98.8%	Adams-Oliver syndrome 5, 616028;Aortic valve disease 1, 109730
NOTCH2	100%	100%	100%	99.7%	Alagille syndrome 2, 610205;Hajdu-Cheney syndrome, 102500
NOTCH2NLC	100%	100%	100%	97.4%	Tremor, hereditary essential, 6, 618866;Oculopharyngodistal myopathy 3, 619473;Neuronal intranuclear inclusion disease, 603472
NOTCH3	100%	100%	100%	98.6%	Lateral meningocele syndrome, 130720;?Myofibromatosis, infantile 2, 615293;Cerebral arteriopathy with subcortical infarcts and leukoencephalopathy 1, 125310
NOVA2	100%	98.7%	99%	88.1%	Neurodevelopmental disorder with or without autistic features and/or structural brain abnormalities, 618859
NPAT	100%	100%	100%	99.8%	

NPC1	100%	100%	100%	99.4%	Niemann-Pick disease, type C1, 257220;Niemann-Pick disease, type D, 257220
NPC2	100%	100%	100%	99.1%	Niemann-pick disease, type C2, 607625
NPHP1	100%	100%	100%	99.3%	Joubert syndrome 4, 609583;Nephronophthisis 1, juvenile, 256100;Senior-Loken syndrome-1, 266900
NPHP3	100%	100%	100%	99.6%	Nephronophthisis 3, 604387;Renal-hepatic-pancreatic dysplasia 1, 208540;Meckel syndrome 7, 267010
NPHP4	100%	100%	100%	99.1%	Senior-Loken syndrome 4, 606996;Nephronophthisis 4, 606966
NPHS1	100%	100%	100%	99.3%	Nephrotic syndrome, type 1, 256300
NPHS2	100%	100%	100%	98.8%	Nephrotic syndrome, type 2, 600995
NPL	100%	100%	100%	99.8%	
NPM1	87.6%	87.6%	100%	98.7%	Leukemia, acute myeloid, somatic, 601626
NPPA	100%	100%	100%	99%	Atrial standstill 2, 615745;Atrial fibrillation, familial, 6, 612201
NPPB	100%	100%	100%	99.6%	
NPPC	100%	100%	100%	99.7%	

NPR2	100%	99.9%	100%	99%	Epiphyseal chondrodysplasia, Miura type, 615923;Short stature with nonspecific skeletal abnormalities, 616255;Acromesomelic dysplasia 1, Maroteaux type, 602875
NPR3	100%	100%	100%	98.5%	Boudin-Mortier syndrome, 619543
NPRL2	100%	100%	100%	99.5%	Epilepsy, familial focal, with variable foci 2, 617116
NPRL3	100%	100%	100%	98.7%	Epilepsy, familial focal, with variable foci 3, 617118
NPTX1	100%	100%	100%	96.9%	Spinocerebellar ataxia 50, 620158
NR0B1	100%	99.6%	98.7%	71.8%	Adrenal hypoplasia, congenital, 300200;46XY sex reversal 2, dosage-sensitive, 300018
NR0B2	100%	100%	100%	98.8%	Obesity, mild, early-onset, 601665
NR1H4	100%	100%	100%	99.7%	Cholestasis, progressive familial intrahepatic, 5, 617049
NR2E3	100%	100%	100%	99.6%	Retinitis pigmentosa 37, 611131;Enhanced S-cone syndrome, 268100
NR2F1	100%	99.7%	100%	96.4%	Bosch-Boonstra-Schaaf optic atrophy syndrome, 615722

NR2F2	100%	100%	100%	98.1%	46XX sex reversal 5, 618901;Congenital heart defects, multiple types, 4, 615779
NR3C1	100%	100%	100%	99.8%	Glucocorticoid resistance, 615962
NR3C2	100%	100%	100%	98.5%	Pseudohypoaldosteronism type I, autosomal dominant, 177735;Hypertension, early-onset, autosomal dominant, with exacerbation in pregnancy, 605115
NR4A2	100%	100%	100%	99.1%	Intellectual developmental disorder with language impairment and early-onset DOPA-responsive dystonia-parkinsonism, 619911
NR4A3	100%	100%	100%	96.7%	Chondrosarcoma, extraskeletal myxoid, 612237
NR5A1	100%	99.9%	100%	98.9%	46XX sex reversal 4, 617480;Premature ovarian failure 7, 612964;46XY sex reversal 3, 612965;Adrenocortical insufficiency, 612964;Spermatogenic failure 8, 613957
NRAP	100%	100%	100%	99.4%	

NRAS	100%	100%	100%	99.6%	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
NRCAM	100%	100%	100%	99.8%	Neurodevelopmental disorder with neuromuscular and skeletal abnormalities, 619833
NRG1	100%	100%	100%	98.9%	{?Schizophrenia, susceptibility to}, 603013
NRIP1	100%	100%	100%	99.6%	?Congenital anomalies of kidney and urinary tract 3, 618270
NRL	100%	100%	100%	98%	Retinitis pigmentosa 27, 613750;Retinal degeneration, autosomal recessive, clumped pigment type

NRROS	100%	100%	100%	98.8%	Seizures, early-onset, with neurodegeneration and brain calcification, 618875
NRXN1	100%	100%	100%	99.2%	Pitt-Hopkins-like syndrome 2, 614325;{Schizophrenia, susceptibility to, 17}, 614332
NSD1	100%	100%	100%	99.5%	Sotos syndrome, 117550
NSD2	99.5%	99.4%	100%	98.9%	Rauch-Steindl syndrome, 619695
NSDHL	100%	99.3%	99%	72.3%	CK syndrome, 300831;CHILD syndrome, 308050
NSF	100%	100%	99.8%	92.1%	Developmental and epileptic encephalopathy 96, 619340
NSMCE2	100%	100%	100%	99.4%	Seckel syndrome 10, 617253
NSMCE3	100%	100%	100%	98.4%	Lung disease, immunodeficiency, and chromosome breakage syndrome, 617241
NSMF	100%	100%	100%	98.8%	Hypogonadotropic hypogonadism 9 with or without anosmia, 614838
NSRP1	91%	91%	100%	99.4%	Neurodevelopmental disorder with spasticity, seizures, and brain abnormalities, 620001
NSUN2	100%	100%	100%	99.5%	Intellectual developmental disorder, autosomal recessive 5, 611091

NSUN3	100%	100%	100%	99.6%	Combined oxidative phosphorylation deficiency 48, 619012
NSUN6	100%	100%	100%	99.5%	Intellectual developmental disorder, autosomal recessive 82, 620779
NT5C2	100%	100%	100%	99.6%	Spastic paraplegia 45, autosomal recessive, 613162
NT5C3A	100%	100%	100%	99.8%	Anemia, congenital, nonspherocytic hemolytic, 8, 266120
NT5E	100%	100%	100%	98.3%	Calcification of joints and arteries, 211800
NTF4	100%	100%	100%	99.1%	Glaucoma 1, open angle, 1O, 613100
NTHL1	100%	100%	100%	99.3%	Familial adenomatous polyposis 3, 616415
NTN1	100%	100%	100%	98.7%	Mirror movements 4, 618264
NTNG2	100%	100%	100%	99.1%	Neurodevelopmental disorder with behavioral abnormalities, absent speech, and hypotonia, 618718
NTRK1	100%	100%	100%	99.3%	Insensitivity to pain, congenital, with anhidrosis, 256800

NTRK2	89.5%	89.5%	100%	99.3%	Developmental and epileptic encephalopathy 58, 617830;Obesity, hyperphagia, and developmental delay, 613886
NUAK2	100%	100%	100%	98.4%	?Anencephaly 2, 619452
NUBPL	100%	100%	100%	99.9%	Mitochondrial complex I deficiency, nuclear type 21, 618242
NUDCD3	100%	100%	100%	99.7%	
NUDT2	100%	100%	100%	99.8%	Intellectual developmental disorder with or without peripheral neuropathy, 619844
NUMA1	100%	100%	100%	98.8%	
NUP107	100%	100%	100%	99.8%	?Ovarian dysgenesis 6, 618078;Galloway-Mowat syndrome 7, 618348;Nephrotic syndrome, type 11, 616730
NUP133	100%	100%	100%	99.7%	?Galloway-Mowat syndrome 8, 618349;Nephrotic syndrome, type 18, 618177
NUP155	100%	100%	100%	99.7%	?Atrial fibrillation 15, 615770
NUP160	100%	100%	100%	99.6%	?Nephrotic syndrome, type 19, 618178
NUP188	100%	100%	100%	99.3%	Sandestig-Stefanova syndrome, 618804

NUP205	100%	100%	100%	99.6%	?Nephrotic syndrome, type 13, 616893
NUP214	100%	100%	100%	99.4%	Leukemia, T-cell acute lymphoblastic, somatic, 613065;Leukemia, acute myeloid, somatic, 601626;{Encephalopathy, acute, infection-induced, susceptibility to, 9}, 618426
NUP37	100%	100%	100%	99.9%	?Microcephaly 24, primary, autosomal recessive, 618179
NUP54	100%	100%	100%	99.5%	Dystonia 37, early-onset, with striatal lesions, 620427
NUP62	100%	100%	99.9%	98.7%	Striatonigral degeneration, infantile, 271930
NUP85	100%	100%	100%	99.2%	Nephrotic syndrome, type 17, 618176
NUP88	93.4%	93.4%	100%	99.8%	Fetal akinesia deformation sequence 4, 618393
NUP93	95.5%	95.5%	100%	99.4%	Nephrotic syndrome, type 12, 616892
NUS1	100%	100%	100%	99.1%	Intellectual developmental disorder, autosomal dominant 55, with seizures, 617831;?Congenital disorder of glycosylation, type 1aa, 617082
NUTF2	97.5%	96.9%	100%	98.5%	

NUTM2B-AS1					?Oculopharyngeal myopathy with leukoencephalopathy 1, 618637
NXF5					
NXN	100%	100%	100%	98.4%	Robinow syndrome, autosomal recessive 2, 618529
NYNRIN	100%	100%	100%	99.3%	
NYX	100%	100%	98.2%	65.7%	Night blindness, congenital stationary (complete), 1A, X-linked, 310500
OAS1	100%	100%	100%	99%	Immunodeficiency 100 with pulmonary alveolar proteinosis and hypogammaglobulinemia, 618042
OAT	100%	100%	100%	99.7%	Gyrate atrophy of choroid and retina with or without ornithinemia, 258870
OBSL1	100%	100%	100%	98.7%	3-M syndrome 2, 612921
OCA2	100%	100%	100%	99.5%	[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

OCLN	94.5%	94.5%	100%	98.4%	Pseudo-TORCH syndrome 1, 251290
OCRL	100%	99.9%	99.5%	75.4%	Dent disease 2, 300555;Lowe syndrome, 309000
ODAD1	100%	100%	100%	98.6%	Ciliary dyskinesia, primary, 20, 615067
ODAD2	96.1%	96.1%	100%	99.8%	Ciliary dyskinesia, primary, 23, 615451
ODAD3	100%	100%	100%	97.8%	Ciliary dyskinesia, primary, 30, 616037
ODAD4	100%	100%	100%	98.5%	Ciliary dyskinesia, primary, 35, 617092
ODAM	100%	100%	100%	99.6%	
ODAPH	100%	100%	100%	99.7%	Amelogenesis imperfecta, type IIA4, 614832
ODC1	100%	100%	100%	99.8%	Bachmann-Bupp syndrome, 619075
OFD1	100%	99.9%	99%	74.1%	Simpson-Golabi-Behmel syndrome, type 2, 300209;?Retinitis pigmentosa 23, 300424;Orofaciodigital syndrome I, 311200;Joubert syndrome 10, 300804
OGDH	100%	100%	100%	99.3%	Oxoglutarate dehydrogenase deficiency, 203740

OGDHL	100%	100%	100%	99.4%	Yoon-Bellen neurodevelopmental syndrome, 619701
OGG1	100%	100%	100%	99.2%	Renal cell carcinoma, clear cell, somatic, 144700
OGT	100%	99.6%	99.1%	74.9%	Intellectual developmental disorder, X-linked 106, 300997
OOEP	100%	100%	100%	98.3%	
OPA1	100%	100%	100%	99.6%	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
OPA3	100%	100%	100%	99%	3-methylglutaconic aciduria, type III, 258501;Optic atrophy 3 with cataract, 165300
OPCML	100%	100%	100%	99.4%	Ovarian cancer, somatic, 167000
OPHN1	94%	93.8%	99.3%	72.4%	Intellectual developmental disorder, X-linked syndromic, Billuart type, 300486
OPLAH	100%	100%	100%	98.7%	5-oxoprolinase deficiency, 260005

OPN1LW	99.5%	98.5%	92.5%	63.4%	Blue cone monochromacy, 303700;Colorblindness, protan, 303900
OPN1MW	96.8%	90.8%	73.5%	46.8%	Colorblindness, deutan, 303800;Blue cone monochromacy, 303700
OPN1SW	100%	100%	100%	99.1%	Colorblindness, tritan, 190900
OPTN	100%	100%	100%	99.3%	Glaucoma 1, open angle, E, 137760;Amyotrophic lateral sclerosis 12 with or without frontotemporal dementia, 613435;{Glaucoma, normal tension, susceptibility to}, 606657
ORAI1	100%	100%	100%	97.8%	Immunodeficiency 9, 612782;Myopathy, tubular aggregate, 2, 615883
ORC1	100%	100%	100%	99.4%	Meier-Gorlin syndrome 1, 224690
ORC4	100%	100%	100%	99.9%	Meier-Gorlin syndrome 2, 613800
ORC6	100%	100%	100%	99.7%	Meier-Gorlin syndrome 3, 613803
OSBPL2	100%	100%	100%	98.9%	Deafness, autosomal dominant 67, 616340
OSGEP	100%	100%	100%	99.3%	Galloway-Mowat syndrome 3, 617729
OSMR	100%	100%	100%	99.8%	Amyloidosis, primary localized cutaneous, 1, 105250

OSTC	84.7%	84.7%	100%	99.5%	
OSTM1	99.8%	98.5%	100%	99.5%	Osteopetrosis, autosomal recessive 5, 259720
OTC	100%	99.8%	99.3%	75.6%	Ornithine transcarbamylase deficiency, 311250
OTOA	100%	100%	100%	99.3%	Deafness, autosomal recessive 22, 607039
OTOF	100%	100%	100%	98.6%	Auditory neuropathy, autosomal recessive, 1, 601071;Deafness, autosomal recessive 9, 601071
OTOG	100%	100%	100%	99.3%	Deafness, autosomal recessive 18B, 614945
OTOGL	100%	100%	100%	99.6%	Deafness, autosomal recessive 84B, 614944
OTUD5	100%	99.1%	97.4%	64.8%	Multiple congenital anomalies-neurodevelopmental syndrome, X-linked, 301056
OTUD6B	100%	100%	100%	99.6%	Intellectual developmental disorder with dysmorphic facies, seizures, and distal limb anomalies, 617452
OTUD7A	99.3%	97%	99.9%	94.2%	Neurodevelopmental disorder with hypotonia and seizures, 620790

OTULIN	100%	100%	100%	99.2%	Autoinflammation, panniculitis, and dermatosis syndrome, 617099;{Immunodeficiency 107, susceptibility to invasive staphylococcus aureus infection}, 619986
OTX2	100%	100%	100%	99.7%	Retinal dystrophy, early-onset, with or without pituitary dysfunction, 610125;Pituitary hormone deficiency, combined, 6, 613986;Microphthalmia, syndromic 5, 610125
OVOL2	100%	99.9%	100%	99%	Corneal dystrophy, posterior polymorphous, 1, 122000
OXA1L	100%	100%	100%	99.6%	
OXCT1	100%	100%	100%	99.8%	Succinyl CoA:3-oxoacid CoA transferase deficiency, 245050
OXGR1	100%	100%	100%	99.8%	Nephrolithiasis, calcium oxalate, 2, with nephrocalcinosis, 620374
OXR1	100%	100%	100%	99.6%	Cerebellar hypoplasia/atrophy, epilepsy, and global developmental delay, 213000
P2RX2	100%	100%	100%	97.4%	Deafness, autosomal dominant 41, 608224
P2RY12	100%	100%	100%	99.8%	Bleeding disorder, platelet-type, 8, 609821

P3H1	100%	100%	100%	99.3%	Osteogenesis imperfecta, type VIII, 610915
P3H2	100%	100%	100%	99.3%	Myopia, high, with cataract and vitreoretinal degeneration, 614292
P4HA2	100%	100%	100%	99.6%	Myopia 25, autosomal dominant, 617238
P4HB	96%	94.6%	100%	99%	Cole-Carpenter syndrome 1, 112240
P4HTM	100%	100%	99.9%	97.3%	Hypotonia, hypoventilation, impaired intellectual development, dysautonomia, epilepsy, and eye abnormalities, 618493
PABPC1	98.8%	98.5%	100%	99.6%	
PABPC1L	100%	100%	100%	99.4%	
PABPN1	100%	100%	100%	95%	Oculopharyngeal muscular dystrophy, 164300
PACS1	100%	100%	100%	98.3%	Schuurs-Hoeijmakers syndrome, 615009
PACS2	100%	99.9%	100%	98.4%	Developmental and epileptic encephalopathy 66, 618067
PADI3	100%	100%	100%	99.3%	Uncombable hair syndrome, 191480
PADI6	100%	99.9%	100%	97.6%	Oocyte/zygote/embryo maturation arrest 16, 617234

PAFAH1B1	100%	100%	100%	99.4%	Subcortical laminar heterotopia, 607432;Lissencephaly 1, 607432
PAH	100%	100%	100%	99.8%	[Hyperphenylalaninemia, non-PKU mild], 261600;Phenylketonuria, 261600
PAICS	100%	100%	100%	99.8%	?Phosphoribosylaminoimidazole carboxylase deficiency, 619859
PAK1	100%	100%	100%	99.4%	Intellectual developmental disorder with macrocephaly, seizures, and speech delay, 618158
PAK2	100%	100%	100%	99.8%	?Knobloch syndrome 2, 618458
PAK3	100%	99.9%	99.1%	73.9%	Intellectual developmental disorder, X-linked 30, 300558
PALB2	100%	100%	100%	99.7%	{Breast-ovarian cancer, familial, susceptibility to, 5}, 620442;{Pancreatic cancer, susceptibility to, 3}, 613348;Fanconi anemia, complementation group N, 610832
PALS1	100%	100%	100%	99.6%	
PAM16	84.5%	84.5%	100%	99%	Spondylometaphyseal dysplasia, Megarbane-Dagher-Melike type, 613320

PAN2	100%	100%	100%	99.4%	
PANK2	100%	100%	100%	99.8%	Neurodegeneration with brain iron accumulation 1, 234200
PANK4	100%	100%	100%	98.6%	?Cataract 49, 619593
PANX1	100%	100%	100%	99.7%	Oocyte/zygote/embryo maturation arrest 7, 618550
PAPPA2	100%	100%	100%	99.4%	Short stature, Dauber-Argente type, 619489
PAPSS2	100%	100%	100%	99.3%	Brachyolmia 4 with mild epiphyseal and metaphyseal changes, 612847
PARK7	100%	100%	100%	99.8%	Parkinson disease 7, autosomal recessive early-onset, 606324
PARN	97.3%	95.3%	100%	99.7%	Dyskeratosis congenita, autosomal recessive 6, 616353;Pulmonary fibrosis and/or bone marrow failure syndrome, telomere-related, 4, 616371
PARP4	100%	100%	100%	99.5%	
PARP6	100%	100%	100%	99.5%	
PARS2	100%	100%	100%	99.3%	Developmental and epileptic encephalopathy 75, 618437
PATL2	100%	100%	100%	99.4%	Oocyte/zygote/embryo maturation arrest 4, 617743

PAX1	100%	100%	100%	97.3%	Otofaciocervical syndrome 2 with T-cell deficiency, 615560
PAX2	100%	100%	100%	98.9%	Glomerulosclerosis, focal segmental, 7, 616002;Papillorenal syndrome, 120330
PAX3	100%	100%	100%	99.1%	Craniofacial-deafness-hand syndrome, 122880;Waardenburg syndrome, type 3, 148820;Waardenburg syndrome, type 1, 193500;Rhabdomyosarcoma 2, alveolar, 268220
PAX4	100%	100%	100%	99%	{Diabetes mellitus, ketosis-prone, susceptibility to}, 612227;Maturity-onset diabetes of the young, type IX, 612225;Diabetes mellitus, type 2, 125853
PAX5	100%	100%	100%	99.1%	{Leukemia, acute lymphoblastic, susceptibility to, 3}, 615545

PAX6	100%	100%	100%	99.1%	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
PAX7	100%	100%	100%	99%	Congenital myopathy 19, 618578;Rhabdomyosarcoma 2, alveolar, 268220
PAX8	100%	100%	100%	99.3%	Hypothyroidism, congenital, due to thyroid dysgenesis or hypoplasia, 218700
PAX9	100%	100%	100%	98.1%	Tooth agenesis, selective, 3, 604625
PBRM1	100%	100%	100%	99.7%	?Renal cell carcinoma, clear cell, 144700
PBX1	100%	100%	100%	99%	Congenital anomalies of kidney and urinary tract syndrome with or without hearing loss, abnormal ears, or developmental delay, 617641
PC	100%	100%	100%	98.5%	Pyruvate carboxylase deficiency, 266150

PCARE	100%	100%	100%	99.1%	Retinitis pigmentosa 54, 613428
PCBD1	100%	100%	100%	99.4%	Hyperphenylalaninemia, BH4-deficient, D, 264070
PCCA	100%	100%	100%	99.8%	Propionicacidemia, 606054
PCCB	99.2%	96.1%	100%	99.7%	Propionicacidemia, 606054
PCDH12	100%	100%	100%	99.2%	Diencephalic-mesencephalic junction dysplasia syndrome 1, 251280
PCDH15	100%	100%	100%	99.5%	Usher syndrome, type 1D/F digenic, 601067;Deafness, autosomal recessive 23, 609533;Usher syndrome, type 1F, 602083
PCDH19	100%	99.8%	98.8%	68.3%	Developmental and epileptic encephalopathy 9, 300088
PCDHGC4	100%	100%	100%	98.9%	Neurodevelopmental disorder with poor growth and skeletal anomalies, 619880
PCGF2	100%	100%	100%	98.9%	Turnpenny-Fry syndrome, 618371
PCK1	100%	100%	100%	99.1%	Phosphoenolpyruvate carboxykinase deficiency, cytosolic, 261680
PCK2	100%	100%	100%	99%	PEPCK deficiency, mitochondrial, 261650
PCLO	100%	100%	100%	99%	?Pontocerebellar hypoplasia, type 3, 608027

PCNA	100%	100%	100%	99.7%	?Ataxia-telangiectasia-like disorder 2, 615919
PCNT	100%	100%	100%	99.2%	Microcephalic osteodysplastic primordial dwarfism, type II, 210720
PCSK1	100%	100%	100%	99.5%	{Obesity, susceptibility to, BMIQ12}, 612362;Endocrinopathy due to proprotein convertase 1/3 deficiency, 600955
PCSK9	100%	100%	100%	99.4%	{Low density lipoprotein cholesterol level QTL 1}, 603776;Hypercholesterolemia, familial, 3, 603776
PCYT1A	100%	100%	100%	99.6%	Spondylometaphyseal dysplasia with cone-rod dystrophy, 608940;Lipodystrophy, congenital generalized, type 5, 620680
PCYT2	100%	100%	100%	98.9%	Spastic paraplegia 82, autosomal recessive, 618770
PDCD1	100%	100%	100%	98.6%	?Autoimmune disease with susceptibility to mycobacterium tuberculosis, 621004
PDCD10	100%	100%	100%	99.7%	Cerebral cavernous malformations-3, 603285
PDCD6IP	100%	100%	100%	99.6%	?Microcephaly 29, primary, autosomal recessive, 620047

PDE10A	99.5%	97.9%	99.6%	91.8%	Striatal degeneration, autosomal dominant, 616922;Dyskinesia, limb and orofacial, infantile-onset, 616921
PDE11A	100%	100%	100%	99.6%	Pigmented nodular adrenocortical disease, primary, 2, 610475
PDE12	100%	100%	100%	99.4%	
PDE1C	100%	100%	100%	99.7%	?Deafness, autosomal dominant 74, 618140
PDE2A	100%	100%	100%	99.2%	Intellectual developmental disorder with paroxysmal dyskinesia or seizures, 619150
PDE3A	100%	100%	100%	99.3%	Hypertension and brachydactyly syndrome, 112410
PDE4D	100%	100%	100%	99.2%	Acrodysostosis 2, with or without hormone resistance, 614613
PDE6A	100%	100%	100%	99.5%	Retinitis pigmentosa 43, 613810
PDE6B	100%	100%	100%	98.8%	Retinitis pigmentosa-40, 613801;Night blindness, congenital stationary, autosomal dominant 2, 163500
PDE6C	100%	100%	100%	99.7%	Cone dystrophy 4, 613093
PDE6D	100%	100%	100%	99.6%	Joubert syndrome 22, 615665

PDE6G	100%	100%	100%	99.8%	Retinitis pigmentosa 57, 613582
PDE6H	100%	100%	100%	98.9%	Retinal cone dystrophy 3, 610024;Achromatopsia 6, 610024
PDE8B	100%	100%	100%	99.4%	Pigmented nodular adrenocortical disease, primary, 3, 614190;Striatal degeneration, autosomal dominant, 609161
PDGFB	100%	100%	100%	99.2%	Meningioma, SIS-related, 607174;Basal ganglia calcification, idiopathic, 5, 615483;Dermatofibrosarcoma protuberans, 607907
PDGFRA	100%	100%	100%	99.4%	Gastrointestinal stromal tumor/GIST-plus syndrome, somatic or familial, 175510;Hypereosinophilic syndrome, idiopathic, resistant to imatinib, 607685
PDGFRB	100%	100%	100%	99%	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

PDGFRL	100%	100%	100%	98.4%	Hepatocellular cancer, somatic, 114550;Colorectal cancer, somatic, 114500
PDHA1	99.1%	93.7%	98.6%	71.4%	Pyruvate dehydrogenase E1-alpha deficiency, 312170
PDHA2	100%	100%	100%	99.6%	Spermatogenic failure 70, 619828
PDHB	100%	100%	100%	99.7%	Pyruvate dehydrogenase E1-beta deficiency, 614111
PDHX	100%	100%	100%	99.3%	Lacticacidemia due to PDX1 deficiency, 245349
PDIA6	100%	100%	100%	99.4%	
PDK1	100%	100%	100%	99.6%	
PDK2	100%	100%	100%	98.8%	
PDK3	100%	99.7%	99.5%	73.1%	?Charcot-Marie-Tooth disease, X-linked dominant, 6, 300905
PDK4	100%	100%	100%	99.8%	
PDLIM3	100%	100%	100%	99.8%	
PDLIM5	99.5%	97.3%	100%	99.3%	
PDP1	100%	100%	100%	99.7%	Pyruvate dehydrogenase phosphatase deficiency, 608782
PDSS1	100%	100%	100%	99.5%	Coenzyme Q10 deficiency, primary, 2, 614651
PDSS2	100%	100%	100%	99.3%	Coenzyme Q10 deficiency, primary, 3, 614652

PDX1	100%	100%	100%	98.5%	{Diabetes mellitus, type II, susceptibility to}, 125853;Pancreatic agenesis 1, 260370;MODY, type IV, 606392
PDXK	99.9%	98%	100%	99%	Neuropathy, hereditary motor and sensory, type VIC, with optic atrophy, 618511
PDYN	100%	100%	100%	99.6%	Spinocerebellar ataxia 23, 610245
PDZD7	99.9%	99.1%	100%	99%	Deafness, autosomal recessive 57, 618003;{Retinal disease in Usher syndrome type IIA, modifier of}, 276901;Usher syndrome, type IIC, GPR98/PDZD7 digenic, 605472
PDZD8	100%	100%	100%	97.3%	Intellectual developmental disorder with autism and dysmorphic facies, 620021
PEPD	93.9%	93.9%	100%	99.4%	Prolidase deficiency, 170100
PER2	100%	100%	100%	99.3%	?Advanced sleep phase syndrome, familial, 1, 604348
PER3	100%	100%	100%	99.1%	?Advanced sleep phase syndrome, familial, 3, 616882
PERCC1	100%	100%	100%	99.3%	Diarrhea 11, malabsorptive, congenital, 618662

PERP	100%	100%	100%	98.5%	Erythrokeratodermia variabilis et progressiva 7, 619209;Olmsted syndrome 2, 619208
PET100	100%	100%	100%	97.8%	Mitochondrial complex IV deficiency, nuclear type 12, 619055
PET117	100%	100%	100%	98%	?Mitochondrial complex IV deficiency, nuclear type 19, 619063
PEX1	100%	100%	100%	99.8%	Heimler syndrome 1, 234580;Peroxisome biogenesis disorder 1B (NALD/IRD), 601539;Peroxisome biogenesis disorder 1A (Zellweger), 214100
PEX10	100%	100%	100%	99.6%	Peroxisome biogenesis disorder 6A (Zellweger), 614870;Peroxisome biogenesis disorder 6B, 614871
PEX11B	100%	100%	100%	98.8%	Peroxisome biogenesis disorder 14B, 614920
PEX12	100%	100%	100%	99.7%	Peroxisome biogenesis disorder 3B, 266510;Peroxisome biogenesis disorder 3A (Zellweger), 614859

PEX13	100%	100%	100%	99.7%	Peroxisome biogenesis disorder 11A (Zellweger), 614883;Peroxisome biogenesis disorder 11B, 614885
PEX14	100%	100%	100%	98.4%	Peroxisome biogenesis disorder 13A (Zellweger), 614887
PEX16	100%	100%	100%	99.1%	Peroxisome biogenesis disorder 8B, 614877;Peroxisome biogenesis disorder 8A (Zellweger), 614876
PEX19	100%	100%	100%	99.5%	Peroxisome biogenesis disorder 12A (Zellweger), 614886
PEX2	100%	100%	100%	99.7%	Peroxisome biogenesis disorder 5A (Zellweger), 614866;Peroxisome biogenesis disorder 5B, 614867
PEX26	100%	100%	100%	99.3%	Peroxisome biogenesis disorder 7B, 614873;Peroxisome biogenesis disorder 7A (Zellweger), 614872
PEX3	100%	100%	100%	99.8%	Peroxisome biogenesis disorder 10A (Zellweger), 614882;?Peroxisome biogenesis disorder 10B, 617370

PEX5	100%	100%	100%	98.5%	Peroxisome biogenesis disorder 2B, 202370;Peroxisome biogenesis disorder 2A (Zellweger), 214110;Rhizomelic chondrodysplasia punctata, type 5, 616716
PEX6	100%	100%	100%	98.5%	Peroxisome biogenesis disorder 4B, 614863;Peroxisome biogenesis disorder 4A (Zellweger), 614862;Heimler syndrome 2, 616617
PEX7	97.9%	97.9%	100%	99.6%	Rhizomelic chondrodysplasia punctata, type 1, 215100;Peroxisome biogenesis disorder 9B, 614879
PFKM	100%	100%	100%	99.4%	Glycogen storage disease VII, 232800
PFN1	100%	100%	100%	98.7%	Amyotrophic lateral sclerosis 18, 614808
PGAM2	100%	100%	100%	99.5%	Glycogen storage disease X, 261670
PGAP1	100%	100%	100%	99.6%	Neurodevelopmental disorder with dysmorphic features, spasticity, and brain abnormalities, 615802

PGAP2	100%	100%	100%	99.3%	Hyperphosphatasia with impaired intellectual development syndrome 3, 614207
PGAP3	100%	100%	100%	99.6%	Hyperphosphatasia with impaired intellectual development syndrome 4, 615716
PGK1	100%	98.1%	98.9%	73.1%	Phosphoglycerate kinase 1 deficiency, 300653
PGM1	94%	94%	100%	99.4%	Congenital disorder of glycosylation, type It, 614921
PGM2L1	100%	100%	100%	99.9%	Neurodevelopmental disorder with hypotonia, dysmorphic facies, and skin abnormalities, 620191
PGM3	100%	100%	100%	99.6%	Immunodeficiency 23, 615816
PHACTR1	100%	99.9%	100%	97.6%	Developmental and epileptic encephalopathy 70, 618298
PHC1	100%	100%	100%	98.9%	?Microcephaly 11, primary, autosomal recessive, 615414
PHEX	100%	99.8%	99.3%	75.2%	Hypophosphatemic rickets, X-linked dominant, 307800

PHF21A	100%	100%	100%	99.3%	Intellectual developmental disorder with behavioral abnormalities and craniofacial dysmorphism with or without seizures, 618725
PHF5A	100%	100%	100%	99.6%	
PHF6	100%	100%	99.6%	76.3%	Borjeson-Forssman-Lehmann syndrome, 301900
PHF8	100%	99.7%	98.9%	70.6%	Intellectual developmental disorder, X-linked syndromic, Siderius type, 300263
PHGDH	100%	100%	100%	99.4%	Neu-Laxova syndrome 1, 256520;Phosphoglycerate dehydrogenase deficiency, 601815
PHIP	99.4%	99%	100%	98.7%	Chung-Jansen syndrome, 617991
PHKA1	100%	99.8%	99.3%	74.3%	Muscle glycogenosis, 300559
PHKA2	100%	99.4%	99.1%	72.2%	Glycogen storage disease, type IXa2, 306000;Glycogen storage disease, type IXa1, 306000
PHKB	100%	100%	100%	99.8%	Phosphorylase kinase deficiency of liver and muscle, autosomal recessive, 261750
PHKG1	100%	100%	100%	99.1%	

PHKG2	100%	100%	100%	98.9%	Glycogen storage disease IXc, 613027
PHLDB1	100%	100%	100%	98.6%	Osteogenesis imperfecta, type XXIII, 620639
PHOX2A	100%	99.9%	100%	98%	Fibrosis of extraocular muscles, congenital, 2, 602078
PHOX2B	100%	100%	100%	97.6%	{Neuroblastoma, susceptibility to, 2}, 613013;Neuroblastoma with Hirschsprung disease, 613013;Central hypoventilation syndrome, congenital, 1, with or without Hirschsprung disease, 209880
PHYH	100%	100%	100%	99.2%	Refsum disease, 266500
PI4K2A	100%	100%	100%	98.5%	Neurodevelopmental disorder with hyperkinetic movements, seizures and structural brain abnormalities, 620732
PI4KA	100%	99.7%	100%	99%	Spastic paraplegia 84, autosomal recessive, 619621;Gastrointestinal defects and immunodeficiency syndrome 2, 619708;Polymicrogyria, perisylvian, with cerebellar hypoplasia and arthrogryposis, 616531

PI4KB	100%	100%	100%	99.1%	Deafness, autosomal dominant 87, 620281
PIBF1	100%	100%	100%	99.7%	Joubert syndrome 33, 617767
PICALM	100%	100%	100%	99.7%	Leukemia, acute myeloid, somatic, 601626
PICK1	100%	100%	100%	99.2%	
PIDD1	100%	100%	100%	99.2%	Intellectual developmental disorder, autosomal recessive 75, with neuropsychiatric features and variant lissencephaly, 619827
PIEZO1	100%	100%	100%	99%	[ER blood group system], 620207;Lymphatic malformation 6, 616843;Dehydrated hereditary stomatocytosis with or without pseudohyperkalemia and/or perinatal edema, 194380
PIEZO2	100%	100%	100%	99.4%	Arthrogryposis, distal, type 5, 108145;Arthrogryposis, distal, with impaired proprioception and touch, 617146;Arthrogryposis, distal, type 3, 114300;?Marden-Walker syndrome, 248700

PIGA	100%	99.9%	98.4%	74%	Paroxysmal nocturnal hemoglobinuria, somatic, 300818;Multiple congenital anomalies-hypotonia-seizures syndrome 2, 300868;Neurodevelopmental disorder with epilepsy and hemochromatosis, 301072
PIGB	100%	100%	100%	99.7%	Developmental and epileptic encephalopathy 80, 618580
PIGC	100%	100%	100%	99.9%	Glycosylphosphatidylinositol biosynthesis defect 16, 617816
PIGF	100%	100%	100%	99.9%	Onychodystrophy, osteodystrophy, impaired intellectual development, and seizures syndrome, 619356
PIGG	100%	100%	100%	99.4%	[Blood group, EMM system], 619812;Neurodevelopmental disorder with or without hypotonia, seizures, and cerebellar atrophy, 616917
PIGH	83.1%	75.6%	100%	98.4%	Glycosylphosphatidylinositol biosynthesis defect 17, 618010
PIGK	100%	100%	100%	99.8%	Neurodevelopmental disorder with hypotonia and cerebellar atrophy, with or without seizures, 618879
PIGL	100%	100%	100%	99.7%	CHIME syndrome, 280000

PIGM	100%	100%	100%	99.7%	Glycosylphosphatidylinositol deficiency, 610293
PIGN	100%	100%	100%	99.7%	Multiple congenital anomalies-hypotonia-seizures syndrome 1, 614080
PIGO	100%	100%	100%	99.3%	Hyperphosphatasia with impaired intellectual development syndrome 2, 614749
PIGP	100%	100%	100%	99.8%	Developmental and epileptic encephalopathy 55, 617599
PIGQ	100%	100%	100%	99.1%	Multiple congenital anomalies-hypotonia-seizures syndrome 4, 618548
PIGS	98.3%	94.6%	100%	98.8%	Developmental and epileptic encephalopathy 95, 618143
PIGT	100%	100%	100%	99.1%	?Paroxysmal nocturnal hemoglobinuria 2, 615399;Multiple congenital anomalies-hypotonia-seizures syndrome 3, 615398
PIGU	100%	100%	100%	99.3%	Neurodevelopmental disorder with brain anomalies, seizures, and scoliosis, 618590
PIGV	100%	99.8%	100%	99.7%	Hyperphosphatasia with impaired intellectual development syndrome 1, 239300

PIGW	100%	100%	100%	99.7%	Glycosylphosphatidylinositol biosynthesis defect 11, 616025
PIGY	100%	100%	100%	99.9%	Hyperphosphatasia with impaired intellectual development syndrome 6, 616809
PIK3C2A	100%	100%	100%	99.4%	Oculoskeletodental syndrome, 618440

PIK3CA	100%	100%	100%	99.7%	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;Nonsmall cell lung cancer, somatic, 211980;Megalencephaly-capillary malformation-polymicrogyria syndrome, somatic, 602501;Cowden syndrome 5, 615108
PIK3CD	100%	100%	100%	98.8%	Immunodeficiency 14A, autosomal dominant, 615513;Immunodeficiency 14B, autosomal recessive, 619281;?Roifman-Chitayat syndrome, digenic, 613328
PIK3CG	100%	100%	100%	99.3%	Immunodeficiency 97 with autoinflammation, 619802

PIK3R1	100%	99.9%	100%	99.9%	Immunodeficiency 36, 616005;?Agammaglobulinemia 7, autosomal recessive, 615214;SHORT syndrome, 269880
PIK3R2	100%	100%	100%	98.6%	Megalencephaly-polymicrogyria-polydactyly-hydrocephalus syndrome 1, 603387
PIK3R5	100%	100%	100%	98.6%	Ataxia-oculomotor apraxia 3, 615217
PIKFYVE	100%	100%	100%	99.7%	Corneal fleck dystrophy, 121850
PINK1	100%	100%	100%	98.6%	Parkinson disease 6, early onset, 605909
PIP5K1C	100%	100%	100%	98.9%	Lethal congenital contractural syndrome 3, 611369
PISD	100%	100%	100%	99.4%	Liberfarb syndrome, 618889
PITPNM3	100%	100%	100%	98%	Cone-rod dystrophy 5, 600977
PITRM1	100%	100%	100%	99.5%	Spinocerebellar ataxia, autosomal recessive 30, 619405
PITX1	100%	100%	100%	95.8%	Clubfoot, congenital, with or without deficiency of long bones and/or mirror-image polydactyly, 119800

PITX2	100%	100%	100%	97.9%	Ring dermoid of cornea, 180550;Axenfeld-Rieger syndrome, type 1, 180500;Anterior segment dysgenesis 4, 137600
PITX3	100%	100%	100%	98%	Cataract 11, multiple types, 610623;Anterior segment dysgenesis 1, multiple subtypes, 107250;Cataract 11, syndromic, autosomal recessive, 610623
PIWIL2	100%	100%	100%	99.5%	
PJA1	100%	99.9%	98.4%	69.2%	
PJVK	100%	100%	100%	99.7%	Deafness, autosomal recessive 59, 610220
PKD1	99.9%	99.6%	100%	98.3%	Polycystic kidney disease 1, 173900
PKD1L1	100%	100%	100%	99.4%	Heterotaxy, visceral, 8, autosomal, 617205
PKD2	100%	100%	100%	97.4%	Polycystic kidney disease 2, 613095
PKDCC	100%	100%	99.7%	88.9%	Rhizomelic limb shortening with dysmorphic features, 618821
PKHD1	100%	100%	100%	99.5%	Polycystic kidney disease 4, with or without hepatic disease, 263200
PKHD1L1	100%	100%	100%	99.8%	Deafness, autosomal recessive 124, 620794

PKLR	100%	100%	100%	98.9%	Anemia, congenital, nonspherocytic hemolytic, 2, pyruvate kinase deficient, 266200;[Adenosine triphosphate, elevated, of erythrocytes], 102900
PKP1	100%	100%	100%	99.4%	Ectodermal dysplasia/skin fragility syndrome, 604536
PKP2	99%	97.1%	100%	99.3%	Arrhythmogenic right ventricular dysplasia 9, 609040
PLA2G4A	100%	100%	100%	99.9%	Gastrointestinal ulceration, recurrent, with dysfunctional platelets, 618372
PLA2G5	100%	100%	100%	99.4%	[Fleck retina, familial benign], 228980
PLA2G6	100%	99.7%	100%	99.1%	Parkinson disease 14, autosomal recessive, 612953;Neurodegeneration with brain iron accumulation 2B, 610217;Infantile neuroaxonal dystrophy 1, 256600
PLA2G7	100%	100%	100%	99.9%	Platelet-activating factor acetylhydrolase deficiency, 614278
PLAA	100%	100%	100%	99.8%	Neurodevelopmental disorder with progressive microcephaly, spasticity, and brain anomalies, 617527

PLAAT3	100%	100%	100%	99.4%	Lipodystrophy, familial partial, type 9, 620683
PLAG1	100%	100%	100%	99.7%	Adenomas, salivary gland pleomorphic, somatic, 181030;Silver-Russell syndrome 4, 618907
PLAT	96%	96%	100%	99.6%	
PLAU	100%	100%	100%	99.4%	Quebec platelet disorder, 601709;{Alzheimer disease, late-onset, susceptibility to}, 104300
PLCB1	100%	100%	100%	99.8%	Developmental and epileptic encephalopathy 12, 613722
PLCB3	100%	100%	100%	98.9%	Spondylometaphyseal dysplasia with corneal dystrophy, 618961
PLCB4	99%	99%	100%	99.8%	Auriculocondylar syndrome 2B, 620458;Auriculocondylar syndrome 2A, 614669
PLCD1	100%	100%	100%	99.3%	Nail disorder, nonsyndromic congenital, 3, (leukonychia), 151600
PLCE1	100%	100%	100%	99.6%	Nephrotic syndrome, type 3, 610725
PLCG1	100%	100%	100%	99.3%	?Immune dysregulation, autoimmunity, and autoinflammation, 620514

PLCG2	100%	100%	100%	99.3%	Autoinflammation, antibody deficiency, and immune dysregulation syndrome, 614878;Familial cold autoinflammatory syndrome 3, 614468
PLCH1	100%	100%	100%	99.7%	Holoprosencephaly 14, 619895
PLCZ1	100%	100%	100%	98.4%	Spermatogenic failure 17, 617214
PLD1	100%	100%	100%	99.7%	Cardiac valvular dysplasia 1, 212093
PLD3	100%	100%	100%	98.4%	?Spinocerebellar ataxia 46, 617770
PLD6	100%	100%	100%	99.1%	
PLEC	100%	100%	100%	98.4%	?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

PLEKHG2	100%	100%	100%	98.9%	Leukodystrophy and acquired microcephaly with or without dystonia, 616763
PLEKHG5	100%	100%	100%	98.9%	Neuronopathy, distal hereditary motor, autosomal recessive 4, 611067;Charcot-Marie-Tooth disease, recessive intermediate C, 615376
PLEKHM1	100%	100%	100%	98.6%	?Osteopetrosis, autosomal recessive 6, 611497;Osteopetrosis, autosomal dominant 3, 618107
PLEKHM2	100%	100%	100%	98.8%	
PLG	100%	100%	100%	99.6%	Dysplasminogenemia, 217090;Angioedema, hereditary, 4, 619360;Plasminogen deficiency, type I, 217090
PLIN1	100%	100%	100%	98.8%	Lipodystrophy, familial partial, type 4, 613877
PLIN4	99.7%	99.7%	100%	98.2%	Myopathy with rimmed ubiquitin-positive autophagic vacuolation, autosomal dominant, 601846
PLK1	100%	100%	100%	99.2%	
PLK4	100%	100%	100%	99.5%	Microcephaly and chorioretinopathy, autosomal recessive, 2, 616171

PLN	100%	100%	100%	99.1%	Cardiomyopathy, dilated, 1P, 609909;Cardiomyopathy, hypertrophic, 18, 613874
PLOD1	100%	100%	100%	98.4%	Ehlers-Danlos syndrome, kyphoscoliotic type, 1, 225400
PLOD2	100%	100%	100%	99.7%	Bruck syndrome 2, 609220
PLOD3	100%	100%	100%	98.4%	BCARD syndrome (lysyl hydroxylase 3 deficiency), 612394
PLP1	100%	99.6%	99.1%	71.8%	Pelizaeus-Merzbacher disease, 312080;Spastic paraplegia 2, X-linked, 312920
PLPBP	100%	100%	100%	98.9%	Epilepsy, early-onset, 1, vitamin B6-dependent, 617290
PLPP6	100%	100%	100%	97.7%	
PLS1	100%	100%	100%	99.8%	Deafness, autosomal dominant 76, 618787
PLS3	96.8%	96.6%	99%	73.5%	Bone mineral density QTL18, osteoporosis, 300910;Diaphragmatic hernia 5, X-linked, 306950
PLVAP	100%	100%	100%	98.5%	Diarrhea 10, protein-losing enteropathy type, 618183
PLXNA1	100%	100%	100%	99.4%	Dworschak-Punetha neurodevelopmental syndrome, 619955

PLXNA2	100%	100%	100%	99.3%	
PLXNB2	100%	100%	100%	98.4%	
PLXND1	100%	99.9%	100%	98.7%	Congenital heart defects, multiple types, 9, 620294
PMEPA1	99.7%	95.6%	100%	94.8%	
PMFBP1	100%	100%	100%	99.6%	Spermatogenic failure 31, 618112
PML	100%	100%	100%	99%	
PMM2	100%	100%	100%	99.7%	Congenital disorder of glycosylation, type Ia, 212065
PMP2	100%	100%	100%	99.8%	Charcot-Marie-Tooth disease, demyelinating, type 1G, 618279
PMP22	100%	100%	100%	99.5%	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
PMPCA	96%	96%	100%	99.1%	Spinocerebellar ataxia, autosomal recessive 2, 213200
PMPCB	91.4%	91.4%	100%	99.7%	Multiple mitochondrial dysfunctions syndrome 6, 617954

PMS2	93.7%	93.4%	100%	98.8%	Lynch syndrome 4, 614337;Mismatch repair cancer syndrome 4, 619101
PMS2CL					
PMVK	100%	100%	100%	99.2%	Porokeratosis 1, multiple types, 175800
PNKD	100%	100%	100%	98.7%	Paroxysmal nonkinesigenic dyskinesia 1, 118800
PNKP	100%	100%	100%	97.9%	?Charcot-Marie-Tooth disease, type 2B2, 605589;Ataxia-oculomotor apraxia 4, 616267;Microcephaly, seizures, and developmental delay, 613402
PNLDC1	100%	100%	100%	99.5%	Spermatogenic failure 57, 619528
PNLIP	100%	100%	100%	99.5%	?Pancreatic lipase deficiency, 614338
PNMT	100%	100%	100%	99.7%	
PNP	100%	100%	100%	99.2%	Immunodeficiency due to purine nucleoside phosphorylase deficiency, 613179
PNPLA1	100%	100%	100%	99.5%	Ichthyosis, congenital, autosomal recessive 10, 615024
PNPLA2	100%	100%	100%	98.6%	Neutral lipid storage disease with myopathy, 610717

PNPLA6	100%	100%	100%	98.8%	Spastic paraplegia 39, autosomal recessive, 612020;Oliver-McFarlane syndrome, 275400;?Laurence-Moon syndrome, 245800;Boucher-Neuhauser syndrome, 215470
PNPLA8	100%	100%	100%	99.7%	?Mitochondrial myopathy with lactic acidosis, 251950
PNPO	100%	100%	100%	99.1%	Pyridoxamine 5'-phosphate oxidase deficiency, 610090
PNPT1	100%	100%	100%	99.8%	Spinocerebellar ataxia 25, 608703;Deafness, autosomal recessive 70, with or without adult-onset neurodegeneration, 614934;Combined oxidative phosphorylation deficiency 13, 614932
POC1A	100%	100%	100%	99.4%	Short stature, onychodysplasia, facial dysmorphism, and hypotrichosis, 614813
POC1B	100%	100%	100%	99.8%	Cone-rod dystrophy 20, 615973
POC5	100%	100%	100%	99.8%	
PODXL	93.8%	93.8%	100%	98.4%	
POF1B	100%	99.8%	99.3%	75.1%	?Premature ovarian failure 2B, 300604

POFUT1	100%	100%	100%	99.2%	Dowling-Degos disease 2, 615327
POGLUT1	100%	100%	100%	99.8%	Dowling-Degos disease 4, 615696;Muscular dystrophy, limb-girdle, autosomal recessive 21, 617232
POGZ	100%	99.5%	100%	99.4%	White-Sutton syndrome, 616364
POLA1	100%	99.8%	99.2%	74.4%	Pigmentary disorder, reticulate, with systemic manifestations, X-linked, 301220;Van Esch-O'Driscoll syndrome, 301030
POLD1	100%	100%	100%	98.3%	Mandibular hypoplasia, deafness, progeroid features, and lipodystrophy syndrome, 615381;Immunodeficiency 120, 620836;{Colorectal cancer, susceptibility to, 10}, 612591
POLD3	100%	100%	100%	99.8%	Immunodeficiency 122, 620869
POLE	100%	100%	100%	99%	{Colorectal cancer, susceptibility to, 12}, 615083;FILS syndrome, 615139;IMAGE-I syndrome, 618336
POLE2	100%	100%	100%	99.9%	

POLG	100%	100%	100%	99.5%	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
POLG2	100%	100%	100%	99.8%	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
POLH	100%	100%	100%	99.6%	Xeroderma pigmentosum, variant type, 278750
POLL	100%	100%	100%	99.4%	

POLR1A	100%	100%	100%	99.4%	Leukodystrophy, hypomyelinating, 27, 620675;Acrofacial dysostosis, Cincinnati type, 616462
POLR1B	100%	100%	100%	99.8%	Treacher-Collins syndrome 4, 618939
POLR1C	83.5%	83.2%	100%	98.8%	Leukodystrophy, hypomyelinating, 11, 616494;Treacher Collins syndrome 3, 248390
POLR1D	100%	100%	100%	99.8%	Treacher Collins syndrome 2, 613717
POLR2A	100%	100%	100%	97.7%	Neurodevelopmental disorder with hypotonia and variable intellectual and behavioral abnormalities, 618603
POLR3A	100%	100%	100%	99.3%	Wiedemann-Rautenstrauch syndrome, 264090;Leukodystrophy, hypomyelinating, 7, with or without oligodontia and/or hypogonadotropic hypogonadism, 607694

POLR3B	100%	100%	100%	99.8%	Leukodystrophy, hypomyelinating, 8, with or without oligodontia and/or hypogonadotropic hypogonadism, 614381;Charcot-Marie-Tooth disease, demyelinating, type 1I, 619742
POLR3F	91.6%	90.5%	100%	99.9%	?Immunodeficiency 101 (varicella zoster virus-specific), 619872
POLR3GL	100%	100%	100%	99.4%	Short stature, oligodontia, dysmorphic facies, and motor delay, 619234
POLR3K	100%	100%	100%	99.6%	Leukodystrophy, hypomyelinating, 21, 619310
POLRMT	100%	100%	100%	98.6%	Combined oxidative phosphorylation deficiency 55, 619743
POMC	100%	100%	100%	99.5%	{Obesity, early-onset, susceptibility to}, 601665;Obesity, adrenal insufficiency, and red hair due to POMC deficiency, 609734

POMGNT1	100%	100%	100%	99%	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
POMGNT2	100%	100%	100%	98.9%	Muscular dystrophy-dystroglycanopathy (congenital with brain and eye anomalies), type A, 8, 614830;Muscular dystrophy-dystroglycanopathy (limb-girdle) type C, 8, 618135
POMK	100%	100%	100%	99.1%	?Muscular dystrophy-dystroglycanopathy (limb-girdle), type C, 12, 616094;Muscular dystrophy-dystroglycanopathy (congenital with brain and eye anomalies), type A, 12, 615249

POMP	83.2%	83.2%	100%	99.9%	Proteasome-associated autoinflammatory syndrome 2, 618048;Keratosis linearis with ichthyosis congenita and sclerosing keratoderma, 601952
POMT1	100%	100%	100%	99%	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
POMT2	100%	100%	99.9%	98.1%	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

POP1	100%	100%	100%	99.6%	Anauxetic dysplasia 2, 617396
POPDC3	100%	100%	100%	99.4%	Muscular dystrophy, limb-girdle, autosomal recessive 26, 618848
POR	100%	100%	100%	99.3%	Antley-Bixler syndrome with genital anomalies and disordered steroidogenesis, 201750;Disordered steroidogenesis due to cytochrome P450 oxidoreductase, 613571
PORCN	100%	99.5%	98.3%	69.5%	Focal dermal hypoplasia, 305600
POT1	100%	100%	100%	99.8%	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
POU1F1	100%	100%	100%	99.6%	Pituitary hormone deficiency, combined or isolated, 1, 613038
POU2AF1	100%	100%	100%	99.1%	
POU3F2	100%	100%	100%	97.8%	
POU3F3	98.2%	90.4%	98.3%	73.8%	Snijders Blok-Fisher syndrome, 618604

POU3F4	100%	100%	98.4%	68%	Deafness, X-linked 2, 304400
POU4F1	93%	86.6%	99.9%	91.4%	Ataxia, intention tremor, and hypotonia syndrome, childhood-onset, 619352
POU4F3	100%	100%	100%	98.6%	Deafness, autosomal dominant 15/52, 602459
POU6F2	100%	100%	100%	99.2%	{Wilms tumor susceptibility-5}, 601583
PPA2	100%	100%	100%	99.5%	?Sudden cardiac failure, alcohol-induced, 617223;Sudden cardiac failure, infantile, 617222
PPARG	100%	100%	100%	99.1%	{Diabetes, type 2}, 125853;Insulin resistance, severe, digenic, 125853;Lipodystrophy, familial partial, type 3, 604367;Obesity, severe, 601665
PPCDC	100%	100%	100%	99.4%	
PPCS	100%	100%	100%	99.2%	Cardiomyopathy, dilated, 2C, 618189
PPFIA3	100%	100%	100%	98.4%	
PPFIBP1	100%	100%	100%	99.7%	Neurodevelopmental disorder with seizures, microcephaly, and brain abnormalities, 620024
PPIB	100%	100%	100%	98.5%	Osteogenesis imperfecta, type IX, 259440

PPIL1	100%	100%	100%	99.8%	Pontocerebellar hypoplasia, type 14, 619301
PPIP5K2	100%	100%	100%	99.6%	Deafness, autosomal recessive 100, 618422
PPM1D	100%	100%	100%	99.5%	Breast cancer, somatic, 114480;Jansen-de Vries syndrome, 617450
PPM1K	100%	100%	100%	99.6%	Maple syrup urine disease, mild variant, 615135
PPOX	100%	100%	100%	99%	Variegate porphyria, childhood-onset, 620483;Variegate porphyria, 176200
PPP1CB	88%	87.4%	100%	99.6%	Noonan syndrome-like disorder with loose anagen hair 2, 617506
PPP1R12A	100%	100%	100%	99.7%	Genitourinary and/or/brain malformation syndrome, 618820
PPP1R13L	100%	100%	100%	96.2%	Arrhythmogenic cardiomyopathy with variable ectodermal abnormalities, 620519
PPP1R15B	100%	100%	100%	99.7%	Microcephaly, short stature, and impaired glucose metabolism 2, 616817
PPP1R21	100%	100%	100%	99.7%	Neurodevelopmental disorder with hypotonia, facial dysmorphism, and brain abnormalities, 619383

PPP1R3A	100%	100%	100%	99.7%	Insulin resistance, severe, digenic, 125853
PPP1R3F	100%	99.9%	97.9%	67.5%	
PPP2CA	100%	100%	100%	99.7%	Houge-Janssens syndrome 3, 618354
PPP2R1A	93.9%	93.9%	100%	99.4%	Houge-Janssens syndrome 2, 616362
PPP2R1B	100%	100%	100%	99.8%	Lung cancer, somatic, 211980
PPP2R2B	100%	100%	100%	99.5%	Spinocerebellar ataxia 12, 604326
PPP2R3C	100%	100%	100%	99.6%	Spermatogenic failure 36, 618420;Myoectodermal gonadal dysgenesis syndrome, 618419
PPP2R5B	100%	100%	100%	99%	
PPP2R5C	100%	99.9%	100%	99.3%	
PPP2R5D	100%	100%	100%	98.2%	Houge-Janssens syndrome 1, 616355
PPP3CA	100%	100%	100%	99.4%	Arthrogryposis, cleft palate, craniosynostosis, and impaired intellectual development, 618265;Developmental and epileptic encephalopathy 91, 617711
PPT1	90.3%	90.3%	100%	99.7%	Ceroid lipofuscinosis, neuronal, 1, 256730
PQBP1	100%	99.9%	98.5%	66.1%	Renpenning syndrome, 309500

PRCC	100%	100%	100%	99.1%	Renal cell carcinoma, papillary, 605074
PRCD	100%	100%	100%	99.2%	Retinitis pigmentosa 36, 610599
PRDM10	100%	100%	100%	99.1%	?Birt-Hogg-Dube syndrome 2, 620459
PRDM12	94.3%	91.2%	100%	98.2%	Neuropathy, hereditary sensory and autonomic, type VIII, 616488
PRDM13	100%	100%	100%	98.1%	Pontocerebellar hypoplasia, type 17, 619909;Cerebellar dysfunction, impaired intellectual development, and hypogonadotropic hypogonadism, 619761
PRDM15	100%	100%	100%	98.8%	
PRDM16	100%	100%	100%	97.8%	Left ventricular noncompaction 8, 615373;Cardiomyopathy, dilated, 1LL, 615373
PRDM5	100%	100%	100%	99.6%	Brittle cornea syndrome 2, 614170
PRDM6	100%	100%	99.9%	96.3%	Patent ductus arteriosus 3, 617039
PRDM8	100%	100%	99.9%	93.3%	?Epilepsy, progressive myoclonic, 10, 616640
PRDX1	100%	100%	100%	99.4%	Methylmalonic aciduria and homocystinuria, cblC type, digenic, 277400
PRDX2	100%	100%	100%	99.5%	

PRDX3	100%	100%	100%	99.8%	Spinocerebellar ataxia, autosomal recessive 32, 619862;Corneal dystrophy, punctiform and polychromatic pre-Descemet, 619871
PREPL	100%	100%	100%	99.6%	Myasthenic syndrome, congenital, 22, 616224
PRF1	100%	100%	100%	99%	Hemophagocytic lymphohistiocytosis, familial, 2, 603553;Aplastic anemia, 609135;Lymphoma, non-Hodgkin, 605027
PRG4	100%	100%	100%	94.3%	Camptodactyly-arthropathy-coxa vara-pericarditis syndrome, 208250
PRICKLE1	100%	100%	100%	99.6%	Epilepsy, progressive myoclonic 1B, 612437
PRICKLE2	100%	100%	100%	99%	
PRIM1	91.8%	91.8%	100%	99.4%	Primordial dwarfism-immunodeficiency-lipodystrophy syndrome, 620005
PRIMPOL	100%	100%	100%	99.3%	Myopia 22, autosomal dominant, 615420
PRKAA1	100%	100%	100%	99.7%	
PRKACA	100%	99.9%	100%	99.1%	Cushing syndrome, ACTH-independent adrenal, somatic, 615830;Cardioacrofacial dysplasia 1, 619142

PRKACB	100%	100%	100%	99.5%	Cardioacrofacial dysplasia 2, 619143
PRKACG	100%	100%	100%	98.7%	?Bleeding disorder, platelet-type, 19, 616176
PRKAG2	100%	100%	100%	98.7%	Glycogen storage disease of heart, lethal congenital, 261740;Wolff-Parkinson-White syndrome, 194200;Cardiomyopathy, hypertrophic 6, 600858
PRKAR1A	100%	100%	100%	99.4%	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
PRKAR1B	100%	100%	100%	98.4%	Marbach-Schaaf neurodevelopmental syndrome, 619680
PRKCA	100%	100%	100%	99.5%	Pituitary tumor, invasive
PRKCB	100%	100%	100%	99.2%	
PRKCD	100%	100%	100%	99.2%	Autoimmune lymphoproliferative syndrome, type III, 615559
PRKCG	100%	100%	100%	97.5%	Spinocerebellar ataxia 14, 605361

PRKCSH	100%	100%	100%	99.2%	Polycystic liver disease 1, 174050
PRKD1	100%	100%	99.9%	97.2%	Congenital heart defects and ectodermal dysplasia, 617364
PRKDC	100%	100%	100%	99.6%	Immunodeficiency 26, with or without neurologic abnormalities, 615966
PRKG1	95.9%	95.9%	100%	99.3%	Aortic aneurysm, familial thoracic 8, 615436
PRKG2	100%	100%	100%	99.7%	Spondylometaphyseal dysplasia, Pagnamenta type, 619638;Acromesomelic dysplasia 4, 619636
PRKN	100%	100%	100%	99.2%	Adenocarcinoma of lung, somatic, 211980;Parkinson disease, juvenile, type 2, 600116;Ovarian cancer, somatic, 167000
PRKRA	100%	100%	100%	99.8%	Dystonia 16, 612067
PRLR	100%	100%	100%	99.6%	Multiple fibroadenomas of the breast, 615554;Hyperprolactinemia, 615555
PRMT7	100%	100%	100%	98.6%	Short stature, brachydactyly, intellectual developmental disability, and seizures, 617157

PRNP	100%	100%	100%	99.7%	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
PROC	100%	100%	100%	99.3%	Thrombophilia 3 due to protein C deficiency, autosomal dominant, 176860;Thrombophilia 3 due to protein C deficiency, autosomal recessive, 612304
PRODH	100%	100%	100%	99.2%	{Schizophrenia, susceptibility to, 4}, 600850;Hyperprolinemia, type I, 239500
PROK2	100%	100%	100%	99.7%	Hypogonadotropic hypogonadism 4 with or without anosmia, 610628
PROKR2	100%	100%	100%	99.3%	Hypogonadotropic hypogonadism 3 with or without anosmia, 244200

PROM1	100%	100%	100%	99.8%	Macular dystrophy, retinal, 2, 608051;Retinitis pigmentosa 41, 612095;Stargardt disease 4, 603786;Cone-rod dystrophy 12, 612657
PROP1	100%	100%	100%	98.8%	Pituitary hormone deficiency, combined, 2, 262600
PRORP	100%	100%	100%	99.7%	Combined oxidative phosphorylation deficiency 54, 619737
PROS1	100%	100%	100%	99.4%	Thrombophilia 5 due to protein S deficiency, autosomal recessive, 614514;Thrombophilia 5 due to protein S deficiency, autosomal dominant, 612336
PROZ	100%	100%	100%	99.2%	[Protein Z deficiency], 614024
PRPF19	100%	100%	100%	98.9%	
PRPF3	100%	100%	100%	99.2%	Retinitis pigmentosa 18, 601414
PRPF31	100%	100%	100%	99.1%	Retinitis pigmentosa 11, 600138
PRPF4	100%	100%	100%	99.5%	Retinitis pigmentosa 70, 615922
PRPF6	100%	100%	100%	98.7%	Retinitis pigmentosa 60, 613983

PRPF8	100%	100%	100%	99.3%	Retinitis pigmentosa 13, 600059
PRPH2	100%	100%	100%	98.7%	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
PRPS1	100%	99.9%	98.5%	70.4%	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
PRR11	100%	100%	100%	99.7%	
PRR12	100%	100%	100%	97.6%	Neuroocular syndrome, 619539

PRRT2	100%	100%	100%	98.1%	Convulsions, familial infantile, with paroxysmal choreoathetosis, 602066;Seizures, benign familial infantile, 2, 605751;Episodic kinesigenic dyskinesia 1, 128200
PRRX1	100%	100%	100%	98.4%	Agnathia-otocephaly complex, 202650
PRSS1	100%	100%	100%	94.5%	Pancreatitis, hereditary, 167800
PRSS12	100%	100%	100%	98.7%	Intellectual developmental disorder, autosomal recessive 1, 249500
PRSS56	100%	100%	100%	98.9%	Microphthalmia, isolated 6, 613517
PRUNE1	93.1%	93.1%	100%	99.2%	Neurodevelopmental disorder with microcephaly, hypotonia, and variable brain anomalies, 617481
PRX	100%	100%	100%	99.6%	Charcot-Marie-Tooth disease, type 4F, 614895;Dejerine-Sottas disease, 145900
PRY	50%	50%	48.3%	25.5%	
PRY2	50%	50%	49.4%	28.9%	

PSAP	100%	100%	100%	99.3%	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
PSAT1	100%	100%	100%	99.5%	Neu-Laxova syndrome 2, 616038;Phosphoserine aminotransferase deficiency, 610992
PSEN1	100%	100%	100%	99.7%	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
PSEN2	73.5%	73.5%	100%	98.8%	Alzheimer disease-4, 606889;Cardiomyopathy, dilated, 1V, 613697
PSENEN	100%	100%	100%	96.6%	Acne inversa, familial, 2, with or without Dowling-Degos disease, 613736
PSIP1	100%	100%	100%	99.5%	

PSKH1	100%	100%	100%	99.1%	Cholestasis, progressive familial intrahepatic, 13, 620962
PSMA3	100%	100%	100%	99.6%	
PSMB1	100%	100%	100%	99.9%	?Neurodevelopmental disorder with microcephaly, hypotonia, and absent language, 620038
PSMB10	100%	100%	100%	99.5%	Immunodeficiency 121 with autoinflammation, 620807;Proteasome-associated autoinflammatory syndrome 5, 619175
PSMB4	100%	100%	100%	99.8%	?Proteasome-associated autoinflammatory syndrome 3 and digenic forms, 617591
PSMB8	100%	100%	100%	99.7%	Proteasome-associated autoinflammatory syndrome 1 and digenic forms, 256040
PSMB9	100%	100%	100%	99.4%	Proteasome-associated autoinflammatory syndrome 6, 620796
PSMC1	100%	100%	100%	99.7%	?Birk-Aharoni syndrome, 620071
PSMC3	100%	100%	100%	98.9%	?Deafness, cataract, impaired intellectual development, and polyneuropathy, 619354

PSMC3IP	100%	100%	100%	99.8%	Ovarian dysgenesis 3, 614324
PSMC5	100%	100%	100%	99.4%	
PSMD11	100%	100%	100%	99.7%	
PSMD12	100%	100%	100%	99.8%	Stankiewicz-Isidor syndrome, 617516
PSMG2	88.4%	88.4%	100%	99.9%	?Proteasome-associated autoinflammatory syndrome 4, 619183
PSPH	100%	100%	100%	99.6%	Phosphoserine phosphatase deficiency, 614023
PSTPIP1	100%	100%	100%	99.4%	Autoinflammatory syndrome with cytopenia, hyperzincemia, and hypercalprotectinemia, 601979;Pyogenic sterile arthritis, pyoderma gangrenosum, and acne, 604416
PTBP1	100%	100%	100%	97.9%	
PTCD3	100%	100%	100%	99.5%	Combined oxidative phosphorylation deficiency 51, 619057
PTCH1	100%	100%	100%	98.7%	Basal cell nevus syndrome 1, 109400;Basal cell carcinoma, somatic, 605462;Holoprosencephaly 7, 610828

PTCH2	100%	100%	100%	98.9%	Medulloblastoma, somatic, 155255;Basal cell carcinoma, somatic, 605462
PTCHD1	100%	99.9%	98.5%	70.7%	{Autism, susceptibility to, X-linked 4}, 300830
PTCRA	100%	100%	100%	99.1%	Immunodeficiency 126, 620931
PTDSS1	100%	100%	100%	99.4%	Lenz-Majewski hyperostotic dwarfism, 151050
PTEN	94.5%	94.5%	99.9%	96.5%	{Glioma susceptibility 2}, 613028;{Meningioma}, 607174;Cowden syndrome 1, 158350;Lhermitte-Duclos disease, 158350;Prostate cancer, somatic, 176807;Macrocephaly/autism syndrome, 605309
PTF1A	100%	100%	100%	98.1%	Pancreatic and cerebellar agenesis, 609069;Pancreatic agenesis 2, 615935
PTGIS	100%	100%	100%	99%	Hypertension, essential, 145500
PTGS1	100%	100%	100%	99.7%	
PTH	100%	100%	100%	100%	Hypoparathyroidism, familial isolated 1, 146200

PTH1R	100%	100%	100%	98.9%	Metaphyseal chondrodysplasia, Murk Jansen type, 156400;Eiken syndrome, 600002;Failure of tooth eruption, primary, 125350;Chondrodysplasia, Blomstrand type, 215045
PTHLH	100%	100%	100%	99.6%	Brachydactyly, type E2, 613382
PTPA	100%	100%	100%	98.4%	Parkinson disease 25, autosomal recessive early-onset, with impaired intellectual development, 620482
PTPMT1	100%	100%	100%	99.3%	
PTPN11	89.7%	89.2%	100%	99.8%	Noonan syndrome 1, 163950;LEOPARD syndrome 1, 151100;Metachondromatosis, 156250;Leukemia, juvenile myelomonocytic, somatic, 607785
PTPN12	100%	100%	100%	99.7%	Colon cancer, somatic, 114500
PTPN14	100%	100%	100%	99.3%	Choanal atresia and lymphedema, 613611
PTPN22	100%	100%	100%	99.8%	{Rheumatoid arthritis, susceptibility to}, 180300;{Systemic lupus erythematosus susceptibility to}, 152700;{Diabetes, type 1, susceptibility to}, 222100

PTPN23	100%	100%	100%	98.1%	Neurodevelopmental disorder and structural brain anomalies with or without seizures and spasticity, 618890
PTPRC	100%	100%	100%	99.7%	Immunodeficiency 105, severe combined, 619924
PTPRF	100%	100%	100%	99.3%	?Breasts and/or nipples, aplasia or hypoplasia of, 2, 616001
PTPRJ	100%	100%	100%	99.2%	Colon cancer, somatic, 114500;Thrombocytopenia 10, 620484
PTPRO	100%	100%	100%	99.8%	Nephrotic syndrome, type 6, 614196
PTPRQ	100%	100%	100%	99.8%	Deafness, autosomal dominant 73, 617663;Deafness, autosomal recessive 84A, 613391
PTRH2	100%	100%	100%	99.2%	Infantile-onset multisystem neurologic, endocrine, and pancreatic disease, 616263
PTRHD1	100%	100%	100%	99.5%	Neurodevelopmental disorder with early-onset parkinsonism and behavioral abnormalities, 620747
PTS	100%	100%	100%	99.2%	Hyperphenylalaninemia, BH4-deficient, A, 261640
PUF60	100%	100%	100%	98.7%	Verheij syndrome, 615583

PUM1	100%	100%	99.9%	98.1%	Spinocerebellar ataxia 47, 617931;Neurodevelopmental disorder with motor abnormalities, seizures, and facial dysmorphism, 620719
PURA	100%	100%	100%	99.9%	Neurodevelopmental disorder with neonatal respiratory insufficiency, hypotonia, and feeding difficulties, 616158
PUS1	100%	100%	100%	98.9%	Myopathy, lactic acidosis, and sideroblastic anemia 1, 600462
PUS3	100%	100%	100%	99.7%	Neurodevelopmental disorder with microcephaly and gray sclerae, 617051
PUS7	100%	100%	100%	99.6%	Intellectual developmental disorder with abnormal behavior, microcephaly, and short stature, 618342
PXDN	100%	99.5%	100%	99.4%	Anterior segment dysgenesis 7, with sclerocornea, 269400
PYCR1	100%	100%	100%	99.4%	Cutis laxa, autosomal recessive, type IIIB, 614438;Cutis laxa, autosomal recessive, type IIB, 612940
PYCR2	100%	100%	100%	99.4%	Leukodystrophy, hypomyelinating, 10, 616420

PYGL	100%	100%	100%	99.6%	Glycogen storage disease VI, 232700
PYGM	100%	100%	100%	98.6%	McArdle disease, 232600
PYROXD1	100%	100%	100%	99.8%	Myopathy, myofibrillar, 8, 617258
PYROXD2	91.1%	88.5%	100%	98.8%	
QARS1	100%	100%	100%	99%	Microcephaly, progressive, seizures, and cerebral and cerebellar atrophy, 615760
QDPR	100%	100%	100%	99.6%	Hyperphenylalaninemia, BH4-deficient, C, 261630
QRICH1	100%	100%	100%	99.1%	Ververi-Brady syndrome, 617982
QRICH2	100%	100%	100%	99.1%	Spermatogenic failure 35, 618341
QRSL1	100%	100%	100%	99.8%	Combined oxidative phosphorylation deficiency 40, 618835
RAB11A	100%	100%	100%	99.7%	
RAB11B	100%	100%	100%	99.2%	Neurodevelopmental disorder with ataxic gait, absent speech, and decreased cortical white matter, 617807
RAB14	100%	100%	100%	98.8%	
RAB18	100%	100%	100%	99.8%	Warburg micro syndrome 3, 614222
RAB23	100%	100%	100%	99.6%	Carpenter syndrome, 201000

RAB27A	100%	100%	100%	99.5%	Griscelli syndrome, type 2, 607624
RAB28	100%	100%	100%	99.3%	Cone-rod dystrophy 18, 615374
RAB33B	100%	100%	100%	99.6%	Smith-McCort dysplasia 2, 615222
RAB34	100%	100%	100%	98.5%	Orofaciodigital syndrome XX, 620718
RAB39B	100%	100%	99.4%	72.6%	Intellectual developmental disorder, X-linked 72, 300271;Waisman syndrome, 311510
RAB3GAP1	100%	100%	100%	99.7%	Martsolf syndrome 2, 619420;Warburg micro syndrome 1, 600118
RAB3GAP2	94.4%	94.4%	100%	99.5%	Martsolf syndrome 1, 212720;Warburg micro syndrome 2, 614225
RAB5C	100%	100%	100%	98%	
RAB5IF	100%	100%	100%	98.6%	?Craniofacial dysmorphism, skeletal anomalies, and impaired intellectual development syndrome 2, 616994
RAB7A	100%	99.3%	100%	99.7%	Charcot-Marie-Tooth disease, type 2B, 600882
RABGAP1	100%	100%	100%	99.5%	
RAC1	86.4%	86.4%	100%	99.6%	Intellectual developmental disorder, autosomal dominant 48, 617751

RAC2	100%	100%	100%	97.9%	Immunodeficiency 73A with defective neutrophil chemotaxix and leukocytosis, 608203;?Immunodeficiency 73C with defective neutrophil chemotaxis and hypogammaglobulinemia, 618987;Immunodeficiency 73B with defective neutrophil chemotaxis and lymphopenia, 618986
RAC3	100%	100%	100%	98.5%	Neurodevelopmental disorder with structural brain anomalies and dysmorphic facies, 618577
RACGAP1	100%	100%	100%	99.6%	Anemia, congenital dyserythropoietic, type IIIb, autosomal recessive, 619789
RAD21	100%	100%	100%	99.8%	Cornelia de Lange syndrome 4, 614701;?Mungan syndrome, 611376
RAD21L1	100%	100%	100%	99.7%	
RAD50	100%	100%	100%	99.7%	Nijmegen breakage syndrome-like disorder, 613078

RAD51	89.3%	89.3%	100%	99.4%	Mirror movements 2, 614508;{Breast cancer, susceptibility to}, 114480;Fanconi anemia, complementation group R, 617244
RAD51C	90.3%	90.3%	100%	99.4%	{Breast-ovarian cancer, familial, susceptibility to, 3}, 613399;Fanconi anemia, complementation group O, 613390
RAD51D	100%	100%	100%	99%	{Breast-ovarian cancer, familial, susceptibility to, 4}, 614291
RAD54B	100%	100%	100%	99.9%	Colon cancer, somatic, 114500;Lymphoma, non-Hodgkin, somatic, 605027
RAD54L	100%	100%	100%	99.4%	{Breast cancer, invasive ductal}, 114480;Adenocarcinoma, colonic, somatic;Lymphoma, non-Hodgkin, somatic, 605027
RAF1	96.6%	93.5%	100%	99.7%	Cardiomyopathy, dilated, 1NN, 615916;Noonan syndrome 5, 611553;LEOPARD syndrome 2, 611554

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
RAG2	100%	100%	100%	100%	Severe combined immunodeficiency, B cell-negative, 601457;Combined cellular and humoral immune defects with granulomas, 233650;Omenn syndrome, 603554
RAI1	100%	100%	100%	98.7%	Smith-Magenis syndrome, 182290
RALA	100%	100%	100%	100%	Hiatt-Neu-Cooper neurodevelopmental syndrome, 619311
RALGAPA1	100%	100%	100%	99.8%	Neurodevelopmental disorder with hypotonia, neonatal respiratory insufficiency, and thermodysregulation, 618797

RANBP2	100%	100%	100%	99.8%	{Encephalopathy, acute, infection-induced, 3, susceptibility to}, 608033
RANGRF	100%	100%	100%	99.3%	
RAP1B	100%	100%	100%	99.8%	Thrombocytopenia 11 with multiple congenital anomalies and dysmorphic facies, 620654
RAP1GDS1	100%	100%	100%	99.8%	Alfadhel syndrome, 620655
RAPGEF2	100%	100%	100%	99.5%	?Epilepsy, familial adult myoclonic, 7, 618075
RAPSN	100%	100%	100%	99.2%	Fetal akinesia deformation sequence 2, 618388;Myasthenic syndrome, congenital, 11, associated with acetylcholine receptor deficiency, 616326
RARB	98%	98%	100%	99.5%	Microphthalmia, syndromic 12, 615524
RARS1	94.5%	94.3%	100%	99.3%	Leukodystrophy, hypomyelinating, 9, 616140
RARS2	94%	92.7%	100%	99.7%	Pontocerebellar hypoplasia, type 6, 611523
RASA1	100%	100%	100%	99.9%	Capillary malformation-arteriovenous malformation 1, 608354;Basal cell carcinoma, somatic, 605462
RASGRP1	95%	95%	100%	99.5%	Immunodeficiency 64, 618534

RASGRP2	100%	100%	100%	98.6%	?Bleeding disorder, platelet-type, 18, 615888
RAX	100%	100%	100%	98.4%	Microphthalmia, syndromic 16, 611038
RAX2	100%	100%	100%	99.1%	Retinitis pigmentosa 95, 620102;Cone-rod dystrophy 11, 610381;?Macular degeneration, age-related, 6, 613757
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
RB1CC1	100%	100%	100%	99.8%	Breast cancer, somatic, 114480
RBBP5	100%	100%	100%	99.7%	
RBBP6	100%	100%	100%	99.6%	
RBBP7	100%	99.1%	98.5%	70.6%	
RBBP8	100%	100%	100%	99.5%	Seckel syndrome 2, 606744;Jawad syndrome, 251255;Pancreatic carcinoma, somatic
RBCK1	100%	100%	100%	97.5%	Polyglucosan body myopathy 1 with or without immunodeficiency, 615895

RBFOX1	99.9%	99%	100%	99.2%	
RBFOX2	92.6%	91.1%	100%	99.1%	
RBL2	100%	100%	100%	99.7%	Brunet-Wagner neurodevelopmental syndrome, 619690
RBM10	100%	99.3%	98.3%	68.6%	TARP syndrome, 311900
RBM20	100%	100%	100%	99.3%	Cardiomyopathy, dilated, 1DD, 613172
RBM28	100%	100%	100%	99.6%	?Alopecia, neurologic defects, and endocrinopathy syndrome, 612079
RBM8A	100%	100%	100%	99.6%	Thrombocytopenia-absent radius syndrome, 274000
RBMX	100%	99.6%	99.5%	80.1%	?Intellectual developmental disorder, X-linked syndromic, Gustavson type, 309555;?Intellectual developmental disorder, X-linked syndromic, Shashi type, 300238
RBMY1A1	50%	50%	49.8%	46.6%	
RBMY1B	50%	49.6%	49%	40%	
RBMY1D	49.5%	48.2%	46.9%	41%	
RBMY1E	49.8%	49.2%	48.4%	42.5%	
RBMY1F	49.2%	48.6%	48.6%	31.5%	
RBMY1J	50%	49.9%	49.7%	34.2%	
RBP3	100%	100%	100%	99.3%	?Retinitis pigmentosa 66, 615233

RBP4	100%	100%	100%	98.4%	Microphthalmia/coloboma 10, 616428;Retinal dystrophy, iris coloboma, and comedogenic acne syndrome, 615147
RBPJ	100%	100%	100%	99.8%	Adams-Oliver syndrome 3, 614814
RBSN	100%	100%	100%	99.4%	Myelofibrosis, congenital, with anemia, neutropenia, developmental delay, and ocular abnormalities, 620939;Kariminejad-Reversade neurodevelopmental syndrome, 620937
RC3H1	100%	100%	100%	99.4%	?Immune dysregulation and systemic hyperinflammation syndrome, 618998
RCBTB1	100%	100%	100%	99.6%	Retinal dystrophy with or without extraocular anomalies, 617175
RD3	100%	100%	100%	99.8%	Leber congenital amaurosis 12, 610612
RDH11	100%	100%	100%	99.6%	?Retinal dystrophy, juvenile cataracts, and short stature syndrome, 616108
RDH12	100%	100%	100%	99.1%	Leber congenital amaurosis 13, 612712
RDH5	100%	100%	100%	99.3%	Fundus albipunctatus, 136880

RDX	100%	100%	100%	99.8%	Deafness, autosomal recessive 24, 611022
REC114	100%	100%	100%	99.7%	Oocyte/zygote/embryo maturation arrest 10, 619176
RECQL	100%	100%	100%	99.7%	RECON progeroid syndrome, 620370
RECQL4	100%	100%	100%	98.9%	Baller-Gerold syndrome, 218600;Rothmund-Thomson syndrome, type 2, 268400;RAPADILINO syndrome, 266280
REEP1	85.8%	85.8%	100%	99.2%	Neuronopathy, distal hereditary motor, autosomal recessive 6, 620011;Spastic paraplegia 31, autosomal dominant, 610250;?Neuronopathy, distal hereditary motor, autosomal dominant 12, 614751
REEP2	100%	100%	100%	98.2%	Spastic paraplegia 72A, autosomal dominant, 615625;?Spastic paraplegia 72B, autosomal recessive, 620606
REEP6	100%	100%	100%	98%	Retinitis pigmentosa 77, 617304
REL	100%	99.4%	100%	99.8%	Immunodeficiency 92, 619652

RELA	100%	100%	100%	98.8%	Autoinflammatory disease, familial, Behcet-like-3, 618287
RELB	100%	99.8%	100%	96.4%	?Immunodeficiency 53, 617585
RELN	100%	100%	100%	99.5%	{Epilepsy, familial temporal lobe, 7}, 616436;Lissencephaly 2 (Norman-Roberts type), 257320
RELT	100%	100%	100%	99%	Amelogenesis imperfecta, type IIIC, 618386
REN	100%	100%	100%	99.4%	Renal tubular dysgenesis, 267430;[Hyperproreninemia];Tubulointerstitial kidney disease, autosomal dominant, 4, 613092
REPS1	100%	100%	100%	99.6%	?Neurodegeneration with brain iron accumulation 7, 617916
RERE	99%	99%	99.9%	96.3%	Neurodevelopmental disorder with or without anomalies of the brain, eye, or heart, 616975
REST	100%	100%	100%	99.7%	Deafness, autosomal dominant 27, 612431;{Wilms tumor 6, susceptibility to}, 616806;Fibromatosis, gingival, 5, 617626

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
RETREG1	91.3%	91.3%	100%	99.2%	Neuropathy, hereditary sensory and autonomic, type IIB, 613115
REV3L	100%	100%	100%	99.7%	
RFC1	100%	100%	100%	99.6%	Cerebellar ataxia, neuropathy, and vestibular areflexia syndrome, 614575
RFC4	100%	100%	100%	99.8%	Morimoto-Ryu-Malicdan neuromuscular syndrome, 621010
RFT1	100%	100%	100%	99.5%	Congenital disorder of glycosylation, type In, 612015
RFWD3	100%	100%	100%	99.8%	?Fanconi anemia, complementation group W, 617784
RFX3	100%	100%	100%	99.8%	
RFX4	100%	100%	100%	98.5%	

RFX5	100%	100%	100%	99.3%	?MHC class II deficiency 5, 620818;MHC class II deficiency 3, 620816
RFX6	100%	100%	100%	99.5%	Mitchell-Riley syndrome, 615710
RFX7	100%	100%	100%	99.2%	Intellectual developmental disorder, autosomal dominant 71, with behavioral abnormalities, 620330
RFXANK	100%	100%	100%	98.2%	MHC class II deficiency 2, 620815
RFXAP	100%	100%	100%	96.5%	MHC class II deficiency 4, 620817
RGR	100%	100%	100%	99.3%	Retinitis pigmentosa 44, 613769
RGS10	100%	100%	100%	99.4%	
RGS9	100%	100%	100%	99.4%	Prolonged electroretinal response suppression 1, 608415
RGS9BP	100%	100%	100%	98.9%	Prolonged electroretinal response suppression 2, 620344
RHAG	100%	100%	100%	99.5%	Overhydrated hereditary stomatocytosis, 185000;Anemia, hemolytic, Rh-null, regulator type, 268150
RHBDF2	100%	99.9%	99.9%	97.5%	Tylosis with esophageal cancer, 148500

RHCE	96.8%	96.8%	96.3%	90.9%	Rh-null disease, amorph type, 617970
RHEB	100%	100%	100%	99.1%	
RHO	100%	100%	100%	98.8%	Night blindness, congenital stationary, autosomal dominant 1, 610445;Retinitis pigmentosa 4, autosomal dominant or recessive, 613731;Retinitis punctata albescens, 136880
RHOA	80.4%	80.4%	100%	98.8%	Ectodermal dysplasia with facial dysmorphism and acral, ocular, and brain anomalies, somatic mosaic, 618727
RHOBTB2	98.7%	98.7%	100%	99.2%	Developmental and epileptic encephalopathy 64, 618004
RHOG	100%	100%	100%	99.4%	
RHOH	100%	100%	100%	99.5%	{?Epidermodysplasia verruciformis, susceptibility to, 4}, 618307
RIC1	100%	100%	100%	99.8%	CATIFA syndrome, 618761
RICTOR	100%	100%	100%	99.8%	
RIGI	98.6%	98.6%	100%	99.6%	Singleton-Merten syndrome 2, 616298
RILPL1	100%	100%	100%	98.7%	Oculopharyngodistal myopathy 4, 619790
RIMS1	100%	100%	100%	99.6%	

RIMS2	99.2%	99.2%	100%	99.4%	Cone-rod synaptic disorder syndrome, congenital nonprogressive, 618970
RIN2	100%	100%	100%	99.3%	Macrocephaly, alopecia, cutis laxa, and scoliosis, 613075
RINT1	100%	100%	100%	99.6%	Infantile liver failure syndrome 3, 618641
RIPK1	100%	100%	100%	99.3%	Immunodeficiency 57 with autoinflammation, 618108;Autoinflammation with episodic fever and lymphadenopathy, 618852
RIPK4	100%	100%	100%	99.4%	CHAND syndrome, 214350;Popliteal pterygium syndrome, Bartsocas-Papas type 1, 263650
RIPOR2	100%	100%	100%	99.6%	Deafness, autosomal dominant 21, 607017;?Deafness, autosomal recessive 104, 616515
RIPPLY2	100%	100%	100%	99.1%	?Spondylocostal dysostosis 6, 616566
RIT1	100%	100%	100%	99%	Noonan syndrome 8, 615355
RLBP1	100%	100%	100%	99.6%	Bothnia retinal dystrophy, 607475;Newfoundland rod-cone dystrophy, 607476;Retinitis punctata albescens, 136880;Fundus albipunctatus, 136880

RLF	100%	100%	100%	99.7%	
RLIM	100%	99.8%	99.4%	72.6%	Tonne-Kalscheuer syndrome, 300978
RMND1	85.6%	85.6%	100%	99.6%	Combined oxidative phosphorylation deficiency 11, 614922
RMRP					Anauxetic dysplasia 1, 607095;Metaphyseal dysplasia without hypotrichosis, 250460;Cartilage-hair hypoplasia, 250250
RNASEH1	100%	99.7%	100%	99.2%	Progressive external ophthalmoplegia with mitochondrial DNA deletions, autosomal recessive 2, 616479
RNASEH2A	100%	100%	100%	99.2%	Aicardi-Goutieres syndrome 4, 610333
RNASEH2B	91.4%	91.4%	100%	99.2%	Aicardi-Goutieres syndrome 2, 610181
RNASEH2C	100%	100%	100%	98.1%	Aicardi-Goutieres syndrome 3, 610329
RNASEL	100%	100%	100%	99.7%	Prostate cancer 1, 601518
RNASET2	100%	100%	100%	99%	Leukoencephalopathy, cystic, without megalencephaly, 612951
RNF111	100%	100%	100%	99.3%	
RNF113A	100%	99.5%	97.6%	69.4%	Trichothiodystrophy 5, nonphotosensitive, 300953

RNF125	100%	100%	100%	99%	Tenorio syndrome, 616260
RNF13	100%	100%	100%	99.8%	Developmental and epileptic encephalopathy 73, 618379
RNF135	100%	100%	100%	99.6%	
RNF139	100%	100%	100%	99.5%	Renal cell carcinoma, 144700
RNF168	100%	100%	100%	99.4%	RIDDLE syndrome, 611943
RNF170	100%	100%	100%	99.7%	Ataxia, sensory, 1, autosomal dominant, 608984;Spastic paraplegia 85, autosomal recessive, 619686
RNF2	100%	100%	100%	99.6%	Luo-Schoch-Yamamoto syndrome, 619460
RNF212	100%	100%	100%	99%	?Spermatogenic failure 62, 619673;Recombination rate QTL 1, 612042
RNF213	100%	100%	100%	99.3%	{Moyamoya disease 2, susceptibility to}, 607151
RNF216	100%	100%	100%	99.8%	Cerebellar ataxia and hypogonadotropic hypogonadism, 212840
RNF220	100%	100%	100%	99.5%	Leukodystrophy, hypomyelinating, 23, with ataxia, deafness, liver dysfunction, and dilated cardiomyopathy, 619688
RNF31	100%	100%	100%	98.8%	Immunodeficiency 115 with autoinflammation, 620632

RNF43	100%	100%	100%	98.6%	Sessile serrated polyposis cancer syndrome, 617108
RNF6	100%	100%	100%	99.7%	Esophageal carcinoma, somatic, 133239
RNPC3	100%	100%	100%	99.8%	Pituitary hormone deficiency, combined or isolated, 7, 618160
RNU12					CDAGS syndrome, 603116;?Spinocerebellar ataxia, autosomal recessive 33, 620208
RNU2-2P					
RNU4-2					ReNU syndrome, 620851
RNU4ATAC					Roifman syndrome, 616651;Lowry-Wood syndrome, 226960;Microcephalic osteodysplastic primordial dwarfism, type I, 210710
RNU5B-1					
RNU6-1					
RNU6-2					
RNU6-8					
RNU6-9					
RNU7-1					Aicardi-Goutieres syndrome 9, 619487

ROBO1	100%	100%	100%	99.7%	Pituitary hormone deficiency, combined or isolated, 8, 620303;Neurooculorenal syndrome, 620305;?Nystagmus 8, congenital, autosomal recessive, 257400
ROBO2	100%	100%	100%	99.5%	Vesicoureteral reflux 2, 610878
ROBO3	100%	100%	100%	98.6%	Gaze palsy, familial horizontal, with progressive scoliosis, 1, 607313
ROBO4	100%	100%	100%	98.9%	Aortic valve disease 3, 618496
ROGDI	100%	100%	100%	98.4%	Kohlschutter-Tonz syndrome, 226750
ROM1	100%	100%	100%	99.4%	Retinitis pigmentosa 7, digenic form, 608133
ROR1	100%	100%	100%	99.4%	?Deafness, autosomal recessive 108, 617654
ROR2	100%	100%	100%	99.4%	Brachydactyly, type B1, 113000;Robinow syndrome, autosomal recessive, 268310
RORA	100%	100%	100%	98.4%	Intellectual developmental disorder with or without epilepsy or cerebellar ataxia, 618060

RORB	100%	100%	100%	99.7%	{Epilepsy, idiopathic generalized, susceptibility to, 15}, 618357
RORC	100%	100%	100%	99.1%	Immunodeficiency 42, 616622
RP1	100%	100%	100%	99.8%	Retinitis pigmentosa 1, 180100
RP1L1	100%	100%	100%	98.4%	Occult macular dystrophy, 613587;Retinitis pigmentosa 88, 618826
RP2	100%	99.9%	98.5%	72.9%	Retinitis pigmentosa 2, 312600
RP9	100%	100%	100%	95%	?Retinitis pigmentosa 9, 180104
RPA1	100%	100%	100%	99.3%	Pulmonary fibrosis and/or bone marrow failure syndrome, telomere-related, 6, 619767
RPE65	100%	100%	100%	99.7%	Retinitis pigmentosa 20, 613794;Retinitis pigmentosa 87 with choroidal involvement, 618697;Leber congenital amaurosis 2, 204100

RPGR	99.7%	96.4%	87.3%	58.5%	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
RPGRIP1	100%	100%	100%	98.9%	Cone-rod dystrophy 13, 608194;Leber congenital amaurosis 6, 613826
RPGRIP1L	100%	100%	100%	99.7%	Joubert syndrome 7, 611560;Meckel syndrome 5, 611561;?COACH syndrome 3, 619113
RPH3A	100%	100%	100%	99.6%	
RPIA	100%	100%	100%	99%	Ribose 5-phosphate isomerase deficiency, 608611
RPL10	100%	99.3%	98.3%	70.8%	{Autism, susceptibility to, X-linked 5}, 300847;Intellectual developmental disorder, X-linked syndromic 35, 300998
RPL10L	100%	100%	100%	99.7%	?Spermatogenic failure 63, 619689
RPL11	100%	100%	100%	99.3%	Diamond-Blackfan anemia 7, 612562

RPL13	100%	99.2%	100%	98.8%	Spondyloepimetaphyseal dysplasia, Isidor-Toutain type, 618728
RPL15	97.6%	89.4%	100%	99.6%	Diamond-Blackfan anemia 12, 615550
RPL18	100%	100%	100%	98.8%	?Diamond-Blackfan anemia 18, 618310
RPL21	100%	99.7%	100%	99.7%	Hypotrichosis 12, 615885
RPL26	100%	100%	100%	100%	?Diamond-Blackfan anemia 11, 614900
RPL27	100%	99.8%	100%	99.3%	?Diamond-Blackfan anemia 16, 617408
RPL31	100%	99.7%	100%	99.8%	
RPL35	100%	100%	100%	99.5%	?Diamond-Blackfan anemia 19, 618312
RPL35A	100%	100%	100%	99.8%	Diamond-Blackfan anemia 5, 612528
RPL3L	100%	100%	100%	97.3%	Cardiomyopathy, dilated, 2D, 619371
RPL4	100%	100%	100%	99.6%	
RPL5	100%	100%	100%	99.8%	Diamond-Blackfan anemia 6, 612561
RPL9	100%	100%	100%	99.9%	
RPN2	100%	100%	100%	99.3%	
RPS10	100%	100%	100%	99.5%	Diamond-Blackfan anemia 9, 613308

RPS14	100%	100%	100%	99.4%	Macrocytic anemia, refractory, due to 5q deletion, somatic, 153550
RPS15A	79.7%	79.7%	100%	99.8%	?Diamond-Blackfan anemia 20, 618313
RPS17	100%	100%	100%	99.1%	Diamond-Blackfan anemia 4, 612527
RPS19	100%	100%	100%	99.2%	Diamond-Blackfan anemia 1, 105650
RPS20	100%	100%	100%	99.1%	
RPS23	100%	100%	100%	99.9%	Brachycephaly, trichomegaly, and developmental delay, 617412
RPS24	100%	100%	100%	99.7%	Diamond-blackfan anemia 3, 610629
RPS26	100%	98.1%	100%	99.7%	Diamond-Blackfan anemia 10, 613309
RPS27	100%	100%	100%	99.5%	?Diamond-Blackfan anemia 17, 617409
RPS28	100%	100%	100%	99.2%	Diamond Blackfan anemia 15 with mandibulofacial dysostosis, 606164
RPS29	100%	100%	100%	99.7%	Diamond-Blackfan anemia 13, 615909
RPS4Y2	50%	50%	49.3%	24.3%	
RPS6KA3	100%	99.7%	99.3%	74.9%	Intellectual developmental disorder, X-linked 19, 300844;Coffin-Lowry syndrome, 303600

RPS6KB1	100%	100%	100%	99.2%	
RPS7	100%	100%	100%	98.2%	Diamond-Blackfan anemia 8, 612563
RPSA	100%	100%	100%	100%	Asplenia, isolated congenital, 271400
RRAD	100%	100%	100%	95.4%	
RRAGC	100%	100%	100%	99.3%	Long-Olsen-Distelmaier syndrome, 620609
RRAGD	100%	100%	100%	99.3%	Hypomagnesemia 7, renal, with or without dilated cardiomyopathy, 620152
RRAS	100%	99.7%	100%	97.2%	
RRAS2	100%	100%	100%	99.3%	Ovarian carcinoma;Noonan syndrome 12, 618624
RREB1	100%	100%	100%	98.3%	
RRM1	100%	100%	100%	99.5%	Progressive external ophthalmoplegia with mitochondrial DNA deletions, autosomal recessive 6, 620647

RRM2B	100%	100%	100%	99.9%	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
RRP7A	99.9%	99.6%	100%	96.3%	?Microcephaly 28, primary, autosomal recessive, 619453
RS1	100%	99.8%	99.2%	70.5%	Retinoschisis, 312700
RSPH1	100%	100%	100%	99.6%	Ciliary dyskinesia, primary, 24, 615481
RSPH3	100%	100%	100%	99.2%	Ciliary dyskinesia, primary, 32, 616481
RSPH4A	100%	100%	100%	99.2%	Ciliary dyskinesia, primary, 11, 612649
RSPH9	100%	100%	100%	99.4%	Ciliary dyskinesia, primary, 12, 612650

RSPO1	100%	100%	100%	99.5%	Palmoplantar hyperkeratosis and true hermaphroditism, 610644;Palmoplantar hyperkeratosis with squamous cell carcinoma of skin and sex reversal, 610644
RSPO2	100%	100%	100%	99.9%	?Humerofemoral hypoplasia with radiotibial ray deficiency, 618022;Tetraamelia syndrome 2, 618021
RSPO4	100%	100%	100%	99.2%	Anonychia congenita, 206800
RSPRY1	100%	100%	100%	99.6%	Spondyloepimetaphyseal dysplasia, Faden-Alkuraya type, 616723
RSRC1	100%	100%	100%	99.9%	Intellectual developmental disorder, autosomal recessive 70, 618402
RTEL1	100%	100%	100%	99.1%	Dyskeratosis congenita, autosomal dominant 4, 615190;Dyskeratosis congenita, autosomal recessive 5, 615190;Pulmonary fibrosis and/or bone marrow failure syndrome, telomere-related, 3, 616373

RTN2	100%	100%	100%	98.9%	Neuronopathy, distal hereditary motor, autosomal recessive 11, with spasticity, 620854;Spastic paraplegia 12, autosomal dominant, 604805
RTN4IP1	100%	100%	100%	99.7%	Optic atrophy 10 with or without ataxia, impaired intellectual development and seizures, 616732
RTTN	100%	100%	100%	99.7%	Microcephaly, short stature, and polymicrogyria with seizures, 614833
RUBCN	100%	100%	100%	99.1%	Spinocerebellar ataxia, autosomal recessive 15, 615705
RUNX1	100%	100%	99%	92.2%	Platelet disorder, familial, with associated myeloid malignancy, 601399;Leukemia, acute myeloid, 601626
RUNX1T1	100%	100%	100%	99.2%	
RUNX2	100%	100%	100%	98.4%	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

RUSC2	100%	100%	100%	99.6%	Intellectual developmental disorder, autosomal recessive 61, 617773
RXFP2	100%	100%	100%	99.9%	
RXYLT1	100%	100%	100%	99.6%	Muscular dystrophy-dystroglycanopathy (congenital with brain and eye anomalies), type A, 10, 615041
RYBP	100%	100%	100%	99.2%	
RYR1	100%	99.8%	100%	98.4%	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
RYR2	100%	100%	100%	99.6%	Ventricular tachycardia, catecholaminergic polymorphic, 1, 604772;Ventricular arrhythmias due to cardiac ryanodine receptor calcium release deficiency syndrome, 115000
RYR3	100%	100%	100%	99.6%	Congenital myopathy 20, 620310
S100A3	100%	100%	100%	99.3%	

S1PR2	100%	100%	100%	99.4%	Deafness, autosomal recessive 68, 610419
SACS	99%	99%	100%	99.8%	Spastic ataxia, Charlevoix-Saguenay type, 270550
SAG	100%	100%	100%	99.4%	Retinitis pigmentosa 47, autosomal recessive, 613758;Retinitis pigmentosa 96, autosomal dominant, 620228;Oguchi disease-1, 258100
SALL1	100%	100%	100%	99.1%	Townes-Brocks syndrome 1, 107480;Townes-Brocks branchiootorenal-like syndrome, 107480
SALL2	100%	100%	100%	99.4%	?Coloboma, ocular, autosomal recessive, 216820
SALL4	100%	100%	100%	99.2%	?IVIC syndrome, 147750;Duane-radial ray syndrome, 607323
SAMD11	100%	100%	100%	98.7%	
SAMD12	100%	100%	100%	99.8%	Epilepsy, familial adult myoclonic, 1, 601068
SAMD7	100%	100%	100%	98.7%	Macular dystrophy with or without cone dysfunction, 620762

SAMD9	100%	100%	100%	99.6%	Tumoral calcinosis, familial, normophosphatemic, 610455;Monosomy 7 myelodysplasia and leukemia syndrome 2, 619041;MIRAGE syndrome, 617053
SAMD9L	100%	100%	100%	99.8%	Ataxia-pancytopenia syndrome, 159550;?Spinocerebellar ataxia 49, 619806;Monosomy 7 myelodysplasia and leukemia syndrome 1, 252270
SAMHD1	100%	100%	100%	99.8%	?Chilblain lupus 2, 614415;Aicardi-Goutieres syndrome 5, 612952
SAR1B	100%	100%	100%	99.1%	Chylomicron retention disease, 246700
SARDH	91.7%	91.7%	100%	99.2%	[Sarcosinemia], 268900
SARS1	100%	100%	100%	99.4%	Neurodevelopmental disorder with microcephaly, ataxia, and seizures, 617709
SARS2	100%	100%	100%	98.5%	Hyperuricemia, pulmonary hypertension, renal failure, and alkalosis, 613845
SART3	100%	100%	100%	99.5%	

SASH1	100%	100%	100%	99%	Dyschromatosis universalis hereditaria 1, 127500;?Cancer, alopecia, pigment dyscrasia, onychodystrophy, and keratoderma, 618373
SASH3	100%	99.7%	98%	70.2%	Immunodeficiency 102, 301082
SASS6	100%	100%	100%	99.7%	Microcephaly 14, primary, autosomal recessive, 616402
SAT1	100%	100%	99.6%	74%	
SATB1	100%	100%	100%	99.6%	den Hoed-de Boer-Voisin syndrome, 619229;Developmental delay with dysmorphic facies and dental anomalies, 619228
SATB2	100%	100%	100%	99.5%	Glass syndrome, 612313
SBDS	100%	100%	100%	99.7%	{Aplastic anemia, susceptibility to}, 609135;Shwachman-Diamond syndrome 1, 260400
SBF1	100%	100%	100%	99%	Charcot-Marie-Tooth disease, type 4B3, 615284
SBF2	93.7%	93.7%	100%	99.7%	Charcot-Marie-Tooth disease, type 4B2, 604563
SC5D	100%	100%	100%	99.8%	Lathosterolosis, 607330
SCAF4	100%	100%	100%	99%	Fliedner-Zweier syndrome, 620511

SCAMP5	100%	100%	100%	98.7%	
SCAPER	100%	100%	100%	99.7%	Intellectual developmental disorder and retinitis pigmentosa, 618195
SCARB2	100%	100%	100%	99.5%	Epilepsy, progressive myoclonic 4, with or without renal failure, 254900
SCARF2	100%	100%	100%	95.1%	Van den Ende-Gupta syndrome, 600920
SCD5	100%	100%	100%	98.3%	?Deafness, autosomal dominant 79, 619086
SCLT1	95.2%	95.2%	100%	99.8%	
SCN10A	100%	100%	100%	99.5%	Episodic pain syndrome, familial, 2, 615551
SCN11A	100%	99.9%	100%	99.3%	Episodic pain syndrome, familial, 3, 615552;Neuropathy, hereditary sensory and autonomic, type VII, 615548
SCN1A	100%	100%	100%	99.7%	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

SCN1B	100%	99.1%	100%	99.1%	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
SCN2A	100%	100%	100%	99.8%	Seizures, benign familial infantile, 3, 607745;Developmental and epileptic encephalopathy 11, 613721;Episodic ataxia, type 9, 618924
SCN2B	100%	100%	100%	98.8%	Atrial fibrillation, familial, 14, 615378
SCN3A	100%	100%	100%	99.5%	Epilepsy, familial focal, with variable foci 4, 617935;Developmental and epileptic encephalopathy 62, 617938
SCN3B	100%	100%	100%	99.1%	Atrial fibrillation, familial, 16, 613120;Brugada syndrome 7, 613120

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
SCN4B	100%	100%	100%	98.3%	Atrial fibrillation, familial, 17, 611819;Long QT syndrome 10, 611819
SCN5A	100%	100%	100%	99.2%	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
SCN7A	100%	100%	100%	99.7%	

SCN8A	100%	100%	100%	99.2%	?Myoclonus, familial, 2, 618364;Seizures, benign familial infantile, 5, 617080;Cognitive impairment with or without cerebellar ataxia, 614306;Developmental and epileptic encephalopathy 13, 614558
SCN9A	100%	100%	100%	98.8%	Erythermalgia, primary, 133020;Insensitivity to pain, congenital, 243000;Small fiber neuropathy, 133020;Paroxysmal extreme pain disorder, 167400;Neuropathy, hereditary sensory and autonomic, type IID, 243000
SCNM1	100%	100%	100%	99.1%	Orofaciodigital syndrome XIX, 620107
SCNN1A	100%	100%	100%	98.6%	Pseudohypoaldosteronism, type IB1, autosomal recessive, 264350;?Liddle syndrome 3, 618126;Bronchiectasis with or without elevated sweat chloride 2, 613021
SCNN1B	100%	100%	100%	99.6%	Bronchiectasis with or without elevated sweat chloride 1, 211400;Pseudohypoaldosteronism, type IB2, autosomal recessive, 620125;Liddle syndrome 1, 177200

SCNN1G	100%	100%	100%	99.2%	Bronchiectasis with or without elevated sweat chloride 3, 613071;Pseudohypoaldosteronism, type IB3, autosomal recessive, 620126;Liddle syndrome 2, 618114
SCO1	100%	100%	100%	99.7%	Mitochondrial complex IV deficiency, nuclear type 4, 619048
SCO2	100%	100%	100%	99.4%	Myopia 6, 608908;Mitochondrial complex IV deficiency, nuclear type 2, 604377
SCP2	100%	100%	100%	99.7%	?Leukoencephalopathy with dystonia and motor neuropathy, 613724
SCUBE3	100%	100%	100%	99%	Short stature, facial dysmorphism, and skeletal anomalies with or without cardiac anomalies, 619184
SCYL1	100%	100%	100%	98.6%	Spinocerebellar ataxia, autosomal recessive 21, 616719
SCYL2	100%	100%	100%	99.7%	Arthrogryposis multiplex congenita 4, neurogenic, with agenesis of the corpus callosum, 618766
SDCCAG8	100%	100%	100%	99.7%	Senior-Loken syndrome 7, 613615;Bardet-Biedl syndrome 16, 615993

SDHA	100%	100%	100%	99.4%	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
SDHAF1	100%	100%	100%	99.6%	Mitochondrial complex II deficiency, nuclear type 2, 619166
SDHAF2	99.8%	97.8%	100%	99.7%	Pheochromocytoma/paraganglioma syndrome 2, 601650
SDHB	100%	100%	100%	99.7%	Pheochromocytoma/paraganglioma syndrome 4, 115310;Mitochondrial complex II deficiency, nuclear type 4, 619224;Gastrointestinal stromal tumor, 606764;Paraganglioma and gastric stromal sarcoma, 606864
SDHC	100%	100%	100%	99.8%	Pheochromocytoma/paraganglioma syndrome 3, 605373;Paraganglioma and gastric stromal sarcoma, 606864;Gastrointestinal stromal tumor, 606764

SDHD	78.9%	78.9%	100%	98.8%	Pheochromocytoma/paraganglioma syndrome 1, 168000;Paraganglioma and gastric stromal sarcoma, 606864;Mitochondrial complex II deficiency, nuclear type 3, 619167
SDR9C7	100%	100%	100%	99.2%	Ichthyosis, congenital, autosomal recessive 13, 617574
SEC23A	100%	100%	100%	99.7%	Craniolenticulosutural dysplasia, 607812
SEC23B	100%	100%	100%	99.7%	?Cowden syndrome 7, 616858;Dyserythropoietic anemia, congenital, type II, 224100
SEC24D	100%	100%	100%	99.3%	Cole-Carpenter syndrome 2, 616294
SEC31A	100%	100%	100%	99.6%	?Halperin-Birk syndrome, 618651
SEC61A1	100%	100%	100%	99.6%	Immunodeficiency, common variable, 15, 620670;?Neutropenia, severe congenital, 11, autosomal dominant, 620674;Tubulointerstitial kidney disease, autosomal dominant, 5, 617056
SEC61B	100%	100%	100%	98%	
SEC63	100%	100%	100%	99.6%	Polycystic liver disease 2, 617004

SECISBP2	100%	100%	100%	99.6%	Thyroid hormone metabolism, abnormal, 1, 609698
SELENBP1	100%	100%	100%	99.2%	Extraoral halitosis due to MTO deficiency, 618148
SELENOI	100%	100%	100%	99.9%	Spastic paraplegia 81, autosomal recessive, 618768
SELENON	91.6%	88.5%	100%	98.3%	Congenital myopathy 3 with rigid spine, 602771
SEMA3A	100%	100%	100%	99.8%	{Hypogonadotropic hypogonadism 16 with or without anosmia}, 614897
SEMA3E	100%	100%	100%	99.7%	
SEMA4A	100%	100%	100%	98.6%	Retinitis pigmentosa 35, 610282;Cone-rod dystrophy 10, 610283
SEMA6B	100%	100%	100%	97.2%	Epilepsy, progressive myoclonic, 11, 618876
SEMA7A	100%	100%	100%	98.8%	?Cholestasis, progressive familial intrahepatic, 11, 619874;[Blood group, John-Milton-Hagen system], 614745
SEPHS1	100%	100%	100%	99.6%	
SEPSECS	99.4%	95.2%	100%	99.4%	Pontocerebellar hypoplasia type 2D, 613811
SEPTIN12	100%	100%	100%	98.9%	Spermatogenic failure 10, 614822
SEPTIN4	100%	100%	100%	98.6%	

SEPTIN9	100%	100%	100%	97.6%	Amyotrophy, hereditary neuralgic, 162100
SERAC1	100%	100%	100%	99.8%	3-methylglutaconic aciduria with deafness, encephalopathy, and Leigh-like syndrome, 614739
SERPINA1	100%	100%	100%	98.4%	Hemorrhagic diathesis due to antithrombin Pittsburgh, 613490;Emphysema due to AAT deficiency, 613490;Emphysema-cirrhosis, due to AAT deficiency, 613490
SERPINA12	100%	100%	100%	99.3%	
SERPINA3	100%	100%	100%	99.6%	Alpha-1-antichymotrypsin deficiency;Cerebrovascular disease, occlusive
SERPINA6	100%	100%	100%	99.7%	Corticosteroid-binding globulin deficiency, 611489
SERPINB6	100%	100%	100%	99.4%	?Deafness, autosomal recessive 91, 613453
SERPINB7	100%	100%	100%	99.6%	Palmoplantar keratoderma, Nagashima type, 615598
SERPINB8	100%	100%	100%	99.3%	Peeling skin syndrome 5, 617115
SERPINC1	100%	100%	100%	99.6%	Thrombophilia 7 due to antithrombin III deficiency, 613118
SERPIND1	100%	100%	100%	99.5%	Thrombophilia 10 due to heparin cofactor II deficiency, 612356

SERPINE1	100%	100%	100%	99.3%	Plasminogen activator inhibitor-1 deficiency, 613329;{Transcription of plasminogen activator inhibitor, modulator of}
SERPINF1	100%	100%	100%	98.8%	Osteogenesis imperfecta, type VI, 613982
SERPINF2	100%	100%	100%	99.2%	Alpha-2-plasmin inhibitor deficiency, 262850
SERPING1	100%	100%	100%	99.5%	Angioedema, hereditary, 1 and 2, 106100;Complement component 4, partial deficiency of, 120790
SERPINH1	100%	100%	100%	99.3%	{Preterm premature rupture of the membranes, susceptibility to}, 610504;Osteogenesis imperfecta, type X, 613848
SERPINI1	100%	100%	100%	99.9%	Encephalopathy, familial, with neuroserpin inclusion bodies, 604218
SET	100%	99.2%	100%	98.2%	Intellectual developmental disorder, autosomal dominant 58, 618106
SETBP1	100%	100%	100%	99%	Schinzel-Giedion midface retraction syndrome, 269150;Intellectual developmental disorder, autosomal dominant 29, 616078

SETD1A	100%	100%	100%	98.6%	Epilepsy, early-onset, 2, with or without developmental delay, 618832;Neurodevelopmental disorder with speech impairment and dysmorphic facies, 619056
SETD1B	100%	100%	99.9%	96.5%	Intellectual developmental disorder with seizures and language delay, 619000
SETD2	100%	100%	100%	99.5%	Luscan-Lumish syndrome, 616831;Intellectual developmental disorder, autosomal dominant 70, 620157;Rabin-Pappas syndrome, 620155
SETD5	99.4%	98.4%	100%	99.5%	Intellectual developmental disorder, autosomal dominant 23, 615761
SETX	100%	100%	100%	99.5%	Spinocerebellar ataxia, autosomal recessive, with axonal neuropathy 2, 606002;Amyotrophic lateral sclerosis 4, juvenile, 602433
SEZ6	100%	100%	100%	98.5%	
SF3B1	100%	100%	100%	99.8%	Myelodysplastic syndrome, somatic, 614286
SF3B2	100%	100%	100%	99%	Craniofacial microsomia, 164210
SF3B4	100%	100%	100%	98.3%	Acrofacial dysostosis 1, Nager type, 154400

SFRP4	100%	100%	100%	99.4%	Pyle disease, 265900
SFTPA1	100%	100%	100%	99.7%	Interstitial lung disease 1, 619611
SFTPA2	100%	100%	100%	99.2%	Interstitial lung disease 2, 178500
SFTPB	100%	100%	100%	99.3%	Surfactant metabolism dysfunction, pulmonary, 1, 265120
SFTPC	100%	100%	100%	99%	Surfactant metabolism dysfunction, pulmonary, 2, 610913
SFXN4	100%	100%	100%	99.5%	Combined oxidative phosphorylation deficiency 18, 615578
SGCA	100%	100%	100%	98.8%	Muscular dystrophy, limb-girdle, autosomal recessive 3, 608099
SGCB	100%	100%	100%	99.7%	Muscular dystrophy, limb-girdle, autosomal recessive 4, 604286
SGCD	100%	100%	100%	99%	Cardiomyopathy, dilated, 1L, 606685;Muscular dystrophy, limb-girdle, autosomal recessive 6, 601287
SGCE	90.8%	90.8%	100%	99.6%	Dystonia-11, myoclonic, 159900
SGCG	100%	100%	100%	99.8%	Muscular dystrophy, limb-girdle, autosomal recessive 5, 253700

SGMS1	100%	100%	100%	99.6%	
SGMS2	100%	100%	100%	99.8%	Calvarial doughnut lesions with bone fragility with or without spondylometaphyseal dysplasia, 126550
SGO1	100%	100%	100%	99.7%	Chronic atrial and intestinal dysrhythmia, 616201
SGPL1	96.6%	96.6%	100%	99.5%	RENI syndrome, 617575
SGSH	100%	100%	100%	99.3%	Mucopolysaccharidosis type IIIA (Sanfilippo A), 252900
SGSM3	100%	100%	100%	99.4%	
SH2B3	100%	100%	100%	98.7%	Thrombocythemia, somatic, 187950;Myelofibrosis, somatic, 254450;Erythrocytosis, somatic, 133100
SH2D1A	100%	100%	99.1%	73.8%	Lymphoproliferative syndrome, X-linked, 1, 308240
SH3BP2	99.9%	99%	100%	98.8%	Cherubism, 118400
SH3KBP1	98.8%	98.4%	99%	72.4%	?Immunodeficiency 61, 300310
SH3PXD2B	100%	100%	100%	98.8%	Frank-ter Haar syndrome, 249420
SH3TC2	100%	100%	100%	99.7%	Charcot-Marie-Tooth disease, type 4C, 601596;Mononeuropathy of the median nerve, mild, 613353

SHANK1	100%	100%	100%	95.1%	
SHANK2	100%	100%	100%	98.5%	{Autism susceptibility 17}, 613436
SHANK3	99.3%	97.7%	99.8%	95.4%	Phelan-McDermid syndrome, 606232;{Schizophrenia 15}, 613950
SHARPIN	100%	100%	100%	99.5%	Autoinflammation with episodic fever and immune dysregulation, 620795
SHH	100%	100%	100%	97.1%	Single median maxillary central incisor, 147250;Holoprosencephaly 3, 142945;Microphthalmia/coloboma 5, 611638
SHMT2	100%	100%	100%	99.3%	Neurodevelopmental disorder with cardiomyopathy, spasticity, and brain abnormalities, 619121
SHOC1	100%	100%	100%	99.8%	Spermatogenic failure 75, 619949
SHOC2	100%	100%	100%	99.7%	Noonan syndrome-like with loose anagen hair 1, 607721

SHOX	47.4%	47.4%	50%	49.4%	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
SHOX2	100%	100%	99.9%	96.8%	
SHQ1	100%	100%	100%	99.6%	Neurodevelopmental disorder with dystonia and seizures, 619922;?Dystonia 35, childhood-onset, 619921
SHROOM3	100%	100%	100%	99.2%	
SHROOM4	100%	99.7%	99.1%	71.1%	
SI	100%	100%	99.8%	99%	Sucrase-isomaltase deficiency, congenital, 222900
SIAH1	100%	100%	100%	99.9%	Buratti-Harel syndrome, 619314
SIGMAR1	100%	100%	100%	99.6%	?Neuronopathy, distal hereditary motor, autosomal recessive 2, 605726;?Amyotrophic lateral sclerosis 16, juvenile, 614373
SIK1	100%	100%	100%	99.4%	Developmental and epileptic encephalopathy 30, 616341

SIK3	100%	100%	100%	98.8%	?Spondyloepimetaphyseal dysplasia, Krakow type, 618162
SIL1	100%	100%	100%	99.5%	Marinesco-Sjogren syndrome, 248800
SIN3A	100%	100%	100%	99.6%	Witteveen-Kolk syndrome, 613406
SIN3B	100%	100%	100%	99%	
SIPA1L3	100%	100%	100%	98.7%	?Cataract 45, 616851
SIRT5	100%	100%	100%	99.6%	
SIX1	100%	100%	100%	98.6%	Deafness, autosomal dominant 23, 605192;Branchiootic syndrome 3, 608389
SIX3	100%	100%	100%	96.2%	Schizencephaly, 269160;Holoprosencephaly 2, 157170
SIX5	100%	100%	100%	97%	Branchiootorenal syndrome 2, 610896
SIX6	100%	100%	100%	97.5%	Optic disc anomalies with retinal and/or macular dystrophy, 212550
SKI	100%	99.8%	100%	96.8%	Shprintzen-Goldberg syndrome, 182212
SKIC2	100%	100%	100%	99.2%	Trichohepatoenteric syndrome 2, 614602
SKIC3	98.9%	98.9%	100%	99.6%	Trichohepatoenteric syndrome 1, 222470

SLC10A1	100%	100%	100%	99.1%	Hypercholanemia, familial 2, 619256
SLC10A2	100%	100%	100%	99.6%	?Bile acid malabsorption, primary, 1, 613291
SLC10A7	92.8%	92.8%	100%	99.8%	Short stature, amelogenesis imperfecta, and skeletal dysplasia with scoliosis, 618363
SLC11A2	100%	100%	100%	99.8%	Anemia, hypochromic microcytic, with iron overload 1, 206100
SLC12A1	96.3%	96.3%	100%	99.8%	Bartter syndrome, type 1, 601678
SLC12A2	100%	100%	100%	99.4%	Kilquist syndrome, 619080;Delpire-McNeill syndrome, 619083;Deafness, autosomal dominant 78, 619081
SLC12A3	100%	100%	100%	98.8%	Gitelman syndrome, 263800
SLC12A5	100%	100%	100%	98.8%	{Epilepsy, idiopathic generalized, susceptibility to, 14}, 616685;Developmental and epileptic encephalopathy 34, 616645
SLC12A6	100%	99.9%	100%	99.7%	Agenesis of the corpus callosum with peripheral neuropathy, 218000;Charcot-Marie-Tooth disease, axonal, type 2II, 620068

SLC12A9	100%	100%	100%	97%	
SLC13A3	100%	100%	100%	99.3%	Leukoencephalopathy, acute reversible, with increased urinary alpha-ketoglutarate, 618384
SLC13A5	100%	100%	100%	99.4%	Developmental and epileptic encephalopathy 25, with amelogenesis imperfecta, 615905
SLC16A1	100%	100%	100%	99.4%	Hyperinsulinemic hypoglycemia, familial, 7, 610021;Erythrocyte lactate transporter defect, 245340;Monocarboxylate transporter 1 deficiency, 616095
SLC16A12	100%	100%	100%	99.8%	Cataract 47, juvenile, with microcornea, 612018
SLC16A2	100%	99.8%	99%	70.5%	Allan-Herndon-Dudley syndrome, 300523
SLC17A5	100%	100%	100%	99.9%	Salla disease, 604369;Sialic acid storage disorder, infantile, 269920
SLC17A8	100%	100%	100%	99.8%	Deafness, autosomal dominant 25, 605583
SLC17A9	100%	100%	100%	99.3%	Porokeratosis 8, disseminated superficial actinic type, 616063
SLC18A2	100%	100%	100%	99.5%	Parkinsonism-dystonia, infantile, 2, 618049

SLC18A3	100%	100%	100%	99.3%	Myasthenic syndrome, congenital, 21, presynaptic, 617239
SLC19A1	100%	100%	100%	97.9%	Immunodeficiency 114, folate-responsive, 620603;?Megaloblastic anemia, folate-responsive, 601775
SLC19A2	100%	100%	100%	99.8%	Thiamine-responsive megaloblastic anemia syndrome, 249270
SLC19A3	99.8%	98.7%	100%	99.6%	Thiamine metabolism dysfunction syndrome 2 (biotin/thiamine-responsive basal ganglia disease type), 607483
SLC1A1	100%	100%	100%	99.6%	Dicarboxylic aminoaciduria, 222730;{?Schizophrenia susceptibility 18}, 615232
SLC1A2	100%	100%	100%	99.3%	Developmental and epileptic encephalopathy 41, 617105
SLC1A3	97.1%	94.1%	100%	99.8%	Episodic ataxia, type 6, 612656
SLC1A4	100%	100%	100%	99.4%	Spastic tetraplegia, thin corpus callosum, and progressive microcephaly, 616657
SLC20A2	100%	100%	100%	99.3%	Basal ganglia calcification, idiopathic, 1, 213600
SLC22A12	100%	99.7%	100%	97.5%	Hypouricemia, renal, 220150

SLC22A18	100%	100%	100%	99.2%	Breast cancer, somatic, 114480;Lung cancer, somatic, 211980;Rhabdomyosarcoma, somatic, 268210
SLC22A4	100%	100%	100%	99.3%	{Rheumatoid arthritis, susceptibility to}, 180300
SLC22A5	100%	100%	100%	99.2%	Carnitine deficiency, systemic primary, 212140
SLC24A1	100%	100%	100%	98.7%	Night blindness, congenital stationary (complete), 1D, autosomal recessive, 613830
SLC24A4	100%	100%	100%	99.3%	[Skin/hair/eye pigmentation 6, blond/brown hair], 210750;Amelogenesis imperfecta, type IIA5, 615887;[Skin/hair/eye pigmentation 6, blue/green eyes], 210750
SLC24A5	100%	100%	100%	99.8%	[Skin/hair/eye pigmentation 4, fair/dark skin], 113750;Albinism, oculocutaneous, type VI, 113750
SLC25A1	100%	100%	100%	97.4%	Combined D-2- and L-2-hydroxyglutaric aciduria, 615182;Myasthenic syndrome, congenital, 23, presynaptic, 618197

SLC25A10	100%	100%	100%	99.1%	?Mitochondrial DNA depletion syndrome 19, 618972
SLC25A11	100%	100%	100%	98.9%	Pheochromocytoma/paraganglioma syndrome 6, 618464
SLC25A12	100%	100%	100%	99.7%	Developmental and epileptic encephalopathy 39, 612949
SLC25A13	100%	100%	100%	99.7%	Citrullinemia, type II, neonatal-onset, 605814;Citrullinemia, adult-onset type II, 603471
SLC25A15	100%	100%	100%	99.5%	Hyperornithinemia-hyperammonemia-homocitrullinemia syndrome, 238970
SLC25A19	100%	100%	100%	99.5%	Microcephaly, Amish type, 607196;Thiamine metabolism dysfunction syndrome 4 (progressive polyneuropathy type), 613710
SLC25A20	100%	100%	100%	99.3%	Carnitine-acylcarnitine translocase deficiency, 212138
SLC25A21	100%	100%	100%	99.7%	?Mitochondrial DNA depletion syndrome 18, 618811
SLC25A22	100%	100%	100%	98.9%	Developmental and epileptic encephalopathy 3, 609304

SLC25A24	99.5%	99.5%	99.6%	97.4%	Fontaine progeroid syndrome, 612289
SLC25A26	100%	100%	100%	99.8%	Combined oxidative phosphorylation deficiency 28, 616794
SLC25A3	100%	100%	100%	99.9%	Mitochondrial phosphate carrier deficiency, 610773
SLC25A32	100%	100%	100%	99.8%	?Exercise intolerance, riboflavin-responsive, 616839
SLC25A36	100%	100%	100%	99.2%	Hyperinsulinemic hypoglycemia, familial, 8, 620211
SLC25A38	100%	100%	100%	99.7%	Anemia, sideroblastic, 2, pyridoxine-refractory, 205950
SLC25A4	100%	100%	100%	98.8%	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

SLC25A42	100%	100%	100%	99.1%	Metabolic crises, recurrent, with variable encephalomyopathic features and neurologic regression, 618416
SLC25A46	100%	100%	100%	99.6%	Neuropathy, hereditary motor and sensory, type VIB, 616505;Pontocerebellar hypoplasia, type 1E, 619303
SLC26A1	100%	100%	100%	99.5%	?Hypersulfaturia, 620372;?Nephrolithiasis, calcium oxalate, 1, 167030
SLC26A2	100%	100%	100%	99.7%	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
SLC26A3	100%	100%	100%	99.6%	Diarrhea 1, secretory chloride, congenital, 214700
SLC26A4	100%	100%	100%	99.5%	Deafness, autosomal recessive 4, with enlarged vestibular aqueduct, 600791;Pendred syndrome, 274600

SLC26A5	100%	100%	100%	99.7%	?Deafness, autosomal recessive 61, 613865
SLC26A8	100%	100%	100%	98.5%	Spermatogenic failure 3, 606766
SLC27A4	100%	100%	100%	98.5%	Ichthyosis prematurity syndrome, 608649
SLC28A1	100%	100%	100%	99.1%	[Uridine-cytidineuria], 618477
SLC29A3	100%	100%	100%	99.3%	Histiocytosis-lymphadenopathy plus syndrome, 602782
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
SLC2A10	100%	100%	100%	99.2%	Arterial tortuosity syndrome, 208050
SLC2A2	100%	100%	100%	99.8%	Fanconi-Bickel syndrome, 227810;{Diabetes mellitus, noninsulin-dependent}, 125853
SLC2A9	100%	100%	100%	99%	{Uric acid concentration, serum, QTL 2}, 612076;Hypouricemia, renal, 2, 612076

SLC30A10	100%	100%	100%	98.8%	Hypermanganesemia with dystonia 1, 613280
SLC30A2	100%	100%	100%	99.3%	Zinc deficiency, transient neonatal, 608118
SLC30A5	100%	100%	100%	98.8%	
SLC30A7	100%	100%	100%	99.8%	Ziegler-Huang syndrome, 620501
SLC30A9	100%	100%	100%	99.7%	Birk-Landau-Perez syndrome, 617595
SLC31A1	100%	100%	100%	99.9%	Neurodegeneration and seizures due to copper transport defect, 620306
SLC32A1	100%	100%	100%	99.3%	Generalized epilepsy with febrile seizures plus, type 12, 620755;Developmental and epileptic encephalopathy 114, 620774
SLC33A1	100%	100%	100%	99.4%	Spastic paraplegia 42, autosomal dominant, 612539;Huppke-Brendel syndrome, 614482
SLC34A1	100%	100%	100%	98.5%	?Fanconi renotubular syndrome 2, 613388;Hypercalcemia, infantile, 2, 616963;Nephrolithiasis/osteoporosis, hypophosphatemic, 1, 612286

SLC34A2	100%	100%	100%	99%	Pulmonary alveolar microlithiasis, 265100
SLC34A3	100%	100%	100%	97.3%	Hypophosphatemic rickets with hypercalciuria, 241530
SLC35A1	100%	100%	100%	99.7%	Congenital disorder of glycosylation, type IIf, 603585
SLC35A2	100%	99.9%	99.1%	70.9%	Congenital disorder of glycosylation, type IIm, 300896
SLC35A3	94.4%	86.9%	100%	99.7%	Arthrogryposis, impaired intellectual development, and seizures, 615553
SLC35B2	100%	100%	100%	97.8%	Leukodystrophy, hypomyelinating, 26, with chondrodysplasia, 620269
SLC35C1	100%	100%	100%	99.2%	Congenital disorder of glycosylation, type IIc, 266265
SLC35D1	100%	100%	100%	99.7%	Schneckenbecken dysplasia, 269250
SLC36A2	100%	100%	100%	99.3%	[Iminoglycinuria], 242600;[Hyperglycinuria], 138500
SLC37A3	100%	100%	100%	99.7%	
SLC37A4	100%	100%	100%	99.5%	Glycogen storage disease Ib, 232220;Congenital disorder of glycosylation, type IIw, 619525;Glycogen storage disease Ic, 232240

SLC38A3	100%	100%	100%	98.9%	Developmental and epileptic encephalopathy 102, 619881
SLC38A8	100%	99.9%	100%	98.7%	Foveal hypoplasia 2, with or without optic nerve misrouting and/or anterior segment dysgenesis, 609218
SLC39A12	100%	100%	100%	99.7%	
SLC39A13	100%	100%	100%	97.7%	Ehlers-Danlos syndrome, spondylodysplastic type, 3, 612350
SLC39A14	93.6%	93.6%	100%	99.4%	?Hyperostosis cranalis interna, 144755;Hypermanganesemia with dystonia 2, 617013
SLC39A4	100%	100%	100%	99.5%	Acrodermatitis enteropathica, 201100
SLC39A5	100%	100%	100%	98.9%	Myopia 24, autosomal dominant, 615946
SLC39A7	100%	100%	100%	98.6%	Agammaglobulinemia 9, autosomal recessive, 619693
SLC39A8	100%	99.1%	100%	99.3%	Congenital disorder of glycosylation, type IIn, 616721
SLC3A1	96.2%	96.2%	100%	99.1%	Cystinuria, 220100
SLC40A1	100%	100%	100%	99.7%	Hemochromatosis, type 4, 606069

SLC41A1	100%	100%	100%	99.3%	?Nephronophthisis-like nephropathy 2, 619468
SLC44A1	100%	100%	100%	99.2%	Neurodegeneration, childhood-onset, with ataxia, tremor, optic atrophy, and cognitive decline, 618868
SLC44A4	100%	100%	100%	99%	?Deafness, autosomal dominant 72, 617606
SLC45A1	100%	100%	100%	98.9%	Intellectual developmental disorder with neuropsychiatric features, 617532
SLC45A2	100%	100%	100%	99.6%	[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
SLC46A1	100%	100%	100%	98.7%	Folate malabsorption, hereditary, 229050

SLC4A1	100%	100%	100%	98.8%	[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
SLC4A10	100%	100%	100%	99.8%	Neurodevelopmental disorder with hypotonia and characteristic brain abnormalities, 620746
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
SLC4A2	100%	100%	100%	98.9%	?Osteopetrosis, autosomal recessive 9, 620366
SLC4A3	100%	100%	100%	98.8%	Short QT syndrome 7, 620231

SLC4A4	97.3%	97.3%	100%	99.7%	Proximal renal tubular acidosis-ocular anomaly syndrome, 604278
SLC4A7	100%	100%	100%	99.7%	
SLC51A	100%	100%	100%	98.4%	?Cholestasis, progressive familial intrahepatic, 6, 619484
SLC51B	100%	100%	100%	99.3%	?Bile acid malabsorption, primary, 2, 619481
SLC52A1	100%	100%	100%	99.4%	Riboflavin deficiency, 615026
SLC52A2	100%	100%	100%	99.6%	Brown-Vialetto-Van Laere syndrome 2, 614707
SLC52A3	100%	100%	100%	99.1%	?Fazio-Londe disease, 211500;Brown-Vialetto-Van Laere syndrome 1, 211530
SLC5A1	100%	100%	100%	99.3%	Glucose/galactose malabsorption, 606824
SLC5A2	100%	100%	100%	99%	Renal glucosuria, 233100
SLC5A5	100%	100%	100%	98.7%	Thyroid dyshormonogenesis 1, 274400
SLC5A6	100%	100%	100%	99.4%	Sodium-dependent multivitamin transporter deficiency, 618973;Peripheral motor neuropathy, childhood-onset, biotin-responsive, 619903

SLC5A7	100%	100%	100%	99.3%	Neuronopathy, distal hereditary motor, autosomal dominant 7, 158580;Myasthenic syndrome, congenital, 20, presynaptic, 617143
SLC66A1	100%	100%	100%	99.1%	
SLC6A1	100%	100%	100%	99.1%	Myoclonic-atonic epilepsy, 616421
SLC6A17	100%	100%	100%	99.2%	Intellectual developmental disorder, autosomal recessive 48, 616269
SLC6A19	100%	100%	100%	98.9%	Hartnup disorder, 234500
SLC6A2	100%	100%	100%	99.3%	?Orthostatic intolerance, 604715
SLC6A20	100%	100%	100%	99.5%	
SLC6A3	100%	100%	100%	99.2%	Parkinsonism-dystonia, infantile, 1, 613135;{Nicotine dependence, protection against}, 188890
SLC6A5	100%	100%	100%	98.7%	Hyperekplexia 3, 614618
SLC6A6	100%	100%	100%	99%	Hypotaurinemic retinal degeneration and cardiomyopathy, 145350
SLC6A8	100%	98.5%	97.7%	68.3%	Cerebral creatine deficiency syndrome 1, 300352
SLC6A9	100%	100%	100%	99.3%	Glycine encephalopathy with normal serum glycine, 617301

SLC7A14	100%	100%	100%	99.6%	Retinitis pigmentosa 68, 615725
SLC7A6OS	100%	100%	100%	99.6%	Epilepsy, progressive myoclonic, 12, 619191
SLC7A7	100%	100%	100%	97.5%	Lysinuric protein intolerance, 222700
SLC7A9	100%	100%	100%	99.5%	Cystinuria, 220100
SLC8B1	100%	100%	100%	99.1%	
SLC9A1	100%	100%	100%	99.1%	Lichtenstein-Knorr syndrome, 616291
SLC9A3	100%	99.3%	100%	96.3%	Diarrhea 8, secretory sodium, congenital, 616868
SLC9A6	100%	100%	99.5%	72.8%	Intellectual developmental disorder, X-linked syndromic, Christianson type, 300243
SLC9A7	100%	99.5%	98.7%	71.4%	Intellectual developmental disorder, X-linked 108, 301024
SLCO1B1	100%	99.9%	99.9%	99.1%	Hyperbilirubinemia, Rotor type, digenic, 237450
SLCO1B3	100%	100%	100%	99.9%	Hyperbilirubinemia, Rotor type, digenic, 237450
SLCO2A1	100%	100%	100%	99%	Hypertrophic osteoarthropathy, primary, autosomal dominant, 167100;PHOAR2-enteropathy syndrome, 614441
SLCO5A1	100%	100%	100%	99.5%	

SLF2	100%	100%	100%	99.7%	Atelis syndrome 1, 620184
SLFN14	100%	100%	100%	99.4%	Bleeding disorder, platelet-type, 20, 616913
SLIRP	100%	100%	100%	98.9%	
SLIT3	100%	100%	100%	99.3%	
SLITRK1	100%	100%	100%	99.2%	Tourette syndrome, 137580;?Trichotillomania, 613229
SLITRK2	100%	100%	98.7%	71.7%	Intellectual developmental disorder, X-linked 111, 301107
SLITRK6	100%	100%	100%	99.7%	Deafness and myopia, 221200
SLMAP	100%	100%	100%	99.6%	
SLURP1	100%	100%	100%	99.2%	Meleda disease, 248300
SLX4	100%	100%	100%	99%	Fanconi anemia, complementation group P, 613951
SMAD1	100%	100%	100%	99.7%	
SMAD2	100%	100%	100%	99.6%	Loeys-Dietz syndrome 6, 619656;Congenital heart defects, multiple types, 8, with or without heterotaxy, 619657
SMAD3	100%	100%	100%	98.7%	Loeys-Dietz syndrome 3, 613795

SMAD4	100%	100%	100%	99.9%	Pancreatic cancer, somatic, 260350;Myhre syndrome, 139210;Polyposis, juvenile intestinal, 174900;Juvenile polyposis/hereditary hemorrhagic telangiectasia syndrome, 175050
SMAD5	100%	100%	100%	99.9%	
SMAD6	100%	100%	100%	96.5%	Aortic valve disease 2, 614823;{Radioulnar synostosis, nonsyndromic}, 179300;{Craniosynostosis 7, susceptibility to}, 617439
SMAD9	100%	100%	100%	98.8%	Pulmonary hypertension, primary, 2, 615342
SMARCA1	100%	99.8%	99.1%	75.2%	
SMARCA2	97.9%	97.6%	100%	99.3%	Nicolaides-Baraitser syndrome, 601358;Blepharophimosis-impaired intellectual development syndrome, 619293
SMARCA4	100%	100%	100%	98.7%	Coffin-Siris syndrome 4, 614609;{Rhabdoid tumor predisposition syndrome 2}, 613325;?Otosclerosis 12, 620792
SMARCA5	100%	100%	100%	99.2%	
SMARCAD1	100%	100%	100%	99.9%	Basan syndrome, 129200;Huriez syndrome, 181600;Adermatoglyphia, 136000

SMARCAL1	100%	100%	100%	99.4%	Schimke immunoosseous dysplasia, 242900
SMARCB1	100%	99.9%	100%	98.1%	Rhabdoid tumors, somatic, 609322;{Schwannomatosis-1, susceptibility to}, 162091;Coffin-Siris syndrome 3, 614608;{Rhabdoid tumor predisposition syndrome 1}, 609322
SMARCC2	100%	100%	100%	98.8%	Coffin-Siris syndrome 8, 618362
SMARCD1	100%	100%	100%	98.6%	Coffin-Siris syndrome 11, 618779
SMARCD2	100%	100%	100%	98.3%	Specific granule deficiency 2, 617475
SMARCE1	100%	100%	100%	99.5%	{Meningioma, familial, susceptibility to}, 607174;Coffin-Siris syndrome 5, 616938
SMC1A	100%	99.6%	98.9%	69.4%	Cornelia de Lange syndrome 2, 300590;Developmental and epileptic encephalopathy 85, with or without midline brain defects, 301044
SMC3	100%	100%	100%	99.7%	Cornelia de Lange syndrome 3, 610759
SMC5	100%	100%	100%	99.8%	Atelis syndrome 2, 620185

SMCHD1	100%	100%	100%	99.7%	Facioscapulohumeral muscular dystrophy 2, digenic, 158901;Bosma arhinia microphthalmia syndrome, 603457
SMDT1	100%	100%	100%	99.4%	
SMG8	100%	100%	100%	99.7%	Alzahrani-Kuwahara syndrome, 619268
SMG9	100%	100%	100%	99%	Heart and brain malformation syndrome, 616920;Neurodevelopmental disorder with intention tremor, pyramidal signs, dyspraxia, and ocular anomalies, 619995
SMN1	93.9%	93.9%	99.7%	96.1%	Spinal muscular atrophy-2, 253550;Spinal muscular atrophy-4, 271150;Spinal muscular atrophy-3, 253400;Spinal muscular atrophy-1, 253300
SMO	100%	100%	100%	99.2%	Pallister-Hall-like syndrome, 241800;Basal cell carcinoma, somatic, 605462;Curry-Jones syndrome, somatic mosaic, 601707
SMOC1	100%	100%	100%	99.6%	Microphthalmia with limb anomalies, 206920
SMOC2	100%	100%	100%	99.6%	Dentin dysplasia, type I, with microdontia and misshapen teeth, 125400

SMPD1	100%	100%	100%	99.1%	Niemann-Pick disease, type B, 607616;Niemann-Pick disease, type A, 257200
SMPD4	100%	100%	100%	99.2%	Neurodevelopmental disorder with microcephaly, arthrogryposis, and structural brain anomalies, 618622
SMPX	100%	100%	99%	77.8%	Myopathy, distal, 7, adult-onset, X-linked, 301075;Deafness, X-linked 4, 300066
SMS	99.9%	97.3%	99.1%	74.7%	Intellectual developmental disorder, X-linked syndromic, Snyder-Robinson type, 309583
SNAI2	100%	100%	100%	99.7%	
SNAP25	100%	100%	100%	99.5%	?Myasthenic syndrome, congenital, 18, 616330
SNAP29	100%	100%	100%	99.1%	Cerebral dysgenesis, neuropathy, ichthyosis, and palmoplantar keratoderma syndrome, 609528
SNAPC4	100%	100%	100%	98.6%	Neurodevelopmental disorder with motor regression, progressive spastic paraplegia, and oromotor dysfunction, 620515

SNCA	100%	100%	100%	99.6%	Dementia, Lewy body, 127750;Parkinson disease 1, 168601;Parkinson disease 4, 605543
SNCB	100%	100%	100%	98.7%	Dementia, Lewy body, 127750
SNF8	100%	100%	100%	99.5%	Developmental and epileptic encephalopathy 115, 620783;Neurodevelopmental disorder plus optic atrophy, 620784
SNIP1	100%	100%	100%	99.7%	Neurodevelopmental disorder with hypotonia, craniofacial abnormalities, and seizures, 614501
SNORA31					{Encephalopathy, acute, infection-induced (herpes-specific), susceptibility to, 10}, 619396
SNORD118					Leukoencephalopathy, brain calcifications, and cysts, 614561
SNRNP200	100%	100%	100%	99.5%	Retinitis pigmentosa 33, 610359
SNRPB	100%	100%	100%	99.5%	Cerebrocostomandibular syndrome, 117650
SNRPE	100%	100%	100%	99.7%	Hypotrichosis 11, 615059
SNRPN	100%	100%	100%	99.4%	
SNTA1	100%	100%	100%	98.8%	Long QT syndrome 12, 612955

SNUPN	100%	100%	100%	99.3%	Muscular dystrophy, limb-girdle, autosomal recessive 29, 620793
SNX10	89.3%	89.3%	100%	99.8%	Osteopetrosis, autosomal recessive 8, 615085
SNX14	95%	95%	100%	99.9%	Spinocerebellar ataxia, autosomal recessive 20, 616354
SNX27	100%	100%	100%	99.3%	
SOBP	99.9%	99%	99.8%	92.3%	?Impaired intellectual development, anterior maxillary protrusion, and strabismus, 613671
SOCS1	100%	100%	100%	94.4%	Autoinflammatory syndrome, familial, with or without immunodeficiency, 619375
SOCS4	100%	100%	100%	99.5%	
SOD1	100%	100%	100%	99.7%	Spastic tetraplegia and axial hypotonia, progressive, 618598;Amyotrophic lateral sclerosis 1, 105400
SOD2	100%	100%	100%	99.3%	{Microvascular complications of diabetes 6}, 612634
SOHLH1	100%	100%	100%	98.6%	Ovarian dysgenesis 5, 617690;Spermatogenic failure 32, 618115
SON	100%	100%	100%	99.5%	ZTTK syndrome, 617140

SORD	95.8%	90.7%	98%	89.6%	Neuronopathy, distal hereditary motor, autosomal recessive 8, 618912
SOS1	98.7%	98.3%	100%	99.7%	Noonan syndrome 4, 610733;Fibromatosis, gingival, 1, 135300
SOS2	100%	100%	100%	99.3%	Noonan syndrome 9, 616559
SOST	100%	100%	100%	99.4%	Sclerosteosis 1, 269500;Craniodiaphyseal dysplasia, autosomal dominant, 122860
SOX10	97.8%	97.8%	100%	98.3%	Waardenburg syndrome, type 4C, 613266;PCWH syndrome, 609136;Waardenburg syndrome, type 2E, with or without neurologic involvement, 611584
SOX11	100%	100%	100%	94.3%	Intellectual developmental disorder with microcephaly and with or without ocular malformations or hypogonadotropic hypogonadism, 615866
SOX17	100%	100%	100%	98.8%	Vesicoureteral reflux 3, 613674

SOX18	100%	98.3%	100%	97.1%	Hypotrichosis-lymphedema-telangiectasia syndrome, 607823;Hypotrichosis-lymphedema-telangiectasia-renal defect syndrome, 137940
SOX2	100%	99.9%	100%	96.8%	Optic nerve hypoplasia and abnormalities of the central nervous system, 206900;Microphthalmia, syndromic 3, 206900
SOX3	100%	100%	95.2%	58.4%	Intellectual developmental disorder, X-linked, with isolated growth hormone deficiency, 300123;Panhypopituitarism, X-linked, 312000
SOX4	100%	100%	99.9%	93.3%	Coffin-Siris syndrome 10, 618506
SOX5	100%	100%	100%	99.6%	Lamb-Shaffer syndrome, 616803
SOX6	100%	100%	100%	99.6%	Tolchin-Le Caignec syndrome, 618971
SOX7	100%	100%	100%	96%	
SOX9	100%	100%	100%	96.9%	Campomelic dysplasia with autosomal sex reversal, 114290;Acampomelic campomelic dysplasia, 114290;Campomelic dysplasia, 114290

SP110	100%	100%	100%	99.3%	{Mycobacterium tuberculosis, susceptibility to}, 607948;Hepatic venoocclusive disease with immunodeficiency, 235550
SP6	100%	100%	100%	99.1%	Amelogenesis imperfecta, type IK, 620104
SP7	100%	100%	100%	98.9%	Osteogenesis imperfecta, type XII, 613849
SP9	100%	100%	100%	96.8%	
SPACA1	100%	100%	100%	99.9%	?Spermatogenic failure 85, 620490
SPAG1	100%	100%	100%	99.4%	Ciliary dyskinesia, primary, 28, 615505
SPAG17	100%	100%	100%	99.8%	?Spermatogenic failure 55, 619380
SPAG6	100%	100%	100%	99.7%	
SPARC	100%	100%	100%	99.1%	Osteogenesis imperfecta, type XVII, 616507
SPARCL1	100%	100%	100%	99.7%	
SPART	100%	100%	100%	99.7%	Troyer syndrome, 275900
SPAST	100%	99.9%	100%	98.5%	Spastic paraplegia 4, autosomal dominant, 182601
SPATA16	100%	100%	100%	99.9%	?Spermatogenic failure 6, 102530
SPATA22	100%	100%	100%	99.9%	Premature ovarian failure 25, 621002;Spermatogenic failure 96, 621001

SPATA7	100%	100%	100%	99.8%	Leber congenital amaurosis 3, 604232;Retinitis pigmentosa 94, variable age at onset, autosomal recessive, 604232
SPECC1L	100%	99.2%	100%	99.4%	Teebi hypertelorism syndrome 1, 145420;?Facial clefting, oblique, 1, 600251
SPEF2	100%	100%	100%	99.6%	Spermatogenic failure 43, 618751
SPEG	100%	100%	100%	98.3%	Centronuclear myopathy 5, 615959
SPEN	100%	100%	100%	99.1%	Radio-Tartaglia syndrome, 619312
SPG11	99.6%	99.6%	100%	99.5%	Amyotrophic lateral sclerosis 5, juvenile, 602099;Charcot-Marie-Tooth disease, axonal, type 2X, 616668;Spastic paraplegia 11, autosomal recessive, 604360
SPG21	100%	100%	100%	99.7%	Mast syndrome, 248900
SPG7	100%	100%	100%	98.7%	Spastic paraplegia 7, autosomal recessive, 607259
SPI1	100%	100%	100%	98.3%	Agammaglobulinemia 10, autosomal dominant, 619707
SPIDR	100%	100%	100%	99.4%	Ovarian dysgenesis 9, 619665

SPIN4	100%	100%	99.5%	72.9%	?Lui-Jee-Baron syndrome, 301114
SPINK1	100%	100%	100%	100%	Tropical calcific pancreatitis, 608189;Pancreatitis, hereditary, 167800;{Fibrocalculous pancreatic diabetes, susceptibility to}, 608189
SPINK2	100%	100%	100%	99.3%	?Spermatogenic failure 29, 618091
SPINK5	100%	100%	100%	99.8%	Netherton syndrome, 256500
SPINT2	100%	100%	100%	98.5%	Diarrhea 3, secretory sodium, congenital, syndromic, 270420
SPNS2	99.9%	99.5%	100%	96.7%	?Deafness, autosomal recessive 115, 618457
SPO11	100%	100%	100%	99.9%	
SPOCK1	100%	100%	100%	98.8%	
SPOP	100%	100%	100%	99.8%	Nabais Sa-de Vries syndrome, type 1, 618828;Nabais Sa-de Vries syndrome, type 2, 618829
SPP2	100%	100%	100%	99.4%	
SPPL2A	100%	100%	100%	99.9%	Immunodeficiency 86, mycobacteriosis, 619549
SPR	100%	100%	100%	98.9%	Dystonia, dopa-responsive, due to sepiapterin reductase deficiency, 612716

SPRED1	100%	100%	100%	99.7%	Legius syndrome, 611431
SPRED2	100%	100%	100%	99.3%	Noonan syndrome 14, 619745
SPRTN	100%	100%	100%	99.9%	Ruijs-Aalfs syndrome, 616200
SPRY4	100%	100%	100%	98.9%	Hypogonadotropic hypogonadism 17 with or without anosmia, 615266
SPTA1	100%	100%	100%	99.5%	Spherocytosis, type 3, 270970;Elliptocytosis-2, 130600;Pyropoikilocytosis, 266140
SPTAN1	99.3%	98.9%	100%	99.4%	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
SPTB	100%	100%	100%	99.2%	Anemia, neonatal hemolytic, fatal or near-fatal, 617948;Elliptocytosis-3, 617948;Spherocytosis, type 2, 616649

SPTBN1	100%	100%	100%	99.4%	Developmental delay, impaired speech, and behavioral abnormalities, 619475
SPTBN2	100%	100%	100%	99%	Spinocerebellar ataxia 5, 600224;Spinocerebellar ataxia, autosomal recessive 14, 615386
SPTBN4	100%	100%	100%	98%	Neurodevelopmental disorder with hypotonia, neuropathy, and deafness, 617519
SPTLC1	88.7%	88.7%	100%	99.8%	Amyotrophic lateral sclerosis 27, juvenile, 620285;Neuropathy, hereditary sensory and autonomic, type IA, 162400
SPTLC2	100%	100%	100%	99.6%	Neuropathy, hereditary sensory and autonomic, type IC, 613640
SPTLC3	100%	100%	100%	99.5%	
SPTSSA	100%	100%	100%	97.5%	Spastic paraplegia 90A, autosomal dominant, 620416;?Spastic paraplegia 90B, autosomal recessive, 620417
SQOR	100%	100%	100%	99.4%	Sulfide:quinone oxidoreductase deficiency, 619221

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
SRC	96.3%	96.3%	100%	98.9%	?Thrombocytopenia 6, 616937;Colon cancer, advanced, somatic, 114500
SRCAP	100%	100%	100%	98.7%	Developmental delay, hypotonia, musculoskeletal defects, and behavioral abnormalities, 619595;Floating-Harbor syndrome, 136140
SRD5A2	100%	100%	100%	99.7%	Pseudovaginal perineoscrotal hypospadias, 264600
SRD5A3	100%	100%	100%	99.4%	Kahrizi syndrome, 612713;Congenital disorder of glycosylation, type Iq, 612379
SREBF1	100%	100%	99.9%	97.1%	Ichthyosis, follicular, with atrichia and photophobia syndrome 2, 619016;Mucoepithelial dysplasia, hereditary, 158310

SRF	100%	100%	100%	96.6%	
SRI	100%	100%	100%	99.6%	
SRP54	100%	100%	100%	99.8%	Neutropenia, severe congenital, 8, autosomal dominant, 618752
SRP68	100%	99.8%	100%	99.5%	?Neutropenia, severe congenital, 10, autosomal recessive, 620534
SRP72	100%	100%	100%	99.5%	Bone marrow failure syndrome 1, 614675
SRPK3	100%	98.7%	98.1%	66.7%	Intellectual developmental disorder, X-linked 114, 301134
SRPX2	100%	98.9%	98.7%	70.8%	?Rolandic epilepsy, impaired intellectual development, and speech dyspraxia, 300643
SRRM2	100%	100%	100%	98.2%	Intellectual developmental disorder, autosomal dominant 72, 620439
SRSF1	100%	100%	100%	99.1%	Neurodevelopmental disorder with dysmorphic facies and behavioral abnormalities, 620489
SRY	50%	50%	48.4%	22.2%	46XY sex reversal 1, 400044;46XX sex reversal 1, 400045
SSBP1	100%	100%	100%	99.7%	Optic atrophy 13 with retinal and foveal abnormalities, 165510

SSR3	100%	100%	100%	99.6%	
SSR4	100%	99.4%	97.4%	65.1%	Congenital disorder of glycosylation, type Iy, 300934
SSTR5	100%	100%	100%	97.9%	
SSX1	100%	100%	98.7%	73.4%	Spermatogenic failure, X-linked, 5, 301099
SSX2	100%	100%	99.1%	77.3%	?Sarcoma, synovial, 300813
ST14	100%	100%	100%	99%	Ichthyosis, congenital, autosomal recessive 11, 602400
ST3GAL3	95.4%	95%	100%	98.9%	Developmental and epileptic encephalopathy 15, 615006;Intellectual developmental disorder, autosomal recessive 12, 611090
ST3GAL5	98.3%	98.3%	100%	99.4%	Salt and pepper developmental regression syndrome, 609056
STAB2	100%	100%	100%	99.6%	
STAC3	100%	100%	100%	99.6%	Congenital myopathy 13, 255995
STAG1	100%	100%	100%	99.6%	Intellectual developmental disorder, autosomal dominant 47, 617635

STAG2	100%	99.9%	99.1%	73.1%	Holoprosencephaly 13, X-linked, 301043;Mullegama-Klein-Martinez syndrome, 301022
STAG3	100%	100%	100%	99.1%	Spermatogenic failure 61, 619672;Premature ovarian failure 8, 615723
STAMBP	96.3%	96.2%	100%	99.3%	Microcephaly-capillary malformation syndrome, 614261
STAR	100%	100%	100%	99.2%	Lipoid adrenal hyperplasia, 201710
STARD7	100%	100%	100%	99.1%	Epilepsy, familial adult myoclonic, 2, 607876
STAT1	95.9%	95.9%	100%	99.7%	Immunodeficiency 31C, chronic mucocutaneous candidiasis, autosomal dominant, 614162;Immunodeficiency 31A, mycobacteriosis, autosomal dominant, 614892;Immunodeficiency 31B, mycobacterial and viral infections, autosomal recessive, 613796
STAT2	100%	100%	100%	99.4%	Pseudo-TORCH syndrome 3, 618886;Immunodeficiency 44, 616636

STAT3	100%	100%	100%	99.7%	Hyper-IgE syndrome 1, autosomal dominant, with recurrent infections, 147060;Autoimmune disease, multisystem, infantile-onset, 1, 615952
STAT4	100%	100%	100%	99.9%	Disabling pansclerotic morphea of childhood, 620443;{Systemic lupus erythematosus, susceptibility to, 11}, 612253
STAT5B	100%	100%	100%	99.2%	Growth hormone insensitivity with immune dysregulation 1, autosomal recessive, 245590;Growth hormone insensitivity with immune dysregulation 2, autosomal dominant, 618985
STAT6	100%	100%	100%	98.6%	Hyper-IgE syndrome 6, autosomal dominant, with recurrent infections, 620532
STEAP3	100%	100%	100%	98.7%	?Anemia, hypochromic microcytic, with iron overload 2, 615234
STEEP1	100%	98.7%	98.8%	67.2%	?Intellectual developmental disorder, X-linked 107, 301013
STIL	100%	100%	100%	99.7%	Microcephaly 7, primary, autosomal recessive, 612703

STIM1	100%	99.8%	100%	99.2%	Myopathy, tubular aggregate, 1, 160565;Stormorken syndrome, 185070;Immunodeficiency 10, 612783
STING1	100%	100%	100%	99.4%	STING-associated vasculopathy, infantile-onset, 615934
STK11	100%	100%	100%	98.7%	Melanoma, malignant, somatic, 155600;Pancreatic cancer, somatic, 260350;Peutz-Jeghers syndrome, 175200;Testicular tumor, somatic, 273300
STK33	100%	100%	100%	99.8%	?Spermatogenic failure 93, 620849
STK36	100%	100%	100%	99.2%	?Ciliary dyskinesia, primary, 46, 619436
STK4	100%	100%	100%	99.8%	T-cell immunodeficiency, recurrent infections, autoimmunity, and cardiac malformations, 614868
STN1	87.1%	87%	100%	99.6%	Cerebroretinal microangiopathy with calcifications and cysts 2, 617341
STOX1	98.8%	97.2%	93%	83.7%	Preeclampsia/eclampsia 4, 609404

STRA6	100%	100%	100%	99.3%	Microphthalmia, syndromic 9, 601186;Microphthalmia, isolated, with coloboma 8, 601186
STRA8	100%	100%	100%	99%	
STRADA	100%	100%	100%	98.9%	Polyhydramnios, megalencephaly, and symptomatic epilepsy, 611087
STRC	100%	100%	99.9%	98.5%	Deafness, autosomal recessive 16, 603720
STS	97.1%	96.8%	98.7%	72.4%	Ichthyosis, X-linked, 308100
STT3A	100%	100%	100%	99.5%	Congenital disorder of glycosylation, type Iw, autosomal dominant, 619714;Congenital disorder of glycosylation, type Iw, autosomal recessive, 615596
STT3B	100%	100%	100%	98.1%	Congenital disorder of glycosylation, type Ix, 615597
STUB1	100%	100%	100%	97.8%	Spinocerebellar ataxia 48, 618093;Spinocerebellar ataxia, autosomal recessive 16, 615768
STX11	100%	100%	100%	99%	Hemophagocytic lymphohistiocytosis, familial, 4, 603552
STX16	100%	100%	100%	99.4%	Pseudohypoparathyroidism Ib, 603233

STX1A	100%	100%	100%	98.5%	
STX1B	100%	100%	100%	97.3%	Generalized epilepsy with febrile seizures plus, type 9, 616172
STX2	100%	100%	100%	99.9%	
STX3	100%	100%	100%	99.6%	Retinal dystrophy and microvillus inclusion disease, 619446;Diarrhea 12, with microvillus atrophy, 619445
STX4	100%	100%	100%	99.4%	?Deafness, autosomal recessive 123, 620745
STX5	100%	100%	100%	99.1%	?Congenital disorder of glycosylation, type IIaa, 620454
STXBP1	100%	100%	100%	99.2%	Developmental and epileptic encephalopathy 4, 612164
STXBP2	100%	100%	100%	98.2%	Hemophagocytic lymphohistiocytosis, familial, 5, with or without microvillus inclusion disease, 613101
SUCLA2	100%	99.7%	100%	99.8%	Mitochondrial DNA depletion syndrome 5 (encephalomyopathic with or without methylmalonic aciduria), 612073
SUCLG1	100%	100%	100%	99.3%	Mitochondrial DNA depletion syndrome 9 (encephalomyopathic type with methylmalonic aciduria), 245400

SUCLG2	100%	100%	100%	99.8%	
SUFU	100%	100%	100%	98.7%	{Meningioma, familial, susceptibility to}, 607174;Joubert syndrome 32, 617757;Basal cell nevus syndrome 2, 620343;{Medulloblastoma}, 155255
SUGCT	100%	100%	100%	99.5%	Glutaric aciduria III, 231690
SULF1	100%	100%	100%	99.6%	
SULT2B1	100%	100%	100%	98.6%	Ichthyosis, congenital, autosomal recessive 14, 617571
SUMF1	100%	100%	100%	99.5%	Multiple sulfatase deficiency, 272200
SUMO1	71%	71%	100%	99.1%	?Orofacial cleft 10, 613705
SUN5	100%	100%	100%	99.3%	Spermatogenic failure 16, 617187
SUOX	100%	100%	100%	98.7%	Sulfite oxidase deficiency, 272300
SUPT16H	100%	100%	100%	99.5%	Neurodevelopmental disorder with dysmorphic facies and thin corpus callosum, 619480
SUPV3L1	100%	100%	100%	99.5%	
SURF1	100%	100%	100%	98.5%	Charcot-Marie-Tooth disease, type 4K, 616684;Mitochondrial complex IV deficiency, nuclear type 1, 220110

SUZ12	100%	100%	100%	99.8%	Imagawa-Matsumoto syndrome, 618786
SV2A	100%	100%	100%	98.6%	Developmental and epileptic encephalopathy 113, 620772
SVBP	100%	100%	100%	99.9%	Neurodevelopmental disorder with ataxia, hypotonia, and microcephaly, 618569
SVIL	100%	100%	100%	99.5%	Myofibrillar myopathy 10, 619040
SYCE1	100%	100%	100%	99.1%	?Spermatogenic failure 15, 616950;?Premature ovarian failure 12, 616947
SYCP2	100%	100%	100%	99.8%	Spermatogenic failure 1, 258150
SYCP2L	100%	100%	100%	99.6%	Premature ovarian failure 24, 620840
SYCP3	100%	100%	100%	99.4%	Pregnancy loss, recurrent, 4, 270960;Spermatogenic failure 4, 270960
SYK	100%	100%	100%	99.8%	Immunodeficiency 82 with systemic inflammation, 619381
SYN1	100%	99.9%	97.7%	65.3%	Epilepsy, X-linked 1, with variable learning disabilities and behavior disorders, 300491;Intellectual developmental disorder, X-linked 50, 300115
SYNCRIP	92.9%	92.9%	100%	99.2%	

SYNE1	100%	100%	100%	99.6%	Arthrogryposis multiplex congenita 3, myogenic type, 618484;Emery-Dreifuss muscular dystrophy 4, autosomal dominant, 612998;Spinocerebellar ataxia, autosomal recessive 8, 610743
SYNE2	100%	100%	100%	99.7%	Emery-Dreifuss muscular dystrophy 5, autosomal dominant, 612999
SYNE4	100%	100%	100%	98.6%	Deafness, autosomal recessive 76, 615540
SYNGAP1	100%	99.9%	100%	97.8%	Intellectual developmental disorder, autosomal dominant 5, 612621
SYNJ1	100%	100%	100%	99.6%	Parkinson disease 20, early-onset, 615530;Developmental and epileptic encephalopathy 53, 617389
SYP	100%	99.5%	97.7%	68.4%	Intellectual developmental disorder, X-linked 96, 300802
SYT1	96%	96%	100%	99.7%	Baker-Gordon syndrome, 618218
SYT14	100%	100%	100%	99.5%	?Spinocerebellar ataxia, autosomal recessive 11, 614229

SYT2	100%	100%	100%	99.1%	Myasthenic syndrome, congenital, 7A, presynaptic, and distal motor neuropathy, autosomal dominant, 616040;Myasthenic syndrome, congenital, 7B, presynaptic, autosomal recessive, 619461
SZT2	100%	99.9%	100%	99.3%	Developmental and epileptic encephalopathy 18, 615476
TAB2	100%	100%	100%	98.8%	Congenital heart defects, nonsyndromic, 2, 614980
TAC3	100%	100%	100%	98.1%	Hypogonadotropic hypogonadism 10 with or without anosmia, 614839
TACO1	100%	100%	100%	99.6%	Mitochondrial complex IV deficiency, nuclear type 8, 619052
TACR3	100%	100%	100%	99.6%	Hypogonadotropic hypogonadism 11 with or without anosmia, 614840
TACSTD2	100%	100%	100%	98.1%	Corneal dystrophy, gelatinous drop-like, 204870
TAF1	98.7%	98.4%	98.7%	70.9%	Intellectual developmental disorder, X-linked syndromic 33, 300966;Dystonia-Parkinsonism, X-linked, 314250

TAF13	84.7%	76.3%	100%	99.9%	Intellectual developmental disorder, autosomal recessive 60, 617432
TAF1A	100%	100%	100%	99.8%	
TAF1C	100%	100%	100%	99.2%	
TAF2	96.3%	96.3%	100%	99.9%	Intellectual developmental disorder, autosomal recessive 40, 615599
TAF4	86.2%	80.7%	99.3%	82.7%	Intellectual developmental disorder, autosomal dominant 73, 620450
TAF4B	100%	100%	100%	99.3%	?Spermatogenic failure 13, 615841
TAF6	100%	100%	100%	98.5%	Alazami-Yuan syndrome, 617126
TAF8	85.3%	85.3%	100%	98.3%	Neurodevelopmental disorder with severe motor impairment, absent language, cerebral hypomyelination, and brain atrophy, 619972
TAFAZZIN	100%	99.6%	97.2%	66.7%	Barth syndrome, 302060
TAL1	100%	100%	100%	98.3%	Leukemia, T-cell acute lymphocytic, somatic, 613065
TAL2	100%	100%	100%	99.6%	Leukemia, T-cell acute lymphocytic, somatic, 613065
TALDO1	100%	100%	100%	97.6%	Transaldolase deficiency, 606003

TAMM41	100%	100%	100%	99.3%	Combined oxidative phosphorylation deficiency 56, 620139
TANC2	100%	100%	100%	99.5%	Intellectual developmental disorder with autistic features and language delay, with or without seizures, 618906
TANGO2	100%	100%	100%	99.5%	Metabolic encephalomyopathic crises, recurrent, with rhabdomyolysis, cardiac arrhythmias, and neurodegeneration, 616878
TAOK1	100%	100%	100%	99.6%	Developmental delay with or without intellectual impairment or behavioral abnormalities, 619575
TAOK2	100%	100%	100%	99%	
TAP1	99.7%	97.1%	100%	99.5%	MHC class I deficiency 1, 604571
TAP2	98.1%	97.9%	100%	98%	MHC class I deficiency 2, 620813
TAPBP	88.8%	88.8%	100%	98%	?MHC class I deficiency 3, 620814
TAPT1	100%	100%	100%	99.4%	Osteochondrodysplasia, complex lethal, Symoens-Barnes-Gistelinck type, 616897

TARDBP	100%	99.7%	100%	99.5%	Frontotemporal lobar degeneration, TARDBP-related, 612069;Amyotrophic lateral sclerosis 10, with or without FTD, 612069
TARS1	100%	100%	100%	99.5%	Trichothiodystrophy 7, nonphotosensitive, 618546
TARS2	100%	100%	100%	99.1%	Combined oxidative phosphorylation deficiency 21, 615918
TASP1	100%	100%	100%	99.8%	Suleiman-El-Hattab syndrome, 618950
TAT	100%	100%	100%	99.7%	Tyrosinemia, type II, 276600
TBC1D20	92.2%	92.2%	100%	99.5%	Warburg micro syndrome 4, 615663
TBC1D23	100%	100%	100%	99.8%	Pontocerebellar hypoplasia, type 11, 617695
TBC1D24	100%	100%	100%	99%	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

TBC1D2B	99.9%	99.2%	100%	99.3%	Neurodevelopmental disorder with seizures and gingival overgrowth, 619323
TBC1D32	100%	100%	100%	99.9%	
TBC1D7	100%	100%	100%	99.9%	Macrocephaly/megalencephaly syndrome, autosomal recessive, 248000
TBC1D8B	100%	99.8%	99.1%	75.4%	Nephrotic syndrome, type 20, 301028
TBCD	91.2%	90.5%	100%	99.2%	Encephalopathy, progressive, early-onset, with brain atrophy and thin corpus callosum, 617193
TBCE	100%	100%	100%	99.4%	Kenny-Caffey syndrome, type 1, 244460;Hypoparathyroidism-retardation-dysmorphism syndrome, 241410;Encephalopathy, progressive, with amyotrophy and optic atrophy, 617207
TBCK	100%	100%	100%	99.5%	Hypotonia, infantile, with psychomotor retardation and characteristic facies 3, 616900

TBK1	100%	100%	100%	99.9%	{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
TBL1X	100%	99.8%	98.9%	70.5%	Hypothyroidism, congenital, nongoitrous, 8, 301033
TBL1XR1	100%	100%	100%	99.5%	Intellectual developmental disorder, autosomal dominant 41, 616944;Pierpont syndrome, 602342
TBL1Y	50%	49.1%	48.8%	19.7%	?Deafness, Y-linked 2, 400047
TBP	100%	100%	100%	98.8%	Spinocerebellar ataxia 17, 607136;{Parkinson disease, susceptibility to}, 168600
TBPL2	100%	100%	100%	99.7%	
TBR1	100%	100%	100%	97.8%	Intellectual developmental disorder with autism and speech delay, 606053

TBX1	96.8%	93%	99.7%	89.1%	Tetralogy of Fallot, 187500;DiGeorge syndrome, 188400;Conotruncal anomaly face syndrome, 217095;Velocardiofacial syndrome, 192430
TBX15	100%	100%	100%	99.4%	Cousin syndrome, 260660
TBX18	100%	100%	100%	98.8%	Congenital anomalies of kidney and urinary tract 2, 143400
TBX19	100%	100%	100%	99.2%	Adrenocorticotropic hormone deficiency, 201400
TBX2	99.8%	97.6%	100%	97.7%	Vertebral anomalies and variable endocrine and T-cell dysfunction, 618223
TBX20	100%	100%	100%	99.1%	Atrial septal defect 4, 611363
TBX21	100%	100%	100%	98.3%	Asthma and nasal polyps, 208550;?Immunodeficiency 88, 619630;{Asthma, aspirin-induced, susceptibility to}, 208550
TBX22	100%	99.9%	98.8%	70%	Cleft palate with ankyloglossia, 303400;?Abruzzo-Erickson syndrome, 302905
TBX3	100%	100%	100%	97.8%	Ulnar-mammary syndrome, 181450

TBX4	100%	100%	100%	98.7%	Ischiocoxopodopatellar syndrome with or without pulmonary arterial hypertension, 147891;Amelia, posterior, with pelvic and pulmonary hypoplasia syndrome, 601360
TBX5	100%	100%	100%	99.1%	Holt-Oram syndrome, 142900
TBX6	100%	100%	100%	98.7%	Spondylocostal dysostosis 5, 122600
TBXA2R	99.8%	97.5%	100%	98.2%	{Bleeding disorder, platelet-type, 13, susceptibility to}, 614009
TBXAS1	100%	100%	100%	99.4%	Ghosal hematodiaphyseal syndrome, 231095
TBXT	100%	100%	100%	99.4%	Sacral agenesis with vertebral anomalies, 615709;{Neural tube defects, susceptibility to}, 182940
TCAP	100%	100%	100%	99.4%	Cardiomyopathy, hypertrophic, 25, 607487;Muscular dystrophy, limb-girdle, autosomal recessive 7, 601954
TCEAL1	100%	99.9%	99.6%	66.1%	Hijazi-Reis syndrome, 301094

TCF12	100%	100%	100%	99.8%	Craniosynostosis 3, 615314;Hypogonadotropic hypogonadism 26 with or without anosmia, 619718
TCF20	100%	100%	100%	99.5%	Developmental delay with variable intellectual impairment and behavioral abnormalities, 618430
TCF3	100%	100%	100%	99%	Agammaglobulinemia 8B, autosomal recessive, 619824;Agammaglobulinemia 8A, autosomal dominant, 616941
TCF4	100%	100%	100%	99.4%	Pitt-Hopkins syndrome, 610954;Corneal dystrophy, Fuchs endothelial, 3, 613267
TCF7L2	100%	99.9%	100%	97.5%	{Diabetes mellitus, type 2, susceptibility to}, 125853
TCHH	100%	100%	99.9%	96%	?Uncombable hair syndrome 3, 617252
TCIRG1	100%	100%	100%	99%	Osteopetrosis, autosomal recessive 1, 259700
TCN2	94.2%	94.2%	100%	99.1%	Transcobalamin II deficiency, 275350
TCOF1	100%	100%	100%	99.1%	Treacher Collins syndrome 1, 154500
TCP1	100%	100%	100%	99.5%	
TCTN1	98.7%	96.6%	100%	99.8%	Joubert syndrome 13, 614173

TCTN2	98.5%	98.5%	100%	99.5%	Joubert syndrome 24, 616654;?Meckel syndrome 8, 613885
TCTN3	100%	100%	100%	99.8%	Joubert syndrome 18, 614815;Orofaciodigital syndrome IV, 258860
TDGF1	100%	100%	100%	99.8%	
TDP1	100%	100%	100%	99.8%	?Spinocerebellar ataxia, autosomal recessive, with axonal neuropathy 1, 607250
TDP2	100%	100%	100%	99.7%	Spinocerebellar ataxia, autosomal recessive 23, 616949
TDRD12	100%	100%	100%	99.6%	
TDRD7	100%	100%	100%	99.7%	Cataract 36, 613887
TDRD9	100%	100%	100%	99.1%	?Spermatogenic failure 30, 618110
TDRKH	100%	100%	99.9%	98.1%	
TEAD1	100%	100%	100%	99.5%	Sveinsson chorioretinal atrophy, 108985
TECPR2	100%	100%	100%	99.4%	Neuropathy, hereditary sensory and autonomic, type IX, with developmental delay, 615031
TECR	100%	100%	100%	98.4%	Intellectual developmental disorder, autosomal recessive 14, 614020

TECRL	100%	100%	100%	99.7%	Ventricular tachycardia, catecholaminergic polymorphic, 3, 614021
TECTA	100%	100%	100%	99.5%	Deafness, autosomal dominant 8/12, 601543;Deafness, autosomal recessive 21, 603629
TEFM	100%	100%	100%	99.5%	Combined oxidative phosphorylation deficiency 58, 620451
TEK	100%	100%	100%	99.8%	Venous malformations, multiple cutaneous and mucosal, 600195;Glaucoma 3, primary congenital, E, 617272
TEKT3	100%	100%	100%	99.3%	Spermatogenic failure 81, 620277
TELO2	100%	99.8%	100%	99.2%	You-Hoover-Fong syndrome, 616954
TENM1	100%	99.7%	99%	72.8%	
TENM3	100%	100%	100%	99%	Microphthalmia, syndromic 15, 615145;?Microphthalmia/coloboma 9, 615145
TENM4	100%	100%	100%	99.3%	Essential tremor, hereditary, 5, 616736
TENT5A	100%	100%	100%	99.4%	Osteogenesis imperfecta, type XVIII, 617952
TENT5D	100%	100%	99.2%	78.1%	

TERB1	100%	100%	100%	99.5%	Spermatogenic failure 60, 619646
TERB2	100%	100%	100%	99.9%	?Spermatogenic failure 59, 619645
TERC					Pulmonary fibrosis and/or bone marrow failure syndrome, telomere-related, 2, 614743;Dyskeratosis congenita, autosomal dominant 1, 127550
TERF2IP	98.5%	91.1%	100%	99%	
TERT	100%	100%	100%	98.4%	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
TES	100%	100%	100%	99.8%	
TET2	100%	100%	100%	99.6%	Myelodysplastic syndrome, somatic, 614286;Immunodeficiency 75, 619126
TET3	100%	100%	100%	98.6%	Beck-Fahrner syndrome, 618798
TEX11	97.1%	96.9%	99.3%	75.3%	Spermatogenic failure, X-linked 2, 309120

TEX14	100%	100%	100%	99.7%	Spermatogenic failure 23, 617707
TEX15	100%	100%	100%	99.8%	Spermatogenic failure 25, 617960
TF	100%	100%	100%	99.2%	Atransferrinemia, 209300
TFAM	100%	100%	100%	99.5%	?Mitochondrial DNA depletion syndrome 15 (hepatocerebral type), 617156
TFAP2A	100%	100%	99.9%	97.8%	Branchiooculofacial syndrome, 113620
TFAP2B	100%	100%	100%	97.8%	Patent ductus arteriosus 2, 617035;Char syndrome, 169100
TFB2M	100%	100%	100%	99.7%	
TFE3	100%	99.4%	98.2%	67.5%	Intellectual developmental disorder, X-linked syndromic, with pigmentary mosaicism and coarse facies, 301066;Renal cell carcinoma, papillary, 1, 300854
TFG	100%	100%	100%	99.8%	?Spastic paraplegia 57, autosomal recessive, 615658;Hereditary motor and sensory neuropathy, Okinawa type, 604484
TFPT	100%	100%	100%	98.6%	
TFR2	100%	100%	100%	99%	Hemochromatosis, type 3, 604250

TFRC	95.5%	95.5%	100%	99.2%	Immunodeficiency 46, 616740
TG	100%	100%	100%	99%	{Autoimmune thyroid disease, susceptibility to, 3}, 608175;Thyroid dyshormonogenesis 3, 274700
TGDS	100%	100%	100%	99.7%	Catel-Manzke syndrome, 616145
TGFB1	100%	99.8%	100%	96.7%	Inflammatory bowel disease, immunodeficiency, and encephalopathy, 618213;Camurati-Engelmann disease, 131300;{Cystic fibrosis lung disease, modifier of}, 219700
TGFB2	100%	100%	100%	99.5%	Loeys-Dietz syndrome 4, 614816
TGFB3	100%	100%	100%	99.1%	Arrhythmogenic right ventricular dysplasia 1, 107970;Loeys-Dietz syndrome 5, 615582

TGFBI	100%	100%	100%	99.5%	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
TGFBR1	100%	99.8%	100%	99.1%	{Multiple self-healing squamous epithelioma, susceptibility to}, 132800;Loeys-Dietz syndrome 1, 609192
TGFBR2	100%	100%	100%	99.6%	Loeys-Dietz syndrome 2, 610168;Colorectal cancer, hereditary nonpolyposis, type 6, 614331;Esophageal cancer, somatic, 133239
TGIF1	100%	100%	100%	99.6%	Holoprosencephaly 4, 142946
TGM1	100%	100%	100%	99.2%	Ichthyosis, congenital, autosomal recessive 1, 242300
TGM3	100%	100%	100%	99.3%	?Uncombable hair syndrome 2, 617251
TGM5	100%	100%	100%	99.4%	Peeling skin syndrome 2, 609796

TGM6	100%	100%	100%	99.4%	Spinocerebellar ataxia 35, 613908
TH	100%	100%	100%	99.1%	Segawa syndrome, recessive, 605407
THAP1	100%	100%	100%	99.6%	Dystonia 6, torsion, 602629
THAP11	100%	100%	100%	97.4%	?Methylmalonic aciduria and homocystinuria, cblL type, 620940;Spinocerebellar ataxia 51, 620947
THBD	100%	100%	100%	96.6%	Thrombophilia 12 due to thrombomodulin defect, 614486;{Hemolytic uremic syndrome, atypical, susceptibility to, 6}, 612926
THBS1	100%	100%	100%	99.6%	
THBS2	100%	100%	100%	98.8%	?Ehlers-Danlos syndrome, classic-like, 3, 620865;{Lumbar disc herniation, susceptibility to}, 603932
THBS4	100%	100%	100%	99.4%	
THG1L	100%	100%	100%	99.7%	Spinocerebellar ataxia, autosomal recessive 28, 618800
THOC1	100%	100%	100%	99.9%	?Deafness, autosomal dominant 86, 620280

THOC2	100%	99.8%	99.2%	74.6%	Arthrogryposis multiplex congenita 7, X-linked, 301127;Intellectual developmental disorder, X-linked syndromic, Kumar type, 300957
THOC6	100%	100%	100%	98.8%	Beaulieu-Boycott-Innes syndrome, 613680
THPO	100%	100%	100%	99.6%	Thrombocythemia 1, 187950;Thrombocytopenia 9, 620478;Amegakaryocytic thrombocytopenia, congenital, 2, 620481
THRA	100%	100%	100%	99.1%	Hypothyroidism, congenital, nongoitrous, 6, 614450
THRB	100%	100%	100%	99.6%	Thyroid hormone resistance, autosomal recessive, 274300;Thyroid hormone resistance, 188570;Thyroid hormone resistance, selective pituitary, 145650
THSD1	100%	100%	100%	99.4%	?Aneurysm, intracranial berry, 12, 618734;Lymphatic malformation 13, 620244
THSD4	100%	100%	100%	98.9%	Aortic aneurysm, familial thoracic 12, 619825
THUMPD1	100%	99.7%	100%	99.6%	Neurodevelopmental disorder with speech delay and variable ocular anomalies, 619989

TIA1	100%	100%	100%	99.7%	Welander distal myopathy, 604454;Amyotrophic lateral sclerosis 26 with or without frontotemporal dementia, 619133
TIAM1	100%	100%	100%	99.3%	Neurodevelopmental disorder with language delay and seizures, 619908
TICAM1	100%	100%	100%	99.4%	{Encephalopathy, acute, infection-induced (herpes-specific), susceptibility to, 6}, 614850
TIE1	100%	100%	100%	99%	Lymphatic malformation 11, 619401
TIMELESS	100%	100%	100%	99.4%	?Advance sleep phase syndrome, familial, 4, 620015
TIMM22	100%	100%	100%	97.4%	?Combined oxidative phosphorylation deficiency 43, 618851
TIMM44	100%	100%	100%	98.1%	
TIMM50	100%	100%	100%	98.5%	3-methylglutaconic aciduria, type IX, 617698
TIMM8A	100%	99.5%	99.2%	71%	Mohr-Tranebjaerg syndrome, 304700
TIMMDC1	100%	100%	100%	99.4%	Mitochondrial complex I deficiency, nuclear type 31, 618251
TIMP3	100%	100%	100%	98.3%	Sorsby fundus dystrophy, 136900

TINF2	100%	100%	100%	99.4%	Dyskeratosis congenita, autosomal dominant 3, 613990;Revesz syndrome, 268130
TIRAP	100%	100%	100%	99.5%	{Malaria, protection against}, 611162;{Tuberculosis, protection against}, 607948;{Bacteremia, protection against}, 614382
TJP1	100%	100%	100%	99.6%	
TJP2	100%	100%	100%	99.6%	Hypercholanemia, familial 1, 607748;Cholestasis, progressive familial intrahepatic 4, 615878
TK2	100%	100%	100%	99.9%	Mitochondrial DNA depletion syndrome 2 (myopathic type), 609560;?Progressive external ophthalmoplegia with mitochondrial DNA deletions, autosomal recessive 3, 617069
TKFC	100%	100%	100%	98.9%	Triokinase and FMN cyclase deficiency syndrome, 618805
TKT	98.1%	98.1%	100%	99.1%	Short stature, developmental delay, and congenital heart defects, 617044
TKTL1	100%	98.1%	99.3%	70.8%	

TLCD3B	100%	100%	100%	98.7%	Cone-rod dystrophy 22, 619531
TLE6	100%	100%	100%	99%	Oocyte/zygote/embryo maturation arrest 15, 616814
TLK2	100%	100%	100%	99.5%	Intellectual developmental disorder, autosomal dominant 57, 618050
TLL1	100%	100%	100%	99.6%	Atrial septal defect 6, 613087
TLR3	100%	100%	100%	100%	{HIV1 infection, resistance to}, 609423;{Immunodeficiency 83, susceptibility to viral infections}, 613002
TLR4	100%	100%	100%	99.8%	
TLR5	100%	100%	100%	99.7%	{Melioidosis, susceptibility to}, 615557;{Systemic lupus erythematosus, susceptibility to, 1}, 601744;{Systemic lupus erythematosus, resistance to}, 601744;{Legionnaire disease, susceptibility to}, 608556
TLR7	100%	100%	99.1%	75.2%	Immunodeficiency 74, COVID19-related, X-linked, 301051;Systemic lupus erythematosus 17, 301080
TLR8	100%	100%	99.1%	75.4%	Immunodeficiency 98 with autoinflammation, X-linked, 301078

TMC1	100%	100%	100%	99.4%	Deafness, autosomal dominant 36, 606705;Deafness, autosomal recessive 7, 600974
TMC6	100%	100%	100%	98.8%	{Epidermodysplasia verruciformis, susceptibility to, 1}, 226400
TMC8	100%	100%	100%	99%	{Epidermodysplasia verruciformis, susceptibility to, 2}, 618231
TMCO1	87.7%	87.7%	100%	98.4%	Craniofacial dysmorphism, skeletal anomalies, and impaired intellectual development 1, 213980
TMCO3	100%	100%	100%	99.5%	
TMEM106B	100%	100%	100%	99.8%	Leukodystrophy, hypomyelinating, 16, 617964
TMEM107	100%	100%	100%	98%	Orofaciodigital syndrome XVI, 617563;Meckel syndrome 13, 617562;?Joubert syndrome 29, 617562
TMEM126A	100%	100%	100%	99.9%	Optic atrophy 7, 612989
TMEM126B	100%	100%	100%	99.7%	Mitochondrial complex I deficiency, nuclear type 29, 618250
TMEM127	100%	100%	100%	98.8%	{Pheochromocytoma, susceptibility to}, 171300

TMEM132E	100%	100%	100%	99.1%	Deafness, autosomal recessive 99, 618481
TMEM138	99.8%	96.8%	100%	99.2%	Joubert syndrome 16, 614465
TMEM147	100%	100%	100%	99.2%	Neurodevelopmental disorder with facial dysmorphism, absent language, and pseudo-Pelger-Huet anomaly, 620075
TMEM14C	100%	100%	100%	99.9%	
TMEM151A	100%	100%	100%	97.6%	Episodic kinesigenic dyskinesia 3, 620245
TMEM163	100%	100%	99.7%	96.7%	Leukodystrophy, hypomyelinating, 25, 620243
TMEM165	100%	100%	100%	99.1%	Congenital disorder of glycosylation, type IIk, 614727
TMEM184B	100%	100%	100%	99.3%	
TMEM186	100%	100%	100%	99.9%	
TMEM199	100%	100%	100%	99.4%	Congenital disorder of glycosylation, type IIp, 616829
TMEM216	100%	100%	100%	99.9%	Joubert syndrome 2, 608091;Retinitis pigmentosa 98, 620996;Meckel syndrome 2, 603194

TMEM218	100%	100%	100%	99.3%	Joubert syndrome 39, 619562
TMEM222	100%	100%	100%	99.6%	Neurodevelopmental disorder with motor and speech delay and behavioral abnormalities, 619470
TMEM231	93.3%	93.2%	100%	98.9%	Joubert syndrome 20, 614970;Meckel syndrome 11, 615397
TMEM237	98.2%	98.2%	100%	99.7%	Joubert syndrome 14, 614424
TMEM240	100%	100%	100%	95.1%	Spinocerebellar ataxia 21, 607454
TMEM260	100%	100%	100%	99.6%	Structural heart defects and renal anomalies syndrome, 617478
TMEM38B	100%	100%	100%	99.9%	Osteogenesis imperfecta, type XIV, 615066
TMEM43	100%	100%	100%	99%	Arrhythmogenic right ventricular dysplasia 5, 604400;Auditory neuropathy, autosomal dominant 3, 619832;Emery-Dreifuss muscular dystrophy 7, AD, 614302
TMEM53	100%	100%	100%	99.1%	Craniotubular dysplasia, Ikegawa type, 619727
TMEM63A	100%	100%	100%	99.2%	Leukodystrophy, hypomyelinating, 19, transient infantile, 618688

TMEM63B	100%	100%	100%	99.1%	
TMEM63C	100%	100%	100%	99%	Spastic paraplegia 87, autosomal recessive, 619966
TMEM65	100%	97.1%	99.9%	94.9%	
TMEM67	96.1%	96.1%	100%	99.8%	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
TMEM70	100%	100%	100%	99.3%	Mitochondrial complex V (ATP synthase) deficiency, nuclear type 2, 614052
TMEM94	100%	100%	100%	99.5%	Intellectual developmental disorder with cardiac defects and dysmorphic facies, 618316
TMEM98	100%	100%	100%	99.6%	Nanophthalmos 4, 615972
TMIE	100%	100%	100%	99.7%	Deafness, autosomal recessive 6, 600971
TMLHE	100%	99.7%	98.9%	78.6%	{Autism, susceptibility to, X-linked 6}, 300872
TMPO	100%	100%	100%	99.4%	
TMPRSS15	100%	100%	100%	99.7%	Enterokinase deficiency, 226200
TMPRSS3	100%	100%	100%	99.9%	Deafness, autosomal recessive 8/10, 601072

TMPRSS6	100%	100%	100%	99.1%	Iron-refractory iron deficiency anemia, 206200
TMTC2	97.3%	97.1%	100%	99.8%	
TMTC3	100%	100%	100%	99.7%	Lissencephaly 8, 617255
TMTC4	100%	100%	100%	99.6%	?Deafness, autosomal recessive 122, 620714
TMX2	100%	100%	100%	99.2%	Neurodevelopmental disorder with microcephaly, cortical malformations, and spasticity, 618730
TNC	100%	100%	100%	99.3%	Deafness, autosomal dominant 56, 615629
TNF	100%	100%	100%	98.6%	{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
TNFAIP3	100%	100%	100%	99.5%	Autoinflammatory syndrome, familial, Behcet-like 1, 616744
TNFRSF10B	100%	100%	100%	99.3%	Squamous cell carcinoma, head and neck, 275355

TNFRSF11A	99.9%	99.2%	100%	98.3%	Osteopetrosis, autosomal recessive 7, 612301;{Paget disease of bone 2, early-onset}, 602080;Osteolysis, familial expansile, 174810
TNFRSF11B	100%	100%	100%	99.8%	Paget disease of bone 5, juvenile-onset, 239000
TNFRSF13B	100%	100%	100%	98.6%	Immunodeficiency, common variable, 2, 240500;Immunoglobulin A deficiency 2, 609529
TNFRSF13C	100%	100%	100%	98.9%	Immunodeficiency, common variable, 4, 613494
TNFRSF1A	100%	100%	100%	98.7%	{Multiple sclerosis, susceptibility to, 5}, 614810;Periodic fever, familial, 142680
TNFRSF4	100%	100%	100%	98%	?Immunodeficiency 16, 615593
TNFRSF9	100%	100%	100%	99.7%	Immunodeficiency 109 with lymphoproliferation, 620282
TNFSF11	100%	100%	100%	98.6%	Osteopetrosis, autosomal recessive 2, 259710
TNFSF12	100%	100%	100%	99.2%	
TNFSF13	100%	99.9%	100%	98.7%	
TNIK	100%	100%	100%	99.5%	Intellectual developmental disorder, autosomal recessive 54, 617028

TNNC1	100%	100%	100%	99.3%	Cardiomyopathy, dilated, 1Z, 611879;Cardiomyopathy, hypertrophic, 13, 613243
TNNC2	100%	100%	100%	98.5%	Congenital myopathy 15, 620161
TNNI2	100%	100%	100%	98.9%	Arthrogryposis, distal, type 2B1, 601680
TNNI3	100%	100%	100%	97.5%	?Cardiomyopathy, dilated, 2A, 611880;Cardiomyopathy, hypertrophic, 7, 613690;Cardiomyopathy, familial restrictive, 1, 115210;Cardiomyopathy, dilated, 1FF, 613286
TNNI3K	100%	100%	100%	99.6%	Cardiac conduction disease with or without dilated cardiomyopathy, 616117
TNNT1	100%	100%	100%	98%	Nemaline myopathy 5C, autosomal dominant, 620389;Nemaline myopathy 5A, autosomal recessive, severe infantile, 605355;Nemaline myopathy 5B, autosomal recessive, childhood-onset, 620386

TNNT2	100%	100%	100%	99.1%	Cardiomyopathy, dilated, 1D, 601494;Cardiomyopathy, hypertrophic, 2, 115195;Cardiomyopathy, familial restrictive, 3, 612422;Left ventricular noncompaction 6, 601494
TNNT3	100%	100%	100%	98.7%	Arthrogryposis, distal, type 2B2, 618435
TNPO2	100%	100%	100%	98.4%	Intellectual developmental disorder with hypotonia, impaired speech, and dysmorphic facies, 619556
TNPO3	100%	100%	100%	99.7%	Muscular dystrophy, limb-girdle, autosomal dominant 2, 608423
TNR	100%	100%	100%	99.3%	Neurodevelopmental disorder, nonprogressive, with spasticity and transient opisthotonus, 619653
TNRC6A	100%	100%	100%	99.6%	?Epilepsy, familial adult myoclonic, 6, 618074
TNRC6B	100%	100%	100%	99.3%	Global developmental delay with speech and behavioral abnormalities, 619243
TNS1	100%	100%	100%	99.2%	
TNS2	100%	100%	100%	98.8%	

TNXB	100%	100%	100%	98.4%	Ehlers-Danlos syndrome, classic-like, 1, 606408;Vesicoureteral reflux 8, 615963
TOE1	100%	100%	100%	99.6%	Pontocerebellar hypoplasia, type 7, 614969
TOGARAM1	100%	100%	100%	99.7%	Joubert syndrome 37, 619185
TOM1	100%	100%	100%	99.3%	?Immunodeficiency 85 and autoimmunity, 619510
TOMM40L	100%	100%	100%	99.4%	
TOMM7	100%	99.9%	100%	99%	Garg-Mishra progeroid syndrome, 620601
TOMM70	100%	100%	100%	99.4%	
TONSL	100%	100%	100%	99.3%	Spondyloepimetaphyseal dysplasia, sponastrime type, 271510
TOP1	100%	100%	100%	99.6%	DNA topoisomerase I, camptothecin-resistant
TOP2A	100%	100%	100%	99.7%	DNA topoisomerase II, resistance to inhibition of, by amsacrine
TOP2B	100%	100%	100%	99.5%	B-cell immunodeficiency, distal limb anomalies, and urogenital malformations, 609296

TOP3A	100%	100%	100%	99.2%	Microcephaly, growth restriction, and increased sister chromatid exchange 2, 618097;Progressive external ophthalmoplegia with mitochondrial DNA deletions, autosomal recessive 5, 618098
TOPORS	100%	100%	100%	99.8%	Retinitis pigmentosa 31, 609923
TOR1A	93.4%	90.8%	100%	98.8%	{Dystonia-1, modifier of};Arthrogryposis multiplex congenita 5, 618947;Dystonia-1, torsion, 128100
TOR1AIP1	100%	100%	100%	99.1%	?Muscular dystrophy, autosomal recessive, with rigid spine and distal joint contractures, 617072

TP53	94.7%	94.7%	100%	98.8%	{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
TP53RK	100%	100%	100%	99.4%	Galloway-Mowat syndrome 4, 617730
TP63	100%	100%	100%	99.6%	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

TP73	100%	100%	100%	98.1%	Ciliary dyskinesia, primary, 47, and lissencephaly, 619466
TPCN2	100%	100%	100%	99.1%	[Skin/hair/eye pigmentation 10, blond/brown hair], 612267
TPI1	100%	100%	100%	99.2%	Hemolytic anemia due to triosephosphate isomerase deficiency, 615512
TPK1	100%	100%	100%	99.6%	Thiamine metabolism dysfunction syndrome 5 (episodic encephalopathy type), 614458
TPM1	100%	100%	100%	98.9%	Left ventricular noncompaction 9, 611878;Cardiomyopathy, hypertrophic, 3, 115196;Cardiomyopathy, dilated, 1Y, 611878
TPM2	100%	100%	100%	98.5%	Arthrogryposis, distal, type 2B4, 108120;Arthrogryposis, distal, type 1A, 108120;Congenital myopathy 23, 609285
TPM3	100%	100%	100%	99.6%	Congenital myopathy 4A, autosomal dominant, 255310;Congenital myopathy 4B, autosomal recessive, 609284
TPM4	100%	99.8%	100%	98.4%	Bleeding disorder, platelet-type, 25, 620486

TPMT	100%	99.8%	100%	99.9%	{Thiopurines, poor metabolism of, 1}, 610460
TPO	100%	100%	100%	99.2%	Thyroid dyshormonogenesis 2A, 274500
TPP1	100%	100%	100%	99.2%	Ceroid lipofuscinosis, neuronal, 2, 204500;Spinocerebellar ataxia, autosomal recessive 7, 609270
TPP2	100%	100%	100%	99.7%	Immunodeficiency 78 with autoimmunity and developmental delay, 619220
TPR	100%	100%	100%	99.3%	?Intellectual developmental disorder, autosomal recessive 79, 620393
TPRKB	84.4%	81.3%	100%	99.3%	Galloway-Mowat syndrome 5, 617731
TPRN	96.3%	93.7%	99.7%	89.8%	Deafness, autosomal recessive 79, 613307
TRA2B	100%	100%	100%	99.6%	
TRAC	100%	100%	100%	99.2%	Immunodeficiency 7, TCR-alpha/beta deficient, 615387
TRAF3	100%	100%	100%	97.9%	{?Encephalopathy, acute, infection-induced (herpes-specific), susceptibility to, 5}, 614849
TRAF3IP1	100%	100%	100%	98.7%	Senior-Loken syndrome 9, 616629

TRAF3IP2	99.7%	97.9%	100%	99.4%	?Candidiasis, familial, 8, 615527;{Psoriasis susceptibility 13}, 614070
TRAF6	100%	100%	100%	99.7%	
TRAF7	100%	100%	100%	98.5%	Cardiac, facial, and digital anomalies with developmental delay, 618164
TRAIP	100%	100%	100%	99.4%	Seckel syndrome 9, 616777
TRAK1	100%	100%	100%	98.9%	Developmental and epileptic encephalopathy 68, 618201
TRAPPC10	100%	100%	100%	99.4%	Neurodevelopmental disorder with microcephaly, short stature, and speech delay, 620027
TRAPPC11	100%	100%	100%	99.7%	Muscular dystrophy, limb-girdle, autosomal recessive 18, 615356
TRAPPC12	100%	100%	100%	98.5%	Encephalopathy, progressive, early-onset, with brain atrophy and spasticity, 617669
TRAPPC14	100%	100%	100%	99%	?Microcephaly 25, primary, autosomal recessive, 618351
TRAPPC2	100%	99.7%	99.4%	75.5%	Spondyloepiphyseal dysplasia tarda, 313400
TRAPPC2L	100%	100%	100%	99.6%	Encephalopathy, progressive, early-onset, with episodic rhabdomyolysis, 618331

TRAPPC4	100%	100%	100%	98.8%	Neurodevelopmental disorder with epilepsy, spasticity, and brain atrophy, 618741
TRAPPC6B	100%	100%	100%	99.9%	Neurodevelopmental disorder with microcephaly, epilepsy, and brain atrophy, 617862
TRAPPC9	100%	100%	100%	99.4%	Intellectual developmental disorder, autosomal recessive 13, 613192
TRDN	100%	100%	100%	99.7%	Cardiac arrhythmia syndrome, with or without skeletal muscle weakness, 615441
TREH	100%	100%	100%	99.3%	Trehalase deficiency, 612119
TREM2	100%	100%	100%	99.5%	{Alzhieimer disease 17, susceptibility to}, 615080;Polycystic lipomembranous osteodysplasia with sclerosing leukoencephalopathy 2, 618193

TREX1	100%	100%	100%	99.7%	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
TRH	100%	100%	100%	98.8%	Thyrotropin-releasing hormone deficiency, 275120
TRHR	100%	100%	100%	99.8%	Hypothyroidism, congenital, nongoitrous, 7, 618573
TRIM2	96.1%	96.1%	100%	99.7%	Charcot-Marie-Tooth disease, type 2R, 615490
TRIM22	100%	100%	100%	99.4%	
TRIM28	100%	100%	100%	98.9%	
TRIM32	100%	100%	100%	99.8%	?Bardet-Biedl syndrome 11, 615988;Muscular dystrophy, limb-girdle, autosomal recessive 8, 254110
TRIM36	100%	100%	100%	99.7%	?Anencephaly 1, 206500
TRIM37	98.3%	98.3%	100%	99.4%	Mulibrey nanism, 253250
TRIM44	100%	100%	100%	98.5%	?Aniridia 3, 617142
TRIM63	100%	100%	100%	99.4%	
TRIM71	100%	100%	100%	96.4%	Hydrocephalus, congenital, 4, 618667

TRIM8	100%	100%	100%	98%	Focal segmental glomerulosclerosis and neurodevelopmental syndrome, 619428
TRIO	99.2%	98.8%	100%	98.9%	Intellectual developmental disorder, autosomal dominant 44, with microcephaly, 617061;Intellectual developmental disorder, autosomal dominant 63, with macrocephaly, 618825
TRIOBP	100%	100%	100%	98.8%	Deafness, autosomal recessive 28, 609823
TRIP11	100%	100%	100%	99.7%	Odontochondrodysplasia 1, 184260;Achondrogenesis, type IA, 200600
TRIP12	100%	100%	100%	99.8%	Intellectual developmental disorder, autosomal dominant 49, 617752
TRIP13	100%	100%	100%	99.3%	Oocyte/zygote/embryo maturation arrest 9, 619011;Mosaic variegated aneuploidy syndrome 3, 617598
TRIP4	100%	100%	100%	99.7%	?Muscular dystrophy, congenital, Davignon-Chauveau type, 617066;Spinal muscular atrophy with congenital bone fractures 1, 616866

TRIT1	100%	100%	100%	99.5%	Combined oxidative phosphorylation deficiency 35, 617873
TRMT1	100%	100%	100%	98.7%	Intellectual developmental disorder, autosomal recessive 68, 618302
TRMT10A	100%	100%	100%	99.9%	Microcephaly, short stature, and impaired glucose metabolism 1, 616033
TRMT10C	100%	100%	100%	99.9%	Combined oxidative phosphorylation deficiency 30, 616974
TRMT5	100%	100%	100%	99.8%	Peripheral neuropathy with variable spasticity, exercise intolerance, and developmental delay, 616539
TRMU	100%	100%	100%	99.4%	{Deafness, mitochondrial, modifier of}, 580000;Liver failure, transient infantile, 613070
TRNT1	91.9%	91.8%	100%	99.9%	Sideroblastic anemia with B-cell immunodeficiency, periodic fevers, and developmental delay, 616084;Retinitis pigmentosa and erythrocytic microcytosis, 616959
TRPA1	100%	100%	100%	99.7%	?Episodic pain syndrome, familial, 1, 615040
TRPC3	100%	100%	100%	99.4%	?Spinocerebellar ataxia 41, 616410

TRPC6	100%	100%	100%	99.7%	Glomerulosclerosis, focal segmental, 2, 603965
TRPM1	100%	100%	100%	99.5%	Night blindness, congenital stationary (complete), 1C, autosomal recessive, 613216
TRPM3	97.8%	97.8%	100%	99.6%	?Cataract 50 with or without glaucoma, 620253;Neurodevelopmental disorder with hypotonia, dysmorphic facies, and skeletal anomalies, with or without seizures, 620224
TRPM4	100%	100%	100%	98.5%	Progressive familial heart block, type IB, 604559;Erythrokeratodermia variabilis et progressiva 6, 618531
TRPM6	100%	100%	100%	99.7%	Hypomagnesemia 1, intestinal, 602014
TRPM7	100%	100%	100%	99.7%	{Amyotrophic lateral sclerosis-parkinsonism/dementia complex, susceptibility to}, 105500
TRPM8	100%	100%	100%	99.3%	
TRPS1	100%	100%	100%	99.7%	Trichorhinophalangeal syndrome, type III, 190351;Trichorhinophalangeal syndrome, type I, 190350
TRPV1	100%	100%	100%	98%	

TRPV3	100%	100%	100%	98.8%	?Palmoplantar keratoderma, nonepidermolytic, focal 2, 616400;Olmsted syndrome 1, 614594
TRPV4	100%	100%	100%	98.4%	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
TRPV6	100%	100%	100%	99.3%	Hyperparathyroidism, transient neonatal, 618188

TRRAP	100%	100%	100%	99.2%	?Deafness, autosomal dominant 75, 618778;Developmental delay with or without dysmorphic facies and autism, 618454
TSC1	100%	100%	100%	99.3%	Focal cortical dysplasia, type II, somatic, 607341;Tuberous sclerosis-1, 191100;Lymphangioleiomyomatosis, 606690
TSC2	100%	100%	100%	99.4%	Lymphangioleiomyomatosis, somatic, 606690;?Focal cortical dysplasia, type II, somatic, 607341;Tuberous sclerosis-2, 613254
TSEN15	100%	100%	100%	99.5%	Pontocerebellar hypoplasia, type 2F, 617026
TSEN2	88.4%	88.4%	100%	99.1%	Pontocerebellar hypoplasia type 2B, 612389
TSEN34	100%	100%	100%	99.1%	?Pontocerebellar hypoplasia type 2C, 612390
TSEN54	100%	100%	100%	99%	Pontocerebellar hypoplasia type 2A, 277470;Pontocerebellar hypoplasia type 4, 225753;?Pontocerebellar hypoplasia type 5, 610204
TSFM	94.3%	94.3%	100%	98.9%	Combined oxidative phosphorylation deficiency 3, 610505

TSGA10	100%	100%	100%	99.7%	?Spermatogenic failure 26, 617961
TSHB	100%	100%	100%	99.9%	Hypothyroidism, congenital, nongoitrous 4, 275100
TSHR	100%	100%	100%	99.5%	Hyperthyroidism, familial gestational, 603373;Hyperthyroidism, nonautoimmune, 609152;Thyroid adenoma, hyperfunctioning, somatic, 609152;Hypothyroidism, congenital, nongoitrous, 1, 275200;Thyroid carcinoma with thyrotoxicosis, somatic, 609152
TSHZ1	100%	100%	100%	98.7%	Aural atresia, congenital, 607842
TSPAN12	100%	100%	100%	99.6%	Exudative vitreoretinopathy 5, 613310
TSPAN7	100%	99.6%	99.8%	74.5%	Intellectual developmental disorder, X-linked 58, 300210
TSPEAR	100%	100%	100%	97.9%	Tooth agenesis, selective, 10, 620173;?Deafness, autosomal recessive 98, 614861;Ectodermal dysplasia 14, hair/tooth type with or without hypohidrosis, 618180
TSPOAP1	100%	100%	100%	98.7%	Dystonia 22, juvenile-onset, 620453;?Dystonia 22, adult-onset, 620456

TSPYL1	100%	100%	100%	99.3%	Sudden infant death with dysgenesis of the testes syndrome, 608800
TSR2	100%	100%	99.4%	71.2%	?Diamond-Blackfan anemia 14 with mandibulofacial dysostosis, 300946
TTBK2	100%	100%	100%	99.6%	Spinocerebellar ataxia 11, 604432
TTC12	100%	100%	100%	99.5%	Ciliary dyskinesia, primary, 45, 618801
TTC19	100%	100%	100%	99.6%	Mitochondrial complex III deficiency, nuclear type 2, 615157
TTC21A	100%	100%	100%	99.5%	Spermatogenic failure 37, 618429
TTC21B	98.3%	97.8%	100%	99.8%	Short-rib thoracic dysplasia 4 with or without polydactyly, 613819;Nephronophthisis 12, 613820
TTC29	100%	100%	100%	100%	Spermatogenic failure 42, 618745
TTC5	100%	100%	100%	99.3%	Neurodevelopmental disorder with cerebral atrophy and variable facial dysmorphism, 619244
TTC7A	100%	100%	100%	98.8%	Gastrointestinal defects and immunodeficiency syndrome, 243150

TTC8	100%	100%	100%	99.8%	Bardet-Biedl syndrome 8, 615985;?Retinitis pigmentosa 51, 613464
TTI1	100%	100%	100%	99.3%	Neurodevelopmental disorder with microcephaly and movement abnormalities, 620445
TTI2	100%	100%	100%	99.5%	Intellectual developmental disorder, autosomal recessive 39, 615541
TTLL5	100%	100%	100%	99.4%	Cone-rod dystrophy 19, 615860
TTN	100%	100%	100%	99.6%	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
TTPA	100%	100%	100%	99%	Ataxia with isolated vitamin E deficiency, 277460

TTR	100%	100%	100%	99.6%	Amyloidosis, hereditary, transthyretin-related, 105210;Carpal tunnel syndrome, familial, 115430;[Dystransthyretinemic hyperthyroxinemia], 145680
TUB	100%	100%	100%	99.1%	?Retinal dystrophy and obesity, 616188
TUBA1A	100%	100%	100%	99.1%	Lissencephaly 3, 611603
TUBA3D	100%	100%	100%	98.7%	Keratoconus 9, 617928
TUBA4A	100%	100%	100%	99.2%	Amyotrophic lateral sclerosis 22 with or without frontotemporal dementia, 616208
TUBA8	95.2%	95%	100%	99.2%	Macrothrombocytopenia, isolated, 2, autosomal dominant, 619840
TUBB	99.8%	99%	100%	99.2%	Symmetric circumferential skin creases, congenital, 1, 156610;Cortical dysplasia, complex, with other brain malformations 6, 615771
TUBB1	100%	100%	100%	99.3%	Macrothrombocytopenia, isolated, 1, autosomal dominant, 613112
TUBB2A	100%	100%	100%	98.3%	Cortical dysplasia, complex, with other brain malformations 5, 615763

TUBB2B	100%	100%	100%	99.2%	Cortical dysplasia, complex, with other brain malformations 7, 610031
TUBB3	100%	100%	100%	98.9%	Fibrosis of extraocular muscles, congenital, 3A, 600638;Cortical dysplasia, complex, with other brain malformations 1, 614039
TUBB4A	99.4%	96.9%	100%	98.7%	Dystonia 4, torsion, autosomal dominant, 128101;Leukodystrophy, hypomyelinating, 6, 612438
TUBB4B	100%	100%	100%	99.5%	Leber congenital amaurosis with early-onset deafness, 617879
TUBB6	100%	100%	100%	99.6%	?Facial palsy, congenital, with ptosis and velopharyngeal dysfunction, 617732
TUBB8	100%	100%	100%	99.5%	Oocyte/zygote/embryo maturation arrest 2, 616780
TUBG1	100%	100%	100%	99%	Cortical dysplasia, complex, with other brain malformations 4, 615412
TUBGCP2	97%	97%	100%	99.3%	Pachygyria, microcephaly, developmental delay, and dysmorphic facies, with or without seizures, 618737
TUBGCP4	100%	99.9%	99.7%	97%	Microcephaly and chorioretinopathy, autosomal recessive, 3, 616335

TUBGCP6	100%	100%	100%	99%	Microcephaly and chorioretinopathy, autosomal recessive, 1, 251270
TUFM	100%	100%	100%	98.3%	Combined oxidative phosphorylation deficiency 4, 610678
TUFT1	100%	100%	100%	98.9%	Woolly hair-skin fragility syndrome, 620415
TULP1	100%	100%	100%	99%	Leber congenital amaurosis 15, 613843;Retinitis pigmentosa 14, 600132
TULP3	100%	100%	100%	99.5%	Hepatorenocardiac degenerative fibrosis, 619902
TUSC3	100%	100%	100%	99.6%	Intellectual developmental disorder, autosomal recessive 7, 611093
TWIST1	100%	99.9%	100%	98.3%	Craniosynostosis 1, 123100;Robinow-Sorauf syndrome, 180750;Sweeney-Cox syndrome, 617746;Saethre-Chotzen syndrome with or without eyelid anomalies, 101400
TWIST2	100%	100%	100%	97.3%	Ablepharon-macrostomia syndrome, 200110;Barber-Say syndrome, 209885;Focal facial dermal dysplasia 3, Setleis type, 227260

TWNK	100%	100%	100%	99.5%	Mitochondrial DNA depletion syndrome 7 (hepatocerebral type), 271245;Progressive external ophthalmoplegia with mitochondrial DNA deletions, autosomal dominant 3, 609286;Perrault syndrome 5, 616138
TXN2	100%	100%	100%	99.8%	?Combined oxidative phosphorylation deficiency 29, 616811
TXNDC15	100%	100%	100%	99.4%	Meckel syndrome 14, 619879
TXNL4A	100%	100%	100%	99%	Burn-McKeown syndrome, 608572
TXNRD2	100%	100%	100%	98.8%	?Glucocorticoid deficiency 5, 617825
TYK2	100%	100%	100%	98.1%	Immunodeficiency 35, 611521
TYMP	100%	100%	100%	98.8%	Mitochondrial DNA depletion syndrome 1 (MNGIE type), 603041
TYMS	100%	100%	100%	98.9%	Dyskeratosis congenita, digenic, 620040

TYR	100%	100%	100%	99.9%	[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
TYROBP	100%	100%	100%	98.9%	Polycystic lipomembranous osteodysplasia with sclerosing leukoencephalopathy 1, 221770
TYRP1	100%	100%	100%	99.6%	[Skin/hair/eye pigmentation, variation in, 11 (Melanesian blond hair)], 612271;Albinism, oculocutaneous, type III, 203290
U2AF2	100%	99.9%	100%	98.2%	Developmental delay, dysmorphic facies, and brain anomalies, 620535
UBA1	99.9%	99.3%	98.7%	68.8%	Spinal muscular atrophy, X-linked 2, infantile, 301830;VEXAS syndrome, somatic, 301054
UBA2	100%	100%	100%	99.7%	ACCES syndrome, 619959

UBA5	99.6%	97.2%	100%	99.9%	?Spinocerebellar ataxia, autosomal recessive 24, 617133;Developmental and epileptic encephalopathy 44, 617132
UBAP1	100%	100%	100%	98.7%	Spastic paraplegia 80, autosomal dominant, 618418
UBAP1L	100%	100%	100%	99.3%	
UBAP2L	100%	100%	100%	99.4%	Neurodevelopmental disorder with impaired language, behavioral abnormalities, and dysmorphic facies, 620494
UBB	100%	100%	100%	99.5%	
UBE2A	95%	89.5%	99.5%	71.2%	Intellectual developmental disorder, X-linked syndromic, Nascimento type, 300860
UBE2T	100%	100%	100%	99%	Fanconi anemia, complementation group T, 616435
UBE3A	100%	100%	100%	99.8%	Angelman syndrome, 105830
UBE3B	100%	100%	100%	99.4%	Kaufman oculocerebrofacial syndrome, 244450
UBE3C	100%	100%	100%	99.6%	Neurodevelopmental disorder with absent speech and movement and behavioral abnormalities, 620270

UBE4A	100%	100%	100%	99.5%	Neurodevelopmental disorder with hypotonia and gross motor and speech delay, 619639
UBIAD1	100%	100%	100%	99.8%	Corneal dystrophy, Schnyder type, 121800
UBQLN2	100%	100%	99.3%	76.6%	Amyotrophic lateral sclerosis 15, with or without frontotemporal dementia, 300857
UBR1	98%	98%	100%	99.6%	Johanson-Blizzard syndrome, 243800
UBR2	100%	99.7%	99.7%	99%	
UBR5	100%	100%	100%	99.7%	
UBR7	100%	100%	100%	99.7%	Li-Campeau syndrome, 619189
UBTF	100%	100%	100%	98.8%	Neurodegeneration, childhood-onset, with brain atrophy, 617672
UCHL1	100%	100%	100%	99.7%	{?Parkinson disease 5, susceptibility to}, 613643;Spastic paraplegia 79A, autosomal dominant, 620221;Spastic paraplegia 79B, autosomal recessive, 615491
UFC1	100%	100%	100%	99.1%	Neurodevelopmental disorder with spasticity and poor growth, 618076

UFM1	100%	100%	100%	99.6%	Leukodystrophy, hypomyelinating, 14, 617899
UFSP2	100%	100%	100%	99.6%	?Hip dysplasia, Beukes type, 142669;Spondyloepimetaphyseal dysplasia, Di Rocco type, 617974;Developmental and epileptic encephalopathy 106, 620028
UGCG	100%	100%	100%	99.8%	
UGDH	100%	100%	100%	99.6%	Developmental and epileptic encephalopathy 84, 618792
UGP2	98.5%	96.7%	100%	99.7%	Developmental and epileptic encephalopathy 83, 618744
UGT1A1	100%	100%	100%	99.3%	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
UMOD	100%	100%	100%	99.7%	Tubulointerstitial kidney disease, autosomal dominant, 1, 162000
UMPS	100%	100%	100%	99.7%	Orotic aciduria, 258900
UNC119	100%	100%	100%	97.9%	Cone-rod dystrophy 24, 620342;?Immunodeficiency 13, 615518

UNC13A	100%	100%	100%	98.7%	
UNC13D	100%	100%	100%	99.2%	Hemophagocytic lymphohistiocytosis, familial, 3, 608898
UNC45A	100%	100%	100%	99.3%	Osteootohepatoenteric syndrome, 619377
UNC45B	100%	100%	100%	99.1%	?Cataract 43, 616279;Myofibrillar myopathy 11, 619178
UNC79	100%	100%	100%	99.6%	
UNC80	100%	100%	100%	99.6%	Hypotonia, infantile, with psychomotor retardation and characteristic facies 2, 616801
UNC93B1	100%	99.6%	100%	95.9%	{Encephalopathy, acute, infection-induced (herpes-specific), susceptibility to, 1}, 610551
UNG	96.5%	96.4%	100%	99%	Immunodeficiency with hyper IgM, type 5, 608106
UPB1	100%	100%	100%	99.2%	Beta-ureidopropionase deficiency, 613161
UPF1	99.7%	98.8%	100%	98.9%	
UPF3B	100%	99.4%	99.1%	71.3%	Intellectual developmental disorder, X-linked syndromic 14, 300676
UPK3A	100%	100%	100%	99.1%	
UQCC1	100%	100%	100%	99.7%	

UQCC2	100%	100%	100%	98.5%	Mitochondrial complex III deficiency, nuclear type 7, 615824
UQCC3	100%	100%	100%	99.4%	?Mitochondrial complex III deficiency, nuclear type 9, 616111
UQCR10	100%	100%	100%	98.6%	
UQCR11	100%	100%	100%	97.5%	
UQCRB	100%	100%	100%	99.7%	Mitochondrial complex III deficiency, nuclear type 3, 615158
UQCRC1	100%	100%	100%	99.5%	Parkinsonism with polyneuropathy, 619279
UQCRC2	100%	100%	100%	99.7%	Mitochondrial complex III deficiency, nuclear type 5, 615160
UQCRFS1	100%	100%	100%	99.1%	Mitochondrial complex III deficiency, nuclear type 10, 618775
UQCRH	100%	100%	100%	99.6%	?Mitochondrial complex III deficiency, nuclear type 11, 620137
UQCRQ	100%	100%	100%	100%	Mitochondrial complex III deficiency, nuclear type 4, 615159
UROC1	100%	100%	100%	99.1%	?Urocanase deficiency, 276880

UROD	100%	100%	100%	99.6%	Porphyria, hepatoerythropoietic, 176100;Porphyria cutanea tarda, 176100
UROS	100%	100%	100%	99.6%	Porphyria, congenital erythropoietic, 263700
USB1	95.5%	93.2%	100%	99.2%	Poikiloderma with neutropenia, 604173
USH1C	100%	100%	100%	98.9%	Usher syndrome, type 1C, 276904;Deafness, autosomal recessive 18A, 602092
USH1G	100%	100%	100%	99.1%	Usher syndrome, type 1G, 606943
USH2A	100%	100%	100%	99.6%	Usher syndrome, type 2A, 276901;Retinitis pigmentosa 39, 613809
USP14	100%	100%	100%	99.7%	
USP18	100%	100%	100%	98.7%	Pseudo-TORCH syndrome 2, 617397
USP26	100%	100%	99.6%	74.8%	Spermatogenic failure, X-linked, 6, 301101
USP27X	100%	100%	98.6%	70.8%	Intellectual developmental disorder, X-linked 105, 300984
USP45	100%	100%	100%	99.9%	?Leber congenital amaurosis 19, 618513
USP48	100%	100%	100%	99.5%	Deafness, autosomal dominant 85, 620227

USP53	100%	100%	100%	99.5%	Cholestasis, progressive familial intrahepatic, 7, with or without hearing loss, 619658
USP7	100%	99.7%	100%	98.1%	Hao-Fountain syndrome, 616863
USP8	100%	100%	100%	99.6%	Pituitary adenoma 4, ACTH-secreting, somatic, 219090
USP9X	100%	99.8%	99.3%	75%	Intellectual developmental disorder, X-linked 99, 300919;Intellectual developmental disorder, X-linked 99, syndromic, female-restricted, 300968
USP9Y	50%	49.6%	49.4%	25.7%	Spermatogenic failure, Y-linked, 2, 415000
UVSSA	100%	100%	100%	98.6%	UV-sensitive syndrome 3, 614640
VAC14	100%	100%	100%	99.4%	Striatonigral degeneration, childhood-onset, 617054
VAMP1	100%	100%	100%	99.8%	Myasthenic syndrome, congenital, 25, 618323;Spastic ataxia 1, autosomal dominant, 108600
VAMP2	100%	100%	100%	97.5%	Neurodevelopmental disorder with hypotonia and autistic features with or without hyperkinetic movements, 618760

VANGL1	100%	100%	100%	99.9%	{Neural tube defects, susceptibility to}, 182940;Caudal regression syndrome, 600145
VANGL2	100%	99.7%	100%	98.5%	Neural tube defects, 182940
VAPB	100%	100%	100%	99.5%	Spinal muscular atrophy, late-onset, Finkel type, 182980;Amyotrophic lateral sclerosis 8, 608627
VARS1	100%	100%	100%	98.6%	Neurodevelopmental disorder with microcephaly, seizures, and cortical atrophy, 617802
VARS2	100%	100%	100%	98.6%	Combined oxidative phosphorylation deficiency 20, 615917
VAV1	98.3%	98.3%	100%	98.6%	
VAX1	100%	98.1%	100%	96.6%	?Microphthalmia, syndromic 11, 614402
VCAN	100%	100%	100%	99.8%	Wagner syndrome 1, 143200
VCL	100%	100%	100%	99.6%	Cardiomyopathy, dilated, 1W, 611407;Cardiomyopathy, hypertrophic, 15, 613255

VCP	100%	100%	100%	99.6%	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
VDR	100%	100%	100%	99.2%	Rickets, vitamin D-resistant, type IIA, 277440
VEGFC	100%	100%	100%	99.5%	Lymphatic malformation 4, 615907
VEZF1	100%	100%	100%	98.3%	?Cardiomyopathy, dilated, 1OO, 620247
VHL	88%	88%	100%	99.1%	Hemangioblastoma, cerebellar, somatic;Erythrocytosis, familial, 2, 263400;von Hippel-Lindau syndrome, 193300;Renal cell carcinoma, somatic, 144700;Pheochromocytoma, 171300
VIM	100%	100%	100%	99.1%	Cataract 30, pulverulent, 116300
VIPAS39	100%	100%	100%	99.7%	Arthrogryposis, renal dysfunction, and cholestasis 2, 613404

VKORC1	98.5%	94.3%	100%	98.6%	Vitamin K-dependent clotting factors, combined deficiency of, 2, 607473;Warfarin resistance, 122700
VLDLR	100%	100%	100%	99.4%	Cerebellar hypoplasia, impaired intellectual development, and dysequilibrium syndrome 1, 224050
VMA21	100%	99.9%	97.8%	66.4%	Myopathy, X-linked, with excessive autophagy, 310440
VPS11	100%	100%	100%	99.2%	?Dystonia 32, 619637;Leukodystrophy, hypomyelinating, 12, 616683
VPS13A	100%	100%	100%	99.8%	Choreoacanthocytosis, 200150
VPS13B	100%	100%	100%	99.7%	Cohen syndrome, 216550
VPS13C	100%	100%	100%	99.8%	Parkinson disease 23, autosomal recessive, early onset, 616840
VPS13D	100%	100%	100%	99.7%	Spinocerebellar ataxia, autosomal recessive 4, 607317
VPS16	100%	100%	100%	99.3%	Dystonia 30, 619291
VPS33A	89.5%	89.5%	100%	99.4%	Mucopolysaccharidosis-plus syndrome, 617303

VPS33B	100%	100%	100%	99.4%	Keratoderma-ichthyosis-deafness syndrome, autosomal recessive, 620009;Cholestasis, progressive familial intrahepatic, 12, 620010;Arthrogryposis, renal dysfunction, and cholestasis 1, 208085
VPS35	100%	100%	100%	99.4%	{Parkinson disease 17}, 614203
VPS35L	100%	100%	100%	99.4%	Ritscher-Schinzel syndrome 3, 619135
VPS37A	100%	100%	100%	99.8%	Spastic paraplegia 53, autosomal recessive, 614898
VPS41	100%	100%	100%	99.7%	Spinocerebellar ataxia, autosomal recessive 29, 619389
VPS45	87.3%	86.8%	100%	99.6%	Neutropenia, severe congenital, 5, autosomal recessive, 615285
VPS4A	100%	100%	100%	96.7%	CIMDAG syndrome, 619273
VPS50	100%	100%	100%	99.9%	Neurodevelopmental disorder with microcephaly, seizures, and neonatal cholestasis, 619685
VPS51	100%	100%	100%	98.8%	Pontocerebellar hypoplasia, type 13, 618606
VPS53	83%	81.7%	100%	99.4%	Pontocerebellar hypoplasia, type 2E, 615851

VRK1	99.3%	97.7%	100%	100%	Pontocerebellar hypoplasia type 1A, 607596;Neuronopathy, distal hereditary motor, autosomal recessive 10, 620542
VSX1	100%	100%	100%	99.3%	?Craniofacial anomalies and anterior segment dysgenesis syndrome, 614195;Keratoconus 1, 148300
VSX2	100%	100%	100%	99.5%	Microphthalmia, isolated 2, 610093;Microphthalmia/coloboma 3, 610092
VTN	100%	100%	100%	99.1%	
VWA1	99.9%	99.8%	100%	99%	Neuronopathy, distal hereditary motor, autosomal recessive 7, 619216
VWA3B	100%	100%	100%	99.4%	?Spinocerebellar ataxia, autosomal recessive 22, 616948
VWA8	100%	100%	100%	99.5%	?Retinitis pigmentosa 97, 620422
VWF	100%	100%	100%	99.3%	von Willebrand disease, type 1, 193400;von Willebrand disease, types 2A, 2B, 2M, and 2N, 613554;von Willebrand disease, type 3, 277480
WAC	94%	94%	100%	98.6%	Desanto-Shinawi syndrome, 616708

WARS1	100%	100%	100%	99.4%	Neuronopathy, distal hereditary motor, autosomal dominant 9, 617721;Neurodevelopmental disorder with microcephaly and speech delay, with or without brain abnormalities, 620317
WARS2	100%	100%	100%	99.4%	Parkinsonism-dystonia 3, childhood-onset, 619738;Neurodevelopmental disorder, mitochondrial, with abnormal movements and lactic acidosis, with or without seizures, 617710
WAS	97.7%	89.9%	97.7%	65.4%	Wiskott-Aldrich syndrome, 301000;Neutropenia, severe congenital, X-linked, 300299;Thrombocytopenia, X-linked, intermittent, 313900;Thrombocytopenia, X-linked, 313900
WASF1	100%	100%	100%	99.6%	Neurodevelopmental disorder with absent language and variable seizures, 618707
WASHC4	100%	100%	100%	99.8%	Intellectual developmental disorder, autosomal recessive 43, 615817
WASHC5	100%	100%	100%	99.7%	Ritscher-Schinzel syndrome 1, 220210;Spastic paraplegia 8, autosomal dominant, 603563

WBP11	100%	99.9%	100%	99.8%	Vertebral, cardiac, tracheoesophageal, renal, and limb defects, 619227
WBP2	100%	100%	100%	99%	Deafness, autosomal recessive 107, 617639
WBP4	100%	100%	100%	99.6%	Neurodevelopemental disorder with hypotonia, feeding difficulties, facial dysmorphism, and brain abnormalities, 620852
WDFY3	100%	100%	100%	99.7%	?Microcephaly 18, primary, autosomal dominant, 617520
WDPCP	97.7%	97.7%	100%	99.8%	Bardet-Biedl syndrome 15, 615992;Congenital heart defects, hamartomas of tongue, and polysyndactyly, 217085
WDR1	100%	100%	100%	98.7%	Periodic fever, immunodeficiency, and thrombocytopenia syndrome, 150550
WDR11	100%	100%	100%	99.7%	Intellectual developmental disorder, autosomal recessive 78, 620237;Hypogonadotropic hypogonadism 14 with or without anosmia, 614858
WDR13	100%	99.1%	98%	69.2%	

WDR19	100%	100%	100%	99.7%	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
WDR26	93.8%	93.8%	100%	99.2%	Skraban-Deardorff syndrome, 617616
WDR35	100%	100%	100%	99.6%	Short-rib thoracic dysplasia 7 with or without polydactyly, 614091;Cranioectodermal dysplasia 2, 613610
WDR36	100%	100%	100%	99.5%	Glaucoma 1, open angle, G, 609887
WDR37	90.3%	89.9%	100%	99.7%	Neurooculocardiogenitourinary syndrome, 618652
WDR4	100%	100%	100%	99.1%	Galloway-Mowat syndrome 6, 618347;Microcephaly, growth deficiency, seizures, and brain malformations, 618346
WDR44	100%	99.8%	99.1%	73.4%	
WDR45	100%	99.6%	98.1%	67.7%	Neurodegeneration with brain iron accumulation 5, 300894

WDR45B	100%	100%	100%	99.5%	Neurodevelopmental disorder with spastic quadriplegia and brain abnormalities with or without seizures, 617977
WDR47	100%	100%	100%	99.9%	
WDR5	100%	100%	100%	99.4%	
WDR62	98.8%	98.7%	100%	99.3%	Microcephaly 2, primary, autosomal recessive, with or without cortical malformations, 604317
WDR72	96.8%	96.8%	100%	99.6%	Amelogenesis imperfecta, type IIA3, 613211
WDR73	100%	100%	100%	99.2%	Galloway-Mowat syndrome 1, 251300
WDR81	100%	100%	100%	99.2%	Cerebellar ataxia, impaired intellectual development, and dysquilibrium syndrome 2, 610185;Hydrocephalus, congenital, 3, with brain anomalies, 617967
WDR83OS	100%	100%	100%	99.4%	
WEE2	100%	100%	100%	99.7%	Oocyte/zygote/embryo maturation arrest 5, 617996
WFDC2	100%	100%	100%	99.2%	Bronchiectasis and nasal polyposis, 620984

WFS1	91.2%	91.2%	100%	99.1%	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
WHRN	100%	100%	100%	98.5%	Deafness, autosomal recessive 31, 607084;Usher syndrome, type 2D, 611383
WIPF1	100%	100%	100%	98.8%	Wiskott-Aldrich syndrome 2, 614493
WIPI2	100%	100%	100%	98%	?Intellectual developmental disorder with short stature and variable skeletal anomalies, 618453
WLS	100%	100%	100%	99.7%	Zaki syndrome, 619648
WNK1	100%	100%	100%	98.9%	Neuropathy, hereditary sensory and autonomic, type II, 201300;Pseudohypoaldosteronism, type IIC, 614492
WNK3	100%	99.8%	99.2%	74.1%	Prieto syndrome, 309610
WNK4	100%	100%	100%	98%	Pseudohypoaldosteronism, type IIB, 614491

WNT1	100%	100%	100%	98.5%	{Osteoporosis, early-onset, susceptibility to, autosomal dominant}, 615221;Osteogenesis imperfecta, type XV, 615220
WNT10A	100%	100%	100%	98.9%	Schopf-Schulz-Passarge syndrome, 224750;Tooth agenesis, selective, 4, 150400;Ectodermal dysplasia 16 (odontoonychodermal dysplasia), 257980
WNT10B	100%	100%	100%	99.3%	Tooth agenesis, selective, 8, 617073;Split-hand/foot malformation 6, 225300
WNT2B	100%	100%	100%	99.2%	Diarrhea 9, 618168
WNT3	100%	99.8%	100%	99.4%	?Tetra-amelia syndrome 1, 273395
WNT4	100%	97.3%	100%	97.5%	?SERKAL syndrome, 611812;Mullerian aplasia and hyperandrogenism, 158330
WNT5A	100%	100%	99.9%	96.2%	Robinow syndrome, autosomal dominant 1, 180700
WNT6	100%	100%	100%	98.2%	
WNT7A	100%	100%	100%	99.5%	Fuhrmann syndrome, 228930;Ulna and fibula, absence of, with severe limb deficiency, 276820

WRAP53	100%	100%	100%	98.8%	Dyskeratosis congenita, autosomal recessive 3, 613988
WRN	100%	100%	100%	99.8%	Werner syndrome, 277700
WT1	100%	100%	100%	98%	Mesothelioma, somatic, 156240;Meacham syndrome, 608978;Frasier syndrome, 136680;Nephrotic syndrome, type 4, 256370;Denys-Drash syndrome, 194080;Wilms tumor, type 1, 194070
WWOX	100%	100%	100%	99.5%	Esophageal squamous cell carcinoma, somatic, 133239;Developmental and epileptic encephalopathy 28, 616211;Spinocerebellar ataxia, autosomal recessive 12, 614322
XDH	100%	100%	100%	99.6%	Xanthinuria, type I, 278300
XIAP	100%	99.9%	99.2%	74%	Lymphoproliferative syndrome, X-linked, 2, 300635
XIRP2	100%	100%	100%	99.7%	
XIST					X-inactivation, familial skewed, 300087
XK	100%	98.9%	99.2%	70.9%	McLeod syndrome, 300842
XKR8	100%	100%	100%	99.1%	
XKRY					

XKRYP7					
XPA	100%	100%	100%	99.9%	Xeroderma pigmentosum, group A, 278700
XPC	100%	100%	100%	99.3%	Xeroderma pigmentosum, group C, 278720
XPNPEP3	100%	100%	100%	99.5%	Nephronophthisis-like nephropathy 1, 613159
XPO5	100%	100%	100%	99.8%	
XPR1	100%	100%	100%	99.7%	Basal ganglia calcification, idiopathic, 6, 616413
XRCC1	100%	100%	100%	98.7%	?Spinocerebellar ataxia, autosomal recessive 26, 617633
XRCC2	100%	100%	100%	99.5%	Spermatogenic failure 50, 619145;?Premature ovarian failure 17, 619146;?Fanconi anemia, complementation group U, 617247
XRCC4	100%	100%	100%	99.8%	Short stature, microcephaly, and endocrine dysfunction, 616541
XYLT1	99.9%	98.8%	100%	98%	Desbuquois dysplasia 2, 615777;{Pseudoxanthoma elasticum, modifier of severity of}, 264800
XYLT2	99.7%	97.3%	100%	99.1%	{Pseudoxanthoma elasticum, modifier of severity of}, 264800;Spondyloocular syndrome, 605822

YAP1	100%	100%	100%	99.8%	Coloboma, ocular, with or without hearing impairment, cleft lip/palate, and/or impaired intellectual development, 120433
YARS1	100%	100%	100%	99.5%	Infantile-onset multisystem neurologic, endocrine, and pancreatic disease 2, 619418;Charcot-Marie-Tooth disease, dominant intermediate C, 608323
YARS2	100%	100%	100%	99.6%	Myopathy, lactic acidosis, and sideroblastic anemia 2, 613561
YEATS2	100%	100%	100%	99.6%	?Epilepsy, myoclonic, familial adult, 4, 615127
YIF1B	90%	90%	100%	99.1%	Kaya-Barakat-Masson syndrome, 619125
YIPF5	100%	100%	100%	99.9%	Microcephaly, epilepsy, and diabetes syndrome 2, 619278
YME1L1	100%	100%	100%	99.8%	?Optic atrophy 11, 617302
YPEL2	100%	100%	100%	99.4%	
YRDC	100%	100%	100%	99.1%	Galloway-Mowat syndrome 10, 619609
YWHAE	100%	100%	100%	99.7%	
YWHAG	100%	100%	100%	99.2%	Developmental and epileptic encephalopathy 56, 617665
YWHAZ	100%	100%	100%	99.4%	

YY1	100%	100%	99.9%	94.2%	Gabriele-de Vries syndrome, 617557
YY1AP1	100%	100%	100%	99.5%	Grange syndrome, 602531
ZAP70	100%	100%	100%	99.3%	Immunodeficiency 48, 269840;Autoimmune disease, multisystem, infantile-onset, 2, 617006
ZBTB11	100%	100%	100%	99.6%	Intellectual developmental disorder, autosomal recessive 69, 618383
ZBTB16	96%	96%	100%	99%	Leukemia, acute promyelocytic, PL2F/RARA type
ZBTB17	100%	100%	100%	99.4%	
ZBTB18	93.2%	93.1%	100%	99%	Intellectual developmental disorder, autosomal dominant 22, 612337
ZBTB20	100%	100%	100%	99.3%	Primrose syndrome, 259050
ZBTB24	100%	100%	100%	99.1%	Immunodeficiency-centromeric instability-facial anomalies syndrome 2, 614069
ZBTB42	100%	100%	100%	99.4%	?Lethal congenital contracture syndrome 6, 616248
ZBTB47	99.5%	99.4%	100%	97.7%	

ZBTB7A	100%	99.8%	100%	92.5%	Macrocephaly, neurodevelopmental delay, lymphoid hyperplasia, and persistent fetal hemoglobin, 619769
ZC3H14	100%	100%	100%	99.7%	Intellectual developmental disorder, autosomal recessive 56, 617125
ZC4H2	100%	99.8%	99.1%	71.6%	Wieacker-Wolff syndrome, 314580;Wieacker-Wolff syndrome, female-restricted, 301041
ZCCHC8	100%	100%	100%	99.5%	?Pulmonary fibrosis and/or bone marrow failure syndrome, telomere-related, 5, 618674
ZDHHC9	100%	99.3%	99.3%	71.1%	Intellectual developmental disorder, X-linked syndromic, Raymond type, 300799
ZEB1	100%	100%	100%	99.4%	Corneal dystrophy, posterior polymorphous, 3, 609141;Corneal dystrophy, Fuchs endothelial, 6, 613270
ZEB2	100%	100%	100%	99.1%	Mowat-Wilson syndrome, 235730
ZFHX2	100%	100%	100%	98.9%	?Marsili syndrome, 147430

ZFHX3	100%	100%	100%	98.6%	Prostate cancer, somatic, 176807;{Atrial fibrillation 8, susceptibility to}, 613055;Spinocerebellar ataxia 4, 600223
ZFHX4	100%	100%	100%	99.2%	?Ptosis, congenital, 178300
ZFP36L2	100%	100%	100%	98.2%	Oocyte/zygote/embryo maturation arrest 13, 620154
ZFP57	100%	100%	100%	99.3%	Diabetes mellitus, transient neonatal 1, 601410
ZFPM2	100%	100%	100%	99.5%	Diaphragmatic hernia 3, 610187;46XY sex reversal 9, 616067;Tetralogy of Fallot, 187500
ZFTRAF1	93.8%	89.8%	99.9%	91.4%	
ZFX	100%	99.9%	99.3%	74.1%	Intellectual developmental disorder, X-linked syndromic 37, 301118
ZFYVE19	100%	100%	100%	99.6%	Cholestasis, progressive familial intrahepatic, 9, 619849
ZFYVE26	100%	100%	100%	99.5%	Spastic paraplegia 15, autosomal recessive, 270700
ZFYVE27	100%	100%	100%	99.1%	

ZIC1	100%	100%	100%	97.1%	?Craniosynostosis 6, 616602;Structural brain anomalies with impaired intellectual development and craniosynostosis, 618736
ZIC2	100%	99.2%	99.8%	92.6%	Holoprosencephaly 5, 609637
ZIC3	100%	100%	98.2%	68.6%	Congenital heart defects, nonsyndromic, 1, X-linked, 306955;Heterotaxy, visceral, 1, X-linked, 306955;VACTERL association, X-linked, 314390
ZMIZ1	100%	99.9%	100%	98.6%	Neurodevelopmental disorder with dysmorphic facies and distal skeletal anomalies, 618659
ZMPSTE24	100%	100%	100%	99.5%	Mandibuloacral dysplasia with type B lipodystrophy, 608612;Restrictive dermopathy 1, 275210
ZMYM2	100%	100%	100%	99.5%	Neurodevelopmental-craniofacial syndrome with variable renal and cardiac abnormalities, 619522
ZMYM3	100%	99.3%	98.6%	69.3%	Intellectual developmental disorder, X-linked 112, 301111
ZMYND10	100%	100%	100%	98.7%	Ciliary dyskinesia, primary, 22, 615444

ZMYND11	100%	100%	100%	99.8%	Intellectual developmental disorder, autosomal dominant 30, 616083
ZMYND15	100%	100%	100%	98.7%	?Spermatogenic failure 14, 615842
ZMYND8	100%	100%	100%	99.5%	
ZNF141	100%	100%	100%	99.7%	?Polydactyly, postaxial, type A6, 615226
ZNF142	100%	100%	100%	99.3%	Neurodevelopmental disorder with impaired speech and hyperkinetic movements, 618425
ZNF148	100%	100%	100%	99.7%	Global developmental delay, absent or hypoplastic corpus callosum, and dysmorphic facies, 617260
ZNF292	99.4%	99.4%	100%	99.7%	Intellectual developmental disorder, autosomal dominant 64, 619188
ZNF335	100%	100%	100%	99.2%	Microcephaly 10, primary, autosomal recessive, 615095
ZNF341	100%	100%	100%	98.5%	Hyper-IgE syndrome 3, autosomal recessive, with recurrent infections, 618282
ZNF407	100%	100%	100%	99.5%	SIMHA syndrome, 619557
ZNF408	100%	100%	100%	99.4%	Retinitis pigmentosa 72, 616469;?Exudative vitreoretinopathy 6, 616468
ZNF41	100%	100%	99.3%	76.4%	

ZNF423	100%	100%	100%	99.1%	Nephronophthisis 14, 614844;Joubert syndrome 19, 614844
ZNF462	100%	100%	100%	99.5%	Weiss-Kruszka syndrome, 618619
ZNF469	100%	100%	100%	98.8%	Brittle cornea syndrome 1, 229200
ZNF513	100%	100%	100%	99.2%	?Retinitis pigmentosa 58, 613617
ZNF526	100%	100%	100%	99.2%	Dentici-Novelli neurodevelopmental syndrome, 619877
ZNF541	100%	100%	100%	99.2%	
ZNF592	100%	100%	100%	99.2%	
ZNF644	100%	100%	100%	99.9%	Myopia 21, autosomal dominant, 614167
ZNF668	100%	100%	100%	98.8%	Neurodevelopmental disorder with poor growth, large ears, and dysmorphic facies, 620194
ZNF687	100%	100%	100%	99%	Paget disease of bone 6, 616833
ZNF699	100%	100%	100%	99.4%	DEGCAGS syndrome, 619488
ZNF711	100%	99.9%	99.2%	74.8%	Intellectual developmental disorder, X-linked 97, 300803
ZNF750	100%	100%	100%	99.5%	?Seborrhea-like dermatitis with psoriasiform elements, 610227

ZNF808	100%	100%	100%	99.9%	Pancreatic agenesis 3, 620991
ZNF862	100%	100%	100%	99.3%	?Fibromatosis, gingival 6, 620999
ZNFX1	100%	100%	100%	99.5%	Immunodeficiency 91 and hyperinflammation, 619644
ZNG1A	98%	97.3%	98%	94.4%	
ZNHIT3	76.2%	76.2%	100%	99.2%	PEHO syndrome, 260565
ZNRF3	97.2%	95.6%	100%	97.9%	
ZP1	100%	100%	100%	99.6%	Oocyte/zygote/embryo maturation arrest 1, 615774
ZP2	100%	100%	100%	99.5%	Oocyte/zygote/embryo maturation arrest 6, 618353
ZP3	100%	100%	100%	98.5%	Oocyte/zygote/embryo maturation arrest 3, 617712
ZPBP	100%	100%	100%	99.9%	?Spermatogenic failure 66, 619799
ZPR1	100%	100%	100%	99.4%	?Growth restriction, hypoplastic kidneys, alopecia, and distinctive facies, 619321
ZRSR2	84.1%	84%	98.5%	71.7%	Orofaciodigital syndrome XXI, 301132
ZSCAN10	100%	100%	100%	98.5%	Otofacial neurodevelopmental syndrome, 620910

ZSWIM6	96.6%	93.7%	97.4%	93.2%	Neurodevelopmental disorder with movement abnormalities, abnormal gait, and autistic features, 617865;Acromelic frontonasal dysostosis, 603671
ZSWIM7	88.9%	88.9%	100%	99.9%	Spermatogenic failure 71, 619831;?Ovarian dysgenesis 10, 619834

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.
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.
srWGS GRCh38 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 GRCh38 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 : March 17th, 2023.
This list is accurate for panel version DG 4.0.0

Ad 1. "No OMIM phenotype" signifies a gene without a current OMIM association Ad 2. OMIM phenotype descriptions between {} signify risk factors