PRECONCEPTION SCREENING GENE PANEL DG 3.1.0 (2351 genes)

Releasedate: 23-03-2021

Gene	Agilent V5 covered >10x	Agilent V5 covered >20x	TWIST covered >10x	TWIST covered >20x	Associated Phenotype description and OMIM disease ID
AARS1	100	99,9	100	100	Developmental and epileptic encephalopathy 29, 616339 Charcot-Marie-Tooth disease, axonal, type 2N, 613287
AARS2	100	99,4	100	100	Combined oxidative phosphorylation deficiency 8, 614096 Leukoencephalopathy, progressive, with ovarian failure, 615889
AASS	100	99,7	100	100	Hyperlysinemia, 238700
ABAT	100	99,4	100	100	GABA-transaminase deficiency, 613163
ABCA1	99,9	99,1	100	100	HDL deficiency, familial, 1, 604091 Tangier disease, 205400
ABCA3	99,9	99,3	100	100	Surfactant metabolism dysfunction, pulmonary, 3, 610921
ABCA4	99,9	99,3	96,5	96,5	Retinal dystrophy, early-onset severe, 248200 Stargardt disease 1, 248200 Fundus flavimaculatus, 248200 {Macular degeneration, age-related, 2}, 153800 Cone-rod dystrophy 3, 604116 Retinitis pigmentosa 19, 601718
ABCB11	100	99,7	100	100	Cholestasis, progressive familial intrahepatic 2, 601847 Cholestasis, benign recurrent intrahepatic, 2, 605479
ABCB4	99,9	99,6	100	100	Gallbladder disease 1, 600803 Cholestasis, intrahepatic, of pregnancy, 3, 614972 Cholestasis, progressive familial intrahepatic 3, 602347
ABCC2	100	99,9	100	100	Dubin-Johnson syndrome, 237500
ABCC6	93,6	92,4	100	100	Pseudoxanthoma elasticum, 264800 Pseudoxanthoma elasticum, forme fruste, 177850 Arterial calcification, generalized, of infancy, 2, 614473
ABCC8	100	99,8	100	100	Diabetes mellitus, noninsulin-dependent, 125853 Diabetes mellitus, permanent neonatal 3, with or without neurologic features, 618857 Diabetes mellitus, transient neonatal 2, 610374 Hyperinsulinemic hypoglycemia, familial, 1, 256450 Hypoglycemia of infancy, leucine-sensitive, 240800
ABCD3	99,8	97,7	100	100	?Bile acid synthesis defect, congenital, 5, 616278

ABCD4	99,9	98,6	100	100	Methylmalonic aciduria and homocystinuria, cblJ type, 614857
ABCG5	100	100	100	100	Sitosterolemia 2, 618666
ABCG8	99,1	97,3	100	100	{Gallbladder disease 4}, 611465 Sitosterolemia 1, 210250
ABHD12	91,2	85,2	100	99,4	Polyneuropathy, hearing loss, ataxia, retinitis pigmentosa, and cataract, 612674
ABHD5	100	100	100	100	Chanarin-Dorfman syndrome, 275630
ACACA	98,4	98,1	100	100	Acetyl-CoA carboxylase deficiency, 613933
ACAD8	100	100	100	100	Isobutyryl-CoA dehydrogenase deficiency, 611283
ACAD9	100	99,9	100	100	Mitochondrial complex I deficiency, nuclear type 20, 611126
ACADM	99,8	99	100	100	Acyl-CoA dehydrogenase, medium chain, deficiency of, 201450
ACADS	99,9	98,2	100	100	Acyl-CoA dehydrogenase, short-chain, deficiency of, 201470
ACADSB	100	99,2	100	100	2-methylbutyrylglycinuria, 610006
ACADVL	99,4	97,3	100	100	VLCAD deficiency, 201475
ACAN	96,6	92,9	98,9	98,7	Short stature and advanced bone age, with or without early-onset osteoarthritis and/or osteochondritis dissecans, 165800 Spondyloepimetaphyseal dysplasia, aggrecan type, 612813 ?Spondyloepiphyseal dysplasia, Kimberley type, 608361
ACAT1	99,9	97,6	100	100	Alpha-methylacetoacetic aciduria, 203750
ACD	100	99,9	100	100	?Dyskeratosis congenita, autosomal dominant 6, 616553 ?Dyskeratosis congenita, autosomal recessive 7, 616553
ACE	99,9	98,4	100	100	Renal tubular dysgenesis, 267430 {Microvascular complications of diabetes 3}, 612624 {Stroke, hemorrhagic}, 614519 {Myocardial infarction, susceptibility to}, 0 [Angiotensin I-converting enzyme, benign serum increase], 0 {SARS, progression of}, 0
ACER3	99,8	98,6	100	100	?Leukodystrophy, progressive, early childhood-onset, 617762
ACO2	96,3	90,3	100	100	?Optic atrophy 9, 616289 Infantile cerebellar-retinal degeneration, 614559
ACOX1	100	99,9	100	100	Mitchell syndrome, 618960 Peroxisomal acyl-CoA oxidase deficiency, 264470
ACOX2	100	99,2	100	100	Bile acid synthesis defect, congenital, 6, 617308
ACP5	99,8	98,3	100	100	Spondyloenchondrodysplasia with immune dysregulation, 607944
ACSF3	100	99,9	100	100	Combined malonic and methylmalonic aciduria, 614265
ACTA1	99,6	92,3	100	100	Myopathy, actin, congenital, with cores, 161800 Nemaline myopathy 3, autosomal dominant or recessive, 161800 Myopathy, congenital, with fiber-type disproportion 1, 255310

					Myopathy, actin, congenital, with excess of thin myofilaments, 161800 ?Myopathy, scapulohumeroperoneal, 616852
ACTL6B	100	99,8	100	100	Developmental and epileptic encephalopathy 76, 618468 Intellectual developmental disorder with severe speech and ambulation defects, 618470
ACY1	100	98,8	100	100	Aminoacylase 1 deficiency, 609924
ADA	100	99,7	100	100	Adenosine deaminase deficiency, partial, 102700 Severe combined immunodeficiency due to ADA deficiency, 102700
ADA2	100	99	100	100	?Sneddon syndrome, 182410 Vasculitis, autoinflammation, immunodeficiency, and hematologic defects syndrome, 615688
ADAM17	99,9	99	100	100	?Inflammatory skin and bowel disease, neonatal, 1, 614328
ADAM22	99,9	99,5	100	100	Developmental and epileptic encephalopathy 61, 617933
ADAM9	99,8	99,1	100	100	Cone-rod dystrophy 9, 612775
ADAMTS10	99,9	98,5	100	100	Weill-Marchesani syndrome 1, recessive, 277600
ADAMTS13	97,1	93,8	99,9	99,5	Thrombotic thrombocytopenic purpura, hereditary, 274150
ADAMTS17	92,8	89	97,6	95,8	Weill-Marchesani 4 syndrome, recessive, 613195
ADAMTS18	100	99,7	100	100	Microcornea, myopic chorioretinal atrophy, and telecanthus, 615458
ADAMTS2	99	96,7	98,1	97,9	Ehlers-Danlos syndrome, dermatosparaxis type, 225410
ADAMTS3	100	100	100	100	Hennekam lymphangiectasia-lymphedema syndrome 3, 618154
ADAMTSL2	97,1	93,3	99,8	99,4	Geleophysic dysplasia 1, 231050
ADAMTSL4	100	99,2	100	100	Ectopia lentis et pupillae, 225200 Ectopia lentis, isolated, autosomal recessive, 225100
ADAR	100	99,8	100	100	Aicardi-Goutieres syndrome 6, 615010 Dyschromatosis symmetrica hereditaria, 127400
ADARB1	97,9	95,2	95,1	95,1	Neurodevelopmental disorder with hypotonia, microcephaly, and seizures, 618862
ADAT3	100	99,7	100	100	Mental retardation, autosomal recessive 36, 615286
ADCY1	95,2	93,8	98,5	97,9	?Deafness, autosomal recessive 44, 610154
ADCY6	100	100	100	100	Lethal congenital contracture syndrome 8, 616287
ADD3	99,9	99,5	100	100	Cerebral palsy, spastic quadriplegic, 3, 617008
ADGRG1	100	100	100	100	Polymicrogyria, bilateral perisylvian, 615752 Polymicrogyria, bilateral frontoparietal, 606854
ADGRG6	99,9	99	100	100	Lethal congenital contracture syndrome 9, 616503
ADGRV1	99,6	98,6	100	100	Usher syndrome, type 2C, 605472 ?Febrile seizures, familial, 4, 604352 Usher syndrome, type 2C, GPR98/PDZD7 digenic, 605472
ADK	84,1	81	84,5	84,5	Hypermethioninemia due to adenosine kinase deficiency, 614300
ADPRS	100	99,8	100	100	Neurodegeneration, childhood-onset, stress-induced, with variable ataxia and seizures, 618170
ADSL	99,2	98,7	100	100	Adenylosuccinase deficiency, 103050

ADSS1	90,2	87,5	100	100	Myopathy, distal, 5, 617030
AEBP1	100	100	100	100	Ehlers-Danlos syndrome, classic-like, 2, 618000
AFG3L2	95	91,1	100	99,9	Spastic ataxia 5, autosomal recessive, 614487 Optic atrophy 12, 618977 Spinocerebellar ataxia 28, 610246
AGA	100	100	100	100	Aspartylglucosaminuria, 208400
AGBL5	99,9	99,3	100	100	Retinitis pigmentosa 75, 617023
AGK	90,6	88,6	91,2	91,2	Sengers syndrome, 212350 Cataract 38, autosomal recessive, 614691
AGL	100	99,4	100	100	Glycogen storage disease IIIb, 232400 Glycogen storage disease IIIa, 232400
AGPAT2	99,6	96,1	100	100	Lipodystrophy, congenital generalized, type 1, 608594
AGPS	99,3	95,4	100	99,9	Rhizomelic chondrodysplasia punctata, type 3, 600121
AGRN	96,9	92,6	100	99,9	Myasthenic syndrome, congenital, 8, with pre- and postsynaptic defects, 615120
AGT	100	100	100	100	Renal tubular dysgenesis, 267430 {Hypertension, essential, susceptibility to}, 145500 {Preeclampsia, susceptibility to}, 0
AGTPBP1	96	94,1	100	100	Neurodegeneration, childhood-onset, with cerebellar atrophy, 618276
AGTR1	92	91,8	100	100	Renal tubular dysgenesis, 267430 {Hypertension, essential}, 145500
AGXT	100	100	100	100	Hyperoxaluria, primary, type 1, 259900
AHCY	100	99,2	100	100	Hypermethioninemia with deficiency of S-adenosylhomocysteine hydrolase, 613752
AHI1	99,7	97,9	100	100	Joubert syndrome 3, 608629
AHR	99,2	98,8	100	100	?Retinitis pigmentosa 85, 618345
AHSG	99,9	99,5	100	100	?Alopecia-mental retardation syndrome 1, 203650
AICDA	100	100	100	100	Immunodeficiency with hyper-IgM, type 2, 605258
AIMP1	99,2	94,5	100	99,9	Leukodystrophy, hypomyelinating, 3, 260600
AIMP2	88,9	86	100	100	Leukodystrophy, hypomyelinating, 17, 618006
AIPL1	100	99,8	100	100	Leber congenital amaurosis 4, 604393 Retinitis pigmentosa, juvenile, 604393 Cone-rod dystrophy, 604393
AIRE	100	99,8	100	100	Autoimmune polyendocrinopathy syndrome , type I, with or without reversible metaphyseal dysplasia, 240300
AK1	100	100	100	100	Hemolytic anemia due to adenylate kinase deficiency, 612631
AK2	98,9	94,9	100	99,9	Reticular dysgenesis, 267500
AKR1D1	100	99,4	100	100	Bile acid synthesis defect, congenital, 2, 235555

ALAD	99,3	94,1	100	100	Porphyria, acute hepatic, 612740 {Lead poisoning, susceptibility to}, 612740
ALB	100	99,4	100	100	[Dysalbuminemic hyperthyroxinemia], 615999 Analbuminemia, 616000
ALDH18A1	100	99,9	100	100	Cutis laxa, autosomal recessive, type IIIA, 219150 Cutis laxa, autosomal dominant 3, 616603 Spastic paraplegia 9B, autosomal recessive, 616586 Spastic paraplegia 9A, autosomal dominant, 601162
ALDH1A3	97,2	94,5	100	99,9	Microphthalmia, isolated 8, 615113
ALDH3A2	88,8	88,1	93,2	93,2	Sjogren-Larsson syndrome, 270200
ALDH4A1	100	99,4	100	100	Hyperprolinemia, type II, 239510
ALDH5A1	91	81,5	100	100	Succinic semialdehyde dehydrogenase deficiency, 271980
ALDH6A1	100	99,9	100	100	Methylmalonate semialdehyde dehydrogenase deficiency, 614105
ALDH7A1	94,4	88,8	100	100	Epilepsy, pyridoxine-dependent, 266100
ALDOA	98,9	96,9	100	100	Glycogen storage disease XII, 611881
ALDOB	99,4	96,6	100	100	Fructose intolerance, hereditary, 229600
ALG1	53	45,8	100	100	Congenital disorder of glycosylation, type Ik, 608540
ALG11	96,8	96,8	96,8	96,8	Congenital disorder of glycosylation, type Ip, 613661
ALG12	100	100	100	100	Congenital disorder of glycosylation, type Ig, 607143
ALG14	100	99,9	100	100	?Myasthenic syndrome, congenital, 15, without tubular aggregates, 616227 Myopathy, epilepsy, and progressive cerebral atrophy, 619036 Intellectual developmental disorder with epilepsy, behavioral abnormalities, and coarse facies, 619031
ALG2	100	100	100	100	Myasthenic syndrome, congenital, 14, with tubular aggregates, 616228 ?Congenital disorder of glycosylation, type Ii, 607906
ALG3	100	99,7	100	100	Congenital disorder of glycosylation, type Id, 601110
ALG6	98,6	94,8	100	100	Congenital disorder of glycosylation, type Ic, 603147
ALG8	97,2	95,6	96,6	96,6	Congenital disorder of glycosylation, type Ih, 608104 Polycystic liver disease 3 with or without kidney cysts, 617874
ALG9	100	99,7	100	100	Gillessen-Kaesbach-Nishimura syndrome, 263210 Congenital disorder of glycosylation, type Il, 608776
ALKBH8	99,8	98,9	100	100	Intellectual developmental disorder, autosomal recessive 71, 618504
ALMS1	99,8	99,5	100	100	Alstrom syndrome, 203800
ALOX12B	100	100	100	100	Ichthyosis, congenital, autosomal recessive 2, 242100
ALOXE3	100	99,5	100	100	Ichthyosis, congenital, autosomal recessive 3, 606545
ALPK3	97,8	94,6	100	100	Cardiomyopathy, familial hypertrophic 27, 618052

ALPL	100	100	100	100	Hypophosphatasia, adult, 146300 Odontohypophosphatasia, 146300 Hypophosphatasia, childhood, 241510 Hypophosphatasia, infantile, 241500
ALS2	100	99,9	100	100	Primary lateral sclerosis, juvenile, 606353 Amyotrophic lateral sclerosis 2, juvenile, 205100 Spastic paralysis, infantile onset ascending, 607225
ALX1	99,7	97,1	100	100	Frontonasal dysplasia 3, 613456
ALX3	77,9	73,3	100	100	Frontonasal dysplasia 1, 136760
ALX4	100	99,3	100	100	Frontonasal dysplasia 2, 613451 Parietal foramina 2, 609597 {Craniosynostosis 5, susceptibility to}, 615529
AMACR	100	100	100	100	Bile acid synthesis defect, congenital, 4, 214950 Alpha-methylacyl-CoA racemase deficiency, 614307
AMBN	99,8	98,5	100	100	Amelogenesis imperfecta, type IF, 616270
AMN	89,7	80	100	100	Imerslund-Grasbeck syndrome 2, 618882
AMPD1	99,9	98,6	100	100	Myopathy due to myoadenylate deaminase deficiency, 615511
AMPD2	99,8	98,9	100	100	?Spastic paraplegia 63, 615686 Pontocerebellar hypoplasia, type 9, 615809
AMT	100	100	100	100	Glycine encephalopathy, 605899
ANAPC1	59,4	57,7	100	99,9	Rothmund-Thomson syndrome, type 1, 618625
ANGPTL3	98,8	95,4	100	100	Hypobetalipoproteinemia, familial, 2, 605019
ANK1	100	99,4	100	100	Spherocytosis, type 1, 182900
ANK3	99,3	99	100	100	Mental retardation, autosomal recessive, 37, 615493
ANKH	100	100	100	100	Craniometaphyseal dysplasia, 123000 Chondrocalcinosis 2, 118600
ANKLE2	99,9	98,6	100	99,8	Microcephaly 16, primary, autosomal recessive, 616681
ANKS6	93,8	89,5	97,9	95,8	Nephronophthisis 16, 615382
ANO10	99,8	97,9	100	100	Spinocerebellar ataxia, autosomal recessive 10, 613728
ANO5	99,5	97,3	100	100	Miyoshi muscular dystrophy 3, 613319 Muscular dystrophy, limb-girdle, autosomal recessive 12, 611307 Gnathodiaphyseal dysplasia, 166260
ANO6	99,9	98,7	100	100	Scott syndrome, 262890
ANTXR1	99,7	97,9	100	100	GAPO syndrome, 230740 {?Hemangioma, capillary infantile, susceptibility to}, 602089
ANTXR2	100	98,2	100	100	Hyaline fibromatosis syndrome, 228600
AP1B1	100	99,5	100	100	Keratitis-ichthyosis-deafness syndrome, autosomal recessive, 242150

AP1S1	99,9	99,5	100	100	MEDNIK syndrome, 609313
AP3B1	99,2	95,8	100	100	Hermansky-Pudlak syndrome 2, 608233
AP3B2	93,3	89,5	99,8	98,6	Developmental and epileptic encephalopathy 48, 617276
AP3D1	99,8	98,6	100	100	?Hermansky-Pudlak syndrome 10, 617050
AP4B1	99,9	98,7	100	100	Spastic paraplegia 47, autosomal recessive, 614066
AP4E1	99,8	98,7	100	100	Stuttering, familial persistent, 1, 184450 Spastic paraplegia 51, autosomal recessive, 613744
AP4M1	99,9	98,9	100	100	Spastic paraplegia 50, autosomal recessive, 612936
AP4S1	78,9	71,3	87,9	87,9	Spastic paraplegia 52, autosomal recessive, 614067
AP5Z1	100	99,8	100	100	Spastic paraplegia 48, autosomal recessive, 613647
APC2	97,6	92,7	99,9	99,1	?Sotos syndrome 3, 617169 Cortical dysplasia, complex, with other brain malformations 10, 618677
APOB	99,8	99,3	100	100	Hypobetalipoproteinemia, 615558 Hypercholesterolemia, familial, 2, 144010
APOC2	100	100	100	100	Hyperlipoproteinemia, type Ib, 207750
APOE	98,9	90,7	100	100	Hyperlipoproteinemia, type III, 617347 {Coronary artery disease, severe, susceptibility to}, 617347 {?Alzheimer disease, protection against, due to APOE3-Christchurch}, 607822 Lipoprotein glomerulopathy, 611771 Sea-blue histiocyte disease, 269600 {?Macular degeneration, age-related}, 603075 Alzheimer disease 2, 104310
APRT	100	99,5	100	100	Adenine phosphoribosyltransferase deficiency, 614723
APTX	94,9	92,4	100	100	Ataxia, early-onset, with oculomotor apraxia and hypoalbuminemia, 208920
AQP2	100	98,6	100	100	Diabetes insipidus, nephrogenic, 125800
ARFGEF2	99,9	99,1	100	100	Periventricular heterotopia with microcephaly, 608097
ARG1	92,9	92,9	92,9	92,9	Argininemia, 207800
ARHGDIA	100	100	100	100	Nephrotic syndrome, type 8, 615244
ARHGEF18	95,4	92,3	100	100	Retinitis pigmentosa 78, 617433
ARHGEF2	93	93	100	100	?Neurodevelopmental disorder with midbrain and hindbrain malformations, 617523
ARL13B	100	99,2	100	100	Joubert syndrome 8, 612291
ARL2BP	95,9	88,3	100	100	Retinitis pigmentosa with or without situs inversus, 615434
ARL3	100	98,4	100	100	Joubert syndrome 35, 618161 Retinitis pigmentosa 83, 618173
ARL6	99,9	98,6	100	100	Retinitis pigmentosa 55, 613575 Bardet-Biedl syndrome 3, 600151 {Bardet-Biedl syndrome 1, modifier of}, 209900

ARL6IP1	99,4	92,6	100	100	?Spastic paraplegia 61, autosomal recessive, 615685
ARMC4	92,1	90	96,3	96,3	Ciliary dyskinesia, primary, 23, 615451
ARMC9	100	99,8	100	100	Joubert syndrome 30, 617622
ARNT2	100	100	100	99,6	?Webb-Dattani syndrome, 615926
ARPC1B	100	100	100	100	Immunodeficiency 71 with inflammatory disease and congenital thrombocytopenia, 617718
ARSA	100	99,8	100	100	Metachromatic leukodystrophy, 250100
ARSB	97	88,7	100	100	Mucopolysaccharidosis type VI (Maroteaux-Lamy), 253200
ARSG	100	99,5	100	100	Usher syndrome, type IV, 618144
ARV1	100	99,9	100	100	Developmental and epileptic encephalopathy 38, 617020
ASAH1	99,7	98,6	100	100	Farber lipogranulomatosis, 228000 Spinal muscular atrophy with progressive myoclonic epilepsy, 159950
ASCC1	93,4	90,3	87,1	87,1	Barrett esophagus/esophageal adenocarcinoma, 614266 Spinal muscular atrophy with congenital bone fractures 2, 616867
ASL	100	99,6	100	100	Argininosuccinic aciduria, 207900
ASNS	99,4	95,2	100	100	Asparagine synthetase deficiency, 615574
ASPA	99,9	98,3	100	100	Canavan disease, 271900
ASPH	99,9	98,8	100	100	Traboulsi syndrome, 601552
ASPM	99,7	98,2	100	100	Microcephaly 5, primary, autosomal recessive, 608716
ASS1	95,4	87,9	100	100	Citrullinemia, 215700
ATAD1	99,6	95,1	100	100	Hyperekplexia 4, 618011
ATAD3A	91,9	83,2	100	100	Harel-Yoon syndrome, 617183 Pontocerebellar hypoplasia, hypotonia, and respiratory insufficiency syndrome, neonatal lethal, 618810
ATCAY	100	99,8	100	100	Ataxia, cerebellar, Cayman type, 601238
ATF6	100	99,9	100	100	Achromatopsia 7, 616517
ATG5	99,4	97,8	100	100	?Spinocerebellar ataxia, autosomal recessive 25, 617584
ATIC	99,9	99,3	100	100	AICA-ribosiduria due to ATIC deficiency, 608688
ATM	99,8	98,1	100	100	Ataxia-telangiectasia, 208900 {Breast cancer, susceptibility to}, 114480 Lymphoma, mantle cell, somatic, 0 Lymphoma, B-cell non-Hodgkin, somatic, 0 T-cell prolymphocytic leukemia, somatic, 0
ATOH7	96	91,2	99,1	94,4	Persistent hyperplastic primary vitreous, autosomal recessive, 221900
ATP13A2	100	99,5	100	100	Kufor-Rakeb syndrome, 606693 Spastic paraplegia 78, autosomal recessive, 617225
ATP2A1	100	100	100	100	Brody myopathy, 601003

ATP5F1A	95,2	87,6	100	100	?Mitochondrial complex V (ATP synthase) deficiency, nuclear type 4, 615228 ?Combined oxidative phosphorylation deficiency 22, 616045
ATP5F1D	96,2	89,3	100	100	Mitochondrial complex V (ATP synthase) deficiency, 618120
ATP5F1E	100	100	100	100	?Mitochondrial complex V (ATP synthase) deficiency, nuclear type 3, 614053
ATP6V0A2	100	99,5	100	100	Wrinkly skin syndrome, 278250 Cutis laxa, autosomal recessive, type IIA, 219200
ATP6V0A4	100	99,9	100	100	Distal renal tubular acidosis 3, with or without sensorineural hearing loss, 602722
ATP6V1A	99,9	98,7	100	100	Cutis laxa, autosomal recessive, type IID, 617403 Developmental and epileptic encephalopathy 93, 618012
ATP6V1B1	100	100	100	100	Distal renal tubular acidosis 2 with progressive sensorineural hearing loss, 267300
ATP6V1E1	93,1	88,3	100	100	Cutis laxa, autosomal recessive, type IIC, 617402
ATP7B	99,9	99,2	100	100	Wilson disease, 277900
ATP8A2	100	99,7	100	100	?Cerebellar ataxia, mental retardation, and dysequilibrium syndrome 4, 615268
ATP8B1	96,5	94	100	100	Cholestasis, intrahepatic, of pregnancy, 1, 147480 Cholestasis, progressive familial intrahepatic 1, 211600 Cholestasis, benign recurrent intrahepatic, 243300
ATPAF2	100	100	100	100	?Mitochondrial complex V (ATP synthase) deficiency, nuclear type 1, 604273
ATR	99,9	99,4	100	100	Seckel syndrome 1, 210600 ?Cutaneous telangiectasia and cancer syndrome, familial, 614564
AUH	100	99,8	100	100	3-methylglutaconic aciduria, type I, 250950
AURKC	100	99,2	100	100	Spermatogenic failure 5, 243060
B2M	100	100	100	100	Immunodeficiency 43, 241600 ?Amyloidosis, familial visceral, 105200
B3GALNT2	93,8	89,4	92,5	92,5	Muscular dystrophy-dystroglycanopathy (congenital with brain and eye anomalies, type A, 11, 615181
B3GALT6	75,7	69,7	89,8	81,6	Al-Gazali syndrome, 609465 Ehlers-Danlos syndrome, spondylodysplastic type, 2, 615349 Spondyloepimetaphyseal dysplasia with joint laxity, type 1, with or without fractures, 271640
B3GAT3	99,9	98,2	94,8	94,8	Multiple joint dislocations, short stature, craniofacial dysmorphism, with or without congenital heart defects, 245600
B3GLCT	99,6	96,3	99,9	99,2	Peters-plus syndrome, 261540
B4GALNT1	99,3	95	100	100	Spastic paraplegia 26, autosomal recessive, 609195
B4GALT1	100	99,8	100	100	Congenital disorder of glycosylation, type IId, 607091
B4GALT7	99,8	97,4	99,9	98,6	Ehlers-Danlos syndrome, spondylodysplastic type, 1, 130070
B4GAT1	100	100	100	100	Muscular dystrophy-dystroglycanopathy (congenital with brain and eye anomalies), type A, 13, 615287
B9D1	85,2	85,1	94,2	93,9	?Meckel syndrome 9, 614209 Joubert syndrome 27, 617120

B9D2	100	100	100	100	Joubert syndrome 34, 614175 ?Meckel syndrome 10, 614175
BAAT	99,8	98,4	100	100	Hypercholanemia, familial, 607748
BANF1	98,3	86,6	100	100	Nestor-Guillermo progeria syndrome, 614008
BBIP1	98,6	92,4	100	100	?Bardet-Biedl syndrome 18, 615995
BBS1	100	100	100	100	Bardet-Biedl syndrome 1, 209900
BBS10	100	99,8	100	100	Bardet-Biedl syndrome 10, 615987
BBS12	100	100	100	100	Bardet-Biedl syndrome 12, 615989
BBS2	100	99,5	100	100	Bardet-Biedl syndrome 2, 615981 Retinitis pigmentosa 74, 616562
BBS4	99,9	99,3	100	100	Bardet-Biedl syndrome 4, 615982
BBS5	99	93,9	100	100	Bardet-Biedl syndrome 5, 615983
BBS7	98,7	95,5	100	100	Bardet-Biedl syndrome 7, 615984
BBS9	92,3	90,4	95,8	95,8	Bardet-Biedl syndrome 9, 615986
BCKDHA	99,9	99,2	100	100	Maple syrup urine disease, type Ia, 248600
BCKDHB	99,5	94,4	100	100	Maple syrup urine disease, type Ib, 248600
BCKDK	100	100	100	100	Branched-chain ketoacid dehydrogenase kinase deficiency, 614923
BCL10	100	100	100	100	?Immunodeficiency 37, 616098 {Male germ cell tumor, somatic}, 273300 Lymphoma, MALT, somatic, 137245 {Lymphoma, follicular, somatic}, 605027 {Mesothelioma, somatic}, 156240 {Sezary syndrome, somatic}, 0
BCS1L	100	100	100	100	GRACILE syndrome, 603358 Bjornstad syndrome, 262000 Mitochondrial complex III deficiency, nuclear type 1, 124000
BFSP1	99	89,9	100	99,9	Cataract 33, multiple types, 611391
BFSP2	99,8	97,6	100	100	Cataract 12, multiple types, 611597
BHLHA9	70,9	50,4	99,8	97,3	Syndactyly, mesoaxial synostotic, with phalangeal reduction, 609432 ?Camptosynpolydactyly, complex, 607539
BIN1	99,6	95,7	100	100	Centronuclear myopathy 2, 255200
BLM	99,8	98,3	100	100	Bloom syndrome, 210900
BLNK	97,1	95,5	100	100	?Agammaglobulinemia 4, 613502
BLOC1S3	98,5	81,3	100	100	Hermansky-Pudlak syndrome 8, 614077
BLOC1S6	99,9	97,8	100	100	?Hermansky-pudlak syndrome 9, 614171
BLVRA	100	99,4	100	100	Hyperbiliverdinemia, 614156
BMP1	100	100	100	100	Osteogenesis imperfecta, type XIII, 614856

BMPER	100	99,8	100	100	Diaphanospondylodysostosis, 608022
BMPR1B	100	99,9	100	100	Brachydactyly, type A2, 112600 Brachydactyly, type A1, D, 616849 Acromesomelic dysplasia, Demirhan type, 609441
BOLA3	99,4	90,2	100	100	Multiple mitochondrial dysfunctions syndrome 2 with hyperglycinemia, 614299
BPGM	100	100	100	100	Erythrocytosis, familial, 8, 222800
BRAT1	99,7	98,2	100	100	Rigidity and multifocal seizure syndrome, lethal neonatal, 614498 Neurodevelopmental disorder with cerebellar atrophy and with or without seizures, 618056
BRCA1	99,4	98,8	100	100	Fanconi anemia, complementation group S, 617883 {Pancreatic cancer, susceptibility to, 4}, 614320 {Breast-ovarian cancer, familial, 1}, 604370
BRCA2	99,8	98,5	100	100	{Pancreatic cancer 2}, 613347 {Breast cancer, male, susceptibility to}, 114480 {Glioblastoma 3}, 613029 Wilms tumor, 194070 Fanconi anemia, complementation group D1, 605724 {Medulloblastoma}, 155255 {Prostate cancer}, 176807 {Breast-ovarian cancer, familial, 2}, 612555
BRF1	99,9	98,4	100	100	Cerebellofaciodental syndrome, 616202
BRIP1	99,9	99	100	100	Fanconi anemia, complementation group J, 609054 {Breast cancer, early-onset, susceptibility to}, 114480
BSCL2	100	100	100	100	Neuropathy, distal hereditary motor, type VC, 619112 Lipodystrophy, congenital generalized, type 2, 269700 Silver spastic paraplegia syndrome, 270685 Encephalopathy, progressive, with or without lipodystrophy, 615924
BSND	100	100	100	100	Sensorineural deafness with mild renal dysfunction, 602522 Bartter syndrome, type 4a, 602522
BTD	83,1	83	83,1	83,1	Biotinidase deficiency, 253260
BUB1B	99,6	98,9	100	100	Colorectal cancer, somatic, 114500 [Premature chromatid separation trait], 176430 Mosaic variegated aneuploidy syndrome 1, 257300
BVES	99,9	98,8	100	100	Muscular dystrophy, limb-girdle, autosomal recessive 25, 616812
C12orf4	100	99,3	100	100	Mental retardation, autosomal recessive 66, 618221
C12orf57	100	98,9	100	100	Temtamy syndrome, 218340
C12orf65	99,8	98,5	100	100	Spastic paraplegia 55, autosomal recessive, 615035 Combined oxidative phosphorylation deficiency 7, 613559
C15orf41	85,9	85,6	100	100	Dyserythropoietic anemia, congenital, type Ib, 615631

C19orf12	100	99,8	100	100	Neurodegeneration with brain iron accumulation 4, 614298 ?Spastic paraplegia 43, autosomal recessive, 615043
C1QA	100	100	100	100	C1q deficiency, 613652
C1QB	100	100	100	100	C1q deficiency, 613652
C1QBP	86,9	77,3	100	100	Combined oxidative phosphorylation deficiency 33, 617713
C1QC	100	99,2	100	100	C1q deficiency, 613652
C1S	99,9	99	99,5	97,7	Ehlers-Danlos syndrome, periodontal type, 2, 617174 C1s deficiency, 613783
C2	100	100	100	100	C2 deficiency, 217000 {Macular degeneration, age-related, 14, reduced risk of}, 615489
C2CD3	95,8	95,6	95,9	95,9	Orofaciodigital syndrome XIV, 615948
C3	99,9	99,2	100	100	{Hemolytic uremic syndrome, atypical, susceptibility to, 5}, 612925 C3 deficiency, 613779 {Macular degeneration, age-related, 9}, 611378
C4A	98,4	95,1	99,6	99,2	C4a deficiency, 614380 [Blood group, Rodgers], 614374
C4B	99,2	96,9	99,9	99,8	C4B deficiency, 614379
C5	99,9	98,5	100	100	[Eculizumab, poor response to], 615749 C5 deficiency, 609536
C8A	100	99,6	100	100	C8 deficiency, type I, 613790
C8B	100	99,2	100	100	C8 deficiency, type II, 613789
C8orf37	100	99,4	100	100	Retinitis pigmentosa 64, 614500 Bardet-Biedl syndrome 21, 617406 Cone-rod dystrophy 16, 614500
C9	99,9	99,5	100	100	{Macular degeneration, age-related, 15, susceptibility to}, 615591 C9 deficiency, 613825
CA12	100	100	100	100	Hyperchlorhidrosis, isolated, 143860
CA2	100	100	100	100	Osteopetrosis, autosomal recessive 3, with renal tubular acidosis, 259730
CA5A	87,4	85,2	87,7	87,7	Hyperammonemia due to carbonic anhydrase VA deficiency, 615751
CA8	99,6	97,3	100	100	Cerebellar ataxia and mental retardation with or without quadrupedal locomotion 3, 613227
CABP2	75,9	68	100	100	Deafness, autosomal recessive 93, 614899
CABP4	100	99,9	100	100	Cone-rod synaptic disorder, congenital nonprogressive, 610427
CACNA1B	97,5	95,7	99,1	97,7	Neurodevelopmental disorder with seizures and nonepileptic hyperkinetic movements, 618497
CACNA1D	98	97,9	100	100	Primary aldosteronism, seizures, and neurologic abnormalities, 615474 Sinoatrial node dysfunction and deafness, 614896
CACNA2D2	94	93,2	99,2	97,6	Cerebellar atrophy with seizures and variable developmental delay, 618501
CAD	100	99,2	100	100	Developmental and epileptic encephalopathy 50, 616457

CAMK2A	99,9	99	99,8	98,7	?Mental retardation, autosomal recessive 63, 618095 Mental retardation, autosomal dominant 53, 617798
CANT1	100	99,9	100	100	Desbuquois dysplasia 1, 251450 Epiphyseal dysplasia, multiple, 7, 617719
CAPN1	100	100	100	100	Spastic paraplegia 76, autosomal recessive, 616907
CAPN10	100	99,6	100	100	{Diabetes mellitus, noninsulin-dependent 1}, 601283
CAPN3	97,8	97,2	97,9	97,9	Muscular dystrophy, limb-girdle, autosomal recessive 1, 253600 Muscular dystrophy, limb-girdle, autosomal dominant 4, 618129
CARD11	100	99,9	100	100	Immunodeficiency 11B with atopic dermatitis, 617638 B-cell expansion with NFKB and T-cell anergy, 616452 Immunodeficiency 11A, 615206
CARD9	99,9	98,4	100	100	Candidiasis, familial, 2, autosomal recessive, 212050
CARS2	100	100	100	99,2	Combined oxidative phosphorylation deficiency 27, 616672
CASP14	100	100	100	100	Ichthyosis, congenital, autosomal recessive 12, 617320
CASP8	95,6	95,4	95,6	95,6	{Lung cancer, protection against}, 211980 ?Autoimmune lymphoproliferative syndrome, type IIB, 607271 Hepatocellular carcinoma, somatic, 114550 {Breast cancer, protection against}, 114480
CASQ2	100	100	100	100	Ventricular tachycardia, catecholaminergic polymorphic, 2, 611938
CASR	100	99,9	100	100	Hypocalcemia, autosomal dominant, with Bartter syndrome, 601198 Hypocalciuric hypercalcemia, type I, 145980 {Epilepsy idiopathic generalized, susceptibility to, 8}, 612899 Hypocalcemia, autosomal dominant, 601198 Hyperparathyroidism, neonatal, 239200
CAST	98,3	95,4	100	100	Peeling skin with leukonychia, acral punctate keratoses, cheilitis, and knuckle pads, 616295
CAT	100	100	100	100	Acatalasemia, 614097
CATSPER1	100	100	100	100	Spermatogenic failure 7, 612997
CAV1	100	100	100	100	Lipodystrophy, familial partial, type 7, 606721 ?Lipodystrophy, congenital generalized, type 3, 612526 Pulmonary hypertension, primary, 3, 615343
CAVIN1	100	100	100	100	Lipodystrophy, congenital generalized, type 4, 613327
CBLIF	100	99,7	100	100	Intrinsic factor deficiency, 261000
CBS	99,8	98,3	100	100	Homocystinuria, B6-responsive and nonresponsive types, 236200 Thrombosis, hyperhomocysteinemic, 236200
CBX2	100	99,8	100	100	?46XY sex reversal 5, 613080
CC2D1A	100	99,3	100	100	Mental retardation, autosomal recessive 3, 608443

CC2D2A	98,5	96,5	97,1	97,1	Meckel syndrome 6, 612284 Joubert syndrome 9, 612285 COACH syndrome 2, 619111
CCBE1	99,8	98,8	100	100	Hennekam lymphangiectasia-lymphedema syndrome 1, 235510
CCDC103	100	100	100	100	Ciliary dyskinesia, primary, 17, 614679
CCDC114	100	100	100	100	Ciliary dyskinesia, primary, 20, 615067
CCDC115	95,3	90	100	100	Congenital disorder of glycosylation, type IIo, 616828
CCDC151	100	99,7	100	100	Ciliary dyskinesia, primary, 30, 616037
CCDC174	99,5	97,1	100	100	Hypotonia, infantile, with psychomotor retardation, 616816
CCDC39	99,5	96,5	100	100	Ciliary dyskinesia, primary, 14, 613807
CCDC40	99,1	98,1	100	100	Ciliary dyskinesia, primary, 15, 613808
CCDC47	99,4	97,5	100	100	Trichohepatoneurodevelopmental syndrome, 618268
CCDC65	99,6	97,1	100	100	Ciliary dyskinesia, primary, 27, 615504
CCDC8	100	100	100	100	3-M syndrome 3, 614205
CCDC88A	96,4	93,1	97,5	97,5	?PEHO syndrome-like, 617507
CCDC88C	100	99,3	100	100	?Spinocerebellar ataxia 40, 616053 Hydrocephalus, congenital, 1, 236600
CCN6	84,7	84,6	84,6	84,6	Progressive pseudorheumatoid dysplasia, 208230
CCNO	100	99,2	100	100	Ciliary dyskinesia, primary, 29, 615872
CCT5	100	99,7	100	100	Neuropathy, hereditary sensory, with spastic paraplegia, 256840
CD151	100	100	100	100	Nephropathy with pretibial epidermolysis bullosa and deafness, 609057 [Blood group, Raph], 179620
CD19	100	100	100	100	Immunodeficiency, common variable, 3, 613493
CD247	100	100	100	100	?Immunodeficiency 25, 610163
CD27	99,9	96,9	100	100	Lymphoproliferative syndrome 2, 615122
CD2AP	99,9	98,8	100	100	Glomerulosclerosis, focal segmental, 3, 607832
CD320	100	99,8	100	100	Methylmalonic aciduria, transient, due to transcobalamin receptor defect, 613646
CD3D	100	100	100	100	Immunodeficiency 19, 615617
CD3E	100	99,5	100	100	Immunodeficiency 18, SCID variant, 615615 Immunodeficiency 18, 615615
CD3G	100	100	100	100	Immunodeficiency 17, CD3 gamma deficient, 615607
CD40	100	100	100	100	Immunodeficiency with hyper-IgM, type 3, 606843
CD55	92,9	85,4	94,1	92	[Blood group Cromer], 613793 Complement hyperactivation, angiopathic thrombosis, and protein-losing enteropathy, 226300
CD59	80	71,6	64,5	64,5	Hemolytic anemia, CD59-mediated, with or without immune-mediated polyneuropathy, 612300
CD79A	100	100	100	100	Agammaglobulinemia 3, 613501
CD79B	100	100	100	100	Agammaglobulinemia 6, 612692

CD81	100	99,9	100	100	Immunodeficiency, common variable, 6, 613496
CD8A	100	99,8	100	100	CD8 deficiency, familial, 608957
CDAN1	100	99,6	100	100	Dyserythropoietic anemia, congenital, type Ia, 224120
CDC14A	100	99	100	100	Deafness, autosomal recessive 32, with or without immotile sperm, 608653
CDC45	99,8	98,5	100	100	Meier-Gorlin syndrome 7, 617063
CDC6	100	100	100	100	?Meier-Gorlin syndrome 5, 613805
CDCA7	100	99,6	100	100	Immunodeficiency-centromeric instability-facial anomalies syndrome 3, 616910
CDH11	100	100	100	100	Elsahy-Waters syndrome, 211380
CDH23	100	100	100	100	{Pituitary adenoma 5, multiple types}, 617540 Deafness, autosomal recessive 12, 601386 Usher syndrome, type 1D/F digenic, 601067 Usher syndrome, type 1D, 601067
CDH3	100	99,5	100	100	Ectodermal dysplasia, ectrodactyly, and macular dystrophy, 225280 Hypotrichosis, congenital, with juvenile macular dystrophy, 601553
CDHR1	99,2	98,1	100	100	Cone-rod dystrophy 15, 613660 Retinitis pigmentosa 65, 613660
CDK10	100	99,9	100	100	Al Kaissi syndrome, 617694
CDK5	100	100	100	100	?Lissencephaly 7 with cerebellar hypoplasia, 616342
CDK5RAP2	99,8	98,9	100	100	Microcephaly 3, primary, autosomal recessive, 604804
CDK6	100	99,6	100	100	?Microcephaly 12, primary, autosomal recessive, 616080
CDSN	100	100	100	100	Hypotrichosis 2, 146520 Peeling skin syndrome 1, 270300
CDT1	99,7	97,5	100	99,1	Meier-Gorlin syndrome 4, 613804
CEACAM16	100	99,5	100	100	Deafness, autosomal recessive 113, 618410 Deafness, autosomal dominant 4B, 614614
CEBPE	100	100	100	100	Specific granule deficiency, 245480
CENPE	98,2	92,2	100	100	?Microcephaly 13, primary, autosomal recessive, 616051
CENPF	99,8	98,5	100	100	Stromme syndrome, 243605
CENPJ	100	99,6	100	100	Microcephaly 6, primary, autosomal recessive, 608393 ?Seckel syndrome 4, 613676
CEP104	100	99,2	100	100	Joubert syndrome 25, 616781
CEP120	100	99,5	100	100	Joubert syndrome 31, 617761 Short-rib thoracic dysplasia 13 with or without polydactyly, 616300
CEP135	99,1	93,6	100	100	Microcephaly 8, primary, autosomal recessive, 614673
CEP152	99,7	98,2	100	100	Microcephaly 9, primary, autosomal recessive, 614852 Seckel syndrome 5, 613823
CEP164	99,9	98,3	100	100	Nephronophthisis 15, 614845

CEP19	100	100	100	100	Morbid obesity and spermatogenic failure, 615703
CEP290	96,1	90	100	100	?Bardet-Biedl syndrome 14, 615991 Leber congenital amaurosis 10, 611755 Senior-Loken syndrome 6, 610189 Meckel syndrome 4, 611134 Joubert syndrome 5, 610188
CEP41	99,8	97,4	100	100	Joubert syndrome 15, 614464
CEP55	100	99,8	100	100	Multinucleated neurons, anhydramnios, renal dysplasia, cerebellar hypoplasia, and hydranencephaly, 236500
CEP57	99,2	93	100	100	Mosaic variegated aneuploidy syndrome 2, 614114
CEP63	99,3	96,5	100	100	?Seckel syndrome 6, 614728
CEP78	98,9	96,8	100	100	Cone-rod dystrophy and hearing loss, 617236
CEP83	99,8	97,4	100	100	Nephronophthisis 18, 615862
CERKL	99,5	96,9	100	100	Retinitis pigmentosa 26, 608380
CERS1	75,4	63,7	94,2	86,4	?Epilepsy, progressive myoclonic, 8, 616230
CERS3	99,9	98,9	100	100	Ichthyosis, congenital, autosomal recessive 9, 615023
CFAP298	100	99,7	100	100	Ciliary dyskinesia, primary, 26, 615500
CFAP410	100	99,3	100	100	Spondylometaphyseal dysplasia, axial, 602271 Retinal dystrophy with macular staphyloma, 617547
CFAP53	99,6	97,4	100	100	Heterotaxy, visceral, 6, autosomal recessive, 614779
CFD	89,3	83,7	100	100	Complement factor D deficiency, 613912
CFH	99,9	99	100	99,9	Basal laminar drusen, 126700 Complement factor H deficiency, 609814 {Hemolytic uremic syndrome, atypical, susceptibility to, 1}, 235400 {Macular degeneration, age-related, 4}, 610698
CFI	99,2	96,8	100	100	Complement factor I deficiency, 610984 {Hemolytic uremic syndrome, atypical, susceptibility to, 3}, 612923 {Macular degeneration, age-related, 13, susceptibility to}, 615439
CFL2	100	99,6	100	100	Nemaline myopathy 7, autosomal recessive, 610687
CFTR	99,6	97,9	100	100	{Bronchiectasis with or without elevated sweat chloride 1, modifier of}, 211400 Cystic fibrosis, 219700 Congenital bilateral absence of vas deferens, 277180 {Pancreatitis, hereditary}, 167800 Sweat chloride elevation without CF, 0 {Hypertrypsinemia, neonatal}, 0
CHAT	93,5	85,7	100	100	Myasthenic syndrome, congenital, 6, presynaptic, 254210
CHKB	100	99,7	100	100	Muscular dystrophy, congenital, megaconial type, 602541

CHMP1A	100	99,8	100	100	Pontocerebellar hypoplasia, type 8, 614961
CHP1	98,5	89,1	100	100	?Spastic ataxia 9, autosomal recessive, 618438
CHRM3	100	100	100	100	Prune belly syndrome, 100100
CHRNA1	100	99,2	100	100	Multiple pterygium syndrome, lethal type, 253290 Myasthenic syndrome, congenital, 1A, slow-channel, 601462 Myasthenic syndrome, congenital, 1B, fast-channel, 608930
CHRNB1	100	99,4	100	100	?Myasthenic syndrome, congenital, 2C, associated with acetylcholine receptor deficiency, 616314 Myasthenic syndrome, congenital, 2A, slow-channel, 616313
CHRND	99,7	97,9	100	100	?Myasthenic syndrome, congenital, 3A, slow-channel, 616321 Myasthenic syndrome, congenital, 3B, fast-channel, 616322 Multiple pterygium syndrome, lethal type, 253290 ?Myasthenic syndrome, congenital, 3C, associated with acetylcholine receptor deficiency, 616323
CHRNE	100	100	100	100	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	100	Escobar syndrome, 265000 Multiple pterygium syndrome, lethal type, 253290
CHST11	100	100	100	100	?Osteochondrodysplasia, brachydactyly, and overlapping malformed digits, 618167
CHST14	99,9	98,9	100	100	Ehlers-Danlos syndrome, musculocontractural type 1, 601776
CHST3	100	99,4	100	100	Spondyloepiphyseal dysplasia with congenital joint dislocations, 143095
CHST6	100	100	100	100	Macular corneal dystrophy, 217800
CHST8	100	100	100	100	?Peeling skin syndrome 3, 616265
CHSY1	97,2	95,7	99,7	98	Temtamy preaxial brachydactyly syndrome, 605282
CHUK	100	99,1	100	100	Cocoon syndrome, 613630
CIB2	99,7	97	100	99,9	Deafness, autosomal recessive 48, 609439 Usher syndrome, type IJ, 614869
CIDEC	100	97,9	100	100	?Lipodystrophy, familial partial, type 5, 615238
CIITA	100	99,5	100	100	Bare lymphocyte syndrome, type II, complementation group A, 209920 {Rheumatoid arthritis, susceptibility to}, 180300
CILK1	99,9	98,7	100	100	Endocrine-cerebroosteodysplasia, 612651 {Epilepsy, juvenile myoclonic, susceptibility to, 10}, 617924
CISD2	83,4	83,4	100	100	Wolfram syndrome 2, 604928
CIT	100	99,4	100	100	Microcephaly 17, primary, autosomal recessive, 617090
CKAP2L	99,7	98,6	100	100	Filippi syndrome, 272440
CLCF1	100	99,4	100	100	Cold-induced sweating syndrome 2, 610313

CLCN1	100	99,2	100	100	Myotonia congenita, dominant, 160800 Myotonia congenita, recessive, 255700 Myotonia levior, recessive, 0
CLCN2	100	99,5	100	100	{Epilepsy, juvenile absence, susceptibility to, 2}, 607628 {Epilepsy, idiopathic generalized, susceptibility to, 11}, 607628 {Epilepsy, juvenile myoclonic, susceptibility to, 8}, 607628 Leukoencephalopathy with ataxia, 615651 Hyperaldosteronism, familial, type II, 605635
CLCN7	99,7	98,4	100	100	Osteopetrosis, autosomal recessive 4, 611490 Osteopetrosis, autosomal dominant 2, 166600 Hypopigmentation, organomegaly, and delayed myelination and development, 618541
CLCNKB	99,1	95,9	100	100	Bartter syndrome, type 3, 607364 Bartter syndrome, type 4b, digenic, 613090
CLDN1	100	100	100	100	Ichthyosis, leukocyte vacuoles, alopecia, and sclerosing cholangitis, 607626
CLDN10	100	100	100	100	HELIX syndrome, 617671
CLDN14	100	99,7	100	100	Deafness, autosomal recessive 29, 614035
CLDN16	100	100	100	100	Hypomagnesemia 3, renal, 248250
CLDN19	98,5	93,1	100	100	Hypomagnesemia 5, renal, with ocular involvement, 248190
CLIC5	89,9	88	100	100	?Deafness, autosomal recessive 103, 616042
CLIP1	100	99	100	100	No OMIM disease ID
CLMP	100	99,6	100	100	Congenital short bowel syndrome, 615237
CLN3	92,5	91,8	92,5	92,5	Ceroid lipofuscinosis, neuronal, 3, 204200
CLN5	69,3	66,3	72,1	71,6	Ceroid lipofuscinosis, neuronal, 5, 256731
CLN6	99,9	97,1	100	100	Ceroid lipofuscinosis, neuronal, Kufs type, adult onset, 204300 Ceroid lipofuscinosis, neuronal, 6, 601780
CLN8	83,5	83,5	100	100	Ceroid lipofuscinosis, neuronal, 8, 600143 Ceroid lipofuscinosis, neuronal, 8, Northern epilepsy variant, 610003
CLP1	100	100	100	100	Pontocerebellar hypoplasia, type 10, 615803
CLPB	94,9	94,9	100	100	3-methylglutaconic aciduria, type VII, with cataracts, neurologic involvement and neutropenia, 616271
CLPP	100	99,1	100	100	Perrault syndrome 3, 614129
CLRN1	100	99,8	100	100	Retinitis pigmentosa 61, 614180 Usher syndrome, type 3A, 276902
CNGA1	91,7	86,3	91	91	Retinitis pigmentosa 49, 613756
CNGA3	100	99,7	100	100	Achromatopsia 2, 216900
CNGB1	99,4	97,5	100	100	Retinitis pigmentosa 45, 613767
CNGB3	99,4	95,9	100	100	Achromatopsia 3, 262300

CNNM2	100	100	100	100	Hypomagnesemia 6, renal, 613882 Hypomagnesemia, seizures, and mental retardation, 616418
CNNM4	99,8	98,9	99,7	98,8	Jalili syndrome, 217080
CNPY3	100	99,3	100	100	Developmental and epileptic encephalopathy 60, 617929
CNTN1	99,9	98,9	100	100	?Myopathy, congenital, Compton-North, 612540
CNTN2	92,7	92,7	100	100	?Epilepsy, myoclonic, familial adult, 5, 615400
CNTNAP1	100	99,8	100	100	Lethal congenital contracture syndrome 7, 616286 Hypomyelinating neuropathy, congenital, 3, 618186
CNTNAP2	100	99,8	100	100	{Autism susceptibility 15}, 612100 Pitt-Hopkins like syndrome 1, 610042 Cortical dysplasia-focal epilepsy syndrome, 610042
COA5	99,1	88,9	85,2	85,2	?Mitochondrial complex IV, deficiency, nuclear type 9, 616500
COA6	99,9	98,4	100	100	Mitochondrial complex IV deficiency, nuclear type 13, 616501
COA7	100	100	100	100	Spinocerebellar ataxia, autosomal recessive, with axonal neuropathy 3, 618387
COA8	81,9	80,7	93,5	93,4	Mitochondrial complex IV deficiency, nuclear type 17, 619061
COASY	100	100	100	100	Neurodegeneration with brain iron accumulation 6, 615643 Pontocerebellar hypoplasia, type 12, 618266
COCH	95,2	93,2	100	100	?Deafness, autosomal recessive 110, 618094 Deafness, autosomal dominant 9, 601369
COG1	100	100	100	100	Congenital disorder of glycosylation, type IIg, 611209
COG2	99,9	98,5	100	100	?Congenital disorder of glycosylation, type IIq, 617395
COG4	100	99,9	100	100	Saul-Wilson syndrome, 618150 Congenital disorder of glycosylation, type IIj, 613489
COG5	99,7	97,6	100	100	Congenital disorder of glycosylation, type IIi, 613612
COG6	99,1	93,9	100	100	Congenital disorder of glycosylation, type III, 614576 Shaheen syndrome, 615328
COG7	100	100	100	100	Congenital disorder of glycosylation, type IIe, 608779
COG8	99,9	98,6	100	100	Congenital disorder of glycosylation, type IIh, 611182
COL11A1	96,2	92,8	100	100	Stickler syndrome, type II, 604841 Marshall syndrome, 154780 ?Deafness, autosomal dominant 37, 618533 {Lumbar disc herniation, susceptibility to}, 603932 Fibrochondrogenesis 1, 228520
COL11A2	100	99,7	100	100	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	99,4	100	100	Bethlem myopathy 2, 616471 ?Ullrich congenital muscular dystrophy 2, 616470
COL13A1	93,9	93,8	100	100	Myasthenic syndrome, congenital, 19, 616720
COL17A1	98,7	96,8	100	100	Epithelial recurrent erosion dystrophy, 122400 Epidermolysis bullosa, junctional, non-Herlitz type, 226650 Epidermolysis bullosa, junctional, localisata variant, 226650
COL18A1	98,1	95,6	100	100	Knobloch syndrome, type 1, 267750 Glaucoma, primary closed-angle, 618880
COL1A2	99,4	96,9	100	100	Combined osteogenesis imperfecta and Ehlers-Danlos syndrome 2, 619120 {Osteoporosis, postmenopausal}, 166710 Ehlers-Danlos syndrome, cardiac valvular type, 225320 Ehlers-Danlos syndrome, arthrochalasia type, 2, 617821 Osteogenesis imperfecta, type II, 166210 Osteogenesis imperfecta, type IV, 166220 Osteogenesis imperfecta, type III, 259420
COL25A1	95,8	95,3	99,9	99,9	Fibrosis of extraocular muscles, congenital, 5, 616219
COL27A1	99,9	99,7	100	100	Steel syndrome, 615155
COL3A1	99,6	97,6	100	100	Ehlers-Danlos syndrome, vascular type, 130050 Polymicrogyria with or without vascular-type EDS, 618343
COL4A3	98,7	98	100	100	Alport syndrome 2, autosomal recessive, 203780 Alport syndrome 3, autosomal dominant, 104200 Hematuria, benign familial, 141200
COL4A4	99,9	98,2	100	100	Alport syndrome 2, autosomal recessive, 203780 Hematuria, familial benign, 141200
COL6A1	100	99,4	100	100	Ullrich congenital muscular dystrophy 1, 254090 Bethlem myopathy 1, 158810
COL6A2	100	99,8	100	100	Bethlem myopathy 1, 158810 Ullrich congenital muscular dystrophy 1, 254090 ?Myosclerosis, congenital, 255600
COL6A3	100	99,8	100	100	Bethlem myopathy 1, 158810 Dystonia 27, 616411 Ullrich congenital muscular dystrophy 1, 254090
COL7A1	99,9	99,1	100	100	EBD inversa, 226600 Epidermolysis bullosa dystrophica, AR, 226600 Toenail dystrophy, isolated, 607523 EBD, Bart type, 132000

					Transient bullous of the newborn, 131705 Epidermolysis bullosa dystrophica, AD, 131750 Epidermolysis bullosa pruriginosa, 604129 Epidermolysis bullosa, pretibial, 131850 EBD, localisata variant, 0
COL9A1	100	99,2	100	100	?Epiphyseal dysplasia, multiple, 6, 614135 Stickler syndrome, type IV, 614134
COL9A2	99,9	99	100	100	?Stickler syndrome, type V, 614284 Epiphyseal dysplasia, multiple, 2, 600204
COLEC10	100	100	100	100	3MC syndrome 3, 248340
COLEC11	100	100	100	100	3MC syndrome 2, 265050
COLGALT1	93,3	89	98,6	97	Brain small vessel disease 3, 618360
COLQ	100	99,2	100	100	Myasthenic syndrome, congenital, 5, 603034
COPB2	99,9	99,3	100	100	?Microcephaly 19, primary, autosomal recessive, 617800
COQ2	98	95,3	97,2	97,2	{Multiple system atrophy, susceptibility to}, 146500 Coenzyme Q10 deficiency, primary, 1, 607426
COQ4	90,9	89,3	100	100	Coenzyme Q10 deficiency, primary, 7, 616276
COQ6	99,9	98,4	100	100	Coenzyme Q10 deficiency, primary, 6, 614650
COQ7	100	99,8	100	100	?Coenzyme Q10 deficiency, primary, 8, 616733
COQ8A	100	99,5	100	100	Coenzyme Q10 deficiency, primary, 4, 612016
COQ8B	100	99,3	100	100	Nephrotic syndrome, type 9, 615573
COQ9	100	97,9	100	100	Coenzyme Q10 deficiency, primary, 5, 614654
CORO1A	100	98,6	100	100	Immunodeficiency 8, 615401
COX10	100	100	100	100	Mitochondrial complex IV deficiency, nuclear type 3, 619046
COX14	100	100	100	100	?Mitochondrial complex IV deficiency, nuclear type 10, 619053
COX15	99,9	98,8	100	100	Mitochondrial complex IV deficiency, nuclear type 6, 615119
COX20	97,8	88,3	100	100	Mitochondrial complex IV deficiency, nuclear type 11, 619054
COX4I2	100	100	100	100	Exocrine pancreatic insufficiency, dyserythropoietic anemia, and calvarial hyperostosis, 612714
COX6A1	100	99,5	100	100	Charcot-Marie-Tooth disease, recessive intermediate D, 616039
COX6A2	99,2	93,7	100	100	Mitochondrial complex IV deficiency, nuclear type 18, 619062
COX6B1	100	100	100	100	Mitochondrial complex IV deficiency, nuclear type 7, 619051
COX8A	100	100	100	100	?Mitochondrial complex IV deficiency, nuclear type 15, 619059
CP	94,8	88,9	100	100	[Hypoceruloplasminemia, hereditary], 604290 Hemosiderosis, systemic, due to aceruloplasminemia, 604290 Cerebellar ataxia, 604290
CPA6	99,6	97,5	100	100	Febrile seizures, familial, 11, 614418 Epilepsy, familial temporal lobe, 5, 614417

CPAMD8	95,8	92,8	99,9	99,6	Anterior segment dysgenesis 8, 617319
CPLANE1	99,7	98,4	100	100	Joubert syndrome 17, 614615 Orofaciodigital syndrome VI, 277170
CPLX1	100	100	100	100	Developmental and epileptic encephalopathy 63, 617976
CPN1	99,9	99,4	100	100	Carboxypeptidase N deficiency, 212070
CPOX	99,9	95,4	100	100	Harderoporphyria, 618892 Coproporphyria, 121300
CPS1	100	99,9	100	100	Carbamoylphosphate synthetase I deficiency, 237300 {Pulmonary hypertension, neonatal, susceptibility to}, 615371
CPT1A	100	98,9	100	100	CPT deficiency, hepatic, type IA, 255120
CPT2	98,2	97,8	100	100	CPT II deficiency, myopathic, stress-induced, 255110 CPT II deficiency, infantile, 600649 CPT II deficiency, lethal neonatal, 608836 {Encephalopathy, acute, infection-induced, 4, susceptibility to}, 614212
CR2	100	99,8	100	100	{Systemic lupus erythematosus, susceptibility to, 9}, 610927 Immunodeficiency, common variable, 7, 614699
CRADD	99,5	96,3	100	100	Mental retardation, autosomal recessive 34, with variant lissencephaly, 614499
CRAT	100	99,8	100	100	?Neurodegeneration with brain iron accumulation 8, 617917
CRB1	100	99,9	100	100	Pigmented paravenous chorioretinal atrophy, 172870 Retinitis pigmentosa-12, 600105 Leber congenital amaurosis 8, 613835
CRB2	98,5	93	100	100	Ventriculomegaly with cystic kidney disease, 219730 Focal segmental glomerulosclerosis 9, 616220
CRBN	88,2	87,7	97	92,9	Mental retardation, autosomal recessive 2, 607417
CREB3L1	100	99,9	100	100	Osteogenesis imperfecta, type XVI, 616229
CRIPT	98,1	93,2	100	100	Short stature with microcephaly and distinctive facies, 615789
CRLF1	91	89,8	97,9	95,2	Cold-induced sweating syndrome 1, 272430
CRPPA	98,5	94,8	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	99,8	98,8	100	100	Osteogenesis imperfecta, type VII, 610682
CRYAA	99,9	97,5	100	100	Cataract 9, multiple types, 604219
CRYAB	100	99,2	100	100	Myopathy, myofibrillar, 2, 608810 Cardiomyopathy, dilated, 1II, 615184 Cataract 16, multiple types, 613763 Myopathy, myofibrillar, fatal infantile hypertonic, alpha-B crystallin-related, 613869
CRYBB1	100	100	100	100	Cataract 17, multiple types, 611544
CRYBB3	100	100	100	100	Cataract 22, 609741

CSF1R	99,9	99,3	100	100	Brain abnormalities, neurodegeneration, and dysosteosclerosis, 618476 Leukoencephalopathy, diffuse hereditary, with spheroids, 221820
CSF2RB	100	99	100	100	Surfactant metabolism dysfunction, pulmonary, 5, 614370
CSPP1	99,8	98,7	100	100	Joubert syndrome 21, 615636
CSTA	100	99,8	100	100	Peeling skin syndrome 4, 607936
CSTB	99,6	89,8	100	100	Epilepsy, progressive myoclonic 1A (Unverricht and Lundborg), 254800
CTC1	100	99,6	100	100	Cerebroretinal microangiopathy with calcifications and cysts, 612199
CTDP1	88,4	84,3	100	99,4	Congenital cataracts, facial dysmorphism, and neuropathy, 604168
CTNNA2	100	99,8	100	100	Cortical dysplasia, complex, with other brain malformations 9, 618174
CTNS	100	99,8	100	100	Cystinosis, nephropathic, 219800 Cystinosis, ocular nonnephropathic, 219750 Cystinosis, late-onset juvenile or adolescent nephropathic, 219900 Cystinosis, atypical nephropathic, 219800
CTPS1	93	93	93	93	Immunodeficiency 24, 615897
CTSA	100	100	100	100	Galactosialidosis, 256540
CTSC	100	100	100	100	Periodontitis 1, juvenile, 170650 Papillon-Lefevre syndrome, 245000 Haim-Munk syndrome, 245010
CTSD	98,4	95	100	100	Ceroid lipofuscinosis, neuronal, 10, 610127
CTSF	84	79,3	100	99,9	Ceroid lipofuscinosis, neuronal, 13, Kufs type, 615362
CTSK	100	99,9	100	100	Pycnodysostosis, 265800
CTU2	99,7	97,7	100	100	Microcephaly, facial dysmorphism, renal agenesis, and ambiguous genitalia syndrome, 618142
CUBN	99,7	98,3	100	100	[Proteinuria, chronic benign], 618884 Imerslund-Grasbeck syndrome 1, 261100
CUL7	100	99,3	100	100	3-M syndrome 1, 273750
CWC27	99,3	96,5	100	100	Retinitis pigmentosa with or without skeletal anomalies, 250410
CWF19L1	100	99,8	100	100	Spinocerebellar ataxia, autosomal recessive 17, 616127
CYB5A	100	100	100	100	Methemoglobinemia and ambiguous genitalia, 250790
CYB5R3	98,4	98	99,8	98,9	Methemoglobinemia, type I, 250800 Methemoglobinemia, type II, 250800
CYBA	95	82,4	100	100	Chronic granulomatous disease 4, autosomal recessive, 233690
CYC1	97,5	89,2	99,9	98,7	Mitochondrial complex III deficiency, nuclear type 6, 615453
CYP11A1	99,3	96,1	100	100	Adrenal insufficiency, congenital, with 46XY sex reversal, partial or complete, 613743
CYP11B1	100	100	100	100	Adrenal hyperplasia, congenital, due to 11-beta-hydroxylase deficiency, 202010 Aldosteronism, glucocorticoid-remediable, 103900
CYP11B2	100	100	100	100	Hypoaldosteronism, congenital, due to CMO II deficiency, 610600 Hypoaldosteronism, congenital, due to CMO I deficiency, 203400

					Aldosterone to renin ratio raised, 0 {Low renin hypertension, susceptibility to}, 0
CYP17A1	100	99,5	100	100	17-alpha-hydroxylase/17,20-lyase deficiency, 202110 17,20-lyase deficiency, isolated, 202110
CYP19A1	98,8	96,8	100	100	Aromatase deficiency, 613546 Aromatase excess syndrome, 139300
CYP1B1	100	100	100	100	Glaucoma 3A, primary open angle, congenital, juvenile, or adult onset, 231300 Anterior segment dysgenesis 6, multiple subtypes, 617315
CYP21A2	97,8	88,4	100	100	Adrenal hyperplasia, congenital, due to 21-hydroxylase deficiency, 201910 Hyperandrogenism, nonclassic type, due to 21-hydroxylase deficiency, 201910
CYP24A1	100	99,9	100	100	Hypercalcemia, infantile, 1, 143880
CYP26B1	100	99,9	100	100	Craniosynostosis with radiohumeral fusions and other skeletal and craniofacial anomalies, 614416
CYP26C1	99,7	97,1	100	99,8	Focal facial dermal dysplasia 4, 614974
CYP27A1	98,9	96,7	100	100	Cerebrotendinous xanthomatosis, 213700
CYP27B1	99,9	99,3	100	100	Vitamin D-dependent rickets, type I, 264700
CYP2C8	99,9	98,6	100	100	{Drug metabolism, altered, CYP2C8-related}, 618018
CYP2R1	99,4	95,6	100	100	Rickets due to defect in vitamin D 25-hydroxylation deficiency, 600081
CYP2U1	94,8	91,5	100	99,9	Spastic paraplegia 56, autosomal recessive, 615030
CYP4F22	100	99,4	100	100	Ichthyosis, congenital, autosomal recessive 5, 604777
CYP4V2	99,9	98,4	100	100	Bietti crystalline corneoretinal dystrophy, 210370
CYP7B1	98	92,8	100	100	Spastic paraplegia 5A, autosomal recessive, 270800 Bile acid synthesis defect, congenital, 3, 613812
D2HGDH	99,2	97,2	100	100	D-2-hydroxyglutaric aciduria, 600721
DAG1	100	100	100	100	Muscular dystrophy-dystroglycanopathy (limb-girdle), type C, 9, 613818 Muscular dystrophy-dystroglycanopathy (congenital with brain and eye anomalies), type A, 9, 616538
DARS1	100	99,3	100	100	Hypomyelination with brainstem and spinal cord involvement and leg spasticity, 615281
DARS2	94,9	94,3	100	100	Leukoencephalopathy with brain stem and spinal cord involvement and lactate elevation, 611105
DBH	100	100	100	100	Orthostatic hypotension 1, due to DBH deficiency, 223360
DBT	99,8	98	100	100	Maple syrup urine disease, type II, 248600
DCAF17	98,9	93,3	100	100	Woodhouse-Sakati syndrome, 241080
DCC	100	100	100	100	Esophageal carcinoma, somatic, 133239 Gaze palsy, familial horizontal, with progressive scoliosis, 2, 617542 Mirror movements 1 and/or agenesis of the corpus callosum, 157600 Colorectal cancer, somatic, 114500
DCDC2	100	99,9	100	100	Sclerosing cholangitis, neonatal, 617394 Nephronophthisis 19, 616217 ?Deafness, autosomal recessive 66, 610212

DCHS1	99,8	99,1	100	100	Mitral valve prolapse 2, 607829 Van Maldergem syndrome 1, 601390
DCLRE1C	100	99,4	100	100	Omenn syndrome, 603554 Severe combined immunodeficiency, Athabascan type, 602450
DCPS	91,3	91,2	100	100	Al-Raqad syndrome, 616459
DDB2	99,6	97,5	100	100	Xeroderma pigmentosum, group E, DDB-negative subtype, 278740
DDC	99,7	96,4	100	100	Aromatic L-amino acid decarboxylase deficiency, 608643
DDHD1	97,9	95,8	100	100	Spastic paraplegia 28, autosomal recessive, 609340
DDHD2	100	99,6	100	100	Spastic paraplegia 54, autosomal recessive, 615033
DDOST	100	99,9	100	100	?Congenital disorder of glycosylation, type Ir, 614507
DDR2	100	99,9	100	100	Spondylometaepiphyseal dysplasia, short limb-hand type, 271665 Warburg-Cinotti syndrome, 618175
DDRGK1	100	99,9	100	100	Spondyloepimetaphyseal dysplasia, Shohat type, 602557
DDX11	85,2	80,7	100	100	Warsaw breakage syndrome, 613398
DDX59	100	100	100	100	Orofaciodigital syndrome V, 174300
DEAF1	97,3	88,8	100	98,7	Vulto-van Silfout-de Vries syndrome, 615828 Neurodevelopmental disorder with hypotonia, impaired expressive language, and with or without seizures, 617171
DEGS1	100	100	100	100	Leukodystrophy, hypomyelinating, 18, 618404
DENND5A	100	99,4	100	100	Developmental and epileptic encephalopathy 49, 617281
DES	100	99,7	100	100	Cardiomyopathy, dilated, 1I, 604765 Scapuloperoneal syndrome, neurogenic, Kaeser type, 181400 Myopathy, myofibrillar, 1, 601419
DGAT1	91,9	87,6	99,7	98,6	?Diarrhea 7, protein-losing enteropathy type, 615863
DGKE	99,8	98,1	100	100	{Hemolytic uremic syndrome, atypical, susceptibility to, 7}, 615008 Nephrotic syndrome, type 7, 615008
DGUOK	100	99,4	100	100	Progressive external ophthalmoplegia with mitochondrial DNA deletions, autosomal recessive 4, 617070 Portal hypertension, noncirrhotic, 617068 Mitochondrial DNA depletion syndrome 3 (hepatocerebral type), 251880
DHCR24	97,7	97,7	97,7	97,7	Desmosterolosis, 602398
DHCR7	100	100	100	100	Smith-Lemli-Opitz syndrome, 270400
DHDDS	99	95	95,2	95,2	Retinitis pigmentosa 59, 613861 Developmental delay and seizures with or without movement abnormalities, 617836 ?Congenital disorder of glycosylation, type 1bb, 613861
DHFR	92,1	78,9	100	100	Megaloblastic anemia due to dihydrofolate reductase deficiency, 613839

DHH	100	100	100	100	46XY sex reversal 7, 233420 46XY gonadal dysgenesis with minifascicular neuropathy, 607080
DHODH	100	100	100	100	Miller syndrome, 263750
DHPS	100	99,7	93,3	93,2	Neurodevelopmental disorder with seizures and speech and walking impairment, 618480
DHTKD1	99,9	98,9	100	100	2-aminoadipic 2-oxoadipic aciduria, 204750 ?Charcot-Marie-Tooth disease, axonal, type 2Q, 615025
DHX38	100	99,3	100	100	Retinitis pigmentosa 84, 618220
DIAPH1	99,8	99	99,5	98	Seizures, cortical blindness, microcephaly syndrome, 616632 Deafness, autosomal dominant 1, with or without thrombocytopenia, 124900
DIS3L2	100	99,8	100	100	Perlman syndrome, 267000
DLAT	100	99,7	100	100	Pyruvate dehydrogenase E2 deficiency, 245348
DLD	100	99,7	100	100	Dihydrolipoamide dehydrogenase deficiency, 246900
DLL3	92,1	87	100	99,1	Spondylocostal dysostosis 1, autosomal recessive, 277300
DLX5	100	99,9	100	100	Split-hand/foot malformation 1, 183600 ?Split-hand/foot malformation 1 with sensorineural hearing loss, 220600
DMGDH	100	99,7	100	100	Dimethylglycine dehydrogenase deficiency, 605850
DMP1	100	99,9	100	100	Hypophosphatemic rickets, AR, 241520
DMXL2	99,9	99,1	100	100	Developmental and epileptic encephalopathy 81, 618663 ?Deafness, autosomal dominant 71, 617605 ?Polyendocrine-polyneuropathy syndrome, 616113
DNA2	99,8	98,3	100	100	Progressive external ophthalmoplegia with mitochondrial DNA deletions, autosomal dominant 6, 615156 ?Seckel syndrome 8, 615807
DNAAF1	100	99,8	100	100	Ciliary dyskinesia, primary, 13, 613193
DNAAF2	99,9	98,9	100	100	Ciliary dyskinesia, primary, 10, 612518
DNAAF3	99,5	96,1	100	100	Ciliary dyskinesia, primary, 2, 606763
DNAAF4	99,8	97	100	100	{Dyslexia, susceptibility to, 1}, 127700 Ciliary dyskinesia, primary, 25, 615482
DNAAF5	84,6	78,6	99,1	97,5	Ciliary dyskinesia, primary, 18, 614874
DNAH1	100	99,7	100	100	?Ciliary dyskinesia, primary, 37, 617577 Spermatogenic failure 18, 617576
DNAH11	99,9	99	100	100	Ciliary dyskinesia, primary, 7, with or without situs inversus, 611884
DNAH5	99,9	99,3	100	100	Ciliary dyskinesia, primary, 3, with or without situs inversus, 608644
DNAH9	99,5	98,3	100	100	Ciliary dyskinesia, primary, 40, 618300
DNAI1	100	100	100	100	Ciliary dyskinesia, primary, 1, with or without situs inversus, 244400
DNAI2	98,6	96,2	100	100	Ciliary dyskinesia, primary, 9, with or without situs inversus, 612444
DNAJB2	100	100	100	100	Spinal muscular atrophy, distal, autosomal recessive, 5, 614881

DNAJC12	87,4	87,4	100	100	Hyperphenylalaninemia, mild, non-BH4-deficient, 617384
DNAJC19	98,9	96,2	100	100	3-methylglutaconic aciduria, type V, 610198
DNAJC21	99,8	98,7	100	100	Bone marrow failure syndrome 3, 617052
DNAJC3	100	99,7	100	100	?Ataxia, combined cerebellar and peripheral, with hearing loss and diabetes mellitus, 616192
DNAJC6	100	99,4	100	100	Parkinson disease 19b, early-onset, 615528 Parkinson disease 19a, juvenile-onset, 615528
DNAL1	99	96,8	100	100	Ciliary dyskinesia, primary, 16, 614017
DNASE1L3	100	100	100	100	Systemic lupus erythematosus 16, 614420
DNM1L	99,9	98,5	100	100	Encephalopathy, lethal, due to defective mitochondrial peroxisomal fission 1, 614388 Optic atrophy 5, 610708
DNM2	98,1	94,5	100	100	Lethal congenital contracture syndrome 5, 615368 Charcot-Marie-Tooth disease, axonal type 2M, 606482 Centronuclear myopathy 1, 160150 Charcot-Marie-Tooth disease, dominant intermediate B, 606482
DNMBP	100	99,7	100	100	Cataract 48, 618415
DNMT3B	100	100	100	100	Immunodeficiency-centromeric instability-facial anomalies syndrome 1, 242860
DOCK2	100	99,6	100	100	Immunodeficiency 40, 616433
DOCK3	100	99	100	100	Neurodevelopmental disorder with impaired intellectual development, hypotonia, and ataxia, 618292
DOCK6	99,3	98,9	100	100	Adams-Oliver syndrome 2, 614219
DOCK7	99,8	98,2	100	99,9	Developmental and epileptic encephalopathy 23, 615859
DOCK8	100	99,6	100	100	Hyper-IgE recurrent infection syndrome, autosomal recessive, 243700
DOK7	95,1	91,6	100	100	Fetal akinesia deformation sequence 3, 618389 Myasthenic syndrome, congenital, 10, 254300
DOLK	100	100	100	100	Congenital disorder of glycosylation, type Im, 610768
DONSON	91,7	85,3	100	100	Microcephaly-micromelia syndrome, 251230 Microcephaly, short stature, and limb abnormalities, 617604
DPAGT1	100	100	100	100	Congenital disorder of glycosylation, type Ij, 608093 Myasthenic syndrome, congenital, 13, with tubular aggregates, 614750
DPH1	100	99,9	100	100	Developmental delay with short stature, dysmorphic facial features, and sparse hair, 616901
DPM1	98,2	91,3	99,7	97,1	Congenital disorder of glycosylation, type Ie, 608799
DPM2	100	98,7	100	100	Congenital disorder of glycosylation, type Iu, 615042
DPM3	100	100	100	100	Muscular dystrophy-dystroglycanopathy (limb-girdle), type C, 15, 612937 ?Muscular dystrophy-dystroglycanopathy (congenital with impaired intellectual development), type B, 15, 618992
DPY19L2	74,5	71,2	100	100	Spermatogenic failure 9, 613958
DPYD	99,7	97,7	100	100	Dihydropyrimidine dehydrogenase deficiency, 274270 5-fluorouracil toxicity, 274270

DPYS	100	99,9	100	100	Dihydropyrimidinuria, 222748
DRAM2	100	99,9	100	100	Cone-rod dystrophy 21, 616502
DRC1	100	99,5	100	100	Ciliary dyskinesia, primary, 21, 615294
DSC2	99,8	98,4	100	100	Arrhythmogenic right ventricular dysplasia 11 with mild palmoplantar keratoderma and woolly hair, 610476 Arrhythmogenic right ventricular dysplasia 11, 610476
DSC3	99,5	96,8	100	100	?Hypotrichosis and recurrent skin vesicles, 613102
DSE	99	96,1	100	100	Ehlers-Danlos syndrome, musculocontractural type 2, 615539
DSG4	100	99,2	100	100	Hypotrichosis 6, 607903
DSP	100	99,6	100	100	Keratosis palmoplantaris striata II, 612908 Arrhythmogenic right ventricular dysplasia 8, 607450 Epidermolysis bullosa, lethal acantholytic, 609638 Cardiomyopathy, dilated, with woolly hair and keratoderma, 605676 Skin fragility-woolly hair syndrome, 607655 Dilated cardiomyopathy with woolly hair, keratoderma, and tooth agenesis, 615821
DST	95,5	95	95,6	95,6	?Neuropathy, hereditary sensory and autonomic, type VI, 614653 Epidermolysis bullosa simplex, autosomal recessive 2, 615425
DSTYK	99,9	99,2	100	100	Congenital anomalies of kidney and urinary tract 1, 610805 Spastic paraplegia 23, 270750
DTNBP1	99,8	98,7	100	100	Hermansky-Pudlak syndrome 7, 614076
DUOX2	96,7	94,7	100	100	Thyroid dyshormonogenesis 6, 607200
DUOXA2	100	100	100	100	Thyroid dyshormonogenesis 5, 274900
DYM	97,4	96,5	100	100	Smith-McCort dysplasia, 607326 Dyggve-Melchior-Clausen disease, 223800
DYNC1I2	84,4	68,8	100	100	Neurodevelopmental disorder with microcephaly and structural brain anomalies, 618492
DYNC2H1	98,8	95,5	100	100	Short-rib thoracic dysplasia 3 with or without polydactyly, 613091
DYNC2LI1	99,7	97,6	100	100	Short-rib thoracic dysplasia 15 with polydactyly, 617088
DYSF	100	99,9	100	100	Miyoshi muscular dystrophy 1, 254130 Myopathy, distal, with anterior tibial onset, 606768 Muscular dystrophy, limb-girdle, autosomal recessive 2, 253601
DZIP1L	99,9	99	100	100	Polycystic kidney disease 5, 617610
EARS2	99,8	97,7	100	100	Combined oxidative phosphorylation deficiency 12, 614924
ECEL1	95,4	90	100	100	Arthrogryposis, distal, type 5D, 615065
ECHS1	99,9	99	100	100	Mitochondrial short-chain enoyl-CoA hydratase 1 deficiency, 616277
ECM1	100	99,6	100	100	Urbach-Wiethe disease, 247100

EDAR	100	99,9	100	100	Ectodermal dysplasia 10A, hypohidrotic/hair/nail type, autosomal dominant, 129490 Ectodermal dysplasia 10B, hypohidrotic/hair/tooth type, autosomal recessive, 224900 [Hair morphology 1, hair thickness], 612630
EDARADD	99,9	98,8	100	100	Ectodermal dysplasia 11B, hypohidrotic/hair/tooth type, autosomal recessive, 614941 Ectodermal dysplasia 11A, hypohidrotic/hair/tooth type, autosomal dominant, 614940
EDC3	100	99,9	100	100	?Mental retardation, autosomal recessive 50, 616460
EDN1	100	100	100	100	Auriculocondylar syndrome 3, 615706 Question mark ears, isolated, 612798 {High density lipoprotein cholesterol level QTL 7}, 0
EDN3	98,8	98,8	100	100	Waardenburg syndrome, type 4B, 613265 Central hypoventilation syndrome, congenital, 209880 {Hirschsprung disease, susceptibility to, 4}, 613712
EDNRB	98	93,8	100	100	{Hirschsprung disease, susceptibility to, 2}, 600155 Waardenburg syndrome, type 4A, 277580 ABCD syndrome, 600501
EFEMP2	100	100	100	100	Cutis laxa, autosomal recessive, type IB, 614437
EFL1	99,6	98,5	100	100	Shwachman-Diamond syndrome 2, 617941
EGF	99,9	99,7	100	100	Hypomagnesemia 4, renal, 611718
EGFR	100	100	100	99,8	?Inflammatory skin and bowel disease, neonatal, 2, 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
EGR2	100	100	100	100	Dejerine-Sottas disease, 145900 Hypomyelinating neuropathy, congenital, 1, 605253 Charcot-Marie-Tooth disease, type 1D, 607678
EIF2AK3	97,2	94,5	100	100	Wolcott-Rallison syndrome, 226980
EIF2AK4	99,8	98,6	100	100	Pulmonary venoocclusive disease 2, 234810
EIF2B1	100	100	100	100	Leukoencephalopathy with vanishing white matter, 603896
EIF2B2	99,9	99,5	100	100	Ovarioleukodystrophy, 603896 Leukoencephalopathy with vanishing white matter, 603896
EIF2B3	100	100	100	100	Leukoencephalopathy with vanishing white matter, 603896
EIF2B4	100	99,9	100	100	Leukoencephalopathy with vanishing white matter, 603896 Ovarioleukodystrophy, 603896
EIF2B5	100	99,1	100	100	Leukoencephalopathy with vanishing white matter, 603896 Ovarioleukodystrophy, 603896
EIF3F	96,8	84,1	100	100	Mental retardation, autosomal recessive 67, 618295
EIF4A3	100	99,5	100	100	Robin sequence with cleft mandible and limb anomalies, 268305

ELAC2	100	99,7	100	100	Combined oxidative phosphorylation deficiency 17, 615440 {Prostate cancer, hereditary, 2, susceptibility to}, 614731
ELMO2	99,9	99	100	100	Vascular malformation, primary intraosseous, 606893
ELMOD3	100	99,8	100	100	?Deafness, autosomal recessive 88, 615429
ELOVL4	100	99,5	100	100	Spinocerebellar ataxia 34, 133190 Stargardt disease 3, 600110 Ichthyosis, spastic quadriplegia, and mental retardation, 614457
ELP1	99,8	99	100	100	Dysautonomia, familial, 223900
ELP2	99,9	98,8	100	100	Mental retardation, autosomal recessive 58, 617270
EMC1	100	99,3	100	100	Cerebellar atrophy, visual impairment, and psychomotor retardation, 616875
EMG1	100	100	100	100	Bowen-Conradi syndrome, 211180
EML1	99,7	98,4	100	100	Band heterotopia, 600348
EMP2	99,9	96,7	100	100	Nephrotic syndrome, type 10, 615861
ENAM	100	100	100	100	Amelogenesis imperfecta, type IC, 204650 Amelogenesis imperfecta, type IB, 104500
ENO3	100	99,9	100	100	?Glycogen storage disease XIII, 612932
ENPP1	96,4	91,2	98,7	97,8	Hypophosphatemic rickets, autosomal recessive, 2, 613312 Cole disease, 615522 {Obesity, susceptibility to}, 601665 Arterial calcification, generalized, of infancy, 1, 208000 {Diabetes mellitus, non-insulin-dependent, susceptibility to}, 125853
ENTPD1	100	100	100	100	Spastic paraplegia 64, autosomal recessive, 615683
EOGT	79,4	78,4	91,9	89	Adams-Oliver syndrome 4, 615297
EPB41	85,3	83,9	100	100	Elliptocytosis-1, 611804
EPB42	100	99,5	100	100	Spherocytosis, type 5, 612690
EPCAM	98,6	90,3	99,8	98,3	Colorectal cancer, hereditary nonpolyposis, type 8, 613244 Diarrhea 5, with tufting enteropathy, congenital, 613217
EPG5	99,5	98,5	100	100	Vici syndrome, 242840
EPHX1	99,9	98,8	100	100	?Hypercholanemia, familial, 607748
EPM2A	94,2	91,5	100	97,7	Epilepsy, progressive myoclonic 2A (Lafora), 254780
EPO	99,9	97,6	100	100	Erythrocytosis, familial, 5, 617907 {Microvascular complications of diabetes 2}, 612623 ?Diamond-Blackfan anemia-like, 617911
EPRS1	100	99,6	100	100	Leukodystrophy, hypomyelinating, 15, 617951
EPS8	97	96,2	100	100	?Deafness, autosomal recessive 102, 615974
EPS8L2	84,5	82,5	88	88	Deafness autosomal recessive 106, 617637
ERAL1	100	99,7	100	100	Perrault syndrome 6, 617565

ERBB3	100	99,8	100	100	{?Erythroleukemia, familial, susceptibility to}, 133180 ?Lethal congenital contractural syndrome 2, 607598
ERCC1	100	99,3	100	100	Cerebrooculofacioskeletal syndrome 4, 610758
ERCC2	100	99,7	100	100	Trichothiodystrophy 1, photosensitive, 601675 ?Cerebrooculofacioskeletal syndrome 2, 610756 Xeroderma pigmentosum, group D, 278730
ERCC3	96,9	96,3	100	100	Xeroderma pigmentosum, group B, 610651 Trichothiodystrophy 2, photosensitive, 616390
ERCC4	100	99,9	100	100	Xeroderma pigmentosum, type F/Cockayne syndrome, 278760 Fanconi anemia, complementation group Q, 615272 XFE progeroid syndrome, 610965 Xeroderma pigmentosum, group F, 278760
ERCC5	100	99,7	100	100	Xeroderma pigmentosum, group G/Cockayne syndrome, 278780 Xeroderma pigmentosum, group G, 278780 Cerebrooculofacioskeletal syndrome 3, 616570
ERCC6	100	100	100	100	{Macular degeneration, age-related, susceptibility to, 5}, 613761 {Lung cancer, susceptibility to}, 211980 Cerebrooculofacioskeletal syndrome 1, 214150 Cockayne syndrome, type B, 133540 Premature ovarian failure 11, 616946 UV-sensitive syndrome 1, 600630 De Sanctis-Cacchione syndrome, 278800
ERCC6L2	99,9	99,2	100	100	Bone marrow failure syndrome 2, 615715
ERCC8	99,5	95,8	100	100	Cockayne syndrome, type A, 216400 UV-sensitive syndrome 2, 614621
ERGIC1	95,2	94,6	98,4	98,4	?Arthrogryposis multiplex congenita 2, neurogenic type, 208100
ERLIN1	100	100	100	100	Spastic paraplegia 62, 615681
ERLIN2	100	99,9	100	100	Spastic paraplegia 18, autosomal recessive, 611225
ESCO2	98,7	95,2	100	100	Roberts-SC phocomelia syndrome, 268300
ESPN	44,6	35,8	100	99,8	?Usher syndrome, type 1M, 618632 Deafness, autosomal recessive 36, 609006 Deafness, neurosensory, without vestibular involvement, autosomal dominant, 609006
ESR1	100	99,8	100	100	{Myocardial infarction, susceptibility to}, 608446 Estrogen resistance, 615363 Breast cancer, somatic, 114480 {Migraine, susceptibility to}, 157300
ESRP1	99,9	98,9	100	100	?Deafness, autosomal recessive 109, 618013
ESRRB	96,8	95	100	100	Deafness, autosomal recessive 35, 608565

ETFA	100	100	100	100	Glutaric acidemia IIA, 231680
ETFB	100	99,8	100	100	Glutaric acidemia IIB, 231680
ETFDH	100	99,8	100	100	Glutaric acidemia IIC, 231680
ETHE1	99,9	97,4	100	100	Ethylmalonic encephalopathy, 602473
EVC	93,9	88,6	96,9	94,8	Ellis-van Creveld syndrome, 225500 ?Weyers acrofacial dysostosis, 193530
EVC2	97,7	96,1	100	100	Ellis-van Creveld syndrome, 225500 Weyers acrofacial dysostosis, 193530
EXOC6B	99,1	97,6	100	100	Spondyloepimetaphyseal dysplasia with joint laxity, type 3, 618395
EXOSC2	100	100	100	100	Short stature, hearing loss, retinitis pigmentosa, and distinctive facies, 617763
EXOSC3	99,5	94,9	100	100	Pontocerebellar hypoplasia, type 1B, 614678
EXOSC8	97,9	91,2	100	100	Pontocerebellar hypoplasia, type 1C, 616081
EXOSC9	99,7	97,2	100	100	Pontocerebellar hypoplasia, type 1D, 618065
EXPH5	100	100	100	100	Epidermolysis bullosa, nonspecific, autosomal recessive, 615028
EXT2	100	99,3	100	100	Seizures, scoliosis, and macrocephaly syndrome, 616682 Exostoses, multiple, type 2, 133701
EXTL3	100	100	100	100	Immunoskeletal dysplasia with neurodevelopmental abnormalities, 617425
EYS	99,7	98,2	100	100	Retinitis pigmentosa 25, 602772
F10	99,8	99,1	100	100	Factor X deficiency, 227600
F11	100	100	100	100	Factor XI deficiency, autosomal dominant, 612416 Factor XI deficiency, autosomal recessive, 612416
F12	99,9	98,8	100	100	Factor XII deficiency, 234000 Angioedema, hereditary, type III, 610618
F13A1	100	100	100	100	{Myocardial infarction, protection against}, 608446 Factor XIIIA deficiency, 613225 {Venous thrombosis, protection against}, 188050
F13B	98,7	93,5	100	100	Factor XIIIB deficiency, 613235
F2	99,9	97,9	100	100	{Pregnancy loss, recurrent, susceptibility to, 2}, 614390 Hypoprothrombinemia, 613679 Dysprothrombinemia, 613679 Thrombophilia due to thrombin defect, 188050 {Stroke, ischemic, susceptibility to}, 601367
F5	99,4	98,5	100	100	{Pregnancy loss, recurrent, susceptibility to, 1}, 614389 Thrombophilia due to activated protein C resistance, 188055 Factor V deficiency, 227400 {Budd-Chiari syndrome}, 600880

					{Stroke, ischemic, susceptibility to}, 601367 {Thrombophilia, susceptibility to, due to factor V Leiden}, 188055
F7	100	100	100	100	Factor VII deficiency, 227500 {Myocardial infarction, decreased susceptibility to}, 608446
FA2H	92	83,1	100	100	Spastic paraplegia 35, autosomal recessive, 612319
FADD	100	100	100	100	Infections, recurrent, with encephalopathy, hepatic dysfunction, and cardiovascular malformations, 613759
FAH	100	100	100	100	Tyrosinemia, type I, 276700
FAM126A	100	99,4	100	100	Leukodystrophy, hypomyelinating, 5, 610532
FAM161A	100	99,7	100	100	Retinitis pigmentosa 28, 606068
FAM20A	99,6	94,7	100	100	Amelogenesis imperfecta, type IG (enamel-renal syndrome), 204690
FAM20C	100	100	100	99,8	Raine syndrome, 259775
FAN1	100	99,8	100	100	Interstitial nephritis, karyomegalic, 614817
FANCA	100	99,4	100	100	Fanconi anemia, complementation group A, 227650
FANCB	98,6	94,1	100	100	Fanconi anemia, complementation group B, 300514
FANCC	97,2	96,6	97,3	97,3	Fanconi anemia, complementation group C, 227645
FANCD2	99,5	97,5	98,8	98,8	Fanconi anemia, complementation group D2, 227646
FANCE	89,8	85,1	100	99,9	Fanconi anemia, complementation group E, 600901
FANCF	100	100	100	100	Fanconi anemia, complementation group F, 603467
FANCG	100	99,9	100	100	Fanconi anemia, complementation group G, 614082
FANCI	99,9	99,2	100	100	Fanconi anemia, complementation group I, 609053
FANCL	100	98,6	100	100	Fanconi anemia, complementation group L, 614083
FAR1	97,6	92,8	100	100	Peroxisomal fatty acyl-CoA reductase 1 disorder, 616154
FARS2	100	100	100	100	Spastic paraplegia 77, autosomal recessive, 617046 Combined oxidative phosphorylation deficiency 14, 614946
FARSB	98,8	94,6	100	100	Rajab interstitial lung disease with brain calcifications 1, 613658
FASTKD2	99,8	98,9	100	100	Combined oxidative phosphorylation deficiency 44, 618855
FAT4	100	100	100	100	Van Maldergem syndrome 2, 615546 Hennekam lymphangiectasia-lymphedema syndrome 2, 616006
FBLN5	91,8	91,8	91,8	91,8	Macular degeneration, age-related, 3, 608895 ?Cutis laxa, autosomal dominant 2, 614434 Neuropathy, hereditary, with or without age-related macular degeneration, 608895 Cutis laxa, autosomal recessive, type IA, 219100
FBP1	93,7	93,3	93,7	93,7	Fructose-1,6-bisphosphatase deficiency, 229700
FBXL3	100	100	100	100	Intellectual developmental disorder with short stature, facial anomalies, and speech defects, 606220
FBXL4	100	100	100	100	Mitochondrial DNA depletion syndrome 13 (encephalomyopathic type), 615471
FBXO31	96	93,1	100	99,9	?Mental retardation, autosomal recessive 45, 615979

FBXO7	99,8	97,9	100	100	Parkinson disease 15, autosomal recessive, 260300
FCGR3A	99	97,1	100	100	Immunodeficiency 20, 615707
FCN3	100	100	100	100	Immunodeficiency due to ficolin 3 deficiency, 613860
FCSK	97,7	95,4	100	100	Congenital disorder of glycosylation with defective fucosylation 2, 618324
FDFT1	97,7	96	100	100	Squalene synthase deficiency, 618156
FDX2	100	100	100	100	Mitochondrial myopathy, episodic, with optic atrophy and reversible leukoencephalopathy, 251900
FDXR	100	99,3	100	100	Auditory neuropathy and optic atrophy, 617717
FECH	100	100	100	100	Protoporphyria, erythropoietic, 1, 177000
FERMT1	99,9	97,9	100	100	Kindler syndrome, 173650
FERMT3	100	100	100	100	Leukocyte adhesion deficiency, type III, 612840
FEZF1	100	99,9	100	100	Hypogonadotropic hypogonadism 22, with or without anosmia, 616030
FGA	99,1	97,2	100	100	Dysfibrinogenemia, congenital, 616004 Amyloidosis, familial visceral, 105200 Hypodysfibrinogenemia, congenital, 616004 Afibrinogenemia, congenital, 202400
FGB	99,8	99,1	100	100	Dysfibrinogenemia, congenital, 616004 Afibrinogenemia, congenital, 202400 Hypofibrinogenemia, congenital, 202400
FGD4	99,9	99,4	100	100	Charcot-Marie-Tooth disease, type 4H, 609311
FGF20	97,5	87,6	100	100	?Renal hypodysplasia/aplasia 2, 615721
FGF23	99,6	97,5	100	100	Tumoral calcinosis, hyperphosphatemic, familial, 2, 617993 Hypophosphatemic rickets, autosomal dominant, 193100
FGF3	99,8	95,1	100	100	Deafness, congenital with inner ear agenesis, microtia, and microdontia, 610706
FGG	99,7	98,2	100	100	Hypodysfibrinogenemia, 616004 Hypofibrinogenemia, congenital, 202400 Dysfibrinogenemia, congenital, 616004 Afibrinogenemia, congenital, 202400
FH	92,1	88,3	100	100	Fumarase deficiency, 606812 Leiomyomatosis and renal cell cancer, 150800
FIBP	100	100	100	100	Thauvin-Robinet-Faivre syndrome, 617107
FIG4	100	99,8	100	100	Yunis-Varon syndrome, 216340 ?Polymicrogyria, bilateral temporooccipital, 612691 Charcot-Marie-Tooth disease, type 4J, 611228 Amyotrophic lateral sclerosis 11, 612577
FITM2	100	100	100	100	Siddiqi syndrome, 618635
FKBP10	98,8	97,2	100	100	Bruck syndrome 1, 259450 Osteogenesis imperfecta, type XI, 610968

FKBP14	100	99,9	100	100	Ehlers-Danlos syndrome, kyphoscoliotic type, 2, 614557
FKRP	100	100	100	99,9	Muscular dystrophy-dystroglycanopathy (limb-girdle), type C, 5, 607155 Muscular dystrophy-dystroglycanopathy (congenital with or without mental retardation), type B, 5, 606612 Muscular dystrophy-dystroglycanopathy (congenital with brain and eye anomalies), type A, 5, 613153
FKTN	99,7	97	100	100	Muscular dystrophy-dystroglycanopathy (limb-girdle), type C, 4, 611588 Cardiomyopathy, dilated, 1X, 611615 Muscular dystrophy-dystroglycanopathy (congenital without mental retardation), type B, 4, 613152 Muscular dystrophy-dystroglycanopathy (congenital with brain and eye anomalies), type A, 4, 253800
FLAD1	100	99,8	100	100	Lipid storage myopathy due to flavin adenine dinucleotide synthetase deficiency, 255100
FLG	100	99,9	100	100	{Dermatitis, atopic, susceptibility to, 2}, 605803 Ichthyosis vulgaris, 146700
FLNB	99,5	98,8	100	100	Larsen syndrome, 150250 Atelosteogenesis, type I, 108720 Boomerang dysplasia, 112310 Spondylocarpotarsal synostosis syndrome, 272460 Atelosteogenesis, type III, 108721
FLVCR1	100	98,9	100	100	Ataxia, posterior column, with retinitis pigmentosa, 609033
FLVCR2	100	100	100	100	Proliferative vasculopathy and hydranencephaly-hydrocephaly syndrome, 225790
FMN2	85,5	82,5	100	100	Mental retardation, autosomal recessive 47, 616193
FMO3	99,9	99,7	100	100	Trimethylaminuria, 602079
FOLR1	100	100	100	100	Neurodegeneration due to cerebral folate transport deficiency, 613068
FOXE1	96,9	78,5	99,9	99,1	Bamforth-Lazarus syndrome, 241850 {Thyroid cancer, nonmedullary, 4}, 616534
FOXE3	82,6	72	94,4	87,8	Cataract 34, multiple types, 612968 Anterior segment dysgenesis 2, multiple subtypes, 610256 {Aortic aneurysm, familial thoracic 11, susceptibility to}, 617349
FOXI1	100	100	100	100	Enlarged vestibular aqueduct, 600791
FOXN1	100	99,6	100	100	T-cell lymphopenia, infantile, with or without nail dystrophy, autosomal dominant, 618806 T-cell immunodeficiency, congenital alopecia, and nail dystrophy, 601705
FOXRED1	100	99,9	100	100	Mitochondrial complex I deficiency, nuclear type 19, 618241
FRAS1	100	99,4	100	100	Fraser syndrome 1, 219000
FREM1	99,9	99,1	100	100	Manitoba oculotrichoanal syndrome, 248450 Trigonocephaly 2, 614485 Bifid nose with or without anorectal and renal anomalies, 608980
FREM2	100	99,3	100	100	Fraser syndrome 2, 617666 Cryptophthalmos, unilateral or bilateral, isolated, 123570
FRMD4A	90,7	87,3	96,6	96,6	?Corpus callosum, agenesis of, with facial anomalies and cerebellar ataxia, 616819

FRRS1L	79,7	69,1	99,2	95,8	Developmental and epileptic encephalopathy 37, 616981
FSHB	100	100	100	100	Hypogonadotropic hypogonadism 24 without anosmia, 229070
FSHR	99,5	97,2	100	100	Ovarian hyperstimulation syndrome, 608115 Ovarian dysgenesis 1, 233300 Ovarian response to FSH stimulation, 276400
FTCD	95,7	91	100	100	Glutamate formiminotransferase deficiency, 229100
FTO	83,8	83,7	94,2	94,2	{Obesity, susceptibility to, BMIQ14}, 612460 Growth retardation, developmental delay, facial dysmorphism, 612938
FUCA1	100	99,9	100	100	Fucosidosis, 230000
FUT8	100	99,2	100	100	Congenital disorder of glycosylation with defective fucosylation 1, 618005
FXN	95,5	80,1	100	100	Friedreich ataxia with retained reflexes, 229300 Friedreich ataxia, 229300
FYCO1	100	99,9	100	100	Cataract 18, autosomal recessive, 610019
FZD6	100	100	100	100	Nail disorder, nonsyndromic congenital, 1, 161050
G6PC	100	100	100	100	Glycogen storage disease Ia, 232200
G6PC3	100	99,9	100	100	Dursun syndrome, 612541 Neutropenia, severe congenital 4, autosomal recessive, 612541
GAA	100	99,9	100	100	Glycogen storage disease II, 232300
GAB1	100	99,4	100	100	?Deafness, autosomal recessive 26, 605428
GAD1	100	99,9	100	100	?Cerebral palsy, spastic quadriplegic, 1, 603513 Developmental and epileptic encephalopathy 89, 619124
GALC	99,8	98,3	100	100	Krabbe disease, 245200
GALE	100	100	100	100	Galactose epimerase deficiency, 230350
GALK1	100	99,1	100	100	Galactokinase deficiency with cataracts, 230200
GALNS	100	99,8	100	100	Mucopolysaccharidosis IVA, 253000
GALNT2	99,6	97	100	100	Congenital disorder of glycosylation, type IIt, 618885
GALNT3	99,8	99	100	100	Tumoral calcinosis, hyperphosphatemic, familial, 1, 211900
GALT	100	99,7	100	100	Galactosemia, 230400
GAMT	93,1	82,7	100	100	Cerebral creatine deficiency syndrome 2, 612736
GAN	100	99,6	100	100	Giant axonal neuropathy-1, 256850
GAS2L2	100	100	100	100	?Ciliary dyskinesia, primary, 41, 618449
GAS8	99,9	99,3	100	100	Ciliary dyskinesia, primary, 33, 616726
GATM	100	100	100	100	Cerebral creatine deficiency syndrome 3, 612718 Fanconi renotubular syndrome 1, 134600
GBA	100	100	100	100	Gaucher disease, type III, 231000 {Parkinson disease, late-onset, susceptibility to}, 168600 Gaucher disease, type IIIC, 231005

					Gaucher disease, type I, 230800 Gaucher disease, perinatal lethal, 608013 Gaucher disease, type II, 230900 {Lewy body dementia, susceptibility to}, 127750
GBA2	100	99,7	100	100	Spastic paraplegia 46, autosomal recessive, 614409
GBE1	100	99,6	100	100	Polyglucosan body disease, adult form, 263570 Glycogen storage disease IV, 232500
GCDH	100	99,2	100	100	Glutaricaciduria, type I, 231670
GCH1	99,9	95,5	100	100	Hyperphenylalaninemia, BH4-deficient, B, 233910 Dystonia, DOPA-responsive, with or without hyperphenylalaninemia, 128230
GCK	95,4	95,4	92,5	92,2	Diabetes mellitus, noninsulin-dependent, late onset, 125853 MODY, type II, 125851 Diabetes mellitus, permanent neonatal 1, 606176 Hyperinsulinemic hypoglycemia, familial, 3, 602485
GCLC	99,8	98	100	100	Hemolytic anemia due to gamma-glutamylcysteine synthetase deficiency, 230450 {Myocardial infarction, susceptibility to}, 608446
GCNT2	99,5	99,5	100	100	Adult i phenotype without cataract, 110800 Cataract 13 with adult i phenotype, 116700 [Blood group, Ii], 110800
GCSH	75,7	68,9	100	100	?Glycine encephalopathy, 605899
GDAP1	99,8	99,3	100	100	Charcot-Marie-Tooth disease, recessive intermediate, A, 608340 Charcot-Marie-Tooth disease, type 4A, 214400 Charcot-Marie-Tooth disease, axonal, type 2K, 607831 Charcot-Marie-Tooth disease, axonal, with vocal cord paresis, 607706
GDAP2	100	99,2	100	100	Spinocerebellar ataxia, autosomal recessive 27, 618369
GDF1	73,9	54	98,7	92	Right atrial isomerism (Ivemark), 208530 Congenital heart defects, multiple types, 6, 613854
GDF5	100	100	100	100	?Acromesomelic dysplasia, Hunter-Thompson type, 201250 Symphalangism, proximal, 1B, 615298 Brachydactyly, type A1, C, 615072 Chondrodysplasia, Grebe type, 200700 Brachydactyly, type A2, 112600 Du Pan syndrome, 228900 {Osteoarthritis-5}, 612400 Brachydactyly, type C, 113100 Multiple synostoses syndrome 2, 610017
GEMIN4	100	99,5	100	100	Neurodevelopmental disorder with microcephaly, cataracts, and renal abnormalities, 617913
GFER	99,6	93,9	100	100	Myopathy, mitochondrial progressive, with congenital cataract and developmental delay, 613076

GFM1	99,9	99,4	100	100	Combined oxidative phosphorylation deficiency 1, 609060
GFM2	98,9	95,2	100	100	Combined oxidative phosphorylation deficiency 39, 618397
GFPT1	100	99,4	100	100	Myasthenia, congenital, 12, with tubular aggregates, 610542
GGCX	100	99,9	100	100	Vitamin K-dependent clotting factors, combined deficiency of, 1, 277450 Pseudoxanthoma elasticum-like disorder with multiple coagulation factor deficiency, 610842
GGT1	19,9	18,4	100	100	?Glutathioninuria, 231950
GH1	100	100	100	100	Kowarski syndrome, 262650 Growth hormone deficiency, isolated, type IA, 262400 Growth hormone deficiency, isolated, type IB, 612781 Growth hormone deficiency, isolated, type II, 173100
GHR	99,6	99,5	99,8	99,8	{Hypercholesterolemia, familial, modifier of}, 143890 Increased responsiveness to growth hormone, 604271 Laron dwarfism, 262500 Growth hormone insensitivity, partial, 604271
GHRHR	96,4	96,1	100	100	Growth hormone deficiency, isolated, type IV, 618157
GHSR	98,5	95,8	100	100	Growth hormone deficiency, isolated partial, 615925
GINS1	99,3	94,9	100	100	Immunodeficiency 55, 617827
GIPC3	24,8	23	99,9	99,6	Deafness, autosomal recessive 15, 601869
GJA1	100	100	100	100	Erythrokeratodermia variabilis et progressiva 3, 617525 Craniometaphyseal dysplasia, autosomal recessive, 218400 Atrioventricular septal defect 3, 600309 Oculodentodigital dysplasia, 164200 Syndactyly, type III, 186100 Oculodentodigital dysplasia, autosomal recessive, 257850 Hypoplastic left heart syndrome 1, 241550 Palmoplantar keratoderma with congenital alopecia, 104100
GJB2	100	100	100	100	Deafness, autosomal dominant 3A, 601544 Deafness, autosomal recessive 1A, 220290 Bart-Pumphrey syndrome, 149200 Vohwinkel syndrome, 124500 Keratoderma, palmoplantar, with deafness, 148350 Keratitis-ichthyosis-deafness syndrome, 148210 Hystrix-like ichthyosis with deafness, 602540
GJB6	100	100	100	100	Deafness, autosomal dominant 3B, 612643 Deafness, autosomal recessive 1B, 612645 Deafness, digenic GJB2/GJB6, 220290 Ectodermal dysplasia 2, Clouston type, 129500

GJC2	78,2	58,7	96,9	91,4	Spastic paraplegia 44, autosomal recessive, 613206 Lymphatic malformation 3, 613480 Leukodystrophy, hypomyelinating, 2, 608804
GLB1	99,9	97,4	100	100	GM1-gangliosidosis, type III, 230650 GM1-gangliosidosis, type I, 230500 Mucopolysaccharidosis type IVB (Morquio), 253010 GM1-gangliosidosis, type II, 230600
GLDC	89,9	82	100	99,9	Glycine encephalopathy, 605899
GLDN	94,6	91	100	100	Lethal congenital contracture syndrome 11, 617194
GLE1	100	100	100	100	Congenital arthrogryposis with anterior horn cell disease, 611890 Lethal congenital contracture syndrome 1, 253310
GLIS2	100	99,8	100	100	Nephronophthisis 7, 611498
GLIS3	98,6	98,2	100	100	Diabetes mellitus, neonatal, with congenital hypothyroidism, 610199
GLRA1	100	99,8	100	100	Hyperekplexia 1, 149400
GLRB	99,2	95,1	100	100	Hyperekplexia 2, 614619
GLRX5	97,3	89,1	99,6	95,4	Anemia, sideroblastic, 3, pyridoxine-refractory, 616860 Spasticity, childhood-onset, with hyperglycinemia, 616859
GLS	96,3	87,2	100	99,9	?Infantile cataract, skin abnormalities, glutamate excess, and impaired intellectual development, 618339 Global developmental delay, progressive ataxia, and elevated glutamine, 618412 Developmental and epileptic encephalopathy 71, 618328
GLUL	74,4	73,2	100	100	Glutamine deficiency, congenital, 610015
GLYCTK	98,8	97,3	100	100	D-glyceric aciduria, 220120
GM2A	100	100	100	100	GM2-gangliosidosis, AB variant, 272750
GMPPA	100	100	100	100	Alacrima, achalasia, and mental retardation syndrome, 615510
GMPPB	100	100	100	100	Muscular dystrophy-dystroglycanopathy (congenital with brain and eye anomalies), type A, 14, 615350 Muscular dystrophy-dystroglycanopathy (congenital with mental retardation), type B, 14, 615351 Muscular dystrophy-dystroglycanopathy (limb-girdle), type C, 14, 615352
GNAT2	99,9	99	100	100	Achromatopsia 4, 613856
GNB3	100	100	100	100	{Hypertension, essential, susceptibility to}, 145500 Night blindness, congenital stationary, type 1H, 617024
GNB5	100	98,8	100	100	Intellectual developmental disorder with cardiac arrhythmia, 617173 Language delay and ADHD/cognitive impairment with or without cardiac arrhythmia, 617182
GNE	100	99,7	100	100	Sialuria, 269921 Nonaka myopathy, 605820
GNMT	100	100	100	100	Glycine N-methyltransferase deficiency, 606664

GNPAT	99,7	97,3	100	100	Rhizomelic chondrodysplasia punctata, type 2, 222765
GNPTAB	100	99,9	100	100	Mucolipidosis II alpha/beta, 252500 Mucolipidosis III alpha/beta, 252600
GNPTG	99,1	94,3	100	99,9	Mucolipidosis III gamma, 252605
GNRHR	100	100	100	100	Hypogonadotropic hypogonadism 7 without anosmia, 146110
GNS	98,4	94,8	100	100	Mucopolysaccharidosis type IIID, 252940
GORAB	100	99,1	100	100	Geroderma osteodysplasticum, 231070
GOSR2	95,9	94,6	100	100	Epilepsy, progressive myoclonic 6, 614018
GP1BA	98,6	95,9	100	100	Bernard-Soulier syndrome, type A1 (recessive), 231200 {Nonarteritic anterior ischemic optic neuropathy, susceptibility to}, 258660 von Willebrand disease, platelet-type, 177820 Bernard-Soulier syndrome, type A2 (dominant), 153670
GP1BB	72,9	59,6	99,5	95	Giant platelet disorder, isolated, 231200 Bernard-Soulier syndrome, type B, 231200
GP6	100	100	94,9	91,7	Bleeding disorder, platelet-type, 11, 614201
GP9	96,5	89,3	100	100	Bernard-Soulier syndrome, type C, 231200
GPAA1	98,9	95,9	100	100	Glycosylphosphatidylinositol biosynthesis defect 15, 617810
GPC6	100	100	100	100	Omodysplasia 1, 258315
GPD1	100	100	100	100	Hypertriglyceridemia, transient infantile, 614480
GPHN	100	99,5	100	100	Molybdenum cofactor deficiency C, 615501
GPI	100	100	100	100	Hemolytic anemia, nonspherocytic, due to glucose phosphate isomerase deficiency, 613470
GPIHBP1	100	100	100	100	Hyperlipoproteinemia, type 1D, 615947
GPNMB	95,5	95,5	95,5	95,5	Amyloidosis, primary localized cutaneous, 3, 617920
GPR179	100	100	100	100	Night blindness, congenital stationary (complete), 1E, autosomal recessive, 614565
GPR88	99,4	95,1	98,8	94,9	?Chorea, childhood-onset, with psychomotor retardation, 616939
GPSM2	99,9	99,2	100	100	Chudley-McCullough syndrome, 604213
GPT2	99,2	93,6	100	99,8	Neurodevelopmental disorder with microcephaly and spastic paraplegia, 616281
GPX4	90,5	85,8	98,2	94,9	Spondylometaphyseal dysplasia, Sedaghatian type, 250220
GRAP	82,8	78,3	100	100	Deafness, autosomal recessive 114, 618456
GRHPR	84,2	81,3	100	99,3	Hyperoxaluria, primary, type II, 260000
GRID2	100	99,8	100	100	Spinocerebellar ataxia, autosomal recessive 18, 616204
GRIK2	96,2	95,4	96,3	96,3	Mental retardation, autosomal recessive, 6, 611092
GRIN1	100	100	100	100	Neurodevelopmental disorder with or without hyperkinetic movements and seizures, autosomal recessive, 617820 Neurodevelopmental disorder with or without hyperkinetic movements and seizures, autosomal dominant, 614254
GRIP1	100	99,7	100	100	Fraser syndrome 3, 617667

GRK1	100	100	100	100	Oguchi disease-2, 613411
GRM1	100	99,7	100	100	Spinocerebellar ataxia 44, 617691 Spinocerebellar ataxia, autosomal recessive 13, 614831
GRM6	90,2	80,6	98,3	96,3	Night blindness, congenital stationary (complete), 1B, autosomal recessive, 257270
GRN	100	100	100	100	Ceroid lipofuscinosis, neuronal, 11, 614706 Aphasia, primary progressive, 607485 Frontotemporal lobar degeneration with ubiquitin-positive inclusions, 607485
GRXCR1	100	99,8	100	100	Deafness, autosomal recessive 25, 613285
GRXCR2	100	100	100	100	?Deafness, autosomal recessive 101, 615837
GSC	99,2	92,4	100	100	Short stature, auditory canal atresia, mandibular hypoplasia, skeletal abnormalities, 602471
GSS	96,5	96,4	100	100	Glutathione synthetase deficiency, 266130 Hemolytic anemia due to glutathione synthetase deficiency, 231900
GSX2	100	100	100	100	Diencephalic-mesencephalic junction dysplasia syndrome 2, 618646
GTF2E2	100	99,8	100	100	Trichothiodystrophy 6, nonphotosensitive, 616943
GTF2H5	72,5	72,2	72,5	72,5	Trichothiodystrophy 3, photosensitive, 616395
GTPBP2	100	99,3	100	99,9	Jaberi-Elahi syndrome, 617988
GTPBP3	100	99,8	100	100	Combined oxidative phosphorylation deficiency 23, 616198
GUCY1A1	100	99,8	100	100	Moyamoya 6 with achalasia, 615750
GUCY2C	100	99,6	100	100	Diarrhea 6, 614616 Meconium ileus, 614665
GUCY2D	99,6	96,2	100	100	Cone-rod dystrophy 6, 601777 Leber congenital amaurosis 1, 204000 Night blindness, congenital stationary, type 1I, 618555 ?Choroidal dystrophy, central areolar 1, 215500
GUF1	99,7	97,8	100	100	?Developmental and epileptic encephalopathy 40, 617065
GUSB	92,9	91,7	100	100	Mucopolysaccharidosis VII, 253220
GYG1	99,9	99,2	100	100	?Glycogen storage disease XV, 613507 Polyglucosan body myopathy 2, 616199
GYS1	100	98,6	100	100	Glycogen storage disease 0, muscle, 611556
GYS2	99,8	99	100	100	Glycogen storage disease 0, liver, 240600
GZF1	100	99,6	100	100	Joint laxity, short stature, and myopia, 617662
H6PD	99	99	100	100	Cortisone reductase deficiency 1, 604931
HAAO	100	99,8	100	100	Vertebral, cardiac, renal, and limb defects syndrome 1, 617660
HACE1	100	99,3	100	100	Spastic paraplegia and psychomotor retardation with or without seizures, 616756
HADH	99	97,5	100	100	3-hydroxyacyl-CoA dehydrogenase deficiency, 231530 Hyperinsulinemic hypoglycemia, familial, 4, 609975

HADHA	97,2	91,6	100	100	LCHAD deficiency, 609016 HELLP syndrome, maternal, of pregnancy, 609016 Mitochondrial trifunctional protein deficiency, 609015 Fatty liver, acute, of pregnancy, 609016
HADHB	98,8	89,7	100	100	Trifunctional protein deficiency, 609015
HAMP	100	100	100	100	Hemochromatosis, type 2B, 613313
HARS1	100	100	100	100	Charcot-Marie-Tooth disease, axonal, type 2W, 616625 Usher syndrome type 3B, 614504
HARS2	100	100	100	100	Perrault syndrome 2, 614926
HAVCR2	100	100	100	100	T-cell lymphoma, subcutaneous panniculitis-like, 618398
HAX1	100	100	100	100	Neutropenia, severe congenital 3, autosomal recessive, 610738
HBB	100	100	100	100	Thalassemia, beta, 613985 Erythrocytosis 6, 617980 Heinz body anemia, 140700 Delta-beta thalassemia, 141749 Thalassemia-beta, dominant inclusion-body, 603902 Hereditary persistence of fetal hemoglobin, 141749 Methemoglobinemia, beta type, 617971 {Malaria, resistance to}, 611162 Sickle cell anemia, 603903
HELLS	97,8	92,1	100	100	Immunodeficiency-centromeric instability-facial anomalies syndrome 4, 616911
HEPACAM	86	78,9	100	100	Megalencephalic leukoencephalopathy with subcortical cysts 2A, 613925 Megalencephalic leukoencephalopathy with subcortical cysts 2B, remitting, with or without mental retardation, 613926
HEPHL1	100	99,9	100	100	?Abnormal hair, joint laxity, and developmental delay, 261990
HERC1	100	100	100	100	Macrocephaly, dysmorphic facies, and psychomotor retardation, 617011
HERC2	79,9	77,2	100	100	[Skin/hair/eye pigmentation 1, blue/nonblue eyes], 227220 Mental retardation, autosomal recessive 38, 615516 [Skin/hair/eye pigmentation 1, blond/brown hair], 227220
HES7	84,4	53,9	100	100	Spondylocostal dysostosis 4, autosomal recessive, 613686
HESX1	99,7	97,3	100	100	Pituitary hormone deficiency, combined, 5, 182230 Septooptic dysplasia, 182230 Growth hormone deficiency with pituitary anomalies, 182230
HEXA	93,8	93,3	100	100	GM2-gangliosidosis, several forms, 272800 Tay-Sachs disease, 272800 [Hex A pseudodeficiency], 272800
HEXB	99,6	96,9	100	99,9	Sandhoff disease, infantile, juvenile, and adult forms, 268800
HFM1	96,3	89,6	100	100	Premature ovarian failure 9, 615724

HGD	100	100	100	100	Alkaptonuria, 203500
HGF	100	99,4	100	100	Deafness, autosomal recessive 39, 608265
HGSNAT	86,4	86,3	91,2	89,3	Retinitis pigmentosa 73, 616544 Mucopolysaccharidosis type IIIC (Sanfilippo C), 252930
HIBCH	98,2	88,5	100	100	3-hydroxyisobutryl-CoA hydrolase deficiency, 250620
HIKESHI	98,2	90,4	100	100	Leukodystrophy, hypomyelinating, 13, 616881
HINT1	98,3	89,3	100	100	Neuromyotonia and axonal neuropathy, autosomal recessive, 137200
HJV	100	100	100	100	Hemochromatosis, type 2A, 602390
HK1	100	100	100	100	Hemolytic anemia due to hexokinase deficiency, 235700 Neuropathy, hereditary motor and sensory, Russe type, 605285 Neurodevelopmental disorder with visual defects and brain anomalies, 618547 Retinitis pigmentosa 79, 617460
HLCS	100	100	100	100	Holocarboxylase synthetase deficiency, 253270
HMGCL	100	99,8	100	100	HMG-CoA lyase deficiency, 246450
HMGCS2	100	99,6	100	100	HMG-CoA synthase-2 deficiency, 605911
HMOX1	98,4	89,9	100	100	{Pulmonary disease, chronic obstructive, susceptibility to}, 606963 Heme oxygenase-1 deficiency, 614034
HMX1	62,4	42,9	99,7	96,1	Oculoauricular syndrome, 612109
HNMT	100	99,8	100	100	{Asthma, susceptibility to}, 600807 Mental retardation, autosomal recessive 51, 616739
HOGA1	100	96,4	100	100	Hyperoxaluria, primary, type III, 613616
HOXA1	100	100	100	100	Athabaskan brainstem dysgenesis syndrome, 601536 Bosley-Salih-Alorainy syndrome, 601536
HOXA2	100	99,9	100	100	Microtia with or without hearing impairment (AD), 612290 ?Microtia, hearing impairment, and cleft palate (AR), 612290
HOXB1	100	100	100	100	Facial paresis, hereditary congenital, 3, 614744
HOXC13	100	99,9	100	100	Ectodermal dysplasia 9, hair/nail type, 614931
HPCA	100	100	100	100	Dystonia 2, torsion, autosomal recessive, 224500
HPD	100	100	100	100	Tyrosinemia, type III, 276710 Hawkinsinuria, 140350
HPDL	100	100	100	100	Spastic paraplegia 83, autosomal recessive, 619027 Neurodevelopmental disorder with progressive spasticity and brain white matter abnormalities, 619026
HPGD	100	98,9	100	100	Hypertrophic osteoarthropathy, primary, autosomal recessive 1, 259100 ?Digital clubbing, isolated congenital, 119900 Cranioosteoarthropathy, 259100
HPS1	100	100	100	100	Hermansky-Pudlak syndrome 1, 203300

HPS3	99,7	97,5	100	100	Hermansky-Pudlak syndrome 3, 614072
HPS4	100	100	100	100	Hermansky-Pudlak syndrome 4, 614073
HPS5	100	99,7	100	100	Hermansky-Pudlak syndrome 5, 614074
HPS6	97,1	88,9	100	100	Hermansky-Pudlak syndrome 6, 614075
HPSE2	100	99,9	100	100	Urofacial syndrome 1, 236730
HR	98,5	95,6	100	100	Hypotrichosis 4, 146550 Alopecia universalis, 203655 Atrichia with papular lesions, 209500
HSD11B2	86	82,7	99,9	98,1	Apparent mineralocorticoid excess, 218030
HSD17B3	97,8	97,8	97,8	97,8	Pseudohermaphroditism, male, with gynecomastia, 264300
HSD17B4	95,4	93,1	96,6	96,6	D-bifunctional protein deficiency, 261515 Perrault syndrome 1, 233400
HSD3B2	100	99,7	100	100	Adrenal hyperplasia, congenital, due to 3-beta-hydroxysteroid dehydrogenase 2 deficiency, 201810
HSD3B7	99,1	95,5	100	100	Bile acid synthesis defect, congenital, 1, 607765
HSPA9	88,5	84,5	100	100	Even-plus syndrome, 616854 Anemia, sideroblastic, 4, 182170
HSPD1	98,8	93,7	100	100	Leukodystrophy, hypomyelinating, 4, 612233 Spastic paraplegia 13, autosomal dominant, 605280
HSPG2	99,2	97,7	100	99,9	Dyssegmental dysplasia, Silverman-Handmaker type, 224410 Schwartz-Jampel syndrome, type 1, 255800
HTRA1	75,8	72,3	87,2	83,2	{Macular degeneration, age-related, neovascular type}, 610149 {Macular degeneration, age-related, 7}, 610149 Cerebral arteriopathy, autosomal dominant, with subcortical infarcts and leukoencephalopathy, type 2, 616779 CARASIL syndrome, 600142
HTRA2	100	99,9	100	100	{Parkinson disease 13}, 610297 3-methylglutaconic aciduria, type VIII, 617248
HYAL1	100	100	100	100	?Mucopolysaccharidosis type IX, 601492
HYDIN	99,9	99,3	100	100	Ciliary dyskinesia, primary, 5, 608647
HYLS1	100	100	100	100	Hydrolethalus syndrome, 236680
IARS1	100	99,6	100	100	Growth retardation, impaired intellectual development, hypotonia, and hepatopathy, 617093
IARS2	100	99,9	100	100	?Cataracts, growth hormone deficiency, sensory neuropathy, sensorineural hearing loss, and skeletal dysplasia, 616007
IBA57	93,7	90,1	100	100	?Spastic paraplegia 74, autosomal recessive, 616451 Multiple mitochondrial dysfunctions syndrome 3, 615330
ICOS	99,9	99,8	100	100	Immunodeficiency, common variable, 1, 607594
IDH3B	95,4	95,4	100	100	Retinitis pigmentosa 46, 612572

IDUA	93,7	86,8	100	100	Mucopolysaccharidosis Ih/s, 607015 Mucopolysaccharidosis Ih, 607014 Mucopolysaccharidosis Is, 607016
IER3IP1	91,9	82,6	100	100	Microcephaly, epilepsy, and diabetes syndrome, 614231
IFNAR2	100	99,7	100	100	?Immunodeficiency 45, 616669 {Hepatitis B virus, susceptibility to}, 610424
IFNGR1	98,2	97,5	100	100	Immunodeficiency 27A, mycobacteriosis, AR, 209950 {Tuberculosis infection, protection against}, 607948 {Tuberculosis, susceptibility to}, 607948 Immunodeficiency 27B, mycobacteriosis, AD, 615978 {H. pylori infection, susceptibility to}, 600263 {Hepatitis B virus infection, susceptibility to}, 610424
IFNGR2	93,3	93,2	100	99,8	Immunodeficiency 28, mycobacteriosis, 614889
IFT122	100	99,6	100	100	Cranioectodermal dysplasia 1, 218330
IFT140	99,8	98,8	100	100	Retinitis pigmentosa 80, 617781 Short-rib thoracic dysplasia 9 with or without polydactyly, 266920
IFT172	99,9	99,1	100	100	Retinitis pigmentosa 71, 616394 Short-rib thoracic dysplasia 10 with or without polydactyly, 615630
IFT27	100	100	100	100	?Bardet-Biedl syndrome 19, 615996
IFT43	100	100	100	100	?Cranioectodermal dysplasia 3, 614099 Short-rib thoracic dysplasia 18 with polydactyly, 617866 ?Retinitis pigmentosa 81, 617871
IFT52	100	99,9	100	100	Short-rib thoracic dysplasia 16 with or without polydactyly, 617102
IFT57	99,9	99,1	100	100	?Orofaciodigital syndrome XVIII, 617927
IFT74	98,4	93,9	100	100	?Bardet-Biedl syndrome 20, 617119
IFT80	97,6	88,2	100	100	Short-rib thoracic dysplasia 2 with or without polydactyly, 611263
IFT81	93,5	90,1	95	94,9	Short-rib thoracic dysplasia 19 with or without polydactyly, 617895
IGF1	100	99,9	100	100	Growth retardation with deafness and mental retardation due to IGF1 deficiency, 608747
IGF1R	100	99,9	100	100	Insulin-like growth factor I, resistance to, 270450
IGFALS	99,9	99,6	100	100	Acid-labile subunit, deficiency of, 615961
IGFBP7	92,7	87,2	100	100	Retinal arterial macroaneurysm with supravalvular pulmonic stenosis, 614224
IGHM	100	100	100	100	Agammaglobulinemia 1, 601495
IGHMBP2	98,8	95,1	100	100	Neuronopathy, distal hereditary motor, type VI, 604320 Charcot-Marie-Tooth disease, axonal, type 2S, 616155
IGKC	100	100	100	100	Kappa light chain deficiency, 614102
IGLL1	99,9	96,9	100	100	Agammaglobulinemia 2, 613500

IHH	100	100	100	100	Acrocapitofemoral dysplasia, 607778 Brachydactyly, type A1, 112500
IKBKB	99,8	97,4	100	100	Immunodeficiency 15A, 618204 Immunodeficiency 15B, 615592
IL10RA	100	100	100	100	Inflammatory bowel disease 28, early onset, autosomal recessive, 613148
IL10RB	99,8	98	100	100	{Hepatitis B virus, susceptibility to}, 610424 Inflammatory bowel disease 25, early onset, autosomal recessive, 612567
IL11RA	100	99,9	100	100	Craniosynostosis and dental anomalies, 614188
IL12B	100	99,3	100	100	Immunodeficiency 29, mycobacteriosis, 614890
IL12RB1	98,9	96,3	94,1	94,1	Immunodeficiency 30, 614891
IL17RA	100	99,4	100	100	Immunodeficiency 51, 613953
IL17RC	100	99,9	100	100	Candidiasis, familial, 9, 616445
IL1RN	100	100	100	100	{Gastric cancer risk after H. pylori infection}, 137215 {Microvascular complications of diabetes 4}, 612628 Interleukin 1 receptor antagonist deficiency, 612852
IL21R	100	100	100	100	[IgE, elevated level of], 147050 Immunodeficiency 56, 615207
IL2RA	100	99,7	100	100	Immunodeficiency 41 with lymphoproliferation and autoimmunity, 606367 {Diabetes, mellitus, insulin-dependent, susceptibility to, 10}, 601942
IL2RB	100	99,7	100	100	Immunodeficiency 63 with lymphoproliferation and autoimmunity, 618495
IL36RN	100	100	100	100	Psoriasis 14, pustular, 614204
IL6ST	96,4	90,3	100	100	Hyper-IgE recurrent infection syndrome 4, autosomal recessive, 618523
IL7R	100	99,8	100	100	Severe combined immunodeficiency, T-cell negative, B-cell/natural killer cell-positive type, 608971
ILDR1	99,9	98,4	100	100	Deafness, autosomal recessive 42, 609646
IMPA1	97	87	100	100	Mental retardation, autosomal recessive 59, 617323
IMPAD1	100	100	100	100	Chondrodysplasia with joint dislocations, GPAPP type, 614078
IMPG2	99,8	98,4	100	100	Macular dystrophy, vitelliform, 5, 616152 Retinitis pigmentosa 56, 613581
INPP5E	97,1	92,7	100	100	Mental retardation, truncal obesity, retinal dystrophy, and micropenis, 610156 Joubert syndrome 1, 213300
INPP5K	100	100	100	100	Muscular dystrophy, congenital, with cataracts and intellectual disability, 617404
INPPL1	98,4	94,5	99,9	99,7	Opsismodysplasia, 258480
INSR	97,8	94,7	99,9	99,2	Hyperinsulinemic hypoglycemia, familial, 5, 609968 Rabson-Mendenhall syndrome, 262190 Diabetes mellitus, insulin-resistant, with acanthosis nigricans, 610549 Leprechaunism, 246200
INTS1	99,8	98,5	100	100	Neurodevelopmental disorder with cataracts, poor growth, and dysmorphic facies, 618571

INTS8	99,9	98,8	100	100	?Neurodevelopmental disorder with cerebellar hypoplasia and spasticity, 618572
INTU	99,7	98,1	100	100	?Short-rib thoracic dysplasia 20 with polydactyly, 617925 ?Orofaciodigital syndrome XVII, 617926
INVS	100	100	100	100	Nephronophthisis 2, infantile, 602088
IQCB1	93,9	85	100	100	Senior-Loken syndrome 5, 609254
IQSEC1	89,1	86,3	97,6	95,2	Intellectual developmental disorder with short stature and behavioral abnormalities, 618687
IRAK4	99,8	97,7	100	100	Immunodeficiency 67, 607676
IREB2	100	99,8	100	100	Neurodegeneration, early-onset, with choreoathetoid movements and microcytic anemia, 618451
IRF7	100	99,9	100	100	?Immunodeficiency 39, 616345
IRF8	99	95,7	100	100	Immunodeficiency 32B, monocyte and dendritic cell deficiency, autosomal recessive, 226990 Immunodeficiency 32A, mycobacteriosis, autosomal dominant, 614893
IRF9	100	100	100	100	Immunodeficiency 65, susceptibility to viral infections, 618648
IRX5	99,9	98,2	100	99,8	Hamamy syndrome, 611174
ISCA1	94,2	85,9	95,1	95,1	Multiple mitochondrial dysfunctions syndrome 5, 617613
ISCA2	100	98,8	100	100	Multiple mitochondrial dysfunctions syndrome 4, 616370
ISCU	100	100	100	100	Myopathy with lactic acidosis, hereditary, 255125
ISG15	100	100	100	100	Immunodeficiency 38, 616126
ITCH	91,6	91,3	95,9	95	Autoimmune disease, multisystem, with facial dysmorphism, 613385
ITGA2B	99,7	97,8	100	100	Glanzmann thrombasthenia, 273800 Bleeding disorder, platelet-type, 16, autosomal dominant, 187800 Thrombocytopenia, neonatal alloimmune, BAK antigen related, 0
ITGA3	99,5	97,4	100	100	Interstitial lung disease, nephrotic syndrome, and epidermolysis bullosa, congenital, 614748
ITGA6	99,9	98,9	100	100	Epidermolysis bullosa, junctional, with pyloric stenosis, 226730
ITGA7	99,6	98	100	100	Muscular dystrophy, congenital, due to ITGA7 deficiency, 613204
ITGA8	99,9	99,7	100	100	Renal hypodysplasia/aplasia 1, 191830
ITGB2	97,2	97,2	97,2	97,2	Leukocyte adhesion deficiency, 116920
ITGB3	100	99,4	100	100	Bleeding disorder, platelet-type, 16, autosomal dominant, 187800 {Myocardial infarction, susceptibility to}, 608446 Glanzmann thrombasthenia, 273800 Purpura, posttransfusion, 0 Thrombocytopenia, neonatal alloimmune, 0
ITGB4	98,4	96,2	100	100	Epidermolysis bullosa of hands and feet, 131800 Epidermolysis bullosa, junctional, with pyloric atresia, 226730 Epidermolysis bullosa, junctional, non-Herlitz type, 226650
ITGB6	97,2	95,8	100	100	Amelogenesis imperfecta, type IH, 616221
ITK	100	98,9	100	100	Lymphoproliferative syndrome 1, 613011

ITPA	100	100	100	100	Developmental and epileptic encephalopathy 35, 616647 [Inosine triphosphatase deficiency], 613850
ITPR1	100	99,9	100	100	Spinocerebellar ataxia 29, congenital nonprogressive, 117360 Spinocerebellar ataxia 15, 606658 Gillespie syndrome, 206700
IVD	100	100	100	100	Isovaleric acidemia, 243500
IYD	99,5	95,7	100	100	Thyroid dyshormonogenesis 4, 274800
JAGN1	100	100	99,7	98	Neutropenia, severe congenital, 6, autosomal recessive, 616022
JAK3	99,9	98,7	100	100	SCID, autosomal recessive, T-negative/B-positive type, 600802
JAM2	100	99,9	92,3	92,3	Basal ganglia calcification, idiopathic, 8, autosomal recessive, 618824
JAM3	100	99,9	100	100	Hemorrhagic destruction of the brain, subependymal calcification, and cataracts, 613730
JPH1	100	99,9	100	100	?Charcot-Marie-Tooth disease, axonal, autosomal dominant, type 2K, 607831
JUP	100	99,5	100	100	Arrhythmogenic right ventricular dysplasia 12, 611528 Naxos disease, 601214
KALRN	99,9	99,6	100	100	No OMIM disease ID
KANK2	100	100	100	100	Palmoplantar keratoderma and woolly hair, 616099 Nephrotic syndrome, type 16, 617783
KARS1	100	99,9	100	100	Deafness, autosomal recessive 89, 613916 ?Charcot-Marie-Tooth disease, recessive intermediate, B, 613641 Leukoencephalopathy, progressive, infantile-onset, with or without deafness, 619147 Deafness, congenital, and adult-onset progressive leukoencephalopathy, 619196
KATNB1	100	99,9	100	100	Lissencephaly 6, with microcephaly, 616212
KCNE1	100	100	100	100	Long QT syndrome 5, 613695 Jervell and Lange-Nielsen syndrome 2, 612347
KCNJ1	100	100	100	100	Bartter syndrome, type 2, 241200
KCNJ10	89,3	89	100	100	Enlarged vestibular aqueduct, digenic, 600791 SESAME syndrome, 612780
KCNJ11	100	100	100	100	Maturity-onset diabetes of the young, type 13, 616329 {Diabetes mellitus, type 2, susceptibility to}, 125853 Diabetes, permanent neonatal 2, with or without neurologic features, 618856 Diabetes mellitus, transient neonatal 3, 610582 Hyperinsulinemic hypoglycemia, familial, 2, 601820
KCNJ13	100	100	100	100	Leber congenital amaurosis 16, 614186 Snowflake vitreoretinal degeneration, 193230
KCNMA1	94,4	93,6	100	100	Liang-Wang syndrome, 618729 {Epilepsy, idiopathic generalized, susceptibility to, 16}, 618596 Cerebellar atrophy, developmental delay, and seizures, 617643 Paroxysmal nonkinesigenic dyskinesia, 3, with or without generalized epilepsy, 609446

KCNQ1	93,3	90,6	100	99,8	Long QT syndrome 1, 192500 Jervell and Lange-Nielsen syndrome, 220400 Short QT syndrome 2, 609621 {Long QT syndrome 1, acquired, susceptibility to}, 192500 Atrial fibrillation, familial, 3, 607554
KCNV2	100	99,9	100	100	Retinal cone dystrophy 3B, 610356
KCTD7	95	95	100	100	Epilepsy, progressive myoclonic 3, with or without intracellular inclusions, 611726
KDM5B	94,6	92,3	93,9	92,9	Mental retardation, autosomal recessive 65, 618109
KERA	100	100	100	100	Cornea plana 2, autosomal recessive, 217300
KHDC3L	100	99,8	100	100	Hydatidiform mole, recurrent, 2, 614293
KIAA0556	100	99,9	100	100	Joubert syndrome 26, 616784
KIAA0586	97,3	93,1	95,8	95,8	Joubert syndrome 23, 616490 Short-rib thoracic dysplasia 14 with polydactyly, 616546
KIAA0753	100	99,3	100	100	?Orofaciodigital syndrome XV, 617127
KIAA1109	99,8	99,2	100	100	Alkuraya-Kucinskas syndrome, 617822
KIAA1549	97,9	96,4	98,8	98	Retinitis pigmentosa 86, 618613
KIF14	99,6	97,7	100	100	Microcephaly 20, primary, autosomal recessive, 617914 ?Meckel syndrome 12, 616258
KIF1A	97,4	95,2	98	98	NESCAV syndrome, 614255 Spastic paraplegia 30, autosomal dominant, 610357 Neuropathy, hereditary sensory, type IIC, 614213 Spastic paraplegia 30, autosomal recessive, 610357
KIF1C	100	100	100	100	Spastic ataxia 2, autosomal recessive, 611302
KIF7	93,6	90,6	99,1	97,8	?Hydrolethalus syndrome 2, 614120 Acrocallosal syndrome, 200990 Joubert syndrome 12, 200990 ?Al-Gazali-Bakalinova syndrome, 607131
KIFBP	96,1	96,1	96,1	96,1	Goldberg-Shprintzen megacolon syndrome, 609460
KISS1R	100	99,5	100	100	Hypogonadotropic hypogonadism 8 with or without anosmia, 614837 ?Precocious puberty, central, 1, 176400
KIZ	100	99,2	100	100	Retinitis pigmentosa 69, 615780
KL	98,2	97,2	98,5	97,5	?Tumoral calcinosis, hyperphosphatemic, familial, 3, 617994
KLC2	99,2	97,9	100	100	Spastic paraplegia, optic atrophy, and neuropathy, 609541
KLHL3	100	99,3	100	100	Pseudohypoaldosteronism, type IID, 614495
KLHL40	100	100	100	100	Nemaline myopathy 8, autosomal recessive, 615348
KLHL41	100	99,9	100	100	Nemaline myopathy 9, 615731

KLHL7	99,9	99,8	100	100	Retinitis pigmentosa 42, 612943 PERCHING syndrome, 617055
KLK4	100	100	100	100	Amelogenesis imperfecta, type IIA1, 204700
KLKB1	100	99,5	100	100	Fletcher factor (prekallikrein) deficiency, 612423
KMT2B	95,8	94	98,7	97,9	Dystonia 28, childhood-onset, 617284
KNL1	99,2	98,1	98,9	98,8	Microcephaly 4, primary, autosomal recessive, 604321
KPTN	100	100	100	100	Mental retardation, autosomal recessive 41, 615637
KRT10	100	99,3	100	100	Ichthyosis with confetti, 609165 Epidermolytic hyperkeratosis, 113800 Ichthyosis, cyclic, with epidermolytic hyperkeratosis, 607602
KRT14	89	81,9	100	100	Naegeli-Franceschetti-Jadassohn syndrome, 161000 Epidermolysis bullosa simplex, Dowling-Meara type, 131760 Dermatopathia pigmentosa reticularis, 125595 Epidermolysis bullosa simplex, Koebner type, 131900 Epidermolysis bullosa simplex, recessive 1, 601001 Epidermolysis bullosa simplex, Weber-Cockayne type, 131800
KRT18	86,7	70,9	100	100	{Cirrhosis, noncryptogenic, susceptibility to}, 215600 Cirrhosis, cryptogenic, 215600
KRT5	100	100	100	100	Epidermolysis bullosa simplex, Weber-Cockayne type, 131800 Epidermolysis bullosa simplex-MCR, 609352 Epidermolysis bullosa simplex-MP, 131960 Dowling-Degos disease 1, 179850 Epidermolysis bullosa simplex, Koebner type, 131900 Epidermolysis bullosa simplex, Dowling-Meara type, 131760 Epidermolysis bullosa simplex, recessive 1, 601001
KRT8	90,6	69,6	100	100	{Cirrhosis, noncryptogenic, susceptibility to}, 215600 Cirrhosis, cryptogenic, 215600
KRT85	99	93,6	100	100	Ectodermal dysplasia 4, hair/nail type, 602032
KY	100	99,7	100	100	Myopathy, myofibrillar, 7, 617114
KYNU	99,6	97,1	100	100	Vertebral, cardiac, renal, and limb defects syndrome 2, 617661 ?Hydroxykynureninuria, 236800
L2HGDH	99	97,2	100	100	L-2-hydroxyglutaric aciduria, 236792
LAMA1	100	99,7	100	100	Poretti-Boltshauser syndrome, 615960
LAMA2	100	99,6	100	100	Muscular dystrophy, limb-girdle, autosomal recessive 23, 618138 Muscular dystrophy, congenital, merosin deficient or partially deficient, 607855
LAMA3	100	99,7	100	100	Epidermolysis bullosa, junctional, Herlitz type, 226700 Laryngoonychocutaneous syndrome, 245660 Epidermolysis bullosa, generalized atrophic benign, 226650

LAMB1	100	99,9	100	100	Lissencephaly 5, 615191
LAMB2	100	99,7	100	100	Pierson syndrome, 609049 Nephrotic syndrome, type 5, with or without ocular abnormalities, 614199
LAMB3	100	99,6	100	100	Amelogenesis imperfecta, type IA, 104530 Epidermolysis bullosa, junctional, Herlitz type, 226700 Epidermolysis bullosa, junctional, non-Herlitz type, 226650
LAMC2	99,8	98	100	100	Epidermolysis bullosa, junctional, non-Herlitz type, 226650 Epidermolysis bullosa, junctional, Herlitz type, 226700
LAMC3	98,6	97,1	100	99,6	Cortical malformations, occipital, 614115
LAMTOR2	100	99,7	100	100	:Immunodeficiency due to defect in MAPBP-interacting protein, 610798
LARGE1	100	99,6	100	100	Muscular dystrophy-dystroglycanopathy (congenital with mental retardation), type B, 6, 608840 Muscular dystrophy-dystroglycanopathy (congenital with brain and eye anomalies), type A, 6, 613154
LARP7	88,5	78,4	100	100	Alazami syndrome, 615071
LARS1	99,8	98,4	100	100	?Infantile liver failure syndrome 1, 615438
LARS2	100	100	100	100	Perrault syndrome 4, 615300 Hydrops, lactic acidosis, and sideroblastic anemia, 617021
LAT	100	99,2	100	100	Immunodeficiency 52, 617514
LBR	99,4	94,5	100	100	Pelger-Huet anomaly, 169400 Greenberg skeletal dysplasia, 215140 ?Reynolds syndrome, 613471 Pelger-Huet anomaly with mild skeletal anomalies, 618019
LCA5	99,9	99,2	100	100	Leber congenital amaurosis 5, 604537
LCAT	99	93,8	100	100	Norum disease, 245900 Fish-eye disease, 136120
LCK	98,9	96,6	100	100	?Immunodeficiency 22, 615758
LCT	99,8	98,5	100	100	Lactase deficiency, congenital, 223000
LDHA	95	91,7	100	100	Glycogen storage disease XI, 612933
LDHD	100	99,5	100	100	D-lactic aciduria with susceptibility to gout, 245450
LDLRAP1	98,8	94,2	100	100	Hypercholesterolemia, familial, 4, 603813
LEMD2	98,7	92	100	100	Cataract 46, juvenile-onset, 212500
LEP	99,9	97,3	100	100	Obesity, morbid, due to leptin deficiency, 614962
LEPR	94,3	92,6	94,6	94,6	Obesity, morbid, due to leptin receptor deficiency, 614963
LFNG	87,9	86,4	92,2	87,7	Spondylocostal dysostosis 3, autosomal recessive, 609813
LGI4	99,9	97,9	100	100	Arthrogryposis multiplex congenita 1, neurogenic, with myelin defect, 617468
LHB	90,4	38,9	100	100	Hypogonadotropic hypogonadism 23 with or without anosmia, 228300
LHCGR	94,1	92,3	100	100	Leydig cell adenoma, somatic, with precocious puberty, 176410 Precocious puberty, male, 176410

					Luteinizing hormone resistance, female, 238320 Leydig cell hypoplasia with pseudohermaphroditism, 238320 Leydig cell hypoplasia with hypergonadotropic hypogonadism, 238320
LHFPL5	100	100	100	100	Deafness, autosomal recessive 67, 610265
LHX3	96,6	96,5	100	100	Pituitary hormone deficiency, combined, 3, 221750
LIAS	100	99,1	100	100	Hyperglycinemia, lactic acidosis, and seizures, 614462
LIFR	99,7	98	100	100	Stuve-Wiedemann syndrome/Schwartz-Jampel type 2 syndrome, 601559
LIG4	100	99,9	100	100	{Multiple myeloma, resistance to}, 254500 LIG4 syndrome, 606593
LIM2	100	100	100	100	Cataract 19, multiple types, 615277
LIMS2	93	92,7	99,8	98,9	?Muscular dystrophy, autosomal recessive, with cardiomyopathy and triangular tongue, 616827
LINGO1	100	100	100	100	Mental retardation, autosomal recessive 64, 618103
LINS1	100	99,1	100	100	Mental retardation, autosomal recessive 27, 614340
LIPA	99,2	95,2	95,2	95,2	Wolman disease, 278000 Cholesteryl ester storage disease, 278000
LIPC	100	99,4	100	100	[High density lipoprotein cholesterol level QTL 12], 612797 Hepatic lipase deficiency, 614025 {Diabetes mellitus, noninsulin-dependent}, 125853
LIPE	100	99	100	100	Lipodystrophy, familial partial, type 6, 615980
LIPH	100	99,8	100	100	Woolly hair, autosomal recessive 2 with or without hypotrichosis, 604379 Hypotrichosis 7, 604379
LIPN	100	98,9	100	100	Ichthyosis, congenital, autosomal recessive 8, 613943
LIPT1	100	99,9	100	100	Lipoyltransferase 1 deficiency, 616299
LIPT2	94,9	75,2	100	100	Encephalopathy, neonatal severe, with lactic acidosis and brain abnormalities, 617668
LMAN1	99,8	99,2	100	100	Combined factor V and VIII deficiency, 227300
LMAN2L	100	99,7	100	100	?Mental retardation, autosomal recessive, 52, 616887
LMBR1	98,1	96,2	98,7	98,7	Triphalangeal thumb-polysyndactyly syndrome, 174500 Syndactyly, type IV, 186200 Triphalangeal thumb, type I, 174500 Acheiropody, 200500 Laurin-Sandrow syndrome, 135750 Hypoplastic or aplastic tibia with polydactyly, 188740 Polydactyly, preaxial type II, 174500
LMBRD1	94,7	90,2	96,1	96,1	Methylmalonic aciduria and homocystinuria, cblF type, 277380
LMF1	100	99,6	100	100	Lipase deficiency, combined, 246650
LMNA	97,4	91,9	100	100	Muscular dystrophy, congenital, 613205 Lipodystrophy, familial partial, type 2, 151660

					Charcot-Marie-Tooth disease, type 2B1, 605588 Cardiomyopathy, dilated, 1A, 115200 Heart-hand syndrome, Slovenian type, 610140 Hutchinson-Gilford progeria, 176670 Restrictive dermopathy, lethal, 275210 Mandibuloacral dysplasia, 248370 Emery-Dreifuss muscular dystrophy 2, autosomal dominant, 181350 Emery-Dreifuss muscular dystrophy 3, autosomal recessive, 616516 Malouf syndrome, 212112
LMNB2	97,5	94,3	98,2	96,9	{Lipodystrophy, partial, acquired, susceptibility to}, 608709 Microcephaly 27, primary, autosomal dominant, 619180 ?Epilepsy, progressive myoclonic, 9, 616540
LMOD3	100	99,7	100	100	Nemaline myopathy 10, 616165
LNPK	98,4	92,8	93,3	93,3	Neurodevelopmental disorder with epilepsy and hypoplasia of the corpus callosum, 618090
LONP1	100	99,8	100	100	CODAS syndrome, 600373
LOXHD1	100	99,7	100	100	Deafness, autosomal recessive 77, 613079
LPAR6	99,6	97,8	100	100	Hypotrichosis 8, 278150 Woolly hair, autosomal recessive 1, with or without hypotrichosis, 278150
LPIN1	99,6	97,3	100	100	Myoglobinuria, acute recurrent, autosomal recessive, 268200
LPIN2	100	100	100	100	Majeed syndrome, 609628
LPL	100	100	100	100	Lipoprotein lipase deficiency, 238600 [High density lipoprotein cholesterol level QTL 11], 238600 Combined hyperlipidemia, familial, 144250
LRAT	100	100	100	100	Retinal dystrophy, early-onset severe, 613341 Leber congenital amaurosis 14, 613341 Retinitis pigmentosa, juvenile, 613341
LRBA	99,9	99,6	100	100	Immunodeficiency, common variable, 8, with autoimmunity, 614700
LRIG2	99,6	98,8	100	100	Urofacial syndrome 2, 615112
LRIT3	93,9	91,9	100	100	Night blindness, congenital stationary (complete), 1F, autosomal recessive, 615058
LRMDA	96,8	95,6	99,6	99,6	Albinism, oculocutaneous, type VII, 615179
LRP1	99,7	98,9	100	100	?Keratosis pilaris atrophicans, 604093
LRP2	100	99,9	100	100	Donnai-Barrow syndrome, 222448
LRP4	99,1	98,8	100	100	?Myasthenic syndrome, congenital, 17, 616304 Sclerosteosis 2, 614305 Cenani-Lenz syndactyly syndrome, 212780
LRP5	98,5	98,1	100	99,7	van Buchem disease, type 2, 607636 Exudative vitreoretinopathy 4, 601813 Hyperostosis, endosteal, 144750

					Osteosclerosis, 144750 Polycystic liver disease 4 with or without kidney cysts, 617875 Osteoporosis-pseudoglioma syndrome, 259770 Osteopetrosis, autosomal dominant 1, 607634 {Osteoporosis}, 166710 [Bone mineral density variability 1], 601884
LRPAP1	100	100	100	100	Myopia 23, autosomal recessive, 615431
LRPPRC	99,9	99,1	100	100	Mitochondrial complex IV deficiency, nuclear type 5, (French-Canadian), 220111
LRRC56	100	99	100	100	Ciliary dyskinesia, primary, 39, 618254
LRRC6	99,2	96,3	100	100	Ciliary dyskinesia, primary, 19, 614935
LRSAM1	100	99,9	100	100	Charcot-Marie-Tooth disease, axonal, type 2P, 614436
LRTOMT	100	99,2	100	100	Deafness, autosomal recessive 63, 611451
LSS	100	99,9	100	100	Alopecia-mental retardation syndrome 4, 618840 Cataract 44, 616509 Hypotrichosis 14, 618275
LTBP2	99,9	99	100	100	Glaucoma 3, primary congenital, D, 613086 ?Weill-Marchesani syndrome 3, recessive, 614819 Microspherophakia and/or megalocornea, with ectopia lentis and with or without secondary glaucoma, 251750
LTBP3	99,6	98,1	100	100	Dental anomalies and short stature, 601216 Geleophysic dysplasia 3, 617809
LTBP4	99,9	97,5	100	100	Cutis laxa, autosomal recessive, type IC, 613177
LTC4S	74,2	68,5	100	100	Leukotriene C4 synthase deficiency, 614037
LYRM4	68,5	66,2	66,3	66,3	?Combined oxidative phosphorylation deficiency 19, 615595
LYRM7	95,9	86,2	100	100	Mitochondrial complex III deficiency, nuclear type 8, 615838
LYST	99,6	98,3	100	100	Chediak-Higashi syndrome, 214500
LZTFL1	99,9	99,2	100	100	Bardet-Biedl syndrome 17, 615994
LZTR1	100	99,9	100	100	{Schwannomatosis-2, susceptibility to}, 615670 Noonan syndrome 2, 605275 Noonan syndrome 10, 616564
MAB21L1	100	100	100	100	Cerebellar, ocular, craniofacial, and genital syndrome, 618479
MAB21L2	100	100	100	100	Microphthalmia/coloboma and skeletal dysplasia syndrome, 615877
MAD2L2	100	99,9	100	100	?Fanconi anemia, complementation group V, 617243
MAG	100	100	100	100	Spastic paraplegia 75, autosomal recessive, 616680
MAGI2	94,5	92,4	94,7	93,3	Nephrotic syndrome, type 15, 617609
MAK	98,7	96,8	100	100	Retinitis pigmentosa 62, 614181
MALT1	91,2	89,4	100	100	Immunodeficiency 12, 615468

MAN1B1	100	99,7	100	99,9	Rafiq syndrome, 614202
MAN2B1	99,8	97,9	100	100	Mannosidosis, alpha-, types I and II, 248500
MANBA	87,8	86,5	100	100	Mannosidosis, beta, 248510
MAP11	100	99,4	100	100	?Microcephaly 25, primary, autosomal recessive, 618351
MAP3K20	100	99,5	100	100	Split-foot malformation with mesoaxial polydactyly, 616890 Centronuclear myopathy 6 with fiber-type disproportion, 617760
MAPKBP1	100	100	100	100	Nephronophthisis 20, 617271
MAPT	100	99,5	100	100	Pick disease, 172700 Dementia, frontotemporal, with or without parkinsonism, 600274 {Parkinson disease, susceptibility to}, 168600 Supranuclear palsy, progressive, 601104 Supranuclear palsy, progressive atypical, 260540
MARS1	99,7	97,4	100	100	Charcot-Marie-Tooth disease, axonal, type 2U, 616280 Interstitial lung and liver disease, 615486
MARS2	100	100	100	100	Spastic ataxia 3, autosomal recessive, 611390 ?Combined oxidative phosphorylation deficiency 25, 616430
MARVELD2	99,2	96,1	100	100	Deafness, autosomal recessive 49, 610153
MASP1	100	99,9	100	100	3MC syndrome 1, 257920
MAT1A	99,7	97,7	100	100	Methionine adenosyltransferase deficiency, autosomal recessive, 250850 Hypermethioninemia, persistent, autosomal dominant, due to methionine adenosyltransferase I/III deficiency, 250850
MATN3	84,7	84,6	100	100	{Osteoarthritis susceptibility 2}, 140600 ?Spondyloepimetaphyseal dysplasia, Borochowitz Cormier-Daire type, 608728 Epiphyseal dysplasia, multiple, 5, 607078
MBOAT7	100	99,5	100	100	Mental retardation, autosomal recessive 57, 617188
MBTPS1	99,6	98,4	100	100	?Spondyloepiphyseal dysplasia, Kondo-Fu type, 618392
MC2R	99,9	98,3	100	100	Glucocorticoid deficiency, due to ACTH unresponsiveness, 202200
MCCC1	100	99,8	100	100	3-Methylcrotonyl-CoA carboxylase 1 deficiency, 210200
MCCC2	99,9	98,4	100	100	3-Methylcrotonyl-CoA carboxylase 2 deficiency, 210210
MCEE	100	100	100	100	Methylmalonyl-CoA epimerase deficiency, 251120
MCFD2	99,5	96,9	100	100	Factor V and factor VIII, combined deficiency of, 613625
MCM3AP	99,9	99,1	100	100	Peripheral neuropathy, autosomal recessive, with or without impaired intellectual development, 618124
MCM4	95,5	95	95,5	95,5	Immunodeficiency 54, 609981
MCM5	100	99,7	100	100	?Meier-Gorlin syndrome 8, 617564
MCM9	99,9	99,8	100	100	Ovarian dysgenesis 4, 616185
MCOLN1	99,8	98,4	100	100	Mucolipidosis IV, 252650

MCPH1	100	99,4	100	100	Microcephaly 1, primary, autosomal recessive, 251200
MDH2	98	97,9	100	100	Developmental and epileptic encephalopathy 51, 617339
MECR	100	98,9	100	100	Dystonia, childhood-onset, with optic atrophy and basal ganglia abnormalities, 617282
MED17	96,3	93,5	100	100	Microcephaly, postnatal progressive, with seizures and brain atrophy, 613668
MED23	100	99,7	100	100	Mental retardation, autosomal recessive 18, 614249
MED25	100	99,8	100	100	Basel-Vanagait-Smirin-Yosef syndrome, 616449
MEFV	99,9	98,6	96,4	96,4	Neutrophilic dermatosis, acute febrile, 608068 Familial Mediterranean fever, AR, 249100 Familial Mediterranean fever, AD, 134610
MEGF10	100	100	100	100	Myopathy, areflexia, respiratory distress, and dysphagia, early-onset, mild variant, 614399 Myopathy, areflexia, respiratory distress, and dysphagia, early-onset, 614399
MEGF8	99,9	99	100	100	Carpenter syndrome 2, 614976
MEOX1	100	98,9	100	100	Klippel-Feil syndrome 2, 214300
MERTK	99,5	98,8	99,1	99,1	Retinitis pigmentosa 38, 613862
MESD	100	99,9	100	100	Osteogenesis imperfecta, type XX, 618644
MESP2	93,9	86,9	97,5	97,5	Spondylocostal dysostosis 2, autosomal recessive, 608681
MET	100	99,5	100	100	{Osteofibrous dysplasia, susceptibility to}, 607278 Hepatocellular carcinoma, childhood type, somatic, 114550 ?Deafness, autosomal recessive 97, 616705 Renal cell carcinoma, papillary, 1, familial and somatic, 605074
METTL23	100	100	100	100	Mental retardation, autosomal recessive 44, 615942
MFF	94,3	89,9	100	100	Encephalopathy due to defective mitochondrial and peroxisomal fission 2, 617086
MFN2	100	99,9	100	100	Hereditary motor and sensory neuropathy VIA, 601152 Charcot-Marie-Tooth disease, axonal, type 2A2B, 617087 Charcot-Marie-Tooth disease, axonal, type 2A2A, 609260
MFRP	100	100	100	100	Nanophthalmos 2, 609549 Microphthalmia, isolated 5, 611040
MFSD2A	99,7	98,5	100	100	Neurodevelopmental disorder with progressive microcephaly, spasticity, and brain abnormalities, 616486
MFSD8	100	99,7	100	100	Macular dystrophy with central cone involvement, 616170 Ceroid lipofuscinosis, neuronal, 7, 610951
MGAT2	100	100	100	100	Congenital disorder of glycosylation, type IIa, 212066
MGME1	100	100	100	100	Mitochondrial DNA depletion syndrome 11, 615084
MGP	98,7	95,1	100	100	Keutel syndrome, 245150
MICOS13	100	99,7	100	99,7	Combined oxidative phosphorylation deficiency 37, 618329
MICU1	98,9	95,2	100	100	Myopathy with extrapyramidal signs, 615673
MIPEP	99,2	96,5	100	100	Combined oxidative phosphorylation deficiency 31, 617228

MITF	100	99,9	100	100	COMMAD syndrome, 617306 {Melanoma, cutaneous malignant, susceptibility to, 8}, 614456 Waardenburg syndrome, type 2A, 193510 Waardenburg syndrome/ocular albinism, digenic, 103470 Tietz albinism-deafness syndrome, 103500
MKKS	100	100	100	100	Bardet-Biedl syndrome 6, 605231 McKusick-Kaufman syndrome, 236700
MKS1	99,8	97,9	100	100	Bardet-Biedl syndrome 13, 615990 Joubert syndrome 28, 617121 Meckel syndrome 1, 249000
MLC1	100	99	100	100	Megalencephalic leukoencephalopathy with subcortical cysts, 604004
MLH1	100	99,9	100	100	Colorectal cancer, hereditary nonpolyposis, type 2, 609310 Muir-Torre syndrome, 158320 Mismatch repair cancer syndrome 1, 276300
MLPH	100	98,8	100	100	Griscelli syndrome, type 3, 609227
MLYCD	96	90,4	100	98,9	Malonyl-CoA decarboxylase deficiency, 248360
MMAA	100	100	100	100	Methylmalonic aciduria, vitamin B12-responsive, cblA type, 251100
MMAB	100	99,6	100	100	Methylmalonic aciduria, vitamin B12-responsive, cblB type, 251110
MMACHC	100	100	100	100	Methylmalonic aciduria and homocystinuria, cblC type, 277400
MMADHC	94,4	83,5	89,7	89,7	Homocystinuria, cblD type, variant 1, 277410 Methylmalonic aciduria and homocystinuria, cblD type, 277410 Methylmalonic aciduria, cblD type, variant 2, 277410
MME	99,8	98,7	98	98	Charcot-Marie-Tooth disease, axonal, type 2T, 617017 ?Spinocerebellar ataxia 43, 617018
MMP13	95,2	92,2	92,4	92,4	Metaphyseal dysplasia, Spahr type, 250400 Metaphyseal anadysplasia 1, 602111 ?Spondyloepimetaphyseal dysplasia, Missouri type, 602111
MMP14	100	98,9	100	100	?Winchester syndrome, 277950
MMP2	100	100	100	100	Multicentric osteolysis, nodulosis, and arthropathy, 259600
MMP20	100	100	100	100	Amelogenesis imperfecta, type IIA2, 612529
MMP21	99,9	98,8	100	100	Heterotaxy, visceral, 7, autosomal, 616749
MMP9	99,1	96,1	100	100	Metaphyseal anadysplasia 2, 613073
MMUT	99,8	98,3	100	100	Methylmalonic aciduria, mut(0) type, 251000
MOCOS	99,8	97,7	100	100	Xanthinuria, type II, 603592
MOCS1	99,2	95,1	100	100	Molybdenum cofactor deficiency A, 252150
MOCS2	99,6	99,5	100	100	Molybdenum cofactor deficiency B, 252160
MOGS	100	99,9	100	100	Congenital disorder of glycosylation, type IIb, 606056

MPC1	100	99,6	100	100	Mitochondrial pyruvate carrier deficiency, 614741
MPDU1	100	100	100	100	Congenital disorder of glycosylation, type If, 609180
MPDZ	99,8	98,8	100	100	Hydrocephalus, congenital, 2, with or without brain or eye anomalies, 615219
MPI	100	99,9	100	100	Congenital disorder of glycosylation, type Ib, 602579
MPL	100	99,5	100	100	Myelofibrosis with myeloid metaplasia, somatic, 254450 Thrombocytopenia, congenital amegakaryocytic, 604498 Thrombocythemia 2, 601977
MPLKIP	100	99,4	100	100	Trichothiodystrophy 4, nonphotosensitive, 234050
MPO	100	99,9	100	100	Myeloperoxidase deficiency, 254600 {Alzheimer disease, susceptibility to}, 104300 {Lung cancer, protection against, in smokers}, 0
MPV17	100	97,2	100	100	Charcot-Marie-Tooth disease, axonal, type 2EE, 618400 Mitochondrial DNA depletion syndrome 6 (hepatocerebral type), 256810
MPZ	87,9	84,1	100	100	Charcot-Marie-Tooth disease, type 2J, 607736 Charcot-Marie-Tooth disease, type 1B, 118200 Dejerine-Sottas disease, 145900 Hypomyelinating neuropathy, congenital, 2, 618184 Charcot-Marie-Tooth disease, dominant intermediate D, 607791 Roussy-Levy syndrome, 180800 Charcot-Marie-Tooth disease, type 2I, 607677
MPZL2	100	99,9	100	100	Deafness, autosomal recessive 111, 618145
MRAP	100	100	100	100	Glucocorticoid deficiency 2, 607398
MRE11	98,9	93,3	100	100	Ataxia-telangiectasia-like disorder 1, 604391
MRM2	100	99,4	98,9	98,9	?Mitochondrial DNA depletion syndrome 17, 618567
MRPL3	93,2	87,2	100	100	Combined oxidative phosphorylation deficiency 9, 614582
MRPL44	99,9	98,7	100	100	?Combined oxidative phosphorylation deficiency 16, 615395
MRPS14	100	100	100	100	?Combined oxidative phosphorylation deficiency 38, 618378
MRPS16	100	99,6	100	100	Combined oxidative phosphorylation deficiency 2, 610498
MRPS2	99,6	96,9	100	100	Combined oxidative phosphorylation deficiency 36, 617950
MRPS22	99,8	99,1	100	100	Combined oxidative phosphorylation deficiency 5, 611719 Ovarian dysgenesis 7, 618117
MRPS34	97,6	92	100	100	Combined oxidative phosphorylation deficiency 32, 617664
MRPS7	100	100	100	100	?Combined oxidative phosphorylation deficiency 34, 617872
MS4A1	99,8	98,8	100	100	?Immunodeficiency, common variable, 5, 613495
MSH2	99	96,9	100	100	Mismatch repair cancer syndrome 2, 619096 Muir-Torre syndrome, 158320 Colorectal cancer, hereditary nonpolyposis, type 1, 120435

MSH3	98	97,3	98	98	Familial adenomatous polyposis 4, 617100 Endometrial carcinoma, somatic, 608089
MSH6	100	99,8	100	100	{Endometrial cancer, familial}, 608089 Mismatch repair cancer syndrome 3, 619097 Colorectal cancer, hereditary nonpolyposis, type 5, 614350
MSMO1	96,3	88,9	100	100	Microcephaly, congenital cataract, and psoriasiform dermatitis, 616834
MSRB3	100	99,4	100	100	Deafness, autosomal recessive 74, 613718
MSTO1	99,6	96,8	100	100	Myopathy, mitochondrial, and ataxia, 617675
MTFMT	100	99,8	100	100	Combined oxidative phosphorylation deficiency 15, 614947 Mitochondrial complex I deficiency, nuclear type 27, 618248
MTHFD1	100	99,5	100	100	Combined immunodeficiency and megaloblastic anemia with or without hyperhomocysteinemia, 617780 {Neural tube defects, folate-sensitive, susceptibility to}, 601634
MTHFR	97,3	96	100	100	{Schizophrenia, susceptibility to}, 181500 Homocystinuria due to MTHFR deficiency, 236250 {Neural tube defects, susceptibility to}, 601634 {Thromboembolism, susceptibility to}, 188050 {Vascular disease, susceptibility to}, 0
MTHFS	75	74,9	100	100	Neurodevelopmental disorder with microcephaly, epilepsy, and hypomyelination, 618367
MTMR2	100	99	100	100	Charcot-Marie-Tooth disease, type 4B1, 601382
MTO1	91,3	90,4	91,6	91,4	Combined oxidative phosphorylation deficiency 10, 614702
MTPAP	99,5	96,1	100	100	?Spastic ataxia 4, autosomal recessive, 613672
MTR	100	100	100	100	{Neural tube defects, folate-sensitive, susceptibility to}, 601634 Homocystinuria-megaloblastic anemia, cblG complementation type, 250940
MTRR	100	99,6	100	100	{Neural tube defects, folate-sensitive, susceptibility to}, 601634 Homocystinuria-megaloblastic anemia, cbl E type, 236270
MTTP	100	99,6	100	100	Abetalipoproteinemia, 200100 {Metabolic syndrome, protection against}, 605552
MUSK	100	99,9	100	100	Fetal akinesia deformation sequence 1, 208150 Myasthenic syndrome, congenital, 9, associated with acetylcholine receptor deficiency, 616325
MUTYH	100	100	100	100	Gastric cancer, somatic, 613659 Adenomas, multiple colorectal, 608456
MVK	90,9	90,5	90,5	90,5	Hyper-IgD syndrome, 260920 Porokeratosis 3, multiple types, 175900 Mevalonic aciduria, 610377
MYBPC1	99,9	99,5	100	100	Arthrogryposis, distal, type 1B, 614335 Myopathy, congenital, with tremor, 618524 Lethal congenital contracture syndrome 4, 614915

MYD88	100	100	100	100	Immunodeficiency 68, 612260 Macroglobulinemia, Waldenstrom, somatic, 153600
MYF5	100	100	100	100	Ophthalmoplegia, external, with rib and vertebral anomalies, 618155
MYH2	99,9	99,4	100	100	Proximal myopathy and ophthalmoplegia, 605637
MYH3	99,9	99	100	100	Contractures, pterygia, and spondylocarpotarsal fusion syndrome 1B, 618469 Arthrogryposis, distal, type 2B3 (Sheldon-Hall), 618436 Arthrogryposis, distal, type 2A (Freeman-Sheldon), 193700 Contractures, pterygia, and spondylocarpostarsal fusion syndrome 1A, 178110
MYL1	100	99,7	100	100	Myopathy, congenital, with fast-twitch (type II) fiber atrophy, 618414
MYL3	100	100	100	100	Cardiomyopathy, hypertrophic, 8, 608751
MYLK	100	99,9	100	100	Aortic aneurysm, familial thoracic 7, 613780 Megacystis-microcolon-intestinal hypoperistalsis syndrome, 249210
MYMK	100	100	100	100	Carey-Fineman-Ziter syndrome, 254940
MYO15A	98,8	97	100	99,9	Deafness, autosomal recessive 3, 600316
MYO18B	100	99,1	100	100	Klippel-Feil syndrome 4, autosomal recessive, with myopathy and facial dysmorphism, 616549
MYO1E	99,9	99,5	100	100	Glomerulosclerosis, focal segmental, 6, 614131
MYO3A	99,6	96,6	100	100	Deafness, autosomal recessive 30, 607101
MYO5A	99,8	98,9	100	100	Griscelli syndrome, type 1, 214450
MYO5B	99,1	96,2	100	100	Microvillus inclusion disease, 251850
MYO6	99,5	96,6	100	100	Deafness, autosomal recessive 37, 607821 Deafness, autosomal dominant 22, 606346 Deafness, autosomal dominant 22, with hypertrophic cardiomyopathy, 606346
MYO7A	99,3	97,3	100	100	Deafness, autosomal recessive 2, 600060 Deafness, autosomal dominant 11, 601317 Usher syndrome, type 1B, 276900
MYO9A	99,9	99,1	100	100	Myasthenic syndrome, congenital, 24, presynaptic, 618198
MYPN	100	99,7	100	100	Cardiomyopathy, dilated, 1KK, 615248 Cardiomyopathy, familial restrictive, 4, 615248 Nemaline myopathy 11, autosomal recessive, 617336 Cardiomyopathy, hypertrophic, 22, 615248
MYSM1	96,4	95,5	96,4	96,4	Bone marrow failure syndrome 4, 618116
NADK2	99,9	97,2	99	96,3	2,4-dienoyl-CoA reductase deficiency, 616034
NADSYN1	100	100	100	100	Vertebral, cardiac, renal, and limb defects syndrome 3, 618845
NAGA	100	100	100	100	Kanzaki disease, 609242 Schindler disease, type I, 609241 Schindler disease, type III, 609241

NAGLU	92,9	89,9	99,9	99,2	Mucopolysaccharidosis type IIIB (Sanfilippo B), 252920 ?Charcot-Marie-Tooth disease, axonal, type 2V, 616491
NAGS	99,7	95	100	100	N-acetylglutamate synthase deficiency, 237310
NALCN	100	99,5	99,8	99,8	Hypotonia, infantile, with psychomotor retardation and characteristic facies 1, 615419 Congenital contractures of the limbs and face, hypotonia, and developmental delay, 616266
NANS	100	99,9	100	100	Spondyloepimetaphyseal dysplasia, Camera-Genevieve type, 610442
NARS2	98,3	97,4	100	100	?Deafness, autosomal recessive 94, 618434 Combined oxidative phosphorylation deficiency 24, 616239
NAT8L	99,5	94,2	95,6	90,8	?N-acetylaspartate deficiency, 614063
NAXD	100	100	100	100	Encephalopathy, progressive, early-onset, with brain edema and/or leukoencephalopathy, 2, 618321
NAXE	100	99,8	100	100	Encephalopathy, progressive, early-onset, with brain edema and/or leukoencephalopathy, 617186
NBAS	100	99,6	100	100	Infantile liver failure syndrome 2, 616483 Short stature, optic nerve atrophy, and Pelger-Huet anomaly, 614800
NBEAL2	99,4	99,4	100	100	Gray platelet syndrome, 139090
NBN	99,9	98,6	100	100	Leukemia, acute lymphoblastic, 613065 Nijmegen breakage syndrome, 251260 Aplastic anemia, 609135
NCAPD2	100	99,7	100	100	?Microcephaly 21, primary, autosomal recessive, 617983
NCAPD3	99,9	98,9	100	100	Microcephaly 22, primary, autosomal recessive, 617984
NCAPG2	99,9	99,2	100	100	Khan-Khan-Katsanis syndrome, 618460
NCAPH	100	100	100	100	?Microcephaly 23, primary, autosomal recessive, 617985
NCF1	26	25,8	100	99,8	Chronic granulomatous disease 1, autosomal recessive, 233700
NCF2	99,9	98,3	100	100	Chronic granulomatous disease 2, autosomal recessive, 233710
NCF4	100	100	100	100	Chronic granulomatous disease 3, autosomal recessive, 613960
NDE1	100	100	100	100	Lissencephaly 4 (with microcephaly), 614019 ?Microhydranencephaly, 605013
NDRG1	100	100	100	100	Charcot-Marie-Tooth disease, type 4D, 601455
NDST1	100	100	100	100	Mental retardation, autosomal recessive 46, 616116
NDUFA10	99,8	98,6	100	100	Mitochondrial complex I deficiency, nuclear type 22, 618243
NDUFA11	100	100	100	99,8	Mitochondrial complex I deficiency, nuclear type 14, 618236
NDUFA12	100	100	100	100	?Mitochondrial complex I deficiency, nuclear type 23, 618244
NDUFA13	92,2	89,2	100	100	?Mitochondrial complex I deficiency, nuclear type 28, 618249 {Thyroid carcinoma, Hurthle cell}, 607464
NDUFA2	100	100	100	100	Mitochondrial complex I deficiency, nuclear type 13, 618235
NDUFA6	100	100	100	100	Mitochondrial complex I deficiency, nuclear type 33, 618253
NDUFA9	99,9	96,5	100	100	Mitochondrial complex I deficiency, nuclear type 26, 618247
NDUFAF1	100	100	100	100	Mitochondrial complex I deficiency, nuclear type 11, 618234

NDUFAF2	95	83,4	100	99,9	Mitochondrial complex I deficiency, nuclear type 10, 618233
NDUFAF3	100	99,9	100	100	Mitochondrial complex I deficiency, nuclear type 18, 618240
NDUFAF4	99,8	98,2	100	100	Mitochondrial complex I deficiency, nuclear type 15, 618237
NDUFAF5	100	99,5	100	100	Mitochondrial complex I deficiency, nuclear type 16, 618238
NDUFAF6	100	96,8	100	100	Fanconi renotubular syndrome 5, 618913 Mitochondrial complex I deficiency, nuclear type 17, 618239
NDUFB11	99,5	96,5	100	99,5	Linear skin defects with multiple congenital anomalies 3, 300952 ?Mitochondrial complex I deficiency, nuclear type 30, 301021
NDUFB3	95,8	80,5	100	100	Mitochondrial complex I deficiency, nuclear type 25, 618246
NDUFB8	100	99,9	100	100	Mitochondrial complex I deficiency, nuclear type 32, 618252
NDUFB9	96,5	92,5	98,7	98,7	?Mitochondrial complex I deficiency, nuclear type 24, 618245
NDUFS1	100	99,5	100	100	Mitochondrial complex I deficiency, nuclear type 5, 618226
NDUFS2	100	100	100	100	Mitochondrial complex I deficiency, nuclear type 6, 618228
NDUFS3	90,7	90,6	91,9	90,7	Mitochondrial complex I deficiency, nuclear type 8, 618230
NDUFS4	100	99,4	100	100	Mitochondrial complex I deficiency, nuclear type 1, 252010
NDUFS6	100	99,9	100	100	Mitochondrial complex I deficiency, nuclear type 9, 618232
NDUFS7	100	99,2	100	100	Mitochondrial complex I deficiency, nuclear type 3, 618224
NDUFS8	100	99,4	100	100	Mitochondrial complex I deficiency, nuclear type 2, 618222
NDUFV1	98	96,1	100	100	Mitochondrial complex I deficiency, nuclear type 4, 618225
NDUFV2	86,9	76,9	100	100	Mitochondrial complex I deficiency, nuclear type 7, 618229
NEB	83	82,6	99,9	99,9	Nemaline myopathy 2, autosomal recessive, 256030
NECAP1	100	100	100	100	Developmental and epileptic encephalopathy 21, 615833
NECTIN1	100	99,9	100	100	Orofacial cleft 7, 225060 Cleft lip/palate-ectodermal dysplasia syndrome, 225060
NECTIN4	100	100	100	100	Ectodermal dysplasia-syndactyly syndrome 1, 613573
NEK1	99,8	98	100	100	{Amyotrophic lateral sclerosis, susceptibility to, 24}, 617892 Short-rib thoracic dysplasia 6 with or without polydactyly, 263520
NEK2	99,7	95,5	96,1	96,1	?Retinitis pigmentosa 67, 615565
NEK8	100	99,9	100	100	?Nephronophthisis 9, 613824 Renal-hepatic-pancreatic dysplasia 2, 615415
NEK9	100	99,6	100	100	Lethal congenital contracture syndrome 10, 617022 Nevus comedonicus, somatic, 617025 ?Arthrogryposis, Perthes disease, and upward gaze palsy, 614262
NEPRO	100	99,7	100	100	Anauxetic dysplasia 3, 618853
NEU1	99,7	97,7	100	100	Sialidosis, type II, 256550 Sialidosis, type I, 256550
NEUROG3	100	100	100	100	Diarrhea 4, malabsorptive, congenital, 610370

NFASC	100	99,9	100	100	Neurodevelopmental disorder with central and peripheral motor dysfunction, 618356
NFU1	98,8	90,8	100	100	Multiple mitochondrial dysfunctions syndrome 1, 605711
NGF	100	100	100	100	Neuropathy, hereditary sensory and autonomic, type V, 608654
NGLY1	100	99,8	100	100	Congenital disorder of deglycosylation, 615273
NHEJ1	100	96,2	100	100	Severe combined immunodeficiency with microcephaly, growth retardation, and sensitivity to ionizing radiation, 611291
NHLRC1	100	98,7	100	100	Epilepsy, progressive myoclonic 2B (Lafora), 254780
NHLRC2	99,6	98,2	100	100	FINCA syndrome, 618278
NHP2	100	100	100	100	Dyskeratosis congenita, autosomal recessive 2, 613987
NIN	100	99,5	99,1	99,1	?Seckel syndrome 7, 614851
NIPAL4	100	99,1	100	100	Ichthyosis, congenital, autosomal recessive 6, 612281
NKX2-6	100	99,5	100	100	Persistent truncus arteriosus, 217095 Conotruncal heart malformations, 217095
NKX3-2	99,8	97	100	100	Spondylo-megaepiphyseal-metaphyseal dysplasia, 613330
NKX6-2	89	81,8	100	100	Spastic ataxia 8, autosomal recessive, with hypomyelinating leukodystrophy, 617560
NLRP1	99,6	98	100	100	Palmoplantar carcinoma, multiple self-healing, 615225 Autoinflammation with arthritis and dyskeratosis, 617388 {Vitiligo-associated multiple autoimmune disease susceptibility 1}, 606579 ?Respiratory papillomatosis, juvenile recurrent, congenital, 618803
NLRP7	100	99,6	100	100	Hydatidiform mole, recurrent, 1, 231090
NME8	99,2	95,3	100	100	Ciliary dyskinesia, primary, 6, 610852
NMNAT1	100	99,2	98,3	95,6	Leber congenital amaurosis 9, 608553
NNT	96,4	95,9	96,4	96,4	Glucocorticoid deficiency 4, with or without mineralocorticoid deficiency, 614736
NOP10	100	99,8	100	100	Dyskeratosis congenita, autosomal recessive 1, 224230
NPC1	99,6	98,7	100	100	Niemann-Pick disease, type D, 257220 Niemann-Pick disease, type C1, 257220
NPC2	100	99,6	100	100	Niemann-pick disease, type C2, 607625
NPHP1	100	99	100	100	Nephronophthisis 1, juvenile, 256100 Senior-Loken syndrome-1, 266900 Joubert syndrome 4, 609583
NPHP3	99,7	98,4	100	100	Meckel syndrome 7, 267010 Renal-hepatic-pancreatic dysplasia 1, 208540 Nephronophthisis 3, 604387
NPHP4	100	99,8	100	100	Nephronophthisis 4, 606966 Senior-Loken syndrome 4, 606996
NPHS1	99,8	99,1	100	100	Nephrotic syndrome, type 1, 256300
NPHS2	100	99,5	100	100	Nephrotic syndrome, type 2, 600995

NPPA	100	100	100	100	Atrial standstill 2, 615745 Atrial fibrillation, familial, 6, 612201
NPR2	100	99,6	100	100	Short stature with nonspecific skeletal abnormalities, 616255 Epiphyseal chondrodysplasia, Miura type, 615923 Acromesomelic dysplasia, Maroteaux type, 602875
NR0B2	100	99,3	100	100	Obesity, mild, early-onset, 601665
NR1H4	99,8	98,5	100	100	Cholestasis, progressive familial intrahepatic, 5, 617049
NR2E3	100	99,6	100	100	Enhanced S-cone syndrome, 268100 Retinitis pigmentosa 37, 611131
NRROS	100	100	100	100	Seizures, early-onset, with neurodegeneration and brain calcification, 618875
NRXN1	97,4	96,9	100	99,8	Pitt-Hopkins-like syndrome 2, 614325 {Schizophrenia, susceptibility to, 17}, 614332
NSMCE2	99,7	98,2	100	100	Seckel syndrome 10, 617253
NSMCE3	100	100	100	100	Lung disease, immunodeficiency, and chromosome breakage syndrome, 617241
NSUN2	96	95,1	100	100	Mental retardation, autosomal recessive 5, 611091
NT5C2	98	96,5	100	100	Spastic paraplegia 45, autosomal recessive, 613162
NT5C3A	97,8	88,2	100	100	Anemia, hemolytic, due to UMPH1 deficiency, 266120
NT5E	100	99,9	100	100	Calcification of joints and arteries, 211800
NTHL1	100	99,8	100	100	Familial adenomatous polyposis 3, 616415
NTNG2	98,5	96,7	99,9	99	Neurodevelopmental disorder with behavioral abnormalities, absent speech, and hypotonia, 618718
NTRK1	99,8	98,2	100	100	Insensitivity to pain, congenital, with anhidrosis, 256800
NUBPL	99,7	98,4	100	100	Mitochondrial complex I deficiency, nuclear type 21, 618242
NUP107	99,8	98,5	100	100	Galloway-Mowat syndrome 7, 618348 ?Ovarian dysgenesis 6, 618078 Nephrotic syndrome, type 11, 616730
NUP133	99,7	98,3	100	100	Nephrotic syndrome, type 18, 618177 ?Galloway-Mowat syndrome 8, 618349
NUP160	100	99,9	100	100	?Nephrotic syndrome, type 19, 618178
NUP188	100	99,6	100	100	Sandestig-Stefanova syndrome, 618804
NUP205	99,9	99,4	100	100	?Nephrotic syndrome, type 13, 616893
NUP214	100	99,7	100	100	Leukemia, acute myeloid, somatic, 601626 {Encephalopathy, acute, infection-induced, susceptibility to, 9}, 618426 Leukemia, T-cell acute lymphoblastic, somatic, 613065
NUP37	100	100	100	100	?Microcephaly 24, primary, autosomal recessive, 618179
NUP62	100	100	100	100	Striatonigral degeneration, infantile, 271930
NUP85	100	100	100	100	Nephrotic syndrome, type 17, 618176
NUP88	100	100	100	100	Fetal akinesia deformation sequence 4, 618393

NUP93	98	94,2	95,5	95,5	Nephrotic syndrome, type 12, 616892
NUS1	60	44,5	100	100	Mental retardation, autosomal dominant 55, with seizures, 617831 ?Congenital disorder of glycosylation, type 1aa, 617082
NXN	100	100	99,9	99,5	Robinow syndrome, autosomal recessive 2, 618529
OAT	85,2	76,3	100	100	Gyrate atrophy of choroid and retina with or without ornithinemia, 258870
OBSL1	100	99,3	100	100	3-M syndrome 2, 612921
OCA2	99,9	98,7	100	100	[Skin/hair/eye pigmentation 1, blue/nonblue eyes], 227220 [Skin/hair/eye pigmentation 1, blond/brown hair], 227220 Albinism, oculocutaneous, type II, 203200 Albinism, brown oculocutaneous, 203200
OCLN	100	100	100	100	Pseudo-TORCH syndrome 1, 251290
ODAPH	100	100	100	100	Amelogenesis imperfecta, type IIA4, 614832
OGDH	100	99,9	100	100	Alpha-ketoglutarate dehydrogenase deficiency, 203740
OPA1	99,6	97,6	100	100	{Glaucoma, normal tension, susceptibility to}, 606657 Behr syndrome, 210000 Optic atrophy 1, 165500 Optic atrophy plus syndrome, 125250 ?Mitochondrial DNA depletion syndrome 14 (encephalocardiomyopathic type), 616896
OPA3	100	99	100	100	Optic atrophy 3 with cataract, 165300 3-methylglutaconic aciduria, type III, 258501
OPLAH	100	99,8	100	100	5-oxoprolinase deficiency, 260005
ORAI1	99,1	96,4	99,6	97,1	Myopathy, tubular aggregate, 2, 615883 Immunodeficiency 9, 612782
ORC1	100	99,4	100	100	Meier-Gorlin syndrome 1, 224690
ORC4	98,7	93,6	100	100	Meier-Gorlin syndrome 2, 613800
ORC6	100	99,9	100	100	Meier-Gorlin syndrome 3, 613803
OSGEP	100	99,4	100	100	Galloway-Mowat syndrome 3, 617729
OSTM1	98,6	94	100	100	Osteopetrosis, autosomal recessive 5, 259720
OTOA	99,4	97,6	100	99,9	Deafness, autosomal recessive 22, 607039
OTOF	100	99,9	100	100	Auditory neuropathy, autosomal recessive, 1, 601071 Deafness, autosomal recessive 9, 601071
OTOG	99,4	98,6	100	100	Deafness, autosomal recessive 18B, 614945
OTOGL	99,5	97,4	100	100	Deafness, autosomal recessive 84B, 614944
OTUD6B	99,9	98,8	100	100	Intellectual developmental disorder with dysmorphic facies, seizures, and distal limb anomalies, 617452
OTULIN	92,6	86,5	99,2	95	Autoinflammation, panniculitis, and dermatosis syndrome, 617099
OXCT1	99,8	98,1	100	100	Succinyl CoA:3-oxoacid CoA transferase deficiency, 245050

P2RY12	100	100	100	100	Bleeding disorder, platelet-type, 8, 609821
P3H1	100	100	100	100	Osteogenesis imperfecta, type VIII, 610915
P3H2	99,8	98	100	100	Myopia, high, with cataract and vitreoretinal degeneration, 614292
P4HTM	99	97,4	100	99,4	Hypotonia, hypoventilation, impaired intellectual development, dysautonomia, epilepsy, and eye abnormalities, 618493
PAH	100	100	100	100	Phenylketonuria, 261600 [Hyperphenylalaninemia, non-PKU mild], 261600
PAM16	65,3	65,2	82,9	82,9	Spondylometaphyseal dysplasia, Megarbane-Dagher-Melike type, 613320
PANK2	100	99,3	100	100	HARP syndrome, 607236 Neurodegeneration with brain iron accumulation 1, 234200
PAPSS2	100	99,5	100	100	Brachyolmia 4 with mild epiphyseal and metaphyseal changes, 612847
PARK7	100	100	100	100	Parkinson disease 7, autosomal recessive early-onset, 606324
PARN	81,2	81,1	88,1	87,6	Pulmonary fibrosis and/or bone marrow failure, telomere-related, 4, 616371 Dyskeratosis congenita, autosomal recessive 6, 616353
PARS2	100	100	100	100	Developmental and epileptic encephalopathy 75, 618437
PATL2	100	99	100	100	Oocyte maturation defect 4, 617743
PAX1	92,4	87,9	100	99,6	Otofaciocervical syndrome 2, 615560
PAX3	100	99,9	100	100	Craniofacial-deafness-hand syndrome, 122880 Rhabdomyosarcoma 2, alveolar, 268220 Waardenburg syndrome, type 1, 193500 Waardenburg syndrome, type 3, 148820
PAX7	100	100	100	100	Myopathy, congenital, progressive, with scoliosis, 618578 Rhabdomyosarcoma 2, alveolar, 268220
PC	99,8	97,3	100	100	Pyruvate carboxylase deficiency, 266150
PCARE	99,6	98,5	100	100	Retinitis pigmentosa 54, 613428
PCBD1	100	99,6	100	99,7	Hyperphenylalaninemia, BH4-deficient, D, 264070
PCCA	99,5	96,7	100	100	Propionicacidemia, 606054
PCCB	97,9	96	98,7	96,2	Propionicacidemia, 606054
PCDH12	100	100	100	100	Diencephalic-mesencephalic junction dysplasia syndrome 1, 251280
PCDH15	97,8	96,7	100	100	Usher syndrome, type 1D/F digenic, 601067 Usher syndrome, type 1F, 602083 Deafness, autosomal recessive 23, 609533
PCK1	100	100	100	100	?Phosphoenolpyruvate carboxykinase deficiency, cytosolic, 261680
PCK2	100	100	100	100	PEPCK deficiency, mitochondrial, 261650
PCLO	99,7	98,7	100	100	?Pontocerebellar hypoplasia, type 3, 608027
PCNA	100	98,4	100	100	?Ataxia-telangiectasia-like disorder 2, 615919
PCNT	99,6	97,1	100	100	Microcephalic osteodysplastic primordial dwarfism, type II, 210720

PCSK1	100	99,5	100	100	{Obesity, susceptibility to, BMIQ12}, 612362 Obesity with impaired prohormone processing, 600955
PCYT1A	98,9	95,5	100	100	Spondylometaphyseal dysplasia with cone-rod dystrophy, 608940
PDE10A	65,5	64,5	86,9	84,1	Dyskinesia, limb and orofacial, infantile-onset, 616921 Striatal degeneration, autosomal dominant, 616922
PDE6A	100	99,6	100	100	Retinitis pigmentosa 43, 613810
PDE6B	100	99,9	100	100	Night blindness, congenital stationary, autosomal dominant 2, 163500 Retinitis pigmentosa-40, 613801
PDE6C	99,9	97,8	100	100	Cone dystrophy 4, 613093
PDE6D	100	100	100	100	Joubert syndrome 22, 615665
PDE6G	100	100	100	100	Retinitis pigmentosa 57, 613582
PDE6H	100	97,9	100	100	Retinal cone dystrophy 3, 610024 Achromatopsia 6, 610024
PDHB	99,1	97,5	100	100	Pyruvate dehydrogenase E1-beta deficiency, 614111
PDHX	99,9	99,4	100	100	Lacticacidemia due to PDX1 deficiency, 245349
PDP1	100	100	100	100	Pyruvate dehydrogenase phosphatase deficiency, 608782
PDSS1	94,7	87,6	97,3	96,6	Coenzyme Q10 deficiency, primary, 2, 614651
PDSS2	99,8	97,1	100	100	Coenzyme Q10 deficiency, primary, 3, 614652
PDX1	93	82,4	100	100	{Diabetes mellitus, type II, susceptibility to}, 125853 Pancreatic agenesis 1, 260370 MODY, type IV, 606392
PDXK	79,3	76,6	99,4	96,7	Neuropathy, hereditary motor and sensory, type VIC, with optic atrophy, 618511
PDZD7	97	93	100	99,8	Usher syndrome, type IIC, GPR98/PDZD7 digenic, 605472 Deafness, autosomal recessive 57, 618003 {Retinal disease in Usher syndrome type IIA, modifier of}, 276901
PEPD	100	98,8	100	100	Prolidase deficiency, 170100
PET100	100	99,6	100	100	Mitochondrial complex IV deficiency, nuclear type 12, 619055
PEX1	99,9	99,4	100	100	Heimler syndrome 1, 234580 Peroxisome biogenesis disorder 1B (NALD/IRD), 601539 Peroxisome biogenesis disorder 1A (Zellweger), 214100
PEX10	96,8	89,7	100	99,9	Peroxisome biogenesis disorder 6B, 614871 Peroxisome biogenesis disorder 6A (Zellweger), 614870
PEX11B	100	99,6	100	100	Peroxisome biogenesis disorder 14B, 614920
PEX12	100	100	100	100	Peroxisome biogenesis disorder 3A (Zellweger), 614859 Peroxisome biogenesis disorder 3B, 266510
PEX13	100	100	100	100	Peroxisome biogenesis disorder 11A (Zellweger), 614883 Peroxisome biogenesis disorder 11B, 614885

PEX14	96,7	90,8	100	100	Peroxisome biogenesis disorder 13A (Zellweger), 614887
PEX16	97,9	94,2	100	100	Peroxisome biogenesis disorder 8A (Zellweger), 614876 Peroxisome biogenesis disorder 8B, 614877
PEX19	99,9	98,5	100	100	Peroxisome biogenesis disorder 12A (Zellweger), 614886
PEX2	100	100	100	100	Peroxisome biogenesis disorder 5A (Zellweger), 614866 Peroxisome biogenesis disorder 5B, 614867
PEX26	100	100	100	100	Peroxisome biogenesis disorder 7A (Zellweger), 614872 Peroxisome biogenesis disorder 7B, 614873
PEX3	100	99,3	100	100	Peroxisome biogenesis disorder 10A (Zellweger), 614882 ?Peroxisome biogenesis disorder 10B, 617370
PEX5	99,9	99	100	100	Peroxisome biogenesis disorder 2B, 202370 Rhizomelic chondrodysplasia punctata, type 5, 616716 Peroxisome biogenesis disorder 2A (Zellweger), 214110
PEX6	94,5	86,7	100	100	Peroxisome biogenesis disorder 4B, 614863 Heimler syndrome 2, 616617 Peroxisome biogenesis disorder 4A (Zellweger), 614862
PEX7	87,8	80,7	91,3	91,3	Peroxisome biogenesis disorder 9B, 614879 Rhizomelic chondrodysplasia punctata, type 1, 215100
PFKM	100	99,5	100	100	Glycogen storage disease VII, 232800
PGAM2	100	100	100	100	Glycogen storage disease X, 261670
PGAP1	99	94,4	100	100	Mental retardation, autosomal recessive 42, 615802
PGAP2	100	99,9	100	100	Hyperphosphatasia with mental retardation syndrome 3, 614207
PGAP3	63,5	59,6	100	100	Hyperphosphatasia with mental retardation syndrome 4, 615716
PGM1	94,2	94,2	94,2	94,2	Congenital disorder of glycosylation, type It, 614921
PGM3	100	99,8	91,7	91,7	Immunodeficiency 23, 615816
PHGDH	99,9	98,8	100	100	Neu-Laxova syndrome 1, 256520 Phosphoglycerate dehydrogenase deficiency, 601815
PHKB	99,9	99,2	100	100	Phosphorylase kinase deficiency of liver and muscle, autosomal recessive, 261750
PHKG2	100	99,9	100	100	Glycogen storage disease IXc, 613027 Cirrhosis due to liver phosphorylase kinase deficiency, 0
PHOX2A	91,6	72,7	100	99,8	Fibrosis of extraocular muscles, congenital, 2, 602078
PHYH	100	99,6	100	100	Refsum disease, 266500
PI4KA	92,6	88,8	99,9	99,9	Polymicrogyria, perisylvian, with cerebellar hypoplasia and arthrogryposis, 616531
PIBF1	99,5	96,2	100	100	Joubert syndrome 33, 617767
PIEZO1	99,9	98,8	100	100	Lymphatic malformation 6, 616843 Dehydrated hereditary stomatocytosis with or without pseudohyperkalemia and/or perinatal edema, 194380

PIEZO2	100	99,5	100	100	Arthrogryposis, distal, with impaired proprioception and touch, 617146 Arthrogryposis, distal, type 5, 108145 ?Marden-Walker syndrome, 248700 Arthrogryposis, distal, type 3, 114300
PIGB	99,9	97,8	100	100	Developmental and epileptic encephalopathy 80, 618580
PIGC	99,2	90,9	100	100	Glycosylphosphatidylinositol biosynthesis defect 16, 617816
PIGG	100	99,7	100	100	Mental retardation, autosomal recessive 53, 616917
PIGH	82,1	68,1	75,2	74,4	Glycosylphosphatidylinositol biosynthesis defect 17, 618010
PIGK	99,2	95,1	100	100	Neurodevelopmental disorder with hypotonia and cerebellar atrophy, with or without seizures, 618879
PIGL	100	100	100	100	CHIME syndrome, 280000
PIGM	100	100	100	100	Glycosylphosphatidylinositol deficiency, 610293
PIGN	93,8	91,5	98,8	98,8	Multiple congenital anomalies-hypotonia-seizures syndrome 1, 614080
PIGO	100	99,9	100	100	Hyperphosphatasia with mental retardation syndrome 2, 614749
PIGP	95,8	87,3	100	100	Developmental and epileptic encephalopathy 55, 617599
PIGQ	92,8	90,8	100	100	Developmental and epileptic encephalopathy 77, 618548
PIGS	100	100	100	100	Glycosylphosphatidylinositol biosynthesis defect 18, 618143
PIGT	98,1	98,1	100	100	?Paroxysmal nocturnal hemoglobinuria 2, 615399 Multiple congenital anomalies-hypotonia-seizures syndrome 3, 615398
PIGU	100	99,1	100	100	Neurodevelopmental disorder with brain anomalies, seizures, and scoliosis, 618590
PIGV	100	100	100	100	Hyperphosphatasia with mental retardation syndrome 1, 239300
PIGW	100	99,8	100	100	Glycosylphosphatidylinositol biosynthesis defect 11, 616025
PIGY	100	99,9	100	100	Hyperphosphatasia with mental retardation syndrome 6, 616809
PIK3C2A	99,2	96,9	100	100	Oculoskeletodental syndrome, 618440
PIK3R1	99,8	99	100	100	SHORT syndrome, 269880 Immunodeficiency 36, 616005 ?Agammaglobulinemia 7, autosomal recessive, 615214
PIK3R5	100	99,9	100	100	Ataxia-oculomotor apraxia 3, 615217
PINK1	90,7	86,9	99,9	99,4	Parkinson disease 6, early onset, 605909
PIP5K1C	98	95,8	99,9	99,8	Lethal congenital contractural syndrome 3, 611369
PJVK	100	99,7	100	100	Deafness, autosomal recessive 59, 610220
PKD1L1	100	99,8	100	100	Heterotaxy, visceral, 8, autosomal, 617205
PKHD1	100	99,6	100	100	Polycystic kidney disease 4, with or without hepatic disease, 263200
PKLR	100	99,2	100	100	Pyruvate kinase deficiency, 266200 Adenosine triphosphate, elevated, of erythrocytes, 102900
PKP1	100	99,1	100	100	Ectodermal dysplasia/skin fragility syndrome, 604536

PLA2G6	92,2	90,7	92,3	92,3	Infantile neuroaxonal dystrophy 1, 256600 Parkinson disease 14, autosomal recessive, 612953 Neurodegeneration with brain iron accumulation 2B, 610217
PLA2G7	99,9	99	100	100	{Asthma, susceptibility to}, 600807 Platelet-activating factor acetylhydrolase deficiency, 614278 {Atopy, susceptibility to}, 147050
PLAA	100	99,2	100	100	Neurodevelopmental disorder with progressive microcephaly, spasticity, and brain anomalies, 617527
PLCB1	100	99,8	100	100	Developmental and epileptic encephalopathy 12, 613722
PLCD1	99,9	97,8	100	100	Nail disorder, nonsyndromic congenital, 3, (leukonychia), 151600
PLCE1	99,9	99,3	100	100	Nephrotic syndrome, type 3, 610725
PLD1	100	99,6	100	100	Cardiac valvular defect, developmental, 212093
PLEC	100	99,8	100	100	Muscular dystrophy, limb-girdle, autosomal recessive 17, 613723 Epidermolysis bullosa simplex with pyloric atresia, 612138 Epidermolysis bullosa simplex with muscular dystrophy, 226670 ?Epidermolysis bullosa simplex with nail dystrophy, 616487 Epidermolysis bullosa simplex, Ogna type, 131950
PLEKHG2	100	99,3	100	100	Leukodystrophy and acquired microcephaly with or without dystonia, 616763
PLEKHG5	95,3	91,1	96,3	96,2	Spinal muscular atrophy, distal, autosomal recessive, 4, 611067 Charcot-Marie-Tooth disease, recessive intermediate C, 615376
PLEKHM1	100	99,8	100	100	Osteopetrosis, autosomal dominant 3, 618107 ?Osteopetrosis, autosomal recessive 6, 611497
PLG	87,8	87,5	100	100	Dysplasminogenemia, 217090 Plasminogen deficiency, type I, 217090
PLK4	99,9	98,2	100	100	Microcephaly and chorioretinopathy, autosomal recessive, 2, 616171
PLOD1	100	98,4	100	100	Ehlers-Danlos syndrome, kyphoscoliotic type, 1, 225400
PLOD2	99,3	97,3	100	100	Bruck syndrome 2, 609220
PLOD3	99,8	98	100	100	Lysyl hydroxylase 3 deficiency, 612394
PLPBP	98,2	90,1	100	100	Epilepsy, early-onset, vitamin B6-dependent, 617290
PLVAP	100	100	100	100	Diarrhea 10, protein-losing enteropathy type, 618183
PMM2	100	100	100	100	Congenital disorder of glycosylation, type Ia, 212065
PMPCA	97,7	94,2	100	100	Spinocerebellar ataxia, autosomal recessive 2, 213200
PMPCB	100	99,7	100	100	Multiple mitochondrial dysfunctions syndrome 6, 617954
PMS2	84,3	82,8	100	100	Colorectal cancer, hereditary nonpolyposis, type 4, 614337 Mismatch repair cancer syndrome 4, 619101
PNKP	100	100	100	100	Ataxia-oculomotor apraxia 4, 616267 ?Charcot-Marie-Tooth disease, type 2B2, 605589 Microcephaly, seizures, and developmental delay, 613402

PNLIP	100	99,8	100	100	?Pancreatic lipase deficiency, 614338
PNP	99,8	98,9	100	100	Immunodeficiency due to purine nucleoside phosphorylase deficiency, 613179
PNPLA1	100	100	100	100	Ichthyosis, congenital, autosomal recessive 10, 615024
PNPLA2	99,7	96,1	100	100	Neutral lipid storage disease with myopathy, 610717
PNPLA6	100	99,7	100	100	Spastic paraplegia 39, autosomal recessive, 612020 Boucher-Neuhauser syndrome, 215470 Oliver-McFarlane syndrome, 275400 ?Laurence-Moon syndrome, 245800
PNPLA8	100	99,8	100	100	?Mitochondrial myopathy with lactic acidosis, 251950
PNPO	99,9	97,7	100	100	Pyridoxamine 5'-phosphate oxidase deficiency, 610090
PNPT1	97,7	89,7	100	100	Deafness, autosomal recessive 70, 614934 Combined oxidative phosphorylation deficiency 13, 614932
POC1A	100	100	100	100	Short stature, onychodysplasia, facial dysmorphism, and hypotrichosis, 614813
POC1B	99,8	98,8	100	100	Cone-rod dystrophy 20, 615973
POGLUT1	99,4	94,6	100	100	Dowling-Degos disease 4, 615696 ?Muscular dystrophy, limb-girdle, autosomal recessive 21, 617232
POLE	100	99,8	100	100	FILS syndrome, 615139 IMAGE-I syndrome, 618336 {Colorectal cancer, susceptibility to, 12}, 615083
POLG	100	99,3	100	100	Progressive external ophthalmoplegia, autosomal dominant 1, 157640 Mitochondrial DNA depletion syndrome 4B (MNGIE type), 613662 Mitochondrial recessive ataxia syndrome (includes SANDO and SCAE), 607459 Mitochondrial DNA depletion syndrome 4A (Alpers type), 203700 Progressive external ophthalmoplegia, autosomal recessive 1, 258450
POLH	100	99,6	100	100	Xeroderma pigmentosum, variant type, 278750
POLR1C	90,5	87	82,8	82,8	Treacher Collins syndrome 3, 248390 Leukodystrophy, hypomyelinating, 11, 616494
POLR1D	91,6	91,6	100	100	Treacher Collins syndrome 2, 613717
POLR3A	100	99,7	100	100	Leukodystrophy, hypomyelinating, 7, with or without oligodontia and/or hypogonadotropic hypogonadism, 607694 Wiedemann-Rautenstrauch syndrome, 264090
POLR3B	99,9	98,6	100	100	Leukodystrophy, hypomyelinating, 8, with or without oligodontia and/or hypogonadotropic hypogonadism, 614381
POMC	100	100	100	100	{Obesity, early-onset, susceptibility to}, 601665 Obesity, adrenal insufficiency, and red hair due to POMC deficiency, 609734
POMGNT1	100	99,9	100	100	Muscular dystrophy-dystroglycanopathy (congenital with mental retardation), type B, 3, 613151 Muscular dystrophy-dystroglycanopathy (limb-girdle), type C, 3, 613157

					Retinitis pigmentosa 76, 617123 Muscular dystrophy-dystroglycanopathy (congenital with brain and eye anomalies), type A, 3, 253280
POMGNT2	100	100	100	100	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	100	?Muscular dystrophy-dystroglycanopathy (limb-girdle), type C, 12, 616094 Muscular dystrophy-dystroglycanopathy (congenital with brain and eye anomalies), type A, 12, 615249
POMP	100	99,1	100	100	Proteasome-associated autoinflammatory syndrome 2, 618048 Keratosis linearis with ichthyosis congenita and sclerosing keratoderma, 601952
POMT1	99,3	97,5	100	100	Muscular dystrophy-dystroglycanopathy (limb-girdle), type C, 1, 609308 Muscular dystrophy-dystroglycanopathy (congenital with brain and eye anomalies), type A, 1, 236670 Muscular dystrophy-dystroglycanopathy (congenital with mental retardation), type B, 1, 613155
POMT2	99,4	96,4	100	100	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 mental retardation), type B, 2, 613156
POP1	100	99,7	100	100	Anauxetic dysplasia 2, 617396
POR	99,8	98,6	100	100	Disordered steroidogenesis due to cytochrome P450 oxidoreductase, 613571 Antley-Bixler syndrome with genital anomalies and disordered steroidogenesis, 201750
POU1F1	100	99,2	100	100	Pituitary hormone deficiency, combined, 1, 613038
PPA2	98,7	94	100	100	Sudden cardiac failure, infantile, 617222 ?Sudden cardiac failure, alcohol-induced, 617223
PPCS	99,8	99,5	100	100	Cardiomyopathy, dilated, 2C, 618189
PPIB	100	100	100	100	Osteogenesis imperfecta, type IX, 259440
PPIP5K2	98,9	95,2	100	100	Deafness, autosomal recessive 100, 618422
PPM1K	100	100	100	100	?Maple syrup urine disease, mild variant, 615135
PPP1R15B	100	99,6	100	100	Microcephaly, short stature, and impaired glucose metabolism 2, 616817
PPP2R3C	99,6	94,9	100	100	Gonadal dysgenesis, dysmorphic facies, retinal dystrophy, and myopathy, 618419 Spermatogenic failure 36, 618420
PPT1	90,3	90,3	82,5	82,5	Ceroid lipofuscinosis, neuronal, 1, 256730
PRCD	100	100	100	100	Retinitis pigmentosa 36, 610599
PRDM12	90,8	88	93,4	91,7	Neuropathy, hereditary sensory and autonomic, type VIII, 616488
PRDM5	99,9	99,2	100	100	Brittle cornea syndrome 2, 614170
PRDM8	92,9	88,6	100	99,8	?Epilepsy, progressive myoclonic, 10, 616640
PRDX1	100	100	100	100	Methylmalonic aciduria and homocystinuria, cblC type, digenic, 277400
PREPL	99,8	98,2	100	100	Myasthenic syndrome, congenital, 22, 616224

PRF1	91,2	90,8	100	100	Hemophagocytic lymphohistiocytosis, familial, 2, 603553 Aplastic anemia, 609135 Lymphoma, non-Hodgkin, 605027
PRG4	87,4	80,9	100	100	Camptodactyly-arthropathy-coxa vara-pericarditis syndrome, 208250
PRICKLE1	100	100	100	100	Epilepsy, progressive myoclonic 1B, 612437
PRKCD	100	100	100	100	Autoimmune lymphoproliferative syndrome, type III, 615559
PRKDC	99,7	98	100	100	Immunodeficiency 26, with or without neurologic abnormalities, 615966
PRKN	67	66,2	75,3	75,3	Parkinson disease, juvenile, type 2, 600116 Ovarian cancer, somatic, 167000 Adenocarcinoma of lung, somatic, 211980
PRKRA	100	99,4	100	100	Dystonia 16, 612067
PRMT7	100	99,9	100	100	Short stature, brachydactyly, intellectual developmental disability, and seizures, 617157
PROC	100	100	100	100	Thrombophilia due to protein C deficiency, autosomal dominant, 176860 Thrombophilia due to protein C deficiency, autosomal recessive, 612304
PRODH	85	80,6	100	100	Hyperprolinemia, type I, 239500 {Schizophrenia, susceptibility to, 4}, 600850
PROM1	97,2	96,1	100	100	Retinitis pigmentosa 41, 612095 Stargardt disease 4, 603786 Cone-rod dystrophy 12, 612657 Macular dystrophy, retinal, 2, 608051
PROP1	92,6	82,6	100	100	Pituitary hormone deficiency, combined, 2, 262600
PROS1	96,7	92,1	98,4	98,4	Thrombophilia due to protein S deficiency, autosomal recessive, 614514 Thrombophilia due to protein S deficiency, autosomal dominant, 612336
PRSS12	100	99,9	100	100	Mental retardation, autosomal recessive 1, 249500
PRSS56	99,9	96,5	100	100	Microphthalmia, isolated 6, 613517
PRUNE1	93,6	93,5	93,6	93,6	Neurodevelopmental disorder with microcephaly, hypotonia, and variable brain anomalies, 617481
PRX	96	95,5	96,5	96,1	Charcot-Marie-Tooth disease, type 4F, 614895 Dejerine-Sottas disease, 145900
PSAP	100	100	100	100	Gaucher disease, atypical, 610539 Krabbe disease, atypical, 611722 Combined SAP deficiency, 611721 Metachromatic leukodystrophy due to SAP-b deficiency, 249900
PSAT1	95,3	81,6	100	100	Neu-Laxova syndrome 2, 616038 ?Phosphoserine aminotransferase deficiency, 610992
PSMB4	100	100	100	100	?Proteasome-associated autoinflammatory syndrome 3 and digenic forms, 617591
PSMB8	99,9	98,5	100	100	Proteasome-associated autoinflammatory syndrome 1 and digenic forms, 256040
PSMB9	99,9	97,7	100	100	?Proteasome-associated autoinflammatory syndrome 3, digenic, 617591

PSMC3IP	100	100	100	100	Ovarian dysgenesis 3, 614324
PSPH	100	100	100	100	Phosphoserine phosphatase deficiency, 614023
PTF1A	95,8	85,6	98,6	93,3	Pancreatic and cerebellar agenesis, 609069 Pancreatic agenesis 2, 615935
PTH1R	100	98,7	100	100	Metaphyseal chondrodysplasia, Murk Jansen type, 156400 Failure of tooth eruption, primary, 125350 Eiken syndrome, 600002 Chondrodysplasia, Blomstrand type, 215045
PTPN14	99,7	97,4	100	100	Choanal atresia and lymphedema, 613611
PTPRC	99	95,1	100	100	{Hepatitis C virus, susceptibility to}, 609532 Severe combined immunodeficiency, T cell-negative, B-cell/natural killer-cell positive, 608971
PTPRO	99,9	99,4	100	100	Nephrotic syndrome, type 6, 614196
PTPRQ	94,6	92,5	92,8	92,7	Deafness, autosomal dominant 73, 617663 Deafness, autosomal recessive 84A, 613391
PTRH2	100	100	100	100	Infantile-onset multisystem neurologic, endocrine, and pancreatic disease, 616263
PTS	99,9	99,1	100	100	Hyperphenylalaninemia, BH4-deficient, A, 261640
PUS1	100	99,5	99,6	97,2	Myopathy, lactic acidosis, and sideroblastic anemia 1, 600462
PUS3	100	100	100	100	Neurodevelopmental disorder with microcephaly and gray sclerae, 617051
PUS7	100	99,8	100	100	Intellectual developmental disorder with abnormal behavior, microcephaly, and short stature, 618342
PXDN	100	99,6	100	100	Anterior segment dysgenesis 7, with sclerocornea, 269400
PYCR1	99,9	97,7	100	100	Cutis laxa, autosomal recessive, type IIIB, 614438 Cutis laxa, autosomal recessive, type IIB, 612940
PYCR2	100	99,1	100	100	Leukodystrophy, hypomyelinating, 10, 616420
PYGL	100	100	100	100	Glycogen storage disease VI, 232700
PYGM	100	99,9	100	100	McArdle disease, 232600
PYROXD1	95,2	83,9	100	100	Myopathy, myofibrillar, 8, 617258
QARS1	100	100	100	100	Microcephaly, progressive, seizures, and cerebral and cerebellar atrophy, 615760
QDPR	100	99,7	100	100	Hyperphenylalaninemia, BH4-deficient, C, 261630
RAB18	99,5	97,4	100	100	Warburg micro syndrome 3, 614222
RAB23	100	99,5	100	100	Carpenter syndrome, 201000
RAB27A	100	100	100	100	Griscelli syndrome, type 2, 607624
RAB28	99,7	96	100	100	Cone-rod dystrophy 18, 615374
RAB33B	85	85	100	100	Smith-McCort dysplasia 2, 615222
RAB3GAP1	99,4	98,9	99,4	99,4	Warburg micro syndrome 1, 600118
RAB3GAP2	99,5	97	100	100	Warburg micro syndrome 2, 614225 Martsolf syndrome, 212720
RAD50	97,5	91,6	100	100	Nijmegen breakage syndrome-like disorder, 613078

RAD51C	100	99,8	100	100	{Breast-ovarian cancer, familial, susceptibility to, 3}, 613399 Fanconi anemia, complementation group O, 613390
RAG1	100	100	100	100	Omenn syndrome, 603554 Severe combined immunodeficiency, B cell-negative, 601457 Alpha/beta T-cell lymphopenia with gamma/delta T-cell expansion, severe cytomegalovirus infection, and autoimmunity, 609889 Combined cellular and humoral immune defects with granulomas, 233650
RAG2	100	100	100	100	Severe combined immunodeficiency, B cell-negative, 601457 Combined cellular and humoral immune defects with granulomas, 233650 Omenn syndrome, 603554
RALGAPA1	74,5	63,9	100	100	Neurodevelopmental disorder with hypotonia, neonatal respiratory insufficiency, and thermodysregulation, 618797
RAPSN	100	99,7	100	100	Fetal akinesia deformation sequence 2, 618388 Myasthenic syndrome, congenital, 11, associated with acetylcholine receptor deficiency, 616326
RARB	100	100	100	100	Microphthalmia, syndromic 12, 615524
RARS1	94,2	91,6	94,4	94,3	Leukodystrophy, hypomyelinating, 9, 616140
RARS2	100	99,8	100	100	Pontocerebellar hypoplasia, type 6, 611523
RASGRP1	100	99,6	100	100	Immunodeficiency 64, 618534
RASGRP2	99,7	97,3	100	100	?Bleeding disorder, platelet-type, 18, 615888
RAX	96	87	100	98,4	Microphthalmia, isolated 3, 611038
RBBP8	100	99,7	100	100	Jawad syndrome, 251255 Seckel syndrome 2, 606744 Pancreatic carcinoma, somatic, 0
RBCK1	99,9	98,2	100	100	Polyglucosan body myopathy 1 with or without immunodeficiency, 615895
RBM28	100	100	100	100	?Alopecia, neurologic defects, and endocrinopathy syndrome, 612079
RBM8A	99,8	97,9	100	100	Thrombocytopenia-absent radius syndrome, 274000
RBP3	100	100	100	100	?Retinitis pigmentosa 66, 615233
RBP4	99,9	97,7	100	100	Microphthalmia, isolated, with coloboma 10, 616428 Retinal dystrophy, iris coloboma, and comedogenic acne syndrome, 615147
RCBTB1	99,9	99,5	100	100	Retinal dystrophy with or without extraocular anomalies, 617175
RD3	100	100	100	100	Leber congenital amaurosis 12, 610612
RDH11	100	99	100	100	?Retinal dystrophy, juvenile cataracts, and short stature syndrome, 616108
RDH12	100	98,6	100	100	Leber congenital amaurosis 13, 612712
RDH5	100	99,9	100	100	Fundus albipunctatus, 136880
RDX	89,1	71,5	100	100	Deafness, autosomal recessive 24, 611022

RECQL4	99,8	98,1	100	99,9	RAPADILINO syndrome, 266280 Baller-Gerold syndrome, 218600 Rothmund-Thomson syndrome, type 2, 268400
REEP2	99,9	98,6	100	100	?Spastic paraplegia 72, autosomal dominant, 615625 ?Spastic paraplegia 72, autosomal recessive, 615625
REEP6	100	100	91,5	87,4	Retinitis pigmentosa 77, 617304
RELB	98,8	88,7	100	100	?Immunodeficiency 53, 617585
RELN	100	99,8	100	100	{Epilepsy, familial temporal lobe, 7}, 616436 Lissencephaly 2 (Norman-Roberts type), 257320
REN	100	100	100	100	Renal tubular dysgenesis, 267430 Tubulointerstitial kidney disease, autosomal dominant, 4, 613092 [Hyperproreninemia], 0
REPS1	99,6	97,5	100	100	?Neurodegeneration with brain iron accumulation 7, 617916
RETREG1	98,8	95,1	100	100	Neuropathy, hereditary sensory and autonomic, type IIB, 613115
RFC1	99,9	98,9	100	100	Cerebellar ataxia, neuropathy, and vestibular areflexia syndrome, 614575
RFT1	99,8	99,6	100	100	Congenital disorder of glycosylation, type In, 612015
RFWD3	100	99,8	100	100	?Fanconi anemia, complementation group W, 617784
RFX5	99,7	98,1	100	100	Bare lymphocyte syndrome, type II, complementation group C, 209920 Bare lymphocyte syndrome, type II, complementation group E, 209920
RFX6	100	99,6	100	100	Mitchell-Riley syndrome, 615710
RFXANK	100	99,5	100	100	MHC class II deficiency, complementation group B, 209920
RFXAP	99,3	97	100	99,9	Bare lymphocyte syndrome, type II, complementation group D, 209920
RGR	99	98,2	99	99	Retinitis pigmentosa 44, 613769
RHO	100	100	100	100	Night blindness, congenital stationary, autosomal dominant 1, 610445 Retinitis punctata albescens, 136880 Retinitis pigmentosa 4, autosomal dominant or recessive, 613731
RIMS2	96,7	95,3	97,8	97,7	Cone-rod synaptic disorder syndrome, congenital nonprogressive, 618970
RIN2	100	100	100	100	Macrocephaly, alopecia, cutis laxa, and scoliosis, 613075
RINT1	99,9	98,6	100	100	Infantile liver failure syndrome 3, 618641
RIPK1	100	99	100	100	Immunodeficiency 57 with autoinflammation, 618108 Autoinflammation with episodic fever and lymphadenopathy, 618852
RIPK4	100	99,9	100	100	Popliteal pterygium syndrome, Bartsocas-Papas type, 263650 CHAND syndrome, 214350
RIPOR2	100	99,8	100	100	?Deafness, autosomal recessive 104, 616515
RIPPLY2	100	97,9	100	100	?Spondylocostal dysostosis 6, 616566
RLBP1	100	99,9	100	100	Retinitis punctata albescens, 136880 Bothnia retinal dystrophy, 607475

					Newfoundland rod-cone dystrophy, 607476 Fundus albipunctatus, 136880
RMND1	100	98,6	100	100	Combined oxidative phosphorylation deficiency 11, 614922
RMRP	NC	NC	NC	NC	Anauxetic dysplasia 1, 607095 Cartilage-hair hypoplasia, 250250 Metaphyseal dysplasia without hypotrichosis, 250460
RNASEH1	98,5	95,3	100	100	Progressive external ophthalmoplegia with mitochondrial DNA deletions, autosomal recessive 2, 616479
RNASEH2A	100	100	100	100	Aicardi-Goutieres syndrome 4, 610333
RNASEH2B	80,6	78,1	91	90,9	Aicardi-Goutieres syndrome 2, 610181
RNASEH2C	100	99,5	100	100	Aicardi-Goutieres syndrome 3, 610329
RNASET2	97,4	93,1	100	100	Leukoencephalopathy, cystic, without megalencephaly, 612951
RNF168	100	99,8	100	100	RIDDLE syndrome, 611943
RNF216	99,8	98,7	100	100	Cerebellar ataxia and hypogonadotropic hypogonadism, 212840
RNPC3	91,5	70,7	100	100	?Growth hormone deficiency, isolated, type V, 618160
ROBO3	98,9	96,1	100	100	Gaze palsy, familial horizontal, with progressive scoliosis, 1, 607313
ROGDI	98,4	95,2	99,9	99,1	Kohlschutter-Tonz syndrome, 226750
ROR1	97	96,8	99,9	99,3	?Deafness, autosomal recessive 108, 617654
ROR2	100	99,9	97	97	Brachydactyly, type B1, 113000 Robinow syndrome, autosomal recessive, 268310
RORC	100	100	100	100	Immunodeficiency 42, 616622
RP1	91,5	90,6	100	100	Retinitis pigmentosa 1, 180100
RPE65	99,8	97,8	100	100	Leber congenital amaurosis 2, 204100 Retinitis pigmentosa 87 with choroidal involvement, 618697 Retinitis pigmentosa 20, 613794
RPGRIP1	100	99,9	100	100	Cone-rod dystrophy 13, 608194 Leber congenital amaurosis 6, 613826
RPGRIP1L	96,7	95,7	100	99,5	?COACH syndrome 3, 619113 Meckel syndrome 5, 611561 Joubert syndrome 7, 611560
RPIA	98,6	94,9	100	100	Ribose 5-phosphate isomerase deficiency, 608611
RRM2B	100	99,7	100	100	Progressive external ophthalmoplegia with mitochondrial DNA deletions, autosomal dominant 5, 613077 Mitochondrial DNA depletion syndrome 8A (encephalomyopathic type with renal tubulopathy), 612075 Mitochondrial DNA depletion syndrome 8B (MNGIE type), 612075
RSPH1	100	100	100	100	Ciliary dyskinesia, primary, 24, 615481

RSPH3	99,9	98,8	100	100	Ciliary dyskinesia, primary, 32, 616481
RSPH4A	98,1	95,6	100	100	Ciliary dyskinesia, primary, 11, 612649
RSPH9	99,9	97,9	100	100	Ciliary dyskinesia, primary, 12, 612650
RSPO1	100	99,9	100	100	Palmoplantar hyperkeratosis with squamous cell carcinoma of skin and sex reversal, 610644 Palmoplantar hyperkeratosis and true hermaphroditism, 610644
RSPO2	97,1	90,7	100	100	Tetraamelia syndrome 2, 618021 ?Humerofemoral hypoplasia with radiotibial ray deficiency, 618022
RSPO4	100	100	100	100	Anonychia congenita, 206800
RSPRY1	100	100	100	100	Spondyloepimetaphyseal dysplasia, Faden-Alkuraya type, 616723
RSRC1	99,8	96,8	100	100	Intellectual developmental disorder, autosomal recessive 70, 618402
RTEL1	99,5	96,8	100	100	Dyskeratosis congenita, autosomal recessive 5, 615190 Pulmonary fibrosis and/or bone marrow failure, telomere-related, 3, 616373 Dyskeratosis congenita, autosomal dominant 4, 615190
RTN4IP1	99,9	98,7	100	100	Optic atrophy 10 with or without ataxia, mental retardation, and seizures, 616732
RTTN	98,9	98	100	100	Microcephaly, short stature, and polymicrogyria with seizures, 614833
RUBCN	99,4	97,5	100	100	Spinocerebellar ataxia, autosomal recessive 15, 615705
RUSC2	100	100	100	100	Mental retardation, autosomal recessive 61, 617773
RXYLT1	99,5	96,8	100	99,9	Muscular dystrophy-dystroglycanopathy (congenital with brain and eye anomalies), type A, 10, 615041
RYR1	96,9	93,9	99,4	99	Central core disease, 117000 King-Denborough syndrome, 145600 {Malignant hyperthermia susceptibility 1}, 145600 Minicore myopathy with external ophthalmoplegia, 255320 Neuromuscular disease, congenital, with uniform type 1 fiber, 117000
S1PR2	99,4	96,9	100	100	Deafness, autosomal recessive 68, 610419
SACS	100	99,9	100	100	Spastic ataxia, Charlevoix-Saguenay type, 270550
SAG	100	100	100	100	Oguchi disease-1, 258100 Retinitis pigmentosa 47, 613758
SALL2	100	100	100	100	?Coloboma, ocular, autosomal recessive, 216820
SAMD9	100	99,8	100	100	MIRAGE syndrome, 617053 Tumoral calcinosis, familial, normophosphatemic, 610455 Monosomy 7 myelodysplasia and leukemia syndrome 2, 619041
SAMHD1	98,7	98,4	100	100	?Chilblain lupus 2, 614415 Aicardi-Goutieres syndrome 5, 612952
SAR1B	97	89,7	100	100	Chylomicron retention disease, 246700
SARS1	100	99,3	100	100	?Neurodevelopmental disorder with microcephaly, ataxia, and seizures, 617709
SARS2	95,8	94,6	100	100	Hyperuricemia, pulmonary hypertension, renal failure, and alkalosis, 613845

SASH1	99,9	98,7	100	100	?Cancer, alopecia, pigment dyscrasia, onychodystrophy, and keratoderma, 618373 Dyschromatosis universalis hereditaria 1, 127500
SASS6	99,9	98,5	100	100	?Microcephaly 14, primary, autosomal recessive, 616402
SBDS	100	100	100	100	{Aplastic anemia, susceptibility to}, 609135 Shwachman-Diamond syndrome, 260400
SBF1	99	97,7	100	100	Charcot-Marie-Tooth disease, type 4B3, 615284
SBF2	99,9	99,4	100	100	Charcot-Marie-Tooth disease, type 4B2, 604563
SC5D	100	99,5	100	100	Lathosterolosis, 607330
SCAPER	99,7	98,2	100	100	Intellectual developmental disorder and retinitis pigmentosa, 618195
SCARB2	100	99,8	100	100	Epilepsy, progressive myoclonic 4, with or without renal failure, 254900
SCARF2	95,4	86,2	99,8	99,2	Van den Ende-Gupta syndrome, 600920
SCN1B	98	96,4	99,8	99,3	Atrial fibrillation, familial, 13, 615377 Developmental and epileptic encephalopathy 52, 617350 Cardiac conduction defect, nonspecific, 612838 Epilepsy, generalized, with febrile seizures plus, type 1, 604233 Brugada syndrome 5, 612838
SCN4A	100	99,6	100	100	Hyperkalemic periodic paralysis, type 2, 170500 Paramyotonia congenita, 168300 Myotonia congenita, atypical, acetazolamide-responsive, 608390 Myasthenic syndrome, congenital, 16, 614198 Hypokalemic periodic paralysis, type 2, 613345
SCN9A	99,3	97,9	100	100	Neuropathy, hereditary sensory and autonomic, type IID, 243000 Small fiber neuropathy, 133020 Paroxysmal extreme pain disorder, 167400 Insensitivity to pain, congenital, 243000 Erythermalgia, primary, 133020
SCNN1A	99,7	98,2	100	100	Pseudohypoaldosteronism, type I, 264350 ?Liddle syndrome 3, 618126 Bronchiectasis with or without elevated sweat chloride 2, 613021
SCNN1B	100	99,7	100	100	Bronchiectasis with or without elevated sweat chloride 1, 211400 Pseudohypoaldosteronism, type I, 264350 Liddle syndrome 1, 177200
SCNN1G	99,8	98,3	100	100	Bronchiectasis with or without elevated sweat chloride 3, 613071 Liddle syndrome 2, 618114 Pseudohypoaldosteronism, type I, 264350
SCO1	97,1	93,8	100	100	Mitochondrial complex IV deficiency, nuclear type 4, 619048
SCO2	100	100	100	100	Myopia 6, 608908 Mitochondrial complex IV deficiency, nuclear type 2, 604377

SCP2	100	99,2	100	100	?Leukoencephalopathy with dystonia and motor neuropathy, 613724
SCYL1	100	100	100	100	Spinocerebellar ataxia, autosomal recessive 21, 616719
SDCCAG8	100	99,9	100	100	Bardet-Biedl syndrome 16, 615993 Senior-Loken syndrome 7, 613615
SDHA	85,8	80,4	100	100	Cardiomyopathy, dilated, 1GG, 613642 Paragangliomas 5, 614165 Mitochondrial complex II deficiency, nuclear type 1, 252011
SDHAF1	99,9	93,2	100	100	Mitochondrial complex II deficiency, nuclear type 2, 619166
SDHD	54	51,6	80,1	80,1	Paragangliomas 1, with or without deafness, 168000 Mitochondrial complex II deficiency, nuclear type 3, 619167 Paraganglioma and gastric stromal sarcoma, 606864 Pheochromocytoma, 171300
SDR9C7	100	100	100	100	Ichthyosis, congenital, autosomal recessive 13, 617574
SEC23A	99,7	98,2	100	100	Craniolenticulosutural dysplasia, 607812
SEC23B	99,9	99,3	100	100	?Cowden syndrome 7, 616858 Dyserythropoietic anemia, congenital, type II, 224100
SEC24D	100	99,7	100	100	Cole-Carpenter syndrome 2, 616294
SEC31A	99,3	97,1	100	100	?Neurodevelopmental disorder with spastic quadriplegia, optic atrophy, seizures, and structural brain anomalies, 618651
SECISBP2	99,8	97,4	100	100	Thyroid hormone metabolism, abnormal, 609698
SELENON	84,5	84	87,7	85,1	Muscular dystrophy, rigid spine, 1, 602771 Myopathy, congenital, with fiber-type disproportion, 255310
SEMA4A	100	99,8	100	100	Cone-rod dystrophy 10, 610283 Retinitis pigmentosa 35, 610282
SEPSECS	100	100	100	100	Pontocerebellar hypoplasia type 2D, 613811
SERAC1	99,9	99,5	100	100	3-methylglutaconic aciduria with deafness, encephalopathy, and Leigh-like syndrome, 614739
SERPINA1	100	100	100	100	Hemorrhagic diathesis due to antithrombin Pittsburgh, 613490 Emphysema-cirrhosis, due to AAT deficiency, 613490 Emphysema due to AAT deficiency, 613490
SERPINA6	100	100	100	100	Corticosteroid-binding globulin deficiency, 611489
SERPINB6	93,4	93,4	100	100	?Deafness, autosomal recessive 91, 613453
SERPINB7	100	99,9	100	100	Palmoplantar keratoderma, Nagashima type, 615598
SERPINB8	95	95	100	100	Peeling skin syndrome 5, 617115
SERPINC1	100	100	100	100	Thrombophilia due to antithrombin III deficiency, 613118
SERPINE1	100	100	100	100	Plasminogen activator inhibitor-1 deficiency, 613329 {Transcription of plasminogen activator inhibitor, modulator of}, 0
SERPINF1	100	100	100	100	Osteogenesis imperfecta, type VI, 613982

SERPINF2	100	99,8	100	100	Alpha-2-plasmin inhibitor deficiency, 262850
SERPING1	99,7	97,5	100	100	Angioedema, hereditary, types I and II, 106100 Complement component 4, partial deficiency of, 120790
SERPINH1	100	98,3	100	100	Osteogenesis imperfecta, type X, 613848 {Preterm premature rupture of the membranes, susceptibility to}, 610504
SETX	100	99,8	100	100	Spinocerebellar ataxia, autosomal recessive, with axonal neuropathy 2, 606002 Amyotrophic lateral sclerosis 4, juvenile, 602433
SFRP4	100	99,8	100	100	Pyle disease, 265900
SFTPB	99,4	96,7	100	100	Surfactant metabolism dysfunction, pulmonary, 1, 265120
SFXN4	99,9	98,9	100	100	Combined oxidative phosphorylation deficiency 18, 615578
SGCA	100	99,9	100	100	Muscular dystrophy, limb-girdle, autosomal recessive 3, 608099
SGCB	97,7	96,5	100	100	Muscular dystrophy, limb-girdle, autosomal recessive 4, 604286
SGCD	100	98,9	100	100	Cardiomyopathy, dilated, 1L, 606685 Muscular dystrophy, limb-girdle, autosomal recessive 6, 601287
SGCG	100	99,2	100	100	Muscular dystrophy, limb-girdle, autosomal recessive 5, 253700
SGO1	99,9	98,9	100	100	Chronic atrial and intestinal dysrhythmia, 616201
SGPL1	100	100	100	100	Nephrotic syndrome, type 14, 617575
SGSH	94,4	94,1	100	100	Mucopolysaccharidosis type IIIA (Sanfilippo A), 252900
SH3PXD2B	100	100	100	100	Frank-ter Haar syndrome, 249420
SH3TC2	100	99,7	100	100	Charcot-Marie-Tooth disease, type 4C, 601596 Mononeuropathy of the median nerve, mild, 613353
SI	99,2	96,1	100	100	Sucrase-isomaltase deficiency, congenital, 222900
SIGMAR1	100	100	100	100	?Amyotrophic lateral sclerosis 16, juvenile, 614373 ?Spinal muscular atrophy, distal, autosomal recessive, 2, 605726
SIK3	99,8	98,7	99,3	98,1	?Spondyloepimetaphyseal dysplasia, Krakow type, 618162
SIL1	99,2	96,7	100	100	Marinesco-Sjogren syndrome, 248800
SIX6	100	100	100	100	Optic disc anomalies with retinal and/or macular dystrophy, 212550
SKIV2L	100	99,8	100	100	Trichohepatoenteric syndrome 2, 614602
SLC10A2	100	100	100	100	Bile acid malabsorption, primary, 613291
SLC10A7	99,7	98	100	100	Short stature, amelogenesis imperfecta, and skeletal dysplasia with scoliosis, 618363
SLC11A2	98,2	98	100	100	Anemia, hypochromic microcytic, with iron overload 1, 206100
SLC12A1	96,2	96,1	96,2	96,2	Bartter syndrome, type 1, 601678
SLC12A3	100	99,9	100	100	Gitelman syndrome, 263800
SLC12A5	83,9	83,8	97,4	97,4	Developmental and epileptic encephalopathy 34, 616645 {Epilepsy, idiopathic generalized, susceptibility to, 14}, 616685
SLC12A6	100	100	100	100	Agenesis of the corpus callosum with peripheral neuropathy, 218000
SLC13A3	99,4	97,5	100	100	Leukoencephalopathy, acute reversible, with increased urinary alpha-ketoglutarate, 618384

SLC13A5	100	99,9	100	100	Developmental and epileptic encephalopathy 25, with amelogenesis imperfecta, 615905
SLC16A1	100	99,3	100	100	Monocarboxylate transporter 1 deficiency, 616095 Hyperinsulinemic hypoglycemia, familial, 7, 610021 Erythrocyte lactate transporter defect, 245340
SLC17A5	99,6	97	100	100	Sialic acid storage disorder, infantile, 269920 Salla disease, 604369
SLC18A2	100	99,7	100	100	?Parkinsonism-dystonia, infantile, 2, 618049
SLC18A3	100	100	100	100	Myasthenic syndrome, congenital, 21, presynaptic, 617239
SLC19A2	100	99,7	100	100	Thiamine-responsive megaloblastic anemia syndrome, 249270
SLC19A3	97,8	97,6	98,7	98,7	Thiamine metabolism dysfunction syndrome 2 (biotin- or thiamine-responsive encephalopathy type 2), 607483
SLC1A1	99,9	99,6	100	100	{?Schizophrenia susceptibility 18}, 615232 Dicarboxylic aminoaciduria, 222730
SLC1A4	99	95,8	100	100	Spastic tetraplegia, thin corpus callosum, and progressive microcephaly, 616657
SLC22A12	100	99,8	100	100	Hypouricemia, renal, 220150
SLC22A5	100	100	100	100	Carnitine deficiency, systemic primary, 212140
SLC24A1	100	99,9	100	100	Night blindness, congenital stationary (complete), 1D, autosomal recessive, 613830
SLC24A4	100	99,8	100	100	[Skin/hair/eye pigmentation 6, blue/green eyes], 210750 Amelogenesis imperfecta, type IIA5, 615887 [Skin/hair/eye pigmentation 6, blond/brown hair], 210750
SLC24A5	99,9	99,1	100	100	[Skin/hair/eye pigmentation 4, fair/dark skin], 113750 Albinism, oculocutaneous, type VI, 113750
SLC25A1	95,8	88,6	99,5	97,8	Myasthenic syndrome, congenital, 23, presynaptic, 618197 Combined D-2- and L-2-hydroxyglutaric aciduria, 615182
SLC25A12	99,9	99,5	100	100	Developmental and epileptic encephalopathy 39, 612949
SLC25A13	100	99,7	100	100	Citrullinemia, adult-onset type II, 603471 Citrullinemia, type II, neonatal-onset, 605814
SLC25A15	99,8	98,1	100	100	Hyperornithinemia-hyperammonemia-homocitrullinemia syndrome, 238970
SLC25A19	100	98,5	100	100	Thiamine metabolism dysfunction syndrome 4 (progressive polyneuropathy type), 613710 Microcephaly, Amish type, 607196
SLC25A20	100	100	100	100	Carnitine-acylcarnitine translocase deficiency, 212138
SLC25A22	98,6	95,8	100	100	Developmental and epileptic encephalopathy 3, 609304
SLC25A26	100	99,5	100	100	Combined oxidative phosphorylation deficiency 28, 616794
SLC25A3	99,8	98	100	100	Mitochondrial phosphate carrier deficiency, 610773
SLC25A38	97,9	95,3	100	100	Anemia, sideroblastic, 2, pyridoxine-refractory, 205950
SLC25A4	100	100	100	100	Mitochondrial DNA depletion syndrome 12B (cardiomyopathic type) AR, 615418 Mitochondrial DNA depletion syndrome 12A (cardiomyopathic type) AD, 617184

					Progressive external ophthalmoplegia with mitochondrial DNA deletions, autosomal dominant 2, 609283
SLC25A42	96,5	93,2	100	100	Metabolic crises, recurrent, with variable encephalomyopathic features and neurologic regression, 618416
SLC25A46	99,7	97,3	100	100	Neuropathy, hereditary motor and sensory, type VIB, 616505
SLC26A1	100	99,6	100	100	?Nephrolithiasis, calcium oxalate, 167030
SLC26A2	100	100	100	100	De la Chapelle dysplasia, 256050 Atelosteogenesis, type II, 256050 Diastrophic dysplasia, broad bone-platyspondylic variant, 222600 Diastrophic dysplasia, 222600 Achondrogenesis Ib, 600972 Epiphyseal dysplasia, multiple, 4, 226900
SLC26A3	100	99,5	100	100	Diarrhea 1, secretory chloride, congenital, 214700
SLC26A4	100	99,7	100	100	Pendred syndrome, 274600 Deafness, autosomal recessive 4, with enlarged vestibular aqueduct, 600791
SLC26A5	99,1	96,8	100	100	?Deafness, autosomal recessive 61, 613865
SLC27A4	100	99,8	100	100	Ichthyosis prematurity syndrome, 608649
SLC29A3	100	99,6	100	100	Histiocytosis-lymphadenopathy plus syndrome, 602782
SLC2A1	92,8	92,8	100	100	Dystonia 9, 601042 GLUT1 deficiency syndrome 1, infantile onset, severe, 606777 Stomatin-deficient cryohydrocytosis with neurologic defects, 608885 GLUT1 deficiency syndrome 2, childhood onset, 612126 {Epilepsy, idiopathic generalized, susceptibility to, 12}, 614847
SLC2A10	97,7	97,7	100	100	Arterial tortuosity syndrome, 208050
SLC2A2	100	100	100	100	Fanconi-Bickel syndrome, 227810 {Diabetes mellitus, noninsulin-dependent}, 125853
SLC2A9	99,8	96,1	100	100	{Uric acid concentration, serum, QTL 2}, 612076 Hypouricemia, renal, 2, 612076
SLC30A10	100	100	100	100	Hypermanganesemia with dystonia 1, 613280
SLC30A9	98,8	94,2	100	100	?Birk-Landau-Perez syndrome, 617595
SLC33A1	99,9	98,9	100	100	Spastic paraplegia 42, autosomal dominant, 612539 Congenital cataracts, hearing loss, and neurodegeneration, 614482
SLC34A1	99,9	99,1	100	100	Nephrolithiasis/osteoporosis, hypophosphatemic, 1, 612286 Hypercalcemia, infantile, 2, 616963 ?Fanconi renotubular syndrome 2, 613388
SLC34A2	100	100	100	100	Pulmonary alveolar microlithiasis, 265100
SLC34A3	100	99,4	100	100	Hypophosphatemic rickets with hypercalciuria, 241530
SLC35A1	100	99,7	100	100	Congenital disorder of glycosylation, type IIf, 603585

SLC35A3	80,7	78,6	81,1	81	?Arthrogryposis, mental retardation, and seizures, 615553
SLC35C1	99,9	98,7	100	100	Congenital disorder of glycosylation, type IIc, 266265
SLC35D1	100	97,7	100	100	Schneckenbecken dysplasia, 269250
SLC37A4	100	99,2	100	100	Glycogen storage disease Ic, 232240 Glycogen storage disease Ib, 232220
SLC38A8	99,9	97,3	100	100	Foveal hypoplasia 2, with or without optic nerve misrouting and/or anterior segment dysgenesis, 609218
SLC39A13	99,8	98,2	100	100	Ehlers-Danlos syndrome, spondylodysplastic type, 3, 612350
SLC39A14	100	99,4	93,5	93,5	?Hyperostosis cranalis interna, 144755 Hypermanganesemia with dystonia 2, 617013
SLC39A4	99,5	95,5	100	100	Acrodermatitis enteropathica, 201100
SLC39A8	100	99,7	100	100	Congenital disorder of glycosylation, type IIn, 616721
SLC3A1	100	99,8	96,6	96,6	Cystinuria, 220100
SLC45A1	100	99,6	100	100	Intellectual developmental disorder with neuropsychiatric features, 617532
SLC45A2	100	99,9	100	100	[Skin/hair/eye pigmentation 5, dark/fair skin], 227240 [Skin/hair/eye pigmentation 5, black/nonblack hair], 227240 Albinism, oculocutaneous, type IV, 606574 [Skin/hair/eye pigmentation 5, dark/light eyes], 227240
SLC46A1	99,9	98,5	100	100	Folate malabsorption, hereditary, 229050
SLC4A1	100	99,8	96,1	96,1	[Blood group, Swann], 601550 [Blood group, Froese], 601551 [Blood group, Waldner], 112010 Spherocytosis, type 4, 612653 Cryohydrocytosis, 185020 Distal renal tubular acidosis 4 with hemolytic anemia, 611590 Ovalocytosis, SA type, 166900 [Malaria, resistance to], 611162 [Blood group, Diego], 110500 Distal renal tubular acidosis 1, 179800 [Blood group, Wright], 112050
SLC4A11	100	99,9	100	100	Corneal endothelial dystrophy, autosomal recessive, 217700 Corneal dystrophy, Fuchs endothelial, 4, 613268 Corneal endothelial dystrophy and perceptive deafness, 217400
SLC4A4	99,8	99,2	100	100	Renal tubular acidosis, proximal, with ocular abnormalities, 604278
SLC52A2	100	100	100	100	Brown-Vialetto-Van Laere syndrome 2, 614707
SLC52A3	100	100	100	100	Brown-Vialetto-Van Laere syndrome 1, 211530 ?Fazio-Londe disease, 211500
SLC5A1	100	100	100	100	Glucose/galactose malabsorption, 606824

SLC5A2	100	100	100	100	Renal glucosuria, 233100
SLC5A5	100	99,8	100	100	Thyroid dyshormonogenesis 1, 274400
SLC5A7	100	99,9	100	100	Neuronopathy, distal hereditary motor, type VIIA, 158580 Myasthenic syndrome, congenital, 20, presynaptic, 617143
SLC6A17	100	100	100	100	Mental retardation, autosomal recessive 48, 616269
SLC6A19	100	100	100	100	Hartnup disorder, 234500 Hyperglycinuria, 138500 Iminoglycinuria, digenic, 242600
SLC6A3	100	100	100	100	{Nicotine dependence, protection against}, 188890 Parkinsonism-dystonia, infantile, 1, 613135
SLC6A5	100	100	100	100	Hyperekplexia 3, 614618
SLC6A9	100	100	100	100	Glycine encephalopathy with normal serum glycine, 617301
SLC7A14	100	100	100	100	Retinitis pigmentosa 68, 615725
SLC7A7	100	99,9	100	100	Lysinuric protein intolerance, 222700
SLC7A9	100	99,9	100	100	Cystinuria, 220100
SLC9A1	100	100	100	100	Lichtenstein-Knorr syndrome, 616291
SLC9A3	90,6	86	96,4	94,1	Diarrhea 8, secretory sodium, congenital, 616868
SLCO2A1	100	99,4	100	100	Hypertrophic osteoarthropathy, primary, autosomal recessive 2, 614441
SLITRK6	100	100	100	100	Deafness and myopia, 221200
SLURP1	100	99,3	100	100	Meleda disease, 248300
SLX4	100	99,8	100	100	Fanconi anemia, complementation group P, 613951
SMARCAL1	100	99,9	100	100	Schimke immunoosseous dysplasia, 242900
SMARCD2	87	85,9	99,6	97	Specific granule deficiency 2, 617475
SMG9	100	100	100	100	Heart and brain malformation syndrome, 616920
SMN1	99,5	94,7	94,6	94,6	Spinal muscular atrophy-2, 253550 Spinal muscular atrophy-3, 253400 Spinal muscular atrophy-1, 253300 Spinal muscular atrophy-4, 271150
SMOC1	99,9	98,4	100	100	Microphthalmia with limb anomalies, 206920
SMOC2	76,8	76,6	100	100	Dentin dysplasia, type I, with microdontia and misshapen teeth, 125400
SMPD1	100	100	100	100	Niemann-Pick disease, type A, 257200 Niemann-Pick disease, type B, 607616
SMPD4	99,4	94,2	100	100	Neurodevelopmental disorder with microcephaly, arthrogryposis, and structural brain anomalies, 618622
SNAI2	100	99,1	100	100	Waardenburg syndrome, type 2D, 608890 Piebaldism, 172800
SNAP29	100	100	100	100	Cerebral dysgenesis, neuropathy, ichthyosis, and palmoplantar keratoderma syndrome, 609528

SNIP1	98,9	97,1	100	100	Psychomotor retardation, epilepsy, and craniofacial dysmorphism, 614501
SNORD118	NC	NC	NC	NC	Leukoencephalopathy, brain calcifications, and cysts, 614561
SNX10	96,2	95,7	100	99,6	Osteopetrosis, autosomal recessive 8, 615085
SNX14	99,6	95,9	100	100	Spinocerebellar ataxia, autosomal recessive 20, 616354
SOBP	97,5	92,9	97	95,3	Mental retardation, anterior maxillary protrusion, and strabismus, 613671
SOD1	100	99,9	100	100	Amyotrophic lateral sclerosis 1, 105400 Spastic tetraplegia and axial hypotonia, progressive, 618598
SOST	100	99,5	100	100	Sclerosteosis 1, 269500 Craniodiaphyseal dysplasia, autosomal dominant, 122860
SOX18	70,7	55,2	96,1	92,6	Hypotrichosis-lymphedema-telangiectasia-renal defect syndrome, 137940 Hypotrichosis-lymphedema-telangiectasia syndrome, 607823
SP110	100	100	100	100	{Mycobacterium tuberculosis, susceptibility to}, 607948 Hepatic venoocclusive disease with immunodeficiency, 235550
SP7	100	99,8	100	100	Osteogenesis imperfecta, type XII, 613849
SPAG1	99,3	95,8	99,9	98,6	Ciliary dyskinesia, primary, 28, 615505
SPARC	100	100	100	100	Osteogenesis imperfecta, type XVII, 616507
SPART	99,7	96,8	100	100	Troyer syndrome, 275900
SPATA5	100	99,7	100	100	Epilepsy, hearing loss, and mental retardation syndrome, 616577
SPATA7	99,8	98,2	100	100	Retinitis pigmentosa, juvenile, autosomal recessive, 604232 Leber congenital amaurosis 3, 604232
SPEG	96,1	88,7	99,7	99,7	Centronuclear myopathy 5, 615959
SPG11	100	99,3	100	100	Charcot-Marie-Tooth disease, axonal, type 2X, 616668 Spastic paraplegia 11, autosomal recessive, 604360 Amyotrophic lateral sclerosis 5, juvenile, 602099
SPG21	99,4	96,8	100	100	Mast syndrome, 248900
SPG7	88,2	86,2	100	100	Spastic paraplegia 7, autosomal recessive, 607259
SPINK5	99,9	99,5	100	100	Netherton syndrome, 256500
SPINT2	98,5	83,8	100	100	Diarrhea 3, secretory sodium, congenital, syndromic, 270420
SPNS2	92,1	89,3	97,6	95,7	?Deafness, autosomal recessive 115, 618457
SPR	99,8	96,3	100	100	Dystonia, dopa-responsive, due to sepiapterin reductase deficiency, 612716
SPRTN	100	100	100	100	Ruijs-Aalfs syndrome, 616200
SPTA1	99,9	99,2	100	100	Pyropoikilocytosis, 266140 Elliptocytosis-2, 130600 Spherocytosis, type 3, 270970
SPTB	100	100	100	100	Elliptocytosis-3, 617948 Spherocytosis, type 2, 616649 Anemia, neonatal hemolytic, fatal or near-fatal, 617948

SPTBN2	100	99,3	99,9	99,9	Spinocerebellar ataxia, autosomal recessive 14, 615386 Spinocerebellar ataxia 5, 600224
SPTBN4	97,3	91	100	100	Neurodevelopmental disorder with hypotonia, neuropathy, and deafness, 617519
SQSTM1	98,8	95,5	100	100	Frontotemporal dementia and/or amyotrophic lateral sclerosis 3, 616437 Neurodegeneration with ataxia, dystonia, and gaze palsy, childhood-onset, 617145 Myopathy, distal, with rimmed vacuoles, 617158 Paget disease of bone 3, 167250
SRD5A2	99,9	99	100	100	Pseudovaginal perineoscrotal hypospadias, 264600
SRD5A3	99,9	99,1	100	100	Kahrizi syndrome, 612713 Congenital disorder of glycosylation, type Iq, 612379
ST14	99,9	98,6	100	100	Ichthyosis, congenital, autosomal recessive 11, 602400
ST3GAL3	68,8	68,6	95,3	95,2	Developmental and epileptic encephalopathy 15, 615006 Intellectual developmental disorder, autosomal recessive 12, 611090
ST3GAL5	85	84,2	98,7	98,4	Salt and pepper developmental regression syndrome, 609056
STAC3	100	100	100	100	Myopathy, congenital, Baily-Bloch, 255995
STAMBP	100	99,4	100	100	Microcephaly-capillary malformation syndrome, 614261
STAR	100	100	100	100	Lipoid adrenal hyperplasia, 201710
STAT1	93,7	91,7	95,7	94,8	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	99,9	100	100	Immunodeficiency 44, 616636 Pseudo-TORCH syndrome 3, 618886
STAT5B	100	98,5	100	100	Growth hormone insensitivity with immune dysregulation 2, autosomal dominant, 618985 Growth hormone insensitivity with immune dysregulation 1, autosomal recessive, 245590 Leukemia, acute promyelocytic, somatic, 102578
STIL	100	99,8	100	100	Microcephaly 7, primary, autosomal recessive, 612703
STIM1	99,8	98	100	100	Myopathy, tubular aggregate, 1, 160565 Immunodeficiency 10, 612783 Stormorken syndrome, 185070
STK4	100	99,8	100	100	T-cell immunodeficiency, recurrent infections, autoimmunity, and cardiac malformations, 614868
STN1	100	100	100	100	Cerebroretinal microangiopathy with calcifications and cysts 2, 617341
STRA6	100	99,8	100	100	Microphthalmia, syndromic 9, 601186 Microphthalmia, isolated, with coloboma 8, 601186
STRADA	100	98,9	100	100	Polyhydramnios, megalencephaly, and symptomatic epilepsy, 611087
STRC	99,9	98	100	100	Deafness, autosomal recessive 16, 603720
STT3A	100	100	100	100	Congenital disorder of glycosylation, type Iw, 615596
STT3B	100	99,6	100	100	?Congenital disorder of glycosylation, type Ix, 615597

STUB1	100	98,7	100	100	Spinocerebellar ataxia, autosomal recessive 16, 615768 ?Spinocerebellar ataxia 48, 618093
STX11	100	100	100	100	Hemophagocytic lymphohistiocytosis, familial, 4, 603552
STXBP2	82,1	79,7	99,3	97,1	Hemophagocytic lymphohistiocytosis, familial, 5, 613101
SUCLA2	89,5	82,2	99,9	99,8	Mitochondrial DNA depletion syndrome 5 (encephalomyopathic with or without methylmalonic aciduria), 612073
SUCLG1	99,9	99,8	100	100	Mitochondrial DNA depletion syndrome 9 (encephalomyopathic type with methylmalonic aciduria), 245400
SUFU	100	100	100	100	Basal cell nevus syndrome, 109400 Medulloblastoma, desmoplastic, 155255 {Meningioma, familial, susceptibility to}, 607174 Joubert syndrome 32, 617757
SULT2B1	100	100	100	100	Ichthyosis, congenital, autosomal recessive 14, 617571
SUMF1	97,5	90,8	100	100	Multiple sulfatase deficiency, 272200
SUOX	100	100	100	100	Sulfite oxidase deficiency, 272300
SURF1	89,4	88,2	100	100	Charcot-Marie-Tooth disease, type 4K, 616684 Mitochondrial complex IV deficiency, nuclear type 1, 220110
SVBP	100	100	100	100	Neurodevelopmental disorder with ataxia, hypotonia, and microcephaly, 618569
SYNE1	98,2	97,8	98,8	98,8	Arthrogryposis multiplex congenita 3, myogenic type, 618484 Spinocerebellar ataxia, autosomal recessive 8, 610743 Emery-Dreifuss muscular dystrophy 4, autosomal dominant, 612998
SYNE4	99,7	97	100	100	Deafness, autosomal recessive 76, 615540
SYNJ1	99,9	99,4	100	100	Developmental and epileptic encephalopathy 53, 617389 Parkinson disease 20, early-onset, 615530
SYT14	61	60,5	100	100	?Spinocerebellar ataxia, autosomal recessive 11, 614229
SZT2	99,6	99,5	100	99,9	Developmental and epileptic encephalopathy 18, 615476
TAC3	100	99,6	100	100	Hypogonadotropic hypogonadism 10 with or without anosmia, 614839
TACO1	98,4	93	100	100	Mitochondrial complex IV deficiency, nuclear type 8, 619052
TACR3	100	100	100	100	Hypogonadotropic hypogonadism 11 with or without anosmia, 614840
TACSTD2	99	96,4	100	100	Corneal dystrophy, gelatinous drop-like, 204870
TAF13	100	100	100	100	Mental retardation, autosomal recessive 60, 617432
TAF1C	100	100	100	100	No OMIM disease ID
TAF2	99,9	98,6	100	100	Mental retardation, autosomal recessive 40, 615599
TAF6	99,8	98,9	100	100	Alazami-Yuan syndrome, 617126
TALDO1	100	97,9	100	100	Transaldolase deficiency, 606003
TANGO2	100	99,3	100	100	Metabolic encephalomyopathic crises, recurrent, with rhabdomyolysis, cardiac arrhythmias, and neurodegeneration, 616878

TAP1	100	99,2	100	100	Bare lymphocyte syndrome, type I, 604571
TAP2	99,9	99,3	100	100	Bare lymphocyte syndrome, type I, due to TAP2 deficiency, 604571
TAPBP	96,5	95,5	96,6	96,6	Bare lymphocyte syndrome, type I, 604571
TAPT1	91,7	86,9	98,5	94,8	Osteochondrodysplasia, complex lethal, Symoens-Barnes-Gistelinck type, 616897
TARS2	100	99,3	100	100	?Combined oxidative phosphorylation deficiency 21, 615918
TAT	100	100	100	100	Tyrosinemia, type II, 276600
TBC1D20	94,2	94,2	100	99,9	Warburg micro syndrome 4, 615663
TBC1D23	99,7	97,2	100	100	Pontocerebellar hypoplasia, type 11, 617695
TBC1D24	100	100	100	100	Developmental and epileptic encephalopathy 16, 615338 Epilepsy, rolandic, with proxysmal exercise-induce dystonia and writer's cramp, 608105 DOORS syndrome, 220500 Deafness, autosomal dominant 65, 616044 Myoclonic epilepsy, infantile, familial, 605021 Deafness, autosomal recessive 86, 614617
TBC1D7	100	99,3	100	100	Macrocephaly/megalencephaly syndrome, autosomal recessive, 248000
TBCD	96,2	94,4	100	100	Encephalopathy, progressive, early-onset, with brain atrophy and thin corpus callosum, 617193
TBCE	99,8	97,5	100	100	Kenny-Caffey syndrome, type 1, 244460 Hypoparathyroidism-retardation-dysmorphism syndrome, 241410 Encephalopathy, progressive, with amyotrophy and optic atrophy, 617207
TBCK	99,1	96,8	100	100	Hypotonia, infantile, with psychomotor retardation and characteristic facies 3, 616900
TBX15	100	99,9	100	100	Cousin syndrome, 260660
TBX19	100	100	100	100	Adrenocorticotropic hormone deficiency, 201400
TBX6	99,5	95,5	100	100	Spondylocostal dysostosis 5, 122600
TBXAS1	100	100	100	100	Ghosal hematodiaphyseal syndrome, 231095
TBXT	99,4	96,9	100	100	{Neural tube defects, susceptibility to}, 182940 Sacral agenesis with vertebral anomalies, 615709
TCAP	100	100	100	100	Cardiomyopathy, hypertrophic, 25, 607487 Muscular dystrophy, limb-girdle, autosomal recessive 7, 601954
TCIRG1	97,6	90,1	100	100	Osteopetrosis, autosomal recessive 1, 259700
TCN2	100	100	100	100	Transcobalamin II deficiency, 275350
TCTEX1D2	100	100	100	100	Short-rib thoracic dysplasia 17 with or without polydactyly, 617405
TCTN1	96,7	93	94,7	94,7	Joubert syndrome 13, 614173
TCTN2	100	99,5	100	100	Joubert syndrome 24, 616654 ?Meckel syndrome 8, 613885
TCTN3	100	100	100	100	Orofaciodigital syndrome IV, 258860 Joubert syndrome 18, 614815
TDP1	99,9	99,5	100	100	?Spinocerebellar ataxia, autosomal recessive, with axonal neuropathy 1, 607250

TDP2	100	99,4	100	100	Spinocerebellar ataxia, autosomal recessive 23, 616949
TDRD7	99,9	99,1	100	100	Cataract 36, 613887
TECPR2	100	100	100	100	Spastic paraplegia 49, autosomal recessive, 615031
TECR	100	99	100	100	Mental retardation, autosomal recessive 14, 614020
TECRL	96,3	89,3	100	100	Ventricular tachycardia, catecholaminergic polymorphic, 3, 614021
TECTA	100	99,9	100	100	Deafness, autosomal dominant 8/12, 601543 Deafness, autosomal recessive 21, 603629
TELO2	99,7	96,2	100	100	You-Hoover-Fong syndrome, 616954
TENM3	100	99,7	100	100	Microphthalmia, syndromic 15, 615145 ?Microphthalmia, isolated, with coloboma 9, 615145
TENT5A	100	99,7	100	100	Osteogenesis imperfecta, type XVIII, 617952
TF	100	100	100	100	Atransferrinemia, 209300
TFAM	97,5	83,5	100	100	?Mitochondrial DNA depletion syndrome 15 (hepatocerebral type), 617156
TFG	96,9	96,3	100	100	?Spastic paraplegia 57, autosomal recessive, 615658 Hereditary motor and sensory neuropathy, Okinawa type, 604484
TFR2	99,1	97,8	100	100	Hemochromatosis, type 3, 604250
TFRC	100	99,8	100	100	Immunodeficiency 46, 616740
TG	100	99,4	100	100	{Autoimmune thyroid disease, susceptibility to, 3}, 608175 Thyroid dyshormonogenesis 3, 274700
TGDS	99,4	96,8	100	100	Catel-Manzke syndrome, 616145
TGFB1	100	99,9	100	100	Camurati-Engelmann disease, 131300 {Cystic fibrosis lung disease, modifier of}, 219700 Inflammatory bowel disease, immunodeficiency, and encephalopathy, 618213
TGM1	100	99,9	100	100	Ichthyosis, congenital, autosomal recessive 1, 242300
TGM5	100	99,7	100	100	Peeling skin syndrome 2, 609796
TH	99,3	96,1	100	100	Segawa syndrome, recessive, 605407
THOC6	100	100	100	100	Beaulieu-Boycott-Innes syndrome, 613680
THRB	100	99,7	100	100	Thyroid hormone resistance, 188570 Thyroid hormone resistance, selective pituitary, 145650 Thyroid hormone resistance, autosomal recessive, 274300
TIMM50	98,3	94,4	100	100	3-methylglutaconic aciduria, type IX, 617698
TIMMDC1	100	100	100	100	Mitochondrial complex I deficiency, nuclear type 31, 618251
TJP2	92,8	92,5	98,8	98,8	Hypercholanemia, familial, 607748 Cholestasis, progressive familial intrahepatic 4, 615878
TK2	99,2	96,3	100	100	Mitochondrial DNA depletion syndrome 2 (myopathic type), 609560 ?Progressive external ophthalmoplegia with mitochondrial DNA deletions, autosomal recessive 3, 617069

TKT	98,7	97,8	98,7	98,7	Short stature, developmental delay, and congenital heart defects, 617044
TLE6	100	98,8	100	100	Preimplantation embryonic lethality, 616814
TMC1	99,7	97,3	100	100	Deafness, autosomal recessive 7, 600974 Deafness, autosomal dominant 36, 606705
TMC6	100	99,3	100	100	Epidermodysplasia verruciformis, 226400
TMC8	100	98,7	100	100	Epidermodysplasia verruciformis 2, 618231
TMCO1	88	87,4	88	88	Craniofacial dysmorphism, skeletal anomalies, and mental retardation syndrome, 213980
TMEM107	100	100	100	100	Orofaciodigital syndrome XVI, 617563 Meckel syndrome 13, 617562 ?Joubert syndrome 29, 617562
TMEM126A	96,3	84,4	100	100	Optic atrophy 7, 612989
TMEM126B	99,8	97,4	100	100	Mitochondrial complex I deficiency, nuclear type 29, 618250
TMEM132E	96,9	93,5	100	100	Deafness, autosomal recessive 99, 618481
TMEM138	100	99,1	100	100	Joubert syndrome 16, 614465
TMEM165	100	100	100	100	Congenital disorder of glycosylation, type IIk, 614727
TMEM199	100	99,9	100	100	Congenital disorder of glycosylation, type IIp, 616829
TMEM216	99,9	98,1	100	100	Meckel syndrome 2, 603194 Joubert syndrome 2, 608091
TMEM231	100	99,6	100	100	Meckel syndrome 11, 615397 Joubert syndrome 20, 614970
TMEM237	100	99,9	100	100	Joubert syndrome 14, 614424
TMEM260	97,5	93,4	100	100	Structural heart defects and renal anomalies syndrome, 617478
TMEM38B	100	99,9	100	100	Osteogenesis imperfecta, type XIV, 615066
TMEM67	99,5	95	100	99,9	Meckel syndrome 3, 607361 COACH syndrome 1, 216360 ?RHYNS syndrome, 602152 Nephronophthisis 11, 613550 {Bardet-Biedl syndrome 14, modifier of}, 615991 Joubert syndrome 6, 610688
TMEM70	98	93,9	100	100	Mitochondrial complex V (ATP synthase) deficiency, nuclear type 2, 614052
TMEM94	100	100	100	100	Intellectual developmental disorder with cardiac defects and dysmorphic facies, 618316
TMIE	99,2	95,1	100	100	Deafness, autosomal recessive 6, 600971
TMPRSS15	98,5	95,2	100	100	Enterokinase deficiency, 226200
TMPRSS3	100	99,9	100	100	Deafness, autosomal recessive 8/10, 601072
TMPRSS6	99,9	99,1	100	100	Iron-refractory iron deficiency anemia, 206200
TMTC3	99,6	96,5	100	100	Lissencephaly 8, 617255

TNFRSF11A	94,6	93,3	99,2	98	Osteolysis, familial expansile, 174810 {Paget disease of bone 2, early-onset}, 602080 Osteopetrosis, autosomal recessive 7, 612301
TNFRSF11B	100	100	100	100	Paget disease of bone 5, juvenile-onset, 239000
TNFRSF13B	100	100	100	100	Immunoglobulin A deficiency 2, 609529 Immunodeficiency, common variable, 2, 240500
TNFRSF13C	80,1	75,4	100	99,9	Immunodeficiency, common variable, 4, 613494
TNFRSF4	99,4	95,4	100	100	?Immunodeficiency 16, 615593
TNFSF11	100	99,9	100	100	Osteopetrosis, autosomal recessive 2, 259710
TNIK	100	99,3	100	100	Mental retardation, autosomal recessive 54, 617028
TNNI3	99,7	95,4	100	100	Cardiomyopathy, hypertrophic, 7, 613690 ?Cardiomyopathy, dilated, 2A, 611880 Cardiomyopathy, dilated, 1FF, 613286 Cardiomyopathy, familial restrictive, 1, 115210
TNNT1	99,9	97,6	100	100	Nemaline myopathy 5, Amish type, 605355
TNXB	99,1	93,7	100	99,9	Ehlers-Danlos syndrome, classic-like, 1, 606408 Vesicoureteral reflux 8, 615963
TOE1	100	100	100	100	Pontocerebellar hypoplasia, type 7, 614969
TONSL	99,8	97,8	100	100	Spondyloepimetaphyseal dysplasia, sponastrime type, 271510
TOP3A	100	98,7	100	100	Microcephaly, growth restriction, and increased sister chromatid exchange 2, 618097 ?Progressive external ophthalmoplegia with mitochondrial DNA deletions, autosomal recessive 5, 618098
TOR1AIP1	99,9	98	100	100	?Muscular dystrophy, autosomal recessive, with rigid spine and distal joint contractures, 617072
TP53RK	92,5	79,6	100	100	Galloway-Mowat syndrome 4, 617730
TPI1	99,8	97,5	100	100	Hemolytic anemia due to triosephosphate isomerase deficiency, 615512
TPK1	99,8	99	100	100	Thiamine metabolism dysfunction syndrome 5 (episodic encephalopathy type), 614458
TPM3	89,2	87,2	100	100	CAP myopathy 1, 609284 Nemaline myopathy 1, autosomal dominant or recessive, 609284 Myopathy, congenital, with fiber-type disproportion, 255310
TPO	99,9	98,2	100	100	Thyroid dyshormonogenesis 2A, 274500
TPP1	100	100	100	100	Spinocerebellar ataxia, autosomal recessive 7, 609270 Ceroid lipofuscinosis, neuronal, 2, 204500
TPRKB	81,1	75,9	81,9	81,9	Galloway-Mowat syndrome 5, 617731
TPRN	87,9	79,3	94,4	89,8	Deafness, autosomal recessive 79, 613307
TRAC	100	100	100	100	Immunodeficiency 7, TCR-alpha/beta deficient, 615387
TRAF3IP1	99,6	97,6	100	100	Senior-Loken syndrome 9, 616629
TRAIP	100	100	100	100	Seckel syndrome 9, 616777

TRAK1	93,3	92,9	100	99,9	Developmental and epileptic encephalopathy 68, 618201
TRAPPC11	100	99,2	100	100	Muscular dystrophy, limb-girdle, autosomal recessive 18, 615356
TRAPPC12	100	99,6	100	100	Encephalopathy, progressive, early-onset, with brain atrophy and spasticity, 617669
TRAPPC2L	100	100	100	100	Encephalopathy, progressive, early-onset, with episodic rhabdomyolysis, 618331
TRAPPC6B	99,9	98	100	100	Neurodevelopmental disorder with microcephaly, epilepsy, and brain atrophy, 617862
TRAPPC9	100	99,6	100	100	Mental retardation, autosomal recessive 13, 613192
TRDN	96,2	86,7	100	100	Ventricular tachycardia, catecholaminergic polymorphic, 5, with or without muscle weakness, 615441
TREM2	100	99,8	100	100	Polycystic lipomembranous osteodysplasia with sclerosing leukoencephalopathy 2, 618193
TREX1	100	100	100	100	{Systemic lupus erythematosus, susceptibility to}, 152700 Vasculopathy, retinal, with cerebral leukoencephalopathy and systemic manifestations, 192315 Aicardi-Goutieres syndrome 1, dominant and recessive, 225750 Chilblain lupus, 610448
TRH	99,6	96,5	100	100	Thyrotropin-releasing hormone deficiency, 275120
TRIM2	93,9	93,3	93,9	93,9	Charcot-Marie-Tooth disease, type 2R, 615490
TRIM32	100	100	100	100	?Bardet-Biedl syndrome 11, 615988 Muscular dystrophy, limb-girdle, autosomal recessive 8, 254110
TRIM36	100	99,2	100	100	?Anencephaly, 206500
TRIM37	98,6	98,1	98,7	98,7	Mulibrey nanism, 253250
TRIOBP	97,8	96,1	99,9	99,6	Deafness, autosomal recessive 28, 609823
TRIP11	98,4	94	100	100	Osteochondrodysplasia, 184260 Achondrogenesis, type IA, 200600
TRIP13	100	100	100	100	Mosaic variegated aneuploidy syndrome 3, 617598 Oocyte maturation defect 9, 619011
TRIP4	100	99,1	100	100	Spinal muscular atrophy with congenital bone fractures 1, 616866 ?Muscular dystrophy, congenital, Davignon-Chauveau type, 617066
TRIT1	100	100	100	100	Combined oxidative phosphorylation deficiency 35, 617873
TRMT1	99,4	96,2	100	100	Mental retardation, autosomal recessive 68, 618302
TRMT10A	100	99,7	100	100	Microcephaly, short stature, and impaired glucose metabolism 1, 616033
TRMT10C	100	100	100	99,9	Combined oxidative phosphorylation deficiency 30, 616974
TRMT5	100	99,3	100	100	Combined oxidative phosphorylation deficiency 26, 616539
TRMU	100	100	100	99,9	Liver failure, transient infantile, 613070 {Deafness, mitochondrial, modifier of}, 580000
TRNT1	99,5	96,5	100	100	Sideroblastic anemia with B-cell immunodeficiency, periodic fevers, and developmental delay, 616084 Retinitis pigmentosa and erythrocytic microcytosis, 616959
TRPM1	100	99,8	100	100	Night blindness, congenital stationary (complete), 1C, autosomal recessive, 613216
TRPM6	99,9	99,5	100	100	Hypomagnesemia 1, intestinal, 602014
TSEN15	79	77,2	100	100	Pontocerebellar hypoplasia, type 2F, 617026

TSEN2	100	99,6	100	100	Pontocerebellar hypoplasia type 2B, 612389
TSEN34	90,8	86,4	100	100	?Pontocerebellar hypoplasia type 2C, 612390
TSEN54	96,3	94,3	99,9	98,9	Pontocerebellar hypoplasia type 4, 225753 Pontocerebellar hypoplasia type 2A, 277470 ?Pontocerebellar hypoplasia type 5, 610204
TSFM	100	99,5	94,9	94,9	Combined oxidative phosphorylation deficiency 3, 610505
TSHB	100	100	100	100	Hypothyroidism, congenital, nongoitrous 4, 275100
TSHR	96	95,4	100	100	Hyperthyroidism, nonautoimmune, 609152 Hypothyroidism, congenital, nongoitrous, 1, 275200 Hyperthyroidism, familial gestational, 603373 Thyroid adenoma, hyperfunctioning, somatic, 0 Thyroid carcinoma with thyrotoxicosis, 0
TSPAN12	100	99,8	100	100	Exudative vitreoretinopathy 5, 613310
TSPEAR	100	99,2	100	100	Ectodermal dysplasia 14, hair/tooth type with or without hypohidrosis, 618180 ?Deafness, autosomal recessive 98, 614861
TSPYL1	100	100	100	100	Sudden infant death with dysgenesis of the testes syndrome, 608800
TTC19	81,5	73,8	100	99,2	Mitochondrial complex III deficiency, nuclear type 2, 615157
TTC21B	99,9	99,3	100	100	Nephronophthisis 12, 613820 Short-rib thoracic dysplasia 4 with or without polydactyly, 613819
TTC25	100	100	100	100	Ciliary dyskinesia, primary, 35, 617092
TTC37	100	99,3	100	100	Trichohepatoenteric syndrome 1, 222470
TTC7A	99,3	95,4	100	100	Gastrointestinal defects and immunodeficiency syndrome, 243150
TTC8	99,6	98,1	100	100	Bardet-Biedl syndrome 8, 615985 ?Retinitis pigmentosa 51, 613464
TTI2	100	100	100	100	Mental retardation, autosomal recessive 39, 615541
TTLL5	100	99,7	100	100	Cone-rod dystrophy 19, 615860
TTPA	94,7	87,1	100	100	Ataxia with isolated vitamin E deficiency, 277460
TUB	99,4	97,1	100	100	?Retinal dystrophy and obesity, 616188
TUBA8	99,9	99,5	100	100	Cortical dysplasia, complex, with other brain malformations 8, 613180
TUBGCP2	99,7	96,2	97	97	Pachygyria, microcephaly, developmental delay, and dysmorphic facies, with or without seizures, 618737
TUBGCP4	99,2	96,4	100	100	Microcephaly and chorioretinopathy, autosomal recessive, 3, 616335
TUBGCP6	100	99,3	100	100	Microcephaly and chorioretinopathy, autosomal recessive, 1, 251270
TUFM	100	99	100	100	Combined oxidative phosphorylation deficiency 4, 610678
TULP1	100	99,5	100	100	Retinitis pigmentosa 14, 600132 Leber congenital amaurosis 15, 613843
TUSC3	100	99,5	100	100	Mental retardation, autosomal recessive 7, 611093

TWIST2	100	100	100	100	Focal facial dermal dysplasia 3, Setleis type, 227260 Ablepharon-macrostomia syndrome, 200110 Barber-Say syndrome, 209885
TWNK	100	100	100	100	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	100	?Combined oxidative phosphorylation deficiency 29, 616811
TXNL4A	100	99,4	100	100	Burn-McKeown syndrome, 608572
TYK2	99,9	99	100	100	Immunodeficiency 35, 611521
TYMP	100	97	100	100	Mitochondrial DNA depletion syndrome 1 (MNGIE type), 603041
TYR	100	100	100	100	Waardenburg syndrome/albinism, digenic, 103470 Albinism, oculocutaneous, type IB, 606952 [Skin/hair/eye pigmentation 3, blue/green eyes], 601800 {Melanoma, cutaneous malignant, susceptibility to, 8}, 601800 Albinism, oculocutaneous, type IA, 203100 [Skin/hair/eye pigmentation 3, light/dark/freckling skin], 601800
TYROBP	100	100	100	100	Polycystic lipomembranous osteodysplasia with sclerosing leukoencephalopathy 1, 221770
TYRP1	100	99,8	100	100	[Skin/hair/eye pigmentation, variation in, 11 (Melanesian blond hair)], 612271 Albinism, oculocutaneous, type III, 203290
UBA5	97,8	86,8	100	100	?Spinocerebellar ataxia, autosomal recessive 24, 617133 Developmental and epileptic encephalopathy 44, 617132
UBE2T	100	99,9	100	100	Fanconi anemia, complementation group T, 616435
UBE3B	100	99,9	100	100	Kaufman oculocerebrofacial syndrome, 244450
UBR1	99,9	99,1	98	98	Johanson-Blizzard syndrome, 243800
UCHL1	99,8	92,5	100	100	Spastic paraplegia 79, autosomal recessive, 615491 {?Parkinson disease 5, susceptibility to}, 613643
UFC1	100	100	100	100	Neurodevelopmental disorder with spasticity and poor growth, 618076
UFM1	74	69,4	100	100	Leukodystrophy, hypomyelinating, 14, 617899
UGT1A1	100	100	100	100	[Gilbert syndrome], 143500 Hyperbilirubinemia, familial transient neonatal, 237900 Crigler-Najjar syndrome, type I, 218800 Crigler-Najjar syndrome, type II, 606785 [Bilirubin, serum level of, QTL1], 601816
UMPS	100	99,4	97	97	Orotic aciduria, 258900
UNC13D	99,7	98,1	100	100	Hemophagocytic lymphohistiocytosis, familial, 3, 608898
UNC80	97,9	97,4	100	100	Hypotonia, infantile, with psychomotor retardation and characteristic facies 2, 616801
UNG	100	98,8	99,9	99,3	Immunodeficiency with hyper IgM, type 5, 608106

UPB1	100	100	100	100	Beta-ureidopropionase deficiency, 613161
UQCC2	100	99,7	100	100	Mitochondrial complex III deficiency, nuclear type 7, 615824
UQCC3	100	98,7	100	100	?Mitochondrial complex III deficiency, nuclear type 9, 616111
UQCRB	99,4	95,1	100	100	Mitochondrial complex III deficiency, nuclear type 3, 615158
UQCRC2	99,9	99,3	100	100	Mitochondrial complex III deficiency, nuclear type 5, 615160
UQCRFS1	91,9	84,9	100	100	Mitochondrial complex III deficiency, nuclear type 10, 618775
UQCRQ	100	100	100	100	Mitochondrial complex III deficiency, nuclear type 4, 615159
UROC1	100	100	100	100	?Urocanase deficiency, 276880
UROD	98,9	96,1	100	100	Porphyria, hepatoerythropoietic, 176100 Porphyria cutanea tarda, 176100
UROS	100	99,9	100	100	Porphyria, congenital erythropoietic, 263700
USB1	100	99,4	100	100	Poikiloderma with neutropenia, 604173
USH1C	100	99,8	100	100	Usher syndrome, type 1C, 276904 Deafness, autosomal recessive 18A, 602092
USH1G	99,6	97,9	100	100	Usher syndrome, type 1G, 606943
USH2A	100	99,8	99,5	99,5	Retinitis pigmentosa 39, 613809 Usher syndrome, type 2A, 276901
USP18	95,9	95,9	100	100	Pseudo-TORCH syndrome 2, 617397
USP45	99,6	98,1	100	100	?Leber congenital amaurosis 19, 618513
UVSSA	100	100	100	100	UV-sensitive syndrome 3, 614640
VAC14	99,9	98,5	100	100	Striatonigral degeneration, childhood-onset, 617054
VAMP1	100	100	100	100	Spastic ataxia 1, autosomal dominant, 108600 Myasthenic syndrome, congenital, 25, 618323
VARS1	100	99,9	100	100	Neurodevelopmental disorder with microcephaly, seizures, and cortical atrophy, 617802
VARS2	100	99,4	100	100	Combined oxidative phosphorylation deficiency 20, 615917
VAX1	97,5	91,5	95,7	91,7	?Microphthalmia, syndromic 11, 614402
VDR	97,2	94,9	98,2	95,2	Rickets, vitamin D-resistant, type IIA, 277440
VHL	96,3	91,4	100	100	Pheochromocytoma, 171300 Erythrocytosis, familial, 2, 263400 von Hippel-Lindau syndrome, 193300 Renal cell carcinoma, somatic, 144700 Hemangioblastoma, cerebellar, somatic, 0
VIPAS39	100	100	100	100	Arthrogryposis, renal dysfunction, and cholestasis 2, 613404
VKORC1	100	100	93	93	Warfarin resistance, 122700 Vitamin K-dependent clotting factors, combined deficiency of, 2, 607473
VLDLR	100	99,8	100	100	Cerebellar hypoplasia and mental retardation with or without quadrupedal locomotion 1, 224050
VPS11	94,9	93,6	100	100	Leukodystrophy, hypomyelinating, 12, 616683

VPS13A	99,4	95,6	100	100	Choreoacanthocytosis, 200150
VPS13B	99,5	98,2	99,5	99,4	Cohen syndrome, 216550
VPS13C	99,4	96,9	100	100	Parkinson disease 23, autosomal recessive, early onset, 616840
VPS13D	100	99,7	100	100	Spinocerebellar ataxia, autosomal recessive 4, 607317
VPS33A	91,3	89,8	89,9	89,9	Mucopolysaccharidosis-plus syndrome, 617303
VPS33B	100	100	100	100	Arthrogryposis, renal dysfunction, and cholestasis 1, 208085
VPS37A	91,3	78,2	100	100	Spastic paraplegia 53, autosomal recessive, 614898
VPS45	99,2	95,7	95,3	95,3	Neutropenia, severe congenital, 5, autosomal recessive, 615285
VPS51	95	83,2	100	100	Pontocerebellar hypoplasia, type 13, 618606
VPS53	91,5	90,7	100	99,3	Pontocerebellar hypoplasia, type 2E, 615851
VRK1	99,7	98,5	100	100	Pontocerebellar hypoplasia type 1A, 607596
VSX2	100	99,3	100	100	Microphthalmia with coloboma 3, 610092 Microphthalmia, isolated 2, 610093
VWA3B	100	99,7	100	100	?Spinocerebellar ataxia, autosomal recessive 22, 616948
VWF	99,8	98,6	100	100	von Willebrand disease, type 1, 193400 von Willebrand disease, types 2A, 2B, 2M, and 2N, 613554 von Willebrand disease, type 3, 277480
WARS2	100	99,4	100	100	Neurodevelopmental disorder, mitochondrial, with abnormal movements and lactic acidosis, with or without seizures, 617710
WASHC4	99,1	95,5	100	100	?Mental retardation, autosomal recessive 43, 615817
WASHC5	100	99,8	100	100	Ritscher-Schinzel syndrome 1, 220210 Spastic paraplegia 8, autosomal dominant, 603563
WBP2	100	99,7	100	100	Deafness, autosomal recessive 107, 617639
WDPCP	98,2	94,4	98,1	98,1	?Bardet-Biedl syndrome 15, 615992 Congenital heart defects, hamartomas of tongue, and polysyndactyly, 217085
WDR19	100	99,4	100	100	Nephronophthisis 13, 614377 Senior-Loken syndrome 8, 616307 ?Cranioectodermal dysplasia 4, 614378 ?Short-rib thoracic dysplasia 5 with or without polydactyly, 614376
WDR34	100	99,6	100	100	Short-rib thoracic dysplasia 11 with or without polydactyly, 615633
WDR35	99,8	98,9	100	100	Short-rib thoracic dysplasia 7 with or without polydactyly, 614091 Cranioectodermal dysplasia 2, 613610
WDR4	100	100	100	100	Microcephaly, growth deficiency, seizures, and brain malformations, 618346 Galloway-Mowat syndrome 6, 618347
WDR45B	98	89,2	100	100	Neurodevelopmental disorder with spastic quadriplegia and brain abnormalities with or without seizures, 617977
WDR60	99,5	97	100	100	Short-rib thoracic dysplasia 8 with or without polydactyly, 615503

WDR62	100	99,5	100	100	Microcephaly 2, primary, autosomal recessive, with or without cortical malformations, 604317
WDR72	96,8	96,4	96,9	96,9	Amelogenesis imperfecta, type IIA3, 613211
WDR73	100	100	100	100	Galloway-Mowat syndrome 1, 251300
WDR81	100	100	100	100	Hydrocephalus, congenital, 3, with brain anomalies, 617967 Cerebellar ataxia, mental retardation, and dysequilibrium syndrome 2, 610185
WEE2	100	99,6	100	100	Oocyte maturation defect 5, 617996
WFS1	100	99,9	100	100	?Cataract 41, 116400 Deafness, autosomal dominant 6/14/38, 600965 {Diabetes mellitus, noninsulin-dependent, association with}, 125853 Wolfram-like syndrome, autosomal dominant, 614296 Wolfram syndrome 1, 222300
WHRN	99,8	98	100	100	Deafness, autosomal recessive 31, 607084 Usher syndrome, type 2D, 611383
WIPF1	100	99,9	100	100	Wiskott-Aldrich syndrome 2, 614493
WIPI2	100	99,3	100	100	?Intellectual developmental disorder with short stature and variable skeletal anomalies, 618453
WNK1	99,9	99,6	100	100	Pseudohypoaldosteronism, type IIC, 614492 Neuropathy, hereditary sensory and autonomic, type II, 201300
WNT1	99,3	95,3	100	100	{Osteoporosis, early-onset, susceptibility to, autosomal dominant}, 615221 Osteogenesis imperfecta, type XV, 615220
WNT10A	100	99,4	100	100	Schopf-Schulz-Passarge syndrome, 224750 Tooth agenesis, selective, 4, 150400 Odontoonychodermal dysplasia, 257980
WNT10B	100	99,4	100	100	Split-hand/foot malformation 6, 225300 Tooth agenesis, selective, 8, 617073
WNT3	100	99,6	100	100	?Tetra-amelia syndrome 1, 273395
WNT4	99,1	94,8	98,9	96,2	?SERKAL syndrome, 611812 Mullerian aplasia and hyperandrogenism, 158330
WNT7A	100	100	100	100	Fuhrmann syndrome, 228930 Ulna and fibula, absence of, with severe limb deficiency, 276820
WRAP53	100	100	100	100	Dyskeratosis congenita, autosomal recessive 3, 613988
WRN	99,9	98,8	100	100	Werner syndrome, 277700
WWOX	100	100	100	100	Spinocerebellar ataxia, autosomal recessive 12, 614322 Esophageal squamous cell carcinoma, somatic, 133239 Developmental and epileptic encephalopathy 28, 616211
XDH	100	99,9	100	100	Xanthinuria, type I, 278300
XPA	99,6	95,6	100	100	Xeroderma pigmentosum, group A, 278700
XPC	100	100	100	100	Xeroderma pigmentosum, group C, 278720

XPNPEP3	100	100	100	100	Nephronophthisis-like nephropathy 1, 613159
XRCC1	100	98,8	100	100	?Spinocerebellar ataxia, autosomal recessive 26, 617633
XRCC2	99,8	97,4	100	100	Spermatogenic failure, 619145 ?Premature ovarian failure 17, 619146 ?Fanconi anemia, complementation group U, 617247
XRCC4	99,9	99,3	100	100	Short stature, microcephaly, and endocrine dysfunction, 616541
XYLT1	97,4	89,6	98,1	94,8	{Pseudoxanthoma elasticum, modifier of severity of}, 264800 Desbuquois dysplasia 2, 615777
XYLT2	100	98,3	96,7	96,7	{Pseudoxanthoma elasticum, modifier of severity of}, 264800 Spondyloocular syndrome, 605822
YARS2	100	99,8	100	100	Myopathy, lactic acidosis, and sideroblastic anemia 2, 613561
YME1L1	99	95,2	100	100	?Optic atrophy 11, 617302
YY1AP1	99,3	98,2	100	100	Grange syndrome, 602531
ZAP70	100	99,3	100	100	Autoimmune disease, multisystem, infantile-onset, 2, 617006 Immunodeficiency 48, 269840
ZBTB11	99,9	99,6	100	100	Intellectual developmental disorder, autosomal recessive 69, 618383
ZBTB16	100	99,9	100	100	Skeletal defects, genital hypoplasia, and mental retardation, 612447 Leukemia, acute promyelocytic, PL2F/RARA type, 0
ZBTB24	100	100	100	100	Immunodeficiency-centromeric instability-facial anomalies syndrome 2, 614069
ZBTB42	100	100	100	100	?Lethal congenital contracture syndrome 6, 616248
ZC3H14	99,9	98,9	100	100	Mental retardation, autosomal recessive 56, 617125
ZFYVE26	100	99,1	100	100	Spastic paraplegia 15, autosomal recessive, 270700
ZMPSTE24	100	99,9	100	100	Restrictive dermopathy, lethal, 275210 Mandibuloacral dysplasia with type B lipodystrophy, 608612
ZMYND10	100	100	100	100	Ciliary dyskinesia, primary, 22, 615444
ZNF142	100	99,9	100	100	Neurodevelopmental disorder with impaired speech and hyperkinetic movements, 618425
ZNF335	100	99,9	100	100	Microcephaly 10, primary, autosomal recessive, 615095
ZNF341	97,2	95	100	100	Hyper-IgE recurrent infection syndrome 3, autosomal recessive, 618282
ZNF408	100	100	100	100	?Exudative vitreoretinopathy 6, 616468 Retinitis pigmentosa 72, 616469
ZNF423	100	100	100	100	Nephronophthisis 14, 614844 Joubert syndrome 19, 614844
ZNF469	100	100	100	100	Brittle cornea syndrome 1, 229200
ZNF513	100	100	100	100	?Retinitis pigmentosa 58, 613617
ZNHIT3	74,4	74,4	74,6	74,4	PEHO syndrome, 260565
ZP1	100	100	100	100	Oocyte maturation defect 1, 615774

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.
Agilent V5 is the default chemistry, and used for all exome analyses apart from the (in-house) TURBO/RAPID WES route.
TWIST is the chemistry used for (in-house) TURBO/RAPID WES analysis.
Covered 10x describes the percentage of a gene's coding sequence that is covered at least 10x.
Covered 20x describes the percentage of a gene's coding sequence that is covered at least 20x.
Genes with coverage denoting NC are non-protein coding genes.
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 23rd , 2021.
This list is accurate for panel version DG 3.1.0
Ad 1. "No OMIM Disease ID" signifies a gene without a current OMIM association Ad 2. OMIM phenotype descriptions between {} signify risk factors