COMPREHENSIVE PRECONCEPTION CARRIER TEST PANEL[1] DG-5.1.0 (2810 GENES)

Gene	*Twist X2 covered 10x*	*Twist X2 covered 20x*	*srWGS covered 10x*	*srWGS covered 15x*	*srWGS covered 20x*	*Associated Phenotype description and OMIM disease ID*
AAAS	100%	100%	100%	100%	99.7%	Achalasia-addisonianism-alacrimia syndrome, 231550
AARS1	100%	100%	100%	100%	99.4%	Developmental and epileptic encephalopathy 29, 616339;Charcot-Marie-Tooth disease, axonal, type 2N, 613287;?Leukoencephalopathy, hereditary diffuse, with spheroids 2, 619661;Trichothiodystrophy 8, nonphotosensitive, 619691
AARS2	100%	100%	100%	100%	99.5%	Leukoencephalopathy, progressive, with ovarian failure, 615889;Combined oxidative phosphorylation deficiency 8, 614096
AASS	100%	100%	100%	100%	99.8%	Hyperlysinemia, 238700
ABAT	100%	100%	100%	100%	99.4%	GABA-transaminase deficiency, 613163
ABCA1	100%	100%	100%	100%	99.8%	Tangier disease, 205400;HDL deficiency, familial, 1, 604091
ABCA12	100%	100%	100%	100%	99.6%	Ichthyosis, congenital, autosomal recessive 4B (harlequin), 242500;Ichthyosis, congenital, autosomal recessive 4A, 601277

ABCA2	100%	100%	100%	99.9%	98.7%	Intellectual developmental disorder with poor growth and with or without seizures or ataxia, 618808
ABCA3	97.2%	97.2%	100%	100%	99.5%	Surfactant metabolism dysfunction, pulmonary, 3, 610921
ABCA4	100%	100%	100%	100%	99.7%	Retinal dystrophy, early-onset severe, 248200;Retinitis pigmentosa 19, 601718;{Macular degeneration, age-related, 2}, 153800;Cone-rod dystrophy 3, 604116;Fundus flavimaculatus, 248200;Stargardt disease 1, 248200
ABCB1	100%	100%	100%	100%	99.7%	Encephalopathy, acute transient, 620950;{Inflammatory bowel disease 13}, 612244;{Colchicine resistance}, 120080
ABCB11	100%	100%	100%	100%	99.7%	Cholestasis, benign recurrent intrahepatic, 2, 605479;Cholestasis, progressive familial intrahepatic 2, 601847
ABCB4	100%	100%	100%	100%	99.8%	Gallbladder disease 1, 600803;Cholestasis, intrahepatic, of pregnancy, 3, 614972;Cholestasis, progressive familial intrahepatic 3, 602347
ABCC2	100%	100%	100%	100%	99.8%	Dubin-Johnson syndrome, 237500

ABCC6	94.4%	94.4%	100%	100%	99.5%	Pseudoxanthoma elasticum, 264800;Arterial calcification, generalized, of infancy, 2, 614473;Pseudoxanthoma elasticum, forme fruste, 177850
ABCC8	100%	100%	100%	100%	99.5%	Diabetes mellitus, permanent neonatal 3, 618857;Maturity-onset diabetes of the young, type 12, 621196;Diabetes mellitus, transient neonatal 2, 610374;Diabetes mellitus, noninsulin-dependent, 125853;Hypoglycemia of infancy, leucine-sensitive, 240800;Hyperinsulinemic hypoglycemia, familial, 1, 256450
ABCC9	96%	96%	100%	100%	99.8%	Cardiomyopathy, dilated, 1O, 608569;Hypertrichotic osteochondrodysplasia (Cantu syndrome), 239850;?Atrial fibrillation, familial, 12, 614050;Intellectual disability and myopathy syndrome, 619719
ABCD4	100%	99.6%	100%	99.9%	99.3%	Methylmalonic aciduria and homocystinuria, cblJ type, 614857
ABCG5	100%	100%	100%	100%	99.5%	Sitosterolemia 2, 618666
ABCG8	100%	100%	100%	100%	99.7%	Sitosterolemia 1, 210250;{Gallbladder disease 4}, 611465
ABHD12	100%	100%	100%	100%	99.6%	Polyneuropathy, hearing loss, ataxia, retinitis pigmentosa, and cataract, 612674

ABHD16A	97.5%	97.5%	100%	100%	99.4%	Spastic paraplegia 86, autosomal recessive, 619735
ABHD5	100%	100%	100%	100%	99.9%	Chanarin-Dorfman syndrome, 275630
ACACA	100%	100%	100%	100%	99.7%	Acetyl-CoA carboxylase deficiency, 613933
ACAD8	100%	99.9%	100%	99.9%	99.5%	Isobutyryl-CoA dehydrogenase deficiency, 611283
ACAD9	100%	100%	100%	100%	99.7%	Mitochondrial complex I deficiency, nuclear type 20, 611126
ACADM	93.1%	91.3%	100%	100%	99.7%	Acyl-CoA dehydrogenase, medium chain, deficiency of, 201450
ACADS	100%	100%	100%	100%	99.5%	Acyl-CoA dehydrogenase, short-chain, deficiency of, 201470
ACADSB	100%	100%	100%	99.9%	99.7%	2-methylbutyrylglycinuria, 610006
ACADVL	100%	100%	100%	100%	99.3%	VLCAD deficiency, 201475
ACAN	99.1%	98.9%	98.5%	97.5%	96.1%	?Spondyloepiphyseal dysplasia, Kimberley type, 608361;Short stature and advanced bone age, with or without early-onset osteoarthritis and/or osteochondritis dissecans, 165800;Spondyloepimetaphyseal dysplasia, aggrecan type, 612813
ACAT1	100%	100%	100%	100%	99.6%	Alpha-methylacetoacetic aciduria, 203750
ACBD5	85.6%	85.6%	100%	100%	99.5%	Retinal dystrophy with leukodystrophy, 618863

ACBD6	79.5%	79.5%	100%	99.9%	99.6%	Neurodevelopmental disorder with progressive movement abnormalities, 620785
ACE	100%	100%	100%	99.8%	98.5%	Renal tubular dysgenesis, 267430;{Microvascular complications of diabetes 3}, 612624;[Angiotensin I-converting enzyme, benign serum increase], 621420
ACER3	96.3%	96.3%	100%	100%	99.9%	?Leukodystrophy, progressive, early childhood-onset, 617762
ACO2	90.7%	86.8%	100%	100%	99.7%	Optic atrophy 9, 616289;Infantile cerebellar-retinal degeneration, 614559
ACOX1	97.9%	97.9%	100%	100%	99.7%	Mitchell syndrome, 618960;Peroxisomal acyl-CoA oxidase deficiency, 264470
ACOX2	100%	100%	100%	100%	99.7%	Bile acid synthesis defect, congenital, 6, 617308
ACP4	100%	100%	100%	99.9%	99.4%	Amelogenesis imperfecta, type IJ, 617297
ACP5	100%	100%	100%	100%	99.6%	Spondyloenchondrodysplasia with immune dysregulation, 607944
ACSF3	94.7%	94.7%	100%	100%	99.3%	Combined malonic and methylmalonic aciduria, 614265

ACTA1	100%	100%	100%	99.8%	97.9%	Congenital myopathy 2B, severe infantile, autosomal recessive, 620265;?Myopathy, scapulohumeroperoneal, 616852;Congenital myopathy 2C, severe infantile, autosomal dominant, 620278;Congenital myopathy 2A, typical, autosomal dominant, 161800
ACTL6B	95.9%	94.6%	100%	100%	99.4%	Developmental and epileptic encephalopathy 76, 618468;Intellectual developmental disorder with severe speech and ambulation defects, 618470
ACY1	100%	100%	100%	99.9%	99.4%	Aminoacylase 1 deficiency, 609924
ADA	85.1%	84.4%	100%	99.9%	99.5%	Adenosine deaminase deficiency, partial, 102700;Severe combined immunodeficiency due to ADA deficiency, 102700
ADA2	95.1%	93.2%	100%	100%	99.8%	Sneddon syndrome, 182410;Vasculitis, autoinflammation, immunodeficiency, and hematologic defects syndrome, 615688
ADAM17	96.1%	96.1%	100%	100%	99.8%	?Inflammatory skin and bowel disease, neonatal, 1, 614328;?Hypotichosis 16, 621490
ADAM22	98.7%	98.7%	100%	100%	99.7%	Developmental and epileptic encephalopathy 61, 617933
ADAM9	95%	95%	100%	100%	99.6%	Cone-rod dystrophy 9, 612775

ADAMTS10	100%	100%	100%	100%	99.6%	Weill-Marchesani syndrome 1, recessive, 277600
ADAMTS13	100%	100%	100%	100%	99.4%	Thrombotic thrombocytopenic purpura, hereditary, 274150
ADAMTS15	100%	100%	100%	99.9%	99%	Arthrogryposis, distal, type 12, 620545
ADAMTS17	100%	100%	100%	100%	99.3%	Weill-Marchesani 4 syndrome, recessive, 613195
ADAMTS18	100%	100%	100%	100%	99.7%	Microcornea, myopic chorioretinal atrophy, and telecanthus, 615458
ADAMTS19	94%	94%	100%	99.9%	99.3%	Cardiac valvular dysplasia 2, 620067
ADAMTS2	97.9%	97.9%	100%	99.9%	99.5%	Ehlers-Danlos syndrome, dermatosparaxis type, 225410
ADAMTS3	100%	100%	100%	100%	99.8%	Hennekam lymphangiectasia-lymphedema syndrome 3, 618154
ADAMTSL2	100%	100%	100%	99.9%	99.4%	Geleophysic dysplasia 1, 231050
ADAMTSL4	100%	100%	100%	100%	99.4%	Ectopia lentis et pupillae, 225200;Ectopia lentis, isolated, autosomal recessive, 225100
ADAR	100%	100%	100%	100%	99.8%	Dyschromatosis symmetrica hereditaria, 127400;Aicardi-Goutieres syndrome 6, 615010
ADARB1	94.8%	94.7%	100%	100%	99.4%	Neurodevelopmental disorder with hypotonia, microcephaly, and seizures, 618862

ADAT3	100%	100%	100%	100%	99.7%	Neurodevelopmental disorder with brain abnormalities, poor growth, and dysmorphic facies, 615286
ADCY5	97.5%	96.9%	100%	99.9%	99.1%	Dyskinesia with orofacial involvement, autosomal dominant, 606703;Neurodevelopmental disorder with hyperkinetic movements and dyskinesia, 619651;Dyskinesia with orofacial involvement, autosomal recessive, 619647
ADCY6	100%	100%	100%	100%	99.6%	Lethal congenital contracture syndrome 8, 616287
ADD3	97.6%	97.4%	100%	100%	99.6%	Cerebral palsy, spastic quadriplegic, 3, 617008
ADGRG1	100%	100%	100%	100%	99.3%	Cortical dysplasia, complex, with other brain malformations 14B, (bilateral perisylvian), 615752;Cortical dysplasia, complex, with other brain malformations 14A, (bilateral frontoparietal), 606854
ADGRG6	100%	100%	100%	100%	99.7%	Lethal congenital contracture syndrome 9, 616503
ADGRV1	100%	100%	100%	100%	99.8%	Usher syndrome, type 2C, 605472;Usher syndrome, type 2C, GPR98/PDZD7 digenic, 605472;?Febrile seizures, familial, 4, 604352
ADK	88%	88%	100%	100%	99.8%	Hypermethioninemia due to adenosine kinase deficiency, 614300

ADPRS	100%	100%	100%	100%	99.4%	Neurodegeneration, childhood-onset, stress-induced, with variable ataxia and seizures, 618170
ADSL	99.5%	97.5%	100%	100%	99.7%	Adenylosuccinase deficiency, 103050
ADSS1	100%	100%	100%	99.9%	99%	Myopathy, distal, 5, 617030
AEBP1	100%	100%	100%	99.9%	99.3%	Ehlers-Danlos syndrome, classic-like, 2, 618000
AFG2A	100%	100%	100%	100%	99.9%	Neurodevelopmental disorder with hearing loss, seizures, and brain abnormalities, 616577
AFG2B	100%	100%	100%	100%	99.8%	Deafness, autosomal recessive 119, 619615;Neurodevelopmental disorder with hearing loss and spasticity, 619616
AFG3L2	94.2%	92.2%	100%	100%	99.5%	Spastic ataxia 5, autosomal recessive, 614487;Optic atrophy 12, 618977;Spinocerebellar ataxia 28, 610246
AGA	91.6%	91.6%	100%	100%	99.5%	Aspartylglucosaminuria, 208400
AGBL5	100%	100%	100%	100%	99.3%	Retinitis pigmentosa 75, 617023
AGK	91.6%	88.9%	100%	100%	100%	Cataract 38, autosomal recessive, 614691;Sengers syndrome, 212350
AGL	100%	100%	100%	100%	99.7%	Glycogen storage disease IIIa, 232400;Glycogen storage disease IIIb, 232400

AGPAT2	100%	99.9%	100%	100%	98.9%	Lipodystrophy, congenital generalized, type 1, 608594
AGPS	97.3%	97.3%	100%	100%	99.8%	Rhizomelic chondrodysplasia punctata, type 3, 600121
AGR2	100%	100%	100%	100%	99%	Respiratory infections, recurrent, and failure to thrive with or without diarrhea, 620233
AGRN	100%	100%	100%	99.9%	98.9%	Myasthenic syndrome, congenital, 8, with pre- and postsynaptic defects, 615120
AGT	95.7%	95.3%	100%	100%	99.5%	Renal tubular dysgenesis, 267430
AGTPBP1	96.5%	96.5%	100%	100%	99.7%	Neurodegeneration, childhood-onset, with cerebellar atrophy, 618276
AGTR1	100%	100%	100%	100%	99.9%	{Hypertension, essential}, 145500;Renal tubular dysgenesis, 267430
AGXT	97.4%	96.4%	100%	100%	99.5%	Hyperoxaluria, primary, type 1, 259900
AHCY	100%	100%	100%	99.9%	99.1%	Hypermethioninemia with deficiency of S-adenosylhomocysteine hydrolase, 613752
AHI1	98.7%	98.7%	100%	99.9%	99.6%	Joubert syndrome 3, 608629
AHR	100%	100%	100%	100%	99.8%	Foveal hypoplasia 3, 620958;?Retinitis pigmentosa 85, 618345
AICDA	92.1%	92.1%	100%	100%	99.7%	Immunodeficiency with hyper-IgM, type 2, 605258
AIMP1	100%	100%	100%	100%	99.9%	Leukodystrophy, hypomyelinating, 3, 260600

AIMP2	100%	100%	100%	99.8%	99.1%	Leukodystrophy, hypomyelinating, 17, 618006
AIPL1	100%	100%	100%	99.9%	98.9%	Leber congenital amaurosis 4, 604393;Retinitis pigmentosa, juvenile, 604393;Cone-rod dystrophy, 604393
AIRE	100%	100%	100%	99.8%	99.1%	Autoimmune polyendocrinopathy syndrome , type I, with or without reversible metaphyseal dysplasia, 240300
AK1	100%	100%	100%	100%	99.8%	Anemia, congenital, nonspherocytic hemolytic, 3, adenylate kinase deficient, 612631
AK2	98.1%	93.2%	100%	100%	99.1%	Reticular dysgenesis, 267500
AKR1C2	96.7%	92.5%	99.9%	99.7%	98.6%	46XY sex reversal 8, 614279
AKR1D1	88.4%	88.4%	100%	100%	99.8%	Bile acid synthesis defect, congenital, 2, 235555
ALAD	100%	100%	100%	100%	99.4%	Porphyria, acute hepatic, 612740;{Lead poisoning, susceptibility to}, 612740
ALB	100%	99.1%	100%	100%	99.9%	?[Dysalbuminemic hypertriiodothyroninemia], 615999;Analbuminemia, 616000;[Dysalbuminemic hyperthyroxinemia], 615999

ALDH18A1	100%	100%	100%	100%	99.6%	Spastic paraplegia 9A, autosomal dominant, 601162;Cutis laxa, autosomal recessive, type IIIA, 219150;Spastic paraplegia 9B, autosomal recessive, 616586;Cutis laxa, autosomal dominant 3, 616603
ALDH1A2	88.9%	88.9%	100%	100%	99.5%	Diaphragmatic hernia 4, with cardiovascular defects, 620025
ALDH1A3	94.4%	94.4%	100%	99.9%	99.4%	Microphthalmia, isolated 8, 615113
ALDH3A2	93.5%	93.5%	100%	100%	99.7%	Sjogren-Larsson syndrome, 270200
ALDH4A1	97.9%	96.1%	100%	100%	99.5%	Hyperprolinemia, type II, 239510
ALDH5A1	100%	100%	100%	100%	99.5%	Succinic semialdehyde dehydrogenase deficiency, 271980
ALDH6A1	95.2%	95.2%	100%	100%	99.7%	Methylmalonate semialdehyde dehydrogenase deficiency, 614105
ALDH7A1	97.2%	97.2%	100%	100%	99.8%	Epilepsy, early-onset, 4, vitamin B6-dependent, 266100
ALDOA	100%	100%	100%	100%	99.4%	Glycogen storage disease XII, 611881
ALDOB	100%	100%	100%	99.9%	99.8%	Fructose intolerance, hereditary, 229600
ALG1	100%	100%	100%	100%	99.7%	Congenital disorder of glycosylation, type Ik, 608540
ALG11	91%	91%	100%	100%	99.9%	Congenital disorder of glycosylation, type Ip, 613661
ALG12	100%	100%	100%	100%	99.6%	Congenital disorder of glycosylation, type Ig, 607143

ALG14	100%	100%	100%	100%	99.8%	Intellectual developmental disorder with epilepsy, behavioral abnormalities, and coarse facies, 619031;Myopathy, epilepsy, and progressive cerebral atrophy, 619036;?Myasthenic syndrome, congenital, 15, without tubular aggregates, 616227
ALG2	100%	100%	100%	100%	99.8%	Congenital disorder of glycosylation, type Ii, 607906;Myasthenic syndrome, congenital, 14, with tubular aggregates, 616228
ALG3	100%	100%	100%	100%	99.4%	Congenital disorder of glycosylation, type Id, 601110
ALG6	100%	100%	100%	100%	99.9%	Congenital disorder of glycosylation, type Ic, 603147
ALG8	78.4%	77.9%	100%	100%	99.7%	Congenital disorder of glycosylation, type Ih, 608104;Polycystic liver disease 3 with or without kidney cysts, 617874
ALG9	100%	100%	100%	99.9%	99.5%	Gillessen-Kaesbach-Nishimura syndrome, 263210;Congenital disorder of glycosylation, type Il, 608776
ALKBH8	96.3%	96.3%	100%	100%	99.7%	Intellectual developmental disorder, autosomal recessive 71, 618504
ALMS1	100%	100%	100%	100%	99.8%	Alstrom syndrome, 203800
ALOX12B	100%	100%	100%	100%	99.5%	Ichthyosis, congenital, autosomal recessive 2, 242100

ALOXE3	100%	100%	100%	100%	99.4%	Ichthyosis, congenital, autosomal recessive 3, 606545
ALPK3	100%	100%	100%	100%	99.5%	Cardiomyopathy, familial hypertrophic 27, 618052
ALPL	100%	100%	100%	100%	99.3%	Odontohypophosphatasia, 146300;Hypophosphatasia, infantile, 241500;Hypophosphatasia, childhood, 241510;Hypophosphatasia, adult, 146300
ALS2	97%	96.3%	100%	100%	99.8%	Primary lateral sclerosis, juvenile, 606353;Spastic paralysis, infantile onset ascending, 607225;Amyotrophic lateral sclerosis 2, juvenile, 205100
ALX1	100%	100%	100%	100%	99.7%	Frontonasal dysplasia 3, 613456
ALX3	100%	100%	100%	99.5%	98.7%	Frontonasal dysplasia 1, 136760
ALX4	100%	100%	100%	100%	99.4%	Parietal foramina 2, 609597;{Craniosynostosis 5, susceptibility to}, 615529;Frontonasal dysplasia 2, 613451
AMACR	100%	100%	100%	99.8%	99.4%	Alpha-methylacyl-CoA racemase deficiency, 614307;Bile acid synthesis defect, congenital, 4, 214950
AMBN	100%	100%	100%	100%	99.9%	Amelogenesis imperfecta, type IF, 616270
AMFR	100%	100%	100%	100%	99.6%	Spastic paraplegia 89, autosomal recessive, 620379
AMH	100%	100%	100%	99.9%	99%	Persistent Mullerian duct syndrome, type I, 261550

AMHR2	100%	100%	100%	100%	99.4%	Persistent Mullerian duct syndrome, type II, 261550
AMN	100%	100%	100%	99.9%	99.1%	Imerslund-Grasbeck syndrome 2, 618882
AMPD1	100%	100%	100%	100%	99.7%	Myopathy due to myoadenylate deaminase deficiency, 615511
AMPD2	100%	100%	100%	99.9%	99.6%	Pontocerebellar hypoplasia, type 9, 615809;?Spastic paraplegia 63, autosomal recessive, 615686
AMT	100%	100%	100%	99.9%	99.3%	Glycine encephalopathy 2, 620398
ANAPC1	100%	100%	100%	99.9%	99.5%	Rothmund-Thomson syndrome, type 1, 618625
ANAPC7	91%	91%	100%	100%	99.6%	Ferguson-Bonni neurodevelopmental syndrome, 619699
ANGPTL3	100%	100%	100%	100%	99.6%	Hypobetalipoproteinemia, familial, 2, 605019
ANK1	100%	100%	100%	100%	99.5%	Spherocytosis, type 1, 182900
ANK3	99.7%	99.7%	100%	100%	99.8%	Intellectual developmental disorder, autosomal recessive 37, 615493
ANKH	100%	100%	100%	100%	99.4%	Chondrocalcinosis 2, 118600;Craniometaphyseal dysplasia, 123000
ANKLE2	100%	100%	100%	99.8%	99.1%	Microcephaly 16, primary, autosomal recessive, 616681
ANKS6	100%	99.9%	100%	100%	99.5%	Nephronophthisis 16, 615382
ANO10	94.9%	94.9%	100%	100%	100%	Spinocerebellar ataxia, autosomal recessive 10, 613728

ANO5	100%	100%	100%	100%	99.8%	Muscular dystrophy, limb-girdle, autosomal recessive 12, 611307;Miyoshi muscular dystrophy 3, 613319;Gnathodiaphyseal dysplasia, 166260
ANO6	95.9%	94.9%	100%	100%	99.8%	Scott syndrome, 262890
ANTXR1	100%	100%	100%	99.9%	99.4%	GAPO syndrome, 230740;{?Hemangioma, capillary infantile, susceptibility to}, 602089
ANTXR2	94.3%	94.3%	100%	100%	99.8%	Hyaline fibromatosis syndrome, 228600
AOPEP	100%	100%	100%	100%	99.5%	Dystonia 31, 619565
AP1B1	93.7%	91.9%	100%	100%	99.6%	Keratitis-ichthyosis-deafness syndrome, autosomal recessive, 242150
AP1G1	100%	100%	100%	100%	99.7%	Usmani-Riazuddin syndrome, autosomal recessive, 619548;Usmani-Riazuddin syndrome, autosomal dominant, 619467
AP1S1	100%	100%	100%	100%	99.2%	MEDNIK syndrome, 609313
AP3B1	100%	100%	100%	100%	99.8%	Hermansky-Pudlak syndrome 2, 608233
AP3B2	100%	100%	100%	100%	99.5%	Developmental and epileptic encephalopathy 48, 617276
AP3D1	100%	100%	100%	100%	99.7%	?Hermansky-Pudlak syndrome 10, 617050
AP4B1	100%	100%	100%	100%	99.8%	Spastic paraplegia 47, autosomal recessive, 614066

AP4E1	100%	100%	100%	100%	99.9%	Stuttering, familial persistent, 1, 184450;Spastic paraplegia 51, autosomal recessive, 613744
AP4M1	100%	100%	100%	100%	99.7%	Spastic paraplegia 50, autosomal recessive, 612936
AP4S1	79.1%	79.1%	100%	99.9%	99.4%	Spastic paraplegia 52, autosomal recessive, 614067
AP5Z1	91.2%	91%	100%	100%	99.5%	Spastic paraplegia 48, autosomal recessive, 613647
APC2	100%	100%	100%	100%	99%	Cortical dysplasia, complex, with other brain malformations 10, 618677;Intellectual developmental disorder, autosomal recessive 74, 617169
APOA1	100%	100%	100%	99.8%	99.3%	Hypoalphalipoproteinemia, primary, 2, 618463;Amyloidosis, hereditary systemic 3, 620657;Hypoalphalipoproteinemia, primary, 2, intermediate, 619836
APOB	100%	100%	100%	100%	99.7%	Hypercholesterolemia, familial, 2, 144010;Hypobetalipoproteinemia, 615558
APOC2	100%	100%	100%	100%	98.3%	Hyperlipoproteinemia, type Ib, 207750

APOE	93.8%	93.4%	100%	100%	99.1%	Alzheimer disease 2, 104310;Sea-blue histiocyte disease, 269600;{?Alzheimer disease, protection against, due to APOE3-Christchurch}, 607822;{Coronary artery disease, severe, susceptibility to}, 617347;Lipoprotein glomerulopathy, 611771;{?Macular degeneration, age-related}, 603075;Hyperlipoproteinemia, type III, 617347
APRT	100%	100%	100%	100%	99.1%	Adenine phosphoribosyltransferase deficiency, 614723
APTX	100%	100%	100%	100%	99.7%	Ataxia, early-onset, with oculomotor apraxia and hypoalbuminemia, 208920
AQP2	100%	100%	100%	100%	99.8%	Diabetes insipidus, nephrogenic, 2, 125800
ARFGEF2	100%	100%	100%	100%	99.5%	Periventricular heterotopia with microcephaly, 608097
ARG1	93%	93%	100%	100%	99.8%	Argininemia, 207800
ARHGAP19	94.5%	94.1%	100%	100%	99.6%	Charcot-Marie-Tooth disease, axonal, type 2KK, 621466
ARHGDIA	100%	100%	100%	100%	99%	Nephrotic syndrome, type 8, 615244
ARHGEF18	100%	100%	100%	100%	99.3%	Retinitis pigmentosa 78, 617433
ARHGEF2	97.9%	97.9%	100%	100%	99.3%	?Neurodevelopmental disorder with midbrain and hindbrain malformations, 617523
ARL13B	93.4%	93.3%	100%	100%	99.7%	Joubert syndrome 8, 612291

ARL2BP	100%	100%	100%	100%	99.9%	Retinitis pigmentosa 82 with or without situs inversus, 615434
ARL3	86.8%	86.8%	100%	100%	99.8%	Retinitis pigmentosa 83, 618173;Joubert syndrome 35, 618161
ARL6	100%	100%	100%	100%	99.8%	Retinitis pigmentosa 55, 613575;{Bardet-Biedl syndrome 1, modifier of}, 209900;Bardet-Biedl syndrome 3, 600151
ARL6IP1	100%	100%	100%	100%	99.9%	Spastic paraplegia 61, autosomal recessive, 615685
ARMC9	96.3%	94.2%	100%	100%	99.3%	Joubert syndrome 30, 617622
ARNT2	91%	91%	100%	100%	99.5%	?Webb-Dattani syndrome, 615926
ARPC1B	99.4%	95.9%	100%	100%	99.3%	Immunodeficiency 71 with inflammatory disease and congenital thrombocytopenia, 617718
ARPC5	100%	100%	100%	100%	100%	Immunodeficiency 113 with autoimmunity and autoinflammation, 620565
ARSA	100%	100%	100%	99.9%	99%	Metachromatic leukodystrophy, 250100
ARSB	100%	100%	100%	100%	99.3%	Mucopolysaccharidosis type VI (Maroteaux-Lamy), 253200
ARSG	100%	100%	100%	100%	99.1%	Usher syndrome, type IV, 618144
ARSK	100%	100%	100%	100%	99.8%	Mucopolysaccharidosis, type X, 619698

ARV1	87.3%	86.5%	100%	100%	99.8%	Developmental and epileptic encephalopathy 38, 617020
ASAH1	100%	100%	100%	99.9%	99.3%	Spinal muscular atrophy with progressive myoclonic epilepsy, 159950;Farber lipogranulomatosis, 228000
ASCC1	86.6%	86.6%	100%	100%	99.6%	Spinal muscular atrophy with congenital bone fractures 2, 616867;Barrett esophagus/esophageal adenocarcinoma, 614266
ASCC3	100%	100%	100%	100%	99.8%	Intellectual developmental disorder, autosomal recessive 81, 620700
ASL	100%	100%	100%	100%	99.1%	Argininosuccinic aciduria, 207900
ASNS	100%	100%	100%	100%	99.8%	Asparagine synthetase deficiency, 615574
ASPA	100%	100%	100%	100%	99.8%	Canavan disease, 271900
ASPH	100%	100%	100%	100%	99.7%	Traboulsi syndrome, 601552
ASPM	97.9%	97.9%	100%	100%	99.7%	Microcephaly 5, primary, autosomal recessive, 608716
ASS1	92.3%	90%	100%	100%	99.6%	Citrullinemia, 215700
ATAD1	96.8%	96.8%	100%	100%	100%	Hyperekplexia 4, 618011
ATAD3A	100%	100%	100%	99.9%	98.7%	Harel-Yoon syndrome, 617183;Pontocerebellar hypoplasia, hypotonia, and respiratory insufficiency syndrome, neonatal lethal, 618810

ATCAY	100%	100%	100%	100%	99.2%	Ataxia, cerebellar, Cayman type, 601238
ATF6	90.9%	90.9%	100%	100%	99.7%	Achromatopsia 7, 616517
ATG5	90.4%	90.4%	100%	100%	99.9%	?Spinocerebellar ataxia, autosomal recessive 25, 617584
ATG7	100%	100%	100%	100%	99.8%	Spinocerebellar ataxia, autosomal recessive 31, 619422
ATIC	100%	100%	100%	100%	99.9%	AICA-ribosiduria due to ATIC deficiency, 608688
ATM	100%	100%	100%	100%	99.7%	Lymphoma, B-cell non-Hodgkin, somatic;Ataxia-telangiectasia, 208900;{Breast cancer, susceptibility to}, 114480;T-cell prolymphocytic leukemia, somatic;Lymphoma, mantle cell, somatic
ATOH7	100%	100%	100%	100%	99.6%	Persistent hyperplastic primary vitreous, autosomal recessive, 221900
ATP13A2	100%	100%	100%	100%	99.5%	Spastic paraplegia 78, autosomal recessive, 617225;Kufor-Rakeb syndrome, 606693
ATP13A3	97%	97%	100%	100%	99.8%	Pulmonary hypertension, primary, 5, 265400

ATP1A2	100%	100%	100%	99.9%	99.4%	Developmental and epileptic encephalopathy 98, 619605;Fetal akinesia, respiratory insufficiency, microcephaly, polymicrogyria, and dysmorphic facies, 619602;Alternating hemiplegia of childhood 1, 104290;Migraine, familial basilar, 602481;Migraine, familial hemiplegic, 2, 602481
ATP2A1	100%	100%	100%	100%	99.3%	Brody myopathy, 601003
ATP5F1A	100%	100%	100%	99.9%	99.7%	Mitochondrial complex V (ATP synthase) deficiency, nuclear type 4A, 620358;?Combined oxidative phosphorylation deficiency 22, 616045;?Mitochondrial complex V (ATP synthase) deficiency, nuclear type 4B, encephalopathic type, 615228
ATP5F1D	100%	100%	100%	99.9%	98.9%	Mitochondrial complex V (ATP synthase) deficiency, 618120
ATP5F1E	100%	100%	100%	100%	100%	Mitochondrial complex V (ATP synthase) deficiency, nuclear type 3, 614053
ATP5MK	100%	100%	100%	100%	99.8%	Mitochondrial complex V (ATP synthase) deficiency, nuclear type 6, 618683
ATP5PO	100%	100%	100%	100%	99.7%	Mitochondrial complex V (ATP synthase) deficiency, nuclear type 7, 620359

ATP6V0A1	92.9%	92.9%	100%	100%	99.7%	Neurodevelopmental disorder with epilepsy and brain atrophy, 619971;Developmental and epileptic encephalopathy 104, 619970
ATP6V0A2	100%	100%	100%	100%	99.6%	Wrinkly skin syndrome, 278250;Cutis laxa, autosomal recessive, type IIA, 219200
ATP6V0A4	100%	100%	100%	99.9%	99%	Distal renal tubular acidosis 3, with or without sensorineural hearing loss, 602722
ATP6V1A	100%	100%	100%	100%	99.8%	Cutis laxa, autosomal recessive, type IID, 617403;Developmental and epileptic encephalopathy 93, 618012
ATP6V1B1	100%	100%	100%	100%	99.5%	Distal renal tubular acidosis 2 with progressive sensorineural hearing loss, 267300
ATP6V1E1	100%	100%	100%	100%	99.6%	Cutis laxa, autosomal recessive, type IIC, 617402
ATP7B	100%	100%	100%	100%	99.6%	Wilson disease, 277900
ATP8A2	100%	100%	100%	100%	99.6%	Cerebellar ataxia, impaired intellectual development, and dysequilibrium syndrome 4, 615268
ATP8B1	100%	100%	100%	100%	99.7%	Cholestasis, progressive familial intrahepatic 1, 211600;Cholestasis, intrahepatic, of pregnancy, 1, 147480;Cholestasis, benign recurrent intrahepatic, 243300

ATP9A	100%	100%	100%	100%	99.6%	Neurodevelopmental disorder with poor growth and behavioral abnormalities, 620242
ATPAF2	100%	100%	100%	100%	99.5%	?Mitochondrial complex V (ATP synthase) deficiency, nuclear type 1, 604273
ATR	100%	100%	99.9%	99.7%	99.2%	Seckel syndrome 1, 210600;?Cutaneous telangiectasia and cancer syndrome, familial, 614564
AUH	100%	100%	100%	100%	99.7%	3-methylglutaconic aciduria, type I, 250950
AURKC	100%	100%	100%	100%	99.5%	Spermatogenic failure 5, 243060
AVIL	100%	100%	100%	100%	99.7%	Nephrotic syndrome, type 21, 618594
AXIN1	100%	100%	100%	100%	99.5%	Hepatocellular carcinoma, somatic, 114550;Craniometadiaphyseal osteosclerosis with hip dysplasia, 620558;?Caudal duplication anomaly, 607864
B2M	100%	100%	100%	100%	99.8%	Amyloidosis, hereditary systemic 6, 620659;Immunodeficiency 43, 241600
B3GALNT2	92.6%	92.6%	100%	100%	99.7%	Muscular dystrophy-dystroglycanopathy (congenital with brain and eye anomalies), type A, 11, 615181

B3GALT6	100%	98.6%	100%	99.9%	98.6%	Ehlers-Danlos syndrome, spondylodysplastic type, 2, 615349;Spondyloepimetaphyseal dysplasia with joint laxity, type 1, with or without fractures, 271640;Al-Gazali syndrome, 609465
B3GAT3	94.7%	93.8%	100%	100%	99.8%	Multiple joint dislocations, short stature, craniofacial dysmorphism, with or without congenital heart defects, 245600
B3GLCT	100%	100%	100%	100%	99.9%	Peters-plus syndrome, 261540
B4GALNT1	100%	100%	100%	100%	99.5%	Spastic paraplegia 26, autosomal recessive, 609195
B4GALT1	91%	88.3%	100%	100%	99.6%	Combined low LDL and fibrinogen, 620364;Congenital disorder of glycosylation, type IId, 607091
B4GALT7	86.8%	86.8%	100%	100%	99.9%	Ehlers-Danlos syndrome, spondylodysplastic type, 1, 130070
B4GAT1	100%	100%	100%	100%	99.3%	Muscular dystrophy-dystroglycanopathy (congenital with brain and eye anomalies), type A, 13, 615287
B9D1	100%	100%	100%	100%	99.4%	?Meckel syndrome 9, 614209;Joubert syndrome 27, 617120
B9D2	100%	100%	100%	100%	99.7%	?Meckel syndrome 10, 614175;Joubert syndrome 34, 614175
BAAT	100%	100%	100%	100%	99.8%	Hypercholanemia, familial 3, 619232

BAG5	100%	100%	100%	100%	98.8%	Cardiomyopathy, dilated, 2F, 619747
BANF1	100%	100%	100%	99.8%	98.5%	Nestor-Guillermo progeria syndrome, 614008
BBIP1	100%	100%	100%	100%	99.7%	Bardet-Biedl syndrome 18, 615995
BBS1	93.7%	93.7%	100%	100%	99.3%	Bardet-Biedl syndrome 1, 209900
BBS10	100%	100%	100%	100%	99.8%	Bardet-Biedl syndrome 10, 615987
BBS12	100%	100%	100%	100%	100%	Bardet-Biedl syndrome 12, 615989
BBS2	98%	98%	100%	100%	99.8%	Retinitis pigmentosa 74, 616562;Bardet-Biedl syndrome 2, 615981
BBS4	100%	100%	100%	100%	99.7%	Bardet-Biedl syndrome 4, 615982
BBS5	100%	100%	100%	100%	99.9%	Bardet-Biedl syndrome 5, 615983
BBS7	100%	100%	100%	100%	99.9%	Bardet-Biedl syndrome 7, 615984
BBS9	91.9%	91.9%	100%	100%	99.7%	Bardet-Biedl syndrome 9, 615986
BCAS3	100%	100%	100%	100%	99.8%	Hengel-Maroofian-Schols syndrome, 619641
BCAT2	100%	100%	100%	100%	99.4%	Hypervalinemia and hyperleucine-isoleucinemia, 618850
BCKDHA	86.3%	86.3%	100%	99.9%	99.3%	Maple syrup urine disease, type Ia, 248600
BCKDHB	89.9%	87.7%	100%	99.9%	99.3%	Maple syrup urine disease, type Ib, 620698
BCKDK	100%	100%	100%	100%	99.6%	Branched-chain keto acid dehydrogenase kinase deficiency, 614923

BCL10	100%	100%	100%	100%	99.7%	{Lymphoma, follicular, somatic}, 605027;?Immunodeficiency 37, 616098;{Sezary syndrome, somatic};{Male germ cell tumor, somatic}, 273300;Lymphoma, MALT, somatic, 137245;{Mesothelioma, somatic}, 156240
BCS1L	100%	100%	100%	100%	99.6%	GRACILE syndrome, 603358;Mitochondrial complex III deficiency, nuclear type 1, 124000;Bjornstad syndrome, 262000
BET1	100%	100%	100%	100%	99.8%	Muscular dystrophy, congenital, with rapid progression, 254100
BFSP1	100%	100%	100%	99.9%	99.4%	Cataract 33, multiple types, 611391
BFSP2	100%	100%	100%	99.9%	99.1%	Cataract 12, multiple types, 611597
BHLHA9	100%	100%	100%	100%	99.4%	?Camptosynpolydactyly, complex, 607539;Syndactyly, mesoaxial synostotic, with phalangeal reduction, 609432
BIN1	100%	100%	100%	100%	99.3%	Centronuclear myopathy 2, 255200
BLM	94.7%	94.6%	100%	100%	99.9%	Bloom syndrome, 210900
BLOC1S3	100%	100%	100%	100%	98.5%	Hermansky-Pudlak syndrome 8, 614077
BLOC1S5	100%	100%	100%	100%	99.7%	Hermansky-Pudlak syndrome 11, 619172
BLOC1S6	100%	100%	100%	100%	99.9%	Hermansky-Pudlak syndrome 9, 614171
BLTP1	100%	100%	100%	100%	99.8%	Alkuraya-Kucinskas syndrome, 617822

BLVRA	95.2%	95.2%	100%	100%	99.8%	Hyperbiliverdinemia, 614156
BMP1	100%	100%	100%	100%	99.3%	Osteogenesis imperfecta, type XIII, 614856
BMPER	100%	100%	100%	100%	99.6%	Diaphanospondylodysostosis, 608022
BMPR1B	95.6%	94.1%	100%	100%	99.7%	Acromesomelic dysplasia 3, 609441;Brachydactyly, type A2, 112600;Brachydactyly, type A1, D, 616849
BOLA3	100%	100%	100%	100%	98.9%	Multiple mitochondrial dysfunctions syndrome 2 with hyperglycinemia, 614299
BORCS8	84.1%	84.1%	100%	99.7%	98.8%	Neurodegeneration, infantile-onset, with optic atrophy and brain abnormalities, 620987
BPGM	100%	100%	100%	100%	99.9%	Erythrocytosis, familial, 8, 222800
BPNT2	94.9%	94.9%	100%	100%	99.5%	Chondrodysplasia with joint dislocations, GPAPP type, 614078
BRAT1	97.3%	96.5%	100%	99.9%	99.3%	Neurodevelopmental disorder with cerebellar atrophy and with or without seizures, 618056;Rigidity and multifocal seizure syndrome, lethal neonatal, 614498
BRCA1	100%	100%	100%	100%	99.8%	Fanconi anemia, complementation group S, 617883;{Breast-ovarian cancer, familial, 1}, 604370;{Pancreatic cancer, susceptibility to, 4}, 614320

BRCA2	100%	100%	100%	100%	99.8%	Fanconi anemia, complementation group D1, 605724;{Glioblastoma 3}, 613029;{Medulloblastoma}, 155255;{Prostate cancer}, 176807;{Breast-ovarian cancer, familial, 2}, 612555;{Breast cancer, male, susceptibility to}, 114480;{Pancreatic cancer 2}, 613347;Wilms tumor, 194070
BRF1	100%	100%	100%	100%	99.5%	Cerebellofaciodental syndrome, 616202
BRIP1	96%	96%	100%	100%	99.8%	Fanconi anemia, complementation group J, 609054;{Breast cancer, early-onset, susceptibility to}, 114480
BSCL2	100%	100%	100%	100%	99.6%	Lipodystrophy, congenital generalized, type 2, 269700;Neuronopathy, distal hereditary motor, autosomal dominant 13, 619112;Silver spastic paraplegia syndrome, 270685;Encephalopathy, progressive, with or without lipodystrophy, 615924
BSND	100%	100%	100%	100%	99.8%	Sensorineural deafness with mild renal dysfunction, 602522;Bartter syndrome, type 4a, 602522
BTD	94.2%	94.2%	100%	100%	99.9%	Biotinidase deficiency, 253260

BUB1	97.3%	97.3%	100%	100%	99.7%	Colorectal cancer with chromosomal instability, somatic, 114500;Microcephaly 30, primary, autosomal recessive, 620183
BUB1B	96.3%	96.3%	100%	100%	99.8%	Colorectal cancer, somatic, 114500;[Premature chromatid separation trait], 176430;Mosaic variegated aneuploidy syndrome 1, 257300
BUD13	100%	100%	100%	100%	99.4%	Achalasia-progeroid syndrome, 621123
C12orf57	100%	100%	100%	99.8%	98.3%	Temtamy syndrome, 218340
C19orf12	100%	100%	100%	99.9%	98.7%	Neurodegeneration with brain iron accumulation 4, 614298;?Spastic paraplegia 43, autosomal recessive, 615043
C1QA	76.9%	73.9%	100%	100%	99.6%	C1q deficiency 1, 613652
C1QB	77.2%	77.2%	100%	100%	99.3%	C1q deficiency 2, 620321
C1QBP	100%	100%	100%	100%	99.6%	Combined oxidative phosphorylation deficiency 33, 617713
C1QC	99.7%	98%	100%	100%	98.7%	C1q deficiency 3, 620322
C1S	100%	100%	100%	100%	99.5%	C1s deficiency, 613783;Ehlers-Danlos syndrome, periodontal type, 2, 617174
C2CD3	96%	96%	100%	100%	99.5%	Orofaciodigital syndrome XIV, 615948
C2orf69	100%	100%	100%	100%	99.9%	Combined oxidative phosphorylation deficiency 53, 619423

C3	97%	97%	100%	99.9%	99.2%	C3 deficiency, 613779;{Hemolytic uremic syndrome, atypical, susceptibility to, 5}, 612925;{Macular degeneration, age-related, 9}, 611378
C4A	99.5%	99.1%	99.2%	97.9%	94.6%	[Blood group, Rodgers], 614374;C4a deficiency, 614380
C4B	100%	99.9%	99.2%	97.8%	94.3%	C4B deficiency, 614379
C5	100%	100%	100%	100%	99.8%	C5 deficiency, 609536;[Eculizumab, poor response to], 615749
C6	100%	100%	100%	100%	99.9%	C6 deficiency, 612446
C8A	100%	100%	100%	100%	99.9%	C8 deficiency, type I, 613790
C8B	100%	100%	100%	100%	99.8%	C8 deficiency, type II, 613789
CA12	100%	100%	100%	100%	99.5%	Hyperchlorhidrosis, isolated, 143860
CA2	100%	100%	100%	100%	99.8%	Osteopetrosis, autosomal recessive 3, with renal tubular acidosis, 259730
CA5A	84.7%	84.7%	100%	100%	99.6%	Hyperammonemia due to carbonic anhydrase VA deficiency, 615751
CA8	100%	100%	100%	100%	99.9%	Spinocerebellar ataxia, autosomal recessive 34, 613227
CABP2	100%	100%	100%	99.7%	97.3%	Deafness, autosomal recessive 93, 614899
CABP4	100%	100%	100%	100%	99.5%	Cone-rod synaptic disorder, congenital nonprogressive, 610427
CACHD1	100%	100%	100%	100%	99.7%	Davis-Wells syndrome, 621566

CACNA1B	97.2%	97.2%	100%	99.9%	99%	Neurodevelopmental disorder with seizures and nonepileptic hyperkinetic movements, 618497
CACNA1D	100%	100%	100%	100%	99.7%	Primary aldosteronism, seizures, and neurologic abnormalities, 615474;Sinoatrial node dysfunction and deafness, 614896
CACNA1S	100%	100%	100%	100%	99.4%	{Thyrotoxic periodic paralysis, susceptibility to, 1}, 188580;Congenital myopathy 18 due to dihydropyridine receptor defect, 620246;Hypokalemic periodic paralysis, type 1, 170400;{Malignant hyperthermia susceptibility 5}, 601887
CACNA2D1	100%	100%	100%	100%	99.4%	Developmental and epileptic encephalopathy 110, 620149
CACNA2D2	100%	100%	100%	100%	99.3%	Cerebellar atrophy with seizures and variable developmental delay, 618501
CAD	99.7%	98.5%	100%	100%	99.4%	Developmental and epileptic encephalopathy 50, 616457
CAMK2A	100%	100%	100%	100%	99.5%	Intellectual developmental disorder, autosomal dominant 53, 617798;?Intellectual developmental disorder, autosomal recessive 63, 618095

CAMSAP1	100%	100%	100%	99.9%	99.3%	Cortical dysplasia, complex, with other brain malformations 12, 620316
CANT1	100%	100%	100%	100%	99.7%	Desbuquois dysplasia 1, 251450;Epiphyseal dysplasia, multiple, 7, 617719
CAP2	82%	82%	100%	99.9%	99.8%	Cardiomyopathy, dilated, 2I, 620462
CAPN1	100%	100%	100%	100%	99.2%	Spastic paraplegia 76, autosomal recessive, 616907
CAPN10	100%	100%	100%	99.9%	99.4%	{Diabetes mellitus, noninsulin-dependent 1}, 601283
CAPN15	100%	100%	100%	99.9%	98.9%	Oculogastrointestinal neurodevelopmental syndrome, 619318
CAPN3	100%	100%	100%	100%	99.5%	Muscular dystrophy, limb-girdle, autosomal recessive 1, 253600;Muscular dystrophy, limb-girdle, autosomal dominant 4, 618129
CARD11	100%	100%	100%	100%	99.5%	B-cell expansion with NFKB and T-cell anergy, 616452;Immunodeficiency 11B with atopic dermatitis, 617638;Immunodeficiency 11A, 615206
CARD9	100%	100%	100%	100%	99.6%	Immunodeficiency 103, susceptibility to fungal infection, 212050
CARMIL2	100%	100%	100%	100%	99.2%	Immunodeficiency 58, 618131
CARS1	100%	100%	100%	100%	99.6%	Microcephaly, developmental delay, and brittle hair syndrome, 618891

CARS2	100%	99.9%	100%	100%	99.2%	Combined oxidative phosphorylation deficiency 27, 616672
CASP14	100%	100%	100%	100%	99.6%	Ichthyosis, congenital, autosomal recessive 12, 617320
CASP2	100%	99.9%	100%	100%	99.7%	Intellectual developmental disorder, autosomal recessive 80, with variant lissencephaly, 620653
CASP8	97%	97%	100%	100%	99.7%	{Breast cancer, protection against}, 114480;?Caspase 8 lymphadenopathy syndrome, 607271;Hepatocellular carcinoma, somatic, 114550;{Lung cancer, protection against}, 211980
CASQ2	87.5%	87.5%	100%	100%	99.8%	Ventricular tachycardia, catecholaminergic polymorphic, 2, 611938
CASR	100%	100%	100%	100%	99.5%	Hypocalcemia, autosomal dominant, with Bartter syndrome, 601198;Hyperparathyroidism, neonatal, 239200;Hypocalcemia, autosomal dominant, 601198;Hypocalciuric hypercalcemia, type I, 145980;{?Epilepsy idiopathic generalized, susceptibility to, 8}, 612899
CAST	100%	100%	100%	100%	99.8%	Peeling skin with leukonychia, acral punctate keratoses, cheilitis, and knuckle pads, 616295
CAT	98.1%	98.1%	100%	100%	99.8%	Acatalasemia, 614097
CATSPER1	100%	100%	100%	100%	98.9%	Spermatogenic failure 7, 612997

CAV1	64.8%	64.8%	100%	100%	98.8%	Lipodystrophy, congenital generalized, type 3, 612526;Pulmonary hypertension, primary, 3, 615343;Lipodystrophy, familial partial, type 7, 606721
CAVIN1	100%	100%	100%	99.6%	97.4%	Lipodystrophy, congenital generalized, type 4, 613327
CBLB	97.1%	97.1%	100%	100%	99.8%	Autoimmune disease, multisystem, infantile-onset, 3, 620430
CBLIF	100%	100%	100%	100%	99.8%	Intrinsic factor deficiency, 261000
CBS	87.7%	85.5%	100%	100%	99.5%	Thrombosis, hyperhomocysteinemic, 236200;Homocystinuria, B6-responsive and nonresponsive types, 236200
CC2D1A	100%	100%	100%	99.9%	98.9%	Intellectual developmental disorder, autosomal recessive 3, 608443
CC2D2A	98.2%	98.2%	100%	100%	99.8%	COACH syndrome 2, 619111;Retinitis pigmentosa 93, 619845;Meckel syndrome 6, 612284;Joubert syndrome 9, 612285
CCBE1	100%	100%	100%	100%	99.7%	Hennekam lymphangiectasia-lymphedema syndrome 1, 235510
CCDC134	100%	100%	100%	100%	99.7%	Osteogenesis imperfecta, type XXII, 619795
CCDC174	100%	100%	100%	100%	99.8%	Hypotonia, infantile, with psychomotor retardation, 616816

CCDC32	100%	100%	100%	99.9%	99.5%	Cardiofacioneurodevelopmental syndrome, 619123
CCDC39	100%	100%	100%	100%	99.9%	Ciliary dyskinesia, primary, 14, 613807
CCDC40	100%	100%	100%	99.9%	99%	Ciliary dyskinesia, primary, 15, 613808
CCDC47	100%	100%	100%	100%	99.9%	Trichohepatoneurodevelopmental syndrome, 618268
CCDC65	100%	100%	100%	99.9%	99.4%	
CCDC8	99.8%	97.4%	100%	100%	99.5%	3-M syndrome 3, 614205
CCDC88A	97.4%	97.4%	100%	100%	99.7%	PEHO syndrome-like, 617507
CCDC88C	100%	100%	100%	99.9%	98.8%	?Spinocerebellar ataxia 40, 616053;Hydrocephalus, congenital, 1, 236600
CCN2	100%	100%	100%	100%	99.7%	?Spondyloepimetaphyseal dysplasia, Li-Shao-Li type, 621099;Kyphomelic dysplasia, 211350
CCN6	100%	100%	100%	100%	99.7%	Progressive pseudorheumatoid dysplasia, 208230
CCNO	100%	100%	100%	100%	99.7%	Ciliary dyskinesia, primary, 29, 615872
CCR2	100%	100%	100%	100%	99.9%	{HIV infection, susceptibility/resistance to}, 609423;Polycystic lung disease, 219600
CCT5	100%	100%	100%	100%	99.5%	?Neuropathy, hereditary sensory, with spastic paraplegia, 256840
CD151	100%	100%	100%	100%	99.7%	[Blood group, Raph], 179620;Epidermolysis bullosa simplex 7, with nephropathy and deafness, 609057

CD19	100%	100%	100%	100%	99%	Immunodeficiency, common variable, 3, 613493
CD247	77.2%	71.4%	100%	100%	99.5%	?Immunodeficiency 25, 610163
CD27	100%	100%	100%	100%	99.7%	Lymphoproliferative syndrome 2, 615122
CD2AP	100%	100%	100%	100%	99.8%	Glomerulosclerosis, focal segmental, 3, 607832
CD320	73.6%	73.6%	100%	100%	99.3%	Methylmalonic aciduria, transient, due to transcobalamin receptor defect, 613646
CD3D	100%	100%	100%	100%	99.7%	Immunodeficiency 19, severe combined, 615617
CD3E	100%	100%	100%	100%	99.8%	Immunodeficiency 18, 615615;Immunodeficiency 18, SCID variant, 615615
CD3G	100%	100%	100%	100%	99.7%	Immunodeficiency 17, CD3 gamma deficient, 615607
CD4	100%	100%	100%	100%	99.6%	Immunodeficiency 79, 619238;OKT4 epitope deficiency, 613949
CD40	100%	100%	100%	100%	99.2%	Immunodeficiency with hyper-IgM, type 3, 606843
CD55	96.6%	93.7%	100%	100%	99.5%	[Blood group Cromer], 613793;Complement hyperactivation, angiopathic thrombosis, and protein-losing enteropathy, 226300
CD59	89.3%	89.3%	100%	100%	100%	Hemolytic anemia, CD59-mediated, with or without immune-mediated polyneuropathy, 612300

CD70	100%	100%	100%	100%	99.1%	Lymphoproliferative syndrome 3, 618261
CD79A	100%	100%	100%	100%	98.9%	Agammaglobulinemia 3, 613501
CD79B	100%	100%	100%	100%	99.5%	Agammaglobulinemia 6, 612692
CD81	100%	99.6%	100%	100%	99.4%	Immunodeficiency, common variable, 6, 613496
CD8A	100%	100%	100%	100%	99.5%	Immunodeficiency 116, 608957
CDAN1	100%	100%	100%	100%	98.9%	Dyserythropoietic anemia, congenital, type Ia, 224120
CDC14A	96.9%	96.9%	100%	100%	99.8%	Deafness, autosomal recessive 32, with or without immotile sperm, 608653
CDC45	100%	100%	100%	100%	99.5%	Meier-Gorlin syndrome 7, 617063
CDC6	100%	100%	100%	100%	99.4%	Meier-Gorlin syndrome 5, 613805
CDCA7	100%	100%	100%	100%	99.7%	Immunodeficiency-centromeric instability-facial anomalies syndrome 3, 616910
CDH11	100%	100%	100%	100%	99.9%	Teebi hypertelorism syndrome 2, 619736;Elsahy-Waters syndrome, 211380
CDH23	100%	100%	100%	100%	99.4%	Usher syndrome, type 1D, 601067;{Pituitary adenoma 5, multiple types}, 617540;Usher syndrome, type 1D/F digenic, 601067;Deafness, autosomal recessive 12, 601386

CDH3	100%	100%	100%	100%	99.6%	Hypotrichosis, congenital, with juvenile macular dystrophy, 601553;Ectodermal dysplasia, ectrodactyly, and macular dystrophy, 225280
CDHR1	100%	100%	100%	100%	99.7%	Macular dystrophy, retinal, 613660;Cone-rod dystrophy 15, 613660;Retinitis pigmentosa 65, 613660
CDIN1	100%	100%	100%	100%	99.7%	Dyserythropoietic anemia, congenital, type Ib, 615631
CDK10	100%	100%	100%	100%	99.6%	Al Kaissi syndrome, 617694
CDK4	100%	100%	100%	100%	99.6%	{Melanoma, cutaneous malignant, 3}, 609048;Microcephaly 31, primary, autosomal recessive, 621507
CDK5	100%	100%	100%	100%	99.4%	?Lissencephaly 7 with cerebellar hypoplasia, 616342
CDK5RAP2	100%	100%	100%	100%	99.7%	Microcephaly 3, primary, autosomal recessive, 604804
CDK6	100%	100%	100%	100%	99%	?Microcephaly 12, primary, autosomal recessive, 616080
CDSN	100%	100%	100%	100%	99.5%	Hypotrichosis 2, 146520;Peeling skin syndrome 1, 270300
CDT1	100%	100%	100%	100%	99.5%	Meier-Gorlin syndrome 4, 613804
CEACAM16	100%	100%	100%	100%	99.6%	Deafness, autosomal dominant 4B, 614614;Deafness, autosomal recessive 113, 618410

CEBPE	100%	100%	100%	100%	99.4%	?Immunodeficiency 108 with autoinflammation, 260570;Specific granule deficiency, 245480
CENPE	100%	100%	100%	99.9%	99.5%	?Microcephaly 13, primary, autosomal recessive, 616051
CENPF	98.6%	98.6%	100%	100%	99.8%	Stromme syndrome, 243605
CEP104	95.1%	95.1%	100%	100%	99.6%	Joubert syndrome 25, 616781;Intellectual developmental disorder, autosomal recessive 77, 619988
CEP120	100%	100%	100%	100%	99.6%	Short-rib thoracic dysplasia 13 with or without polydactyly, 616300;Joubert syndrome 31, 617761
CEP135	95.8%	95.8%	100%	100%	99.7%	Microcephaly 8, primary, autosomal recessive, 614673
CEP152	100%	100%	100%	100%	99.9%	Microcephaly 9, primary, autosomal recessive, 614852;Seckel syndrome 5, 613823
CEP164	100%	100%	100%	100%	99.5%	Nephronophthisis 15, 614845
CEP19	100%	100%	100%	100%	99.7%	?Morbid obesity and spermatogenic failure, 615703
CEP290	100%	100%	100%	100%	99.8%	Leber congenital amaurosis 10, 611755;Joubert syndrome 5, 610188;Senior-Loken syndrome 6, 610189;?Bardet-Biedl syndrome 14, 615991;Meckel syndrome 4, 611134
CEP41	100%	100%	100%	100%	99.8%	Joubert syndrome 15, 614464

CEP55	100%	100%	100%	100%	99.9%	Multinucleated neurons, anhydramnios, renal dysplasia, cerebellar hypoplasia, and hydranencephaly, 236500
CEP57	100%	100%	100%	100%	99.7%	Mosaic variegated aneuploidy syndrome 2, 614114
CEP63	92.8%	92.8%	100%	100%	99.7%	?Seckel syndrome 6, 614728
CEP78	100%	100%	100%	100%	99.6%	Cone-rod dystrophy and hearing loss, 617236
CEP83	100%	100%	100%	100%	99.8%	Nephronophthisis 18, 615862
CERKL	100%	100%	100%	100%	99.8%	Retinitis pigmentosa 26, 608380
CERS1	100%	99.6%	100%	100%	99%	Epilepsy, progressive myoclonic, 8, 616230
CERS3	100%	100%	100%	100%	99.8%	Ichthyosis, congenital, autosomal recessive 9, 615023
CFAP20	100%	100%	100%	100%	99.5%	
CFAP298	89.6%	86.3%	100%	100%	99.5%	Ciliary dyskinesia, primary, 26, 615500
CFAP300	100%	100%	100%	100%	100%	Ciliary dyskinesia, primary, 38, 618063
CFAP410	100%	100%	100%	100%	99.3%	Retinal dystrophy with macular staphyloma, 617547;Spondylometaphyseal dysplasia, axial, 602271
CFAP418	100%	100%	100%	100%	99.5%	Retinitis pigmentosa 64, 614500;Cone-rod dystrophy 16, 614500;Bardet-Biedl syndrome 21, 617406
CFAP53	100%	100%	100%	100%	99.6%	Heterotaxy, visceral, 6, autosomal recessive, 614779

CFD	100%	100%	100%	100%	98%	Complement factor D deficiency, 613912
CFH	97.5%	97.5%	100%	100%	99.9%	{Macular degeneration, age-related, 4}, 610698;Basal laminar drusen, 126700;Complement factor H deficiency, 609814;{Hemolytic uremic syndrome, atypical, susceptibility to, 1}, 235400
CFI	98.6%	95.9%	100%	100%	99.8%	{Hemolytic uremic syndrome, atypical, susceptibility to, 3}, 612923;{Macular degeneration, age-related, 13, susceptibility to}, 615439;Complement factor I deficiency, 610984
CFL2	100%	100%	100%	100%	100%	Nemaline myopathy 7, autosomal recessive, 610687
CFTR	100%	100%	100%	100%	99.6%	Cystic fibrosis, 219700;Sweat chloride elevation without CF;Congenital bilateral absence of vas deferens, 277180;{Pancreatitis, hereditary}, 167800;{Bronchiectasis with or without elevated sweat chloride 1, modifier of}, 211400;{Hypertrypsinemia, neonatal}
CHAT	100%	100%	100%	100%	99.6%	Myasthenic syndrome, congenital, 6, presynaptic, 254210
CHKA	100%	100%	100%	100%	99.1%	Neurodevelopmental disorder with microcephaly, movement abnormalities, and seizures, 620023

CHKB	100%	100%	100%	100%	99.5%	Muscular dystrophy, congenital, megaconial type, 602541
CHMP1A	100%	100%	100%	99.9%	99.2%	Pontocerebellar hypoplasia, type 8, 614961
CHP1	100%	100%	100%	100%	99.5%	?Spastic ataxia 9, autosomal recessive, 618438
CHRM3	100%	100%	100%	100%	99.8%	Prune belly syndrome, 100100
CHRNA1	100%	100%	100%	100%	99.6%	Myasthenic syndrome, congenital, 1B, fast-channel, 608930;Myasthenic syndrome, congenital, 1A, slow-channel, 601462;Multiple pterygium syndrome, lethal type, 253290
CHRNA3	92%	92%	100%	99.9%	99.4%	{Lung cancer susceptibility 2}, 612052;Bladder dysfunction, autonomic, with impaired pupillary reflex and secondary CAKUT, 191800
CHRNB1	100%	100%	100%	99.8%	98.5%	?Myasthenic syndrome, congenital, 2C, associated with acetylcholine receptor deficiency, 616314;Myasthenic syndrome, congenital, 2A, slow-channel, 616313

CHRND	100%	100%	100%	100%	99.5%	?Myasthenic syndrome, congenital, 3C, associated with acetylcholine receptor deficiency, 616323;Multiple pterygium syndrome, lethal type, 253290;Myasthenic syndrome, congenital, 3B, fast-channel, 616322;?Myasthenic syndrome, congenital, 3A, slow-channel, 616321
CHRNE	100%	100%	100%	99.9%	98.9%	Myasthenic syndrome, congenital, 4A, slow-channel, 605809;Myasthenic syndrome, congenital, 4C, associated with acetylcholine receptor deficiency, 608931;Myasthenic syndrome, congenital, 4B, fast-channel, 616324
CHRNG	100%	100%	100%	99.9%	99.6%	Multiple pterygium syndrome, lethal type, 253290;Escobar syndrome, 265000
CHST11	100%	100%	100%	100%	99.7%	?Osteochondrodysplasia, brachydactyly, and overlapping malformed digits, 618167
CHST14	100%	100%	100%	99.9%	98.4%	Ehlers-Danlos syndrome, musculocontractural type 1, 601776
CHST3	100%	100%	100%	100%	98.9%	Spondyloepiphyseal dysplasia with congenital joint dislocations, 143095
CHST6	100%	100%	100%	100%	99.6%	Macular corneal dystrophy, 217800
CHST8	100%	100%	100%	99.9%	99.1%	

CHSY1	100%	99.9%	100%	99.8%	99.1%	Temtamy preaxial brachydactyly syndrome, 605282
CIAO1	100%	100%	100%	99.7%	98.8%	Multiple mitochondrial dysfunctions syndrome 10, 620960
CIB2	100%	100%	100%	100%	99.9%	Deafness, autosomal recessive 48, 609439
CIDEC	100%	100%	100%	100%	99.8%	?Lipodystrophy, familial partial, type 5, 615238
CIITA	100%	100%	100%	100%	99.2%	{Rheumatoid arthritis, susceptibility to}, 180300;MHC class II deficiency 1, 209920
CILK1	100%	100%	100%	100%	99.8%	Cranioectodermal dysplasia 6, 621337;{Epilepsy, juvenile myoclonic, susceptibility to, 10}, 617924;Endocrine-cerebroosteodysplasia, 612651
CIROP	98.3%	93.9%	100%	100%	99.7%	Heterotaxy, visceral, 12, autosomal, 619702
CIROZ	100%	100%	100%	100%	99.7%	Heterotaxy, visceral, 14, autosomal, 621080
CISD2	100%	100%	100%	99.9%	99.4%	Wolfram syndrome 2, 604928
CIT	95.8%	95.8%	100%	100%	99.4%	Microcephaly 17, primary, autosomal recessive, 617090
CKAP2L	100%	100%	100%	100%	99.7%	Filippi syndrome, 272440
CLCF1	100%	100%	100%	100%	99.8%	Cold-induced sweating syndrome 2, 610313
CLCN1	100%	100%	100%	100%	99.6%	Myotonia congenita, recessive, 255700;Myotonia congenita, dominant, 160800;Myotonia levior, 160800

CLCN2	100%	99.1%	100%	100%	99.6%	Leukoencephalopathy with ataxia, 615651;Hyperaldosteronism, familial, type II, 605635;{Epilepsy, juvenile myoclonic, susceptibility to, 8}, 607628;{Epilepsy, juvenile absence, susceptibility to, 2}, 607628;{Epilepsy, idiopathic generalized, susceptibility to, 11}, 607628
CLCN3	100%	100%	100%	100%	99.8%	?Neurodevelopmental disorder with seizures and brain abnormalities, 619517;Neurodevelopmental disorder with hypotonia and brain abnormalities, 619512
CLCN7	97.5%	97.5%	100%	100%	99.7%	Hypopigmentation, organomegaly, and delayed myelination and development, 618541;Osteopetrosis, autosomal recessive 4, 611490;Osteopetrosis, autosomal dominant 2, 166600
CLCNKB	98.6%	97.4%	100%	100%	99.5%	Bartter syndrome, type 3, 607364;Bartter syndrome, type 4b, digenic, 613090
CLDN1	100%	100%	100%	100%	99.9%	Ichthyosis, leukocyte vacuoles, alopecia, and sclerosing cholangitis, 607626
CLDN10	100%	100%	100%	100%	99.8%	HELIX syndrome, 617671
CLDN14	100%	100%	100%	100%	99.4%	Deafness, autosomal recessive 29, 614035
CLDN16	95%	95%	100%	100%	99.8%	Hypomagnesemia 3, renal, 248250

CLDN19	100%	100%	100%	100%	99.5%	Hypomagnesemia 5, renal, with ocular involvement, 248190
CLDN9	100%	100%	100%	100%	99.2%	Deafness, autosomal recessive 116, 619093
CLIP1	100%	100%	100%	100%	99.8%	
CLMP	100%	100%	100%	100%	99.7%	Congenital short bowel syndrome, 615237
CLN3	93.1%	93.1%	100%	100%	99.4%	Ceroid lipofuscinosis, neuronal, 3, 204200;Retinitis pigmentosa 101, 621548
CLN5	83%	83%	100%	100%	99.8%	Ceroid lipofuscinosis, neuronal, 5, 256731
CLN6	95.1%	95.1%	100%	100%	99.4%	Ceroid lipofuscinosis, neuronal, 6B (Kufs type), 204300;Ceroid lipofuscinosis, neuronal, 6A, 601780
CLN8	100%	100%	100%	100%	99.5%	Ceroid lipofuscinosis, neuronal, 8, Northern epilepsy variant, 610003;Ceroid lipofuscinosis, neuronal, 8, 600143
CLP1	100%	100%	100%	100%	99.5%	Pontocerebellar hypoplasia, type 10, 615803
CLPB	98.2%	98.2%	100%	100%	99.7%	Neutropenia, severe congenital, 9, autosomal dominant, 619813;3-methylglutaconic aciduria, type VIIB, autosomal recessive, 616271;3-methylglutaconic aciduria, type VIIA, autosomal dominant, 619835
CLPP	100%	100%	100%	100%	98.9%	Perrault syndrome 3, 614129
CLRN1	100%	100%	100%	99.9%	99.1%	Usher syndrome, type 3A, 276902;Retinitis pigmentosa 61, 614180

CLRN2	100%	100%	100%	100%	99.6%	Deafness, autosomal recessive 117, 619174
CLXN	100%	100%	100%	100%	99.8%	Ciliary dyskinesia, primary, 53, 620642
CMPK2	100%	100%	100%	100%	99.1%	Basal ganglia calcification, idiopathic, 10, autosomal recessive, 621018
CNGA1	100%	100%	100%	100%	99.7%	Retinitis pigmentosa 49, 613756
CNGA3	100%	100%	100%	100%	99.2%	Achromatopsia 2, 216900
CNGB1	100%	100%	100%	100%	99.4%	Retinitis pigmentosa 45, 613767
CNGB3	100%	100%	100%	99.9%	99.5%	Achromatopsia 3, 262300
CNNM2	100%	100%	100%	100%	99.4%	Hypomagnesemia 6, renal, 613882;Hypomagnesemia, seizures, and impaired intellectual development 1, 616418
CNNM4	100%	100%	100%	100%	99.4%	Jalili syndrome, 217080
CNPY3	99.1%	95.7%	100%	100%	99.3%	Developmental and epileptic encephalopathy 60, 617929
CNTN1	100%	100%	100%	100%	99.8%	Congenital myopathy 12, 612540
CNTN2	100%	99.9%	100%	99.9%	99.5%	Epilepsy, early-onset, 5, with or without developmental delay, 615400
CNTNAP1	100%	100%	100%	100%	99.1%	Lethal congenital contracture syndrome 7, 616286;Hypomyelinating neuropathy, congenital, 3, 618186

CNTNAP2	100%	100%	100%	100%	99.7%	Pitt-Hopkins like syndrome 1, 610042;{Autism susceptibility 15}, 612100
COA5	69.5%	69.5%	100%	99.8%	98.1%	?Mitochondrial complex IV, deficiency, nuclear type 9, 616500
COA6	100%	100%	100%	100%	99.6%	Mitochondrial complex IV deficiency, nuclear type 13, 616501
COA7	100%	100%	100%	100%	99.6%	Spinocerebellar ataxia, autosomal recessive, with axonal neuropathy 3, 618387
COA8	100%	100%	100%	100%	99.7%	Mitochondrial complex IV deficiency, nuclear type 17, 619061
COASY	100%	100%	100%	99.8%	99.1%	Pontocerebellar hypoplasia, type 12, 618266;Neurodegeneration with brain iron accumulation 6, 615643
COCH	100%	100%	100%	100%	99.9%	Deafness, autosomal dominant 9, 601369;?Deafness, autosomal recessive 110, 618094
COG1	100%	100%	100%	100%	99.4%	Congenital disorder of glycosylation, type IIg, 611209
COG3	100%	100%	100%	100%	99.8%	Congenital disorder of glycosylation, type IIbb, 620546
COG4	97%	97%	100%	100%	99.3%	Congenital disorder of glycosylation, type IIj, 613489;Saul-Wilson syndrome, 618150
COG5	96.8%	96.8%	100%	100%	99.8%	Congenital disorder of glycosylation, type IIi, 613612

COG6	100%	100%	100%	100%	99.9%	Shaheen syndrome, 615328;Congenital disorder of glycosylation, type III, 614576
COG7	95.1%	95.1%	100%	100%	99.6%	Congenital disorder of glycosylation, type IIe, 608779
COG8	100%	100%	100%	100%	99.6%	Congenital disorder of glycosylation, type IIh, 611182
COL11A1	100%	100%	100%	100%	99.8%	Fibrochondrogenesis 1, 228520;Stickler syndrome, type II, 604841;Marshall syndrome, 154780;Deafness, autosomal dominant 37, 618533;{Lumbar disc herniation, susceptibility to}, 603932
COL11A2	100%	100%	100%	100%	99.1%	Deafness, autosomal dominant 13, 601868;Otospondylomegaepiphyseal dysplasia, autosomal recessive, 215150;Fibrochondrogenesis 2, 614524;Deafness, autosomal recessive 53, 609706;Otospondylomegaepiphyseal dysplasia, autosomal dominant, 184840
COL12A1	100%	100%	100%	100%	99.8%	Bethlem myopathy 2, 616471;?Ullrich congenital muscular dystrophy 2, 616470
COL13A1	100%	100%	100%	100%	99.4%	Myasthenic syndrome, congenital, 19, 616720
COL17A1	100%	100%	100%	100%	99.4%	Epithelial recurrent erosion dystrophy, 122400;Epidermolysis bullosa, junctional 4, intermediate, 619787

COL18A1	99.2%	98%	100%	99.9%	99.2%	Knobloch syndrome, type 1, 267750;Glaucoma, primary closed-angle, 618880
COL1A2	100%	100%	100%	100%	99.8%	Osteogenesis imperfecta, type III, 259420;{Osteoporosis, postmenopausal}, 166710;Ehlers-Danlos syndrome, arthrochalasia type, 2, 617821;Combined osteogenesis imperfecta and Ehlers-Danlos syndrome 2, 619120;Ehlers-Danlos syndrome, cardiac valvular type, 225320;Osteogenesis imperfecta, type IV, 166220;Osteogenesis imperfecta, type II, 166210
COL25A1	100%	100%	100%	100%	99.7%	Fibrosis of extraocular muscles, congenital, 5, 616219
COL27A1	100%	100%	100%	100%	99.4%	Steel syndrome, 615155
COL3A1	100%	100%	100%	100%	99.7%	Ehlers-Danlos syndrome, vascular type, 130050;Polymicrogyria with or without vascular-type EDS, 618343
COL4A3	100%	100%	100%	100%	99.6%	Alport syndrome 3A, autosomal dominant, 104200;Hematuria, benign familial, 2, 620320;Alport syndrome 3B, autosomal recessive, 620536

COL4A4	99.7%	98.9%	100%	100%	99.7%	Hematuria, familial benign, 1, 141200;Alport syndrome 2, autosomal recessive, 203780
COL6A1	100%	100%	100%	100%	99.4%	Ullrich congenital muscular dystrophy 1A, 254090;Bethlem myopathy 1A, 158810
COL6A2	100%	99.8%	100%	99.9%	99.2%	?Myosclerosis, congenital, 255600;Ullrich congenital muscular dystrophy 1B, 620727;Bethlem myopathy 1B, 620725
COL6A3	100%	100%	100%	100%	99.7%	Bethlem myopathy 1C, 620726;Ullrich congenital muscular dystrophy 1C, 620728;Dystonia 27, 616411
COL7A1	100%	100%	100%	100%	99.4%	Nail disorder, nonsyndromic congenital, 8, 607523;Epidermolysis bullosa dystrophica, Bart type, 132000;Epidermolysis bullosa dystrophica inversa, 226600;Epidermolysis bullosa dystrophica, autosomal recessive, 226600;Epidermolysis bullosa, pretibial, 131850;Epidermolysis bullosa dystrophica, autosomal dominant, 131750;Transient bullous of the newborn, 131705;Epidermolysis bullosa pruriginosa, 604129;Epidermolysis bullosa dystrophica, localisata variant, 226600

COL9A1	100%	100%	100%	100%	99.7%	Stickler syndrome, type IV, 614134;?Epiphyseal dysplasia, multiple, 6, 614135
COL9A2	100%	100%	100%	100%	99.3%	Epiphyseal dysplasia, multiple, 2, 600204;?Stickler syndrome, type V, 614284
COL9A3	100%	100%	100%	100%	99.4%	{Intervertebral disc disease, susceptibility to}, 603932;Epiphyseal dysplasia, multiple, 3, with or without myopathy, 600969;Stickler syndrome, type VI, 620022
COLEC10	95%	86.3%	100%	100%	99.8%	3MC syndrome 3, 248340
COLEC11	100%	100%	100%	100%	99.6%	3MC syndrome 2, 265050
COLGALT1	96%	92.4%	100%	100%	99%	Brain small vessel disease 3, 618360
COLQ	100%	100%	100%	100%	99.6%	Myasthenic syndrome, congenital, 5, 603034
COPB1	96.3%	96.3%	100%	99.9%	99.7%	Baralle-Macken syndrome, 619255
COPB2	96.7%	96.7%	100%	100%	99.8%	Osteoporosis, childhood- or juvenile-onset, with developmental delay, 619884;?Microcephaly 19, primary, autosomal recessive, 617800
COPZ1	86.8%	86.2%	100%	100%	99.8%	Neutropenia, severe congenital, 12, autosomal recessive, 621439
COQ2	96.3%	96.3%	100%	100%	99.5%	{Multiple system atrophy, susceptibility to}, 146500;Coenzyme Q10 deficiency, primary, 1, 607426

COQ4	96.3%	96.3%	100%	100%	99.6%	Coenzyme Q10 deficiency, primary, 7, 616276;Spastic ataxia 10, autosomal recessive, 620666
COQ6	100%	100%	100%	100%	99.6%	Coenzyme Q10 deficiency, primary, 6, 614650
COQ7	100%	100%	100%	100%	99.5%	Coenzyme Q10 deficiency, primary, 8, 616733;Neuronopathy, distal hereditary motor, autosomal recessive 9, 620402
COQ8A	89.1%	87.2%	100%	99.9%	99.5%	Coenzyme Q10 deficiency, primary, 4, 612016
COQ8B	92.1%	92.1%	100%	100%	99.6%	Nephrotic syndrome, type 9, 615573
COQ9	91.9%	91.9%	100%	99.9%	99.2%	Coenzyme Q10 deficiency, primary, 5, 614654
CORO1A	100%	100%	100%	100%	99.7%	Immunodeficiency 8, 615401
COX10	100%	100%	100%	100%	99.7%	Mitochondrial complex IV deficiency, nuclear type 3, 619046
COX11	100%	100%	100%	99.9%	98.7%	Mitochondrial complex IV deficiency, nuclear type 23, 620275
COX14	100%	100%	100%	100%	100%	?Mitochondrial complex IV deficiency, nuclear type 10, 619053
COX15	100%	100%	100%	100%	99.4%	Mitochondrial complex IV deficiency, nuclear type 6, 615119
COX16	61.9%	61.9%	100%	99.8%	99.4%	Mitochondrial complex IV deficiency, nuclear type 22, 619355

COX18	100%	100%	100%	99.9%	99.4%	?Mitochondrial complex IV deficiency, nuclear type 25, 621487;Charcot-Marie-Tooth disease, axonal, type 2MM, 621488
COX20	100%	100%	100%	100%	99.8%	Mitochondrial complex IV deficiency, nuclear type 11, 619054
COX4I1	100%	100%	100%	100%	99.4%	Mitochondrial complex IV deficiency, nuclear type 16, 619060
COX4I2	100%	99.1%	100%	100%	99.6%	Exocrine pancreatic insufficiency, dyserythropoietic anemia, and calvarial hyperostosis, 612714
COX5A	100%	100%	100%	100%	99.6%	Mitochondrial complex IV deficiency, nuclear type 20, 619064
COX6A1	100%	100%	100%	100%	99.5%	Charcot-Marie-Tooth disease, recessive intermediate D, 616039
COX6A2	100%	100%	100%	99.9%	97.6%	Mitochondrial complex IV deficiency, nuclear type 18, 619062
COX6B1	80.9%	80.9%	100%	99.8%	98.7%	Mitochondrial complex IV deficiency, nuclear type 7, 619051
COX8A	100%	100%	100%	99.7%	97.3%	?Mitochondrial complex IV deficiency, nuclear type 15, 619059
CP	100%	100%	100%	100%	99.9%	Aceruloplasminemia, 604290
CPA6	100%	100%	100%	100%	99.8%	Febrile seizures, familial, 11, 614418;Epilepsy, familial temporal lobe, 5, 614417
CPAMD8	100%	100%	100%	99.9%	99.3%	Anterior segment dysgenesis 8, 617319

CPAP	100%	100%	100%	100%	99.7%	Microcephaly 6, primary, autosomal recessive, 608393;Seckel syndrome 4, 613676
CPE	97.5%	97.5%	100%	99.9%	99.1%	BDV syndrome, 619326
CPLANE1	100%	100%	100%	100%	99.8%	Orofaciodigital syndrome VI, 277170;Joubert syndrome 17, 614615
CPLX1	100%	100%	100%	100%	99.6%	Developmental and epileptic encephalopathy 63, 617976
CPN1	100%	100%	100%	99.9%	99%	Carboxypeptidase N deficiency, 212070
CPOX	93.4%	93.4%	100%	99.9%	99%	Coproporphyria, 121300;Harderoporphyria, 618892
CPS1	100%	100%	100%	100%	99.7%	Carbamoylphosphate synthetase I deficiency, 237300
CPSF3	95.9%	95.9%	100%	100%	99.9%	Neurodevelopmental disorder with microcephaly, hypotonia, nystagmus, and seizures, 619876
CPT1A	100%	100%	100%	100%	99.5%	CPT deficiency, hepatic, type IA, 255120
CPT2	97.3%	96.3%	100%	100%	99.7%	{Encephalopathy, acute, infection-induced, 4, susceptibility to}, 614212;CPT II deficiency, infantile, 600649;CPT II deficiency, lethal neonatal, 608836;CPT II deficiency, myopathic, stress-induced, 255110

CR2	100%	100%	100%	100%	99.9%	{Systemic lupus erythematosus, susceptibility to, 9}, 610927;?Immunodeficiency, common variable, 7, 614699
CRADD	100%	100%	100%	100%	99.4%	Intellectual developmental disorder, autosomal recessive 34, with variant lissencephaly, 614499
CRAT	99.2%	96.9%	100%	99.8%	99.1%	?Neurodegeneration with brain iron accumulation 8, 617917
CRB1	98.6%	98.6%	100%	100%	99.9%	Leber congenital amaurosis 8, 613835;Retinitis pigmentosa-12, 600105;Pigmented paravenous chorioretinal atrophy, 172870
CRB2	100%	100%	100%	100%	99.6%	Focal segmental glomerulosclerosis 9, 616220;Ventriculomegaly with cystic kidney disease, 219730
CRBN	100%	99.6%	100%	100%	99.8%	Intellectual developmental disorder, autosomal recessive 2, 607417
CREB3L1	100%	100%	100%	100%	99.2%	Osteogenesis imperfecta, type XVI, 616229
CRELD1	100%	100%	100%	100%	99.6%	Atrioventricular septal defect, partial, with heterotaxy syndrome, 606217;Jeffries-Lakhani neurodevelopmental syndrome, 620771;{Atrioventricular septal defect, susceptibility to, 2}, 606217

CRIPT	100%	100%	100%	100%	99.9%	Rothmund-Thomson syndrome, type 3, 615789
CRLF1	94.8%	90.1%	99.7%	98.1%	93.3%	Cold-induced sweating syndrome 1, 272430
CRLS1	89.6%	84.4%	100%	99.7%	99.4%	Combined oxidative phosphorylation deficiency 57, 620167
CRPPA	100%	100%	100%	100%	99.3%	Muscular dystrophy-dystroglycanopathy (limb-girdle), type C, 7, 616052;Muscular dystrophy-dystroglycanopathy (congenital with brain and eye anomalies), type A, 7, 614643
CRTAP	100%	100%	100%	100%	99.2%	Osteogenesis imperfecta, type VII, 610682
CRYAA	100%	100%	100%	100%	99.3%	Cataract 9, multiple types, 604219
CRYAB	100%	100%	100%	100%	99.7%	Myopathy, myofibrillar, 2B, infantile-onset, 613869;Myopathy, myofibrillar, 2A, adult-onset, 608810;Cataract 16, multiple types, 613763;Cardiomyopathy, dilated, 1II, 615184
CRYBB1	100%	100%	100%	100%	99.5%	Cataract 17, multiple types, 611544
CRYBB3	100%	100%	100%	100%	99.6%	Cataract 22, 609741
CSF1R	97.3%	97.3%	100%	100%	99.6%	Brain abnormalities, neurodegeneration, and dysosteosclerosis, 618476;Leukoencephalopathy, diffuse hereditary, with spheroids 1, 221820
CSF2RB	100%	100%	100%	100%	99.5%	Surfactant metabolism dysfunction, pulmonary, 5, 614370

CSF3R	100%	100%	100%	100%	99.5%	Neutropenia, severe congenital, 7, autosomal recessive, 617014;?Neutrophilia, hereditary, 162830
CSGALNACT1	100%	100%	100%	100%	99.9%	Skeletal dysplasia, mild, with joint laxity and advanced bone age, 618870
CSPP1	96.9%	96.9%	100%	100%	99.6%	Joubert syndrome 21, 615636
CSTA	100%	100%	100%	100%	100%	Peeling skin syndrome 4, 607936
CSTB	100%	100%	100%	100%	99.4%	Epilepsy, progressive myoclonic 1A (Unverricht and Lundborg), 254800
CTC1	100%	100%	100%	100%	99.4%	Cerebroretinal microangiopathy with calcifications and cysts, 612199
CTDP1	90.8%	90.5%	100%	100%	99.2%	Congenital cataracts, facial dysmorphism, and neuropathy, 604168
CTNNA2	100%	100%	100%	100%	99.6%	Cortical dysplasia, complex, with other brain malformations 9, 618174
CTNS	100%	100%	100%	100%	99.4%	Cystinosis, nephropathic, 219800;Cystinosis, ocular nonnephropathic, 219750;Cystinosis, late-onset juvenile or adolescent nephropathic, 219900;Cystinosis, atypical nephropathic, 219800
CTPS1	100%	100%	100%	100%	99.7%	Immunodeficiency 24, 615897

CTSA	100%	100%	100%	100%	99.5%	Galactosialidosis, 256540;Brain small vessel disease 6 with leukoencephalopathy, 621394
CTSC	94.8%	94.8%	100%	100%	99.8%	Periodontitis 1, aggressive, 170650;Haim-Munk syndrome, 245010;Papillon-Lefevre syndrome, 245000
CTSD	86%	86%	100%	100%	99.5%	Ceroid lipofuscinosis, neuronal, 10, 610127
CTSF	100%	100%	100%	99.9%	99.4%	Ceroid lipofuscinosis, neuronal, 13 (Kufs type), 615362
CTSK	100%	99.8%	100%	100%	99.7%	Pycnodysostosis, 265800
CTU2	100%	100%	100%	100%	99.2%	Microcephaly, facial dysmorphism, renal agenesis, and ambiguous genitalia syndrome, 618142
CUBN	100%	100%	100%	100%	99.8%	[Proteinuria, chronic benign], 618884;Imerslund-Grasbeck syndrome 1, 261100
CUL7	100%	100%	100%	100%	99.4%	3-M syndrome 1, 273750
CWC27	82.6%	82.6%	100%	100%	99.8%	Retinitis pigmentosa with or without skeletal anomalies, 250410
CWF19L1	100%	100%	100%	100%	99.8%	Spinocerebellar ataxia, autosomal recessive 17, 616127
CYB561	100%	100%	100%	100%	99.2%	Orthostatic hypotension 2, 618182
CYB5A	57.7%	57.7%	100%	100%	99.9%	Methemoglobinemia and ambiguous genitalia, 250790

CYB5R3	95.7%	95%	100%	100%	99.2%	Methemoglobinemia, type I, 250800;Methemoglobinemia, type II, 250800
CYBA	69.7%	69.3%	100%	100%	99.3%	Chronic granulomatous disease 4, autosomal recessive, 233690
CYBC1	100%	100%	100%	100%	99.6%	Chronic granulomatous disease 5, autosomal recessive, 618935
CYC1	96.9%	91.9%	100%	100%	99.3%	Mitochondrial complex III deficiency, nuclear type 6, 615453
CYP11A1	100%	98%	100%	100%	99.3%	Adrenal insufficiency, congenital, with 46XY sex reversal, partial or complete, 613743
CYP11B1	100%	100%	100%	100%	99.4%	Aldosteronism, glucocorticoid-remediable, 103900;Adrenal hyperplasia, congenital, due to 11-beta-hydroxylase deficiency, 202010
CYP11B2	100%	100%	100%	100%	99.3%	Hypoaldosteronism, congenital, due to CMO I deficiency, 203400;Aldosterone to renin ratio raised;{Low renin hypertension, susceptibility to};Hypoaldosteronism, congenital, due to CMO II deficiency, 610600
CYP17A1	100%	100%	100%	100%	99.5%	17,20-lyase deficiency, isolated, 202110;17-alpha-hydroxylase/17,20-lyase deficiency, 202110
CYP19A1	100%	100%	100%	100%	99.8%	Aromatase deficiency, 613546

CYP1B1	100%	100%	100%	100%	99.5%	Glaucoma 3A, primary open angle, congenital, juvenile, or adult onset, 231300;Anterior segment dysgenesis 6, multiple subtypes, 617315
CYP24A1	100%	100%	100%	100%	99.6%	Hypercalcemia, infantile, 1, 143880
CYP26B1	100%	100%	100%	100%	99.3%	Craniosynostosis with radiohumeral fusions and other skeletal and craniofacial anomalies, 614416
CYP26C1	100%	100%	100%	100%	99.4%	Focal facial dermal dysplasia 4, 614974
CYP27A1	100%	100%	100%	100%	99.5%	Cerebrotendinous xanthomatosis, 213700
CYP27B1	100%	100%	100%	100%	99.6%	Vitamin D-dependent rickets, type I, 264700
CYP2C8	92.3%	92.3%	100%	100%	99.9%	{Drug metabolism, altered, CYP2C8-related}, 618018
CYP2R1	100%	100%	100%	100%	99.5%	Rickets due to defect in vitamin D 25-hydroxylation deficiency, 600081
CYP2U1	100%	100%	100%	99.9%	98.8%	Spastic paraplegia 56, autosomal recessive, 615030
CYP4F22	100%	100%	100%	100%	99.1%	Ichthyosis, congenital, autosomal recessive 5, 604777
CYP4V2	100%	100%	100%	100%	99.8%	Bietti crystalline corneoretinal dystrophy, 210370
CYP7B1	100%	100%	100%	100%	100%	Spastic paraplegia 5A, autosomal recessive, 270800;Bile acid synthesis defect, congenital, 3, 613812

D2HGDH	99.9%	98.4%	100%	99.9%	99.4%	D-2-hydroxyglutaric aciduria, 600721
DAG1	100%	100%	100%	100%	99.3%	Muscular dystrophy-dystroglycanopathy (congenital with brain and eye anomalies), type A, 9, 616538;Muscular dystrophy-dystroglycanopathy (limb-girdle), type C, 9, 613818
DALRD3	100%	100%	100%	100%	99.3%	Developmental and epileptic encephalopathy 86, 618910
DAND5	78.8%	78.8%	100%	99.9%	99.1%	Heterotaxy, visceral, 13, autosomal, 621079
DAP3	91.7%	90.8%	100%	100%	99.6%	Perrault syndrome 7, 621101
DARS1	100%	100%	100%	100%	99.8%	Hypomyelination with brainstem and spinal cord involvement and leg spasticity, 615281
DARS2	100%	100%	100%	100%	99.8%	Leukoencephalopathy with brain stem and spinal cord involvement and lactate elevation, 611105;Charcot-Marie-Tooth disease, axonal, type 2LL, 621485
DAW1	100%	100%	100%	100%	99.9%	Ciliary dyskinesia, primary, 52, 620570
DBH	100%	100%	100%	100%	99.5%	Orthostatic hypotension 1, due to DBH deficiency, 223360
DBR1	100%	100%	100%	100%	99.6%	Xerosis and growth failure with immune and pulmonary dysfunction syndrome, 620510;{Encephalitis, acute, infection (viral)-induced, susceptibility to, 11}, 619441

DBT	100%	100%	100%	100%	99.9%	Maple syrup urine disease, type II, 620699
DCAF17	96.5%	96.5%	100%	99.9%	99.7%	Woodhouse-Sakati syndrome, 241080
DCC	100%	100%	100%	100%	99.7%	Mirror movements 1 and/or agenesis of the corpus callosum, 157600;Esophageal carcinoma, somatic, 133239;Colorectal cancer, somatic, 114500;Gaze palsy, familial horizontal, with progressive scoliosis, 2, 617542
DCDC2	100%	100%	100%	100%	99.8%	Nephronophthisis 19, 616217;?Deafness, autosomal recessive 66, 610212;Sclerosing cholangitis, neonatal, 617394
DCHS1	100%	100%	100%	100%	99.6%	Mitral valve prolapse 2, 607829;Van Maldergem syndrome 1, 601390
DCLRE1B	100%	100%	100%	100%	99.6%	Dyskeratosis congenita, autosomal recessive 8, 620133
DCLRE1C	97.1%	97.1%	100%	100%	99.9%	Severe combined immunodeficiency, Athabascan type, 602450;Omenn syndrome, 603554
DCPS	100%	100%	100%	100%	99.6%	Al-Raqad syndrome, 616459
DDB2	100%	100%	100%	100%	99.8%	Xeroderma pigmentosum, group E, DDB-negative subtype, 278740
DDC	100%	100%	100%	100%	99.7%	Aromatic L-amino acid decarboxylase deficiency, 608643

DDHD1	100%	100%	100%	100%	99.3%	Spastic paraplegia 28, autosomal recessive, 609340
DDHD2	100%	100%	100%	100%	99.7%	Spastic paraplegia 54, autosomal recessive, 615033
DDOST	100%	100%	100%	99.9%	99.2%	Congenital disorder of glycosylation, type Ir, 614507
DDR2	100%	100%	100%	100%	99.8%	Warburg-Cinotti syndrome, 618175;Spondylometaepiphyseal dysplasia, short limb-hand type, 271665
DDRGK1	100%	100%	100%	100%	99.6%	Spondyloepimetaphyseal dysplasia, Shohat type, 602557
DDX11	100%	100%	100%	100%	99.5%	Warsaw breakage syndrome, 613398
DDX59	100%	100%	100%	100%	99.8%	Orofaciodigital syndrome V, 174300
DEAF1	93.3%	88.8%	100%	99.9%	98%	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%	99.8%	Leukodystrophy, hypomyelinating, 18, 618404
DENND5A	100%	100%	100%	100%	99.7%	Developmental and epileptic encephalopathy 49, 617281
DEPDC5	100%	100%	100%	99.9%	99.4%	Epilepsy, familial focal, with variable foci 1, 604364;Developmental and epileptic encephalopathy 111, 620504

DES	92.6%	92.6%	100%	99.9%	98.6%	Scapuloperoneal syndrome, neurogenic, Kaeser type, 181400;Cardiomyopathy, dilated, 1I, 604765;Myopathy, myofibrillar, 1, 601419
DGAT1	100%	100%	100%	100%	99.6%	Diarrhea 7, protein-losing enteropathy type, 615863
DGKE	100%	100%	100%	100%	99.7%	{Hemolytic uremic syndrome, atypical, susceptibility to, 7}, 615008;Nephrotic syndrome, type 7, 615008
DGUOK	100%	100%	100%	100%	99.9%	Portal hypertension, noncirrhotic, 1, 617068;Progressive external ophthalmoplegia with mitochondrial DNA deletions, autosomal recessive 4, 617070;Mitochondrial DNA depletion syndrome 3 (hepatocerebral type), 251880
DHCR24	97.8%	97.8%	100%	99.7%	98.6%	Desmosterolosis, 602398
DHCR7	96.2%	96.2%	100%	100%	99.6%	Smith-Lemli-Opitz syndrome, 270400
DHDDS	74.4%	74.4%	100%	100%	99.8%	Developmental delay and seizures with or without movement abnormalities, 617836;?Congenital disorder of glycosylation, type 1bb, 621567;Retinitis pigmentosa 59, 613861
DHFR	100%	100%	100%	100%	99.9%	Megaloblastic anemia due to dihydrofolate reductase deficiency, 613839

DHH	100%	100%	100%	100%	99.7%	46XY gonadal dysgenesis with minifascicular neuropathy, 607080;46XY sex reversal 7, 233420
DHODH	89%	86.1%	100%	100%	99.5%	Miller syndrome, 263750
DHPS	96.4%	93.1%	100%	99.9%	99%	Neurodevelopmental disorder with seizures and speech and walking impairment, 618480
DHRS3	100%	100%	100%	100%	99.5%	Craniosynostosis-scoliosis syndrome, 621499
DHTKD1	92.7%	92.7%	100%	100%	99.4%	?Charcot-Marie-Tooth disease, axonal, type 2Q, 615025;Alpha-aminoadipic and alpha-ketoadipic aciduria, 204750
DHX38	100%	100%	100%	100%	99.6%	Retinitis pigmentosa 84, 618220
DIAPH1	100%	100%	100%	100%	99.5%	Deafness, autosomal dominant 1, with or without thrombocytopenia, 124900;Seizures, cortical blindness, microcephaly syndrome, 616632
DIS3L2	100%	100%	100%	99.9%	99.5%	Perlman syndrome, 267000
DISP1	100%	100%	100%	100%	99.8%	Holoprosencephaly 10, 621143
DLAT	100%	100%	100%	100%	99.4%	Pyruvate dehydrogenase E2 deficiency, 245348
DLD	100%	100%	100%	100%	99.8%	Dihydrolipoamide dehydrogenase deficiency, 246900
DLG5	100%	100%	100%	100%	99.5%	Yuksel-Vogel-Bauser syndrome, 620703

DLL3	100%	100%	100%	100%	98.8%	Spondylocostal dysostosis 1, autosomal recessive, 277300
DLX5	100%	100%	100%	100%	99.2%	Split-hand/foot malformation 1, 183600;?Split-hand/foot malformation 1 with sensorineural hearing loss, 220600
DMP1	100%	100%	100%	100%	99.1%	Hypophosphatemic rickets, AR, 241520
DMRT2	100%	100%	100%	100%	99.7%	Spondylocostal dysostosis 7, autosomal recessive, 621523
DMXL2	100%	100%	100%	100%	99.8%	Developmental and epileptic encephalopathy 81, 618663;?Deafness, autosomal dominant 71, 617605;?Polyendocrine-polyneuropathy syndrome, 616113
DNA2	97.1%	97.1%	100%	100%	99.8%	Progressive external ophthalmoplegia with mitochondrial DNA deletions, autosomal dominant 6, 615156;Rothmund-Thomson syndrome, type 4, 620819;Seckel syndrome 8, 615807
DNAAF1	96.2%	95.9%	100%	100%	99.5%	Ciliary dyskinesia, primary, 13, 613193
DNAAF11	100%	100%	100%	100%	99.8%	Ciliary dyskinesia, primary, 19, 614935
DNAAF19	100%	100%	100%	100%	99.1%	Ciliary dyskinesia, primary, 17, 614679
DNAAF2	100%	100%	100%	99.9%	99.2%	Ciliary dyskinesia, primary, 10, 612518
DNAAF3	100%	100%	100%	100%	99.4%	Ciliary dyskinesia, primary, 2, 606763

DNAAF4	100%	100%	100%	100%	99.9%	{Dyslexia, susceptibility to, 1}, 127700;Ciliary dyskinesia, primary, 25, 615482
DNAAF5	100%	99.9%	100%	99.9%	99.1%	Ciliary dyskinesia, primary, 18, 614874
DNAH1	100%	100%	100%	100%	99.4%	Spermatogenic failure 18, 617576;Ciliary dyskinesia, primary, 37, 617577
DNAH11	100%	100%	100%	100%	99.8%	Ciliary dyskinesia, primary, 7, with or without situs inversus, 611884
DNAH5	100%	100%	100%	100%	99.7%	Ciliary dyskinesia, primary, 3, with or without situs inversus, 608644
DNAH9	100%	100%	100%	100%	99.7%	Ciliary dyskinesia, primary, 40, 618300
DNAI1	95.7%	95.7%	100%	100%	99.8%	Ciliary dyskinesia, primary, 1, with or without situs inversus, 244400
DNAI2	100%	100%	100%	100%	99.6%	Ciliary dyskinesia, primary, 9, with or without situs inversus, 612444
DNAJB2	100%	100%	100%	99.9%	99.5%	Neuronopathy, distal hereditary motor, autosomal recessive 5, 614881
DNAJB4	100%	100%	100%	100%	99.9%	Congenital myopathy 21 with early respiratory failure, 620326
DNAJC12	100%	100%	100%	100%	99.7%	Hyperphenylalaninemia, mild, non-BH4-deficient, 617384
DNAJC19	100%	100%	100%	100%	99.9%	3-methylglutaconic aciduria, type V, 610198

DNAJC21	94.7%	94.7%	100%	100%	99.3%	Bone marrow failure syndrome 3, 617052
DNAJC3	92.1%	92.1%	100%	100%	99.8%	Ataxia, combined cerebellar and peripheral, with hearing loss and diabetes mellitus, 616192
DNAJC30	100%	100%	100%	100%	99.7%	Leber-like hereditary optic neuropathy, autosomal recessive 1, 619382
DNAJC6	100%	100%	100%	99.9%	99.6%	Parkinson disease 19a, juvenile-onset, 615528;Parkinson disease 19b, early-onset, 615528
DNAL1	100%	100%	100%	100%	99.6%	Ciliary dyskinesia, primary, 16, 614017
DNASE1L3	100%	100%	100%	100%	99.9%	Systemic lupus erythematosus 16, 614420
DNASE2	92.7%	92.7%	100%	100%	99.3%	Autoinflammatory-panc ytopenia syndrome, 619858
DNM1	100%	100%	100%	100%	99.2%	Developmental and epileptic encephalopathy 31B, autosomal recessive, 620352;Developmental and epileptic encephalopathy 31A, autosomal dominant, 616346
DNM1L	100%	100%	100%	100%	99.9%	Optic atrophy 5, 610708;Encephalopath y, lethal, due to defective mitochondrial peroxisomal fission 1, 614388

DNM2	93.3%	91.7%	100%	99.9%	99.4%	Centronuclear myopathy 1, 160150;Charcot-Marie-Tooth disease, axonal type 2M, 606482;Charcot-Marie-Tooth disease, dominant intermediate B, 606482;Lethal congenital contracture syndrome 5, 615368
DNMBP	100%	100%	100%	100%	99.6%	Cataract 48, 618415
DNMT3B	100%	100%	100%	100%	99.7%	Immunodeficiency-centromeric instability-facial anomalies syndrome 1, 242860;Facioscapulohumeral muscular dystrophy 4, digenic, 619478
DOCK2	100%	100%	100%	100%	99.7%	Immunodeficiency 40, 616433
DOCK3	100%	100%	100%	100%	99.7%	Neurodevelopmental disorder with impaired intellectual development, hypotonia, and ataxia, 618292
DOCK6	100%	100%	100%	99.9%	99.3%	Adams-Oliver syndrome 2, 614219
DOCK7	100%	100%	100%	100%	99.8%	Developmental and epileptic encephalopathy 23, 615859
DOCK8	98.6%	98.6%	100%	100%	99.7%	Hyper-IgE syndrome 2, autosomal recessive, with recurrent infections, 243700
DOHH	98.4%	92.9%	100%	100%	99.6%	Neurodevelopmental disorder with microcephaly, cerebral atrophy, and visual impairment, 620066

DOK7	100%	100%	100%	99.9%	99.3%	Fetal akinesia deformation sequence 3, 618389;Myasthenic syndrome, congenital, 10, 254300
DOLK	100%	100%	100%	100%	99.6%	Congenital disorder of glycosylation, type Im, 610768
DONSON	100%	99.7%	100%	100%	99.7%	Microcephaly, short stature, and limb abnormalities, 617604;Microcephaly-micromelia syndrome, 251230;Meier-Gorlin syndrome 10, 621528
DPAGT1	100%	100%	100%	100%	99.4%	Myasthenic syndrome, congenital, 13, with tubular aggregates, 614750;Congenital disorder of glycosylation, type Ij, 608093
DPH1	100%	100%	100%	100%	99.3%	Developmental delay with short stature, dysmorphic facial features, and sparse hair, 616901
DPH2	100%	100%	100%	100%	99.7%	Developmental delay with short stature, dysmorphic facial features, and sparse hair 2, 620062
DPH5	75.4%	75.4%	100%	100%	99.9%	Neurodevelopmental disorder with short stature, prominent forehead, and feeding difficulties, 620070
DPM1	99.6%	96.9%	99.6%	98.4%	96.6%	Congenital disorder of glycosylation, type Ie, 608799
DPM2	100%	99.9%	100%	100%	99.5%	Congenital disorder of glycosylation, type Iu, 615042

DPM3	100%	100%	100%	100%	98.8%	?Muscular dystrophy-dystroglycanopathy (congenital with impaired intellectual development), type B, 15, 618992;Muscular dystrophy-dystroglycanopathy (limb-girdle), type C, 15, 612937
DPP9	100%	100%	100%	99.9%	99.5%	Hatipoglu immunodeficiency syndrome, 620331
DPY19L2	100%	100%	100%	99.8%	99%	Spermatogenic failure 9, 613958
DPYD	86.9%	86.9%	100%	100%	99.9%	Dihydropyrimidine dehydrogenase deficiency, 274270;5-fluorouracil toxicity, 274270
DPYS	100%	100%	100%	100%	99.5%	Dihydropyrimidinuria, 222748
DRAM2	98.4%	93.3%	100%	100%	99.7%	Cone-rod dystrophy 21, 616502
DRC1	100%	100%	100%	100%	99.5%	Spermatogenic failure 80, 620222;Ciliary dyskinesia, primary, 21, 615294
DRG1	92.1%	92.1%	100%	100%	99.7%	Tan-Almurshedi syndrome, 620641
DSC2	100%	100%	100%	99.9%	99.5%	Arrhythmogenic right ventricular dysplasia 11 with mild palmoplantar keratoderma and woolly hair, 610476;Arrhythmogenic right ventricular dysplasia 11, 610476
DSC3	100%	100%	100%	100%	99.7%	Hypotrichosis and recurrent skin vesicles, 613102
DSE	100%	100%	100%	100%	99.7%	Ehlers-Danlos syndrome, musculocontractural type 2, 615539

DSG1	100%	100%	100%	100%	99.8%	Keratosis palmoplantaris striata I, AD, 148700;Erythroderma, congenital, with palmoplantar keratoderma, hypotrichosis, and hyper IgE, 615508
DSG2	88%	88%	100%	100%	99.7%	Cardiomyopathy, dilated, 1BB, 612877;Arrhythmogenic right ventricular dysplasia 10, 610193
DSG3	100%	100%	100%	100%	99.8%	Blistering, acantholytic, of oral and laryngeal mucosa, 619226
DSG4	100%	100%	100%	100%	99.8%	Hypotrichosis 6, 607903
DSP	99.4%	99.2%	100%	100%	99.5%	Arrhythmogenic right ventricular dysplasia 8, 607450;Epidermolysis bullosa, lethal acantholytic, 609638;Keratosis palmoplantaris striata II, 612908;Dilated cardiomyopathy with woolly hair, keratoderma, and tooth agenesis, 615821;Cardiomyopathy, dilated, with woolly hair and keratoderma, 605676
DST	100%	100%	100%	100%	99.7%	Congenital myopathy 29 with contractures, 621510;Neuropathy, hereditary sensory and autonomic, type VI, 614653;Lethal congenital contracture syndrome 12, 621511;Epidermolysis bullosa simplex 3, localized or generalized intermediate, with bp230 deficiency, 615425

DSTYK	100%	100%	100%	100%	99.5%	Spastic paraplegia 23, autosomal recessive, 270750;Congenital anomalies of kidney and urinary tract 1, 610805
DTNBP1	96.4%	96.4%	100%	100%	99.7%	Hermansky-Pudlak syndrome 7, 614076
DTYMK	91.4%	84.2%	100%	100%	99.1%	Neurodegeneration, childhood-onset, with progressive microcephaly, 619847
DUOX2	100%	100%	100%	99.9%	99.3%	Thyroid dyshormonogenesis 6, 607200
DUOXA2	100%	100%	100%	100%	99.1%	Thyroid dyshormonogenesis 5, 274900
DUT	100%	100%	100%	99.8%	99%	Bone marrow failure and diabetes mellitus syndrome, 620044
DYM	100%	100%	100%	100%	99.9%	Smith-McCort dysplasia, 607326;Dyggve-Melchior-Clausen disease, 223800
DYNC1I2	92.4%	92.4%	100%	100%	99.9%	Neurodevelopmental disorder with microcephaly and structural brain anomalies, 618492
DYNC2H1	100%	100%	100%	100%	99.8%	Short-rib thoracic dysplasia 3 with or without polydactyly, 613091
DYNC2I1	95.7%	95.7%	100%	100%	99.5%	Short-rib thoracic dysplasia 8 with or without polydactyly, 615503
DYNC2I2	100%	100%	100%	100%	99.4%	Short-rib thoracic dysplasia 11 with or without polydactyly, 615633

DYNC2LI1	81.5%	81.4%	100%	100%	99.8%	Short-rib thoracic dysplasia 15 with polydactyly, 617088
DYNLT2B	58.6%	58.6%	100%	100%	99.3%	Short-rib thoracic dysplasia 17 with or without polydactyly, 617405
DYSF	96.9%	96.9%	100%	100%	99.6%	Muscular dystrophy, limb-girdle, autosomal recessive 2, 253601;Miyoshi muscular dystrophy 1, 254130;Myopathy, distal, with anterior tibial onset, 606768
DZIP1L	100%	100%	100%	100%	99.7%	Polycystic kidney disease 5, 617610
EARS2	100%	100%	100%	99.9%	99.3%	Combined oxidative phosphorylation deficiency 12, 614924
ECEL1	100%	100%	100%	100%	99.3%	Arthrogryposis, distal, type 5D, 615065
ECHS1	71.2%	69%	100%	100%	99.3%	Mitochondrial short-chain enoyl-CoA hydratase 1 deficiency, 616277
ECM1	100%	100%	100%	100%	99.3%	Urbach-Wiethe disease, 247100
EDAR	100%	100%	100%	100%	99.7%	[Hair morphology 1, hair thickness], 612630;Ectodermal dysplasia 10A, hypohidrotic/hair/nail type, autosomal dominant, 129490;Ectodermal dysplasia 10B, hypohidrotic/hair/tooth type, autosomal recessive, 224900

EDARADD	100%	100%	100%	100%	99.7%	Ectodermal dysplasia 11B, hypohidrotic/hair/tooth type, autosomal recessive, 614941;Ectodermal dysplasia 11A, hypohidrotic/hair/tooth type, autosomal dominant, 614940
EDC3	100%	100%	100%	100%	99.5%	?Intellectual developmental disorder, autosomal recessive 50, 616460
EDEM3	100%	100%	100%	100%	99.8%	Congenital disorder of glycosylation, type IIv, 619493
EDN1	100%	100%	100%	100%	99.8%	Question mark ears, isolated, 612798;Auriculocondylar syndrome 3, 615706
EDN3	100%	100%	100%	99.9%	99.4%	Waardenburg syndrome, type 4B, 613265;{Hirschsprung disease, susceptibility to, 4}, 613712
EDNRB	100%	100%	100%	100%	99.5%	{Hirschsprung disease, susceptibility to, 2}, 600155;?ABCD syndrome, 600501;Waardenburg syndrome, type 4A, 277580
EEF1D	100%	100%	100%	100%	99.5%	Neurodevelopmental disorder with thin corpus callosum, hypotonia, and absent language, 621150
EEFSEC	100%	100%	100%	100%	98.7%	Neurodevelopmental disorder with progressive spasticity and brain abnormalities, 621102

EFEMP1	84.9%	84.9%	100%	100%	99.8%	Doyne honeycomb degeneration of retina, 126600;Cutis laxa, autosomal recessive, type ID, 620780;Glaucoma 1, open angle, H, 611276
EFEMP2	99.6%	95.2%	100%	100%	99.5%	Cutis laxa, autosomal recessive, type IB, 614437
EFL1	96.3%	96.3%	100%	100%	99.9%	Shwachman-Diamond syndrome 2, 617941
EGF	100%	100%	100%	100%	99.8%	?Hypomagnesemia 4, renal, 611718
EGFR	100%	100%	100%	100%	99.7%	Neonatal nephrocutaneous inflammatory syndrome, 616069;Nonsmall cell lung cancer, response to tyrosine kinase inhibitor in, 211980;Adenocarcinoma of lung, response to tyrosine kinase inhibitor in, 211980;{Nonsmall cell lung cancer, susceptibility to}, 211980
EGR2	100%	100%	100%	100%	99.6%	Dejerine-Sottas disease, 145900;Charcot-Marie-Tooth disease, type 1D, 607678;Hypomyelinating neuropathy, congenital, 1, 605253
EIF2AK2	100%	100%	100%	100%	99.8%	Leukoencephalopathy, developmental delay, and episodic neurologic regression syndrome, 618877;Dystonia 33, 619687
EIF2AK3	100%	100%	100%	100%	99.7%	Wolcott-Rallison syndrome, 226980
EIF2AK4	99%	99%	100%	100%	99.8%	Pulmonary venoocclusive disease 2, 234810

EIF2B1	93.5%	90.6%	100%	100%	99.8%	Leukoencephalopathy with vanishing white matter 1, with or without ovarian failure, 603896
EIF2B2	100%	100%	100%	99.9%	99.2%	Leukoencephalopathy with vanishing white matter 2, with or without ovarian failure, 620312
EIF2B3	100%	100%	100%	100%	99.5%	Leukoencephalopathy with vanishing white matter 3, with or without ovarian failure, 620313
EIF2B4	100%	100%	100%	100%	99.9%	Leukoencephalopathy with vanishing white matter 4, with or without ovarian failure, 620314
EIF2B5	100%	100%	100%	100%	99.6%	Leukoencephalopathy with vanishing white matter 5, with or without ovarian failure, 620315
EIF3F	100%	100%	100%	100%	99.3%	Intellectual developmental disorder, autosomal recessive 67, 618295
EIF4A2	100%	100%	100%	100%	99.9%	Neurodevelopmental disorder with hypotonia and speech delay, with or without seizures, 620455
EIF4A3	100%	100%	100%	100%	99.4%	Robin sequence with cleft mandible and limb anomalies, 268305
EIPR1	91.2%	91.2%	100%	100%	99.7%	
ELAC2	94.8%	94.8%	100%	99.9%	99.5%	{Prostate cancer, hereditary, 2, susceptibility to}, 614731;Combined oxidative phosphorylation deficiency 17, 615440

ELFN1	100%	100%	100%	99.9%	98.5%	Dursun-Ozgul neurodevelopmental syndrome, 621344
ELMO2	97.9%	97.9%	100%	100%	99.6%	Vascular malformation, primary intraosseous, 606893
ELOVL1	100%	100%	100%	100%	99.9%	Ichthyotic keratoderma, spasticity, hypomyelination, and dysmorphic facies, 618527
ELOVL4	100%	100%	100%	100%	99.8%	Spinocerebellar ataxia 34, 133190;Stargardt disease 3, 600110;Ichthyosis, spastic quadriplegia, and impaired intellectual development, 614457
ELP1	100%	100%	100%	100%	99.8%	{Medulloblastoma}, 155255;Dysautonomia, familial, 223900
ELP2	95%	94.7%	100%	100%	99.9%	Intellectual developmental disorder, autosomal recessive 58, 617270
EMC1	99.6%	98.2%	100%	100%	99.6%	Cerebellar atrophy, visual impairment, and psychomotor retardation, 616875
EMC10	100%	100%	100%	100%	99.4%	Neurodevelopmental disorder with dysmorphic facies and variable seizures, 619264
EMG1	86.7%	86.7%	100%	99.9%	98.9%	Bowen-Conradi syndrome, 211180
EMILIN1	100%	99.7%	100%	100%	99.3%	Neuronopathy, distal hereditary motor, autosomal dominant 10, 620080;Arterial tortuosity-bone fragility syndrome, 620908
EML1	90.9%	90.9%	100%	100%	99.4%	Band heterotopia, 600348

EMP2	100%	100%	100%	100%	98.7%	Nephrotic syndrome, type 10, 615861
ENAM	100%	100%	100%	100%	99.8%	Amelogenesis imperfecta, type IC, 204650;Amelogenesis imperfecta, type IB, 104500
ENO3	100%	100%	100%	100%	98.9%	Glycogen storage disease XIII, 612932
ENPP1	100%	100%	100%	100%	99.8%	{Obesity, susceptibility to}, 601665;Hypophosphatemic rickets, autosomal recessive, 2, 613312;{Diabetes mellitus, non-insulin-dependent, susceptibility to}, 125853;Arterial calcification, generalized, of infancy, 1, 208000;Cole disease, 615522
ENTPD1	100%	100%	100%	100%	99.6%	Spastic paraplegia 64, autosomal recessive, 615683
EOGT	95.8%	92.4%	100%	100%	99.6%	Adams-Oliver syndrome 4, 615297
EPB41	100%	100%	100%	100%	99.8%	Elliptocytosis-1, 611804
EPB42	100%	100%	100%	100%	99.5%	Spherocytosis, type 5, 612690
EPCAM	100%	100%	100%	100%	99.3%	Diarrhea 5, with tufting enteropathy, congenital, 613217;Lynch syndrome 8, 613244
EPG5	100%	100%	100%	100%	99.8%	Vici syndrome, 242840;Neurodevelopmental disorder with parkinsonism or other movement abnormalities, 621506
EPHX1	91.3%	90.8%	100%	100%	99.5%	

EPM2A	100%	100%	100%	99.6%	98.1%	Myoclonic epilepsy of Lafora 1, 254780
EPO	100%	100%	100%	100%	98.9%	{Microvascular complications of diabetes 2}, 612623;Erythrocytosis, familial, 5, 617907;?Diamond-Blackfan anemia-like, 617911
EPRS1	96.2%	96.2%	100%	100%	99.4%	Leukodystrophy, hypomyelinating, 15, 617951
EPS8L2	100%	100%	100%	99.9%	99.1%	Deafness autosomal recessive 106, 617637
ERAL1	100%	100%	100%	99.9%	99.3%	Perrault syndrome 6, 617565
ERBB3	100%	100%	100%	99.9%	98.9%	?Lethal congenital contractural syndrome 2, 607598;{?Erythroleukemia, familial, susceptibility to}, 133180;Visceral neuropathy, familial, 1, autosomal recessive, 243180
ERCC1	100%	100%	100%	99.9%	99.2%	Cerebrooculofacioskeletal syndrome 4, 610758
ERCC2	96.3%	94.6%	100%	100%	99.3%	Xeroderma pigmentosum, group D, 278730;Trichothiodystrophy 1, photosensitive, 601675;?Cerebrooculofacioskeletal syndrome 2, 610756
ERCC3	100%	100%	100%	100%	99.7%	Trichothiodystrophy 2, photosensitive, 616390;Xeroderma pigmentosum, group B, 610651

ERCC4	100%	100%	100%	100%	99.6%	Xeroderma pigmentosum, type F/Cockayne syndrome, 278760;XFE progeroid syndrome, 610965;Xeroderma pigmentosum, group F, 278760;Fanconi anemia, complementation group Q, 615272
ERCC5	100%	100%	100%	100%	99.8%	Xeroderma pigmentosum, group G, 278780;Cerebrooculofacioskeletal syndrome 3, 616570;Xeroderma pigmentosum, group G/Cockayne syndrome, 278780
ERCC6	100%	100%	100%	100%	99.6%	UV-sensitive syndrome 1, 600630;Cerebrooculofacioskeletal syndrome 1, 214150;?De Sanctis-Cacchione syndrome, 278800;Cockayne syndrome, type B, 133540;{Macular degeneration, age-related, susceptibility to, 5}, 613761;Premature ovarian failure 11, 616946;{Lung cancer, susceptibility to}, 211980
ERCC6L2	100%	100%	100%	100%	99.9%	Bone marrow failure syndrome 2, 615715
ERCC8	95.2%	95.2%	100%	99.9%	98.8%	UV-sensitive syndrome 2, 614621;Cockayne syndrome, type A, 216400
ERI1	100%	100%	100%	100%	99.6%	Hoxha-Aliu syndrome, 620662;Spondyloepimetaphyseal dysplasia, Guo-Campeau type, 620663

ERLIN1	100%	100%	100%	100%	99.8%	Spastic paraplegia 62, autosomal recessive, 615681
ERLIN2	100%	100%	100%	100%	99.5%	Spastic paraplegia 18A, autosomal dominant, 620512;Spastic paraplegia 18B, autosomal recessive, 611225
ESAM	99.7%	96.8%	100%	99.9%	99.1%	Neurodevelopmental disorder with intracranial hemorrhage, seizures, and spasticity, 620371
ESCO2	100%	100%	100%	100%	99.8%	Juberg-Hayward syndrome, 216100;Roberts-SC phocomelia syndrome, 268300
ESPN	100%	100%	100%	99.8%	98.2%	Deafness, autosomal recessive 36, 609006;Deafness, autosomal dominant 91, 621556;?Usher syndrome, type 1M, 618632
ESR1	100%	100%	100%	100%	99.7%	Breast cancer, somatic, 114480;{Migraine, susceptibility to}, 157300;Estrogen resistance, 615363;{Myocardial infarction, susceptibility to}, 608446
ESRRB	100%	100%	100%	100%	99.7%	Deafness, autosomal recessive 35, 608565
ETFA	82.4%	82.4%	100%	100%	99.6%	Glutaric acidemia IIA, 231680
ETFB	92.9%	92.9%	100%	100%	99.3%	Glutaric acidemia IIB, 231680
ETFDH	94.1%	92.3%	100%	100%	99.9%	Glutaric acidemia IIC, 231680
ETHE1	77.1%	77.1%	100%	100%	99.7%	Ethylmalonic encephalopathy, 602473

EVC	100%	100%	100%	100%	99.5%	Ellis-van Creveld syndrome, 225500;?Weyers acrofacial dysostosis, 193530
EVC2	100%	100%	100%	100%	99.7%	Ellis-van Creveld syndrome, 225500;Weyers acrofacial dysostosis, 193530
EXOC2	100%	100%	100%	100%	99.9%	Neurodevelopmental disorder with dysmorphic facies and cerebellar hypoplasia, 619306
EXOC3L2	100%	100%	100%	100%	98.9%	Brain malformation renal syndrome, 620943
EXOC6B	97.1%	97.1%	100%	100%	99.7%	Spondyloepimetaphyseal dysplasia with joint laxity, type 3, 618395
EXOC7	98.6%	98.6%	100%	100%	99.6%	Neurodevelopmental disorder with seizures and brain atrophy, 619072
EXOSC2	91.1%	91%	100%	100%	99.7%	Short stature, hearing loss, retinitis pigmentosa, and distinctive facies, 617763
EXOSC3	100%	100%	100%	100%	99.7%	Pontocerebellar hypoplasia, type 1B, 614678
EXOSC5	100%	100%	100%	100%	98.6%	Cerebellar ataxia, brain abnormalities, and cardiac conduction defects, 619576
EXOSC8	100%	100%	100%	100%	99.8%	Pontocerebellar hypoplasia, type 1C, 616081
EXOSC9	100%	100%	100%	100%	99.9%	Pontocerebellar hypoplasia, type 1D, 618065

EXPH5	100%	100%	100%	100%	99.8%	Epidermolysis bullosa simplex 4, localized or generalized intermediate, autosomal recessive, 615028
EXT2	100%	100%	100%	100%	99.6%	Seizures, scoliosis, and macrocephaly syndrome, 616682;Exostoses, multiple, type 2, 133701
EXTL3	100%	99.9%	100%	100%	99.7%	Immunoskeletal dysplasia with neurodevelopmental abnormalities, 617425
EYS	100%	100%	100%	100%	99.9%	Retinitis pigmentosa 25, 602772
F10	100%	100%	100%	100%	99.4%	Factor X deficiency, 227600
F11	92.5%	92.5%	100%	100%	99.6%	Factor XI deficiency, autosomal dominant, 612416;Factor XI deficiency, autosomal recessive, 612416
F12	100%	100%	100%	100%	99.7%	Angioedema, hereditary, 3, 610618;Factor XII deficiency, 234000
F13A1	100%	100%	100%	100%	99.7%	Factor XIIIA deficiency, 613225;{Myocardial infarction, protection against}, 608446;{Venous thrombosis, protection against}, 188050
F13B	100%	100%	100%	100%	99.9%	Factor XIIIB deficiency, 613235

F2	90.8%	90.8%	100%	99.9%	99.4%	Hypoprothrombinemia, 613679;{Pregnancy loss, recurrent, susceptibility to, 2}, 614390;Dysprothrombinemia, 613679;Thrombophilia 1 due to thrombin defect, 188050;{Stroke, ischemic, susceptibility to}, 601367
F5	100%	100%	100%	100%	99.8%	Thrombophilia 2 due to activated protein C resistance, 188055;{Pregnancy loss, recurrent, susceptibility to, 1}, 614389;{Thrombophilia, susceptibility to, due to factor V Leiden}, 188055;{Budd-Chiari syndrome}, 600880;{Stroke, ischemic, susceptibility to}, 601367;Factor V deficiency, 227400
F7	99.5%	96.6%	100%	100%	99.6%	{Myocardial infarction, decreased susceptibility to}, 608446;Factor VII deficiency, 227500
FA2H	100%	100%	100%	100%	99.7%	Spastic paraplegia 35, autosomal recessive, 612319
FAAP100	100%	100%	100%	100%	99.6%	Fanconi anemia, complementation group X, 621258
FADD	98.2%	92.7%	100%	99.9%	99.2%	Immunodeficiency 90 with encephalopathy, functional hyposplenia, and hepatic dysfunction, 613759
FAH	95.2%	93.3%	100%	100%	99.5%	Tyrosinemia, type I, 276700
FAM149B1	100%	100%	100%	100%	99.9%	Joubert syndrome 36, 618763

FAM161A	100%	100%	100%	100%	99.5%	Retinitis pigmentosa 28, 606068
FAM177A1	86.5%	86.5%	100%	99.9%	99.3%	Neurodevelopmental disorder with white matter abnormalities and gait disturbance, 621152
FAM20A	100%	99.9%	100%	99.8%	99%	Amelogenesis imperfecta, type IG (enamel-renal syndrome), 204690
FAM20C	99.4%	97.2%	100%	100%	99.2%	Raine syndrome, 259775
FAM210A	100%	100%	100%	100%	99.6%	
FAN1	95.9%	94%	100%	100%	99.6%	Interstitial nephritis, karyomegalic, 614817
FANCA	100%	100%	100%	100%	99.5%	Fanconi anemia, complementation group A, 227650
FANCC	100%	100%	100%	100%	99.4%	Fanconi anemia, complementation group C, 227645
FANCD2	100%	100%	100%	100%	99.7%	Fanconi anemia, complementation group D2, 227646
FANCE	100%	100%	100%	100%	99.3%	Fanconi anemia, complementation group E, 600901
FANCF	100%	100%	100%	100%	99.2%	Fanconi anemia, complementation group F, 603467
FANCG	100%	99.5%	100%	100%	99.3%	Fanconi anemia, complementation group G, 614082
FANCI	100%	99.9%	100%	100%	99.6%	Fanconi anemia, complementation group I, 609053
FANCL	91.5%	89.3%	100%	100%	99.1%	Fanconi anemia, complementation group L, 614083

FAR1	91%	90.9%	100%	100%	99.9%	Peroxisomal fatty acyl-CoA reductase 1 disorder, 616154;Cataracts, spastic paraparesis, and speech delay, 619338
FARS2	100%	100%	100%	100%	99.5%	Combined oxidative phosphorylation deficiency 14, 614946;Spastic paraplegia 77, autosomal recessive, 617046
FARSB	94%	94%	100%	100%	99.7%	Rajab interstitial lung disease with brain calcifications 1, 613658
FASTKD2	100%	100%	100%	100%	99.8%	Combined oxidative phosphorylation deficiency 44, 618855
FASTKD5	100%	100%	100%	100%	99.9%	Mitochondrial complex IV deficiency, nuclear type 24, 621431
FAT4	100%	100%	100%	99.9%	99.5%	Van Maldergem syndrome 2, 615546;Hennekam lymphangiectasia-lymphedema syndrome 2, 616006
FBLN5	92.8%	92.8%	100%	99.9%	99.4%	Cutis laxa, autosomal recessive, type IA, 219100;Charcot-Marie-Tooth disease, demyelinating, type 1H, 619764;Macular degeneration, age-related, 3, 608895;?Cutis laxa, autosomal dominant 2, 614434
FBP1	95.5%	94.8%	100%	100%	99.7%	Fructose-1,6-bisphosphatase deficiency, 229700

FBXL3	100%	100%	100%	100%	99.8%	Intellectual developmental disorder with short stature, facial anomalies, and speech defects, 606220
FBXL4	95.6%	95.6%	100%	100%	99.8%	Mitochondrial DNA depletion syndrome 13 (encephalomyopathic type), 615471
FBXO22	100%	100%	100%	100%	99.6%	Tayoun-Maawali syndrome, 621184
FBXO31	100%	100%	100%	100%	99.2%	?Intellectual developmental disorder, autosomal recessive 45, 615979
FBXO7	99.1%	98.1%	100%	100%	99.6%	Parkinson disease 15, autosomal recessive, 260300
FCGR3A	91.7%	89%	100%	99.9%	99.6%	Immunodeficiency 20, 615707
FCHO1	98.6%	96.4%	100%	99.9%	99.2%	Immunodeficiency 76, 619164
FCN3	89.9%	89.9%	100%	99.9%	98.8%	Immunodeficiency due to ficolin 3 deficiency, 613860
FCSK	100%	100%	100%	100%	99.6%	Congenital disorder of glycosylation with defective fucosylation 2, 618324
FDFT1	100%	98.7%	100%	100%	99.5%	Squalene synthase deficiency, 618156
FDX2	100%	100%	100%	100%	99.3%	Mitochondrial myopathy, episodic, with optic atrophy and reversible leukoencephalopathy, 251900
FDXR	100%	100%	100%	100%	99.4%	Multiple mitochondrial dysfunctions syndrome 9B, 620887;Auditory neuropathy and optic atrophy, 617717

FECH	100%	100%	100%	100%	99.8%	Protoporphyria, erythropoietic, 1, 177000
FERMT1	100%	100%	100%	100%	99.7%	Kindler syndrome, 173650
FERMT3	100%	100%	100%	100%	99.4%	Leukocyte adhesion deficiency, type III, 612840
FERRY3	100%	100%	100%	100%	99.8%	Intellectual developmental disorder, autosomal recessive 66, 618221
FEZF1	100%	100%	100%	99.9%	99.6%	Hypogonadotropic hypogonadism 22, with or without anosmia, 616030
FGA	100%	99.7%	100%	100%	99.7%	Amyloidosis, hereditary systemic 2, 105200;Hypodysfibrinogenemia, congenital, 616004;Dysfibrinogenemia, congenital, 616004;Afibrinogenemia, congenital, 202400
FGB	100%	100%	100%	100%	99.6%	Hypofibrinogenemia, congenital, 202400;Dysfibrinogenemia, congenital, 616004;Afibrinogenemia, congenital, 202400
FGD4	100%	100%	100%	100%	99.8%	Charcot-Marie-Tooth disease, type 4H, 609311
FGF20	100%	100%	100%	100%	99.6%	?Renal hypodysplasia/aplasia 2, 615721
FGF23	100%	100%	100%	100%	99.2%	Tumoral calcinosis, hyperphosphatemic, familial, 2, 617993;Hypophosphatemic rickets, autosomal dominant, 193100

FGF3	100%	100%	100%	99.8%	97.8%	Deafness, congenital with inner ear agenesis, microtia, and microdontia, 610706
FGF4	100%	100%	100%	99.9%	98.4%	Short-rib thoracic dysplasia 22 without polydactyly, 621260
FGG	100%	100%	100%	100%	99.8%	Dysfibrinogenemia, congenital, 616004;Hypodysfibrinogenemia, 616004;Hypofibrinogenemia, congenital, 202400;Afibrinogenemia, congenital, 202400
FH	100%	100%	100%	100%	99.8%	Leiomyomatosis and renal cell cancer, 150800;Fumarase deficiency, 606812
FIBP	98.7%	95%	100%	100%	99.5%	Thauvin-Robinet-Faivre syndrome, 617107
FIG4	98.4%	98.4%	100%	100%	99.9%	Yunis-Varon syndrome, 216340;?Polymicrogyria, bilateral temporooccipital, 612691;Amyotrophic lateral sclerosis 11, 612577;Charcot-Marie-Tooth disease, type 4J, 611228
FILIP1	100%	100%	100%	100%	99.7%	Neuromuscular disorder, congenital, with dysmorphic facies, 620775
FITM2	100%	100%	100%	100%	99.2%	Siddiqi syndrome, 618635
FKBP10	100%	100%	100%	100%	99.3%	Osteogenesis imperfecta, type XI, 610968;Bruck syndrome 1, 259450
FKBP14	100%	100%	100%	100%	99.9%	Ehlers-Danlos syndrome, kyphoscoliotic type, 2, 614557

FKRP	100%	100%	100%	100%	99.3%	Muscular dystrophy-dystroglycanopathy (congenital with or without impaired intellectual development), type B, 5, 606612;Muscular dystrophy-dystroglycanopathy (limb-girdle), type C, 5, 607155;Muscular dystrophy-dystroglycanopathy (congenital with brain and eye anomalies), type A, 5, 613153
FKTN	95.2%	94.9%	100%	100%	99.9%	Muscular dystrophy-dystroglycanopathy (limb-girdle), type C, 4, 611588;Muscular dystrophy-dystroglycanopathy (congenital with brain and eye anomalies), type A, 4, 253800;Cardiomyopathy, dilated, 1X, 611615;Muscular dystrophy-dystroglycanopathy (congenital without impaired intellectual development), type B, 4, 613152
FLAD1	100%	100%	100%	100%	99.5%	Lipid storage myopathy due to flavin adenine dinucleotide synthetase deficiency, 255100
FLG2	100%	100%	99.9%	99.9%	99.5%	Peeling skin syndrome 6, 618084
FLII	100%	100%	100%	100%	99.5%	Cardiomyopathy, dilated, 2J, 620635

FLNB	100%	100%	100%	100%	99.6%	Larsen syndrome, 150250;Atelosteogenesis, type I, 108720;Atelosteogenesis, type III, 108721;Spondylocarpotarsal synostosis syndrome, 272460;Boomerang dysplasia, 112310
FLVCR1	95%	94.6%	100%	100%	99.7%	Retinopathy-sensory neuropathy syndrome, 609033;Neurodevelopmental disorder with microcephaly, absent speech, and hypotonia, 621060
FLVCR2	100%	100%	100%	100%	99.7%	Proliferative vasculopathy and hydranencephaly-hydrocephaly syndrome, 225790
FMN2	100%	100%	99.9%	99.7%	97.9%	Intellectual developmental disorder, autosomal recessive 47, 616193
FMO3	100%	100%	100%	100%	99.9%	Trimethylaminuria, 602079
FNIP1	100%	100%	100%	99.9%	99.7%	Immunodeficiency 93 and hypertrophic cardiomyopathy, 619705
FOCAD	100%	100%	100%	99.9%	99.7%	Liver disease, severe congenital, 619991
FOLR1	100%	100%	100%	100%	99.5%	Neurodegeneration due to cerebral folate transport deficiency, 613068
FOXE1	100%	100%	100%	99.9%	97.1%	Bamforth-Lazarus syndrome, 241850;{Thyroid cancer, nonmedullary, 4}, 616534

FOXE3	100%	100%	100%	99.8%	97.4%	Anterior segment dysgenesis 2, multiple subtypes, 610256;{Aortic aneurysm, familial thoracic 11, susceptibility to}, 617349;Cataract 34, multiple types, 612968
FOXI1	100%	100%	100%	100%	99.8%	Enlarged vestibular aqueduct, 600791
FOXI3	100%	99.9%	100%	99.5%	97.6%	Craniofacial microsomia 2, 620444
FOXN1	100%	100%	100%	100%	99.8%	T-cell lymphopenia, infantile, with or without nail dystrophy, autosomal dominant, 618806;T-cell immunodeficiency, congenital alopecia, and nail dystrophy, 601705;?T-cell immunodeficiency with thymic aplasia, 242700
FOXRED1	100%	100%	100%	100%	99.1%	Mitochondrial complex I deficiency, nuclear type 19, 618241
FRA10AC1	92.2%	92.2%	100%	99.9%	99.5%	Neurodevelopmental disorder with growth retardation, dysmorphic facies, and corpus callosum abnormalities, 620113
FRAS1	100%	100%	100%	100%	99.7%	Fraser syndrome 1, 219000
FREM1	100%	100%	100%	100%	99.6%	Manitoba oculotrichoanal syndrome, 248450;Bifid nose with or without anorectal and renal anomalies, 608980;Trigonocephaly 2, 614485

FREM2	100%	100%	100%	100%	99.7%	Fraser syndrome 2, 617666;Cryptophthalmos, unilateral or bilateral, isolated, 123570
FRRS1L	100%	100%	100%	100%	98.2%	Developmental and epileptic encephalopathy 37, 616981
FSD1L	100%	100%	100%	99.9%	99.6%	
FSHB	100%	100%	100%	100%	99.6%	Hypogonadotropic hypogonadism 24 without anosmia, 229070
FSHR	100%	100%	100%	100%	99.8%	Ovarian hyperstimulation syndrome, 608115;Ovarian dysgenesis 1, 233300
FTCD	100%	100%	100%	99.8%	98.7%	Glutamate formiminotransferase deficiency, 229100
FTO	94.5%	94.5%	100%	100%	99.9%	Growth retardation, developmental delay, facial dysmorphism, 612938;{Obesity, susceptibility to, BMIQ14}, 612460
FUCA1	100%	100%	100%	100%	99.5%	Fucosidosis, 230000
FUT8	97.2%	97.2%	100%	100%	99.8%	Congenital disorder of glycosylation with defective fucosylation 1, 618005
FXN	100%	100%	100%	100%	98.9%	Friedreich ataxia with retained reflexes, 229300;Friedreich ataxia, 229300
FYCO1	100%	100%	100%	100%	99.5%	Cataract 18, autosomal recessive, 610019
FZD6	99.9%	99.1%	100%	100%	99.8%	Nail disorder, nonsyndromic congenital, 1, 161050

G6PC1	100%	100%	100%	100%	99.7%	Glycogen storage disease Ia, 232200
G6PC3	96.8%	96.8%	100%	100%	99.8%	Dursun syndrome, 612541;Neutropenia, severe congenital 4, autosomal recessive, 612541
GAA	100%	100%	100%	100%	99.5%	Pompe disease, late-onset, 621314;Pompe disease, infantile-onset, 232300
GAD1	100%	100%	100%	100%	99.5%	Developmental and epileptic encephalopathy 89, 619124
GALC	100%	100%	100%	100%	99.6%	Krabbe disease, 245200
GALE	100%	100%	100%	100%	99.6%	Thrombocytopenia 13, syndromic, 620776;Galactose epimerase deficiency, 230350
GALK1	92.3%	92.3%	100%	100%	99.8%	Galactokinase deficiency with cataracts, 230200
GALNS	100%	100%	100%	100%	99%	Mucopolysaccharidosis IVA, 253000
GALNT2	100%	100%	100%	100%	99.4%	Congenital disorder of glycosylation, type IIt, 618885
GALNT3	100%	100%	100%	100%	99.8%	Tumoral calcinosis, hyperphosphatemic, familial, 1, 211900
GALT	100%	100%	100%	100%	99.5%	Galactosemia, 230400
GAMT	97%	93.2%	100%	99.8%	98.8%	Cerebral creatine deficiency syndrome 2, 612736
GAN	100%	100%	100%	99.9%	99.5%	Giant axonal neuropathy-1, 256850
GAS8	100%	100%	100%	99.9%	99.6%	

GATM	100%	100%	100%	100%	99.9%	Cerebral creatine deficiency syndrome 3, 612718;Fanconi renotubular syndrome 1, 134600
GBA1	100%	98.3%	100%	100%	99.5%	{Lewy body dementia, susceptibility to}, 127750;Gaucher disease, type II, 230900;Gaucher disease, type IIIC, 231005;Gaucher disease, type III, 231000;Gaucher disease, type I, 230800;Gaucher disease, perinatal lethal, 608013;{Parkinson disease, late-onset, susceptibility to}, 168600
GBA2	100%	100%	100%	99.9%	99.4%	Spastic paraplegia 46, autosomal recessive, 614409
GBE1	100%	100%	100%	100%	99.7%	Glycogen storage disease IV, 232500;Polyglucosan body disease, adult form, 263570
GCDH	79.8%	79.8%	100%	99.9%	99.5%	Glutaricaciduria, type I, 231670
GCGR	100%	100%	100%	99.9%	99.4%	Mahvash disease, 619290
GCH1	100%	100%	100%	100%	99.5%	Dystonia, DOPA-responsive, 128230;Hyperphenylalaninemia, BH4-deficient, B, 233910

GCK	100%	100%	100%	99.9%	99.2%	MODY, type II, 125851;Diabetes mellitus, permanent neonatal 1, 606176;Hyperinsulinemic hypoglycemia, familial, 3, 602485;Diabetes mellitus, noninsulin-dependent, late onset, 125853
GCLC	100%	100%	100%	100%	99.6%	{Myocardial infarction, susceptibility to}, 608446;Anemia, congenital, nonspherocytic hemolytic, 7, 230450
GCM2	100%	100%	100%	100%	99.7%	Hypoparathyroidism, familial isolated 2, 618883;Hyperparathyroidism 4, 617343
GCNT2	100%	100%	100%	100%	99.6%	[Blood group, Ii], 110800;Adult i phenotype without cataract, 110800;Cataract 13 with adult i phenotype, 116700
GCSH	100%	100%	100%	100%	99.9%	Multiple mitochondrial dysfunctions syndrome 7, 620423
GDAP1	86.2%	86.2%	98.3%	98%	97.1%	Charcot-Marie-Tooth disease, axonal, with vocal cord paresis, 607706;Charcot-Marie-Tooth disease, recessive intermediate, A, 608340;Charcot-Marie-Tooth disease, axonal, type 2K, 607831;Charcot-Marie-Tooth disease, type 4A, 214400
GDAP2	100%	100%	100%	100%	99.9%	Spinocerebellar ataxia, autosomal recessive 27, 618369

GDF1	100%	100%	100%	100%	98.9%	Congenital heart defects, multiple types, 6, 613854;Right atrial isomerism (Ivemark), 208530
GDF5	100%	100%	100%	100%	99.2%	Acromesomelic dysplasia 2A, 200700;Acromesomelic dysplasia 2B, 228900;Multiple synostoses syndrome 2, 610017;Symphalangism, proximal, 1B, 615298;Brachydactyly, type A2, 112600;?Acromesomelic dysplasia 2C, Hunter-Thompson type, 201250;Brachydactyly, type C, 113100;{Osteoarthritis-5}, 612400;Brachydactyly, type A1, C, 615072
GEMIN4	100%	100%	100%	100%	99.5%	Neurodevelopmental disorder with microcephaly, cataracts, and renal abnormalities, 617913
GEMIN5	100%	100%	100%	99.9%	99.6%	Neurodevelopmental disorder with cerebellar atrophy and motor dysfunction, 619333
GFER	100%	100%	100%	99.9%	99.4%	Myopathy, mitochondrial progressive, with congenital cataract and developmental delay, 613076
GFM1	96.6%	96.6%	100%	100%	99.7%	Combined oxidative phosphorylation deficiency 1, 609060
GFM2	100%	100%	100%	100%	99.9%	Combined oxidative phosphorylation deficiency 39, 618397

GFPT1	97.5%	97.5%	100%	100%	99.8%	Myasthenia, congenital, 12, with tubular aggregates, 610542
GFRA1	94%	94%	100%	100%	99.5%	Renal hypodysplasia/aplasia 4, 619887
GGCX	100%	100%	100%	100%	99.4%	Vitamin K-dependent clotting factors, combined deficiency of, 1, 277450;Pseudoxanthoma elasticum-like disorder with multiple coagulation factor deficiency, 610842
GGPS1	100%	100%	100%	100%	99.9%	Muscular dystrophy, congenital hearing loss, and ovarian insufficiency syndrome, 619518
GGT1	100%	100%	100%	99.9%	99.4%	?Glutathioninuria, 231950
GH1	100%	100%	100%	100%	99.5%	Kowarski syndrome, 262650;Growth hormone deficiency, isolated, type II, 173100;Growth hormone deficiency, isolated, type IB, 612781;Growth hormone deficiency, isolated, type IA, 262400
GHR	99.6%	99.6%	99.8%	99.8%	99.1%	Laron dwarfism, 262500;Increased responsiveness to growth hormone, 604271;Growth hormone insensitivity, partial, 604271;{Hypercholesterolemia, familial, modifier of}, 143890
GHRHR	100%	100%	100%	100%	99.4%	Growth hormone deficiency, isolated, type IV, 618157

GHSR	100%	100%	100%	99.9%	99.4%	Growth hormone deficiency, isolated partial, 615925
GIMAP5	100%	100%	100%	100%	99.9%	Portal hypertension, noncirrhotic, 2, 619463
GINS1	81%	81%	100%	100%	99.8%	Immunodeficiency 55, 617827
GINS3	100%	99.9%	100%	100%	99.6%	Meier-Gorlin syndrome 9, 621512
GIPC3	100%	100%	100%	100%	99%	Deafness, autosomal recessive 15, 601869
GJA1	100%	100%	100%	100%	99.7%	Erythrokeratodermia variabilis et progressiva 3, 617525;Craniometaphyseal dysplasia, autosomal recessive, 218400;Oculodentodigital dysplasia, 164200;Palmoplantar keratoderma with congenital alopecia, 104100;Syndactyly, type III, 186100;Oculodentodigital dysplasia, autosomal recessive, 257850
GJB2	100%	100%	100%	100%	99.6%	Keratoderma, palmoplantar, with deafness, 148350;Deafness, autosomal recessive 1A, 220290;Deafness, autosomal dominant 3A, 601544;Hystrix-like ichthyosis with deafness, 602540;Bart-Pumphrey syndrome, 149200;Keratitis-ichthyosis-deafness syndrome, 148210;Vohwinkel syndrome, 124500

GJB3	100%	100%	100%	100%	99.5%	Deafness, digenic, GJB2/GJB3, 220290;Erythrokeratodermia variabilis et progressiva 1, 133200;Deafness, autosomal dominant 2B, with or without peripheral neuropathy, 612644
GJB6	100%	100%	100%	99.9%	99.8%	Ectodermal dysplasia 2, Clouston type, 129500;Deafness, autosomal dominant 3B, 612643;Deafness, autosomal recessive 1B, 612645;Deafness, digenic GJB2/GJB6, 220290
GJC2	100%	99.4%	100%	99.9%	98.3%	Lymphatic malformation 3, 613480;?Spastic paraplegia 44, autosomal recessive, 613206;Leukodystrophy, hypomyelinating, 2, 608804
GLB1	100%	100%	100%	100%	99.5%	GM1-gangliosidosis, type I, 230500;GM1-gangliosidosis, type III, 230650;Mucopolysaccharidosis type IVB (Morquio), 253010;GM1-gangliosidosis, type II, 230600
GLDC	97.3%	97.3%	100%	100%	99.6%	Glycine encephalopathy1, 605899
GLDN	100%	100%	100%	100%	99.6%	Lethal congenital contracture syndrome 11, 617194
GLE1	100%	100%	100%	100%	99.7%	Lethal congenital contracture syndrome 1, 253310;Congenital arthrogryposis with anterior horn cell disease, 611890

GLI1	100%	100%	100%	99.9%	99.1%	Polydactyly, preaxial I, 174400;Polydactyly, postaxial, type A8, 618123
GLIS2	100%	100%	100%	99.8%	99.1%	Nephronophthisis 7, 611498
GLIS3	100%	100%	100%	100%	99.6%	Diabetes mellitus, neonatal, with congenital hypothyroidism, 610199
GLRA1	100%	100%	100%	100%	99.7%	Hyperekplexia 1, 149400
GLRB	93.7%	93.7%	100%	100%	99.7%	Hyperekplexia 2, 614619
GLRX5	100%	100%	100%	100%	99.7%	Anemia, sideroblastic, 3, pyridoxine-refractory, 616860;Spasticity, childhood-onset, with hyperglycinemia, 616859
GLS	95.8%	95.8%	100%	100%	99.8%	CASGID syndrome, 618339;Global developmental delay, progressive ataxia, and elevated glutamine, 618412;Developmental and epileptic encephalopathy 71, 618328
GLUL	100%	100%	100%	99.9%	99.2%	Glutamine deficiency, congenital, 610015;Developmental and epileptic encephalopathy 116, 620806
GLYCTK	100%	100%	100%	99.9%	99.3%	D-glyceric aciduria, 220120
GM2A	100%	100%	100%	100%	99.7%	GM2-gangliosidosis, AB variant, 272750
GMPPA	100%	100%	100%	100%	98.8%	Alacrima, achalasia, and impaired intellectual development syndrome, 615510

GMPPB	100%	100%	100%	100%	99.7%	Muscular dystrophy-dystroglycanopathy (limb-girdle), type C, 14, 615352;Muscular dystrophy-dystroglycanopathy (congenital with impaired intellectual development), type B, 14, 615351;Muscular dystrophy-dystroglycanopathy (congenital with brain and eye anomalies), type A, 14, 615350
GNAT2	100%	100%	100%	100%	99.8%	Achromatopsia 4, 613856
GNB3	100%	100%	100%	100%	99.1%	Night blindness, congenital stationary, type 1H, 617024;{Hypertension, essential, susceptibility to}, 145500
GNB5	100%	100%	100%	100%	99.8%	Lodder-Merla syndrome, type 2, with developmental delay and with or without cardiac arrhythmia, 617182;Lodder-Merla syndrome, type 1, with impaired intellectual development and cardiac arrhythmia, 617173
GNE	100%	100%	100%	100%	99.8%	Sialuria, 269921;Thrombocytopenia 12 with or without myopathy, 620757;Nonaka myopathy, 605820
GNMT	100%	100%	100%	99.9%	98.2%	Glycine N-methyltransferase deficiency, 606664
GNPAT	100%	100%	100%	100%	100%	Rhizomelic chondrodysplasia punctata, type 2, 222765

GNPTAB	100%	100%	100%	100%	99.8%	Mucolipidosis III alpha/beta, 252600;Mucolipidosis II alpha/beta, 252500
GNPTG	90.1%	90.1%	100%	99.8%	98.5%	Mucolipidosis III gamma, 252605
GNRHR	100%	100%	100%	100%	99.8%	Hypogonadotropic hypogonadism 7 without anosmia, 146110
GNS	94.3%	94.3%	100%	100%	99.6%	Mucopolysaccharidosis type IIID, 252940
GOLGA2	100%	100%	100%	99.9%	99.1%	Developmental delay with hypotonia, myopathy, and brain abnormalities, 620240
GON4L	100%	100%	100%	100%	99.7%	Li-Takada-Miyake syndrome, 621212
GON7	100%	100%	100%	100%	100%	Galloway-Mowat syndrome 9, 619603
GORAB	100%	100%	100%	100%	99.5%	Geroderma osteodysplasticum, 231070
GOSR2	100%	100%	100%	100%	99.5%	Epilepsy, progressive myoclonic 6, 614018;Muscular dystrophy, congenital, with or without seizures, 620166
GOT2	92.8%	92.8%	100%	100%	99.5%	Developmental and epileptic encephalopathy 82, 618721

GP1BA	100%	100%	99.9%	99.7%	98.6%	Bernard-Soulier syndrome, type A1 (recessive), 231200;Bernard-Soulier syndrome, type A2 (dominant), 153670;von Willebrand disease, platelet-type, 177820;{Nonarteritic anterior ischemic optic neuropathy, susceptibility to}, 258660
GP1BB	100%	100%	100%	99.4%	97.5%	Giant platelet disorder, isolated, 231200;Bernard-Soulier syndrome, type B, 231200
GP6	100%	98.2%	100%	100%	99.6%	Bleeding disorder, platelet-type, 11, 614201
GP9	100%	100%	100%	100%	99%	Bernard-Soulier syndrome, type C, 231200
GPAA1	88.5%	88.5%	100%	100%	99.4%	Glycosylphosphatidylinositol biosynthesis defect 15, 617810
GPC6	100%	100%	100%	100%	99.7%	Omodysplasia 1, 258315
GPD1	100%	100%	100%	100%	99.7%	Hypertriglyceridemia, transient infantile, 614480
GPHN	95.9%	95.9%	100%	100%	99.8%	Molybdenum cofactor deficiency C, 615501
GPI	100%	100%	100%	100%	99.4%	Anemia, congenital, nonspherocytic hemolytic, 4, glucose phosphate isomerase deficient, 613470
GPIHBP1	94.6%	90.9%	100%	100%	99.9%	Hyperlipoproteinemia, type 1D, 615947
GPNMB	95.1%	95.1%	100%	100%	99.8%	Amyloidosis, primary localized cutaneous, 3, 617920

GPR156	100%	100%	100%	100%	99.6%	Deafness, autosomal recessive 121, 620551
GPR179	100%	100%	100%	100%	99.5%	Night blindness, congenital stationary (complete), 1E, autosomal recessive, 614565
GPSM2	95.5%	95.5%	100%	100%	99.8%	Chudley-McCullough syndrome, 604213
GPT2	92.9%	92.9%	100%	100%	99.7%	Neurodevelopmental disorder with microcephaly and spastic paraplegia, 616281
GPX4	100%	100%	100%	99.9%	98.7%	Spondylometaphyseal dysplasia, Sedaghatian type, 250220
GRAP	100%	100%	100%	99.7%	99.5%	Deafness, autosomal recessive 114, 618456
GRHL2	100%	100%	100%	100%	99.8%	Deafness, autosomal dominant 28, 608641;Ectodermal dysplasia/short stature syndrome, 616029;Corneal dystrophy, posterior polymorphous, 4, 618031
GRHPR	86.7%	86.6%	100%	100%	99.7%	Hyperoxaluria, primary, type II, 260000
GRID2	100%	100%	100%	100%	99.8%	Spinocerebellar ataxia, autosomal recessive 18, 616204
GRIK2	95.3%	95.3%	100%	99.9%	99.3%	Neurodevelopmental disorder with impaired language and ataxia and with or without seizures, 619580;Intellectual developmental disorder, autosomal recessive 6, 611092

GRIN1	100%	100%	100%	100%	99.2%	Neurodevelopmental disorder with or without hyperkinetic movements and seizures, autosomal recessive, 617820;Developmental and epileptic encephalopathy 101, 619814;Neurodevelopmental disorder with or without hyperkinetic movements and seizures, autosomal dominant, 614254
GRIP1	100%	100%	100%	100%	99.9%	Fraser syndrome 3, 617667
GRK1	100%	100%	100%	100%	98.7%	Oguchi disease-2, 613411
GRM1	100%	100%	100%	100%	99.6%	Spinocerebellar ataxia, autosomal recessive 13, 614831;Spinocerebellar ataxia 44, 617691
GRM6	100%	100%	100%	99.9%	99.2%	Night blindness, congenital stationary (complete), 1B, autosomal recessive, 257270
GRM7	100%	100%	100%	99.8%	98.4%	Neurodevelopmental disorder with seizures, hypotonia, and brain abnormalities, 618922
GRN	100%	100%	100%	100%	99.7%	Frontotemporal dementia 2, 607485;Aphasia, primary progressive, 607485;Ceroid lipofuscinosis, neuronal, 11, 614706
GRXCR1	100%	100%	100%	100%	99.5%	Deafness, autosomal recessive 25, 613285
GSC	100%	100%	100%	99.9%	98.6%	Short stature, auditory canal atresia, mandibular hypoplasia, skeletal abnormalities, 602471

GSS	100%	100%	100%	99.9%	99.5%	Anemia, congenital, nonspherocytic hemolytic, 6, glutatione synthetase deficient, 231900;Glutathione synthetase deficiency, 266130
GSX2	100%	100%	100%	100%	99.4%	Diencephalic-mesence phalic junction dysplasia syndrome 2, 618646
GTF2E2	78.7%	78.7%	100%	100%	99.9%	Trichothiodystrophy 6, nonphotosensitive, 616943
GTF2H5	44.2%	44.2%	100%	100%	99.8%	Trichothiodystrophy 3, photosensitive, 616395
GTF3C3	98.6%	98.5%	100%	100%	99.7%	Neurodevelopmental disorder with dysmorphic facies, brain anomalies, and seizures, 621201
GTPBP1	99.9%	98.3%	100%	99.9%	98.8%	Neurodevelopmental disorder with characteristic facial and ectodermal features and tetraparesis 1, 620888
GTPBP2	100%	100%	100%	100%	99.9%	Jaberi-Elahi syndrome, 617988
GTPBP3	100%	100%	100%	99.9%	99.3%	Combined oxidative phosphorylation deficiency 23, 616198
GUCY1A1	100%	100%	100%	100%	99.8%	Moyamoya 6 with achalasia, 615750
GUCY2C	98.7%	98.7%	100%	100%	99.8%	Diarrhea 6, 614616;Meconium ileus, 614665

GUCY2D	100%	100%	100%	100%	99.5%	Cone-rod dystrophy 6, 601777;?Choroidal dystrophy, central areolar 1, 215500;Leber congenital amaurosis 1, 204000;Night blindness, congenital stationary, type 1I, 618555
GUK1	100%	100%	100%	100%	99.6%	Mitochondrial DNA depletion syndrome 21, 621071
GUSB	98.4%	98.4%	100%	100%	99.1%	Mucopolysaccharidosis VII, 253220
GYG1	93.7%	93.7%	100%	100%	100%	?Glycogen storage disease XV, 613507;Polyglucosan body myopathy 2, 616199
GYS1	100%	100%	100%	100%	99.5%	Glycogen storage disease 0, muscle, 611556
GYS2	100%	100%	100%	100%	99.8%	Glycogen storage disease 0, liver, 240600
GZF1	99.3%	97.6%	100%	100%	99.7%	Joint laxity, short stature, and myopia, 617662
H6PD	100%	100%	100%	100%	99.5%	Cortisone reductase deficiency 1, 604931
HAAO	100%	98.9%	100%	100%	99.5%	Vertebral, cardiac, renal, and limb defects syndrome 1, 617660
HACD1	71.3%	71.3%	100%	100%	99.6%	Congenital myopathy 11, 619967
HACE1	95.9%	95.9%	100%	100%	99.7%	Spastic paraplegia and psychomotor retardation with or without seizures, 616756

HADH	87.7%	87.7%	100%	100%	99.6%	Hyperinsulinemic hypoglycemia, familial, 4, 609975;3-hydroxyacyl-CoA dehydrogenase deficiency, 231530
HADHA	96.1%	95.3%	100%	100%	99.6%	HELLP syndrome, maternal, of pregnancy, 609016;LCHAD deficiency, 609016;Mitochondrial trifunctional protein deficiency 1, 609015;Fatty liver, acute, of pregnancy, 609016
HADHB	94.6%	94.6%	100%	100%	99.7%	Mitochondrial trifunctional protein deficiency 2, 620300
HAMP	100%	100%	100%	100%	99.9%	Hemochromatosis, type 2B, 613313
HARS1	93.2%	93.2%	100%	99.9%	99.9%	Charcot-Marie-Tooth disease, axonal, type 2W, 616625;Usher syndrome type 3B, 614504
HARS2	100%	100%	100%	99.9%	98.9%	Perrault syndrome 2, 614926
HAVCR2	100%	100%	100%	100%	99.3%	T-cell lymphoma, subcutaneous panniculitis-like, 618398
HAX1	100%	100%	100%	100%	99.7%	Neutropenia, severe congenital 3, autosomal recessive, 610738

HBB	100%	100%	100%	100%	99.9%	Methemoglobinemia, beta type, 617971;Thalassemia-beta, dominant inclusion-body, 603902;Sickle cell disease, 603903;Thalassemia, beta, 613985;Delta-beta thalassemia, 141749;{Malaria, resistance to}, 611162;Hereditary persistence of fetal hemoglobin, 141749;Erythrocytosis, familial, 6, 617980;Heinz body anemia, 140700
HEATR3	100%	100%	100%	100%	99.8%	Diamond-Blackfan anemia 21, 620072
HECTD4	100%	100%	100%	100%	99.6%	Neurodevelopmental disorder with seizures, spasticity, and complete or partial agenesis of the corpus callosum, 620250
HELLS	97.7%	97.4%	100%	100%	99.8%	Immunodeficiency-centromeric instability-facial anomalies syndrome 4, 616911
HEPACAM	100%	100%	100%	100%	99.5%	Megalencephalic leukoencephalopathy with subcortical cysts 2A, 613925;Megalencephalic leukoencephalopathy with subcortical cysts 2B, remitting, with or without impaired intellectual development, 613926
HERC1	100%	100%	100%	100%	99.7%	Macrocephaly, dysmorphic facies, and psychomotor retardation, 617011

HERC2	100%	100%	100%	99.9%	99.5%	Intellectual developmental disorder, autosomal recessive 38, 615516;[Skin/hair/eye pigmentation 1, blond/brown hair], 227220;[Skin/hair/eye pigmentation 1, blue/nonblue eyes], 227220
HES7	100%	100%	100%	99.8%	98.2%	Spondylocostal dysostosis 4, autosomal recessive, 613686
HESX1	100%	100%	100%	100%	99.7%	Pituitary hormone deficiency, combined, 5, 182230;Septooptic dysplasia, 182230;Growth hormone deficiency with pituitary anomalies, 182230
HEXA	100%	100%	100%	100%	99.4%	[Hex A pseudodeficiency], 272800;GM2-gangliosidosis, several forms, 272800;Tay-Sachs disease, 272800
HEXB	100%	100%	100%	99.9%	99.5%	Sandhoff disease, infantile, juvenile, and adult forms, 268800
HGD	90.1%	90%	100%	100%	99.9%	Alkaptonuria, 203500
HGF	100%	100%	100%	100%	99.4%	Deafness, autosomal recessive 39, 608265
HGSNAT	83.5%	83.5%	100%	100%	99.8%	Mucopolysaccharidosis type IIIC (Sanfilippo C), 252930;Retinitis pigmentosa 73, 616544
HHAT	100%	100%	100%	100%	99.6%	Nivelon-Nivelon-Mabille syndrome, 600092
HIBCH	100%	100%	100%	100%	100%	3-hydroxyisobutryl-CoA hydrolase deficiency, 250620

HID1	100%	100%	100%	100%	99.5%	Developmental and epileptic encephalopathy 105 with hypopituitarism, 619983
HIKESHI	100%	100%	100%	100%	99.4%	Leukodystrophy, hypomyelinating, 13, 616881
HINT1	100%	100%	100%	100%	99.8%	Neuromyotonia and axonal neuropathy, autosomal recessive, 137200
HJV	100%	100%	100%	100%	99.8%	Hemochromatosis, type 2A, 602390
HK1	98.2%	96.5%	100%	99.9%	99.5%	Anemia, congenital, nonspherocytic hemolytic, 5, hexokinase deficient, 235700;Retinitis pigmentosa 79, 617460;Neuropathy, hereditary motor and sensory, Russe type, 605285;Neurodevelop mental disorder with visual defects and brain anomalies, 618547
HLCS	100%	100%	100%	100%	99.2%	Holocarboxylase synthetase deficiency, 253270
HLX	100%	100%	100%	100%	99.3%	
HMBS	100%	100%	100%	100%	99.9%	Leukoencephalopathy, porphyria-related, 620711;Encephalopath y, porphyria-related, 620704;Porphyria, acute intermittent, nonerythroid variant, 176000;Porphyria, acute intermittent, 176000
HMGCL	76.2%	76.2%	100%	100%	99.5%	HMG-CoA lyase deficiency, 246450

HMGCR	100%	100%	100%	100%	99.7%	Muscular dystrophy, limb-girdle, autosomal recessive 28, 620375;[Statins, response to], 620410;[Low density lipoprotein cholesterol level QTL 3], 620410
HMGCS1	97.2%	97.2%	100%	100%	99.7%	Congenital myopathy 28 with rigid spine, 621433
HMGCS2	97.5%	97.5%	100%	100%	99.2%	HMG-CoA synthase-2 deficiency, 605911
HMOX1	100%	100%	100%	100%	99.5%	Heme oxygenase-1 deficiency, 614034;{Pulmonary disease, chronic obstructive, susceptibility to}, 606963
HMX1	100%	100%	99.9%	99.5%	97.7%	Oculoauricular syndrome, 612109
HNMT	100%	100%	100%	100%	99.1%	Intellectual developmental disorder, autosomal recessive 51, 616739;{Asthma, susceptibility to}, 600807
HOGA1	100%	100%	100%	100%	99.5%	Hyperoxaluria, primary, type III, 613616
HOXA1	100%	100%	100%	100%	99.5%	Bosley-Salih-Alorainy syndrome, 601536;Athabaskan brainstem dysgenesis syndrome, 601536
HOXA2	100%	100%	100%	99.6%	98%	Microtia with or without hearing impairment (AD), 612290;?Microtia, hearing impairment, and cleft palate (AR), 612290
HOXB1	100%	100%	100%	100%	99.9%	Facial paresis, hereditary congenital, 3, 614744

HOXC13	100%	100%	100%	100%	99.4%	Ectodermal dysplasia 9, hair/nail type, 614931
HPCA	100%	100%	100%	99.9%	98.8%	Dystonia 2, torsion, autosomal recessive, 224500
HPD	93.7%	90.9%	100%	99.9%	98.9%	Hawkinsinuria, 140350;Tyrosinemia, type III, 276710
HPDL	100%	100%	100%	100%	99.8%	Neurodevelopmental disorder with progressive spasticity and brain white matter abnormalities, 619026;Spastic paraplegia 83, autosomal recessive, 619027
HPGD	100%	100%	100%	100%	99.9%	?Digital clubbing, isolated congenital, 119900;Hypertrophic osteoarthropathy, primary, autosomal recessive 1, 259100;Cranioosteoarthropathy, 259100
HPS1	100%	100%	100%	100%	99.7%	Hermansky-Pudlak syndrome 1, 203300
HPS3	100%	100%	100%	100%	99.8%	Hermansky-Pudlak syndrome 3, 614072
HPS4	100%	100%	100%	100%	99.8%	Hermansky-Pudlak syndrome 4, 614073
HPS5	100%	100%	100%	100%	99.8%	Hermansky-Pudlak syndrome 5, 614074
HPS6	100%	100%	100%	100%	99.4%	Hermansky-Pudlak syndrome 6, 614075
HPSE2	100%	100%	100%	100%	99.5%	Urofacial syndrome 1, 236730
HR	100%	100%	100%	100%	99.5%	Atrichia with papular lesions, 209500;Alopecia universalis, 203655

HS2ST1	100%	100%	100%	100%	99.7%	Neurofacioskeletal syndrome with or without renal agenesis, 619194
HSD11B2	100%	100%	100%	100%	99.4%	Apparent mineralocorticoid excess, 218030
HSD17B3	100%	100%	100%	100%	99.8%	Pseudohermaphroditism, male, with gynecomastia, 264300
HSD17B4	100%	100%	100%	100%	99.9%	D-bifunctional protein deficiency, 261515;Perrault syndrome 1, 233400
HSD3B2	100%	100%	100%	100%	99.6%	Adrenal hyperplasia, congenital, due to 3-beta-hydroxysteroid dehydrogenase 2 deficiency, 201810
HSD3B7	100%	100%	100%	100%	99.5%	Bile acid synthesis defect, congenital, 1, 607765
HSPA9	100%	100%	100%	100%	99.6%	Even-plus syndrome, 616854;Anemia, sideroblastic, 4, 182170
HSPD1	99.9%	98.9%	100%	100%	99.9%	Spastic paraplegia 13, autosomal dominant, 605280;Leukodystrophy, hypomyelinating, 4, 612233
HSPG2	100%	100%	100%	100%	99.4%	Dyssegmental dysplasia, Silverman-Handmaker type, 224410;Schwartz-Jampel syndrome, type 1, 255800

HTRA1	100%	100%	100%	99.9%	99.3%	Cerebral arteriopathy, autosomal dominant, with subcortical infarcts and leukoencephalopathy 2, 616779;Cerebral arteriopathy, autosomal recessive, with subcortical infarcts and leukoencephalopathy 2, 600142
HTRA2	100%	100%	100%	100%	99.5%	{Parkinson disease 13}, 610297;3-methylglutaconic aciduria, type VIII, 617248
HYAL1	100%	100%	100%	99.9%	98.9%	Mucopolysaccharidosis type IX, 601492
HYAL2	100%	100%	100%	100%	99.5%	Muggenthaler-Chowdhury-Chioza syndrome, 621063
HYCC1	100%	100%	100%	100%	100%	Leukodystrophy, hypomyelinating, 5, 610532
HYDIN	100%	99.9%	100%	99.9%	99.5%	Ciliary dyskinesia, primary, 5, 608647
HYLS1	100%	100%	100%	100%	99.8%	Hydrolethalus syndrome, 236680
IARS1	100%	100%	100%	100%	99.6%	Growth retardation, impaired intellectual development, hypotonia, and hepatopathy, 617093
IARS2	100%	99.7%	100%	99.9%	99.6%	Cataracts, growth hormone deficiency, sensory neuropathy, sensorineural hearing loss, and skeletal dysplasia, 616007
IBA57	100%	100%	100%	100%	99.5%	Multiple mitochondrial dysfunctions syndrome 3, 615330;?Spastic paraplegia 74, autosomal recessive, 616451

ICOS	100%	100%	100%	99.9%	99.7%	Immunodeficiency, common variable, 1, 607594
IDH3A	100%	100%	100%	100%	99.9%	Retinitis pigmentosa 90, 619007
IDH3B	100%	100%	100%	100%	99%	Retinitis pigmentosa 46, 612572
IDUA	96%	96%	100%	99.8%	98.7%	Mucopolysaccharidosis Is, 607016;Mucopolysaccharidosis Ih/s, 607015;Mucopolysaccharidosis Ih, 607014
IER3IP1	90.2%	74.9%	100%	100%	99.3%	Microcephaly, epilepsy, and diabetes syndrome, 614231
IFIH1	100%	100%	100%	100%	99.8%	Immunodeficiency 95, 619773;Aicardi-Goutieres syndrome 7, 615846;Singleton-Merten syndrome 1, 182250
IFNAR1	94.5%	94.5%	100%	100%	99.6%	Immunodeficiency 106, susceptibility to viral infections, 619935
IFNAR2	100%	100%	100%	100%	99.9%	{Hepatitis B virus, susceptibility to}, 610424;Immunodeficiency 45, 616669
IFNGR1	100%	100%	100%	100%	99.8%	{H. pylori infection, susceptibility to}, 600263;Immunodeficiency 27A, mycobacteriosis, AR, 209950;Immunodeficiency 27B, mycobacteriosis, AD, 615978;{Tuberculosis infection, protection against}, 607948;{Tuberculosis, susceptibility to}, 607948;{Hepatitis B virus infection, susceptibility to}, 610424

IFNGR2	91.6%	91.6%	100%	99.9%	99.4%	Immunodeficiency 28, mycobacteriosis, 614889
IFT122	100%	100%	100%	100%	99.6%	Cranioectodermal dysplasia 1, 218330
IFT140	100%	100%	100%	100%	99.6%	{Polycystic kidney disease 9, susceptibility to}, 621164;Short-rib thoracic dysplasia 9 with or without polydactyly, 266920;Retinitis pigmentosa 80, 617781;Cranioectodermal dysplasia 5, 621180
IFT172	100%	100%	100%	100%	99.7%	Retinitis pigmentosa 71, 616394;Bardet-Biedl syndrome 20, 619471;Short-rib thoracic dysplasia 10 with or without polydactyly, 615630
IFT27	99.8%	96.5%	100%	100%	99.9%	Bardet-Biedl syndrome 19, 615996
IFT43	93.5%	93.5%	100%	100%	99.7%	?Cranioectodermal dysplasia 3, 614099;?Retinitis pigmentosa 81, 617871;Short-rib thoracic dysplasia 18 with polydactyly, 617866
IFT52	92.9%	92.9%	100%	100%	99.8%	Short-rib thoracic dysplasia 16 with or without polydactyly, 617102
IFT56	100%	100%	100%	100%	99.8%	Biliary, renal, neurologic, and skeletal syndrome, 619534
IFT57	92.4%	92.4%	100%	100%	99.9%	?Orofaciodigital syndrome XVIII, 617927

IFT74	100%	100%	100%	100%	100%	Bardet-Biedl syndrome 22, 617119;Spermatogenic failure 58, 619585;Joubert syndrome 40, 619582
IFT80	100%	100%	100%	100%	99.6%	Short-rib thoracic dysplasia 2 with or without polydactyly, 611263
IFT81	92.5%	92.5%	100%	100%	99.7%	Short-rib thoracic dysplasia 19 with or without polydactyly, 617895
IGF1	100%	100%	100%	99.9%	99.1%	Insulin-like growth factor I deficiency, 608747
IGF1R	100%	100%	100%	100%	99.7%	Insulin-like growth factor I, resistance to, 270450
IGFALS	95.7%	95.7%	100%	100%	99.2%	Acid-labile subunit, deficiency of, 615961
IGFBP7	85.5%	85.5%	100%	99.9%	98.8%	Retinal arterial macroaneurysm with supravalvular pulmonic stenosis, 614224
IGHM	100%	100%	100%	100%	99.8%	
IGHMBP2	100%	100%	100%	100%	99.4%	Charcot-Marie-Tooth disease, axonal, type 2S, 616155;Neuronopathy, distal hereditary motor, autosomal recessive 1, 604320
IGKC	100%	100%	100%	100%	99.8%	
IGLL1	100%	100%	100%	100%	99.2%	Agammaglobulinemia 2, 613500
IHH	100%	100%	100%	99.8%	99%	Acrocapitofemoral dysplasia, 607778;Brachydactyly, type A1, 112500

IKBKB	100%	100%	100%	100%	99.4%	Immunodeficiency 15B, 615592;Immunodeficiency 15A, 618204
IL10RA	100%	100%	100%	100%	99.6%	Inflammatory bowel disease 28, early onset, autosomal recessive, 613148
IL10RB	100%	100%	100%	100%	99.7%	{Hepatitis B virus, susceptibility to}, 610424;Inflammatory bowel disease 25, early onset, autosomal recessive, 612567
IL11RA	92.1%	92.1%	100%	100%	99.3%	Craniosynostosis and dental anomalies, 614188
IL12B	100%	100%	100%	100%	100%	Immunodeficiency 29, mycobacteriosis, 614890
IL12RB1	94.1%	94.1%	100%	99.9%	99.2%	Immunodeficiency 30, 614891
IL17RA	100%	100%	100%	100%	99.5%	Immunodeficiency 51, 613953
IL17RC	100%	100%	100%	99.9%	99.3%	Candidiasis, familial, 9, 616445
IL1RN	100%	100%	100%	100%	99.6%	Chronic recurrent multifocal osteomyelitis 2, with periostitis and pustulosis, 612852;{Gastric cancer risk after H. pylori infection}, 613659;{Microvascular complications of diabetes 4}, 612628;Interleukin 1 receptor antagonist deficiency, 612852
IL21R	100%	100%	100%	100%	99.2%	Immunodeficiency 56, 615207
IL27RA	100%	100%	100%	99.9%	99.2%	Immunodeficiency 134 (Epstein-Barr virus-specific), 621405

IL2RA	100%	100%	100%	100%	99.7%	Immunodeficiency 41 with lymphoproliferation and autoimmunity, 606367;{Diabetes, mellitus, insulin-dependent, susceptibility to, 10}, 601942
IL2RB	96.1%	96.1%	100%	100%	99.7%	Immunodeficiency 63 with lymphoproliferation and autoimmunity, 618495
IL36RN	100%	100%	100%	100%	99.9%	Psoriasis 14, pustular, 614204
IL6R	86.3%	86.3%	100%	99.9%	99.4%	[Interleukin 6, serum level of, QTL], 614752;Hyper-IgE syndrome 5, autosomal recessive, with recurrent infections, 618944;[Interleukin-6 receptor, soluble, serum level of, QTL], 614689
IL6ST	96.7%	96.7%	100%	100%	99.6%	Hyper-IgE syndrome 4A, autosomal dominant, with recurrent infections, 619752;Stuve-Wiedemann syndrome 2, 619751;Hyper-IgE syndrome 4B, autosomal recessive, with recurrent infections, 618523;?Immunodeficiency 94 with autoinflammation and dysmorphic facies, 619750
IL7R	100%	100%	100%	100%	99.7%	Immunodeficiency 104, severe combined, 608971
ILDR1	100%	100%	100%	100%	99.8%	Deafness, autosomal recessive 42, 609646

IMPA1	81.6%	81.6%	100%	100%	99.9%	Intellectual developmental disorder, autosomal recessive 59, 617323
IMPG2	100%	100%	100%	100%	99.8%	Retinitis pigmentosa 56, 613581;Macular dystrophy, vitelliform, 5, 616152
INPP4A	100%	100%	100%	100%	99.7%	Neurodevelopmental disorder with growth impairment, quadriparesis, and poor or absent speech, 621354
INPP5E	100%	100%	100%	99.8%	98.1%	Impaired intellectual development, truncal obesity, retinal dystrophy, and micropenis syndrome, 610156;Joubert syndrome 1, 213300
INPP5K	100%	98.5%	100%	100%	99.8%	Muscular dystrophy, congenital, with cataracts and intellectual disability, 617404
INPPL1	100%	100%	100%	99.9%	99.4%	Opsismodysplasia, 258480
INSR	100%	100%	100%	100%	99.5%	Rabson-Mendenhall syndrome, 262190;Diabetes mellitus, insulin-resistant, with acanthosis nigricans, 610549;Donohue syndrome, 246200;Hyperinsulinemic hypoglycemia, familial, 5, 609968
INTS1	99.6%	98.7%	100%	100%	99.4%	Neurodevelopmental disorder with cataracts, poor growth, and dysmorphic facies, 618571

INTS11	100%	100%	100%	99.9%	98.6%	Neurodevelopmental disorder with motor and language delay, ocular defects, and brain abnormalities, 620428
INTS8	100%	100%	100%	100%	99.4%	?Neurodevelopmental disorder with cerebellar hypoplasia and spasticity, 618572
INTU	100%	100%	100%	100%	99.7%	?Orofaciodigital syndrome XVII, 617926;?Short-rib thoracic dysplasia 20 with polydactyly, 617925
INVS	100%	100%	100%	100%	99.9%	Nephronophthisis 2, infantile, 602088
IPO8	97%	97%	100%	100%	99.8%	VISS syndrome, 619472
IQCB1	95.8%	95.8%	100%	100%	99.7%	Senior-Loken syndrome 5, 609254
IQCE	100%	100%	100%	99.9%	99%	Polydactyly, postaxial, type A7, 617642
IQSEC1	100%	100%	100%	99.8%	98.6%	Intellectual developmental disorder with short stature and behavioral abnormalities, 618687
IRAK4	100%	100%	100%	100%	99.9%	Immunodeficiency 67, 607676
IREB2	98.6%	98.6%	100%	100%	99.8%	Neurodegeneration, early-onset, with choreoathetoid movements and microcytic anemia, 618451
IRF4	100%	100%	100%	99.9%	99.1%	[Skin/hair/eye pigmentation, variation in, 8], 611724;Immunodeficiency 131, 621097
IRF7	100%	100%	100%	100%	99.2%	Immunodeficiency 39, 616345

IRF8	100%	100%	100%	100%	99.8%	Immunodeficiency 32A, mycobacteriosis, autosomal dominant, 614893;Immunodeficiency 32B, monocyte and dendritic cell deficiency, autosomal recessive, 226990
IRF9	100%	100%	100%	99.9%	99%	Immunodeficiency 65, susceptibility to viral infections, 618648
IRX5	100%	100%	99.9%	99.6%	97.1%	Hamamy syndrome, 611174
ISCA1	92.4%	92.4%	100%	100%	99.6%	Multiple mitochondrial dysfunctions syndrome 5, 617613
ISCA2	100%	100%	100%	100%	99.7%	Multiple mitochondrial dysfunctions syndrome 4, 616370
ISCU	100%	100%	100%	100%	99.7%	Myopathy with lactic acidosis, hereditary, 255125
ISG15	100%	100%	100%	100%	99.6%	Immunodeficiency 38, 616126
ITCH	92.6%	92.6%	100%	100%	99.8%	Autoimmune disease, multisystem, with facial dysmorphism, 613385
ITGA2B	100%	100%	100%	100%	99.4%	Fetomaternal alloimmune thrombocytopenia 2, 621266;Glanzmann thrombasthenia 1, 273800;Bleeding disorder, platelet-type, 16, autosomal dominant, 187800
ITGA3	100%	100%	100%	100%	99.5%	Epidermolysis bullosa, junctional 7, with interstitial lung disease and nephrotic syndrome, 614748
ITGA6	100%	100%	100%	100%	99.8%	Epidermolysis bullosa, junctional 6, with pyloric atresia, 619817

ITGA7	100%	99.9%	100%	100%	99.5%	Muscular dystrophy, congenital, due to ITGA7 deficiency, 613204
ITGA8	100%	100%	100%	100%	99.7%	Renal hypodysplasia/aplasia 1, 191830
ITGAV	99.5%	99.5%	100%	100%	99.8%	Immune dysregulation, neurodevelopmental defects, and colitis, 621375
ITGB2	100%	100%	100%	99.9%	99.7%	Leukocyte adhesion deficiency, 116920
ITGB3	100%	100%	100%	100%	99.5%	Bleeding disorder, platelet-type, 24, autosomal dominant, 619271;Glanzmann thrombasthenia 2, 619267;Fetomaternal alloimmune thrombocytopenia 1, 621264
ITGB4	100%	99.3%	100%	100%	99.4%	Epidermolysis bullosa, junctional 5B, with pyloric atresia, 226730;Epidermolysis bullosa, junctional 5A, intermediate, 619816
ITGB6	96.2%	96.2%	100%	100%	99.9%	Amelogenesis imperfecta, type IH, 616221
ITK	100%	100%	100%	100%	99.6%	Lymphoproliferative syndrome 1, 613011
ITPA	76.9%	76.9%	100%	99.9%	99.4%	[Inosine triphosphatase deficiency], 613850;Developmental and epileptic encephalopathy 35, 616647
ITPR1	100%	100%	100%	100%	99.6%	Gillespie syndrome, 206700;Spinocerebellar ataxia 29, congenital nonprogressive, 117360;Spinocerebellar ataxia 15, 606658

IVD	90%	90%	100%	100%	99.6%	Isovaleric acidemia, 243500
IYD	89.2%	89.2%	100%	100%	99.8%	Thyroid dyshormonogenesis 4, 274800
JAG2	100%	100%	100%	100%	99.1%	Muscular dystrophy, limb-girdle, autosomal recessive 27, 619566
JAGN1	100%	100%	100%	100%	99.6%	Neutropenia, severe congenital, 6, autosomal recessive, 616022
JAK3	100%	100%	100%	99.9%	98.9%	Severe combined immunodeficiency, autosomal recessive, T-negative/B-positive type, 600802
JAM2	87.3%	87.3%	100%	100%	99.7%	Basal ganglia calcification, idiopathic, 8, autosomal recessive, 618824
JAM3	98.1%	93.6%	100%	100%	99.5%	Hemorrhagic destruction of the brain, subependymal calcification, and cataracts, 613730
JKAMP	100%	100%	100%	100%	99.8%	Neurodevelopmental disorder with seizures and impaired intellectual and language development, 621533
JPH2	92.6%	92.6%	100%	99.9%	98.5%	Cardiomyopathy, dilated, 2E, 619492;Cardiomyopathy, hypertrophic, 17, 613873
JUP	100%	100%	100%	100%	99.5%	Naxos disease, 601214;?Arrhythmogenic right ventricular dysplasia 12, 611528
KALRN	98.3%	98.3%	100%	100%	99.4%	

KANK2	100%	100%	100%	100%	99.6%	Nephrotic syndrome, type 16, 617783;Palmoplantar keratoderma and woolly hair, 616099
KARS1	94.3%	94.3%	100%	100%	99.8%	Deafness, autosomal recessive 89, 613916;Leukoencephalopathy, progressive, infantile-onset, with or without deafness, 619147;?Charcot-Marie-Tooth disease, recessive intermediate, B, 613641;Deafness, congenital, and adult-onset progressive leukoencephalopathy, 619196
KATNB1	100%	100%	100%	100%	99.6%	Lissencephaly 6, with microcephaly, 616212
KATNIP	100%	100%	100%	100%	99.4%	Joubert syndrome 26, 616784
KCNA4	100%	100%	100%	100%	99.8%	Microcephaly, cataracts, impaired intellectual development, and dystonia with abnormal striatum, 618284
KCNE1	100%	100%	100%	100%	100%	Jervell and Lange-Nielsen syndrome 2, 612347;Long QT syndrome 5, 613695
KCNJ1	100%	100%	100%	100%	99.9%	Bartter syndrome, type 2, 241200
KCNJ10	100%	100%	100%	100%	99.7%	Enlarged vestibular aqueduct, digenic, 600791;SESAME syndrome, 612780

KCNJ11	100%	100%	100%	100%	99.5%	Diabetes, permanent neonatal 2, with or without neurologic features, 618856;Maturity-onset diabetes of the young, type 13, 616329;Diabetes mellitus, transient neonatal 3, 610582;Hyperinsulinemic hypoglycemia, familial, 2, 601820
KCNJ13	100%	100%	100%	100%	99.5%	Snowflake vitreoretinal degeneration, 193230;Leber congenital amaurosis 16, 614186
KCNJ16	100%	100%	100%	100%	99.9%	Hypokalemic tubulopathy and deafness, 619406
KCNMA1	100%	100%	100%	99.9%	99.4%	{Epilepsy, idiopathic generalized, susceptibility to, 16}, 618596;Paroxysmal nonkinesigenic dyskinesia, 3, with or without generalized epilepsy, 609446;Cerebellar atrophy, developmental delay, and seizures, 617643;Liang-Wang syndrome, 618729
KCNQ1	100%	100%	100%	99.9%	99.3%	Short QT syndrome 2, 609621;Atrial fibrillation, familial, 3, 607554;Long QT syndrome 1, 192500;{Long QT syndrome 1, acquired, susceptibility to}, 192500;Jervell and Lange-Nielsen syndrome, 220400
KCNV2	100%	100%	100%	100%	99.7%	Cone dystrophy with supernormal rod responses, 610356

KCTD7	100%	100%	100%	100%	99.9%	Epilepsy, progressive myoclonic 3, with or without intracellular inclusions, 611726
KDELR2	86.8%	86.8%	100%	100%	99.7%	Osteogenesis imperfecta, type XXI, 619131
KDM5A	100%	100%	100%	100%	99.8%	El Hayek-Chahrour neurodevelopmental syndrome, 620820
KDM5B	97.6%	96.9%	100%	100%	99.7%	Intellectual developmental disorder, autosomal recessive 65, 618109
KDSR	91.6%	91.6%	100%	100%	98.4%	Erythrokeratodermia variabilis et progressiva 4, 617526
KERA	100%	100%	100%	99.7%	98.6%	Cornea plana 2, autosomal recessive, 217300
KHDC3L	100%	100%	100%	100%	99.8%	Hydatidiform mole, recurrent, 2, 614293
KIAA0586	95.6%	95.6%	100%	100%	99.7%	Short-rib thoracic dysplasia 14 with polydactyly, 616546;Joubert syndrome 23, 616490
KIAA0753	100%	100%	100%	100%	99.8%	?Orofaciodigital syndrome XV, 617127;?Joubert syndrome 38, 619476;Short-rib thoracic dysplasia 21 without polydactyly, 619479
KIAA0825	100%	100%	100%	99.9%	99.6%	Polydactyly, postaxial, type A10, 618498
KIAA1549	99.5%	99.3%	100%	100%	99.3%	Retinitis pigmentosa 86, 618613
KICS2	100%	100%	100%	100%	99.8%	Intellectual developmental disorder, autosomal recessive 83, 621100

KIDINS220	100%	100%	100%	100%	99.8%	Spastic paraplegia, intellectual disability, nystagmus, and obesity, 617296;Ventriculomegaly and arthrogryposis, 619501
KIF12	100%	100%	100%	100%	99.3%	Cholestasis, progressive familial intrahepatic, 8, 619662
KIF14	97.6%	97.6%	100%	100%	99.8%	Microcephaly 20, primary, autosomal recessive, 617914;?Meckel syndrome 12, 616258
KIF1A	100%	100%	100%	100%	99.3%	NESCAV syndrome, 614255;Neuropathy, hereditary sensory, type IIC, 614213;Spastic paraplegia 30, autosomal dominant, 610357;Spastic paraplegia 30, autosomal recessive, 620607
KIF1C	100%	100%	100%	100%	99.5%	Spastic ataxia 2, autosomal recessive, 611302
KIF26A	97.6%	97.6%	100%	99.9%	99.4%	Cortical dysplasia, complex, with other brain malformations 11, 620156
KIF7	100%	100%	100%	100%	99.2%	Joubert syndrome 12, 200990;Acrocallosal syndrome, 200990;?Hydrolethalus syndrome 2, 614120;?Al-Gazali-Bakalinova syndrome, 607131
KIFBP	95.9%	95.9%	100%	99.9%	99.5%	Goldberg-Shprintzen megacolon syndrome, 609460
KIRREL1	100%	100%	100%	100%	99.2%	Nephrotic syndrome, type 23, 619201

KISS1R	100%	100%	100%	100%	98.8%	Hypogonadotropic hypogonadism 8 with or without anosmia, 614837;?Precocious puberty, central, 1, 176400
KITLG	100%	100%	100%	100%	99.9%	Hyperpigmentation with or without hypopigmentation, 145250;Waardenburg syndrome, type 2F, 619947;Deafness, autosomal dominant 69, unilateral or asymmetric, 616697;[Skin/hair/eye pigmentation 7, blond/brown hair], 611664
KIZ	100%	100%	100%	100%	99.7%	Retinitis pigmentosa 69, 615780
KL	100%	99.8%	99.8%	99.3%	98.4%	?Tumoral calcinosis, hyperphosphatemic, familial, 3, 617994
KLC2	100%	100%	100%	100%	99.4%	Spastic paraplegia, optic atrophy, and neuropathy, 609541
KLF1	100%	100%	100%	99.9%	98.6%	Blood group--Lutheran inhibitor, 111150;[Hereditary persistence of fetal hemoglobin], 613566;Anemia, dyserythropoietic congenital, type IVa, 613673;Anemia, congenital dyserythropoietic, type IVb, 620969

KLHL24	91.5%	91.3%	100%	100%	99.7%	Cardiomyopathy, familial hypertrophic, 29, with polyglucosan bodies, 620236;Epidermolysis bullosa simplex 6, generalized intermediate, with or without cardiomyopathy, 617294
KLHL3	100%	100%	100%	100%	99.6%	Pseudohypoaldosteronism, type IID, 614495
KLHL40	100%	100%	100%	99.9%	99.5%	Nemaline myopathy 8, autosomal recessive, 615348
KLHL41	100%	100%	100%	100%	99.7%	Nemaline myopathy 9, 615731
KLHL7	100%	100%	100%	100%	99.8%	Retinitis pigmentosa 42, 612943;PERCHING syndrome, 617055
KLK4	100%	100%	100%	99.9%	99.2%	Amelogenesis imperfecta, type IIA1, 204700
KLKB1	100%	100%	100%	100%	99.8%	Fletcher factor (prekallikrein) deficiency, 612423
KMT2B	100%	99.7%	100%	99.9%	98.8%	Intellectual developmental disorder, autosomal dominant 68, 619934;Dystonia 28, childhood-onset, 617284
KNL1	98.7%	98.7%	100%	100%	99.8%	Microcephaly 4, primary, autosomal recessive, 604321
KPTN	100%	100%	100%	100%	99%	Intellectual developmental disorder, autosomal recessive 41, 615637
KREMEN1	85.3%	83.4%	100%	100%	99.2%	Ectodermal dysplasia 13, hair/tooth type, 617392

KRT10	100%	100%	100%	100%	99.3%	Ichthyosis, annular epidermolytic 1, 607602;Epidermolytic hyperkeratosis 2B, autosomal recessive, 620707;Epidermolytic hyperkeratosis 2A, autosomal dominant, 620150;?Ichthyosis histrix, Lambert type, 146600;Ichthyosis with confetti, 609165
KRT14	100%	100%	100%	100%	99.3%	Epidermolysis bullosa simplex 1D, generalized, intermediate or severe, autosomal recessive, 601001;Epidermolysis bullosa simplex 1C, localized, 131800;Dermatopathia pigmentosa reticularis, 125595;Epidermolysis bullosa simplex 1A, generalized severe, 131760;Naegeli-France schetti-Jadassohn syndrome, 161000;Epidermolysis bullosa simplex 1B, generalized intermediate, 131900
KRT18	100%	100%	100%	100%	99.4%	Cirrhosis, cryptogenic, 215600;{Cirrhosis, noncryptogenic, susceptibility to}, 215600

KRT5	100%	100%	100%	100%	99.1%	Epidermolysis bullosa simplex 2A, generalized severe, 619555;Dowling-Degos disease 1, 179850;Epidermolysis bullosa simplex 2F, with mottled pigmentation, 131960;Epidermolysis bullosa simplex 2D, generalized, intermediate or severe, autosomal recessive, 619599;Epidermolysis bullosa simplex 2B, generalized intermediate, 619588;Epidermolysis bullosa simplex 2C, localized, 619594;Epidermolysis bullosa simplex 2E, with migratory circinate erythema, 609352
KRT8	100%	100%	100%	100%	99.3%	
KRT85	100%	100%	100%	100%	99.6%	Ectodermal dysplasia 4, hair/nail type, 602032
KY	100%	100%	100%	100%	99.1%	Myopathy, myofibrillar, 7, 617114
KYNU	87.7%	86.6%	100%	100%	99.9%	?Hydroxykynureninuria, 236800;Vertebral, cardiac, renal, and limb defects syndrome 2, 617661
L2HGDH	92.8%	92.8%	100%	100%	99.6%	L-2-hydroxyglutaric aciduria, 236792
LACC1	94.1%	94.1%	100%	100%	99.7%	Juvenile arthritis, 618795
LAMA1	98.9%	98.9%	100%	100%	99.6%	Poretti-Boltshauser syndrome, 615960

LAMA2	100%	100%	100%	100%	99.8%	Muscular dystrophy, limb-girdle, autosomal recessive 23, 618138;Muscular dystrophy, congenital, merosin deficient or partially deficient, 607855
LAMA3	100%	100%	100%	100%	99.6%	Epidermolysis bullosa, junctional 2A, intermediate, 619783;Epidermolysis bullosa, junctional 2C, laryngoonychocutaneous, 245660;Epidermolysis bullosa, junctional 2B, severe, 619784
LAMA5	100%	100%	100%	99.9%	99.2%	Nephrotic syndrome, type 26, 620049;?Bent bone dysplasia syndrome 2, 620076
LAMB1	100%	100%	100%	100%	99.7%	Leukoencephalopathy without lacunae, adult-onset, 621424;Leukoencephalopathy with variable cortical brain malformations and/or hydrocephalus, 615191
LAMB2	100%	100%	100%	100%	99.7%	Nephrotic syndrome, type 5, with or without ocular abnormalities, 614199;Pierson syndrome, 609049
LAMB3	100%	100%	100%	99.9%	99.6%	Epidermolysis bullosa, junctional 1B, severe, 226700;Epidermolysis bullosa, junctional 1A, intermediate, 226650;Amelogenesis imperfecta, type IA, 104530
LAMC2	100%	100%	100%	99.9%	99.5%	Epidermolysis bullosa, junctional 3B, severe, 619786;Epidermolysis bullosa, junctional 3A, intermediate, 619785

LAMC3	100%	100%	100%	100%	99.4%	Cortical malformations, occipital, 614115
LAMTOR2	96.6%	85.3%	100%	100%	99.3%	?Immunodeficiency due to defect in MAPBP-interacting protein, 610798
LARGE1	96.9%	96.9%	100%	100%	99.6%	Muscular dystrophy-dystroglycan opathy (congenital with impaired intellectual development), type B, 6, 608840;Muscular dystrophy-dystroglycan opathy (congenital with brain and eye anomalies), type A, 6, 613154
LARP7	100%	100%	100%	100%	99.8%	Alazami syndrome, 615071
LARS1	94.3%	94.3%	100%	100%	99.7%	?Infantile liver failure syndrome 1, 615438
LARS2	96.2%	96.2%	100%	100%	99.8%	Perrault syndrome 4, 615300;Hydrops, lactic acidosis, and sideroblastic anemia, 617021
LAT	100%	100%	100%	100%	99.7%	Immunodeficiency 52, 617514
LBR	100%	100%	100%	100%	99.7%	Pelger-Huet anomaly, 169400;?Reynolds syndrome, 613471;Rhizomelic skeletal dysplasia with or without Pelger-Huet anomaly, 618019;Greenberg skeletal dysplasia, 215140
LCA5	100%	100%	100%	100%	99.5%	Leber congenital amaurosis 5, 604537
LCAT	100%	100%	100%	99.9%	99%	Fish-eye disease, 136120;Norum disease, 245900
LCK	98%	98%	100%	100%	99.7%	Immunodeficiency 22, 615758

LCP2	100%	100%	100%	100%	99.6%	Immunodeficiency 81, 619374
LCT	100%	100%	100%	100%	99.5%	Lactase deficiency, congenital, 223000
LDHA	100%	100%	100%	100%	99.6%	Glycogen storage disease XI, 612933
LDHD	100%	100%	100%	100%	99.5%	D-lactic aciduria with susceptibility to gout, 245450
LDLR	100%	100%	100%	100%	99.1%	LDL cholesterol level QTL2, 143890;Hypercholesterolemia, familial, 1, 143890
LEMD2	100%	100%	100%	100%	99.8%	Marbach-Rustad progeroid syndrome, 619322;Cataract 46, juvenile-onset, 212500
LEP	100%	100%	100%	100%	99.5%	Obesity, morbid, due to leptin deficiency, 614962
LEPR	94.6%	94.6%	100%	100%	99.7%	Obesity, morbid, due to leptin receptor deficiency, 614963
LETM1	92.4%	89.8%	100%	100%	99.5%	Neurodegeneration, childhood-onset, with multisystem involvement due to mitochondrial dysfunction, 620089
LFNG	99.9%	97.1%	100%	99.9%	98.8%	Spondylocostal dysostosis 3, autosomal recessive, 609813
LGI1	100%	100%	100%	100%	99.8%	Developmental and epileptic encephalopathy 121, 621475;Epilepsy, familial temporal lobe, 1, 600512

LGI3	100%	100%	100%	100%	99.4%	Intellectual developmental disorder with muscle tone abnormalities and distal skeletal defects, 620007
LGI4	100%	100%	100%	99.9%	99.4%	Arthrogryposis multiplex congenita 1, neurogenic, with myelin defect, 617468
LHB	100%	100%	100%	99.8%	98%	Hypogonadotropic hypogonadism 23 with or without anosmia, 228300
LHCGR	100%	100%	100%	100%	99.7%	Leydig cell adenoma, somatic, with precocious puberty, 176410;Leydig cell hypoplasia with pseudohermaphroditism, 238320;Leydig cell hypoplasia with hypergonadotropic hypogonadism, 238320;Luteinizing hormone resistance, female, 238320;Precocious puberty, male, 176410
LHFPL5	100%	100%	100%	99.8%	99.2%	Deafness, autosomal recessive 67, 610265
LHX3	100%	100%	100%	99.6%	98.9%	Pituitary hormone deficiency, combined, 3, 221750
LIAS	100%	100%	100%	100%	99.9%	Hyperglycinemia, lactic acidosis, and seizures, 614462
LIFR	100%	100%	100%	100%	99.7%	Stuve-Wiedemann syndrome/Schwartz-Jampel type 2 syndrome, 601559
LIG1	100%	100%	100%	99.9%	99.1%	Immunodeficiency 96, 619774
LIG3	100%	100%	100%	100%	99.6%	Mitochondrial DNA depletion syndrome 20 (MNGIE type), 619780

LIG4	100%	100%	100%	100%	99.8%	LIG4 syndrome, 606593;{Multiple myeloma, resistance to}, 254500
LIM2	100%	100%	100%	100%	98.6%	Cataract 19, multiple types, 615277
LIMS2	100%	99.9%	100%	100%	99.8%	?Muscular dystrophy, autosomal recessive, with cardiomyopathy and triangular tongue, 616827
LINGO1	100%	100%	100%	99.9%	98.1%	Intellectual developmental disorder, autosomal recessive 64, 618103
LINS1	100%	100%	100%	100%	99.9%	Intellectual developmental disorder, autosomal recessive 27, 614340
LIPA	95.6%	95.2%	100%	100%	99.9%	Wolman disease, 620151;Cholesteryl ester storage disease, 278000
LIPE	100%	100%	100%	100%	99.4%	Lipodystrophy, familial partial, type 6, 615980
LIPH	100%	100%	100%	100%	99.6%	Hypotrichosis 7, 604379;Woolly hair, autosomal recessive 2 with or without hypotrichosis, 604379
LIPN	100%	100%	100%	100%	99.8%	Ichthyosis, congenital, autosomal recessive 8, 613943
LIPT1	100%	100%	100%	100%	99.8%	Lipoyltransferase 1 deficiency, 616299
LIPT2	100%	100%	100%	100%	99%	Encephalopathy, neonatal severe, with lactic acidosis and brain abnormalities, 617668
LMAN1	100%	100%	100%	100%	99.7%	Combined factor V and VIII deficiency, 227300

LMAN2L	100%	100%	100%	100%	99.9%	?Intellectual developmental disorder, autosomal dominant 69, 617863;?Intellectual developmental disorder, autosomal recessive 52, 616887
LMBR1	96.1%	95.5%	100%	100%	99.7%	Syndactyly, type IV, 186200;Laurin-Sandrow syndrome, 135750;Acheiropody, 200500;Triphalangeal thumb-polysyndactyly syndrome, 190605
LMBRD1	86.4%	86.4%	100%	99.9%	99.6%	Methylmalonic aciduria and homocystinuria, cblF type, 277380
LMF1	91.5%	91.5%	100%	100%	99.8%	Lipase deficiency, combined, 246650
LMNA	100%	100%	100%	99.9%	99.3%	Mandibuloacral dysplasia, 248370;Heart-hand syndrome, Slovenian type, 610140;Cardiomyopathy, dilated, 1A, 115200;Emery-Dreifuss muscular dystrophy 3, autosomal recessive, 616516;Restrictive dermopathy 2, 619793;Charcot-Marie-Tooth disease, type 2B1, 605588;Emery-Dreifuss muscular dystrophy 2, autosomal dominant, 181350;Hutchinson-Gilford progeria, 176670;Lipodystrophy, familial partial, type 2, 151660;Muscular dystrophy, congenital, 613205;Malouf syndrome, 212112

LMNB2	92.9%	92.9%	100%	99.8%	98.3%	Microcephaly 27, primary, autosomal dominant, 619180;?Epilepsy, progressive myoclonic, 9, 616540;{Lipodystrophy, partial, acquired, susceptibility to}, 608709
LMOD2	100%	100%	100%	100%	99.2%	Cardiomyopathy, dilated, 2G, 619897
LMOD3	100%	100%	100%	99.9%	99.2%	Nemaline myopathy 10, 616165
LNPK	85.8%	85.8%	100%	100%	99.8%	Neurodevelopmental disorder with epilepsy and hypoplasia of the corpus callosum, 618090
LONP1	99.3%	97%	100%	100%	99.5%	CODAS syndrome, 600373
LOXHD1	100%	100%	100%	100%	99.7%	Deafness, autosomal recessive 77, 613079
LPAR6	100%	100%	100%	100%	99.9%	Hypotrichosis 8, 278150;Woolly hair, autosomal recessive 1, with or without hypotrichosis, 278150
LPIN1	94%	94%	100%	100%	99.7%	Myoglobinuria, acute recurrent, autosomal recessive, 268200
LPIN2	99.3%	99.2%	100%	99.9%	99.4%	Majeed syndrome, 609628
LPL	100%	100%	100%	100%	99.6%	Lipoprotein lipase deficiency, 238600;[High density lipoprotein cholesterol level QTL 11], 238600;Combined hyperlipidemia, familial, 144250

LRAT	100%	100%	100%	100%	99.4%	Leber congenital amaurosis 14, 613341;Retinal dystrophy, early-onset severe, 613341;Retinitis pigmentosa, juvenile, 613341
LRBA	99.8%	99.8%	100%	100%	99.8%	Immunodeficiency, common variable, 8, with autoimmunity, 614700
LRIG2	98%	98%	100%	100%	99.8%	Urofacial syndrome 2, 615112
LRIT3	100%	100%	100%	100%	99.9%	Night blindness, congenital stationary (complete), 1F, autosomal recessive, 615058
LRMDA	97.8%	97.8%	100%	100%	99.4%	Albinism, oculocutaneous, type VII, 615179
LRP1	100%	100%	100%	100%	99.3%	?Keratosis pilaris atrophicans, 604093;Developmental dysplasia of the hip 3, 620690
LRP2	100%	100%	100%	100%	99.8%	Donnai-Barrow syndrome, 222448
LRP4	100%	100%	100%	99.9%	99.4%	?Myasthenic syndrome, congenital, 17, 616304;Sclerosteosis 2, 614305;Cenani-Lenz syndactyly syndrome, 212780

LRP5	100%	100%	100%	100%	99%	Osteopetrosis, autosomal dominant 1, 607634;Polycystic liver disease 4 with or without kidney cysts, 617875;Endosteal hyperostosis, 144750;Osteoporosis-pseudoglioma syndrome, 259770;[Bone mineral density variability 1, high bone mass], 601884;Exudative vitreoretinopathy 4, 601813
LRPAP1	84.8%	84.8%	100%	100%	99.4%	Myopia 23, autosomal recessive, 615431
LRPPRC	96.8%	96.7%	100%	100%	99.8%	Mitochondrial complex IV deficiency, nuclear type 5, (French-Canadian), 220111
LRRC56	100%	100%	100%	100%	99.6%	Ciliary dyskinesia, primary, 39, 618254
LRRK1	100%	100%	100%	100%	99.5%	Osteosclerotic metaphyseal dysplasia, 615198
LRSAM1	100%	100%	100%	100%	99.4%	Charcot-Marie-Tooth disease, axonal, type 2P, 614436
LRTOMT	100%	100%	100%	100%	99.7%	Deafness, autosomal recessive 63, 611451
LSM1	100%	100%	100%	100%	99.2%	FICUS syndrome, 621193
LSM7	100%	100%	100%	100%	98.7%	Leukodystrophy and cerebellar atrophy, 621191
LSS	100%	100%	100%	100%	99.8%	Hypotrichosis 14, 618275;Cataract 44, 616509;Alopecia-intellectual disability syndrome 4, 618840

LTBP1	100%	100%	100%	100%	99.5%	Cutis laxa, autosomal recessive, type IIE, 619451
LTBP2	100%	100%	100%	100%	99.7%	Glaucoma 3, primary congenital, D, 613086;Microspherophakia and/or megalocornea, with ectopia lentis and with or without secondary glaucoma, 251750;?Weill-Marchesani syndrome 3, recessive, 614819
LTBP3	100%	100%	100%	99.9%	98.9%	Dental anomalies and short stature, 601216;Geleophysic dysplasia 3, 617809
LTBP4	100%	100%	100%	100%	99%	Cutis laxa, autosomal recessive, type IC, 613177
LTC4S	100%	100%	99.8%	99.2%	98.3%	Leukotriene C4 synthase deficiency, 614037
LYRM7	100%	100%	100%	100%	99.4%	Mitochondrial complex III deficiency, nuclear type 8, 615838
LYSET	100%	100%	100%	100%	99.7%	Dysostosis multiplex, Ain-Naz type, 619345
LYST	99.5%	99.5%	100%	100%	99.9%	Chediak-Higashi syndrome, 214500
LZTFL1	100%	100%	100%	100%	99.9%	Bardet-Biedl syndrome 17, 615994
LZTR1	100%	100%	100%	100%	99.6%	Noonan syndrome 2, 605275;Noonan syndrome 10, 616564;{Schwannomatosis-2, susceptibility to}, 615670
MAB21L1	100%	100%	100%	100%	99.3%	Cerebellar, ocular, craniofacial, and genital syndrome, 618479

MAB21L2	100%	100%	100%	100%	99.2%	Microphthalmia/coloboma and skeletal dysplasia syndrome, 615877
MAD1L1	100%	98.9%	100%	100%	99.5%	Prostate cancer, somatic, 176807;Mosaic variegated aneuploidy syndrome 7 with inflammation and tumor predisposition, 620189;Lymphoma, B-cell, somatic
MADD	98.7%	98.7%	100%	100%	99.5%	Neurodevelopmental disorder with dysmorphic facies, impaired speech and hypotonia, 619005;DEEAH syndrome, 619004
MAG	100%	100%	100%	100%	99.3%	Spastic paraplegia 75, autosomal recessive, 616680
MAGI2	99.6%	97.9%	100%	99.8%	99%	Nephrotic syndrome, type 15, 617609
MAK	100%	100%	100%	100%	99.8%	Retinitis pigmentosa 62, 614181
MALT1	93.9%	93.9%	100%	100%	99.7%	Immunodeficiency 12, 615468
MAN1B1	99.8%	98.1%	100%	100%	99.6%	Rafiq syndrome, 614202
MAN2A2	100%	100%	100%	100%	99.4%	
MAN2B1	100%	100%	100%	100%	99.1%	Mannosidosis, alpha-, types I and II, 248500
MAN2B2	89.8%	87.3%	100%	100%	99.6%	Congenital disorder of glycosylation type 1EE with or without immunodeficiency, 621140
MAN2C1	98.4%	98.4%	100%	99.9%	99.5%	Congenital disorder of deglycosylation 2, 619775

MANBA	99.4%	97.5%	100%	100%	99.5%	Mannosidosis, beta, 248510
MANF	100%	100%	100%	99.9%	99%	Diabetes, deafness, developmental delay, and short stature syndrome, 620651
MAP3K20	100%	100%	100%	100%	99.9%	Centronuclear myopathy 6 with fiber-type disproportion, 617760;Split-foot malformation with mesoaxial polydactyly, 616890
MAPKAPK5	100%	100%	100%	100%	99.4%	Neurocardiofaciodigital syndrome, 619869
MAPKBP1	100%	100%	100%	100%	99.8%	Nephronophthisis 20, 617271
MAPT	95.4%	95.4%	100%	99.9%	99.2%	Supranuclear palsy, progressive, 601104;Frontotemporal dementia 1, with or without parkinsonism, 600274;Supranuclear palsy, progressive atypical, 260540;{Parkinson disease, susceptibility to}, 168600;Pick disease, 172700
MARS1	89.3%	88.7%	100%	100%	99.5%	Spastic paraplegia 70, autosomal recessive, 620323;Interstitial lung and liver disease, 615486;?Trichothiodystrophy 9, nonphotosensitive, 619692;Charcot-Marie-Tooth disease, axonal, type 2U, 616280
MARS2	100%	100%	100%	99.9%	99.2%	?Combined oxidative phosphorylation deficiency 25, 616430;Spastic ataxia 3, autosomal recessive, 611390
MARVELD2	100%	100%	100%	100%	99.8%	Deafness, autosomal recessive 49, 610153

MASP1	100%	100%	100%	100%	99.6%	3MC syndrome 1, 257920
MAT1A	100%	99.9%	100%	99.9%	99.4%	Hypermethioninemia, persistent, autosomal dominant, due to methionine adenosyltransferase I/III deficiency, 250850;Methionine adenosyltransferase deficiency, autosomal recessive, 250850
MATN3	100%	100%	100%	100%	99.8%	{Osteoarthritis susceptibility 2}, 140600;Spondyloepimetaphyseal dysplasia, Borochowitz-Cormier-Daire type, 608728;Epiphyseal dysplasia, multiple, 5, 607078
MBOAT7	100%	100%	100%	100%	99.6%	Intellectual developmental disorder, autosomal recessive 57, 617188
MBTPS1	100%	100%	100%	100%	99.4%	Spondyloepiphyseal dysplasia, Kondo-Fu type, 618392;CAOP syndrome, 621252
MC2R	100%	100%	100%	100%	99.2%	Glucocorticoid deficiency, due to ACTH unresponsiveness, 202200
MCAT	95.9%	95.9%	100%	100%	99.6%	Optic atrophy 15, 620583
MCCC1	94.5%	94.5%	100%	100%	99.8%	3-Methylcrotonyl-CoA carboxylase 1 deficiency, 210200
MCCC2	93.5%	93.5%	100%	100%	99.9%	3-Methylcrotonyl-CoA carboxylase 2 deficiency, 210210
MCEE	100%	100%	100%	100%	99.5%	Methylmalonyl-CoA epimerase deficiency, 251120

MCFD2	100%	100%	100%	100%	99.6%	Factor V and factor VIII, combined deficiency of, 613625
MCIDAS	100%	100%	100%	99.9%	98.9%	Ciliary dyskinesia, primary, 42, 618695
MCM10	100%	100%	100%	100%	99.7%	Immunodeficiency 80 with or without cardiomyopathy, 619313
MCM3AP	100%	100%	100%	100%	99.5%	Peripheral neuropathy, autosomal recessive, with or without impaired intellectual development, 618124
MCM4	95.4%	95.4%	100%	100%	99.8%	Immunodeficiency 54, 609981
MCM5	96.9%	96.9%	100%	100%	99.6%	?Meier-Gorlin syndrome 8, 617564
MCM9	100%	100%	100%	100%	99.8%	Ovarian dysgenesis 4, 616185
MCOLN1	98.3%	95.7%	100%	100%	99.7%	Lisch epithelial corneal dystrophy, 620763;Mucolipidosis IV, 252650
MCPH1	94.6%	92.8%	100%	100%	99.7%	Microcephaly 1, primary, autosomal recessive, 251200
MDFIC	100%	100%	100%	100%	99.8%	Lymphatic malformation 12, 620014
MDGA2	100%	100%	100%	100%	99.9%	
MDH2	100%	100%	100%	100%	98.7%	Developmental and epileptic encephalopathy 51, 617339
MDM1	100%	100%	100%	100%	99.6%	
MECR	93.5%	93.5%	100%	100%	99.6%	?Optic atrophy 16, 620629;Dystonia, childhood-onset, with optic atrophy and basal ganglia abnormalities, 617282

MED16	100%	100%	99.9%	99.6%	97.3%	Guillouet-Gordon syndrome, 621220
MED17	100%	100%	100%	100%	99.6%	Microcephaly, postnatal progressive, with seizures and brain atrophy, 613668
MED23	100%	100%	100%	100%	99.8%	Intellectual developmental disorder, autosomal recessive 18, with or without epilepsy, 614249
MED25	100%	100%	100%	100%	99.4%	Basel-Vanagaite-Smirin-Yosef syndrome, 616449
MED27	100%	100%	100%	100%	99.5%	Neurodevelopmental disorder with spasticity, cataracts, and cerebellar hypoplasia, 619286
MEFV	96.1%	96.1%	100%	100%	99.6%	Neutrophilic dermatosis, acute febrile, 608068;Familial Mediterranean fever, AR, 249100;Familial Mediterranean fever, AD, 134610
MEGF10	100%	100%	100%	100%	99.6%	Congenital myopathy 10A, severe variant, 614399;Congenital myopathy 10B, mild variant, 620249
MEGF8	100%	100%	100%	99.8%	98.9%	Carpenter syndrome 2, 614976
MEOX1	100%	100%	100%	100%	98.8%	Klippel-Feil syndrome 2, 214300
MERTK	100%	100%	100%	100%	99.8%	Retinitis pigmentosa 38, 613862
MESD	91.8%	91.8%	100%	99.9%	99.1%	Osteogenesis imperfecta, type XX, 618644

MESP2	100%	99.9%	100%	100%	99.8%	Spondylocostal dysostosis 2, autosomal recessive, 608681
MET	100%	100%	100%	100%	99.8%	Renal cell carcinoma, papillary, 1, familial and somatic, 605074;?Arthrogryposis, distal, type 11, 620019;Hepatocellular carcinoma, childhood type, somatic, 114550;{Osteofibrous dysplasia, susceptibility to}, 607278;?Deafness, autosomal recessive 97, 616705
METTL23	100%	100%	100%	100%	100%	Intellectual developmental disorder, autosomal recessive 44, 615942
METTL5	100%	100%	100%	100%	99.8%	Intellectual developmental disorder, autosomal recessive 72, 618665
MFF	89.1%	89.1%	100%	100%	99.9%	Encephalopathy due to defective mitochondrial and peroxisomal fission 2, 617086
MFN2	100%	100%	100%	100%	99.7%	Lipomatosis, multiple symmetric, with or without peripheral neuropathy, 151800;Charcot-Marie-Tooth disease, axonal, type 2A2A, 609260;Charcot-Marie-Tooth disease, axonal, type 2A2B, 617087;Hereditary motor and sensory neuropathy VIA, 601152
MFRP	100%	100%	100%	100%	99.4%	Microphthalmia, isolated 5, 611040;Nanophthalmos 2, 609549

MFSD2A	100%	100%	100%	100%	99.6%	Neurodevelopmental disorder with progressive microcephaly, spasticity, and brain abnormalities, 616486
MFSD8	100%	100%	100%	100%	100%	Macular dystrophy with central cone involvement, 616170;Ceroid lipofuscinosis, neuronal, 7, 610951
MGAT2	100%	100%	100%	99.8%	99%	Congenital disorder of glycosylation, type IIa, 212066
MGME1	100%	100%	100%	100%	99.5%	Mitochondrial DNA depletion syndrome 11, 615084
MGP	100%	100%	100%	100%	99.7%	Keutel syndrome, 245150
MICOS13	100%	100%	100%	100%	99.6%	Combined oxidative phosphorylation deficiency 37, 618329
MICU1	86.7%	86.7%	100%	100%	99.8%	Myopathy with extrapyramidal signs, 615673
MINAR2	100%	100%	100%	100%	99.8%	Deafness, autosomal recessive 120, 620238
MINPP1	100%	100%	100%	100%	99.9%	{Thyroid carcinoma, follicular}, 188470;Pontocerebellar hypoplasia, type 16, 619527
MIPEP	100%	100%	100%	100%	99.9%	Combined oxidative phosphorylation deficiency 31, 617228

MITF	94.7%	94.7%	100%	100%	99.9%	Waardenburg syndrome, type 2A, 193510;{Melanoma, cutaneous malignant, susceptibility to, 8}, 614456;Tietz albinism-deafness syndrome, 103500;COMMAD syndrome, 617306
MKKS	100%	100%	100%	100%	100%	McKusick-Kaufman syndrome, 236700;Bardet-Biedl syndrome 6, 605231
MKS1	99.1%	99%	100%	100%	99.7%	Bardet-Biedl syndrome 13, 615990;Meckel syndrome 1, 249000;Joubert syndrome 28, 617121
MLC1	100%	100%	100%	100%	99.2%	Megalencephalic leukoencephalopathy with subcortical cysts 1, 604004
MLH1	100%	100%	100%	100%	99.7%	Lynch syndrome 2, 609310;Muir-Torre syndrome, 158320;Mismatch repair cancer syndrome 1, 276300
MLIP	100%	100%	100%	100%	99.7%	Myopathy with myalgia, increased serum creatine kinase, and with or without episodic rhabdomyolysis, 620138
MLYCD	100%	100%	100%	100%	99.4%	Malonyl-CoA decarboxylase deficiency, 248360
MMAA	100%	100%	100%	100%	99.8%	Methylmalonic aciduria, vitamin B12-responsive, cblA type, 251100
MMAB	91.8%	91.8%	100%	99.9%	99.7%	Methylmalonic aciduria, vitamin B12-responsive, cblB type, 251110

MMACHC	100%	100%	100%	100%	99.5%	Methylmalonic aciduria and homocystinuria, cblC type, 277400
MMADHC	79.3%	79.3%	100%	100%	99.7%	Methylmalonic aciduria and homocystinuria, cblD type, 277410;Methylmalonic aciduria, cblD type, 620953;Homocystinuria -megaloblastic anemia, cblD type, 620952
MME	97.7%	97.7%	100%	100%	99.7%	?Spinocerebellar ataxia 43, 617018;Charcot-Marie-Tooth disease, axonal, type 2T, 617017
MMP13	92.2%	92.2%	100%	100%	99.4%	?Spondyloepimetaphyseal dysplasia, Missouri type, 602111;Metaphyseal anadysplasia 1, 602111;Metaphyseal dysplasia, Spahr type, 250400
MMP14	94.9%	94.9%	100%	100%	99.5%	Winchester syndrome, 277950
MMP2	100%	100%	100%	100%	99.5%	Multicentric osteolysis, nodulosis, and arthropathy, 259600
MMP20	100%	100%	100%	100%	99.8%	Amelogenesis imperfecta, type IIA2, 612529
MMP21	100%	100%	100%	100%	99.4%	Heterotaxy, visceral, 7, autosomal, 616749
MMP9	100%	100%	100%	99.9%	98.8%	Metaphyseal anadysplasia 2, 613073
MMUT	100%	100%	100%	100%	99.7%	Methylmalonic aciduria, mut(0) type, 251000
MOCOS	97.6%	97.6%	100%	100%	99.6%	Xanthinuria, type II, 603592
MOCS1	100%	100%	100%	100%	99.4%	Molybdenum cofactor deficiency A, 252150

MOCS2	100%	100%	100%	100%	99.9%	Molybdenum cofactor deficiency B1, 252160
MOGS	100%	100%	100%	100%	99.5%	Congenital disorder of glycosylation, type IIb, 606056
MPC1	100%	100%	100%	100%	99.9%	Mitochondrial pyruvate carrier deficiency, 614741
MPDU1	100%	100%	100%	100%	98.8%	Congenital disorder of glycosylation, type If, 609180
MPDZ	100%	100%	100%	100%	99.7%	Hydrocephalus, congenital, 2, with or without brain or eye anomalies, 615219
MPI	100%	100%	100%	100%	99.7%	Congenital disorder of glycosylation, type Ib, 602579
MPL	100%	100%	100%	99.9%	99.3%	Myelofibrosis with myeloid metaplasia, somatic, 254450;Amegakaryocytic thrombocytopenia, congenital, 1, 604498;Thrombocythemia 2, 601977
MPLKIP	100%	100%	100%	100%	99%	Trichothiodystrophy 4, nonphotosensitive, 234050
MPO	100%	100%	100%	100%	99.5%	{Alzheimer disease, susceptibility to}, 104300;Myeloperoxidase deficiency, 254600;{Lung cancer, protection against, in smokers}
MPV17	100%	100%	100%	100%	99.6%	Charcot-Marie-Tooth disease, axonal, type 2EE, 618400;Mitochondrial DNA depletion syndrome 6 (hepatocerebral type), 256810

MPZ	100%	100%	100%	100%	98.9%	Charcot-Marie-Tooth disease, type 2I, 607677;Dejerine-Sottas disease, 145900;Charcot-Marie-Tooth disease, type 1B, 118200;Roussy-Levy syndrome, 180800;Charcot-Marie-Tooth disease, dominant intermediate D, 607791;Hypomyelinating neuropathy, congenital, 2, 618184;Charcot-Marie-Tooth disease, type 2J, 607736
MPZL2	87.9%	87.9%	100%	100%	99.4%	Deafness, autosomal recessive 111, 618145
MRAP	100%	100%	100%	100%	99.6%	Glucocorticoid deficiency 2, 607398
MRE11	97.4%	97.4%	100%	100%	99.9%	Ataxia-telangiectasia-like disorder 1, 604391
MRM2	87.4%	87.3%	100%	99.7%	98.3%	Mitochondrial DNA depletion syndrome 17, 618567
MRPL3	100%	100%	100%	100%	99.9%	Combined oxidative phosphorylation deficiency 9, 614582
MRPL39	100%	100%	100%	100%	99.6%	Combined oxidative phosphorylation deficiency 59, 620646
MRPL44	100%	100%	100%	100%	99.7%	Combined oxidative phosphorylation deficiency 16, 615395
MRPL49	100%	100%	100%	100%	99.3%	Combined oxidative phosphorylation deficiency 60, 621195
MRPS14	85.2%	85.2%	100%	100%	99.7%	Combined oxidative phosphorylation deficiency 38, 618378
MRPS16	100%	100%	100%	100%	99%	Combined oxidative phosphorylation deficiency 2, 610498

MRPS2	100%	100%	100%	99.9%	99.6%	Combined oxidative phosphorylation deficiency 36, 617950
MRPS22	86.6%	86.6%	100%	100%	99.9%	Ovarian dysgenesis 7, 618117;Combined oxidative phosphorylation deficiency 5, 611719
MRPS34	100%	100%	100%	100%	99.5%	Combined oxidative phosphorylation deficiency 32, 617664
MS4A1	100%	100%	100%	100%	99.9%	?Immunodeficiency, common variable, 5, 613495
MSH2	90.6%	90.6%	100%	100%	99.6%	Lynch syndrome 1, 120435;Muir-Torre syndrome, 158320;Mismatch repair cancer syndrome 2, 619096
MSH3	98%	98%	100%	100%	99.7%	Familial adenomatous polyposis 4, 617100;Endometrial carcinoma, somatic, 608089
MSH6	100%	100%	100%	100%	99.5%	Lynch syndrome 5, 614350;Mismatch repair cancer syndrome 3, 619097;{Endometrial cancer, familial}, 608089
MSMO1	100%	100%	100%	100%	99.9%	Microcephaly, congenital cataract, and psoriasiform dermatitis, 616834
MSRB3	100%	100%	100%	100%	99.9%	Deafness, autosomal recessive 74, 613718
MSTO1	100%	100%	99.9%	99.1%	97.7%	Myopathy, mitochondrial, and ataxia, 617675

MTFMT	91.8%	91.8%	100%	99.9%	98.6%	Combined oxidative phosphorylation deficiency 15, 614947;Mitochondrial complex I deficiency, nuclear type 27, 618248
MTHFD1	94.9%	92%	100%	100%	99.7%	{Neural tube defects, folate-sensitive, susceptibility to}, 601634;Combined immunodeficiency and megaloblastic anemia with or without hyperhomocysteinemia, 617780
MTHFR	100%	100%	100%	100%	99.5%	{Vascular disease, susceptibility to};Homocystinuria due to MTHFR deficiency, 236250;{Thromboembolism, susceptibility to}, 188050;{Schizophrenia, susceptibility to}, 181500;{Neural tube defects, susceptibility to}, 601634
MTHFS	100%	100%	100%	100%	99.7%	Neurodevelopmental disorder with microcephaly, epilepsy, and hypomyelination, 618367
MTMR2	100%	100%	100%	99.9%	99.6%	Charcot-Marie-Tooth disease, type 4B1, 601382
MTO1	94.8%	91.9%	100%	100%	99.7%	Combined oxidative phosphorylation deficiency 10, 614702
MTPAP	96.6%	96.6%	100%	100%	99.4%	?Spastic ataxia 4, autosomal recessive, 613672
MTR	100%	100%	100%	100%	99.7%	{Neural tube defects, folate-sensitive, susceptibility to}, 601634;Homocystinuria-megaloblastic anemia, cblG complementation type, 250940

MTRFR	100%	100%	100%	100%	99.7%	Spastic paraplegia 55, autosomal recessive, 615035;Combined oxidative phosphorylation deficiency 7, 613559
MTRR	100%	100%	100%	100%	99.7%	Homocystinuria-megaloblastic anemia, cbl E type, 236270;{Neural tube defects, folate-sensitive, susceptibility to}, 601634
MTTP	100%	100%	100%	100%	99.8%	Abetalipoproteinemia, 200100
MTX2	87.1%	87.1%	100%	100%	99.2%	Mandibuloacral dysplasia progeroid syndrome, 619127
MUSK	100%	100%	100%	100%	99.7%	Fetal akinesia deformation sequence 1, 208150;Myasthenic syndrome, congenital, 9, associated with acetylcholine receptor deficiency, 616325
MUTYH	100%	100%	100%	99.9%	99.5%	Adenomas, multiple colorectal, 608456;Gastric cancer, somatic, 613659
MVK	100%	100%	100%	100%	99.8%	Hyper-IgD syndrome, 260920;Porokeratosis 3, multiple types, 175900;Mevalonic aciduria, 610377
MYBPC1	100%	100%	100%	100%	99.8%	Congenital myopathy 16, 618524;Lethal congenital contracture syndrome 4, 614915;Arthrogryposis, distal, type 1B, 614335
MYD88	100%	100%	100%	100%	99.9%	Macroglobulinemia, Waldenstrom, somatic, 153600;Immunodeficiency 68, 612260

MYF5	100%	100%	100%	100%	99.9%	Ophthalmoplegia, external, with rib and vertebral anomalies, 618155
MYH11	100%	100%	100%	100%	99.3%	Megacystis-microcolon-intestinal hypoperistalsis syndrome 2, 619351;Aortic aneurysm, familial thoracic 4, 132900;Visceral myopathy 2, 619350
MYH2	100%	100%	100%	99.9%	99.8%	Congenital myopathy 6 with ophthalmoplegia, 605637
MYH3	100%	100%	100%	100%	99.5%	Contractures, pterygia, and spondylocarpostarsal fusion syndrome 1A, 178110;Contractures, pterygia, and spondylocarpotarsal fusion syndrome 1B, 618469;Arthrogryposis, distal, type 2B3 (Sheldon-Hall), 618436;Arthrogryposis, distal, type 2A (Freeman-Sheldon), 193700
MYL1	100%	100%	100%	100%	99.7%	Congenital myopathy 14, 618414
MYL11	100%	100%	100%	100%	99.1%	Arthrogryposis, distal, type 1C, 619110
MYL2	100%	100%	100%	100%	99.4%	Cardiomyopathy, hypertrophic, 10, 608758;Myopathy, myofibrillar, 12, infantile-onset, with cardiomyopathy, 619424
MYL3	81.6%	79.9%	100%	100%	99.2%	Cardiomyopathy, hypertrophic, 8, 608751

MYLK	99.2%	99.2%	100%	100%	99.4%	Megacystis-microcolon-intestinal hypoperistalsis syndrome 1, 249210;Aortic aneurysm, familial thoracic 7, 613780
MYMK	100%	100%	100%	100%	99.4%	Carey-Fineman-Ziter syndrome, 254940
MYMX	100%	100%	100%	100%	100%	Carey-Fineman-Ziter syndrome 2, 619941
MYO15A	100%	100%	100%	100%	99.3%	Deafness, autosomal recessive 3, 600316
MYO18B	100%	100%	100%	99.9%	99.4%	Klippel-Feil syndrome 4, autosomal recessive, with myopathy and facial dysmorphism, 616549
MYO1E	100%	100%	100%	100%	99.6%	Glomerulosclerosis, focal segmental, 6, 614131
MYO3A	100%	100%	100%	100%	99.8%	Deafness, autosomal recessive 30, 607101;Deafness, autosomal dominant 90, 620722
MYO5A	99%	99%	100%	100%	99.7%	Griscelli syndrome, type 1, 214450
MYO5B	100%	100%	100%	100%	99.5%	Diarrhea 2, with microvillus atrophy, with or without cholestasis, 251850;Cholestasis, progressive familial intrahepatic, 10, 619868
MYO6	100%	100%	100%	100%	99.9%	Deafness, autosomal dominant 22, with hypertrophic cardiomyopathy, 606346;Deafness, autosomal dominant 22, 606346;Deafness, autosomal recessive 37, 607821

MYO7A	100%	100%	100%	100%	99.5%	Deafness, autosomal recessive 2, 600060;Usher syndrome, type 1B, 276900;Deafness, autosomal dominant 11, 601317
MYO9A	100%	100%	100%	100%	99.7%	Myasthenic syndrome, congenital, 24, presynaptic, 618198
MYOD1	100%	100%	100%	99.9%	97.5%	Congenital myopathy 17, 618975
MYORG	100%	100%	100%	99.9%	99.1%	Basal ganglia calcification, idiopathic, 7, autosomal recessive, 618317
MYPN	98.4%	98.4%	100%	100%	99.6%	Cardiomyopathy, hypertrophic, 22, 615248;Congenital myopathy 24, 617336;Cardiomyopathy, familial restrictive, 4, 615248;Cardiomyopathy, dilated, 1KK, 615248
MYSM1	100%	100%	100%	100%	99.7%	Bone marrow failure syndrome 4, 618116
MYZAP	80%	80%	100%	99.9%	99.7%	Cardiomyopathy, dilated, 2K, 620894
NAA20	100%	100%	100%	100%	99.9%	Intellectual developmental disorder, autosomal recessive 73, 619717
NAA60	100%	100%	100%	100%	99.5%	Basal ganglia calcification, idiopathic, 9, autosomal recessive, 620786
NADK2	100%	100%	100%	99.9%	99.2%	2,4-dienoyl-CoA reductase deficiency, 616034
NADSYN1	93.5%	93.5%	100%	100%	99.5%	Vertebral, cardiac, renal, and limb defects syndrome 3, 618845

NAE1	100%	100%	100%	100%	99.8%	Neurodevelopmental disorder with dysmorphic facies and ischiopubic hypoplasia, 620210
NAGA	90%	88%	100%	100%	99.5%	Schindler disease, type I, 609241;Kanzaki disease, 609242;Schindler disease, type III, 609241
NAGLU	100%	100%	100%	100%	99.4%	?Charcot-Marie-Tooth disease, axonal, type 2V, 616491;Mucopolysaccharidosis type IIIB (Sanfilippo B), 252920
NAGS	100%	100%	100%	100%	99.2%	N-acetylglutamate synthase deficiency, 237310
NALCN	100%	100%	100%	100%	99.8%	Congenital contractures of the limbs and face, hypotonia, and developmental delay, 616266;Hypotonia, infantile, with psychomotor retardation and characteristic facies 1, 615419
NANS	94.8%	94.3%	100%	99.9%	99.7%	Spondyloepimetaphyseal dysplasia, Genevieve type, 610442
NAPB	100%	100%	100%	100%	99.7%	Developmental and epileptic encephalopathy 107, 620033

NARS1	100%	100%	100%	100%	99.5%	Neurodevelopmental disorder with microcephaly, impaired language, epilepsy, and gait abnormalities, autosomal dominant, 619092;Neurodevelopmental disorder with microcephaly, impaired language, and gait abnormalities, autosomal recessive, 619091
NARS2	92.3%	92.3%	100%	100%	99.7%	Combined oxidative phosphorylation deficiency 24, 616239;?Deafness, autosomal recessive 94, 618434
NAT8L	100%	98%	100%	99.5%	95.3%	?N-acetylaspartate deficiency, 614063
NAV3	100%	100%	100%	100%	99.7%	Neurodevelopmental disorder with poor or absent speech, dysmorphic facies, and behavioral abnormalities, 621182
NAXD	95.5%	89.7%	100%	100%	99.2%	Encephalopathy, progressive, early-onset, with brain edema and/or leukoencephalopathy, 2, 618321
NAXE	97.6%	93.5%	100%	100%	99.5%	Encephalopathy, progressive, early-onset, with brain edema and/or leukoencephalopathy, 617186
NBAS	100%	100%	100%	100%	99.7%	Short stature, optic nerve atrophy, and Pelger-Huet anomaly, 614800;Infantile liver failure syndrome 2, 616483
NBEAL2	100%	100%	100%	100%	99.2%	Gray platelet syndrome, 139090

NBN	97.5%	97.5%	100%	100%	99.8%	Leukemia, acute lymphoblastic, 613065;Aplastic anemia, 609135;Nijmegen breakage syndrome, 251260
NCAPD2	97.1%	97.1%	100%	100%	99.7%	Microcephaly 21, primary, autosomal recessive, 617983
NCAPD3	100%	100%	100%	100%	99.7%	Microcephaly 22, primary, autosomal recessive, 617984
NCAPG2	100%	100%	100%	100%	99.8%	Khan-Khan-Katsanis syndrome, 618460
NCAPH	100%	100%	100%	99.9%	99.4%	?Microcephaly 23, primary, autosomal recessive, 617985
NCF1	99.7%	96.1%	100%	99.7%	97.8%	Chronic granulomatous disease 1, autosomal recessive, 233700
NCF2	89.5%	89.5%	100%	100%	99.5%	Chronic granulomatous disease 2, autosomal recessive, 233710
NCF4	100%	100%	100%	100%	99.3%	Chronic granulomatous disease 3, autosomal recessive, 613960
NCKAP1L	100%	100%	100%	100%	99.7%	Immunodeficiency 72 with autoinflammation, 618982
NDC1	100%	100%	100%	100%	99.7%	Neurodevelopmental disorder with achalasia, polyneuropathy, and alacrima, 621328
NDE1	93.5%	93.5%	100%	100%	99.1%	Microhydranencephaly, 605013;Lissencephaly 4 (with microcephaly), 614019
NDRG1	100%	100%	100%	100%	99.7%	Charcot-Marie-Tooth disease, type 4D, 601455

NDST1	100%	100%	100%	100%	99.2%	Intellectual developmental disorder, autosomal recessive 46, 616116
NDUFA10	82.9%	80.5%	100%	100%	99.8%	Mitochondrial complex I deficiency, nuclear type 22, 618243
NDUFA11	96.1%	95.9%	100%	100%	99.2%	Mitochondrial complex I deficiency, nuclear type 14, 618236
NDUFA12	80.4%	80.4%	100%	99.9%	99.7%	Mitochondrial complex I deficiency, nuclear type 23, 618244
NDUFA13	99.1%	94.7%	100%	100%	99.5%	{Thyroid carcinoma, Hurthle cell}, 607464;Mitochondrial complex I deficiency, nuclear type 28, 618249
NDUFA2	100%	100%	100%	100%	99.7%	Mitochondrial complex I deficiency, nuclear type 13, 618235
NDUFA6	100%	100%	100%	100%	99.6%	Mitochondrial complex I deficiency, nuclear type 33, 618253
NDUFA8	100%	100%	100%	100%	99.6%	Mitochondrial complex I deficiency, nuclear type 37, 619272
NDUFA9	87%	85.2%	100%	100%	99.9%	Mitochondrial complex I deficiency, nuclear type 26, 618247
NDUFAF1	91.6%	91.5%	100%	100%	99.2%	Mitochondrial complex I deficiency, nuclear type 11, 618234
NDUFAF2	67.4%	67.4%	100%	100%	99.6%	Mitochondrial complex I deficiency, nuclear type 10, 618233
NDUFAF3	100%	100%	100%	100%	99.9%	Mitochondrial complex I deficiency, nuclear type 18, 618240
NDUFAF4	100%	100%	100%	100%	99.8%	Mitochondrial complex I deficiency, nuclear type 15, 618237

NDUFAF5	90.9%	90.9%	100%	100%	99.8%	Mitochondrial complex I deficiency, nuclear type 16, 618238
NDUFAF6	100%	100%	100%	100%	99.7%	Mitochondrial complex I deficiency, nuclear type 17, 618239;Fanconi renotubular syndrome 5, 618913
NDUFAF8	100%	100%	100%	100%	99.4%	Mitochondrial complex I deficiency, nuclear type 34, 618776
NDUFB11	99.9%	99.3%	93.6%	79.3%	59.7%	Linear skin defects with multiple congenital anomalies 3, 300952;?Mitochondrial complex I deficiency, nuclear type 30, 301021
NDUFB3	100%	100%	100%	100%	99.7%	Mitochondrial complex I deficiency, nuclear type 25, 618246
NDUFB8	100%	100%	100%	99.7%	98.9%	Mitochondrial complex I deficiency, nuclear type 32, 618252
NDUFB9	86.8%	86.6%	100%	100%	99.5%	?Mitochondrial complex I deficiency, nuclear type 24, 618245
NDUFC2	100%	100%	100%	100%	99.7%	Mitochondrial complex I deficiency, nuclear type 36, 619170
NDUFS1	100%	100%	100%	100%	99.8%	Mitochondrial complex I deficiency, nuclear type 5, 618226
NDUFS2	99.9%	98.3%	100%	100%	99.6%	?Leber-like hereditary optic neuropathy, autosomal recessive 2, 620569;Mitochondrial complex I deficiency, nuclear type 6, 618228
NDUFS3	98%	92.5%	100%	100%	99.5%	Mitochondrial complex I deficiency, nuclear type 8, 618230

NDUFS4	100%	100%	100%	100%	99.8%	Mitochondrial complex I deficiency, nuclear type 1, 252010
NDUFS6	94.7%	86.1%	100%	100%	99.7%	Mitochondrial complex I deficiency, nuclear type 9, 618232
NDUFS7	89.1%	85%	100%	100%	99.4%	Mitochondrial complex I deficiency, nuclear type 3, 618224
NDUFS8	100%	100%	100%	100%	99.3%	Mitochondrial complex I deficiency, nuclear type 2, 618222
NDUFV1	100%	100%	100%	100%	98.7%	Mitochondrial complex I deficiency, nuclear type 4, 618225
NDUFV2	100%	100%	100%	100%	99.5%	Mitochondrial complex I deficiency, nuclear type 7, 618229
NEB	99.9%	99.8%	99.6%	99.2%	98.3%	Nemaline myopathy 2, autosomal recessive, 256030;Arthrogryposis multiplex congenita 6, 619334
NECAP1	100%	100%	100%	100%	99.9%	Developmental and epileptic encephalopathy 21, 615833
NECTIN1	93.4%	93.4%	100%	99.9%	99.1%	Cleft lip/palate-ectodermal dysplasia syndrome, 225060;Orofacial cleft 7, 225060
NECTIN4	100%	100%	100%	100%	99.4%	Ectodermal dysplasia-syndactyly syndrome 1, 613573
NEK1	100%	100%	100%	100%	99.8%	Short-rib thoracic dysplasia 6 with or without polydactyly, 263520;?Orofaciodigital syndrome II, 252100;{Amyotrophic lateral sclerosis, susceptibility to, 24}, 617892

NEK10	100%	100%	100%	100%	99.9%	Ciliary dyskinesia, primary, 44, 618781
NEK2	95.9%	95.9%	100%	100%	100%	?Retinitis pigmentosa 67, 615565
NEK8	100%	100%	100%	100%	99.8%	Renal-hepatic-pancreatic dysplasia 2, 615415;Polycystic kidney disease 8, 620903;?Nephronophthisis 9, 613824
NEK9	100%	100%	100%	100%	99.7%	?Arthrogryposis, Perthes disease, and upward gaze palsy, 614262;Nevus comedonicus, somatic, 617025;Lethal congenital contracture syndrome 10, 617022
NEMF	100%	100%	100%	99.8%	99.3%	Intellectual developmental disorder with speech delay and axonal peripheral neuropathy, 619099
NEPRO	100%	100%	100%	100%	99.5%	
NEU1	100%	100%	100%	99.9%	99.5%	Sialidosis, type II, 256550;Sialidosis, type I, 256550
NEUROG1	100%	100%	100%	99.6%	97%	Cranial dysinnervation disorder, congenital, with absent corneal reflex and developmental delay, 620469
NEUROG3	100%	100%	100%	99.8%	98.6%	Diarrhea 4, malabsorptive, congenital, 610370
NEXN	100%	100%	100%	100%	99.7%	Cardiomyopathy, dilated, 2M, autosomal recessive, 621261;Cardiomyopathy, dilated, 1CC, 613122;Cardiomyopathy, hypertrophic, 20, 613876

NFASC	100%	100%	100%	99.9%	99.5%	Neurodevelopmental disorder with central and peripheral motor dysfunction, 618356
NFS1	83.4%	83.4%	100%	100%	99.5%	Combined oxidative phosphorylation deficiency 52, 619386
NFU1	98.9%	94.6%	100%	100%	99.7%	Spastic paraplegia 93, autosomal recessive, 620938;Multiple mitochondrial dysfunctions syndrome 1, 605711
NGF	100%	100%	100%	100%	98.9%	Neuropathy, hereditary sensory and autonomic, type V, 608654
NGLY1	98.4%	97.2%	100%	100%	99.8%	Congenital disorder of deglycosylation 1, 615273
NHEJ1	92.9%	92.9%	100%	100%	99.7%	Microphthalmia/colobo ma 13, 620968;Immunodeficie ncy 124, severe combined, 611291
NHLRC1	100%	100%	100%	99.9%	99.1%	Myoclonic epilepsy of Lafora 2, 620681
NHLRC2	100%	100%	100%	100%	99.8%	FINCA syndrome, 618278
NHP2	100%	100%	100%	100%	99.6%	Dyskeratosis congenita, autosomal recessive 2, 613987
NIN	100%	100%	100%	100%	99.7%	?Seckel syndrome 7, 614851
NIPAL4	100%	100%	100%	99.7%	98.5%	Ichthyosis, congenital, autosomal recessive 6, 612281
NIT1	100%	100%	100%	100%	99.8%	Brain small vessel disease 4, 621313

NKX2-6	100%	100%	100%	100%	99.5%	Persistent truncus arteriosus, 217095;Conotruncal heart malformations, 217095
NKX3-2	100%	100%	100%	100%	99%	Spondylo-megaepiphyseal-metaphyseal dysplasia, 613330
NKX6-2	100%	100%	100%	99.6%	97.1%	Spastic ataxia 8, autosomal recessive, with hypomyelinating leukodystrophy, 617560
NLRP1	98.1%	98.1%	100%	100%	99.7%	{Vitiligo-associated multiple autoimmune disease susceptibility 1}, 606579;?Respiratory papillomatosis, juvenile recurrent, congenital, 618803;Autoinflammation with arthritis and dyskeratosis, 617388;Palmoplantar carcinoma, multiple self-healing, 615225
NLRP7	100%	100%	100%	100%	99.3%	?Ooxyte/zygote/embryo maturation arrest 25, 621471;Hydatidiform mole, recurrent, 1, 231090
NME5	86.6%	86.6%	100%	100%	100%	Ciliary dyskinesia, primary, 48, without situs inversus, 620032
NME8	100%	100%	100%	100%	99.8%	?Ciliary dyskinesia, primary, 6, 610852
NMNAT1	100%	99.5%	100%	100%	99.4%	Spondyloepiphyseal dysplasia, sensorineural hearing loss, intellectual developmental disorder, and Leber congenital amaurosis, 619260;Leber congenital amaurosis 9, 608553

NNT	96.3%	96.3%	100%	100%	99.8%	Glucocorticoid deficiency 4, with or without mineralocorticoid deficiency, 614736
NOP10	92.5%	92.5%	100%	100%	99.9%	?Pulmonary fibrosis and/or bone marrow failure syndrome, telomere-related, 9, 620400;?Cataracts, hearing impairment, nephrotic syndrome, and enterocolitis 2, 620425;?Dyskeratosis congenita, autosomal recessive 1, 224230
NOS1AP	100%	100%	100%	100%	99.6%	Nephrotic syndrome, type 22, 619155
NOTCH3	99.9%	99.4%	100%	100%	99.4%	Lateral meningocele syndrome, 130720;Lipodystrophy, familial partial, type 1, 608600;Cerebral arteriopathy, autosomal recessive, with subcortical infarcts and leukoencephalopathy 1, 621295;?Myofibromatosis, infantile 2, 615293;Cerebral arteriopathy, autosomal dominant, with subcortical infarcts and leukoencephalopathy 1, 125310
NPC1	100%	100%	100%	100%	99.7%	Niemann-Pick disease, type C1, 257220;Niemann-Pick disease, type D, 257220
NPC2	99%	95.2%	100%	100%	99.6%	Niemann-pick disease, type C2, 607625
NPHP1	100%	100%	100%	100%	99.8%	Joubert syndrome 4, 609583;Nephronophthisis 1, juvenile, 256100;Senior-Loken syndrome-1, 266900

NPHP3	100%	100%	100%	100%	99.6%	Nephronophthisis 3, 604387;Renal-hepatic-pancreatic dysplasia 1, 208540;Meckel syndrome 7, 267010
NPHP4	100%	100%	100%	100%	99.5%	Senior-Loken syndrome 4, 606996;Nephronophthisis 4, 606966
NPHS1	100%	100%	100%	100%	99.3%	Nephrotic syndrome, type 1, 256300
NPHS2	100%	100%	100%	100%	99.3%	Nephrotic syndrome, type 2, 600995
NPPA	100%	100%	100%	100%	99.7%	Atrial standstill 2, 615745;Atrial fibrillation, familial, 6, 612201
NPR2	100%	100%	100%	100%	99.5%	Epiphyseal chondrodysplasia, Miura type, 615923;Short stature with nonspecific skeletal abnormalities, 616255;Acromesomelic dysplasia 1, Maroteaux type, 602875
NPR3	100%	100%	100%	100%	99.1%	Boudin-Mortier syndrome, 619543
NR0B2	100%	100%	100%	100%	99.5%	Obesity, mild, early-onset, 601665
NR1H4	100%	100%	100%	100%	99.7%	Cholestasis, progressive familial intrahepatic, 5, 617049
NR2E3	100%	100%	100%	100%	99.5%	Retinitis pigmentosa 37, 611131;Enhanced S-cone syndrome 1, 268100
NRAP	100%	100%	100%	100%	99.7%	
NRCAM	100%	100%	100%	100%	99.9%	Neurodevelopmental disorder with neuromuscular and skeletal abnormalities, 619833

NRL	100%	100%	100%	100%	99.4%	Enhanced S-cone syndrome 2, 621371;Retinitis pigmentosa 27, 613750
NRROS	100%	100%	100%	99.9%	99.2%	Seizures, early-onset, with neurodegeneration and brain calcification, 618875
NRXN1	100%	100%	100%	100%	99.5%	Pitt-Hopkins-like syndrome 2, 614325;{Schizophrenia, susceptibility to, 17}, 621407
NSMCE2	92.1%	92.1%	100%	100%	99.9%	Seckel syndrome 10, 617253
NSMCE3	100%	100%	100%	99.9%	98.4%	Lung disease, immunodeficiency, and chromosome breakage syndrome, 617241
NSRP1	91%	91%	100%	100%	99.5%	Neurodevelopmental disorder with spasticity, seizures, and brain abnormalities, 620001
NSUN2	98.5%	98.5%	100%	100%	99.5%	Intellectual developmental disorder, autosomal recessive 5, 611091
NSUN3	100%	100%	100%	100%	99.9%	Combined oxidative phosphorylation deficiency 48, 619012
NSUN6	100%	100%	100%	100%	99.9%	Intellectual developmental disorder, autosomal recessive 82, 620779
NT5C2	91.4%	91.4%	100%	100%	99.8%	Spastic paraplegia 45, autosomal recessive, 613162
NT5C3A	100%	100%	100%	100%	99.9%	Anemia, congenital, nonspherocytic hemolytic, 8, 266120
NT5E	91%	91%	100%	100%	99.5%	Calcification of joints and arteries, 211800

NTHL1	100%	100%	100%	99.9%	99.1%	Familial adenomatous polyposis 3, 616415
NTNG2	100%	100%	100%	100%	99.3%	Neurodevelopmental disorder with behavioral abnormalities, absent speech, and hypotonia, 618718
NTRK1	100%	100%	100%	99.9%	99.3%	Insensitivity to pain, congenital, with anhidrosis, 256800
NUBPL	89.8%	89.8%	100%	100%	99.9%	Mitochondrial complex I deficiency, nuclear type 21, 618242
NUDT2	100%	100%	100%	100%	99.8%	Intellectual developmental disorder with or without peripheral neuropathy, 619844
NUP107	97.8%	97.8%	100%	100%	99.9%	?Ovarian dysgenesis 6, 618078;Galloway-Mowat syndrome 7, 618348;Nephrotic syndrome, type 11, 616730
NUP133	97.9%	97.9%	100%	100%	99.8%	?Galloway-Mowat syndrome 8, 618349;Nephrotic syndrome, type 18, 618177
NUP160	94.6%	94.6%	100%	100%	99.8%	?Nephrotic syndrome, type 19, 618178
NUP188	95.4%	95.4%	100%	100%	99.6%	Sandestig-Stefanova syndrome, 618804
NUP205	97%	97%	100%	100%	99.6%	?Nephrotic syndrome, type 13, 616893
NUP214	100%	100%	100%	100%	99.5%	Leukemia, T-cell acute lymphoblastic, somatic, 613065;Leukemia, acute myeloid, somatic, 601626;{Encephalopathy, acute, infection-induced, susceptibility to, 9}, 618426

NUP37	100%	100%	100%	100%	100%	?Microcephaly 24, primary, autosomal recessive, 618179
NUP54	100%	100%	100%	100%	99.7%	Dystonia 37, early-onset, with striatal lesions, 620427
NUP62	100%	100%	100%	100%	99.3%	Striatonigral degeneration, infantile, 271930
NUP85	100%	99.8%	100%	100%	99.4%	Nephrotic syndrome, type 17, 618176
NUP88	93.5%	93.5%	100%	100%	99.7%	Fetal akinesia deformation sequence 4, 618393
NUP93	95.5%	95.5%	100%	100%	99.7%	Nephrotic syndrome, type 12, 616892
NUS1	93.9%	93.9%	100%	100%	99.4%	Intellectual developmental disorder, autosomal dominant 55, with seizures, 617831;?Congenital disorder of glycosylation, type 1aa, 617082
NXN	100%	100%	100%	99.9%	99%	Robinow syndrome, autosomal recessive 2, 618529
OAT	100%	100%	100%	99.9%	99.3%	Gyrate atrophy of choroid and retina with or without ornithinemia, 258870
OBSL1	100%	100%	100%	99.8%	99.2%	3-M syndrome 2, 612921
OCA2	93.4%	93.4%	100%	100%	99.7%	[Skin/hair/eye pigmentation 1, blue/nonblue eyes], 227220;[Skin/hair/eye pigmentation 1, blond/brown hair], 227220;Albinism, brown oculocutaneous, 203200;Albinism, oculocutaneous, type II, 203200

OCLN	94.5%	94.5%	100%	99.9%	99.4%	Pseudo-TORCH syndrome 1, 251290
ODAD1	100%	100%	100%	99.9%	99.4%	Ciliary dyskinesia, primary, 20, 615067
ODAD2	96.1%	96.1%	100%	100%	99.7%	Ciliary dyskinesia, primary, 23, 615451
ODAD3	100%	100%	100%	99.9%	98.7%	Ciliary dyskinesia, primary, 30, 616037
ODAD4	100%	100%	100%	99.9%	99.2%	Ciliary dyskinesia, primary, 35, 617092
ODAPH	100%	100%	100%	100%	99.8%	Amelogenesis imperfecta, type IIA4, 614832
OGDH	100%	100%	100%	100%	99.5%	Oxoglutarate dehydrogenase deficiency, 203740
OGDHL	100%	100%	100%	100%	99.5%	
OPA1	94.7%	94.7%	100%	100%	99.7%	Optic atrophy plus syndrome, 125250;{Glaucoma, normal tension, susceptibility to}, 606657;Optic atrophy 1, 165500;Behr syndrome, 210000;Mitochondrial DNA depletion syndrome 14A (encephalomyopathic type), 621481;?Mitochondrial DNA depletion syndrome 14B (cardioencephalomyopathic type), 616896
OPA3	100%	100%	100%	100%	99.6%	3-methylglutaconic aciduria, type III, 258501;Optic atrophy 3 with cataract, 165300
OPLAH	100%	100%	100%	100%	99.3%	5-oxoprolinase deficiency, 260005

ORAI1	100%	100%	100%	99.8%	98.6%	Immunodeficiency 9, 612782;Myopathy, tubular aggregate, 2, 615883
ORC1	100%	100%	100%	100%	99.8%	Meier-Gorlin syndrome 1, 224690
ORC4	90.8%	90.8%	100%	100%	99.7%	Meier-Gorlin syndrome 2, 613800
ORC6	100%	100%	100%	100%	99.7%	Meier-Gorlin syndrome 3, 613803
OSGEP	100%	100%	100%	100%	99.8%	Galloway-Mowat syndrome 3, 617729
OSTM1	100%	99.7%	100%	100%	99.5%	Osteopetrosis, autosomal recessive 5, 259720
OTOA	100%	100%	100%	99.9%	99.3%	Deafness, autosomal recessive 22, 607039
OTOF	100%	100%	100%	100%	99.3%	Auditory neuropathy, autosomal recessive, 1, 601071;Deafness, autosomal recessive 9, 601071
OTOG	100%	100%	100%	99.9%	99.3%	Deafness, autosomal recessive 18B, 614945
OTOGL	100%	100%	100%	100%	99.7%	Deafness, autosomal recessive 84B, 614944
OTUD6B	100%	100%	100%	100%	99.7%	Intellectual developmental disorder with dysmorphic facies, seizures, and distal limb anomalies, 617452
OTUD7A	99.9%	99.1%	100%	99.9%	97.8%	Neurodevelopmental disorder with hypotonia and seizures, 620790

OTULIN	100%	100%	100%	100%	99.4%	Autoinflammation, panniculitis, and dermatosis syndrome, autosomal recessive, 617099;{Immunodeficiency 107, susceptibility to invasive staphylococcus aureus infection}, 619986;Autoinflammation, panniculitis, and dermatosis syndrome, autosomal dominant, 621030
OXCT1	90.5%	90.5%	100%	100%	99.7%	Succinyl CoA:3-oxoacid CoA transferase deficiency, 245050
OXR1	100%	100%	100%	100%	99.8%	Cerebellar hypoplasia/atrophy, epilepsy, and global developmental delay, 213000
P2RY12	100%	100%	100%	100%	99.8%	Bleeding disorder, platelet-type, 8, 609821
P3H1	95.3%	93.2%	100%	99.9%	99.1%	Osteogenesis imperfecta, type VIII, 610915
P3H2	100%	100%	100%	99.8%	99.2%	Myopia, high, with cataract and vitreoretinal degeneration, 614292
P4HTM	89.4%	89.4%	100%	99.9%	99.2%	Hypotonia, hypoventilation, impaired intellectual development, dysautonomia, epilepsy, and eye abnormalities, 618493
PACSIN3	100%	100%	100%	100%	99.7%	Congenital myopathy 27, 621343
PAH	94.9%	93.7%	100%	100%	99.8%	[Hyperphenylalaninemia, non-PKU mild], 261600;Phenylketonuria, 261600

PAM16	85.8%	85.5%	100%	100%	99.3%	Spondylometaphyseal dysplasia, Megarbane-Dagher-Me like type, 613320
PAN2	100%	100%	100%	100%	99.7%	Developmental delay with variable cardiac and renal congenital anomalies and dysmorphic facies, 621384
PANK2	100%	100%	100%	100%	99.6%	Neurodegeneration with brain iron accumulation 1, 234200
PAPPA2	100%	100%	100%	100%	99.7%	Short stature, Dauber-Argente type, 619489
PAPSS2	100%	100%	100%	100%	99.6%	Brachyolmia 4 with mild epiphyseal and metaphyseal changes, 612847
PARK7	97.5%	93.1%	100%	99.8%	99.4%	Parkinson disease 7, autosomal recessive early-onset, 606324
PARN	97.2%	96%	100%	100%	99.8%	Dyskeratosis congenita, autosomal recessive 6, 616353;Pulmonary fibrosis and/or bone marrow failure syndrome, telomere-related, 4, 616371
PARS2	100%	100%	100%	100%	99.7%	Developmental and epileptic encephalopathy 75, 618437
PATL2	100%	100%	100%	100%	99.8%	Oocyte/zygote/embryo maturation arrest 4, 617743
PAX1	100%	100%	100%	100%	98.6%	Otofaciocervical syndrome 2 with T-cell deficiency, 615560

PAX3	100%	100%	100%	100%	99.6%	Craniofacial-deafness-hand syndrome, 122880;Waardenburg syndrome, type 3, 148820;Waardenburg syndrome, type 1, 193500;Rhabdomyosarcoma 2, alveolar, 268220
PAX7	94.2%	94.2%	100%	99.9%	99.1%	Congenital myopathy 19, 618578;Rhabdomyosarcoma 2, alveolar, 268220
PC	99.9%	99.2%	100%	100%	99.5%	Pyruvate carboxylase deficiency, 266150
PCARE	100%	100%	100%	100%	99.6%	Retinitis pigmentosa 54, 613428
PCBD1	100%	100%	100%	100%	99.5%	Hyperphenylalaninemia, BH4-deficient, D, 264070
PCCA	84.6%	84.4%	100%	100%	99.7%	Propionicacidemia, 606054
PCCB	86.8%	85.3%	100%	100%	99.8%	Propionicacidemia, 606054
PCDH12	100%	100%	100%	100%	99.5%	Diencephalic-mesencephalic junction dysplasia syndrome 1, 251280
PCDH15	100%	100%	100%	100%	99.6%	Usher syndrome, type 1D/F digenic, 601067;Deafness, autosomal recessive 23, 609533;Usher syndrome, type 1F, 602083
PCDHGC4	100%	100%	100%	100%	99.6%	Neurodevelopmental disorder with poor growth and skeletal anomalies, 619880
PCK1	100%	100%	100%	100%	99.7%	Phosphoenolpyruvate carboxykinase deficiency, cytosolic, 261680

PCK2	100%	100%	100%	100%	99.5%	PEPCK deficiency, mitochondrial, 261650
PCLO	100%	100%	100%	100%	99.5%	Pontocerebellar hypoplasia, type 3, 608027
PCNT	100%	100%	100%	100%	99.6%	Microcephalic osteodysplastic primordial dwarfism, type II, 210720
PCSK1	100%	100%	100%	100%	99.9%	{Obesity, susceptibility to, BMIQ12}, 612362;Endocrinopathy due to proprotein convertase 1/3 deficiency, 600955
PCYT1A	100%	100%	100%	100%	99.7%	Spondylometaphyseal dysplasia with cone-rod dystrophy, 608940;Lipodystrophy, congenital generalized, type 5, 620680
PCYT2	99.9%	97.1%	100%	100%	99.5%	Spastic paraplegia 82, autosomal recessive, 618770
PDE10A	100%	99.4%	99.9%	99.7%	98.3%	Striatal degeneration, autosomal dominant, 616922;Dyskinesia, limb and orofacial, infantile-onset, 616921
PDE2A	100%	100%	100%	100%	99.3%	Intellectual developmental disorder with paroxysmal dyskinesia or seizures, 619150
PDE6A	100%	100%	100%	100%	99.6%	Retinitis pigmentosa 43, 613810
PDE6B	100%	100%	100%	100%	99.2%	Retinitis pigmentosa-40, 613801;Night blindness, congenital stationary, autosomal dominant 2, 163500
PDE6C	100%	100%	100%	100%	99.9%	Cone dystrophy 4, 613093

PDE6D	100%	100%	100%	100%	99.1%	Joubert syndrome 22, 615665
PDE6G	100%	100%	100%	100%	99.2%	Retinitis pigmentosa 57, 613582
PDE6H	100%	100%	100%	100%	99.9%	Achromatopsia 6, 610024
PDHB	100%	100%	100%	100%	99.8%	Pyruvate dehydrogenase E1-beta deficiency, 614111
PDHX	91.6%	91.6%	100%	100%	99.6%	Lacticacidemia due to PDX1 deficiency, 245349
PDIA6	100%	100%	100%	100%	99.5%	
PDP1	100%	100%	100%	100%	99.9%	Pyruvate dehydrogenase phosphatase deficiency, 608782
PDSS1	100%	100%	100%	100%	99.7%	Coenzyme Q10 deficiency, primary, 2, 614651
PDSS2	100%	100%	100%	100%	99.4%	Coenzyme Q10 deficiency, primary, 3, 614652
PDX1	100%	100%	100%	100%	99.5%	{Diabetes mellitus, type II, susceptibility to}, 125853;Pancreatic agenesis 1, 260370;MODY, type IV, 606392
PDXK	91.6%	88.3%	100%	100%	99.3%	Neuropathy, hereditary motor and sensory, type VIC, with optic atrophy, 618511
PDZD7	100%	99.8%	100%	100%	99.3%	Deafness, autosomal recessive 57, 618003;{Retinal disease in Usher syndrome type IIA, modifier of}, 276901;Usher syndrome, type IIC, GPR98/PDZD7 digenic, 605472

PDZD8	100%	100%	100%	99.8%	98.5%	Intellectual developmental disorder with autism and dysmorphic facies, 620021
PEPD	94%	94%	100%	100%	99.6%	Prolidase deficiency, 170100
PERCC1	100%	100%	100%	100%	99.8%	Diarrhea 11, malabsorptive, congenital, 618662
PERP	100%	100%	100%	100%	99.4%	Erythrokeratodermia variabilis et progressiva 7, 619209;Olmsted syndrome 2, 619208
PET100	100%	100%	100%	100%	99.2%	Mitochondrial complex IV deficiency, nuclear type 12, 619055
PEX1	100%	100%	100%	100%	99.9%	Heimler syndrome 1, 234580;Peroxisome biogenesis disorder 1B (NALD/IRD), 601539;Peroxisome biogenesis disorder 1A (Zellweger), 214100
PEX10	100%	100%	100%	99.6%	98.7%	Peroxisome biogenesis disorder 6A (Zellweger), 614870;Peroxisome biogenesis disorder 6B, 614871
PEX11B	100%	100%	100%	99.9%	99.1%	Peroxisome biogenesis disorder 14B, 614920
PEX12	100%	100%	100%	100%	99.5%	Peroxisome biogenesis disorder 3B, 266510;Peroxisome biogenesis disorder 3A (Zellweger), 614859
PEX13	92.4%	92.4%	100%	100%	99.9%	Peroxisome biogenesis disorder 11A (Zellweger), 614883;Peroxisome biogenesis disorder 11B, 614885

PEX14	100%	100%	100%	100%	99.2%	Peroxisome biogenesis disorder 13A (Zellweger), 614887
PEX16	100%	100%	100%	100%	99.6%	Peroxisome biogenesis disorder 8B, 614877;Peroxisome biogenesis disorder 8A (Zellweger), 614876
PEX19	88.6%	88.6%	100%	100%	99.9%	Peroxisome biogenesis disorder 12A (Zellweger), 614886
PEX2	100%	100%	100%	100%	99.7%	Peroxisome biogenesis disorder 5A (Zellweger), 614866;Peroxisome biogenesis disorder 5B, 614867
PEX26	91.9%	91.7%	100%	99.9%	99.1%	Peroxisome biogenesis disorder 7B, 614873;Peroxisome biogenesis disorder 7A (Zellweger), 614872
PEX3	100%	100%	100%	100%	99.9%	Peroxisome biogenesis disorder 10A (Zellweger), 614882;?Peroxisome biogenesis disorder 10B, 617370
PEX5	100%	100%	100%	99.8%	99%	Peroxisome biogenesis disorder 2B, 202370;Peroxisome biogenesis disorder 2A (Zellweger), 214110;Rhizomelic chondrodysplasia punctata, type 5, 616716
PEX6	100%	100%	100%	100%	99.1%	Peroxisome biogenesis disorder 4B, 614863;Peroxisome biogenesis disorder 4A (Zellweger), 614862;Heimler syndrome 2, 616617

PEX7	97.9%	97.9%	100%	100%	99.6%	Rhizomelic chondrodysplasia punctata, type 1, 215100;Peroxisome biogenesis disorder 9B, 614879
PFKM	100%	100%	100%	100%	99.7%	Glycogen storage disease VII, 232800
PGAM2	100%	100%	100%	99.8%	99.4%	Glycogen storage disease X, 261670
PGAP1	96.7%	96.7%	100%	100%	99.9%	Neurodevelopmental disorder with dysmorphic features, spasticity, and brain abnormalities, 615802
PGAP2	100%	100%	100%	99.9%	98.9%	Hyperphosphatasia with impaired intellectual development syndrome 3, 614207
PGAP3	100%	100%	100%	100%	99.4%	Hyperphosphatasia with impaired intellectual development syndrome 4, 615716
PGBD5	100%	100%	100%	99.9%	99.4%	Neurodevelopmental disorder with seizures, hypotonia, and variable spasticity, 621482
PGM1	87.5%	87.5%	100%	100%	99.4%	Congenital disorder of glycosylation, type It, 614921
PGM2L1	100%	100%	100%	100%	99.9%	Neurodevelopmental disorder with hypotonia, dysmorphic facies, and skin abnormalities, 620191
PGM3	100%	100%	100%	100%	99.8%	Immunodeficiency 23, 615816
PHGDH	100%	100%	100%	100%	99.6%	Neu-Laxova syndrome 1, 256520;Phosphoglycerate dehydrogenase deficiency, 601815

PHKB	100%	100%	100%	100%	99.7%	Phosphorylase kinase deficiency of liver and muscle, autosomal recessive, 261750
PHKG2	100%	100%	100%	100%	99.1%	Glycogen storage disease IXc, 613027
PHLDB1	97.7%	97.5%	100%	99.9%	99.1%	Osteogenesis imperfecta, type XXIII, 620639
PHOX2A	100%	100%	100%	99.9%	99.2%	Fibrosis of extraocular muscles, congenital, 2, 602078
PHYH	100%	100%	100%	99.8%	99.1%	Refsum disease, 266500
PI4K2A	86.9%	86.9%	100%	100%	99.1%	Neurodevelopmental disorder with hyperkinetic movements, seizures and structural brain abnormalities, 620732
PI4KA	99.3%	99.2%	100%	99.9%	99%	Spastic paraplegia 84, autosomal recessive, 619621;Gastrointestinal defects and immunodeficiency syndrome 2, 619708;Polymicrogyria, perisylvian, with cerebellar hypoplasia and arthrogryposis, 616531
PIBF1	100%	100%	100%	100%	99.8%	Joubert syndrome 33, 617767
PIDD1	100%	100%	100%	100%	99.6%	Intellectual developmental disorder, autosomal recessive 75, with neuropsychiatric features and variant lissencephaly, 619827

PIEZO1	100%	100%	100%	100%	99.2%	[ER blood group system], 620207;Lymphatic malformation 6, 616843;Dehydrated hereditary stomatocytosis with or without pseudohyperkalemia and/or perinatal edema, 194380
PIEZO2	100%	100%	100%	100%	99.7%	Arthrogryposis, distal, type 5, 108145;Arthrogryposis, distal, with impaired proprioception and touch, 617146;Arthrogryposis, distal, type 3, 114300;?Marden-Walker syndrome, 248700
PIGB	100%	100%	100%	100%	99.8%	Developmental and epileptic encephalopathy 80, 618580
PIGC	100%	100%	100%	100%	99.8%	Glycosylphosphatidylinositol biosynthesis defect 16, 617816
PIGG	100%	100%	100%	100%	99.5%	[Blood group, EMM system], 619812;Neurodevelopmental disorder with or without hypotonia, seizures, and cerebellar atrophy, 616917
PIGH	82.9%	76.2%	100%	100%	99.8%	Glycosylphosphatidylinositol biosynthesis defect 17, 618010
PIGK	88.2%	88.2%	100%	100%	99.9%	Neurodevelopmental disorder with hypotonia and cerebellar atrophy, with or without seizures, 618879
PIGL	100%	100%	100%	100%	99.7%	CHIME syndrome, 280000

PIGM	100%	100%	100%	100%	99.8%	Glycosylphosphatidylinositol deficiency, 610293
PIGN	95.9%	94.4%	100%	100%	99.7%	Multiple congenital anomalies-hypotonia-seizures syndrome 1, 614080
PIGO	100%	100%	100%	100%	99.8%	Hyperphosphatasia with impaired intellectual development syndrome 2, 614749
PIGP	100%	100%	100%	99.9%	99.4%	Developmental and epileptic encephalopathy 55, 617599
PIGQ	100%	100%	100%	100%	99.6%	Multiple congenital anomalies-hypotonia-seizures syndrome 4, 618548
PIGS	98.4%	95.1%	100%	99.9%	99.1%	Developmental and epileptic encephalopathy 95, 618143
PIGT	100%	100%	100%	100%	99.5%	?Paroxysmal nocturnal hemoglobinuria 2, 615399;Multiple congenital anomalies-hypotonia-seizures syndrome 3, 615398
PIGU	97.1%	97.1%	100%	100%	99.7%	Neurodevelopmental disorder with brain anomalies, seizures, and scoliosis, 618590
PIGV	100%	100%	100%	100%	99.7%	Hyperphosphatasia with impaired intellectual development syndrome 1, 239300
PIGW	100%	100%	100%	100%	99.9%	Glycosylphosphatidylinositol biosynthesis defect 11, 616025

PIGY	100%	100%	100%	99.8%	99.3%	Hyperphosphatasia with impaired intellectual development syndrome 6, 616809
PIK3C2A	100%	100%	100%	100%	99.9%	Oculoskeletodental syndrome, 618440
PIK3CD	100%	100%	100%	100%	99.3%	Immunodeficiency 14A, autosomal dominant, 615513;Immunodeficiency 14B, autosomal recessive, 619281;?Roifman-Chitayat syndrome, digenic, 613328
PIK3R1	100%	100%	100%	100%	99.7%	Immunodeficiency 36, 616005;SHORT syndrome, 269880;Agammaglobulinemia 7, autosomal recessive, 615214
PIK3R5	100%	100%	100%	100%	98.9%	Ataxia-oculomotor apraxia 3, 615217
PINK1	96.9%	96.9%	100%	100%	99.2%	Parkinson disease 6, early onset, 605909
PIP5K1C	90.6%	90.2%	100%	99.9%	99.4%	Lethal congenital contractural syndrome 3, 611369
PISD	100%	100%	100%	99.9%	99.5%	Liberfarb syndrome, 618889
PJVK	100%	100%	100%	100%	99.9%	Deafness, autosomal recessive 59, 610220
PKD1L1	100%	100%	100%	100%	99.7%	Heterotaxy, visceral, 8, autosomal, 617205
PKDCC	100%	100%	100%	99.9%	98.2%	Rhizomelic limb shortening with dysmorphic features, 618821
PKHD1	100%	100%	100%	100%	99.7%	Polycystic kidney disease 4, with or without hepatic disease, 263200

PKHD1L1	100%	100%	100%	100%	99.8%	Deafness, autosomal recessive 124, 620794
PKLR	100%	100%	100%	100%	99.4%	Anemia, congenital, nonspherocytic hemolytic, 2, pyruvate kinase deficient, 266200;[Adenosine triphosphate, elevated, of erythrocytes], 102900
PKP1	100%	100%	100%	100%	99.6%	Ectodermal dysplasia/skin fragility syndrome, 604536
PLA2G4A	100%	100%	100%	100%	99.8%	Gastrointestinal ulceration, recurrent, with dysfunctional platelets, 618372
PLA2G6	100%	100%	100%	100%	99.4%	Parkinson disease 14, autosomal recessive, 612953;Neurodegeneration with brain iron accumulation 2B, 610217;Infantile neuroaxonal dystrophy 1, 256600
PLA2G7	100%	100%	100%	100%	99.6%	Platelet-activating factor acetylhydrolase deficiency, 614278
PLAA	100%	100%	100%	99.9%	99.8%	Neurodevelopmental disorder with progressive microcephaly, spasticity, and brain anomalies, 617527
PLAAT3	65.6%	63.1%	100%	100%	99.7%	Lipodystrophy, familial partial, type 9, 620683
PLCB1	100%	100%	100%	100%	99.6%	Developmental and epileptic encephalopathy 12, 613722
PLCB3	100%	100%	100%	100%	99.6%	Spondylometaphyseal dysplasia with corneal dystrophy, 618961

PLCB4	99%	99%	100%	100%	99.8%	Auriculocondylar syndrome 2B, 620458;Auriculocondylar syndrome 2A, 614669
PLCD1	100%	100%	100%	100%	99.6%	Nail disorder, nonsyndromic congenital, 3, (leukonychia), 151600
PLCE1	100%	100%	100%	100%	99.8%	Nephrotic syndrome, type 3, 610725
PLCH1	100%	100%	100%	100%	99.8%	Holoprosencephaly 14, 619895
PLD1	100%	100%	100%	100%	99.8%	Cardiac valvular dysplasia 1, 212093
PLD4	100%	100%	100%	100%	99.7%	Systemic lupus erythematosus 18, 621369
PLEC	100%	100%	100%	100%	98.9%	?Epidermolysis bullosa simplex 5D, generalized intermediate, autosomal recessive, 616487;Epidermolysis bullosa simplex 5B, with muscular dystrophy, 226670;Epidermolysis bullosa simplex 5C, with pyloric atresia, 612138;Epidermolysis bullosa simplex 5A, Ogna type, 131950;Muscular dystrophy, limb-girdle, autosomal recessive 17, 613723
PLEKHG2	100%	100%	100%	100%	99.4%	Leukodystrophy and acquired microcephaly with or without dystonia, 616763

PLEKHG5	100%	100%	100%	99.9%	99.3%	Neuronopathy, distal hereditary motor, autosomal recessive 4, 611067;Charcot-Marie-Tooth disease, recessive intermediate C, 615376
PLEKHM1	100%	100%	100%	99.8%	98.8%	?Osteopetrosis, autosomal recessive 6, 611497;Osteopetrosis, autosomal dominant 3, 618107
PLG	89.3%	89.3%	100%	100%	99.8%	Dysplasminogenemia, 217090;Angioedema, hereditary, 4, 619360;Plasminogen deficiency, type I, 217090
PLK4	100%	100%	100%	100%	99.9%	Microcephaly and chorioretinopathy, autosomal recessive, 2, 616171
PLOD1	96.5%	96.5%	100%	100%	99.2%	Ehlers-Danlos syndrome, kyphoscoliotic type, 1, 225400
PLOD2	100%	100%	100%	99.9%	99.4%	Bruck syndrome 2, 609220
PLOD3	98.5%	96.8%	100%	99.8%	98.9%	BCARD syndrome (lysyl hydroxylase 3 deficiency), 612394
PLPBP	100%	100%	100%	99.8%	98.6%	Epilepsy, early-onset, 1, vitamin B6-dependent, 617290
PLVAP	94.1%	94.1%	100%	99.8%	98.6%	Diarrhea 10, protein-losing enteropathy type, 618183
PLXNA1	100%	100%	100%	100%	99.6%	Dworschak-Punetha neurodevelopmental syndrome, 619955
PLXND1	100%	100%	100%	99.9%	99.2%	Congenital heart defects, multiple types, 9, 620294

PMM2	94.6%	94.6%	100%	100%	99.7%	Congenital disorder of glycosylation, type Ia, 212065
PMPCA	96.2%	96.2%	100%	100%	99.1%	Spinocerebellar ataxia, autosomal recessive 2, 213200
PMPCB	91.4%	91.4%	100%	100%	99.8%	Multiple mitochondrial dysfunctions syndrome 6, 617954
PMS2	93.4%	93.4%	99.8%	99.6%	98.9%	Lynch syndrome 4, 614337;Mismatch repair cancer syndrome 4, 619101
PNKP	100%	100%	100%	99.9%	99.2%	?Charcot-Marie-Tooth disease, type 2B2, 605589;Ataxia-oculomotor apraxia 4, 616267;Microcephaly, seizures, and developmental delay, 613402
PNLIP	100%	100%	100%	100%	99.6%	?Pancreatic lipase deficiency, 614338
PNP	100%	100%	100%	100%	99.8%	Immunodeficiency due to purine nucleoside phosphorylase deficiency, 613179
PNPLA1	100%	100%	100%	99.9%	98.9%	Ichthyosis, congenital, autosomal recessive 10, 615024
PNPLA2	80.4%	80.4%	100%	100%	99.6%	Neutral lipid storage disease with myopathy, 610717
PNPLA6	100%	100%	100%	100%	99.3%	Spastic paraplegia 39, autosomal recessive, 612020;Oliver-McFarlane syndrome, 275400;?Laurence-Moon syndrome, 245800;Boucher-Neuhauser syndrome, 215470
PNPLA8	100%	100%	100%	100%	99.5%	Mitochondrial myopathy with lactic acidosis, 251950

PNPO	100%	100%	100%	100%	99.6%	Pyridoxamine 5'-phosphate oxidase deficiency, 610090
PNPT1	100%	100%	100%	100%	99.8%	Spinocerebellar ataxia 25, 608703;Deafness, autosomal recessive 70, with or without adult-onset neurodegeneration, 614934;Combined oxidative phosphorylation deficiency 13, 614932
POC1A	100%	100%	100%	99.8%	99.1%	Short stature, onychodysplasia, facial dysmorphism, and hypotrichosis, 614813
POC1B	100%	100%	100%	100%	99.7%	Cone-rod dystrophy 20, 615973
POGLUT1	99.9%	98%	100%	100%	99.8%	Dowling-Degos disease 4, 615696;Muscular dystrophy, limb-girdle, autosomal recessive 21, 617232
POLD1	100%	100%	100%	99.9%	99%	Mandibular hypoplasia, deafness, progeroid features, and lipodystrophy syndrome, 615381;Immunodeficiency 120, 620836;{Colorectal cancer, susceptibility to, 10}, 612591
POLD3	100%	100%	100%	100%	99.5%	Immunodeficiency 122, 620869
POLE	100%	100%	100%	100%	99.3%	{Colorectal cancer, susceptibility to, 12}, 615083;FILS syndrome, 615139;IMAGE-I syndrome, 618336

POLG	100%	100%	100%	100%	99.8%	Mitochondrial recessive ataxia syndrome (includes SANDO and SCAE), 607459;Mitochondrial DNA depletion syndrome 4B (MNGIE type), 613662;Mitochondrial DNA depletion syndrome 4A (Alpers type), 203700;Progressive external ophthalmoplegia, autosomal dominant 1, 157640;Progressive external ophthalmoplegia, autosomal recessive 1, 258450
POLH	100%	100%	100%	100%	99.8%	Xeroderma pigmentosum, variant type, 278750
POLR1A	100%	100%	100%	100%	99.7%	Leukodystrophy, hypomyelinating, 27, 620675;Acrofacial dysostosis, Cincinnati type, 616462
POLR1C	83.6%	83.5%	100%	100%	99.2%	Leukodystrophy, hypomyelinating, 11, 616494;Treacher Collins syndrome 3, 248390
POLR1D	100%	100%	100%	100%	99.8%	Treacher Collins syndrome 2, 613717
POLR3A	100%	100%	100%	100%	99.6%	Wiedemann-Rautenstrauch syndrome, 264090;Leukodystrophy, hypomyelinating, 7, with or without oligodontia and/or hypogonadotropic hypogonadism, 607694

POLR3B	95.4%	95.4%	100%	100%	99.7%	Leukodystrophy, hypomyelinating, 8, with or without oligodontia and/or hypogonadotropic hypogonadism, 614381;Charcot-Marie-Tooth disease, demyelinating, type 1I, 619742
POLR3GL	100%	100%	100%	100%	99.5%	Short stature, oligodontia, dysmorphic facies, and motor delay, 619234
POLR3K	100%	100%	100%	100%	99.6%	Leukodystrophy, hypomyelinating, 21, 619310
POLRMT	99.7%	98.7%	100%	100%	99%	Combined oxidative phosphorylation deficiency 55, 619743
POMC	100%	100%	100%	100%	99.7%	{Obesity, early-onset, susceptibility to}, 601665;Obesity, adrenal insufficiency, and red hair due to POMC deficiency, 609734
POMGNT1	100%	100%	100%	100%	99.6%	Muscular dystrophy-dystroglycanopathy (limb-girdle), type C, 3, 613157;Muscular dystrophy-dystroglycanopathy (congenital with impaired intellectual development), type B, 3, 613151;Retinitis pigmentosa 76, 617123;Muscular dystrophy-dystroglycanopathy (congenital with brain and eye anomalies), type A, 3, 253280

POMGNT2	100%	100%	100%	100%	99.5%	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%	99.8%	?Muscular dystrophy-dystroglycanopathy (limb-girdle), type C, 12, 616094;Muscular dystrophy-dystroglycanopathy (congenital with brain and eye anomalies), type A, 12, 615249
POMP	83.2%	83.2%	100%	100%	99.6%	Proteasome-associated autoinflammatory syndrome 2, 618048;Keratosis linearis with ichthyosis congenita and sclerosing keratoderma, 601952
POMT1	100%	99.7%	100%	100%	99.3%	Muscular dystrophy-dystroglycanopathy (congenital with brain and eye anomalies), type A, 1, 236670;Muscular dystrophy-dystroglycanopathy (limb-girdle), type C, 1, 609308;Muscular dystrophy-dystroglycanopathy (congenital with impaired intellectual development), type B, 1, 613155

POMT2	94.9%	94.9%	100%	99.8%	98.4%	Muscular dystrophy-dystroglycanopathy (limb-girdle), type C, 2, 613158;Muscular dystrophy-dystroglycanopathy (congenital with brain and eye anomalies), type A, 2, 613150;Muscular dystrophy-dystroglycanopathy (congenital with impaired intellectual development), type B, 2, 613156
POP1	100%	100%	100%	100%	99.6%	Anauxetic dysplasia 2, 617396
POPDC1	100%	100%	100%	100%	99.8%	Muscular dystrophy, limb-girdle, autosomal recessive 25, 616812
POPDC2	100%	100%	100%	100%	99.7%	Cardiac conduction disease with or without cardiomyopathy 2, 621367
POPDC3	100%	100%	100%	100%	99.6%	Muscular dystrophy, limb-girdle, autosomal recessive 26, 618848
POR	98%	94.9%	100%	100%	99.6%	Antley-Bixler syndrome with genital anomalies and disordered steroidogenesis, 201750;Disordered steroidogenesis due to cytochrome P450 oxidoreductase, 613571
POU1F1	100%	100%	100%	100%	99.7%	Pituitary hormone deficiency, combined or isolated, 1, 613038
PPA2	82.6%	82.6%	100%	100%	99.7%	?Sudden cardiac failure, alcohol-induced, 617223;Sudden cardiac failure, infantile, 617222
PPCS	100%	100%	100%	100%	99.3%	Cardiomyopathy, dilated, 2C, 618189

PPFIBP1	100%	100%	100%	100%	99.8%	Neurodevelopmental disorder with seizures, microcephaly, and brain abnormalities, 620024
PPIB	100%	100%	100%	100%	99.6%	Osteogenesis imperfecta, type IX, 259440
PPIL1	80.1%	80.1%	100%	100%	99.2%	Pontocerebellar hypoplasia, type 14, 619301
PPIP5K2	100%	100%	100%	100%	99.6%	Deafness, autosomal recessive 100, 618422
PPM1K	100%	100%	100%	100%	99.8%	Maple syrup urine disease, mild variant, 615135
PPOX	99.9%	97.6%	100%	100%	99.3%	Variegate porphyria, childhood-onset, 620483;Variegate porphyria, 176200
PPP1R13L	100%	100%	100%	99.9%	98.5%	Arrhythmogenic cardiomyopathy with variable ectodermal abnormalities, 620519
PPP1R15B	100%	100%	100%	100%	99.8%	Microcephaly, short stature, and impaired glucose metabolism 2, 616817
PPP1R21	100%	100%	100%	100%	99.8%	Neurodevelopmental disorder with hypotonia, facial dysmorphism, and brain abnormalities, 619383
PPP2R3C	100%	100%	100%	100%	99.9%	Spermatogenic failure 36, 618420;Myoectodermal gonadal dysgenesis syndrome, 618419
PPT1	90.3%	90.3%	100%	100%	99.7%	Ceroid lipofuscinosis, neuronal, 1, 256730
PRCD	100%	100%	100%	99.8%	99.7%	Retinitis pigmentosa 36, 610599

PRDM12	97.1%	92.6%	100%	99.9%	98.8%	Neuropathy, hereditary sensory and autonomic, type VIII, 616488
PRDM13	100%	100%	100%	99.9%	98.7%	Pontocerebellar hypoplasia, type 17, 619909;Cerebellar dysfunction, impaired intellectual development, and hypogonadotropic hypogonadism, 619761
PRDM5	100%	100%	100%	100%	99.8%	Brittle cornea syndrome 2, 614170
PRDX1	100%	100%	100%	100%	99.3%	Methylmalonic aciduria and homocystinuria, cblC type, digenic, 277400
PRDX3	100%	98.8%	100%	100%	99.7%	Spinocerebellar ataxia, autosomal recessive 32, 619862;Corneal dystrophy, punctiform and polychromatic pre-Descemet, 619871
PREPL	100%	100%	100%	100%	99.7%	Myasthenic syndrome, congenital, 22, 616224
PRF1	100%	100%	100%	100%	99.6%	Hemophagocytic lymphohistiocytosis, familial, 2, 603553;Aplastic anemia, 609135;Lymphoma, non-Hodgkin, 605027
PRG4	100%	100%	100%	99.8%	98.4%	Camptodactyly-arthropathy-coxa vara-pericarditis syndrome, 208250
PRICKLE1	100%	100%	100%	100%	99.8%	Epilepsy, progressive myoclonic 1B, 612437
PRIM1	91.8%	91.8%	100%	100%	99.7%	Primordial dwarfism-immunodeficiency-lipodystrophy syndrome, 620005

PRKCD	100%	100%	100%	100%	99.7%	Autoimmune lymphoproliferative syndrome, type III, 615559
PRKDC	100%	100%	100%	100%	99.7%	Immunodeficiency 26, with or without neurologic abnormalities, 615966
PRKG2	87.9%	87.9%	100%	100%	99.8%	Acromesomelic dysplasia 4, 619636;?Spondylometaphyseal dysplasia, Pagnamenta type, 619638
PRKN	100%	100%	100%	100%	99.8%	Adenocarcinoma of lung, somatic, 211980;Parkinson disease, juvenile, type 2, 600116;Ovarian cancer, somatic, 167000
PRKRA	100%	100%	100%	100%	99.8%	Dystonia 16, 612067
PRMT7	100%	100%	100%	100%	99.6%	Short stature, brachydactyly, intellectual developmental disability, and seizures, 617157
PRMT9	97.7%	97.7%	99.9%	98.9%	96.2%	
PROC	100%	100%	100%	100%	99.6%	Thrombophilia 3 due to protein C deficiency, autosomal dominant, 176860;Thrombophilia 3 due to protein C deficiency, autosomal recessive, 612304
PRODH	91.9%	91.9%	100%	99.9%	99.5%	{Schizophrenia, susceptibility to, 4}, 600850;Hyperprolinemia, type I, 239500

PROM1	100%	100%	100%	100%	99.7%	Macular dystrophy, retinal, 2, 608051;Retinitis pigmentosa 41, 612095;Stargardt disease 4, 603786;Cone-rod dystrophy 12, 612657
PROP1	100%	100%	100%	99.9%	99.1%	Pituitary hormone deficiency, combined, 2, 262600
PRORP	100%	100%	100%	100%	99.8%	Combined oxidative phosphorylation deficiency 54, 619737
PROS1	100%	100%	100%	100%	99.7%	Thrombophilia 5 due to protein S deficiency, autosomal recessive, 614514;Thrombophilia 5 due to protein S deficiency, autosomal dominant, 612336
PRSS12	100%	100%	100%	100%	99.3%	Intellectual developmental disorder, autosomal recessive 1, 249500
PRSS56	100%	100%	100%	100%	99.5%	Microphthalmia, isolated 6, 613517
PRUNE1	93.1%	93.1%	100%	100%	99.6%	Neurodevelopmental disorder with microcephaly, hypotonia, and variable brain anomalies, 617481
PRX	100%	100%	100%	100%	99.5%	Charcot-Marie-Tooth disease, type 4F, 614895;Dejerine-Sottas disease, 145900

PSAP	99.6%	97.1%	100%	100%	99.4%	Combined SAP deficiency, 611721;Krabbe disease, atypical, 611722;Metachromatic leukodystrophy due to SAP-b deficiency, 249900;Gaucher disease, atypical, 610539;{Parkinson disease 24, autosomal dominant, susceptibility to}, 619491
PSAT1	100%	100%	100%	100%	99.9%	Neu-Laxova syndrome 2, 616038;Phosphoserine aminotransferase deficiency, 610992
PSKH1	100%	100%	100%	100%	99.4%	Cholestasis, progressive familial intrahepatic, 13, 620962
PSMB10	100%	100%	100%	100%	99%	Immunodeficiency 121 with autoinflammation, 620807;Proteasome-associated autoinflammatory syndrome 5, 619175
PSMB4	100%	100%	100%	100%	99.8%	?Proteasome-associated autoinflammatory syndrome 3 and digenic forms, 617591
PSMB8	100%	100%	100%	99.9%	99.2%	Proteasome-associated autoinflammatory syndrome 1 and digenic forms, 256040
PSMB9	100%	100%	100%	100%	99.7%	Proteasome-associated autoinflammatory syndrome 6, 620796
PSMC3IP	100%	100%	100%	100%	99.5%	Ovarian dysgenesis 3, 614324
PSPH	100%	100%	100%	100%	99.8%	Phosphoserine phosphatase deficiency, 614023

PTCD3	100%	100%	100%	100%	99.7%	Combined oxidative phosphorylation deficiency 51, 619057
PTF1A	100%	100%	100%	99.5%	97.6%	Pancreatic and cerebellar agenesis, 609069;Pancreatic agenesis 2, 615935
PTH	100%	100%	100%	100%	100%	Hypoparathyroidism, familial isolated 1, 146200
PTH1R	100%	100%	100%	100%	99%	Metaphyseal chondrodysplasia, Murk Jansen type, 156400;Eiken syndrome, 600002;Failure of tooth eruption, primary, 125350;Chondrodysplasia, Blomstrand type, 215045
PTPA	100%	100%	100%	100%	99.1%	Parkinson disease 25, autosomal recessive early-onset, with impaired intellectual development, 620482
PTPMT1	100%	100%	100%	100%	99.2%	Neurodevelopmental disorder with ataxia and brain abnormalities, 621199
PTPN14	100%	100%	100%	100%	99.5%	Choanal atresia and lymphedema, 613611
PTPN23	100%	100%	100%	100%	99%	Neurodevelopmental disorder and structural brain anomalies with or without seizures and spasticity, 618890
PTPRC	100%	100%	100%	100%	99.8%	Immunodeficiency 105, severe combined, 619924
PTPRJ	100%	100%	100%	99.9%	99.5%	Colon cancer, somatic, 114500;Thrombocytopenia 10, 620484
PTPRO	100%	100%	100%	100%	99.8%	Nephrotic syndrome, type 6, 614196

PTPRQ	100%	100%	100%	99.9%	99.7%	Deafness, autosomal dominant 73, 617663;Deafness, autosomal recessive 84A, 613391
PTRH2	100%	100%	100%	100%	99.9%	Infantile-onset multisystem neurologic, endocrine, and pancreatic disease, 616263
PTRHD1	100%	100%	100%	99.8%	99.4%	Neurodevelopmental disorder with early-onset parkinsonism and behavioral abnormalities, 620747
PTS	100%	100%	100%	100%	99.6%	Hyperphenylalaninemia, BH4-deficient, A, 261640
PUS1	100%	100%	100%	99.9%	99.5%	Myopathy, lactic acidosis, and sideroblastic anemia 1, 600462
PUS3	100%	100%	100%	100%	100%	Neurodevelopmental disorder with microcephaly and gray sclerae, 617051
PUS7	100%	100%	100%	100%	99.5%	Intellectual developmental disorder with abnormal behavior, microcephaly, and short stature, 618342
PXDN	100%	99.6%	100%	99.9%	99.3%	Anterior segment dysgenesis 7, with sclerocornea, 269400
PYCR1	100%	100%	100%	100%	99.8%	Cutis laxa, autosomal recessive, type IIIB, 614438;Cutis laxa, autosomal recessive, type IIB, 612940
PYCR2	100%	100%	100%	100%	99.5%	Leukodystrophy, hypomyelinating, 10, 616420

PYGL	100%	100%	100%	99.9%	99.4%	Glycogen storage disease VI, 232700
PYGM	100%	100%	100%	100%	99.6%	McArdle disease, 232600
PYROXD1	92.6%	92.6%	100%	100%	99.6%	Myopathy, myofibrillar, 8, 617258
QARS1	100%	100%	100%	100%	99.2%	Microcephaly, progressive, seizures, and cerebral and cerebellar atrophy, 615760
QDPR	93.7%	93.6%	100%	100%	99.3%	Hyperphenylalaninemia, BH4-deficient, C, 261630
QRSL1	91.9%	89.5%	100%	100%	99.7%	Combined oxidative phosphorylation deficiency 40, 618835
RAB18	100%	100%	100%	100%	99.5%	Warburg micro syndrome 3, 614222
RAB23	100%	100%	100%	100%	99.4%	Carpenter syndrome 1, 201000
RAB27A	100%	100%	100%	100%	99.6%	Griscelli syndrome, type 2, 607624
RAB28	100%	100%	100%	100%	99.6%	Cone-rod dystrophy 18, 615374
RAB33B	100%	100%	100%	100%	99.4%	Smith-McCort dysplasia 2, 615222
RAB34	100%	100%	100%	100%	99.5%	Orofaciodigital syndrome XX, 620718
RAB3GAP1	95.7%	95.7%	100%	100%	99.6%	Martsolf syndrome 2, 619420;Warburg micro syndrome 1, 600118
RAB3GAP2	94.4%	94.4%	100%	100%	99.8%	Martsolf syndrome 1, 212720;Warburg micro syndrome 2, 614225
RACGAP1	95.4%	92.8%	100%	100%	99.7%	Anemia, congenital dyserythropoietic, type IIIb, autosomal recessive, 619789

RAD50	100%	100%	100%	100%	99.7%	Nijmegen breakage syndrome-like disorder, 613078
RAD51C	87.3%	85.8%	100%	100%	99.2%	{Breast-ovarian cancer, familial, susceptibility to, 3}, 613399;Fanconi anemia, complementation group O, 613390
RAG1	100%	100%	100%	100%	99.7%	Omenn syndrome, 603554;Severe combined immunodeficiency, B cell-negative, 601457;Combined cellular and humoral immune defects with granulomas, 233650;Alpha/beta T-cell lymphopenia with gamma/delta T-cell expansion, severe cytomegalovirus infection, and autoimmunity, 609889
RAG2	100%	100%	100%	100%	100%	Severe combined immunodeficiency, B cell-negative, 601457;Combined cellular and humoral immune defects with granulomas, 233650;Omenn syndrome, 603554
RALGAPA1	100%	100%	100%	100%	99.9%	Neurodevelopmental disorder with hypotonia, neonatal respiratory insufficiency, and thermodysregulation, 618797
RAP1GDS1	94.7%	94.7%	100%	100%	99.9%	Alfadhel syndrome, 620655
RAPSN	100%	100%	100%	100%	99.6%	Fetal akinesia deformation sequence 2, 618388;Myasthenic syndrome, congenital, 11, associated with acetylcholine receptor deficiency, 616326

RARB	98%	98%	100%	100%	99.7%	Microphthalmia, syndromic 12, 615524
RARS1	90.9%	90.5%	100%	99.9%	99.4%	Leukodystrophy, hypomyelinating, 9, 616140
RARS2	94.2%	93.8%	100%	100%	99.9%	Pontocerebellar hypoplasia, type 6, 611523
RASGRP1	95%	95%	100%	100%	99.8%	Immunodeficiency 64, 618534
RAX	100%	100%	100%	100%	99.1%	Microphthalmia, syndromic 16, 611038
RAX2	100%	100%	100%	100%	99.6%	Retinitis pigmentosa 95, 620102;Cone-rod dystrophy 11, 610381;?Macular degeneration, age-related, 6, 613757
RBBP8	100%	100%	100%	100%	99.8%	Seckel syndrome 2, 606744;Jawad syndrome, 251255;Pancreatic carcinoma, somatic
RBCK1	100%	100%	100%	99.8%	98.8%	Polyglucosan body myopathy 1 with or without immunodeficiency, 615895
RBL2	100%	100%	100%	100%	99.6%	Brunet-Wagner neurodevelopmental syndrome, 619690
RBM8A	100%	100%	100%	99.7%	98.9%	Thrombocytopenia-absent radius syndrome, 274000
RBP3	100%	100%	100%	100%	99.5%	?Retinitis pigmentosa 66, 615233
RBP4	100%	100%	100%	100%	98.8%	Microphthalmia/coloboma 10, 616428;Retinal dystrophy, iris coloboma, and comedogenic acne syndrome, 615147

RBSN	96.1%	96.1%	100%	100%	99.5%	Myelofibrosis, congenital, with anemia, neutropenia, developmental delay, and ocular abnormalities, 620939;Kariminejad neurodevelopmental syndrome, 620937
RCBTB1	100%	100%	100%	100%	99.7%	Retinal dystrophy with or without extraocular anomalies, 617175
RCC1	100%	100%	100%	99.9%	99.3%	Infection-induced acute-onset axonal neuropathy, 621333
RD3	100%	100%	100%	100%	99.4%	Leber congenital amaurosis 12, 610612
RDH11	94.9%	94%	100%	100%	99.5%	?Retinal dystrophy, juvenile cataracts, and short stature syndrome, 616108
RDH12	100%	100%	100%	100%	99.6%	Leber congenital amaurosis 13, 612712
RDH5	100%	100%	100%	100%	99.5%	Fundus albipunctatus, 136880
RDH8	100%	100%	100%	100%	99%	Stargardt disease 5, 621259
RDX	100%	100%	100%	100%	99.9%	Deafness, autosomal recessive 24, 611022
RECQL	97.9%	97.9%	100%	100%	99.7%	RECON progeroid syndrome, 620370
RECQL4	100%	100%	100%	100%	99.4%	Baller-Gerold syndrome, 218600;Rothmund-Thomson syndrome, type 2, 268400;RAPADILINO syndrome, 266280
REEP2	100%	100%	100%	99.8%	99.3%	Spastic paraplegia 72A, autosomal dominant, 615625;?Spastic paraplegia 72B, autosomal recessive, 620606

REEP6	100%	100%	100%	100%	99.5%	Retinitis pigmentosa 77, 617304
REL	100%	100%	100%	100%	99.7%	Immunodeficiency 92, 619652
RELB	100%	100%	100%	99.9%	98.6%	Immunodeficiency 53, 617585
RELN	100%	100%	100%	100%	99.8%	{Epilepsy, familial temporal lobe, 7}, 616436;Lissencephaly 2 (Norman-Roberts type), 257320
RELT	100%	100%	100%	100%	99.7%	Amelogenesis imperfecta, type IIIC, 618386
REN	100%	100%	100%	100%	99.4%	Renal tubular dysgenesis, 267430;[Hyperprorenin emia];Tubulointerstitial kidney disease, autosomal dominant, 4, 613092
REPS1	100%	100%	100%	100%	99.9%	?Neurodegeneration with brain iron accumulation 7, 617916
RETREG1	91.3%	91.3%	100%	99.9%	99.5%	Neuropathy, hereditary sensory and autonomic, type IIB, 613115
RFC1	100%	100%	100%	100%	99.7%	Cerebellar ataxia, neuropathy, and vestibular areflexia syndrome, 614575
RFC4	100%	100%	100%	100%	99.8%	Morimoto-Ryu-Malicda n neuromuscular syndrome, 621010
RFT1	84.9%	84.9%	100%	100%	99.8%	Congenital disorder of glycosylation, type In, 612015
RFWD3	100%	100%	100%	100%	99.8%	?Fanconi anemia, complementation group W, 617784

RFX5	100%	100%	100%	100%	99.8%	?MHC class II deficiency 5, 620818;MHC class II deficiency 3, 620816
RFX6	100%	100%	100%	100%	99.8%	Mitchell-Riley syndrome, 615710
RFXANK	100%	100%	100%	100%	99.4%	MHC class II deficiency 2, 620815
RFXAP	100%	100%	100%	99.8%	98.8%	MHC class II deficiency 4, 620817
RGR	100%	100%	100%	100%	99.1%	Retinitis pigmentosa 44, 613769
RGS9	95.9%	95.9%	100%	100%	99.6%	Prolonged electroretinal response suppression 1, 608415
RGS9BP	100%	100%	100%	100%	99.4%	Prolonged electroretinal response suppression 2, 620344
RHAG	100%	100%	100%	100%	99.5%	Overhydrated hereditary stomatocytosis, 185000;Anemia, hemolytic, Rh-null, regulator type, 268150
RHCE	97.1%	97.1%	96.8%	95.5%	91.9%	Rh-null disease, amorph type, 617970
RHO	100%	100%	100%	100%	99.8%	Night blindness, congenital stationary, autosomal dominant 1, 610445;Retinitis pigmentosa 4, autosomal dominant or recessive, 613731;Retinitis punctata albescens, 136880
RHOH	100%	100%	100%	100%	99.1%	Immunodeficiency 129, 618307
RIC1	100%	100%	100%	100%	99.8%	CATIFA syndrome, 618761

RIMS2	99.2%	99.2%	100%	100%	99.8%	Cone-rod synaptic disorder syndrome, congenital nonprogressive, 618970
RIN2	98.3%	98.3%	100%	100%	99.2%	Macrocephaly, alopecia, cutis laxa, and scoliosis, 613075
RINT1	93.6%	93.6%	100%	100%	99.8%	Infantile liver failure syndrome 3, 618641
RIPK1	100%	99.8%	100%	100%	99.5%	Immunodeficiency 57 with autoinflammation, 618108;Autoinflammation with episodic fever and lymphadenopathy, 618852
RIPK4	100%	100%	100%	100%	99.7%	CHAND syndrome, 214350;Popliteal pterygium syndrome, Bartsocas-Papas type 1, 263650
RIPOR2	100%	100%	100%	100%	99.8%	Deafness, autosomal dominant 21, 607017;?Deafness, autosomal recessive 104, 616515
RIPPLY2	100%	100%	100%	100%	99.9%	Spondylocostal dysostosis 6, 616566
RLBP1	100%	100%	100%	100%	99.8%	Bothnia retinal dystrophy, 607475;Newfoundland rod-cone dystrophy, 607476;Retinitis punctata albescens, 136880;Fundus albipunctatus, 136880
RMND1	85.8%	85.8%	100%	100%	99.7%	Combined oxidative phosphorylation deficiency 11, 614922
RMRP						Anauxetic dysplasia 1, 607095;Metaphyseal dysplasia without hypotrichosis, 250460;Cartilage-hair hypoplasia, 250250

RNASEH1	88.3%	88.3%	100%	100%	99.4%	Progressive external ophthalmoplegia with mitochondrial DNA deletions, autosomal recessive 2, 616479
RNASEH2A	100%	100%	100%	100%	98.8%	Aicardi-Goutieres syndrome 4, 610333
RNASEH2B	91.5%	91.5%	100%	99.9%	99.5%	Aicardi-Goutieres syndrome 2, 610181
RNASEH2C	100%	100%	100%	99.9%	98.6%	Aicardi-Goutieres syndrome 3, 610329
RNASET2	79.4%	74.3%	100%	100%	99.4%	Leukoencephalopathy, cystic, without megalencephaly, 612951
RNF168	100%	100%	100%	100%	99.3%	RIDDLE syndrome, 611943
RNF170	100%	100%	100%	100%	99.9%	Ataxia, sensory, 1, autosomal dominant, 608984;Spastic paraplegia 85, autosomal recessive, 619686
RNF216	100%	100%	100%	100%	99.7%	Cerebellar ataxia and hypogonadotropic hypogonadism, 212840
RNF220	100%	100%	100%	100%	99.7%	Leukodystrophy, hypomyelinating, 23, with ataxia, deafness, liver dysfunction, and dilated cardiomyopathy, 619688
RNF31	100%	99.9%	100%	100%	99.2%	Immunodeficiency 115 with autoinflammation, 620632
RNPC3	100%	100%	100%	100%	99.8%	Pituitary hormone deficiency, combined or isolated, 7, 618160
RNU12						CDAGS syndrome, 603116;?Spinocerebellar ataxia, autosomal recessive 33, 620208

RNU2-2						Developmental and epileptic encephalopathy 119, 621304
RNU4ATAC						Roifman syndrome, 616651;Lowry-Wood syndrome, 226960;Microcephalic osteodysplastic primordial dwarfism, type I, 210710
RNU6ATAC						
RNU7-1						Aicardi-Goutieres syndrome 9, 619487
ROBO1	100%	100%	100%	100%	99.6%	Pituitary hormone deficiency, combined or isolated, 8, 620303;Neurooculoren al syndrome, 620305;?Nystagmus 8, congenital, autosomal recessive, 257400
ROBO3	100%	100%	100%	100%	99.4%	Gaze palsy, familial horizontal, with progressive scoliosis, 1, 607313
ROGDI	100%	100%	100%	99.9%	98.8%	Kohlschutter-Tonz syndrome, 226750
ROR2	100%	100%	100%	100%	99.6%	Brachydactyly, type B1, 113000;Robinow syndrome, autosomal recessive 1, 268310
RORC	100%	100%	100%	100%	99.5%	Immunodeficiency 42, 616622
RP1	100%	100%	100%	100%	99.8%	Retinitis pigmentosa 1, 180100
RP1L1	100%	100%	100%	99.9%	99%	Occult macular dystrophy, 613587;Retinitis pigmentosa 88, 618826

RPE65	100%	100%	100%	100%	99.8%	Retinitis pigmentosa 20, 613794;Retinitis pigmentosa 87 with choroidal involvement, 618697;Leber congenital amaurosis 2, 204100
RPGRIP1	100%	100%	100%	99.9%	99.1%	Cone-rod dystrophy 13, 608194;Leber congenital amaurosis 6, 613826
RPGRIP1L	100%	100%	100%	100%	99.7%	Joubert syndrome 7, 611560;Meckel syndrome 5, 611561;?COACH syndrome 3, 619113
RPIA	100%	100%	100%	100%	99.7%	Ribose 5-phosphate isomerase deficiency, 608611
RPL3L	100%	99.9%	100%	99.8%	99.2%	Cardiomyopathy, dilated, 2D, 619371
RPS6KC1	97.6%	97.6%	100%	100%	99.5%	Neurodevelopmental disorder with spasticity, thin corpus callosum, and decreased brain white matter, 621460
RRM1	100%	100%	100%	100%	99.7%	Progressive external ophthalmoplegia with mitochondrial DNA deletions, autosomal recessive 6, 620647

RRM2B	100%	100%	100%	100%	99.9%	Mitochondrial DNA depletion syndrome 8B (MNGIE type), 612075;Mitochondrial DNA depletion syndrome 8A (encephalomyopathic type with renal tubulopathy), 612075;Rod-cone dystrophy, sensorineural deafness, and Fanconi-type renal dysfunction, 268315;Progressive external ophthalmoplegia with mitochondrial DNA deletions, autosomal dominant 5, 613077
RRP12	100%	100%	100%	100%	99.2%	Basal ganglia calcification, idiopathic, 11, autosomal recessive, 621452
RSPH1	100%	100%	100%	100%	99.5%	Ciliary dyskinesia, primary, 24, 615481
RSPH3	100%	100%	100%	99.9%	99.6%	Ciliary dyskinesia, primary, 32, 616481
RSPH4A	100%	100%	100%	100%	99.7%	Ciliary dyskinesia, primary, 11, 612649
RSPH9	100%	100%	100%	100%	99.7%	Ciliary dyskinesia, primary, 12, 612650
RSPO1	100%	100%	100%	100%	99.9%	Palmoplantar hyperkeratosis and true hermaphroditism, 610644;Palmoplantar hyperkeratosis with squamous cell carcinoma of skin and sex reversal, 610644
RSPO2	100%	100%	100%	99.9%	99.5%	?Humerofemoral hypoplasia with radiotibial ray deficiency, 618022;Tetraamelia syndrome 2, 618021

RSPO4	100%	100%	100%	100%	99.4%	Anonychia congenita, 206800
RSPRY1	95.5%	95.5%	100%	100%	99.4%	Spondyloepimetaphyseal dysplasia, Faden-Alkuraya type, 616723
RSRC1	84.2%	84.2%	100%	99.9%	99.5%	Intellectual developmental disorder, autosomal recessive 70, 618402
RTEL1	100%	100%	100%	99.9%	99.4%	Dyskeratosis congenita, autosomal dominant 4, 615190;Dyskeratosis congenita, autosomal recessive 5, 615190;Pulmonary fibrosis and/or bone marrow failure syndrome, telomere-related, 3, 616373
RTN2	100%	100%	100%	100%	99.2%	Neuronopathy, distal hereditary motor, autosomal recessive 11, with spasticity, 620854;Spastic paraplegia 12, autosomal dominant, 604805
RTN4IP1	100%	100%	100%	100%	99.8%	Optic atrophy 10 with or without ataxia, impaired intellectual development and seizures, 616732
RTTN	100%	100%	100%	100%	99.8%	Microcephaly, short stature, and polymicrogyria with seizures, 614833
RUBCN	100%	100%	100%	99.9%	99.5%	Spinocerebellar ataxia, autosomal recessive 15, 615705
RUSC2	100%	100%	100%	100%	99.7%	Intellectual developmental disorder, autosomal recessive 61, 617773

RXYLT1	100%	100%	100%	99.8%	99.3%	Muscular dystrophy-dystroglycanopathy (congenital with brain and eye anomalies), type A, 10, 615041
RYR1	100%	100%	100%	99.9%	99%	Congenital myopathy 1B, autosomal recessive, 255320;Congenital myopathy 1A, autosomal dominant, with susceptibility to malignant hyperthermia, 117000;King-Denborough syndrome, 619542;{Malignant hyperthermia susceptibility 1}, 145600
RYR3	100%	100%	100%	100%	99.7%	Congenital myopathy 20, 620310
S1PR2	100%	100%	100%	100%	99.7%	Deafness, autosomal recessive 68, 610419
SACS	99%	99%	100%	100%	99.9%	Spastic ataxia, Charlevoix-Saguenay type, 270550
SAG	100%	100%	100%	100%	99.6%	Retinitis pigmentosa 47, autosomal recessive, 613758;Retinitis pigmentosa 96, autosomal dominant, 620228;Oguchi disease-1, 258100
SAMD7	100%	100%	100%	100%	99.2%	Macular dystrophy with or without cone dysfunction, 620762
SAMD9	100%	100%	100%	100%	99.8%	Tumoral calcinosis, familial, normophosphatemic, 610455;Monosomy 7 myelodysplasia and leukemia syndrome 2, 619041;MIRAGE syndrome, 617053

SAMHD1	92%	89.8%	100%	100%	99.5%	?Chilblain lupus 2, 614415;Aicardi-Goutieres syndrome 5, 612952
SAR1B	88.5%	84.5%	100%	100%	98.9%	Chylomicron retention disease, 246700
SARS1	89.1%	89.1%	100%	100%	99.3%	Neurodevelopmental disorder with microcephaly, ataxia, and seizures, 617709
SARS2	100%	100%	100%	100%	99.7%	Hyperuricemia, pulmonary hypertension, renal failure, and alkalosis, 613845
SASH1	98.3%	97.2%	100%	100%	99.5%	Dyschromatosis universalis hereditaria 1, 127500;?Cancer, alopecia, pigment dyscrasia, onychodystrophy, and keratoderma, 618373
SASS6	100%	100%	100%	100%	99.7%	Microcephaly 14, primary, autosomal recessive, 616402
SBDS	100%	100%	100%	100%	99.9%	{Aplastic anemia, susceptibility to}, 609135;Shwachman-Diamond syndrome 1, 260400
SBF1	100%	100%	100%	100%	99.2%	Charcot-Marie-Tooth disease, type 4B3, 615284
SBF2	93.7%	93.7%	100%	100%	99.7%	Charcot-Marie-Tooth disease, type 4B2, 604563
SC5D	100%	100%	100%	100%	99.9%	Lathosterolosis, 607330
SCAPER	97.9%	97.9%	100%	100%	99.8%	Intellectual developmental disorder and retinitis pigmentosa, 618195

SCARB2	100%	100%	100%	100%	99.7%	Epilepsy, progressive myoclonic 4, with or without renal failure, 254900
SCARF2	100%	100%	100%	99.8%	97.6%	Van den Ende-Gupta syndrome, 600920
SCN1B	100%	100%	100%	100%	99.5%	Generalized epilepsy with febrile seizures plus, type 1, 604233;Developmental and epileptic encephalopathy 52, 617350;Cardiac conduction defect, nonspecific, 612838;Atrial fibrillation, familial, 13, 615377;Brugada syndrome 5, 612838
SCN4A	100%	100%	100%	100%	99.4%	Paramyotonia congenita, 168300;Hyperkalemic periodic paralysis, 170500;Congenital myopathy 22B, severe fetal, 620369;Hypokalemic periodic paralysis, type 2, 613345;Myotonia congenita, atypical, acetazolamide-responsive, 608390;Myasthenic syndrome, congenital, 16, 614198;Congenital myopathy 22A, classic, 620351

SCN5A	100%	100%	100%	100%	99.5%	Ventricular fibrillation, familial, 1, 603829;Heart block, progressive, type IA, 113900;Cardiomyopathy, dilated, 1E, 601154;Heart block, nonprogressive, 113900;Long QT syndrome 3, 603830;Sick sinus syndrome 1, 608567;Brugada syndrome 1, 601144;Atrial fibrillation, familial, 10, 614022;{Sudden infant death syndrome, susceptibility to}, 272120
SCN9A	100%	100%	100%	100%	99.5%	Erythermalgia, primary, 133020;Insensitivity to pain, congenital, 243000;Small fiber neuropathy, 133020;Paroxysmal extreme pain disorder, 167400;Neuropathy, hereditary sensory and autonomic, type IID, 243000
SCNM1	100%	100%	100%	99.7%	98.4%	Orofaciodigital syndrome XIX, 620107
SCNN1A	100%	100%	100%	99.9%	99.5%	Pseudohypoaldosteronism, type IB1, autosomal recessive, 264350;?Liddle syndrome 3, 618126;Bronchiectasis with or without elevated sweat chloride 2, 613021
SCNN1B	100%	100%	100%	100%	99.6%	Bronchiectasis with or without elevated sweat chloride 1, 211400;Pseudohypoaldosteronism, type IB2, autosomal recessive, 620125;Liddle syndrome 1, 177200

SCNN1G	100%	100%	100%	100%	99.2%	Bronchiectasis with or without elevated sweat chloride 3, 613071;Pseudohypoaldosteronism, type IB3, autosomal recessive, 620126;Liddle syndrome 2, 618114
SCO1	100%	100%	100%	100%	99.6%	Mitochondrial complex IV deficiency, nuclear type 4, 619048
SCO2	100%	100%	100%	100%	99.8%	Myopia 6, 608908;Mitochondrial complex IV deficiency, nuclear type 2, 604377
SCP2	95.5%	95.5%	100%	100%	99.7%	?Leukoencephalopathy with dystonia and motor neuropathy, 613724
SCUBE3	97.5%	97.5%	100%	100%	99.8%	Short stature, facial dysmorphism, and skeletal anomalies with or without cardiac anomalies 2, 619184
SCYL1	100%	100%	100%	99.9%	99.1%	Spinocerebellar ataxia, autosomal recessive 21, 616719
SCYL2	97.3%	97.3%	100%	100%	99.9%	Arthrogryposis multiplex congenita 4, neurogenic, with agenesis of the corpus callosum, 618766
SDCCAG8	100%	100%	100%	100%	99.9%	Senior-Loken syndrome 7, 613615;Bardet-Biedl syndrome 16, 615993
SDHA	92.6%	89.3%	100%	100%	99.7%	Cardiomyopathy, dilated, 1GG, 613642;Mitochondrial complex II deficiency, nuclear type 1, 252011;Neurodegeneration with ataxia and late-onset optic atrophy, 619259;Pheochromocytoma/paraganglioma syndrome 5, 614165

SDHAF1	100%	100%	100%	100%	99.3%	Mitochondrial complex II deficiency, nuclear type 2, 619166
SDHB	94%	94%	100%	100%	99.7%	Pheochromocytoma/paraganglioma syndrome 4, 115310;Mitochondrial complex II deficiency, nuclear type 4, 619224;Gastrointestinal stromal tumor, 606764;Paraganglioma and gastric stromal sarcoma, 606864
SDHD	78.9%	78.9%	100%	100%	99.5%	Pheochromocytoma/paraganglioma syndrome 1, 168000;Paraganglioma and gastric stromal sarcoma, 606864;Mitochondrial complex II deficiency, nuclear type 3, 619167
SDR9C7	100%	100%	100%	100%	99.9%	Ichthyosis, congenital, autosomal recessive 13, 617574
SEC23A	100%	100%	100%	100%	99.7%	Craniolenticulosutural dysplasia, 607812
SEC23B	100%	100%	100%	100%	99.7%	?Cowden syndrome 7, 616858;Dyserythropoietic anemia, congenital, type II, 224100
SEC24D	100%	100%	100%	100%	99.6%	Cole-Carpenter syndrome 2, 616294
SEC31A	96.6%	96.6%	100%	99.9%	99.7%	?Halperin-Birk syndrome, 618651
SECISBP2	100%	100%	100%	100%	99.7%	Thyroid hormone metabolism, abnormal, 1, 609698
SELENBP1	93.1%	93.1%	100%	100%	99.4%	Extraoral halitosis due to MTO deficiency, 618148
SELENOI	100%	100%	100%	100%	99.9%	Spastic paraplegia 81, autosomal recessive, 618768

SELENON	93.9%	91.8%	100%	99.8%	99.3%	Congenital myopathy 3 with rigid spine, 602771
SEMA4A	100%	100%	100%	100%	98.9%	Retinitis pigmentosa 35, 610282;Cone-rod dystrophy 10, 610283
SEPSECS	99.4%	96.3%	100%	100%	99.5%	Pontocerebellar hypoplasia type 2D, 613811
SERAC1	100%	100%	100%	100%	99.9%	3-methylglutaconic aciduria with deafness, encephalopathy, and Leigh-like syndrome, 614739
SERPINA1	100%	100%	100%	100%	99.3%	Hemorrhagic diathesis due to antithrombin Pittsburgh, 613490;Emphysema due to AAT deficiency, 613490;Emphysema-cirrhosis, due to AAT deficiency, 613490
SERPINA6	93%	93%	100%	100%	99.8%	Corticosteroid-binding globulin deficiency, 611489
SERPINB7	100%	100%	100%	100%	99.7%	Palmoplantar keratoderma, Nagashima type, 615598
SERPINB8	100%	100%	100%	100%	99.6%	Peeling skin syndrome 5, 617115
SERPINC1	90.4%	88.6%	100%	100%	99.9%	Thrombophilia 7 due to antithrombin III deficiency, 613118
SERPINE1	100%	100%	100%	100%	99.6%	Plasminogen activator inhibitor-1 deficiency, 613329;{Transcription of plasminogen activator inhibitor, modulator of}
SERPINF1	100%	100%	100%	100%	99%	Osteogenesis imperfecta, type VI, 613982

SERPINF2	92.1%	91.2%	100%	100%	99.4%	Alpha-2-plasmin inhibitor deficiency, 262850
SERPING1	100%	100%	100%	100%	99.6%	Angioedema, hereditary, 1 and 2, 106100;Complement component 4, partial deficiency of, 120790
SERPINH1	100%	100%	100%	100%	99.6%	{Preterm premature rupture of the membranes, susceptibility to}, 610504;Osteogenesis imperfecta, type X, 613848
SETX	100%	100%	100%	100%	99.8%	Spinocerebellar ataxia, autosomal recessive, with axonal neuropathy 2, 606002;Amyotrophic lateral sclerosis 4, juvenile, 602433
SFRP4	100%	100%	100%	100%	99.8%	Pyle disease, 265900
SFTPB	100%	100%	100%	100%	99.3%	Surfactant metabolism dysfunction, pulmonary, 1, 265120
SFXN4	100%	100%	100%	99.9%	99.6%	Combined oxidative phosphorylation deficiency 18, 615578
SGCA	97.5%	97.5%	100%	100%	99.2%	Muscular dystrophy, limb-girdle, autosomal recessive 3, 608099
SGCB	100%	100%	100%	100%	99.8%	Muscular dystrophy, limb-girdle, autosomal recessive 4, 604286
SGCD	94%	94%	100%	100%	99.7%	Cardiomyopathy, dilated, 1L, 606685;Muscular dystrophy, limb-girdle, autosomal recessive 6, 601287
SGCG	83.2%	83.2%	100%	100%	99.8%	Muscular dystrophy, limb-girdle, autosomal recessive 5, 253700

SGO1	100%	100%	100%	100%	99.9%	Chronic atrial and intestinal dysrhythmia, 616201
SGPL1	95.5%	95.5%	100%	100%	99.7%	RENI syndrome, 617575
SGSH	100%	100%	100%	100%	99.5%	Mucopolysaccharidosis type IIIA (Sanfilippo A), 252900
SGSM3	100%	100%	100%	100%	99.7%	Intellectual developmental disorder, autosomal recessive 84, 620401
SH3PXD2B	99.9%	98.6%	100%	100%	99.2%	Frank-ter Haar syndrome, 249420
SH3TC2	100%	100%	100%	100%	99.5%	Charcot-Marie-Tooth disease, type 4C, 601596;Mononeuropathy of the median nerve, mild, 613353
SHARPIN	100%	100%	100%	100%	99.3%	Autoinflammation with episodic fever and immune dysregulation, 620795
SHMT2	100%	100%	100%	100%	99.7%	Neurodevelopmental disorder with cardiomyopathy, spasticity, and brain abnormalities, 619121
SHQ1	100%	100%	100%	100%	99.7%	Neurodevelopmental disorder with dystonia and seizures, 619922;?Dystonia 35, childhood-onset, 619921
SI	100%	100%	100%	99.7%	99.1%	Sucrase-isomaltase deficiency, congenital, 222900
SIGMAR1	100%	100%	100%	100%	100%	?Neuronopathy, distal hereditary motor, autosomal recessive 2, 605726;?Amyotrophic lateral sclerosis 16, juvenile, 614373

SIK3	100%	100%	100%	100%	99.4%	?Spondyloepimetaphyseal dysplasia, Krakow type, 618162
SIL1	91.5%	91.5%	100%	100%	99.5%	Marinesco-Sjogren syndrome, 248800
SIX6	100%	100%	100%	100%	99.3%	Optic disc anomalies with retinal and/or macular dystrophy, 212550
SKIC2	99.7%	98.5%	100%	100%	99.6%	Trichohepatoenteric syndrome 2, 614602
SKIC3	98.9%	98.9%	100%	100%	99.9%	Trichohepatoenteric syndrome 1, 222470
SKOR2	100%	100%	100%	99.8%	97.1%	Valence-Farazi cerebellar ataxia syndrome, 621386
SLC10A7	92.8%	92.8%	100%	100%	100%	Short stature, amelogenesis imperfecta, and skeletal dysplasia with scoliosis, 618363
SLC11A2	100%	100%	100%	100%	99.6%	Anemia, hypochromic microcytic, with iron overload 1, 206100
SLC12A1	96.3%	96.3%	100%	100%	99.8%	Bartter syndrome, type 1, 601678
SLC12A2	100%	100%	100%	100%	99.6%	Kilquist syndrome, 619080;Delpire-McNeill syndrome, 619083;Deafness, autosomal dominant 78, 619081
SLC12A3	94.7%	94.7%	100%	100%	99.7%	Gitelman syndrome, 263800
SLC12A5	100%	100%	100%	99.9%	99.4%	{Epilepsy, idiopathic generalized, susceptibility to, 14}, 616685;Developmental and epileptic encephalopathy 34, 616645

SLC12A6	100%	100%	100%	100%	99.9%	Agenesis of the corpus callosum with peripheral neuropathy, 218000;Charcot-Marie-Tooth disease, axonal, type 2II, 620068
SLC13A1	100%	100%	100%	100%	99.8%	
SLC13A3	100%	100%	100%	100%	99.6%	Leukoencephalopathy, acute reversible, with increased urinary alpha-ketoglutarate, 618384
SLC13A5	100%	100%	100%	100%	99.8%	Developmental and epileptic encephalopathy 25, with amelogenesis imperfecta, 615905
SLC16A1	100%	100%	100%	100%	99.9%	Hyperinsulinemic hypoglycemia, familial, 7, 610021;Erythrocyte lactate transporter defect, 245340;Monocarboxylate transporter 1 deficiency, 616095
SLC17A5	92.7%	92.7%	100%	100%	99.8%	Salla disease, 604369;Sialic acid storage disorder, infantile, 269920
SLC18A2	99.6%	96.7%	100%	100%	99.8%	Parkinsonism-dystonia, infantile, 2, 618049
SLC18A3	100%	100%	100%	100%	98.8%	Myasthenic syndrome, congenital, 21, presynaptic, 617239
SLC19A1	97.7%	97.4%	100%	99.9%	98.9%	Immunodeficiency 114, folate-responsive, 620603;?Megaloblastic anemia, folate-responsive, 601775
SLC19A2	100%	100%	100%	100%	99.7%	Thiamine-responsive megaloblastic anemia syndrome, 249270

SLC19A3	99.9%	98.9%	100%	100%	99.7%	Thiamine metabolism dysfunction syndrome 2 (biotin/thiamine-responsive basal ganglia disease type), 607483
SLC1A1	100%	100%	100%	99.9%	99.4%	Dicarboxylic aminoaciduria, 222730;{?Schizophrenia susceptibility 18}, 615232
SLC1A4	100%	100%	100%	99.9%	99.3%	Spastic tetraplegia, thin corpus callosum, and progressive microcephaly, 616657
SLC22A12	100%	100%	100%	99.9%	98.7%	Hypouricemia, renal, 220150
SLC22A5	97.1%	97.1%	100%	100%	99.5%	Carnitine deficiency, systemic primary, 212140
SLC24A1	100%	100%	100%	100%	99.4%	Night blindness, congenital stationary (complete), 1D, autosomal recessive, 613830
SLC24A4	100%	100%	100%	100%	99.3%	[Skin/hair/eye pigmentation 6, blond/brown hair], 210750;Amelogenesis imperfecta, type IIA5, 615887;[Skin/hair/eye pigmentation 6, blue/green eyes], 210750
SLC24A5	100%	100%	100%	100%	99.7%	[Skin/hair/eye pigmentation 4, fair/dark skin], 113750;Albinism, oculocutaneous, type VI, 113750
SLC25A1	100%	100%	100%	99.9%	98.8%	Combined D-2- and L-2-hydroxyglutaric aciduria, 615182;Myasthenic syndrome, congenital, 23, presynaptic, 618197

SLC25A12	90.5%	89.5%	100%	100%	99.9%	Developmental and epileptic encephalopathy 39, 612949
SLC25A13	91.7%	91.7%	100%	100%	99.6%	Citrullinemia, type II, neonatal-onset, 605814;Citrullinemia, adult-onset type II, 603471
SLC25A15	100%	100%	100%	100%	99.8%	Hyperornithinemia-hyperammonemia-homocitrullinemia syndrome, 238970
SLC25A19	100%	100%	100%	100%	99.8%	Microcephaly, Amish type, 607196;Thiamine metabolism dysfunction syndrome 4 (progressive polyneuropathy type), 613710
SLC25A20	100%	100%	100%	100%	99.4%	Carnitine-acylcarnitine translocase deficiency, 212138
SLC25A22	100%	100%	100%	99.9%	99.2%	Developmental and epileptic encephalopathy 3, 609304
SLC25A26	95.5%	95.5%	100%	100%	99.7%	Combined oxidative phosphorylation deficiency 28, 616794
SLC25A3	100%	100%	100%	100%	99.6%	Mitochondrial phosphate carrier deficiency, 610773
SLC25A36	95%	95%	100%	100%	99.4%	Hyperinsulinemic hypoglycemia, familial, 8, 620211
SLC25A38	90.6%	90.6%	100%	100%	99.4%	Anemia, sideroblastic, 2, pyridoxine-refractory, 205950

SLC25A4	100%	100%	100%	100%	99.4%	Mitochondrial DNA depletion syndrome 12B (cardiomyopathic type) AR, 615418;Progressive external ophthalmoplegia with mitochondrial DNA deletions, autosomal dominant 2, 609283;Mitochondrial DNA depletion syndrome 12A (cardiomyopathic type) AD, 617184
SLC25A42	100%	100%	100%	100%	99.2%	Metabolic crises, recurrent, with variable encephalomyopathic features and neurologic regression, 618416
SLC25A46	100%	100%	100%	100%	99.7%	Neuropathy, hereditary motor and sensory, type VIB, 616505;Pontocerebellar hypoplasia, type 1E, 619303
SLC26A1	100%	100%	100%	100%	99.5%	?Hypersulfaturia, 620372;?Nephrolithiasis, calcium oxalate, 1, 167030
SLC26A2	100%	100%	100%	100%	99.8%	Epiphyseal dysplasia, multiple, 4, 226900;De la Chapelle dysplasia, 256050;Diastrophic dysplasia, 222600;Diastrophic dysplasia, broad bone-platyspondylic variant, 222600;Achondrogenesis IB, 600972;Atelosteogenesis, type II, 256050
SLC26A3	100%	100%	100%	100%	99.9%	Diarrhea 1, secretory chloride, congenital, 214700

SLC26A4	100%	100%	100%	100%	99.8%	Deafness, autosomal recessive 4, with enlarged vestibular aqueduct, 600791;Pendred syndrome, 274600
SLC27A4	100%	100%	100%	100%	99.6%	Ichthyosis prematurity syndrome, 608649
SLC29A3	100%	100%	100%	100%	99.5%	Histiocytosis-lymphadenopathy plus syndrome, 602782
SLC2A1	100%	100%	100%	100%	99.3%	Dystonia 9, 601042;GLUT1 deficiency syndrome 1, infantile onset, severe, 606777;Stomatin-deficient cryohydrocytosis with neurologic defects, 608885;{Epilepsy, idiopathic generalized, susceptibility to, 12}, 614847;GLUT1 deficiency syndrome 2, childhood onset, 612126
SLC2A10	94.7%	94.7%	100%	100%	99.8%	Arterial tortuosity syndrome, 208050
SLC2A2	100%	100%	100%	100%	99.8%	Fanconi-Bickel syndrome, 227810;{Diabetes mellitus, noninsulin-dependent}, 125853
SLC2A9	100%	100%	100%	100%	99.7%	{Uric acid concentration, serum, QTL 2}, 612076;Hypouricemia, renal, 2, 612076
SLC30A10	100%	100%	100%	100%	99.3%	Hypermanganesemia with dystonia 1, 613280
SLC30A7	100%	100%	100%	100%	99.9%	Ziegler-Huang syndrome, 620501
SLC30A9	100%	100%	100%	100%	99.9%	Birk-Landau-Perez syndrome, 617595

SLC31A1	100%	100%	100%	99.9%	99.5%	Neurodegeneration and seizures due to copper transport defect, 620306
SLC33A1	92.6%	92.6%	100%	100%	99.7%	Spastic paraplegia 42, autosomal dominant, 612539;Huppke-Brendel syndrome, 614482
SLC34A1	100%	100%	100%	99.9%	99.4%	?Fanconi renotubular syndrome 2, 613388;Hypercalcemia, infantile, 2, 616963;Nephrolithiasis/osteoporosis, hypophosphatemic, 1, 612286
SLC34A2	100%	100%	100%	100%	99.5%	Pulmonary alveolar microlithiasis, 265100
SLC34A3	100%	100%	100%	99.8%	98.6%	Hypophosphatemic rickets with hypercalciuria, 241530
SLC35A1	94.9%	94.9%	100%	100%	100%	Congenital disorder of glycosylation, type IIf, 603585
SLC35A3	91%	87.6%	100%	100%	99.8%	Arthrogryposis, impaired intellectual development, and seizures, 615553
SLC35B2	100%	100%	100%	100%	99.2%	Leukodystrophy, hypomyelinating, 26, with chondrodysplasia, 620269
SLC35C1	100%	100%	100%	100%	98.9%	Congenital disorder of glycosylation, type IIc, 266265
SLC35D1	100%	100%	100%	100%	99.8%	Schneckenbecken dysplasia, 269250
SLC37A4	100%	100%	100%	100%	99.8%	Glycogen storage disease Ib, 232220;Congenital disorder of glycosylation, type IIw, 619525;Glycogen storage disease Ic, 232240

SLC38A3	96.3%	96.3%	100%	100%	99.4%	Developmental and epileptic encephalopathy 102, 619881
SLC38A8	100%	100%	100%	100%	99.5%	Foveal hypoplasia 2, with or without optic nerve misrouting and/or anterior segment dysgenesis, 609218
SLC39A13	99.8%	97.5%	100%	100%	99.5%	Ehlers-Danlos syndrome, spondylodysplastic type, 3, 612350
SLC39A14	94%	94%	100%	100%	99.5%	?Hyperostosis cranalis interna, 144755;Hypermanganesemia with dystonia 2, 617013
SLC39A4	100%	100%	100%	100%	99.5%	Acrodermatitis enteropathica, 201100
SLC39A7	100%	100%	100%	100%	99.6%	Agammaglobulinemia 9, autosomal recessive, 619693
SLC39A8	77.5%	77.4%	100%	100%	100%	Congenital disorder of glycosylation, type IIn, 616721
SLC3A1	96.2%	96.2%	100%	100%	99.8%	Cystinuria, 220100
SLC44A1	97.5%	97.5%	100%	100%	99.8%	Neurodegeneration, childhood-onset, with ataxia, tremor, optic atrophy, and cognitive decline, 618868
SLC45A1	100%	100%	100%	100%	99.4%	Intellectual developmental disorder with neuropsychiatric features, 617532

SLC45A2	100%	100%	100%	100%	99.6%	[Skin/hair/eye pigmentation 5, dark/light eyes], 227240;[Skin/hair/eye pigmentation 5, black/nonblack hair], 227240;Albinism, oculocutaneous, type IV, 606574;[Skin/hair/eye pigmentation 5, dark/fair skin], 227240
SLC46A1	100%	100%	99.9%	99.7%	99%	Folate malabsorption, hereditary, 229050
SLC4A1	100%	100%	100%	100%	99.3%	[Blood group, Swann], 601550;[Blood group, Wright], 112050;Distal renal tubular acidosis 1, 179800;[Blood group, Waldner], 112010;Spherocytosis, type 4, 612653;[Blood group, Froese], 601551;Distal renal tubular acidosis 4 with hemolytic anemia, 611590;{Malaria, resistance to}, 611162;Cryohydrocytosis, 185020;Ovalocytosis, SA type, 166900;[Blood group, Diego], 110500
SLC4A10	100%	100%	100%	100%	99.9%	Neurodevelopmental disorder with hypotonia and characteristic brain abnormalities, 620746
SLC4A11	100%	100%	100%	100%	99.6%	Corneal endothelial dystrophy, autosomal recessive, 217700;Corneal dystrophy, Fuchs endothelial, 4, 613268;Corneal endothelial dystrophy and perceptive deafness, 217400

SLC4A4	95%	95%	100%	100%	99.9%	Proximal renal tubular acidosis-ocular anomaly syndrome, 604278
SLC52A2	100%	100%	100%	99.9%	99.7%	Brown-Vialetto-Van Laere syndrome 2, 614707
SLC52A3	100%	100%	100%	100%	99.4%	?Fazio-Londe disease, 211500;Brown-Vialetto-Van Laere syndrome 1, 211530
SLC5A1	100%	100%	100%	100%	99.4%	Glucose/galactose malabsorption, 606824
SLC5A2	100%	100%	100%	99.9%	99.4%	Renal glucosuria, 233100
SLC5A5	100%	100%	100%	100%	99.4%	Thyroid dyshormonogenesis 1, 274400
SLC5A6	100%	100%	100%	100%	99.5%	Sodium-dependent multivitamin transporter deficiency, 618973;Peripheral motor neuropathy, childhood-onset, biotin-responsive, 619903
SLC5A7	100%	100%	100%	99.9%	99.4%	Neuronopathy, distal hereditary motor, autosomal dominant 7, 158580;Myasthenic syndrome, congenital, 20, presynaptic, 617143
SLC6A17	100%	100%	100%	100%	99.2%	Intellectual developmental disorder, autosomal recessive 48, 616269
SLC6A19	100%	100%	100%	99.9%	99%	Hartnup disorder, 234500
SLC6A3	93.9%	92.4%	100%	100%	99.6%	Parkinsonism-dystonia, infantile, 1, 613135;{Nicotine dependence, protection against}, 188890

SLC6A5	100%	100%	100%	100%	99.6%	Hyperekplexia 3, 614618
SLC6A6	100%	100%	100%	100%	99.4%	Hypotaurinemic retinal degeneration and cardiomyopathy, 145350
SLC6A9	100%	100%	100%	100%	99.2%	Glycine encephalopathy with normal serum glycine, 617301;{Scoliosis, isolated, susceptibility to, 6}, 621428
SLC7A14	100%	100%	100%	99.9%	99.6%	Retinitis pigmentosa 68, 615725
SLC7A6OS	87%	87%	100%	100%	99.6%	Epilepsy, progressive myoclonic, 12, 619191
SLC7A7	100%	100%	100%	100%	98.8%	Lysinuric protein intolerance, 222700
SLC7A9	100%	100%	100%	100%	99.5%	Cystinuria, 220100
SLC9A1	100%	100%	100%	99.9%	99.1%	Lichtenstein-Knorr syndrome, 616291
SLC9A3	100%	100%	100%	99.7%	98.5%	Diarrhea 8, secretory sodium, congenital, 616868
SLCO2A1	100%	100%	100%	100%	99.3%	Hypertrophic osteoarthropathy, primary, autosomal dominant, 167100;PHOAR2-enteropathy syndrome, 614441
SLF2	100%	100%	100%	100%	99.8%	Atelis syndrome 1, 620184
SLITRK6	100%	100%	100%	100%	99.7%	Deafness and myopia, 221200
SLURP1	100%	100%	100%	100%	99.2%	Meleda disease, 248300
SLX4	100%	100%	100%	100%	99.4%	Fanconi anemia, complementation group P, 613951

SMARCAL1	95.6%	95.6%	100%	100%	99.7%	Schimke immunoosseous dysplasia, 242900
SMARCD2	100%	100%	100%	99.9%	99%	Specific granule deficiency 2, 617475
SMC5	97.3%	97.3%	100%	100%	99.8%	Atelis syndrome 2, 620185
SMG8	100%	100%	100%	100%	99.7%	Alzahrani-Kuwahara syndrome, 619268
SMG9	100%	100%	100%	100%	99.6%	Heart and brain malformation syndrome, 616920;Neurodevelopmental disorder with intention tremor, pyramidal signs, dyspraxia, and ocular anomalies, 619995
SMN1	93.9%	93.9%	100%	99.5%	98.3%	Spinal muscular atrophy-2, 253550;Spinal muscular atrophy-4, 271150;Spinal muscular atrophy-3, 253400;Spinal muscular atrophy-1, 253300
SMO	100%	100%	100%	100%	99.7%	Pallister-Hall-like syndrome, 241800;Basal cell carcinoma, somatic, 605462;Curry-Jones syndrome, somatic mosaic, 601707
SMOC1	96%	96%	100%	100%	99.7%	Microphthalmia with limb anomalies, 206920
SMOC2	100%	100%	100%	100%	99.6%	Dentin dysplasia, type IA, 125400
SMPD1	100%	100%	100%	100%	99.6%	Niemann-Pick disease, type B, 607616;Niemann-Pick disease, type A, 257200

SMPD4	100%	100%	100%	100%	99.6%	Neurodevelopmental disorder with microcephaly, arthrogryposis, and structural brain anomalies, 618622
SNAI2	100%	100%	100%	100%	99.7%	
SNAP29	100%	100%	100%	99.7%	99.3%	Cerebral dysgenesis, neuropathy, ichthyosis, and palmoplantar keratoderma syndrome, 609528
SNAPC4	100%	100%	100%	100%	99.3%	Neurodevelopmental disorder with motor regression, progressive spastic paraplegia, and oromotor dysfunction, 620515
SNAPIN	100%	100%	100%	100%	99.3%	Neurodevelopmental disorder with structural brain abnormalities and craniofacial abnormalities, 621393
SNF8	94.6%	94.6%	100%	100%	99.6%	Developmental and epileptic encephalopathy 115, 620783;Neurodevelopmental disorder plus optic atrophy, 620784
SNIP1	100%	100%	100%	100%	99.8%	Neurodevelopmental disorder with hypotonia, craniofacial abnormalities, and seizures, 614501
SNORD118						Leukoencephalopathy, brain calcifications, and cysts, 614561
SNUPN	100%	100%	100%	100%	99.8%	Muscular dystrophy, limb-girdle, autosomal recessive 29, 620793
SNX10	89.3%	89.3%	100%	100%	99.6%	Osteopetrosis, autosomal recessive 8, 615085

SNX14	95%	95%	100%	100%	99.8%	Spinocerebellar ataxia, autosomal recessive 20, 616354
SNX27	100%	100%	100%	100%	99.5%	
SOBP	100%	100%	100%	99.8%	97%	?Impaired intellectual development, anterior maxillary protrusion, and strabismus, 613671
SOD1	100%	100%	100%	100%	99.7%	Spastic tetraplegia and axial hypotonia, progressive, 618598;Amyotrophic lateral sclerosis 1, 105400
SORD	83%	78.5%	98%	95.9%	93.1%	Neuronopathy, distal hereditary motor, autosomal recessive 8, 618912
SOST	100%	100%	100%	99.9%	98.7%	Sclerosteosis 1, 269500;Craniodiaphyseal dysplasia, autosomal dominant, 122860
SOX18	100%	100%	100%	99.5%	98%	Hypotrichosis-lymphedema-telangiectasia syndrome, 607823;Hypotrichosis-lymphedema-telangiectasia-renal defect syndrome, 137940
SP110	100%	100%	100%	100%	99.8%	{Mycobacterium tuberculosis, susceptibility to}, 607948;Hepatic venoocclusive disease with immunodeficiency, 235550
SP7	100%	100%	100%	100%	99.5%	Osteogenesis imperfecta, type XII, 613849
SPAG1	100%	100%	100%	100%	99.6%	Ciliary dyskinesia, primary, 28, 615505
SPARC	100%	100%	100%	100%	99.4%	Osteogenesis imperfecta, type XVII, 616507

SPART	95.8%	95.8%	100%	100%	99.7%	Troyer syndrome, 275900
SPATA7	100%	100%	100%	100%	99.9%	Leber congenital amaurosis 3, 604232;Retinitis pigmentosa 94, variable age at onset, autosomal recessive, 604232
SPEG	100%	100%	100%	99.9%	98.7%	Centronuclear myopathy 5, 615959
SPG11	99.6%	99.6%	100%	100%	99.7%	Amyotrophic lateral sclerosis 5, juvenile, 602099;Charcot-Marie-Tooth disease, axonal, type 2X, 616668;Spastic paraplegia 11, autosomal recessive, 604360
SPG21	100%	100%	100%	100%	99.7%	Mast syndrome, 248900
SPG7	100%	100%	100%	99.9%	99.3%	Spastic paraplegia 7, autosomal recessive, 607259
SPINK5	100%	100%	100%	100%	99.8%	Netherton syndrome, 256500
SPINT2	100%	100%	100%	100%	99.2%	Diarrhea 3, secretory sodium, congenital, syndromic, 270420
SPOUT1	100%	100%	100%	100%	99.4%	Neurodevelopmental disorder with poor growth, seizures, and brain abnormalities, 621154
SPR	100%	100%	100%	100%	99.8%	Dystonia, dopa-responsive, due to sepiapterin reductase deficiency, 612716
SPRED2	100%	100%	100%	100%	99.8%	Noonan syndrome 14, 619745
SPRTN	100%	100%	100%	100%	99.6%	Ruijs-Aalfs progeroid syndrome, 616200

SPTA1	100%	100%	100%	100%	99.7%	Spherocytosis, type 3, 270970;Elliptocytosis-2, 130600;Pyropoikilocytosis, 266140
SPTB	100%	100%	100%	100%	99.4%	Anemia, neonatal hemolytic, fatal or near-fatal, 617948;Elliptocytosis-3, 617948;Spherocytosis, type 2, 616649
SPTBN2	100%	100%	100%	99.9%	99.3%	Spinocerebellar ataxia 5, 600224;Spinocerebellar ataxia, autosomal recessive 14, 615386
SPTBN4	100%	100%	100%	99.9%	98.7%	Neurodevelopmental disorder with hypotonia, neuropathy, and deafness, 617519
SQOR	100%	100%	100%	100%	99.7%	Sulfide:quinone oxidoreductase deficiency, 619221
SQSTM1	100%	100%	100%	100%	98.9%	Neurodegeneration with ataxia, dystonia, and gaze palsy, childhood-onset, 617145;Frontotemporal dementia and/or amyotrophic lateral sclerosis 3, 616437;Myopathy, distal, with rimmed vacuoles, 617158;Paget disease of bone 3, 167250
SRD5A2	87.5%	87.5%	100%	100%	99.3%	Pseudovaginal perineoscrotal hypospadias, 264600
SRD5A3	100%	100%	100%	100%	99.9%	Kahrizi syndrome, 612713;Congenital disorder of glycosylation, type Iq, 612379
ST14	100%	100%	100%	100%	99.5%	Ichthyosis, congenital, autosomal recessive 11, 602400

ST3GAL3	89.5%	88.2%	100%	100%	99.5%	Developmental and epileptic encephalopathy 15, 615006;Intellectual developmental disorder, autosomal recessive 12, 611090
ST3GAL5	98.4%	98.4%	100%	100%	99.6%	Salt and pepper developmental regression syndrome, 609056
STAC3	100%	100%	100%	100%	99.2%	Congenital myopathy 13, 255995
STAMBP	96.3%	96.3%	100%	100%	99.8%	Microcephaly-capillary malformation syndrome, 614261
STAR	100%	100%	100%	100%	99.4%	Lipoid adrenal hyperplasia, 201710
STAT1	96%	96%	100%	100%	99.8%	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%	100%	100%	100%	99.5%	Pseudo-TORCH syndrome 3, 618886;Immunodeficiency 44, 616636
STAT5B	100%	100%	100%	100%	99.5%	Growth hormone insensitivity with immune dysregulation 1, autosomal recessive, 245590;Growth hormone insensitivity with immune dysregulation 2, autosomal dominant, 618985

STIL	100%	100%	100%	100%	99.8%	Microcephaly 7, primary, autosomal recessive, 612703
STIM1	100%	99.9%	100%	99.9%	99.7%	Myopathy, tubular aggregate, 1, 160565;Stormorken syndrome, 185070;Immunodeficiency 10, 612783
STK4	100%	100%	100%	100%	100%	T-cell immunodeficiency, recurrent infections, autoimmunity, and cardiac malformations, 614868
STN1	87.1%	87.1%	100%	100%	99.6%	Cerebroretinal microangiopathy with calcifications and cysts 2, 617341
STRA6	100%	100%	100%	100%	99.3%	Microphthalmia, syndromic 9, 601186;Microphthalmia, isolated, with coloboma 8, 601186
STRADA	100%	100%	100%	99.9%	99.4%	Polyhydramnios, megalencephaly, and symptomatic epilepsy, 611087
STRC	100%	100%	100%	99.9%	99.2%	Deafness, autosomal recessive 16, 603720
STT3A	99.9%	98.8%	100%	100%	99.8%	Congenital disorder of glycosylation, type Iw, autosomal dominant, 619714;Congenital disorder of glycosylation, type Iw, autosomal recessive, 615596
STT3B	100%	100%	100%	100%	99.5%	Congenital disorder of glycosylation, type Ix, 615597
STUB1	100%	100%	100%	100%	99.4%	Spinocerebellar ataxia 48, 618093;Spinocerebellar ataxia, autosomal recessive 16, 615768

STX11	100%	100%	100%	100%	99.6%	Hemophagocytic lymphohistiocytosis, familial, 4, 603552
STX3	100%	100%	100%	100%	99.7%	Retinal dystrophy and microvillus inclusion disease, 619446;Diarrhea 12, with microvillus atrophy, 619445
STXBP1	100%	100%	100%	100%	99.5%	Developmental and epileptic encephalopathy 4, 612164
STXBP2	100%	100%	100%	99.9%	99.3%	Hemophagocytic lymphohistiocytosis, familial, 5, with or without microvillus inclusion disease, 613101
SUCLA2	96.3%	96.3%	100%	100%	99.9%	Mitochondrial DNA depletion syndrome 5 (encephalomyopathic with or without methylmalonic aciduria), 612073
SUCLG1	100%	100%	100%	100%	99.8%	Mitochondrial DNA depletion syndrome 9 (encephalomyopathic type with methylmalonic aciduria), 245400
SUFU	100%	100%	100%	100%	99.6%	{Meningioma, familial, susceptibility to}, 607174;Joubert syndrome 32, 617757;Basal cell nevus syndrome 2, 620343;{Medulloblastoma}, 155255
SULT2B1	100%	100%	100%	100%	98.7%	Ichthyosis, congenital, autosomal recessive 14, 617571
SUMF1	100%	100%	100%	100%	99.9%	Multiple sulfatase deficiency, 272200
SUOX	100%	100%	100%	100%	99.5%	Sulfite oxidase deficiency, 272300

SUPT7L	100%	100%	100%	100%	99.9%	Fischer-Zirnsak progeroid syndrome, 621130
SURF1	100%	100%	100%	100%	99.4%	Charcot-Marie-Tooth disease, type 4K, 616684;Mitochondrial complex IV deficiency, nuclear type 1, 220110
SV2A	99.8%	98.2%	100%	100%	99.6%	Developmental and epileptic encephalopathy 113, 620772
SVBP	100%	100%	100%	100%	99.9%	Neurodevelopmental disorder with ataxia, hypotonia, and microcephaly, 618569
SVIL	100%	100%	100%	100%	99.4%	Myofibrillar myopathy 10, 619040
SYNE1	100%	100%	100%	100%	99.8%	Arthrogryposis multiplex congenita 3, myogenic type, 618484;Emery-Dreifuss muscular dystrophy 4, autosomal dominant, 612998;Spinocerebellar ataxia, autosomal recessive 8, 610743
SYNE4	100%	99.7%	100%	100%	99.7%	Deafness, autosomal recessive 76, 615540
SYNJ1	100%	100%	100%	100%	99.8%	Parkinson disease 20, early-onset, 615530;Developmental and epileptic encephalopathy 53, 617389
SYT14	100%	100%	100%	100%	99.7%	?Spinocerebellar ataxia, autosomal recessive 11, 614229

SYT2	100%	100%	100%	100%	99.4%	Myasthenic syndrome, congenital, 7A, presynaptic, and distal motor neuropathy, autosomal dominant, 616040;Myasthenic syndrome, congenital, 7B, presynaptic, autosomal recessive, 619461
SZT2	100%	100%	100%	100%	99.6%	Developmental and epileptic encephalopathy 18, 615476
TAC3	100%	100%	100%	100%	99.4%	Hypogonadotropic hypogonadism 10 with or without anosmia, 614839
TACO1	100%	100%	100%	99.8%	99.3%	Mitochondrial complex IV deficiency, nuclear type 8, 619052
TACR3	100%	100%	100%	100%	99%	Hypogonadotropic hypogonadism 11 with or without anosmia, 614840
TACSTD2	100%	100%	100%	99.6%	97.6%	Corneal dystrophy, gelatinous drop-like, 204870
TAF13	85.1%	77.2%	100%	100%	99.8%	Intellectual developmental disorder, autosomal recessive 60, 617432
TAF1C	100%	99.2%	100%	100%	99.4%	
TAF2	96.3%	96.3%	100%	100%	99.8%	Intellectual developmental disorder, autosomal recessive 40, 615599
TAF6	100%	100%	100%	100%	99.3%	Alazami-Yuan syndrome, 617126

TAF8	85.3%	85.3%	100%	100%	99.3%	Neurodevelopmental disorder with severe motor impairment, absent language, cerebral hypomyelination, and brain atrophy, 619972
TALDO1	89.1%	89.1%	100%	100%	99.7%	Transaldolase deficiency, 606003
TAMM41	100%	100%	100%	100%	99.6%	Combined oxidative phosphorylation deficiency 56, 620139
TANGO2	100%	100%	100%	100%	99.6%	Metabolic encephalomyopathic crises, recurrent, with rhabdomyolysis, cardiac arrhythmias, and neurodegeneration, 616878
TAP1	99.7%	97.9%	100%	100%	99.7%	MHC class I deficiency 1, 604571
TAP2	97.9%	97.9%	100%	99.9%	99%	MHC class I deficiency 2, 620813
TAPBP	88.8%	88.8%	100%	99.8%	98%	?MHC class I deficiency 3, 620814
TAPT1	100%	100%	100%	100%	99.7%	Osteochondrodysplasia, complex lethal, Symoens-Barnes-Gistelinck type, 616897
TARS1	100%	100%	100%	100%	99.9%	Trichothiodystrophy 7, nonphotosensitive, 618546
TARS2	100%	100%	100%	100%	99.8%	Combined oxidative phosphorylation deficiency 21, 615918
TASP1	93.2%	93.2%	100%	100%	99.8%	Suleiman-El-Hattab syndrome, 618950
TAT	100%	100%	100%	100%	99.8%	Tyrosinemia, type II, 276600
TBC1D20	92.2%	92.2%	100%	100%	99.7%	Warburg micro syndrome 4, 615663

TBC1D23	96.3%	96.3%	100%	100%	99.8%	Pontocerebellar hypoplasia, type 11, 617695
TBC1D24	100%	100%	100%	100%	99.4%	Deafness, autosomal recessive 86, 614617;Epilepsy, rolandic, with paroxysmal exercise-induce dystonia and writer's cramp, 608105;Myoclonic epilepsy, infantile, familial, 605021;Deafness, autosomal dominant 65, 616044;Developmental and epileptic encephalopathy 16, 615338;DOORS syndrome, 220500
TBC1D2B	100%	100%	100%	100%	99.5%	Neurodevelopmental disorder with seizures and gingival overgrowth, 619323
TBC1D32	100%	100%	100%	100%	99.8%	Orofaciodigital syndrome IX, 258865;Alsahan-Harris syndrome, 621307;Retinitis pigmentosa 100, 621280
TBC1D7	88.9%	88.9%	100%	100%	99.4%	Macrocephaly/megalencephaly syndrome, autosomal recessive, 248000
TBCB	77.5%	77.5%	100%	100%	99.4%	Neurodevelopmental disorder with behavioral abnormalities and childhood onset spastic paraplegia, 621382
TBCD	91.2%	90.7%	100%	99.9%	99.2%	Encephalopathy, progressive, early-onset, with brain atrophy and thin corpus callosum, 617193

TBCE	96.7%	96.6%	100%	100%	99.6%	Kenny-Caffey syndrome, type 1, 244460;Hypoparathyroidism-retardation-dysmorphism syndrome, 241410;Encephalopathy, progressive, with amyotrophy and optic atrophy, 617207
TBCK	100%	100%	100%	100%	99.9%	Hypotonia, infantile, with psychomotor retardation and characteristic facies 3, 616900
TBK1	100%	100%	100%	100%	99.9%	{Encephalopathy, acute, infection-induced (herpes-specific), susceptibility to, 8}, 617900;Frontotemporal dementia and/or amyotrophic lateral sclerosis 4, 616439;Autoinflammation with arthritis and vasculitis, 620880
TBX15	100%	100%	100%	100%	99.8%	Cousin syndrome, 260660
TBX19	100%	100%	100%	100%	99.6%	Adrenocorticotropic hormone deficiency, 201400
TBX4	100%	100%	100%	100%	99%	Ischiocoxopodopatellar syndrome with or without pulmonary arterial hypertension, 147891;Amelia, posterior, with pelvic and pulmonary hypoplasia syndrome, 601360
TBX6	100%	99.5%	100%	100%	99.3%	Spondylocostal dysostosis 5, 122600
TBXAS1	100%	100%	100%	100%	99.8%	Ghosal hematodiaphyseal syndrome, 231095

TBXT	100%	100%	100%	100%	99.6%	Sacral agenesis with vertebral anomalies, 615709;{Neural tube defects, susceptibility to}, 182940
TCAP	100%	100%	100%	100%	99.7%	Cardiomyopathy, hypertrophic, 25, 607487;Muscular dystrophy, limb-girdle, autosomal recessive 7, 601954
TCF12	96.1%	96.1%	100%	100%	99.7%	Craniosynostosis 3, 615314;Hypogonadotropic hypogonadism 26 with or without anosmia, 619718
TCF3	100%	100%	100%	100%	99.3%	Agammaglobulinemia 8B, autosomal recessive, 619824;Agammaglobulinemia 8A, autosomal dominant, 616941
TCIRG1	100%	100%	100%	100%	99.7%	Osteopetrosis, autosomal recessive 1, 259700
TCN2	91.8%	87.7%	100%	100%	99.9%	Transcobalamin II deficiency, 275350
TCTN1	92.6%	90.8%	100%	100%	99.5%	Joubert syndrome 13, 614173
TCTN2	98.5%	98.5%	100%	100%	99.5%	Joubert syndrome 24, 616654;?Meckel syndrome 8, 613885
TCTN3	100%	100%	100%	100%	99.5%	Joubert syndrome 18, 614815;Orofaciodigital syndrome IV, 258860
TDP1	87.9%	87.5%	100%	100%	99.8%	?Spinocerebellar ataxia, autosomal recessive, with axonal neuropathy 1, 607250
TDP2	100%	100%	100%	100%	99.8%	Spinocerebellar ataxia, autosomal recessive 23, 616949
TDRD7	100%	100%	100%	100%	99.8%	Cataract 36, 613887

TECPR2	100%	100%	100%	99.9%	99.6%	Neuropathy, hereditary sensory and autonomic, type IX, with developmental delay, 615031
TECR	87.3%	87.3%	100%	100%	98.9%	Intellectual developmental disorder, autosomal recessive 14, 614020
TECRL	78%	78%	100%	100%	99.7%	Ventricular tachycardia, catecholaminergic polymorphic, 3, 614021
TECTA	100%	100%	100%	100%	99.7%	Deafness, autosomal dominant 8/12, 601543;Deafness, autosomal recessive 21, 603629
TEFM	100%	100%	100%	100%	99.8%	Combined oxidative phosphorylation deficiency 58, 620451
TELO2	94.4%	94.3%	100%	99.9%	99.4%	You-Hoover-Fong syndrome, 616954
TENM3	99.6%	99.6%	100%	100%	99.6%	Microphthalmia, syndromic 15, 615145;?Microphthalmia/coloboma 9, 615145
TENT5A	100%	100%	100%	100%	99.4%	Osteogenesis imperfecta, type XVIII, 617952
TET2	100%	100%	100%	100%	99.9%	Myelodysplastic syndrome, somatic, 614286;Immunodeficiency 75, 619126
TET3	100%	100%	100%	100%	98.9%	Beck-Fahrner syndrome, 618798
TF	100%	100%	100%	100%	99.5%	Atransferrinemia, 209300
TFAM	100%	100%	100%	99.9%	99.4%	?Mitochondrial DNA depletion syndrome 15 (hepatocerebral type), 617156

TFG	100%	100%	100%	100%	99.4%	?Spastic paraplegia 57, autosomal recessive, 615658;Hereditary motor and sensory neuropathy, Okinawa type, 604484
TFR2	95.8%	94.2%	100%	100%	99%	Hemochromatosis, type 3, 604250
TFRC	94.7%	94.7%	100%	99.9%	99.4%	Immunodeficiency 46, 616740
TG	100%	100%	100%	99.9%	99.2%	{Autoimmune thyroid disease, susceptibility to, 3}, 608175;Thyroid dyshormonogenesis 3, 274700
TGDS	100%	100%	100%	100%	99.9%	Catel-Manzke syndrome, 616145
TGFB1	100%	100%	100%	99.9%	98.5%	Inflammatory bowel disease, immunodeficiency, and encephalopathy, 618213;{Cystic fibrosis lung disease, modifier of}, 219700;Camurati-Engelmann disease 1, 131300
TGM1	100%	100%	100%	100%	99.6%	Ichthyosis, congenital, autosomal recessive 1, 242300
TGM5	100%	100%	100%	100%	99.3%	Peeling skin syndrome 2, 609796
TH	100%	99.8%	100%	100%	99.2%	Segawa syndrome, recessive, 605407
THG1L	100%	100%	100%	99.7%	99.3%	Spinocerebellar ataxia, autosomal recessive 28, 618800
THOC6	100%	100%	100%	100%	99.5%	Beaulieu-Boycott-Innes syndrome, 613680

THPO	100%	100%	100%	100%	99.3%	Thrombocythemia 1, 187950;Thrombocytopenia 9, 620478;Amegakaryocytic thrombocytopenia, congenital, 2, 620481
THRB	100%	100%	100%	100%	99.9%	Thyroid hormone resistance, autosomal recessive, 274300;Thyroid hormone resistance, 188570;Thyroid hormone resistance, selective pituitary, 145650
THSD1	100%	100%	100%	100%	99.6%	?Aneurysm, intracranial berry, 12, 618734;Lymphatic malformation 13, 620244
THUMPD1	100%	100%	100%	99.8%	99.4%	Neurodevelopmental disorder with speech delay and variable ocular anomalies, 619989
TIAM1	98.3%	98.3%	100%	100%	99.5%	Neurodevelopmental disorder with language delay and seizures, 619908
TIMM50	100%	100%	100%	100%	99.6%	3-methylglutaconic aciduria, type IX, 617698
TIMMDC1	100%	100%	100%	100%	99.4%	Mitochondrial complex I deficiency, nuclear type 31, 618251
TJP2	97.1%	95.8%	100%	100%	99.8%	Hypercholanemia, familial 1, 607748;Cholestasis, progressive familial intrahepatic 4, 615878

TK2	100%	100%	100%	100%	99.9%	Mitochondrial DNA depletion syndrome 2 (myopathic type), 609560;?Progressive external ophthalmoplegia with mitochondrial DNA deletions, autosomal recessive 3, 617069
TKT	89%	89%	100%	100%	99.5%	Short stature, developmental delay, and congenital heart defects, 617044
TLCD3B	100%	100%	100%	99.8%	99%	Cone-rod dystrophy 22, 619531
TLE6	100%	100%	100%	100%	99.4%	Oocyte/zygote/embryo maturation arrest 15, 616814
TLR1	100%	100%	100%	100%	100%	{Leprosy, susceptibility to, 5}, 613223;{Leprosy, protection against}, 613223
TM2D3	100%	100%	100%	100%	99.6%	Neurocardiorenal malformation syndrome, 621379
TMC1	100%	100%	100%	100%	99.7%	Deafness, autosomal dominant 36, 606705;Deafness, autosomal recessive 7, 600974
TMC6	94.9%	94.4%	100%	99.9%	99.1%	{Epidermodysplasia verruciformis, susceptibility to, 1}, 226400
TMC8	100%	100%	100%	100%	99.4%	{Epidermodysplasia verruciformis, susceptibility to, 2}, 618231
TMCO1	72.3%	72.3%	100%	100%	99.6%	Craniofacial dysmorphism, skeletal anomalies, and impaired intellectual development 1, 213980

TMEM107	100%	100%	100%	100%	99.2%	Orofaciodigital syndrome XVI, 617563;Meckel syndrome 13, 617562;?Joubert syndrome 29, 617562
TMEM126A	100%	100%	100%	100%	100%	Optic atrophy 7, 612989
TMEM126B	100%	100%	100%	100%	99.9%	Mitochondrial complex I deficiency, nuclear type 29, 618250
TMEM132E	100%	100%	100%	100%	99.6%	Deafness, autosomal recessive 99, 618481
TMEM138	100%	98.8%	100%	99.9%	99.7%	Joubert syndrome 16, 614465
TMEM147	100%	100%	100%	100%	99.6%	Neurodevelopmental disorder with facial dysmorphism, absent language, and pseudo-Pelger-Huet anomaly, 620075
TMEM165	88.9%	88.9%	100%	100%	99.7%	Congenital disorder of glycosylation, type IIk, 614727
TMEM216	100%	100%	100%	100%	99.8%	Joubert syndrome 2, 608091;Retinitis pigmentosa 98, 620996;Meckel syndrome 2, 603194
TMEM218	100%	100%	100%	100%	99.6%	Joubert syndrome 39, 619562
TMEM222	100%	100%	100%	100%	99.7%	Neurodevelopmental disorder with motor and speech delay and behavioral abnormalities, 619470
TMEM231	93.5%	93.5%	100%	100%	99.5%	Joubert syndrome 20, 614970;Meckel syndrome 11, 615397
TMEM237	98.2%	98.2%	100%	100%	99.7%	Joubert syndrome 14, 614424

TMEM260	94.8%	94.8%	100%	100%	99.7%	Structural heart defects and renal anomalies syndrome, 617478
TMEM38B	89.5%	89.5%	100%	100%	99.9%	Osteogenesis imperfecta, type XIV, 615066
TMEM53	100%	100%	100%	100%	99.3%	Craniotubular dysplasia, Ikegawa type, 619727
TMEM63B	100%	100%	100%	100%	99.5%	Developmental and epileptic encephalopathy 118, 621250
TMEM63C	100%	100%	100%	100%	99.1%	Spastic paraplegia 87, autosomal recessive, 619966
TMEM67	96.1%	96.1%	100%	100%	99.7%	Nephronophthisis 11, 613550;{Bardet-Biedl syndrome 14, modifier of}, 615991;Joubert syndrome 6, 610688;Meckel syndrome 3, 607361;?RHYNS syndrome, 602152;COACH syndrome 1, 216360
TMEM70	100%	100%	100%	100%	99.3%	Mitochondrial complex V (ATP synthase) deficiency, nuclear type 2, 614052
TMEM94	100%	100%	100%	100%	99.6%	Intellectual developmental disorder with cardiac defects and dysmorphic facies, 618316
TMIE	100%	100%	100%	99.9%	99.4%	Deafness, autosomal recessive 6, 600971
TMPRSS15	100%	100%	100%	100%	99.7%	Enterokinase deficiency, 226200
TMPRSS3	100%	100%	100%	100%	99.5%	Deafness, autosomal recessive 8/10, 601072

TMPRSS6	100%	99.9%	100%	100%	99.5%	Iron-refractory iron deficiency anemia, 206200
TMTC3	100%	100%	100%	100%	99.7%	Lissencephaly 8, 617255
TMX2	99.8%	97%	100%	100%	99.7%	Neurodevelopmental disorder with microcephaly, cortical malformations, and spasticity, 618730
TNFRSF11A	100%	100%	100%	100%	99.5%	Osteopetrosis, autosomal recessive 7, 612301;{Paget disease of bone 2, early-onset}, 602080;Osteolysis, familial expansile, 174810
TNFRSF11B	89.8%	89.8%	100%	100%	99.9%	Paget disease of bone 5, juvenile-onset, 239000
TNFRSF13B	100%	100%	100%	100%	99%	Immunodeficiency, common variable, 2, 240500;Immunoglobulin A deficiency 2, 609529
TNFRSF13C	100%	100%	100%	99.9%	98.5%	Immunodeficiency, common variable, 4, 613494
TNFRSF4	100%	99.8%	100%	99.7%	98.9%	?Immunodeficiency 16, 615593
TNFRSF9	100%	100%	100%	100%	99.9%	Immunodeficiency 109 with lymphoproliferation, 620282
TNFSF11	100%	100%	100%	100%	99.9%	Osteopetrosis, autosomal recessive 2, 259710
TNIK	98.6%	98.6%	100%	100%	99.6%	Intellectual developmental disorder, autosomal recessive 54, 617028

TNNI3	100%	100%	100%	100%	98.2%	?Cardiomyopathy, dilated, 2A, 611880;Cardiomyopathy, hypertrophic, 7, 613690;Cardiomyopathy, familial restrictive, 1, 115210;Cardiomyopathy, dilated, 1FF, 613286
TNNT1	99.9%	97.6%	100%	99.9%	98.5%	Nemaline myopathy 5C, autosomal dominant, 620389;Nemaline myopathy 5A, autosomal recessive, severe infantile, 605355;Nemaline myopathy 5B, autosomal recessive, childhood-onset, 620386
TNR	100%	100%	100%	99.9%	99.5%	Neurodevelopmental disorder, nonprogressive, with spasticity and transient opisthotonus, 619653
TNXB	100%	100%	100%	99.9%	98.8%	Ehlers-Danlos syndrome, classic-like, 1, 606408;Vesicoureteral reflux 8, 615963
TOE1	100%	100%	100%	99.9%	99.4%	Pontocerebellar hypoplasia, type 7, 614969
TOGARAM1	100%	100%	100%	100%	99.7%	Joubert syndrome 37, 619185
TOMM7	100%	100%	100%	100%	99.6%	Garg-Mishra progeroid syndrome, 620601
TONSL	96.1%	96.1%	100%	99.9%	99.3%	Spondyloepimetaphyseal dysplasia, sponastrime type, 271510

TOP3A	95.7%	95%	100%	100%	99.5%	Microcephaly, growth restriction, and increased sister chromatid exchange 2, 618097;Progressive external ophthalmoplegia with mitochondrial DNA deletions, autosomal recessive 5, 618098
TOR1A	93.1%	90.7%	100%	99.9%	99.4%	{Dystonia-1, modifier of};Arthrogryposis multiplex congenita 5, 618947;Dystonia-1, torsion, 128100
TOR1AIP1	100%	100%	100%	100%	99.8%	?Muscular dystrophy, autosomal recessive, with rigid spine and distal joint contractures, 617072
TP53RK	100%	100%	100%	100%	99.8%	Galloway-Mowat syndrome 4, 617730
TP73	100%	100%	100%	100%	99.5%	Ciliary dyskinesia, primary, 47, and lissencephaly, 619466
TPI1	100%	100%	100%	100%	99.6%	Hemolytic anemia due to triosephosphate isomerase deficiency, 615512
TPK1	93.3%	93.3%	100%	100%	99.9%	Thiamine metabolism dysfunction syndrome 5 (episodic encephalopathy type), 614458
TPM3	100%	100%	100%	99.9%	99.8%	Congenital myopathy 4A, autosomal dominant, 255310;Congenital myopathy 4B, autosomal recessive, 609284
TPO	100%	100%	100%	100%	99.5%	Thyroid dyshormonogenesis 2A, 274500

TPP1	100%	100%	100%	100%	99.3%	Ceroid lipofuscinosis, neuronal, 2, 204500;Spinocerebellar ataxia, autosomal recessive 7, 609270
TPP2	100%	100%	100%	100%	99.9%	Immunodeficiency 78 with autoimmunity and developmental delay, 619220
TPRKB	83.2%	81.2%	100%	100%	99.8%	Galloway-Mowat syndrome 5, 617731
TPRN	97.4%	95.3%	100%	99.7%	96.6%	Deafness, autosomal recessive 79, 613307
TRAC	100%	100%	100%	100%	99.7%	
TRAF3IP1	100%	100%	100%	100%	99.6%	
TRAIP	100%	100%	100%	100%	99.8%	Seckel syndrome 9, 616777
TRAK1	100%	100%	100%	100%	99.7%	Developmental and epileptic encephalopathy 68, 618201
TRAPPC10	97.6%	97.6%	100%	99.9%	99.4%	Neurodevelopmental disorder with microcephaly, short stature, and speech delay, 620027
TRAPPC11	100%	100%	100%	100%	99.8%	Muscular dystrophy, limb-girdle, autosomal recessive 18, 615356
TRAPPC12	86.3%	86.3%	100%	99.9%	99%	Encephalopathy, progressive, early-onset, with brain atrophy and spasticity, 617669
TRAPPC14	100%	100%	100%	100%	99.5%	?Microcephaly 25, primary, autosomal recessive, 618351
TRAPPC2L	100%	100%	100%	100%	99.6%	Encephalopathy, progressive, early-onset, with episodic rhabdomyolysis, 618331

TRAPPC4	100%	100%	100%	99.9%	99.3%	Neurodevelopmental disorder with epilepsy, spasticity, and brain atrophy, 618741
TRAPPC6B	100%	100%	100%	100%	99.8%	Neurodevelopmental disorder with microcephaly, epilepsy, and brain atrophy, 617862
TRAPPC9	100%	100%	100%	100%	99.5%	Intellectual developmental disorder, autosomal recessive 13, 613192
TRDN	92.7%	92.7%	100%	100%	99.8%	Cardiac arrhythmia syndrome, with or without skeletal muscle weakness, 615441
TREM2	100%	100%	100%	100%	99.9%	{Alzheimer disease 17, susceptibility to}, 615080;Polycystic lipomembranous osteodysplasia with sclerosing leukoencephalopathy 2, 618193
TREX1	100%	100%	100%	100%	99.5%	Vasculopathy, retinal, with cerebral leukoencephalopathy and systemic manifestations, 192315;Aicardi-Goutieres syndrome 1, dominant and recessive, 225750;{Systemic lupus erythematosus, susceptibility to}, 152700;Chilblain lupus, 610448
TRH	100%	100%	100%	100%	99.5%	Thyrotropin-releasing hormone deficiency, 275120
TRHR	100%	100%	100%	100%	99.9%	Hypothyroidism, congenital, nongoitrous, 7, 618573

TRIM2	96.1%	96.1%	100%	100%	99.7%	Charcot-Marie-Tooth disease, type 2R, 615490
TRIM32	100%	100%	100%	100%	99.6%	?Bardet-Biedl syndrome 11, 615988;Muscular dystrophy, limb-girdle, autosomal recessive 8, 254110
TRIM37	98.3%	97.6%	100%	100%	99.7%	Mulibrey nanism, 253250
TRIM63	100%	100%	100%	100%	99.7%	Cardiomyopathy, familial hypertrophic, 31, 621270
TRIOBP	100%	100%	100%	99.9%	99.1%	Deafness, autosomal recessive 28, 609823
TRIP11	100%	100%	100%	100%	99.8%	Odontochondrodysplasia 1, 184260;Achondrogenesis, type IA, 200600
TRIP13	100%	100%	100%	100%	99.7%	Oocyte/zygote/embryo maturation arrest 9, 619011;Mosaic variegated aneuploidy syndrome 3, 617598
TRIP4	95.9%	94.9%	100%	100%	99.7%	?Muscular dystrophy, congenital, Davignon-Chauveau type, 617066;Spinal muscular atrophy with congenital bone fractures 1, 616866
TRIT1	100%	100%	100%	100%	99.8%	Combined oxidative phosphorylation deficiency 35, 617873
TRMT1	100%	100%	100%	100%	99%	Intellectual developmental disorder, autosomal recessive 68, 618302
TRMT10A	100%	100%	100%	100%	99.7%	Microcephaly, short stature, and impaired glucose metabolism 1, 616033

TRMT10C	100%	100%	100%	100%	99.8%	Combined oxidative phosphorylation deficiency 30, 616974
TRMT5	100%	100%	100%	100%	99.8%	Peripheral neuropathy with variable spasticity, exercise intolerance, and developmental delay, 616539
TRMU	88%	88%	100%	100%	99.4%	{Deafness, mitochondrial, modifier of}, 580000;Liver failure, transient infantile, 613070
TRNT1	91.9%	91.9%	100%	100%	99.9%	Sideroblastic anemia with B-cell immunodeficiency, periodic fevers, and developmental delay, 616084;Retinitis pigmentosa and erythrocytic microcytosis, 616959
TRPM1	100%	100%	100%	100%	99.8%	Night blindness, congenital stationary (complete), 1C, autosomal recessive, 613216
TRPM6	100%	100%	100%	100%	99.8%	Hypomagnesemia 1, intestinal, 602014
TRPV6	100%	100%	100%	100%	99.2%	Hyperparathyroidism, transient neonatal, 618188
TSEN15	100%	100%	100%	100%	99.8%	Pontocerebellar hypoplasia, type 2F, 617026
TSEN2	88.4%	88.4%	100%	100%	99.6%	Pontocerebellar hypoplasia type 2B, 612389
TSEN34	100%	100%	100%	100%	98.8%	?Pontocerebellar hypoplasia type 2C, 612390

TSEN54	100%	100%	100%	100%	99.7%	Pontocerebellar hypoplasia type 2A, 277470;Pontocerebellar hypoplasia type 4, 225753;?Pontocerebellar hypoplasia type 5, 610204
TSFM	91.5%	91.1%	100%	100%	99.3%	Combined oxidative phosphorylation deficiency 3, 610505
TSHB	100%	100%	100%	100%	99.7%	Hypothyroidism, congenital, nongoitrous 4, 275100
TSHR	100%	100%	100%	100%	99.7%	Hyperthyroidism, familial gestational, 603373;Hyperthyroidism, nonautoimmune, 609152;Thyroid adenoma, hyperfunctioning, somatic, 609152;Hypothyroidism, congenital, nongoitrous, 1, 275200;Thyroid carcinoma with thyrotoxicosis, somatic, 609152
TSPAN12	100%	100%	100%	100%	99.7%	Exudative vitreoretinopathy 5, 613310
TSPEAR	93.9%	93.9%	100%	100%	99.1%	Tooth agenesis, selective, 10, 620173;?Deafness, autosomal recessive 98, 614861;Ectodermal dysplasia 14, hypohidrotic/hair/tooth/nail type, 618180
TSPOAP1	100%	100%	100%	100%	99.5%	Dystonia 22, juvenile-onset, 620453;?Dystonia 22, adult-onset, 620456
TSPYL1	100%	100%	100%	100%	99.1%	Sudden infant death with dysgenesis of the testes syndrome, 608800

TTC12	100%	100%	100%	100%	99.8%	Ciliary dyskinesia, primary, 45, 618801
TTC19	100%	100%	100%	100%	99.7%	Mitochondrial complex III deficiency, nuclear type 2, 615157
TTC21B	98.4%	98.1%	100%	100%	99.8%	Short-rib thoracic dysplasia 4 with or without polydactyly, 613819;Nephronophthisis 12, 613820
TTC5	100%	100%	100%	100%	99.8%	Neurodevelopmental disorder with cerebral atrophy and variable facial dysmorphism, 619244
TTC7A	100%	99.8%	100%	100%	99.6%	Gastrointestinal defects and immunodeficiency syndrome, 243150
TTC8	91.4%	91.4%	100%	100%	99.9%	Bardet-Biedl syndrome 8, 615985;?Retinitis pigmentosa 51, 613464
TTI1	100%	100%	100%	100%	99.7%	Neurodevelopmental disorder with microcephaly and movement abnormalities, 620445
TTI2	100%	100%	100%	100%	99.7%	Intellectual developmental disorder, autosomal recessive 39, 615541
TTLL5	100%	100%	100%	100%	99.7%	Cone-rod dystrophy 19, 615860
TTPA	87.1%	87.1%	100%	100%	99.7%	Ataxia with isolated vitamin E deficiency, 277460
TUB	100%	100%	100%	100%	99.5%	?Retinal dystrophy and obesity, 616188
TUBA8	95.3%	95.1%	100%	99.8%	99.2%	Macrothrombocytopenia, isolated, 2, autosomal dominant, 619840

TUBGCP2	97%	97%	100%	100%	99.6%	Pachygyria, microcephaly, developmental delay, and dysmorphic facies, with or without seizures, 618737
TUBGCP4	100%	100%	100%	99.8%	98.7%	Microcephaly and chorioretinopathy, autosomal recessive, 3, 616335
TUBGCP6	100%	100%	100%	99.9%	99.2%	Microcephaly and chorioretinopathy, autosomal recessive, 1, 251270
TUFM	99.7%	96.7%	100%	99.9%	99.4%	Combined oxidative phosphorylation deficiency 4, 610678
TULP1	100%	100%	100%	100%	99.5%	Leber congenital amaurosis 15, 613843;Retinitis pigmentosa 14, 600132
TULP3	93.4%	93.4%	100%	100%	99.5%	Hepatorenocardiac degenerative fibrosis, 619902
TUSC3	93.3%	93.3%	100%	100%	99.7%	Intellectual developmental disorder, autosomal recessive 7, 611093
TWIST2	100%	100%	100%	99.9%	96.6%	Ablepharon-macrostomia syndrome, 200110;Barber-Say syndrome, 209885;Focal facial dermal dysplasia 3, Setleis type, 227260
TWNK	100%	100%	100%	100%	99.6%	Mitochondrial DNA depletion syndrome 7 (hepatocerebral type), 271245;Progressive external ophthalmoplegia with mitochondrial DNA deletions, autosomal dominant 3, 609286;Perrault syndrome 5, 616138

TXNDC15	100%	98.5%	100%	99.9%	99.5%	Meckel syndrome 14, 619879
TXNL4A	100%	100%	100%	99.7%	99.5%	Burn-McKeown syndrome, 608572
TYK2	100%	99.8%	100%	99.9%	98.7%	Immunodeficiency 35, 611521
TYMP	100%	100%	100%	100%	99.4%	Mitochondrial DNA depletion syndrome 1 (MNGIE type), 603041
TYR	100%	100%	100%	100%	99.7%	[Skin/hair/eye pigmentation 3, light/dark/freckling skin], 601800;[Skin/hair/eye pigmentation 3, blue/green eyes], 601800;{Melanoma, cutaneous malignant, susceptibility to, 8}, 601800;Albinism, oculocutaneous, type IB, 606952;Albinism, oculocutaneous, type IA, 203100
TYROBP	91.4%	91.2%	100%	99.8%	99.2%	Polycystic lipomembranous osteodysplasia with sclerosing leukoencephalopathy 1, 221770
TYRP1	100%	100%	100%	100%	99.7%	[Skin/hair/eye pigmentation, variation in, 11 (Melanesian blond hair)], 612271;Albinism, oculocutaneous, type III, 203290
UBA5	94.1%	92.2%	100%	100%	99.8%	?Spinocerebellar ataxia, autosomal recessive 24, 617133;Developmental and epileptic encephalopathy 44, 617132
UBAP1L	100%	100%	100%	100%	99.5%	Retinal dystrophy, Zeitz-Han type, 621558

UBE2T	92.6%	92.6%	100%	100%	99.7%	Fanconi anemia, complementation group T, 616435
UBE3B	100%	100%	100%	100%	99.8%	Kaufman oculocerebrofacial syndrome, 244450
UBE3C	94.8%	94.8%	100%	100%	99.6%	Neurodevelopmental disorder with absent speech and movement and behavioral abnormalities, 620270
UBE4A	100%	100%	100%	100%	99.7%	Neurodevelopmental disorder with hypotonia and gross motor and speech delay, 619639
UBR1	98%	97.9%	100%	100%	99.7%	Johanson-Blizzard syndrome, 243800
UBR7	100%	100%	100%	100%	99.4%	Li-Campeau syndrome, 619189
UCHL1	100%	100%	100%	100%	99.8%	{?Parkinson disease 5, susceptibility to}, 613643;Spastic paraplegia 79A, autosomal dominant, 620221;Spastic paraplegia 79B, autosomal recessive, 615491
UFC1	100%	100%	100%	99.9%	99.6%	Neurodevelopmental disorder with spasticity and poor growth, 618076
UFM1	100%	100%	100%	100%	99.8%	Leukodystrophy, hypomyelinating, 14, 617899
UFSP2	100%	100%	100%	100%	99.8%	?Hip dysplasia, Beukes type, 142669;Spondyloepimetaphyseal dysplasia, Di Rocco type, 617974;Developmental and epileptic encephalopathy 106, 620028

UGDH	97%	94%	100%	100%	99.7%	Developmental and epileptic encephalopathy 84, 618792
UGGT1	100%	100%	100%	100%	99.8%	Congenital disorder of glycosylation, type IICC, 621381
UGP2	91.8%	90.6%	100%	100%	99.8%	Developmental and epileptic encephalopathy 83, 618744
UGT1A1	100%	100%	100%	100%	99.6%	Crigler-Najjar syndrome, type I, 218800;[Bilirubin, serum level of, QTL1], 601816;Hyperbilirubinemia, familial transient neonatal, 237900;Crigler-Najjar syndrome, type II, 606785;[Gilbert syndrome], 143500
UMPS	90.6%	90.6%	100%	100%	99.6%	Orotic aciduria, 258900
UNC13A	100%	100%	100%	99.9%	99%	?Intellectual development disorder with seizures and dysmorphic facies, 621457;Neurodevelopmental disorder with speech delay, movement abnormalities, and seizures, 621456;Neurodevelopmental disorder with hypotonia, epilepsy, and absent speech, 621455
UNC13D	100%	100%	100%	100%	99.5%	Hemophagocytic lymphohistiocytosis, familial, 3, 608898
UNC45A	100%	100%	100%	100%	99.4%	Osteootohepatoenteric syndrome, 619377
UNC45B	96.1%	95.9%	100%	100%	99.7%	?Cataract 43, 616279;Myofibrillar myopathy 11, 619178

UNC80	100%	100%	100%	100%	99.7%	Hypotonia, infantile, with psychomotor retardation and characteristic facies 2, 616801
UNG	96.5%	96.5%	100%	100%	99.3%	Immunodeficiency with hyper IgM, type 5, 608106
UPB1	94.3%	91.8%	100%	100%	99.5%	Beta-ureidopropionase deficiency, 613161
UQCC2	100%	100%	100%	100%	99.9%	Mitochondrial complex III deficiency, nuclear type 7, 615824
UQCC3	100%	100%	100%	100%	99.6%	?Mitochondrial complex III deficiency, nuclear type 9, 616111
UQCRB	100%	100%	100%	100%	99.8%	Mitochondrial complex III deficiency, nuclear type 3, 615158
UQCRC2	74.7%	74.7%	100%	100%	99.8%	Mitochondrial complex III deficiency, nuclear type 5, 615160
UQCRFS1	100%	100%	100%	100%	99.4%	Mitochondrial complex III deficiency, nuclear type 10, 618775
UQCRQ	100%	100%	100%	100%	99.7%	Mitochondrial complex III deficiency, nuclear type 4, 615159
UROC1	96.6%	96.6%	100%	100%	99.6%	?Urocanase deficiency, 276880
UROD	100%	100%	100%	100%	99.6%	Porphyria, hepatoerythropoietic, 176100;Porphyria cutanea tarda, 176100
UROS	95.3%	95.3%	100%	100%	99.9%	Porphyria, congenital erythropoietic, 263700
USB1	93.9%	93.2%	100%	100%	99.1%	Poikiloderma with neutropenia, 604173
USH1C	100%	100%	100%	100%	99.6%	Usher syndrome, type 1C, 276904;Deafness, autosomal recessive 18A, 602092

USH1G	100%	100%	100%	100%	99.1%	Usher syndrome, type 1G, 606943
USH2A	100%	100%	100%	100%	99.7%	Usher syndrome, type 2A, 276901;Retinitis pigmentosa 39, 613809
USP18	100%	100%	100%	100%	99.2%	Pseudo-TORCH syndrome 2, 617397
USP45	100%	100%	100%	100%	99.8%	?Leber congenital amaurosis 19, 618513
USP53	100%	100%	100%	100%	99.8%	Cholestasis, progressive familial intrahepatic, 7, with or without hearing loss, 619658
UVSSA	100%	100%	100%	100%	99.5%	UV-sensitive syndrome 3, 614640
VAC14	96.4%	96.4%	100%	100%	99.7%	Striatonigral degeneration, childhood-onset, 617054
VAMP1	100%	100%	100%	99.9%	98.9%	Myasthenic syndrome, congenital, 25, 618323;Spastic ataxia 1, autosomal dominant, 108600
VARS1	100%	100%	100%	100%	99.3%	Neurodevelopmental disorder with microcephaly, seizures, and cortical atrophy, 617802
VARS2	100%	100%	100%	100%	98.9%	Combined oxidative phosphorylation deficiency 20, 615917
VAX1	100%	100%	100%	100%	98.5%	?Microphthalmia, syndromic 11, 614402
VDR	100%	100%	100%	100%	99.1%	Rickets, vitamin D-resistant, type IIA, 277440

VHL	87.7%	87.7%	100%	99.9%	98.8%	Hemangioblastoma, cerebellar, somatic;Erythrocytosis, familial, 2, 263400;von Hippel-Lindau syndrome, 193300;Renal cell carcinoma, somatic, 144700;Pheochromocytoma, 171300
VIPAS39	99.8%	97.9%	100%	100%	99.7%	Arthrogryposis, renal dysfunction, and cholestasis 2, 613404
VKORC1	98.6%	93.5%	100%	100%	99.2%	Vitamin K-dependent clotting factors, combined deficiency of, 2, 607473;Warfarin resistance, 122700
VLDLR	100%	100%	100%	100%	99.8%	Cerebellar hypoplasia, impaired intellectual development, and dysequilibrium syndrome 1, 224050
VMA12	100%	100%	100%	99.9%	99.2%	Congenital disorder of glycosylation, type IIp, 616829
VMA22	100%	98.5%	100%	99.9%	97.9%	Congenital disorder of glycosylation, type IIo, 616828
VPS11	100%	100%	100%	100%	99.6%	?Dystonia 32, 619637;Leukodystrophy, hypomyelinating, 12, 616683
VPS13A	100%	100%	100%	100%	99.9%	Choreoacanthocytosis, 200150
VPS13B	100%	100%	100%	100%	99.7%	Cohen syndrome, 216550
VPS13C	100%	100%	100%	100%	99.8%	Parkinson disease 23, autosomal recessive, early onset, 616840
VPS13D	100%	100%	100%	100%	99.8%	Spinocerebellar ataxia, autosomal recessive 4, 607317

VPS33A	90.3%	90.2%	100%	100%	99.7%	Mucopolysaccharidosis-plus syndrome, 617303
VPS33B	100%	100%	100%	100%	99.9%	Keratoderma-ichthyosis-deafness syndrome, autosomal recessive, 620009;Cholestasis, progressive familial intrahepatic, 12, 620010;Arthrogryposis, renal dysfunction, and cholestasis 1, 208085
VPS35L	93.6%	93%	100%	100%	99.4%	Ritscher-Schinzel syndrome 3, 619135
VPS37A	92.3%	92.3%	100%	99.9%	99.5%	Spastic paraplegia 53, autosomal recessive, 614898
VPS41	98.4%	96.8%	100%	100%	99.9%	Spinocerebellar ataxia, autosomal recessive 29, 619389
VPS45	88.2%	87.2%	100%	100%	99.6%	Neutropenia, severe congenital, 5, autosomal recessive, 615285
VPS50	96.2%	95.3%	100%	100%	99.7%	Neurodevelopmental disorder with microcephaly, seizures, and neonatal cholestasis, 619685
VPS51	100%	100%	100%	100%	99%	Pontocerebellar hypoplasia, type 13, 618606
VPS53	83.1%	81.5%	100%	99.9%	99.4%	Pontocerebellar hypoplasia, type 2E, 615851
VRK1	99.6%	98%	100%	100%	99.9%	Pontocerebellar hypoplasia type 1A, 607596;Neuronopathy, distal hereditary motor, autosomal recessive 10, 620542
VSX2	100%	100%	100%	99.9%	99.1%	Microphthalmia, isolated 2, 610093;Microphthalmia/coloboma 3, 610092

VWA1	100%	100%	100%	100%	99.1%	Neuronopathy, distal hereditary motor, autosomal recessive 7, 619216
VWA3B	100%	100%	100%	100%	99.7%	?Spinocerebellar ataxia, autosomal recessive 22, 616948
VWF	100%	100%	100%	100%	99.5%	von Willebrand disease, type 1, 193400;von Willebrand disease, types 2A, 2B, 2M, and 2N, 613554;von Willebrand disease, type 3, 277480
WARS1	91.8%	91.8%	100%	100%	99.6%	Neuronopathy, distal hereditary motor, autosomal dominant 9, 617721;Neurodevelopmental disorder with microcephaly and speech delay, with or without brain abnormalities, 620317
WARS2	93.8%	93.8%	100%	100%	100%	Parkinsonism-dystonia 3, childhood-onset, 619738;Neurodevelopmental disorder, mitochondrial, with abnormal movements and lactic acidosis, with or without seizures, 617710
WASHC4	100%	100%	100%	100%	99.8%	Intellectual developmental disorder, autosomal recessive 43, 615817
WASHC5	100%	100%	100%	100%	99.8%	Ritscher-Schinzel syndrome 1, 220210;Spastic paraplegia 8, autosomal dominant, 603563
WBP2	100%	100%	100%	100%	99.3%	Deafness, autosomal recessive 107, 617639

WBP4	100%	100%	100%	100%	99.2%	Neurodevelopemental disorder with hypotonia, feeding difficulties, facial dysmorphism, and brain abnormalities, 620852
WDPCP	92.1%	92.1%	100%	100%	99.8%	Bardet-Biedl syndrome 15, 615992;Congenital heart defects, hamartomas of tongue, and polysyndactyly, 217085
WDR1	100%	100%	100%	100%	99.4%	Periodic fever, immunodeficiency, and thrombocytopenia syndrome, 150550
WDR11	100%	100%	100%	100%	99.9%	Intellectual developmental disorder, autosomal recessive 78, 620237;Hypogonadotropic hypogonadism 14 with or without anosmia, 614858
WDR19	100%	100%	100%	100%	99.8%	Nephronophthisis 13, 614377;Cranioectodermal dysplasia 4, 614378;Senior-Loken syndrome 8, 616307;Short-rib thoracic dysplasia 5 with or without polydactyly, 614376;?Spermatogenic failure 72, 619867
WDR35	100%	100%	100%	100%	99.7%	Short-rib thoracic dysplasia 7 with or without polydactyly, 614091;Cranioectodermal dysplasia 2, 613610
WDR4	100%	100%	100%	99.9%	99.3%	Galloway-Mowat syndrome 6, 618347;Microcephaly, growth deficiency, seizures, and brain malformations, 618346

WDR45B	89.6%	89.6%	100%	100%	99.6%	Neurodevelopmental disorder with spastic quadriplegia and brain abnormalities with or without seizures, 617977
WDR62	98.8%	98.8%	100%	100%	99.6%	Microcephaly 2, primary, autosomal recessive, with or without cortical malformations, 604317
WDR72	100%	100%	100%	100%	99.6%	Amelogenesis imperfecta, type IIA3, 613211
WDR73	100%	100%	100%	100%	99.3%	Galloway-Mowat syndrome 1, 251300
WDR81	100%	100%	100%	100%	99.5%	Cerebellar ataxia, impaired intellectual development, and dysquilibrium syndrome 2, 610185;Hydrocephalus, congenital, 3, with brain anomalies, 617967
WDR83OS	100%	100%	100%	100%	98.4%	Neurodevelopmental disorder with variable familial hypercholanemia, 621016
WFDC2	100%	100%	100%	100%	98.6%	Bronchiectasis and nasal polyposis, 620984
WFS1	91.5%	90.5%	100%	100%	99.4%	Deafness, autosomal dominant 6/14/38, 600965;?Cataract 41, 116400;Wolfram-like syndrome, autosomal dominant, 614296;{Diabetes mellitus, noninsulin-dependent, association with}, 125853;Wolfram syndrome 1, 222300

WHRN	100%	100%	100%	100%	99.4%	Deafness, autosomal recessive 31, 607084;Usher syndrome, type 2D, 611383
WIPF1	100%	100%	100%	100%	99.7%	Wiskott-Aldrich syndrome 2, 614493
WIPI2	100%	100%	100%	100%	99.5%	?Intellectual developmental disorder with short stature and variable skeletal anomalies, 618453
WLS	100%	100%	100%	100%	99.9%	Zaki syndrome, 619648
WNK1	100%	100%	100%	99.9%	99%	Neuropathy, hereditary sensory and autonomic, type II, 201300;Pseudohypoaldosteronism, type IIC, 614492
WNT1	100%	100%	100%	99.9%	99%	{Osteoporosis, early-onset, susceptibility to, autosomal dominant}, 615221;Osteogenesis imperfecta, type XV, 615220
WNT10A	79.3%	79.3%	100%	100%	99.7%	Schopf-Schulz-Passarge syndrome, 224750;Tooth agenesis, selective, 4, 150400;Ectodermal dysplasia 16 (odontoonychodermal dysplasia), 257980
WNT10B	100%	100%	100%	99.7%	98.8%	Tooth agenesis, selective, 8, 617073;Split-hand/foot malformation 6, 225300
WNT2B	96.4%	95.9%	100%	100%	99.8%	Diarrhea 9, 618168
WNT3	100%	100%	100%	99.8%	99.3%	?Tetra-amelia syndrome 1, 273395
WNT4	100%	100%	100%	100%	99.1%	Mullerian aplasia and hyperandrogenism, 158330;SERKAL syndrome, 611812

WNT7A	100%	100%	100%	100%	99.6%	Fuhrmann syndrome, 228930;Ulna and fibula, absence of, with severe limb deficiency, 276820;?Santos syndrome, 613005
WRAP53	100%	99.9%	100%	99.9%	99.2%	Dyskeratosis congenita, autosomal recessive 3, 613988
WRN	100%	100%	100%	100%	99.7%	Werner syndrome, 277700
WSB2	100%	100%	100%	99.9%	99.1%	Luo-Agrawal neurodevelopmental syndrome, 621552
WWOX	100%	100%	100%	100%	99.7%	Esophageal squamous cell carcinoma, somatic, 133239;Developmental and epileptic encephalopathy 28, 616211;Spinocerebellar ataxia, autosomal recessive 12, 614322
XDH	100%	100%	100%	100%	99.6%	Xanthinuria, type I, 278300
XPA	100%	100%	100%	100%	99.9%	Xeroderma pigmentosum, group A, 278700
XPC	100%	100%	100%	100%	99.6%	Xeroderma pigmentosum, group C, 278720
XPNPEP3	100%	100%	100%	99.9%	99.6%	Nephronophthisis-like nephropathy 1, 613159
XRCC1	100%	100%	100%	100%	99.2%	?Spinocerebellar ataxia, autosomal recessive 26, 617633
XRCC2	100%	100%	100%	100%	99.9%	Spermatogenic failure 50, 619145;?Premature ovarian failure 17, 619146;?Fanconi anemia, complementation group U, 617247

XRCC4	100%	100%	100%	100%	99.9%	Short stature, microcephaly, and endocrine dysfunction, 616541
XYLT1	100%	100%	100%	99.9%	99.3%	Desbuquois dysplasia 2, 615777;{Pseudoxanthoma elasticum, modifier of severity of}, 264800
XYLT2	99.9%	98.8%	100%	99.9%	99.5%	{Pseudoxanthoma elasticum, modifier of severity of}, 264800;Spondyloocular syndrome, 605822
YARS1	100%	98.5%	100%	100%	99.5%	Infantile-onset multisystem neurologic, endocrine, and pancreatic disease 2, 619418;Charcot-Marie-Tooth disease, dominant intermediate C, 608323
YARS2	100%	100%	100%	100%	99.8%	Myopathy, lactic acidosis, and sideroblastic anemia 2, 613561
YIF1B	90%	90%	100%	99.9%	99.4%	Kaya-Barakat-Masson syndrome, 619125
YIPF5	100%	100%	100%	100%	100%	Microcephaly, epilepsy, and diabetes syndrome 2, 619278
YME1L1	96.7%	96.7%	100%	100%	99.7%	?Optic atrophy 11, 617302
YRDC	100%	100%	100%	99.9%	98.7%	Galloway-Mowat syndrome 10, 619609
YY1AP1	100%	100%	100%	100%	99.9%	Grange syndrome, 602531
ZAP70	100%	100%	100%	100%	99.4%	Immunodeficiency 48, 269840;Autoimmune disease, multisystem, infantile-onset, 2, 617006

ZBTB11	100%	100%	100%	100%	99.8%	Intellectual developmental disorder, autosomal recessive 69, 618383
ZBTB16	87.8%	87.8%	100%	100%	99.3%	Leukemia, acute promyelocytic, PL2F/RARA type
ZBTB24	100%	100%	100%	100%	99.7%	Immunodeficiency-centromeric instability-facial anomalies syndrome 2, 614069
ZBTB42	100%	100%	100%	100%	99.3%	?Lethal congenital contracture syndrome 6, 616248
ZC3H14	100%	100%	100%	100%	99.8%	Intellectual developmental disorder, autosomal recessive 56, 617125
ZFYVE19	100%	100%	100%	100%	99.6%	Cholestasis, progressive familial intrahepatic, 9, 619849
ZFYVE26	100%	100%	100%	100%	99.7%	Spastic paraplegia 15, autosomal recessive, 270700
ZMPSTE24	100%	100%	100%	100%	100%	Mandibuloacral dysplasia with type B lipodystrophy, 608612;Restrictive dermopathy 1, 275210
ZMYND10	100%	100%	100%	100%	99.6%	Ciliary dyskinesia, primary, 22, 615444
ZNF142	100%	100%	100%	100%	99.6%	Neurodevelopmental disorder with impaired speech and hyperkinetic movements, 618425
ZNF335	100%	100%	100%	100%	99.5%	Microcephaly 10, primary, autosomal recessive, 615095
ZNF341	100%	100%	100%	100%	99.3%	Hyper-IgE syndrome 3, autosomal recessive, with recurrent infections, 618282

ZNF407	100%	100%	100%	100%	99.8%	SIMHA syndrome, 619557
ZNF408	96.2%	96.2%	100%	100%	99.6%	Retinitis pigmentosa 72, 616469;?Exudative vitreoretinopathy 6, 616468
ZNF423	100%	100%	100%	100%	99.4%	Nephronophthisis 14, 614844;Joubert syndrome 19, 614844
ZNF469	100%	100%	100%	99.9%	99.1%	Brittle cornea syndrome 1, 229200
ZNF513	100%	100%	100%	100%	99.4%	?Retinitis pigmentosa 58, 613617
ZNF526	100%	100%	100%	100%	99.3%	Dentici-Novelli neurodevelopmental syndrome, 619877
ZNF668	100%	100%	100%	100%	99.4%	Neurodevelopmental disorder with poor growth, large ears, and dysmorphic facies, 620194
ZNF699	100%	100%	100%	99.9%	99.3%	DEGCAGS syndrome, 619488
ZNF808	100%	100%	100%	100%	99.9%	Pancreatic agenesis 3, 620991
ZNFX1	100%	100%	100%	100%	99.6%	Immunodeficiency 91 and hyperinflammation, 619644
ZNHIT3	78.8%	76.2%	100%	100%	99.4%	PEHO syndrome, 260565
ZP1	100%	100%	100%	100%	99.4%	Oocyte/zygote/embryo maturation arrest 1, 615774
ZPR1	100%	100%	100%	99.9%	99.3%	Growth restriction, hypoplastic kidneys, alopecia, and distinctive facies, 619321
ZSCAN10	100%	100%	100%	100%	99.2%	Otofacial neurodevelopmental syndrome, 620910

Gene symbols used follow HGNC guidelines: Gray KA, Yates B, Seal RL, Wright MW, Bruford EA. Nucleic Acids Res. 2015 Jan 43(Database issue):D1079-85.

TWIST X2 covered 10x describes the percentage of a gene's coding sequence that is covered at least 10x when analyzed by WES using TWIST X2 chemistry mapped against GRCh38.
TWIST X2 covered 20x describes the percentage of a gene's coding sequence that is covered at least 20x when analyzed by WES using TWIST X2 chemistry mapped against GRCh38.
srWGS covered 10x describes the percentage of a gene's coding sequence that is covered at least 10x when analyzed by WGS mapped against GRCh38.
srWGS covered 15x describes the percentage of a gene's coding sequence that is covered at least 15x when analyzed by WGS mapped against GRCh38.
srWGS covered 20x describes the percentage of a gene's coding sequence that is covered at least 20x when analyzed by WGS mapped against GRCh38.
non-protein coding genes are covered, but as coverage statistics are based on protein coding regions, statistics could not be generated.
OMIM release used for OMIM disease identifiers and descriptions : November 25th, 2024.
This list is accurate for panel version DG 5.1.0
Ad 1. Blank field signifies a gene without a current OMIM association Ad 2. OMIM phenotype descriptions between {} signify risk factors