MENDELIOME GENE PANEL DG 3.3.0 (4837 genes)

Releasedate: 13-01-2022

Gene	*TWIST covered >10x*	*TWIST covered >20x*	*Associated Phenotype Description and OMIM disease ID*
A2M	100%	100%	No OMIM disease ID
A2ML1	100%	100%	No OMIM disease ID
A4GALT	100%	100%	NOR polyagglutination syndrome, 111400
AAAS	100%	100%	Achalasia-addisonianism-alacrimia syndrome, 231550
AAGAB	100%	100%	Keratoderma, palmoplantar, punctate type IA, 148600
AARS1	100%	100%	Developmental and epileptic encephalopathy 29, 616339 Charcot-Marie-Tooth disease, axonal, type 2N, 613287
AARS2	100%	100%	Leukoencephalopathy, progressive, with ovarian failure, 615889 Combined oxidative phosphorylation deficiency 8, 614096
AASS	100%	100%	Hyperlysinemia, 238700
ABAT	100%	100%	GABA-transaminase deficiency, 613163
ABCA1	100%	100%	Tangier disease, 205400 HDL deficiency, familial, 1, 604091
ABCA12	100%	100%	Ichthyosis, congenital, autosomal recessive 4B (harlequin), 242500 Ichthyosis, congenital, autosomal recessive 4A, 601277
ABCA2	100%	100%	Intellectual developmental disorder with poor growth and with or without seizures or ataxia, 618808
ABCA3	100%	100%	Surfactant metabolism dysfunction, pulmonary, 3, 610921
ABCA4	96%	96%	Retinal dystrophy, early-onset severe, 248200 Retinitis pigmentosa 19, 601718 Cone-rod dystrophy 3, 604116 Fundus flavimaculatus, 248200 Stargardt disease 1, 248200
ABCA5	100%	100%	?Hypertrichosis, congenital generalized, with gingival hyperplasia, 135400
ABCB10	100%	100%	No OMIM disease ID
ABCB11	100%	100%	Cholestasis, benign recurrent intrahepatic, 2, 605479 Cholestasis, progressive familial intrahepatic 2, 601847

ABCB4	100%	100%	Gallbladder disease 1, 600803 Cholestasis, intrahepatic, of pregnancy, 3, 614972 Cholestasis, progressive familial intrahepatic 3, 602347
ABCB6	100%	100%	Microphthalmia, isolated, with coloboma 7, 614497 Dyschromatosis universalis hereditaria 3, 615402 Pseudohyperkalemia, familial, 2, due to red cell leak, 609153
ABCB7	99%	99%	Anemia, sideroblastic, with ataxia, 301310
ABCC1	100%	100%	?Deafness, autosomal dominant 77, 618915
ABCC2	100%	100%	Dubin-Johnson syndrome, 237500
ABCC6	100%	100%	Pseudoxanthoma elasticum, 264800 Arterial calcification, generalized, of infancy, 2, 614473 Pseudoxanthoma elasticum, forme fruste, 177850
ABCC8	100%	100%	Diabetes mellitus, permanent neonatal 3, with or without neurologic features, 618857 Diabetes mellitus, transient neonatal 2, 610374 Diabetes mellitus, noninsulin-dependent, 125853 Hypoglycemia of infancy, leucine-sensitive, 240800 Hyperinsulinemic hypoglycemia, familial, 1, 256450
ABCC9	100%	100%	Cardiomyopathy, dilated, 1O, 608569 Hypertrichotic osteochondrodysplasia, 239850 ?Atrial fibrillation, familial, 12, 614050
ABCD1	100%	100%	Adrenoleukodystrophy, 300100 Adrenomyeloneuropathy, adult, 300100
ABCD2	100%	100%	No OMIM disease ID
ABCD3	100%	100%	?Bile acid synthesis defect, congenital, 5, 616278
ABCD4	100%	100%	Methylmalonic aciduria and homocystinuria, cblJ type, 614857
ABCG5	100%	100%	Sitosterolemia 2, 618666
ABCG8	100%	100%	Sitosterolemia 1, 210250
ABHD12	100%	100%	Polyneuropathy, hearing loss, ataxia, retinitis pigmentosa, and cataract, 612674
ABHD16A	100%	100%	No OMIM disease ID
ABHD5	100%	100%	Chanarin-Dorfman syndrome, 275630
ABL1	100%	100%	Leukemia, Philadelphia chromosome-positive, resistant to imatinib, 608232 Congenital heart defects and skeletal malformations syndrome, 617602
ACACA	100%	100%	No OMIM disease ID
ACAD8	100%	100%	Isobutyryl-CoA dehydrogenase deficiency, 611283
ACAD9	100%	100%	Mitochondrial complex I deficiency, nuclear type 20, 611126
ACADM	100%	100%	Acyl-CoA dehydrogenase, medium chain, deficiency of, 201450

ACADS	100%	100%	Acyl-CoA dehydrogenase, short-chain, deficiency of, 201470
ACADSB	100%	100%	2-methylbutyrylglycinuria, 610006
ACADVL	100%	100%	VLCAD deficiency, 201475
ACAN	98%	98%	?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%	Alpha-methylacetoacetic aciduria, 203750
ACAT2	100%	100%	No OMIM disease ID
ACBD5	100%	100%	Retinal dystrophy with leukodystrophy, 618863
ACD	100%	100%	?Dyskeratosis congenita, autosomal recessive 7, 616553 ?Dyskeratosis congenita, autosomal dominant 6, 616553
ACE	100%	100%	Renal tubular dysgenesis, 267430
ACER3	100%	100%	?Leukodystrophy, progressive, early childhood-onset, 617762
ACKR3	100%	100%	?Oculomotor-abducens synkinesis, 619215
ACO2	100%	100%	?Optic atrophy 9, 616289 Infantile cerebellar-retinal degeneration, 614559
ACOX1	100%	100%	Mitchell syndrome, 618960 Peroxisomal acyl-CoA oxidase deficiency, 264470
ACOX2	100%	100%	Bile acid synthesis defect, congenital, 6, 617308
ACP4	100%	100%	Amelogenesis imperfecta, type IJ, 617297
ACP5	100%	100%	Spondyloenchondrodysplasia with immune dysregulation, 607944
ACSF3	100%	100%	Combined malonic and methylmalonic aciduria, 614265
ACSL4	100%	100%	Intellectual developmental disorder, X-linked 63, 300387
ACSL6	97%	97%	Myelodysplastic syndrome, Myelogenous leukemia, acute,
ACTA1	100%	100%	?Myopathy, scapulohumeroperoneal, 616852 Nemaline myopathy 3, autosomal dominant or recessive, 161800 Myopathy, actin, congenital, with excess of thin myofilaments, 161800 Myopathy, actin, congenital, with cores, 161800 Myopathy, congenital, with fiber-type disproportion 1, 255310
ACTA2	100%	100%	Multisystemic smooth muscle dysfunction syndrome, 613834 Aortic aneurysm, familial thoracic 6, 611788 Moyamoya disease 5, 614042
ACTB	100%	100%	Baraitser-Winter syndrome 1, 243310 ?Dystonia, juvenile-onset, 607371

ACTC1	100%	100%	Left ventricular noncompaction 4, 613424 Cardiomyopathy, hypertrophic, 11, 612098 Atrial septal defect 5, 612794 Cardiomyopathy, dilated, 1R, 613424
ACTG1	100%	100%	Deafness, autosomal dominant 20/26, 604717 Baraitser-Winter syndrome 2, 614583
ACTG2	100%	100%	Megacystis-microcolon-intestinal hypoperistalsis syndrome 5, 619431 Visceral myopathy 1, 155310
ACTL6A	100%	100%	No OMIM disease ID
ACTL6B	100%	100%	Developmental and epileptic encephalopathy 76, 618468 Intellectual developmental disorder with severe speech and ambulation defects, 618470
ACTL9	100%	100%	Spermatogenic failure 53, 619258
ACTN1	100%	100%	Bleeding disorder, platelet-type, 15, 615193
ACTN2	100%	100%	Myopathy, distal, 6, adult onset, 618655 Cardiomyopathy, hypertrophic, 23, with or without LVNC, 612158 Cardiomyopathy, dilated, 1AA, with or without LVNC, 612158 Myopathy, congenital with structured cores and Z-line abnormalities, 618654
ACTN4	100%	100%	Glomerulosclerosis, focal segmental, 1, 603278
ACVR1	100%	100%	Fibrodysplasia ossificans progressiva, 135100
ACVR1B	100%	100%	Pancreatic cancer, somatic,
ACVR2B	100%	100%	Heterotaxy, visceral, 4, autosomal, 613751
ACVRL1	100%	100%	Telangiectasia, hereditary hemorrhagic, type 2, 600376
ACY1	100%	100%	Aminoacylase 1 deficiency, 609924

ADA	100%	100%	Adenosine deaminase deficiency, partial, 102700 Severe combined immunodeficiency due to ADA deficiency, 102700
ADA2	100%	100%	Sneddon syndrome, 182410 Vasculitis, autoinflammation, immunodeficiency, and hematologic defects syndrome, 615688
ADAD2	100%	100%	No OMIM disease ID
ADAM10	100%	100%	Reticulate acropigmentation of Kitamura, 615537
ADAM17	100%	100%	?Inflammatory skin and bowel disease, neonatal, 1, 614328
ADAM22	100%	100%	Developmental and epileptic encephalopathy 61, 617933
ADAM9	100%	100%	Cone-rod dystrophy 9, 612775
ADAMTS1	100%	100%	No OMIM disease ID
ADAMTS10	100%	100%	Weill-Marchesani syndrome 1, recessive, 277600
ADAMTS13	100%	100%	Thrombotic thrombocytopenic purpura, hereditary, 274150
ADAMTS17	99%	99%	Weill-Marchesani 4 syndrome, recessive, 613195
ADAMTS18	100%	100%	Microcornea, myopic chorioretinal atrophy, and telecanthus, 615458
ADAMTS19	100%	100%	No OMIM disease ID
ADAMTS2	98%	98%	Ehlers-Danlos syndrome, dermatosparaxis type, 225410
ADAMTS3	100%	100%	Hennekam lymphangiectasia-lymphedema syndrome 3, 618154
ADAMTS9	100%	100%	No OMIM disease ID
ADAMTSL2	99%	99%	Geleophysic dysplasia 1, 231050
ADAMTSL4	100%	100%	Ectopia lentis et pupillae, 225200 Ectopia lentis, isolated, autosomal recessive, 225100
ADAR	100%	100%	Dyschromatosis symmetrica hereditaria, 127400 Aicardi-Goutieres syndrome 6, 615010
ADARB1	95%	95%	Neurodevelopmental disorder with hypotonia, microcephaly, and seizures, 618862
ADAT3	100%	100%	Neurodevelopmental disorder with brain abnormalities, poor growth, and dysmorphic facies, 615286
ADCK2	100%	100%	No OMIM disease ID
ADCK5	100%	100%	No OMIM disease ID
ADCY1	99%	98%	?Deafness, autosomal recessive 44, 610154

ADCY10	100%	100%	No OMIM disease ID
ADCY3	100%	100%	No OMIM disease ID
ADCY5	100%	99%	Dyskinesia, familial, with facial myokymia, 606703
ADCY6	100%	100%	Lethal congenital contracture syndrome 8, 616287
ADD3	100%	100%	Cerebral palsy, spastic quadriplegic, 3, 617008
ADGRE2	99%	98%	Vibratory urticaria, 125630
ADGRG1	100%	100%	Polymicrogyria, bilateral frontoparietal, 606854 Polymicrogyria, bilateral perisylvian, 615752
ADGRG2	100%	100%	Congenital bilateral absence of vas deferens, X-linked, 300985
ADGRG6	100%	100%	Lethal congenital contracture syndrome 9, 616503
ADGRV1	100%	100%	Usher syndrome, type 2C, 605472 Usher syndrome, type 2C, GPR98/PDZD7 digenic, 605472 ?Febrile seizures, familial, 4, 604352
ADH5	100%	100%	AMED syndrome, digenic, 619151
ADIPOQ	100%	100%	Adiponectin deficiency, 612556
ADIPOR1	100%	100%	No OMIM disease ID
ADK	84%	84%	Hypermethioninemia due to adenosine kinase deficiency, 614300
ADNP	95%	95%	Helsmoortel-van der Aa syndrome, 615873
ADPRS	100%	100%	Neurodegeneration, childhood-onset, stress-induced, with variable ataxia and seizures, 618170
ADRB2	100%	100%	Beta-2-adrenoreceptor agonist, reduced response to,
ADSL	100%	100%	Adenylosuccinase deficiency, 103050
ADSS1	100%	100%	Myopathy, distal, 5, 617030
AEBP1	100%	100%	Ehlers-Danlos syndrome, classic-like, 2, 618000
AFF2	100%	99%	Intellectual developmental disorder, X-linked 109, 309548
AFF3	100%	100%	KINSSHIP syndrome, 619297
AFF4	100%	100%	CHOPS syndrome, 616368
AFG3L2	100%	100%	Spastic ataxia 5, autosomal recessive, 614487 Optic atrophy 12, 618977 Spinocerebellar ataxia 28, 610246
AFP	100%	100%	Alpha-fetoprotein deficiency, 615969
AGA	100%	100%	Aspartylglucosaminuria, 208400

AGAP1	100%	100%	No OMIM disease ID
AGBL1	100%	100%	Corneal dystrophy, Fuchs endothelial, 8, 615523
AGBL5	100%	100%	Retinitis pigmentosa 75, 617023
AGK	91%	91%	Cataract 38, autosomal recessive, 614691 Sengers syndrome, 212350
AGL	100%	100%	Glycogen storage disease IIIa, 232400 Glycogen storage disease IIIb, 232400
AGMO	100%	100%	No OMIM disease ID
AGO1	100%	100%	No OMIM disease ID
AGO2	100%	99%	Lessel-Kreienkamp syndrome, 619149
AGPAT2	100%	100%	Lipodystrophy, congenital generalized, type 1, 608594
AGPS	100%	100%	Rhizomelic chondrodysplasia punctata, type 3, 600121
AGRN	100%	100%	Myasthenic syndrome, congenital, 8, with pre- and postsynaptic defects, 615120
AGT	100%	100%	Renal tubular dysgenesis, 267430
AGTPBP1	100%	100%	Neurodegeneration, childhood-onset, with cerebellar atrophy, 618276
AGTR1	100%	100%	Renal tubular dysgenesis, 267430
AGXT	100%	100%	Hyperoxaluria, primary, type 1, 259900
AHCY	100%	100%	Hypermethioninemia with deficiency of S-adenosylhomocysteine hydrolase, 613752
AHDC1	100%	100%	Xia-Gibbs syndrome, 615829
AHI1	100%	100%	Joubert syndrome 3, 608629
AHNAK2	97%	97%	No OMIM disease ID
AHR	100%	100%	?Retinitis pigmentosa 85, 618345
AHSG	100%	100%	?Alopecia-mental retardation syndrome 1, 203650
AICDA	100%	100%	Immunodeficiency with hyper-IgM, type 2, 605258
AIFM1	100%	100%	Combined oxidative phosphorylation deficiency 6, 300816 Cowchock syndrome, 310490 Spondyloepimetaphyseal dysplasia, X-linked, with hypomyelinating leukodystrophy, 300232 Deafness, X-linked 5, 300614
AIMP1	100%	100%	Leukodystrophy, hypomyelinating, 3, 260600
AIMP2	100%	100%	Leukodystrophy, hypomyelinating, 17, 618006

AIP	100%	100%	Pituitary adenoma 1, multiple types, 102200 Pituitary adenoma predisposition, 102200
AIPL1	100%	100%	Leber congenital amaurosis 4, 604393 Retinitis pigmentosa, juvenile, 604393 Cone-rod dystrophy, 604393
AIRE	100%	100%	Autoimmune polyendocrinopathy syndrome , type I, with or without reversible metaphyseal dysplasia, 240300
AK1	100%	100%	Hemolytic anemia due to adenylate kinase deficiency, 612631
AK2	100%	100%	Reticular dysgenesis, 267500
AK7	100%	100%	?Spermatogenic failure 27, 617965
AKAP9	100%	100%	?Long QT syndrome 11, 611820
AKR1C1	100%	100%	No OMIM disease ID
AKR1C2	100%	100%	46XY sex reversal 8, 614279
AKR1D1	100%	100%	Bile acid synthesis defect, congenital, 2, 235555
AKT1	100%	100%	Breast cancer, somatic, 114480 Cowden syndrome 6, 615109 Colorectal cancer, somatic, 114500 Proteus syndrome, somatic, 176920 Ovarian cancer, somatic, 167000
AKT2	100%	100%	Diabetes mellitus, type II, 125853 Hypoinsulinemic hypoglycemia with hemihypertrophy, 240900
AKT3	100%	100%	Megalencephaly-polymicrogyria-polydactyly-hydrocephalus syndrome 2, 615937
ALAD	100%	100%	Porphyria, acute hepatic, 612740
ALAS2	100%	100%	Anemia, sideroblastic, 1, 300751 Protoporphyria, erythropoietic, X-linked, 300752
ALB	100%	100%	Analbuminemia, 616000

ALDH18A1	100%	100%	Spastic paraplegia 9A, autosomal dominant, 601162 Cutis laxa, autosomal recessive, type IIIA, 219150 Spastic paraplegia 9B, autosomal recessive, 616586 Cutis laxa, autosomal dominant 3, 616603
ALDH1A2	100%	100%	No OMIM disease ID
ALDH1A3	100%	100%	Microphthalmia, isolated 8, 615113
ALDH1B1	100%	100%	No OMIM disease ID
ALDH2	100%	100%	Alcohol sensitivity, acute, 610251
ALDH3A2	93%	93%	Sjogren-Larsson syndrome, 270200
ALDH4A1	100%	100%	Hyperprolinemia, type II, 239510
ALDH5A1	100%	100%	Succinic semialdehyde dehydrogenase deficiency, 271980
ALDH6A1	100%	100%	Methylmalonate semialdehyde dehydrogenase deficiency, 614105
ALDH7A1	100%	100%	Epilepsy, pyridoxine-dependent, 266100
ALDOA	100%	100%	Glycogen storage disease XII, 611881
ALDOB	100%	100%	Fructose intolerance, hereditary, 229600
ALG1	100%	100%	Congenital disorder of glycosylation, type Ik, 608540
ALG10	100%	100%	No OMIM disease ID
ALG11	96%	96%	Congenital disorder of glycosylation, type Ip, 613661
ALG12	100%	100%	Congenital disorder of glycosylation, type Ig, 607143
ALG13	100%	99%	?Congenital disorder of glycosylation, type Is, 300884 Developmental and epileptic encephalopathy 36, 300884

ALG14	100%	100%	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%	?Congenital disorder of glycosylation, type Ii, 607906 Myasthenic syndrome, congenital, 14, with tubular aggregates, 616228
ALG3	100%	100%	Congenital disorder of glycosylation, type Id, 601110
ALG6	100%	100%	Congenital disorder of glycosylation, type Ic, 603147
ALG8	96%	96%	Congenital disorder of glycosylation, type Ih, 608104 Polycystic liver disease 3 with or without kidney cysts, 617874
ALG9	100%	100%	Gillessen-Kaesbach-Nishimura syndrome, 263210 Congenital disorder of glycosylation, type Il, 608776
ALK	100%	100%	No OMIM disease ID
ALKBH1	100%	100%	No OMIM disease ID
ALKBH8	100%	100%	Intellectual developmental disorder, autosomal recessive 71, 618504
ALMS1	100%	100%	Alstrom syndrome, 203800
ALOX12B	100%	100%	Ichthyosis, congenital, autosomal recessive 2, 242100
ALOXE3	100%	100%	Ichthyosis, congenital, autosomal recessive 3, 606545
ALPI	100%	100%	No OMIM disease ID
ALPK1	100%	100%	ROSAH syndrome, 614979
ALPK3	100%	100%	Cardiomyopathy, familial hypertrophic 27, 618052

ALPL	100%	100%	Odontohypophosphatasia, 146300 Hypophosphatasia, infantile, 241500 Hypophosphatasia, childhood, 241510 Hypophosphatasia, adult, 146300
ALS2	100%	100%	Primary lateral sclerosis, juvenile, 606353 Spastic paralysis, infantile onset ascending, 607225 Amyotrophic lateral sclerosis 2, juvenile, 205100
ALX1	100%	100%	Frontonasal dysplasia 3, 613456
ALX3	100%	100%	Frontonasal dysplasia 1, 136760
ALX4	100%	100%	Parietal foramina 2, 609597 Frontonasal dysplasia 2, 613451
AMACR	100%	100%	Alpha-methylacyl-CoA racemase deficiency, 614307 Bile acid synthesis defect, congenital, 4, 214950
AMBN	100%	100%	Amelogenesis imperfecta, type IF, 616270
AMELX	100%	100%	Amelogenesis imperfecta, type 1E, 301200
AMER1	100%	100%	Osteopathia striata with cranial sclerosis, 300373
AMH	100%	100%	Persistent Mullerian duct syndrome, type I, 261550
AMHR2	100%	100%	Persistent Mullerian duct syndrome, type II, 261550
AMMECR1	100%	100%	Midface hypoplasia, hearing impairment, elliptocytosis, and nephrocalcinosis, 300990
AMN	100%	100%	Imerslund-Grasbeck syndrome 2, 618882
AMPD1	100%	100%	Myopathy due to myoadenylate deaminase deficiency, 615511
AMPD2	100%	100%	?Spastic paraplegia 63, 615686 Pontocerebellar hypoplasia, type 9, 615809
AMPD3	100%	100%	No OMIM disease ID
AMT	100%	100%	Glycine encephalopathy, 605899
AMTN	100%	100%	?Amelogenesis imperfecta, type IIIB, 617607
ANAPC1	100%	100%	Rothmund-Thomson syndrome, type 1, 618625
ANG	100%	100%	Amyotrophic lateral sclerosis 9, 611895

ANGPT1	100%	100%	?Angioedema, hereditary, 5, 619361
ANGPT2	100%	100%	Lymphatic malformation 10, 619369
ANGPTL3	100%	100%	Hypobetalipoproteinemia, familial, 2, 605019
ANGPTL4	100%	100%	Plasma triglyceride level QTL, low, 615881
ANK1	100%	100%	Spherocytosis, type 1, 182900
ANK2	100%	100%	Long QT syndrome 4, 600919 Cardiac arrhythmia, ankyrin-B-related, 600919
ANK3	100%	100%	Mental retardation, autosomal recessive, 37, 615493
ANKFY1	100%	100%	No OMIM disease ID
ANKH	100%	100%	Chondrocalcinosis 2, 118600 Craniometaphyseal dysplasia, 123000
ANKLE2	100%	100%	Microcephaly 16, primary, autosomal recessive, 616681
ANKRD1	100%	100%	No OMIM disease ID
ANKRD11	100%	100%	KBG syndrome, 148050
ANKRD17	100%	100%	Chopra-Amiel-Gordon syndrome, 619504
ANKRD26	97%	97%	Thrombocytopenia 2, 188000
ANKS1B	100%	100%	No OMIM disease ID
ANKS6	99%	99%	Nephronophthisis 16, 615382
ANLN	100%	100%	Focal segmental glomerulosclerosis 8, 616032
ANO10	100%	100%	Spinocerebellar ataxia, autosomal recessive 10, 613728
ANO3	100%	100%	Dystonia 24, 615034
ANO5	100%	100%	Muscular dystrophy, limb-girdle, autosomal recessive 12, 611307 Miyoshi muscular dystrophy 3, 613319 Gnathodiaphyseal dysplasia, 166260
ANO6	100%	100%	Scott syndrome, 262890
ANOS1	100%	99%	Hypogonadotropic hypogonadism 1 with or without anosmia (Kallmann syndrome 1), 308700
ANTXR1	100%	100%	GAPO syndrome, 230740
ANTXR2	100%	100%	Hyaline fibromatosis syndrome, 228600
ANXA11	100%	100%	Amyotrophic lateral sclerosis 23, 617839
AP1B1	100%	100%	Keratitis-ichthyosis-deafness syndrome, autosomal recessive, 242150

AP1G1	100%	100%	Usmani-Riazuddin syndrome, autosomal recessive, 619548 Usmani-Riazuddin syndrome, autosomal dominant, 619467
AP1S1	100%	100%	MEDNIK syndrome, 609313
AP1S2	100%	100%	Pettigrew syndrome, 304340
AP1S3	90%	90%	No OMIM disease ID
AP2M1	100%	100%	Intellectual developmental disorder 60 with seizures, 618587
AP2S1	100%	100%	Hypocalciuric hypercalcemia, type III, 600740
AP3B1	100%	100%	Hermansky-Pudlak syndrome 2, 608233
AP3B2	100%	99%	Developmental and epileptic encephalopathy 48, 617276
AP3D1	100%	100%	?Hermansky-Pudlak syndrome 10, 617050
AP4B1	100%	100%	Spastic paraplegia 47, autosomal recessive, 614066
AP4E1	100%	100%	Stuttering, familial persistent, 1, 184450 Spastic paraplegia 51, autosomal recessive, 613744
AP4M1	100%	100%	Spastic paraplegia 50, autosomal recessive, 612936
AP4S1	87%	87%	Spastic paraplegia 52, autosomal recessive, 614067
AP5Z1	100%	100%	Spastic paraplegia 48, autosomal recessive, 613647
APC	100%	100%	Colorectal cancer, somatic, 114500 Brain tumor-polyposis syndrome 2, 175100 Desmoid disease, hereditary, 135290 Adenoma, periampullary, somatic, 175100 Hepatoblastoma, somatic, 114550 Gastric cancer, somatic, 613659 Gastric adenocarcinoma and proximal polyposis of the stomach, 619182 Gardner syndrome, 175100 Adenomatous polyposis coli, 175100
APC2	100%	100%	Cortical dysplasia, complex, with other brain malformations 10, 618677 ?Sotos syndrome 3, 617169

APCDD1	100%	100%	Hypotrichosis 1, 605389
APOA1	100%	100%	Hypoalphalipoproteinemia, primary, 2, with or without corneal clouding, 618463 Amyloidosis, 3 or more types, 105200 ApoA-I and apoC-III deficiency, combined, 618463
APOA2	100%	100%	Apolipoprotein A-II deficiency,
APOA5	100%	99%	Hyperchylomicronemia, late-onset, 144650
APOB	100%	100%	Hypercholesterolemia, familial, 2, 144010 Hypobetalipoproteinemia, 615558
APOC2	100%	100%	Hyperlipoproteinemia, type Ib, 207750
APOC3	100%	100%	Apolipoprotein C-III deficiency, 614028
APOE	100%	100%	Alzheimer disease 2, 104310 Sea-blue histiocyte disease, 269600 Lipoprotein glomerulopathy, 611771 Hyperlipoproteinemia, type III, 617347
APOL1	100%	100%	No OMIM disease ID
APOO	100%	100%	No OMIM disease ID
APP	100%	100%	Cerebral amyloid angiopathy, Dutch, Italian, Iowa, Flemish, Arctic variants, 605714 Alzheimer disease 1, familial, 104300
APRT	100%	100%	Adenine phosphoribosyltransferase deficiency, 614723
APTX	100%	100%	Ataxia, early-onset, with oculomotor apraxia and hypoalbuminemia, 208920
AQP2	100%	100%	Diabetes insipidus, nephrogenic, 2, 125800
AQP5	100%	100%	Palmoplantar keratoderma, Bothnian type, 600231
AR	100%	100%	Androgen insensitivity, partial, with or without breast cancer, 312300 Androgen insensitivity, 300068 Spinal and bulbar muscular atrophy of Kennedy, 313200 Hypospadias 1, X-linked, 300633

ARCN1	97%	96%	Short stature, rhizomelic, with microcephaly, micrognathia, and developmental delay, 617164
ARF1	100%	100%	Periventricular nodular heterotopia 8, 618185
ARF3	100%	100%	No OMIM disease ID
ARFGEF1	100%	100%	No OMIM disease ID
ARFGEF2	100%	100%	Periventricular heterotopia with microcephaly, 608097
ARG1	92%	92%	Argininemia, 207800
ARHGAP24	100%	100%	No OMIM disease ID
ARHGAP26	100%	100%	Leukemia, juvenile myelomonocytic, somatic, 607785
ARHGAP29	100%	100%	No OMIM disease ID
ARHGAP31	100%	100%	Adams-Oliver syndrome 1, 100300
ARHGAP35	100%	100%	No OMIM disease ID
ARHGDIA	100%	100%	Nephrotic syndrome, type 8, 615244
ARHGEF1	100%	100%	?Immunodeficiency 62, 618459
ARHGEF10	100%	100%	?Slowed nerve conduction velocity, AD, 608236
ARHGEF18	100%	100%	Retinitis pigmentosa 78, 617433
ARHGEF2	100%	100%	?Neurodevelopmental disorder with midbrain and hindbrain malformations, 617523
ARHGEF28	100%	100%	No OMIM disease ID
ARHGEF6	100%	100%	No OMIM disease ID
ARHGEF9	97%	97%	Developmental and epileptic encephalopathy 8, 300607
ARID1A	100%	100%	Coffin-Siris syndrome 2, 614607
ARID1B	98%	98%	Coffin-Siris syndrome 1, 135900
ARID2	100%	100%	Coffin-Siris syndrome 6, 617808
ARIH1	100%	100%	No OMIM disease ID
ARL13B	100%	100%	Joubert syndrome 8, 612291
ARL2	100%	100%	?Microcornea, rod-cone dystrophy, cataract, and posterior staphyloma 1, 619082
ARL2BP	100%	100%	Retinitis pigmentosa with or without situs inversus, 615434
ARL3	100%	100%	Retinitis pigmentosa 83, 618173 Joubert syndrome 35, 618161
ARL6	100%	100%	Retinitis pigmentosa 55, 613575 Bardet-Biedl syndrome 3, 600151
ARL6IP1	100%	100%	?Spastic paraplegia 61, autosomal recessive, 615685
ARMC2	100%	100%	Spermatogenic failure 38, 618433
ARMC5	100%	100%	ACTH-independent macronodular adrenal hyperplasia 2, 615954
ARMC9	100%	100%	Joubert syndrome 30, 617622

ARNT2	100%	100%	?Webb-Dattani syndrome, 615926
ARPC1B	100%	100%	Immunodeficiency 71 with inflammatory disease and congenital thrombocytopenia, 617718
ARR3	100%	100%	Myopia 26, X-linked, female-limited, 301010
ARSA	100%	100%	Metachromatic leukodystrophy, 250100
ARSB	100%	100%	Mucopolysaccharidosis type VI (Maroteaux-Lamy), 253200
ARSG	100%	100%	Usher syndrome, type IV, 618144
ARSL	100%	100%	Chondrodysplasia punctata, X-linked recessive, 302950
ARV1	100%	100%	Developmental and epileptic encephalopathy 38, 617020
ARX	98%	95%	Proud syndrome, 300004 Hydranencephaly with abnormal genitalia, 300215 Partington syndrome, 309510 Developmental and epileptic encephalopathy 1, 308350 Lissencephaly, X-linked 2, 300215 Intellectual developmental disorder, X-linked 29, 300419
ASAH1	100%	100%	Spinal muscular atrophy with progressive myoclonic epilepsy, 159950 Farber lipogranulomatosis, 228000
ASB10	100%	100%	Glaucoma 1, open angle, F, 603383
ASCC1	87%	87%	Spinal muscular atrophy with congenital bone fractures 2, 616867 Barrett esophagus/esophageal adenocarcinoma, 614266
ASCL1	100%	100%	No OMIM disease ID
ASH1L	98%	98%	Mental retardation, autosomal dominant 52, 617796
ASIP	100%	100%	No OMIM disease ID
ASL	100%	100%	Argininosuccinic aciduria, 207900
ASNS	100%	100%	Asparagine synthetase deficiency, 615574
ASPA	100%	100%	Canavan disease, 271900
ASPH	100%	100%	Traboulsi syndrome, 601552
ASPM	100%	100%	Microcephaly 5, primary, autosomal recessive, 608716
ASPRV1	100%	100%	Ichthyosis, lamellar, autosomal dominant, 146750

ASPSCR1	100%	100%	Alveolar soft-part sarcoma, 606243
ASRGL1	100%	100%	No OMIM disease ID
ASS1	100%	100%	Citrullinemia, 215700
ASXL1	100%	100%	Myelodysplastic syndrome, somatic, 614286 Bohring-Opitz syndrome, 605039
ASXL2	100%	100%	Shashi-Pena syndrome, 617190
ASXL3	100%	100%	Bainbridge-Ropers syndrome, 615485
ATAD1	100%	100%	Hyperekplexia 4, 618011
ATAD3A	100%	100%	Harel-Yoon syndrome, 617183 Pontocerebellar hypoplasia, hypotonia, and respiratory insufficiency syndrome, neonatal lethal, 618810
ATAD3B	100%	100%	No OMIM disease ID
ATCAY	100%	100%	Ataxia, cerebellar, Cayman type, 601238
ATF3	100%	100%	No OMIM disease ID
ATF6	100%	100%	Achromatopsia 7, 616517
ATG4A	100%	99%	No OMIM disease ID
ATG5	100%	100%	?Spinocerebellar ataxia, autosomal recessive 25, 617584
ATG7	100%	100%	Spinocerebellar ataxia, autosomal recessive 31, 619422
ATIC	100%	100%	AICA-ribosiduria due to ATIC deficiency, 608688
ATL1	100%	100%	Spastic paraplegia 3A, autosomal dominant, 182600 Neuropathy, hereditary sensory, type ID, 613708
ATL3	100%	100%	Neuropathy, hereditary sensory, type IF, 615632
ATM	100%	100%	Ataxia-telangiectasia, 208900 Lymphoma, B-cell non-Hodgkin, somatic, T-cell prolymphocytic leukemia, somatic, Lymphoma, mantle cell, somatic,

ATN1	100%	100%	Dentatorubral-pallidoluysian atrophy, 125370 Congenital hypotonia, epilepsy, developmental delay, and digital anomalies, 618494
ATOH1	100%	100%	No OMIM disease ID
ATOH7	100%	99%	Persistent hyperplastic primary vitreous, autosomal recessive, 221900
ATP11C	100%	99%	?Hemolytic anemia, congenital, X-linked, 301015
ATP13A2	100%	100%	Spastic paraplegia 78, autosomal recessive, 617225 Kufor-Rakeb syndrome, 606693
ATP1A1	100%	100%	Hypomagnesemia, seizures, and mental retardation 2, 618314 Charcot-Marie-Tooth disease, axonal, type 2DD, 618036
ATP1A2	100%	100%	Developmental and epileptic encephalopathy 98, 619605 Fetal akinesia, respiratory insufficiency, microcephaly, polymicrogyria, and dysmorphic facies, 619602 Alternating hemiplegia of childhood 1, 104290 Migraine, familial basilar, 602481 Migraine, familial hemiplegic, 2, 602481
ATP1A3	100%	100%	Alternating hemiplegia of childhood 2, 614820 Dystonia-12, 128235 CAPOS syndrome, 601338 Developmental and epileptic encephalopathy 99, 619606
ATP2A1	100%	100%	Brody myopathy, 601003
ATP2A2	100%	100%	Acrokeratosis verruciformis, 101900 Darier disease, 124200

ATP2B2	100%	100%	No OMIM disease ID
ATP2B3	100%	100%	?Spinocerebellar ataxia, X-linked 1, 302500
ATP2C1	100%	100%	Hailey-Hailey disease, 169600
ATP4A	100%	100%	No OMIM disease ID
ATP5F1A	100%	100%	?Mitochondrial complex V (ATP synthase) deficiency, nuclear type 4, 615228 ?Combined oxidative phosphorylation deficiency 22, 616045
ATP5F1B	100%	100%	No OMIM disease ID
ATP5F1C	100%	100%	No OMIM disease ID
ATP5F1D	100%	100%	Mitochondrial complex V (ATP synthase) deficiency, 618120
ATP5F1E	100%	100%	?Mitochondrial complex V (ATP synthase) deficiency, nuclear type 3, 614053
ATP5IF1	100%	100%	No OMIM disease ID
ATP5MC1	100%	100%	No OMIM disease ID
ATP5MC2	100%	100%	No OMIM disease ID
ATP5MC3	100%	100%	No OMIM disease ID
ATP5ME	100%	100%	No OMIM disease ID
ATP5MF	100%	100%	No OMIM disease ID
ATP5MG	100%	100%	No OMIM disease ID
ATP5MGL	100%	100%	No OMIM disease ID
ATP5MK	100%	100%	Mitochondrial complex V (ATP synthase) deficiency, nuclear type 6, 618683
ATP5PB	100%	100%	No OMIM disease ID
ATP5PD	100%	100%	No OMIM disease ID
ATP5PF	100%	100%	No OMIM disease ID
ATP5PO	100%	100%	No OMIM disease ID
ATP6AP1	100%	100%	Immunodeficiency 47, 300972
ATP6AP2	100%	100%	Intellectual developmental disorder, X-linked, syndromic, Hedera type, 300423 ?Parkinsonism with spasticity, X-linked, 300911 Congenital disorder of glycosylation, type IIr, 301045

ATP6V0A1	100%	100%	No OMIM disease ID
ATP6V0A2	100%	100%	Wrinkly skin syndrome, 278250 Cutis laxa, autosomal recessive, type IIA, 219200
ATP6V0A4	100%	100%	Distal renal tubular acidosis 3, with or without sensorineural hearing loss, 602722
ATP6V0C	100%	100%	No OMIM disease ID
ATP6V1A	100%	100%	Cutis laxa, autosomal recessive, type IID, 617403 Developmental and epileptic encephalopathy 93, 618012
ATP6V1B1	100%	100%	Distal renal tubular acidosis 2 with progressive sensorineural hearing loss, 267300
ATP6V1B2	100%	100%	Zimmermann-Laband syndrome 2, 616455 Deafness, congenital, with onychodystrophy, autosomal dominant, 124480
ATP6V1E1	100%	100%	Cutis laxa, autosomal recessive, type IIC, 617402
ATP7A	100%	100%	Occipital horn syndrome, 304150 Spinal muscular atrophy, distal, X-linked 3, 300489 Menkes disease, 309400
ATP7B	100%	100%	Wilson disease, 277900
ATP8A2	100%	100%	?Cerebellar ataxia, mental retardation, and dysequilibrium syndrome 4, 615268
ATP8B1	100%	100%	Cholestasis, progressive familial intrahepatic 1, 211600 Cholestasis, intrahepatic, of pregnancy, 1, 147480 Cholestasis, benign recurrent intrahepatic, 243300
ATP9A	100%	100%	No OMIM disease ID
ATPAF1	100%	100%	No OMIM disease ID
ATPAF2	100%	100%	?Mitochondrial complex V (ATP synthase) deficiency, nuclear type 1, 604273
ATR	100%	100%	Seckel syndrome 1, 210600 ?Cutaneous telangiectasia and cancer syndrome, familial, 614564

ATRX	100%	100%	Alpha-thalassemia/mental retardation syndrome, 301040 Alpha-thalassemia myelodysplasia syndrome, somatic, 300448 Mental retardation-hypotonic facies syndrome, X-linked, 309580
ATXN1	100%	100%	Spinocerebellar ataxia 1, 164400
ATXN10	100%	100%	Spinocerebellar ataxia 10, 603516
ATXN2	100%	100%	Spinocerebellar ataxia 2, 183090
ATXN2L	100%	100%	No OMIM disease ID
ATXN3	95%	95%	Machado-Joseph disease, 109150
ATXN7	100%	99%	Spinocerebellar ataxia 7, 164500
ATXN8OS	NC	NC	Spinocerebellar ataxia 8, 608768
AUH	100%	100%	3-methylglutaconic aciduria, type I, 250950
AURKC	100%	100%	Spermatogenic failure 5, 243060
AUTS2	100%	100%	Intellectual developmental disorder, autosomal dominant 26, 615834
AVIL	100%	100%	Nephrotic syndrome, type 21, 618594
AVP	100%	100%	Diabetes insipidus, neurohypophyseal, 125700
AVPR2	100%	100%	Diabetes insipidus, nephrogenic, 1, 304800 Nephrogenic syndrome of inappropriate antidiuresis, 300539
AXIN1	100%	100%	Hepatocellular carcinoma, somatic, 114550 ?Caudal duplication anomaly, 607864
AXIN2	100%	100%	Colorectal cancer, somatic, 114500 Oligodontia-colorectal cancer syndrome, 608615
AXL	100%	100%	No OMIM disease ID
B2M	100%	100%	?Amyloidosis, familial visceral, 105200 Immunodeficiency 43, 241600
B3GALNT1	100%	100%	No OMIM disease ID
B3GALNT2	92%	92%	Muscular dystrophy-dystroglycanopathy (congenital with brain and eye anomalies, type A, 11, 615181

B3GALT6	99%	95%	Ehlers-Danlos syndrome, spondylodysplastic type, 2, 615349 Spondyloepimetaphyseal dysplasia with joint laxity, type 1, with or without fractures, 271640 Al-Gazali syndrome, 609465
B3GAT3	96%	95%	Multiple joint dislocations, short stature, craniofacial dysmorphism, with or without congenital heart defects, 245600
B3GLCT	100%	100%	Peters-plus syndrome, 261540
B4GALNT1	100%	100%	Spastic paraplegia 26, autosomal recessive, 609195
B4GALT1	100%	100%	Congenital disorder of glycosylation, type IId, 607091
B4GALT7	100%	100%	Ehlers-Danlos syndrome, spondylodysplastic type, 1, 130070
B4GAT1	100%	100%	Muscular dystrophy-dystroglycanopathy (congenital with brain and eye anomalies), type A, 13, 615287
B9D1	97%	94%	?Meckel syndrome 9, 614209 Joubert syndrome 27, 617120
B9D2	100%	100%	?Meckel syndrome 10, 614175 Joubert syndrome 34, 614175
BAAT	100%	100%	Hypercholanemia, familial, 607748 Bile acid conjugation defect 1, 619232
BACH2	100%	100%	Immunodeficiency 60 and autoimmunity, 618394
BAG3	100%	100%	Cardiomyopathy, dilated, 1HH, 613881 Myopathy, myofibrillar, 6, 612954
BANF1	100%	100%	Nestor-Guillermo progeria syndrome, 614008
BAP1	100%	100%	Tumor predisposition syndrome, 614327
BARD1	100%	100%	No OMIM disease ID
BAX	100%	100%	Colorectal cancer, somatic, 114500 T-cell acute lymphoblastic leukemia, somatic, 613065
BAZ2B	100%	100%	No OMIM disease ID
BBIP1	100%	100%	?Bardet-Biedl syndrome 18, 615995
BBS1	100%	100%	Bardet-Biedl syndrome 1, 209900
BBS10	100%	100%	Bardet-Biedl syndrome 10, 615987
BBS12	100%	100%	Bardet-Biedl syndrome 12, 615989
BBS2	100%	100%	Retinitis pigmentosa 74, 616562 Bardet-Biedl syndrome 2, 615981

BBS4	100%	100%	Bardet-Biedl syndrome 4, 615982
BBS5	100%	100%	Bardet-Biedl syndrome 5, 615983
BBS7	100%	100%	Bardet-Biedl syndrome 7, 615984
BBS9	95%	95%	Bardet-Biedl syndrome 9, 615986
BCAP31	100%	99%	Deafness, dystonia, and cerebral hypomyelination, 300475
BCAS3	100%	100%	Hengel-Maroofian-Schols syndrome, 619641
BCAT1	100%	100%	No OMIM disease ID
BCAT2	100%	100%	?Hypervalinemia or hyperleucine-isoleucinemia, 618850
BCHE	100%	100%	Butyrylcholinesterase deficiency, 617936
BCKDHA	100%	100%	Maple syrup urine disease, type Ia, 248600
BCKDHB	100%	100%	Maple syrup urine disease, type Ib, 248600
BCKDK	100%	100%	Branched-chain ketoacid dehydrogenase kinase deficiency, 614923
BCL10	100%	100%	?Immunodeficiency 37, 616098 Lymphoma, MALT, somatic, 137245
BCL11A	100%	100%	Dias-Logan syndrome, 617101
BCL11B	99%	99%	Intellectual developmental disorder with dysmorphic facies, speech delay, and T-cell abnormalities, 618092 Immunodeficiency 49, 617237
BCL2	100%	100%	Leukemia/lymphoma, B-cell, 2,
BCL7A	100%	100%	B-cell non-Hodgkin lymphoma, high-grade,
BCO1	100%	100%	?Hypercarotenemia and vitamin A deficiency, autosomal dominant, 115300
BCOR	100%	100%	Microphthalmia, syndromic 2, 300166
BCORL1	100%	100%	Shukla-Vernon syndrome, 301029
BCS1L	100%	100%	GRACILE syndrome, 603358 Mitochondrial complex III deficiency, nuclear type 1, 124000 Bjornstad syndrome, 262000
BDP1	100%	100%	?Deafness, autosomal recessive 112, 618257
BEAN1	92%	92%	Spinocerebellar ataxia 31, 117210

BEST1	100%	99%	Macular dystrophy, vitelliform, 2, 153700 ?Microcornea, rod-cone dystrophy, cataract, and posterior staphyloma 2, 193220 Retinitis pigmentosa-50, 613194 Retinitis pigmentosa, concentric, 613194 Vitreoretinochoroidopathy, 193220 Bestrophinopathy, autosomal recessive, 611809
BFSP1	100%	100%	Cataract 33, multiple types, 611391
BFSP2	100%	100%	Cataract 12, multiple types, 611597
BGN	100%	100%	Meester-Loeys syndrome, 300989 Spondyloepimetaphyseal dysplasia, X-linked, 300106
BHLHA9	100%	100%	?Camptosynpolydactyly, complex, 607539 Syndactyly, mesoaxial synostotic, with phalangeal reduction, 609432
BICC1	100%	100%	No OMIM disease ID
BICD2	100%	100%	Spinal muscular atrophy, lower extremity-predominant, 2B, autosomal dominant, 618291 Spinal muscular atrophy, lower extremity-predominant, 2A, autosomal dominant, 615290
BICRA	100%	100%	Coffin-Siris syndrome 12, 619325
BIN1	100%	100%	Centronuclear myopathy 2, 255200
BLK	100%	100%	Maturity-onset diabetes of the young, type 11, 613375
BLM	100%	100%	Bloom syndrome, 210900
BLNK	100%	100%	?Agammaglobulinemia 4, 613502
BLOC1S1	100%	100%	No OMIM disease ID
BLOC1S3	100%	100%	Hermansky-Pudlak syndrome 8, 614077
BLOC1S5	100%	100%	Hermansky-Pudlak syndrome 11, 619172

BLOC1S6	100%	100%	?Hermansky-Pudlak syndrome 9, 614171
BLVRA	100%	100%	Hyperbiliverdinemia, 614156
BMP1	100%	100%	Osteogenesis imperfecta, type XIII, 614856
BMP15	100%	100%	Premature ovarian failure 4, 300510 Ovarian dysgenesis 2, 300510
BMP2	100%	100%	Short stature, facial dysmorphism, and skeletal anomalies with or without cardiac anomalies 1, 617877 Brachydactyly, type A2, 112600
BMP4	100%	100%	Orofacial cleft 11, 600625 Microphthalmia, syndromic 6, 607932
BMP6	100%	99%	No OMIM disease ID
BMP7	100%	100%	No OMIM disease ID
BMPER	100%	100%	Diaphanospondylodysostosis, 608022
BMPR1A	100%	100%	Polyposis syndrome, hereditary mixed, 2, 610069 Polyposis, juvenile intestinal, 174900
BMPR1B	100%	100%	Acromesomelic dysplasia 3, 609441 Brachydactyly, type A2, 112600 Brachydactyly, type A1, D, 616849
BMPR2	99%	99%	Pulmonary hypertension, familial primary, 1, with or without HHT, 178600 Pulmonary hypertension, primary, fenfluramine or dexfenfluramine-associated, 178600 Pulmonary venoocclusive disease 1, 265450
BMS1	100%	100%	?Aplasia cutis congenita, nonsyndromic, 107600
BNC1	100%	99%	?Premature ovarian failure 16, 618723
BNC2	100%	100%	Lower urinary tract obstruction, congenital, 618612
BOLA1	100%	100%	No OMIM disease ID
BOLA2	100%	100%	No OMIM disease ID
BOLA3	100%	100%	Multiple mitochondrial dysfunctions syndrome 2 with hyperglycinemia, 614299
BPGM	100%	100%	Erythrocytosis, familial, 8, 222800

BPNT2	100%	100%	Chondrodysplasia with joint dislocations, GPAPP type, 614078
BPTF	100%	100%	Neurodevelopmental disorder with dysmorphic facies and distal limb anomalies, 617755
BRAF	100%	100%	Melanoma, malignant, somatic, 155600 LEOPARD syndrome 3, 613707 Cardiofaciocutaneous syndrome, 115150 Adenocarcinoma of lung, somatic, 211980 Noonan syndrome 7, 613706 Colorectal cancer, somatic, 114500 Nonsmall cell lung cancer, somatic, 211980
BRAT1	100%	100%	Neurodevelopmental disorder with cerebellar atrophy and with or without seizures, 618056 Rigidity and multifocal seizure syndrome, lethal neonatal, 614498
BRCA1	100%	100%	Fanconi anemia, complementation group S, 617883
BRCA2	100%	100%	Fanconi anemia, complementation group D1, 605724 Wilms tumor, 194070
BRDT	100%	100%	?Spermatogenic failure 21, 617644
BRF1	100%	100%	Cerebellofaciodental syndrome, 616202
BRIP1	100%	100%	Fanconi anemia, complementation group J, 609054
BRPF1	100%	100%	Intellectual developmental disorder with dysmorphic facies and ptosis, 617333
BRSK2	100%	100%	No OMIM disease ID
BRWD3	100%	100%	Intellectual developmental disorder, X-linked 93, 300659

BSCL2	100%	100%	Lipodystrophy, congenital generalized, type 2, 269700 Neuropathy, distal hereditary motor, type VC, 619112 Silver spastic paraplegia syndrome, 270685 Encephalopathy, progressive, with or without lipodystrophy, 615924
BSND	100%	100%	Sensorineural deafness with mild renal dysfunction, 602522 Bartter syndrome, type 4a, 602522
BTD	83%	83%	Biotinidase deficiency, 253260
BTG4	100%	100%	Oocyte maturation defect 8, 619009
BTK	100%	100%	Agammaglobulinemia, X-linked 1, 300755 Isolated growth hormone deficiency, type III, with agammaglobulinemia, 307200
BTRC	100%	100%	No OMIM disease ID
BUB1	100%	100%	Colorectal cancer with chromosomal instability, somatic, 114500
BUB1B	100%	100%	Colorectal cancer, somatic, 114500 Mosaic variegated aneuploidy syndrome 1, 257300
BUB3	100%	100%	No OMIM disease ID
BVES	100%	100%	Muscular dystrophy, limb-girdle, autosomal recessive 25, 616812
C11orf80	91%	91%	Hydatidiform mole, recurrent, 4, 618432
C12orf4	100%	100%	Mental retardation, autosomal recessive 66, 618221
C12orf57	100%	100%	Temtamy syndrome, 218340
C14orf39	100%	100%	Spermatogenic failure 52, 619202 ?Premature ovarian failure 18, 619203
C19orf12	100%	100%	Neurodegeneration with brain iron accumulation 4, 614298 ?Spastic paraplegia 43, autosomal recessive, 615043

C1GALT1C1	100%	100%	Tn polyagglutination syndrome, somatic, 300622
C1QA	100%	100%	C1q deficiency, 613652
C1QB	100%	100%	C1q deficiency, 613652
C1QBP	100%	100%	Combined oxidative phosphorylation deficiency 33, 617713
C1QC	100%	100%	C1q deficiency, 613652
C1QTNF5	100%	100%	Retinal degeneration, late-onset, autosomal dominant, 605670
C1R	100%	99%	Ehlers-Danlos syndrome, periodontal type, 1, 130080
C1S	99%	99%	C1s deficiency, 613783 Ehlers-Danlos syndrome, periodontal type, 2, 617174
C2	100%	100%	C2 deficiency, 217000
C2CD3	95%	95%	Orofaciodigital syndrome XIV, 615948
C2orf69	100%	100%	Combined oxidative phosphorylation deficiency 53, 619423
C3	100%	100%	C3 deficiency, 613779
C4A	99%	99%	C4a deficiency, 614380
C4B	99%	99%	C4B deficiency, 614379
C5	100%	100%	C5 deficiency, 609536
C6	100%	100%	C6 deficiency, 612446 Combined C6/C7 deficiency,
C7	100%	100%	C7 deficiency, 610102
C8A	100%	100%	C8 deficiency, type I, 613790
C8B	100%	100%	C8 deficiency, type II, 613789
C8G	100%	100%	No OMIM disease ID
C9	100%	100%	C9 deficiency, 613825
C9orf72	100%	100%	Frontotemporal dementia and/or amyotrophic lateral sclerosis 1, 105550
CA12	100%	100%	Hyperchlorhidrosis, isolated, 143860
CA2	100%	100%	Osteopetrosis, autosomal recessive 3, with renal tubular acidosis, 259730
CA4	100%	100%	No OMIM disease ID
CA5A	87%	87%	Hyperammonemia due to carbonic anhydrase VA deficiency, 615751
CA8	100%	100%	Cerebellar ataxia and mental retardation with or without quadrupedal locomotion 3, 613227
CABIN1	100%	100%	No OMIM disease ID
CABP2	100%	100%	Deafness, autosomal recessive 93, 614899
CABP4	100%	100%	Cone-rod synaptic disorder, congenital nonprogressive, 610427

CACNA1A	100%	100%	Spinocerebellar ataxia 6, 183086 Episodic ataxia, type 2, 108500 Developmental and epileptic encephalopathy 42, 617106 Migraine, familial hemiplegic, 1, with progressive cerebellar ataxia, 141500 Migraine, familial hemiplegic, 1, 141500
CACNA1B	100%	100%	Neurodevelopmental disorder with seizures and nonepileptic hyperkinetic movements, 618497
CACNA1C	100%	100%	Timothy syndrome, 601005 Long QT syndrome 8, 618447 Brugada syndrome 3, 611875
CACNA1D	100%	100%	Primary aldosteronism, seizures, and neurologic abnormalities, 615474 Sinoatrial node dysfunction and deafness, 614896
CACNA1E	100%	100%	Developmental and epileptic encephalopathy 69, 618285
CACNA1F	100%	100%	Cone-rod dystrophy, X-linked, 3, 300476 Night blindness, congenital stationary (incomplete), 2A, X-linked, 300071 Aland Island eye disease, 300600
CACNA1G	100%	100%	Spinocerebellar ataxia 42, 616795 Spinocerebellar ataxia 42, early-onset, severe, with neurodevelopmental deficits, 618087
CACNA1H	100%	100%	Hyperaldosteronism, familial, type IV, 617027
CACNA1I	100%	100%	No OMIM disease ID
CACNA1S	100%	100%	Hypokalemic periodic paralysis, type 1, 170400
CACNA2D1	100%	100%	No OMIM disease ID
CACNA2D2	100%	100%	Cerebellar atrophy with seizures and variable developmental delay, 618501
CACNA2D4	100%	100%	Retinal cone dystrophy 4, 610478
CACNB2	100%	100%	Brugada syndrome 4, 611876
CACNB4	100%	100%	Episodic ataxia, type 5, 613855
CACNG2	100%	100%	?Mental retardation, autosomal dominant 10, 614256

CAD	100%	100%	Developmental and epileptic encephalopathy 50, 616457
CADM3	100%	100%	Charcot-Marie-Tooth disease, axonal, type 2FF, 619519
CALCRL	100%	100%	?Lymphatic malformation 8, 618773
CALM1	100%	100%	Ventricular tachycardia, catecholaminergic polymorphic, 4, 614916 Long QT syndrome 14, 616247
CALM2	72%	72%	Long QT syndrome 15, 616249
CALM3	100%	100%	Long QT syndrome 16, 618782 ?Ventricular tachycardia, catecholaminergic polymorphic 6, 618782
CALR	100%	100%	Myelofibrosis, somatic, 254450 Thrombocythemia, somatic, 187950
CAMK2A	100%	100%	Mental retardation, autosomal dominant 53, 617798 ?Mental retardation, autosomal recessive 63, 618095
CAMK2B	100%	100%	Mental retardation, autosomal dominant 54, 617799
CAMK2G	100%	100%	Mental retardation, autosomal dominant 59, 618522
CAMK4	100%	100%	No OMIM disease ID
CAMTA1	100%	100%	Cerebellar ataxia, nonprogressive, with mental retardation, 614756
CANT1	100%	100%	Desbuquois dysplasia 1, 251450 Epiphyseal dysplasia, multiple, 7, 617719
CAPN1	100%	100%	Spastic paraplegia 76, autosomal recessive, 616907
CAPN10	100%	100%	No OMIM disease ID
CAPN12	100%	100%	No OMIM disease ID
CAPN15	100%	100%	Oculogastrointestinal neurodevelopmental syndrome, 619318
CAPN3	97%	97%	Muscular dystrophy, limb-girdle, autosomal recessive 1, 253600 Muscular dystrophy, limb-girdle, autosomal dominant 4, 618129
CAPN5	100%	100%	Vitreoretinopathy, neovascular inflammatory, 193235
CARD10	100%	100%	?Immunodeficiency 89 and autoimmunity, 619632

CARD11	100%	100%	B-cell expansion with NFKB and T-cell anergy, 616452 Immunodeficiency 11B with atopic dermatitis, 617638 Immunodeficiency 11A, 615206
CARD14	100%	100%	Psoriasis 2, 602723 Pityriasis rubra pilaris, 173200
CARD8	100%	100%	?Inflammatory bowel disease (Crohn disease) 30, 619079
CARD9	100%	100%	Candidiasis, familial, 2, autosomal recessive, 212050
CARMIL2	100%	100%	Immunodeficiency 58, 618131
CARS1	100%	100%	Microcephaly, developmental delay, and brittle hair syndrome, 618891
CARS2	100%	100%	Combined oxidative phosphorylation deficiency 27, 616672
CASK	100%	100%	Mental retardation, with or without nystagmus, 300422 Intellectual developmental disorder and microcephaly with pontine and cerebellar hypoplasia, 300749 FG syndrome 4, 300422
CASP10	100%	100%	Autoimmune lymphoproliferative syndrome, type II, 603909 Gastric cancer, somatic, 613659 Lymphoma, non-Hodgkin, somatic, 605027
CASP14	100%	100%	Ichthyosis, congenital, autosomal recessive 12, 617320
CASP8	95%	95%	Hepatocellular carcinoma, somatic, 114550 ?Autoimmune lymphoproliferative syndrome, type IIB, 607271
CASQ1	100%	100%	Myopathy, vacuolar, with CASQ1 aggregates, 616231
CASQ2	100%	100%	Ventricular tachycardia, catecholaminergic polymorphic, 2, 611938

CASR	100%	100%	Hypocalcemia, autosomal dominant, with Bartter syndrome, 601198 Hyperparathyroidism, neonatal, 239200 Hypocalcemia, autosomal dominant, 601198 Hypocalciuric hypercalcemia, type I, 145980
CAST	100%	100%	Peeling skin with leukonychia, acral punctate keratoses, cheilitis, and knuckle pads, 616295
CASZ1	99%	98%	No OMIM disease ID
CAT	100%	100%	Acatalasemia, 614097
CATIP	100%	100%	?Spermatogenic failure 54, 619379
CATSPER1	100%	100%	Spermatogenic failure 7, 612997
CATSPER2	100%	100%	No OMIM disease ID
CAV1	100%	100%	?Lipodystrophy, congenital generalized, type 3, 612526 Pulmonary hypertension, primary, 3, 615343 Lipodystrophy, familial partial, type 7, 606721
CAV3	100%	100%	Myopathy, distal, Tateyama type, 614321 Creatine phosphokinase, elevated serum, 123320 Cardiomyopathy, familial hypertrophic, 192600 Rippling muscle disease 2, 606072 Long QT syndrome 9, 611818
CAVIN1	100%	100%	Lipodystrophy, congenital generalized, type 4, 613327
CBL	100%	100%	Noonan syndrome-like disorder with or without juvenile myelomonocytic leukemia, 613563 ?Juvenile myelomonocytic leukemia, 607785
CBLB	100%	100%	No OMIM disease ID

CBLIF	100%	100%	Intrinsic factor deficiency, 261000
CBS	100%	100%	Thrombosis, hyperhomocysteinemic, 236200 Homocystinuria, B6-responsive and nonresponsive types, 236200
CBWD1	99%	98%	No OMIM disease ID
CBX2	100%	100%	?46XY sex reversal 5, 613080
CBY1	100%	100%	No OMIM disease ID
CC2D1A	100%	100%	Mental retardation, autosomal recessive 3, 608443
CC2D2A	97%	97%	COACH syndrome 2, 619111 Meckel syndrome 6, 612284 Joubert syndrome 9, 612285
CCBE1	100%	100%	Hennekam lymphangiectasia-lymphedema syndrome 1, 235510
CCDC103	100%	100%	Ciliary dyskinesia, primary, 17, 614679
CCDC115	100%	100%	Congenital disorder of glycosylation, type IIo, 616828
CCDC134	100%	100%	No OMIM disease ID
CCDC141	100%	100%	No OMIM disease ID
CCDC174	100%	100%	Hypotonia, infantile, with psychomotor retardation, 616816
CCDC186	100%	100%	No OMIM disease ID
CCDC22	100%	100%	Ritscher-Schinzel syndrome 2, 300963
CCDC28B	100%	100%	No OMIM disease ID
CCDC32	100%	100%	Cardiofacioneurodevelopmental syndrome, 619123
CCDC39	100%	100%	Ciliary dyskinesia, primary, 14, 613807
CCDC40	100%	100%	Ciliary dyskinesia, primary, 15, 613808
CCDC47	100%	100%	Trichohepatoneurodevelopmental syndrome, 618268
CCDC50	100%	100%	?Deafness, autosomal dominant 44, 607453
CCDC65	100%	100%	Ciliary dyskinesia, primary, 27, 615504
CCDC78	100%	100%	?Centronuclear myopathy 4, 614807
CCDC8	100%	100%	3-M syndrome 3, 614205
CCDC88A	97%	97%	?PEHO syndrome-like, 617507
CCDC88C	100%	100%	?Spinocerebellar ataxia 40, 616053 Hydrocephalus, congenital, 1, 236600
CCL2	100%	100%	No OMIM disease ID
CCM2	100%	100%	Cerebral cavernous malformations-2, 603284

CCN6	85%	84%	Progressive pseudorheumatoid dysplasia, 208230
CCND2	100%	100%	Megalencephaly-polymicrogyria-polydactyly-hydrocephalus syndrome 3, 615938
CCNF	100%	100%	Frontotemporal dementia and/or amyotrophic lateral sclerosis 5, 619141
CCNK	100%	99%	?Intellectual developmental disorder with hypertelorism and distinctive facies, 618147
CCNO	100%	100%	Ciliary dyskinesia, primary, 29, 615872
CCNQ	100%	99%	STAR syndrome, 300707
CCT2	100%	100%	No OMIM disease ID
CCT5	100%	100%	Neuropathy, hereditary sensory, with spastic paraplegia, 256840
CD151	100%	100%	Epidermolysis bullosa simplex 7, with nephropathy and deafness, 609057
CD164	100%	100%	?Deafness, autosomal dominant 66, 616969
CD19	100%	100%	Immunodeficiency, common variable, 3, 613493
CD247	100%	100%	?Immunodeficiency 25, 610163
CD27	100%	100%	Lymphoproliferative syndrome 2, 615122
CD28	100%	100%	No OMIM disease ID
CD2AP	100%	100%	Glomerulosclerosis, focal segmental, 3, 607832
CD320	100%	100%	Methylmalonic aciduria, transient, due to transcobalamin receptor defect, 613646
CD36	100%	100%	Platelet glycoprotein IV deficiency, 608404
CD3D	100%	100%	Immunodeficiency 19, 615617
CD3E	100%	100%	Immunodeficiency 18, 615615 Immunodeficiency 18, SCID variant, 615615
CD3G	100%	100%	Immunodeficiency 17, CD3 gamma deficient, 615607
CD4	100%	100%	Immunodeficiency 79, 619238 OKT4 epitope deficiency, 613949
CD40	100%	100%	Immunodeficiency with hyper-IgM, type 3, 606843
CD40LG	100%	100%	Immunodeficiency, X-linked, with hyper-IgM, 308230
CD46	100%	100%	No OMIM disease ID
CD48	100%	100%	No OMIM disease ID
CD55	96%	94%	Complement hyperactivation, angiopathic thrombosis, and protein-losing enteropathy, 226300
CD59	64%	64%	Hemolytic anemia, CD59-mediated, with or without immune-mediated polyneuropathy, 612300
CD70	100%	100%	Lymphoproliferative syndrome 3, 618261
CD79A	100%	100%	Agammaglobulinemia 3, 613501
CD79B	100%	100%	Agammaglobulinemia 6, 612692
CD81	100%	100%	Immunodeficiency, common variable, 6, 613496

CD8A	100%	100%	CD8 deficiency, familial, 608957
CD96	100%	100%	C syndrome, 211750
CDAN1	100%	100%	Dyserythropoietic anemia, congenital, type Ia, 224120
CDC14A	100%	100%	Deafness, autosomal recessive 32, with or without immotile sperm, 608653
CDC40	100%	100%	?Pontocerebellar hypoplasia, type 15, 619302
CDC42	100%	100%	Takenouchi-Kosaki syndrome, 616737
CDC42BPB	100%	100%	No OMIM disease ID
CDC45	100%	100%	Meier-Gorlin syndrome 7, 617063
CDC6	100%	100%	?Meier-Gorlin syndrome 5, 613805
CDC73	100%	100%	Hyperparathyroidism, familial primary, 145000 Parathyroid adenoma with cystic changes, 145001 Parathyroid carcinoma, 608266 Hyperparathyroidism-jaw tumor syndrome, 145001
CDCA7	100%	100%	Immunodeficiency-centromeric instability-facial anomalies syndrome 3, 616910
CDH1	99%	99%	Ovarian cancer, somatic, 167000 Blepharocheilodontic syndrome 1, 119580 Endometrial carcinoma, somatic, 608089 Gastric cancer, hereditary diffuse, with or without cleft lip and/or palate, 137215
CDH11	100%	100%	Elsahy-Waters syndrome, 211380
CDH15	100%	100%	Mental retardation, autosomal dominant 3, 612580
CDH2	100%	100%	Arrhythmogenic right ventricular dysplasia, familial, 14, 618920 Agenesis of corpus callosum, cardiac, ocular, and genital syndrome, 618929

CDH23	100%	100%	Usher syndrome, type 1D, 601067 Usher syndrome, type 1D/F digenic, 601067 Deafness, autosomal recessive 12, 601386
CDH3	100%	100%	Hypotrichosis, congenital, with juvenile macular dystrophy, 601553 Ectodermal dysplasia, ectrodactyly, and macular dystrophy, 225280
CDHR1	100%	100%	Cone-rod dystrophy 15, 613660 Retinitis pigmentosa 65, 613660
CDIN1	100%	100%	Dyserythropoietic anemia, congenital, type Ib, 615631
CDK10	100%	100%	Al Kaissi syndrome, 617694
CDK13	100%	100%	Congenital heart defects, dysmorphic facial features, and intellectual developmental disorder, 617360
CDK19	100%	100%	Developmental and epileptic encephalopathy 87, 618916
CDK4	100%	100%	No OMIM disease ID
CDK5	100%	100%	?Lissencephaly 7 with cerebellar hypoplasia, 616342
CDK5RAP2	100%	100%	Microcephaly 3, primary, autosomal recessive, 604804
CDK6	100%	100%	?Microcephaly 12, primary, autosomal recessive, 616080
CDK8	100%	100%	Intellectual developmental disorder with hypotonia and behavioral abnormalities, 618748
CDKL5	92%	91%	Developmental and epileptic encephalopathy 2, 300672
CDKN1A	100%	100%	No OMIM disease ID
CDKN1B	100%	100%	Multiple endocrine neoplasia, type IV, 610755
CDKN1C	100%	100%	IMAGE syndrome, 614732 Beckwith-Wiedemann syndrome, 130650
CDKN2A	100%	100%	No OMIM disease ID
CDKN2B	100%	100%	No OMIM disease ID
CDKN2C	100%	100%	No OMIM disease ID
CDON	100%	100%	Holoprosencephaly 11, 614226
CDSN	100%	100%	Hypotrichosis 2, 146520 Peeling skin syndrome 1, 270300
CDT1	100%	100%	Meier-Gorlin syndrome 4, 613804

CEACAM16	100%	100%	Deafness, autosomal dominant 4B, 614614 Deafness, autosomal recessive 113, 618410
CEBPA	100%	100%	Leukemia, acute myeloid, somatic, 601626 ?Leukemia, acute myeloid, 601626
CEBPE	100%	100%	Specific granule deficiency, 245480
CEL	100%	100%	Maturity-onset diabetes of the young, type VIII, 609812
CELA2A	100%	100%	Abdominal obesity-metabolic syndrome 4, 618620
CELF2	100%	100%	Developmental and epileptic encephalopathy 97, 619561
CELSR1	99%	99%	Lymphatic malformation 9, 619319
CENPE	100%	100%	?Microcephaly 13, primary, autosomal recessive, 616051
CENPF	100%	100%	Stromme syndrome, 243605
CENPJ	100%	100%	Microcephaly 6, primary, autosomal recessive, 608393 ?Seckel syndrome 4, 613676
CENPS	100%	100%	No OMIM disease ID
CENPT	100%	100%	?Short stature and microcephaly with genital anomalies, 618702
CEP104	100%	100%	Joubert syndrome 25, 616781
CEP112	100%	100%	Spermatogenic failure 44, 619044
CEP120	100%	100%	Short-rib thoracic dysplasia 13 with or without polydactyly, 616300 Joubert syndrome 31, 617761
CEP135	100%	100%	Microcephaly 8, primary, autosomal recessive, 614673
CEP152	100%	100%	Microcephaly 9, primary, autosomal recessive, 614852 Seckel syndrome 5, 613823
CEP164	100%	100%	Nephronophthisis 15, 614845
CEP19	100%	100%	Morbid obesity and spermatogenic failure, 615703
CEP250	100%	100%	Cone-rod dystrophy and hearing loss 2, 618358

CEP290	100%	100%	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%	Joubert syndrome 15, 614464
CEP55	100%	100%	Multinucleated neurons, anhydramnios, renal dysplasia, cerebellar hypoplasia, and hydranencephaly, 236500
CEP57	100%	100%	Mosaic variegated aneuploidy syndrome 2, 614114
CEP63	100%	100%	?Seckel syndrome 6, 614728
CEP78	100%	100%	Cone-rod dystrophy and hearing loss, 617236
CEP83	100%	100%	Nephronophthisis 18, 615862
CEP85L	100%	100%	Lissencephaly 10, 618873
CEP89	100%	100%	No OMIM disease ID
CERKL	100%	100%	Retinitis pigmentosa 26, 608380
CERS1	98%	96%	?Epilepsy, progressive myoclonic, 8, 616230
CERS3	100%	100%	Ichthyosis, congenital, autosomal recessive 9, 615023
CERT1	100%	100%	Mental retardation, autosomal dominant 34, 616351
CES1	99%	99%	Drug metabolism, altered, CES1-related, 618057
CETP	100%	100%	Hyperalphalipoproteinemia, 143470
CFAP251	100%	100%	Spermatogenic failure 33, 618152
CFAP276	100%	100%	No OMIM disease ID
CFAP298	100%	100%	Ciliary dyskinesia, primary, 26, 615500
CFAP300	100%	100%	Ciliary dyskinesia, primary, 38, 618063
CFAP410	100%	100%	Retinal dystrophy with macular staphyloma, 617547 Spondylometaphyseal dysplasia, axial, 602271
CFAP418	100%	100%	Retinitis pigmentosa 64, 614500 Cone-rod dystrophy 16, 614500 Bardet-Biedl syndrome 21, 617406
CFAP43	100%	100%	Hydrocephalus, normal pressure, 1, 236690 Spermatogenic failure 19, 617592
CFAP44	100%	100%	?Spermatogenic failure 20, 617593

CFAP47	100%	99%	Spermatogenic failure, X-linked, 3, 301059
CFAP53	100%	100%	Heterotaxy, visceral, 6, autosomal recessive, 614779
CFAP58	100%	100%	Spermatogenic failure 49, 619144
CFAP65	100%	100%	Spermatogenic failure 40, 618664
CFAP69	100%	100%	Spermatogenic failure 24, 617959
CFAP70	100%	100%	?Spermatogenic failure 41, 618670
CFAP91	100%	100%	Spermatogenic failure 51, 619177
CFB	100%	100%	?Complement factor B deficiency, 615561
CFC1	100%	100%	Heterotaxy, visceral, 2, autosomal, 605376
CFD	100%	100%	Complement factor D deficiency, 613912
CFH	100%	100%	Basal laminar drusen, 126700 Complement factor H deficiency, 609814
CFHR1	96%	94%	No OMIM disease ID
CFHR2	76%	76%	No OMIM disease ID
CFHR3	96%	95%	No OMIM disease ID
CFHR4	100%	100%	No OMIM disease ID
CFHR5	100%	100%	Nephropathy due to CFHR5 deficiency, 614809
CFI	100%	100%	Complement factor I deficiency, 610984
CFL2	100%	100%	Nemaline myopathy 7, autosomal recessive, 610687
CFP	100%	100%	Properdin deficiency, X-linked, 312060
CFTR	100%	100%	Cystic fibrosis, 219700 Congenital bilateral absence of vas deferens, 277180 Sweat chloride elevation without CF,
CHAMP1	100%	100%	Mental retardation, autosomal dominant 40, 616579
CHAT	100%	100%	Myasthenic syndrome, congenital, 6, presynaptic, 254210
CHCHD10	100%	100%	?Myopathy, isolated mitochondrial, autosomal dominant, 616209 Spinal muscular atrophy, Jokela type, 615048 Frontotemporal dementia and/or amyotrophic lateral sclerosis 2, 615911
CHCHD2	100%	100%	Parkinson disease 22, autosomal dominant, 616710

CHD1	100%	100%	Pilarowski-Bjornsson syndrome, 617682
CHD2	100%	100%	Developmental and epileptic encephalopathy 94, 615369
CHD3	100%	99%	Snijders Blok-Campeau syndrome, 618205
CHD4	100%	100%	Sifrim-Hitz-Weiss syndrome, 617159
CHD5	100%	100%	No OMIM disease ID
CHD7	100%	100%	Hypogonadotropic hypogonadism 5 with or without anosmia, 612370 CHARGE syndrome, 214800
CHD8	100%	100%	No OMIM disease ID
CHEK2	100%	100%	Osteosarcoma, somatic, 259500 Li-Fraumeni syndrome 2, 609265
CHIT1	100%	100%	No OMIM disease ID
CHKB	100%	100%	Muscular dystrophy, congenital, megaconial type, 602541
CHM	99%	97%	Choroideremia, 303100
CHMP1A	100%	100%	Pontocerebellar hypoplasia, type 8, 614961
CHMP2B	100%	100%	Frontotemporal dementia and/or amyotrophic lateral sclerosis 7, 600795
CHMP4B	100%	100%	Cataract 31, multiple types, 605387
CHN1	97%	97%	Duane retraction syndrome 2, 604356
CHP1	100%	100%	?Spastic ataxia 9, autosomal recessive, 618438
CHRDL1	100%	100%	Megalocornea 1, X-linked, 309300
CHRM1	100%	100%	No OMIM disease ID
CHRM2	100%	100%	No OMIM disease ID
CHRM3	100%	100%	Prune belly syndrome, 100100
CHRNA1	100%	100%	Myasthenic syndrome, congenital, 1B, fast-channel, 608930 Myasthenic syndrome, congenital, 1A, slow-channel, 601462 Multiple pterygium syndrome, lethal type, 253290
CHRNA2	100%	100%	Epilepsy, nocturnal frontal lobe, type 4, 610353
CHRNA3	100%	100%	Bladder dysfunction, autonomic, with impaired pupillary reflex and secondary CAKUT, 191800
CHRNA4	100%	100%	Epilepsy, nocturnal frontal lobe, 1, 600513

CHRNB1	100%	100%	?Myasthenic syndrome, congenital, 2C, associated with acetylcholine receptor deficiency, 616314 Myasthenic syndrome, congenital, 2A, slow-channel, 616313
CHRNB2	100%	100%	Epilepsy, nocturnal frontal lobe, 3, 605375
CHRND	100%	100%	?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%	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%	Multiple pterygium syndrome, lethal type, 253290 Escobar syndrome, 265000
CHST11	100%	100%	?Osteochondrodysplasia, brachydactyly, and overlapping malformed digits, 618167
CHST14	100%	100%	Ehlers-Danlos syndrome, musculocontractural type 1, 601776
CHST3	100%	100%	Spondyloepiphyseal dysplasia with congenital joint dislocations, 143095
CHST6	100%	100%	Macular corneal dystrophy, 217800
CHST8	100%	100%	?Peeling skin syndrome 3, 616265
CHSY1	99%	99%	Temtamy preaxial brachydactyly syndrome, 605282
CHUK	100%	100%	?Popliteal pterygium syndrome, Bartsocas-Papas type 2, 619339 Cocoon syndrome, 613630
CIB1	100%	100%	Epidermodysplasia verruciformis 3, 618267

CIB2	100%	100%	Deafness, autosomal recessive 48, 609439 Usher syndrome, type IJ, 614869
CIBAR1	100%	100%	?Polydactyly, postaxial, type A9, 618219
CIC	100%	100%	Mental retardation, autosomal dominant 45, 617600
CIDEC	100%	100%	?Lipodystrophy, familial partial, type 5, 615238
CIITA	100%	100%	Bare lymphocyte syndrome, type II, complementation group A, 209920
CILK1	100%	100%	Endocrine-cerebroosteodysplasia, 612651
CISD2	100%	100%	Wolfram syndrome 2, 604928
CIT	100%	100%	Microcephaly 17, primary, autosomal recessive, 617090
CITED2	100%	100%	Atrial septal defect 8, 614433 Ventricular septal defect 2, 614431
CKAP2L	100%	100%	Filippi syndrome, 272440
CLCC1	100%	100%	Retinitis pigmentosa 32, 609913
CLCF1	100%	100%	Cold-induced sweating syndrome 2, 610313
CLCN1	100%	100%	Myotonia congenita, recessive, 255700 Myotonia congenita, dominant, 160800 Myotonia levior, recessive,
CLCN2	100%	100%	Leukoencephalopathy with ataxia, 615651 Hyperaldosteronism, familial, type II, 605635
CLCN3	96%	96%	Neurodevelopmental disorder with seizures and brain abnormalities, 619517 Neurodevelopmental disorder with hypotonia and brain abnormalities, 619512
CLCN4	100%	100%	Raynaud-Claes syndrome, 300114

CLCN5	100%	100%	Proteinuria, low molecular weight, with hypercalciuric nephrocalcinosis, 308990 Hypophosphatemic rickets, 300554 Dent disease 1, 300009 Nephrolithiasis, type I, 310468
CLCN6	100%	100%	Neurodegeneration, childhood-onset, hypotonia, respiratory insufficiency and brain imaging abnormalities, 619173
CLCN7	100%	100%	Hypopigmentation, organomegaly, and delayed myelination and development, 618541 Osteopetrosis, autosomal recessive 4, 611490 Osteopetrosis, autosomal dominant 2, 166600
CLCNKA	100%	100%	Bartter syndrome, type 4b, digenic, 613090
CLCNKB	100%	100%	Bartter syndrome, type 3, 607364 Bartter syndrome, type 4b, digenic, 613090
CLDN1	100%	100%	Ichthyosis, leukocyte vacuoles, alopecia, and sclerosing cholangitis, 607626
CLDN10	100%	100%	HELIX syndrome, 617671
CLDN11	100%	100%	Leukodystrophy, hypomyelinating, 22, 619328
CLDN14	100%	100%	Deafness, autosomal recessive 29, 614035
CLDN16	100%	100%	Hypomagnesemia 3, renal, 248250
CLDN19	100%	100%	Hypomagnesemia 5, renal, with ocular involvement, 248190
CLDN2	100%	100%	?Azoospermia, obstructive, with nephrolithiasis, 301060
CLDN9	100%	100%	?Deafness, autosomal recessive 116, 619093
CLEC4D	100%	100%	No OMIM disease ID
CLEC7A	100%	100%	Candidiasis, familial, 4, autosomal recessive, 613108
CLIC2	100%	100%	?Intellectual developmental disorder, X-linked syndromic 32, 300886
CLIC5	100%	100%	?Deafness, autosomal recessive 103, 616042
CLIP1	100%	100%	No OMIM disease ID
CLMP	100%	100%	Congenital short bowel syndrome, 615237
CLN3	92%	92%	Ceroid lipofuscinosis, neuronal, 3, 204200
CLN5	71%	71%	Ceroid lipofuscinosis, neuronal, 5, 256731

CLN6	100%	100%	Ceroid lipofuscinosis, neuronal, 6B (Kufs type), 204300 Ceroid lipofuscinosis, neuronal, 6A, 601780
CLN8	100%	100%	Ceroid lipofuscinosis, neuronal, 8, Northern epilepsy variant, 610003 Ceroid lipofuscinosis, neuronal, 8, 600143
CLP1	100%	100%	Pontocerebellar hypoplasia, type 10, 615803
CLPB	100%	100%	3-methylglutaconic aciduria, type VII, with cataracts, neurologic involvement and neutropenia, 616271
CLPP	100%	100%	Perrault syndrome 3, 614129
CLPX	100%	100%	?Protoporphyria, erythropoietic, 2, 618015
CLRN1	100%	100%	Usher syndrome, type 3A, 276902 Retinitis pigmentosa 61, 614180
CLRN2	100%	100%	?Deafness, autosomal recessive 117, 619174
CLTC	100%	100%	Mental retardation, autosomal dominant 56, 617854
CLTCL1	100%	100%	No OMIM disease ID
CLUAP1	100%	100%	No OMIM disease ID
CMAS	100%	100%	No OMIM disease ID
CNBP	100%	100%	Myotonic dystrophy 2, 602668
CNGA1	91%	91%	Retinitis pigmentosa 49, 613756
CNGA3	100%	100%	Achromatopsia 2, 216900
CNGB1	100%	100%	Retinitis pigmentosa 45, 613767
CNGB3	100%	100%	Achromatopsia 3, 262300
CNKSR2	100%	100%	Intellectual developmental disorder, X-linked, syndromic, Houge type, 301008
CNNM2	100%	100%	Hypomagnesemia 6, renal, 613882 Hypomagnesemia, seizures, and mental retardation, 616418
CNNM4	100%	99%	Jalili syndrome, 217080
CNOT1	100%	100%	Vissers-Bodmer syndrome, 619033 Holoprosencephaly 12, with or without pancreatic agenesis, 618500
CNOT2	100%	100%	Intellectual developmental disorder with nasal speech, dysmorphic facies, and variable skeletal anomalies, 618608
CNOT3	100%	100%	Intellectual developmental disorder with speech delay, autism, and dysmorphic facies, 618672
CNP	100%	100%	?Leukodystrophy, hypomyelinating, 20, 619071

CNPY3	100%	100%	Developmental and epileptic encephalopathy 60, 617929
CNTN1	100%	100%	?Myopathy, congenital, Compton-North, 612540
CNTN2	100%	100%	?Epilepsy, myoclonic, familial adult, 5, 615400
CNTNAP1	100%	100%	Lethal congenital contracture syndrome 7, 616286 Hypomyelinating neuropathy, congenital, 3, 618186
CNTNAP2	100%	100%	Pitt-Hopkins like syndrome 1, 610042 Cortical dysplasia-focal epilepsy syndrome, 610042
COA1	100%	100%	No OMIM disease ID
COA3	100%	100%	?Mitochondrial complex IV deficiency, nuclear type 14, 619058
COA5	85%	85%	?Mitochondrial complex IV, deficiency, nuclear type 9, 616500
COA6	100%	100%	Mitochondrial complex IV deficiency, nuclear type 13, 616501
COA7	100%	100%	Spinocerebellar ataxia, autosomal recessive, with axonal neuropathy 3, 618387
COA8	93%	93%	Mitochondrial complex IV deficiency, nuclear type 17, 619061
COASY	100%	100%	Pontocerebellar hypoplasia, type 12, 618266 Neurodegeneration with brain iron accumulation 6, 615643
COCH	100%	100%	Deafness, autosomal dominant 9, 601369 ?Deafness, autosomal recessive 110, 618094
COG1	100%	100%	Congenital disorder of glycosylation, type IIg, 611209
COG2	100%	100%	?Congenital disorder of glycosylation, type IIq, 617395
COG4	100%	100%	Congenital disorder of glycosylation, type IIj, 613489 Saul-Wilson syndrome, 618150
COG5	100%	100%	Congenital disorder of glycosylation, type IIi, 613612
COG6	100%	100%	Shaheen syndrome, 615328 Congenital disorder of glycosylation, type III, 614576
COG7	100%	100%	Congenital disorder of glycosylation, type IIe, 608779
COG8	100%	100%	Congenital disorder of glycosylation, type IIh, 611182

COL10A1	100%	100%	Metaphyseal chondrodysplasia, Schmid type, 156500
COL11A1	100%	100%	Fibrochondrogenesis 1, 228520 Stickler syndrome, type II, 604841 Marshall syndrome, 154780 Deafness, autosomal dominant 37, 618533
COL11A2	100%	100%	Deafness, autosomal dominant 13, 601868 Otospondylomegaepiphyseal dysplasia, autosomal recessive, 215150 Fibrochondrogenesis 2, 614524 Deafness, autosomal recessive 53, 609706 Otospondylomegaepiphyseal dysplasia, autosomal dominant, 184840
COL12A1	100%	100%	Bethlem myopathy 2, 616471 ?Ullrich congenital muscular dystrophy 2, 616470
COL13A1	100%	100%	Myasthenic syndrome, congenital, 19, 616720
COL14A1	100%	100%	No OMIM disease ID
COL17A1	100%	100%	Epithelial recurrent erosion dystrophy, 122400 Epidermolysis bullosa, junctional, localisata variant, 226650 Epidermolysis bullosa, junctional, non-Herlitz type, 226650
COL18A1	100%	100%	Knobloch syndrome, type 1, 267750 Glaucoma, primary closed-angle, 618880

COL1A1	100%	100%	Osteogenesis imperfecta, type II, 166210 Caffey disease, 114000 Ehlers-Danlos syndrome, arthrochalasia type, 1, 130060 Osteogenesis imperfecta, type I, 166200 Combined osteogenesis imperfecta and Ehlers-Danlos syndrome 1, 619115 Osteogenesis imperfecta, type IV, 166220 Osteogenesis imperfecta, type III, 259420
COL1A2	100%	100%	Osteogenesis imperfecta, type III, 259420 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	99%	99%	Fibrosis of extraocular muscles, congenital, 5, 616219
COL27A1	100%	100%	Steel syndrome, 615155

COL2A1	100%	100%	?Vitreoretinopathy with phalangeal epiphyseal dysplasia, 619248 Czech dysplasia, 609162 Achondrogenesis, type II or hypochondrogenesis, 200610 Spondyloperipheral dysplasia, 271700 SMED Strudwick type, 184250 Stickler sydrome, type I, nonsyndromic ocular, 609508 ?Epiphyseal dysplasia, multiple, with myopia and deafness, 132450 SED congenita, 183900 Kniest dysplasia, 156550 Osteoarthritis with mild chondrodysplasia, 604864 Stickler syndrome, type I, 108300 Platyspondylic skeletal dysplasia, Torrance type, 151210 Spondyloepiphyseal dysplasia, Stanescu type, 616583 Avascular necrosis of the femoral head, 608805 Legg-Calve-Perthes disease, 150600
COL3A1	100%	100%	Ehlers-Danlos syndrome, vascular type, 130050 Polymicrogyria with or without vascular-type EDS, 618343

COL4A1	100%	100%	?Retinal arteries, tortuosity of, 180000 Angiopathy, hereditary, with nephropathy, aneurysms, and muscle cramps, 611773 Microangiopathy and leukoencephalopathy, pontine, autosomal dominant, 618564 Brain small vessel disease with or without ocular anomalies, 175780
COL4A2	100%	100%	Brain small vessel disease 2, 614483
COL4A3	100%	100%	Hematuria, benign familial, 141200 Alport syndrome 3, autosomal dominant, 104200 Alport syndrome 2, autosomal recessive, 203780
COL4A4	100%	100%	Hematuria, familial benign, 141200 Alport syndrome 2, autosomal recessive, 203780
COL4A5	100%	100%	Alport syndrome 1, X-linked, 301050
COL4A6	100%	100%	?Deafness, X-linked 6, 300914
COL5A1	100%	100%	Ehlers-Danlos syndrome, classic type, 1, 130000 Fibromuscular dysplasia, multifocal, 619329
COL5A2	100%	100%	Ehlers-Danlos syndrome, classic type, 2, 130010
COL6A1	100%	100%	Bethlem myopathy 1, 158810 Ullrich congenital muscular dystrophy 1, 254090
COL6A2	100%	100%	Bethlem myopathy 1, 158810 ?Myosclerosis, congenital, 255600 Ullrich congenital muscular dystrophy 1, 254090

COL6A3	100%	100%	Ullrich congenital muscular dystrophy 1, 254090 Dystonia 27, 616411 Bethlem myopathy 1, 158810
COL6A5	100%	100%	No OMIM disease ID
COL7A1	100%	100%	Epidermolysis bullosa, pretibial, 131850 Transient bullous of the newborn, 131705 EBD, Bart type, 132000 Epidermolysis bullosa dystrophica, AD, 131750 Epidermolysis bullosa pruriginosa, 604129 EBD inversa, 226600 Epidermolysis bullosa dystrophica, AR, 226600 Toenail dystrophy, isolated, 607523 EBD, localisata variant,
COL8A2	100%	100%	Corneal dystrophy, posterior polymorphous 2, 609140 Corneal dystrophy, Fuchs endothelial, 1, 136800
COL9A1	100%	100%	Stickler syndrome, type IV, 614134 ?Epiphyseal dysplasia, multiple, 6, 614135
COL9A2	100%	100%	Epiphyseal dysplasia, multiple, 2, 600204 ?Stickler syndrome, type V, 614284
COL9A3	100%	100%	Epiphyseal dysplasia, multiple, 3, with or without myopathy, 600969
COLEC10	100%	100%	3MC syndrome 3, 248340
COLEC11	100%	100%	3MC syndrome 2, 265050
COLGALT1	100%	100%	Brain small vessel disease 3, 618360
COLQ	100%	100%	Myasthenic syndrome, congenital, 5, 603034

COMP	100%	100%	Pseudoachondroplasia, 177170 Carpal tunnel syndrome 2, 619161 Epiphyseal dysplasia, multiple, 1, 132400
COMT	100%	100%	No OMIM disease ID
COPA	100%	100%	No OMIM disease ID
COPB1	100%	100%	Baralle-Macken syndrome, 619255
COPB2	100%	100%	?Microcephaly 19, primary, autosomal recessive, 617800
COQ2	97%	97%	Coenzyme Q10 deficiency, primary, 1, 607426
COQ4	100%	100%	Coenzyme Q10 deficiency, primary, 7, 616276
COQ5	100%	100%	?Coenzyme Q10 deficiency, primary, 9, 619028
COQ6	100%	100%	Coenzyme Q10 deficiency, primary, 6, 614650
COQ7	100%	100%	?Coenzyme Q10 deficiency, primary, 8, 616733
COQ8A	100%	100%	Coenzyme Q10 deficiency, primary, 4, 612016
COQ8B	100%	100%	Nephrotic syndrome, type 9, 615573
COQ9	100%	100%	Coenzyme Q10 deficiency, primary, 5, 614654
CORIN	100%	100%	Preeclampsia/eclampsia 5, 614595
CORO1A	100%	100%	Immunodeficiency 8, 615401
COX10	100%	100%	Mitochondrial complex IV deficiency, nuclear type 3, 619046
COX14	100%	100%	?Mitochondrial complex IV deficiency, nuclear type 10, 619053
COX15	100%	100%	Mitochondrial complex IV deficiency, nuclear type 6, 615119
COX16	100%	100%	Mitochondrial complex IV deficiency, nuclear type 22, 619355
COX20	100%	100%	Mitochondrial complex IV deficiency, nuclear type 11, 619054
COX4I1	100%	100%	Mitochondrial complex IV deficiency, nuclear type 16, 619060
COX4I2	100%	100%	Exocrine pancreatic insufficiency, dyserythropoietic anemia, and calvarial hyperostosis, 612714
COX5A	100%	100%	?Mitochondrial complex IV deficiency, nuclear type 20, 619064
COX5B	100%	100%	No OMIM disease ID
COX6A1	100%	100%	Charcot-Marie-Tooth disease, recessive intermediate D, 616039
COX6A2	100%	100%	Mitochondrial complex IV deficiency, nuclear type 18, 619062
COX6B1	100%	100%	Mitochondrial complex IV deficiency, nuclear type 7, 619051
COX6B2	100%	100%	No OMIM disease ID
COX6C	100%	100%	No OMIM disease ID
COX7A1	100%	100%	No OMIM disease ID
COX7A2	100%	100%	No OMIM disease ID

COX7B	100%	100%	Linear skin defects with multiple congenital anomalies 2, 300887
COX7B2	100%	100%	No OMIM disease ID
COX7C	100%	100%	No OMIM disease ID
COX8A	100%	100%	?Mitochondrial complex IV deficiency, nuclear type 15, 619059
COX8C	100%	100%	No OMIM disease ID
CP	100%	100%	Cerebellar ataxia, 604290 Hemosiderosis, systemic, due to aceruloplasminemia, 604290
CPA6	100%	100%	Febrile seizures, familial, 11, 614418 Epilepsy, familial temporal lobe, 5, 614417
CPAMD8	100%	100%	Anterior segment dysgenesis 8, 617319
CPE	100%	100%	BDV syndrome, 619326
CPLANE1	100%	100%	Orofaciodigital syndrome VI, 277170 Joubert syndrome 17, 614615
CPLX1	100%	100%	Developmental and epileptic encephalopathy 63, 617976
CPN1	100%	100%	Carboxypeptidase N deficiency, 212070
CPOX	100%	100%	Coproporphyria, 121300 Harderoporphyria, 618892
CPS1	100%	100%	Carbamoylphosphate synthetase I deficiency, 237300
CPSF1	100%	100%	Myopia 27, 618827
CPT1A	100%	100%	CPT deficiency, hepatic, type IA, 255120
CPT1C	100%	100%	?Spastic paraplegia 73, autosomal dominant, 616282
CPT2	100%	100%	CPT II deficiency, infantile, 600649 CPT II deficiency, lethal neonatal, 608836 CPT II deficiency, myopathic, stress-induced, 255110
CR2	100%	100%	Immunodeficiency, common variable, 7, 614699
CRADD	100%	100%	Mental retardation, autosomal recessive 34, with variant lissencephaly, 614499
CRAT	100%	100%	?Neurodegeneration with brain iron accumulation 8, 617917
CRB1	100%	100%	Leber congenital amaurosis 8, 613835 Retinitis pigmentosa-12, 600105 Pigmented paravenous chorioretinal atrophy, 172870

CRB2	100%	100%	Focal segmental glomerulosclerosis 9, 616220 Ventriculomegaly with cystic kidney disease, 219730
CRBN	99%	96%	Mental retardation, autosomal recessive 2, 607417
CREB1	100%	100%	Histiocytoma, angiomatoid fibrous, somatic, 612160
CREB3L1	100%	100%	Osteogenesis imperfecta, type XVI, 616229
CREB3L3	100%	100%	Hypertriglyceridemia 2, 619324
CREBBP	100%	100%	Menke-Hennekam syndrome 1, 618332 Rubinstein-Taybi syndrome 1, 180849
CRELD1	100%	100%	Atrioventricular septal defect, partial, with heterotaxy syndrome, 606217
CRIPT	100%	100%	Short stature with microcephaly and distinctive facies, 615789
CRLF1	99%	97%	Cold-induced sweating syndrome 1, 272430
CRPPA	100%	100%	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%	Osteogenesis imperfecta, type VII, 610682
CRTC1	100%	100%	Mucoepidermoid salivary gland carcinoma,
CRX	100%	100%	Leber congenital amaurosis 7, 613829 Cone-rod retinal dystrophy-2, 120970
CRYAA	100%	100%	Cataract 9, multiple types, 604219
CRYAB	100%	100%	Myopathy, myofibrillar, fatal infantile hypertonic, alpha-B crystallin-related, 613869 Myopathy, myofibrillar, 2, 608810 Cataract 16, multiple types, 613763 Cardiomyopathy, dilated, 1II, 615184
CRYBA1	100%	100%	Cataract 10, multiple types, 600881
CRYBA2	100%	100%	?Cataract 42, 115900
CRYBA4	100%	100%	Cataract 23, 610425

CRYBB1	100%	100%	Cataract 17, multiple types, 611544
CRYBB2	100%	100%	Cataract 3, multiple types, 601547
CRYBB3	100%	100%	Cataract 22, 609741
CRYGB	100%	100%	Cataract 39, multiple types, autosomal dominant, 615188
CRYGC	100%	100%	Cataract 2, multiple types, 604307
CRYGD	100%	100%	Cataract 4, multiple types, 115700
CRYGS	100%	100%	Cataract 20, multiple types, 116100
CRYL1	100%	100%	No OMIM disease ID
CRYM	100%	100%	Deafness, autosomal dominant 40, 616357
CSDE1	100%	100%	No OMIM disease ID
CSF1R	100%	100%	Brain abnormalities, neurodegeneration, and dysosteosclerosis, 618476 Leukoencephalopathy, diffuse hereditary, with spheroids, 221820
CSF2RA	95%	91%	Surfactant metabolism dysfunction, pulmonary, 4, 300770
CSF2RB	100%	100%	Surfactant metabolism dysfunction, pulmonary, 5, 614370
CSF3R	100%	100%	Neutropenia, severe congenital, 7, autosomal recessive, 617014
CSGALNACT1	100%	100%	Skeletal dysplasia, mild, with joint laxity and advanced bone age, 618870
CSNK1D	100%	100%	Advanced sleep-phase syndrome, familial, 2, 615224
CSNK1G1	100%	100%	No OMIM disease ID
CSNK2A1	94%	94%	Okur-Chung neurodevelopmental syndrome, 617062
CSNK2B	100%	100%	Poirier-Bienvenu neurodevelopmental syndrome, 618732
CSPP1	100%	100%	Joubert syndrome 21, 615636
CSRP3	100%	100%	?Cardiomyopathy, dilated, 1M, 607482 Cardiomyopathy, hypertrophic, 12, 612124
CST3	100%	100%	Cerebral amyloid angiopathy, 105150
CST6	100%	100%	?Ectodermal dysplasia 15, hypohidrotic/hair type, 618535
CSTA	100%	100%	Peeling skin syndrome 4, 607936
CSTB	100%	100%	Epilepsy, progressive myoclonic 1A (Unverricht and Lundborg), 254800
CTBP1	99%	99%	Hypotonia, ataxia, developmental delay, and tooth enamel defect syndrome, 617915
CTC1	100%	100%	Cerebroretinal microangiopathy with calcifications and cysts, 612199

CTCF	100%	100%	Mental retardation, autosomal dominant 21, 615502
CTDP1	100%	100%	Congenital cataracts, facial dysmorphism, and neuropathy, 604168
CTH	100%	100%	Cystathioninuria, 219500
CTHRC1	100%	100%	Barrett esophagus/esophageal adenocarcinoma, 614266
CTLA4	100%	100%	Immune dysregulation with autoimmunity, immunodeficiency, and lymphoproliferation, 616100
CTNNA1	100%	100%	Macular dystrophy, patterned, 2, 608970
CTNNA2	100%	100%	Cortical dysplasia, complex, with other brain malformations 9, 618174
CTNNA3	100%	100%	Arrhythmogenic right ventricular dysplasia, familial, 13, 615616
CTNNB1	100%	100%	Exudative vitreoretinopathy 7, 617572 Pilomatricoma, somatic, 132600 Colorectal cancer, somatic, 114500 Neurodevelopmental disorder with spastic diplegia and visual defects, 615075 Medulloblastoma, somatic, 155255 Ovarian cancer, somatic, 167000 Hepatocellular carcinoma, somatic, 114550
CTNNBL1	100%	100%	No OMIM disease ID
CTNND1	100%	100%	Blepharocheilodontic syndrome 2, 617681
CTNND2	100%	99%	No OMIM disease ID
CTNS	100%	100%	Cystinosis, nephropathic, 219800 Cystinosis, ocular nonnephropathic, 219750 Cystinosis, late-onset juvenile or adolescent nephropathic, 219900 Cystinosis, atypical nephropathic, 219800
CTPS1	93%	93%	Immunodeficiency 24, 615897
CTR9	100%	100%	No OMIM disease ID
CTSA	100%	100%	Galactosialidosis, 256540
CTSB	100%	100%	No OMIM disease ID
CTSC	100%	100%	Periodontitis 1, juvenile, 170650 Haim-Munk syndrome, 245010 Papillon-Lefevre syndrome, 245000

CTSD	100%	100%	Ceroid lipofuscinosis, neuronal, 10, 610127
CTSF	100%	100%	Ceroid lipofuscinosis, neuronal, 13 (Kufs type), autosomal dominant, 615362
CTSH	100%	100%	No OMIM disease ID
CTSK	100%	100%	Pycnodysostosis, 265800
CTTNBP2	100%	100%	No OMIM disease ID
CTU2	100%	100%	Microcephaly, facial dysmorphism, renal agenesis, and ambiguous genitalia syndrome, 618142
CUBN	100%	100%	Imerslund-Grasbeck syndrome 1, 261100
CUL3	100%	100%	Neurodevelopmental disorder with or without autism or seizures, 619239 Pseudohypoaldosteronism, type IIE, 614496
CUL4B	100%	99%	Mental retardation, X-linked, syndromic 15 (Cabezas type), 300354
CUL7	100%	100%	3-M syndrome 1, 273750
CUX1	100%	99%	Global developmental delay with or without impaired intellectual development, 618330
CUX2	100%	100%	Developmental and epileptic encephalopathy 67, 618141
CWC27	100%	100%	Retinitis pigmentosa with or without skeletal anomalies, 250410
CWF19L1	100%	100%	Spinocerebellar ataxia, autosomal recessive 17, 616127
CXCR2	100%	100%	?WHIM syndrome 2, 619407
CXCR4	100%	100%	WHIM syndrome 1, 193670 Myelokathexis, isolated, 193670
CYB561	100%	100%	Orthostatic hypotension 2, 618182
CYB5A	100%	100%	Methemoglobinemia and ambiguous genitalia, 250790
CYB5R3	100%	100%	Methemoglobinemia, type I, 250800 Methemoglobinemia, type II, 250800
CYBA	100%	100%	Chronic granulomatous disease 4, autosomal recessive, 233690
CYBB	100%	100%	Immunodeficiency 34, mycobacteriosis, X-linked, 300645 Chronic granulomatous disease, X-linked, 306400
CYBC1	100%	100%	Chronic granulomatous disease 5, autosomal recessive, 618935
CYBRD1	100%	100%	No OMIM disease ID
CYC1	100%	100%	Mitochondrial complex III deficiency, nuclear type 6, 615453
CYCS	100%	100%	Thrombocytopenia 4, 612004
CYFIP2	100%	100%	Developmental and epileptic encephalopathy 65, 618008

CYLD	100%	100%	Brooke-Spiegler syndrome, 605041 Cylindromatosis, familial, 132700 Trichoepithelioma, multiple familial, 1, 601606 ?Frontotemporal dementia and/or amyotrophic lateral sclerosis 8, 619132
CYP11A1	100%	100%	Adrenal insufficiency, congenital, with 46XY sex reversal, partial or complete, 613743
CYP11B1	100%	100%	Aldosteronism, glucocorticoid-remediable, 103900 Adrenal hyperplasia, congenital, due to 11-beta-hydroxylase deficiency, 202010
CYP11B2	100%	100%	Hypoaldosteronism, congenital, due to CMO I deficiency, 203400 Hypoaldosteronism, congenital, due to CMO II deficiency, 610600 Aldosterone to renin ratio raised,
CYP17A1	100%	100%	17,20-lyase deficiency, isolated, 202110 17-alpha-hydroxylase/17,20-lyase deficiency, 202110
CYP19A1	100%	100%	Aromatase deficiency, 613546 Aromatase excess syndrome, 139300
CYP1B1	100%	100%	Glaucoma 3A, primary open angle, congenital, juvenile, or adult onset, 231300 Anterior segment dysgenesis 6, multiple subtypes, 617315
CYP21A2	100%	100%	Hyperandrogenism, nonclassic type, due to 21-hydroxylase deficiency, 201910 Adrenal hyperplasia, congenital, due to 21-hydroxylase deficiency, 201910
CYP24A1	100%	100%	Hypercalcemia, infantile, 1, 143880

CYP26B1	100%	100%	Craniosynostosis with radiohumeral fusions and other skeletal and craniofacial anomalies, 614416
CYP26C1	100%	100%	Focal facial dermal dysplasia 4, 614974
CYP27A1	100%	100%	Cerebrotendinous xanthomatosis, 213700
CYP27B1	100%	100%	Vitamin D-dependent rickets, type I, 264700
CYP2A6	100%	100%	Coumarin resistance, 122700
CYP2B6	100%	100%	Efavirenz, poor metabolism of, 614546
CYP2C19	100%	100%	Proguanil poor metabolizer, 609535 Mephenytoin poor metabolizer, 609535 Clopidogrel, impaired responsiveness to, 609535 Omeprazole poor metabolizer, 609535
CYP2C8	100%	100%	No OMIM disease ID
CYP2C9	100%	100%	Warfarin sensitivity, 122700 Tolbutamide poor metabolizer,
CYP2R1	100%	100%	Rickets due to defect in vitamin D 25-hydroxylation deficiency, 600081
CYP2U1	100%	100%	Spastic paraplegia 56, autosomal recessive, 615030
CYP3A4	100%	99%	Vitamin D-dependent rickets, type 3, 619073
CYP4F22	100%	100%	Ichthyosis, congenital, autosomal recessive 5, 604777
CYP4V2	100%	100%	Bietti crystalline corneoretinal dystrophy, 210370
CYP7B1	100%	100%	Spastic paraplegia 5A, autosomal recessive, 270800 Bile acid synthesis defect, congenital, 3, 613812
D2HGDH	100%	100%	D-2-hydroxyglutaric aciduria, 600721
DAAM2	100%	100%	Nephrotic syndrome, type 24, 619263
DAB1	100%	100%	Spinocerebellar ataxia 37, 615945
DACT1	100%	100%	?Townes-Brocks syndrome 2, 617466

DAG1	100%	100%	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%	?Developmental and epileptic encephalopathy 86, 618910
DAO	100%	100%	No OMIM disease ID
DARS1	100%	100%	Hypomyelination with brainstem and spinal cord involvement and leg spasticity, 615281
DARS2	100%	100%	Leukoencephalopathy with brain stem and spinal cord involvement and lactate elevation, 611105
DBF4	100%	100%	No OMIM disease ID
DBH	100%	100%	Orthostatic hypotension 1, due to DBH deficiency, 223360
DBR1	100%	100%	No OMIM disease ID
DBT	100%	100%	Maple syrup urine disease, type II, 248600
DCAF17	100%	100%	Woodhouse-Sakati syndrome, 241080
DCAF8	100%	100%	?Giant axonal neuropathy 2, autosomal dominant, 610100
DCC	100%	100%	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%	Nephronophthisis 19, 616217 ?Deafness, autosomal recessive 66, 610212 Sclerosing cholangitis, neonatal, 617394
DCHS1	100%	100%	Mitral valve prolapse 2, 607829 Van Maldergem syndrome 1, 601390
DCLRE1C	100%	100%	Severe combined immunodeficiency, Athabascan type, 602450 Omenn syndrome, 603554
DCN	95%	95%	Corneal dystrophy, congenital stromal, 610048

DCPS	100%	100%	Al-Raqad syndrome, 616459
DCT	100%	100%	Oculocutaneous albinism, type VIII, 619165
DCTN1	100%	100%	Neuronopathy, distal hereditary motor, type VIIB, 607641 Perry syndrome, 168605
DCTN2	100%	100%	No OMIM disease ID
DCX	100%	100%	Subcortical laminal heterotopia, X-linked, 300067 Lissencephaly, X-linked, 300067
DCXR	100%	100%	No OMIM disease ID
DDB1	100%	100%	White-Kernohan syndrome, 619426
DDB2	100%	100%	Xeroderma pigmentosum, group E, DDB-negative subtype, 278740
DDC	100%	100%	Aromatic L-amino acid decarboxylase deficiency, 608643
DDHD1	100%	100%	Spastic paraplegia 28, autosomal recessive, 609340
DDHD2	100%	100%	Spastic paraplegia 54, autosomal recessive, 615033
DDOST	100%	100%	?Congenital disorder of glycosylation, type Ir, 614507
DDR2	100%	100%	Warburg-Cinotti syndrome, 618175 Spondylometaepiphyseal dysplasia, short limb-hand type, 271665
DDRGK1	100%	100%	Spondyloepimetaphyseal dysplasia, Shohat type, 602557
DDX11	100%	100%	Warsaw breakage syndrome, 613398
DDX23	100%	100%	No OMIM disease ID
DDX3X	99%	97%	Intellectual developmental disorder, X-linked, syndrome, Snijders Blok type, 300958
DDX41	100%	100%	No OMIM disease ID
DDX58	100%	100%	Singleton-Merten syndrome 2, 616298
DDX59	100%	100%	Orofaciodigital syndrome V, 174300
DDX6	100%	100%	Intellectual developmental disorder with impaired language and dysmorphic facies, 618653
DEAF1	100%	100%	Vulto-van Silfout-de Vries syndrome, 615828 Neurodevelopmental disorder with hypotonia, impaired expressive language, and with or without seizures, 617171
DEF6	100%	100%	Immunodeficiency 87 and autoimmunity, 619573
DEGS1	100%	100%	Leukodystrophy, hypomyelinating, 18, 618404

DENND5A	100%	100%	Developmental and epileptic encephalopathy 49, 617281
DEPDC5	100%	100%	Epilepsy, familial focal, with variable foci 1, 604364
DES	100%	100%	Scapuloperoneal syndrome, neurogenic, Kaeser type, 181400 Cardiomyopathy, dilated, 1I, 604765 Myopathy, myofibrillar, 1, 601419
DGAT1	100%	100%	?Diarrhea 7, protein-losing enteropathy type, 615863
DGAT2	100%	100%	No OMIM disease ID
DGKE	100%	100%	Nephrotic syndrome, type 7, 615008
DGUOK	100%	100%	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%	97%	Desmosterolosis, 602398
DHCR7	100%	100%	Smith-Lemli-Opitz syndrome, 270400
DHDDS	95%	95%	Developmental delay and seizures with or without movement abnormalities, 617836 ?Congenital disorder of glycosylation, type 1bb, 613861 Retinitis pigmentosa 59, 613861
DHFR	100%	100%	Megaloblastic anemia due to dihydrofolate reductase deficiency, 613839
DHH	100%	100%	46XY gonadal dysgenesis with minifascicular neuropathy, 607080 46XY sex reversal 7, 233420
DHODH	100%	100%	Miller syndrome, 263750
DHPS	93%	93%	Neurodevelopmental disorder with seizures and speech and walking impairment, 618480
DHTKD1	100%	100%	?Charcot-Marie-Tooth disease, axonal, type 2Q, 615025 Alpha-aminoadipic and alpha-ketoadipic aciduria, 204750

DHX16	100%	100%	Neuromuscular disease and ocular or auditory anomalies with or without seizures, 618733
DHX30	100%	100%	Neurodevelopmental disorder with severe motor impairment and absent language, 617804
DHX37	100%	100%	Neurodevelopmental disorder with brain anomalies and with or without vertebral or cardiac anomalies, 618731 46, XY sex reversal 11, 273250
DHX38	100%	100%	Retinitis pigmentosa 84, 618220
DIABLO	100%	100%	Deafness, autosomal dominant 64, 614152
DIAPH1	100%	99%	Deafness, autosomal dominant 1, with or without thrombocytopenia, 124900 Seizures, cortical blindness, microcephaly syndrome, 616632
DIAPH2	100%	99%	?Premature ovarian failure 2A, 300511
DIAPH3	100%	100%	Auditory neuropathy, autosomal dominant, 1, 609129
DICER1	100%	100%	Pleuropulmonary blastoma, 601200 Goiter, multinodular 1, with or without Sertoli-Leydig cell tumors, 138800 GLOW syndrome, somatic mosaic, 618272 Rhabdomyosarcoma, embryonal, 2, 180295
DIP2B	100%	100%	Mental retardation, FRA12A type, 136630
DIS3L2	100%	100%	Perlman syndrome, 267000
DISP1	100%	100%	No OMIM disease ID
DKC1	100%	100%	Dyskeratosis congenita, X-linked, 305000
DLAT	100%	100%	Pyruvate dehydrogenase E2 deficiency, 245348
DLC1	100%	100%	Colorectal cancer, somatic, 114500
DLD	100%	100%	Dihydrolipoamide dehydrogenase deficiency, 246900
DLG3	100%	100%	Intellectual developmental disorder, X-linked 90, 300850
DLG4	98%	98%	Intellectual developmental disorder 62, 618793
DLL1	100%	100%	Neurodevelopmental disorder with nonspecific brain abnormalities and with or without seizures, 618709
DLL3	100%	100%	Spondylocostal dysostosis 1, autosomal recessive, 277300

DLL4	100%	100%	Adams-Oliver syndrome 6, 616589
DLST	100%	100%	Paragangliomas 7, 618475
DLX3	100%	100%	Trichodontoosseous syndrome, 190320 Amelogenesis imperfecta, type IV, 104510
DLX4	100%	100%	?Orofacial cleft 15, 616788
DLX5	100%	100%	Split-hand/foot malformation 1, 183600 ?Split-hand/foot malformation 1 with sensorineural hearing loss, 220600
DLX6	100%	100%	No OMIM disease ID
DMAC1	100%	100%	No OMIM disease ID
DMAC2	100%	100%	No OMIM disease ID
DMAC2L	100%	100%	No OMIM disease ID
DMC1	100%	100%	No OMIM disease ID
DMD	100%	100%	Becker muscular dystrophy, 300376 Cardiomyopathy, dilated, 3B, 302045 Duchenne muscular dystrophy, 310200
DMGDH	100%	100%	Dimethylglycine dehydrogenase deficiency, 605850
DMP1	100%	100%	Hypophosphatemic rickets, AR, 241520
DMPK	100%	100%	Myotonic dystrophy 1, 160900
DMRT1	100%	100%	No OMIM disease ID
DMRT2	100%	100%	No OMIM disease ID
DMXL2	100%	100%	Developmental and epileptic encephalopathy 81, 618663 ?Deafness, autosomal dominant 71, 617605 ?Polyendocrine-polyneuropathy syndrome, 616113
DNA2	100%	100%	?Seckel syndrome 8, 615807 Progressive external ophthalmoplegia with mitochondrial DNA deletions, autosomal dominant 6, 615156

DNAAF1	100%	100%	Ciliary dyskinesia, primary, 13, 613193
DNAAF11	100%	100%	Ciliary dyskinesia, primary, 19, 614935
DNAAF2	100%	100%	Ciliary dyskinesia, primary, 10, 612518
DNAAF3	100%	100%	Ciliary dyskinesia, primary, 2, 606763
DNAAF4	100%	100%	Ciliary dyskinesia, primary, 25, 615482
DNAAF5	100%	99%	Ciliary dyskinesia, primary, 18, 614874
DNAAF6	100%	100%	Ciliary dyskinesia, primary, 36, X-linked, 300991
DNAH1	100%	100%	Spermatogenic failure 18, 617576 ?Ciliary dyskinesia, primary, 37, 617577
DNAH10	100%	100%	Spermatogenic failure 56, 619515
DNAH11	100%	100%	Ciliary dyskinesia, primary, 7, with or without situs inversus, 611884
DNAH17	100%	100%	Spermatogenic failure 39, 618643
DNAH2	99%	99%	Spermatogenic failure 45, 619094
DNAH5	100%	100%	Ciliary dyskinesia, primary, 3, with or without situs inversus, 608644
DNAH8	100%	100%	Spermatogenic failure 46, 619095
DNAH9	100%	100%	Ciliary dyskinesia, primary, 40, 618300
DNAI1	100%	100%	Ciliary dyskinesia, primary, 1, with or without situs inversus, 244400
DNAI2	100%	100%	Ciliary dyskinesia, primary, 9, with or without situs inversus, 612444
DNAJA3	100%	100%	No OMIM disease ID
DNAJB11	100%	100%	Polycystic kidney disease 6 with or without polycystic liver disease, 618061
DNAJB13	100%	100%	Ciliary dyskinesia, primary, 34, 617091
DNAJB2	100%	100%	Spinal muscular atrophy, distal, autosomal recessive, 5, 614881
DNAJB5	100%	100%	No OMIM disease ID
DNAJB6	100%	100%	Muscular dystrophy, limb-girdle, autosomal dominant 1, 603511
DNAJC12	100%	100%	Hyperphenylalaninemia, mild, non-BH4-deficient, 617384
DNAJC19	100%	100%	3-methylglutaconic aciduria, type V, 610198
DNAJC21	100%	100%	Bone marrow failure syndrome 3, 617052
DNAJC3	100%	100%	?Ataxia, combined cerebellar and peripheral, with hearing loss and diabetes mellitus, 616192
DNAJC30	100%	100%	Leber hereditary optic neuropathy, autosomal recessive, 619382
DNAJC5	100%	100%	Ceroid lipofuscinosis, neuronal, 4 (Kufs type), 162350

DNAJC6	100%	100%	Parkinson disease 19a, juvenile-onset, 615528 Parkinson disease 19b, early-onset, 615528
DNAL1	100%	100%	Ciliary dyskinesia, primary, 16, 614017
DNAL4	100%	100%	?Mirror movements 3, 616059
DNASE1	100%	100%	No OMIM disease ID
DNASE1L3	100%	100%	Systemic lupus erythematosus 16, 614420
DNASE2	100%	100%	No OMIM disease ID
DNM1	97%	97%	Developmental and epileptic encephalopathy 31, 616346
DNM1L	100%	100%	Optic atrophy 5, 610708 Encephalopathy, lethal, due to defective mitochondrial peroxisomal fission 1, 614388
DNM2	100%	100%	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%	Cataract 48, 618415
DNMT1	100%	99%	Neuropathy, hereditary sensory, type IE, 614116 Cerebellar ataxia, deafness, and narcolepsy, autosomal dominant, 604121
DNMT3A	100%	100%	Tatton-Brown-Rahman syndrome, 615879 Acute myeloid leukemia, somatic, 601626 Heyn-Sproul-Jackson syndrome, 618724

DNMT3B	100%	100%	Immunodeficiency-centromeric instability-facial anomalies syndrome 1, 242860 Facioscapulohumeral muscular dystrophy 4, digenic, 619478
DOCK2	100%	100%	Immunodeficiency 40, 616433
DOCK3	100%	100%	Neurodevelopmental disorder with impaired intellectual development, hypotonia, and ataxia, 618292
DOCK6	100%	100%	Adams-Oliver syndrome 2, 614219
DOCK7	100%	100%	Developmental and epileptic encephalopathy 23, 615859
DOCK8	100%	100%	Hyper-IgE recurrent infection syndrome, autosomal recessive, 243700
DOK7	100%	100%	Fetal akinesia deformation sequence 3, 618389 Myasthenic syndrome, congenital, 10, 254300
DOLK	100%	100%	Congenital disorder of glycosylation, type Im, 610768
DONSON	100%	100%	Microcephaly, short stature, and limb abnormalities, 617604 Microcephaly-micromelia syndrome, 251230
DOT1L	100%	100%	No OMIM disease ID
DPAGT1	100%	100%	Myasthenic syndrome, congenital, 13, with tubular aggregates, 614750 Congenital disorder of glycosylation, type Ij, 608093
DPCD	100%	100%	No OMIM disease ID
DPF2	100%	100%	Coffin-Siris syndrome 7, 618027
DPH1	100%	100%	Developmental delay with short stature, dysmorphic facial features, and sparse hair, 616901
DPM1	99%	97%	Congenital disorder of glycosylation, type Ie, 608799
DPM2	100%	100%	Congenital disorder of glycosylation, type Iu, 615042

DPM3	100%	100%	?Muscular dystrophy-dystroglycanopathy (congenital with impaired intellectual development), type B, 15, 618992 Muscular dystrophy-dystroglycanopathy (limb-girdle), type C, 15, 612937
DPP6	100%	99%	Mental retardation, autosomal dominant 33, 616311
DPY19L2	100%	100%	Spermatogenic failure 9, 613958
DPYD	100%	100%	Dihydropyrimidine dehydrogenase deficiency, 274270 5-fluorouracil toxicity, 274270
DPYS	100%	100%	Dihydropyrimidinuria, 222748
DPYSL5	100%	100%	Ritscher-Schinzel syndrome 4, 619435
DRAM2	100%	100%	Cone-rod dystrophy 21, 616502
DRC1	100%	100%	Ciliary dyskinesia, primary, 21, 615294
DRD4	100%	100%	Autonomic nervous system dysfunction,
DRP2	100%	100%	No OMIM disease ID
DSC2	100%	100%	Arrhythmogenic right ventricular dysplasia 11 with mild palmoplantar keratoderma and woolly hair, 610476 Arrhythmogenic right ventricular dysplasia 11, 610476
DSC3	100%	100%	Hypotrichosis and recurrent skin vesicles, 613102
DSE	100%	100%	Ehlers-Danlos syndrome, musculocontractural type 2, 615539
DSG1	100%	100%	Keratosis palmoplantaris striata I, AD, 148700 Erythroderma, congenital, with palmoplantar keratoderma, hypotrichosis, and hyper IgE, 615508
DSG2	100%	100%	Cardiomyopathy, dilated, 1BB, 612877 Arrhythmogenic right ventricular dysplasia 10, 610193
DSG3	100%	100%	Blistering, acantholytic, of oral and laryngeal mucosa, 619226
DSG4	100%	100%	Hypotrichosis 6, 607903

DSP	100%	100%	Arrhythmogenic right ventricular dysplasia 8, 607450 Skin fragility-woolly hair syndrome, 607655 Epidermolysis bullosa, lethal acantholytic, 609638 Keratosis palmoplantaris striata II, 612908 Dilated cardiomyopathy with woolly hair, keratoderma, and tooth agenesis, 615821 Cardiomyopathy, dilated, with woolly hair and keratoderma, 605676
DSPP	100%	100%	Dentinogenesis imperfecta, Shields type III, 125500 Dentinogenesis imperfecta, Shields type II, 125490 Dentin dysplasia, type II, 125420 Deafness, autosomal dominant 39, with dentinogenesis, 605594
DST	95%	95%	?Neuropathy, hereditary sensory and autonomic, type VI, 614653 Epidermolysis bullosa simplex 3, localized or generalized intermediate, with bp230 deficiency, 615425
DSTYK	100%	100%	Congenital anomalies of kidney and urinary tract 1, 610805 Spastic paraplegia 23, 270750
DTNA	100%	100%	Left ventricular noncompaction 1, with or without congenital heart defects, 604169
DTNBP1	100%	100%	Hermansky-Pudlak syndrome 7, 614076
DTYMK	100%	100%	No OMIM disease ID
DUOX2	100%	100%	Thyroid dyshormonogenesis 6, 607200
DUOXA2	100%	100%	Thyroid dyshormonogenesis 5, 274900
DUSP6	100%	100%	Hypogonadotropic hypogonadism 19 with or without anosmia, 615269
DVL1	100%	100%	Robinow syndrome, autosomal dominant 2, 616331

DVL3	100%	100%	Robinow syndrome, autosomal dominant 3, 616894
DYM	100%	100%	Smith-McCort dysplasia, 607326 Dyggve-Melchior-Clausen disease, 223800
DYNC1H1	100%	100%	Spinal muscular atrophy, lower extremity-predominant 1, AD, 158600 Charcot-Marie-Tooth disease, axonal, type 20, 614228 Mental retardation, autosomal dominant 13, 614563
DYNC1I2	100%	100%	Neurodevelopmental disorder with microcephaly and structural brain anomalies, 618492
DYNC2H1	100%	100%	Short-rib thoracic dysplasia 3 with or without polydactyly, 613091
DYNC2I1	100%	100%	Short-rib thoracic dysplasia 8 with or without polydactyly, 615503
DYNC2I2	100%	100%	Short-rib thoracic dysplasia 11 with or without polydactyly, 615633
DYNC2LI1	100%	100%	Short-rib thoracic dysplasia 15 with polydactyly, 617088
DYNLT2B	100%	100%	Short-rib thoracic dysplasia 17 with or without polydactyly, 617405
DYRK1A	100%	100%	Mental retardation, autosomal dominant 7, 614104
DYRK1B	100%	100%	Abdominal obesity-metabolic syndrome 3, 615812
DYSF	100%	100%	Muscular dystrophy, limb-girdle, autosomal recessive 2, 253601 Miyoshi muscular dystrophy 1, 254130 Myopathy, distal, with anterior tibial onset, 606768
DZIP1	100%	100%	Spermatogenic failure 47, 619102 ?Mitral valve prolapse 3, 610840
DZIP1L	100%	100%	Polycystic kidney disease 5, 617610
E2F1	99%	99%	No OMIM disease ID
EARS2	100%	100%	Combined oxidative phosphorylation deficiency 12, 614924
EBF3	100%	100%	Hypotonia, ataxia, and delayed development syndrome, 617330
EBP	100%	100%	MEND syndrome, 300960 Chondrodysplasia punctata, X-linked dominant, 302960
ECE1	90%	90%	?Hirschsprung disease, cardiac defects, and autonomic dysfunction, 613870
ECEL1	100%	100%	Arthrogryposis, distal, type 5D, 615065

ECHS1	100%	100%	Mitochondrial short-chain enoyl-CoA hydratase 1 deficiency, 616277
ECM1	100%	100%	Urbach-Wiethe disease, 247100
ECSIT	100%	100%	No OMIM disease ID
EDA	100%	100%	Tooth agenesis, selective, X-linked 1, 313500 Ectodermal dysplasia 1, hypohidrotic, X-linked, 305100
EDAR	100%	100%	Ectodermal dysplasia 10A, hypohidrotic/hair/nail type, autosomal dominant, 129490 Ectodermal dysplasia 10B, hypohidrotic/hair/tooth type, autosomal recessive, 224900
EDARADD	100%	100%	Ectodermal dysplasia 11B, hypohidrotic/hair/tooth type, autosomal recessive, 614941 Ectodermal dysplasia 11A, hypohidrotic/hair/tooth type, autosomal dominant, 614940
EDC3	100%	100%	?Intellectual developmental disorder, autosomal recessive 50, 616460
EDEM3	100%	100%	Congenital disorder of glycosylation, type 2V, 619493
EDN1	100%	100%	Question mark ears, isolated, 612798 Auriculocondylar syndrome 3, 615706
EDN3	100%	100%	Waardenburg syndrome, type 4B, 613265
EDNRA	100%	100%	Mandibulofacial dysostosis with alopecia, 616367
EDNRB	100%	100%	ABCD syndrome, 600501 Waardenburg syndrome, type 4A, 277580
EED	100%	100%	Cohen-Gibson syndrome, 617561
EEF1A2	100%	100%	Mental retardation, autosomal dominant 38, 616393 Developmental and epileptic encephalopathy 33, 616409
EEF2	100%	100%	?Spinocerebellar ataxia 26, 609306
EFEMP1	100%	100%	Doyne honeycomb degeneration of retina, 126600

EFEMP2	100%	100%	Cutis laxa, autosomal recessive, type IB, 614437
EFHC1	98%	98%	No OMIM disease ID
EFL1	100%	100%	Shwachman-Diamond syndrome 2, 617941
EFNA4	100%	100%	No OMIM disease ID
EFNB1	100%	100%	Craniofrontonasal dysplasia, 304110
EFNB2	100%	100%	No OMIM disease ID
EFTUD2	100%	100%	Mandibulofacial dysostosis, Guion-Almeida type, 610536
EGF	100%	100%	?Hypomagnesemia 4, renal, 611718
EGFR	100%	100%	?Inflammatory skin and bowel disease, neonatal, 2, 616069 Nonsmall cell lung cancer, response to tyrosine kinase inhibitor in, 211980 Adenocarcinoma of lung, response to tyrosine kinase inhibitor in, 211980
EGLN1	100%	100%	Erythrocytosis, familial, 3, 609820
EGLN2	100%	100%	No OMIM disease ID
EGR2	100%	100%	Dejerine-Sottas disease, 145900 Charcot-Marie-Tooth disease, type 1D, 607678 Hypomyelinating neuropathy, congenital, 1, 605253
EHHADH	100%	100%	?Fanconi renotubular syndrome 3, 615605
EHMT1	99%	99%	Kleefstra syndrome 1, 610253
EIF2AK1	100%	100%	?Leukoencephalopathy, motor delay, spasticity, and dysarthria syndrome, 618878
EIF2AK2	100%	100%	Leukoencephalopathy, developmental delay, and episodic neurologic regression syndrome, 618877
EIF2AK3	100%	100%	Wolcott-Rallison syndrome, 226980
EIF2AK4	100%	100%	Pulmonary venoocclusive disease 2, 234810
EIF2B1	100%	100%	Leukoencephalopathy with vanishing white matter, 603896
EIF2B2	100%	100%	Leukoencephalopathy with vanishing white matter, 603896 Ovarioleukodystrophy, 603896
EIF2B3	100%	100%	Leukoencephalopathy with vanishing white matter, 603896
EIF2B4	100%	100%	Ovarioleukodystrophy, 603896 Leukoencephalopathy with vanishing white matter, 603896

EIF2B5	100%	100%	Ovarioleukodystrophy, 603896 Leukoencephalopathy with vanishing white matter, 603896
EIF2S3	100%	100%	MEHMO syndrome, 300148
EIF3F	100%	100%	Intellectual developmental disorder, autosomal recessive 67, 618295
EIF4A3	100%	100%	Robin sequence with cleft mandible and limb anomalies, 268305
EIF5A	100%	100%	Faundes-Banka syndrome, 619376
ELAC2	100%	100%	Combined oxidative phosphorylation deficiency 17, 615440
ELANE	100%	100%	Neutropenia, cyclic, 162800 Neutropenia, severe congenital 1, autosomal dominant, 202700
ELF2	100%	100%	No OMIM disease ID
ELF4	100%	100%	No OMIM disease ID
ELMO2	100%	100%	Vascular malformation, primary intraosseous, 606893
ELMOD3	100%	100%	?Deafness, autosomal recessive 88, 615429 ?Deafness, autosomal dominant 81, 619500
ELN	100%	100%	Cutis laxa, autosomal dominant, 123700 Supravalvar aortic stenosis, 185500
ELOVL1	100%	100%	Ichthyotic keratoderma, spasticity, hypomyelination, and dysmorphic facies, 618527
ELOVL4	100%	100%	Spinocerebellar ataxia 34, 133190 Stargardt disease 3, 600110 Ichthyosis, spastic quadriplegia, and mental retardation, 614457
ELOVL5	100%	100%	Spinocerebellar ataxia 38, 615957
ELP1	100%	100%	Dysautonomia, familial, 223900 Medulloblastoma, 155255
ELP2	100%	100%	Mental retardation, autosomal recessive 58, 617270
ELP4	87%	87%	?Aniridia 2, 617141
EMC1	100%	100%	Cerebellar atrophy, visual impairment, and psychomotor retardation, 616875
EMC10	100%	100%	Neurodevelopmental disorder with dysmorphic facies and variable seizures, 619264
EMD	100%	100%	Emery-Dreifuss muscular dystrophy 1, X-linked, 310300
EMG1	100%	100%	Bowen-Conradi syndrome, 211180

EMILIN1	100%	100%	No OMIM disease ID
EML1	100%	100%	Band heterotopia, 600348
EMP2	100%	100%	Nephrotic syndrome, type 10, 615861
EMX2	100%	100%	Schizencephaly, 269160
EN1	100%	100%	?ENDOVE syndrome, limb-brain type, 619218
ENAM	100%	100%	Amelogenesis imperfecta, type IC, 204650 Amelogenesis imperfecta, type IB, 104500
ENG	100%	100%	Telangiectasia, hereditary hemorrhagic, type 1, 187300
ENO3	100%	100%	Glycogen storage disease XIII, 612932
ENPP1	99%	99%	Hypophosphatemic rickets, autosomal recessive, 2, 613312 Arterial calcification, generalized, of infancy, 1, 208000 Cole disease, 615522
ENTPD1	100%	100%	Spastic paraplegia 64, autosomal recessive, 615683
EOGT	94%	90%	Adams-Oliver syndrome 4, 615297
EP300	100%	100%	Menke-Hennekam syndrome 2, 618333 Colorectal cancer, somatic, 114500 Rubinstein-Taybi syndrome 2, 613684
EPAS1	100%	100%	Erythrocytosis, familial, 4, 611783
EPB41	100%	100%	Elliptocytosis-1, 611804
EPB41L1	97%	97%	?Intellectual developmental disorder, autosomal dominant 11, 614257
EPB42	100%	100%	Spherocytosis, type 5, 612690
EPCAM	100%	100%	Colorectal cancer, hereditary nonpolyposis, type 8, 613244 Diarrhea 5, with tufting enteropathy, congenital, 613217
EPG5	100%	100%	Vici syndrome, 242840
EPHA2	100%	100%	Cataract 6, multiple types, 116600
EPHA7	100%	100%	No OMIM disease ID
EPHB2	99%	99%	?Bleeding disorder, platelet-type, 22, 618462

EPHB4	100%	100%	Capillary malformation-arteriovenous malformation 2, 618196 Lymphatic malformation 7, 617300
EPHX1	100%	100%	No OMIM disease ID
EPHX2	100%	100%	No OMIM disease ID
EPM2A	100%	100%	Epilepsy, progressive myoclonic 2A (Lafora), 254780
EPO	100%	100%	Erythrocytosis, familial, 5, 617907 ?Diamond-Blackfan anemia-like, 617911
EPRS1	100%	100%	Leukodystrophy, hypomyelinating, 15, 617951
EPS8	100%	100%	?Deafness, autosomal recessive 102, 615974
EPS8L2	88%	88%	Deafness autosomal recessive 106, 617637
EPS8L3	100%	100%	?Hypotrichosis 5, 612841
ERAL1	100%	100%	Perrault syndrome 6, 617565
ERBB2	100%	100%	Gastric cancer, somatic, 613659 Adenocarcinoma of lung, somatic, 211980 Ovarian cancer, somatic, 167000 ?Visceral neuropathy, familial, 2, autosomal recessive, 619465 Glioblastoma, somatic, 137800
ERBB3	100%	100%	?Lethal congenital contractural syndrome 2, 607598 Visceral neuropathy, familial, 1, autosomal recessive, 243180
ERBB4	100%	100%	Amyotrophic lateral sclerosis 19, 615515
ERCC1	100%	100%	Cerebrooculofacioskeletal syndrome 4, 610758
ERCC2	100%	100%	Xeroderma pigmentosum, group D, 278730 Trichothiodystrophy 1, photosensitive, 601675 ?Cerebrooculofacioskeletal syndrome 2, 610756

ERCC3	100%	100%	Trichothiodystrophy 2, photosensitive, 616390 Xeroderma pigmentosum, group B, 610651
ERCC4	100%	100%	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%	Xeroderma pigmentosum, group G, 278780 Cerebrooculofacioskeletal syndrome 3, 616570 Xeroderma pigmentosum, group G/Cockayne syndrome, 278780
ERCC6	100%	100%	UV-sensitive syndrome 1, 600630 Cerebrooculofacioskeletal syndrome 1, 214150 Cockayne syndrome, type B, 133540 De Sanctis-Cacchione syndrome, 278800 Premature ovarian failure 11, 616946
ERCC6L2	100%	100%	Bone marrow failure syndrome 2, 615715
ERCC8	100%	100%	UV-sensitive syndrome 2, 614621 Cockayne syndrome, type A, 216400
ERF	100%	100%	Craniosynostosis 4, 600775 Chitayat syndrome, 617180
ERGIC1	98%	98%	?Arthrogryposis multiplex congenita 2, neurogenic type, 208100
ERLIN1	100%	100%	Spastic paraplegia 62, 615681
ERLIN2	100%	100%	Spastic paraplegia 18, autosomal recessive, 611225
ERMARD	100%	100%	?Periventricular nodular heterotopia 6, 615544

ESCO2	100%	100%	Juberg-Hayward syndrome, 216100 Roberts-SC phocomelia syndrome, 268300
ESPN	100%	100%	Deafness, neurosensory, without vestibular involvement, autosomal dominant, 609006 Deafness, autosomal recessive 36, 609006 ?Usher syndrome, type 1M, 618632
ESR1	100%	100%	Breast cancer, somatic, 114480 Estrogen resistance, 615363
ESR2	100%	100%	?Ovarian dysgenesis 8, 618187
ESRP1	100%	100%	?Deafness, autosomal recessive 109, 618013
ESRRB	100%	100%	Deafness, autosomal recessive 35, 608565
ETFA	100%	100%	Glutaric acidemia IIA, 231680
ETFB	100%	100%	Glutaric acidemia IIB, 231680
ETFDH	100%	100%	Glutaric acidemia IIC, 231680
ETHE1	100%	100%	Ethylmalonic encephalopathy, 602473
ETV6	100%	100%	Thrombocytopenia 5, 616216 Leukemia, acute myeloid, somatic, 601626
EVC	99%	99%	Ellis-van Creveld syndrome, 225500 ?Weyers acrofacial dysostosis, 193530
EVC2	100%	100%	Ellis-van Creveld syndrome, 225500 Weyers acrofacial dysostosis, 193530
EWSR1	100%	100%	Neuroepithelioma, 612219 Ewing sarcoma, 612219
EXOC2	100%	100%	Neurodevelopmental disorder with dysmorphic facies and cerebellar hypoplasia, 619306
EXOC6	100%	100%	No OMIM disease ID
EXOC6B	100%	100%	Spondyloepimetaphyseal dysplasia with joint laxity, type 3, 618395
EXOC7	100%	100%	Neurodevelopmental disorder with seizures and brain atrophy, 619072
EXOC8	100%	100%	?Neurodevelopmental disorder with microcephaly, seizures, and brain atrophy, 619076
EXOSC1	100%	100%	?Pontocerebellar hypoplasia, type 1F, 619304
EXOSC2	100%	100%	Short stature, hearing loss, retinitis pigmentosa, and distinctive facies, 617763
EXOSC3	100%	100%	Pontocerebellar hypoplasia, type 1B, 614678

EXOSC5	100%	100%	Cerebellar ataxia, brain abnormalities, and cardiac conduction defects, 619576
EXOSC8	100%	100%	Pontocerebellar hypoplasia, type 1C, 616081
EXOSC9	100%	100%	Pontocerebellar hypoplasia, type 1D, 618065
EXPH5	100%	100%	Epidermolysis bullosa simplex 4, localized or generalized intermediate, autosomal recessive, 615028
EXT1	100%	100%	Exostoses, multiple, type 1, 133700 Chondrosarcoma, 215300
EXT2	100%	100%	Seizures, scoliosis, and macrocephaly syndrome, 616682 Exostoses, multiple, type 2, 133701
EXTL3	100%	100%	Immunoskeletal dysplasia with neurodevelopmental abnormalities, 617425
EYA1	100%	100%	Branchiootic syndrome 1, 602588 Branchiootorenal syndrome 1, with or without cataracts, 113650 Anterior segment anomalies with or without cataract, 602588 ?Otofaciocervical syndrome, 166780
EYA4	100%	100%	?Cardiomyopathy, dilated, 1J, 605362 Deafness, autosomal dominant 10, 601316
EYS	100%	100%	Retinitis pigmentosa 25, 602772
EZH2	100%	100%	Weaver syndrome, 277590
F10	100%	100%	Factor X deficiency, 227600
F11	100%	100%	Factor XI deficiency, autosomal dominant, 612416 Factor XI deficiency, autosomal recessive, 612416
F12	100%	100%	Angioedema, hereditary, 3, 610618 Factor XII deficiency, 234000
F13A1	100%	100%	Factor XIIIA deficiency, 613225
F13B	100%	100%	Factor XIIIB deficiency, 613235
F2	100%	100%	Hypoprothrombinemia, 613679 Dysprothrombinemia, 613679 Thrombophilia due to thrombin defect, 188050
F2RL3	100%	100%	No OMIM disease ID

F5	100%	100%	Thrombophilia due to activated protein C resistance, 188055 Factor V deficiency, 227400
F7	100%	100%	Factor VII deficiency, 227500
F8	100%	100%	Hemophilia A, 306700
F9	100%	99%	Thrombophilia, X-linked, due to factor IX defect, 300807 Hemophilia B, 306900
FA2H	100%	100%	Spastic paraplegia 35, autosomal recessive, 612319
FAAH	100%	100%	No OMIM disease ID
FAAP24	100%	100%	No OMIM disease ID
FADD	100%	100%	Immunodeficiency 90 with encephalopathy, functional hyposplenia, and hepatic dysfunction, 613759
FAH	100%	100%	Tyrosinemia, type I, 276700
FAM111A	100%	100%	Kenny-Caffey syndrome, type 2, 127000 Gracile bone dysplasia, 602361
FAM111B	100%	100%	Poikiloderma, hereditary fibrosing, with tendon contractures, myopathy, and pulmonary fibrosis, 615704
FAM126A	100%	100%	Leukodystrophy, hypomyelinating, 5, 610532
FAM149B1	100%	100%	Joubert syndrome 36, 618763
FAM161A	100%	100%	Retinitis pigmentosa 28, 606068
FAM20A	100%	100%	Amelogenesis imperfecta, type IG (enamel-renal syndrome), 204690
FAM20B	100%	100%	No OMIM disease ID
FAM20C	100%	100%	Raine syndrome, 259775
FAM50A	100%	100%	Intellectual developmental disorder, X-linked, syndromic, Armfield type, 300261
FAM83G	100%	100%	No OMIM disease ID
FAM83H	100%	100%	Amelogenesis imperfecta, type IIIA, 130900
FAN1	100%	100%	Interstitial nephritis, karyomegalic, 614817
FANCA	100%	100%	Fanconi anemia, complementation group A, 227650
FANCB	100%	100%	Fanconi anemia, complementation group B, 300514
FANCC	97%	97%	Fanconi anemia, complementation group C, 227645
FANCD2	98%	98%	Fanconi anemia, complementation group D2, 227646
FANCE	100%	100%	Fanconi anemia, complementation group E, 600901
FANCF	100%	100%	Fanconi anemia, complementation group F, 603467
FANCG	100%	100%	Fanconi anemia, complementation group G, 614082

FANCI	100%	100%	Fanconi anemia, complementation group I, 609053
FANCL	100%	100%	Fanconi anemia, complementation group L, 614083
FANCM	100%	100%	?Premature ovarian failure 15, 618096 Spermatogenic failure 28, 618086
FAR1	100%	100%	Peroxisomal fatty acyl-CoA reductase 1 disorder, 616154 Cataracts, spastic paraparesis, and speech delay, 619338
FARS2	100%	100%	Combined oxidative phosphorylation deficiency 14, 614946 Spastic paraplegia 77, autosomal recessive, 617046
FARSA	100%	100%	?Rajab interstitial lung disease with brain calcifications 2, 619013
FARSB	100%	100%	Rajab interstitial lung disease with brain calcifications 1, 613658
FAS	100%	100%	Autoimmune lymphoproliferative syndrome, type IA, 601859 Squamous cell carcinoma, burn scar-related, somatic,
FASLG	100%	100%	Autoimmune lymphoproliferative syndrome, type IB, 601859
FASTKD2	100%	100%	Combined oxidative phosphorylation deficiency 44, 618855
FAT1	100%	100%	No OMIM disease ID
FAT2	100%	100%	Spinocerebellar ataxia 45, 617769
FAT4	100%	100%	Van Maldergem syndrome 2, 615546 Hennekam lymphangiectasia-lymphedema syndrome 2, 616006
FBLN1	100%	100%	No OMIM disease ID

FBLN5	91%	91%	Cutis laxa, autosomal recessive, type IA, 219100 Macular degeneration, age-related, 3, 608895 Neuropathy, hereditary, with or without age-related macular degeneration, 608895 ?Cutis laxa, autosomal dominant 2, 614434
FBN1	100%	100%	Geleophysic dysplasia 2, 614185 Weill-Marchesani syndrome 2, dominant, 608328 Ectopia lentis, familial, 129600 MASS syndrome, 604308 Marfan lipodystrophy syndrome, 616914 Acromicric dysplasia, 102370 Marfan syndrome, 154700 Stiff skin syndrome, 184900
FBN2	100%	100%	Macular degeneration, early-onset, 616118 Contractural arachnodactyly, congenital, 121050
FBP1	93%	93%	Fructose-1,6-bisphosphatase deficiency, 229700
FBP2	100%	100%	No OMIM disease ID
FBRSL1	99%	98%	No OMIM disease ID
FBXL3	100%	100%	Intellectual developmental disorder with short stature, facial anomalies, and speech defects, 606220
FBXL4	100%	100%	Mitochondrial DNA depletion syndrome 13 (encephalomyopathic type), 615471
FBXO11	100%	100%	Intellectual developmental disorder with dysmorphic facies and behavioral abnormalities, 618089
FBXO31	100%	100%	?Mental retardation, autosomal recessive 45, 615979
FBXO32	100%	100%	No OMIM disease ID
FBXO38	100%	100%	Neuronopathy, distal hereditary motor, type IID, 615575
FBXO7	100%	100%	Parkinson disease 15, autosomal recessive, 260300
FBXW11	100%	100%	Neurodevelopmental, jaw, eye, and digital syndrome, 618914
FBXW4	91%	86%	No OMIM disease ID

FBXW7	100%	100%	No OMIM disease ID
FCGR1A	100%	100%	No OMIM disease ID
FCGR2A	100%	100%	No OMIM disease ID
FCGR2B	100%	100%	No OMIM disease ID
FCGR2C	99%	98%	No OMIM disease ID
FCGR3A	100%	100%	Immunodeficiency 20, 615707
FCGR3B	98%	97%	No OMIM disease ID
FCHO1	100%	100%	Immunodeficiency 76, 619164
FCN3	100%	100%	Immunodeficiency due to ficolin 3 deficiency, 613860
FCSK	100%	100%	Congenital disorder of glycosylation with defective fucosylation 2, 618324
FDFT1	100%	100%	Squalene synthase deficiency, 618156
FDPS	100%	100%	Porokeratosis 9, multiple types, 616631
FDX2	100%	100%	Mitochondrial myopathy, episodic, with optic atrophy and reversible leukoencephalopathy, 251900
FDXR	100%	100%	Auditory neuropathy and optic atrophy, 617717
FECH	100%	100%	Protoporphyria, erythropoietic, 1, 177000
FERMT1	100%	100%	Kindler syndrome, 173650
FERMT3	100%	100%	Leukocyte adhesion deficiency, type III, 612840
FEZF1	100%	100%	Hypogonadotropic hypogonadism 22, with or without anosmia, 616030
FGA	100%	100%	Hypodysfibrinogenemia, congenital, 616004 Dysfibrinogenemia, congenital, 616004 Amyloidosis, familial visceral, 105200 Afibrinogenemia, congenital, 202400
FGB	100%	100%	Hypofibrinogenemia, congenital, 202400 Dysfibrinogenemia, congenital, 616004 Afibrinogenemia, congenital, 202400
FGD1	100%	100%	Mental retardation, X-linked syndromic 16, 305400 Aarskog-Scott syndrome, 305400
FGD4	100%	100%	Charcot-Marie-Tooth disease, type 4H, 609311

FGF10	100%	100%	Aplasia of lacrimal and salivary glands, 180920 LADD syndrome, 149730
FGF12	100%	100%	Developmental and epileptic encephalopathy 47, 617166
FGF13	100%	100%	Developmental and epileptic encephalopathy 90, 301058
FGF14	100%	100%	Spinocerebellar ataxia 27, 609307
FGF16	100%	100%	Metacarpal 4-5 fusion, 309630
FGF17	100%	100%	Hypogonadotropic hypogonadism 20 with or without anosmia, 615270
FGF20	100%	100%	?Renal hypodysplasia/aplasia 2, 615721
FGF23	100%	100%	Tumoral calcinosis, hyperphosphatemic, familial, 2, 617993 Hypophosphatemic rickets, autosomal dominant, 193100
FGF3	100%	100%	Deafness, congenital with inner ear agenesis, microtia, and microdontia, 610706
FGF5	100%	100%	Trichomegaly, 190330
FGF8	100%	100%	Hypogonadotropic hypogonadism 6 with or without anosmia, 612702
FGF9	100%	100%	Multiple synostoses syndrome 3, 612961
FGFR1	100%	100%	Pfeiffer syndrome, 101600 Hypogonadotropic hypogonadism 2 with or without anosmia, 147950 Jackson-Weiss syndrome, 123150 Hartsfield syndrome, 615465 Trigonocephaly 1, 190440 Osteoglophonic dysplasia, 166250 Encephalocraniocutaneous lipomatosis, somatic mosaic, 613001

FGFR2	100%	100%	Bent bone dysplasia syndrome, 614592 LADD syndrome, 149730 Scaphocephaly, maxillary retrusion, and mental retardation, 609579 Antley-Bixler syndrome without genital anomalies or disordered steroidogenesis, 207410 Jackson-Weiss syndrome, 123150 Gastric cancer, somatic, 613659 Craniofacial-skeletal-dermatologic dysplasia, 101600 Apert syndrome, 101200 Pfeiffer syndrome, 101600 Beare-Stevenson cutis gyrata syndrome, 123790 Crouzon syndrome, 123500 Saethre-Chotzen syndrome, 101400 Scaphocephaly and Axenfeld-Rieger anomaly, Craniosynostosis, nonspecific,

FGFR3	100%	100%	Muenke syndrome, 602849 SADDAN, 616482 Hypochondroplasia, 146000 LADD syndrome, 149730 Thanatophoric dysplasia, type II, 187601 Nevus, epidermal, somatic, 162900 CATSHL syndrome, 610474 Thanatophoric dysplasia, type I, 187600 Spermatocytic seminoma, somatic, 273300 Bladder cancer, somatic, 109800 Achondroplasia, 100800 Cervical cancer, somatic, 603956 Colorectal cancer, somatic, 114500 Crouzon syndrome with acanthosis nigricans, 612247
FGG	100%	100%	Dysfibrinogenemia, congenital, 616004 Hypodysfibrinogenemia, 616004 Hypofibrinogenemia, congenital, 202400 Afibrinogenemia, congenital, 202400
FH	100%	100%	Leiomyomatosis and renal cell cancer, 150800 Fumarase deficiency, 606812

FHL1	100%	100%	Myopathy, X-linked, with postural muscle atrophy, 300696 Emery-Dreifuss muscular dystrophy 6, X-linked, 300696 ?Uruguay faciocardiomusculoskeletal syndrome, 300280 Scapuloperoneal myopathy, X-linked dominant, 300695 Reducing body myopathy, X-linked 1b, with late childhood or adult onset, 300718 Reducing body myopathy, X-linked 1a, severe, infantile or early childhood onset, 300717
FHL2	100%	100%	No OMIM disease ID
FHOD3	100%	100%	Cardiomyopathy, familial hypertrophic, 28, 619402
FIBP	100%	100%	Thauvin-Robinet-Faivre syndrome, 617107
FIG4	100%	100%	Yunis-Varon syndrome, 216340 ?Polymicrogyria, bilateral temporooccipital, 612691 Amyotrophic lateral sclerosis 11, 612577 Charcot-Marie-Tooth disease, type 4J, 611228
FIGLA	100%	100%	Premature ovarian failure 6, 612310
FIGN	100%	100%	No OMIM disease ID
FITM2	100%	100%	Siddiqi syndrome, 618635
FKBP10	100%	100%	Osteogenesis imperfecta, type XI, 610968 Bruck syndrome 1, 259450
FKBP14	100%	100%	Ehlers-Danlos syndrome, kyphoscoliotic type, 2, 614557

FKRP	100%	100%	Muscular dystrophy-dystroglycanopathy (congenital with or without mental retardation), type B, 5, 606612 Muscular dystrophy-dystroglycanopathy (limb-girdle), type C, 5, 607155 Muscular dystrophy-dystroglycanopathy (congenital with brain and eye anomalies), type A, 5, 613153
FKTN	100%	100%	Muscular dystrophy-dystroglycanopathy (limb-girdle), type C, 4, 611588 Muscular dystrophy-dystroglycanopathy (congenital with brain and eye anomalies), type A, 4, 253800 Muscular dystrophy-dystroglycanopathy (congenital without mental retardation), type B, 4, 613152 Cardiomyopathy, dilated, 1X, 611615
FLAD1	100%	100%	Lipid storage myopathy due to flavin adenine dinucleotide synthetase deficiency, 255100
FLCN	100%	100%	Birt-Hogg-Dube syndrome, 135150 Colorectal cancer, somatic, 114500 Pneumothorax, primary spontaneous, 173600 Renal carcinoma, chromophobe, somatic, 144700
FLG	100%	100%	Ichthyosis vulgaris, 146700
FLG2	100%	100%	Peeling skin syndrome 6, 618084
FLI1	100%	100%	Bleeding disorder, platelet-type, 21, 617443

FLNA	100%	100%	Otopalatodigital syndrome, type II, 304120 Intestinal pseudoobstruction, neuronal, 300048 Cardiac valvular dysplasia, X-linked, 314400 ?FG syndrome 2, 300321 Melnick-Needles syndrome, 309350 Terminal osseous dysplasia, 300244 Congenital short bowel syndrome, 300048 Otopalatodigital syndrome, type I, 311300 Heterotopia, periventricular, 1, 300049 Frontometaphyseal dysplasia 1, 305620
FLNB	100%	100%	Larsen syndrome, 150250 Atelosteogenesis, type I, 108720 Atelosteogenesis, type III, 108721 Spondylocarpotarsal synostosis syndrome, 272460 Boomerang dysplasia, 112310
FLNC	100%	100%	Cardiomyopathy, familial hypertrophic, 26, 617047 Cardiomyopathy, familial restrictive 5, 617047 Myopathy, distal, 4, 614065 Myopathy, myofibrillar, 5, 609524
FLRT3	100%	100%	Hypogonadotropic hypogonadism 21 with anosmia, 615271

FLT3	100%	100%	Leukemia, acute lymphoblastic, somatic, 613065 Leukemia, acute myeloid, reduced survival in, somatic, 601626 Leukemia, acute myeloid, somatic, 601626
FLT4	100%	100%	Hemangioma, capillary infantile, somatic, 602089 Lymphatic malformation 1, 153100 Congenital heart defects, multiple types, 7, 618780
FLVCR1	100%	100%	Ataxia, posterior column, with retinitis pigmentosa, 609033
FLVCR2	100%	100%	Proliferative vasculopathy and hydranencephaly-hydrocephaly syndrome, 225790
FMN1	100%	100%	No OMIM disease ID
FMN2	100%	100%	Mental retardation, autosomal recessive 47, 616193
FMO3	100%	100%	Trimethylaminuria, 602079
FMR1	100%	100%	Fragile X tremor/ataxia syndrome, 300623 Fragile X syndrome, 300624 Premature ovarian failure 1, 311360
FN1	100%	100%	Spondylometaphyseal dysplasia, corner fracture type, 184255 Glomerulopathy with fibronectin deposits 2, 601894
FNIP1	100%	100%	No OMIM disease ID
FOLR1	100%	100%	Neurodegeneration due to cerebral folate transport deficiency, 613068
FOXC1	100%	100%	Axenfeld-Rieger syndrome, type 3, 602482 Anterior segment dysgenesis 3, multiple subtypes, 601631
FOXC2	100%	100%	Lymphedema-distichiasis syndrome, 153400 Lymphedema-distichiasis syndrome with renal disease and diabetes mellitus, 153400

FOXD4	100%	100%	No OMIM disease ID
FOXE1	100%	100%	Bamforth-Lazarus syndrome, 241850
FOXE3	100%	99%	Anterior segment dysgenesis 2, multiple subtypes, 610256 Cataract 34, multiple types, 612968
FOXF1	100%	100%	Alveolar capillary dysplasia with misalignment of pulmonary veins, 265380
FOXF2	98%	97%	No OMIM disease ID
FOXG1	100%	99%	Rett syndrome, congenital variant, 613454
FOXH1	100%	100%	No OMIM disease ID
FOXI1	100%	100%	Enlarged vestibular aqueduct, 600791
FOXJ1	100%	100%	Ciliary dyskinesia, primary, 43, 618699
FOXL1	100%	100%	No OMIM disease ID
FOXL2	100%	100%	Blepharophimosis, epicanthus inversus, and ptosis, type 2, 110100 Blepharophimosis, epicanthus inversus, and ptosis, type 1, 110100 Premature ovarian failure 3, 608996
FOXN1	100%	100%	T-cell lymphopenia, infantile, with or without nail dystrophy, autosomal dominant, 618806 T-cell immunodeficiency, congenital alopecia, and nail dystrophy, 601705
FOXO1	100%	100%	Rhabdomyosarcoma, alveolar, 268220
FOXP1	100%	100%	Mental retardation with language impairment and with or without autistic features, 613670
FOXP2	100%	100%	Speech-language disorder-1, 602081
FOXP3	100%	100%	Immunodysregulation, polyendocrinopathy, and enteropathy, X-linked, 304790
FOXRED1	100%	100%	Mitochondrial complex I deficiency, nuclear type 19, 618241
FPR1	100%	100%	No OMIM disease ID
FRAS1	100%	100%	Fraser syndrome 1, 219000
FREM1	100%	100%	Manitoba oculotrichoanal syndrome, 248450 Bifid nose with or without anorectal and renal anomalies, 608980 Trigonocephaly 2, 614485

FREM2	100%	100%	Fraser syndrome 2, 617666 Cryptophthalmos, unilateral or bilateral, isolated, 123570
FRMD4A	96%	96%	?Corpus callosum, agenesis of, with facial anomalies and cerebellar ataxia, 616819
FRMD7	100%	99%	Nystagmus, infantile periodic alternating, X-linked, 310700 Nystagmus 1, congenital, X-linked, 310700
FRMPD4	98%	98%	Intellectual developmental disorder, X-linked 104, 300983
FRRS1L	100%	100%	Developmental and epileptic encephalopathy 37, 616981
FSCN2	100%	100%	Retinitis pigmentosa 30, 607921
FSHB	100%	100%	Hypogonadotropic hypogonadism 24 without anosmia, 229070
FSHR	100%	100%	Ovarian response to FSH stimulation, 276400 Ovarian hyperstimulation syndrome, 608115 Ovarian dysgenesis 1, 233300
FSIP2	100%	100%	Spermatogenic failure 34, 618153
FTCD	100%	100%	Glutamate formiminotransferase deficiency, 229100
FTH1	100%	100%	?Hemochromatosis, type 5, 615517
FTL	100%	100%	Hyperferritinemia-cataract syndrome, 600886 L-ferritin deficiency, dominant and recessive, 615604 Neurodegeneration with brain iron accumulation 3, 606159
FTO	94%	94%	Growth retardation, developmental delay, facial dysmorphism, 612938
FTSJ1	100%	100%	Intellectual developmental disorder, X-linked 9, 309549
FUCA1	100%	100%	Fucosidosis, 230000
FURIN	100%	100%	No OMIM disease ID
FUS	100%	100%	Amyotrophic lateral sclerosis 6, with or without frontotemporal dementia, 608030 Essential tremor, hereditary, 4, 614782

FUT2	98%	98%	No OMIM disease ID
FUT6	100%	100%	No OMIM disease ID
FUT8	100%	100%	Congenital disorder of glycosylation with defective fucosylation 1, 618005
FUZ	100%	100%	No OMIM disease ID
FXN	100%	100%	Friedreich ataxia with retained reflexes, 229300 Friedreich ataxia, 229300
FXR1	100%	100%	?Myopathy, congenital, with respiratory insufficiency and bone fractures, 618822 ?Myopathy, congenital proximal, with minicore lesions, 618823
FXYD2	100%	100%	Hypomagnesemia 2, renal, 154020
FYB1	100%	100%	Thrombocytopenia 3, 273900
FYCO1	100%	100%	Cataract 18, autosomal recessive, 610019
FZD2	100%	100%	Omodysplasia 2, 164745
FZD4	100%	100%	Retinopathy of prematurity, 133780 Exudative vitreoretinopathy 1, 133780
FZD6	100%	100%	Nail disorder, nonsyndromic congenital, 1, 161050
G6PC1	100%	100%	Glycogen storage disease Ia, 232200
G6PC3	100%	100%	Dursun syndrome, 612541 Neutropenia, severe congenital 4, autosomal recessive, 612541
G6PD	100%	100%	Hemolytic anemia, G6PD deficient (favism), 300908
GAA	100%	100%	Glycogen storage disease II, 232300
GAB1	100%	100%	?Deafness, autosomal recessive 26, 605428
GABBR2	100%	99%	Developmental and epileptic encephalopathy 59, 617904 Neurodevelopmental disorder with poor language and loss of hand skills, 617903
GABRA1	100%	100%	Developmental and epileptic encephalopathy 19, 615744
GABRA2	100%	100%	Developmental and epileptic encephalopathy 78, 618557
GABRA3	100%	99%	No OMIM disease ID
GABRA5	100%	100%	Developmental and epileptic encephalopathy 79, 618559

GABRB1	100%	100%	Developmental and epileptic encephalopathy 45, 617153
GABRB2	100%	100%	Developmental and epileptic encephalopathy 92, 617829
GABRB3	100%	100%	Developmental and epileptic encephalopathy 43, 617113
GABRD	100%	99%	No OMIM disease ID
GABRG2	93%	93%	Developmental and epileptic encephalopathy 74, 618396 Febrile seizures, familial, 8, 607681 Generalized epilepsy with febrile seizures plus, type 3, 607681
GAD1	100%	100%	Developmental and epileptic encephalopathy 89, 619124
GAL	100%	100%	?Epilepsy, familial temporal lobe, 8, 616461
GALC	100%	100%	Krabbe disease, 245200
GALE	100%	100%	Galactose epimerase deficiency, 230350
GALK1	100%	100%	Galactokinase deficiency with cataracts, 230200
GALM	100%	100%	Galactosemia IV, 618881
GALNS	100%	100%	Mucopolysaccharidosis IVA, 253000
GALNT12	100%	99%	No OMIM disease ID
GALNT2	100%	100%	Congenital disorder of glycosylation, type IIt, 618885
GALNT3	100%	100%	Tumoral calcinosis, hyperphosphatemic, familial, 1, 211900
GALNTL5	100%	100%	No OMIM disease ID
GALT	100%	100%	Galactosemia, 230400
GAMT	100%	100%	Cerebral creatine deficiency syndrome 2, 612736
GAN	100%	100%	Giant axonal neuropathy-1, 256850
GANAB	100%	100%	Polycystic kidney disease 3, 600666
GAPVD1	100%	100%	No OMIM disease ID
GARS1	100%	100%	Spinal muscular atrophy, infantile, James type, 619042 Neuronopathy, distal hereditary motor, type VA, 600794 Charcot-Marie-Tooth disease, type 2D, 601472
GAS2	100%	100%	No OMIM disease ID
GAS2L2	100%	100%	?Ciliary dyskinesia, primary, 41, 618449
GAS8	100%	100%	Ciliary dyskinesia, primary, 33, 616726

GATA1	100%	100%	Leukemia, megakaryoblastic, with or without Down syndrome, somatic, 190685 Thrombocytopenia, X-linked, with or without dyserythropoietic anemia, 300367 Anemia, X-linked, with/without neutropenia and/or platelet abnormalities, 300835 Thrombocytopenia with beta-thalassemia, X-linked, 314050
GATA2	100%	100%	Emberger syndrome, 614038 Immunodeficiency 21, 614172
GATA3	100%	100%	Hypoparathyroidism, sensorineural deafness, and renal dysplasia, 146255
GATA4	100%	100%	Tetralogy of Fallot, 187500 Atrial septal defect 2, 607941 Ventricular septal defect 1, 614429 Atrioventricular septal defect 4, 614430 ?Testicular anomalies with or without congenital heart disease, 615542
GATA5	100%	100%	Congenital heart defects, multiple types, 5, 617912
GATA6	100%	100%	Atrial septal defect 9, 614475 Persistent truncus arteriosus, 217095 Pancreatic agenesis and congenital heart defects, 600001 Atrioventricular septal defect 5, 614474 Tetralogy of Fallot, 187500
GATAD1	100%	100%	?Cardiomyopathy, dilated, 2B, 614672
GATAD2B	100%	100%	GAND syndrome, 615074
GATB	100%	100%	?Combined oxidative phosphorylation deficiency 41, 618838
GATC	100%	100%	Combined oxidative phosphorylation deficiency 42, 618839

GATM	100%	100%	Cerebral creatine deficiency syndrome 3, 612718 Fanconi renotubular syndrome 1, 134600
GBA	100%	100%	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
GBA2	100%	100%	Spastic paraplegia 46, autosomal recessive, 614409
GBE1	100%	100%	Glycogen storage disease IV, 232500 Polyglucosan body disease, adult form, 263570
GBF1	100%	100%	Charcot-Marie-Tooth disease, axonal, type 2GG, 606483
GCDH	100%	100%	Glutaricaciduria, type I, 231670
GCGR	100%	100%	Mahvash disease, 619290
GCH1	100%	100%	Dystonia, DOPA-responsive, with or without hyperphenylalaninemia, 128230 Hyperphenylalaninemia, BH4-deficient, B, 233910
GCK	98%	93%	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%	Hemolytic anemia due to gamma-glutamylcysteine synthetase deficiency, 230450
GCLM	100%	100%	No OMIM disease ID
GCM2	100%	100%	Hypoparathyroidism, familial isolated 2, 618883 Hyperparathyroidism 4, 617343
GCNA	100%	100%	No OMIM disease ID

GCNT2	100%	100%	Adult i phenotype without cataract, 110800 Cataract 13 with adult i phenotype, 116700
GCSH	100%	100%	?Glycine encephalopathy, 605899
GDAP1	100%	100%	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%	Spinocerebellar ataxia, autosomal recessive 27, 618369
GDF1	100%	99%	Congenital heart defects, multiple types, 6, 613854 Right atrial isomerism (Ivemark), 208530
GDF11	100%	99%	?Vertebral hypersegmentation and orofacial anomalies, 619122
GDF2	100%	100%	Telangiectasia, hereditary hemorrhagic, type 5, 615506
GDF3	100%	100%	Klippel-Feil syndrome 3, autosomal dominant, 613702 Microphthalmia with coloboma 6, 613703 Microphthalmia, isolated 7, 613704
GDF5	100%	100%	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 Brachydactyly, type A1, C, 615072

GDF6	100%	100%	Microphthalmia with coloboma 6, digenic, 613703 Microphthalmia, isolated 4, 613094 Leber congenital amaurosis 17, 615360 Multiple synostoses syndrome 4, 617898 Klippel-Feil syndrome 1, autosomal dominant, 118100
GDF9	100%	100%	?Premature ovarian failure 14, 618014
GDI1	100%	100%	Intellectual developmental disorder, X-linked 41, 300849
GDNF	100%	100%	No OMIM disease ID
GDPD1	100%	100%	No OMIM disease ID
GEMIN4	100%	100%	Neurodevelopmental disorder with microcephaly, cataracts, and renal abnormalities, 617913
GEMIN5	100%	100%	Neurodevelopmental disorder with cerebellar atrophy and motor dysfunction, 619333
GFAP	100%	100%	Alexander disease, 203450
GFER	100%	100%	Myopathy, mitochondrial progressive, with congenital cataract and developmental delay, 613076
GFI1	100%	100%	?Neutropenia, nonimmune chronic idiopathic, of adults, 607847 Neutropenia, severe congenital 2, autosomal dominant, 613107
GFI1B	100%	100%	Bleeding disorder, platelet-type, 17, 187900
GFM1	100%	100%	Combined oxidative phosphorylation deficiency 1, 609060
GFM2	100%	100%	Combined oxidative phosphorylation deficiency 39, 618397
GFPT1	100%	100%	Myasthenia, congenital, 12, with tubular aggregates, 610542
GFRA1	100%	100%	No OMIM disease ID
GGCX	100%	100%	Vitamin K-dependent clotting factors, combined deficiency of, 1, 277450 Pseudoxanthoma elasticum-like disorder with multiple coagulation factor deficiency, 610842
GGPS1	100%	100%	Muscular dystrophy, congenital hearing loss, and ovarian insufficiency syndrome, 619518
GGT1	100%	100%	?Glutathioninuria, 231950

GH1	100%	100%	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%	99%	Laron dwarfism, 262500 Increased responsiveness to growth hormone, 604271 Growth hormone insensitivity, partial, 604271
GHRHR	100%	100%	Growth hormone deficiency, isolated, type IV, 618157
GHSR	100%	100%	Growth hormone deficiency, isolated partial, 615925
GIGYF1	100%	100%	No OMIM disease ID
GIMAP5	100%	100%	Portal hypertension, noncirrhotic, 2, 619463
GINS1	100%	100%	Immunodeficiency 55, 617827
GINS2	100%	100%	No OMIM disease ID
GIPC1	100%	100%	Oculopharyngodistal myopathy 2, 618940
GIPC3	100%	100%	Deafness, autosomal recessive 15, 601869
GJA1	100%	100%	Erythrokeratodermia variabilis et progressiva 3, 617525 Craniometaphyseal dysplasia, autosomal recessive, 218400 Oculodentodigital dysplasia, 164200 Hypoplastic left heart syndrome 1, 241550 Palmoplantar keratoderma with congenital alopecia, 104100 Syndactyly, type III, 186100 Oculodentodigital dysplasia, autosomal recessive, 257850 Atrioventricular septal defect 3, 600309
GJA3	100%	100%	Cataract 14, multiple types, 601885

GJA5	100%	100%	Atrial fibrillation, familial, 11, 614049 Atrial standstill, digenic (GJA5/SCN5A), 108770
GJA8	100%	100%	Cataract 1, multiple types, 116200
GJB1	100%	100%	Charcot-Marie-Tooth neuropathy, X-linked dominant, 1, 302800
GJB2	100%	100%	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%	Deafness, digenic, GJB2/GJB3, 220290 Deafness, autosomal dominant 2B, 612644 Erythrokeratodermia variabilis et progressiva 1, 133200 Deafness, autosomal recessive, Deafness, autosomal dominant, with peripheral neuropathy,
GJB4	100%	100%	Erythrokeratodermia variabilis et progressiva 2, 617524
GJB6	100%	100%	Ectodermal dysplasia 2, Clouston type, 129500 Deafness, autosomal dominant 3B, 612643 Deafness, autosomal recessive 1B, 612645 Deafness, digenic GJB2/GJB6, 220290

GJC2	99%	98%	Lymphatic malformation 3, 613480 ?Spastic paraplegia 44, autosomal recessive, 613206 Leukodystrophy, hypomyelinating, 2, 608804
GK	100%	100%	Glycerol kinase deficiency, 307030
GLA	91%	91%	Fabry disease, cardiac variant, 301500 Fabry disease, 301500
GLB1	100%	100%	GM1-gangliosidosis, type I, 230500 GM1-gangliosidosis, type III, 230650 Mucopolysaccharidosis type IVB (Morquio), 253010 GM1-gangliosidosis, type II, 230600
GLDC	100%	100%	Glycine encephalopathy, 605899
GLDN	100%	100%	Lethal congenital contracture syndrome 11, 617194
GLE1	100%	100%	Lethal congenital contracture syndrome 1, 253310 Congenital arthrogryposis with anterior horn cell disease, 611890
GLI1	100%	100%	Polydactyly, preaxial I, 174400 Polydactyly, postaxial, type A8, 618123
GLI2	100%	100%	Culler-Jones syndrome, 615849 Holoprosencephaly 9, 610829
GLI3	100%	100%	Greig cephalopolysyndactyly syndrome, 175700 Polydactyly, postaxial, types A1 and B, 174200 Pallister-Hall syndrome, 146510 Polydactyly, preaxial, type IV, 174700
GLIS2	100%	100%	Nephronophthisis 7, 611498
GLIS3	100%	100%	Diabetes mellitus, neonatal, with congenital hypothyroidism, 610199
GLMN	100%	100%	Glomuvenous malformations, 138000
GLRA1	100%	100%	Hyperekplexia 1, 149400
GLRB	100%	100%	Hyperekplexia 2, 614619

GLRX5	100%	100%	Anemia, sideroblastic, 3, pyridoxine-refractory, 616860 Spasticity, childhood-onset, with hyperglycinemia, 616859
GLS	100%	100%	Global developmental delay, progressive ataxia, and elevated glutamine, 618412 ?Infantile cataract, skin abnormalities, glutamate excess, and impaired intellectual development, 618339 Developmental and epileptic encephalopathy 71, 618328
GLUD1	100%	100%	Hyperinsulinism-hyperammonemia syndrome, 606762
GLUL	100%	100%	Glutamine deficiency, congenital, 610015
GLYCTK	100%	100%	D-glyceric aciduria, 220120
GM2A	100%	100%	GM2-gangliosidosis, AB variant, 272750
GMNN	100%	100%	Meier-Gorlin syndrome 6, 616835
GMPPA	100%	100%	Alacrima, achalasia, and mental retardation syndrome, 615510
GMPPB	100%	100%	Muscular dystrophy-dystroglycanopathy (limb-girdle), type C, 14, 615352 Muscular dystrophy-dystroglycanopathy (congenital with mental retardation), type B, 14, 615351 Muscular dystrophy-dystroglycanopathy (congenital with brain and eye anomalies), type A, 14, 615350
GMPR	100%	100%	No OMIM disease ID
GMPS	100%	100%	No OMIM disease ID
GNA11	100%	100%	Hypocalciuric hypercalcemia, type II, 145981 Hypocalcemia, autosomal dominant 2, 615361

GNA14	100%	100%	No OMIM disease ID
GNAI1	100%	100%	No OMIM disease ID
GNAI2	100%	100%	Ventricular tachycardia, idiopathic, 192605 Pituitary adenoma, ACTH-secreting, somatic,
GNAI3	100%	100%	Auriculocondylar syndrome 1, 602483
GNAL	100%	100%	Dystonia 25, 615073
GNAO1	100%	100%	Developmental and epileptic encephalopathy 17, 615473 Neurodevelopmental disorder with involuntary movements, 617493
GNAQ	100%	100%	Capillary malformations, congenital, 1, somatic, mosaic, 163000 Sturge-Weber syndrome, somatic, mosaic, 185300
GNAS	84%	82%	ACTH-independent macronodular adrenal hyperplasia, 219080 Pituitary adenoma 3, multiple types, somatic, 617686 Pseudohypoparathyroidism Ic, 612462 Pseudohypoparathyroidism Ia, 103580 Osseous heteroplasia, progressive, 166350 Pseudohypoparathyroidism Ib, 603233 McCune-Albright syndrome, somatic, mosaic, 174800 Pseudopseudohypoparathyroidism, 612463
GNAS-AS1	NC	NC	Pseudohypoparathyroidism, type IB, 603233

GNAT1	100%	100%	Night blindness, congenital stationary, autosomal dominant 3, 610444 Night blindness, congenital stationary, type 1G, 616389
GNAT2	100%	100%	Achromatopsia 4, 613856
GNB1	100%	100%	Myelodysplastic syndrome, somatic, 614286 Leukemia, acute lymphoblastic, somatic, 613065 Intellectual developmental disorder, autosomal dominant 42, 616973
GNB2	100%	100%	Neurodevelopmental disorder with hypotonia and dysmorphic facies, 619503 ?Sick sinus syndrome 4, 619464
GNB3	100%	100%	Night blindness, congenital stationary, type 1H, 617024
GNB4	100%	100%	Charcot-Marie-Tooth disease, dominant intermediate F, 615185
GNB5	100%	100%	Language delay and ADHD/cognitive impairment with or without cardiac arrhythmia, 617182 Intellectual developmental disorder with cardiac arrhythmia, 617173
GNE	100%	100%	Sialuria, 269921 Nonaka myopathy, 605820
GNMT	100%	100%	Glycine N-methyltransferase deficiency, 606664
GNPAT	100%	100%	Rhizomelic chondrodysplasia punctata, type 2, 222765
GNPNAT1	100%	100%	?Rhizomelic dysplasia, Ain-Naz type, 616510
GNPTAB	100%	100%	Mucolipidosis III alpha/beta, 252600 Mucolipidosis II alpha/beta, 252500
GNPTG	100%	100%	Mucolipidosis III gamma, 252605
GNRH1	100%	100%	?Hypogonadotropic hypogonadism 12 with or without anosmia, 614841
GNRHR	100%	100%	Hypogonadotropic hypogonadism 7 without anosmia, 146110
GNS	100%	100%	Mucopolysaccharidosis type IIID, 252940
GORAB	100%	100%	Geroderma osteodysplasticum, 231070
GOSR2	100%	100%	Epilepsy, progressive myoclonic 6, 614018

GOT1	100%	100%	Aspartate aminotransferase, serum level of, QTL1, 614419
GOT2	100%	100%	Developmental and epileptic encephalopathy 82, 618721
GP1BA	100%	100%	Bernard-Soulier syndrome, type A1 (recessive), 231200 Bernard-Soulier syndrome, type A2 (dominant), 153670 von Willebrand disease, platelet-type, 177820
GP1BB	100%	100%	Giant platelet disorder, isolated, 231200 Bernard-Soulier syndrome, type B, 231200
GP6	99%	96%	Bleeding disorder, platelet-type, 11, 614201
GP9	100%	100%	Bernard-Soulier syndrome, type C, 231200
GPAA1	100%	100%	Glycosylphosphatidylinositol biosynthesis defect 15, 617810
GPC3	100%	100%	Wilms tumor, somatic, 194070 Simpson-Golabi-Behmel syndrome, type 1, 312870
GPC4	100%	100%	Keipert syndrome, 301026
GPC6	100%	100%	Omodysplasia 1, 258315
GPD1	100%	100%	Hypertriglyceridemia, transient infantile, 614480
GPD1L	100%	100%	Brugada syndrome 2, 611777
GPHN	100%	100%	Molybdenum cofactor deficiency C, 615501
GPI	100%	100%	Hemolytic anemia, nonspherocytic, due to glucose phosphate isomerase deficiency, 613470
GPIHBP1	100%	100%	Hyperlipoproteinemia, type 1D, 615947
GPNMB	95%	95%	Amyloidosis, primary localized cutaneous, 3, 617920
GPR101	100%	100%	Pituitary adenoma 2, GH-secreting, 300943
GPR143	100%	99%	Ocular albinism, type I, Nettleship-Falls type, 300500 Nystagmus 6, congenital, X-linked, 300814
GPR161	100%	100%	No OMIM disease ID
GPR179	100%	100%	Night blindness, congenital stationary (complete), 1E, autosomal recessive, 614565
GPR68	100%	100%	Amelogenesis imperfecta, hypomaturation type, IIA6, 617217
GPR88	100%	99%	?Chorea, childhood-onset, with psychomotor retardation, 616939

GPRASP2	100%	100%	?Deafness, X-linked 7, 301018
GPSM2	100%	100%	Chudley-McCullough syndrome, 604213
GPT2	100%	100%	Neurodevelopmental disorder with microcephaly and spastic paraplegia, 616281
GPX1	100%	100%	No OMIM disease ID
GPX4	100%	100%	Spondylometaphyseal dysplasia, Sedaghatian type, 250220
GRAP	100%	100%	Deafness, autosomal recessive 114, 618456
GREB1L	100%	100%	Deafness, autosomal dominant 80, 619274 Renal hypodysplasia/aplasia 3, 617805
GREM1	100%	100%	No OMIM disease ID
GREM2	100%	100%	Tooth agenesis, selective, 9, 617275
GRHL2	100%	100%	Deafness, autosomal dominant 28, 608641 Ectodermal dysplasia/short stature syndrome, 616029 Corneal dystrophy, posterior polymorphous, 4, 618031
GRHL3	100%	100%	Van der Woude syndrome 2, 606713
GRHPR	100%	99%	Hyperoxaluria, primary, type II, 260000
GRIA2	100%	100%	Neurodevelopmental disorder with language impairment and behavioral abnormalities, 618917
GRIA3	99%	99%	Intellectual developmental disorder, X-linked, syndromic, Wu type, 300699
GRIA4	100%	100%	Neurodevelopmental disorder with or without seizures and gait abnormalities, 617864
GRID2	100%	100%	Spinocerebellar ataxia, autosomal recessive 18, 616204
GRIK2	96%	96%	Neurodevelopmental disorder with impaired language and ataxia and with or without seizures, 619580 Intellectual developmental disorder, autosomal recessive 6, 611092

GRIN1	100%	100%	Neurodevelopmental disorder with or without hyperkinetic movements and seizures, autosomal recessive, 617820 Neurodevelopmental disorder with or without hyperkinetic movements and seizures, autosomal dominant, 614254
GRIN2A	100%	100%	Epilepsy, focal, with speech disorder and with or without impaired intellectual development, 245570
GRIN2B	100%	100%	Developmental and epileptic encephalopathy 27, 616139 Intellectual developmental disorder, autosomal dominant 6, with or without seizures, 613970
GRIN2D	99%	98%	Developmental and epileptic encephalopathy 46, 617162
GRIP1	100%	100%	Fraser syndrome 3, 617667
GRK1	100%	100%	Oguchi disease-2, 613411
GRM1	100%	100%	Spinocerebellar ataxia, autosomal recessive 13, 614831 Spinocerebellar ataxia 44, 617691
GRM6	100%	99%	Night blindness, congenital stationary (complete), 1B, autosomal recessive, 257270
GRM7	100%	100%	Neurodevelopmental disorder with seizures, hypotonia, and brain abnormalities, 618922
GRN	100%	100%	Aphasia, primary progressive, 607485 Frontotemporal lobar degeneration with ubiquitin-positive inclusions, 607485 Ceroid lipofuscinosis, neuronal, 11, 614706
GRXCR1	100%	100%	Deafness, autosomal recessive 25, 613285
GRXCR2	100%	100%	?Deafness, autosomal recessive 101, 615837
GSC	100%	100%	Short stature, auditory canal atresia, mandibular hypoplasia, skeletal abnormalities, 602471
GSDME	100%	100%	Deafness, autosomal dominant 5, 600994
GSE1	100%	100%	No OMIM disease ID
GSN	100%	100%	Amyloidosis, Finnish type, 105120
GSR	100%	100%	Hemolytic anemia due to glutathione reductase deficiency, 618660

GSS	100%	100%	Hemolytic anemia due to glutathione synthetase deficiency, 231900 Glutathione synthetase deficiency, 266130
GSX2	100%	100%	Diencephalic-mesencephalic junction dysplasia syndrome 2, 618646
GTF2E2	100%	100%	Trichothiodystrophy 6, nonphotosensitive, 616943
GTF2H5	72%	72%	Trichothiodystrophy 3, photosensitive, 616395
GTPBP2	100%	100%	Jaberi-Elahi syndrome, 617988
GTPBP3	100%	100%	Combined oxidative phosphorylation deficiency 23, 616198
GUCA1A	100%	100%	Cone-rod dystrophy 14, 602093 Cone dystrophy-3, 602093
GUCA1B	100%	100%	Retinitis pigmentosa 48, 613827
GUCY1A1	100%	100%	Moyamoya 6 with achalasia, 615750
GUCY2C	100%	100%	Diarrhea 6, 614616 Meconium ileus, 614665
GUCY2D	100%	100%	Cone-rod dystrophy 6, 601777 ?Choroidal dystrophy, central areolar 1, 215500 Leber congenital amaurosis 1, 204000 Night blindness, congenital stationary, type 1I, 618555
GUF1	100%	100%	?Developmental and epileptic encephalopathy 40, 617065
GULOP	NC	NC	Scurvy,
GUSB	100%	100%	Mucopolysaccharidosis VII, 253220
GYG1	100%	100%	?Glycogen storage disease XV, 613507 Polyglucosan body myopathy 2, 616199
GYS1	100%	100%	Glycogen storage disease 0, muscle, 611556
GYS2	100%	100%	Glycogen storage disease 0, liver, 240600
GZF1	100%	100%	Joint laxity, short stature, and myopia, 617662
H1-4	100%	100%	Rahman syndrome, 617537
H19	NC	NC	No OMIM disease ID
H4C3	100%	100%	No OMIM disease ID
H6PD	100%	100%	Cortisone reductase deficiency 1, 604931
HAAO	100%	100%	Vertebral, cardiac, renal, and limb defects syndrome 1, 617660

HABP2	100%	100%	No OMIM disease ID
HACE1	100%	100%	Spastic paraplegia and psychomotor retardation with or without seizures, 616756
HADH	100%	100%	Hyperinsulinemic hypoglycemia, familial, 4, 609975 3-hydroxyacyl-CoA dehydrogenase deficiency, 231530
HADHA	100%	100%	HELLP syndrome, maternal, of pregnancy, 609016 Mitochondrial trifunctional protein deficiency, 609015 LCHAD deficiency, 609016 Fatty liver, acute, of pregnancy, 609016
HADHB	100%	100%	Trifunctional protein deficiency, 609015
HAGH	100%	99%	No OMIM disease ID
HAMP	100%	100%	Hemochromatosis, type 2B, 613313
HAND1	100%	100%	No OMIM disease ID
HAND2	100%	100%	No OMIM disease ID
HARS1	100%	100%	Charcot-Marie-Tooth disease, axonal, type 2W, 616625 Usher syndrome type 3B, 614504
HARS2	100%	100%	Perrault syndrome 2, 614926
HAVCR2	100%	100%	T-cell lymphoma, subcutaneous panniculitis-like, 618398
HAX1	100%	100%	Neutropenia, severe congenital 3, autosomal recessive, 610738
HBA1	100%	100%	Hemoglobin H disease, nondeletional, 613978 Thalassemias, alpha-, 604131 Heinz body anemias, alpha-, 140700 Methemoglobinemia, alpha type, 617973 Erythrocytosis 7, 617981

HBA2	100%	100%	Heinz body anemia, 140700 Erythrocytosis 7, 617981 Thalassemia, alpha-, 604131 Hemoglobin H disease, deletional and nondeletional, 613978
HBB	100%	100%	Methemoglobinemia, beta type, 617971 Thalassemia-beta, dominant inclusion-body, 603902 Sickle cell anemia, 603903 Thalassemia, beta, 613985 Delta-beta thalassemia, 141749 Hereditary persistence of fetal hemoglobin, 141749 Heinz body anemia, 140700 Erythrocytosis 6, 617980
HBD	100%	100%	Thalassemia due to Hb Lepore, Thalassemia, delta-,
HBG1	98%	97%	Fetal hemoglobin quantitative trait locus 1, 141749
HBG2	100%	100%	Fetal hemoglobin quantitative trait locus 1, 141749 Cyanosis, transient neonatal, 613977
HCCS	100%	100%	Linear skin defects with multiple congenital anomalies 1, 309801
HCFC1	100%	100%	Methylmalonic acidemia and homocysteinemia, cblX type, 309541
HCN1	98%	98%	Developmental and epileptic encephalopathy 24, 615871 Generalized epilepsy with febrile seizures plus, type 10, 618482
HCN2	93%	89%	Febrile seizures, familial, 2, 602477 Generalized epilepsy with febrile seizures plus, type 11, 602477
HCN3	100%	100%	No OMIM disease ID
HCN4	100%	100%	Sick sinus syndrome 2, 163800 Brugada syndrome 8, 613123
HCRT	100%	100%	?Narcolepsy 1, 161400

HDAC4	100%	100%	No OMIM disease ID
HDAC6	100%	100%	?Chondrodysplasia with platyspondyly, distinctive brachydactyly, hydrocephaly, and microphthalmia, 300863
HDAC8	96%	96%	Cornelia de Lange syndrome 5, 300882
HEATR5B	100%	100%	No OMIM disease ID
HECW2	100%	100%	Neurodevelopmental disorder with hypotonia, seizures, and absent language, 617268
HELLS	100%	100%	Immunodeficiency-centromeric instability-facial anomalies syndrome 4, 616911
HEPACAM	100%	100%	Megalencephalic leukoencephalopathy with subcortical cysts 2A, 613925 Megalencephalic leukoencephalopathy with subcortical cysts 2B, remitting, with or without mental retardation, 613926
HEPH	100%	100%	No OMIM disease ID
HEPHL1	100%	100%	?Abnormal hair, joint laxity, and developmental delay, 261990
HERC1	100%	100%	Macrocephaly, dysmorphic facies, and psychomotor retardation, 617011
HERC2	100%	100%	Mental retardation, autosomal recessive 38, 615516
HES7	100%	100%	Spondylocostal dysostosis 4, autosomal recessive, 613686
HESX1	100%	100%	Pituitary hormone deficiency, combined, 5, 182230 Septooptic dysplasia, 182230 Growth hormone deficiency with pituitary anomalies, 182230
HEXA	100%	100%	GM2-gangliosidosis, several forms, 272800 Tay-Sachs disease, 272800
HEXB	100%	100%	Sandhoff disease, infantile, juvenile, and adult forms, 268800
HEY2	100%	100%	No OMIM disease ID
HFE	100%	100%	Hemochromatosis, 235200
HFM1	100%	100%	Premature ovarian failure 9, 615724
HGD	100%	100%	Alkaptonuria, 203500
HGF	100%	100%	Deafness, autosomal recessive 39, 608265
HGSNAT	92%	91%	Mucopolysaccharidosis type IIIC (Sanfilippo C), 252930 Retinitis pigmentosa 73, 616544

HHAT	100%	100%	Nivelon-Nivelon-Mabille syndrome, 600092
HIBADH	100%	100%	No OMIM disease ID
HIBCH	100%	100%	3-hydroxyisobutryl-CoA hydrolase deficiency, 250620
HID1	100%	100%	No OMIM disease ID
HIKESHI	100%	100%	Leukodystrophy, hypomyelinating, 13, 616881
HINT1	100%	100%	Neuromyotonia and axonal neuropathy, autosomal recessive, 137200
HIVEP2	100%	100%	Mental retardation, autosomal dominant 43, 616977
HJV	100%	100%	Hemochromatosis, type 2A, 602390
HK1	100%	100%	Retinitis pigmentosa 79, 617460 Neuropathy, hereditary motor and sensory, Russe type, 605285 Neurodevelopmental disorder with visual defects and brain anomalies, 618547 Hemolytic anemia due to hexokinase deficiency, 235700
HKDC1	100%	100%	Retinitis pigmentosa 92, 619614
HLCS	100%	100%	Holocarboxylase synthetase deficiency, 253270
HMBS	100%	100%	Porphyria, acute intermittent, nonerythroid variant, 176000 Porphyria, acute intermittent, 176000
HMGA2	95%	84%	Silver-Russell syndrome 5, 618908
HMGB3	100%	100%	?Microphthalmia, syndromic 13, 300915
HMGCL	100%	100%	HMG-CoA lyase deficiency, 246450
HMGCS2	100%	100%	HMG-CoA synthase-2 deficiency, 605911
HMOX1	100%	100%	Heme oxygenase-1 deficiency, 614034
HMX1	100%	100%	Oculoauricular syndrome, 612109
HNF1A	100%	100%	Hepatic adenoma, somatic, 142330 Diabetes mellitus, insulin-dependent, 20, 612520 MODY, type III, 600496 Renal cell carcinoma, 144700
HNF1B	100%	100%	Type 2 diabetes mellitus, 125853 Renal cysts and diabetes syndrome, 137920

HNF4A	100%	100%	Fanconi renotubular syndrome 4, with maturity-onset diabetes of the young, 616026 MODY, type I, 125850
HNMT	100%	100%	Mental retardation, autosomal recessive 51, 616739
HNRNPA1	100%	100%	?Inclusion body myopathy with early-onset Paget disease without frontotemporal dementia 3, 615424 Amyotrophic lateral sclerosis 20, 615426
HNRNPA2B1	100%	100%	?Inclusion body myopathy with early-onset Paget disease with or without frontotemporal dementia 2, 615422
HNRNPD	100%	100%	No OMIM disease ID
HNRNPDL	100%	100%	Muscular dystrophy, limb-girdle, autosomal dominant 3, 609115
HNRNPH1	100%	100%	No OMIM disease ID
HNRNPH2	100%	100%	Intellectual developmental disorder, X-linked, syndromic, Bain type, 300986
HNRNPK	100%	100%	Au-Kline syndrome, 616580
HNRNPU	100%	100%	Developmental and epileptic encephalopathy 54, 617391
HOGA1	100%	100%	Hyperoxaluria, primary, type III, 613616
HOMER2	100%	100%	?Deafness, autosomal dominant 68, 616707
HOXA1	100%	100%	Bosley-Salih-Alorainy syndrome, 601536 Athabaskan brainstem dysgenesis syndrome, 601536
HOXA11	100%	100%	Radioulnar synostosis with amegakaryocytic thrombocytopenia 1, 605432
HOXA13	99%	97%	Hand-foot-uterus syndrome, 140000 ?Guttmacher syndrome, 176305
HOXA2	100%	100%	Microtia with or without hearing impairment (AD), 612290 ?Microtia, hearing impairment, and cleft palate (AR), 612290
HOXB1	100%	100%	Facial paresis, hereditary congenital, 3, 614744
HOXB13	100%	100%	No OMIM disease ID
HOXC13	100%	100%	Ectodermal dysplasia 9, hair/nail type, 614931

HOXD10	100%	100%	Vertical talus, congenital, 192950 Charcot-Marie-Tooth disease, foot deformity of, 192950
HOXD13	100%	100%	Syndactyly, type V, 186300 Synpolydactyly 1, 186000 Brachydactyly, type E, 113300 Brachydactyly, type D, 113200 ?Brachydactyly-syndactyly syndrome, 610713
HPCA	100%	100%	Dystonia 2, torsion, autosomal recessive, 224500
HPD	100%	100%	Hawkinsinuria, 140350 Tyrosinemia, type III, 276710
HPDL	100%	100%	Neurodevelopmental disorder with progressive spasticity and brain white matter abnormalities, 619026 Spastic paraplegia 83, autosomal recessive, 619027
HPGD	100%	100%	?Digital clubbing, isolated congenital, 119900 Hypertrophic osteoarthropathy, primary, autosomal recessive 1, 259100 Cranioosteoarthropathy, 259100
HPRT1	100%	100%	Hyperuricemia, HRPT-related, 300323 Lesch-Nyhan syndrome, 300322
HPS1	100%	100%	Hermansky-Pudlak syndrome 1, 203300
HPS3	100%	100%	Hermansky-Pudlak syndrome 3, 614072
HPS4	100%	100%	Hermansky-Pudlak syndrome 4, 614073
HPS5	100%	100%	Hermansky-Pudlak syndrome 5, 614074
HPS6	100%	100%	Hermansky-Pudlak syndrome 6, 614075
HPSE2	100%	100%	Urofacial syndrome 1, 236730
HR	100%	100%	Atrichia with papular lesions, 209500 Alopecia universalis, 203655

HRAS	100%	100%	Bladder cancer, somatic, 109800 Thyroid carcinoma, follicular, somatic, 188470 Congenital myopathy with excess of muscle spindles, 218040 Nevus sebaceous or woolly hair nevus, somatic, 162900 Schimmelpenning-Feuerstein-Mims syndrome, somatic mosaic, 163200 Spitz nevus or nevus spilus, somatic, 137550 Costello syndrome, 218040
HRG	100%	100%	Thrombophilia due to HRG deficiency, 613116
HS2ST1	100%	100%	Neurofacioskeletal syndrome with or without renal agenesis, 619194
HS3ST6	99%	98%	?Angioedema, hereditary, 8, 619367
HS6ST1	100%	100%	No OMIM disease ID
HS6ST2	100%	100%	?Paganini-Miozzo syndrome, 301025
HSCB	100%	100%	?Anemia, sideroblastic, 5, 619523
HSD11B1	100%	100%	Cortisone reductase deficiency 2, 614662
HSD11B2	100%	100%	Apparent mineralocorticoid excess, 218030
HSD17B10	100%	100%	HSD10 mitochondrial disease, 300438
HSD17B3	100%	100%	Pseudohermaphroditism, male, with gynecomastia, 264300
HSD17B4	96%	96%	D-bifunctional protein deficiency, 261515 Perrault syndrome 1, 233400
HSD3B2	100%	100%	Adrenal hyperplasia, congenital, due to 3-beta-hydroxysteroid dehydrogenase 2 deficiency, 201810
HSD3B7	100%	100%	Bile acid synthesis defect, congenital, 1, 607765
HSF2	100%	100%	No OMIM disease ID
HSF2BP	100%	100%	Premature ovarian failure 19, 619245
HSF4	100%	100%	Cataract 5, multiple types, 116800
HSPA9	100%	100%	Even-plus syndrome, 616854 Anemia, sideroblastic, 4, 182170

HSPB1	100%	100%	Neuronopathy, distal hereditary motor, type IIB, 608634 Charcot-Marie-Tooth disease, axonal, type 2F, 606595
HSPB3	100%	100%	?Neuronopathy, distal hereditary motor, type IIC, 613376
HSPB6	100%	100%	No OMIM disease ID
HSPB8	100%	100%	Neuronopathy, distal hereditary motor, type IIA, 158590 Charcot-Marie-Tooth disease, axonal, type 2L, 608673
HSPD1	100%	100%	Spastic paraplegia 13, autosomal dominant, 605280 Leukodystrophy, hypomyelinating, 4, 612233
HSPG2	100%	100%	Dyssegmental dysplasia, Silverman-Handmaker type, 224410 Schwartz-Jampel syndrome, type 1, 255800
HTR1A	100%	100%	Periodic fever, menstrual cycle dependent, 614674
HTRA1	90%	90%	CARASIL syndrome, 600142 Cerebral arteriopathy, autosomal dominant, with subcortical infarcts and leukoencephalopathy, type 2, 616779
HTRA2	100%	100%	3-methylglutaconic aciduria, type VIII, 617248
HTT	100%	100%	Lopes-Maciel-Rodan syndrome, 617435 Huntington disease, 143100
HUWE1	100%	100%	Intellectual developmental disorder, X-linked, Turner type, 309590
HYAL1	100%	100%	?Mucopolysaccharidosis type IX, 601492
HYAL2	100%	100%	No OMIM disease ID
HYDIN	100%	100%	Ciliary dyskinesia, primary, 5, 608647
HYLS1	100%	100%	Hydrolethalus syndrome, 236680
HYOU1	100%	100%	?Immunodeficiency 59 and hypoglycemia, 233600
IARS1	100%	100%	Growth retardation, impaired intellectual development, hypotonia, and hepatopathy, 617093

IARS2	100%	100%	Cataracts, growth hormone deficiency, sensory neuropathy, sensorineural hearing loss, and skeletal dysplasia, 616007
IBA57	100%	100%	Multiple mitochondrial dysfunctions syndrome 3, 615330 ?Spastic paraplegia 74, autosomal recessive, 616451
ICOS	100%	100%	Immunodeficiency, common variable, 1, 607594
ICOSLG	100%	100%	No OMIM disease ID
ID4	100%	99%	No OMIM disease ID
IDH1	100%	100%	No OMIM disease ID
IDH2	100%	100%	D-2-hydroxyglutaric aciduria 2, 613657
IDH3A	100%	100%	Retinitis pigmentosa 90, 619007
IDH3B	100%	100%	Retinitis pigmentosa 46, 612572
IDI1	100%	100%	No OMIM disease ID
IDS	100%	100%	Mucopolysaccharidosis II, 309900
IDUA	100%	100%	Mucopolysaccharidosis Is, 607016 Mucopolysaccharidosis Ih/s, 607015 Mucopolysaccharidosis Ih, 607014
IER3IP1	100%	100%	Microcephaly, epilepsy, and diabetes syndrome, 614231
IFIH1	100%	100%	Aicardi-Goutieres syndrome 7, 615846 Singleton-Merten syndrome 1, 182250
IFITM5	100%	100%	Osteogenesis imperfecta, type V, 610967
IFNAR1	97%	97%	No OMIM disease ID
IFNAR2	100%	100%	?Immunodeficiency 45, 616669
IFNG	100%	100%	?Immunodeficiency 69, mycobacteriosis, 618963
IFNGR1	100%	100%	Immunodeficiency 27A, mycobacteriosis, AR, 209950 Immunodeficiency 27B, mycobacteriosis, AD, 615978
IFNGR2	100%	100%	Immunodeficiency 28, mycobacteriosis, 614889
IFNLR1	100%	100%	No OMIM disease ID
IFRD1	100%	100%	No OMIM disease ID
IFT122	100%	100%	Cranioectodermal dysplasia 1, 218330

IFT140	100%	100%	Short-rib thoracic dysplasia 9 with or without polydactyly, 266920 Retinitis pigmentosa 80, 617781
IFT172	100%	100%	Retinitis pigmentosa 71, 616394 Bardet-Biedl syndrome 20, 619471 Short-rib thoracic dysplasia 10 with or without polydactyly, 615630
IFT27	100%	100%	Bardet-Biedl syndrome 19, 615996
IFT43	100%	100%	?Cranioectodermal dysplasia 3, 614099 ?Retinitis pigmentosa 81, 617871 Short-rib thoracic dysplasia 18 with polydactyly, 617866
IFT52	100%	100%	Short-rib thoracic dysplasia 16 with or without polydactyly, 617102
IFT57	100%	100%	?Orofaciodigital syndrome XVIII, 617927
IFT74	100%	100%	Spermatogenic failure 58, 619585 Joubert syndrome 40, 619582 ?Bardet-Biedl syndrome 22, 617119
IFT80	100%	100%	Short-rib thoracic dysplasia 2 with or without polydactyly, 611263
IFT81	95%	95%	Short-rib thoracic dysplasia 19 with or without polydactyly, 617895
IFT88	100%	100%	No OMIM disease ID
IGBP1	100%	100%	?Corpus callosum, agenesis of, with impaired intellectual development, ocular coloboma and micrognathia, 300472
IGF1	100%	100%	Growth retardation with deafness and mental retardation due to IGF1 deficiency, 608747
IGF1R	100%	100%	Insulin-like growth factor I, resistance to, 270450
IGF2	100%	100%	Silver-Russell syndrome 3, 616489
IGF2R	100%	99%	Hepatocellular carcinoma, somatic, 114550
IGFALS	100%	100%	Acid-labile subunit, deficiency of, 615961
IGFBP7	100%	100%	Retinal arterial macroaneurysm with supravalvular pulmonic stenosis, 614224
IGHG2	100%	100%	IgG2 deficiency, selective,
IGHM	100%	100%	Agammaglobulinemia 1, 601495
IGHMBP2	100%	100%	Neuronopathy, distal hereditary motor, type VI, 604320 Charcot-Marie-Tooth disease, axonal, type 2S, 616155
IGKC	100%	100%	Kappa light chain deficiency, 614102

IGLL1	100%	100%	Agammaglobulinemia 2, 613500
IGSF1	100%	100%	Hypothyroidism, central, and testicular enlargement, 300888
IGSF10	100%	100%	No OMIM disease ID
IGSF3	100%	100%	?Lacrimal duct defect, 149700
IHH	100%	100%	Acrocapitofemoral dysplasia, 607778 Brachydactyly, type A1, 112500
IKBKB	100%	100%	Immunodeficiency 15B, 615592 Immunodeficiency 15A, 618204
IKBKG	100%	100%	Incontinentia pigmenti, 308300 Ectodermal dysplasia and immunodeficiency 1, 300291 Immunodeficiency 33, 300636
IKZF1	100%	100%	Immunodeficiency, common variable, 13, 616873
IKZF3	100%	100%	?Immunodeficiency 84, 619437
IKZF5	100%	100%	Thrombocytopenia, autosomal dominant, 7, 619130
IL10	100%	100%	No OMIM disease ID
IL10RA	100%	100%	Inflammatory bowel disease 28, early onset, autosomal recessive, 613148
IL10RB	100%	100%	Inflammatory bowel disease 25, early onset, autosomal recessive, 612567
IL11RA	100%	100%	Craniosynostosis and dental anomalies, 614188
IL12B	100%	100%	Immunodeficiency 29, mycobacteriosis, 614890
IL12RB1	94%	94%	Immunodeficiency 30, 614891
IL17F	100%	100%	?Candidiasis, familial, 6, autosomal dominant, 613956
IL17RA	100%	100%	Immunodeficiency 51, 613953
IL17RC	100%	100%	Candidiasis, familial, 9, 616445
IL17RD	100%	100%	Hypogonadotropic hypogonadism 18 with or without anosmia, 615267
IL18BP	100%	100%	No OMIM disease ID
IL1RAPL1	100%	100%	Intellectual developmental disorder, X-linked 21, 300143
IL1RN	100%	100%	Interleukin 1 receptor antagonist deficiency, 612852
IL2	100%	100%	No OMIM disease ID
IL21	100%	100%	?Immunodeficiency, common variable, 11, 615767
IL21R	100%	100%	Immunodeficiency 56, 615207
IL2RA	100%	100%	Immunodeficiency 41 with lymphoproliferation and autoimmunity, 606367
IL2RB	100%	100%	Immunodeficiency 63 with lymphoproliferation and autoimmunity, 618495

IL2RG	100%	100%	Combined immunodeficiency, X-linked, moderate, 312863 Severe combined immunodeficiency, X-linked, 300400
IL31RA	100%	100%	?Amyloidosis, primary localized cutaneous, 2, 613955
IL36RN	100%	100%	Psoriasis 14, pustular, 614204
IL37	100%	100%	?Inflammatory bowel disease (infantile ulcerative colitis) 31, 619398
IL6R	92%	92%	Hyper-IgE recurrent infection syndrome 5, autosomal recessive, 618944
IL6ST	100%	100%	Hyper-IgE recurrent infection syndrome 4, autosomal recessive, 618523
IL7R	100%	100%	Severe combined immunodeficiency, T-cell negative, B-cell/natural killer cell-positive type, 608971
ILDR1	100%	100%	Deafness, autosomal recessive 42, 609646
ILK	100%	100%	No OMIM disease ID
IMPA1	100%	100%	Mental retardation, autosomal recessive 59, 617323
IMPDH1	100%	100%	Retinitis pigmentosa 10, 180105 Leber congenital amaurosis 11, 613837
IMPG1	100%	100%	Macular dystrophy, vitelliform, 4, 616151 Retinitis pigmentosa 91, 153870
IMPG2	100%	100%	Retinitis pigmentosa 56, 613581 Macular dystrophy, vitelliform, 5, 616152
INF2	100%	100%	Glomerulosclerosis, focal segmental, 5, 613237 Charcot-Marie-Tooth disease, dominant intermediate E, 614455
ING1	100%	100%	Squamous cell carcinoma, head and neck, somatic, 275355
INO80	100%	100%	No OMIM disease ID
INPP5E	100%	100%	Joubert syndrome 1, 213300 Mental retardation, truncal obesity, retinal dystrophy, and micropenis, 610156
INPP5K	100%	100%	Muscular dystrophy, congenital, with cataracts and intellectual disability, 617404
INPPL1	100%	100%	Opsismodysplasia, 258480

INS	100%	100%	Diabetes mellitus, insulin-dependent, 2, 125852 Maturity-onset diabetes of the young, type 10, 613370 Hyperproinsulinemia, 616214 Diabetes mellitus, permanent neonatal 4, 618858
INSL3	80%	80%	Cryptorchidism, 219050
INSR	100%	100%	Rabson-Mendenhall syndrome, 262190 Leprechaunism, 246200 Diabetes mellitus, insulin-resistant, with acanthosis nigricans, 610549 Hyperinsulinemic hypoglycemia, familial, 5, 609968
INTS1	100%	100%	Neurodevelopmental disorder with cataracts, poor growth, and dysmorphic facies, 618571
INTS8	100%	100%	?Neurodevelopmental disorder with cerebellar hypoplasia and spasticity, 618572
INTU	100%	100%	?Orofaciodigital syndrome XVII, 617926 ?Short-rib thoracic dysplasia 20 with polydactyly, 617925
INVS	100%	100%	Nephronophthisis 2, infantile, 602088
IPMK	100%	100%	No OMIM disease ID
IPO8	100%	100%	VISS syndrome, 619472
IQCB1	100%	100%	Senior-Loken syndrome 5, 609254
IQCE	100%	100%	Polydactyly, postaxial, type A7, 617642
IQSEC1	99%	98%	Intellectual developmental disorder with short stature and behavioral abnormalities, 618687
IQSEC2	99%	98%	Intellectual developmental disorder, X-linked 1, 309530
IRAK1	100%	100%	No OMIM disease ID
IRAK4	100%	100%	Immunodeficiency 67, 607676
IREB2	100%	100%	Neurodegeneration, early-onset, with choreoathetoid movements and microcytic anemia, 618451

IRF1	100%	100%	Nonsmall cell lung cancer, somatic, 211980 Gastric cancer, somatic, 613659 Myelodysplastic syndrome, preleukemic, Myelogenous leukemia, acute,
IRF2BP2	100%	100%	?Immunodeficiency, common variable, 14, 617765
IRF2BPL	100%	100%	Neurodevelopmental disorder with regression, abnormal movements, loss of speech, and seizures, 618088
IRF3	100%	100%	No OMIM disease ID
IRF4	100%	100%	No OMIM disease ID
IRF6	100%	100%	Popliteal pterygium syndrome 1, 119500 van der Woude syndrome, 119300
IRF7	100%	100%	?Immunodeficiency 39, 616345
IRF8	100%	100%	Immunodeficiency 32A, mycobacteriosis, autosomal dominant, 614893 Immunodeficiency 32B, monocyte and dendritic cell deficiency, autosomal recessive, 226990
IRF9	100%	100%	Immunodeficiency 65, susceptibility to viral infections, 618648
IRGM	100%	100%	No OMIM disease ID
IRS4	100%	100%	Hypothyroidism, congenital, nongoitrous, 9, 301035
IRX1	100%	99%	No OMIM disease ID
IRX5	100%	100%	Hamamy syndrome, 611174
ISCA1	95%	95%	Multiple mitochondrial dysfunctions syndrome 5, 617613
ISCA2	100%	100%	Multiple mitochondrial dysfunctions syndrome 4, 616370
ISCU	100%	100%	Myopathy with lactic acidosis, hereditary, 255125
ISG15	100%	100%	Immunodeficiency 38, 616126
ITCH	95%	93%	Autoimmune disease, multisystem, with facial dysmorphism, 613385
ITGA2	100%	100%	No OMIM disease ID

ITGA2B	100%	100%	Glanzmann thrombasthenia 1, 273800 Bleeding disorder, platelet-type, 16, autosomal dominant, 187800 Thrombocytopenia, neonatal alloimmune, BAK antigen related,
ITGA3	100%	100%	Interstitial lung disease, nephrotic syndrome, and epidermolysis bullosa, congenital, 614748
ITGA6	100%	100%	Epidermolysis bullosa, junctional, with pyloric stenosis, 226730
ITGA7	100%	100%	Muscular dystrophy, congenital, due to ITGA7 deficiency, 613204
ITGA8	100%	100%	Renal hypodysplasia/aplasia 1, 191830
ITGB2	97%	97%	Leukocyte adhesion deficiency, 116920
ITGB3	100%	100%	Bleeding disorder, platelet-type, 24, autosomal dominant, 619271 Glanzmann thrombasthenia 2, 619267 Thrombocytopenia, neonatal alloimmune, Purpura, posttransfusion,
ITGB4	100%	100%	Epidermolysis bullosa of hands and feet, 131800 Epidermolysis bullosa, junctional, with pyloric atresia, 226730 Epidermolysis bullosa, junctional, non-Herlitz type, 226650
ITGB6	100%	100%	Amelogenesis imperfecta, type IH, 616221
ITK	100%	100%	Lymphoproliferative syndrome 1, 613011
ITM2B	100%	100%	?Retinal dystrophy with inner retinal dysfunction and ganglion cell abnormalities, 616079 Dementia, familial British, 176500 Dementia, familial Danish, 117300
ITPA	100%	100%	Developmental and epileptic encephalopathy 35, 616647
ITPKB	100%	100%	No OMIM disease ID

ITPR1	100%	100%	Gillespie syndrome, 206700 Spinocerebellar ataxia 29, congenital nonprogressive, 117360 Spinocerebellar ataxia 15, 606658
ITPR2	100%	100%	?Anhidrosis, isolated, with normal sweat glands, 106190
ITPR3	100%	100%	No OMIM disease ID
ITSN1	100%	100%	No OMIM disease ID
ITSN2	100%	100%	No OMIM disease ID
IVD	100%	100%	Isovaleric acidemia, 243500
IVNS1ABP	100%	100%	Immunodeficiency 70, 618969
IYD	100%	100%	Thyroid dyshormonogenesis 4, 274800
JAG1	100%	100%	?Deafness, congenital heart defects, and posterior embryotoxon, 617992 Charcot-Marie-Tooth disease, axonal, type 2HH, 619574 Alagille syndrome 1, 118450 Tetralogy of Fallot, 187500
JAGN1	100%	100%	Neutropenia, severe congenital, 6, autosomal recessive, 616022
JAK1	100%	100%	Autoinflammation, immune dysregulation, and eosinophilia, 618999
JAK2	100%	100%	Myelofibrosis, somatic, 254450 Erythrocytosis, somatic, 133100 Leukemia, acute myeloid, somatic, 601626 Thrombocythemia 3, 614521 Polycythemia vera, somatic, 263300
JAK3	100%	100%	SCID, autosomal recessive, T-negative/B-positive type, 600802
JAM2	92%	92%	Basal ganglia calcification, idiopathic, 8, autosomal recessive, 618824
JAM3	100%	100%	Hemorrhagic destruction of the brain, subependymal calcification, and cataracts, 613730
JARID2	100%	100%	No OMIM disease ID
JMJD1C	100%	100%	No OMIM disease ID
JPH1	100%	100%	?Charcot-Marie-Tooth disease, axonal, autosomal dominant, type 2K, 607831
JPH2	100%	100%	Cardiomyopathy, dilated, 2E, 619492 Cardiomyopathy, hypertrophic, 17, 613873
JPH3	100%	100%	Huntington disease-like 2, 606438

JUP	100%	100%	Naxos disease, 601214 ?Arrhythmogenic right ventricular dysplasia 12, 611528
KALRN	100%	100%	No OMIM disease ID
KANK1	100%	100%	Cerebral palsy, spastic quadriplegic, 2, 612900
KANK2	100%	100%	Nephrotic syndrome, type 16, 617783 Palmoplantar keratoderma and woolly hair, 616099
KANSL1	100%	100%	Koolen-De Vries syndrome, 610443
KARS1	100%	100%	Deafness, autosomal recessive 89, 613916 Leukoencephalopathy, progressive, infantile-onset, with or without deafness, 619147 ?Charcot-Marie-Tooth disease, recessive intermediate, B, 613641 Deafness, congenital, and adult-onset progressive leukoencephalopathy, 619196
KASH5	100%	100%	No OMIM disease ID
KAT5	100%	100%	Neurodevelopmental disorder wtih dysmorphic facies, sleep disturbance, and brain abnormalities, 619103
KAT6A	100%	100%	Arboleda-Tham syndrome, 616268
KAT6B	100%	100%	SBBYSS syndrome, 603736 Genitopatellar syndrome, 606170
KAT8	100%	100%	Li-Ghorgani-Weisz-Hubshman syndrome, 618974
KATNB1	100%	100%	Lissencephaly 6, with microcephaly, 616212
KATNIP	100%	100%	Joubert syndrome 26, 616784
KBTBD13	100%	100%	Nemaline myopathy 6, autosomal dominant, 609273
KCNA1	100%	100%	Episodic ataxia/myokymia syndrome, 160120
KCNA2	100%	100%	Developmental and epileptic encephalopathy 32, 616366
KCNA4	100%	100%	Microcephaly, cataracts, impaired intellectual development, and dystonia with abnormal striatum, 618284
KCNA5	100%	100%	Atrial fibrillation, familial, 7, 612240
KCNB1	100%	100%	Developmental and epileptic encephalopathy 26, 616056
KCNC1	100%	100%	Epilepsy, progressive myoclonic 7, 616187
KCNC3	99%	98%	Spinocerebellar ataxia 13, 605259

KCND2	100%	100%	No OMIM disease ID
KCND3	100%	100%	Spinocerebellar ataxia 19, 607346 Brugada syndrome 9, 616399
KCNE1	100%	100%	Jervell and Lange-Nielsen syndrome 2, 612347 Long QT syndrome 5, 613695
KCNE2	100%	100%	Long QT syndrome 6, 613693 Atrial fibrillation, familial, 4, 611493
KCNE3	100%	100%	?Brugada syndrome 6, 613119
KCNE4	100%	100%	No OMIM disease ID
KCNE5	100%	100%	No OMIM disease ID
KCNH1	98%	98%	Zimmermann-Laband syndrome 1, 135500 Temple-Baraitser syndrome, 611816
KCNH2	100%	100%	Short QT syndrome 1, 609620 Long QT syndrome 2, 613688
KCNJ1	100%	100%	Bartter syndrome, type 2, 241200
KCNJ10	100%	100%	Enlarged vestibular aqueduct, digenic, 600791 SESAME syndrome, 612780
KCNJ11	100%	100%	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%	Snowflake vitreoretinal degeneration, 193230 Leber congenital amaurosis 16, 614186
KCNJ16	100%	100%	Hypokalemic tubulopathy and deafness, 619406

KCNJ2	100%	100%	Atrial fibrillation, familial, 9, 613980 Andersen syndrome, 170390 Short QT syndrome 3, 609622
KCNJ5	100%	100%	Long QT syndrome 13, 613485 Hyperaldosteronism, familial, type III, 613677
KCNJ6	100%	100%	Keppen-Lubinsky syndrome, 614098
KCNJ8	100%	100%	No OMIM disease ID
KCNK3	100%	100%	Pulmonary hypertension, primary, 4, 615344
KCNK4	100%	100%	Facial dysmorphism, hypertrichosis, epilepsy, intellectual/developmental delay, and gingival overgrowth syndrome, 618381
KCNK9	97%	97%	Birk-Barel syndrome, 612292
KCNMA1	100%	100%	Paroxysmal nonkinesigenic dyskinesia, 3, with or without generalized epilepsy, 609446 Cerebellar atrophy, developmental delay, and seizures, 617643 Liang-Wang syndrome, 618729
KCNN2	100%	100%	No OMIM disease ID
KCNN3	100%	100%	Zimmermann-Laband syndrome 3, 618658
KCNN4	100%	100%	Dehydrated hereditary stomatocytosis 2, 616689
KCNQ1	100%	100%	Short QT syndrome 2, 609621 Atrial fibrillation, familial, 3, 607554 Long QT syndrome 1, 192500 Jervell and Lange-Nielsen syndrome, 220400
KCNQ1OT1	NC	NC	Beckwith-Wiedemann syndrome, 130650
KCNQ2	100%	100%	Developmental and epileptic encephalopathy 7, 613720 Seizures, benign neonatal, 1, 121200 Myokymia, 121200
KCNQ3	100%	100%	Seizures, benign neonatal, 2, 121201
KCNQ4	99%	99%	Deafness, autosomal dominant 2A, 600101
KCNQ5	100%	100%	Mental retardation, autosomal dominant 46, 617601

KCNT1	99%	99%	Developmental and epileptic encephalopathy 14, 614959 Epilepsy nocturnal frontal lobe, 5, 615005
KCNT2	100%	100%	Developmental and epileptic encephalopathy 57, 617771
KCNV2	100%	100%	Retinal cone dystrophy 3B, 610356
KCTD1	100%	100%	Scalp-ear-nipple syndrome, 181270
KCTD17	100%	100%	Dystonia 26, myoclonic, 616398
KCTD3	100%	100%	No OMIM disease ID
KCTD7	100%	100%	Epilepsy, progressive myoclonic 3, with or without intracellular inclusions, 611726
KDELR2	100%	100%	Osteogenesis imperfecta 21, 619131
KDF1	100%	100%	?Ectodermal dysplasia 12, hypohidrotic/hair/tooth/nail type, 617337
KDM1A	100%	100%	Cleft palate, psychomotor retardation, and distinctive facial features, 616728
KDM3B	100%	100%	Diets-Jongmans syndrome, 618846
KDM4B	100%	100%	Intellectual developmental disorder, autosomal dominant 65, 619320
KDM5B	95%	94%	Mental retardation, autosomal recessive 65, 618109
KDM5C	100%	100%	Intellectual developmental disorder, X-linked syndromic, Claes-Jensen type, 300534
KDM6A	100%	100%	Kabuki syndrome 2, 300867
KDM6B	100%	100%	Neurodevelopmental disorder with coarse facies and mild distal skeletal abnormalities, 618505
KDR	100%	100%	Hemangioma, capillary infantile, somatic, 602089
KDSR	100%	100%	Erythrokeratodermia variabilis et progressiva 4, 617526
KERA	100%	100%	Cornea plana 2, autosomal recessive, 217300
KHDC3L	100%	100%	Hydatidiform mole, recurrent, 2, 614293
KIAA0586	95%	95%	Short-rib thoracic dysplasia 14 with polydactyly, 616546 Joubert syndrome 23, 616490
KIAA0753	100%	100%	?Orofaciodigital syndrome XV, 617127 ?Joubert syndrome 38, 619476 Short-rib thoracic dysplasia 21 without polydactyly, 619479
KIAA0825	100%	100%	Polydactyly, postaxial, type A10, 618498
KIAA1109	100%	100%	Alkuraya-Kucinskas syndrome, 617822
KIAA1549	99%	99%	Retinitis pigmentosa 86, 618613

KIDINS220	100%	100%	Spastic paraplegia, intellectual disability, nystagmus, and obesity, 617296 Ventriculomegaly and arthrogryposis, 619501
KIF11	100%	100%	Microcephaly with or without chorioretinopathy, lymphedema, or mental retardation, 152950
KIF14	100%	100%	Microcephaly 20, primary, autosomal recessive, 617914 ?Meckel syndrome 12, 616258
KIF1A	98%	98%	NESCAV syndrome, 614255 Neuropathy, hereditary sensory, type IIC, 614213 Spastic paraplegia 30, autosomal dominant, 610357 Spastic paraplegia 30, autosomal recessive, 610357
KIF1B	100%	100%	Pheochromocytoma, 171300 Charcot-Marie-Tooth disease, type 2A1, 118210
KIF1C	100%	100%	Spastic ataxia 2, autosomal recessive, 611302
KIF20A	100%	100%	?Cardiomyopathy, familial restrictive, 6, 619433
KIF21A	100%	100%	Fibrosis of extraocular muscles, congenital, 3B, 135700 Fibrosis of extraocular muscles, congenital, 1, 135700
KIF21B	100%	100%	No OMIM disease ID
KIF22	100%	100%	Spondyloepimetaphyseal dysplasia with joint laxity, type 2, 603546
KIF23	100%	100%	No OMIM disease ID
KIF2A	100%	100%	Cortical dysplasia, complex, with other brain malformations 3, 615411
KIF3B	100%	100%	Retinitis pigmentosa 89, 618955
KIF4A	100%	100%	?Intellectual developmental disorder, X-linked 100, 300923
KIF5A	100%	100%	Myoclonus, intractable, neonatal, 617235 Spastic paraplegia 10, autosomal dominant, 604187

KIF5C	99%	99%	Cortical dysplasia, complex, with other brain malformations 2, 615282
KIF7	100%	99%	Joubert syndrome 12, 200990 Acrocallosal syndrome, 200990 ?Hydrolethalus syndrome 2, 614120 ?Al-Gazali-Bakalinova syndrome, 607131
KIFBP	96%	96%	Goldberg-Shprintzen megacolon syndrome, 609460
KIRREL1	100%	100%	Nephrotic syndrome, type 23, 619201
KIRREL3	100%	100%	No OMIM disease ID
KISS1	100%	100%	?Hypogonadotropic hypogonadism 13 with or without anosmia, 614842
KISS1R	100%	100%	Hypogonadotropic hypogonadism 8 with or without anosmia, 614837 ?Precocious puberty, central, 1, 176400
KIT	100%	100%	Gastrointestinal stromal tumor, familial, 606764 Mastocytosis, cutaneous, 154800 Piebaldism, 172800 Germ cell tumors, somatic, 273300 Mastocytosis, systemic, somatic, 154800 Leukemia, acute myeloid, somatic, 601626
KITLG	100%	100%	Hyperpigmentation with or without hypopigmentation, 145250 Deafness, autosomal dominant 69, unilateral or asymmetric, 616697
KIZ	100%	100%	Retinitis pigmentosa 69, 615780
KL	99%	99%	?Tumoral calcinosis, hyperphosphatemic, familial, 3, 617994
KLB	100%	100%	No OMIM disease ID
KLC2	100%	100%	Spastic paraplegia, optic atrophy, and neuropathy, 609541
KLF1	100%	100%	Blood group--Lutheran inhibitor, 111150 Dyserythropoietic anemia, congenital, type IV, 613673

KLF10	100%	100%	No OMIM disease ID
KLF11	100%	100%	Maturity-onset diabetes of the young, type VII, 610508
KLF6	100%	100%	Gastric cancer, somatic, 613659 Prostate cancer, somatic, 176807
KLF7	100%	100%	No OMIM disease ID
KLHL10	100%	100%	Spermatogenic failure 11, 615081
KLHL15	100%	100%	Intellectual developmental disorder, X-linked 103, 300982
KLHL24	100%	100%	Epidermolysis bullosa simplex 6, generalized, with scarring and hair loss, 617294
KLHL3	100%	100%	Pseudohypoaldosteronism, type IID, 614495
KLHL40	100%	100%	Nemaline myopathy 8, autosomal recessive, 615348
KLHL41	100%	100%	Nemaline myopathy 9, 615731
KLHL7	100%	100%	Retinitis pigmentosa 42, 612943 PERCHING syndrome, 617055
KLHL9	100%	100%	No OMIM disease ID
KLK4	100%	100%	Amelogenesis imperfecta, type IIA1, 204700
KLKB1	100%	100%	Fletcher factor (prekallikrein) deficiency, 612423
KLLN	100%	100%	Cowden syndrome 4, 615107
KMT2A	100%	100%	Wiedemann-Steiner syndrome, 605130
KMT2B	99%	99%	Dystonia 28, childhood-onset, 617284
KMT2C	100%	100%	Kleefstra syndrome 2, 617768
KMT2D	100%	100%	Kabuki syndrome 1, 147920
KMT2E	100%	100%	O'Donnell-Luria-Rodan syndrome, 618512
KMT5B	100%	100%	Mental retardation, autosomal dominant 51, 617788
KNG1	100%	100%	Angioedema, hereditary, 6, 619363
KNL1	98%	98%	Microcephaly 4, primary, autosomal recessive, 604321
KNSTRN	100%	100%	?Roifman-Chitayat syndrome, digenic, 613328
KPTN	100%	100%	Mental retardation, autosomal recessive 41, 615637

KRAS	100%	100%	Gastric cancer, somatic, 613659 Oculoectodermal syndrome, somatic, 600268 Breast cancer, somatic, 114480 Noonan syndrome 3, 609942 RAS-associated autoimmune leukoproliferative disorder, 614470 Arteriovenous malformation of the brain, somatic, 108010 Lung cancer, somatic, 211980 Pancreatic carcinoma, somatic, 260350 Leukemia, acute myeloid, somatic, 601626 Schimmelpenning-Feuerstein-Mims syndrome, somatic mosaic, 163200 Cardiofaciocutaneous syndrome 2, 615278 Bladder cancer, somatic, 109800
KREMEN1	100%	100%	Ectodermal dysplasia 13, hair/tooth type, 617392
KRIT1	100%	100%	Hyperkeratotic cutaneous capillary-venous malformations associated with cerebral capillary malformations, 116860 Cerebral cavernous malformations-1, 116860 Cavernous malformations of CNS and retina, 116860

KRT1	100%	100%	Ichthyosis, cyclic, with epidermolytic hyperkeratosis, 607602 Epidermolytic hyperkeratosis, 113800 Palmoplantar keratoderma, nonepidermolytic, 600962 Keratosis palmoplantaris striata III, 607654 Palmoplantar keratoderma, epidermolytic, 144200 Ichthyosis histrix, Curth-Macklin type, 146590
KRT10	100%	100%	Epidermolytic hyperkeratosis, 113800 Ichthyosis with confetti, 609165 Ichthyosis, cyclic, with epidermolytic hyperkeratosis, 607602
KRT12	100%	100%	Meesmann corneal dystrophy 1, 122100
KRT13	100%	100%	White sponge nevus 2, 615785
KRT14	100%	100%	Epidermolysis bullosa simplex 1D, generalized, intermediate or severe, autosomal recessive, 601001 Epidermolysis bullosa simplex 1C, localized, 131800 Dermatopathia pigmentosa reticularis, 125595 Epidermolysis bullosa simplex 1A, generalized severe, 131760 Naegeli-Franceschetti-Jadassohn syndrome, 161000 Epidermolysis bullosa simplex 1B, generalized intermediate, 131900
KRT16	100%	100%	Palmoplantar keratoderma, nonepidermolytic, focal, 613000 Pachyonychia congenita 1, 167200
KRT17	100%	100%	Steatocystoma multiplex, 184500 Pachyonychia congenita 2, 167210

KRT18	100%	100%	Cirrhosis, cryptogenic, 215600
KRT2	100%	100%	Ichthyosis bullosa of Siemens, 146800
KRT25	100%	100%	Woolly hair, autosomal recessive 3, 616760
KRT3	100%	100%	Meesmann corneal dystrophy 2, 618767
KRT4	100%	100%	White sponge nevus 1, 193900
KRT5	100%	100%	Epidermolysis bullosa simplex 2A, generalized severe, 619555 Dowling-Degos disease 1, 179850 Epidermolysis bullosa simplex 2F, with mottled pigmentation, 131960 Epidermolysis bullosa simplex 2D, generalized, intermediate or severe, autosomal recessive, 619599 Epidermolysis bullosa simplex 2B, generalized intermediate, 619588 Epidermolysis bullosa simplex 2C, localized, 619594 Epidermolysis bullosa simplex 2E, with migratory circinate erythema, 609352
KRT6A	100%	100%	Pachyonychia congenita 3, 615726
KRT6B	100%	100%	Pachyonychia congenita 4, 615728
KRT6C	100%	99%	Palmoplantar keratoderma, nonepidermolytic, focal or diffuse, 615735
KRT71	100%	100%	?Hypotrichosis 13, 615896
KRT74	100%	100%	Woolly hair, autosomal dominant, 194300 ?Hypotrichosis 3, 613981 ?Ectodermal dysplasia 7, hair/nail type, 614929
KRT75	100%	100%	No OMIM disease ID
KRT8	100%	100%	Cirrhosis, cryptogenic, 215600
KRT81	100%	100%	Monilethrix, 158000
KRT83	100%	100%	Monilethrix, 158000 Erythrokeratodermia variabilis et progressiva 5, 617756

KRT85	100%	100%	Ectodermal dysplasia 4, hair/nail type, 602032
KRT86	100%	100%	Monilethrix, 158000
KRT9	100%	100%	Palmoplantar keratoderma, epidermolytic, 144200
KY	100%	100%	Myopathy, myofibrillar, 7, 617114
KYNU	100%	100%	?Hydroxykynureninuria, 236800 Vertebral, cardiac, renal, and limb defects syndrome 2, 617661
L1CAM	100%	100%	MASA syndrome, 303350 Hydrocephalus with congenital idiopathic intestinal pseudoobstruction, 307000 Corpus callosum, partial agenesis of, 304100 CRASH syndrome, 303350 Hydrocephalus with Hirschsprung disease, 307000 Hydrocephalus due to aqueductal stenosis, 307000
L2HGDH	100%	100%	L-2-hydroxyglutaric aciduria, 236792
LACC1	100%	100%	Juvenile arthritis, 618795
LACTB	100%	100%	No OMIM disease ID
LAGE3	100%	100%	Galloway-Mowat syndrome 2, X-linked, 301006
LAMA1	100%	100%	Poretti-Boltshauser syndrome, 615960
LAMA2	100%	100%	Muscular dystrophy, limb-girdle, autosomal recessive 23, 618138 Muscular dystrophy, congenital, merosin deficient or partially deficient, 607855
LAMA3	100%	100%	Laryngoonychocutaneous syndrome, 245660 Epidermolysis bullosa, junctional, Herlitz type, 226700 Epidermolysis bullosa, generalized atrophic benign, 226650
LAMA4	100%	100%	Cardiomyopathy, dilated, 1JJ, 615235

LAMA5	100%	100%	No OMIM disease ID
LAMB1	100%	100%	Lissencephaly 5, 615191
LAMB2	100%	100%	Nephrotic syndrome, type 5, with or without ocular abnormalities, 614199 Pierson syndrome, 609049
LAMB3	100%	100%	Epidermolysis bullosa, junctional, non-Herlitz type, 226650 Epidermolysis bullosa, junctional, Herlitz type, 226700 Amelogenesis imperfecta, type IA, 104530
LAMC2	100%	100%	Epidermolysis bullosa, junctional, non-Herlitz type, 226650 Epidermolysis bullosa, junctional, Herlitz type, 226700
LAMC3	100%	100%	Cortical malformations, occipital, 614115
LAMP2	100%	100%	Danon disease, 300257
LAMTOR2	100%	100%	Immunodeficiency due to defect in MAPBP-interacting protein, 610798
LAPTM5	100%	100%	No OMIM disease ID
LARGE1	100%	100%	Muscular dystrophy-dystroglycanopathy (congenital with mental retardation), type B, 6, 608840 Muscular dystrophy-dystroglycanopathy (congenital with brain and eye anomalies), type A, 6, 613154
LARP7	100%	100%	Alazami syndrome, 615071
LARS1	100%	100%	?Infantile liver failure syndrome 1, 615438
LARS2	100%	100%	Perrault syndrome 4, 615300 Hydrops, lactic acidosis, and sideroblastic anemia, 617021
LAS1L	100%	100%	Wilson-Turner syndrome, 309585
LAT	100%	100%	Immunodeficiency 52, 617514

LBR	100%	100%	Pelger-Huet anomaly, 169400 Pelger-Huet anomaly with mild skeletal anomalies, 618019 ?Reynolds syndrome, 613471 Greenberg skeletal dysplasia, 215140
LBX1	100%	100%	?Central hypoventilation syndrome, congenital, 3, 619483
LCA5	100%	100%	Leber congenital amaurosis 5, 604537
LCAT	100%	100%	Fish-eye disease, 136120 Norum disease, 245900
LCK	100%	100%	?Immunodeficiency 22, 615758
LCP2	100%	100%	?Immunodeficiency 81, 619374
LCT	100%	100%	Lactase deficiency, congenital, 223000
LDB3	100%	100%	Left ventricular noncompaction 3, 601493 Cardiomyopathy, hypertrophic, 24, 601493 Myopathy, myofibrillar, 4, 609452 Cardiomyopathy, dilated, 1C, with or without LVNC, 601493
LDHA	100%	100%	Glycogen storage disease XI, 612933
LDHB	100%	100%	No OMIM disease ID
LDHD	100%	100%	D-lactic aciduria with susceptibility to gout, 245450
LDLR	100%	100%	LDL cholesterol level QTL2, 143890 Hypercholesterolemia, familial, 1, 143890
LDLRAP1	100%	100%	Hypercholesterolemia, familial, 4, 603813
LEF1	100%	100%	Sebaceous tumors, somatic,
LEFTY2	100%	100%	No OMIM disease ID
LEMD2	100%	100%	Marbach-Rustad progeroid syndrome, 619322 Cataract 46, juvenile-onset, 212500
LEMD3	100%	100%	Buschke-Ollendorff syndrome, 166700 Osteopoikilosis with or without melorheostosis, 166700

LEP	100%	100%	Obesity, morbid, due to leptin deficiency, 614962
LEPR	94%	94%	Obesity, morbid, due to leptin receptor deficiency, 614963
LFNG	98%	93%	Spondylocostal dysostosis 3, autosomal recessive, 609813
LGI1	100%	100%	Epilepsy, familial temporal lobe, 1, 600512
LGI4	100%	100%	Arthrogryposis multiplex congenita 1, neurogenic, with myelin defect, 617468
LHB	100%	100%	Hypogonadotropic hypogonadism 23 with or without anosmia, 228300
LHCGR	100%	100%	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%	Deafness, autosomal recessive 67, 610265
LHX1	100%	100%	No OMIM disease ID
LHX3	100%	100%	Pituitary hormone deficiency, combined, 3, 221750
LHX4	100%	100%	Pituitary hormone deficiency, combined, 4, 262700
LIAS	100%	100%	Hyperglycinemia, lactic acidosis, and seizures, 614462
LIFR	100%	100%	Stuve-Wiedemann syndrome/Schwartz-Jampel type 2 syndrome, 601559
LIG1	100%	100%	No OMIM disease ID
LIG3	100%	100%	No OMIM disease ID
LIG4	100%	100%	LIG4 syndrome, 606593
LIM2	100%	100%	Cataract 19, multiple types, 615277
LIMS2	100%	100%	?Muscular dystrophy, autosomal recessive, with cardiomyopathy and triangular tongue, 616827
LINGO1	100%	100%	Mental retardation, autosomal recessive 64, 618103
LINS1	100%	100%	Mental retardation, autosomal recessive 27, 614340
LIPA	95%	95%	Wolman disease, 278000 Cholesteryl ester storage disease, 278000
LIPC	100%	100%	Hepatic lipase deficiency, 614025
LIPE	100%	100%	Lipodystrophy, familial partial, type 6, 615980

LIPH	100%	100%	Hypotrichosis 7, 604379 Woolly hair, autosomal recessive 2 with or without hypotrichosis, 604379
LIPN	100%	100%	Ichthyosis, congenital, autosomal recessive 8, 613943
LIPT1	100%	100%	Lipoyltransferase 1 deficiency, 616299
LIPT2	100%	100%	Encephalopathy, neonatal severe, with lactic acidosis and brain abnormalities, 617668
LITAF	100%	100%	Charcot-Marie-Tooth disease, type 1C, 601098
LMAN1	100%	100%	Combined factor V and VIII deficiency, 227300
LMAN2L	100%	100%	?Mental retardation, autosomal recessive, 52, 616887
LMBR1	98%	98%	Triphalangeal thumb, type I, 174500 Syndactyly, type IV, 186200 Laurin-Sandrow syndrome, 135750 Hypoplastic or aplastic tibia with polydactyly, 188740 Polydactyly, preaxial type II, 174500 Acheiropody, 200500 Triphalangeal thumb-polysyndactyly syndrome, 174500
LMBRD1	96%	96%	Methylmalonic aciduria and homocystinuria, cblF type, 277380
LMBRD2	100%	100%	No OMIM disease ID
LMF1	100%	100%	Lipase deficiency, combined, 246650

LMNA	100%	100%	Mandibuloacral dysplasia, 248370 Heart-hand syndrome, Slovenian type, 610140 Cardiomyopathy, dilated, 1A, 115200 Restrictive dermopathy, lethal, 275210 Emery-Dreifuss muscular dystrophy 3, autosomal recessive, 616516 Charcot-Marie-Tooth disease, type 2B1, 605588 Emery-Dreifuss muscular dystrophy 2, autosomal dominant, 181350 Hutchinson-Gilford progeria, 176670 Lipodystrophy, familial partial, type 2, 151660 Muscular dystrophy, congenital, 613205 Malouf syndrome, 212112
LMNB1	100%	100%	Leukodystrophy, adult-onset, autosomal dominant, 169500 Microcephaly 26, primary, autosomal dominant, 619179
LMNB2	100%	99%	Microcephaly 27, primary, autosomal dominant, 619180 ?Epilepsy, progressive myoclonic, 9, 616540
LMOD1	100%	100%	?Megacystis-microcolon-intestinal hypoperistalsis syndrome 3, 619362
LMOD3	100%	100%	Nemaline myopathy 10, 616165
LMX1A	100%	100%	Deafness, autosomal dominant 7, 601412
LMX1B	100%	100%	Focal segmental glomerulosclerosis 10, 256020 Nail-patella syndrome, 161200
LNPK	93%	93%	Neurodevelopmental disorder with epilepsy and hypoplasia of the corpus callosum, 618090
LONP1	100%	100%	CODAS syndrome, 600373
LORICRIN	100%	100%	Vohwinkel syndrome with ichthyosis, 604117

LOX	100%	100%	Aortic aneurysm, familial thoracic 10, 617168
LOXHD1	100%	100%	Deafness, autosomal recessive 77, 613079
LOXL3	100%	100%	No OMIM disease ID
LPAR6	100%	100%	Hypotrichosis 8, 278150 Woolly hair, autosomal recessive 1, with or without hypotrichosis, 278150
LPIN1	100%	100%	Myoglobinuria, acute recurrent, autosomal recessive, 268200
LPIN2	100%	100%	Majeed syndrome, 609628
LPL	100%	100%	Lipoprotein lipase deficiency, 238600 Combined hyperlipidemia, familial, 144250
LPP	100%	100%	Leukemia, acute myeloid, 601626 Lipoma,
LRAT	100%	100%	Leber congenital amaurosis 14, 613341 Retinal dystrophy, early-onset severe, 613341 Retinitis pigmentosa, juvenile, 613341
LRBA	100%	100%	Immunodeficiency, common variable, 8, with autoimmunity, 614700
LRIF1	100%	100%	?Facioscapulohumeral muscular dystrophy 3, digenic, 619477
LRIG2	100%	100%	Urofacial syndrome 2, 615112
LRIG3	100%	100%	No OMIM disease ID
LRIT3	100%	100%	Night blindness, congenital stationary (complete), 1F, autosomal recessive, 615058
LRMDA	99%	99%	Albinism, oculocutaneous, type VII, 615179
LRP1	100%	100%	?Keratosis pilaris atrophicans, 604093
LRP12	100%	100%	Oculopharyngodistal myopathy 1, 164310
LRP2	100%	100%	Donnai-Barrow syndrome, 222448
LRP4	100%	100%	?Myasthenic syndrome, congenital, 17, 616304 Sclerosteosis 2, 614305 Cenani-Lenz syndactyly syndrome, 212780

LRP5	100%	100%	Osteopetrosis, autosomal dominant 1, 607634 Hyperostosis, endosteal, 144750 Osteosclerosis, 144750 Polycystic liver disease 4 with or without kidney cysts, 617875 Osteoporosis-pseudoglioma syndrome, 259770 Exudative vitreoretinopathy 4, 601813 van Buchem disease, type 2, 607636
LRP6	100%	100%	Tooth agenesis, selective, 7, 616724
LRPAP1	100%	100%	Myopia 23, autosomal recessive, 615431
LRPPRC	100%	100%	Mitochondrial complex IV deficiency, nuclear type 5, (French-Canadian), 220111
LRRC10	100%	100%	No OMIM disease ID
LRRC32	100%	100%	Cleft palate, proliferative retinopathy, and developmental delay, 619074
LRRC56	100%	100%	Ciliary dyskinesia, primary, 39, 618254
LRRC8A	100%	100%	?Agammaglobulinemia 5, 613506
LRRK1	100%	100%	No OMIM disease ID
LRRK2	100%	100%	No OMIM disease ID
LRSAM1	100%	100%	Charcot-Marie-Tooth disease, axonal, type 2P, 614436
LRTOMT	100%	100%	Deafness, autosomal recessive 63, 611451
LSM11	100%	100%	?Aicardi-Goutieres syndrome 8, 619486
LSS	100%	100%	Hypotrichosis 14, 618275 Cataract 44, 616509 Alopecia-mental retardation syndrome 4, 618840
LTBP1	100%	100%	Cutis laxa, autosomal recessive, type IIE, 619451
LTBP2	100%	100%	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%	Dental anomalies and short stature, 601216 Geleophysic dysplasia 3, 617809
LTBP4	100%	100%	Cutis laxa, autosomal recessive, type IC, 613177
LTC4S	100%	100%	No OMIM disease ID
LYRM4	66%	66%	?Combined oxidative phosphorylation deficiency 19, 615595
LYRM7	100%	100%	Mitochondrial complex III deficiency, nuclear type 8, 615838
LYST	100%	100%	Chediak-Higashi syndrome, 214500
LYZ	100%	100%	Amyloidosis, renal, 105200
LZTFL1	100%	100%	Bardet-Biedl syndrome 17, 615994
LZTR1	100%	100%	Noonan syndrome 2, 605275 Noonan syndrome 10, 616564
LZTS1	100%	100%	Esophageal squamous cell carcinoma, somatic, 133239
M1AP	100%	100%	Spermatogenic failure 48, 619108
MAB21L1	100%	100%	Cerebellar, ocular, craniofacial, and genital syndrome, 618479
MAB21L2	100%	100%	Microphthalmia/coloboma and skeletal dysplasia syndrome, 615877
MACF1	100%	100%	Lissencephaly 9 with complex brainstem malformation, 618325
MAD1L1	100%	100%	Prostate cancer, somatic, 176807 Lymphoma, somatic,
MAD2L2	100%	100%	?Fanconi anemia, complementation group V, 617243
MADD	100%	100%	Neurodevelopmental disorder with dysmorphic facies, impaired speech and hypotonia, 619005 DEEAH syndrome, 619004
MAF	93%	90%	Cataract 21, multiple types, 610202 Ayme-Gripp syndrome, 601088
MAFA	100%	99%	Insulinomatosis and diabetes mellitus, 147630
MAFB	100%	100%	Duane retraction syndrome 3, 617041 Multicentric carpotarsal osteolysis syndrome, 166300
MAG	100%	100%	Spastic paraplegia 75, autosomal recessive, 616680
MAGED2	100%	99%	Bartter syndrome, type 5, antenatal, transient, 300971
MAGEL2	100%	100%	Schaaf-Yang syndrome, 615547
MAGI2	97%	95%	Nephrotic syndrome, type 15, 617609

MAGT1	98%	98%	Immunodeficiency, X-linked, with magnesium defect, Epstein-Barr virus infection and neoplasia, 300853 Congenital disorder of glycosylation, type Icc, 301031
MAK	100%	100%	Retinitis pigmentosa 62, 614181
MAL2	100%	100%	No OMIM disease ID
MALT1	100%	100%	Immunodeficiency 12, 615468
MAML2	100%	100%	Mucoepidermoid salivary gland carcinoma,
MAMLD1	100%	100%	Hypospadias 2, X-linked, 300758
MAN1B1	100%	100%	Rafiq syndrome, 614202
MAN2B1	100%	100%	Mannosidosis, alpha-, types I and II, 248500
MAN2B2	100%	100%	No OMIM disease ID
MANBA	100%	100%	Mannosidosis, beta, 248510
MAOA	99%	98%	Brunner syndrome, 300615
MAP1B	100%	100%	Periventricular nodular heterotopia 9, 618918
MAP1LC3B2	100%	100%	No OMIM disease ID
MAP2K1	100%	100%	Cardiofaciocutaneous syndrome 3, 615279 Melorheostosis, isolated, somatic mosaic, 155950
MAP2K2	100%	100%	Cardiofaciocutaneous syndrome 4, 615280
MAP3K1	100%	100%	46XY sex reversal 6, 613762
MAP3K14	100%	100%	No OMIM disease ID
MAP3K20	100%	100%	Centronuclear myopathy 6 with fiber-type disproportion, 617760 Split-foot malformation with mesoaxial polydactyly, 616890
MAP3K7	100%	100%	Frontometaphyseal dysplasia 2, 617137 Cardiospondylocarpofacial syndrome, 157800
MAP3K8	100%	100%	Lung cancer, somatic, 211980
MAP4K4	100%	100%	No OMIM disease ID
MAPK1	100%	100%	Noonan syndrome 13, 619087

MAPK8	100%	100%	No OMIM disease ID
MAPK8IP3	100%	100%	Neurodevelopmental disorder with or without variable brain abnormalities, 618443
MAPKAPK3	100%	100%	?Macular dystrophy, patterned, 3, 617111
MAPKAPK5	100%	100%	No OMIM disease ID
MAPKBP1	100%	100%	Nephronophthisis 20, 617271
MAPRE2	100%	100%	Symmetric circumferential skin creases, congenital, 2, 616734
MAPT	100%	100%	Supranuclear palsy, progressive, 601104 Supranuclear palsy, progressive atypical, 260540 Dementia, frontotemporal, with or without parkinsonism, 600274 Pick disease, 172700
MARCHF6	100%	100%	Epilepsy, familial adult myoclonic, 3, 613608
MARK3	100%	100%	?Visual impairment and progressive phthisis bulbi, 618283
MARS1	100%	100%	Interstitial lung and liver disease, 615486 Charcot-Marie-Tooth disease, axonal, type 2U, 616280
MARS2	100%	100%	?Combined oxidative phosphorylation deficiency 25, 616430 Spastic ataxia 3, autosomal recessive, 611390
MARVELD2	100%	100%	Deafness, autosomal recessive 49, 610153
MASP1	100%	100%	3MC syndrome 1, 257920
MASP2	100%	100%	MASP2 deficiency, 613791
MAST1	100%	100%	Mega-corpus-callosum syndrome with cerebellar hypoplasia and cortical malformations, 618273
MASTL	100%	100%	No OMIM disease ID

MAT1A	100%	100%	Hypermethioninemia, persistent, autosomal dominant, due to methionine adenosyltransferase I/III deficiency, 250850 Methionine adenosyltransferase deficiency, autosomal recessive, 250850
MAT2A	100%	100%	No OMIM disease ID
MATN3	100%	100%	Spondyloepimetaphyseal dysplasia, Borochowitz-Cormier-Daire type, 608728 Epiphyseal dysplasia, multiple, 5, 607078
MATR3	100%	100%	Amyotrophic lateral sclerosis 21, 606070
MAX	100%	100%	No OMIM disease ID
MBD5	100%	100%	Mental retardation, autosomal dominant 1, 156200
MBL2	100%	100%	No OMIM disease ID
MBOAT7	100%	100%	Mental retardation, autosomal recessive 57, 617188
MBTPS1	100%	100%	?Spondyloepiphyseal dysplasia, Kondo-Fu type, 618392
MBTPS2	100%	100%	Keratosis follicularis spinulosa decalvans, X-linked, 308800 Osteogenesis imperfecta, type XIX, 301014 IFAP syndrome with or without BRESHECK syndrome, 308205 ?Olmsted syndrome, X-linked, 300918
MC1R	100%	100%	No OMIM disease ID
MC2R	100%	100%	Glucocorticoid deficiency, due to ACTH unresponsiveness, 202200
MC4R	100%	100%	Obesity (BMIQ20), 618406
MCAT	100%	100%	No OMIM disease ID
MCC	100%	100%	Colorectal cancer, somatic, 114500
MCCC1	100%	100%	3-Methylcrotonyl-CoA carboxylase 1 deficiency, 210200
MCCC2	100%	100%	3-Methylcrotonyl-CoA carboxylase 2 deficiency, 210210
MCEE	100%	100%	Methylmalonyl-CoA epimerase deficiency, 251120
MCFD2	100%	100%	Factor V and factor VIII, combined deficiency of, 613625

MCIDAS	100%	100%	Ciliary dyskinesia, primary, 42, 618695
MCM10	100%	100%	Immunodeficiency 80 with or without cardiomyopathy, 619313
MCM2	100%	100%	?Deafness, autosomal dominant 70, 616968
MCM3AP	100%	100%	Peripheral neuropathy, autosomal recessive, with or without impaired intellectual development, 618124
MCM4	95%	95%	Immunodeficiency 54, 609981
MCM5	100%	100%	?Meier-Gorlin syndrome 8, 617564
MCM6	100%	100%	Lactase persistence/nonpersistence, 223100
MCM8	94%	94%	?Premature ovarian failure 10, 612885
MCM9	100%	100%	Ovarian dysgenesis 4, 616185
MCOLN1	100%	100%	Mucolipidosis IV, 252650
MCPH1	100%	100%	Microcephaly 1, primary, autosomal recessive, 251200
MCTP2	100%	100%	No OMIM disease ID
MCUR1	100%	100%	No OMIM disease ID
MDH1	100%	100%	?Developmental and epileptic encephalopathy 88, 618959
MDH2	100%	100%	Developmental and epileptic encephalopathy 51, 617339
MDM2	92%	92%	?Lessel-Kubisch syndrome, 618681
MDM4	100%	100%	?Bone marrow failure syndrome 6, 618849
MECOM	100%	100%	Radioulnar synostosis with amegakaryocytic thrombocytopenia 2, 616738
MECP2	100%	99%	Intellectual developmental disorder, X-linked, syndromic 13, 300055 Rett syndrome, atypical, 312750 Encephalopathy, neonatal severe, 300673 Intellectual developmental disorder, X-linked syndromic, Lubs type, 300260 Rett syndrome, 312750 Rett syndrome, preserved speech variant, 312750
MECR	100%	100%	Dystonia, childhood-onset, with optic atrophy and basal ganglia abnormalities, 617282
MED12	100%	100%	Lujan-Fryns syndrome, 309520 Ohdo syndrome, X-linked, 300895 Opitz-Kaveggia syndrome, 305450
MED12L	100%	100%	Nizon-Isidor syndrome, 618872
MED13	100%	100%	Intellectual developmental disorder 61, 618009
MED13L	100%	100%	Impaired intellectual development and distinctive facial features with or without cardiac defects, 616789

MED17	100%	100%	Microcephaly, postnatal progressive, with seizures and brain atrophy, 613668
MED23	100%	100%	Mental retardation, autosomal recessive 18, 614249
MED25	100%	100%	Basel-Vanagait-Smirin-Yosef syndrome, 616449
MED27	84%	84%	Neurodevelopmental disorder with spasticity, cataracts, and cerebellar hypoplasia, 619286
MEF2C	100%	100%	Neurodevelopmental disorder with hypotonia, stereotypic hand movements, and impaired language, 613443
MEFV	96%	96%	Neutrophilic dermatosis, acute febrile, 608068 Familial Mediterranean fever, AR, 249100 Familial Mediterranean fever, AD, 134610
MEGF10	100%	100%	Myopathy, areflexia, respiratory distress, and dysphagia, early-onset, mild variant, 614399 Myopathy, areflexia, respiratory distress, and dysphagia, early-onset, 614399
MEGF8	100%	100%	Carpenter syndrome 2, 614976
MEI1	100%	100%	Hydatidiform mole, recurrent, 3, 618431
MEIOB	100%	100%	?Spermatogenic failure 22, 617706
MEIS2	100%	100%	Cleft palate, cardiac defects, and mental retardation, 600987
MEN1	100%	100%	Multiple endocrine neoplasia 1, 131100 Lipoma, somatic, Angiofibroma, somatic, Carcinoid tumor of lung, Adrenal adenoma, somatic, Parathyroid adenoma, somatic,
MEOX1	100%	100%	Klippel-Feil syndrome 2, 214300
MERTK	99%	99%	Retinitis pigmentosa 38, 613862
MESD	100%	100%	Osteogenesis imperfecta, type XX, 618644
MESP2	97%	97%	Spondylocostal dysostosis 2, autosomal recessive, 608681

MET	100%	100%	Renal cell carcinoma, papillary, 1, familial and somatic, 605074 Hepatocellular carcinoma, childhood type, somatic, 114550 ?Deafness, autosomal recessive 97, 616705
METTL13	100%	100%	No OMIM disease ID
METTL23	100%	100%	Mental retardation, autosomal recessive 44, 615942
METTL5	100%	99%	Intellectual developmental disorder, autosomal recessive 72, 618665
MFAP5	100%	100%	Aortic aneurysm, familial thoracic 9, 616166
MFF	100%	100%	Encephalopathy due to defective mitochondrial and peroxisomal fission 2, 617086
MFN2	100%	100%	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%	Microphthalmia, isolated 5, 611040 Nanophthalmos 2, 609549
MFSD2A	100%	100%	Neurodevelopmental disorder with progressive microcephaly, spasticity, and brain abnormalities, 616486
MFSD8	100%	100%	Macular dystrophy with central cone involvement, 616170 Ceroid lipofuscinosis, neuronal, 7, 610951
MGAT2	100%	100%	Congenital disorder of glycosylation, type IIa, 212066
MGME1	100%	100%	Mitochondrial DNA depletion syndrome 11, 615084
MGP	100%	100%	Keutel syndrome, 245150
MIA3	100%	100%	?Ondontochondrodysplasia 2 with hearing loss and diabetes, 619269
MIB1	100%	100%	Left ventricular noncompaction 7, 615092
MICOS13	100%	100%	Combined oxidative phosphorylation deficiency 37, 618329
MICU1	100%	100%	Myopathy with extrapyramidal signs, 615673
MICU2	100%	100%	No OMIM disease ID
MID1	100%	100%	Opitz GBBB syndrome, type I, 300000
MID2	100%	100%	?Intellectual developmental disorder, X-linked 101, 300928
MIEF2	100%	100%	?Combined oxidative phosphorylation deficiency 49, 619024

MINPP1	100%	100%	Pontocerebellar hypoplasia, type 16, 619527
MIP	100%	100%	Cataract 15, multiple types, 615274
MIPEP	100%	100%	Combined oxidative phosphorylation deficiency 31, 617228
MIR140	NC	NC	Spondyloepiphyseal dysplasia, Nishimura type, 618618
MIR17HG	NC	NC	No OMIM disease ID
MIR184	NC	NC	EDICT syndrome, 614303
MIR204	NC	NC	?Retinal dystrophy and iris coloboma with or without cataract, 616722
MIR96	NC	NC	Deafness, autosomal dominant 50, 613074
MITF	100%	100%	Waardenburg syndrome, type 2A, 193510 Tietz albinism-deafness syndrome, 103500 Waardenburg syndrome/ocular albinism, digenic, 103470 COMMAD syndrome, 617306
MKKS	100%	100%	McKusick-Kaufman syndrome, 236700 Bardet-Biedl syndrome 6, 605231
MKRN3	96%	96%	Precocious puberty, central, 2, 615346
MKS1	100%	100%	Bardet-Biedl syndrome 13, 615990 Meckel syndrome 1, 249000 Joubert syndrome 28, 617121
MLC1	100%	100%	Megalencephalic leukoencephalopathy with subcortical cysts, 604004
MLH1	100%	100%	Colorectal cancer, hereditary nonpolyposis, type 2, 609310 Muir-Torre syndrome, 158320 Mismatch repair cancer syndrome 1, 276300
MLH3	100%	100%	Colorectal cancer, somatic, 114500 Colorectal cancer, hereditary nonpolyposis, type 7, 614385
MLIP	100%	100%	No OMIM disease ID
MLLT10	97%	97%	Leukemia, acute myeloid, 601626
MLLT6	100%	100%	No OMIM disease ID
MLPH	100%	100%	Griscelli syndrome, type 3, 609227
MLYCD	100%	100%	Malonyl-CoA decarboxylase deficiency, 248360

MMAA	100%	100%	Methylmalonic aciduria, vitamin B12-responsive, cblA type, 251100
MMAB	100%	100%	Methylmalonic aciduria, vitamin B12-responsive, cblB type, 251110
MMACHC	100%	100%	Methylmalonic aciduria and homocystinuria, cblC type, 277400
MMADHC	89%	89%	Methylmalonic aciduria, cblD type, variant 2, 277410 Methylmalonic aciduria and homocystinuria, cblD type, 277410 Homocystinuria, cblD type, variant 1, 277410
MME	98%	98%	?Spinocerebellar ataxia 43, 617018 Charcot-Marie-Tooth disease, axonal, type 2T, 617017
MMGT1	100%	100%	No OMIM disease ID
MMP1	100%	100%	COPD, rate of decline of lung function in, 606963
MMP13	92%	92%	?Spondyloepimetaphyseal dysplasia, Missouri type, 602111 Metaphyseal anadysplasia 1, 602111 Metaphyseal dysplasia, Spahr type, 250400
MMP14	100%	100%	?Winchester syndrome, 277950
MMP19	100%	100%	Cavitary optic disc anomalies, 611543
MMP2	100%	100%	Multicentric osteolysis, nodulosis, and arthropathy, 259600
MMP20	100%	100%	Amelogenesis imperfecta, type IIA2, 612529
MMP21	100%	100%	Heterotaxy, visceral, 7, autosomal, 616749
MMP9	100%	100%	Metaphyseal anadysplasia 2, 613073
MMUT	100%	100%	Methylmalonic aciduria, mut(0) type, 251000
MN1	100%	100%	CEBALID syndrome, 618774 Meningioma, 607174
MNS1	100%	100%	Heterotaxy, visceral, 9, autosomal, with male infertility, 618948
MNX1	96%	89%	Currarino syndrome, 176450
MOCOS	100%	100%	Xanthinuria, type II, 603592
MOCS1	100%	100%	Molybdenum cofactor deficiency A, 252150
MOCS2	100%	100%	Molybdenum cofactor deficiency B, 252160
MOG	100%	100%	?Narcolepsy 7, 614250
MOGS	100%	100%	Congenital disorder of glycosylation, type IIb, 606056

MORC2	100%	100%	Charcot-Marie-Tooth disease, axonal, type 2Z, 616688 Developmental delay, impaired growth, dysmorphic facies, and axonal neuropathy, 619090
MPC1	100%	100%	Mitochondrial pyruvate carrier deficiency, 614741
MPDU1	100%	100%	Congenital disorder of glycosylation, type If, 609180
MPDZ	100%	100%	Hydrocephalus, congenital, 2, with or without brain or eye anomalies, 615219
MPEG1	100%	100%	Immunodeficiency 77, 619223
MPI	100%	100%	Congenital disorder of glycosylation, type Ib, 602579
MPIG6B	100%	100%	?Thrombocytopenia, anemia, and myelofibrosis, 617441
MPL	100%	100%	Myelofibrosis with myeloid metaplasia, somatic, 254450 Thrombocythemia 2, 601977 Thrombocytopenia, congenital amegakaryocytic, 604498
MPLKIP	100%	100%	Trichothiodystrophy 4, nonphotosensitive, 234050
MPO	100%	100%	Myeloperoxidase deficiency, 254600
MPV17	100%	100%	Charcot-Marie-Tooth disease, axonal, type 2EE, 618400 Mitochondrial DNA depletion syndrome 6 (hepatocerebral type), 256810
MPZ	100%	100%	Charcot-Marie-Tooth disease, type 2I, 607677 Dejerine-Sottas disease, 145900 Charcot-Marie-Tooth disease, type 1B, 118200 Roussy-Levy syndrome, 180800 Charcot-Marie-Tooth disease, dominant intermediate D, 607791 Hypomyelinating neuropathy, congenital, 2, 618184 Charcot-Marie-Tooth disease, type 2J, 607736

MPZL2	100%	100%	Deafness, autosomal recessive 111, 618145
MRAP	100%	100%	Glucocorticoid deficiency 2, 607398
MRAS	100%	100%	Noonan syndrome 11, 618499
MRE11	100%	100%	Ataxia-telangiectasia-like disorder 1, 604391
MRM2	98%	98%	?Mitochondrial DNA depletion syndrome 17, 618567
MRPL12	100%	100%	?Combined oxidative phosphorylation deficiency 45, 618951
MRPL24	100%	100%	No OMIM disease ID
MRPL3	100%	100%	Combined oxidative phosphorylation deficiency 9, 614582
MRPL40	100%	100%	No OMIM disease ID
MRPL44	100%	100%	Combined oxidative phosphorylation deficiency 16, 615395
MRPL57	100%	100%	No OMIM disease ID
MRPS14	100%	100%	?Combined oxidative phosphorylation deficiency 38, 618378
MRPS16	100%	100%	Combined oxidative phosphorylation deficiency 2, 610498
MRPS2	100%	100%	Combined oxidative phosphorylation deficiency 36, 617950
MRPS22	100%	100%	Ovarian dysgenesis 7, 618117 Combined oxidative phosphorylation deficiency 5, 611719
MRPS23	100%	100%	?Combined oxidative phosphorylation defiency 46, 618952
MRPS25	82%	82%	?Combined oxidative phosphorylation deficiency 50, 619025
MRPS28	86%	86%	?Combined oxidative phosphorylation deficiency 47, 618958
MRPS34	100%	100%	Combined oxidative phosphorylation deficiency 32, 617664
MRPS36	100%	100%	No OMIM disease ID
MRPS7	100%	100%	?Combined oxidative phosphorylation deficiency 34, 617872
MRRF	100%	100%	No OMIM disease ID
MRTFA	92%	92%	?Immunodeficiency 66, 618847
MS4A1	100%	100%	?Immunodeficiency, common variable, 5, 613495
MSH2	100%	100%	Muir-Torre syndrome, 158320 Colorectal cancer, hereditary nonpolyposis, type 1, 120435 Mismatch repair cancer syndrome 2, 619096
MSH3	100%	100%	Familial adenomatous polyposis 4, 617100 Endometrial carcinoma, somatic, 608089

MSH4	100%	100%	No OMIM disease ID
MSH5	100%	100%	?Premature ovarian failure 13, 617442
MSH6	100%	100%	Colorectal cancer, hereditary nonpolyposis, type 5, 614350 Mismatch repair cancer syndrome 3, 619097
MSL2	100%	100%	No OMIM disease ID
MSL3	98%	97%	Basilicata-Akhtar syndrome, 301032
MSMO1	100%	100%	Microcephaly, congenital cataract, and psoriasiform dermatitis, 616834
MSN	100%	100%	Immunodeficiency 50, 300988
MSR1	100%	100%	Barrett esophagus/esophageal adenocarcinoma, 614266
MSRB3	100%	100%	Deafness, autosomal recessive 74, 613718
MSTN	100%	100%	?Muscle hypertrophy, 614160
MSTO1	100%	100%	Myopathy, mitochondrial, and ataxia, 617675
MSX1	100%	100%	Tooth agenesis, selective, 1, with or without orofacial cleft, 106600 Ectodermal dysplasia 3, Witkop type, 189500 Orofacial cleft 5, 608874
MSX2	100%	100%	Parietal foramina with cleidocranial dysplasia, 168550 Craniosynostosis 2, 604757 Parietal foramina 1, 168500
MTAP	100%	100%	Diaphyseal medullary stenosis with malignant fibrous histiocytoma, 112250
MTFMT	100%	100%	Combined oxidative phosphorylation deficiency 15, 614947 Mitochondrial complex I deficiency, nuclear type 27, 618248
MTHFD1	100%	100%	Combined immunodeficiency and megaloblastic anemia with or without hyperhomocysteinemia, 617780
MTHFR	100%	100%	Homocystinuria due to MTHFR deficiency, 236250
MTHFS	100%	100%	Neurodevelopmental disorder with microcephaly, epilepsy, and hypomyelination, 618367
MTM1	100%	100%	Myotubular myopathy, X-linked, 310400
MTMR2	100%	100%	Charcot-Marie-Tooth disease, type 4B1, 601382
MTO1	94%	92%	Combined oxidative phosphorylation deficiency 10, 614702

MTOR	100%	100%	Focal cortical dysplasia, type II, somatic, 607341 Smith-Kingsmore syndrome, 616638
MTPAP	100%	100%	?Spastic ataxia 4, autosomal recessive, 613672
MTR	100%	100%	Homocystinuria-megaloblastic anemia, cblG complementation type, 250940
MTRFR	100%	100%	Spastic paraplegia 55, autosomal recessive, 615035 Combined oxidative phosphorylation deficiency 7, 613559
MTRR	100%	100%	Homocystinuria-megaloblastic anemia, cbl E type, 236270
MTTP	100%	100%	Abetalipoproteinemia, 200100
MTX2	100%	100%	Mandibuloacral dysplasia progeroid syndrome, 619127
MUC1	100%	100%	Tubulointerstitial kidney disease, autosomal dominant, 2, 174000
MUSK	100%	100%	Fetal akinesia deformation sequence 1, 208150 Myasthenic syndrome, congenital, 9, associated with acetylcholine receptor deficiency, 616325
MUTYH	100%	100%	Adenomas, multiple colorectal, 608456 Gastric cancer, somatic, 613659
MVD	100%	100%	Porokeratosis 7, multiple types, 614714
MVK	90%	90%	Hyper-IgD syndrome, 260920 Porokeratosis 3, multiple types, 175900 Mevalonic aciduria, 610377
MXI1	100%	99%	Prostate cancer, somatic, 176807 Neurofibrosarcoma, somatic,
MYBPC1	100%	100%	Myopathy, congenital, with tremor, 618524 Lethal congenital contracture syndrome 4, 614915 Arthrogryposis, distal, type 1B, 614335

MYBPC3	100%	100%	Cardiomyopathy, hypertrophic, 4, 115197 Cardiomyopathy, dilated, 1MM, 615396 Left ventricular noncompaction 10, 615396
MYBPHL	100%	100%	No OMIM disease ID
MYC	100%	100%	Burkitt lymphoma, somatic, 113970
MYCN	100%	100%	Feingold syndrome 1, 164280
MYD88	100%	100%	Macroglobulinemia, Waldenstrom, somatic, 153600 Immunodeficiency 68, 612260
MYF5	100%	100%	Ophthalmoplegia, external, with rib and vertebral anomalies, 618155
MYH11	100%	100%	Megacystis-microcolon-intestinal hypoperistalsis syndrome 2, 619351 Aortic aneurysm, familial thoracic 4, 132900 Visceral myopathy 2, 619350
MYH14	100%	100%	?Peripheral neuropathy, myopathy, hoarseness, and hearing loss, 614369 Deafness, autosomal dominant 4A, 600652
MYH2	100%	100%	Proximal myopathy and ophthalmoplegia, 605637
MYH3	100%	100%	Contractures, pterygia, and spondylocarpostarsal fusion syndrome 1A, 178110 Contractures, pterygia, and spondylocarpotarsal fusion syndrome 1B, 618469 Arthrogryposis, distal, type 2B3 (Sheldon-Hall), 618436 Arthrogryposis, distal, type 2A (Freeman-Sheldon), 193700

MYH6	100%	100%	Atrial septal defect 3, 614089 Cardiomyopathy, dilated, 1EE, 613252 Cardiomyopathy, hypertrophic, 14, 613251
MYH7	100%	100%	Laing distal myopathy, 160500 Cardiomyopathy, hypertrophic, 1, 192600 Left ventricular noncompaction 5, 613426 Cardiomyopathy, dilated, 1S, 613426 Scapuloperoneal syndrome, myopathic type, 181430 Myopathy, myosin storage, autosomal dominant, 608358 Myopathy, myosin storage, autosomal recessive, 255160
MYH7B	100%	100%	No OMIM disease ID
MYH8	100%	100%	Carney complex variant, 608837 Trismus-pseudocamptodactyly syndrome, 158300
MYH9	100%	100%	Macrothrombocytopenia and granulocyte inclusions with or without nephritis or sensorineural hearing loss, 155100 Deafness, autosomal dominant 17, 603622
MYL1	100%	100%	Myopathy, congenital, with fast-twitch (type II) fiber atrophy, 618414
MYL2	99%	99%	Cardiomyopathy, hypertrophic, 10, 608758 Myopathy, myofibrillar, 12, infantile-onset, with cardiomyopathy, 619424
MYL3	100%	100%	Cardiomyopathy, hypertrophic, 8, 608751
MYL4	100%	100%	?Atrial fibrillation, familial, 18, 617280
MYL7	100%	100%	No OMIM disease ID
MYL9	100%	100%	?Megacystis-microcolon-intestinal hypoperistalsis syndrome 4, 619365

MYLK	100%	100%	Megacystis-microcolon-intestinal hypoperistalsis syndrome 1, 249210 Aortic aneurysm, familial thoracic 7, 613780
MYLK2	100%	100%	Cardiomyopathy, hypertrophic, 1, digenic, 192600
MYLK3	100%	100%	No OMIM disease ID
MYLPF	100%	100%	Arthrogryposis, distal, type 1C, 619110
MYMK	100%	100%	Carey-Fineman-Ziter syndrome, 254940
MYO15A	100%	100%	Deafness, autosomal recessive 3, 600316
MYO18B	100%	100%	Klippel-Feil syndrome 4, autosomal recessive, with myopathy and facial dysmorphism, 616549
MYO1A	100%	100%	No OMIM disease ID
MYO1E	100%	100%	Glomerulosclerosis, focal segmental, 6, 614131
MYO1H	100%	100%	?Central hypoventilation syndrome, congenital, 2, and autonomic dysfunction, 619482
MYO3A	100%	100%	Deafness, autosomal recessive 30, 607101
MYO5A	100%	100%	Griscelli syndrome, type 1, 214450
MYO5B	100%	100%	Diarrhea 2, with microvillus atrophy, 251850
MYO6	100%	100%	Deafness, autosomal dominant 22, with hypertrophic cardiomyopathy, 606346 Deafness, autosomal dominant 22, 606346 Deafness, autosomal recessive 37, 607821
MYO7A	100%	100%	Deafness, autosomal recessive 2, 600060 Usher syndrome, type 1B, 276900 Deafness, autosomal dominant 11, 601317
MYO9A	100%	100%	Myasthenic syndrome, congenital, 24, presynaptic, 618198
MYOC	100%	100%	Glaucoma 1A, primary open angle, 137750
MYOCD	100%	100%	Megabladder, congenital, 618719
MYOD1	100%	100%	Myopathy, congenital, with diaphragmatic defects, respiratory insufficiency, and dysmorphic facies, 618975
MYOF	100%	100%	?Angioedema, hereditary, 7, 619366
MYOM1	100%	100%	No OMIM disease ID
MYORG	100%	100%	Basal ganglia calcification, idiopathic, 7, autosomal recessive, 618317

MYOT	100%	100%	Myopathy, myofibrillar, 3, 609200 Myopathy, spheroid body, 182920
MYOZ2	100%	100%	Cardiomyopathy, hypertrophic, 16, 613838
MYPN	100%	100%	Cardiomyopathy, hypertrophic, 22, 615248 Cardiomyopathy, familial restrictive, 4, 615248 Cardiomyopathy, dilated, 1KK, 615248 Nemaline myopathy 11, autosomal recessive, 617336
MYRF	100%	100%	Encephalitis/encephalopathy, mild, with reversible myelin vacuolization, 618113 Cardiac-urogenital syndrome, 618280
MYSM1	96%	96%	Bone marrow failure syndrome 4, 618116
MYT1L	90%	90%	Mental retardation, autosomal dominant 39, 616521
NAA10	100%	100%	Microphthalmia, syndromic 1, 309800 Ogden syndrome, 300855
NAA15	96%	96%	Intellectual developmental disorder, autosomal dominant 50, with behavioral abnormalities, 617787
NAA20	100%	100%	No OMIM disease ID
NACC1	100%	100%	Neurodevelopmental disorder with epilepsy, cataracts, feeding difficulties, and delayed brain myelination, 617393
NADK2	100%	100%	2,4-dienoyl-CoA reductase deficiency, 616034
NADSYN1	100%	100%	Vertebral, cardiac, renal, and limb defects syndrome 3, 618845
NAGA	100%	100%	Schindler disease, type I, 609241 Kanzaki disease, 609242 Schindler disease, type III, 609241
NAGLU	100%	100%	?Charcot-Marie-Tooth disease, axonal, type 2V, 616491 Mucopolysaccharidosis type IIIB (Sanfilippo B), 252920
NAGS	100%	100%	N-acetylglutamate synthase deficiency, 237310

NALCN	99%	99%	Congenital contractures of the limbs and face, hypotonia, and developmental delay, 616266 Hypotonia, infantile, with psychomotor retardation and characteristic facies 1, 615419
NANOS1	99%	99%	Spermatogenic failure 12, 615413
NANS	100%	100%	Spondyloepimetaphyseal dysplasia, Camera-Genevieve type, 610442
NARS1	100%	100%	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	100%	100%	Combined oxidative phosphorylation deficiency 24, 616239 ?Deafness, autosomal recessive 94, 618434
NAT8L	99%	96%	?N-acetylaspartate deficiency, 614063
NAXD	100%	100%	Encephalopathy, progressive, early-onset, with brain edema and/or leukoencephalopathy, 2, 618321
NAXE	100%	100%	Encephalopathy, progressive, early-onset, with brain edema and/or leukoencephalopathy, 617186
NBAS	100%	100%	Short stature, optic nerve atrophy, and Pelger-Huet anomaly, 614800 Infantile liver failure syndrome 2, 616483
NBEA	100%	100%	Neurodevelopmental disorder with or without early-onset generalized epilepsy, 619157
NBEAL2	100%	100%	Gray platelet syndrome, 139090
NBN	100%	100%	Leukemia, acute lymphoblastic, 613065 Aplastic anemia, 609135 Nijmegen breakage syndrome, 251260
NCAPD2	100%	100%	?Microcephaly 21, primary, autosomal recessive, 617983

NCAPD3	100%	100%	Microcephaly 22, primary, autosomal recessive, 617984
NCAPG2	100%	100%	Khan-Khan-Katsanis syndrome, 618460
NCAPH	100%	100%	?Microcephaly 23, primary, autosomal recessive, 617985
NCDN	100%	100%	Neurodevelopmental disorder with infantile epileptic spasms, 619373
NCF1	100%	99%	Chronic granulomatous disease 1, autosomal recessive, 233700
NCF2	100%	100%	Chronic granulomatous disease 2, autosomal recessive, 233710
NCF4	100%	100%	Chronic granulomatous disease 3, autosomal recessive, 613960
NCKAP1	100%	100%	No OMIM disease ID
NCKAP1L	100%	100%	Immunodeficiency 72 with autoinflammation, 618982
NCOA3	100%	100%	No OMIM disease ID
NCOA4	100%	100%	No OMIM disease ID
NCSTN	100%	100%	Acne inversa, familial, 1, 142690
NDE1	100%	100%	Lissencephaly 4 (with microcephaly), 614019 ?Microhydranencephaly, 605013
NDN	100%	100%	Prader-Willi syndrome, 176270
NDNF	100%	100%	Hypogonadotropic hypogonadism 25 with anosmia, 618841
NDP	100%	100%	Exudative vitreoretinopathy 2, X-linked, 305390 Norrie disease, 310600
NDRG1	100%	100%	Charcot-Marie-Tooth disease, type 4D, 601455
NDST1	100%	100%	Mental retardation, autosomal recessive 46, 616116
NDUFA1	100%	100%	Mitochondrial complex I deficiency, nuclear type 12, 301020
NDUFA10	100%	100%	Mitochondrial complex I deficiency, nuclear type 22, 618243
NDUFA11	100%	100%	Mitochondrial complex I deficiency, nuclear type 14, 618236
NDUFA12	100%	100%	Mitochondrial complex I deficiency, nuclear type 23, 618244
NDUFA13	100%	100%	?Mitochondrial complex I deficiency, nuclear type 28, 618249
NDUFA2	100%	100%	Mitochondrial complex I deficiency, nuclear type 13, 618235
NDUFA3	94%	88%	No OMIM disease ID
NDUFA4	100%	100%	?Mitochondrial complex IV deficiency, nuclear type 21, 619065
NDUFA5	100%	100%	No OMIM disease ID
NDUFA6	100%	100%	Mitochondrial complex I deficiency, nuclear type 33, 618253
NDUFA7	100%	100%	No OMIM disease ID
NDUFA8	100%	100%	Mitochondrial complex I deficiency, nuclear type 37, 619272

NDUFA9	100%	100%	Mitochondrial complex I deficiency, nuclear type 26, 618247
NDUFAB1	100%	100%	No OMIM disease ID
NDUFAF1	100%	100%	Mitochondrial complex I deficiency, nuclear type 11, 618234
NDUFAF2	100%	100%	Mitochondrial complex I deficiency, nuclear type 10, 618233
NDUFAF3	100%	100%	Mitochondrial complex I deficiency, nuclear type 18, 618240
NDUFAF4	100%	100%	Mitochondrial complex I deficiency, nuclear type 15, 618237
NDUFAF5	100%	100%	Mitochondrial complex I deficiency, nuclear type 16, 618238
NDUFAF6	100%	100%	Mitochondrial complex I deficiency, nuclear type 17, 618239 Fanconi renotubular syndrome 5, 618913
NDUFAF7	100%	100%	No OMIM disease ID
NDUFAF8	100%	100%	Mitochondrial complex I deficiency, nuclear type 34, 618776
NDUFB1	100%	100%	No OMIM disease ID
NDUFB10	100%	100%	?Mitochondrial complex I deficiency, nuclear type 35, 619003
NDUFB11	100%	99%	Linear skin defects with multiple congenital anomalies 3, 300952 ?Mitochondrial complex I deficiency, nuclear type 30, 301021
NDUFB2	100%	100%	No OMIM disease ID
NDUFB3	100%	100%	Mitochondrial complex I deficiency, nuclear type 25, 618246
NDUFB4	100%	100%	No OMIM disease ID
NDUFB5	100%	100%	No OMIM disease ID
NDUFB6	100%	100%	No OMIM disease ID
NDUFB7	100%	100%	No OMIM disease ID
NDUFB8	100%	100%	Mitochondrial complex I deficiency, nuclear type 32, 618252
NDUFB9	98%	98%	?Mitochondrial complex I deficiency, nuclear type 24, 618245
NDUFC1	100%	100%	No OMIM disease ID
NDUFC2	100%	100%	Mitochondrial complex I deficiency, nuclear type 36, 619170
NDUFS1	100%	100%	Mitochondrial complex I deficiency, nuclear type 5, 618226
NDUFS2	100%	100%	Mitochondrial complex I deficiency, nuclear type 6, 618228
NDUFS3	95%	91%	Mitochondrial complex I deficiency, nuclear type 8, 618230
NDUFS4	100%	100%	Mitochondrial complex I deficiency, nuclear type 1, 252010
NDUFS5	100%	100%	No OMIM disease ID

NDUFS6	100%	100%	Mitochondrial complex I deficiency, nuclear type 9, 618232
NDUFS7	100%	100%	Mitochondrial complex I deficiency, nuclear type 3, 618224
NDUFS8	100%	100%	Mitochondrial complex I deficiency, nuclear type 2, 618222
NDUFV1	100%	100%	Mitochondrial complex I deficiency, nuclear type 4, 618225
NDUFV2	100%	100%	Mitochondrial complex I deficiency, nuclear type 7, 618229
NDUFV3	100%	100%	No OMIM disease ID
NEB	99%	99%	Nemaline myopathy 2, autosomal recessive, 256030 Arthrogryposis multiplex congenita 6, 619334
NEBL	100%	100%	No OMIM disease ID
NECAP1	100%	100%	Developmental and epileptic encephalopathy 21, 615833
NECTIN1	100%	100%	Cleft lip/palate-ectodermal dysplasia syndrome, 225060 Orofacial cleft 7, 225060
NECTIN4	100%	100%	Ectodermal dysplasia-syndactyly syndrome 1, 613573
NEDD4L	100%	100%	Periventricular nodular heterotopia 7, 617201
NEFH	100%	100%	Charcot-Marie-Tooth disease, axonal, type 2CC, 616924
NEFL	100%	100%	Charcot-Marie-Tooth disease, type 1F, 607734 Charcot-Marie-Tooth disease, dominant intermediate G, 617882 Charcot-Marie-Tooth disease, type 2E, 607684
NEK1	100%	100%	Short-rib thoracic dysplasia 6 with or without polydactyly, 263520
NEK10	100%	100%	Ciliary dyskinesia, primary, 44, 618781
NEK11	100%	100%	No OMIM disease ID
NEK2	96%	96%	?Retinitis pigmentosa 67, 615565
NEK4	94%	94%	No OMIM disease ID
NEK8	100%	100%	Renal-hepatic-pancreatic dysplasia 2, 615415 ?Nephronophthisis 9, 613824

NEK9	100%	100%	?Arthrogryposis, Perthes disease, and upward gaze palsy, 614262 Nevus comedonicus, somatic, 617025 Lethal congenital contracture syndrome 10, 617022
NEMF	100%	100%	Intellectual developmental disorder with speech delay and axonal peripheral neuropathy, 619099
NEPRO	100%	100%	Anauxetic dysplasia 3, 618853
NEU1	100%	100%	Sialidosis, type II, 256550 Sialidosis, type I, 256550
NEUROD1	100%	100%	Maturity-onset diabetes of the young 6, 606394
NEUROD2	100%	100%	Developmental and epileptic encephalopathy 72, 618374
NEUROG3	100%	100%	Diarrhea 4, malabsorptive, congenital, 610370
NEXMIF	100%	100%	Intellectual developmental disorder, X-linked 98, 300912
NEXN	100%	100%	Cardiomyopathy, dilated, 1CC, 613122 Cardiomyopathy, hypertrophic, 20, 613876
NF1	100%	100%	Watson syndrome, 193520 Leukemia, juvenile myelomonocytic, 607785 Neurofibromatosis, familial spinal, 162210 Neurofibromatosis, type 1, 162200 Neurofibromatosis-Noonan syndrome, 601321
NF2	100%	100%	Neurofibromatosis, type 2, 101000 Meningioma, NF2-related, somatic, 607174 Schwannomatosis, somatic, 162091
NFASC	100%	100%	Neurodevelopmental disorder with central and peripheral motor dysfunction, 618356
NFAT5	100%	100%	No OMIM disease ID
NFATC1	100%	100%	No OMIM disease ID
NFE2	100%	100%	No OMIM disease ID
NFE2L2	100%	100%	Immunodeficiency, developmental delay, and hypohomocysteinemia, 617744
NFIA	99%	99%	Brain malformations with or without urinary tract defects, 613735
NFIB	100%	100%	Macrocephaly, acquired, with impaired intellectual development, 618286

NFIX	99%	99%	Marshall-Smith syndrome, 602535 Sotos syndrome 2, 614753
NFKB1	100%	100%	Immunodeficiency, common variable, 12, 616576
NFKB2	100%	100%	Immunodeficiency, common variable, 10, 615577
NFKBIA	100%	100%	Ectodermal dysplasia and immunodeficiency 2, 612132
NFS1	89%	89%	Combined oxidative phosphorylation deficiency 52, 619386
NFU1	100%	100%	Multiple mitochondrial dysfunctions syndrome 1, 605711
NGF	100%	100%	Neuropathy, hereditary sensory and autonomic, type V, 608654
NGLY1	100%	100%	Congenital disorder of deglycosylation, 615273
NHEJ1	100%	100%	Severe combined immunodeficiency with microcephaly, growth retardation, and sensitivity to ionizing radiation, 611291
NHLRC1	100%	100%	Epilepsy, progressive myoclonic 2B (Lafora), 254780
NHLRC2	100%	100%	FINCA syndrome, 618278
NHP2	100%	100%	Dyskeratosis congenita, autosomal recessive 2, 613987
NHS	100%	100%	Cataract 40, X-linked, 302200 Nance-Horan syndrome, 302350
NIN	99%	99%	?Seckel syndrome 7, 614851
NIPA1	100%	100%	Spastic paraplegia 6, autosomal dominant, 600363
NIPAL4	100%	100%	Ichthyosis, congenital, autosomal recessive 6, 612281
NIPBL	100%	100%	Cornelia de Lange syndrome 1, 122470
NKAP	100%	100%	Intellectual developmental disorder, X-linked, syndromic, Hackman-Di Donato type, 301039
NKX2-1	100%	100%	Chorea, hereditary benign, 118700 Choreoathetosis, hypothyroidism, and neonatal respiratory distress, 610978
NKX2-5	100%	100%	Hypoplastic left heart syndrome 2, 614435 Tetralogy of Fallot, 187500 Hypothyroidism, congenital nongoitrous, 5, 225250 Conotruncal heart malformations, variable, 217095 Ventricular septal defect 3, 614432 Atrial septal defect 7, with or without AV conduction defects, 108900

NKX2-6	100%	100%	Persistent truncus arteriosus, 217095 Conotruncal heart malformations, 217095
NKX3-2	100%	100%	Spondylo-megaepiphyseal-metaphyseal dysplasia, 613330
NKX6-2	100%	100%	Spastic ataxia 8, autosomal recessive, with hypomyelinating leukodystrophy, 617560
NLGN2	100%	100%	No OMIM disease ID
NLGN3	100%	100%	No OMIM disease ID
NLGN4X	100%	100%	Intellectual developmental disorder, X-linked, 300495
NLRC4	100%	100%	?Familial cold autoinflammatory syndrome 4, 616115 Autoinflammation with infantile enterocolitis, 616050
NLRP1	100%	100%	?Respiratory papillomatosis, juvenile recurrent, congenital, 618803 Autoinflammation with arthritis and dyskeratosis, 617388 Palmoplantar carcinoma, multiple self-healing, 615225
NLRP12	100%	100%	Familial cold autoinflammatory syndrome 2, 611762
NLRP3	100%	100%	CINCA syndrome, 607115 Familial cold inflammatory syndrome 1, 120100 Keratoendothelitis fugax hereditaria, 148200 Deafness, autosomal dominant 34, with or without inflammation, 617772 Muckle-Wells syndrome, 191900
NLRP6	100%	100%	No OMIM disease ID
NLRP7	100%	100%	Hydatidiform mole, recurrent, 1, 231090
NME1	100%	100%	No OMIM disease ID
NME3	100%	100%	No OMIM disease ID
NME5	100%	100%	No OMIM disease ID
NME8	100%	100%	Ciliary dyskinesia, primary, 6, 610852

NMNAT1	99%	98%	Spondyloepiphyseal dysplasia, sensorineural hearing loss, intellectual developmental disorder, and Leber congenital amaurosis, 619260 Leber congenital amaurosis 9, 608553
NMNAT2	100%	100%	No OMIM disease ID
NNT	96%	96%	Glucocorticoid deficiency 4, with or without mineralocorticoid deficiency, 614736
NOBOX	100%	100%	Premature ovarian failure 5, 611548
NOD2	100%	100%	Blau syndrome, 186580
NODAL	100%	100%	Heterotaxy, visceral, 5, 270100
NOG	100%	100%	Symphalangism, proximal, 1A, 185800 Brachydactyly, type B2, 611377 Stapes ankylosis with broad thumbs and toes, 184460 Tarsal-carpal coalition syndrome, 186570 Multiple synostoses syndrome 1, 186500
NOL3	100%	100%	?Myoclonus, familial, 1, 614937
NONO	100%	100%	Intellectual developmental disorder, X-linked syndromic 34, 300967
NOP10	100%	100%	Dyskeratosis congenita, autosomal recessive 1, 224230
NOP56	100%	100%	Spinocerebellar ataxia 36, 614153
NOS1AP	100%	100%	Nephrotic syndrome, type 22, 619155
NOS2	100%	100%	No OMIM disease ID
NOTCH1	100%	100%	Adams-Oliver syndrome 5, 616028 Aortic valve disease 1, 109730
NOTCH2	100%	100%	Alagille syndrome 2, 610205 Hajdu-Cheney syndrome, 102500
NOTCH2NLC	100%	100%	Tremor, hereditary essential, 6, 618866 Oculopharyngodistal myopathy 3, 619473 Neuronal intranuclear inclusion disease, 603472

NOTCH3	100%	100%	Lateral meningocele syndrome, 130720 ?Myofibromatosis, infantile 2, 615293 Cerebral arteriopathy with subcortical infarcts and leukoencephalopathy 1, 125310
NOVA2	100%	99%	Neurodevelopmental disorder with or without autistic features and/or structural brain abnormalities, 618859
NPAT	100%	100%	No OMIM disease ID
NPC1	100%	100%	Niemann-Pick disease, type C1, 257220 Niemann-Pick disease, type D, 257220
NPC2	100%	100%	Niemann-pick disease, type C2, 607625
NPHP1	100%	100%	Joubert syndrome 4, 609583 Nephronophthisis 1, juvenile, 256100 Senior-Loken syndrome-1, 266900
NPHP3	100%	100%	Nephronophthisis 3, 604387 Renal-hepatic-pancreatic dysplasia 1, 208540 Meckel syndrome 7, 267010
NPHP4	100%	100%	Senior-Loken syndrome 4, 606996 Nephronophthisis 4, 606966
NPHS1	100%	100%	Nephrotic syndrome, type 1, 256300
NPHS2	100%	100%	Nephrotic syndrome, type 2, 600995
NPL	100%	100%	No OMIM disease ID
NPM1	100%	100%	Leukemia, acute myeloid, somatic, 601626
NPPA	100%	100%	Atrial standstill 2, 615745 Atrial fibrillation, familial, 6, 612201
NPPB	100%	100%	No OMIM disease ID
NPPC	100%	100%	No OMIM disease ID
NPR2	100%	100%	Epiphyseal chondrodysplasia, Miura type, 615923 Short stature with nonspecific skeletal abnormalities, 616255 Acromesomelic dysplasia 1, Maroteaux type, 602875

NPR3	100%	100%	Boudin-Mortier syndrome, 619543
NPRL2	100%	100%	Epilepsy, familial focal, with variable foci 2, 617116
NPRL3	100%	100%	Epilepsy, familial focal, with variable foci 3, 617118
NR0B1	100%	100%	Adrenal hypoplasia, congenital, 300200 46XY sex reversal 2, dosage-sensitive, 300018
NR0B2	100%	100%	Obesity, mild, early-onset, 601665
NR1H4	100%	100%	Cholestasis, progressive familial intrahepatic, 5, 617049
NR2E3	100%	100%	Retinitis pigmentosa 37, 611131 Enhanced S-cone syndrome, 268100
NR2F1	100%	99%	Bosch-Boonstra-Schaaf optic atrophy syndrome, 615722
NR2F2	100%	100%	46,XX sex reversal 5, 618901 Congenital heart defects, multiple types, 4, 615779
NR3C1	100%	100%	Glucocorticoid resistance, 615962
NR3C2	100%	100%	Pseudohypoaldosteronism type I, autosomal dominant, 177735 Hypertension, early-onset, autosomal dominant, with exacerbation in pregnancy, 605115
NR4A2	100%	100%	No OMIM disease ID
NR4A3	100%	100%	Chondrosarcoma, extraskeletal myxoid, 612237
NR5A1	100%	100%	46, XX sex reversal 4, 617480 Premature ovarian failure 7, 612964 46XY sex reversal 3, 612965 Adrenocortical insufficiency, 612964 Spermatogenic failure 8, 613957

NRAS	100%	100%	Noonan syndrome 6, 613224 ?RAS-associated autoimmune lymphoproliferative syndrome type IV, somatic, 614470 Melanocytic nevus syndrome, congenital, somatic, 137550 Epidermal nevus, somatic, 162900 Schimmelpenning-Feuerstein-Mims syndrome, somatic mosaic, 163200 Thyroid carcinoma, follicular, somatic, 188470 Neurocutaneous melanosis, somatic, 249400 Colorectal cancer, somatic, 114500
NRIP1	100%	100%	?Congenital anomalies of kidney and urinary tract 3, 618270
NRL	100%	100%	Retinitis pigmentosa 27, 613750 Retinal degeneration, autosomal recessive, clumped pigment type,
NRROS	100%	100%	Seizures, early-onset, with neurodegeneration and brain calcification, 618875
NRXN1	100%	100%	Pitt-Hopkins-like syndrome 2, 614325
NSD1	100%	100%	Sotos syndrome 1, 117550
NSD2	100%	100%	No OMIM disease ID
NSDHL	100%	100%	CK syndrome, 300831 CHILD syndrome, 308050
NSF	100%	100%	Developmental and epileptic encephalopathy 96, 619340
NSMCE2	100%	100%	Seckel syndrome 10, 617253
NSMCE3	100%	100%	Lung disease, immunodeficiency, and chromosome breakage syndrome, 617241
NSMF	100%	100%	Hypogonadotropic hypogonadism 9 with or without anosmia, 614838
NSUN2	100%	100%	Mental retardation, autosomal recessive 5, 611091
NSUN3	100%	100%	Combined oxidative phosphorylation deficiency 48, 619012
NT5C2	100%	100%	Spastic paraplegia 45, autosomal recessive, 613162
NT5C3A	100%	100%	Anemia, hemolytic, due to UMPH1 deficiency, 266120
NT5E	100%	100%	Calcification of joints and arteries, 211800
NTF4	100%	100%	Glaucoma 1, open angle, 1O, 613100
NTHL1	100%	100%	Familial adenomatous polyposis 3, 616415

NTN1	100%	100%	Mirror movements 4, 618264
NTNG2	100%	100%	Neurodevelopmental disorder with behavioral abnormalities, absent speech, and hypotonia, 618718
NTRK1	100%	100%	Insensitivity to pain, congenital, with anhidrosis, 256800
NTRK2	100%	100%	Developmental and epileptic encephalopathy 58, 617830 Obesity, hyperphagia, and developmental delay, 613886
NUAK2	100%	100%	?Anencephaly 2, 619452
NUBPL	100%	100%	Mitochondrial complex I deficiency, nuclear type 21, 618242
NUDT2	100%	100%	No OMIM disease ID
NUMA1	100%	100%	Leukemia, acute promyelocytic, somatic, 612376
NUP107	100%	100%	?Ovarian dysgenesis 6, 618078 Galloway-Mowat syndrome 7, 618348 Nephrotic syndrome, type 11, 616730
NUP133	100%	100%	?Galloway-Mowat syndrome 8, 618349 Nephrotic syndrome, type 18, 618177
NUP155	100%	100%	?Atrial fibrillation 15, 615770
NUP160	100%	100%	?Nephrotic syndrome, type 19, 618178
NUP188	100%	100%	Sandestig-Stefanova syndrome, 618804
NUP205	100%	100%	?Nephrotic syndrome, type 13, 616893
NUP214	100%	100%	Leukemia, T-cell acute lymphoblastic, somatic, 613065 Leukemia, acute myeloid, somatic, 601626
NUP37	100%	100%	?Microcephaly 24, primary, autosomal recessive, 618179
NUP62	100%	100%	Striatonigral degeneration, infantile, 271930
NUP85	100%	100%	Nephrotic syndrome, type 17, 618176
NUP88	100%	100%	Fetal akinesia deformation sequence 4, 618393
NUP93	95%	95%	Nephrotic syndrome, type 12, 616892
NUS1	100%	100%	Mental retardation, autosomal dominant 55, with seizures, 617831 ?Congenital disorder of glycosylation, type 1aa, 617082

NUTM2B-AS1	NC	NC	?Oculopharyngeal myopathy with leukoencephalopathy 1, 618637
NXF5	100%	100%	No OMIM disease ID
NXN	100%	100%	Robinow syndrome, autosomal recessive 2, 618529
NYX	100%	100%	Night blindness, congenital stationary (complete), 1A, X-linked, 310500
OAS1	100%	100%	No OMIM disease ID
OAT	100%	100%	Gyrate atrophy of choroid and retina with or without ornithinemia, 258870
OBSCN	100%	100%	No OMIM disease ID
OBSL1	100%	100%	3-M syndrome 2, 612921
OCA2	100%	100%	Albinism, brown oculocutaneous, 203200 Albinism, oculocutaneous, type II, 203200
OCLN	100%	100%	Pseudo-TORCH syndrome 1, 251290
OCRL	100%	100%	Dent disease 2, 300555 Lowe syndrome, 309000
ODAD1	100%	100%	Ciliary dyskinesia, primary, 20, 615067
ODAD2	96%	96%	Ciliary dyskinesia, primary, 23, 615451
ODAD3	100%	100%	Ciliary dyskinesia, primary, 30, 616037
ODAD4	100%	100%	Ciliary dyskinesia, primary, 35, 617092
ODAM	100%	100%	No OMIM disease ID
ODAPH	100%	100%	Amelogenesis imperfecta, type IIA4, 614832
ODC1	100%	100%	Bachmann-Bupp syndrome, 619075
OFD1	100%	100%	Simpson-Golabi-Behmel syndrome, type 2, 300209 ?Retinitis pigmentosa 23, 300424 Orofaciodigital syndrome I, 311200 Joubert syndrome 10, 300804
OGDH	100%	100%	No OMIM disease ID
OGDHL	100%	100%	No OMIM disease ID
OGG1	100%	100%	Renal cell carcinoma, clear cell, somatic, 144700
OGT	100%	100%	Intellectual developmental disorder, X-linked 106, 300997

OPA1	100%	100%	Optic atrophy plus syndrome, 125250 Optic atrophy 1, 165500 Behr syndrome, 210000 ?Mitochondrial DNA depletion syndrome 14 (encephalocardiomyopathic type), 616896
OPA3	100%	100%	3-methylglutaconic aciduria, type III, 258501 Optic atrophy 3 with cataract, 165300
OPCML	100%	100%	Ovarian cancer, somatic, 167000
OPHN1	100%	99%	Intellectual developmental disorder, X-linked syndromic, Billuart type, 300486
OPLAH	100%	100%	5-oxoprolinase deficiency, 260005
OPN1LW	98%	98%	Blue cone monochromacy, 303700 Colorblindness, protan, 303900
OPN1MW	97%	96%	Colorblindness, deutan, 303800 Blue cone monochromacy, 303700
OPN1SW	100%	100%	Colorblindness, tritan, 190900
OPTN	100%	100%	Glaucoma 1, open angle, E, 137760 Amyotrophic lateral sclerosis 12 with or without frontotemporal dementia, 613435
ORAI1	100%	99%	Immunodeficiency 9, 612782 Myopathy, tubular aggregate, 2, 615883
ORC1	100%	100%	Meier-Gorlin syndrome 1, 224690
ORC4	100%	100%	Meier-Gorlin syndrome 2, 613800
ORC6	100%	100%	Meier-Gorlin syndrome 3, 613803
OSBPL2	100%	100%	Deafness, autosomal dominant 67, 616340
OSGEP	100%	100%	Galloway-Mowat syndrome 3, 617729
OSMR	100%	100%	Amyloidosis, primary localized cutaneous, 1, 105250
OSTM1	100%	100%	Osteopetrosis, autosomal recessive 5, 259720
OTC	100%	100%	Ornithine transcarbamylase deficiency, 311250
OTOA	100%	100%	Deafness, autosomal recessive 22, 607039

OTOF	100%	100%	Auditory neuropathy, autosomal recessive, 1, 601071 Deafness, autosomal recessive 9, 601071
OTOG	100%	100%	Deafness, autosomal recessive 18B, 614945
OTOGL	100%	100%	Deafness, autosomal recessive 84B, 614944
OTUD5	100%	99%	Multiple congenital anomalies-neurodevelopmental syndrome, X-linked, 301056
OTUD6B	100%	100%	Intellectual developmental disorder with dysmorphic facies, seizures, and distal limb anomalies, 617452
OTUD7A	99%	97%	No OMIM disease ID
OTULIN	100%	99%	Autoinflammation, panniculitis, and dermatosis syndrome, 617099
OTX2	100%	100%	Retinal dystrophy, early-onset, with or without pituitary dysfunction, 610125 Pituitary hormone deficiency, combined, 6, 613986 Microphthalmia, syndromic 5, 610125
OVOL2	100%	100%	Corneal dystrophy, posterior polymorphous, 1, 122000
OXA1L	100%	100%	No OMIM disease ID
OXCT1	100%	100%	Succinyl CoA:3-oxoacid CoA transferase deficiency, 245050
OXR1	100%	100%	Cerebellar hypoplasia/atrophy, epilepsy, and global developmental delay, 213000
P2RX2	100%	100%	Deafness, autosomal dominant 41, 608224
P2RY12	100%	100%	Bleeding disorder, platelet-type, 8, 609821
P3H1	100%	100%	Osteogenesis imperfecta, type VIII, 610915
P3H2	100%	100%	Myopia, high, with cataract and vitreoretinal degeneration, 614292
P4HA2	100%	100%	Myopia 25, autosomal dominant, 617238
P4HB	100%	100%	Cole-Carpenter syndrome 1, 112240
P4HTM	100%	100%	Hypotonia, hypoventilation, impaired intellectual development, dysautonomia, epilepsy, and eye abnormalities, 618493
PABPN1	100%	100%	Oculopharyngeal muscular dystrophy, 164300
PACS1	100%	100%	Schuurs-Hoeijmakers syndrome, 615009
PACS2	100%	100%	Developmental and epileptic encephalopathy 66, 618067
PADI3	100%	100%	Uncombable hair syndrome, 191480
PADI6	100%	100%	Preimplantation embryonic lethality 2, 617234
PAFAH1B1	100%	100%	Subcortical laminar heterotopia, 607432 Lissencephaly 1, 607432

PAH	100%	100%	Phenylketonuria, 261600
PAK1	100%	100%	Intellectual developmental disorder with macrocephaly, seizures, and speech delay, 618158
PAK3	100%	99%	Intellectual developmental disorder, X-linked 30, 300558
PALB2	100%	100%	Fanconi anemia, complementation group N, 610832
PALS1	100%	100%	No OMIM disease ID
PAM16	82%	82%	Spondylometaphyseal dysplasia, Megarbane-Dagher-Melike type, 613320
PANK2	100%	100%	HARP syndrome, 607236 Neurodegeneration with brain iron accumulation 1, 234200
PANK4	100%	100%	?Cataract 49, 619593
PANX1	100%	100%	Oocyte maturation defect 7, 618550
PAPPA2	100%	100%	Short stature, Dauber-Argente type, 619489
PAPSS2	100%	100%	Brachyolmia 4 with mild epiphyseal and metaphyseal changes, 612847
PARK7	100%	100%	Parkinson disease 7, autosomal recessive early-onset, 606324
PARN	90%	87%	Dyskeratosis congenita, autosomal recessive 6, 616353 Pulmonary fibrosis and/or bone marrow failure, telomere-related, 4, 616371
PARP1	100%	100%	No OMIM disease ID
PARP4	100%	100%	No OMIM disease ID
PARP6	100%	100%	No OMIM disease ID
PARS2	100%	100%	Developmental and epileptic encephalopathy 75, 618437
PATL2	100%	100%	Oocyte maturation defect 4, 617743
PAX1	100%	100%	Otofaciocervical syndrome 2, 615560
PAX2	100%	100%	Glomerulosclerosis, focal segmental, 7, 616002 Papillorenal syndrome, 120330

PAX3	100%	100%	Craniofacial-deafness-hand syndrome, 122880 Waardenburg syndrome, type 3, 148820 Waardenburg syndrome, type 1, 193500 Rhabdomyosarcoma 2, alveolar, 268220
PAX4	100%	100%	Maturity-onset diabetes of the young, type IX, 612225 Diabetes mellitus, type 2, 125853
PAX5	100%	100%	No OMIM disease ID
PAX6	100%	100%	Optic nerve hypoplasia, 165550 Cataract with late-onset corneal dystrophy, 106210 ?Coloboma, ocular, 120200 ?Coloboma of optic nerve, 120430 Aniridia, 106210 Anterior segment dysgenesis 5, multiple subtypes, 604229 ?Morning glory disc anomaly, 120430 Foveal hypoplasia 1, 136520 Keratitis, 148190
PAX7	100%	100%	Rhabdomyosarcoma 2, alveolar, 268220 Myopathy, congenital, progressive, with scoliosis, 618578
PAX8	100%	100%	Hypothyroidism, congenital, due to thyroid dysgenesis or hypoplasia, 218700
PAX9	100%	100%	Tooth agenesis, selective, 3, 604625
PBRM1	100%	100%	?Renal cell carcinoma, clear cell, 144700
PBX1	100%	100%	Congenital anomalies of kidney and urinary tract syndrome with or without hearing loss, abnormal ears, or developmental delay, 617641
PC	100%	100%	Pyruvate carboxylase deficiency, 266150
PCARE	100%	100%	Retinitis pigmentosa 54, 613428
PCBD1	100%	100%	Hyperphenylalaninemia, BH4-deficient, D, 264070

PCCA	100%	100%	Propionicacidemia, 606054
PCCB	99%	98%	Propionicacidemia, 606054
PCDH12	100%	100%	Diencephalic-mesencephalic junction dysplasia syndrome 1, 251280
PCDH15	100%	100%	Usher syndrome, type 1D/F digenic, 601067 Deafness, autosomal recessive 23, 609533 Usher syndrome, type 1F, 602083
PCDH19	100%	100%	Developmental and epileptic encephalopathy 9, 300088
PCDHGC4	100%	100%	No OMIM disease ID
PCGF2	100%	100%	Turnpenny-Fry syndrome, 618371
PCK1	100%	100%	?Phosphoenolpyruvate carboxykinase deficiency, cytosolic, 261680
PCK2	100%	100%	No OMIM disease ID
PCLO	100%	100%	?Pontocerebellar hypoplasia, type 3, 608027
PCNA	100%	100%	?Ataxia-telangiectasia-like disorder 2, 615919
PCNT	100%	100%	Microcephalic osteodysplastic primordial dwarfism, type II, 210720
PCSK1	100%	100%	Obesity with impaired prohormone processing, 600955
PCSK9	100%	100%	Hypercholesterolemia, familial, 3, 603776
PCYT1A	100%	100%	Spondylometaphyseal dysplasia with cone-rod dystrophy, 608940
PCYT2	100%	99%	Spastic paraplegia 82, autosomal recessive, 618770
PDCD1	100%	100%	No OMIM disease ID
PDCD10	100%	100%	Cerebral cavernous malformations-3, 603285
PDE10A	89%	87%	Striatal degeneration, autosomal dominant, 616922 Dyskinesia, limb and orofacial, infantile-onset, 616921
PDE11A	100%	100%	Pigmented nodular adrenocortical disease, primary, 2, 610475
PDE1C	100%	100%	?Deafness, autosomal dominant 74, 618140
PDE2A	100%	100%	Intellectual developmental disorder with paroxysmal dyskinesia or seizures, 619150
PDE3A	100%	100%	Hypertension and brachydactyly syndrome, 112410
PDE4D	100%	100%	Acrodysostosis 2, with or without hormone resistance, 614613
PDE6A	100%	100%	Retinitis pigmentosa 43, 613810
PDE6B	100%	100%	Retinitis pigmentosa-40, 613801 Night blindness, congenital stationary, autosomal dominant 2, 163500

PDE6C	100%	100%	Cone dystrophy 4, 613093
PDE6D	100%	100%	Joubert syndrome 22, 615665
PDE6G	100%	100%	Retinitis pigmentosa 57, 613582
PDE6H	100%	100%	Retinal cone dystrophy 3, 610024 Achromatopsia 6, 610024
PDE8B	100%	100%	Pigmented nodular adrenocortical disease, primary, 3, 614190 Striatal degeneration, autosomal dominant, 609161
PDGFB	100%	100%	Meningioma, SIS-related, 607174 Basal ganglia calcification, idiopathic, 5, 615483 Dermatofibrosarcoma protuberans, 607907
PDGFRA	100%	100%	Gastrointestinal stromal tumor/GIST-plus syndrome, somatic or familial, 175510 Hypereosinophilic syndrome, idiopathic, resistant to imatinib, 607685
PDGFRB	100%	100%	Premature aging syndrome, Penttinen type, 601812 Kosaki overgrowth syndrome, 616592 Myofibromatosis, infantile, 1, 228550 Basal ganglia calcification, idiopathic, 4, 615007
PDGFRL	100%	100%	Hepatocellular cancer, somatic, 114550 Colorectal cancer, somatic, 114500
PDHA1	100%	100%	Pyruvate dehydrogenase E1-alpha deficiency, 312170
PDHB	100%	100%	Pyruvate dehydrogenase E1-beta deficiency, 614111
PDHX	100%	100%	Lacticacidemia due to PDX1 deficiency, 245349
PDK1	100%	100%	No OMIM disease ID
PDK2	100%	100%	No OMIM disease ID
PDK3	100%	100%	?Charcot-Marie-Tooth disease, X-linked dominant, 6, 300905

PDK4	100%	100%	No OMIM disease ID
PDLIM3	100%	100%	No OMIM disease ID
PDLIM5	99%	96%	No OMIM disease ID
PDP1	100%	100%	Pyruvate dehydrogenase phosphatase deficiency, 608782
PDSS1	97%	97%	Coenzyme Q10 deficiency, primary, 2, 614651
PDSS2	100%	100%	Coenzyme Q10 deficiency, primary, 3, 614652
PDX1	100%	100%	Pancreatic agenesis 1, 260370 MODY, type IV, 606392
PDXK	100%	99%	Neuropathy, hereditary motor and sensory, type VIC, with optic atrophy, 618511
PDYN	100%	100%	Spinocerebellar ataxia 23, 610245
PDZD7	100%	99%	Deafness, autosomal recessive 57, 618003 Usher syndrome, type IIC, GPR98/PDZD7 digenic, 605472
PEPD	100%	100%	Prolidase deficiency, 170100
PER2	100%	100%	?Advanced sleep phase syndrome, familial, 1, 604348
PER3	100%	100%	?Advanced sleep phase syndrome, familial, 3, 616882
PERCC1	100%	100%	Diarrhea 11, malabsorptive, congenital, 618662
PERP	100%	100%	Erythrokeratodermia variabilis et progressiva 7, 619209 Olmsted syndrome 2, 619208
PET100	100%	100%	Mitochondrial complex IV deficiency, nuclear type 12, 619055
PET117	100%	100%	?Mitochondrial complex IV deficiency, nuclear type 19, 619063
PEX1	100%	100%	Heimler syndrome 1, 234580 Peroxisome biogenesis disorder 1B (NALD/IRD), 601539 Peroxisome biogenesis disorder 1A (Zellweger), 214100
PEX10	100%	100%	Peroxisome biogenesis disorder 6A (Zellweger), 614870 Peroxisome biogenesis disorder 6B, 614871
PEX11B	100%	100%	Peroxisome biogenesis disorder 14B, 614920

PEX12	100%	100%	Peroxisome biogenesis disorder 3B, 266510 Peroxisome biogenesis disorder 3A (Zellweger), 614859
PEX13	100%	100%	Peroxisome biogenesis disorder 11A (Zellweger), 614883 Peroxisome biogenesis disorder 11B, 614885
PEX14	100%	100%	Peroxisome biogenesis disorder 13A (Zellweger), 614887
PEX16	100%	100%	Peroxisome biogenesis disorder 8B, 614877 Peroxisome biogenesis disorder 8A (Zellweger), 614876
PEX19	100%	100%	Peroxisome biogenesis disorder 12A (Zellweger), 614886
PEX2	100%	100%	Peroxisome biogenesis disorder 5A (Zellweger), 614866 Peroxisome biogenesis disorder 5B, 614867
PEX26	100%	100%	Peroxisome biogenesis disorder 7B, 614873 Peroxisome biogenesis disorder 7A (Zellweger), 614872
PEX3	100%	100%	Peroxisome biogenesis disorder 10A (Zellweger), 614882 ?Peroxisome biogenesis disorder 10B, 617370
PEX5	100%	100%	Peroxisome biogenesis disorder 2B, 202370 Peroxisome biogenesis disorder 2A (Zellweger), 214110 Rhizomelic chondrodysplasia punctata, type 5, 616716

PEX6	100%	100%	Peroxisome biogenesis disorder 4B, 614863 Peroxisome biogenesis disorder 4A (Zellweger), 614862 Heimler syndrome 2, 616617
PEX7	91%	91%	Rhizomelic chondrodysplasia punctata, type 1, 215100 Peroxisome biogenesis disorder 9B, 614879
PFKM	100%	100%	Glycogen storage disease VII, 232800
PFN1	100%	100%	Amyotrophic lateral sclerosis 18, 614808
PGAM2	100%	100%	Glycogen storage disease X, 261670
PGAP1	100%	100%	Neurodevelopmental disorder with dysmorphic features, spasticity, and brain abnormalities, 615802
PGAP2	100%	100%	Hyperphosphatasia with mental retardation syndrome 3, 614207
PGAP3	100%	100%	Hyperphosphatasia with mental retardation syndrome 4, 615716
PGK1	100%	100%	Phosphoglycerate kinase 1 deficiency, 300653
PGM1	94%	94%	Congenital disorder of glycosylation, type It, 614921
PGM2L1	100%	100%	No OMIM disease ID
PGM3	91%	91%	Immunodeficiency 23, 615816
PHACTR1	100%	100%	Developmental and epileptic encephalopathy 70, 618298
PHC1	100%	100%	?Microcephaly 11, primary, autosomal recessive, 615414
PHEX	100%	100%	Hypophosphatemic rickets, X-linked dominant, 307800
PHF21A	100%	100%	Intellectual developmental disorder with behavioral abnormalities and craniofacial dysmorphism with or without seizures, 618725
PHF6	100%	99%	Borjeson-Forssman-Lehmann syndrome, 301900
PHF8	100%	100%	Intellectual developmental disorder, X-linked, syndromic, Siderius type, 300263
PHGDH	100%	100%	Neu-Laxova syndrome 1, 256520 Phosphoglycerate dehydrogenase deficiency, 601815
PHIP	100%	99%	Chung-Jansen syndrome, 617991
PHKA1	100%	99%	Muscle glycogenosis, 300559
PHKA2	100%	99%	Glycogen storage disease, type IXa2, 306000 Glycogen storage disease, type IXa1, 306000
PHKB	100%	100%	Phosphorylase kinase deficiency of liver and muscle, autosomal recessive, 261750

PHKG1	100%	100%	No OMIM disease ID
PHKG2	100%	100%	Glycogen storage disease IXc, 613027
PHOX2A	100%	100%	Fibrosis of extraocular muscles, congenital, 2, 602078
PHOX2B	100%	100%	Neuroblastoma with Hirschsprung disease, 613013 Central hypoventilation syndrome, congenital, 1, with or without Hirschsprung disease, 209880
PHYH	100%	100%	Refsum disease, 266500
PI4K2A	100%	100%	No OMIM disease ID
PI4KA	100%	100%	Spastic paraplegia 84, autosomal recessive, 619621 Polymicrogyria, perisylvian, with cerebellar hypoplasia and arthrogryposis, 616531
PI4KB	100%	100%	No OMIM disease ID
PIBF1	100%	100%	Joubert syndrome 33, 617767
PICALM	100%	100%	Leukemia, acute myeloid, somatic, 601626
PIDD1	100%	100%	No OMIM disease ID
PIEZO1	100%	100%	Lymphatic malformation 6, 616843 Dehydrated hereditary stomatocytosis with or without pseudohyperkalemia and/or perinatal edema, 194380
PIEZO2	100%	100%	Arthrogryposis, distal, type 5, 108145 Arthrogryposis, distal, with impaired proprioception and touch, 617146 Arthrogryposis, distal, type 3, 114300 ?Marden-Walker syndrome, 248700
PIGA	100%	100%	Paroxysmal nocturnal hemoglobinuria, somatic, 300818 Multiple congenital anomalies-hypotonia-seizures syndrome 2, 300868
PIGB	100%	100%	Developmental and epileptic encephalopathy 80, 618580

PIGC	100%	100%	Glycosylphosphatidylinositol biosynthesis defect 16, 617816
PIGF	100%	100%	Onychodystrophy, osteodystrophy, impaired intellectual development, and seizures syndrome, 619356
PIGG	100%	100%	Mental retardation, autosomal recessive 53, 616917
PIGH	81%	75%	Glycosylphosphatidylinositol biosynthesis defect 17, 618010
PIGK	100%	100%	Neurodevelopmental disorder with hypotonia and cerebellar atrophy, with or without seizures, 618879
PIGL	100%	100%	CHIME syndrome, 280000
PIGM	100%	100%	Glycosylphosphatidylinositol deficiency, 610293
PIGN	98%	98%	Multiple congenital anomalies-hypotonia-seizures syndrome 1, 614080
PIGO	100%	100%	Hyperphosphatasia with mental retardation syndrome 2, 614749
PIGP	100%	100%	Developmental and epileptic encephalopathy 55, 617599
PIGQ	100%	100%	Developmental and epileptic encephalopathy 77, 618548
PIGS	100%	100%	Developmental and epileptic encephalopathy 95, 618143
PIGT	100%	100%	?Paroxysmal nocturnal hemoglobinuria 2, 615399 Multiple congenital anomalies-hypotonia-seizures syndrome 3, 615398
PIGU	100%	99%	Neurodevelopmental disorder with brain anomalies, seizures, and scoliosis, 618590
PIGV	100%	100%	Hyperphosphatasia with mental retardation syndrome 1, 239300
PIGW	100%	100%	Glycosylphosphatidylinositol biosynthesis defect 11, 616025
PIGY	100%	100%	Hyperphosphatasia with mental retardation syndrome 6, 616809
PIK3C2A	100%	100%	Oculoskeletodental syndrome, 618440

PIK3CA	100%	100%	CLOVE syndrome, somatic, 612918 Hepatocellular carcinoma, somatic, 114550 Breast cancer, somatic, 114480 Ovarian cancer, somatic, 167000 Colorectal cancer, somatic, 114500 CLAPO syndrome, somatic, 613089 Keratosis, seborrheic, somatic, 182000 Nevus, epidermal, somatic, 162900 Gastric cancer, somatic, 613659 Nonsmall cell lung cancer, somatic, 211980 Megalencephaly-capillary malformation-polymicrogyria syndrome, somatic, 602501 Cowden syndrome 5, 615108 Macrodactyly, somatic,,
PIK3CD	100%	100%	Immunodeficiency 14A, autosomal dominant, 615513 Immunodeficiency 14B, autosomal recessive, 619281 ?Roifman-Chitayat syndrome, digenic, 613328
PIK3CG	100%	100%	No OMIM disease ID
PIK3R1	100%	100%	Immunodeficiency 36, 616005 ?Agammaglobulinemia 7, autosomal recessive, 615214 SHORT syndrome, 269880
PIK3R2	100%	100%	Megalencephaly-polymicrogyria-polydactyly-hydrocephalus syndrome 1, 603387
PIK3R5	100%	100%	Ataxia-oculomotor apraxia 3, 615217
PIKFYVE	100%	100%	Corneal fleck dystrophy, 121850
PINK1	100%	100%	Parkinson disease 6, early onset, 605909
PIP5K1C	100%	100%	Lethal congenital contractural syndrome 3, 611369
PISD	100%	100%	Liberfarb syndrome, 618889
PITPNM3	100%	100%	Cone-rod dystrophy 5, 600977

PITRM1	100%	100%	Spinocerebellar ataxia, autosomal recessive 30, 619405
PITX1	100%	100%	Clubfoot, congenital, with or without deficiency of long bones and/or mirror-image polydactyly, 119800
PITX2	100%	100%	Ring dermoid of cornea, 180550 Axenfeld-Rieger syndrome, type 1, 180500 Anterior segment dysgenesis 4, 137600
PITX3	100%	100%	Cataract 11, multiple types, 610623 Anterior segment dysgenesis 1, multiple subtypes, 107250 Cataract 11, syndromic, autosomal recessive, 610623
PIWIL2	100%	100%	No OMIM disease ID
PJA1	100%	100%	No OMIM disease ID
PJVK	100%	100%	Deafness, autosomal recessive 59, 610220
PKD1	99%	99%	Polycystic kidney disease 1, 173900
PKD1L1	100%	100%	Heterotaxy, visceral, 8, autosomal, 617205
PKD2	100%	100%	Polycystic kidney disease 2, 613095
PKDCC	100%	99%	Rhizomelic limb shortening with dysmorphic features, 618821
PKHD1	100%	100%	Polycystic kidney disease 4, with or without hepatic disease, 263200
PKLR	100%	100%	Adenosine triphosphate, elevated, of erythrocytes, 102900 Pyruvate kinase deficiency, 266200
PKP1	100%	100%	Ectodermal dysplasia/skin fragility syndrome, 604536
PKP2	95%	95%	Arrhythmogenic right ventricular dysplasia 9, 609040
PKP4	100%	100%	No OMIM disease ID
PLA2G4A	100%	100%	Gastrointestinal ulceration, recurrent, with dysfunctional platelets, 618372
PLA2G5	100%	100%	No OMIM disease ID
PLA2G6	92%	92%	Parkinson disease 14, autosomal recessive, 612953 Neurodegeneration with brain iron accumulation 2B, 610217 Infantile neuroaxonal dystrophy 1, 256600
PLA2G7	100%	100%	Platelet-activating factor acetylhydrolase deficiency, 614278

PLAA	100%	100%	Neurodevelopmental disorder with progressive microcephaly, spasticity, and brain anomalies, 617527
PLAG1	100%	100%	Adenomas, salivary gland pleomorphic, somatic, 181030 Silver-Russell syndrome 4, 618907
PLAT	100%	100%	No OMIM disease ID
PLAU	100%	100%	Quebec platelet disorder, 601709
PLCB1	100%	100%	Developmental and epileptic encephalopathy 12, 613722
PLCB3	100%	100%	Spondylometaphyseal dysplasia with corneal dystrophy, 618961
PLCB4	100%	100%	Auriculocondylar syndrome 2, 614669
PLCD1	100%	100%	Nail disorder, nonsyndromic congenital, 3, (leukonychia), 151600
PLCE1	100%	100%	Nephrotic syndrome, type 3, 610725
PLCG2	100%	100%	Autoinflammation, antibody deficiency, and immune dysregulation syndrome, 614878 Familial cold autoinflammatory syndrome 3, 614468
PLCZ1	100%	100%	Spermatogenic failure 17, 617214
PLD1	100%	100%	Cardiac valvular defect, developmental, 212093
PLD3	100%	100%	?Spinocerebellar ataxia 46, 617770
PLEC	100%	100%	?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%	Leukodystrophy and acquired microcephaly with or without dystonia, 616763
PLEKHG5	96%	96%	Spinal muscular atrophy, distal, autosomal recessive, 4, 611067 Charcot-Marie-Tooth disease, recessive intermediate C, 615376

PLEKHM1	100%	100%	?Osteopetrosis, autosomal recessive 6, 611497 Osteopetrosis, autosomal dominant 3, 618107
PLEKHM2	100%	100%	No OMIM disease ID
PLG	100%	100%	Dysplasminogenemia, 217090 Angioedema, hereditary, 4, 619360 Plasminogen deficiency, type I, 217090
PLIN1	100%	100%	Lipodystrophy, familial partial, type 4, 613877
PLK1	100%	100%	No OMIM disease ID
PLK4	100%	100%	Microcephaly and chorioretinopathy, autosomal recessive, 2, 616171
PLN	100%	100%	Cardiomyopathy, dilated, 1P, 609909 Cardiomyopathy, hypertrophic, 18, 613874
PLOD1	100%	100%	Ehlers-Danlos syndrome, kyphoscoliotic type, 1, 225400
PLOD2	100%	100%	Bruck syndrome 2, 609220
PLOD3	100%	100%	Lysyl hydroxylase 3 deficiency, 612394
PLP1	100%	100%	Pelizaeus-Merzbacher disease, 312080 Spastic paraplegia 2, X-linked, 312920
PLPBP	100%	100%	Epilepsy, early-onset, vitamin B6-dependent, 617290
PLPP6	100%	100%	No OMIM disease ID
PLS1	100%	100%	Deafness, autosomal dominant 76, 618787
PLS3	97%	97%	Bone mineral density QTL18, osteoporosis, 300910
PLVAP	100%	100%	Diarrhea 10, protein-losing enteropathy type, 618183
PLXNA1	100%	100%	No OMIM disease ID
PLXNA2	100%	100%	No OMIM disease ID
PLXND1	100%	99%	No OMIM disease ID
PMEPA1	100%	99%	No OMIM disease ID
PMFBP1	100%	100%	Spermatogenic failure 31, 618112
PML	100%	100%	Leukemia, acute promyelocytic, PML/RARA type,
PMM2	100%	100%	Congenital disorder of glycosylation, type Ia, 212065
PMP2	100%	100%	Charcot-Marie-Tooth disease, demyelinating, type 1G, 618279

PMP22	100%	100%	Charcot-Marie-Tooth disease, type 1A, 118220 Roussy-Levy syndrome, 180800 Charcot-Marie-Tooth disease, type 1E, 118300 ?Neuropathy, inflammatory demyelinating, 139393 Neuropathy, recurrent, with pressure palsies, 162500 Dejerine-Sottas disease, 145900
PMPCA	100%	100%	Spinocerebellar ataxia, autosomal recessive 2, 213200
PMPCB	100%	100%	Multiple mitochondrial dysfunctions syndrome 6, 617954
PMS2	100%	100%	Colorectal cancer, hereditary nonpolyposis, type 4, 614337 Mismatch repair cancer syndrome 4, 619101
PMS2CL	NC	NC	No OMIM disease ID
PMVK	100%	100%	Porokeratosis 1, multiple types, 175800
PNKD	100%	100%	Paroxysmal nonkinesigenic dyskinesia 1, 118800
PNKP	100%	100%	?Charcot-Marie-Tooth disease, type 2B2, 605589 Ataxia-oculomotor apraxia 4, 616267 Microcephaly, seizures, and developmental delay, 613402
PNLDC1	100%	100%	Spermatogenic failure 57, 619528
PNLIP	100%	100%	?Pancreatic lipase deficiency, 614338
PNMT	100%	100%	No OMIM disease ID
PNP	100%	100%	Immunodeficiency due to purine nucleoside phosphorylase deficiency, 613179
PNPLA1	100%	100%	Ichthyosis, congenital, autosomal recessive 10, 615024
PNPLA2	100%	100%	Neutral lipid storage disease with myopathy, 610717

PNPLA6	100%	100%	Spastic paraplegia 39, autosomal recessive, 612020 Oliver-McFarlane syndrome, 275400 ?Laurence-Moon syndrome, 245800 Boucher-Neuhauser syndrome, 215470
PNPLA8	100%	100%	?Mitochondrial myopathy with lactic acidosis, 251950
PNPO	100%	100%	Pyridoxamine 5'-phosphate oxidase deficiency, 610090
PNPT1	100%	100%	Deafness, autosomal recessive 70, 614934 Combined oxidative phosphorylation deficiency 13, 614932
POC1A	100%	100%	Short stature, onychodysplasia, facial dysmorphism, and hypotrichosis, 614813
POC1B	100%	100%	Cone-rod dystrophy 20, 615973
POC5	100%	100%	No OMIM disease ID
PODXL	94%	94%	No OMIM disease ID
POF1B	100%	100%	?Premature ovarian failure 2B, 300604
POFUT1	100%	100%	Dowling-Degos disease 2, 615327
POGLUT1	100%	100%	Dowling-Degos disease 4, 615696 ?Muscular dystrophy, limb-girdle, autosomal recessive 21, 617232
POGZ	100%	100%	White-Sutton syndrome, 616364
POLA1	100%	100%	Pigmentary disorder, reticulate, with systemic manifestations, X-linked, 301220 Van Esch-O'Driscoll syndrome, 301030
POLD1	100%	100%	Mandibular hypoplasia, deafness, progeroid features, and lipodystrophy syndrome, 615381
POLE	100%	100%	FILS syndrome, 615139 IMAGE-I syndrome, 618336
POLE2	100%	100%	No OMIM disease ID

POLG	100%	100%	Mitochondrial recessive ataxia syndrome (includes SANDO and SCAE), 607459 Mitochondrial DNA depletion syndrome 4B (MNGIE type), 613662 Mitochondrial DNA depletion syndrome 4A (Alpers type), 203700 Progressive external ophthalmoplegia, autosomal dominant 1, 157640 Progressive external ophthalmoplegia, autosomal recessive 1, 258450
POLG2	100%	100%	Progressive external ophthalmoplegia with mitochondrial DNA deletions, autosomal dominant 4, 610131 ?Mitochondrial DNA depletion syndrome 16 (hepatic type), 618528 ?Mitochondrial DNA depletion syndrome 16B (neuroophthalmic type), 619425
POLH	100%	100%	Xeroderma pigmentosum, variant type, 278750
POLL	100%	100%	No OMIM disease ID
POLR1A	100%	100%	Acrofacial dysostosis, Cincinnati type, 616462
POLR1B	100%	100%	Treacher-Collins syndrome 4, 618939
POLR1C	83%	82%	Leukodystrophy, hypomyelinating, 11, 616494 Treacher Collins syndrome 3, 248390
POLR1D	100%	100%	Treacher Collins syndrome 2, 613717
POLR2A	100%	100%	Neurodevelopmental disorder with hypotonia and variable intellectual and behavioral abnormalities, 618603

POLR3A	100%	100%	Wiedemann-Rautenstrauch syndrome, 264090 Leukodystrophy, hypomyelinating, 7, with or without oligodontia and/or hypogonadotropic hypogonadism, 607694
POLR3B	100%	100%	Leukodystrophy, hypomyelinating, 8, with or without oligodontia and/or hypogonadotropic hypogonadism, 614381
POLR3GL	100%	100%	Short stature, oligodontia, dysmorphic facies, and motor delay, 619234
POLR3K	100%	100%	Leukodystrophy, hypomyelinating, 21, 619310
POLRMT	100%	100%	No OMIM disease ID
POMC	100%	100%	Obesity, adrenal insufficiency, and red hair due to POMC deficiency, 609734
POMGNT1	100%	100%	Muscular dystrophy-dystroglycanopathy (congenital with mental retardation), type B, 3, 613151 Muscular dystrophy-dystroglycanopathy (limb-girdle), type C, 3, 613157 Retinitis pigmentosa 76, 617123 Muscular dystrophy-dystroglycanopathy (congenital with brain and eye anomalies), type A, 3, 253280
POMGNT2	100%	100%	Muscular dystrophy-dystroglycanopathy (limb-girdle) type C, 8, 618135 Muscular dystrophy-dystroglycanopathy (congenital with brain and eye anomalies, type A, 8, 614830

POMK	100%	100%	?Muscular dystrophy-dystroglycanopathy (limb-girdle), type C, 12, 616094 Muscular dystrophy-dystroglycanopathy (congenital with brain and eye anomalies), type A, 12, 615249
POMP	100%	100%	Proteasome-associated autoinflammatory syndrome 2, 618048 Keratosis linearis with ichthyosis congenita and sclerosing keratoderma, 601952
POMT1	100%	100%	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 mental retardation), type B, 1, 613155
POMT2	100%	100%	Muscular dystrophy-dystroglycanopathy (limb-girdle), type C, 2, 613158 Muscular dystrophy-dystroglycanopathy (congenital with mental retardation), type B, 2, 613156 Muscular dystrophy-dystroglycanopathy (congenital with brain and eye anomalies), type A, 2, 613150
POP1	100%	100%	Anauxetic dysplasia 2, 617396
POPDC3	100%	100%	Muscular dystrophy, limb-girdle, autosomal recessive 26, 618848

POR	100%	100%	Antley-Bixler syndrome with genital anomalies and disordered steroidogenesis, 201750 Disordered steroidogenesis due to cytochrome P450 oxidoreductase, 613571
PORCN	100%	100%	Focal dermal hypoplasia, 305600
POT1	100%	100%	No OMIM disease ID
POU1F1	100%	100%	Pituitary hormone deficiency, combined or isolated, 1, 613038
POU2AF1	100%	100%	No OMIM disease ID
POU3F3	99%	96%	Snijders Blok-Fisher syndrome, 618604
POU3F4	100%	100%	Deafness, X-linked 2, 304400
POU4F1	94%	90%	Ataxia, intention tremor, and hypotonia syndrome, childhood-onset, 619352
POU4F3	100%	100%	Deafness, autosomal dominant 15, 602459
POU6F2	100%	100%	No OMIM disease ID
PPA2	100%	100%	?Sudden cardiac failure, alcohol-induced, 617223 Sudden cardiac failure, infantile, 617222
PPARG	98%	98%	Insulin resistance, severe, digenic, 604367 Lipodystrophy, familial partial, type 3, 604367 Obesity, severe, 601665 Carotid intimal medial thickness 1, 609338
PPCS	100%	100%	Cardiomyopathy, dilated, 2C, 618189
PPIB	100%	100%	Osteogenesis imperfecta, type IX, 259440
PPIL1	100%	100%	Pontocerebellar hypoplasia, type 14, 619301
PPIP5K2	100%	100%	Deafness, autosomal recessive 100, 618422
PPM1D	100%	100%	Breast cancer, somatic, 114480 Jansen de Vries syndrome, 617450
PPM1K	100%	100%	?Maple syrup urine disease, mild variant, 615135
PPOX	100%	100%	Porphyria variegata, 176200
PPP1CB	100%	100%	Noonan syndrome-like disorder with loose anagen hair 2, 617506

PPP1R12A	100%	100%	Genitourinary and/or/brain malformation syndrome, 618820
PPP1R15B	100%	100%	Microcephaly, short stature, and impaired glucose metabolism 2, 616817
PPP1R21	100%	100%	Neurodevelopmental disorder with hypotonia, facial dysmorphism, and brain abnormalities, 619383
PPP1R3A	100%	100%	Insulin resistance, severe, digenic, 125853
PPP2CA	100%	100%	Neurodevelopmental disorder and language delay with or without structural brain abnormalities, 618354
PPP2R1A	93%	93%	Mental retardation, autosomal dominant 36, 616362
PPP2R1B	100%	100%	Lung cancer, somatic, 211980
PPP2R2B	100%	100%	Spinocerebellar ataxia 12, 604326
PPP2R3C	100%	100%	Gonadal dysgenesis, dysmorphic facies, retinal dystrophy, and myopathy, 618419 Spermatogenic failure 36, 618420
PPP2R5B	100%	100%	No OMIM disease ID
PPP2R5C	100%	100%	No OMIM disease ID
PPP2R5D	100%	100%	Mental retardation, autosomal dominant 35, 616355
PPP3CA	100%	100%	Arthrogryposis, cleft palate, craniosynostosis, and impaired intellectual development, 618265 Developmental and epileptic encephalopathy 91, 617711
PPT1	82%	82%	Ceroid lipofuscinosis, neuronal, 1, 256730
PQBP1	100%	100%	Renpenning syndrome, 309500
PRCC	100%	100%	Renal cell carcinoma, papillary, 605074
PRCD	100%	100%	Retinitis pigmentosa 36, 610599
PRDM12	95%	93%	Neuropathy, hereditary sensory and autonomic, type VIII, 616488
PRDM13	100%	100%	No OMIM disease ID
PRDM15	100%	99%	No OMIM disease ID
PRDM16	100%	100%	Left ventricular noncompaction 8, 615373 Cardiomyopathy, dilated, 1LL, 615373
PRDM5	100%	100%	Brittle cornea syndrome 2, 614170
PRDM6	100%	100%	Patent ductus arteriosus 3, 617039
PRDM8	100%	100%	?Epilepsy, progressive myoclonic, 10, 616640
PRDX1	100%	100%	Methylmalonic aciduria and homocystinuria, cblC type, digenic, 277400
PRDX2	100%	100%	No OMIM disease ID

PRDX3	100%	100%	No OMIM disease ID
PREPL	100%	100%	Myasthenic syndrome, congenital, 22, 616224
PRF1	100%	100%	Hemophagocytic lymphohistiocytosis, familial, 2, 603553 Aplastic anemia, 609135 Lymphoma, non-Hodgkin, 605027
PRG4	100%	100%	Camptodactyly-arthropathy-coxa vara-pericarditis syndrome, 208250
PRICKLE1	100%	100%	Epilepsy, progressive myoclonic 1B, 612437
PRICKLE2	100%	100%	No OMIM disease ID
PRIMPOL	100%	100%	Myopia 22, autosomal dominant, 615420
PRKAA1	100%	100%	No OMIM disease ID
PRKACA	100%	100%	Cushing syndrome, ACTH-independent adrenal, somatic, 615830 Cardioacrofacial dysplasia 1, 619142
PRKACB	100%	100%	Cardioacrofacial dysplasia 2, 619143
PRKACG	100%	100%	?Bleeding disorder, platelet-type, 19, 616176
PRKAG2	100%	100%	Glycogen storage disease of heart, lethal congenital, 261740 Wolff-Parkinson-White syndrome, 194200 Cardiomyopathy, hypertrophic 6, 600858
PRKAR1A	100%	100%	Pigmented nodular adrenocortical disease, primary, 1, 610489 Acrodysostosis 1, with or without hormone resistance, 101800 Carney complex, type 1, 160980 Myxoma, intracardiac, 255960 Adrenocortical tumor, somatic,
PRKAR1B	100%	100%	No OMIM disease ID
PRKCA	100%	100%	Pituitary tumor, invasive,
PRKCB	100%	100%	No OMIM disease ID
PRKCD	100%	100%	Autoimmune lymphoproliferative syndrome, type III, 615559
PRKCG	100%	100%	Spinocerebellar ataxia 14, 605361

PRKCSH	100%	100%	Polycystic liver disease 1, 174050
PRKD1	100%	100%	Congenital heart defects and ectodermal dysplasia, 617364
PRKDC	100%	100%	Immunodeficiency 26, with or without neurologic abnormalities, 615966
PRKG1	92%	92%	Aortic aneurysm, familial thoracic 8, 615436
PRKG2	100%	100%	Spondylometaphyseal dysplasia, Pagnamenta type, 619638 Acromesomelic dysplasia 4, 619636
PRKN	75%	75%	Adenocarcinoma of lung, somatic, 211980 Parkinson disease, juvenile, type 2, 600116 Ovarian cancer, somatic, 167000
PRKRA	100%	100%	Dystonia 16, 612067
PRLR	100%	100%	Multiple fibroadenomas of the breast, 615554 Hyperprolactinemia, 615555
PRMT7	100%	100%	Short stature, brachydactyly, intellectual developmental disability, and seizures, 617157
PRNP	100%	100%	Spongiform encephalopathy with neuropsychiatric features, 606688 Gerstmann-Straussler disease, 137440 Huntington disease-like 1, 603218 Insomnia, fatal familial, 600072 Cerebral amyloid angiopathy, PRNP-related, 137440 Creutzfeldt-Jakob disease, 123400
PROC	100%	100%	Thrombophilia due to protein C deficiency, autosomal recessive, 612304 Thrombophilia due to protein C deficiency, autosomal dominant, 176860
PRODH	100%	100%	Hyperprolinemia, type I, 239500
PROK2	100%	100%	Hypogonadotropic hypogonadism 4 with or without anosmia, 610628
PROKR2	100%	100%	Hypogonadotropic hypogonadism 3 with or without anosmia, 244200

PROM1	100%	100%	Macular dystrophy, retinal, 2, 608051 Retinitis pigmentosa 41, 612095 Stargardt disease 4, 603786 Cone-rod dystrophy 12, 612657
PROP1	100%	100%	Pituitary hormone deficiency, combined, 2, 262600
PRORP	100%	100%	No OMIM disease ID
PROS1	98%	98%	Thrombophilia due to protein S deficiency, autosomal dominant, 612336 Thrombophilia due to protein S deficiency, autosomal recessive, 614514
PROZ	100%	100%	No OMIM disease ID
PRPF3	100%	100%	Retinitis pigmentosa 18, 601414
PRPF31	100%	100%	Retinitis pigmentosa 11, 600138
PRPF4	100%	100%	Retinitis pigmentosa 70, 615922
PRPF6	100%	100%	Retinitis pigmentosa 60, 613983
PRPF8	100%	100%	Retinitis pigmentosa 13, 600059
PRPH2	100%	100%	Macular dystrophy, patterned, 1, 169150 Choroidal dystrophy, central areolar 2, 613105 Retinitis punctata albescens, 136880 Leber congenital amaurosis 18, 608133 Macular dystrophy, vitelliform, 3, 608161 Retinitis pigmentosa 7 and digenic form, 608133

PRPS1	100%	100%	Arts syndrome, 301835 Phosphoribosylpyrophosphate synthetase superactivity, 300661 Charcot-Marie-Tooth disease, X-linked recessive, 5, 311070 Deafness, X-linked 1, 304500 Gout, PRPS-related, 300661
PRR11	100%	100%	No OMIM disease ID
PRR12	100%	100%	Neuroocular syndrome, 619539
PRRT2	100%	100%	Convulsions, familial infantile, with paroxysmal choreoathetosis, 602066 Seizures, benign familial infantile, 2, 605751 Episodic kinesigenic dyskinesia 1, 128200
PRRX1	100%	100%	Agnathia-otocephaly complex, 202650
PRSS1	100%	100%	Pancreatitis, hereditary, 167800
PRSS12	100%	100%	Mental retardation, autosomal recessive 1, 249500
PRSS56	100%	100%	Microphthalmia, isolated 6, 613517
PRUNE1	93%	93%	Neurodevelopmental disorder with microcephaly, hypotonia, and variable brain anomalies, 617481
PRX	98%	97%	Charcot-Marie-Tooth disease, type 4F, 614895 Dejerine-Sottas disease, 145900
PSAP	100%	100%	Combined SAP deficiency, 611721 Krabbe disease, atypical, 611722 Metachromatic leukodystrophy due to SAP-b deficiency, 249900 Gaucher disease, atypical, 610539
PSAT1	100%	100%	Neu-Laxova syndrome 2, 616038 ?Phosphoserine aminotransferase deficiency, 610992

PSEN1	100%	100%	Pick disease, 172700 Alzheimer disease, type 3, with spastic paraparesis and apraxia, 607822 Dementia, frontotemporal, 600274 ?Acne inversa, familial, 3, 613737 Cardiomyopathy, dilated, 1U, 613694 Alzheimer disease, type 3, with spastic paraparesis and unusual plaques, 607822 Alzheimer disease, type 3, 607822
PSEN2	100%	100%	Alzheimer disease-4, 606889 Cardiomyopathy, dilated, 1V, 613697
PSENEN	100%	100%	Acne inversa, familial, 2, with or without Dowling-Degos disease, 613736
PSIP1	100%	100%	No OMIM disease ID
PSMA3	100%	100%	No OMIM disease ID
PSMB1	100%	100%	No OMIM disease ID
PSMB10	100%	100%	Proteasome-associated autoinflammatory syndrome 5, 619175
PSMB4	100%	100%	?Proteasome-associated autoinflammatory syndrome 3 and digenic forms, 617591
PSMB8	100%	100%	Proteasome-associated autoinflammatory syndrome 1 and digenic forms, 256040
PSMB9	100%	100%	?Proteasome-associated autoinflammatory syndrome 3, digenic, 617591
PSMC3	100%	100%	?Deafness, cataract, impaired intellectual development, and polyneuropathy, 619354
PSMC3IP	100%	100%	Ovarian dysgenesis 3, 614324
PSMC5	100%	100%	No OMIM disease ID
PSMD12	100%	100%	Stankiewicz-Isidor syndrome, 617516
PSMG2	100%	100%	?Proteasome-associated autoinflammatory syndrome 4, 619183
PSPH	100%	100%	Phosphoserine phosphatase deficiency, 614023
PSTPIP1	100%	100%	Pyogenic sterile arthritis, pyoderma gangrenosum, and acne, 604416
PTCD3	100%	100%	?Combined oxidative phosphorylation deficiency 51, 619057
PTCH1	100%	100%	Basal cell carcinoma, somatic, 605462 Holoprosencephaly 7, 610828 Basal cell nevus syndrome, 109400
PTCH2	100%	100%	Medulloblastoma, somatic, 155255 Basal cell nevus syndrome, 109400 Basal cell carcinoma, somatic, 605462

PTCHD1	100%	100%	No OMIM disease ID
PTDSS1	100%	100%	Lenz-Majewski hyperostotic dwarfism, 151050
PTEN	100%	100%	Lhermitte-Duclos syndrome, 158350 Cowden syndrome 1, 158350 Prostate cancer, somatic, 176807 Macrocephaly/autism syndrome, 605309
PTF1A	100%	100%	Pancreatic and cerebellar agenesis, 609069 Pancreatic agenesis 2, 615935
PTGIS	100%	100%	Hypertension, essential, 145500
PTGS1	100%	100%	No OMIM disease ID
PTH	100%	100%	Hypoparathyroidism, familial isolated 1, 146200
PTH1R	100%	100%	Metaphyseal chondrodysplasia, Murk Jansen type, 156400 Eiken syndrome, 600002 Failure of tooth eruption, primary, 125350 Chondrodysplasia, Blomstrand type, 215045
PTHLH	100%	100%	Brachydactyly, type E2, 613382
PTPN11	100%	100%	Noonan syndrome 1, 163950 LEOPARD syndrome 1, 151100 Metachondromatosis, 156250 Leukemia, juvenile myelomonocytic, somatic, 607785
PTPN12	100%	100%	Colon cancer, somatic, 114500
PTPN14	100%	100%	Choanal atresia and lymphedema, 613611
PTPN22	100%	100%	No OMIM disease ID
PTPN23	100%	100%	Neurodevelopmental disorder and structural brain anomalies with or without seizures and spasticity, 618890
PTPRC	100%	100%	Severe combined immunodeficiency, T cell-negative, B-cell/natural killer-cell positive, 608971
PTPRF	100%	100%	?Breasts and/or nipples, aplasia or hypoplasia of, 2, 616001
PTPRJ	100%	100%	Colon cancer, somatic, 114500
PTPRO	100%	100%	Nephrotic syndrome, type 6, 614196

PTPRQ	92%	92%	Deafness, autosomal dominant 73, 617663 Deafness, autosomal recessive 84A, 613391
PTRH2	100%	100%	Infantile-onset multisystem neurologic, endocrine, and pancreatic disease, 616263
PTRHD1	100%	100%	No OMIM disease ID
PTS	100%	100%	Hyperphenylalaninemia, BH4-deficient, A, 261640
PUF60	100%	100%	Verheij syndrome, 615583
PUM1	100%	100%	Spinocerebellar ataxia 47, 617931
PURA	100%	100%	Neurodevelopmental disorder with neonatal respiratory insufficiency, hypotonia, and feeding difficulties, 616158
PUS1	100%	99%	Myopathy, lactic acidosis, and sideroblastic anemia 1, 600462
PUS3	100%	100%	Neurodevelopmental disorder with microcephaly and gray sclerae, 617051
PUS7	100%	100%	Intellectual developmental disorder with abnormal behavior, microcephaly, and short stature, 618342
PXDN	100%	100%	Anterior segment dysgenesis 7, with sclerocornea, 269400
PYCR1	100%	100%	Cutis laxa, autosomal recessive, type IIIB, 614438 Cutis laxa, autosomal recessive, type IIB, 612940
PYCR2	100%	100%	Leukodystrophy, hypomyelinating, 10, 616420
PYGL	100%	100%	Glycogen storage disease VI, 232700
PYGM	100%	100%	McArdle disease, 232600
PYROXD1	100%	100%	Myopathy, myofibrillar, 8, 617258
QARS1	100%	100%	Microcephaly, progressive, seizures, and cerebral and cerebellar atrophy, 615760
QDPR	100%	100%	Hyperphenylalaninemia, BH4-deficient, C, 261630
QRICH1	100%	100%	Ververi-Brady syndrome, 617982
QRICH2	100%	100%	Spermatogenic failure 35, 618341
QRSL1	100%	100%	Combined oxidative phosphorylation deficiency 40, 618835
RAB11B	100%	100%	Neurodevelopmental disorder with ataxic gait, absent speech, and decreased cortical white matter, 617807
RAB14	100%	100%	No OMIM disease ID
RAB18	100%	100%	Warburg micro syndrome 3, 614222
RAB23	100%	100%	Carpenter syndrome, 201000
RAB27A	100%	100%	Griscelli syndrome, type 2, 607624
RAB28	100%	100%	Cone-rod dystrophy 18, 615374
RAB33B	100%	100%	Smith-McCort dysplasia 2, 615222

RAB39B	100%	100%	Intellectual developmental disorder, X-linked 72, 300271 Waisman syndrome, 311510
RAB3GAP1	99%	99%	Martsolf syndrome 2, 619420 Warburg micro syndrome 1, 600118
RAB3GAP2	100%	100%	Martsolf syndrome 1, 212720 Warburg micro syndrome 2, 614225
RAB7A	100%	100%	Charcot-Marie-Tooth disease, type 2B, 600882
RAC1	100%	100%	Mental retardation, autosomal dominant 48, 617751
RAC2	100%	100%	Immunodeficiency 73A with defective neutrophil chemotaxix and leukocytosis, 608203 ?Immunodeficiency 73C with defective neutrophil chemotaxis and hypogammaglobulinemia, 618987 Immunodeficiency 73B with defective neutrophil chemotaxis and lymphopenia, 618986
RAC3	100%	100%	Neurodevelopmental disorder with structural brain anomalies and dysmorphic facies, 618577
RAD21	100%	100%	Cornelia de Lange syndrome 4, 614701 ?Mungan syndrome, 611376
RAD21L1	100%	100%	No OMIM disease ID
RAD50	100%	100%	Nijmegen breakage syndrome-like disorder, 613078
RAD51	89%	89%	Mirror movements 2, 614508 Fanconi anemia, complementation group R, 617244
RAD51B	97%	93%	No OMIM disease ID
RAD51C	100%	100%	Fanconi anemia, complementation group O, 613390
RAD51D	100%	100%	No OMIM disease ID
RAD54B	100%	100%	Colon cancer, somatic, 114500 Lymphoma, non-Hodgkin, somatic, 605027

RAD54L	100%	100%	Lymphoma, non-Hodgkin, somatic, 605027 Adenocarcinoma, colonic, somatic,
RAF1	100%	100%	Cardiomyopathy, dilated, 1NN, 615916 Noonan syndrome 5, 611553 LEOPARD syndrome 2, 611554
RAG1	100%	100%	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%	Severe combined immunodeficiency, B cell-negative, 601457 Combined cellular and humoral immune defects with granulomas, 233650 Omenn syndrome, 603554
RAI1	100%	100%	Smith-Magenis syndrome, 182290
RALA	100%	100%	Hiatt-Neu-Cooper neurodevelopmental syndrome, 619311
RALGAPA1	100%	100%	Neurodevelopmental disorder with hypotonia, neonatal respiratory insufficiency, and thermodysregulation, 618797
RANBP2	100%	100%	No OMIM disease ID
RANGRF	100%	100%	No OMIM disease ID
RAP1GDS1	100%	100%	Lymphocytic leukemia, acute T-cell,
RAPGEF2	100%	100%	?Epilepsy, familial adult myoclonic, 7, 618075
RAPSN	100%	100%	Fetal akinesia deformation sequence 2, 618388 Myasthenic syndrome, congenital, 11, associated with acetylcholine receptor deficiency, 616326

RARB	100%	100%	Microphthalmia, syndromic 12, 615524
RARS1	94%	94%	Leukodystrophy, hypomyelinating, 9, 616140
RARS2	100%	100%	Pontocerebellar hypoplasia, type 6, 611523
RASA1	100%	100%	Capillary malformation-arteriovenous malformation 1, 608354 Basal cell carcinoma, somatic, 605462
RASEF	100%	100%	No OMIM disease ID
RASGRP1	100%	100%	Immunodeficiency 64, 618534
RASGRP2	100%	100%	?Bleeding disorder, platelet-type, 18, 615888
RAX	100%	100%	Microphthalmia, isolated 3, 611038
RAX2	100%	100%	Cone-rod dystrophy 11, 610381 ?Macular degeneration, age-related, 6, 613757
RB1	100%	100%	Small cell cancer of the lung, somatic, 182280 Bladder cancer, somatic, 109800 Retinoblastoma, trilateral, 180200 Osteosarcoma, somatic, 259500 Retinoblastoma, 180200
RB1CC1	100%	100%	Breast cancer, somatic, 114480
RBBP6	100%	100%	No OMIM disease ID
RBBP8	100%	100%	Seckel syndrome 2, 606744 Jawad syndrome, 251255 Pancreatic carcinoma, somatic,
RBCK1	100%	100%	Polyglucosan body myopathy 1 with or without immunodeficiency, 615895
RBFOX1	100%	99%	No OMIM disease ID
RBM10	100%	100%	TARP syndrome, 311900
RBM20	100%	100%	Cardiomyopathy, dilated, 1DD, 613172
RBM28	100%	100%	?Alopecia, neurologic defects, and endocrinopathy syndrome, 612079
RBM8A	100%	100%	Thrombocytopenia-absent radius syndrome, 274000
RBMX	100%	100%	?Intellectual developmental disorder, syndromic 11, Shashi type, 300238
RBP3	100%	100%	?Retinitis pigmentosa 66, 615233

RBP4	100%	100%	Microphthalmia, isolated, with coloboma 10, 616428 Retinal dystrophy, iris coloboma, and comedogenic acne syndrome, 615147
RBPJ	100%	100%	Adams-Oliver syndrome 3, 614814
RC3H1	100%	100%	?Immune dysregulation and systemic hyperinflammation syndrome, 618998
RCBTB1	100%	100%	Retinal dystrophy with or without extraocular anomalies, 617175
RD3	100%	100%	Leber congenital amaurosis 12, 610612
RDH11	100%	100%	?Retinal dystrophy, juvenile cataracts, and short stature syndrome, 616108
RDH12	100%	100%	Leber congenital amaurosis 13, 612712
RDH5	100%	100%	Fundus albipunctatus, 136880
RDX	100%	100%	Deafness, autosomal recessive 24, 611022
REC114	100%	100%	Oocyte maturation defect 10, 619176
RECQL4	100%	100%	Baller-Gerold syndrome, 218600 Rothmund-Thomson syndrome, type 2, 268400 RAPADILINO syndrome, 266280
REEP1	100%	100%	?Neuronopathy, distal hereditary motor, type VB, 614751 Spastic paraplegia 31, autosomal dominant, 610250
REEP2	100%	100%	?Spastic paraplegia 72, autosomal dominant, 615625 ?Spastic paraplegia 72, autosomal recessive, 615625
REEP6	99%	94%	Retinitis pigmentosa 77, 617304
REL	99%	98%	No OMIM disease ID
RELA	100%	100%	?Mucocutaneous ulceration, chronic, 618287
RELB	100%	100%	?Immunodeficiency 53, 617585
RELN	100%	100%	Lissencephaly 2 (Norman-Roberts type), 257320
RELT	100%	100%	Amelogenesis imperfecta, type IIIC, 618386
REN	100%	100%	Renal tubular dysgenesis, 267430 Tubulointerstitial kidney disease, autosomal dominant, 4, 613092
REPS1	100%	100%	?Neurodegeneration with brain iron accumulation 7, 617916

RERE	99%	99%	Neurodevelopmental disorder with or without anomalies of the brain, eye, or heart, 616975
REST	98%	98%	?Deafness, autosomal dominant 27, 612431 Fibromatosis, gingival, 5, 617626
RET	100%	100%	Multiple endocrine neoplasia IIA, 171400 Medullary thyroid carcinoma, 155240 Pheochromocytoma, 171300 Multiple endocrine neoplasia IIB, 162300
RETREG1	100%	100%	Neuropathy, hereditary sensory and autonomic, type IIB, 613115
REV3L	97%	97%	No OMIM disease ID
RFC1	100%	100%	Cerebellar ataxia, neuropathy, and vestibular areflexia syndrome, 614575
RFT1	100%	100%	Congenital disorder of glycosylation, type In, 612015
RFWD3	100%	100%	?Fanconi anemia, complementation group W, 617784
RFX3	100%	100%	No OMIM disease ID
RFX4	100%	100%	No OMIM disease ID
RFX5	100%	100%	Bare lymphocyte syndrome, type II, complementation group C, 209920 Bare lymphocyte syndrome, type II, complementation group E, 209920
RFX6	100%	100%	Mitchell-Riley syndrome, 615710
RFX7	99%	99%	No OMIM disease ID
RFXANK	100%	100%	MHC class II deficiency, complementation group B, 209920
RFXAP	100%	100%	Bare lymphocyte syndrome, type II, complementation group D, 209920
RGR	99%	99%	Retinitis pigmentosa 44, 613769
RGS10	100%	100%	No OMIM disease ID
RGS9	100%	100%	Bradyopsia, 608415
RGS9BP	100%	100%	Bradyopsia, 608415
RHAG	100%	100%	Overhydrated hereditary stomatocytosis, 185000 Anemia, hemolytic, Rh-null, regulator type, 268150
RHBDF2	100%	100%	Tylosis with esophageal cancer, 148500
RHCE	96%	96%	Rh-null disease, amorph type, 617970

RHEB	100%	100%	No OMIM disease ID
RHO	100%	100%	Night blindness, congenital stationary, autosomal dominant 1, 610445 Retinitis pigmentosa 4, autosomal dominant or recessive, 613731 Retinitis punctata albescens, 136880
RHOA	80%	80%	Ectodermal dysplasia with facial dysmorphism and acral, ocular, and brain anomalies, somatic mosaic, 618727
RHOBTB2	100%	100%	Developmental and epileptic encephalopathy 64, 618004
RHOG	100%	100%	No OMIM disease ID
RHOH	100%	100%	No OMIM disease ID
RIC1	100%	100%	CATIFA syndrome, 618761
RIMS1	100%	100%	Cone-rod dystrophy 7, 603649
RIMS2	97%	97%	Cone-rod synaptic disorder syndrome, congenital nonprogressive, 618970
RIN2	100%	100%	Macrocephaly, alopecia, cutis laxa, and scoliosis, 613075
RINT1	100%	100%	Infantile liver failure syndrome 3, 618641
RIPK1	100%	100%	Immunodeficiency 57 with autoinflammation, 618108 Autoinflammation with episodic fever and lymphadenopathy, 618852
RIPK4	100%	100%	CHAND syndrome, 214350 Popliteal pterygium syndrome, Bartsocas-Papas type 1, 263650
RIPOR2	100%	100%	?Deafness, autosomal recessive 104, 616515
RIPPLY2	100%	100%	?Spondylocostal dysostosis 6, 616566
RIT1	100%	100%	Noonan syndrome 8, 615355
RLBP1	100%	100%	Bothnia retinal dystrophy, 607475 Newfoundland rod-cone dystrophy, 607476 Retinitis punctata albescens, 136880 Fundus albipunctatus, 136880
RLIM	100%	100%	Tonne-Kalscheuer syndrome, 300978
RMND1	100%	100%	Combined oxidative phosphorylation deficiency 11, 614922

RMRP	NC	NC	Anauxetic dysplasia 1, 607095 Metaphyseal dysplasia without hypotrichosis, 250460 Cartilage-hair hypoplasia, 250250
RNASEH1	100%	100%	Progressive external ophthalmoplegia with mitochondrial DNA deletions, autosomal recessive 2, 616479
RNASEH2A	100%	100%	Aicardi-Goutieres syndrome 4, 610333
RNASEH2B	91%	91%	Aicardi-Goutieres syndrome 2, 610181
RNASEH2C	100%	100%	Aicardi-Goutieres syndrome 3, 610329
RNASEL	100%	100%	Prostate cancer 1, 601518
RNASET2	100%	100%	Leukoencephalopathy, cystic, without megalencephaly, 612951
RNF113A	100%	100%	Trichothiodystrophy 5, nonphotosensitive, 300953
RNF125	100%	100%	Tenorio syndrome, 616260
RNF13	100%	100%	Developmental and epileptic encephalopathy 73, 618379
RNF139	100%	100%	Renal cell carcinoma, 144700
RNF168	100%	100%	RIDDLE syndrome, 611943
RNF170	100%	100%	Ataxia, sensory, 1, autosomal dominant, 608984
RNF2	100%	100%	Luo-Schoch-Yamamoto syndrome, 619460
RNF212	100%	100%	Recombination rate QTL 1, 612042
RNF216	100%	100%	Cerebellar ataxia and hypogonadotropic hypogonadism, 212840
RNF220	100%	100%	No OMIM disease ID
RNF31	100%	100%	No OMIM disease ID
RNF43	100%	100%	Sessile serrated polyposis cancer syndrome, 617108
RNF6	100%	100%	Esophageal carcinoma, somatic, 133239
RNPC3	100%	100%	Pituitary hormone deficiency, combined or isolated, 7, 618160
RNU4ATAC	NC	NC	Roifman syndrome, 616651 Lowry-Wood syndrome, 226960 Microcephalic osteodysplastic primordial dwarfism, type I, 210710
RNU7-1	NC	NC	Aicardi-Goutieres syndrome 9, 619487
ROBO1	100%	100%	No OMIM disease ID
ROBO2	100%	100%	Vesicoureteral reflux 2, 610878
ROBO3	100%	100%	Gaze palsy, familial horizontal, with progressive scoliosis, 1, 607313
ROBO4	100%	100%	Aortic valve disease 3, 618496
ROGDI	100%	100%	Kohlschutter-Tonz syndrome, 226750

ROM1	100%	100%	Retinitis pigmentosa 7, digenic form, 608133
ROR1	100%	100%	?Deafness, autosomal recessive 108, 617654
ROR2	97%	97%	Brachydactyly, type B1, 113000 Robinow syndrome, autosomal recessive, 268310
RORA	100%	100%	Intellectual developmental disorder with or without epilepsy or cerebellar ataxia, 618060
RORC	100%	100%	Immunodeficiency 42, 616622
RP1	100%	100%	Retinitis pigmentosa 1, 180100
RP1L1	100%	100%	Occult macular dystrophy, 613587 Retinitis pigmentosa 88, 618826
RP2	100%	100%	Retinitis pigmentosa 2, 312600
RP9	100%	100%	?Retinitis pigmentosa 9, 180104
RPA1	100%	100%	No OMIM disease ID
RPE65	100%	100%	Retinitis pigmentosa 20, 613794 Retinitis pigmentosa 87 with choroidal involvement, 618697 Leber congenital amaurosis 2, 204100
RPGR	100%	99%	Retinitis pigmentosa, X-linked, and sinorespiratory infections, with or without deafness, 300455 Cone-rod dystrophy, X-linked, 1, 304020 Retinitis pigmentosa 3, 300029 Macular degeneration, X-linked atrophic, 300834
RPGRIP1	100%	100%	Cone-rod dystrophy 13, 608194 Leber congenital amaurosis 6, 613826
RPGRIP1L	99%	99%	Joubert syndrome 7, 611560 Meckel syndrome 5, 611561 ?COACH syndrome 3, 619113
RPIA	100%	100%	Ribose 5-phosphate isomerase deficiency, 608611
RPL10	100%	100%	Intellectual developmental disorder, X-linked, syndromic, 35, 300998
RPL11	100%	100%	Diamond-Blackfan anemia 7, 612562
RPL13	100%	100%	Spondyloepimetaphyseal dysplasia, Isidor-Toutain type, 618728
RPL15	100%	100%	?Diamond-Blackfan anemia 12, 615550

RPL18	100%	100%	?Diamond-Blackfan anemia 18, 618310
RPL21	100%	100%	Hypotrichosis 12, 615885
RPL26	100%	100%	?Diamond-Blackfan anemia 11, 614900
RPL27	100%	100%	?Diamond-Blackfan anemia 16, 617408
RPL31	100%	100%	No OMIM disease ID
RPL35	100%	100%	?Diamond-Blackfan anemia 19, 618312
RPL35A	100%	100%	Diamond-Blackfan anemia 5, 612528
RPL3L	100%	100%	Cardiomyopathy, dilated, 2D, 619371
RPL4	100%	100%	No OMIM disease ID
RPL5	100%	100%	Diamond-Blackfan anemia 6, 612561
RPL9	100%	100%	No OMIM disease ID
RPN2	100%	100%	No OMIM disease ID
RPS10	100%	100%	Diamond-Blackfan anemia 9, 613308
RPS14	100%	100%	Macrocytic anemia, refractory, due to 5q deletion, somatic, 153550
RPS15A	80%	80%	?Diamond-Blackfan anemia 20, 618313
RPS17	100%	100%	Diamond-Blackfan anemia 4, 612527
RPS19	100%	100%	Diamond-Blackfan anemia 1, 105650
RPS20	100%	100%	No OMIM disease ID
RPS23	100%	100%	Brachycephaly, trichomegaly, and developmental delay, 617412
RPS24	100%	100%	Diamond-blackfan anemia 3, 610629
RPS26	100%	100%	Diamond-Blackfan anemia 10, 613309
RPS27	100%	100%	?Diamond-Blackfan anemia 17, 617409
RPS28	100%	100%	Diamond Blackfan anemia 15 with mandibulofacial dysostosis, 606164
RPS29	100%	100%	Diamond-Blackfan anemia 13, 615909
RPS6KA3	100%	99%	Intellectual developmental disorder, X-linked 19, 300844 Coffin-Lowry syndrome, 303600
RPS7	100%	100%	Diamond-Blackfan anemia 8, 612563
RPSA	100%	100%	Asplenia, isolated congenital, 271400
RRAD	100%	100%	No OMIM disease ID
RRAGC	100%	100%	No OMIM disease ID
RRAS	100%	100%	No OMIM disease ID
RRAS2	100%	100%	Noonan syndrome 12, 618624 Ovarian carcinoma,

RREB1	100%	100%	No OMIM disease ID
RRM1	100%	100%	No OMIM disease ID
RRM2B	100%	100%	Mitochondrial DNA depletion syndrome 8B (MNGIE type), 612075 Mitochondrial DNA depletion syndrome 8A (encephalomyopathic type with renal tubulopathy), 612075 Progressive external ophthalmoplegia with mitochondrial DNA deletions, autosomal dominant 5, 613077
RRP7A	100%	100%	?Microcephaly 28, primary, autosomal recessive, 619453
RS1	100%	100%	Retinoschisis, 312700
RSPH1	100%	100%	Ciliary dyskinesia, primary, 24, 615481
RSPH3	100%	100%	Ciliary dyskinesia, primary, 32, 616481
RSPH4A	100%	100%	Ciliary dyskinesia, primary, 11, 612649
RSPH9	100%	100%	Ciliary dyskinesia, primary, 12, 612650
RSPO1	100%	100%	Palmoplantar hyperkeratosis and true hermaphroditism, 610644 Palmoplantar hyperkeratosis with squamous cell carcinoma of skin and sex reversal, 610644
RSPO2	100%	100%	?Humerofemoral hypoplasia with radiotibial ray deficiency, 618022 Tetraamelia syndrome 2, 618021
RSPO4	100%	100%	Anonychia congenita, 206800
RSPRY1	100%	100%	Spondyloepimetaphyseal dysplasia, Faden-Alkuraya type, 616723
RSRC1	100%	100%	Intellectual developmental disorder, autosomal recessive 70, 618402
RTEL1	100%	100%	Pulmonary fibrosis and/or bone marrow failure, telomere-related, 3, 616373 Dyskeratosis congenita, autosomal dominant 4, 615190 Dyskeratosis congenita, autosomal recessive 5, 615190
RTN2	100%	100%	Spastic paraplegia 12, autosomal dominant, 604805

RTN4IP1	100%	100%	Optic atrophy 10 with or without ataxia, mental retardation, and seizures, 616732
RTTN	100%	100%	Microcephaly, short stature, and polymicrogyria with seizures, 614833
RUBCN	100%	100%	Spinocerebellar ataxia, autosomal recessive 15, 615705
RUNX1	100%	100%	Platelet disorder, familial, with associated myeloid malignancy, 601399 Leukemia, acute myeloid, 601626
RUNX2	100%	100%	Metaphyseal dysplasia with maxillary hypoplasia with or without brachydactyly, 156510 Cleidocranial dysplasia, forme fruste, with brachydactyly, 119600 Cleidocranial dysplasia, forme fruste, dental anomalies only, 119600 Cleidocranial dysplasia, 119600
RUSC2	100%	100%	Mental retardation, autosomal recessive 61, 617773
RXYLT1	100%	100%	Muscular dystrophy-dystroglycanopathy (congenital with brain and eye anomalies), type A, 10, 615041
RYR1	100%	99%	Neuromuscular disease, congenital, with uniform type 1 fiber, 117000 Central core disease, 117000 King-Denborough syndrome, 619542 Minicore myopathy with external ophthalmoplegia, 255320
RYR2	100%	100%	Ventricular tachycardia, catecholaminergic polymorphic, 1, 604772 Ventricular arrythmias due to cardiac ryanodine receptor calcium release deficiency syndrome, 115000 Arrhythmogenic right ventricular dysplasia 2, 600996
S1PR2	100%	100%	Deafness, autosomal recessive 68, 610419
SACS	100%	100%	Spastic ataxia, Charlevoix-Saguenay type, 270550
SAG	100%	100%	Retinitis pigmentosa 47, 613758 Oguchi disease-1, 258100

SALL1	100%	100%	Townes-Brocks syndrome 1, 107480 Townes-Brocks branchiootorenal-like syndrome, 107480
SALL2	100%	100%	?Coloboma, ocular, autosomal recessive, 216820
SALL4	100%	100%	?IVIC syndrome, 147750 Duane-radial ray syndrome, 607323
SAMD11	100%	100%	No OMIM disease ID
SAMD12	100%	100%	Epilepsy, familial adult myoclonic, 1, 601068
SAMD9	100%	100%	Tumoral calcinosis, familial, normophosphatemic, 610455 Monosomy 7 myelodysplasia and leukemia syndrome 2, 619041 MIRAGE syndrome, 617053
SAMD9L	100%	100%	Ataxia-pancytopenia syndrome, 159550 Monosomy 7 myelodysplasia and leukemia syndrome 1, 252270
SAMHD1	100%	100%	?Chilblain lupus 2, 614415 Aicardi-Goutieres syndrome 5, 612952
SAR1B	100%	100%	Chylomicron retention disease, 246700
SARDH	91%	91%	No OMIM disease ID
SARS1	100%	100%	?Neurodevelopmental disorder with microcephaly, ataxia, and seizures, 617709
SARS2	100%	100%	Hyperuricemia, pulmonary hypertension, renal failure, and alkalosis, 613845
SART3	100%	100%	No OMIM disease ID
SASH1	100%	100%	Dyschromatosis universalis hereditaria 1, 127500 ?Cancer, alopecia, pigment dyscrasia, onychodystrophy, and keratoderma, 618373
SASH3	100%	100%	No OMIM disease ID
SASS6	100%	100%	?Microcephaly 14, primary, autosomal recessive, 616402
SAT1	100%	100%	No OMIM disease ID

SATB1	100%	100%	Kohlschutter-Tonz syndrome-like, 619229 Developmental delay with dysmorphic facies and dental anomalies, 619228
SATB2	100%	100%	Glass syndrome, 612313
SBDS	100%	100%	Shwachman-Diamond syndrome, 260400
SBF1	100%	100%	Charcot-Marie-Tooth disease, type 4B3, 615284
SBF2	100%	100%	Charcot-Marie-Tooth disease, type 4B2, 604563
SC5D	100%	100%	Lathosterolosis, 607330
SCAF4	100%	100%	No OMIM disease ID
SCAMP5	100%	100%	No OMIM disease ID
SCAPER	100%	100%	Intellectual developmental disorder and retinitis pigmentosa, 618195
SCARB2	100%	100%	Epilepsy, progressive myoclonic 4, with or without renal failure, 254900
SCARF2	100%	100%	Van den Ende-Gupta syndrome, 600920
SCD5	100%	100%	?Deafness, autosomal dominant 79, 619086
SCLT1	95%	95%	No OMIM disease ID
SCN10A	100%	100%	Episodic pain syndrome, familial, 2, 615551
SCN11A	100%	100%	Episodic pain syndrome, familial, 3, 615552 Neuropathy, hereditary sensory and autonomic, type VII, 615548
SCN1A	100%	100%	Developmental and epileptic encephalopathy 6B, non-Dravet, 619317 Migraine, familial hemiplegic, 3, 609634 Dravet syndrome, 607208 Febrile seizures, familial, 3A, 604403 Generalized epilepsy with febrile seizures plus, type 2, 604403

SCN1B	100%	99%	Generalized epilepsy with febrile seizures plus, type 1, 604233 Developmental and epileptic encephalopathy 52, 617350 Cardiac conduction defect, nonspecific, 612838 Atrial fibrillation, familial, 13, 615377 Brugada syndrome 5, 612838
SCN2A	100%	100%	Seizures, benign familial infantile, 3, 607745 Developmental and epileptic encephalopathy 11, 613721 Episodic ataxia, type 9, 618924
SCN2B	100%	100%	Atrial fibrillation, familial, 14, 615378
SCN3A	100%	100%	Epilepsy, familial focal, with variable foci 4, 617935 Developmental and epileptic encephalopathy 62, 617938
SCN3B	100%	100%	Atrial fibrillation, familial, 16, 613120 Brugada syndrome 7, 613120
SCN4A	100%	100%	Paramyotonia congenita, 168300 Hypokalemic periodic paralysis, type 2, 613345 Myotonia congenita, atypical, acetazolamide-responsive, 608390 Myasthenic syndrome, congenital, 16, 614198 Hyperkalemic periodic paralysis, type 2, 170500
SCN4B	100%	100%	Atrial fibrillation, familial, 17, 611819 Long QT syndrome 10, 611819

SCN5A	100%	100%	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
SCN7A	100%	100%	No OMIM disease ID
SCN8A	100%	100%	?Myoclonus, familial, 2, 618364 Seizures, benign familial infantile, 5, 617080 Cognitive impairment with or without cerebellar ataxia, 614306 Developmental and epileptic encephalopathy 13, 614558
SCN9A	100%	100%	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
SCNN1A	100%	100%	Pseudohypoaldosteronism, type I, 264350 ?Liddle syndrome 3, 618126 Bronchiectasis with or without elevated sweat chloride 2, 613021

SCNN1B	100%	100%	Bronchiectasis with or without elevated sweat chloride 1, 211400 Pseudohypoaldosteronism, type I, 264350 Liddle syndrome 1, 177200
SCNN1G	100%	100%	Bronchiectasis with or without elevated sweat chloride 3, 613071 Pseudohypoaldosteronism, type I, 264350 Liddle syndrome 2, 618114
SCO1	100%	100%	Mitochondrial complex IV deficiency, nuclear type 4, 619048
SCO2	100%	100%	Myopia 6, 608908 Mitochondrial complex IV deficiency, nuclear type 2, 604377
SCP2	100%	100%	?Leukoencephalopathy with dystonia and motor neuropathy, 613724
SCUBE3	100%	100%	Short stature, facial dysmorphism, and skeletal anomalies with or without cardiac anomalies, 619184
SCYL1	100%	100%	Spinocerebellar ataxia, autosomal recessive 21, 616719
SCYL2	100%	100%	Arthrogryposis multiplex congenita 4, neurogenic, with agenesis of the corpus callosum, 618766
SDCCAG8	100%	100%	Senior-Loken syndrome 7, 613615 Bardet-Biedl syndrome 16, 615993
SDHA	100%	100%	Cardiomyopathy, dilated, 1GG, 613642 Mitochondrial complex II deficiency, nuclear type 1, 252011 Neurodegeneration with ataxia and late-onset optic atrophy, 619259 Paragangliomas 5, 614165
SDHAF1	100%	100%	Mitochondrial complex II deficiency, nuclear type 2, 619166
SDHAF2	100%	99%	Paragangliomas 2, 601650

SDHB	100%	100%	Paragangliomas 4, 115310 Mitochondrial complex II deficiency, nuclear type 4, 619224 Gastrointestinal stromal tumor, 606764 Pheochromocytoma, 171300 Paraganglioma and gastric stromal sarcoma, 606864
SDHC	100%	100%	Paragangliomas 3, 605373 Paraganglioma and gastric stromal sarcoma, 606864 Gastrointestinal stromal tumor, 606764
SDHD	80%	80%	Paragangliomas 1, with or without deafness, 168000 Paraganglioma and gastric stromal sarcoma, 606864 Mitochondrial complex II deficiency, nuclear type 3, 619167 Pheochromocytoma, 171300
SDR9C7	100%	100%	Ichthyosis, congenital, autosomal recessive 13, 617574
SEC23A	100%	100%	Craniolenticulosutural dysplasia, 607812
SEC23B	100%	100%	?Cowden syndrome 7, 616858 Dyserythropoietic anemia, congenital, type II, 224100
SEC24D	100%	100%	Cole-Carpenter syndrome 2, 616294
SEC31A	100%	100%	?Halperin-Birk syndrome, 618651
SEC61A1	100%	100%	Tubulointerstitial kidney disease, autosomal dominant, 5, 617056
SEC61B	100%	100%	No OMIM disease ID
SEC63	100%	100%	Polycystic liver disease 2, 617004
SECISBP2	100%	100%	Thyroid hormone metabolism, abnormal, 609698
SELENBP1	100%	100%	Extraoral halitosis due to MTO deficiency, 618148
SELENOI	100%	100%	Spastic paraplegia 81, autosomal recessive, 618768

SELENON	91%	90%	Myopathy, congenital, with fiber-type disproportion, 255310 Muscular dystrophy, rigid spine, 1, 602771
SEMA3A	100%	100%	No OMIM disease ID
SEMA3E	100%	100%	?CHARGE syndrome, 214800
SEMA4A	100%	100%	Retinitis pigmentosa 35, 610282 Cone-rod dystrophy 10, 610283
SEMA6B	100%	100%	Epilepsy, progressive myoclonic, 11, 618876
SEPSECS	100%	100%	Pontocerebellar hypoplasia type 2D, 613811
SEPTIN12	100%	100%	Spermatogenic failure 10, 614822
SEPTIN9	100%	100%	Amyotrophy, hereditary neuralgic, 162100
SERAC1	100%	100%	3-methylglutaconic aciduria with deafness, encephalopathy, and Leigh-like syndrome, 614739
SERPINA1	100%	100%	Hemorrhagic diathesis due to antithrombin Pittsburgh, 613490 Emphysema due to AAT deficiency, 613490 Emphysema-cirrhosis, due to AAT deficiency, 613490
SERPINA12	100%	100%	No OMIM disease ID
SERPINA3	100%	100%	Alpha-1-antichymotrypsin deficiency, Cerebrovascular disease, occlusive,
SERPINA6	100%	100%	Corticosteroid-binding globulin deficiency, 611489
SERPINB6	100%	100%	?Deafness, autosomal recessive 91, 613453
SERPINB7	100%	100%	Palmoplantar keratoderma, Nagashima type, 615598
SERPINB8	100%	100%	Peeling skin syndrome 5, 617115
SERPINC1	100%	100%	Thrombophilia due to antithrombin III deficiency, 613118
SERPIND1	100%	100%	Thrombophilia due to heparin cofactor II deficiency, 612356
SERPINE1	100%	100%	Plasminogen activator inhibitor-1 deficiency, 613329
SERPINF1	100%	100%	Osteogenesis imperfecta, type VI, 613982
SERPINF2	100%	100%	Alpha-2-plasmin inhibitor deficiency, 262850
SERPING1	100%	100%	Angioedema, hereditary, 1 and 2, 106100 Complement component 4, partial deficiency of, 120790

SERPINH1	100%	100%	Osteogenesis imperfecta, type X, 613848
SERPINI1	100%	100%	Encephalopathy, familial, with neuroserpin inclusion bodies, 604218
SET	100%	99%	Mental retardation, autosomal dominant 58, 618106
SETBP1	100%	100%	Schinzel-Giedion midface retraction syndrome, 269150 Mental retardation, autosomal dominant 29, 616078
SETD1A	100%	100%	Epilepsy, early-onset, with or without developmental delay, 618832 Neurodevelopmental disorder with speech impairment and dysmorphic facies, 619056
SETD1B	100%	100%	Intellectual developmental disorder with seizures and language delay, 619000
SETD2	100%	100%	Luscan-Lumish syndrome, 616831
SETD5	98%	98%	Mental retardation, autosomal dominant 23, 615761
SETX	100%	100%	Spinocerebellar ataxia, autosomal recessive, with axonal neuropathy 2, 606002 Amyotrophic lateral sclerosis 4, juvenile, 602433
SEZ6	100%	100%	No OMIM disease ID
SF3B1	100%	100%	Myelodysplastic syndrome, somatic, 614286
SF3B2	100%	100%	No OMIM disease ID
SF3B4	100%	100%	Acrofacial dysostosis 1, Nager type, 154400
SFRP4	100%	100%	Pyle disease, 265900
SFTPA1	100%	100%	Interstitial lung disease 1, 619611
SFTPA2	100%	100%	Interstitial lung disease 2, 178500
SFTPB	100%	100%	Surfactant metabolism dysfunction, pulmonary, 1, 265120
SFTPC	100%	100%	Surfactant metabolism dysfunction, pulmonary, 2, 610913
SFXN4	100%	100%	Combined oxidative phosphorylation deficiency 18, 615578
SGCA	100%	100%	Muscular dystrophy, limb-girdle, autosomal recessive 3, 608099
SGCB	100%	100%	Muscular dystrophy, limb-girdle, autosomal recessive 4, 604286
SGCD	100%	100%	Cardiomyopathy, dilated, 1L, 606685 Muscular dystrophy, limb-girdle, autosomal recessive 6, 601287

SGCE	91%	91%	Dystonia-11, myoclonic, 159900
SGCG	100%	100%	Muscular dystrophy, limb-girdle, autosomal recessive 5, 253700
SGMS2	100%	100%	Calvarial doughnut lesions with bone fragility with or without spondylometaphyseal dysplasia, 126550
SGO1	100%	100%	Chronic atrial and intestinal dysrhythmia, 616201
SGPL1	100%	100%	Nephrotic syndrome, type 14, 617575
SGSH	100%	100%	Mucopolysaccharidosis type IIIA (Sanfilippo A), 252900
SH2B3	100%	100%	Thrombocythemia, somatic, 187950 Myelofibrosis, somatic, 254450 Erythrocytosis, somatic, 133100
SH2D1A	100%	100%	Lymphoproliferative syndrome, X-linked, 1, 308240
SH3BP2	99%	98%	Cherubism, 118400
SH3KBP1	100%	100%	?Immunodeficiency 61, 300310
SH3PXD2B	100%	100%	Frank-ter Haar syndrome, 249420
SH3TC2	100%	100%	Charcot-Marie-Tooth disease, type 4C, 601596 Mononeuropathy of the median nerve, mild, 613353
SHANK2	98%	98%	No OMIM disease ID
SHANK3	97%	96%	Phelan-McDermid syndrome, 606232
SHH	100%	100%	Microphthalmia with coloboma 5, 611638 Schizencephaly, 269160 Single median maxillary central incisor, 147250 Holoprosencephaly 3, 142945
SHMT2	100%	100%	Neurodevelopmental disorder with cardiomyopathy, spasticity, and brain abnormalities, 619121
SHOC1	100%	100%	No OMIM disease ID
SHOC2	100%	100%	Noonan syndrome-like with loose anagen hair 1, 607721

SHOX	95%	95%	Short stature, idiopathic familial, 300582 Leri-Weill dyschondrosteosis, 127300 Langer mesomelic dysplasia, 249700 Short stature, idiopathic familial, 300582 Langer mesomelic dysplasia, 249700 Leri-Weill dyschondrosteosis, 127300
SHROOM3	100%	100%	No OMIM disease ID
SHROOM4	100%	100%	Intellectual developmental disorder, X-linked syndromic, Stocco dos Santos type, 300434
SI	100%	100%	Sucrase-isomaltase deficiency, congenital, 222900
SIAH1	100%	100%	Buratti-Harel syndrome, 619314
SIGLEC7	100%	100%	No OMIM disease ID
SIGMAR1	100%	100%	?Spinal muscular atrophy, distal, autosomal recessive, 2, 605726 ?Amyotrophic lateral sclerosis 16, juvenile, 614373
SIK1	100%	100%	Developmental and epileptic encephalopathy 30, 616341
SIK3	100%	99%	?Spondyloepimetaphyseal dysplasia, Krakow type, 618162
SIL1	100%	100%	Marinesco-Sjogren syndrome, 248800
SIN3A	100%	100%	Witteveen-Kolk syndrome, 613406
SIN3B	100%	100%	No OMIM disease ID
SIPA1L3	100%	100%	?Cataract 45, 616851
SIX1	100%	100%	Deafness, autosomal dominant 23, 605192 Branchiootic syndrome 3, 608389
SIX3	100%	100%	Schizencephaly, 269160 Holoprosencephaly 2, 157170
SIX5	100%	100%	Branchiootorenal syndrome 2, 610896
SIX6	100%	100%	Optic disc anomalies with retinal and/or macular dystrophy, 212550
SKI	100%	100%	Shprintzen-Goldberg syndrome, 182212
SKIV2L	100%	100%	Trichohepatoenteric syndrome 2, 614602
SLC10A1	100%	100%	Hypercholanemia, familial 2, 619256
SLC10A2	100%	100%	?Bile acid malabsorption, primary, 1, 613291
SLC10A7	100%	100%	Short stature, amelogenesis imperfecta, and skeletal dysplasia with scoliosis, 618363

SLC11A2	100%	100%	Anemia, hypochromic microcytic, with iron overload 1, 206100
SLC12A1	96%	96%	Bartter syndrome, type 1, 601678
SLC12A2	100%	100%	Kilquist syndrome, 619080 Delpire-McNeill syndrome, 619083 Deafness, autosomal dominant 78, 619081
SLC12A3	100%	100%	Gitelman syndrome, 263800
SLC12A5	97%	97%	Developmental and epileptic encephalopathy 34, 616645
SLC12A6	100%	100%	Agenesis of the corpus callosum with peripheral neuropathy, 218000
SLC13A3	100%	100%	Leukoencephalopathy, acute reversible, with increased urinary alpha-ketoglutarate, 618384
SLC13A5	100%	100%	Developmental and epileptic encephalopathy 25, with amelogenesis imperfecta, 615905
SLC16A1	100%	100%	Hyperinsulinemic hypoglycemia, familial, 7, 610021 Erythrocyte lactate transporter defect, 245340 Monocarboxylate transporter 1 deficiency, 616095
SLC16A12	100%	100%	Cataract 47, juvenile, with microcornea, 612018
SLC16A2	100%	100%	Allan-Herndon-Dudley syndrome, 300523
SLC17A5	100%	100%	Salla disease, 604369 Sialic acid storage disorder, infantile, 269920
SLC17A8	100%	100%	Deafness, autosomal dominant 25, 605583
SLC17A9	100%	100%	Porokeratosis 8, disseminated superficial actinic type, 616063
SLC18A2	100%	100%	?Parkinsonism-dystonia, infantile, 2, 618049
SLC18A3	100%	100%	Myasthenic syndrome, congenital, 21, presynaptic, 617239
SLC19A1	100%	100%	?Megaloblastic anemia, folate-responsive, 601775
SLC19A2	100%	100%	Thiamine-responsive megaloblastic anemia syndrome, 249270
SLC19A3	98%	98%	Thiamine metabolism dysfunction syndrome 2 (biotin- or thiamine-responsive encephalopathy type 2), 607483
SLC1A1	100%	100%	Dicarboxylic aminoaciduria, 222730
SLC1A2	100%	100%	Developmental and epileptic encephalopathy 41, 617105
SLC1A3	100%	100%	Episodic ataxia, type 6, 612656
SLC1A4	100%	100%	Spastic tetraplegia, thin corpus callosum, and progressive microcephaly, 616657
SLC20A2	100%	100%	Basal ganglia calcification, idiopathic, 1, 213600
SLC22A12	100%	100%	Hypouricemia, renal, 220150

SLC22A18	100%	100%	Breast cancer, somatic, 114480 Lung cancer, somatic, 211980 Rhabdomyosarcoma, somatic, 268210
SLC22A4	100%	100%	No OMIM disease ID
SLC22A5	100%	100%	Carnitine deficiency, systemic primary, 212140
SLC24A1	100%	100%	Night blindness, congenital stationary (complete), 1D, autosomal recessive, 613830
SLC24A4	100%	100%	Amelogenesis imperfecta, type IIA5, 615887
SLC24A5	100%	100%	Albinism, oculocutaneous, type VI, 113750
SLC25A1	100%	100%	Combined D-2- and L-2-hydroxyglutaric aciduria, 615182 Myasthenic syndrome, congenital, 23, presynaptic, 618197
SLC25A10	100%	100%	?Mitochondrial DNA depletion syndrome 19, 618972
SLC25A11	100%	100%	Paragangliomas 6, 618464
SLC25A12	100%	100%	Developmental and epileptic encephalopathy 39, 612949
SLC25A13	100%	100%	Citrullinemia, type II, neonatal-onset, 605814 Citrullinemia, adult-onset type II, 603471
SLC25A15	100%	100%	Hyperornithinemia-hyperammonemia-homocitrullinemia syndrome, 238970
SLC25A19	100%	100%	Microcephaly, Amish type, 607196 Thiamine metabolism dysfunction syndrome 4 (progressive polyneuropathy type), 613710
SLC25A20	100%	100%	Carnitine-acylcarnitine translocase deficiency, 212138
SLC25A21	100%	100%	?Mitochondrial DNA depletion syndrome 18, 618811
SLC25A22	100%	100%	Developmental and epileptic encephalopathy 3, 609304
SLC25A24	99%	99%	Fontaine progeroid syndrome, 612289
SLC25A26	100%	100%	Combined oxidative phosphorylation deficiency 28, 616794
SLC25A3	100%	100%	Mitochondrial phosphate carrier deficiency, 610773
SLC25A32	100%	100%	?Exercise intolerance, riboflavin-responsive, 616839
SLC25A37	100%	100%	No OMIM disease ID
SLC25A38	100%	100%	Anemia, sideroblastic, 2, pyridoxine-refractory, 205950

SLC25A4	100%	100%	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%	Metabolic crises, recurrent, with variable encephalomyopathic features and neurologic regression, 618416
SLC25A46	100%	100%	Neuropathy, hereditary motor and sensory, type VIB, 616505 Pontocerebellar hypoplasia, type 1E, 619303
SLC26A1	100%	100%	?Nephrolithiasis, calcium oxalate, 167030
SLC26A2	100%	100%	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%	Diarrhea 1, secretory chloride, congenital, 214700
SLC26A4	100%	100%	Deafness, autosomal recessive 4, with enlarged vestibular aqueduct, 600791 Pendred syndrome, 274600
SLC26A5	100%	100%	?Deafness, autosomal recessive 61, 613865
SLC26A8	100%	100%	Spermatogenic failure 3, 606766
SLC27A4	100%	100%	Ichthyosis prematurity syndrome, 608649
SLC28A1	100%	100%	No OMIM disease ID
SLC29A3	100%	100%	Histiocytosis-lymphadenopathy plus syndrome, 602782

SLC2A1	100%	100%	Dystonia 9, 601042 GLUT1 deficiency syndrome 1, infantile onset, severe, 606777 Stomatin-deficient cryohydrocytosis with neurologic defects, 608885 GLUT1 deficiency syndrome 2, childhood onset, 612126
SLC2A10	100%	100%	Arterial tortuosity syndrome, 208050
SLC2A2	100%	100%	Fanconi-Bickel syndrome, 227810
SLC2A9	100%	100%	Hypouricemia, renal, 2, 612076
SLC30A10	100%	100%	Hypermanganesemia with dystonia 1, 613280
SLC30A2	100%	100%	Zinc deficiency, transient neonatal, 608118
SLC30A5	100%	100%	No OMIM disease ID
SLC30A9	100%	100%	?Birk-Landau-Perez syndrome, 617595
SLC33A1	100%	100%	Spastic paraplegia 42, autosomal dominant, 612539 Congenital cataracts, hearing loss, and neurodegeneration, 614482
SLC34A1	100%	100%	?Fanconi renotubular syndrome 2, 613388 Hypercalcemia, infantile, 2, 616963 Nephrolithiasis/osteoporosis, hypophosphatemic, 1, 612286
SLC34A2	100%	100%	Pulmonary alveolar microlithiasis, 265100
SLC34A3	100%	100%	Hypophosphatemic rickets with hypercalciuria, 241530
SLC35A1	100%	100%	Congenital disorder of glycosylation, type IIf, 603585
SLC35A2	100%	100%	Congenital disorder of glycosylation, type IIm, 300896
SLC35A3	81%	81%	?Arthrogryposis, mental retardation, and seizures, 615553
SLC35C1	100%	100%	Congenital disorder of glycosylation, type IIc, 266265
SLC35D1	100%	100%	Schneckenbecken dysplasia, 269250
SLC36A2	100%	100%	Iminoglycinuria, digenic, 242600 Hyperglycinuria, 138500

SLC37A4	100%	100%	Glycogen storage disease Ib, 232220 Congenital disorder of glycosylation, type IIw, 619525 Glycogen storage disease Ic, 232240
SLC38A8	100%	100%	Foveal hypoplasia 2, with or without optic nerve misrouting and/or anterior segment dysgenesis, 609218
SLC39A13	100%	100%	Ehlers-Danlos syndrome, spondylodysplastic type, 3, 612350
SLC39A14	93%	93%	?Hyperostosis cranalis interna, 144755 Hypermanganesemia with dystonia 2, 617013
SLC39A4	100%	100%	Acrodermatitis enteropathica, 201100
SLC39A5	100%	100%	Myopia 24, autosomal dominant, 615946
SLC39A7	100%	100%	No OMIM disease ID
SLC39A8	100%	100%	Congenital disorder of glycosylation, type IIn, 616721
SLC3A1	96%	96%	Cystinuria, 220100
SLC40A1	100%	100%	Hemochromatosis, type 4, 606069
SLC41A1	100%	100%	?Nephronophthisis-like nephropathy 2, 619468
SLC44A1	100%	100%	Neurodegeneration, childhood-onset, with ataxia, tremor, optic atrophy, and cognitive decline, 618868
SLC44A4	100%	100%	?Deafness, autosomal dominant 72, 617606
SLC45A1	100%	100%	Intellectual developmental disorder with neuropsychiatric features, 617532
SLC45A2	100%	100%	Albinism, oculocutaneous, type IV, 606574
SLC46A1	100%	100%	Folate malabsorption, hereditary, 229050
SLC4A1	96%	96%	Distal renal tubular acidosis 1, 179800 Spherocytosis, type 4, 612653 Distal renal tubular acidosis 4 with hemolytic anemia, 611590 Cryohydrocytosis, 185020 Ovalocytosis, SA type, 166900
SLC4A11	100%	100%	Corneal endothelial dystrophy, autosomal recessive, 217700 Corneal dystrophy, Fuchs endothelial, 4, 613268 Corneal endothelial dystrophy and perceptive deafness, 217400
SLC4A4	100%	100%	Renal tubular acidosis, proximal, with ocular abnormalities, 604278

SLC51A	100%	100%	?Cholestasis, progressive familial intrahepatic, 6, 619484
SLC51B	100%	100%	?Bile acid malabsorption, primary, 2, 619481
SLC52A1	100%	100%	Riboflavin deficiency, 615026
SLC52A2	100%	100%	Brown-Vialetto-Van Laere syndrome 2, 614707
SLC52A3	100%	100%	?Fazio-Londe disease, 211500 Brown-Vialetto-Van Laere syndrome 1, 211530
SLC5A1	100%	100%	Glucose/galactose malabsorption, 606824
SLC5A2	100%	100%	Renal glucosuria, 233100
SLC5A5	100%	100%	Thyroid dyshormonogenesis 1, 274400
SLC5A6	100%	100%	Neurodegeneration, infantile-onset, biotin-responsive, 618973
SLC5A7	100%	100%	Neuronopathy, distal hereditary motor, type VIIA, 158580 Myasthenic syndrome, congenital, 20, presynaptic, 617143
SLC6A1	100%	100%	Myoclonic-atonic epilepsy, 616421
SLC6A17	100%	100%	Mental retardation, autosomal recessive 48, 616269
SLC6A19	100%	100%	Iminoglycinuria, digenic, 242600 Hartnup disorder, 234500 Hyperglycinuria, 138500
SLC6A2	100%	100%	?Orthostatic intolerance, 604715
SLC6A20	100%	100%	Iminoglycinuria, digenic, 242600 Hyperglycinuria, 138500
SLC6A3	100%	100%	Parkinsonism-dystonia, infantile, 1, 613135
SLC6A5	100%	100%	Hyperekplexia 3, 614618
SLC6A6	100%	100%	Hypotaurinemic retinal degeneration and cardiomyopathy, 145350
SLC6A8	100%	100%	Cerebral creatine deficiency syndrome 1, 300352
SLC6A9	100%	100%	Glycine encephalopathy with normal serum glycine, 617301
SLC7A14	100%	100%	Retinitis pigmentosa 68, 615725
SLC7A6OS	100%	100%	Epilepsy, progressive myoclonic, 12, 619191
SLC7A7	100%	100%	Lysinuric protein intolerance, 222700
SLC7A9	100%	100%	Cystinuria, 220100
SLC8B1	100%	100%	No OMIM disease ID
SLC9A1	100%	100%	Lichtenstein-Knorr syndrome, 616291

SLC9A3	99%	97%	Diarrhea 8, secretory sodium, congenital, 616868
SLC9A3R1	100%	100%	Nephrolithiasis/osteoporosis, hypophosphatemic, 2, 612287
SLC9A6	100%	99%	Intellectual developmental disorder, X-linked syndromic, Christianson type, 300243
SLC9A7	100%	100%	Intellectual developmental disorder, X-linked 108, 301024
SLCO1B1	100%	100%	Hyperbilirubinemia, Rotor type, digenic, 237450
SLCO1B3	100%	100%	Hyperbilirubinemia, Rotor type, digenic, 237450
SLCO2A1	100%	100%	Hypertrophic osteoarthropathy, primary, autosomal dominant, 167100 Hypertrophic osteoarthropathy, primary, autosomal recessive 2, 614441
SLCO5A1	100%	100%	No OMIM disease ID
SLFN14	100%	100%	Bleeding disorder, platelet-type, 20, 616913
SLIRP	100%	100%	No OMIM disease ID
SLIT3	100%	100%	No OMIM disease ID
SLITRK1	100%	100%	Tourette syndrome, 137580 ?Trichotillomania, 613229
SLITRK6	100%	100%	Deafness and myopia, 221200
SLMAP	100%	100%	No OMIM disease ID
SLURP1	100%	100%	Meleda disease, 248300
SLX4	100%	100%	Fanconi anemia, complementation group P, 613951
SMAD1	100%	100%	No OMIM disease ID
SMAD2	100%	100%	No OMIM disease ID
SMAD3	100%	100%	Loeys-Dietz syndrome 3, 613795
SMAD4	100%	100%	Pancreatic cancer, somatic, 260350 Myhre syndrome, 139210 Polyposis, juvenile intestinal, 174900 Juvenile polyposis/hereditary hemorrhagic telangiectasia syndrome, 175050
SMAD6	100%	100%	Aortic valve disease 2, 614823
SMAD9	100%	100%	Pulmonary hypertension, primary, 2, 615342
SMARCA1	100%	99%	No OMIM disease ID

SMARCA2	98%	97%	Nicolaides-Baraitser syndrome, 601358 Blepharophimosis-impaired intellectual development syndrome, 619293
SMARCA4	100%	100%	Coffin-Siris syndrome 4, 614609
SMARCA5	100%	100%	No OMIM disease ID
SMARCAD1	100%	100%	Basan syndrome, 129200 Huriez syndrome, 181600 Adermatoglyphia, 136000
SMARCAL1	100%	100%	Schimke immunoosseous dysplasia, 242900
SMARCB1	100%	100%	Rhabdoid tumors, somatic, 609322 Coffin-Siris syndrome 3, 614608
SMARCC2	100%	100%	Coffin-Siris syndrome 8, 618362
SMARCD1	100%	100%	Coffin-Siris syndrome 11, 618779
SMARCD2	100%	100%	Specific granule deficiency 2, 617475
SMARCE1	100%	100%	Coffin-Siris syndrome 5, 616938
SMC1A	100%	100%	Cornelia de Lange syndrome 2, 300590 Developmental and epileptic encephalopathy 85, with or without midline brain defects, 301044
SMC3	100%	100%	Cornelia de Lange syndrome 3, 610759
SMCHD1	100%	100%	Bosma arhinia microphthalmia syndrome, 603457 Fascioscapulohumeral muscular dystrophy 2, digenic, 158901
SMDT1	100%	100%	No OMIM disease ID
SMG8	100%	100%	Alzahrani-Kuwahara syndrome, 619268
SMG9	100%	100%	Heart and brain malformation syndrome, 616920
SMN1	94%	94%	Spinal muscular atrophy-2, 253550 Spinal muscular atrophy-4, 271150 Spinal muscular atrophy-3, 253400 Spinal muscular atrophy-1, 253300

SMO	100%	100%	Pallister-Hall-like syndrome, 241800 Basal cell carcinoma, somatic, 605462 Curry-Jones syndrome, somatic mosaic, 601707
SMOC1	100%	100%	Microphthalmia with limb anomalies, 206920
SMOC2	100%	100%	Dentin dysplasia, type I, with microdontia and misshapen teeth, 125400
SMPD1	100%	100%	Niemann-Pick disease, type B, 607616 Niemann-Pick disease, type A, 257200
SMPD4	100%	100%	Neurodevelopmental disorder with microcephaly, arthrogryposis, and structural brain anomalies, 618622
SMPX	100%	100%	Deafness, X-linked 4, 300066
SMS	100%	100%	Intellectual developmental disorder, X-linked syndromic, Snyder-Robinson type, 309583
SNAI2	100%	100%	Waardenburg syndrome, type 2D, 608890 Piebaldism, 172800
SNAP25	100%	100%	?Myasthenic syndrome, congenital, 18, 616330
SNAP29	100%	100%	Cerebral dysgenesis, neuropathy, ichthyosis, and palmoplantar keratoderma syndrome, 609528
SNCA	79%	79%	Dementia, Lewy body, 127750 Parkinson disease 1, 168601 Parkinson disease 4, 605543
SNCB	100%	100%	Dementia, Lewy body, 127750
SNIP1	100%	100%	Neurodevelopmental disorder with hypotonia, craniofacial abnormalities, and seizures, 614501
SNORA31	NC	NC	No OMIM disease ID
SNORD118	NC	NC	Leukoencephalopathy, brain calcifications, and cysts, 614561
SNRNP200	100%	100%	Retinitis pigmentosa 33, 610359
SNRPB	100%	100%	Cerebrocostomandibular syndrome, 117650
SNRPE	100%	100%	Hypotrichosis 11, 615059
SNRPN	100%	100%	Prader-Willi syndrome, 176270
SNTA1	100%	100%	Long QT syndrome 12, 612955
SNX10	100%	99%	Osteopetrosis, autosomal recessive 8, 615085
SNX14	100%	100%	Spinocerebellar ataxia, autosomal recessive 20, 616354
SNX27	100%	100%	No OMIM disease ID
SOBP	100%	99%	Mental retardation, anterior maxillary protrusion, and strabismus, 613671
SOCS1	100%	100%	Autoinflammatory syndrome, familial, with or without immunodeficiency, 619375
SOCS4	100%	100%	No OMIM disease ID

SOD1	100%	100%	Spastic tetraplegia and axial hypotonia, progressive, 618598 Amyotrophic lateral sclerosis 1, 105400
SOD2	100%	100%	No OMIM disease ID
SOHLH1	100%	100%	Ovarian dysgenesis 5, 617690 Spermatogenic failure 32, 618115
SON	100%	100%	ZTTK syndrome, 617140
SORD	98%	96%	Sorbitol dehydrogenase deficiency with peripheral neuropathy, 618912
SOS1	100%	100%	Noonan syndrome 4, 610733 ?Fibromatosis, gingival, 1, 135300
SOS2	100%	100%	Noonan syndrome 9, 616559
SOST	100%	100%	Sclerosteosis 1, 269500 Craniodiaphyseal dysplasia, autosomal dominant, 122860
SOX10	100%	100%	Waardenburg syndrome, type 4C, 613266 PCWH syndrome, 609136 Waardenburg syndrome, type 2E, with or without neurologic involvement, 611584
SOX11	100%	100%	Coffin-Siris syndrome 9, 615866
SOX17	100%	100%	Vesicoureteral reflux 3, 613674
SOX18	99%	98%	Hypotrichosis-lymphedema-telangiectasia syndrome, 607823 Hypotrichosis-lymphedema-telangiectasia-renal defect syndrome, 137940
SOX2	100%	100%	Optic nerve hypoplasia and abnormalities of the central nervous system, 206900 Microphthalmia, syndromic 3, 206900

SOX3	100%	100%	Intellectual developmental disorder, X-linked, with isolated growth hormone deficiency, 300123 Panhypopituitarism, X-linked, 312000
SOX4	100%	100%	Coffin-Siris syndrome 10, 618506
SOX5	100%	100%	Lamb-Shaffer syndrome, 616803
SOX6	100%	100%	Tolchin-Le Caignec syndrome, 618971
SOX9	100%	100%	Campomelic dysplasia with autosomal sex reversal, 114290 Acampomelic campomelic dysplasia, 114290 Campomelic dysplasia, 114290
SP110	100%	100%	Hepatic venoocclusive disease with immunodeficiency, 235550
SP7	100%	100%	Osteogenesis imperfecta, type XII, 613849
SPAG1	100%	100%	Ciliary dyskinesia, primary, 28, 615505
SPAG17	100%	100%	?Spermatogenic failure 55, 619380
SPAG6	100%	100%	No OMIM disease ID
SPARC	100%	100%	Osteogenesis imperfecta, type XVII, 616507
SPART	100%	100%	Troyer syndrome, 275900
SPAST	100%	100%	Spastic paraplegia 4, autosomal dominant, 182601
SPATA16	100%	100%	?Spermatogenic failure 6, 102530
SPATA5	100%	100%	Neurodevelopmental disorder with hearing loss, seizures, and brain abnormalities, 616577
SPATA7	100%	100%	Retinitis pigmentosa, juvenile, autosomal recessive, 604232 Leber congenital amaurosis 3, 604232
SPECC1L	97%	96%	Opitz GBBB syndrome, type II, 145410 Teebi hypertelorism syndrome, 145420 ?Facial clefting, oblique, 1, 600251
SPEF2	100%	100%	Spermatogenic failure 43, 618751
SPEG	99%	99%	Centronuclear myopathy 5, 615959
SPEN	100%	100%	Radio-Tartaglia syndrome, 619312

SPG11	100%	100%	Amyotrophic lateral sclerosis 5, juvenile, 602099 Charcot-Marie-Tooth disease, axonal, type 2X, 616668 Spastic paraplegia 11, autosomal recessive, 604360
SPG21	100%	100%	Mast syndrome, 248900
SPG7	100%	100%	Spastic paraplegia 7, autosomal recessive, 607259
SPINK1	100%	100%	Tropical calcific pancreatitis, 608189 Pancreatitis, hereditary, 167800
SPINK2	99%	99%	?Spermatogenic failure 29, 618091
SPINK5	100%	100%	Netherton syndrome, 256500
SPINT2	100%	100%	Diarrhea 3, secretory sodium, congenital, syndromic, 270420
SPNS2	99%	99%	?Deafness, autosomal recessive 115, 618457
SPO11	100%	100%	No OMIM disease ID
SPOCK1	100%	100%	No OMIM disease ID
SPOP	100%	100%	Nabais Sa-de Vries syndrome, type 1, 618828 Nabais Sa-de Vries syndrome, type 2, 618829
SPP2	100%	100%	No OMIM disease ID
SPPL2A	100%	100%	Immunodeficiency 86, mycobacteriosis, 619549
SPR	100%	100%	Dystonia, dopa-responsive, due to sepiapterin reductase deficiency, 612716
SPRED1	100%	100%	Legius syndrome, 611431
SPRED2	100%	100%	No OMIM disease ID
SPRTN	100%	100%	Ruijs-Aalfs syndrome, 616200
SPRY4	100%	100%	Hypogonadotropic hypogonadism 17 with or without anosmia, 615266
SPTA1	100%	100%	Spherocytosis, type 3, 270970 Elliptocytosis-2, 130600 Pyropoikilocytosis, 266140
SPTAN1	100%	100%	Developmental and epileptic encephalopathy 5, 613477
SPTB	100%	100%	Anemia, neonatal hemolytic, fatal or near-fatal, 617948 Elliptocytosis-3, 617948 Spherocytosis, type 2, 616649

SPTBN1	100%	100%	Developmental delay, impaired speech, and behavioral abnormalities, 619475
SPTBN2	100%	99%	Spinocerebellar ataxia 5, 600224 Spinocerebellar ataxia, autosomal recessive 14, 615386
SPTBN4	100%	100%	Neurodevelopmental disorder with hypotonia, neuropathy, and deafness, 617519
SPTLC1	100%	100%	Neuropathy, hereditary sensory and autonomic, type IA, 162400
SPTLC2	100%	100%	Neuropathy, hereditary sensory and autonomic, type IC, 613640
SPTLC3	100%	100%	No OMIM disease ID
SQOR	100%	100%	Sulfide:quinone oxidoreductase deficiency, 619221
SQSTM1	100%	100%	Neurodegeneration with ataxia, dystonia, and gaze palsy, childhood-onset, 617145 Frontotemporal dementia and/or amyotrophic lateral sclerosis 3, 616437 Myopathy, distal, with rimmed vacuoles, 617158 Paget disease of bone 3, 167250
SRC	100%	100%	?Thrombocytopenia 6, 616937 Colon cancer, advanced, somatic, 114500
SRCAP	100%	100%	Developmental delay, hypotonia, musculoskeletal defects, and behavioral abnormalities, 619595 Floating-Harbor syndrome, 136140
SRD5A2	100%	100%	Pseudovaginal perineoscrotal hypospadias, 264600
SRD5A3	100%	100%	Kahrizi syndrome, 612713 Congenital disorder of glycosylation, type Iq, 612379
SREBF1	96%	96%	Ichthyosis, follicular, with atrichia and photophobia syndrome 2, 619016 Mucoepithelial dysplasia, hereditary, 158310
SRF	100%	100%	No OMIM disease ID
SRI	100%	100%	No OMIM disease ID

SRP54	100%	100%	Neutropenia, severe congenital, 8, autosomal dominant, 618752
SRP72	100%	100%	Bone marrow failure syndrome 1, 614675
SRPK3	100%	100%	No OMIM disease ID
SRPX2	100%	100%	?Rolandic epilepsy, impaired intellectual development, and speech dyspraxia, 300643
SRRM2	100%	100%	No OMIM disease ID
SRY	50%	50%	46XY sex reversal 1, 400044
SSBP1	100%	100%	Optic atrophy 13 with retinal and foveal abnormalities, 165510
SSR4	100%	100%	Congenital disorder of glycosylation, type Iy, 300934
SSTR5	100%	100%	No OMIM disease ID
SSX1	100%	100%	?Sarcoma, synovial, 300813
SSX2	100%	100%	?Sarcoma, synovial, 300813
ST14	100%	100%	Ichthyosis, congenital, autosomal recessive 11, 602400
ST3GAL3	95%	95%	Developmental and epileptic encephalopathy 15, 615006 Intellectual developmental disorder, autosomal recessive 12, 611090
ST3GAL5	98%	98%	Salt and pepper developmental regression syndrome, 609056
STAB2	100%	100%	No OMIM disease ID
STAC3	100%	100%	Myopathy, congenital, Baily-Bloch, 255995
STAG1	100%	100%	Mental retardation, autosomal dominant 47, 617635
STAG2	100%	100%	Holoprosencephaly 13, X-linked, 301043 Mullegama-Klein-Martinez syndrome, 301022
STAG3	100%	100%	Premature ovarian failure 8, 615723
STAMBP	100%	100%	Microcephaly-capillary malformation syndrome, 614261
STAR	100%	100%	Lipoid adrenal hyperplasia, 201710
STARD7	100%	100%	Epilepsy, familial adult myoclonic, 2, 607876

STAT1	95%	95%	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%	Pseudo-TORCH syndrome 3, 618886 Immunodeficiency 44, 616636
STAT3	100%	100%	Hyper-IgE recurrent infection syndrome, 147060 Autoimmune disease, multisystem, infantile-onset, 1, 615952
STAT4	100%	100%	No OMIM disease ID
STAT5B	100%	100%	Growth hormone insensitivity with immune dysregulation 1, autosomal recessive, 245590 Growth hormone insensitivity with immune dysregulation 2, autosomal dominant, 618985 Leukemia, acute promyelocytic, somatic, 102578
STAT6	100%	100%	No OMIM disease ID
STEAP3	100%	100%	?Anemia, hypochromic microcytic, with iron overload 2, 615234
STEEP1	100%	100%	?Intellectual developmental disorder, X-linked 107, 301013
STIL	100%	100%	Microcephaly 7, primary, autosomal recessive, 612703
STIM1	100%	100%	Myopathy, tubular aggregate, 1, 160565 Stormorken syndrome, 185070 Immunodeficiency 10, 612783
STING1	100%	100%	STING-associated vasculopathy, infantile-onset, 615934

STK11	100%	100%	Melanoma, malignant, somatic, 155600 Pancreatic cancer, somatic, 260350 Peutz-Jeghers syndrome, 175200 Testicular tumor, somatic, 273300
STK36	100%	100%	?Ciliary dyskinesia, primary, 46, 619436
STK4	100%	100%	T-cell immunodeficiency, recurrent infections, autoimmunity, and cardiac malformations, 614868
STN1	100%	100%	Cerebroretinal microangiopathy with calcifications and cysts 2, 617341
STOX1	97%	95%	Preeclampsia/eclampsia 4, 609404
STRA6	100%	100%	Microphthalmia, syndromic 9, 601186 Microphthalmia, isolated, with coloboma 8, 601186
STRADA	100%	100%	Polyhydramnios, megalencephaly, and symptomatic epilepsy, 611087
STRC	100%	100%	Deafness, autosomal recessive 16, 603720
STS	97%	97%	Ichthyosis, X-linked, 308100
STT3A	100%	100%	Congenital disorder of glycosylation, type Iw, 615596
STT3B	100%	100%	?Congenital disorder of glycosylation, type Ix, 615597
STUB1	100%	100%	Spinocerebellar ataxia 48, 618093 Spinocerebellar ataxia, autosomal recessive 16, 615768
STX11	100%	100%	Hemophagocytic lymphohistiocytosis, familial, 4, 603552
STX16	100%	100%	Pseudohypoparathyroidism, type IB, 603233
STX1B	100%	100%	Generalized epilepsy with febrile seizures plus, type 9, 616172
STX3	100%	100%	Retinal dystrophy and microvillus inclusion disease, 619446 Diarrhea 12, with microvillus atrophy, 619445
STX5	100%	100%	No OMIM disease ID
STXBP1	100%	100%	Developmental and epileptic encephalopathy 4, 612164
STXBP2	99%	99%	Hemophagocytic lymphohistiocytosis, familial, 5, with or without microvillus inclusion disease, 613101
SUCLA2	100%	99%	Mitochondrial DNA depletion syndrome 5 (encephalomyopathic with or without methylmalonic aciduria), 612073
SUCLG1	100%	100%	Mitochondrial DNA depletion syndrome 9 (encephalomyopathic type with methylmalonic aciduria), 245400
SUCLG2	100%	100%	No OMIM disease ID

SUFU	100%	100%	Joubert syndrome 32, 617757 Medulloblastoma, desmoplastic, 155255 Basal cell nevus syndrome, 109400
SUGCT	100%	100%	Glutaric aciduria III, 231690
SULF1	100%	100%	No OMIM disease ID
SULT2B1	100%	100%	Ichthyosis, congenital, autosomal recessive 14, 617571
SUMF1	100%	100%	Multiple sulfatase deficiency, 272200
SUMO1	69%	69%	?Orofacial cleft 10, 613705
SUN5	100%	100%	Spermatogenic failure 16, 617187
SUOX	100%	100%	Sulfite oxidase deficiency, 272300
SUPT16H	100%	100%	Neurodevelopmental disorder with dysmorphic facies and thin corpus callosum, 619480
SURF1	100%	100%	Charcot-Marie-Tooth disease, type 4K, 616684 Mitochondrial complex IV deficiency, nuclear type 1, 220110
SUZ12	100%	100%	Imagawa-Matsumoto syndrome, 618786
SVBP	100%	100%	Neurodevelopmental disorder with ataxia, hypotonia, and microcephaly, 618569
SVIL	100%	100%	Myofibrillar myopathy 10, 619040
SYCE1	100%	100%	?Spermatogenic failure 15, 616950 ?Premature ovarian failure 12, 616947
SYCP2	100%	100%	Spermatogenic failure 1, 258150
SYCP3	100%	100%	Pregnancy loss, recurrent, 4, 270960 Spermatogenic failure 4, 270960
SYK	100%	100%	Immunodeficiency 82 with systemic inflammation, 619381
SYN1	100%	100%	Intellectual developmental disorder, X-linked 50, 300115 Epilepsy, X-linked, with variable learning disabilities and behavior disorders, 300491
SYNCRIP	100%	100%	No OMIM disease ID

SYNE1	98%	98%	Arthrogryposis multiplex congenita 3, myogenic type, 618484 Emery-Dreifuss muscular dystrophy 4, autosomal dominant, 612998 Spinocerebellar ataxia, autosomal recessive 8, 610743
SYNE2	100%	100%	Emery-Dreifuss muscular dystrophy 5, autosomal dominant, 612999
SYNE4	100%	100%	Deafness, autosomal recessive 76, 615540
SYNGAP1	100%	100%	Mental retardation, autosomal dominant 5, 612621
SYNJ1	100%	100%	Parkinson disease 20, early-onset, 615530 Developmental and epileptic encephalopathy 53, 617389
SYP	100%	100%	Intellectual developmental disorder, X-linked 96, 300802
SYT1	100%	100%	Baker-Gordon syndrome, 618218
SYT14	100%	100%	?Spinocerebellar ataxia, autosomal recessive 11, 614229
SYT2	100%	100%	Myasthenic syndrome, congenital, 7A, presynaptic, and distal motor neuropathy, autosomal dominant, 616040 Myasthenic syndrome, congenital, 7B, presynaptic, autosomal recessive, 619461
SZT2	100%	100%	Developmental and epileptic encephalopathy 18, 615476
TAB2	100%	100%	Congenital heart defects, nonsyndromic, 2, 614980
TAC3	100%	100%	Hypogonadotropic hypogonadism 10 with or without anosmia, 614839
TACO1	100%	100%	Mitochondrial complex IV deficiency, nuclear type 8, 619052
TACR3	100%	100%	Hypogonadotropic hypogonadism 11 with or without anosmia, 614840
TACSTD2	100%	100%	Corneal dystrophy, gelatinous drop-like, 204870
TAF1	100%	100%	Intellectual developmental disorder, X-linked syndromic 33, 300966 Dystonia-Parkinsonism, X-linked, 314250
TAF13	100%	100%	Mental retardation, autosomal recessive 60, 617432
TAF1C	100%	100%	No OMIM disease ID

TAF2	100%	100%	Mental retardation, autosomal recessive 40, 615599
TAF4B	100%	100%	?Spermatogenic failure 13, 615841
TAF6	100%	100%	Alazami-Yuan syndrome, 617126
TAFAZZIN	100%	100%	Barth syndrome, 302060
TAL1	100%	100%	Leukemia, T-cell acute lymphocytic, somatic, 613065
TAL2	100%	100%	Leukemia, T-cell acute lymphocytic, somatic, 613065
TALDO1	100%	100%	Transaldolase deficiency, 606003
TANC2	100%	100%	Intellectual developmental disorder with autistic features and language delay, with or without seizures, 618906
TANGO2	100%	100%	Metabolic encephalomyopathic crises, recurrent, with rhabdomyolysis, cardiac arrhythmias, and neurodegeneration, 616878
TAOK1	100%	100%	Developmental delay with or without intellectual impairment or behavioral abnormalities, 619575
TAP1	100%	100%	Bare lymphocyte syndrome, type I, 604571
TAP2	100%	100%	Bare lymphocyte syndrome, type I, due to TAP2 deficiency, 604571
TAPBP	96%	96%	Bare lymphocyte syndrome, type I, 604571
TAPT1	100%	100%	Osteochondrodysplasia, complex lethal, Symoens-Barnes-Gistelinck type, 616897
TARDBP	100%	100%	Frontotemporal lobar degeneration, TARDBP-related, 612069 Amyotrophic lateral sclerosis 10, with or without FTD, 612069
TARS1	100%	100%	Trichothiodystrophy 7, nonphotosensitive, 618546
TARS2	100%	100%	?Combined oxidative phosphorylation deficiency 21, 615918
TASP1	100%	100%	Suleiman-El-Hattab syndrome, 618950
TAT	100%	100%	Tyrosinemia, type II, 276600
TAX1BP3	100%	100%	No OMIM disease ID
TBC1D20	100%	100%	Warburg micro syndrome 4, 615663
TBC1D23	100%	100%	Pontocerebellar hypoplasia, type 11, 617695

TBC1D24	100%	100%	Deafness, autosomal recessive 86, 614617 Epilepsy, rolandic, with proxysmal exercise-induce dystonia and writer's cramp, 608105 Myoclonic epilepsy, infantile, familial, 605021 Deafness, autosomal dominant 65, 616044 Developmental and epileptic encephalopathy 16, 615338 DOORS syndrome, 220500
TBC1D2B	99%	99%	Neurodevelopmental disorder with seizures and gingival overgrowth, 619323
TBC1D32	100%	100%	No OMIM disease ID
TBC1D7	100%	100%	Macrocephaly/megalencephaly syndrome, autosomal recessive, 248000
TBC1D8B	100%	100%	Nephrotic syndrome, type 20, 301028
TBCD	100%	100%	Encephalopathy, progressive, early-onset, with brain atrophy and thin corpus callosum, 617193
TBCE	100%	100%	Kenny-Caffey syndrome, type 1, 244460 Hypoparathyroidism-retardation-dysmorphism syndrome, 241410 Encephalopathy, progressive, with amyotrophy and optic atrophy, 617207
TBCK	100%	100%	Hypotonia, infantile, with psychomotor retardation and characteristic facies 3, 616900
TBK1	100%	100%	Frontotemporal dementia and/or amyotrophic lateral sclerosis 4, 616439
TBL1X	100%	100%	Hypothyroidism, congenital, nongoitrous, 8, 301033
TBL1XR1	100%	100%	Pierpont syndrome, 602342 Mental retardation, autosomal dominant 41, 616944
TBL1Y	50%	49%	?Deafness, Y-linked 2, 400047
TBP	100%	100%	Spinocerebellar ataxia 17, 607136
TBR1	100%	100%	Intellectual developmental disorder with autism and speech delay, 606053

TBX1	97%	94%	Tetralogy of Fallot, 187500 DiGeorge syndrome, 188400 Conotruncal anomaly face syndrome, 217095 Velocardiofacial syndrome, 192430
TBX15	100%	100%	Cousin syndrome, 260660
TBX18	100%	100%	Congenital anomalies of kidney and urinary tract 2, 143400
TBX19	100%	100%	Adrenocorticotropic hormone deficiency, 201400
TBX2	100%	99%	Vertebral anomalies and variable endocrine and T-cell dysfunction, 618223
TBX20	100%	100%	Atrial septal defect 4, 611363
TBX21	100%	100%	Asthma and nasal polyps, 208550 ?Immunodeficiency 88, 619630
TBX22	100%	100%	Cleft palate with ankyloglossia, 303400 ?Abruzzo-Erickson syndrome, 302905
TBX3	100%	100%	Ulnar-mammary syndrome, 181450
TBX4	100%	100%	Ischiocoxopodopatellar syndrome with or without pulmonary arterial hypertension, 147891 Amelia, posterior, with pelvic and pulmonary hypoplasia syndrome, 601360
TBX5	100%	100%	Holt-Oram syndrome, 142900
TBX6	100%	100%	Spondylocostal dysostosis 5, 122600
TBXA2R	100%	99%	No OMIM disease ID
TBXAS1	100%	100%	Ghosal hematodiaphyseal syndrome, 231095
TBXT	100%	100%	Sacral agenesis with vertebral anomalies, 615709
TCAP	100%	100%	Cardiomyopathy, hypertrophic, 25, 607487 Muscular dystrophy, limb-girdle, autosomal recessive 7, 601954
TCF12	100%	100%	Craniosynostosis 3, 615314
TCF20	100%	100%	Developmental delay with variable intellectual impairment and behavioral abnormalities, 618430
TCF3	100%	100%	Agammaglobulinemia 8, autosomal dominant, 616941

TCF4	100%	100%	Pitt-Hopkins syndrome, 610954 Corneal dystrophy, Fuchs endothelial, 3, 613267
TCF7L2	100%	100%	No OMIM disease ID
TCHH	100%	100%	?Uncombable hair syndrome 3, 617252
TCIRG1	100%	100%	Osteopetrosis, autosomal recessive 1, 259700
TCN2	100%	100%	Transcobalamin II deficiency, 275350
TCOF1	100%	100%	Treacher Collins syndrome 1, 154500
TCTN1	95%	94%	Joubert syndrome 13, 614173
TCTN2	100%	100%	Joubert syndrome 24, 616654 ?Meckel syndrome 8, 613885
TCTN3	100%	100%	Joubert syndrome 18, 614815 Orofaciodigital syndrome IV, 258860
TDGF1	100%	100%	Forebrain defects,
TDP1	100%	100%	?Spinocerebellar ataxia, autosomal recessive, with axonal neuropathy 1, 607250
TDP2	100%	100%	Spinocerebellar ataxia, autosomal recessive 23, 616949
TDRD7	100%	100%	Cataract 36, 613887
TDRD9	100%	100%	?Spermatogenic failure 30, 618110
TDRKH	100%	100%	No OMIM disease ID
TEAD1	100%	100%	Sveinsson chorioretinal atrophy, 108985
TECPR2	100%	100%	Neuropathy, hereditary sensory and autonomic, type IX, with developmental delay, 615031
TECR	100%	100%	Mental retardation, autosomal recessive 14, 614020
TECRL	100%	100%	Ventricular tachycardia, catecholaminergic polymorphic, 3, 614021
TECTA	100%	100%	Deafness, autosomal dominant 8/12, 601543 Deafness, autosomal recessive 21, 603629
TEK	100%	100%	Venous malformations, multiple cutaneous and mucosal, 600195 Glaucoma 3, primary congenital, E, 617272
TELO2	100%	100%	You-Hoover-Fong syndrome, 616954

TENM3	100%	100%	Microphthalmia, syndromic 15, 615145 ?Microphthalmia, isolated, with coloboma 9, 615145
TENM4	100%	100%	Essential tremor, hereditary, 5, 616736
TENT5A	100%	100%	Osteogenesis imperfecta, type XVIII, 617952
TERB1	100%	100%	Spermatogenic failure 60, 619646
TERB2	100%	100%	?Spermatogenic failure 59, 619645
TERC	NC	NC	Dyskeratosis congenita, autosomal dominant 1, 127550
TERF1	100%	100%	No OMIM disease ID
TERF2	100%	100%	No OMIM disease ID
TERF2IP	83%	83%	No OMIM disease ID
TERT	100%	100%	Dyskeratosis congenita, autosomal dominant 2, 613989 Dyskeratosis congenita, autosomal recessive 4, 613989 Pulmonary fibrosis and/or bone marrow failure, telomere-related, 1, 614742
TES	100%	100%	No OMIM disease ID
TET2	100%	100%	Myelodysplastic syndrome, somatic, 614286 Immunodeficiency 75, 619126
TET3	100%	100%	Beck-Fahrner syndrome, 618798
TEX11	97%	97%	Spermatogenic failure, X-linked, 2, 309120
TEX14	100%	100%	Spermatogenic failure 23, 617707
TEX15	100%	100%	Spermatogenic failure 25, 617960
TF	100%	100%	Atransferrinemia, 209300
TFAM	100%	100%	?Mitochondrial DNA depletion syndrome 15 (hepatocerebral type), 617156
TFAP2A	100%	100%	Branchiooculofacial syndrome, 113620
TFAP2B	100%	100%	Patent ductus arteriosus 2, 617035 Char syndrome, 169100
TFB2M	100%	100%	No OMIM disease ID

TFE3	100%	100%	Intellectual developmental disorder, X-linked, syndromic, with pigmentary mosaicism and coarse facies, 301066 Renal cell carcinoma, papillary, 1, 300854
TFG	100%	100%	?Spastic paraplegia 57, autosomal recessive, 615658 Hereditary motor and sensory neuropathy, Okinawa type, 604484
TFPT	100%	100%	No OMIM disease ID
TFR2	100%	100%	Hemochromatosis, type 3, 604250
TFRC	100%	100%	Immunodeficiency 46, 616740
TG	100%	100%	Thyroid dyshormonogenesis 3, 274700
TGDS	100%	100%	Catel-Manzke syndrome, 616145
TGFB1	100%	100%	Inflammatory bowel disease, immunodeficiency, and encephalopathy, 618213 Camurati-Engelmann disease, 131300
TGFB2	100%	100%	Loeys-Dietz syndrome 4, 614816
TGFB3	100%	100%	Arrhythmogenic right ventricular dysplasia 1, 107970 Loeys-Dietz syndrome 5, 615582

TGFBI	100%	100%	Corneal dystrophy, Avellino type, 607541 Corneal dystrophy, Reis-Bucklers type, 608470 Corneal dystrophy, Thiel-Behnke type, 602082 Corneal dystrophy, Groenouw type I, 121900 Corneal dystrophy, epithelial basement membrane, 121820 Corneal dystrophy, lattice type I, 122200 Corneal dystrophy, lattice type IIIA, 608471
TGFBR1	100%	99%	Loeys-Dietz syndrome 1, 609192
TGFBR2	100%	100%	Loeys-Dietz syndrome 2, 610168 Colorectal cancer, hereditary nonpolyposis, type 6, 614331 Esophageal cancer, somatic, 133239
TGIF1	100%	100%	Holoprosencephaly 4, 142946
TGM1	100%	100%	Ichthyosis, congenital, autosomal recessive 1, 242300
TGM3	100%	100%	?Uncombable hair syndrome 2, 617251
TGM5	100%	100%	Peeling skin syndrome 2, 609796
TGM6	100%	100%	Spinocerebellar ataxia 35, 613908
TH	100%	100%	Segawa syndrome, recessive, 605407
THAP1	100%	100%	Dystonia 6, torsion, 602629
THBD	100%	100%	Thrombophilia due to thrombomodulin defect, 614486
THBS4	100%	100%	No OMIM disease ID
THG1L	100%	100%	Spinocerebellar ataxia, autosomal recessive 28, 618800
THOC1	100%	100%	No OMIM disease ID
THOC2	100%	100%	Intellectual developmental disorder, X-linked 12, 300957
THOC6	100%	100%	Beaulieu-Boycott-Innes syndrome, 613680
THPO	100%	100%	Thrombocythemia 1, 187950
THRA	100%	100%	Hypothyroidism, congenital, nongoitrous, 6, 614450

THRB	100%	100%	Thyroid hormone resistance, autosomal recessive, 274300 Thyroid hormone resistance, 188570 Thyroid hormone resistance, selective pituitary, 145650
THSD1	100%	100%	?Aneurysm, intracranial berry, 12, 618734
THSD4	100%	100%	No OMIM disease ID
TIA1	100%	100%	Welander distal myopathy, 604454 Amyotrophic lateral sclerosis 26 with or without frontotemporal dementia, 619133
TICAM1	100%	100%	No OMIM disease ID
TIE1	100%	100%	Lymphatic malformation 11, 619401
TIMM22	100%	100%	?Combined oxidative phosphorylation deficiency 43, 618851
TIMM44	100%	100%	No OMIM disease ID
TIMM50	100%	100%	3-methylglutaconic aciduria, type IX, 617698
TIMM8A	100%	100%	Mohr-Tranebjaerg syndrome, 304700
TIMMDC1	100%	100%	Mitochondrial complex I deficiency, nuclear type 31, 618251
TIMP3	100%	100%	Sorsby fundus dystrophy, 136900
TINF2	100%	100%	Dyskeratosis congenita, autosomal dominant 3, 613990 Revesz syndrome, 268130
TIRAP	100%	100%	No OMIM disease ID
TJP1	100%	100%	No OMIM disease ID
TJP2	98%	98%	Hypercholanemia, familial 1, 607748 Cholestasis, progressive familial intrahepatic 4, 615878
TK2	100%	100%	Mitochondrial DNA depletion syndrome 2 (myopathic type), 609560 ?Progressive external ophthalmoplegia with mitochondrial DNA deletions, autosomal recessive 3, 617069
TKFC	100%	100%	Triokinase and FMN cyclase deficiency syndrome, 618805
TKT	98%	98%	Short stature, developmental delay, and congenital heart defects, 617044

TLCD3B	100%	100%	Cone-rod dystrophy 22, 619531
TLE6	100%	100%	Preimplantation embryonic lethality, 616814
TLK2	100%	100%	Mental retardation, autosomal dominant 57, 618050
TLL1	100%	100%	Atrial septal defect 6, 613087
TLN1	100%	100%	No OMIM disease ID
TLR3	100%	100%	No OMIM disease ID
TLR4	100%	100%	No OMIM disease ID
TLR5	100%	100%	No OMIM disease ID
TLR7	100%	100%	Immunodeficiency 74, COVID19-related, X-linked, 301051
TLR8	100%	100%	No OMIM disease ID
TMC1	100%	100%	Deafness, autosomal dominant 36, 606705 Deafness, autosomal recessive 7, 600974
TMC6	100%	100%	Epidermodysplasia verruciformis, 226400
TMC8	100%	100%	Epidermodysplasia verruciformis 2, 618231
TMCO1	88%	88%	Craniofacial dysmorphism, skeletal anomalies, and mental retardation syndrome, 213980
TMCO3	100%	100%	No OMIM disease ID
TMEM106B	100%	100%	Leukodystrophy, hypomyelinating, 16, 617964
TMEM107	100%	100%	Orofaciodigital syndrome XVI, 617563 Meckel syndrome 13, 617562 ?Joubert syndrome 29, 617562
TMEM126A	100%	100%	Optic atrophy 7, 612989
TMEM126B	100%	100%	Mitochondrial complex I deficiency, nuclear type 29, 618250
TMEM127	100%	100%	No OMIM disease ID
TMEM132E	100%	100%	Deafness, autosomal recessive 99, 618481
TMEM138	100%	100%	Joubert syndrome 16, 614465
TMEM14C	100%	100%	No OMIM disease ID
TMEM165	100%	100%	Congenital disorder of glycosylation, type IIk, 614727
TMEM186	100%	100%	No OMIM disease ID
TMEM199	100%	100%	Congenital disorder of glycosylation, type IIp, 616829
TMEM216	100%	100%	Joubert syndrome 2, 608091 Meckel syndrome 2, 603194
TMEM218	100%	100%	Joubert syndrome 39, 619562

TMEM222	100%	100%	Neurodevelopmental disorder with motor and speech delay and behavioral abnormalities, 619470
TMEM231	100%	100%	Joubert syndrome 20, 614970 Meckel syndrome 11, 615397
TMEM237	100%	100%	Joubert syndrome 14, 614424
TMEM240	100%	100%	Spinocerebellar ataxia 21, 607454
TMEM251	100%	100%	Dysostosis multiplex, Ain-Naz type, 619345
TMEM260	100%	100%	Structural heart defects and renal anomalies syndrome, 617478
TMEM38B	100%	100%	Osteogenesis imperfecta, type XIV, 615066
TMEM43	100%	100%	Arrhythmogenic right ventricular dysplasia 5, 604400 Emery-Dreifuss muscular dystrophy 7, AD, 614302
TMEM63A	100%	100%	Leukodystrophy, hypomyelinating, 19, transient infantile, 618688
TMEM65	99%	95%	No OMIM disease ID
TMEM67	100%	99%	Nephronophthisis 11, 613550 Joubert syndrome 6, 610688 Meckel syndrome 3, 607361 ?RHYNS syndrome, 602152 COACH syndrome 1, 216360
TMEM70	100%	100%	Mitochondrial complex V (ATP synthase) deficiency, nuclear type 2, 614052
TMEM94	100%	100%	Intellectual developmental disorder with cardiac defects and dysmorphic facies, 618316
TMEM98	100%	100%	Nanophthalmos 4, 615972
TMIE	100%	100%	Deafness, autosomal recessive 6, 600971
TMLHE	100%	99%	No OMIM disease ID
TMPO	100%	100%	No OMIM disease ID
TMPRSS15	100%	100%	Enterokinase deficiency, 226200
TMPRSS3	100%	100%	Deafness, autosomal recessive 8/10, 601072
TMPRSS6	100%	100%	Iron-refractory iron deficiency anemia, 206200
TMTC2	97%	97%	No OMIM disease ID
TMTC3	100%	100%	Lissencephaly 8, 617255
TMX2	100%	100%	Neurodevelopmental disorder with microcephaly, cortical malformations, and spasticity, 618730
TNC	100%	100%	Deafness, autosomal dominant 56, 615629
TNFAIP3	100%	100%	Autoinflammatory syndrome, familial, Behcet-like, 616744
TNFRSF10B	100%	100%	Squamous cell carcinoma, head and neck, 275355

TNFRSF11A	99%	99%	Osteopetrosis, autosomal recessive 7, 612301 Osteolysis, familial expansile, 174810
TNFRSF11B	100%	100%	Paget disease of bone 5, juvenile-onset, 239000
TNFRSF13B	100%	100%	Immunodeficiency, common variable, 2, 240500 Immunoglobulin A deficiency 2, 609529
TNFRSF13C	100%	100%	Immunodeficiency, common variable, 4, 613494
TNFRSF1A	92%	92%	Periodic fever, familial, 142680
TNFRSF4	100%	100%	?Immunodeficiency 16, 615593
TNFRSF9	100%	100%	No OMIM disease ID
TNFSF11	100%	100%	Osteopetrosis, autosomal recessive 2, 259710
TNFSF12	100%	100%	No OMIM disease ID
TNFSF13	100%	100%	No OMIM disease ID
TNIK	100%	100%	Intellectual developmental disorder 54, 617028
TNNC1	100%	100%	Cardiomyopathy, dilated, 1Z, 611879 Cardiomyopathy, hypertrophic, 13, 613243
TNNI2	100%	100%	Arthrogryposis, distal, type 2B1, 601680
TNNI3	100%	100%	?Cardiomyopathy, dilated, 2A, 611880 Cardiomyopathy, hypertrophic, 7, 613690 Cardiomyopathy, familial restrictive, 1, 115210 Cardiomyopathy, dilated, 1FF, 613286
TNNI3K	100%	100%	Cardiac conduction disease with or without dilated cardiomyopathy, 616117
TNNT1	100%	100%	Nemaline myopathy 5, Amish type, 605355
TNNT2	100%	100%	Cardiomyopathy, dilated, 1D, 601494 Cardiomyopathy, hypertrophic, 2, 115195 Cardiomyopathy, familial restrictive, 3, 612422 Left ventricular noncompaction 6, 601494

TNNT3	100%	100%	Arthrogryposis, distal, type 2B2, 618435
TNPO2	100%	100%	Intellectual developmental disorder with hypotonia, impaired speech, and dysmorphic facies, 619556
TNPO3	100%	100%	Muscular dystrophy, limb-girdle, autosomal dominant 2, 608423
TNR	100%	100%	No OMIM disease ID
TNRC6A	100%	100%	?Epilepsy, familial adult myoclonic, 6, 618074
TNRC6B	100%	100%	Global developmental delay with speech and behavioral abnormalities, 619243
TNS2	100%	100%	No OMIM disease ID
TNXB	100%	100%	Ehlers-Danlos syndrome, classic-like, 1, 606408 Vesicoureteral reflux 8, 615963
TOE1	100%	100%	Pontocerebellar hypoplasia, type 7, 614969
TOGARAM1	100%	100%	Joubert syndrome 37, 619185
TOM1	100%	100%	?Immunodeficiency 85 and autoimmunity, 619510
TOMM70	100%	100%	No OMIM disease ID
TONSL	100%	100%	Spondyloepimetaphyseal dysplasia, sponastrime type, 271510
TOP1	100%	100%	DNA topoisomerase I, camptothecin-resistant,
TOP2A	100%	100%	DNA topoisomerase II, resistance to inhibition of, by amsacrine,
TOP2B	100%	100%	B-cell immunodeficiency, distal limb anomalies, and urogenital malformations, 609296
TOP3A	100%	100%	?Progressive external ophthalmoplegia with mitochondrial DNA deletions, autosomal recessive 5, 618098 Microcephaly, growth restriction, and increased sister chromatid exchange 2, 618097
TOPORS	100%	100%	Retinitis pigmentosa 31, 609923
TOR1A	94%	91%	Arthrogryposis multiplex congenita 5, 618947 Dystonia-1, torsion, 128100
TOR1AIP1	100%	100%	?Muscular dystrophy, autosomal recessive, with rigid spine and distal joint contractures, 617072

TP53	91%	91%	Hepatocellular carcinoma, somatic, 114550 Breast cancer, somatic, 114480 Li-Fraumeni syndrome, 151623 Pancreatic cancer, somatic, 260350 Nasopharyngeal carcinoma, somatic, 607107 Bone marrow failure syndrome 5, 618165
TP53RK	100%	100%	Galloway-Mowat syndrome 4, 617730
TP63	100%	100%	Ectrodactyly, ectodermal dysplasia, and cleft lip/palate syndrome 3, 604292 Hay-Wells syndrome, 106260 Split-hand/foot malformation 4, 605289 Orofacial cleft 8, 618149 Rapp-Hodgkin syndrome, 129400 ADULT syndrome, 103285 Limb-mammary syndrome, 603543
TP73	100%	100%	Ciliary dyskinesia, primary, 47, and lissencephaly, 619466
TPCN2	100%	100%	No OMIM disease ID
TPI1	100%	100%	Hemolytic anemia due to triosephosphate isomerase deficiency, 615512
TPK1	100%	100%	Thiamine metabolism dysfunction syndrome 5 (episodic encephalopathy type), 614458
TPM1	100%	99%	Left ventricular noncompaction 9, 611878 Cardiomyopathy, hypertrophic, 3, 115196 Cardiomyopathy, dilated, 1Y, 611878

TPM2	100%	100%	Arthrogryposis, distal, type 2B4, 108120 Arthrogryposis, distal, type 1A, 108120 Nemaline myopathy 4, autosomal dominant, 609285 CAP myopathy 2, 609285
TPM3	100%	100%	CAP myopathy 1, 609284 Myopathy, congenital, with fiber-type disproportion, 255310 Nemaline myopathy 1, autosomal dominant or recessive, 609284
TPM4	100%	100%	No OMIM disease ID
TPMT	100%	100%	No OMIM disease ID
TPO	100%	100%	Thyroid dyshormonogenesis 2A, 274500
TPP1	100%	100%	Ceroid lipofuscinosis, neuronal, 2, 204500 Spinocerebellar ataxia, autosomal recessive 7, 609270
TPP2	100%	100%	Immunodeficiency 78 with autoimmunity and developmental delay, 619220
TPRKB	82%	81%	Galloway-Mowat syndrome 5, 617731
TPRN	97%	95%	Deafness, autosomal recessive 79, 613307
TRAC	100%	100%	Immunodeficiency 7, TCR-alpha/beta deficient, 615387
TRAF3	100%	100%	No OMIM disease ID
TRAF3IP1	100%	100%	Senior-Loken syndrome 9, 616629
TRAF3IP2	100%	100%	?Candidiasis, familial, 8, 615527
TRAF6	100%	100%	No OMIM disease ID
TRAF7	100%	100%	Cardiac, facial, and digital anomalies with developmental delay, 618164
TRAIP	100%	100%	Seckel syndrome 9, 616777
TRAK1	100%	100%	Developmental and epileptic encephalopathy 68, 618201
TRAPPC11	100%	100%	Muscular dystrophy, limb-girdle, autosomal recessive 18, 615356
TRAPPC12	100%	100%	Encephalopathy, progressive, early-onset, with brain atrophy and spasticity, 617669
TRAPPC14	100%	100%	?Microcephaly 25, primary, autosomal recessive, 618351
TRAPPC2	100%	100%	Spondyloepiphyseal dysplasia tarda, 313400
TRAPPC2L	100%	100%	Encephalopathy, progressive, early-onset, with episodic rhabdomyolysis, 618331

TRAPPC4	100%	100%	Neurodevelopmental disorder with epilepsy, spasticity, and brain atrophy, 618741
TRAPPC6B	100%	100%	Neurodevelopmental disorder with microcephaly, epilepsy, and brain atrophy, 617862
TRAPPC9	100%	100%	Mental retardation, autosomal recessive 13, 613192
TRDN	100%	100%	Cardiac arrhythmia syndrome, with or without skeletal muscle weakness, 615441
TREH	100%	100%	Trehalase deficiency, 612119
TREM2	100%	100%	Polycystic lipomembranous osteodysplasia with sclerosing leukoencephalopathy 2, 618193
TREX1	100%	100%	Vasculopathy, retinal, with cerebral leukoencephalopathy and systemic manifestations, 192315 Aicardi-Goutieres syndrome 1, dominant and recessive, 225750 Chilblain lupus, 610448
TRH	100%	100%	No OMIM disease ID
TRHR	100%	100%	Hypothyroidism, congenital, nongoitrous, 7, 618573
TRIM2	93%	93%	Charcot-Marie-Tooth disease, type 2R, 615490
TRIM22	100%	100%	No OMIM disease ID
TRIM28	100%	100%	No OMIM disease ID
TRIM32	100%	100%	?Bardet-Biedl syndrome 11, 615988 Muscular dystrophy, limb-girdle, autosomal recessive 8, 254110
TRIM36	100%	100%	?Anencephaly 1, 206500
TRIM37	98%	98%	Mulibrey nanism, 253250
TRIM44	100%	100%	?Aniridia 3, 617142
TRIM63	100%	100%	No OMIM disease ID
TRIM71	100%	100%	Hydrocephalus, congenital communicating, 1, 618667
TRIM8	100%	100%	Focal segmental glomerulosclerosis and neurodevelopmental syndrome, 619428
TRIO	99%	99%	Intellectual developmental disorder, autosomal dominant 44, with microcephaly, 617061 Intellectual developmental disorder, autosomal dominant 63, with macrocephaly, 618825
TRIOBP	100%	100%	Deafness, autosomal recessive 28, 609823
TRIP11	100%	100%	Odontochondrodysplasia 1, 184260 Achondrogenesis, type IA, 200600

TRIP12	100%	100%	Mental retardation, autosomal dominant 49, 617752
TRIP13	100%	100%	Oocyte maturation defect 9, 619011 Mosaic variegated aneuploidy syndrome 3, 617598
TRIP4	100%	100%	?Muscular dystrophy, congenital, Davignon-Chauveau type, 617066 Spinal muscular atrophy with congenital bone fractures 1, 616866
TRIT1	100%	100%	Combined oxidative phosphorylation deficiency 35, 617873
TRMT1	100%	100%	Intellectual developmental disorder, autosomal recessive 68, 618302
TRMT10A	100%	100%	Microcephaly, short stature, and impaired glucose metabolism 1, 616033
TRMT10C	100%	100%	Combined oxidative phosphorylation deficiency 30, 616974
TRMT5	100%	100%	Combined oxidative phosphorylation deficiency 26, 616539
TRMU	100%	100%	Liver failure, transient infantile, 613070
TRNT1	100%	100%	Sideroblastic anemia with B-cell immunodeficiency, periodic fevers, and developmental delay, 616084 Retinitis pigmentosa and erythrocytic microcytosis, 616959
TRPA1	100%	100%	?Episodic pain syndrome, familial, 1, 615040
TRPC3	100%	100%	?Spinocerebellar ataxia 41, 616410
TRPC6	100%	100%	Glomerulosclerosis, focal segmental, 2, 603965
TRPM1	100%	100%	Night blindness, congenital stationary (complete), 1C, autosomal recessive, 613216
TRPM3	100%	100%	No OMIM disease ID
TRPM4	100%	100%	Progressive familial heart block, type IB, 604559 Erythrokeratodermia veriabilis et progressiva 6, 618531
TRPM6	100%	100%	Hypomagnesemia 1, intestinal, 602014
TRPM8	100%	100%	No OMIM disease ID
TRPS1	100%	100%	Trichorhinophalangeal syndrome, type III, 190351 Trichorhinophalangeal syndrome, type I, 190350
TRPV1	100%	100%	No OMIM disease ID

TRPV3	97%	97%	?Palmoplantar keratoderma, nonepidermolytic, focal 2, 616400 Olmsted syndrome 1, 614594
TRPV4	100%	100%	Spondylometaphyseal dysplasia, Kozlowski type, 184252 Digital arthropathy-brachydactyly, familial, 606835 SED, Maroteaux type, 184095 Metatropic dysplasia, 156530 Scapuloperoneal spinal muscular atrophy, 181405 Hereditary motor and sensory neuropathy, type IIc, 606071 ?Avascular necrosis of femoral head, primary, 2, 617383 Neuronopathy, distal hereditary motor, type VIII, 600175 Parastremmatic dwarfism, 168400 Brachyolmia type 3, 113500
TRPV6	100%	99%	Hyperparathyroidism, transient neonatal, 618188
TRRAP	100%	100%	?Deafness, autosomal dominant 75, 618778 Developmental delay with or without dysmorphic facies and autism, 618454
TSC1	100%	100%	Focal cortical dysplasia, type II, somatic, 607341 Tuberous sclerosis-1, 191100 Lymphangioleiomyomatosis, 606690
TSC2	100%	100%	Lymphangioleiomyomatosis, somatic, 606690 ?Focal cortical dysplasia, type II, somatic, 607341 Tuberous sclerosis-2, 613254
TSEN15	100%	100%	Pontocerebellar hypoplasia, type 2F, 617026
TSEN2	100%	100%	Pontocerebellar hypoplasia type 2B, 612389

TSEN34	100%	100%	?Pontocerebellar hypoplasia type 2C, 612390
TSEN54	100%	100%	Pontocerebellar hypoplasia type 2A, 277470 Pontocerebellar hypoplasia type 4, 225753 ?Pontocerebellar hypoplasia type 5, 610204
TSFM	94%	94%	Combined oxidative phosphorylation deficiency 3, 610505
TSGA10	100%	100%	?Spermatogenic failure 26, 617961
TSHB	100%	100%	Hypothyroidism, congenital, nongoitrous 4, 275100
TSHR	100%	100%	Hyperthyroidism, familial gestational, 603373 Hyperthyroidism, nonautoimmune, 609152 Hypothyroidism, congenital, nongoitrous, 1, 275200 Thyroid adenoma, hyperfunctioning, somatic, Thyroid carcinoma with thyrotoxicosis,
TSHZ1	100%	100%	Aural atresia, congenital, 607842
TSPAN12	100%	100%	Exudative vitreoretinopathy 5, 613310
TSPAN7	100%	100%	Intellectual developmental disorder, X-linked 58, 300210
TSPEAR	100%	100%	?Deafness, autosomal recessive 98, 614861 Ectodermal dysplasia 14, hair/tooth type with or without hypohidrosis, 618180
TSPYL1	100%	100%	Sudden infant death with dysgenesis of the testes syndrome, 608800
TSR2	100%	100%	?Diamond-Blackfan anemia 14 with mandibulofacial dysostosis, 300946
TTBK2	100%	100%	Spinocerebellar ataxia 11, 604432
TTC12	100%	100%	Ciliary dyskinesia, primary, 45, 618801
TTC19	100%	100%	Mitochondrial complex III deficiency, nuclear type 2, 615157
TTC21A	100%	100%	Spermatogenic failure 37, 618429

TTC21B	100%	100%	Short-rib thoracic dysplasia 4 with or without polydactyly, 613819 Nephronophthisis 12, 613820
TTC26	100%	100%	Biliary, renal, neurologic, and skeletal syndrome, 619534
TTC29	100%	100%	Spermatogenic failure 42, 618745
TTC37	100%	100%	Trichohepatoenteric syndrome 1, 222470
TTC5	100%	100%	Neurodevelopmental disorder with cerebral atrophy and variable facial dysmorphism, 619244
TTC7A	100%	100%	Gastrointestinal defects and immunodeficiency syndrome, 243150
TTC8	100%	100%	Bardet-Biedl syndrome 8, 615985 ?Retinitis pigmentosa 51, 613464
TTI2	100%	100%	Mental retardation, autosomal recessive 39, 615541
TTLL5	100%	100%	Cone-rod dystrophy 19, 615860
TTN	100%	100%	Muscular dystrophy, limb-girdle, autosomal recessive 10, 608807 Cardiomyopathy, familial hypertrophic, 9, 613765 Tibial muscular dystrophy, tardive, 600334 Salih myopathy, 611705 Cardiomyopathy, dilated, 1G, 604145 Myopathy, myofibrillar, 9, with early respiratory failure, 603689
TTPA	100%	100%	Ataxia with isolated vitamin E deficiency, 277460
TTR	94%	94%	Amyloidosis, hereditary, transthyretin-related, 105210 Carpal tunnel syndrome, familial, 115430
TUB	100%	100%	?Retinal dystrophy and obesity, 616188
TUBA1A	100%	100%	Lissencephaly 3, 611603
TUBA3D	100%	100%	Keratoconus 9, 617928
TUBA4A	100%	100%	Amyotrophic lateral sclerosis 22 with or without frontotemporal dementia, 616208
TUBA8	100%	100%	No OMIM disease ID

TUBB	100%	99%	Symmetric circumferential skin creases, congenital, 1, 156610 Cortical dysplasia, complex, with other brain malformations 6, 615771
TUBB1	100%	100%	Macrothrombocytopenia, autosomal dominant, TUBB1-related, 613112
TUBB2A	100%	100%	Cortical dysplasia, complex, with other brain malformations 5, 615763
TUBB2B	100%	100%	Cortical dysplasia, complex, with other brain malformations 7, 610031
TUBB3	100%	100%	Fibrosis of extraocular muscles, congenital, 3A, 600638 Cortical dysplasia, complex, with other brain malformations 1, 614039
TUBB4A	99%	98%	Dystonia 4, torsion, autosomal dominant, 128101 Leukodystrophy, hypomyelinating, 6, 612438
TUBB4B	100%	100%	Leber congenital amaurosis with early-onset deafness, 617879
TUBB6	100%	100%	?Facial palsy, congenitla, with ptosis and velopharyngeal dysfunction, 617732
TUBB8	100%	100%	Oocyte maturation defect 2, 616780
TUBG1	100%	100%	Cortical dysplasia, complex, with other brain malformations 4, 615412
TUBGCP2	97%	97%	Pachygyria, microcephaly, developmental delay, and dysmorphic facies, with or without seizures, 618737
TUBGCP4	100%	100%	Microcephaly and chorioretinopathy, autosomal recessive, 3, 616335
TUBGCP6	100%	100%	Microcephaly and chorioretinopathy, autosomal recessive, 1, 251270
TUFM	100%	100%	Combined oxidative phosphorylation deficiency 4, 610678
TULP1	100%	100%	Leber congenital amaurosis 15, 613843 Retinitis pigmentosa 14, 600132
TUSC3	100%	100%	Mental retardation, autosomal recessive 7, 611093
TWIST1	100%	99%	Craniosynostosis 1, 123100 Robinow-Sorauf syndrome, 180750 Sweeney-Cox syndrome, 617746 Saethre-Chotzen syndrome with or without eyelid anomalies, 101400

TWIST2	100%	100%	Blepharon-macrostomia syndrome, 200110 Barber-Say syndrome, 209885 Focal facial dermal dysplasia 3, Setleis type, 227260
TWNK	100%	100%	Mitochondrial DNA depletion syndrome 7 (hepatocerebral type), 271245 Progressive external ophthalmoplegia with mitochondrial DNA deletions, autosomal dominant 3, 609286 Perrault syndrome 5, 616138
TXN2	100%	100%	?Combined oxidative phosphorylation deficiency 29, 616811
TXNL4A	100%	100%	Burn-McKeown syndrome, 608572
TXNRD2	100%	100%	?Glucocorticoid deficiency 5, 617825
TYK2	100%	100%	Immunodeficiency 35, 611521
TYMP	100%	100%	Mitochondrial DNA depletion syndrome 1 (MNGIE type), 603041
TYMS	100%	100%	No OMIM disease ID
TYR	100%	100%	Albinism, oculocutaneous, type IB, 606952 Waardenburg syndrome/albinism, digenic, 103470 Albinism, oculocutaneous, type IA, 203100
TYROBP	100%	100%	Polycystic lipomembranous osteodysplasia with sclerosing leukoencephalopathy 1, 221770
TYRP1	100%	100%	Albinism, oculocutaneous, type III, 203290
U2AF2	100%	100%	No OMIM disease ID
UBA1	100%	99%	Spinal muscular atrophy, X-linked 2, infantile, 301830 VEXAS syndrome, somatic, 301054
UBA5	100%	100%	?Spinocerebellar ataxia, autosomal recessive 24, 617133 Developmental and epileptic encephalopathy 44, 617132
UBAP1	100%	100%	Spastic paraplegia 80, autosomal dominant, 618418

UBB	100%	100%	No OMIM disease ID
UBE2A	100%	100%	Intellectual developmental disorder, X-linked syndromic, Nascimento type, 300860
UBE2T	100%	100%	Fanconi anemia, complementation group T, 616435
UBE3A	100%	100%	Angelman syndrome, 105830
UBE3B	100%	100%	Kaufman oculocerebrofacial syndrome, 244450
UBE4A	100%	100%	Neurodevelopmental disorder with hypotonia and gross motor and seech delay, 619639
UBIAD1	100%	100%	Corneal dystrophy, Schnyder type, 121800
UBQLN2	100%	100%	Amyotrophic lateral sclerosis 15, with or without frontotemporal dementia, 300857
UBR1	98%	98%	Johanson-Blizzard syndrome, 243800
UBR2	100%	99%	No OMIM disease ID
UBR7	100%	100%	Li-Campeau syndrome, 619189
UBTF	100%	100%	Neurodegeneration, childhood-onset, with brain atrophy, 617672
UCHL1	100%	100%	Spastic paraplegia 79, autosomal recessive, 615491
UFC1	100%	100%	Neurodevelopmental disorder with spasticity and poor growth, 618076
UFM1	100%	100%	Leukodystrophy, hypomyelinating, 14, 617899
UFSP2	100%	100%	?Hip dysplasia, Beukes type, 142669 ?Spondyloepimetaphyseal dysplasia, Di Rocco type, 617974
UGDH	100%	100%	Developmental and epileptic encephalopathy 84, 618792
UGP2	97%	96%	Developmental and epileptic encephalopathy 83, 618744
UGT1A1	100%	100%	Crigler-Najjar syndrome, type I, 218800 Hyperbilirubinemia, familial transient neonatal, 237900 Crigler-Najjar syndrome, type II, 606785
UMOD	100%	100%	Tubulointerstitial kidney disease, autosomal dominant, 1, 162000
UMPS	97%	97%	Orotic aciduria, 258900
UNC119	100%	100%	?Immunodeficiency 13, 615518 ?Cone-rod dystrophy,
UNC13A	100%	99%	No OMIM disease ID
UNC13D	100%	100%	Hemophagocytic lymphohistiocytosis, familial, 3, 608898
UNC45A	100%	100%	Osteootohepatoenteric syndrome, 619377
UNC45B	100%	100%	?Cataract 43, 616279 Myofibrillar myopathy 11, 619178

UNC80	100%	100%	Hypotonia, infantile, with psychomotor retardation and characteristic facies 2, 616801
UNC93B1	100%	100%	No OMIM disease ID
UNG	100%	100%	Immunodeficiency with hyper IgM, type 5, 608106
UPB1	100%	100%	Beta-ureidopropionase deficiency, 613161
UPF1	99%	98%	No OMIM disease ID
UPF3B	100%	100%	Intellectual developmental disorder, X-linked syndromic 14, 300676
UPK3A	100%	100%	No OMIM disease ID
UQCC1	100%	100%	No OMIM disease ID
UQCC2	100%	100%	Mitochondrial complex III deficiency, nuclear type 7, 615824
UQCC3	100%	100%	?Mitochondrial complex III deficiency, nuclear type 9, 616111
UQCR10	100%	100%	No OMIM disease ID
UQCR11	100%	100%	No OMIM disease ID
UQCRB	100%	100%	Mitochondrial complex III deficiency, nuclear type 3, 615158
UQCRC1	100%	100%	Parkinsonism with polyneuropathy, 619279
UQCRC2	100%	100%	Mitochondrial complex III deficiency, nuclear type 5, 615160
UQCRFS1	100%	100%	Mitochondrial complex III deficiency, nuclear type 10, 618775
UQCRH	100%	100%	No OMIM disease ID
UQCRQ	100%	100%	Mitochondrial complex III deficiency, nuclear type 4, 615159
UROC1	100%	100%	?Urocanase deficiency, 276880
UROD	100%	100%	Porphyria, hepatoerythropoietic, 176100 Porphyria cutanea tarda, 176100
UROS	100%	100%	Porphyria, congenital erythropoietic, 263700
USB1	100%	100%	Poikiloderma with neutropenia, 604173
USH1C	100%	100%	Usher syndrome, type 1C, 276904 Deafness, autosomal recessive 18A, 602092
USH1G	100%	100%	Usher syndrome, type 1G, 606943
USH2A	99%	99%	Usher syndrome, type 2A, 276901 Retinitis pigmentosa 39, 613809
USP18	100%	100%	Pseudo-TORCH syndrome 2, 617397
USP26	100%	100%	No OMIM disease ID
USP27X	100%	100%	Intellectual developmental disorder, X-linked 105, 300984
USP45	100%	100%	?Leber congenital amaurosis 19, 618513

USP48	100%	100%	No OMIM disease ID
USP7	94%	94%	Hao-Fountain syndrome, 616863
USP8	100%	100%	Pituitary adenoma 4, ACTH-secreting, somatic, 219090
USP9X	100%	100%	Intellectual developmental disorder, X-linked 99, 300919 Intellectual developmental disorder, X-linked 99, syndromic, female-restricted, 300968
UST	100%	100%	No OMIM disease ID
UVSSA	100%	100%	UV-sensitive syndrome 3, 614640
VAC14	100%	100%	Striatonigral degeneration, childhood-onset, 617054
VAMP1	100%	100%	Myasthenic syndrome, congenital, 25, 618323 Spastic ataxia 1, autosomal dominant, 108600
VAMP2	100%	100%	Neurodevelopmental disorder with hypotonia and autistic features with or without hyperkinetic movements, 618760
VANGL1	100%	100%	Caudal regression syndrome, 600145
VANGL2	100%	100%	Neural tube defects, 182940
VAPB	100%	100%	Spinal muscular atrophy, late-onset, Finkel type, 182980 Amyotrophic lateral sclerosis 8, 608627
VARS1	100%	100%	Neurodevelopmental disorder with microcephaly, seizures, and cortical atrophy, 617802
VARS2	100%	100%	Combined oxidative phosphorylation deficiency 20, 615917
VAV1	97%	97%	No OMIM disease ID
VAX1	99%	99%	?Microphthalmia, syndromic 11, 614402
VCAN	100%	100%	Wagner syndrome 1, 143200
VCL	100%	100%	Cardiomyopathy, dilated, 1W, 611407 Cardiomyopathy, hypertrophic, 15, 613255

VCP	100%	100%	Frontotemporal dementia and/or amyotrophic lateral sclerosis 6, 613954 Charcot-Marie-Tooth disease, type 2Y, 616687 Inclusion body myopathy with early-onset Paget disease and frontotemporal dementia 1, 167320
VDR	100%	99%	Rickets, vitamin D-resistant, type IIA, 277440
VEGFC	100%	100%	Lymphatic malformation 4, 615907
VHL	100%	100%	Erythrocytosis, familial, 2, 263400 von Hippel-Lindau syndrome, 193300 Renal cell carcinoma, somatic, 144700 Pheochromocytoma, 171300 Hemangioblastoma, cerebellar, somatic,
VIM	100%	100%	Cataract 30, pulverulent, 116300
VIPAS39	100%	100%	Arthrogryposis, renal dysfunction, and cholestasis 2, 613404
VKORC1	93%	93%	Vitamin K-dependent clotting factors, combined deficiency of, 2, 607473 Warfarin resistance, 122700
VLDLR	100%	100%	Cerebellar hypoplasia and mental retardation with or without quadrupedal locomotion 1, 224050
VMA21	100%	100%	Myopathy, X-linked, with excessive autophagy, 310440
VPS11	100%	100%	?Dystonia 32, 619637 Leukodystrophy, hypomyelinating, 12, 616683
VPS13A	100%	100%	Choreoacanthocytosis, 200150
VPS13B	99%	99%	Cohen syndrome, 216550
VPS13C	100%	100%	Parkinson disease 23, autosomal recessive, early onset, 616840
VPS13D	100%	100%	Spinocerebellar ataxia, autosomal recessive 4, 607317
VPS16	100%	100%	Dystonia 30, 619291
VPS33A	89%	89%	Mucopolysaccharidosis-plus syndrome, 617303
VPS33B	100%	100%	Arthrogryposis, renal dysfunction, and cholestasis 1, 208085
VPS35	100%	100%	No OMIM disease ID
VPS35L	100%	100%	Ritscher-Schinzel syndrome 3, 619135

VPS37A	100%	100%	Spastic paraplegia 53, autosomal recessive, 614898
VPS41	100%	100%	Spinocerebellar ataxia, autosomal recessive 29, 619389
VPS45	95%	95%	Neutropenia, severe congenital, 5, autosomal recessive, 615285
VPS4A	100%	100%	CIMDAG syndrome, 619273
VPS50	100%	100%	No OMIM disease ID
VPS51	100%	100%	Pontocerebellar hypoplasia, type 13, 618606
VPS53	100%	99%	Pontocerebellar hypoplasia, type 2E, 615851
VRK1	100%	100%	Pontocerebellar hypoplasia type 1A, 607596
VSX1	100%	100%	?Craniofacial anomalies and anterior segment dysgenesis syndrome, 614195 Keratoconus 1, 148300
VSX2	100%	100%	Microphthalmia, isolated 2, 610093 Microphthalmia with coloboma 3, 610092
VWA1	100%	99%	Neuropathy, hereditary motor, with myopathic features, 619216
VWA3B	100%	100%	?Spinocerebellar ataxia, autosomal recessive 22, 616948
VWF	100%	100%	von Willebrand disease, type 1, 193400 von Willebrand disease, types 2A, 2B, 2M, and 2N, 613554 von Willebrand disease, type 3, 277480
WAC	100%	100%	Desanto-Shinawi syndrome, 616708
WARS1	100%	100%	Neuronopathy, distal hereditary motor, type IX, 617721
WARS2	100%	100%	Neurodevelopmental disorder, mitochondrial, with abnormal movements and lactic acidosis, with or without seizures, 617710
WAS	100%	100%	Wiskott-Aldrich syndrome, 301000 Neutropenia, severe congenital, X-linked, 300299 Thrombocytopenia, X-linked, intermittent, 313900 Thrombocytopenia, X-linked, 313900
WASF1	100%	100%	Neurodevelopmental disorder with absent language and variable seizures, 618707
WASHC4	100%	100%	?Mental retardation, autosomal recessive 43, 615817

WASHC5	100%	100%	Ritscher-Schinzel syndrome 1, 220210 Spastic paraplegia 8, autosomal dominant, 603563
WBP11	100%	100%	Vertebral, cardiac, tracheoesophageal, renal, and limb defects, 619227
WBP2	100%	100%	Deafness, autosomal recessive 107, 617639
WDFY3	100%	100%	?Microcephaly 18, primary, autosomal dominant, 617520
WDPCP	98%	98%	?Bardet-Biedl syndrome 15, 615992 Congenital heart defects, hamartomas of tongue, and polysyndactyly, 217085
WDR1	100%	100%	Periodic fever, immunodeficiency, and thrombocytopenia syndrome, 150550
WDR11	100%	100%	Hypogonadotropic hypogonadism 14 with or without anosmia, 614858
WDR13	100%	100%	No OMIM disease ID
WDR19	100%	100%	Nephronophthisis 13, 614377 Senior-Loken syndrome 8, 616307 ?Short-rib thoracic dysplasia 5 with or without polydactyly, 614376 ?Cranioectodermal dysplasia 4, 614378
WDR26	97%	95%	Skraban-Deardorff syndrome, 617616
WDR35	100%	100%	Short-rib thoracic dysplasia 7 with or without polydactyly, 614091 Cranioectodermal dysplasia 2, 613610
WDR36	100%	100%	Glaucoma 1, open angle, G, 609887
WDR37	86%	86%	Neurooculocardiogenitourinary syndrome, 618652
WDR4	100%	100%	Galloway-Mowat syndrome 6, 618347 Microcephaly, growth deficiency, seizures, and brain malformations, 618346
WDR45	100%	100%	Neurodegeneration with brain iron accumulation 5, 300894
WDR45B	100%	100%	Neurodevelopmental disorder with spastic quadriplegia and brain abnormalities with or without seizures, 617977
WDR62	100%	100%	Microcephaly 2, primary, autosomal recessive, with or without cortical malformations, 604317
WDR72	96%	96%	Amelogenesis imperfecta, type IIA3, 613211
WDR73	100%	100%	Galloway-Mowat syndrome 1, 251300

WDR81	100%	100%	Cerebellar ataxia, mental retardation, and dysequilibrium syndrome 2, 610185 Hydrocephalus, congenital, 3, with brain anomalies, 617967
WEE2	100%	100%	Oocyte maturation defect 5, 617996
WFS1	100%	100%	Deafness, autosomal dominant 6/14/38, 600965 ?Cataract 41, 116400 Wolfram-like syndrome, autosomal dominant, 614296 Wolfram syndrome 1, 222300
WHRN	100%	100%	Deafness, autosomal recessive 31, 607084 Usher syndrome, type 2D, 611383
WIPF1	100%	100%	Wiskott-Aldrich syndrome 2, 614493
WIPI2	100%	100%	?Intellectual developmental disorder with short stature and variable skeletal anomalies, 618453
WNK1	100%	100%	Neuropathy, hereditary sensory and autonomic, type II, 201300 Pseudohypoaldosteronism, type IIC, 614492
WNK4	100%	100%	Pseudohypoaldosteronism, type IIB, 614491
WNT1	100%	100%	Osteogenesis imperfecta, type XV, 615220
WNT10A	100%	100%	Schopf-Schulz-Passarge syndrome, 224750 Tooth agenesis, selective, 4, 150400 Odontoonychodermal dysplasia, 257980
WNT10B	100%	100%	Tooth agenesis, selective, 8, 617073 Split-hand/foot malformation 6, 225300
WNT2B	100%	100%	Diarrhea 9, 618168
WNT3	100%	100%	?Tetra-amelia syndrome 1, 273395

WNT4	100%	99%	?SERKAL syndrome, 611812 Mullerian aplasia and hyperandrogenism, 158330
WNT5A	100%	100%	Robinow syndrome, autosomal dominant 1, 180700
WNT6	100%	100%	No OMIM disease ID
WNT7A	100%	100%	Fuhrmann syndrome, 228930 Ulna and fibula, absence of, with severe limb deficiency, 276820
WRAP53	100%	100%	Dyskeratosis congenita, autosomal recessive 3, 613988
WRN	100%	100%	Werner syndrome, 277700
WT1	97%	97%	Mesothelioma, somatic, 156240 Meacham syndrome, 608978 Frasier syndrome, 136680 Nephrotic syndrome, type 4, 256370 Denys-Drash syndrome, 194080 Wilms tumor, type 1, 194070
WWOX	100%	100%	Esophageal squamous cell carcinoma, somatic, 133239 Developmental and epileptic encephalopathy 28, 616211 Spinocerebellar ataxia, autosomal recessive 12, 614322
XDH	100%	100%	Xanthinuria, type I, 278300
XIAP	100%	100%	Lymphoproliferative syndrome, X-linked, 2, 300635
XIRP2	100%	100%	No OMIM disease ID
XIST	NC	NC	X-inactivation, familial skewed, 300087
XK	100%	100%	McLeod syndrome with or without chronic granulomatous disease, 300842
XPA	100%	100%	Xeroderma pigmentosum, group A, 278700
XPC	100%	100%	Xeroderma pigmentosum, group C, 278720
XPNPEP3	100%	100%	Nephronophthisis-like nephropathy 1, 613159
XPO5	100%	100%	No OMIM disease ID
XPR1	100%	100%	Basal ganglia calcification, idiopathic, 6, 616413
XRCC1	100%	100%	?Spinocerebellar ataxia, autosomal recessive 26, 617633

XRCC2	100%	100%	Spermatogenic failure, 619145 ?Premature ovarian failure 17, 619146 ?Fanconi anemia, complementation group U, 617247
XRCC4	100%	100%	Short stature, microcephaly, and endocrine dysfunction, 616541
XYLT1	99%	98%	Desbuquois dysplasia 2, 615777
XYLT2	96%	96%	Spondyloocular syndrome, 605822
YAP1	100%	100%	Coloboma, ocular, with or without hearing impairment, cleft lip/palate, and/or mental retardation, 120433
YARS1	100%	100%	Infantile-onset multisystem neurologic, endocrine, and pancreatic disease 2, 619418 Charcot-Marie-Tooth disease, dominant intermediate C, 608323
YARS2	100%	100%	Myopathy, lactic acidosis, and sideroblastic anemia 2, 613561
YEATS2	100%	100%	?Epilepsy, myoclonic, familial adult, 4, 615127
YIF1B	90%	90%	Kaya-Barakat-Masson syndrome, 619125
YIPF5	100%	100%	Microcephaly, epilepsy, and diabetes syndrome 2, 619278
YME1L1	100%	100%	?Optic atrophy 11, 617302
YPEL2	100%	99%	No OMIM disease ID
YWHAE	100%	100%	No OMIM disease ID
YWHAG	100%	100%	Developmental and epileptic encephalopathy 56, 617665
YWHAZ	100%	100%	No OMIM disease ID
YY1	100%	100%	Gabriele-de Vries syndrome, 617557
YY1AP1	100%	100%	Grange syndrome, 602531
ZAP70	100%	100%	Immunodeficiency 48, 269840 Autoimmune disease, multisystem, infantile-onset, 2, 617006
ZBTB11	100%	100%	Intellectual developmental disorder, autosomal recessive 69, 618383
ZBTB16	100%	100%	Skeletal defects, genital hypoplasia, and mental retardation, 612447 Leukemia, acute promyelocytic, PL2F/RARA type,
ZBTB17	100%	100%	No OMIM disease ID
ZBTB18	100%	100%	Mental retardation, autosomal dominant 22, 612337

ZBTB20	100%	100%	Primrose syndrome, 259050
ZBTB24	100%	100%	Immunodeficiency-centromeric instability-facial anomalies syndrome 2, 614069
ZBTB42	100%	100%	?Lethal congenital contracture syndrome 6, 616248
ZC3H14	100%	100%	Intellectual developmental disorder, autosomal recessive 56, 617125
ZC4H2	100%	100%	Wieacker-Wolff syndrome, 314580 Wieacker-Wolff syndrome, female-restricted, 301041
ZCCHC8	100%	100%	?Pulmonary fibrosis and/or bone marrow failure, telomere-related, 5, 618674
ZDHHC9	100%	100%	Mental retardation, X-linked syndromic, Raymond type, 300799
ZEB1	100%	100%	Corneal dystrophy, posterior polymorphous, 3, 609141 Corneal dystrophy, Fuchs endothelial, 6, 613270
ZEB2	97%	97%	Mowat-Wilson syndrome, 235730
ZFHX2	100%	100%	?Marsili syndrome, 147430
ZFHX3	100%	100%	Prostate cancer, somatic, 176807
ZFHX4	100%	100%	No OMIM disease ID
ZFP57	100%	100%	Diabetes mellitus, transient neonatal 1, 601410
ZFPM2	100%	100%	Diaphragmatic hernia 3, 610187 46XY sex reversal 9, 616067 Tetralogy of Fallot, 187500
ZFYVE26	100%	100%	Spastic paraplegia 15, autosomal recessive, 270700
ZFYVE27	100%	100%	Spastic paraplegia 33, autosomal dominant, 610244
ZIC1	100%	100%	?Craniosynostosis 6, 616602 Structural brain anomalies with impaired intellectual development and craniosynostosis, 618736
ZIC2	100%	99%	Holoprosencephaly 5, 609637
ZIC3	100%	100%	Congenital heart defects, nonsyndromic, 1, X-linked, 306955 Heterotaxy, visceral, 1, X-linked, 306955 VACTERL association, X-linked, 314390
ZMIZ1	100%	100%	Neurodevelopmental disorder with dysmorphic facies and distal skeletal anomalies, 618659

ZMPSTE24	100%	100%	Mandibuloacral dysplasia with type B lipodystrophy, 608612 Restrictive dermopathy, lethal, 275210
ZMYM2	100%	100%	Neurodevelopmental-craniofacial syndrome with variable renal and cardiac abnormalities, 619522
ZMYND10	100%	100%	Ciliary dyskinesia, primary, 22, 615444
ZMYND11	100%	100%	Mental retardation, autosomal dominant 30, 616083
ZMYND15	100%	100%	?Spermatogenic failure 14, 615842
ZNF141	100%	100%	?Polydactyly, postaxial, type A6, 615226
ZNF142	100%	100%	Neurodevelopmental disorder with impaired speech and hyperkinetic movements, 618425
ZNF148	100%	100%	Global developmental delay, absent or hypoplastic corpus callosum, and dysmorphic facies, 617260
ZNF292	99%	99%	Intellectual developmental disorder, autosomal dominant 64, 619188
ZNF335	100%	100%	Microcephaly 10, primary, autosomal recessive, 615095
ZNF341	100%	100%	Hyper-IgE recurrent infection syndrome 3, autosomal recessive, 618282
ZNF407	100%	100%	SIMHA syndrome, 619557
ZNF408	100%	100%	Retinitis pigmentosa 72, 616469 ?Exudative vitreoretinopathy 6, 616468
ZNF41	100%	100%	No OMIM disease ID
ZNF423	100%	100%	Nephronophthisis 14, 614844 Joubert syndrome 19, 614844
ZNF462	100%	100%	Weiss-Kruszka syndrome, 618619
ZNF469	100%	100%	Brittle cornea syndrome 1, 229200
ZNF513	100%	100%	?Retinitis pigmentosa 58, 613617
ZNF526	100%	100%	No OMIM disease ID
ZNF592	100%	100%	No OMIM disease ID
ZNF644	100%	100%	Myopia 21, autosomal dominant, 614167
ZNF687	100%	100%	Paget disease of bone 6, 616833
ZNF699	100%	100%	DEGCAGS syndrome, 619488
ZNF711	100%	99%	Intellectual developmental disorder, X-linked 97, 300803
ZNF750	100%	100%	Seborrhea-like dermatitis with psoriasiform elements, 610227
ZNFX1	100%	100%	No OMIM disease ID
ZNHIT3	75%	74%	PEHO syndrome, 260565
ZP1	100%	100%	Oocyte maturation defect 1, 615774

ZP2	100%	100%	Oocyte maturation defect 6, 618353
ZP3	100%	100%	Oocyte maturation defect 3, 617712
ZPR1	100%	100%	?Growth restriction, hypoplastic kidneys, alopecia, and distinctive facies, 619321
ZSWIM6	97%	95%	Neurodevelopmental disorder with movement abnormalities, abnormal gait, and autistic features, 617865 Acromelic frontonasal dysostosis, 603671

Gene symbols used follow HGCN guidelines: Gray KA, Yates B, Seal RL, Wright MW, Bruford EA. Nucleic Acids Res. 2015 Jan 43(Database issue):D1079-85.
Gene symbols used follow HGCN guidelines: Gray KA, Yates B, Seal RL, Wright MW, Bruford EA. Nucleic Acids Res. 2015 Jan 43(Database issue):D1079-85.
TWIST is the chemistry used for WES analysis.
Covered 10x describes the percentage of a gene's coding sequence that is covered at least 10x.
Covered 20x describes the percentage of a gene's coding sequence that is covered at least 20x.
Genes with coverage denoting NC are non-protein coding genes.
non-protein coding genes are covered, but as coverage statistics are based on protein coding regions, statistics could not be generated.
OMIM release used for OMIM disease identifiers and descriptions : January 13th , 2022.
This list is accurate for panel version DG 3.3.0
Ad 1. "No OMIM Disease ID" signifies a gene without a current OMIM association Ad 2. OMIM phenotype descriptions between {} signify risk factors