Supplementary Table 1. 2742 confirmed disease-causing genes targeted in this study

Gene	Disorder*
A2M	Alpha-2-macroglobulin deficiency, 614036 (1)/Alzheimer disease, susceptibility to, 104300 (3)
AAAS	Achalasia-addisonianism-alacrimia syndrome, 231550 (3)
AAGAB	Keratoderma, palmoplantar, punctate type IA, 148600 (3)
AARS	Charcot-Marie-Tooth disease, axonal, type 2N, 613287 (3)
AARS2	Combined oxidative phosphorylation deficiency 8, 614096 (3)/Leukoencephalopathy, progressive, with ovarian failure, 615889 (3)
AASS	Hyperlysinemia, 238700 (3)/Saccharopinuria, 268700 (1)
ABAT	GABA-transaminase deficiency, 613163 (3)
ABCA1	HDL deficiency, type 2, 604091 (3)/Tangier disease, 205400 (3)/Coronary artery disease in familial hypercholesterolemia, protection against, 143890 (3)
ABCA12	Ichthyosis, autosomal recessive 4B (harlequin), 242500 (3)/Ichthyosis, congenital, autosomal recessive 4A, 601277 (3)
ABCA3	Surfactant metabolism dysfunction, pulmonary, 3, 610921 (3)
ABCA4	Cone-rod dystrophy 3, 604116 (3)/Fundus flavimaculatus, 248200 (3)/Retinal dystrophy, early-onset severe, 248200 (3)/Retinitis pigmentosa 19, 601718 (3)/Stargardt disease 1, 248200 (3)/Macular degeneration, age-related, 2, 153800 (3)
ABCB1	Colchicine resistance, 120080 (3)/Inflammatory bowel disease 13, 612244 (3)
ABCB11	Cholestasis, benign recurrent intrahepatic, 2, 605479 (3)/Cholestasis, progressive familial intrahepatic 2, 601847 (3)
ABCB4	Cholestasis, intrahepatic, of pregnancy, 3, 614972 (3)/Cholestasis, progressive familial intrahepatic 3, 602347 (3)/Gallbladder disease 1, 600803 (3)
ABCB6	Dyschromatosis universalis hereditaria 3, 615402 (3)/Microphthalmia, isolated, with coloboma 7, 614497 (3)/[Blood group, Langereis system], 111600 (3)
ABCB7	Anemia, sideroblastic, with ataxia, 301310 (3)
ABCC11	[Axillary odor, variation in], 117800 (3)/[Colostrum secretion, variation in], 117800 (3)/[Earwax, wet/dry], 117800 (3)
ABCC2	Dubin-Johnson syndrome, 237500 (3)
ABCC6	Arterial calcification, generalized, of infancy, 2, 614473 (3)/Pseudoxanthoma elasticum, 264800 (3)/Pseudoxanthoma elasticum, forme fruste, 177850 (3)
ABCC8	Diabetes mellitus, noninsulin-dependent, 125853 (3)/Diabetes mellitus, permanent neonatal, 606176 (3)/Diabetes mellitus, transient neonatal 2, 610374 (3)/Hyperinsulinemic hypoglycemia, familial, 1, 256450 (3)/Hypoglycemia of infancy, leucine-sensitive, 240800 (3)
ABCC9	Atrial fibrillation, familial, 12, 614050 (3)/Cardiomyopathy, dilated, 1O, 608569 (3)/Hypertrichotic osteochondrodysplasia, 239850 (3)
ABCD1	Adrenoleukodystrophy, 300100 (3)/Adrenomyeloneuropathy, adult, 300100 (3)
ABCD3	Bile acid synthesis defect, congenital, 5
ABCD4	Methylmalonic aciduria and homocystinuria, cblJ type, 614857 (3)
ABCG5	Sitosterolemia, 210250 (3)
ABCG8	Gallbladder disease 4, 611465 (3)/Sitosterolemia, 210250 (3)
ABHD12	Polyneuropathy, hearing loss, ataxia, retinitis pigmentosa, and cataract, 612674 (3)
ABHD5	Chanarin-Dorfman syndrome, 275630 (3)
ABL1	Leukemia, Philadelphia chromosome-positive, resistant to imatinib (3)
ACACA	Acetyl-CoA carboxylase deficiency, 613933 (1)
ACAD8	Isobutyryl-CoA dehydrogenase deficiency, 611283 (3)
ACAD9	ACAD9 deficiency, 611126 (3)
ACADL	long-chain acyl-CoA dehydrogenase (LCAD) deficiency
ACADM	Acyl-CoA dehydrogenase, medium chain, deficiency of, 201450 (3)
ACADS	Acyl-CoA dehydrogenase, short-chain, deficiency of, 201470 (3)
ACADSB	2-methylbutyrylglycinuria, 610006 (3)
ACADVL	VLCAD deficiency, 201475 (3)
ACAN	Osteochondritis dissecans, short stature, and early-onset osteoarthritis, 165800 (3)/Spondyloepimetaphyseal dysplasia, aggrecan type, 612813 (3)/Spondyloepiphyseal dysplasia, Kimberley type, 608361 (3)

ACAT1	Alpha-methylacetoacetic aciduria, 203750 (3)
ACAT2	ACAT2 deficiency, 614055 (1)
ACE	Renal tubular dysgenesis, 267430 (3)/[Angiotensin I-converting enzyme, benign serum increase] (3)/Alzheimer disease, susceptibility to, 104300 (3)/Microvascular complications of diabetes 3, 612624 (3)/Myocardial infarction, susceptibility to (3)/SARS, progression of (3)/Stroke, hemorrhagic, 614519 (3)
ACO2	Infantile cerebellar-retinal degeneration, 614559 (3)
ACOX1	Peroxisomal acyl-CoA oxidase deficiency, 264470 (3)
ACP2	Lysosomal acid phosphatase deficiency, 200950 (1)
ACP5	Spondyloenchondrodysplasia with immune dysregulation, 607944 (3)
ACSF3	Combined malonic and methylmalonic aciduria, 614265 (3)
ACSL4	Mental retardation, X-linked 63, 300387 (3)
ACTA1	Myopathy, actin, congenital, with cores, 161800 (3)/Myopathy, actin, congenital, with excess of thin myofilaments, 161800 (3)/Myopathy, congenital, with fiber-type disproportion 1, 255310 (3)/Nemaline myopathy 3, autosomal dominant or recessive, 161800 (3)
ACTA2	Aortic aneurysm, familial thoracic 6, 611788 (3)/Moyamoya disease 5, 614042 (3)/Multisystemic smooth muscle dysfunction syndrome, 613834 (3)
ACTB	Baraitser-Winter syndrome 1, 243310 (3)/Dystonia, juvenile-onset, 607371 (3)
ACTC1	Atrial septal defect 5, 612794 (3)/Cardiomyopathy, dilated, 1R, 613424 (3)/Cardiomyopathy, familial hypertrophic, 11, 612098 (3)/Left ventricular noncompaction 4, 613424 (3)
ACTG1	Baraitser-Winter syndrome 2, 614583 (3)/Deafness, autosomal dominant 20/26, 604717 (3)
ACTN2	Cardiomyopathy, dilated, 1AA, 612158 (3)
ACTN4	Glomerulosclerosis, focal segmental, 1, 603278 (3)
ACVR1	Fibrodysplasia ossificans progressiva, 135100 (3)
ACVR2B	Heterotaxy, visceral, 4, autosomal, 613751 (3)
ACVRL1	Telangiectasia, hereditary hemorrhagic, type 2, 600376 (3)
ACY1	Aminoacylase 1 deficiency, 609924 (3)
ADA	Adenosine deaminase deficiency, partial, 102700 (3)/Severe combined immunodeficiency due to ADA deficiency, 102700 (3)
ADAM17	Inflammatory skin and bowel disease, neonatal, 1, 614328 (3)
ADAM9	Cone-rod dystrophy 9, 612775 (3)
ADAMTS10	Weill-Marchesani syndrome 1, recessive, 277600 (3)
ADAMTS13	Thrombotic thrombocytopenic purpura, familial, 274150 (3)
ADAMTS17	Weill-Marchesani-like syndrome, 613195 (3)
ADAMTS18	Microcornea, myopic chorioretinal atrophy, and telecanthus, 615458 (3)
ADAMTS2	Ehlers-Danlos syndrome, type VIIC, 225410 (3)
ADAMTSL2	Geleophysic dysplasia 1, 231050 (3)
ADAMTSL4	Ectopia lentis et pupillae, 225200 (3)/Ectopia lentis, isolated, autosomal recessive, 225100 (3)
ADAR	Aicardi-Goutieres syndrome 6, 615010 (3)/Dyschromatosis symmetrica hereditaria, 127400 (3)
ADCK3	Coenzyme Q10 deficiency, primary, 4, 612016 (3)
ADCY5	Dyskinesia, familial, with facial myokymia, 606703 (3)
ADIPOQ	Adiponectin deficiency, 612556 (3)
ADK	Hypermethioninemia due to adenosine kinase deficiency, 614300 (3)
ADRA2B	Epilepsy, familial adult myoclonic 2
ADSL	Adenylosuccinase deficiency, 103050 (3)
AFF2	Mental retardation, X-linked, FRAXE type, 309548 (3)
AFG3L2	Ataxia, spastic, 5, autosomal recessive, 614487 (3)/Spinocerebellar ataxia 28, 610246 (3)
AFP	Alpha-fetoprotein deficiency, 615969 (3)/[Hereditary persistence of alpha-fetoprotein], 615970 (3)
AGA	Aspartylglucosaminuria, 208400 (3)
AGK	Cataract 38, autosomal recessive, 614691 (3)/Sengers syndrome, 212350 (3)
AGL	Glycogen storage disease IIIa, 232400 (3)/Glycogen storage disease IIIb, 232400 (3)
AGPAT2	Lipodystrophy, congenital generalized, type 1, 608594 (3)
AGPS	Rhizomelic chondrodysplasia punctata, type 3, 600121 (3)
AGRN	Myasthenic syndrome, congenital, with pre- and postsynaptic defects, 615120 (3)

AGT	Renal tubular dysgenesis, 267430 (3)/Hypertension, essential, susceptibility to, 145500 (3)/Preeclampsia, susceptibility to (3)
AGTR1	Hypertension, essential, 145500 (3)/Renal tubular dysgenesis, 267430 (3)
AGTR2	AGTR2-Related X-Linked Intellectual Disability (XLID)
AGXT	Hyperoxaluria, primary, type 1, 259900 (3)
AHCY	Hypermethioninemia with deficiency of S-adenosylhomocysteine hydrolase, 613752 (3)
AHI1	Joubert syndrome-3, 608629 (3)
AICDA	Immunodeficiency with hyper-IgM, type 2, 605258 (3)
AIFM1	Combined oxidative phosphorylation deficiency 6, 300816 (3)/Cowchock syndrome, 310490 (3)
AIMP1	Leukodystrophy, hypomyelinating, 3, 260600 (3)
AIP	Pituitary adenoma, ACTH-secreting, 219090 (3)/Pituitary adenoma, growth hormone-secreting, 102200 (3)/Pituitary adenoma, prolactin-secreting, 600634 (3)
AIPL1	Cone-rod dystrophy, 604393 (3)/Leber congenital amaurosis 4, 604393 (3)/Retinitis pigmentosa, juvenile, 604393 (3)
AIRE	Autoimmune polyendocrinopathy syndrome , type I, with or without reversible metaphyseal dysplasia, 240300 (3)
AK1	Hemolytic anemia due to adenylate kinase deficiency, 612631 (3)
AK2	Reticular dysgenesis, 267500 (3)
AKAP9	Long QT syndrome-11, 611820 (3)
AKR1C2	46XY sex reversal 8, 614279 (3)/Obesity, hyperphagia, and developmental delay (3)
AKR1D1	Bile acid synthesis defect, congenital, 2, 235555 (3)
AKT1	Breast cancer, somatic, 114480 (3)/Colorectal cancer, somatic, 114500 (3)/Cowden syndrome 6, 615109 (3)/Ovarian cancer, somatic, 167000 (3)/Proteus syndrome, somatic, 176920 (3)/Schizophrenia, susceptibility to, 181500 (2)
AKT2	Diabetes mellitus, type II, 125853 (3)/Hypoinsulinemic hypoglycemia with hemihypertrophy, 240900 (3)
AKT3	Megalencephaly-polymicrogyria-polydactyly-hydrocephalus syndrome 2, 615937 (3)
ALAD	Porphyria, acute hepatic, 612740 (3)/Lead poisoning, susceptibility to, 612740 (3)
ALAS2	Anemia, sideroblastic, X-linked, 300751 (3)/Protoporphyria, erythropoietic, X-linked, 300752 (3)
ALDH18A1	Cutis laxa, autosomal recessive, type IIIA, 219150 (3)
ALDH2	Alcohol sensitivity, acute
ALDH3A2	Sjogren-Larsson syndrome, 270200 (3)
ALDH4A1	Hyperprolinemia, type II, 239510 (3)
ALDH5A1	Succinic semialdehyde dehydrogenase deficiency, 271980 (3)
ALDH6A1	Methylmalonate semialdehyde dehydrogenase deficiency, 614105 (3)
ALDH7A1	Epilepsy, pyridoxine-dependent, 266100 (3)
ALDOA	Glycogen storage disease XII, 611881 (3)
ALDOB	Fructose intolerance, 229600 (3)
ALG1	Congenital disorder of glycosylation, type Ik, 608540 (3)
ALG10	Long QT syndrome, acquired, reduced susceptibility to, 613688 (3)
ALG11	Congenital disorder of glycosylation, type Ip, 613661 (3)
ALG12	Congenital disorder of glycosylation, type Ig, 607143 (3)
ALG13	Congenital disorder of glycosylation, type Is, 300884 (3)
ALG2	Congenital disorder of glycosylation, type Ii, 607906 (3)/Myasthenic syndrome, congenital, with tubular aggregates 3, 616228 (3)
ALG3	Congenital disorder of glycosylation, type Id, 601110 (3)
ALG6	Congenital disorder of glycosylation, type Ic, 603147 (3)
ALG8	Congenital disorder of glycosylation, type Ih, 608104 (3)
ALG9	Congenital disorder of glycosylation, type Il, 608776 (3)
ALK	Neuroblastoma, susceptibility to, 3, 613014 (3)
ALMS1	Alstrom syndrome, 203800 (3)
ALOX12B	Ichthyosis, congenital, autosomal recessive 2, 242100 (3)
ALOXE3	Ichthyosis, congenital, autosomal recessive 3, 606545 (3)
ALPL	Hypophosphatasia, adult, 146300 (3)/Hypophosphatasia, childhood, 241510 (3)/Hypophosphatasia, infantile, 241500 (3)/Odontohypophosphatasia, 146300 (3)

ALS2	Amyotrophic lateral sclerosis 2, juvenile, 205100 (3)/Primary lateral sclerosis, juvenile, 606353 (3)/Spastic paralysis, infantile onset ascending, 607225 (3)
ALX1	Frontonasal dysplasia 3, 613456 (3)
ALX3	Frontonasal dysplasia 1, 136760 (3)
ALX4	Frontonasal dysplasia 2, 613451 (3)/Parietal foramina 2, 609597 (3)/Craniosynostosis 5, susceptibility to, 615529 (3)
AMACR	Alpha-methylacyl-CoA racemase deficiency, 614307 (3)/Bile acid synthesis defect, congenital, 4, 214950 (3)
AMELX	Amelogenesis imperfecta, type 1E, 301200 (3)
AMELY	Oligozoospermia
AMER1	Osteopathia striata with cranial sclerosis, 300373 (3)
AMH	Persistent Mullerian duct syndrome, type I, 261550 (3)
AMHR2	Persistent Mullerian duct syndrome, type II, 261550 (3)
AMN	Megaloblastic anemia-1, Norwegian type, 261100 (3)
AMPD1	Myopathy due to myoadenylate deaminase deficiency, 615511 (3)
AMT	Glycine encephalopathy, 605899 (3)
ANG	Amyotrophic lateral sclerosis 9, 611895 (3)
ANGPTL3	Hypobetalipoproteinemia, familial, 2, 605019 (3)
ANK1	Spherocytosis, type 1, 182900 (3)
ANK2	Cardiac arrhythmia, ankyrin-B-related, 600919 (3)/Long QT syndrome 4, 600919 (3)
ANKH	Chondrocalcinosis 2, 118600 (3)/Craniometaphyseal dysplasia, 123000 (3)
ANKRD11	KBG syndrome, 148050 (3)
ANKRD26	Thrombocytopenia 2, 188000 (3)
ANO10	Spinocerebellar ataxia, autosomal recessive 10, 613728 (3)
ANO5	Gnathodiaphyseal dysplasia, 166260 (3)/Miyoshi muscular dystrophy 3, 613319 (3)/Muscular dystrophy, limb-girdle, type 2L, 611307 (3)
ANO6	Scott syndrome, 262890 (3)
ANTXR2	Hyaline fibromatosis syndrome, 228600 (3)
AP1S2	Mental retardation, X-linked syndromic 5, 304340 (3)
AP3B1	Hermansky-Pudlak syndrome 2, 608233 (3)
AP4B1	Spastic paraplegia 47, autosomal recessive, 614066 (3)
AP4E1	Spastic paraplegia 51, autosomal recessive, 613744 (3)
AP4M1	Spastic paraplegia 50, autosomal recessive, 612936 (3)
AP4S1	Spastic paraplegia 52, autosomal recessive, 614067 (3)
AP5Z1	Spastic paraplegia 48
APC	Adenoma, periampullary, somatic (3)/Adenomatous polyposis coli, 175100 (3)/Brain tumor-polyposis syndrome 2, 175100 (3)/Colorectal cancer, somatic, 114500 (3)/Desmoid disease, hereditary, 135290 (3)/Gardner syndrome, 175100 (3)/Gastric cancer, somatic, 613659 (3)/Hepatoblastoma, somatic, 114550 (3)
APCDD1	Hypotrichosis 1, 605389 (3)
APOA1	Amyloidosis, 3 or more types, 105200 (3)/ApoA-I and apoC-III deficiency, combined (3)/Corneal clouding, autosomal recessive (3)/Hypoalphalipoproteinemia, 604091 (3)
APOA4	Hyperlipidaemia
APOA5	Hyperchylomicronemia, late-onset, 144650 (3)/Hypertriglyceridemia, susceptibility to, 145750 (3)
APOB	Hypercholesterolemia, due to ligand-defective apo B, 144010 (3)/Hypobetalipoproteinemia, 615558 (3)
APOC2	Hyperlipoproteinemia, type Ib, 207750 (3)
APOC3	Apolipoprotein C-III deficiency, 614028 (3)
APOE	Alzheimer disease-2, 104310 (3)/Hyperlipoproteinemia, type III (3)/Lipoprotein glomerulopathy, 611771 (3)/Sea-blue histiocyte disease, 269600 (3)/
APOL1	End-stage renal disease, nondiabetic, susceptibility to, 612551 (3)/Glomerulosclerosis, focal segmental, 4, susceptibility to, 612551 (3)
APP	Alzheimer disease 1, familial, 104300 (3)/Cerebral amyloid angiopathy, Dutch, Italian, Iowa, Flemish, Arctic variants, 605714 (3)
APRT	Adenine phosphoribosyltransferase deficiency, 614723 (3)
APTX	Ataxia, early-onset, with oculomotor apraxia and hypoalbuminemia, 208920 (3)
AQP2	Diabetes insipidus, nephrogenic, 125800 (3)

AR	Androgen insensitivity, 300068 (3)/Androgen insensitivity, partial, with or without breast cancer, 312300 (3)/Hypospadias 1, X-linked, 300633 (3)/Spinal and bulbar muscular atrophy of Kennedy, 313200 (3)/Prostate cancer, susceptibility to, 176807 (3)
ARFGEF2	Periventricular heterotopia with microcephaly, 608097 (3)
ARG1	Argininemia, 207800 (3)
ARHGAP26	Leukemia, juvenile myelomonocytic, 607785 (3)
ARHGAP31	Adams-Oliver syndrome 1, 100300 (3)
ARHGEF10	Slowed nerve conduction velocity, AD, 608236 (3)
ARHGEF12	Leukemia, acute myeloid, 601626 (3)
ARHGEF6	Mental retardation, X-linked 46, 300436 (3)
ARHGEF9	Epileptic encephalopathy, early infantile, 8, 300607 (3)
ARID1A	Mental retardation, autosomal dominant 14, 614607 (3)
ARID1B	Mental retardation, autosomal dominant 12, 614562 (3)
ARL13B	Joubert syndrome 8, 612291 (3)
ARL6	Retinitis pigmentosa 55, 613575 (3)/Bardet-Biedl syndrome 3, 600151 (3)/Bardet-Biedl syndrome 1, modifier of, 209900 (3)
ARSA	Metachromatic leukodystrophy, 250100 (3)
ARSB	Mucopolysaccharidosis type VI (Maroteaux-Lamy), 253200 (3)
ARSE	Chondrodysplasia punctata, X-linked recessive, 302950 (3)
ARX	Epileptic encephalopathy, early infantile, 1, 308350 (3)/Hydranencephaly with abnormal genitalia, 300215 (3)/Lissencephaly, X-linked 2, 300215 (3)/Mental retardation, X-linked 29 and others, 300419 (3)/Partington syndrome, 309510 (3)/Proud syndrome, 300004 (3)
ASAH1	Farber lipogranulomatosis, 228000 (3)/Spinal muscular atrophy with progressive myoclonic epilepsy, 159950 (3)
ASCC1	Barrett esophagus/esophageal adenocarcinoma, 614266 (3)
ASCC3	Intellectual disability
ASCL1	Central hypoventilation syndrome, congenital, 209880 (3)/Haddad syndrome, 209880 (3)
ASL	Argininosuccinic aciduria, 207900 (3)
ASNS	Asparagine synthetase deficiency, 615574 (3)
ASPA	Canavan disease, 271900 (3)
ASPM	Microcephaly 5, primary, autosomal recessive, 608716 (3)
ASPSCR1	Alveolar soft-part sarcoma, 606243 (3)
ASS1	Citrullinemia, 215700 (3)
ASXL1	Bohring-Opitz syndrome, 605039 (3)/Myelodysplastic syndrome, somatic, 614286 (3)
ASXL3	Bainbridge-Ropers syndrome, 615485 (3)
ATCAY	Ataxia, cerebellar, Cayman type, 601238 (3)
ATIC	AICA-ribosiduria due to ATIC deficiency, 608688 (3)
ATL1	Neuropathy, hereditary sensory, type ID, 613708 (3)/Spastic paraplegia 3A, autosomal dominant, 182600 (3)
ATM	Ataxia-telangiectasia, 208900 (3)/Lymphoma, B-cell non-Hodgkin, somatic (3)/Lymphoma, mantle cell (3)/T-cell prolymphocytic leukemia, somatic (3)/Breast cancer, susceptibility to, 114480 (3)
ATN1	Dentatorubro-pallidoluysian atrophy, 125370 (3)
ATP13A2	Ceroid lipofuscinosis, neuronal, 12, 606693 (3)/Kufor-Rakeb syndrome, 606693 (3)
ATP1A2	Alternating hemiplegia of childhood, 104290 (3)/Migraine, familial basilar, 602481 (3)/Migraine, familial hemiplegic, 2, 602481 (3)
ATP1A3	Alternating hemiplegia of childhood 2, 614820 (3)/CAPOS syndrome, 601338 (3)/Dystonia-12, 128235 (3)
ATP2A1	Brody myopathy, 601003 (3)
ATP2A2	Acrokeratosis verruciformis, 101900 (3)/Darier disease, 124200 (3)
ATP2B3	Spinocerebellar ataxia, X-linked 1, 302500 (3)
ATP2C1	Hailey-Hailey disease, 169600 (3)
ATP5E	Mitochondrial complex V (ATP synthase) deficiency, nuclear type 3, 614053 (3)
ATP6AP2	Mental retardation, X-linked, syndromic, Hedera type, 300423 (3)/
ATP6V0A2	Cutis laxa, autosomal recessive, type IIA, 219200 (3)/Wrinkly skin syndrome, 278250 (3)
ATP6V0A4	Renal tubular acidosis, distal, autosomal recessive, 602722 (3)
ATP6V1B1	Renal tubular acidosis

ATP7A	Menkes disease, 309400 (3)/Occipital horn syndrome, 304150 (3)/Spinal muscular atrophy, distal, X-linked 3, 300489 (3)
ATP7B	Wilson disease, 277900 (3)
ATP8B1	Cholestasis, benign recurrent intrahepatic, 243300 (3)/Cholestasis, intrahepatic, of pregnancy, 1, 147480 (3)/Cholestasis, progressive familial intrahepatic 1, 211600 (3)
ATPAF2	Mitochondrial complex V (ATP synthase) deficiency, nuclear type 1, 604273 (3)
ATR	Cutaneous telangiectasia and cancer syndrome, familial, 614564 (3)/Seckel syndrome 1, 210600 (3)
ATRX	Alpha-thalassemia myelodysplasia syndrome, somatic, 300448 (3)/Alpha-thalassemia/mental retardation syndrome, 301040 (3)/Mental retardation-hypotonic facies syndrome, X-linked, 309580 (3)
ATXN1	Spinocerebellar ataxia 1, 164400 (3)
ATXN10	Spinocerebellar ataxia 10, 603516 (3)
ATXN2	Spinocerebellar ataxia 2, 183090 (3)/Amyotrophic lateral sclerosis, susceptibility to, 13, 183090 (3)
ATXN3	Machado-Joseph disease, 109150 (3)
ATXN7	Spinocerebellar ataxia 7, 164500 (3)
ATXN8	Spinocerebellar ataxia 8, 608768 (3)
AUH	3-methylglutaconic aciduria, type I, 250950 (3)
AURKC	Infertility, male
AVP	Diabetes insipidus, neurohypophyseal, 125700 (3)
AVPR2	Diabetes insipidus, nephrogenic, 304800 (3)/Nephrogenic syndrome of inappropriate antidiuresis, 300539 (3)
AXIN1	Caudal duplication anomaly, 607864 (3)/Hepatocellular carcinoma, somatic, 114550 (3)
AXIN2	Colorectal cancer, somatic, 114500 (3)/Oligodontia-colorectal cancer syndrome, 608615 (3)
B2M	Hypoproteinemia, hypercatabolic, 241600 (3)
B3GALTL	Peters-plus syndrome, 261540 (3)
B3GAT3	Multiple joint dislocations, short stature, craniofacial dysmorphism, and congenital heart defects, 245600 (3)
B4GALT1	Congenital disorder of glycosylation, type IId, 607091 (3)
B4GALT1-AS1	Congenital disorder of glycosylation, type IId 607091 (3)
B4GALT7	Ehlers-Danlos syndrome, progeroid type, 1, 130070 (3)
B9D1	Meckel syndrome 9, 614209 (3)
B9D2	Meckel syndrome 10, 614175 (3)
BAAT	Hypercholanemia, familial, 607748 (3)
BAG3	Cardiomyopathy, dilated, 1HH, 613881 (3)/Myopathy, myofibrillar, 6, 612954 (3)
BANF1	Nestor-Guillermo progeria syndrome, 614008 (3)
BAP1	Tumor predisposition syndrome, 614327 (3)
BBS1	Bardet-Biedl syndrome 1, 209900 (3)
BBS10	Bardet-Biedl syndrome 10, 615987 (3)
BBS12	Bardet-Biedl syndrome 12, 615989 (3)
BBS2	Bardet-Biedl syndrome 2, 615981 (3)
BBS4	Bardet-Biedl syndrome 4, 615982 (3)
BBS5	Bardet-Biedl syndrome 5, 615983 (3)
BBS7	Bardet-Biedl syndrome 7, 615984 (3)
BBS9	Bardet-Biedl syndrome
BCHE	Apnea, postanesthetic (3)
BCKDHA	Maple syrup urine disease, type Ia, 248600 (3)
BCKDHB	Maple syrup urine disease, type Ib, 248600 (3)
BCKDK	Branched-chain ketoacid dehydrogenase kinase deficiency, 614923 (3)
BCL10	Immunodeficiency 37, 616098 (3)/Lymphoma, MALT, somatic, 137245 (3)/Lymphoma, follicular, somatic, 613024 (3)/Male germ cell tumor, somatic, 273300, (3)/Mesothelioma, somatic, 156240 (3)/Sezary syndrome, somatic, (3)
BCL11A	Dias-Logan syndrome, 617101 (3)
BCL2	Leukemia/lymphoma, B-cell, 2 (3)
BCL3	Leukemia/lymphoma, B-cell, 3 (2)
BCMO1	Hypercarotenemia and vitamin A deficiency, autosomal dominant, 115300 (3)
BCOR	Microphthalmia, syndromic 2, 300166 (3)

BCR	Leukemia, acute lymphocytic, 613065 (3)/Leukemia, chronic myeloid, 608232 (3)
BCS1L	Bjornstad syndrome, 262000 (3)/GRACILE syndrome, 603358 (3)/Leigh syndrome, 256000 (3)/Mitochondrial complex III deficiency, nuclear type 1, 124000 (3)
BDNF	Central hypoventilation syndrome, congenital, 209880 (3)/Anorexia nervosa, susceptibility to, 610269 (3)/Bulimia nervosa, age of onset of weight loss in, 607499 (3)/Memory impairment, susceptibility to (3)/Obsessive-compulsive disorder, protection against, 164230 (3)
BEAN1	Spinocerebellar ataxia 31
BEST1	Bestrophinopathy, autosomal recessive, 611809 (3)/Microcornea, rod-cone dystrophy, cataract, and posterior staphyloma, 193220 (3)/Retinitis pigmentosa, concentric, 613194 (3)/Retinitis pigmentosa-50, 613194 (3)/Vitelliform macular dystrophy 2, 153700 (3)/Vitreoretinochoroidopathy, 193220 (3)
BFSP1	Cataract 33, 611391 (3)
BFSP2	Cataract 12, multiple types, 611597 (3)
BHLHA9	Syndactyly, mesoaxial synostotic, with phalangeal reduction, 609432 (3)
BIN1	Myopathy, centronuclear, autosomal recessive, 255200 (3)
BLK	Maturity-onset diabetes of the young, type 11, 613375 (3)
BLM	Bloom syndrome
BLNK	Agammaglobulinemia 4, 613502 (3)
BLOC1S3	Hermansky-Pudlak syndrome 8, 614077 (3)
BLOC1S6	Hermansky-pudlak syndrome 9, 614171 (3)
BLVRA	Hyperbiliverdinemia, 614156 (3)
BMP1	Osteogenesis imperfecta, type XIII, 614856 (3)
BMP15	Ovarian dysgenesis 2, 300510 (3)/Premature ovarian failure 4, 300510 (3)
BMP2	Brachydactyly, type A2, 112600 (3)/HFE hemochromatosis, modifier of, 235200 (3)
BMP4	Microphthalmia, syndromic 6, 607932 (3)/Orofacial cleft 11, 600625 (3)
BMPER	Diaphanospondylodysostosis, 608022 (3)
BMPR1A	Juvenile polyposis syndrome, infantile form, 174900 (3)/Polyposis syndrome, hereditary mixed, 2, 610069 (3)/Polyposis, juvenile intestinal, 174900 (3)
BMPR1B	Brachydactyly, type A2, 112600 (3)/Chrondrodysplasia, acromesomelic, with genital anomalies, 609441 (3)
BMPR2	Pulmonary hypertension, familial primary, 1, with or without HHT, 178600 (3)/Pulmonary hypertension, primary, fenfluramine or dexfenfluramine-associated, 178600 (3)/Pulmonary venoocclusive disease 1, 265450 (3)
BOLA3	Multiple mitochondrial dysfunctions syndrome 2, 614299 (3)
BPGM	Erythrocytosis due to bisphosphoglycerate mutase deficiency, 222800 (3)
BRAF	Adenocarcinoma of lung, somatic, 211980 (3)/Cardiofaciocutaneous syndrome, 115150 (3)/Colorectal cancer, somatic (3)/LEOPARD syndrome 3, 613707 (3)/Melanoma, malignant, somatic (3)/Nonsmall cell lung cancer, somatic (3)/Noonan syndrome 7, 613706 (3)
BRAT1	Rigidity and multifocal seizure syndrome, lethal neonatal, 614498 (3)
BRCA1	Breast-ovarian cancer, familial, 1, 604370 (3)/Pancreatic cancer, susceptibility to, 4, 614320 (3)
BRCA2	Fanconi anemia, complementation group D1, 605724 (3)/Pancreatic cancer, 613347 (3)/Prostate cancer, 176807 (3)/Wilms tumor, 194070 (3)/Breast cancer, male, susceptibility to, 114480 (3)/Breast-ovarian cancer, familial, 2, 612555 (3)/Glioblastoma 3, 613029 (3)/Medulloblastoma, 155255 (3)/Pre-B-cell acute lymphoblastic leukemia (3)
BRIP1	Breast cancer, early-onset, 114480 (3)/Fanconi anemia, complementation group J, 609054 (3)
BRWD3	Mental retardation, X-linked 93, 300659 (3)
BSCL2	Encephalopathy, progressive, with or without lipodystrophy, 615924 (3)/Lipodystrophy, congenital generalized, type 2, 269700 (3)/Neuropathy, distal hereditary motor, type VA, 600794 (3)/Silver spastic paraplegia syndrome, 270685 (3)
BSND	Bartter syndrome, type 4a, 602522 (3)/Sensorineural deafness with mild renal dysfunction, 602522 (3)
BTD	Biotinidase deficiency, 253260 (3)
BTK	Agammaglobulinemia and isolated hormone deficiency, 307200 (3)/Agammaglobulinemia, X-linked 1, 300755 (3)
BUB1B	Colorectal cancer, somatic, 114500 (3)/Mosaic variegated aneuploidy syndrome 1, 257300 (3)/[Premature chromatid separation trait], 176430 (3)

C10orf2	Mitochondrial DNA depletion syndrome 7 (hepatocerebral type) 271245 (3)/Perrault syndrome 5 616138 (3)/Progressive external ophthalmoplegia with mitochondrial DNA deletions, autosomal dominant 3 609286 (3)
C11ORF46	Intellectual disability
C12ORF57	Temtamy syndrome, 218340 (3)
C12ORF65	Combined oxidative phosphorylation deficiency 7, 613559 (3)/Spastic paraplegia 55, autosomal recessive, 615035 (3)
C14ORF133	Arthrogryposis, renal dysfunction, and cholestasis 2, 613404 (3)
C19ORF12	Spastic paraplegia 43, autosomal recessive, 615043 (3)/Neurodegeneration with brain iron accumulation 4, 614298 (3)
C1GALT1C1	Tn polyagglutination syndrome, somatic, 300622 (3)
C1QA	C1q deficiency, 613652 (3)
C1QB	C1q deficiency, 613652 (3)
C1QC	C1q deficiency, 613652 (3)
C1QTNF5	Retinal degeneration, late-onset, autosomal dominant, 605670 (3)
C1R	C1r/C1s deficiency, combined, 216950 (1)
C1S	C1s deficiency, 613783 (3)
C2	C2 deficiency, 217000 (3)/Macular degeneration, age-related, 14, reduced risk of, 615489 (3)
C20ORF7	Complex I deficiency
C2ORF71	Retinitis pigmentosa 54, 613428 (3)
C3	C3 deficiency, 613779 (3)/Hemolytic uremic syndrome, atypical, susceptibility to, 5, 612925 (3)/Macular degeneration, age-related, 9, 611378 (3)
C4A	Systemic lupus erythematosus, susceptibility to or protection against, 152700 (2)/C4a deficiency, 614380 (3)/[Blood group, Rodgers], 614374 (3)
C4B	C4B deficiency 614379 (3)
C4ORF26	Amelogenesis imperfecta, type IIA4, 614832 (3)
C5	C5 deficiency, 609536 (3)/[Eculizumab, poor response to], 615749 (3)
C5ORF42	Joubert syndrome 17, 614615 (3)/Orofaciodigital syndrome VI, 277170 (3)
C6	C6 deficiency, 612446 (3)/Combined C6/C7 deficiency (3)
C7	C7 deficiency, 610102 (3)
C8A	C8 deficiency, type I, 613790 (3)
C8B	C8 deficiency, type II, 613789 (3)
C8ORF37	Cone-rod dystrophy 16, 614500 (3)/Retinitis pigmentosa 64, 614500 (3)
C8ORF38	Complex I deficiency
C9	C9 deficiency, 613825 (3)/Macular degeneration, age-related, 15, suscepbitility to, 615591 (3)
C9ORF72	Amyotrophic lateral sclerosis and/or frontotemporal dementia, 105550 (3)
C9ORF86	Intellectual disability
CA12	Hyperchlorhidrosis, isolated, 143860 (3)
CA2	Osteopetrosis, autosomal recessive 3, with renal tubular acidosis, 259730 (3)
CA4	Retinitis pigmentosa 17, 600852 (3)
CA8	Cerebellar ataxia and mental retardation with or without quadrupedal locomotion 3, 613227 (3)
CABP2	Deafness, autosomal recessive 93, 614899 (3)
CABP4	Night blindness, congenital stationary (incomplete), 2B, autosomal recessive, 610427 (3)
CACNA1A	Episodic ataxia, type 2, 108500 (3)/Migraine, familial hemiplegic, 1, 141500 (3)/Migraine, familial hemiplegic, 1, with progressive cerebellar ataxia, 141500 (3)/Spinocerebellar ataxia 6, 183086 (3)
CACNA1C	Brugada syndrome 3, 611875 (3)/Timothy syndrome, 601005 (3)
CACNA1D	Primary aldosteronism, seizures, and neurologic abnormalities, 615474 (3)/Sinoatrial node dysfunction and deafness, 614896 (3)
CACNA1F	Aland Island eye disease, 300600 (3)/Cone-rod dystropy, X-linked, 3, 300476 (3)/Night blindness, congenital stationary (incomplete), 2A, X-linked, 300071 (3)
CACNA1G	Myoclonic epilepsy, juvenile
CACNA1H	Epilepsy, childhood absence, susceptibility to, 6, 611942 (3)/Epilepsy, idiopathic generalized, susceptibility to, 6, 611942 (3)

CACNA1S	Hypokalemic periodic paralysis, type 1, 170400 (3)/Malignant hyperthermia susceptibility 5, 601887 (3)/Thyrotoxic periodic paralysis, susceptibility to, 1, 188580 (3)
CACNA2D4	Retinal cone dystrophy 4, 610478 (3)
cacnb2	Brugada syndrome
CACNB4	Episodic ataxia, type 5, 613855 (3)/Epilepsy, idiopathic generalized, susceptibility to, 9, 607682 (3)/Epilepsy, juvenile myoclonic, susceptibility to, 6, 607682 (3)
CACNG2	Mental retardation, autosomal dominant 10, 614256 (3)
CALM1	Ventricular tachycardia, catecholaminergic polymorphic, 4, 614916 (3)
CALR3	Cardiomyopathy, familial hypertrophic, 19, 613875 (3)
CAMTA1	Cerebellar ataxia, nonprogressive, with mental retardation, 614756 (3)
CANT1	Desbuquois dysplasia, 251450 (3)
CAPN10	Diabetes mellitus, noninsulin-dependent 1, 601283 (3)
CAPN3	Muscular dystrophy, limb-girdle, type 2A, 253600 (3)
CARD14	Pityriasis rubra pilaris, 173200 (3)/Psoriasis 2, 602723 (3)
CARD9	Candidiasis, familial, 2, autosomal recessive, 212050 (3)
CASC5	Microcephaly 4, primary, autosomal recessive, 604321 (3)
CASK	FG syndrome 4, 300422 (3)/Mental retardation and microcephaly with pontine and cerebellar hypoplasia, 300749 (3)/Mental retardation, with or without nystagmus, 300422 (3)
CASP10	Autoimmune lymphoproliferative syndrome, type II, 603909 (3)/Gastric cancer, somatic, 613659 (3)/Non-Hodgkin lymphoma, somatic, 605027 (3)
CASP2	Intellectual disability
CASP8	Autoimmune lymphoproliferative syndrome, type IIB, 607271 (3)/Hepatocellular carcinoma, somatic, 114550 (3)/Breast cancer, protection against, 114480 (3)/Lung cancer, protection against, 211980 (3)
CASQ2	Ventricular tachycardia, catecholaminergic polymorphic, 2, 611938 (3)
CASR	Hypercalciuric hypercalcemia (3)/Hyperparathyroidism, neonatal, 239200 (3)/Hypocalcemia, autosomal dominant, 601198 (3)/Hypocalcemia, autosomal dominant, with Bartter syndrome, 601198 (3)/Hypocalciuric hypercalcemia, type I, 145980 (3)/Calcium, serum level of (3)/Epilepsy idiopathic generalized, susceptibility to, 8, 612899 (3)
CAT	Acatalasemia, 614097 (3)
CATSPER1	Spermatogenic failure 7, 612997 (3)
CAV1	Lipodystrophy, congenital generalized, type 3, 612526 (3)/
CAV3	Cardiomyopathy, familial hypertrophic, 192600 (3)/Creatine phosphokinase, elevated serum, 123320 (3)/Long QT syndrome 9, 611818 (3)/Muscular dystrophy, limb-girdle, type IC, 607801 (3)/Myopathy, distal, Tateyama type, 614321 (3)/Rippling muscle disease, 606072 (3)
CBFB	Myeloid leukemia, acute, M4/M4Eo subtype, somatic, 601626 (1)
CBL	Noonan syndrome-like disorder with or without juvenile myelomonocytic leukemia, 613563 (3)
CBS	Homocystinuria, B6-responsive and nonresponsive types, 236200 (3)/Thrombosis, hyperhomocysteinemic, 236200 (3)
CBX2	46XY sex reversal 5, 613080 (3)
CC2D1A	Mental retardation, autosomal recessive 3, 608443 (3)
CC2D2A	COACH syndrome, 216360 (3)/Joubert syndrome 9, 612285 (3)/Meckel syndrome 6, 612284 (3)
CCBE1	Hennekam lymphangiectasia-lymphedema syndrome 1, 235510 (3)
CCDC103	Ciliary dyskinesia, primary, 17, 614679 (3)
CCDC11	Heterotaxy, visceral, 6, autosomal recessive, 614779 (3)
CCDC39	Ciliary dyskinesia, primary, 14, 613807 (3)
CCDC39	Ciliary dyskinesia, primary, 14 613807 (3)
CCDC40	Ciliary dyskinesia, primary, 15, 613808 (3)
CCDC50	Deafness, autosomal dominant 44, 607453 (3)
CCDC6	Thyroid papillary carcinoma, 188550 (1)
CCDC78	Myopathy, centronuclear, 4, 614807 (3)
CCDC8	3-M syndrome 3, 614205 (3)
CCDC88C	Spinocerebellar ataxia 40, 616053 (3)/Hydrocephalus, nonsyndromic, autosomal recessive, 236600 (3)
CCL11	Asthma, susceptibility to, 600807 (3)/HIV1, resistance to, 609423 (3)
CCM2	Cerebral cavernous malformations

CCND1	Colorectal cancer, susceptibility to, 114500 (3)/Multiple myeloma, susceptibility to, 254500 (3)/von Hippel-Lindau syndrome, modifier of, 193300 (3)
CCT5	Neuropathy, hereditary sensory, with spastic paraplegia, 256840 (3)
CD151	Nephropathy with pretibial epidermolysis bullosa and deafness, 609057 (3)/[Blood group, Raph], 179620 (3)
CD19	Immunodeficiency, common variable, 3, 613493 (3)
CD247	Immunodeficiency 25, 610163 (3)
CD2AP	Glomerulosclerosis, focal segmental, 3, 607832 (3)
CD320	Methylmalonic aciduria due to transcobalamin receptor defect, 613646 (3)
CD36	Platelet glycoprotein IV deficiency, 608404 (3)/[Macrothrombocytopenia] (1)/Coronary heart disease, susceptibility to, 7, 610938 (3)/Malaria, cerebral, reduced risk of, 611162 (3)/Malaria, cerebral, susceptibility to, 611162 (3)
CD3D	Immunodeficiency 19, 615617 (3)
CD3E	Immunodeficiency 18, 615615 (3)/Immunodeficiency 18, SCID variant, 615615 (3)
CD3G	Immunodeficiency 17, CD3 gamma deficient, 615607 (3)
CD4	OKT4 epitope deficiency, 613949 (3)
CD40	Immunodeficiency with hyper-IgM, type 3, 606843 (3)
CD40LG	Hyper-IgM syndrome
CD59	Hemolytic anemia, CD59-mediated, with or without immune-mediated polyneuropathy, 612300 (3)
CD79A	Agammaglobulinemia 3, 613501 (3)
CD79B	Agammaglobulinemia 6, 612692 (3)
CD81	Immunodeficiency, common variable, 6, 613496 (3)
CD81-AS1	Immunodeficiency, common variable, 6 613496 (3)
CD82	Prostate cancer, susceptibility to, 176807 (2)
CD8A	CD8 deficiency, familial, 608957 (3)
CD96	C syndrome, 211750 (3)
CDAN1	Dyserythropoietic anemia, congenital, type Ia, 224120 (3)
CDC6	Meier-Gorlin syndrome 5, 613805 (3)
CDC73	Hyperparathyroidism-jaw tumour syndrome
CDH1	Endometrial carcinoma, somatic, 608089 (3)/Gastric cancer, familial diffuse, with or without cleft lip and/or palate, 137215 (3)/Ovarian carcinoma, somatic, 167000 (3)/Breast cancer, lobular, 114480 (3)/Prostate cancer, susceptibility to, 176807 (3)
CDH15	Mental retardation, autosomal dominant 3, 612580 (3)
CDH23	Deafness, autosomal recessive 12, 601386 (3)/Usher syndrome, type 1D, 601067 (3)/Usher syndrome, type 1D/F digenic, 601067 (3)
CDH3	Ectodermal dysplasia, ectrodactyly, and macular dystrophy, 225280 (3)/Hypotrichosis, congenital, with juvenile macular dystrophy, 601553 (3)
CDHR1	Cone-rod dystrophy 15, 613660 (3)/Retinitis pigmentosa 65, 613660 (3)
CDK4	Melanoma, cutaneous malignant, 3, 609048 (3)
CDK5RAP2	Microcephaly 3, primary, autosomal recessive, 604804 (3)
CDKL5	Angelman syndrome-like, 105830 (3)/Epileptic encephalopathy, early infantile, 2, 300672 (3)
CDKN1B	Multiple endocrine neoplasia, type IV, 610755 (3)
CDKN1C	Beckwith-Wiedemann syndrome, 130650 (3)/IMAGE syndrome, 614732 (3)
CDKN2A	Melanoma
CDON	Holoprosencephaly 11, 614226 (3)
CDSN	Hypotrichosis 2, 146520 (3)/Peeling skin syndrome 1, 270300 (3)
CDT1	Meier-Gorlin syndrome 4, 613804 (3)
CEACAM16	Deafness, autosomal dominant 4B, 614614 (3)
CEBPA	Leukemia, acute myeloid, 601626 (3)/Leukemia, acute myeloid, somatic, 601626 (3)
CEBPE	Specific granule deficiency, 245480 (3)
CEL	Maturity-onset diabetes of the young, type VIII, 609812 (3)
CENPJ	Microcephaly 6, primary, autosomal recessive, 608393 (3)/Seckel syndrome 4, 613676 (3)
CEP135	Microcephaly 8, primary, autosomal recessive, 614673 (3)
CEP152	Microcephaly 9, primary, autosomal recessive, 614852 (3)/Seckel syndrome 5, 613823 (3)
CEP164	Nephronophthisis 15, 614845 (3)

CEP290	Bardet-Biedl syndrome 14, 615991 (3)/Joubert syndrome 5, 610188 (3)/Leber congenital amaurosis 10, 611755 (3)/Meckel syndrome 4, 611134 (3)/Senior-Loken syndrome 6, 610189 (3)
CEP41	Joubert syndrome 15, 614464 (3)
CEP57	Mosaic variegated aneuploidy syndrome 2, 614114 (3)
CEP63	Seckel syndrome 6, 614728 (3)
CERKL	Retinitis pigmentosa 26, 608380 (3)
CES1	Carboxylesterase 1 deficiency (3)
CETP	Hyperalphalipoproteinemia,143470 (3)/[High density lipoprotein cholesterol level QTL 10], 143470 (3)
CFC1	Double-outlet right ventricle, 217095 (3)/Heterotaxy, visceral, 2, autosomal, 605376 (3)/Transposition of the great arteries, dextro-looped 2, 613853 (3)
CFD	Complement factor D deficiency, 613912 (3)
CFH	Haemolytic uraemic syndrome, atypical
CFHR2	Lowe C3 plasma levels & Factor B activation
CFHR5	Nephropathy due to CFHR5 deficiency, 614809 (3)
CFI	Complement factor I deficiency, 610984 (3)/Hemolytic uremic syndrome, atypical, susceptibility to, 3, 612923 (3)/Macular degeneration,age-related, 13, susceptibility to, 615439 (3)
CFL2	Nemaline myopathy 7, autosomal recessive, 610687 (3)
CFP	Properdin deficiency
CFTR	Congenital bilateral absence of vas deferens, 277180 (3)/Cystic fibrosis, 219700 (3)/Sweat chloride elevation without CF (3)/Bronchiectasis with or without elevated sweat chloride 1, modifier of, 211400 (3)/Hypertrypsinemia, neonatal (3)/Pancreatitis, idiopathic, 167800 (3)
CHAT	Myasthenic syndrome, congenital, associated with episodic apnea, 254210 (3)
CHD2	Epileptic encephalopathy, childhood-onset, 615369 (3)
CHD7	CHARGE syndrome, 214800 (3)/Hypogonadotropic hypogonadism 5 with or without anosmia, 612370 (3)/Scoliosis, idiopathic 3, 608765 (3)
CHEK2	Li-Fraumeni syndrome, 609265 (3)/Osteosarcoma, somatic, 259500 (3)/Breast and colorectal cancer, susceptibility to (3)/Breast cancer, susceptibility to, 114480 (3)/Prostate cancer, familial, susceptibility to, 176807 (3)
CHKB	Muscular dystrophy, congenital, megaconial type, 602541 (3)
CHM	Choroideremia, 303100 (3)
CHMP1A	Pontocerebellar hypoplasia, type 8, 614961 (3)
CHMP2B	Amyotrophic lateral sclerosis 17, 614696 (3)/Dementia, familial, nonspecific, 600795 (3)
CHMP4B	Cataract 31, multiple types, 605387 (3)
CHN1	Duane retraction syndrome 2, 604356 (3)
CHRDL1	Megalocornea 1, X-linked 309300 (3)
CHRM2	Cardiomyopathy, dilated
CHRM3	Eagle-Barrett syndrome, 100100 (3)
CHRNA1	Multiple pterygium syndrome, lethal type, 253290 (3)/Myasthenic syndrome, fast-channel congenital, 608930 (3)/Myasthenic syndrome, slow-channel congenital, 601462 (3)
CHRNA2	Epilepsy, nocturnal frontal lobe, type 4, 610353 (3)
CHRNA4	Epilepsy, nocturnal frontal lobe, 1, 600513 (3)/Nicotine addiction, susceptibility to, 188890 (3)
CHRNA7	Schizophrenia, neurophysiologic defect in (2)
CHRNB1	Myasthenic syndrome, congenital, associated with acetylcholine receptor deficiency, 608931 (3)/Myasthenic syndrome, slow-channel congenital, 601462 (3)
CHRNB2	Epilepsy, nocturnal frontal lobe, 3, 605375 (3)
CHRND	Multiple pterygium syndrome, lethal type, 253290 (3)/Myasthenic syndrome, fast-channel congenital, 608930 (3)/Myasthenic syndrome, slow-channel congenital, 601462 (3)
CHRNE	Myasthenic syndrome, congenital, associated with acetylcholine receptor deficiency, 608931 (3)/Myasthenic syndrome, fast-channel congenital, 608930 (3)/Myasthenic syndrome, slow-channel congenital, 601462 (3)
CHRNG	Escobar syndrome, 265000 (3)/Multiple pterygium syndrome, lethal type, 253290 (3)/Myasthenia gravis, neonatal transient (2)
CHST14	Ehlers-Danlos syndrome, musculocontractural type 1, 601776 (3)
CHST3	Spondyloepiphyseal dysplasia with congenital joint dislocations, 143095 (3)
CHST6	Macular corneal dystrophy, 217800 (3)

CHSY1	Temtamy preaxial brachydactyly syndrome, 605282 (3)
CHUK	Cocoon syndrome, 613630 (3)
CIB2	Deafness, autosomal recessive 48, 609439 (3)/Usher syndrome, type IJ, 614869 (3)
CIITA	Bare lymphocyte syndrome, type II, complementation group A (3)
CIRH1A	Cirrhosis, North American Indian childhood type, 604901 (3)
CISD2	Wolfram syndrome 2, 604928 (3)
CITED2	Atrial septal defect 8, 614433 (3)/Ventricular septal defect 2, 614431 (3)
CLCF1	Cold-induced sweating syndrome 2, 610313 (3)
CLCN1	Myotonia congenita, dominant, 160800 (3)/Myotonia congenita, recessive, 255700 (3)/Myotonia levior, recessive (3)
CLCN2	Leukoencephalopathy with ataxia, 615651 (3)/Epilepsy, idiopathic generalized, susceptibility to, 11, 607628 (3)/Epilepsy, juvenile absence, susceptibility to, 2, 607628 (3)/Epilepsy, juvenile myoclonic, susceptibility to, 8, 607628 (3)
CLCN4	Epileptic encephalopathy
CLCN5	Dent disease, 300009 (3)/Hypophosphatemic rickets, 300554 (3)/Nephrolithiasis, type I, 310468 (3)/Proteinuria, low molecular weight, with hypercalciuric nephrocalcinosis, 308990 (3)
CLCN7	Osteopetrosis, autosomal dominant 2, 166600 (3)/Osteopetrosis, autosomal recessive 4, 611490 (3)
CLCNKA	Bartter syndrome, type 4b, digenic, 613090 (3)
CLCNKB	Bartter syndrome, type 3, 607364 (3)/Bartter syndrome, type 4b, digenic, 613090 (3)
CLDN1	Ichthyosis, leukocyte vacuoles, alopecia, and sclerosing cholangitis, 607626 (3)
CLDN14	Deafness, autosomal recessive 29, 614035 (3)
CLDN16	Hypomagnesemia 3, renal, 248250 (3)
CLDN19	Hypomagnesemia 5, renal, with ocular involvement, 248190 (3)
CLEC7A	Candidiasis, familial, 4, autosomal recessive, 613108 (3)/Aspergillosis, susceptibility to, 614079 (3)
CLIC2	Mental retardation, X-linked, syndromic 32, 300886 (3)
CLMP	Congenital short bowel syndrome, 615237 (3)
CLN3	Ceroid lipofuscinosis, neuronal, 3, 204200 (3)
CLN5	Ceroid lipofuscinosis, neuronal, 5, 256731 (3)
CLN6	Ceroid lipofuscinosis, neuronal, 6, 601780 (3)/Ceroid lipofuscinosis, neuronal, Kufs type, adult onset, 204300 (3)
CLN8	Ceroid lipofuscinosis, neuronal, 8, 600143 (3)/Ceroid lipofuscinosis, neuronal, 8, Northern epilepsy variant, 610003 (3)
CLRN1	Retinitis pigmentosa 61, 614180 (3)/Usher syndrome, type 3A, 276902 (3)
CNBP	Myotonic dystrophy
CNGA1	Retinitis pigmentosa 49, 613756 (3)
CNGA3	Achromatopsia-2, 216900 (3)
CNGB1	Retinitis pigmentosa 45, 613767 (3)
CNGB3	Achromatopsia-3, 262300 (3)/Macular degeneration, juvenile, 248200 (3)
CNKSR1	Intellectual disability
CNKSR2	Intellectual disability, X-linked non-syndromic
CNNM2	Hypomagnesemia 6, renal, 613882 (3)
CNNM4	Jalili syndrome, 217080 (3)
CNTN1	Myopathy, congenital, Compton-North, 612540 (3)
CNTNAP2	Cortical dysplasia-focal epilepsy syndrome, 610042 (3)/Pitt-Hopkins like syndrome 1, 610042 (3)/Autism susceptibility 15, 612100 (3)
COA5	Mitochondrial complex IV deficiency, 220110 (3)
COCH	Deafness, autosomal dominant 9, 601369 (3)
COG1	Congenital disorder of glycosylation, type IIg, 611209 (3)
COG4	Congenital disorder of glycosylation, type IIj, 613489 (3)
COG5	Congenital disorder of glycosylation, type IIi, 613612 (3)
COG6	Congenital disorder of glycosylation, type IIl, 614576 (3)/Shaheen syndrome, 615328 (3)
COG7	Congenital disorder of glycosylation, type IIe, 608779 (3)
COG8	Congenital disorder of glycosylation, type IIh, 611182 (3)
COL10A1	Metaphyseal chondrodysplasia, Schmid type, 156500 (3)

COL11A1	Fibrochondrogenesis, 228520 (3)/Marshall syndrome, 154780 (3)/Stickler syndrome, type II, 604841 (3)/Lumbar disc herniation, susceptibility to, 603932 (3)
COL11A2	Deafness, autosomal dominant 13, 601868 (3)/Deafness, autosomal recessive 53, 609706 (3)/Fibrochondrogenesis 2, 614524 (3)/Otospondylomegaepiphyseal dysplasia, 215150 (3)/Stickler syndrome, type III, 184840 (3)/Weissenbacher-Zweymuller syndrome, 277610 (3)
COL14A1	Keratoderma, palmoplantar, punctate
COL17A1	Epidermolysis bullosa, junctional, non-Herlitz type, 226650 (3)
COL18A1	Knobloch syndrome, type 1, 267750 (3)
COL1A1	Caffey disease, 114000 (3)/Ehlers-Danlos syndrome, type I, 130000 (3)/Ehlers-Danlos syndrome, type VIIA, 130060 (3)/OI type II, 166210 (3)/OI type III, 259420 (3)/OI type IV, 166220 (3)/Osteogenesis imperfecta, type I, 166200 (3)/[Bone mineral density variation QTL], 166710 (3)/Osteoporosis, 166710 (3)
COL1A2	Ehlers-Danlos syndrome, cardiac valvular form, 225320 (3)/Ehlers-Danlos syndrome, type VIIB, 130060 (3)/Osteogenesis imperfecta, type II, 166210 (3)/Osteogenesis imperfecta, type III, 259420 (3)/Osteogenesis imperfecta, type IV, 166220 (3)/Osteoporosis, postmenopausal, 166710 (3)
COL2A1	Achondrogenesis, type II or hypochondrogenesis, 200610 (3)/Avascular necrosis of the femoral head, 608805 (3)/Czech dysplasia, 609162 (3)/Epiphyseal dysplasia, multiple, with myopia and deafness, 132450 (3)/Kniest dysplasia, 156550 (3)/Legg-Calve-Perthes disease, 150600 (3)/Osteoarthritis with mild chondrodysplasia, 604864 (3)/Otospondylomegaepiphyseal dysplasia, 215150 (3)/Platyspondylic skeletal dysplasia, Torrance type, 151210 (3)/SED congenita, 183900 (3)/SED, Namaqualand type (3)/SMED Strudwick type, 184250 (3)/Spondyloperipheral dysplasia, 271700 (3)/Stickler sydrome, type I, nonsyndromic ocular, 609508 (3)/Stickler syndrome, type I, 108300 (3)/Vitreoretinopathy with phalangeal epiphyseal dysplasia (3)
COL3A1	Ehlers-Danlos syndrome, type III, 130020 (3)/Ehlers-Danlos syndrome, type IV, 130050 (3)
COL4A1	Angiopathy, hereditary, with nephropathy, aneurysms, and muscle cramps, 611773 (3)/Brain small vessel disease with or without ocular anomalies, 607595 (3)/Porencephaly 1, 175780 (3)/Hemorrhage, intracerebral, susceptibility to, 614519 (3)
COL4A2	Porencephaly 2, 614483 (3)/Hemorrhage, intracerebral, susceptibility to, 614519 (3)
COL4A3	Alport syndrome, autosomal dominant, 104200 (3)/Alport syndrome, autosomal recessive, 203780 (3)/Hematuria, benign familial, 141200 (3)
COL4A3BP	Mental retardation, autosomal dominant 34, 616351 (3)
COL4A4	Alport syndrome, autosomal recessive, 203780 (3)/Hematuria, familial benign (3)
COL4A5	Alport syndrome, 301050 (3)
COL4A6	Deafness, X-linked 6, 300914 (3)
COL5A1	Ehlers-Danlos syndrome, type I, 130000 (3)/Ehlers-Danlos syndrome, type II, 130010 (3)
COL5A2	Ehlers-Danlos syndrome, type I, 130000 (3)
COL6A1	Bethlem myopathy, 158810 (3)/Ullrich congenital muscular dystrophy, 254090 (3)
COL6A2	Myosclerosis, congenital, 255600 (3)/Bethlem myopathy, 158810 (3)/Ullrich congenital muscular dystrophy, 254090 (3)
COL6A3	Bethlem myopathy, 158810 (3)/Ullrich congenital muscular dystrophy, 254090 (3)
COL7A1	EBD inversa, 226600 (3)/EBD, Bart type, 132000 (3)/EBD, localisata variant (3)/Epidermolysis bullosa dystrophica, AD, 131750 (3)/Epidermolysis bullosa dystrophica, AR, 226600 (3)/Epidermolysis bullosa pruriginosa, 604129 (3)/Epidermolysis bullosa, pretibial, 131850 (3)/Toenail dystrophy, isolated, 607523 (3)/Transient bullous of the newborn, 131705 (3)
COL8A2	Corneal dystrophy, Fuchs endothelial, 1, 136800 (3)/Corneal dystrophy, posterior polymorphous 2, 609140 (3)
COL9A1	Epiphyseal dysplasia, multiple, 6, 614135 (3)/Stickler syndrome, type IV, 614134 (3)
COL9A2	Stickler syndrome, type V, 614284 (3)/Epiphyseal dysplasia, multiple, 2, 600204 (3)/Intervertebral disc disease, susceptibility to, 603932 (3)
COL9A3	Epiphyseal dysplasia, multiple, 3, 600969 (3)/Epiphyseal dysplasia, multiple, with myopathy (3)/Intervertebral disc disease, susceptibility to, 603932 (3)
COLEC11	3MC syndrome 2, 265050 (3)
COLQ	Endplate acetylcholinesterase deficiency, 603034 (3)
COMP	Epiphyseal dysplasia, multiple 1, 132400 (3)/Pseudoachondroplasia, 177170 (3)
COMT	Panic disorder, susceptibility to, 167870 (3)/Schizophrenia, susceptibility to, 181500 (3)

COPA	{Autoimmune interstitial lung, joint, and kidney disease} 616414 (3)
COQ2	Coenzyme Q10 deficiency, primary, 1, 607426 (3)/Multiple system atrophy, susceptibility to, 146500 (3)
COQ5	Intellectual disability
COQ6	Coenzyme Q10 deficiency, primary, 6, 614650 (3)
COQ9	Coenzyme Q10 deficiency, primary, 5, 614654 (3)
CORIN	Preeclampsia/eclampsia 5, 614595 (3)
CORO1A	Immunodeficiency 8, 615401 (3)
COX10	Leigh syndrome due to mitochondrial COX4 deficiency, 256000 (3)/Mitochondrial complex IV deficiency, 220110 (3)
COX14	Lactic acidosis, fatal neonatal
COX15	Cardioencephalomyopathy, fatal infantile, due to cytochrome c oxidase deficiency 2, 615119 (3)/Leigh syndrome due to cytochrome c oxidase deficiency, 256000 (3)
COX4I2	Exocrine pancreatic insufficiency, dyserythropoietic anemia, and calvarial hyperostosis, 612714 (3)
COX6B1	Mitochondrial complex IV deficiency, 220110 (3)
COX7B	Aplasia cutis congenita, reticulolinear, with mmicrocephaly, facial dysmorphism and other congenital anomalies, 300887 (3)
CP	Cerebellar ataxia, 604290 (3)/Hemosiderosis, systemic, due to aceruloplasminemia, 604290 (3)/[Hypoceruloplasminemia, hereditary], 604290 (3)
CPA6	Epilepsy, familial temporal lobe, 5, 614417 (3)/Febrile seizures, familial, 11, 614418 (3)
CPN1	Carboxypeptidase N deficiency, 212070 (3)
CPOX	Coproporphyria, 121300 (3)/Harderoporphyria, 121300 (3)
CPS1	Carbamoylphosphate synthetase I deficiency, 237300 (3)/Pulmonary hypertension, neonatal, susceptibility to, 615371 (3)/Venoocclusive disease after bone marrow transplantation (3)
CPT1A	CPT deficiency, hepatic, type IA, 255120 (3)
CPT2	CPT II deficiency, lethal neonatal, 608836 (3)/CPT deficiency, hepatic, type II, 600649 (3)/Myopathy due to CPT II deficiency, 255110 (3)/Encephalopathy, acute, infection-induced, 4, susceptibility to, 614212 (3)
CR2	Immunodeficiency, common variable, 7, 614699 (3)/Systemic lupus erythematosus, susceptibility to, 9, 610927 (3)
CRADD	Mental retardation, autosomal recessive 34, 614499 (3)
CRB1	Leber congenital amaurosis 8, 613835 (3)/Pigmented paravenous chorioretinal atrophy, 172870 (3)/Retinitis pigmentosa-12, autosomal recessive, 600105 (3)
CRBN	Mental retardation, autosomal recessive 2, 607417 (3)
CREB1	Histiocytoma, angiomatoid fibrous, somatic, 612160 (3)
CREBBP	Rubinstein-Taybi syndrome, 180849 (3)
CRELD1	Atrioventricular septal defect, partial, with heterotaxy syndrome, 606217 (3)/Atrioventricular septal defect, susceptibility to, 2, 606217 (3)
CRH	Nocturnal frontal lobe epilepsy, autosomal dominant
CRLF1	Cold-induced sweating syndrome 1, 272430 (3)
CRTAP	Osteogenesis imperfecta, type VII, 610682 (3)
CRX	Cone-rod retinal dystrophy-2, 120970 (3)/Leber congenital amaurosis 7, 613829 (3)
CRYAB	Cardiomyopathy, dilated, 1II, 615184 (3)/Cataract 16, multiple types, 613763 (3)/Myopathy, myofibrillar, 2, 608810 (3)/Myopathy, myofibrillar, fatal infantile hypertrophy, alpha-B crystallin-related, 613869 (3)
CRYBA1	Cataract 10, multiple types, 600881 (3)
CRYBA4	Cataract 23, 610425 (3)
CRYBB1	Cataract 17, multiple types, 611544 (3)
CRYBB2	Cataract 3, multiple types, 601547 (3)
CRYBB3	Cataract 22, autosomal recessive, 609741 (3)
CRYGC	Cataract 2, multiple types, 604307 (3)
CRYGD	Cataract 4, multiple types, 115700 (3)
CSF1R	Leukoencephalopathy, diffuse hereditary, with spheroids, 221820 (3)
CSF2RA	Surfactant metabolism dysfunction, pulmonary, 4, 300770 (3)
CSF2RB	Surfactant metabolism dysfunction, pulmonary, 5, 614370 (3)
CSF3R	Neutrophilia, hereditary, 162830 (3)
CSRP3	Cardiomyopathy, dilated, 1M, 607482 (3)/Cardiomyopathy, familial hypertrophic, 12, 612124 (3)

CST3	Cerebral amyloid angiopathy, 105150 (3)/Macular degeneration, age-related, 11, 611953 (3)
CSTA	Exfoliative ichthyosis, autosomal recessive, ichthyosis bullosa of Siemens-like, 607936 (3)
CSTB	Epilepsy, progressive myoclonic 1A (Unverricht and Lundborg), 254800 (3)
CTC1	Cerebroretinal microangiopathy with calcifications and cysts, 612199 (3)
CTDP1	Congenital cataracts, facial dysmorphism, and neuropathy, 604168 (3)
CTH	Cystathioninuria, 219500 (3)/Homocysteine, total plasma, elevated (3)
CTHRC1	Barrett esophagus/esophageal adenocarcinoma, 614266 (3)
CTLA4	Autoimmune lymphoproliferative syndrome, type V, 616100 (3)
CTNNB1	Colorectal cancer, somatic, 114500 (3)/Hepatocellular carcinoma, somatic, 114550 (3)/Mental retardation, autosomal dominant 19, 615075 (3)/Ovarian cancer, somatic, 167000 (3)/Pilomatricoma, somatic, 132600
CTNS	Cystinosis, atypical nephropathic, 219800 (3)/Cystinosis, late-onset juvenile or adolescent nephropathic, 219900 (3)/Cystinosis, nephropathic, 219800 (3)/Cystinosis, ocular nonnephropathic, 219750 (3)
CTSA	Galactosialidosis, 256540 (3)
CTSC	Haim-Munk syndrome, 245010 (3)/Papillon-Lefevre syndrome, 245000 (3)/Periodontitis 1, juvenile, 170650 (3)
CTSD	Ceroid lipofuscinosis, neuronal, 10, 610127 (3)
CTSK	Pycnodysostosis, 265800 (3)
CUBN	Megaloblastic anemia-1, Finnish type, 261100 (3)
CUL3	Pseudohypoaldosteronism, type IIE, 614496 (3)
CUL4B	Mental retardation, X-linked, syndromic 15 (Cabezas type), 300354 (3)
CUL7	3-M syndrome 1, 273750 (3)
CXCR4	Myelokathexis, isolated (3)/WHIM syndrome, 193670 (3)
CYB5A	Methemoglobinemia, type IV, 250790 (3)
CYB5R3	Methemoglobinemia, type I, 250800 (3)/Methemoglobinemia, type II, 250800 (3)
CYBA	Chronic granulomatous disease, autosomal, due to deficiency of CYBA, 233690 (3)
CYBB	Chronic granulomatous disease, X-linked, 306400 (3)/Immunodeficiency 34, mycobacteriosis, X-linked, 300645 (3)
CYCS	Thrombocytopenia 4, 612004 (3)
CYLD	Brooke-Spiegler syndrome, 605041 (3)/Cylindromatosis, familial, 132700 (3)/Trichoepithelioma, multiple familial, 1, 601606 (3)
CYP11A1	Adrenal insufficiency, congenital, with 46XY sex reversal, partial or complete, 613743 (3)
CYP11B1	Adrenal hyperplasia, congenital, due to 11-beta-hydroxylase deficiency, 202010 (3)/Aldosteronism, glucocorticoid-remediable, 103900 (3)
CYP11B2	Aldosterone to renin ratio raised (3)/Hypoaldosteronism, congenital, due to CMO I deficiency, 203400 (3)/Hypoaldosteronism, congenital, due to CMO II deficiency, 610600 (3)/Low renin hypertension, susceptibility to (3)
CYP17A1	17,20-lyase deficiency, isolated, 202110 (3)/17-alpha-hydroxylase/17,20-lyase deficiency, 202110 (3)
CYP19A1	Aromatase deficiency, 613546 (3)/Aromatase excess syndrome, 139300 (3)
CYP1B1	Glaucoma 3A, primary open angle, congenital, juvenile, or adult onset, 231300 (3)/Peters anomaly, 604229 (3)
CYP21A1P	Adrenal hyperplasia, congenital, due to 21-hydroxylase deficiency 201910 (3)/Hyperandrogenism, nonclassic type, due to 21-hydroxylase deficiency 201910 (3)
CYP21A2	Adrenal hyperplasia, congenital, due to 21-hydroxylase deficiency, 201910 (3)/Hyperandrogenism, nonclassic type, due to 21-hydroxylase deficiency, 201910 (3)
CYP24A1	Hypercalcemia, infantile, 143880 (3)
CYP26B1	Craniosynostosis with radiohumeral fusions and other skeletal and craniofacial anomalies, 614416 (3)
CYP27A1	Cerebrotendinous xanthomatosis, 213700 (3)
CYP27B1	Vitamin D-dependent rickets, type I, 264700 (3)
CYP2A6	Coumarin resistance, 122700 (3)/Lung cancer, resistance to, 211980 (3)/Nicotine addiction, protection from, 188890 (3)
CYP2B6	Efavirenz, poor metabolism of, 614546 (3)/Efavirenz central nervous system toxicity, susceptibility to, 614546 (3)
CYP2C19	Clopidogrel, impaired responsiveness to, 609535 (3)/Mephenytoin poor metabolizer, 609535(3)/Omeprazole poor metabolizer, 609535(3)/Proguanil poor metabolizer, 609535(3)

CYP2C9	Tolbutamide poor metabolizer (3)/Warfarin sensitivity, 122700 (3)
CYP2D6	Codeine sensitivity, 608902 (3)/Debrisoquine sensitivity, 608902 (3)
CYP2R1	Rickets due to defect in vitamin D 25-hydroxylation, 600081 (3)
CYP4F22	Ichthyosis, congenital, autosomal recessive 5, 604777 (3)
CYP4V2	Bietti crystalline corneoretinal dystrophy, 210370 (3)
CYP7B1	Bile acid synthesis defect, congenital, 3, 613812 (3)/Spastic paraplegia 5A, autosomal recessive, 270800 (3)
D2HGDH	D-2-hydroxyglutaric aciduria, 600721 (3)
DACT1	Neural tube defects
DAG1	Muscular dystrophy-dystroglycanopathy (limb-girdle), type C, 9, 613818 (3)
DARS2	Leukoencephalopathy with brain stem and spinal cord involvement and lactate elevation, 611105 (3)
DAZ1	Oligozoospermia
DBH	Dopamine beta-hydroxylase deficiency, 223360 (3)/[Dopamine-beta-hydroxylase activity levels, plasma] (3)
DBT	Maple syrup urine disease, type II, 248600 (3)
DCAF17	Woodhouse-Sakati syndrome, 241080 (3)
DCC	Colorectal cancer, somatic, 114500 (3)/Esophageal carcinoma, somatic 133239 (3)/Mirror movements 1, 157600 (3)
DCLRE1C	Omenn syndrome, 603554 (3)/Severe combined immunodeficiency, Athabascan type, 602450 (3)
DCN	Corneal dystrophy, congenital stromal, 610048 (3)
DCR	Down syndrome (4)
DCTN1	Neuropathy, distal hereditary motor, type VIIB, 607641 (3)/Perry syndrome, 168605 (3)/Amyotrophic lateral sclerosis, susceptibility to, 105400 (3)
DCX	Lissencephaly, X-linked, 300067 (3)/Subcortical laminal heteropia, X-linked, 300067 (3)
DCXR	[Pentosuria] 260800 (3)
DDB2	Xeroderma pigmentosum, group E, DDB-negative subtype, 278740 (3)
DDC	Aromatic L-amino acid decarboxylase deficiency, 608643 (3)
DDHD1	Spastic paraplegia 28, autosomal recessive, 609340 (3)
DDIT3	Myxoid liposarcoma, 613488 (1)
DDOST	Congenital disorder of glycosylation, type Ir, 614507 (3)
DDR2	Spondylometaepiphyseal dysplasia, short limb-hand type, 271665 (3)
DDX11	Warsaw breakage syndrome, 613398 (3)
DEAF1	Mental retardation, autosomal dominant 24, 615828 (3)
DEC1	Esophageal squamous cell carcinoma 133239 (1)
DES	Muscular dystrophy, limb-girdle, type 2R, 615325 (3)/Cardiomyopathy, dilated, 1I, 604765 (3)/Myopathy, myofibrillar, 1, 601419 (3)/Scapuloperoneal syndrome, neurogenic, Kaeser type, 181400 (3)
DFNA5	Deafness, autosomal dominant 5, 600994 (3)
DFNB31	Deafness, non-syndromic
DFNB59	Progressive hearing loss, autosomal recessive
DGCR	DiGeorge syndrome 188400 (3)
DGKE	Nephrotic syndrome, type 7, 615008 (3)
DGUOK	Mitochondrial DNA depletion syndrome 3 (hepatocerebral type), 251880 (3)
DHCR24	Desmosterolosis, 602398 (3)
DHCR7	Smith-Lemli-Opitz syndrome, 270400 (3)
DHDDS	Retinitis pigmentosa 59, 613861 (3)
DHFR	Megaloblastic anemia due to dihydrofolate reductase deficiency, 613839 (3)
DHH	46XY partial gonadal dysgenesis, with minifascicular neuropathy, 607080 (3)/46XY sex reversal 7, 233420 (3)
DHODH	Miller syndrome, 263750 (3)
DHTKD1	2-aminoadipic 2-oxoadipic aciduria, 204750 (3)/
DIABLO	Progressive hearing loss
DIAPH1	Deafness, autosomal dominant 1, 124900 (3)
DIAPH2	Premature ovarian failure, 300511 (3)
DIAPH3	Auditory neuropathy, autosomal dominant, 1, 609129 (3)
DICER1	Goiter, multinodular 1, with or without Sertoli-Leydig cell tumors, 138800 (3)/Pleuropulmonary blastoma, 601200 (3)/Rhabdomyosarcoma, embryonal, 2, 180295 (3)

DIP2B	Mental retardation, FRA12A type, 136630 (3)
DIRC2	Renal cell carcinoma, 144700 (2)
DIS3L2	Perlman syndrome, 267000 (3)
DISP1	Craniofacial and neuro-developmental abnormalities
DKC1	Dyskeratosis congenita, X-linked, 305000 (3)
DLAT	Pyruvate dehydrogenase E2 deficiency, 245348 (3)
DLD	Dihydrolipoamide dehydrogenase deficiency, 246900 (3)
DLEC1	Esophageal cancer, 133239 (1)/Lung cancer, 211980 (1)
DLG3	Mental retardation, X-linked 90, 300850 (3)
DLL3	Spondylocostal dysostosis 1, autosomal recessive, 277300 (3)
DLX3	Amelogenesis imperfecta, type IV, 104510 (3)/Trichodontoosseous syndrome, 190320 (3)
DLX5	Split-hand/foot malformation 1 with sensorineural hearing loss, 220600 (3)
DMD	Becker muscular dystrophy, 300376 (3)/Cardiomyopathy, dilated, 3B, 302045 (3)/Duchenne muscular dystrophy, 310200 (3)
DMGDH	Dimethylglycine dehydrogenase deficiency, 605850 (3)
DMP1	Hypophosphatemic rickets, AR, 241520 (3)
DMPK	Myotonic dystrophy 1, 160900 (3)
DMRT1	XY gonadal dysgenesis
DNAAF1	Primary ciliary dyskinesia
DNAAF2	Ciliary dyskinesia, primary
DNAAF3	Ciliary dyskinesia, primary, 2, 606763 (3)
DNAH11	Ciliary dyskinesia, primary, 7, with or without situs inversus, 611884 (3)
DNAH5	Ciliary dyskinesia, primary, 3, with or without situs inversus, 608644 (3)
DNAI1	Ciliary dyskinesia, primary, 1, with or without situs inversus, 244400 (3)
DNAI2	Ciliary dyskinesia, primary, 9, with or without situs inversus, 612444 (3)
DNAJB2	Charcot-Marie-Tooth disease, axonal, type 2T, 616233 (3)/Spinal muscular atrophy, distal, autosomal recessive, 5, 614881 (3)
DNAJB6	Muscular dystrophy, limb-girdle, type 1E, 603511 (3)
DNAJC19	3-methylglutaconic aciduria, type V, 610198 (3)
DNAJC5	Ceroid lipofuscinosis, neuronal, 4, Parry type, 162350 (3)
DNAL1	Ciliary dyskinesia, primary, 16, 614017 (3)
DNASE1L3	Systemic lupus erythematosus 16, 614420 (3)
DNM1L	Encephalopahty, lethal, due to defective mitochondrial peroxisomal fission, 614388 (3)
DNM2	Charcot-Marie-Tooth disease, axonal, type 2M, 606482 (3)/Charcot-Marie-Tooth disease, dominant intermediate B, 606482 (3)/Lethal congenital contracture syndrome 5, 615368 (3)/Myopathy, centronuclear, 160150 (3)
DNMT1	Cerebellar ataxia, deafness, and narcolepsy, autosomal dominant, 604121 (3)/Neuropathy, hereditary sensory, type IE, 614116 (3)
DNMT3B	Immunodeficiency-centromeric instability-facial anomalies syndrome 1, 242860 (3)
DOCK6	Adams-Oliver syndrome 2, 614219 (3)
DOCK8	Hyper-IgE recurrent infection syndrome, autosomal recessive, 243700 (3)/Mental retardation, autosomal dominant 2, 614113 (3)
DOK7	Fetal akinesia deformation sequence, 208150 (3)/Myasthenia, limb-girdle, familial, 254300 (3)
DOLK	Dolichol kinase deficiency
DPAGT1	Congenital disorder of glycosylation, type Ij, 608093 (3)/Myasthenic syndrome, congenital, with tubular aggregates 2, 614750 (3)
DPM1	Congenital disorder of glycosylation, type Ie, 608799 (3)
DPM2	Congenital disorder of glycosylation, type Iu, 615042 (3)
DPM3	Congenital disorder of glycosylation, type Io, 612937 (3)
DPP6	Ventricular fibrillation, paroxysmal familial, 2, 612956 (3)
DPYD	5-fluorouracil toxicity, 274270 (3)/Dihydropyrimidine dehydrogenase deficiency, 274270 (3)
DPYS	Dihydropyrimidinuria, 222748 (3)
DRD2	Dystonia, myoclonic, 159900 (3)

DSC2	Arrhythmogenic right ventricular dysplasia 11 with mild palmoplantar keratoderma and woolly hair, 610476 (3)/Arrhythmogenic right ventricular dysplasia 11, 610476 (3)
DSC3	?Hypotrichosis and recurrent skin vesicles, 613102 (3)
DSG1	Erythroderma, congenital, with palmoplantar keratoderma, hypotrichosis, and hyper IgE, 615508 (3)/Keratosis palmoplantaris striata I, AD, 148700 (3)
DSG2	Arrhythmogenic right ventricular dysplasia 10, 610193 (3)/Cardiomyopathy, dilated, 1BB, 612877 (3)
DSG4	Hypotrichosis 6, 607903 (3)
DSP	Arrhythmogenic right ventricular dysplasia 8, 607450 (3)/Cardiomyopathy, dilated, with woolly hair and keratoderma, 605676 (3)/Dilated cardiomyopathy with woolly hair, keratoderma, and tooth agenesis, 615821 (3)/Epidermolysis bullosa, lethal acantholytic, 609638 (3)/Keratosis palmoplantaris striata II, 612908 (3)/Skin fragility-woolly hair syndrome, 607655 (3)
DSPP	Deafness, autosomal dominant 36, with dentinogenesis, 605594 (3)/Dentin dysplasia, type II, 125420 (3)/Dentinogenesis imperfecta, Shields type II, 125490 (3)/Dentinogenesis imperfecta, Shields type III, 125500 (3)
DST	Neuropathy, hereditary sensory and autonomic, type VI, 614653 (3)/Epidermolysis bullosa simplex, sutosomal recessive 2, 615425 (3)
DTNA	Left ventricular noncompaction 1, with or without congenital heart defects, 604169 (3)
DTNBP1	Hermansky-Pudlak syndrome 7, 614076 (3)/Schizophrenia, 181500 (2)
DUOX2	Thryoid dyshormonogenesis 6, 607200 (3)
DUOXA2	Thyroid dyshormonogenesis 5, 274900 (3)
DUX4	Facioscapulohumeral muscular dystrophy
DYM	Dyggve-Melchior-Clausen disease, 223800 (3)/Smith-McCort dysplasia, 607326 (3)
DYNC1H1	Charcot-Marie-Tooth disease, axonal, type 20, 614228 (3)/Mental retardation, autosomal dominant 13, 614563 (3)/Spinal muscular atrophy, lower extremity-predominant 1, AD, 158600 (3)
DYNC2H1	Short-rib thoracic dysplasia 3 with or without polydactyly, 613091 (3)
DYRK1A	Mental retardation, autosomal dominant 7, 614104 (3)
DYSF	Miyoshi muscular dystrophy 1, 254130 (3)/Muscular dystrophy, limb-girdle, type 2B, 253601 (3)/Myopathy, distal, with anterior tibial onset, 606768 (3)
EARS2	Combined oxidative phosphorylation deficiency 12, 614924 (3)
EBP	Chondrodysplasia punctata, X-linked dominant, 302960 (3)
ECE1	Hirschsprung disease, cardiac defects, and autonomic dysfunction, 613870 (3)/Hypertension, essential, susceptibility to, 145500 (3)
ECM1	Urbach-Wiethe disease, 247100 (3)
EDA	Ectodermal dysplasia
EDAR	Ectodermal dysplasia 10A, hypohidrotic/hair/nail type, autosomal dominant, 129490 (3)/Ectodermal dysplasia 10B, hypohidrotic/hair/tooth type, autosomal recessive, 224900 (3)/[Hair morphology 1, hair thickness], 612630 (3)
EDARADD	Ectodermal dysplasia 11A, hypohidrotic/hair/tooth type, autosomal dominant, 614940 (3)/Ectodermal dysplasia 11B, hypohidrotic/hair/tooth type, autosomal recessive, 614941 (3)
EDN3	Central hypoventilation syndrome, congenital, 209880 (3)/Waardenburg syndrome, type 4B, 613265 (3)/Hirschsprung disease, susceptibility to, 4, 613712 (3)
EDNRA	Migraine, resistance to, 157300 (3)
EDNRB	ABCD syndrome, 600501 (3)/Waardenburg syndrome, type 4A, 277580 (3)/Hirschsprung disease, susceptibility to, 2, 600155 (3)
EEF1A2	Epileptic encephalopathy, early infantile, 33 616409/Mental retardation, autosomal dominant 38 616393 (3)
EEF1B2	Intellectual disability
EFEMP1	Doyne honeycomb degeneration of retina, 126600 (3)
EFEMP2	Cutis laxa, autosomal recessive, type IB, 614437 (3)
EFHC1	Epilepsy, juvenile absence, susceptibility to, 1, 607631 (3)/Myoclonic epilepsy, juvenile, susceptibility to, 1, 254770 (3)
EFNB1	Craniofrontonasal dysplasia, 304110 (3)
EFTUD2	Mandibulofacial dysostosis, Guion-Almeida type, 610536 (3)
EGF	Hypomagnesemia 4, renal, 611718 (3)

EGFR	Inflammatory skin and bowel disease, neonatal, 2, 616069 (3)/Adenocarcinoma of lung, response to tyrosine kinase inhibitor in, 211980 (3)/Nonsmall cell lung cancer, response to tyrosine kinase inhibitor in, 211980 (3)/Nonsmall cell lung cancer, susceptibility to, 211980 (3)
EGLN1	Erythrocytosis, familial, 3, 609820 (3)/[Hemoglobin, high altitude adaptation], 609070 (3)
EGR2	Charcot-Marie-Tooth disease, type 1D, 607678 (3)/Dejerine-Sottas disease, 145900 (3)/Neuropathy, congenital hypomyelinating, 1, 605253 (3)
EHMT1	Kleefstra syndrome, 610253 (3)
EIF2AK3	Wolcott-Rallison syndrome, 226980 (3)
EIF2B1	Leukoencephalopathy with vanishing white matter, 603896 (3)
EIF2B2	Leukoencephalopathy with vanishing white matter, 603896 (3)/Ovarioleukodystrophy, 603896 (3)
EIF2B3	Leukoencephalopathy with vanishing white matter, 603896 (3)
EIF2B4	Leukoencephaly with vanishing white matter, 603896 (3)/Ovarioleukodystrophy, 603896 (3)
EIF2B5	Leukoencephalopathy with vanishing white matter, 603896 (3)/Ovarioleukodystrophy, 603896 (3)
EIF2S3	Intellectual disability
EIF4G1	Parkinson disease 18, 614251 (3)
ELANE	Neutropenia, cyclic, 162800 (3)/Neutropenia, severe congenital 1, autosomal dominant, 202700 (3)
ELK1	Mental retardation, X-linked
ELN	Cutis laxa, AD, 123700 (3)/Supravalvar aortic stenosis, 185500 (3)
ELOVL4	Spinocerebellar ataxia 34, 133190 (3)/Ichthyosis, spastic quadriplegia, and mental retardation, 614457 (3)/Stargardt disease 3, 600110 (3)
ELP2	Intellectual disability
EMD	Emery-Dreifuss muscular dystrophy 1, X-linked, 310300 (3)
EMG1	Bowen-Conradi syndrome, 211180 (3)
EMX2	Schizencephaly, 269160 (3)
ENAM	Amelogenesis imperfecta, type IB, 104500 (3)/Amelogenesis imperfecta, type IC, 204650 (3)
ENG	Telangiectasia, hereditary hemorrhagic, type 1, 187300 (3)
ENO3	Glycogen storage disease XIII, 612932 (3)
ENPP1	Arterial calcification, generalized, of infancy, 1, 208000 (3)/Cole disease, 615522 (3)/Hypophosphatemic rickets, autosomal recessive, 2, 613312 (3)/Diabetes mellitus, non-insulin-dependent, susceptibility to, 125853 (3)/Obesity, susceptibility to, 601665 (3)
ENTPD1	Spastic paraplegia 64, 615683 (3)
EP300	Colorectal cancer, somatic, 114500 (3)/Rubinstein-Taybi syndrome 2, 613684 (3)
EPAS1	Erythrocytosis, familial, 4, 611783 (3)
EPB41	Elliptocytosis-1, 611804 (3)
EPB41L1	Mental retardation, autosomal dominant 11, 614257 (3)
EPB42	Spherocytosis, hereditary, type 5, 612690 (3)
EPCAM	Colorectal cancer, hereditary nonpolyposis, type 8, 613244 (3)/Diarrhea 5, with tufting enteropathy, congenital, 613217 (3)
EPHA2	Cataract 6, multiple types, 116600 (3)
EPHB2	Prostate cancer/brain cancer susceptibility, somatic, 603688 (3)
EPHX1	Fetal hydantoin syndrome (1)/Diphenylhydantoin toxicity (1)/Hypercholanemia, familial, 607748 (3)/Preeclampsia, susceptibility to, 189800 (3)
EPHX2	Hypercholesterolemia, familial, due to LDLR defect, modifier of, 143890 (3)
EPM2A	Epilepsy, progressive myoclonic 2A (Lafora), 254780 (3)
EPX	Eosinophil peroxidase deficiency, 261500 (3)
ERBB2	Adenocarcinoma of lung, somatic, 211980 (3)/Gastric cancer, somatic, 613659 (3)/Glioblastoma, somatic, 137800 (3)/Ovarian cancer, somatic, (3)
ERBB3	Lethal congenital contractural syndrome 2, 607598 (3)
ERCC1	Cerebrooculofacioskeletal syndrome 4, 610758 (3)
ERCC2	Cerebrooculofacioskeletal syndrome 2, 610756 (3)/Trichothiodystrophy, 601675 (3)/Xeroderma pigmentosum, group D, 278730 (3)
ERCC3	Trichothiodystrophy, 601675 (3)/Xeroderma pigmentosum, group B, 610651 (3)
ERCC4	Fanconi anemia, complementation group Q, 615272 (3)/XFE progeroid syndrome, 610965 (3)/Xeroderma pigmentosum, group F, 278760 (3)/Xeroderma pigmentosum, type F/Cockayne syndrome, 278760 (3)

ERCC5	Xeroderma pigmentosum, group G, 278780 (3)/Xeroderma pigmentosum, group G/Cockayne syndrome, 278780 (3)
ERCC6	Cerebrooculofacioskeletal syndrome 1, 214150 (3)/Cockayne syndrome, type B, 133540 (3)/De Sanctis-Cacchione syndrome, 278800 (3)/UV-sensitive syndrome 1, 600630 (3)/Lung cancer, susceptibility to, 211980 (3)/Macular degeneration, age-related, susceptibility to 5, 613761 (3)
ERCC8	Cockayne syndrome, type A, 216400 (3)/UV-sensitive syndrome 2, 614621 (3)
ERLIN2	Spastic paraplegia 18, autosomal recessive, 611225 (3)
ERMAP	[Blood group, Radin], 111620 (3)/[Blood group, Scianna system], 111750 (3)
ESCO2	Roberts syndrome, 268300 (3)/SC phocomelia syndrome, 269000 (3)
ESPN	Deafness, autosomal recessive 36, 609006 (3)/Deafness, neurosensory, without vestibular involvement, autosomal dominant (3)
ESRRB	Deafness, autosomal recessive 35, 608565 (3)
ETFA	Glutaric acidemia IIA, 231680 (3)
ETFB	Glutaric acidemia IIB, 231680 (3)
ETFDH	Glutaric acidemia IIC, 231680 (3)
ETHE1	Ethylmalonic encephalopathy, 602473 (3)
ETV6	Leukemia, acute myeloid, somatic, 601626 (3)/Thrombocytopenia 5, 616216 (3)
EVC	Ellis-van Creveld syndrome, 225500 (3)/Weyers acrodental dysostosis, 193530 (3)
EVC2	Weyers acrofacial dysostosis
EWSR1	Ewing sarcoma, 612219 (3)/Neuroepithelioma, 612219 (3)
EXOSC3	Pontocerebellar hypoplasia, type 1B, 614678 (3)
EXT1	Chondrosarcoma, 215300 (3)/Exostoses, multiple, type 1, 133700 (3)
EXT2	Exostoses, multiple, type 2, 133701 (3)
EYA1	Otofaciocervical syndrome, 166780 (3)/Anterior segment anomalies with or without cataract, 113650 (3)/Branchiootic syndrome 1, 602588 (3)/Branchiootorenal syndrome 1, with or without cataracts, 113650 (3)
EYA4	Cardiomyopathy, dilated, 1J, 605362 (3)/Deafness, autosomal dominant 10, 601316 (3)
EYS	Retinitis pigmentosa 25, 602772 (3)
EZH2	Weaver syndrome, 277590 (3)
F10	Factor X deficiency, 227600 (3)
F11	Factor XI deficiency, autosomal dominant, 612416 (3)/Factor XI deficiency, autosomal recessive, 612416 (3)
F11-AS1	Factor XI deficiency, autosomal dominant 612416 (3)/ Factor XI deficiency, autosomal recessive 612416 (3)
F12	Angioedema, hereditary, type III, 610618 (3)/Factor XII deficiency, 234000 (3)
F13A1	Factor XIIIA deficiency, 613225 (3)/Myocardial infarction, protection against, 608446 (3)/Venous thrombosis, protection against, 188050 (3)
F13B	Factor XIIIB deficiency, 613235 (3)
F2	Dysprothrombinemia, 613679 (3)/Hypoprothrombinemia, 613679 (3)/Thrombophilia due to thrombin defect, 188050 (3)/Pregnancy loss, recurrent, susceptibility to, 2, 614390 (3)/Stroke, ischemic, susceptibility to, 601367 (3)
F5	Factor V deficiency, 227400 (3)/Thrombophilia due to activated protein C resistance, 188055 (3)/Budd-Chiari syndrome, 600880 (3)/Pregnancy loss, recurrent, susceptibility to, 1, 614389 (3)/Stroke, ischemic, susceptibility to, 601367 (3)/Thrombophilia, susceptibility to, due to factor V Leiden, 188055 (3)
F7	Factor VII deficiency, 227500 (3)/Myocardial infarction, decreased susceptibility to, 608446 (3)
F8	Hemophilia A, 306700 (3)
F9	Hemophilia B, 306900 (3)/Thrombophilia, X-linked, due to factor IX defect, 300807 (3)/Deep venous thrombosis, protection against, 300807 (3)/Warfarin sensitivity, 122700 (3)
FA2H	Spastic paraplegia 35, autosomal recessive, 612319 (3)
FADD	Infections, recurrent, with encephalopathy, hepatic dysfunction, and cardiovasuclar malformations, 613759 (3)
FAH	Tyrosinemia, type I, 276700 (3)
FAM126A	Leukodystrophy, hypomyelinating, 5, 610532 (3)
FAM134B	Neuropathy, hereditary sensory and autonomic, type IIB, 613115 (3)
FAM161A	Retinitis pigmentosa 28, 606068 (3)
FAM20A	Amelogenesis imperfecta, type IG (enamel-renal syndrome), 204690 (3)

FAM20C	Raine syndrome, 259775 (3)
FAM58A	STAR syndrome, 300707 (3)
FAM83H	Amelogenesis imperfecta, type III, 130900 (3)
FAN1	Interstitial nephritis, karyomegalic, 614817 (3)
FANCA	Fanconi anemia, complementation group A, 227650 (3)
FANCB	VACTERL with Fanconi anaemia
FANCC	Fanconi anemia, complementation group C, 227645 (3)
FANCD2	Fanconi anemia, complementation group D2, 227646 (3)
FANCE	Fanconi anemia, complementation group E, 600901 (3)
FANCF	Fanconi anemia, complementation group F, 603467 (3)
FANCG	Fanconi anaemia
FANCI	Fanconi anemia, complementation group I, 609053 (3)
FANCL	Fanconi anaemia
FANCM	Fanconi anemia, complementation group M, 614087 (3)
FARS2	Combined oxidative phosphorylation deficiency 14, 614946 (3)
FAS	Autoimmune lymphoproliferative syndrome, type IA, 601859 (3)/Squamous cell carcinoma, burn scar-related, somatic (3)/Autoimmune lymphoproliferative syndrome, 601859 (3)
FASLG	Autoimmune lymphoproliferative syndrome, type IB, 601859 (3)/Lung cancer, susceptibility to, 211980 (3)
FASN	Intellectual disability
FASTKD2	Mitochondrial complex IV deficiency, 220110 (3)
FBLN1	Synpolydactyly, 3/3'4, associated with metacarpal and metatarsal synostoses, 608180 (4)
FBLN5	Cutis laxa, autosomal dominant 2, 614434 (3)/Cutis laxa, autosomal recessive, type IA, 219100 (3)/Macular degeneration, age-related, 3, 608895 (3)
FBN1	Acromicric dysplasia, 102370 (3)/Aortic aneurysm, ascending, and dissection (3)/Ectopia lentis, familial, 129600 (3)/Geleophysic dysplasia 2, 614185 (3)/MASS syndrome, 604308 (3)/Marfan syndrome, 154700 (3)/Stiff skin syndrome, 184900 (3)/Weill-Marchesani syndrome 2, dominant, 608328 (3)
FBN2	Contractural arachnodactyly, congenital, 121050 (3)/Macular degeneration, early-onset, 616118 (3)
FBP1	Fructose-1,6-bisphosphatase deficiency, 229700 (3)
FBXO7	Parkinson disease 15, autosomal recessive, 260300 (3)
FBXW4	Split hand/foot malformation 3
FCGR2C	Thrombocytopenic purpura, autoimmune, 188030 (1)
FCN3	Immunodeficiency due to ficolin 3 deficiency, 613860 (3)
FECH	Protoporphyria, erythropoietic, autosomal recessive, 177000 (3)
FERMT1	Kindler syndrome
FERMT3	Integrin activation deficiency disease
FGA	Afibrinogenemia, congenital, 202400 (3)/Amyloidosis, familial visceral, 105200 (3)/Dysfibrinogenemia, congenital, 616004 (3)/Hypodysfibrinogenemia, congenital, 616004 (3)
FGB	Afibrinogenemia, congenital, 202400 (3)/Dysfibrinogenemia,congenital, 616004 (3)/Hypofibrinogenemia, congenital, 202400 (3)
FGD1	Aarskog-Scott syndrome, 305400 (3)/Mental retardation, X-linked syndromic 16, 305400 (3)
FGD4	Charcot-Marie-Tooth disease, type 4H, 609311 (3)
FGF10	Aplasia of lacrimal and salivary glands, 180920 (3)/LADD syndrome, 149730 (3)
FGF14	Spinocerebellar ataxia 27, 609307 (3)
FGF23	Hypophosphatemic rickets, autosomal dominant, 193100 (3)/Osteomalacia, tumor-induced (1)/Tumoral calcinosis, hyperphosphatemic, familial, 211900 (3)
FGF3	Deafness, congenital with inner ear agenesis, microtia, and microdontia, 610706 (3)
FGF8	Hypogonadotropic hypogonadism 6 with or without anosmia, 612702 (3)
FGF9	Multiple synostoses syndrome 3, 612961 (3)
FGFR1	Hartsfield syndrome, 615465 (3)/Hypogonadotropic hypogonadism 2 with or without anosmia, 147950 (3)/Jackson-Weiss syndrome, 123150 (3)/Osteoglophonic dysplasia, 166250 (3)/Pfeiffer syndrome, 101600 (3)/Trigonocephaly 1, 190440 (3)

FGFR2	Antley-Bixler syndrome without genital anomalies or disordered steroidogenesis, 207410 (3)/Apert syndrome, 101200 (3)/Beare-Stevenson cutis gyrata syndrome, 123790 (3)/Bent bone dysplasia syndrome, 614592 (3)/Craniofacial-skeletal-dermatologic dysplasia, 101600 (3)/Craniosynostosis, nonspecific (3)/Crouzon syndrome, 123500 (3)/Gastric cancer, somatic, 613659 (3)/Jackson-Weiss syndrome, 123150 (3)/LADD syndrome, 149730 (3)/Pfeiffer syndrome, 101600 (3)/Saethre-Chotzen syndrome, 101400 (3)/Scaphocephaly and Axenfeld-Rieger anomaly (3)/Scaphocephaly, maxillary retrusion, and mental retardation, 609579 (3)
FGFR3	Achondroplasia, 100800 (3)/Bladder cancer, somatic, 109800 (3)/CATSHL syndrome, 610474 (3)/Cervical cancer, somatic, 603956 (3)/Colorectal cancer, somatic, 114500 (3)/Crouzon syndrome with acanthosis nigricans, 612247 (3)/Hypochondroplasia, 146000 (3)/LADD syndrome, 149730 (3)/Muenke syndrome, 602849 (3)/Nevus, epidermal, somatic, 162900 (3)/Spermatocytic seminoma, somatic, 273300 (3)/Thanatophoric dysplasia, type I, 187600 (3)/Thanatophoric dysplasia, type II, 187601 (3)
FH	Fumarase deficiency, 606812 (3)/Leiomyomatosis and renal cell cancer, 150800 (3)
FHL1	Emery-Dreifuss muscular dystrophy 6, X-linked, 300696 (3)/Hemophagocytic lymphohistiocytosis, familial, 1 (2)/Myopathy, X-linked, with postural muscle atrophy, 300696 (3)/Myopathy, reducing body, X-linked, childhood-onset, 300718 (3)/Myopathy, reducing body, X-linked, severe early-onset, 300717 (3)/Scapuloperoneal myopathy, X-linked dominant, 300695 (3)
FIG4	Polymicrogyria, bilateral temporooccipital, 612691 (3)/Amyotrophic lateral sclerosis 11, 612577 (3)/Charcot-Marie-Tooth disease, type 4J, 611228 (3)/Yunis-Varon syndrome, 216340 (3)
FIGLA	Premature ovarian failure 6, 612310 (3)
FKBP10	Bruck syndrome 1, 259450 (3)/Osteogenesis imperfecta, type XI, 610968 (3)
FKBP14	Ehlers-Danlos syndrome with progressive kyphoscoliosis, myopathy, and hearing loss, 614557 (3)
FKBPL	Infertility, male
FKRP	Muscular dystrophy-dystroglycanopathy (congenital with brain and eye anomalies), type A, 5, 613153 (3)/Muscular dystrophy-dystroglycanopathy (congenital with or without mental retardation), type B, 5, 606612 (3)/Muscular dystrophy-dystroglycanopathy (limb-girdle), type C, 5, 607155 (3)
FKTN	Cardiomyopathy, dilated, 1X, 611615 (3)/Muscular dystrophy-dystroglycanopathy (congenital with brain and eye anomalies), type A, 4, 253800 (3)/Muscular dystrophy-dystroglycanopathy (congenital without mental retardation), type B, 4, 613152 (3)/Muscular dystrophy-dystroglycanopathy (limb-girdle), type C, 4, 611588 (3)
FLCN	Birt-Hogg-Dube syndrome, 135150 (3)/Colorectal cancer, somatic, 114500 (3)/Pneumothorax, primary spontaneous, 173600 (3)/Renal carcinoma, chromophobe, somatic, 144700 (3)
FLG	Ichthyosis vulgaris, 146700 (3)/Dermatitis, atopic, susceptibility to, 2, 605803 (3)
FLNA	Cardiac valvular dysplasia, X-linked, 314400 (3)/Congenital short bowel syndrome, 300048 (3)/FG syndrome 2, 300321 (3)/Frontometaphyseal dysplasia, 305620 (3)/Heterotopia, periventricular, 300049 (3)/Heterotopia, periventricular, ED variant, 300537 (3)/Intestinal pseudoobstruction, neuronal, 300048 (3)/Melnick-Needles syndrome, 309350 (3)/Otopalatodigital syndrome, type I, 311300 (3)/Otopalatodigital syndrome, type II, 304120 (3)/Terminal osseous dysplasia, 300244 (3)
FLNB	Atelosteogenesis, type I, 108720 (3)/Atelosteogenesis, type III, 108721 (3)/Boomerang dysplasia, 112310 (3)/Larsen syndrome, 150250 (3)/Spondylocarpotarsal synostosis syndrome, 272460 (3)
FLNC	Myopathy, distal, 4, 614065 (3)/Myopathy, myofibrillar, 5, 609524 (3)
FLT3	Leukemia, acute lymphoblastic, somatic, 613065 (3)/Leukemia, acute myeloid, 601626 (3)/Leukemia, acute myeloid, reduced survival in, 601626 (3)
FLT4	Hemangioma, capillary infantile, somatic, 602089 (3)/Lymphedema, hereditary, IA, 153100 (3)
FLVCR1	Ataxia, posterior column, with retinitis pigmentosa, 609033 (3)
FLVCR2	Proliferative vasculopathy and hydraencephaly-hydrocephaly syndrome, 225790 (3)
FMO3	Trimethylaminuria, 602079 (3)
FMR1	Fragile X syndrome, 300624 (3)/Fragile X tremor/ataxia syndrome, 300623 (3)/Premature ovarian failure 1, 311360 (3)
FN1	Glomerulopathy with fibronectin deposits 2, 601894 (3)/Plasma fibronectin deficiency, 614101 (1)
FOLR1	Neurodegeneration due to cerebral folate transport deficiency, 613068 (3)
FOXC1	Axenfeld-Rieger syndrome, type 3, 602482 (3)/Iridogoniodysgenesis, type 1, 601631 (3)/Iris hypoplasia and glaucoma, 601631 (3)/Rieger or Axenfeld anomalies, 602482 (3)

FOXC2	Lymphedema-distichiasis syndrome with renal disease and diabetes mellitus, 153400 (3)/Lymphedema-distichiasis syndrome, 153400 (3)
FOXE1	Bamforth-Lazarus syndrome, 241850 (3)
FOXE3	Anterior segment mesenchymal dysgenesis, 107250 (3)/Aphakia, congenital primary, 610256 (3)
FOXF1	Alveolar capillary dysplasia with misalignment of pulmonary veins, 265380 (3)
FOXG1	Rett syndrome, congenital variant, 613454 (3)
FOXH1	Congenital heart defects
FOXI1	Enlarged vestibular aqueduct, 600791 (3)
FOXL2	Blepharophimosis, epicanthus inversus, and ptosis, type 1, 110100 (3)/Blepharophimosis, epicanthus inversus, and ptosis, type 2, 110100 (3)/Premature ovarian failure 3, 608996 (3)
FOXN1	T-cell immunodeficiency, congenital alopecia, and nail dystrophy, 601705 (3)
FOXO1	Rhabdomyosarcoma, alveolar, 268220 (3)
FOXP1	Mental retardation with language impairment and autistic features, 613670 (3)
FOXP2	Speech-language disorder-1, 602081 (3)
FOXP3	Immunodysregulation, polyendocrinopathy, and enteropathy, X-linked, 304790 (3)/Diabetes mellitus, type I, susceptibility to, 222100 (3)
FOXRED1	Leigh syndrome due to mitochondrial complex I deficiency, 256000 (3)/Mitochondrial complex I deficiency, 252010 (3)
FRA10A	Fragile site, FRA10A
FRAS1	Fraser syndrome, 219000 (3)
FREM1	Bifid nose with or without anorectal and renal anomalies, 608980 (3)/Manitoba oculotrichoanal syndrome, 248450 (3)/Trigonocephaly 2, 614485 (3)
FREM2	Fraser syndrome, 219000 (3)
FRMD7	Nystagmus 1, congenital, X-linked, 310700 (3)/Nystagmus, infantile periodic alternating, X-linked, 310700 (3)
FRMPD4	Mental retardation, X-linked
FRY	Intellectual disability
FSCN2	Retinitis pigmentosa 30, 607921 (3)
FSHB	Follicle-stimulating hormone deficiency, isolated, 229070 (3)
FSHR	Ovarian dysgenesis 1, 233300 (3)/Ovarian hyperstimulation syndrome, 608115 (3)/Ovarian response to FSH stimulation, 276400 (3)
FTCD	Glutamate formiminotransferase deficiency, 229100 (3)
FTL	Hyperferritinemia-cataract syndrome, 600886 (3)/L-ferritin deficiency, dominant and recessive, 615604 (3)/Neurodegeneration with brain iron accumulation 3, 606159 (3)
FTO	Growth retardation, developmental delay, coarse facies, and early death, 612938 (3)
FTSJ1	Mental retardation, X-linked 9, 309549 (3)
FUCA1	Fucosidosis, 230000 (3)
FUS	Amyotrophic lateral sclerosis 6, autosomal recessive, with or without frontotemporal dementia, 608030 (3)/Tremor, hereditary essential, 4, 614782 (3)
FUT1	[Bombay phenotype] (3)
FUT6	Fucosyltransferase 6 deficiency, 613852 (3)
FUZ	Neural tube defects, 182940 (3)
FXN	Friedreich ataxia with retained reflexes, 229300 (3)/Friedreich ataxia, 229300 (3)
FXYD2	Hypomagnesemia-2, renal, 154020 (3)
FYCO1	Cataract 18, autosomal recessive, 610019 (3)
FZD4	Exudative vitreoretinopathy 1, 133780 (3)/Retinopathy of prematurity, 133780 (3)
FZD6	Nail disorder, nonsyndromic congenital, 10, (claw-shaped nails), 614157 (3)
G6PC	Glycogen storage disease Ia, 232200 (3)
G6PC3	Dursun syndrome, 612541 (3)/Neutropenia, severe congenital 4, autosomal recessive, 612541 (3)
G6PD	Favism, 134700 (3)/Hemolytic anemia due to G6PD deficiency, 300908 (3)/Resistance to malaria due to G6PD deficiency, 611162 (3)
GAA	Glycogen storage disease II, 232300 (3)
GABRA1	Epileptic encephalopathy, early infantile, 19, 615744 (3)/Epilepsy, childhood absence, susceptibility to, 4, 611136 (3)/Epilepsy, juvenile myoclonic, susceptibility to, 5, 611136 (3)

GABRB3	Epilepsy, childhood absence, susceptibility to, 5, 612269 (3)
GABRD	Epilepsy, generalized, with febrile seizures plus, type 5, susceptibility to, 613060 (3)/Epilepsy, idiopathic generalized, 10, 613060 (3)/Epilepsy, juvenile myoclonic, susceptibility to, 613060 (3)
GABRG2	Epilepsy, generalized, with febrile seizures plus, type 3, 611277 (3)/Febrile seizures, familial, 8, 611277 (3)/Epilepsy, childhood absence, susceptibility to, 2, 607681 (3)
GAD1	Cerebral palsy, spastic quadriplegic, 1, 603513 (3)
GALC	Krabbe disease, 245200 (3)
GALE	Galactose epimerase deficiency, 230350 (3)
GALK1	Galactokinase deficiency with cataracts, 230200 (3)
GALNS	Mucopolysaccharidosis IVA, 253000 (3)
GALNT3	Tumoral calcinosis, hyperphosphatemic, familial, 211900 (3)
GALT	Galactosemia, 230400 (3)
GAMT	Cerebral creatine deficiency syndrome 2, 612736 (3)
GAN	Giant axonal neuropathy-1, 256850 (3)
GARS	Charcot-Marie-Tooth disease, type 2D, 601472 (3)/Neuropathy, distal hereditary motor, type VA, 600794 (3)
GATA1	Anemia, X-linked, with/without neutropenia and/or platelet abnormalities, 300835 (3)/Leukemia, megakaryoblastic, with or without Down syndrome, somatic, 190685 (3)/Thrombocytopenia with beta-thalassemia, X-linked, 314050 (3)/Thrombocytopenia, X-linked, with or without dyserythropoietic anemia, 300367 (3)
GATA2	Emberger syndrome, 614038 (3)/Immunodeficiency 21, 614172 (3)/Leukemia, acute myeloid, susceptibility to, 601626 (3)/Myelodysplastic syndrome, susceptibility to, 614286 (3)
GATA3	Hypoparathyroidism, sensorineural deafness, and renal dysplasia, 146255 (3)
GATA4	Testicular anomalies with or without congenital heart disease, 615542 (3)/Atrial septal defect 2, 607941 (3)/Atrioventricular septal defect 4, 614430 (3)/Tetralogy of Fallot, 187500 (3)/Ventricular septal defect 1, 614429 (3)
GATA6	Atrial septal defect 9, 614475 (3)/Atrioventricular septal defect 5, 614474 (3)/Pancreatic agenesis and congenital heart defects, 600001 (3)/Persistent truncus arteriosus, 217095 (3)/Tetralogy of Fallot, 187500 (3)
GATAD1	Cardiomyopathy, dilated, 2B, 614672 (3)
GATAD2B	Mental retardation, autosomal dominant 18, 615074 (3)
GATM	Cerebral creatine deficiency syndrome 3, 612718 (3)
GBA	Gaucher disease, perinatal lethal, 608013 (3)/Gaucher disease, type I, 230800(3)/Gaucher disease, type II, 230900 (3)/Gaucher disease, type III, 231000 (3)/Gaucher disease, type IIIC, 231005 (3)/Lewy body dementia, susceptibility to, 127750 (3)/Parkinson disease, late-onset, susceptibility to, 168600 (3)
GBE1	Glycogen storage disease IV, 232500 (3)/Polyglucosan body disease, adult form, 263570 (3)
GCDH	Glutaricaciduria, type I, 231670 (3)
GCH1	Dystonia, DOPA-responsive, with or without hyperphenylalaninemia, 128230 (3)/Hyperphenylalaninemia, BH4-deficient, B, 233910 (3)
GCK	Diabetes mellitus, gestational, 125851 (3)/Diabetes mellitus, noninsulin-dependent, late onset, 125853 (3)/Diabetes mellitus, permanent neonatal, 606176 (3)/Hyperinsulinemic hypoglycemia, familial, 3, 602485 (3)/MODY, type II, 125851 (3)
GCLC	Hemolytic anemia due to gamma-glutamylcysteine synthetase deficiency, 230450 (3)/Myocardial infarction, susceptibility to, 608446 (3)
GCM2	Hypoparathyroidism
GCNT2	Adult i phenotype without cataract, 110800 (3)/Cataract 13 with adult i phenotype, 110800 (3)/[Blood group, Ii], 110800 (3)
GCSH	Glycine encephalopathy, 605899 (3)
GDAP1	Charcot-Marie-Tooth disease, axonal, type 2K, 607831 (3)/Charcot-Marie-Tooth disease, axonal, with vocal cord paresis, 607706 (3)/Charcot-Marie-Tooth disease, recessive intermediate, A, 608340 (3)/Charcot-Marie-Tooth disease, type 4A, 214400 (3)
GDF1	Double-outlet right ventricle, 217095 (3)/Right atrial isomerism, 208530 (3)/Tetralogy of Fallot, 187500 (3)/Transposition of great arteries, dextro-looped 3, 613854 (3)
GDF3	Klippel-Feil syndrome 3, autosomal dominant, 613702 (3)/Microphthalmia with coloboma 6, 613703 (3)/Microphthalmia, isolated 7, 613704 (3)

GDF5	Acromesomelic dysplasia, Hunter-Thompson type, 201250 (3)/Brachydactyly, type A1, C, 615072 (3)/Brachydactyly, type A2, 112600 (3)/Brachydactyly, type C, 113100 (3)/Chondrodysplasia, Grebe type, 200700 (3)/Du Pan syndrome, 228900 (3)/Multiple synostoses syndrome 2, 610017 (3)/Symphalangism, proximal, 1B, 615298 (3)/Osteoarthritis-5, 612400 (3)
GDF6	Klippel-Feil syndrome 1, autosomal dominant, 118100 (3)/Leber congenital amaurosis 17, 615360 (3)/Microphthalmia with coloboma 6, digenic, 613703 (3)/Microphthalmia, isolated 4, 613094 (3)
GDI1	Mental retardation, X-linked 41, 300849 (3)
GDNF	Central hypoventilation syndrome, 209880 (3)/Hirschsprung disease, susceptibility to, 3, 613711 (3)/Pheochromocytoma, modifier of, 171300 (3)
GFAP	Alexander disease, 203450 (3)
GFER	Myopathy, mitochondrial progressive, with congenital cataract, hearing loss, and developmental delay, 613076 (3)
GFI1	Neutropenia, nonimmune chronic idiopathic, of adults, 607847 (3)/Neutropenia, severe congenital 2, autosomal dominant, 613107 (3)
GFM1	Combined oxidative phosphorylation deficiency 1, 609060 (3)
GFPT1	Myasthenia, congenital, with tubular aggregates 1, 610542 (3)
GGCX	Pseudoxanthoma elasticum-like disorder with multiple coagulation factor deficiency, 610842 (3)/Vitamin K-dependent clotting factors, combined deficiency of, 1, 277450 (3)
GH1	Growth hormone deficiency, isolated, type IA, 262400 (3)/Growth hormone deficiency, isolated, type IB, 612781 (3)/Growth hormone deficiency, isolated, type II, 173100 (3)/Kowarski syndrome, 262650 (3)
GHR	Increased responsiveness to growth hormone (3) Growth hormone insensitivity, partial, 604271 (3)/Laron dwarfism, 262500 (3)/Hypercholesterolemia, familial, modifier of, 143890 (3)
GHRH	Isolated growth hormone deficiency due to defect in GHRF (1)/Gigantism due to GHRF hypersecretion (1)
GHRHR	Growth hormone deficiency, isolated, type IB, 612781 (3)
GHSR	Growth hormone deficiency, isolated partial, 615925 (3)
GIF	Intrinsic factor deficiency, 261000 (3)
GIGYF2	Parkinson disease 11, 607688 (3)
GIPC3	Deafness, autosomal recessive 15, 601869 (3)
GJA1	Atrioventricular septal defect 3, 600309 (3)/Craniometaphyseal dysplasia, autosomal recessive, 218400 (3)/Hypoplastic left heart syndrome 1, 241550 (3)/Oculodentodigital dysplasia, 164200 (3)/Oculodentodigital dysplasia, autosomal recessive, 257850 (3)/Syndactyly, type III, 186100 (3)
GJA3	Cataract 14, multiple types, 601885 (3)
GJA5	Atrial fibrillation, familial, 11, 614049 (3)/Atrial standstill, digenic (GJA5/SCN5A), 108770 (3)
GJA8	Cataract 1, multiple types, 116200 (3)
GJB1	Charcot-Marie-Tooth neuropathy, X-linked dominant, 1, 302800 (3)
GJB2	Bart-Pumphrey syndrome, 149200 (3)/Deafness, autosomal dominant 3A, 601544 (3)/Deafness, autosomal recessive 1A, 220290 (3)/Hystrix-like ichthyosis with deafness, 602540 (3)/Keratitis-ichthyosis-deafness syndrome, 148210 (3)/Keratoderma, palmoplantar, with deafness, 148350 (3)/Vohwinkel syndrome, 124500 (3)
GJB3	Deafness, autosomal dominant 2B, 612644 (3)/Deafness, autosomal dominant, with peripheral neuropathy (3)/Deafness, autosomal recessive (3)/Deafness, digenic, GJB2/GJB3, 220290 (3)/Erythrokeratodermia variabilis et progressiva, 133200 (3)
GJB4	Erythrokeratodermia variabilis with erythema gyratum repens, 133200 (3)
GJB6	Deafness, autosomal dominant 3B, 612643 (3)/Deafness, autosomal recessive 1B, 612645 (3)/Deafness, digenic GJB2/GJB6, 220290 (3)/Ectodermal dysplasia 2, Clouston type, 129500 (3)
GJC2	Leukodystrophy, hypomyelinating, 2, 608804 (3)/Lymphedema, hereditary, IC, 613480 (3)/Spastic paraplegia 44, autosomal recessive, 613206 (3)
GK	Glycerol kinase deficiency, 307030 (3)
GLA	Fabry disease, 301500 (3)/Fabry disease, cardiac variant, 301500 (3)
GLB1	GM1-gangliosidosis, type I, 230500 (3)/GM1-gangliosidosis, type II, 230600 (3)/GM1-gangliosidosis, type III, 230650 (3)/Mucopolysaccharidosis type IVB (Morquio), 253010 (3)
GLDC	Glycine encephalopathy, 605899 (3)
GLE1	Arthrogryposis, lethal, with anterior horn cell disease, 611890 (3)/Lethal congenital contracture syndrome 1, 253310 (3)

GLI2	Culler-Jones syndrome, 615849 (3)/Holoprosencephaly-9, 610829 (3)
GLI3	Greig cephalopolysyndactyly syndrome, 175700 (3)/Pallister-Hall syndrome, 146510 (3)/Polydactyly, postaxial, types A1 and B, 174200 (3)/Polydactyly, preaxial, type IV, 174700 (3)/Hypothalamic hamartomas, somatic, 241800 (3)
GLIS2	Nephronophthisis 7, 611498 (3)
GLIS3	Diabetes mellitus, neonatal, with congenital hypothyroidism, 610199 (3)
GLMN	Glomuvenous malformations
GLRA1	Hyperekplexia, hereditary 1, autosomal dominant or recessive, 149400 (3)
GLRB	Hyperekplexia 2, autosomal recessive, 614619 (3)
GLRX5	Anemia, sideroblastic, pyridoxine-refractory, autosomal recessive, 205950 (3)
GLUD1	Hyperinsulinism-hyperammonemia syndrome, 606762 (3)
GLUL	Glutamine deficiency, congenital, 610015 (3)
GLYCTK	D-glyceric aciduria, 220120 (3)
GM2A	GM2-gangliosidosis, AB variant, 272750 (3)
GMPS	Leukemia, acute myelogenous, 601626 (3)
GNAI2	Pituitary ACTH-secreting adenoma (3)/Ventricular tachycardia, idiopathic, 192605 (3)
GNAI3	Auriculocondylar syndrome 1, 602483 (3)
GNAS	ACTH-independent macronodular adrenal hyperplasia, 219080 (3)/Acromegaly, somatic, 102200 (3)/McCune-Albright syndrome, somatic, mosaic 174800 (3)/Osseous heteroplasia, progressive, 166350 (3)/Pseudohypoparathyroidism Ia, 103580 (3)/Pseudohypoparathyroidism Ib, 603233 (3)/Pseudohypoparathyroidism Ic, 612462 (3)/Pseudopseudohypoparathyroidism,612463 (3)
GNAT1	Night blindness, congenital stationary, autosomal dominant 3, 610444 (3)
GNAT2	Achromatopsia-4, 613856 (3)
GNE	Inclusion body myopathy, autosomal recessive, 600737 (3)/Nonaka myopathy, 605820 (3)/Sialuria, 269921 (3)
GNMT	Glycine N-methyltransferase deficiency, 606664 (3)
GNPAT	Chondrodysplasia punctata, rhizomelic, type 2, 222765 (3)
GNPTAB	Mucolipidosis II alpha/beta, 252500 (3)/Mucolipidosis III alpha/beta, 252600 (3)
GNPTG	Stuttering
GNRH1	Hypogonadotropic hypogonadism 12 with or without anosmia, 614841 (3)
GNRHR	Fertile eunuch syndrome, 228300 (3)/Hypogonadotropic hypogonadism 7 without anosmia, 146110 (3)
GNS	Mucopolysaccharidosis type IIID, 252940 (3)
GOLGA5	Thyroid carcinoma, papillary, 188550 (3)
GON4L	Intellectual disability
GOPC	Globozoospermia, 102530 (1)
GORAB	Geroderma osteodysplasticum, 231070 (3)
GOSR2	Epilepsy, progressive myoclonic 6, 614018 (3)
GOT1	Aspartate aminotransferase, serum level of, QTL1, 614419 (3)
GP1BA	Bernard-Soulier syndrome, type A1 (recessive), 231200 (3)/Bernard-Soulier syndrome, type A2 (dominant), 153670 (3)/von Willebrand disease, platelet-type, 177820 (3)/Nonarteritic anterior ischemic optic neuropathy, susceptibility to, 258660 (3)
GP1BB	Bernard-Soulier syndrome, type B, 231200 (3)/Giant platelet disorder, isolated, 231200 (3)
GP6	Bleeding disorder, platelet-type, 11, 614201 (3)
GP9	Bernard-Soulier syndrome, type C, 231200 (3)
GPC3	Simpson-Golabi-Behmel syndrome, type 1, 312870 (3)/Wilms tumor, somatic, 194070 (3)
GPC6	Omodysplasia 1, 258315 (3)
GPD1	Hypertriglyceridemia, transient infantile, 614480 (3)
GPD1L	Brugada syndrome 2 611777 (3)
GPHN	Molybdenum cofactor deficiency C, 615501 (3)
GPI	Hemolytic anemia, nonspherocytic, due to glucose phosphate isomerase deficiency, 613470 (3)
GPR143	Nystagmus 6, congenital, X-linked, 300814 (3)/Ocular albinism, type I, Nettleship-Falls type, 300500 (3)
GPR179	Night blindness, congenital stationary (complete), 1E, autosomal recessive, 614565 (3)
GPR56	Polymicrogyria, bilateral frontoparietal, 606854 (3)/Polymicrogyria, bilateral perisylvian, 615752 (3)

GPR98	Febrile seizures, familial, 4, 604352 (3)/Usher syndrome, type 2C, 605472 (3)/Usher syndrome, type 2C, GPR98/PDZD7 digenic, 605472 (3)
GPSM2	Chudley-McCullough syndrome, 604213 (3)
GPX1	Hemolytic anemia due to glutathione peroxidase deficiency, 614164 (1)
GRHL2	Deafness, autosomal dominant 28, 608641 (3)/Ectodermal dysplasia/short stature syndrome, 616029 (3)
GRHPR	Hyperoxaluria, primary, type II, 260000 (3)
GRIA3	Mental retardation, X-linked 94, 300699 (3)
GRIK2	Mental retardation, autosomal recessive, 6, 611092 (3)
GRIN1	Mental retardation, autosomal dominant 8, 614254 (3)
GRIN2A	Epilepsy, focal, with speech disorder and with or without mental retardation, 245570 (3)
GRIN2B	Epileptic encephalopathy, early infantile, 27, 616139 (3)/Mental retardation, autosomal dominant 6, 613970 (3)
GRIP1	Fraser syndrome, 219000 (3)
GRK1	Oguchi disease-2, 613411 (3)
GRM1	Spinocerebellar ataxia, autosomal recessive 13, 614831 (3)
GRM6	Night blindness, congenital stationary (complete), 1B, autosomal recessive, 257270 (3)
GRN	Aphasia, primary progressive, 607485 (3)/Ceroid lipofuscinosis, neuronal, 11, 614706 (3)/Frontotemporal lobar degeneration with ubiquitin-positive inclusions, 607485 (3)
GRPR	Autism and multiple exostoses
GRXCR1	Deafness, autosomal recessive 25, 613285 (3)
GSN	Amyloidosis, Finnish type, 105120 (3)
GSR	Hemolytic anemia due to glutathione reductase deficiency (1)
GSS	Glutathione synthetase deficiency, 266130 (3)/Hemolytic anemia due to glutathione synthetase deficiency, 231900 (3)
GTDC2	Walker-Warburg syndrome
GTF2H5	Trichothiodystrophy, complementation group A, 601675 (3)
GUCA1A	Cone dystrophy-3, 602093 (3)/Cone-rod dystrophy 14, 602093 (3)
GUCA1B	Retinitis pigmentosa 48, 613827 (3)
GUCY2C	Diarrhea 6, 614616 (3)/Meconium ileus, 614665 (3)
GUCY2D	Cone-rod dystrophy 6, 601777(3)/Leber congenital amaurosis 1, 204000 (3)
GUSB	Mucopolysaccharidosis VII, 253220 (3)
GYG1	Glycogen storage disease XV, 613507 (3)/Polyglucosan body myopathy 2, 616199 (3)
GYS1	Glycogen storage disease 0, muscle, 611556 (3)
GYS2	Glycogen storage disease 0, liver, 240600 (3)
H6PD	Cortisone reductase deficiency 1, 604931 (3)
HADH	Hypoglycaemia, hyperinsulinaemic
HADHA	Fatty liver, acute, of pregnancy, 609016 (3)/HELLP syndrome, maternal, of pregnancy, 609016 (3)/LCHAD deficiency, 609016 (3)/Trifunctional protein deficiency, 609015 (3)
HADHB	Trifunctional protein deficiency, 609015 (3)
HAGH	[Glyoxalase II deficiency], 614033 (1)
HAMP	Hemochromatosis, type 2B, 613313 (3)
HARS	Usher syndrome type 3B, 614504 (3)
HARS2	Perrault syndrome 2, 614926 (3)
HAX1	Neutropenia, severe congenital 3, autosomal recessive, 610738 (3)
HBA1	Erythremias, alpha- (3)/Heinz body anemias, alpha-, 140700 (3)/Hemoglobin H disease, nondeletional, 613978 (3)/Methemoglobinemias, alpha- (3)/Thalassemias, alpha-, 604131 (3)
HBA2	Erythrocytosis (3)/Heinz body anemia, 140700 (3)/Hemoglobin H disease, nondeletional, 613978 (3)/Hypochromic microcytic anemia (3)/Thalassemia, alpha-, 604131 (3)
HBB	Delta-beta thalassemia, 141749 (3)/Erythremias, beta- (3)/Heinz body anemias, beta-, 140700 (3)/Hereditary persistence of fetal hemoglobin, 141749 (3)/Methemoglobinemias, beta- (3)/Sickle cell anemia, 603903 (3)/Thalassemia-beta, dominant inclusion-body, 603902 (3)/Thalassemias, beta-, 613985 (3)/Malaria, resistance to, 611162 (3)
HBD	Thalassemia due to Hb Lepore (3)/Thalassemia, delta- (3)
HBE1	Thalassaemia gamma-delta-beta

HBEGF	Diphtheria, susceptibility to (1)
HBG1	Fetal hemoglobin quantitative trait locus 1, 141749 (3)
HBG2	Cyanosis, transient neonatal, 613977 (3)/Fetal hemoglobin quantitative trait locus 1, 141749 (3)
HCCS	Microphthalmia, syndromic 7, 309801 (3)
HCFC1	Mental retardation, X-linked 3 (methylmalonic acidemia and homocysteinemia, cblX type), 309541 (3)
HCN4	Brugada syndrome 8, 613123 (3)/Sick sinus syndrome 2, 163800 (3)
HCRT	Narcolepsy 1, 161400 (3)
HDAC4	Brachydactyly-mental retardation syndrome, 600430 (3)
HDAC8	Cornelia de Lange syndrome 5, 300882 (3)/Wilson-Turner syndrome, 309585 (3)
HEATR2	Ciliary dyskinesia, primary, 18, 614874 (3)
HEPACAM	Megalencephalic leukoencephalopathy with subcortical cysts 2A, 613925 (3)/Megalencephalic leukoencephalopathy with subcortical cysts 2B, remitting, with or without mental retardation, 613926 (3)
HES7	Spondylocostal dysostosis 4, autosomal recessive, 613686 (3)
HESX1	Growth hormone deficiency with pituitary anomalies, 182230 (3)/Pituitary hormone deficiency, combined, 5, 182230 (3)/Septooptic dysplasia, 182230 (3)
HEXA	GM2-gangliosidosis, several forms, 272800 (3)/Tay-Sachs disease, 272800 (3)/[Hex A pseudodeficiency], 272800 (3)
HEXB	Sandhoff disease, infantile, juvenile, and adult forms, 268800 (3)
HFE	Hemochromatosis, 235200 (3)/[Transferrin serum level QTL2], 614193 (3)/Alzheimer disease, susceptibility to, 104300 (3)/Microvascular complications of diabetes 7, 612635 (3)/Porphyria cutanea tarda, susceptibility to, 176100 (3)/Porphyria variegata, susceptibility to, 176200 (3)
HFE2	Haemochromatosis
HGD	Alkaptonuria, 203500 (3)
HGF	Deafness, autosomal recessive 39, 608265 (3)
HGSNAT	Mucopolysaccharidosis type IIIC (Sanfilippo C), 252930 (3)
HIBCH	3-hydroxyisobutryl-CoA hydrolase deficiency, 250620 (3)
HINT1	Neuromyotonia and axonal neuropathy, autosomal recessive, 137200 (3)
HIP1	Prostate cancer, progression of, 176807 (1)
HIST3H3	Intellectual disability
HIVEP2	Mental retardation, autosomal dominant 43 616977 (3)
HK1	Hemolytic anemia due to hexokinase deficiency, 235700 (3)/Neuropathy, hereditary motor and sensory, Russe type, 605285 (3)
HLA-A	Hypersensitivity syndrome, carbamazepine-induced, susceptibility to, 608579 (3)
HLA-DQB1	Celiac disease, susceptibility to, 212750 (3)/Creutzfeldt-Jakob disease, variant, resistance to, 123400 (3)/Multiple sclerosis, susceptibility to, 1, 126200 (3)
HLA-DRB1	Multiple sclerosis, susceptibility to, 1, 126200 (3)/Pemphigoid, susceptibility to (2)/Rheumatoid arthritis, susceptibility to, 180300 (3)/Sarcoidosis, susceptibility to, 1, 181000 (3)
HLCS	Holocarboxylase synthetase deficiency, 253270 (3)
HMBS	Porphyria, acute intermittent, 176000 (3)/Porphyria, acute intermittent, nonerythroid variant, 176000 (3)
HMGA1	Diabetes mellitus, noninsulin-dependent, susceptibility to, 125853 (3)
HMGA2	Leiomyoma, uterine, somatic, 150699 (2)
HMGCL	HMG-CoA lyase deficiency, 246450 (3)
HMGCS2	HMG-CoA synthase-2 deficiency, 605911 (3)
HMOX1	Heme oxygenase-1 deficiency, 614034 (3)/Pulmonary disease, chronic obstructive, susceptibility to, 606963 (3)
HMX1	Oculoauricular syndrome, 612109 (3)
HNF1A	Diabetes mellitus, insulin-dependent, 20, 612520 (3)/Hepatic adenoma, somatic, 142330 (3)/MODY, type III, 600496 (3)/Renal cell carcinoma, 144700 (3)/Diabetes mellitus, insulin-dependent, 222100 (3)/Diabetes mellitus, noninsulin-dependent, 2, 125853 (3)
HNF1B	Diabetes mellitus, noninsulin-dependent, 125853 (3)/Renal cysts and diabetes syndrome, 137920 (3)/Renal cell carcinoma, 144700 (3)
HNF4A	Fanconi renotubular syndrome 4, with maturity-onset diabetes of the young, 616026 (3)/MODY, type I, 125850 (3)/Diabetes mellitus, noninsulin-dependent, 125853 (3)
HOGA1	Hyperoxaluria, primary, type III, 613616 (3)

HOXA1	Athabaskan brainstem dysgenesis syndrome, 601536 (3)/Bosley-Salih-Alorainy syndrome, 601536 (3)
HOXA11	Radioulnar synostosis with amegakaryocytic thrombocytopenia, 605432 (3)
HOXA13	Guttmacher syndrome, 176305 (3)/Hand-foot-uterus syndrome, 140000 (3)
HOXA2	Microtia with or without hearing impairment (AD), 612290 (3)/
HOXB1	Facial paresis, hereditary congenital, 3, 614744 (3)
HOXC13	Ectodermal dysplasia 9, hair/nail type, 614931 (3)
HOXC13-AS	Ectodermal dysplasia 9, hair/nail type 614931 (3)
HOXD10	Charcot-Marie-Tooth disease, foot deformity of, 192950 (3)/Vertical talus, congenital, 192950 (3)
HOXD13	VACTERL association, 192350 (3)/Brachydactyly, type D, 113200 (3)/Brachydactyly, type E, 113300 (3)/Brachydactyly-syndactyly syndrome, 610713 (3)/Syndactyly, type V, 186300 (3)/Synpolydactyly with foot anomalies, 186000 (3)/Synpolydactyly, type II, 186000 (3)
HPD	Hawkinsinuria, 140350 (3)/Tyrosinemia, type III, 276710 (3)
HPGD	Cranioosteoarthropathy, 259100 (3)/Digital clubbing, isolated congenital, 119900 (3)/Hypertrophic osteoarthropathy, primary, autosomal recessive 1, 259100 (3)
HPRT1	HPRT-related gout, 300323 (3)/Lesch-Nyhan syndrome, 300322 (3)
HPS1	Hermansky-Pudlak syndrome 1, 203300 (3)
HPS3	Hermansky-Pudlak syndrome 3, 614072 (3)
HPS4	Hermansky-Pudlak syndrome 4, 614073 (3)
HPS5	Hermansky-Pudlak syndrome 5, 614074 (3)
HPS6	Hermansky-Pudlak syndrome 6, 614075 (3)
HPSE2	Urofacial syndrome 1, 236730 (3)
HR	Alopecia universalis, 203655 (3)/Atrichia with papular lesions, 209500 (3)/Hypotrichosis 4, 146550 (3)
HRAS	Congenital myopathy with excess of muscle spindles, 218040 (3)/Costello syndrome, 218040 (3)/Schimmelpenning-Feuerstein-Mims syndrome, somatic mosaic, 163200 (3)/Bladder cancer, somatic, 109800 (3)/Nevus sebaceous, somatic, 162900 (3)/Thyroid carcinoma, follicular, somatic, 188470 (3)
HRG	Thrombophilia due to HRG deficiency, 613116 (3)/Thrombophilia due to elevated HRG, 613116 (1)
HS6ST1	Hypogonadotropic hypogonadism 15 with or without anosmia, 614880 (3)
HSD11B1	Cortisone reductase deficiency 2, 614662 (3)
HSD11B2	Apparent mineralocorticoid excess, 218030 (3)
HSD17B10	17-beta-hydroxysteroid dehydrogenase X deficiency, 300438 (3)/
HSD17B3	Pseudohermaphroditism, male, with gynecomastia, 264300 (3)
HSD17B4	D-bifunctional protein deficiency, 261515 (3)/Perrault syndrome 1, 233400 (3)
HSD3B2	3-beta-hydroxysteroid dehydrogenase, type II, deficiency, 201810 (3)
HSD3B7	Bile acid synthesis defect, congenital, 1, 607765 (3)
HSF4	Cataract 5, multiple types, 116800 (3)
HSPB1	Charcot-Marie-Tooth disease, axonal, type 2F, 606595 (3)/Neuropathy, distal hereditary motor, type IIB, 608634 (3)
HSPB3	Neuronopathy, distal hereditary motor, type IIC, 613376 (3)
HSPB8	Charcot-Marie-Tooth disease, axonal, type 2L, 608673 (3)/Neuropathy, distal hereditary motor, type IIA, 158590 (3)
HSPD1	Leukodystrophy, hypomyelinating, 4, 612233 (3)/Spastic paraplegia 13, autosomal dominant, 605280 (3)
HSPG2	Dyssegmental dysplasia, Silverman-Handmaker type, 224410 (3)/Schwartz-Jampel syndrome, type 1, 255800 (3)
HTR1A	Periodic fever, menstrual cycle dependent, 614674 (3)
HTRA1	CARASIL syndrome, 600142 (3)/Macular degeneration, age-related, 7, 610149 (3)/Macular degeneration, age-related, neovascular type, 610149 (3)
HTRA2	Parkinson disease 13, 610297 (3)
HTT	Huntington disease, 143100 (3)
HUWE1	Mental retardation, X-linked syndromic, Turner type, 300706 (3)
HYAL1	Mucopolysaccharidosis type IX, 601492 (3)
HYDIN	Ciliary dyskinesia, primary, 5, 608647 (3)
HYLS1	Hydrolethalus syndrome, 236680 (3)
ICAM1	Malaria, cerebral, susceptibility to, 611162 (3)
ICK	Endocrine-cerebroosteodysplasia, 612651 (3)

ICOS	Immunodeficiency, common variable, 1, 607594 (3)
IDH2	D-2-hydroxyglutaric aciduria 2, 613657 (3)
IDH3B	Retinitis pigmentosa 46, 612572 (3)
IDS	Mucopolysaccharidosis II, 309900 (3)
IDUA	Mucopolysaccharidosis Ih, 607014 (3)/Mucopolysaccharidosis Ih/s, 607015 (3)/Mucopolysaccharidosis Is, 607016 (3)
IER3IP1	Microcephaly, epilepsy, and diabetes syndrome, 614231 (3)
IFITM5	Osteogenesis imperfecta, type V, 610967 (3)
IFNAR2	Hepatitis B virus, susceptibility to, 610424 (3)
IFNG	AIDS, rapid progression to, 609423 (3)/Aplastic anemia, 609135 (3)/Hepatitis C virus, response to therapy of, 609532 (3)/TSC2 angiomyolipomas, renal, modifier of, 613254 (3)/Tuberculosis, protection against, 607948 (3)
IFNGR1	Immunodeficiency 27A, mycobacteriosis, AR, 209950 (3)/Immunodeficiency 27B, mycobacteriosis, AD, 615978 (3)/H. pylori infection, susceptibility to, 600263 (3)/Hepatitis B virus infection, susceptibility to, 610424 (3)/Tuberculosis infection, protection against, 607948 (3)/Tuberculosis, susceptibility to, 607948 (3)
IFNGR2	Immunodeficiency 28, mycobacteriosis, 614889 (3)
IFT122	Cranioectodermal dysplasia 1, 218330 (3)
IFT140	Short-rib thoracic dysplasia 9 with or without polydactyly, 266920 (3)
IFT43	Cranioectodermal dysplasia 3, 614099 (3)
IFT80	Short-rib thoracic dysplasia 2 with or without polydactyly, 611263 (3)
IGBP1	Corpus callosum, agenesis of, with mental retardation, ocular coloboma and micrognathia, 300472 (3)
IGF1	Growth retardation with deafness and mental retardation due to IGF1 deficiency, 608747 (3)
IGF1R	Insulin-like growth factor I, resistance to, 270450 (3)
IGF2	?Growth restriction, severe, with distinctive facies 616489 (3)
IGFBP7	Retinal arterial macroaneurysm with supravalvular pulmonic stenosis, 614224 (3)
IGFBP7-AS1	Retinal arterial macroaneurysm with supravalvular pulmonic stenosis 614224 (3)
IGHM	Agammaglobulinemia 1, 601495 (3)
IGHMBP2	Charcot-Marie-Tooth disease, axonal, type 2S, 616155 (3)/Neuronopathy, distal hereditary motor, type VI, 604320 (3)
IGKC	[Kappa light chain deficiency], 614102 (3)
IGLL1	Agammaglobulinemia 2, 613500 (3)
IGSF1	Hypothyroidism, central, and testicular enlargement, 300888 (3)
IHH	Acrocapitofemoral dysplasia, 607778 (3)/Brachydactyly, type A1, 112500 (3)
IKBKAP	Dysautonomia, familial, 223900 (3)
IKBKG	Ectodermal dysplasia, hypohidrotic, with immune deficiency, 300291 (3)/Ectodermal, dysplasia, anhidrotic, lymphedema and immunodeficiency, 300301 (3)/Immunodeficiency 33, 300636 (3)/Immunodeficiency, isolated, 300584 (3)/Incontinentia pigmenti, type II, 308300 (3)/Invasive pneumococcal disease, recurrent isolated, 2, 300640 (3)
IL10RA	Inflammatory bowel disease 28, early onset, autosomal recessive, 613148 (3)
IL10RB	Enterocolitis, early-onset
IL11RA	Craniosynostosis and dental anomalies, 614188 (3)
IL12B	Immunodeficiency 29, mycobacteriosis, 614890 (3)/Asthma, susceptibility to, 600807 (3)
IL12RB1	Immunodeficiency 30, 614891 (3)
IL17F	Candidiasis, familial, 6, autosomal dominant, 613956 (3)
IL17RA	Candidiasis, familial, 5, autosomal recessive, 613953 (3)
IL1RAPL1	Mental retardation, X-linked 21/34, 300143 (3)
IL1RN	Interleukin 1 receptor antagonist deficiency, 612852 (3)/Gastric cancer risk after H. pylori infection, 137215 (3)/Microvascular complications of diabetes 4, 612628 (3)
IL21R	Immunodeficiency, primary, autosomal recessive, IL21R-related, 615207 (3)/[IgE, elevated level of], 147050 (3)
IL2RA	Interleukin-2 receptor, alpha chain, deficiency of, 606367 (3)/Diabetes, mellitus, insulin-dependent, susceptibility to, 10, 601942 (3)
IL2RG	Combined immunodeficiency, X-linked, moderate, 312863 (3)/Severe combined immunodeficiency, X-linked, 300400 (3)

IL31RA	Amyloidosis, primary localized cutaneous, 2, 613955 (3)
IL36RN	Psoriasis 14, pustular, 614204 (3)
IL4R	AIDS, slow progression to, 609423 (3)/Atopy, susceptibility to, 147050 (3)
IL6	{Crohn disease-associated growth failure} 266600 (3)/{Diabetes, susceptibility to},222100 (3)/{Intracranial hemorrhage in brain cerebrovascular malformations, susceptibility to},108010 (3)/{Kaposi sarcoma, susceptibility to},148000 (3)/{Rheumatoid arthritis, systemic juvenile}, 604302 (3)
IL7R	Severe combined immunodeficiency, T-cell negative, B-cell/natural killer cell-positive type, 608971 (3)
ILDR1	Deafness, autosomal recessive 42, 609646 (3)
ILK	Cardiomyopathy, dilated
IMPAD1	Chondrodysplasia with joint dislocations, GRAPP type, 614078 (3)
IMPDH1	Leber congenital amaurosis 11, 613837 (3)/Retinitis pigmentosa 10, 180105 (3)
IMPG2	Macular dystrophy, vitelliform, 5, 616152 (3)/Retinitis pigmentosa 56, 613581 (3)
INF2	Charcot-Marie-Tooth disease, dominant intermediate E, 614455 (3)/Glomerulosclerosis, focal segmental, 5, 613237 (3)
ING1	Squamous cell carcinoma, head and neck, somatic, 275355 (3)
INPP5E	Joubert syndrome 1, 213300 (3)/Mental retardation, truncal obesity, retinal dystrophy, and micropenis, 610156 (3)
INS	Diabetes mellitus, insulin-dependent, 2, 125852 (3)/Diabetes mellitus, permanent neonatal, 606176 (3)/Hyperproinsulinemia, 616214 (3)/Maturity-onset diabetes of the young, type 10, 613370 (3)
INSL3	Cryptorchidism, 219050 (3)
INSR	Diabetes mellitus, insulin-resistant, with acanthosis nigricans, 610549 (3)/Hyperinsulinemic hypoglycemia, familial, 5, 609968 (3)/Leprechaunism, 246200 (3)/Rabson-Mendenhall syndrome, 262190 (3)
INVS	Nephronophthisis 2, infantile, 602088 (3)
IQCB1	Senior-Loken syndrome 5, 609254 (3)
IQSEC2	Mental retardation, X-linked 1, 309530 (3)
IRAK4	IRAK4 deficiency, 607676 (3)/Invasive pneumococcal disease, recurrent isolated, 1, 610799 (3)
IRF1	Gastric cancer, somatic, 613659 (3)/Myelodysplastic syndrome, preleukemic (3)/Myelogenous leukemia, acute (3)/Nonsmall cell lung cancer, somatic, 211980 (3)
IRF4	[Skin/hair/eye pigmentation, variation in, 8], 611724 (3)
IRF6	Orofacial cleft 6, 608864 (3)/Popliteal pterygium syndrome 1, 119500 (3)/van der Woude syndrome, 119300 (3)
IRF8	Immunodeficiency 32A, mycobacteriosis, autosomal dominant, 614893 (3)/Immunodeficiency 32B, monocyte and dendritic cell deficiency, autosomal recessive, 614894 (3)
IRGM	Inflammatory bowel disease 19, 612278 (3)/Mycobacterium tuberculosis, protection against, 607948 (3)
IRX5	Hamamy syndrome, 611174 (3)
ISCU	Myopathy with lactic acidosis, hereditary, 255125 (3)
ISPD	Muscular dystrophy-dystroglycanopathy (congenital with brain and eye anomalies), type A, 7, 614643 (3)/Muscular dystrophy-dystroglycanopathy (limb-girdle), type C, 7, 616052 (3)
ITCH	Autoimmune disease, multisystem, with facial dysmorphism, 613385 (3)
ITGA2	Glycoprotein Ia deficiency, 614200 (1)
ITGA2B	Bleeding disorder, platelet-type, 16, autosomal dominant, 187800 (3)/Glanzmann thrombasthenia, 273800 (3)/Thrombocytopenia, neonatal alloimmune, BAK antigen related (3)
ITGA3	Interstitial lung disease, nephrotic syndrome, and epidermolysis bullosa, congenital, 614748 (3)
ITGA6	Epidermolysis bullosa, junctional, with pyloric stenosis, 226730 (3)
ITGA7	Muscular dystrophy, congenital, due to ITGA7 deficiency, 613204 (3)
ITGAM	Systemic lupus erythematous, association with susceptibility to, 6, 609939 (3)
ITGB2	Leukocyte adhesion deficiency, 116920 (3)
ITGB3	Bleeding disorder, platelet-type, 16, autosomal dominant, 187800 (3)/Glanzmann thrombasthenia, 273800 (3)/Purpura, posttransfusion (3)/Thrombocytopenia, neonatal alloimmune (3)/Myocardial infarction, susceptibility to, 608446 (3)
ITGB4	Epidermolysis bullosa of hands and feet, 131800 (3)/Epidermolysis bullosa, junctional, non-Herlitz type, 226650 (3)/Epidermolysis bullosa, junctional, with pyloric atresia, 226730 (3)
ITK	Lymphoproliferative syndrome 1, 613011 (3)
ITM2B	Dementia, familial British 176500 (3)/Dementia, familial Danish 117300 (3)

ITPR1	Spinocerebellar ataxia 15, 606658 (3)/Spinocerebellar ataxia 29, congenital nonprogressive, 117360 (3)
IVD	Isovaleric acidemia, 243500 (3)
IYD	Thyroid dyshormonogenesis 4, 274800 (3)
JAG1	Alagille syndrome, 118450 (3)/Deafness, congenital heart defects, and posterior embryotoxon (3)/Tetralogy of Fallot, 187500 (3)
JAK2	Erythrocytosis, somatic, 133100 (3)/Leukemia, acute myelogenous, 601626 (3)/Myelofibrosis, somatic, 254450 (3)/Polycythemia vera, 263300 (3)/Thrombocythemia 3, 614521 (3)/Budd-Chiari syndrome, 600880 (3)
JAK3	SCID, autosomal recessive, T-negative/B-positive type, 600802 (3)
JAM3	Hemorrhagic destruction of the brain, subependymal calcification, and cataracts, 613730 (3)
JPH2	Cardiomyopathy, familial hypertrophic 17, 613873 (3)
JPH3	Huntington disease-like 2, 606438 (3)
JUP	Arrhythmogenic right ventricular dysplasia 12, 611528 (3)/Naxos disease, 601214 (3)
KAL1	Hypogonadotropic hypogonadism 1 with or without anosmia (Kallmann syndrome 1), 308700 (3)
KANK1	Cerebral palsy, spastic quadriplegic, 2, 612900 (3)
KANSL1	Koolen-De Vries syndrome, 610443 (3)
KARS	Charcot-Marie-Tooth disease, recessive intermediate, B, 613641 (3)/Deafness, autosomal recessive 89, 613916 (3)
KAT6B	Genitopatellar syndrome, 606170 (3)/SBBYSS syndrome, 603736 (3)
KBTBD13	Nemaline myopathy 6, autosomal dominant, 609273 (3)
KCNA1	Episodic ataxia/myokymia syndrome, 160120 (3)
KCNA5	Atrial fibrillation, familial, 7, 612240 (3)
KCNC3	Spinocerebellar ataxia 13, 605259 (3)
KCNE1	Jervell and Lange-Nielsen syndrome 2, 612347 (3)/Long QT syndrome 5, 613695 (3)
KCNE1L	Idiopathic ventricular fibrillation
KCNE2	Atrial fibrillation, familial, 4, 611493 (3)/Long QT syndrome 6, 613693 (3)
KCNE3	Brugada syndrome 6, 613119 (3)
KCNH2	Long QT syndrome 2, 613688 (3)/Short QT syndrome 1, 609620 (3)/Long QT syndrome 2, acquired, susceptibility to, 613688 (3)
KCNJ1	Bartter syndrome, type 2, 241200 (3)
KCNJ10	Enlarged vestibular aqueduct, digenic, 600791 (3)/SESAME syndrome, 612780 (3)
KCNJ11	Diabetes mellitus, permanent neonatal, with neurologic features, 606176 (3)/Diabetes mellitus, transient neonatal, 3, 610582 (3)/Diabetes, permanent neonatal, 606176 (3)/Hyperinsulinemic hypoglycemia, familial, 2, 601820 (3)/Diabetes mellitus, type 2, susceptibility to, 125853 (3)
KCNJ13	Leber congenital amaurosis 16, 614186 (3)/Snowflake vitreoretinal degeneration, 193230 (3)
KCNJ2	Andersen syndrome, 170390 (3)/Atrial fibrillation, familial, 9, 613980 (3)/Short QT syndrome 3, 609622 (3)
KCNJ5	Hyperaldosteronism, familial, type III, 613677 (3)/Long QT syndrome 13, 613485 (3)
KCNJ8	Sudden infant death syndrome
KCNK9	Birk-Barel mental retardation dysmorphism syndrome, 612292 (3)
KCNMA1	Generalized epilepsy and paroxysmal dyskinesia, 609446 (3)
KCNQ1	Atrial fibrillation, familial, 3, 607554 (3)/Jervell and Lange-Nielsen syndrome, 220400 (3)/Long QT syndrome 1, 192500 (3)/Short QT syndrome 2, 609621 (3)/Long QT syndrome 1, acquired, susceptibility to, 192500 (3)
KCNQ1OT1	Beckwith-Wiedemann syndrome 130650 (3)
KCNQ2	Epileptic encephalopathy, early infantile, 7, 613720 (3)/Myokymia, 121200 (3)/Seizures, benign neonatal, 1, 121200 (3)
KCNQ3	Seizures, benign neonatal, type 2, 121201 (3)
KCNQ4	Deafness, autosomal dominant 2A, 600101 (3)
KCNT1	Epilepsy, nocturnal frontal lobe, 5, 615005 (3)/Epileptic encephalopathy, early infantile, 14, 614959 (3)
KCNV2	Retinal cone dystrophy 3B, 610356 (3)
KCTD7	Epilepsy, progressive myoclonic 3, with or without intracellular inclusions, 611726 (3)
KDM5A	Congenital heart disease
KDM5C	Mental retardation, X-linked, syndromic, Claes-Jensen type, 300534 (3)
KDM6A	Kabuki syndrome 2, 300867 (3)

KDM6B	Intellectual disability
KDR	Hemangioma, capillary infantile, somatic, 602089 (3)/Hemangioma,capillary infantile, susceptibility to, 602089 (3)
KERA	Cornea plana congenita, recessive, 217300 (3)
KHDC3L	Hydatidiform mole, recurrent, 2, 614293 (3)
KIAA0196	Ritscher-Schinzel syndrome, 220210 (3)/Spastic paraplegia 8, autosomal dominant, 603563 (3)
KIAA0226	Spinocerebellar ataxia, autosomal recessive 15, 615705 (3)
KIAA1279	Goldberg-Shprintzen megacolon syndrome, 609460 (3)
KIAA2022	Mental retardation, X-linked 98, 300912 (3)
KIF11	Microcephaly with or without chorioretinopathy, lymphedema, or mental retardation, 152950 (3)
KIF1A	Mental retardation, autosomal dominant 9, 614255 (3)/Neuropathy, hereditary sensory, type IIC, 614213 (3)/Spastic paraplegia 30, autosomal recessive, 610357 (3)
KIF1B	Charcot-Marie-Tooth disease, type 2A1, 118210 (3)/Pheochromocytoma, 171300 (3)/Neuroblastoma, susceptibility to, 1, 256700 (3)
KIF21A	Fibrosis of extraocular muscles, congenital, 1, 135700 (3)/Fibrosis of extraocular muscles, congenital, 3B, 135700 (3)
KIF22	Spondyloepimetaphyseal dysplasia with joint laxity, type 2, 603546 (3)
KIF4A	Mental retardation, X-linked 100, 300923 (3)
KIF5A	Spastic paraplegia 10, autosomal dominant, 604187 (3)
KIF5C	Cortical dysplasia, complex, with other brain malformations 2, 615282 (3)
KIF7	Acrocallosal syndrome, 200990 (3)/Hydrolethalus syndrome 2, 614120 (3)/Joubert syndrome 12, 200990 (3)
KIRREL3	Mental retardation, autosomal dominant 4, 612581 (3)
KISS1	Hypogonadotropic hypogonadism 13 with or without anosmia, 614842 (3)
KISS1R	Precocious puberty, central, 1, 176400 (3)/Hypogonadotropic hypogonadism 8 with or without anosmia, 614837 (3)
KIT	Gastrointestinal stromal tumor, familial, 606764 (3)/Germ cell tumors, 273300 (3)/Leukemia, acute myeloid, 601626 (3)/Mast cell disease, 154800 (3)/Piebaldism, 172800 (3)
KITLG	Hyperpigmentation, familial progressive, 2, 145250 (3)/[Skin/hair/eye pigmentation 7, blond/brown hair], 611664 (3)
KL	Tumoral calcinosis, hyperphosphatemic, 211900 (3)/Coronary artery disease, susceptibility to (3)
KLF1	Blood group--Lutheran inhibitor, 111150 (3)/Dyserythropoietic anemia, congenital, type IV, 613673 (3)/[Hereditary persistence of fetal hemoglobin], 613566 (3)
KLF11	Maturity-onset diabetes of the young, type VII, 610508 (3)
KLF6	Gastric cancer, somatic, 613659 (3)/Prostate cancer, somatic, 176807 (3)
KLF8	Intellectual disability, absent speech & behavioural problems
KLHDC8B	Hodgkin lymphoma, susceptibility to, 246000 (3)
KLHL3	Pseudohypoaldosteronism, type IID, 614495 (3)
KLHL7	Retinitis pigmentosa 42, 612943 (3)
KLK4	Amelogenesis imperfecta, type IIA1, 204700 (3)
KLKB1	Fletcher factor deficiency, 612423 (3)
KMT2A	Leukemia, myeloid/lymphoid or mixed-lineage 159555 (3)/Wiedemann-Steiner syndrome 605130 (3)
KMT2D	Kabuki syndrome 1 147920 (3)
KRAS	Bladder cancer, somatic, 109800 (3)/Breast cancer, somatic, 114480 (3)/Cardiofaciocutaneous syndrome 2, 615278 (3)/Gastric cancer, somatic, 137215 (3)/Leukemia, acute myelogenous (3)/Lung cancer, somatic, 211980 (3)/Noonan syndrome 3, 609942 (3)/Pancreatic carcinoma, somatic, 260350 (3)/SFM syndrome, somatic mosaic, 163200 (3)
KRIT1	Cerebral cavernous malformations
KRT1	Epidermolytic hyperkeratosis, 113800 (3)/Ichthyosis histrix, Curth-Macklin type, 146590 (3)/Ichthyosis, cyclic, with epidermolytic hyperkeratosis, 607602 (3)/Keratosis palmoplantaris striata III, 607654 (3)/Palmoplantar keratoderma, epidermolytic, 144200 (3)/Palmoplantar keratoderma, nonepidermolytic, 600962 (3)
KRT10	Epidermolytic hyperkeratosis, 113800 (3)/Ichthyosis with confetti, 609165 (3)/Ichthyosis, cyclic, with epidermolytic hyperkeratosis, 607602 (3)
KRT12	Meesmann corneal dystrophy, 122100 (3)

KRT13	White sponge nevus 2, 615785 (3)
KRT14	Dermatopathia pigmentosa reticularis, 125595 (3)/Epidermolysis bullosa simplex, Dowling-Meara type, 131760 (3)/Epidermolysis bullosa simplex, Koebner type, 131900 (3)/Epidermolysis bullosa simplex, Weber-Cockayne type, 131800 (3)/Epidermolysis bullosa simplex, recessive 1, 601001 (3)/Naegeli-Franceschetti-Jadassohn syndrome, 161000 (3)
KRT16	Pachyonychia congenita 1, 167200 (3)/Palmoplantar keratoderma, nonepidermolytic, focal, 613000 (3)
KRT17	Pachyonychia congenita 2, 167210 (3)/Steatocystoma multiplex, 184500 (3)
KRT2	Ichthyosis bullosa of Siemens 146800 (3)
KRT3	Meesmann corneal dystrophy, 122100 (3)
KRT4	White sponge nevus 1, 193900 (3)
KRT5	Dowling-Degos disease 1, 179850 (3)/Epidermolysis bullosa simplex, Dowling-Meara type, 131760 (3)/Epidermolysis bullosa simplex, Koebner type, 131900 (3)/Epidermolysis bullosa simplex, Weber-Cockayne type, 131800 (3)/Epidermolysis bullosa simplex, recessive 1, 601001 (3)/Epidermolysis bullosa simplex-MP, 131960 (3)/Epidermylysis bullosa simplex-MCR, 609352 (3)
KRT6A	Pachyonychia congenita 3, 615726 (3)
KRT6B	Pachyonychia congenita 4, 615728 (3)
KRT74	Ectodermal dysplasia 7, hair/nail type, 614929 (3)/
KRT81	Monilethrix, 158000 (3)
KRT83	Monilethrix, 158000 (3)
KRT85	Ectodermal dysplasia 4, hair/nail type, 602032 (3)
KRT86	Monilethrix, 158000 (3)
KRT9	Palmoplantar keratoderma, epidermolytic, 144200 (3)
L1CAM	CRASH syndrome, 303350 (3)/Corpus callosum, partial agenesis of, 304100 (3)/Hydrocephalus due to aqueductal stenosis, 307000 (3)/Hydrocephalus with Hirschsprung disease, 307000 (3)/Hydrocephalus with congenital idiopathic intestinal pseudoobstruction, 307000 (3)/MASA syndrome, 303350 (3)
L2HGDH	L-2-hydroxyglutaric aciduria, 236792 (3)
LAMA1	Poretti-Boltshauser syndrome, 615960 (3)
LAMA2	Muscular dystrophy, congenital merosin-deficient, 607855 (3)/Muscular dystrophy, congenital, due to partial LAMA2 deficiency, 607855 (3)
LAMA3	Epidermolysis bullosa, generalized atrophic benign, 226650 (3)/Epidermolysis bullosa, junctional, Herlitz type, 226700 (3)/Laryngoonychocutaneous syndrome, 245660 (3)
LAMA4	Cardiomyopathy, dilated, 1JJ, 615235 (3)
LAMB1	Lissencephaly 5, 615191 (3)
LAMB2	Nephrotic syndrome, type 5, with or without ocular abnormalities, 614199 (3)/Pierson syndrome, 609049 (3)
LAMB3	Amelogenesis imperfecta, type IA, 104530 (3)/Epidermolysis bullosa, junctional, Herlitz type, 226700 (3)/Epidermolysis bullosa, junctional, non-Herlitz type, 226650 (3)
LAMC2	Epidermolysis bullosa, junctional, Herlitz type, 226700 (3)/Epidermolysis bullosa, junctional, non-Herlitz type, 226650 (3)
LAMC3	Cortical malformations, occipital, 614115 (3)
LAMP2	Danon disease, 300257 (3)
LAMTOR2	Immunodeficiency due to defect in MAPBP-interacting protein, 610798 (3)
LARGE	Muscular dystrophy-dystroglycanopathy (congenital with brain and eye anomalies), type A, 6, 613154 (3)/Muscular dystrophy-dystroglycanopathy (congenital with mental retardation), type B, 6, 608840 (3)
LARP7	Alazami syndrome, 615071 (3)
LBR	Reynolds syndrome, 613471 (3)/Greenberg skeletal dysplasia, 215140 (3)/Pelger-Huet anomaly, 169400 (3)
LCA5	Leber congenital amaurosis 5, 604537 (3)
LCAT	Fish-eye disease, 136120 (3)/Norum disease, 245900 (3)
LCT	Lactase deficiency, congenital, 223000 (3)
LDB3	Cardiomyopathy, dilated 1C, 601493 (3)/Left ventricular noncompaction 3, with or without dilated cardiomyopathy, 601493 (3)/Myopathy, myofibrillar, 4, 609452 (3)
LDHA	Glycogen storage disease XI, 612933 (3)
LDHB	Lactate dehydrogenase-B deficiency, 614128 (3)
LDLR	Hypercholesterolemia, familial 143890 (3)/LDL cholesterol level QTL2 143890 (3)
LDLRAP1	Hypercholesterolemia, familial, autosomal recessive, 603813 (3)

LEMD3	Buschke-Ollendorff syndrome, 166700 (3)/Melorheostosis with osteopoikilosis, 155950 (3)/Osteopoikilosis, 166700 (3)
LEP	Obesity, morbid, due to leptin deficiency, 614962 (3)
LEPR	Obesity, morbid, due to leptin receptor deficiency, 614963 (3)
LEPRE1	Osteogenesis imperfecta, non-lethal
LEPREL1	High myopia
LFNG	Spondylocostal dysostosis 3, autosomal recessive, 609813 (3)
LGI1	Epilepsy, familial temporal lobe, 1, 600512 (3)
LHB	Male pseudohermaphroditism due to defective LH (1)/Hypogonadism, hypergonadotropic (3)
LHCGR	Leydig cell adenoma, somatic, with precocious puberty, 176410 (3)/Leydig cell hypoplasia with hypergonadotropic hypogonadism, 238320 (3)/Leydig cell hypoplasia with pseudohermaphroditism, 238320 (3)/Luteinizing hormone resistance, female, 238320 (3)/Precocious puberty, male, 176410 (3)
LHFPL5	Deafness, autosomal recessive 67, 610265 (3)
LHX3	Pituitary hormone deficiency, combined, 3, 221750 (3)
LHX4	Pituitary hormone deficiency, combined, 4, 262700 (3)
LIAS	Pyruvate dehydrogenase lipoic acid synthetase deficiency, 614462 (3)
LIFR	Stuve-Wiedemann syndrome/Schwartz-Jampel type 2 syndrome, 601559 (3)
LIG4	LIG4 syndrome, 606593 (3)/Multiple myeloma, resistance to, 254500 (3)
LINS	Intellectual disability
LIPA	Cholesteryl ester storage disease, 278000 (3)/Wolman disease, 278000 (3)
LIPC	Hepatic lipase deficiency, 614025 (3)/[High density lipoprotein cholesterol level QTL 12], 612797 (3)/Diabetes mellitus, noninsulin-dependent, 125853 (3)
LIPH	Hypotrichosis 7, 604379 (3)/Woolly hair, autosomal recessive 2 with or without hypotrichosis, 604379 (3)
LIPN	Ichthyosis, congenital, autosomal recessive 8, 613943 (3)
LITAF	Charcot-Marie-Tooth disease, type 1C, 601098 (3)
LMAN1	Combined factor V and VIII deficiency, 227300 (3)
LMBR1	Acheiropody, 200500 (3)/Hypoplastic or aplastic tibia with polydactyly, 188740 (3)/Laurin-Sandrow syndrome, 135750 (3)/Polydactyly, preaxial type II, 174500 (3)/Syndactyly, type IV, 186200 (3)/Triphalangeal thumb, type I, 174500 (3)/Triphalangeal thumb-polysyndactyly syndrome, 174500 (3)
LMBRD1	Methylmalonic aciduria and homocystinuria, cblF type, 277380 (3)
LMF1	Lipase deficiency, combined, 246650 (3)
LMNA	Cardiomyopathy, dilated, 1A, 115200 (3)/Charcot-Marie-Tooth disease, type 2B1, 605588 (3)/Emery-Dreifuss muscular dystrophy 2, AD, 181350 (3)/Emery-Dreifuss muscular dystrophy 3, AR, 181350 (3)/Heart-hand syndrome, Slovenian type, 610140 (3)/Hutchinson-Gilford progeria, 176670 (3)/Lipodystrophy, familial partial, 2, 151660 (3)/Malouf syndrome, 212112 (3)/Mandibuloacral dysplasia, 248370 (3)/Muscular dystrophy, congenital, 613205 (3)/Muscular dystrophy, limb-girdle, type 1B, 159001 (3)/Restrictive dermopathy, lethal, 275210 (3)
LMNB1	Leukodystrophy, adult-onset, autosomal dominant, 169500 (3)
LMX1B	Nail-patella syndrome, 161200 (3)
LOR	Vohwinkel syndrome with ichthyosis, 604117 (3)
LOXHD1	Deafness, autosomal recessive 77, 613079 (3)
LPA	[LPA deficiency, congenital] (3)/Coronary artery disease, susceptibility to (1)
LPAR6	Hypotrichosis 8, 278150 (3)/Woolly hair, autosomal recessive 1, with or without hypotrichosis, 278150 (3)
LPIN1	Myoglobinuria, acute recurrent, autosomal recessive, 268200 (3)
LPIN2	Majeed syndrome, 609628 (3)
LPL	Combined hyperlipidemia, familial, 144250 (3)/Lipoprotein lipase deficiency, 238600 (3)/[High density lipoprotein cholesterol level QTL 11] (3)
LPP	Leukemia, acute myeloid, 601626 (3)/Lipoma (3)
LRAT	Leber congenital amaurosis 14, 613341 (3)/Retinal dystrophy, early-onset severe, 613341 (3)/Retinitis pigmentosa, juvenile, 613341 (3)
LRBA	Immunodeficiency, common variable, 8, with autoimmunity, 614700 (3)
LRP1	?Keratosis pilaris atrophicans 604093 (3)
LRP2	Donnai-Barrow syndrome, 222448 (3)
LRP4	Cenani-Lenz syndactyly syndrome, 212780 (3)/Sclerosteosis 2, 614305 (3)

LRP5	Exudative vitreoretinopathy 4, 601813 (3)/Hyperostosis, endosteal, 144750 (3)/Osteopetrosis, autosomal dominant 1, 607634 (3)/Osteoporosis-pseudoglioma syndrome, 259770 (3)/Osteosclerosis, 144750 (3)/[Bone mineral density variability 1], 601884 (3)/van Buchem disease, type 2, 607636 (3)/Osteoporosis, 166710 (3)
LRPPRC	Leigh syndrome, French-Canadian type, 220111 (3)
LRRC6	Ciliary dyskinesia, primary, 19, 614935 (3)
LRRC8A	Agammaglobulinemia 5, 613506 (3)
LRRK2	Parkinson disease 8, 607060 (3)
LRSAM1	Charcot-Marie-Toothe disease, axonal, type 2P, 614436 (3)
LRTOMT	Deafness, autosomal recessive 63, 611451 (3)
LTBP2	Glaucoma 3, primary congenital, D, 613086 (3)/Microspherophakia and/or megalocornea, with ectopia lentis and with or without secondary glaucoma, 251750 (3)/Weill-Marchesani syndrome 3, recessive, 614819 (3)
LTBP3	Tooth agenesis, selective, 6, 613097 (3)
LTBP4	Cutis laxa, autosomal recessive, type IC, 613177 (3)
LTC4S	Leukotriene C4 synthase deficiency, 614037 (1)
LYL1	Leukemia, T-cell acute lymphoblastoid (2)
LYST	Chediak-Higashi syndrome, 214500 (3)
LYZ	Amyloidosis, renal, 105200 (3)
LZTFL1	Bardet-Biedl syndrome 17, 615994 (3)
LZTS1	Esophageal squamous cell carcinoma, 133239 (3)
MAD1L1	Lymphoma, somatic (3)/Prostate cancer, somatic, 176807 (3)
MAF	Cataract 21, multiple types, 610202 (3)
MAFB	Multicentric carpotarsal osteolysis syndrome, 166300 (3)
MAGEL2	Prader-Willi-like syndrome, 615547 (3)
MAGT1	Immunodeficiency, X-linked, with magnesium defect, Epstein-Barr virus infection and neoplasia, 300853 (3)
MAK	Retinitis pigmentosa 62, 614181 (3)
MAMLD1	Hypospadias 2, X-linked, 300758 (3)
MAN1B1	Mental retardation, autosomal recessive 15, 614202 (3)
MAN2B1	Mannosidosis, alpha-, types I and II, 248500 (3)
MANBA	Mannosidosis, beta, 248510 (3)
MAOA	Brunner syndrome, 300615 (3)
MAP2K1	Cardiofaciocutaneous syndrome 3, 615279 (3)
MAP2K2	Cardiofaciocutaneous syndrome 4, 615280 (3)
MAP3K1	46XY sex reversal 6, 613762 (3)
MAP3K8	Lung cancer, somatic, 211980 (3)
MAPK10	Hirschsprung disease, modifier of
MAPT	Dementia, frontotemporal, with or without parkinsonism, 600274 (3)/Pick disease, 172700 (3)/Supranuclear palsy, progressive atypical, 260540 (3)/Supranuclear palsy, progressive, 601104 (3)/Parkinson disease, susceptibility to, 168600 (3)
MARVELD2	Deafness, autosomal recessive 49, 610153 (3)
MASP1	3MC syndrome 1, 257920 (3)
MASP2	MASP2 deficiency, 613791 (3)
MASTL	Thrombocytopenia-2, 188000 (3)
MAT1A	Hypermethioninemia, persistent, autosomal dominant, due to methionine adenosyltransferase I/III deficiency, 250850 (3)/Methionine adenosyltransferase deficiency, autosomal recessive, 250850 (3)
MATN3	Epiphyseal dysplasia, multiple, 5, 607078 (3)/Spondyloepimetaphyseal dysplasia, 608728 (3)/Osteoarthritis susceptibility 2, 140600 (3)
MATR3	Amyotrophic lateral sclerosis 21, 606070 (3)
MBD5	Mental retardation, autosomal dominant 1, 156200 (3)
MBTPS2	Olmsted syndrome, X-linked, 300918 (3)/IFAP syndrome with or without BRESHECK syndrome, 308205 (3)/Keratosis follicularis spinulosa decalvans, X-linked, 308800 (3)

MC1R	[Analgesia from kappa-opioid receptor agonist, female-specific], 613098 (3)/[Skin/hair/eye pigmentation 2, blond hair/fair skin], 266300 (3)/[Skin/hair/eye pigmentation 2, red hair/fair skin], 266300 (3)/Albinism, oculocutaneous, type II, modifier of, 203200 (3)/Melanoma, cutaneous malignant, 5, 613099 (3)/UV-induced skin damage, 266300 (3)
MC2R	Glucocorticoid deficiency, due to ACTH unresponsiveness, 202200 (3)
MC4R	Obesity, autosomal dominant, 601665 (3)
MCCC1	3-Methylcrotonyl-CoA carboxylase 1 deficiency, 210200 (3)
MCCC2	3-Methylcrotonyl-CoA carboxylase 2 deficiency, 210210 (3)
MCEE	Methylmalonyl-CoA epimerase deficiency, 251120 (3)
MCFD2	Factor V and factor VIII, combined deficiency of, 613625 (3)
MCM4	Natural killer cell and glucocorticoid deficiency with DNA repair defect, 609981 (3)
MCM6	Lactase persistence/nonpersistence, 223100 (3)
MCOLN1	Mucolipidosis IV, 252650 (3)
MCPH1	Microcephaly 1, primary, autosomal recessive, 251200 (3)
MECP2	Angelman syndrome, 105830 (3)/Encephalopathy, neonatal severe, 300673 (3)/Mental retardation, X-linked syndromic, Lubs type, 300260 (3)/Mental retardation, X-linked, syndromic 13, 300055 (3)/Rett syndrome, 312750 (3)/Rett syndrome, preserved speech variant, 312750 (3)/Autism susceptibility, X-linked 3, 300496 (3)
MED12	Lujan-Fryns syndrome, 309520 (3)/Ohdo syndrome, X-linked, 300895 (3)/Opitz-Kaveggia syndrome, 305450 (3)
MED13L	Transposition of the great arteries, dextro-looped 1, 608808 (3)
MED17	Microcephaly, postnatal progressive, with seizures and brain atrophy, 613668 (3)
MED23	Mental retardation, autosomal recessive 18, 614249 (3)
MED25	Charcot-Marie-Tooth disease, type 2B2, 605589 (3)
MEF2C	Chromosome 5q14.3 deletion syndrome, 613443 (4)/Mental retardation, stereotypic movements, epilepsy, and/or cerebral malformations, 613443 (3)
MEFV	Familial Mediterranean fever, AD, 134610 (3)/Familial Mediterranean fever, AR, 249100 (3)
MEGF10	Myopathy, areflexia, respiratory distress, and dysphagia, early-onset, 614399 (3)/Myopathy, areflexia, respiratory distress, and dysphagia, early-onset, mild variant, 614399 (3)
MEN1	Adrenal adenoma, somatic (3)/Angiofibroma, somatic (3)/Carcinoid tumor of lung (3)/Lipoma, somatic (3)/Multiple endocrine neoplasia 1, 131100 (3)/Parathyroid adenoma, somatic (3)
MERTK	Retinitis pigmentosa 38, 613862 (3)
MESP2	Spondylocostal dysostosis 2, autosomal recessive, 608681 (3)
MET	Hepatocellular carcinoma, childhood type, 114550 (3)/Renal cell carcinoma, papillary, 1, familial and somatic, 605074 (3)
MFN2	Charcot-Marie-Tooth disease, type 2A2, 609260 (3)/Hereditary motor and sensory neuropathy VI, 601152 (3)
MFRP	Microphthalmia, isolated 5, 611040 (3)/Nanophthalmos 2, 609549 (3)
MFSD8	Ceroid lipofuscinosis, neuronal, 7, 610951 (3)/Macular dystrophy with central cone involvement, 616170 (3)
MGAT2	Congenital disorder of glycosylation, type IIa, 212066 (3)
MGP	Keutel syndrome, 245150 (3)
MIB1	Left ventricular noncompaction 7, 615092 (3)
MID1	Opitz GBBB syndrome, type I 300000 (3)
MIF	Rheumatoid arthritis, systemic juvenile, susceptibility to, 604302 (3)
MINPP1	Thyroid carcinoma, follicular, 188470 (3)
MIPOL1	Craniofacial/acallosal CNS midline defects
MITF	Tietz albinism-deafness syndrome, 103500 (3)/Waardenburg syndrome, type 2A, 193510 (3)/Waardenburg syndrome/ocular albinism, digenic, 103470 (3)/Melanoma, cutaneous malignant, susceptibility to, 8, 614456 (3)
MKKS	Bardet-Biedl syndrome 6, 605231 (3)/McKusick-Kaufman syndrome, 236700 (3)
MKS1	Bardet-Biedl syndrome 13, 615990 (3)/Meckel syndrome 1, 249000 (3)
MLC1	Megalencephalic leukoencephalopathy with subcortical cysts, 604004 (3)
MLF1	Leukemia, acute myeloid, 601626 (1)
MLH1	Colorectal cancer, hereditary nonpolyposis, type 2, 609310 (3)/Mismatch repair cancer syndrome, 276300 (3)/Muir-Torre syndrome, 158320 (3)

MLH3	Colorectal cancer, hereditary nonpolyposis, type 7, 614385 (3)/Colorectal cancer, somatic, 114500 (3)/Endometrial cancer, susceptibility to, 608089 (3)
MLL	Wiedemann-Steiner syndrome
MLL2	Kabuki syndrome
MLLT10	Leukemia, acute myeloid 601626 (3)
MLLT3	Neuromotor dev. delay, cerebellar ataxia, epilepsy
MLPH	Griscelli syndrome, type 3, 609227 (3)
MLYCD	Malonyl-CoA decarboxylase deficiency, 248360 (3)
MMAA	Methylmalonic aciduria, vitamin B12-responsive, 251100 (3)
MMAB	Methylmalonic aciduria, vitamin B12-responsive, due to defect in synthesis of adenosylcobalamin, cblB complementation type, 251110 (3)
MMACHC	Methylmalonic aciduria and homocystinuria, cblC type, 277400 (3)
MMADHC	Homocystinuria, cblD type
MMP1	COPD, rate of decline of lung function in, 606963 (3)/Epidermolysis bullosa dystrophica, autosomal recessive, modifier of, 226600 (3)
MMP13	Metaphyseal anadysplasia 1, 602111 (3)/Spondyloepimetaphyseal dysplasia, Missouri type, 602111 (3)
MMP2	Multicentric osteolysis, nodulosis, and arthropathy, 259600 (3)
MMP20	Amelogenesis imperfecta, type IIA2, 612529 (3)
MMP9	Metaphyseal anadysplasia 2, 613073 (3)
MN1	Meningioma, 607174 (3)
MNX1	Currarino syndrome, 176450 (3)
MOCS1	Molybdenum cofactor deficiency A, 252150 (3)
MOCS2	Molybdenum cofactor deficiency B, 252160 (3)
MOG	Narcolepsy 7, 614250 (3)
MOGS	Congenital disorder of glycosylation, type IIb, 606056 (3)
MPDU1	Congenital disorder of glycosylation, type If, 609180 (3)
MPI	Congenital disorder of glycosylation, type Ib, 602579 (3)
MPL	Myelofibrosis with myeloid metaplasia, somatic, 254450 (3)/Thrombocythemia 2, 601977 (3)/Thrombocytopenia, congenital amegakaryocytic, 604498 (3)
MPLKIP	Trichothiodystrophy, nonphotosensitive 1, 234050 (3)
MPO	Myeloperoxidase deficiency, 254600 (3)/Alzheimer disease, susceptibility to, 104300 (3)/Lung cancer, protection against, in smokers (3)
MPV17	Mitochondrial DNA depletion syndrome 6 (hepatocerebral type), 256810 (3)
MPZ	Charcot-Marie-Tooth disease, dominant intermediate D, 607791 (3)/Charcot-Marie-Tooth disease, type 1B, 118200 (3)/Charcot-Marie-Tooth disease, type 2I, 607677 (3)/Charcot-Marie-Tooth disease, type 2J, 607736 (3)/Dejerine-Sottas disease, 145900 (3)/Neuropathy, congenital hypomyelinating, 605253 (3)/Roussy-Levy syndrome, 180800 (3)
MRAP	Glucocorticoid deficiency 2, 607398 (3)
MRE11A	Ataxia-telangiectasia-like disorder, 604391 (3)
MRPL3	Combined oxidative phosphorylation deficiency 9, 614582 (3)
MRPS16	Combined oxidative phosphorylation deficiency 2, 610498 (3)
MRPS22	Combined oxidative phosphorylation deficiency 5, 611719 (3)
MS4A1	Immunodeficiency, common variable, 5, 613495 (3)
MS4A2	{Atopy, susceptibility to}, 147050 (3)
MSH2	Colorectal cancer, hereditary nonpolyposis, type 1, 120435 (3)/Mismatch repair cancer syndrome, 276300 (3)/Muir-Torre syndrome, 158320 (3)
MSH6	Colorectal cancer, hereditary nonpolyposis, type 5, 614350 (3)/Endometrial cancer, familial, 608089 (3)/Mismatch repair cancer syndrome, 276300 (3)
MSR1	Barrett esophagus/esophageal adenocarcinoma, 614266 (3)/Prostate cancer, hereditary, 176807 (3)
MSRB3	Deafness, autosomal recessive 74, 613718 (3)
MSTN	Muscle hypertrophy, gross
MSX1	Ectodermal dysplasia 3, Witkop type, 189500 (3)/Orofacial cleft 5, 608874 (3)/Tooth agenesis, selective, 1, with or without orofacial cleft, 106600 (3)

MSX2	Craniosynostosis, type 2, 604757 (3)/Parietal foramina 1, 168500 (3)/Parietal foramina with cleidocranial dysplasia, 168550 (3)
MTAP	Diaphyseal medullary stenosis with malignant fibrous histiocytoma, 112250 (3)
MTFMT	Combined oxidative phosphorylation deficiency 15, 614947 (3)
MTHFR	Homocystinuria due to MTHFR deficiency, 236250 (3)/Neural tube defects, susceptibility to, 601634 (3)/Schizophrenia, susceptibility to, 181500 (3)/Thromboembolism, susceptibility to, 188050 (3)/Vascular disease, susceptibility to (3)
MTM1	Myotubular myopathy, X-linked, 310400 (3)
MTMR2	Charcot-Marie-Tooth disease, type 4B1, 601382 (3)
MTO1	Combined oxidative phosphorylation deficiency 10, 614702 (3)
MTPAP	Ataxia, spastic, 4, 613672 (3)
MTR	Homocystinuria-megaloblastic anemia, cblG complementation type, 250940 (3)/Neural tube defects, folate-sensitive, susceptibility to, 601634 (3)
MTRR	Homocystinuria-megaloblastic anemia, cbl E type, 236270 (3)/Neural tube defects, folate-sensitive, susceptibility to, 601634 (3)
MTTP	Abetalipoproteinaemia
MUSK	Myasthenic syndrome, congenital, associated with acetylcholine receptor deficiency, 608931 (3)
MUT	Methylmalonic aciduria, mut(0) type, 251000 (3)
MUTYH	Adenomas, multiple colorectal, 608456 (3)/Colorectal adenomatous polyposis, autosomal recessive, with pilomatricomas, 132600 (3)/Gastric cancer, somatic, 613659 (3)
MVK	Hyper-IgD syndrome, 260920 (3)/Mevalonic aciduria, 610377 (3)/Porokeratosis 3, disseminated superficial actinic, 175900 (3)
MYB	T-cell acute lymphoblastic leukemia (3)
MYBPC1	Arthrogryposis, distal, type 1B, 614335 (3)/Lethal congenital contracture syndrome 4, 614915 (3)
MYBPC3	Cardiomyopathy, dilated, 1MM, 615396 (3)/Cardiomyopathy, familial hypertrophic, 4, 115197 (3)/Left ventricular noncompaction 10, 615396 (3)
MYC	Burkitt lymphoma, 113970 (3)
MYCN	Feingold syndrome, 164280 (3)
MYD88	Macroglobulinemia, Waldenstrom, somatic, 153600 (3)/Pyogenic bacterial infections, recurrent, due to MYD88 deficiency, 612260 (3)
MYF6	Myopathy, centronuclear, 3, 614408 (3)
MYH11	Aortic aneurysm, familial thoracic 4, 132900 (3)
MYH14	Peripheral neuropathy, myopathy, hoarseness, and hearing loss, 614369 (3)/Deafness, autosomal dominant 4A, 600652 (3)
MYH2	Inclusion body myopathy-3, 605637 (3)
MYH3	Arthrogryposis, distal, type 2A, 193700 (3)/Arthrogryposis, distal, type 2B, 601680 (3)
MYH6	Atrial septal defect 3, 614089 (3)/Cardiomyopathy, dilated, 1EE, 613252 (3)/Cardiomyopathy, familial hypertrophic, 14, 613251 (3)/Sick sinus syndrome 3, 614090 (3)
MYH7	Cardiomyopathy, dilated, 1S, 613426 (3)/Cardiomyopathy, familial hypertrophic, 1, 192600 (3)/Laing distal myopathy, 160500 (3)/Left ventricular noncompaction 5, 613426 (3)/Myopathy, myosin storage, 608358 (3)/Scapuloperoneal syndrome, myopathic type, 181430 (3)
MYH8	Carney complex variant, 608837 (3)/Trismus-pseudocamptodactyly syndrome, 158300 (3)
MYH9	Deafness, autosomal dominant 17, 603622 (3)/Epstein syndrome, 153650 (3)/Fechtner syndrome, 153640 (3)/Macrothrombocytopenia and progressive sensorineural deafness, 600208 (3)/May-Hegglin anomaly, 155100 (3)/Sebastian syndrome, 605249 (3)
MYL2	Cardiomyopathy, familial hypertrophic, 10, 608758 (3)
MYL3	Cardiomyopathy, familial hypertrophic, 8, 608751 (3)
MYLK	Aortic aneurysm, familial thoracic 7, 613780 (3)
MYLK2	Cardiomyopathy, hypertrophic, midventricular, digenic, 192600 (3)
MYO15A	Deafness, autosomal recessive 3, 600316 (3)
MYO1A	Deafness, autosomal dominant 48, 607841 (3)
MYO1E	Glomerulosclerosis, focal segmental, 6, 614131 (3)
MYO3A	Deafness, autosomal recessive 30, 607101 (3)
MYO5A	Griscelli syndrome, type 1, 214450 (3)

MYO5B	Microvillus inclusion disease, 251850 (3)
MYO6	Deafness, autosomal dominant 22, 606346 (3)/Deafness, autosomal dominant 22, with hypertrophic cardiomyopathy, 606346 (3)/Deafness, autosomal recessive 37, 607821 (3)
MYO7A	Deafness, autosomal dominant 11, 601317 (3)/Deafness, autosomal recessive 2, 600060 (3)/Usher syndrome, type 1B, 276900 (3)
MYOC	Glaucoma 1A, primary open angle, 137750 (3)
MYOT	Muscular dystrophy, limb-girdle, type 1A, 159000 (3)/Myopathy, myofibrillar, 3, 609200 (3)/Myopathy, spheroid body, 182920 (3)
MYOZ2	Cardiomyopathy, familial hypertrophic, 16, 613838 (3)
MYPN	Cardiomyopathy, dilated, 1KK, 615248 (3)/Cardiomyopathy, familial hypertrophic, 22, 615248 (3)/Cardiomyopathy, familial restrictive, 4, 615248 (3)
MYT1L	Intellectual disability
NAA10	Microphthalmia, syndromic 1, 309800 (3)/N-terminal acetyltransferase deficiency, 300855 (3)
NAGA	Kanzaki disease, 609242 (3)/Schindler disease, type I, 609241 (3)/Schindler disease, type III, 609241 (3)
NAGLU	Mucopolysaccharidosis type IIIB (Sanfilippo B), 252920 (3)
NAGS	N-acetylglutamate synthase deficiency, 237310 (3)
NAT8L	N-acetylaspartate deficiency, 614063 (3)
NBAS	Short stature, optic nerve atrophy, and Pelger-Huet anomaly, 614800 (3)
NBEAL2	Gray platelet syndrome, 139090 (3)
NBN	Nijmegen breakage syndrome 251260/Leukemia, acute lymphoblastic 613065/Aplastic anemia 609135 (3)
NCF1	Chronic granulomatous disease due to deficiency of NCF-1, 233700 (3)
NCF2	Chronic granulomatous disease due to deficiency of NCF-2, 233710 (3)
NCF4	Granulomatous disease, chronic, autosomal recessive, cytochrome b-positive, type III, 613960 (3)
NCOA4	Thyroid carcinoma, papillary, 188550 (3)
NCSTN	Acne inversa, familial, 1, 142690 (3)
NDE1	Microhydranencephaly, 605013 (3)/Lissencephaly 4 (with microcephaly), 614019 (3)
NDN	Prader-Willi syndrome, 176270 (3)
NDP	Exudative vitreoretinopathy 2, X-linked, 305390 (3)/Norrie disease, 310600 (3)
NDRG1	Charcot-Marie-Tooth disease, type 4D, 601455 (3)
NDST1	Mental retardation, autosomal recessive 46, 616116 (3)
NDUFA1	Mitochondrial complex I deficiency, 252010 (3)
NDUFA10	Leigh syndrome, 256000 (3)
NDUFA11	Mitochondrial complex I deficiency, 252010 (3)
NDUFA12	Leigh syndrome due to mitochondrial complex 1 deficiency, 256000 (3)
NDUFA2	Leigh syndrome due to mitochondrial complex I deficiency, 256000 (3)
NDUFA9	Leigh syndrome due to mitochondrial complex I deficiency, 256000 (3)
NDUFAF1	Mitochondrial complex I deficiency, 252010 (3)
NDUFAF2	Leigh syndrome, 256000 (3)/Mitochondrial complex I deficiency, 252010 (3)
NDUFAF3	Mitochondrial complex I deficiency, 252010 (3)
NDUFAF4	Mitochondrial complex I deficiency, 252010 (3)
NDUFAF5	Mitochondrial complex 1 deficiency 252010 (3)
NDUFAF6	Leigh syndrome due to mitochondrial complex I deficiency 256000 (3)
NDUFB3	Mitochondrial complex I deficiency, 252010 (3)
NDUFS1	Mitochondrial complex I deficiency, 252010 (3)
NDUFS2	Mitochondrial complex I deficiency, 252010 (3)
NDUFS3	Leigh syndrome due to mitochondrial complex I deficiency, 256000 (3)/Mitochondrial complex I deficiency, 252010 (3)
NDUFS4	Leigh syndrome, 256000 (3)/Mitochondrial complex I deficiency, 252010 (3)
NDUFS5	Complex I deficiency
NDUFS6	Complex I, mitochondrial respiratory chain, deficiency of, 252010 (3)
NDUFS7	Leigh syndrome, 256000 (3)
NDUFS8	Leigh syndrome due to mitochondrial complex I deficiency, 256000 (3)
NDUFV1	Mitochondrial complex I deficiency, 252010 (3)
NDUFV2	Mitochondrial complex I deficiency, 252010 (3)

NEB	Nemaline myopathy 2, autosomal recessive, 256030 (3)
NEFL	Charcot-Marie-Tooth disease, type 1F, 607734 (3)/Charcot-Marie-Tooth disease, type 2E, 607684 (3)
NEK1	Short-rib thoracic dysplasia 6 with or without polydactyly, 263520 (3)
NEK8	Nephronophthisis 9, 613824 (3)/
NELF	Hypogonadotropic hypogonadism, idiopathic
NEU1	Sialidosis, type I, 256550 (3)/Sialidosis, type II, 256550 (3)
NEUROD1	Maturity-onset diabetes of the young 6, 606394 (3)/Diabetes mellitus, noninsulin-dependent, 125853 (3)
NEUROG3	Diarrhea 4, malabsorptive, congenital, 610370 (3)
NEXN	Cardiomyopathy, dilated, 1CC, 613122 (3)/Cardiomyopathy, familial hypertrophic, 20, 613876 (3)
NF1	Leukemia, juvenile myelomonocytic, 607785 (3)/Neurofibromatosis, familial spinal, 162210 (3)/Neurofibromatosis, type 1, 162200 (3)/Neurofibromatosis-Noonan syndrome, 601321 (3)/Watson syndrome, 193520 (3)
NF2	Meningioma, NF2-related, somatic, 607174 (3)/Neurofibromatosis, type 2, 101000 (3)/Schwannomatosis, 162091 (3)
NFIX	Marshall-Smith syndrome, 602535 (3)/Sotos syndrome 2, 614753 (3)
NFKBIA	Ectodermal dysplasia, anhidrotic, with T-cell immunodeficiency, 612132 (3)
NFU1	Multiple mitochondrial dysfunctions syndrome 1, 605711 (3)
NGF	Neuropathy, hereditary sensory and autonomic, type V, 608654 (3)
NHEJ1	Severe combined immunodeficiency with microcephaly, growth retardation, and sensitivity to ionizing radiation, 611291 (3)
NHLRC1	Epilepsy, progressive myoclonic 2B (Lafora) 254780 (3)
NHP2	Dyskeratosis congenita
NHS	Cataract 40, X-linked, 302200 (3)/Nance-Horan syndrome, 302350 (3)
NIN	Seckel syndrome 7, 614851 (3)
NIPA1	Spastic paraplegia 6, autosomal dominant, 600363 (3)
NIPAL4	Ichthyosis, congenital, autosomal recessive 6, 612281 (3)
NIPBL	Cornelia de Lange syndrome 1, 122470 (3)
NKX2-1	Chorea, hereditary benign, 118700 (3)/Choreoathetosis, hypothyroidism, and neonatal respiratory distress, 610978 (3)/Goiter, familial, due to TTF-1 defect (1)
NKX2-5	Atrial septal defect 7, with or without AV conduction defects, 108900 (3)/Conotruncal heart malformations, variable, 217095 (3)/Hypoplastic left heart syndrome 2, 614435 (3)/Hypothyroidism, congenital nongoitrous, 5, 225250 (3)/Tetrology of Fallot, 187500 (3)/Ventricular septal defect 3, 614432 (3)
NKX2-6	Conotruncal heart malformations, 217095 (3)/Persistent truncus arteriosus, 217095 (3)
NKX3-2	Spondylo-megaepiphyseal-metaphyseal dysplasia, 613330 (3)
NLGN3	Asperger syndrome susceptibility, X-linked 1, 300494 (3)/Autism susceptibility, X-linked 1, 300425 (3)
NLGN4X	Mental retardation
NLRP1	Corneal intraepithelial dyskeratosis and ectodermal dysplasia, 615225 (3)/Vitiligo-associated multiple autoimmune disease susceptibility 1, 606579 (3)
NLRP12	Familial cold autoinflammatory syndrome 2, 611762 (3)
NLRP3	CINCA syndrome, 607115 (3)/Familial cold-induced inflammatory syndrome 1, 120100 (3)/Muckle-Wells syndrome, 191900 (3)
NLRP7	Hydatidiform mole
NME1	Neuroblastoma, 256700 (3)
NME8	Ciliary dyskinesia, primary, 6, 610852 (3)
NMNAT1	Leber congenital amaurosis 9, 608553 (3)
NNT	Glucocorticoid deficiency 4, 614736 (3)
NOBOX	Premature ovarian failure 5, 611548 (3)
NOD2	Blau syndrome, 186580 (3)/Sarcoidosis, early-onset, 609464 (3)/Inflammatory bowel disease 1, 266600 (3)/Psoriatic arthritis, susceptibility to, 607507 (3)
NODAL	Heterotaxy, visceral, 5, 270100 (3)
NOG	Brachydactyly, type B2, 611377 (3)/Multiple synostoses syndrome 1, 186500 (3)/Stapes ankylosis with broad thumb and toes, 184460 (3)/Symphalangism, proximal, 185800 (3)/Tarsal-carpal coalition syndrome, 186570 (3)
NOL3	Myoclonus, familial cortical, 614937 (3)

NOP10	Dyskeratosis congenita, autosomal recessive 1 224230 (3)
NOP56	Spinocerebellar ataxia 36, 614153 (3)
NOTCH1	Adams-Oliver syndrome 5, 616028 (3)/Aortic valve disease 1, 109730 (3)
NOTCH2	Alagille syndrome 2, 610205 (3)/Hajdu-Cheney syndrome, 102500 (3)
NOTCH3	Myofibromatosis, infantile 2, 615293 (3)/Cerebral arteriopathy with subcortical infarcts and leukoencephalopathy, 125310 (3)
NPC1	Niemann-Pick disease, type C1, 257220 (3)/Niemann-Pick disease, type D, 257220 (3)/Nasopharyngeal carcinoma 1 (2)
NPC2	Niemann-pick disease, type C2, 607625 (3)
NPHP1	Joubert syndrome 4, 609583 (3)/Nephronophthisis 1, juvenile, 256100 (3)/Senior-Loken syndrome-1, 266900 (3)
NPHP3	Meckel syndrome 7, 267010 (3)/Nephronophthisis 3, 604387 (3)/Renal-hepatic-pancreatic dysplasia 1, 208540 (3)
NPHP4	Nephronophthisis 4, 606966 (3)/Senior-Loken syndrome 4, 606996 (3)
NPHS1	Nephrotic syndrome, type 1, 256300 (3)
NPHS2	Nephrotic syndrome, type 2 600995 (3)
NPM1	Leukemia, acute myeloid, 601626 (3)/Leukemia, acute promyelocytic, NPM/RARA type (3)
NPPA	Atrial fibrillation, familial, 6, 612201 (3)/Atrial standstill 2, 615745 (3)
NPR2	Acromesomelic dysplasia, Maroteaux type, 602875 (3)/Epiphyseal chondrodysplasia, Miura type, 615923 (3)
NR0B1	Adrenal hypoplasia & hypogonadotrophic hypogonadism
NR0B2	Obesity, mild, early-onset, 601665 (3)
NR2E3	Enhanced S-cone syndrome, 268100 (3)/Retinitis pigmentosa 37, 611131 (3)
NR3C1	Glucocorticoid resistance, 615962 (3)
NR3C2	Hypertension, early-onset, autosomal dominant, with exacerbation in pregnancy, 605115 (3)/Pseudohypoaldosteronism type I, autosomal dominant, 177735 (3)
NR4A3	Chondrosarcoma, extraskeletal myxoid 612237 (3)
NR5A1	46XY sex reversal 3, 612965 (3)/Adrenocortical insufficiency (3)/Premature ovarian failure 7, 612964 (3)/Spermatogenic failure 8, 613957 (3)
NRAS	Autoimmune lymphoproliferative syndrome type IV, 614470 (3)/Colorectal cancer, somatic, 114500 (3)/Epidermal nevus, somatic, 162900 (3)/Melanocytic nevus syndrome, congenital, somatic, 137550 (3)/Neurocutaneous melanosis, somatic, 249400 (3)/Noonan syndrome 6, 613224 (3)/Schimmelpenning-Feuerstein-Mims syndrome, somatic mosaic, 163200 (3)/Thyroid carcinoma, follicular, somatic, 188470 (3)
NRL	Retinal degeneration, autosomal recessive, clumped pigment type (3)/Retinitis pigmentosa 27, 613750 (3)
NRTN	Aganglionosis, in Hirschsprung disease
NRXN1	Pitt-Hopkins-like syndrome 2, 614325 (3)/Schizophrenia, susceptibility to, 17, 614332 (3)
NSD1	Beckwith-Wiedemann syndrome, 130650 (3)/Leukemia, acute myeloid, 601626 (1)/Sotos syndrome 1, 117550 (3)
NSDHL	CHILD syndrome, 308050 (3)/CK syndrome, 300831 (3)
NSUN2	Mental retardation, autosomal recessive 5, 611091 (3)
NT5C3	Haemolytic anaemia
NT5C3A	Anemia, hemolytic, due to UMPH1 deficiency 266120 (3)
NT5E	Calcification of joints and arteries, 211800 (3)
NTF4	Glaucoma 1, open angle, 1O, 613100 (3)
NTNG1	Rett syndrome
NTRK1	Insensitivity to pain, congenital, with anhidrosis, 256800 (3)/Medullary thyroid carcinoma, familial, 155240 (3)
NTRK2	Obesity, hyperphagia, and developmental delay, 613886 (3)
NUBPL	Mitochondrial complex I deficiency, 252010 (3)
NUP214	Leukemia, T-cell acute lymphoblastic (3)/Leukemia, acute myeloid, 601626 (3)
NUP62	Striatonigral degeneration, infantile, 271930 (3)
NYX	Night blindness, congenital stationary (complete), 1A, X-linked, 310500 (3)
OAT	Gyrate atrophy of choroid and retina with or without ornithinemia, 258870 (3)
OBSCN	Glioblastoma
OBSL1	3-M syndrome 2, 612921 (3)

OCA2	Albinism, brown oculocutaneous, 203200 (3)/Albinism, oculocutaneous, type II, 203200 (3)/[Skin/hair/eye pigmentation 1, blond/brown hair], 227220 (3)/[Skin/hair/eye pigmentation 1, blue/nonblue eyes], 227220 (3)
OCLN	Band-like calcification with simplified gyration and polymicrogyria, 251290 (3)
OCRL	Dent disease 2, 300555 (3)/Lowe syndrome, 309000 (3)
OFD1	Retinitis pigmentosa 23, 300424 (3)/Joubert syndrome 10, 300804 (3)/Orofaciodigital syndrome I, 311200 (3)/Simpson-Golabi-Behmel syndrome, type 2, 300209 (3)
OGDH	Alpha-ketoglutarate dehydrogenase deficiency, 203740 (1)
OGG1	Renal cell carcinoma, clear cell, somatic, 144700 (3)
OPA1	Optic atrophy 1, 165500 (3)/Optic atrophy plus syndrome, 125250 (3)/Glaucoma, normal tension, susceptibility to, 606657 (3)
OPA3	3-methylglutaconic aciduria, type III, 258501 (3)/Optic atrophy 3 with cataract, 165300 (3)
OPHN1	Mental retardation, X-linked, with cerebellar hypoplasia and distinctive facial appearance, 300486 (3)
OPLAH	5-oxoprolinase deficiency, 260005 (3)
OPN1LW	Blue cone monochromacy, 303700 (3)/Colorblindness, protan, 303900 (3)
OPN1MW	Blue cone monochromacy, 303700 (3)/Colorblindness, deutan, 303800 (3)
OPN1SW	Colorblindness, tritan, 190900 (3)
OPTN	Amyotrophic lateral sclerosis 12, 613435 (3)/Glaucoma 1, open angle, E, 137760 (3)/Glaucoma, normal tension, susceptibility to, 606657 (3)
ORAI1	Immunodeficiency 9, 612782 (3)/
ORC1	Meier-Gorlin syndrome 1, 224690 (3)
ORC4	Meier-Gorlin syndrome 2, 613800 (3)
ORC6	Meier-Gorlin syndrome 3, 613803 (3)
OSMR	Amyloidosis, primary localized cutaneous, 1, 105250 (3)
OSTM1	Osteopetrosis, autosomal recessive 5, 259720 (3)
OTC	Ornithine transcarbamylase deficiency, 311250 (3)
OTOA	Deafness, autosomal recessive 22, 607039 (3)
OTOF	Auditory neuropathy, autosomal recessive, 1, 601071 (3)/Deafness, autosomal recessive 9, 601071 (3)
OTOG	Deafness, autosomal recessive 18B, 614945 (3)
OTOGL	Deafness, autosomal recessive 84B, 614944 (3)
OTX2	Microphthalmia, syndromic 5, 610125 (3)/Pituitary hormone deficiency, combined, 6, 613986 (3)/Retinal dystrophy, early-onset, and pituitary dysfunction, 610125 (3)
OXCT1	Succinyl CoA:3-oxoacid CoA transferase deficiency, 245050 (3)
P2RY12	Bleeding disorder, platelet-type, 8, 609821 (3)
P4HB	Cole-Carpenter syndrome 1 112240 (3)
PABPN1	Oculopharyngeal muscular dystrophy, 164300 (3)
PAFAH1B1	Lissencephaly 1, 607432 (3)/Subcortical laminar heterotopia, 607432 (3)
PAH	Phenylketonuria, 261600 (3)/[Hyperphenylalaninemia, non-PKU mild], 261600 (3)
PAK3	Mental retardation, X-linked 30/47, 300558 (3)
PALB2	Fanconi anemia, complementation group N, 610832 (3)/Breast cancer, susceptibility to, 114480 (3)/Pancreatic cancer, susceptibility to, 3, 613348 (3)
PANK2	HARP syndrome, 607236 (3)/Neurodegeneration with brain iron accumulation 1, 234200 (3)
PAPSS2	Brachyolmia 4 with mild epiphyseal and metaphyseal changes, 612847 (3)
PARK2	Parkinson disease, early-onset
PARK7	Parkinson disease, autosomal recessive
PARP1	Intellectual disability
PAX2	Glomerulosclerosis, focal segmental, 7, 616002 (3)/Papillorenal syndrome, 120330 (3)/Renal hypoplasia, isolated, 191830 (3)
PAX3	Craniofacial-deafness-hand syndrome, 122880 (3)/Rhabdomyosarcoma2, alveolar, 268220 (3)/Waardenburg syndrome, type 1, 193500 (3)/Waardenburg syndrome, type 3, 148820 (3)
PAX4	Diabetes mellitus, ketosis-prone, 612227 (3)/Diabetes mellitus, type 2, 125853 (3)/Maturity-onset diabetes of the young, type IX, 612225 (3)

PAX6	Morning glory disc anomaly, 120430 (3)/Aniridia, 106210 (3)/Cataract with late-onset corneal dystrophy, 106210 (3)/Coloboma of optic nerve, 120430 (3)/Coloboma, ocular, 120200 (3)/Foveal hypoplasia 1, 136520 (3)/Gillespie syndrome, 206700 (3)/Keratitis, 148190 (3)/Optic nerve hypoplasia, 165550 (3)/Peters anomaly, 604229 (3)
PAX7	Rhabdomyosarcoma 2, alveolar, 268220 (3)
PAX8	Hypothyroidism, congenital, due to thyroid dysgenesis or hypoplasia, 218700 (3)
PAX8-AS1	Hypothyroidism, congenital, due to thyroid dysgenesis or hypoplasia 218700 (3)
PAX9	Tooth agenesis, selective, 3, 604625 (3)
PBX1	Leukemia, acute pre-B-cell (2)
PC	Pyruvate carboxylase deficiency, 266150 (3)
PCBD1	Hyperphenylalaninemia, BH4-deficient, D, 264070 (3)
PCCA	Propionicacidemia, 606054 (3)
PCCB	Propionicacidemia, 606054 (3)
PCDH15	Deafness, autosomal recessive 23, 609533 (3)/Usher syndrome, type 1D/F digenic, 601067 (3)/Usher syndrome, type 1F, 602083 (3)
PCDH19	Epileptic encephalopathy, early infantile, 9, 300088 (3)
PCK1	Phosphoenolpyruvate carboxykinase-1, cytosolic, deficiency, 261680 (1)
PCK2	PEPCK deficiency, mitochondrial, 261650 (1)
PCM1	Thyroid carcinoma, papillary, 188550 (3)
PCNT	Microcephalic osteodysplastic primordial dwarfism, type II, 210720 (3)
PCSK1	Obesity with impaired prohormone processing, 600955 (3)/Obesity, susceptibility to, BMIQ12, 612362 (3)
PCSK9	Hypercholesterolemia, familial, 3, 603776 (3)/Low density lipoprotein cholesterol level QTL 1, 603776 (3)
PDCD10	Cerebral cavernous malformations 3, 603285 (3)
PDE11A	Pigmented nodular adrenocortical disease, primary, 2, 610475 (3)
PDE4D	Acrodysostosis 2, with or without hormone resistance, 614613 (3)/Stroke, susceptibility to, 1, 606799 (3)
PDE6A	Retinitis pigmentosa 43, 613810 (3)
PDE6B	Night blindness, congenital stationary, autosomal dominant 2, 163500 (3)/Retinitis pigmentosa-40, 613801 (3)
PDE6C	Cone dystrophy 4, 613093 (3)
PDE6G	Retinitis pigmentosa 57, 613582 (3)
PDE6H	Achromatopsia 6, 610024 (3)/Retinal cone dystrophy 3, 610024 (3)
PDE8B	Pigmented nodular adrenocortical disease, primary, 3, 614190 (3)/Striatal degeneration, autosomal dominant, 609161 (3)
PDGFB	Basal ganglia calcification, idiopathic, 5, 615483 (3)/Dermatofibrosarcoma protuberans, 607907 (3)/Meningioma, SIS-related, 607174 (3)
PDGFRA	Gastrointestinal stromal tumor, somatic, 606764 (3)/Hypereosinophilic syndrome, idiopathic, resistant to imatinib, 607685 (3)
PDGFRB	Basal ganglia calcification, idiopathic, 4, 615007 (3)/Myeloproliferative disorder with eosinophilia, 131440 (4)/Myofibromatosis, infantile, 1, 228550 (3)
PDGFRL	Colorectal cancer, somatic, 114500 (3)/Hepatocellular cancer, somatic, 114550 (3)
PDHA1	Pyruvate dehydrogenase E1-alpha deficiency, 312170 (3)
PDHB	Pyruvate dehydrogenase E1-beta deficiency, 614111 (3)
PDHX	Pyruvate dehydrogenase complex deficiency
PDLIM3	Cardiomyopathy, dilated
PDP1	Pyruvate dehydrogenase phosphatase deficiency, 608782 (3)
PDSS1	Coenzyme Q10 deficiency, primary, 2, 614651 (3)
PDSS2	Coenzyme Q10 deficiency, primary, 3, 614652 (3)
PDX1	Lacticacidemia due to PDX1 deficiency, 245349 (3)
PDYN	Spinocerebellar ataxia 23, 610245 (3)
PDZD7	Usher syndrome, type IIC, GPR98/PDZD7 digenic, 605472 (3)/Retinal disease in Usher syndrome type IIA, modifier of, 276901 (3)
PECR	Intellectual disability
PEPD	Prolidase deficiency, 170100 (3)
PER2	Advanced sleep phase syndrome, familial, 1, 604348 (3)

PEX1	Peroxisome biogenesis disorder 1A (Zellweger), 214100 (3)/Peroxisome biogenesis disorder 1B (NALD/IRD), 601539 (3)
PEX10	Peroxisome biogenesis disorder 6A (Zellweger), 614870 (3)/Peroxisome biogenesis disorder 6B, 614871 (3)
PEX11B	Peroxisome biogenesis disorder 14B, 614920 (3)
PEX12	Peroxisome biogenesis disorder 3A (Zellweger), 614859 (3)/Peroxisome biogenesis disorder 3B, 266510 (3)
PEX13	Peroxisome biogenesis disorder 11A (Zellweger), 614883 (3)/Peroxisome biogenesis disorder 11B, 614885 (3)
PEX14	Peroxisome biogenesis disorder 13A (Zellweger), 614887 (3)
PEX16	Peroxisome biogenesis disorder 8A, (Zellweger), 614876 (3)/Peroxisome biogenesis disorder 8B, 614877 (3)
PEX19	Peroxisome biogenesis disorder 12A (Zellweger), 614886 (3)
PEX2	Peroxisome biogenesis disorder 5A (Zellweger), 614866 (3)/Peroxisome biogenesis disorder 5B, 614867 (3)
PEX26	Peroxisome biogenesis disorder 7A (Zellweger), 614872 (3)/Peroxisome biogenesis disorder 7B, 614873 (3)
PEX3	Peroxisome biogenesis disorder 10A (Zellweger), 614882 (3)
PEX5	Peroxisome biogenesis disorder 2A (Zellweger), 214110 (3)/Peroxisome biogenesis disorder 2B, 202370 (3)
PEX6	Peroxisome biogenesis disorder 4A (Zellweger), 614862 (3)/Peroxisome biogenesis disorder 4B, 614863 (3)
PEX7	Peroxisome biogenesis disorder 9B, 614879 (3)/Rhizomelic chondrodysplasia punctata, type 1, 215100 (3)
PFKM	Glycogen storage disease VII, 232800 (3)
PFN1	Amyotrophic lateral sclerosis 18, 614808 (3)
PGAM2	Glycogen storage disease X, 261670 (3)
PGK1	Phosphoglycerate kinase 1 deficiency, 300653 (3)
PGM1	Congenital disorder of glycosylation, type It, 614921 (3)
PGR	Progesterone resistance, 264080 (2)
PHB	Breast cancer, susceptibility to, 114480 (3)
PHEX	Hypophosphatemic rickets, X-linked dominant, 307800 (3)
PHF6	Borjeson-Forssman-Lehmann syndrome, 301900 (3)
PHF8	Mental retardation syndrome, X-linked, Siderius type, 300263 (3)
PHGDH	Neu-Laxova syndrome 1 256520 (3)/Phosphoglycerate dehydrogenase deficiency 601815 (3)
PHIP	Glaucoma, primary congenital
PHKA1	Muscle glycogenosis, 300559 (3)
PHKA2	Glycogen storage disease, type IXa1, 306000 (3)/Glycogen storage disease, type IXa2, 306000 (3)
PHKB	Phosphorylase kinase deficiency of liver and muscle, autosomal recessive, 261750 (3)
PHKG2	Cirrhosis due to liver phosphorylase kinase deficiency (3)/Glycogen storage disease IXc, 613027 (3)
PHOX2A	Fibrosis of extraocular muscles, congenital, 2, 602078 (3)
PHOX2B	Central hypoventilation syndrome, congenital, with or without Hirschsprung disease (3)
PHYH	Refsum disease, 266500 (3)
PICALM	Leukemia, acute T-cell lymphoblastic (3)/Leukemia, acute myeloid, 601626 (3)
PIGA	Multiple congenital anomalies-hypotonia-seizures syndrome 2, 300868 (3)/Paroxysmal nocturnal hemoglobinuria, somatic, 300818 (3)
PIGL	CHIME syndrome, 280000 (3)
PIGM	Glycosylphosphatidylinositol deficiency, 610293 (3)
PIGN	Multiple congenital anomalies-hypotonia-seizures syndrome 1, 614080 (3)
PIGO	Hyperphosphatasia with mental retardation syndrome 2, 614749 (3)
PIGV	Hyperphosphatasia with mental retardation syndrome 1, 239300 (3)
PIK3CA	Breast cancer, somatic, 114480 (3)/CLOVE syndrome, somatic, 612918 (3)/Colorectal cancer, somatic, 114500 (3)/Cowden syndrome 5, 615108 (3)/Gastric cancer, somatic, 613659 (3)/Hepatocellular carcinoma, somatic, 114550 (3)/Keratosis, seborrheic, somatic, 182000 (3)/Megalencephaly-capillary malformation-polymicrogyria syndrome, somatic, 602501 (3)/Nevus, epidermal, somatic, 162900 (3)/Nonsmall cell lung cancer, somatic, 211980 (3)/Ovarian cancer, somatic, 167000 (3)
PIK3R2	Megalencephaly-polymicrogyria-polydactyly-hydrocephalus syndrome 1, 603387 (3)
PIKFYVE	Corneal fleck dystrophy, 121850 (3)
PINK1	Parkinson disease 6, early onset, 605909 (3)
PIP5K1C	Lethal congenital contractural syndrome 3, 611369 (3)
PITPNM3	Cone-rod dystrophy 5, 600977 (3)
PITX1	Clubfoot, congenital, with or without deficiency of long bones and/or mirror-image polydactyly, 119800 (3)/Liebenberg syndrome, 186550 (4)

PITX2	Axenfeld-Rieger syndrome, type 1, 180500 (3)/Iridogoniodysgenesis, type 2, 137600 (3)/Peters anomaly, 604229 (3)/Ring dermoid of cornea, 180550 (3)
PITX3	Anterior segment mesenchymal dysgenesis, 107250 (3)/Cataract 11, multiple types, 610623 (3)/Cataract 11, syndromic, 610623 (3)
PKD1	Polycystic kidney disease, adult type I, 173900 (3)
PKD2	Polycystic kidney disease 2, 613095 (3)
PKHD1	Polycystic kidney disease
PKLR	Adenosine triphosphate, elevated, of erythrocytes, 102900 (3)/Pyruvate kinase deficiency, 266200 (3)
PKP1	Ectodermal dysplasia/skin fragility syndrome, 604536 (3)
PKP2	Arrhythmogenic right ventricular dysplasia 9, 609040 (3)
PLA2G5	[Fleck retina, familial benign], 228980 (3)
PLA2G6	Infantile neuroaxonal dystrophy 1, 256600 (3)/Neurodegeneration with brain iron accumulation 2B, 610217 (3)/Parkinson disease 14, 612953 (3)
PLA2G7	Platelet-activating factor acetylhydrolase deficiency, 614278 (3)/Asthma, susceptibility to, 600807 (3)/Atopy, susceptibility to, 147050 (3)
PLAG1	Adenomas, salivary gland pleomorphic, 181030 (3)
PLAT	Hyperfibrinolysis, familial, due to increased release of PLAT, 612348 (1)/Thrombophilia, familial, due to decreased release of PLAT, 612348 (1)
PLAU	Quebec platelet disorder, 601709 (3)/Alzheimer disease, late-onset, susceptibility to, 104300 (3)
PLCB1	Epileptic encephalopathy, early infantile, 12, 613722 (3)
PLCB4	Auriculocondylar syndrome 2, 614669 (3)
PLCD1	Nail disorder, nonsyndromic congenital, 3, (leukonychia), 151600 (3)
PLCE1	Nephrotic syndrome, type 3, 610725 (3)
PLCG2	Autoinflammation, antibody deficiency, and immune dysregulation syndrome, 614878 (3)/Familial cold autoinflammatory syndrome 3, 614468 (3)
PLEC	Muscular dystrophy with epidermolysis bullosa
PLEKHG4	Cerebellar ataxia, autosomal dominant
PLEKHG5	Charcot-Marie-Tooth disease, recessive intermediate C, 615376 (3)/Spinal muscular atrophy, distal, autosomal recessive, 4, 611067 (3)
PLEKHM1	Osteopetrosis, autosomal recessive 6, 611497 (3)
PLG	Dysplasminogenemia, 217090 (3)/Plasminogen deficiency, type I, 217090 (3)
PLIN1	Lipodystrophy, familial partial, type 4, 613877 (3)
PLN	Cardiomyopathy, dilated, 1P, 609909 (3)/Cardiomyopathy, familial hypertrophic, 18, 613874 (3)
PLOD1	Ehlers-Danlos syndrome, type VI, 225400 (3)
PLOD2	Bruck syndrome 2, 609220 (3)
PLOD3	Lysyl hydroxylase 3 deficiency, 612394 (3)
PLP1	Pelizaeus-Merzbacher disease, 312080 (3)/Spastic paraplegia 2, X-linked, 312920 (3)
PMM2	Congenital disorder of glycosylation, type Ia, 212065 (3)
PMP22	Charcot-Marie-Tooth disease, type 1A, 118220 (3)/Charcot-Marie-Tooth disease, type 1E, 118300 (3)/Dejerine-Sottas disease, 145900 (3)/Neuropathy, inflammatory demyelinating, 139393 (3)/Neuropathy, recurrent, with pressure palsies, 162500 (3)/Roussy-Levy syndrome, 180800 (3)
PMS2	Colorectal cancer, hereditary nonpolyposis, type 4, 614337 (3)/Mismatch repair cancer syndrome, 276300 (3)
PNKD	Paroxysmal nonkinesiogenic dyskinesia
PNKP	Microcephaly, seizures, and developmental delay, 613402 (3)
PNLIP	Pancreatic lipase deficiency, 614338 (1)
PNP	Immunodeficiency due to purine nucleoside phosphorylase deficiency, 613179 (3)
PNPLA1	Ichthyosis, congenital, autosomal recessive 10, 615024 (3)
PNPLA2	Neutral lipid storage disease with myopathy, 610717 (3)
PNPLA6	Boucher-Neuhauser syndrome, 215470 (3)/Spastic paraplegia 39, autosomal recessive, 612020 (3)
PNPO	Pyridoxamine 5'-phosphate oxidase deficiency, 610090 (3)
PNPT1	Combined oxidative phosphorylation deficiency 13, 614932 (3)/Deafness, autosomal recessive 70, 614934 (3)
POC1A	Short stature, onychodysplasia, facial dysmorphism, and hypotrichosis, 614813 (3)
POF1B	?Premature ovarian failure 2B 300604 (3)
POLA1	N syndrome, 310465 (1)

POLG	Mitochondrial DNA depletion syndrome 4A (Alpers type), 203700 (3)/Mitochondrial DNA depletion syndrome 4B (MNGIE type), 613662 (3)/Mitochondrial recessive ataxia syndrome (includes SANDO and SCAE), 607459 (3)/Progressive external ophthalmoplegia, autosomal dominant, 157640 (3)/Progressive external ophthalmoplegia, autosomal recessive, 258450 (3)
POLG2	Progressive external ophthalmoplegia with mitochondrial DNA deletions, autosomal dominant 4, 610131 (3)
POLH	Xeroderma pigmentosum, variant type, 278750 (3)
POLR1C	Treacher Collins syndrome 3, 248390 (3)
POLR1D	Treacher Collins syndrome 2, 613717 (3)
POLR3A	Leukodystrophy, hypomyelinating,7, with or without oligodontia and/or hypogonadotropic hypogonadism, 607694 (3)
POLR3B	Leukodystrophy, hypomyelinating,8, with or without oligodontia and/or hypogonadotropic hypogonadism, 614381 (3)
POMC	Obesity, adrenal insufficiency, and red hair due to POMC deficiency, 609734 (3)/Obesity, early-onset, susceptibility to, 601665 (3)
POMGNT1	Muscular dystrophy-dystroglycanopathy (congenital with brain and eye anomalies), type A, 3, 253280 (3)/Muscular dystrophy-dystroglycanopathy (congenital with mental retardation), type B, 3, 613151 (3)/Muscular dystrophy-dystroglycanopathy (limb-girdle), type C, 3, 613157 (3)
POMGNT2	Muscular dystrophy-dystroglycanopathy (congenital with brain and eye anomalies, type A, 8 614830 (3)
POMP	Keratosis linearis with ichthyosis congenita and sclerosing keratoderma, 601952 (3)
POMT1	Muscular dystrophy-dystroglycanopathy (congenital with brain and eye anomalies), type A, 1, 236670 (3)/Muscular dystrophy-dystroglycanopathy (congenital with mental retardation), type B, 1, 613155 (3)/Muscular dystrophy-dystroglycanopathy (limb-girdle), type C, 1, 609308 (3)
POMT2	Muscular dystrophy-dystroglycanopathy (congenital with brain and eye anomalies), type A, 2, 613150 (3)/Muscular dystrophy-dystroglycanopathy (congenital with mental retardation), type B, 2, 613156 (3)/Muscular dystrophy-dystroglycanopathy (limb-girdle), type C, 2, 613158 (3)
PON1	Coronary artery disease, susceptibility to (3)/Coronary artery spasm 2, susceptibility to (3)/Microvascular complications of diabetes 5, 612633 (3)/Organophosphate poisoning, sensitivity to (3)
POR	Antley-Bixler syndrome with genital anomalies and disordered steroidogenesis, 201750 (3)/Disordered steroidogenesis due to cytochrome P450 oxidoreductase, 613571 (3)
PORCN	Focal dermal hypoplasia, 305600 (3)
POU1F1	Pituitary hormone deficiency, combined, 1, 613038 (3)
POU3F4	Deafness, X-linked 2, 304400 (3)
POU4F3	Deafness, autosomal dominant 15, 602459 (3)
POU6F2	Wilms tumor susceptibility-5, 601583 (3)
PPARG	Carotid intimal medial thickness 1, 609338 (3)/Insulin resistance, severe, digenic, 604367 (3)/Lipodystrophy, familial partial, type 3, 604367 (3)/Obesity, severe, 601665 (3)/[Obesity, resistance to] (3)/Diabetes, type 2, 125853 (3)
PPIB	Osteogenesis imperfecta, type IX, 259440 (3)
PPM1D	Breast cancer, 114480 (3)
PPOX	Porphyria variegata, 176200 (3)
PPP1R3A	Insulin resistance, severe, digenic, 604367 (3)
PPP2R1B	Lung cancer, 211980 (3)
PPP2R2B	Spinocerebellar ataxia 12, 604326 (3)
PPP2R5D	Mental retardation, autosomal dominant 35 616355 (3)
PPT1	Ceroid lipofuscinosis, neuronal, 1, 256730 (3)
PQBP1	Renpenning syndrome, 309500 (3)
PRCC	Renal cell carcinoma, papillary, 605074 (3)
PRCD	Retinitis pigmentosa 36 610599 (3)
PRDM5	Brittle cornea syndrome 2, 614170 (3)
PRF1	Hemophagocytic lymphohistiocytosis, familial, 2, 603553 (3)/Lymphoma, non-Hodgkin, 605027 (3)
PRG4	Camptodactyly-arthropathy-coxa vara-pericarditis syndrome, 208250 (3)
PRICKLE1	Epilepsy, progressive myoclonic 1B, 612437 (3)
PRICKLE2	Epilepsy, progressive myoclonic 5, 613832 (3)

PRKAG2	Cardiomyopathy, familial hypertrophic 6, 600858 (3)/Glycogen storage disease of heart, lethal congenital, 261740 (3)/Wolff-Parkinson-White syndrome, 194200 (3)
PRKAR1A	Acrodysostosis 1, with or without hormone resistance, 101800 (3)/Adrenocortical tumor, somatic, (3)/Carney complex, type 1, 160980 (3)/Myxoma, intracardiac, 255960 (3)/Pigmented nodular adrenocortical disease, primary, 1, 610489 (3)/Thyroid carcinoma, papillary, somatic, 188550 (3)
PRKCA	Pituitary tumor, invasive (3)
PRKCG	Spinocerebellar ataxia 14, 605361 (3)
PRKCSH	Polycystic liver disease, 174050 (3)
PRKDC	Immunodeficiency 16, with or without neurologic abnormalities, 615966 (3)
PRKRA	Dystonia 16, 612067 (3)
PRMT10	Intellectual disability
PRNP	Cerebral amyloid angiopathy, PRNP-related 137440 (3)
PROC	Thrombophilia due to protein C deficiency, autosomal dominant, 176860 (3)/Thrombophilia due to protein C deficiency, autosomal recessive, 612304 (3)
PRODH	Hyperprolinemia, type I, 239500 (3)/Schizophrenia, susceptibility to, 4, 600850 (3)
PROK2	Hypogonadotropic hypogonadism 4 with or without anosmia, 610628 (3)
PROKR2	Hypogonadotropic hypogonadism 3 with or without anosmia, 244200 (3)
PROM1	Cone-rod dystrophy 12, 612657 (3)/Macular dystrophy, retinal, 2, 608051 (3)/Retinitis pigmentosa 41, 612095 (3)/Stargardt disease 4, 603786 (3)
PROP1	Pituitary hormone deficiency, combined, 2, 262600 (3)
PROS1	Thrombophilia due to protein S deficiency, autosomal dominant, 612336 (3)/Thrombophilia due to protein S deficiency, autosomal recessive, 614514 (3)
PRPF3	Retinitis pigmentosa
PRPF31	Retinitis pigmentosa 11, 600138 (3)
PRPF6	Retinitis pigmentosa 60, 613983 (3)
PRPF8	Retinitis pigmentosa 13, 600059 (3)
PRPH2	Choriodal dystrophy, central areolar 2, 613105 (3)/Leber congenital amaurosis 18, 608133 (3)/Macular dystrophy, patterned, 1, 169150 (3)/Macular dystrophy, vitelliform, 3, 608161 (3)/Retinitis pigmentosa 7 and digenic, 608133 (3)/Retinitis punctata albescens, 136880 (3)
PRPS1	Arts syndrome, 301835 (3)/Charcot-Marie-Tooth disease, X-linked recessive, 5, 311070 (3)/Deafness, X-linked 1, 304500 (3)/Gout, PRPS-related, 300661 (3)/Phosphoribosylpyrophosphate synthetase superactivity, 300661 (3)
PRRT2	Convulsions, familial infantile, with paroxysmal choreoathetosis, 602066 (3)/Episodic kinesigenic dyskinesia 1, 128200 (3)/Seizures, benign familial infantile, 2, 605751 (3)
PRRX1	Agnathia-otocephaly complex, 202650 (3)
PRSS1	Pancreatitis, hereditary, 167800 (3)/Trypsinogen deficiency, 614044 (1)
PRSS12	Mental retardation, autosomal recessive 1, 249500 (3)
PRSS56	Microphthalmia, isolated 6, 613517 (3)
PRX	Charcot-Marie-Tooth disease, type 4F, 614895 (3)/Dejerine-Sottas disease, 145900 (3)
PSAP	Combined SAP deficiency, 611721 (3)/Gaucher disease, atypical, 610539 (3)/Krabbe disease, atypical, 611722 (3)/Metachromatic leukodystrophy due to SAP-b deficiency, 249900 (3)
PSAT1	Phosphoserine aminotransferase deficiency, 610992 (3)/Neu-Laxova syndrome 2, 616038 (3)
PSEN1	Acne inversa, familial, 3, 613737 (3)/Alzheimer disease, type 3, 607822 (3)/Alzheimer disease, type 3, with spastic paraparesis and apraxia, 607822 (3)/Alzheimer disease, type 3, with spastic paraparesis and unusual plaques, 607822 (3)/Cardiomyopathy, dilated, 1U, 613694 (3)/Dementia, frontotemporal, 600274 (3)/Pick disease, 172700 (3)
PSEN2	Alzheimer disease-4, 606889 (3)/Cardiomyopathy, dilated, 1V, 613697 (3)
PSENEN	Acne inversa, familial, 2, 613736 (3)
PSMB8	Autoinflammation, lipodystrophy, and dermatosis syndrome, 256040 (3)
PSMC3IP	Ovarian dysgenesis 3, 614324 (3)
PSPH	Phosphoserine phosphatase deficiency, 614023 (3)
PSTPIP1	Pyogenic sterile arthritis, pyoderma gangrenosum, and acne, 604416 (3)
PTCH1	Basal cell carcinoma, somatic, 605462 (3)/Basal cell nevus syndrome, 109400 (3)/Holoprosencephaly-7, 610828 (3)

PTCH2	Basal cell carcinoma, somatic, 605462 (3)/Basal cell nevus syndrome, 109400 (3)/Medulloblastoma, 155255 (3)
PTCHD1	Intellectual disability, X-linked
PTEN	Bannayan-Riley-Ruvalcaba syndrome, 153480 (3)/Cowden syndrome 1, 158350 (3)/Endometrial carcinoma, somatic, 608089 (3)/Lhermitte-Duclos syndrome, 158350 (3)/Macrocephaly/autism syndrome, 605309 (3)/Malignant melanoma, somatic, 155600 (3)/PTEN hamartoma tumor syndrome (3)/Squamous cell carcinoma, head and neck, somatic, 275355 (3)/Thyroid carcinoma, follicular, somatic, 188470 (3)/VATER association with macrocephaly and ventriculomegaly, 276950 (3)/Glioma susceptibility 2, 613028 (3)/Meningioma, 607174 (3)/Prostate cancer, somatic, 176807 (3)
PTF1A	Pancreatic agenesis 2, 615935 (3)/Pancreatic and cerebellar agenesis, 609069 (3)
PTGIS	Hypertension, essential, 145500 (3)
PTH	Hypoparathyroidism, autosomal dominant, 146200 (3)/Hypoparathyroidism, autosomal recessive, 146200 (3)
PTH1R	Osteochondrodysplasia, Blomstrand, type 1
PTHLH	Brachydactyly, type E2, 613382 (3)/Humoral hypercalcemia of malignancy (1)
PTPN11	LEOPARD syndrome 1, 151100 (3)/Leukemia, juvenile myelomonocytic, 607785 (3)/Metachondromatosis, 156250 (3)/Noonan syndrome 1, 163950 (3)
PTPN14	Choanal atresia and lymphedema, 613611 (3)
PTPRC	Severe combined immunodeficiency, T cell-negative, B-cell/natural killer-cell positive, 608971 (3)/Hepatitic C virus, susceptibility to, 609532 (3)
PTPRJ	Colon cancer, somatic, 114500 (3)
PTPRO	Nephrotic syndrome, type 6, 614196 (3)
PTPRQ	Deafness, autosomal recessive 84A, 613391 (3)
PTRF	Lipodystrophy, congenital generalized, type 4, 613327 (3)
PTS	Hyperphenylalaninemia, BH4-deficient, A, 261640 (3)
PUS1	Mitochondrial myopathy and sideroblastic anemia 1, 600462 (3)
PVRL1	Cleft lip / palate
PVRL4	Ectodermal dysplasia-syndactyly syndrome 1, 613573 (3)
PYCR1	Cutis laxa, autosomal recessive, type IIB, 612940 (3)/Cutis laxa, autosomal recessive, type IIIB, 614438 (3)
PYGL	Glycogen storage disease VI, 232700 (3)
PYGM	McArdle disease, 232600 (3)
QDPR	Hyperphenylalaninemia, BH4-deficient, C, 261630 (3)
RAB18	Warburg micro syndrome 3, 614222 (3)
RAB23	Carpenter syndrome, 201000 (3)
RAB27A	Griscelli syndrome, type 2, 607624 (3)
RAB39B	Waisman syndrome, 311510 (3)/Mental retardation, X-linked 72, 300271 (3)
RAB3GAP1	Warburg micro syndrome 1, 600118 (3)
RAB3GAP2	Martsolf syndrome, 212720 (3)/Warburg micro syndrome 2, 614225 (3)
RAB40AL	Martin-Probst syndrome
RAB7A	Charcot-Marie-Tooth disease 2b
RAC2	Neutrophil immunodeficiency syndrome, 608203 (3)
RAD21	Cornelia de Lange syndrome 4, 614701 (3)
RAD50	Nijmegen breakage syndrome-like disorder, 613078 (3)
RAD51	Mirror movements, congenital
RAD51C	Fanconi anemia, complementation group O, 613390 (3)/Breast-ovarian cancer, familial, susceptibility to, 3, 613399 (3)
RAD54L	Adenocarcinoma, colonic, somatic (3)/Lymphoma, non-Hodgkin, somatic, 605027 (3)/Breast cancer, invasive ductal, 114480 (3)
RAF1	Cardiomyopathy, dilated, 1NN, 615916 (3)/LEOPARD syndrome 2, 611554 (3)/Noonan syndrome 5, 611553 (3)
RAG1	Alpha/beta T-cell lymphopenia with gamma/delta T-cell expansion, severe cytomegalovirus infection, and autoimmunity, 609889 (3)/Combined cellular and humoral immune defects with granulomas, 233650 (3)/Omenn syndrome, 603554 (3)/Severe combined immunodeficiency, B cell-negative, 601457 (3)
RAG2	Combined cellular and humoral immune defects with granulomas, 233650 (3)/Omenn syndrome, 603554 (3)/Severe combined immunodeficiency, B cell-negative, 601457 (3)

RAI1	Smith-Magenis syndrome, 182290 (3)
RALGDS	Intellectual disability
RANBP2	Encephalopathy, acute, infection-induced, 3, susceptibility to, 608033 (3)
RANGRF	Brugada syndrome
RAPSN	Fetal akinesia deformation sequence, 208150 (3)/Myasthenic syndrome, congenital, associated with acetylcholine receptor deficiency, 608931 (3)/Myasthenic syndrome, congenital, associated with facial dysmorphism and acetylcholine receptor deficiency, 608931 (3)
RARA	Leukemia, acute promyelocytic, 612376 (1)
RARA-AS1	Leukemia, acute promyelocytic 612376 (1)
RARS2	Pontocerebellar hypoplasia, type 6, 611523 (3)
RASA1	Basal cell carcinoma, somatic, 605462 (3)/Capillary malformation-arteriovenous malformation, 608354 (3)/Parkes Weber syndrome, 608355 (3)
RASSF1	Lung cancer, 211980 (2)
RASSF1-AS1	Lung cancer 211980 (2)
RAX	Microphthalmia, isolated 3, 611038 (3)
RAX2	Cone-rod dystrophy
RB1	Bladder cancer, somatic, 109800 (3)/Osteosarcoma, somatic, 259500 (3)/Retinoblastoma, 180200 (3)/Retinoblastoma, trilateral, 180200 (3)/Small cell cancer of the lung, somatic, 182280 (3)
RB1CC1	Breast cancer, somatic, 114480 (3)
RBBP8	Jawad syndrome, 251255 (3)/Pancreatic carcinoma, somatic (3)/Seckel syndrome 2, 606744 (3)
RBM10	TARP syndrome, 311900 (3)
RBM20	Cardiomyopathy, dilated, 1DD, 613172 (3)
RBM28	Alopecia, neurologic defects, and endocrinopathy syndrome, 612079 (3)
RBM8A	Thrombocytopenia-absent radius syndrome, 274000 (3)
RBP3	Retinitis pigmentosa 66, 615233 (3)
RBP4	Retinol dystrophy, iris coloboma, and comedogenic acne syndrome, 615147 (3)
RBPJ	Adams-Oliver syndrome 3, 614814 (3)
RD3	Leber congenital amaurosis 12 610612 (3)
RDH12	Leber congenital amaurosis 13 612712 (3)
RDH5	Fundus albipunctatus, 136880 (3)
RDX	Deafness, autosomal recessive 24, 611022 (3)
RECQL4	Baller-Gerold syndrome, 218600 (3)/RAPADILINO syndrome, 266280 (3)/Rothmund-Thomson syndrome, 268400 (3)
REEP1	Neuronopathy, distal hereditary motor, type VB, 614751 (3)/Spastic paraplegia 31, autosomal dominant, 610250 (3)
RELN	Lissencephaly 2 (Norman-Roberts type), 257320 (3)
REN	Hyperuricemic nephropathy, familial juvenile 2, 613092 (3)/Renal tubular dysgenesis, 267430 (3)/[Hyperproreninemia] (3)
RET	Central hypoventilation syndrome, congenital, 209880 (3)/Medullary thyroid carcinoma, 155240 (3)/Multiple endocrine neoplasia IIA, 171400 (3)/Multiple endocrine neoplasia IIB, 162300 (3)/Pheochromocytoma, 171300 (3)/Renal agenesis, 191830 (3)/Hirschsprung disease, susceptibility to, 1, 142623 (3)
RFT1	Congenital disorder of glycosylation, type In, 612015 (3)
RFX5	Bare lymphocyte syndrome, type II, complementation group C, 209920 (3)/Bare lymphocyte syndrome, type II, complementation group E, 209920 (3)
RFX6	Mitchell-Riley syndrome, 615710 (3)
RFXANK	MHC class II deficiency, complementation group B, 209920 (3)
RFXAP	Bare lymphocyte syndrome, type II, complementation group D, 209920 (3)
RGR	Retinitis pigmentosa 44, 613769 (3)
RGS7	Intellectual disability
RGS9	Bradyopsia, 608415 (3)
RGS9BP	Bradyopsia, 608415 (3)
RHAG	Anemia, hemolytic, Rh-null, regulator type, 268150 (3)/Rh-mod syndrome (3)
RHBDF2	Tylosis with esophageal cancer, 148500 (3)

RHO	Night blindness, congenital stationary, autosomal dominant 1, 610445 (3)/Retinitis pigmentosa 4, autosomal dominant or recessive, 613731 (3)/Retinitis punctata albescens, 136880 (3)
RIMS1	Cone-rod dystrophy 7, 603649 (3)
RIN2	Macrocephaly, alopecia, cutis laxa, and scoliosis, 613075 (3)
RIPK4	Popliteal pterygium syndrome 2, lethal type, 263650 (3)
RLBP1	Bothnia retinal dystrophy, 607475 (3)/Fundus albipunctatus, 136880 (3)/Newfoundland rod-cone dystrophy, 607476 (3)/Retinitis punctata albescens, 136880 (3)
RMND1	Combined oxidative phosphorylation deficiency 11, 614922 (3)
RNASEH2A	Aicardi-Goutieres syndrome 4, 610333 (3)
RNASEH2B	Aicardi-Goutieres syndrome 2, 610181 (3)
RNASEH2C	Aicardi-Goutieres syndrome 3, 610329 (3)
RNASEL	Prostate cancer 1, 601518 (3)
RNASET2	Leukoencephalopathy, cystic, without megalencephaly, 612951 (3)
RNF135	Macrocephaly, macrosomia, facial dysmorphism syndrome, 614192 (3)
RNF139	Renal cell carcinoma, 144700 (3)
RNF168	RIDDLE syndrome, 611943 (3)
RNF170	Ataxia, sensory, 1, autosomal dominant, 608984 (3)
RNF212	Recombination rate QTL 1, 612042 (3)
RNF213	Moyamoya disease 2, susceptibility to, 607151 (3)
RNF6	Esophageal carcinoma, somatic, 133239 (3)
ROBO2	Vesicoureteral reflux 2, 610878 (3)
ROBO3	Gaze palsy, horizontal, with progressive scoliosis, 607313 (3)
ROGDI	Kohlschutter-Tonz syndrome, 226750 (3)
ROM1	Retinitis pigmentosa 7, digenic, 608133 (3)
ROR2	Brachydactyly, type B1, 113000 (3)/Robinow syndrome, autosomal recessive, 268310 (3)
RP1	Retinitis pigmentosa 1, 180100 (3)
RP1L1	Occult macular dystrophy, 613587 (3)
RP2	Retinitis pigmentosa 2, 312600 (3)
RP9	Retinitis pigmentosa 9, 180104 (3)
RPE65	Leber congenital amaurosis 2, 204100 (3)/Retinitis pigmentosa 20, 613794 (3)
RPGR	Cone-rod dystrophy, X-linked, 1, 304020 (3)/Macular degeneration, X-linked atrophic, 300834 (3)/Retinitis pigmentosa 3, 300029 (3)/Retinitis pigmentosa, X-linked, and sinorespiratory infections, with or without deafness, 300455 (3)
RPGRIP1	Cone-rod dystrophy 13, 608194 (3)/Leber congenital amaurosis 6, 613826 (3)
RPGRIP1L	COACH syndrome, 216360 (3)/Joubert syndrome 7, 611560 (3)/Meckel syndrome 5, 611561 (3)
RPIA	Ribose 5-phosphate isomerase deficiency, 608611 (3)
RPL10	Autism, susceptibility to, X-linked 5, 300847 (3)
RPL11	Diamond-Blackfan anemia 7, 612562 (3)
RPL21	Hypotrichosis 12, 615885 (3)
RPL26	Diamond-Blackfan anemia 11, 614900 (3)
RPL35A	Diamond-Blackfan anemia 5, 612528 (3)
RPL5	Diamond-Blackfan anemia 6, 612561 (3)
RPS10	Diamond-Blackfan anemia 9, 613308 (3)
RPS14	Macrocytic anemia, refractory, due to 5q deletion, somatic, 153550 (3)
RPS17	Diamond-Blackfan anemia 4, 612527 (3)
RPS19	Diamond-Blackfan anemia 1, 105650 (3)
RPS24	Diamond-blackfan anemia 3, 610629 (3)
RPS26	Diamond-Blackfan anemia 10, 613309 (3)
RPS6KA3	Coffin-Lowry syndrome, 303600 (3)/Mental retardation, X-linked 19, 300844 (3)
RPS7	Diamond-Blackfan anemia 8, 612563 (3)
RPSA	Asplenia, isolated congenital, 271400 (3)
RRM2B	Mitochondrial DNA depletion syndrome 8A (encephalomyopathic type with renal tubulopathy), 612075 (3)/Mitochondrial DNA depletion syndrome 8B (MNGIE type), 612075 (3)/Progressive external ophthalmoplegia with mitochondrial DNA deletions, autosomal dominant, 5, 613077 (3)

RS1	Retinoschisis, 312700 (3)
RSPH4A	Ciliary dyskinesia, primary, 11, 612649 (3)
RSPH9	Ciliary dyskinesia, primary, 12, 612650 (3)
RSPO1	Palmoplantar hyperkeratosis and true hermaphroditism, 610644 (3)/Palmoplantar hyperkeratosis with squamous cell carcinoma of skin and sex reversal, 610644 (3)
RSPO4	Anonychia congenita, 206800 (3)
RTN2	Spastic paraplegia 12, autosomal dominant, 604805 (3)
RTTN	Polymicrogyria with seizures, 614833 (3)
RUNX1	Leukemia, acute myeloid, 601626 (3)/Platelet disorder, familial, with associated myeloid malignancy, 601399 (3)
RUNX2	Cleidocranial dysplasia, 119600 (3)/Cleidocranial dysplasia, forme fruste, dental anomalies only, 119600 (3)/Cleidocranial dysplasia, forme fruste, with brachydactyly, 119600 (3)/Metaphyseal dysplasia with maxillary hypoplasia with or without brachydactyly, 156510 (3)
RXFP2	Cryptorchidism/Testicular Diseases/Disorder Of Male Reproductive System
RYR1	Central core disease, 117000 (3)/King-Denborough syndrome, 145600 (3)/Minicore myopathy with external ophthalmoplegia, 255320 (3)/Neuromuscular disease, congenital, with uniform type 1 fiber, 117000 (3)/Malignant hyperthermia susceptibility 1, 145600 (3)
RYR2	Arrhythmogenic right ventricular dysplasia 2, 600996 (3)/Ventricular tachycardia, catecholaminergic polymorphic, 1, 604772 (3)
SACS	Spastic ataxia, Charlevoix-Saguenay type, 270550 (3)
SAG	Oguchi disease-1, 258100 (3)/Retinitis pigmentosa 47, 613758 (3)
SALL1	Townes-Brocks branchiootorenal-like syndrome, 107480 (3)/Townes-Brocks syndrome, 107480 (3)
SALL4	Duane-radial ray syndrome, 607323 (3)/IVIC syndrome, 147750 (3)
SAMD9	Tumoral calcinosis, familial, normophosphatemic, 610455 (3)
SAMHD1	Aicardi-Goutieres syndrome 5, 612952 (3)/Chilblain lupus 2, 614415 (3)
SAR1B	Chylomicron retention disease, 246700 (3)
SARS2	Hyperuricemia, pulmonary hypertension, renal failure, and alkalosis, 613845 (3)
SART3	Disseminated superficial actinic porokeratosis
SAT1	Keratosis follicularis spinulosa decalvans, 308800 (3)
SATB2	Glass syndrome, 612313 (3)
SBDS	Shwachman-Bodian-Diamond syndrome, 260400 (3)
SBF2	Charcot-Marie-Tooth disease, type 4B2, 604563 (3)
SC5D	Lathosterolosis 607330 (3)
SCARB2	Epilepsy, progressive myoclonic 4, with or without renal failure, 254900 (3)
SCARF2	Van den Ende-Gupta syndrome, 600920 (3)
SCN1A	Dravet syndrome, 607208 (3)/Epilepsy, generalized, with febrile seizures plus, type 2, 604403 (3)/Febrile seizures, familial, 3A, 604403 (3)/Migraine, familial hemiplegic, 3, 609634 (3)
SCN1B	Atrial fibrillation, familial, 13, 615377 (3)/Brugada syndrome 5, 612838 (3)/Cardiac conduction defect, nonspecific, 612838 (3)/Epilepsy, generalized, with febrile seizures plus, type 1, 604233 (3)
SCN2A	Epileptic encephalopathy, early infantile, 11, 613721 (3)/Seizures, benign familial infantile, 3, 607745 (3)
SCN2B	Atrial fibrillation, familial, 14, 615378 (3)
SCN3B	Atrial fibrillation, familial, 16, 613120 (3)/Brugada syndrome 7, 613120 (3)
SCN4A	Hyperkalemic periodic paralysis, type 2, 170500 (3)/Hypokalemic periodic paralysis, type 2, 613345 (3)/Myasthenic syndrome, acetazolamide-responsive, 614198 (3)/Myotonia congenita, atypical, acetazolamide-responsive, 608390 (3)/Paramyotonia congenita, 168300 (3)
SCN4B	Atrial fibrillation, familial, 17, 611819 (3)/Long QT syndrome-10, 611819 (3)
SCN5A	Atrial fibrillation, familial, 10, 614022 (3)/Brugada syndrome 1, 601144 (3)/Cardiomyopathy, dilated, 1E, 601154 (3)/Heart block, nonprogressive, 113900 (3)/Heart block, progressive, type IA, 113900 (3)/Long QT syndrome-3, 603830 (3)/Sick sinus syndrome 1, 608567 (3)/Ventricular fibrillation, familial, 1, 603829 (3)/Sudden infant death syndrome, susceptibility to, 272120 (3)
SCN8A	Cognitive impairment with or without cerebellar ataxia, 614306 (3)/Epileptic encephalopathy, early infantile, 13, 614558 (3)

SCN9A	Epilepsy, generalized, with febrile seizures plus, type 7, 613863 (3)/Erythermalgia,primary, 133020 (3)/Febrile seizures, familial, 3B, 613863 (3)/HSAN2D, autosomal recessive, 243000 (3)/Insensitivity to pain, congenital, 243000 (3)/Paroxysmal extreme pain disorder, 167400, (3)/Small fiber neuropathy, 133020 (3)/Dravet syndrome, modifier of, 607208 (3)
SCNN1A	Bronchiectasis with or without elevated sweat chloride 2, 613021 (3)/Pseudohypoaldosteronism, type I, 264350 (3)
SCNN1B	Bronchiectasis with or without elevated sweat chloride 1, 211400 (3)/Liddle syndrome, 177200 (3)/Pseudohypoaldosteronism, type I, 264350 (3)
SCNN1G	Bronchiectasis with or without elevated sweat chloride 3, 613071 (3)/Liddle syndrome, 177200 (3)/Pseudohypoaldosteronism, type I, 264350 (3)
SCO1	Cytochrome c oxidase deficiency
SCO2	Cardioencephalomyopathy, fatal infantile, due to cytochrome c oxidase deficiency 1, 604377 (3)/Myopia 6, 608908 (3)
SCP2	Leukoencephalopathy with dystonia and motor neuropathy, 613724 (3)
SDCCAG8	Bardet-Biedl syndrome 16, 615993 (3)/Senior-Loken syndrome 7, 613615 (3)
SDHA	Cardiomyopathy, dilated, 1GG, 613642 (3)/Leigh syndrome, 256000 (3)/Mitochondrial respiratory chain complex II deficiency, 252011 (3)/Paragangliomas 5, 614165 (3)
SDHAF1	Mitochondrial complex II deficiency, 252011 (3)
SDHAF2	Paragangliomas 2, 601650 (3)
SDHB	Cowden syndrome 2, 612359 (3)/Gastrointestinal stromal tumor, 606764 (3)/Paraganglioma and gastric stromal sarcoma, 606864 (3)/Paragangliomas 4, 115310 (3)/Pheochromocytoma, 171300 (3)
SDHC	Gastrointestinal stromal tumor, 606764 (3)/Paraganglioma and gastric stromal sarcoma, 606864 (3)/Paragangliomas 3, 605373 (3)
SDHD	Carcinoid tumors, intestinal, 114900 (3)/Cowden syndrome 3, 615106 (3)/Merkel cell carcinoma, somatic (3)/Paraganglioma and gastric stromal sarcoma, 606864 (3)/Paragangliomas 1, with or without deafness, 168000 (3)/Pheochromocytoma, 171300 (3)
SEC23A	Craniolenticulosutural dysplasia, 607812 (3)
SEC23B	Dyserythropoietic anemia, congenital, type II, 224100 (3)
SEC63	Polycystic liver disease, 174050 (3)
SECISBP2	Thyroid hormone metabolism, abnormal, 609698 (3)
SEMA3A	Hypogonadotropic hypogonadism 16 with or without anosmia, 614897 (3)
SEMA3E	CHARGE syndrome, 214800 (3)
SEMA4A	Cone-rod dystrophy 10, 610283 (3)/Retinitis pigmentosa 35, 610282 (3)
SEPN1	Muscular dystrophy, rigid spine, 1, 602771 (3)/Myopathy, congenital, with fiber-type disproportion, 255310 (3)
SEPSECS	Pontocerebellar hypoplasia type 2D, 613811 (3)
SEPT12	Spermatogenic failure 10 614822 (3)
SEPT9	Amyotrophy, hereditary neuralgic 162100(3)
SERAC1	3-methylglutaconic aciduria with deafness, encephalopathy, and Leigh-like syndrome, 614739 (3)
SERPINA1	Emphysema due to AAT deficiency, 613490 (3)/Emphysema-cirrhosis, due to AAT deficiency, 613490 (3)/Hemorrhagic diathesis due to \'antithrombin\' Pittsburgh, 613490 (3)/Pulmonary disease, chronic obstructive, susceptibility to, 606963 (1)
SERPINA6	Corticosteroid-binding globulin deficiency
SERPINA7	Thyroxine-binding globulin deficiency
SERPINB6	Deafness, autosomal recessive 91, 613453 (3)
SERPINC1	Thrombophilia due to antithrombin III deficiency, 613118 (3)
SERPIND1	Heparin cofactor 2 deficiency
SERPINE1	Plasminogen activator inhibitor 1 deficiency
SERPINF1	Osteogenesis imperfecta, type VI, 613982 (3)
SERPINF2	Antiplasmin alpha 2 deficiency
SERPING1	Angioneurotic oedema
SERPINH1	Osteogenesis imperfecta, type X, 613848 (3)/Preterm premature rupture of the membranes, susceptibility to, 610504 (3)
SERPINI1	Encephalopathy, familial, with neuroserpin inclusion bodies, 604218 (3)

SETBP1	Mental retardation, autosomal dominant 29, 616078 (3)/Schinzel-Giedion midface retraction syndrome, 269150 (3)
SETD5	Mental retardation, autosomal dominant 23, 615761 (3)
SETX	Amyotrophic lateral sclerosis 4, juvenile, 602433 (3)/Ataxia-ocular apraxia-2, 606002 (3)
SF3B1	Myelodysplastic syndrome, somatic, 614286 (3)
SF3B4	Acrofacial dysostosis 1, Nager type, 154400 (3)
SFTPA1	{Pulmonary fibrosis, idiopathic, susceptibility to} 178500 (3)
SFTPA2	Pulmonary fibrosis, idiopathic 178500 (3)
SFTPB	Surfactant metabolism dysfunction, pulmonary, 1, 265120 (3)
SFTPC	Surfactant metabolism dysfunction, pulmonary, 2, 610913 (3)
SGCA	Muscular dystrophy, limb-girdle, type 2D, 608099 (3)
SGCB	Muscular dystrophy, limb-girdle, type 2E, 604286 (3)
SGCD	Cardiomyopathy, dilated, 1L, 606685 (3)/Muscular dystrophy, limb-girdle, type 2F, 601287 (3)
SGCE	Dystonia-11, myoclonic, 159900 (3)
SGCG	Muscular dystrophy, limb-girdle, type 2C, 253700 (3)
SGSH	Mucopolysaccharidisis type IIIA (Sanfilippo A), 252900 (3)
SH2B3	Erythrocytosis, somatic, 133100 (3)/Myelofibrosis, somatic, 254450 (3)/Thrombocythemia, somatic, 187950 (3)
SH2D1A	Lymphoproliferative syndrome, X-linked, 1, 308240 (3)
SH3BP2	Cherubism, 118400 (3)
SH3GL1	Leukemia, acute myeloid, 601626 (1)
SH3PXD2B	Frank-ter Haar syndrome, 249420 (3)
SH3TC2	Charcot-Marie-Tooth disease, type 4C, 601596 (3)/Mononeuropathy of the median nerve, mild, 613353 (3)
SHANK2	Autism susceptibility 17, 613436 (3)
SHANK3	Phelan-McDermid syndrome, 606232 (3)/Schizophrenia 15, 613950 (3)
SHFM1	Split hand/foot malformation 1 (4)
SHH	Holoprosencephaly-3, 142945 (3)/Microphthalmia with coloboma 5, 611638 (3)/Schizencephaly, 269160 (3)/Single median maxillary central incisor, 147250 (3)
SHOC2	Noonan-like syndrome with loose anagen hair, 607721 (3)
SHOX	Langer mesomelic dysplasia, 249700
SHROOM4	Stocco dos Santos X-linked mental retardation syndrome, 300434 (3)
SI	Sucrase-isomaltase deficiency, congenital, 222900 (3)
SIGMAR1	Amyotrophic lateral sclerosis 16, juvenile, 614373 (3)
SIL1	Marinesco-Sjogren syndrome, 248800 (3)
SIM1	Obesity, severe, 601665 (3)
SIX1	Brachiootic syndrome 3, 608389 (3)/Deafness, autosomal dominant 23, 605192 (3)
SIX3	Holoprosencephaly-2, 157170 (3)/Schizencephaly, 269160 (3)
SIX5	Branchiootorenal syndrome 2, 610896 (3)
SIX6	Microphthalmia with cataract 2, 212550 (3)
SKI	Shprintzen-Goldberg syndrome, 182212 (3)
SKIV2L	Trichohepatoenteric syndrome 2, 614602 (3)
SLC10A2	Bile acid malabsorption, primary, 613291 (3)
SLC11A2	Microcytic anaemia & iron overload
SLC12A1	Bartter syndrome, type 1, 601678 (3)
SLC12A3	Gitelman syndrome, 263800 (3)
SLC12A6	Agenesis of the corpus callosum with peripheral neuropathy, 218000 (3)
SLC16A1	Erythrocyte lactate transporter defect, 245340 (3)/Hyperinsulinemic hypoglycemia, familial, 7, 610021 (3)/Monocarboxylate transporter 1 deficiency, 616095 (3)
SLC16A12	Cataract, juvenile, with microcornea and glucosuria, 612018 (3)
SLC16A2	Allan-Herndon-Dudley syndrome, 300523 (3)
SLC17A5	Salla disease, 604369 (3)/Sialic acid storage disorder, infantile, 269920 (3)
SLC17A8	Deafness, autosomal dominant 25, 605583 (3)
SLC19A2	Thiamine-responsive megaloblastic anemia syndrome, 249270 (3)

SLC19A3	Thiamine metabolism dysfunction syndrome 2 (biotin- or thiamine-responsive encephalopathy type 2), 607483 (3)
SLC1A1	Dicarboxylic aminoaciduria, 222730 (3)/
SLC1A3	Episodic ataxia, type 6, 612656 (3)
SLC20A2	Basal ganglia calcification, idiopathic, 1, 213600 (3)
SLC22A12	Hypouricemia, renal, 220150 (3)
SLC22A18	Breast cancer, somatic 114480 (3)/Lung cancer, somatic 211980 (3)/Rhabdomyosarcoma, somatic 268210 (3)
SLC22A5	Carnitine deficiency, systemic primary, 212140 (3)
SLC24A1	Night blindness, congenital stationary (complete), 1D, autosomal recessive, 613830 (3)
SLC25A12	Hypomyelination, global cerebral, 612949 (3)
SLC25A13	Citrullinemia, adult-onset type II, 603471 (3)/Citrullinemia, type II, neonatal-onset, 605814 (3)
SLC25A15	Hyperornithinemia-hyperammonemia-homocitrullinemia syndrome, 238970 (3)
SLC25A19	Microcephaly, Amish type, 607196 (3)/Thiamine metabolism dysfunction syndrome 4 (progressive polyneuropathy type), 613710 (3)
SLC25A20	Carnitine-acylcarnitine translocase deficiency, 212138 (3)
SLC25A22	Epileptic encephalopathy, early infantile, 3, 609304 (3)
SLC25A3	Mitochondrial phosphate carrier deficiency, 610773 (3)
SLC25A38	Anemia, sideroblastic, pyridoxine-refractory, autosomal recessive, 205950 (3)
SLC25A4	Mitochondrial DNA depletion syndrome 12 (cardiomyopathic type), 615418 (3)/Progressive external ophthalmoplegia with mitochondrial DNA deletions 3, 609283 (3)
SLC26A1	?Nephrolithiasis, calcium oxalate 167030 (3)
SLC26A2	Achondrogenesis Ib, 600972 (3)/Atelosteogenesis II, 256050 (3)/De la Chapelle dysplasia, 256050 (3)/Diastrophic dysplasia, 222600 (3)/Diastrophic dysplasia, broad bone-platyspondylic variant, 222600 (3)/Epiphyseal dysplasia, multiple, 4, 226900 (3)
SLC26A3	Diarrhea 1, secretory chloride, congenital, 214700 (3)
SLC26A4	Deafness, autosomal recessive 4, with enlarged vestibular aqueduct, 600791 (3)/Pendred syndrome, 274600 (3)
SLC26A5	Deafness, autosomal recessive 61, 613865 (3)
SLC27A4	Ichthyosis prematurity syndrome, 608649 (3)
SLC29A3	Histiocytosis-lymphadenopathy plus syndrome, 602782 (3)
SLC2A1	Dystonia 9, 601042 (3)/GLUT1 deficiency syndrome 1, 606777 (3)/GLUT1 deficiency syndrome 2, 612126 (3)/Epilepsy, idiopathic generalized, suscpetibility to, 12, 614847 (3)
SLC2A10	Arterial tortuosity syndrome, 208050 (3)
SLC2A2	Fanconi-Bickel syndrome, 227810 (3)/Diabetes mellitus, noninsulin-dependent, 135853 (3)
SLC2A9	Hypouricemia, renal, 2, 612076 (3)/Uric acid concentration, serum, QTL 2, 612076 (3)
SLC30A10	Hypermanganesemia with dystonia, polycythemia, and cirrhosis, 613280 (3)
SLC30A2	Zinc deficiency, transient neonatal, 608118 (3)
SLC31A1	Intellectual disability
SLC33A1	Congenital cataracts, hearing loss, and neurodegeneration, 614482 (3)/Spastic paraplegia 42, autosomal dominant, 612539 (3)
SLC34A1	Fanconi renotubular syndrome 2, 613388 (3)/Nephrolithiasis/osteoporosis, hypophosphatemic, 1, 612286 (3)
SLC34A2	Testicular microlithiasis, 610441 (3)/Pulmonary alveolar microlithiasis, 265100 (3)
SLC34A3	Hypophosphatemic rickets with hypercalciuria, 241530 (3)
SLC35A1	Congenital disorder of glycosylation, type IIf, 603585 (3)
SLC35C1	Congenital disorder of glycosylation, type IIc, 266265 (3)
SLC35D1	Schneckenbecken dysplasia, 269250 (3)
SLC36A2	Hyperglycinuria, 138500 (3)/Iminoglycinuria, digenic, 242600 (3)
SLC37A4	Glycogen storage disease Ib, 232220 (3)/Glycogen storage disease Ic, 232240 (3)
SLC39A13	Spondylocheirodysplasia, Ehlers-Danlos syndrome-like, 612350 (3)
SLC39A4	Acrodermatitis enteropathica, 201100 (3)
SLC3A1	Cystinuria, 220100 (3)
SLC40A1	Hemochromatosis, type 4, 606069 (3)

SLC45A2	Albinism, oculocutaneous, type IV, 606574 (3)/[Skin/hair/eye pigmentation 5, black/nonblack hair], 227240 (3)/[Skin/hair/eye pigmentation 5, dark/fair skin], 227240 (3)/[Skin/hair/eye pigmentation 5, dark/light eyes], 227240 (3)
SLC46A1	Folate malabsorption, hereditary, 229050 (3)
SLC4A1	Ovalocytosis (3)/Renal tubular acidosis, distal, AD, 179800 (3)/Renal tubular acidosis, distal, AR, 611590 (3)/Spherocytosis, type 4, 612653 (3)/[Blood group, Diego], 110500 (3)/[Blood group, Froese], 601551 (3)/[Blood group, Swann], 601550 (3)/[Blood group, Waldner], 112010 (3)/[Blood group, Wright], 112050 (3)/[Malaria, resistance to], 611162 (3)
SLC4A11	Corneal dystrophy, Fuchs endothelial, 4, 613268 (3)/Corneal endothelial dystrophy 2, autosomal recessive, 217700 (3)/Corneal endothelial dystrophy and perceptive deafness, 217400 (3)
SLC4A4	Renal tubular acidosis, proximal, with ocular abnormalities, 604278 (3)
SLC52A2	Brown-Vialetto-Van Laere syndrome 2, 614707 (3)
SLC52A3	Brown-Vialetto-Van Laere syndrome 1, 211530 (3)/Fazio-Londe disease, 211500 (3)
SLC5A1	Glucose/galactose malabsorption, 606824 (3)
SLC5A2	Renal glucosuria, 233100 (3)
SLC5A5	Thyroid dyshormonogenesis 1, 274400 (3)
SLC5A7	Neuronopathy, distal hereditary motor, type VIIA, 158580 (3)
SLC6A1	Myoclonic-atonic epilepsy 616421 (3)
SLC6A19	Hartnup disorder, 234500 (3)/Hyperglycinuria, 138500 (3)/Iminoglycinuria, digenic, 242600 (3)
SLC6A2	Orthostatic intolerance, 604715 (3)
SLC6A20	Hyperglycinuria, 138500 (3)/Iminoglycinuria, digenic, 242600 (3)
SLC6A3	Parkinsonism-dystonia, infantile, 613135 (3)/Nicotine dependence, protection against, 188890 (3)
SLC6A5	Hyperekplexia 3, 614618 (3)
SLC6A8	Cerebral creatine deficiency syndrome 1, 300352 (3)
SLC7A7	Lysinuric protein intolerance, 222700 (3)
SLC7A9	Cystinuria, 220100 (3)
SLC9A3R1	Nephrolithiasis/osteoporosis, hypophosphatemic, 2, 612287 (3)
SLC9A6	Mental retardation, X-linked syndromic, Christianson type, 300243 (3)
SLCO1B1	Hyperbilirubinemia, Rotor type, digenic, 237450 (3)
SLCO1B3	Hyperbilirubinemia, Rotor type, digenic, 237450 (3)
SLCO2A1	Hypertrophic osteoarthropathy, primary, autosomal recessive 2, 614441 (3)
SLITRK1	Tourette syndrome, 137580 (3)/Trichotillomania, 613229 (3)
SLURP1	Meleda disease, 248300 (3)
SLX4	Fanconi anemia, complementation group P, 613951 (3)
SMAD3	Loeys-Dietz syndrome, type 3, 613795 (3)
SMAD4	Pulmonary arterial hypertension
SMAD6	Aortic valve disease 2, 614823 (3)
SMAD9	Pulmonary arterial hypertension
SMARCA2	Nicolaides-Baraitser syndrome, 601358 (3)
SMARCA4	Mental retardation, autosomal dominant 16, 614609 (3)/Rhabdoid tumor predisposition syndrome 2, 613325 (3)
SMARCAD1	Adermatoglyphia, 136000 (3)
SMARCAL1	Schimke immunoosseous dysplasia, 242900 (3)
SMARCB1	Mental retardation, autosomal dominant 15, 614608 (3)/Rhabdoid tumors, somatic, 609322 (3)/Rhabdoid predisposition syndrome 1, 609322 (3)/Schwannomatosis-1, susceptibility to, 162091 (3)
SMARCE1	Coffin-Siris syndrome 5 616938 (3)
SMC1A	Cornelia de Lange syndrome
SMC3	Cornelia de Lange syndrome
SMN1	Spinal muscular atrophy-1, 253300 (3)/Spinal muscular atrophy-2, 253550 (3)/Spinal muscular atrophy-3, 253400 (3)/Spinal muscular atrophy-4, 271150 (3)
SMN2	{Spinal muscular atrophy, type III, modifier of} 253400
SMOC1	Microphthalmia with limb anomalies, 206920 (3)
SMOC2	Dentin dysplasia, type I, with microdontia and misshapen teeth, 125400 (3)
SMPD1	Niemann-Pick disease, type A 257200 (3)/Niemann-Pick disease, type B 607616 (3)

SMPX	Deafness, X-linked 4, 300066 (3)
SNAI2	Piebaldism, 172800 (3)/Waardenburg syndrome, type 2D, 608890 (3)
SNAP29	Cerebral dysgenesis, neuropathy, ichthyosis, and palmoplantar keratoderma syndrome, 609528 (3)
SNCA	Dementia, Lewy body, 127750 (3)/Parkinson disease 1, 168601 (3)/Parkinson disease 4, 605543 (3)
SNCB	Dementia, Lewy body, 127750 (3)
SNIP1	Psychomotor retardation, epilepsy, and craniofacial dysmorphism, 614501 (3)
SNRNP200	Retinitis pigmentosa 33, 610359 (3)
SNRPN	Prader-Willi syndrome, 176270 (3)
SNTA1	Sudden infant death syndrome
SOBP	Mental retardation, anterior maxillary protrusion, and strabismus, 613671 (3)
SOD1	Amyotrophic lateral sclerosis 1, 105400 (3)
SOS1	Fibromatosis, gingival, 135300 (3)/Noonan syndrome 4, 610733 (3)
SOST	Craniodiaphyseal dysplasia, autosomal dominant, 122860 (3)/Sclerosteosis 1, 269500 (3)/Van Buchem disease, 239100 (3)
SOX10	PCWH syndrome, 609136 (3)/Waardenburg syndrome, type 2E, with or without neurologic involvement, 611584 (3)/Waardenburg syndrome, type 4C, 613266 (3)
SOX17	Vesicoureteral reflux 3, 613674 (3)
SOX18	Hypotrichosis-lymphedema-telangiectasia syndrome, 607823 (3)
SOX2	Microphthalmia, syndromic 3, 206900 (3)/Optic nerve hypoplasia and abnormalities of the central nervous system, 206900 (3)
SOX3	Langer mesomelic dysplasia
SOX9	Acampomelic campomelic dysplasia, 114290 (3)/Campomelic dysplasia with autosomal sex reversal, 114290 (3)/Campomelic dysplasia, 114290 (3)
SP110	Hepatic venoocclusive disease with immunodeficiency, 235550 (3)/Mycobacterium tuberculosis, susceptibility to, 607948 (3)
SP7	Osteogenesis imperfecta, type XII, 613849 (3)
SPAST	Spastic paraplegia 4, autosomal dominant, 182601 (3)
SPATA16	Spermatogenic failure 6, 102530 (3)
SPATA7	Leber congenital amaurosis 3, 604232 (3)/Retinitis pigmentosa, juvenile, autosomal recessive, 604232 (3)
SPECC1L	Facial clefting, oblique, 1, 600251 (3)
SPG11	Spastic paraplegia 11, autosomal recessive, 604360 (3)
SPG20	Troyer syndrome, 275900 (3)
SPG21	Mast syndrome 248900 (3)
SPG7	Spastic paraplegia 7, autosomal recessive 607259 (3)
SPINK1	Pancreatitis, hereditary, 167800 (3)/Tropical calcific pancreatitis, 608189 (3)/Fibrocalculous pancreatic diabetes, susceptibility to, 608189 (3)
SPINK5	Atopy, 147050 (3)/Netherton syndrome, 256500 (3)
SPINT2	Diarrhea 3, secretory sodium, congenital, syndromic, 270420 (3)
SPR	Dystonia, dopa-responsive, due to sepiapterin reductase deficiency, 612716 (3)
SPRED1	Legius syndrome, 611431 (3)
SPTA1	Elliptocytosis-2, 130600 (3)/Pyropoikilocytosis, 266140 (3)/Spherocytosis, type 3, 270970 (3)
SPTAN1	Epileptic encephalopathy, early infantile, 5, 613477 (3)
SPTB	Anemia, neonatal hemolytic, fatal and near-fatal (3)/Elliptocytosis-3 (3)/Spherocytosis, type 2 (3)
SPTBN2	Spinocerebellar ataxia 5, 600224 (3)/Spinocerebellar ataxia, autosomal recessive 14, 615386 (3)
SPTLC1	Neuropathy, hereditary sensory and autonomic, type IA, 162400 (3)
SPTLC2	Neuropathy, hereditary sensory and autonomic, type IC, 613640 (3)
SQSTM1	Paget disease of bone, 602080 (3)
SRCAP	Floating-Harbor syndrome, 136140 (3)
SRD5A2	Pseudovaginal perineoscrotal hypospadias, 264600 (3)
SRD5A3	Congenital disorder of glycosylation, type Iq, 612379 (3)/Kahrizi syndrome, 612713 (3)
SRP72	Bone marrow failure syndrome 1, 614675 (3)
SRPX2	Rolandic epilepsy, mental retardation, and speech dyspraxia, 300643 (3)
SRY	46XX sex reversal 1, 400045 (3)/46XY sex reversal 1, 400044 (3)
SSTR5	Somatostatin analog, resistance to, 102200 (3)

ST14	Ichthyosis, congenital, autosomal recessive 11, 602400 (3)
ST3GAL3	Epileptic encephalopathy, early infantile, 15, 615006 (3)/Mental retardation, autosomal recessive 12, 611090 (3)
ST3GAL5	Amish infantile epilepsy syndrome
STAR	Lipoid adrenal hyperplasia, 201710 (3)
STAT1	Immunodeficiency 31A, mycobacteriosis, autosomal dominant, 614892 (3)/Immunodeficiency 31B, mycobacterial and viral infections, autosomal recessive, 613796 (3)/Immunodeficiency 31C, autosomal dominant, 614162 (3)
STAT3	Autoimmune disease, multisystem, infantile-onset, 615952 (3)/Hyper-IgE recurrent infection syndrome, 147060 (3)
STAT5B	Growth hormone insensitivity with immunodeficiency, 245590 (3)/Leukemia, acute promyelocytic, STAT5B/RARA type (3)
STIL	Microcephaly 7, primary, autosomal recessive, 612703 (3)
STIM1	Immunodeficiency 10, 612783 (3)/Myopathy, tubular aggregate, 1 160565 (3)/Stormorken syndrome, 185070 (3)
STK11	Melanoma, malignant, somatic (3)/Pancreatic cancer, 260350 (3)/Peutz-Jeghers syndrome, 175200 (3)/Testicular tumor, somatic, 273300 (3)
STK4	T-cell immunodeficiency, recurrent infections, autoimmunity, and cardiac malformations, 614868 (3)
STOX1	Preeclampsia/eclampsia 4, 609404 (3)
STRA6	Microphthalmia, isolated, with coloboma 8, 601186 (3)/Microphthalmia, syndromic 9, 601186 (3)
STRADA	Polyhydramnios, megalencephaly, and symptomatic epilepsy, 611087 (3)
STRC	Deafness, autosomal recessive 16, 603720 (3)
STS	Ichthyosis, X-linked, 308100 (3)
STX11	Hemophagocytic lymphohistiocytosis, familial, 4, 603552 (3)
STX16	Pseudohypoparathyroidism, type IB, 603233 (3)
STXBP1	Epileptic encephalopathy, early infantile, 4, 612164 (3)
STXBP2	Hemophagocytic lymphohistiocytosis, familial, 5, 613101 (3)
SUCLA2	Mitochondrial DNA depletion syndrome 5 (encephalomyopathic with or without methylmalonic aciduria), 612073 (3)
SUCLG1	Mitochondrial DNA depletion syndrome 9 (encephalomyopathic type with methylmalonic aciduria), 245400 (3)
SUFU	Basal cell nevus syndrome, 109400 (3)/Medulloblastoma, desmoplastic, 155255 (3)/Meningioma, familial, susceptibility to, 607174 (3)
SUMF1	Multiple sulfatase deficiency, 272200 (3)
SUMO1	Orofacial cleft 10, 613705 (3)
SUOX	Sulfite oxidase deficiency, 272300 (3)
SURF1	Leigh syndrome, due to COX deficiency, 256000 (3)
SYCP3	Spermatogenic failure 4, 270960 (3)/Pregnancy loss, susceptibility to (3)
SYN1	Epilepsy, X-linked, with variable learning disabilities and behavior disorders, 300491 (3)
SYNE1	Emery-Dreifuss muscular dystrophy 4, autosomal dominant, 612998 (3)/Spinocerebellar ataxia, autosomal recessive 8, 610743 (3)
SYNE2	Emery-Dreifuss muscular dystrophy 5, autosomal dominant, 612999 (3)
SYNGAP1	Mental retardation, autosomal dominant 5, 612621 (3)
SYP	Mental retardation, X-linked 96, 300802 (3)
SYT14	Spinocerebellar ataxia, autosomal recessive 11, 614229 (3)
TAB2	Congenital heart defects, nonsyndromic, 2, 614980 (3)
TAC3	Hypogonadotropic hypogonadism 10 with or without anosmia, 614839 (3)
TACR3	Hypogonadotropic hypogonadism 11 with or without anosmia, 614840 (3)
TACSTD2	Corneal dystrophy, gelatinous drop-like, 204870 (3)
TAF1	Dystonia-Parkinsonism, X-linked, 314250 (3)
TAF15	Chondrosarcoma, extraskeletal myxoid, 612237 (1)
TAF2	Mental retardation, autosomal recessive 40, 615599 (3)
TAF6	Alazami-Yuan syndrome 617126 (3)
TAL1	Leukemia-1, T-cell acute lymphocytic (3)

TALDO1	Transaldolase deficiency, 606003 (3)
TAP1	Bare lymphocyte syndrome, type I, 604571 (3)
TAP2	Bare lymphocyte syndrome, type I, due to TAP2 deficiency, 604571 (3)/Wegener-like granulomatosis (3)
TAPBP	Bare lymphocyte syndrome, type I, 604571 (3)
TARDBP	Amyotrophic lateral sclerosis 10, with or without FTD, 612069 (3)/Frontotemporal lobar degeneration, TARDBP-related, 612069 (3)
TAT	Tyrosinemia, type II, 276600 (3)
TAZ	Barth syndrome, 302060 (3)
TBC1D24	DOOR syndrome, 220500 (3)/Deafness , autosomal recessive 86, 614617 (3)/Deafness, autosomal dominant 65, 616044 (3)/Epileptic encephalopathy, early infantile, 16, 615338 (3)/Myoclonic epilepsy, infantile, familial, 605021 (3)
TBCE	Hypoparathyroidism-retardation-dysmorphism syndrome, 241410 (3)/Kenny-Caffey syndrome-1, 244460 (3)
TBP	Spinocerebellar ataxia 17, 607136 (3)/Parkinson disease, susceptibility to, 168600 (3)
TBX1	DiGeorge syndrome 188400 (3)/Tetralogy of Fallot 187500 (3)/Velocardiofacial syndrome 192430 (3)
TBX15	Cousin syndrome, 260660 (3)
TBX19	Adrenocorticotropic hormone deficiency, 201400 (3)
TBX20	Atrial septal defect 4, 611363 (3)
TBX21	Asthma and nasal polyps, 208550 (3)/Asthma, aspirin-induced, susceptibility to, 208550 (3)
TBX22	Abruzzo-Erickson syndrome, 302905 (3)/Cleft palate with ankyloglossia, 303400 (3)
TBX3	Ulnar-mammary syndrome, 181450 (3)
TBX4	Small patella syndrome, 147891 (3)
TBX5	Holt-Oram syndrome, 142900 (3)
TBXAS1	Thromboxane synthase deficiency, 614158 (1)/Ghosal hematodiaphyseal syndrome, 231095 (3)
TCAP	Cardiomyopathy, dilated, 1N, 607487 (3)/Muscular dystrophy, limb-girdle, type 2G, 601954 (3)
TCF4	Pitt-Hopkins syndrome, 610954 (3)
TCIRG1	Osteopetrosis, autosomal recessive 1, 259700 (3)
TCL1A	Leukemia/lymphoma, T-cell (2)
TCN2	Transcobalamin II deficiency, 275350 (3)
TCOF1	Treacher Collins syndrome 1, 154500 (3)
TCTN1	Joubert syndrome
TCTN2	Meckel syndrome 8, 613885 (3)
TCTN3	Joubert syndrome 18, 614815 (3)/Orofaciodigital syndrome IV, 258860 (3)
TDGF1	Forebrain defects (3)
TDP1	Spinocerebellar ataxia, autosomal recessive with axonal neuropathy, 607250 (3)
TDRD7	Cataract 36, 613887 (3)
TEAD1	Sveinsson choreoretinal atrophy, 108985 (3)
TECR	Mental retardation, autosomal recessive 14, 614020 (3)
TECTA	Deafness, autosomal dominant 8/12, 601543 (3)/Deafness, autosomal recessive 21, 603629 (3)
TEK	Venous malformations, multiple cutaneous and mucosal, 600195 (3)
TERT	Bone marrow failure, telomere-related, 1, 614742 (3)/Coronary artery disease (3)/Dyskeratosis congenita, autosomal dominant 2, 613989 (3)/Dyskeratosis congenita, autosomal recessive 4, 613989 (3)/Leukemia, acute myeloid, 601626 (3)/Melanoma, cutaneous malignant, 9, 615134 (3)/Pulmonary fibrosis, telomere-related, 1, 614742 (3)
TET2	Myelodysplastic syndrome, somatic, 614286 (3)
TF	Atransferrinemia, 209300 (3)
TFAP2A	Branchiooculofacial syndrome, 113620 (3)
TFAP2A-AS1	Branchiooculofacial syndrome 113620 (3)
TFAP2B	Char syndrome, 169100 (3)
TFE3	Renal cell carcinoma, papillary, 1, 300854 (3)
TFG	Spastic paraplegia 57, autosomal recessive, 615658 (3)/Hereditary motor and sensory neuropathy, proximal type, 604484 (3)
TFR2	Hemochromatosis, type 3, 604250 (3)
TG	Thyroid dyshormonogenesis 3, 274700 (3)/Autoimmune thyroid disease, susceptibility to, 3, 608175 (3)
TGFB1	Camurati-Engelmann disease, 131300 (3)/Cystic fibrosis lung disease, modifier of, 219700 (3)

TGFB2	Loeys-Dietz syndrome, type 4, 614816 (3)
TGFB3	Rienhoff syndrome, 615582 (3)/Arrhythmogenic right ventricular dysplasia 1, 107970 (3)
TGFBI	Corneal dystrophy, Avellino type, 607541 (3)/Corneal dystrophy, Groenouw type I, 121900 (3)/Corneal dystrophy, Reis-Bucklers type, 608470 (3)/Corneal dystrophy, Thiel-Behnke type, 602082 (3)/Corneal dystrophy, epithelial basement membrane, 121820 (3)/Corneal dystrophy, lattice type I, 122200 (3)/Corneal dystrophy, lattice type IIIA, 608471 (3)
TGFBR1	Loeys-Dietz syndrome, type 1, 609192 (3)/Multiple self-healing squamous epithelioma, susceptiblity to, 132800 (3)
TGFBR2	Colorectal cancer, hereditary nonpolyposis, type 6, 614331 (3)/Esophageal cancer, somatic, 133239 (3)/Loeys-Dietz syndrome, type 2, 610168 (3)
TGIF1	Holoprosencephaly
TGM1	Ichthyosis, congenital, autosomal recessive 1, 242300 (3)
TGM5	Peeling skin syndrome 2, 609796 (3)
TGM6	Spinocerebellar ataxia 35, 613908 (3)
TH	Segawa syndrome, recessive, 605407 (3)
THAP1	Dystonia 6, torsion, 602629 (3)
THBD	Thrombophilia due to thrombomodulin defect, 614486 (3)/Hemolytic uremic syndrome, atypical, susceptibility to, 6, 612926 (3)
THOC2	Psychomotor retardation & cerebellar hypoplasia
THPO	Thrombocythemia 1, 187950 (3)
THRA	Hypothyroidism, congenital, nongoitrous, 6, 614450 (3)
THRB	Thyroid hormone resistance, 188570 (3)/Thyroid hormone resistance, autosomal recessive, 274300 (3)/Thyroid hormone resistance, selective pituitary, 145650 (3)
TICAM1	Encephalopathy, acute, infection-induced, susceptibility to, 6, 614850 (3)
TIMM8A	Deafness, X-linked 1, progressive (3)/Jensen syndrome, 311150 (3)/Mohr-Tranebjaerg syndrome, 304700 (3)
TIMP3	Sorsby fundus dystrophy, 136900 (3)
TINF2	Dyskeratosis congenita, autosomal dominant 3, 613990 (3)/Revesz syndrome, 268130 (3)
TJP2	Cholestasis, progressive familial intrahepatic 4, 615878 (3)/Hypercholanemia, familial, 607748 (3)
TK2	Mitochondrial DNA depletion syndrome 2 (myopathic type), 609560 (3)
TLL1	Atrial septal defect 6, 613087 (3)
TLR3	HIV1 infection, resistance to, 609423 (3)/Herpes simplex encephalitis, susceptibility to, 2 613002 (3)
TMC1	Deafness, autosomal dominant 36, 606705 (3)/Deafness, autosomal recessive 7, 600974 (3)
TMC6	Epidermodysplasia verruciformis, 226400 (3)
TMC8	Epidermodysplasia verruciformis, 226400 (3)
TMCO1	Craniofacial dysmorphism, skeletal anomalies, and mental retardation syndrome, 213980 (3)
TMEM114	Cataract, congenital
TMEM126A	Optic atrophy-7, 612989 (3)
TMEM135	Intellectual disability
TMEM138	Joubert syndrome 16, 614465 (3)
TMEM165	Congenital disorder of glycosylation, type IIk, 614727 (3)
TMEM216	Joubert syndrome 2, 608091 (3)/Meckel syndrome 2, 603194 (3)
TMEM231	Joubert syndrome 20, 614970 (3)/Meckel syndrome, type 11, 615397 (3)
TMEM237	Joubert syndrome 14, 614424 (3)
TMEM43	Arrhythmogenic right ventricular dysplasia 5, 604400 (3)/Emery-Dreifuss muscular dystrophy 7, AD, 614302 (3)
TMEM5	Muscular dystrophy-dystroglycanopathy (congenital with brain and eye anomalies), type A, 10, 615041 (3)
TMEM67	COACH syndrome, 216360 (3)/Joubert syndrome 6, 610688 (3)/Meckel syndrome 3, 607361 (3)/Nephronophthisis 11, 613550 (3)/Bardet-Biedl syndrome 14, modifier of, 209900 (3)
TMEM70	Mitochondrial complex V (ATP synthase) deficiency, nuclear type 2, 614052 (3)
TMIE	Deafness, autosomal recessive 6, 600971 (3)
TMLHE	Epsilon-trimethyllysine hydroxylase deficiency, 300872 (3)
TMPO	Cardiomyopathy, dilated, 1T, 613740 (3)
TMPRSS15	Enteropeptidase deficiency
TMPRSS3	Deafness, autosomal recessive 8/10, 601072 (3)

TMPRSS6	Iron-refractory iron deficiency anemia, 206200 (3)
TNF	Asthma, susceptibility to, 600807 (3)/Dementia, vascular, susceptibility to (3)/Malaria, cerebral, susceptibility to, 611162 (3)/Migraine without aura, susceptibility to, 157300 (3)/Septic shock, susceptibility to (3)
TNFRSF10B	Squamous cell carcinoma, head and neck, 275355 (3)
TNFRSF11A	Osteolysis, familial expansile, 174810 (3)/Osteopetrosis, autosomal recessive 7, 612301 (3)/Paget disease of bone, 602080 (3)
TNFRSF11B	Paget disease, juvenile, 239000 (3)
TNFRSF13B	Immunodeficiency, common variable, 2, 240500 (3)/Immunoglobulin A deficiency 2, 609529 (3)
TNFRSF13C	Immunodeficiency, common variable, 4, 613494 (3)
TNFRSF1A	Periodic fever, familial, 142680 (3)/Multiple sclerosis, susceptibility to, 5, 614810 (3)
TNFSF11	Osteopetrosis, autosomal recessive 2, 259710 (3)
TNNC1	Cardiomyopathy, dilated, 1Z, 611879 (3)/Cardiomyopathy, familial hypertrophic, 13, 613243 (3)
TNNI2	Arthrogryposis multiplex congenita, distal, type 2B, 601680 (3)
TNNI3	Cardiomyopathy, dilated, 1FF, 613286 (3)/Cardiomyopathy, dilated, 2A, 611880 (3)/Cardiomyopathy, familial hypertrophic, 7, 613690 (3)/Cardiomyopathy, familial restrictive, 1, 115210 (3)
TNNT1	Nemaline myopathy 5, Amish type, 605355 (3)
TNNT2	Cardiomyopathy, dilated, 1D, 601494 (3)/Cardiomyopathy, familial hypertrophic, 2, 115195 (3)/Cardiomyopathy, familial restrictive, 3, 612422 (3)/Left ventricular noncompaction 6, 601494 (3)
TNNT3	Arthyrgryposis, distal, type 2B, 601680 (3)
TNXB	Ehlers-Danlos syndrome, autosomal dominant, hypermobility type, 130020 (3)/Ehlers-Danlos syndrome, autosomal recessive, due to tenascin X deficiency, 606408 (3)/Vesicoureteral reflux 8, 615963 (3)
TOPORS	Retinitis pigmentosa 31, 609923 (3)
TOR1A	Torsion dystonia, early onset
TP53	Adrenal cortical carcinoma, 202300 (3)/Breast cancer, 114480 (3)/Choroid plexus papilloma, 260500 (3)/Colorectal cancer, 114500 (3)/Hepatocellular carcinoma, 114550 (3)/Li-Fraumeni syndrome, 151623 (3)/Nasopharyngeal carcinoma, 607107 (3)/Osteosarcoma, 259500 (3)/Pancreatic cancer, 260350 (3)/Basal cell carcinoma 7, 614740 (3)/Glioma susceptibility 1, 137800 (3)
TP63	ADULT syndrome, 103285 (3)/Ectrodactyly, ectodermal dysplasia, and cleft lip/palate syndrome 3, 604292 (3)/Hay-Wells syndrome, 106260 (3)/Limb-mammary syndrome, 603543 (3)/Orofacial cleft 8, 129400 (3)/Rapp-Hodgkin syndrome, 129400 (3)/Split-hand/foot malformation 4, 605289 (3)
TPI1	Hemolytic anemia due to triosephosphate isomerase deficiency, 615512 (3)
TPK1	Thiamine metabolism dysfunction syndrome 5 (episodic encephalopathy type), 614458 (3)
TPM1	Cardiomyopathy, dilated, 1Y, 611878 (3)/Cardiomyopathy, familial hypertrophic, 3, 115196 (3)/Left ventricular noncompaction 9, 611878 (3)
TPM2	Arthrogryposis multiplex congenita, distal, type 1, 108120 (3)/Arthrogryposis, distal, type 2B, 601680 (3)/CAP myopathy 2, 609285 (3)/Nemaline myopathy 4, autosomal dominant, 609285 (3)
TPM3	CAP myopathy 1, 609284 (3)/Myopathy, congenital, with fiber-type disproportion, 255310 (3)/Nemaline myopathy 1, autosomal dominant or recessive, 609284 (3)
TPMT	6-mercaptopurine sensitivity, 610460 (3)
TPO	Thyroid dyshormonogenesis 2A, 274500 (3)
TPP1	Ceroid lipofuscinosis, neuronal, 2, 204500 (3)/Spinocerebellar ataxia, autosomal recessive 7, 609270 (3)
TPRN	Deafness, autosomal recessive 79, 613307 (3)
TRAPPC2	Spondyloepiphyseal dysplasia tarda, 313400 (3)
TRAPPC9	Mental retardation, autosomal recessive 13, 613192 (3)
TRDN	Ventricular tachycardia, catecholaminergic polymorphic, 5, with or without muscle weakness, 615441 (3)
TREH	Trehalase deficiency, 612119 (1)
TREM2	Nasu-Hakola disease, 221770 (3)
TREX1	Aicardi-Goutieres syndrome 1, dominant and recessive, 225750 (3)/Chilblain lupus, 610448 (3)/Vasculopathy, retinal, with cerebral leukodystrophy, 192315 (3)/Systemic lupus erythematosus, susceptibility to, 152700 (3)
TRH	Thyrotropin-releasing hormone deficiency, 275120 (1)
TRHR	Thyrotropin-releasing hormone resistance, generalized (3)
TRIM24	Thyroid carcinoma, papillary, 188550 (3)

TRIM32	Bardet-Biedl syndrome 11, 615988 (3)/Muscular dystrophy, limb-girdle, type 2H, 254110 (3)
TRIM33	Thyroid carcinoma, papillary, 188550 (3)
TRIM37	Mulibrey nanism, 253250 (3)
TRIOBP	Deafness, autosomal recessive 28, 609823 (3)
TRIP11	Achondrogenesis, type IA, 200600 (3)
TRMT1	Intellectual disability
TRMU	Liver failure, transient infantile, 613070 (3)/Deafness, mitochondrial, modifier of, 580000 (3)
TRPC6	Glomerulosclerosis, focal segmental, 2, 603965 (3)
TRPM1	Night blindness, congenital stationary (complete), 1C, autosomal recessive, 613216 (3)
TRPM4	Progressive familial heart block, type IB, 604559 (3)
TRPM6	Hypomagnesemia 1, intestinal, 602014 (3)
TRPS1	Trichorhinophalangeal syndrome, type I, 190350 (3)/Trichorhinophalangealsyndrome, type III, 190351 (3)
TRPV3	Olmsted syndrome, 614594 (3)
TRPV4	Brachyolmia type 3, 113500 (3)/Digital arthropathy-brachydactyly, familial, 606835 (3)/Hereditary motor and sensory neuropathy, type IIc, 606071 (3)/Metatropic dysplasia, 156530 (3)/Parastremmatic dwarfism, 168400 (3)/SED, Maroteaux type, 184095 (3)/Scapuloperoneal spinal muscular atrophy, 181405 (3)/Spinal muscular atrophy, distal, congenital nonprogressive, 600175 (3)/Spondylometaphyseal dysplasia, Kozlowski type, 184252 (3)/[Sodium serum level QTL 1], 613508 (3)
TSC1	Focal cortical dysplasia, Taylor balloon cell type, 607341 (3)/Lymphangioleiomyomatosis, 606690 (3)/Tuberous sclerosis-1, 191100 (3)
TSC2	Lymphangioleiomyomatosis, somatic, 606690 (3)/Tuberous sclerosis-2, 613254 (3)
TSEN2	Pontocerebellar hypoplasia type 2B, 612389 (3)
TSEN34	Pontocerebellar hypoplasia type 2C, 612390 (3)
TSEN54	Pontocerebellar hypoplasia type 5, 610204 (3)/Pontocerebellar hypoplasia type 2A, 277470 (3)/Pontocerebellar hypoplasia type 4, 225753 (3)
TSFM	Combined oxidative phosphorylation deficiency 3, 610505 (3)
TSG101	Breast cancer, somatic, 114480 (3)
TSHB	Hypothryoidism, congenital, nongoitrous 4, 275100 (3)
TSHR	Hyperthyroidism, familial gestational, 603373 (3)/Hyperthyroidism, nonautoimmune, 609152 (3)/Hypothyroidism, congenital, nongoitrous, 1 275200 (3)/Thyroid adenoma, hyperfunctioning, somatic (3)/Thyroid carcinoma with thyrotoxicosis (3)
TSHZ1	Aural atresia, congenital, 607842 (3)
TSPAN12	Exudative vitreoretinopathy 5, 613310 (3)
TSPAN7	Mental retardation, X-linked 58, 300210 (3)
TSPEAR	Deafness, autosomal recessive 98, 614861 (3)
TSPYL1	Sudden infant death with dysgenesis of the testes syndrome, 608800 (3)
TTBK2	Spinocerebellar ataxia 11, 604432 (3)
TTC19	Mitochondrial complex III deficiency, nuclear type 2, 615157 (3)
TTC21B	Nephronophthisis 12, 613820 (3)/Short-rib thoracic dysplasia 4 with or without polydactyly, 613819 (3)
TTC37	Trichohepatoenteric syndrome 1, 222470 (3)
TTC7A	Gastrointestinal defects and immunodeficiency syndrome 243150 (3)
TTC8	Retinitis pigmentosa 51, 613464 (3)/Bardet-Biedl syndrome 8, 615985 (3)
TTI2	Mental retardation, autosomal recessive 39, 615541 (3)
TTN	Cardiomyopathy, dilated, 1G, 604145 (3)/Cardiomyopathy, familial hypertrophic, 9, 613765 (3)/Muscular dystrophy, limb-girdle, type 2J, 608807 (3)/Myopathy, early-onset, with fatal cardiomyopathy, 611705 (3)/Myopathy, proximal, with early respiratory muscle involvement, 603689 (3)/Tibial muscular dystrophy, tardive, 600334 (3)
TTPA	Ataxia with isolated vitamin E deficiency, 277460 (3)
TTR	Amyloidosis, hereditary, transthyretin-related, 105210 (3)/Carpal tunnel syndrome, familial, 115430 (3)/[Dystransthyretinemic hyperthyroxinemia], 145680 (3)
TUBA1A	Lissencephaly 3, 611603 (3)
TUBA8	Polymicrogyria with optic nerve hypoplasia, 613180 (3)
TUBB1	Macrothrombocytopenia, autosomal dominant, TUBB1-related, 613112 (3)
TUBB2B	Polymicrogyria, symmetric or asymmetric, 610031 (3)

TUBB3	Cortical dysplasia, complex, with other brain malformations 1, 614039 (3)/Fibrosis of extraocular muscles, congenital, 3A, 600638 (3)
TUBGCP6	Microcephaly and chorioretinopathy, autosomal recessive, 1, 251270 (3)
TUFM	Combined oxidative phosphorylation deficiency 4, 610678 (3)
TULP1	Leber congenital amaurosis 15, 613843 (3)/Retinitis pigmentosa 14, 600132 (3)
TUSC3	Mental retardation, autosomal recessive 7, 611093 (3)
TWIST1	Craniosynostosis, type 1, 123100 (3)/Robinow-Sorauf syndrome, 180750 (3)/Saethre-Chotzen syndrome with eyelid anomalies, 101400 (3)/Saethre-Chotzen syndrome, 101400 (3)
TWIST2	Focal facial dermal dysplasia 3, Setleis type, 227260 (3)
TXNRD2	Cardiomyopathy, dilated
TYK2	Immunodeficiency 35, 611521 (3)
TYMP	Mitochondrial DNA depletion syndrome 1 (MNGIE type), 603041 (3)
TYR	Albinism, oculocutaneous, type IA, 203100 (3)/Albinism, oculocutaneous, type IB, 606952 (3)/Waardenburg syndrome/albinism, digenic, 103470 (3)/[Skin/hair/eye pigmentation 3, blue/green eyes], 601800 (3)/[Skin/hair/eye pigmentation 3, light/dark/freckling skin], 601800 (3)/Melanoma, cutaneous malignant, susceptibility to, 8, 601800 (3)
TYROBP	Nasu-Hakola disease, 221770 (3)
TYRP1	Albinism, oculocutaneous, type III, 203290 (3)/[Skin/hair/eye pigmentation, variation in, 11 (Melanesian blond hair)], 612271 (3)
UBA1	Spinal muscular atrophy, X-linked 2, infantile, 301830 (3)
UBB	Cleft palate, isolated, 119540 (2)
UBE2A	Mental retardation, X-linked syndromic, Nascimento-type, 300860 (3)
UBE3A	Angelman syndrome, 105830 (3)
UBIAD1	Corneal dystrophy, Schnyder type, 121800 (3)
UBQLN2	Amyotrophic lateral sclerosis 15, with or without frontotemporal dementia, 300857 (3)
UBR1	Johanson-Blizzard syndrome, 243800 (3)
UBR7	Intellectual disability
UFD1L	Catch 22 syndrome
UGT1A1	Crigler-Najjar syndrome, type I, 218800 (3)/Crigler-Najjar syndrome, type II, 606785 (3)/Hyperbilirubinemia, familial transient neonatal, 237900 (3)/[Bilirubin, serum level of, QTL1], 601816 (3)/[Gilbert syndrome], 143500 (3)
UMOD	Glomerulocystic kidney disease with hyperuricemia and isosthenuria, 609886 (3)/Hyperuricemic nephropathy, familial juvenile 1, 162000 (3)/Medullary cystic kidney disease 2, 603860 (3)
UMPS	Orotic aciduria, 258900 (3)
UNC119	Cone-rod dystrophy (3)/
UNC13D	Hemophagocytic lymphohistiocytosis, familial, 3, 608898 (3)
UNC93B1	Herpes simplex encephalitis, susceptibility to, 1, 610551 (3)
UNG	Immunodeficiency with hyper IgM, type 5, 608106 (3)
UPB1	Beta-ureidopropionase deficiency, 613161 (3)
UPF3B	Mental retardation, X-linked, syndromic 14, 300676 (3)
UPK3A	Renal hypodysplasia
UQCRB	Mitochondrial complex III deficiency, nuclear type 3, 615158 (3)
UQCRQ	Mitochondrial complex III deficiency, nuclear type 4, 615159 (3)
UROC1	Urocanase deficiency, 276880 (3)
UROD	Porphyria cutanea tarda, 176100 (3)/Porphyria, hepatoerythropoietic, 176100 (3)
UROS	Porphyria, congenital erythropoietic, 263700 (3)
USB1	Poikiloderma with neutropenia 604173 (3)
USH1C	Deafness, autosomal recessive 18A, 602092 (3)/Usher syndrome, type 1C, 276904 (3)
USH1G	Usher syndrome 1g
USH2A	Retinitis pigmentosa 39, 613809 (3)/Usher syndrome, type 2A, 276901 (3)
USP9Y	Spermatogenic failure, Y-linked, 2, 415000 (3)
UVSSA	UV-sensitive syndrome 3, 614640 (3)
VANGL1	Caudal regression syndrome, 600145 (3)/Neural tube defects, 182940 (3)
VAPB	Amyotrophic lateral sclerosis 8, 608627 (3)/Spinal muscular atrophy, late-onset, Finkel type, 182980 (3)

VCAN	Wagner syndrome 1, 143200 (3)
VCL	Cardiomyopathy, dilated, 1W, 611407 (3)/Cardiomyopathy, familial hypertrophic, 15, 613255 (3)
VCP	Amyotrophic lateral sclerosis 14, with or without frontotemporal dementia, 613954 (3)/Inclusion body myopathy with early-onset Paget disease and frontotemporal dementia 1, 167320 (3)
VDR	Osteoporosis, involutional, 166710 (1)/Rickets, vitamin D-resistant, type IIA, 277440 (3)
VHL	Erythrocytosis, familial, 2, 263400 (3)/Hemangioblastoma, cerebellar, somatic (3)/Pheochromocytoma, 171300 (3)/Renal cell carcinoma, somatic, 144700 (3)/von Hippel-Lindau syndrome, 193300 (3)
VIPAS39	Arthrogryposis, renal dysfunction, and cholestasis 2 613404 (3)
VKORC1	Vitamin K-dependent clotting factors, combined deficiency of, 2 607473 (3)/Warfarin resistance 122700 (3)
VLDLR	Cerebellar hypoplasia and mental retardation with or without quadrupedal locomotion 1, 224050 (3)
VLDLR-AS1	Cerebellar hypoplasia and mental retardation with or without quadrupedal locomotion 1 224050 (3)
VMA21	X-linked myopathy with excessive autophagy
VPS13A	Choreoacanthocytosis, 200150 (3)
VPS13B	Cohen syndrome, 216550 (3)
VPS33B	Arthrogryposis, renal dysfunction, and cholestasis 1, 208085 (3)
VPS35	Parkinson disease 17, 614203 (3)
VPS37A	Spastic paraplegia 53, autosomal recessive, 614898 (3)
VRK1	Pontocerebellar hypoplasia type 1A, 607596 (3)
VSX1	Corneal dystrophy, posterior polymorphous, 1, 122000 (3)/Craniofacial anomalies and anterior segment dysgenesis syndrome, 614195 (3)/Keratoconus 1, 148300 (3)
VSX2	Microphthalmia with coloboma 3, 610092
VWF	von Willebrand disease, type 1, 193400 (3)/von Willebrand disease, types 2A, 2B, 2M, and 2N, 613554 (3)/von Willibrand disease, type 3, 277480 (3)
WAS	Neutropenia, severe congenital, X-linked, 300299 (3)/Thrombocytopenia, X-linked, 313900 (3)/Thrombocytopenia, X-linked, intermittent, 313900 (3)/Wiskott-Aldrich syndrome, 301000 (3)
WDPCP	Bardet-Biedl syndrome 15, 615992 (3)/
WDR11	Hypogonadotropic hypogonadism 14 with or without anosmia, 614858 (3)
WDR19	Cranioectodermal dysplasia 4, 614378 (3)/Nephronophthisis 13, 614377 (3)/Short-rib thoracic dysplasia 5 with or without polydactyly, 614376 (3)
WDR35	Cranioectodermal dysplasia 2, 613610 (3)/Short-rib thoracic dysplasia 7 with or without polydactyly, 614091 (3)
WDR36	Glaucoma 1, open angle, G, 609887 (3)
WDR45L	Intellectual disability
WDR62	Microcephaly 2, primary, autosomal recessive, with or without cortical malformations, 604317 (3)
WDR65	Van der Woude syndrome 2 606713 (3)
WDR72	Amelogenesis imperfecta, type IIA3, 613211 (3)
WDR81	Cerebellar ataxia, mental retardation, and dysequilibrium syndrome 2, 610185 (3)
WFS1	Cataract 41, 116400 (3)/Deafness, autosomal dominant 6/14/38, 600965 (3)/Wolfram syndrome, 222300 (3)/Wolfram-like syndrome, autosomal dominant, 614296 (3)/Diabetes mellitus, noninsulin-dependent, association with, 125853 (3)
WHSC1	Wolf-Hirschhorn syndrome
WHSC1L1	Leukemia, acute myeloid, 601626 (3)
WHSC2	Wolf-Hirschhorn syndrome
WIPF1	Wiskott-Aldrich syndrome 2, 614493 (3)
WISP3	Arthropathy, progressive pseudorheumatoid, of childhood, 208230 (3)/Spondyloepiphyseal dysplasia tarda with progressive arthropathy, 208230 (3)
WNK1	Neuropathy, hereditary sensory and autonomic, type II, 201300 (3)/Pseudohypoaldosteronism, type IIC, 614492 (3)
WNK4	Pseudohypoaldosteronism, type IIB, 614491 (3)
WNT10A	Odontoonychodermal dysplasia, 257980 (3)/Schopf-Schulz-Passarge syndrome, 224750 (3)/Tooth agenesis, selective, 4, 150400 (3)
WNT10B	Split-hand/foot malformation 6, 225300 (3)
WNT3	Tetra-amelia syndrome, 273395 (3)
WNT4	Mullerian aplasia and hyperandrogenism, 158330 (3)/SERKAL syndrome, 611812 (3)

WNT5A	Robinow syndrome, autosomal dominant, 180700 (3)
WNT7A	Fuhrmann syndrome, 228930 (3)/Ulna and fibula, absence of, with severe limb deficiency, 276820 (3)
WRAP53	Dyskeratosis congenita, autosomal recessive 3, 613988 (3)
WRN	Werner syndrome
WT1	Denys-Drash syndrome, 194080 (3)/Frasier syndrome, 136680 (3)/Meacham syndrome, 608978 (3)/Mesothelioma, somatic, 156240 (3)/Nephrotic syndrome, type 4, 256370 (3)/Wilms tumor, type 1, 194070 (3)
WWOX	Epileptic encephalopathy, early infantile, 28, 616211 (3)/Esophageal squamous cell carcinoma, somatic, 133239 (3)/Spinocrebellar ataxia, autosomal recessive 12, 614322 (3)
XDH	Xanthinuria, type I, 278300 (3)
XIAP	Lymphoproliferative syndrome, X-linked, 2, 300635 (3)
XK	McLeod syndrome with or without chronic granulomatous disease, 300842 (3)
XPA	Xeroderma pigmentosum, group A, 278700 (3)
XPC	Xeroderma pigmentosum, group C, 278720 (3)
XPNPEP3	Nephronophthisis-like nephropathy 1, 613159 (3)
YARS	Charcot-Marie-Tooth disease, dominant intermediate C, 608323 (3)
YARS2	Myopathy, lactic acidosis, and sideroblastic anemia 2, 613561 (3)
YWHAE	Left ventricular noncompaction & hypoplasia of the corpus callosum
ZAP70	Selective T-cell defect, 269840 (3)
ZBTB16	Leukemia, acute promyelocytic, PL2F/RARA type (3)/Skeletal defects, genital hypoplasia, and mental retardation, 612447 (3)
ZBTB24	Immunodeficiency-centromeric instability-facial anomalies syndrome-2, 614069 (3)
ZBTB40	Intellectual disability
ZC3H14	Intellectual disability, nonsyndromic, autosomal recessive
ZC4H2	Wieacker-Wolff syndrome, 314580 (3)
ZCCHC12	Mental retardation
ZCCHC8	Intellectual disability
ZDHHC15	Mental retardation, X-linked 91, 300577 (3)
ZDHHC9	Mental retardation, X-linked syndromic, Raymond type, 300799 (3)
ZEB1	Corneal dystrophy, Fuchs endothelial, 6, 613270 (3)/Corneal dystrophy, posterior polymorphous, 3, 609141 (3)
ZEB2	Mowat-Wilson syndrome 235730 (3)
ZFHX4	Ptosis, congenital, 178300 (2)
ZFP57	Diabetes mellitus, transient neonatal, 1, 601410 (3)
ZFPM2	46,XY sex reversal 9, 616067 (3)/Diaphragmatic hernia 3, 610187 (3)/Tetralogy of Fallot, 187500 (3)
ZFYVE26	Spastic paraplegia 15, autosomal recessive, 270700 (3)
ZFYVE27	Spastic paraplegia 33, autosomal dominant, 610244 (3)
ZIC2	Holoprosencephaly-5, 609637 (3)
ZIC3	Congenital heart defects, nonsyndromic, 1, X-linked, 306955 (3)/Heterotaxy, visceral, 1, X-linked 306955 (3)/VACTERL association, X-linked, 314390 (3)
ZMPSTE24	Mandibuloacral dysplasia with type B lipodystrophy, 608612 (3)/Restrictive dermopathy, lethal, 275210 (3)
ZNF238	Abnormalities of the corpus callosum
ZNF41	Mental retardation
ZNF423	Joubert syndrome 19, 614844 (3)/Nephronophthisis 14, 614844 (3)
ZNF469	Brittle cornea syndrome 1 229200 (3)
ZNF513	Retinitis pigmentosa 58, 613617 (3)
ZNF526	Intellectual disability
ZNF592	Spinocerebellar ataxia, autosomal recessive 5, 606937 (3)
ZNF644	Myopia 21, autosomal dominant, 614167 (3)
ZNF674	Mental retardation, X-linked
ZNF711	Mental retardation, X-linked 97, 300803 (3)
ZNF750	Seborrhea-like dermatitis with psoriasiform elements, 610227 (3)
ZNF81	Mental retardation, X-linked 45, 300498 (3)

*: (1)The disorder is placed on the map due to its association with a gene, but the underlying defect is not known; (2)The disorder was placed on the map by statistical methods; (3)The molecular basis of the disorder is known