Mendelioom panel

versie	13-Mar-2018 (3380 genen)	Centrum voor Medische Genetica Gent

Gene	OMIM gene ID	Associated phenotype, OMIM phenotype ID, phenotype mapping key and inheritance pattern
A2M	103950	Alpha-2-macroglobulin deficiency, 614036 (1), Autosomal dominant; {Alzheimer disease, susceptibility to}, 104300 (3), Autosomal dominant
A4GALT	607922	[Blood group, P1Pk system, P(2) phenotype], 111400 (3); [Blood group, P1Pk system, p phenotype], 111400 (3); NOR polyagglutination syndrome, 111400 (3)
AAAS	605378	Achalasia-addisonianism-alacrimia syndrome, 231550 (3), Autosomal recessive
AANAT	600950	No OMIM phenotype
AARS	601065	Charcot-Marie-Tooth disease, axonal, type 2N, 613287 (3), Autosomal dominant; Epileptic encephalopathy, early infantile, 29, 616339 (3), Autosomal recessive
AARS2	612035	Combined oxidative phosphorylation deficiency 8, 614096 (3), Autosomal recessive; Leukoencephalopathy, progressive, with ovarian failure, 615889 (3), Autosomal recessive
AASS	605113	Hyperlysinemia, 238700 (3), Autosomal recessive; Saccharopinuria, 268700 (1), Autosomal recessive
ABAT	137150	GABA-transaminase deficiency, 613163 (3), Autosomal recessive
ABCA1	600046	{Coronary artery disease in familial hypercholesterolemia, protection against}, 143890 (3), Autosomal dominant; HDL deficiency, type 2, 604091 (3); Tangier disease, 205400 (3), Autosomal recessive
ABCA12	607800	Ichthyosis, congenital, autosomal recessive 4A, 601277 (3), Autosomal recessive; Ichthyosis, congenital, autosomal recessive 4B (harlequin), 242500 (3), Autosomal recessive
ABCA3	601615	Surfactant metabolism dysfunction, pulmonary, 3, 610921 (3), Autosomal recessive
ABCA4	601691	Cone-rod dystrophy 3, 604116 (3); Fundus flavimaculatus, 248200 (3), Autosomal recessive; {Macular degeneration, age-related, 2}, 153800 (3), Autosomal dominant; Retinal dystrophy, early-onset severe, 248200 (3), Autosomal recessive; Retinitis pigmentosa 19, 601718 (3), Autosomal recessive; Stargardt disease 1, 248200 (3), Autosomal recessive
ABCB1	171050	{Colchicine resistance}, 120080 (3); {Inflammatory bowel disease 13}, 612244 (3)
ABCB11	603201	Cholestasis, benign recurrent intrahepatic, 2, 605479 (3), Autosomal recessive; Cholestasis, progressive familial intrahepatic 2, 601847 (3), Autosomal recessive

ABCB6	605452	[Blood group, Langereis system], 111600 (3); Dyschromatosis universalis hereditaria 3, 615402 (3), Autosomal dominant; Microphthalmia, isolated, with coloboma 7, 614497 (3), Autosomal dominant; Pseudohyperkalemia, familial, 2, due to red cell leak, 609153 (3), Autosomal dominant
ABCB7	300135	Anemia, sideroblastic, with ataxia, 301310 (3), X-linked recessive
ABCC11	607040	[Axillary odor, variation in], 117800 (3), Autosomal dominant; [Colostrum secretion, variation in], 117800 (3), Autosomal dominant; [Earwax, wet/dry], 117800 (3), Autosomal dominant
ABCC2	601107	Dubin-Johnson syndrome, 237500 (3), Autosomal recessive
ABCC6	603234	Arterial calcification, generalized, of infancy, 2, 614473 (3), Autosomal recessive; Pseudoxanthoma elasticum, 264800 (3), Autosomal recessive; Pseudoxanthoma elasticum, forme fruste, 177850 (3), Autosomal dominant
ABCC9	601439	Atrial fibrillation, familial, 12, 614050 (3), Autosomal dominant; Cardiomyopathy, dilated, 1O, 608569 (3); Hypertrichotic osteochondrodysplasia, 239850 (3), Autosomal dominant
ABCD1	300371	Adrenoleukodystrophy, 300100 (3), X-linked recessive; Adrenomyeloneuropathy, adult, 300100 (3), X-linked recessive
ABCD3	170995	?Bile acid synthesis defect, congenital, 5, 616278 (3), Autosomal recessive
ABCD4	603214	Methylmalonic aciduria and homocystinuria, cblJ type, 614857 (3), Autosomal recessive
ABCG2	603756	[Junior blood group system], 614490 (3); [Uric acid concentration, serum, QTL1], 138900 (3), ?Autosomal dominant
ABCG5	605459	Sitosterolemia, 210250 (3), Autosomal recessive
ABCG8	605460	{Gallbladder disease 4}, 611465 (3); Sitosterolemia, 210250 (3), Autosomal recessive
ABHD12	613599	Polyneuropathy, hearing loss, ataxia, retinitis pigmentosa, and cataract, 612674 (3), Autosomal recessive
ABHD5	604780	Chanarin-Dorfman syndrome, 275630 (3), Autosomal recessive
ABO	110300	[Blood group, ABO system], 616093 (3)
ACAD8	604773	Isobutyryl-CoA dehydrogenase deficiency, 611283 (3)
ACAD9	611103	Mitochondrial complex I deficiency due to ACAD9 deficiency, 611126 (3), Autosomal recessive
ACADM	607008	Acyl-CoA dehydrogenase, medium chain, deficiency of, 201450 (3), Autosomal recessive
ACADS	606885	Acyl-CoA dehydrogenase, short-chain, deficiency of, 201470 (3), Autosomal recessive
ACADSB	600301	2-methylbutyrylglycinuria, 610006 (3), Autosomal recessive
ACADVL	609575	VLCAD deficiency, 201475 (3), Autosomal recessive
ACAT1	607809	Alpha-methylacetoacetic aciduria, 203750 (3), Autosomal recessive
ACAT2	100678	?ACAT2 deficiency, 614055 (1), Isolated cases

ACD	609377	?Dyskeratosis congenita, autosomal dominant 6, 616553 (3), Autosomal recessive, Autosomal dominant; ?Dyskeratosis congenita, autosomal recessive 7, 616553 (3), Autosomal recessive, Autosomal dominant
ACE	106180	[Angiotensin I-converting enzyme, benign serum increase] (3); {Microvascular complications of diabetes 3}, 612624 (3); {Myocardial infarction, susceptibility to} (3); Renal tubular dysgenesis, 267430 (3), Autosomal recessive; {SARS, progression of} (3); {Stroke, hemorrhagic}, 614519 (3)
ACKR1	613665	[Blood group, Duffy system], 110700 (3), Autosomal recessive, Autosomal dominant; {Malaria, vivax, protection against}, 611162 (3); [White blood cell count QTL], 611862 (3), Autosomal recessive
ACO2	100850	Infantile cerebellar-retinal degeneration, 614559 (3), Autosomal recessive; ?Optic atrophy 9, 616289 (3), Autosomal recessive
ACOX1	609751	Peroxisomal acyl-CoA oxidase deficiency, 264470 (3), Autosomal recessive
ACOX2	601641	Bile acid synthesis defect, congenital, 6, 617308 (3), Autosomal recessive
ACP2	171650	?Lysosomal acid phosphatase deficiency, 200950 (1), Autosomal recessive
ACP4	606362	Amelogenesis imperfecta, type IJ, 617297 (3), Autosomal recessive
ACP5	171640	Spondyloenchondrodysplasia with immune dysregulation, 607944 (3), Autosomal recessive
ACSF3	614245	Combined malonic and methylmalonic aciduria, 614265 (3)
ACSL4	300157	Mental retardation, X-linked 63, 300387 (3), X-linked dominant
ACTA1	102610	Myopathy, actin, congenital, with cores, 161800 (3), Autosomal recessive, Autosomal dominant; Myopathy, actin, congenital, with excess of thin myofilaments, 161800 (3), Autosomal recessive, Autosomal dominant; Myopathy, congenital, with fiber-type disproportion 1, 255310 (3), Autosomal recessive, Autosomal dominant; ?Myopathy, scapulohumeroperoneal, 616852 (3), Autosomal dominant; Nemaline myopathy 3, autosomal dominant or recessive, 161800 (3), Autosomal recessive, Autosomal dominant
ACTA2	102620	Aortic aneurysm, familial thoracic 6, 611788 (3), Autosomal dominant; Moyamoya disease 5, 614042 (3); Multisystemic smooth muscle dysfunction syndrome, 613834 (3), Autosomal dominant
ACTB	102630	Baraitser-Winter syndrome 1, 243310 (3), Autosomal dominant; ?Dystonia, juvenile-onset, 607371 (3), Autosomal dominant
ACTC1	102540	Atrial septal defect 5, 612794 (3), Autosomal dominant; Cardiomyopathy, dilated, 1R, 613424 (3), Autosomal dominant; Cardiomyopathy, hypertrophic, 11, 612098 (3), Autosomal dominant; Left ventricular noncompaction 4, 613424 (3), Autosomal dominant
ACTG1	102560	Baraitser-Winter syndrome 2, 614583 (3), Autosomal dominant; Deafness, autosomal dominant 20/26, 604717 (3), Autosomal dominant

ACTN1	102575	Bleeding disorder, platelet-type, 15, 615193 (3), Autosomal dominant
ACTN2	102573	Cardiomyopathy, dilated, 1AA, with or without LVNC, 612158 (3), Autosomal dominant; Cardiomyopathy, hypertrophic, 23, with or without LVNC, 612158 (3), Autosomal dominant
ACTN4	604638	Glomerulosclerosis, focal segmental, 1, 603278 (3), Autosomal dominant
ACVR2B	602730	Heterotaxy, visceral, 4, autosomal, 613751 (3)
ACY1	104620	Aminoacylase 1 deficiency, 609924 (3), Autosomal recessive
ADA	608958	Adenosine deaminase deficiency, partial, 102700 (3), Autosomal recessive, Somatic mosaicism; Severe combined immunodeficiency due to ADA deficiency, 102700 (3), Autosomal recessive, Somatic mosaicism
ADAM10	602192	{Alzheimer disease 18, susceptibility to}, 615590 (3); Reticulate acropigmentation of Kitamura, 615537 (3), Autosomal dominant
ADAM17	603639	?Inflammatory skin and bowel disease, neonatal, 1, 614328 (3), Autosomal recessive
ADAM9	602713	Cone-rod dystrophy 9, 612775 (3)
ADAMTS10	608990	Weill-Marchesani syndrome 1, recessive, 277600 (3), Autosomal recessive
ADAMTS13	604134	Thrombotic thrombocytopenic purpura, familial, 274150 (3), Autosomal recessive
ADAMTS17	607511	Weill-Marchesani 4 syndrome, recessive, 613195 (3), Autosomal recessive
ADAMTS18	607512	Microcornea, myopic chorioretinal atrophy, and telecanthus, 615458 (3), Autosomal recessive
ADAMTS2	604539	Ehlers-Danlos syndrome, dermatosparaxis type, 225410 (3), Autosomal recessive
ADAMTSL2	612277	Geleophysic dysplasia 1, 231050 (3), Autosomal recessive
ADAMTSL4	610113	Ectopia lentis et pupillae, 225200 (3), Autosomal recessive; Ectopia lentis, isolated, autosomal recessive, 225100 (3), Autosomal recessive
ADAT3	615302	Mental retardation, autosomal recessive 36, 615286 (3), Autosomal recessive
ADCY1	103072	?Deafness, autosomal recessive 44, 610154 (3), Autosomal recessive
ADCY10	605205	{Hypercalciuria, absorptive, susceptibility to}, 143870 (3), Autosomal dominant
ADCY6	600294	?Lethal congenital contracture syndrome 8, 616287 (3), Autosomal recessive
ADD1	102680	{Hypertension, essential, salt-sensitive}, 145500 (3), Multifactorial
ADD3	601568	Cerebral palsy, spastic quadriplegic, 3, 617008 (3), Autosomal recessive
ADGRE2	606100	Vibratory urticaria, 125630 (3), Autosomal dominant
ADGRG2	300572	Vas deferens, congenital bilateral aplasia of, X-linked, 300985 (3), X-linked
ADGRG6	612243	Lethal congenital contracture syndrome 9, 616503 (3), Autosomal recessive

ADGRV1	602851	?Febrile seizures, familial, 4, 604352 (3), Autosomal dominant; Usher syndrome, type 2C, 605472 (3), Autosomal recessive; Usher syndrome, type 2C, GPR98/PDZD7 digenic, 605472 (3), Autosomal recessive
ADH1B	103720	{Aerodigestive tract cancer, squamous cell, alcohol-related, protection against}, 103780 (3), Multifactorial; {Alcohol dependence, protection against}, 103780 (3), Multifactorial
ADH1C	103730	{Alcohol dependence, protection against}, 103780 (3), Multifactorial; {Parkinson disease, susceptibility to}, 168600 (3), Isolated cases, Multifactorial
ADIPOQ	605441	Adiponectin deficiency, 612556 (3)
ADK	102750	Hypermethioninemia due to adenosine kinase deficiency, 614300 (3), Autosomal recessive
ADRA2B	104260	Epilepsy, myoclonic, familial adult, 2, 607876 (3), Autosomal dominant
ADRB1	109630	{Congestive heart failure and beta-blocker response, modifier of} (3); [Resting heart rate], 607276 (3)
ADRB2	109690	{Asthma, nocturnal, susceptibility to}, 600807 (3), Autosomal dominant; Beta-2-adrenoreceptor agonist, reduced response to (3); {Obesity, susceptibility to}, 601665 (3), Autosomal recessive, Autosomal dominant, Multifactorial
ADRB3	109691	{Obesity, susceptibility to}, 601665 (3), Autosomal recessive, Autosomal dominant, Multifactorial
ADSL	608222	Adenylosuccinase deficiency, 103050 (3), Autosomal recessive
ADSSL1	612498	Myopathy, distal, 5, 617030 (3), Autosomal recessive
AFF2	300806	Mental retardation, X-linked, FRAXE type, 309548 (3), X-linked recessive
AFF4	604417	CHOPS syndrome, 616368 (3), Autosomal dominant
AFG3L2	604581	Spastic ataxia 5, autosomal recessive, 614487 (3), Autosomal recessive; Spinocerebellar ataxia 28, 610246 (3), Autosomal dominant
AFP	104150	Alpha-fetoprotein deficiency, 615969 (3), Autosomal recessive; [Hereditary persistence of alpha-fetoprotein], 615970 (3), Autosomal dominant
AGA	613228	Aspartylglucosaminuria, 208400 (3), Autosomal recessive
AGBL1	615496	Corneal dystrophy, Fuchs endothelial, 8, 615523 (3), Autosomal dominant
AGBL5	615900	Retinitis pigmentosa 75, 617023 (3), Autosomal recessive
AGK	610345	Cataract 38, autosomal recessive, 614691 (3), Autosomal recessive; Sengers syndrome, 212350 (3), Autosomal recessive
AGL	610860	Glycogen storage disease IIIa, 232400 (3), Autosomal recessive; Glycogen storage disease IIIb, 232400 (3), Autosomal recessive
AGPAT2	603100	Lipodystrophy, congenital generalized, type 1, 608594 (3), Autosomal recessive
AGPS	603051	Rhizomelic chondrodysplasia punctata, type 3, 600121 (3), Autosomal recessive
AGRP	602311	{Leanness, inherited} (3); {Obesity, late-onset}, 601665 (3), Autosomal recessive, Autosomal dominant, Multifactorial

AGT	106150	{Hypertension, essential, susceptibility to}, 145500 (3), Multifactorial; {Preeclampsia, susceptibility to} (3); Renal tubular dysgenesis, 267430 (3), Autosomal recessive
AGTR1	106165	{Hypertension, essential}, 145500 (3), Multifactorial; Renal tubular dysgenesis, 267430 (3), Autosomal recessive
AGXT	604285	Hyperoxaluria, primary, type 1, 259900 (3), Autosomal recessive
AHDC1	615790	Xia-Gibbs syndrome, 615829 (3), Autosomal dominant
AHI1	608894	Joubert syndrome 3, 608629 (3), Autosomal recessive
AICDA	605257	Immunodeficiency with hyper-IgM, type 2, 605258 (3), Autosomal recessive
AIFM1	300169	Combined oxidative phosphorylation deficiency 6, 300816 (3), X-linked recessive; Cowchock syndrome, 310490 (3), X-linked recessive; Deafness, X-linked 5, 300614 (3), X-linked recessive
AIMP1	603605	Leukodystrophy, hypomyelinating, 3, 260600 (3), Autosomal recessive
AIP	605555	Pituitary adenoma 1, multiple types, 102200 (3), Autosomal dominant, Somatic mutation; Pituitary adenoma predisposition, 102000 (3), Autosomal dominant
AIPL1	604392	Cone-rod dystrophy, 604393 (3), Autosomal recessive; Leber congenital amaurosis 4, 604393 (3), Autosomal recessive; Retinitis pigmentosa, juvenile, 604393 (3), Autosomal recessive
AIRE	607358	Autoimmune polyendocrinopathy syndrome , type I, with or without reversible metaphyseal dysplasia, 240300 (3), Autosomal recessive, Autosomal dominant
AK1	103000	Hemolytic anemia due to adenylate kinase deficiency, 612631 (3), Autosomal recessive
AK2	103020	Reticular dysgenesis, 267500 (3), Autosomal recessive
AKAP10	604694	{Cardiac conduction defect, susceptibility to}, 115080 (3), Autosomal dominant
AKAP9	604001	?Long QT syndrome-11, 611820 (3), Autosomal dominant
AKR1C2	600450	46XY sex reversal 8, 614279 (3), Autosomal recessive
AKR1C4	600451	{46XY sex reversal 8, modifier of}, 614279 (3), Autosomal recessive
AKR1D1	604741	Bile acid synthesis defect, congenital, 2, 235555 (3), Autosomal recessive
AKT1	164730	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), Autosomal dominant
AKT2	164731	Diabetes mellitus, type II, 125853 (3), Autosomal dominant; Hypoinsulinemic hypoglycemia with hemihypertrophy, 240900 (3), Autosomal dominant
AKT3	611223	Megalencephaly-polymicrogyria-polydactyly-hydrocephalus syndrome 2, 615937 (3), Autosomal dominant
ALAD	125270	{Lead poisoning, susceptibility to}, 612740 (3), Autosomal recessive; Porphyria, acute hepatic, 612740 (3), Autosomal recessive

ALB	103600	Analbuminemia, 616000 (3); [Dysalbuminemic hyperthyroxinemia], 615999 (3)
ALDH18A1	138250	Cutis laxa, autosomal dominant 3, 616603 (3), Autosomal dominant; Cutis laxa, autosomal recessive, type IIIA, 219150 (3), Autosomal recessive, Isolated cases; Spastic paraplegia 9A, autosomal dominant, 601162 (3), Autosomal dominant; Spastic paraplegia 9B, autosomal recessive, 616586 (3), Autosomal recessive
ALDH1A3	600463	Microphthalmia, isolated 8, 615113 (3), Autosomal recessive
ALDH2	100650	Alcohol sensitivity, acute, 610251 (3), Autosomal dominant; {Esophageal cancer, alcohol-related, susceptibility to} (3); {Hangover, susceptibility to}, 610251 (3), Autosomal dominant; {Sublingual nitroglycerin, susceptibility to poor response to} (3)
ALDH3A2	609523	Sjogren-Larsson syndrome, 270200 (3), Autosomal recessive
ALDH4A1	606811	Hyperprolinemia, type II, 239510 (3), Autosomal recessive
ALDH5A1	610045	Succinic semialdehyde dehydrogenase deficiency, 271980 (3), Autosomal recessive
ALDH6A1	603178	Methylmalonate semialdehyde dehydrogenase deficiency, 614105 (3), Autosomal recessive
ALDH7A1	107323	Epilepsy, pyridoxine-dependent, 266100 (3), Autosomal recessive
ALDOB	612724	Fructose intolerance, hereditary, 229600 (3), Autosomal recessive
ALG1	605907	Congenital disorder of glycosylation, type Ik, 608540 (3), Autosomal recessive
ALG10	603313	{Long QT syndrome, acquired, reduced susceptibility to}, 613688 (3), Autosomal dominant
ALG11	613666	Congenital disorder of glycosylation, type Ip, 613661 (3), Autosomal recessive
ALG12	607144	Congenital disorder of glycosylation, type Ig, 607143 (3), Autosomal recessive
ALG14	612866	?Myasthenic syndrome, congenital, 15, without tubular aggregates, 616227 (3)
ALG2	607905	?Congenital disorder of glycosylation, type Ii, 607906 (3), Autosomal recessive; Myasthenic syndrome, congenital, 14, with tubular aggregates, 616228 (3), Autosomal recessive
ALG6	604566	Congenital disorder of glycosylation, type Ic, 603147 (3), Autosomal recessive
ALG8	608103	Congenital disorder of glycosylation, type Ih, 608104 (3), Autosomal recessive; Polycystic liver disease 3 with or without kidney cysts, 617874 (3), Autosomal dominant
ALG9	606941	Congenital disorder of glycosylation, type Il, 608776 (3), Autosomal recessive; Gillessen-Kaesbach-Nishimura syndrome, 263210 (3), Autosomal recessive
ALK	105590	{Neuroblastoma, susceptibility to, 3}, 613014 (3)
ALLC	612396	No OMIM phenotype
ALMS1	606844	Alstrom syndrome, 203800 (3), Autosomal recessive

ALOX12B	603741	Ichthyosis, congenital, autosomal recessive 2, 242100 (3), Autosomal recessive
ALOX5	152390	{Asthma, diminished response to antileukotriene treatment in}, 600807 (3), Autosomal dominant; {Atherosclerosis, susceptibility to} (3)
ALOX5AP	603700	{Stroke, susceptibility to}, 601367 (3), Multifactorial
ALOXE3	607206	Ichthyosis, congenital, autosomal recessive 3, 606545 (3), Autosomal recessive
ALPL	171760	Hypophosphatasia, adult, 146300 (3), Autosomal recessive, Autosomal dominant; Hypophosphatasia, childhood, 241510 (3), Autosomal recessive; Hypophosphatasia, infantile, 241500 (3), Autosomal recessive; Odontohypophosphatasia, 146300 (3), Autosomal recessive, Autosomal dominant
ALS2	606352	Amyotrophic lateral sclerosis 2, juvenile, 205100 (3), Autosomal recessive; Primary lateral sclerosis, juvenile, 606353 (3), Autosomal recessive; Spastic paralysis, infantile onset ascending, 607225 (3), Autosomal recessive
ALX1	601527	?Frontonasal dysplasia 3, 613456 (3)
ALX3	606014	Frontonasal dysplasia 1, 136760 (3), Autosomal recessive
ALX4	605420	{Craniosynostosis 5, susceptibility to}, 615529 (3), Autosomal dominant; Frontonasal dysplasia 2, 613451 (3), Autosomal recessive; Parietal foramina 2, 609597 (3), Autosomal dominant
AMACR	604489	Alpha-methylacyl-CoA racemase deficiency, 614307 (3), Autosomal recessive; Bile acid synthesis defect, congenital, 4, 214950 (3), Autosomal recessive
AMBN	601259	Amelogenesis imperfecta, type IF, 616270 (3), Autosomal recessive
AMELX	300391	Amelogenesis imperfecta, type 1E, 301200 (3), X-linked dominant
AMER1	300647	Osteopathia striata with cranial sclerosis, 300373 (3), X-linked dominant
AMH	600957	Persistent Mullerian duct syndrome, type I, 261550 (3), Autosomal recessive
AMHR2	600956	Persistent Mullerian duct syndrome, type II, 261550 (3), Autosomal recessive
AMMECR1	300195	Midface hypoplasia, hearing impairment, elliptocytosis, and nephrocalcinosis, 300990 (3), X-linked recessive
AMN	605799	Megaloblastic anemia-1, Norwegian type, 261100 (3), Autosomal recessive
AMPD1	102770	Myopathy due to myoadenylate deaminase deficiency, 615511 (3), Autosomal recessive
AMPD2	102771	Pontocerebellar hypoplasia, type 9, 615809 (3), Autosomal recessive; ?Spastic paraplegia 63, 615686 (3), Autosomal recessive
AMPD3	102772	[AMP deaminase deficiency, erythrocytic], 612874 (3), Autosomal recessive
AMT	238310	Glycine encephalopathy, 605899 (3), Autosomal recessive
ANG	105850	Amyotrophic lateral sclerosis 9, 611895 (3)
ANGPTL3	604774	Hypobetalipoproteinemia, familial, 2, 605019 (3), Autosomal recessive

ANGPTL4	605910	Plasma triglyceride level QTL, low, 615881 (3), Autosomal dominant
ANK2	106410	Cardiac arrhythmia, ankyrin-B-related, 600919 (3), Autosomal dominant; Long QT syndrome 4, 600919 (3), Autosomal dominant
ANK3	600465	?Mental retardation, autosomal recessive, 37, 615493 (3), Autosomal recessive
ANKH	605145	Chondrocalcinosis 2, 118600 (3), Autosomal dominant; Craniometaphyseal dysplasia, 123000 (3), Autosomal dominant
ANKLE2	616062	?Microcephaly 16, primary, autosomal recessive, 616681 (3), Autosomal recessive
ANKRD26	610855	Thrombocytopenia 2, 188000 (3), Autosomal dominant
ANKS6	615370	Nephronophthisis 16, 615382 (3), Autosomal recessive
ANLN	616027	Focal segmental glomerulosclerosis 8, 616032 (3), Autosomal dominant
ANO10	613726	Spinocerebellar ataxia, autosomal recessive 10, 613728 (3), Autosomal recessive
ANO3	610110	Dystonia 24, 615034 (3), Autosomal dominant
ANO5	608662	Gnathodiaphyseal dysplasia, 166260 (3), Autosomal dominant; Miyoshi muscular dystrophy 3, 613319 (3), Autosomal recessive; Muscular dystrophy, limb-girdle, type 2L, 611307 (3), Autosomal recessive
ANOS1	300836	Hypogonadotropic hypogonadism 1 with or without anosmia (Kallmann syndrome 1), 308700 (3), X-linked recessive
ANTXR1	606410	GAPO syndrome, 230740 (3), Autosomal recessive; {?Hemangioma, capillary infantile, susceptibility to}, 602089 (3), Autosomal dominant
ANTXR2	608041	Hyaline fibromatosis syndrome, 228600 (3), Autosomal recessive
ANXA5	131230	{Pregnancy loss, recurrent, susceptibility to, 3}, 614391 (3), Autosomal dominant
AP1S1	603531	MEDNIK syndrome, 609313 (3), Autosomal recessive
AP1S3	615781	{Psoriasis 15, pustular, susceptibility to}, 616106 (3), Autosomal dominant
AP2S1	602242	Hypocalciuric hypercalcemia, type III, 600740 (3), Autosomal dominant
AP3B1	603401	Hermansky-Pudlak syndrome 2, 608233 (3), Autosomal recessive
AP3B2	602166	Epileptic encephalopathy, early infantile, 48, 617276 (3), Autosomal recessive
AP3D1	607246	?Hermansky-Pudlak syndrome 10, 617050 (3), Autosomal recessive
AP4B1	607245	Spastic paraplegia 47, autosomal recessive, 614066 (3), Autosomal recessive
AP4E1	607244	Spastic paraplegia 51, autosomal recessive, 613744 (3), Autosomal recessive; Stuttering, familial persistent, 1, 184450 (3), Autosomal dominant
AP4M1	602296	Spastic paraplegia 50, autosomal recessive, 612936 (3), Autosomal recessive
AP4S1	607243	Spastic paraplegia 52, autosomal recessive, 614067 (3), Autosomal recessive
AP5Z1	613653	Spastic paraplegia 48, autosomal recessive, 613647 (3), Autosomal recessive

APBB2	602710	No OMIM phenotype
APCDD1	607479	Hypotrichosis 1, 605389 (3), Autosomal dominant
APOA1	107680	Amyloidosis, 3 or more types, 105200 (3), Autosomal dominant; ApoA-I and apoC-III deficiency, combined (3); Corneal clouding, autosomal recessive (3); Hypoalphalipoproteinemia, 604091 (3)
APOA2	107670	Apolipoprotein A-II deficiency (3); {Hypercholesterolemia, familial, modifier of}, 143890 (3), Autosomal dominant
APOA5	606368	Hyperchylomicronemia, late-onset, 144650 (3), Autosomal dominant; {Hypertriglyceridemia, susceptibility to}, 145750 (3), Autosomal dominant
APOB	107730	Hypercholesterolemia, due to ligand-defective apo B, 144010 (3), Autosomal dominant; Hypobetalipoproteinemia, 615558 (3), Autosomal recessive
APOC2	608083	Hyperlipoproteinemia, type Ib, 207750 (3), Autosomal recessive
APOC3	107720	Apolipoprotein C-III deficiency, 614028 (3)
APOE	107741	Alzheimer disease-2, 104310 (3), Autosomal dominant; {Coronary artery disease, severe, susceptibility to}, 617347 (3); Hyperlipoproteinemia, type III, 617347 (3); Lipoprotein glomerulopathy, 611771 (3); {?Macular degeneration, age-related}, 603075 (3), Autosomal dominant; Sea-blue histiocyte disease, 269600 (3), Autosomal recessive
APOL1	603743	{End-stage renal disease, nondiabetic, susceptibility to}, 612551 (3); {Glomerulosclerosis, focal segmental, 4, susceptibility to}, 612551 (3)
APOL4	607254	{Schizophrenia}, 181500 (1), Autosomal dominant
APOPT1	616003	Mitochondrial complex IV deficiency, 220110 (3), Autosomal recessive, Mitochondrial
APP	104760	Alzheimer disease 1, familial, 104300 (3), Autosomal dominant; Cerebral amyloid angiopathy, Dutch, Italian, Iowa, Flemish, Arctic variants, 605714 (3), Autosomal dominant
APPL1	604299	{Maturity-onset diabetes of the young, type 14}, 616511 (3), Autosomal dominant
APRT	102600	Adenine phosphoribosyltransferase deficiency, 614723 (3), Autosomal recessive
APTX	606350	Ataxia, early-onset, with oculomotor apraxia and hypoalbuminemia, 208920 (3), Autosomal recessive
AQP1	107776	[Aquaporin-1 deficiency], 110450 (3); [Blood group, Colton], 110450 (3)
AQP2	107777	Diabetes insipidus, nephrogenic, 125800 (3), Autosomal recessive, Autosomal dominant
AQP3	600170	[Blood group GIL], 607457 (3)
AQP7	602974	[Glycerol quantitative trait locus], 614411 (3), Autosomal recessive

AR	313700	Androgen insensitivity, 300068 (3), X-linked recessive; Androgen insensitivity, partial, with or without breast cancer, 312300 (3), X-linked recessive; Hypospadias 1, X-linked, 300633 (3), X-linked recessive; {Prostate cancer, susceptibility to}, 176807 (3), Autosomal dominant; Spinal and bulbar muscular atrophy of Kennedy, 313200 (3), X-linked recessive
ARCN1	600820	Short stature, rhizomelic, with microcephaly, micrognathia, and developmental delay, 617164 (3), Autosomal dominant
ARFGEF2	605371	Periventricular heterotopia with microcephaly, 608097 (3), Autosomal recessive
ARG1	608313	Argininemia, 207800 (3), Autosomal recessive
ARHGAP26	605370	Leukemia, juvenile myelomonocytic, somatic, 607785 (3)
ARHGAP31	610911	Adams-Oliver syndrome 1, 100300 (3), Autosomal dominant
ARHGDIA	601925	Nephrotic syndrome, type 8, 615244 (3), Autosomal recessive
ARHGEF6	300267	?Mental retardation, X-linked 46, 300436 (3), X-linked recessive
ARHGEF9	300429	Epileptic encephalopathy, early infantile, 8, 300607 (3), X-linked recessive
ARID1A	603024	Coffin-Siris syndrome 2, 614607 (3), Autosomal dominant
ARID1B	614556	Coffin-Siris syndrome 1, 135900 (3), Autosomal dominant
ARL13B	608922	Joubert syndrome 8, 612291 (3), Autosomal recessive
ARL2BP	615407	Retinitis pigmentosa with or without situs inversus, 615434 (3), Autosomal recessive
ARL6IP1	607669	?Spastic paraplegia 61, autosomal recessive, 615685 (3), Autosomal recessive
ARMC4	615408	Ciliary dyskinesia, primary, 23, 615451 (3), Autosomal recessive
ARMC5	615549	ACTH-independent macronodular adrenal hyperplasia 2, 615954 (3), Autosomal dominant, Somatic mutation
ARMS2	611313	{Macular degeneration, age-related, 8}, 613778 (3)
ARNT2	606036	?Webb-Dattani syndrome, 615926 (3), Autosomal recessive
ARSA	607574	Metachromatic leukodystrophy, 250100 (3), Autosomal recessive
ARSB	611542	Mucopolysaccharidosis type VI (Maroteaux-Lamy), 253200 (3), Autosomal recessive
ART4	110600	[Blood group, Dombrock], 616060 (3)
ARV1	611647	Epileptic encephalopathy, early infantile, 38, 617020 (3), Autosomal recessive
ARX	300382	Epileptic encephalopathy, early infantile, 1, 308350 (3), X-linked recessive; Hydranencephaly with abnormal genitalia, 300215 (3), X-linked; Lissencephaly, X-linked 2, 300215 (3), X-linked; Mental retardation, X-linked 29 and others, 300419 (3), X-linked recessive; Partington syndrome, 309510 (3), X-linked recessive; Proud syndrome, 300004 (3), X-linked
ASAH1	613468	Farber lipogranulomatosis, 228000 (3), Autosomal recessive; Spinal muscular atrophy with progressive myoclonic epilepsy, 159950 (3), Autosomal recessive
ASB10	615054	Glaucoma 1, open angle, F, 603383 (3)

ASCC1	614215	Barrett esophagus/esophageal adenocarcinoma, 614266 (3); ?Spinal muscular atrophy with congenital bone fractures 2, 616867 (3), Autosomal recessive
ASCL1	100790	Central hypoventilation syndrome, congenital, 209880 (3), Autosomal dominant; Haddad syndrome, 209880 (3), Autosomal dominant
ASIP	600201	[Skin/hair/eye pigmentation 9, brown/nonbrown eyes], 611742 (3); [Skin/hair/eye pigmentation 9, dark/light hair], 611742 (3)
ASL	608310	Argininosuccinic aciduria, 207900 (3), Autosomal recessive
ASNS	108370	Asparagine synthetase deficiency, 615574 (3), Autosomal recessive
ASPA	608034	Canavan disease, 271900 (3), Autosomal recessive
ASPH	600582	Traboulsi syndrome, 601552 (3), Autosomal recessive
ASPM	605481	Microcephaly 5, primary, autosomal recessive, 608716 (3), Autosomal recessive
ASPN	608135	{Lumbar disc degeneration}, 603932 (3); {Osteoarthritis susceptibility 3}, 607850 (3), Autosomal dominant
ASXL2	612991	Shashi-Pena syndrome, 617190 (3), Autosomal dominant
ASXL3	615115	Bainbridge-Ropers syndrome, 615485 (3), Autosomal dominant
ATAD3A	612316	Harel-Yoon syndrome, 617183 (3), Autosomal recessive, Autosomal dominant
ATCAY	608179	Ataxia, cerebellar, Cayman type, 601238 (3), Autosomal recessive
ATF6	605537	Achromatopsia 7, 616517 (3), Autosomal recessive
ATG16L1	610767	{Inflammatory bowel disease (Crohn disease) 10}, 611081 (3)
ATIC	601731	AICA-ribosiduria due to ATIC deficiency, 608688 (3), Autosomal recessive
ATL3	609369	Neuropathy, hereditary sensory, type IF, 615632 (3), Autosomal dominant
ATN1	607462	Dentatorubro-pallidoluysian atrophy, 125370 (3), Autosomal dominant
ATP13A2	610513	Kufor-Rakeb syndrome, 606693 (3), Autosomal recessive; Spastic paraplegia 78, autosomal recessive, 617225 (3), Autosomal recessive
ATP1A2	182340	Alternating hemiplegia of childhood 1, 104290 (3), Autosomal dominant; Migraine, familial basilar, 602481 (3), Autosomal dominant; Migraine, familial hemiplegic, 2, 602481 (3), Autosomal dominant
ATP1A3	182350	Alternating hemiplegia of childhood 2, 614820 (3), Autosomal dominant; CAPOS syndrome, 601338 (3), Autosomal dominant; Dystonia-12, 128235 (3), Autosomal dominant
ATP1B1	182330	[Blood pressure regulation QTL], 145500 (2), Multifactorial
ATP2A1	108730	Brody myopathy, 601003 (3), Autosomal recessive
ATP2A2	108740	Acrokeratosis verruciformis, 101900 (3), Autosomal dominant; Darier disease, 124200 (3), Autosomal dominant
ATP5F1A (ATP5A1)	164360	?Combined oxidative phosphorylation deficiency 22, 616045 (3), Autosomal recessive; ?Mitochondrial complex (ATP synthase) deficiency, nuclear type 4, 615228 (3), Autosomal recessive
ATP5F1E (ATP5E)	606153	?Mitochondrial complex V (ATP synthase) deficiency, nuclear type 3, 614053 (3)

ATP6AP1	300197	Immunodeficiency 47, 300972 (3), X-linked recessive
ATP6AP2	300556	Mental retardation, X-linked, syndromic, Hedera type, 300423 (3), X-linked recessive; ?Parkinsonism with spasticity, X-linked, 300911 (3), X-linked recessive
ATP6V0A2	611716	Cutis laxa, autosomal recessive, type IIA, 219200 (3), Autosomal recessive; Wrinkly skin syndrome, 278250 (3), Autosomal recessive
ATP6V0A4	605239	Renal tubular acidosis, distal, autosomal recessive, 602722 (3)
ATP6V1A	607027	Cutis laxa, autosomal recessive, type IID, 617403 (3), Autosomal recessive; Epileptic encephalopathy, infantile or early childhood, 3, 618012 (3), Autosomal dominant
ATP6V1B1	192132	Renal tubular acidosis with deafness, 267300 (3), Autosomal recessive
ATP6V1B2	606939	Deafness, congenital, with onychodystrophy, autosomal dominant, 124480 (3), Autosomal dominant; Zimmermann-Laband syndrome 2, 616455 (3), Autosomal dominant
ATP6V1E1	108746	Cutis laxa, autosomal recessive, type IIC, 617402 (3), Autosomal recessive
ATP7A	300011	Menkes disease, 309400 (3), X-linked recessive; Occipital horn syndrome, 304150 (3), X-linked recessive; Spinal muscular atrophy, distal, X-linked 3, 300489 (3), X-linked recessive
ATP7B	606882	Wilson disease, 277900 (3), Autosomal recessive
ATP8A2	605870	?Cerebellar ataxia, mental retardation, and dysequilibrium syndrome 4, 615268 (3), Autosomal recessive
ATP8B1	602397	Cholestasis, benign recurrent intrahepatic, 243300 (3), Autosomal recessive; Cholestasis, intrahepatic, of pregnancy, 1, 147480 (3), Autosomal dominant; Cholestasis, progressive familial intrahepatic 1, 211600 (3), Autosomal recessive
ATPAF2	608918	?Mitochondrial complex V (ATP synthase) deficiency, nuclear type 1, 604273 (3), Autosomal recessive
ATR	601215	?Cutaneous telangiectasia and cancer syndrome, familial, 614564 (3), Autosomal dominant; Seckel syndrome 1, 210600 (3), Autosomal recessive
ATRX	300032	Alpha-thalassemia myelodysplasia syndrome, somatic, 300448 (3); Alpha-thalassemia/mental retardation syndrome, 301040 (3), X-linked dominant; Mental retardation-hypotonic facies syndrome, X-linked, 309580 (3), X-linked recessive
ATXN1	601556	Spinocerebellar ataxia 1, 164400 (3), Autosomal dominant
ATXN10	611150	Spinocerebellar ataxia 10, 603516 (3), Autosomal dominant
ATXN2	601517	{Amyotrophic lateral sclerosis, susceptibility to, 13}, 183090 (3), Autosomal dominant; {Parkinson disease, late-onset, susceptibility to}, 168600 (3), Isolated cases, Multifactorial; Spinocerebellar ataxia 2, 183090 (3), Autosomal dominant
ATXN3	607047	Machado-Joseph disease, 109150 (3), Autosomal dominant
ATXN7	607640	Spinocerebellar ataxia 7, 164500 (3), Autosomal dominant
AUH	600529	3-methylglutaconic aciduria, type I, 250950 (3), Autosomal recessive
AURKC	603495	Spermatogenic failure 5, 243060 (3), Autosomal recessive

AUTS2	607270	Mental retardation, autosomal dominant 26, 615834 (3), Autosomal dominant
AVP	192340	Diabetes insipidus, neurohypophyseal, 125700 (3), Autosomal dominant
AVPR2	300538	Diabetes insipidus, nephrogenic, 304800 (3), X-linked recessive; Nephrogenic syndrome of inappropriate antidiuresis, 300539 (3), X-linked recessive
AXIN1	603816	?Caudal duplication anomaly, 607864 (3); Hepatocellular carcinoma, somatic, 114550 (3)
AXIN2	604025	Colorectal cancer, somatic, 114500 (3); Oligodontia-colorectal cancer syndrome, 608615 (3), Autosomal dominant
AXL	109135	No OMIM phenotype
B3GALNT2	610194	Muscular dystrophy-dystroglycanopathy (congenital with brain and eye anomalies, type A, 11, 615181 (3), Autosomal recessive
B3GALT6	615291	Ehlers-Danlos syndrome, spondylodysplastic type, 2, 615349 (3), Autosomal recessive; Spondyloepimetaphyseal dysplasia with joint laxity, type 1, with or without fractures, 271640 (3), Autosomal recessive
B3GAT3	606374	Multiple joint dislocations, short stature, craniofacial dysmorphism, with or without congenital heart defects, 245600 (3), Autosomal recessive
B3GLCT	610308	Peters-plus syndrome, 261540 (3), Autosomal recessive
B4GALT1	137060	Congenital disorder of glycosylation, type IId, 607091 (3), Autosomal recessive
B4GALT7	604327	Ehlers-Danlos syndrome, spondylodysplastic type, 1, 130070 (3), Autosomal recessive
B4GAT1	605517	Muscular dystrophy-dystroglycanopathy (congenital with brain and eye anomalies), type A, 13, 615287 (3), Autosomal recessive
B9D1	614144	Joubert syndrome 27, 617120 (3), Autosomal recessive; ?Meckel syndrome 9, 614209 (3), Autosomal recessive
B9D2	611951	Joubert syndrome 34, 614175 (3), Autosomal recessive; ?Meckel syndrome 10, 614175 (3), Autosomal recessive
BAAT	602938	Hypercholanemia, familial, 607748 (3)
BAG3	603883	Cardiomyopathy, dilated, 1HH, 613881 (3), Autosomal dominant; Myopathy, myofibrillar, 6, 612954 (3), Autosomal dominant
BANF1	603811	Nestor-Guillermo progeria syndrome, 614008 (3), Autosomal recessive
BANK1	610292	No OMIM phenotype
BAP1	603089	Tumor predisposition syndrome, 614327 (3), Autosomal dominant
BARD1	601593	{Breast cancer, susceptibility to}, 114480 (3), Autosomal dominant
BBIP1	613605	?Bardet-Biedl syndrome 18, 615995 (3), Autosomal recessive
BBS1	209901	Bardet-Biedl syndrome 1, 209900 (3), Autosomal recessive, Digenic recessive
BBS10	610148	Bardet-Biedl syndrome 10, 615987 (3), Autosomal recessive
BBS12	610683	Bardet-Biedl syndrome 12, 615989 (3), Autosomal recessive

BBS2	606151	Bardet-Biedl syndrome 2, 615981 (3), Autosomal recessive; Retinitis pigmentosa 74, 616562 (3), Autosomal recessive
BBS4	600374	Bardet-Biedl syndrome 4, 615982 (3), Autosomal recessive
BBS5	603650	Bardet-Biedl syndrome 5, 615983 (3), Autosomal recessive
BBS7	607590	Bardet-Biedl syndrome 7, 615984 (3), Autosomal recessive
BBS9	607968	Bardet-Biedl syndrome 9, 615986 (3), Autosomal recessive
BCAM	612773	[Blood group, Auberger system], 111200 (3); [Blood group, Lutheran null], 247420 (3), Autosomal recessive; [Blood group, Lutheran system], 111200 (3)
BCAN	600347	No OMIM phenotype
BCAP31	300398	Deafness, dystonia, and cerebral hypomyelination, 300475 (3), X-linked recessive
BCKDHA	608348	Maple syrup urine disease, type Ia, 248600 (3), Autosomal recessive
BCKDHB	248611	Maple syrup urine disease, type Ib, 248600 (3), Autosomal recessive
BCKDK	614901	Branched-chain ketoacid dehydrogenase kinase deficiency, 614923 (3)
BCL10	603517	?Immunodeficiency 37, 616098 (3), Autosomal recessive; Lymphoma, MALT, somatic, 137245 (3); {Lymphoma, follicular, somatic}, 605027 (3); {Male germ cell tumor, somatic},, 273300 (3); {Mesothelioma, somatic}, 156240 (3); {Sezary syndrome, somatic} (3)
BCL11B	606558	?Immunodeficiency 49, 617237 (3), Autosomal dominant
BCO1	605748	?Hypercarotenemia and vitamin A deficiency, autosomal dominant, 115300 (3), Autosomal dominant
BCOR	300485	Microphthalmia, syndromic 2, 300166 (3), X-linked dominant
BCR	151410	Leukemia, acute lymphocytic, somatic, 613065 (3); Leukemia, chronic myeloid, somatic, 608232 (3)
BCS1L	603647	Bjornstad syndrome, 262000 (3), Autosomal recessive; GRACILE syndrome, 603358 (3); Leigh syndrome, 256000 (3), Autosomal recessive, Mitochondrial; Mitochondrial complex III deficiency, nuclear type 1, 124000 (3), Autosomal recessive
BDNF	113505	No OMIM phenotype
BEAN1	612051	Spinocerebellar ataxia 31, 117210 (3), Autosomal dominant
BEST1	607854	Bestrophinopathy, autosomal recessive, 611809 (3); Macular dystrophy, vitelliform, 2, 153700 (3), Autosomal dominant; Microcornea, rod-cone dystrophy, cataract, and posterior staphyloma, 193220 (3), Autosomal dominant; Retinitis pigmentosa, concentric, 613194 (3); Retinitis pigmentosa-50, 613194 (3); Vitreoretinochoroidopathy, 193220 (3), Autosomal dominant
BFSP1	603307	Cataract 33, multiple types, 611391 (3), Autosomal recessive, Autosomal dominant
BFSP2	603212	Cataract 12, multiple types, 611597 (3), Autosomal dominant
BGN	301870	Meester-Loeys syndrome, 300989 (3), X-linked; Spondyloepimetaphyseal dysplasia, X-linked, 300106 (3), X-linked recessive

BHLHA9	615416	?Camptosynpolydactyly, complex, 607539 (3), Autosomal recessive; Syndactyly, mesoaxial synostotic, with phalangeal reduction, 609432 (3), Autosomal recessive
BHLHE41	606200	[Short sleeper], 612975 (3), Autosomal dominant
BICC1	614295	{Renal dysplasia, cystic, susceptibility to}, 601331 (3), Autosomal dominant
BICD2	609797	Spinal muscular atrophy, lower extremity-predominant, 2, AD, 615290 (3), Autosomal dominant
BIN1	601248	Centronuclear myopathy 2, 255200 (3), Autosomal recessive
BLK	191305	Maturity-onset diabetes of the young, type 11, 613375 (3), Autosomal dominant
BLM	604610	Bloom syndrome, 210900 (3), Autosomal recessive
BLMH	602403	No OMIM phenotype
BLNK	604515	?Agammaglobulinemia 4, 613502 (3), Autosomal recessive
BLOC1S3	609762	Hermansky-Pudlak syndrome 8, 614077 (3), Autosomal recessive
BLOC1S6	604310	?Hermansky-pudlak syndrome 9, 614171 (3), Autosomal recessive
BLVRA	109750	Hyperbiliverdinemia, 614156 (3), Autosomal recessive, Autosomal dominant
BMP1	112264	Osteogenesis imperfecta, type XIII, 614856 (3), Autosomal recessive
BMP15	300247	Ovarian dysgenesis 2, 300510 (3), X-linked; Premature ovarian failure 4, 300510 (3), X-linked
BMP2	112261	Brachydactyly, type A2, 112600 (3), Autosomal dominant; {HFE hemochromatosis, modifier of}, 235200 (3), Autosomal recessive; Short stature, facial dysmorphism, and skeletal anomalies with or without cardiac anomalies, 617877 (3), Autosomal dominant
BMPER	608699	Diaphanospondylodysostosis, 608022 (3), Autosomal recessive
BMPR1A	601299	Juvenile polyposis syndrome, infantile form, 174900 (3), Autosomal dominant; Polyposis syndrome, hereditary mixed, 2, 610069 (3); Polyposis, juvenile intestinal, 174900 (3), Autosomal dominant
BMPR2	600799	Pulmonary hypertension, familial primary, 1, with or without HHT, 178600 (3), Autosomal dominant; Pulmonary hypertension, primary, fenfluramine or dexfenfluramine-associated, 178600 (3), Autosomal dominant; Pulmonary venoocclusive disease 1, 265450 (3), Autosomal dominant
BMS1	611448	?Aplasia cutis congenita, nonsyndromic, 107600 (3), Autosomal dominant
BOLA3	613183	Multiple mitochondrial dysfunctions syndrome 2 with hyperglycinemia, 614299 (3), Autosomal recessive
BPGM	613896	Erythrocytosis due to bisphosphoglycerate mutase deficiency, 222800 (3), Autosomal recessive
BPY2	400013	No OMIM phenotype

BRAF	164757	Adenocarcinoma of lung, somatic, 211980 (3); Cardiofaciocutaneous syndrome, 115150 (3), Autosomal dominant; Colorectal cancer, somatic (3); LEOPARD syndrome 3, 613707 (3), Autosomal dominant; Melanoma, malignant, somatic (3); Nonsmall cell lung cancer, somatic (3); Noonan syndrome 7, 613706 (3), Autosomal dominant
BRAT1	614506	Rigidity and multifocal seizure syndrome, lethal neonatal, 614498 (3), Autosomal recessive
BRCA1	113705	{Breast-ovarian cancer, familial, 1}, 604370 (3), Autosomal dominant, Multifactorial; Fanconi anemia, complementation group S, 617883 (3), Autosomal recessive; {Pancreatic cancer, susceptibility to, 4}, 614320 (3)
BRCA2	600185	{Breast cancer, male, susceptibility to}, 114480 (3), Autosomal dominant; {Breast-ovarian cancer, familial, 2}, 612555 (3), Autosomal dominant; Fanconi anemia, complementation group D1, 605724 (3), Autosomal recessive; {Glioblastoma 3}, 613029 (3), Autosomal recessive; {Medulloblastoma}, 155255 (3), Autosomal recessive, Autosomal dominant; {Pancreatic cancer 2}, 613347 (3); {Prostate cancer}, 176807 (3), Autosomal dominant; Wilms tumor, 194070 (3), Autosomal dominant, Somatic mutation
BRF1	604902	Cerebellofaciodental syndrome, 616202 (3), Autosomal recessive
BRIP1	605882	Breast cancer, early-onset, 114480 (3), Autosomal dominant; Fanconi anemia, complementation group J, 609054 (3)
BRPF1	602410	Intellectual developmental disorder with dysmorphic facies and ptosis, 617333 (3), Autosomal dominant
BRWD3	300553	Mental retardation, X-linked 93, 300659 (3), X-linked recessive
BSCL2	606158	Encephalopathy, progressive, with or without lipodystrophy, 615924 (3), Autosomal recessive; Lipodystrophy, congenital generalized, type 2, 269700 (3), Autosomal recessive; Neuropathy, distal hereditary motor, type VA, 600794 (3), Autosomal dominant; Silver spastic paraplegia syndrome, 270685 (3), Autosomal dominant
BSG	109480	[Blood group, OK], 111380 (3)
BSND	606412	Bartter syndrome, type 4a, 602522 (3), Autosomal recessive; Sensorineural deafness with mild renal dysfunction, 602522 (3), Autosomal recessive
BTD	609019	Biotinidase deficiency, 253260 (3), Autosomal recessive
BTK	300300	Agammaglobulinemia and isolated hormone deficiency, 307200 (3), X-linked recessive; Agammaglobulinemia, X-linked 1, 300755 (3), X-linked recessive
BTNL2	606000	{Sarcoidosis, susceptibility to, 2}, 612387 (3), Autosomal dominant
BUB1B	602860	Colorectal cancer, somatic, 114500 (3); Mosaic variegated aneuploidy syndrome 1, 257300 (3), Autosomal recessive; [Premature chromatid separation trait], 176430 (3), Autosomal dominant
BVES	604577	?Muscular dystrophy, limb-girdle, type 2X, 616812 (3), Autosomal recessive

C12orf65	613541	Combined oxidative phosphorylation deficiency 7, 613559 (3), Autosomal recessive; Spastic paraplegia 55, autosomal recessive, 615035 (3), Autosomal recessive
C15orf41	615626	Dyserythropoietic anemia, congenital, type Ib, 615631 (3), Autosomal recessive
C19orf12	614297	Neurodegeneration with brain iron accumulation 4, 614298 (3), Autosomal recessive; ?Spastic paraplegia 43, autosomal recessive, 615043 (3), Autosomal recessive
C1GALT1C1	300611	Tn polyagglutination syndrome, somatic, 300622 (3)
C1QA	120550	C1q deficiency, 613652 (3), Autosomal recessive
C1QB	120570	C1q deficiency, 613652 (3), Autosomal recessive
C1QC	120575	C1q deficiency, 613652 (3), Autosomal recessive
C1R	613785	Ehlers-Danlos syndrome, periodontal type, 1, 130080 (3), Autosomal dominant
C1S	120580	C1s deficiency, 613783 (3); Ehlers-Danlos syndrome, periodontal type, 2, 617174 (3), Autosomal dominant
C2	613927	C2 deficiency, 217000 (3), Autosomal recessive; {Macular degeneration, age-related, 14, reduced risk of}, 615489 (3)
C2CD3	615944	?Orofaciodigital syndrome XIV, 615948 (3), Autosomal recessive
C3	120700	C3 deficiency, 613779 (3), Autosomal recessive; {Hemolytic uremic syndrome, atypical, susceptibility to, 5}, 612925 (3), Autosomal dominant; {Macular degeneration, age-related, 9}, 611378 (3)
C4A	120810	[Blood group, Rodgers], 614374 (3); C4a deficiency, 614380 (3), Autosomal recessive
C4B	120820	C4B deficiency, 614379 (3)
C5	120900	C5 deficiency, 609536 (3); [Eculizumab, poor response to], 615749 (3), Autosomal dominant
C7	217070	C7 deficiency, 610102 (3)
C8A	120950	C8 deficiency, type I, 613790 (3), Autosomal recessive
C8B	120960	C8 deficiency, type II, 613789 (3), Autosomal recessive
C8orf37	614477	Bardet-Biedl syndrome 21, 617406 (3), Autosomal recessive; Cone-rod dystrophy 16, 614500 (3), Autosomal recessive; Retinitis pigmentosa 64, 614500 (3), Autosomal recessive
C9	120940	C9 deficiency, 613825 (3); {Macular degeneration, age-related, 15, susceptibility to}, 615591 (3)
C9orf72	614260	Frontotemporal dementia and/or amyotrophic lateral sclerosis 1, 105550 (3), Autosomal dominant
CA12	603263	Hyperchlorhidrosis, isolated, 143860 (3), Autosomal recessive
CA2	611492	Osteopetrosis, autosomal recessive 3, with renal tubular acidosis, 259730 (3), Autosomal recessive
CA4	114760	Retinitis pigmentosa 17, 600852 (3), Autosomal dominant
CA5A	114761	Hyperammonemia due to carbonic anhydrase VA deficiency, 615751 (3), Autosomal recessive

CA8	114815	Cerebellar ataxia and mental retardation with or without quadrupedal locomotion 3, 613227 (3), Autosomal recessive
CABP2	607314	Deafness, autosomal recessive 93, 614899 (3), Autosomal recessive
CACNA1A	601011	Epileptic encephalopathy, early infantile, 42, 617106 (3), Autosomal dominant; Episodic ataxia, type 2, 108500 (3), Autosomal dominant; Migraine, familial hemiplegic, 1, 141500 (3), Autosomal dominant; Migraine, familial hemiplegic, 1, with progressive cerebellar ataxia, 141500 (3), Autosomal dominant; Spinocerebellar ataxia 6, 183086 (3), Autosomal dominant
CACNA1B	601012	?Dystonia 23, 614860 (3), Autosomal dominant
CACNA1D	114206	Primary aldosteronism, seizures, and neurologic abnormalities, 615474 (3), Autosomal dominant; Sinoatrial node dysfunction and deafness, 614896 (3), Autosomal recessive
CACNA1F	300110	Aland Island eye disease, 300600 (3), X-linked; Cone-rod dystrophy, X-linked, 3, 300476 (3), X-linked recessive; Night blindness, congenital stationary (incomplete), 2A, X-linked, 300071 (3), X-linked
CACNA1G	604065	Spinocerebellar ataxia 42, 616795 (3), Autosomal dominant
CACNA1H	607904	{Epilepsy, childhood absence, susceptibility to, 6}, 611942 (3); {Epilepsy, idiopathic generalized, susceptibility to, 6}, 611942 (3); Hyperaldosteronism, familial, type IV, 617027 (3), Autosomal dominant
CACNA1S	114208	Hypokalemic periodic paralysis, type 1, 170400 (3), Autosomal dominant; {Malignant hyperthermia susceptibility 5}, 601887 (3), Autosomal dominant; {Thyrotoxic periodic paralysis, susceptibility to, 1}, 188580 (3), Isolated cases
CACNA2D4	608171	Retinal cone dystrophy 4, 610478 (3), Autosomal recessive
CACNB2	600003	Brugada syndrome 4, 611876 (3)
CACNG2	602911	?Mental retardation, autosomal dominant 10, 614256 (3)
CAD	114010	Epileptic encephalopathy, early infantile, 50, 616457 (3), Autosomal recessive
CALCR	114131	{Osteoporosis, postmenopausal, susceptibility}, 166710 (3), Autosomal dominant
CALM1	114180	Long QT syndrome 14, 616247 (3), Autosomal dominant; Ventricular tachycardia, catecholaminergic polymorphic, 4, 614916 (3), Autosomal dominant
CALM2	114182	Long QT syndrome 15, 616249 (3), Autosomal dominant
CALR	109091	Myelofibrosis, somatic, 254450 (3); Thrombocythemia, somatic, 187950 (3)
CALR3	611414	?Cardiomyopathy, hypertrophic, 19, 613875 (3), Autosomal dominant
CAMTA1	611501	Cerebellar ataxia, nonprogressive, with mental retardation, 614756 (3), Autosomal dominant
CANT1	613165	Desbuquois dysplasia 1, 251450 (3), Autosomal recessive; Epiphyseal dysplasia, multiple, 7, 617719 (3), Autosomal recessive
CAPN1	114220	Spastic paraplegia 76, autosomal recessive, 616907 (3), Autosomal recessive

CAPN10	605286	{Diabetes mellitus, noninsulin-dependent 1}, 601283 (3)
CAPN3	114240	Muscular dystrophy, limb-girdle, type 2A, 253600 (3), Autosomal recessive
CARD11	607210	B-cell expansion with NFKB and T-cell anergy, 616452 (3), Autosomal dominant; Immunodeficiency 11A, 615206 (3), Autosomal recessive; Immunodeficiency 11B with atopic dermatitis, 617638 (3), Autosomal dominant
CARD14	607211	Pityriasis rubra pilaris, 173200 (3), Autosomal dominant; Psoriasis 2, 602723 (3), Autosomal dominant
CARD9	607212	Candidiasis, familial, 2, autosomal recessive, 212050 (3), Autosomal recessive
CARS2	612800	Combined oxidative phosphorylation deficiency 27, 616672 (3), Autosomal recessive
CARTPT	602606	{?Obesity, susceptibility to}, 601665 (3), Autosomal recessive, Autosomal dominant, Multifactorial
CASK	300172	FG syndrome 4, 300422 (3); Mental retardation and microcephaly with pontine and cerebellar hypoplasia, 300749 (3), X-linked dominant; Mental retardation, with or without nystagmus, 300422 (3)
CASP10	601762	Autoimmune lymphoproliferative syndrome, type II, 603909 (3), Autosomal dominant; Gastric cancer, somatic, 613659 (3); Lymphoma, non-Hodgkin, somatic, 605027 (3)
CASP14	605848	Ichthyosis, congenital, autosomal recessive 12, 617320 (3), Autosomal recessive
CASP8	601763	?Autoimmune lymphoproliferative syndrome, type IIB, 607271 (3), Autosomal recessive; {Breast cancer, protection against}, 114480 (3), Autosomal dominant; Hepatocellular carcinoma, somatic, 114550 (3); {Lung cancer, protection against}, 211980 (3), Autosomal recessive
CASQ1	114250	Myopathy, vacuolar, with CASQ1 aggregates, 616231 (3), Autosomal dominant
CASQ2	114251	Ventricular tachycardia, catecholaminergic polymorphic, 2, 611938 (3), Autosomal recessive
CASR	601199	{Calcium, serum level of} (3); {Epilepsy idiopathic generalized, susceptibility to, 8}, 612899 (3); Hypercalciuric hypercalcemia (3); Hyperparathyroidism, neonatal, 239200 (3), Autosomal recessive, Autosomal dominant; Hypocalcemia, autosomal dominant, 601198 (3), Autosomal dominant; Hypocalcemia, autosomal dominant, with Bartter syndrome, 601198 (3), Autosomal dominant; Hypocalciuric hypercalcemia, type I, 145980 (3), Autosomal dominant
CAT	115500	Acatalasemia, 614097 (3)
CATSPER1	606389	Spermatogenic failure 7, 612997 (3), Autosomal recessive
CATSPER2	607249	No OMIM phenotype

CAV1	601047	?Lipodystrophy, congenital generalized, type 3, 612526 (3); ?Partial lipodystrophy, congenital cataracts, and neurodegeneration syndrome, 606721 (3), Autosomal dominant; Pulmonary hypertension, primary, 3, 615343 (3), Autosomal dominant
CAV3	601253	Cardiomyopathy, familial hypertrophic, 192600 (3), Autosomal dominant; Creatine phosphokinase, elevated serum, 123320 (3), Autosomal dominant; Long QT syndrome 9, 611818 (3), Autosomal dominant; Muscular dystrophy, limb-girdle, type IC, 607801 (3), Autosomal recessive, Autosomal dominant; Myopathy, distal, Tateyama type, 614321 (3), Autosomal dominant; Rippling muscle disease, 606072 (3), Autosomal dominant
CAVIN1	603198	Lipodystrophy, congenital generalized, type 4, 613327 (3), Autosomal recessive
CBL	165360	?Juvenile myelomonocytic leukemia, 607785 (3), Autosomal dominant, Somatic mutation; Noonan syndrome-like disorder with or without juvenile myelomonocytic leukemia, 613563 (3), Autosomal dominant
CBS	613381	Homocystinuria, B6-responsive and nonresponsive types, 236200 (3), Autosomal recessive; Thrombosis, hyperhomocysteinemic, 236200 (3), Autosomal recessive
CBX2	602770	?46XY sex reversal 5, 613080 (3), Autosomal recessive
CC2D1A	610055	Mental retardation, autosomal recessive 3, 608443 (3), Autosomal recessive
CC2D2A	612013	COACH syndrome, 216360 (3), Autosomal recessive; Joubert syndrome 9, 612285 (3), Autosomal recessive; Meckel syndrome 6, 612284 (3), Autosomal recessive
CCBE1	612753	Hennekam lymphangiectasia-lymphedema syndrome 1, 235510 (3), Autosomal recessive
CCDC114	615038	Ciliary dyskinesia, primary, 20, 615067 (3), Autosomal recessive
CCDC115	613734	Congenital disorder of glycosylation, type IIo, 616828 (3), Autosomal recessive
CCDC141	616031	No OMIM phenotype
CCDC151	615956	Ciliary dyskinesia, primary, 30, 616037 (3), Autosomal recessive
CCDC174	616735	Hypotonia, infantile, with psychomotor retardation, 616816 (3), Autosomal recessive
CCDC22	300859	Ritscher-Schinzel syndrome 2, 300963 (3), X-linked recessive
CCDC39	613798	Ciliary dyskinesia, primary, 14, 613807 (3)
CCDC40	613799	Ciliary dyskinesia, primary, 15, 613808 (3)
CCDC50	611051	?Deafness, autosomal dominant 44, 607453 (3), Autosomal dominant
CCDC6	601985	No OMIM phenotype
CCDC65	611088	Ciliary dyskinesia, primary, 27, 615504 (3), Autosomal recessive
CCDC78	614666	?Centronuclear myopathy 4, 614807 (3), Autosomal dominant
CCDC8	614145	3-M syndrome 3, 614205 (3), Autosomal recessive
CCDC88A	609736	?PEHO syndrome-like, 617507 (3), Autosomal recessive

CCDC88C	611204	Hydrocephalus, nonsyndromic, autosomal recessive, 236600 (3), Autosomal recessive; ?Spinocerebellar ataxia 40, 616053 (3), Autosomal dominant
CCL11	601156	{Asthma, susceptibility to}, 600807 (3), Autosomal dominant; {HIV1, resistance to}, 609423 (3)
CCL2	158105	{Coronary artery disease, modifier of} (3); {HIV-1, resistance to}, 609423 (3); {Mycobacterium tuberculosis, susceptibility to}, 607948 (3); {Spina bifida, susceptibility to}, 182940 (3), Autosomal dominant
CCL3	182283	{HIV infection, resistance to}, 609423 (2)
CCM2	607929	Cerebral cavernous malformations-2, 603284 (3), Autosomal dominant
CCND1	168461	{Colorectal cancer, susceptibility to}, 114500 (3), Autosomal dominant; {Multiple myeloma, susceptibility to}, 254500 (3), Somatic mutation; {von Hippel-Lindau syndrome, modifier of}, 193300 (3), Autosomal dominant
CCND2	123833	Megalencephaly-polymicrogyria-polydactyly-hydrocephalus syndrome 3, 615938 (3), Autosomal dominant
CCNO	607752	Ciliary dyskinesia, primary, 29, 615872 (3), Autosomal recessive
CCNQ (FAM58A)	300708	STAR syndrome, 300707 (3), X-linked dominant
CCR5	601373	{Diabetes mellitus, insulin-dependent, 22}, 612522 (3); {HIV infection, susceptibility/resistance to} (3); {Hepatitis C virus, resistance to}, 609532 (3); {West nile virus, susceptibility to}, 610379 (3)
CCT5	610150	Neuropathy, hereditary sensory, with spastic paraplegia, 256840 (3), Autosomal recessive
CD151	602243	[Blood group, Raph], 179620 (3); Nephropathy with pretibial epidermolysis bullosa and deafness, 609057 (3)
CD164	603356	?Deafness, autosomal dominant 66, 616969 (3), Autosomal dominant
CD19	107265	Immunodeficiency, common variable, 3, 613493 (3), Autosomal recessive
CD207	604862	[?Birbeck granule deficiency], 613393 (3)
CD209	604672	{Dengue fever, protection against}, 614371 (3); {HIV type 1, susceptibility to}, 609423 (3); {Mycobacterium tuberculosis, susceptibility to}, 607948 (3)
CD244	605554	{Rheumatoid arthritis, susceptibility to}, 180300 (3)
CD247	186780	?Immunodeficiency 25, 610163 (3), Autosomal recessive
CD27	186711	Lymphoproliferative syndrome 2, 615122 (3), Autosomal recessive
CD2AP	604241	Glomerulosclerosis, focal segmental, 3, 607832 (3)
CD320	606475	Methylmalonic aciduria, transient, due to transcobalamin receptor defect, 613646 (3)
CD3D	186790	Immunodeficiency 19, 615617 (3), Autosomal recessive
CD3E	186830	Immunodeficiency 18, 615615 (3), Autosomal recessive; Immunodeficiency 18, SCID variant, 615615 (3), Autosomal recessive
CD3G	186740	Immunodeficiency 17, CD3 gamma deficient, 615607 (3), Autosomal recessive
CD4	186940	OKT4 epitope deficiency, 613949 (3)

CD40	109535	Immunodeficiency with hyper-IgM, type 3, 606843 (3), Autosomal recessive
CD40LG	300386	Immunodeficiency, X-linked, with hyper-IgM, 308230 (3), X-linked recessive
CD44	107269	[Blood group, Indian system], 609027 (3)
CD46	120920	{Hemolytic uremic syndrome, atypical, susceptibility to, 2}, 612922 (3), Autosomal recessive, Autosomal dominant
CD55	125240	[Blood group Cromer], 613793 (3), Autosomal recessive; Complement hyperactivation, angiopathic thrombosis, and protein-losing enteropathy, 226300 (3), Autosomal recessive
CD59	107271	Hemolytic anemia, CD59-mediated, with or without immune-mediated polyneuropathy, 612300 (3), Autosomal recessive
CD79A	112205	Agammaglobulinemia 3, 613501 (3), Autosomal recessive
CD79B	147245	Agammaglobulinemia 6, 612692 (3), Autosomal recessive
CD81	186845	Immunodeficiency, common variable, 6, 613496 (3), Autosomal recessive
CD82	600623	{Prostate cancer, susceptibility to}, 176807 (2), Autosomal dominant
CD8A	186910	CD8 deficiency, familial, 608957 (3), Autosomal recessive
CD96	606037	C syndrome, 211750 (3), Autosomal dominant
CDAN1	607465	Dyserythropoietic anemia, congenital, type Ia, 224120 (3), Autosomal recessive
CDC14A	603504	Deafness, autosomal recessive 105, 616958 (3), Autosomal recessive
CDC42	116952	Takenouchi-Kosaki syndrome, 616737 (3), Autosomal dominant
CDC45	603465	Meier-Gorlin syndrome 7, 617063 (3), Autosomal recessive
CDC6	602627	?Meier-Gorlin syndrome 5, 613805 (3), Autosomal recessive
CDC73	607393	Hyperparathyroidism, familial primary, 145000 (3), Autosomal dominant; Hyperparathyroidism-jaw tumor syndrome, 145001 (3), Autosomal dominant; Parathyroid adenoma with cystic changes, 145001 (3), Autosomal dominant; Parathyroid carcinoma, 608266 (3)
CDCA7	609937	Immunodeficiency-centromeric instability-facial anomalies syndrome 3, 616910 (3), Autosomal recessive
CDH1	192090	Blepharocheilodontic syndrome 1, 119580 (3), Autosomal dominant; {Breast cancer, lobular}, 114480 (3), Autosomal dominant; Endometrial carcinoma, somatic, 608089 (3); Gastric cancer, familial diffuse, with or without cleft lip and/or palate, 137215 (3), Autosomal dominant; Ovarian carcinoma, somatic, 167000 (3); {Prostate cancer, susceptibility to}, 176807 (3), Autosomal dominant
CDH15	114019	Mental retardation, autosomal dominant 3, 612580 (3)
CDH23	605516	Deafness, autosomal recessive 12, 601386 (3), Autosomal recessive; {Pituitary adenoma 5, multiple types}, 617540 (3), Autosomal dominant; Usher syndrome, type 1D, 601067 (3), Autosomal recessive, Digenic recessive; Usher syndrome, type 1D/F digenic, 601067 (3), Autosomal recessive, Digenic recessive

CDH3	114021	Ectodermal dysplasia, ectrodactyly, and macular dystrophy, 225280 (3), Autosomal recessive; Hypotrichosis, congenital, with juvenile macular dystrophy, 601553 (3), Autosomal recessive
CDHR1	609502	Cone-rod dystrophy 15, 613660 (3), Autosomal recessive; Retinitis pigmentosa 65, 613660 (3), Autosomal recessive
CDK13	603309	Congenital heart defects, dysmorphic facial features, and intellectual developmental disorder, 617360 (3), Autosomal dominant
CDK4	123829	{Melanoma, cutaneous malignant, 3}, 609048 (3), Autosomal dominant
CDK5	123831	?Lissencephaly 7 with cerebellar hypoplasia, 616342 (3), Autosomal recessive
CDK5RAP2	608201	Microcephaly 3, primary, autosomal recessive, 604804 (3), Autosomal recessive
CDK6	603368	?Microcephaly 12, primary, autosomal recessive, 616080 (3), Autosomal recessive
CDKAL1	611259	No OMIM phenotype
CDKL5	300203	Epileptic encephalopathy, early infantile, 2, 300672 (3), X-linked dominant
CDKN1B	600778	Multiple endocrine neoplasia, type IV, 610755 (3), Autosomal dominant
CDKN1C	600856	Beckwith-Wiedemann syndrome, 130650 (3), Autosomal dominant; IMAGE syndrome, 614732 (3), Autosomal dominant
CDKN2A	600160	Melanoma and neural system tumor syndrome, 155755 (3), Autosomal dominant; {Melanoma, cutaneous malignant, 2}, 155601 (3), Autosomal dominant; Orolaryngeal cancer, multiple (3); Pancreatic cancer/melanoma syndrome, 606719 (3), Autosomal dominant
CDON	608707	Holoprosencephaly 11, 614226 (3), Autosomal dominant, Isolated cases
CDSN	602593	Hypotrichosis 2, 146520 (3), Autosomal dominant; Peeling skin syndrome 1, 270300 (3), Autosomal recessive
CDT1	605525	Meier-Gorlin syndrome 4, 613804 (3), Autosomal recessive
CDY1	400016	No OMIM phenotype
CDY2A	400018	No OMIM phenotype
CEACAM16	614591	Deafness, autosomal dominant 4B, 614614 (3), Autosomal dominant
CEBPA	116897	?Leukemia, acute myeloid, 601626 (3), Autosomal dominant; Leukemia, acute myeloid, somatic, 601626 (3)
CEBPE	600749	Specific granule deficiency, 245480 (3), Autosomal recessive
CEL	114840	Maturity-onset diabetes of the young, type VIII, 609812 (3), Autosomal dominant
CENPE	117143	?Microcephaly 13, primary, autosomal recessive, 616051 (3), Autosomal recessive
CENPF	600236	Stromme syndrome, 243605 (3), Autosomal recessive
CEP104	616690	Joubert syndrome 25, 616781 (3), Autosomal recessive

CEP120	613446	Joubert syndrome 31, 617761 (3), Autosomal recessive; Short-rib thoracic dysplasia 13 with or without polydactyly, 616300 (3), Autosomal recessive
CEP135	611423	Microcephaly 8, primary, autosomal recessive, 614673 (3), Autosomal recessive
CEP152	613529	Microcephaly 9, primary, autosomal recessive, 614852 (3), Autosomal recessive; Seckel syndrome 5, 613823 (3), Autosomal recessive
CEP164	614848	Nephronophthisis 15, 614845 (3), Autosomal recessive
CEP19	615586	Morbid obesity and spermatogenic failure, 615703 (3), Autosomal recessive
CEP290	610142	?Bardet-Biedl syndrome 14, 615991 (3), Autosomal recessive; Joubert syndrome 5, 610188 (3), Autosomal recessive; Leber congenital amaurosis 10, 611755 (3); Meckel syndrome 4, 611134 (3), Autosomal recessive; Senior-Loken syndrome 6, 610189 (3), Autosomal recessive
CEP41	610523	Joubert syndrome 15, 614464 (3), Autosomal recessive
CEP78	617110	Cone-rod dystrophy and hearing loss, 617236 (3), Autosomal recessive
CEP83	615847	Nephronophthisis 18, 615862 (3), Autosomal recessive
CERKL	608381	Retinitis pigmentosa 26, 608380 (3)
CERS1	606919	?Epilepsy, progressive myoclonic, 8, 616230 (3), Autosomal recessive
CETP	118470	[High density lipoprotein cholesterol level QTL 10], 143470 (3), Autosomal dominant; Hyperalphalipoproteinemia, 143470 (3), Autosomal dominant
CFAP298 (C21orf59)	615494	Ciliary dyskinesia, primary, 26, 615500 (3), Autosomal recessive
CFAP53	614759	Heterotaxy, visceral, 6, autosomal recessive, 614779 (3), Autosomal recessive
CFB	138470	?Complement factor B deficiency, 615561 (3); {Hemolytic uremic syndrome, atypical, susceptibility to, 4}, 612924 (3), Autosomal dominant; {Macular degeneration, age-related, 14, reduced risk of}, 615489 (3)
CFC1	605194	Heterotaxy, visceral, 2, autosomal, 605376 (3), Autosomal dominant
CFD	134350	Complement factor D deficiency, 613912 (3), Autosomal recessive
CFH	134370	Basal laminar drusen, 126700 (3), Autosomal dominant; Complement factor H deficiency, 609814 (3), Autosomal recessive, Autosomal dominant; {Hemolytic uremic syndrome, atypical, susceptibility to, 1}, 235400 (3), Autosomal recessive, Autosomal dominant; {Macular degeneration, age-related, 4}, 610698 (3)
CFHR1	134371	{Hemolytic uremic syndrome, atypical, susceptibility to}, 235400 (3), Autosomal recessive, Autosomal dominant; {Macular degeneration, age-related, reduced risk of}, 603075 (3), Autosomal dominant
CFHR3	605336	{Hemolytic uremic syndrome, atypical, susceptibility to}, 235400 (3), Autosomal recessive, Autosomal dominant; {Macular degeneration, age-related, reduced risk of}, 603075 (3), Autosomal dominant
CFHR5	608593	Nephropathy due to CFHR5 deficiency, 614809 (3), Autosomal dominant

CFI	217030	Complement factor I deficiency, 610984 (3), Autosomal recessive; {Hemolytic uremic syndrome, atypical, susceptibility to, 3}, 612923 (3), Autosomal dominant; {Macular degeneration, age-related, 13, susceptibility to}, 615439 (3), Autosomal dominant
CFL2	601443	Nemaline myopathy 7, autosomal recessive, 610687 (3), Autosomal recessive
CFP	300383	Properdin deficiency, X-linked, 312060 (3), X-linked recessive
CFTR	602421	{Bronchiectasis with or without elevated sweat chloride 1, modifier of}, 211400 (3), Autosomal dominant; Congenital bilateral absence of vas deferens, 277180 (3), Autosomal recessive; Cystic fibrosis, 219700 (3), Autosomal recessive; {Hypertrypsinemia, neonatal} (3); {Pancreatitis, hereditary}, 167800 (3), Autosomal dominant; Sweat chloride elevation without CF (3)
CHAMP1	616327	Mental retardation, autosomal dominant 40, 616579 (3), Autosomal dominant
CHCHD10	615903	Frontotemporal dementia and/or amyotrophic lateral sclerosis 2, 615911 (3), Autosomal dominant; ?Myopathy, isolated mitochondrial, autosomal dominant, 616209 (3), Autosomal dominant; Spinal muscular atrophy, Jokela type, 615048 (3), Autosomal dominant
CHCHD2	616244	Parkinson disease 22, autosomal dominant, 616710 (3), Autosomal dominant
CHD2	602119	Epileptic encephalopathy, childhood-onset, 615369 (3), Autosomal dominant
CHD4	603277	Sifrim-Hitz-Weiss syndrome, 617159 (3), Autosomal dominant
CHD7	608892	CHARGE syndrome, 214800 (3), Autosomal dominant; Hypogonadotropic hypogonadism 5 with or without anosmia, 612370 (3), Autosomal dominant
CHD8	610528	{Autism, susceptibility to, 18}, 615032 (3), Autosomal dominant
CHEK2	604373	{Breast and colorectal cancer, susceptibility to} (3); {Breast cancer, susceptibility to}, 114480 (3), Autosomal dominant; Li-Fraumeni syndrome, 609265 (3); Osteosarcoma, somatic, 259500 (3); {Prostate cancer, familial, susceptibility to}, 176807 (3), Autosomal dominant
CHI3L1	601525	{Asthma-related traits, susceptibility to, 7}, 611960 (3); {Schizophrenia, susceptibility to}, 181500 (3), Autosomal dominant
CHIT1	600031	[Chitotriosidase deficiency], 614122 (3), Autosomal recessive
CHKB	612395	Muscular dystrophy, congenital, megaconial type, 602541 (3), Autosomal recessive
CHM	300390	Choroideremia, 303100 (3), X-linked dominant
CHMP1A	164010	Pontocerebellar hypoplasia, type 8, 614961 (3), Autosomal recessive
CHMP2B	609512	Amyotrophic lateral sclerosis 17, 614696 (3), Autosomal dominant; Dementia, familial, nonspecific, 600795 (3), Autosomal dominant
CHMP4B	610897	Cataract 31, multiple types, 605387 (3), Autosomal dominant
CHN1	118423	Duane retraction syndrome 2, 604356 (3)
CHRM3	118494	?Prune belly syndrome, 100100 (3), Autosomal recessive

CHRNA1	100690	Multiple pterygium syndrome, lethal type, 253290 (3), Autosomal recessive; Myasthenic syndrome, congenital, 1A, slow-channel, 601462 (3), Autosomal dominant; Myasthenic syndrome, congenital, 1B, fast-channel, 608930 (3), Autosomal recessive, Autosomal dominant
CHRNA2	118502	Epilepsy, nocturnal frontal lobe, type 4, 610353 (3), Autosomal dominant
CHRNA3	118503	{Lung cancer susceptibility 2}, 612052 (3)
CHRNA4	118504	Epilepsy, nocturnal frontal lobe, 1, 600513 (3), Autosomal dominant; {Nicotine addiction, susceptibility to}, 188890 (3)
CHRNA5	118505	{Lung cancer susceptibility 2}, 612052 (3); {Nicotine dependence, susceptibility to}, 612052 (3)
CHRNA7	118511	No OMIM phenotype
CHRNB1	100710	Myasthenic syndrome, congenital, 2A, slow-channel, 616313 (3), Autosomal dominant; ?Myasthenic syndrome, congenital, 2C, associated with acetylcholine receptor deficiency, 616314 (3), Autosomal recessive
CHRNB2	118507	Epilepsy, nocturnal frontal lobe, 3, 605375 (3)
CHRND	100720	Multiple pterygium syndrome, lethal type, 253290 (3), Autosomal recessive; ?Myasthenic syndrome, congenital, 3A, slow-channel, 616321 (3), Autosomal dominant; Myasthenic syndrome, congenital, 3B, fast-channel, 616322 (3), Autosomal recessive; ?Myasthenic syndrome, congenital, 3C, associated with acetylcholine receptor deficiency, 616323 (3), Autosomal recessive
CHRNE	100725	Myasthenic syndrome, congenital, 4A, slow-channel, 605809 (3), Autosomal recessive, Autosomal dominant; Myasthenic syndrome, congenital, 4B, fast-channel, 616324 (3), Autosomal recessive; Myasthenic syndrome, congenital, 4C, associated with acetylcholine receptor deficiency, 608931 (3), Autosomal recessive
CHRNG	100730	Escobar syndrome, 265000 (3), Autosomal recessive; Multiple pterygium syndrome, lethal type, 253290 (3), Autosomal recessive
CHST3	603799	Spondyloepiphyseal dysplasia with congenital joint dislocations, 143095 (3), Autosomal recessive
CHST6	605294	Macular corneal dystrophy, 217800 (3), Autosomal recessive
CHST8	610190	?Peeling skin syndrome 3, 616265 (3), Autosomal recessive
CHSY1	608183	Temtamy preaxial brachydactyly syndrome, 605282 (3), Autosomal recessive
CHUK	600664	Cocoon syndrome, 613630 (3)
CIB2	605564	Deafness, autosomal recessive 48, 609439 (3), Autosomal recessive; Usher syndrome, type IJ, 614869 (3), Autosomal recessive
CIDEC	612120	?Lipodystrophy, familial partial, type 5, 615238 (3), Autosomal recessive
CIITA	600005	Bare lymphocyte syndrome, type II, complementation group A, 209920 (3), Autosomal recessive; {Rheumatoid arthritis, susceptibility to}, 180300 (3)
CILP	603489	{Lumbar disc disease, susceptibility to}, 603932 (3)

CISD2	611507	Wolfram syndrome 2, 604928 (3), Autosomal recessive
CISH	602441	{Bacteremia, susceptibility to}, 614383 (3); {Malaria, susceptibility to}, 611162 (3); {Tuberculosis, susceptibility to}, 607948 (3)
CIT	605629	Microcephaly 17, primary, autosomal recessive, 617090 (3), Autosomal recessive
CITED2	602937	Atrial septal defect 8, 614433 (3), Autosomal dominant; Ventricular septal defect 2, 614431 (3), Autosomal dominant
CKAP2L	616174	Filippi syndrome, 272440 (3), Autosomal recessive
CLCF1	607672	Cold-induced sweating syndrome 2, 610313 (3), Autosomal recessive
CLCN1	118425	Myotonia congenita, dominant, 160800 (3), Autosomal dominant; Myotonia congenita, recessive, 255700 (3), Autosomal recessive; Myotonia levior, recessive (3)
CLCN2	600570	{Epilepsy, idiopathic generalized, susceptibility to, 11}, 607628 (3), Autosomal dominant; {Epilepsy, juvenile absence, susceptibility to, 2}, 607628 (3), Autosomal dominant; {Epilepsy, juvenile myoclonic, susceptibility to, 8}, 607628 (3), Autosomal dominant; Leukoencephalopathy with ataxia, 615651 (3), Autosomal recessive
CLCN4	302910	Mental retardation, X-linked 49/15, 300114 (3), X-linked recessive
CLCN5	300008	Dent disease, 300009 (3), X-linked recessive; Hypophosphatemic rickets, 300554 (3), X-linked recessive; Nephrolithiasis, type I, 310468 (3), X-linked recessive; Proteinuria, low molecular weight, with hypercalciuric nephrocalcinosis, 308990 (3), X-linked recessive
CLCN7	602727	Osteopetrosis, autosomal dominant 2, 166600 (3), Autosomal dominant; Osteopetrosis, autosomal recessive 4, 611490 (3)
CLCNKA	602024	Bartter syndrome, type 4b, digenic, 613090 (3), Digenic recessive
CLCNKB	602023	Bartter syndrome, type 3, 607364 (3), Autosomal recessive; Bartter syndrome, type 4b, digenic, 613090 (3), Digenic recessive
CLDN1	603718	Ichthyosis, leukocyte vacuoles, alopecia, and sclerosing cholangitis, 607626 (3), Autosomal recessive
CLDN14	605608	Deafness, autosomal recessive 29, 614035 (3), Autosomal recessive
CLDN16	603959	Hypomagnesemia 3, renal, 248250 (3), Autosomal recessive
CLDN19	610036	Hypomagnesemia 5, renal, with ocular involvement, 248190 (3), Autosomal recessive
CLEC7A	606264	{Aspergillosis, susceptibility to}, 614079 (3); Candidiasis, familial, 4, autosomal recessive, 613108 (3), Autosomal recessive
CLIC2	300138	?Mental retardation, X-linked, syndromic 32, 300886 (3), X-linked recessive
CLIC5	607293	?Deafness, autosomal recessive 103, 616042 (3), Autosomal recessive
CLMP	611693	Congenital short bowel syndrome, 615237 (3), Autosomal recessive
CLN3	607042	Ceroid lipofuscinosis, neuronal, 3, 204200 (3), Autosomal recessive
CLN5	608102	Ceroid lipofuscinosis, neuronal, 5, 256731 (3), Autosomal recessive
CLN6	606725	Ceroid lipofuscinosis, neuronal, 6, 601780 (3), Autosomal recessive; Ceroid lipofuscinosis, neuronal, Kufs type, adult onset, 204300 (3), Autosomal recessive

CLP1	608757	Pontocerebellar hypoplasia, type 10, 615803 (3), Autosomal recessive
CLPB	616254	3-methylglutaconic aciduria, type VII, with cataracts, neurologic involvement and neutropenia, 616271 (3), Autosomal recessive
CLPP	601119	Perrault syndrome 3, 614129 (3), Autosomal recessive
CLRN1	606397	Retinitis pigmentosa 61, 614180 (3); Usher syndrome, type 3A, 276902 (3), Autosomal recessive
CNBP	116955	Myotonic dystrophy 2, 602668 (3), Autosomal dominant
CNGA1	123825	Retinitis pigmentosa 49, 613756 (3)
CNGA3	600053	Achromatopsia 2, 216900 (3), Autosomal recessive
CNGB1	600724	Retinitis pigmentosa 45, 613767 (3), Autosomal recessive
CNGB3	605080	Achromatopsia 3, 262300 (3), Autosomal recessive; Macular degeneration, juvenile, 248200 (3), Autosomal recessive
CNNM2	607803	Hypomagnesemia 6, renal, 613882 (3), Autosomal dominant; Hypomagnesemia, seizures, and mental retardation, 616418 (3), Autosomal recessive, Autosomal dominant
CNNM4	607805	Jalili syndrome, 217080 (3), Autosomal recessive
CNTN2	190197	?Epilepsy, myoclonic, familial adult, 5, 615400 (3), Autosomal recessive
CNTNAP1	602346	Lethal congenital contracture syndrome 7, 616286 (3), Autosomal recessive
CNTNAP2	604569	{Autism susceptibility 15}, 612100 (3); Cortical dysplasia-focal epilepsy syndrome, 610042 (3), Autosomal recessive; Pitt-Hopkins like syndrome 1, 610042 (3), Autosomal recessive
COA5	613920	?Cardioencephalomyopathy, fatal infantile, due to cytochrome c oxidase deficiency 3, 616500 (3), Autosomal recessive
COA6	614772	Cardioencephalomyopathy, fatal infantile, due to cytochrome c oxidase deficiency 4, 616501 (3), Autosomal recessive
COASY	609855	Neurodegeneration with brain iron accumulation 6, 615643 (3), Autosomal recessive
COCH	603196	Deafness, autosomal dominant 9, 601369 (3), Autosomal dominant
COG1	606973	Congenital disorder of glycosylation, type IIg, 611209 (3)
COG2	606974	?Congenital disorder of glycosylation, type IIq, 617395 (3), Autosomal recessive
COG4	606976	Congenital disorder of glycosylation, type IIj, 613489 (3), Autosomal recessive
COG5	606821	Congenital disorder of glycosylation, type IIi, 613612 (3)
COG6	606977	Congenital disorder of glycosylation, type III, 614576 (3), Autosomal recessive; Shaheen syndrome, 615328 (3), Autosomal recessive
COG7	606978	Congenital disorder of glycosylation, type IIe, 608779 (3)
COG8	606979	Congenital disorder of glycosylation, type IIh, 611182 (3)
COL10A1	120110	Metaphyseal chondrodysplasia, Schmid type, 156500 (3), Autosomal dominant

COL11A1	120280	Fibrochondrogenesis 1, 228520 (3), Autosomal recessive; {Lumbar disc herniation, susceptibility to}, 603932 (3); Marshall syndrome, 154780 (3), Autosomal dominant; Stickler syndrome, type II, 604841 (3), Autosomal dominant
COL11A2	120290	Deafness, autosomal dominant 13, 601868 (3), Autosomal dominant; Deafness, autosomal recessive 53, 609706 (3), Autosomal recessive; Fibrochondrogenesis 2, 614524 (3), Autosomal recessive, Autosomal dominant; Otospondylomegaepiphyseal dysplasia, autosomal dominant, 184840 (3), Autosomal dominant; Otospondylomegaepiphyseal dysplasia, autosomal recessive, 215150 (3), Autosomal recessive
COL13A1	120350	Myasthenic syndrome, congenital, 19, 616720 (3), Autosomal recessive
COL17A1	113811	Epidermolysis bullosa, junctional, localisata variant, 226650 (3), Autosomal recessive; Epidermolysis bullosa, junctional, non-Herlitz type, 226650 (3), Autosomal recessive; Epithelial recurrent erosion dystrophy, 122400 (3), Autosomal dominant
COL18A1	120328	Knobloch syndrome, type 1, 267750 (3), Autosomal recessive
COL1A1	120150	{Bone mineral density variation QTL, osteoporosis}, 166710 (3), Autosomal dominant; Caffey disease, 114000 (3), Autosomal dominant; Ehlers-Danlos syndrome, arthrochalasia type, 1, 130060 (3), Autosomal dominant; Osteogenesis imperfecta, type I, 166200 (3), Autosomal dominant; Osteogenesis imperfecta, type II, 166210 (3), Autosomal dominant; Osteogenesis imperfecta, type III, 259420 (3), Autosomal dominant; Osteogenesis imperfecta, type IV, 166220 (3), Autosomal dominant
COL1A2	120160	Ehlers-Danlos syndrome, arthrochalasia type, 2, 617821 (3), Autosomal dominant; Ehlers-Danlos syndrome, cardiac valvular type, 225320 (3), Autosomal recessive; imperfecta, type III, 259420 (3), Autosomal dominant; Osteogenesis imperfecta, type II, 166210 (3), Autosomal dominant; Osteogenesis imperfecta, type IV, 166220 (3), Autosomal dominant; {Osteoporosis, postmenopausal}, 166710 (3), Autosomal dominant
COL25A1	610004	Fibrosis of extraocular muscles, congenital, 5, 616219 (3), Autosomal recessive
COL27A1	608461	?Steel syndrome, 615155 (3), Autosomal recessive

COL2A1	120140	Achondrogenesis, type II or hypochondrogenesis, 200610 (3), Autosomal dominant; Avascular necrosis of the femoral head, 608805 (3), Autosomal dominant; Czech dysplasia, 609162 (3), Autosomal dominant; Epiphyseal dysplasia, multiple, with myopia and deafness, 132450 (3), Autosomal dominant; Kniest dysplasia, 156550 (3), Autosomal dominant; Legg-Calve-Perthes disease, 150600 (3), Autosomal dominant; Osteoarthritis with mild chondrodysplasia, 604864 (3), Autosomal dominant; Platyspondylic skeletal dysplasia, Torrance type, 151210 (3), Autosomal dominant; SED congenita, 183900 (3), Autosomal dominant; SMED Strudwick type, 184250 (3), Autosomal dominant; Spondyloepiphyseal dysplasia, Stanescu type, 616583 (3), Autosomal dominant; Spondyloperipheral dysplasia, 271700 (3), Autosomal dominant; Stickler sydrome, type I, nonsyndromic ocular, 609508 (3), Autosomal dominant; Stickler syndrome, type I, 108300 (3), Autosomal dominant; Vitreoretinopathy with phalangeal epiphyseal dysplasia (3)
COL3A1	120180	Ehlers-Danlos syndrome, vascular type, 130050 (3), Autosomal dominant
COL4A1	120130	Angiopathy, hereditary, with nephropathy, aneurysms, and muscle cramps, 611773 (3), Autosomal dominant; Brain small vessel disease with or without ocular anomalies, 607595 (3), Autosomal dominant; {Hemorrhage, intracerebral, susceptibility to}, 614519 (3); Porencephaly 1, 175780 (3), Autosomal dominant; ?Retinal arteries, tortuosity of, 180000 (3), Autosomal dominant; Schizencephaly, 269160 (3)
COL4A2	120090	{Hemorrhage, intracerebral, susceptibility to}, 614519 (3); Porencephaly 2, 614483 (3), Autosomal dominant
COL4A3	120070	Alport syndrome, autosomal dominant, 104200 (3), Autosomal dominant; Alport syndrome, autosomal recessive, 203780 (3), Autosomal recessive; Hematuria, benign familial, 141200 (3), Autosomal dominant
COL4A3BP	604677	Mental retardation, autosomal dominant 34, 616351 (3), Autosomal dominant
COL4A4	120131	Alport syndrome, autosomal recessive, 203780 (3), Autosomal recessive; Hematuria, familial benign (3)
COL4A5	303630	Alport syndrome, 301050 (3), X-linked dominant
COL5A1	120215	Ehlers-Danlos syndrome, classic type, 1, 130000 (3), Autosomal dominant
COL5A2	120190	Ehlers-Danlos syndrome, classic type, 2, 130010 (3), Autosomal dominant
COL6A1	120220	Bethlem myopathy 1, 158810 (3), Autosomal recessive, Autosomal dominant; Ullrich congenital muscular dystrophy 1, 254090 (3), Autosomal recessive, Autosomal dominant
COL6A2	120240	Bethlem myopathy 1, 158810 (3), Autosomal recessive, Autosomal dominant; ?Myosclerosis, congenital, 255600 (3), Autosomal recessive; Ullrich congenital muscular dystrophy 1, 254090 (3), Autosomal recessive, Autosomal dominant

COL7A1	120120	EBD inversa, 226600 (3), Autosomal recessive; EBD, Bart type, 132000 (3), Autosomal dominant; EBD, localisata variant (3); Epidermolysis bullosa dystrophica, AD, 131750 (3), Autosomal dominant; Epidermolysis bullosa dystrophica, AR, 226600 (3), Autosomal recessive; Epidermolysis bullosa pruriginosa, 604129 (3), Autosomal recessive, Autosomal dominant; Epidermolysis bullosa, pretibial, 131850 (3), Autosomal recessive, Autosomal dominant; Toenail dystrophy, isolated, 607523 (3), Autosomal dominant; Transient bullous of the newborn, 131705 (3), Autosomal recessive, Autosomal dominant
COL8A2	120252	Corneal dystrophy, Fuchs endothelial, 1, 136800 (3), Autosomal dominant; Corneal dystrophy, posterior polymorphous 2, 609140 (3), Autosomal dominant
COL9A1	120210	?Epiphyseal dysplasia, multiple, 6, 614135 (3), Autosomal dominant; Stickler syndrome, type IV, 614134 (3)
COL9A2	120260	Epiphyseal dysplasia, multiple, 2, 600204 (3), Autosomal dominant; ?Stickler syndrome, type V, 614284 (3), Autosomal recessive
COL9A3	120270	Epiphyseal dysplasia, multiple, 3, with or without myopathy, 600969 (3), Autosomal dominant; {Intervertebral disc disease, susceptibility to}, 603932 (3)
COLQ	603033	Myasthenic syndrome, congenital, 5, 603034 (3), Autosomal recessive
COMP	600310	Epiphyseal dysplasia, multiple, 1, 132400 (3), Autosomal dominant; Pseudoachondroplasia, 177170 (3), Autosomal dominant
COPA	601924	{Autoimmune interstitial lung, joint, and kidney disease}, 616414 (3), Autosomal dominant
COQ2	609825	Coenzyme Q10 deficiency, primary, 1, 607426 (3), Autosomal recessive; {Multiple system atrophy, susceptibility to}, 146500 (3), Autosomal recessive, Autosomal dominant
COQ4	612898	Coenzyme Q10 deficiency, primary, 7, 616276 (3), Autosomal recessive
COQ7	601683	?Coenzyme Q10 deficiency, primary, 8, 616733 (3), Autosomal recessive
COQ8A	606980	Coenzyme Q10 deficiency, primary, 4, 612016 (3), Autosomal recessive
COQ8B	615567	Nephrotic syndrome, type 9, 615573 (3), Autosomal recessive
COQ9	612837	Coenzyme Q10 deficiency, primary, 5, 614654 (3), Autosomal recessive
CORIN	605236	Preeclampsia/eclampsia 5, 614595 (3)
COX10	602125	Leigh syndrome due to mitochondrial COX4 deficiency, 256000 (3), Autosomal recessive, Mitochondrial; Mitochondrial complex IV deficiency, 220110 (3), Autosomal recessive, Mitochondrial
COX14	614478	?Mitochondrial complex IV deficiency, 220110 (3), Autosomal recessive, Mitochondrial
COX20	614698	Mitochondrial complex IV deficiency, 220110 (3), Autosomal recessive, Mitochondrial
COX4I2	607976	Exocrine pancreatic insufficiency, dyserythropoietic anemia, and calvarial hyperostosis, 612714 (3), Autosomal recessive

COX6A1	602072	Charcot-Marie-Tooth disease, recessive intermediate D, 616039 (3), Autosomal recessive
COX6B1	124089	Mitochondrial complex IV deficiency, 220110 (3), Autosomal recessive, Mitochondrial
COX7B	300885	Linear skin defects with multiple congenital anomalies 2, 300887 (3), X-linked dominant
CP	117700	Cerebellar ataxia, 604290 (3), Autosomal recessive; Hemosiderosis, systemic, due to aceruloplasminemia, 604290 (3), Autosomal recessive; [Hypoceruloplasminemia, hereditary], 604290 (3), Autosomal recessive
CPA6	609562	Epilepsy, familial temporal lobe, 5, 614417 (3), Autosomal recessive, Autosomal dominant; Febrile seizures, familial, 11, 614418 (3), Autosomal recessive
CPAMD8	608841	Anterior segment dysgenesis 8, 617319 (3), Autosomal recessive
CPLANE1 (C5orf42)	614571	Joubert syndrome 17, 614615 (3), Autosomal recessive; Orofaciodigital syndrome VI, 277170 (3), Autosomal recessive
CPLX1	605032	Epileptic encephalopathy, early infantile, 63, 617976 (3), Autosomal recessive
CPN1	603103	Carboxypeptidase N deficiency, 212070 (3), Autosomal recessive
CPOX	612732	Coproporphyria, 121300 (3), Autosomal dominant; Harderoporphyria, 121300 (3), Autosomal dominant
CPS1	608307	Carbamoylphosphate synthetase I deficiency, 237300 (3), Autosomal recessive; {Pulmonary hypertension, neonatal, susceptibility to}, 615371 (3); {Venoocclusive disease after bone marrow transplantation} (3)
CPT1C	608846	?Spastic paraplegia 73, autosomal dominant, 616282 (3), Autosomal dominant
CPT2	600650	CPT II deficiency, infantile, 600649 (3), Autosomal recessive; CPT II deficiency, lethal neonatal, 608836 (3), Autosomal recessive; CPT II deficiency, myopathic, stress-induced, 255110 (3), Autosomal recessive, Autosomal dominant; {Encephalopathy, acute, infection-induced, 4, susceptibility to}, 614212 (3), Autosomal recessive, Autosomal dominant
CR1	120620	[Blood group, Knops system], 607486 (3); CR1 deficiency (1); {Malaria, severe, resistance to}, 611162 (3); {?SLE susceptibility} (1)
CR2	120650	Immunodeficiency, common variable, 7, 614699 (3), Autosomal recessive; {Systemic lupus erythematosus, susceptibility to, 9}, 610927 (3)
CRADD	603454	Mental retardation, autosomal recessive 34, with variant lissencephaly, 614499 (3), Autosomal recessive
CRB1	604210	Leber congenital amaurosis 8, 613835 (3); Pigmented paravenous chorioretinal atrophy, 172870 (3), Autosomal dominant; Retinitis pigmentosa-12, autosomal recessive, 600105 (3), Autosomal recessive
CRB2	609720	Focal segmental glomerulosclerosis 9, 616220 (3), Autosomal recessive; Ventriculomegaly with cystic kidney disease, 219730 (3), Autosomal recessive

CRBN	609262	Mental retardation, autosomal recessive 2, 607417 (3), Autosomal recessive
CREB1	123810	Histiocytoma, angiomatoid fibrous, somatic, 612160 (3)
CREBBP	600140	Rubinstein-Taybi syndrome 1, 180849 (3), Autosomal dominant
CRELD1	607170	Atrioventricular septal defect, partial, with heterotaxy syndrome, 606217 (3), Autosomal dominant; {Atrioventricular septal defect, susceptibility to, 2}, 606217 (3), Autosomal dominant
CRIPT	604594	Short stature with microcephaly and distinctive facies, 615789 (3), Autosomal recessive
CRLF1	604237	Cold-induced sweating syndrome 1, 272430 (3), Autosomal recessive
CRTAP	605497	Osteogenesis imperfecta, type VII, 610682 (3), Autosomal recessive
CRX	602225	Cone-rod retinal dystrophy-2, 120970 (3), Autosomal dominant; Leber congenital amaurosis 7, 613829 (3)
CRYAA	123580	Cataract 9, multiple types, 604219 (3), Autosomal recessive, Autosomal dominant
CRYAB	123590	Cardiomyopathy, dilated, 1II, 615184 (3), Autosomal dominant; Cataract 16, multiple types, 613763 (3), Autosomal recessive, Autosomal dominant; Myopathy, myofibrillar, 2, 608810 (3), Autosomal dominant; Myopathy, myofibrillar, fatal infantile hypertonic, alpha-B crystallin-related, 613869 (3), Autosomal recessive
CRYBA1	123610	Cataract 10, multiple types, 600881 (3), Autosomal dominant
CRYBA2	600836	?Cataract 42, 115900 (3), Autosomal dominant
CRYBA4	123631	Cataract 23, 610425 (3)
CRYBB1	600929	Cataract 17, multiple types, 611544 (3), Autosomal recessive, Autosomal dominant
CRYBB2	123620	Cataract 3, multiple types, 601547 (3), Autosomal dominant
CRYBB3	123630	Cataract 22, 609741 (3), Autosomal recessive, Autosomal dominant
CRYGB	123670	Cataract 39, multiple types, autosomal dominant, 615188 (3), Autosomal dominant
CRYGC	123680	Cataract 2, multiple types, 604307 (3), Autosomal dominant
CRYGD	123690	Cataract 4, multiple types, 115700 (3), Autosomal dominant
CRYGS	123730	Cataract 20, multiple types, 116100 (3), Autosomal dominant
CRYM	123740	Deafness, autosomal dominant 40, 616357 (3), Autosomal dominant
CSF1R	164770	Leukoencephalopathy, diffuse hereditary, with spheroids, 221820 (3), Autosomal dominant
CSF2RB	138981	Surfactant metabolism dysfunction, pulmonary, 5, 614370 (3), Autosomal recessive
CSF3R	138971	Neutropenia, severe congenital, 7, autosomal recessive, 617014 (3), Autosomal recessive
CSNK1D	600864	Advanced sleep-phase syndrome, familial, 2, 615224 (3), Autosomal dominant
CSNK2A1	115440	Okur-Chung neurodevelopmental syndrome, 617062 (3), Autosomal dominant
CSPP1	611654	Joubert syndrome 21, 615636 (3), Autosomal recessive

CSRP3	600824	?Cardiomyopathy, dilated, 1M, 607482 (3); Cardiomyopathy, hypertrophic, 12, 612124 (3), Autosomal dominant
CST3	604312	Cerebral amyloid angiopathy, 105150 (3), Autosomal dominant; {Macular degeneration, age-related, 11}, 611953 (3)
CSTA	184600	Peeling skin syndrome 4, 607936 (3), Autosomal recessive
CSTB	601145	Epilepsy, progressive myoclonic 1A (Unverricht and Lundborg), 254800 (3), Autosomal recessive
CTBP1	602618	Hypotonia, ataxia, developmental delay, and tooth enamel defect syndrome, 617915 (3), Autosomal dominant
CTC1	613129	Cerebroretinal microangiopathy with calcifications and cysts, 612199 (3), Autosomal recessive
CTCF	604167	Mental retardation, autosomal dominant 21, 615502 (3), Autosomal dominant
CTDP1	604927	Congenital cataracts, facial dysmorphism, and neuropathy, 604168 (3), Autosomal recessive
CTH	607657	Cystathioninuria, 219500 (3), Autosomal recessive; Homocysteine, total plasma, elevated (3)
CTHRC1	610635	Barrett esophagus/esophageal adenocarcinoma, 614266 (3)
CTLA4	123890	Autoimmune lymphoproliferative syndrome, type V, 616100 (3), Autosomal dominant; {Celiac disease, susceptibility to, 3}, 609755 (3); {Diabetes mellitus, insulin-dependent, 12}, 601388 (3); {Hashimoto thyroiditis}, 140300 (3), Autosomal dominant; {Systemic lupus erythematosus, susceptibility to}, 152700 (3), Autosomal dominant
CTNNA1	116805	Macular dystrophy, patterned, 2, 608970 (3), Autosomal dominant
CTNNA3	607667	Arrhythmogenic right ventricular dysplasia, familial, 13, 615616 (3), Autosomal dominant
CTNNB1	116806	Colorectal cancer, somatic, 114500 (3); Exudative vitreoretinopathy 7, 617572 (3), Autosomal dominant; Hepatocellular carcinoma, somatic, 114550 (3); Medulloblastoma, somatic, 155255 (3); Mental retardation, autosomal dominant 19, 615075 (3), Autosomal dominant; Ovarian cancer, somatic, 167000 (3); Pilomatricoma, somatic, 132600 (3)
CTNS	606272	Cystinosis, atypical nephropathic, 219800 (3), Autosomal recessive; Cystinosis, late-onset juvenile or adolescent nephropathic, 219900 (3), Autosomal recessive; Cystinosis, nephropathic, 219800 (3), Autosomal recessive; Cystinosis, ocular nonnephropathic, 219750 (3), Autosomal recessive
CTPS1	123860	Immunodeficiency 24, 615897 (3), Autosomal recessive
CTRC	601405	{Pancreatitis, chronic, susceptibility to}, 167800 (3), Autosomal dominant
CTSA	613111	Galactosialidosis, 256540 (3), Autosomal recessive
CTSC	602365	Haim-Munk syndrome, 245010 (3), Autosomal recessive; Papillon-Lefevre syndrome, 245000 (3), Autosomal recessive; Periodontitis 1, juvenile, 170650 (3), Autosomal recessive
CTSD	116840	Ceroid lipofuscinosis, neuronal, 10, 610127 (3), Autosomal recessive

CTSF	603539	Ceroid lipofuscinosis, neuronal, 13, Kufs type, 615362 (3), Autosomal recessive
CTSK	601105	Pycnodysostosis, 265800 (3), Autosomal recessive
CUBN	602997	Megaloblastic anemia-1, Finnish type, 261100 (3), Autosomal recessive
CUL3	603136	Pseudohypoaldosteronism, type IIE, 614496 (3), Autosomal dominant
CUL4B	300304	Mental retardation, X-linked, syndromic 15 (Cabezas type), 300354 (3), X-linked recessive
CWF19L1	616120	Spinocerebellar ataxia, autosomal recessive 17, 616127 (3), Autosomal recessive
CX3CR1	601470	{Coronary artery disease, resistance to}, 607339 (3); {Macular degeneration, age-related, 12}, 613784 (3); {Rapid progression to AIDS from HIV1 infection}, 609423 (3)
CXCL12	600835	{AIDS, resistance to}, 609423 (3)
CXCR1	146929	{AIDS, slow progression to}, 609423 (3)
CXCR4	162643	Myelokathexis, isolated (3); WHIM syndrome, 193670 (3), Autosomal dominant
CYB5A	613218	Methemoglobinemia and ambiguous genitalia, 250790 (3), Autosomal recessive
CYB5R3	613213	Methemoglobinemia, type I, 250800 (3), Autosomal recessive; Methemoglobinemia, type II, 250800 (3), Autosomal recessive
CYBA	608508	Chronic granulomatous disease, autosomal, due to deficiency of CYBA, 233690 (3), Autosomal recessive
CYBB	300481	Chronic granulomatous disease, X-linked, 306400 (3), X-linked recessive; Immunodeficiency 34, mycobacteriosis, X-linked, 300645 (3), X-linked recessive
CYC1	123980	Mitochondrial complex III deficiency, nuclear type 6, 615453 (3), Autosomal recessive
CYCS	123970	Thrombocytopenia 4, 612004 (3), Autosomal dominant
CYP11A1	118485	Adrenal insufficiency, congenital, with 46XY sex reversal, partial or complete, 613743 (3)
CYP11B1	610613	Adrenal hyperplasia, congenital, due to 11-beta-hydroxylase deficiency, 202010 (3), Autosomal recessive; Aldosteronism, glucocorticoid-remediable, 103900 (3), Autosomal dominant
CYP11B2	124080	Aldosterone to renin ratio raised (3); Hypoaldosteronism, congenital, due to CMO I deficiency, 203400 (3), Autosomal recessive; Hypoaldosteronism, congenital, due to CMO II deficiency, 610600 (3), Autosomal recessive; {Low renin hypertension, susceptibility to} (3)
CYP17A1	609300	17-alpha-hydroxylase/17,20-lyase deficiency, 202110 (3), Autosomal recessive; 17,20-lyase deficiency, isolated, 202110 (3), Autosomal recessive
CYP19A1	107910	Aromatase deficiency, 613546 (3); Aromatase excess syndrome, 139300 (3), Autosomal dominant

CYP1B1	601771	Anterior segment dysgenesis 6, multiple subtypes, 617315 (3); Glaucoma 3A, primary open angle, congenital, juvenile, or adult onset, 231300 (3), Autosomal recessive
CYP21A2	613815	Adrenal hyperplasia, congenital, due to 21-hydroxylase deficiency, 201910 (3), Autosomal recessive; Hyperandrogenism, nonclassic type, due to 21-hydroxylase deficiency, 201910 (3), Autosomal recessive
CYP24A1	126065	Hypercalcemia, infantile, 1, 143880 (3), Autosomal recessive
CYP26B1	605207	Craniosynostosis with radiohumeral fusions and other skeletal and craniofacial anomalies, 614416 (3)
CYP26C1	608428	Focal facial dermal dysplasia 4, 614974 (3), Autosomal recessive
CYP27A1	606530	Cerebrotendinous xanthomatosis, 213700 (3), Autosomal recessive
CYP27B1	609506	Vitamin D-dependent rickets, type I, 264700 (3), Autosomal recessive
CYP2A6	122720	Coumarin resistance, 122700 (3), Autosomal dominant; {Lung cancer, resistance to}, 211980 (3), Autosomal recessive; {Nicotine addiction, protection from}, 188890 (3)
CYP2B6	123930	{Efavirenz central nervous system toxicity, susceptibility to}, 614546 (3); Efavirenz, poor metabolism of, 614546 (3)
CYP2C19	124020	Clopidogrel, impaired responsiveness to, 609535 (3), Autosomal recessive; Mephenytoin poor metabolizer, 609535 (3), Autosomal recessive; Omeprazole poor metabolizer, 609535 (3), Autosomal recessive; Proguanil poor metabolizer, 609535 (3), Autosomal recessive
CYP2C9	601130	Tolbutamide poor metabolizer (3); Warfarin sensitivity, 122700 (3), Autosomal dominant
CYP2D6	124030	{Codeine sensitivity}, 608902 (3), Autosomal recessive; {Debrisoquine sensitivity}, 608902 (3), Autosomal recessive
CYP2R1	608713	Rickets due to defect in vitamin D 25-hydroxylation, 600081 (3), Autosomal recessive
CYP2U1	610670	Spastic paraplegia 56, autosomal recessive, 615030 (3), Autosomal recessive
CYP3A5	605325	{Hypertension, salt-sensitive essential, susceptibility to}, 145500 (3), Multifactorial
CYP4V2	608614	Bietti crystalline corneoretinal dystrophy, 210370 (3), Autosomal recessive
CYP7B1	603711	Bile acid synthesis defect, congenital, 3, 613812 (3), Autosomal recessive; Spastic paraplegia 5A, autosomal recessive, 270800 (3), Autosomal recessive
D2HGDH	609186	D-2-hydroxyglutaric aciduria, 600721 (3), Autosomal recessive
DAO	124050	No OMIM phenotype
DAOA	607408	{Schizophrenia}, 181500 (2), Autosomal dominant
DARS	603084	Hypomyelination with brainstem and spinal cord involvement and leg spasticity, 615281 (3), Autosomal recessive
DARS2	610956	Leukoencephalopathy with brain stem and spinal cord involvement and lactate elevation, 611105 (3), Autosomal recessive
DAZ1	400003	?Sertoli-cell-only syndrome (1)

DAZ2	400026	No OMIM phenotype
DAZ3	400027	No OMIM phenotype
DBH	609312	Dopamine beta-hydroxylase deficiency, 223360 (3), Autosomal recessive; [Dopamine-beta-hydroxylase activity levels, plasma] (3)
DBT	248610	Maple syrup urine disease, type II, 248600 (3), Autosomal recessive
DCAF17	612515	Woodhouse-Sakati syndrome, 241080 (3), Autosomal recessive
DCAF8	615820	?Giant axonal neuropathy 2, autosomal dominant, 610100 (3), Autosomal dominant
DCC	120470	Colorectal cancer, somatic, 114500 (3); Esophageal carcinoma, somatic, 133239 (3); Gaze palsy, familial horizontal, with progressive scoliosis, 2, 617542 (3), Autosomal recessive; Mirror movements 1 and/or agenesis of the corpus callosum, 157600 (3), Autosomal dominant
DCDC2	605755	?Deafness, autosomal recessive 66, 610212 (3), Autosomal recessive; Nephronophthisis 19, 616217 (3), Autosomal recessive; Sclerosing cholangitis, neonatal, 617394 (3), Autosomal recessive
DCHS1	603057	Mitral valve prolapse 2, 607829 (3), Autosomal dominant; Van Maldergem syndrome 1, 601390 (3), Autosomal recessive
DCN	125255	Corneal dystrophy, congenital stromal, 610048 (3), Autosomal dominant
DCPS	610534	Al-Raqad syndrome, 616459 (3), Autosomal recessive
DCTN1	601143	{Amyotrophic lateral sclerosis, susceptibility to}, 105400 (3), Autosomal recessive, Autosomal dominant; Neuropathy, distal hereditary motor, type VIIB, 607641 (3), Autosomal dominant; Perry syndrome, 168605 (3), Autosomal dominant
DCX	300121	Lissencephaly, X-linked, 300067 (3), X-linked; Subcortical laminal heterotopia, X-linked, 300067 (3), X-linked
DCXR	608347	[Pentosuria], 260800 (3), Autosomal recessive
DDB2	600811	Xeroderma pigmentosum, group E, DDB-negative subtype, 278740 (3), Autosomal recessive
DDC	107930	Aromatic L-amino acid decarboxylase deficiency, 608643 (3), Autosomal recessive
DDHD1	614603	Spastic paraplegia 28, autosomal recessive, 609340 (3), Autosomal recessive
DDHD2	615003	Spastic paraplegia 54, autosomal recessive, 615033 (3), Autosomal recessive
DDIT3	126337	Myxoid liposarcoma, 613488 (1)
DDOST	602202	?Congenital disorder of glycosylation, type Ir, 614507 (3), Autosomal recessive
DDR2	191311	Spondylometaepiphyseal dysplasia, short limb-hand type, 271665 (3), Autosomal recessive
DDX11	601150	Warsaw breakage syndrome, 613398 (3), Autosomal recessive
DDX3X	300160	Mental retardation, X-linked 102, 300958 (3), X-linked recessive, X-linked dominant

DDX41	608170	{Myeloproliferative/lymphoproliferative neoplasms, familial (multiple types), susceptibility to}, 616871 (3), Autosomal dominant
DDX58	609631	Singleton-Merten syndrome 2, 616298 (3), Autosomal dominant
DDX59	615464	Orofaciodigital syndrome V, 174300 (3), Autosomal recessive
DEAF1	602635	?Dyskinesia, seizures, and intellectual developmental disorder, 617171 (3), Autosomal recessive; Mental retardation, autosomal dominant 24, 615828 (3), Autosomal dominant
DEC1	604767	Esophageal squamous cell carcinoma, 133239 (1), Autosomal dominant
DENND5A	617278	Epileptic encephalopathy, early infantile, 49, 617281 (3), Autosomal recessive
DEPDC5	614191	Epilepsy, familial focal, with variable foci 1, 604364 (3), Autosomal dominant
DES	125660	Cardiomyopathy, dilated, 1I, 604765 (3); ?Muscular dystrophy, limb-girdle, type 2R, 615325 (3), Autosomal recessive; Myopathy, myofibrillar, 1, 601419 (3), Autosomal recessive, Autosomal dominant; Scapuloperoneal syndrome, neurogenic, Kaeser type, 181400 (3), Autosomal dominant
DGAT1	604900	?Diarrhea 7, 615863 (3), Autosomal recessive
DGCR2	600594	No OMIM phenotype
DGCR6	601279	No OMIM phenotype
DGCR8	609030	No OMIM phenotype
DGKE	601440	{Hemolytic uremic syndrome, atypical, susceptibility to, 7}, 615008 (3), Autosomal recessive; Nephrotic syndrome, type 7, 615008 (3), Autosomal recessive
DGUOK	601465	Mitochondrial DNA depletion syndrome 3 (hepatocerebral type), 251880 (3), Autosomal recessive; Portal hypertension, noncirrhotic, 617068 (3), Autosomal recessive; Progressive external ophthalmoplegia with mitochondrial DNA deletions, autosomal recessive 4, 617070 (3), Autosomal recessive
DHCR24	606418	Desmosterolosis, 602398 (3), Autosomal recessive
DHCR7	602858	Smith-Lemli-Opitz syndrome, 270400 (3), Autosomal recessive
DHDDS	608172	?Congenital disorder of glycosylation, type 1bb, 613861 (3), Autosomal recessive; Developmental delay and seizures with or without movement abnormalities, 617836 (3), Autosomal dominant; Retinitis pigmentosa 59, 613861 (3), Autosomal recessive
DHFR	126060	Megaloblastic anemia due to dihydrofolate reductase deficiency, 613839 (3), Autosomal recessive
DHH	605423	46XY partial gonadal dysgenesis, with minifascicular neuropathy, 607080 (3); 46XY sex reversal 7, 233420 (3), Autosomal recessive
DHODH	126064	Miller syndrome, 263750 (3), Autosomal recessive
DHTKD1	614984	2-aminoadipic 2-oxoadipic aciduria, 204750 (3), Autosomal recessive; ?Charcot-Marie-Tooth disease, axonal, type 2Q, 615025 (3), Autosomal dominant
DIABLO	605219	Deafness, autosomal dominant 64, 614152 (3), Autosomal dominant

DIAPH1	602121	Deafness, autosomal dominant 1, 124900 (3), Autosomal dominant; Seizures, cortical blindness, microcephaly syndrome, 616632 (3), Autosomal recessive
DIAPH2	300108	?Premature ovarian failure 2A, 300511 (3), X-linked dominant
DIP2B	611379	Mental retardation, FRA12A type, 136630 (3), Autosomal dominant
DIRC2	602773	Renal cell carcinoma, 144700 (1)
DIS3L2	614184	Perlman syndrome, 267000 (3), Autosomal recessive
DISC1	605210	{Schizophrenia 9, susceptibility to}, 604906 (3)
DKC1	300126	Dyskeratosis congenita, X-linked, 305000 (3), X-linked recessive
DLAT	608770	Pyruvate dehydrogenase E2 deficiency, 245348 (3), Autosomal recessive
DLC1	604258	Colorectal cancer, somatic, 114500 (3)
DLD	238331	Dihydrolipoamide dehydrogenase deficiency, 246900 (3), Autosomal recessive
DLEC1	604050	Esophageal cancer, 133239 (1), Autosomal dominant; Lung cancer, 211980 (1), Autosomal recessive
DLG3	300189	Mental retardation, X-linked 90, 300850 (3), X-linked recessive
DLL3	602768	Spondylocostal dysostosis 1, autosomal recessive, 277300 (3), Autosomal recessive
DLL4	605185	Adams-Oliver syndrome 6, 616589 (3), Autosomal dominant
DLX3	600525	Amelogenesis imperfecta, type IV, 104510 (3), Autosomal dominant; Trichodontoosseous syndrome, 190320 (3), Autosomal dominant
DLX4	601911	?Orofacial cleft 15, 616788 (3), Autosomal dominant
DLX5	600028	?Split-hand/foot malformation 1 with sensorineural hearing loss, 220600 (3), Autosomal recessive
DMGDH	605849	Dimethylglycine dehydrogenase deficiency, 605850 (3), Autosomal recessive
DMP1	600980	Hypophosphatemic rickets, AR, 241520 (3), Autosomal recessive
DMPK	605377	Myotonic dystrophy 1, 160900 (3), Autosomal dominant
DMXL2	612186	?Deafness, autosomal dominant 71, 617605 (3), Autosomal dominant; ?Polyendocrine-polyneuropathy syndrome, 616113 (3), Autosomal recessive
DNA2	601810	Progressive external ophthalmoplegia with mitochondrial DNA deletions, autosomal dominant 6, 615156 (3), Autosomal dominant; ?Seckel syndrome 8, 615807 (3), Autosomal recessive
DNAAF1	613190	Ciliary dyskinesia, primary, 13, 613193 (3), Autosomal recessive
DNAAF2	612517	Ciliary dyskinesia, primary, 10, 612518 (3)
DNAAF3	614566	Ciliary dyskinesia, primary, 2, 606763 (3), Autosomal recessive
DNAAF4	608706	Ciliary dyskinesia, primary, 25, 615482 (3), Autosomal recessive; {Dyslexia, susceptibility to, 1}, 127700 (3), Autosomal dominant
DNAAF5	614864	Ciliary dyskinesia, primary, 18, 614874 (3), Autosomal recessive
DNAH11	603339	Ciliary dyskinesia, primary, 7, with or without situs inversus, 611884 (3), Autosomal recessive
DNAH5	603335	Ciliary dyskinesia, primary, 3, with or without situs inversus, 608644 (3)

DNAI1	604366	Ciliary dyskinesia, primary, 1, with or without situs inversus, 244400 (3), Autosomal recessive
DNAI2	605483	Ciliary dyskinesia, primary, 9, with or without situs inversus, 612444 (3)
DNAJB13	610263	Ciliary dyskinesia, primary, 34, 617091 (3), Autosomal recessive
DNAJB2	604139	Spinal muscular atrophy, distal, autosomal recessive, 5, 614881 (3), Autosomal recessive
DNAJB6	611332	Muscular dystrophy, limb-girdle, type 1E, 603511 (3), Autosomal dominant
DNAJC12	606060	Hyperphenylalaninemia, mild, non-BH4-deficient, 617384 (3), Autosomal recessive
DNAJC13	614334	No OMIM phenotype
DNAJC19	608977	3-methylglutaconic aciduria, type V, 610198 (3), Autosomal recessive
DNAJC21	617048	Bone marrow failure syndrome 3, 617052 (3), Autosomal recessive
DNAJC3	601184	?Ataxia, combined cerebellar and peripheral, with hearing loss and diabetes mellitus, 616192 (3), Autosomal recessive
DNAJC6	608375	Parkinson disease 19a, juvenile-onset, 615528 (3), Autosomal recessive; Parkinson disease 19b, early-onset, 615528 (3), Autosomal recessive
DNAL4	610565	?Mirror movements 3, 616059 (3), Autosomal recessive
DNASE1	125505	{Systemic lupus erythematosus, susceptibility to}, 152700 (3), Autosomal dominant
DNASE1L3	602244	Systemic lupus erythematosus 16, 614420 (3), Autosomal recessive
DNM1	602377	Epileptic encephalopathy, early infantile, 31, 616346 (3), Autosomal dominant
DNM1L	603850	Encephalopathy, lethal, due to defective mitochondrial peroxisomal fission 1, 614388 (3), Autosomal recessive, Autosomal dominant; Optic atrophy 5, 610708 (3), Autosomal dominant
DNM2	602378	Centronuclear myopathy 1, 160150 (3), Autosomal dominant; Charcot-Marie-Tooth disease, axonal type 2M, 606482 (3), Autosomal dominant; Charcot-Marie-Tooth disease, dominant intermediate B, 606482 (3), Autosomal dominant; Lethal congenital contracture syndrome 5, 615368 (3), Autosomal recessive
DNMT1	126375	Cerebellar ataxia, deafness, and narcolepsy, autosomal dominant, 604121 (3), Autosomal dominant; Neuropathy, hereditary sensory, type IE, 614116 (3), Autosomal dominant
DNMT3A	602769	Acute myeloid leukemia, somatic, 601626 (3); Tatton-Brown-Rahman syndrome, 615879 (3), Autosomal dominant
DNMT3B	602900	Immunodeficiency-centromeric instability-facial anomalies syndrome 1, 242860 (3), Autosomal recessive
DOCK2	603122	Immunodeficiency 40, 616433 (3), Autosomal recessive
DOCK6	614194	Adams-Oliver syndrome 2, 614219 (3), Autosomal recessive
DOCK8	611432	Hyper-IgE recurrent infection syndrome, autosomal recessive, 243700 (3), Autosomal recessive

DOK7	610285	?Fetal akinesia deformation sequence, 208150 (3), Autosomal recessive; Myasthenic syndrome, congenital, 10, 254300 (3), Autosomal recessive
DOLK	610746	Congenital disorder of glycosylation, type Im, 610768 (3), Autosomal recessive
DPAGT1	191350	Congenital disorder of glycosylation, type Ij, 608093 (3), Autosomal recessive; Myasthenic syndrome, congenital, 13, with tubular aggregates, 614750 (3), Autosomal recessive
DPH1	603527	Developmental delay with short stature, dysmorphic features, and sparse hair, 616901 (3), Autosomal recessive
DPM1	603503	Congenital disorder of glycosylation, type Ie, 608799 (3), Autosomal recessive
DPM2	603564	Congenital disorder of glycosylation, type Iu, 615042 (3), Autosomal recessive
DPM3	605951	Congenital disorder of glycosylation, type Io, 612937 (3)
DPP6	126141	Mental retardation, autosomal dominant 33, 616311 (3); {Ventricular fibrillation, paroxysmal familial, 2}, 612956 (3), Autosomal dominant
DPY19L2	613893	Spermatogenic failure 9, 613958 (3), Autosomal recessive
DPYD	612779	Dihydropyrimidine dehydrogenase deficiency, 274270 (3), Autosomal recessive; 5-fluorouracil toxicity, 274270 (3), Autosomal recessive
DPYS	613326	Dihydropyrimidinuria, 222748 (3), Autosomal recessive
DRAM2	613360	Cone-rod dystrophy 21, 616502 (3), Autosomal recessive
DRC1	615288	Ciliary dyskinesia, primary, 21, 615294 (3), Autosomal recessive
DRD2	126450	No OMIM phenotype
DRD3	126451	{Essential tremor, hereditary, 1}, 190300 (3), Autosomal dominant; {Schizophrenia, susceptibility to}, 181500 (3), Autosomal dominant
DRD4	126452	{Attention deficit-hyperactivity disorder}, 143465 (3), Autosomal dominant; Autonomic nervous system dysfunction (3); [Novelty seeking personality], 601696 (1), ?Autosomal dominant
DRD5	126453	{Attention deficit-hyperactivity disorder, susceptibility to}, 143465 (3), Autosomal dominant; {Blepharospasm, primary benign}, 606798 (3), Isolated cases
DSC2	125645	Arrhythmogenic right ventricular dysplasia 11, 610476 (3), Autosomal recessive, Autosomal dominant; Arrhythmogenic right ventricular dysplasia 11 with mild palmoplantar keratoderma and woolly hair, 610476 (3), Autosomal recessive, Autosomal dominant
DSG1	125670	Erythroderma, congenital, with palmoplantar keratoderma, hypotrichosis, and hyper IgE, 615508 (3), Autosomal recessive; Keratosis palmoplantaris striata I, AD, 148700 (3), Autosomal dominant
DSG2	125671	Arrhythmogenic right ventricular dysplasia 10, 610193 (3), Autosomal dominant; Cardiomyopathy, dilated, 1BB, 612877 (3)
DSG4	607892	Hypotrichosis 6, 607903 (3), Autosomal recessive

DSP	125647	Arrhythmogenic right ventricular dysplasia 8, 607450 (3), Autosomal dominant; Cardiomyopathy, dilated, with woolly hair and keratoderma, 605676 (3), Autosomal recessive; Dilated cardiomyopathy with woolly hair, keratoderma, and tooth agenesis, 615821 (3), Autosomal dominant; Epidermolysis bullosa, lethal acantholytic, 609638 (3), Autosomal recessive; Keratosis palmoplantaris striata II, 612908 (3), Autosomal dominant; Skin fragility-woolly hair syndrome, 607655 (3), Autosomal recessive
DSPP	125485	Deafness, autosomal dominant 39, with dentinogenesis, 605594 (3), Autosomal dominant; Dentin dysplasia, type II, 125420 (3), Autosomal dominant; Dentinogenesis imperfecta, Shields type II, 125490 (3), Autosomal dominant; Dentinogenesis imperfecta, Shields type III, 125500 (3), Autosomal dominant
DST	113810	Epidermolysis bullosa simplex, autosomal recessive 2, 615425 (3), Autosomal recessive; ?Neuropathy, hereditary sensory and autonomic, type VI, 614653 (3), Autosomal recessive
DSTYK	612666	Congenital anomalies of kidney and urinary tract 1, 610805 (3), Autosomal dominant; Spastic paraplegia 23, 270750 (3), Autosomal recessive
DTNA	601239	Left ventricular noncompaction 1, with or without congenital heart defects, 604169 (3), Autosomal dominant
DTNBP1	607145	Hermansky-Pudlak syndrome 7, 614076 (3), Autosomal recessive
DUOX2	606759	Thyroid dyshormonogenesis 6, 607200 (3), Autosomal recessive
DUOXA2	612772	Thyroid dyshormonogenesis 5, 274900 (3), Autosomal recessive
DUSP6	602748	Hypogonadotropic hypogonadism 19 with or without anosmia, 615269 (3), Autosomal dominant
DVL1	601365	Robinow syndrome, autosomal dominant 2, 616331 (3), Autosomal dominant
DVL3	601368	Robinow syndrome, autosomal dominant 3, 616894 (3), Autosomal dominant
DYM	607461	Dyggve-Melchior-Clausen disease, 223800 (3), Autosomal recessive; Smith-McCort dysplasia, 607326 (3), Autosomal recessive
DYNC1H1	600112	Charcot-Marie-Tooth disease, axonal, type 20, 614228 (3), Autosomal dominant; Mental retardation, autosomal dominant 13, 614563 (3), Autosomal dominant; Spinal muscular atrophy, lower extremity-predominant 1, AD, 158600 (3), Autosomal dominant
DYNC2LI1	617083	Short-rib thoracic dysplasia 15 with polydactyly, 617088 (3), Autosomal recessive
DYRK1A	600855	Mental retardation, autosomal dominant 7, 614104 (3), Autosomal dominant
DYRK1B	604556	Abdominal obesity-metabolic syndrome 3, 615812 (3), Autosomal dominant
DYSF	603009	Miyoshi muscular dystrophy 1, 254130 (3), Autosomal recessive; Muscular dystrophy, limb-girdle, type 2B, 253601 (3), Autosomal recessive; Myopathy, distal, with anterior tibial onset, 606768 (3), Autosomal recessive

EBP	300205	Chondrodysplasia punctata, X-linked dominant, 302960 (3), X-linked dominant; MEND syndrome, 300960 (3), X-linked recessive
ECE1	600423	?Hirschsprung disease, cardiac defects, and autonomic dysfunction, 613870 (3), Autosomal dominant; {Hypertension, essential, susceptibility to}, 145500 (3), Multifactorial
ECEL1	605896	Arthrogryposis, distal, type 5D, 615065 (3), Autosomal recessive
ECHS1	602292	Mitochondrial short-chain enoyl-CoA hydratase 1 deficiency, 616277 (3), Autosomal recessive
ECM1	602201	Urbach-Wiethe disease, 247100 (3), Autosomal recessive
EDA	300451	Ectodermal dysplasia 1, hypohidrotic, X-linked, 305100 (3), X-linked recessive; Tooth agenesis, selective, X-linked 1, 313500 (3), X-linked dominant
EDAR	604095	Ectodermal dysplasia 10A, hypohidrotic/hair/nail type, autosomal dominant, 129490 (3), Autosomal dominant; Ectodermal dysplasia 10B, hypohidrotic/hair/tooth type, autosomal recessive, 224900 (3), Autosomal recessive; [Hair morphology 1, hair thickness], 612630 (3)
EDARADD	606603	Ectodermal dysplasia 11A, hypohidrotic/hair/tooth type, autosomal dominant, 614940 (3), Autosomal dominant; Ectodermal dysplasia 11B, hypohidrotic/hair/tooth type, autosomal recessive, 614941 (3), Autosomal recessive
EDC3	609842	?Mental retardation, autosomal recessive 50, 616460 (3), Autosomal recessive
EDN1	131240	Auriculocondylar syndrome 3, 615706 (3), Autosomal recessive; {High density lipoprotein cholesterol level QTL 7} (3); Question mark ears, isolated, 612798 (3), Autosomal dominant
EDN3	131242	Central hypoventilation syndrome, congenital, 209880 (3), Autosomal dominant; {Hirschsprung disease, susceptibility to, 4}, 613712 (3), Autosomal dominant; Waardenburg syndrome, type 4B, 613265 (3), Autosomal recessive, Autosomal dominant
EDNRA	131243	Mandibulofacial dysostosis with alopecia, 616367 (3), Autosomal dominant; {Migraine, resistance to}, 157300 (3), Autosomal dominant
EDNRB	131244	ABCD syndrome, 600501 (3), Autosomal recessive; {Hirschsprung disease, susceptibility to, 2}, 600155 (3), Autosomal dominant; Waardenburg syndrome, type 4A, 277580 (3), Autosomal recessive, Autosomal dominant
EEF1A2	602959	Epileptic encephalopathy, early infantile, 33, 616409 (3), Autosomal dominant; Mental retardation, autosomal dominant 38, 616393 (3), Autosomal dominant
EEF2	130610	?Spinocerebellar ataxia 26, 609306 (3), Autosomal dominant
EFEMP1	601548	Doyne honeycomb degeneration of retina, 126600 (3), Autosomal dominant
EFEMP2	604633	Cutis laxa, autosomal recessive, type IB, 614437 (3), Autosomal recessive

EFHC1	608815	{Epilepsy, juvenile absence, susceptibility to, 1}, 607631 (3), Autosomal dominant; {Myoclonic epilepsy, juvenile, susceptibility to, 1}, 254770 (3), Autosomal dominant
EFNB1	300035	Craniofrontonasal dysplasia, 304110 (3), X-linked dominant
EGF	131530	Hypomagnesemia 4, renal, 611718 (3)
EGFR	131550	Adenocarcinoma of lung, response to tyrosine kinase inhibitor in, 211980 (3), Autosomal recessive; ?Inflammatory skin and bowel disease, neonatal, 2, 616069 (3), Autosomal recessive; Nonsmall cell lung cancer, response to tyrosine kinase inhibitor in, 211980 (3), Autosomal recessive; {Nonsmall cell lung cancer, susceptibility to}, 211980 (3), Autosomal recessive
EGLN1	606425	Erythrocytosis, familial, 3, 609820 (3), Autosomal dominant; [Hemoglobin, high altitude adaptation], 609070 (3), Autosomal dominant
EHBP1	609922	{Prostate cancer, hereditary, 12}, 611868 (3)
EHHADH	607037	?Fanconi renotubular syndrome 3, 615605 (3), Autosomal dominant
EHMT1	607001	Kleefstra syndrome 1, 610253 (3), Autosomal dominant
EIF2AK4	609280	Pulmonary venoocclusive disease 2, 234810 (3), Autosomal recessive
EIF2B1	606686	Leukoencephalopathy with vanishing white matter, 603896 (3), Autosomal recessive
EIF2B2	606454	Leukoencephalopathy with vanishing white matter, 603896 (3), Autosomal recessive; Ovarioleukodystrophy, 603896 (3), Autosomal recessive
EIF2B3	606273	Leukoencephalopathy with vanishing white matter, 603896 (3), Autosomal recessive
EIF2B5	603945	Leukoencephalopathy with vanishing white matter, 603896 (3), Autosomal recessive; Ovarioleukodystrophy, 603896 (3), Autosomal recessive
EIF2S3	300161	MEHMO syndrome, 300148 (3), X-linked recessive
EIF4A3	608546	Robin sequence with cleft mandible and limb anomalies, 268305 (3), Autosomal recessive
EIF4E	133440	{Autism, susceptibility to, 19}, 615091 (3)
EIF4G1	600495	{Parkinson disease 18}, 614251 (3), Autosomal dominant
ELAC2	605367	Combined oxidative phosphorylation deficiency 17, 615440 (3), Autosomal recessive; {Prostate cancer, hereditary, 2, susceptibility to}, 614731 (3)
ELANE	130130	Neutropenia, cyclic, 162800 (3), Autosomal dominant; Neutropenia, severe congenital 1, autosomal dominant, 202700 (3), Autosomal dominant
ELMO2	606421	Vascular malformation, primary intraosseous, 606893 (3), Autosomal recessive
ELN	130160	Cutis laxa, autosomal dominant, 123700 (3), Autosomal dominant; Supravalvar aortic stenosis, 185500 (3), Autosomal dominant
ELOVL5	611805	Spinocerebellar ataxia 38, 615957 (3), Autosomal dominant
ELP1	603722	Dysautonomia, familial, 223900 (3), Autosomal recessive

ELP2	616054	Mental retardation, autosomal recessive 58, 617270 (3), Autosomal recessive
ELP4	606985	?Aniridia 2, 617141 (3), Autosomal dominant
EMC1	616846	Cerebellar atrophy, visual impairment, and psychomotor retardation, 616875 (3), Autosomal recessive
EMD	300384	Emery-Dreifuss muscular dystrophy 1, X-linked, 310300 (3), X-linked recessive
EMG1	611531	Bowen-Conradi syndrome, 211180 (3), Autosomal recessive
EML1	602033	Band heterotopia, 600348 (3), Autosomal recessive
EMP2	602334	Nephrotic syndrome, type 10, 615861 (3), Autosomal recessive
EMX2	600035	Schizencephaly, 269160 (3)
ENAM	606585	Amelogenesis imperfecta, type IB, 104500 (3), Autosomal dominant; Amelogenesis imperfecta, type IC, 204650 (3), Autosomal recessive
ENG	131195	Telangiectasia, hereditary hemorrhagic, type 1, 187300 (3), Autosomal dominant
ENO3	131370	?Glycogen storage disease XIII, 612932 (3), Autosomal recessive
ENO4	No OMIM gene	No OMIM phenotype
ENPP1	173335	Arterial calcification, generalized, of infancy, 1, 208000 (3), Autosomal recessive; Cole disease, 615522 (3), Autosomal dominant; {Diabetes mellitus, non-insulin-dependent, susceptibility to}, 125853 (3), Autosomal dominant; Hypophosphatemic rickets, autosomal recessive, 2, 613312 (3), Autosomal dominant; {Obesity, susceptibility to}, 601665 (3), Autosomal recessive, Autosomal dominant, Multifactorial
ENTPD1	601752	Spastic paraplegia 64, autosomal recessive, 615683 (3), Autosomal recessive
EOGT	614789	Adams-Oliver syndrome 4, 615297 (3), Autosomal recessive
EP300	602700	Colorectal cancer, somatic, 114500 (3); Rubinstein-Taybi syndrome 2, 613684 (3), Autosomal dominant
EPAS1	603349	Erythrocytosis, familial, 4, 611783 (3)
EPB41	130500	Elliptocytosis-1, 611804 (3), Autosomal recessive, Autosomal dominant
EPCAM	185535	Colorectal cancer, hereditary nonpolyposis, type 8, 613244 (3); Diarrhea 5, with tufting enteropathy, congenital, 613217 (3), Autosomal recessive
EPG5	615068	Vici syndrome, 242840 (3), Autosomal recessive
EPHA2	176946	Cataract 6, multiple types, 116600 (3), Autosomal dominant
EPHB2	600997	{Prostate cancer/brain cancer susceptibility, somatic}, 603688 (3)
EPHB4	600011	{Hydrops fetalis, nonimmune, and/or atrial septal defect}, 617300 (3), Autosomal dominant
EPHX1	132810	?Hypercholanemia, familial, 607748 (3)
EPHX2	132811	{Hypercholesterolemia, familial, due to LDLR defect, modifier of}, 143890 (3), Autosomal dominant
EPM2A	607566	Epilepsy, progressive myoclonic 2A (Lafora), 254780 (3), Autosomal recessive

EPO	133170	?Diamond-Blackfan anemia-like, 617911 (3), Autosomal recessive; Erythrocytosis, familial, 5, 617907 (3), Autosomal dominant; {Microvascular complications of diabetes 2}, 612623 (3)
EPOR	133171	[Erythrocytosis, familial, 1], 133100 (3), Autosomal dominant
EPS8	600206	?Deafness, autosomal recessive 102, 615974 (3), Autosomal recessive
EPX	131399	[Eosinophil peroxidase deficiency], 261500 (3), Autosomal recessive
ERBB2	164870	Adenocarcinoma of lung, somatic, 211980 (3); Gastric cancer, somatic, 613659 (3); Glioblastoma, somatic, 137800 (3); Ovarian cancer, somatic (3)
ERBB3	190151	Lethal congenital contractural syndrome 2, 607598 (3), Autosomal recessive
ERBB4	600543	Amyotrophic lateral sclerosis 19, 615515 (3), Autosomal dominant
ERCC1	126380	Cerebrooculofacioskeletal syndrome 4, 610758 (3), Autosomal recessive
ERCC2	126340	?Cerebrooculofacioskeletal syndrome 2, 610756 (3), Autosomal recessive; Trichothiodystrophy 1, photosensitive, 601675 (3), Autosomal recessive; Xeroderma pigmentosum, group D, 278730 (3), Autosomal recessive
ERCC3	133510	Trichothiodystrophy 2, photosensitive, 616390 (3), Autosomal recessive; Xeroderma pigmentosum, group B, 610651 (3), Autosomal recessive
ERCC4	133520	Fanconi anemia, complementation group Q, 615272 (3), Autosomal recessive; ?XFE progeroid syndrome, 610965 (3); Xeroderma pigmentosum, group F, 278760 (3), Autosomal recessive; Xeroderma pigmentosum, type F/Cockayne syndrome, 278760 (3), Autosomal recessive
ERCC5	133530	Cerebrooculofacioskeletal syndrome 3, 616570 (3), Autosomal recessive; Xeroderma pigmentosum, group G, 278780 (3), Autosomal recessive; Xeroderma pigmentosum, group G/Cockayne syndrome, 278780 (3), Autosomal recessive
ERCC6	609413	Cerebrooculofacioskeletal syndrome 1, 214150 (3), Autosomal recessive; Cockayne syndrome, type B, 133540 (3), Autosomal recessive; De Sanctis-Cacchione syndrome, 278800 (3), Autosomal recessive; {Lung cancer, susceptibility to}, 211980 (3), Autosomal recessive; {Macular degeneration, age-related, susceptibility to, 5}, 613761 (3); Premature ovarian failure 11, 616946 (3), Autosomal dominant; UV-sensitive syndrome 1, 600630 (3), Autosomal recessive
ERCC6L2	615667	Bone marrow failure syndrome 2, 615715 (3), Autosomal recessive
ERCC8	609412	Cockayne syndrome, type A, 216400 (3), Autosomal recessive; UV-sensitive syndrome 2, 614621 (3), Autosomal recessive
ERLIN1	611604	Spastic paraplegia 62, 615681 (3), Autosomal recessive
ERLIN2	611605	Spastic paraplegia 18, autosomal recessive, 611225 (3), Autosomal recessive
ERMAP	609017	[Blood group, Radin], 111620 (3); [Blood group, Scianna system], 111750 (3)

ERMARD	615532	?Periventricular nodular heterotopia 6, 615544 (3), Autosomal dominant
ESCO2	609353	Roberts syndrome, 268300 (3), Autosomal recessive; SC phocomelia syndrome, 269000 (3), Autosomal recessive
ESPN	606351	Deafness, autosomal recessive 36, 609006 (3), Autosomal recessive; Deafness, neurosensory, without vestibular involvement, autosomal dominant (3)
ESS2	601755	No OMIM phenotype
ETFA	608053	Glutaric acidemia IIA, 231680 (3), Autosomal recessive
ETFB	130410	Glutaric acidemia IIB, 231680 (3), Autosomal recessive
ETFDH	231675	Glutaric acidemia IIC, 231680 (3), Autosomal recessive
ETHE1	608451	Ethylmalonic encephalopathy, 602473 (3), Autosomal recessive
ETV6	600618	Leukemia, acute myeloid, somatic, 601626 (3); Thrombocytopenia 5, 616216 (3), Autosomal dominant
EVC	604831	Ellis-van Creveld syndrome, 225500 (3), Autosomal recessive; ?Weyers acrofacial dysostosis, 193530 (3), Autosomal dominant
EVC2	607261	Ellis-van Creveld syndrome, 225500 (3), Autosomal recessive; Weyers acrofacial dysostosis, 193530 (3), Autosomal dominant
EWSR1	133450	Ewing sarcoma, 612219 (3); Neuroepithelioma, 612219 (3)
EXOSC3	606489	Pontocerebellar hypoplasia, type 1B, 614678 (3), Autosomal recessive
EXOSC8	606019	Pontocerebellar hypoplasia, type 1C, 616081 (3), Autosomal recessive
EXT1	608177	Chondrosarcoma, 215300 (3), Autosomal recessive; Exostoses, multiple, type 1, 133700 (3), Autosomal dominant
EYA1	601653	Anterior segment anomalies with or without cataract, 602588 (3), Autosomal dominant; Branchiootic syndrome 1, 602588 (3), Autosomal dominant; Branchiootorenal syndrome 1, with or without cataracts, 113650 (3), Autosomal dominant; ?Otofaciocervical syndrome, 166780 (3), Autosomal dominant
EYA4	603550	?Cardiomyopathy, dilated, 1J, 605362 (3), Autosomal dominant; Deafness, autosomal dominant 10, 601316 (3), Autosomal dominant
EYS	612424	Retinitis pigmentosa 25, 602772 (3), Autosomal recessive
F10	613872	Factor X deficiency, 227600 (3), Autosomal recessive
F11	264900	Factor XI deficiency, autosomal dominant, 612416 (3); Factor XI deficiency, autosomal recessive, 612416 (3)
F12	610619	Angioedema, hereditary, type III, 610618 (3), Autosomal dominant; Factor XII deficiency, 234000 (3), Autosomal recessive
F13A1	134570	Factor XIIIA deficiency, 613225 (3), Autosomal recessive; {Myocardial infarction, protection against}, 608446 (3); {Venous thrombosis, protection against}, 188050 (3), Autosomal dominant
F13B	134580	Factor XIIIB deficiency, 613235 (3), Autosomal recessive

F2	176930	Dysprothrombinemia, 613679 (3), Autosomal recessive; Hypoprothrombinemia, 613679 (3), Autosomal recessive; {Pregnancy loss, recurrent, susceptibility to, 2}, 614390 (3), Autosomal dominant; {Stroke, ischemic, susceptibility to}, 601367 (3), Multifactorial; Thrombophilia due to thrombin defect, 188050 (3), Autosomal dominant
F5	612309	{Budd-Chiari syndrome}, 600880 (3), Autosomal recessive; Factor V deficiency, 227400 (3), Autosomal recessive; {Pregnancy loss, recurrent, susceptibility to, 1}, 614389 (3), Autosomal dominant; {Stroke, ischemic, susceptibility to}, 601367 (3), Multifactorial; Thrombophilia due to activated protein C resistance, 188055 (3), Autosomal dominant; {Thrombophilia, susceptibility to, due to factor V Leiden}, 188055 (3), Autosomal dominant
F7	613878	Factor VII deficiency, 227500 (3), Autosomal recessive; {Myocardial infarction, decreased susceptibility to}, 608446 (3)
F8	300841	Hemophilia A, 306700 (3), X-linked recessive
F9	300746	{Deep venous thrombosis, protection against}, 300807 (3); Hemophilia B, 306900 (3), X-linked recessive; Thrombophilia, X-linked, due to factor IX defect, 300807 (3); {Warfarin sensitivity}, 122700 (3), Autosomal dominant
FA2H	611026	Spastic paraplegia 35, autosomal recessive, 612319 (3), Autosomal recessive
FAAH	602935	{Drug addiction, susceptibility to}, 606581 (3)
FADD	602457	Infections, recurrent, with encephalopathy, hepatic dysfunction, and cardiovascular malformations, 613759 (3), Autosomal recessive
FAH	613871	Tyrosinemia, type I, 276700 (3), Autosomal recessive
FAM111A	615292	Gracile bone dysplasia, 602361 (3), Autosomal dominant; Kenny-Caffey syndrome, type 2, 127000 (3), Autosomal dominant
FAM111B	615584	Poikiloderma, hereditary fibrosing, with tendon contractures, myopathy, and pulmonary fibrosis, 615704 (3), Autosomal dominant
FAM126A	610531	Leukodystrophy, hypomyelinating, 5, 610532 (3), Autosomal recessive
FAM161A	613596	Retinitis pigmentosa 28, 606068 (3)
FAM187A	No OMIM gene	No OMIM phenotype
FAM20A	611062	Amelogenesis imperfecta, type IG (enamel-renal syndrome), 204690 (3), Autosomal recessive
FAM20C	611061	Raine syndrome, 259775 (3), Autosomal recessive
FAM83H	611927	Amelogenesis imperfecta, type IIIA, 130900 (3), Autosomal dominant
FAN1	613534	Interstitial nephritis, karyomegalic, 614817 (3), Autosomal recessive
FANCA	607139	Fanconi anemia, complementation group A, 227650 (3), Autosomal recessive
FANCC	613899	Fanconi anemia, complementation group C, 227645 (3), Autosomal recessive
FANCD2	613984	Fanconi anemia, complementation group D2, 227646 (3), Autosomal recessive
FANCE	613976	Fanconi anemia, complementation group E, 600901 (3), Autosomal recessive

FANCF	613897	Fanconi anemia, complementation group F, 603467 (3)
FANCG	602956	Fanconi anemia, complementation group G, 614082 (3)
FANCI	611360	Fanconi anemia, complementation group I, 609053 (3), Autosomal recessive
FANCL	608111	Fanconi anemia, complementation group L, 614083 (3), Autosomal recessive
FANCM	609644	No OMIM phenotype
FAR1	616107	Peroxisomal fatty acyl-CoA reductase 1 disorder, 616154 (3), Autosomal recessive
FARS2	611592	Combined oxidative phosphorylation deficiency 14, 614946 (3), Autosomal recessive; Spastic paraplegia 77, autosomal recessive, 617046 (3), Autosomal recessive
FAS	134637	Autoimmune lymphoproliferative syndrome, type IA, 601859 (3), Autosomal dominant; {Autoimmune lymphoproliferative syndrome}, 601859 (3), Autosomal dominant; Squamous cell carcinoma, burn scar-related, somatic (3)
FASLG	134638	Autoimmune lymphoproliferative syndrome, type IB, 601859 (3), Autosomal dominant; {Lung cancer, susceptibility to}, 211980 (3), Autosomal recessive
FASTKD2	612322	?Mitochondrial complex IV deficiency, 220110 (3), Autosomal recessive, Mitochondrial
FAT4	612411	Hennekam lymphangiectasia-lymphedema syndrome 2, 616006 (3), Autosomal recessive; Van Maldergem syndrome 2, 615546 (3), Autosomal recessive
FBLN1	135820	Synpolydactyly, 3/3'4, associated with metacarpal and metatarsal synostoses, 608180 (4), Autosomal dominant
FBLN5	604580	Cutis laxa, autosomal dominant 2, 614434 (3); Cutis laxa, autosomal recessive, type IA, 219100 (3), Autosomal recessive; Macular degeneration, age-related, 3, 608895 (3), Autosomal dominant; Neuropathy, hereditary, with or without age-related macular degeneration, 608895 (3), Autosomal dominant
FBN1	134797	Acromicric dysplasia, 102370 (3), Autosomal dominant; Ectopia lentis, familial, 129600 (3), Autosomal dominant; Geleophysic dysplasia 2, 614185 (3), Autosomal dominant; MASS syndrome, 604308 (3); Marfan lipodystrophy syndrome, 616914 (3), Autosomal dominant; Marfan syndrome, 154700 (3), Autosomal dominant; Stiff skin syndrome, 184900 (3), Autosomal dominant; Weill-Marchesani syndrome 2, dominant, 608328 (3), Autosomal dominant
FBN2	612570	Contractural arachnodactyly, congenital, 121050 (3), Autosomal dominant; Macular degeneration, early-onset, 616118 (3), Autosomal dominant
FBP1	611570	Fructose-1,6-bisphosphatase deficiency, 229700 (3), Autosomal recessive
FBXL4	605654	Mitochondrial DNA depletion syndrome 13 (encephalomyopathic type), 615471 (3), Autosomal recessive

FBXO31	609102	?Mental retardation, autosomal recessive 45, 615979 (3), Autosomal recessive
FBXO38	608533	Neuronopathy, distal hereditary motor, type IID, 615575 (3), Autosomal dominant
FBXO7	605648	Parkinson disease 15, autosomal recessive, 260300 (3), Autosomal recessive
FCGR2A	146790	{Lupus nephritis, susceptibility to}, 152700 (3), Autosomal dominant; {Malaria, severe, susceptibility to}, 611162 (3); {Pseudomonas aeruginosa, susceptibility to chronic infection by, in cystic fibrosis}, 219700 (3), Autosomal recessive
FCGR2B	604590	{Malaria, resistance to}, 611162 (3); {Systemic lupus erythematosus, susceptibility to}, 152700 (3), Autosomal dominant
FCGR3A	146740	Immunodeficiency 20, 615707 (3), Autosomal recessive
FCN3	604973	Immunodeficiency due to ficolin 3 deficiency, 613860 (3), Autosomal recessive
FDPS	134629	Porokeratosis 9, multiple types, 616631 (3), Autosomal dominant
FECH	612386	Protoporphyria, erythropoietic, 1, 177000 (3), Autosomal recessive
FERMT1	607900	Kindler syndrome, 173650 (3), Autosomal recessive
FERMT3	607901	Leukocyte adhesion deficiency, type III, 612840 (3), Autosomal recessive
FEZF1	613301	Hypogonadotropic hypogonadism 22, with or without anosmia, 616030 (3), Autosomal recessive
FFAR4	609044	{Obesity, susceptibility to}, 607514 (3)
FGA	134820	Afibrinogenemia, congenital, 202400 (3), Autosomal recessive; Amyloidosis, familial visceral, 105200 (3), Autosomal dominant; Dysfibrinogenemia, congenital, 616004 (3); Hypodysfibrinogenemia, congenital, 616004 (3)
FGB	134830	Afibrinogenemia, congenital, 202400 (3), Autosomal recessive; Dysfibrinogenemia, congenital, 616004 (3); Hypofibrinogenemia, congenital, 202400 (3), Autosomal recessive
FGD1	300546	Aarskog-Scott syndrome, 305400 (3), X-linked recessive; Mental retardation, X-linked syndromic 16, 305400 (3), X-linked recessive
FGD4	611104	Charcot-Marie-Tooth disease, type 4H, 609311 (3), Autosomal recessive
FGF10	602115	Aplasia of lacrimal and salivary glands, 180920 (3), Autosomal dominant; LADD syndrome, 149730 (3), Autosomal dominant
FGF14	601515	Spinocerebellar ataxia 27, 609307 (3), Autosomal dominant
FGF16	300827	Metacarpal 4-5 fusion, 309630 (3), X-linked recessive
FGF17	603725	Hypogonadotropic hypogonadism 20 with or without anosmia, 615270 (3), Autosomal dominant
FGF20	605558	?Renal hypodysplasia/aplasia 2, 615721 (3), Autosomal recessive
FGF23	605380	Hypophosphatemic rickets, autosomal dominant, 193100 (3), Autosomal dominant; Osteomalacia, tumor-induced (1); Tumoral calcinosis, hyperphosphatemic, familial, 2, 617993 (3)

FGF3	164950	Deafness, congenital with inner ear agenesis, microtia, and microdontia, 610706 (3), Autosomal recessive
FGF5	165190	Trichomegaly, 190330 (3), Autosomal recessive
FGF8	600483	Hypogonadotropic hypogonadism 6 with or without anosmia, 612702 (3), Autosomal dominant
FGF9	600921	Multiple synostoses syndrome 3, 612961 (3), Autosomal dominant
FGFR2	176943	Antley-Bixler syndrome without genital anomalies or disordered steroidogenesis, 207410 (3), Autosomal dominant; Apert syndrome, 101200 (3), Autosomal dominant; Beare-Stevenson cutis gyrata syndrome, 123790 (3), Autosomal dominant; Bent bone dysplasia syndrome, 614592 (3), Autosomal dominant; Craniofacial-skeletal-dermatologic dysplasia, 101600 (3), Autosomal dominant; Craniosynostosis, nonspecific (3); Crouzon syndrome, 123500 (3), Autosomal dominant; Gastric cancer, somatic, 613659 (3); Jackson-Weiss syndrome, 123150 (3), Autosomal dominant; LADD syndrome, 149730 (3), Autosomal dominant; Pfeiffer syndrome, 101600 (3), Autosomal dominant; Saethre-Chotzen syndrome, 101400 (3), Autosomal dominant; Scaphocephaly and Axenfeld-Rieger anomaly (3); Scaphocephaly, maxillary retrusion, and mental retardation, 609579 (3)
FGFR3	134934	Achondroplasia, 100800 (3), Autosomal dominant; Bladder cancer, somatic, 109800 (3); CATSHL syndrome, 610474 (3), Autosomal recessive, Autosomal dominant; Cervical cancer, somatic, 603956 (3); Colorectal cancer, somatic, 114500 (3); Crouzon syndrome with acanthosis nigricans, 612247 (3), Autosomal dominant; Hypochondroplasia, 146000 (3), Autosomal dominant; LADD syndrome, 149730 (3), Autosomal dominant; Muenke syndrome, 602849 (3), Autosomal dominant; Nevus, epidermal, somatic, 162900 (3); SADDAN, 616482 (3), Autosomal dominant; Spermatocytic seminoma, somatic, 273300 (3); Thanatophoric dysplasia, type I, 187600 (3), Autosomal dominant; Thanatophoric dysplasia, type II, 187601 (3), Autosomal dominant
FGFRL1	605830	No OMIM phenotype
FGG	134850	Afibrinogenemia, congenital, 202400 (3), Autosomal recessive; Dysfibrinogenemia, congenital, 616004 (3); Hypodysfibrinogenemia, 616004 (3); Hypofibrinogenemia, congenital, 202400 (3), Autosomal recessive
FH	136850	Fumarase deficiency, 606812 (3), Autosomal recessive; Leiomyomatosis and renal cell cancer, 150800 (3), Autosomal dominant

FHL1	300163	Emery-Dreifuss muscular dystrophy 6, X-linked, 300696 (3), X-linked recessive; Myopathy, X-linked, with postural muscle atrophy, 300696 (3), X-linked recessive; Reducing body myopathy, X-linked 1a, severe, infantile or early childhood onset, 300717 (3), X-linked dominant; Reducing body myopathy, X-linked 1b, with late childhood or adult onset, 300718 (3), X-linked; Scapuloperoneal myopathy, X-linked dominant, 300695 (3), X-linked dominant; ?Uruguay faciocardiomusculoskeletal syndrome, 300280 (3), X-linked recessive
FIBP	608296	Thauvin-Robinet-Faivre syndrome, 617107 (3), Autosomal recessive
FIG4	609390	Amyotrophic lateral sclerosis 11, 612577 (3), Autosomal dominant; Charcot-Marie-Tooth disease, type 4J, 611228 (3), Autosomal recessive; ?Polymicrogyria, bilateral temporooccipital, 612691 (3), Autosomal recessive; Yunis-Varon syndrome, 216340 (3), Autosomal recessive
FIGLA	608697	Premature ovarian failure 6, 612310 (3), Autosomal dominant
FKBP10	607063	Bruck syndrome 1, 259450 (3), Autosomal recessive; Osteogenesis imperfecta, type XI, 610968 (3), Autosomal recessive
FKBP14	614505	Ehlers-Danlos syndrome, kyphoscoliotic type, 2, 614557 (3), Autosomal recessive
FKBP5	602623	{Major depressive disorder and accelerated response to antidepressant drug treatment}, 608516 (3)
FLAD1	610595	Lipid storage myopathy due to flavin adenine dinucleotide synthetase deficiency, 255100 (3), Autosomal recessive
FLCN	607273	Birt-Hogg-Dube syndrome, 135150 (3), Autosomal dominant; Colorectal cancer, somatic, 114500 (3); Pneumothorax, primary spontaneous, 173600 (3), Autosomal dominant; Renal carcinoma, chromophobe, somatic, 144700 (3)
FLG	135940	{Dermatitis, atopic, susceptibility to, 2}, 605803 (3); Ichthyosis vulgaris, 146700 (3), Autosomal dominant
FLNA	300017	Cardiac valvular dysplasia, X-linked, 314400 (3), X-linked recessive; Congenital short bowel syndrome, 300048 (3), X-linked recessive; ?FG syndrome 2, 300321 (3), X-linked; Frontometaphyseal dysplasia 1, 305620 (3), X-linked recessive; Heterotopia, periventricular, 300049 (3), X-linked dominant; Intestinal pseudoobstruction, neuronal, 300048 (3), X-linked recessive; Melnick-Needles syndrome, 309350 (3), X-linked dominant; Otopalatodigital syndrome, type I, 311300 (3), X-linked dominant; Otopalatodigital syndrome, type II, 304120 (3), X-linked dominant; Terminal osseous dysplasia, 300244 (3), X-linked dominant
FLNB	603381	Atelosteogenesis, type I, 108720 (3), Autosomal dominant; Atelosteogenesis, type III, 108721 (3), Autosomal dominant; Boomerang dysplasia, 112310 (3), Autosomal dominant; Larsen syndrome, 150250 (3), Autosomal dominant; Spondylocarpotarsal synostosis syndrome, 272460 (3), Autosomal recessive

FLNC	102565	Cardiomyopathy, familial hypertrophic, 26 (3); Cardiomyopathy, familial restrictive 5, 617047 (3), Autosomal dominant; Myopathy, distal, 4, 614065 (3), Autosomal dominant; Myopathy, myofibrillar, 5, 609524 (3), Autosomal dominant
FLRT3	604808	Hypogonadotropic hypogonadism 21 with anosmia, 615271 (3), Autosomal dominant
FLT3	136351	Leukemia, acute lymphoblastic, somatic, 613065 (3); Leukemia, acute myeloid, reduced survival in, somatic, 601626 (3); Leukemia, acute myeloid, somatic, 601626 (3)
FLVCR1	609144	Ataxia, posterior column, with retinitis pigmentosa, 609033 (3), Autosomal recessive
FLVCR2	610865	Proliferative vasculopathy and hydranencephaly-hydrocephaly syndrome, 225790 (3), Autosomal recessive
FMN2	606373	Mental retardation, autosomal recessive 47, 616193 (3), Autosomal recessive
FMO3	136132	Trimethylaminuria, 602079 (3), Autosomal recessive
FMR1	309550	Fragile X syndrome, 300624 (3), X-linked dominant; Fragile X tremor/ataxia syndrome, 300623 (3), X-linked dominant; Premature ovarian failure 1, 311360 (3), X-linked
FN1	135600	Glomerulopathy with fibronectin deposits 2, 601894 (3), Autosomal dominant; Plasma fibronectin deficiency, 614101 (1), Autosomal dominant; Spondylometaphyseal dysplasia, corner fracture type, 184255 (3), Autosomal dominant
FOLR1	136430	Neurodegeneration due to cerebral folate transport deficiency, 613068 (3), Autosomal recessive
FOXC1	601090	Anterior segment dysgenesis 3, multiple subtypes, 601631 (3), Autosomal dominant; Axenfeld-Rieger syndrome, type 3, 602482 (3), Autosomal dominant
FOXC2	602402	Lymphedema-distichiasis syndrome, 153400 (3), Autosomal dominant; Lymphedema-distichiasis syndrome with renal disease and diabetes mellitus, 153400 (3), Autosomal dominant
FOXD3	611539	{Autoimmune disease, susceptibility to, 1}, 607836 (3), Autosomal dominant
FOXE1	602617	Bamforth-Lazarus syndrome, 241850 (3), Autosomal recessive; {Thyroid cancer, nonmedullary, 4}, 616534 (3), Autosomal dominant
FOXE3	601094	Anterior segment dysgenesis 2, multiple subtypes, 610256 (3), Autosomal recessive; {Aortic aneurysm, familial thoracic 11, susceptibility to}, 617349 (3), Autosomal dominant; Cataract 34, multiple types, 612968 (3)
FOXF1	601089	Alveolar capillary dysplasia with misalignment of pulmonary veins, 265380 (3), Autosomal dominant
FOXG1	164874	Rett syndrome, congenital variant, 613454 (3), Autosomal dominant
FOXI1	601093	Enlarged vestibular aqueduct, 600791 (3), Autosomal recessive

FOXL2	605597	Blepharophimosis, epicanthus inversus, and ptosis, type 1, 110100 (3), Autosomal dominant; Blepharophimosis, epicanthus inversus, and ptosis, type 2, 110100 (3), Autosomal dominant; Premature ovarian failure 3, 608996 (3), Autosomal dominant
FOXO1	136533	Rhabdomyosarcoma, alveolar, 268220 (3), Autosomal recessive
FOXP1	605515	Mental retardation with language impairment and with or without autistic features, 613670 (3), Autosomal dominant
FOXP2	605317	Speech-language disorder-1, 602081 (3), Autosomal dominant
FOXP3	300292	{Diabetes mellitus, type I, susceptibility to}, 222100 (3), Autosomal recessive; Immunodysregulation, polyendocrinopathy, and enteropathy, X-linked, 304790 (3), X-linked recessive
FOXRED1	613622	Leigh syndrome due to mitochondrial complex I deficiency, 256000 (3), Autosomal recessive, Mitochondrial; Mitochondrial complex I deficiency, 252010 (3), Autosomal recessive, X-linked dominant, Mitochondrial
FRAS1	607830	Fraser syndrome 1, 219000 (3), Autosomal recessive
FREM2	608945	Fraser syndrome 2, 617666 (3), Autosomal recessive
FRG1	601278	No OMIM phenotype
FRMD4A	616305	?Corpus callosum, agenesis of, with facial anomalies and cerebellar ataxia, 616819 (3), Autosomal recessive
FRMD7	300628	Nystagmus 1, congenital, X-linked, 310700 (3), X-linked; Nystagmus, infantile periodic alternating, X-linked, 310700 (3), X-linked
FRMPD4	300838	Mental retardation, X-linked 104, 300983 (3), X-linked recessive
FRRS1L	604574	Epileptic encephalopathy, early infantile, 37, 616981 (3), Autosomal recessive
FRZB	605083	{Osteoarthritis susceptibility 1}, 165720 (3), Multifactorial
FSHB	136530	Hypogonadotropic hypogonadism 24 without anosmia, 229070 (3), Autosomal recessive
FSHR	136435	Ovarian dysgenesis 1, 233300 (3), Autosomal recessive; Ovarian hyperstimulation syndrome, 608115 (3), Autosomal dominant; Ovarian response to FSH stimulation, 276400 (3), Autosomal recessive
FTCD	606806	Glutamate formiminotransferase deficiency, 229100 (3), Autosomal recessive
FTH1	134770	?Hemochromatosis, type 5, 615517 (3), Autosomal dominant
FTL	134790	Hyperferritinemia-cataract syndrome, 600886 (3), Autosomal dominant; L-ferritin deficiency, dominant and recessive, 615604 (3), Autosomal recessive, Autosomal dominant; Neurodegeneration with brain iron accumulation 3, 606159 (3), Autosomal dominant
FTSJ1	300499	Mental retardation, X-linked 9/44, 309549 (3), X-linked recessive
FUCA1	612280	Fucosidosis, 230000 (3), Autosomal recessive
FUS	137070	Amyotrophic lateral sclerosis 6, with or without frontotemporal dementia, 608030 (3); Essential tremor, hereditary, 4, 614782 (3), Autosomal dominant
FUT1	211100	[Bombay phenotype], 616754 (3), Autosomal recessive

FUT2	182100	[Bombay phenotype] (3); {Norwalk virus infection, resistance to} (3); {Vitamin B12 plasma level QTL1}, 612542 (3)
FUT6	136836	Fucosyltransferase 6 deficiency, 613852 (3)
FUZ	610622	Neural tube defects, 182940 (3), Autosomal dominant
FXN	606829	Friedreich ataxia, 229300 (3), Autosomal recessive; Friedreich ataxia with retained reflexes, 229300 (3), Autosomal recessive
FXYD2	601814	Hypomagnesemia 2, renal, 154020 (3), Autosomal dominant
FYCO1	607182	Cataract 18, autosomal recessive, 610019 (3), Autosomal recessive
FZD4	604579	Exudative vitreoretinopathy 1, 133780 (3), Autosomal dominant; Retinopathy of prematurity, 133780 (3), Autosomal dominant
FZD6	603409	Nail disorder, nonsyndromic congenital, 10, (claw-shaped nails), 614157 (3), Autosomal recessive
G6PC	613742	Glycogen storage disease Ia, 232200 (3), Autosomal recessive
G6PC3	611045	Dursun syndrome, 612541 (3), Autosomal recessive; Neutropenia, severe congenital 4, autosomal recessive, 612541 (3), Autosomal recessive
G6PD	305900	Hemolytic anemia, G6PD deficient (favism), 300908 (3), X-linked dominant; {Resistance to malaria due to G6PD deficiency}, 611162 (3)
GAA	606800	Glycogen storage disease II, 232300 (3), Autosomal recessive
GABBR2	607340	Epileptic encephalopathy, early infantile, 59, 617904 (3), Autosomal dominant; Neurodevelopmental disorder with poor language and loss of hand skills, 617903 (3), Autosomal dominant; {Nicotine dependence, protection against}, 188890 (3); {Nicotine dependence, susceptibility to}, 188890 (3)
GABRA1	137160	{Epilepsy, childhood absence, susceptibility to, 4}, 611136 (3); {Epilepsy, juvenile myoclonic, susceptibility to, 5}, 611136 (3); Epileptic encephalopathy, early infantile, 19, 615744 (3), Autosomal dominant
GABRA2	137140	{Alcohol dependence, susceptibility to}, 103780 (3), Multifactorial
GABRB1	137190	Epileptic encephalopathy, early infantile, 45, 617153 (3), Autosomal dominant
GABRB3	137192	{Epilepsy, childhood absence, susceptibility to, 5}, 612269 (3); Epileptic encephalopathy, early infantile, 43, 617113 (3), Autosomal dominant
GABRD	137163	{Epilepsy, generalized, with febrile seizures plus, type 5, susceptibility to}, 613060 (3), Autosomal dominant; {Epilepsy, idiopathic generalized, 10}, 613060 (3), Autosomal dominant; {Epilepsy, juvenile myoclonic, susceptibility to}, 613060 (3), Autosomal dominant
GABRG2	137164	{Epilepsy, childhood absence, susceptibility to, 2}, 607681 (3), Autosomal dominant; Epilepsy, generalized, with febrile seizures plus, type 3, 611277 (3), Autosomal dominant; Febrile seizures, familial, 8, 611277 (3), Autosomal dominant
GAD1	605363	?Cerebral palsy, spastic quadriplegic, 1, 603513 (3), Autosomal recessive
GAL	137035	?Epilepsy, familial temporal lobe, 8, 616461 (3), Autosomal dominant
GALE	606953	Galactose epimerase deficiency, 230350 (3), Autosomal recessive

GALK1	604313	Galactokinase deficiency with cataracts, 230200 (3), Autosomal recessive
GALK2 (GK2)	137028	No OMIM phenotype
GALNS	612222	Mucopolysaccharidosis IVA, 253000 (3), Autosomal recessive
GALNT12	610290	{Colorectal cancer, susceptibility to, 1}, 608812 (3)
GALT	606999	Galactosemia, 230400 (3), Autosomal recessive
GAMT	601240	Cerebral creatine deficiency syndrome 2, 612736 (3), Autosomal recessive
GAN	605379	Giant axonal neuropathy-1, 256850 (3), Autosomal recessive
GARS	600287	Charcot-Marie-Tooth disease, type 2D, 601472 (3), Autosomal dominant; Neuropathy, distal hereditary motor, type VA, 600794 (3), Autosomal dominant
GAS8	605178	Ciliary dyskinesia, primary, 33, 616726 (3), Autosomal recessive
GATA1	305371	Anemia, X-linked, with/without neutropenia and/or platelet abnormalities, 300835 (3), X-linked recessive; Leukemia, megakaryoblastic, with or without Down syndrome, somatic, 190685 (3); Thrombocytopenia with beta-thalassemia, X-linked, 314050 (3), X-linked recessive; Thrombocytopenia, X-linked, with or without dyserythropoietic anemia, 300367 (3), X-linked recessive
GATA2	137295	Emberger syndrome, 614038 (3), Autosomal dominant; Immunodeficiency 21, 614172 (3), Autosomal dominant; {Leukemia, acute myeloid, susceptibility to}, 601626 (3), Autosomal dominant; {Myelodysplastic syndrome, susceptibility to}, 614286 (3)
GATA3	131320	Hypoparathyroidism, sensorineural deafness, and renal dysplasia, 146255 (3), Autosomal dominant
GATA4	600576	Atrial septal defect 2, 607941 (3), Autosomal dominant; Atrioventricular septal defect 4, 614430 (3), Autosomal dominant; ?Testicular anomalies with or without congenital heart disease, 615542 (3), Autosomal dominant; Tetralogy of Fallot, 187500 (3), Autosomal dominant; Ventricular septal defect 1, 614429 (3), Autosomal dominant
GATA6	601656	Atrial septal defect 9, 614475 (3), Autosomal dominant; Atrioventricular septal defect 5, 614474 (3), Autosomal dominant; Pancreatic agenesis and congenital heart defects, 600001 (3), Autosomal dominant; Persistent truncus arteriosus, 217095 (3); Tetralogy of Fallot, 187500 (3), Autosomal dominant
GATAD1	614518	?Cardiomyopathy, dilated, 2B, 614672 (3), Autosomal recessive
GATAD2B	614998	Mental retardation, autosomal dominant 18, 615074 (3), Autosomal dominant
GATM	602360	Cerebral creatine deficiency syndrome 3, 612718 (3), Autosomal recessive

GBA	606463	Gaucher disease, perinatal lethal, 608013 (3), Autosomal recessive; Gaucher disease, type I, 230800 (3), Autosomal recessive; Gaucher disease, type II, 230900 (3), Autosomal recessive; Gaucher disease, type III, 231000 (3), Autosomal recessive; Gaucher disease, type IIIC, 231005 (3), Autosomal recessive; {Lewy body dementia, susceptibility to}, 127750 (3), Autosomal dominant; {Parkinson disease, late-onset, susceptibility to}, 168600 (3), Isolated cases, Multifactorial
GBE1	607839	Glycogen storage disease IV, 232500 (3), Autosomal recessive; Polyglucosan body disease, adult form, 263570 (3), Autosomal recessive
GCGR	138033	{Diabetes mellitus, noninsulin-dependent}, 125853 (3), Autosomal dominant
GCH1	600225	Dystonia, DOPA-responsive, with or without hyperphenylalaninemia, 128230 (3), Autosomal recessive, Autosomal dominant; Hyperphenylalaninemia, BH4-deficient, B, 233910 (3), Autosomal recessive
GCK	138079	Diabetes mellitus, noninsulin-dependent, late onset, 125853 (3), Autosomal dominant; Diabetes mellitus, permanent neonatal, 606176 (3), Autosomal recessive, Autosomal dominant; Hyperinsulinemic hypoglycemia, familial, 3, 602485 (3), Autosomal dominant; MODY, type II, 125851 (3), Autosomal dominant
GCKR	600842	[Fasting plasma glucose level QTL 5], 613463 (3)
GCLC	606857	Hemolytic anemia due to gamma-glutamylcysteine synthetase deficiency, 230450 (3), Autosomal recessive; {Myocardial infarction, susceptibility to}, 608446 (3)
GCLM	601176	{Myocardial infarction, susceptibility to}, 608446 (3)
GCM2	603716	Hyperparathyroidism 4, 617343 (3), Autosomal dominant; Hypoparathyroidism, familial isolated, 146200 (3), Autosomal dominant
GCNT2	600429	Adult i phenotype without cataract, 110800 (3), Autosomal dominant; [Blood group, Ii], 110800 (3), Autosomal dominant; Cataract 13 with adult i phenotype, 116700 (3), Autosomal recessive
GCSH	238330	?Glycine encephalopathy, 605899 (3), Autosomal recessive
GDAP1	606598	Charcot-Marie-Tooth disease, axonal, type 2K, 607831 (3), Autosomal recessive, Autosomal dominant; Charcot-Marie-Tooth disease, axonal, with vocal cord paresis, 607706 (3), Autosomal recessive; Charcot-Marie-Tooth disease, recessive intermediate, A, 608340 (3), Autosomal recessive; Charcot-Marie-Tooth disease, type 4A, 214400 (3), Autosomal recessive
GDF2	605120	Telangiectasia, hereditary hemorrhagic, type 5, 615506 (3), Autosomal dominant
GDF3	606522	Klippel-Feil syndrome 3, autosomal dominant, 613702 (3); Microphthalmia with coloboma 6, 613703 (3), Autosomal dominant; Microphthalmia, isolated 7, 613704 (3), Autosomal dominant

GDF5	601146	?Acromesomelic dysplasia, Hunter-Thompson type, 201250 (3), Autosomal recessive; Brachydactyly, type A1, C, 615072 (3), Autosomal recessive, Autosomal dominant; Brachydactyly, type A2, 112600 (3), Autosomal dominant; Brachydactyly, type C, 113100 (3), Autosomal dominant; Chondrodysplasia, Grebe type, 200700 (3), Autosomal recessive; Du Pan syndrome, 228900 (3), Autosomal recessive; Multiple synostoses syndrome 2, 610017 (3), Autosomal dominant; {Osteoarthritis-5}, 612400 (3); Symphalangism, proximal, 1B, 615298 (3)
GDF6	601147	Klippel-Feil syndrome 1, autosomal dominant, 118100 (3), Autosomal dominant; Leber congenital amaurosis 17, 615360 (3), Autosomal recessive; Microphthalmia with coloboma 6, digenic, 613703 (3), Autosomal dominant; Microphthalmia, isolated 4, 613094 (3); Multiple synostoses syndrome 4, 617898 (3), Autosomal dominant
GDI1	300104	Mental retardation, X-linked 41, 300849 (3), X-linked dominant
GDNF	600837	Central hypoventilation syndrome, 209880 (3), Autosomal dominant; {Hirschsprung disease, susceptibility to, 3}, 613711 (3), Autosomal dominant; {Pheochromocytoma, modifier of}, 171300 (3), Autosomal dominant
GFAP	137780	Alexander disease, 203450 (3), Autosomal dominant
GFER	600924	Myopathy, mitochondrial progressive, with congenital cataract, hearing loss, and developmental delay, 613076 (3)
GFI1	600871	?Neutropenia, nonimmune chronic idiopathic, of adults, 607847 (3), Autosomal dominant; ?Neutropenia, severe congenital 2, autosomal dominant, 613107 (3), Autosomal dominant
GFI1B	604383	Bleeding disorder, platelet-type, 17, 187900 (3), Autosomal recessive, Autosomal dominant
GFM1	606639	Combined oxidative phosphorylation deficiency 1, 609060 (3), Autosomal recessive
GFPT1	138292	Myasthenia, congenital, 12, with tubular aggregates, 610542 (3), Autosomal recessive
GGCX	137167	Pseudoxanthoma elasticum-like disorder with multiple coagulation factor deficiency, 610842 (3); Vitamin K-dependent clotting factors, combined deficiency of, 1, 277450 (3), Autosomal recessive
GH1	139250	Growth hormone deficiency, isolated, type IA, 262400 (3), Autosomal recessive; Growth hormone deficiency, isolated, type IB, 612781 (3); Growth hormone deficiency, isolated, type II, 173100 (3), Autosomal dominant; Kowarski syndrome, 262650 (3), Autosomal recessive
GHRHR	139191	Growth hormone deficiency, isolated, type IB, 612781 (3)
GHRL	605353	{Obesity, susceptibility to}, 601665 (3), Autosomal recessive, Autosomal dominant, Multifactorial
GIF	609342	Intrinsic factor deficiency, 261000 (3), Autosomal recessive
GIGYF2	612003	{Parkinson disease 11}, 607688 (3)
GIPC3	608792	Deafness, autosomal recessive 15, 601869 (3), Autosomal recessive

GJA1	121014	Atrioventricular septal defect 3, 600309 (3), Autosomal dominant; Craniometaphyseal dysplasia, autosomal recessive, 218400 (3), Autosomal recessive; Erythrokeratodermia variabilis et progressiva 3, 617525 (3), Autosomal dominant; Hypoplastic left heart syndrome 1, 241550 (3), Autosomal recessive; Oculodentodigital dysplasia, 164200 (3), Autosomal dominant; Oculodentodigital dysplasia, autosomal recessive, 257850 (3), Autosomal recessive; Palmoplantar keratoderma with congenital alopecia, 104100 (3), Autosomal dominant; Syndactyly, type III, 186100 (3), Autosomal dominant
GJA3	121015	Cataract 14, multiple types, 601885 (3), Autosomal dominant
GJA5	121013	Atrial fibrillation, familial, 11, 614049 (3), Autosomal dominant; Atrial standstill, digenic (GJA5/SCN5A), 108770 (3), Autosomal dominant
GJA8	600897	Cataract 1, multiple types, 116200 (3), Autosomal dominant
GJB1	304040	Charcot-Marie-Tooth neuropathy, X-linked dominant, 1, 302800 (3), X-linked dominant
GJB2	121011	Bart-Pumphrey syndrome, 149200 (3), Autosomal dominant; Deafness, autosomal dominant 3A, 601544 (3), Autosomal dominant; Deafness, autosomal recessive 1A, 220290 (3), Autosomal recessive; Hystrix-like ichthyosis with deafness, 602540 (3), Autosomal dominant; Keratitis-ichthyosis-deafness syndrome, 148210 (3), Autosomal dominant; Keratoderma, palmoplantar, with deafness, 148350 (3), Autosomal dominant; Vohwinkel syndrome, 124500 (3), Autosomal dominant
GJB3	603324	Deafness, autosomal dominant 2B, 612644 (3), Autosomal dominant; Deafness, autosomal dominant, with peripheral neuropathy (3); Deafness, autosomal recessive (3); Deafness, digenic, GJB2/GJB3, 220290 (3), Autosomal recessive; Erythrokeratodermia variabilis et progressiva 1, 133200 (3), Autosomal recessive, Autosomal dominant
GJB4	605425	Erythrokeratodermia variabilis et progressiva 2, 617524 (3), Autosomal dominant
GJB6	604418	Deafness, autosomal dominant 3B, 612643 (3), Autosomal dominant; Deafness, autosomal recessive 1B, 612645 (3), Autosomal recessive; Deafness, digenic GJB2/GJB6, 220290 (3), Autosomal recessive; Ectodermal dysplasia 2, Clouston type, 129500 (3), Autosomal dominant
GJC2	608803	Leukodystrophy, hypomyelinating, 2, 608804 (3), Autosomal recessive; Lymphedema, hereditary, IC, 613480 (3), Autosomal dominant; Spastic paraplegia 44, autosomal recessive, 613206 (3), Autosomal recessive
GK	300474	Glycerol kinase deficiency, 307030 (3), X-linked recessive
GLA	300644	Fabry disease, 301500 (3), X-linked; Fabry disease, cardiac variant, 301500 (3), X-linked
GLB1	611458	GM1-gangliosidosis, type I, 230500 (3), Autosomal recessive; GM1-gangliosidosis, type II, 230600 (3), Autosomal recessive; GM1-gangliosidosis, type III, 230650 (3), Autosomal recessive; Mucopolysaccharidosis type IVB (Morquio), 253010 (3), Autosomal recessive

GLCCI1	614283	{Glucocorticoid therapy, response to}, 614400 (3)
GLDC	238300	Glycine encephalopathy, 605899 (3), Autosomal recessive
GLE1	603371	Arthrogryposis, lethal, with anterior horn cell disease, 611890 (3); Lethal congenital contracture syndrome 1, 253310 (3), Autosomal recessive
GLI2	165230	Culler-Jones syndrome, 615849 (3), Autosomal dominant; Holoprosencephaly 9, 610829 (3), Autosomal dominant
GLI3	165240	Greig cephalopolysyndactyly syndrome, 175700 (3), Autosomal dominant; {Hypothalamic hamartomas, somatic}, 241800 (3); Pallister-Hall syndrome, 146510 (3), Autosomal dominant; Polydactyly, postaxial, types A1 and B, 174200 (3), Autosomal dominant; Polydactyly, preaxial, type IV, 174700 (3), Autosomal dominant
GLIS2	608539	Nephronophthisis 7, 611498 (3)
GLIS3	610192	Diabetes mellitus, neonatal, with congenital hypothyroidism, 610199 (3), Autosomal recessive
GLMN	601749	Glomuvenous malformations, 138000 (3), Autosomal dominant
GLRA1	138491	Hyperekplexia 1, 149400 (3), Autosomal recessive, Autosomal dominant
GLRB	138492	Hyperekplexia 2, 614619 (3), Autosomal recessive
GLRX5	609588	Anemia, sideroblastic, 3, pyridoxine-refractory, 616860 (3), Autosomal recessive; Spasticity, childhood-onset, with hyperglycinemia, 616859 (3), Autosomal recessive
GLUD1	138130	Hyperinsulinism-hyperammonemia syndrome, 606762 (3), Autosomal dominant
GLUD2	300144	{Parkinson disease, age of onset, modifier}, 168600 (3), Isolated cases, Multifactorial
GLUL	138290	Glutamine deficiency, congenital, 610015 (3), Autosomal recessive
GLYCTK	610516	D-glyceric aciduria, 220120 (3), Autosomal recessive
GM2A	613109	GM2-gangliosidosis, AB variant, 272750 (3), Autosomal recessive
GMPPB	615320	Muscular dystrophy-dystroglycanopathy (congenital with brain and eye anomalies), type A, 14, 615350 (3), Autosomal recessive; Muscular dystrophy-dystroglycanopathy (congenital with mental retardation), type B, 14, 615351 (3), Autosomal recessive; Muscular dystrophy-dystroglycanopathy (limb-girdle), type C, 14, 615352 (3), Autosomal recessive
GNAI3	139370	Auriculocondylar syndrome 1, 602483 (3), Autosomal dominant
GNAL	139312	Dystonia 25, 615073 (3), Autosomal dominant
GNAO1	139311	Epileptic encephalopathy, early infantile, 17, 615473 (3), Autosomal dominant; Neurodevelopmental disorder with involuntary movements, 617493 (3), Autosomal dominant
GNAQ	600998	Capillary malformations, congenital, 1, somatic, mosaic, 163000 (3); Sturge-Weber syndrome, somatic, mosaic, 185300 (3)

GNAS	139320	ACTH-independent macronodular adrenal hyperplasia, 219080 (3), Isolated cases; McCune-Albright syndrome, somatic, mosaic, 174800 (3); Osseous heteroplasia, progressive, 166350 (3), Autosomal dominant; Pituitary adenoma 3, multiple types, somatic, 617686 (3); Pseudohypoparathyroidism Ia, 103580 (3), Autosomal dominant; Pseudohypoparathyroidism Ib, 603233 (3), Autosomal dominant; Pseudohypoparathyroidism Ic, 612462 (3), Autosomal dominant; Pseudopseudohypoparathyroidism, 612463 (3), Autosomal dominant
GNAT1	139330	Night blindness, congenital stationary, autosomal dominant 3, 610444 (3), Autosomal dominant; Night blindness, congenital stationary, type 1G, 616389 (3), Autosomal recessive
GNAT2	139340	Achromatopsia 4, 613856 (3)
GNB1	139380	Leukemia, acute lymphoblastic, somatic, 613065 (3); Mental retardation, autosomal dominant 42, 616973 (3), Autosomal dominant
GNB3	139130	{Hypertension, essential, susceptibility to}, 145500 (3), Multifactorial; Night blindness, congenital stationary, type 1H, 617024 (3), Autosomal recessive
GNB4	610863	Charcot-Marie-Tooth disease, dominant intermediate F, 615185 (3), Autosomal dominant
GNB5	604447	Intellectual developmental disorder with cardiac arrhythmia, 617173 (3), Autosomal recessive; Language delay and ADHD/cognitive impairment with or without cardiac arrhythmia, 617182 (3), Autosomal recessive
GNE	603824	Nonaka myopathy, 605820 (3), Autosomal recessive; Sialuria, 269921 (3), Autosomal dominant
GNMT	606628	Glycine N-methyltransferase deficiency, 606664 (3), Autosomal recessive
GNPAT	602744	Rhizomelic chondrodysplasia punctata, type 2, 222765 (3), Autosomal recessive
GNPTAB	607840	Mucolipidosis II alpha/beta, 252500 (3), Autosomal recessive; Mucolipidosis III alpha/beta, 252600 (3), Autosomal recessive
GNPTG	607838	Mucolipidosis III gamma, 252605 (3), Autosomal recessive
GNRHR	138850	Hypogonadotropic hypogonadism 7 without anosmia, 146110 (3), Autosomal recessive
GNS	607664	Mucopolysaccharidosis type IIID, 252940 (3), Autosomal recessive
GOLGA5	606918	No OMIM phenotype
GORAB	607983	Geroderma osteodysplasticum, 231070 (3), Autosomal recessive
GOSR2	604027	Epilepsy, progressive myoclonic 6, 614018 (3), Autosomal recessive
GOT1	138180	Aspartate aminotransferase, serum level of, QTL1, 614419 (3)
GP1BA	606672	Bernard-Soulier syndrome, type A1 (recessive), 231200 (3), Autosomal recessive; Bernard-Soulier syndrome, type A2 (dominant), 153670 (3), Autosomal dominant; {Nonarteritic anterior ischemic optic neuropathy, susceptibility to}, 258660 (3), Autosomal recessive; von Willebrand disease, platelet-type, 177820 (3), Autosomal dominant

GP1BB	138720	Bernard-Soulier syndrome, type B, 231200 (3), Autosomal recessive; Giant platelet disorder, isolated, 231200 (3), Autosomal recessive
GP6	605546	Bleeding disorder, platelet-type, 11, 614201 (3), Autosomal recessive
GP9	173515	Bernard-Soulier syndrome, type C, 231200 (3), Autosomal recessive
GPC3	300037	Simpson-Golabi-Behmel syndrome, type 1, 312870 (3), X-linked recessive; Wilms tumor, somatic, 194070 (3)
GPC4	300168	No OMIM phenotype
GPC6	604404	Omodysplasia 1, 258315 (3), Autosomal recessive
GPD1	138420	Hypertriglyceridemia, transient infantile, 614480 (3), Autosomal recessive
GPD1L	611778	Brugada syndrome 2, 611777 (3)
GPHN	603930	Molybdenum cofactor deficiency C, 615501 (3), Autosomal recessive
GPI	172400	Hemolytic anemia, nonspherocytic, due to glucose phosphate isomerase deficiency, 613470 (3), Autosomal recessive
GPIHBP1	612757	Hyperlipoproteinemia, type 1D, 615947 (3), Autosomal recessive
GPR101	300393	Pituitary adenoma 2, GH-secreting, 300943 (3)
GPR143	300808	Nystagmus 6, congenital, X-linked, 300814 (3); Ocular albinism, type I, Nettleship-Falls type, 300500 (3), X-linked
GPR179	614515	Night blindness, congenital stationary (complete), 1E, autosomal recessive, 614565 (3), Autosomal recessive
GPR88	607468	?Chorea, childhood-onset, with psychomotor retardation, 616939 (3), Autosomal recessive
GPT2	138210	Mental retardation, autosomal recessive 49, 616281 (3), Autosomal recessive
GPX1	138320	Hemolytic anemia due to glutathione peroxidase deficiency, 614164 (1), Autosomal recessive
GPX4	138322	Spondylometaphyseal dysplasia, Sedaghatian type, 250220 (3), Autosomal recessive
GREM2	608832	Tooth agenesis, selective, 9, 617275 (3), Autosomal dominant
GRHPR	604296	Hyperoxaluria, primary, type II, 260000 (3), Autosomal recessive
GRIA3	305915	Mental retardation, X-linked 94, 300699 (3), X-linked recessive
GRID2	602368	Spinocerebellar ataxia, autosomal recessive 18, 616204 (3), Autosomal recessive
GRIN1	138249	Neurodevelopmental disorder with or without hyperkinetic movements and seizures, autosomal dominant, 614254 (3), Autosomal dominant; Neurodevelopmental disorder with or without hyperkinetic movements and seizures, autosomal recessive, 617820 (3), Autosomal recessive
GRIN2B	138252	Epileptic encephalopathy, early infantile, 27, 616139 (3), Autosomal dominant; Mental retardation, autosomal dominant 6, 613970 (3), Autosomal dominant
GRIN2D	602717	Epileptic encephalopathy, early infantile, 46, 617162 (3), Autosomal dominant
GRIP1	604597	Fraser syndrome 3, 617667 (3), Autosomal recessive
GRK1	180381	Oguchi disease-2, 613411 (3)

GRM1	604473	Spinocerebellar ataxia 44, 617691 (3), Autosomal dominant; Spinocerebellar ataxia, autosomal recessive 13, 614831 (3), Autosomal recessive
GRM6	604096	Night blindness, congenital stationary (complete), 1B, autosomal recessive, 257270 (3), Autosomal recessive
GRN	138945	Aphasia, primary progressive, 607485 (3), Autosomal dominant; Ceroid lipofuscinosis, neuronal, 11, 614706 (3), Autosomal recessive; Frontotemporal lobar degeneration with ubiquitin-positive inclusions, 607485 (3), Autosomal dominant
GRXCR1	613283	Deafness, autosomal recessive 25, 613285 (3), Autosomal recessive
GRXCR2	615762	?Deafness, autosomal recessive 101, 615837 (3), Autosomal recessive
GSC	138890	Short stature, auditory canal atresia, mandibular hypoplasia, skeletal abnormalities, 602471 (3), Autosomal recessive
GSDME (DFNA5)	608798	Deafness, autosomal dominant 5, 600994 (3), Autosomal dominant
GSS	601002	Glutathione synthetase deficiency, 266130 (3), Autosomal recessive; Hemolytic anemia due to glutathione synthetase deficiency, 231900 (3), Autosomal recessive
GTF2E2	189964	Trichothiodystrophy 6, nonphotosensitive, 616943 (3), Autosomal recessive
GTF2H5	608780	Trichothiodystrophy 3, photosensitive, 616395 (3)
GTPBP3	608536	Combined oxidative phosphorylation deficiency 23, 616198 (3), Autosomal recessive
GUCA1A	600364	Cone dystrophy-3, 602093 (3), Autosomal dominant; Cone-rod dystrophy 14, 602093 (3), Autosomal dominant
GUCA1B	602275	Retinitis pigmentosa 48, 613827 (3)
GUCY1A1 (GUCY1A3)	139396	Moyamoya 6 with achalasia, 615750 (3), Autosomal recessive
GUCY2C	601330	Diarrhea 6, 614616 (3), Autosomal dominant; Meconium ileus, 614665 (3), Autosomal recessive
GUCY2D	600179	?Central areolar choroidal dystrophy 1, 215500 (3), Autosomal recessive; Cone-rod dystrophy 6, 601777 (3), Autosomal recessive, Autosomal dominant; Leber congenital amaurosis 1, 204000 (3), Autosomal recessive
GUF1	617064	?Epileptic encephalopathy, early infantile, 40, 617065 (3), Autosomal recessive
GUSB	611499	Mucopolysaccharidosis VII, 253220 (3), Autosomal recessive
GYG1	603942	?Glycogen storage disease XV, 613507 (3), Autosomal recessive; Polyglucosan body myopathy 2, 616199 (3), Autosomal recessive
GYPA	617922	[Blood group, MNSs system], 111300 (3); {Malaria, resistance to}, 611162 (3)
GYPB	617923	[Blood group, Ss], 111740 (3); {Malaria, resistance to}, 611162 (3)
GYPC	110750	[Blood group, Gerbich], 616089 (3); {Malaria, resistance to}, 611162 (3)
GYS1	138570	Glycogen storage disease 0, muscle, 611556 (3), Autosomal recessive

GYS2	138571	Glycogen storage disease 0, liver, 240600 (3), Autosomal recessive
H6PD	138090	Cortisone reductase deficiency 1, 604931 (3), Autosomal recessive
HABP2	603924	{?Thyroid cancer, nonmedullary, 5}, 616535 (3), Autosomal dominant; {Venous thromboembolism, susceptibility to}, 188050 (3), Autosomal dominant
HACE1	610876	Spastic paraplegia and psychomotor retardation with or without seizures, 616756 (3), Autosomal recessive
HADH	601609	3-hydroxyacyl-CoA dehydrogenase deficiency, 231530 (3), Autosomal recessive; Hyperinsulinemic hypoglycemia, familial, 4, 609975 (3), Autosomal recessive
HADHA	600890	Fatty liver, acute, of pregnancy, 609016 (3), Autosomal recessive; HELLP syndrome, maternal, of pregnancy, 609016 (3), Autosomal recessive; LCHAD deficiency, 609016 (3), Autosomal recessive; Trifunctional protein deficiency, 609015 (3), Autosomal recessive
HADHB	143450	Trifunctional protein deficiency, 609015 (3), Autosomal recessive
HAGH	138760	[Glyoxalase II deficiency], 614033 (1), Autosomal dominant
HAL	609457	[Histidinemia], 235800 (3), Autosomal recessive, Autosomal dominant
HAMP	606464	Hemochromatosis, type 2B, 613313 (3), Autosomal recessive
HARS	142810	Charcot-Marie-Tooth disease, axonal, type 2W, 616625 (3), Autosomal dominant; Usher syndrome type 3B, 614504 (3), Autosomal recessive
HARS2	600783	?Perrault syndrome 2, 614926 (3), Autosomal recessive
HAVCR1	606518	No OMIM phenotype
HAX1	605998	Neutropenia, severe congenital 3, autosomal recessive, 610738 (3), Autosomal recessive
HBA1	141800	Erythrocytosis, 7, 617981 (3); Heinz body anemias, alpha-, 140700 (3), Autosomal dominant; Hemoglobin H disease, nondeletional, 613978 (3); Methemoglobinemia, alpha type, 617973 (3); Thalassemias, alpha-, 604131 (3)
HBA2	141850	Erythrocytosis 7, 617981 (3); Heinz body anemia, 140700 (3), Autosomal dominant; Hemoglobin H disease, deletional and nondeletional, 613978 (3); Thalassemia, alpha-, 604131 (3)
HBB	141900	Delta-beta thalassemia, 141749 (3), Autosomal dominant; Erythrocytosis 6, 617980 (3); Heinz body anemia, 140700 (3), Autosomal dominant; Hereditary persistence of fetal hemoglobin, 141749 (3), Autosomal dominant; {Malaria, resistance to}, 611162 (3); Methmoglobinemia, beta type, 617971 (3); Sickle cell anemia, 603903 (3), Autosomal recessive; Thalassemia, beta, 613985 (3); Thalassemia-beta, dominant inclusion-body, 603902 (3)
HBG1	142200	Fetal hemoglobin quantitative trait locus 1, 141749 (3), Autosomal dominant
HBG2	142250	Cyanosis, transient neonatal, 613977 (3), Autosomal dominant; Fetal hemoglobin quantitative trait locus 1, 141749 (3), Autosomal dominant
HCCS	300056	Linear skin defects with multiple congenital anomalies 1, 309801 (3), X-linked dominant

HCFC1	300019	Mental retardation, X-linked 3 (methylmalonic acidemia and homocysteinemia, cblX type), 309541 (3), X-linked recessive
HCN1	602780	Epileptic encephalopathy, early infantile, 24, 615871 (3), Autosomal dominant
HCN4	605206	Brugada syndrome 8, 613123 (3); Sick sinus syndrome 2, 163800 (3), Autosomal dominant
HCRT	602358	?Narcolepsy 1, 161400 (3), Autosomal dominant
HDAC6	300272	?Chondrodysplasia with platyspondyly, distinctive brachydactyly, hydrocephaly, and microphthalmia, 300863 (3), X-linked dominant
HDC	142704	{Gilles de la Tourette syndrome, susceptibility to}, 137580 (3), Autosomal dominant
HELLS	603946	Immunodeficiency-centromeric instability-facial anomalies syndrome 4, 616911 (3), Autosomal recessive
HEPACAM	611642	Megalencephalic leukoencephalopathy with subcortical cysts 2A, 613925 (3), Autosomal recessive; Megalencephalic leukoencephalopathy with subcortical cysts 2B, remitting, with or without mental retardation, 613926 (3), Autosomal dominant
HERC1	605109	Macrocephaly, dysmorphic facies, and psychomotor retardation, 617011 (3), Autosomal recessive
HERC2	605837	Mental retardation, autosomal recessive 38, 615516 (3), Autosomal recessive; [Skin/hair/eye pigmentation 1, blond/brown hair], 227220 (3), Autosomal recessive; [Skin/hair/eye pigmentation 1, blue/nonblue eyes], 227220 (3), Autosomal recessive
HERC3	605200	No OMIM phenotype
HES7	608059	Spondylocostal dysostosis 4, autosomal recessive, 613686 (3), Autosomal recessive
HESX1	601802	Growth hormone deficiency with pituitary anomalies, 182230 (3), Autosomal recessive, Autosomal dominant; Pituitary hormone deficiency, combined, 5, 182230 (3), Autosomal recessive, Autosomal dominant; Septooptic dysplasia, 182230 (3), Autosomal recessive, Autosomal dominant
HEXA	606869	GM2-gangliosidosis, several forms, 272800 (3), Autosomal recessive; [Hex A pseudodeficiency], 272800 (3), Autosomal recessive; Tay-Sachs disease, 272800 (3), Autosomal recessive
HEXB	606873	Sandhoff disease, infantile, juvenile, and adult forms, 268800 (3), Autosomal recessive
HFE	613609	{Alzheimer disease, susceptibility to}, 104300 (3), Autosomal dominant; Hemochromatosis, 235200 (3), Autosomal recessive; {Microvascular complications of diabetes 7}, 612635 (3); {Porphyria cutanea tarda, susceptibility to}, 176100 (3), Autosomal recessive, Autosomal dominant; {Porphyria variegata, susceptibility to}, 176200 (3), Autosomal dominant; [Transferrin serum level QTL2], 614193 (3)
HFE2	No OMIM gene	No OMIM phenotype
HFM1	615684	Premature ovarian failure 9, 615724 (3), Autosomal recessive
HGD	607474	Alkaptonuria, 203500 (3), Autosomal recessive

HGF	142409	Deafness, autosomal recessive 39, 608265 (3), Autosomal recessive
HGSNAT	610453	Mucopolysaccharidosis type IIIC (Sanfilippo C), 252930 (3), Autosomal recessive; Retinitis pigmentosa 73, 616544 (3), Autosomal recessive
HIBCH	610690	3-hydroxyisobutryl-CoA hydrolase deficiency, 250620 (3), Autosomal recessive
HINT1	601314	Neuromyotonia and axonal neuropathy, autosomal recessive, 137200 (3), Autosomal recessive
HIVEP2	143054	Mental retardation, autosomal dominant 43, 616977 (3), Autosomal dominant
HLA-A	142800	{Hypersensitivity syndrome, carbamazepine-induced, susceptibility to}, 608579 (3)
HLA-B	142830	{Abacavir hypersensitivity, susceptibility to} (3); {Drug-induced liver injury due to flucloxacillin} (3); {Spondyloarthropathy, susceptibility to, 1}, 106300 (3), Multifactorial; {Stevens-Johnson syndrome, susceptibility to}, 608579 (3); {Synovitis, chronic, susceptibility to} (3); {Toxic epidermal necrolysis, susceptibility to}, 608579 (3)
HLA-C	142840	{HIV-1 viremia, susceptibility to}, 609423 (3); {Psoriasis susceptibility 1}, 177900 (3), Multifactorial
HLA-DQA1	146880	{Celiac disease, susceptibility to}, 212750 (3), Autosomal recessive, Multifactorial
HLA-DQB1	604305	{Celiac disease, susceptibility to}, 212750 (3), Autosomal recessive, Multifactorial; {Creutzfeldt-Jakob disease, variant, resistance to}, 123400 (3), Autosomal dominant; {Multiple sclerosis, susceptibility to, 1}, 126200 (3), Multifactorial
HLA-DRB1	142857	{Multiple sclerosis, susceptibility to, 1}, 126200 (3), Multifactorial; {Sarcoidosis, susceptibility to, 1}, 181000 (3), Autosomal dominant
HLA-G	142871	{Asthma, susceptibility to}, 600807 (2), Autosomal dominant
HLCS	609018	Holocarboxylase synthetase deficiency, 253270 (3), Autosomal recessive
HMCN1	608548	{Macular degeneration, age-related, 1}, 603075 (3), Autosomal dominant
HMGA1	600701	{Diabetes mellitus, noninsulin-dependent, susceptibility to}, 125853 (3), Autosomal dominant
HMGCL	613898	HMG-CoA lyase deficiency, 246450 (3), Autosomal recessive
HMGCS2	600234	HMG-CoA synthase-2 deficiency, 605911 (3), Autosomal recessive
HMMR	600936	{Breast cancer, susceptibility to}, 114480 (3), Autosomal dominant
HMOX1	141250	Heme oxygenase-1 deficiency, 614034 (3); {Pulmonary disease, chronic obstructive, susceptibility to}, 606963 (3)
HMX1	142992	Oculoauricular syndrome, 612109 (3), Autosomal recessive
HNF1A	142410	Diabetes mellitus, insulin-dependent, 20, 612520 (3); {Diabetes mellitus, insulin-dependent}, 222100 (3), Autosomal recessive; {Diabetes mellitus, noninsulin-dependent, 2}, 125853 (3), Autosomal dominant; Hepatic adenoma, somatic, 142330 (3); MODY, type III, 600496 (3), Autosomal dominant; Renal cell carcinoma, 144700 (3)

HNF1B	189907	Diabetes mellitus, noninsulin-dependent, 125853 (3), Autosomal dominant; {Renal cell carcinoma}, 144700 (3); Renal cysts and diabetes syndrome, 137920 (3), Autosomal dominant
HNF4A	600281	{Diabetes mellitus, noninsulin-dependent}, 125853 (3), Autosomal dominant; Fanconi renotubular syndrome 4, with maturity-onset diabetes of the young, 616026 (3), Autosomal dominant; MODY, type I, 125850 (3), Autosomal dominant
HNMT	605238	{Asthma, susceptibility to}, 600807 (3), Autosomal dominant; Mental retardation, autosomal recessive 51, 616739 (3), Autosomal recessive
HNRNPA1	164017	Amyotrophic lateral sclerosis 20, 615426 (3), Autosomal dominant; ?Inclusion body myopathy with early-onset Paget disease without frontotemporal dementia 3, 615424 (3), Autosomal dominant
HNRNPA2B1	600124	?Inclusion body myopathy with early-onset Paget disease with or without frontotemporal dementia 2, 615422 (3)
HNRNPDL	607137	Muscular dystrophy, limb-girdle, type 1G, 609115 (3), Autosomal dominant
HNRNPH2	300610	Mental retardation, X-linked, syndromic, Bain type, 300986 (3), X-linked dominant
HNRNPK	600712	Au-Kline syndrome, 616580 (3), Autosomal dominant
HNRNPU	602869	Epileptic encephalopathy, early infantile, 54, 617391 (3), Autosomal dominant
HOGA1	613597	Hyperoxaluria, primary, type III, 613616 (3)
HOMER2	604799	?Deafness, autosomal dominant 68, 616707 (3), Autosomal dominant
HOXA1	142955	Athabaskan brainstem dysgenesis syndrome, 601536 (3); Bosley-Salih-Alorainy syndrome, 601536 (3)
HOXA11	142958	Radioulnar synostosis with amegakaryocytic thrombocytopenia 1, 605432 (3), Autosomal dominant
HOXA13	142959	Guttmacher syndrome, 176305 (3), Autosomal dominant; Hand-foot-uterus syndrome, 140000 (3), Autosomal dominant
HOXA2	604685	Microtia with or without hearing impairment (AD), 612290 (3), Autosomal recessive, Autosomal dominant; ?Microtia, hearing impairment, and cleft palate (AR), 612290 (3), Autosomal recessive, Autosomal dominant
HOXC13	142976	Ectodermal dysplasia 9, hair/nail type, 614931 (3), Autosomal recessive
HOXD10	142984	Charcot-Marie-Tooth disease, foot deformity of, 192950 (3), Autosomal dominant; Vertical talus, congenital, 192950 (3), Autosomal dominant
HOXD13	142989	Brachydactyly, type D, 113200 (3), Autosomal dominant; Brachydactyly, type E, 113300 (3), Autosomal dominant; ?Brachydactyly-syndactyly syndrome, 610713 (3); Syndactyly, type V, 186300 (3), Autosomal dominant; Synpolydactyly 1, 186000 (3), Autosomal dominant
HP	140100	[Anhaptoglobinemia], 614081 (3); [Hypohaptoglobinemia], 614081 (3)
HPCA	142622	Dystonia 2, torsion, autosomal recessive, 224500 (3), Autosomal recessive

HPD	609695	Hawkinsinuria, 140350 (3), Autosomal dominant; Tyrosinemia, type III, 276710 (3), Autosomal recessive
HPGD	601688	Cranioosteoarthropathy, 259100 (3), Autosomal recessive; Digital clubbing, isolated congenital, 119900 (3), Autosomal recessive; Hypertrophic osteoarthropathy, primary, autosomal recessive 1, 259100 (3), Autosomal recessive
HPRT1	308000	HPRT-related gout, 300323 (3), X-linked recessive; Lesch-Nyhan syndrome, 300322 (3), X-linked recessive
HPS1	604982	Hermansky-Pudlak syndrome 1, 203300 (3), Autosomal recessive
HPS3	606118	Hermansky-Pudlak syndrome 3, 614072 (3), Autosomal recessive
HPS4	606682	Hermansky-Pudlak syndrome 4, 614073 (3), Autosomal recessive
HPS5	607521	Hermansky-Pudlak syndrome 5, 614074 (3), Autosomal recessive
HPS6	607522	Hermansky-Pudlak syndrome 6, 614075 (3), Autosomal recessive
HPSE2	613469	Urofacial syndrome 1, 236730 (3), Autosomal recessive
HR	602302	Alopecia universalis, 203655 (3), Autosomal recessive; Atrichia with papular lesions, 209500 (3), Autosomal recessive; Hypotrichosis 4, 146550 (3), Autosomal dominant
HRAS	190020	{Bladder cancer, somatic}, 109800 (3); Congenital myopathy with excess of muscle spindles, 218040 (3), Autosomal dominant, Isolated cases; Costello syndrome, 218040 (3), Autosomal dominant, Isolated cases; {Nevus sebaceous or woolly hair nevus, somatic}, 162900 (3); Schimmelpenning-Feuerstein-Mims syndrome, somatic mosaic, 163200 (3); {Spitz nevus or nevus spilus, somatic}, 137550 (3); {Thyroid carcinoma, follicular, somatic}, 188470 (3)
HRG	142640	Thrombophilia due to HRG deficiency, 613116 (3), Autosomal dominant; Thrombophilia due to elevated HRG, 613116 (1), Autosomal dominant
HS6ST1	604846	{Hypogonadotropic hypogonadism 15 with or without anosmia}, 614880 (3), Autosomal dominant
HSD11B1	600713	Cortisone reductase deficiency 2, 614662 (3), Autosomal dominant
HSD11B2	614232	Apparent mineralocorticoid excess, 218030 (3), Autosomal recessive
HSD17B10	300256	HSD10 mitochondrial disease, 300438 (3), X-linked dominant
HSD17B3	605573	Pseudohermaphroditism, male, with gynecomastia, 264300 (3), Autosomal recessive
HSD17B4	601860	D-bifunctional protein deficiency, 261515 (3), Autosomal recessive; Perrault syndrome 1, 233400 (3), Autosomal recessive
HSD3B2	613890	Adrenal hyperplasia, congenital, due to 3-beta-hydroxysteroid dehydrogenase 2 deficiency, 201810 (3), Autosomal recessive
HSD3B7	607764	Bile acid synthesis defect, congenital, 1, 607765 (3), Autosomal recessive
HSF4	602438	Cataract 5, multiple types, 116800 (3), Autosomal dominant
HSFY1	400029	No OMIM phenotype
HSPA9	600548	Anemia, sideroblastic, 4, 182170 (3), Autosomal dominant; Even-plus syndrome, 616854 (3), Autosomal recessive

HSPB1	602195	Charcot-Marie-Tooth disease, axonal, type 2F, 606595 (3), Autosomal dominant; Neuropathy, distal hereditary motor, type IIB, 608634 (3), Autosomal dominant
HSPB3	604624	?Neuronopathy, distal hereditary motor, type IIC, 613376 (3), Autosomal dominant
HSPB8	608014	Charcot-Marie-Tooth disease, axonal, type 2L, 608673 (3), Autosomal dominant; Neuropathy, distal hereditary motor, type IIA, 158590 (3), Autosomal dominant
HSPD1	118190	Leukodystrophy, hypomyelinating, 4, 612233 (3), Autosomal recessive; Spastic paraplegia 13, autosomal dominant, 605280 (3), Autosomal dominant
HSPG2	142461	Dyssegmental dysplasia, Silverman-Handmaker type, 224410 (3), Autosomal recessive; Schwartz-Jampel syndrome, type 1, 255800 (3), Autosomal recessive
HTR1A	109760	Periodic fever, menstrual cycle dependent, 614674 (3), Autosomal dominant
HTR2A	182135	{Alcohol dependence, susceptibility to}, 103780 (3), Multifactorial; {Anorexia nervosa, susceptibility to}, 606788 (3); {Major depressive disorder, response to citalopram therapy in}, 608516 (3); {Obsessive-compulsive disorder, susceptibility to}, 164230 (3), Autosomal dominant; {Schizophrenia, susceptibility to}, 181500 (3), Autosomal dominant; {Seasonal affective disorder, susceptibility to}, 608516 (3)
HTRA1	602194	CARASIL syndrome, 600142 (3), Autosomal recessive; Cerebral arteriopathy, autosomal dominant, with subcortical infarcts and leukoencephalopathy, type 2, 616779 (3), Autosomal dominant; {Macular degeneration, age-related, 7}, 610149 (3); {Macular degeneration, age-related, neovascular type}, 610149 (3)
HTRA2	606441	3-methylglutaconic aciduria, type VIII, 617248 (3), Autosomal recessive; {Parkinson disease 13}, 610297 (3)
HTT	613004	Huntington disease, 143100 (3), Autosomal dominant; Lopes-Maciel-Rodan syndrome, 617435 (3), Autosomal recessive
HUWE1	300697	Mental retardation, X-linked syndromic, Turner type, 300706 (3)
HYAL1	607071	?Mucopolysaccharidosis type IX, 601492 (3), Autosomal recessive
HYDIN	610812	Ciliary dyskinesia, primary, 5, 608647 (3), Autosomal recessive
HYLS1	610693	Hydrolethalus syndrome, 236680 (3), Autosomal recessive
IARS2	612801	?Cataracts, growth hormone deficiency, sensory neuropathy, sensorineural hearing loss, and skeletal dysplasia, 616007 (3), Autosomal recessive
IBA57	615316	Multiple mitochondrial dysfunctions syndrome 3, 615330 (3), Autosomal recessive; ?Spastic paraplegia 74, autosomal recessive, 616451 (3), Autosomal recessive
ICAM1	147840	{Malaria, cerebral, susceptibility to}, 611162 (3)
ICAM4	614088	[Blood group, Landsteiner-Wiener], 111250 (3)
ICOS	604558	Immunodeficiency, common variable, 1, 607594 (3), Autosomal recessive

IDH1	147700	{Glioma, susceptibility to, somatic}, 137800 (3)
IDH2	147650	D-2-hydroxyglutaric aciduria 2, 613657 (3)
IDH3B	604526	Retinitis pigmentosa 46, 612572 (3)
IDS	300823	Mucopolysaccharidosis II, 309900 (3), X-linked recessive
IDUA	252800	Mucopolysaccharidosis Ih, 607014 (3), Autosomal recessive; Mucopolysaccharidosis Ih/s, 607015 (3), Autosomal recessive; Mucopolysaccharidosis Is, 607016 (3), Autosomal recessive
IER3IP1	609382	Microcephaly, epilepsy, and diabetes syndrome, 614231 (3), Autosomal recessive
IFIH1	606951	Aicardi-Goutieres syndrome 7, 615846 (3), Autosomal dominant; Singleton-Merten syndrome 1, 182250 (3), Autosomal dominant
IFITM3	605579	{Influenza, severe, susceptibility to}, 614680 (3)
IFITM5	614757	Osteogenesis imperfecta, type V, 610967 (3), Autosomal dominant
IFNAR2	602376	{Hepatitis B virus, susceptibility to}, 610424 (3); ?Immunodeficiency 45, 616669 (3), Autosomal recessive
IFNG	147570	{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), Autosomal dominant; {Tuberculosis, protection against}, 607948 (3)
IFNGR1	107470	{H. pylori infection, susceptibility to}, 600263 (3); {Hepatitis B virus infection, susceptibility to}, 610424 (3); Immunodeficiency 27A, mycobacteriosis, AR, 209950 (3), Autosomal recessive; Immunodeficiency 27B, mycobacteriosis, AD, 615978 (3), Autosomal dominant; {Tuberculosis infection, protection against}, 607948 (3); {Tuberculosis, susceptibility to}, 607948 (3)
IFNGR2	147569	Immunodeficiency 28, mycobacteriosis, 614889 (3), Autosomal recessive
IFNL3	607402	{Hepatitis C virus infection, response to therapy of}, 609532 (3)
IFT122	606045	Cranioectodermal dysplasia 1, 218330 (3), Autosomal recessive
IFT140	614620	Retinitis pigmentosa 80, 617781 (3), Autosomal recessive; Short-rib thoracic dysplasia 9 with or without polydactyly, 266920 (3), Autosomal recessive
IFT172	607386	Retinitis pigmentosa 71, 616394 (3), Autosomal recessive; Short-rib thoracic dysplasia 10 with or without polydactyly, 615630 (3), Autosomal recessive
IFT27	615870	?Bardet-Biedl syndrome 19, 615996 (3), Autosomal recessive
IFT43	614068	?Cranioectodermal dysplasia 3, 614099 (3), Autosomal recessive; ?Retinitis pigmentosa 81, 617871 (3), Autosomal recessive; Short-rib thoracic dysplasia 18 with polydactyly, 617866 (3), Autosomal recessive
IFT52	617094	Short-rib thoracic dysplasia 16 with or without polydactyly, 617102 (3), Autosomal recessive
IFT80	611177	Short-rib thoracic dysplasia 2 with or without polydactyly, 611263 (3), Autosomal recessive
IGBP1	300139	Corpus callosum, agenesis of, with mental retardation, ocular coloboma and micrognathia, 300472 (3), X-linked recessive

IGF1	147440	Growth retardation with deafness and mental retardation due to IGF1 deficiency, 608747 (3), Autosomal recessive
IGF1R	147370	Insulin-like growth factor I, resistance to, 270450 (3), Autosomal recessive, Autosomal dominant
IGF2BP2	608289	{Diabetes mellitus, noninsulin-dependent, susceptibility to}, 125853 (3), Autosomal dominant
IGF2R	147280	Hepatocellular carcinoma, somatic, 114550 (3)
IGFALS	601489	Acid-labile subunit, deficiency of, 615961 (3)
IGFBP7	602867	Retinal arterial macroaneurysm with supravalvular pulmonic stenosis, 614224 (3), Autosomal recessive
IGHMBP2	600502	Charcot-Marie-Tooth disease, axonal, type 2S, 616155 (3), Autosomal recessive; Neuronopathy, distal hereditary motor, type VI, 604320 (3), Autosomal recessive
IGLL1	146770	Agammaglobulinemia 2, 613500 (3), Autosomal recessive
IGSF1	300137	Hypothyroidism, central, and testicular enlargement, 300888 (3), X-linked recessive
IGSF3	603491	?Lacrimal duct defect, 149700 (3), Autosomal recessive
IHH	600726	Acrocapitofemoral dysplasia, 607778 (3), Autosomal recessive; Brachydactyly, type A1, 112500 (3), Autosomal dominant
IKBKB	603258	Immunodeficiency 15, 615592 (3), Autosomal recessive
IKBKG	300248	Ectodermal dysplasia, hypohidrotic, with immune deficiency, 300291 (3); Ectodermal, dysplasia, anhidrotic, lymphedema and immunodeficiency, 300301 (3); Immunodeficiency 33, 300636 (3), X-linked recessive; Immunodeficiency, isolated, 300584 (3); Incontinentia pigmenti, 308300 (3), X-linked dominant; Invasive pneumococcal disease, recurrent isolated, 2, 300640 (3)
IKZF1	603023	Immunodeficiency, common variable, 13, 616873 (3), Autosomal dominant
IL10	124092	{Graft-versus-host disease, protection against}, 614395 (3); {HIV-1, susceptibility to}, 609423 (3); {Rheumatoid arthritis, progression of}, 180300 (3)
IL10RA	146933	Inflammatory bowel disease 28, early onset, autosomal recessive, 613148 (3), Autosomal recessive
IL10RB	123889	{Hepatitis B virus, susceptibility to}, 610424 (3); Inflammatory bowel disease 25, early onset, autosomal recessive, 612567 (3), Autosomal recessive
IL11RA	600939	Craniosynostosis and dental anomalies, 614188 (3), Autosomal recessive
IL12B	161561	Immunodeficiency 29, mycobacteriosis, 614890 (3), Autosomal recessive
IL13	147683	{Allergic rhinitis, susceptibility to}, 607154 (3); {Asthma, susceptibility to}, 600807 (3), Autosomal dominant
IL17F	606496	?Candidiasis, familial, 6, autosomal dominant, 613956 (3)
IL17RA	605461	Immunodeficiency 51, 613953 (3), Autosomal recessive
IL17RC	610925	Candidiasis, familial, 9, 616445 (3), Autosomal recessive

IL17RD	606807	Hypogonadotropic hypogonadism 18 with or without anosmia, 615267 (3), Autosomal recessive, Autosomal dominant
IL1B	147720	{Gastric cancer risk after H. pylori infection}, 137215 (3), Autosomal dominant
IL1RAPL1	300206	Mental retardation, X-linked 21/34, 300143 (3), X-linked recessive
IL1RN	147679	{Gastric cancer risk after H. pylori infection}, 137215 (3), Autosomal dominant; Interleukin 1 receptor antagonist deficiency, 612852 (3), Autosomal recessive; {Microvascular complications of diabetes 4}, 612628 (3)
IL21	605384	?Immunodeficiency, common variable, 11, 615767 (3), Autosomal recessive
IL23R	607562	{Inflammatory bowel disease 17, protection against}, 612261 (3); {Psoriasis, protection against}, 605606 (3)
IL2RA	147730	{Diabetes, mellitus, insulin-dependent, susceptibility to, 10}, 601942 (3); Immunodeficiency 41 with lymphoproliferation and autoimmunity, 606367 (3), Autosomal recessive
IL2RG	308380	Combined immunodeficiency, X-linked, moderate, 312863 (3), X-linked recessive; Severe combined immunodeficiency, X-linked, 300400 (3), X-linked recessive
IL31RA	609510	?Amyloidosis, primary localized cutaneous, 2, 613955 (3), Autosomal dominant
IL36RN	605507	Psoriasis 14, pustular, 614204 (3), Autosomal recessive
IL4R	147781	{AIDS, slow progression to}, 609423 (3); {Atopy, susceptibility to}, 147050 (3), Autosomal dominant
IL6	147620	{Crohn disease-associated growth failure}, 266600 (3), Multifactorial; {Diabetes, susceptibility to},, 222100 (3), Autosomal recessive; {Intracranial hemorrhage in brain cerebrovascular malformations, susceptibility to}, 108010 (3), Autosomal dominant; {Kaposi sarcoma, susceptibility to}, 148000 (3), Autosomal dominant; {Rheumatoid arthritis, systemic juvenile}, 604302 (3)
IL6R	147880	[Interleukin 6, serum level of, QTL], 614752 (3); [Interleukin-6 receptor, soluble, serum level of, QTL], 614689 (3)
IL7R	146661	Severe combined immunodeficiency, T-cell negative, B-cell/natural killer cell-positive type, 608971 (3), Autosomal recessive
ILDR1	609739	Deafness, autosomal recessive 42, 609646 (3), Autosomal recessive
IMPA1	602064	Mental retardation, autosomal recessive 59, 617323 (3), Autosomal recessive
IMPAD1	614010	Chondrodysplasia with joint dislocations, GPAPP type, 614078 (3), Autosomal recessive
IMPDH1	146690	Leber congenital amaurosis 11, 613837 (3); Retinitis pigmentosa 10, 180105 (3), Autosomal dominant
IMPG1	602870	Macular dystrophy, vitelliform, 4, 616151 (3), Autosomal dominant
IMPG2	607056	Macular dystrophy, vitelliform, 5, 616152 (3), Autosomal dominant; Retinitis pigmentosa 56, 613581 (3), Autosomal recessive

INF2	610982	Charcot-Marie-Tooth disease, dominant intermediate E, 614455 (3), Autosomal dominant; Glomerulosclerosis, focal segmental, 5, 613237 (3)
ING1	601566	Squamous cell carcinoma, head and neck, somatic, 275355 (3)
INPP5E	613037	Joubert syndrome 1, 213300 (3), Autosomal recessive; Mental retardation, truncal obesity, retinal dystrophy, and micropenis, 610156 (3), Autosomal recessive
INPPL1	600829	Opsismodysplasia, 258480 (3), Autosomal recessive
INS	176730	Diabetes mellitus, insulin-dependent, 2, 125852 (3), Autosomal dominant; Diabetes mellitus, permanent neonatal, 606176 (3), Autosomal recessive, Autosomal dominant; Hyperproinsulinemia, 616214 (3), Autosomal dominant; Maturity-onset diabetes of the young, type 10, 613370 (3), Autosomal dominant
INSL3	146738	Cryptorchidism, 219050 (3), Autosomal dominant
INSR	147670	Diabetes mellitus, insulin-resistant, with acanthosis nigricans, 610549 (3); Hyperinsulinemic hypoglycemia, familial, 5, 609968 (3), Autosomal dominant; Leprechaunism, 246200 (3), Autosomal recessive; Rabson-Mendenhall syndrome, 262190 (3), Autosomal recessive
INVS	243305	Nephronophthisis 2, infantile, 602088 (3), Autosomal recessive
IQCB1	609237	Senior-Loken syndrome 5, 609254 (3), Autosomal recessive
IQSEC2	300522	Mental retardation, X-linked 1/78, 309530 (3), X-linked dominant
IRAK3	604459	{Asthma susceptibility 5}, 611064 (3)
IRAK4	606883	IRAK4 deficiency, 607676 (3); Invasive pneumococcal disease, recurrent isolated, 1, 610799 (3)
IRF1	147575	Gastric cancer, somatic, 613659 (3); Myelodysplastic syndrome, preleukemic (3); Myelogenous leukemia, acute (3); Nonsmall cell lung cancer, somatic, 211980 (3)
IRF4	601900	[Skin/hair/eye pigmentation, variation in, 8], 611724 (3)
IRF6	607199	{Orofacial cleft 6}, 608864 (3), Isolated cases; Popliteal pterygium syndrome 1, 119500 (3), Autosomal dominant; van der Woude syndrome, 119300 (3), Autosomal dominant
IRF7	605047	?Immunodeficiency 39, 616345 (3), Autosomal recessive
IRF8	601565	Immunodeficiency 32A, mycobacteriosis, autosomal dominant, 614893 (3), Autosomal dominant; Immunodeficiency 32B, monocyte and dendritic cell deficiency, autosomal recessive, 226990 (3), Autosomal recessive
IRGM	608212	{Inflammatory bowel disease (Crohn disease) 19}, 612278 (3); {Mycobacterium tuberculosis, protection against}, 607948 (3)
IRS1	147545	{Coronary artery disease, susceptibility to} (3); {Diabetes mellitus, noninsulin-dependent}, 125853 (3), Autosomal dominant
IRS2	600797	{Diabetes mellitus, noninsulin-dependent}, 125853 (3), Autosomal dominant
IRX5	606195	Hamamy syndrome, 611174 (3), Autosomal recessive
ISCA2	615317	Multiple mitochondrial dysfunctions syndrome 4, 616370 (3), Autosomal recessive

ISCU	611911	Myopathy with lactic acidosis, hereditary, 255125 (3), Autosomal recessive
ISG15	147571	Immunodeficiency 38, 616126 (3), Autosomal recessive
ISPD	614631	Muscular dystrophy-dystroglycanopathy (congenital with brain and eye anomalies), type A, 7, 614643 (3), Autosomal recessive; Muscular dystrophy-dystroglycanopathy (limb-girdle), type C, 7, 616052 (3), Autosomal recessive
ITGA2	192974	?Glycoprotein Ia deficiency, 614200 (1), Autosomal dominant
ITGA2B	607759	Bleeding disorder, platelet-type, 16, autosomal dominant, 187800 (3), Autosomal dominant; Glanzmann thrombasthenia, 273800 (3), Autosomal recessive; Thrombocytopenia, neonatal alloimmune, BAK antigen related (3)
ITGA3	605025	Interstitial lung disease, nephrotic syndrome, and epidermolysis bullosa, congenital, 614748 (3), Autosomal recessive
ITGA6	147556	Epidermolysis bullosa, junctional, with pyloric stenosis, 226730 (3), Autosomal recessive
ITGA7	600536	Muscular dystrophy, congenital, due to ITGA7 deficiency, 613204 (3), Autosomal recessive
ITGA8	604063	Renal hypodysplasia/aplasia 1, 191830 (3), Autosomal recessive
ITGAM	120980	{Systemic lupus erythematous, association with susceptibility to, 6}, 609939 (3)
ITGB2	600065	Leukocyte adhesion deficiency, 116920 (3), Autosomal recessive
ITGB3	173470	Bleeding disorder, platelet-type, 16, autosomal dominant, 187800 (3), Autosomal dominant; Glanzmann thrombasthenia, 273800 (3), Autosomal recessive; {Myocardial infarction, susceptibility to}, 608446 (3); Purpura, posttransfusion (3); Thrombocytopenia, neonatal alloimmune (3)
ITGB4	147557	Epidermolysis bullosa of hands and feet, 131800 (3), Autosomal dominant; Epidermolysis bullosa, junctional, non-Herlitz type, 226650 (3), Autosomal recessive; Epidermolysis bullosa, junctional, with pyloric atresia, 226730 (3), Autosomal recessive
ITGB6	147558	Amelogenesis imperfecta, type IH, 616221 (3), Autosomal recessive
ITIH4	600564	{Hypercholesterolemia, susceptibility to}, 143890 (3), Autosomal dominant
ITK	186973	Lymphoproliferative syndrome 1, 613011 (3), Autosomal recessive
ITM2B	603904	Dementia, familial British, 176500 (3), Autosomal dominant; Dementia, familial Danish, 117300 (3), Autosomal dominant; ?Retinal dystrophy with inner retinal dysfunction and ganglion cell abnormalities, 616079 (3), Autosomal dominant
ITPA	147520	Epileptic encephalopathy, early infantile, 35, 616647 (3), Autosomal recessive; [Inosine triphosphatase deficiency], 613850 (3)
ITPKC	606476	{Kawasaki disease, susceptibility to}, 611775 (3)
ITPR2	600144	?Anhidrosis, isolated, with normal sweat glands, 106190 (3), Autosomal recessive
ITPR3	147267	{Diabetes, type 1, susceptibility to}, 222100 (2), Autosomal recessive

IVD	607036	Isovaleric acidemia, 243500 (3), Autosomal recessive
IYD	612025	Thyroid dyshormonogenesis 4, 274800 (3), Autosomal recessive
JAG1	601920	Alagille syndrome 1, 118450 (3), Autosomal dominant; ?Deafness, congenital heart defects, and posterior embryotoxon, 617992 (3); Tetralogy of Fallot, 187500 (3), Autosomal dominant
JAGN1	616012	Neutropenia, severe congenital, 6, autosomal recessive, 616022 (3), Autosomal recessive
JAK2	147796	{Budd-Chiari syndrome, somatic}, 600880 (3); Erythrocytosis, somatic, 133100 (3); Leukemia, acute myeloid, somatic, 601626 (3); Myelofibrosis, somatic, 254450 (3); Polycythemia vera, somatic, 263300 (3); Thrombocythemia 3, 614521 (3), Autosomal dominant, Somatic mutation
JAK3	600173	SCID, autosomal recessive, T-negative/B-positive type, 600802 (3), Autosomal recessive
JAM3	606871	Hemorrhagic destruction of the brain, subependymal calcification, and cataracts, 613730 (3), Autosomal recessive
JPH1	605266	?Charcot-Marie-Tooth disease, axonal, autosomal dominant, type 2K, 607831 (3), Autosomal recessive, Autosomal dominant
JPH2	605267	Cardiomyopathy, hypertrophic, 17, 613873 (3), Autosomal dominant
JPH3	605268	?Huntington disease-like 2, 606438 (3), Autosomal dominant
KANK2	614610	Nephrotic syndrome 16, 617783 (3), Autosomal recessive; Palmoplantar keratoderma and woolly hair, 616099 (3), Autosomal recessive
KARS	601421	?Charcot-Marie-Tooth disease, recessive intermediate, B, 613641 (3), Autosomal recessive; Deafness, autosomal recessive 89, 613916 (3), Autosomal recessive
KAT6A	601408	Mental retardation, autosomal dominant 32, 616268 (3), Autosomal dominant
KAT6B	605880	Genitopatellar syndrome, 606170 (3), Autosomal dominant; SBBYSS syndrome, 603736 (3), Autosomal dominant
KATNB1	602703	Lissencephaly 6, with microcephaly, 616212 (3), Autosomal recessive
KBTBD13	613727	Nemaline myopathy 6, autosomal dominant, 609273 (3), Autosomal dominant
KCNA1	176260	Episodic ataxia/myokymia syndrome, 160120 (3), Autosomal dominant
KCNA5	176267	Atrial fibrillation, familial, 7, 612240 (3), Autosomal dominant
KCNB1	600397	Epileptic encephalopathy, early infantile, 26, 616056 (3), Autosomal dominant
KCNC1	176258	Epilepsy, progressive myoclonic 7, 616187 (3), Autosomal dominant
KCND3	605411	Brugada syndrome 9, 616399 (3), Autosomal dominant; Spinocerebellar ataxia 19, 607346 (3), Autosomal dominant
KCNE1	176261	Jervell and Lange-Nielsen syndrome 2, 612347 (3), Autosomal recessive; Long QT syndrome 5, 613695 (3), Autosomal dominant
KCNE2	603796	Atrial fibrillation, familial, 4, 611493 (3); Long QT syndrome 6, 613693 (3), Autosomal dominant

KCNH1	603305	Temple-Baraitser syndrome, 611816 (3), Autosomal dominant; Zimmermann-Laband syndrome 1, 135500 (3), Autosomal dominant
KCNH2	152427	Long QT syndrome 2, 613688 (3), Autosomal dominant; {Long QT syndrome 2, acquired, susceptibility to}, 613688 (3), Autosomal dominant; Short QT syndrome 1, 609620 (3)
KCNJ10	602208	Enlarged vestibular aqueduct, digenic, 600791 (3), Autosomal recessive; SESAME syndrome, 612780 (3), Autosomal recessive
KCNJ11	600937	Diabetes mellitus, transient neonatal, 3, 610582 (3), Autosomal dominant; {Diabetes mellitus, type 2, susceptibility to}, 125853 (3), Autosomal dominant; Diabetes, permanent neonatal, with or without neurologic features, 606176 (3), Autosomal recessive, Autosomal dominant; Hyperinsulinemic hypoglycemia, familial, 2, 601820 (3), Autosomal recessive; Maturity-onset diabetes of the young, type 13, 616329 (3), Autosomal dominant
KCNJ13	603208	Leber congenital amaurosis 16, 614186 (3), Autosomal recessive; Snowflake vitreoretinal degeneration, 193230 (3), Autosomal dominant
KCNJ18	613236	{Thyrotoxic periodic paralysis, susceptibility to, 2}, 613239 (3), Isolated cases
KCNJ2	600681	Andersen syndrome, 170390 (3), Autosomal dominant; Atrial fibrillation, familial, 9, 613980 (3), Autosomal dominant; Short QT syndrome 3, 609622 (3)
KCNJ5	600734	Hyperaldosteronism, familial, type III, 613677 (3), Autosomal dominant; Long QT syndrome 13, 613485 (3), Autosomal dominant
KCNJ6	600877	Keppen-Lubinsky syndrome, 614098 (3), Autosomal dominant
KCNK18	613655	{Migraine, with or without aura, susceptibility to, 13}, 613656 (3)
KCNK3	603220	Pulmonary hypertension, primary, 4, 615344 (3), Autosomal dominant
KCNK9	605874	Birk-Barel mental retardation dysmorphism syndrome, 612292 (3)
KCNMA1	600150	?Cerebellar atrophy, developmental delay, and seizures, 617643 (3), Autosomal recessive; Paroxysmal nonkinesigenic dyskinesia, 3, with or without generalized epilepsy, 609446 (3), Autosomal dominant
KCNMB1	603951	{Hypertension, diastolic, resistance to}, 608622 (3), Autosomal dominant
KCNN4	602754	Dehydrated hereditary stomatocytosis 2, 616689 (3), Autosomal dominant
KCNQ1	607542	Atrial fibrillation, familial, 3, 607554 (3), Autosomal dominant; Jervell and Lange-Nielsen syndrome, 220400 (3), Autosomal recessive; Long QT syndrome 1, 192500 (3), Autosomal dominant; {Long QT syndrome 1, acquired, susceptibility to}, 192500 (3), Autosomal dominant; Short QT syndrome 2, 609621 (3), Autosomal dominant
KCNQ2	602235	Epileptic encephalopathy, early infantile, 7, 613720 (3), Autosomal dominant; Myokymia, 121200 (3), Autosomal dominant; Seizures, benign neonatal, 1, 121200 (3), Autosomal dominant
KCNQ4	603537	Deafness, autosomal dominant 2A, 600101 (3), Autosomal dominant

KCNT1	608167	Epilepsy, nocturnal frontal lobe, 5, 615005 (3), Autosomal dominant; Epileptic encephalopathy, early infantile, 14, 614959 (3), Autosomal dominant
KCNV2	607604	Retinal cone dystrophy 3B, 610356 (3), Autosomal recessive
KCTD1	613420	Scalp-ear-nipple syndrome, 181270 (3), Autosomal dominant
KCTD17	616386	Dystonia 26, myoclonic, 616398 (3), Autosomal dominant
KCTD7	611725	Epilepsy, progressive myoclonic 3, with or without intracellular inclusions, 611726 (3), Autosomal recessive
KDF1	616758	?Ectodermal dysplasia 12, hypohidrotic/hair/tooth/nail type, 617337 (3), Autosomal dominant
KDM5C	314690	Mental retardation, X-linked, syndromic, Claes-Jensen type, 300534 (3), X-linked recessive
KDM5D	426000	No OMIM phenotype
KDM6A	300128	Kabuki syndrome 2, 300867 (3), X-linked dominant
KDR	191306	Hemangioma, capillary infantile, somatic, 602089 (3); {Hemangioma, capillary infantile, susceptibility to}, 602089 (3), Autosomal dominant
KEL	613883	[Blood group, Kell], 110900 (3)
KERA	603288	Cornea plana 2, autosomal recessive, 217300 (3), Autosomal recessive
KHDC3L	611687	Hydatidiform mole, recurrent, 2, 614293 (3), Autosomal recessive
KHK	614058	[Fructosuria], 229800 (3), Autosomal recessive
KIAA0319	609269	No OMIM phenotype
KIAA0556	616650	Joubert syndrome 26, 616784 (3), Autosomal recessive
KIAA0586	610178	Joubert syndrome 23, 616490 (3), Autosomal recessive; Short-rib thoracic dysplasia 14 with polydactyly, 616546 (3), Autosomal recessive
KIDINS220	615759	Spastic paraplegia, intellectual disability, nystagmus, and obesity, 617296 (3), Autosomal dominant
KIF11	148760	Microcephaly with or without chorioretinopathy, lymphedema, or mental retardation, 152950 (3), Autosomal dominant
KIF14	611279	?Meckel syndrome 12, 616258 (3), Autosomal recessive; Microcephaly 20, primary, autosomal recessive, 617914 (3), Autosomal recessive
KIF1A	601255	Mental retardation, autosomal dominant 9, 614255 (3), Autosomal dominant; Neuropathy, hereditary sensory, type IIC, 614213 (3), Autosomal recessive; Spastic paraplegia 30, autosomal recessive, 610357 (3), Autosomal recessive
KIF1B	605995	?Charcot-Marie-Tooth disease, type 2A1, 118210 (3), Autosomal dominant; {Neuroblastoma, susceptibility to, 1}, 256700 (3), Autosomal dominant, Isolated cases; Pheochromocytoma, 171300 (3), Autosomal dominant
KIF1BP	609367	Goldberg-Shprintzen megacolon syndrome, 609460 (3), Autosomal recessive
KIF1C	603060	Spastic ataxia 2, autosomal recessive, 611302 (3), Autosomal recessive
KIF2A	602591	Cortical dysplasia, complex, with other brain malformations 3, 615411 (3), Autosomal dominant
KIF4A	300521	?Mental retardation, X-linked 100, 300923 (3), X-linked recessive

KIF5A	602821	{Amyotrophic lateral sclerosis, susceptibility to, 25}, 617921 (3), Autosomal dominant; Myoclonus, intractable, neonatal, 617235 (3), Autosomal dominant; Spastic paraplegia 10, autosomal dominant, 604187 (3), Autosomal dominant
KIF5C	604593	Cortical dysplasia, complex, with other brain malformations 2, 615282 (3), Autosomal dominant
KIF7	611254	Acrocallosal syndrome, 200990 (3), Autosomal recessive; ?Al-Gazali-Bakalinova syndrome, 607131 (3), Autosomal recessive; ?Hydrolethalus syndrome 2, 614120 (3), Autosomal recessive; Joubert syndrome 12, 200990 (3), Autosomal recessive
KIR3DL1	604946	{AIDS, delayed/rapid progression to}, 609423 (3)
KIRREL3	607761	No OMIM phenotype
KISS1	603286	?Hypogonadotropic hypogonadism 13 with or without anosmia, 614842 (3), Autosomal recessive
KISS1R	604161	Hypogonadotropic hypogonadism 8 with or without anosmia, 614837 (3), Autosomal recessive; ?Precocious puberty, central, 1, 176400 (3), Autosomal dominant
KIT	164920	Gastrointestinal stromal tumor, familial, 606764 (3), Autosomal dominant, Isolated cases; Germ cell tumors, somatic, 273300 (3); Leukemia, acute myeloid, 601626 (3), Autosomal dominant; Mastocytosis, cutaneous, 154800 (3), Autosomal dominant; Mastocytosis, systemic, somatic, 154800 (3); Piebaldism, 172800 (3), Autosomal dominant
KITLG	184745	Deafness, autosomal dominant 69, unilateral or asymmetric, 616697 (3), Autosomal dominant; Hyperpigmentation with or without hypopigmentation, 145250 (3), Autosomal dominant; [Skin/hair/eye pigmentation 7, blond/brown hair], 611664 (3)
KLF1	600599	Blood group--Lutheran inhibitor, 111150 (3); Dyserythropoietic anemia, congenital, type IV, 613673 (3), Autosomal dominant; [Hereditary persistence of fetal hemoglobin], 613566 (3)
KLF11	603301	Maturity-onset diabetes of the young, type VII, 610508 (3)
KLF13	605328	No OMIM phenotype
KLF6	602053	Gastric cancer, somatic, 613659 (3); Prostate cancer, somatic, 176807 (3)
KLHDC8B	613169	{Hodgkin lymphoma, susceptibility to}, 236000 (3), Autosomal recessive
KLHL10	608778	Spermatogenic failure 11, 615081 (3), Autosomal dominant
KLHL15	300980	Mental retardation, X-linked 103, 300982 (3), X-linked recessive
KLHL24	611295	Epidermolysis bullosa simplex, generalized, with scarring and hair loss, 617294 (3), Autosomal dominant
KLHL3	605775	Pseudohypoaldosteronism, type IID, 614495 (3), Autosomal recessive, Autosomal dominant
KLHL40	615340	Nemaline myopathy 8, autosomal recessive, 615348 (3)
KLHL41	607701	Nemaline myopathy 9, 615731 (3), Autosomal recessive
KLK1	147910	[Kallikrein, decreased urinary activity of], 615953 (3)

KLK4	603767	Amelogenesis imperfecta, type IIA1, 204700 (3), Autosomal recessive
KLKB1	229000	Fletcher factor (prekallikrein) deficiency, 612423 (3), Autosomal recessive
KLLN	612105	Cowden syndrome 4, 615107 (3)
KMT2B	606834	Dystonia 28, childhood-onset, 617284 (3), Autosomal dominant
KMT2D	602113	Kabuki syndrome 1, 147920 (3), Autosomal dominant
KNG1	612358	[High molecular weight kininogen deficiency], 228960 (3), Autosomal recessive; [Kininogen deficiency], 228960 (3), Autosomal recessive
KPTN	615620	Mental retardation, autosomal recessive 41, 615637 (3), Autosomal recessive
KREMEN1	609898	Ectodermal dysplasia 13, hair/tooth type, 617392 (3), Autosomal recessive
KRIT1	604214	Cavernous malformations of CNS and retina, 116860 (3), Autosomal dominant; Cerebral cavernous malformations-1, 116860 (3), Autosomal dominant; Hyperkeratotic cutaneous capillary-venous malformations associated with cerebral capillary malformations, 116860 (3), Autosomal dominant
KRT1	139350	Epidermolytic hyperkeratosis, 113800 (3), Autosomal recessive, Autosomal dominant; Ichthyosis histrix, Curth-Macklin type, 146590 (3), Autosomal dominant; Ichthyosis, cyclic, with epidermolytic hyperkeratosis, 607602 (3), Autosomal dominant; Keratosis palmoplantaris striata III, 607654 (3); Palmoplantar keratoderma, epidermolytic, 144200 (3), Autosomal dominant; Palmoplantar keratoderma, nonepidermolytic, 600962 (3), Autosomal dominant
KRT10	148080	Epidermolytic hyperkeratosis, 113800 (3), Autosomal recessive, Autosomal dominant; Ichthyosis with confetti, 609165 (3), Autosomal dominant; Ichthyosis, cyclic, with epidermolytic hyperkeratosis, 607602 (3), Autosomal dominant
KRT12	601687	Meesmann corneal dystrophy, 122100 (3), Autosomal dominant
KRT13	148065	White sponge nevus 2, 615785 (3), Autosomal dominant
KRT14	148066	Dermatopathia pigmentosa reticularis, 125595 (3), Autosomal dominant; Epidermolysis bullosa simplex, Dowling-Meara type, 131760 (3), Autosomal dominant; Epidermolysis bullosa simplex, Koebner type, 131900 (3), Autosomal dominant; Epidermolysis bullosa simplex, Weber-Cockayne type, 131800 (3), Autosomal dominant; Epidermolysis bullosa simplex, recessive 1, 601001 (3), Autosomal recessive; Naegeli-Franceschetti-Jadassohn syndrome, 161000 (3), Autosomal dominant
KRT16	148067	Pachyonychia congenita 1, 167200 (3), Autosomal dominant; Palmoplantar keratoderma, nonepidermolytic, focal, 613000 (3), Autosomal dominant
KRT17	148069	Pachyonychia congenita 2, 167210 (3), Autosomal dominant; Steatocystoma multiplex, 184500 (3), Autosomal dominant
KRT2	600194	Ichthyosis bullosa of Siemens, 146800 (3), Autosomal dominant
KRT25	616646	Woolly hair, autosomal recessive 3, 616760 (3), Autosomal recessive

KRT3	148043	Meesmann corneal dystrophy, 122100 (3), Autosomal dominant
KRT4	123940	White sponge nevus 1, 193900 (3), Autosomal dominant
KRT5	148040	Dowling-Degos disease 1, 179850 (3), Autosomal dominant; Epidermolysis bullosa simplex, Dowling-Meara type, 131760 (3), Autosomal dominant; Epidermolysis bullosa simplex, Koebner type, 131900 (3), Autosomal dominant; Epidermolysis bullosa simplex, Weber-Cockayne type, 131800 (3), Autosomal dominant; Epidermolysis bullosa simplex, recessive 1, 601001 (3), Autosomal recessive; Epidermolysis bullosa simplex-MCR, 609352 (3); Epidermolysis bullosa simplex-MP, 131960 (3), Autosomal dominant
KRT6A	148041	Pachyonychia congenita 3, 615726 (3)
KRT6B	148042	Pachyonychia congenita 4, 615728 (3)
KRT6C	612315	Palmoplantar keratoderma, nonepidermolytic, focal or diffuse, 615735 (3), Autosomal dominant
KRT71	608245	?Hypotrichosis 13, 615896 (3), Autosomal dominant
KRT74	608248	?Ectodermal dysplasia 7, hair/nail type, 614929 (3), Autosomal recessive; ?Hypotrichosis 3, 613981 (3), Autosomal dominant; Woolly hair, autosomal dominant, 194300 (3), Autosomal dominant
KRT75	609025	{Pseudofolliculitis barbae, susceptibility to}, 612318 (3)
KRT81	602153	Monilethrix, 158000 (3), Autosomal dominant
KRT83	602765	Erythrokeratodermia variabilis et progressiva 5, 617756 (3), Autosomal recessive; Monilethrix, 158000 (3), Autosomal dominant
KRT85	602767	Ectodermal dysplasia 4, hair/nail type, 602032 (3), Autosomal recessive
KRT86	601928	Monilethrix, 158000 (3), Autosomal dominant
KRT9	607606	Palmoplantar keratoderma, epidermolytic, 144200 (3), Autosomal dominant
KY	605739	Myopathy, myofibrillar, 7, 617114 (3), Autosomal recessive
KYNU	605197	?Hydroxykynureninuria, 236800 (3), Autosomal recessive; Vertebral, cardiac, renal, and limb defects syndrome 2, 617661 (3), Autosomal recessive
L1CAM	308840	CRASH syndrome, 303350 (3), X-linked recessive; Corpus callosum, partial agenesis of, 304100 (3), X-linked recessive; Hydrocephalus due to aqueductal stenosis, 307000 (3), X-linked recessive; Hydrocephalus with Hirschsprung disease, 307000 (3), X-linked recessive; Hydrocephalus with congenital idiopathic intestinal pseudoobstruction, 307000 (3), X-linked recessive; MASA syndrome, 303350 (3), X-linked recessive
L2HGDH	609584	L-2-hydroxyglutaric aciduria, 236792 (3), Autosomal recessive
LAMA1	150320	Poretti-Boltshauser syndrome, 615960 (3), Autosomal recessive
LAMA2	156225	Muscular dystrophy, congenital merosin-deficient, 607855 (3), Autosomal recessive; Muscular dystrophy, congenital, due to partial LAMA2 deficiency, 607855 (3), Autosomal recessive

LAMA3	600805	Epidermolysis bullosa, generalized atrophic benign, 226650 (3), Autosomal recessive; Epidermolysis bullosa, junctional, Herlitz type, 226700 (3), Autosomal recessive; Laryngoonychocutaneous syndrome, 245660 (3), Autosomal recessive
LAMA4	600133	Cardiomyopathy, dilated, 1JJ, 615235 (3), Autosomal dominant
LAMB1	150240	Lissencephaly 5, 615191 (3), Autosomal recessive
LAMB2	150325	Nephrotic syndrome, type 5, with or without ocular abnormalities, 614199 (3); Pierson syndrome, 609049 (3), Autosomal recessive
LAMB3	150310	Amelogenesis imperfecta, type IA, 104530 (3), Autosomal dominant; Epidermolysis bullosa, junctional, Herlitz type, 226700 (3), Autosomal recessive; Epidermolysis bullosa, junctional, non-Herlitz type, 226650 (3), Autosomal recessive
LAMC2	150292	Epidermolysis bullosa, junctional, Herlitz type, 226700 (3), Autosomal recessive; Epidermolysis bullosa, junctional, non-Herlitz type, 226650 (3), Autosomal recessive
LAMC3	604349	Cortical malformations, occipital, 614115 (3), Autosomal recessive
LAMP2	309060	Danon disease, 300257 (3), X-linked dominant
LAMTOR2	610389	Immunodeficiency due to defect in MAPBP-interacting protein, 610798 (3), Autosomal recessive
LARGE1	603590	Muscular dystrophy-dystroglycanopathy (congenital with brain and eye anomalies), type A, 6, 613154 (3), Autosomal recessive; Muscular dystrophy-dystroglycanopathy (congenital with mental retardation), type B, 6, 608840 (3), Autosomal recessive
LARP7	612026	Alazami syndrome, 615071 (3), Autosomal recessive
LARS2	604544	?Hydrops, lactic acidosis, and sideroblastic anemia, 617021 (3), Autosomal recessive; Perrault syndrome 4, 615300 (3), Autosomal recessive
LAS1L	300964	Wilson-Turner syndrome, 309585 (3), X-linked recessive
LBR	600024	Greenberg skeletal dysplasia, 215140 (3), Autosomal recessive; Pelger-Huet anomaly, 169400 (3), Autosomal dominant; Pelger-Huet anomaly with mild skeletal anomalies, 618019 (3); ?Reynolds syndrome, 613471 (3), Autosomal dominant
LCA5	611408	Leber congenital amaurosis 5, 604537 (3)
LCAT	606967	Fish-eye disease, 136120 (3), Autosomal recessive; Norum disease, 245900 (3), Autosomal recessive
LCK	153390	?Immunodeficiency 22, 615758 (3), Autosomal recessive
LCT	603202	Lactase deficiency, congenital, 223000 (3), Autosomal recessive
LDB3	605906	Cardiomyopathy, dilated, 1C, with or without LVNC, 601493 (3), Autosomal dominant; Cardiomyopathy, hypertrophic, 24, 601493 (3), Autosomal dominant; Left ventricular noncompaction 3, 601493 (3), Autosomal dominant; Myopathy, myofibrillar, 4, 609452 (3), Autosomal dominant
LDHA	150000	Glycogen storage disease XI, 612933 (3), Autosomal recessive
LDLR	606945	Hypercholesterolemia, familial, 143890 (3), Autosomal dominant; LDL cholesterol level QTL2, 143890 (3), Autosomal dominant

LDLRAP1	605747	Hypercholesterolemia, familial, autosomal recessive, 603813 (3), Autosomal recessive
LEMD3	607844	Buschke-Ollendorff syndrome, 166700 (3), Autosomal dominant; Osteopoikilosis with or without melorheostosis, 166700 (3), Autosomal dominant
LEP	164160	Obesity, morbid, due to leptin deficiency, 614962 (3), Autosomal recessive
LEPR	601007	Obesity, morbid, due to leptin receptor deficiency, 614963 (3)
LETM1	604407	No OMIM phenotype
LFNG	602576	?Spondylocostal dysostosis 3, autosomal recessive, 609813 (3)
LGALS2	150571	{Myocardial infarction, susceptibility to}, 608446 (3)
LGI1	604619	Epilepsy, familial temporal lobe, 1, 600512 (3), Autosomal dominant
LGR4	606666	{Bone mineral density, low, susceptibility to}, 615311 (3)
LHB	152780	Hypogonadotropic hypogonadism 23 with or without anosmia, 228300 (3), Autosomal recessive
LHCGR	152790	Leydig cell adenoma, somatic, with precocious puberty, 176410 (3); Leydig cell hypoplasia with hypergonadotropic hypogonadism, 238320 (3), Autosomal recessive; Leydig cell hypoplasia with pseudohermaphroditism, 238320 (3), Autosomal recessive; Luteinizing hormone resistance, female, 238320 (3), Autosomal recessive; Precocious puberty, male, 176410 (3), Autosomal dominant
LHFPL5	609427	Deafness, autosomal recessive 67, 610265 (3), Autosomal recessive
LHX3	600577	Pituitary hormone deficiency, combined, 3, 221750 (3), Autosomal recessive
LHX4	602146	Pituitary hormone deficiency, combined, 4, 262700 (3), Autosomal dominant
LIAS	607031	Hyperglycinemia, lactic acidosis, and seizures, 614462 (3), Autosomal recessive
LIFR	151443	Stuve-Wiedemann syndrome/Schwartz-Jampel type 2 syndrome, 601559 (3), Autosomal recessive
LIM2	154045	Cataract 19, multiple types, 615277 (3), Autosomal recessive
LIMS2	607908	Muscular dystrophy, limb-girdle, type 2W, 616827 (3), Autosomal recessive
LINS1	610350	Mental retardation, autosomal recessive 27, 614340 (3), Autosomal recessive
LIPA	613497	Cholesteryl ester storage disease, 278000 (3), Autosomal recessive; Wolman disease, 278000 (3), Autosomal recessive
LIPE	151750	Lipodystrophy, familial partial, type 6, 615980 (3), Autosomal recessive
LIPH	607365	Hypotrichosis 7, 604379 (3), Autosomal recessive; Woolly hair, autosomal recessive 2 with or without hypotrichosis, 604379 (3), Autosomal recessive
LIPI	609252	{Hypertriglyceridemia, susceptibility to}, 145750 (3), Autosomal dominant
LIPN	613924	Ichthyosis, congenital, autosomal recessive 8, 613943 (3), Autosomal recessive

LMAN1	601567	Combined factor V and VIII deficiency, 227300 (3), Autosomal recessive
LMAN2L	609552	?Mental retardation, autosomal recessive, 52, 616887 (3), Autosomal recessive
LMBR1	605522	Acheiropody, 200500 (3), Autosomal recessive; Hypoplastic or aplastic tibia with polydactyly, 188740 (3), Autosomal dominant; Laurin-Sandrow syndrome, 135750 (3), Autosomal dominant; Polydactyly, preaxial type II, 174500 (3), Autosomal dominant; Syndactyly, type IV, 186200 (3), Autosomal dominant; Triphalangeal thumb, type I, 174500 (3), Autosomal dominant; Triphalangeal thumb-polysyndactyly syndrome, 174500 (3), Autosomal dominant
LMBRD1	612625	Methylmalonic aciduria and homocystinuria, cblF type, 277380 (3), Autosomal recessive
LMNA	150330	Cardiomyopathy, dilated, 1A, 115200 (3), Autosomal dominant; Charcot-Marie-Tooth disease, type 2B1, 605588 (3), Autosomal recessive; Emery-Dreifuss muscular dystrophy 2, AD, 181350 (3), Autosomal dominant; Emery-Dreifuss muscular dystrophy 3, AR, 616516 (3), Autosomal recessive; Heart-hand syndrome, Slovenian type, 610140 (3), Autosomal dominant; Hutchinson-Gilford progeria, 176670 (3), Autosomal recessive, Autosomal dominant; Lipodystrophy, familial partial, type 2, 151660 (3), Autosomal dominant; Malouf syndrome, 212112 (3), Autosomal dominant; Mandibuloacral dysplasia, 248370 (3), Autosomal recessive; Muscular dystrophy, congenital, 613205 (3), Autosomal dominant; Muscular dystrophy, limb-girdle, type 1B, 159001 (3), Autosomal dominant; Restrictive dermopathy, lethal, 275210 (3), Autosomal recessive
LMNB1	150340	Leukodystrophy, adult-onset, autosomal dominant, 169500 (3), Autosomal dominant
LMNB2	150341	?Epilepsy, progressive myoclonic, 9, 616540 (3), Autosomal recessive; {Lipodystrophy, partial, acquired, susceptibility to}, 608709 (3), Autosomal dominant
LMOD3	616112	Nemaline myopathy 10, 616165 (3), Autosomal recessive
LMX1B	602575	Nail-patella syndrome, 161200 (3), Autosomal dominant
LONP1	605490	CODAS syndrome, 600373 (3), Autosomal recessive
LOR	152445	Vohwinkel syndrome with ichthyosis, 604117 (3), Autosomal dominant
LOX	153455	Aortic aneurysm, familial thoracic 10, 617168 (3), Autosomal dominant
LOXHD1	613072	Deafness, autosomal recessive 77, 613079 (3), Autosomal recessive
LOXL1	153456	{Exfoliation syndrome, susceptibility to}, 177650 (3), Autosomal dominant
LPAR6	609239	Hypotrichosis 8, 278150 (3), Autosomal recessive; Woolly hair, autosomal recessive 1, with or without hypotrichosis, 278150 (3), Autosomal recessive
LPIN2	605519	Majeed syndrome, 609628 (3)
LPL	609708	Combined hyperlipidemia, familial, 144250 (3), Autosomal dominant; [High density lipoprotein cholesterol level QTL 11] (3); Lipoprotein lipase deficiency, 238600 (3), Autosomal recessive

LRAT	604863	Leber congenital amaurosis 14, 613341 (3), Autosomal recessive; Retinal dystrophy, early-onset severe, 613341 (3), Autosomal recessive; Retinitis pigmentosa, juvenile, 613341 (3), Autosomal recessive
LRBA	606453	Immunodeficiency, common variable, 8, with autoimmunity, 614700 (3), Autosomal recessive
LRIG2	608869	Urofacial syndrome 2, 615112 (3), Autosomal recessive
LRIT3	615004	Night blindness, congenital stationary (complete), 1F, autosomal recessive, 615058 (3), Autosomal recessive
LRMDA	614537	Albinism, oculocutaneous, type VII, 615179 (3), Autosomal recessive
LRP1	107770	?Keratosis pilaris atrophicans, 604093 (3), Autosomal recessive
LRP2	600073	Donnai-Barrow syndrome, 222448 (3), Autosomal recessive
LRP4	604270	Cenani-Lenz syndactyly syndrome, 212780 (3), Autosomal recessive; ?Myasthenic syndrome, congenital, 17, 616304 (3), Autosomal recessive; Sclerosteosis 2, 614305 (3), Autosomal recessive, Autosomal dominant
LRP5	603506	[Bone mineral density variability 1], 601884 (3), Autosomal dominant; Exudative vitreoretinopathy 4, 601813 (3), Autosomal recessive, Autosomal dominant; Hyperostosis, endosteal, 144750 (3), Autosomal dominant; Osteopetrosis, autosomal dominant 1, 607634 (3), Autosomal dominant; Osteoporosis-pseudoglioma syndrome, 259770 (3), Autosomal recessive; {Osteoporosis}, 166710 (3), Autosomal dominant; Osteosclerosis, 144750 (3), Autosomal dominant; Polycystic liver disease 4 with or without kidney cysts, 617875 (3), Autosomal dominant; van Buchem disease, type 2, 607636 (3), Autosomal dominant
LRP6	603507	{Coronary artery disease, autosomal dominant, 2}, 610947 (3), Autosomal dominant; Tooth agenesis, selective, 7, 616724 (3), Autosomal dominant
LRP8	602600	{Myocardial infarction, susceptibility to}, 608446 (3)
LRPAP1	104225	Myopia 23, autosomal recessive, 615431 (3), Autosomal recessive
LRPPRC	607544	Leigh syndrome, French-Canadian type, 220111 (3), Autosomal recessive
LRRC6	614930	Ciliary dyskinesia, primary, 19, 614935 (3), Autosomal recessive
LRRC8A	608360	?Agammaglobulinemia 5, 613506 (3), Autosomal dominant
LRRK2	609007	{Parkinson disease 8}, 607060 (3), Autosomal dominant
LRSAM1	610933	Charcot-Marie-Tooth disease, axonal, type 2P, 614436 (3), Autosomal recessive, Autosomal dominant
LRTOMT	612414	Deafness, autosomal recessive 63, 611451 (3), Autosomal recessive
LTA	153440	{Leprosy, susceptibility to, 4}, 610988 (3); {Myocardial infarction, susceptibility to}, 608446 (3); {Psoriatic arthritis, susceptibility to}, 607507 (3)
LTBP2	602091	Glaucoma 3, primary congenital, D, 613086 (3); Microspherophakia and/or megalocornea, with ectopia lentis and with or without secondary glaucoma, 251750 (3), Autosomal recessive; ?Weill-Marchesani syndrome 3, recessive, 614819 (3), Autosomal recessive

LTBP3	602090	Dental anomalies and short stature, 601216 (3), Autosomal recessive; Geleophysic dysplasia 3, 617809 (3), Autosomal dominant
LTC4S	246530	Leukotriene C4 synthase deficiency, 614037 (1), Autosomal recessive
LYRM4	613311	?Combined oxidative phosphorylation deficiency 19, 615595 (3), Autosomal recessive
LYRM7	615831	Mitochondrial complex III deficiency, nuclear type 8, 615838 (3), Autosomal recessive
LYST	606897	Chediak-Higashi syndrome, 214500 (3), Autosomal recessive
LYZ	153450	Amyloidosis, renal, 105200 (3), Autosomal dominant
LZTFL1	606568	Bardet-Biedl syndrome 17, 615994 (3), Autosomal recessive
LZTR1	600574	Noonan syndrome 10, 616564 (3), Autosomal dominant; {Schwannomatosis-2, susceptibility to}, 615670 (3), Autosomal dominant
LZTS1	606551	Esophageal squamous cell carcinoma, 133239 (3), Autosomal dominant
MAB21L2	604357	Microphthalmia/coloboma and skeletal dysplasia syndrome, 615877 (3), Autosomal recessive, Autosomal dominant
MAD2L2	604094	?Fanconi anemia, complementation group V, 617243 (3), Autosomal recessive
MAF	177075	Ayme-Gripp syndrome, 601088 (3), Autosomal dominant; Cataract 21, multiple types, 610202 (3), Autosomal dominant
MAFB	608968	Duane retraction syndrome 3, 617041 (3), Autosomal dominant; Multicentric carpotarsal osteolysis syndrome, 166300 (3), Autosomal dominant
MAG	159460	Spastic paraplegia 75, autosomal recessive, 616680 (3), Autosomal recessive
MAGED2	300470	Bartter syndrome, type 5, antenatal, transient, 300971 (3), X-linked recessive
MAGEL2	605283	Schaaf-Yang syndrome, 615547 (3), Autosomal dominant
MAGT1	300715	Immunodeficiency, X-linked, with magnesium defect, Epstein-Barr virus infection and neoplasia, 300853 (3)
MAK	154235	Retinitis pigmentosa 62, 614181 (3), Autosomal recessive
MALT1	604860	Immunodeficiency 12, 615468 (3), Autosomal recessive
MAMLD1	300120	Hypospadias 2, X-linked, 300758 (3), X-linked recessive
MAN1B1	604346	Mental retardation, autosomal recessive 15, 614202 (3), Autosomal recessive
MAN2B1	609458	Mannosidosis, alpha-, types I and II, 248500 (3), Autosomal recessive
MANBA	609489	Mannosidosis, beta, 248510 (3), Autosomal recessive
MAOA	309850	{Antisocial behavior}, 300615 (3), X-linked recessive; Brunner syndrome, 300615 (3), X-linked recessive
MAP2K1	176872	Cardiofaciocutaneous syndrome 3, 615279 (3)
MAP2K2	601263	Cardiofaciocutaneous syndrome 4, 615280 (3)
MAP3K1	600982	46XY sex reversal 6, 613762 (3), Autosomal dominant

MAP3K20	609479	Centronuclear myopathy 6 with fiber-type disproportion, 617760 (3), Autosomal recessive; Split-foot malformation with mesoaxial polydactyly, 616890 (3), Autosomal recessive
MAP3K7	602614	Cardiospondylocarpofacial syndrome, 157800 (3), Autosomal dominant; Frontometaphyseal dysplasia 2, 617137 (3), Autosomal dominant
MAP3K8	191195	Lung cancer, somatic, 211980 (3)
MAPK8IP1	604641	{Diabetes mellitus, noninsulin-dependent}, 125853 (3), Autosomal dominant
MAPKAPK3	602130	?Macular dystrophy, patterned, 3, 617111 (3), Autosomal dominant
MAPKBP1	616786	Nephronophthisis 20, 617271 (3), Autosomal recessive
MAPRE2	605789	Symmetric circumferential skin creases, congenital, 2, 616734 (3), Autosomal dominant
MAPT	157140	Dementia, frontotemporal, with or without parkinsonism, 600274 (3), Autosomal dominant; {Parkinson disease, susceptibility to}, 168600 (3), Isolated cases, Multifactorial; Pick disease, 172700 (3), Autosomal dominant, Isolated cases; Supranuclear palsy, progressive, 601104 (3), Autosomal dominant; Supranuclear palsy, progressive atypical, 260540 (3), Autosomal recessive
MARS	156560	Charcot-Marie-Tooth disease, axonal, type 2U, 616280 (3), Autosomal dominant; Interstitial lung and liver disease, 615486 (3), Autosomal recessive
MARS2	609728	?Combined oxidative phosphorylation deficiency 25, 616430 (3), Autosomal recessive; Spastic ataxia 3, autosomal recessive, 611390 (3), Autosomal recessive
MARVELD2	610572	Deafness, autosomal recessive 49, 610153 (3), Autosomal recessive
MASP1	600521	3MC syndrome 1, 257920 (3), Autosomal recessive
MASP2	605102	MASP2 deficiency, 613791 (3), Autosomal recessive
MASTL	608221	No OMIM phenotype
MAT1A	610550	Hypermethioninemia, persistent, autosomal dominant, due to methionine adenosyltransferase I/III deficiency, 250850 (3), Autosomal recessive, Autosomal dominant; Methionine adenosyltransferase deficiency, autosomal recessive, 250850 (3), Autosomal recessive, Autosomal dominant
MATN3	602109	Epiphyseal dysplasia, multiple, 5, 607078 (3), Autosomal dominant; {Osteoarthritis susceptibility 2}, 140600 (3), Autosomal dominant; ?Spondyloepimetaphyseal dysplasia, 608728 (3), Autosomal recessive
MATR3	164015	Amyotrophic lateral sclerosis 21, 606070 (3), Autosomal dominant
MAX	154950	{Pheochromocytoma, susceptibility to}, 171300 (3), Autosomal dominant
MBL2	154545	{Chronic infections, due to MBL deficiency}, 614372 (3), Autosomal dominant
MBOAT7	606048	Mental retardation, autosomal recessive 57, 617188 (3), Autosomal recessive

MBTPS2	300294	IFAP syndrome with or without BRESHECK syndrome, 308205 (3), X-linked recessive; Keratosis follicularis spinulosa decalvans, X-linked, 308800 (3), X-linked recessive; ?Olmsted syndrome, X-linked, 300918 (3), X-linked recessive
MC1R	155555	{Albinism, oculocutaneous, type II, modifier of}, 203200 (3), Autosomal recessive; [Analgesia from kappa-opioid receptor agonist, female-specific], 613098 (3); {Melanoma, cutaneous malignant, 5}, 613099 (3); [Skin/hair/eye pigmentation 2, blond hair/fair skin], 266300 (3), Autosomal recessive; [Skin/hair/eye pigmentation 2, red hair/fair skin], 266300 (3), Autosomal recessive; {UV-induced skin damage}, 266300 (3), Autosomal recessive
MC3R	155540	{Mycobacterium tuberculosis, protection against}, 607948 (3); {Obesity, severe, susceptibility to, BMIQ9}, 602025 (3)
MC4R	155541	Obesity, autosomal dominant, 601665 (3), Autosomal recessive, Autosomal dominant, Multifactorial
MCCC2	609014	3-Methylcrotonyl-CoA carboxylase 2 deficiency, 210210 (3), Autosomal recessive
MCEE	608419	Methylmalonyl-CoA epimerase deficiency, 251120 (3), Autosomal recessive
MCM2	116945	?Deafness, autosomal dominant 70, 616968 (3), Autosomal dominant
MCM4	602638	Immunodeficiency 54, 609981 (3), Autosomal recessive
MCM6	601806	Lactase persistence/nonpersistence, 223100 (3), Autosomal dominant
MCM8	608187	?Premature ovarian failure 10, 612885 (3), Autosomal recessive
MCM9	610098	Ovarian dysgenesis 4, 616185 (3), Autosomal recessive
MCOLN1	605248	Mucolipidosis IV, 252650 (3), Autosomal recessive
MCPH1	607117	Microcephaly 1, primary, autosomal recessive, 251200 (3), Autosomal recessive
MDH2	154100	Epileptic encephalopathy, early infantile, 51, 617339 (3), Autosomal recessive
MDM2	164785	{Accelerated tumor formation, susceptibility to}, 614401 (3)
MECOM	165215	Radioulnar synostosis with amegakaryocytic thrombocytopenia 2, 616738 (3), Autosomal dominant
MECP2	300005	{Autism susceptibility, X-linked 3}, 300496 (3), Isolated cases, X-linked, Multifactorial; Encephalopathy, neonatal severe, 300673 (3), X-linked recessive; Mental retardation, X-linked syndromic, Lubs type, 300260 (3), X-linked recessive; Mental retardation, X-linked, syndromic 13, 300055 (3), X-linked recessive; Rett syndrome, 312750 (3), X-linked dominant; Rett syndrome, atypical, 312750 (3), X-linked dominant; Rett syndrome, preserved speech variant, 312750 (3), X-linked dominant
MECR	608205	Dystonia, childhood-onset, with optic atrophy and basal ganglia abnormalities, 617282 (3), Autosomal recessive
MED12	300188	Lujan-Fryns syndrome, 309520 (3), X-linked recessive; Ohdo syndrome, X-linked, 300895 (3), X-linked recessive; Opitz-Kaveggia syndrome, 305450 (3), X-linked recessive

MED13L	608771	Mental retardation and distinctive facial features with or without cardiac defects, 616789 (3), Autosomal dominant; Transposition of the great arteries, dextro-looped 1, 608808 (3), Autosomal dominant
MED17	603810	Microcephaly, postnatal progressive, with seizures and brain atrophy, 613668 (3), Autosomal recessive
MED25	610197	Basel-Vanagait-Smirin-Yosef syndrome, 616449 (3), Autosomal recessive; ?Charcot-Marie-Tooth disease, type 2B2, 605589 (3), Autosomal recessive
MEF2A	600660	{Coronary artery disease, autosomal dominant, 1}, 608320 (3), Autosomal dominant
MEF2C	600662	Chromosome 5q14.3 deletion syndrome, 613443 (4), Autosomal dominant; Mental retardation, stereotypic movements, epilepsy, and/or cerebral malformations, 613443 (3), Autosomal dominant
MEFV	608107	Familial Mediterranean fever, AD, 134610 (3), Autosomal dominant; Familial Mediterranean fever, AR, 249100 (3), Autosomal recessive
MEGF10	612453	Myopathy, areflexia, respiratory distress, and dysphagia, early-onset, 614399 (3), Autosomal recessive; Myopathy, areflexia, respiratory distress, and dysphagia, early-onset, mild variant, 614399 (3), Autosomal recessive
MEGF8	604267	Carpenter syndrome 2, 614976 (3), Autosomal recessive
MEN1	613733	Adrenal adenoma, somatic (3); Angiofibroma, somatic (3); Carcinoid tumor of lung (3); Lipoma, somatic (3); Multiple endocrine neoplasia 1, 131100 (3), Autosomal dominant; Parathyroid adenoma, somatic (3)
MEOX1	600147	Klippel-Feil syndrome 2, 214300 (3), Autosomal recessive
MERTK	604705	Retinitis pigmentosa 38, 613862 (3), Autosomal recessive
MESP2	605195	Spondylocostal dysostosis 2, autosomal recessive, 608681 (3)
MET	164860	?Deafness, autosomal recessive 97, 616705 (3), Autosomal recessive; Hepatocellular carcinoma, childhood type, somatic, 114550 (3); {Osteofibrous dysplasia, susceptibility to}, 607278 (3), Autosomal dominant; Renal cell carcinoma, papillary, 1, familial and somatic, 605074 (3)
METTL23	615262	Mental retardation, autosomal recessive 44, 615942 (3), Autosomal recessive
MFAP5	601103	Aortic aneurysm, familial thoracic 9, 616166 (3), Autosomal dominant
MFN2	608507	Charcot-Marie-Tooth disease, axonal, type 2A2A, 609260 (3), Autosomal dominant; Charcot-Marie-Tooth disease, axonal, type 2A2B, 617087 (3), Autosomal recessive; Hereditary motor and sensory neuropathy VIA, 601152 (3), Autosomal dominant
MFRP	606227	Microphthalmia, isolated 5, 611040 (3), Autosomal recessive; Nanophthalmos 2, 609549 (3)
MFSD2A	614397	Microcephaly 15, primary, autosomal recessive, 616486 (3), Autosomal recessive
MFSD8	611124	Ceroid lipofuscinosis, neuronal, 7, 610951 (3), Autosomal recessive; Macular dystrophy with central cone involvement, 616170 (3), Autosomal recessive

MGAT2	602616	Congenital disorder of glycosylation, type IIa, 212066 (3), Autosomal recessive
MGME1	615076	Mitochondrial DNA depletion syndrome 11, 615084 (3), Autosomal recessive
MGP	154870	Keutel syndrome, 245150 (3), Autosomal recessive
MIB1	608677	Left ventricular noncompaction 7, 615092 (3), Autosomal dominant
MICU1	605084	Myopathy with extrapyramidal signs, 615673 (3), Autosomal recessive
MID1	300552	Opitz GBBB syndrome, type I, 300000 (3), X-linked recessive
MID2	300204	?Mental retardation, X-linked 101, 300928 (3), X-linked recessive
MIF	153620	{Rheumatoid arthritis, systemic juvenile, susceptibility to}, 604302 (3)
MINPP1	605391	Thyroid carcinoma, follicular, 188470 (3), Autosomal dominant
MIP	154050	Cataract 15, multiple types, 615274 (3), Autosomal dominant
MIPEP	602241	Combined oxidative phosphorylation deficiency 31, 617228 (3), Autosomal recessive
MIR204	610942	?Retinal dystrophy and iris coloboma with or without cataract, 616722 (3), Autosomal dominant
MITF	156845	COMMAD syndrome, 617306 (3), Autosomal recessive; {Melanoma, cutaneous malignant, susceptibility to, 8}, 614456 (3); Tietz albinism-deafness syndrome, 103500 (3), Autosomal dominant; Waardenburg syndrome, type 2A, 193510 (3), Autosomal dominant; Waardenburg syndrome/ocular albinism, digenic, 103470 (3), Autosomal dominant
MKKS	604896	Bardet-Biedl syndrome 6, 605231 (3), Autosomal recessive; McKusick-Kaufman syndrome, 236700 (3), Autosomal recessive
MKRN3	603856	Precocious puberty, central, 2, 615346 (3), Autosomal dominant
MKS1	609883	Bardet-Biedl syndrome 13, 615990 (3), Autosomal recessive; Joubert syndrome 28, 617121 (3), Autosomal recessive; Meckel syndrome 1, 249000 (3), Autosomal recessive
MLC1	605908	Megalencephalic leukoencephalopathy with subcortical cysts, 604004 (3), Autosomal recessive
MLH3	604395	Colorectal cancer, hereditary nonpolyposis, type 7, 614385 (3); Colorectal cancer, somatic, 114500 (3); {Endometrial cancer, susceptibility to}, 608089 (3)
MLLT11	604684	No OMIM phenotype
MLPH	606526	Griscelli syndrome, type 3, 609227 (3), Autosomal recessive
MLXIPL	605678	No OMIM phenotype
MLYCD	606761	Malonyl-CoA decarboxylase deficiency, 248360 (3), Autosomal recessive
MMAA	607481	Methylmalonic aciduria, vitamin B12-responsive, 251100 (3), Autosomal recessive
MMAB	607568	Methylmalonic aciduria, vitamin B12-responsive, due to defect in synthesis of adenosylcobalamin, cblB complementation type, 251110 (3), Autosomal recessive
MMACHC	609831	Methylmalonic aciduria and homocystinuria, cblC type, 277400 (3), Autosomal recessive

MMADHC	611935	Homocystinuria, cblD type, variant 1, 277410 (3), Autosomal recessive; Methylmalonic aciduria and homocystinuria, cblD type, 277410 (3), Autosomal recessive; Methylmalonic aciduria, cblD type, variant 2, 277410 (3), Autosomal recessive
MME	120520	Charcot-Marie-Tooth disease, axonal, type 2T, 617017 (3), Autosomal recessive, Autosomal dominant; ?Spinocerebellar ataxia 43, 617018 (3), Autosomal dominant
MMP1	120353	COPD, rate of decline of lung function in, 606963 (3); {Epidermolysis bullosa dystrophica, autosomal recessive, modifier of}, 226600 (3), Autosomal recessive
MMP13	600108	Metaphyseal anadysplasia 1, 602111 (3), Autosomal dominant; Metaphyseal dysplasia, Spahr type, 250400 (3), Autosomal recessive; Spondyloepimetaphyseal dysplasia, Missouri type, 602111 (3), Autosomal dominant
MMP14	600754	?Winchester syndrome, 277950 (3)
MMP19	601807	Cavitary optic disc anomalies, 611543 (3), Autosomal dominant
MMP20	604629	Amelogenesis imperfecta, type IIA2, 612529 (3), Autosomal recessive
MMP21	608416	Heterotaxy, visceral, 7, autosomal, 616749 (3), Autosomal recessive
MMP3	185250	{Coronary heart disease, susceptibility to, 6}, 614466 (3)
MMP9	120361	Metaphyseal anadysplasia 2, 613073 (3)
MN1	156100	Meningioma, 607174 (3), Autosomal dominant
MNX1	142994	Currarino syndrome, 176450 (3), Autosomal dominant
MOCOS	613274	Xanthinuria, type II, 603592 (3), Autosomal recessive
MOCS2	603708	Molybdenum cofactor deficiency B, 252160 (3), Autosomal recessive
MOG	159465	?Narcolepsy 7, 614250 (3), Autosomal dominant
MOGS	601336	Congenital disorder of glycosylation, type IIb, 606056 (3), Autosomal recessive
MORC2	616661	Charcot-Marie-Tooth disease, axonal, type 2Z, 616688 (3), Autosomal dominant
MPDU1	604041	Congenital disorder of glycosylation, type If, 609180 (3), Autosomal recessive
MPDZ	603785	Hydrocephalus, nonsyndromic, autosomal recessive 2, 615219 (3), Autosomal recessive
MPI	154550	Congenital disorder of glycosylation, type Ib, 602579 (3), Autosomal recessive
MPL	159530	Myelofibrosis with myeloid metaplasia, somatic, 254450 (3); Thrombocythemia 2, 601977 (3), Autosomal dominant, Somatic mutation; Thrombocytopenia, congenital amegakaryocytic, 604498 (3), Autosomal recessive
MPLKIP	609188	Trichothiodystrophy 4, nonphotosensitive, 234050 (3), Autosomal recessive
MPO	606989	{Alzheimer disease, susceptibility to}, 104300 (3), Autosomal dominant; {Lung cancer, protection against, in smokers} (3); Myeloperoxidase deficiency, 254600 (3), Autosomal recessive

MPV17	137960	Mitochondrial DNA depletion syndrome 6 (hepatocerebral type), 256810 (3), Autosomal recessive
MPZ	159440	Charcot-Marie-Tooth disease, dominant intermediate D, 607791 (3), Autosomal dominant; Charcot-Marie-Tooth disease, type 1B, 118200 (3), Autosomal dominant; Charcot-Marie-Tooth disease, type 2I, 607677 (3), Autosomal dominant; Charcot-Marie-Tooth disease, type 2J, 607736 (3), Autosomal dominant; Dejerine-Sottas disease, 145900 (3), Autosomal recessive, Autosomal dominant; Neuropathy, congenital hypomyelinating, 605253 (3), Autosomal recessive, Autosomal dominant; Roussy-Levy syndrome, 180800 (3), Autosomal dominant
MRAP	609196	Glucocorticoid deficiency 2, 607398 (3), Autosomal recessive
MRAP2	615410	{?Obesity, susceptibility to, BMIQ18}, 615457 (3), Autosomal dominant
MRE11	600814	Ataxia-telangiectasia-like disorder 1, 604391 (3), Autosomal recessive
MRPL3	607118	Combined oxidative phosphorylation deficiency 9, 614582 (3), Autosomal recessive
MRPL44	611849	?Combined oxidative phosphorylation deficiency 16, 615395 (3), Autosomal recessive
MRPS16	609204	Combined oxidative phosphorylation deficiency 2, 610498 (3), Autosomal recessive
MRPS22	605810	Combined oxidative phosphorylation deficiency 5, 611719 (3), Autosomal recessive
MS4A1	112210	Immunodeficiency, common variable, 5, 613495 (3), Autosomal recessive
MS4A2	147138	{Atopy, susceptibility to}, 147050 (3), Autosomal dominant
MSH2	609309	Colorectal cancer, hereditary nonpolyposis, type 1, 120435 (3), Autosomal dominant; Mismatch repair cancer syndrome, 276300 (3), Autosomal recessive; Muir-Torre syndrome, 158320 (3), Autosomal dominant
MSH3	600887	Endometrial carcinoma, somatic, 608089 (3); Familial adenomatous polyposis 4, 617100 (3), Autosomal recessive
MSH6	600678	Colorectal cancer, hereditary nonpolyposis, type 5, 614350 (3), Autosomal dominant; Endometrial cancer, familial, 608089 (3); Mismatch repair cancer syndrome, 276300 (3), Autosomal recessive
MSMB	157145	{Prostate cancer, hereditary, 13}, 611928 (3)
MSMO1	607545	Microcephaly, congenital cataract, and psoriasiform dermatitis, 616834 (3), Autosomal recessive
MSN	309845	Immunodeficiency 50, 300988 (3), X-linked recessive
MSR1	153622	Barrett esophagus/esophageal adenocarcinoma, 614266 (3)
MSRB3	613719	Deafness, autosomal recessive 74, 613718 (3), Autosomal recessive
MSTN	601788	Muscle hypertrophy, 614160 (3)
MSX1	142983	Ectodermal dysplasia 3, Witkop type, 189500 (3), Autosomal dominant; Orofacial cleft 5, 608874 (3); Tooth agenesis, selective, 1, with or without orofacial cleft, 106600 (3), Autosomal dominant

MSX2	123101	Craniosynostosis 2, 604757 (3), Autosomal dominant; Parietal foramina 1, 168500 (3), Autosomal dominant; Parietal foramina with cleidocranial dysplasia, 168550 (3), Autosomal dominant
MTFMT	611766	Combined oxidative phosphorylation deficiency 15, 614947 (3), Autosomal recessive
MTHFD1	172460	Combined immunodeficiency and megaloblastic anemia with or without hyperhomocysteinemia, 617780 (3), Autosomal recessive; {Neural tube defects, folate-sensitive, susceptibility to}, 601634 (3), Autosomal recessive
MTHFR	607093	Homocystinuria due to MTHFR deficiency, 236250 (3), Autosomal recessive; {Neural tube defects, susceptibility to}, 601634 (3), Autosomal recessive; {Schizophrenia, susceptibility to}, 181500 (3), Autosomal dominant; {Thromboembolism, susceptibility to}, 188050 (3), Autosomal dominant; {Vascular disease, susceptibility to} (3)
MTM1	300415	Myotubular myopathy, X-linked, 310400 (3), X-linked recessive
MTMR14	611089	{Centronuclear myopathy, autosomal, modifier of}, 160150 (3), Autosomal dominant
MTNR1B	600804	{Diabetes mellitus, type 2, susceptibility to}, 125853 (3), Autosomal dominant
MTO1	614667	Combined oxidative phosphorylation deficiency 10, 614702 (3), Autosomal recessive
MTOR	601231	Focal cortical dysplasia, type II, somatic, 607341 (3); Smith-Kingsmore syndrome, 616638 (3), Autosomal dominant
MTPAP	613669	?Spastic ataxia 4, autosomal recessive, 613672 (3), Autosomal recessive
MTR	156570	Homocystinuria-megaloblastic anemia, cblG complementation type, 250940 (3), Autosomal recessive; {Neural tube defects, folate-sensitive, susceptibility to}, 601634 (3), Autosomal recessive
MTRR	602568	Homocystinuria-megaloblastic anemia, cbl E type, 236270 (3), Autosomal recessive; {Neural tube defects, folate-sensitive, susceptibility to}, 601634 (3), Autosomal recessive
MTTP	157147	Abetalipoproteinemia, 200100 (3), Autosomal recessive; {Metabolic syndrome, protection against}, 605552 (3), Autosomal dominant
MUC1	158340	Medullary cystic kidney disease 1, 174000 (3), Autosomal dominant
MUC5B	600770	{Pulmonary fibrosis, idiopathic, susceptibility to}, 178500 (3), Autosomal dominant
MUC7	158375	{Asthma, protection against}, 600807 (3), Autosomal dominant
MUSK	601296	Fetal akinesia deformation sequence, 208150 (3), Autosomal recessive; Myasthenic syndrome, congenital, 9, associated with acetylcholine receptor deficiency, 616325 (3), Autosomal recessive
MUT	609058	Methylmalonic aciduria, mut(0) type, 251000 (3), Autosomal recessive
MVD	603236	Porokeratosis 7, multiple types, 614714 (3), Autosomal dominant
MVK	251170	Hyper-IgD syndrome, 260920 (3), Autosomal recessive; Mevalonic aciduria, 610377 (3), Autosomal recessive; Porokeratosis 3, multiple types, 175900 (3), Autosomal dominant

MXI1	600020	Neurofibrosarcoma (3); {Prostate cancer, susceptibility to}, 176807 (3), Autosomal dominant
MYBPC3	600958	Cardiomyopathy, dilated, 1MM, 615396 (3), Autosomal dominant; Cardiomyopathy, hypertrophic, 4, 115197 (3), Autosomal dominant; Left ventricular noncompaction 10, 615396 (3), Autosomal dominant
MYC	190080	Burkitt lymphoma, 113970 (3), Isolated cases
MYCN	164840	Feingold syndrome 1, 164280 (3), Autosomal dominant
MYD88	602170	Macroglobulinemia, Waldenstrom, somatic, 153600 (3); Pyogenic bacterial infections, recurrent, due to MYD88 deficiency, 612260 (3)
MYF6	159991	Centronuclear myopathy 3, 614408 (3), Autosomal dominant
MYH11	160745	Aortic aneurysm, familial thoracic 4, 132900 (3), Autosomal dominant
MYH2	160740	Proximal myopathy and ophthalmoplegia, 605637 (3), Autosomal recessive, Autosomal dominant
MYH3	160720	Arthrogryposis, distal, type 2A, 193700 (3), Autosomal dominant; Arthrogryposis, distal, type 2B, 601680 (3), Autosomal dominant; Arthrogryposis, distal, type 8, 178110 (3), Autosomal dominant
MYH6	160710	Atrial septal defect 3, 614089 (3); Cardiomyopathy, dilated, 1EE, 613252 (3); Cardiomyopathy, hypertrophic, 14, 613251 (3), Autosomal dominant; {Sick sinus syndrome 3}, 614090 (3)
MYH7	160760	Cardiomyopathy, dilated, 1S, 613426 (3), Autosomal dominant; Cardiomyopathy, hypertrophic, 1, 192600 (3), Autosomal dominant; Laing distal myopathy, 160500 (3), Autosomal dominant; Left ventricular noncompaction 5, 613426 (3), Autosomal dominant; Myopathy, myosin storage, autosomal dominant, 608358 (3), Autosomal dominant; Myopathy, myosin storage, autosomal recessive, 255160 (3), Autosomal recessive; Scapuloperoneal syndrome, myopathic type, 181430 (3), Autosomal dominant
MYH8	160741	Carney complex variant, 608837 (3); Trismus-pseudocamptodactyly syndrome, 158300 (3), Autosomal dominant
MYH9	160775	Deafness, autosomal dominant 17, 603622 (3), Autosomal dominant; Macrothrombocytopenia and granulocyte inclusions with or without nephritis or sensorineural hearing loss, 155100 (3), Autosomal dominant
MYL2	160781	Cardiomyopathy, hypertrophic, 10, 608758 (3), Autosomal dominant
MYL3	160790	Cardiomyopathy, hypertrophic, 8, 608751 (3), Autosomal dominant
MYL4	160770	?Atrial fibrillation, familial, 18, 617280 (3), Autosomal dominant
MYLK	600922	Aortic aneurysm, familial thoracic 7, 613780 (3), Autosomal dominant
MYLK2	606566	Cardiomyopathy, hypertrophic, 1, digenic, 192600 (3), Autosomal dominant
MYO18B	607295	Klippel-Feil syndrome 4, autosomal recessive, with myopathy and facial dysmorphism, 616549 (3), Autosomal recessive
MYO1A	601478	No OMIM phenotype
MYO1E	601479	Glomerulosclerosis, focal segmental, 6, 614131 (3), Autosomal recessive
MYO3A	606808	Deafness, autosomal recessive 30, 607101 (3), Autosomal recessive

MYO5B	606540	Microvillus inclusion disease, 251850 (3), Autosomal recessive
MYO6	600970	Deafness, autosomal dominant 22, 606346 (3), Autosomal dominant; Deafness, autosomal dominant 22, with hypertrophic cardiomyopathy, 606346 (3), Autosomal dominant; Deafness, autosomal recessive 37, 607821 (3), Autosomal recessive
MYO9B	602129	{Celiac disease, susceptibility to, 4}, 609753 (3)
MYOC	601652	Glaucoma 1A, primary open angle, 137750 (3), Autosomal dominant
MYOT	604103	Muscular dystrophy, limb-girdle, type 1A, 159000 (3), Autosomal dominant; Myopathy, myofibrillar, 3, 609200 (3), Autosomal dominant; Myopathy, spheroid body, 182920 (3), Autosomal dominant
MYOZ2	605602	Cardiomyopathy, hypertrophic, 16, 613838 (3), Autosomal dominant
MYT1L	613084	Mental retardation, autosomal dominant 39, 616521 (3), Autosomal dominant
NAA10	300013	?Microphthalmia, syndromic 1, 309800 (3), X-linked; Ogden syndrome, 300855 (3), X-linked recessive, X-linked dominant
NACC1	610672	Neurodevelopmental disorder with epilepsy, cataracts, feeding difficulties, and delayed brain myelination, 617393 (3), Autosomal dominant
NADK2	615787	?2,4-dienoyl-CoA reductase deficiency, 616034 (3), Autosomal recessive
NAGA	104170	Kanzaki disease, 609242 (3), Autosomal recessive; Schindler disease, type I, 609241 (3), Autosomal recessive; Schindler disease, type III, 609241 (3), Autosomal recessive
NAGLU	609701	?Charcot-Marie-Tooth disease, axonal, type 2V, 616491 (3), Autosomal dominant; Mucopolysaccharidosis type IIIB (Sanfilippo B), 252920 (3), Autosomal recessive
NAGS	608300	N-acetylglutamate synthase deficiency, 237310 (3), Autosomal recessive
NALCN	611549	Congenital contractures of the limbs and face, hypotonia, and developmental delay, 616266 (3), Autosomal dominant; Hypotonia, infantile, with psychomotor retardation and characteristic facies 1, 615419 (3), Autosomal recessive
NANOS1	608226	Spermatogenic failure 12, 615413 (3), Autosomal dominant
NANS	605202	Spondyloepimetaphyseal dysplasia, Camera-Genevieve type, 610442 (3), Autosomal recessive
NARS2	612803	Combined oxidative phosphorylation deficiency 24, 616239 (3), Autosomal recessive
NAT2	612182	[Acetylation, slow], 243400 (3), Autosomal recessive
NAT8L	610647	?N-acetylaspartate deficiency, 614063 (3), Autosomal recessive
NAXE	608862	Encephalopathy, progressive, early-onset, with brain edema and/or leukoencephalopathy, 617186 (3), Autosomal recessive
NBAS	608025	Infantile liver failure syndrome 2, 616483 (3), Autosomal recessive; Short stature, optic nerve atrophy, and Pelger-Huet anomaly, 614800 (3), Autosomal recessive
NBEAL2	614169	Gray platelet syndrome, 139090 (3), Autosomal recessive

NBN	602667	Aplastic anemia, 609135 (3); Leukemia, acute lymphoblastic, 613065 (3); Nijmegen breakage syndrome, 251260 (3), Autosomal recessive
NCF1	608512	Chronic granulomatous disease due to deficiency of NCF-1, 233700 (3), Autosomal recessive
NCF2	608515	Chronic granulomatous disease due to deficiency of NCF-2, 233710 (3), Autosomal recessive
NCF4	601488	?Granulomatous disease, chronic, autosomal recessive, cytochrome b-positive, type III, 613960 (3), Autosomal recessive
NCOA4	601984	No OMIM phenotype
NCR3	611550	{Malaria, mild, susceptibility to}, 609148 (3)
NCSTN	605254	Acne inversa, familial, 1, 142690 (3), Autosomal dominant
NDN	602117	Prader-Willi syndrome, 176270 (3), Isolated cases
NDP	300658	Exudative vitreoretinopathy 2, X-linked, 305390 (3); Norrie disease, 310600 (3), X-linked recessive
NDRG1	605262	Charcot-Marie-Tooth disease, type 4D, 601455 (3), Autosomal recessive
NDST1	600853	Mental retardation, autosomal recessive 46, 616116 (3), Autosomal recessive
NDUFA1	300078	Mitochondrial complex I deficiency, 252010 (3), Autosomal recessive, X-linked dominant, Mitochondrial
NDUFA10	603835	Leigh syndrome, 256000 (3), Autosomal recessive, Mitochondrial
NDUFA11	612638	Mitochondrial complex I deficiency, 252010 (3), Autosomal recessive, X-linked dominant, Mitochondrial
NDUFA12	614530	Leigh syndrome due to mitochondrial complex 1 deficiency, 256000 (3), Autosomal recessive, Mitochondrial
NDUFA13	609435	{Thyroid carcinoma, Hurthle cell}, 607464 (3)
NDUFA2	602137	Leigh syndrome due to mitochondrial complex I deficiency, 256000 (3), Autosomal recessive, Mitochondrial
NDUFA9	603834	Leigh syndrome due to mitochondrial complex I deficiency, 256000 (3), Autosomal recessive, Mitochondrial
NDUFAF1	606934	Mitochondrial complex I deficiency, 252010 (3), Autosomal recessive, X-linked dominant, Mitochondrial
NDUFAF2	609653	Mitochondrial complex I deficiency, 252010 (3), Autosomal recessive, X-linked dominant, Mitochondrial
NDUFAF3	612911	Mitochondrial complex I deficiency, 252010 (3), Autosomal recessive, X-linked dominant, Mitochondrial
NDUFAF4	611776	Mitochondrial complex I deficiency, 252010 (3), Autosomal recessive, X-linked dominant, Mitochondrial
NDUFAF5	612360	Mitochondrial complex 1 deficiency, 252010 (3), Autosomal recessive, X-linked dominant, Mitochondrial
NDUFB11	300403	Linear skin defects with multiple congenital anomalies 3, 300952 (3), X-linked dominant; ?Mitochondrial complex I deficiency, 252010 (3), Autosomal recessive, X-linked dominant, Mitochondrial

NDUFB3	603839	Mitochondrial complex I deficiency, 252010 (3), Autosomal recessive, X-linked dominant, Mitochondrial
NDUFB9	601445	?Mitochondrial complex I deficiency, 252010 (3), Autosomal recessive, X-linked dominant, Mitochondrial
NDUFS1	157655	Mitochondrial complex I deficiency, 252010 (3), Autosomal recessive, X-linked dominant, Mitochondrial
NDUFS2	602985	Mitochondrial complex I deficiency, 252010 (3), Autosomal recessive, X-linked dominant, Mitochondrial
NDUFS3	603846	Leigh syndrome due to mitochondrial complex I deficiency, 256000 (3), Autosomal recessive, Mitochondrial; Mitochondrial complex I deficiency, 252010 (3), Autosomal recessive, X-linked dominant, Mitochondrial
NDUFS4	602694	Leigh syndrome, 256000 (3), Autosomal recessive, Mitochondrial; Mitochondrial complex I deficiency, 252010 (3), Autosomal recessive, X-linked dominant, Mitochondrial
NDUFS6	603848	Mitochondrial complex I deficiency, 252010 (3), Autosomal recessive, X-linked dominant, Mitochondrial
NDUFS7	601825	Leigh syndrome, 256000 (3), Autosomal recessive, Mitochondrial
NDUFS8	602141	Leigh syndrome due to mitochondrial complex I deficiency, 256000 (3), Autosomal recessive, Mitochondrial
NDUFV1	161015	Mitochondrial complex I deficiency, 252010 (3), Autosomal recessive, X-linked dominant, Mitochondrial
NDUFV2	600532	Mitochondrial complex I deficiency, 252010 (3), Autosomal recessive, X-linked dominant, Mitochondrial
NEB	161650	Nemaline myopathy 2, autosomal recessive, 256030 (3), Autosomal recessive
NECAP1	611623	?Epileptic encephalopathy, early infantile, 21, 615833 (3), Autosomal recessive
NECTIN1	600644	Cleft lip/palate-ectodermal dysplasia syndrome, 225060 (3), Autosomal recessive; Orofacial cleft 7, 225060 (3), Autosomal recessive
NECTIN4	609607	Ectodermal dysplasia-syndactyly syndrome 1, 613573 (3), Autosomal recessive
NEFH	162230	?{Amyotrophic lateral sclerosis, susceptibility to}, 105400 (3), Autosomal recessive, Autosomal dominant; Charcot-Marie-Tooth disease, axonal, type 2CC, 616924 (3), Autosomal dominant
NEFL	162280	Charcot-Marie-Tooth disease, dominant intermediate G, 617882 (3), Autosomal dominant; Charcot-Marie-Tooth disease, type 1F, 607734 (3), Autosomal recessive, Autosomal dominant; Charcot-Marie-Tooth disease, type 2E, 607684 (3), Autosomal dominant
NEK1	604588	{Amyotrophic lateral sclerosis, susceptibility to, 24}, 617892 (3); Short-rib thoracic dysplasia 6 with or without polydactyly, 263520 (3), Autosomal recessive, Digenic recessive
NEK2	604043	?Retinitis pigmentosa 67, 615565 (3), Autosomal recessive

NEK8	609799	?Nephronophthisis 9, 613824 (3); Renal-hepatic-pancreatic dysplasia 2, 615415 (3), Autosomal recessive
NEK9	609798	?Arthrogryposis, Perthes disease, and upward gaze palsy, 614262 (3), Autosomal recessive; Lethal congenital contracture syndrome 10, 617022 (3), Autosomal recessive; Nevus comedonicus, somatic, 617025 (3)
NEU1	608272	Sialidosis, type I, 256550 (3), Autosomal recessive; Sialidosis, type II, 256550 (3), Autosomal recessive
NEUROD1	601724	{Diabetes mellitus, noninsulin-dependent}, 125853 (3), Autosomal dominant; Maturity-onset diabetes of the young 6, 606394 (3)
NEUROG3	604882	Diarrhea 4, malabsorptive, congenital, 610370 (3), Autosomal recessive
NEXMIF	300524	Mental retardation, X-linked 98, 300912 (3), X-linked dominant
NEXN	613121	Cardiomyopathy, dilated, 1CC, 613122 (3), Autosomal dominant; Cardiomyopathy, hypertrophic, 20, 613876 (3), Autosomal dominant
NF1	613113	Leukemia, juvenile myelomonocytic, 607785 (3), Autosomal dominant, Somatic mutation; Neurofibromatosis, familial spinal, 162210 (3), Autosomal dominant; Neurofibromatosis, type 1, 162200 (3), Autosomal dominant; Neurofibromatosis-Noonan syndrome, 601321 (3), Autosomal dominant; Watson syndrome, 193520 (3), Autosomal dominant
NF2	607379	Meningioma, NF2-related, somatic, 607174 (3); Neurofibromatosis, type 2, 101000 (3), Autosomal dominant; Schwannomatosis, somatic, 162091 (3)
NFKB1	164011	Immunodeficiency, common variable, 12, 616576 (3), Autosomal dominant
NFKB2	164012	Immunodeficiency, common variable, 10, 615577 (3), Autosomal dominant
NFKBIA	164008	Ectodermal dysplasia, anhidrotic, with T-cell immunodeficiency, 612132 (3), Autosomal dominant
NFU1	608100	Multiple mitochondrial dysfunctions syndrome 1, 605711 (3), Autosomal recessive
NGF	162030	Neuropathy, hereditary sensory and autonomic, type V, 608654 (3), Autosomal recessive
NGLY1	610661	Congenital disorder of deglycosylation, 615273 (3), Autosomal recessive
NHEJ1	611290	Severe combined immunodeficiency with microcephaly, growth retardation, and sensitivity to ionizing radiation, 611291 (3)
NHLRC1	608072	Epilepsy, progressive myoclonic 2B (Lafora), 254780 (3), Autosomal recessive
NHP2	606470	Dyskeratosis congenita, autosomal recessive 2, 613987 (3), Autosomal recessive
NIN	608684	?Seckel syndrome 7, 614851 (3), Autosomal recessive
NIPA1	608145	Spastic paraplegia 6, autosomal dominant, 600363 (3), Autosomal dominant

NIPAL4	609383	Ichthyosis, congenital, autosomal recessive 6, 612281 (3), Autosomal recessive
NIPBL	608667	Cornelia de Lange syndrome 1, 122470 (3), Autosomal dominant
NKX2-1	600635	Chorea, hereditary benign, 118700 (3), Autosomal dominant; Choreoathetosis, hypothyroidism, and neonatal respiratory distress, 610978 (3), Autosomal dominant; {Thyroid cancer, nonmedullary, 1}, 188550 (3), Autosomal dominant
NKX2-5	600584	Atrial septal defect 7, with or without AV conduction defects, 108900 (3), Autosomal dominant; Conotruncal heart malformations, variable, 217095 (3); Hypoplastic left heart syndrome 2, 614435 (3), Autosomal dominant; Hypothyroidism, congenital nongoitrous, 5, 225250 (3), Autosomal dominant; Tetralogy of Fallot, 187500 (3), Autosomal dominant; Ventricular septal defect 3, 614432 (3), Autosomal dominant
NKX2-6	611770	Conotruncal heart malformations, 217095 (3); Persistent truncus arteriosus, 217095 (3)
NKX3-2	602183	Spondylo-megaepiphyseal-metaphyseal dysplasia, 613330 (3), Autosomal recessive
NLGN3	300336	{Asperger syndrome susceptibility, X-linked 1}, 300494 (3), Isolated cases, X-linked, Multifactorial; {Autism susceptibility, X-linked 1}, 300425 (3), Isolated cases, X-linked, Multifactorial
NLGN4X	300427	{Asperger syndrome susceptibility, X-linked 2}, 300497 (3), Isolated cases, X-linked, Multifactorial; {Autism susceptibility, X-linked 2}, 300495 (3), Isolated cases, X-linked, Multifactorial; Mental retardation, X-linked, 300495 (3), Isolated cases, X-linked, Multifactorial
NLRC4	606831	Autoinflammation with infantile enterocolitis, 616050 (3), Autosomal dominant; ?Familial cold autoinflammatory syndrome 4, 616115 (3), Autosomal dominant
NLRP1	606636	Autoinflammation with arthritis and dyskeratosis, 617388 (3), Autosomal recessive, Autosomal dominant; Palmoplantar carcinoma, multiple self-healing, 615225 (3), Autosomal dominant; {Vitiligo-associated multiple autoimmune disease susceptibility 1}, 606579 (3)
NLRP12	609648	Familial cold autoinflammatory syndrome 2, 611762 (3), Autosomal dominant
NLRP3	606416	CINCA syndrome, 607115 (3), Autosomal dominant; Deafness, autosomal dominant 34, with or without inflammation, 617772 (3), Autosomal dominant; Familial cold inflammatory syndrome 1, 120100 (3), Autosomal dominant; Keratoendothelitis fugax hereditaria, 148200 (3), Autosomal dominant; Muckle-Wells syndrome, 191900 (3), Autosomal dominant
NME1	156490	Neuroblastoma, 256700 (3), Autosomal dominant, Isolated cases
NME8	607421	Ciliary dyskinesia, primary, 6, 610852 (3), Autosomal recessive
NMNAT1	608700	Leber congenital amaurosis 9, 608553 (3), Autosomal recessive
NNT	607878	Glucocorticoid deficiency 4, with or without mineralocorticoid deficiency, 614736 (3), Autosomal recessive

NOBOX	610934	Premature ovarian failure 5, 611548 (3), Autosomal dominant
NOD2	605956	Blau syndrome, 186580 (3), Autosomal dominant; {Inflammatory bowel disease 1, Crohn disease}, 266600 (3), Multifactorial; {Psoriatic arthritis, susceptibility to}, 607507 (2); {Yao syndrome}, 617321 (3), Multifactorial
NODAL	601265	Heterotaxy, visceral, 5, 270100 (3), Autosomal dominant
NOG	602991	Brachydactyly, type B2, 611377 (3), Autosomal dominant; Multiple synostoses syndrome 1, 186500 (3), Autosomal dominant; Stapes ankylosis with broad thumbs and toes, 184460 (3), Autosomal dominant; Symphalangism, proximal, 1A, 185800 (3), Autosomal dominant; Tarsal-carpal coalition syndrome, 186570 (3), Autosomal dominant
NOL3	605235	Myoclonus, familial cortical, 614937 (3), Autosomal dominant
NONO	300084	Mental retardation, X-linked, syndromic 34, 300967 (3), X-linked
NOP10	606471	Dyskeratosis congenita, autosomal recessive 1, 224230 (3), Autosomal recessive
NOP56	614154	Spinocerebellar ataxia 36, 614153 (3), Autosomal dominant
NOS2	163730	{Hypertension, susceptibility to}, 145500 (2), Multifactorial; {Malaria, resistance to}, 611162 (3)
NOS3	163729	{Alzheimer disease, late-onset, susceptibility to}, 104300 (3), Autosomal dominant; {Coronary artery spasm 1, susceptibility to} (3); {Hypertension, pregnancy-induced}, 189800 (3), Autosomal dominant; {Hypertension, susceptibility to}, 145500 (3), Multifactorial; {Ischemic stroke, susceptibility to}, 601367 (3), Multifactorial; {Placental abruption} (3)
NOTCH1	190198	Adams-Oliver syndrome 5, 616028 (3), Autosomal dominant; Aortic valve disease 1, 109730 (3), Autosomal dominant
NOTCH2	600275	Alagille syndrome 2, 610205 (3), Autosomal dominant; Hajdu-Cheney syndrome, 102500 (3), Autosomal dominant
NOTCH3	600276	Cerebral arteriopathy with subcortical infarcts and leukoencephalopathy 1, 125310 (3), Autosomal dominant; Lateral meningocele syndrome, 130720 (3), Autosomal dominant; ?Myofibromatosis, infantile 2, 615293 (3), Autosomal dominant
NPAP1	610922	No OMIM phenotype
NPC1	607623	Niemann-Pick disease, type C1, 257220 (3), Autosomal recessive; Niemann-Pick disease, type D, 257220 (3), Autosomal recessive
NPC2	601015	Niemann-pick disease, type C2, 607625 (3), Autosomal recessive
NPHP1	607100	Joubert syndrome 4, 609583 (3), Autosomal recessive; Nephronophthisis 1, juvenile, 256100 (3), Autosomal recessive; Senior-Loken syndrome-1, 266900 (3), Autosomal recessive
NPHP3	608002	Meckel syndrome 7, 267010 (3), Autosomal recessive; Nephronophthisis 3, 604387 (3), Autosomal recessive; Renal-hepatic-pancreatic dysplasia 1, 208540 (3), Autosomal recessive
NPHP4	607215	Nephronophthisis 4, 606966 (3), Autosomal recessive; Senior-Loken syndrome 4, 606996 (3), Autosomal recessive

NPHS1	602716	Nephrotic syndrome, type 1, 256300 (3), Autosomal recessive
NPHS2	604766	Nephrotic syndrome, type 2, 600995 (3), Autosomal recessive
NPPA	108780	Atrial fibrillation, familial, 6, 612201 (3), Autosomal dominant; Atrial standstill 2, 615745 (3), Autosomal recessive
NPR2	108961	Acromesomelic dysplasia, Maroteaux type, 602875 (3), Autosomal recessive; Epiphyseal chondrodysplasia, Miura type, 615923 (3), Autosomal dominant; Short stature with nonspecific skeletal abnormalities, 616255 (3), Autosomal dominant
NPRL2	607072	Epilepsy, familial focal, with variable foci 2, 617116 (3), Autosomal dominant
NPRL3	600928	Epilepsy, familial focal, with variable foci 3, 617118 (3), Autosomal dominant
NPSR1	608595	{Asthma, susceptibility to, 2}, 608584 (3)
NQO2	160998	{?Breast cancer susceptibility}, 114480 (1), Autosomal dominant
NR0B1	300473	Adrenal hypoplasia, congenital, 300200 (3), X-linked recessive; 46XY sex reversal 2, dosage-sensitive, 300018 (3), X-linked
NR0B2	604630	Obesity, mild, early-onset, 601665 (3), Autosomal recessive, Autosomal dominant, Multifactorial
NR2E3	604485	Enhanced S-cone syndrome, 268100 (3), Autosomal recessive; Retinitis pigmentosa 37, 611131 (3), Autosomal recessive, Autosomal dominant
NR2F1	132890	Bosch-Boonstra-Schaaf optic atrophy syndrome, 615722 (3), Autosomal dominant
NR2F2	107773	Congenital heart defects, multiple types, 4, 615779 (3), Autosomal dominant
NR3C2	600983	Hypertension, early-onset, autosomal dominant, with exacerbation in pregnancy, 605115 (3); Pseudohypoaldosteronism type I, autosomal dominant, 177735 (3), Autosomal dominant
NR4A2	601828	No OMIM phenotype
NR4A3	600542	Chondrosarcoma, extraskeletal myxoid, 612237 (3)
NR5A1	184757	Adrenocortical insufficiency, 612964 (3), Autosomal dominant; Premature ovarian failure 7, 612964 (3), Autosomal dominant; Spermatogenic failure 8, 613957 (3), Autosomal dominant; 46, XX sex reversal 4, 617480 (3), Autosomal dominant; 46XY sex reversal 3, 612965 (3), Autosomal dominant
NRAS	164790	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), Autosomal dominant; ?RAS-associated autoimmune lymphoproliferative syndrome type IV, somatic, 614470 (3); Schimmelpenning-Feuerstein-Mims syndrome, somatic mosaic, 163200 (3); Thyroid carcinoma, follicular, somatic, 188470 (3)
NRG1	142445	{?Schizophrenia, susceptibility to}, 603013 (1)
NRL	162080	Retinal degeneration, autosomal recessive, clumped pigment type (3); Retinitis pigmentosa 27, 613750 (3), Autosomal dominant

NRXN1	600565	Pitt-Hopkins-like syndrome 2, 614325 (3), Autosomal recessive; {Schizophrenia, susceptibility to, 17}, 614332 (3)
NSD1	606681	Leukemia, acute myeloid, 601626 (1), Autosomal dominant; Sotos syndrome 1, 117550 (3), Autosomal dominant
NSD2	602952	No OMIM phenotype
NSMCE3	608243	Lung disease, immunodeficiency, and chromosome breakage syndrome, 617241 (3), Autosomal recessive
NSMF	608137	Hypogonadotropic hypogonadism 9 with or without anosmia, 614838 (3), Autosomal dominant
NSUN2	610916	Mental retardation, autosomal recessive 5, 611091 (3), Autosomal recessive
NT5C3A	606224	Anemia, hemolytic, due to UMPH1 deficiency, 266120 (3), Autosomal recessive
NT5E	129190	Calcification of joints and arteries, 211800 (3), Autosomal recessive
NTF4	162662	Glaucoma 1, open angle, 1O, 613100 (3)
NTHL1	602656	Familial adenomatous polyposis 3, 616415 (3), Autosomal recessive
NTRK1	191315	Insensitivity to pain, congenital, with anhidrosis, 256800 (3), Autosomal recessive; Medullary thyroid carcinoma, familial, 155240 (3), Autosomal dominant
NTRK2	600456	Epileptic encephalopathy, early infantile, 58, 617830 (3), Autosomal dominant; Obesity, hyperphagia, and developmental delay, 613886 (3), Autosomal dominant
NUBPL	613621	Mitochondrial complex I deficiency, 252010 (3), Autosomal recessive, X-linked dominant, Mitochondrial
NUDT15	615792	{Thiopurines, poor metabolism of, 2}, 616903 (3), Autosomal dominant
NUMA1	164009	Leukemia, acute promyelocytic, somatic, 612376 (3)
NUP107	607617	Nephrotic syndrome, type 11, 616730 (3), Autosomal recessive
NUP155	606694	?Atrial fibrillation 15, 615770 (3), Autosomal recessive
NUP205	614352	?Nephrotic syndrome, type 13, 616893 (3)
NUP214	114350	Leukemia, T-cell acute lymphoblastic, somatic, 613065 (3); Leukemia, acute myeloid, somatic, 601626 (3)
NUP62	605815	Striatonigral degeneration, infantile, 271930 (3), Autosomal recessive
NUP93	614351	Nephrotic syndrome, type 12, 616892 (3), Autosomal recessive
NUS1	610463	?Congenital disorder of glycosylation, type 1aa, 617082 (3), Autosomal recessive; Mental retardation, autosomal dominant 55, with seizures, 617831 (3), Autosomal dominant
NYX	300278	Night blindness, congenital stationary (complete), 1A, X-linked, 310500 (3), X-linked recessive
OAS1	164350	No OMIM phenotype
OAT	613349	Gyrate atrophy of choroid and retina with or without ornithinemia, 258870 (3), Autosomal recessive
OBSL1	610991	3-M syndrome 2, 612921 (3)

OCA2	611409	Albinism, brown oculocutaneous, 203200 (3), Autosomal recessive; Albinism, oculocutaneous, type II, 203200 (3), Autosomal recessive; [Skin/hair/eye pigmentation 1, blond/brown hair], 227220 (3), Autosomal recessive; [Skin/hair/eye pigmentation 1, blue/nonblue eyes], 227220 (3), Autosomal recessive
OCLN	602876	Pseudo-TORCH syndrome 1, 251290 (3), Autosomal recessive
OCRL	300535	Dent disease 2, 300555 (3), X-linked recessive; Lowe syndrome, 309000 (3), X-linked recessive
ODAPH (C4orf26)	614829	Amelogenesis imperfecta, type IIA4, 614832 (3), Autosomal recessive
OFD1	300170	Joubert syndrome 10, 300804 (3), X-linked recessive; Orofaciodigital syndrome I, 311200 (3), X-linked dominant; ?Retinitis pigmentosa 23, 300424 (3), X-linked recessive; Simpson-Golabi-Behmel syndrome, type 2, 300209 (3), X-linked recessive
OGDH	613022	Alpha-ketoglutarate dehydrogenase deficiency, 203740 (1), Autosomal recessive
OGG1	601982	Renal cell carcinoma, clear cell, somatic, 144700 (3)
OLR1	602601	{Myocardial infarction, susceptibility to}, 608446 (3)
OPA1	605290	Behr syndrome, 210000 (3), Autosomal recessive; {Glaucoma, normal tension, susceptibility to}, 606657 (3); ?Mitochondrial DNA depletion syndrome 14 (encephalocardiomyopathic type), 616896 (3); Optic atrophy 1, 165500 (3), Autosomal dominant; Optic atrophy plus syndrome, 125250 (3), Autosomal dominant
OPA3	606580	3-methylglutaconic aciduria, type III, 258501 (3), Autosomal recessive; Optic atrophy 3 with cataract, 165300 (3), Autosomal dominant
OPCML	600632	{Ovarian cancer, somatic}, 167000 (3)
OPHN1	300127	Mental retardation, X-linked, with cerebellar hypoplasia and distinctive facial appearance, 300486 (3), X-linked recessive
OPLAH	614243	5-oxoprolinase deficiency, 260005 (3), Autosomal recessive, Autosomal dominant
OPN1LW	300822	Blue cone monochromacy, 303700 (3), X-linked recessive; Colorblindness, protan, 303900 (3), X-linked
OPN1MW	300821	Blue cone monochromacy, 303700 (3), X-linked recessive; Colorblindness, deutan, 303800 (3), X-linked
OPN1SW	613522	Colorblindness, tritan, 190900 (3), Autosomal dominant
OPTN	602432	Amyotrophic lateral sclerosis 12, 613435 (3); Glaucoma 1, open angle, E, 137760 (3), Autosomal dominant; {Glaucoma, normal tension, susceptibility to}, 606657 (3)
OR2J3	615016	[C3HEX, ability to smell], 615082 (3), Autosomal dominant
ORC1	601902	Meier-Gorlin syndrome 1, 224690 (3), Autosomal recessive
ORC4	603056	Meier-Gorlin syndrome 2, 613800 (3), Autosomal recessive
ORC6	607213	Meier-Gorlin syndrome 3, 613803 (3), Autosomal recessive
OSBPL2	606731	Deafness, autosomal dominant 67, 616340 (3), Autosomal dominant

OSMR	601743	Amyloidosis, primary localized cutaneous, 1, 105250 (3), Autosomal dominant
OSTM1	607649	Osteopetrosis, autosomal recessive 5, 259720 (3), Autosomal recessive
OTC	300461	Ornithine transcarbamylase deficiency, 311250 (3), X-linked recessive
OTOA	607038	Deafness, autosomal recessive 22, 607039 (3), Autosomal recessive
OTOF	603681	Auditory neuropathy, autosomal recessive, 1, 601071 (3), Autosomal recessive; Deafness, autosomal recessive 9, 601071 (3), Autosomal recessive
OTOG	604487	Deafness, autosomal recessive 18B, 614945 (3), Autosomal recessive
OTOGL	614925	Deafness, autosomal recessive 84B, 614944 (3), Autosomal recessive
OTULIN	615712	Autoinflammation, panniculitis, and dermatosis syndrome, 617099 (3), Autosomal recessive
OVOL2	616441	Corneal dystrophy, posterior polymorphous, 1, 122000 (3), Autosomal dominant
OXCT1	601424	Succinyl CoA:3-oxoacid CoA transferase deficiency, 245050 (3)
P2RX1	600845	No OMIM phenotype
P2RX2	600844	Deafness, autosomal dominant 41, 608224 (3), Autosomal dominant
P2RY12	600515	Bleeding disorder, platelet-type, 8, 609821 (3), Autosomal recessive
P3H1	610339	Osteogenesis imperfecta, type VIII, 610915 (3), Autosomal recessive
P3H2	610341	Myopia, high, with cataract and vitreoretinal degeneration, 614292 (3), Autosomal recessive
P4HA2	600608	Myopia 25, autosomal dominant, 617238 (3), Autosomal dominant
P4HB	176790	Cole-Carpenter syndrome 1, 112240 (3), Autosomal dominant
PABPN1	602279	Oculopharyngeal muscular dystrophy, 164300 (3), Autosomal dominant
PACS1	607492	Schuurs-Hoeijmakers syndrome, 615009 (3), Autosomal dominant
PADI3	606755	Uncombable hair syndrome, 191480 (3), Autosomal recessive
PADI4	605347	{Rheumatoid arthritis, susceptibility to}, 180300 (3)
PADI6	610363	Preimplantation embryonic lethality 2, 617234 (3), Autosomal recessive
PAFAH1B1	601545	Lissencephaly 1, 607432 (3), Isolated cases; Subcortical laminar heterotopia, 607432 (3), Isolated cases
PAH	612349	[Hyperphenylalaninemia, non-PKU mild], 261600 (3), Autosomal recessive; Phenylketonuria, 261600 (3), Autosomal recessive
PALB2	610355	{Breast cancer, susceptibility to}, 114480 (3), Autosomal dominant; Fanconi anemia, complementation group N, 610832 (3); {Pancreatic cancer, susceptibility to, 3}, 613348 (3)
PALLD	608092	{Pancreatic cancer, susceptibility to, 1}, 606856 (3)
PAM16	614336	Spondylometaphyseal dysplasia, Megarbane-Dagher-Melike type, 613320 (3), Autosomal recessive
PAPSS2	603005	Brachyolmia 4 with mild epiphyseal and metaphyseal changes, 612847 (3), Autosomal recessive
PARK7	602533	Parkinson disease 7, autosomal recessive early-onset, 606324 (3), Autosomal recessive

PARN	604212	Dyskeratosis congenita, autosomal recessive 6, 616353 (3), Autosomal recessive; Pulmonary fibrosis and/or bone marrow failure, telomere-related, 4, 616371 (3), Autosomal dominant
PAX1	167411	?Otofaciocervical syndrome 2, 615560 (3), Autosomal recessive
PAX3	606597	Craniofacial-deafness-hand syndrome, 122880 (3), Autosomal dominant; Rhabdomyosarcoma 2, alveolar, 268220 (3), Autosomal recessive; Waardenburg syndrome, type 1, 193500 (3), Autosomal dominant; Waardenburg syndrome, type 3, 148820 (3), Autosomal recessive, Autosomal dominant
PAX4	167413	{Diabetes mellitus, ketosis-prone, susceptibility to}, 612227 (3), Autosomal recessive, Autosomal dominant; Diabetes mellitus, type 2, 125853 (3), Autosomal dominant; Maturity-onset diabetes of the young, type IX, 612225 (3)
PAX5	167414	{Leukemia, acute lymphoblastic, susceptibility to, 3}, 615545 (3)
PAX6	607108	Aniridia, 106210 (3), Autosomal dominant; Anterior segment dysgenesis 5, multiple subtypes, 604229 (3); Cataract with late-onset corneal dystrophy, 106210 (3), Autosomal dominant; ?Coloboma of optic nerve, 120430 (3), Autosomal dominant; ?Coloboma, ocular, 120200 (3), Autosomal dominant; Foveal hypoplasia 1, 136520 (3), Autosomal dominant; Keratitis, 148190 (3), Autosomal dominant; ?Morning glory disc anomaly, 120430 (3), Autosomal dominant; Optic nerve hypoplasia, 165550 (3), Autosomal dominant
PAX7	167410	Rhabdomyosarcoma 2, alveolar, 268220 (3), Autosomal recessive
PAX8	167415	Hypothyroidism, congenital, due to thyroid dysgenesis or hypoplasia, 218700 (3), Autosomal dominant
PAX9	167416	Tooth agenesis, selective, 3, 604625 (3), Autosomal dominant
PAXIP1	608254	No OMIM phenotype
PC	608786	Pyruvate carboxylase deficiency, 266150 (3), Autosomal recessive
PCARE (C2orf71)	613425	Retinitis pigmentosa 54, 613428 (3)
PCBD1	126090	Hyperphenylalaninemia, BH4-deficient, D, 264070 (3), Autosomal recessive
PCCA	232000	Propionicacidemia, 606054 (3), Autosomal recessive
PCCB	232050	Propionicacidemia, 606054 (3), Autosomal recessive
PCDH15	605514	Deafness, autosomal recessive 23, 609533 (3), Autosomal recessive; Usher syndrome, type 1D/F digenic, 601067 (3), Autosomal recessive, Digenic recessive; Usher syndrome, type 1F, 602083 (3), Autosomal recessive
PCDH19	300460	Epileptic encephalopathy, early infantile, 9, 300088 (3), X-linked
PCK1	614168	?Phosphoenolpyruvate carboxykinase deficiency, cytosolic, 261680 (3), Autosomal recessive
PCK2	614095	PEPCK deficiency, mitochondrial, 261650 (1), Autosomal recessive
PCLO	604918	?Pontocerebellar hypoplasia, type 3, 608027 (3), Autosomal recessive
PCM1	600299	No OMIM phenotype
PCNA	176740	?Ataxia-telangiectasia-like disorder 2, 615919 (3), Autosomal recessive

PCNT	605925	Microcephalic osteodysplastic primordial dwarfism, type II, 210720 (3), Autosomal recessive
PCSK1	162150	Obesity with impaired prohormone processing, 600955 (3), Autosomal recessive; {Obesity, susceptibility to, BMIQ12}, 612362 (3)
PCSK9	607786	Hypercholesterolemia, familial, 3, 603776 (3); {Low density lipoprotein cholesterol level QTL 1}, 603776 (3)
PCYT1A	123695	Spondylometaphyseal dysplasia with cone-rod dystrophy, 608940 (3), Autosomal recessive
PDE11A	604961	Pigmented nodular adrenocortical disease, primary, 2, 610475 (3), Autosomal dominant
PDE3A	123805	Hypertension and brachydactyly syndrome, 112410 (3), Autosomal dominant
PDE6A	180071	Retinitis pigmentosa 43, 613810 (3)
PDE6B	180072	Night blindness, congenital stationary, autosomal dominant 2, 163500 (3), Autosomal dominant; Retinitis pigmentosa-40, 613801 (3), Autosomal recessive
PDE6C	600827	Cone dystrophy 4, 613093 (3), Autosomal recessive
PDE6D	602676	?Joubert syndrome 22, 615665 (3), Autosomal recessive
PDE6G	180073	Retinitis pigmentosa 57, 613582 (3), Autosomal recessive
PDE6H	601190	Achromatopsia 6, 610024 (3), Autosomal recessive, Autosomal dominant; Retinal cone dystrophy 3, 610024 (3), Autosomal recessive, Autosomal dominant
PDE8B	603390	Pigmented nodular adrenocortical disease, primary, 3, 614190 (3); Striatal degeneration, autosomal dominant, 609161 (3), Autosomal dominant
PDGFB	190040	Basal ganglia calcification, idiopathic, 5, 615483 (3), Autosomal dominant; Dermatofibrosarcoma protuberans, 607907 (3); Meningioma, SIS-related, 607174 (3), Autosomal dominant
PDGFRA	173490	Gastrointestinal stromal tumor, somatic, 606764 (3); Hypereosinophilic syndrome, idiopathic, resistant to imatinib, 607685 (3), Isolated cases, Somatic mutation
PDGFRB	173410	Basal ganglia calcification, idiopathic, 4, 615007 (3), Autosomal dominant; Kosaki overgrowth syndrome, 616592 (3), Autosomal dominant; Myeloproliferative disorder with eosinophilia, 131440 (4), Autosomal dominant; Myofibromatosis, infantile, 1, 228550 (3), Autosomal dominant; Premature aging syndrome, Penttinen type, 601812 (3), Autosomal dominant
PDGFRL	604584	Colorectal cancer, somatic, 114500 (3); Hepatocellular cancer, somatic, 114550 (3)
PDHA1	300502	Pyruvate dehydrogenase E1-alpha deficiency, 312170 (3), X-linked dominant
PDHB	179060	Pyruvate dehydrogenase E1-beta deficiency, 614111 (3)
PDHX	608769	Lacticacidemia due to PDX1 deficiency, 245349 (3), Autosomal recessive
PDLIM4	603422	{Osteoporosis, susceptibility to}, 166710 (3), Autosomal dominant

PDSS1	607429	Coenzyme Q10 deficiency, primary, 2, 614651 (3), Autosomal recessive
PDSS2	610564	Coenzyme Q10 deficiency, primary, 3, 614652 (3), Autosomal recessive
PDX1	600733	{Diabetes mellitus, type II, susceptibility to}, 125853 (3), Autosomal dominant; MODY, type IV, 606392 (3); Pancreatic agenesis 1, 260370 (3), Autosomal recessive
PDYN	131340	Spinocerebellar ataxia 23, 610245 (3), Autosomal dominant
PDZD7	612971	Deafness, autosomal recessive 57, 618003 (3), Autosomal recessive; {Retinal disease in Usher syndrome type IIA, modifier of}, 276901 (3), Autosomal recessive; Usher syndrome, type IIC, GPR98/PDZD7 digenic, 605472 (3), Autosomal recessive
PEPD	613230	Prolidase deficiency, 170100 (3), Autosomal recessive
PER2	603426	Advanced sleep phase syndrome, familial, 1, 604348 (3), Autosomal dominant
PER3	603427	?Advanced sleep phase syndrome, familial, 3, 616882 (3), Autosomal dominant
PET100	614770	Mitochondrial complex IV deficiency, 220110 (3), Autosomal recessive, Mitochondrial
PEX1	602136	Heimler syndrome 1, 234580 (3), Autosomal recessive; Peroxisome biogenesis disorder 1A (Zellweger), 214100 (3), Autosomal recessive; Peroxisome biogenesis disorder 1B (NALD/IRD), 601539 (3), Autosomal recessive
PEX10	602859	Peroxisome biogenesis disorder 6A (Zellweger), 614870 (3), Autosomal recessive; Peroxisome biogenesis disorder 6B, 614871 (3), Autosomal recessive
PEX11B	603867	?Peroxisome biogenesis disorder 14B, 614920 (3), Autosomal recessive
PEX12	601758	Peroxisome biogenesis disorder 3A (Zellweger), 614859 (3), Autosomal recessive; Peroxisome biogenesis disorder 3B, 266510 (3), Autosomal recessive
PEX13	601789	Peroxisome biogenesis disorder 11A (Zellweger), 614883 (3), Autosomal recessive; Peroxisome biogenesis disorder 11B, 614885 (3), Autosomal recessive
PEX14	601791	Peroxisome biogenesis disorder 13A (Zellweger), 614887 (3), Autosomal recessive
PEX16	603360	Peroxisome biogenesis disorder 8A (Zellweger), 614876 (3), Autosomal recessive; Peroxisome biogenesis disorder 8B, 614877 (3), Autosomal recessive
PEX19	600279	Peroxisome biogenesis disorder 12A (Zellweger), 614886 (3), Autosomal recessive
PEX26	608666	Peroxisome biogenesis disorder 7A (Zellweger), 614872 (3), Autosomal recessive; Peroxisome biogenesis disorder 7B, 614873 (3), Autosomal recessive
PEX3	603164	Peroxisome biogenesis disorder 10A (Zellweger), 614882 (3), Autosomal recessive; ?Peroxisome biogenesis disorder 10B, 617370 (3), Autosomal recessive

PEX6	601498	Heimler syndrome 2, 616617 (3), Autosomal recessive; Peroxisome biogenesis disorder 4A (Zellweger), 614862 (3), Autosomal recessive; Peroxisome biogenesis disorder 4B, 614863 (3), Autosomal recessive, Autosomal dominant
PEX7	601757	Peroxisome biogenesis disorder 9B, 614879 (3); Rhizomelic chondrodysplasia punctata, type 1, 215100 (3), Autosomal recessive
PFKM	610681	Glycogen storage disease VII, 232800 (3), Autosomal recessive
PFN1	176610	Amyotrophic lateral sclerosis 18, 614808 (3)
PGA3	169710	No OMIM phenotype
PGAM2	612931	Glycogen storage disease X, 261670 (3), Autosomal recessive
PGAP1	611655	Mental retardation, autosomal recessive 42, 615802 (3), Autosomal recessive
PGAP3	611801	Hyperphosphatasia with mental retardation syndrome 4, 615716 (3), Autosomal recessive
PGK1	311800	Phosphoglycerate kinase 1 deficiency, 300653 (3), X-linked recessive
PGM3	172100	Immunodeficiency 23, 615816 (3), Autosomal recessive
PGR	607311	?Progesterone resistance, 264080 (2), Autosomal recessive
PHB	176705	{Breast cancer, susceptibility to}, 114480 (3), Autosomal dominant
PHEX	300550	Hypophosphatemic rickets, X-linked dominant, 307800 (3), X-linked dominant
PHF11	607796	No OMIM phenotype
PHF6	300414	Borjeson-Forssman-Lehmann syndrome, 301900 (3), X-linked recessive
PHKA1	311870	Muscle glycogenosis, 300559 (3), X-linked recessive
PHKA2	300798	Glycogen storage disease, type IXa1, 306000 (3), X-linked recessive; Glycogen storage disease, type IXa2, 306000 (3), X-linked recessive
PHKB	172490	Phosphorylase kinase deficiency of liver and muscle, autosomal recessive, 261750 (3), Autosomal recessive
PHKG2	172471	Cirrhosis due to liver phosphorylase kinase deficiency (3); Glycogen storage disease IXc, 613027 (3), Autosomal recessive
PHOX2A	602753	Fibrosis of extraocular muscles, congenital, 2, 602078 (3), Autosomal recessive
PHOX2B	603851	Central hypoventilation syndrome, congenital, with or without Hirschsprung disease, 209880 (3), Autosomal dominant; Neuroblastoma with Hirschsprung disease, 613013 (3); {Neuroblastoma, susceptibility to, 2}, 613013 (3)
PHYH	602026	Refsum disease, 266500 (3), Autosomal recessive
PHYKPL	614683	[?Phosphohydroxylysinuria], 615011 (3)
PI4KA	600286	Polymicrogyria, perisylvian, with cerebellar hypoplasia and arthrogryposis, 616531 (3), Autosomal recessive
PIEZO1	611184	Dehydrated hereditary stomatocytosis with or without pseudohyperkalemia and/or perinatal edema, 194380 (3), Autosomal dominant; Lymphedema, hereditary, III, 616843 (3), Autosomal recessive

PIEZO2	613629	Arthrogryposis, distal, type 3, 114300 (3), Autosomal dominant; Arthrogryposis, distal, type 5, 108145 (3), Autosomal dominant; Arthrogryposis, distal, with impaired proprioception and touch, 617146 (3), Autosomal recessive; ?Marden-Walker syndrome, 248700 (3), Autosomal dominant
PIGA	311770	Multiple congenital anomalies-hypotonia-seizures syndrome 2, 300868 (3), X-linked recessive; Paroxysmal nocturnal hemoglobinuria, somatic, 300818 (3)
PIGG	616918	Mental retardation, autosomal recessive 53, 616917 (3), Autosomal recessive
PIGL	605947	CHIME syndrome, 280000 (3), Autosomal recessive
PIGM	610273	Glycosylphosphatidylinositol deficiency, 610293 (3), Autosomal recessive
PIGO	614730	Hyperphosphatasia with mental retardation syndrome 2, 614749 (3), Autosomal recessive
PIGT	610272	Multiple congenital anomalies-hypotonia-seizures syndrome 3, 615398 (3), Autosomal recessive; ?Paroxysmal nocturnal hemoglobinuria 2, 615399 (3), Autosomal dominant, Somatic mutation
PIGV	610274	Hyperphosphatasia with mental retardation syndrome 1, 239300 (3), Autosomal recessive
PIGW	610275	Glycosylphosphatidylinositol biosynthesis defect 11, 616025 (3), Autosomal recessive
PIH1D3	300933	Ciliary dyskinesia, primary, 36, X-linked, 300991 (3), X-linked recessive
PIK3CA	171834	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)
PIK3CD	602839	Immunodeficiency 14, 615513 (3), Autosomal dominant
PIK3R1	171833	?Agammaglobulinemia 7, autosomal recessive, 615214 (3), Autosomal recessive; Immunodeficiency 36, 616005 (3), Autosomal dominant; SHORT syndrome, 269880 (3), Autosomal dominant
PIK3R2	603157	Megalencephaly-polymicrogyria-polydactyly-hydrocephalus syndrome 1, 603387 (3), Autosomal dominant
PIK3R5	611317	Ataxia-oculomotor apraxia 3, 615217 (3), Autosomal recessive
PIKFYVE	609414	Corneal fleck dystrophy, 121850 (3), Autosomal dominant
PINK1	608309	Parkinson disease 6, early onset, 605909 (3), Autosomal recessive
PITPNM3	608921	Cone-rod dystrophy 5, 600977 (3), Autosomal dominant
PITX1	602149	Clubfoot, congenital, with or without deficiency of long bones and/or mirror-image polydactyly, 119800 (3), Autosomal dominant; Liebenberg syndrome, 186550 (4), Autosomal dominant

PITX3	602669	Anterior segment dysgenesis 1, multiple subtypes, 107250 (3), Autosomal dominant; Cataract 11, multiple types, 610623 (3), Autosomal dominant; Cataract 11, syndromic, 610623 (3), Autosomal dominant
PJVK (DFNB59)	610219	Deafness, autosomal recessive 59, 610220 (3), Autosomal recessive
PKD1	601313	Polycystic kidney disease 1, 173900 (3), Autosomal dominant
PKD1L1	609721	Heterotaxy, visceral, 8, autosomal, 617205 (3), Autosomal recessive
PKD2	173910	Polycystic kidney disease 2, 613095 (3), Autosomal dominant
PKHD1	606702	Polycystic kidney disease 4, with or without hepatic disease, 263200 (3), Autosomal recessive
PKLR	609712	Adenosine triphosphate, elevated, of erythrocytes, 102900 (3), Autosomal dominant; Pyruvate kinase deficiency, 266200 (3), Autosomal recessive
PKP1	601975	Ectodermal dysplasia/skin fragility syndrome, 604536 (3)
PKP2	602861	Arrhythmogenic right ventricular dysplasia 9, 609040 (3), Autosomal dominant
PLA2G5	601192	[Fleck retina, familial benign], 228980 (3), Autosomal recessive
PLA2G6	603604	Infantile neuroaxonal dystrophy 1, 256600 (3), Autosomal recessive; Neurodegeneration with brain iron accumulation 2B, 610217 (3), Autosomal recessive; Parkinson disease 14, autosomal recessive, 612953 (3), Autosomal recessive
PLA2G7	601690	{Asthma, susceptibility to}, 600807 (3), Autosomal dominant; {Atopy, susceptibility to}, 147050 (3), Autosomal dominant; Platelet-activating factor acetylhydrolase deficiency, 614278 (3), Autosomal recessive
PLAG1	603026	Adenomas, salivary gland pleomorphic, somatic, 181030 (3)
PLAT	173370	Hyperfibrinolysis, familial, due to increased release of PLAT, 612348 (1); Thrombophilia, familial, due to decreased release of PLAT, 612348 (1)
PLAU	191840	{Alzheimer disease, late-onset, susceptibility to}, 104300 (3), Autosomal dominant; Quebec platelet disorder, 601709 (3), Autosomal dominant
PLCB4	600810	Auriculocondylar syndrome 2, 614669 (3), Autosomal recessive, Autosomal dominant
PLCD1	602142	Nail disorder, nonsyndromic congenital, 3, (leukonychia), 151600 (3), Autosomal recessive, Autosomal dominant
PLCE1	608414	Nephrotic syndrome, type 3, 610725 (3), Autosomal recessive
PLCG2	600220	Autoinflammation, antibody deficiency, and immune dysregulation syndrome, 614878 (3), Autosomal dominant; Familial cold autoinflammatory syndrome 3, 614468 (3), Autosomal dominant
PLCZ1	608075	?Spermatogenic failure 17, 617214 (3), Autosomal recessive
PLD3	615698	?Spinocerebellar ataxia 46, 617770 (3), Autosomal dominant

PLEC	601282	Epidermolysis bullosa simplex with muscular dystrophy, 226670 (3), Autosomal recessive; ?Epidermolysis bullosa simplex with nail dystrophy, 616487 (3), Autosomal recessive; Epidermolysis bullosa simplex with pyloric atresia, 612138 (3), Autosomal recessive; Epidermolysis bullosa simplex, Ogna type, 131950 (3), Autosomal dominant; Muscular dystrophy, limb-girdle, type 2Q, 613723 (3), Autosomal recessive
PLEKHA1	607772	No OMIM phenotype
PLEKHG5	611101	Charcot-Marie-Tooth disease, recessive intermediate C, 615376 (3), Autosomal recessive; Spinal muscular atrophy, distal, autosomal recessive, 4, 611067 (3), Autosomal recessive
PLEKHM1	611466	Osteopetrosis, autosomal recessive 6, 611497 (3)
PLG	173350	Dysplasminogenemia, 217090 (3), Autosomal recessive; Plasminogen deficiency, type I, 217090 (3), Autosomal recessive
PLIN1	170290	Lipodystrophy, familial partial, type 4, 613877 (3), Autosomal dominant
PLK4	605031	Microcephaly and chorioretinopathy, autosomal recessive, 2, 616171 (3), Autosomal recessive
PLN	172405	Cardiomyopathy, dilated, 1P, 609909 (3); Cardiomyopathy, hypertrophic, 18, 613874 (3), Autosomal dominant
PLOD1	153454	Ehlers-Danlos syndrome, kyphoscoliotic type, 1, 225400 (3), Autosomal recessive
PLOD2	601865	Bruck syndrome 2, 609220 (3), Autosomal recessive
PLOD3	603066	Lysyl hydroxylase 3 deficiency, 612394 (3), Autosomal recessive
PLP1	300401	Pelizaeus-Merzbacher disease, 312080 (3), X-linked recessive; Spastic paraplegia 2, X-linked, 312920 (3), X-linked recessive
PLPBP	604436	Epilepsy, early-onset, vitamin B6-dependent, 617290 (3), Autosomal recessive
PLS3	300131	Bone mineral density QTL18, osteoporosis, 300910 (3), X-linked dominant
PMM2	601785	Congenital disorder of glycosylation, type Ia, 212065 (3), Autosomal recessive
PMP22	601097	Charcot-Marie-Tooth disease, type 1A, 118220 (3), Autosomal dominant; Charcot-Marie-Tooth disease, type 1E, 118300 (3), Autosomal dominant; Dejerine-Sottas disease, 145900 (3), Autosomal recessive, Autosomal dominant; ?Neuropathy, inflammatory demyelinating, 139393 (3), ?Autosomal dominant; Neuropathy, recurrent, with pressure palsies, 162500 (3), Autosomal dominant; Roussy-Levy syndrome, 180800 (3), Autosomal dominant
PMPCA	613036	Spinocerebellar ataxia, autosomal recessive 2, 213200 (3), Autosomal recessive
PMS1	600258	No OMIM phenotype
PMS2	600259	Colorectal cancer, hereditary nonpolyposis, type 4, 614337 (3); Mismatch repair cancer syndrome, 276300 (3), Autosomal recessive
PMVK	607622	Porokeratosis 1, multiple types, 175800 (3), Autosomal dominant

PNKD	609023	Paroxysmal nonkinesigenic dyskinesia 1, 118800 (3), Autosomal dominant
PNKP	605610	Ataxia-oculomotor apraxia 4, 616267 (3), Autosomal recessive; Microcephaly, seizures, and developmental delay, 613402 (3), Autosomal recessive
PNLIP	246600	?Pancreatic lipase deficiency, 614338 (3), Autosomal recessive
PNP	164050	Immunodeficiency due to purine nucleoside phosphorylase deficiency, 613179 (3), Autosomal recessive
PNPLA1	612121	Ichthyosis, congenital, autosomal recessive 10, 615024 (3), Autosomal recessive
PNPLA2	609059	Neutral lipid storage disease with myopathy, 610717 (3), Autosomal recessive
PNPLA6	603197	Boucher-Neuhauser syndrome, 215470 (3), Autosomal recessive; ?Laurence-Moon syndrome, 245800 (3), Autosomal recessive; Oliver-McFarlane syndrome, 275400 (3), Autosomal recessive; Spastic paraplegia 39, autosomal recessive, 612020 (3), Autosomal recessive
PNPLA8	612123	?Mitochondrial myopathy with lactic acidosis, 251950 (3), Autosomal recessive
PNPO	603287	Pyridoxamine 5'-phosphate oxidase deficiency, 610090 (3), Autosomal recessive
PNPT1	610316	Combined oxidative phosphorylation deficiency 13, 614932 (3), Autosomal recessive; Deafness, autosomal recessive 70, 614934 (3), Autosomal recessive
POC1A	614783	Short stature, onychodysplasia, facial dysmorphism, and hypotrichosis, 614813 (3), Autosomal recessive
POC1B	614784	Cone-rod dystrophy 20, 615973 (3), Autosomal recessive
POF1B	300603	?Premature ovarian failure 2B, 300604 (3), X-linked recessive
POFUT1	607491	Dowling-Degos disease 2, 615327 (3), Autosomal dominant
POGLUT1	615618	Dowling-Degos disease 4, 615696 (3), Autosomal dominant; ?Muscular dystrophy, limb-girdle, type 2Z, 617232 (3), Autosomal recessive
POLE	174762	{Colorectal cancer, susceptibility to, 12}, 615083 (3), Autosomal dominant; FILS syndrome, 615139 (3), Autosomal recessive
POLG	174763	Mitochondrial DNA depletion syndrome 4A (Alpers type), 203700 (3), Autosomal recessive; Mitochondrial DNA depletion syndrome 4B (MNGIE type), 613662 (3), Autosomal recessive; Mitochondrial recessive ataxia syndrome (includes SANDO and SCAE), 607459 (3), Autosomal recessive; Progressive external ophthalmoplegia, autosomal dominant 1, 157640 (3), Autosomal dominant; Progressive external ophthalmoplegia, autosomal recessive 1, 258450 (3), Autosomal recessive
POLG2	604983	Progressive external ophthalmoplegia with mitochondrial DNA deletions, autosomal dominant 4, 610131 (3), Autosomal dominant
POLH	603968	Xeroderma pigmentosum, variant type, 278750 (3), Autosomal recessive

POLR1C	610060	Leukodystrophy, hypomyelinating, 11, 616494 (3), Autosomal recessive; Treacher Collins syndrome 3, 248390 (3), Autosomal recessive
POLR1D	613715	Treacher Collins syndrome 2, 613717 (3), Autosomal recessive, Autosomal dominant
POLR3A	614258	Leukodystrophy, hypomyelinating, 7, with or without oligodontia and/or hypogonadotropic hypogonadism, 607694 (3), Autosomal recessive
POLR3B	614366	Leukodystrophy, hypomyelinating, 8, with or without oligodontia and/or hypogonadotropic hypogonadism, 614381 (3), Autosomal recessive
POMC	176830	Obesity, adrenal insufficiency, and red hair due to POMC deficiency, 609734 (3), Autosomal recessive; {Obesity, early-onset, susceptibility to}, 601665 (3), Autosomal recessive, Autosomal dominant, Multifactorial
POMGNT1	606822	Muscular dystrophy-dystroglycanopathy (congenital with brain and eye anomalies), type A, 3, 253280 (3), Autosomal recessive; Muscular dystrophy-dystroglycanopathy (congenital with mental retardation), type B, 3, 613151 (3), Autosomal recessive; Muscular dystrophy-dystroglycanopathy (limb-girdle), type C, 3, 613157 (3), Autosomal recessive; Retinitis pigmentosa 76, 617123 (3), Autosomal recessive
POMGNT2	614828	Muscular dystrophy-dystroglycanopathy (congenital with brain and eye anomalies, type A, 8, 614830 (3), Autosomal recessive
POMK	615247	Muscular dystrophy-dystroglycanopathy (congenital with brain and eye anomalies), type A, 12, 615249 (3), Autosomal recessive; ?Muscular dystrophy-dystroglycanopathy (limb-girdle), type C, 12, 616094 (3), Autosomal recessive
POMP	613386	Keratosis linearis with ichthyosis congenita and sclerosing keratoderma, 601952 (3), Autosomal recessive
POMT1	607423	Muscular dystrophy-dystroglycanopathy (congenital with brain and eye anomalies), type A, 1, 236670 (3), Autosomal recessive; Muscular dystrophy-dystroglycanopathy (congenital with mental retardation), type B, 1, 613155 (3), Autosomal recessive; Muscular dystrophy-dystroglycanopathy (limb-girdle), type C, 1, 609308 (3), Autosomal recessive
POMT2	607439	Muscular dystrophy-dystroglycanopathy (congenital with brain and eye anomalies), type A, 2, 613150 (3), Autosomal recessive; Muscular dystrophy-dystroglycanopathy (congenital with mental retardation), type B, 2, 613156 (3), Autosomal recessive; Muscular dystrophy-dystroglycanopathy (limb-girdle), type C, 2, 613158 (3), Autosomal recessive
PON1	168820	{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)
POP1	602486	Anauxetic dysplasia 2, 617396 (3), Autosomal recessive

POR	124015	Antley-Bixler syndrome with genital anomalies and disordered steroidogenesis, 201750 (3), Autosomal recessive; Disordered steroidogenesis due to cytochrome P450 oxidoreductase, 613571 (3)
PORCN	300651	Focal dermal hypoplasia, 305600 (3), X-linked dominant
POT1	606478	{Glioma susceptibility 9}, 616568 (3), Autosomal dominant; {Melanoma, cutaneous malignant, susceptibility to, 10}, 615848 (3), Autosomal dominant
POU1F1	173110	Pituitary hormone deficiency, combined, 1, 613038 (3), Autosomal recessive, Autosomal dominant
POU3F4	300039	Deafness, X-linked 2, 304400 (3), X-linked recessive
POU4F3	602460	Deafness, autosomal dominant 15, 602459 (3), Autosomal dominant
POU6F2	609062	{Wilms tumor susceptibility-5}, 601583 (3), Autosomal dominant, Somatic mutation
PPA2	609988	?Sudden cardiac failure, alcohol-induced, 617223 (3), Autosomal recessive; Sudden cardiac failure, infantile, 617222 (3), Autosomal recessive
PPARG	601487	Carotid intimal medial thickness 1, 609338 (3); {Diabetes, type 2}, 125853 (3), Autosomal dominant; Insulin resistance, severe, digenic, 604367 (3), Autosomal dominant; Lipodystrophy, familial partial, type 3, 604367 (3), Autosomal dominant; [Obesity, resistance to] (3); Obesity, severe, 601665 (3), Autosomal recessive, Autosomal dominant, Multifactorial
PPARGC1B	608886	{Obesity, variation in}, 601665 (3), Autosomal recessive, Autosomal dominant, Multifactorial
PPIB	123841	Osteogenesis imperfecta, type IX, 259440 (3), Autosomal recessive
PPM1D	605100	Breast cancer, somatic, 114480 (3); Intellectual developmental disorder with gastrointestinal difficulties and high pain threshold, 617450 (3), Autosomal dominant
PPM1K	611065	?Maple syrup urine disease, mild variant, 615135 (3)
PPOX	600923	Porphyria variegata, 176200 (3), Autosomal dominant
PPP1R15B	613257	Microcephaly, short stature, and impaired glucose metabolism 2, 616817 (3), Autosomal recessive
PPP1R17	604088	{Hypercholesterolemia, susceptibility to}, 143890 (3), Autosomal dominant
PPP1R3A	600917	Insulin resistance, severe, digenic, 125853 (3), Autosomal dominant
PPP2R1A	605983	Mental retardation, autosomal dominant 36, 616362 (3), Autosomal dominant
PPP2R1B	603113	Lung cancer, 211980 (3), Autosomal recessive
PPP2R2B	604325	Spinocerebellar ataxia 12, 604326 (3), Autosomal dominant
PPP2R5D	601646	Mental retardation, autosomal dominant 35, 616355 (3), Autosomal dominant
PPT1	600722	Ceroid lipofuscinosis, neuronal, 1, 256730 (3), Autosomal recessive
PQBP1	300463	Renpenning syndrome, 309500 (3), X-linked recessive
PRCC	179755	Renal cell carcinoma, papillary, 605074 (3)
PRCD	610598	Retinitis pigmentosa 36, 610599 (3)

PRDM12	616458	Neuropathy, hereditary sensory and autonomic, type VIII, 616488 (3), Autosomal recessive
PRDM16	605557	Cardiomyopathy, dilated, 1LL, 615373 (3), Autosomal dominant; Left ventricular noncompaction 8, 615373 (3), Autosomal dominant
PRDM5	614161	Brittle cornea syndrome 2, 614170 (3), Autosomal recessive
PRDM6	616982	Patent ductus arteriosus 3, 617039 (3), Autosomal dominant
PREPL	609557	?Myasthenic syndrome, congenital, 22, 616224 (3), Autosomal recessive
PRF1	170280	Aplastic anemia, 609135 (3); Hemophagocytic lymphohistiocytosis, familial, 2, 603553 (3), Autosomal recessive; Lymphoma, non-Hodgkin, 605027 (3)
PRG4	604283	Camptodactyly-arthropathy-coxa vara-pericarditis syndrome, 208250 (3), Autosomal recessive
PRICKLE1	608500	Epilepsy, progressive myoclonic 1B, 612437 (3), Autosomal recessive
PRIMPOL	615421	Myopia 22, autosomal dominant, 615420 (3), Autosomal dominant
PRKACA	601639	Cushing syndrome, ACTH-independent adrenal, somatic, 615830 (3)
PRKACG	176893	?Bleeding disorder, platelet-type, 19, 616176 (3), Autosomal recessive
PRKAG2	602743	Cardiomyopathy, hypertrophic 6, 600858 (3), Autosomal dominant; Glycogen storage disease of heart, lethal congenital, 261740 (3), Autosomal dominant; Wolff-Parkinson-White syndrome, 194200 (3), ?Autosomal dominant
PRKAR1A	188830	Acrodysostosis 1, with or without hormone resistance, 101800 (3), Autosomal dominant; Adrenocortical tumor, somatic (3); Carney complex, type 1, 160980 (3), Autosomal dominant; Myxoma, intracardiac, 255960 (3), Autosomal dominant; Pigmented nodular adrenocortical disease, primary, 1, 610489 (3), Autosomal dominant
PRKCG	176980	Spinocerebellar ataxia 14, 605361 (3), Autosomal dominant
PRKCH	605437	{Cerebral infarction, susceptibility to}, 601367 (3), Multifactorial
PRKCSH	177060	Polycystic liver disease 1, 174050 (3), Autosomal dominant
PRKD1	605435	Congenital heart defects and ectodermal dysplasia, 617364 (3), Autosomal dominant
PRKDC	600899	Immunodeficiency 26, with or without neurologic abnormalities, 615966 (3), Autosomal recessive
PRKG1	176894	Aortic aneurysm, familial thoracic 8, 615436 (3), Autosomal dominant
PRKN	602544	Adenocarcinoma of lung, somatic, 211980 (3); Adenocarcinoma, ovarian, somatic, 167000 (3); {Leprosy, susceptibility to}, 607572 (3); Parkinson disease, juvenile, type 2, 600116 (3), Autosomal recessive
PRKRA	603424	Dystonia 16, 612067 (3), Autosomal recessive
PRLR	176761	?Hyperprolactinemia, 615555 (3), Autosomal dominant; Multiple fibroadenomas of the breast, 615554 (3), Autosomal dominant

PRNP	176640	Cerebral amyloid angiopathy, PRNP-related, 137440 (3), Autosomal dominant; Creutzfeldt-Jakob disease, 123400 (3), Autosomal dominant; Gerstmann-Straussler disease, 137440 (3), Autosomal dominant; Huntington disease-like 1, 603218 (3), Autosomal dominant; Insomnia, fatal familial, 600072 (3), Autosomal dominant; {Kuru, susceptibility to}, 245300 (3); Prion disease with protracted course, 606688 (3), Autosomal dominant
PROC	612283	Thrombophilia due to protein C deficiency, autosomal dominant, 176860 (3), Autosomal dominant; Thrombophilia due to protein C deficiency, autosomal recessive, 612304 (3), Autosomal recessive
PRODH	606810	Hyperprolinemia, type I, 239500 (3), Autosomal recessive; {Schizophrenia, susceptibility to, 4}, 600850 (3), Autosomal dominant
PROK2	607002	Hypogonadotropic hypogonadism 4 with or without anosmia, 610628 (3), Autosomal dominant
PROKR2	607123	Hypogonadotropic hypogonadism 3 with or without anosmia, 244200 (3), Autosomal dominant
PROM1	604365	Cone-rod dystrophy 12, 612657 (3); Macular dystrophy, retinal, 2, 608051 (3), Autosomal dominant; Retinitis pigmentosa 41, 612095 (3), Autosomal recessive; Stargardt disease 4, 603786 (3)
PROP1	601538	Pituitary hormone deficiency, combined, 2, 262600 (3), Autosomal recessive
PROS1	176880	Thrombophilia due to protein S deficiency, autosomal dominant, 612336 (3), Autosomal dominant; Thrombophilia due to protein S deficiency, autosomal recessive, 614514 (3), Autosomal recessive
PROZ	176895	[Protein Z deficiency], 614024 (3)
PRPF3	607301	Retinitis pigmentosa 18, 601414 (3), Autosomal dominant
PRPF31	606419	Retinitis pigmentosa 11, 600138 (3), Autosomal dominant
PRPF4	607795	Retinitis pigmentosa 70, 615922 (3), Autosomal dominant
PRPF6	613979	Retinitis pigmentosa 60, 613983 (3), Autosomal dominant
PRPF8	607300	Retinitis pigmentosa 13, 600059 (3), Autosomal dominant
PRPH	170710	{Amyotrophic lateral sclerosis, susceptibility to}, 105400 (3), Autosomal recessive, Autosomal dominant
PRPH2	179605	Choroidal dystrophy, central areolar 2, 613105 (3), Autosomal dominant; Leber congenital amaurosis 18, 608133 (3), Autosomal recessive, Autosomal dominant; Macular dystrophy, patterned, 1, 169150 (3), Autosomal dominant; Macular dystrophy, vitelliform, 3, 608161 (3), Autosomal dominant; Retinitis pigmentosa 7 and digenic, 608133 (3), Autosomal recessive, Autosomal dominant; Retinitis punctata albescens, 136880 (3), Autosomal recessive, Autosomal dominant
PRPS1	311850	Arts syndrome, 301835 (3), X-linked recessive; Charcot-Marie-Tooth disease, X-linked recessive, 5, 311070 (3), X-linked recessive; Deafness, X-linked 1, 304500 (3), X-linked; Gout, PRPS-related, 300661 (3), X-linked recessive; Phosphoribosylpyrophosphate synthetase superactivity, 300661 (3), X-linked recessive

PRRX1	167420	Agnathia-otocephaly complex, 202650 (3), Autosomal recessive, Autosomal dominant
PRSS1	276000	Pancreatitis, hereditary, 167800 (3), Autosomal dominant; Trypsinogen deficiency, 614044 (1), Autosomal recessive
PRSS12	606709	Mental retardation, autosomal recessive 1, 249500 (3), Autosomal recessive
PRSS2	601564	{Pancreatitis, chronic, protection against}, 167800 (3), Autosomal dominant
PRSS56	613858	Microphthalmia, isolated 6, 613517 (3), Autosomal recessive
PRX	605725	Charcot-Marie-Tooth disease, type 4F, 614895 (3), Autosomal recessive; Dejerine-Sottas disease, 145900 (3), Autosomal recessive, Autosomal dominant
PRY	400019	No OMIM phenotype
PRY2	400041	No OMIM phenotype
PSAP	176801	Combined SAP deficiency, 611721 (3), Autosomal recessive; Gaucher disease, atypical, 610539 (3); Krabbe disease, atypical, 611722 (3), Autosomal recessive; Metachromatic leukodystrophy due to SAP-b deficiency, 249900 (3), Autosomal recessive
PSAT1	610936	Neu-Laxova syndrome 2, 616038 (3), Autosomal recessive; ?Phosphoserine aminotransferase deficiency, 610992 (3), Autosomal recessive
PSEN1	104311	?Acne inversa, familial, 3, 613737 (3), Autosomal dominant; Alzheimer disease, type 3, 607822 (3), Autosomal dominant; Alzheimer disease, type 3, with spastic paraparesis and apraxia, 607822 (3), Autosomal dominant; Alzheimer disease, type 3, with spastic paraparesis and unusual plaques, 607822 (3), Autosomal dominant; Cardiomyopathy, dilated, 1U, 613694 (3), Autosomal dominant; Dementia, frontotemporal, 600274 (3), Autosomal dominant; Pick disease, 172700 (3), Autosomal dominant, Isolated cases
PSEN2	600759	Alzheimer disease-4, 606889 (3), Autosomal dominant; Cardiomyopathy, dilated, 1V, 613697 (3), Autosomal dominant
PSENEN	607632	Acne inversa, familial, 2, with or without Dowling-Degos disease, 613736 (3), Autosomal dominant
PSMB8	177046	Autoinflammation, lipodystrophy, and dermatosis syndrome, 256040 (3), Autosomal recessive
PSMC3IP	608665	Ovarian dysgenesis 3, 614324 (3), Autosomal recessive
PSPH	172480	Phosphoserine phosphatase deficiency, 614023 (3), Autosomal recessive
PSTPIP1	606347	Pyogenic sterile arthritis, pyoderma gangrenosum, and acne, 604416 (3), Autosomal dominant
PTCH1	601309	Basal cell carcinoma, somatic, 605462 (3); Basal cell nevus syndrome, 109400 (3), Autosomal dominant; Holoprosencephaly 7, 610828 (3), Autosomal dominant
PTCH2	603673	Basal cell carcinoma, somatic, 605462 (3); Basal cell nevus syndrome, 109400 (3), Autosomal dominant; Medulloblastoma, somatic, 155255 (3)

PTCHD1	300828	{Autism, susceptibility to, X-linked 4}, 300830 (3), X-linked recessive
PTDSS1	612792	Lenz-Majewski hyperostotic dwarfism, 151050 (3), Autosomal dominant
PTEN	601728	Bannayan-Riley-Ruvalcaba syndrome, 153480 (3); Cowden syndrome 1, 158350 (3), Autosomal dominant; Endometrial carcinoma, somatic, 608089 (3); {Glioma susceptibility 2}, 613028 (3); Lhermitte-Duclos syndrome, 158350 (3), Autosomal dominant; Macrocephaly/autism syndrome, 605309 (3), Autosomal dominant; Malignant melanoma, somatic, 155600 (3); {Meningioma}, 607174 (3), Autosomal dominant; PTEN hamartoma tumor syndrome (3); {Prostate cancer, somatic}, 176807 (3); Squamous cell carcinoma, head and neck, somatic, 275355 (3); VATER association with macrocephaly and ventriculomegaly, 276950 (3), Autosomal recessive
PTF1A	607194	Pancreatic agenesis 2, 615935 (3), Autosomal recessive; Pancreatic and cerebellar agenesis, 609069 (3), Autosomal recessive
PTGDR	604687	{Asthma, susceptibility to, 1}, 607277 (3)
PTGER2	176804	{Asthma, aspirin-induced, susceptibility to}, 208550 (3), Autosomal recessive
PTGIS	601699	Hypertension, essential, 145500 (3), Multifactorial
PTH	168450	Hypoparathyroidism, autosomal dominant, 146200 (3), Autosomal dominant; Hypoparathyroidism, autosomal recessive, 146200 (3), Autosomal dominant
PTHLH	168470	Brachydactyly, type E2, 613382 (3), Autosomal dominant
PTPN1	176885	{Insulin resistance, susceptibility to}, 125853 (3), Autosomal dominant
PTPN11	176876	LEOPARD syndrome 1, 151100 (3), Autosomal dominant; Leukemia, juvenile myelomonocytic, somatic, 607785 (3); Metachondromatosis, 156250 (3), Autosomal dominant; Noonan syndrome 1, 163950 (3), Autosomal dominant
PTPN14	603155	?Choanal atresia and lymphedema, 613611 (3), Autosomal recessive
PTPN22	600716	{Diabetes, type 1, susceptibility to}, 222100 (3), Autosomal recessive; {Rheumatoid arthritis, susceptibility to}, 180300 (3); {Systemic lupus erythematosus susceptibility to}, 152700 (3), Autosomal dominant
PTPRC	151460	{Hepatitis C virus, susceptibility to}, 609532 (3); Severe combined immunodeficiency, T cell-negative, B-cell/natural killer-cell positive, 608971 (3), Autosomal recessive
PTPRF	179590	?Breasts and/or nipples, aplasia or hypoplasia of, 2, 616001 (3), Autosomal recessive
PTPRO	600579	Nephrotic syndrome, type 6, 614196 (3), Autosomal recessive
PTRH2	608625	Infantile-onset multisystem neurologic, endocrine, and pancreatic disease, 616263 (3), Autosomal recessive
PTS	612719	Hyperphenylalaninemia, BH4-deficient, A, 261640 (3), Autosomal recessive
PUF60	604819	Verheij syndrome, 615583 (3), Autosomal dominant
PURA	600473	Mental retardation, autosomal dominant 31, 616158 (3), Autosomal dominant

PUS1	608109	Myopathy, lactic acidosis, and sideroblastic anemia 1, 600462 (3), Autosomal recessive
PUS3	616283	?Mental retardation, autosomal recessive 55, 617051 (3), Autosomal recessive
PXDN	605158	Anterior segment dysgenesis 7, with sclerocornea, 269400 (3), Autosomal recessive
PYCR2	616406	Leukodystrophy, hypomyelinating, 10, 616420 (3), Autosomal recessive
PYGL	613741	Glycogen storage disease VI, 232700 (3), Autosomal recessive
PYGM	608455	McArdle disease, 232600 (3), Autosomal recessive
PYROXD1	617220	Myopathy, myofibrillar, 8, 617258 (3), Autosomal recessive
QARS	603727	Microcephaly, progressive, seizures, and cerebral and cerebellar atrophy, 615760 (3), Autosomal recessive
QDPR	612676	Hyperphenylalaninemia, BH4-deficient, C, 261630 (3), Autosomal recessive
RAB18	602207	Warburg micro syndrome 3, 614222 (3), Autosomal recessive
RAB23	606144	Carpenter syndrome, 201000 (3), Autosomal recessive
RAB28	612994	Cone-rod dystrophy 18, 615374 (3), Autosomal recessive
RAB33B	605950	Smith-McCort dysplasia 2, 615222 (3), Autosomal recessive
RAB3GAP1	602536	Warburg micro syndrome 1, 600118 (3), Autosomal recessive
RAB3GAP2	609275	Martsolf syndrome, 212720 (3), Autosomal recessive; Warburg micro syndrome 2, 614225 (3), Autosomal recessive
RAB7A	602298	Charcot-Marie-Tooth disease, type 2B, 600882 (3), Autosomal dominant
RAC2	602049	Neutrophil immunodeficiency syndrome, 608203 (3)
RAD21	606462	Cornelia de Lange syndrome 4, 614701 (3), Autosomal dominant
RAD50	604040	Nijmegen breakage syndrome-like disorder, 613078 (3)
RAD51	179617	{Breast cancer, susceptibility to}, 114480 (3), Autosomal dominant; ?Fanconi anemia, complementation group R, 617244 (3), Autosomal dominant; Mirror movements 2, 614508 (3), Autosomal dominant
RAD51C	602774	{Breast-ovarian cancer, familial, susceptibility to, 3}, 613399 (3); Fanconi anemia, complementation group O, 613390 (3), Autosomal recessive
RAD51D	602954	{Breast-ovarian cancer, familial, susceptibility to, 4}, 614291 (3)
RAD54B	604289	Colon cancer, somatic, 114500 (3); Lymphoma, non-Hodgkin, somatic, 605027 (3)
RAD54L	603615	Adenocarcinoma, colonic, somatic (3); {Breast cancer, invasive ductal}, 114480 (3), Autosomal dominant; Lymphoma, non-Hodgkin, somatic, 605027 (3)
RAF1	164760	Cardiomyopathy, dilated, 1NN, 615916 (3), Autosomal dominant; LEOPARD syndrome 2, 611554 (3); Noonan syndrome 5, 611553 (3), Autosomal dominant

RAG1	179615	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), Autosomal recessive; Omenn syndrome, 603554 (3), Autosomal recessive; Severe combined immunodeficiency, B cell-negative, 601457 (3), Autosomal recessive
RAG2	179616	Combined cellular and humoral immune defects with granulomas, 233650 (3), Autosomal recessive; Omenn syndrome, 603554 (3), Autosomal recessive; Severe combined immunodeficiency, B cell-negative, 601457 (3), Autosomal recessive
RANBP2	601181	{Encephalopathy, acute, infection-induced, 3, susceptibility to}, 608033 (3), Autosomal dominant
RAPSN	601592	Fetal akinesia deformation sequence, 208150 (3), Autosomal recessive; Myasthenic syndrome, congenital, 11, associated with acetylcholine receptor deficiency, 616326 (3), Autosomal recessive
RARA	180240	Leukemia, acute promyelocytic, 612376 (1)
RARB	180220	Microphthalmia, syndromic 12, 615524 (3), Autosomal recessive, Autosomal dominant
RARS	107820	Leukodystrophy, hypomyelinating, 9, 616140 (3), Autosomal recessive
RARS2	611524	Pontocerebellar hypoplasia, type 6, 611523 (3), Autosomal recessive
RASA1	139150	Basal cell carcinoma, somatic, 605462 (3); Capillary malformation-arteriovenous malformation, 608354 (3), Autosomal dominant; Parkes Weber syndrome, 608355 (3), Autosomal dominant
RASSF1	605082	Lung cancer, 211980 (2), Autosomal recessive
RAX	601881	Microphthalmia, isolated 3, 611038 (3), Autosomal recessive
RAX2	610362	Cone-rod dystrophy 11, 610381 (3), Autosomal dominant; ?Macular degeneration, age-related, 6, 613757 (3)
RB1	614041	Bladder cancer, somatic, 109800 (3); Osteosarcoma, somatic, 259500 (3); Retinoblastoma, 180200 (3), Autosomal dominant, Somatic mutation; Retinoblastoma, trilateral, 180200 (3), Autosomal dominant, Somatic mutation; Small cell cancer of the lung, somatic, 182280 (3)
RB1CC1	606837	Breast cancer, somatic, 114480 (3)
RBBP8	604124	Jawad syndrome, 251255 (3), Autosomal recessive; Pancreatic carcinoma, somatic (3); Seckel syndrome 2, 606744 (3), Autosomal recessive
RBCK1	610924	Polyglucosan body myopathy 1 with or without immunodeficiency, 615895 (3), Autosomal recessive
RBM10	300080	TARP syndrome, 311900 (3), X-linked recessive
RBM20	613171	Cardiomyopathy, dilated, 1DD, 613172 (3), Autosomal dominant
RBM28	612074	?Alopecia, neurologic defects, and endocrinopathy syndrome, 612079 (3), Autosomal recessive
RBM8A	605313	Thrombocytopenia-absent radius syndrome, 274000 (3), Autosomal recessive

RBMX	300199	?Mental retardation, X-linked, syndromic 11, Shashi type, 300238 (3), X-linked recessive
RBMY1A1	400006	No OMIM phenotype
RBP3	180290	?Retinitis pigmentosa 66, 615233 (3), Autosomal recessive
RBP4	180250	Microphthalmia, isolated, with coloboma 10, 616428 (3), Autosomal dominant; Retinal dystrophy, iris coloboma, and comedogenic acne syndrome, 615147 (3), Autosomal recessive
RCBTB1	607867	Retinal dystrophy with or without extraocular anomalies, 617175 (3), Autosomal recessive
RD3	180040	Leber congenital amaurosis 12, 610612 (3), Autosomal recessive
RDH11	607849	?Retinal dystrophy, juvenile cataracts, and short stature syndrome, 616108 (3), Autosomal recessive
RDH12	608830	Leber congenital amaurosis 13, 612712 (3), Autosomal recessive
RDH5	601617	Fundus albipunctatus, 136880 (3), Autosomal recessive, Autosomal dominant
RDX	179410	Deafness, autosomal recessive 24, 611022 (3), Autosomal recessive
RECQL4	603780	Baller-Gerold syndrome, 218600 (3), Autosomal recessive; RAPADILINO syndrome, 266280 (3), Autosomal recessive; Rothmund-Thomson syndrome, 268400 (3), Autosomal recessive
REEP2	609347	?Spastic paraplegia 72, autosomal dominant, 615625 (3), Autosomal recessive, Autosomal dominant; ?Spastic paraplegia 72, autosomal recessive, 615625 (3), Autosomal recessive, Autosomal dominant
REEP6	609346	Retinitis pigmentosa 77, 617304 (3), Autosomal recessive
RELN	600514	{Epilepsy, familial temporal lobe, 7}, 616436 (3), Autosomal dominant; Lissencephaly 2 (Norman-Roberts type), 257320 (3), Autosomal recessive
REN	179820	[Hyperproreninemia] (3); Hyperuricemic nephropathy, familial juvenile 2, 613092 (3), Autosomal dominant; Renal tubular dysgenesis, 267430 (3), Autosomal recessive
RERE	605226	Neurodevelopmental disorder with or without anomalies of the brain, eye, or heart, 616975 (3), Autosomal dominant
REST	600571	Fibromatosis, gingival, 5, 617626 (3), Autosomal dominant; {Wilms tumor 6, susceptibility to}, 616806 (3)
RET	164761	Central hypoventilation syndrome, congenital, 209880 (3), Autosomal dominant; {Hirschsprung disease, protection against}, 142623 (3), Autosomal dominant; {Hirschsprung disease, susceptibility to, 1}, 142623 (3), Autosomal dominant; Medullary thyroid carcinoma, 155240 (3), Autosomal dominant; Multiple endocrine neoplasia IIA, 171400 (3), Autosomal dominant; Multiple endocrine neoplasia IIB, 162300 (3), Autosomal dominant; Pheochromocytoma, 171300 (3), Autosomal dominant
RETN	605565	{Diabetes mellitus, noninsulin-dependent, susceptibility to}, 125853 (3), Autosomal dominant; {Hypertension, insulin resistance-related, susceptibility to}, 125853 (3), Autosomal dominant

RETREG1	613114	Neuropathy, hereditary sensory and autonomic, type IIB, 613115 (3), Autosomal recessive
RFT1	611908	Congenital disorder of glycosylation, type In, 612015 (3), Autosomal recessive
RFX5	601863	Bare lymphocyte syndrome, type II, complementation group C, 209920 (3), Autosomal recessive; Bare lymphocyte syndrome, type II, complementation group E, 209920 (3), Autosomal recessive
RFX6	612659	Mitchell-Riley syndrome, 615710 (3), Autosomal recessive
RFXAP	601861	Bare lymphocyte syndrome, type II, complementation group D, 209920 (3), Autosomal recessive
RGR	600342	Retinitis pigmentosa 44, 613769 (3)
RGS5	603276	[Blood pressure regulation QTL], 145500 (2), Multifactorial
RGS9	604067	Bradyopsia, 608415 (3)
RGS9BP	607814	Bradyopsia, 608415 (3)
RHAG	180297	Anemia, hemolytic, Rh-null, regulator type, 268150 (3), Autosomal dominant; Overhydrated hereditary stomatocytosis, 185000 (3), Autosomal dominant
RHBDF2	614404	Tylosis with esophageal cancer, 148500 (3), Autosomal dominant
RHCE	111700	[Blood group, Rhesus] (3); Rh-null disease, amorph type, 617970 (3)
RHO	180380	Night blindness, congenital stationary, autosomal dominant 1, 610445 (3); Retinitis pigmentosa 4, autosomal dominant or recessive, 613731 (3), Autosomal recessive, Autosomal dominant; Retinitis punctata albescens, 136880 (3), Autosomal recessive, Autosomal dominant
RIMS1	606629	Cone-rod dystrophy 7, 603649 (3)
RIN2	610222	Macrocephaly, alopecia, cutis laxa, and scoliosis, 613075 (3), Autosomal recessive
RIPK4	605706	Popliteal pterygium syndrome, Bartsocas-Papas type, 263650 (3), Autosomal recessive
RIPOR2	611410	?Deafness, autosomal recessive 104, 616515 (3), Autosomal recessive
RIPPLY2	609891	?Spondylocostal dysostosis 6, 616566 (3), Autosomal recessive
RIT1	609591	Noonan syndrome 8, 615355 (3), Autosomal dominant
RLBP1	180090	Bothnia retinal dystrophy, 607475 (3), Autosomal recessive; Fundus albipunctatus, 136880 (3), Autosomal recessive, Autosomal dominant; Newfoundland rod-cone dystrophy, 607476 (3); Retinitis punctata albescens, 136880 (3), Autosomal recessive, Autosomal dominant
RLIM	300379	Mental retardation, X-linked 61, 300978 (3), X-linked recessive
RMND1	614917	Combined oxidative phosphorylation deficiency 11, 614922 (3), Autosomal recessive
RMRP	157660	Anauxetic dysplasia 1, 607095 (3), Autosomal recessive; Cartilage-hair hypoplasia, 250250 (3), Autosomal recessive; Metaphyseal dysplasia without hypotrichosis, 250460 (3), Autosomal recessive
RNASEH1	604123	Progressive external ophthalmoplegia with mitochondrial DNA deletions, autosomal recessive 2, 616479 (3), Autosomal recessive
RNASEH2A	606034	Aicardi-Goutieres syndrome 4, 610333 (3), Autosomal recessive
RNASEH2B	610326	Aicardi-Goutieres syndrome 2, 610181 (3), Autosomal recessive

RNASEH2C	610330	Aicardi-Goutieres syndrome 3, 610329 (3), Autosomal recessive
RNASEL	180435	Prostate cancer 1, 601518 (3), Autosomal dominant
RNASET2	612944	Leukoencephalopathy, cystic, without megalencephaly, 612951 (3), Autosomal recessive
RNF113A	300951	?Trichothiodystrophy 5, nonphotosensitive, 300953 (3), X-linked dominant
RNF125	610432	Tenorio syndrome, 616260 (3), Autosomal dominant
RNF135	611358	Macrocephaly, macrosomia, facial dysmorphism syndrome, 614192 (3)
RNF139	603046	Renal cell carcinoma, 144700 (3)
RNF168	612688	RIDDLE syndrome, 611943 (3), Autosomal recessive
RNF170	614649	Ataxia, sensory, 1, autosomal dominant, 608984 (3), Autosomal dominant
RNF212	612041	Recombination rate QTL 1, 612042 (3)
RNF216	609948	Cerebellar ataxia and hypogonadotropic hypogonadism, 212840 (3), Autosomal recessive
RNF43	612482	Sessile serrated polyposis cancer syndrome, 617108 (3), Autosomal dominant
RNF6	604242	Esophageal carcinoma, somatic, 133239 (3)
ROBO3	608630	Gaze palsy, familial horizontal, with progressive scoliosis, 1, 607313 (3), Autosomal recessive
ROGDI	614574	Kohlschutter-Tonz syndrome, 226750 (3), Autosomal recessive
ROM1	180721	Retinitis pigmentosa 7, digenic, 608133 (3), Autosomal recessive, Autosomal dominant
ROR2	602337	Brachydactyly, type B1, 113000 (3), Autosomal dominant; Robinow syndrome, autosomal recessive, 268310 (3), Autosomal recessive
RORC	602943	Immunodeficiency 42, 616622 (3), Autosomal recessive
RP1	603937	Retinitis pigmentosa 1, 180100 (3), Autosomal recessive, Autosomal dominant
RP1L1	608581	Occult macular dystrophy, 613587 (3), Autosomal dominant
RP2	300757	Retinitis pigmentosa 2, 312600 (3), X-linked
RP9	607331	?Retinitis pigmentosa 9, 180104 (3), Autosomal dominant
RPE65	180069	Leber congenital amaurosis 2, 204100 (3), Autosomal recessive; Retinitis pigmentosa 20, 613794 (3), Autosomal recessive
RPGR	312610	Cone-rod dystrophy, X-linked, 1, 304020 (3), X-linked; Macular degeneration, X-linked atrophic, 300834 (3), X-linked recessive; Retinitis pigmentosa 3, 300029 (3); Retinitis pigmentosa, X-linked, and sinorespiratory infections, with or without deafness, 300455 (3)
RPGRIP1	605446	Cone-rod dystrophy 13, 608194 (3); Leber congenital amaurosis 6, 613826 (3)
RPGRIP1L	610937	COACH syndrome, 216360 (3), Autosomal recessive; Joubert syndrome 7, 611560 (3), Autosomal recessive; Meckel syndrome 5, 611561 (3), Autosomal recessive
RPIA	180430	?Ribose 5-phosphate isomerase deficiency, 608611 (3), Autosomal recessive

RPL10	312173	{Autism, susceptibility to, X-linked 5}, 300847 (3); Mental retardation, X-linked, syndromic, 35, 300998 (3), X-linked recessive
RPL11	604175	Diamond-Blackfan anemia 7, 612562 (3), Autosomal dominant
RPL15	604174	?Diamond-Blackfan anemia 12, 615550 (3), Autosomal dominant
RPL21	603636	Hypotrichosis 12, 615885 (3), Autosomal dominant
RPL26	603704	?Diamond-Blackfan anemia 11, 614900 (3), Autosomal dominant
RPL35A	180468	Diamond-Blackfan anemia 5, 612528 (3), Autosomal dominant
RPL5	603634	Diamond-Blackfan anemia 6, 612561 (3), Autosomal dominant
RPS10	603632	Diamond-Blackfan anemia 9, 613308 (3), Autosomal dominant
RPS14	130620	Macrocytic anemia, refractory, due to 5q deletion, somatic, 153550 (3)
RPS17	180472	Diamond-Blackfan anemia 4, 612527 (3), Autosomal dominant
RPS19	603474	Diamond-Blackfan anemia 1, 105650 (3), Autosomal dominant
RPS24	602412	Diamond-blackfan anemia 3, 610629 (3), Autosomal dominant
RPS26	603701	Diamond-Blackfan anemia 10, 613309 (3), Autosomal dominant
RPS27	603702	?Diamond-Blackfan anemia 17, 617409 (3), Autosomal dominant
RPS28	603685	Diamond Blackfan anemia 15 with mandibulofacial dysostosis, 606164 (3), Autosomal dominant
RPS29	603633	Diamond-Blackfan anemia 13, 615909 (3), Autosomal dominant
RPS4Y2	400030	No OMIM phenotype
RPS6KA3	300075	Coffin-Lowry syndrome, 303600 (3), X-linked dominant, Isolated cases; Mental retardation, X-linked 19, 300844 (3), X-linked dominant
RPS7	603658	Diamond-Blackfan anemia 8, 612563 (3), Autosomal dominant
RPSA	150370	Asplenia, isolated congenital, 271400 (3), Autosomal dominant
RRM2B	604712	Mitochondrial DNA depletion syndrome 8A (encephalomyopathic type with renal tubulopathy), 612075 (3), Autosomal recessive; Mitochondrial DNA depletion syndrome 8B (MNGIE type), 612075 (3), Autosomal recessive; Progressive external ophthalmoplegia with mitochondrial DNA deletions, autosomal dominant 5, 613077 (3), Autosomal dominant
RS1	300839	Retinoschisis, 312700 (3), X-linked recessive
RSPH1	609314	Ciliary dyskinesia, primary, 24, 615481 (3), Autosomal recessive
RSPH3	615876	Ciliary dyskinesia, primary, 32, 616481 (3), Autosomal recessive
RSPH4A	612647	Ciliary dyskinesia, primary, 11, 612649 (3)
RSPH9	612648	Ciliary dyskinesia, primary, 12, 612650 (3)
RSPO1	609595	Palmoplantar hyperkeratosis and true hermaphroditism, 610644 (3), Autosomal recessive; Palmoplantar hyperkeratosis with squamous cell carcinoma of skin and sex reversal, 610644 (3), Autosomal recessive
RSPO4	610573	Anonychia congenita, 206800 (3), Autosomal recessive
RSPRY1	616585	Spondyloepimetaphyseal dysplasia, Faden-Alkuraya type, 616723 (3), Autosomal recessive
RTEL1	608833	Dyskeratosis congenita, autosomal dominant 4, 615190 (3), Autosomal recessive, Autosomal dominant; Dyskeratosis congenita, autosomal recessive 5, 615190 (3), Autosomal recessive, Autosomal dominant; Pulmonary fibrosis and/or bone marrow failure, telomere-related, 3, 616373 (3), Autosomal dominant

RTN2	603183	Spastic paraplegia 12, autosomal dominant, 604805 (3), Autosomal dominant
RTN4IP1	610502	Optic atrophy 10 with or without ataxia, mental retardation, and seizures, 616732 (3), Autosomal recessive
RTN4R	605566	{Schizophrenia, susceptibility to}, 181500 (3), Autosomal dominant
RTTN	610436	Microcephaly, short stature, and polymicrogyria with seizures, 614833 (3), Autosomal recessive
RUBCN	613516	?Spinocerebellar ataxia, autosomal recessive 15, 615705 (3), Autosomal recessive
RUNX2	600211	Cleidocranial dysplasia, 119600 (3), Autosomal dominant; Cleidocranial dysplasia, forme fruste, dental anomalies only, 119600 (3), Autosomal dominant; Cleidocranial dysplasia, forme fruste, with brachydactyly, 119600 (3), Autosomal dominant; Metaphyseal dysplasia with maxillary hypoplasia with or without brachydactyly, 156510 (3), Autosomal dominant
RXFP2	606655	No OMIM phenotype
RXYLT1 (TMEM5)	605862	Muscular dystrophy-dystroglycanopathy (congenital with brain and eye anomalies), type A, 10, 615041 (3), Autosomal recessive
RYR1	180901	Central core disease, 117000 (3), Autosomal recessive, Autosomal dominant; King-Denborough syndrome, 145600 (3), Autosomal dominant; {Malignant hyperthermia susceptibility 1}, 145600 (3), Autosomal dominant; Minicore myopathy with external ophthalmoplegia, 255320 (3), Autosomal recessive; Neuromuscular disease, congenital, with uniform type 1 fiber, 117000 (3), Autosomal recessive, Autosomal dominant
RYR2	180902	Arrhythmogenic right ventricular dysplasia 2, 600996 (3), Autosomal dominant; Ventricular tachycardia, catecholaminergic polymorphic, 1, 604772 (3), Autosomal dominant
S1PR2	605111	Deafness, autosomal recessive 68, 610419 (3), Autosomal recessive
SAG	181031	Oguchi disease-1, 258100 (3), Autosomal recessive; Retinitis pigmentosa 47, 613758 (3)
SALL1	602218	Townes-Brocks branchiootorenal-like syndrome, 107480 (3), Autosomal dominant; Townes-Brocks syndrome 1, 107480 (3), Autosomal dominant
SALL2	602219	?Coloboma, ocular, autosomal recessive, 216820 (3), Autosomal recessive
SALL4	607343	Duane-radial ray syndrome, 607323 (3), Autosomal dominant; IVIC syndrome, 147750 (3), Autosomal dominant
SAMD9	610456	MIRAGE syndrome, 617053 (3), Autosomal dominant; Tumoral calcinosis, familial, normophosphatemic, 610455 (3), Autosomal recessive
SAMHD1	606754	Aicardi-Goutieres syndrome 5, 612952 (3), Autosomal recessive; ?Chilblain lupus 2, 614415 (3), Autosomal dominant
SAR1B	607690	Chylomicron retention disease, 246700 (3), Autosomal recessive
SARS2	612804	Hyperuricemia, pulmonary hypertension, renal failure, and alkalosis, 613845 (3), Autosomal recessive

SASS6	609321	?Microcephaly 14, primary, autosomal recessive, 616402 (3), Autosomal recessive
SAT1	313020	No OMIM phenotype
SBDS	607444	{Aplastic anemia, susceptibility to}, 609135 (3); Shwachman-Diamond syndrome, 260400 (3), Autosomal recessive
SBF1	603560	Charcot-Marie-Tooth disease, type 4B3, 615284 (3), Autosomal recessive
SBF2	607697	Charcot-Marie-Tooth disease, type 4B2, 604563 (3), Autosomal recessive
SCARB1	601040	[High density lipoprotein cholesterol level QTL6], 610762 (3)
SCARB2	602257	Epilepsy, progressive myoclonic 4, with or without renal failure, 254900 (3), Autosomal recessive
SCARF2	613619	Van den Ende-Gupta syndrome, 600920 (3), Autosomal recessive
SCGB3A2	606531	{Asthma, susceptibility to}, 600807 (3), Autosomal dominant
SCN10A	604427	Episodic pain syndrome, familial, 2, 615551 (3), Autosomal dominant
SCN11A	604385	Episodic pain syndrome, familial, 3, 615552 (3), Autosomal dominant; Neuropathy, hereditary sensory and autonomic, type VII, 615548 (3), Autosomal dominant
SCN1B	600235	Atrial fibrillation, familial, 13, 615377 (3), Autosomal dominant; Brugada syndrome 5, 612838 (3); Cardiac conduction defect, nonspecific, 612838 (3); Epilepsy, generalized, with febrile seizures plus, type 1, 604233 (3), Autosomal dominant; Epileptic encephalopathy, early infantile, 52, 617350 (3), Autosomal recessive
SCN2A	182390	Epileptic encephalopathy, early infantile, 11, 613721 (3), Autosomal dominant; Seizures, benign familial infantile, 3, 607745 (3), Autosomal dominant
SCN2B	601327	Atrial fibrillation, familial, 14, 615378 (3), Autosomal dominant
SCN3B	608214	Atrial fibrillation, familial, 16, 613120 (3), Autosomal dominant; Brugada syndrome 7, 613120 (3), Autosomal dominant
SCN4A	603967	Hyperkalemic periodic paralysis, type 2, 170500 (3), Autosomal dominant; Hypokalemic periodic paralysis, type 2, 613345 (3), Autosomal dominant; Myasthenic syndrome, congenital, 16, 614198 (3), Autosomal recessive; Myotonia congenita, atypical, acetazolamide-responsive, 608390 (3), Autosomal dominant; Paramyotonia congenita, 168300 (3), Autosomal dominant
SCN4B	608256	Atrial fibrillation, familial, 17, 611819 (3), Autosomal dominant; Long QT syndrome-10, 611819 (3), Autosomal dominant

SCN5A	600163	Atrial fibrillation, familial, 10, 614022 (3), Autosomal dominant; Brugada syndrome 1, 601144 (3), Autosomal dominant; Cardiomyopathy, dilated, 1E, 601154 (3), Autosomal dominant; Heart block, nonprogressive, 113900 (3), Autosomal dominant; Heart block, progressive, type IA, 113900 (3), Autosomal dominant; Long QT syndrome-3, 603830 (3), Autosomal dominant; Sick sinus syndrome 1, 608567 (3), Autosomal recessive; {Sudden infant death syndrome, susceptibility to}, 272120 (3), Autosomal recessive; Ventricular fibrillation, familial, 1, 603829 (3)
SCN8A	600702	?Cognitive impairment with or without cerebellar ataxia, 614306 (3), Autosomal dominant; Epileptic encephalopathy, early infantile, 13, 614558 (3), Autosomal dominant; Seizures, benign familial infantile, 5, 617080 (3), Autosomal dominant
SCNN1A	600228	Bronchiectasis with or without elevated sweat chloride 2, 613021 (3), Autosomal dominant; Pseudohypoaldosteronism, type I, 264350 (3), Autosomal recessive
SCNN1B	600760	Bronchiectasis with or without elevated sweat chloride 1, 211400 (3), Autosomal dominant; Liddle syndrome, 177200 (3), Autosomal dominant; Pseudohypoaldosteronism, type I, 264350 (3), Autosomal recessive
SCNN1G	600761	Bronchiectasis with or without elevated sweat chloride 3, 613071 (3), Autosomal dominant; Liddle syndrome, 177200 (3), Autosomal dominant; Pseudohypoaldosteronism, type I, 264350 (3), Autosomal recessive
SCO1	603644	Mitochondrial complex IV deficiency, 220110 (3), Autosomal recessive, Mitochondrial
SCO2	604272	Cardioencephalomyopathy, fatal infantile, due to cytochrome c oxidase deficiency 1, 604377 (3), Autosomal recessive; Myopia 6, 608908 (3), Autosomal dominant
SCP2	184755	?Leukoencephalopathy with dystonia and motor neuropathy, 613724 (3), Autosomal recessive
SCYL1	607982	Spinocerebellar ataxia, autosomal recessive 21, 616719 (3), Autosomal recessive
SDCCAG8	613524	Bardet-Biedl syndrome 16, 615993 (3), Autosomal recessive; Senior-Loken syndrome 7, 613615 (3)
SDHA	600857	Cardiomyopathy, dilated, 1GG, 613642 (3); Leigh syndrome, 256000 (3), Autosomal recessive, Mitochondrial; Mitochondrial respiratory chain complex II deficiency, 252011 (3), Autosomal recessive; Paragangliomas 5, 614165 (3), Autosomal dominant
SDHAF1	612848	Mitochondrial complex II deficiency, 252011 (3), Autosomal recessive
SDHAF2	613019	Paragangliomas 2, 601650 (3), Autosomal dominant

SDHB	185470	Cowden syndrome 2, 612359 (3), Autosomal dominant; Gastrointestinal stromal tumor, 606764 (3), Autosomal dominant, Isolated cases; Paraganglioma and gastric stromal sarcoma, 606864 (3); Paragangliomas 4, 115310 (3), Autosomal dominant; Pheochromocytoma, 171300 (3), Autosomal dominant
SDHC	602413	Gastrointestinal stromal tumor, 606764 (3), Autosomal dominant, Isolated cases; Paraganglioma and gastric stromal sarcoma, 606864 (3); Paragangliomas 3, 605373 (3), Autosomal dominant
SDHD	602690	Carcinoid tumors, intestinal, 114900 (3), Autosomal dominant; Cowden syndrome 3, 615106 (3); Merkel cell carcinoma, somatic (3); Mitochondrial complex II deficiency, 252011 (3), Autosomal recessive; Paraganglioma and gastric stromal sarcoma, 606864 (3); Paragangliomas 1, with or without deafness, 168000 (3), Autosomal dominant; Pheochromocytoma, 171300 (3), Autosomal dominant
SEC23A	610511	Craniolenticulosutural dysplasia, 607812 (3), Autosomal recessive
SEC24D	607186	Cole-Carpenter syndrome 2, 616294 (3), Autosomal recessive
SEC61A1	609213	Hyperuricemic nephropathy, familial juvenile, 4, 617056 (3), Autosomal dominant
SEC63	608648	Polycystic liver disease 2, 617004 (3), Autosomal dominant
SECISBP2	607693	Thyroid hormone metabolism, abnormal, 609698 (3)
SELE	131210	No OMIM phenotype
SELENON	606210	Muscular dystrophy, rigid spine, 1, 602771 (3), Autosomal recessive; Myopathy, congenital, with fiber-type disproportion, 255310 (3), Autosomal recessive, Autosomal dominant
SELP	173610	No OMIM phenotype
SEMA3A	603961	{Hypogonadotropic hypogonadism 16 with or without anosmia}, 614897 (3), Autosomal dominant
SEMA3E	608166	?CHARGE syndrome, 214800 (3), Autosomal dominant
SEMA7A	607961	[Blood group, John-Milton-Hagen system], 614745 (3)
SEPSECS	613009	Pontocerebellar hypoplasia type 2D, 613811 (3), Autosomal recessive
SEPT12	611562	Spermatogenic failure 10, 614822 (3), Autosomal dominant
SEPT9	604061	Amyotrophy, hereditary neuralgic, 162100 (3), Autosomal dominant; Leukemia, acute myeloid, therapy-related (1); Ovarian carcinoma (1)
SERAC1	614725	3-methylglutaconic aciduria with deafness, encephalopathy, and Leigh-like syndrome, 614739 (3), Autosomal recessive
SERPINA1	107400	Emphysema due to AAT deficiency, 613490 (3), Autosomal recessive; Emphysema-cirrhosis, due to AAT deficiency, 613490 (3), Autosomal recessive; Hemorrhagic diathesis due to antithrombin Pittsburgh, 613490 (3), Autosomal recessive; {Pulmonary disease, chronic obstructive, susceptibility to}, 606963 (1)
SERPINA6	122500	Corticosteroid-binding globulin deficiency, 611489 (3), Autosomal recessive, Autosomal dominant
SERPINA7	314200	[Thyroxine-binding globulin QTL], 300932 (3)

SERPINB7	603357	Palmoplantar keratoderma, Nagashima type, 615598 (3), Autosomal recessive
SERPINB8	601697	Peeling skin syndrome 5, 617115 (3), Autosomal recessive
SERPINC1	107300	Thrombophilia due to antithrombin III deficiency, 613118 (3), Autosomal recessive, Autosomal dominant
SERPIND1	142360	Thrombophilia due to heparin cofactor II deficiency, 612356 (3), Autosomal dominant
SERPINE1	173360	Plasminogen activator inhibitor-1 deficiency, 613329 (3), Autosomal recessive, Autosomal dominant; {Transcription of plasminogen activator inhibitor, modulator of} (3)
SERPINF1	172860	Osteogenesis imperfecta, type VI, 613982 (3)
SERPINI1	602445	Encephalopathy, familial, with neuroserpin inclusion bodies, 604218 (3), Autosomal dominant
SETBP1	611060	Mental retardation, autosomal dominant 29, 616078 (3), Autosomal dominant; Schinzel-Giedion midface retraction syndrome, 269150 (3), Autosomal dominant
SETD2	612778	Luscan-Lumish syndrome, 616831 (3), Autosomal dominant
SETX	608465	Amyotrophic lateral sclerosis 4, juvenile, 602433 (3), Autosomal dominant; Spinocerebellar ataxia, autosomal recessive 1, 606002 (3), Autosomal recessive
SF3B1	605590	Myelodysplastic syndrome, somatic, 614286 (3)
SF3B4	605593	Acrofacial dysostosis 1, Nager type, 154400 (3), Autosomal dominant
SFRP4	606570	Pyle disease, 265900 (3), Autosomal recessive
SFTPA1	178630	No OMIM phenotype
SFTPA2	178642	Pulmonary fibrosis, idiopathic, 178500 (3), Autosomal dominant
SFTPC	178620	Surfactant metabolism dysfunction, pulmonary, 2, 610913 (3), Autosomal dominant
SFXN4	615564	Combined oxidative phosphorylation deficiency 18, 615578 (3), Autosomal recessive
SGCA	600119	Muscular dystrophy, limb-girdle, type 2D, 608099 (3), Autosomal recessive
SGCB	600900	Muscular dystrophy, limb-girdle, type 2E, 604286 (3), Autosomal recessive
SGCD	601411	Cardiomyopathy, dilated, 1L, 606685 (3); Muscular dystrophy, limb-girdle, type 2F, 601287 (3), Autosomal recessive
SGCE	604149	Dystonia-11, myoclonic, 159900 (3), Autosomal dominant
SGCG	608896	Muscular dystrophy, limb-girdle, type 2C, 253700 (3), Autosomal recessive
SGO1	609168	Chronic atrial and intestinal dysrhythmia, 616201 (3), Autosomal recessive
SGSH	605270	Mucopolysaccharidosis type IIIA (Sanfilippo A), 252900 (3), Autosomal recessive
SH2B3	605093	Erythrocytosis, somatic, 133100 (3); Myelofibrosis, somatic, 254450 (3); Thrombocythemia, somatic, 187950 (3)

SH2D1A	300490	Lymphoproliferative syndrome, X-linked, 1, 308240 (3), X-linked recessive
SH3BP2	602104	Cherubism, 118400 (3), Autosomal dominant
SH3PXD2B	613293	Frank-ter Haar syndrome, 249420 (3), Autosomal recessive
SH3TC2	608206	Charcot-Marie-Tooth disease, type 4C, 601596 (3), Autosomal recessive; Mononeuropathy of the median nerve, mild, 613353 (3), Autosomal dominant
SHANK2	603290	{Autism susceptibility 17}, 613436 (3)
SHANK3	606230	Phelan-McDermid syndrome, 606232 (3), Autosomal dominant; {Schizophrenia 15}, 613950 (3)
SHH	600725	Holoprosencephaly 3, 142945 (3), Autosomal dominant; Microphthalmia with coloboma 5, 611638 (3), Autosomal dominant; Schizencephaly, 269160 (3); Single median maxillary central incisor, 147250 (3), Autosomal dominant
SHOC2	602775	Noonan-like syndrome with loose anagen hair, 607721 (3), Autosomal dominant
SHPK	605060	[Sedoheptulokinase deficiency], 617213 (3), Autosomal recessive
SHROOM4	300579	Stocco dos Santos X-linked mental retardation syndrome, 300434 (3), X-linked
SI	609845	Sucrase-isomaltase deficiency, congenital, 222900 (3), Autosomal recessive
SIAE	610079	{Autoimmune disease, susceptibility to, 6}, 613551 (3)
SIGMAR1	601978	?Amyotrophic lateral sclerosis 16, juvenile, 614373 (3), Autosomal recessive; ?Spinal muscular atrophy, distal, autosomal recessive, 2, 605726 (3), Autosomal recessive
SIK1	605705	Epileptic encephalopathy, early infantile, 30, 616341 (3), Autosomal dominant
SIL1	608005	Marinesco-Sjogren syndrome, 248800 (3), Autosomal recessive
SIM1	603128	Obesity, severe, 601665 (3), Autosomal recessive, Autosomal dominant, Multifactorial
SIN3A	607776	Witteveen-Kolk syndrome, 613406 (3), Autosomal dominant
SIPA1L3	616655	?Cataract 45, 616851 (3), Autosomal recessive
SIX1	601205	Branchiootic syndrome 3, 608389 (3), Autosomal dominant; Deafness, autosomal dominant 23, 605192 (3), Autosomal dominant
SIX3	603714	Holoprosencephaly 2, 157170 (3), Autosomal dominant; Schizencephaly, 269160 (3)
SIX5	600963	Branchiootorenal syndrome 2, 610896 (3)
SIX6	606326	Optic disc anomalies with retinal and/or macular dystrophy, 212550 (3), Autosomal recessive
SKI	164780	Shprintzen-Goldberg syndrome, 182212 (3), Autosomal dominant
SKIV2L	600478	Trichohepatoenteric syndrome 2, 614602 (3), Autosomal recessive
SLC10A2	601295	Bile acid malabsorption, primary, 613291 (3), Autosomal recessive
SLC11A1	600266	{Buruli ulcer, susceptibility to}, 610446 (3); {Mycobacterium tuberculosis, susceptibility to infection by}, 607948 (3)
SLC12A1	600839	Bartter syndrome, type 1, 601678 (3), Autosomal recessive

SLC12A3	600968	Gitelman syndrome, 263800 (3), Autosomal recessive
SLC12A5	606726	{Epilepsy, idiopathic generalized, susceptibility to, 14}, 616685 (3), Autosomal dominant; Epileptic encephalopathy, early infantile, 34, 616645 (3), Autosomal recessive
SLC12A6	604878	Agenesis of the corpus callosum with peripheral neuropathy, 218000 (3), Autosomal recessive
SLC13A5	608305	Epileptic encephalopathy, early infantile, 25, 615905 (3), Autosomal recessive
SLC14A1	613868	[Blood group, Kidd], 111000 (3)
SLC16A1	600682	Erythrocyte lactate transporter defect, 245340 (3), Autosomal dominant; Hyperinsulinemic hypoglycemia, familial, 7, 610021 (3), Autosomal dominant; Monocarboxylate transporter 1 deficiency, 616095 (3), Autosomal recessive, Autosomal dominant
SLC16A12	611910	Cataract 47, juvenile, with microcornea, 612018 (3), Autosomal dominant
SLC16A2	300095	Allan-Herndon-Dudley syndrome, 300523 (3), X-linked
SLC17A3	611034	{Gout susceptibility 4}, 612671 (3), Autosomal dominant; [Uric acid concentration, serum, QTL4], 612671 (3), Autosomal dominant
SLC17A5	604322	Salla disease, 604369 (3), Autosomal recessive; Sialic acid storage disorder, infantile, 269920 (3), Autosomal recessive
SLC17A8	607557	Deafness, autosomal dominant 25, 605583 (3), Autosomal dominant
SLC17A9	612107	Porokeratosis 8, disseminated superficial actinic type, 616063 (3), Autosomal dominant
SLC18A3	600336	Myasthenic syndrome, congenital, 21, presynaptic, 617239 (3), Autosomal recessive
SLC19A2	603941	Thiamine-responsive megaloblastic anemia syndrome, 249270 (3), Autosomal recessive
SLC19A3	606152	Thiamine metabolism dysfunction syndrome 2 (biotin- or thiamine-responsive encephalopathy type 2), 607483 (3), Autosomal recessive
SLC1A1	133550	Dicarboxylic aminoaciduria, 222730 (3), Autosomal recessive; {?Schizophrenia susceptibility 18}, 615232 (3)
SLC1A2	600300	Epileptic encephalopathy, early infantile, 41, 617105 (3), Autosomal dominant
SLC1A3	600111	Episodic ataxia, type 6, 612656 (3), Autosomal dominant
SLC1A4	600229	Spastic tetraplegia, thin corpus callosum, and progressive microcephaly, 616657 (3), Autosomal recessive
SLC22A12	607096	Hypouricemia, renal, 220150 (3), Autosomal recessive
SLC22A18	602631	Breast cancer, somatic, 114480 (3); Lung cancer, somatic, 211980 (3); Rhabdomyosarcoma, somatic, 268210 (3)
SLC22A4	604190	{Rheumatoid arthritis, susceptibility to}, 180300 (3)
SLC22A5	603377	Carnitine deficiency, systemic primary, 212140 (3), Autosomal recessive

SLC24A4	609840	Amelogenesis imperfecta, type IIA5, 615887 (3), Autosomal recessive; [Skin/hair/eye pigmentation 6, blond/brown hair], 210750 (3), Autosomal recessive; [Skin/hair/eye pigmentation 6, blue/green eyes], 210750 (3), Autosomal recessive
SLC24A5	609802	Albinism, oculocutaneous, type VI, 113750 (3), Autosomal recessive; [Skin/hair/eye pigmentation 4, fair/dark skin], 113750 (3), Autosomal recessive
SLC25A1	190315	Combined D-2- and L-2-hydroxyglutaric aciduria, 615182 (3), Autosomal recessive
SLC25A12	603667	Epileptic encephalopathy, early infantile, 39, 612949 (3), Autosomal recessive
SLC25A13	603859	Citrullinemia, adult-onset type II, 603471 (3), Autosomal recessive; Citrullinemia, type II, neonatal-onset, 605814 (3), Autosomal recessive
SLC25A15	603861	Hyperornithinemia-hyperammonemia-homocitrullinemia syndrome, 238970 (3), Autosomal recessive
SLC25A19	606521	Microcephaly, Amish type, 607196 (3), Autosomal recessive; Thiamine metabolism dysfunction syndrome 4 (progressive polyneuropathy type), 613710 (3), Autosomal recessive
SLC25A20	613698	Carnitine-acylcarnitine translocase deficiency, 212138 (3), Autosomal recessive
SLC25A22	609302	Epileptic encephalopathy, early infantile, 3, 609304 (3), Autosomal recessive
SLC25A26	611037	Combined oxidative phosphorylation deficiency 28, 616794 (3), Autosomal recessive
SLC25A3	600370	Mitochondrial phosphate carrier deficiency, 610773 (3)
SLC25A32	610815	?Exercise intolerance, riboflavin-responsive, 616839 (3), Autosomal recessive
SLC25A38	610819	Anemia, sideroblastic, 2, pyridoxine-refractory, 205950 (3), Autosomal recessive
SLC25A4	103220	Mitochondrial DNA depletion syndrome 12A (cardiomyopathic type) AD, 617184 (3), Autosomal dominant; Mitochondrial DNA depletion syndrome 12B (cardiomyopathic type) AR, 615418 (3), Autosomal recessive; Progressive external ophthalmoplegia with mitochondrial DNA deletions, autosomal dominant 2, 609283 (3), Autosomal dominant
SLC25A46	610826	Neuropathy, hereditary motor and sensory, type VIB, 616505 (3), Autosomal recessive
SLC26A1	610130	?Nephrolithiasis, calcium oxalate, 167030 (3), Autosomal recessive
SLC26A2	606718	Achondrogenesis Ib, 600972 (3), Autosomal recessive; Atelosteogenesis, type II, 256050 (3), Autosomal recessive; De la Chapelle dysplasia, 256050 (3), Autosomal recessive; Diastrophic dysplasia, 222600 (3), Autosomal recessive; Diastrophic dysplasia, broad bone-platyspondylic variant, 222600 (3), Autosomal recessive; Epiphyseal dysplasia, multiple, 4, 226900 (3), Autosomal recessive

SLC26A3	126650	Diarrhea 1, secretory chloride, congenital, 214700 (3), Autosomal recessive
SLC26A4	605646	Deafness, autosomal recessive 4, with enlarged vestibular aqueduct, 600791 (3), Autosomal recessive; Pendred syndrome, 274600 (3), Autosomal recessive
SLC26A5	604943	?Deafness, autosomal recessive 61, 613865 (3), Autosomal recessive
SLC26A8	608480	Spermatogenic failure 3, 606766 (3), Autosomal dominant
SLC27A4	604194	Ichthyosis prematurity syndrome, 608649 (3)
SLC29A3	612373	Histiocytosis-lymphadenopathy plus syndrome, 602782 (3), Autosomal recessive
SLC2A1	138140	Dystonia 9, 601042 (3), Autosomal dominant; {Epilepsy, idiopathic generalized, susceptibility to, 12}, 614847 (3), Autosomal dominant; GLUT1 deficiency syndrome 1, infantile onset, severe, 606777 (3), Autosomal recessive, Autosomal dominant; GLUT1 deficiency syndrome 2, childhood onset, 612126 (3), Autosomal dominant; Stomatin-deficient cryohydrocytosis with neurologic defects, 608885 (3), Autosomal dominant
SLC2A10	606145	Arterial tortuosity syndrome, 208050 (3), Autosomal recessive
SLC2A2	138160	{Diabetes mellitus, noninsulin-dependent}, 125853 (3), Autosomal dominant; Fanconi-Bickel syndrome, 227810 (3), Autosomal recessive
SLC2A9	606142	Hypouricemia, renal, 2, 612076 (3), Autosomal recessive, Autosomal dominant; {Uric acid concentration, serum, QTL 2}, 612076 (3), Autosomal recessive, Autosomal dominant
SLC30A10	611146	Hypermanganesemia with dystonia 1, 613280 (3), Autosomal recessive
SLC30A2	609617	Zinc deficiency, transient neonatal, 608118 (3), Autosomal dominant
SLC30A8	611145	{Diabetes mellitus, noninsulin-dependent, susceptibility to}, 125853 (3), Autosomal dominant
SLC33A1	603690	Congenital cataracts, hearing loss, and neurodegeneration, 614482 (3), Autosomal recessive; Spastic paraplegia 42, autosomal dominant, 612539 (3), Autosomal dominant
SLC34A1	182309	?Fanconi renotubular syndrome 2, 613388 (3), Autosomal recessive; Hypercalcemia, infantile, 2, 616963 (3), Autosomal recessive; Nephrolithiasis/osteoporosis, hypophosphatemic, 1, 612286 (3), Autosomal dominant
SLC34A3	609826	Hypophosphatemic rickets with hypercalciuria, 241530 (3), Autosomal recessive
SLC35A1	605634	Congenital disorder of glycosylation, type IIf, 603585 (3), Autosomal recessive
SLC35A2	314375	Congenital disorder of glycosylation, type IIm, 300896 (3), X-linked dominant, Somatic mosaicism
SLC35A3	605632	?Arthrogryposis, mental retardation, and seizures, 615553 (3), Autosomal recessive
SLC35C1	605881	Congenital disorder of glycosylation, type IIc, 266265 (3), Autosomal recessive
SLC35D1	610804	Schneckenbecken dysplasia, 269250 (3), Autosomal recessive

SLC36A2	608331	Hyperglycinuria, 138500 (3), Autosomal dominant; Iminoglycinuria, digenic, 242600 (3), Autosomal recessive, Digenic recessive
SLC38A8	615585	Foveal hypoplasia 2, with or without optic nerve misrouting and/or anterior segment dysgenesis, 609218 (3), Autosomal recessive
SLC39A13	608735	Ehlers-Danlos syndrome, spondylodysplastic type, 3, 612350 (3), Autosomal recessive
SLC39A14	608736	Hypermanganesemia with dystonia 2, 617013 (3), Autosomal recessive; ?Hyperostosis cranalis interna, 144755 (3), Autosomal dominant
SLC39A4	607059	Acrodermatitis enteropathica, 201100 (3), Autosomal recessive
SLC39A5	608730	Myopia 24, autosomal dominant, 615946 (3), Autosomal dominant
SLC39A8	608732	Congenital disorder of glycosylation, type IIn, 616721 (3), Autosomal recessive
SLC3A1	104614	Cystinuria, 220100 (3), Autosomal recessive, Autosomal dominant
SLC40A1	604653	Hemochromatosis, type 4, 606069 (3), Autosomal dominant
SLC45A2	606202	Albinism, oculocutaneous, type IV, 606574 (3); [Skin/hair/eye pigmentation 5, black/nonblack hair], 227240 (3), Autosomal recessive; [Skin/hair/eye pigmentation 5, dark/fair skin], 227240 (3), Autosomal recessive; [Skin/hair/eye pigmentation 5, dark/light eyes], 227240 (3), Autosomal recessive
SLC46A1	611672	Folate malabsorption, hereditary, 229050 (3), Autosomal recessive
SLC4A1	109270	[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); Cryohydrocytosis, 185020 (3), Autosomal dominant; [Malaria, resistance to], 611162 (3); Ovalocytosis, SA type, 166900 (3), Autosomal dominant; Renal tubular acidosis, distal, AD, 179800 (3), Autosomal dominant; Renal tubular acidosis, distal, AR, 611590 (3), Autosomal recessive; Spherocytosis, type 4, 612653 (3), Autosomal dominant
SLC52A1	607883	Riboflavin deficiency, 615026 (3), Autosomal dominant
SLC52A2	607882	Brown-Vialetto-Van Laere syndrome 2, 614707 (3), Autosomal recessive
SLC52A3	613350	Brown-Vialetto-Van Laere syndrome 1, 211530 (3), Autosomal recessive; ?Fazio-Londe disease, 211500 (3), Autosomal recessive
SLC5A1	182380	Glucose/galactose malabsorption, 606824 (3), Autosomal recessive
SLC5A2	182381	Renal glucosuria, 233100 (3), Autosomal recessive, Autosomal dominant
SLC5A5	601843	Thyroid dyshormonogenesis 1, 274400 (3), Autosomal recessive
SLC5A7	608761	Myasthenic syndrome, congenital, 20, presynaptic, 617143 (3), Autosomal recessive; Neuronopathy, distal hereditary motor, type VIIA, 158580 (3), Autosomal dominant
SLC6A1	137165	Myoclonic-atonic epilepsy, 616421 (3), Autosomal dominant
SLC6A14	300444	{Obesity, susceptibility to, BMIQ11}, 300306 (3)
SLC6A17	610299	Mental retardation, autosomal recessive 48, 616269 (3), Autosomal recessive

SLC6A19	608893	Hartnup disorder, 234500 (3), Autosomal recessive; Hyperglycinuria, 138500 (3), Autosomal dominant; Iminoglycinuria, digenic, 242600 (3), Autosomal recessive, Digenic recessive
SLC6A2	163970	Orthostatic intolerance, 604715 (3)
SLC6A20	605616	Hyperglycinuria, 138500 (3), Autosomal dominant; Iminoglycinuria, digenic, 242600 (3), Autosomal recessive, Digenic recessive
SLC6A3	126455	{Nicotine dependence, protection against}, 188890 (3); Parkinsonism-dystonia, infantile, 613135 (3), Autosomal recessive
SLC6A4	182138	{Anxiety-related personality traits}, 607834 (3); {Obsessive-compulsive disorder}, 164230 (3), Autosomal dominant
SLC6A5	604159	Hyperekplexia 3, 614618 (3), Autosomal recessive, Autosomal dominant
SLC6A8	300036	Cerebral creatine deficiency syndrome 1, 300352 (3), X-linked recessive
SLC7A14	615720	Retinitis pigmentosa 68, 615725 (3), Autosomal recessive
SLC7A7	603593	Lysinuric protein intolerance, 222700 (3), Autosomal recessive
SLC7A9	604144	Cystinuria, 220100 (3), Autosomal recessive, Autosomal dominant
SLC9A1	107310	?Lichtenstein-Knorr syndrome, 616291 (3), Autosomal recessive
SLC9A3	182307	Diarrhea 8, secretory sodium, congenital, 616868 (3), Autosomal recessive
SLC9A3R1	604990	Nephrolithiasis/osteoporosis, hypophosphatemic, 2, 612287 (3), Autosomal dominant
SLC9A9	608396	{?Autism susceptibility 16}, 613410 (3)
SLCO1B1	604843	Hyperbilirubinemia, Rotor type, digenic, 237450 (3), Digenic recessive
SLCO2A1	601460	Hypertrophic osteoarthropathy, primary, autosomal recessive 2, 614441 (3), Autosomal recessive
SLFN14	614958	Bleeding disorder, platelet-type, 20, 616913 (3), Autosomal dominant
SLITRK1	609678	Tourette syndrome, 137580 (3), Autosomal dominant; ?Trichotillomania, 613229 (3), Autosomal dominant, Multifactorial
SLITRK6	609681	Deafness and myopia, 221200 (3), Autosomal recessive
SLURP1	606119	Meleda disease, 248300 (3), Autosomal recessive
SLX4	613278	Fanconi anemia, complementation group P, 613951 (3), Autosomal recessive
SMAD3	603109	Loeys-Dietz syndrome 3, 613795 (3), Autosomal dominant
SMAD4	600993	Juvenile polyposis/hereditary hemorrhagic telangiectasia syndrome, 175050 (3), Autosomal dominant; Myhre syndrome, 139210 (3), Autosomal dominant; Pancreatic cancer, somatic, 260350 (3); Polyposis, juvenile intestinal, 174900 (3), Autosomal dominant
SMAD6	602931	Aortic valve disease 2, 614823 (3), Autosomal dominant; {Craniosynostosis 7, susceptibility to}, 617439 (3), Autosomal dominant
SMAD7	602932	{Colorectal cancer, susceptibility to, 3}, 612229 (3)
SMAD9	603295	Pulmonary hypertension, primary, 2, 615342 (3), Autosomal dominant
SMARCA2	600014	Nicolaides-Baraitser syndrome, 601358 (3), Autosomal dominant

SMARCA4	603254	Coffin-Siris syndrome 4, 614609 (3), Autosomal dominant; {Rhabdoid tumor predisposition syndrome 2}, 613325 (3), Autosomal dominant
SMARCAD1	612761	Adermatoglyphia, 136000 (3), Autosomal dominant; Basan syndrome, 129200 (3), Autosomal dominant
SMARCAL1	606622	Schimke immunoosseous dysplasia, 242900 (3), Autosomal recessive
SMARCB1	601607	Coffin-Siris syndrome 3, 614608 (3), Autosomal dominant; {Rhabdoid tumor predisposition syndrome 1}, 609322 (3), Autosomal dominant; Rhabdoid tumors, somatic, 609322 (3); {Schwannomatosis-1, susceptibility to}, 162091 (3), Autosomal dominant
SMARCE1	603111	Coffin-Siris syndrome 5, 616938 (3), Autosomal dominant; {Meningioma, familial, susceptibility to}, 607174 (3), Autosomal dominant
SMC1A	300040	Cornelia de Lange syndrome 2, 300590 (3), X-linked dominant
SMC3	606062	Cornelia de Lange syndrome 3, 610759 (3), Autosomal dominant
SMCHD1	614982	Bosma arhinia microphthalmia syndrome, 603457 (3), Autosomal dominant; Fascioscapulohumeral muscular dystrophy 2, digenic, 158901 (3)
SMG9	613176	Heart and brain malformation syndrome, 616920 (3), Autosomal recessive
SMIM1	615242	[Blood group, Vel system], 615264 (3), Autosomal recessive
SMN1	600354	Spinal muscular atrophy-1, 253300 (3), Autosomal recessive; Spinal muscular atrophy-2, 253550 (3), Autosomal recessive; Spinal muscular atrophy-3, 253400 (3), Autosomal recessive; Spinal muscular atrophy-4, 271150 (3), Autosomal recessive
SMN2	601627	{Spinal muscular atrophy, type III, modifier of}, 253400 (3), Autosomal recessive
SMO	601500	Basal cell carcinoma, somatic, 605462 (3); Curry-Jones syndrome, somatic mosaic, 601707 (3)
SMOC1	608488	Microphthalmia with limb anomalies, 206920 (3), Autosomal recessive
SMOC2	607223	Dentin dysplasia, type I, with microdontia and misshapen teeth, 125400 (3), Autosomal recessive
SMPD1	607608	Niemann-Pick disease, type A, 257200 (3), Autosomal recessive; Niemann-Pick disease, type B, 607616 (3), Autosomal recessive
SMPX	300226	Deafness, X-linked 4, 300066 (3), X-linked dominant
SMS	300105	Mental retardation, X-linked, Snyder-Robinson type, 309583 (3), X-linked recessive
SNAI2	602150	Piebaldism, 172800 (3), Autosomal dominant; Waardenburg syndrome, type 2D, 608890 (3), Autosomal recessive
SNAP25	600322	?Myasthenic syndrome, congenital, 18, 616330 (3), Autosomal dominant
SNAP29	604202	Cerebral dysgenesis, neuropathy, ichthyosis, and palmoplantar keratoderma syndrome, 609528 (3), Autosomal recessive
SNCB	602569	Dementia, Lewy body, 127750 (3), Autosomal dominant
SNIP1	608241	Psychomotor retardation, epilepsy, and craniofacial dysmorphism, 614501 (3), Autosomal recessive

SNRNP200	601664	Retinitis pigmentosa 33, 610359 (3), Autosomal dominant
SNRPB	182282	Cerebrocostomandibular syndrome, 117650 (3), Autosomal dominant
SNRPE	128260	Hypotrichosis 11, 615059 (3), Autosomal dominant
SNRPN	182279	Prader-Willi syndrome, 176270 (3), Isolated cases
SNTA1	601017	Long QT syndrome 12, 612955 (3), Autosomal dominant
SNX10	614780	Osteopetrosis, autosomal recessive 8, 615085 (3), Autosomal recessive
SNX14	616105	Spinocerebellar ataxia, autosomal recessive 20, 616354 (3), Autosomal recessive
SOBP	613667	Mental retardation, anterior maxillary protrusion, and strabismus, 613671 (3), Autosomal recessive
SOD1	147450	Amyotrophic lateral sclerosis 1, 105400 (3), Autosomal recessive, Autosomal dominant
SOD2	147460	{Microvascular complications of diabetes 6}, 612634 (3)
SON	182465	ZTTK syndrome, 617140 (3), Autosomal dominant
SORL1	602005	No OMIM phenotype
SORT1	602458	[Low density lipoprotein cholesterol level QTL6], 613589 (3), Autosomal dominant
SOS1	182530	?Fibromatosis, gingival, 1, 135300 (3), Autosomal dominant; Noonan syndrome 4, 610733 (3), Autosomal dominant
SOS2	601247	Noonan syndrome 9, 616559 (3), Autosomal dominant
SOST	605740	Craniodiaphyseal dysplasia, autosomal dominant, 122860 (3), Autosomal dominant; Sclerosteosis 1, 269500 (3), Autosomal recessive; Van Buchem disease, 239100 (3), Autosomal recessive
SOX10	602229	PCWH syndrome, 609136 (3), Autosomal dominant; Waardenburg syndrome, type 2E, with or without neurologic involvement, 611584 (3), Autosomal dominant; Waardenburg syndrome, type 4C, 613266 (3), Autosomal dominant
SOX11	600898	Mental retardation, autosomal dominant 27, 615866 (3), Autosomal dominant
SOX17	610928	Vesicoureteral reflux 3, 613674 (3), Autosomal dominant
SOX18	601618	Hypotrichosis-lymphedema-telangiectasia syndrome, 607823 (3), Autosomal recessive; Hypotrichosis-lymphedema-telangiectasia-renal defect syndrome, 137940 (3), Autosomal dominant
SOX2	184429	Microphthalmia, syndromic 3, 206900 (3), Autosomal dominant; Optic nerve hypoplasia and abnormalities of the central nervous system, 206900 (3), Autosomal dominant
SOX3	313430	Mental retardation, X-linked, with isolated growth hormone deficiency, 300123 (3); Panhypopituitarism, X-linked, 312000 (3), X-linked
SOX5	604975	Lamb-Shaffer syndrome, 616803 (3), Autosomal dominant
SOX9	608160	Acampomelic campomelic dysplasia, 114290 (3), Autosomal dominant; Campomelic dysplasia, 114290 (3), Autosomal dominant; Campomelic dysplasia with autosomal sex reversal, 114290 (3), Autosomal dominant

SP110	604457	Hepatic venoocclusive disease with immunodeficiency, 235550 (3), Autosomal recessive; {Mycobacterium tuberculosis, susceptibility to}, 607948 (3)
SP7	606633	?Osteogenesis imperfecta, type XII, 613849 (3), Autosomal recessive
SPAG1	603395	Ciliary dyskinesia, primary, 28, 615505 (3), Autosomal recessive
SPARC	182120	Osteogenesis imperfecta, type XVII, 616507 (3), Autosomal recessive
SPART	607111	Troyer syndrome, 275900 (3), Autosomal recessive
SPATA16	609856	?Spermatogenic failure 6, 102530 (3), Autosomal recessive
SPATA5	613940	Epilepsy, hearing loss, and mental retardation syndrome, 616577 (3), Autosomal recessive
SPECC1L	614140	?Facial clefting, oblique, 1, 600251 (3), Autosomal dominant; Opitz GBBB syndrome, type II, 145410 (3), Autosomal dominant
SPEG	615950	Centronuclear myopathy 5, 615959 (3), Autosomal recessive
SPG11	610844	Amyotrophic lateral sclerosis 5, juvenile, 602099 (3), Autosomal recessive; Charcot-Marie-Tooth disease, axonal, type 2X, 616668 (3), Autosomal recessive; Spastic paraplegia 11, autosomal recessive, 604360 (3), Autosomal recessive
SPG21	608181	Mast syndrome, 248900 (3), Autosomal recessive
SPG7	602783	Spastic paraplegia 7, autosomal recessive, 607259 (3), Autosomal recessive, Autosomal dominant
SPINK1	167790	{Fibrocalculous pancreatic diabetes, susceptibility to}, 608189 (3), Autosomal recessive, Autosomal dominant; Pancreatitis, hereditary, 167800 (3), Autosomal dominant; Tropical calcific pancreatitis, 608189 (3), Autosomal recessive, Autosomal dominant
SPINK5	605010	Netherton syndrome, 256500 (3), Autosomal recessive
SPINT2	605124	Diarrhea 3, secretory sodium, congenital, syndromic, 270420 (3), Autosomal recessive
SPR	182125	Dystonia, dopa-responsive, due to sepiapterin reductase deficiency, 612716 (3), ?Autosomal dominant, Autosomal recessive
SPRED1	609291	Legius syndrome, 611431 (3), Autosomal dominant
SPRTN	616086	Ruijs-Aalfs syndrome, 616200 (3), Autosomal recessive
SPRY2	602466	{?IgA nephropathy, susceptibility to, 3}, 616818 (3), Autosomal dominant
SPRY4	607984	Hypogonadotropic hypogonadism 17 with or without anosmia, 615266 (3), Autosomal dominant
SPTA1	182860	Elliptocytosis-2, 130600 (3), Autosomal dominant; Pyropoikilocytosis, 266140 (3), Autosomal recessive; Spherocytosis, type 3, 270970 (3), Autosomal recessive
SPTAN1	182810	Epileptic encephalopathy, early infantile, 5, 613477 (3), Autosomal dominant
SPTB	182870	Anemia, neonatal hemolytic, fatal or near-fatal, 617948 (3); Elliptocytosis-3, 617948 (3); Spherocytosis, type 2, 616649 (3), Autosomal dominant
SPTLC1	605712	Neuropathy, hereditary sensory and autonomic, type IA, 162400 (3), Autosomal dominant

SPTLC2	605713	Neuropathy, hereditary sensory and autonomic, type IC, 613640 (3), Autosomal dominant
SRA1	603819	No OMIM phenotype
SRC	190090	Colon cancer, advanced, somatic, 114500 (3); ?Thrombocytopenia 6, 616937 (3), Autosomal dominant
SRCAP	611421	Floating-Harbor syndrome, 136140 (3), Autosomal dominant
SRD5A2	607306	Pseudovaginal perineoscrotal hypospadias, 264600 (3), Autosomal recessive
SRD5A3	611715	Congenital disorder of glycosylation, type Iq, 612379 (3), Autosomal recessive; Kahrizi syndrome, 612713 (3), Autosomal recessive
SRGAP1	606523	{Thyroid cancer, nonmedullary, 2}, 188470 (3), Autosomal dominant
SRP72	602122	Bone marrow failure syndrome 1, 614675 (3), Autosomal dominant
SRPX2	300642	?Rolandic epilepsy, mental retardation, and speech dyspraxia, 300643 (3)
SRY	480000	46XX sex reversal 1, 400045 (3); 46XY sex reversal 1, 400044 (3)
SSX1	312820	?Sarcoma, synovial, 300813 (3)
SSX2	300192	?Sarcoma, synovial, 300813 (3)
STAC3	615521	Native American myopathy, 255995 (3), Autosomal recessive
STAR	600617	Lipoid adrenal hyperplasia, 201710 (3), Autosomal recessive
STAT1	600555	Immunodeficiency 31A, mycobacteriosis, autosomal dominant, 614892 (3), Autosomal dominant; Immunodeficiency 31B, mycobacterial and viral infections, autosomal recessive, 613796 (3), Autosomal recessive; Immunodeficiency 31C, autosomal dominant, 614162 (3), Autosomal dominant
STAT2	600556	Immunodeficiency 44, 616636 (3), Autosomal recessive
STAT4	600558	{Systemic lupus erythematosus, susceptibility to, 11}, 612253 (3)
STAT5B	604260	Growth hormone insensitivity with immunodeficiency, 245590 (3); Leukemia, acute promyelocytic, somatic, 102578 (3)
STIL	181590	Microcephaly 7, primary, autosomal recessive, 612703 (3), Autosomal recessive
STIM1	605921	Immunodeficiency 10, 612783 (3), Autosomal recessive; Myopathy, tubular aggregate, 1, 160565 (3), Autosomal dominant; Stormorken syndrome, 185070 (3), Autosomal dominant
STK11	602216	Melanoma, malignant, somatic (3); Pancreatic cancer, 260350 (3), Autosomal dominant, Somatic mutation, Multifactorial; Peutz-Jeghers syndrome, 175200 (3), Autosomal dominant; Testicular tumor, somatic, 273300 (3)
STK4	604965	T-cell immunodeficiency, recurrent infections, autoimmunity, and cardiac malformations, 614868 (3)
STN1	613128	Cerebroretinal microangiopathy with calcifications and cysts 2, 617341 (3), Autosomal recessive
STOM	133090	No OMIM phenotype
STOX1	609397	Preeclampsia/eclampsia 4, 609404 (3)

STRA6	610745	Microphthalmia, isolated, with coloboma 8, 601186 (3), Autosomal recessive; Microphthalmia, syndromic 9, 601186 (3), Autosomal recessive
STRC	606440	Deafness, autosomal recessive 16, 603720 (3), Autosomal recessive
STS	300747	Ichthyosis, X-linked, 308100 (3), X-linked recessive
STT3A	601134	?Congenital disorder of glycosylation, type Iw, 615596 (3), Autosomal recessive
STT3B	608605	?Congenital disorder of glycosylation, type Ix, 615597 (3), Autosomal recessive
STUB1	607207	Spinocerebellar ataxia, autosomal recessive 16, 615768 (3), Autosomal recessive
STX11	605014	Hemophagocytic lymphohistiocytosis, familial, 4, 603552 (3), Autosomal recessive
STX16	603666	Pseudohypoparathyroidism, type IB, 603233 (3), Autosomal dominant
STX1B	601485	Generalized epilepsy with febrile seizures plus, type 9, 616172 (3), Autosomal dominant
STXBP1	602926	Epileptic encephalopathy, early infantile, 4, 612164 (3), Autosomal dominant
STXBP2	601717	Hemophagocytic lymphohistiocytosis, familial, 5, 613101 (3)
SUCLA2	603921	Mitochondrial DNA depletion syndrome 5 (encephalomyopathic with or without methylmalonic aciduria), 612073 (3), Autosomal recessive
SUCLG1	611224	Mitochondrial DNA depletion syndrome 9 (encephalomyopathic type with methylmalonic aciduria), 245400 (3), Autosomal recessive
SUFU	607035	Basal cell nevus syndrome, 109400 (3), Autosomal dominant; Joubert syndrome 32, 617757 (3), Autosomal recessive; Medulloblastoma, desmoplastic, 155255 (3), Autosomal recessive, Autosomal dominant; {Meningioma, familial, susceptibility to}, 607174 (3), Autosomal dominant
SUGCT	609187	Glutaric aciduria III, 231690 (3), Autosomal recessive
SUMF1	607939	Multiple sulfatase deficiency, 272200 (3), Autosomal recessive
SUMO1	601912	?Orofacial cleft 10, 613705 (3), Isolated cases
SUMO4	608829	{Diabetes mellitus, insulin-dependent, 5}, 600320 (3)
SUN5	613942	Spermatogenic failure 16, 617187 (3), Autosomal recessive
SURF1	185620	Charcot-Marie-Tooth disease, type 4K, 616684 (3), Autosomal recessive; Leigh syndrome, due to COX IV deficiency, 256000 (3), Autosomal recessive, Mitochondrial
SYCE1	611486	?Premature ovarian failure 12, 616947 (3), Autosomal recessive; ?Spermatogenic failure 15, 616950 (3), Autosomal recessive
SYN1	313440	Epilepsy, X-linked, with variable learning disabilities and behavior disorders, 300491 (3), X-linked recessive, X-linked dominant
SYN2	600755	{Schizophrenia, susceptibility to}, 181500 (3), Autosomal dominant
SYNE1	608441	Emery-Dreifuss muscular dystrophy 4, autosomal dominant, 612998 (3), Autosomal dominant; Spinocerebellar ataxia, autosomal recessive 8, 610743 (3), Autosomal recessive

SYNE2	608442	Emery-Dreifuss muscular dystrophy 5, autosomal dominant, 612999 (3), Autosomal dominant
SYNE4	615535	Deafness, autosomal recessive 76, 615540 (3), Autosomal recessive
SYNGAP1	603384	Mental retardation, autosomal dominant 5, 612621 (3), Autosomal dominant
SYNJ1	604297	Epileptic encephalopathy, early infantile, 53, 617389 (3), Autosomal recessive; Parkinson disease 20, early-onset, 615530 (3), Autosomal recessive
SYP	313475	Mental retardation, X-linked 96, 300802 (3), X-linked dominant
SZT2	615463	Epileptic encephalopathy, early infantile, 18, 615476 (3), Autosomal recessive
TAC3	162330	Hypogonadotropic hypogonadism 10 with or without anosmia, 614839 (3), Autosomal recessive
TACO1	612958	Mitochondrial complex IV deficiency, 220110 (3), Autosomal recessive, Mitochondrial
TACR3	162332	Hypogonadotropic hypogonadism 11 with or without anosmia, 614840 (3), Autosomal recessive
TACSTD2	137290	Corneal dystrophy, gelatinous drop-like, 204870 (3), Autosomal recessive
TAF1	313650	Dystonia-Parkinsonism, X-linked, 314250 (3), X-linked recessive; Mental retardation, X-linked, syndromic 33, 300966 (3), X-linked recessive
TAF15	601574	Chondrosarcoma, extraskeletal myxoid, 612237 (1)
TAF2	604912	Mental retardation, autosomal recessive 40, 615599 (3), Autosomal recessive
TAF4B	601689	?Spermatogenic failure 13, 615841 (3), Autosomal recessive
TAL1	187040	Leukemia, T-cell acute lymphocytic, somatic, 613065 (3)
TAL2	186855	Leukemia, T-cell acute lymphocytic, somatic, 613065 (3)
TALDO1	602063	Transaldolase deficiency, 606003 (3), Autosomal recessive
TANGO2	616830	Metabolic encephalomyopathic crises, recurrent, with rhabdomyolysis, cardiac arrhythmias, and neurodegeneration, 616878 (3), Autosomal recessive
TAP1	170260	Bare lymphocyte syndrome, type I, 604571 (3), Autosomal recessive
TAP2	170261	Bare lymphocyte syndrome, type I, due to TAP2 deficiency, 604571 (3), Autosomal recessive; Wegener-like granulomatosis (3)
TAPBP	601962	Bare lymphocyte syndrome, type I, 604571 (3), Autosomal recessive
TAPT1	612758	Osteochondrodysplasia, complex lethal, Symoens-Barnes-Gistelinck type, 616897 (3), Autosomal recessive
TARDBP	605078	Amyotrophic lateral sclerosis 10, with or without FTD, 612069 (3), Autosomal dominant; Frontotemporal lobar degeneration, TARDBP-related, 612069 (3), Autosomal dominant
TARS2	612805	?Combined oxidative phosphorylation deficiency 21, 615918 (3), Autosomal recessive
TAS2R16	604867	[Beta-glycopyranoside tasting], (3) {Alcohol dependence, susceptibility to}, 617956 (3)

TAS2R38	607751	[Phenylthiocarbamide tasting], 171200 (3), Autosomal dominant
TAT	613018	Tyrosinemia, type II, 276600 (3), Autosomal recessive
TAZ	300394	Barth syndrome, 302060 (3), X-linked recessive
TBC1D20	611663	Warburg micro syndrome 4, 615663 (3), Autosomal recessive
TBC1D24	613577	DOORS syndrome, 220500 (3), Autosomal recessive; Deafness , autosomal recessive 86, 614617 (3), Autosomal recessive; Deafness, autosomal dominant 65, 616044 (3), Autosomal dominant; Epileptic encephalopathy, early infantile, 16, 615338 (3), Autosomal recessive; Myoclonic epilepsy, infantile, familial, 605021 (3), Autosomal recessive
TBC1D4	612465	{Diabetes mellitus, noninsulin-dependent, 5}, 616087 (3)
TBCD	604649	Encephalopathy, progressive, early-onset, with brain atrophy and thin corpus callosum, 617193 (3), Autosomal recessive
TBCE	604934	Encephalopathy, progressive, with amyotrophy and optic atrophy, 617207 (3), Autosomal recessive; Hypoparathyroidism-retardation-dysmorphism syndrome, 241410 (3), Autosomal recessive; Kenny-Caffey syndrome, type 1, 244460 (3), Autosomal recessive
TBCK	616899	Hypotonia, infantile, with psychomotor retardation and characteristic facies 3, 616900 (3), Autosomal recessive
TBK1	604834	{Encephalopathy, acute, infection-induced (herpes-specific), susceptibility to, 8}, 617900 (3), Autosomal dominant; Frontotemporal dementia and/or amyotrophic lateral sclerosis 4, 616439 (3), Autosomal dominant
TBL1XR1	608628	Mental retardation, autosomal dominant 41, 616944 (3), Autosomal dominant; Pierpont syndrome, 602342 (3), Autosomal dominant
TBP	600075	{Parkinson disease, susceptibility to}, 168600 (3), Isolated cases, Multifactorial; Spinocerebellar ataxia 17, 607136 (3), Autosomal dominant
TBX1	602054	Conotruncal anomaly face syndrome, 217095 (3); DiGeorge syndrome, 188400 (3), Autosomal dominant; Tetralogy of Fallot, 187500 (3), Autosomal dominant; Velocardiofacial syndrome, 192430 (3), Autosomal dominant
TBX18	604613	Congenital anomalies of kidney and urinary tract 2, 143400 (3), Autosomal dominant
TBX19	604614	Adrenocorticotropic hormone deficiency, 201400 (3), Autosomal recessive
TBX20	606061	Atrial septal defect 4, 611363 (3)
TBX21	604895	Asthma and nasal polyps, 208550 (3), Autosomal recessive; {Asthma, aspirin-induced, susceptibility to}, 208550 (3), Autosomal recessive
TBX22	300307	?Abruzzo-Erickson syndrome, 302905 (3), X-linked; Cleft palate with ankyloglossia, 303400 (3), X-linked
TBX3	601621	Ulnar-mammary syndrome, 181450 (3), Autosomal dominant
TBX4	601719	Ischiocoxopodopatellar syndrome, 147891 (3), Autosomal dominant
TBX5	601620	Holt-Oram syndrome, 142900 (3), Autosomal dominant
TBX6	602427	Spondylocostal dysostosis 5, 122600 (3), Autosomal recessive, Autosomal dominant

TBXA2R	188070	{Bleeding disorder, platelet-type, 13, susceptibility to}, 614009 (3), Autosomal dominant
TBXAS1	274180	Ghosal hematodiaphyseal syndrome, 231095 (3), Autosomal recessive; ?Thromboxane synthase deficiency, 614158 (1), Autosomal dominant
TBXT (T)	601397	{Neural tube defects, susceptibility to}, 182940 (3), Autosomal dominant; Sacral agenesis with vertebral anomalies, 615709 (3), Autosomal recessive
TCAP	604488	Cardiomyopathy, hypertrophic, 25, 607487 (3), Autosomal dominant; Muscular dystrophy, limb-girdle, type 2G, 601954 (3), Autosomal recessive
TCHH	190370	?Uncombable hair syndrome 3, 617252 (3), Autosomal recessive
TCIRG1	604592	Osteopetrosis, autosomal recessive 1, 259700 (3), Autosomal recessive
TCN2	613441	Transcobalamin II deficiency, 275350 (3), Autosomal recessive
TCOF1	606847	Treacher Collins syndrome 1, 154500 (3), Autosomal dominant
TCTEX1D2	617353	Short-rib thoracic dysplasia 17 with or without polydactyly, 617405 (3), Autosomal recessive
TCTN1	609863	Joubert syndrome 13, 614173 (3), Autosomal recessive
TCTN2	613846	Joubert syndrome 24, 616654 (3), Autosomal recessive; ?Meckel syndrome 8, 613885 (3), Autosomal recessive
TCTN3	613847	Joubert syndrome 18, 614815 (3), Autosomal recessive; Orofaciodigital syndrome IV, 258860 (3), Autosomal recessive
TDP1	607198	Spinocerebellar ataxia, autosomal recessive with axonal neuropathy, 607250 (3)
TDP2	605764	Spinocerebellar ataxia, autosomal recessive 23, 616949 (3), Autosomal recessive
TDRD7	611258	Cataract 36, 613887 (3), Autosomal recessive
TEAD1	189967	Sveinsson chorioretinal atrophy, 108985 (3), Autosomal dominant
TEC	600583	No OMIM phenotype
TECPR2	615000	Spastic paraplegia 49, autosomal recessive, 615031 (3), Autosomal recessive
TECR	610057	Mental retardation, autosomal recessive 14, 614020 (3), Autosomal recessive
TECTA	602574	Deafness, autosomal dominant 8/12, 601543 (3), Autosomal dominant; Deafness, autosomal recessive 21, 603629 (3), Autosomal recessive
TEK	600221	Glaucoma 3, primary congenital, E, 617272 (3), Autosomal dominant; Venous malformations, multiple cutaneous and mucosal, 600195 (3), Autosomal dominant
TELO2	611140	You-Hoover-Fong syndrome, 616954 (3), Autosomal recessive
TENM3	610083	Microphthalmia, isolated, with coloboma 9, 615145 (3), Autosomal recessive
TENM4	610084	Essential tremor, hereditary, 5, 616736 (3), Autosomal dominant

TERT	187270	{Dyskeratosis congenita, autosomal dominant 2}, 613989 (3), Autosomal recessive, Autosomal dominant; {Dyskeratosis congenita, autosomal recessive 4}, 613989 (3), Autosomal recessive, Autosomal dominant; {Leukemia, acute myeloid}, 601626 (3), Autosomal dominant; {Melanoma, cutaneous malignant, 9}, 615134 (3); {Pulmonary fibrosis and/or bone marrow failure, telomere-related, 1}, 614742 (3), Autosomal dominant
TET2	612839	Myelodysplastic syndrome, somatic, 614286 (3)
TEX11	300311	Spermatogenic failure, X-linked, 2, 309120 (3), X-linked recessive
TF	190000	Atransferrinemia, 209300 (3), Autosomal recessive
TFAM	600438	?Mitochondrial DNA depletion syndrome 15 (hepatocerebral type), 617156 (3), Autosomal recessive
TFAP2A	107580	Branchiooculofacial syndrome, 113620 (3), Autosomal dominant
TFAP2B	601601	Char syndrome, 169100 (3), Autosomal dominant; Patent ductus arteriosus 2, 617035 (3), Autosomal dominant
TFE3	314310	Renal cell carcinoma, papillary, 1, 300854 (3)
TFG	602498	Hereditary motor and sensory neuropathy, Okinawa type, 604484 (3), Autosomal dominant; ?Spastic paraplegia 57, autosomal recessive, 615658 (3), Autosomal recessive
TFR2	604720	Hemochromatosis, type 3, 604250 (3), Autosomal recessive
TFRC	190010	Immunodeficiency 46, 616740 (3), Autosomal recessive
TG	188450	{Autoimmune thyroid disease, susceptibility to, 3}, 608175 (3); Thyroid dyshormonogenesis 3, 274700 (3), Autosomal recessive
TGDS	616146	Catel-Manzke syndrome, 616145 (3), Autosomal recessive
TGFB1	190180	Camurati-Engelmann disease, 131300 (3), Autosomal dominant; {Cystic fibrosis lung disease, modifier of}, 219700 (3), Autosomal recessive
TGFB2	190220	Loeys-Dietz syndrome 4, 614816 (3), Autosomal dominant
TGFB3	190230	Arrhythmogenic right ventricular dysplasia 1, 107970 (3), Autosomal dominant; Loeys-Dietz syndrome 5, 615582 (3), Autosomal dominant
TGFBI	601692	Corneal dystrophy, Avellino type, 607541 (3), Autosomal dominant; Corneal dystrophy, Groenouw type I, 121900 (3), Autosomal dominant; Corneal dystrophy, Reis-Bucklers type, 608470 (3); Corneal dystrophy, Thiel-Behnke type, 602082 (3), Autosomal dominant; Corneal dystrophy, epithelial basement membrane, 121820 (3), Autosomal dominant; Corneal dystrophy, lattice type I, 122200 (3), Autosomal dominant; Corneal dystrophy, lattice type IIIA, 608471 (3), Autosomal dominant
TGFBR1	190181	Loeys-Dietz syndrome 1, 609192 (3), Autosomal dominant; {Multiple self-healing squamous epithelioma, susceptibility to}, 132800 (3), Autosomal dominant
TGFBR2	190182	Colorectal cancer, hereditary nonpolyposis, type 6, 614331 (3); Esophageal cancer, somatic, 133239 (3); Loeys-Dietz syndrome 2, 610168 (3), Autosomal dominant
TGM1	190195	Ichthyosis, congenital, autosomal recessive 1, 242300 (3), Autosomal recessive

TGM3	600238	?Uncombable hair syndrome 2, 617251 (3), Autosomal recessive
TGM5	603805	Peeling skin syndrome 2, 609796 (3), Autosomal recessive
TGM6	613900	Spinocerebellar ataxia 35, 613908 (3), Autosomal dominant
TH	191290	Segawa syndrome, recessive, 605407 (3), Autosomal recessive
THAP1	609520	Dystonia 6, torsion, 602629 (3), Autosomal dominant
THBD	188040	{Hemolytic uremic syndrome, atypical, susceptibility to, 6}, 612926 (3), Autosomal dominant; Thrombophilia due to thrombomodulin defect, 614486 (3)
THBS2	188061	{Lumbar disc herniation, susceptibility to}, 603932 (3)
THOC2	300395	Mental retardation, X-linked 12/35, 300957 (3), X-linked recessive
THOC6	615403	Beaulieu-Boycott-Innes syndrome, 613680 (3), Autosomal recessive
THPO	600044	Thrombocythemia 1, 187950 (3), Autosomal dominant
THRA	190120	Hypothyroidism, congenital, nongoitrous, 6, 614450 (3), Autosomal dominant
TIA1	603518	Welander distal myopathy, 604454 (3), Autosomal recessive, Autosomal dominant
TICAM1	607601	{Encephalopathy, acute, infection-induced (herpes-specific), susceptibility to, 6}, 614850 (3), Autosomal recessive, Autosomal dominant
TIMM8A	300356	Mohr-Tranebjaerg syndrome, 304700 (3), X-linked recessive
TIMP3	188826	Sorsby fundus dystrophy, 136900 (3), Autosomal dominant
TINF2	604319	Dyskeratosis congenita, autosomal dominant 3, 613990 (3), Autosomal dominant; Revesz syndrome, 268130 (3), Autosomal dominant
TIRAP	606252	{Bacteremia, protection against}, 614382 (3); {Malaria, protection against}, 611162 (3); {Pneumococcal disease, invasive, protection against}, 610799 (3); {Tuberculosis, protection against}, 607948 (3)
TJP2	607709	Cholestasis, progressive familial intrahepatic 4, 615878 (3), Autosomal recessive; Hypercholanemia, familial, 607748 (3)
TKT	606781	Short stature, developmental delay, and congenital heart defects, 617044 (3), Autosomal recessive
TLE6	612399	Preimplantation embryonic lethality, 616814 (3), Autosomal recessive
TLL1	606742	Atrial septal defect 6, 613087 (3), Autosomal dominant
TLR1	601194	{Leprosy, protection against}, 613223 (3); {Leprosy, susceptibility to, 5}, 613223 (3)
TLR2	603028	{Colorectal cancer, susceptibility to}, 114500 (3), Autosomal dominant; {Leprosy, susceptibility to}, 246300 (3), Autosomal dominant; {Mycobacterium tuberculosis, susceptibility to}, 607948 (3)
TLR3	603029	{Encephalopathy, acute, infection-induced (herpes-specific), susceptibility to, 2}, 613002 (3), Autosomal recessive, Autosomal dominant; {HIV1 infection, resistance to}, 609423 (3)
TLR4	603030	No OMIM phenotype
TLR5	603031	{Legionnaire disease, susceptibility to}, 608556 (3); {Melioidosis, susceptibility to}, 615557 (3); {Systemic lupus erythematosus, resistance to}, 601744 (3); {Systemic lupus erythematosus, susceptibility to, 1}, 601744 (3)

TM4SF20	615404	{Specific language impairment 5}, 615432 (3), Autosomal dominant
TMC6	605828	Epidermodysplasia verruciformis, 226400 (3), Autosomal recessive
TMC8	605829	Epidermodysplasia verruciformis, 226400 (3), Autosomal recessive
TMEM126A	612988	Optic atrophy 7, 612989 (3), Autosomal recessive
TMEM138	614459	Joubert syndrome 16, 614465 (3), Autosomal recessive
TMEM165	614726	Congenital disorder of glycosylation, type IIk, 614727 (3), Autosomal recessive
TMEM173	612374	STING-associated vasculopathy, infantile-onset, 615934 (3), Autosomal dominant
TMEM199	616815	Congenital disorder of glycosylation, type IIp, 616829 (3), Autosomal recessive
TMEM216	613277	Joubert syndrome 2, 608091 (3), Autosomal recessive; Meckel syndrome 2, 603194 (3), Autosomal recessive
TMEM231	614949	Joubert syndrome 20, 614970 (3), Autosomal recessive; Meckel syndrome 11, 615397 (3), Autosomal recessive
TMEM237	614423	Joubert syndrome 14, 614424 (3), Autosomal recessive
TMEM240	616101	Spinocerebellar ataxia 21, 607454 (3), Autosomal dominant
TMEM38B	611236	Osteogenesis imperfecta, type XIV, 615066 (3)
TMEM43	612048	Arrhythmogenic right ventricular dysplasia 5, 604400 (3), Autosomal dominant; Emery-Dreifuss muscular dystrophy 7, AD, 614302 (3), Autosomal dominant
TMEM67	609884	{Bardet-Biedl syndrome 14, modifier of}, 615991 (3), Autosomal recessive; COACH syndrome, 216360 (3), Autosomal recessive; Joubert syndrome 6, 610688 (3), Autosomal recessive; Meckel syndrome 3, 607361 (3), Autosomal recessive; Nephronophthisis 11, 613550 (3), Autosomal recessive
TMEM70	612418	Mitochondrial complex V (ATP synthase) deficiency, nuclear type 2, 614052 (3), Autosomal recessive
TMIE	607237	Deafness, autosomal recessive 6, 600971 (3), Autosomal recessive
TMLHE	300777	{Autism, susceptibility to, X-linked 6}, 300872 (3), X-linked recessive
TMPRSS15	606635	Enterokinase deficiency, 226200 (3), Autosomal recessive
TMPRSS6	609862	Iron-refractory iron deficiency anemia, 206200 (3), Autosomal recessive
TMTC3	617218	Lissencephaly 8, 617255 (3), Autosomal recessive
TNC	187380	Deafness, autosomal dominant 56, 615629 (3), Autosomal dominant
TNF	191160	{Asthma, susceptibility to}, 600807 (3), Autosomal dominant; {Dementia, vascular, susceptibility to} (3); {Malaria, cerebral, susceptibility to}, 611162 (3); {Migraine without aura, susceptibility to}, 157300 (3), Autosomal dominant; {Septic shock, susceptibility to} (3)
TNFAIP3	191163	Autoinflammatory syndrome, familial, Behcet-like, 616744 (3), Autosomal dominant
TNFRSF10B	603612	Squamous cell carcinoma, head and neck, 275355 (3), Autosomal recessive

TNFRSF11A	603499	Osteolysis, familial expansile, 174810 (3), Autosomal dominant; Osteopetrosis, autosomal recessive 7, 612301 (3); {Paget disease of bone 2, early-onset}, 602080 (3), Autosomal dominant
TNFRSF11B	602643	Paget disease of bone 5, juvenile-onset, 239000 (3), Autosomal recessive
TNFRSF13B	604907	Immunodeficiency, common variable, 2, 240500 (3), Autosomal recessive, Autosomal dominant; Immunoglobulin A deficiency 2, 609529 (3)
TNFRSF13C	606269	Immunodeficiency, common variable, 4, 613494 (3), Autosomal recessive
TNFRSF1A	191190	{Multiple sclerosis, susceptibility to, 5}, 614810 (3); Periodic fever, familial, 142680 (3), Autosomal dominant
TNFRSF4	600315	?Immunodeficiency 16, 615593 (3), Autosomal recessive
TNFSF11	602642	Osteopetrosis, autosomal recessive 2, 259710 (3), Autosomal recessive
TNFSF4	603594	{Myocardial infarction, susceptibility to}, 608446 (3)
TNIK	610005	Mental retardation, autosomal recessive 54, 617028 (3), Autosomal recessive
TNNC1	191040	Cardiomyopathy, dilated, 1Z, 611879 (3); Cardiomyopathy, hypertrophic, 13, 613243 (3), Autosomal dominant
TNNI2	191043	Arthrogryposis multiplex congenita, distal, type 2B, 601680 (3), Autosomal dominant
TNNI3	191044	Cardiomyopathy, dilated, 1FF, 613286 (3); ?Cardiomyopathy, dilated, 2A, 611880 (3), Autosomal recessive; Cardiomyopathy, familial restrictive, 1, 115210 (3), Autosomal dominant; Cardiomyopathy, hypertrophic, 7, 613690 (3), Autosomal dominant
TNNI3K	613932	?Cardiac conduction disease with or without dilated cardiomyopathy, 616117 (3), Autosomal dominant
TNNT1	191041	Nemaline myopathy 5, Amish type, 605355 (3), Autosomal recessive
TNNT2	191045	Cardiomyopathy, dilated, 1D, 601494 (3), Autosomal dominant; Cardiomyopathy, familial restrictive, 3, 612422 (3), Autosomal dominant; Cardiomyopathy, hypertrophic, 2, 115195 (3), Autosomal dominant; Left ventricular noncompaction 6, 601494 (3), Autosomal dominant
TNXB	600985	Ehlers-Danlos syndrome, classic-like, 1, 606408 (3), Autosomal recessive; Vesicoureteral reflux 8, 615963 (3), Autosomal dominant
TOPORS	609507	Retinitis pigmentosa 31, 609923 (3)
TOR1A	605204	{Dystonia-1, modifier of} (3); Dystonia-1, torsion, 128100 (3), Autosomal dominant
TOR1AIP1	614512	?Muscular dystrophy, limb-girdle, type 2Y, 617072 (3), Autosomal recessive

TP53	191170	Adrenal cortical carcinoma, 202300 (3), Autosomal recessive; {Basal cell carcinoma 7}, 614740 (3); Breast cancer, 114480 (3), Autosomal dominant; Choroid plexus papilloma, 260500 (3), Autosomal dominant; Colorectal cancer, 114500 (3), Autosomal dominant; {Glioma susceptibility 1}, 137800 (3), Autosomal dominant, Somatic mutation; Hepatocellular carcinoma, 114550 (3), Somatic mutation; Li-Fraumeni syndrome, 151623 (3), Autosomal dominant; Nasopharyngeal carcinoma, 607107 (3); Osteosarcoma, 259500 (3), Autosomal recessive; Pancreatic cancer, 260350 (3), Autosomal dominant, Somatic mutation, Multifactorial
TP63	603273	ADULT syndrome, 103285 (3), Autosomal dominant; Ectrodactyly, ectodermal dysplasia, and cleft lip/palate syndrome 3, 604292 (3), Autosomal dominant; Hay-Wells syndrome, 106260 (3), Autosomal dominant; Limb-mammary syndrome, 603543 (3), Autosomal dominant; Orofacial cleft 8, 129400 (3), Autosomal dominant; Rapp-Hodgkin syndrome, 129400 (3), Autosomal dominant; Split-hand/foot malformation 4, 605289 (3), Autosomal dominant
TPCN2	612163	[Skin/hair/eye pigmentation 10, blond/brown hair], 612267 (3)
TPH2	607478	{Attention deficit-hyperactivity disorder, susceptibility to, 7}, 613003 (3); {Unipolar depression, susceptibility to}, 608516 (3)
TPI1	190450	Hemolytic anemia due to triosephosphate isomerase deficiency, 615512 (3), Autosomal recessive
TPK1	606370	Thiamine metabolism dysfunction syndrome 5 (episodic encephalopathy type), 614458 (3), Autosomal recessive
TPM1	191010	Cardiomyopathy, dilated, 1Y, 611878 (3), Autosomal dominant; Cardiomyopathy, hypertrophic, 3, 115196 (3), Autosomal dominant; Left ventricular noncompaction 9, 611878 (3), Autosomal dominant
TPM2	190990	Arthrogryposis multiplex congenita, distal, type 1, 108120 (3), Autosomal dominant; Arthrogryposis, distal, type 2B, 601680 (3), Autosomal dominant; CAP myopathy 2, 609285 (3), Autosomal dominant; Nemaline myopathy 4, autosomal dominant, 609285 (3), Autosomal dominant
TPM3	191030	CAP myopathy 1, 609284 (3), Autosomal recessive, Autosomal dominant; Myopathy, congenital, with fiber-type disproportion, 255310 (3), Autosomal recessive, Autosomal dominant; Nemaline myopathy 1, autosomal dominant or recessive, 609284 (3), Autosomal recessive, Autosomal dominant
TPMT	187680	{Thiopurines, poor metabolism of, 1}, 610460 (3), Autosomal recessive
TPP1	607998	Ceroid lipofuscinosis, neuronal, 2, 204500 (3), Autosomal recessive; Spinocerebellar ataxia, autosomal recessive 7, 609270 (3), Autosomal recessive
TPRN	613354	Deafness, autosomal recessive 79, 613307 (3), Autosomal recessive
TRAF3IP1	607380	Senior-Loken syndrome 9, 616629 (3), Autosomal recessive
TRAF3IP2	607043	?Candidiasis, familial, 8, 615527 (3), Autosomal recessive; {Psoriasis susceptibility 13}, 614070 (3)
TRAIP	605958	Seckel syndrome 9, 616777 (3), Autosomal recessive

TRAPPC11	614138	Muscular dystrophy, limb-girdle, type 2S, 615356 (3), Autosomal recessive
TRAPPC2	300202	Spondyloepiphyseal dysplasia tarda, 313400 (3), X-linked recessive
TRAPPC9	611966	Mental retardation, autosomal recessive 13, 613192 (3), Autosomal recessive
TRDN	603283	Ventricular tachycardia, catecholaminergic polymorphic, 5, with or without muscle weakness, 615441 (3), Autosomal recessive
TREH	275360	Trehalase deficiency, 612119 (3), Autosomal recessive
TREM2	605086	Nasu-Hakola disease, 221770 (3), Autosomal recessive
TREX1	606609	Aicardi-Goutieres syndrome 1, dominant and recessive, 225750 (3), Autosomal recessive, Autosomal dominant; Chilblain lupus, 610448 (3), Autosomal dominant; {Systemic lupus erythematosus, susceptibility to}, 152700 (3), Autosomal dominant; Vasculopathy, retinal, with cerebral leukodystrophy, 192315 (3), Autosomal dominant
TRH	613879	Thyrotropin-releasing hormone deficiency, 275120 (1), Autosomal recessive
TRIM2	614141	Charcot-Marie-Tooth disease, type 2R, 615490 (3), Autosomal recessive
TRIM24	603406	No OMIM phenotype
TRIM32	602290	?Bardet-Biedl syndrome 11, 615988 (3), Autosomal recessive; Muscular dystrophy, limb-girdle, type 2H, 254110 (3), Autosomal recessive
TRIM33	605769	No OMIM phenotype
TRIM44	612298	?Aniridia 3, 617142 (3), Autosomal dominant
TRIO	601893	Mental retardation, autosomal dominant 44, 617061 (3), Autosomal dominant
TRIOBP	609761	Deafness, autosomal recessive 28, 609823 (3), Autosomal recessive
TRIP11	604505	Achondrogenesis, type IA, 200600 (3), Autosomal recessive
TRIP4	604501	?Muscular dystrophy, congenital, Davignon-Chauveau type, 617066 (3), Autosomal recessive; Spinal muscular atrophy with congenital bone fractures 1, 616866 (3), Autosomal recessive
TRMT10A	616013	Microcephaly, short stature, and impaired glucose metabolism 1, 616033 (3), Autosomal recessive
TRMT10C	615423	Combined oxidative phosphorylation deficiency 30, 616974 (3), Autosomal recessive
TRMT5	611023	Combined oxidative phosphorylation deficiency 26, 616539 (3), Autosomal recessive
TRMU	610230	{Deafness, mitochondrial, modifier of}, 580000 (3), Mitochondrial; Liver failure, transient infantile, 613070 (3), Autosomal recessive
TRNT1	612907	Retinitis pigmentosa and erythrocytic microcytosis, 616959 (3), Autosomal recessive; Sideroblastic anemia with B-cell immunodeficiency, periodic fevers, and developmental delay, 616084 (3), Autosomal recessive
TRPA1	604775	?Episodic pain syndrome, familial, 1, 615040 (3), Autosomal dominant
TRPC3	602345	?Spinocerebellar ataxia 41, 616410 (3), Autosomal dominant

TRPC6	603652	Glomerulosclerosis, focal segmental, 2, 603965 (3)
TRPM1	603576	Night blindness, congenital stationary (complete), 1C, autosomal recessive, 613216 (3)
TRPM4	606936	Progressive familial heart block, type IB, 604559 (3), Autosomal dominant
TRPM6	607009	Hypomagnesemia 1, intestinal, 602014 (3), Autosomal recessive
TRPM7	605692	{Amyotrophic lateral sclerosis-parkinsonism/dementia complex, susceptibility to}, 105500 (3), Autosomal dominant
TRPV3	607066	Olmsted syndrome, 614594 (3), Autosomal dominant; ?Palmoplantar keratoderma, nonepidermolytic, focal 2, 616400 (3), Autosomal dominant
TRPV4	605427	?Avascular necrosis of femoral head, primary, 2, 617383 (3), Autosomal dominant; Brachyolmia type 3, 113500 (3), Autosomal dominant; Digital arthropathy-brachydactyly, familial, 606835 (3), Autosomal dominant; Hereditary motor and sensory neuropathy, type IIc, 606071 (3), Autosomal dominant; Metatropic dysplasia, 156530 (3), Autosomal dominant; Parastremmatic dwarfism, 168400 (3), Autosomal dominant; SED, Maroteaux type, 184095 (3), Autosomal dominant; Scapuloperoneal spinal muscular atrophy, 181405 (3), Autosomal dominant; [Sodium serum level QTL 1], 613508 (3); Spinal muscular atrophy, distal, congenital nonprogressive, 600175 (3), Autosomal dominant; Spondylometaphyseal dysplasia, Kozlowski type, 184252 (3), Autosomal dominant
TSC1	605284	Focal cortical dysplasia, type II, somatic, 607341 (3); Lymphangioleiomyomatosis, 606690 (3); Tuberous sclerosis-1, 191100 (3), Autosomal dominant
TSC2	191092	?Focal cortical dysplasia, type II, somatic, 607341 (3); Lymphangioleiomyomatosis, somatic, 606690 (3); Tuberous sclerosis-2, 613254 (3), Autosomal dominant
TSEN15	608756	Pontocerebellar hypoplasia, type 2F, 617026 (3), Autosomal recessive
TSEN54	608755	Pontocerebellar hypoplasia type 2A, 277470 (3), Autosomal recessive; Pontocerebellar hypoplasia type 4, 225753 (3), Autosomal recessive; ?Pontocerebellar hypoplasia type 5, 610204 (3), Autosomal recessive
TSFM	604723	Combined oxidative phosphorylation deficiency 3, 610505 (3), Autosomal recessive
TSG101	601387	Breast cancer, somatic, 114480 (3)
TSHB	188540	Hypothyroidism, congenital, nongoitrous 4, 275100 (3), Autosomal recessive
TSHR	603372	Hyperthyroidism, familial gestational, 603373 (3); Hyperthyroidism, nonautoimmune, 609152 (3), Autosomal dominant, Isolated cases; Hypothyroidism, congenital, nongoitrous, 1, 275200 (3), Autosomal recessive; Thyroid adenoma, hyperfunctioning, somatic (3); Thyroid carcinoma with thyrotoxicosis (3)
TSHZ1	614427	Aural atresia, congenital, 607842 (3), Autosomal dominant
TSPAN12	613138	Exudative vitreoretinopathy 5, 613310 (3), Autosomal dominant
TSPAN7	300096	Mental retardation, X-linked 58, 300210 (3), X-linked recessive

TSPEAR	612920	?Deafness, autosomal recessive 98, 614861 (3), Autosomal recessive
TSPYL1	604714	Sudden infant death with dysgenesis of the testes syndrome, 608800 (3), Autosomal recessive
TSR2	300945	?Diamond-Blackfan anemia 14 with mandibulofacial dysostosis, 300946 (3), X-linked recessive
TTBK2	611695	Spinocerebellar ataxia 11, 604432 (3), Autosomal dominant
TTC19	613814	Mitochondrial complex III deficiency, nuclear type 2, 615157 (3), Autosomal recessive
TTC21B	612014	Nephronophthisis 12, 613820 (3), Autosomal recessive, Autosomal dominant; Short-rib thoracic dysplasia 4 with or without polydactyly, 613819 (3), Autosomal recessive
TTC25	617095	Ciliary dyskinesia, primary, 35, 617092 (3), Autosomal recessive
TTC37	614589	Trichohepatoenteric syndrome 1, 222470 (3), Autosomal recessive
TTC7A	609332	Gastrointestinal defects and immunodeficiency syndrome, 243150 (3), Autosomal recessive
TTI2	614426	Mental retardation, autosomal recessive 39, 615541 (3), Autosomal recessive
TTLL5	612268	Cone-rod dystrophy 19, 615860 (3), Autosomal recessive
TTN	188840	Cardiomyopathy, dilated, 1G, 604145 (3); Cardiomyopathy, familial hypertrophic, 9, 613765 (3), Autosomal dominant; Muscular dystrophy, limb-girdle, type 2J, 608807 (3), Autosomal recessive; Myopathy, proximal, with early respiratory muscle involvement, 603689 (3); Salih myopathy, 611705 (3), Autosomal recessive; Tibial muscular dystrophy, tardive, 600334 (3), Autosomal dominant
TTPA	600415	Ataxia with isolated vitamin E deficiency, 277460 (3), Autosomal recessive
TTR	176300	Amyloidosis, hereditary, transthyretin-related, 105210 (3), Autosomal dominant; Carpal tunnel syndrome, familial, 115430 (3), Autosomal dominant; [Dystransthyretinemic hyperthyroxinemia], 145680 (3), Autosomal dominant
TUB	601197	?Retinal dystrophy and obesity, 616188 (3), Autosomal recessive
TUBA1A	602529	Lissencephaly 3, 611603 (3), Autosomal dominant
TUBA4A	191110	Amyotrophic lateral sclerosis 22 with or without frontotemporal dementia, 616208 (3), Autosomal dominant
TUBA8	605742	Cortical dysplasia, complex, with other brain malformations 8, 613180 (3), Autosomal recessive
TUBB	191130	Cortical dysplasia, complex, with other brain malformations 6, 615771 (3), Autosomal dominant; Symmetric circumferential skin creases, congenital, 1, 156610 (3), Autosomal dominant
TUBB1	612901	Macrothrombocytopenia, autosomal dominant, TUBB1-related, 613112 (3), Autosomal dominant
TUBB2A	615101	Cortical dysplasia, complex, with other brain malformations 5, 615763 (3), Autosomal dominant

TUBB2B	612850	Cortical dysplasia, complex, with other brain malformations 7, 610031 (3), Autosomal dominant
TUBB3	602661	Cortical dysplasia, complex, with other brain malformations 1, 614039 (3), Autosomal dominant; Fibrosis of extraocular muscles, congenital, 3A, 600638 (3), Autosomal dominant
TUBB8	616768	Oocyte maturation defect 2, 616780 (3), Autosomal recessive, Autosomal dominant
TUBG1	191135	Cortical dysplasia, complex, with other brain malformations 4, 615412 (3), Autosomal dominant
TUBGCP4	609610	Microcephaly and chorioretinopathy, autosomal recessive, 3, 616335 (3), Autosomal recessive
TUBGCP6	610053	Microcephaly and chorioretinopathy, autosomal recessive, 1, 251270 (3), Autosomal recessive
TUFM	602389	Combined oxidative phosphorylation deficiency 4, 610678 (3), Autosomal recessive
TULP1	602280	Leber congenital amaurosis 15, 613843 (3), Autosomal recessive; Retinitis pigmentosa 14, 600132 (3), Autosomal recessive
TUSC3	601385	Mental retardation, autosomal recessive 7, 611093 (3), Autosomal recessive
TWIST1	601622	Craniosynostosis 1, 123100 (3), Autosomal dominant; Robinow-Sorauf syndrome, 180750 (3), Autosomal dominant; Saethre-Chotzen syndrome with or without eyelid anomalies, 101400 (3), Autosomal dominant; Sweeney-Cox syndrome, 617746 (3), Autosomal dominant
TWIST2	607556	Ablepharon-macrostomia syndrome, 200110 (3), Autosomal dominant; Barber-Say syndrome, 209885 (3), Autosomal dominant; Focal facial dermal dysplasia 3, Setleis type, 227260 (3), Autosomal recessive
TWNK	606075	Mitochondrial DNA depletion syndrome 7 (hepatocerebral type), 271245 (3), Autosomal recessive; Perrault syndrome 5, 616138 (3), Autosomal recessive; Progressive external ophthalmoplegia with mitochondrial DNA deletions, autosomal dominant 3, 609286 (3), Autosomal dominant
TXN2	609063	?Combined oxidative phosphorylation deficiency 29, 616811 (3), Autosomal recessive
TXNL4A	611595	Burn-McKeown syndrome, 608572 (3), Autosomal recessive
TYMP	131222	Mitochondrial DNA depletion syndrome 1 (MNGIE type), 603041 (3), Autosomal recessive
TYR	606933	Albinism, oculocutaneous, type IA, 203100 (3), Autosomal recessive; Albinism, oculocutaneous, type IB, 606952 (3); {Melanoma, cutaneous malignant, susceptibility to, 8}, 601800 (3); [Skin/hair/eye pigmentation 3, blue/green eyes], 601800 (3); [Skin/hair/eye pigmentation 3, light/dark/freckling skin], 601800 (3); Waardenburg syndrome/albinism, digenic, 103470 (3), Autosomal dominant
TYROBP	604142	Nasu-Hakola disease, 221770 (3), Autosomal recessive

TYRP1	115501	Albinism, oculocutaneous, type III, 203290 (3), Autosomal recessive; [Skin/hair/eye pigmentation, variation in, 11 (Melanesian blond hair)], 612271 (3)
UBA1	314370	Spinal muscular atrophy, X-linked 2, infantile, 301830 (3), X-linked recessive
UBA5	610552	Epileptic encephalopathy, early infantile, 44, 617132 (3), Autosomal recessive; ?Spinocerebellar ataxia, autosomal recessive 24, 617133 (3), Autosomal recessive
UBB	191339	Cleft palate, isolated, 119540 (2), Autosomal dominant
UBE2A	312180	Mental retardation, X-linked syndromic, Nascimento-type, 300860 (3), X-linked recessive
UBE2T	610538	Fanconi anemia, complementation group T, 616435 (3), Autosomal recessive
UBQLN2	300264	Amyotrophic lateral sclerosis 15, with or without frontotemporal dementia, 300857 (3), X-linked dominant
UBR1	605981	Johanson-Blizzard syndrome, 243800 (3), Autosomal recessive
UCHL1	191342	{?Parkinson disease 5, susceptibility to}, 613643 (3); Spastic paraplegia 79, autosomal recessive, 615491 (3), Autosomal recessive
UCP1	113730	{Obesity, susceptibility to}, 601665 (3), Autosomal recessive, Autosomal dominant, Multifactorial
UCP2	601693	{Obesity, susceptibility to, BMIQ4}, 607447 (3)
UCP3	602044	{Obesity, severe, and type II diabetes}, 601665 (3), Autosomal recessive, Autosomal dominant, Multifactorial
UFSP2	611482	?Hip dysplasia, Beukes type, 142669 (3), Autosomal dominant; ?Spondyloepimetaphyseal dysplasia, Di Rocco type, 617974 (3), Autosomal dominant
UGT1A1	191740	[Bilirubin, serum level of, QTL1], 601816 (3); Crigler-Najjar syndrome, type I, 218800 (3), Autosomal recessive; Crigler-Najjar syndrome, type II, 606785 (3), Autosomal recessive; [Gilbert syndrome], 143500 (3), Autosomal recessive; Hyperbilirubinemia, familial transient neonatal, 237900 (3), Autosomal recessive
UGT2B17	601903	{Bone mineral density QTL 12, osteoporosis}, 612560 (3)
UMOD	191845	Glomerulocystic kidney disease with hyperuricemia and isosthenuria, 609886 (3); Hyperuricemic nephropathy, familial juvenile 1, 162000 (3), Autosomal dominant; Medullary cystic kidney disease 2, 603860 (3)
UMPS	613891	Orotic aciduria, 258900 (3), Autosomal recessive
UNC119	604011	?Cone-rod dystrophy (3); ?Immunodeficiency 13, 615518 (3), Autosomal dominant
UNC45B	611220	?Cataract 43, 616279 (3), Autosomal dominant
UNC80	612636	Hypotonia, infantile, with psychomotor retardation and characteristic facies 2, 616801 (3), Autosomal recessive
UNC93B1	608204	{Encephalopathy, acute, infection-induced (herpes-specific), susceptibility to, 1}, 610551 (3)

UNG	191525	Immunodeficiency with hyper IgM, type 5, 608106 (3), Autosomal recessive
UPB1	606673	Beta-ureidopropionase deficiency, 613161 (3), Autosomal recessive
UPF3B	300298	Mental retardation, X-linked, syndromic 14, 300676 (3), X-linked recessive
UQCC2	614461	?Mitochondrial complex III deficiency, nuclear type 7, 615824 (3), Autosomal recessive
UQCC3	616097	?Mitochondrial complex III deficiency, nuclear type 9, 616111 (3), Autosomal recessive
UQCRB	191330	Mitochondrial complex III deficiency, nuclear type 3, 615158 (3), Autosomal recessive
UQCRC2	191329	Mitochondrial complex III deficiency, nuclear type 5, 615160 (3), Autosomal recessive
UQCRQ	612080	Mitochondrial complex III deficiency, nuclear type 4, 615159 (3), Autosomal recessive
UROC1	613012	?Urocanase deficiency, 276880 (3), Autosomal recessive
UROD	613521	Porphyria cutanea tarda, 176100 (3), Autosomal recessive, Autosomal dominant; Porphyria, hepatoerythropoietic, 176100 (3), Autosomal recessive, Autosomal dominant
UROS	606938	Porphyria, congenital erythropoietic, 263700 (3), Autosomal recessive
USB1	613276	Poikiloderma with neutropenia, 604173 (3), Autosomal recessive
USF1	191523	{Hyperlipidemia, familial combined, susceptibility to}, 602491 (3)
USH1C	605242	Deafness, autosomal recessive 18A, 602092 (3), Autosomal recessive; Usher syndrome, type 1C, 276904 (3), Autosomal recessive
USH1G	607696	Usher syndrome, type 1G, 606943 (3), Autosomal recessive
USH2A	608400	Retinitis pigmentosa 39, 613809 (3); Usher syndrome, type 2A, 276901 (3), Autosomal recessive
USP18	607057	Pseudo-TORCH syndrome 2, 617397 (3), Autosomal recessive
USP27X	300975	Mental retardation, X-linked 105, 300984 (3), X-linked recessive
USP9X	300072	Mental retardation, X-linked 99, 300919 (3), X-linked recessive; Mental retardation, X-linked 99, syndromic, female-restricted, 300968 (3), X-linked dominant
USP9Y	400005	Spermatogenic failure, Y-linked, 2, 415000 (3), Y-linked
UTP4	607456	No OMIM phenotype
UVSSA	614632	UV-sensitive syndrome 3, 614640 (3), Autosomal recessive
VAC14	604632	Striatonigral degeneration, childhood-onset, 617054 (3), Autosomal recessive
VAMP1	185880	Spastic ataxia 1, autosomal dominant, 108600 (3), Autosomal dominant
VANGL1	610132	Caudal regression syndrome, 600145 (3), Autosomal dominant; {Neural tube defects, susceptibility to}, 182940 (3), Autosomal dominant
VANGL2	600533	Neural tube defects, 182940 (3), Autosomal dominant

VAPB	605704	Amyotrophic lateral sclerosis 8, 608627 (3), Autosomal dominant; Spinal muscular atrophy, late-onset, Finkel type, 182980 (3), Autosomal dominant
VARS2	612802	Combined oxidative phosphorylation deficiency 20, 615917 (3), Autosomal recessive
VAX1	604294	?Microphthalmia, syndromic 11, 614402 (3), Autosomal recessive
VCAN	118661	Wagner syndrome 1, 143200 (3), Autosomal dominant
VCL	193065	Cardiomyopathy, dilated, 1W, 611407 (3); Cardiomyopathy, hypertrophic, 15, 613255 (3), Autosomal dominant
VCP	601023	Amyotrophic lateral sclerosis 14, with or without frontotemporal dementia, 613954 (3); Charcot-Marie-Tooth disease, type 2Y, 616687 (3), Autosomal dominant; Inclusion body myopathy with early-onset Paget disease and frontotemporal dementia 1, 167320 (3), Autosomal dominant
VCY	400012	No OMIM phenotype
VDR	601769	?Osteoporosis, involutional, 166710 (1), Autosomal dominant; Rickets, vitamin D-resistant, type IIA, 277440 (3), Autosomal recessive
VEGFA	192240	{Microvascular complications of diabetes 1}, 603933 (3)
VEGFC	601528	Lymphedema, hereditary, ID, 615907 (3), Autosomal dominant
VHL	608537	Erythrocytosis, familial, 2, 263400 (3), Autosomal recessive; Hemangioblastoma, cerebellar, somatic (3); Pheochromocytoma, 171300 (3), Autosomal dominant; Renal cell carcinoma, somatic, 144700 (3); von Hippel-Lindau syndrome, 193300 (3), Autosomal dominant
VIM	193060	Cataract 30, pulverulent, 116300 (3), Autosomal dominant
VIPAS39	613401	Arthrogryposis, renal dysfunction, and cholestasis 2, 613404 (3), Autosomal recessive
VKORC1	608547	Vitamin K-dependent clotting factors, combined deficiency of, 2, 607473 (3); Warfarin resistance, 122700 (3), Autosomal dominant
VLDLR	192977	Cerebellar hypoplasia and mental retardation with or without quadrupedal locomotion 1, 224050 (3), Autosomal recessive
VMA21	300913	Myopathy, X-linked, with excessive autophagy, 310440 (3), X-linked recessive
VPS11	608549	Leukodystrophy, hypomyelinating, 12, 616683 (3), Autosomal recessive
VPS13A	605978	Choreoacanthocytosis, 200150 (3), Autosomal recessive
VPS13B	607817	Cohen syndrome, 216550 (3), Autosomal recessive
VPS13C	608879	Parkinson disease 23, autosomal recessive, early onset, 616840 (3), Autosomal recessive
VPS33A	610034	Mucopolysaccharidosis-plus syndrome, 617303 (3), Autosomal recessive
VPS33B	608552	Arthrogryposis, renal dysfunction, and cholestasis 1, 208085 (3), Autosomal recessive
VPS35	601501	{Parkinson disease 17}, 614203 (3), Autosomal dominant
VPS37A	609927	Spastic paraplegia 53, autosomal recessive, 614898 (3), Autosomal recessive

VPS45	610035	Neutropenia, severe congenital, 5, autosomal recessive, 615285 (3), Autosomal recessive
VPS53	615850	Pontocerebellar hypoplasia, type 2E, 615851 (3), Autosomal recessive
VRK1	602168	Pontocerebellar hypoplasia type 1A, 607596 (3), Autosomal recessive
VSX1	605020	?Craniofacial anomalies and anterior segment dysgenesis syndrome, 614195 (3); Keratoconus 1, 148300 (3), Autosomal dominant
VSX2	142993	Microphthalmia with coloboma 3, 610092 (3); Microphthalmia, isolated 2, 610093 (3)
VWA3B	614884	?Spinocerebellar ataxia, autosomal recessive 22, 616948 (3), Autosomal recessive
VWF	613160	von Willebrand disease, type 1, 193400 (3), Autosomal dominant; von Willebrand disease, types 2A, 2B, 2M, and 2N, 613554 (3), Autosomal recessive, Autosomal dominant; von Willibrand disease, type 3, 277480 (3), Autosomal recessive
WAC	615049	Desanto-Shinawi syndrome, 616708 (3), Autosomal dominant
WAS	300392	Neutropenia, severe congenital, X-linked, 300299 (3), X-linked recessive; Thrombocytopenia, X-linked, 313900 (3), X-linked recessive; Thrombocytopenia, X-linked, intermittent, 313900 (3), X-linked recessive; Wiskott-Aldrich syndrome, 301000 (3), X-linked recessive
WASHC4	615748	?Mental retardation, autosomal recessive 43, 615817 (3), Autosomal recessive
WASHC5	610657	Ritscher-Schinzel syndrome 1, 220210 (3), Autosomal recessive; Spastic paraplegia 8, autosomal dominant, 603563 (3), Autosomal dominant
WDPCP	613580	?Bardet-Biedl syndrome 15, 615992 (3), Autosomal recessive; ?Congenital heart defects, hamartomas of tongue, and polysyndactyly, 217085 (3), Autosomal recessive
WDR11	606417	Hypogonadotropic hypogonadism 14 with or without anosmia, 614858 (3), Autosomal dominant
WDR19	608151	?Cranioectodermal dysplasia 4, 614378 (3), Autosomal recessive; Nephronophthisis 13, 614377 (3), Autosomal recessive; Senior-Loken syndrome 8, 616307 (3), Autosomal recessive; ?Short-rib thoracic dysplasia 5 with or without polydactyly, 614376 (3), Autosomal recessive
WDR34	613363	Short-rib thoracic dysplasia 11 with or without polydactyly, 615633 (3), Autosomal recessive
WDR35	613602	Cranioectodermal dysplasia 2, 613610 (3), Autosomal recessive; Short-rib thoracic dysplasia 7 with or without polydactyly, 614091 (3), Autosomal recessive
WDR36	609669	Glaucoma 1, open angle, G, 609887 (3)
WDR45	300526	Neurodegeneration with brain iron accumulation 5, 300894 (3), X-linked dominant
WDR60	615462	Short-rib thoracic dysplasia 8 with or without polydactyly, 615503 (3), Autosomal recessive

WDR62	613583	Microcephaly 2, primary, autosomal recessive, with or without cortical malformations, 604317 (3), Autosomal recessive
WDR72	613214	Amelogenesis imperfecta, type IIA3, 613211 (3), Autosomal recessive
WDR73	616144	Galloway-Mowat syndrome 1, 251300 (3), Autosomal recessive
WDR81	614218	Cerebellar ataxia, mental retardation, and dysequilibrium syndrome 2, 610185 (3), Autosomal recessive; Hydrocephalus, nonsyndromic, autosomal recessive 3, 617967 (3)
WFS1	606201	?Cataract 41, 116400 (3), Autosomal dominant; Deafness, autosomal dominant 6/14/38, 600965 (3), Autosomal dominant; {Diabetes mellitus, noninsulin-dependent, association with}, 125853 (3), Autosomal dominant; Wolfram syndrome 1, 222300 (3), Autosomal recessive; Wolfram-like syndrome, autosomal dominant, 614296 (3), Autosomal dominant
WHRN	607928	Deafness, autosomal recessive 31, 607084 (3), Autosomal recessive; Usher syndrome, type 2D, 611383 (3), Autosomal recessive
WISP3	603400	Arthropathy, progressive pseudorheumatoid, of childhood, 208230 (3), Autosomal recessive; Spondyloepiphyseal dysplasia tarda with progressive arthropathy, 208230 (3), Autosomal recessive
WNK1	605232	Neuropathy, hereditary sensory and autonomic, type II, 201300 (3), Autosomal recessive; Pseudohypoaldosteronism, type IIC, 614492 (3), Autosomal dominant
WNK4	601844	Pseudohypoaldosteronism, type IIB, 614491 (3), Autosomal dominant
WNT1	164820	Osteogenesis imperfecta, type XV, 615220 (3), Autosomal recessive; {Osteoporosis, early-onset, susceptibility to, autosomal dominant}, 615221 (3)
WNT10A	606268	Odontoonychodermal dysplasia, 257980 (3), Autosomal recessive; Schopf-Schulz-Passarge syndrome, 224750 (3), Autosomal recessive; Tooth agenesis, selective, 4, 150400 (3), Autosomal recessive, Autosomal dominant
WNT10B	601906	Split-hand/foot malformation 6, 225300 (3), Autosomal recessive; Tooth agenesis, selective, 8, 617073 (3), Autosomal dominant
WNT3	165330	?Tetra-amelia syndrome 1, 273395 (3), Autosomal recessive
WNT4	603490	Mullerian aplasia and hyperandrogenism, 158330 (3), Autosomal dominant; ?SERKAL syndrome, 611812 (3), Autosomal recessive
WNT5A	164975	Robinow syndrome, autosomal dominant 1, 180700 (3), Autosomal dominant
WNT7A	601570	Fuhrmann syndrome, 228930 (3), Autosomal recessive; Ulna and fibula, absence of, with severe limb deficiency, 276820 (3), Autosomal recessive
WRAP53	612661	Dyskeratosis congenita, autosomal recessive 3, 613988 (3), Autosomal recessive
WRN	604611	Werner syndrome, 277700 (3), Autosomal recessive

WT1	607102	Denys-Drash syndrome, 194080 (3), Autosomal dominant, Somatic mutation; Frasier syndrome, 136680 (3), Autosomal dominant, Somatic mutation; Meacham syndrome, 608978 (3); Mesothelioma, somatic, 156240 (3); Nephrotic syndrome, type 4, 256370 (3), Autosomal dominant; Wilms tumor, type 1, 194070 (3), Autosomal dominant, Somatic mutation
WWC1	610533	[Memory, enhanced, QTL], 615602 (3)
WWOX	605131	Epileptic encephalopathy, early infantile, 28, 616211 (3), Autosomal recessive; Esophageal squamous cell carcinoma, somatic, 133239 (3); Spinocerebellar ataxia, autosomal recessive 12, 614322 (3), Autosomal recessive
XBP1	194355	{Major affective disorder-7, susceptibility to}, 612371 (3)
XDH	607633	Xanthinuria, type I, 278300 (3), Autosomal recessive
XIAP	300079	Lymphoproliferative syndrome, X-linked, 2, 300635 (3), X-linked recessive
XK	314850	McLeod syndrome with or without chronic granulomatous disease, 300842 (3), X-linked
XPA	611153	Xeroderma pigmentosum, group A, 278700 (3), Autosomal recessive
XPC	613208	Xeroderma pigmentosum, group C, 278720 (3), Autosomal recessive
XPNPEP2	300145	{Angioedema induced by ACE inhibitors, susceptibility to}, 300909 (3)
XPNPEP3	613553	Nephronophthisis-like nephropathy 1, 613159 (3), Autosomal recessive
XPR1	605237	Basal ganglia calcification, idiopathic, 6, 616413 (3), Autosomal dominant
XRCC2	600375	?Fanconi anemia, complementation group U, 617247 (3), Autosomal recessive
XRCC3	600675	{Breast cancer, susceptibility to}, 114480 (3), Autosomal dominant; {Melanoma, cutaneous malignant, 6}, 613972 (3)
XRCC4	194363	Short stature, microcephaly, and endocrine dysfunction, 616541 (3), Autosomal recessive
XYLT1	608124	Desbuquois dysplasia 2, 615777 (3), Autosomal recessive; {Pseudoxanthoma elasticum, modifier of severity of}, 264800 (3), Autosomal recessive
XYLT2	608125	{Pseudoxanthoma elasticum, modifier of severity of}, 264800 (3), Autosomal recessive; Spondyloocular syndrome, 605822 (3), Autosomal recessive
YAP1	606608	Coloboma, ocular, with or without hearing impairment, cleft lip/palate, and/or mental retardation, 120433 (3), Autosomal dominant
YARS	603623	Charcot-Marie-Tooth disease, dominant intermediate C, 608323 (3), Autosomal dominant
YARS2	610957	Myopathy, lactic acidosis, and sideroblastic anemia 2, 613561 (3), Autosomal recessive
YME1L1	607472	?Optic atrophy 11, 617302 (3), Autosomal recessive

ZAP70	176947	Autoimmune disease, multisystem, infantile-onset, 2, 617006 (3), Autosomal recessive; Immunodeficiency 48, 269840 (3), Autosomal recessive
ZBTB16	176797	Leukemia, acute promyelocytic, PL2F/RARA type (3); Skeletal defects, genital hypoplasia, and mental retardation, 612447 (3), Autosomal recessive
ZBTB18	608433	Mental retardation, autosomal dominant 22, 612337 (3), Autosomal dominant
ZBTB24	614064	Immunodeficiency-centromeric instability-facial anomalies syndrome 2, 614069 (3), Autosomal recessive
ZBTB42	613915	?Lethal congenital contracture syndrome 6, 616248 (3), Autosomal recessive
ZC3H14	613279	Mental retardation, autosomal recessive 56, 617125 (3), Autosomal recessive
ZDHHC15	300576	?Mental retardation, X-linked 91, 300577 (3), X-linked dominant
ZDHHC9	300646	Mental retardation, X-linked syndromic, Raymond type, 300799 (3)
ZFAT	610931	{Autoimmune thyroid disease, susceptibility to, 3}, 608175 (3)
ZFHX3	104155	{Prostate cancer, susceptibility to, somatic}, 176807 (3)
ZFHX4	606940	?Ptosis, congenital, 178300 (2), Autosomal dominant
ZFP57	612192	Diabetes mellitus, transient neonatal, 1, 601410 (3)
ZFPM2	603693	Diaphragmatic hernia 3, 610187 (3); Tetralogy of Fallot, 187500 (3), Autosomal dominant; 46XY sex reversal 9, 616067 (3), Autosomal dominant
ZFYVE26	612012	Spastic paraplegia 15, autosomal recessive, 270700 (3), Autosomal recessive
ZIC1	600470	Craniosynostosis 6, 616602 (3), Autosomal dominant
ZIC2	603073	Holoprosencephaly 5, 609637 (3), Autosomal dominant
ZIC3	300265	Congenital heart defects, nonsyndromic, 1, X-linked, 306955 (3), X-linked recessive; Heterotaxy, visceral, 1, X-linked, 306955 (3), X-linked recessive; VACTERL association, X-linked, 314390 (3), X-linked recessive
ZMPSTE24	606480	Mandibuloacral dysplasia with type B lipodystrophy, 608612 (3), Autosomal recessive; Restrictive dermopathy, lethal, 275210 (3), Autosomal recessive
ZMYND10	607070	Ciliary dyskinesia, primary, 22, 615444 (3), Autosomal recessive
ZMYND15	614312	?Spermatogenic failure 14, 615842 (3), Autosomal recessive
ZNF141	194648	?Polydactyly, postaxial, type A6, 615226 (3), Autosomal recessive
ZNF148	601897	Global developmental delay, absent or hypoplastic corpus callosum, and dysmorphic facies, 617260 (3), Autosomal dominant
ZNF335	610827	?Microcephaly 10, primary, autosomal recessive, 615095 (3), Autosomal recessive
ZNF365	607818	{Nephrolithiasis, uric acid, susceptibility to}, 605990 (3)
ZNF408	616454	?Exudative vitreoretinopathy 6, 616468 (3), Autosomal dominant; Retinitis pigmentosa 72, 616469 (3), Autosomal recessive
ZNF469	612078	Brittle cornea syndrome 1, 229200 (3), Autosomal recessive
ZNF513	613598	?Retinitis pigmentosa 58, 613617 (3), Autosomal recessive

ZNF592	613624	No OMIM phenotype
ZNF644	614159	Myopia 21, autosomal dominant, 614167 (3), Autosomal dominant
ZNF750	610226	Seborrhea-like dermatitis with psoriasiform elements, 610227 (3)
ZNF81	314998	No OMIM phenotype
ZP1	195000	Oocyte maturation defect 1, 615774 (3), Autosomal recessive
ZSWIM6	615951	Acromelic frontonasal dysostosis, 603671 (3), Autosomal dominant; Neurodevelopmental disorder with movement abnormalities, abnormal gait, and autistic features, 617865 (3), Autosomal dominant

Gene symbols used are according to the HGNC guidelines. For some genes a previously HGNC-approved symbol is in brackets.
Each Phenotype is followed by its MIM number, phenotype mapping key and inheritance pattern.
OMIM release used for OMIM disease identifiers and descriptions: July 04, 2018

Possible phenotype mapping keys
(1) the disorder is placed on the map based on its association with a gene, but the underlying defect is not known
(2) the disorder has been placed on the map by linkage; no mutation has been found
(3) the molecular basis for the disorder is known; a mutation has been found in the gene
(4) a contiguous gene deletion or duplication syndrome, multiple genes are deleted or duplicated causing the phenotype

Brackets, "[ ]", indicate "nondiseases," mainly genetic variations that lead to apparently abnormal laboratory test values (e.g., dysalbuminemic euthyroidal hyperthyroxinemia).
Braces, "{ }", indicate mutations that contribute to susceptibility to multifactorial disorders (e.g., diabetes, asthma) or to susceptibility to infection (e.g., malaria).
A question mark, "?", before the phenotype name indicates that the relationship between the phenotype and gene is provisional. More details about this relationship are provided in the comment field of the map and in the gene and phenotype OMIM entries.