

Mendiolome panel		
versie	V3 (4362 genen)	Centrum voor Medische Genetica Gent

Gene	OMIM gene ID	Associated phenotype, OMIM phenotype ID, phenotype mapping key and inheritance pattern
<i>A2M</i>	103950	{Alzheimer disease, susceptibility to}, 104300 (3), Autosomal dominant
<i>A2ML1</i>	610627	{Otitis media, susceptibility to}, 166760 (3), Autosomal dominant
<i>A4GALT</i>	607922	[Blood group, P1Pk system, p phenotype], 111400 (3); NOR polyagglutination syndrome, 111400 (3); [Blood group, P1Pk system, P(2) phenotype], 111400 (3)
<i>AAAS</i>	605378	Achalasia-addisonianism-alacrimia syndrome, 231550 (3), Autosomal recessive
<i>AAGAB</i>	614888	Keratoderma, palmoplantar, punctate type IA, 148600 (3), Autosomal dominant
<i>AARS1</i> (AARS)	601065	Epileptic encephalopathy, early infantile, 29, 616339 (3), Autosomal recessive; Charcot-Marie-Tooth disease, axonal, type 2N, 613287 (3), Autosomal dominant
<i>AARS2</i>	612035	Combined oxidative phosphorylation deficiency 8, 614096 (3), Autosomal recessive; Leukoencephalopathy, progressive, with ovarian failure, 615889 (3), Autosomal recessive
<i>AASS</i>	605113	Hyperlysinemia, 238700 (3), Autosomal recessive
<i>ABAT</i>	137150	GABA-transaminase deficiency, 613163 (3), Autosomal recessive
<i>ABCA1</i>	600046	HDL deficiency, familial, 1, 604091 (3); Tangier disease, 205400 (3), Autosomal recessive
<i>ABCA12</i>	607800	Ichthyosis, congenital, autosomal recessive 4B (harlequin), 242500 (3), Autosomal recessive; Ichthyosis, congenital, autosomal recessive 4A, 601277 (3), Autosomal recessive
<i>ABCA2</i>	600047	Intellectual developmental disorder with poor growth and with or without seizures or ataxia, 618808 (3), Autosomal recessive
<i>ABCA3</i>	601615	Surfactant metabolism dysfunction, pulmonary, 3, 610921 (3), Autosomal recessive
<i>ABCA4</i>	601691	Retinal dystrophy, early-onset severe, 248200 (3), Autosomal recessive; Stargardt disease 1, 248200 (3), Autosomal recessive; Fundus flavimaculatus, 248200 (3), Autosomal recessive; {Macular degeneration, age-related, 2}, 153800 (3), Autosomal dominant; Cone-rod dystrophy 3, 604116 (3); Retinitis pigmentosa 19, 601718 (3), Autosomal recessive
<i>ABCA5</i>	612503	?Hypertrichosis, congenital generalized, with gingival hyperplasia, 135400 (3), Autosomal recessive
<i>ABCA7</i>	605414	{Alzheimer disease 9, susceptibility to}, 608907 (3), Autosomal dominant

<i>ABCB1</i>	171050	{Inflammatory bowel disease 13}, 612244 (3); {Colchicine resistance}, 120080 (3)
<i>ABCB11</i>	603201	Cholestasis, progressive familial intrahepatic 2, 601847 (3), Autosomal recessive; Cholestasis, benign recurrent intrahepatic, 2, 605479 (3), Autosomal recessive
<i>ABCB4</i>	171060	Gallbladder disease 1, 600803 (3), Autosomal recessive, Autosomal dominant; Cholestasis, intrahepatic, of pregnancy, 3, 614972 (3), Autosomal recessive, Autosomal dominant; Cholestasis, progressive familial intrahepatic 3, 602347 (3), Autosomal recessive
<i>ABCB6</i>	605452	Dyschromatosis universalis hereditaria 3, 615402 (3), Autosomal dominant; Microphthalmia, isolated, with coloboma 7, 614497 (3), Autosomal dominant; [Blood group, Langereis system], 111600 (3); Pseudohyperkalemia, familial, 2, due to red cell leak, 609153 (3), Autosomal dominant
<i>ABCB7</i>	300135	Anemia, sideroblastic, with ataxia, 301310 (3), X-linked recessive
<i>ABCC1</i>	158343	?Deafness, autosomal dominant 77, 618915 (3), Autosomal dominant
<i>ABCC11</i>	607040	[Colostrum secretion, variation in], 117800 (3), Autosomal dominant; [Earwax, wet/dry], 117800 (3), Autosomal dominant; [Axillary odor, variation in], 117800 (3), Autosomal dominant
<i>ABCC2</i>	601107	Dubin-Johnson syndrome, 237500 (3), Autosomal recessive
<i>ABCC6</i>	603234	Pseudoxanthoma elasticum, 264800 (3), Autosomal recessive; Pseudoxanthoma elasticum, forme fruste, 177850 (3), Autosomal dominant; Arterial calcification, generalized, of infancy, 2, 614473 (3), Autosomal recessive
<i>ABCC8</i>	600509	Diabetes mellitus, noninsulin-dependent, 125853 (3), Autosomal dominant; Diabetes mellitus, permanent neonatal 3, with or without neurologic features, 618857 (3), Autosomal recessive, Autosomal dominant; Diabetes mellitus, transient neonatal 2, 610374 (3); Hyperinsulinemic hypoglycemia, familial, 1, 256450 (3), Autosomal recessive, Autosomal dominant; Hypoglycemia of infancy, leucine-sensitive, 240800 (3), Autosomal dominant
<i>ABCC9</i>	601439	Hypertrichotic osteochondrodysplasia, 239850 (3), Autosomal dominant; ?Atrial fibrillation, familial, 12, 614050 (3), Autosomal dominant; Cardiomyopathy, dilated, 10, 608569 (3), Autosomal dominant
<i>ABCD1</i>	300371	Adrenomyeloneuropathy, adult, 300100 (3), X-linked recessive; Adrenoleukodystrophy, 300100 (3), X-linked recessive
<i>ABCD3</i>	170995	?Bile acid synthesis defect, congenital, 5, 616278 (3), Autosomal recessive
<i>ABCD4</i>	603214	Methylmalonic aciduria and homocystinuria, cblJ type, 614857 (3), Autosomal recessive
<i>ABCG2</i>	603756	[Junior blood group system], 614490 (3); [Uric acid concentration, serum, QTL1], 138900 (3), ?Autosomal dominant
<i>ABCG5</i>	605459	Sitosterolemia 2, 618666 (3)

<i>ABCG8</i>	605460	{Gallbladder disease 4}, 611465 (3); Sitosterolemia 1, 210250 (3), Autosomal recessive
<i>ABHD12</i>	613599	Polyneuropathy, hearing loss, ataxia, retinitis pigmentosa, and cataract, 612674 (3), Autosomal recessive
<i>ABHD5</i>	604780	Chanarin-Dorfman syndrome, 275630 (3), Autosomal recessive
<i>ABL1</i>	189980	Leukemia, Philadelphia chromosome-positive, resistant to imatinib, 608232 (3), Somatic mutation; Congenital heart defects and skeletal malformations syndrome, 617602 (3), Autosomal dominant
<i>ABO</i>	110300	[Blood group, ABO system], 616093 (3)
<i>ACACA</i>	200350	Acetyl-CoA carboxylase deficiency, 613933 (1), Autosomal recessive
<i>ACAD8</i>	604773	Isobutyryl-CoA dehydrogenase deficiency, 611283 (3), Autosomal recessive
<i>ACAD9</i>	611103	Mitochondrial complex I deficiency, nuclear type 20, 611126 (3), Autosomal recessive
<i>ACADM</i>	607008	Acyl-CoA dehydrogenase, medium chain, deficiency of, 201450 (3), Autosomal recessive
<i>ACADS</i>	606885	Acyl-CoA dehydrogenase, short-chain, deficiency of, 201470 (3), Autosomal recessive
<i>ACADSB</i>	600301	2-methylbutyrylglycinuria, 610006 (3), Autosomal recessive
<i>ACADVL</i>	609575	VLCAD deficiency, 201475 (3), Autosomal recessive
<i>ACAN</i>	155760	Short stature and advanced bone age, with or without early-onset osteoarthritis and/or osteochondritis dissecans, 165800 (3), Autosomal dominant; Spondyloepimetaphyseal dysplasia, aggrecan type, 612813 (3), Autosomal recessive; ?Spondyloepiphyseal dysplasia, Kimberley type, 608361 (3), Autosomal dominant
<i>ACAT1</i>	607809	Alpha-methylacetoacetic aciduria, 203750 (3), Autosomal recessive
<i>ACAT2</i>	100678	?ACAT2 deficiency, 614055 (1), Isolated cases
<i>ACBD5</i>	616618	Retinal dystrophy with leukodystrophy, 618863 (3), Autosomal recessive
<i>ACD</i>	609377	?Dyskeratosis congenita, autosomal dominant 6, 616553 (3), Autosomal recessive, Autosomal dominant; ?Dyskeratosis congenita, autosomal recessive 7, 616553 (3), Autosomal recessive, Autosomal dominant
<i>ACE</i>	106180	Renal tubular dysgenesis, 267430 (3), Autosomal recessive; {Myocardial infarction, susceptibility to} (3); {Microvascular complications of diabetes 3}, 612624 (3); [Angiotensin I-converting enzyme, benign serum increase] (3); {SARS, progression of} (3); {Stroke, hemorrhagic}, 614519 (3)
<i>ACER3</i>	617036	?Leukodystrophy, progressive, early childhood-onset, 617762 (3), Autosomal recessive
<i>ACHE</i>	100740	[Blood group, Yt system], 112100 (3)

<i>ACKR1</i>	613665	{Malaria, vivax, protection against}, 611162 (3); [Blood group, Duffy system], 110700 (3), Autosomal recessive, Autosomal dominant; [White blood cell count QTL], 611862 (3), Autosomal recessive
<i>ACO2</i>	100850	Infantile cerebellar-retinal degeneration, 614559 (3), Autosomal recessive; ?Optic atrophy 9, 616289 (3), Autosomal recessive
<i>ACOX1</i>	609751	Mitchell syndrome, 618960 (3); Peroxisomal acyl-CoA oxidase deficiency, 264470 (3), Autosomal recessive
<i>ACOX2</i>	601641	Bile acid synthesis defect, congenital, 6, 617308 (3), Autosomal recessive
<i>ACP2</i>	171650	?Lysosomal acid phosphatase deficiency, 200950 (1), Autosomal recessive
<i>ACP4</i>	606362	Amelogenesis imperfecta, type IJ, 617297 (3), Autosomal recessive
<i>ACP5</i>	171640	Spondyloenchondrodysplasia with immune dysregulation, 607944 (3), Autosomal recessive
<i>ACR</i>	102480	?Male infertility due to acrosin deficiency, 102480 (2)
<i>ACSF3</i>	614245	Combined malonic and methylmalonic aciduria, 614265 (3)
<i>ACSL4</i>	300157	Mental retardation, X-linked 63, 300387 (3), X-linked dominant
<i>ACSL6</i>	604443	Myelodysplastic syndrome (3); Myelogenous leukemia, acute (3)
<i>ACSM3</i>	145505	{?Hypertension, essential} (1)
<i>ACTA1</i>	102610	Myopathy, actin, congenital, with cores, 161800 (3), Autosomal recessive, Autosomal dominant; Nemaline myopathy 3, autosomal dominant or recessive, 161800 (3), Autosomal recessive, Autosomal dominant; Myopathy, congenital, with fiber-type disproportion 1, 255310 (3), Autosomal recessive, Autosomal dominant; Myopathy, actin, congenital, with excess of thin myofilaments, 161800 (3), Autosomal recessive, Autosomal dominant; ?Myopathy, scapulohumeroperoneal, 616852 (3), Autosomal dominant
<i>ACTA2</i>	102620	Aortic aneurysm, familial thoracic 6, 611788 (3), Autosomal dominant; Multisystemic smooth muscle dysfunction syndrome, 613834 (3), Autosomal dominant; Moyamoya disease 5, 614042 (3)
<i>ACTB</i>	102630	?Dystonia, juvenile-onset, 607371 (3), Autosomal dominant; Baraitser-Winter syndrome 1, 243310 (3), Autosomal dominant
<i>ACTC1</i>	102540	Left ventricular noncompaction 4, 613424 (3), Autosomal dominant; Atrial septal defect 5, 612794 (3), Autosomal dominant; Cardiomyopathy, dilated, 1R, 613424 (3), Autosomal dominant; Cardiomyopathy, hypertrophic, 11, 612098 (3), Autosomal dominant
<i>ACTG1</i>	102560	Baraitser-Winter syndrome 2, 614583 (3), Autosomal dominant; Deafness, autosomal dominant 20/26, 604717 (3), Autosomal dominant
<i>ACTG2</i>	102545	Visceral myopathy, 155310 (3), Autosomal dominant

<i>ACTL6B</i>	612458	Intellectual developmental disorder with severe speech and ambulation defects, 618470 (3), Autosomal dominant; Epileptic encephalopathy, early infantile, 76, 618468 (3), Autosomal recessive
<i>ACTN1</i>	102575	Bleeding disorder, platelet-type, 15, 615193 (3), Autosomal dominant
<i>ACTN2</i>	102573	Myopathy, distal, 6, adult onset, 618655 (3), Autosomal dominant; Cardiomyopathy, dilated, 1AA, with or without LVNC, 612158 (3), Autosomal dominant; Cardiomyopathy, hypertrophic, 23, with or without LVNC, 612158 (3), Autosomal dominant; Myopathy, congenital with structured cores and Z-line abnormalities, 618654 (3), Autosomal dominant
<i>ACTN3</i>	102574	[Alpha-actinin-3 deficiency], 617749 (3); [Sprinting performance], 617749 (3)
<i>ACTN4</i>	604638	Glomerulosclerosis, focal segmental, 1, 603278 (3), Autosomal dominant
<i>ACVR1</i>	102576	Fibrodysplasia ossificans progressiva, 135100 (3), Autosomal dominant
<i>ACVR1B</i>	601300	Pancreatic cancer, somatic (3)
<i>ACVR2B</i>	602730	Heterotaxy, visceral, 4, autosomal, 613751 (3)
<i>ACVRL1</i>	601284	Telangiectasia, hereditary hemorrhagic, type 2, 600376 (3), Autosomal dominant
<i>ACY1</i>	104620	Aminoacylase 1 deficiency, 609924 (3), Autosomal recessive
<i>ADA</i>	608958	Adenosine deaminase deficiency, partial, 102700 (3), Autosomal recessive, Somatic mosaicism; Severe combined immunodeficiency due to ADA deficiency, 102700 (3), Autosomal recessive, Somatic mosaicism
<i>ADA2</i>	607575	Vasculitis, autoinflammation, immunodeficiency, and hematologic defects syndrome, 615688 (3), Autosomal recessive; ?Sneddon syndrome, 182410 (3), Autosomal recessive
<i>ADAM10</i>	602192	{Alzheimer disease 18, susceptibility to}, 615590 (3); Reticulate acropigmentation of Kitamura, 615537 (3), Autosomal dominant
<i>ADAM17</i>	603639	?Inflammatory skin and bowel disease, neonatal, 1, 614328 (3), Autosomal recessive
<i>ADAM22</i>	603709	?Epileptic encephalopathy, early infantile, 61, 617933 (3), Autosomal recessive
<i>ADAM9</i>	602713	Cone-rod dystrophy 9, 612775 (3), Autosomal recessive
<i>ADAMTS10</i>	608990	Weill-Marchesani syndrome 1, recessive, 277600 (3), Autosomal recessive
<i>ADAMTS13</i>	604134	Thrombotic thrombocytopenic purpura, hereditary, 274150 (3), Autosomal recessive
<i>ADAMTS17</i>	607511	Weill-Marchesani 4 syndrome, recessive, 613195 (3), Autosomal recessive
<i>ADAMTS18</i>	607512	Microcornea, myopic chorioretinal atrophy, and telecanthus, 615458 (3), Autosomal recessive

<i>ADAMTS2</i>	604539	Ehlers-Danlos syndrome, dermatosparaxis type, 225410 (3), Autosomal recessive
<i>ADAMTS3</i>	605011	Hennekam lymphangiectasia-lymphedema syndrome 3, 618154 (3), Autosomal recessive
<i>ADAMTSL2</i>	612277	Geleophysic dysplasia 1, 231050 (3), Autosomal recessive
<i>ADAMTSL4</i>	610113	Ectopia lentis et pupillae, 225200 (3), Autosomal recessive; Ectopia lentis, isolated, autosomal recessive, 225100 (3), Autosomal recessive
<i>ADAR</i>	146920	Dyschromatosis symmetrica hereditaria, 127400 (3), Autosomal dominant; Aicardi-Goutieres syndrome 6, 615010 (3), Autosomal recessive
<i>ADARB1</i>	601218	Neurodevelopmental disorder with hypotonia, microcephaly, and seizures, 618862 (3), Autosomal recessive
<i>ADAT3</i>	615302	Mental retardation, autosomal recessive 36, 615286 (3), Autosomal recessive
<i>ADCY1</i>	103072	?Deafness, autosomal recessive 44, 610154 (3), Autosomal recessive
<i>ADCY10</i>	605205	{Hypercalciuria, absorptive, susceptibility to}, 143870 (3), Autosomal dominant
<i>ADCY3</i>	600291	{Obesity, susceptibility to, BMIQ19}, 617885 (3), Autosomal recessive
<i>ADCY5</i>	600293	Dyskinesia, familial, with facial myokymia, 606703 (3), Autosomal dominant
<i>ADCY6</i>	600294	?Lethal congenital contracture syndrome 8, 616287 (3), Autosomal recessive
<i>ADD1</i>	102680	{Hypertension, essential, salt-sensitive}, 145500 (3), Multifactorial
<i>ADD3</i>	601568	Cerebral palsy, spastic quadriplegic, 3, 617008 (3), Autosomal recessive
<i>ADGRE2</i>	606100	Vibratory urticaria, 125630 (3), Autosomal dominant
<i>ADGRG1</i>	604110	Polymicrogyria, bilateral perisylvian, 615752 (3); Polymicrogyria, bilateral frontoparietal, 606854 (3), Autosomal recessive
<i>ADGRG2</i>	300572	Congenital bilateral absence of vas deferens, X-linked, 300985 (3), X-linked
<i>ADGRG6</i>	612243	Lethal congenital contracture syndrome 9, 616503 (3), Autosomal recessive
<i>ADGRV1</i>	602851	Usher syndrome, type 2C, 605472 (3), Autosomal recessive, Digenic dominant; ?Febrile seizures, familial, 4, 604352 (3), Autosomal dominant; Usher syndrome, type 2C, GPR98/PDZD7 digenic, 605472 (3), Autosomal recessive, Digenic dominant
<i>ADH1B</i>	103720	{Alcohol dependence, protection against}, 103780 (3), Multifactorial; {Aerodigestive tract cancer, squamous cell, alcohol-related, protection against}, 103780 (3), Multifactorial
<i>ADH1C</i>	103730	{Parkinson disease, susceptibility to}, 168600 (3), Autosomal dominant, Multifactorial; {Alcohol dependence, protection against}, 103780 (3), Multifactorial

<i>ADIPOQ</i>	605441	Adiponectin deficiency, 612556 (3)
<i>ADK</i>	102750	Hypermethioninemia due to adenosine kinase deficiency, 614300 (3), Autosomal recessive
<i>ADNP</i>	611386	Helsmoortel-van der Aa syndrome, 615873 (3), Autosomal dominant
<i>ADPRS</i> (<i>ADPRHL2</i>)	610624	Neurodegeneration, childhood-onset, stress-induced, with variable ataxia and seizures, 618170 (3), Autosomal recessive
<i>ADRA2C</i>	104250	{Congestive heart failure and beta-blocker response, modifier of} (3)
<i>ADRB1</i>	109630	[Resting heart rate], 607276 (3); [Short sleep, familial natural, 2], 618591 (3), Autosomal dominant
<i>ADRB2</i>	109690	{Asthma, nocturnal, susceptibility to}, 600807 (3), Autosomal dominant; {Obesity, susceptibility to}, 601665 (3), Autosomal recessive, Autosomal dominant, Multifactorial; Beta-2-adrenoreceptor agonist, reduced response to (3)
<i>ADRB3</i>	109691	{Obesity, susceptibility to}, 601665 (3), Autosomal recessive, Autosomal dominant, Multifactorial
<i>ADSL</i>	608222	Adenylosuccinase deficiency, 103050 (3), Autosomal recessive
<i>ADSS1</i> (<i>ADSSL1</i>)	612498	Myopathy, distal, 5, 617030 (3), Autosomal recessive
<i>AEBP1</i>	602981	Ehlers-Danlos syndrome, classic-like, 2, 618000 (3), Autosomal recessive
<i>AFF2</i>	300806	Mental retardation, X-linked, FRAXE type, 309548 (3), X-linked recessive
<i>AFF4</i>	604417	CHOPS syndrome, 616368 (3), Autosomal dominant
<i>AFG3L2</i>	604581	Spastic ataxia 5, autosomal recessive, 614487 (3), Autosomal recessive; Optic atrophy 12, 618977 (3); Spinocerebellar ataxia 28, 610246 (3), Autosomal dominant
<i>AFP</i>	104150	Alpha-fetoprotein deficiency, 615969 (3), Autosomal recessive; [Hereditary persistence of alpha-fetoprotein], 615970 (3), Autosomal dominant
<i>AGA</i>	613228	Aspartylglucosaminuria, 208400 (3), Autosomal recessive
<i>AGBL1</i>	615496	Corneal dystrophy, Fuchs endothelial, 8, 615523 (3), Autosomal dominant
<i>AGBL5</i>	615900	Retinitis pigmentosa 75, 617023 (3), Autosomal recessive
<i>AGK</i>	610345	Sengers syndrome, 212350 (3), Autosomal recessive; Cataract 38, autosomal recessive, 614691 (3), Autosomal recessive
<i>AGL</i>	610860	Glycogen storage disease IIb, 232400 (3), Autosomal recessive; Glycogen storage disease IIIa, 232400 (3), Autosomal recessive
<i>AGPAT2</i>	603100	Lipodystrophy, congenital generalized, type 1, 608594 (3), Autosomal recessive
<i>AGPS</i>	603051	Rhizomelic chondrodysplasia punctata, type 3, 600121 (3), Autosomal recessive
<i>AGRN</i>	103320	Myasthenic syndrome, congenital, 8, with pre- and postsynaptic defects, 615120 (3), Autosomal recessive

<i>AGRP</i>	602311	{Obesity, late-onset}, 601665 (3), Autosomal recessive, Autosomal dominant, Multifactorial; {Leanness, inherited}, 601665 (3), Autosomal recessive, Autosomal dominant, Multifactorial
<i>AGT</i>	106150	{Preeclampsia, susceptibility to} (3); Renal tubular dysgenesis, 267430 (3), Autosomal recessive; {Hypertension, essential, susceptibility to}, 145500 (3), Multifactorial
<i>AGTPBP1</i>	606830	Neurodegeneration, childhood-onset, with cerebellar atrophy, 618276 (3), Autosomal recessive
<i>AGTR1</i>	106165	Renal tubular dysgenesis, 267430 (3), Autosomal recessive; {Hypertension, essential}, 145500 (3), Multifactorial
<i>AGXT</i>	604285	Hyperoxaluria, primary, type 1, 259900 (3), Autosomal recessive
<i>AGXT2</i>	612471	[Beta-aminoisobutyric acid, urinary excretion of], 210100 (3), Autosomal recessive
<i>AHCY</i>	180960	Hypermethioninemia with deficiency of S-adenosylhomocysteine hydrolase, 613752 (3), Autosomal recessive
<i>AHDC1</i>	615790	Xia-Gibbs syndrome, 615829 (3), Autosomal dominant
<i>AHI1</i>	608894	Joubert syndrome 3, 608629 (3), Autosomal recessive
<i>AHR</i>	600253	?Retinitis pigmentosa 85, 618345 (3), Autosomal recessive
<i>AHSG</i>	138680	?Alopecia-mental retardation syndrome 1, 203650 (3), Autosomal recessive
<i>AICDA</i>	605257	Immunodeficiency with hyper-IgM, type 2, 605258 (3), Autosomal recessive
<i>AIFM1</i>	300169	Cowchock syndrome, 310490 (3), X-linked recessive; Spondyloepimetaphyseal dysplasia, X-linked, with hypomyelinating leukodystrophy, 300232 (3), X-linked recessive; Combined oxidative phosphorylation deficiency 6, 300816 (3), X-linked recessive; Deafness, X-linked 5, 300614 (3), X-linked recessive
<i>AIMP1</i>	603605	Leukodystrophy, hypomyelinating, 3, 260600 (3), Autosomal recessive
<i>AIMP2</i>	600859	Leukodystrophy, hypomyelinating, 17, 618006 (3), Autosomal recessive
<i>AIP</i>	605555	Pituitary adenoma 1, multiple types, 102200 (3), Autosomal dominant, Somatic mutation; Pituitary adenoma predisposition, 102200 (3), Autosomal dominant, Somatic mutation
<i>AIPL1</i>	604392	Leber congenital amaurosis 4, 604393 (3), Autosomal recessive, Autosomal dominant; Retinitis pigmentosa, juvenile, 604393 (3), Autosomal recessive, Autosomal dominant; Cone-rod dystrophy, 604393 (3), Autosomal recessive, Autosomal dominant
<i>AIRE</i>	607358	Autoimmune polyendocrinopathy syndrome, type I, with or without reversible metaphyseal dysplasia, 240300 (3), Autosomal recessive, Autosomal dominant
<i>AK1</i>	103000	Hemolytic anemia due to adenylate kinase deficiency, 612631 (3), Autosomal recessive
<i>AK2</i>	103020	Reticular dysgenesis, 267500 (3), Autosomal recessive

<i>AK7</i>	615364	?Spermatogenic failure 27, 617965 (3), Autosomal recessive
<i>AKAP10</i>	604694	{Cardiac conduction defect, susceptibility to}, 115080 (3), Autosomal dominant
<i>AKAP9</i>	604001	?Long QT syndrome 11, 611820 (3), Autosomal dominant
<i>AKR1C2</i>	600450	46XY sex reversal 8, 614279 (3), Autosomal recessive
<i>AKR1C4</i>	600451	{46XY sex reversal 8, modifier of}, 614279 (3), Autosomal recessive
<i>AKR1D1</i>	604741	Bile acid synthesis defect, congenital, 2, 235555 (3), Autosomal recessive
<i>AKT1</i>	164730	Breast cancer, somatic, 114480 (3); Cowden syndrome 6, 615109 (3); Proteus syndrome, somatic, 176920 (3); Ovarian cancer, somatic, 167000 (3); Colorectal cancer, somatic, 114500 (3)
<i>AKT2</i>	164731	Hypoinsulinemic hypoglycemia with hemihypertrophy, 240900 (3), Autosomal dominant; Diabetes mellitus, type II, 125853 (3), Autosomal dominant
<i>AKT3</i>	611223	Megalencephaly-polymicrogyria-polydactyly-hydrocephalus syndrome 2, 615937 (3), Autosomal dominant
<i>AL032819.3</i>	No OMIM gene	No OMIM phenotype
<i>ALAD</i>	125270	Porphyria, acute hepatic, 612740 (3), Autosomal recessive; {Lead poisoning, susceptibility to}, 612740 (3), Autosomal recessive
<i>ALAS2</i>	301300	Protoporphyrin, erythropoietic, X-linked, 300752 (3), X-linked; Anemia, sideroblastic, 1, 300751 (3), X-linked recessive
<i>ALB</i>	103600	[Dysalbuminemic hyperthyroxinemia], 615999 (3); Analbuminemia, 616000 (3)
<i>ALDH18A1</i>	138250	Cutis laxa, autosomal recessive, type IIIA, 219150 (3), Autosomal recessive; Cutis laxa, autosomal dominant 3, 616603 (3), Autosomal dominant; Spastic paraplegia 9B, autosomal recessive, 616586 (3), Autosomal recessive; Spastic paraplegia 9A, autosomal dominant, 601162 (3), Autosomal dominant
<i>ALDH1A3</i>	600463	Microphthalmia, isolated 8, 615113 (3), Autosomal recessive
<i>ALDH2</i>	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)
<i>ALDH3A2</i>	609523	Sjogren-Larsson syndrome, 270200 (3), Autosomal recessive
<i>ALDH4A1</i>	606811	Hyperprolinemia, type II, 239510 (3), Autosomal recessive
<i>ALDH5A1</i>	610045	Succinic semialdehyde dehydrogenase deficiency, 271980 (3), Autosomal recessive
<i>ALDH6A1</i>	603178	Methylmalonate semialdehyde dehydrogenase deficiency, 614105 (3), Autosomal recessive
<i>ALDH7A1</i>	107323	Epilepsy, pyridoxine-dependent, 266100 (3), Autosomal recessive
<i>ALDOA</i>	103850	Glycogen storage disease XII, 611881 (3), Autosomal recessive
<i>ALDOB</i>	612724	Fructose intolerance, hereditary, 229600 (3), Autosomal recessive

<i>ALG1</i>	605907	Congenital disorder of glycosylation, type Ik, 608540 (3), Autosomal recessive
<i>ALG10B</i>	603313	{Long QT syndrome, acquired, reduced susceptibility to}, 613688 (3), Autosomal dominant
<i>ALG11</i>	613666	Congenital disorder of glycosylation, type Ip, 613661 (3), Autosomal recessive
<i>ALG12</i>	607144	Congenital disorder of glycosylation, type Ig, 607143 (3), Autosomal recessive
<i>ALG13</i>	300776	Epileptic encephalopathy, early infantile, 36, 300884 (3), X-linked dominant; ?Congenital disorder of glycosylation, type Is, 300884 (3), X-linked dominant
<i>ALG14</i>	612866	?Myasthenic syndrome, congenital, 15, without tubular aggregates, 616227 (3), Autosomal recessive
<i>ALG2</i>	607905	Myasthenic syndrome, congenital, 14, with tubular aggregates, 616228 (3), Autosomal recessive; ?Congenital disorder of glycosylation, type Ii, 607906 (3), Autosomal recessive
<i>ALG3</i>	608750	Congenital disorder of glycosylation, type Id, 601110 (3), Autosomal recessive
<i>ALG6</i>	604566	Congenital disorder of glycosylation, type Ic, 603147 (3), Autosomal recessive
<i>ALG8</i>	608103	Congenital disorder of glycosylation, type Ih, 608104 (3), Autosomal recessive; Polycystic liver disease 3 with or without kidney cysts, 617874 (3), Autosomal dominant
<i>ALG9</i>	606941	Gillessen-Kaesbach-Nishimura syndrome, 263210 (3), Autosomal recessive; Congenital disorder of glycosylation, type Il, 608776 (3), Autosomal recessive
<i>ALK</i>	105590	{Neuroblastoma, susceptibility to, 3}, 613014 (3)
<i>ALKBH8</i>	613306	Intellectual developmental disorder, autosomal recessive 71, 618504 (3), Autosomal recessive
<i>ALMS1</i>	606844	Alstrom syndrome, 203800 (3), Autosomal recessive
<i>ALOX12B</i>	603741	Ichthyosis, congenital, autosomal recessive 2, 242100 (3), Autosomal recessive
<i>ALOX5</i>	152390	{Atherosclerosis, susceptibility to} (3); {Asthma, diminished response to antileukotriene treatment in}, 600807 (3), Autosomal dominant
<i>ALOX5AP</i>	603700	{Stroke, susceptibility to}, 601367 (3), Multifactorial
<i>ALOXE3</i>	607206	Ichthyosis, congenital, autosomal recessive 3, 606545 (3), Autosomal recessive
<i>ALPK3</i>	617608	Cardiomyopathy, familial hypertrophic 27, 618052 (3), Autosomal recessive
<i>ALPL</i>	171760	Hypophosphatasia, adult, 146300 (3), Autosomal recessive, Autosomal dominant; Odontohypophosphatasia, 146300 (3), Autosomal recessive, Autosomal dominant; Hypophosphatasia, childhood, 241510 (3), Autosomal recessive; Hypophosphatasia, infantile, 241500 (3), Autosomal recessive

<i>ALS2</i>	606352	Primary lateral sclerosis, juvenile, 606353 (3), Autosomal recessive; Amyotrophic lateral sclerosis 2, juvenile, 205100 (3), Autosomal recessive; Spastic paralysis, infantile onset ascending, 607225 (3), Autosomal recessive
<i>ALX1</i>	601527	Frontonasal dysplasia 3, 613456 (3), Autosomal recessive
<i>ALX3</i>	606014	Frontonasal dysplasia 1, 136760 (3), Autosomal recessive
<i>ALX4</i>	605420	Frontonasal dysplasia 2, 613451 (3), Autosomal recessive; Parietal foramina 2, 609597 (3), Autosomal dominant; {Craniosynostosis 5, susceptibility to}, 615529 (3), Autosomal dominant
<i>AMACR</i>	604489	Bile acid synthesis defect, congenital, 4, 214950 (3), Autosomal recessive; Alpha-methylacyl-CoA racemase deficiency, 614307 (3), Autosomal recessive
<i>AMBN</i>	601259	Amelogenesis imperfecta, type IF, 616270 (3), Autosomal recessive
<i>AMELX</i>	300391	Amelogenesis imperfecta, type 1E, 301200 (3), X-linked dominant
<i>AMER1</i>	300647	Osteopathia striata with cranial sclerosis, 300373 (3), X-linked dominant
<i>AMH</i>	600957	Persistent Mullerian duct syndrome, type I, 261550 (3), Autosomal recessive
<i>AMHR2</i>	600956	Persistent Mullerian duct syndrome, type II, 261550 (3), Autosomal recessive
<i>AMMECR1</i>	300195	Midface hypoplasia, hearing impairment, elliptocytosis, and nephrocalcinosis, 300990 (3), X-linked recessive
<i>AMN</i>	605799	Imerslund-Grasbeck syndrome 2, 618882 (3), Autosomal recessive
<i>AMPD1</i>	102770	Myopathy due to myoadenylate deaminase deficiency, 615511 (3), Autosomal recessive
<i>AMPD2</i>	102771	?Spastic paraplegia 63, 615686 (3), Autosomal recessive; Pontocerebellar hypoplasia, type 9, 615809 (3), Autosomal recessive
<i>AMPD3</i>	102772	[AMP deaminase deficiency, erythrocytic], 612874 (3), Autosomal recessive
<i>AMT</i>	238310	Glycine encephalopathy, 605899 (3), Autosomal recessive
<i>AMTN</i>	610912	?Amelogenesis imperfecta, type IIIB, 617607 (3), Autosomal dominant
<i>ANAPC1</i>	608473	Rothmund-Thomson syndrome, type 1, 618625 (3), Autosomal recessive
<i>ANG</i>	105850	Amyotrophic lateral sclerosis 9, 611895 (3)
<i>ANGPTL3</i>	604774	Hypobetalipoproteinemia, familial, 2, 605019 (3), Autosomal recessive
<i>ANGPTL4</i>	605910	Plasma triglyceride level QTL, low, 615881 (3), Autosomal dominant
<i>ANK1</i>	612641	Spherocytosis, type 1, 182900 (3), Autosomal recessive, Autosomal dominant

<i>ANK2</i>	106410	Cardiac arrhythmia, ankyrin-B-related, 600919 (3), Autosomal dominant; Long QT syndrome 4, 600919 (3), Autosomal dominant
<i>ANK3</i>	600465	Mental retardation, autosomal recessive, 37, 615493 (3), Autosomal recessive
<i>ANKH</i>	605145	Craniometaphyseal dysplasia, 123000 (3), Autosomal dominant; Chondrocalcinosis 2, 118600 (3), Autosomal dominant
<i>ANKLE2</i>	616062	Microcephaly 16, primary, autosomal recessive, 616681 (3), Autosomal recessive
<i>ANKRD11</i>	611192	KBG syndrome, 148050 (3), Autosomal dominant
<i>ANKRD26</i>	610855	Thrombocytopenia 2, 188000 (3), Autosomal dominant
<i>ANKS6</i>	615370	Nephronophthisis 16, 615382 (3), Autosomal recessive
<i>ANLN</i>	616027	Focal segmental glomerulosclerosis 8, 616032 (3), Autosomal dominant
<i>ANO10</i>	613726	Spinocerebellar ataxia, autosomal recessive 10, 613728 (3), Autosomal recessive
<i>ANO3</i>	610110	Dystonia 24, 615034 (3), Autosomal dominant
<i>ANO5</i>	608662	Miyoshi muscular dystrophy 3, 613319 (3), Autosomal recessive; Muscular dystrophy, limb-girdle, autosomal recessive 12, 611307 (3), Autosomal recessive; Gnathodiaphyseal dysplasia, 166260 (3), Autosomal dominant
<i>ANO6</i>	608663	Scott syndrome, 262890 (3), Autosomal recessive
<i>ANOS1</i>	300836	Hypogonadotropic hypogonadism 1 with or without anosmia (Kallmann syndrome 1), 308700 (3), X-linked recessive
<i>ANTXR1</i>	606410	GAPO syndrome, 230740 (3), Autosomal recessive; {?Hemangioma, capillary infantile, susceptibility to}, 602089 (3), Autosomal dominant
<i>ANTXR2</i>	608041	Hyaline fibromatosis syndrome, 228600 (3), Autosomal recessive
<i>ANXA11</i>	602572	Amyotrophic lateral sclerosis 23, 617839 (3), Autosomal dominant
<i>ANXA5</i>	131230	{Pregnancy loss, recurrent, susceptibility to, 3}, 614391 (3), Autosomal dominant
<i>AP1B1</i>	600157	Keratitis-ichthyosis-deafness syndrome, autosomal recessive, 242150 (3), Autosomal recessive
<i>AP1S1</i>	603531	MEDNIK syndrome, 609313 (3), Autosomal recessive
<i>AP1S2</i>	300629	Mental retardation, X-linked syndromic 5, 304340 (3), X-linked recessive
<i>AP1S3</i>	615781	{Psoriasis 15, pustular, susceptibility to}, 616106 (3), Autosomal dominant
<i>AP2M1</i>	601024	Intellectual developmental disorder 60 with seizures, 618587 (3), Autosomal dominant
<i>AP2S1</i>	602242	Hypocalciuric hypercalcemia, type III, 600740 (3), Autosomal dominant
<i>AP3B1</i>	603401	Hermansky-Pudlak syndrome 2, 608233 (3), Autosomal recessive
<i>AP3B2</i>	602166	Epileptic encephalopathy, early infantile, 48, 617276 (3), Autosomal recessive

<i>AP3D1</i>	607246	?Hermansky-Pudlak syndrome 10, 617050 (3), Autosomal recessive
<i>AP4B1</i>	607245	Spastic paraplegia 47, autosomal recessive, 614066 (3), Autosomal recessive
<i>AP4E1</i>	607244	Stuttering, familial persistent, 1, 184450 (3), Autosomal dominant; Spastic paraplegia 51, autosomal recessive, 613744 (3), Autosomal recessive
<i>AP4M1</i>	602296	Spastic paraplegia 50, autosomal recessive, 612936 (3), Autosomal recessive
<i>AP4S1</i>	607243	Spastic paraplegia 52, autosomal recessive, 614067 (3), Autosomal recessive
<i>AP5Z1</i>	613653	Spastic paraplegia 48, autosomal recessive, 613647 (3), Autosomal recessive
<i>APC</i>	611731	Adenoma, periampullary, somatic (3); Desmoid disease, hereditary, 135290 (3), Autosomal dominant; Adenomatous polyposis coli, 175100 (3), Autosomal dominant; Gardner syndrome, 175100 (3), Autosomal dominant; Hepatoblastoma, somatic, 114550 (3); Colorectal cancer, somatic, 114500 (3); Brain tumor-polypoid syndrome 2, 175100 (3), Autosomal dominant; Gastric cancer, somatic, 613659 (3)
<i>APC2</i>	612034	?Sotos syndrome 3, 617169 (3), Autosomal recessive; Cortical dysplasia, complex, with other brain malformations 10, 618677 (3), Autosomal recessive
<i>APCDD1</i>	607479	Hypotrichosis 1, 605389 (3), Autosomal dominant
<i>APCS</i>	104770	{?Amyloidosis, secondary, susceptibility to} (1)
<i>APOA1</i>	107680	Hypoalphalipoproteinemia, primary, 2, with or without corneal clouding, 618463 (3); Amyloidosis, 3 or more types, 105200 (3), Autosomal dominant; ApoA-I and apoC-III deficiency, combined, 618463 (3)
<i>APOA2</i>	107670	Apolipoprotein A-II deficiency (3); {Hypercholesterolemia, familial, modifier of}, 143890 (3), Autosomal dominant
<i>APOA5</i>	606368	{Hypertriglyceridemia, susceptibility to}, 145750 (3), Autosomal dominant; Hyperchylomicronemia, late-onset, 144650 (3), Autosomal dominant
<i>APOB</i>	107730	Hypobetalipoproteinemia, 615558 (3), Autosomal recessive; Hypercholesterolemia, familial, 2, 144010 (3), Autosomal dominant
<i>APOC2</i>	608083	Hyperlipoproteinemia, type Ib, 207750 (3), Autosomal recessive
<i>APOC3</i>	107720	Apolipoprotein C-III deficiency, 614028 (3)
<i>APOE</i>	107741	Hyperlipoproteinemia, type III, 617347 (3); {Coronary artery disease, severe, susceptibility to}, 617347 (3); {?Alzheimer disease, protection against, due to APOE3-Christchurch}, 607822 (3), Autosomal dominant; Lipoprotein glomerulopathy, 611771 (3); Sea-blue histiocyte disease, 269600 (3), Autosomal recessive; {?Macular degeneration, age-related}, 603075 (3), Autosomal dominant; Alzheimer disease 2, 104310 (3), Autosomal dominant

<i>APOL1</i>	603743	{End-stage renal disease, nondiabetic, susceptibility to}, 612551 (3); {Glomerulosclerosis, focal segmental, 4, susceptibility to}, 612551 (3)
<i>APOL2</i>	607252	{Schizophrenia}, 181500 (1), Autosomal dominant
<i>APOL4</i>	607254	{Schizophrenia}, 181500 (1), Autosomal dominant
<i>APP</i>	104760	Cerebral amyloid angiopathy, Dutch, Italian, Iowa, Flemish, Arctic variants, 605714 (3), Autosomal dominant; Alzheimer disease 1, familial, 104300 (3), Autosomal dominant
<i>APPL1</i>	604299	{Maturity-onset diabetes of the young, type 14}, 616511 (3), Autosomal dominant
<i>APRT</i>	102600	Adenine phosphoribosyltransferase deficiency, 614723 (3), Autosomal recessive
<i>APTX</i>	606350	Ataxia, early-onset, with oculomotor apraxia and hypoalbuminemia, 208920 (3), Autosomal recessive
<i>AQP1</i>	107776	[Blood group, Colton], 110450 (3); [Aquaporin-1 deficiency], 110450 (3)
<i>AQP2</i>	107777	Diabetes insipidus, nephrogenic, 125800 (3), Autosomal recessive, Autosomal dominant
<i>AQP3</i>	600170	[Blood group GIL], 607457 (3)
<i>AQP5</i>	600442	Palmoplantar keratoderma, Bothnian type, 600231 (3), Autosomal dominant
<i>AQP7</i>	602974	[Glycerol quantitative trait locus], 614411 (3), Autosomal recessive
<i>AR</i>	313700	Hypospadias 1, X-linked, 300633 (3), X-linked recessive; Androgen insensitivity, 300068 (3), X-linked recessive; {Prostate cancer, susceptibility to}, 176807 (3), Autosomal dominant, Somatic mutation; Androgen insensitivity, partial, with or without breast cancer, 312300 (3), X-linked recessive; Spinal and bulbar muscular atrophy of Kennedy, 313200 (3), X-linked recessive
<i>ARCNI</i>	600820	Short stature, rhizomelic, with microcephaly, micrognathia, and developmental delay, 617164 (3), Autosomal dominant
<i>ARF1</i>	103180	Periventricular nodular heterotopia 8, 618185 (3), Autosomal dominant
<i>ARFGEF2</i>	605371	Periventricular heterotopia with microcephaly, 608097 (3), Autosomal recessive
<i>ARG1</i>	608313	Argininemia, 207800 (3), Autosomal recessive
<i>ARHGAP26</i>	605370	Leukemia, juvenile myelomonocytic, somatic, 607785 (3)
<i>ARHGAP31</i>	610911	Adams-Oliver syndrome 1, 100300 (3), Autosomal dominant
<i>ARHGDIA</i>	601925	Nephrotic syndrome, type 8, 615244 (3), Autosomal recessive
<i>ARHGEF1</i>	601855	?Immunodeficiency 62, 618459 (3), Autosomal recessive
<i>ARHGEF10</i>	608136	?Slowed nerve conduction velocity, AD, 608236 (3), Autosomal dominant
<i>ARHGEF18</i>	616432	Retinitis pigmentosa 78, 617433 (3), Autosomal recessive
<i>ARHGEF2</i>	607560	?Neurodevelopmental disorder with midbrain and hindbrain malformations, 617523 (3), Autosomal recessive

<i>ARHGEF6</i>	300267/300436	-/Mental retardation, X-linked 46, 300436 (2), X-linked recessive
<i>ARHGEF9</i>	300429	Epileptic encephalopathy, early infantile, 8, 300607 (3), X-linked recessive
<i>ARID1A</i>	603024	Coffin-Siris syndrome 2, 614607 (3), Autosomal dominant
<i>ARID1B</i>	614556	Coffin-Siris syndrome 1, 135900 (3), Autosomal dominant
<i>ARID2</i>	609539	Coffin-Siris syndrome 6, 617808 (3), Autosomal dominant
<i>ARL13B</i>	608922	Joubert syndrome 8, 612291 (3), Autosomal recessive
<i>ARL2BP</i>	615407	Retinitis pigmentosa with or without situs inversus, 615434 (3), Autosomal recessive
<i>ARL3</i>	604695	Joubert syndrome 35, 618161 (3), Autosomal recessive; Retinitis pigmentosa 83, 618173 (3), Autosomal dominant
<i>ARL6</i>	608845	?Retinitis pigmentosa 55, 613575 (3); Bardet-Biedl syndrome 3, 600151 (3), Autosomal recessive; {Bardet-Biedl syndrome 1, modifier of}, 209900 (3), Autosomal recessive, Digenic recessive
<i>ARL6IP1</i>	607669	?Spastic paraplegia 61, autosomal recessive, 615685 (3), Autosomal recessive
<i>ARMC2</i>	618424	Spermatogenic failure 38, 618433 (3), Autosomal recessive
<i>ARMC4</i>	615408	Ciliary dyskinesia, primary, 23, 615451 (3), Autosomal recessive
<i>ARMC5</i>	615549	ACTH-independent macronodular adrenal hyperplasia 2, 615954 (3), Autosomal dominant, Somatic mutation
<i>ARMC9</i>	617612	Joubert syndrome 30, 617622 (3), Autosomal recessive
<i>ARMS2</i>	611313	{Macular degeneration, age-related, 8}, 613778 (3)
<i>ARNT2</i>	606036	?Webb-Dattani syndrome, 615926 (3), Autosomal recessive
<i>ARPC1B</i>	604223	Immunodeficiency 71 with inflammatory disease and congenital thrombocytopenia, 617718 (3), Autosomal recessive
<i>ARR3</i>	301770	Myopia 26, X-linked, female-limited, 301010 (3), X-linked
<i>ARSA</i>	607574	Metachromatic leukodystrophy, 250100 (3), Autosomal recessive
<i>ARSB</i>	611542	Mucopolysaccharidosis type VI (Maroteaux-Lamy), 253200 (3), Autosomal recessive
<i>ARSG</i>	610008	Usher syndrome, type IV, 618144 (3), Autosomal recessive
<i>ARSL (ARSE)</i>	300180	Chondrodysplasia punctata, X-linked recessive, 302950 (3), X-linked recessive
<i>ART4</i>	110600	[Blood group, Dombrock], 616060 (3)
<i>ARV1</i>	611647	Epileptic encephalopathy, early infantile, 38, 617020 (3), Autosomal recessive
<i>ARX</i>	300382	Epileptic encephalopathy, early infantile, 1, 308350 (3), X-linked recessive; Lissencephaly, X-linked 2, 300215 (3), X-linked; Proud syndrome, 300004 (3), X-linked; Mental retardation, X-linked 29 and others, 300419 (3), X-linked recessive; Partington syndrome, 309510 (3), X-linked recessive; Hydranencephaly with abnormal genitalia, 300215 (3), X-linked
<i>ASAH1</i>	613468	Farber lipogranulomatosis, 228000 (3), Autosomal recessive; Spinal muscular atrophy with progressive myoclonic epilepsy, 159950 (3), Autosomal recessive

<i>ASB10</i>	615054	Glaucoma 1, open angle, F, 603383 (3)
<i>ASCC1</i>	614215	Spinal muscular atrophy with congenital bone fractures 2, 616867 (3), Autosomal recessive; Barrett esophagus/esophageal adenocarcinoma, 614266 (3)
<i>ASCL1</i>	100790	Haddad syndrome, 209880 (3), Autosomal dominant; Central hypoventilation syndrome, congenital, 209880 (3), Autosomal dominant
<i>ASH1L</i>	607999	Mental retardation, autosomal dominant 52, 617796 (3), Autosomal dominant
<i>ASIP</i>	600201	[Skin/hair/eye pigmentation 9, brown/nonbrown eyes], 611742 (3); [Skin/hair/eye pigmentation 9, dark/light hair], 611742 (3)
<i>ASL</i>	608310	Argininosuccinic aciduria, 207900 (3), Autosomal recessive
<i>ASNS</i>	108370	Asparagine synthetase deficiency, 615574 (3), Autosomal recessive
<i>ASPA</i>	608034	Canavan disease, 271900 (3), Autosomal recessive
<i>ASPH</i>	600582	Traboulsi syndrome, 601552 (3), Autosomal recessive
<i>ASPM</i>	605481	Microcephaly 5, primary, autosomal recessive, 608716 (3), Autosomal recessive
<i>ASPN</i>	608135	{Osteoarthritis susceptibility 3}, 607850 (3), Autosomal dominant; {Lumbar disc degeneration}, 603932 (3)
<i>ASPRV1</i>	611765	Ichthyosis, lamellar, autosomal dominant, 146750 (3), Autosomal dominant
<i>ASPSCR1</i>	606236	Alveolar soft-part sarcoma, 606243 (3)
<i>ASS1</i>	603470	Citrullinemia, 215700 (3), Autosomal recessive
<i>ASXL1</i>	612990	Bohring-Opitz syndrome, 605039 (3), Autosomal dominant; Myelodysplastic syndrome, somatic, 614286 (3)
<i>ASXL2</i>	612991	Shashi-Pena syndrome, 617190 (3), Autosomal dominant
<i>ASXL3</i>	615115	Bainbridge-Ropers syndrome, 615485 (3), Autosomal dominant
<i>ATAD1</i>	614452	Hyperekplexia 4, 618011 (3), Autosomal recessive
<i>ATAD3A</i>	612316	Harel-Yoon syndrome, 617183 (3), Autosomal recessive, Autosomal dominant; Pontocerebellar hypoplasia, hypotonia, and respiratory insufficiency syndrome, neonatal lethal, 618810 (3), Autosomal recessive
<i>ATCAY</i>	608179	Ataxia, cerebellar, Cayman type, 601238 (3), Autosomal recessive
<i>ATF6</i>	605537	Achromatopsia 7, 616517 (3), Autosomal recessive
<i>ATG16L1</i>	610767	{Inflammatory bowel disease (Crohn disease) 10}, 611081 (3)
<i>ATG5</i>	604261	?Spinocerebellar ataxia, autosomal recessive 25, 617584 (3), Autosomal recessive
<i>ATIC</i>	601731	AICA-ribosiduria due to ATIC deficiency, 608688 (3), Autosomal recessive
<i>ATL1</i>	606439	Spastic paraplegia 3A, autosomal dominant, 182600 (3), Autosomal dominant; Neuropathy, hereditary sensory, type ID, 613708 (3), Autosomal dominant

<i>ATL3</i>	609369	Neuropathy, hereditary sensory, type IF, 615632 (3), Autosomal dominant
<i>ATM</i>	607585	Lymphoma, mantle cell, somatic (3); Lymphoma, B-cell non-Hodgkin, somatic (3); Ataxia-telangiectasia, 208900 (3), Autosomal recessive; {Breast cancer, susceptibility to}, 114480 (3), Autosomal dominant, Somatic mutation; T-cell prolymphocytic leukemia, somatic (3)
<i>ATN1</i>	607462	Dentatorubral-pallidoluysian atrophy, 125370 (3), Autosomal dominant; Congenital hypotonia, epilepsy, developmental delay, and digital anomalies, 618494 (3), Autosomal dominant
<i>ATOH7</i>	609875	Persistent hyperplastic primary vitreous, autosomal recessive, 221900 (3), Autosomal recessive
<i>ATP11C</i>	300516	?Hemolytic anemia, congenital, X-linked, 301015 (3), X-linked recessive
<i>ATP13A2</i>	610513	Kufor-Rakeb syndrome, 606693 (3), Autosomal recessive; Spastic paraplegia 78, autosomal recessive, 617225 (3), Autosomal recessive
<i>ATP1A1</i>	182310	Charcot-Marie-Tooth disease, axonal, type 2DD, 618036 (3), Autosomal dominant; Hypomagnesemia, seizures, and mental retardation 2, 618314 (3), Autosomal dominant
<i>ATP1A2</i>	182340	Migraine, familial hemiplegic, 2, 602481 (3), Autosomal dominant; Migraine, familial basilar, 602481 (3), Autosomal dominant; Alternating hemiplegia of childhood 1, 104290 (3), Autosomal dominant
<i>ATP1A3</i>	182350	CAPOS syndrome, 601338 (3), Autosomal dominant; Alternating hemiplegia of childhood 2, 614820 (3), Autosomal dominant; Dystonia-12, 128235 (3), Autosomal dominant
<i>ATP1B1</i>	182330	[Blood pressure regulation QTL], 145500 (2), Multifactorial
<i>ATP2A1</i>	108730	Brody myopathy, 601003 (3), Autosomal recessive
<i>ATP2A2</i>	108740	Acrokeratosis verruciformis, 101900 (3), Autosomal dominant; Darier disease, 124200 (3), Autosomal dominant
<i>ATP2B2</i>	108733	{Deafness, autosomal recessive 12, modifier of}, 601386 (3), Autosomal recessive
<i>ATP2B3</i>	300014	?Spinocerebellar ataxia, X-linked 1, 302500 (3), X-linked recessive
<i>ATP2C1</i>	604384	Hailey-Hailey disease, 169600 (3), Autosomal dominant
<i>ATP5F1A</i>	164360	?Mitochondrial complex V (ATP synthase) deficiency, nuclear type 4, 615228 (3), Autosomal recessive; ?Combined oxidative phosphorylation deficiency 22, 616045 (3), Autosomal recessive
<i>ATP5F1D</i>	603150	Mitochondrial complex V (ATP synthase) deficiency, 618120 (3), Autosomal recessive
<i>ATP5F1E</i>	606153	?Mitochondrial complex V (ATP synthase) deficiency, nuclear type 3, 614053 (3)
<i>ATP5MD</i>	615204	Mitochondrial complex V (ATP synthase) deficiency, nuclear type 6, 618683 (3), Autosomal recessive
<i>ATP6AP1</i>	300197	Immunodeficiency 47, 300972 (3), X-linked recessive

<i>ATP6AP2</i>	300556	Congenital disorder of glycosylation, type IIr, 301045 (3), X-linked recessive; Mental retardation, X-linked, syndromic, Hedera type, 300423 (3), X-linked recessive; ?Parkinsonism with spasticity, X-linked, 300911 (3), X-linked recessive
<i>ATP6VOA2</i>	611716	Wrinkly skin syndrome, 278250 (3), Autosomal recessive; Cutis laxa, autosomal recessive, type IIA, 219200 (3), Autosomal recessive
<i>ATP6VOA4</i>	605239	Renal tubular acidosis, distal, autosomal recessive, 602722 (3), Autosomal recessive
<i>ATP6V1A</i>	607027	Epileptic encephalopathy, infantile or early childhood, 3, 618012 (3), Autosomal dominant; Cutis laxa, autosomal recessive, type IID, 617403 (3), Autosomal recessive
<i>ATP6V1B1</i>	192132	Renal tubular acidosis with deafness, 267300 (3), Autosomal recessive
<i>ATP6V1B2</i>	606939	Zimmermann-Laband syndrome 2, 616455 (3), Autosomal dominant; Deafness, congenital, with onychodystrophy, autosomal dominant, 124480 (3), Autosomal dominant
<i>ATP6V1E1</i>	108746	Cutis laxa, autosomal recessive, type IIC, 617402 (3), Autosomal recessive
<i>ATP7A</i>	300011	Occipital horn syndrome, 304150 (3), X-linked recessive; Menkes disease, 309400 (3), X-linked recessive; Spinal muscular atrophy, distal, X-linked 3, 300489 (3), X-linked recessive
<i>ATP7B</i>	606882	Wilson disease, 277900 (3), Autosomal recessive
<i>ATP8A2</i>	605870	?Cerebellar ataxia, mental retardation, and dysequilibrium syndrome 4, 615268 (3), Autosomal recessive
<i>ATP8B1</i>	602397	Cholestasis, intrahepatic, of pregnancy, 1, 147480 (3), Autosomal dominant; Cholestasis, progressive familial intrahepatic 1, 211600 (3), Autosomal recessive; Cholestasis, benign recurrent intrahepatic, 243300 (3), Autosomal recessive
<i>ATPAF2</i>	608918	?Mitochondrial complex V (ATP synthase) deficiency, nuclear type 1, 604273 (3), Autosomal recessive
<i>ATR</i>	601215	Seckel syndrome 1, 210600 (3), Autosomal recessive; ?Cutaneous telangiectasia and cancer syndrome, familial, 614564 (3), Autosomal dominant
<i>ATRX</i>	300032	Alpha-thalassemia/mental retardation syndrome, 301040 (3), X-linked dominant; Alpha-thalassemia myelodysplasia syndrome, somatic, 300448 (3); Mental retardation-hypotonic facies syndrome, X-linked, 309580 (3), X-linked recessive
<i>ATXN1</i>	601556	Spinocerebellar ataxia 1, 164400 (3), Autosomal dominant
<i>ATXN10</i>	611150	Spinocerebellar ataxia 10, 603516 (3), Autosomal dominant
<i>ATXN2</i>	601517	{Amyotrophic lateral sclerosis, susceptibility to, 13}, 183090 (3), Autosomal dominant; {Parkinson disease, late-onset, susceptibility to}, 168600 (3), Autosomal dominant, Multifactorial; Spinocerebellar ataxia 2, 183090 (3), Autosomal dominant

<i>ATXN3</i>	607047	Machado-Joseph disease, 109150 (3), Autosomal dominant
<i>ATXN7</i>	607640	Spinocerebellar ataxia 7, 164500 (3), Autosomal dominant
<i>AUH</i>	600529	3-methylglutaconic aciduria, type I, 250950 (3), Autosomal recessive
<i>AURKA</i>	603072	{Colon cancer, susceptibility to}, 114500 (3), Autosomal dominant, Somatic mutation
<i>AURKC</i>	603495	Spermatogenic failure 5, 243060 (3), Autosomal recessive
<i>AUTS2</i>	607270	Mental retardation, autosomal dominant 26, 615834 (3), Autosomal dominant
<i>AVIL</i>	613397	Nephrotic syndrome, type 21, 618594 (3), Autosomal recessive
<i>AVP</i>	192340	Diabetes insipidus, neurohypophyseal, 125700 (3), Autosomal dominant
<i>AVPR2</i>	300538	Nephrogenic syndrome of inappropriate antidiuresis, 300539 (3), X-linked recessive; Diabetes insipidus, nephrogenic, 304800 (3), X-linked recessive
<i>AXIN1</i>	603816	Hepatocellular carcinoma, somatic, 114550 (3); ?Caudal duplication anomaly, 607864 (3)
<i>AXIN2</i>	604025	Colorectal cancer, somatic, 114500 (3); Oligodontia-colorectal cancer syndrome, 608615 (3), Autosomal dominant
<i>B2M</i>	109700	Immunodeficiency 43, 241600 (3), Autosomal recessive; ?Amyloidosis, familial visceral, 105200 (3), Autosomal dominant
<i>B3GALNT1</i>	603094	[Blood group, globoside system], 615021 (3); [Blood group, P1PK system, P(k) phenotype], 111400 (3)
<i>B3GALNT2</i>	610194	Muscular dystrophy-dystroglycanopathy (congenital with brain and eye anomalies, type A, 11, 615181 (3), Autosomal recessive
<i>B3GALT6</i>	615291	Al-Gazali syndrome, 609465 (3); 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
<i>B3GAT3</i>	606374	Multiple joint dislocations, short stature, craniofacial dysmorphism, with or without congenital heart defects, 245600 (3), Autosomal recessive
<i>B3GLCT</i>	610308	Peters-plus syndrome, 261540 (3), Autosomal recessive
<i>B4GALNT1</i>	601873	Spastic paraplegia 26, autosomal recessive, 609195 (3), Autosomal recessive
<i>B4GALT1</i>	137060	Congenital disorder of glycosylation, type IIId, 607091 (3), Autosomal recessive
<i>B4GALT7</i>	604327	Ehlers-Danlos syndrome, spondylodysplastic type, 1, 130070 (3), Autosomal recessive
<i>B4GAT1</i>	605517	Muscular dystrophy-dystroglycanopathy (congenital with brain and eye anomalies), type A, 13, 615287 (3), Autosomal recessive
<i>B9D1</i>	614144	?Meckel syndrome 9, 614209 (3), Autosomal recessive; Joubert syndrome 27, 617120 (3), Autosomal recessive
<i>B9D2</i>	611951	Joubert syndrome 34, 614175 (3), Autosomal recessive; ?Meckel syndrome 10, 614175 (3), Autosomal recessive

<i>BAAT</i>	602938	Hypercholanemia, familial, 607748 (3), Autosomal recessive
<i>BACH2</i>	605394	Immunodeficiency 60, 618394 (3), Autosomal dominant
<i>BAG3</i>	603883	Cardiomyopathy, dilated, 1HH, 613881 (3), Autosomal dominant; Myopathy, myofibrillar, 6, 612954 (3), Autosomal dominant
<i>BANF1</i>	603811	Nestor-Guillermo progeria syndrome, 614008 (3), Autosomal recessive
<i>BAP1</i>	603089	Tumor predisposition syndrome, 614327 (3), Autosomal dominant
<i>BARD1</i>	601593	{Breast cancer, susceptibility to}, 114480 (3), Autosomal dominant, Somatic mutation
<i>BAX</i>	600040	T-cell acute lymphoblastic leukemia, somatic, 613065 (3); Colorectal cancer, somatic, 114500 (3)
<i>BBIP1</i>	613605	?Bardet-Biedl syndrome 18, 615995 (3), Autosomal recessive
<i>BBS1</i>	209901	Bardet-Biedl syndrome 1, 209900 (3), Autosomal recessive, Digenic recessive
<i>BBS10</i>	610148	Bardet-Biedl syndrome 10, 615987 (3), Autosomal recessive
<i>BBS12</i>	610683	Bardet-Biedl syndrome 12, 615989 (3), Autosomal recessive
<i>BBS2</i>	606151	Bardet-Biedl syndrome 2, 615981 (3), Autosomal recessive; Retinitis pigmentosa 74, 616562 (3), Autosomal recessive
<i>BBS4</i>	600374	Bardet-Biedl syndrome 4, 615982 (3), Autosomal recessive
<i>BBS5</i>	603650	Bardet-Biedl syndrome 5, 615983 (3), Autosomal recessive
<i>BBS7</i>	607590	Bardet-Biedl syndrome 7, 615984 (3), Autosomal recessive
<i>BBS9</i>	607968	Bardet-Biedl syndrome 9, 615986 (3), Autosomal recessive
<i>BCAM</i>	612773	[Blood group, Lutheran null], 247420 (3), Autosomal recessive; [Blood group, Lutheran system], 111200 (3); [Blood group, Auberger system], 111200 (3)
<i>BCAP31</i>	300398	Deafness, dystonia, and cerebral hypomyelination, 300475 (3), X-linked recessive
<i>BCAT2</i>	113530	?Hypervalinemia or hyperleucine-isoleucinemia, 618850 (3), Autosomal recessive
<i>BCHE</i>	177400	{Apnea, postanesthetic, susceptibility to, due to BCHE deficiency}, 617936 (3); Butyrylcholinesterase deficiency, 617936 (3)
<i>BCKDHA</i>	608348	Maple syrup urine disease, type Ia, 248600 (3), Autosomal recessive
<i>BCKDHB</i>	248611	Maple syrup urine disease, type Ib, 248600 (3), Autosomal recessive
<i>BCKDK</i>	614901	Branched-chain ketoacid dehydrogenase kinase deficiency, 614923 (3)
<i>BCL10</i>	603517	?Immunodeficiency 37, 616098 (3), Autosomal recessive; {Male germ cell tumor, somatic}, 273300 (3); Lymphoma, MALT, somatic, 137245 (3); {Sezary syndrome, somatic} (3); {Lymphoma, follicular, somatic}, 605027 (3); {Mesothelioma, somatic}, 156240 (3)
<i>BCL11A</i>	606557	Dias-Logan syndrome, 617101 (3), Autosomal dominant

<i>BCL11B</i>	606558	Intellectual developmental disorder with dysmorphic facies, speech delay, and T-cell abnormalities, 618092 (3), Autosomal dominant; Immunodeficiency 49, 617237 (3), Autosomal dominant
<i>BCL2</i>	151430	Leukemia/lymphoma, B-cell, 2 (3)
<i>BCL3</i>	109560	Leukemia/lymphoma, B-cell, 3, 109560 (2)
<i>BCL6</i>	109565	Lymphoma, B-cell, 109565 (2)
<i>BCL7A</i>	601406	B-cell non-Hodgkin lymphoma, high-grade (3)
<i>BCO1</i>	605748	?Hypercarotenemia and vitamin A deficiency, autosomal dominant, 115300 (3), Autosomal dominant
<i>BCOR</i>	300485	Microphthalmia, syndromic 2, 300166 (3), X-linked dominant
<i>BCORL1</i>	300688	Shukla-Vernon syndrome, 301029 (3), X-linked recessive
<i>BCR</i>	151410	Leukemia, chronic myeloid, Philadelphia chromosome positive, somatic, 608232 (4); Leukemia, acute lymphocytic, Philadelphia chromosome positive, somatic, 613065 (4)
<i>BCS1L</i>	603647	Leigh syndrome, 256000 (3), Autosomal recessive, Mitochondrial; GRACILE syndrome, 603358 (3), Autosomal recessive; Bjornstad syndrome, 262000 (3), Autosomal recessive; Mitochondrial complex III deficiency, nuclear type 1, 124000 (3), Autosomal recessive
<i>BDP1</i>	607012	?Deafness, autosomal recessive 112, 618257 (3), Autosomal recessive
<i>BEAN1</i>	612051	Spinocerebellar ataxia 31, 117210 (3), Autosomal dominant
<i>BEST1</i>	607854	Retinitis pigmentosa-50, 613194 (3); Bestrophinopathy, autosomal recessive, 611809 (3); Retinitis pigmentosa, concentric, 613194 (3); Vitreoretinchoroidopathy, 193220 (3), Autosomal dominant; Microcornea, rod-cone dystrophy, cataract, and posterior staphyloma, 193220 (3), Autosomal dominant; Macular dystrophy, vitelliform, 2, 153700 (3), Autosomal dominant
<i>BFSP1</i>	603307	Cataract 33, multiple types, 611391 (3), Autosomal recessive, Autosomal dominant
<i>BFSP2</i>	603212	Cataract 12, multiple types, 611597 (3), Autosomal dominant
<i>BGN</i>	301870	Meester-Loeys syndrome, 300989 (3), X-linked; Spondyloepimetaphyseal dysplasia, X-linked, 300106 (3), X-linked recessive
<i>BHLHA9</i>	615416	Syndactyly, mesoaxial synostotic, with phalangeal reduction, 609432 (3), Autosomal recessive; ?Camptosynpolydactyly, complex, 607539 (3), Autosomal recessive
<i>BHLHE41</i>	606200	[Short sleep, familial natural, 1], 612975 (3), Autosomal dominant
<i>BICC1</i>	614295	{Renal dysplasia, cystic, susceptibility to}, 601331 (3), Autosomal dominant

<i>BICD2</i>	609797	Spinal muscular atrophy, lower extremity-predominant, 2B, autosomal dominant, 618291 (3), Autosomal dominant; Spinal muscular atrophy, lower extremity-predominant, 2A, autosomal dominant, 615290 (3), Autosomal dominant
<i>BIN1</i>	601248	Centronuclear myopathy 2, 255200 (3), Autosomal recessive
<i>BLK</i>	191305	Maturity-onset diabetes of the young, type 11, 613375 (3), Autosomal dominant
<i>BLM</i>	604610	Bloom syndrome, 210900 (3), Autosomal recessive
<i>BLNK</i>	604515	?Agammaglobulinemia 4, 613502 (3), Autosomal recessive
<i>BLOC1S3</i>	609762	Hermansky-Pudlak syndrome 8, 614077 (3), Autosomal recessive
<i>BLOC1S6</i>	604310	?Hermansky-pudlak syndrome 9, 614171 (3), Autosomal recessive
<i>BLVRA</i>	109750	Hyperbiliverdinemia, 614156 (3), Autosomal recessive, Autosomal dominant
<i>BMP1</i>	112264	Osteogenesis imperfecta, type XIII, 614856 (3), Autosomal recessive
<i>BMP15</i>	300247	Ovarian dysgenesis 2, 300510 (3), X-linked; Premature ovarian failure 4, 300510 (3), X-linked
<i>BMP2</i>	112261	Brachydactyly, type A2, 112600 (3), Autosomal dominant; Short stature, facial dysmorphism, and skeletal anomalies with or without cardiac anomalies, 617877 (3), Autosomal dominant; {HFE hemochromatosis, modifier of}, 235200 (3), Autosomal recessive
<i>BMP4</i>	112262	Microphthalmia, syndromic 6, 607932 (3), Autosomal dominant; Orofacial cleft 11, 600625 (3)
<i>BMPER</i>	608699	Diaphanospondylodysostosis, 608022 (3), Autosomal recessive
<i>BMPR1A</i>	601299	Polyposis syndrome, hereditary mixed, 2, 610069 (3); Polyposis, juvenile intestinal, 174900 (3), Autosomal dominant; Juvenile polyposis syndrome, infantile form, 174900 (3), Autosomal dominant
<i>BMPR1B</i>	603248	Brachydactyly, type A2, 112600 (3), Autosomal dominant; Brachydactyly, type A1, D, 616849 (3), Autosomal dominant; Acromesomelic dysplasia, Demirhan type, 609441 (3), Autosomal recessive
<i>BMPR2</i>	600799	Pulmonary venoocclusive disease 1, 265450 (3), Autosomal dominant; Pulmonary hypertension, familial primary, 1, with or without HHT, 178600 (3), Autosomal dominant; Pulmonary hypertension, primary, fenfluramine or dexfenfluramine-associated, 178600 (3), Autosomal dominant
<i>BMS1</i>	611448	?Aplasia cutis congenita, nonsyndromic, 107600 (3), Autosomal dominant
<i>BNC1</i>	601930	?Premature ovarian failure 16, 618723 (3), Autosomal dominant
<i>BNC2</i>	608669	Lower urinary tract obstruction, congenital, 618612 (3), Autosomal dominant
<i>BOLA3</i>	613183	Multiple mitochondrial dysfunctions syndrome 2 with hyperglycinemia, 614299 (3), Autosomal recessive

<i>BPGM</i>	613896	Erythrocytosis, familial, 8, 222800 (3), Autosomal recessive
<i>BPNT2</i> (<i>IMPAD1</i>)	614010	Chondrodysplasia with joint dislocations, GPAPP type, 614078 (3), Autosomal recessive
<i>BPTF</i>	601819	Neurodevelopmental disorder with dysmorphic facies and distal limb anomalies, 617755 (3), Autosomal dominant
<i>BRAF</i>	164757	Nonsmall cell lung cancer, somatic (3); Melanoma, malignant, somatic (3); Noonan syndrome 7, 613706 (3), Autosomal dominant; Cardiofaciocutaneous syndrome, 115150 (3), Autosomal dominant; Colorectal cancer, somatic (3); Adenocarcinoma of lung, somatic, 211980 (3); LEOPARD syndrome 3, 613707 (3), Autosomal dominant
<i>BRAT1</i>	614506	Rigidity and multifocal seizure syndrome, lethal neonatal, 614498 (3), Autosomal recessive; Neurodevelopmental disorder with cerebellar atrophy and with or without seizures, 618056 (3), Autosomal recessive
<i>BRCA1</i>	113705	Fanconi anemia, complementation group S, 617883 (3), Autosomal recessive; {Pancreatic cancer, susceptibility to, 4}, 614320 (3); {Breast-ovarian cancer, familial, 1}, 604370 (3), Autosomal dominant, Multifactorial
<i>BRCA2</i>	600185	{Pancreatic cancer 2}, 613347 (3); {Breast cancer, male, susceptibility to}, 114480 (3), Autosomal dominant, Somatic mutation; {Glioblastoma 3}, 613029 (3), Autosomal recessive; Wilms tumor, 194070 (3), Autosomal dominant, Somatic mutation; Fanconi anemia, complementation group D1, 605724 (3), Autosomal recessive; {Medulloblastoma}, 155255 (3), Autosomal recessive, Autosomal dominant, Somatic mutation; {Prostate cancer}, 176807 (3), Autosomal dominant, Somatic mutation; {Breast-ovarian cancer, familial, 2}, 612555 (3), Autosomal dominant
<i>BRDT</i>	602144	?Spermatogenic failure 21, 617644 (3), Autosomal recessive
<i>BRF1</i>	604902	Cerebellofaciodental syndrome, 616202 (3), Autosomal recessive
<i>BRIP1</i>	605882	Fanconi anemia, complementation group J, 609054 (3); {Breast cancer, early-onset, susceptibility to}, 114480 (3), Autosomal dominant, Somatic mutation
<i>BRPF1</i>	602410	Intellectual developmental disorder with dysmorphic facies and ptosis, 617333 (3), Autosomal dominant
<i>BRWD3</i>	300553	Mental retardation, X-linked 93, 300659 (3), X-linked recessive
<i>BSCL2</i>	606158	Lipodystrophy, congenital generalized, type 2, 269700 (3), Autosomal recessive; Silver spastic paraplegia syndrome, 270685 (3), Autosomal dominant; Neuropathy, distal hereditary motor, type VA, 600794 (3), Autosomal dominant; Encephalopathy, progressive, with or without lipodystrophy, 615924 (3), Autosomal recessive
<i>BSG</i>	109480	[Blood group, OK], 111380 (3)
<i>BSND</i>	606412	Sensorineural deafness with mild renal dysfunction, 602522 (3), Autosomal recessive; Bartter syndrome, type 4a, 602522 (3), Autosomal recessive

<i>BTB</i>	609019	Biotinidase deficiency, 253260 (3), Autosomal recessive
<i>BTK</i>	300300	Isolated growth hormone deficiency, type III, with agammaglobulinemia, 307200 (3), X-linked recessive; Agammaglobulinemia, X-linked 1, 300755 (3), X-linked recessive
<i>BTNL2</i>	606000	{Sarcoidosis, susceptibility to, 2}, 612387 (3), Autosomal dominant
<i>BUB1</i>	602452	Colorectal cancer with chromosomal instability, somatic, 114500 (3)
<i>BUB1B</i>	602860	Colorectal cancer, somatic, 114500 (3); [Premature chromatid separation trait], 176430 (3), Autosomal dominant; Mosaic variegated aneuploidy syndrome 1, 257300 (3), Autosomal recessive
<i>BVES</i>	604577	Muscular dystrophy, limb-girdle, autosomal recessive 25, 616812 (3), Autosomal recessive
<i>C11orf80</i>	616109	Hydatidiform mole, recurrent, 4, 618432 (3), Autosomal recessive
<i>C12orf4</i>	616082	Mental retardation, autosomal recessive 66, 618221 (3), Autosomal recessive
<i>C12orf57</i>	615140	Temtamy syndrome, 218340 (3), Autosomal recessive
<i>C12orf65</i>	613541	Spastic paraplegia 55, autosomal recessive, 615035 (3), Autosomal recessive; Combined oxidative phosphorylation deficiency 7, 613559 (3), Autosomal recessive
<i>C19orf12</i>	614297	Neurodegeneration with brain iron accumulation 4, 614298 (3), Autosomal recessive, Autosomal dominant; ?Spastic paraplegia 43, autosomal recessive, 615043 (3), Autosomal recessive
<i>C1GALT1C1</i>	300611	Tn polyagglutination syndrome, somatic, 300622 (3)
<i>C1QA</i>	120550	C1q deficiency, 613652 (3), Autosomal recessive
<i>C1QB</i>	120570	C1q deficiency, 613652 (3), Autosomal recessive
<i>C1QBP</i>	601269	Combined oxidative phosphorylation deficiency 33, 617713 (3), Autosomal recessive
<i>C1QC</i>	120575	C1q deficiency, 613652 (3), Autosomal recessive
<i>C1QTNF5</i>	608752	Retinal degeneration, late-onset, autosomal dominant, 605670 (3), Autosomal dominant
<i>C1R</i>	613785	Ehlers-Danlos syndrome, periodontal type, 1, 130080 (3), Autosomal dominant
<i>C1S</i>	120580	Ehlers-Danlos syndrome, periodontal type, 2, 617174 (3), Autosomal dominant; C1s deficiency, 613783 (3)
<i>C2</i>	613927	C2 deficiency, 217000 (3), Autosomal recessive; {Macular degeneration, age-related, 14, reduced risk of}, 615489 (3)
<i>C2CD3</i>	615944	Orofaciodigital syndrome XIV, 615948 (3), Autosomal recessive
<i>C3</i>	120700	{Hemolytic uremic syndrome, atypical, susceptibility to, 5}, 612925 (3), Autosomal dominant; C3 deficiency, 613779 (3), Autosomal recessive; {Macular degeneration, age-related, 9}, 611378 (3)

<i>C4A</i>	120810	C4a deficiency, 614380 (3), Autosomal recessive; [Blood group, Rodgers], 614374 (3)
<i>C4B</i>	120820	C4B deficiency, 614379 (3)
<i>C5</i>	120900	[Eculizumab, poor response to], 615749 (3), Autosomal dominant; C5 deficiency, 609536 (3)
<i>C6</i>	217050	C6 deficiency, 612446 (3); Combined C6/C7 deficiency (3)
<i>C7</i>	217070	C7 deficiency, 610102 (3)
<i>C8A</i>	120950	C8 deficiency, type I, 613790 (3), Autosomal recessive
<i>C8B</i>	120960	C8 deficiency, type II, 613789 (3), Autosomal recessive
<i>C8orf37</i>	614477	Retinitis pigmentosa 64, 614500 (3), Autosomal recessive; Bardet-Biedl syndrome 21, 617406 (3), Autosomal recessive; Cone-rod dystrophy 16, 614500 (3), Autosomal recessive
<i>C9</i>	120940	{Macular degeneration, age-related, 15, susceptibility to}, 615591 (3); C9 deficiency, 613825 (3)
<i>C9orf72</i>	614260	Frontotemporal dementia and/or amyotrophic lateral sclerosis 1, 105550 (3), Autosomal dominant
<i>CA12</i>	603263	Hyperchlorhidrosis, isolated, 143860 (3), Autosomal recessive
<i>CA2</i>	611492	Osteopetrosis, autosomal recessive 3, with renal tubular acidosis, 259730 (3), Autosomal recessive
<i>CA4</i>	114760	Retinitis pigmentosa 17, 600852 (3), Autosomal dominant
<i>CA5A</i>	114761	Hyperammonemia due to carbonic anhydrase VA deficiency, 615751 (3), Autosomal recessive
<i>CA8</i>	114815	Cerebellar ataxia and mental retardation with or without quadrupedal locomotion 3, 613227 (3), Autosomal recessive
<i>CABP2</i>	607314	Deafness, autosomal recessive 93, 614899 (3), Autosomal recessive
<i>CABP4</i>	608965	Cone-rod synaptic disorder, congenital nonprogressive, 610427 (3), Autosomal recessive
<i>CACNA1A</i>	601011	Spinocerebellar ataxia 6, 183086 (3), Autosomal dominant; Epileptic encephalopathy, early infantile, 42, 617106 (3), Autosomal dominant; Migraine, familial hemiplegic, 1, with progressive cerebellar ataxia, 141500 (3), Autosomal dominant; Episodic ataxia, type 2, 108500 (3), Autosomal dominant; Migraine, familial hemiplegic, 1, 141500 (3), Autosomal dominant
<i>CACNA1B</i>	601012	Neurodevelopmental disorder with seizures and nonepileptic hyperkinetic movements, 618497 (3), Autosomal recessive
<i>CACNA1C</i>	114205	Timothy syndrome, 601005 (3), Autosomal dominant; Long QT syndrome 8, 618447 (3); Brugada syndrome 3, 611875 (3)
<i>CACNA1D</i>	114206	Primary aldosteronism, seizures, and neurologic abnormalities, 615474 (3), Autosomal dominant; Sinoatrial node dysfunction and deafness, 614896 (3), Autosomal recessive
<i>CACNA1E</i>	601013	Epileptic encephalopathy, early infantile, 69, 618285 (3), Autosomal dominant

<i>CACNA1F</i>	300110	Cone-rod dystrophy, X-linked, 3, 300476 (3), X-linked recessive; Night blindness, congenital stationary (incomplete), 2A, X-linked, 300071 (3), X-linked; Aland Island eye disease, 300600 (3), X-linked
<i>CACNA1G</i>	604065	Spinocerebellar ataxia 42, 616795 (3), Autosomal dominant; Spinocerebellar ataxia 42, early-onset, severe, with neurodevelopmental deficits, 618087 (3), Autosomal dominant
<i>CACNA1H</i>	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
<i>CACNA1S</i>	114208	{Malignant hyperthermia susceptibility 5}, 601887 (3), Autosomal dominant; {Thyrotoxic periodic paralysis, susceptibility to, 1}, 188580 (3), Autosomal dominant; {Malignant hyperthermia, susceptibility to, 5}, 601887 (3), Autosomal dominant; Hypokalemic periodic paralysis, type 1, 170400 (3), Autosomal dominant
<i>CACNA2D1</i>	114204/154276	-/{Malignant hyperthermia susceptibility 3}, 154276 (2), Autosomal dominant
<i>CACNA2D2</i>	607082	Cerebellar atrophy with seizures and variable developmental delay, 618501 (3), Autosomal recessive
<i>CACNA2D4</i>	608171	Retinal cone dystrophy 4, 610478 (3), Autosomal recessive
<i>CACNB2</i>	600003	Brugada syndrome 4, 611876 (3)
<i>CACNB4</i>	601949	{Epilepsy, idiopathic generalized, susceptibility to, 9}, 607682 (3), Autosomal dominant; {Epilepsy, juvenile myoclonic, susceptibility to, 6}, 607682 (3), Autosomal dominant; Episodic ataxia, type 5, 613855 (3), Autosomal dominant
<i>CACNG2</i>	602911	?Mental retardation, autosomal dominant 10, 614256 (3), Autosomal dominant
<i>CAD</i>	114010	Epileptic encephalopathy, early infantile, 50, 616457 (3), Autosomal recessive
<i>CALCR</i>	114131	{Osteoporosis, postmenopausal, susceptibility}, 166710 (3), Autosomal dominant
<i>CALCRL</i>	114190	?Lymphatic malformation 8, 618773 (3), Autosomal recessive
<i>CALM1</i>	114180	Long QT syndrome 14, 616247 (3), Autosomal dominant; Ventricular tachycardia, catecholaminergic polymorphic, 4, 614916 (3), Autosomal dominant
<i>CALM2</i>	114182	Long QT syndrome 15, 616249 (3), Autosomal dominant
<i>CALM3</i>	114183	Long QT syndrome 16, 618782 (3), Autosomal dominant; ?Ventricular tachycardia, catecholaminergic polymorphic 6, 618782 (3), Autosomal dominant
<i>CALR</i>	109091	Myelofibrosis, somatic, 254450 (3); Thrombocythemia, somatic, 187950 (3)
<i>CAMK2A</i>	114078	?Mental retardation, autosomal recessive 63, 618095 (3), Autosomal recessive; Mental retardation, autosomal dominant 53, 617798 (3), Autosomal dominant

<i>CAMK2B</i>	607707	Mental retardation, autosomal dominant 54, 617799 (3), Autosomal dominant
<i>CAMK2G</i>	602123	Mental retardation, autosomal dominant 59, 618522 (3), Autosomal dominant
<i>CAMTA1</i>	611501	Cerebellar ataxia, nonprogressive, with mental retardation, 614756 (3), Autosomal dominant
<i>CANT1</i>	613165	Desbuquois dysplasia 1, 251450 (3), Autosomal recessive; Epiphyseal dysplasia, multiple, 7, 617719 (3), Autosomal recessive
<i>CAPN1</i>	114220	Spastic paraplegia 76, autosomal recessive, 616907 (3), Autosomal recessive
<i>CAPN10</i>	605286	{Diabetes mellitus, noninsulin-dependent 1}, 601283 (3)
<i>CAPN3</i>	114240	Muscular dystrophy, limb-girdle, autosomal recessive 1, 253600 (3), Autosomal recessive; Muscular dystrophy, limb-girdle, autosomal dominant 4, 618129 (3), Autosomal dominant
<i>CAPN5</i>	602537	Vitreoretinopathy, neovascular inflammatory, 193235 (3), Autosomal dominant
<i>CARD11</i>	607210	Immunodeficiency 11B with atopic dermatitis, 617638 (3), Autosomal dominant; B-cell expansion with NFKB and T-cell anergy, 616452 (3), Autosomal dominant; Immunodeficiency 11A, 615206 (3), Autosomal recessive
<i>CARD14</i>	607211	Pityriasis rubra pilaris, 173200 (3), Autosomal dominant; Psoriasis 2, 602723 (3), Autosomal dominant
<i>CARD9</i>	607212	Candidiasis, familial, 2, autosomal recessive, 212050 (3), Autosomal recessive
<i>CARMIL2</i>	610859	Immunodeficiency 58, 618131 (3), Autosomal recessive
<i>CARS1 (CARS)</i>	123859	Microcephaly, developmental delay, and brittle hair syndrome, 618891 (3), Autosomal recessive
<i>CARS2</i>	612800	Combined oxidative phosphorylation deficiency 27, 616672 (3), Autosomal recessive
<i>CARTPT</i>	602606	{?Obesity, susceptibility to}, 601665 (3), Autosomal recessive, Autosomal dominant, Multifactorial
<i>CASK</i>	300172	Mental retardation, with or without nystagmus, 300422 (3); Mental retardation and microcephaly with pontine and cerebellar hypoplasia, 300749 (3), X-linked dominant; FG syndrome 4, 300422 (3)
<i>CASP10</i>	601762	Lymphoma, non-Hodgkin, somatic, 605027 (3); Gastric cancer, somatic, 613659 (3); Autoimmune lymphoproliferative syndrome, type II, 603909 (3), Autosomal dominant
<i>CASP12</i>	608633	{Sepsis, susceptibility to} (3)
<i>CASP14</i>	605848	Ichthyosis, congenital, autosomal recessive 12, 617320 (3), Autosomal recessive

<i>CASP8</i>	601763	{Lung cancer, protection against}, 211980 (3), Autosomal dominant, Somatic mutation; ?Autoimmune lymphoproliferative syndrome, type IIB, 607271 (3), Autosomal recessive; Hepatocellular carcinoma, somatic, 114550 (3); {Breast cancer, protection against}, 114480 (3), Autosomal dominant, Somatic mutation
<i>CASQ1</i>	114250	Myopathy, vacuolar, with CASQ1 aggregates, 616231 (3), Autosomal dominant
<i>CASQ2</i>	114251	Ventricular tachycardia, catecholaminergic polymorphic, 2, 611938 (3), Autosomal recessive
<i>CASR</i>	601199	Hypocalcemia, autosomal dominant, with Bartter syndrome, 601198 (3), Autosomal dominant; Hypocalciuric hypercalcemia, type I, 145980 (3), Autosomal dominant; {Epilepsy idiopathic generalized, susceptibility to, 8}, 612899 (3); Hypocalcemia, autosomal dominant, 601198 (3), Autosomal dominant; Hyperparathyroidism, neonatal, 239200 (3), Autosomal recessive, Autosomal dominant
<i>CAST</i>	114090	Peeling skin with leukonychia, acral punctate keratoses, cheilitis, and knuckle pads, 616295 (3), Autosomal recessive
<i>CAT</i>	115500	Acatalasemia, 614097 (3)
<i>CATSPER1</i>	606389	Spermatogenic failure 7, 612997 (3), Autosomal recessive
<i>CAV1</i>	601047	Lipodystrophy, familial partial, type 7, 606721 (3), Autosomal dominant; ?Lipodystrophy, congenital generalized, type 3, 612526 (3), Autosomal recessive; Pulmonary hypertension, primary, 3, 615343 (3), Autosomal dominant
<i>CAV3</i>	601253	Creatine phosphokinase, elevated serum, 123320 (3), Autosomal dominant; Long QT syndrome 9, 611818 (3), Autosomal dominant; Myopathy, distal, Tateyama type, 614321 (3), Autosomal dominant; Rippling muscle disease 2, 606072 (3), Autosomal dominant; Cardiomyopathy, familial hypertrophic, 192600 (3), Digenic dominant, Autosomal dominant
<i>CAVIN1</i>	603198	Lipodystrophy, congenital generalized, type 4, 613327 (3), Autosomal recessive
<i>CBFB</i>	121360	Myeloid leukemia, acute, M4/M4Eo subtype, somatic, 601626 (1)
<i>CBL</i>	165360	Noonan syndrome-like disorder with or without juvenile myelomonocytic leukemia, 613563 (3), Autosomal dominant; ?Juvenile myelomonocytic leukemia, 607785 (3), Autosomal dominant, Somatic mutation
<i>CBLIF (GIF)</i>	609342	Intrinsic factor deficiency, 261000 (3), Autosomal recessive
<i>CBS</i>	613381	Homocystinuria, B6-responsive and nonresponsive types, 236200 (3), Autosomal recessive; Thrombosis, hyperhomocysteinemic, 236200 (3), Autosomal recessive
<i>CBX2</i>	602770	?46XY sex reversal 5, 613080 (3), Autosomal recessive
<i>CC2D1A</i>	610055	Mental retardation, autosomal recessive 3, 608443 (3), Autosomal recessive

<i>CC2D2A</i>	612013	Meckel syndrome 6, 612284 (3), Autosomal recessive; Joubert syndrome 9, 612285 (3), Autosomal recessive; COACH syndrome, 216360 (3), Autosomal recessive
<i>CCBE1</i>	612753	Hennekam lymphangiectasia-lymphedema syndrome 1, 235510 (3), Autosomal recessive
<i>CCDC103</i>	614677	Ciliary dyskinesia, primary, 17, 614679 (3), Autosomal recessive
<i>CCDC114</i>	615038	Ciliary dyskinesia, primary, 20, 615067 (3), Autosomal recessive
<i>CCDC115</i>	613734	Congenital disorder of glycosylation, type Ilo, 616828 (3), Autosomal recessive
<i>CCDC151</i>	615956	Ciliary dyskinesia, primary, 30, 616037 (3), Autosomal recessive
<i>CCDC174</i>	616735	Hypotonia, infantile, with psychomotor retardation, 616816 (3), Autosomal recessive
<i>CCDC22</i>	300859	Ritscher-Schinzel syndrome 2, 300963 (3), X-linked recessive
<i>CCDC28B</i>	610162	{Bardet-Biedl syndrome 1, modifier of}, 209900 (3), Autosomal recessive, Digenic recessive
<i>CCDC39</i>	613798	Ciliary dyskinesia, primary, 14, 613807 (3)
<i>CCDC40</i>	613799	Ciliary dyskinesia, primary, 15, 613808 (3)
<i>CCDC47</i>	618260	Trichohepatoneurodevelopmental syndrome, 618268 (3), Autosomal recessive
<i>CCDC50</i>	611051	?Deafness, autosomal dominant 44, 607453 (3), Autosomal dominant
<i>CCDC65</i>	611088	Ciliary dyskinesia, primary, 27, 615504 (3), Autosomal recessive
<i>CCDC78</i>	614666	?Centronuclear myopathy 4, 614807 (3), Autosomal dominant
<i>CCDC8</i>	614145	3-M syndrome 3, 614205 (3), Autosomal recessive
<i>CCDC88A</i>	609736	?PEHO syndrome-like, 617507 (3), Autosomal recessive
<i>CCDC88C</i>	611204	?Spinocerebellar ataxia 40, 616053 (3), Autosomal dominant; Hydrocephalus, congenital, 1, 236600 (3), Autosomal recessive
<i>CCL11</i>	601156	{HIV1, resistance to}, 609423 (3); {Asthma, susceptibility to}, 600807 (3), Autosomal dominant
<i>CCL2</i>	158105	{HIV-1, resistance to}, 609423 (3); {Mycobacterium tuberculosis, susceptibility to}, 607948 (3); {Spina bifida, susceptibility to}, 182940 (3), Autosomal dominant; {Coronary artery disease, modifier of} (3)
<i>CCL3</i>	182283	{HIV infection, resistance to}, 609423 (2)
<i>CCL3L1</i>	601395	{HIV/AIDS, susceptibility to}, 609423 (3)
<i>CCL5</i>	187011	{HIV-1 disease, rapid progression of} (3); {HIV-1 disease, delayed progression of} (3)
<i>CCM2</i>	607929	Cerebral cavernous malformations-2, 603284 (3), Autosomal dominant
<i>CCN6 (WISP3)</i>	603400	Spondyloepiphyseal dysplasia tarda with progressive arthropathy, 208230 (3), Autosomal recessive; Arthropathy, progressive pseudorheumatoid, of childhood, 208230 (3), Autosomal recessive

<i>CCND1</i>	168461	{Colorectal cancer, susceptibility to}, 114500 (3), Autosomal dominant, Somatic mutation; {von Hippel-Lindau syndrome, modifier of}, 193300 (3), Autosomal dominant; {Multiple myeloma, susceptibility to}, 254500 (3), Somatic mutation
<i>CCND2</i>	123833	Megalencephaly-polymicrogyria-polydactyly-hydrocephalus syndrome 3, 615938 (3), Autosomal dominant
<i>CCNK</i>	603544	?Intellectual developmental disorder with hypertelorism and distinctive facies, 618147 (3), Autosomal dominant
<i>CCNO</i>	607752	Ciliary dyskinesia, primary, 29, 615872 (3), Autosomal recessive
<i>CCNQ</i>	300708	STAR syndrome, 300707 (3), X-linked dominant
<i>CCR2</i>	601267	{HIV infection, susceptibility/resistance to} (3)
<i>CCR5</i>	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)
<i>CCT5</i>	610150	Neuropathy, hereditary sensory, with spastic paraplegia, 256840 (3), Autosomal recessive
<i>CD151</i>	602243	Nephropathy with pretibial epidermolysis bullosa and deafness, 609057 (3); [Blood group, Raph], 179620 (3)
<i>CD164</i>	603356	?Deafness, autosomal dominant 66, 616969 (3), Autosomal dominant
<i>CD19</i>	107265	Immunodeficiency, common variable, 3, 613493 (3), Autosomal recessive
<i>CD207</i>	604862	[?Birbeck granule deficiency], 613393 (3)
<i>CD209</i>	604672	{Mycobacterium tuberculosis, susceptibility to}, 607948 (3); {HIV type 1, susceptibility to}, 609423 (3); {Dengue fever, protection against}, 614371 (3)
<i>CD244</i>	605554	{Rheumatoid arthritis, susceptibility to}, 180300 (3)
<i>CD247</i>	186780	?Immunodeficiency 25, 610163 (3), Autosomal recessive
<i>CD27</i>	186711	Lymphoproliferative syndrome 2, 615122 (3), Autosomal recessive
<i>CD2AP</i>	604241	Glomerulosclerosis, focal segmental, 3, 607832 (3)
<i>CD320</i>	606475	Methylmalonic aciduria, transient, due to transcobalamin receptor defect, 613646 (3)
<i>CD36</i>	173510	{Coronary heart disease, susceptibility to, 7}, 610938 (3); {Malaria, cerebral, susceptibility to}, 611162 (3); Platelet glycoprotein IV deficiency, 608404 (3), Autosomal recessive; {Malaria, cerebral, reduced risk of}, 611162 (3)
<i>CD3D</i>	186790	Immunodeficiency 19, 615617 (3), Autosomal recessive
<i>CD3E</i>	186830	Immunodeficiency 18, SCID variant, 615615 (3), Autosomal recessive; Immunodeficiency 18, 615615 (3), Autosomal recessive
<i>CD3G</i>	186740	Immunodeficiency 17, CD3 gamma deficient, 615607 (3), Autosomal recessive
<i>CD4</i>	186940	OKT4 epitope deficiency, 613949 (3)

<i>CD40</i>	109535	Immunodeficiency with hyper-IgM, type 3, 606843 (3), Autosomal recessive
<i>CD40LG</i>	300386	Immunodeficiency, X-linked, with hyper-IgM, 308230 (3), X-linked recessive
<i>CD44</i>	107269	[Blood group, Indian system], 609027 (3)
<i>CD46</i>	120920	{Hemolytic uremic syndrome, atypical, susceptibility to, 2}, 612922 (3), Autosomal recessive, Autosomal dominant
<i>CD55</i>	125240	[Blood group Cromer], 613793 (3), Autosomal recessive; Complement hyperactivation, angiopathic thrombosis, and protein-losing enteropathy, 226300 (3), Autosomal recessive
<i>CD59</i>	107271	Hemolytic anemia, CD59-mediated, with or without immune-mediated polyneuropathy, 612300 (3), Autosomal recessive
<i>CD70</i>	602840	Lymphoproliferative syndrome 3, 618261 (3), Autosomal recessive
<i>CD79A</i>	112205	Agammaglobulinemia 3, 613501 (3), Autosomal recessive
<i>CD79B</i>	147245	Agammaglobulinemia 6, 612692 (3), Autosomal recessive
<i>CD81</i>	186845	Immunodeficiency, common variable, 6, 613496 (3), Autosomal recessive
<i>CD8A</i>	186910	CD8 deficiency, familial, 608957 (3), Autosomal recessive
<i>CD96</i>	606037	C syndrome, 211750 (3), Autosomal dominant
<i>CDAN1</i>	607465	Dyserythropoietic anemia, congenital, type Ia, 224120 (3), Autosomal recessive
<i>CDC14A</i>	603504	Deafness, autosomal recessive 32, with or without immotile sperm, 608653 (3), Autosomal recessive
<i>CDC42</i>	116952	Takenouchi-Kosaki syndrome, 616737 (3), Autosomal dominant
<i>CDC45</i>	603465	Meier-Gorlin syndrome 7, 617063 (3), Autosomal recessive
<i>CDC6</i>	602627	?Meier-Gorlin syndrome 5, 613805 (3), Autosomal recessive
<i>CDC73</i>	607393	Parathyroid adenoma with cystic changes, 145001 (3), Autosomal dominant; Hyperparathyroidism-jaw tumor syndrome, 145001 (3), Autosomal dominant; Parathyroid carcinoma, 608266 (3); Hyperparathyroidism, familial primary, 145000 (3), Autosomal dominant
<i>CDCA7</i>	609937	Immunodeficiency-centromeric instability-facial anomalies syndrome 3, 616910 (3), Autosomal recessive
<i>CDH1</i>	192090	Endometrial carcinoma, somatic, 608089 (3); {Prostate cancer, susceptibility to}, 176807 (3), Autosomal dominant, Somatic mutation; Blepharocheilodontic syndrome 1, 119580 (3), Autosomal dominant; Gastric cancer, hereditary diffuse, with or without cleft lip and/or palate, 137215 (3), Autosomal dominant; {Breast cancer, lobular}, 114480 (3), Autosomal dominant, Somatic mutation; Ovarian cancer, somatic, 167000 (3)
<i>CDH11</i>	600023	Elsahy-Waters syndrome, 211380 (3), Autosomal recessive
<i>CDH15</i>	114019	Mental retardation, autosomal dominant 3, 612580 (3)
<i>CDH2</i>	114020	Arrhythmogenic right ventricular dysplasia, familial, 14, 618920 (3), Autosomal dominant; Agenesis of corpus callosum, cardiac, ocular, and genital syndrome, 618929 (3), Autosomal dominant

<i>CDH23</i>	605516	{Pituitary adenoma 5, multiple types}, 617540 (3), Autosomal dominant; Deafness, autosomal recessive 12, 601386 (3), Autosomal recessive; Usher syndrome, type 1D/F digenic, 601067 (3), Autosomal recessive, Digenic recessive; Usher syndrome, type 1D, 601067 (3), Autosomal recessive, Digenic recessive
<i>CDH3</i>	114021	Ectodermal dysplasia, ectrodactyly, and macular dystrophy, 225280 (3), Autosomal recessive; Hypotrichosis, congenital, with juvenile macular dystrophy, 601553 (3), Autosomal recessive
<i>CDHR1</i>	609502	Cone-rod dystrophy 15, 613660 (3), Autosomal recessive; Retinitis pigmentosa 65, 613660 (3), Autosomal recessive
<i>CDIN1</i> (<i>C15orf41</i>)	615626	Dyserythropoietic anemia, congenital, type 1b, 615631 (3), Autosomal recessive
<i>CDK10</i>	603464	Al Kaissi syndrome, 617694 (3), Autosomal recessive
<i>CDK13</i>	603309	Congenital heart defects, dysmorphic facial features, and intellectual developmental disorder, 617360 (3), Autosomal dominant
<i>CDK19</i>	614720	Epileptic encephalopathy, early infantile, 87, 618916 (3), Autosomal dominant
<i>CDK4</i>	123829	{Melanoma, cutaneous malignant, 3}, 609048 (3), Autosomal dominant
<i>CDK5</i>	123831	?Lissencephaly 7 with cerebellar hypoplasia, 616342 (3), Autosomal recessive
<i>CDK5RAP2</i>	608201	Microcephaly 3, primary, autosomal recessive, 604804 (3), Autosomal recessive
<i>CDK6</i>	603368	?Microcephaly 12, primary, autosomal recessive, 616080 (3), Autosomal recessive
<i>CDK8</i>	603184	Intellectual developmental disorder with hypotonia and behavioral abnormalities, 618748 (3), Autosomal dominant
<i>CDKL5</i>	300203	Epileptic encephalopathy, early infantile, 2, 300672 (3), X-linked dominant
<i>CDKN1B</i>	600778	Multiple endocrine neoplasia, type IV, 610755 (3), Autosomal dominant
<i>CDKN1C</i>	600856	IMAGE syndrome, 614732 (3), Autosomal dominant; Beckwith-Wiedemann syndrome, 130650 (3), Autosomal dominant
<i>CDKN2A</i>	600160	{Melanoma and neural system tumor syndrome}, 155755 (3), Autosomal dominant; {Melanoma, cutaneous malignant, 2}, 155601 (3), Autosomal dominant; {Melanoma-pancreatic cancer syndrome}, 606719 (3), Autosomal dominant
<i>CDON</i>	608707	Holoprosencephaly 11, 614226 (3), Autosomal dominant
<i>CDSN</i>	602593	Hypotrichosis 2, 146520 (3), Autosomal dominant; Peeling skin syndrome 1, 270300 (3), Autosomal recessive
<i>CDT1</i>	605525	Meier-Gorlin syndrome 4, 613804 (3), Autosomal recessive
<i>CEACAM16</i>	614591	Deafness, autosomal recessive 113, 618410 (3), Autosomal recessive; Deafness, autosomal dominant 4B, 614614 (3), Autosomal dominant

<i>CEBPA</i>	116897	Leukemia, acute myeloid, somatic, 601626 (3); ?Leukemia, acute myeloid, 601626 (3), Autosomal dominant, Somatic mutation
<i>CEBPE</i>	600749	Specific granule deficiency, 245480 (3), Autosomal recessive
<i>CEL</i>	114840	Maturity-onset diabetes of the young, type VIII, 609812 (3), Autosomal dominant
<i>CELA2A</i>	609443	Abdominal obesity-metabolic syndrome 4, 618620 (3), Autosomal dominant
<i>CENPE</i>	117143	?Microcephaly 13, primary, autosomal recessive, 616051 (3), Autosomal recessive
<i>CENPF</i>	600236	Stromme syndrome, 243605 (3), Autosomal recessive
<i>CENPJ</i>	609279	Microcephaly 6, primary, autosomal recessive, 608393 (3), Autosomal recessive; ?Seckel syndrome 4, 613676 (3), Autosomal recessive
<i>CENPT</i>	611510	?Short stature and microcephaly with genital anomalies, 618702 (3), Autosomal recessive
<i>CEP104</i>	616690	Joubert syndrome 25, 616781 (3), Autosomal recessive
<i>CEP120</i>	613446	Joubert syndrome 31, 617761 (3), Autosomal recessive; Short-rib thoracic dysplasia 13 with or without polydactyly, 616300 (3), Autosomal recessive
<i>CEP135</i>	611423	Microcephaly 8, primary, autosomal recessive, 614673 (3), Autosomal recessive
<i>CEP152</i>	613529	Microcephaly 9, primary, autosomal recessive, 614852 (3), Autosomal recessive; Seckel syndrome 5, 613823 (3), Autosomal recessive
<i>CEP164</i>	614848	Nephronophthisis 15, 614845 (3), Autosomal recessive
<i>CEP19</i>	615586	Morbid obesity and spermatogenic failure, 615703 (3), Autosomal recessive
<i>CEP250</i>	609689	Cone-rod dystrophy and hearing loss 2, 618358 (3), Autosomal recessive
<i>CEP290</i>	610142	?Bardet-Biedl syndrome 14, 615991 (3), Autosomal recessive; Leber congenital amaurosis 10, 611755 (3); Senior-Loken syndrome 6, 610189 (3), Autosomal recessive; Meckel syndrome 4, 611134 (3), Autosomal recessive; Joubert syndrome 5, 610188 (3), Autosomal recessive
<i>CEP41</i>	610523	Joubert syndrome 15, 614464 (3), Autosomal recessive
<i>CEP43 (FGFR1OP)</i>	605392	Myeloproliferative disorder, 605392 (2)
<i>CEP55</i>	610000	Multinucleated neurons, anhydramnios, renal dysplasia, cerebellar hypoplasia, and hydranencephaly, 236500 (3), Autosomal recessive
<i>CEP57</i>	607951	Mosaic variegated aneuploidy syndrome 2, 614114 (3), Autosomal recessive
<i>CEP63</i>	614724	?Seckel syndrome 6, 614728 (3), Autosomal recessive
<i>CEP78</i>	617110	Cone-rod dystrophy and hearing loss, 617236 (3), Autosomal recessive
<i>CEP83</i>	615847	Nephronophthisis 18, 615862 (3), Autosomal recessive

<i>CEP85L</i>	618865	Lissencephaly 10, 618873 (3), Autosomal dominant
<i>CERKL</i>	608381	Retinitis pigmentosa 26, 608380 (3)
<i>CERS1</i>	606919	?Epilepsy, progressive myoclonic, 8, 616230 (3), Autosomal recessive
<i>CERS3</i>	615276	Ichthyosis, congenital, autosomal recessive 9, 615023 (3), Autosomal recessive
<i>CERT1</i> (<i>COL4A3BP</i>)	604677	Mental retardation, autosomal dominant 34, 616351 (3), Autosomal dominant
<i>CES1</i>	114835	Drug metabolism, altered, CES1-related, 618057 (3)
<i>CETP</i>	118470	[High density lipoprotein cholesterol level QTL 10], 143470 (3), Autosomal dominant; Hyperalphalipoproteinemia, 143470 (3), Autosomal dominant
<i>CFAP251</i> (<i>WDR66</i>)	618146	Spermatogenic failure 33, 618152 (3), Autosomal recessive
<i>CFAP298</i>	615494	Ciliary dyskinesia, primary, 26, 615500 (3), Autosomal recessive
<i>CFAP300</i>	618058	Ciliary dyskinesia, primary, 38, 618063 (3), Autosomal recessive
<i>CFAP410</i>	603191	Spondylometaphyseal dysplasia, axial, 602271 (3), Autosomal recessive; Retinal dystrophy with macular staphyloma, 617547 (3), Autosomal recessive
<i>CFAP43</i>	617558	Hydrocephalus, normal pressure, 1, 236690 (3), Autosomal dominant; Spermatogenic failure 19, 617592 (3), Autosomal recessive
<i>CFAP44</i>	617559	?Spermatogenic failure 20, 617593 (3), Autosomal recessive
<i>CFAP53</i>	614759	Heterotaxy, visceral, 6, autosomal recessive, 614779 (3), Autosomal recessive
<i>CFAP65</i>	614270	Spermatogenic failure 40, 618664 (3), Autosomal recessive
<i>CFAP69</i>	617949	Spermatogenic failure 24, 617959 (3), Autosomal recessive
<i>CFAP70</i>	618661	?Spermatogenic failure 41, 618670 (3), Autosomal recessive
<i>CFB</i>	138470	?Complement factor B deficiency, 615561 (3), Autosomal recessive; {Macular degeneration, age-related, 14, reduced risk of}, 615489 (3); {Hemolytic uremic syndrome, atypical, susceptibility to, 4}, 612924 (3), Autosomal dominant
<i>CFC1</i>	605194	Heterotaxy, visceral, 2, autosomal, 605376 (3), Autosomal dominant
<i>CFD</i>	134350	Complement factor D deficiency, 613912 (3), Autosomal recessive
<i>CFH</i>	134370	Complement factor H deficiency, 609814 (3), Autosomal recessive, Autosomal dominant; {Hemolytic uremic syndrome, atypical, susceptibility to, 1}, 235400 (3), Autosomal recessive, Autosomal dominant; Basal laminar drusen, 126700 (3), Autosomal dominant; {Macular degeneration, age-related, 4}, 610698 (3)
<i>CFHR1</i>	134371	{Hemolytic uremic syndrome, atypical, susceptibility to}, 235400 (3), Autosomal recessive, Autosomal dominant; {Macular degeneration, age-related, reduced risk of}, 603075 (3), Autosomal dominant

<i>CFHR3</i>	605336	{Macular degeneration, age-related, reduced risk of}, 603075 (3), Autosomal dominant; {Hemolytic uremic syndrome, atypical, susceptibility to}, 235400 (3), Autosomal recessive, Autosomal dominant
<i>CFHR5</i>	608593	Nephropathy due to CFHR5 deficiency, 614809 (3), Autosomal dominant
<i>CFI</i>	217030	{Hemolytic uremic syndrome, atypical, susceptibility to, 3}, 612923 (3), Autosomal dominant; Complement factor I deficiency, 610984 (3), Autosomal recessive; {Macular degeneration, age-related, 13, susceptibility to}, 615439 (3), Autosomal dominant
<i>CFL2</i>	601443	Nemaline myopathy 7, autosomal recessive, 610687 (3), Autosomal recessive
<i>CFP</i>	300383	Properdin deficiency, X-linked, 312060 (3), X-linked recessive
<i>CFTR</i>	602421	{Bronchiectasis with or without elevated sweat chloride 1, modifier of}, 211400 (3), Autosomal dominant; Sweat chloride elevation without CF (3); {Hypertrypsinemia, neonatal} (3); Cystic fibrosis, 219700 (3), Autosomal recessive; Congenital bilateral absence of vas deferens, 277180 (3), Autosomal recessive; {Pancreatitis, hereditary}, 167800 (3), Autosomal dominant
<i>CHAMP1</i>	616327	Mental retardation, autosomal dominant 40, 616579 (3), Autosomal dominant
<i>CHAT</i>	118490	Myasthenic syndrome, congenital, 6, presynaptic, 254210 (3), Autosomal recessive
<i>CHCHD10</i>	615903	Spinal muscular atrophy, Jokela type, 615048 (3), Autosomal dominant; Frontotemporal dementia and/or amyotrophic lateral sclerosis 2, 615911 (3), Autosomal dominant; ?Myopathy, isolated mitochondrial, autosomal dominant, 616209 (3), Autosomal dominant
<i>CHCHD2</i>	616244	Parkinson disease 22, autosomal dominant, 616710 (3), Autosomal dominant
<i>CHD1</i>	602118	Pilarowski-Bjornsson syndrome, 617682 (3), Autosomal dominant
<i>CHD2</i>	602119	Epileptic encephalopathy, childhood-onset, 615369 (3), Autosomal dominant
<i>CHD3</i>	602120	Snijders Blok-Campeau syndrome, 618205 (3), Autosomal dominant
<i>CHD4</i>	603277	Sifrim-Hitz-Weiss syndrome, 617159 (3), Autosomal dominant
<i>CHD7</i>	608892	Hypogonadotropic hypogonadism 5 with or without anosmia, 612370 (3), Autosomal dominant; CHARGE syndrome, 214800 (3), Autosomal dominant
<i>CHD8</i>	610528	{Autism, susceptibility to, 18}, 615032 (3), Autosomal dominant

<i>CHEK2</i>	604373	Li-Fraumeni syndrome, 609265 (3); Osteosarcoma, somatic, 259500 (3); {Prostate cancer, familial, susceptibility to}, 176807 (3), Autosomal dominant, Somatic mutation; {Breast cancer, susceptibility to}, 114480 (3), Autosomal dominant, Somatic mutation; {Breast and colorectal cancer, susceptibility to} (3)
<i>CHI3L1</i>	601525	{Schizophrenia, susceptibility to}, 181500 (3), Autosomal dominant; {Asthma-related traits, susceptibility to, 7}, 611960 (3)
<i>CHIC2</i>	604332	{Leukemia, acute myeloid}, 601626 (3), Autosomal dominant, Somatic mutation
<i>CHIT1</i>	600031	[Chitotriosidase deficiency], 614122 (3), Autosomal recessive
<i>CHKB</i>	612395	Muscular dystrophy, congenital, megaconial type, 602541 (3), Autosomal recessive
<i>CHM</i>	300390	Choroideremia, 303100 (3), X-linked dominant
<i>CHMP1A</i>	164010	Pontocerebellar hypoplasia, type 8, 614961 (3), Autosomal recessive
<i>CHMP2B</i>	609512	Amyotrophic lateral sclerosis 17, 614696 (3), Autosomal dominant; Dementia, familial, nonspecific, 600795 (3), Autosomal dominant
<i>CHMP4B</i>	610897	Cataract 31, multiple types, 605387 (3), Autosomal dominant
<i>CHN1</i>	118423	Duane retraction syndrome 2, 604356 (3), Autosomal dominant
<i>CHP1</i>	606988	?Spastic ataxia 9, autosomal recessive, 618438 (3), Autosomal recessive
<i>CHRD1</i>	300350	Megalocornea 1, X-linked, 309300 (3), X-linked recessive
<i>CHRM3</i>	118494	Prune belly syndrome, 100100 (3), Autosomal recessive
<i>CHRNA1</i>	100690	Myasthenic syndrome, congenital, 1A, slow-channel, 601462 (3), Autosomal dominant; Myasthenic syndrome, congenital, 1B, fast-channel, 608930 (3), Autosomal recessive, Autosomal dominant; Multiple pterygium syndrome, lethal type, 253290 (3), Autosomal recessive
<i>CHRNA2</i>	118502	Epilepsy, nocturnal frontal lobe, type 4, 610353 (3), Autosomal dominant
<i>CHRNA3</i>	118503	{Lung cancer susceptibility 2}, 612052 (3); Bladder dysfunction, autonomic, with impaired pupillary reflex and secondary CAKUT, 191800 (3), Autosomal recessive
<i>CHRNA4</i>	118504	Epilepsy, nocturnal frontal lobe, 1, 600513 (3), Autosomal dominant; {Nicotine addiction, susceptibility to}, 188890 (3)
<i>CHRNA5</i>	118505	{Lung cancer susceptibility 2}, 612052 (3); {Nicotine dependence, susceptibility to}, 612052 (3)
<i>CHRNA1</i>	100710	?Myasthenic syndrome, congenital, 2C, associated with acetylcholine receptor deficiency, 616314 (3), Autosomal recessive; Myasthenic syndrome, congenital, 2A, slow-channel, 616313 (3), Autosomal dominant
<i>CHRNA2</i>	118507	Epilepsy, nocturnal frontal lobe, 3, 605375 (3)

<i>CHRNA</i>	100720	?Myasthenic syndrome, congenital, 3C, associated with acetylcholine receptor deficiency, 616323 (3), Autosomal recessive; ?Myasthenic syndrome, congenital, 3A, slow-channel, 616321 (3), Autosomal dominant; Myasthenic syndrome, congenital, 3B, fast-channel, 616322 (3), Autosomal recessive; Multiple pterygium syndrome, lethal type, 253290 (3), Autosomal recessive
<i>CHRNA</i>	100725	Myasthenic syndrome, congenital, 4A, slow-channel, 605809 (3), Autosomal recessive, Autosomal dominant; Myasthenic syndrome, congenital, 4C, associated with acetylcholine receptor deficiency, 608931 (3), Autosomal recessive; Myasthenic syndrome, congenital, 4B, fast-channel, 616324 (3), Autosomal recessive
<i>CHRNA</i>	100730	Escobar syndrome, 265000 (3), Autosomal recessive; Multiple pterygium syndrome, lethal type, 253290 (3), Autosomal recessive
<i>CHST11</i>	610128	?Osteochondrodysplasia, brachydactyly, and overlapping malformed digits, 618167 (3), Autosomal recessive
<i>CHST14</i>	608429	Ehlers-Danlos syndrome, musculocontractural type 1, 601776 (3), Autosomal recessive
<i>CHST3</i>	603799	Spondyloepiphyseal dysplasia with congenital joint dislocations, 143095 (3), Autosomal recessive
<i>CHST6</i>	605294	Macular corneal dystrophy, 217800 (3), Autosomal recessive
<i>CHST8</i>	610190	?Peeling skin syndrome 3, 616265 (3), Autosomal recessive
<i>CHSY1</i>	608183	Temtamy preaxial brachydactyly syndrome, 605282 (3), Autosomal recessive
<i>CHUK</i>	600664	Cocoon syndrome, 613630 (3)
<i>CIB1</i>	602293	Epidermodysplasia verruciformis 3, 618267 (3), Autosomal recessive
<i>CIB2</i>	605564	Deafness, autosomal recessive 48, 609439 (3), Autosomal recessive; Usher syndrome, type II, 614869 (3), Autosomal recessive
<i>CIBAR1 (FAM92A)</i>	617273	?Polydactyly, postaxial, type A9, 618219 (3), Autosomal recessive
<i>CIC</i>	612082	Mental retardation, autosomal dominant 45, 617600 (3), Autosomal dominant
<i>CIDEC</i>	612120	?Lipodystrophy, familial partial, type 5, 615238 (3), Autosomal recessive
<i>CIITA</i>	600005	Bare lymphocyte syndrome, type II, complementation group A, 209920 (3), Autosomal recessive; {Rheumatoid arthritis, susceptibility to}, 180300 (3)
<i>CILK1 (ICK)</i>	612325	Endocrine-cerebroosteodysplasia, 612651 (3), Autosomal recessive; {Epilepsy, juvenile myoclonic, susceptibility to, 10}, 617924 (3), Autosomal dominant
<i>CILP</i>	603489	{Lumbar disc disease, susceptibility to}, 603932 (3)
<i>CISD2</i>	611507	Wolfram syndrome 2, 604928 (3), Autosomal recessive

<i>CISH</i>	602441	{Malaria, susceptibility to}, 611162 (3); {Tuberculosis, susceptibility to}, 607948 (3); {Bacteremia, susceptibility to}, 614383 (3)
<i>CIT</i>	605629	Microcephaly 17, primary, autosomal recessive, 617090 (3), Autosomal recessive
<i>CITED2</i>	602937	Atrial septal defect 8, 614433 (3), Autosomal dominant; Ventricular septal defect 2, 614431 (3), Autosomal dominant
<i>CKAP2L</i>	616174	Filippi syndrome, 272440 (3), Autosomal recessive
<i>CLCC1</i>	617539	Retinitis pigmentosa 32, 609913 (3), Autosomal recessive
<i>CLCF1</i>	607672	Cold-induced sweating syndrome 2, 610313 (3), Autosomal recessive
<i>CLCN1</i>	118425	Myotonia congenita, dominant, 160800 (3), Autosomal dominant; Myotonia levior, recessive (3); Myotonia congenita, recessive, 255700 (3), Autosomal recessive
<i>CLCN2</i>	600570	{Epilepsy, juvenile absence, susceptibility to, 2}, 607628 (3), Autosomal dominant; {Epilepsy, idiopathic generalized, susceptibility to, 11}, 607628 (3), Autosomal dominant; {Epilepsy, juvenile myoclonic, susceptibility to, 8}, 607628 (3), Autosomal dominant; Leukoencephalopathy with ataxia, 615651 (3), Autosomal recessive; Hyperaldosteronism, familial, type II, 605635 (3), Autosomal dominant
<i>CLCN4</i>	302910	Raynaud-Claes syndrome, 300114 (3), X-linked dominant
<i>CLCN5</i>	300008	Proteinuria, low molecular weight, with hypercalciuric nephrocalcinosis, 308990 (3), X-linked recessive; Dent disease, 300009 (3), X-linked recessive; Hypophosphatemic rickets, 300554 (3), X-linked recessive; Nephrolithiasis, type I, 310468 (3), X-linked recessive
<i>CLCN7</i>	602727	Osteopetrosis, autosomal recessive 4, 611490 (3), Autosomal recessive; Osteopetrosis, autosomal dominant 2, 166600 (3), Autosomal dominant; Hypopigmentation, organomegaly, and delayed myelination and development, 618541 (3), Autosomal dominant
<i>CLCNKA</i>	602024	Bartter syndrome, type 4b, digenic, 613090 (3), Digenic recessive
<i>CLCNKB</i>	602023	Bartter syndrome, type 3, 607364 (3), Autosomal recessive; Bartter syndrome, type 4b, digenic, 613090 (3), Digenic recessive
<i>CLDN1</i>	603718	Ichthyosis, leukocyte vacuoles, alopecia, and sclerosing cholangitis, 607626 (3), Autosomal recessive
<i>CLDN10</i>	617579	HELIX syndrome, 617671 (3), Autosomal recessive
<i>CLDN14</i>	605608	Deafness, autosomal recessive 29, 614035 (3), Autosomal recessive
<i>CLDN16</i>	603959	Hypomagnesemia 3, renal, 248250 (3), Autosomal recessive
<i>CLDN19</i>	610036	Hypomagnesemia 5, renal, with ocular involvement, 248190 (3), Autosomal recessive
<i>CLEC1A</i>	606782	{Aspergillosis, susceptibility to}, 614079 (3)
<i>CLEC7A</i>	606264	{Aspergillosis, susceptibility to}, 614079 (3); Candidiasis, familial, 4, autosomal recessive, 613108 (3), Autosomal recessive

<i>CLIC2</i>	300138	?Mental retardation, X-linked, syndromic 32, 300886 (3), X-linked recessive
<i>CLIC5</i>	607293	?Deafness, autosomal recessive 103, 616042 (3), Autosomal recessive
<i>CLMP</i>	611693	Congenital short bowel syndrome, 615237 (3), Autosomal recessive
<i>CLN3</i>	607042	Ceroid lipofuscinosis, neuronal, 3, 204200 (3), Autosomal recessive
<i>CLN5</i>	608102	Ceroid lipofuscinosis, neuronal, 5, 256731 (3), Autosomal recessive
<i>CLN6</i>	606725	Ceroid lipofuscinosis, neuronal, Kufs type, adult onset, 204300 (3), Autosomal recessive; Ceroid lipofuscinosis, neuronal, 6, 601780 (3), Autosomal recessive
<i>CLN8</i>	607837	Ceroid lipofuscinosis, neuronal, 8, 600143 (3), Autosomal recessive; Ceroid lipofuscinosis, neuronal, 8, Northern epilepsy variant, 610003 (3), Autosomal recessive
<i>CLP1</i>	608757	Pontocerebellar hypoplasia, type 10, 615803 (3), Autosomal recessive
<i>CLPB</i>	616254	3-methylglutaconic aciduria, type VII, with cataracts, neurologic involvement and neutropenia, 616271 (3), Autosomal recessive
<i>CLPP</i>	601119	Perrault syndrome 3, 614129 (3), Autosomal recessive
<i>CLPX</i>	615611	?Protoporphyrria, erythropoietic, 2, 618015 (3), Autosomal dominant
<i>CLRN1</i>	606397	Retinitis pigmentosa 61, 614180 (3); Usher syndrome, type 3A, 276902 (3), Autosomal recessive
<i>CLTC</i>	118955	Mental retardation, autosomal dominant 56, 617854 (3), Autosomal dominant
<i>CNBP</i>	116955	Myotonic dystrophy 2, 602668 (3), Autosomal dominant
<i>CNGA1</i>	123825	Retinitis pigmentosa 49, 613756 (3)
<i>CNGA3</i>	600053	Achromatopsia 2, 216900 (3), Autosomal recessive
<i>CNGB1</i>	600724	Retinitis pigmentosa 45, 613767 (3), Autosomal recessive
<i>CNGB3</i>	605080	Achromatopsia 3, 262300 (3), Autosomal recessive
<i>CNKS2</i>	300724	Mental retardation, X-linked, syndromic, Houge type, 301008 (3), X-linked
<i>CNNM2</i>	607803	Hypomagnesemia 6, renal, 613882 (3), Autosomal dominant; Hypomagnesemia, seizures, and mental retardation, 616418 (3), Autosomal recessive, Autosomal dominant
<i>CNNM4</i>	607805	Jalili syndrome, 217080 (3), Autosomal recessive
<i>CNOT1</i>	604917	Holoprosencephaly 12, with or without pancreatic agenesis, 618500 (3), Autosomal dominant
<i>CNOT2</i>	604909	Intellectual developmental disorder with nasal speech, dysmorphic facies, and variable skeletal anomalies, 618608 (3), Autosomal dominant
<i>CNOT3</i>	604910	Intellectual developmental disorder with speech delay, autism, and dysmorphic facies, 618672 (3), Autosomal dominant

<i>CNPY3</i>	610774	Epileptic encephalopathy, early infantile, 60, 617929 (3), Autosomal recessive
<i>CNTN1</i>	600016	?Myopathy, congenital, Compton-North, 612540 (3), Autosomal recessive
<i>CNTN2</i>	190197	?Epilepsy, myoclonic, familial adult, 5, 615400 (3), Autosomal recessive
<i>CNTNAP1</i>	602346	Lethal congenital contracture syndrome 7, 616286 (3), Autosomal recessive; Hypomyelinating neuropathy, congenital, 3, 618186 (3), Autosomal recessive
<i>CNTNAP2</i>	604569	{Autism susceptibility 15}, 612100 (3); Pitt-Hopkins like syndrome 1, 610042 (3), Autosomal recessive; Cortical dysplasia-focal epilepsy syndrome, 610042 (3), Autosomal recessive
<i>COA3</i>	614775	?Mitochondrial complex IV deficiency, 220110 (3), Autosomal recessive, Mitochondrial
<i>COA5</i>	613920	?Cardioencephalomyopathy, fatal infantile, due to cytochrome c oxidase deficiency 3, 616500 (3), Autosomal recessive
<i>COA6</i>	614772	Cardioencephalomyopathy, fatal infantile, due to cytochrome c oxidase deficiency 4, 616501 (3), Autosomal recessive
<i>COA7</i>	615623	Spinocerebellar ataxia, autosomal recessive, with axonal neuropathy 3, 618387 (3), Autosomal recessive
<i>COA8 (APOPT1)</i>	616003	Mitochondrial complex IV deficiency, 220110 (3), Autosomal recessive, Mitochondrial
<i>COASY</i>	609855	Neurodegeneration with brain iron accumulation 6, 615643 (3), Autosomal recessive; Pontocerebellar hypoplasia, type 12, 618266 (3), Autosomal recessive
<i>COCH</i>	603196	?Deafness, autosomal recessive 110, 618094 (3), Autosomal recessive; Deafness, autosomal dominant 9, 601369 (3), Autosomal dominant
<i>COG1</i>	606973	Congenital disorder of glycosylation, type IIg, 611209 (3), Autosomal recessive
<i>COG2</i>	606974	?Congenital disorder of glycosylation, type IIq, 617395 (3), Autosomal recessive
<i>COG4</i>	606976	Saul-Wilson syndrome, 618150 (3), Autosomal dominant; Congenital disorder of glycosylation, type IIj, 613489 (3), Autosomal recessive
<i>COG5</i>	606821	Congenital disorder of glycosylation, type Ili, 613612 (3), Autosomal recessive
<i>COG6</i>	606977	Congenital disorder of glycosylation, type III, 614576 (3), Autosomal recessive; Shaheen syndrome, 615328 (3), Autosomal recessive
<i>COG7</i>	606978	Congenital disorder of glycosylation, type Iie, 608779 (3), Autosomal recessive
<i>COG8</i>	606979	Congenital disorder of glycosylation, type IIh, 611182 (3)
<i>COL10A1</i>	120110	Metaphyseal chondrodysplasia, Schmid type, 156500 (3), Autosomal dominant

<i>COL11A1</i>	120280	Stickler syndrome, type II, 604841 (3), Autosomal dominant; Marshall syndrome, 154780 (3), Autosomal dominant; ?Deafness, autosomal dominant 37, 618533 (3), Autosomal dominant; {Lumbar disc herniation, susceptibility to}, 603932 (3); Fibrochondrogenesis 1, 228520 (3), Autosomal recessive
<i>COL11A2</i>	120290	Deafness, autosomal dominant 13, 601868 (3), Autosomal dominant; Otospondylomegaepiphyseal dysplasia, autosomal recessive, 215150 (3), Autosomal recessive; Fibrochondrogenesis 2, 614524 (3), Autosomal recessive, Autosomal dominant; Deafness, autosomal recessive 53, 609706 (3), Autosomal recessive; Otospondylomegaepiphyseal dysplasia, autosomal dominant, 184840 (3), Autosomal dominant
<i>COL12A1</i>	120320	Bethlem myopathy 2, 616471 (3), Autosomal dominant; ?Ullrich congenital muscular dystrophy 2, 616470 (3)
<i>COL13A1</i>	120350	Myasthenic syndrome, congenital, 19, 616720 (3), Autosomal recessive
<i>COL17A1</i>	113811	Epithelial recurrent erosion dystrophy, 122400 (3), Autosomal dominant; Epidermolysis bullosa, junctional, non-Herlitz type, 226650 (3), Autosomal recessive; Epidermolysis bullosa, junctional, localisata variant, 226650 (3), Autosomal recessive
<i>COL18A1</i>	120328	Knobloch syndrome, type 1, 267750 (3), Autosomal recessive; Glaucoma, primary closed-angle, 618880 (3), Autosomal dominant
<i>COL1A1</i>	120150	Osteogenesis imperfecta, type I, 166200 (3), Autosomal dominant; Osteogenesis imperfecta, type IV, 166220 (3), Autosomal dominant; Osteogenesis imperfecta, type II, 166210 (3), Autosomal dominant; {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 III, 259420 (3), Autosomal dominant
<i>COL1A2</i>	120160	{Osteoporosis, postmenopausal}, 166710 (3), Autosomal dominant; Ehlers-Danlos syndrome, cardiac valvular type, 225320 (3), Autosomal recessive; Ehlers-Danlos syndrome, arthrochalasia type, 2, 617821 (3), Autosomal dominant; Osteogenesis imperfecta, type II, 166210 (3), Autosomal dominant; Osteogenesis imperfecta, type IV, 166220 (3), Autosomal dominant; Osteogenesis imperfecta, type III, 259420 (3), Autosomal dominant
<i>COL25A1</i>	610004	Fibrosis of extraocular muscles, congenital, 5, 616219 (3), Autosomal recessive
<i>COL27A1</i>	608461	Steel syndrome, 615155 (3), Autosomal recessive

<i>COL2A1</i>	120140	Vitreoretinopathy with phalangeal epiphyseal dysplasia (3); Achondrogenesis, type II or hypochondrogenesis, 200610 (3), Autosomal dominant; Spondyloperipheral dysplasia, 271700 (3), Autosomal dominant; Kniest dysplasia, 156550 (3), Autosomal dominant; Stickler syndrome, type I, 108300 (3), Autosomal dominant; Osteoarthritis with mild chondrodysplasia, 604864 (3), Autosomal dominant; Platyspondylic skeletal dysplasia, Torrance type, 151210 (3), Autosomal dominant; Avascular necrosis of the femoral head, 608805 (3), Autosomal dominant; ?Epiphyseal dysplasia, multiple, with myopia and deafness, 132450 (3), Autosomal dominant; SED congenita, 183900 (3), Autosomal dominant; Legg-Calve-Perthes disease, 150600 (3), Autosomal dominant; SMED Strudwick type, 184250 (3), Autosomal dominant; Czech dysplasia, 609162 (3), Autosomal dominant; Stickler syndrome, type I, nonsyndromic ocular, 609508 (3), Autosomal dominant; Spondyloepiphyseal dysplasia, Stanescu type, 616583 (3), Autosomal dominant
<i>COL3A1</i>	120180	Ehlers-Danlos syndrome, vascular type, 130050 (3), Autosomal dominant; Polymicrogyria with or without vascular-type EDS, 618343 (3), Autosomal recessive
<i>COL4A1</i>	120130	Angiopathy, hereditary, with nephropathy, aneurysms, and muscle cramps, 611773 (3), Autosomal dominant; Brain small vessel disease with or without ocular anomalies, 175780 (3), Autosomal dominant; {Hemorrhage, intracerebral, susceptibility to}, 614519 (3); ?Retinal arteries, tortuosity of, 180000 (3), Autosomal dominant; Microangiopathy and leukoencephalopathy, pontine, autosomal dominant, 618564 (3), Autosomal dominant
<i>COL4A2</i>	120090	Brain small vessel disease 2, 614483 (3), Autosomal dominant; {Hemorrhage, intracerebral, susceptibility to}, 614519 (3)
<i>COL4A3</i>	120070	Alport syndrome 2, autosomal recessive, 203780 (3), Autosomal recessive; Alport syndrome 3, autosomal dominant, 104200 (3), Autosomal dominant; Hematuria, benign familial, 141200 (3), Autosomal dominant
<i>COL4A4</i>	120131	Alport syndrome 2, autosomal recessive, 203780 (3), Autosomal recessive; Hematuria, familial benign, 141200 (3), Autosomal dominant
<i>COL4A5</i>	303630	Alport syndrome 1, X-linked, 301050 (3), X-linked dominant
<i>COL4A6</i>	303631	?Deafness, X-linked 6, 300914 (3), X-linked recessive
<i>COL5A1</i>	120215	Ehlers-Danlos syndrome, classic type, 1, 130000 (3), Autosomal dominant
<i>COL5A2</i>	120190	Ehlers-Danlos syndrome, classic type, 2, 130010 (3), Autosomal dominant
<i>COL6A1</i>	120220	Ullrich congenital muscular dystrophy 1, 254090 (3), Autosomal recessive, Autosomal dominant; Bethlem myopathy 1, 158810 (3), Autosomal recessive, Autosomal dominant

<i>COL6A2</i>	120240	Bethlem myopathy 1, 158810 (3), Autosomal recessive, Autosomal dominant; Ullrich congenital muscular dystrophy 1, 254090 (3), Autosomal recessive, Autosomal dominant; ?Myosclerosis, congenital, 255600 (3), Autosomal recessive
<i>COL6A3</i>	120250	Bethlem myopathy 1, 158810 (3), Autosomal recessive, Autosomal dominant; Dystonia 27, 616411 (3), Autosomal recessive; Ullrich congenital muscular dystrophy 1, 254090 (3), Autosomal recessive, Autosomal dominant
<i>COL7A1</i>	120120	EBD inversa, 226600 (3), Autosomal recessive; Epidermolysis bullosa dystrophica, AR, 226600 (3), Autosomal recessive; Toenail dystrophy, isolated, 607523 (3), Autosomal dominant; EBD, localisata variant (3); EBD, Bart type, 132000 (3), Autosomal dominant; Transient bullous of the newborn, 131705 (3), Autosomal recessive, Autosomal dominant; Epidermolysis bullosa dystrophica, AD, 131750 (3), Autosomal dominant; Epidermolysis bullosa pruriginosa, 604129 (3), Autosomal recessive, Autosomal dominant; Epidermolysis bullosa, pretibial, 131850 (3), Autosomal recessive, Autosomal dominant
<i>COL8A2</i>	120252	Corneal dystrophy, posterior polymorphous 2, 609140 (3), Autosomal dominant; Corneal dystrophy, Fuchs endothelial, 1, 136800 (3), Autosomal dominant
<i>COL9A1</i>	120210	?Epiphyseal dysplasia, multiple, 6, 614135 (3), Autosomal dominant; Stickler syndrome, type IV, 614134 (3)
<i>COL9A2</i>	120260	?Stickler syndrome, type V, 614284 (3), Autosomal recessive; Epiphyseal dysplasia, multiple, 2, 600204 (3), Autosomal dominant
<i>COL9A3</i>	120270	Epiphyseal dysplasia, multiple, 3, with or without myopathy, 600969 (3), Autosomal dominant; {Intervertebral disc disease, susceptibility to}, 603932 (3)
<i>COLEC10</i>	607620	3MC syndrome 3, 248340 (3), Autosomal recessive
<i>COLEC11</i>	612502	3MC syndrome 2, 265050 (3), Autosomal recessive
<i>COLGALT1</i>	617531	Brain small vessel disease 3, 618360 (3), Autosomal recessive
<i>COLQ</i>	603033	Myasthenic syndrome, congenital, 5, 603034 (3), Autosomal recessive
<i>COMP</i>	600310	Epiphyseal dysplasia, multiple, 1, 132400 (3), Autosomal dominant; Pseudoachondroplasia, 177170 (3), Autosomal dominant
<i>COMT</i>	116790	{Schizophrenia, susceptibility to}, 181500 (3), Autosomal dominant; {Panic disorder, susceptibility to}, 167870 (3), ?Autosomal dominant
<i>COPA</i>	601924	{Autoimmune interstitial lung, joint, and kidney disease}, 616414 (3), Autosomal dominant
<i>COPB2</i>	606990	?Microcephaly 19, primary, autosomal recessive, 617800 (3), Autosomal recessive

<i>COQ2</i>	609825	{Multiple system atrophy, susceptibility to}, 146500 (3), Autosomal recessive, Autosomal dominant; Coenzyme Q10 deficiency, primary, 1, 607426 (3), Autosomal recessive
<i>COQ4</i>	612898	Coenzyme Q10 deficiency, primary, 7, 616276 (3), Autosomal recessive
<i>COQ6</i>	614647	Coenzyme Q10 deficiency, primary, 6, 614650 (3), Autosomal recessive
<i>COQ7</i>	601683	?Coenzyme Q10 deficiency, primary, 8, 616733 (3), Autosomal recessive
<i>COQ8A</i>	606980	Coenzyme Q10 deficiency, primary, 4, 612016 (3), Autosomal recessive
<i>COQ8B</i>	615567	Nephrotic syndrome, type 9, 615573 (3), Autosomal recessive
<i>COQ9</i>	612837	Coenzyme Q10 deficiency, primary, 5, 614654 (3), Autosomal recessive
<i>CORIN</i>	605236	Preeclampsia/eclampsia 5, 614595 (3)
<i>CORO1A</i>	605000	Immunodeficiency 8, 615401 (3), Autosomal recessive
<i>COX10</i>	602125	Mitochondrial complex IV deficiency, 220110 (3), Autosomal recessive, Mitochondrial; Leigh syndrome due to mitochondrial COX4 deficiency, 256000 (3), Autosomal recessive, Mitochondrial
<i>COX14</i>	614478	?Mitochondrial complex IV deficiency, 220110 (3), Autosomal recessive, Mitochondrial
<i>COX15</i>	603646	Cardioencephalomyopathy, fatal infantile, due to cytochrome c oxidase deficiency 2, 615119 (3), Autosomal recessive; Leigh syndrome due to cytochrome c oxidase deficiency, 256000 (3), Autosomal recessive, Mitochondrial
<i>COX20</i>	614698	Mitochondrial complex IV deficiency, 220110 (3), Autosomal recessive, Mitochondrial
<i>COX4I2</i>	607976	Exocrine pancreatic insufficiency, dyserythropoietic anemia, and calvarial hyperostosis, 612714 (3), Autosomal recessive
<i>COX6A1</i>	602072	Charcot-Marie-Tooth disease, recessive intermediate D, 616039 (3), Autosomal recessive
<i>COX6A2</i>	602009	Mitochondrial complex IV deficiency, 220110 (3), Autosomal recessive, Mitochondrial
<i>COX6B1</i>	124089	Mitochondrial complex IV deficiency, 220110 (3), Autosomal recessive, Mitochondrial
<i>COX7B</i>	300885	Linear skin defects with multiple congenital anomalies 2, 300887 (3), X-linked dominant
<i>COX8A</i>	123870	?Mitochondrial complex IV deficiency, 220110 (3), Autosomal recessive, Mitochondrial
<i>CP</i>	117700	[Hypoceruloplasminemia, hereditary], 604290 (3), Autosomal recessive; Hemosiderosis, systemic, due to aceruloplasminemia, 604290 (3), Autosomal recessive; Cerebellar ataxia, 604290 (3), Autosomal recessive

<i>CPA6</i>	609562	Febrile seizures, familial, 11, 614418 (3), Autosomal recessive; Epilepsy, familial temporal lobe, 5, 614417 (3), Autosomal recessive, Autosomal dominant
<i>CPAMD8</i>	608841	Anterior segment dysgenesis 8, 617319 (3), Autosomal recessive
<i>CPLANE1</i>	614571	Joubert syndrome 17, 614615 (3), Autosomal recessive; Orofaciodigital syndrome VI, 277170 (3), Autosomal recessive
<i>CPLX1</i>	605032	Epileptic encephalopathy, early infantile, 63, 617976 (3), Autosomal recessive
<i>CPN1</i>	603103	Carboxypeptidase N deficiency, 212070 (3), Autosomal recessive
<i>CPOX</i>	612732	Harderoporphyria, 618892 (3), Autosomal recessive; Coproporphyria, 121300 (3), Autosomal recessive, Autosomal dominant
<i>CPS1</i>	608307	Carbamoylphosphate synthetase I deficiency, 237300 (3), Autosomal recessive; {Pulmonary hypertension, neonatal, susceptibility to}, 615371 (3)
<i>CPSF1</i>	606027	Myopia 27, 618827 (3), Autosomal dominant
<i>CPT1A</i>	600528	CPT deficiency, hepatic, type IA, 255120 (3), Autosomal recessive
<i>CPT1C</i>	608846	?Spastic paraplegia 73, autosomal dominant, 616282 (3), Autosomal dominant
<i>CPT2</i>	600650	CPT II deficiency, myopathic, stress-induced, 255110 (3), Autosomal recessive, Autosomal dominant; CPT II deficiency, infantile, 600649 (3), Autosomal recessive; CPT II deficiency, lethal neonatal, 608836 (3), Autosomal recessive; {Encephalopathy, acute, infection-induced, 4, susceptibility to}, 614212 (3), Autosomal recessive, Autosomal dominant
<i>CR1</i>	120620	[Blood group, Knops system], 607486 (3); {Malaria, severe, resistance to}, 611162 (3)
<i>CR2</i>	120650	{Systemic lupus erythematosus, susceptibility to, 9}, 610927 (3); Immunodeficiency, common variable, 7, 614699 (3), Autosomal recessive
<i>CRADD</i>	603454	Mental retardation, autosomal recessive 34, with variant lissencephaly, 614499 (3), Autosomal recessive
<i>CRAT</i>	600184	?Neurodegeneration with brain iron accumulation 8, 617917 (3), Autosomal recessive
<i>CRB1</i>	604210	Pigmented paravenous chorioretinal atrophy, 172870 (3), Autosomal dominant; Retinitis pigmentosa-12, 600105 (3), Autosomal recessive; Leber congenital amaurosis 8, 613835 (3), Autosomal recessive
<i>CRB2</i>	609720	Ventriculomegaly with cystic kidney disease, 219730 (3), Autosomal recessive; Focal segmental glomerulosclerosis 9, 616220 (3), Autosomal recessive
<i>CRBN</i>	609262	Mental retardation, autosomal recessive 2, 607417 (3), Autosomal recessive
<i>CREB1</i>	123810	Histiocytoma, angiomatoid fibrous, somatic, 612160 (3)
<i>CREB3L1</i>	616215	Osteogenesis imperfecta, type XVI, 616229 (3), Autosomal recessive

<i>CREBBP</i>	600140	Rubinstein-Taybi syndrome 1, 180849 (3), Autosomal dominant; Menke-Hennekam syndrome 1, 618332 (3), Autosomal dominant
<i>CRELD1</i>	607170	Atrioventricular septal defect, partial, with heterotaxy syndrome, 606217 (3), Autosomal dominant; {Atrioventricular septal defect, susceptibility to, 2}, 606217 (3), Autosomal dominant
<i>CRIP1</i>	604594	Short stature with microcephaly and distinctive facies, 615789 (3), Autosomal recessive
<i>CRLF1</i>	604237	Cold-induced sweating syndrome 1, 272430 (3), Autosomal recessive
<i>CRPPA (ISPD)</i>	614631	Muscular dystrophy-dystroglycanopathy (limb-girdle), type C, 7, 616052 (3), Autosomal recessive; Muscular dystrophy-dystroglycanopathy (congenital with brain and eye anomalies), type A, 7, 614643 (3), Autosomal recessive
<i>CRTAP</i>	605497	Osteogenesis imperfecta, type VII, 610682 (3), Autosomal recessive
<i>CRTC1</i>	607536	Mucoepidermoid salivary gland carcinoma (3)
<i>CRX</i>	602225	Cone-rod retinal dystrophy-2, 120970 (3), Autosomal dominant; Leber congenital amaurosis 7, 613829 (3)
<i>CRY1</i>	601933	{Delayed sleep phase disorder, susceptibility to}, 614163 (3), Autosomal dominant
<i>CRYAA</i>	123580	Cataract 9, multiple types, 604219 (3), Autosomal recessive, Autosomal dominant
<i>CRYAB</i>	123590	Myopathy, myofibrillar, 2, 608810 (3), Autosomal dominant; Cardiomyopathy, dilated, 1II, 615184 (3), Autosomal dominant; Myopathy, myofibrillar, fatal infantile hypertonic, alpha-B crystallin-related, 613869 (3), Autosomal recessive; Cataract 16, multiple types, 613763 (3), Autosomal recessive, Autosomal dominant
<i>CRYBA1</i>	123610	Cataract 10, multiple types, 600881 (3), Autosomal dominant
<i>CRYBA2</i>	600836	?Cataract 42, 115900 (3), Autosomal dominant
<i>CRYBA4</i>	123631	Cataract 23, 610425 (3)
<i>CRYBB1</i>	600929	Cataract 17, multiple types, 611544 (3), Autosomal recessive, Autosomal dominant
<i>CRYBB2</i>	123620	Cataract 3, multiple types, 601547 (3), Autosomal dominant
<i>CRYBB3</i>	123630	Cataract 22, 609741 (3), Autosomal recessive, Autosomal dominant
<i>CRYGB</i>	123670	Cataract 39, multiple types, autosomal dominant, 615188 (3), Autosomal dominant
<i>CRYGC</i>	123680	Cataract 2, multiple types, 604307 (3), Autosomal dominant
<i>CRYGD</i>	123690	Cataract 4, multiple types, 115700 (3), Autosomal dominant
<i>CRYGS</i>	123730	Cataract 20, multiple types, 116100 (3), Autosomal dominant
<i>CRYM</i>	123740	Deafness, autosomal dominant 40, 616357 (3), Autosomal dominant

<i>CSF1R</i>	164770	Brain abnormalities, neurodegeneration, and dysosteosclerosis, 618476 (3), Autosomal recessive; Leukoencephalopathy, diffuse hereditary, with spheroids, 221820 (3), Autosomal dominant
<i>CSF2RA</i>	306250	Surfactant metabolism dysfunction, pulmonary, 4, 300770 (3)
<i>CSF2RB</i>	138981	Surfactant metabolism dysfunction, pulmonary, 5, 614370 (3), Autosomal recessive
<i>CSF3R</i>	138971	Neutropenia, severe congenital, 7, autosomal recessive, 617014 (3), Autosomal recessive
<i>CSGALNACT1</i>	616615	Skeletal dysplasia, mild, with joint laxity and advanced bone age, 618870 (3), Autosomal recessive
<i>CSH1</i>	150200	[Placental lactogen deficiency] (1)
<i>CSNK1D</i>	600864	Advanced sleep-phase syndrome, familial, 2, 615224 (3), Autosomal dominant
<i>CSNK2A1</i>	115440	Okur-Chung neurodevelopmental syndrome, 617062 (3), Autosomal dominant
<i>CSNK2B</i>	115441	Poirier-Bienvenu neurodevelopmental syndrome, 618732 (3), Autosomal dominant
<i>CSPP1</i>	611654	Joubert syndrome 21, 615636 (3), Autosomal recessive
<i>CSR3P</i>	600824	Cardiomyopathy, hypertrophic, 12, 612124 (3), Autosomal dominant; ?Cardiomyopathy, dilated, 1M, 607482 (3)
<i>CST3</i>	604312	Cerebral amyloid angiopathy, 105150 (3), Autosomal dominant; {Macular degeneration, age-related, 11}, 611953 (3)
<i>CST6</i>	601891	?Ectodermal dysplasia 15, hypohidrotic/hair type, 618535 (3), Autosomal recessive
<i>CSTA</i>	184600	Peeling skin syndrome 4, 607936 (3), Autosomal recessive
<i>CSTB</i>	601145	Epilepsy, progressive myoclonic 1A (Unverricht and Lundborg), 254800 (3), Autosomal recessive
<i>CTBP1</i>	602618	Hypotonia, ataxia, developmental delay, and tooth enamel defect syndrome, 617915 (3), Autosomal dominant
<i>CTC1</i>	613129	Cerebroretinal microangiopathy with calcifications and cysts, 612199 (3), Autosomal recessive
<i>CTCF</i>	604167	Mental retardation, autosomal dominant 21, 615502 (3), Autosomal dominant
<i>CTDP1</i>	604927	Congenital cataracts, facial dysmorphism, and neuropathy, 604168 (3), Autosomal recessive
<i>CTH</i>	607657	Cystathioninuria, 219500 (3), Autosomal recessive
<i>CTHRC1</i>	610635	Barrett esophagus/esophageal adenocarcinoma, 614266 (3)
<i>CTLA4</i>	123890	{Systemic lupus erythematosus, susceptibility to}, 152700 (3), Autosomal dominant; {Diabetes mellitus, insulin-dependent, 12}, 601388 (3); {Celiac disease, susceptibility to, 3}, 609755 (3); Autoimmune lymphoproliferative syndrome, type V, 616100 (3), Autosomal dominant; {Hashimoto thyroiditis}, 140300 (3), Autosomal dominant
<i>CTNNA1</i>	116805	Macular dystrophy, patterned, 2, 608970 (3), Autosomal dominant

<i>CTNNA2</i>	114025	Cortical dysplasia, complex, with other brain malformations 9, 618174 (3), Autosomal recessive
<i>CTNNA3</i>	607667	Arrhythmogenic right ventricular dysplasia, familial, 13, 615616 (3), Autosomal dominant
<i>CTNNB1</i>	116806	Ovarian cancer, somatic, 167000 (3); Colorectal cancer, somatic, 114500 (3); Medulloblastoma, somatic, 155255 (3); Hepatocellular carcinoma, somatic, 114550 (3); Pilomatricoma, somatic, 132600 (3); Neurodevelopmental disorder with spastic diplegia and visual defects, 615075 (3), Autosomal dominant; Exudative vitreoretinopathy 7, 617572 (3), Autosomal dominant
<i>CTNND1</i>	601045	Blepharocheilodontic syndrome 2, 617681 (3), Autosomal dominant
<i>CTNS</i>	606272	Cystinosis, nephropathic, 219800 (3), Autosomal recessive; Cystinosis, ocular nonnephropathic, 219750 (3), Autosomal recessive; Cystinosis, late-onset juvenile or adolescent nephropathic, 219900 (3), Autosomal recessive; Cystinosis, atypical nephropathic, 219800 (3), Autosomal recessive
<i>CTPS1</i>	123860	Immunodeficiency 24, 615897 (3), Autosomal recessive
<i>CTRC</i>	601405	{Pancreatitis, chronic, susceptibility to}, 167800 (3), Autosomal dominant
<i>CTSA</i>	613111	Galactosialidosis, 256540 (3), Autosomal recessive
<i>CTSB</i>	116810	Keratolytic winter erythema, 148370 (4), Autosomal dominant
<i>CTSC</i>	602365	Periodontitis 1, juvenile, 170650 (3), Autosomal recessive; Papillon-Lefevre syndrome, 245000 (3), Autosomal recessive; Haim-Munk syndrome, 245010 (3), Autosomal recessive
<i>CTSD</i>	116840	Ceroid lipofuscinosis, neuronal, 10, 610127 (3), Autosomal recessive
<i>CTSF</i>	603539	Ceroid lipofuscinosis, neuronal, 13, Kufs type, 615362 (3), Autosomal recessive
<i>CTSK</i>	601105	Pycnodysostosis, 265800 (3), Autosomal recessive
<i>CTU2</i>	617057	Microcephaly, facial dysmorphism, renal agenesis, and ambiguous genitalia syndrome, 618142 (3), Autosomal recessive
<i>CUBN</i>	602997	[Proteinuria, chronic benign], 618884 (3), Autosomal recessive; Imlerslund-Grasbeck syndrome 1, 261100 (3), Autosomal recessive
<i>CUL3</i>	603136	Pseudohypoaldosteronism, type IIE, 614496 (3), Autosomal dominant
<i>CUL4B</i>	300304	Mental retardation, X-linked, syndromic 15 (Cabezas type), 300354 (3), X-linked recessive
<i>CUL7</i>	609577	3-M syndrome 1, 273750 (3), Autosomal recessive
<i>CUX1</i>	116896	Global developmental delay with or without impaired intellectual development, 618330 (3), Autosomal dominant
<i>CUX2</i>	610648	Epileptic encephalopathy, early infantile, 67, 618141 (3), Autosomal dominant
<i>CWC27</i>	617170	Retinitis pigmentosa with or without skeletal anomalies, 250410 (3), Autosomal recessive

<i>CWF19L1</i>	616120	Spinocerebellar ataxia, autosomal recessive 17, 616127 (3), Autosomal recessive
<i>CX3CR1</i>	601470	{Rapid progression to AIDS from HIV1 infection}, 609423 (3); {Macular degeneration, age-related, 12}, 613784 (3); {Coronary artery disease, resistance to}, 607339 (3)
<i>CXCL12</i>	600835	{AIDS, resistance to}, 609423 (3)
<i>CXCR1</i>	146929	{AIDS, slow progression to}, 609423 (3)
<i>CXCR4</i>	162643	Myelokathexis, isolated (3); WHIM syndrome, 193670 (3), Autosomal dominant
<i>CXorf56</i>	301012	?Mental retardation, X-linked 107, 301013 (3), X-linked
<i>CYB561</i>	600019	Orthostatic hypotension 2, 618182 (3), Autosomal recessive
<i>CYB5A</i>	613218	Methemoglobinemia and ambiguous genitalia, 250790 (3), Autosomal recessive
<i>CYB5R3</i>	613213	Methemoglobinemia, type I, 250800 (3), Autosomal recessive; Methemoglobinemia, type II, 250800 (3), Autosomal recessive
<i>CYBA</i>	608508	Chronic granulomatous disease 4, autosomal recessive, 233690 (3), Autosomal recessive
<i>CYBB</i>	300481	Immunodeficiency 34, mycobacteriosis, X-linked, 300645 (3), X- linked recessive; Chronic granulomatous disease, X-linked, 306400 (3), X-linked recessive
<i>CYBC1</i>	618334	Chronic granulomatous disease 5, autosomal recessive, 618935 (3), Autosomal recessive
<i>CYC1</i>	123980	Mitochondrial complex III deficiency, nuclear type 6, 615453 (3), Autosomal recessive
<i>CYCS</i>	123970	Thrombocytopenia 4, 612004 (3), Autosomal dominant
<i>CYFIP2</i>	606323	Epileptic encephalopathy, early infantile, 65, 618008 (3), Autosomal dominant
<i>CYLD</i>	605018	Cylindromatosis, familial, 132700 (3), Autosomal dominant; Brooke-Spiegler syndrome, 605041 (3), Autosomal dominant; Trichoepithelioma, multiple familial, 1, 601606 (3), Autosomal dominant
<i>CYP11A1</i>	118485	Adrenal insufficiency, congenital, with 46XY sex reversal, partial or complete, 613743 (3)
<i>CYP11B1</i>	610613	Adrenal hyperplasia, congenital, due to 11-beta-hydroxylase deficiency, 202010 (3), Autosomal recessive; Aldosteronism, glucocorticoid-remediable, 103900 (3), Autosomal dominant
<i>CYP11B2</i>	124080	Aldosterone to renin ratio raised (3); Hypoaldosteronism, congenital, due to CMO II deficiency, 610600 (3), Autosomal recessive; {Low renin hypertension, susceptibility to} (3); Hypoaldosteronism, congenital, due to CMO I deficiency, 203400 (3), Autosomal recessive
<i>CYP17A1</i>	609300	17-alpha-hydroxylase/17,20-lyase deficiency, 202110 (3), Autosomal recessive; 17,20-lyase deficiency, isolated, 202110 (3), Autosomal recessive

<i>CYP19A1</i>	107910	Aromatase deficiency, 613546 (3); Aromatase excess syndrome, 139300 (3), Autosomal dominant
<i>CYP1B1</i>	601771	Glaucoma 3A, primary open angle, congenital, juvenile, or adult onset, 231300 (3), Autosomal recessive; Anterior segment dysgenesis 6, multiple subtypes, 617315 (3)
<i>CYP21A2</i>	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
<i>CYP24A1</i>	126065	Hypercalcemia, infantile, 1, 143880 (3), Autosomal recessive
<i>CYP26B1</i>	605207	Craniosynostosis with radiohumeral fusions and other skeletal and craniofacial anomalies, 614416 (3)
<i>CYP26C1</i>	608428	Focal facial dermal dysplasia 4, 614974 (3), Autosomal recessive
<i>CYP27A1</i>	606530	Cerebrotendinous xanthomatosis, 213700 (3), Autosomal recessive
<i>CYP27B1</i>	609506	Vitamin D-dependent rickets, type I, 264700 (3), Autosomal recessive
<i>CYP2A6</i>	122720	Coumarin resistance, 122700 (3), Autosomal dominant; {Lung cancer, resistance to}, 211980 (3), Autosomal dominant, Somatic mutation; {Nicotine addiction, protection from}, 188890 (3)
<i>CYP2B6</i>	123930	{Efavirenz central nervous system toxicity, susceptibility to}, 614546 (3); Efavirenz, poor metabolism of, 614546 (3)
<i>CYP2C19</i>	124020	Clopidogrel, impaired responsiveness to, 609535 (3), Autosomal recessive; Mephenytoin poor metabolizer, 609535 (3), Autosomal recessive; Proguanil poor metabolizer, 609535 (3), Autosomal recessive; Omeprazole poor metabolizer, 609535 (3), Autosomal recessive
<i>CYP2C8</i>	601129	{Drug metabolism, altered, CYP2C8-related}, 618018 (3)
<i>CYP2C9</i>	601130	Warfarin sensitivity, 122700 (3), Autosomal dominant; Tolbutamide poor metabolizer (3)
<i>CYP2D6</i>	124030	{Debrisoquine sensitivity}, 608902 (3), Autosomal recessive; {Codeine sensitivity}, 608902 (3), Autosomal recessive
<i>CYP2R1</i>	608713	Rickets due to defect in vitamin D 25-hydroxylation, 600081 (3), Autosomal recessive
<i>CYP2U1</i>	610670	Spastic paraplegia 56, autosomal recessive, 615030 (3), Autosomal recessive
<i>CYP3A5</i>	605325	{Hypertension, salt-sensitive essential, susceptibility to}, 145500 (3), Multifactorial
<i>CYP4F22</i>	611495	Ichthyosis, congenital, autosomal recessive 5, 604777 (3), Autosomal recessive
<i>CYP4V2</i>	608614	Bietti crystalline corneoretinal dystrophy, 210370 (3), Autosomal recessive
<i>CYP7B1</i>	603711	Spastic paraplegia 5A, autosomal recessive, 270800 (3), Autosomal recessive; Bile acid synthesis defect, congenital, 3, 613812 (3), Autosomal recessive
<i>D2HGDH</i>	609186	D-2-hydroxyglutaric aciduria, 600721 (3), Autosomal recessive

<i>DAB1</i>	603448	Spinocerebellar ataxia 37, 615945 (3), Autosomal dominant
<i>DACT1</i>	607861	?Townes-Brocks syndrome 2, 617466 (3), Autosomal dominant
<i>DAG1</i>	128239	Muscular dystrophy-dystroglycanopathy (limb-girdle), type C, 9, 613818 (3), Autosomal recessive; Muscular dystrophy-dystroglycanopathy (congenital with brain and eye anomalies), type A, 9, 616538 (3), Autosomal recessive
<i>DALRD3</i>	618904	?Epileptic encephalopathy, early infantile, 86, 618910 (3), Autosomal recessive
<i>DAOA</i>	607408	{Schizophrenia}, 181500 (2), Autosomal dominant
<i>DARS1</i> (<i>DARS</i>)	603084	Hypomyelination with brainstem and spinal cord involvement and leg spasticity, 615281 (3), Autosomal recessive
<i>DARS2</i>	610956	Leukoencephalopathy with brain stem and spinal cord involvement and lactate elevation, 611105 (3), Autosomal recessive
<i>DAZL</i>	601486	{Spermatogenic failure, susceptibility to} (3)
<i>DBH</i>	609312	Orthostatic hypotension 1, due to DBH deficiency, 223360 (3), Autosomal recessive
<i>DBT</i>	248610	Maple syrup urine disease, type II, 248600 (3), Autosomal recessive
<i>DCAF17</i>	612515	Woodhouse-Sakati syndrome, 241080 (3), Autosomal recessive
<i>DCAF8</i>	615820	?Giant axonal neuropathy 2, autosomal dominant, 610100 (3), Autosomal dominant
<i>DCC</i>	120470	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; Colorectal cancer, somatic, 114500 (3)
<i>DCDC2</i>	605755	Sclerosing cholangitis, neonatal, 617394 (3), Autosomal recessive; Nephronophthisis 19, 616217 (3), Autosomal recessive; ?Deafness, autosomal recessive 66, 610212 (3), Autosomal recessive
<i>DCHS1</i>	603057	Mitral valve prolapse 2, 607829 (3), Autosomal dominant; Van Maldergem syndrome 1, 601390 (3), Autosomal recessive
<i>DCLRE1C</i>	605988	Omenn syndrome, 603554 (3), Autosomal recessive; Severe combined immunodeficiency, Athabaskan type, 602450 (3), Autosomal recessive
<i>DCN</i>	125255	Corneal dystrophy, congenital stromal, 610048 (3), Autosomal dominant
<i>DCPS</i>	610534	Al-Raqad syndrome, 616459 (3), Autosomal recessive
<i>DCTN1</i>	601143	{Amyotrophic lateral sclerosis, susceptibility to}, 105400 (3), Autosomal recessive, Autosomal dominant; Perry syndrome, 168605 (3), Autosomal dominant; Neuronopathy, distal hereditary motor, type VIIIB, 607641 (3), Autosomal dominant
<i>DCX</i>	300121	Subcortical laminar heterotopia, X-linked, 300067 (3), X-linked; Lissencephaly, X-linked, 300067 (3), X-linked
<i>DCXR</i>	608347	[Pentosuria], 260800 (3), Autosomal recessive

<i>DDB2</i>	600811	Xeroderma pigmentosum, group E, DDB-negative subtype, 278740 (3), Autosomal recessive
<i>DDC</i>	107930	Aromatic L-amino acid decarboxylase deficiency, 608643 (3), Autosomal recessive
<i>DDHD1</i>	614603	Spastic paraplegia 28, autosomal recessive, 609340 (3), Autosomal recessive
<i>DDHD2</i>	615003	Spastic paraplegia 54, autosomal recessive, 615033 (3), Autosomal recessive
<i>DDOST</i>	602202	?Congenital disorder of glycosylation, type 1r, 614507 (3), Autosomal recessive
<i>DDR2</i>	191311	Spondylometaphyseal dysplasia, short limb-hand type, 271665 (3), Autosomal recessive; Warburg-Cinotti syndrome, 618175 (3), Autosomal dominant
<i>DDRKG1</i>	616177	Spondyloepimetaphyseal dysplasia, Shohat type, 602557 (3), Autosomal recessive
<i>DDX11</i>	601150	Warsaw breakage syndrome, 613398 (3), Autosomal recessive
<i>DDX3X</i>	300160	Intellectual developmental disorder, X-linked, syndrome, Snijders Blok type, 300958 (3), X-linked recessive, X-linked dominant
<i>DDX41</i>	608170	{Myeloproliferative/lymphoproliferative neoplasms, familial (multiple types), susceptibility to}, 616871 (3), Autosomal dominant
<i>DDX58</i>	609631	Singleton-Merten syndrome 2, 616298 (3), Autosomal dominant
<i>DDX59</i>	615464	Orofaciodigital syndrome V, 174300 (3), Autosomal recessive
<i>DDX6</i>	600326	Intellectual developmental disorder with impaired language and dysmorphic facies, 618653 (3), Autosomal dominant
<i>DEAF1</i>	602635	Vulto-van Silfout-de Vries syndrome, 615828 (3), Autosomal dominant; Neurodevelopmental disorder with hypotonia, impaired expressive language, and with or without seizures, 617171 (3), Autosomal recessive
<i>DEGS1</i>	615843	Leukodystrophy, hypomyelinating, 18, 618404 (3), Autosomal recessive
<i>DEK</i>	125264	Leukemia, acute nonlymphocytic, 125264 (2)
<i>DENND5A</i>	617278	Epileptic encephalopathy, early infantile, 49, 617281 (3), Autosomal recessive
<i>DEPDC5</i>	614191	Epilepsy, familial focal, with variable foci 1, 604364 (3), Autosomal dominant
<i>DES</i>	125660	?Cardiomyopathy, dilated, 1l, 604765 (3), Autosomal dominant; Scapuloperoneal syndrome, neurogenic, Kaeser type, 181400 (3), Autosomal dominant; Myopathy, myofibrillar, 1, 601419 (3), Autosomal recessive, Autosomal dominant
<i>DGAT1</i>	604900	?Diarrhea 7, protein-losing enteropathy type, 615863 (3), Autosomal recessive
<i>DGKE</i>	601440	{Hemolytic uremic syndrome, atypical, susceptibility to, 7}, 615008 (3), Autosomal recessive; Nephrotic syndrome, type 7, 615008 (3), Autosomal recessive

<i>DGUOK</i>	601465	Progressive external ophthalmoplegia with mitochondrial DNA deletions, autosomal recessive 4, 617070 (3), Autosomal recessive; Portal hypertension, noncirrhotic, 617068 (3), Autosomal recessive; Mitochondrial DNA depletion syndrome 3 (hepatocerebral type), 251880 (3), Autosomal recessive
<i>DHCR24</i>	606418	Desmosterolosis, 602398 (3), Autosomal recessive
<i>DHCR7</i>	602858	Smith-Lemli-Opitz syndrome, 270400 (3), Autosomal recessive
<i>DHDDS</i>	608172	Retinitis pigmentosa 59, 613861 (3), Autosomal recessive; Developmental delay and seizures with or without movement abnormalities, 617836 (3), Autosomal dominant; ?Congenital disorder of glycosylation, type 1bb, 613861 (3), Autosomal recessive
<i>DHFR</i>	126060	Megaloblastic anemia due to dihydrofolate reductase deficiency, 613839 (3), Autosomal recessive
<i>DHH</i>	605423	46XY sex reversal 7, 233420 (3), Autosomal recessive; 46XY partial gonadal dysgenesis, with minifascicular neuropathy, 607080 (3)
<i>DHODH</i>	126064	Miller syndrome, 263750 (3), Autosomal recessive
<i>DHPS</i>	600944	Neurodevelopmental disorder with seizures and speech and walking impairment, 618480 (3), Autosomal recessive
<i>DHTKD1</i>	614984	2-aminoadipic 2-oxoadipic aciduria, 204750 (3), Autosomal recessive; ?Charcot-Marie-Tooth disease, axonal, type 2Q, 615025 (3), Autosomal dominant
<i>DHX16</i>	603405	Neuromuscular disease and ocular or auditory anomalies with or without seizures, 618733 (3), Autosomal dominant
<i>DHX30</i>	616423	Neurodevelopmental disorder with severe motor impairment and absent language, 617804 (3), Autosomal dominant
<i>DHX37</i>	617362	Neurodevelopmental disorder with brain anomalies and with or without vertebral or cardiac anomalies, 618731 (3), Autosomal recessive; 46, XY sex reversal 11, 273250 (3), Autosomal dominant
<i>DHX38</i>	605584	Retinitis pigmentosa 84, 618220 (3), Autosomal recessive
<i>DIABLO</i>	605219	Deafness, autosomal dominant 64, 614152 (3), Autosomal dominant
<i>DIAPH1</i>	602121	Seizures, cortical blindness, microcephaly syndrome, 616632 (3), Autosomal recessive; Deafness, autosomal dominant 1, with or without thrombocytopenia, 124900 (3), Autosomal dominant
<i>DIAPH2</i>	300108	?Premature ovarian failure 2A, 300511 (3), X-linked dominant
<i>DIAPH3</i>	614567	Auditory neuropathy, autosomal dominant, 1, 609129 (3), Autosomal dominant
<i>DICER1</i>	606241	GLOW syndrome, somatic mosaic, 618272 (3); Rhabdomyosarcoma, embryonal, 2, 180295 (3); Goiter, multinodular 1, with or without Sertoli-Leydig cell tumors, 138800 (3), Autosomal dominant; Pleuropulmonary blastoma, 601200 (3), Autosomal dominant

<i>DIP2B</i>	611379	Mental retardation, FRA12A type, 136630 (3), Autosomal dominant
<i>DIS3L2</i>	614184	Perlman syndrome, 267000 (3), Autosomal recessive
<i>DISC1</i>	605210	{Schizophrenia 9, susceptibility to}, 604906 (3)
<i>DKC1</i>	300126	Dyskeratosis congenita, X-linked, 305000 (3), X-linked recessive
<i>DLAT</i>	608770	Pyruvate dehydrogenase E2 deficiency, 245348 (3), Autosomal recessive
<i>DLC1</i>	604258	Colorectal cancer, somatic, 114500 (3)
<i>DLD</i>	238331	Dihydrolipoamide dehydrogenase deficiency, 246900 (3), Autosomal recessive
<i>DLG3</i>	300189	Mental retardation, X-linked 90, 300850 (3), X-linked recessive
<i>DLG4</i>	602887	Intellectual developmental disorder 62, 618793 (3), Autosomal dominant
<i>DLL1</i>	606582	Neurodevelopmental disorder with nonspecific brain abnormalities and with or without seizures, 618709 (3), Autosomal dominant
<i>DLL3</i>	602768	Spondylocostal dysostosis 1, autosomal recessive, 277300 (3), Autosomal recessive
<i>DLL4</i>	605185	Adams-Oliver syndrome 6, 616589 (3), Autosomal dominant
<i>DLST</i>	126063	Parangangliomas 7, 618475 (3), Autosomal dominant
<i>DLX3</i>	600525	Trichodontoosseous syndrome, 190320 (3), Autosomal dominant; Amelogenesis imperfecta, type IV, 104510 (3), Autosomal dominant
<i>DLX4</i>	601911	?Orofacial cleft 15, 616788 (3), Autosomal dominant
<i>DLX5</i>	600028	Split-hand/foot malformation 1, 183600 (3), Autosomal dominant; ?Split-hand/foot malformation 1 with sensorineural hearing loss, 220600 (3), Autosomal recessive
<i>DMD</i>	300377	Cardiomyopathy, dilated, 3B, 302045 (3), X-linked; Becker muscular dystrophy, 300376 (3), X-linked recessive; Duchenne muscular dystrophy, 310200 (3), X-linked recessive
<i>DMGDH</i>	605849	Dimethylglycine dehydrogenase deficiency, 605850 (3), Autosomal recessive
<i>DMP1</i>	600980	Hypophosphatemic rickets, AR, 241520 (3), Autosomal recessive
<i>DMPK</i>	605377	Myotonic dystrophy 1, 160900 (3), Autosomal dominant
<i>DMXL2</i>	612186	?Deafness, autosomal dominant 71, 617605 (3), Autosomal dominant; ?Polyendocrine-polyneuropathy syndrome, 616113 (3), Autosomal recessive; Epileptic encephalopathy, early infantile, 81, 618663 (3), Autosomal recessive
<i>DNA2</i>	601810	Progressive external ophthalmoplegia with mitochondrial DNA deletions, autosomal dominant 6, 615156 (3), Autosomal dominant; ?Seckel syndrome 8, 615807 (3), Autosomal recessive
<i>DNAAF1</i>	613190	Ciliary dyskinesia, primary, 13, 613193 (3), Autosomal recessive
<i>DNAAF2</i>	612517	Ciliary dyskinesia, primary, 10, 612518 (3)
<i>DNAAF3</i>	614566	Ciliary dyskinesia, primary, 2, 606763 (3), Autosomal recessive
<i>DNAAF4</i>	608706	{Dyslexia, susceptibility to, 1}, 127700 (3), Autosomal dominant; Ciliary dyskinesia, primary, 25, 615482 (3), Autosomal recessive

<i>DNAAF5</i>	614864	Ciliary dyskinesia, primary, 18, 614874 (3), Autosomal recessive
<i>DNAAF6</i> (<i>PIH1D3</i>)	300933	Ciliary dyskinesia, primary, 36, X-linked, 300991 (3), X-linked recessive
<i>DNAH1</i>	603332	?Ciliary dyskinesia, primary, 37, 617577 (3), Autosomal recessive; Spermatogenic failure 18, 617576 (3), Autosomal recessive
<i>DNAH11</i>	603339	Ciliary dyskinesia, primary, 7, with or without situs inversus, 611884 (3), Autosomal recessive
<i>DNAH17</i>	610063	Spermatogenic failure 39, 618643 (3), Autosomal recessive
<i>DNAH5</i>	603335	Ciliary dyskinesia, primary, 3, with or without situs inversus, 608644 (3)
<i>DNAH9</i>	603330	Ciliary dyskinesia, primary, 40, 618300 (3), Autosomal recessive
<i>DNAI1</i>	604366	Ciliary dyskinesia, primary, 1, with or without situs inversus, 244400 (3), Autosomal recessive
<i>DNAI2</i>	605483	Ciliary dyskinesia, primary, 9, with or without situs inversus, 612444 (3)
<i>DNAJB11</i>	611341	Polycystic kidney disease 6 with or without polycystic liver disease, 618061 (3), Autosomal dominant
<i>DNAJB13</i>	610263	Ciliary dyskinesia, primary, 34, 617091 (3), Autosomal recessive
<i>DNAJB2</i>	604139	Spinal muscular atrophy, distal, autosomal recessive, 5, 614881 (3), Autosomal recessive
<i>DNAJB6</i>	611332	Muscular dystrophy, limb-girdle, autosomal dominant 1, 603511 (3), Autosomal dominant
<i>DNAJC12</i>	606060	Hyperphenylalaninemia, mild, non-BH4-deficient, 617384 (3), Autosomal recessive
<i>DNAJC19</i>	608977	3-methylglutaconic aciduria, type V, 610198 (3), Autosomal recessive
<i>DNAJC21</i>	617048	Bone marrow failure syndrome 3, 617052 (3), Autosomal recessive
<i>DNAJC3</i>	601184	?Ataxia, combined cerebellar and peripheral, with hearing loss and diabetes mellitus, 616192 (3), Autosomal recessive
<i>DNAJC5</i>	611203	Ceroid lipofuscinosis, neuronal, 4, Parry type, 162350 (3), Autosomal dominant
<i>DNAJC6</i>	608375	Parkinson disease 19b, early-onset, 615528 (3), Autosomal recessive; Parkinson disease 19a, juvenile-onset, 615528 (3), Autosomal recessive
<i>DNAL1</i>	610062	Ciliary dyskinesia, primary, 16, 614017 (3), Autosomal recessive
<i>DNAL4</i>	610565	?Mirror movements 3, 616059 (3), Autosomal recessive
<i>DNASE1</i>	125505	{Systemic lupus erythematosus, susceptibility to}, 152700 (3), Autosomal dominant
<i>DNASE1L3</i>	602244	Systemic lupus erythematosus 16, 614420 (3), Autosomal recessive
<i>DNM1</i>	602377	Epileptic encephalopathy, early infantile, 31, 616346 (3), Autosomal dominant

<i>DNM1L</i>	603850	Encephalopathy, lethal, due to defective mitochondrial peroxisomal fission 1, 614388 (3), Autosomal recessive, Autosomal dominant; Optic atrophy 5, 610708 (3), Autosomal dominant
<i>DNM2</i>	602378	Lethal congenital contracture syndrome 5, 615368 (3), Autosomal recessive; Charcot-Marie-Tooth disease, axonal type 2M, 606482 (3), Autosomal dominant; Centronuclear myopathy 1, 160150 (3), Autosomal dominant; Charcot-Marie-Tooth disease, dominant intermediate B, 606482 (3), Autosomal dominant
<i>DNMBP</i>	611282	Cataract 48, 618415 (3), Autosomal recessive
<i>DNMT1</i>	126375	Cerebellar ataxia, deafness, and narcolepsy, autosomal dominant, 604121 (3), Autosomal dominant; Neuropathy, hereditary sensory, type IE, 614116 (3), Autosomal dominant
<i>DNMT3A</i>	602769	Heyn-Sproul-Jackson syndrome, 618724 (3), Autosomal dominant; Acute myeloid leukemia, somatic, 601626 (3); Tatton-Brown-Rahman syndrome, 615879 (3), Autosomal dominant
<i>DNMT3B</i>	602900	Immunodeficiency-centromeric instability-facial anomalies syndrome 1, 242860 (3), Autosomal recessive
<i>DOCK2</i>	603122	Immunodeficiency 40, 616433 (3), Autosomal recessive
<i>DOCK3</i>	603123	Neurodevelopmental disorder with impaired intellectual development, hypotonia, and ataxia, 618292 (3), Autosomal recessive
<i>DOCK6</i>	614194	Adams-Oliver syndrome 2, 614219 (3), Autosomal recessive
<i>DOCK7</i>	615730	Epileptic encephalopathy, early infantile, 23, 615859 (3), Autosomal recessive
<i>DOCK8</i>	611432	Hyper-IgE recurrent infection syndrome, autosomal recessive, 243700 (3), Autosomal recessive
<i>DOK7</i>	610285	Fetal akinesia deformation sequence 3, 618389 (3), Autosomal recessive; Myasthenic syndrome, congenital, 10, 254300 (3), Autosomal recessive
<i>DOLK</i>	610746	Congenital disorder of glycosylation, type Im, 610768 (3), Autosomal recessive
<i>DONSON</i>	611428	Microcephaly-micromelia syndrome, 251230 (3), Autosomal recessive; Microcephaly, short stature, and limb abnormalities, 617604 (3), Autosomal recessive
<i>DPAGT1</i>	191350	Congenital disorder of glycosylation, type Ij, 608093 (3), Autosomal recessive; Myasthenic syndrome, congenital, 13, with tubular aggregates, 614750 (3), Autosomal recessive
<i>DPF2</i>	601671	Coffin-Siris syndrome 7, 618027 (3), Autosomal dominant
<i>DPH1</i>	603527	Developmental delay with short stature, dysmorphic facial features, and sparse hair, 616901 (3), Autosomal recessive
<i>DPM1</i>	603503	Congenital disorder of glycosylation, type Ie, 608799 (3), Autosomal recessive
<i>DPM2</i>	603564	Congenital disorder of glycosylation, type Iu, 615042 (3), Autosomal recessive

<i>DPM3</i>	605951	Muscular dystrophy-dystroglycanopathy (limb-girdle), type C, 15, 612937 (3), Autosomal recessive; ?Muscular dystrophy-dystroglycanopathy (congenital with impaired intellectual development), type B, 15, 618992 (3)
<i>DPP6</i>	126141	Mental retardation, autosomal dominant 33, 616311 (3); {Ventricular fibrillation, paroxysmal familial, 2}, 612956 (3), Autosomal dominant
<i>DPY19L2</i>	613893	Spermatogenic failure 9, 613958 (3), Autosomal recessive
<i>DPYD</i>	612779	Dihydropyrimidine dehydrogenase deficiency, 274270 (3), Autosomal recessive; 5-fluorouracil toxicity, 274270 (3), Autosomal recessive
<i>DPYS</i>	613326	Dihydropyrimidinuria, 222748 (3), Autosomal recessive
<i>DRAM2</i>	613360	Cone-rod dystrophy 21, 616502 (3), Autosomal recessive
<i>DRC1</i>	615288	Ciliary dyskinesia, primary, 21, 615294 (3), Autosomal recessive
<i>DRD3</i>	126451	{Schizophrenia, susceptibility to}, 181500 (3), Autosomal dominant; {Essential tremor, hereditary, 1}, 190300 (3), Autosomal dominant
<i>DRD4</i>	126452	{Attention deficit-hyperactivity disorder}, 143465 (3), Autosomal dominant; Autonomic nervous system dysfunction (3)
<i>DRD5</i>	126453	{Attention deficit-hyperactivity disorder, susceptibility to}, 143465 (3), Autosomal dominant; {Blepharospasm, primary benign}, 606798 (3), Autosomal dominant
<i>DSC2</i>	125645	Arrhythmogenic right ventricular dysplasia 11 with mild palmoplantar keratoderma and woolly hair, 610476 (3), Autosomal recessive, Autosomal dominant; Arrhythmogenic right ventricular dysplasia 11, 610476 (3), Autosomal recessive, Autosomal dominant
<i>DSC3</i>	600271	?Hypotrichosis and recurrent skin vesicles, 613102 (3), Autosomal recessive
<i>DSE</i>	605942	Ehlers-Danlos syndrome, musculocontractural type 2, 615539 (3), Autosomal recessive
<i>DSG1</i>	125670	Keratosis palmoplantaris striata I, AD, 148700 (3), Autosomal dominant; Erythroderma, congenital, with palmoplantar keratoderma, hypotrichosis, and hyper IgE, 615508 (3), Autosomal recessive
<i>DSG2</i>	125671	Arrhythmogenic right ventricular dysplasia 10, 610193 (3), Autosomal dominant; Cardiomyopathy, dilated, 1BB, 612877 (3)
<i>DSG4</i>	607892	Hypotrichosis 6, 607903 (3), Autosomal recessive
<i>DSP</i>	125647	Keratosis palmoplantaris striata II, 612908 (3), Autosomal dominant; Arrhythmogenic right ventricular dysplasia 8, 607450 (3), Autosomal dominant; Epidermolysis bullosa, lethal acantholytic, 609638 (3), Autosomal recessive; Cardiomyopathy, dilated, with woolly hair and keratoderma, 605676 (3), Autosomal recessive; Skin fragility-woolly hair syndrome, 607655 (3), Autosomal recessive; Dilated cardiomyopathy with woolly hair, keratoderma, and tooth agenesis, 615821 (3), Autosomal dominant

<i>DSPP</i>	125485	Dentin dysplasia, type II, 125420 (3), Autosomal dominant; Deafness, autosomal dominant 39, with dentinogenesis, 605594 (3), Autosomal dominant; Dentinogenesis imperfecta, Shields type II, 125490 (3), Autosomal dominant; Dentinogenesis imperfecta, Shields type III, 125500 (3), Autosomal dominant
<i>DST</i>	113810	?Neuropathy, hereditary sensory and autonomic, type VI, 614653 (3), Autosomal recessive; Epidermolysis bullosa simplex, autosomal recessive 2, 615425 (3), Autosomal recessive
<i>DSTYK</i>	612666	Congenital anomalies of kidney and urinary tract 1, 610805 (3), Autosomal dominant; Spastic paraplegia 23, 270750 (3), Autosomal recessive
<i>DTNA</i>	601239	Left ventricular noncompaction 1, with or without congenital heart defects, 604169 (3), Autosomal dominant
<i>DTNBP1</i>	607145	Hermansky-Pudlak syndrome 7, 614076 (3), Autosomal recessive
<i>DUOX2</i>	606759	Thyroid dyshormonogenesis 6, 607200 (3), Autosomal recessive
<i>DUOXA2</i>	612772	Thyroid dyshormonogenesis 5, 274900 (3), Autosomal recessive
<i>DUSP6</i>	602748	Hypogonadotropic hypogonadism 19 with or without anosmia, 615269 (3), Autosomal dominant
<i>DVL1</i>	601365	Robinow syndrome, autosomal dominant 2, 616331 (3), Autosomal dominant
<i>DVL3</i>	601368	Robinow syndrome, autosomal dominant 3, 616894 (3), Autosomal dominant
<i>DYM</i>	607461	Smith-McCort dysplasia, 607326 (3), Autosomal recessive; Dyggve-Melchior-Clausen disease, 223800 (3), Autosomal recessive
<i>DYNC1H1</i>	600112	Mental retardation, autosomal dominant 13, 614563 (3), Autosomal dominant; Spinal muscular atrophy, lower extremity-predominant 1, AD, 158600 (3), Autosomal dominant; Charcot-Marie-Tooth disease, axonal, type 20, 614228 (3), Autosomal dominant
<i>DYNC1I2</i>	603331	Neurodevelopmental disorder with microcephaly and structural brain anomalies, 618492 (3), Autosomal recessive
<i>DYNC2H1</i>	603297	Short-rib thoracic dysplasia 3 with or without polydactyly, 613091 (3), Autosomal recessive, Digenic recessive
<i>DYNC2I1 (WDR60)</i>	615462	Short-rib thoracic dysplasia 8 with or without polydactyly, 615503 (3), Autosomal recessive
<i>DYNC2I2 (WDR34)</i>	613363	Short-rib thoracic dysplasia 11 with or without polydactyly, 615633 (3), Autosomal recessive
<i>DYNC2LI1</i>	617083	Short-rib thoracic dysplasia 15 with polydactyly, 617088 (3), Autosomal recessive
<i>DYNLT2B (TCTEX1D2)</i>	617353	Short-rib thoracic dysplasia 17 with or without polydactyly, 617405 (3), Autosomal recessive
<i>DYRK1A</i>	600855	Mental retardation, autosomal dominant 7, 614104 (3), Autosomal dominant

<i>DYRK1B</i>	604556	Abdominal obesity-metabolic syndrome 3, 615812 (3), Autosomal dominant
<i>DYSF</i>	603009	Miyoshi muscular dystrophy 1, 254130 (3), Autosomal recessive; Muscular dystrophy, limb-girdle, autosomal recessive 2, 253601 (3), Autosomal recessive; Myopathy, distal, with anterior tibial onset, 606768 (3), Autosomal recessive
<i>DZIP1L</i>	617570	Polycystic kidney disease 5, 617610 (3), Autosomal recessive
<i>EARS2</i>	612799	Combined oxidative phosphorylation deficiency 12, 614924 (3), Autosomal recessive
<i>EBF3</i>	607407	Hypotonia, ataxia, and delayed development syndrome, 617330 (3), Autosomal dominant
<i>EBP</i>	300205	Chondrodysplasia punctata, X-linked dominant, 302960 (3), X-linked dominant; MEND syndrome, 300960 (3), X-linked recessive
<i>ECE1</i>	600423	{Hypertension, essential, susceptibility to}, 145500 (3), Multifactorial; ?Hirschsprung disease, cardiac defects, and autonomic dysfunction, 613870 (3), Autosomal dominant
<i>ECEL1</i>	605896	Arthrogryposis, distal, type 5D, 615065 (3), Autosomal recessive
<i>ECHS1</i>	602292	Mitochondrial short-chain enoyl-CoA hydratase 1 deficiency, 616277 (3), Autosomal recessive
<i>ECM1</i>	602201	Urbach-Wiethe disease, 247100 (3), Autosomal recessive
<i>EDA</i>	300451	Tooth agenesis, selective, X-linked 1, 313500 (3), X-linked dominant; Ectodermal dysplasia 1, hypohidrotic, X-linked, 305100 (3), X-linked recessive
<i>EDAR</i>	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)
<i>EDARADD</i>	606603	Ectodermal dysplasia 11B, hypohidrotic/hair/tooth type, autosomal recessive, 614941 (3), Autosomal recessive; Ectodermal dysplasia 11A, hypohidrotic/hair/tooth type, autosomal dominant, 614940 (3), Autosomal dominant
<i>EDC3</i>	609842	?Mental retardation, autosomal recessive 50, 616460 (3), Autosomal recessive
<i>EDN1</i>	131240	Auriculocondylar syndrome 3, 615706 (3), Autosomal recessive; {High density lipoprotein cholesterol level QTL 7} (3); Question mark ears, isolated, 612798 (3), Autosomal dominant
<i>EDN3</i>	131242	Waardenburg syndrome, type 4B, 613265 (3), Autosomal recessive, Autosomal dominant; Central hypoventilation syndrome, congenital, 209880 (3), Autosomal dominant; {Hirschsprung disease, susceptibility to, 4}, 613712 (3), Autosomal dominant
<i>EDNRA</i>	131243	Mandibulofacial dysostosis with alopecia, 616367 (3), Autosomal dominant; {Migraine, resistance to}, 157300 (3), Autosomal dominant

<i>EDNRB</i>	131244	{Hirschsprung disease, susceptibility to, 2}, 600155 (3), Autosomal dominant; Waardenburg syndrome, type 4A, 277580 (3), Autosomal recessive, Autosomal dominant; ABCD syndrome, 600501 (3), Autosomal recessive
<i>EED</i>	605984	Cohen-Gibson syndrome, 617561 (3), Autosomal dominant
<i>EEF1A2</i>	602959	Epileptic encephalopathy, early infantile, 33, 616409 (3), Autosomal dominant; Mental retardation, autosomal dominant 38, 616393 (3), Autosomal dominant
<i>EEF1AKNMT</i>	617987	?{Deafness, autosomal recessive 26, modifier of}, 605429 (3), Autosomal dominant
<i>EEF2</i>	130610	?Spinocerebellar ataxia 26, 609306 (3), Autosomal dominant
<i>EFEMP1</i>	601548	Doyne honeycomb degeneration of retina, 126600 (3), Autosomal dominant
<i>EFEMP2</i>	604633	Cutis laxa, autosomal recessive, type IB, 614437 (3), Autosomal recessive
<i>EFHC1</i>	608815	{Myoclonic epilepsy, juvenile, susceptibility to, 1}, 254770 (3), Autosomal dominant; {Epilepsy, juvenile absence, susceptibility to, 1}, 607631 (3), Autosomal dominant
<i>EFL1</i>	617538	Shwachman-Diamond syndrome 2, 617941 (3), Autosomal recessive
<i>EFNB1</i>	300035	Craniofrontonasal dysplasia, 304110 (3), X-linked dominant
<i>EFTUD2</i>	603892	Mandibulofacial dysostosis, Guion-Almeida type, 610536 (3), Autosomal dominant
<i>EGF</i>	131530	Hypomagnesemia 4, renal, 611718 (3)
<i>EGFR</i>	131550	?Inflammatory skin and bowel disease, neonatal, 2, 616069 (3), Autosomal recessive; Non-small cell lung cancer, response to tyrosine kinase inhibitor in, 211980 (3), Autosomal dominant, Somatic mutation; Adenocarcinoma of lung, response to tyrosine kinase inhibitor in, 211980 (3), Autosomal dominant, Somatic mutation; {Non-small cell lung cancer, susceptibility to}, 211980 (3), Autosomal dominant, Somatic mutation
<i>EGLN1</i>	606425	[Hemoglobin, high altitude adaptation], 609070 (3), Autosomal dominant; Erythrocytosis, familial, 3, 609820 (3), Autosomal dominant
<i>EGR2</i>	129010	Dejerine-Sottas disease, 145900 (3), Autosomal recessive, Autosomal dominant; Hypomyelinating neuropathy, congenital, 1, 605253 (3), Autosomal recessive, Autosomal dominant; Charcot-Marie-Tooth disease, type 1D, 607678 (3), Autosomal dominant
<i>EHBP1</i>	609922	{Prostate cancer, hereditary, 12}, 611868 (3)
<i>EHHADH</i>	607037	?Fanconi renal tubular syndrome 3, 615605 (3), Autosomal dominant
<i>EHMT1</i>	607001	Kleefstra syndrome 1, 610253 (3), Autosomal dominant
<i>EIF2AK1</i>	613635	?Leukoencephalopathy, motor delay, spasticity, and dysarthria syndrome, 618878 (3)

<i>EIF2AK2</i>	176871	Leukoencephalopathy, developmental delay, and episodic neurologic regression syndrome, 618877 (3), Autosomal dominant
<i>EIF2AK3</i>	604032	Wolcott-Rallison syndrome, 226980 (3), Autosomal recessive
<i>EIF2AK4</i>	609280	Pulmonary venoocclusive disease 2, 234810 (3), Autosomal recessive
<i>EIF2B1</i>	606686	Leukoencephalopathy with vanishing white matter, 603896 (3), Autosomal recessive
<i>EIF2B2</i>	606454	Ovarioleukodystrophy, 603896 (3), Autosomal recessive; Leukoencephalopathy with vanishing white matter, 603896 (3), Autosomal recessive
<i>EIF2B3</i>	606273	Leukoencephalopathy with vanishing white matter, 603896 (3), Autosomal recessive
<i>EIF2B4</i>	606687	Leukoencephalopathy with vanishing white matter, 603896 (3), Autosomal recessive; Ovarioleukodystrophy, 603896 (3), Autosomal recessive
<i>EIF2B5</i>	603945	Leukoencephalopathy with vanishing white matter, 603896 (3), Autosomal recessive; Ovarioleukodystrophy, 603896 (3), Autosomal recessive
<i>EIF2S3</i>	300161	MEHMO syndrome, 300148 (3), X-linked recessive
<i>EIF3F</i>	603914	Mental retardation, autosomal recessive 67, 618295 (3), Autosomal recessive
<i>EIF4A3</i>	608546	Robin sequence with cleft mandible and limb anomalies, 268305 (3), Autosomal recessive
<i>EIF4E</i>	133440	{Autism, susceptibility to, 19}, 615091 (3)
<i>EIF4G1</i>	600495	{Parkinson disease 18}, 614251 (3), Autosomal dominant
<i>ELAC2</i>	605367	Combined oxidative phosphorylation deficiency 17, 615440 (3), Autosomal recessive; {Prostate cancer, hereditary, 2, susceptibility to}, 614731 (3)
<i>ELANE</i>	130130	Neutropenia, severe congenital 1, autosomal dominant, 202700 (3), Autosomal dominant; Neutropenia, cyclic, 162800 (3), Autosomal dominant
<i>ELMO2</i>	606421	Vascular malformation, primary intraosseous, 606893 (3), Autosomal recessive
<i>ELMOD3</i>	615427	?Deafness, autosomal recessive 88, 615429 (3), Autosomal recessive
<i>ELN</i>	130160	Cutis laxa, autosomal dominant, 123700 (3), Autosomal dominant; Supravalvar aortic stenosis, 185500 (3), Autosomal dominant
<i>ELOVL1</i>	611813	Ichthyotic keratoderma, spasticity, hypomyelination, and dysmorphic facies, 618527 (3), Autosomal dominant
<i>ELOVL4</i>	605512	Spinocerebellar ataxia 34, 133190 (3), Autosomal dominant; Stargardt disease 3, 600110 (3), Autosomal dominant; Ichthyosis, spastic quadriplegia, and mental retardation, 614457 (3), Autosomal recessive
<i>ELOVL5</i>	611805	Spinocerebellar ataxia 38, 615957 (3), Autosomal dominant

<i>ELP1</i>	603722	Dysautonomia, familial, 223900 (3), Autosomal recessive
<i>ELP2</i>	616054	Mental retardation, autosomal recessive 58, 617270 (3), Autosomal recessive
<i>ELP4</i>	606985	?Aniridia 2, 617141 (3), Autosomal dominant
<i>EMC1</i>	616846	Cerebellar atrophy, visual impairment, and psychomotor retardation, 616875 (3), Autosomal recessive
<i>EMD</i>	300384	Emery-Dreifuss muscular dystrophy 1, X-linked, 310300 (3), X-linked recessive
<i>EMG1</i>	611531	Bowen-Conradi syndrome, 211180 (3), Autosomal recessive
<i>EML1</i>	602033	Band heterotopia, 600348 (3), Autosomal recessive
<i>EMP2</i>	602334	Nephrotic syndrome, type 10, 615861 (3), Autosomal recessive
<i>EMX2</i>	600035	Schizencephaly, 269160 (3)
<i>ENAM</i>	606585	Amelogenesis imperfecta, type IC, 204650 (3), Autosomal recessive; Amelogenesis imperfecta, type IB, 104500 (3), Autosomal dominant
<i>ENG</i>	131195	Telangiectasia, hereditary hemorrhagic, type 1, 187300 (3), Autosomal dominant
<i>ENO1</i>	172430	Enolase deficiency (1)
<i>ENO3</i>	131370	?Glycogen storage disease XIII, 612932 (3), Autosomal recessive
<i>ENPP1</i>	173335	Hypophosphatemic rickets, autosomal recessive, 2, 613312 (3), Autosomal recessive; Cole disease, 615522 (3), Autosomal dominant; {Obesity, susceptibility to}, 601665 (3), Autosomal recessive, Autosomal dominant, Multifactorial; Arterial calcification, generalized, of infancy, 1, 208000 (3), Autosomal recessive; {Diabetes mellitus, non-insulin-dependent, susceptibility to}, 125853 (3), Autosomal dominant
<i>ENTPD1</i>	601752	Spastic paraplegia 64, autosomal recessive, 615683 (3), Autosomal recessive
<i>EOGT</i>	614789	Adams-Oliver syndrome 4, 615297 (3), Autosomal recessive
<i>EP300</i>	602700	Rubinstein-Taybi syndrome 2, 613684 (3), Autosomal dominant; Menke-Hennekam syndrome 2, 618333 (3), Autosomal dominant; Colorectal cancer, somatic, 114500 (3)
<i>EPAS1</i>	603349	Erythrocytosis, familial, 4, 611783 (3)
<i>EPB41</i>	130500	Elliptocytosis-1, 611804 (3), Autosomal recessive, Autosomal dominant
<i>EPB41L1</i>	602879	?Mental retardation, autosomal dominant 11, 614257 (3), Autosomal dominant
<i>EPB42</i>	177070	Spherocytosis, type 5, 612690 (3)
<i>EPCAM</i>	185535	Colorectal cancer, hereditary nonpolyposis, type 8, 613244 (3); Diarrhea 5, with tufting enteropathy, congenital, 613217 (3), Autosomal recessive
<i>EPG5</i>	615068	Vici syndrome, 242840 (3), Autosomal recessive
<i>EPHA2</i>	176946	Cataract 6, multiple types, 116600 (3), Autosomal dominant
<i>EPHB2</i>	600997	{Prostate cancer/brain cancer susceptibility, somatic}, 603688 (3); ?Bleeding disorder, platelet-type, 22, 618462 (3), Autosomal recessive

<i>EPHB4</i>	600011	Capillary malformation-arteriovenous malformation 2, 618196 (3), Autosomal dominant; Lymphatic malformation 7, 617300 (3), Autosomal dominant
<i>EPHX1</i>	132810	?Hypercholanemia, familial, 607748 (3), Autosomal recessive
<i>EPHX2</i>	132811	{Hypercholesterolemia, familial, due to LDLR defect, modifier of}, 143890 (3), Autosomal dominant
<i>EPM2A</i>	607566	Epilepsy, progressive myoclonic 2A (Lafora), 254780 (3), Autosomal recessive
<i>EPO</i>	133170	Erythrocytosis, familial, 5, 617907 (3), Autosomal dominant; {Microvascular complications of diabetes 2}, 612623 (3); ?Diamond-Blackfan anemia-like, 617911 (3), Autosomal recessive
<i>EPOR</i>	133171	[Erythrocytosis, familial, 1], 133100 (3), Autosomal dominant
<i>EPRS1 (EPRS)</i>	138295	Leukodystrophy, hypomyelinating, 15, 617951 (3), Autosomal recessive
<i>EPS8</i>	600206	?Deafness, autosomal recessive 102, 615974 (3), Autosomal recessive
<i>EPS8L2</i>	614988	Deafness autosomal recessive 106, 617637 (3), Autosomal recessive
<i>EPS8L3</i>	614989	?Hypotrichosis 5, 612841 (3), Autosomal dominant
<i>EPX</i>	131399	[Eosinophil peroxidase deficiency], 261500 (3), Autosomal recessive
<i>ERAL1</i>	607435	Perrault syndrome 6, 617565 (3), Autosomal recessive
<i>ERBB2</i>	164870	Glioblastoma, somatic, 137800 (3); Adenocarcinoma of lung, somatic, 211980 (3); Gastric cancer, somatic, 613659 (3); Ovarian cancer, somatic (3)
<i>ERBB3</i>	190151	{?Erythroleukemia, familial, susceptibility to}, 133180 (3), Autosomal dominant; ?Lethal congenital contractural syndrome 2, 607598 (3), Autosomal recessive
<i>ERBB4</i>	600543	Amyotrophic lateral sclerosis 19, 615515 (3), Autosomal dominant
<i>ERCC1</i>	126380	Cerebrooculofacioskeletal syndrome 4, 610758 (3), Autosomal recessive
<i>ERCC2</i>	126340	Trichothiodystrophy 1, photosensitive, 601675 (3), Autosomal recessive; ?Cerebrooculofacioskeletal syndrome 2, 610756 (3), Autosomal recessive; Xeroderma pigmentosum, group D, 278730 (3), Autosomal recessive
<i>ERCC3</i>	133510	Xeroderma pigmentosum, group B, 610651 (3), Autosomal recessive; Trichothiodystrophy 2, photosensitive, 616390 (3), Autosomal recessive
<i>ERCC4</i>	133520	Xeroderma pigmentosum, type F/Cockayne syndrome, 278760 (3), Autosomal recessive; Fanconi anemia, complementation group Q, 615272 (3), Autosomal recessive; XFE progeroid syndrome, 610965 (3), Autosomal recessive; Xeroderma pigmentosum, group F, 278760 (3), Autosomal recessive

<i>ERCC5</i>	133530	Xeroderma pigmentosum, group G/Cockayne syndrome, 278780 (3), Autosomal recessive; Xeroderma pigmentosum, group G, 278780 (3), Autosomal recessive; Cerebrooculofacioskeletal syndrome 3, 616570 (3), Autosomal recessive
<i>ERCC6</i>	609413	{Macular degeneration, age-related, susceptibility to, 5}, 613761 (3); {Lung cancer, susceptibility to}, 211980 (3), Autosomal dominant, Somatic mutation; Cerebrooculofacioskeletal syndrome 1, 214150 (3), Autosomal recessive; Cockayne syndrome, type B, 133540 (3), Autosomal recessive; Premature ovarian failure 11, 616946 (3), Autosomal dominant; UV-sensitive syndrome 1, 600630 (3), Autosomal recessive; De Sanctis-Cacchione syndrome, 278800 (3), Autosomal recessive
<i>ERCC6L2</i>	615667	Bone marrow failure syndrome 2, 615715 (3), Autosomal recessive
<i>ERCC8</i>	609412	Cockayne syndrome, type A, 216400 (3), Autosomal recessive; UV-sensitive syndrome 2, 614621 (3), Autosomal recessive
<i>ERF</i>	611888	Craniosynostosis 4, 600775 (3), Autosomal dominant; Chitayat syndrome, 617180 (3), Autosomal dominant
<i>ERGIC1</i>	617946	?Arthrogryposis multiplex congenita 2, neurogenic type, 208100 (3), Autosomal recessive
<i>ERLIN1</i>	611604	Spastic paraplegia 62, 615681 (3), Autosomal recessive
<i>ERLIN2</i>	611605	Spastic paraplegia 18, autosomal recessive, 611225 (3), Autosomal recessive
<i>ERMAP</i>	609017	[Blood group, Scianna system], 111750 (3); [Blood group, Radin], 111620 (3)
<i>ERMARD</i>	615532	?Periventricular nodular heterotopia 6, 615544 (3), Autosomal dominant
<i>ESCO2</i>	609353	Roberts syndrome, 268300 (3), Autosomal recessive; SC phocomelia syndrome, 269000 (3), Autosomal recessive
<i>ESPN</i>	606351	?Usher syndrome, type 1M, 618632 (3), Autosomal recessive; Deafness, autosomal recessive 36, 609006 (3), Autosomal recessive; Deafness, neurosensory, without vestibular involvement, autosomal dominant, 609006 (3), Autosomal recessive
<i>ESR1</i>	133430	{Myocardial infarction, susceptibility to}, 608446 (3); Estrogen resistance, 615363 (3), Autosomal recessive; Breast cancer, somatic, 114480 (3); {Migraine, susceptibility to}, 157300 (3), Autosomal dominant
<i>ESR2</i>	601663	?Ovarian dysgenesis 8, 618187 (3), Autosomal dominant
<i>ESRP1</i>	612959	?Deafness, autosomal recessive 109, 618013 (3), Autosomal recessive
<i>ESRRB</i>	602167	Deafness, autosomal recessive 35, 608565 (3), Autosomal recessive
<i>ETFA</i>	608053	Glutaric acidemia IIA, 231680 (3), Autosomal recessive
<i>ETFB</i>	130410	Glutaric acidemia IIB, 231680 (3), Autosomal recessive
<i>ETFDH</i>	231675	Glutaric acidemia IIC, 231680 (3), Autosomal recessive

<i>ETHE1</i>	608451	Ethylmalonic encephalopathy, 602473 (3), Autosomal recessive
<i>ETV6</i>	600618	Leukemia, acute myeloid, somatic, 601626 (3); Thrombocytopenia 5, 616216 (3), Autosomal dominant
<i>EVC</i>	604831	Ellis-van Creveld syndrome, 225500 (3), Autosomal recessive; ?Weyers acrofacial dysostosis, 193530 (3), Autosomal dominant
<i>EVC2</i>	607261	Weyers acrofacial dysostosis, 193530 (3), Autosomal dominant; Ellis-van Creveld syndrome, 225500 (3), Autosomal recessive
<i>EWSR1</i>	133450	Neuroepithelioma, 612219 (3); Ewing sarcoma, 612219 (3)
<i>EXOC6B</i>	607880	Spondyloepimetaphyseal dysplasia with joint laxity, type 3, 618395 (3), Autosomal recessive
<i>EXOSC2</i>	602238	Short stature, hearing loss, retinitis pigmentosa, and distinctive facies, 617763 (3), Autosomal recessive
<i>EXOSC3</i>	606489	Pontocerebellar hypoplasia, type 1B, 614678 (3), Autosomal recessive
<i>EXOSC8</i>	606019	Pontocerebellar hypoplasia, type 1C, 616081 (3), Autosomal recessive
<i>EXOSC9</i>	606180	Pontocerebellar hypoplasia, type 1D, 618065 (3), Autosomal recessive
<i>EXPH5</i>	612878	Epidermolysis bullosa, nonspecific, autosomal recessive, 615028 (3), Autosomal recessive
<i>EXT1</i>	608177	Exostoses, multiple, type 1, 133700 (3), Autosomal dominant; Chondrosarcoma, 215300 (3), Somatic mutation
<i>EXT2</i>	608210	Seizures, scoliosis, and macrocephaly syndrome, 616682 (3), Autosomal recessive; Exostoses, multiple, type 2, 133701 (3), Autosomal dominant
<i>EXTL3</i>	605744	Immunoskeletal dysplasia with neurodevelopmental abnormalities, 617425 (3), Autosomal recessive
<i>EYA1</i>	601653	?Otofaciocervical syndrome, 166780 (3), Autosomal dominant; Anterior segment anomalies with or without cataract, 602588 (3), Autosomal dominant; Branchiootorenal syndrome 1, with or without cataracts, 113650 (3), Autosomal dominant; Branchiootic syndrome 1, 602588 (3), Autosomal dominant
<i>EYA4</i>	603550	Deafness, autosomal dominant 10, 601316 (3), Autosomal dominant; ?Cardiomyopathy, dilated, 1J, 605362 (3), Autosomal dominant
<i>EYS</i>	612424	Retinitis pigmentosa 25, 602772 (3), Autosomal recessive
<i>EZH2</i>	601573	Weaver syndrome, 277590 (3), Autosomal dominant
<i>F10</i>	613872	Factor X deficiency, 227600 (3), Autosomal recessive
<i>F11</i>	264900	Factor XI deficiency, autosomal dominant, 612416 (3); Factor XI deficiency, autosomal recessive, 612416 (3)
<i>F12</i>	610619	Factor XII deficiency, 234000 (3), Autosomal recessive; Angioedema, hereditary, type III, 610618 (3), Autosomal dominant

<i>F13A1</i>	134570	{Myocardial infarction, protection against}, 608446 (3); Factor XIII A deficiency, 613225 (3), Autosomal recessive; {Venous thrombosis, protection against}, 188050 (3), Autosomal dominant
<i>F13B</i>	134580	Factor XIII B deficiency, 613235 (3), Autosomal recessive
<i>F2</i>	176930	{Pregnancy loss, recurrent, susceptibility to, 2}, 614390 (3), Autosomal dominant; Hypoprothrombinemia, 613679 (3), Autosomal recessive; Dysprothrombinemia, 613679 (3), Autosomal recessive; Thrombophilia due to thrombin defect, 188050 (3), Autosomal dominant; {Stroke, ischemic, susceptibility to}, 601367 (3), Multifactorial
<i>F5</i>	612309	{Pregnancy loss, recurrent, susceptibility to, 1}, 614389 (3), Autosomal dominant; Thrombophilia due to activated protein C resistance, 188055 (3), Autosomal dominant; {Thrombophilia, susceptibility to, due to factor V Leiden}, 188055 (3), Autosomal dominant; Factor V deficiency, 227400 (3), Autosomal recessive; {Budd-Chiari syndrome}, 600880 (3), Autosomal recessive; {Stroke, ischemic, susceptibility to}, 601367 (3), Multifactorial
<i>F7</i>	613878	{Myocardial infarction, decreased susceptibility to}, 608446 (3); Factor VII deficiency, 227500 (3), Autosomal recessive
<i>F8</i>	300841	Hemophilia A, 306700 (3), X-linked recessive
<i>F9</i>	300746	Thrombophilia, X-linked, due to factor IX defect, 300807 (3), X-linked recessive; {Deep venous thrombosis, protection against}, 300807 (3), X-linked recessive; Hemophilia B, 306900 (3), X-linked recessive; {Warfarin sensitivity}, 301052 (3), X-linked
<i>FA2H</i>	611026	Spastic paraplegia 35, autosomal recessive, 612319 (3), Autosomal recessive
<i>FAAH</i>	602935	{Drug addiction, susceptibility to}, 606581 (3)
<i>FADD</i>	602457	Infections, recurrent, with encephalopathy, hepatic dysfunction, and cardiovascular malformations, 613759 (3), Autosomal recessive
<i>FAH</i>	613871	Tyrosinemia, type I, 276700 (3), Autosomal recessive
<i>FAM111A</i>	615292	Gracile bone dysplasia, 602361 (3), Autosomal dominant; Kenny-Caffey syndrome, type 2, 127000 (3), Autosomal dominant
<i>FAM111B</i>	615584	Poikiloderma, hereditary fibrosing, with tendon contractures, myopathy, and pulmonary fibrosis, 615704 (3), Autosomal dominant
<i>FAM126A</i>	610531	Leukodystrophy, hypomyelinating, 5, 610532 (3), Autosomal recessive
<i>FAM149B1</i>	618413	Joubert syndrome 36, 618763 (3), Autosomal recessive
<i>FAM161A</i>	613596	Retinitis pigmentosa 28, 606068 (3)
<i>FAM20A</i>	611062	Amelogenesis imperfecta, type IG (enamel-renal syndrome), 204690 (3), Autosomal recessive
<i>FAM20C</i>	611061	Raine syndrome, 259775 (3), Autosomal recessive
<i>FAM50A</i>	300453	Intellectual developmental disorder, X-linked, syndromic, Armfield type, 300261 (3), X-linked recessive

<i>FAM83H</i>	611927	Amelogenesis imperfecta, type IIIA, 130900 (3), Autosomal dominant
<i>FAN1</i>	613534	Interstitial nephritis, karyomegalic, 614817 (3), Autosomal recessive
<i>FANCA</i>	607139	Fanconi anemia, complementation group A, 227650 (3), Autosomal recessive
<i>FANCB</i>	300515	Fanconi anemia, complementation group B, 300514 (3), X-linked recessive
<i>FANCC</i>	613899	Fanconi anemia, complementation group C, 227645 (3), Autosomal recessive
<i>FANCD2</i>	613984	Fanconi anemia, complementation group D2, 227646 (3), Autosomal recessive
<i>FANCE</i>	613976	Fanconi anemia, complementation group E, 600901 (3), Autosomal recessive
<i>FANCF</i>	613897	Fanconi anemia, complementation group F, 603467 (3)
<i>FANCG</i>	602956	Fanconi anemia, complementation group G, 614082 (3)
<i>FANCI</i>	611360	Fanconi anemia, complementation group I, 609053 (3), Autosomal recessive
<i>FANCL</i>	608111	Fanconi anemia, complementation group L, 614083 (3), Autosomal recessive
<i>FANCM</i>	609644	Spermatogenic failure 28, 618086 (3), Autosomal recessive; ?Premature ovarian failure 15, 618096 (3), Autosomal recessive
<i>FAR1</i>	616107	Peroxisomal fatty acyl-CoA reductase 1 disorder, 616154 (3), Autosomal recessive
<i>FARS2</i>	611592	Spastic paraplegia 77, autosomal recessive, 617046 (3), Autosomal recessive; Combined oxidative phosphorylation deficiency 14, 614946 (3), Autosomal recessive
<i>FARSB</i>	609690	Rajab interstitial lung disease with brain calcifications, 613658 (3), Autosomal recessive
<i>FAS</i>	134637	Autoimmune lymphoproliferative syndrome, type IA, 601859 (3), Autosomal dominant; Squamous cell carcinoma, burn scar-related, somatic (3); {Autoimmune lymphoproliferative syndrome}, 601859 (3), Autosomal dominant
<i>FASLG</i>	134638	Autoimmune lymphoproliferative syndrome, type IB, 601859 (3), Autosomal dominant; {Lung cancer, susceptibility to}, 211980 (3), Autosomal dominant, Somatic mutation
<i>FASTKD2</i>	612322	Combined oxidative phosphorylation deficiency 44, 618855 (3), Autosomal recessive
<i>FAT2</i>	604269	Spinocerebellar ataxia 45, 617769 (3), Autosomal dominant
<i>FAT4</i>	612411	Van Maldergem syndrome 2, 615546 (3), Autosomal recessive; Hennekam lymphangiectasia-lymphedema syndrome 2, 616006 (3), Autosomal recessive
<i>FBLN1</i>	135820	Synpolydactyly, 3/3'4, associated with metacarpal and metatarsal synostoses, 608180 (4), Autosomal dominant

<i>FBLN5</i>	604580	Macular degeneration, age-related, 3, 608895 (3), Autosomal dominant; ?Cutis laxa, autosomal dominant 2, 614434 (3), Autosomal dominant; Neuropathy, hereditary, with or without age-related macular degeneration, 608895 (3), Autosomal dominant; Cutis laxa, autosomal recessive, type IA, 219100 (3), Autosomal recessive
<i>FBN1</i>	134797	Marfan lipodystrophy syndrome, 616914 (3), Autosomal dominant; Marfan syndrome, 154700 (3), Autosomal dominant; MASS syndrome, 604308 (3), Autosomal dominant; Ectopia lentis, familial, 129600 (3), Autosomal dominant; Acromicric dysplasia, 102370 (3), Autosomal dominant; Weill-Marchesani syndrome 2, dominant, 608328 (3), Autosomal dominant; Geleophysic dysplasia 2, 614185 (3), Autosomal dominant; Stiff skin syndrome, 184900 (3), Autosomal dominant
<i>FBN2</i>	612570	Contractural arachnodactyly, congenital, 121050 (3), Autosomal dominant; Macular degeneration, early-onset, 616118 (3), Autosomal dominant
<i>FBP1</i>	611570	Fructose-1,6-bisphosphatase deficiency, 229700 (3), Autosomal recessive
<i>FBXL3</i>	605653	Intellectual developmental disorder with short stature, facial anomalies, and speech defects, 606220 (3), Autosomal recessive
<i>FBXL4</i>	605654	Mitochondrial DNA depletion syndrome 13 (encephalomyopathic type), 615471 (3), Autosomal recessive
<i>FBXO11</i>	607871	Intellectual developmental disorder with dysmorphic facies and behavioral abnormalities, 618089 (3), Autosomal dominant
<i>FBXO31</i>	609102	?Mental retardation, autosomal recessive 45, 615979 (3), Autosomal recessive
<i>FBXO38</i>	608533	Neuronopathy, distal hereditary motor, type IID, 615575 (3), Autosomal dominant
<i>FBXO7</i>	605648	Parkinson disease 15, autosomal recessive, 260300 (3), Autosomal recessive
<i>FBXW11</i>	605651	Neurodevelopmental, jaw, eye, and digital syndrome, 618914 (3), Autosomal dominant
<i>FBXW4</i>	608071/246560	-/Split-hand/foot malformation 3, gene duplication syndrome, 246560 (4), Autosomal dominant
<i>FCGR1A</i>	146760	[IgG receptor I, phagocytic, familial deficiency of] (3)
<i>FCGR2A</i>	146790	{Pseudomonas aeruginosa, susceptibility to chronic infection by, in cystic fibrosis}, 219700 (3), Autosomal recessive; {Lupus nephritis, susceptibility to}, 152700 (3), Autosomal dominant; {Malaria, severe, susceptibility to}, 611162 (3)
<i>FCGR2B</i>	604590	{Malaria, resistance to}, 611162 (3); {Systemic lupus erythematosus, susceptibility to}, 152700 (3), Autosomal dominant
<i>FCGR3A</i>	146740	Immunodeficiency 20, 615707 (3), Autosomal recessive
<i>FCGR3B</i>	610665	Neutropenia, alloimmune neonatal (3)

<i>FCN3</i>	604973	Immunodeficiency due to ficolin 3 deficiency, 613860 (3), Autosomal recessive
<i>FCSK (FUK)</i>	608675	Congenital disorder of glycosylation with defective fucosylation 2, 618324 (3), Autosomal recessive
<i>FDFT1</i>	184420	Squalene synthase deficiency, 618156 (3), Autosomal recessive
<i>FDPS</i>	134629	Porokeratosis 9, multiple types, 616631 (3), Autosomal dominant
<i>FDX2</i>	614585	Mitochondrial myopathy, episodic, with optic atrophy and reversible leukoencephalopathy, 251900 (3), Autosomal recessive
<i>FDXR</i>	103270	Auditory neuropathy and optic atrophy, 617717 (3), Autosomal recessive
<i>FECH</i>	612386	Protoporphyrria, erythropoietic, 1, 177000 (3), Autosomal recessive
<i>FERMT1</i>	607900	Kindler syndrome, 173650 (3), Autosomal recessive
<i>FERMT3</i>	607901	Leukocyte adhesion deficiency, type III, 612840 (3), Autosomal recessive
<i>FEZF1</i>	613301	Hypogonadotropic hypogonadism 22, with or without anosmia, 616030 (3), Autosomal recessive
<i>FFAR4</i>	609044	{Obesity, susceptibility to}, 607514 (3)
<i>FGA</i>	134820	Dysfibrinogenemia, congenital, 616004 (3); Amyloidosis, familial visceral, 105200 (3), Autosomal dominant; Hypodysfibrinogenemia, congenital, 616004 (3); Afibrinogenemia, congenital, 202400 (3), Autosomal recessive
<i>FGB</i>	134830	Dysfibrinogenemia, congenital, 616004 (3); Afibrinogenemia, congenital, 202400 (3), Autosomal recessive; Hypofibrinogenemia, congenital, 202400 (3), Autosomal recessive
<i>FGD1</i>	300546	Mental retardation, X-linked syndromic 16, 305400 (3), X-linked recessive; Aarskog-Scott syndrome, 305400 (3), X-linked recessive
<i>FGD4</i>	611104	Charcot-Marie-Tooth disease, type 4H, 609311 (3), Autosomal recessive
<i>FGF10</i>	602115	Aplasia of lacrimal and salivary glands, 180920 (3), Autosomal dominant; LADD syndrome, 149730 (3), Autosomal dominant
<i>FGF12</i>	601513	Epileptic encephalopathy, early infantile, 47, 617166 (3), Autosomal dominant
<i>FGF14</i>	601515	Spinocerebellar ataxia 27, 609307 (3), Autosomal dominant
<i>FGF16</i>	300827	Metacarpal 4-5 fusion, 309630 (3), X-linked recessive
<i>FGF17</i>	603725	Hypogonadotropic hypogonadism 20 with or without anosmia, 615270 (3), Autosomal dominant
<i>FGF20</i>	605558	?Renal hypodysplasia/aplasia 2, 615721 (3), Autosomal recessive
<i>FGF23</i>	605380	Tumoral calcinosis, hyperphosphatemic, familial, 2, 617993 (3), Autosomal recessive; Hypophosphatemic rickets, autosomal dominant, 193100 (3), Autosomal dominant

<i>FGF3</i>	164950	Deafness, congenital with inner ear agenesis, microtia, and microdontia, 610706 (3), Autosomal recessive
<i>FGF5</i>	165190	Trichomegaly, 190330 (3), Autosomal recessive
<i>FGF8</i>	600483	Hypogonadotropic hypogonadism 6 with or without anosmia, 612702 (3), Autosomal dominant
<i>FGF9</i>	600921	Multiple synostoses syndrome 3, 612961 (3), Autosomal dominant
<i>FGFR1</i>	136350	Pfeiffer syndrome, 101600 (3), Autosomal dominant; Jackson-Weiss syndrome, 123150 (3), Autosomal dominant; Trigonocephaly 1, 190440 (3), Autosomal dominant; Hypogonadotropic hypogonadism 2 with or without anosmia, 147950 (3), Autosomal dominant; Hartsfield syndrome, 615465 (3), Autosomal dominant; Osteoglophonic dysplasia, 166250 (3), Autosomal dominant; Encephalocraniocutaneous lipomatosis, somatic mosaic, 613001 (3)
<i>FGFR2</i>	176943	Apert syndrome, 101200 (3), Autosomal dominant; Craniosynostosis, nonspecific (3); Jackson-Weiss syndrome, 123150 (3), Autosomal dominant; Scaphocephaly and Axenfeld-Rieger anomaly (3); Saethre-Chotzen syndrome, 101400 (3), Autosomal dominant; Gastric cancer, somatic, 613659 (3); Scaphocephaly, maxillary retrusion, and mental retardation, 609579 (3); Bent bone dysplasia syndrome, 614592 (3), Autosomal dominant; LADD syndrome, 149730 (3), Autosomal dominant; Craniofacial-skeletal-dermatologic dysplasia, 101600 (3), Autosomal dominant; Pfeiffer syndrome, 101600 (3), Autosomal dominant; Crouzon syndrome, 123500 (3), Autosomal dominant; Beare-Stevenson cutis gyrata syndrome, 123790 (3), Autosomal dominant; Antley-Bixler syndrome without genital anomalies or disordered steroidogenesis, 207410 (3), Autosomal dominant
<i>FGFR3</i>	134934	Muenke syndrome, 602849 (3), Autosomal dominant; Nevus, epidermal, somatic, 162900 (3); Thanatophoric dysplasia, type II, 187601 (3), Autosomal dominant; Bladder cancer, somatic, 109800 (3); CATSHL syndrome, 610474 (3), Autosomal recessive, Autosomal dominant; Crouzon syndrome with acanthosis nigricans, 612247 (3), Autosomal dominant; Hypochondroplasia, 146000 (3), Autosomal dominant; LADD syndrome, 149730 (3), Autosomal dominant; Achondroplasia, 100800 (3), Autosomal dominant; Thanatophoric dysplasia, type I, 187600 (3), Autosomal dominant; Colorectal cancer, somatic, 114500 (3); Spermatocytic seminoma, somatic, 273300 (3); Cervical cancer, somatic, 603956 (3); SADDAN, 616482 (3), Autosomal dominant
<i>FGFR4</i>	134935	{Cancer progression/metastasis} (3)
<i>FGG</i>	134850	Hypofibrinogenemia, congenital, 202400 (3), Autosomal recessive; Hypodysfibrinogenemia, 616004 (3); Dysfibrinogenemia, congenital, 616004 (3); Afibrinogenemia, congenital, 202400 (3), Autosomal recessive

<i>FH</i>	136850	Fumarase deficiency, 606812 (3), Autosomal recessive; Leiomyomatosis and renal cell cancer, 150800 (3), Autosomal dominant
<i>FHL1</i>	300163	Reducing body myopathy, X-linked 1a, severe, infantile or early childhood onset, 300717 (3), X-linked dominant; Scapuloperoneal myopathy, X-linked dominant, 300695 (3), X-linked dominant; Reducing body myopathy, X-linked 1b, with late childhood or adult onset, 300718 (3), X-linked; ?Uruguay faciocardiomusculoskeletal syndrome, 300280 (3), X-linked recessive; Emery-Dreifuss muscular dystrophy 6, X-linked, 300696 (3), X-linked recessive; Myopathy, X-linked, with postural muscle atrophy, 300696 (3), X-linked recessive
<i>FIBP</i>	608296	Thauvin-Robinet-Faivre syndrome, 617107 (3), Autosomal recessive
<i>FIG4</i>	609390	Yunis-Varon syndrome, 216340 (3), Autosomal recessive; ?Polymicrogyria, bilateral temporooccipital, 612691 (3), Autosomal recessive; Charcot-Marie-Tooth disease, type 4J, 611228 (3), Autosomal recessive; Amyotrophic lateral sclerosis 11, 612577 (3), Autosomal dominant
<i>FIGLA</i>	608697	Premature ovarian failure 6, 612310 (3), Autosomal dominant
<i>FITM2</i>	612029	Siddiqi syndrome, 618635 (3), Autosomal recessive
<i>FKBP10</i>	607063	Bruck syndrome 1, 259450 (3), Autosomal recessive; Osteogenesis imperfecta, type XI, 610968 (3), Autosomal recessive
<i>FKBP14</i>	614505	Ehlers-Danlos syndrome, kyphoscoliotic type, 2, 614557 (3), Autosomal recessive
<i>FKBP5</i>	602623	{Major depressive disorder and accelerated response to antidepressant drug treatment}, 608516 (3)
<i>FKRP</i>	606596	Muscular dystrophy-dystroglycanopathy (limb-girdle), type C, 5, 607155 (3), Autosomal recessive; Muscular dystrophy-dystroglycanopathy (congenital with or without mental retardation), type B, 5, 606612 (3), Autosomal recessive; Muscular dystrophy-dystroglycanopathy (congenital with brain and eye anomalies), type A, 5, 613153 (3), Autosomal recessive
<i>FKTN</i>	607440	Muscular dystrophy-dystroglycanopathy (limb-girdle), type C, 4, 611588 (3), Autosomal recessive; Cardiomyopathy, dilated, 1X, 611615 (3), Autosomal recessive; Muscular dystrophy-dystroglycanopathy (congenital without mental retardation), type B, 4, 613152 (3), Autosomal recessive; Muscular dystrophy-dystroglycanopathy (congenital with brain and eye anomalies), type A, 4, 253800 (3), Autosomal recessive
<i>FLAD1</i>	610595	Lipid storage myopathy due to flavin adenine dinucleotide synthetase deficiency, 255100 (3), Autosomal recessive
<i>FLCN</i>	607273	Pneumothorax, primary spontaneous, 173600 (3), Autosomal dominant; Renal carcinoma, chromophobe, somatic, 144700 (3); Birt-Hogg-Dube syndrome, 135150 (3), Autosomal dominant; Colorectal cancer, somatic, 114500 (3)

<i>FLG</i>	135940	{Dermatitis, atopic, susceptibility to, 2}, 605803 (3); Ichthyosis vulgaris, 146700 (3), Autosomal recessive, Autosomal dominant
<i>FLG2</i>	616284	Peeling skin syndrome 6, 618084 (3), Autosomal recessive
<i>FLI1</i>	193067	Bleeding disorder, platelet-type, 21, 617443 (3), Autosomal recessive, Autosomal dominant
<i>FLNA</i>	300017	Otopalatodigital syndrome, type I, 311300 (3), X-linked dominant; Congenital short bowel syndrome, 300048 (3), X-linked recessive; Otopalatodigital syndrome, type II, 304120 (3), X-linked dominant; Intestinal pseudoobstruction, neuronal, 300048 (3), X-linked recessive; Melnick-Needles syndrome, 309350 (3), X-linked dominant; Cardiac valvular dysplasia, X-linked, 314400 (3), X-linked; ?FG syndrome 2, 300321 (3), X-linked; Heterotopia, periventricular, 1, 300049 (3), X-linked dominant; Terminal osseous dysplasia, 300244 (3), X-linked dominant; Frontometaphyseal dysplasia 1, 305620 (3), X-linked recessive
<i>FLNB</i>	603381	Larsen syndrome, 150250 (3), Autosomal dominant; Atelosteogenesis, type I, 108720 (3), Autosomal dominant; Boomerang dysplasia, 112310 (3), Autosomal dominant; Spondylotarsal synostosis syndrome, 272460 (3), Autosomal recessive; Atelosteogenesis, type III, 108721 (3), Autosomal dominant
<i>FLNC</i>	102565	Cardiomyopathy, familial hypertrophic, 26, 617047 (3), Autosomal dominant; Myopathy, myofibrillar, 5, 609524 (3), Autosomal dominant; Cardiomyopathy, familial restrictive 5, 617047 (3), Autosomal dominant; Myopathy, distal, 4, 614065 (3), Autosomal dominant
<i>FLRT3</i>	604808	Hypogonadotropic hypogonadism 21 with anosmia, 615271 (3), Autosomal dominant
<i>FLT3</i>	136351	Leukemia, acute myeloid, somatic, 601626 (3); Leukemia, acute lymphoblastic, somatic, 613065 (3); Leukemia, acute myeloid, reduced survival in, somatic, 601626 (3)
<i>FLT4</i>	136352	Congenital heart defects, multiple types, 7, 618780 (3), Autosomal dominant; Hemangioma, capillary infantile, somatic, 602089 (3); Lymphatic malformation 1, 153100 (3), Autosomal dominant
<i>FLVCR1</i>	609144	Ataxia, posterior column, with retinitis pigmentosa, 609033 (3), Autosomal recessive
<i>FLVCR2</i>	610865	Proliferative vasculopathy and hydranencephaly-hydrocephaly syndrome, 225790 (3), Autosomal recessive
<i>FMN2</i>	606373	Mental retardation, autosomal recessive 47, 616193 (3), Autosomal recessive
<i>FMO3</i>	136132	Trimethylaminuria, 602079 (3), Autosomal recessive
<i>FMR1</i>	309550	Premature ovarian failure 1, 311360 (3), X-linked; Fragile X tremor/ataxia syndrome, 300623 (3), X-linked dominant; Fragile X syndrome, 300624 (3), X-linked dominant

<i>FN1</i>	135600	Glomerulopathy with fibronectin deposits 2, 601894 (3), Autosomal dominant; Spondylometaphyseal dysplasia, corner fracture type, 184255 (3), Autosomal dominant
<i>FOLR1</i>	136430	Neurodegeneration due to cerebral folate transport deficiency, 613068 (3), Autosomal recessive
<i>FOXC1</i>	601090	Axenfeld-Rieger syndrome, type 3, 602482 (3), Autosomal dominant; Anterior segment dysgenesis 3, multiple subtypes, 601631 (3), Autosomal dominant
<i>FOXC2</i>	602402	Lymphedema-distichiasis syndrome, 153400 (3), Autosomal dominant; Lymphedema-distichiasis syndrome with renal disease and diabetes mellitus, 153400 (3), Autosomal dominant
<i>FOXD3</i>	611539	{Autoimmune disease, susceptibility to, 1}, 607836 (3), Autosomal dominant
<i>FOXE1</i>	602617	Bamforth-Lazarus syndrome, 241850 (3), Autosomal recessive; {Thyroid cancer, nonmedullary, 4}, 616534 (3), Autosomal dominant
<i>FOXE3</i>	601094	Cataract 34, multiple types, 612968 (3); Anterior segment dysgenesis 2, multiple subtypes, 610256 (3), Autosomal recessive; {Aortic aneurysm, familial thoracic 11, susceptibility to}, 617349 (3), Autosomal dominant
<i>FOXF1</i>	601089	Alveolar capillary dysplasia with misalignment of pulmonary veins, 265380 (3), Autosomal dominant
<i>FOXG1</i>	164874	Rett syndrome, congenital variant, 613454 (3), Autosomal dominant
<i>FOXI1</i>	601093	Enlarged vestibular aqueduct, 600791 (3), Autosomal recessive
<i>FOXJ1</i>	602291	Ciliary dyskinesia, primary, 43, 618699 (3), Autosomal dominant
<i>FOXL2</i>	605597	Blepharophimosis, epicanthus inversus, and ptosis, type 2, 110100 (3), Autosomal recessive, Autosomal dominant; Premature ovarian failure 3, 608996 (3), Autosomal dominant; Blepharophimosis, epicanthus inversus, and ptosis, type 1, 110100 (3), Autosomal recessive, Autosomal dominant
<i>FOXN1</i>	600838	T-cell lymphopenia, infantile, with or without nail dystrophy, autosomal dominant, 618806 (3); T-cell immunodeficiency, congenital alopecia, and nail dystrophy, 601705 (3), Autosomal recessive
<i>FOXO1</i>	136533	Rhabdomyosarcoma, alveolar, 268220 (3), Somatic mutation
<i>FOXP1</i>	605515	Mental retardation with language impairment and with or without autistic features, 613670 (3), Autosomal dominant
<i>FOXP2</i>	605317	Speech-language disorder-1, 602081 (3), Autosomal dominant
<i>FOXP3</i>	300292	Immunodysregulation, polyendocrinopathy, and enteropathy, X-linked, 304790 (3), X-linked recessive
<i>FOXRED1</i>	613622	Mitochondrial complex I deficiency, nuclear type 19, 618241 (3), Autosomal recessive
<i>FRAS1</i>	607830	Fraser syndrome 1, 219000 (3), Autosomal recessive

<i>FREM1</i>	608944	Manitoba oculotrichoanal syndrome, 248450 (3), Autosomal recessive; Trigonoccephaly 2, 614485 (3), Autosomal dominant; Bifid nose with or without anorectal and renal anomalies, 608980 (3)
<i>FREM2</i>	608945	Fraser syndrome 2, 617666 (3), Autosomal recessive; Cryptophthalmos, unilateral or bilateral, isolated, 123570 (3), Autosomal recessive
<i>FRMD4A</i>	616305	?Corpus callosum, agenesis of, with facial anomalies and cerebellar ataxia, 616819 (3), Autosomal recessive
<i>FRMD7</i>	300628	Nystagmus 1, congenital, X-linked, 310700 (3), X-linked; Nystagmus, infantile periodic alternating, X-linked, 310700 (3), X-linked
<i>FRMPD4</i>	300838	Mental retardation, X-linked 104, 300983 (3), X-linked
<i>FRRS1L</i>	604574	Epileptic encephalopathy, early infantile, 37, 616981 (3), Autosomal recessive
<i>FRZB</i>	605083	{Osteoarthritis susceptibility 1}, 165720 (3), Multifactorial
<i>FSCN2</i>	607643	Retinitis pigmentosa 30, 607921 (3)
<i>FSHB</i>	136530	Hypogonadotropic hypogonadism 24 without anosmia, 229070 (3), Autosomal recessive
<i>FSHR</i>	136435	Ovarian hyperstimulation syndrome, 608115 (3), Autosomal dominant; Ovarian dysgenesis 1, 233300 (3), Autosomal recessive; Ovarian response to FSH stimulation, 276400 (3), Autosomal recessive
<i>FSIP2</i>	615796	Spermatogenic failure 34, 618153 (3), Autosomal recessive
<i>FTCD</i>	606806	Glutamate formiminotransferase deficiency, 229100 (3), Autosomal recessive
<i>FTH1</i>	134770	?Hemochromatosis, type 5, 615517 (3), Autosomal dominant
<i>FTL</i>	134790	Hyperferritinemia-cataract syndrome, 600886 (3), Autosomal dominant; Neurodegeneration with brain iron accumulation 3, 606159 (3), Autosomal dominant; L-ferritin deficiency, dominant and recessive, 615604 (3), Autosomal recessive, Autosomal dominant
<i>FTO</i>	610966	Growth retardation, developmental delay, facial dysmorphism, 612938 (3), Autosomal recessive; {Obesity, susceptibility to, BMIQ14}, 612460 (3), Autosomal recessive
<i>FTSJ1</i>	300499	Mental retardation, X-linked 9/44, 309549 (3), X-linked recessive
<i>FUCA1</i>	612280	Fucosidosis, 230000 (3), Autosomal recessive
<i>FUS</i>	137070	Amyotrophic lateral sclerosis 6, with or without frontotemporal dementia, 608030 (3); Essential tremor, hereditary, 4, 614782 (3), Autosomal dominant
<i>FUT1</i>	211100	[Bombay phenotype], 616754 (3), Autosomal recessive
<i>FUT2</i>	182100	{Vitamin B12 plasma level QTL1}, 612542 (3); [Bombay phenotype, digenic], 616754 (3), Autosomal recessive; {Norwalk virus infection, resistance to} (3)
<i>FUT3</i>	111100	[Blood group, Lewis], 618983 (3)
<i>FUT6</i>	136836	[Fucosyltransferase 6 deficiency], 613852 (3)

<i>FUT8</i>	602589	Congenital disorder of glycosylation with defective fucosylation 1, 618005 (3), Autosomal recessive
<i>FUZ</i>	610622	{Neural tube defects, susceptibility to}, 182940 (3), Autosomal dominant
<i>FXN</i>	606829	Friedreich ataxia with retained reflexes, 229300 (3), Autosomal recessive; Friedreich ataxia, 229300 (3), Autosomal recessive
<i>FXR1</i>	600819	?Myopathy, congenital proximal, with minicore lesions, 618823 (3), Autosomal recessive; ?Myopathy, congenital, with respiratory insufficiency and bone fractures, 618822 (3), Autosomal recessive
<i>FXD2</i>	601814	Hypomagnesemia 2, renal, 154020 (3), Autosomal dominant
<i>FYB1</i>	602731	Thrombocytopenia 3, 273900 (3), Autosomal recessive
<i>FYCO1</i>	607182	Cataract 18, autosomal recessive, 610019 (3), Autosomal recessive
<i>FZD2</i>	600667	Omodysplasia 2, 164745 (3), Autosomal dominant
<i>FZD4</i>	604579	Exudative vitreoretinopathy 1, 133780 (3), Autosomal dominant; Retinopathy of prematurity, 133780 (3), Autosomal dominant
<i>FZD6</i>	603409	Nail disorder, nonsyndromic congenital, 1, 161050 (3), Autosomal recessive
<i>G6PC</i>	613742	Glycogen storage disease Ia, 232200 (3), Autosomal recessive
<i>G6PC3</i>	611045	Dursun syndrome, 612541 (3), Autosomal recessive; Neutropenia, severe congenital 4, autosomal recessive, 612541 (3), Autosomal recessive
<i>G6PD</i>	305900	Hemolytic anemia, G6PD deficient (favism), 300908 (3), X-linked dominant; {Resistance to malaria due to G6PD deficiency}, 611162 (3)
<i>GAA</i>	606800	Glycogen storage disease II, 232300 (3), Autosomal recessive
<i>GAB1</i>	604439	?Deafness, autosomal recessive 26, 605428 (3), Autosomal recessive
<i>GABBR2</i>	607340	Neurodevelopmental disorder with poor language and loss of hand skills, 617903 (3), Autosomal dominant; Epileptic encephalopathy, early infantile, 59, 617904 (3), Autosomal dominant; {Nicotine dependence, susceptibility to}, 188890 (3); {Nicotine dependence, protection against}, 188890 (3)
<i>GABRA1</i>	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
<i>GABRA2</i>	137140	{Alcohol dependence, susceptibility to}, 103780 (3), Multifactorial; Epileptic encephalopathy, early infantile, 78, 618557 (3), Autosomal dominant
<i>GABRA5</i>	137142	Epileptic encephalopathy, early infantile, 79, 618559 (3), Autosomal dominant
<i>GABRB1</i>	137190	Epileptic encephalopathy, early infantile, 45, 617153 (3), Autosomal dominant

<i>GABRB2</i>	600232	Epileptic encephalopathy, infantile or early childhood, 2, 617829 (3), Autosomal dominant
<i>GABRB3</i>	137192	{Epilepsy, childhood absence, susceptibility to, 5}, 612269 (3); Epileptic encephalopathy, early infantile, 43, 617113 (3), Autosomal dominant
<i>GABRD</i>	137163	{Epilepsy, juvenile myoclonic, susceptibility to}, 613060 (3), Autosomal dominant; {Epilepsy, idiopathic generalized, 10}, 613060 (3), Autosomal dominant; {Epilepsy, generalized, with febrile seizures plus, type 5, susceptibility to}, 613060 (3), Autosomal dominant
<i>GABRG2</i>	137164	Epileptic encephalopathy, early infantile, 74, 618396 (3), Autosomal dominant; Febrile seizures, familial, 8, 607681 (3), Autosomal dominant; Epilepsy, generalized, with febrile seizures plus, type 3, 607681 (3), Autosomal dominant
<i>GAD1</i>	605363	?Cerebral palsy, spastic quadriplegic, 1, 603513 (3), Autosomal recessive
<i>GAL</i>	137035	?Epilepsy, familial temporal lobe, 8, 616461 (3), Autosomal dominant
<i>GALC</i>	606890	Krabbe disease, 245200 (3), Autosomal recessive
<i>GALE</i>	606953	Galactose epimerase deficiency, 230350 (3), Autosomal recessive
<i>GALK1</i>	604313	Galactokinase deficiency with cataracts, 230200 (3), Autosomal recessive
<i>GALM</i>	137030	Galactosemia IV, 618881 (3), Autosomal recessive
<i>GALNS</i>	612222	Mucopolysaccharidosis IVA, 253000 (3), Autosomal recessive
<i>GALNT12</i>	610290	{Colorectal cancer, susceptibility to, 1}, 608812 (3)
<i>GALNT2</i>	602274	Congenital disorder of glycosylation, type II, 618885 (3), Autosomal recessive
<i>GALNT3</i>	601756	Tumoral calcinosis, hyperphosphatemic, familial, 1, 211900 (3), Autosomal recessive
<i>GALT</i>	606999	Galactosemia, 230400 (3), Autosomal recessive
<i>GAMT</i>	601240	Cerebral creatine deficiency syndrome 2, 612736 (3), Autosomal recessive
<i>GAN</i>	605379	Giant axonal neuropathy-1, 256850 (3), Autosomal recessive
<i>GANAB</i>	104160	Polycystic kidney disease 3, 600666 (3), Autosomal dominant
<i>GARS1 (GARS)</i>	600287	Charcot-Marie-Tooth disease, type 2D, 601472 (3), Autosomal dominant; Neuronopathy, distal hereditary motor, type VA, 600794 (3), Autosomal dominant
<i>GAS2L2</i>	611398	?Ciliary dyskinesia, primary, 41, 618449 (3), Autosomal recessive
<i>GAS8</i>	605178	Ciliary dyskinesia, primary, 33, 616726 (3), Autosomal recessive
<i>GATA1</i>	305371	Leukemia, megakaryoblastic, with or without Down syndrome, somatic, 190685 (3); Anemia, X-linked, with/without neutropenia and/or platelet abnormalities, 300835 (3), X-linked recessive; Thrombocytopenia, X-linked, with or without dyserythropoietic anemia, 300367 (3), X-linked recessive;

Thrombocytopenia with beta-thalassemia, X-linked, 314050 (3),
X-linked recessive

<i>GATA2</i>	137295	Emberger syndrome, 614038 (3), Autosomal dominant; {Myelodysplastic syndrome, susceptibility to}, 614286 (3); Immunodeficiency 21, 614172 (3), Autosomal dominant; {Leukemia, acute myeloid, susceptibility to}, 601626 (3), Autosomal dominant, Somatic mutation
<i>GATA3</i>	131320	Hypoparathyroidism, sensorineural deafness, and renal dysplasia, 146255 (3), Autosomal dominant
<i>GATA4</i>	600576	?Testicular anomalies with or without congenital heart disease, 615542 (3), Autosomal dominant; Tetralogy of Fallot, 187500 (3), Autosomal dominant; Atrioventricular septal defect 4, 614430 (3), Autosomal dominant; Atrial septal defect 2, 607941 (3), Autosomal dominant; Ventricular septal defect 1, 614429 (3), Autosomal dominant
<i>GATA5</i>	611496	Congenital heart defects, multiple types, 5, 617912 (3), Autosomal recessive, Autosomal dominant
<i>GATA6</i>	601656	Pancreatic agenesis and congenital heart defects, 600001 (3), Autosomal dominant; Atrial septal defect 9, 614475 (3), Autosomal dominant; Atrioventricular septal defect 5, 614474 (3), Autosomal dominant; Persistent truncus arteriosus, 217095 (3); Tetralogy of Fallot, 187500 (3), Autosomal dominant
<i>GATAD1</i>	614518	?Cardiomyopathy, dilated, 2B, 614672 (3), Autosomal recessive
<i>GATAD2B</i>	614998	Mental retardation, autosomal dominant 18, 615074 (3), Autosomal dominant
<i>GATB</i>	603645	?Combined oxidative phosphorylation deficiency 41, 618838 (3), Autosomal recessive
<i>GATC</i>	617210	Combined oxidative phosphorylation deficiency 42, 618839 (3), Autosomal recessive
<i>GATM</i>	602360	Cerebral creatine deficiency syndrome 3, 612718 (3), Autosomal recessive; Fanconi renotubular syndrome 1, 134600 (3), Autosomal dominant
<i>GBA</i>	606463	Gaucher disease, type III, 231000 (3), Autosomal recessive; {Parkinson disease, late-onset, susceptibility to}, 168600 (3), Autosomal dominant, Multifactorial; Gaucher disease, type IIIC, 231005 (3), Autosomal recessive; Gaucher disease, type I, 230800 (3), Autosomal recessive; Gaucher disease, perinatal lethal, 608013 (3), Autosomal recessive; Gaucher disease, type II, 230900 (3), Autosomal recessive; {Lewy body dementia, susceptibility to}, 127750 (3), Autosomal dominant
<i>GBA2</i>	609471	Spastic paraplegia 46, autosomal recessive, 614409 (3), Autosomal recessive

<i>GBE1</i>	607839	Polyglucosan body disease, adult form, 263570 (3), Autosomal recessive; Glycogen storage disease IV, 232500 (3), Autosomal recessive
<i>GCDH</i>	608801	Glutaricaciduria, type I, 231670 (3), Autosomal recessive
<i>GCH1</i>	600225	Hyperphenylalaninemia, BH4-deficient, B, 233910 (3), Autosomal recessive; Dystonia, DOPA-responsive, with or without hyperphenylalaninemia, 128230 (3), Autosomal recessive, Autosomal dominant
<i>GCK</i>	138079	Diabetes mellitus, noninsulin-dependent, late onset, 125853 (3), Autosomal dominant; MODY, type II, 125851 (3), Autosomal dominant; Diabetes mellitus, permanent neonatal 1, 606176 (3), Autosomal recessive; Hyperinsulinemic hypoglycemia, familial, 3, 602485 (3), Autosomal dominant
<i>GCKR</i>	600842	[Fasting plasma glucose level QTL 5], 613463 (3)
<i>GCLC</i>	606857	Hemolytic anemia due to gamma-glutamylcysteine synthetase deficiency, 230450 (3), Autosomal recessive; {Myocardial infarction, susceptibility to}, 608446 (3)
<i>GCLM</i>	601176	{Myocardial infarction, susceptibility to}, 608446 (3)
<i>GCM2</i>	603716	Hyperparathyroidism 4, 617343 (3), Autosomal dominant; Hypoparathyroidism, familial isolated 2, 618883 (3)
<i>GCNT2</i>	600429	Adult i phenotype without cataract, 110800 (3), Autosomal dominant; Cataract 13 with adult i phenotype, 116700 (3), Autosomal recessive; [Blood group, Ii], 110800 (3), Autosomal dominant
<i>GCSH</i>	238330	?Glycine encephalopathy, 605899 (3), Autosomal recessive
<i>GDAP1</i>	606598	Charcot-Marie-Tooth disease, recessive intermediate, A, 608340 (3), Autosomal recessive; Charcot-Marie-Tooth disease, type 4A, 214400 (3), Autosomal recessive; 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
<i>GDAP2</i>	618128	Spinocerebellar ataxia, autosomal recessive 27, 618369 (3), Autosomal recessive
<i>GDF1</i>	602880	Right atrial isomerism (Ivemark), 208530 (3), Autosomal recessive; Congenital heart defects, multiple types, 6, 613854 (3), Autosomal dominant
<i>GDF2</i>	605120	Telangiectasia, hereditary hemorrhagic, type 5, 615506 (3), Autosomal dominant
<i>GDF3</i>	606522	Microphthalmia, isolated 7, 613704 (3), Autosomal dominant; Microphthalmia with coloboma 6, 613703 (3), Autosomal dominant; Klippel-Feil syndrome 3, autosomal dominant, 613702 (3)

<i>GDF5</i>	601146	?Acromesomelic dysplasia, Hunter-Thompson type, 201250 (3), Autosomal recessive; Symphalangism, proximal, 1B, 615298 (3), Autosomal dominant; Brachydactyly, type A1, C, 615072 (3), Autosomal recessive, Autosomal dominant; Chondrodysplasia, Grebe type, 200700 (3), Autosomal recessive; Brachydactyly, type A2, 112600 (3), Autosomal dominant; Du Pan syndrome, 228900 (3), Autosomal recessive; {Osteoarthritis-5}, 612400 (3); Brachydactyly, type C, 113100 (3), Autosomal dominant; Multiple synostoses syndrome 2, 610017 (3), Autosomal dominant
<i>GDF6</i>	601147	Leber congenital amaurosis 17, 615360 (3), Autosomal recessive; Klippel-Feil syndrome 1, autosomal dominant, 118100 (3), Autosomal dominant; Multiple synostoses syndrome 4, 617898 (3), Autosomal dominant; Microphthalmia with coloboma 6, digenic, 613703 (3), Autosomal dominant; Microphthalmia, isolated 4, 613094 (3)
<i>GDF9</i>	601918	?Premature ovarian failure 14, 618014 (3), Autosomal recessive
<i>GDI1</i>	300104	Mental retardation, X-linked 41, 300849 (3), X-linked dominant
<i>GDNF</i>	600837	{Pheochromocytoma, modifier of}, 171300 (3), Autosomal dominant; {Hirschsprung disease, susceptibility to, 3}, 613711 (3), Autosomal dominant; Central hypoventilation syndrome, 209880 (3), Autosomal dominant
<i>GEMIN4</i>	606969	Neurodevelopmental disorder with microcephaly, cataracts, and renal abnormalities, 617913 (3), Autosomal recessive
<i>GFAP</i>	137780	Alexander disease, 203450 (3), Autosomal dominant
<i>GFER</i>	600924	Myopathy, mitochondrial progressive, with congenital cataract and developmental delay, 613076 (3)
<i>GFI1</i>	600871	?Neutropenia, nonimmune chronic idiopathic, of adults, 607847 (3), Autosomal dominant; Neutropenia, severe congenital 2, autosomal dominant, 613107 (3), Autosomal dominant
<i>GFI1B</i>	604383	Bleeding disorder, platelet-type, 17, 187900 (3), Autosomal recessive, Autosomal dominant
<i>GFM1</i>	606639	Combined oxidative phosphorylation deficiency 1, 609060 (3), Autosomal recessive
<i>GFM2</i>	606544	Combined oxidative phosphorylation deficiency 39, 618397 (3), Autosomal recessive
<i>GFPT1</i>	138292	Myasthenia, congenital, 12, with tubular aggregates, 610542 (3), Autosomal recessive
<i>GGCX</i>	137167	Vitamin K-dependent clotting factors, combined deficiency of, 1, 277450 (3), Autosomal recessive; Pseudoxanthoma elasticum-like disorder with multiple coagulation factor deficiency, 610842 (3)
<i>GGT1</i>	612346	?Glutathioninuria, 231950 (3), Autosomal recessive
<i>GGT2</i>	137181	[Gamma-glutamyltransferase, familial high serum], 137181 (2)

<i>GH1</i>	139250	Kowarski syndrome, 262650 (3), Autosomal recessive; 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
<i>GHR</i>	600946	{Hypercholesterolemia, familial, modifier of}, 143890 (3), Autosomal dominant; Increased responsiveness to growth hormone, 604271 (3), Autosomal dominant; Laron dwarfism, 262500 (3), Autosomal recessive; Growth hormone insensitivity, partial, 604271 (3), Autosomal dominant
<i>GHRH</i>	139190	?Isolated growth hormone deficiency due to defect in GHRF (1); Gigantism due to GHRF hypersecretion (1)
<i>GHRHR</i>	139191	Growth hormone deficiency, isolated, type IV, 618157 (3), Autosomal recessive
<i>GHRL</i>	605353	{Obesity, susceptibility to}, 601665 (3), Autosomal recessive, Autosomal dominant, Multifactorial
<i>GHSR</i>	601898	Growth hormone deficiency, isolated partial, 615925 (3), Autosomal recessive, Autosomal dominant
<i>GIGYF2</i>	612003	{Parkinson disease 11}, 607688 (3)
<i>GINS1</i>	610608	Immunodeficiency 55, 617827 (3), Autosomal recessive
<i>GIPC1</i>	605072	Oculopharyngodistal myopathy 2, 618940 (3), Autosomal dominant
<i>GIPC3</i>	608792	Deafness, autosomal recessive 15, 601869 (3), Autosomal recessive
<i>GJA1</i>	121014	Erythrokeratoderma variabilis et progressiva 3, 617525 (3), Autosomal dominant; Craniometaphyseal dysplasia, autosomal recessive, 218400 (3), Autosomal recessive; Atrioventricular septal defect 3, 600309 (3), Autosomal dominant; Oculodentodigital dysplasia, 164200 (3), Autosomal dominant; Syndactyly, type III, 186100 (3), Autosomal dominant; Oculodentodigital dysplasia, autosomal recessive, 257850 (3), Autosomal recessive; Hypoplastic left heart syndrome 1, 241550 (3), Autosomal recessive; Palmoplantar keratoderma with congenital alopecia, 104100 (3), Autosomal dominant
<i>GJA3</i>	121015	Cataract 14, multiple types, 601885 (3), Autosomal dominant
<i>GJA5</i>	121013	Atrial fibrillation, familial, 11, 614049 (3), Autosomal dominant; Atrial standstill, digenic (GJA5/SCN5A), 108770 (3), Autosomal dominant
<i>GJA8</i>	600897	Cataract 1, multiple types, 116200 (3), Autosomal dominant
<i>GJB1</i>	304040	Charcot-Marie-Tooth neuropathy, X-linked dominant, 1, 302800 (3), X-linked dominant

<i>GJB2</i>	121011	Deafness, autosomal dominant 3A, 601544 (3), Autosomal dominant; Deafness, autosomal recessive 1A, 220290 (3), Autosomal recessive, Digenic dominant; Bart-Pumphrey syndrome, 149200 (3), Autosomal dominant; Vohwinkel syndrome, 124500 (3), Autosomal dominant; Keratoderma, palmoplantar, with deafness, 148350 (3), Autosomal dominant; Keratitis-ichthyosis-deafness syndrome, 148210 (3), Autosomal dominant; Hystrix-like ichthyosis with deafness, 602540 (3), Autosomal dominant
<i>GJB3</i>	603324	Deafness, autosomal dominant 2B, 612644 (3), Autosomal dominant; Deafness, autosomal recessive (3); Deafness, autosomal dominant, with peripheral neuropathy (3); Deafness, digenic, GJB2/GJB3, 220290 (3), Autosomal recessive, Digenic dominant; Erythrokeratoderma variabilis et progressiva 1, 133200 (3), Autosomal recessive, Autosomal dominant
<i>GJB4</i>	605425	Erythrokeratoderma variabilis et progressiva 2, 617524 (3), Autosomal dominant
<i>GJB6</i>	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, Digenic dominant; Ectodermal dysplasia 2, Clouston type, 129500 (3), Autosomal dominant
<i>GJC2</i>	608803	Spastic paraplegia 44, autosomal recessive, 613206 (3), Autosomal recessive; Lymphatic malformation 3, 613480 (3), Autosomal dominant; Leukodystrophy, hypomyelinating, 2, 608804 (3), Autosomal recessive
<i>GK</i>	300474	Glycerol kinase deficiency, 307030 (3), X-linked recessive
<i>GLA</i>	300644	Fabry disease, 301500 (3), X-linked; Fabry disease, cardiac variant, 301500 (3), X-linked
<i>GLB1</i>	611458	GM1-gangliosidosis, type III, 230650 (3), Autosomal recessive; GM1-gangliosidosis, type I, 230500 (3), Autosomal recessive; Mucopolysaccharidosis type IVB (Morquio), 253010 (3), Autosomal recessive; GM1-gangliosidosis, type II, 230600 (3), Autosomal recessive
<i>GLCCI1</i>	614283	{Glucocorticoid therapy, response to}, 614400 (3)
<i>GLDC</i>	238300	Glycine encephalopathy, 605899 (3), Autosomal recessive
<i>GLDN</i>	608603	Lethal congenital contracture syndrome 11, 617194 (3), Autosomal recessive
<i>GLE1</i>	603371	Congenital arthrogryposis with anterior horn cell disease, 611890 (3), Autosomal recessive; Lethal congenital contracture syndrome 1, 253310 (3), Autosomal recessive
<i>GLI1</i>	165220	Polydactyly, postaxial, type A8, 618123 (3), Autosomal recessive; Polydactyly, preaxial I, 174400 (3), Autosomal recessive
<i>GLI2</i>	165230	Culler-Jones syndrome, 615849 (3), Autosomal dominant; Holoprosencephaly 9, 610829 (3), Autosomal dominant

<i>GLI3</i>	165240	Polydactyly, postaxial, types A1 and B, 174200 (3), Autosomal dominant; {Hypothalamic hamartomas, somatic}, 241800 (3); Greig cephalopolysyndactyly syndrome, 175700 (3), Autosomal dominant; Polydactyly, preaxial, type IV, 174700 (3), Autosomal dominant; Pallister-Hall syndrome, 146510 (3), Autosomal dominant
<i>GLIS2</i>	608539	Nephronophthisis 7, 611498 (3)
<i>GLIS3</i>	610192	Diabetes mellitus, neonatal, with congenital hypothyroidism, 610199 (3), Autosomal recessive
<i>GLMN</i>	601749	Glomuvenous malformations, 138000 (3), Autosomal dominant
<i>GLRA1</i>	138491	Hyperekplexia 1, 149400 (3), Autosomal recessive, Autosomal dominant
<i>GLRB</i>	138492	Hyperekplexia 2, 614619 (3), Autosomal recessive
<i>GLRX5</i>	609588	Anemia, sideroblastic, 3, pyridoxine-refractory, 616860 (3), Autosomal recessive; Spasticity, childhood-onset, with hyperglycinemia, 616859 (3), Autosomal recessive
<i>GLS</i>	138280	?Infantile cataract, skin abnormalities, glutamate excess, and impaired intellectual development, 618339 (3), Autosomal dominant; Global developmental delay, progressive ataxia, and elevated glutamine, 618412 (3), Autosomal recessive; Epileptic encephalopathy, early infantile, 71, 618328 (3), Autosomal recessive
<i>GLUD1</i>	138130	Hyperinsulinism-hyperammonemia syndrome, 606762 (3), Autosomal dominant
<i>GLUD2</i>	300144	{Parkinson disease, age of onset, modifier}, 168600 (3), Autosomal dominant, Multifactorial
<i>GLUL</i>	138290	Glutamine deficiency, congenital, 610015 (3), Autosomal recessive
<i>GLYCTK</i>	610516	D-glyceric aciduria, 220120 (3), Autosomal recessive
<i>GM2A</i>	613109	GM2-gangliosidosis, AB variant, 272750 (3), Autosomal recessive
<i>GMNN</i>	602842	Meier-Gorlin syndrome 6, 616835 (3), Autosomal dominant
<i>GMPPA</i>	615495	Alacrima, achalasia, and mental retardation syndrome, 615510 (3), Autosomal recessive
<i>GMPPB</i>	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
<i>GNA11</i>	139313	Hypocalciuric hypercalcemia, type II, 145981 (3), Autosomal dominant; Hypocalcemia, autosomal dominant 2, 615361 (3), Autosomal dominant
<i>GNAI2</i>	139360	Ventricular tachycardia, idiopathic, 192605 (3), Autosomal dominant; Pituitary adenoma, ACTH-secreting, somatic (3)
<i>GNAI3</i>	139370	Auriculocondylar syndrome 1, 602483 (3), Autosomal dominant
<i>GNAL</i>	139312	Dystonia 25, 615073 (3), Autosomal dominant

<i>GNAO1</i>	139311	Epileptic encephalopathy, early infantile, 17, 615473 (3), Autosomal dominant; Neurodevelopmental disorder with involuntary movements, 617493 (3), Autosomal dominant
<i>GNAQ</i>	600998	Sturge-Weber syndrome, somatic, mosaic, 185300 (3); Capillary malformations, congenital, 1, somatic, mosaic, 163000 (3)
<i>GNAS</i>	139320	ACTH-independent macronodular adrenal hyperplasia, 219080 (3), Somatic mutation; Pseudohypoparathyroidism 1c, 612462 (3), Autosomal dominant; Pseudohypoparathyroidism 1b, 603233 (3), Autosomal dominant; Pseudopseudohypoparathyroidism, 612463 (3), Autosomal dominant; McCune-Albright syndrome, somatic, mosaic, 174800 (3); Osseous heteroplasia, progressive, 166350 (3), Autosomal dominant; Pituitary adenoma 3, multiple types, somatic, 617686 (3); Pseudohypoparathyroidism 1a, 103580 (3), Autosomal dominant
<i>GNAT1</i>	139330	Night blindness, congenital stationary, type 1G, 616389 (3), Autosomal recessive; Night blindness, congenital stationary, autosomal dominant 3, 610444 (3), Autosomal dominant
<i>GNAT2</i>	139340	Achromatopsia 4, 613856 (3)
<i>GNB1</i>	139380	Mental retardation, autosomal dominant 42, 616973 (3), Autosomal dominant; Leukemia, acute lymphoblastic, somatic, 613065 (3)
<i>GNB3</i>	139130	{Hypertension, essential, susceptibility to}, 145500 (3), Multifactorial; Night blindness, congenital stationary, type 1H, 617024 (3), Autosomal recessive
<i>GNB4</i>	610863	Charcot-Marie-Tooth disease, dominant intermediate F, 615185 (3), Autosomal dominant
<i>GNB5</i>	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
<i>GNE</i>	603824	Sialuria, 269921 (3), Autosomal dominant; Nonaka myopathy, 605820 (3), Autosomal recessive
<i>GNMT</i>	606628	Glycine N-methyltransferase deficiency, 606664 (3), Autosomal recessive
<i>GNPAT</i>	602744	Rhizomelic chondrodysplasia punctata, type 2, 222765 (3), Autosomal recessive
<i>GNPTAB</i>	607840	Mucopolipidosis II alpha/beta, 252500 (3), Autosomal recessive; Mucopolipidosis III alpha/beta, 252600 (3), Autosomal recessive
<i>GNPTG</i>	607838	Mucopolipidosis III gamma, 252605 (3), Autosomal recessive
<i>GNRH1</i>	152760	?Hypogonadotropic hypogonadism 12 with or without anosmia, 614841 (3), Autosomal recessive
<i>GNRHR</i>	138850	Hypogonadotropic hypogonadism 7 without anosmia, 146110 (3), Autosomal recessive
<i>GNS</i>	607664	Mucopolysaccharidosis type IIID, 252940 (3), Autosomal recessive
<i>GORAB</i>	607983	Geroderma osteodysplasticum, 231070 (3), Autosomal recessive

<i>GOSR2</i>	604027	Epilepsy, progressive myoclonic 6, 614018 (3), Autosomal recessive
<i>GOT1</i>	138180	Aspartate aminotransferase, serum level of, QTL1, 614419 (3)
<i>GOT2</i>	138150	Epileptic encephalopathy, early infantile, 82, 618721 (3), Autosomal recessive
<i>GP1BA</i>	606672	Bernard-Soulier syndrome, type A1 (recessive), 231200 (3), Autosomal recessive; {Nonarteritic anterior ischemic optic neuropathy, susceptibility to}, 258660 (3), Autosomal recessive; von Willebrand disease, platelet-type, 177820 (3), Autosomal dominant; Bernard-Soulier syndrome, type A2 (dominant), 153670 (3), Autosomal dominant
<i>GP1BB</i>	138720	Giant platelet disorder, isolated, 231200 (3), Autosomal recessive; Bernard-Soulier syndrome, type B, 231200 (3), Autosomal recessive
<i>GP6</i>	605546	Bleeding disorder, platelet-type, 11, 614201 (3), Autosomal recessive
<i>GP9</i>	173515	Bernard-Soulier syndrome, type C, 231200 (3), Autosomal recessive
<i>GPAA1</i>	603048	Glycosylphosphatidylinositol biosynthesis defect 15, 617810 (3), Autosomal recessive
<i>GPC3</i>	300037	Simpson-Golabi-Behmel syndrome, type 1, 312870 (3), X-linked recessive; Wilms tumor, somatic, 194070 (3)
<i>GPC4</i>	300168	Keipert syndrome, 301026 (3), X-linked recessive
<i>GPC6</i>	604404	Omodysplasia 1, 258315 (3), Autosomal recessive
<i>GPD1</i>	138420	Hypertriglyceridemia, transient infantile, 614480 (3), Autosomal recessive
<i>GPD1L</i>	611778	Brugada syndrome 2, 611777 (3)
<i>GPD2</i>	138430	{Diabetes, type 2, susceptibility to}, 125853 (3), Autosomal dominant
<i>GPHN</i>	603930	Molybdenum cofactor deficiency C, 615501 (3), Autosomal recessive
<i>GPI</i>	172400	Hemolytic anemia, nonspherocytic, due to glucose phosphate isomerase deficiency, 613470 (3), Autosomal recessive
<i>GPIHBP1</i>	612757	Hyperlipoproteinemia, type 1D, 615947 (3), Autosomal recessive
<i>GPNMB</i>	604368	Amyloidosis, primary localized cutaneous, 3, 617920 (3), Autosomal recessive
<i>GPR101</i>	300393	Pituitary adenoma 2, GH-secreting, 300943 (3), X-linked
<i>GPR143</i>	300808	Ocular albinism, type I, Nettleship-Falls type, 300500 (3), X-linked; Nystagmus 6, congenital, X-linked, 300814 (3), X-linked recessive
<i>GPR179</i>	614515	Night blindness, congenital stationary (complete), 1E, autosomal recessive, 614565 (3), Autosomal recessive
<i>GPR68</i>	601404	Amelogenesis imperfecta, hypomaturation type, IIA6, 617217 (3), Autosomal recessive
<i>GPR88</i>	607468	?Chorea, childhood-onset, with psychomotor retardation, 616939 (3), Autosomal recessive

<i>GPRASP2</i>	300969	?Deafness, X-linked 7, 301018 (3), X-linked recessive
<i>GPSM2</i>	609245	Chudley-McCullough syndrome, 604213 (3), Autosomal recessive
<i>GPT2</i>	138210	Mental retardation, autosomal recessive 49, 616281 (3), Autosomal recessive
<i>GPX1</i>	138320	Hemolytic anemia due to glutathione peroxidase deficiency, 614164 (1), Autosomal recessive
<i>GPX4</i>	138322	Spondylometaphyseal dysplasia, Sedaghatian type, 250220 (3), Autosomal recessive
<i>GRAP</i>	604330	Deafness, autosomal recessive 114, 618456 (3), Autosomal recessive
<i>GREB1L</i>	617782	Renal hypodysplasia/aplasia 3, 617805 (3), Autosomal dominant
<i>GREM2</i>	608832	Tooth agenesis, selective, 9, 617275 (3), Autosomal dominant
<i>GRHL2</i>	608576	Deafness, autosomal dominant 28, 608641 (3), Autosomal dominant; Corneal dystrophy, posterior polymorphous, 4, 618031 (3), Autosomal dominant; Ectodermal dysplasia/short stature syndrome, 616029 (3), Autosomal recessive
<i>GRHL3</i>	608317	Van der Woude syndrome 2, 606713 (3), Autosomal dominant
<i>GRHPR</i>	604296	Hyperoxaluria, primary, type II, 260000 (3), Autosomal recessive
<i>GRIA2</i>	138247	Neurodevelopmental disorder with language impairment and behavioral abnormalities, 618917 (3), Autosomal dominant
<i>GRIA3</i>	305915	Intellectual developmental disorder, X-linked, syndromic, Wu type, 300699 (3), X-linked recessive
<i>GRIA4</i>	138246	Neurodevelopmental disorder with or without seizures and gait abnormalities, 617864 (3), Autosomal dominant
<i>GRID2</i>	602368	Spinocerebellar ataxia, autosomal recessive 18, 616204 (3), Autosomal recessive
<i>GRIK2</i>	138244	Mental retardation, autosomal recessive, 6, 611092 (3), Autosomal recessive
<i>GRIN1</i>	138249	Neurodevelopmental disorder with or without hyperkinetic movements and seizures, autosomal recessive, 617820 (3), Autosomal recessive; Neurodevelopmental disorder with or without hyperkinetic movements and seizures, autosomal dominant, 614254 (3), Autosomal dominant
<i>GRIN2A</i>	138253	Epilepsy, focal, with speech disorder and with or without mental retardation, 245570 (3), Autosomal dominant
<i>GRIN2B</i>	138252	Epileptic encephalopathy, early infantile, 27, 616139 (3), Autosomal dominant; Mental retardation, autosomal dominant 6, 613970 (3), Autosomal dominant
<i>GRIN2D</i>	602717	Epileptic encephalopathy, early infantile, 46, 617162 (3), Autosomal dominant
<i>GRIP1</i>	604597	Fraser syndrome 3, 617667 (3), Autosomal recessive
<i>GRK1</i>	180381	Oguchi disease-2, 613411 (3)
<i>GRM1</i>	604473	Spinocerebellar ataxia 44, 617691 (3), Autosomal dominant; Spinocerebellar ataxia, autosomal recessive 13, 614831 (3), Autosomal recessive

<i>GRM6</i>	604096	Night blindness, congenital stationary (complete), 1B, autosomal recessive, 257270 (3), Autosomal recessive
<i>GRM7</i>	604101	Neurodevelopmental disorder with seizures, hypotonia, and brain imaging abnormalities, 618922 (3), Autosomal recessive
<i>GRN</i>	138945	Ceroid lipofuscinosis, neuronal, 11, 614706 (3), Autosomal recessive; Aphasia, primary progressive, 607485 (3), Autosomal dominant; Frontotemporal lobar degeneration with ubiquitin-positive inclusions, 607485 (3), Autosomal dominant
<i>GRXCR1</i>	613283	Deafness, autosomal recessive 25, 613285 (3), Autosomal recessive
<i>GRXCR2</i>	615762	?Deafness, autosomal recessive 101, 615837 (3), Autosomal recessive
<i>GSC</i>	138890	Short stature, auditory canal atresia, mandibular hypoplasia, skeletal abnormalities, 602471 (3), Autosomal recessive
<i>GSDME</i>	608798	Deafness, autosomal dominant 5, 600994 (3), Autosomal dominant
<i>GSN</i>	137350	Amyloidosis, Finnish type, 105120 (3), Autosomal dominant
<i>GSR</i>	138300	Hemolytic anemia due to glutathione reductase deficiency, 618660 (3), Autosomal recessive
<i>GSS</i>	601002	Glutathione synthetase deficiency, 266130 (3), Autosomal recessive; Hemolytic anemia due to glutathione synthetase deficiency, 231900 (3), Autosomal recessive
<i>GSTZ1</i>	603758	[Maleylacetoacetate isomerase deficiency], 617596 (3), Autosomal recessive
<i>GSX2</i>	616253	Diencephalic-mesencephalic junction dysplasia syndrome 2, 618646 (3), Autosomal recessive
<i>GTF2E2</i>	189964	Trichothiodystrophy 6, nonphotosensitive, 616943 (3), Autosomal recessive
<i>GTF2H5</i>	608780	Trichothiodystrophy 3, photosensitive, 616395 (3)
<i>GTPBP2</i>	607434	Jaberi-Elahi syndrome, 617988 (3), Autosomal recessive
<i>GTPBP3</i>	608536	Combined oxidative phosphorylation deficiency 23, 616198 (3), Autosomal recessive
<i>GUCA1A</i>	600364	Cone-rod dystrophy 14, 602093 (3), Autosomal dominant; Cone dystrophy-3, 602093 (3), Autosomal dominant
<i>GUCA1B</i>	602275	Retinitis pigmentosa 48, 613827 (3)
<i>GUCY1A1</i>	139396	Moyamoya 6 with achalasia, 615750 (3), Autosomal recessive
<i>GUCY2C</i>	601330	Diarrhea 6, 614616 (3), Autosomal dominant; Meconium ileus, 614665 (3), Autosomal recessive
<i>GUCY2D</i>	600179	Cone-rod dystrophy 6, 601777 (3), Autosomal recessive, Autosomal dominant; Leber congenital amaurosis 1, 204000 (3), Autosomal recessive; Night blindness, congenital stationary, type 1I, 618555 (3), Autosomal recessive; ?Choroidal dystrophy, central areolar 1, 215500 (3), Autosomal dominant
<i>GUF1</i>	617064	?Epileptic encephalopathy, early infantile, 40, 617065 (3), Autosomal recessive

<i>GUSB</i>	611499	Mucopolysaccharidosis VII, 253220 (3), Autosomal recessive
<i>GYG1</i>	603942	?Glycogen storage disease XV, 613507 (3), Autosomal recessive; Polyglucosan body myopathy 2, 616199 (3), Autosomal recessive
<i>GYPA</i>	617922	[Blood group, MNSs system], 111300 (3); {Malaria, resistance to}, 611162 (3)
<i>GYPB</i>	617923	{Malaria, resistance to}, 611162 (3); [Blood group, Ss], 111740 (3)
<i>GYPC</i>	110750	{Malaria, resistance to}, 611162 (3); [Blood group, Gerbich], 616089 (3)
<i>GYS1</i>	138570	Glycogen storage disease 0, muscle, 611556 (3), Autosomal recessive
<i>GYS2</i>	138571	Glycogen storage disease 0, liver, 240600 (3), Autosomal recessive
<i>GZF1</i>	613842	Joint laxity, short stature, and myopia, 617662 (3), Autosomal recessive
<i>H1-4 (HIST1H1E)</i>	142220	Rahman syndrome, 617537 (3), Autosomal dominant
<i>H6PD</i>	138090	Cortisone reductase deficiency 1, 604931 (3), Autosomal recessive
<i>HAAO</i>	604521	Vertebral, cardiac, renal, and limb defects syndrome 1, 617660 (3), Autosomal recessive
<i>HABP2</i>	603924	{?Thyroid cancer, nonmedullary, 5}, 616535 (3), Autosomal dominant; {Venous thromboembolism, susceptibility to}, 188050 (3), Autosomal dominant
<i>HACE1</i>	610876	Spastic paraplegia and psychomotor retardation with or without seizures, 616756 (3), Autosomal recessive
<i>HADH</i>	601609	3-hydroxyacyl-CoA dehydrogenase deficiency, 231530 (3), Autosomal recessive; Hyperinsulinemic hypoglycemia, familial, 4, 609975 (3), Autosomal recessive
<i>HADHA</i>	600890	LCHAD deficiency, 609016 (3), Autosomal recessive; HELLP syndrome, maternal, of pregnancy, 609016 (3), Autosomal recessive; Mitochondrial trifunctional protein deficiency, 609015 (3), Autosomal recessive; Fatty liver, acute, of pregnancy, 609016 (3), Autosomal recessive
<i>HADHB</i>	143450	Trifunctional protein deficiency, 609015 (3), Autosomal recessive
<i>HAGH</i>	138760	[Glyoxalase II deficiency], 614033 (1), Autosomal dominant
<i>HAL</i>	609457	[Histidinemia], 235800 (3), Autosomal recessive, Autosomal dominant
<i>HAMP</i>	606464	Hemochromatosis, type 2B, 613313 (3), Autosomal recessive
<i>HARS1 (HARS)</i>	142810	Charcot-Marie-Tooth disease, axonal, type 2W, 616625 (3), Autosomal dominant; Usher syndrome type 3B, 614504 (3), Autosomal recessive
<i>HARS2</i>	600783	?Perrault syndrome 2, 614926 (3), Autosomal recessive
<i>HAVCR2</i>	606652	T-cell lymphoma, subcutaneous panniculitis-like, 618398 (3), Autosomal recessive

<i>HAX1</i>	605998	Neutropenia, severe congenital 3, autosomal recessive, 610738 (3), Autosomal recessive
<i>HBA1</i>	141800	Hemoglobin H disease, nondeletional, 613978 (3); Thalassemias, alpha-, 604131 (3); Erythrocytosis, 7, 617981 (3); Methemoglobinemia, alpha type, 617973 (3); Heinz body anemias, alpha-, 140700 (3), Autosomal dominant
<i>HBA2</i>	141850	Thalassemia, alpha-, 604131 (3); Erythrocytosis 7, 617981 (3); Heinz body anemia, 140700 (3), Autosomal dominant; Hemoglobin H disease, deletional and nondeletional, 613978 (3)
<i>HBB</i>	141900	Thalassemia, beta, 613985 (3); Methemoglobinemia, beta type, 617971 (3); Erythrocytosis 6, 617980 (3); Heinz body anemia, 140700 (3), Autosomal dominant; Delta-beta thalassemia, 141749 (3), Autosomal dominant; Thalassemia-beta, dominant inclusion-body, 603902 (3); Hereditary persistence of fetal hemoglobin, 141749 (3), Autosomal dominant; {Malaria, resistance to}, 611162 (3); Sickle cell anemia, 603903 (3), Autosomal recessive
<i>HBD</i>	142000	Thalassemia, delta- (3); Thalassemia due to Hb Lepore (3)
<i>HBEGF</i>	126150	{Diphtheria, susceptibility to} (1)
<i>HBG1</i>	142200	Fetal hemoglobin quantitative trait locus 1, 141749 (3), Autosomal dominant
<i>HBG2</i>	142250	Fetal hemoglobin quantitative trait locus 1, 141749 (3), Autosomal dominant; Cyanosis, transient neonatal, 613977 (3), Autosomal dominant
<i>HCCS</i>	300056	Linear skin defects with multiple congenital anomalies 1, 309801 (3), X-linked dominant
<i>HCFC1</i>	300019	Mental retardation, X-linked 3 (methylmalonic acidemia and homocysteinemia, cblX type), 309541 (3), X-linked recessive
<i>HCN1</i>	602780	Generalized epilepsy with febrile seizures plus, type 10, 618482 (3), Autosomal dominant; Epileptic encephalopathy, early infantile, 24, 615871 (3), Autosomal dominant
<i>HCN4</i>	605206	Brugada syndrome 8, 613123 (3); Sick sinus syndrome 2, 163800 (3), Autosomal dominant
<i>HCRT</i>	602358	?Narcolepsy 1, 161400 (3), Autosomal dominant
<i>HDAC4</i>	605314/600430	-/Chromosome 2q37 deletion syndrome, 600430 (4), Autosomal dominant
<i>HDAC6</i>	300272	?Chondrodysplasia with platyspondyly, distinctive brachydactyly, hydrocephaly, and microphthalmia, 300863 (3), X-linked dominant
<i>HDAC8</i>	300269	Cornelia de Lange syndrome 5, 300882 (3), X-linked dominant
<i>HDC</i>	142704	{Gilles de la Tourette syndrome, susceptibility to}, 137580 (3), Autosomal dominant
<i>HECW2</i>	617245	Neurodevelopmental disorder with hypotonia, seizures, and absent language, 617268 (3), Autosomal dominant
<i>HELLS</i>	603946	Immunodeficiency-centromeric instability-facial anomalies syndrome 4, 616911 (3), Autosomal recessive

<i>HEPACAM</i>	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
<i>HEPHL1</i>	618455	?Abnormal hair, joint laxity, and developmental delay, 261990 (3), Autosomal recessive
<i>HERC1</i>	605109	Macrocephaly, dysmorphic facies, and psychomotor retardation, 617011 (3), Autosomal recessive
<i>HERC2</i>	605837	[Skin/hair/eye pigmentation 1, blue/nonblue eyes], 227220 (3), Autosomal recessive; Mental retardation, autosomal recessive 38, 615516 (3), Autosomal recessive; [Skin/hair/eye pigmentation 1, blond/brown hair], 227220 (3), Autosomal recessive
<i>HES7</i>	608059	Spondylocostal dysostosis 4, autosomal recessive, 613686 (3), Autosomal recessive
<i>HESX1</i>	601802	Pituitary hormone deficiency, combined, 5, 182230 (3), Autosomal recessive, Autosomal dominant; Septo-optic dysplasia, 182230 (3), Autosomal recessive, Autosomal dominant; Growth hormone deficiency with pituitary anomalies, 182230 (3), Autosomal recessive, Autosomal dominant
<i>HEXA</i>	606869	GM2-gangliosidosis, several forms, 272800 (3), Autosomal recessive; Tay-Sachs disease, 272800 (3), Autosomal recessive; [Hex A pseudodeficiency], 272800 (3), Autosomal recessive
<i>HEXB</i>	606873	Sandhoff disease, infantile, juvenile, and adult forms, 268800 (3), Autosomal recessive
<i>HFE</i>	613609	{Porphyria variegata, susceptibility to}, 176200 (3), Autosomal dominant; {Microvascular complications of diabetes 7}, 612635 (3); {Porphyria cutanea tarda, susceptibility to}, 176100 (3), Autosomal recessive, Autosomal dominant; [Transferrin serum level QTL2], 614193 (3); {Alzheimer disease, susceptibility to}, 104300 (3), Autosomal dominant; Hemochromatosis, 235200 (3), Autosomal recessive
<i>HFM1</i>	615684	Premature ovarian failure 9, 615724 (3), Autosomal recessive
<i>HGD</i>	607474	Alkaptonuria, 203500 (3), Autosomal recessive
<i>HGF</i>	142409	Deafness, autosomal recessive 39, 608265 (3), Autosomal recessive
<i>HGSNAT</i>	610453	Retinitis pigmentosa 73, 616544 (3), Autosomal recessive; Mucopolysaccharidosis type IIIC (Sanfilippo C), 252930 (3), Autosomal recessive
<i>HIBCH</i>	610690	3-hydroxyisobutryl-CoA hydrolase deficiency, 250620 (3), Autosomal recessive
<i>HIKESHI</i>	614908	Leukodystrophy, hypomyelinating, 13, 616881 (3), Autosomal recessive
<i>HINT1</i>	601314	Neuromyotonia and axonal neuropathy, autosomal recessive, 137200 (3), Autosomal recessive

<i>HIVEP2</i>	143054	Mental retardation, autosomal dominant 43, 616977 (3), Autosomal dominant
<i>HJV</i>	608374	Hemochromatosis, type 2A, 602390 (3), Autosomal recessive
<i>HK1</i>	142600	Hemolytic anemia due to hexokinase deficiency, 235700 (3), Autosomal recessive; Neuropathy, hereditary motor and sensory, Russe type, 605285 (3), Autosomal recessive; Neurodevelopmental disorder with visual defects and brain anomalies, 618547 (3), Autosomal dominant; Retinitis pigmentosa 79, 617460 (3), Autosomal dominant
<i>HLA-A</i>	142800	{Hypersensitivity syndrome, carbamazepine-induced, susceptibility to}, 608579 (3)
<i>HLA-B</i>	142830	{Stevens-Johnson syndrome, susceptibility to}, 608579 (3); {Spondyloarthritis, susceptibility to, 1}, 106300 (3), Multifactorial; {Abacavir hypersensitivity, susceptibility to} (3); {Drug-induced liver injury due to flucloxacillin} (3); {Synovitis, chronic, susceptibility to} (3); {Toxic epidermal necrolysis, susceptibility to}, 608579 (3)
<i>HLA-C</i>	142840	{Psoriasis susceptibility 1}, 177900 (3), Multifactorial; {HIV-1 viremia, susceptibility to}, 609423 (3)
<i>HLA-DPB1</i>	142858	{Beryllium disease, chronic, susceptibility to} (3)
<i>HLA-DQA1</i>	146880	{Celiac disease, susceptibility to}, 212750 (3), Autosomal recessive, Multifactorial
<i>HLA-DQB1</i>	604305	{Multiple sclerosis, susceptibility to, 1}, 126200 (3), Multifactorial; {Celiac disease, susceptibility to}, 212750 (3), Autosomal recessive, Multifactorial; {Creutzfeldt-Jakob disease, variant, resistance to}, 123400 (3), Autosomal dominant
<i>HLA-DRB1</i>	142857	{Sarcoidosis, susceptibility to, 1}, 181000 (3), Autosomal dominant; {Multiple sclerosis, susceptibility to, 1}, 126200 (3), Multifactorial
<i>HLA-G</i>	142871	{Asthma, susceptibility to}, 600807 (2), Autosomal dominant
<i>HLCS</i>	609018	Holocarboxylase synthetase deficiency, 253270 (3), Autosomal recessive
<i>HMBS</i>	609806	Porphyria, acute intermittent, 176000 (3), Autosomal dominant; Porphyria, acute intermittent, nonerythroid variant, 176000 (3), Autosomal dominant
<i>HMCN1</i>	608548	{Macular degeneration, age-related, 1}, 603075 (3), Autosomal dominant
<i>HMGA1</i>	600701	{Diabetes mellitus, noninsulin-dependent, susceptibility to}, 125853 (3), Autosomal dominant
<i>HMGA2</i>	600698	Silver-Russell syndrome 5, 618908 (3), Autosomal dominant
<i>HMGB3</i>	300193	?Microphthalmia, syndromic 13, 300915 (3), X-linked
<i>HMGCL</i>	613898	HMG-CoA lyase deficiency, 246450 (3), Autosomal recessive
<i>HMGCR</i>	142910	[Statins, attenuated cholesterol lowering by] (3); [Low density lipoprotein cholesterol level QTL 3] (3)
<i>HMGCS2</i>	600234	HMG-CoA synthase-2 deficiency, 605911 (3), Autosomal recessive

<i>HMMR</i>	600936	{Breast cancer, susceptibility to}, 114480 (3), Autosomal dominant, Somatic mutation
<i>HMOX1</i>	141250	{Pulmonary disease, chronic obstructive, susceptibility to}, 606963 (3); Heme oxygenase-1 deficiency, 614034 (3)
<i>HMX1</i>	142992	Oculoauricular syndrome, 612109 (3), Autosomal recessive
<i>HNF1A</i>	142410	{Diabetes mellitus, insulin-dependent}, 222100 (3), Autosomal recessive; MODY, type III, 600496 (3), Autosomal dominant; Hepatic adenoma, somatic, 142330 (3); Renal cell carcinoma, 144700 (3); Diabetes mellitus, insulin-dependent, 20, 612520 (3); {Diabetes mellitus, noninsulin-dependent, 2}, 125853 (3), Autosomal dominant
<i>HNF1B</i>	189907	Diabetes mellitus, noninsulin-dependent, 125853 (3), Autosomal dominant; Renal cysts and diabetes syndrome, 137920 (3), Autosomal dominant; {Renal cell carcinoma}, 144700 (3)
<i>HNF4A</i>	600281	{Diabetes mellitus, noninsulin-dependent}, 125853 (3), Autosomal dominant; MODY, type I, 125850 (3), Autosomal dominant; Fanconi renotubular syndrome 4, with maturity-onset diabetes of the young, 616026 (3), Autosomal dominant
<i>HNMT</i>	605238	{Asthma, susceptibility to}, 600807 (3), Autosomal dominant; Mental retardation, autosomal recessive 51, 616739 (3), Autosomal recessive
<i>HNRNPA1</i>	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
<i>HNRNPA2B1</i>	600124	?Inclusion body myopathy with early-onset Paget disease with or without frontotemporal dementia 2, 615422 (3)
<i>HNRNPDL</i>	607137	Muscular dystrophy, limb-girdle, autosomal dominant 3, 609115 (3), Autosomal dominant
<i>HNRNPH2</i>	300610	Mental retardation, X-linked, syndromic, Bain type, 300986 (3), X-linked dominant
<i>HNRNPK</i>	600712	Au-Kline syndrome, 616580 (3), Autosomal dominant
<i>HNRNPU</i>	602869	Epileptic encephalopathy, early infantile, 54, 617391 (3), Autosomal dominant
<i>HOGA1</i>	613597	Hyperoxaluria, primary, type III, 613616 (3), Autosomal recessive
<i>HOMER2</i>	604799	?Deafness, autosomal dominant 68, 616707 (3), Autosomal dominant
<i>HOXA1</i>	142955	Athabaskan brainstem dysgenesis syndrome, 601536 (3); Bosley-Salih-Alorainy syndrome, 601536 (3)
<i>HOXA11</i>	142958	Radioulnar synostosis with amegakaryocytic thrombocytopenia 1, 605432 (3), Autosomal dominant
<i>HOXA13</i>	142959	?Guttmacher syndrome, 176305 (3), Autosomal dominant; Hand-foot-uterus syndrome, 140000 (3), Autosomal dominant

<i>HOXA2</i>	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
<i>HOXB1</i>	142968	Facial paresis, hereditary congenital, 3, 614744 (3), Autosomal recessive
<i>HOXB13</i>	604607	{Prostate cancer, hereditary, 9}, 610997 (3)
<i>HOXC13</i>	142976	Ectodermal dysplasia 9, hair/nail type, 614931 (3), Autosomal recessive
<i>HOXD10</i>	142984	Charcot-Marie-Tooth disease, foot deformity of, 192950 (3), Autosomal dominant; Vertical talus, congenital, 192950 (3), Autosomal dominant
<i>HOXD13</i>	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
<i>HP</i>	140100	[Hypohaptoglobinemia], 614081 (3); [Anhaptoglobinemia], 614081 (3)
<i>HPCA</i>	142622	Dystonia 2, torsion, autosomal recessive, 224500 (3), Autosomal recessive
<i>HPD</i>	609695	Tyrosinemia, type III, 276710 (3), Autosomal recessive; Hawkinsinuria, 140350 (3), Autosomal dominant
<i>HPGD</i>	601688	Hypertrophic osteoarthropathy, primary, autosomal recessive 1, 259100 (3), Autosomal recessive; ?Digital clubbing, isolated congenital, 119900 (3), Autosomal recessive; Cranioosteoarthropathy, 259100 (3), Autosomal recessive
<i>HPRT1</i>	308000	HPRT-related gout, 300323 (3), X-linked recessive; Lesch-Nyhan syndrome, 300322 (3), X-linked recessive
<i>HPS1</i>	604982	Hermansky-Pudlak syndrome 1, 203300 (3), Autosomal recessive
<i>HPS3</i>	606118	Hermansky-Pudlak syndrome 3, 614072 (3), Autosomal recessive
<i>HPS4</i>	606682	Hermansky-Pudlak syndrome 4, 614073 (3), Autosomal recessive
<i>HPS5</i>	607521	Hermansky-Pudlak syndrome 5, 614074 (3), Autosomal recessive
<i>HPS6</i>	607522	Hermansky-Pudlak syndrome 6, 614075 (3), Autosomal recessive
<i>HPSE2</i>	613469	Urofacial syndrome 1, 236730 (3), Autosomal recessive
<i>HR</i>	602302	Hypotrichosis 4, 146550 (3), Autosomal dominant; Alopecia universalis, 203655 (3), Autosomal recessive; Atrichia with papular lesions, 209500 (3), Autosomal recessive
<i>HRAS</i>	190020	Nevus sebaceous or woolly hair nevus, somatic, 162900 (3); Congenital myopathy with excess of muscle spindles, 218040 (3), Autosomal dominant; Bladder cancer, somatic, 109800 (3); Thyroid carcinoma, follicular, somatic, 188470 (3); Schimmelpenning-Feuerstein-Mims syndrome, somatic mosaic, 163200 (3); Spitz nevus or nevus spilus, somatic, 137550 (3); Costello syndrome, 218040 (3), Autosomal dominant

<i>HRG</i>	142640	Thrombophilia due to HRG deficiency, 613116 (3), Autosomal dominant
<i>HS6ST1</i>	604846	{Hypogonadotropic hypogonadism 15 with or without anosmia}, 614880 (3), Autosomal dominant
<i>HS6ST2</i>	300545	?Paganini-Miozzo syndrome, 301025 (3), X-linked recessive
<i>HSD11B1</i>	600713	Cortisone reductase deficiency 2, 614662 (3), Autosomal dominant
<i>HSD11B2</i>	614232	Apparent mineralocorticoid excess, 218030 (3), Autosomal recessive
<i>HSD17B10</i>	300256	HSD10 mitochondrial disease, 300438 (3), X-linked dominant
<i>HSD17B3</i>	605573	Pseudohermaphroditism, male, with gynecomastia, 264300 (3), Autosomal recessive
<i>HSD17B4</i>	601860	D-bifunctional protein deficiency, 261515 (3), Autosomal recessive; Perrault syndrome 1, 233400 (3), Autosomal recessive
<i>HSD3B2</i>	613890	Adrenal hyperplasia, congenital, due to 3-beta-hydroxysteroid dehydrogenase 2 deficiency, 201810 (3), Autosomal recessive
<i>HSD3B7</i>	607764	Bile acid synthesis defect, congenital, 1, 607765 (3), Autosomal recessive
<i>HSF4</i>	602438	Cataract 5, multiple types, 116800 (3), Autosomal dominant
<i>HSPA9</i>	600548	Even-plus syndrome, 616854 (3), Autosomal recessive; Anemia, sideroblastic, 4, 182170 (3), Autosomal dominant
<i>HSPB1</i>	602195	Neuronopathy, distal hereditary motor, type IIB, 608634 (3), Autosomal dominant; Charcot-Marie-Tooth disease, axonal, type 2F, 606595 (3), Autosomal dominant
<i>HSPB3</i>	604624	?Neuronopathy, distal hereditary motor, type IIC, 613376 (3), Autosomal dominant
<i>HSPB8</i>	608014	Neuronopathy, distal hereditary motor, type IIA, 158590 (3), Autosomal dominant; Charcot-Marie-Tooth disease, axonal, type 2L, 608673 (3), Autosomal dominant
<i>HSPD1</i>	118190	Spastic paraplegia 13, autosomal dominant, 605280 (3), Autosomal dominant; Leukodystrophy, hypomyelinating, 4, 612233 (3), Autosomal recessive
<i>HSPG2</i>	142461	Dyssegmental dysplasia, Silverman-Handmaker type, 224410 (3), Autosomal recessive; Schwartz-Jampel syndrome, type 1, 255800 (3), Autosomal recessive
<i>HTR1A</i>	109760	Periodic fever, menstrual cycle dependent, 614674 (3), Autosomal dominant
<i>HTR2A</i>	182135	{Major depressive disorder, response to citalopram therapy in}, 608516 (3); {Alcohol dependence, susceptibility to}, 103780 (3), Multifactorial; {Schizophrenia, susceptibility to}, 181500 (3), Autosomal dominant; {Anorexia nervosa, susceptibility to}, 606788 (3); {Obsessive-compulsive disorder, susceptibility to}, 164230 (3), Autosomal dominant; {Seasonal affective disorder, susceptibility to}, 608516 (3)

<i>HTRA1</i>	602194	{Macular degeneration, age-related, neovascular type}, 610149 (3); {Macular degeneration, age-related, 7}, 610149 (3); Cerebral arteriopathy, autosomal dominant, with subcortical infarcts and leukoencephalopathy, type 2, 616779 (3), Autosomal dominant; CARASIL syndrome, 600142 (3), Autosomal recessive
<i>HTRA2</i>	606441	{Parkinson disease 13}, 610297 (3); 3-methylglutaconic aciduria, type VIII, 617248 (3), Autosomal recessive
<i>HTT</i>	613004	Huntington disease, 143100 (3), Autosomal dominant; Lopes-Maciél-Rodan syndrome, 617435 (3), Autosomal recessive
<i>HUWE1</i>	300697	Mental retardation, X-linked syndromic, Turner type, 309590 (3), X-linked
<i>HYAL1</i>	607071	?Mucopolysaccharidosis type IX, 601492 (3), Autosomal recessive
<i>HYDIN</i>	610812	Ciliary dyskinesia, primary, 5, 608647 (3), Autosomal recessive
<i>HYLS1</i>	610693	Hydrolethalus syndrome, 236680 (3), Autosomal recessive
<i>HYOU1</i>	601746	?Immunodeficiency 59 and hypoglycemia, 233600 (3), Autosomal recessive
<i>IARS1 (IARS)</i>	600709	Growth retardation, impaired intellectual development, hypotonia, and hepatopathy, 617093 (3), Autosomal recessive
<i>IARS2</i>	612801	?Cataracts, growth hormone deficiency, sensory neuropathy, sensorineural hearing loss, and skeletal dysplasia, 616007 (3), Autosomal recessive
<i>IBA57</i>	615316	?Spastic paraplegia 74, autosomal recessive, 616451 (3), Autosomal recessive; Multiple mitochondrial dysfunctions syndrome 3, 615330 (3), Autosomal recessive
<i>ICAM1</i>	147840	{Malaria, cerebral, susceptibility to}, 611162 (3)
<i>ICAM4</i>	614088	[Blood group, Landsteiner-Wiener], 111250 (3)
<i>ICOS</i>	604558	Immunodeficiency, common variable, 1, 607594 (3), Autosomal recessive
<i>IDH1</i>	147700	{Glioma, susceptibility to, somatic}, 137800 (3)
<i>IDH2</i>	147650	D-2-hydroxyglutaric aciduria 2, 613657 (3)
<i>IDH3B</i>	604526	Retinitis pigmentosa 46, 612572 (3)
<i>IDS</i>	300823	Mucopolysaccharidosis II, 309900 (3), X-linked recessive
<i>IDUA</i>	252800	Mucopolysaccharidosis Ih/s, 607015 (3), Autosomal recessive; Mucopolysaccharidosis Ih, 607014 (3), Autosomal recessive; Mucopolysaccharidosis Is, 607016 (3), Autosomal recessive
<i>IER3IP1</i>	609382	Microcephaly, epilepsy, and diabetes syndrome, 614231 (3), Autosomal recessive
<i>IFIH1</i>	606951	Aicardi-Goutières syndrome 7, 615846 (3), Autosomal dominant; Singleton-Merten syndrome 1, 182250 (3), Autosomal dominant
<i>IFITM3</i>	605579	{Influenza, severe, susceptibility to}, 614680 (3)
<i>IFITM5</i>	614757	Osteogenesis imperfecta, type V, 610967 (3), Autosomal dominant
<i>IFNA1</i>	147660	Interferon, alpha, deficiency (1)

<i>IFNAR2</i>	602376	?Immunodeficiency 45, 616669 (3), Autosomal recessive; {Hepatitis B virus, susceptibility to}, 610424 (3)
<i>IFNG</i>	147570	?Immunodeficiency 69, mycobacteriosis, 618963 (3), Autosomal recessive; {AIDS, rapid progression to}, 609423 (3); {TSC2 angiomyolipomas, renal, modifier of}, 613254 (3), Autosomal dominant; {Hepatitis C virus, response to therapy of}, 609532 (3); {Aplastic anemia}, 609135 (3); {Tuberculosis, protection against}, 607948 (3)
<i>IFNGR1</i>	107470	Immunodeficiency 27A, mycobacteriosis, AR, 209950 (3), Autosomal recessive; {Tuberculosis infection, protection against}, 607948 (3); {Tuberculosis, susceptibility to}, 607948 (3); Immunodeficiency 27B, mycobacteriosis, AD, 615978 (3), Autosomal dominant; {H. pylori infection, susceptibility to}, 600263 (3); {Hepatitis B virus infection, susceptibility to}, 610424 (3)
<i>IFNGR2</i>	147569	Immunodeficiency 28, mycobacteriosis, 614889 (3), Autosomal recessive
<i>IFNL3</i>	607402	{Hepatitis C virus infection, response to therapy of}, 609532 (3)
<i>IFT122</i>	606045	Cranioectodermal dysplasia 1, 218330 (3), Autosomal recessive
<i>IFT140</i>	614620	Retinitis pigmentosa 80, 617781 (3), Autosomal recessive; Short-rib thoracic dysplasia 9 with or without polydactyly, 266920 (3), Autosomal recessive
<i>IFT172</i>	607386	Retinitis pigmentosa 71, 616394 (3), Autosomal recessive; Short-rib thoracic dysplasia 10 with or without polydactyly, 615630 (3), Autosomal recessive
<i>IFT27</i>	615870	?Bardet-Biedl syndrome 19, 615996 (3), Autosomal recessive
<i>IFT43</i>	614068	?Cranioectodermal dysplasia 3, 614099 (3), Autosomal recessive; Short-rib thoracic dysplasia 18 with polydactyly, 617866 (3), Autosomal recessive; ?Retinitis pigmentosa 81, 617871 (3), Autosomal recessive
<i>IFT52</i>	617094	Short-rib thoracic dysplasia 16 with or without polydactyly, 617102 (3), Autosomal recessive
<i>IFT57</i>	606621	?Orofaciodigital syndrome XVIII, 617927 (3), Autosomal recessive
<i>IFT74</i>	608040	?Bardet-Biedl syndrome 20, 617119 (3), Autosomal recessive
<i>IFT80</i>	611177	Short-rib thoracic dysplasia 2 with or without polydactyly, 611263 (3), Autosomal recessive
<i>IFT81</i>	605489	Short-rib thoracic dysplasia 19 with or without polydactyly, 617895 (3), Autosomal recessive
<i>IGBP1</i>	300139	Corpus callosum, agenesis of, with mental retardation, ocular coloboma and micrognathia, 300472 (3), X-linked recessive
<i>IGF1</i>	147440	Growth retardation with deafness and mental retardation due to IGF1 deficiency, 608747 (3), Autosomal recessive
<i>IGF1R</i>	147370	Insulin-like growth factor I, resistance to, 270450 (3), Autosomal recessive, Autosomal dominant
<i>IGF2</i>	147470	Silver-Russell syndrome 3, 616489 (3), Autosomal dominant

<i>IGF2BP2</i>	608289	{Diabetes mellitus, noninsulin-dependent, susceptibility to}, 125853 (3), Autosomal dominant
<i>IGF2R</i>	147280	Hepatocellular carcinoma, somatic, 114550 (3)
<i>IGFALS</i>	601489	Acid-labile subunit, deficiency of, 615961 (3)
<i>IGFBP7</i>	602867	Retinal arterial macroaneurysm with supravalvular pulmonic stenosis, 614224 (3), Autosomal recessive
<i>IGHG2</i>	147110	IgG2 deficiency, selective (3)
<i>IGHM</i>	147020	Agammaglobulinemia 1, 601495 (3), Autosomal recessive
<i>IGHMBP2</i>	600502	Neuronopathy, distal hereditary motor, type VI, 604320 (3), Autosomal recessive; Charcot-Marie-Tooth disease, axonal, type 2S, 616155 (3), Autosomal recessive
<i>IGKC</i>	147200	Kappa light chain deficiency, 614102 (3), Autosomal recessive
<i>IGLL1</i>	146770	Agammaglobulinemia 2, 613500 (3), Autosomal recessive
<i>IGSF1</i>	300137	Hypothyroidism, central, and testicular enlargement, 300888 (3), X-linked recessive
<i>IGSF3</i>	603491	?Lacrimal duct defect, 149700 (3), Autosomal recessive
<i>IHH</i>	600726	Acrocapitofemoral dysplasia, 607778 (3), Autosomal recessive; Brachydactyly, type A1, 112500 (3), Autosomal dominant
<i>IKBKB</i>	603258	Immunodeficiency 15A, 618204 (3), Autosomal dominant; Immunodeficiency 15B, 615592 (3), Autosomal recessive
<i>IKBKG</i>	300248	Immunodeficiency 33, 300636 (3), X-linked recessive; Incontinentia pigmenti, 308300 (3), X-linked dominant; Ectodermal dysplasia and immunodeficiency 1, 300291 (3), X-linked recessive
<i>IKZF1</i>	603023	Immunodeficiency, common variable, 13, 616873 (3), Autosomal dominant
<i>IL10</i>	124092	{Graft-versus-host disease, protection against}, 614395 (3); {HIV-1, susceptibility to}, 609423 (3); {Rheumatoid arthritis, progression of}, 180300 (3)
<i>IL10RA</i>	146933	Inflammatory bowel disease 28, early onset, autosomal recessive, 613148 (3), Autosomal recessive
<i>IL10RB</i>	123889	{Hepatitis B virus, susceptibility to}, 610424 (3); Inflammatory bowel disease 25, early onset, autosomal recessive, 612567 (3), Autosomal recessive
<i>IL11RA</i>	600939	Craniosynostosis and dental anomalies, 614188 (3), Autosomal recessive
<i>IL12B</i>	161561	Immunodeficiency 29, mycobacteriosis, 614890 (3), Autosomal recessive
<i>IL12RB1</i>	601604	Immunodeficiency 30, 614891 (3), Autosomal recessive
<i>IL13</i>	147683	{Allergic rhinitis, susceptibility to}, 607154 (3); {Asthma, susceptibility to}, 600807 (3), Autosomal dominant
<i>IL17F</i>	606496	?Candidiasis, familial, 6, autosomal dominant, 613956 (3)
<i>IL17RA</i>	605461	Immunodeficiency 51, 613953 (3), Autosomal recessive
<i>IL17RC</i>	610925	Candidiasis, familial, 9, 616445 (3), Autosomal recessive

<i>IL17RD</i>	606807	Hypogonadotropic hypogonadism 18 with or without anosmia, 615267 (3), Autosomal recessive, Digenic dominant, Autosomal dominant
<i>IL18BP</i>	604113	{?Hepatitis, fulminant viral, susceptibility to}, 618549 (3), Autosomal recessive
<i>IL1B</i>	147720	{Gastric cancer risk after H. pylori infection}, 137215 (3), Autosomal dominant
<i>IL1RAPL1</i>	300206	Mental retardation, X-linked 21/34, 300143 (3), X-linked recessive
<i>IL1RN</i>	147679	{Gastric cancer risk after H. pylori infection}, 137215 (3), Autosomal dominant; {Microvascular complications of diabetes 4}, 612628 (3); Interleukin 1 receptor antagonist deficiency, 612852 (3), Autosomal recessive
<i>IL21</i>	605384	?Immunodeficiency, common variable, 11, 615767 (3), Autosomal recessive
<i>IL21R</i>	605383	[IgE, elevated level of], 147050 (3), Autosomal dominant; Immunodeficiency 56, 615207 (3), Autosomal recessive
<i>IL23R</i>	607562	{Inflammatory bowel disease 17, protection against}, 612261 (3); {Psoriasis, protection against}, 605606 (3)
<i>IL2RA</i>	147730	Immunodeficiency 41 with lymphoproliferation and autoimmunity, 606367 (3), Autosomal recessive; {Diabetes, mellitus, insulin-dependent, susceptibility to, 10}, 601942 (3)
<i>IL2RB</i>	146710	Immunodeficiency 63 with lymphoproliferation and autoimmunity, 618495 (3), Autosomal recessive
<i>IL2RG</i>	308380	Severe combined immunodeficiency, X-linked, 300400 (3), X-linked recessive; Combined immunodeficiency, X-linked, moderate, 312863 (3), X-linked recessive
<i>IL31RA</i>	609510	?Amyloidosis, primary localized cutaneous, 2, 613955 (3), Autosomal dominant
<i>IL36RN</i>	605507	Psoriasis 14, pustular, 614204 (3), Autosomal recessive
<i>IL4R</i>	147781	{Atopy, susceptibility to}, 147050 (3), Autosomal dominant; {AIDS, slow progression to}, 609423 (3)
<i>IL6</i>	147620	{Intracranial hemorrhage in brain cerebrovascular malformations, susceptibility to}, 108010 (3), Somatic mutation; {Rheumatoid arthritis, systemic juvenile}, 604302 (3); {Diabetes, susceptibility to}, 222100 (3), Autosomal recessive; {Kaposi sarcoma, susceptibility to}, 148000 (3), Autosomal dominant; {Crohn disease-associated growth failure}, 266600 (3), Multifactorial
<i>IL6R</i>	147880	[Interleukin-6 receptor, soluble, serum level of, QTL], 614689 (3); [Interleukin 6, serum level of, QTL], 614752 (3); Hyper-IgE recurrent infection syndrome 5, autosomal recessive, 618944 (3), Autosomal recessive
<i>IL6ST</i>	600694	Hyper-IgE recurrent infection syndrome 4, autosomal recessive, 618523 (3), Autosomal recessive

<i>IL7</i>	146660	{?Epidermodysplasia verruciformis, susceptibility to, 5}, 618309 (3), Autosomal recessive
<i>IL7R</i>	146661	Severe combined immunodeficiency, T-cell negative, B-cell/natural killer cell-positive type, 608971 (3), Autosomal recessive
<i>ILDR1</i>	609739	Deafness, autosomal recessive 42, 609646 (3), Autosomal recessive
<i>IMPA1</i>	602064	Mental retardation, autosomal recessive 59, 617323 (3), Autosomal recessive
<i>IMPDH1</i>	146690	Leber congenital amaurosis 11, 613837 (3), Autosomal dominant; Retinitis pigmentosa 10, 180105 (3), Autosomal dominant
<i>IMPDH2</i>	146691	[IMPDH2 enzyme activity, variation in], 617995 (3)
<i>IMPG1</i>	602870	Macular dystrophy, vitelliform, 4, 616151 (3), Autosomal dominant
<i>IMPG2</i>	607056	Macular dystrophy, vitelliform, 5, 616152 (3), Autosomal dominant; Retinitis pigmentosa 56, 613581 (3), Autosomal recessive
<i>INAVA</i>	618051	{Inflammatory bowel disease 29}, 618077 (3), Autosomal dominant
<i>INF2</i>	610982	Glomerulosclerosis, focal segmental, 5, 613237 (3); Charcot-Marie-Tooth disease, dominant intermediate E, 614455 (3), Autosomal dominant
<i>ING1</i>	601566	Squamous cell carcinoma, head and neck, somatic, 275355 (3)
<i>INPP5E</i>	613037	Mental retardation, truncal obesity, retinal dystrophy, and micropenis, 610156 (3), Autosomal recessive; Joubert syndrome 1, 213300 (3), Autosomal recessive
<i>INPP5K</i>	607875	Muscular dystrophy, congenital, with cataracts and intellectual disability, 617404 (3), Autosomal recessive
<i>INPPL1</i>	600829	Opsismodysplasia, 258480 (3), Autosomal recessive
<i>INS</i>	176730	Maturity-onset diabetes of the young, type 10, 613370 (3), Autosomal dominant; Hyperproinsulinemia, 616214 (3), Autosomal dominant; Diabetes mellitus, insulin-dependent, 2, 125852 (3), Autosomal dominant; Diabetes mellitus, permanent neonatal, 618858 (3), Autosomal recessive, Autosomal dominant
<i>INSL3</i>	146738	Cryptorchidism, 219050 (3), Autosomal dominant
<i>INSR</i>	147670	Hyperinsulinemic hypoglycemia, familial, 5, 609968 (3), Autosomal dominant; Rabson-Mendenhall syndrome, 262190 (3), Autosomal recessive; Diabetes mellitus, insulin-resistant, with acanthosis nigricans, 610549 (3); Leprechaunism, 246200 (3), Autosomal recessive
<i>INTS1</i>	611345	Neurodevelopmental disorder with cataracts, poor growth, and dysmorphic facies, 618571 (3), Autosomal recessive
<i>INTS8</i>	611351	?Neurodevelopmental disorder with cerebellar hypoplasia and spasticity, 618572 (3), Autosomal recessive

<i>INTU</i>	610621	?Short-rib thoracic dysplasia 20 with polydactyly, 617925 (3), Autosomal recessive; ?Orofaciodigital syndrome XVII, 617926 (3), Autosomal recessive
<i>INVS</i>	243305	Nephronophthisis 2, infantile, 602088 (3), Autosomal recessive
<i>IQCB1</i>	609237	Senior-Loken syndrome 5, 609254 (3), Autosomal recessive
<i>IQCE</i>	617631	?Polydactyly, postaxial, type A7, 617642 (3), Autosomal recessive
<i>IQSEC1</i>	610166	Intellectual developmental disorder with short stature and behavioral abnormalities, 618687 (3), Autosomal recessive
<i>IQSEC2</i>	300522	Mental retardation, X-linked 1/78, 309530 (3), X-linked dominant
<i>IRAK3</i>	604459	{Asthma susceptibility 5}, 611064 (3)
<i>IRAK4</i>	606883	Immunodeficiency 67, 607676 (3), Autosomal recessive
<i>IREB2</i>	147582	Neurodegeneration, early-onset, with choreoathetoid movements and microcytic anemia, 618451 (3), Autosomal recessive
<i>IRF1</i>	147575	Gastric cancer, somatic, 613659 (3); Myelogenous leukemia, acute (3); Myelodysplastic syndrome, preleukemic (3); Nonsmall cell lung cancer, somatic, 211980 (3)
<i>IRF2BP2</i>	615332	?Immunodeficiency, common variable, 14, 617765 (3), Autosomal dominant
<i>IRF2BPL</i>	611720	Neurodevelopmental disorder with regression, abnormal movements, loss of speech, and seizures, 618088 (3), Autosomal dominant
<i>IRF3</i>	603734	{Encephalopathy, acute, infection-induced (herpes-specific), susceptibility to, 7}, 616532 (3), Autosomal dominant
<i>IRF4</i>	601900	[Skin/hair/eye pigmentation, variation in, 8], 611724 (3)
<i>IRF5</i>	607218	{Systemic lupus erythematosus, susceptibility to, 10}, 612251 (3); {Inflammatory bowel disease 14}, 612245 (3)
<i>IRF6</i>	607199	Popliteal pterygium syndrome 1, 119500 (3), Autosomal dominant; {Orofacial cleft 6}, 608864 (3), Autosomal dominant; van der Woude syndrome, 119300 (3), Autosomal dominant
<i>IRF7</i>	605047	?Immunodeficiency 39, 616345 (3), Autosomal recessive
<i>IRF8</i>	601565	Immunodeficiency 32B, monocyte and dendritic cell deficiency, autosomal recessive, 226990 (3), Autosomal recessive; Immunodeficiency 32A, mycobacteriosis, autosomal dominant, 614893 (3), Autosomal dominant
<i>IRF9</i>	147574	Immunodeficiency 65, susceptibility to viral infections, 618648 (3), Autosomal recessive
<i>IRGM</i>	608212	{Mycobacterium tuberculosis, protection against}, 607948 (3); {Inflammatory bowel disease (Crohn disease) 19}, 612278 (3)
<i>IRS1</i>	147545	{Coronary artery disease, susceptibility to} (3); {Diabetes mellitus, noninsulin-dependent}, 125853 (3), Autosomal dominant
<i>IRS2</i>	600797	{Diabetes mellitus, noninsulin-dependent}, 125853 (3), Autosomal dominant

<i>IRS4</i>	300904	Hypothyroidism, congenital, nongoitrous, 9, 301035 (3), X-linked recessive
<i>IRX5</i>	606195	Hamamy syndrome, 611174 (3), Autosomal recessive
<i>ISCA1</i>	611006	Multiple mitochondrial dysfunctions syndrome 5, 617613 (3), Autosomal recessive
<i>ISCA2</i>	615317	Multiple mitochondrial dysfunctions syndrome 4, 616370 (3), Autosomal recessive
<i>ISCU</i>	611911	Myopathy with lactic acidosis, hereditary, 255125 (3), Autosomal recessive
<i>ISG15</i>	147571	Immunodeficiency 38, 616126 (3), Autosomal recessive
<i>ITCH</i>	606409	Autoimmune disease, multisystem, with facial dysmorphism, 613385 (3), Autosomal recessive
<i>ITGA2B</i>	607759	Glanzmann thrombasthenia, 273800 (3), Autosomal recessive; Thrombocytopenia, neonatal alloimmune, BAK antigen related (3); Bleeding disorder, platelet-type, 16, autosomal dominant, 187800 (3), Autosomal dominant
<i>ITGA3</i>	605025	Interstitial lung disease, nephrotic syndrome, and epidermolysis bullosa, congenital, 614748 (3), Autosomal recessive
<i>ITGA6</i>	147556	Epidermolysis bullosa, junctional, with pyloric stenosis, 226730 (3), Autosomal recessive
<i>ITGA7</i>	600536	Muscular dystrophy, congenital, due to ITGA7 deficiency, 613204 (3), Autosomal recessive
<i>ITGA8</i>	604063	Renal hypodysplasia/aplasia 1, 191830 (3), Autosomal recessive
<i>ITGB2</i>	600065	Leukocyte adhesion deficiency, 116920 (3), Autosomal recessive
<i>ITGB3</i>	173470	Purpura, posttransfusion (3); Thrombocytopenia, neonatal alloimmune (3); Bleeding disorder, platelet-type, 16, autosomal dominant, 187800 (3), Autosomal dominant; {Myocardial infarction, susceptibility to}, 608446 (3); Glanzmann thrombasthenia, 273800 (3), Autosomal recessive
<i>ITGB4</i>	147557	Epidermolysis bullosa of hands and feet, 131800 (3), Autosomal dominant; Epidermolysis bullosa, junctional, with pyloric atresia, 226730 (3), Autosomal recessive; Epidermolysis bullosa, junctional, non-Herlitz type, 226650 (3), Autosomal recessive
<i>ITGB6</i>	147558	Amelogenesis imperfecta, type IH, 616221 (3), Autosomal recessive
<i>ITK</i>	186973	Lymphoproliferative syndrome 1, 613011 (3), Autosomal recessive
<i>ITM2B</i>	603904	Dementia, familial British, 176500 (3), Autosomal dominant; ?Retinal dystrophy with inner retinal dysfunction and ganglion cell abnormalities, 616079 (3), Autosomal dominant; Dementia, familial Danish, 117300 (3), Autosomal dominant
<i>ITPA</i>	147520	[Inosine triphosphatase deficiency], 613850 (3); Epileptic encephalopathy, early infantile, 35, 616647 (3), Autosomal recessive

<i>ITPR1</i>	147265	Spinocerebellar ataxia 29, congenital nonprogressive, 117360 (3), Autosomal dominant; Spinocerebellar ataxia 15, 606658 (3), Autosomal dominant; Gillespie syndrome, 206700 (3), Autosomal recessive, Autosomal dominant
<i>ITPR2</i>	600144	?Anhidrosis, isolated, with normal sweat glands, 106190 (3), Autosomal recessive
<i>ITPR3</i>	147267	{Diabetes, type 1, susceptibility to}, 222100 (2), Autosomal recessive
<i>IVD</i>	607036	Isovaleric acidemia, 243500 (3), Autosomal recessive
<i>IVNS1ABP</i>	609209	Immunodeficiency 70, 618969 (3), Autosomal dominant
<i>IYD</i>	612025	Thyroid dyshormonogenesis 4, 274800 (3), Autosomal recessive
<i>JAG1</i>	601920	?Deafness, congenital heart defects, and posterior embryotoxon, 617992 (3); Alagille syndrome 1, 118450 (3), Autosomal dominant; Tetralogy of Fallot, 187500 (3), Autosomal dominant
<i>JAGN1</i>	616012	Neutropenia, severe congenital, 6, autosomal recessive, 616022 (3), Autosomal recessive
<i>JAK2</i>	147796	Myelofibrosis, somatic, 254450 (3); Thrombocythemia 3, 614521 (3), Autosomal dominant, Somatic mutation; Polycythemia vera, somatic, 263300 (3); {Budd-Chiari syndrome, somatic}, 600880 (3); Leukemia, acute myeloid, somatic, 601626 (3); Erythrocytosis, somatic, 133100 (3)
<i>JAK3</i>	600173	SCID, autosomal recessive, T-negative/B-positive type, 600802 (3), Autosomal recessive
<i>JAM2</i>	606870	Basal ganglia calcification, idiopathic, 8, autosomal recessive, 618824 (3), Autosomal recessive
<i>JAM3</i>	606871	Hemorrhagic destruction of the brain, subependymal calcification, and cataracts, 613730 (3), Autosomal recessive
<i>JPH1</i>	605266	?Charcot-Marie-Tooth disease, axonal, autosomal dominant, type 2K, 607831 (3), Autosomal recessive, Autosomal dominant
<i>JPH2</i>	605267	Cardiomyopathy, hypertrophic, 17, 613873 (3), Autosomal dominant
<i>JPH3</i>	605268	Huntington disease-like 2, 606438 (3), Autosomal dominant
<i>JUP</i>	173325	Arrhythmogenic right ventricular dysplasia 12, 611528 (3), Autosomal dominant; Naxos disease, 601214 (3), Autosomal recessive
<i>KANK1</i>	607704	Cerebral palsy, spastic quadriplegic, 2, 612900 (3)
<i>KANK2</i>	614610	Palmoplantar keratoderma and woolly hair, 616099 (3), Autosomal recessive; Nephrotic syndrome, type 16, 617783 (3), Autosomal recessive
<i>KANSL1</i>	612452	Koolen-De Vries syndrome, 610443 (3), Autosomal dominant
<i>KARS1</i> (KARS)	601421	?Charcot-Marie-Tooth disease, recessive intermediate, B, 613641 (3), Autosomal recessive; Deafness, autosomal recessive 89, 613916 (3), Autosomal recessive
<i>KAT6A</i>	601408	Arboleda-Tham syndrome, 616268 (3), Autosomal dominant

<i>KAT6B</i>	605880	SBBYSS syndrome, 603736 (3), Autosomal dominant; Genitopatellar syndrome, 606170 (3), Autosomal dominant
<i>KAT8</i>	609912	Li-Ghorgani-Weisz-Hubshman syndrome, 618974 (3), Autosomal dominant
<i>KATNB1</i>	602703	Lissencephaly 6, with microcephaly, 616212 (3), Autosomal recessive
<i>KATNIP</i> (<i>KIAA0556</i>)	616650	Joubert syndrome 26, 616784 (3), Autosomal recessive
<i>KBTBD13</i>	613727	Nemaline myopathy 6, autosomal dominant, 609273 (3), Autosomal dominant
<i>KCNA1</i>	176260	Episodic ataxia/myokymia syndrome, 160120 (3), Autosomal dominant
<i>KCNA2</i>	176262	Epileptic encephalopathy, early infantile, 32, 616366 (3), Autosomal dominant
<i>KCNA4</i>	176266	Microcephaly, cataracts, impaired intellectual development, and dystonia with abnormal striatum, 618284 (3), Autosomal recessive
<i>KCNA5</i>	176267	Atrial fibrillation, familial, 7, 612240 (3), Autosomal dominant
<i>KCNB1</i>	600397	Epileptic encephalopathy, early infantile, 26, 616056 (3), Autosomal dominant
<i>KCNC1</i>	176258	Epilepsy, progressive myoclonic 7, 616187 (3), Autosomal dominant
<i>KCNC3</i>	176264	Spinocerebellar ataxia 13, 605259 (3), Autosomal dominant
<i>KCND3</i>	605411	Brugada syndrome 9, 616399 (3), Autosomal dominant; Spinocerebellar ataxia 19, 607346 (3), Autosomal dominant
<i>KCNE1</i>	176261	Long QT syndrome 5, 613695 (3), Autosomal dominant; Jervell and Lange-Nielsen syndrome 2, 612347 (3), Autosomal recessive
<i>KCNE2</i>	603796	Atrial fibrillation, familial, 4, 611493 (3); Long QT syndrome 6, 613693 (3), Autosomal dominant
<i>KCNE3</i>	604433	?Brugada syndrome 6, 613119 (3)
<i>KCNH1</i>	603305	Temple-Baraitser syndrome, 611816 (3), Autosomal dominant; Zimmermann-Laband syndrome 1, 135500 (3), Autosomal dominant
<i>KCNH2</i>	152427	{Long QT syndrome 2, acquired, susceptibility to}, 613688 (3), Autosomal dominant; Long QT syndrome 2, 613688 (3), Autosomal dominant; Short QT syndrome 1, 609620 (3)
<i>KCNJ1</i>	600359	Bartter syndrome, type 2, 241200 (3), Autosomal recessive
<i>KCNJ10</i>	602208	Enlarged vestibular aqueduct, digenic, 600791 (3), Autosomal recessive; SESAME syndrome, 612780 (3), Autosomal recessive
<i>KCNJ11</i>	600937	Maturity-onset diabetes of the young, type 13, 616329 (3), Autosomal dominant; {Diabetes mellitus, type 2, susceptibility to}, 125853 (3), Autosomal dominant; Diabetes, permanent neonatal 2, with or without neurologic features, 618856 (3), Autosomal dominant; Diabetes mellitus, transient neonatal, 3, 610582 (3), Autosomal dominant; Hyperinsulinemic hypoglycemia, familial, 2, 601820 (3), Autosomal recessive

<i>KCNJ13</i>	603208	Leber congenital amaurosis 16, 614186 (3), Autosomal recessive; Snowflake vitreoretinal degeneration, 193230 (3), Autosomal dominant
<i>KCNJ18</i>	613236	{Thyrotoxic periodic paralysis, susceptibility to, 2}, 613239 (3), Autosomal dominant
<i>KCNJ2</i>	600681	Short QT syndrome 3, 609622 (3); Atrial fibrillation, familial, 9, 613980 (3), Autosomal dominant; Andersen syndrome, 170390 (3), Autosomal dominant
<i>KCNJ5</i>	600734	Long QT syndrome 13, 613485 (3), Autosomal dominant; Hyperaldosteronism, familial, type III, 613677 (3), Autosomal dominant
<i>KCNJ6</i>	600877	Keppen-Lubinsky syndrome, 614098 (3), Autosomal dominant
<i>KCNK18</i>	613655	{Migraine, with or without aura, susceptibility to, 13}, 613656 (3), Autosomal dominant
<i>KCNK3</i>	603220	Pulmonary hypertension, primary, 4, 615344 (3), Autosomal dominant
<i>KCNK4</i>	605720	Facial dysmorphism, hypertrichosis, epilepsy, intellectual/developmental delay, and gingival overgrowth syndrome, 618381 (3), Autosomal dominant
<i>KCNK9</i>	605874	Birk-Barel mental retardation dysmorphism syndrome, 612292 (3)
<i>KCNMA1</i>	600150	Liang-Wang syndrome, 618729 (3), Autosomal dominant; {Epilepsy, idiopathic generalized, susceptibility to, 16}, 618596 (3), Autosomal dominant; Cerebellar atrophy, developmental delay, and seizures, 617643 (3), Autosomal recessive; Paroxysmal nonkinesigenic dyskinesia, 3, with or without generalized epilepsy, 609446 (3), Autosomal dominant
<i>KCNMB1</i>	603951	{Hypertension, diastolic, resistance to}, 608622 (3), Autosomal dominant
<i>KCNN3</i>	602983	Zimmermann-Laband syndrome 3, 618658 (3), Autosomal dominant
<i>KCNN4</i>	602754	Dehydrated hereditary stomatocytosis 2, 616689 (3), Autosomal dominant
<i>KCNQ1</i>	607542	Long QT syndrome 1, 192500 (3), Autosomal dominant; Jervell and Lange-Nielsen syndrome, 220400 (3), Autosomal recessive; Short QT syndrome 2, 609621 (3), Autosomal dominant; {Long QT syndrome 1, acquired, susceptibility to}, 192500 (3), Autosomal dominant; Atrial fibrillation, familial, 3, 607554 (3), Autosomal dominant
<i>KCNQ2</i>	602235	Epileptic encephalopathy, early infantile, 7, 613720 (3), Autosomal dominant; Seizures, benign neonatal, 1, 121200 (3), Autosomal dominant; Myokymia, 121200 (3), Autosomal dominant
<i>KCNQ3</i>	602232	Seizures, benign neonatal, 2, 121201 (3), Autosomal dominant
<i>KCNQ4</i>	603537	Deafness, autosomal dominant 2A, 600101 (3), Autosomal dominant

<i>KCNQ5</i>	607357	Mental retardation, autosomal dominant 46, 617601 (3), Autosomal dominant
<i>KCNT1</i>	608167	Epilepsy, nocturnal frontal lobe, 5, 615005 (3), Autosomal dominant; Epileptic encephalopathy, early infantile, 14, 614959 (3), Autosomal dominant
<i>KCNT2</i>	610044	?Epileptic encephalopathy, early infantile, 57, 617771 (3), Autosomal dominant
<i>KCNV2</i>	607604	Retinal cone dystrophy 3B, 610356 (3), Autosomal recessive
<i>KCTD1</i>	613420	Scalp-ear-nipple syndrome, 181270 (3), Autosomal dominant
<i>KCTD17</i>	616386	Dystonia 26, myoclonic, 616398 (3), Autosomal dominant
<i>KCTD7</i>	611725	Epilepsy, progressive myoclonic 3, with or without intracellular inclusions, 611726 (3), Autosomal recessive
<i>KDF1</i>	616758	?Ectodermal dysplasia 12, hypohidrotic/hair/tooth/nail type, 617337 (3), Autosomal dominant
<i>KDM1A</i>	609132	Cleft palate, psychomotor retardation, and distinctive facial features, 616728 (3), Autosomal dominant
<i>KDM3B</i>	609373	Diets-Jongmans syndrome, 618846 (3), Autosomal dominant
<i>KDM5B</i>	605393	Mental retardation, autosomal recessive 65, 618109 (3), Autosomal recessive
<i>KDM5C</i>	314690	Mental retardation, X-linked, syndromic, Claes-Jensen type, 300534 (3), X-linked recessive
<i>KDM6A</i>	300128	Kabuki syndrome 2, 300867 (3), X-linked dominant
<i>KDM6B</i>	611577	Neurodevelopmental disorder with coarse facies and mild distal skeletal abnormalities, 618505 (3), Autosomal dominant
<i>KDR</i>	191306	{Hemangioma, capillary infantile, susceptibility to}, 602089 (3), Autosomal dominant; Hemangioma, capillary infantile, somatic, 602089 (3)
<i>KDSR</i>	136440	Erythrokeratoderma variabilis et progressiva 4, 617526 (3), Autosomal recessive
<i>KEL</i>	613883	[Blood group, Kell], 110900 (3)
<i>KERA</i>	603288	Cornea plana 2, autosomal recessive, 217300 (3), Autosomal recessive
<i>KHDC3L</i>	611687	Hydatidiform mole, recurrent, 2, 614293 (3), Autosomal recessive
<i>KHK</i>	614058	[Fructosuria], 229800 (3), Autosomal recessive
<i>KIAA0586</i>	610178	Joubert syndrome 23, 616490 (3), Autosomal recessive; Short-rib thoracic dysplasia 14 with polydactyly, 616546 (3), Autosomal recessive
<i>KIAA0753</i>	617112	?Orofaciodigital syndrome XV, 617127 (3), Autosomal recessive
<i>KIAA0825</i>	617266	Polydactyly, postaxial, type A10, 618498 (3), Autosomal recessive
<i>KIAA1109</i>	611565	Alkuraya-Kucinskas syndrome, 617822 (3), Autosomal recessive
<i>KIAA1549</i>	613344	Retinitis pigmentosa 86, 618613 (3), Autosomal recessive
<i>KIDINS220</i>	615759	Spastic paraplegia, intellectual disability, nystagmus, and obesity, 617296 (3), Autosomal dominant

<i>KIF11</i>	148760	Microcephaly with or without chorioretinopathy, lymphedema, or mental retardation, 152950 (3), Autosomal dominant
<i>KIF14</i>	611279	Microcephaly 20, primary, autosomal recessive, 617914 (3), Autosomal recessive; ?Meckel syndrome 12, 616258 (3), Autosomal recessive
<i>KIF1A</i>	601255	NESCAV syndrome, 614255 (3), Autosomal dominant; Spastic paraplegia 30, autosomal dominant, 610357 (3), Autosomal recessive, Autosomal dominant; Neuropathy, hereditary sensory, type IIC, 614213 (3), Autosomal recessive; Spastic paraplegia 30, autosomal recessive, 610357 (3), Autosomal recessive, Autosomal dominant
<i>KIF1B</i>	605995	Pheochromocytoma, 171300 (3), Autosomal dominant; ?Charcot-Marie-Tooth disease, type 2A1, 118210 (3), Autosomal dominant; {Neuroblastoma, susceptibility to, 1}, 256700 (3), Autosomal dominant, Somatic mutation
<i>KIF1BP</i>	No OMIM gene	No OMIM phenotype
<i>KIF1C</i>	603060	Spastic ataxia 2, autosomal recessive, 611302 (3), Autosomal recessive
<i>KIF21A</i>	608283	Fibrosis of extraocular muscles, congenital, 1, 135700 (3), Autosomal dominant; Fibrosis of extraocular muscles, congenital, 3B, 135700 (3), Autosomal dominant
<i>KIF22</i>	603213	Spondyloepimetaphyseal dysplasia with joint laxity, type 2, 603546 (3), Autosomal dominant
<i>KIF2A</i>	602591	Cortical dysplasia, complex, with other brain malformations 3, 615411 (3), Autosomal dominant
<i>KIF3B</i>	603754	Retinitis pigmentosa 89, 618955 (3), Autosomal dominant
<i>KIF4A</i>	300521	?Mental retardation, X-linked 100, 300923 (3), X-linked recessive
<i>KIF5A</i>	602821	Myoclonus, intractable, neonatal, 617235 (3), Autosomal dominant; Spastic paraplegia 10, autosomal dominant, 604187 (3), Autosomal dominant; {Amyotrophic lateral sclerosis, susceptibility to, 25}, 617921 (3), Autosomal dominant
<i>KIF5C</i>	604593	Cortical dysplasia, complex, with other brain malformations 2, 615282 (3), Autosomal dominant
<i>KIF7</i>	611254	?Hydroletharus syndrome 2, 614120 (3), Autosomal recessive; Acrocallosal syndrome, 200990 (3), Autosomal recessive; Joubert syndrome 12, 200990 (3), Autosomal recessive; ?Al-Gazali-Bakalinova syndrome, 607131 (3), Autosomal recessive
<i>KIR3DL1</i>	604946	{AIDS, delayed/rapid progression to}, 609423 (3)
<i>KISS1</i>	603286	?Hypogonadotropic hypogonadism 13 with or without anosmia, 614842 (3), Autosomal recessive
<i>KISS1R</i>	604161	Hypogonadotropic hypogonadism 8 with or without anosmia, 614837 (3), Autosomal recessive; ?Precocious puberty, central, 1, 176400 (3), Autosomal dominant

<i>KIT</i>	164920	Gastrointestinal stromal tumor, familial, 606764 (3), Autosomal dominant, Isolated cases; Mastocytosis, cutaneous, 154800 (3), Autosomal dominant; Germ cell tumors, somatic, 273300 (3); Leukemia, acute myeloid, somatic, 601626 (3); Mastocytosis, systemic, somatic, 154800 (3); Piebaldism, 172800 (3), Autosomal dominant
<i>KITLG</i>	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)
<i>KIZ</i>	615757	Retinitis pigmentosa 69, 615780 (3), Autosomal recessive
<i>KL</i>	604824	?Tumoral calcinosis, hyperphosphatemic, familial, 3, 617994 (3), Autosomal recessive
<i>KLC2</i>	611729	Spastic paraplegia, optic atrophy, and neuropathy, 609541 (3), Autosomal recessive
<i>KLF1</i>	600599	Blood group--Lutheran inhibitor, 111150 (3); [Hereditary persistence of fetal hemoglobin], 613566 (3); Dyserythropoietic anemia, congenital, type IV, 613673 (3), Autosomal dominant
<i>KLF11</i>	603301	Maturity-onset diabetes of the young, type VII, 610508 (3)
<i>KLF6</i>	602053	Gastric cancer, somatic, 613659 (3); Prostate cancer, somatic, 176807 (3)
<i>KLHDC8B</i>	613169	{Hodgkin lymphoma, susceptibility to}, 236000 (3), Autosomal recessive
<i>KLHL10</i>	608778	Spermatogenic failure 11, 615081 (3), Autosomal dominant
<i>KLHL15</i>	300980	Mental retardation, X-linked 103, 300982 (3), X-linked recessive
<i>KLHL24</i>	611295	Epidermolysis bullosa simplex, generalized, with scarring and hair loss, 617294 (3), Autosomal dominant
<i>KLHL3</i>	605775	Pseudohypoaldosteronism, type IID, 614495 (3), Autosomal recessive, Autosomal dominant
<i>KLHL40</i>	615340	Nemaline myopathy 8, autosomal recessive, 615348 (3), Autosomal recessive
<i>KLHL41</i>	607701	Nemaline myopathy 9, 615731 (3), Autosomal recessive
<i>KLHL7</i>	611119	Retinitis pigmentosa 42, 612943 (3), Autosomal dominant; PERCHING syndrome, 617055 (3), Autosomal recessive
<i>KLK1</i>	147910	[Kallikrein, decreased urinary activity of], 615953 (3)
<i>KLK4</i>	603767	Amelogenesis imperfecta, type IIA1, 204700 (3), Autosomal recessive
<i>KLKB1</i>	229000	Fletcher factor (prekallikrein) deficiency, 612423 (3), Autosomal recessive
<i>KLLN</i>	612105	Cowden syndrome 4, 615107 (3)
<i>KMT2A</i>	159555	Wiedemann-Steiner syndrome, 605130 (3), Autosomal dominant
<i>KMT2B</i>	606834	Dystonia 28, childhood-onset, 617284 (3), Autosomal dominant
<i>KMT2C</i>	606833	Kleefstra syndrome 2, 617768 (3), Autosomal dominant
<i>KMT2D</i>	602113	Kabuki syndrome 1, 147920 (3), Autosomal dominant
<i>KMT2E</i>	608444	O'Donnell-Luria-Rodan syndrome, 618512 (3), Autosomal dominant

<i>KMT5B</i>	610881	Mental retardation, autosomal dominant 51, 617788 (3), Autosomal dominant
<i>KNG1</i>	612358	[Kininogen deficiency], 228960 (3), Autosomal recessive; [High molecular weight kininogen deficiency], 228960 (3), Autosomal recessive
<i>KNL1</i>	609173	Microcephaly 4, primary, autosomal recessive, 604321 (3), Autosomal recessive
<i>KPTN</i>	615620	Mental retardation, autosomal recessive 41, 615637 (3), Autosomal recessive
<i>KRAS</i>	190070	Oculoectodermal syndrome, somatic, 600268 (3); Leukemia, acute myeloid, somatic, 601626 (3); Breast cancer, somatic, 114480 (3); RAS-associated autoimmune leukoproliferative disorder, 614470 (3), Autosomal dominant; Cardiofaciocutaneous syndrome 2, 615278 (3), Autosomal dominant; Arteriovenous malformation of the brain, somatic, 108010 (3); Bladder cancer, somatic, 109800 (3); Pancreatic carcinoma, somatic, 260350 (3); Lung cancer, somatic, 211980 (3); Gastric cancer, somatic, 137215 (3); Schimmelpenning- Feuerstein-Mims syndrome, somatic mosaic, 163200 (3); Noonan syndrome 3, 609942 (3), Autosomal dominant
<i>KREMEN1</i>	609898	Ectodermal dysplasia 13, hair/tooth type, 617392 (3), Autosomal recessive
<i>KRIT1</i>	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
<i>KRT1</i>	139350	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; Ichthyosis histris, Curth-Macklin type, 146590 (3), Autosomal dominant; Epidermolytic hyperkeratosis, 113800 (3), Autosomal recessive, Autosomal dominant
<i>KRT10</i>	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
<i>KRT12</i>	601687	Meesmann corneal dystrophy 1, 122100 (3), Autosomal dominant
<i>KRT13</i>	148065	White sponge nevus 2, 615785 (3), Autosomal dominant

<i>KRT14</i>	148066	Naegeli-Franceschetti-Jadassohn syndrome, 161000 (3), Autosomal dominant; Epidermolysis bullosa simplex, Dowling-Meara type, 131760 (3), Autosomal dominant; Dermatopathia pigmentosa reticularis, 125595 (3), Autosomal dominant; Epidermolysis bullosa simplex, Koebner type, 131900 (3), Autosomal dominant; Epidermolysis bullosa simplex, recessive 1, 601001 (3), Autosomal recessive; Epidermolysis bullosa simplex, Weber-Cockayne type, 131800 (3), Autosomal dominant
<i>KRT16</i>	148067	Palmoplantar keratoderma, nonepidermolytic, focal, 613000 (3), Autosomal dominant; Pachyonychia congenita 1, 167200 (3), Autosomal dominant
<i>KRT17</i>	148069	Pachyonychia congenita 2, 167210 (3), Autosomal dominant; Steatocystoma multiplex, 184500 (3), Autosomal dominant
<i>KRT18</i>	148070	{Cirrhosis, noncryptogenic, susceptibility to}, 215600 (3), Autosomal recessive; Cirrhosis, cryptogenic, 215600 (3), Autosomal recessive
<i>KRT2</i>	600194	Ichthyosis bullosa of Siemens, 146800 (3), Autosomal dominant
<i>KRT25</i>	616646	Woolly hair, autosomal recessive 3, 616760 (3), Autosomal recessive
<i>KRT3</i>	148043	Meesmann corneal dystrophy 2, 618767 (3), Autosomal dominant
<i>KRT4</i>	123940	White sponge nevus 1, 193900 (3), Autosomal dominant
<i>KRT5</i>	148040	Epidermolysis bullosa simplex, Weber-Cockayne type, 131800 (3), Autosomal dominant; Epidermolysis bullosa simplex-MCR, 609352 (3); Epidermolysis bullosa simplex-MP, 131960 (3), Autosomal dominant; Dowling-Degos disease 1, 179850 (3), Autosomal dominant; Epidermolysis bullosa simplex, Koebner type, 131900 (3), Autosomal dominant; Epidermolysis bullosa simplex, Dowling-Meara type, 131760 (3), Autosomal dominant; Epidermolysis bullosa simplex, recessive 1, 601001 (3), Autosomal recessive
<i>KRT6A</i>	148041	Pachyonychia congenita 3, 615726 (3), Autosomal dominant
<i>KRT6B</i>	148042	Pachyonychia congenita 4, 615728 (3), Autosomal dominant
<i>KRT6C</i>	612315	Palmoplantar keratoderma, nonepidermolytic, focal or diffuse, 615735 (3), Autosomal dominant
<i>KRT71</i>	608245	?Hypotrichosis 13, 615896 (3), Autosomal dominant
<i>KRT74</i>	608248	?Ectodermal dysplasia 7, hair/nail type, 614929 (3), Autosomal recessive; Woolly hair, autosomal dominant, 194300 (3), Autosomal dominant; ?Hypotrichosis 3, 613981 (3), Autosomal dominant
<i>KRT75</i>	609025	{Pseudofolliculitis barbae, susceptibility to}, 612318 (3)
<i>KRT8</i>	148060	{Cirrhosis, noncryptogenic, susceptibility to}, 215600 (3), Autosomal recessive; Cirrhosis, cryptogenic, 215600 (3), Autosomal recessive
<i>KRT81</i>	602153	Monilethrix, 158000 (3), Autosomal dominant

<i>KRT83</i>	602765	Erythrokeratoderma variabilis et progressiva 5, 617756 (3), Autosomal recessive; Monilethrix, 158000 (3), Autosomal dominant
<i>KRT85</i>	602767	Ectodermal dysplasia 4, hair/nail type, 602032 (3), Autosomal recessive
<i>KRT86</i>	601928	Monilethrix, 158000 (3), Autosomal dominant
<i>KRT9</i>	607606	Palmoplantar keratoderma, epidermolytic, 144200 (3), Autosomal dominant
<i>KY</i>	605739	Myopathy, myofibrillar, 7, 617114 (3), Autosomal recessive
<i>KYNU</i>	605197	Vertebral, cardiac, renal, and limb defects syndrome 2, 617661 (3), Autosomal recessive; ?Hydroxykynureninuria, 236800 (3), Autosomal recessive
<i>L1CAM</i>	308840	MASA syndrome, 303350 (3), X-linked recessive; Hydrocephalus with Hirschsprung disease, 307000 (3), X-linked recessive; Hydrocephalus with congenital idiopathic intestinal pseudoobstruction, 307000 (3), X-linked recessive; Corpus callosum, partial agenesis of, 304100 (3), X-linked recessive; CRASH syndrome, 303350 (3), X-linked recessive; Hydrocephalus due to aqueductal stenosis, 307000 (3), X-linked recessive
<i>L2HGDH</i>	609584	L-2-hydroxyglutaric aciduria, 236792 (3), Autosomal recessive
<i>LACC1</i>	613409	Juvenile arthritis, 618795 (3), Autosomal recessive
<i>LAGE3</i>	300060	Galloway-Mowat syndrome 2, X-linked, 301006 (3), X-linked recessive
<i>LAMA1</i>	150320	Poretti-Boltshauser syndrome, 615960 (3), Autosomal recessive
<i>LAMA2</i>	156225	Muscular dystrophy, limb-girdle, autosomal recessive 23, 618138 (3), Autosomal recessive; Muscular dystrophy, congenital, merosin deficient or partially deficient, 607855 (3), Autosomal recessive
<i>LAMA3</i>	600805	Epidermolysis bullosa, junctional, Herlitz type, 226700 (3), Autosomal recessive; Laryngoonychocutaneous syndrome, 245660 (3), Autosomal recessive; Epidermolysis bullosa, generalized atrophic benign, 226650 (3), Autosomal recessive
<i>LAMA4</i>	600133	Cardiomyopathy, dilated, 1J, 615235 (3), Autosomal dominant
<i>LAMB1</i>	150240	Lissencephaly 5, 615191 (3), Autosomal recessive
<i>LAMB2</i>	150325	Nephrotic syndrome, type 5, with or without ocular abnormalities, 614199 (3); Pierson syndrome, 609049 (3), Autosomal recessive
<i>LAMB3</i>	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
<i>LAMC2</i>	150292	Epidermolysis bullosa, junctional, non-Herlitz type, 226650 (3), Autosomal recessive; Epidermolysis bullosa, junctional, Herlitz type, 226700 (3), Autosomal recessive
<i>LAMC3</i>	604349	Cortical malformations, occipital, 614115 (3), Autosomal recessive

<i>LAMP2</i>	309060	Danon disease, 300257 (3), X-linked dominant
<i>LAMTOR2</i>	610389	Immunodeficiency due to defect in MAPBP-interacting protein, 610798 (3), Autosomal recessive
<i>LARGE1</i>	603590	Muscular dystrophy-dystroglycanopathy (congenital with mental retardation), type B, 6, 608840 (3), Autosomal recessive; Muscular dystrophy-dystroglycanopathy (congenital with brain and eye anomalies), type A, 6, 613154 (3), Autosomal recessive
<i>LARP7</i>	612026	Alazami syndrome, 615071 (3), Autosomal recessive
<i>LARS1 (LARS)</i>	151350	?Infantile liver failure syndrome 1, 615438 (3), Autosomal recessive
<i>LARS2</i>	604544	Perrault syndrome 4, 615300 (3), Autosomal recessive; ?Hydrops, lactic acidosis, and sideroblastic anemia, 617021 (3), Autosomal recessive
<i>LAS1L</i>	300964	Wilson-Turner syndrome, 309585 (3), X-linked recessive
<i>LAT</i>	602354	Immunodeficiency 52, 617514 (3), Autosomal recessive
<i>LBR</i>	600024	Pelger-Huet anomaly, 169400 (3), Autosomal dominant; Greenberg skeletal dysplasia, 215140 (3), Autosomal recessive; ?Reynolds syndrome, 613471 (3), Autosomal dominant; Pelger-Huet anomaly with mild skeletal anomalies, 618019 (3)
<i>LCA5</i>	611408	Leber congenital amaurosis 5, 604537 (3), Autosomal recessive
<i>LCAT</i>	606967	Norum disease, 245900 (3), Autosomal recessive; Fish-eye disease, 136120 (3), Autosomal recessive
<i>LCK</i>	153390	?Immunodeficiency 22, 615758 (3), Autosomal recessive
<i>LCT</i>	603202	Lactase deficiency, congenital, 223000 (3), Autosomal recessive
<i>LDB3</i>	605906	Cardiomyopathy, hypertrophic, 24, 601493 (3), Autosomal dominant; Cardiomyopathy, dilated, 1C, with or without LVNC, 601493 (3), Autosomal dominant; Myopathy, myofibrillar, 4, 609452 (3), Autosomal dominant; Left ventricular noncompaction 3, 601493 (3), Autosomal dominant
<i>LDHA</i>	150000	Glycogen storage disease XI, 612933 (3), Autosomal recessive
<i>LDHB</i>	150100	[Lactate dehydrogenase-B deficiency], 614128 (3)
<i>LDHD</i>	607490	D-lactic aciduria with susceptibility to gout, 245450 (3), Autosomal recessive
<i>LDLR</i>	606945	Hypercholesterolemia, familial, 1, 143890 (3), Autosomal dominant; LDL cholesterol level QTL2, 143890 (3), Autosomal dominant
<i>LDLRAP1</i>	605747	Hypercholesterolemia, familial, 4, 603813 (3), Autosomal recessive
<i>LEF1</i>	153245	Sebaceous tumors, somatic (3)
<i>LEMD2</i>	616312	Cataract 46, juvenile-onset, 212500 (3), Autosomal recessive
<i>LEMD3</i>	607844	Osteopoikilosis with or without melorheostosis, 166700 (3), Autosomal dominant; Buschke-Ollendorff syndrome, 166700 (3), Autosomal dominant
<i>LEP</i>	164160	Obesity, morbid, due to leptin deficiency, 614962 (3), Autosomal recessive

<i>LEPR</i>	601007	Obesity, morbid, due to leptin receptor deficiency, 614963 (3), Autosomal recessive
<i>LFNG</i>	602576	Spondylocostal dysostosis 3, autosomal recessive, 609813 (3), Autosomal recessive
<i>LGALS2</i>	150571	{Myocardial infarction, susceptibility to}, 608446 (3)
<i>LGI1</i>	604619	Epilepsy, familial temporal lobe, 1, 600512 (3), Autosomal dominant
<i>LGI4</i>	608303	Arthrogryposis multiplex congenita 1, neurogenic, with myelin defect, 617468 (3), Autosomal recessive
<i>LGR4</i>	606666	{Bone mineral density, low, susceptibility to}, 615311 (3)
<i>LHB</i>	152780	Hypogonadotropic hypogonadism 23 with or without anosmia, 228300 (3), Autosomal recessive
<i>LHCGR</i>	152790	Leydig cell adenoma, somatic, with precocious puberty, 176410 (3); Precocious puberty, male, 176410 (3), Autosomal dominant; Luteinizing hormone resistance, female, 238320 (3), Autosomal recessive; Leydig cell hypoplasia with pseudohermaphroditism, 238320 (3), Autosomal recessive; Leydig cell hypoplasia with hypergonadotropic hypogonadism, 238320 (3), Autosomal recessive
<i>LHFPL5</i>	609427	Deafness, autosomal recessive 67, 610265 (3), Autosomal recessive
<i>LHX3</i>	600577	Pituitary hormone deficiency, combined, 3, 221750 (3), Autosomal recessive
<i>LHX4</i>	602146	Pituitary hormone deficiency, combined, 4, 262700 (3), Autosomal dominant
<i>LIAS</i>	607031	Hyperglycinemia, lactic acidosis, and seizures, 614462 (3), Autosomal recessive
<i>LIFR</i>	151443	Stuve-Wiedemann syndrome/Schwartz-Jampel type 2 syndrome, 601559 (3), Autosomal recessive
<i>LIG4</i>	601837	{Multiple myeloma, resistance to}, 254500 (3), Somatic mutation; LIG4 syndrome, 606593 (3), Autosomal recessive
<i>LIM2</i>	154045	Cataract 19, multiple types, 615277 (3), Autosomal recessive
<i>LIMA1</i>	608364	[Low density lipoprotein cholesterol level QTL 8], 618079 (3)
<i>LIMS2</i>	607908	?Muscular dystrophy, autosomal recessive, with cardiomyopathy and triangular tongue, 616827 (3), Autosomal recessive
<i>LINGO1</i>	609791	Mental retardation, autosomal recessive 64, 618103 (3), Autosomal recessive
<i>LINS1</i>	610350	Mental retardation, autosomal recessive 27, 614340 (3), Autosomal recessive
<i>LIPA</i>	613497	Wolman disease, 278000 (3), Autosomal recessive; Cholesteryl ester storage disease, 278000 (3), Autosomal recessive
<i>LIPC</i>	151670	[High density lipoprotein cholesterol level QTL 12], 612797 (3); Hepatic lipase deficiency, 614025 (3), Autosomal recessive; {Diabetes mellitus, noninsulin-dependent}, 125853 (3), Autosomal dominant

<i>LIPE</i>	151750	Lipodystrophy, familial partial, type 6, 615980 (3), Autosomal recessive
<i>LIPH</i>	607365	Woolly hair, autosomal recessive 2 with or without hypotrichosis, 604379 (3), Autosomal recessive; Hypotrichosis 7, 604379 (3), Autosomal recessive
<i>LIPN</i>	613924	Ichthyosis, congenital, autosomal recessive 8, 613943 (3), Autosomal recessive
<i>LIPT1</i>	610284	Lipoyltransferase 1 deficiency, 616299 (3), Autosomal recessive
<i>LIPT2</i>	617659	Encephalopathy, neonatal severe, with lactic acidosis and brain abnormalities, 617668 (3), Autosomal recessive
<i>LITAF</i>	603795	Charcot-Marie-Tooth disease, type 1C, 601098 (3), Autosomal dominant
<i>LMAN1</i>	601567	Combined factor V and VIII deficiency, 227300 (3), Autosomal recessive
<i>LMAN2L</i>	609552	?Mental retardation, autosomal recessive, 52, 616887 (3), Autosomal recessive
<i>LMBR1</i>	605522	Triphalangeal thumb-polysyndactyly syndrome, 174500 (3), Autosomal dominant; Syndactyly, type IV, 186200 (3), Autosomal dominant; Triphalangeal thumb, type I, 174500 (3), Autosomal dominant; Acheiropody, 200500 (3), Autosomal recessive; Laurin-Sandrow syndrome, 135750 (3), Autosomal dominant; Hypoplastic or aplastic tibia with polydactyly, 188740 (3), Autosomal dominant; Polydactyly, preaxial type II, 174500 (3), Autosomal dominant
<i>LMBRD1</i>	612625	Methylmalonic aciduria and homocystinuria, cblF type, 277380 (3), Autosomal recessive
<i>LMF1</i>	611761	Lipase deficiency, combined, 246650 (3), Autosomal recessive
<i>LMNA</i>	150330	Muscular dystrophy, congenital, 613205 (3), Autosomal dominant; Lipodystrophy, familial partial, type 2, 151660 (3), Autosomal dominant; Charcot-Marie-Tooth disease, type 2B1, 605588 (3), Autosomal recessive; Cardiomyopathy, dilated, 1A, 115200 (3), Autosomal dominant; Heart-hand syndrome, Slovenian type, 610140 (3), Autosomal dominant; Hutchinson-Gilford progeria, 176670 (3), Autosomal recessive, Autosomal dominant; Restrictive dermopathy, lethal, 275210 (3), Autosomal recessive; Mandibuloacral dysplasia, 248370 (3), Autosomal recessive; Emery-Dreifuss muscular dystrophy 2, autosomal dominant, 181350 (3), Autosomal dominant; Emery-Dreifuss muscular dystrophy 3, autosomal recessive, 616516 (3), Autosomal recessive; Malouf syndrome, 212112 (3), Autosomal dominant
<i>LMNB1</i>	150340	Leukodystrophy, adult-onset, autosomal dominant, 169500 (3), Autosomal dominant
<i>LMNB2</i>	150341	{Lipodystrophy, partial, acquired, susceptibility to}, 608709 (3), Autosomal dominant; ?Epilepsy, progressive myoclonic, 9, 616540 (3), Autosomal recessive

<i>LMO1</i>	186921	Leukemia, T-cell acute lymphoblastic, 186921 (2)
<i>LMO2</i>	180385	Leukemia, acute T-cell, 180385 (2)
<i>LMOD3</i>	616112	Nemaline myopathy 10, 616165 (3), Autosomal recessive
<i>LMX1A</i>	600298	Deafness, autosomal dominant 7, 601412 (3), Autosomal dominant
<i>LMX1B</i>	602575	Nail-patella syndrome, 161200 (3), Autosomal dominant
<i>LNPK</i>	610236	Neurodevelopmental disorder with epilepsy and hypoplasia of the corpus callosum, 618090 (3), Autosomal recessive
<i>LONP1</i>	605490	CODAS syndrome, 600373 (3), Autosomal recessive
<i>LORICRIN (LOR)</i>	152445	Vohwinkel syndrome with ichthyosis, 604117 (3), Autosomal dominant
<i>LOX</i>	153455	Aortic aneurysm, familial thoracic 10, 617168 (3), Autosomal dominant
<i>LOXHD1</i>	613072	Deafness, autosomal recessive 77, 613079 (3), Autosomal recessive
<i>LOXL1</i>	153456	{Exfoliation syndrome, susceptibility to}, 177650 (3), Autosomal dominant
<i>LPA</i>	152200	[LPA deficiency, congenital], 618807 (3); {Coronary artery disease, susceptibility to}, 618807 (3)
<i>LPAR6</i>	609239	Hypotrichosis 8, 278150 (3), Autosomal recessive; Woolly hair, autosomal recessive 1, with or without hypotrichosis, 278150 (3), Autosomal recessive
<i>LPIN1</i>	605518	Myoglobinuria, acute recurrent, autosomal recessive, 268200 (3), Autosomal recessive
<i>LPIN2</i>	605519	Majeed syndrome, 609628 (3)
<i>LPL</i>	609708	Lipoprotein lipase deficiency, 238600 (3), Autosomal recessive; [High density lipoprotein cholesterol level QTL 11], 238600 (3), Autosomal recessive; Combined hyperlipidemia, familial, 144250 (3), Autosomal dominant
<i>LPP</i>	600700	Lipoma (3); Leukemia, acute myeloid, 601626 (3), Autosomal dominant, Somatic mutation
<i>LRAT</i>	604863	Retinal dystrophy, early-onset severe, 613341 (3), Autosomal recessive; Leber congenital amaurosis 14, 613341 (3), Autosomal recessive; Retinitis pigmentosa, juvenile, 613341 (3), Autosomal recessive
<i>LRBA</i>	606453	Immunodeficiency, common variable, 8, with autoimmunity, 614700 (3), Autosomal recessive
<i>LRIG2</i>	608869	Urofacial syndrome 2, 615112 (3), Autosomal recessive
<i>LRIT3</i>	615004	Night blindness, congenital stationary (complete), 1F, autosomal recessive, 615058 (3), Autosomal recessive
<i>LRMDA</i>	614537	Albinism, oculocutaneous, type VII, 615179 (3), Autosomal recessive
<i>LRP1</i>	107770	?Keratosis pilaris atrophicans, 604093 (3), Autosomal recessive
<i>LRP12</i>	618299	Oculopharyngodistal myopathy 1, 164310 (3), Autosomal dominant
<i>LRP2</i>	600073	Donnai-Barrow syndrome, 222448 (3), Autosomal recessive

<i>LRP4</i>	604270	?Myasthenic syndrome, congenital, 17, 616304 (3), Autosomal recessive; Sclerosteosis 2, 614305 (3), Autosomal recessive, Autosomal dominant; Cenani-Lenz syndactyly syndrome, 212780 (3), Autosomal recessive
<i>LRP5</i>	603506	van Buchem disease, type 2, 607636 (3), Autosomal dominant; Exudative vitreoretinopathy 4, 601813 (3), Autosomal recessive, Autosomal dominant; Hyperostosis, endosteal, 144750 (3), Autosomal dominant; Osteosclerosis, 144750 (3), Autosomal dominant; Polycystic liver disease 4 with or without kidney cysts, 617875 (3), Autosomal dominant; Osteoporosis-pseudoglioma syndrome, 259770 (3), Autosomal recessive; Osteopetrosis, autosomal dominant 1, 607634 (3), Autosomal dominant; {Osteoporosis}, 166710 (3), Autosomal dominant; [Bone mineral density variability 1], 601884 (3), Autosomal dominant {Coronary artery disease, autosomal dominant, 2}, 610947 (3), Autosomal dominant; Tooth agenesis, selective, 7, 616724 (3), Autosomal dominant
<i>LRP6</i>	603507	
<i>LRP8</i>	602600	{Myocardial infarction, susceptibility to}, 608446 (3)
<i>LRPAP1</i>	104225	Myopia 23, autosomal recessive, 615431 (3), Autosomal recessive
<i>LRPPRC</i>	607544	Leigh syndrome, French-Canadian type, 220111 (3), Autosomal recessive
<i>LRRC56</i>	618227	Ciliary dyskinesia, primary, 39, 618254 (3), Autosomal recessive
<i>LRRC6</i>	614930	Ciliary dyskinesia, primary, 19, 614935 (3), Autosomal recessive
<i>LRRC8A</i>	608360	?Agammaglobulinemia 5, 613506 (3), Autosomal dominant
<i>LRRK2</i>	609007	{Parkinson disease 8}, 607060 (3), Autosomal dominant
<i>LRSAM1</i>	610933	Charcot-Marie-Tooth disease, axonal, type 2P, 614436 (3), Autosomal recessive, Autosomal dominant
<i>LRTOMT</i>	612414	Deafness, autosomal recessive 63, 611451 (3), Autosomal recessive
<i>LSS</i>	600909	Alopecia-mental retardation syndrome 4, 618840 (3), Autosomal recessive; Cataract 44, 616509 (3), Autosomal recessive; Hypotrichosis 14, 618275 (3), Autosomal recessive
<i>LTA</i>	153440	{Psoriatic arthritis, susceptibility to}, 607507 (3); {Leprosy, susceptibility to, 4}, 610988 (3); {Myocardial infarction, susceptibility to}, 608446 (3)
<i>LTBP2</i>	602091	Microspherophakia and/or megalocornea, with ectopia lentis and with or without secondary glaucoma, 251750 (3), Autosomal recessive; Glaucoma 3, primary congenital, D, 613086 (3); ?Weill-Marchesani syndrome 3, recessive, 614819 (3), Autosomal recessive
<i>LTBP3</i>	602090	Dental anomalies and short stature, 601216 (3), Autosomal recessive; Geleophysic dysplasia 3, 617809 (3), Autosomal dominant
<i>LTBP4</i>	604710	Cutis laxa, autosomal recessive, type IC, 613177 (3), Autosomal recessive

<i>LTC4S</i>	246530	Leukotriene C4 synthase deficiency, 614037 (1), Autosomal recessive
<i>LYL1</i>	151440	Leukemia, T-cell acute lymphoblastoid, 151440 (2)
<i>LYRM4</i>	613311	?Combined oxidative phosphorylation deficiency 19, 615595 (3), Autosomal recessive
<i>LYRM7</i>	615831	Mitochondrial complex III deficiency, nuclear type 8, 615838 (3), Autosomal recessive
<i>LYST</i>	606897	Chediak-Higashi syndrome, 214500 (3), Autosomal recessive
<i>LYZ</i>	153450	Amyloidosis, renal, 105200 (3), Autosomal dominant
<i>LZTFL1</i>	606568	Bardet-Biedl syndrome 17, 615994 (3), Autosomal recessive
<i>LZTR1</i>	600574	{Schwannomatosis-2, susceptibility to}, 615670 (3), Autosomal dominant; Noonan syndrome 2, 605275 (3), Autosomal recessive; Noonan syndrome 10, 616564 (3), Autosomal dominant
<i>LZTS1</i>	606551	Esophageal squamous cell carcinoma, somatic, 133239 (3)
<i>MAB21L1</i>	601280	Cerebellar, ocular, craniofacial, and genital syndrome, 618479 (3), Autosomal recessive
<i>MAB21L2</i>	604357	Microphthalmia/coloboma and skeletal dysplasia syndrome, 615877 (3), Autosomal recessive, Autosomal dominant
<i>MACF1</i>	608271	Lissencephaly 9 with complex brainstem malformation, 618325 (3), Autosomal dominant
<i>MAD1L1</i>	602686	Lymphoma, somatic (3); Prostate cancer, somatic, 176807 (3)
<i>MAD2L2</i>	604094	?Fanconi anemia, complementation group V, 617243 (3), Autosomal recessive
<i>MAF</i>	177075	Ayme-Gripp syndrome, 601088 (3), Autosomal dominant; Cataract 21, multiple types, 610202 (3), Autosomal dominant
<i>MAFA</i>	610303	Insulinomatosis and diabetes mellitus, 147630 (3), Autosomal dominant
<i>MAFB</i>	608968	Duane retraction syndrome 3, 617041 (3), Autosomal dominant; Multicentric carpotarsal osteolysis syndrome, 166300 (3), Autosomal dominant
<i>MAG</i>	159460	Spastic paraplegia 75, autosomal recessive, 616680 (3), Autosomal recessive
<i>MAGED2</i>	300470	Bartter syndrome, type 5, antenatal, transient, 300971 (3), X-linked recessive
<i>MAGEL2</i>	605283	Schaaf-Yang syndrome, 615547 (3), Autosomal dominant
<i>MAGI2</i>	606382	Nephrotic syndrome, type 15, 617609 (3), Autosomal recessive
<i>MAGT1</i>	300715	Immunodeficiency, X-linked, with magnesium defect, Epstein-Barr virus infection and neoplasia, 300853 (3), X-linked recessive; Congenital disorder of glycosylation, type Icc, 301031 (3), X-linked recessive
<i>MAK</i>	154235	Retinitis pigmentosa 62, 614181 (3), Autosomal recessive
<i>MALT1</i>	604860	Immunodeficiency 12, 615468 (3), Autosomal recessive
<i>MAML2</i>	607537	Mucoepidermoid salivary gland carcinoma (3)
<i>MAMLD1</i>	300120	Hypospadias 2, X-linked, 300758 (3), X-linked recessive

<i>MAN1B1</i>	604346	Mental retardation, autosomal recessive 15, 614202 (3), Autosomal recessive
<i>MAN2B1</i>	609458	Mannosidosis, alpha-, types I and II, 248500 (3), Autosomal recessive
<i>MANBA</i>	609489	Mannosidosis, beta, 248510 (3), Autosomal recessive
<i>MAOA</i>	309850	Brunner syndrome, 300615 (3), X-linked recessive; {Antisocial behavior}, 300615 (3), X-linked recessive
<i>MAP11 (C7orf43)</i>	618350	?Microcephaly 25, primary, autosomal recessive, 618351 (3), Autosomal recessive
<i>MAP1B</i>	157129	Periventricular nodular heterotopia 9, 618918 (3), Autosomal dominant
<i>MAP2K1</i>	176872	Cardiofaciocutaneous syndrome 3, 615279 (3), Autosomal dominant; Melorheostosis, isolated, somatic mosaic, 155950 (3)
<i>MAP2K2</i>	601263	Cardiofaciocutaneous syndrome 4, 615280 (3), Autosomal dominant
<i>MAP3K1</i>	600982	46XY sex reversal 6, 613762 (3), Autosomal dominant
<i>MAP3K20</i>	609479	Split-foot malformation with mesoaxial polydactyly, 616890 (3), Autosomal recessive; Centronuclear myopathy 6 with fiber-type disproportion, 617760 (3), Autosomal recessive
<i>MAP3K7</i>	602614	Cardiospondylocarpofacial syndrome, 157800 (3), Autosomal dominant; Frontometaphyseal dysplasia 2, 617137 (3), Autosomal dominant
<i>MAP3K8</i>	191195	Lung cancer, somatic, 211980 (3)
<i>MAPK8IP1</i>	604641	{Diabetes mellitus, noninsulin-dependent}, 125853 (3), Autosomal dominant
<i>MAPK8IP3</i>	605431	Neurodevelopmental disorder with or without variable brain abnormalities, 618443 (3), Autosomal dominant
<i>MAPKAPK3</i>	602130	?Macular dystrophy, patterned, 3, 617111 (3), Autosomal dominant
<i>MAPKBP1</i>	616786	Nephronophthisis 20, 617271 (3), Autosomal recessive
<i>MAPRE2</i>	605789	Symmetric circumferential skin creases, congenital, 2, 616734 (3), Autosomal dominant
<i>MAPT</i>	157140	Pick disease, 172700 (3), Autosomal dominant; Dementia, frontotemporal, with or without parkinsonism, 600274 (3), Autosomal dominant; {Parkinson disease, susceptibility to}, 168600 (3), Autosomal dominant, Multifactorial; Supranuclear palsy, progressive, 601104 (3), Autosomal dominant; Supranuclear palsy, progressive atypical, 260540 (3), Autosomal recessive
<i>MARCHF6 (MARCH6)</i>	613297	Epilepsy, familial adult myoclonic, 3, 613608 (3), Autosomal dominant
<i>MARK3</i>	602678	?Visual impairment and progressive phthisis bulbi, 618283 (3), Autosomal recessive
<i>MARS1 (MARS)</i>	156560	Charcot-Marie-Tooth disease, axonal, type 2U, 616280 (3), Autosomal dominant; Interstitial lung and liver disease, 615486 (3), Autosomal recessive

<i>MARS2</i>	609728	Spastic ataxia 3, autosomal recessive, 611390 (3), Autosomal recessive; ?Combined oxidative phosphorylation deficiency 25, 616430 (3), Autosomal recessive
<i>MARVELD2</i>	610572	Deafness, autosomal recessive 49, 610153 (3), Autosomal recessive
<i>MASP1</i>	600521	3MC syndrome 1, 257920 (3), Autosomal recessive
<i>MASP2</i>	605102	MASP2 deficiency, 613791 (3), Autosomal recessive
<i>MAST1</i>	612256	Mega-corpus-callosum syndrome with cerebellar hypoplasia and cortical malformations, 618273 (3), Autosomal dominant
<i>MAT1A</i>	610550	Methionine adenosyltransferase deficiency, autosomal recessive, 250850 (3), Autosomal recessive, Autosomal dominant; Hypermethioninemia, persistent, autosomal dominant, due to methionine adenosyltransferase I/III deficiency, 250850 (3), Autosomal recessive, Autosomal dominant
<i>MATN3</i>	602109	{Osteoarthritis susceptibility 2}, 140600 (3), Autosomal dominant; ?Spondyloepimetaphyseal dysplasia, 608728 (3), Autosomal recessive; Epiphyseal dysplasia, multiple, 5, 607078 (3), Autosomal dominant
<i>MATR3</i>	164015	Amyotrophic lateral sclerosis 21, 606070 (3), Autosomal dominant
<i>MAX</i>	154950	{Pheochromocytoma, susceptibility to}, 171300 (3), Autosomal dominant
<i>MBD5</i>	611472	Mental retardation, autosomal dominant 1, 156200 (3), Autosomal dominant
<i>MBL2</i>	154545	{Chronic infections, due to MBL deficiency}, 614372 (3), Autosomal dominant
<i>MBOAT7</i>	606048	Mental retardation, autosomal recessive 57, 617188 (3), Autosomal recessive
<i>MBTPS1</i>	603355	?Spondyloepiphyseal dysplasia, Kondo-Fu type, 618392 (3), Autosomal recessive
<i>MBTPS2</i>	300294	IFAP syndrome with or without BRESHECK syndrome, 308205 (3), X-linked recessive; Keratosis follicularis spinulosa decalvans, X-linked, 308800 (3), X-linked recessive; Osteogenesis imperfecta, type XIX, 301014 (3), X-linked recessive; ?Olmsted syndrome, X-linked, 300918 (3), X-linked recessive
<i>MC1R</i>	155555	[Skin/hair/eye pigmentation 2, blond hair/fair skin], 266300 (3), Autosomal recessive; {Albinism, oculocutaneous, type II, modifier of}, 203200 (3), Autosomal recessive; {Melanoma, cutaneous malignant, 5}, 613099 (3); [Analgesia from kappa-opioid receptor agonist, female-specific], 613098 (3); {UV-induced skin damage}, 266300 (3), Autosomal recessive; [Skin/hair/eye pigmentation 2, red hair/fair skin], 266300 (3), Autosomal recessive
<i>MC2R</i>	607397	Glucocorticoid deficiency, due to ACTH unresponsiveness, 202200 (3), Autosomal recessive
<i>MC3R</i>	155540	{Obesity, severe, susceptibility to, BMIQ9}, 602025 (3)

<i>MC4R</i>	155541	Obesity (BMIQ20), 618406 (3), Autosomal recessive, Autosomal dominant; {Obesity, resistance to (BMIQ20)}, 618406 (3), Autosomal recessive, Autosomal dominant
<i>MCC</i>	159350	Colorectal cancer, somatic, 114500 (3)
<i>MCCC1</i>	609010	3-Methylcrotonyl-CoA carboxylase 1 deficiency, 210200 (3), Autosomal recessive
<i>MCCC2</i>	609014	3-Methylcrotonyl-CoA carboxylase 2 deficiency, 210210 (3), Autosomal recessive
<i>MCEE</i>	608419	Methylmalonyl-CoA epimerase deficiency, 251120 (3), Autosomal recessive
<i>MCFD2</i>	607788	Factor V and factor VIII, combined deficiency of, 613625 (3)
<i>MCIDAS</i>	614086	Ciliary dyskinesia, primary, 42, 618695 (3), Autosomal recessive
<i>MCM2</i>	116945	?Deafness, autosomal dominant 70, 616968 (3), Autosomal dominant
<i>MCM3AP</i>	603294	Peripheral neuropathy, autosomal recessive, with or without impaired intellectual development, 618124 (3), Autosomal recessive
<i>MCM4</i>	602638	Immunodeficiency 54, 609981 (3), Autosomal recessive
<i>MCM5</i>	602696	?Meier-Gorlin syndrome 8, 617564 (3), Autosomal recessive
<i>MCM6</i>	601806	Lactase persistence/nonpersistence, 223100 (3), Autosomal dominant
<i>MCM8</i>	608187	?Premature ovarian failure 10, 612885 (3), Autosomal recessive
<i>MCM9</i>	610098	Ovarian dysgenesis 4, 616185 (3), Autosomal recessive
<i>MCOLN1</i>	605248	Mucopolipidosis IV, 252650 (3), Autosomal recessive
<i>MCPH1</i>	607117	Microcephaly 1, primary, autosomal recessive, 251200 (3), Autosomal recessive
<i>MDH1</i>	154200	?Epileptic encephalopathy, early infantile, 88, 618959 (3)
<i>MDH2</i>	154100	Epileptic encephalopathy, early infantile, 51, 617339 (3), Autosomal recessive
<i>MDM2</i>	164785	{Accelerated tumor formation, susceptibility to}, 614401 (3), Autosomal dominant; ?Lessel-Kubisch syndrome, 618681 (3), Autosomal recessive
<i>MDM4</i>	602704	?Bone marrow failure syndrome 6, 618849 (3), Autosomal dominant
<i>MECOM</i>	165215	Radioulnar synostosis with amegakaryocytic thrombocytopenia 2, 616738 (3), Autosomal dominant
<i>MECP2</i>	300005	Mental retardation, X-linked syndromic, Lubs type, 300260 (3), X-linked recessive; Encephalopathy, neonatal severe, 300673 (3), X-linked recessive; Mental retardation, X-linked, syndromic 13, 300055 (3), X-linked recessive; Rett syndrome, atypical, 312750 (3), X-linked dominant; {Autism susceptibility, X-linked 3}, 300496 (3), X-linked; Rett syndrome, 312750 (3), X-linked dominant; Rett syndrome, preserved speech variant, 312750 (3), X-linked dominant
<i>MECR</i>	608205	Dystonia, childhood-onset, with optic atrophy and basal ganglia abnormalities, 617282 (3), Autosomal recessive

<i>MED12</i>	300188	Ohdo syndrome, X-linked, 300895 (3), X-linked recessive; Lujan-Fryns syndrome, 309520 (3), X-linked recessive; Opitz-Kaveggia syndrome, 305450 (3), X-linked recessive
<i>MED12L</i>	611318	Nizon-Isidor syndrome, 618872 (3), Autosomal dominant
<i>MED13</i>	603808	Intellectual developmental disorder 61, 618009 (3), Autosomal dominant
<i>MED13L</i>	608771	Transposition of the great arteries, dextro-looped 1, 608808 (3), Autosomal dominant; Mental retardation and distinctive facial features with or without cardiac defects, 616789 (3), Autosomal dominant
<i>MED17</i>	603810	Microcephaly, postnatal progressive, with seizures and brain atrophy, 613668 (3), Autosomal recessive
<i>MED23</i>	605042	Mental retardation, autosomal recessive 18, 614249 (3), Autosomal recessive
<i>MED25</i>	610197	Basel-Vanagait-Smirin-Yosef syndrome, 616449 (3), Autosomal recessive
<i>MEF2A</i>	600660	{Coronary artery disease, autosomal dominant, 1}, 608320 (3), Autosomal dominant
<i>MEF2C</i>	600662	Chromosome 5q14.3 deletion syndrome, 613443 (4), Autosomal dominant; Mental retardation, stereotypic movements, epilepsy, and/or cerebral malformations, 613443 (3), Autosomal dominant
<i>MEFV</i>	608107	Neutrophilic dermatosis, acute febrile, 608068 (3), Autosomal dominant; Familial Mediterranean fever, AR, 249100 (3), Autosomal recessive; Familial Mediterranean fever, AD, 134610 (3), Autosomal dominant
<i>MEGF10</i>	612453	Myopathy, areflexia, respiratory distress, and dysphagia, early-onset, mild variant, 614399 (3), Autosomal recessive; Myopathy, areflexia, respiratory distress, and dysphagia, early-onset, 614399 (3), Autosomal recessive
<i>MEGF8</i>	604267	Carpenter syndrome 2, 614976 (3), Autosomal recessive
<i>MEI1</i>	608797	Hydatidiform mole, recurrent, 3, 618431 (3), Autosomal recessive
<i>MEIOB</i>	617670	?Spermatogenic failure 22, 617706 (3), Autosomal recessive
<i>MEIS2</i>	601740	Cleft palate, cardiac defects, and mental retardation, 600987 (3), Autosomal dominant
<i>MEN1</i>	613733	Angiofibroma, somatic (3); Adrenal adenoma, somatic (3); Parathyroid adenoma, somatic (3); Lipoma, somatic (3); Carcinoid tumor of lung (3); Multiple endocrine neoplasia 1, 131100 (3), Autosomal dominant
<i>MEOX1</i>	600147	Klippel-Feil syndrome 2, 214300 (3), Autosomal recessive
<i>MERTK</i>	604705	Retinitis pigmentosa 38, 613862 (3), Autosomal recessive
<i>MESD</i>	607783	Osteogenesis imperfecta, type XX, 618644 (3), Autosomal recessive
<i>MESP2</i>	605195	Spondylocostal dysostosis 2, autosomal recessive, 608681 (3), Autosomal recessive

<i>MET</i>	164860	{Osteofibrous dysplasia, susceptibility to}, 607278 (3), Autosomal dominant; Hepatocellular carcinoma, childhood type, somatic, 114550 (3); ?Deafness, autosomal recessive 97, 616705 (3), Autosomal recessive; Renal cell carcinoma, papillary, 1, familial and somatic, 605074 (3)
<i>METTL23</i>	615262	Mental retardation, autosomal recessive 44, 615942 (3), Autosomal recessive
<i>METTL5</i>	618628	Intellectual developmental disorder, autosomal recessive 72, 618665 (3), Autosomal recessive
<i>MFAP5</i>	601103	Aortic aneurysm, familial thoracic 9, 616166 (3), Autosomal dominant
<i>MFF</i>	614785	Encephalopathy due to defective mitochondrial and peroxisomal fission 2, 617086 (3), Autosomal recessive
<i>MFHAS1</i>	605352	Malignant fibrous histiocytoma, 605352 (2)
<i>MFN2</i>	608507	Hereditary motor and sensory neuropathy VIA, 601152 (3), Autosomal dominant; Charcot-Marie-Tooth disease, axonal, type 2A2B, 617087 (3), Autosomal recessive; Charcot-Marie-Tooth disease, axonal, type 2A2A, 609260 (3), Autosomal dominant
<i>MFRP</i>	606227	Nanophthalmos 2, 609549 (3); Microphthalmia, isolated 5, 611040 (3), Autosomal recessive
<i>MFSD2A</i>	614397	Neurodevelopmental disorder with progressive microcephaly, spasticity, and brain imaging abnormalities, 616486 (3), Autosomal recessive
<i>MFSD8</i>	611124	Macular dystrophy with central cone involvement, 616170 (3), Autosomal recessive; Ceroid lipofuscinosis, neuronal, 7, 610951 (3), Autosomal recessive
<i>MGAT2</i>	602616	Congenital disorder of glycosylation, type IIa, 212066 (3), Autosomal recessive
<i>MGME1</i>	615076	Mitochondrial DNA depletion syndrome 11, 615084 (3), Autosomal recessive
<i>MGP</i>	154870	Keutel syndrome, 245150 (3), Autosomal recessive
<i>MIB1</i>	608677	Left ventricular noncompaction 7, 615092 (3), Autosomal dominant
<i>MICOS13 (C19orf70)</i>	616658	Combined oxidative phosphorylation deficiency 37, 618329 (3), Autosomal recessive
<i>MICU1</i>	605084	Myopathy with extrapyramidal signs, 615673 (3), Autosomal recessive
<i>MID1</i>	300552	Opitz GBBB syndrome, type I, 300000 (3), X-linked recessive
<i>MID2</i>	300204	?Mental retardation, X-linked 101, 300928 (3), X-linked recessive
<i>MIF</i>	153620	{Rheumatoid arthritis, systemic juvenile, susceptibility to}, 604302 (3)
<i>MINPP1</i>	605391	{Thyroid carcinoma, follicular}, 188470 (3), Autosomal dominant, Somatic mutation
<i>MIP</i>	154050	Cataract 15, multiple types, 615274 (3), Autosomal dominant
<i>MIPEP</i>	602241	Combined oxidative phosphorylation deficiency 31, 617228 (3), Autosomal recessive

<i>MIR2861</i>	613405	[Bone mineral density QTL 15], 613418 (3), Autosomal recessive, Autosomal dominant
<i>MITF</i>	156845	COMMAD syndrome, 617306 (3), Autosomal recessive; {Melanoma, cutaneous malignant, susceptibility to, 8}, 614456 (3); Waardenburg syndrome, type 2A, 193510 (3), Autosomal dominant; Waardenburg syndrome/ocular albinism, digenic, 103470 (3); Tietz albinism-deafness syndrome, 103500 (3), Autosomal dominant
<i>MKKS</i>	604896	Bardet-Biedl syndrome 6, 605231 (3), Autosomal recessive; McKusick-Kaufman syndrome, 236700 (3), Autosomal recessive
<i>MKRN3</i>	603856	Precocious puberty, central, 2, 615346 (3), Autosomal dominant
<i>MKS1</i>	609883	Bardet-Biedl syndrome 13, 615990 (3), Autosomal recessive; Joubert syndrome 28, 617121 (3), Autosomal recessive; Meckel syndrome 1, 249000 (3), Autosomal recessive
<i>MLC1</i>	605908	Megalencephalic leukoencephalopathy with subcortical cysts, 604004 (3), Autosomal recessive
<i>MLH1</i>	120436	Mismatch repair cancer syndrome, 276300 (3), Autosomal recessive; Colorectal cancer, hereditary nonpolyposis, type 2, 609310 (3); Muir-Torre syndrome, 158320 (3), Autosomal dominant
<i>MLH3</i>	604395	{Endometrial cancer, susceptibility to}, 608089 (3), Autosomal dominant, Somatic mutation; Colorectal cancer, somatic, 114500 (3); Colorectal cancer, hereditary nonpolyposis, type 7, 614385 (3)
<i>MLLT10</i>	602409	Leukemia, acute myeloid, 601626 (3), Autosomal dominant, Somatic mutation
<i>MLPH</i>	606526	Griscelli syndrome, type 3, 609227 (3), Autosomal recessive
<i>MLYCD</i>	606761	Malonyl-CoA decarboxylase deficiency, 248360 (3), Autosomal recessive
<i>MMAA</i>	607481	Methylmalonic aciduria, vitamin B12-responsive, 251100 (3), Autosomal recessive
<i>MMAB</i>	607568	Methylmalonic aciduria, vitamin B12-responsive, due to defect in synthesis of adenosylcobalamin, cblB complementation type, 251110 (3), Autosomal recessive
<i>MMACHC</i>	609831	Methylmalonic aciduria and homocystinuria, cblC type, 277400 (3), Autosomal recessive
<i>MMADHC</i>	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
<i>MME</i>	120520	Charcot-Marie-Tooth disease, axonal, type 2T, 617017 (3), Autosomal recessive, Autosomal dominant; ?Spinocerebellar ataxia 43, 617018 (3), Autosomal dominant

<i>MMP1</i>	120353	COPD, rate of decline of lung function in, 606963 (3); {Epidermolysis bullosa dystrophica, autosomal recessive, modifier of}, 226600 (3), Autosomal recessive
<i>MMP13</i>	600108	Metaphyseal dysplasia, Spahr type, 250400 (3), Autosomal recessive; Metaphyseal anadysplasia 1, 602111 (3), Autosomal dominant; Spondyloepimetaphyseal dysplasia, Missouri type, 602111 (3), Autosomal dominant
<i>MMP14</i>	600754	?Winchester syndrome, 277950 (3)
<i>MMP19</i>	601807	Cavitary optic disc anomalies, 611543 (3), Autosomal dominant
<i>MMP2</i>	120360	Multicentric osteolysis, nodulosis, and arthropathy, 259600 (3), Autosomal recessive
<i>MMP20</i>	604629	Amelogenesis imperfecta, type IIA2, 612529 (3), Autosomal recessive
<i>MMP21</i>	608416	Heterotaxy, visceral, 7, autosomal, 616749 (3), Autosomal recessive
<i>MMP3</i>	185250	{Coronary heart disease, susceptibility to, 6}, 614466 (3)
<i>MMP9</i>	120361	Metaphyseal anadysplasia 2, 613073 (3)
<i>MMUT</i> (<i>MUT</i>)	609058	Methylmalonic aciduria, mut(0) type, 251000 (3), Autosomal recessive
<i>MN1</i>	156100	Meningioma, 607174 (3), Autosomal dominant; CEBALID syndrome, 618774 (3), Autosomal dominant
<i>MNS1</i>	610766	Heterotaxy, visceral, 9, autosomal, with male infertility, 618948 (3), Autosomal recessive
<i>MNX1</i>	142994	Currarino syndrome, 176450 (3), Autosomal dominant
<i>MOCOS</i>	613274	Xanthinuria, type II, 603592 (3), Autosomal recessive
<i>MOCS1</i>	603707	Molybdenum cofactor deficiency A, 252150 (3), Autosomal recessive
<i>MOCS2</i>	603708	Molybdenum cofactor deficiency B, 252160 (3), Autosomal recessive
<i>MOG</i>	159465	?Narcolepsy 7, 614250 (3), Autosomal dominant
<i>MOGS</i>	601336	Congenital disorder of glycosylation, type IIb, 606056 (3), Autosomal recessive
<i>MORC2</i>	616661	Charcot-Marie-Tooth disease, axonal, type 2Z, 616688 (3), Autosomal dominant
<i>MPC1</i>	614738	Mitochondrial pyruvate carrier deficiency, 614741 (3), Autosomal recessive
<i>MPDU1</i>	604041	Congenital disorder of glycosylation, type If, 609180 (3), Autosomal recessive
<i>MPDZ</i>	603785	Hydrocephalus, congenital, 2, with or without brain or eye anomalies, 615219 (3), Autosomal recessive
<i>MPI</i>	154550	Congenital disorder of glycosylation, type Ib, 602579 (3), Autosomal recessive
<i>MPIG6B</i>	606520	?Thrombocytopenia, anemia, and myelofibrosis, 617441 (3), Autosomal recessive

<i>MPL</i>	159530	Myelofibrosis with myeloid metaplasia, somatic, 254450 (3); Thrombocytopenia, congenital amegakaryocytic, 604498 (3), Autosomal recessive; Thrombocythemia 2, 601977 (3), Autosomal dominant, Somatic mutation
<i>MPLKIP</i>	609188	Trichothiodystrophy 4, nonphotosensitive, 234050 (3), Autosomal recessive
<i>MPO</i>	606989	{Lung cancer, protection against, in smokers} (3); Myeloperoxidase deficiency, 254600 (3), Autosomal recessive; {Alzheimer disease, susceptibility to}, 104300 (3), Autosomal dominant
<i>MPV17</i>	137960	Charcot-Marie-Tooth disease, axonal, type 2EE, 618400 (3), Autosomal recessive; Mitochondrial DNA depletion syndrome 6 (hepatocerebral type), 256810 (3), Autosomal recessive
<i>MPZ</i>	159440	Charcot-Marie-Tooth disease, type 2J, 607736 (3), Autosomal dominant; Charcot-Marie-Tooth disease, type 1B, 118200 (3), Autosomal dominant; Dejerine-Sottas disease, 145900 (3), Autosomal recessive, Autosomal dominant; Hypomyelinating neuropathy, congenital, 2, 618184 (3), Autosomal dominant; Charcot-Marie-Tooth disease, dominant intermediate D, 607791 (3), Autosomal dominant; Roussy-Levy syndrome, 180800 (3), Autosomal dominant; Charcot-Marie-Tooth disease, type 2I, 607677 (3), Autosomal dominant
<i>MPZL2</i>	604873	Deafness, autosomal recessive 111, 618145 (3), Autosomal recessive
<i>MRAP</i>	609196	Glucocorticoid deficiency 2, 607398 (3), Autosomal recessive
<i>MRAP2</i>	615410	{?Obesity, susceptibility to, BMIQ18}, 615457 (3), Autosomal dominant
<i>MRAS</i>	608435	Noonan syndrome 11, 618499 (3), Autosomal dominant
<i>MRE11</i>	600814	Ataxia-telangiectasia-like disorder 1, 604391 (3), Autosomal recessive
<i>MRM2</i>	606906	?Mitochondrial DNA depletion syndrome 17, 618567 (3), Autosomal recessive
<i>MRPL12</i>	602375	?Combined oxidative phosphorylation deficiency 45, 618951 (3), Autosomal recessive
<i>MRPL3</i>	607118	Combined oxidative phosphorylation deficiency 9, 614582 (3), Autosomal recessive
<i>MRPL44</i>	611849	?Combined oxidative phosphorylation deficiency 16, 615395 (3), Autosomal recessive
<i>MRPS14</i>	611978	?Combined oxidative phosphorylation deficiency 38, 618378 (3), Autosomal recessive
<i>MRPS16</i>	609204	Combined oxidative phosphorylation deficiency 2, 610498 (3), Autosomal recessive
<i>MRPS2</i>	611971	Combined oxidative phosphorylation deficiency 36, 617950 (3), Autosomal recessive

<i>MRPS22</i>	605810	Combined oxidative phosphorylation deficiency 5, 611719 (3), Autosomal recessive; Ovarian dysgenesis 7, 618117 (3), Autosomal recessive
<i>MRPS23</i>	611985	?Combined oxidative phosphorylation deficiency 46, 618952 (3), Autosomal recessive
<i>MRPS28</i>	611990	?Combined oxidative phosphorylation deficiency 47, 618958 (3), Autosomal recessive
<i>MRPS34</i>	611994	Combined oxidative phosphorylation deficiency 32, 617664 (3), Autosomal recessive
<i>MRPS7</i>	611974	?Combined oxidative phosphorylation deficiency 34, 617872 (3), Autosomal recessive
<i>MRTFA</i>	606078	?Immunodeficiency 66, 618847 (3), Autosomal recessive
<i>MS4A1</i>	112210	Immunodeficiency, common variable, 5, 613495 (3), Autosomal recessive
<i>MS4A2</i>	147138	{Atopy, susceptibility to}, 147050 (3), Autosomal dominant
<i>MSH2</i>	609309	Mismatch repair cancer syndrome, 276300 (3), Autosomal recessive; Muir-Torre syndrome, 158320 (3), Autosomal dominant; Colorectal cancer, hereditary nonpolyposis, type 1, 120435 (3), Autosomal dominant
<i>MSH3</i>	600887	Familial adenomatous polyposis 4, 617100 (3), Autosomal recessive; Endometrial carcinoma, somatic, 608089 (3)
<i>MSH5</i>	603382	?Premature ovarian failure 13, 617442 (3), Autosomal recessive
<i>MSH6</i>	600678	Mismatch repair cancer syndrome, 276300 (3), Autosomal recessive; {Endometrial cancer, familial}, 608089 (3), Autosomal dominant, Somatic mutation; Colorectal cancer, hereditary nonpolyposis, type 5, 614350 (3), Autosomal dominant
<i>MSL3</i>	300609	Basilicata-Akhtar syndrome, 301032 (3), X-linked dominant
<i>MSMB</i>	157145	{Prostate cancer, hereditary, 13}, 611928 (3)
<i>MSMO1</i>	607545	Microcephaly, congenital cataract, and psoriasiform dermatitis, 616834 (3), Autosomal recessive
<i>MSN</i>	309845	Immunodeficiency 50, 300988 (3), X-linked recessive
<i>MSR1</i>	153622	Barrett esophagus/esophageal adenocarcinoma, 614266 (3)
<i>MSRB3</i>	613719	Deafness, autosomal recessive 74, 613718 (3), Autosomal recessive
<i>MST1R</i>	600168	{Nasopharyngeal carcinoma, susceptibility to, 3}, 617075 (3), Autosomal dominant
<i>MSTN</i>	601788	Muscle hypertrophy, 614160 (3)
<i>MSTO1</i>	617619	Myopathy, mitochondrial, and ataxia, 617675 (3), Autosomal recessive, Autosomal dominant
<i>MSX1</i>	142983	Orofacial cleft 5, 608874 (3), Autosomal dominant; Ectodermal dysplasia 3, Witkop type, 189500 (3), Autosomal dominant; Tooth agenesis, selective, 1, with or without orofacial cleft, 106600 (3), Autosomal dominant

<i>MSX2</i>	123101	Parietal foramina 1, 168500 (3), Autosomal dominant; Craniosynostosis 2, 604757 (3), Autosomal dominant; Parietal foramina with cleidocranial dysplasia, 168550 (3), Autosomal dominant
<i>MTAP</i>	156540	Diaphyseal medullary stenosis with malignant fibrous histiocytoma, 112250 (3), Autosomal dominant
<i>MTFMT</i>	611766	Combined oxidative phosphorylation deficiency 15, 614947 (3), Autosomal recessive; Mitochondrial complex I deficiency, nuclear type 27, 618248 (3), Autosomal recessive
<i>MTHFD1</i>	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
<i>MTHFR</i>	607093	{Vascular disease, susceptibility to} (3); {Schizophrenia, susceptibility to}, 181500 (3), Autosomal dominant; Homocystinuria due to MTHFR deficiency, 236250 (3), Autosomal recessive; {Neural tube defects, susceptibility to}, 601634 (3), Autosomal recessive; {Thromboembolism, susceptibility to}, 188050 (3), Autosomal dominant
<i>MTHFS</i>	604197	Neurodevelopmental disorder with microcephaly, epilepsy, and hypomyelination, 618367 (3), Autosomal recessive
<i>MTM1</i>	300415	Myotubular myopathy, X-linked, 310400 (3), X-linked recessive
<i>MTMR14</i>	611089	{Centronuclear myopathy, autosomal, modifier of}, 160150 (3), Autosomal dominant
<i>MTMR2</i>	603557	Charcot-Marie-Tooth disease, type 4B1, 601382 (3), Autosomal recessive
<i>MTNR1B</i>	600804	{Diabetes mellitus, type 2, susceptibility to}, 125853 (3), Autosomal dominant
<i>MTO1</i>	614667	Combined oxidative phosphorylation deficiency 10, 614702 (3), Autosomal recessive
<i>MTOR</i>	601231	Smith-Kingsmore syndrome, 616638 (3), Autosomal dominant; Focal cortical dysplasia, type II, somatic, 607341 (3)
<i>MTPAP</i>	613669	?Spastic ataxia 4, autosomal recessive, 613672 (3), Autosomal recessive
<i>MTR</i>	156570	{Neural tube defects, folate-sensitive, susceptibility to}, 601634 (3), Autosomal recessive; Homocystinuria-megaloblastic anemia, cblG complementation type, 250940 (3), Autosomal recessive
<i>MTRR</i>	602568	{Neural tube defects, folate-sensitive, susceptibility to}, 601634 (3), Autosomal recessive; Homocystinuria-megaloblastic anemia, cbl E type, 236270 (3), Autosomal recessive
<i>MTTP</i>	157147	Abetalipoproteinemia, 200100 (3), Autosomal recessive; {Metabolic syndrome, protection against}, 605552 (3), Autosomal dominant
<i>MUC1</i>	158340	Medullary cystic kidney disease 1, 174000 (3), Autosomal dominant

<i>MUC5B</i>	600770	{Pulmonary fibrosis, idiopathic, susceptibility to}, 178500 (3), Autosomal dominant
<i>MUC7</i>	158375	{Asthma, protection against}, 600807 (3), Autosomal dominant
<i>MUCL3</i>	613928/604809	-/Panbronchiolitis, diffuse, 604809 (2), Multifactorial
<i>MUSK</i>	601296	Fetal akinesia deformation sequence 1, 208150 (3), Autosomal recessive; Myasthenic syndrome, congenital, 9, associated with acetylcholine receptor deficiency, 616325 (3), Autosomal recessive
<i>MUTYH</i>	604933	Gastric cancer, somatic, 613659 (3); Adenomas, multiple colorectal, 608456 (3), Autosomal recessive
<i>MVD</i>	603236	Porokeratosis 7, multiple types, 614714 (3), Autosomal dominant
<i>MVK</i>	251170	Hyper-IgD syndrome, 260920 (3), Autosomal recessive; Porokeratosis 3, multiple types, 175900 (3), Autosomal dominant; Mevalonic aciduria, 610377 (3), Autosomal recessive
<i>MVP</i>	605088/157700	-/Mitral valve prolapse, myxomatous 1, 157700 (2), Autosomal dominant
<i>MXI1</i>	600020	Prostate cancer, somatic, 176807 (3); Neurofibrosarcoma, somatic (3)
<i>MYB</i>	189990	{T-cell acute lymphoblastic leukemia} (3)
<i>MYBPC1</i>	160794	Arthrogryposis, distal, type 1B, 614335 (3), Autosomal dominant; Myopathy, congenital, with tremor, 618524 (3), Autosomal dominant; Lethal congenital contracture syndrome 4, 614915 (3), Autosomal recessive
<i>MYBPC3</i>	600958	Cardiomyopathy, hypertrophic, 4, 115197 (3), Autosomal recessive, Autosomal dominant; Cardiomyopathy, dilated, 1MM, 615396 (3), Autosomal dominant; Left ventricular noncompaction 10, 615396 (3), Autosomal dominant
<i>MYC</i>	190080	Burkitt lymphoma, somatic, 113970 (3)
<i>MYCN</i>	164840	Feingold syndrome 1, 164280 (3), Autosomal dominant
<i>MYD88</i>	602170	Immunodeficiency 68, 612260 (3), Autosomal recessive; Macroglobulinemia, Waldenstrom, somatic, 153600 (3)
<i>MYF5</i>	159990	Ophthalmoplegia, external, with rib and vertebral anomalies, 618155 (3), Autosomal recessive
<i>MYH11</i>	160745	Aortic aneurysm, familial thoracic 4, 132900 (3), Autosomal dominant
<i>MYH14</i>	608568	?Peripheral neuropathy, myopathy, hoarseness, and hearing loss, 614369 (3), Autosomal dominant; Deafness, autosomal dominant 4A, 600652 (3), Autosomal dominant
<i>MYH2</i>	160740	Proximal myopathy and ophthalmoplegia, 605637 (3), Autosomal recessive, Autosomal dominant

<i>MYH3</i>	160720	Contractures, pterygia, and spondylcarpotarsal fusion syndrome 1B, 618469 (3), Autosomal recessive; Arthrogryposis, distal, type 2B3 (Sheldon-Hall), 618436 (3), Autosomal dominant; Arthrogryposis, distal, type 2A (Freeman-Sheldon), 193700 (3), Autosomal dominant; Contractures, pterygia, and spondylcarpostarsal fusion syndrome 1A, 178110 (3), Autosomal dominant
<i>MYH6</i>	160710	Atrial septal defect 3, 614089 (3); Cardiomyopathy, hypertrophic, 14, 613251 (3), Autosomal dominant; {Sick sinus syndrome 3}, 614090 (3); Cardiomyopathy, dilated, 1EE, 613252 (3)
<i>MYH7</i>	160760	Myopathy, myosin storage, autosomal recessive, 255160 (3), Autosomal recessive; Left ventricular noncompaction 5, 613426 (3), Autosomal dominant; Laing distal myopathy, 160500 (3), Autosomal dominant; Myopathy, myosin storage, autosomal dominant, 608358 (3), Autosomal dominant; Cardiomyopathy, dilated, 1S, 613426 (3), Autosomal dominant; Scapuloperoneal syndrome, myopathic type, 181430 (3), Autosomal dominant; Cardiomyopathy, hypertrophic, 1, 192600 (3), Digenic dominant, Autosomal dominant
<i>MYH8</i>	160741	Carney complex variant, 608837 (3); Trismus-pseudocamptodactyly syndrome, 158300 (3), Autosomal dominant
<i>MYH9</i>	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
<i>MYL1</i>	160780	Myopathy, congenital, with fast-twitch (type II) fiber atrophy, 618414 (3), Autosomal recessive
<i>MYL2</i>	160781	Cardiomyopathy, hypertrophic, 10, 608758 (3), Autosomal dominant
<i>MYL3</i>	160790	Cardiomyopathy, hypertrophic, 8, 608751 (3), Autosomal recessive, Autosomal dominant
<i>MYL4</i>	160770	?Atrial fibrillation, familial, 18, 617280 (3), Autosomal dominant
<i>MYLK</i>	600922	Aortic aneurysm, familial thoracic 7, 613780 (3), Autosomal dominant; Megacystis-microcolon-intestinal hypoperistalsis syndrome, 249210 (3), Autosomal recessive
<i>MYLK2</i>	606566	Cardiomyopathy, hypertrophic, 1, digenic, 192600 (3), Digenic dominant, Autosomal dominant
<i>MYMK</i>	615345	Carey-Fineman-Ziter syndrome, 254940 (3), Autosomal recessive
<i>MYO15A</i>	602666	Deafness, autosomal recessive 3, 600316 (3), Autosomal recessive
<i>MYO18B</i>	607295	Klippel-Feil syndrome 4, autosomal recessive, with myopathy and facial dysmorphism, 616549 (3), Autosomal recessive
<i>MYO1A</i>	601478/607841	-/Deafness, autosomal dominant 48, 607841 (2), Autosomal dominant

<i>MYO1E</i>	601479	Glomerulosclerosis, focal segmental, 6, 614131 (3), Autosomal recessive
<i>MYO3A</i>	606808	Deafness, autosomal recessive 30, 607101 (3), Autosomal recessive
<i>MYO5A</i>	160777	Griscelli syndrome, type 1, 214450 (3), Autosomal recessive
<i>MYO5B</i>	606540	Microvillus inclusion disease, 251850 (3), Autosomal recessive
<i>MYO6</i>	600970	Deafness, autosomal recessive 37, 607821 (3), Autosomal recessive; Deafness, autosomal dominant 22, 606346 (3), Autosomal dominant; Deafness, autosomal dominant 22, with hypertrophic cardiomyopathy, 606346 (3), Autosomal dominant
<i>MYO7A</i>	276903	Deafness, autosomal recessive 2, 600060 (3), Autosomal recessive; Deafness, autosomal dominant 11, 601317 (3), Autosomal dominant; Usher syndrome, type 1B, 276900 (3), Autosomal recessive
<i>MYO9A</i>	604875	Myasthenic syndrome, congenital, 24, presynaptic, 618198 (3), Autosomal recessive
<i>MYO9B</i>	602129	{Celiac disease, susceptibility to, 4}, 609753 (3)
<i>MYOC</i>	601652	Glaucoma 1A, primary open angle, 137750 (3), Autosomal dominant
<i>MYOCD</i>	606127	Megabladder, congenital, 618719 (3), Autosomal dominant
<i>MYOD1</i>	159970	Myopathy, congenital, with diaphragmatic defects, respiratory insufficiency, and dysmorphic facies, 618975 (3)
<i>MYORG</i>	618255	Basal ganglia calcification, idiopathic, 7, autosomal recessive, 618317 (3), Autosomal recessive
<i>MYOT</i>	604103	Myopathy, myofibrillar, 3, 609200 (3), Autosomal dominant; Myopathy, spheroid body, 182920 (3), Autosomal dominant
<i>MYOZ2</i>	605602	Cardiomyopathy, hypertrophic, 16, 613838 (3), Autosomal dominant
<i>MYPN</i>	608517	Cardiomyopathy, dilated, 1KK, 615248 (3), Autosomal dominant; Cardiomyopathy, familial restrictive, 4, 615248 (3), Autosomal dominant; Nemaline myopathy 11, autosomal recessive, 617336 (3), Autosomal recessive; Cardiomyopathy, hypertrophic, 22, 615248 (3), Autosomal dominant
<i>MYRF</i>	608329	Cardiac-urogenital syndrome, 618280 (3), Autosomal dominant; Encephalitis/encephalopathy, mild, with reversible myelin vacuolization, 618113 (3), Autosomal dominant
<i>MYSM1</i>	612176	Bone marrow failure syndrome 4, 618116 (3), Autosomal recessive
<i>MYT1L</i>	613084	Mental retardation, autosomal dominant 39, 616521 (3), Autosomal dominant
<i>NAA10</i>	300013	Ogden syndrome, 300855 (3), X-linked recessive, X-linked dominant; Microphthalmia, syndromic 1, 309800 (3), X-linked
<i>NAA15</i>	608000	Mental retardation, autosomal dominant 50, 617787 (3), Autosomal dominant

<i>NACC1</i>	610672	Neurodevelopmental disorder with epilepsy, cataracts, feeding difficulties, and delayed brain myelination, 617393 (3), Autosomal dominant
<i>NADK2</i>	615787	2,4-dienoyl-CoA reductase deficiency, 616034 (3), Autosomal recessive
<i>NADSYN1</i>	608285	Vertebral, cardiac, renal, and limb defects syndrome 3, 618845 (3), Autosomal recessive
<i>NAGA</i>	104170	Kanzaki disease, 609242 (3), Autosomal recessive; Schindler disease, type I, 609241 (3), Autosomal recessive; Schindler disease, type III, 609241 (3), Autosomal recessive
<i>NAGLU</i>	609701	Mucopolysaccharidosis type IIIB (Sanfilippo B), 252920 (3), Autosomal recessive; ?Charcot-Marie-Tooth disease, axonal, type 2V, 616491 (3), Autosomal dominant
<i>NAGS</i>	608300	N-acetylglutamate synthase deficiency, 237310 (3), Autosomal recessive
<i>NALCN</i>	611549	Hypotonia, infantile, with psychomotor retardation and characteristic facies 1, 615419 (3), Autosomal recessive; Congenital contractures of the limbs and face, hypotonia, and developmental delay, 616266 (3), Autosomal dominant
<i>NANOS1</i>	608226	Spermatogenic failure 12, 615413 (3), Autosomal dominant
<i>NANS</i>	605202	Spondyloepimetaphyseal dysplasia, Camera-Genevieve type, 610442 (3), Autosomal recessive
<i>NARS2</i>	612803	?Deafness, autosomal recessive 94, 618434 (3), Autosomal recessive; Combined oxidative phosphorylation deficiency 24, 616239 (3), Autosomal recessive
<i>NAT2</i>	612182	[Acetylation, slow], 243400 (3), Autosomal recessive
<i>NAT8L</i>	610647	?N-acetylaspartate deficiency, 614063 (3), Autosomal recessive
<i>NAXD</i>	615910	Encephalopathy, progressive, early-onset, with brain edema and/or leukoencephalopathy, 2, 618321 (3), Autosomal recessive
<i>NAXE</i>	608862	Encephalopathy, progressive, early-onset, with brain edema and/or leukoencephalopathy, 617186 (3), Autosomal recessive
<i>NBAS</i>	608025	Infantile liver failure syndrome 2, 616483 (3), Autosomal recessive; Short stature, optic nerve atrophy, and Pelger-Huet anomaly, 614800 (3), Autosomal recessive
<i>NBEAL2</i>	614169	Gray platelet syndrome, 139090 (3), Autosomal recessive
<i>NBN</i>	602667	Leukemia, acute lymphoblastic, 613065 (3); Nijmegen breakage syndrome, 251260 (3), Autosomal recessive; Aplastic anemia, 609135 (3)
<i>NCAPD2</i>	615638	?Microcephaly 21, primary, autosomal recessive, 617983 (3), Autosomal recessive
<i>NCAPD3</i>	609276	Microcephaly 22, primary, autosomal recessive, 617984 (3), Autosomal recessive
<i>NCAPG2</i>	608532	Khan-Khan-Katsanis syndrome, 618460 (3), Autosomal recessive

<i>NCAPH</i>	602332	?Microcephaly 23, primary, autosomal recessive, 617985 (3), Autosomal recessive
<i>NCF1</i>	608512	Chronic granulomatous disease 1, autosomal recessive, 233700 (3), Autosomal recessive
<i>NCF2</i>	608515	Chronic granulomatous disease due to deficiency of NCF-2, 233710 (3), Autosomal recessive
<i>NCF4</i>	601488	Chronic granulomatous disease 3, autosomal recessive, 613960 (3), Autosomal recessive
<i>NCKAP1L</i>	141180	Immunodeficiency 72 with autoinflammation, 618982 (3)
<i>NCR3</i>	611550	{Malaria, mild, susceptibility to}, 609148 (3)
<i>NCSTN</i>	605254	Acne inversa, familial, 1, 142690 (3), Autosomal dominant
<i>NDE1</i>	609449	Lissencephaly 4 (with microcephaly), 614019 (3), Autosomal recessive; ?Microhydranencephaly, 605013 (3), Autosomal recessive
<i>NDN</i>	602117	Prader-Willi syndrome, 176270 (3), Autosomal dominant
<i>NDNF</i>	616506	Hypogonadotropic hypogonadism 25 with anosmia, 618841 (3), Autosomal dominant
<i>NDP</i>	300658	Exudative vitreoretinopathy 2, X-linked, 305390 (3), X-linked recessive, X-linked dominant; Norrie disease, 310600 (3), X-linked recessive
<i>NDRG1</i>	605262	Charcot-Marie-Tooth disease, type 4D, 601455 (3), Autosomal recessive
<i>NDST1</i>	600853	Mental retardation, autosomal recessive 46, 616116 (3), Autosomal recessive
<i>NDUFA1</i>	300078	Mitochondrial complex I deficiency, nuclear type 12, 301020 (3), X-linked recessive
<i>NDUFA10</i>	603835	Mitochondrial complex I deficiency, nuclear type 22, 618243 (3), Autosomal recessive
<i>NDUFA11</i>	612638	Mitochondrial complex I deficiency, nuclear type 14, 618236 (3), Autosomal recessive
<i>NDUFA12</i>	614530	?Mitochondrial complex I deficiency, nuclear type 23, 618244 (3), Autosomal recessive
<i>NDUFA13</i>	609435	?Mitochondrial complex I deficiency, nuclear type 28, 618249 (3), Autosomal recessive; {Thyroid carcinoma, Hurthle cell}, 607464 (3)
<i>NDUFA2</i>	602137	?Mitochondrial complex I deficiency, nuclear type 13, 618235 (3), Autosomal recessive
<i>NDUFA6</i>	602138	Mitochondrial complex I deficiency, nuclear type 33, 618253 (3), Autosomal recessive
<i>NDUFA9</i>	603834	Mitochondrial complex I deficiency, nuclear type 26, 618247 (3), Autosomal recessive
<i>NDUFAF1</i>	606934	Mitochondrial complex I deficiency, nuclear type 11, 618234 (3), Autosomal recessive
<i>NDUFAF2</i>	609653	Mitochondrial complex I deficiency, nuclear type 10, 618233 (3), Autosomal recessive

<i>NDUFAF3</i>	612911	Mitochondrial complex I deficiency, nuclear type 18, 618240 (3), Autosomal recessive
<i>NDUFAF4</i>	611776	Mitochondrial complex I deficiency, nuclear type 15, 618237 (3), Autosomal recessive
<i>NDUFAF5</i>	612360	Mitochondrial complex I deficiency, nuclear type 16, 618238 (3), Autosomal recessive
<i>NDUFAF6</i>	612392	Fanconi renotubular syndrome 5, 618913 (3), Autosomal recessive; Mitochondrial complex I deficiency, nuclear type 17, 618239 (3), Autosomal recessive
<i>NDUFAF8</i>	618461	Mitochondrial complex I deficiency, nuclear type 34, 618776 (3), Autosomal recessive
<i>NDUFB11</i>	300403	Linear skin defects with multiple congenital anomalies 3, 300952 (3), X-linked dominant; ?Mitochondrial complex I deficiency, nuclear type 30, 301021 (3), X-linked
<i>NDUFB3</i>	603839	Mitochondrial complex I deficiency, nuclear type 25, 618246 (3), Autosomal recessive
<i>NDUFB8</i>	602140	Mitochondrial complex I deficiency, nuclear type 32, 618252 (3), Autosomal recessive
<i>NDUFB9</i>	601445	?Mitochondrial complex I deficiency, nuclear type 24, 618245 (3), Autosomal recessive
<i>NDUFS1</i>	157655	Mitochondrial complex I deficiency, nuclear type 5, 618226 (3), Autosomal recessive
<i>NDUFS2</i>	602985	Mitochondrial complex I deficiency, nuclear type 6, 618228 (3), Autosomal recessive
<i>NDUFS3</i>	603846	Mitochondrial complex I deficiency, nuclear type 8, 618230 (3), Autosomal recessive
<i>NDUFS4</i>	602694	Mitochondrial complex I deficiency, nuclear type 1, 252010 (3), Autosomal recessive
<i>NDUFS6</i>	603848	Mitochondrial complex I deficiency, nuclear type 9, 618232 (3), Autosomal recessive
<i>NDUFS7</i>	601825	Mitochondrial complex I deficiency, nuclear type 3, 618224 (3), Autosomal recessive
<i>NDUFS8</i>	602141	Mitochondrial complex I deficiency, nuclear type 2, 618222 (3), Autosomal recessive
<i>NDUFV1</i>	161015	Mitochondrial complex I deficiency, nuclear type 4, 618225 (3), Autosomal recessive
<i>NDUFV2</i>	600532	Mitochondrial complex I deficiency, nuclear type 7, 618229 (3), Autosomal recessive
<i>NEB</i>	161650	Nemaline myopathy 2, autosomal recessive, 256030 (3), Autosomal recessive
<i>NECAP1</i>	611623	Epileptic encephalopathy, early infantile, 21, 615833 (3), Autosomal recessive
<i>NECTIN1</i>	600644	Orofacial cleft 7, 225060 (3), Autosomal recessive; Cleft lip/palate-ectodermal dysplasia syndrome, 225060 (3), Autosomal recessive

<i>NECTIN4</i>	609607	Ectodermal dysplasia-syndactyly syndrome 1, 613573 (3), Autosomal recessive
<i>NEDD4L</i>	606384	Periventricular nodular heterotopia 7, 617201 (3), Autosomal dominant
<i>NEFH</i>	162230	Charcot-Marie-Tooth disease, axonal, type 2CC, 616924 (3), Autosomal dominant; ?{Amyotrophic lateral sclerosis, susceptibility to}, 105400 (3), Autosomal recessive, Autosomal dominant
<i>NEFL</i>	162280	Charcot-Marie-Tooth disease, type 1F, 607734 (3), Autosomal recessive, Autosomal dominant; Charcot-Marie-Tooth disease, dominant intermediate G, 617882 (3), Autosomal dominant; Charcot-Marie-Tooth disease, type 2E, 607684 (3), Autosomal dominant
<i>NEK1</i>	604588	{Amyotrophic lateral sclerosis, susceptibility to, 24}, 617892 (3), Autosomal dominant; Short-rib thoracic dysplasia 6 with or without polydactyly, 263520 (3), Autosomal recessive, Digenic recessive
<i>NEK10</i>	618726	Ciliary dyskinesia, primary, 44, 618781 (3), Autosomal recessive
<i>NEK2</i>	604043	?Retinitis pigmentosa 67, 615565 (3), Autosomal recessive
<i>NEK8</i>	609799	?Nephronophthisis 9, 613824 (3); Renal-hepatic-pancreatic dysplasia 2, 615415 (3), Autosomal recessive
<i>NEK9</i>	609798	Lethal congenital contracture syndrome 10, 617022 (3), Autosomal recessive; Nevus comedonicus, somatic, 617025 (3); ?Arthrogryposis, Perthes disease, and upward gaze palsy, 614262 (3), Autosomal recessive
<i>NEPRO</i>	617089	Anauxetic dysplasia 3, 618853 (3), Autosomal recessive
<i>NEU1</i>	608272	Sialidosis, type II, 256550 (3), Autosomal recessive; Sialidosis, type I, 256550 (3), Autosomal recessive
<i>NEUROD1</i>	601724	Maturity-onset diabetes of the young 6, 606394 (3); {Diabetes mellitus, noninsulin-dependent}, 125853 (3), Autosomal dominant
<i>NEUROD2</i>	601725	Epileptic encephalopathy, early infantile, 72, 618374 (3), Autosomal dominant
<i>NEUROG3</i>	604882	Diarrhea 4, malabsorptive, congenital, 610370 (3), Autosomal recessive
<i>NEXMIF</i>	300524	Mental retardation, X-linked 98, 300912 (3), X-linked dominant
<i>NEXN</i>	613121	Cardiomyopathy, hypertrophic, 20, 613876 (3), Autosomal dominant; Cardiomyopathy, dilated, 1CC, 613122 (3), Autosomal dominant
<i>NF1</i>	613113	Neurofibromatosis-Noonan syndrome, 601321 (3), Autosomal dominant; Leukemia, juvenile myelomonocytic, 607785 (3), Autosomal dominant, Somatic mutation; Neurofibromatosis, familial spinal, 162210 (3), Autosomal dominant; Watson syndrome, 193520 (3), Autosomal dominant; Neurofibromatosis, type 1, 162200 (3), Autosomal dominant

<i>NF2</i>	607379	Meningioma, NF2-related, somatic, 607174 (3); Schwannomatosis, somatic, 162091 (3); Neurofibromatosis, type 2, 101000 (3), Autosomal dominant
<i>NFASC</i>	609145	Neurodevelopmental disorder with central and peripheral motor dysfunction, 618356 (3), Autosomal recessive
<i>NFE2L2</i>	600492	Immunodeficiency, developmental delay, and hypohomocysteinemia, 617744 (3), Autosomal dominant
<i>NFIA</i>	600727	Brain malformations with or without urinary tract defects, 613735 (3), Autosomal dominant
<i>NFIB</i>	600728	Macrocephaly, acquired, with impaired intellectual development, 618286 (3), Autosomal dominant
<i>NFIX</i>	164005	Marshall-Smith syndrome, 602535 (3), Autosomal dominant; Sotos syndrome 2, 614753 (3), Autosomal dominant
<i>NFKB1</i>	164011	Immunodeficiency, common variable, 12, 616576 (3), Autosomal dominant
<i>NFKB2</i>	164012	Immunodeficiency, common variable, 10, 615577 (3), Autosomal dominant
<i>NFKBIA</i>	164008	Ectodermal dysplasia and immunodeficiency 2, 612132 (3), Autosomal dominant
<i>NFKBIL1</i>	601022	{Rheumatoid arthritis, susceptibility to}, 180300 (3)
<i>NFU1</i>	608100	Multiple mitochondrial dysfunctions syndrome 1, 605711 (3), Autosomal recessive
<i>NGF</i>	162030	Neuropathy, hereditary sensory and autonomic, type V, 608654 (3), Autosomal recessive
<i>NGLY1</i>	610661	Congenital disorder of deglycosylation, 615273 (3), Autosomal recessive
<i>NHEJ1</i>	611290	Severe combined immunodeficiency with microcephaly, growth retardation, and sensitivity to ionizing radiation, 611291 (3)
<i>NHLRC1</i>	608072	Epilepsy, progressive myoclonic 2B (Lafora), 254780 (3), Autosomal recessive
<i>NHLRC2</i>	618277	FINCA syndrome, 618278 (3), Autosomal recessive
<i>NHP2</i>	606470	Dyskeratosis congenita, autosomal recessive 2, 613987 (3), Autosomal recessive
<i>NHS</i>	300457	Nance-Horan syndrome, 302350 (3), X-linked dominant; Cataract 40, X-linked, 302200 (3), X-linked
<i>NIN</i>	608684	?Seckel syndrome 7, 614851 (3), Autosomal recessive
<i>NIPA1</i>	608145	Spastic paraplegia 6, autosomal dominant, 600363 (3), Autosomal dominant
<i>NIPAL4</i>	609383	Ichthyosis, congenital, autosomal recessive 6, 612281 (3), Autosomal recessive
<i>NIPBL</i>	608667	Cornelia de Lange syndrome 1, 122470 (3), Autosomal dominant
<i>NKAP</i>	300766	Intellectual developmental disorder, X-linked, syndromic, Hackman-Di Donato type, 301039 (3), X-linked recessive

<i>NKX2-1</i>	600635	{Thyroid cancer, nonmedullary, 1}, 188550 (3), Autosomal dominant; Chorea, hereditary benign, 118700 (3), Autosomal dominant; Choreoathetosis, hypothyroidism, and neonatal respiratory distress, 610978 (3), Autosomal dominant
<i>NKX2-5</i>	600584	Atrial septal defect 7, with or without AV conduction defects, 108900 (3), Autosomal dominant; Ventricular septal defect 3, 614432 (3), Autosomal dominant; Hypoplastic left heart syndrome 2, 614435 (3), Autosomal dominant; Conotruncal heart malformations, variable, 217095 (3); Tetralogy of Fallot, 187500 (3), Autosomal dominant; Hypothyroidism, congenital nongoitrous, 5, 225250 (3), Autosomal dominant
<i>NKX2-6</i>	611770	Persistent truncus arteriosus, 217095 (3); Conotruncal heart malformations, 217095 (3)
<i>NKX3-2</i>	602183	Spondylo-megaepiphyseal-metaphyseal dysplasia, 613330 (3), Autosomal recessive
<i>NKX6-2</i>	605955	Spastic ataxia 8, autosomal recessive, with hypomyelinating leukodystrophy, 617560 (3), Autosomal recessive
<i>NLGN1</i>	600568	{Autism, susceptibility to, 20}, 618830 (3), Autosomal dominant
<i>NLGN3</i>	300336	{Autism susceptibility, X-linked 1}, 300425 (3), X-linked; {Asperger syndrome susceptibility, X-linked 1}, 300494 (3), Isolated cases, X-linked, Multifactorial
<i>NLGN4X</i>	300427	Mental retardation, X-linked, 300495 (3), Isolated cases, X-linked, Multifactorial; {Asperger syndrome susceptibility, X-linked 2}, 300497 (3), X-linked; {Autism susceptibility, X-linked 2}, 300495 (3), Isolated cases, X-linked, Multifactorial
<i>NLRC4</i>	606831	Autoinflammation with infantile enterocolitis, 616050 (3), Autosomal dominant; ?Familial cold autoinflammatory syndrome 4, 616115 (3), Autosomal dominant
<i>NLRP1</i>	606636	Palmoplantar carcinoma, multiple self-healing, 615225 (3), Autosomal dominant; Autoinflammation with arthritis and dyskeratosis, 617388 (3), Autosomal recessive, Autosomal dominant; {Vitiligo-associated multiple autoimmune disease susceptibility 1}, 606579 (3); ?Respiratory papillomatosis, juvenile recurrent, congenital, 618803 (3), Autosomal recessive
<i>NLRP12</i>	609648	Familial cold autoinflammatory syndrome 2, 611762 (3), Autosomal dominant
<i>NLRP3</i>	606416	Familial cold inflammatory syndrome 1, 120100 (3), Autosomal dominant; Muckle-Wells syndrome, 191900 (3), Autosomal dominant; CINCA syndrome, 607115 (3), Autosomal dominant; Deafness, autosomal dominant 34, with or without inflammation, 617772 (3), Autosomal dominant; Keratoendothelitis fugax hereditaria, 148200 (3), Autosomal dominant
<i>NLRP7</i>	609661	Hydatidiform mole, recurrent, 1, 231090 (3), Autosomal recessive
<i>NME8</i>	607421	Ciliary dyskinesia, primary, 6, 610852 (3), Autosomal recessive

<i>NMNAT1</i>	608700	Leber congenital amaurosis 9, 608553 (3), Autosomal recessive
<i>NNMT</i>	600008	Homocysteine plasma level, 600008 (2)
<i>NNT</i>	607878	Glucocorticoid deficiency 4, with or without mineralocorticoid deficiency, 614736 (3), Autosomal recessive
<i>NOBOX</i>	610934	Premature ovarian failure 5, 611548 (3), Autosomal dominant
<i>NOD2</i>	605956	{Yao syndrome}, 617321 (3), Multifactorial; Blau syndrome, 186580 (3), Autosomal dominant; {Inflammatory bowel disease 1, Crohn disease}, 266600 (3), Multifactorial
<i>NODAL</i>	601265	Heterotaxy, visceral, 5, 270100 (3), Autosomal dominant
<i>NOG</i>	602991	Tarsal-carpal coalition syndrome, 186570 (3), Autosomal dominant; Symphalangism, proximal, 1A, 185800 (3), Autosomal dominant; Stapes ankylosis with broad thumbs and toes, 184460 (3), Autosomal dominant; Multiple synostoses syndrome 1, 186500 (3), Autosomal dominant; Brachydactyly, type B2, 611377 (3), Autosomal dominant
<i>NOL3</i>	605235	?Myoclonus, familial, 1, 614937 (3), Autosomal dominant
<i>NONO</i>	300084	Mental retardation, X-linked, syndromic 34, 300967 (3), X-linked
<i>NOP10</i>	606471	Dyskeratosis congenita, autosomal recessive 1, 224230 (3), Autosomal recessive
<i>NOP56</i>	614154	Spinocerebellar ataxia 36, 614153 (3), Autosomal dominant
<i>NOS2</i>	163730	{Malaria, resistance to}, 611162 (3)
<i>NOS3</i>	163729	{Coronary artery spasm 1, susceptibility to} (3); {Alzheimer disease, late-onset, susceptibility to}, 104300 (3), Autosomal dominant; {Hypertension, susceptibility to}, 145500 (3), Multifactorial; {Placental abruption} (3); {Ischemic stroke, susceptibility to}, 601367 (3), Multifactorial; {Hypertension, pregnancy-induced}, 189800 (3), Autosomal dominant
<i>NOTCH1</i>	190198	Aortic valve disease 1, 109730 (3), Autosomal dominant; Adams-Oliver syndrome 5, 616028 (3), Autosomal dominant
<i>NOTCH2</i>	600275	Hajdu-Cheney syndrome, 102500 (3), Autosomal dominant; Alagille syndrome 2, 610205 (3), Autosomal dominant
<i>NOTCH3</i>	600276	?Myofibromatosis, infantile 2, 615293 (3), Autosomal dominant; Cerebral arteriopathy with subcortical infarcts and leukoencephalopathy 1, 125310 (3), Autosomal dominant; Lateral meningocele syndrome, 130720 (3), Autosomal dominant
<i>NOVA2</i>	601991	Neurodevelopmental disorder with or without autistic features and/or structural brain abnormalities, 618859 (3), Autosomal dominant
<i>NPC1</i>	607623	Niemann-Pick disease, type D, 257220 (3), Autosomal recessive; Niemann-Pick disease, type C1, 257220 (3), Autosomal recessive
<i>NPC1L1</i>	608010	[Low density lipoprotein cholesterol level QTL 7], 617966 (3); [Ezetimibe, nonresponse to], 617966 (3)
<i>NPC2</i>	601015	Niemann-pick disease, type C2, 607625 (3), Autosomal recessive

<i>NPHP1</i>	607100	Nephronophthisis 1, juvenile, 256100 (3), Autosomal recessive; Senior-Loken syndrome-1, 266900 (3), Autosomal recessive; Joubert syndrome 4, 609583 (3), Autosomal recessive
<i>NPHP3</i>	608002	Meckel syndrome 7, 267010 (3), Autosomal recessive; Renal- hepatic-pancreatic dysplasia 1, 208540 (3), Autosomal recessive; Nephronophthisis 3, 604387 (3), Autosomal recessive
<i>NPHP4</i>	607215	Nephronophthisis 4, 606966 (3), Autosomal recessive; Senior- Loken syndrome 4, 606996 (3), Autosomal recessive
<i>NPHS1</i>	602716	Nephrotic syndrome, type 1, 256300 (3), Autosomal recessive
<i>NPHS2</i>	604766	Nephrotic syndrome, type 2, 600995 (3), Autosomal recessive
<i>NPM1</i>	164040	Leukemia, acute myeloid, somatic, 601626 (3)
<i>NPPA</i>	108780	Atrial standstill 2, 615745 (3), Autosomal recessive; Atrial fibrillation, familial, 6, 612201 (3), Autosomal dominant
<i>NPR2</i>	108961	Short stature with nonspecific skeletal abnormalities, 616255 (3), Autosomal dominant; Epiphyseal chondrodysplasia, Miura type, 615923 (3), Autosomal dominant; Acromesomelic dysplasia, Maroteaux type, 602875 (3), Autosomal recessive
<i>NPR3</i>	108962	?Hypertension, salt-resistant (1)
<i>NPRL2</i>	607072	Epilepsy, familial focal, with variable foci 2, 617116 (3), Autosomal dominant
<i>NPRL3</i>	600928	Epilepsy, familial focal, with variable foci 3, 617118 (3), Autosomal dominant
<i>NPSR1</i>	608595	{Asthma, susceptibility to, 2}, 608584 (3)
<i>NQO1</i>	125860	{Breast cancer, poor survival after chemotherapy for} (3); {Leukemia, post-chemotherapy, susceptibility to} (3); {Benzene toxicity, susceptibility to} (3)
<i>NQO2</i>	160998	{?Breast cancer susceptibility}, 114480 (1), Autosomal dominant, Somatic mutation
<i>NR0B1</i>	300473	Adrenal hypoplasia, congenital, 300200 (3), X-linked recessive; 46XY sex reversal 2, dosage-sensitive, 300018 (3), X-linked
<i>NR0B2</i>	604630	Obesity, mild, early-onset, 601665 (3), Autosomal recessive, Autosomal dominant, Multifactorial
<i>NR1H4</i>	603826	Cholestasis, progressive familial intrahepatic, 5, 617049 (3), Autosomal recessive
<i>NR2E3</i>	604485	Enhanced S-cone syndrome, 268100 (3), Autosomal recessive; Retinitis pigmentosa 37, 611131 (3), Autosomal recessive, Autosomal dominant
<i>NR2F1</i>	132890	Bosch-Boonstra-Schaaf optic atrophy syndrome, 615722 (3), Autosomal dominant
<i>NR2F2</i>	107773	Congenital heart defects, multiple types, 4, 615779 (3), Autosomal dominant; 46,XX sex reversal 5, 618901 (3), Autosomal dominant
<i>NR3C1</i>	138040	Glucocorticoid resistance, 615962 (3), Autosomal dominant

<i>NR3C2</i>	600983	Pseudohypoaldosteronism type I, autosomal dominant, 177735 (3), Autosomal dominant; Hypertension, early-onset, autosomal dominant, with exacerbation in pregnancy, 605115 (3)
<i>NR4A3</i>	600542	Chondrosarcoma, extraskeletal myxoid, 612237 (3)
<i>NR5A1</i>	184757	Adrenocortical insufficiency, 612964 (3), Autosomal dominant; 46, XX sex reversal 4, 617480 (3), Autosomal dominant; Premature ovarian failure 7, 612964 (3), Autosomal dominant; Spermatogenic failure 8, 613957 (3), Autosomal dominant; 46XY sex reversal 3, 612965 (3), Autosomal dominant
<i>NRAS</i>	164790	Epidermal nevus, somatic, 162900 (3); Melanocytic nevus syndrome, congenital, somatic, 137550 (3); Schimmelpenning-Feuerstein-Mims syndrome, somatic mosaic, 163200 (3); Colorectal cancer, somatic, 114500 (3); ?RAS-associated autoimmune lymphoproliferative syndrome type IV, somatic, 614470 (3); Thyroid carcinoma, follicular, somatic, 188470 (3); Neurocutaneous melanosis, somatic, 249400 (3); Noonan syndrome 6, 613224 (3), Autosomal dominant
<i>NRG1</i>	142445	{?Schizophrenia, susceptibility to}, 603013 (1)
<i>NRIP1</i>	602490	?Congenital anomalies of kidney and urinary tract 3, 618270 (3), Autosomal dominant
<i>NRL</i>	162080	Retinitis pigmentosa 27, 613750 (3), Autosomal dominant; Retinal degeneration, autosomal recessive, clumped pigment type (3)
<i>NRROS</i>	615322	Seizures, early-onset, with neurodegeneration and brain calcification, 618875 (3), Autosomal recessive
<i>NRXN1</i>	600565	Pitt-Hopkins-like syndrome 2, 614325 (3), Autosomal recessive; {Schizophrenia, susceptibility to, 17}, 614332 (3)
<i>NSD1</i>	606681	Sotos syndrome 1, 117550 (3), Autosomal dominant
<i>NSDHL</i>	300275	CHILD syndrome, 308050 (3), X-linked dominant; CK syndrome, 300831 (3), X-linked recessive
<i>NSMCE2</i>	617246	Seckel syndrome 10, 617253 (3), Autosomal recessive
<i>NSMCE3</i>	608243	Lung disease, immunodeficiency, and chromosome breakage syndrome, 617241 (3), Autosomal recessive
<i>NSMF</i>	608137	Hypogonadotropic hypogonadism 9 with or without anosmia, 614838 (3), Autosomal dominant
<i>NSUN2</i>	610916	Mental retardation, autosomal recessive 5, 611091 (3), Autosomal recessive
<i>NT5C2</i>	600417	Spastic paraplegia 45, autosomal recessive, 613162 (3), Autosomal recessive
<i>NT5C3A</i>	606224	Anemia, hemolytic, due to UMPH1 deficiency, 266120 (3), Autosomal recessive
<i>NT5E</i>	129190	Calcification of joints and arteries, 211800 (3), Autosomal recessive
<i>NTF4</i>	162662	Glaucoma 1, open angle, 10, 613100 (3)
<i>NTHL1</i>	602656	Familial adenomatous polyposis 3, 616415 (3), Autosomal recessive

<i>NTN1</i>	601614	Mirror movements 4, 618264 (3), Autosomal dominant
<i>NTNG2</i>	618689	Neurodevelopmental disorder with behavioral abnormalities, absent speech, and hypotonia, 618718 (3), Autosomal recessive
<i>NTRK1</i>	191315	Insensitivity to pain, congenital, with anhidrosis, 256800 (3), Autosomal recessive
<i>NTRK2</i>	600456	Obesity, hyperphagia, and developmental delay, 613886 (3), Autosomal dominant; Epileptic encephalopathy, early infantile, 58, 617830 (3), Autosomal dominant
<i>NUBPL</i>	613621	Mitochondrial complex I deficiency, nuclear type 21, 618242 (3), Autosomal recessive
<i>NUDT15</i>	615792	{Thiopurines, poor metabolism of, 2}, 616903 (3), Autosomal dominant
<i>NUMA1</i>	164009	Leukemia, acute promyelocytic, somatic, 612376 (3)
<i>NUP107</i>	607617	Galloway-Mowat syndrome 7, 618348 (3), Autosomal recessive; ?Ovarian dysgenesis 6, 618078 (3), Autosomal recessive; Nephrotic syndrome, type 11, 616730 (3), Autosomal recessive
<i>NUP133</i>	607613	Nephrotic syndrome, type 18, 618177 (3), Autosomal recessive; ?Galloway-Mowat syndrome 8, 618349 (3), Autosomal recessive
<i>NUP155</i>	606694	?Atrial fibrillation 15, 615770 (3), Autosomal recessive
<i>NUP160</i>	607614	?Nephrotic syndrome, type 19, 618178 (3), Autosomal recessive
<i>NUP188</i>	615587	Sandestig-Stefanova syndrome, 618804 (3), Autosomal recessive
<i>NUP205</i>	614352	?Nephrotic syndrome, type 13, 616893 (3)
<i>NUP214</i>	114350	Leukemia, acute myeloid, somatic, 601626 (3); {Encephalopathy, acute, infection-induced, susceptibility to, 9}, 618426 (3), Autosomal recessive; Leukemia, T-cell acute lymphoblastic, somatic, 613065 (3)
<i>NUP37</i>	609264	?Microcephaly 24, primary, autosomal recessive, 618179 (3), Autosomal recessive
<i>NUP62</i>	605815	Striatonigral degeneration, infantile, 271930 (3), Autosomal recessive
<i>NUP85</i>	170285	Nephrotic syndrome, type 17, 618176 (3), Autosomal recessive
<i>NUP88</i>	602552	Fetal akinesia deformation sequence 4, 618393 (3), Autosomal recessive
<i>NUP93</i>	614351	Nephrotic syndrome, type 12, 616892 (3), Autosomal recessive
<i>NUS1</i>	610463	Mental retardation, autosomal dominant 55, with seizures, 617831 (3), Autosomal dominant; ?Congenital disorder of glycosylation, type 1aa, 617082 (3), Autosomal recessive
<i>NXN</i>	612895	Robinow syndrome, autosomal recessive 2, 618529 (3), Autosomal recessive
<i>NYX</i>	300278	Night blindness, congenital stationary (complete), 1A, X-linked, 310500 (3), X-linked recessive
<i>OAT</i>	613349	Gyrate atrophy of choroid and retina with or without ornithinemia, 258870 (3), Autosomal recessive
<i>OBSL1</i>	610991	3-M syndrome 2, 612921 (3), Autosomal recessive

<i>OCA2</i>	611409	[Skin/hair/eye pigmentation 1, blue/nonblue eyes], 227220 (3), Autosomal recessive; [Skin/hair/eye pigmentation 1, blond/brown hair], 227220 (3), Autosomal recessive; Albinism, oculocutaneous, type II, 203200 (3), Autosomal recessive; Albinism, brown oculocutaneous, 203200 (3), Autosomal recessive
<i>OCLN</i>	602876	Pseudo-TORCH syndrome 1, 251290 (3), Autosomal recessive
<i>OCRL</i>	300535	Lowe syndrome, 309000 (3), X-linked recessive; Dent disease 2, 300555 (3), X-linked recessive
<i>ODAPH</i>	614829	Amelogenesis imperfecta, type IIA4, 614832 (3), Autosomal recessive
<i>ODC1</i>	165640	{Colonic adenoma recurrence, reduced risk of}, 114500 (3), Autosomal dominant, Somatic mutation
<i>OFD1</i>	300170	Orofaciodigital syndrome I, 311200 (3), X-linked dominant; ?Retinitis pigmentosa 23, 300424 (3), X-linked recessive; Joubert syndrome 10, 300804 (3), X-linked recessive; Simpson-Golabi-Behmel syndrome, type 2, 300209 (3), X-linked recessive
<i>OGDH</i>	613022	Alpha-ketoglutarate dehydrogenase deficiency, 203740 (1), Autosomal recessive
<i>OGG1</i>	601982	Renal cell carcinoma, clear cell, somatic, 144700 (3)
<i>OGT</i>	300255	Mental retardation, X-linked 106, 300997 (3), X-linked recessive
<i>OLR1</i>	602601	{Myocardial infarction, susceptibility to}, 608446 (3)
<i>OPA1</i>	605290	{Glaucoma, normal tension, susceptibility to}, 606657 (3); Behr syndrome, 210000 (3), Autosomal recessive; Optic atrophy 1, 165500 (3), Autosomal dominant; Optic atrophy plus syndrome, 125250 (3), Autosomal dominant; ?Mitochondrial DNA depletion syndrome 14 (encephalocardiomyopathic type), 616896 (3), Autosomal recessive
<i>OPA3</i>	606580	3-methylglutaconic aciduria, type III, 258501 (3), Autosomal recessive; Optic atrophy 3 with cataract, 165300 (3), Autosomal dominant
<i>OPCML</i>	600632	Ovarian cancer, somatic, 167000 (3)
<i>OPHN1</i>	300127	Mental retardation, X-linked, with cerebellar hypoplasia and distinctive facial appearance, 300486 (3), X-linked recessive
<i>OPLAH</i>	614243	5-oxoprolinase deficiency, 260005 (3), Autosomal recessive, Autosomal dominant
<i>OPN1LW</i>	300822	Blue cone monochromacy, 303700 (3), X-linked recessive; Colorblindness, protan, 303900 (3), X-linked
<i>OPN1MW</i>	300821	Colorblindness, deutan, 303800 (3), X-linked; Blue cone monochromacy, 303700 (3), X-linked recessive
<i>OPN1SW</i>	613522	Colorblindness, tritan, 190900 (3), Autosomal dominant
<i>OPTN</i>	602432	{Glaucoma, normal tension, susceptibility to}, 606657 (3); Glaucoma 1, open angle, E, 137760 (3), Autosomal dominant; Amyotrophic lateral sclerosis 12, 613435 (3)
<i>OR2J3</i>	615016	[C3HEX, ability to smell], 615082 (3), Autosomal dominant

<i>ORAI1</i>	610277	Myopathy, tubular aggregate, 2, 615883 (3), Autosomal dominant; Immunodeficiency 9, 612782 (3), Autosomal recessive
<i>ORC1</i>	601902	Meier-Gorlin syndrome 1, 224690 (3), Autosomal recessive
<i>ORC4</i>	603056	Meier-Gorlin syndrome 2, 613800 (3), Autosomal recessive
<i>ORC6</i>	607213	Meier-Gorlin syndrome 3, 613803 (3), Autosomal recessive
<i>OSBPL2</i>	606731	Deafness, autosomal dominant 67, 616340 (3), Autosomal dominant
<i>OSGEP</i>	610107	Galloway-Mowat syndrome 3, 617729 (3), Autosomal recessive
<i>OSMR</i>	601743	Amyloidosis, primary localized cutaneous, 1, 105250 (3), Autosomal dominant
<i>OSTM1</i>	607649	Osteopetrosis, autosomal recessive 5, 259720 (3), Autosomal recessive
<i>OTC</i>	300461	Ornithine transcarbamylase deficiency, 311250 (3), X-linked recessive
<i>OTOA</i>	607038	Deafness, autosomal recessive 22, 607039 (3), Autosomal recessive
<i>OTOF</i>	603681	Auditory neuropathy, autosomal recessive, 1, 601071 (3), Autosomal recessive; Deafness, autosomal recessive 9, 601071 (3), Autosomal recessive
<i>OTOG</i>	604487	Deafness, autosomal recessive 18B, 614945 (3), Autosomal recessive
<i>OTOGL</i>	614925	Deafness, autosomal recessive 84B, 614944 (3), Autosomal recessive
<i>OTUD6B</i>	612021	Intellectual developmental disorder with dysmorphic facies, seizures, and distal limb anomalies, 617452 (3), Autosomal recessive
<i>OTULIN</i>	615712	Autoinflammation, panniculitis, and dermatosis syndrome, 617099 (3), Autosomal recessive
<i>OTX2</i>	600037	Pituitary hormone deficiency, combined, 6, 613986 (3), Autosomal dominant; Retinal dystrophy, early-onset, with or without pituitary dysfunction, 610125 (3), Autosomal dominant; Microphthalmia, syndromic 5, 610125 (3), Autosomal dominant
<i>OVOL2</i>	616441	Corneal dystrophy, posterior polymorphous, 1, 122000 (3), Autosomal dominant
<i>OXCT1</i>	601424	Succinyl CoA:3-oxoacid CoA transferase deficiency, 245050 (3), Autosomal recessive
<i>OXR1</i>	605609	Cerebellar hypoplasia/atrophy, epilepsy, and global developmental delay, 213000 (3), Autosomal recessive
<i>P2RX2</i>	600844	Deafness, autosomal dominant 41, 608224 (3), Autosomal dominant
<i>P2RY12</i>	600515	Bleeding disorder, platelet-type, 8, 609821 (3), Autosomal recessive
<i>P3H1</i>	610339	Osteogenesis imperfecta, type VIII, 610915 (3), Autosomal recessive
<i>P3H2</i>	610341	Myopia, high, with cataract and vitreoretinal degeneration, 614292 (3), Autosomal recessive

<i>P4HA2</i>	600608	Myopia 25, autosomal dominant, 617238 (3), Autosomal dominant
<i>P4HB</i>	176790	Cole-Carpenter syndrome 1, 112240 (3), Autosomal dominant
<i>P4HTM</i>	614584	Hypotonia, hypoventilation, impaired intellectual development, dysautonomia, epilepsy, and eye abnormalities, 618493 (3), Autosomal recessive
<i>PABPN1</i>	602279	Oculopharyngeal muscular dystrophy, 164300 (3), Autosomal dominant
<i>PACS1</i>	607492	Schuurs-Hoeijmakers syndrome, 615009 (3), Autosomal dominant
<i>PACS2</i>	610423	Epileptic encephalopathy, early infantile, 66, 618067 (3), Autosomal dominant
<i>PADI3</i>	606755	Uncombable hair syndrome, 191480 (3), Autosomal recessive
<i>PADI4</i>	605347	{Rheumatoid arthritis, susceptibility to}, 180300 (3)
<i>PADI6</i>	610363	Preimplantation embryonic lethality 2, 617234 (3), Autosomal recessive
<i>PAFAH1B1</i>	601545	Subcortical laminar heterotopia, 607432 (3), Autosomal dominant; Lissencephaly 1, 607432 (3), Autosomal dominant
<i>PAH</i>	612349	[Hyperphenylalaninemia, non-PKU mild], 261600 (3), Autosomal recessive; Phenylketonuria, 261600 (3), Autosomal recessive
<i>PAK1</i>	602590	Intellectual developmental disorder with macrocephaly, seizures, and speech delay, 618158 (3), Autosomal dominant
<i>PAK3</i>	300142	Mental retardation, X-linked 30/47, 300558 (3), X-linked recessive
<i>PALB2</i>	610355	{Pancreatic cancer, susceptibility to, 3}, 613348 (3); Fanconi anemia, complementation group N, 610832 (3); {Breast cancer, susceptibility to}, 114480 (3), Autosomal dominant, Somatic mutation
<i>PALLD</i>	608092	{Pancreatic cancer, susceptibility to, 1}, 606856 (3), Autosomal dominant
<i>PAM16</i>	614336	Spondylometaphyseal dysplasia, Megarbane-Dagher-Melike type, 613320 (3), Autosomal recessive
<i>PANK2</i>	606157	HARP syndrome, 607236 (3), Autosomal recessive; Neurodegeneration with brain iron accumulation 1, 234200 (3), Autosomal recessive
<i>PANX1</i>	608420	Oocyte maturation defect 7, 618550 (3), Autosomal dominant
<i>PAPSS2</i>	603005	Brachyolmia 4 with mild epiphyseal and metaphyseal changes, 612847 (3), Autosomal recessive
<i>PARK7</i>	602533	Parkinson disease 7, autosomal recessive early-onset, 606324 (3), Autosomal recessive
<i>PARN</i>	604212	Pulmonary fibrosis and/or bone marrow failure, telomere-related, 4, 616371 (3), Autosomal dominant; Dyskeratosis congenita, autosomal recessive 6, 616353 (3), Autosomal recessive
<i>PARS2</i>	612036	Epileptic encephalopathy, early infantile, 75, 618437 (3), Autosomal recessive

<i>PATL2</i>	614661	Oocyte maturation defect 4, 617743 (3), Autosomal recessive
<i>PAX1</i>	167411	Otofaciocervical syndrome 2, 615560 (3), Autosomal recessive
<i>PAX2</i>	167409	Glomerulosclerosis, focal segmental, 7, 616002 (3), Autosomal dominant; Papillorenal syndrome, 120330 (3), Autosomal dominant
<i>PAX3</i>	606597	Craniofacial-deafness-hand syndrome, 122880 (3), Autosomal dominant; Rhabdomyosarcoma 2, alveolar, 268220 (3), Somatic mutation; Waardenburg syndrome, type 1, 193500 (3), Autosomal dominant; Waardenburg syndrome, type 3, 148820 (3), Autosomal recessive, Autosomal dominant
<i>PAX4</i>	167413	Maturity-onset diabetes of the young, type IX, 612225 (3); Diabetes mellitus, type 2, 125853 (3), Autosomal dominant; {Diabetes mellitus, ketosis-prone, susceptibility to}, 612227 (3), Autosomal recessive, Autosomal dominant
<i>PAX5</i>	167414	{Leukemia, acute lymphoblastic, susceptibility to, 3}, 615545 (3) Optic nerve hypoplasia, 165550 (3), Autosomal dominant; ?Coloboma, ocular, 120200 (3), Autosomal dominant; Foveal hypoplasia 1, 136520 (3), Autosomal dominant; Aniridia, 106210 (3), Autosomal dominant; Keratitis, 148190 (3), Autosomal dominant
<i>PAX6</i>	607108	Anterior segment dysgenesis 5, multiple subtypes, 604229 (3), Autosomal dominant; Cataract with late-onset corneal dystrophy, 106210 (3), Autosomal dominant; ?Coloboma of optic nerve, 120430 (3), Autosomal dominant; ?Morning glory disc anomaly, 120430 (3), Autosomal dominant
<i>PAX7</i>	167410	Myopathy, congenital, progressive, with scoliosis, 618578 (3), Autosomal recessive; Rhabdomyosarcoma 2, alveolar, 268220 (3), Somatic mutation
<i>PAX8</i>	167415	Hypothyroidism, congenital, due to thyroid dysgenesis or hypoplasia, 218700 (3), Autosomal dominant
<i>PAX9</i>	167416	Tooth agenesis, selective, 3, 604625 (3), Autosomal dominant
<i>PBX1</i>	176310	Congenital anomalies of kidney and urinary tract syndrome with or without hearing loss, abnormal ears, or developmental delay, 617641 (3), Autosomal dominant
<i>PC</i>	608786	Pyruvate carboxylase deficiency, 266150 (3), Autosomal recessive
<i>PCARE</i>	613425	Retinitis pigmentosa 54, 613428 (3)
<i>PCBD1</i>	126090	Hyperphenylalaninemia, BH4-deficient, D, 264070 (3), Autosomal recessive
<i>PCCA</i>	232000	Propionicacidemia, 606054 (3), Autosomal recessive
<i>PCCB</i>	232050	Propionicacidemia, 606054 (3), Autosomal recessive
<i>PCDH12</i>	605622	Diencephalic-mesencephalic junction dysplasia syndrome 1, 251280 (3), Autosomal recessive
<i>PCDH15</i>	605514	Usher syndrome, type 1D/F digenic, 601067 (3), Autosomal recessive, Digenic recessive; Usher syndrome, type 1F, 602083 (3), Autosomal recessive; Deafness, autosomal recessive 23, 609533 (3), Autosomal recessive

<i>PCDH19</i>	300460	Epileptic encephalopathy, early infantile, 9, 300088 (3), X-linked
<i>PCGF2</i>	600346	Turnpenny-Fry syndrome, 618371 (3), Autosomal dominant
<i>PCK1</i>	614168	?Phosphoenolpyruvate carboxykinase deficiency, cytosolic, 261680 (3), Autosomal recessive
<i>PCK2</i>	614095	PEPCK deficiency, mitochondrial, 261650 (1), Autosomal recessive
<i>PCLO</i>	604918	?Pontocerebellar hypoplasia, type 3, 608027 (3), Autosomal recessive
<i>PCNA</i>	176740	?Ataxia-telangiectasia-like disorder 2, 615919 (3), Autosomal recessive
<i>PCNT</i>	605925	Microcephalic osteodysplastic primordial dwarfism, type II, 210720 (3), Autosomal recessive
<i>PCSK1</i>	162150	{Obesity, susceptibility to, BMIQ12}, 612362 (3); Obesity with impaired prohormone processing, 600955 (3), Autosomal recessive
<i>PCSK9</i>	607786	Hypercholesterolemia, familial, 3, 603776 (3), Autosomal dominant; {Low density lipoprotein cholesterol level QTL 1}, 603776 (3), Autosomal dominant
<i>PCYT1A</i>	123695	Spondylometaphyseal dysplasia with cone-rod dystrophy, 608940 (3), Autosomal recessive
<i>PCYT2</i>	602679	Spastic paraplegia 82, autosomal recessive, 618770 (3), Autosomal recessive
<i>PDCD1</i>	600244	{Systemic lupus erythematosus, susceptibility to, 2}, 605218 (3); {Multiple sclerosis, disease progression, modifier of}, 126200 (3), Multifactorial
<i>PDCD10</i>	609118	Cerebral cavernous malformations 3, 603285 (3)
<i>PDE10A</i>	610652	Dyskinesia, limb and orofacial, infantile-onset, 616921 (3), Autosomal recessive; Striatal degeneration, autosomal dominant, 616922 (3), Autosomal dominant
<i>PDE11A</i>	604961	Pigmented nodular adrenocortical disease, primary, 2, 610475 (3), Autosomal dominant
<i>PDE1C</i>	602987	?Deafness, autosomal dominant 74, 618140 (3), Autosomal dominant
<i>PDE3A</i>	123805	Hypertension and brachydactyly syndrome, 112410 (3), Autosomal dominant
<i>PDE4D</i>	600129	Acrodysostosis 2, with or without hormone resistance, 614613 (3), Autosomal dominant
<i>PDE6A</i>	180071	Retinitis pigmentosa 43, 613810 (3)
<i>PDE6B</i>	180072	Night blindness, congenital stationary, autosomal dominant 2, 163500 (3), Autosomal dominant; Retinitis pigmentosa-40, 613801 (3), Autosomal recessive
<i>PDE6C</i>	600827	Cone dystrophy 4, 613093 (3), Autosomal recessive
<i>PDE6D</i>	602676	?Joubert syndrome 22, 615665 (3), Autosomal recessive
<i>PDE6G</i>	180073	Retinitis pigmentosa 57, 613582 (3), Autosomal recessive

<i>PDE6H</i>	601190	Retinal cone dystrophy 3, 610024 (3), Autosomal recessive, Autosomal dominant; Achromatopsia 6, 610024 (3), Autosomal recessive, Autosomal dominant
<i>PDE8B</i>	603390	Striatal degeneration, autosomal dominant, 609161 (3), Autosomal dominant; Pigmented nodular adrenocortical disease, primary, 3, 614190 (3)
<i>PDGFB</i>	190040	Dermatofibrosarcoma protuberans, 607907 (3); Basal ganglia calcification, idiopathic, 5, 615483 (3), Autosomal dominant; Meningioma, SIS-related, 607174 (3), Autosomal dominant
<i>PDGFRA</i>	173490	Gastrointestinal stromal tumor/GIST-plus syndrome, somatic or familial, 175510 (3); Hypereosinophilic syndrome, idiopathic, resistant to imatinib, 607685 (3), Isolated cases, Somatic mutation
<i>PDGFRB</i>	173410	Myeloproliferative disorder with eosinophilia, 131440 (4), Autosomal dominant; Basal ganglia calcification, idiopathic, 4, 615007 (3), Autosomal dominant; Kosaki overgrowth syndrome, 616592 (3), Autosomal dominant; Premature aging syndrome, Penttinen type, 601812 (3), Autosomal dominant; Myofibromatosis, infantile, 1, 228550 (3), Autosomal dominant
<i>PDGFRL</i>	604584	Hepatocellular cancer, somatic, 114550 (3); Colorectal cancer, somatic, 114500 (3)
<i>PDHA1</i>	300502	Pyruvate dehydrogenase E1-alpha deficiency, 312170 (3), X-linked dominant
<i>PDHB</i>	179060	Pyruvate dehydrogenase E1-beta deficiency, 614111 (3)
<i>PDHX</i>	608769	Lacticacidemia due to PDX1 deficiency, 245349 (3), Autosomal recessive
<i>PDK3</i>	300906	?Charcot-Marie-Tooth disease, X-linked dominant, 6, 300905 (3), X-linked dominant
<i>PDLIM4</i>	603422	{Osteoporosis, susceptibility to}, 166710 (3), Autosomal dominant
<i>PDP1</i>	605993	Pyruvate dehydrogenase phosphatase deficiency, 608782 (3), Autosomal recessive
<i>PDSS1</i>	607429	Coenzyme Q10 deficiency, primary, 2, 614651 (3), Autosomal recessive
<i>PDSS2</i>	610564	Coenzyme Q10 deficiency, primary, 3, 614652 (3), Autosomal recessive
<i>PDX1</i>	600733	{Diabetes mellitus, type II, susceptibility to}, 125853 (3), Autosomal dominant; Pancreatic agenesis 1, 260370 (3), Autosomal recessive; MODY, type IV, 606392 (3)
<i>PDXK</i>	179020	Neuropathy, hereditary motor and sensory, type VIC, with optic atrophy, 618511 (3), Autosomal recessive
<i>PDYN</i>	131340	Spinocerebellar ataxia 23, 610245 (3), Autosomal dominant
<i>PDZD7</i>	612971	Usher syndrome, type IIC, GPR98/PDZD7 digenic, 605472 (3), Autosomal recessive, Digenic dominant; Deafness, autosomal recessive 57, 618003 (3), Autosomal recessive; {Retinal disease

in Usher syndrome type IIA, modifier of}, 276901 (3), Autosomal recessive

<i>PEPD</i>	613230	Prolidase deficiency, 170100 (3), Autosomal recessive
<i>PER2</i>	603426	?Advanced sleep phase syndrome, familial, 1, 604348 (3), Autosomal dominant
<i>PER3</i>	603427	?Advanced sleep phase syndrome, familial, 3, 616882 (3), Autosomal dominant
<i>PET100</i>	614770	Mitochondrial complex IV deficiency, 220110 (3), Autosomal recessive, Mitochondrial
<i>PEX1</i>	602136	Heimler syndrome 1, 234580 (3), Autosomal recessive; Peroxisome biogenesis disorder 1B (NALD/IRD), 601539 (3), Autosomal recessive; Peroxisome biogenesis disorder 1A (Zellweger), 214100 (3), Autosomal recessive
<i>PEX10</i>	602859	Peroxisome biogenesis disorder 6B, 614871 (3), Autosomal recessive; Peroxisome biogenesis disorder 6A (Zellweger), 614870 (3), Autosomal recessive
<i>PEX11B</i>	603867	?Peroxisome biogenesis disorder 14B, 614920 (3), Autosomal recessive
<i>PEX12</i>	601758	Peroxisome biogenesis disorder 3A (Zellweger), 614859 (3), Autosomal recessive; Peroxisome biogenesis disorder 3B, 266510 (3), Autosomal recessive
<i>PEX13</i>	601789	Peroxisome biogenesis disorder 11A (Zellweger), 614883 (3), Autosomal recessive; Peroxisome biogenesis disorder 11B, 614885 (3), Autosomal recessive
<i>PEX14</i>	601791	Peroxisome biogenesis disorder 13A (Zellweger), 614887 (3), Autosomal recessive
<i>PEX16</i>	603360	Peroxisome biogenesis disorder 8A (Zellweger), 614876 (3), Autosomal recessive; Peroxisome biogenesis disorder 8B, 614877 (3), Autosomal recessive
<i>PEX19</i>	600279	Peroxisome biogenesis disorder 12A (Zellweger), 614886 (3), Autosomal recessive
<i>PEX2</i>	170993	Peroxisome biogenesis disorder 5A (Zellweger), 614866 (3), Autosomal recessive; Peroxisome biogenesis disorder 5B, 614867 (3), Autosomal recessive
<i>PEX26</i>	608666	Peroxisome biogenesis disorder 7A (Zellweger), 614872 (3), Autosomal recessive; Peroxisome biogenesis disorder 7B, 614873 (3), Autosomal recessive
<i>PEX3</i>	603164	Peroxisome biogenesis disorder 10A (Zellweger), 614882 (3), Autosomal recessive; ?Peroxisome biogenesis disorder 10B, 617370 (3), Autosomal recessive

<i>PEX5</i>	600414	Peroxisome biogenesis disorder 2B, 202370 (3), Autosomal recessive; Rhizomelic chondrodysplasia punctata, type 5, 616716 (3), Autosomal recessive; Peroxisome biogenesis disorder 2A (Zellweger), 214110 (3), Autosomal recessive
<i>PEX6</i>	601498	Peroxisome biogenesis disorder 4B, 614863 (3), Autosomal recessive, Autosomal dominant; Heimler syndrome 2, 616617 (3), Autosomal recessive; Peroxisome biogenesis disorder 4A (Zellweger), 614862 (3), Autosomal recessive
<i>PEX7</i>	601757	Peroxisome biogenesis disorder 9B, 614879 (3), Autosomal recessive; Rhizomelic chondrodysplasia punctata, type 1, 215100 (3), Autosomal recessive
<i>PFKL</i>	171860	Hemolytic anemia due to phosphofructokinase deficiency (1)
<i>PFKM</i>	610681	Glycogen storage disease VII, 232800 (3), Autosomal recessive
<i>PFN1</i>	176610	Amyotrophic lateral sclerosis 18, 614808 (3)
<i>PGAM2</i>	612931	Glycogen storage disease X, 261670 (3), Autosomal recessive
<i>PGAP1</i>	611655	Mental retardation, autosomal recessive 42, 615802 (3), Autosomal recessive
<i>PGAP2</i>	615187	Hyperphosphatasia with mental retardation syndrome 3, 614207 (3), Autosomal recessive
<i>PGAP3</i>	611801	Hyperphosphatasia with mental retardation syndrome 4, 615716 (3), Autosomal recessive
<i>PGK1</i>	311800	Phosphoglycerate kinase 1 deficiency, 300653 (3), X-linked recessive
<i>PGM1</i>	171900	Congenital disorder of glycosylation, type It, 614921 (3), Autosomal recessive
<i>PGM3</i>	172100	Immunodeficiency 23, 615816 (3), Autosomal recessive
<i>PGR</i>	607311	?Progesterone resistance, 264080 (2), Autosomal recessive
<i>PHACTR1</i>	608723	Epileptic encephalopathy, early infantile, 70, 618298 (3), Autosomal dominant
<i>PHB</i>	176705	{Breast cancer, susceptibility to}, 114480 (3), Autosomal dominant, Somatic mutation
<i>PHC1</i>	602978	?Microcephaly 11, primary, autosomal recessive, 615414 (3), Autosomal recessive
<i>PHEX</i>	300550	Hypophosphatemic rickets, X-linked dominant, 307800 (3), X-linked dominant
<i>PHF21A</i>	608325	Intellectual developmental disorder with behavioral abnormalities and craniofacial dysmorphism with or without seizures, 618725 (3), Autosomal dominant
<i>PHF6</i>	300414	Borjeson-Forssman-Lehmann syndrome, 301900 (3), X-linked recessive
<i>PHF8</i>	300560	Mental retardation syndrome, X-linked, Siderius type, 300263 (3), X-linked recessive
<i>PHGDH</i>	606879	Neu-Laxova syndrome 1, 256520 (3), Autosomal recessive; Phosphoglycerate dehydrogenase deficiency, 601815 (3), Autosomal recessive

<i>PHIP</i>	612870	Chung-Jansen syndrome, 617991 (3), Autosomal dominant
<i>PHKA1</i>	311870	Muscle glycogenosis, 300559 (3), X-linked recessive
<i>PHKA2</i>	300798	Glycogen storage disease, type IXa2, 306000 (3), X-linked recessive; Glycogen storage disease, type IXa1, 306000 (3), X-linked recessive
<i>PHKB</i>	172490	Phosphorylase kinase deficiency of liver and muscle, autosomal recessive, 261750 (3), Autosomal recessive
<i>PHKG2</i>	172471	Glycogen storage disease IXc, 613027 (3), Autosomal recessive; Cirrhosis due to liver phosphorylase kinase deficiency (3)
<i>PHOX2A</i>	602753	Fibrosis of extraocular muscles, congenital, 2, 602078 (3), Autosomal recessive
<i>PHOX2B</i>	603851	Neuroblastoma with Hirschsprung disease, 613013 (3); Central hypoventilation syndrome, congenital, with or without Hirschsprung disease, 209880 (3), Autosomal dominant; {Neuroblastoma, susceptibility to, 2}, 613013 (3)
<i>PHYH</i>	602026	Refsum disease, 266500 (3), Autosomal recessive
<i>PHYKPL</i>	614683	[?Phosphohydroxylysineuria], 615011 (3)
<i>PI4KA</i>	600286	Polymicrogyria, perisylvian, with cerebellar hypoplasia and arthrogryposis, 616531 (3), Autosomal recessive
<i>PIBF1</i>	607532	Joubert syndrome 33, 617767 (3), Autosomal recessive
<i>PICALM</i>	603025	Leukemia, acute myeloid, somatic, 601626 (3)
<i>PIEZO1</i>	611184	Lymphatic malformation 6, 616843 (3), Autosomal recessive; Dehydrated hereditary stomatocytosis with or without pseudohyperkalemia and/or perinatal edema, 194380 (3), Autosomal dominant
<i>PIEZO2</i>	613629	Arthrogryposis, distal, with impaired proprioception and touch, 617146 (3), Autosomal recessive; Arthrogryposis, distal, type 5, 108145 (3), Autosomal dominant; ?Marden-Walker syndrome, 248700 (3), Autosomal dominant; Arthrogryposis, distal, type 3, 114300 (3), Autosomal dominant
<i>PIGA</i>	311770	Multiple congenital anomalies-hypotonia-seizures syndrome 2, 300868 (3), X-linked recessive; Paroxysmal nocturnal hemoglobinuria, somatic, 300818 (3)
<i>PIGB</i>	604122	Epileptic encephalopathy, early infantile, 80, 618580 (3), Autosomal recessive
<i>PIGC</i>	601730	Glycosylphosphatidylinositol biosynthesis defect 16, 617816 (3), Autosomal recessive
<i>PIGG</i>	616918	Mental retardation, autosomal recessive 53, 616917 (3), Autosomal recessive
<i>PIGH</i>	600154	Glycosylphosphatidylinositol biosynthesis defect 17, 618010 (3), Autosomal recessive
<i>PIGK</i>	605087	Neurodevelopmental disorder with hypotonia and cerebellar atrophy, with or without seizures, 618879 (3), Autosomal recessive
<i>PIGL</i>	605947	CHIME syndrome, 280000 (3), Autosomal recessive

<i>PIGM</i>	610273	Glycosylphosphatidylinositol deficiency, 610293 (3), Autosomal recessive
<i>PIGN</i>	606097	Multiple congenital anomalies-hypotonia-seizures syndrome 1, 614080 (3), Autosomal recessive
<i>PIGO</i>	614730	Hyperphosphatasia with mental retardation syndrome 2, 614749 (3), Autosomal recessive
<i>PIGP</i>	605938	Epileptic encephalopathy, early infantile, 55, 617599 (3), Autosomal recessive
<i>PIGQ</i>	605754	Epileptic encephalopathy, early infantile, 77, 618548 (3), Autosomal recessive
<i>PIGS</i>	610271	Glycosylphosphatidylinositol biosynthesis defect 18, 618143 (3), Autosomal recessive
<i>PIGT</i>	610272	Multiple congenital anomalies-hypotonia-seizures syndrome 3, 615398 (3), Autosomal recessive; ?Paroxysmal nocturnal hemoglobinuria 2, 615399 (3), Autosomal dominant, Somatic mutation
<i>PIGU</i>	608528	Neurodevelopmental disorder with brain anomalies, seizures, and scoliosis, 618590 (3), Autosomal recessive
<i>PIGV</i>	610274	Hyperphosphatasia with mental retardation syndrome 1, 239300 (3), Autosomal recessive
<i>PIGW</i>	610275	Glycosylphosphatidylinositol biosynthesis defect 11, 616025 (3), Autosomal recessive
<i>PIGY</i>	610662	Hyperphosphatasia with mental retardation syndrome 6, 616809 (3), Autosomal recessive
<i>PIK3C2A</i>	603601	Oculoskeletodental syndrome, 618440 (3), Autosomal recessive
<i>PIK3CA</i>	171834	Ovarian cancer, somatic, 167000 (3); Colorectal cancer, somatic, 114500 (3); CLAPO syndrome, somatic, 613089 (3); Cowden syndrome 5, 615108 (3); Hepatocellular carcinoma, somatic, 114550 (3); Breast cancer, somatic, 114480 (3); Macrodactyly, somatic, 155500 (3); Keratosis, seborrheic, somatic, 182000 (3); Gastric cancer, somatic, 613659 (3); Megalencephaly-capillary malformation-polymicrogyria syndrome, somatic, 602501 (3); Nevus, epidermal, somatic, 162900 (3); CLOVE syndrome, somatic, 612918 (3); Non-small cell lung cancer, somatic, 211980 (3)
<i>PIK3CD</i>	602839	Immunodeficiency 14, 615513 (3), Autosomal dominant
<i>PIK3R1</i>	171833	?Agammaglobulinemia 7, autosomal recessive, 615214 (3), Autosomal recessive; SHORT syndrome, 269880 (3), Autosomal dominant; Immunodeficiency 36, 616005 (3), Autosomal dominant
<i>PIK3R2</i>	603157	Megalencephaly-polymicrogyria-polydactyly-hydrocephalus syndrome 1, 603387 (3), Autosomal dominant
<i>PIK3R5</i>	611317	Ataxia-oculomotor apraxia 3, 615217 (3), Autosomal recessive
<i>PIKFYVE</i>	609414	Corneal fleck dystrophy, 121850 (3), Autosomal dominant
<i>PINK1</i>	608309	Parkinson disease 6, early onset, 605909 (3), Autosomal recessive

<i>PIP5K1C</i>	606102	Lethal congenital contractural syndrome 3, 611369 (3), Autosomal recessive
<i>PISD</i>	612770	Liberfarb syndrome, 618889 (3), Autosomal recessive
<i>PITPNM3</i>	608921	Cone-rod dystrophy 5, 600977 (3), Autosomal dominant
<i>PITX1</i>	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
<i>PITX2</i>	601542	Axenfeld-Rieger syndrome, type 1, 180500 (3), Autosomal dominant; Ring dermoid of cornea, 180550 (3), Autosomal dominant; Anterior segment dysgenesis 4, 137600 (3), Autosomal dominant
<i>PITX3</i>	602669	Anterior segment dysgenesis 1, multiple subtypes, 107250 (3), Autosomal dominant; Cataract 11, syndromic, autosomal recessive, 610623 (3), Autosomal recessive, Autosomal dominant; Cataract 11, multiple types, 610623 (3), Autosomal recessive, Autosomal dominant
<i>PJVK</i>	610219	Deafness, autosomal recessive 59, 610220 (3), Autosomal recessive
<i>PKD1</i>	601313	Polycystic kidney disease 1, 173900 (3), Autosomal dominant
<i>PKD1L1</i>	609721	Heterotaxy, visceral, 8, autosomal, 617205 (3), Autosomal recessive
<i>PKD2</i>	173910	Polycystic kidney disease 2, 613095 (3), Autosomal dominant
<i>PKDCC</i>	614150	Rhizomelic limb shortening with dysmorphic features, 618821 (3), Autosomal recessive
<i>PKHD1</i>	606702	Polycystic kidney disease 4, with or without hepatic disease, 263200 (3), Autosomal recessive
<i>PKLR</i>	609712	Pyruvate kinase deficiency, 266200 (3), Autosomal recessive; Adenosine triphosphate, elevated, of erythrocytes, 102900 (3), Autosomal dominant
<i>PKP1</i>	601975	Ectodermal dysplasia/skin fragility syndrome, 604536 (3), Autosomal recessive
<i>PKP2</i>	602861	Arrhythmogenic right ventricular dysplasia 9, 609040 (3), Autosomal dominant
<i>PLA2G2A</i>	172411	{?Colorectal cancer, susceptibility to}, 114500 (3), Autosomal dominant, Somatic mutation
<i>PLA2G4A</i>	600522	Gastrointestinal ulceration, recurrent, with dysfunctional platelets, 618372 (3), Autosomal recessive
<i>PLA2G5</i>	601192	[Fleck retina, familial benign], 228980 (3), Autosomal recessive
<i>PLA2G6</i>	603604	Infantile neuroaxonal dystrophy 1, 256600 (3), Autosomal recessive; Parkinson disease 14, autosomal recessive, 612953 (3), Autosomal recessive; Neurodegeneration with brain iron accumulation 2B, 610217 (3), Autosomal recessive
<i>PLA2G7</i>	601690	{Asthma, susceptibility to}, 600807 (3), Autosomal dominant; Platelet-activating factor acetylhydrolase deficiency, 614278 (3),

		Autosomal recessive; {Atopy, susceptibility to}, 147050 (3), Autosomal dominant
<i>PLAA</i>	603873	Neurodevelopmental disorder with progressive microcephaly, spasticity, and brain anomalies, 617527 (3), Autosomal recessive
<i>PLAG1</i>	603026	Silver-Russell syndrome 4, 618907 (3), Autosomal dominant; Adenomas, salivary gland pleomorphic, somatic, 181030 (3)
<i>PLAT</i>	173370	Hyperfibrinolysis, familial, due to increased release of PLAT, 612348 (1); Thrombophilia, familial, due to decreased release of PLAT, 612348 (1)
<i>PLAU</i>	191840	Quebec platelet disorder, 601709 (3), Autosomal dominant; {Alzheimer disease, late-onset, susceptibility to}, 104300 (3), Autosomal dominant
<i>PLCB1</i>	607120	Epileptic encephalopathy, early infantile, 12, 613722 (3), Autosomal recessive
<i>PLCB2</i>	604114	Platelet PLC beta-2 deficiency (1)
<i>PLCB3</i>	600230	Spondylometaphyseal dysplasia with corneal dystrophy, 618961 (3), Autosomal recessive
<i>PLCB4</i>	600810	Auriculocondylar syndrome 2, 614669 (3), Autosomal recessive, Autosomal dominant
<i>PLCD1</i>	602142	Nail disorder, nonsyndromic congenital, 3, (leukonychia), 151600 (3), Autosomal recessive, Autosomal dominant
<i>PLCE1</i>	608414	Nephrotic syndrome, type 3, 610725 (3), Autosomal recessive
<i>PLCG2</i>	600220	Familial cold autoinflammatory syndrome 3, 614468 (3), Autosomal dominant; Autoinflammation, antibody deficiency, and immune dysregulation syndrome, 614878 (3), Autosomal dominant
<i>PLCZ1</i>	608075	?Spermatogenic failure 17, 617214 (3), Autosomal recessive
<i>PLD1</i>	602382	Cardiac valvular defect, developmental, 212093 (3), Autosomal recessive
<i>PLD3</i>	615698	?Spinocerebellar ataxia 46, 617770 (3), Autosomal dominant
<i>PLEC</i>	601282	Muscular dystrophy, limb-girdle, autosomal recessive 17, 613723 (3), Autosomal recessive; Epidermolysis bullosa simplex with pyloric atresia, 612138 (3), Autosomal recessive; Epidermolysis bullosa simplex with muscular dystrophy, 226670 (3), Autosomal recessive; ?Epidermolysis bullosa simplex with nail dystrophy, 616487 (3), Autosomal recessive; Epidermolysis bullosa simplex, Ogna type, 131950 (3), Autosomal dominant
<i>PLEKHG2</i>	611893	Leukodystrophy and acquired microcephaly with or without dystonia, 616763 (3), Autosomal recessive
<i>PLEKHG4</i>	609526/600223	-/Spinocerebellar ataxia 4, 600223 (2), Autosomal dominant
<i>PLEKHG5</i>	611101	Spinal muscular atrophy, distal, autosomal recessive, 4, 611067 (3), Autosomal recessive; Charcot-Marie-Tooth disease, recessive intermediate C, 615376 (3), Autosomal recessive

<i>PLEKHM1</i>	611466	Osteopetrosis, autosomal dominant 3, 618107 (3), Autosomal dominant; ?Osteopetrosis, autosomal recessive 6, 611497 (3), Autosomal recessive
<i>PLG</i>	173350	Dysplasminogenemia, 217090 (3), Autosomal recessive; Plasminogen deficiency, type I, 217090 (3), Autosomal recessive
<i>PLIN1</i>	170290	Lipodystrophy, familial partial, type 4, 613877 (3), Autosomal dominant
<i>PLK4</i>	605031	Microcephaly and chorioretinopathy, autosomal recessive, 2, 616171 (3), Autosomal recessive
<i>PLN</i>	172405	Cardiomyopathy, hypertrophic, 18, 613874 (3), Autosomal dominant; Cardiomyopathy, dilated, 1P, 609909 (3)
<i>PLOD1</i>	153454	Ehlers-Danlos syndrome, kyphoscoliotic type, 1, 225400 (3), Autosomal recessive
<i>PLOD2</i>	601865	Bruck syndrome 2, 609220 (3), Autosomal recessive
<i>PLOD3</i>	603066	Lysyl hydroxylase 3 deficiency, 612394 (3), Autosomal recessive
<i>PLP1</i>	300401	Pelizaeus-Merzbacher disease, 312080 (3), X-linked recessive; Spastic paraplegia 2, X-linked, 312920 (3), X-linked recessive
<i>PLPBP</i>	604436	Epilepsy, early-onset, vitamin B6-dependent, 617290 (3), Autosomal recessive
<i>PLS1</i>	602734	Deafness, autosomal dominant 76, 618787 (3), Autosomal dominant
<i>PLS3</i>	300131	Bone mineral density QTL18, osteoporosis, 300910 (3), X-linked dominant
<i>PLTP</i>	172425	[High density lipoprotein cholesterol level QTL 9] (3)
<i>PLVAP</i>	607647	Diarrhea 10, protein-losing enteropathy type, 618183 (3), Autosomal recessive
<i>PMFBP1</i>	618085	Spermatogenic failure 31, 618112 (3), Autosomal recessive
<i>PML</i>	102578	Leukemia, acute promyelocytic, PML/RARA type (3)
<i>PMM2</i>	601785	Congenital disorder of glycosylation, type Ia, 212065 (3), Autosomal recessive
<i>PMP2</i>	170715	Charcot-Marie-Tooth disease, demyelinating, type 1G, 618279 (3), Autosomal dominant
<i>PMP22</i>	601097	Dejerine-Sottas disease, 145900 (3), Autosomal recessive, Autosomal dominant; ?Neuropathy, inflammatory demyelinating, 139393 (3), ?Autosomal dominant; Charcot-Marie-Tooth disease, type 1E, 118300 (3), Autosomal dominant; Roussy-Levy syndrome, 180800 (3), Autosomal dominant; Neuropathy, recurrent, with pressure palsies, 162500 (3), Autosomal dominant; Charcot-Marie-Tooth disease, type 1A, 118220 (3), Autosomal dominant
<i>PMPCA</i>	613036	Spinocerebellar ataxia, autosomal recessive 2, 213200 (3), Autosomal recessive
<i>PMPCB</i>	603131	Multiple mitochondrial dysfunctions syndrome 6, 617954 (3), Autosomal recessive

<i>PMS2</i>	600259	Mismatch repair cancer syndrome, 276300 (3), Autosomal recessive; Colorectal cancer, hereditary nonpolyposis, type 4, 614337 (3)
<i>PMVK</i>	607622	Porokeratosis 1, multiple types, 175800 (3), Autosomal dominant
<i>PNKD</i>	609023	Paroxysmal nonkinesigenic dyskinesia 1, 118800 (3), Autosomal dominant
<i>PNKP</i>	605610	Microcephaly, seizures, and developmental delay, 613402 (3), Autosomal recessive; Ataxia-oculomotor apraxia 4, 616267 (3), Autosomal recessive; ?Charcot-Marie-Tooth disease, type 2B2, 605589 (3), Autosomal recessive
<i>PNLIP</i>	246600	?Pancreatic lipase deficiency, 614338 (3), Autosomal recessive
<i>PNP</i>	164050	Immunodeficiency due to purine nucleoside phosphorylase deficiency, 613179 (3), Autosomal recessive
<i>PNPLA1</i>	612121	Ichthyosis, congenital, autosomal recessive 10, 615024 (3), Autosomal recessive
<i>PNPLA2</i>	609059	Neutral lipid storage disease with myopathy, 610717 (3), Autosomal recessive
<i>PNPLA6</i>	603197	Spastic paraplegia 39, autosomal recessive, 612020 (3), Autosomal recessive; Boucher-Neuhauser syndrome, 215470 (3), Autosomal recessive; Oliver-McFarlane syndrome, 275400 (3), Autosomal recessive; ?Laurence-Moon syndrome, 245800 (3), Autosomal recessive
<i>PNPLA8</i>	612123	?Mitochondrial myopathy with lactic acidosis, 251950 (3), Autosomal recessive
<i>PNPO</i>	603287	Pyridoxamine 5'-phosphate oxidase deficiency, 610090 (3), Autosomal recessive
<i>PNPT1</i>	610316	Deafness, autosomal recessive 70, 614934 (3), Autosomal recessive; Combined oxidative phosphorylation deficiency 13, 614932 (3), Autosomal recessive
<i>POC1A</i>	614783	Short stature, onychodysplasia, facial dysmorphism, and hypotrichosis, 614813 (3), Autosomal recessive
<i>POC1B</i>	614784	Cone-rod dystrophy 20, 615973 (3), Autosomal recessive
<i>POF1B</i>	300603	?Premature ovarian failure 2B, 300604 (3), X-linked recessive
<i>POFUT1</i>	607491	Dowling-Degos disease 2, 615327 (3), Autosomal dominant
<i>POGLUT1</i>	615618	?Muscular dystrophy, limb-girdle, autosomal recessive 21, 617232 (3), Autosomal recessive; Dowling-Degos disease 4, 615696 (3), Autosomal dominant
<i>POGZ</i>	614787	White-Sutton syndrome, 616364 (3), Autosomal dominant
<i>POLA1</i>	312040	Pigmentary disorder, reticulate, with systemic manifestations, X-linked, 301220 (3), X-linked recessive; Van Esch-O'Driscoll syndrome, 301030 (3), X-linked recessive
<i>POLD1</i>	174761	Mandibular hypoplasia, deafness, progeroid features, and lipodystrophy syndrome, 615381 (3), Autosomal dominant; {Colorectal cancer, susceptibility to, 10}, 612591 (3), Autosomal dominant

<i>POLE</i>	174762	FILS syndrome, 615139 (3), Autosomal recessive; IMAGE-I syndrome, 618336 (3), Autosomal recessive; {Colorectal cancer, susceptibility to, 12}, 615083 (3), Autosomal dominant
<i>POLG</i>	174763	Progressive external ophthalmoplegia, autosomal dominant 1, 157640 (3), Autosomal dominant; Mitochondrial DNA depletion syndrome 4B (MNGIE type), 613662 (3), Autosomal recessive; Mitochondrial recessive ataxia syndrome (includes SANDO and SCAE), 607459 (3), Autosomal recessive; Mitochondrial DNA depletion syndrome 4A (Alpers type), 203700 (3), Autosomal recessive; Progressive external ophthalmoplegia, autosomal recessive 1, 258450 (3), Autosomal recessive
<i>POLG2</i>	604983	Progressive external ophthalmoplegia with mitochondrial DNA deletions, autosomal dominant 4, 610131 (3), Autosomal dominant; Mitochondrial DNA depletion syndrome 16 (hepatic type), 618528 (3), Autosomal recessive
<i>POLH</i>	603968	Xeroderma pigmentosum, variant type, 278750 (3), Autosomal recessive
<i>POLR1A</i>	616404	Acrofacial dysostosis, Cincinnati type, 616462 (3), Autosomal dominant
<i>POLR1B</i>	602000	Treacher-Collins syndrome 4, 618939 (3), Autosomal dominant
<i>POLR1C</i>	610060	Treacher Collins syndrome 3, 248390 (3), Autosomal recessive; Leukodystrophy, hypomyelinating, 11, 616494 (3), Autosomal recessive
<i>POLR1D</i>	613715	Treacher Collins syndrome 2, 613717 (3), Autosomal recessive, Autosomal dominant
<i>POLR2A</i>	180660	Neurodevelopmental disorder with hypotonia and variable intellectual and behavioral abnormalities, 618603 (3), Autosomal dominant
<i>POLR3A</i>	614258	Leukodystrophy, hypomyelinating, 7, with or without oligodontia and/or hypogonadotropic hypogonadism, 607694 (3), Autosomal recessive; Wiedemann-Rautenstrauch syndrome, 264090 (3), Autosomal recessive
<i>POLR3B</i>	614366	Leukodystrophy, hypomyelinating, 8, with or without oligodontia and/or hypogonadotropic hypogonadism, 614381 (3), Autosomal recessive
<i>POMC</i>	176830	{Obesity, early-onset, susceptibility to}, 601665 (3), Autosomal recessive, Autosomal dominant, Multifactorial; Obesity, adrenal insufficiency, and red hair due to POMC deficiency, 609734 (3), Autosomal recessive
<i>POMGNT1</i>	606822	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; Muscular dystrophy-dystroglycanopathy (congenital with brain and eye anomalies), type A, 3, 253280 (3), Autosomal recessive

<i>POMGNT2</i>	614828	Muscular dystrophy-dystroglycanopathy (congenital with brain and eye anomalies, type A, 8, 614830 (3), Autosomal recessive; Muscular dystrophy-dystroglycanopathy (limb-girdle) type C, 8, 618135 (3), Autosomal recessive
<i>POMK</i>	615247	?Muscular dystrophy-dystroglycanopathy (limb-girdle), type C, 12, 616094 (3), Autosomal recessive; Muscular dystrophy-dystroglycanopathy (congenital with brain and eye anomalies), type A, 12, 615249 (3), Autosomal recessive
<i>POMP</i>	613386	Proteasome-associated autoinflammatory syndrome 2, 618048 (3), Autosomal dominant; Keratosis linearis with ichthyosis congenita and sclerosing keratoderma, 601952 (3), Autosomal recessive
<i>POMT1</i>	607423	Muscular dystrophy-dystroglycanopathy (limb-girdle), type C, 1, 609308 (3), Autosomal recessive; 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
<i>POMT2</i>	607439	Muscular dystrophy-dystroglycanopathy (limb-girdle), type C, 2, 613158 (3), Autosomal recessive; 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
<i>PON1</i>	168820	{Microvascular complications of diabetes 5}, 612633 (3); {Organophosphate poisoning, sensitivity to} (3); {Coronary artery disease, susceptibility to} (3); {Coronary artery spasm 2, susceptibility to} (3)
<i>PON2</i>	602447	{Coronary artery disease, susceptibility to} (3)
<i>POP1</i>	602486	Anauxetic dysplasia 2, 617396 (3), Autosomal recessive
<i>POPDC3</i>	605824	Muscular dystrophy, limb-girdle, autosomal recessive 26, 618848 (3), Autosomal recessive
<i>POR</i>	124015	Disordered steroidogenesis due to cytochrome P450 oxidoreductase, 613571 (3); Antley-Bixler syndrome with genital anomalies and disordered steroidogenesis, 201750 (3), Autosomal recessive
<i>PORCN</i>	300651	Focal dermal hypoplasia, 305600 (3), X-linked dominant
<i>POT1</i>	606478	{Glioma susceptibility 9}, 616568 (3), Autosomal dominant; {Melanoma, cutaneous malignant, susceptibility to, 10}, 615848 (3), Autosomal dominant
<i>POU1F1</i>	173110	Pituitary hormone deficiency, combined, 1, 613038 (3), Autosomal recessive, Autosomal dominant
<i>POU3F3</i>	602480	Snijders Blok-Fisher syndrome, 618604 (3), Autosomal dominant
<i>POU3F4</i>	300039	Deafness, X-linked 2, 304400 (3), X-linked recessive
<i>POU4F3</i>	602460	Deafness, autosomal dominant 15, 602459 (3), Autosomal dominant

<i>POU6F2</i>	609062	{Wilms tumor susceptibility-5}, 601583 (3), Autosomal dominant, Somatic mutation
<i>PPA2</i>	609988	?Sudden cardiac failure, alcohol-induced, 617223 (3), Autosomal recessive; Sudden cardiac failure, infantile, 617222 (3), Autosomal recessive
<i>PPARA</i>	170998	{Hyperapobetalipoproteinemia, susceptibility to} (3)
<i>PPARG</i>	601487	[Obesity, resistance to] (3); Carotid intimal medial thickness 1, 609338 (3); {Diabetes, type 2}, 125853 (3), Autosomal dominant; Insulin resistance, severe, digenic, 604367 (3), Autosomal dominant; Obesity, severe, 601665 (3), Autosomal recessive, Autosomal dominant, Multifactorial; Lipodystrophy, familial partial, type 3, 604367 (3), Autosomal dominant
<i>PPCS</i>	609853	Cardiomyopathy, dilated, 2C, 618189 (3), Autosomal recessive
<i>PPIB</i>	123841	Osteogenesis imperfecta, type IX, 259440 (3), Autosomal recessive
<i>PPIP5K2</i>	611648	Deafness, autosomal recessive 100, 618422 (3), Autosomal recessive
<i>PPM1D</i>	605100	Breast cancer, somatic, 114480 (3); Jansen de Vries syndrome, 617450 (3), Autosomal dominant
<i>PPM1K</i>	611065	?Maple syrup urine disease, mild variant, 615135 (3)
<i>PPOX</i>	600923	Porphyria variegata, 176200 (3), Autosomal dominant
<i>PPP1CB</i>	600590	Noonan syndrome-like disorder with loose anagen hair 2, 617506 (3), Autosomal dominant
<i>PPP1R12A</i>	602021	Genitourinary and/or/brain malformation syndrome, 618820 (3), Autosomal dominant
<i>PPP1R15B</i>	613257	Microcephaly, short stature, and impaired glucose metabolism 2, 616817 (3), Autosomal recessive
<i>PPP1R17</i>	604088	{Hypercholesterolemia, susceptibility to}, 143890 (3), Autosomal dominant
<i>PPP1R3A</i>	600917	Insulin resistance, severe, digenic, 125853 (3), Autosomal dominant
<i>PPP2CA</i>	176915	Neurodevelopmental disorder and language delay with or without structural brain abnormalities, 618354 (3), Autosomal dominant
<i>PPP2R1A</i>	605983	Mental retardation, autosomal dominant 36, 616362 (3), Autosomal dominant
<i>PPP2R1B</i>	603113	Lung cancer, somatic, 211980 (3)
<i>PPP2R2B</i>	604325	Spinocerebellar ataxia 12, 604326 (3), Autosomal dominant
<i>PPP2R3C</i>	615902	Gonadal dysgenesis, dysmorphic facies, retinal dystrophy, and myopathy, 618419 (3), Autosomal recessive; Spermatogenic failure 36, 618420 (3), Autosomal dominant
<i>PPP2R5D</i>	601646	Mental retardation, autosomal dominant 35, 616355 (3), Autosomal dominant
<i>PPP3CA</i>	114105	Epileptic encephalopathy, infantile or early childhood, 1, 617711 (3), Autosomal dominant; Arthrogryposis, cleft palate,

		craniosynostosis, and impaired intellectual development, 618265 (3), Autosomal dominant
<i>PPT1</i>	600722	Ceroid lipofuscinosis, neuronal, 1, 256730 (3), Autosomal recessive
<i>PQBP1</i>	300463	Renpenning syndrome, 309500 (3), X-linked recessive
<i>PRCC</i>	179755	Renal cell carcinoma, papillary, 605074 (3)
<i>PRCD</i>	610598	Retinitis pigmentosa 36, 610599 (3)
<i>PRDM12</i>	616458	Neuropathy, hereditary sensory and autonomic, type VIII, 616488 (3), Autosomal recessive
<i>PRDM16</i>	605557	Left ventricular noncompaction 8, 615373 (3), Autosomal dominant; Cardiomyopathy, dilated, 1LL, 615373 (3), Autosomal dominant
<i>PRDM5</i>	614161	Brittle cornea syndrome 2, 614170 (3), Autosomal recessive
<i>PRDM6</i>	616982	Patent ductus arteriosus 3, 617039 (3), Autosomal dominant
<i>PRDM8</i>	616639	?Epilepsy, progressive myoclonic, 10, 616640 (3), Autosomal recessive
<i>PRDX1</i>	176763	Methylmalonic aciduria and homocystinuria, cblC type, digenic, 277400 (3), Autosomal recessive
<i>PREPL</i>	609557	Myasthenic syndrome, congenital, 22, 616224 (3), Autosomal recessive
<i>PRF1</i>	170280	Hemophagocytic lymphohistiocytosis, familial, 2, 603553 (3), Autosomal recessive; Aplastic anemia, 609135 (3); Lymphoma, non-Hodgkin, 605027 (3)
<i>PRG4</i>	604283	Camptodactyly-arthropathy-coxa vara-pericarditis syndrome, 208250 (3), Autosomal recessive
<i>PRICKLE1</i>	608500	Epilepsy, progressive myoclonic 1B, 612437 (3), Autosomal recessive
<i>PRIMPOL</i>	615421	Myopia 22, autosomal dominant, 615420 (3), Autosomal dominant
<i>PRKACA</i>	601639	Cushing syndrome, ACTH-independent adrenal, somatic, 615830 (3)
<i>PRKACG</i>	176893	?Bleeding disorder, platelet-type, 19, 616176 (3), Autosomal recessive
<i>PRKAG2</i>	602743	Glycogen storage disease of heart, lethal congenital, 261740 (3), Autosomal dominant; Wolff-Parkinson-White syndrome, 194200 (3), Autosomal dominant; Cardiomyopathy, hypertrophic 6, 600858 (3), Autosomal dominant
<i>PRKAR1A</i>	188830	Adrenocortical tumor, somatic (3); Myxoma, intracardiac, 255960 (3), Autosomal dominant; Carney complex, type 1, 160980 (3), Autosomal dominant; Pigmented nodular adrenocortical disease, primary, 1, 610489 (3), Autosomal dominant; Acrodysostosis 1, with or without hormone resistance, 101800 (3), Autosomal dominant
<i>PRKCA</i>	176960	Pituitary tumor, invasive (3)

<i>PRKCD</i>	176977	Autoimmune lymphoproliferative syndrome, type III, 615559 (3), Autosomal recessive
<i>PRKCG</i>	176980	Spinocerebellar ataxia 14, 605361 (3), Autosomal dominant
<i>PRKCH</i>	605437	{Cerebral infarction, susceptibility to}, 601367 (3), Multifactorial
<i>PRKCSH</i>	177060	Polycystic liver disease 1, 174050 (3), Autosomal dominant
<i>PRKD1</i>	605435	Congenital heart defects and ectodermal dysplasia, 617364 (3), Autosomal dominant
<i>PRKDC</i>	600899	Immunodeficiency 26, with or without neurologic abnormalities, 615966 (3), Autosomal recessive
<i>PRKG1</i>	176894	Aortic aneurysm, familial thoracic 8, 615436 (3), Autosomal dominant
<i>PRKN</i>	602544	Parkinson disease, juvenile, type 2, 600116 (3), Autosomal recessive; Ovarian cancer, somatic, 167000 (3); Adenocarcinoma of lung, somatic, 211980 (3)
<i>PRKRA</i>	603424	Dystonia 16, 612067 (3), Autosomal recessive
<i>PRLR</i>	176761	Hyperprolactinemia, 615555 (3), Autosomal recessive, Autosomal dominant; Multiple fibroadenomas of the breast, 615554 (3), Autosomal dominant
<i>PRMT7</i>	610087	Short stature, brachydactyly, intellectual developmental disability, and seizures, 617157 (3), Autosomal recessive
<i>PRNP</i>	176640	Insomnia, fatal familial, 600072 (3), Autosomal dominant; {Kuru, susceptibility to}, 245300 (3); Huntington disease-like 1, 603218 (3), Autosomal dominant; Prion disease with protracted course, 606688 (3), Autosomal dominant; Cerebral amyloid angiopathy, PRNP-related, 137440 (3), Autosomal dominant; Creutzfeldt-Jakob disease, 123400 (3), Autosomal dominant; Gerstmann-Straussler disease, 137440 (3), Autosomal dominant
<i>PROC</i>	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
<i>PRODH</i>	606810	Hyperprolinemia, type I, 239500 (3), Autosomal recessive; {Schizophrenia, susceptibility to, 4}, 600850 (3), Autosomal dominant
<i>PROK2</i>	607002	Hypogonadotropic hypogonadism 4 with or without anosmia, 610628 (3), Autosomal dominant
<i>PROKR2</i>	607123	Hypogonadotropic hypogonadism 3 with or without anosmia, 244200 (3), Autosomal dominant
<i>PROM1</i>	604365	Retinitis pigmentosa 41, 612095 (3), Autosomal recessive; Stargardt disease 4, 603786 (3), Autosomal dominant; Cone-rod dystrophy 12, 612657 (3), Autosomal recessive, Autosomal dominant; Macular dystrophy, retinal, 2, 608051 (3), Autosomal dominant
<i>PROP1</i>	601538	Pituitary hormone deficiency, combined, 2, 262600 (3), Autosomal recessive

<i>PROS1</i>	176880	Thrombophilia due to protein S deficiency, autosomal recessive, 614514 (3), Autosomal recessive; Thrombophilia due to protein S deficiency, autosomal dominant, 612336 (3), Autosomal dominant
<i>PROZ</i>	176895	[Protein Z deficiency], 614024 (3)
<i>PRPF3</i>	607301	Retinitis pigmentosa 18, 601414 (3), Autosomal dominant
<i>PRPF31</i>	606419	Retinitis pigmentosa 11, 600138 (3), Autosomal dominant
<i>PRPF4</i>	607795	Retinitis pigmentosa 70, 615922 (3), Autosomal dominant
<i>PRPF6</i>	613979	Retinitis pigmentosa 60, 613983 (3), Autosomal dominant
<i>PRPF8</i>	607300	Retinitis pigmentosa 13, 600059 (3), Autosomal dominant
<i>PRPH</i>	170710	{Amyotrophic lateral sclerosis, susceptibility to}, 105400 (3), Autosomal recessive, Autosomal dominant
<i>PRPH2</i>	179605	Macular dystrophy, patterned, 1, 169150 (3), Autosomal dominant; Retinitis punctata albescens, 136880 (3), Autosomal recessive, Autosomal dominant; Choroidal dystrophy, central areolar 2, 613105 (3), Autosomal dominant; Retinitis pigmentosa 7 and digenic form, 608133 (3), Autosomal recessive, Autosomal dominant; Leber congenital amaurosis 18, 608133 (3), Autosomal recessive, Autosomal dominant; Macular dystrophy, vitelliform, 3, 608161 (3), Autosomal dominant
<i>PRPS1</i>	311850	Charcot-Marie-Tooth disease, X-linked recessive, 5, 311070 (3), X-linked recessive; Phosphoribosylpyrophosphate synthetase superactivity, 300661 (3), X-linked recessive; Deafness, X-linked 1, 304500 (3), X-linked; Arts syndrome, 301835 (3), X-linked recessive; Gout, PRPS-related, 300661 (3), X-linked recessive
<i>PRRT2</i>	614386	Episodic kinesigenic dyskinesia 1, 128200 (3), Autosomal dominant; Seizures, benign familial infantile, 2, 605751 (3), Autosomal dominant; Convulsions, familial infantile, with paroxysmal choreoathetosis, 602066 (3), Autosomal dominant
<i>PRRX1</i>	167420	Agnathia-otocephaly complex, 202650 (3), Autosomal recessive, Autosomal dominant
<i>PRSS1</i>	276000	Pancreatitis, hereditary, 167800 (3), Autosomal dominant
<i>PRSS12</i>	606709	Mental retardation, autosomal recessive 1, 249500 (3), Autosomal recessive
<i>PRSS2</i>	601564	{Pancreatitis, chronic, protection against}, 167800 (3), Autosomal dominant
<i>PRSS56</i>	613858	Microphthalmia, isolated 6, 613517 (3), Autosomal recessive
<i>PRUNE1</i>	617413	Neurodevelopmental disorder with microcephaly, hypotonia, and variable brain anomalies, 617481 (3), Autosomal recessive
<i>PRX</i>	605725	Charcot-Marie-Tooth disease, type 4F, 614895 (3), Autosomal recessive; Dejerine-Sottas disease, 145900 (3), Autosomal recessive, Autosomal dominant

<i>PSAP</i>	176801	Gaucher disease, atypical, 610539 (3); Krabbe disease, atypical, 611722 (3), Autosomal recessive; Combined SAP deficiency, 611721 (3), Autosomal recessive; Metachromatic leukodystrophy due to SAP-b deficiency, 249900 (3), Autosomal recessive
<i>PSAT1</i>	610936	Neu-Laxova syndrome 2, 616038 (3), Autosomal recessive; ?Phosphoserine aminotransferase deficiency, 610992 (3), Autosomal recessive
<i>PSEN1</i>	104311	Alzheimer disease, type 3, with spastic paraparesis and unusual plaques, 607822 (3), Autosomal dominant; Alzheimer disease, type 3, with spastic paraparesis and apraxia, 607822 (3), Autosomal dominant; ?Acne inversa, familial, 3, 613737 (3), Autosomal dominant; Alzheimer disease, type 3, 607822 (3), Autosomal dominant; Dementia, frontotemporal, 600274 (3), Autosomal dominant; Pick disease, 172700 (3), Autosomal dominant; Cardiomyopathy, dilated, 1U, 613694 (3), Autosomal dominant
<i>PSEN2</i>	600759	Alzheimer disease-4, 606889 (3), Autosomal dominant; Cardiomyopathy, dilated, 1V, 613697 (3), Autosomal dominant
<i>PSENEN</i>	607632	Acne inversa, familial, 2, with or without Dowling-Degos disease, 613736 (3), Autosomal dominant
<i>PSMA6</i>	602855	{Myocardial infarction, susceptibility to}, 608446 (3)
<i>PSMB4</i>	602177	?Proteasome-associated autoinflammatory syndrome 3 and digenic forms, 617591 (3), Autosomal recessive
<i>PSMB8</i>	177046	Proteasome-associated autoinflammatory syndrome 1 and digenic forms, 256040 (3), Autosomal recessive
<i>PSMB9</i>	177045	?Proteasome-associated autoinflammatory syndrome 3, digenic, 617591 (3), Autosomal recessive
<i>PSMC3IP</i>	608665	Ovarian dysgenesis 3, 614324 (3), Autosomal recessive
<i>PSMD12</i>	604450	Stankiewicz-Isidor syndrome, 617516 (3), Autosomal dominant
<i>PSPH</i>	172480	Phosphoserine phosphatase deficiency, 614023 (3), Autosomal recessive
<i>PSTPIP1</i>	606347	Pyogenic sterile arthritis, pyoderma gangrenosum, and acne, 604416 (3), Autosomal dominant
<i>PTCH1</i>	601309	Basal cell carcinoma, somatic, 605462 (3); Basal cell nevus syndrome, 109400 (3), Autosomal dominant; Holoprosencephaly 7, 610828 (3), Autosomal dominant
<i>PTCH2</i>	603673	Basal cell carcinoma, somatic, 605462 (3); Basal cell nevus syndrome, 109400 (3), Autosomal dominant; Medulloblastoma, somatic, 155255 (3)
<i>PTCHD1</i>	300828	{Autism, susceptibility to, X-linked 4}, 300830 (3), X-linked recessive
<i>PTDSS1</i>	612792	Lenz-Majewski hyperostotic dwarfism, 151050 (3), Autosomal dominant

<i>PTEN</i>	601728	Prostate cancer, somatic, 176807 (3); {Glioma susceptibility 2}, 613028 (3); Cowden syndrome 1, 158350 (3), Autosomal dominant; Lhermitte-Duclos syndrome, 158350 (3), Autosomal dominant; Macrocephaly/autism syndrome, 605309 (3), Autosomal dominant; {Meningioma}, 607174 (3), Autosomal dominant
<i>PTF1A</i>	607194	Pancreatic and cerebellar agenesis, 609069 (3), Autosomal recessive; Pancreatic agenesis 2, 615935 (3), Autosomal recessive
<i>PTGDR</i>	604687	{Asthma, susceptibility to, 1}, 607277 (3)
<i>PTGER2</i>	176804	{Asthma, aspirin-induced, susceptibility to}, 208550 (3), Autosomal recessive
<i>PTGIS</i>	601699	Hypertension, essential, 145500 (3), Multifactorial
<i>PTH</i>	168450	Hypoparathyroidism, familial isolated 1, 146200 (3), Autosomal recessive, Autosomal dominant
<i>PTH1R</i>	168468	Metaphyseal chondrodysplasia, Murk Jansen type, 156400 (3), Autosomal dominant; Failure of tooth eruption, primary, 125350 (3), Autosomal dominant; Eiken syndrome, 600002 (3), Autosomal recessive; Chondrodysplasia, Blomstrand type, 215045 (3), Autosomal recessive
<i>PTHLH</i>	168470	Brachydactyly, type E2, 613382 (3), Autosomal dominant
<i>PTPN1</i>	176885	{Insulin resistance, susceptibility to}, 125853 (3), Autosomal dominant
<i>PTPN11</i>	176876	LEOPARD syndrome 1, 151100 (3), Autosomal dominant; Metachondromatosis, 156250 (3), Autosomal dominant; Noonan syndrome 1, 163950 (3), Autosomal dominant; Leukemia, juvenile myelomonocytic, somatic, 607785 (3)
<i>PTPN12</i>	600079	Colon cancer, somatic, 114500 (3)
<i>PTPN14</i>	603155	Choanal atresia and lymphedema, 613611 (3), Autosomal recessive
<i>PTPN22</i>	600716	{Diabetes, type 1, susceptibility to}, 222100 (3), Autosomal recessive; {Systemic lupus erythematosus susceptibility to}, 152700 (3), Autosomal dominant; {Rheumatoid arthritis, susceptibility to}, 180300 (3)
<i>PTPN23</i>	606584	Neurodevelopmental disorder and structural brain anomalies with or without seizures and spasticity, 618890 (3), Autosomal recessive
<i>PTPRC</i>	151460	{Hepatitis C virus, susceptibility to}, 609532 (3); Severe combined immunodeficiency, T cell-negative, B-cell/natural killer-cell positive, 608971 (3), Autosomal recessive
<i>PTPRF</i>	179590	?Breasts and/or nipples, aplasia or hypoplasia of, 2, 616001 (3), Autosomal recessive
<i>PTPRJ</i>	600925	Colon cancer, somatic, 114500 (3)
<i>PTPRO</i>	600579	Nephrotic syndrome, type 6, 614196 (3), Autosomal recessive

<i>PTPRQ</i>	603317	Deafness, autosomal dominant 73, 617663 (3), Autosomal dominant; Deafness, autosomal recessive 84A, 613391 (3), Autosomal recessive
<i>PTRH2</i>	608625	Infantile-onset multisystem neurologic, endocrine, and pancreatic disease, 616263 (3), Autosomal recessive
<i>PTS</i>	612719	Hyperphenylalaninemia, BH4-deficient, A, 261640 (3), Autosomal recessive
<i>PUF60</i>	604819	Verheij syndrome, 615583 (3), Autosomal dominant
<i>PUM1</i>	607204	Spinocerebellar ataxia 47, 617931 (3), Autosomal dominant
<i>PURA</i>	600473	Mental retardation, autosomal dominant 31, 616158 (3), Autosomal dominant
<i>PUS1</i>	608109	Myopathy, lactic acidosis, and sideroblastic anemia 1, 600462 (3), Autosomal recessive
<i>PUS3</i>	616283	Mental retardation, autosomal recessive 55, 617051 (3), Autosomal recessive
<i>PUS7</i>	616261	Intellectual developmental disorder with abnormal behavior, microcephaly, and short stature, 618342 (3), Autosomal recessive
<i>PXDN</i>	605158	Anterior segment dysgenesis 7, with sclerocornea, 269400 (3), Autosomal recessive
<i>PYCR1</i>	179035	Cutis laxa, autosomal recessive, type IIIB, 614438 (3); Cutis laxa, autosomal recessive, type IIB, 612940 (3), Autosomal recessive
<i>PYCR2</i>	616406	Leukodystrophy, hypomyelinating, 10, 616420 (3), Autosomal recessive
<i>PYGL</i>	613741	Glycogen storage disease VI, 232700 (3), Autosomal recessive
<i>PYGM</i>	608455	McArdle disease, 232600 (3), Autosomal recessive
<i>PYROXD1</i>	617220	Myopathy, myofibrillar, 8, 617258 (3), Autosomal recessive
<i>QARS1 (QARS)</i>	603727	Microcephaly, progressive, seizures, and cerebral and cerebellar atrophy, 615760 (3), Autosomal recessive
<i>QDPR</i>	612676	Hyperphenylalaninemia, BH4-deficient, C, 261630 (3), Autosomal recessive
<i>QRICH1</i>	617387	Ververi-Brady syndrome, 617982 (3), Autosomal dominant
<i>QRICH2</i>	618304	Spermatogenic failure 35, 618341 (3), Autosomal recessive
<i>QRSL1</i>	617209	Combined oxidative phosphorylation deficiency 40, 618835 (3), Autosomal recessive
<i>RAB11B</i>	604198	Neurodevelopmental disorder with ataxic gait, absent speech, and decreased cortical white matter, 617807 (3), Autosomal dominant
<i>RAB18</i>	602207	Warburg micro syndrome 3, 614222 (3), Autosomal recessive
<i>RAB23</i>	606144	Carpenter syndrome, 201000 (3), Autosomal recessive
<i>RAB27A</i>	603868	GrisCELLI syndrome, type 2, 607624 (3), Autosomal recessive
<i>RAB28</i>	612994	Cone-rod dystrophy 18, 615374 (3), Autosomal recessive
<i>RAB33B</i>	605950	Smith-McCort dysplasia 2, 615222 (3), Autosomal recessive
<i>RAB39B</i>	300774	Waisman syndrome, 311510 (3), X-linked recessive; Mental retardation, X-linked 72, 300271 (3), X-linked recessive

<i>RAB3GAP1</i>	602536	Warburg micro syndrome 1, 600118 (3), Autosomal recessive
<i>RAB3GAP2</i>	609275	Warburg micro syndrome 2, 614225 (3), Autosomal recessive; Martsof syndrome, 212720 (3), Autosomal recessive
<i>RAB7A</i>	602298	Charcot-Marie-Tooth disease, type 2B, 600882 (3), Autosomal dominant
<i>RABL3</i>	618542	{?Pancreatic cancer, susceptibility to, 5}, 618680 (3), Autosomal dominant
<i>RAC1</i>	602048	Mental retardation, autosomal dominant 48, 617751 (3), Autosomal dominant
<i>RAC2</i>	602049	?Immunodeficiency 73C with defective neutrophil chemotaxis and hypogammaglobulinemia, 618987 (3); Immunodeficiency 73B with defective neutrophil chemotaxis and lymphopenia, 618986 (3); Immunodeficiency 73A with defective neutrophil chemotaxis and leukocytosis, 608203 (3)
<i>RAC3</i>	602050	Neurodevelopmental disorder with structural brain anomalies and dysmorphic facies, 618577 (3), Autosomal dominant
<i>RAD21</i>	606462	?Mungan syndrome, 611376 (3), Autosomal recessive; Cornelia de Lange syndrome 4, 614701 (3), Autosomal dominant
<i>RAD50</i>	604040	Nijmegen breakage syndrome-like disorder, 613078 (3)
<i>RAD51</i>	179617	{Breast cancer, susceptibility to}, 114480 (3), Autosomal dominant, Somatic mutation; Mirror movements 2, 614508 (3), Autosomal dominant; Fanconi anemia, complementation group R, 617244 (3), Autosomal dominant
<i>RAD51C</i>	602774	{Breast-ovarian cancer, familial, susceptibility to, 3}, 613399 (3); Fanconi anemia, complementation group O, 613390 (3), Autosomal recessive
<i>RAD51D</i>	602954	{Breast-ovarian cancer, familial, susceptibility to, 4}, 614291 (3)
<i>RAD54B</i>	604289	Colon cancer, somatic, 114500 (3); Lymphoma, non-Hodgkin, somatic, 605027 (3)
<i>RAD54L</i>	603615	Adenocarcinoma, colonic, somatic (3); {Breast cancer, invasive ductal}, 114480 (3), Autosomal dominant, Somatic mutation; Lymphoma, non-Hodgkin, somatic, 605027 (3)
<i>RAF1</i>	164760	LEOPARD syndrome 2, 611554 (3); Noonan syndrome 5, 611553 (3), Autosomal dominant; Cardiomyopathy, dilated, 1NN, 615916 (3), Autosomal dominant
<i>RAG1</i>	179615	Omenn syndrome, 603554 (3), Autosomal recessive; Severe combined immunodeficiency, B cell-negative, 601457 (3), Autosomal recessive; 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
<i>RAG2</i>	179616	Severe combined immunodeficiency, B cell-negative, 601457 (3), Autosomal recessive; Combined cellular and humoral immune defects with granulomas, 233650 (3), Autosomal recessive; Omenn syndrome, 603554 (3), Autosomal recessive

<i>RAI1</i>	607642	Smith-Magenis syndrome, 182290 (3), Autosomal dominant, Isolated cases
<i>RALGAP1</i>	608884	Neurodevelopmental disorder with hypotonia, neonatal respiratory insufficiency, and thermoregulation, 618797 (3), Autosomal recessive
<i>RANBP2</i>	601181	{Encephalopathy, acute, infection-induced, 3, susceptibility to}, 608033 (3), Autosomal dominant
<i>RAP1GDS1</i>	179502	Lymphocytic leukemia, acute T-cell (3)
<i>RAPGEF2</i>	609530	?Epilepsy, familial adult myoclonic, 7, 618075 (3), Autosomal dominant
<i>RAPSN</i>	601592	Fetal akinesia deformation sequence 2, 618388 (3), Autosomal recessive; Myasthenic syndrome, congenital, 11, associated with acetylcholine receptor deficiency, 616326 (3), Autosomal recessive
<i>RARA</i>	180240	Leukemia, acute promyelocytic, 612376 (1)
<i>RARB</i>	180220	Microphthalmia, syndromic 12, 615524 (3), Autosomal recessive, Autosomal dominant
<i>RARS1 (RARS)</i>	107820	Leukodystrophy, hypomyelinating, 9, 616140 (3), Autosomal recessive
<i>RARS2</i>	611524	Pontocerebellar hypoplasia, type 6, 611523 (3), Autosomal recessive
<i>RASA1</i>	139150	Capillary malformation-arteriovenous malformation 1, 608354 (3), Autosomal dominant; Basal cell carcinoma, somatic, 605462 (3)
<i>RASGRP1</i>	603962	Immunodeficiency 64, 618534 (3), Autosomal recessive
<i>RASGRP2</i>	605577	?Bleeding disorder, platelet-type, 18, 615888 (3), Autosomal recessive
<i>RAX</i>	601881	Microphthalmia, isolated 3, 611038 (3), Autosomal recessive
<i>RAX2</i>	610362	?Macular degeneration, age-related, 6, 613757 (3); Cone-rod dystrophy 11, 610381 (3), Autosomal dominant
<i>RB1</i>	614041	Small cell cancer of the lung, somatic, 182280 (3); Bladder cancer, somatic, 109800 (3); Retinoblastoma, trilateral, 180200 (3), Autosomal dominant, Somatic mutation; Osteosarcoma, somatic, 259500 (3); Retinoblastoma, 180200 (3), Autosomal dominant, Somatic mutation
<i>RB1CC1</i>	606837	Breast cancer, somatic, 114480 (3)
<i>RBBP8</i>	604124	Jawad syndrome, 251255 (3), Autosomal recessive; Pancreatic carcinoma, somatic (3); Seckel syndrome 2, 606744 (3), Autosomal recessive
<i>RBCK1</i>	610924	Polyglucosan body myopathy 1 with or without immunodeficiency, 615895 (3), Autosomal recessive
<i>RBM10</i>	300080	TARP syndrome, 311900 (3), X-linked recessive
<i>RBM12</i>	607179	{Schizophrenia 19, susceptibility to}, 617629 (3), Autosomal dominant
<i>RBM15</i>	606077	Megakaryoblastic leukemia, acute, 606077 (2)
<i>RBM20</i>	613171	Cardiomyopathy, dilated, 1DD, 613172 (3), Autosomal dominant

<i>RBM28</i>	612074	?Alopecia, neurologic defects, and endocrinopathy syndrome, 612079 (3), Autosomal recessive
<i>RBM8A</i>	605313	Thrombocytopenia-absent radius syndrome, 274000 (3), Autosomal recessive
<i>RBMX</i>	300199	?Mental retardation, X-linked, syndromic 11, Shashi type, 300238 (3), X-linked recessive
<i>RBP3</i>	180290	?Retinitis pigmentosa 66, 615233 (3), Autosomal recessive
<i>RBP4</i>	180250	Microphthalmia, isolated, with coloboma 10, 616428 (3), Autosomal dominant; Retinal dystrophy, iris coloboma, and comedogenic acne syndrome, 615147 (3), Autosomal recessive
<i>RBPJ</i>	147183	Adams-Oliver syndrome 3, 614814 (3), Autosomal dominant
<i>RCBTB1</i>	607867	Retinal dystrophy with or without extraocular anomalies, 617175 (3), Autosomal recessive
<i>RD3</i>	180040	Leber congenital amaurosis 12, 610612 (3), Autosomal recessive
<i>RDH11</i>	607849	?Retinal dystrophy, juvenile cataracts, and short stature syndrome, 616108 (3), Autosomal recessive
<i>RDH12</i>	608830	Leber congenital amaurosis 13, 612712 (3), Autosomal recessive, Autosomal dominant
<i>RDH5</i>	601617	Fundus albipunctatus, 136880 (3), Autosomal recessive, Autosomal dominant
<i>RDX</i>	179410	Deafness, autosomal recessive 24, 611022 (3), Autosomal recessive
<i>RECQL4</i>	603780	RAPADILINO syndrome, 266280 (3), Autosomal recessive; Baller-Gerold syndrome, 218600 (3), Autosomal recessive; Rothmund-Thomson syndrome, type 2,, 268400 (3), Autosomal recessive
<i>REEP1</i>	609139	Spastic paraplegia 31, autosomal dominant, 610250 (3), Autosomal dominant; ?Neuronopathy, distal hereditary motor, type VB, 614751 (3), Autosomal dominant
<i>REEP2</i>	609347	?Spastic paraplegia 72, autosomal dominant, 615625 (3), Autosomal recessive, Autosomal dominant; ?Spastic paraplegia 72, autosomal recessive, 615625 (3), Autosomal recessive, Autosomal dominant
<i>REEP6</i>	609346	Retinitis pigmentosa 77, 617304 (3), Autosomal recessive
<i>RELA</i>	164014	?Mucocutaneous ulceration, chronic, 618287 (3), Autosomal dominant
<i>RELB</i>	604758	?Immunodeficiency 53, 617585 (3), Autosomal recessive
<i>RELN</i>	600514	{Epilepsy, familial temporal lobe, 7}, 616436 (3), Autosomal dominant; Lissencephaly 2 (Norman-Roberts type), 257320 (3), Autosomal recessive
<i>RELT</i>	611211	Amelogenesis imperfecta, type IIIC, 618386 (3), Autosomal recessive
<i>REN</i>	179820	[Hyperproreninemia] (3); Renal tubular dysgenesis, 267430 (3), Autosomal recessive; Hyperuricemic nephropathy, familial juvenile 2, 613092 (3), Autosomal dominant

<i>REPS1</i>	614825	?Neurodegeneration with brain iron accumulation 7, 617916 (3), Autosomal recessive
<i>RERE</i>	605226	Neurodevelopmental disorder with or without anomalies of the brain, eye, or heart, 616975 (3), Autosomal dominant
<i>REST</i>	600571	{Wilms tumor 6, susceptibility to}, 616806 (3); Fibromatosis, gingival, 5, 617626 (3), Autosomal dominant; ?Deafness, autosomal dominant 27, 612431 (3), Autosomal dominant
<i>RET</i>	164761	Multiple endocrine neoplasia IIB, 162300 (3), Autosomal dominant; Pheochromocytoma, 171300 (3), Autosomal dominant; Multiple endocrine neoplasia IIA, 171400 (3), Autosomal dominant; Medullary thyroid carcinoma, 155240 (3), Autosomal dominant; {Hirschsprung disease, protection against}, 142623 (3), Autosomal dominant; Central hypoventilation syndrome, congenital, 209880 (3), Autosomal dominant; {Hirschsprung disease, susceptibility to, 1}, 142623 (3), Autosomal dominant
<i>RETN</i>	605565	{Diabetes mellitus, noninsulin-dependent, susceptibility to}, 125853 (3), Autosomal dominant; {Hypertension, insulin resistance-related, susceptibility to}, 125853 (3), Autosomal dominant
<i>RETREG1</i>	613114	Neuropathy, hereditary sensory and autonomic, type IIB, 613115 (3), Autosomal recessive
<i>RFC1</i>	102579	Cerebellar ataxia, neuropathy, and vestibular areflexia syndrome, 614575 (3), Autosomal recessive
<i>RFT1</i>	611908	Congenital disorder of glycosylation, type In, 612015 (3), Autosomal recessive
<i>RFWD3</i>	614151	?Fanconi anemia, complementation group W, 617784 (3), Autosomal recessive
<i>RFX5</i>	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
<i>RFX6</i>	612659	Mitchell-Riley syndrome, 615710 (3), Autosomal recessive
<i>RFXANK</i>	603200	MHC class II deficiency, complementation group B, 209920 (3), Autosomal recessive
<i>RFXAP</i>	601861	Bare lymphocyte syndrome, type II, complementation group D, 209920 (3), Autosomal recessive
<i>RGR</i>	600342	Retinitis pigmentosa 44, 613769 (3)
<i>RGS5</i>	603276	[Blood pressure regulation QTL], 145500 (2), Multifactorial
<i>RGS9</i>	604067	Bradyopsia, 608415 (3)
<i>RGS9BP</i>	607814	Bradyopsia, 608415 (3)
<i>RHAG</i>	180297	Overhydrated hereditary stomatocytosis, 185000 (3), Autosomal dominant; Anemia, hemolytic, Rh-null, regulator type, 268150 (3), Autosomal dominant
<i>RHBDF2</i>	614404	Tylosis with esophageal cancer, 148500 (3), Autosomal dominant

<i>RHCE</i>	111700	[Blood group, Rhesus] (3); Rh-null disease, amorph type, 617970 (3)
<i>RHD</i>	111680	[Rh-negative blood type] (3)
<i>RHO</i>	180380	Night blindness, congenital stationary, autosomal dominant 1, 610445 (3); Retinitis punctata albescens, 136880 (3), Autosomal recessive, Autosomal dominant; Retinitis pigmentosa 4, autosomal dominant or recessive, 613731 (3), Autosomal recessive, Autosomal dominant
<i>RHOA</i>	165390	Ectodermal dysplasia with facial dysmorphism and acral, ocular, and brain anomalies, somatic mosaic, 618727 (3)
<i>RHOBTB2</i>	607352	Epileptic encephalopathy, early infantile, 64, 618004 (3), Autosomal dominant
<i>RHOH</i>	602037	{?Epidermodysplasia verruciformis, susceptibility to, 4}, 618307 (3), Autosomal recessive
<i>RIC1</i>	610354	CATIFA syndrome, 618761 (3), Autosomal recessive
<i>RIMS1</i>	606629	Cone-rod dystrophy 7, 603649 (3)
<i>RIMS2</i>	606630	Cone-rod synaptic disorder syndrome, congenital nonprogressive, 618970 (3), Autosomal recessive
<i>RIN2</i>	610222	Macrocephaly, alopecia, cutis laxa, and scoliosis, 613075 (3), Autosomal recessive
<i>RINT1</i>	610089	Infantile liver failure syndrome 3, 618641 (3), Autosomal recessive
<i>RIPK1</i>	603453	Immunodeficiency 57 with autoinflammation, 618108 (3), Autosomal recessive; Autoinflammation with episodic fever and lymphadenopathy, 618852 (3), Autosomal dominant
<i>RIPK4</i>	605706	Popliteal pterygium syndrome, Bartsocas-Papas type, 263650 (3), Autosomal recessive; CHAND syndrome, 214350 (3), Autosomal recessive
<i>RIPOR2</i>	611410	?Deafness, autosomal recessive 104, 616515 (3), Autosomal recessive
<i>RIPPLY2</i>	609891	?Spondylocostal dysostosis 6, 616566 (3), Autosomal recessive
<i>RIT1</i>	609591	Noonan syndrome 8, 615355 (3), Autosomal dominant
<i>RLBP1</i>	180090	Retinitis punctata albescens, 136880 (3), Autosomal recessive, Autosomal dominant; Bothnia retinal dystrophy, 607475 (3), Autosomal recessive; Newfoundland rod-cone dystrophy, 607476 (3); Fundus albipunctatus, 136880 (3), Autosomal recessive, Autosomal dominant
<i>RLIM</i>	300379	Tonne-Kalscheuer syndrome, 300978 (3), X-linked
<i>RMND1</i>	614917	Combined oxidative phosphorylation deficiency 11, 614922 (3), Autosomal recessive
<i>RNASEH1</i>	604123	Progressive external ophthalmoplegia with mitochondrial DNA deletions, autosomal recessive 2, 616479 (3), Autosomal recessive
<i>RNASEH2A</i>	606034	Aicardi-Goutieres syndrome 4, 610333 (3), Autosomal recessive
<i>RNASEH2B</i>	610326	Aicardi-Goutieres syndrome 2, 610181 (3), Autosomal recessive
<i>RNASEH2C</i>	610330	Aicardi-Goutieres syndrome 3, 610329 (3), Autosomal recessive

<i>RNASEL</i>	180435	Prostate cancer 1, 601518 (3), Autosomal dominant
<i>RNASET2</i>	612944	Leukoencephalopathy, cystic, without megalencephaly, 612951 (3), Autosomal recessive
<i>RNF113A</i>	300951	Trichothiodystrophy 5, nonphotosensitive, 300953 (3), X-linked dominant
<i>RNF114</i>	612451/612950	-/{Psoriasis susceptibility 12}, 612950 (2)
<i>RNF125</i>	610432	Tenorio syndrome, 616260 (3), Autosomal dominant
<i>RNF13</i>	609247	Epileptic encephalopathy, early infantile, 73, 618379 (3), Autosomal dominant
<i>RNF139</i>	603046	Renal cell carcinoma, 144700 (3)
<i>RNF168</i>	612688	RIDDLE syndrome, 611943 (3), Autosomal recessive
<i>RNF170</i>	614649	Ataxia, sensory, 1, autosomal dominant, 608984 (3), Autosomal dominant
<i>RNF212</i>	612041	Recombination rate QTL 1, 612042 (3)
<i>RNF213</i>	613768	{Moyamoya disease 2, susceptibility to}, 607151 (3)
<i>RNF216</i>	609948	Cerebellar ataxia and hypogonadotropic hypogonadism, 212840 (3), Autosomal recessive
<i>RNF43</i>	612482	Sessile serrated polyposis cancer syndrome, 617108 (3), Autosomal dominant
<i>RNF6</i>	604242	Esophageal carcinoma, somatic, 133239 (3)
<i>RNPC3</i>	618016	?Growth hormone deficiency, isolated, type V, 618160 (3), Autosomal recessive
<i>ROBO2</i>	602431	Vesicoureteral reflux 2, 610878 (3), Autosomal dominant
<i>ROBO3</i>	608630	Gaze palsy, familial horizontal, with progressive scoliosis, 1, 607313 (3), Autosomal recessive
<i>ROBO4</i>	607528	Aortic valve disease 8, 618496 (3), Autosomal dominant
<i>ROGDI</i>	614574	Kohlschutter-Tonz syndrome, 226750 (3), Autosomal recessive
<i>ROM1</i>	180721	Retinitis pigmentosa 7, digenic form, 608133 (3), Autosomal recessive, Autosomal dominant
<i>ROR1</i>	602336	?Deafness, autosomal recessive 108, 617654 (3), Autosomal recessive
<i>ROR2</i>	602337	Brachydactyly, type B1, 113000 (3), Autosomal dominant; Robinow syndrome, autosomal recessive, 268310 (3), Autosomal recessive
<i>RORA</i>	600825	Intellectual developmental disorder with or without epilepsy or cerebellar ataxia, 618060 (3), Autosomal dominant
<i>RORB</i>	601972	{Epilepsy, idiopathic generalized, susceptibility to, 15}, 618357 (3), Autosomal dominant
<i>RORC</i>	602943	Immunodeficiency 42, 616622 (3), Autosomal recessive
<i>RP1</i>	603937	Retinitis pigmentosa 1, 180100 (3), Autosomal recessive, Autosomal dominant
<i>RP1L1</i>	608581	Retinitis pigmentosa 88, 618826 (3), Autosomal recessive; Occult macular dystrophy, 613587 (3), Autosomal dominant
<i>RP2</i>	300757	Retinitis pigmentosa 2, 312600 (3), X-linked
<i>RP9</i>	607331	?Retinitis pigmentosa 9, 180104 (3), Autosomal dominant

<i>RPE65</i>	180069	Leber congenital amaurosis 2, 204100 (3), Autosomal recessive; Retinitis pigmentosa 87 with choroidal involvement, 618697 (3), Autosomal dominant; Retinitis pigmentosa 20, 613794 (3), Autosomal recessive
<i>RPGR</i>	312610	Cone-rod dystrophy, X-linked, 1, 304020 (3), X-linked; Retinitis pigmentosa 3, 300029 (3); Retinitis pigmentosa, X-linked, and sinorespiratory infections, with or without deafness, 300455 (3); Macular degeneration, X-linked atrophic, 300834 (3), X-linked recessive
<i>RPGRIP1</i>	605446	Cone-rod dystrophy 13, 608194 (3); Leber congenital amaurosis 6, 613826 (3), Autosomal recessive
<i>RPGRIP1L</i>	610937	COACH syndrome, 216360 (3), Autosomal recessive; Meckel syndrome 5, 611561 (3), Autosomal recessive; Joubert syndrome 7, 611560 (3), Autosomal recessive
<i>RPIA</i>	180430	Ribose 5-phosphate isomerase deficiency, 608611 (3), Autosomal recessive
<i>RPL10</i>	312173	Mental retardation, X-linked, syndromic, 35, 300998 (3), X-linked recessive; {Autism, susceptibility to, X-linked 5}, 300847 (3)
<i>RPL11</i>	604175	Diamond-Blackfan anemia 7, 612562 (3), Autosomal dominant
<i>RPL13</i>	113703	Spondyloepimetaphyseal dysplasia, Isidor-Toutain type, 618728 (3), Autosomal dominant
<i>RPL15</i>	604174	?Diamond-Blackfan anemia 12, 615550 (3), Autosomal dominant
<i>RPL18</i>	604179	?Diamond-Blackfan anemia 18, 618310 (3), Autosomal dominant
<i>RPL21</i>	603636	Hypotrichosis 12, 615885 (3), Autosomal dominant
<i>RPL26</i>	603704	?Diamond-Blackfan anemia 11, 614900 (3), Autosomal dominant
<i>RPL27</i>	607526	?Diamond-Blackfan anemia 16, 617408 (3), Autosomal dominant
<i>RPL35</i>	618315	?Diamond-Blackfan anemia 19, 618312 (3), Autosomal dominant
<i>RPL35A</i>	180468	Diamond-Blackfan anemia 5, 612528 (3), Autosomal dominant
<i>RPL5</i>	603634	Diamond-Blackfan anemia 6, 612561 (3), Autosomal dominant
<i>RPS10</i>	603632	Diamond-Blackfan anemia 9, 613308 (3), Autosomal dominant
<i>RPS14</i>	130620	Macrocytic anemia, refractory, due to 5q deletion, somatic, 153550 (3)
<i>RPS15A</i>	603674	?Diamond-Blackfan anemia 20, 618313 (3), Autosomal dominant
<i>RPS17</i>	180472	Diamond-Blackfan anemia 4, 612527 (3), Autosomal dominant
<i>RPS19</i>	603474	Diamond-Blackfan anemia 1, 105650 (3), Autosomal dominant
<i>RPS23</i>	603683	Brachycephaly, trichomegaly, and developmental delay, 617412 (3), Autosomal dominant
<i>RPS24</i>	602412	Diamond-blackfan anemia 3, 610629 (3), Autosomal dominant
<i>RPS26</i>	603701	Diamond-Blackfan anemia 10, 613309 (3), Autosomal dominant
<i>RPS27</i>	603702	?Diamond-Blackfan anemia 17, 617409 (3), Autosomal dominant
<i>RPS28</i>	603685	Diamond Blackfan anemia 15 with mandibulofacial dysostosis, 606164 (3), Autosomal dominant
<i>RPS29</i>	603633	Diamond-Blackfan anemia 13, 615909 (3), Autosomal dominant
<i>RPS6KA3</i>	300075	Mental retardation, X-linked 19, 300844 (3), X-linked dominant; Coffin-Lowry syndrome, 303600 (3), X-linked dominant

<i>RPS7</i>	603658	Diamond-Blackfan anemia 8, 612563 (3), Autosomal dominant
<i>RPSA</i>	150370	Asplenia, isolated congenital, 271400 (3), Autosomal dominant
<i>RRAS2</i>	600098	Ovarian carcinoma (3); Noonan syndrome 12, 618624 (3), Autosomal dominant
<i>RRM2B</i>	604712	Progressive external ophthalmoplegia with mitochondrial DNA deletions, autosomal dominant 5, 613077 (3), Autosomal dominant; 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
<i>RS1</i>	300839	Retinoschisis, 312700 (3), X-linked recessive
<i>RSPH1</i>	609314	Ciliary dyskinesia, primary, 24, 615481 (3), Autosomal recessive
<i>RSPH3</i>	615876	Ciliary dyskinesia, primary, 32, 616481 (3), Autosomal recessive
<i>RSPH4A</i>	612647	Ciliary dyskinesia, primary, 11, 612649 (3)
<i>RSPH9</i>	612648	Ciliary dyskinesia, primary, 12, 612650 (3)
<i>RSPO1</i>	609595	Palmoplantar hyperkeratosis with squamous cell carcinoma of skin and sex reversal, 610644 (3), Autosomal recessive; Palmoplantar hyperkeratosis and true hermaphroditism, 610644 (3), Autosomal recessive
<i>RSPO2</i>	610575	Tetraamelia syndrome 2, 618021 (3), Autosomal recessive; ?Humero femoral hypoplasia with radiotibial ray deficiency, 618022 (3), Autosomal recessive
<i>RSPO4</i>	610573	Anonychia congenita, 206800 (3), Autosomal recessive
<i>RSPRY1</i>	616585	Spondyloepimetaphyseal dysplasia, Faden-Alkuraya type, 616723 (3), Autosomal recessive
<i>RSRC1</i>	613352	Intellectual developmental disorder, autosomal recessive 70, 618402 (3), Autosomal recessive
<i>RTKL1</i>	608833	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; Dyskeratosis congenita, autosomal dominant 4, 615190 (3), Autosomal recessive, Autosomal dominant
<i>RTN2</i>	603183	Spastic paraplegia 12, autosomal dominant, 604805 (3), Autosomal dominant
<i>RTN4IP1</i>	610502	Optic atrophy 10 with or without ataxia, mental retardation, and seizures, 616732 (3), Autosomal recessive
<i>RTN4R</i>	605566	{Schizophrenia, susceptibility to}, 181500 (3), Autosomal dominant
<i>RTTN</i>	610436	Microcephaly, short stature, and polymicrogyria with seizures, 614833 (3), Autosomal recessive
<i>RUBCN</i>	613516	?Spinocerebellar ataxia, autosomal recessive 15, 615705 (3), Autosomal recessive
<i>RUNX1</i>	151385	Platelet disorder, familial, with associated myeloid malignancy, 601399 (3), Autosomal dominant; Leukemia, acute myeloid, 601626 (3), Autosomal dominant, Somatic mutation

<i>RUNX2</i>	600211	Cleidocranial dysplasia, forme fruste, with brachydactyly, 119600 (3), Autosomal dominant; Metaphyseal dysplasia with maxillary hypoplasia with or without brachydactyly, 156510 (3), Autosomal dominant; Cleidocranial dysplasia, 119600 (3), Autosomal dominant; Cleidocranial dysplasia, forme fruste, dental anomalies only, 119600 (3), Autosomal dominant
<i>RUSC2</i>	611053	Mental retardation, autosomal recessive 61, 617773 (3), Autosomal recessive
<i>RXYLT1</i>	605862	Muscular dystrophy-dystroglycanopathy (congenital with brain and eye anomalies), type A, 10, 615041 (3), Autosomal recessive
<i>RYR1</i>	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
<i>RYR2</i>	180902	Ventricular tachycardia, catecholaminergic polymorphic, 1, 604772 (3), Autosomal dominant; Arrhythmogenic right ventricular dysplasia 2, 600996 (3), Autosomal dominant
<i>S1PR2</i>	605111	Deafness, autosomal recessive 68, 610419 (3), Autosomal recessive
<i>SACS</i>	604490	Spastic ataxia, Charlevoix-Saguenay type, 270550 (3), Autosomal recessive
<i>SAG</i>	181031	Oguchi disease-1, 258100 (3), Autosomal recessive; Retinitis pigmentosa 47, 613758 (3)
<i>SALL1</i>	602218	Townes-Brocks syndrome 1, 107480 (3), Autosomal dominant; Townes-Brocks branchiootorenal-like syndrome, 107480 (3), Autosomal dominant
<i>SALL2</i>	602219	?Coloboma, ocular, autosomal recessive, 216820 (3), Autosomal recessive
<i>SALL4</i>	607343	Duane-radial ray syndrome, 607323 (3), Autosomal dominant; IVIC syndrome, 147750 (3), Autosomal dominant
<i>SAMD12</i>	618073	Epilepsy, familial adult myoclonic, 1, 601068 (3), Autosomal dominant
<i>SAMD9</i>	610456	MIRAGE syndrome, 617053 (3), Autosomal dominant; Tumoral calcinosis, familial, normophosphatemic, 610455 (3), Autosomal recessive
<i>SAMD9L</i>	611170	Ataxia-pancytopenia syndrome, 159550 (3), Autosomal dominant
<i>SAMHD1</i>	606754	?Chilblain lupus 2, 614415 (3), Autosomal dominant; Aicardi-Goutieres syndrome 5, 612952 (3), Autosomal recessive
<i>SAR1B</i>	607690	Chylomicron retention disease, 246700 (3), Autosomal recessive
<i>SARDH</i>	604455	[Sarcosinemia], 268900 (3), Autosomal recessive
<i>SARS1 (SARS)</i>	607529	?Neurodevelopmental disorder with microcephaly, ataxia, and seizures, 617709 (3), Autosomal recessive

<i>SARS2</i>	612804	Hyperuricemia, pulmonary hypertension, renal failure, and alkalosis, 613845 (3), Autosomal recessive
<i>SASH1</i>	607955	?Cancer, alopecia, pigment dyscrasia, onychodystrophy, and keratoderma, 618373 (3), Autosomal recessive; Dyschromatosis universalis hereditaria 1, 127500 (3), Autosomal dominant
<i>SASS6</i>	609321	?Microcephaly 14, primary, autosomal recessive, 616402 (3), Autosomal recessive
<i>SATB2</i>	608148	Glass syndrome, 612313 (3), Autosomal dominant
<i>SBDS</i>	607444	{Aplastic anemia, susceptibility to}, 609135 (3); Shwachman-Diamond syndrome, 260400 (3), Autosomal recessive
<i>SBF1</i>	603560	Charcot-Marie-Tooth disease, type 4B3, 615284 (3), Autosomal recessive
<i>SBF2</i>	607697	Charcot-Marie-Tooth disease, type 4B2, 604563 (3), Autosomal recessive
<i>SC5D</i>	602286	Lathosterolosis, 607330 (3), Autosomal recessive
<i>SCAPER</i>	611611	Intellectual developmental disorder and retinitis pigmentosa, 618195 (3), Autosomal recessive
<i>SCARB1</i>	601040	[High density lipoprotein cholesterol level QTL6], 610762 (3)
<i>SCARB2</i>	602257	Epilepsy, progressive myoclonic 4, with or without renal failure, 254900 (3), Autosomal recessive
<i>SCARF2</i>	613619	Van den Ende-Gupta syndrome, 600920 (3), Autosomal recessive
<i>SCGB3A2</i>	606531	{Asthma, susceptibility to}, 600807 (3), Autosomal dominant
<i>SCN10A</i>	604427	Episodic pain syndrome, familial, 2, 615551 (3), Autosomal dominant
<i>SCN11A</i>	604385	Neuropathy, hereditary sensory and autonomic, type VII, 615548 (3), Autosomal dominant; Episodic pain syndrome, familial, 3, 615552 (3), Autosomal dominant
<i>SCN1A</i>	182389	Febrile seizures, familial, 3A, 604403 (3), Autosomal dominant; Migraine, familial hemiplegic, 3, 609634 (3), Autosomal dominant; Epilepsy, generalized, with febrile seizures plus, type 2, 604403 (3), Autosomal dominant; Epileptic encephalopathy, early infantile, 6 (Dravet syndrome), 607208 (3), Autosomal dominant
<i>SCN1B</i>	600235	Epileptic encephalopathy, early infantile, 52, 617350 (3), Autosomal recessive; Atrial fibrillation, familial, 13, 615377 (3), Autosomal dominant; Cardiac conduction defect, nonspecific, 612838 (3); Epilepsy, generalized, with febrile seizures plus, type 1, 604233 (3), Autosomal dominant; Brugada syndrome 5, 612838 (3)
<i>SCN2A</i>	182390	Episodic ataxia, type 9, 618924 (3), Autosomal dominant; Epileptic encephalopathy, early infantile, 11, 613721 (3), Autosomal dominant; Seizures, benign familial infantile, 3, 607745 (3), Autosomal dominant
<i>SCN2B</i>	601327	Atrial fibrillation, familial, 14, 615378 (3), Autosomal dominant

<i>SCN3A</i>	182391	Epilepsy, familial focal, with variable foci 4, 617935 (3), Autosomal dominant; Epileptic encephalopathy, early infantile, 62, 617938 (3), Autosomal dominant
<i>SCN3B</i>	608214	Brugada syndrome 7, 613120 (3), Autosomal dominant; Atrial fibrillation, familial, 16, 613120 (3), Autosomal dominant
<i>SCN4A</i>	603967	Hyperkalemic periodic paralysis, type 2, 170500 (3), Autosomal dominant; Paramyotonia congenita, 168300 (3), Autosomal dominant; Myotonia congenita, atypical, acetazolamide-responsive, 608390 (3), Autosomal dominant; Myasthenic syndrome, congenital, 16, 614198 (3), Autosomal recessive; Hypokalemic periodic paralysis, type 2, 613345 (3), Autosomal dominant
<i>SCN4B</i>	608256	Atrial fibrillation, familial, 17, 611819 (3), Autosomal dominant; Long QT syndrome 10, 611819 (3), Autosomal dominant
<i>SCN5A</i>	600163	Atrial fibrillation, familial, 10, 614022 (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); Long QT syndrome 3, 603830 (3), Autosomal dominant; Heart block, nonprogressive, 113900 (3), Autosomal dominant; Cardiomyopathy, dilated, 1E, 601154 (3), Autosomal dominant; Brugada syndrome 1, 601144 (3), Autosomal dominant; Heart block, progressive, type IA, 113900 (3), Autosomal dominant
<i>SCN8A</i>	600702	Seizures, benign familial infantile, 5, 617080 (3), Autosomal dominant; Cognitive impairment with or without cerebellar ataxia, 614306 (3), Autosomal dominant; ?Myoclonus, familial, 2, 618364 (3), Autosomal dominant; Epileptic encephalopathy, early infantile, 13, 614558 (3), Autosomal dominant
<i>SCN9A</i>	603415	Neuropathy, hereditary sensory and autonomic, type IID, 243000 (3), Autosomal recessive; Generalized epilepsy with febrile seizures plus, type 7, 613863 (3), Autosomal dominant; Small fiber neuropathy, 133020 (3), Autosomal dominant; Paroxysmal extreme pain disorder, 167400 (3), Autosomal dominant; Insensitivity to pain, congenital, 243000 (3), Autosomal recessive; Erythralgia, primary, 133020 (3), Autosomal dominant; Febrile seizures, familial, 3B, 613863 (3), Autosomal dominant
<i>SCNN1A</i>	600228	Pseudohypoaldosteronism, type I, 264350 (3), Autosomal recessive; ?Liddle syndrome 3, 618126 (3), Autosomal dominant; Bronchiectasis with or without elevated sweat chloride 2, 613021 (3), Autosomal dominant
<i>SCNN1B</i>	600760	Bronchiectasis with or without elevated sweat chloride 1, 211400 (3), Autosomal dominant; Pseudohypoaldosteronism, type I, 264350 (3), Autosomal recessive; Liddle syndrome 1, 177200 (3), Autosomal dominant

<i>SCNN1G</i>	600761	Bronchiectasis with or without elevated sweat chloride 3, 613071 (3), Autosomal dominant; Liddle syndrome 2, 618114 (3), Autosomal dominant; Pseudohypoaldosteronism, type I, 264350 (3), Autosomal recessive
<i>SCO1</i>	603644	Mitochondrial complex IV deficiency, 220110 (3), Autosomal recessive, Mitochondrial
<i>SCO2</i>	604272	Myopia 6, 608908 (3), Autosomal dominant; Cardioencephalomyopathy, fatal infantile, due to cytochrome c oxidase deficiency 1, 604377 (3), Autosomal recessive
<i>SCP2</i>	184755	?Leukoencephalopathy with dystonia and motor neuropathy, 613724 (3), Autosomal recessive
<i>SCYL1</i>	607982	Spinocerebellar ataxia, autosomal recessive 21, 616719 (3), Autosomal recessive
<i>SCYL2</i>	616365	Arthrogryposis multiplex congenita 4, neurogenic, with agenesis of the corpus callosum, 618766 (3), Autosomal recessive
<i>SDC3</i>	186357	{Obesity, association with}, 601665 (3), Autosomal recessive, Autosomal dominant, Multifactorial
<i>SDCCAG8</i>	613524	Bardet-Biedl syndrome 16, 615993 (3), Autosomal recessive; Senior-Loken syndrome 7, 613615 (3)
<i>SDHA</i>	600857	Cardiomyopathy, dilated, 1GG, 613642 (3), Autosomal recessive; Leigh syndrome, 256000 (3), Autosomal recessive, Mitochondrial; Paragangliomas 5, 614165 (3), Autosomal dominant; Mitochondrial respiratory chain complex II deficiency, 252011 (3), Autosomal recessive
<i>SDHAF1</i>	612848	Mitochondrial complex II deficiency, 252011 (3), Autosomal recessive
<i>SDHAF2</i>	613019	Paragangliomas 2, 601650 (3), Autosomal dominant
<i>SDHB</i>	185470	Gastrointestinal stromal tumor, 606764 (3), Autosomal dominant, Isolated cases; Pheochromocytoma, 171300 (3), Autosomal dominant; Paragangliomas 4, 115310 (3), Autosomal dominant; Paraganglioma and gastric stromal sarcoma, 606864 (3)
<i>SDHC</i>	602413	Paragangliomas 3, 605373 (3), Autosomal dominant; Paraganglioma and gastric stromal sarcoma, 606864 (3); Gastrointestinal stromal tumor, 606764 (3), Autosomal dominant, Isolated cases
<i>SDHD</i>	602690	Paragangliomas 1, with or without deafness, 168000 (3), Autosomal dominant; Mitochondrial complex II deficiency, 252011 (3), Autosomal recessive; Paraganglioma and gastric stromal sarcoma, 606864 (3); Pheochromocytoma, 171300 (3), Autosomal dominant
<i>SDR9C7</i>	609769	Ichthyosis, congenital, autosomal recessive 13, 617574 (3), Autosomal recessive
<i>SEC23A</i>	610511	Craniolenticulosutural dysplasia, 607812 (3), Autosomal recessive

<i>SEC23B</i>	610512	?Cowden syndrome 7, 616858 (3), Autosomal dominant; Dyserythropoietic anemia, congenital, type II, 224100 (3), Autosomal recessive
<i>SEC24D</i>	607186	Cole-Carpenter syndrome 2, 616294 (3), Autosomal recessive
<i>SEC31A</i>	610257	?Neurodevelopmental disorder with spastic quadriplegia, optic atrophy, seizures, and structural brain anomalies, 618651 (3), Autosomal recessive
<i>SEC61A1</i>	609213	Hyperuricemic nephropathy, familial juvenile, 4, 617056 (3), Autosomal dominant
<i>SEC63</i>	608648	Polycystic liver disease 2, 617004 (3), Autosomal dominant
<i>SECISBP2</i>	607693	Thyroid hormone metabolism, abnormal, 609698 (3), Autosomal recessive
<i>SELENBP1</i>	604188	Extraoral halitosis due to MTO deficiency, 618148 (3), Autosomal recessive
<i>SELENOI</i>	607915	Spastic paraplegia 81, autosomal recessive, 618768 (3), Autosomal recessive
<i>SELENON</i>	606210	Muscular dystrophy, rigid spine, 1, 602771 (3), Autosomal recessive; Myopathy, congenital, with fiber-type disproportion, 255310 (3), Autosomal recessive, Autosomal dominant
<i>SEMA3A</i>	603961	{Hypogonadotropic hypogonadism 16 with or without anosmia}, 614897 (3), Autosomal dominant
<i>SEMA3E</i>	608166	?CHARGE syndrome, 214800 (3), Autosomal dominant
<i>SEMA4A</i>	607292	Cone-rod dystrophy 10, 610283 (3), Autosomal recessive; Retinitis pigmentosa 35, 610282 (3), Autosomal recessive, Autosomal dominant
<i>SEMA6B</i>	608873	Epilepsy, progressive myoclonic, 11, 618876 (3), Autosomal dominant
<i>SEMA7A</i>	607961	[Blood group, John-Milton-Hagen system], 614745 (3)
<i>SEPSECS</i>	613009	Pontocerebellar hypoplasia type 2D, 613811 (3), Autosomal recessive
<i>SEPTIN12</i> (<i>SEPT12</i>)	611562	Spermatogenic failure 10, 614822 (3), Autosomal dominant
<i>SEPTIN9</i> (<i>SEPT9</i>)	604061	Amyotrophy, hereditary neuralgic, 162100 (3), Autosomal dominant
<i>SERAC1</i>	614725	3-methylglutaconic aciduria with deafness, encephalopathy, and Leigh-like syndrome, 614739 (3), Autosomal recessive
<i>SERPINA1</i>	107400	Hemorrhagic diathesis due to antithrombin Pittsburgh, 613490 (3), Autosomal recessive; Emphysema-cirrhosis, due to AAT deficiency, 613490 (3), Autosomal recessive; Emphysema due to AAT deficiency, 613490 (3), Autosomal recessive
<i>SERPINA3</i>	107280	Cerebrovascular disease, occlusive (3); Alpha-1-antichymotrypsin deficiency (3)
<i>SERPINA6</i>	122500	Corticosteroid-binding globulin deficiency, 611489 (3), Autosomal recessive, Autosomal dominant
<i>SERPINA7</i>	314200	[Thyroxine-binding globulin QTL], 300932 (3), X-linked

<i>SERPINB6</i>	173321	?Deafness, autosomal recessive 91, 613453 (3), Autosomal recessive
<i>SERPINB7</i>	603357	Palmoplantar keratoderma, Nagashima type, 615598 (3), Autosomal recessive
<i>SERPINB8</i>	601697	Peeling skin syndrome 5, 617115 (3), Autosomal recessive
<i>SERPINC1</i>	107300	Thrombophilia due to antithrombin III deficiency, 613118 (3), Autosomal recessive, Autosomal dominant
<i>SERPIND1</i>	142360	Thrombophilia due to heparin cofactor II deficiency, 612356 (3), Autosomal dominant
<i>SERPINE1</i>	173360	{Transcription of plasminogen activator inhibitor, modulator of} (3); Plasminogen activator inhibitor-1 deficiency, 613329 (3), Autosomal recessive, Autosomal dominant
<i>SERPINF1</i>	172860	Osteogenesis imperfecta, type VI, 613982 (3), Autosomal recessive
<i>SERPINF2</i>	613168	Alpha-2-plasmin inhibitor deficiency, 262850 (3), Autosomal recessive
<i>SERPING1</i>	606860	Angioedema, hereditary, types I and II, 106100 (3), Autosomal recessive, Autosomal dominant; Complement component 4, partial deficiency of, 120790 (3), Autosomal dominant
<i>SERPINH1</i>	600943	Osteogenesis imperfecta, type X, 613848 (3), Autosomal recessive; {Preterm premature rupture of the membranes, susceptibility to}, 610504 (3)
<i>SERPINI1</i>	602445	Encephalopathy, familial, with neuroserpin inclusion bodies, 604218 (3), Autosomal dominant
<i>SET</i>	600960	Mental retardation, autosomal dominant 58, 618106 (3), Autosomal dominant
<i>SETBP1</i>	611060	Mental retardation, autosomal dominant 29, 616078 (3), Autosomal dominant; Schinzel-Giedion midface retraction syndrome, 269150 (3), Autosomal dominant
<i>SETD1A</i>	611052	Epilepsy, early-onset, with or without developmental delay, 618832 (3), Autosomal dominant
<i>SETD2</i>	612778	Luscan-Lumish syndrome, 616831 (3), Autosomal dominant
<i>SETD5</i>	615743	Mental retardation, autosomal dominant 23, 615761 (3), Autosomal dominant
<i>SETX</i>	608465	Spinocerebellar ataxia, autosomal recessive, with axonal neuropathy 2, 606002 (3), Autosomal recessive; Amyotrophic lateral sclerosis 4, juvenile, 602433 (3), Autosomal dominant
<i>SF3B1</i>	605590	Myelodysplastic syndrome, somatic, 614286 (3)
<i>SF3B4</i>	605593	Acrofacial dysostosis 1, Nager type, 154400 (3), Autosomal dominant
<i>SFRP4</i>	606570	Pyle disease, 265900 (3), Autosomal recessive
<i>SFTPA2</i>	178642	Pulmonary fibrosis, idiopathic, 178500 (3), Autosomal dominant
<i>SFTPB</i>	178640	Surfactant metabolism dysfunction, pulmonary, 1, 265120 (3), Autosomal recessive

<i>SFTPC</i>	178620	Surfactant metabolism dysfunction, pulmonary, 2, 610913 (3), Autosomal dominant
<i>SFXN4</i>	615564	Combined oxidative phosphorylation deficiency 18, 615578 (3), Autosomal recessive
<i>SGCA</i>	600119	Muscular dystrophy, limb-girdle, autosomal recessive 3, 608099 (3), Autosomal recessive
<i>SGCB</i>	600900	Muscular dystrophy, limb-girdle, autosomal recessive 4, 604286 (3), Autosomal recessive
<i>SGCD</i>	601411	Cardiomyopathy, dilated, 1L, 606685 (3); Muscular dystrophy, limb-girdle, autosomal recessive 6, 601287 (3), Autosomal recessive
<i>SGCE</i>	604149	Dystonia-11, myoclonic, 159900 (3), Autosomal dominant
<i>SGCG</i>	608896	Muscular dystrophy, limb-girdle, autosomal recessive 5, 253700 (3), Autosomal recessive
<i>SGMS2</i>	611574	Calvarial doughnut lesions with bone fragility with or without spondylometaphyseal dysplasia, 126550 (3), Autosomal dominant
<i>SGO1</i>	609168	Chronic atrial and intestinal dysrhythmia, 616201 (3), Autosomal recessive
<i>SGPL1</i>	603729	Nephrotic syndrome, type 14, 617575 (3), Autosomal recessive
<i>SGSH</i>	605270	Mucopolysaccharidosis type IIIA (Sanfilippo A), 252900 (3), Autosomal recessive
<i>SH2B3</i>	605093	Myelofibrosis, somatic, 254450 (3); Thrombocythemia, somatic, 187950 (3); Erythrocytosis, somatic, 133100 (3)
<i>SH2D1A</i>	300490	Lymphoproliferative syndrome, X-linked, 1, 308240 (3), X-linked recessive
<i>SH3BP2</i>	602104	Cherubism, 118400 (3), Autosomal dominant
<i>SH3GL1</i>	601768	Leukemia, acute myeloid, 601626 (1), Autosomal dominant, Somatic mutation
<i>SH3KBP1</i>	300374	?Immunodeficiency 61, 300310 (3), X-linked recessive
<i>SH3PXD2B</i>	613293	Frank-ter Haar syndrome, 249420 (3), Autosomal recessive
<i>SH3TC2</i>	608206	Charcot-Marie-Tooth disease, type 4C, 601596 (3), Autosomal recessive; Mononeuropathy of the median nerve, mild, 613353 (3), Autosomal dominant
<i>SHANK2</i>	603290	{Autism susceptibility 17}, 613436 (3)
<i>SHANK3</i>	606230	{Schizophrenia 15}, 613950 (3), Autosomal dominant; Phelan-McDermid syndrome, 606232 (3), Autosomal dominant
<i>SHH</i>	600725	Schizencephaly, 269160 (3); Microphthalmia with coloboma 5, 611638 (3), Autosomal dominant; Single median maxillary central incisor, 147250 (3), Autosomal dominant; Holoprosencephaly 3, 142945 (3), Autosomal dominant
<i>SHOC2</i>	602775	Noonan syndrome-like with loose anagen hair 1, 607721 (3), Autosomal dominant

<i>SHOX</i>	400020	Leri-Weill dyschondrosteosis, 127300 (3), Pseudoautosomal dominant; Langer mesomelic dysplasia, 249700 (3), Pseudoautosomal recessive; Short stature, idiopathic familial, 300582 (3)
<i>SHPK</i>	605060	[Sedoheptulokinase deficiency], 617213 (3), Autosomal recessive
<i>SHROOM4</i>	300579	Stocco dos Santos X-linked mental retardation syndrome, 300434 (3), X-linked
<i>SI</i>	609845	Sucrase-isomaltase deficiency, congenital, 222900 (3), Autosomal recessive
<i>SIAE</i>	610079	{Autoimmune disease, susceptibility to, 6}, 613551 (3)
<i>SIGMAR1</i>	601978	?Amyotrophic lateral sclerosis 16, juvenile, 614373 (3), Autosomal recessive; ?Spinal muscular atrophy, distal, autosomal recessive, 2, 605726 (3), Autosomal recessive
<i>SIK1</i>	605705	Epileptic encephalopathy, early infantile, 30, 616341 (3), Autosomal dominant
<i>SIK3</i>	614776	?Spondyloepimetaphyseal dysplasia, Krakow type, 618162 (3), Autosomal recessive
<i>SIL1</i>	608005	Marinesco-Sjogren syndrome, 248800 (3), Autosomal recessive
<i>SIN3A</i>	607776	Witteveen-Kolk syndrome, 613406 (3), Autosomal dominant
<i>SIPA1L3</i>	616655	?Cataract 45, 616851 (3), Autosomal recessive
<i>SIX1</i>	601205	Deafness, autosomal dominant 23, 605192 (3), Autosomal dominant; Branchiootoc syndrome 3, 608389 (3), Autosomal dominant
<i>SIX3</i>	603714	Holoprosencephaly 2, 157170 (3), Autosomal dominant; Schizencephaly, 269160 (3)
<i>SIX5</i>	600963	Branchiootorenal syndrome 2, 610896 (3)
<i>SIX6</i>	606326	Optic disc anomalies with retinal and/or macular dystrophy, 212550 (3), Autosomal recessive
<i>SKI</i>	164780	Shprintzen-Goldberg syndrome, 182212 (3), Autosomal dominant
<i>SKIV2L</i>	600478	Trichohepatoenteric syndrome 2, 614602 (3), Autosomal recessive
<i>SLC10A2</i>	601295	Bile acid malabsorption, primary, 613291 (3), Autosomal recessive
<i>SLC10A7</i>	611459	Short stature, amelogenesis imperfecta, and skeletal dysplasia with scoliosis, 618363 (3), Autosomal recessive
<i>SLC11A1</i>	600266	{Buruli ulcer, susceptibility to}, 610446 (3); {Mycobacterium tuberculosis, susceptibility to infection by}, 607948 (3)
<i>SLC11A2</i>	600523	Anemia, hypochromic microcytic, with iron overload 1, 206100 (3), Autosomal recessive
<i>SLC12A1</i>	600839	Bartter syndrome, type 1, 601678 (3), Autosomal recessive
<i>SLC12A3</i>	600968	Gitelman syndrome, 263800 (3), Autosomal recessive
<i>SLC12A5</i>	606726	Epileptic encephalopathy, early infantile, 34, 616645 (3), Autosomal recessive; {Epilepsy, idiopathic generalized, susceptibility to, 14}, 616685 (3), Autosomal dominant

<i>SLC12A6</i>	604878	Agenesis of the corpus callosum with peripheral neuropathy, 218000 (3), Autosomal recessive
<i>SLC13A3</i>	606411	Leukoencephalopathy, acute reversible, with increased urinary alpha-ketoglutarate, 618384 (3), Autosomal recessive
<i>SLC13A5</i>	608305	Epileptic encephalopathy, early infantile, 25, 615905 (3), Autosomal recessive
<i>SLC14A1</i>	613868	[Blood group, Kidd], 111000 (3)
<i>SLC16A1</i>	600682	Monocarboxylate transporter 1 deficiency, 616095 (3), Autosomal recessive, Autosomal dominant; Hyperinsulinemic hypoglycemia, familial, 7, 610021 (3), Autosomal dominant; Erythrocyte lactate transporter defect, 245340 (3), Autosomal dominant
<i>SLC16A12</i>	611910	Cataract 47, juvenile, with microcornea, 612018 (3), Autosomal dominant
<i>SLC16A2</i>	300095	Allan-Herndon-Dudley syndrome, 300523 (3), X-linked
<i>SLC17A3</i>	611034	[Uric acid concentration, serum, QTL4], 612671 (3), Autosomal dominant; {Gout susceptibility 4}, 612671 (3), Autosomal dominant
<i>SLC17A5</i>	604322	Sialic acid storage disorder, infantile, 269920 (3), Autosomal recessive; Salla disease, 604369 (3), Autosomal recessive
<i>SLC17A8</i>	607557	Deafness, autosomal dominant 25, 605583 (3), Autosomal dominant
<i>SLC17A9</i>	612107	Porokeratosis 8, disseminated superficial actinic type, 616063 (3), Autosomal dominant
<i>SLC18A2</i>	193001	?Parkinsonism-dystonia, infantile, 2, 618049 (3), Autosomal recessive
<i>SLC18A3</i>	600336	Myasthenic syndrome, congenital, 21, presynaptic, 617239 (3), Autosomal recessive
<i>SLC19A2</i>	603941	Thiamine-responsive megaloblastic anemia syndrome, 249270 (3), Autosomal recessive
<i>SLC19A3</i>	606152	Thiamine metabolism dysfunction syndrome 2 (biotin- or thiamine-responsive encephalopathy type 2), 607483 (3), Autosomal recessive
<i>SLC1A1</i>	133550	{?Schizophrenia susceptibility 18}, 615232 (3); Dicarboxylic aminoaciduria, 222730 (3), Autosomal recessive
<i>SLC1A2</i>	600300	Epileptic encephalopathy, early infantile, 41, 617105 (3), Autosomal dominant
<i>SLC1A3</i>	600111	Episodic ataxia, type 6, 612656 (3), Autosomal dominant
<i>SLC1A4</i>	600229	Spastic tetraplegia, thin corpus callosum, and progressive microcephaly, 616657 (3), Autosomal recessive
<i>SLC20A2</i>	158378	Basal ganglia calcification, idiopathic, 1, 213600 (3), Autosomal dominant
<i>SLC22A12</i>	607096	Hypouricemia, renal, 220150 (3), Autosomal recessive
<i>SLC22A18</i>	602631	Lung cancer, somatic, 211980 (3); Breast cancer, somatic, 114480 (3); Rhabdomyosarcoma, somatic, 268210 (3)
<i>SLC22A4</i>	604190	{Rheumatoid arthritis, susceptibility to}, 180300 (3)

<i>SLC22A5</i>	603377	Carnitine deficiency, systemic primary, 212140 (3), Autosomal recessive
<i>SLC24A1</i>	603617	Night blindness, congenital stationary (complete), 1D, autosomal recessive, 613830 (3), Autosomal recessive
<i>SLC24A4</i>	609840	[Skin/hair/eye pigmentation 6, blue/green eyes], 210750 (3), Autosomal recessive; Amelogenesis imperfecta, type IIA5, 615887 (3), Autosomal recessive; [Skin/hair/eye pigmentation 6, blond/brown hair], 210750 (3), Autosomal recessive
<i>SLC24A5</i>	609802	[Skin/hair/eye pigmentation 4, fair/dark skin], 113750 (3), Autosomal recessive; Albinism, oculocutaneous, type VI, 113750 (3), Autosomal recessive
<i>SLC25A1</i>	190315	Myasthenic syndrome, congenital, 23, presynaptic, 618197 (3), Autosomal recessive; Combined D-2- and L-2-hydroxyglutaric aciduria, 615182 (3), Autosomal recessive
<i>SLC25A10</i>	606794	?Mitochondrial DNA depletion syndrome 19, 618972 (3)
<i>SLC25A11</i>	604165	Parangangliomas 6, 618464 (3), Autosomal dominant
<i>SLC25A12</i>	603667	Epileptic encephalopathy, early infantile, 39, 612949 (3), Autosomal recessive
<i>SLC25A13</i>	603859	Citrullinemia, adult-onset type II, 603471 (3), Autosomal recessive; Citrullinemia, type II, neonatal-onset, 605814 (3), Autosomal recessive
<i>SLC25A15</i>	603861	Hyperornithinemia-hyperammonemia-homocitrullinemia syndrome, 238970 (3), Autosomal recessive
<i>SLC25A19</i>	606521	Thiamine metabolism dysfunction syndrome 4 (progressive polyneuropathy type), 613710 (3), Autosomal recessive; Microcephaly, Amish type, 607196 (3), Autosomal recessive
<i>SLC25A20</i>	613698	Carnitine-acylcarnitine translocase deficiency, 212138 (3), Autosomal recessive
<i>SLC25A21</i>	607571	?Mitochondrial DNA depletion syndrome 18, 618811 (3), Autosomal recessive
<i>SLC25A22</i>	609302	Epileptic encephalopathy, early infantile, 3, 609304 (3), Autosomal recessive
<i>SLC25A24</i>	608744	Fontaine progeroid syndrome, 612289 (3), Autosomal dominant
<i>SLC25A26</i>	611037	Combined oxidative phosphorylation deficiency 28, 616794 (3), Autosomal recessive
<i>SLC25A3</i>	600370	Mitochondrial phosphate carrier deficiency, 610773 (3)
<i>SLC25A32</i>	610815	?Exercise intolerance, riboflavin-responsive, 616839 (3), Autosomal recessive
<i>SLC25A38</i>	610819	Anemia, sideroblastic, 2, pyridoxine-refractory, 205950 (3), Autosomal recessive
<i>SLC25A4</i>	103220	Mitochondrial DNA depletion syndrome 12B (cardiomyopathic type) AR, 615418 (3), Autosomal recessive; Mitochondrial DNA depletion syndrome 12A (cardiomyopathic type) AD, 617184 (3), Autosomal dominant; Progressive external ophthalmoplegia

		with mitochondrial DNA deletions, autosomal dominant 2, 609283 (3), Autosomal dominant
<i>SLC25A42</i>	610823	Metabolic crises, recurrent, with variable encephalomyopathic features and neurologic regression, 618416 (3), Autosomal recessive
<i>SLC25A46</i>	610826	Neuropathy, hereditary motor and sensory, type VIB, 616505 (3), Autosomal recessive
<i>SLC26A1</i>	610130	?Nephrolithiasis, calcium oxalate, 167030 (3), Autosomal recessive
<i>SLC26A2</i>	606718	De la Chapelle dysplasia, 256050 (3), Autosomal recessive; Atelosteogenesis, type II, 256050 (3), Autosomal recessive; Diastrophic dysplasia, broad bone-platyspondylic variant, 222600 (3), Autosomal recessive; Diastrophic dysplasia, 222600 (3), Autosomal recessive; Achondrogenesis Ib, 600972 (3), Autosomal recessive; Epiphyseal dysplasia, multiple, 4, 226900 (3), Autosomal recessive
<i>SLC26A3</i>	126650	Diarrhea 1, secretory chloride, congenital, 214700 (3), Autosomal recessive
<i>SLC26A4</i>	605646	Deafness, autosomal recessive 4, with enlarged vestibular aqueduct, 600791 (3), Autosomal recessive; Pendred syndrome, 274600 (3), Autosomal recessive
<i>SLC26A5</i>	604943	?Deafness, autosomal recessive 61, 613865 (3), Autosomal recessive
<i>SLC26A8</i>	608480	Spermatogenic failure 3, 606766 (3), Autosomal dominant
<i>SLC27A4</i>	604194	Ichthyosis prematurity syndrome, 608649 (3)
<i>SLC28A1</i>	606207	[Uridine-cytidineuria], 618477 (3), Autosomal recessive
<i>SLC29A3</i>	612373	Histiocytosis-lymphadenopathy plus syndrome, 602782 (3), Autosomal recessive
<i>SLC2A1</i>	138140	Dystonia 9, 601042 (3), Autosomal dominant; GLUT1 deficiency syndrome 1, infantile onset, severe, 606777 (3), Autosomal recessive, Autosomal dominant; Stomatin-deficient cryohydrocytosis with neurologic defects, 608885 (3), Autosomal dominant; GLUT1 deficiency syndrome 2, childhood onset, 612126 (3), Autosomal dominant; {Epilepsy, idiopathic generalized, susceptibility to, 12}, 614847 (3), Autosomal dominant
<i>SLC2A10</i>	606145	Arterial tortuosity syndrome, 208050 (3), Autosomal recessive
<i>SLC2A2</i>	138160	Fanconi-Bickel syndrome, 227810 (3), Autosomal recessive; {Diabetes mellitus, noninsulin-dependent}, 125853 (3), Autosomal dominant
<i>SLC2A9</i>	606142	{Uric acid concentration, serum, QTL 2}, 612076 (3), Autosomal recessive, Autosomal dominant; Hypouricemia, renal, 2, 612076 (3), Autosomal recessive, Autosomal dominant

<i>SLC30A10</i>	611146	Hypermanganesemia with dystonia 1, 613280 (3), Autosomal recessive
<i>SLC30A2</i>	609617	Zinc deficiency, transient neonatal, 608118 (3), Autosomal dominant
<i>SLC30A8</i>	611145	{Diabetes mellitus, noninsulin-dependent, susceptibility to}, 125853 (3), Autosomal dominant
<i>SLC30A9</i>	604604	?Birk-Landau-Perez syndrome, 617595 (3), Autosomal recessive
<i>SLC33A1</i>	603690	Spastic paraplegia 42, autosomal dominant, 612539 (3), Autosomal dominant; Congenital cataracts, hearing loss, and neurodegeneration, 614482 (3), Autosomal recessive
<i>SLC34A1</i>	182309	Nephrolithiasis/osteoporosis, hypophosphatemic, 1, 612286 (3), Autosomal dominant; Hypercalcemia, infantile, 2, 616963 (3), Autosomal recessive; ?Fanconi renal tubular syndrome 2, 613388 (3), Autosomal recessive
<i>SLC34A2</i>	604217	Pulmonary alveolar microlithiasis, 265100 (3), Autosomal recessive
<i>SLC34A3</i>	609826	Hypophosphatemic rickets with hypercalciuria, 241530 (3), Autosomal recessive
<i>SLC35A1</i>	605634	Congenital disorder of glycosylation, type II f, 603585 (3), Autosomal recessive
<i>SLC35A2</i>	314375	Congenital disorder of glycosylation, type II m, 300896 (3), Somatic mosaicism, X-linked dominant
<i>SLC35A3</i>	605632	?Arthrogryposis, mental retardation, and seizures, 615553 (3), Autosomal recessive
<i>SLC35C1</i>	605881	Congenital disorder of glycosylation, type II c, 266265 (3), Autosomal recessive
<i>SLC35D1</i>	610804	Schneckenbecken dysplasia, 269250 (3), Autosomal recessive
<i>SLC36A2</i>	608331	Iminoglycinuria, digenic, 242600 (3), Autosomal recessive, Digenic recessive; Hyperglycinuria, 138500 (3), Autosomal dominant
<i>SLC37A4</i>	602671	Glycogen storage disease Ic, 232240 (3), Autosomal recessive; Glycogen storage disease Ib, 232220 (3), Autosomal recessive
<i>SLC38A8</i>	615585	Foveal hypoplasia 2, with or without optic nerve misrouting and/or anterior segment dysgenesis, 609218 (3), Autosomal recessive
<i>SLC39A13</i>	608735	Ehlers-Danlos syndrome, spondylodysplastic type, 3, 612350 (3), Autosomal recessive
<i>SLC39A14</i>	608736	?Hyperostosis cranialis interna, 144755 (3), Autosomal dominant; Hypermanganesemia with dystonia 2, 617013 (3), Autosomal recessive
<i>SLC39A4</i>	607059	Acrodermatitis enteropathica, 201100 (3), Autosomal recessive
<i>SLC39A5</i>	608730	Myopia 24, autosomal dominant, 615946 (3), Autosomal dominant
<i>SLC39A8</i>	608732	Congenital disorder of glycosylation, type II n, 616721 (3), Autosomal recessive

<i>SLC3A1</i>	104614	Cystinuria, 220100 (3), Autosomal recessive, Autosomal dominant
<i>SLC40A1</i>	604653	Hemochromatosis, type 4, 606069 (3), Autosomal dominant
<i>SLC44A1</i>	606105	Neurodegeneration, childhood-onset, with ataxia, tremor, optic atrophy, and cognitive decline, 618868 (3), Autosomal recessive
<i>SLC44A4</i>	606107	?Deafness, autosomal dominant 72, 617606 (3), Autosomal dominant
<i>SLC45A1</i>	605763	Intellectual developmental disorder with neuropsychiatric features, 617532 (3), Autosomal recessive
<i>SLC45A2</i>	606202	[Skin/hair/eye pigmentation 5, dark/fair skin], 227240 (3), Autosomal recessive; [Skin/hair/eye pigmentation 5, black/nonblack hair], 227240 (3), Autosomal recessive; Albinism, oculocutaneous, type IV, 606574 (3), Autosomal recessive; [Skin/hair/eye pigmentation 5, dark/light eyes], 227240 (3), Autosomal recessive
<i>SLC46A1</i>	611672	Folate malabsorption, hereditary, 229050 (3), Autosomal recessive
<i>SLC4A1</i>	109270	Renal tubular acidosis, distal, AD, 179800 (3), Autosomal dominant; [Blood group, Swann], 601550 (3); [Blood group, Froese], 601551 (3); [Blood group, Waldner], 112010 (3); Renal tubular acidosis, distal, AR, 611590 (3), Autosomal recessive; Spherocytosis, type 4, 612653 (3), Autosomal dominant; Cryohydrocytosis, 185020 (3), Autosomal dominant; Ovalocytosis, SA type, 166900 (3), Autosomal dominant; [Malaria, resistance to], 611162 (3); [Blood group, Diego], 110500 (3); [Blood group, Wright], 112050 (3)
<i>SLC4A11</i>	610206	Corneal endothelial dystrophy, autosomal recessive, 217700 (3), Autosomal recessive; Corneal dystrophy, Fuchs endothelial, 4, 613268 (3); Corneal endothelial dystrophy and perceptive deafness, 217400 (3), Autosomal recessive
<i>SLC4A4</i>	603345	Renal tubular acidosis, proximal, with ocular abnormalities, 604278 (3), Autosomal recessive
<i>SLC52A1</i>	607883	Riboflavin deficiency, 615026 (3), Autosomal dominant
<i>SLC52A2</i>	607882	Brown-Vialetto-Van Laere syndrome 2, 614707 (3), Autosomal recessive
<i>SLC52A3</i>	613350	Brown-Vialetto-Van Laere syndrome 1, 211530 (3), Autosomal recessive; ?Fazio-Londe disease, 211500 (3), Autosomal recessive
<i>SLC5A1</i>	182380	Glucose/galactose malabsorption, 606824 (3), Autosomal recessive
<i>SLC5A2</i>	182381	Renal glucosuria, 233100 (3), Autosomal recessive, Autosomal dominant
<i>SLC5A5</i>	601843	Thyroid dyshormonogenesis 1, 274400 (3), Autosomal recessive
<i>SLC5A6</i>	604024	Neurodegeneration, infantile-onset, biotin-responsive, 618973 (3), Autosomal recessive

<i>SLC5A7</i>	608761	Neuronopathy, distal hereditary motor, type VIIA, 158580 (3), Autosomal dominant; Myasthenic syndrome, congenital, 20, presynaptic, 617143 (3), Autosomal recessive
<i>SLC6A1</i>	137165	Myoclonic-atonic epilepsy, 616421 (3), Autosomal dominant
<i>SLC6A17</i>	610299	Mental retardation, autosomal recessive 48, 616269 (3), Autosomal recessive
<i>SLC6A19</i>	608893	Iminoglycinuria, digenic, 242600 (3), Autosomal recessive, Digenic recessive; Hartnup disorder, 234500 (3), Autosomal recessive; Hyperglycinuria, 138500 (3), Autosomal dominant
<i>SLC6A2</i>	163970	?Orthostatic intolerance, 604715 (3)
<i>SLC6A20</i>	605616	Iminoglycinuria, digenic, 242600 (3), Autosomal recessive, Digenic recessive; Hyperglycinuria, 138500 (3), Autosomal dominant
<i>SLC6A3</i>	126455	{Nicotine dependence, protection against}, 188890 (3); Parkinsonism-dystonia, infantile, 1, 613135 (3), Autosomal recessive
<i>SLC6A4</i>	182138	{Anxiety-related personality traits}, 607834 (3); {Obsessive-compulsive disorder}, 164230 (3), Autosomal dominant
<i>SLC6A5</i>	604159	Hyperekplexia 3, 614618 (3), Autosomal recessive, Autosomal dominant
<i>SLC6A8</i>	300036	Cerebral creatine deficiency syndrome 1, 300352 (3), X-linked recessive
<i>SLC6A9</i>	601019	Glycine encephalopathy with normal serum glycine, 617301 (3), Autosomal recessive
<i>SLC7A14</i>	615720	Retinitis pigmentosa 68, 615725 (3), Autosomal recessive
<i>SLC7A7</i>	603593	Lysinuric protein intolerance, 222700 (3), Autosomal recessive
<i>SLC7A9</i>	604144	Cystinuria, 220100 (3), Autosomal recessive, Autosomal dominant
<i>SLC9A1</i>	107310	?Lichtenstein-Knorr syndrome, 616291 (3), Autosomal recessive
<i>SLC9A3</i>	182307	Diarrhea 8, secretory sodium, congenital, 616868 (3), Autosomal recessive
<i>SLC9A3R1</i>	604990	Nephrolithiasis/osteoporosis, hypophosphatemic, 2, 612287 (3), Autosomal dominant
<i>SLC9A6</i>	300231	Mental retardation, X-linked syndromic, Christianson type, 300243 (3), X-linked dominant
<i>SLC9A7</i>	300368	Intellectual developmental disorder, X-linked 108, 301024 (3), X-linked recessive
<i>SLC9A9</i>	608396	{?Autism susceptibility 16}, 613410 (3)
<i>SLCO1B1</i>	604843	Hyperbilirubinemia, Rotor type, digenic, 237450 (3), Digenic recessive
<i>SLCO1B3</i>	605495	Hyperbilirubinemia, Rotor type, digenic, 237450 (3), Digenic recessive
<i>SLCO2A1</i>	601460	Hypertrophic osteoarthropathy, primary, autosomal recessive 2, 614441 (3), Autosomal recessive
<i>SLFN14</i>	614958	Bleeding disorder, platelet-type, 20, 616913 (3), Autosomal dominant

<i>SLITRK1</i>	609678	?Trichotillomania, 613229 (3), Autosomal dominant, Multifactorial; Tourette syndrome, 137580 (3), Autosomal dominant
<i>SLITRK6</i>	609681	Deafness and myopia, 221200 (3), Autosomal recessive
<i>SLURP1</i>	606119	Meleda disease, 248300 (3), Autosomal recessive
<i>SLX4</i>	613278	Fanconi anemia, complementation group P, 613951 (3), Autosomal recessive
<i>SMAD3</i>	603109	Loeys-Dietz syndrome 3, 613795 (3), Autosomal dominant
<i>SMAD4</i>	600993	Polyposis, juvenile intestinal, 174900 (3), Autosomal dominant; Juvenile polyposis/hereditary hemorrhagic telangiectasia syndrome, 175050 (3), Autosomal dominant; Myhre syndrome, 139210 (3), Autosomal dominant; Pancreatic cancer, somatic, 260350 (3)
<i>SMAD6</i>	602931	Aortic valve disease 2, 614823 (3), Autosomal dominant; {Radioulnar synostosis, nonsyndromic}, 179300 (3), Autosomal dominant; {Craniosynostosis 7, susceptibility to}, 617439 (3), Autosomal dominant
<i>SMAD7</i>	602932	{Colorectal cancer, susceptibility to, 3}, 612229 (3)
<i>SMAD9</i>	603295	Pulmonary hypertension, primary, 2, 615342 (3), Autosomal dominant
<i>SMARCA2</i>	600014	Nicolaides-Baraitser syndrome, 601358 (3), Autosomal dominant
<i>SMARCA4</i>	603254	{Rhabdoid tumor predisposition syndrome 2}, 613325 (3), Autosomal dominant; Coffin-Siris syndrome 4, 614609 (3), Autosomal dominant
<i>SMARCA1</i>	612761	Huriez syndrome, 181600 (3), Autosomal dominant; Basan syndrome, 129200 (3), Autosomal dominant; Adermatoglyphia, 136000 (3), Autosomal dominant
<i>SMARCA1</i>	606622	Schimke immunoosseous dysplasia, 242900 (3), Autosomal recessive
<i>SMARCB1</i>	601607	Rhabdoid tumors, somatic, 609322 (3); {Schwannomatosis-1, susceptibility to}, 162091 (3), Autosomal dominant; Coffin-Siris syndrome 3, 614608 (3), Autosomal dominant; {Rhabdoid tumor predisposition syndrome 1}, 609322 (3), Autosomal dominant
<i>SMARCC2</i>	601734	Coffin-Siris syndrome 8, 618362 (3), Autosomal dominant
<i>SMARCD1</i>	601735	Coffin-Siris syndrome 11, 618779 (3), Autosomal dominant
<i>SMARCD2</i>	601736	Specific granule deficiency 2, 617475 (3), Autosomal recessive
<i>SMARCE1</i>	603111	{Meningioma, familial, susceptibility to}, 607174 (3), Autosomal dominant; Coffin-Siris syndrome 5, 616938 (3), Autosomal dominant
<i>SMC1A</i>	300040	Cornelia de Lange syndrome 2, 300590 (3), X-linked dominant; Epileptic encephalopathy, early infantile, 85, with or without midline brain defects, 301044 (3)
<i>SMC3</i>	606062	Cornelia de Lange syndrome 3, 610759 (3), Autosomal dominant
<i>SMCHD1</i>	614982	Fascioscapulohumeral muscular dystrophy 2, digenic, 158901 (3); Bosma arhinia microphthalmia syndrome, 603457 (3), Autosomal dominant

<i>SMG9</i>	613176	Heart and brain malformation syndrome, 616920 (3), Autosomal recessive
<i>SMIM1</i>	615242	[Blood group, Vel system], 615264 (3), Autosomal recessive
<i>SMN1</i>	600354	Spinal muscular atrophy-2, 253550 (3), Autosomal recessive; Spinal muscular atrophy-3, 253400 (3), Autosomal recessive; Spinal muscular atrophy-1, 253300 (3), Autosomal recessive; Spinal muscular atrophy-4, 271150 (3), Autosomal recessive
<i>SMN2</i>	601627	{Spinal muscular atrophy, type III, modifier of}, 253400 (3), Autosomal recessive
<i>SMO</i>	601500	Curry-Jones syndrome, somatic mosaic, 601707 (3); Pallister-Hall-like syndrome, 241800 (3), Autosomal recessive; Basal cell carcinoma, somatic, 605462 (3)
<i>SMOC1</i>	608488	Microphthalmia with limb anomalies, 206920 (3), Autosomal recessive
<i>SMOC2</i>	607223	Dentin dysplasia, type I, with microdontia and misshapen teeth, 125400 (3), Autosomal recessive
<i>SMPD1</i>	607608	Niemann-Pick disease, type A, 257200 (3), Autosomal recessive; Niemann-Pick disease, type B, 607616 (3), Autosomal recessive
<i>SMPD4</i>	610457	Neurodevelopmental disorder with microcephaly, arthrogryposis, and structural brain anomalies, 618622 (3), Autosomal recessive
<i>SMPX</i>	300226	Deafness, X-linked 4, 300066 (3), X-linked dominant
<i>SMS</i>	300105	Mental retardation, X-linked, Snyder-Robinson type, 309583 (3), X-linked recessive
<i>SNAI2</i>	602150	Waardenburg syndrome, type 2D, 608890 (3), Autosomal recessive; Piebaldism, 172800 (3), Autosomal dominant
<i>SNAP25</i>	600322	?Myasthenic syndrome, congenital, 18, 616330 (3), Autosomal dominant
<i>SNAP29</i>	604202	Cerebral dysgenesis, neuropathy, ichthyosis, and palmoplantar keratoderma syndrome, 609528 (3), Autosomal recessive
<i>SNCA</i>	163890	Dementia, Lewy body, 127750 (3), Autosomal dominant; Parkinson disease 1, 168601 (3), Autosomal dominant; Parkinson disease 4, 605543 (3), Autosomal dominant
<i>SNCB</i>	602569	Dementia, Lewy body, 127750 (3), Autosomal dominant
<i>SNIP1</i>	608241	Psychomotor retardation, epilepsy, and craniofacial dysmorphism, 614501 (3), Autosomal recessive
<i>SNRNP200</i>	601664	Retinitis pigmentosa 33, 610359 (3), Autosomal dominant
<i>SNRPB</i>	182282	Cerebrocostomandibular syndrome, 117650 (3), Autosomal dominant
<i>SNRPE</i>	128260	Hypotrichosis 11, 615059 (3), Autosomal dominant
<i>SNRPN</i>	182279	Prader-Willi syndrome, 176270 (3), Autosomal dominant
<i>SNTA1</i>	601017	Long QT syndrome 12, 612955 (3), Autosomal dominant
<i>SNX10</i>	614780	Osteopetrosis, autosomal recessive 8, 615085 (3), Autosomal recessive

<i>SNX14</i>	616105	Spinocerebellar ataxia, autosomal recessive 20, 616354 (3), Autosomal recessive
<i>SOBP</i>	613667	Mental retardation, anterior maxillary protrusion, and strabismus, 613671 (3), Autosomal recessive
<i>SOD1</i>	147450	Spastic tetraplegia and axial hypotonia, progressive, 618598 (3), Autosomal recessive; Amyotrophic lateral sclerosis 1, 105400 (3), Autosomal recessive, Autosomal dominant
<i>SOD2</i>	147460	{Microvascular complications of diabetes 6}, 612634 (3)
<i>SOD3</i>	185490	[Superoxide dismutase, elevated extracellular] (3)
<i>SOHLH1</i>	610224	Ovarian dysgenesis 5, 617690 (3), Autosomal recessive; Spermatogenic failure 32, 618115 (3), Autosomal dominant
<i>SON</i>	182465	ZTTK syndrome, 617140 (3), Autosomal dominant
<i>SORD</i>	182500	Sorbitol dehydrogenase deficiency with peripheral neuropathy, 618912 (3), Autosomal recessive
<i>SORT1</i>	602458	[Low density lipoprotein cholesterol level QTL6], 613589 (3), Autosomal dominant
<i>SOS1</i>	182530	Noonan syndrome 4, 610733 (3), Autosomal dominant; ?Fibromatosis, gingival, 1, 135300 (3), Autosomal dominant
<i>SOS2</i>	601247	Noonan syndrome 9, 616559 (3), Autosomal dominant
<i>SOST</i>	605740	Sclerosteosis 1, 269500 (3), Autosomal recessive; Van Buchem disease, 239100 (3), Autosomal recessive; Craniodiaphyseal dysplasia, autosomal dominant, 122860 (3), Autosomal dominant
<i>SOX10</i>	602229	Waardenburg syndrome, type 2E, with or without neurologic involvement, 611584 (3), Autosomal dominant; PCWH syndrome, 609136 (3), Autosomal dominant; Waardenburg syndrome, type 4C, 613266 (3), Autosomal dominant
<i>SOX11</i>	600898	Coffin-Siris syndrome 9, 615866 (3), Autosomal dominant
<i>SOX17</i>	610928	Vesicoureteral reflux 3, 613674 (3), Autosomal dominant
<i>SOX18</i>	601618	Hypotrichosis-lymphedema-telangiectasia-renal defect syndrome, 137940 (3), Autosomal dominant; Hypotrichosis-lymphedema-telangiectasia syndrome, 607823 (3), Autosomal recessive
<i>SOX2</i>	184429	Microphthalmia, syndromic 3, 206900 (3), Autosomal dominant; Optic nerve hypoplasia and abnormalities of the central nervous system, 206900 (3), Autosomal dominant
<i>SOX3</i>	313430	Mental retardation, X-linked, with isolated growth hormone deficiency, 300123 (3); Panhypopituitarism, X-linked, 312000 (3), X-linked
<i>SOX4</i>	184430	Coffin-Siris syndrome 10, 618506 (3), Autosomal dominant
<i>SOX5</i>	604975	Lamb-Shaffer syndrome, 616803 (3), Autosomal dominant
<i>SOX6</i>	607257	Tolchin-Le Caignec syndrome, 618971 (3), Autosomal dominant
<i>SOX9</i>	608160	Acampomelic campomelic dysplasia, 114290 (3), Autosomal dominant; Campomelic dysplasia, 114290 (3), Autosomal

dominant; Campomelic dysplasia with autosomal sex reversal, 114290 (3), Autosomal dominant

<i>SP110</i>	604457	{Mycobacterium tuberculosis, susceptibility to}, 607948 (3); Hepatic venoocclusive disease with immunodeficiency, 235550 (3), Autosomal recessive
<i>SP7</i>	606633	Osteogenesis imperfecta, type XII, 613849 (3), Autosomal recessive
<i>SPAG1</i>	603395	Ciliary dyskinesia, primary, 28, 615505 (3), Autosomal recessive
<i>SPARC</i>	182120	Osteogenesis imperfecta, type XVII, 616507 (3), Autosomal recessive
<i>SPART</i>	607111	Troyer syndrome, 275900 (3), Autosomal recessive
<i>SPAST</i>	604277	Spastic paraplegia 4, autosomal dominant, 182601 (3), Autosomal dominant
<i>SPATA16</i>	609856	?Spermatogenic failure 6, 102530 (3), Autosomal recessive
<i>SPATA5</i>	613940	Epilepsy, hearing loss, and mental retardation syndrome, 616577 (3), Autosomal recessive
<i>SPATA7</i>	609868	Retinitis pigmentosa, juvenile, autosomal recessive, 604232 (3); Leber congenital amaurosis 3, 604232 (3)
<i>SPECC1L</i>	614140	Hypertelorism, Teebi type, 145420 (3), Autosomal dominant; ?Facial clefting, oblique, 1, 600251 (3), Autosomal dominant; Opitz GBBB syndrome, type II, 145410 (3), Autosomal dominant
<i>SPEF2</i>	610172	Spermatogenic failure 43, 618751 (3), Autosomal recessive
<i>SPEG</i>	615950	Centronuclear myopathy 5, 615959 (3), Autosomal recessive
<i>SPG11</i>	610844	Charcot-Marie-Tooth disease, axonal, type 2X, 616668 (3), Autosomal recessive; Spastic paraplegia 11, autosomal recessive, 604360 (3), Autosomal recessive; Amyotrophic lateral sclerosis 5, juvenile, 602099 (3), Autosomal recessive
<i>SPG21</i>	608181	Mast syndrome, 248900 (3), Autosomal recessive
<i>SPG7</i>	602783	Spastic paraplegia 7, autosomal recessive, 607259 (3), Autosomal recessive, Autosomal dominant
<i>SPINK1</i>	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
<i>SPINK2</i>	605753	?Spermatogenic failure 29, 618091 (3), Autosomal recessive
<i>SPINK5</i>	605010	Netherton syndrome, 256500 (3), Autosomal recessive
<i>SPINT2</i>	605124	Diarrhea 3, secretory sodium, congenital, syndromic, 270420 (3), Autosomal recessive
<i>SPNS2</i>	612584	?Deafness, autosomal recessive 115, 618457 (3), Autosomal recessive
<i>SPOP</i>	602650	Nabais Sa-de Vries syndrome, type 1, 618828 (3), Autosomal dominant; Nabais Sa-de Vries syndrome, type 2, 618829 (3), Autosomal dominant

<i>SPR</i>	182125	Dystonia, dopa-responsive, due to sepiapterin reductase deficiency, 612716 (3), Autosomal recessive, ?Autosomal dominant
<i>SPRED1</i>	609291	Legius syndrome, 611431 (3), Autosomal dominant
<i>SPRTN</i>	616086	Ruijs-Aalfs syndrome, 616200 (3), Autosomal recessive
<i>SPRY2</i>	602466	{?IgA nephropathy, susceptibility to, 3}, 616818 (3), Autosomal dominant
<i>SPRY4</i>	607984	Hypogonadotropic hypogonadism 17 with or without anosmia, 615266 (3), Autosomal dominant
<i>SPTA1</i>	182860	Pyropoikilocytosis, 266140 (3), Autosomal recessive; Elliptocytosis-2, 130600 (3), Autosomal dominant; Spherocytosis, type 3, 270970 (3), Autosomal recessive
<i>SPTAN1</i>	182810	Epileptic encephalopathy, early infantile, 5, 613477 (3), Autosomal dominant
<i>SPTB</i>	182870	Elliptocytosis-3, 617948 (3); Spherocytosis, type 2, 616649 (3), Autosomal dominant; Anemia, neonatal hemolytic, fatal or near-fatal, 617948 (3)
<i>SPTBN2</i>	604985	Spinocerebellar ataxia, autosomal recessive 14, 615386 (3), Autosomal recessive; Spinocerebellar ataxia 5, 600224 (3), Autosomal dominant
<i>SPTBN4</i>	606214	Neurodevelopmental disorder with hypotonia, neuropathy, and deafness, 617519 (3), Autosomal recessive
<i>SPTLC1</i>	605712	Neuropathy, hereditary sensory and autonomic, type IA, 162400 (3), Autosomal dominant
<i>SPTLC2</i>	605713	Neuropathy, hereditary sensory and autonomic, type IC, 613640 (3), Autosomal dominant
<i>SQSTM1</i>	601530	Frontotemporal dementia and/or amyotrophic lateral sclerosis 3, 616437 (3), Autosomal dominant; Neurodegeneration with ataxia, dystonia, and gaze palsy, childhood-onset, 617145 (3), Autosomal recessive; Myopathy, distal, with rimmed vacuoles, 617158 (3), Autosomal dominant; Paget disease of bone 3, 167250 (3), Autosomal dominant
<i>SRC</i>	190090	Colon cancer, advanced, somatic, 114500 (3); ?Thrombocytopenia 6, 616937 (3), Autosomal dominant
<i>SRCAP</i>	611421	Floating-Harbor syndrome, 136140 (3), Autosomal dominant
<i>SRD5A2</i>	607306	Pseudovaginal perineoscrotal hypospadias, 264600 (3), Autosomal recessive
<i>SRD5A3</i>	611715	Kahrizi syndrome, 612713 (3), Autosomal recessive; Congenital disorder of glycosylation, type Iq, 612379 (3), Autosomal recessive
<i>SRGAP1</i>	606523	{Thyroid cancer, nonmedullary, 2}, 188470 (3), Autosomal dominant, Somatic mutation
<i>SRP54</i>	604857	Neutropenia, severe congenital, 8, autosomal dominant, 618752 (3), Autosomal dominant
<i>SRP72</i>	602122	Bone marrow failure syndrome 1, 614675 (3), Autosomal dominant

<i>SRPX2</i>	300642	?Rolandic epilepsy, mental retardation, and speech dyspraxia, 300643 (3)
<i>SRY</i>	480000	46XY sex reversal 1, 400044 (3), Y-linked; 46XX sex reversal 1, 400045 (4), X-linked dominant
<i>SS18</i>	600192	Sarcoma, synovial (1)
<i>SSR4</i>	300090	Congenital disorder of glycosylation, type Iy, 300934 (3), X-linked recessive
<i>SSTR5</i>	182455	Somatostatin analog, resistance to (3)
<i>SSX1</i>	312820	?Sarcoma, synovial, 300813 (3)
<i>SSX2</i>	300192	?Sarcoma, synovial, 300813 (3)
<i>ST14</i>	606797	Ichthyosis, congenital, autosomal recessive 11, 602400 (3), Autosomal recessive
<i>ST3GAL3</i>	606494	Mental retardation, autosomal recessive 12, 611090 (3), Autosomal recessive; ?Epileptic encephalopathy, early infantile, 15, 615006 (3), Autosomal recessive
<i>ST3GAL5</i>	604402	Salt and pepper developmental regression syndrome, 609056 (3), Autosomal recessive
<i>STAC3</i>	615521	Myopathy, congenital, Baily-Bloch, 255995 (3), Autosomal recessive
<i>STAG1</i>	604358	Mental retardation, autosomal dominant 47, 617635 (3), Autosomal dominant
<i>STAG2</i>	300826	Mullegama-Klein-Martinez syndrome, 301022 (3), X-linked; Holoprosencephaly 13, X-linked, 301043 (3), X-linked recessive, X-linked dominant
<i>STAG3</i>	608489	Premature ovarian failure 8, 615723 (3), Autosomal recessive
<i>STAMBP</i>	606247	Microcephaly-capillary malformation syndrome, 614261 (3), Autosomal recessive
<i>STAR</i>	600617	Lipoid adrenal hyperplasia, 201710 (3), Autosomal recessive
<i>STARD7</i>	616712	Epilepsy, familial adult myoclonic, 2, 607876 (3), Autosomal dominant
<i>STAT1</i>	600555	Immunodeficiency 31C, autosomal dominant, 614162 (3), Autosomal dominant; Immunodeficiency 31A, mycobacteriosis, autosomal dominant, 614892 (3), Autosomal dominant; Immunodeficiency 31B, mycobacterial and viral infections, autosomal recessive, 613796 (3), Autosomal recessive
<i>STAT2</i>	600556	Immunodeficiency 44, 616636 (3), Autosomal recessive; Pseudo-TORCH syndrome 3, 618886 (3), Autosomal recessive
<i>STAT3</i>	102582	Hyper-IgE recurrent infection syndrome, 147060 (3), Autosomal dominant; Autoimmune disease, multisystem, infantile-onset, 1, 615952 (3), Autosomal dominant
<i>STAT4</i>	600558	{Systemic lupus erythematosus, susceptibility to, 11}, 612253 (3)
<i>STAT5B</i>	604260	Growth hormone insensitivity with immune dysregulation 2, autosomal dominant, 618985 (3); Growth hormone insensitivity with immune dysregulation 1, autosomal recessive, 245590 (3); Leukemia, acute promyelocytic, somatic, 102578 (3)

<i>STEAP3</i>	609671	?Anemia, hypochromic microcytic, with iron overload 2, 615234 (3), Autosomal dominant
<i>STIL</i>	181590	Microcephaly 7, primary, autosomal recessive, 612703 (3), Autosomal recessive
<i>STIM1</i>	605921	Myopathy, tubular aggregate, 1, 160565 (3), Autosomal dominant; Immunodeficiency 10, 612783 (3), Autosomal recessive; Stormorken syndrome, 185070 (3), Autosomal dominant
<i>STING1 (TMEM173)</i>	612374	STING-associated vasculopathy, infantile-onset, 615934 (3), Autosomal dominant
<i>STK11</i>	602216	Testicular tumor, somatic, 273300 (3); Peutz-Jeghers syndrome, 175200 (3), Autosomal dominant; Melanoma, malignant, somatic (3); Pancreatic cancer, somatic, 260350 (3)
<i>STK4</i>	604965	T-cell immunodeficiency, recurrent infections, autoimmunity, and cardiac malformations, 614868 (3)
<i>STN1</i>	613128	Cerebroretinal microangiopathy with calcifications and cysts 2, 617341 (3), Autosomal recessive
<i>STOX1</i>	609397	Preeclampsia/eclampsia 4, 609404 (3)
<i>STRA6</i>	610745	Microphthalmia, isolated, with coloboma 8, 601186 (3), Autosomal recessive; Microphthalmia, syndromic 9, 601186 (3), Autosomal recessive
<i>STRADA</i>	608626	Polyhydramnios, megalencephaly, and symptomatic epilepsy, 611087 (3), Autosomal recessive
<i>STRC</i>	606440	Deafness, autosomal recessive 16, 603720 (3), Autosomal recessive
<i>STS</i>	300747	Ichthyosis, X-linked, 308100 (3), X-linked recessive
<i>STT3A</i>	601134	?Congenital disorder of glycosylation, type Iw, 615596 (3), Autosomal recessive
<i>STT3B</i>	608605	?Congenital disorder of glycosylation, type Ix, 615597 (3), Autosomal recessive
<i>STUB1</i>	607207	Spinocerebellar ataxia, autosomal recessive 16, 615768 (3), Autosomal recessive; ?Spinocerebellar ataxia 48, 618093 (3), Autosomal dominant
<i>STX11</i>	605014	Hemophagocytic lymphohistiocytosis, familial, 4, 603552 (3), Autosomal recessive
<i>STX16</i>	603666	Pseudohypoparathyroidism, type IB, 603233 (3), Autosomal dominant
<i>STX1B</i>	601485	Generalized epilepsy with febrile seizures plus, type 9, 616172 (3), Autosomal dominant
<i>STXBP1</i>	602926	Epileptic encephalopathy, early infantile, 4, 612164 (3), Autosomal dominant
<i>STXBP2</i>	601717	Hemophagocytic lymphohistiocytosis, familial, 5, 613101 (3)
<i>SUCLA2</i>	603921	Mitochondrial DNA depletion syndrome 5 (encephalomyopathic with or without methylmalonic aciduria), 612073 (3), Autosomal recessive

<i>SUCLG1</i>	611224	Mitochondrial DNA depletion syndrome 9 (encephalomyopathic type with methylmalonic aciduria), 245400 (3), Autosomal recessive
<i>SUFU</i>	607035	Basal cell nevus syndrome, 109400 (3), Autosomal dominant; Medulloblastoma, desmoplastic, 155255 (3), Autosomal recessive, Autosomal dominant, Somatic mutation; {Meningioma, familial, susceptibility to}, 607174 (3), Autosomal dominant; Joubert syndrome 32, 617757 (3), Autosomal recessive
<i>SUGCT</i>	609187	Glutaric aciduria III, 231690 (3), Autosomal recessive
<i>SULT2B1</i>	604125	Ichthyosis, congenital, autosomal recessive 14, 617571 (3), Autosomal recessive
<i>SUMF1</i>	607939	Multiple sulfatase deficiency, 272200 (3), Autosomal recessive
<i>SUMO1</i>	601912	?Orofacial cleft 10, 613705 (3), Isolated cases
<i>SUMO4</i>	608829	{Diabetes mellitus, insulin-dependent, 5}, 600320 (3)
<i>SUN5</i>	613942	Spermatogenic failure 16, 617187 (3), Autosomal recessive
<i>SUOX</i>	606887	Sulfite oxidase deficiency, 272300 (3), Autosomal recessive
<i>SURF1</i>	185620	Leigh syndrome, due to COX IV deficiency, 256000 (3), Autosomal recessive, Mitochondrial; Charcot-Marie-Tooth disease, type 4K, 616684 (3), Autosomal recessive
<i>SUZ12</i>	606245	Imagawa-Matsumoto syndrome, 618786 (3), Autosomal dominant
<i>SVBP</i>	617853	Neurodevelopmental disorder with ataxia, hypotonia, and microcephaly, 618569 (3), Autosomal recessive
<i>SYCE1</i>	611486	?Premature ovarian failure 12, 616947 (3), Autosomal recessive; ?Spermatogenic failure 15, 616950 (3), Autosomal recessive
<i>SYCP2</i>	604105	Spermatogenic failure 1, 258150 (3), Autosomal recessive
<i>SYCP3</i>	604759	Spermatogenic failure 4, 270960 (3), Autosomal dominant; Pregnancy loss, recurrent, 4, 270960 (3), Autosomal dominant
<i>SYN1</i>	313440	Epilepsy, X-linked, with variable learning disabilities and behavior disorders, 300491 (3), X-linked recessive, X-linked dominant
<i>SYN2</i>	600755	{Schizophrenia, susceptibility to}, 181500 (3), Autosomal dominant
<i>SYNE1</i>	608441	Arthrogryposis multiplex congenita 3, myogenic type, 618484 (3), Autosomal recessive; Spinocerebellar ataxia, autosomal recessive 8, 610743 (3), Autosomal recessive; Emery-Dreifuss muscular dystrophy 4, autosomal dominant, 612998 (3), Autosomal dominant
<i>SYNE2</i>	608442	Emery-Dreifuss muscular dystrophy 5, autosomal dominant, 612999 (3), Autosomal dominant
<i>SYNE4</i>	615535	Deafness, autosomal recessive 76, 615540 (3), Autosomal recessive
<i>SYNGAP1</i>	603384	Mental retardation, autosomal dominant 5, 612621 (3), Autosomal dominant

<i>SYNJ1</i>	604297	Epileptic encephalopathy, early infantile, 53, 617389 (3), Autosomal recessive; Parkinson disease 20, early-onset, 615530 (3), Autosomal recessive
<i>SYP</i>	313475	Mental retardation, X-linked 96, 300802 (3), X-linked recessive
<i>SYT1</i>	185605	Baker-Gordon syndrome, 618218 (3), Autosomal dominant
<i>SYT14</i>	610949	?Spinocerebellar ataxia, autosomal recessive 11, 614229 (3), Autosomal recessive
<i>SYT2</i>	600104	Myasthenic syndrome, congenital, 7, presynaptic, 616040 (3), Autosomal dominant
<i>SZT2</i>	615463	Epileptic encephalopathy, early infantile, 18, 615476 (3), Autosomal recessive
<i>TAB2</i>	605101	Congenital heart defects, nonsyndromic, 2, 614980 (3), Autosomal dominant
<i>TAC3</i>	162330	Hypogonadotropic hypogonadism 10 with or without anosmia, 614839 (3), Autosomal recessive
<i>TACO1</i>	612958	Mitochondrial complex IV deficiency, 220110 (3), Autosomal recessive, Mitochondrial
<i>TACR3</i>	162332	Hypogonadotropic hypogonadism 11 with or without anosmia, 614840 (3), Autosomal recessive
<i>TACSTD2</i>	137290	Corneal dystrophy, gelatinous drop-like, 204870 (3), Autosomal recessive
<i>TAF1</i>	313650	Dystonia-Parkinsonism, X-linked, 314250 (3), X-linked recessive; Mental retardation, X-linked, syndromic 33, 300966 (3), X-linked recessive
<i>TAF13</i>	600774	Mental retardation, autosomal recessive 60, 617432 (3), Autosomal recessive
<i>TAF15</i>	601574	Chondrosarcoma, extraskeletal myxoid, 612237 (1)
<i>TAF2</i>	604912	Mental retardation, autosomal recessive 40, 615599 (3), Autosomal recessive
<i>TAF4B</i>	601689	?Spermatogenic failure 13, 615841 (3), Autosomal recessive
<i>TAF6</i>	602955	Alazami-Yuan syndrome, 617126 (3), Autosomal recessive
<i>TAGAP</i>	609667/612521	-/{Diabetes mellitus, insulin-dependent, 21}, 612521 (2)
<i>TAL1</i>	187040	Leukemia, T-cell acute lymphocytic, somatic, 613065 (3)
<i>TAL2</i>	186855	Leukemia, T-cell acute lymphocytic, somatic, 613065 (3)
<i>TALDO1</i>	602063	Transaldolase deficiency, 606003 (3), Autosomal recessive
<i>TANC2</i>	615047	Intellectual developmental disorder with autistic features and language delay, with or without seizures, 618906 (3), Autosomal dominant
<i>TANGO2</i>	616830	Metabolic encephalomyopathic crises, recurrent, with rhabdomyolysis, cardiac arrhythmias, and neurodegeneration, 616878 (3), Autosomal recessive
<i>TAP1</i>	170260	Bare lymphocyte syndrome, type I, 604571 (3), Autosomal recessive
<i>TAP2</i>	170261	Bare lymphocyte syndrome, type I, due to TAP2 deficiency, 604571 (3), Autosomal recessive

<i>TAPBP</i>	601962	Bare lymphocyte syndrome, type I, 604571 (3), Autosomal recessive
<i>TAPT1</i>	612758	Osteochondrodysplasia, complex lethal, Symoens-Barnes-Gistelinck type, 616897 (3), Autosomal recessive
<i>TARDBP</i>	605078	Frontotemporal lobar degeneration, TARDBP-related, 612069 (3), Autosomal dominant; Amyotrophic lateral sclerosis 10, with or without FTD, 612069 (3), Autosomal dominant
<i>TARS1 (TARS)</i>	187790	Trichothiodystrophy 7, nonphotosensitive, 618546 (3), Autosomal recessive
<i>TARS2</i>	612805	?Combined oxidative phosphorylation deficiency 21, 615918 (3), Autosomal recessive
<i>TAS2R16</i>	604867	[Beta-glycopyranoside tasting], (3) {Alcohol dependence, susceptibility to}, 617956 (3), Autosomal dominant
<i>TAS2R38</i>	607751	[Phenylthiocarbamide tasting], 171200 (3), Autosomal dominant
<i>TASP1</i>	608270	Suleiman-El-Hattab syndrome, 618950 (3), Autosomal recessive
<i>TAT</i>	613018	Tyrosinemia, type II, 276600 (3), Autosomal recessive
<i>TAZ</i>	300394	Barth syndrome, 302060 (3), X-linked recessive
<i>TBC1D20</i>	611663	Warburg micro syndrome 4, 615663 (3), Autosomal recessive
<i>TBC1D23</i>	617687	Pontocerebellar hypoplasia, type 11, 617695 (3), Autosomal recessive
<i>TBC1D24</i>	613577	Epilepsy, rolandic, with proxysmal exercise-induce dystonia and writer's cramp, 608105 (3), Autosomal recessive; DOORS syndrome, 220500 (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; Deafness , autosomal recessive 86, 614617 (3), Autosomal recessive
<i>TBC1D4</i>	612465	{Diabetes mellitus, noninsulin-dependent, 5}, 616087 (3)
<i>TBC1D7</i>	612655	Macrocephaly/megalencephaly syndrome, autosomal recessive, 248000 (3), Autosomal recessive
<i>TBC1D8B</i>	301027	Nephrotic syndrome, type 20, 301028 (3), X-linked
<i>TBCD</i>	604649	Encephalopathy, progressive, early-onset, with brain atrophy and thin corpus callosum, 617193 (3), Autosomal recessive
<i>TBCE</i>	604934	Kenny-Caffey syndrome, type 1, 244460 (3), Autosomal recessive; Hypoparathyroidism-retardation-dysmorphism syndrome, 241410 (3), Autosomal recessive; Encephalopathy, progressive, with amyotrophy and optic atrophy, 617207 (3), Autosomal recessive
<i>TBCK</i>	616899	Hypotonia, infantile, with psychomotor retardation and characteristic facies 3, 616900 (3), Autosomal recessive
<i>TBK1</i>	604834	Frontotemporal dementia and/or amyotrophic lateral sclerosis 4, 616439 (3), Autosomal dominant; {Encephalopathy, acute, infection-induced (herpes-specific), susceptibility to, 8}, 617900 (3), Autosomal dominant
<i>TBL1X</i>	300196	Hypothyroidism, congenital, nongoitrous, 8, 301033 (3), X-linked

<i>TBL1XR1</i>	608628	Pierpont syndrome, 602342 (3), Autosomal dominant; Mental retardation, autosomal dominant 41, 616944 (3), Autosomal dominant
<i>TBL1Y</i>	400033	?Deafness, Y-linked 2, 400047 (3), Y-linked
<i>TBP</i>	600075	{Parkinson disease, susceptibility to}, 168600 (3), Autosomal dominant, Multifactorial; Spinocerebellar ataxia 17, 607136 (3), Autosomal dominant
<i>TBR1</i>	604616	Intellectual developmental disorder with autism and speech delay, 606053 (3), Autosomal dominant
<i>TBX1</i>	602054	Conotruncal anomaly face syndrome, 217095 (3); Velocardiofacial syndrome, 192430 (3), Autosomal dominant; DiGeorge syndrome, 188400 (3), Autosomal dominant; Tetralogy of Fallot, 187500 (3), Autosomal dominant
<i>TBX15</i>	604127	Cousin syndrome, 260660 (3), Autosomal recessive
<i>TBX18</i>	604613	Congenital anomalies of kidney and urinary tract 2, 143400 (3), Autosomal dominant
<i>TBX19</i>	604614	Adrenocorticotrophic hormone deficiency, 201400 (3), Autosomal recessive
<i>TBX2</i>	600747	Vertebral anomalies and variable endocrine and T-cell dysfunction, 618223 (3), Autosomal dominant
<i>TBX20</i>	606061	Atrial septal defect 4, 611363 (3)
<i>TBX21</i>	604895	{Asthma, aspirin-induced, susceptibility to}, 208550 (3), Autosomal recessive; Asthma and nasal polyps, 208550 (3), Autosomal recessive
<i>TBX22</i>	300307	?Abruzzo-Erickson syndrome, 302905 (3), X-linked; Cleft palate with ankyloglossia, 303400 (3), X-linked
<i>TBX3</i>	601621	Ulnar-mammary syndrome, 181450 (3), Autosomal dominant
<i>TBX4</i>	601719	Amelia, posterior, with pelvic and pulmonary hypoplasia syndrome, 601360 (3), Autosomal recessive; Ischiocoxopodopatellar syndrome with or without pulmonary arterial hypertension, 147891 (3), Autosomal dominant
<i>TBX5</i>	601620	Holt-Oram syndrome, 142900 (3), Autosomal dominant
<i>TBX6</i>	602427	Spondylocostal dysostosis 5, 122600 (3), Autosomal recessive, Autosomal dominant
<i>TBXA2R</i>	188070	{Bleeding disorder, platelet-type, 13, susceptibility to}, 614009 (3), Autosomal dominant
<i>TBXAS1</i>	274180	Ghosal hematodiaphyseal syndrome, 231095 (3), Autosomal recessive
<i>TBXT</i>	601397	{Neural tube defects, susceptibility to}, 182940 (3), Autosomal dominant; Sacral agenesis with vertebral anomalies, 615709 (3), Autosomal recessive
<i>TCAP</i>	604488	Cardiomyopathy, hypertrophic, 25, 607487 (3), Autosomal dominant; Muscular dystrophy, limb-girdle, autosomal recessive 7, 601954 (3), Autosomal recessive
<i>TCF12</i>	600480	Craniosynostosis 3, 615314 (3), Autosomal dominant

<i>TCF20</i>	603107	Developmental delay with variable intellectual impairment and behavioral abnormalities, 618430 (3), Autosomal dominant
<i>TCF3</i>	147141	Agammaglobulinemia 8, autosomal dominant, 616941 (3), Autosomal dominant
<i>TCF4</i>	602272	Corneal dystrophy, Fuchs endothelial, 3, 613267 (3), Autosomal dominant; Pitt-Hopkins syndrome, 610954 (3), Autosomal dominant
<i>TCF7L2</i>	602228	{Diabetes mellitus, type 2, susceptibility to}, 125853 (3), Autosomal dominant
<i>TCHH</i>	190370	?Uncombable hair syndrome 3, 617252 (3), Autosomal recessive
<i>TCIRG1</i>	604592	Osteopetrosis, autosomal recessive 1, 259700 (3), Autosomal recessive
<i>TCL1A</i>	186960	Leukemia/lymphoma, T-cell, 186960 (2)
<i>TCL1B</i>	603769	Leukemia/lymphoma, T-cell, 603769 (2)
<i>TCN2</i>	613441	Transcobalamin II deficiency, 275350 (3), Autosomal recessive
<i>TCOF1</i>	606847	Treacher Collins syndrome 1, 154500 (3), Autosomal dominant
<i>TCTN1</i>	609863	Joubert syndrome 13, 614173 (3), Autosomal recessive
<i>TCTN2</i>	613846	Joubert syndrome 24, 616654 (3), Autosomal recessive; ?Meckel syndrome 8, 613885 (3), Autosomal recessive
<i>TCTN3</i>	613847	Orofaciodigital syndrome IV, 258860 (3), Autosomal recessive; Joubert syndrome 18, 614815 (3), Autosomal recessive
<i>TDGF1</i>	187395	Forebrain defects (3)
<i>TDO2</i>	191070	[?Hypertryptophanemia], 600627 (3), Autosomal recessive
<i>TDP1</i>	607198	?Spinocerebellar ataxia, autosomal recessive, with axonal neuropathy 1, 607250 (3), Autosomal recessive
<i>TDP2</i>	605764	Spinocerebellar ataxia, autosomal recessive 23, 616949 (3), Autosomal recessive
<i>TDRD7</i>	611258	Cataract 36, 613887 (3), Autosomal recessive
<i>TDRD9</i>	617963	?Spermatogenic failure 30, 618110 (3), Autosomal recessive
<i>TEAD1</i>	189967	Sveinsson chorioretinal atrophy, 108985 (3), Autosomal dominant
<i>TECPR2</i>	615000	Spastic paraplegia 49, autosomal recessive, 615031 (3), Autosomal recessive
<i>TECR</i>	610057	Mental retardation, autosomal recessive 14, 614020 (3), Autosomal recessive
<i>TECRL</i>	617242	Ventricular tachycardia, catecholaminergic polymorphic, 3, 614021 (3), Autosomal recessive
<i>TECTA</i>	602574	Deafness, autosomal dominant 8/12, 601543 (3), Autosomal dominant; Deafness, autosomal recessive 21, 603629 (3), Autosomal recessive
<i>TEK</i>	600221	Glaucoma 3, primary congenital, E, 617272 (3), Autosomal dominant; Venous malformations, multiple cutaneous and mucosal, 600195 (3), Autosomal dominant
<i>TELO2</i>	611140	You-Hoover-Fong syndrome, 616954 (3), Autosomal recessive

<i>TENM3</i>	610083	Microphthalmia, syndromic 15, 615145 (3), Autosomal recessive; ?Microphthalmia, isolated, with coloboma 9, 615145 (3), Autosomal recessive
<i>TENM4</i>	610084	Essential tremor, hereditary, 5, 616736 (3), Autosomal dominant
<i>TENT5A</i>	611357	Osteogenesis imperfecta, type XVIII, 617952 (3), Autosomal recessive
<i>TERT</i>	187270	{Melanoma, cutaneous malignant, 9}, 615134 (3); {Pulmonary fibrosis and/or bone marrow failure, telomere-related, 1}, 614742 (3), Autosomal dominant; {Leukemia, acute myeloid}, 601626 (3), Autosomal dominant, Somatic mutation; {Dyskeratosis congenita, autosomal recessive 4}, 613989 (3), Autosomal recessive, Autosomal dominant; {Dyskeratosis congenita, autosomal dominant 2}, 613989 (3), Autosomal recessive, Autosomal dominant
<i>TET2</i>	612839	Myelodysplastic syndrome, somatic, 614286 (3)
<i>TET3</i>	613555	Beck-Fahrner syndrome, 618798 (3), Autosomal recessive, Autosomal dominant
<i>TEX11</i>	300311	Spermatogenic failure, X-linked, 2, 309120 (3), X-linked recessive
<i>TEX14</i>	605792	Spermatogenic failure 23, 617707 (3), Autosomal recessive
<i>TEX15</i>	605795	Spermatogenic failure 25, 617960 (3), Autosomal recessive
<i>TF</i>	190000	Atransferrinemia, 209300 (3), Autosomal recessive
<i>TFAM</i>	600438	?Mitochondrial DNA depletion syndrome 15 (hepatocerebral type), 617156 (3), Autosomal recessive
<i>TFAP2A</i>	107580	Branchiooculofacial syndrome, 113620 (3), Autosomal dominant
<i>TFAP2B</i>	601601	Char syndrome, 169100 (3), Autosomal dominant; Patent ductus arteriosus 2, 617035 (3), Autosomal dominant
<i>TFE3</i>	314310	Renal cell carcinoma, papillary, 1, 300854 (3)
<i>TFG</i>	602498	?Spastic paraplegia 57, autosomal recessive, 615658 (3), Autosomal recessive; Hereditary motor and sensory neuropathy, Okinawa type, 604484 (3), Autosomal dominant
<i>TFR2</i>	604720	Hemochromatosis, type 3, 604250 (3), Autosomal recessive
<i>TFRC</i>	190010	Immunodeficiency 46, 616740 (3), Autosomal recessive
<i>TG</i>	188450	{Autoimmune thyroid disease, susceptibility to, 3}, 608175 (3); Thyroid dyshormonogenesis 3, 274700 (3), Autosomal recessive
<i>TGDS</i>	616146	Catel-Manzke syndrome, 616145 (3), Autosomal recessive
<i>TGFB1</i>	190180	{Cystic fibrosis lung disease, modifier of}, 219700 (3), Autosomal recessive; Camurati-Engelmann disease, 131300 (3), Autosomal dominant; Inflammatory bowel disease, immunodeficiency, and encephalopathy, 618213 (3), Autosomal recessive
<i>TGFB2</i>	190220	Loeys-Dietz syndrome 4, 614816 (3), Autosomal dominant
<i>TGFB3</i>	190230	Loeys-Dietz syndrome 5, 615582 (3), Autosomal dominant; Arrhythmogenic right ventricular dysplasia 1, 107970 (3), Autosomal dominant

<i>TGFB1</i>	601692	Corneal dystrophy, lattice type IIIA, 608471 (3), Autosomal dominant; Corneal dystrophy, Groenouw type I, 121900 (3), Autosomal dominant; Corneal dystrophy, lattice type I, 122200 (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, Avellino type, 607541 (3), Autosomal dominant
<i>TGFB1</i>	190181	Loeys-Dietz syndrome 1, 609192 (3), Autosomal dominant; {Multiple self-healing squamous epithelioma, susceptibility to}, 132800 (3), Autosomal dominant
<i>TGFB2</i>	190182	Esophageal cancer, somatic, 133239 (3); Colorectal cancer, hereditary nonpolyposis, type 6, 614331 (3); Loeys-Dietz syndrome 2, 610168 (3), Autosomal dominant
<i>TGIF1</i>	602630	Holoprosencephaly 4, 142946 (3), Autosomal dominant
<i>TGM1</i>	190195	Ichthyosis, congenital, autosomal recessive 1, 242300 (3), Autosomal recessive
<i>TGM3</i>	600238	?Uncombable hair syndrome 2, 617251 (3), Autosomal recessive
<i>TGM5</i>	603805	Peeling skin syndrome 2, 609796 (3), Autosomal recessive
<i>TGM6</i>	613900	Spinocerebellar ataxia 35, 613908 (3), Autosomal dominant
<i>TH</i>	191290	Segawa syndrome, recessive, 605407 (3), Autosomal recessive
<i>THAP1</i>	609520	Dystonia 6, torsion, 602629 (3), Autosomal dominant
<i>THBD</i>	188040	Thrombophilia due to thrombomodulin defect, 614486 (3); {Hemolytic uremic syndrome, atypical, susceptibility to, 6}, 612926 (3), Autosomal dominant
<i>THBS2</i>	188061	{Lumbar disc herniation, susceptibility to}, 603932 (3)
<i>THG1L</i>	618802	Spinocerebellar ataxia, autosomal recessive 28, 618800 (3), Autosomal recessive
<i>THOC2</i>	300395	Mental retardation, X-linked 12/35, 300957 (3), X-linked recessive
<i>THOC6</i>	615403	Beaulieu-Boycott-Innes syndrome, 613680 (3), Autosomal recessive
<i>THPO</i>	600044	Thrombocythemia 1, 187950 (3), Autosomal dominant
<i>THRA</i>	190120	Hypothyroidism, congenital, nongoitrous, 6, 614450 (3), Autosomal dominant
<i>THRB</i>	190160	Thyroid hormone resistance, 188570 (3), Autosomal dominant; Thyroid hormone resistance, selective pituitary, 145650 (3), Autosomal dominant; Thyroid hormone resistance, autosomal recessive, 274300 (3), Autosomal recessive
<i>THSD1</i>	616821	?Aneurysm, intracranial berry, 12, 618734 (3)
<i>TIA1</i>	603518	Welander distal myopathy, 604454 (3), Autosomal recessive, Autosomal dominant
<i>TICAM1</i>	607601	{Encephalopathy, acute, infection-induced (herpes-specific), susceptibility to, 6}, 614850 (3), Autosomal recessive, Autosomal dominant

<i>TIMM22</i>	607251	?Combined oxidative phosphorylation deficiency 43, 618851 (3), Autosomal recessive
<i>TIMM50</i>	607381	3-methylglutaconic aciduria, type IX, 617698 (3), Autosomal recessive
<i>TIMM8A</i>	300356	Mohr-Tranebjaerg syndrome, 304700 (3), X-linked recessive
<i>TIMMDC1</i>	615534	Mitochondrial complex I deficiency, nuclear type 31, 618251 (3), Autosomal recessive
<i>TIMP3</i>	188826	Sorsby fundus dystrophy, 136900 (3), Autosomal dominant
<i>TINF2</i>	604319	Revesz syndrome, 268130 (3), Autosomal dominant; Dyskeratosis congenita, autosomal dominant 3, 613990 (3), Autosomal dominant
<i>TIRAP</i>	606252	{Bacteremia, protection against}, 614382 (3); {Malaria, protection against}, 611162 (3); {Tuberculosis, protection against}, 607948 (3)
<i>TJP2</i>	607709	Cholestasis, progressive familial intrahepatic 4, 615878 (3), Autosomal recessive; Hypercholanemia, familial, 607748 (3), Autosomal recessive
<i>TK2</i>	188250	Mitochondrial DNA depletion syndrome 2 (myopathic type), 609560 (3), Autosomal recessive; ?Progressive external ophthalmoplegia with mitochondrial DNA deletions, autosomal recessive 3, 617069 (3), Autosomal recessive
<i>TKFC</i>	615844	Triokinase and FMN cyclase deficiency syndrome, 618805 (3), Autosomal recessive
<i>TKT</i>	606781	Short stature, developmental delay, and congenital heart defects, 617044 (3), Autosomal recessive
<i>TLE6</i>	612399	Preimplantation embryonic lethality, 616814 (3), Autosomal recessive
<i>TLK2</i>	608439	Mental retardation, autosomal dominant 57, 618050 (3), Autosomal dominant
<i>TLL1</i>	606742	Atrial septal defect 6, 613087 (3), Autosomal dominant
<i>TLR1</i>	601194	{Leprosy, protection against}, 613223 (3); {Leprosy, susceptibility to, 5}, 613223 (3)
<i>TLR2</i>	603028	{Colorectal cancer, susceptibility to}, 114500 (3), Autosomal dominant, Somatic mutation; {Mycobacterium tuberculosis, susceptibility to}, 607948 (3); {Leprosy, susceptibility to}, 246300 (3), Autosomal dominant
<i>TLR3</i>	603029	{HIV1 infection, resistance to}, 609423 (3); {Encephalopathy, acute, infection-induced (herpes-specific), susceptibility to, 2}, 613002 (3), Autosomal recessive, Autosomal dominant
<i>TLR4</i>	603030/611488	-/Macular degeneration, age-related, 10, 611488 (2)
<i>TLR5</i>	603031	{Systemic lupus erythematosus, susceptibility to, 1}, 601744 (3); {Systemic lupus erythematosus, resistance to}, 601744 (3); {Meloidosis, susceptibility to}, 615557 (3); {Legionnaire disease, susceptibility to}, 608556 (3)
<i>TLR7</i>	300365	Immunodeficiency 74, COVID19-related, X-linked, 301051 (3)

<i>TM4SF20</i>	615404	{Specific language impairment 5}, 615432 (3), Autosomal dominant
<i>TMC1</i>	606706	Deafness, autosomal recessive 7, 600974 (3), Autosomal recessive; Deafness, autosomal dominant 36, 606705 (3), Autosomal dominant
<i>TMC6</i>	605828	Epidermodysplasia verruciformis, 226400 (3), Autosomal recessive
<i>TMC8</i>	605829	Epidermodysplasia verruciformis 2, 618231 (3), Autosomal recessive
<i>TMCO1</i>	614123	Craniofacial dysmorphism, skeletal anomalies, and mental retardation syndrome, 213980 (3), Autosomal recessive
<i>TMEM106B</i>	613413	Leukodystrophy, hypomyelinating, 16, 617964 (3), Autosomal dominant
<i>TMEM107</i>	616183	Orofaciodigital syndrome XVI, 617563 (3), Autosomal recessive; Meckel syndrome 13, 617562 (3), Autosomal recessive; ?Joubert syndrome 29, 617562 (3), Autosomal recessive
<i>TMEM126A</i>	612988	Optic atrophy 7, 612989 (3), Autosomal recessive
<i>TMEM126B</i>	615533	Mitochondrial complex I deficiency, nuclear type 29, 618250 (3), Autosomal recessive
<i>TMEM127</i>	613403	{Pheochromocytoma, susceptibility to}, 171300 (3), Autosomal dominant
<i>TMEM132E</i>	616178	?Deafness, autosomal recessive 99, 618481 (3), Autosomal recessive
<i>TMEM138</i>	614459	Joubert syndrome 16, 614465 (3), Autosomal recessive
<i>TMEM165</i>	614726	Congenital disorder of glycosylation, type IIk, 614727 (3), Autosomal recessive
<i>TMEM199</i>	616815	Congenital disorder of glycosylation, type IIp, 616829 (3), Autosomal recessive
<i>TMEM216</i>	613277	Meckel syndrome 2, 603194 (3), Autosomal recessive; Joubert syndrome 2, 608091 (3), Autosomal recessive
<i>TMEM231</i>	614949	Meckel syndrome 11, 615397 (3), Autosomal recessive; Joubert syndrome 20, 614970 (3), Autosomal recessive
<i>TMEM237</i>	614423	Joubert syndrome 14, 614424 (3), Autosomal recessive
<i>TMEM240</i>	616101	Spinocerebellar ataxia 21, 607454 (3), Autosomal dominant
<i>TMEM260</i>	617449	Structural heart defects and renal anomalies syndrome, 617478 (3), Autosomal recessive
<i>TMEM38B</i>	611236	Osteogenesis imperfecta, type XIV, 615066 (3)
<i>TMEM43</i>	612048	Emery-Dreifuss muscular dystrophy 7, AD, 614302 (3), Autosomal dominant; Arrhythmogenic right ventricular dysplasia 5, 604400 (3), Autosomal dominant
<i>TMEM63A</i>	618685	Leukodystrophy, hypomyelinating, 19, transient infantile, 618688 (3), Autosomal dominant

<i>TMEM67</i>	609884	Meckel syndrome 3, 607361 (3), Autosomal recessive; ?RHYNS syndrome, 602152 (3), Autosomal recessive; Nephronophthisis 11, 613550 (3), Autosomal recessive; {Bardet-Biedl syndrome 14, modifier of}, 615991 (3), Autosomal recessive; COACH syndrome, 216360 (3), Autosomal recessive; Joubert syndrome 6, 610688 (3), Autosomal recessive
<i>TMEM70</i>	612418	Mitochondrial complex V (ATP synthase) deficiency, nuclear type 2, 614052 (3), Autosomal recessive
<i>TMEM94</i>	618163	Intellectual developmental disorder with cardiac defects and dysmorphic facies, 618316 (3), Autosomal recessive
<i>TMEM98</i>	615949	Nanophthalmos 4, 615972 (3), Autosomal dominant
<i>TMIE</i>	607237	Deafness, autosomal recessive 6, 600971 (3), Autosomal recessive
<i>TMLHE</i>	300777	{Autism, susceptibility to, X-linked 6}, 300872 (3), X-linked recessive
<i>TMPRSS15</i>	606635	Enterokinase deficiency, 226200 (3), Autosomal recessive
<i>TMPRSS3</i>	605511	Deafness, autosomal recessive 8/10, 601072 (3), Autosomal recessive
<i>TMPRSS6</i>	609862	Iron-refractory iron deficiency anemia, 206200 (3), Autosomal recessive
<i>TMTC3</i>	617218	Lissencephaly 8, 617255 (3), Autosomal recessive
<i>TMX2</i>	616715	Neurodevelopmental disorder with microcephaly, cortical malformations, and spasticity, 618730 (3), Autosomal recessive
<i>TNC</i>	187380	Deafness, autosomal dominant 56, 615629 (3), Autosomal dominant
<i>TNF</i>	191160	{Asthma, susceptibility to}, 600807 (3), Autosomal dominant; {Migraine without aura, susceptibility to}, 157300 (3), Autosomal dominant; {Septic shock, susceptibility to} (3); {Dementia, vascular, susceptibility to} (3); {Malaria, cerebral, susceptibility to}, 611162 (3)
<i>TNFAIP3</i>	191163	Autoinflammatory syndrome, familial, Behcet-like, 616744 (3), Autosomal dominant
<i>TNFRSF10B</i>	603612	Squamous cell carcinoma, head and neck, 275355 (3), Autosomal recessive
<i>TNFRSF11A</i>	603499	Osteolysis, familial expansile, 174810 (3), Autosomal dominant; {Paget disease of bone 2, early-onset}, 602080 (3), Autosomal dominant; Osteopetrosis, autosomal recessive 7, 612301 (3), Autosomal recessive
<i>TNFRSF11B</i>	602643	Paget disease of bone 5, juvenile-onset, 239000 (3), Autosomal recessive
<i>TNFRSF13B</i>	604907	Immunoglobulin A deficiency 2, 609529 (3); Immunodeficiency, common variable, 2, 240500 (3), Autosomal recessive, Autosomal dominant
<i>TNFRSF13C</i>	606269	Immunodeficiency, common variable, 4, 613494 (3), Autosomal recessive
<i>TNFRSF1A</i>	191190	{Multiple sclerosis, susceptibility to, 5}, 614810 (3); Periodic fever, familial, 142680 (3), Autosomal dominant

<i>TNFRSF4</i>	600315	?Immunodeficiency 16, 615593 (3), Autosomal recessive
<i>TNFSF11</i>	602642	Osteopetrosis, autosomal recessive 2, 259710 (3), Autosomal recessive
<i>TNFSF4</i>	603594	{Myocardial infarction, susceptibility to}, 608446 (3)
<i>TNIK</i>	610005	Mental retardation, autosomal recessive 54, 617028 (3), Autosomal recessive
<i>TNNC1</i>	191040	Cardiomyopathy, hypertrophic, 13, 613243 (3), Autosomal dominant; Cardiomyopathy, dilated, 1Z, 611879 (3)
<i>TNNI2</i>	191043	Arthrogryposis, distal, type 2B1, 601680 (3), Autosomal dominant
<i>TNNI3</i>	191044	Cardiomyopathy, hypertrophic, 7, 613690 (3), Autosomal dominant; ?Cardiomyopathy, dilated, 2A, 611880 (3), Autosomal recessive; Cardiomyopathy, dilated, 1FF, 613286 (3); Cardiomyopathy, familial restrictive, 1, 115210 (3), Autosomal dominant
<i>TNNI3K</i>	613932	Cardiac conduction disease with or without dilated cardiomyopathy, 616117 (3), Autosomal dominant
<i>TNNT1</i>	191041	Nemaline myopathy 5, Amish type, 605355 (3), Autosomal recessive
<i>TNNT2</i>	191045	Cardiomyopathy, familial restrictive, 3, 612422 (3), Autosomal dominant; Cardiomyopathy, hypertrophic, 2, 115195 (3), Autosomal dominant; Left ventricular noncompaction 6, 601494 (3), Autosomal dominant; Cardiomyopathy, dilated, 1D, 601494 (3), Autosomal dominant
<i>TNNT3</i>	600692	Arthrogryposis, distal, type 2B2, 618435 (3), Autosomal dominant
<i>TNPO3</i>	610032	Muscular dystrophy, limb-girdle, autosomal dominant 2, 608423 (3), Autosomal dominant
<i>TNRC6A</i>	610739	?Epilepsy, familial adult myoclonic, 6, 618074 (3)
<i>TNXB</i>	600985	Ehlers-Danlos syndrome, classic-like, 1, 606408 (3), Autosomal recessive; Vesicoureteral reflux 8, 615963 (3), Autosomal dominant
<i>TOE1</i>	613931	Pontocerebellar hypoplasia, type 7, 614969 (3), Autosomal recessive
<i>TONSL</i>	604546	Spondyloepimetaphyseal dysplasia, sponastrime type, 271510 (3), Autosomal recessive
<i>TOP1</i>	126420	DNA topoisomerase I, camptothecin-resistant (3)
<i>TOP2A</i>	126430	DNA topoisomerase II, resistance to inhibition of, by amsacrine (3)
<i>TOP3A</i>	601243	Microcephaly, growth restriction, and increased sister chromatid exchange 2, 618097 (3), Autosomal recessive; ?Progressive external ophthalmoplegia with mitochondrial DNA deletions, autosomal recessive 5, 618098 (3), Autosomal recessive
<i>TOPORS</i>	609507	Retinitis pigmentosa 31, 609923 (3)

<i>TOR1A</i>	605204	Dystonia-1, torsion, 128100 (3), Autosomal dominant; Arthrogryposis multiplex congenita 5, 618947 (3); {Dystonia-1, modifier of} (3)
<i>TOR1AIP1</i>	614512	?Muscular dystrophy, autosomal recessive, with rigid spine and distal joint contractures, 617072 (3), Autosomal recessive {Adrenocortical carcinoma, pediatric}, 202300 (3), Autosomal dominant; {Glioma susceptibility 1}, 137800 (3), Autosomal dominant, Somatic mutation; {Basal cell carcinoma 7}, 614740 (3), Autosomal dominant; Bone marrow failure syndrome 5, 618165 (3), Autosomal dominant; {Colorectal cancer}, 114500 (3), Autosomal dominant, Somatic mutation; Nasopharyngeal carcinoma, somatic, 607107 (3); Breast cancer, somatic, 114480 (3); {Osteosarcoma}, 259500 (3), Somatic mutation; {Choroid plexus papilloma}, 260500 (3), Autosomal dominant; Li-Fraumeni syndrome, 151623 (3), Autosomal dominant; Hepatocellular carcinoma, somatic, 114550 (3); Pancreatic cancer, somatic, 260350 (3)
<i>TP53</i>	191170	
<i>TP53RK</i>	608679	Galloway-Mowat syndrome 4, 617730 (3), Autosomal recessive Limb-mammary syndrome, 603543 (3), Autosomal dominant; Orofacial cleft 8, 618149 (3); Split-hand/foot malformation 4, 605289 (3), Autosomal dominant; Hay-Wells syndrome, 106260 (3), Autosomal dominant; Ectrodactyly, ectodermal dysplasia, and cleft lip/palate syndrome 3, 604292 (3), Autosomal dominant; Rapp-Hodgkin syndrome, 129400 (3), Autosomal dominant; ADULT syndrome, 103285 (3), Autosomal dominant
<i>TP63</i>	603273	[Skin/hair/eye pigmentation 10, blond/brown hair], 612267 (3)
<i>TPCN2</i>	612163	{Attention deficit-hyperactivity disorder, susceptibility to, 7}, 613003 (3); {Unipolar depression, susceptibility to}, 608516 (3)
<i>TPH2</i>	607478	
<i>TPI1</i>	190450	Hemolytic anemia due to triosephosphate isomerase deficiency, 615512 (3), Autosomal recessive
<i>TPK1</i>	606370	Thiamine metabolism dysfunction syndrome 5 (episodic encephalopathy type), 614458 (3), Autosomal recessive
<i>TPM1</i>	191010	Left ventricular noncompaction 9, 611878 (3), Autosomal dominant; Cardiomyopathy, hypertrophic, 3, 115196 (3), Autosomal dominant; Cardiomyopathy, dilated, 1Y, 611878 (3), Autosomal dominant
<i>TPM2</i>	190990	Nemaline myopathy 4, autosomal dominant, 609285 (3), Autosomal dominant; Arthrogryposis, distal, type 2B4, 108120 (3), Autosomal dominant; Arthrogryposis, distal, type 1A, 108120 (3), Autosomal dominant; CAP myopathy 2, 609285 (3), Autosomal dominant
<i>TPM3</i>	191030	CAP myopathy 1, 609284 (3), Autosomal recessive, Autosomal dominant; Nemaline myopathy 1, autosomal dominant or recessive, 609284 (3), Autosomal recessive, Autosomal dominant; Myopathy, congenital, with fiber-type disproportion, 255310 (3), Autosomal recessive, Autosomal dominant

<i>TPMT</i>	187680	{Thiopurines, poor metabolism of, 1}, 610460 (3), Autosomal recessive
<i>TPO</i>	606765	Thyroid dysmorphogenesis 2A, 274500 (3), Autosomal recessive
<i>TPP1</i>	607998	Spinocerebellar ataxia, autosomal recessive 7, 609270 (3), Autosomal recessive; Ceroid lipofuscinosis, neuronal, 2, 204500 (3), Autosomal recessive
<i>TPRKB</i>	608680	Galloway-Mowat syndrome 5, 617731 (3), Autosomal recessive
<i>TPRN</i>	613354	Deafness, autosomal recessive 79, 613307 (3), Autosomal recessive
<i>TRAC</i>	186880	Immunodeficiency 7, TCR-alpha/beta deficient, 615387 (3), Autosomal recessive
<i>TRAF3</i>	601896	{?Encephalopathy, acute, infection-induced (herpes-specific), susceptibility to, 5}, 614849 (3)
<i>TRAF3IP1</i>	607380	Senior-Loken syndrome 9, 616629 (3), Autosomal recessive
<i>TRAF3IP2</i>	607043	?Candidiasis, familial, 8, 615527 (3), Autosomal recessive; {Psoriasis susceptibility 13}, 614070 (3)
<i>TRAF7</i>	606692	Cardiac, facial, and digital anomalies with developmental delay, 618164 (3), Autosomal dominant
<i>TRAIP</i>	605958	Seckel syndrome 9, 616777 (3), Autosomal recessive
<i>TRAK1</i>	608112	Epileptic encephalopathy, early infantile, 68, 618201 (3), Autosomal recessive
<i>TRAPPC11</i>	614138	Muscular dystrophy, limb-girdle, autosomal recessive 18, 615356 (3), Autosomal recessive
<i>TRAPPC12</i>	614139	Encephalopathy, progressive, early-onset, with brain atrophy and spasticity, 617669 (3), Autosomal recessive
<i>TRAPPC2</i>	300202	Spondyloepiphyseal dysplasia tarda, 313400 (3), X-linked recessive
<i>TRAPPC2L</i>	610970	Encephalopathy, progressive, early-onset, with episodic rhabdomyolysis, 618331 (3), Autosomal recessive
<i>TRAPPC4</i>	610971	Neurodevelopmental disorder with epilepsy, spasticity, and brain atrophy, 618741 (3), Autosomal recessive
<i>TRAPPC6B</i>	610397	Neurodevelopmental disorder with microcephaly, epilepsy, and brain atrophy, 617862 (3), Autosomal recessive
<i>TRAPPC9</i>	611966	Mental retardation, autosomal recessive 13, 613192 (3), Autosomal recessive
<i>TRDN</i>	603283	Ventricular tachycardia, catecholaminergic polymorphic, 5, with or without muscle weakness, 615441 (3), Autosomal recessive
<i>TREH</i>	275360	Trehalase deficiency, 612119 (3), Autosomal recessive
<i>TREM2</i>	605086	Polycystic lipomembranous osteodysplasia with sclerosing leukoencephalopathy 2, 618193 (3), Autosomal dominant

<i>TREX1</i>	606609	{Systemic lupus erythematosus, susceptibility to}, 152700 (3), Autosomal dominant; Vasculopathy, retinal, with cerebral leukodystrophy, 192315 (3), Autosomal dominant; Aicardi-Goutieres syndrome 1, dominant and recessive, 225750 (3), Autosomal recessive, Autosomal dominant; Chilblain lupus, 610448 (3), Autosomal dominant
<i>TRH</i>	613879	Thyrotropin-releasing hormone deficiency, 275120 (1), Autosomal recessive
<i>TRHR</i>	188545	Hypothyroidism, congenital, nongoitrous, 7, 618573 (3), Autosomal recessive
<i>TRIM2</i>	614141	Charcot-Marie-Tooth disease, type 2R, 615490 (3), Autosomal recessive
<i>TRIM32</i>	602290	?Bardet-Biedl syndrome 11, 615988 (3), Autosomal recessive; Muscular dystrophy, limb-girdle, autosomal recessive 8, 254110 (3), Autosomal recessive
<i>TRIM36</i>	609317	?Anencephaly, 206500 (3), Autosomal recessive
<i>TRIM37</i>	605073	Mulibrey nanism, 253250 (3), Autosomal recessive
<i>TRIM44</i>	612298	?Aniridia 3, 617142 (3), Autosomal dominant
<i>TRIM71</i>	618570	Hydrocephalus, congenital communicating, 1, 618667 (3), Autosomal dominant
<i>TRIO</i>	601893	Intellectual developmental disorder, autosomal dominant 44, with microcephaly, 617061 (3), Autosomal dominant; Intellectual developmental disorder, autosomal dominant 63, with macrocephaly, 618825 (3), Autosomal dominant
<i>TRIOBP</i>	609761	Deafness, autosomal recessive 28, 609823 (3), Autosomal recessive
<i>TRIP11</i>	604505	Osteochondrodysplasia, 184260 (3), Autosomal recessive; Achondrogenesis, type IA, 200600 (3), Autosomal recessive
<i>TRIP12</i>	604506	Mental retardation, autosomal dominant 49, 617752 (3), Autosomal dominant
<i>TRIP13</i>	604507	Mosaic variegated aneuploidy syndrome 3, 617598 (3), Autosomal recessive
<i>TRIP4</i>	604501	Spinal muscular atrophy with congenital bone fractures 1, 616866 (3), Autosomal recessive; ?Muscular dystrophy, congenital, Davignon-Chauveau type, 617066 (3), Autosomal recessive
<i>TRIT1</i>	617840	Combined oxidative phosphorylation deficiency 35, 617873 (3), Autosomal recessive
<i>TRMT1</i>	611669	Mental retardation, autosomal recessive 68, 618302 (3), Autosomal recessive
<i>TRMT10A</i>	616013	Microcephaly, short stature, and impaired glucose metabolism 1, 616033 (3), Autosomal recessive
<i>TRMT10C</i>	615423	Combined oxidative phosphorylation deficiency 30, 616974 (3), Autosomal recessive
<i>TRMT5</i>	611023	Combined oxidative phosphorylation deficiency 26, 616539 (3), Autosomal recessive

<i>TRMU</i>	610230	Liver failure, transient infantile, 613070 (3), Autosomal recessive; {Deafness, mitochondrial, modifier of}, 580000 (3), Mitochondrial
<i>TRNT1</i>	612907	Sideroblastic anemia with B-cell immunodeficiency, periodic fevers, and developmental delay, 616084 (3), Autosomal recessive; Retinitis pigmentosa and erythrocytic microcytosis, 616959 (3), Autosomal recessive
<i>TRPA1</i>	604775	?Episodic pain syndrome, familial, 1, 615040 (3), Autosomal dominant
<i>TRPC3</i>	602345	?Spinocerebellar ataxia 41, 616410 (3), Autosomal dominant
<i>TRPC6</i>	603652	Glomerulosclerosis, focal segmental, 2, 603965 (3), Autosomal dominant
<i>TRPM1</i>	603576	Night blindness, congenital stationary (complete), 1C, autosomal recessive, 613216 (3)
<i>TRPM4</i>	606936	Erythrokeratoderma variabilis et progressiva 6, 618531 (3), Autosomal dominant; Progressive familial heart block, type IB, 604559 (3), Autosomal dominant
<i>TRPM6</i>	607009	Hypomagnesemia 1, intestinal, 602014 (3), Autosomal recessive
<i>TRPM7</i>	605692	{Amyotrophic lateral sclerosis-parkinsonism/dementia complex, susceptibility to}, 105500 (3), Autosomal dominant
<i>TRPS1</i>	604386	Trichorhinophalangeal syndrome, type I, 190350 (3), Autosomal dominant; Trichorhinophalangeal syndrome, type III, 190351 (3), Autosomal dominant
<i>TRPV3</i>	607066	?Palmoplantar keratoderma, nonepidermolytic, focal 2, 616400 (3), Autosomal dominant; Olmsted syndrome, 614594 (3), Autosomal dominant
<i>TRPV4</i>	605427	Spondylometaphyseal dysplasia, Kozlowski type, 184252 (3), Autosomal dominant; Parastremmatic dwarfism, 168400 (3), Autosomal dominant; SED, Maroteaux type, 184095 (3), Autosomal dominant; Neuronopathy, distal hereditary motor, type VIII, 600175 (3), Autosomal dominant; [Sodium serum level QTL 1], 613508 (3); Scapuloperoneal spinal muscular atrophy, 181405 (3), Autosomal dominant; Metatropic dysplasia, 156530 (3), Autosomal dominant; Digital arthropathy-brachydactyly, familial, 606835 (3), Autosomal dominant; Hereditary motor and sensory neuropathy, type IIc, 606071 (3), Autosomal dominant; Brachyolmia type 3, 113500 (3), Autosomal dominant; ?Avascular necrosis of femoral head, primary, 2, 617383 (3), Autosomal dominant
<i>TRPV6</i>	606680	Hyperparathyroidism, transient neonatal, 618188 (3), Autosomal recessive
<i>TRRAP</i>	603015	Developmental delay with or without dysmorphic facies and autism, 618454 (3), Autosomal dominant; ?Deafness, autosomal dominant 75, 618778 (3), Autosomal dominant
<i>TSC1</i>	605284	Tuberous sclerosis-1, 191100 (3), Autosomal dominant; Focal cortical dysplasia, type II, somatic, 607341 (3); Lymphangioliomyomatosis, 606690 (3)

<i>TSC2</i>	191092	Tuberous sclerosis-2, 613254 (3), Autosomal dominant; ?Focal cortical dysplasia, type II, somatic, 607341 (3); Lymphangioleiomyomatosis, somatic, 606690 (3)
<i>TSEN15</i>	608756	Pontocerebellar hypoplasia, type 2F, 617026 (3), Autosomal recessive
<i>TSEN2</i>	608753	Pontocerebellar hypoplasia type 2B, 612389 (3), Autosomal recessive
<i>TSEN34</i>	608754	?Pontocerebellar hypoplasia type 2C, 612390 (3), Autosomal recessive
<i>TSEN54</i>	608755	Pontocerebellar hypoplasia type 4, 225753 (3), Autosomal recessive; Pontocerebellar hypoplasia type 2A, 277470 (3), Autosomal recessive; ?Pontocerebellar hypoplasia type 5, 610204 (3), Autosomal recessive
<i>TSFM</i>	604723	Combined oxidative phosphorylation deficiency 3, 610505 (3), Autosomal recessive
<i>TSGA10</i>	607166	?Spermatogenic failure 26, 617961 (3), Autosomal recessive
<i>TSHB</i>	188540	Hypothyroidism, congenital, nongoitrous 4, 275100 (3), Autosomal recessive
<i>TSHR</i>	603372	Thyroid adenoma, hyperfunctioning, somatic (3); Thyroid carcinoma with thyrotoxicosis (3); Hyperthyroidism, nonautoimmune, 609152 (3), Autosomal dominant; Hypothyroidism, congenital, nongoitrous, 1, 275200 (3), Autosomal recessive; Hyperthyroidism, familial gestational, 603373 (3)
<i>TSHZ1</i>	614427	Aural atresia, congenital, 607842 (3), Autosomal dominant
<i>TSPAN12</i>	613138	Exudative vitreoretinopathy 5, 613310 (3), Autosomal dominant
<i>TSPAN7</i>	300096	Mental retardation, X-linked 58, 300210 (3), X-linked recessive
<i>TSPEAR</i>	612920	Ectodermal dysplasia 14, hair/tooth type with or without hypohidrosis, 618180 (3), Autosomal recessive; ?Deafness, autosomal recessive 98, 614861 (3), Autosomal recessive
<i>TSPYL1</i>	604714	Sudden infant death with dysgenesis of the testes syndrome, 608800 (3), Autosomal recessive
<i>TSR2</i>	300945	?Diamond-Blackfan anemia 14 with mandibulofacial dysostosis, 300946 (3), X-linked recessive
<i>TTBK2</i>	611695	Spinocerebellar ataxia 11, 604432 (3), Autosomal dominant
<i>TTC12</i>	610732	Ciliary dyskinesia, primary, 45, 618801 (3), Autosomal recessive
<i>TTC19</i>	613814	Mitochondrial complex III deficiency, nuclear type 2, 615157 (3), Autosomal recessive
<i>TTC21A</i>	611430	Spermatogenic failure 37, 618429 (3), Autosomal recessive
<i>TTC21B</i>	612014	Nephronophthisis 12, 613820 (3), Autosomal recessive, Autosomal dominant; Short-rib thoracic dysplasia 4 with or without polydactyly, 613819 (3), Autosomal recessive
<i>TTC25</i>	617095	Ciliary dyskinesia, primary, 35, 617092 (3), Autosomal recessive
<i>TTC29</i>	618735	Spermatogenic failure 42, 618745 (3), Autosomal recessive
<i>TTC37</i>	614589	Trichohepatoenteric syndrome 1, 222470 (3), Autosomal recessive

<i>TTC7A</i>	609332	Gastrointestinal defects and immunodeficiency syndrome, 243150 (3), Autosomal recessive
<i>TTC8</i>	608132	Bardet-Biedl syndrome 8, 615985 (3), Autosomal recessive; ?Retinitis pigmentosa 51, 613464 (3), Autosomal recessive
<i>TTI2</i>	614426	Mental retardation, autosomal recessive 39, 615541 (3), Autosomal recessive
<i>TTLL5</i>	612268	Cone-rod dystrophy 19, 615860 (3), Autosomal recessive
<i>TTN</i>	188840	Myopathy, myofibrillar, 9, with early respiratory failure, 603689 (3), Autosomal dominant; Cardiomyopathy, familial hypertrophic, 9, 613765 (3), Autosomal dominant; Muscular dystrophy, limb-girdle, autosomal recessive 10, 608807 (3), Autosomal recessive; Cardiomyopathy, dilated, 1G, 604145 (3); Tibial muscular dystrophy, tardive, 600334 (3), Autosomal dominant; Salih myopathy, 611705 (3), Autosomal recessive
<i>TTPA</i>	600415	Ataxia with isolated vitamin E deficiency, 277460 (3), Autosomal recessive
<i>TTR</i>	176300	Amyloidosis, hereditary, transthyretin-related, 105210 (3), Autosomal dominant; [Dystransthyretinemic hyperthyroxinemia], 145680 (3), Autosomal dominant; Carpal tunnel syndrome, familial, 115430 (3), Autosomal dominant
<i>TUB</i>	601197	?Retinal dystrophy and obesity, 616188 (3), Autosomal recessive
<i>TUBA1A</i>	602529	Lissencephaly 3, 611603 (3), Autosomal dominant
<i>TUBA3D</i>	617878	Keratoconus 9, 617928 (3), Autosomal dominant
<i>TUBA4A</i>	191110	Amyotrophic lateral sclerosis 22 with or without frontotemporal dementia, 616208 (3), Autosomal dominant
<i>TUBA8</i>	605742	Cortical dysplasia, complex, with other brain malformations 8, 613180 (3), Autosomal recessive
<i>TUBB</i>	191130	Symmetric circumferential skin creases, congenital, 1, 156610 (3), Autosomal dominant; Cortical dysplasia, complex, with other brain malformations 6, 615771 (3), Autosomal dominant
<i>TUBB1</i>	612901	Macrothrombocytopenia, autosomal dominant, TUBB1-related, 613112 (3), Autosomal dominant
<i>TUBB2A</i>	615101	Cortical dysplasia, complex, with other brain malformations 5, 615763 (3), Autosomal dominant
<i>TUBB2B</i>	612850	Cortical dysplasia, complex, with other brain malformations 7, 610031 (3), Autosomal dominant
<i>TUBB3</i>	602661	Fibrosis of extraocular muscles, congenital, 3A, 600638 (3), Autosomal dominant; Cortical dysplasia, complex, with other brain malformations 1, 614039 (3), Autosomal dominant
<i>TUBB4A</i>	602662	Leukodystrophy, hypomyelinating, 6, 612438 (3), Autosomal dominant; Dystonia 4, torsion, autosomal dominant, 128101 (3), Autosomal dominant
<i>TUBB4B</i>	602660	Leber congenital amaurosis with early-onset deafness, 617879 (3), Autosomal dominant

<i>TUBB6</i>	615103	?Facial palsy, congenitla, with ptosis and velopharyngeal dysfunction, 617732 (3), Autosomal dominant
<i>TUBB8</i>	616768	Oocyte maturation defect 2, 616780 (3), Autosomal recessive, Autosomal dominant
<i>TUBG1</i>	191135	Cortical dysplasia, complex, with other brain malformations 4, 615412 (3), Autosomal dominant
<i>TUBGCP2</i>	617817	Pachygyria, microcephaly, developmental delay, and dysmorphic facies, with or without seizures, 618737 (3), Autosomal recessive
<i>TUBGCP4</i>	609610	Microcephaly and chorioretinopathy, autosomal recessive, 3, 616335 (3), Autosomal recessive
<i>TUBGCP6</i>	610053	Microcephaly and chorioretinopathy, autosomal recessive, 1, 251270 (3), Autosomal recessive
<i>TUFM</i>	602389	Combined oxidative phosphorylation deficiency 4, 610678 (3), Autosomal recessive
<i>TULP1</i>	602280	Retinitis pigmentosa 14, 600132 (3), Autosomal recessive; Leber congenital amaurosis 15, 613843 (3), Autosomal recessive
<i>TUSC3</i>	601385	Mental retardation, autosomal recessive 7, 611093 (3), Autosomal recessive
<i>TWIST1</i>	601622	Robinow-Sorauf syndrome, 180750 (3), Autosomal dominant; Craniosynostosis 1, 123100 (3), Autosomal dominant; Sweeney-Cox syndrome, 617746 (3), Autosomal dominant; Saethre-Chotzen syndrome with or without eyelid anomalies, 101400 (3), Autosomal dominant
<i>TWIST2</i>	607556	Focal facial dermal dysplasia 3, Setleis type, 227260 (3), Autosomal recessive; Ablepharon-macrostomia syndrome, 200110 (3), Autosomal dominant; Barber-Say syndrome, 209885 (3), Autosomal dominant
<i>TWINK</i>	606075	Mitochondrial DNA depletion syndrome 7 (hepatocerebral type), 271245 (3), Autosomal recessive; Progressive external ophthalmoplegia with mitochondrial DNA deletions, autosomal dominant 3, 609286 (3), Autosomal dominant; Perrault syndrome 5, 616138 (3), Autosomal recessive
<i>TXN2</i>	609063	?Combined oxidative phosphorylation deficiency 29, 616811 (3), Autosomal recessive
<i>TXNL4A</i>	611595	Burn-McKeown syndrome, 608572 (3), Autosomal recessive
<i>TXNRD2</i>	606448	?Glucocorticoid deficiency 5, 617825 (3), Autosomal recessive
<i>TYK2</i>	176941	Immunodeficiency 35, 611521 (3), Autosomal recessive
<i>TYMP</i>	131222	Mitochondrial DNA depletion syndrome 1 (MNGIE type), 603041 (3), Autosomal recessive

<i>TYR</i>	606933	Waardenburg syndrome/albinism, digenic, 103470 (3); Albinism, oculocutaneous, type IB, 606952 (3), Autosomal recessive; [Skin/hair/eye pigmentation 3, blue/green eyes], 601800 (3), Autosomal dominant; {Melanoma, cutaneous malignant, susceptibility to, 8}, 601800 (3), Autosomal dominant; Albinism, oculocutaneous, type IA, 203100 (3), Autosomal recessive; [Skin/hair/eye pigmentation 3, light/dark/freckling skin], 601800 (3), Autosomal dominant
<i>TYROBP</i>	604142	Polycystic lipomembranous osteodysplasia with sclerosing leukoencephalopathy 1, 221770 (3), Autosomal recessive
<i>TYRP1</i>	115501	[Skin/hair/eye pigmentation, variation in, 11 (Melanesian blond hair)], 612271 (3); Albinism, oculocutaneous, type III, 203290 (3), Autosomal recessive
<i>UBA1</i>	314370	Spinal muscular atrophy, X-linked 2, infantile, 301830 (3), X-linked recessive
<i>UBA5</i>	610552	?Spinocerebellar ataxia, autosomal recessive 24, 617133 (3), Autosomal recessive; Epileptic encephalopathy, early infantile, 44, 617132 (3), Autosomal recessive
<i>UBAP1</i>	609787	Spastic paraplegia 80, autosomal dominant, 618418 (3), Autosomal dominant
<i>UBE2A</i>	312180	Mental retardation, X-linked syndromic, Nascimento-type, 300860 (3), X-linked recessive
<i>UBE2T</i>	610538	Fanconi anemia, complementation group T, 616435 (3), Autosomal recessive
<i>UBE3A</i>	601623	Angelman syndrome, 105830 (3), Autosomal dominant
<i>UBE3B</i>	608047	Kaufman oculocerebrofacial syndrome, 244450 (3), Autosomal recessive
<i>UBIAD1</i>	611632	Corneal dystrophy, Schnyder type, 121800 (3), Autosomal dominant
<i>UBQLN2</i>	300264	Amyotrophic lateral sclerosis 15, with or without frontotemporal dementia, 300857 (3), X-linked dominant
<i>UBR1</i>	605981	Johanson-Blizzard syndrome, 243800 (3), Autosomal recessive
<i>UBTF</i>	600673	Neurodegeneration, childhood-onset, with brain atrophy, 617672 (3), Autosomal dominant
<i>UCHL1</i>	191342	Spastic paraplegia 79, autosomal recessive, 615491 (3), Autosomal recessive; {?Parkinson disease 5, susceptibility to}, 613643 (3), Autosomal dominant
<i>UCP2</i>	601693	{Obesity, susceptibility to, BMIQ4}, 607447 (3)
<i>UCP3</i>	602044	{Obesity, severe, and type II diabetes}, 601665 (3), Autosomal recessive, Autosomal dominant, Multifactorial
<i>UFC1</i>	610554	Neurodevelopmental disorder with spasticity and poor growth, 618076 (3), Autosomal recessive
<i>UFM1</i>	610553	Leukodystrophy, hypomyelinating, 14, 617899 (3), Autosomal recessive

<i>UFSP2</i>	611482	?Spondyloepimetaphyseal dysplasia, Di Rocco type, 617974 (3), Autosomal dominant; ?Hip dysplasia, Beukes type, 142669 (3), Autosomal dominant
<i>UGDH</i>	603370	Epileptic encephalopathy, early infantile, 84, 618792 (3), Autosomal recessive
<i>UGP2</i>	191760	Epileptic encephalopathy, early infantile, 83, 618744 (3), Autosomal recessive
<i>UGT1A1</i>	191740	[Gilbert syndrome], 143500 (3), Autosomal recessive; Hyperbilirubinemia, familial transient neonatal, 237900 (3), Autosomal recessive; Crigler-Najjar syndrome, type I, 218800 (3), Autosomal recessive; Crigler-Najjar syndrome, type II, 606785 (3), Autosomal recessive; [Bilirubin, serum level of, QTL1], 601816 (3)
<i>UGT2B17</i>	601903	{Bone mineral density QTL 12, osteoporosis}, 612560 (3)
<i>UMOD</i>	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)
<i>UMPS</i>	613891	Orotic aciduria, 258900 (3), Autosomal recessive
<i>UNC119</i>	604011	?Immunodeficiency 13, 615518 (3), Autosomal dominant; ?Cone-rod dystrophy (3)
<i>UNC13D</i>	608897	Hemophagocytic lymphohistiocytosis, familial, 3, 608898 (3), Autosomal recessive
<i>UNC45B</i>	611220	?Cataract 43, 616279 (3), Autosomal dominant
<i>UNC80</i>	612636	Hypotonia, infantile, with psychomotor retardation and characteristic facies 2, 616801 (3), Autosomal recessive
<i>UNC93B1</i>	608204	{Encephalopathy, acute, infection-induced (herpes-specific), susceptibility to, 1}, 610551 (3)
<i>UNG</i>	191525	Immunodeficiency with hyper IgM, type 5, 608106 (3), Autosomal recessive
<i>UPB1</i>	606673	Beta-ureidopropionase deficiency, 613161 (3), Autosomal recessive
<i>UPF3B</i>	300298	Mental retardation, X-linked, syndromic 14, 300676 (3), X-linked recessive
<i>UQCC2</i>	614461	Mitochondrial complex III deficiency, nuclear type 7, 615824 (3), Autosomal recessive
<i>UQCC3</i>	616097	?Mitochondrial complex III deficiency, nuclear type 9, 616111 (3), Autosomal recessive
<i>UQCRB</i>	191330	Mitochondrial complex III deficiency, nuclear type 3, 615158 (3), Autosomal recessive
<i>UQCRC2</i>	191329	Mitochondrial complex III deficiency, nuclear type 5, 615160 (3), Autosomal recessive
<i>UQCRFS1</i>	191327	Mitochondrial complex III deficiency, nuclear type 10, 618775 (3), Autosomal recessive
<i>UQCRQ</i>	612080	Mitochondrial complex III deficiency, nuclear type 4, 615159 (3), Autosomal recessive

<i>UROC1</i>	613012	?Urocanase deficiency, 276880 (3), Autosomal recessive
<i>UROD</i>	613521	Porphyria, hepatoerythropoietic, 176100 (3), Autosomal recessive, Autosomal dominant; Porphyria cutanea tarda, 176100 (3), Autosomal recessive, Autosomal dominant
<i>UROS</i>	606938	Porphyria, congenital erythropoietic, 263700 (3), Autosomal recessive
<i>USB1</i>	613276	Poikiloderma with neutropenia, 604173 (3), Autosomal recessive
<i>USF1</i>	191523	{Hyperlipidemia, familial combined, susceptibility to}, 602491 (3)
<i>USH1C</i>	605242	Deafness, autosomal recessive 18A, 602092 (3), Autosomal recessive; Usher syndrome, type 1C, 276904 (3), Autosomal recessive
<i>USH1G</i>	607696	Usher syndrome, type 1G, 606943 (3), Autosomal recessive
<i>USH2A</i>	608400	Retinitis pigmentosa 39, 613809 (3); Usher syndrome, type 2A, 276901 (3), Autosomal recessive
<i>USP18</i>	607057	Pseudo-TORCH syndrome 2, 617397 (3), Autosomal recessive
<i>USP27X</i>	300975	Mental retardation, X-linked 105, 300984 (3), X-linked recessive
<i>USP45</i>	618439	?Leber congenital amaurosis 19, 618513 (3), Autosomal recessive
<i>USP7</i>	602519	Hao-Fountain syndrome, 616863 (3), Autosomal dominant
<i>USP8</i>	603158	Pituitary adenoma 4, ACTH-secreting, somatic, 219090 (3)
<i>USP9X</i>	300072	Mental retardation, X-linked 99, 300919 (3), X-linked recessive; Mental retardation, X-linked 99, syndromic, female-restricted, 300968 (3), X-linked dominant
<i>USP9Y</i>	400005	Spermatogenic failure, Y-linked, 2, 415000 (3), Y-linked
<i>UTP4</i>	607456/604901	-/North American Indian childhood cirrhosis, 604901 (2)
<i>UVSSA</i>	614632	UV-sensitive syndrome 3, 614640 (3), Autosomal recessive
<i>VAC14</i>	604632	Striatonigral degeneration, childhood-onset, 617054 (3), Autosomal recessive
<i>VAMP1</i>	185880	Spastic ataxia 1, autosomal dominant, 108600 (3), Autosomal dominant; Myasthenic syndrome, congenital, 25, 618323 (3), Autosomal recessive
<i>VAMP2</i>	185881	Neurodevelopmental disorder with hypotonia and autistic features with or without hyperkinetic movements, 618760 (3), Autosomal dominant
<i>VANGL1</i>	610132	Caudal regression syndrome, 600145 (3), Autosomal dominant; {Neural tube defects, susceptibility to}, 182940 (3), Autosomal dominant
<i>VANGL2</i>	600533	Neural tube defects, 182940 (3), Autosomal dominant
<i>VAPB</i>	605704	Spinal muscular atrophy, late-onset, Finkel type, 182980 (3), Autosomal dominant; Amyotrophic lateral sclerosis 8, 608627 (3), Autosomal dominant
<i>VARS1 (VARS)</i>	192150	Neurodevelopmental disorder with microcephaly, seizures, and cortical atrophy, 617802 (3), Autosomal recessive
<i>VARS2</i>	612802	Combined oxidative phosphorylation deficiency 20, 615917 (3), Autosomal recessive

<i>VAX1</i>	604294	?Microphthalmia, syndromic 11, 614402 (3), Autosomal recessive
<i>VCAN</i>	118661	Wagner syndrome 1, 143200 (3), Autosomal dominant
<i>VCL</i>	193065	Cardiomyopathy, dilated, 1W, 611407 (3); Cardiomyopathy, hypertrophic, 15, 613255 (3), Autosomal dominant
<i>VCP</i>	601023	Inclusion body myopathy with early-onset Paget disease and frontotemporal dementia 1, 167320 (3), Autosomal dominant; Charcot-Marie-Tooth disease, type 2Y, 616687 (3), Autosomal dominant; Amyotrophic lateral sclerosis 14, with or without frontotemporal dementia, 613954 (3)
<i>VDR</i>	601769	Rickets, vitamin D-resistant, type IIA, 277440 (3), Autosomal recessive
<i>VEGFA</i>	192240	{Microvascular complications of diabetes 1}, 603933 (3)
<i>VEGFC</i>	601528	Lymphatic malformation 4, 615907 (3), Autosomal dominant
<i>VHL</i>	608537	Pheochromocytoma, 171300 (3), Autosomal dominant; Erythrocytosis, familial, 2, 263400 (3), Autosomal recessive; von Hippel-Lindau syndrome, 193300 (3), Autosomal dominant; Renal cell carcinoma, somatic, 144700 (3); Hemangioblastoma, cerebellar, somatic (3)
<i>VIM</i>	193060	Cataract 30, pulverulent, 116300 (3), Autosomal dominant
<i>VIPAS39</i>	613401	Arthrogryposis, renal dysfunction, and cholestasis 2, 613404 (3), Autosomal recessive
<i>VKORC1</i>	608547	Warfarin resistance, 122700 (3), Autosomal dominant; Vitamin K-dependent clotting factors, combined deficiency of, 2, 607473 (3)
<i>VLDLR</i>	192977	Cerebellar hypoplasia and mental retardation with or without quadrupedal locomotion 1, 224050 (3), Autosomal recessive
<i>VMA21</i>	300913	Myopathy, X-linked, with excessive autophagy, 310440 (3), X-linked recessive
<i>VNN1</i>	603570	[High density lipoprotein cholesterol level QTL 8] (3)
<i>VPS11</i>	608549	Leukodystrophy, hypomyelinating, 12, 616683 (3), Autosomal recessive
<i>VPS13A</i>	605978	Choreoacanthocytosis, 200150 (3), Autosomal recessive
<i>VPS13B</i>	607817	Cohen syndrome, 216550 (3), Autosomal recessive
<i>VPS13C</i>	608879	Parkinson disease 23, autosomal recessive, early onset, 616840 (3), Autosomal recessive
<i>VPS13D</i>	608877	Spinocerebellar ataxia, autosomal recessive 4, 607317 (3), Autosomal recessive
<i>VPS33A</i>	610034	Mucopolysaccharidosis-plus syndrome, 617303 (3), Autosomal recessive
<i>VPS33B</i>	608552	Arthrogryposis, renal dysfunction, and cholestasis 1, 208085 (3), Autosomal recessive
<i>VPS35</i>	601501	{Parkinson disease 17}, 614203 (3), Autosomal dominant
<i>VPS37A</i>	609927	Spastic paraplegia 53, autosomal recessive, 614898 (3), Autosomal recessive

<i>VPS45</i>	610035	Neutropenia, severe congenital, 5, autosomal recessive, 615285 (3), Autosomal recessive
<i>VPS51</i>	615738	Pontocerebellar hypoplasia, type 13, 618606 (3), Autosomal recessive
<i>VPS53</i>	615850	Pontocerebellar hypoplasia, type 2E, 615851 (3), Autosomal recessive
<i>VRK1</i>	602168	Pontocerebellar hypoplasia type 1A, 607596 (3), Autosomal recessive
<i>VSX1</i>	605020	Keratoconus 1, 148300 (3), Autosomal dominant; ?Craniofacial anomalies and anterior segment dysgenesis syndrome, 614195 (3)
<i>VSX2</i>	142993	Microphthalmia with coloboma 3, 610092 (3); Microphthalmia, isolated 2, 610093 (3)
<i>VWA3B</i>	614884	?Spinocerebellar ataxia, autosomal recessive 22, 616948 (3), Autosomal recessive
<i>VWF</i>	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
<i>WAC</i>	615049	Desanto-Shinawi syndrome, 616708 (3), Autosomal dominant
<i>WARS1 (WARS)</i>	191050	Neuronopathy, distal hereditary motor, type IX, 617721 (3), Autosomal dominant
<i>WARS2</i>	604733	Neurodevelopmental disorder, mitochondrial, with abnormal movements and lactic acidosis, with or without seizures, 617710 (3), Autosomal recessive
<i>WAS</i>	300392	Thrombocytopenia, X-linked, intermittent, 313900 (3), X-linked recessive; Thrombocytopenia, X-linked, 313900 (3), X-linked recessive; Wiskott-Aldrich syndrome, 301000 (3), X-linked recessive; Neutropenia, severe congenital, X-linked, 300299 (3), X-linked recessive
<i>WASF1</i>	605035	Neurodevelopmental disorder with absent language and variable seizures, 618707 (3), Autosomal dominant
<i>WASHC4</i>	615748	?Mental retardation, autosomal recessive 43, 615817 (3), Autosomal recessive
<i>WASHC5</i>	610657	Ritscher-Schinzel syndrome 1, 220210 (3), Autosomal recessive; Spastic paraplegia 8, autosomal dominant, 603563 (3), Autosomal dominant
<i>WBP2</i>	606962	Deafness, autosomal recessive 107, 617639 (3), Autosomal recessive
<i>WDFY3</i>	617485	?Microcephaly 18, primary, autosomal dominant, 617520 (3), Autosomal dominant
<i>WDPCP</i>	613580	?Bardet-Biedl syndrome 15, 615992 (3), Autosomal recessive; ?Congenital heart defects, hamartomas of tongue, and polysyndactyly, 217085 (3), Autosomal recessive
<i>WDR1</i>	604734	Periodic fever, immunodeficiency, and thrombocytopenia syndrome, 150550 (3), Autosomal recessive

<i>WDR11</i>	606417	Hypogonadotropic hypogonadism 14 with or without anosmia, 614858 (3), Autosomal dominant
<i>WDR19</i>	608151	Nephronophthisis 13, 614377 (3), Autosomal recessive; Senior-Loken syndrome 8, 616307 (3), Autosomal recessive; ?Cranioectodermal dysplasia 4, 614378 (3), Autosomal recessive; ?Short-rib thoracic dysplasia 5 with or without polydactyly, 614376 (3), Autosomal recessive
<i>WDR26</i>	617424	Skraban-Deardorff syndrome, 617616 (3), Autosomal dominant
<i>WDR35</i>	613602	Short-rib thoracic dysplasia 7 with or without polydactyly, 614091 (3), Autosomal recessive; Cranioectodermal dysplasia 2, 613610 (3), Autosomal recessive
<i>WDR36</i>	609669	Glaucoma 1, open angle, G, 609887 (3)
<i>WDR37</i>	618586	Neurooculocardiogenitourinary syndrome, 618652 (3), Autosomal dominant
<i>WDR4</i>	605924	Microcephaly, growth deficiency, seizures, and brain malformations, 618346 (3), Autosomal recessive; Galloway-Mowat syndrome 6, 618347 (3), Autosomal recessive
<i>WDR45</i>	300526	Neurodegeneration with brain iron accumulation 5, 300894 (3), X-linked dominant
<i>WDR45B</i>	609226	Neurodevelopmental disorder with spastic quadriplegia and brain abnormalities with or without seizures, 617977 (3), Autosomal recessive
<i>WDR62</i>	613583	Microcephaly 2, primary, autosomal recessive, with or without cortical malformations, 604317 (3), Autosomal recessive
<i>WDR72</i>	613214	Amelogenesis imperfecta, type IIA3, 613211 (3), Autosomal recessive
<i>WDR73</i>	616144	Galloway-Mowat syndrome 1, 251300 (3), Autosomal recessive
<i>WDR81</i>	614218	Hydrocephalus, congenital, 3, with brain anomalies, 617967 (3), Autosomal recessive; Cerebellar ataxia, mental retardation, and dysequilibrium syndrome 2, 610185 (3), Autosomal recessive
<i>WEE2</i>	614084	Oocyte maturation defect 5, 617996 (3), Autosomal recessive
<i>WFS1</i>	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-like syndrome, autosomal dominant, 614296 (3), Autosomal dominant; Wolfram syndrome 1, 222300 (3), Autosomal recessive
<i>WHRN</i>	607928	Deafness, autosomal recessive 31, 607084 (3), Autosomal recessive; Usher syndrome, type 2D, 611383 (3), Autosomal recessive
<i>WIPF1</i>	602357	Wiskott-Aldrich syndrome 2, 614493 (3)
<i>WIPI2</i>	609225	?Intellectual developmental disorder with short stature and variable skeletal anomalies, 618453 (3), Autosomal recessive
<i>WNK1</i>	605232	Pseudohypoaldosteronism, type IIC, 614492 (3), Autosomal dominant; Neuropathy, hereditary sensory and autonomic, type II, 201300 (3), Autosomal recessive

<i>WNK4</i>	601844	Pseudohypoaldosteronism, type IIB, 614491 (3), Autosomal dominant
<i>WNT1</i>	164820	{Osteoporosis, early-onset, susceptibility to, autosomal dominant}, 615221 (3); Osteogenesis imperfecta, type XV, 615220 (3), Autosomal recessive
<i>WNT10A</i>	606268	Schopf-Schulz-Passarge syndrome, 224750 (3), Autosomal recessive; Tooth agenesis, selective, 4, 150400 (3), Autosomal recessive, Autosomal dominant; Odontoonychodermal dysplasia, 257980 (3), Autosomal recessive
<i>WNT10B</i>	601906	Split-hand/foot malformation 6, 225300 (3), Autosomal recessive; Tooth agenesis, selective, 8, 617073 (3), Autosomal dominant
<i>WNT2B</i>	601968	Diarrhea 9, 618168 (3), Autosomal recessive
<i>WNT3</i>	165330	?Tetra-amelia syndrome 1, 273395 (3), Autosomal recessive
<i>WNT4</i>	603490	?SERKAL syndrome, 611812 (3), Autosomal recessive; Mullerian aplasia and hyperandrogenism, 158330 (3), Autosomal dominant
<i>WNT5A</i>	164975	Robinow syndrome, autosomal dominant 1, 180700 (3), Autosomal dominant
<i>WNT7A</i>	601570	Fuhrmann syndrome, 228930 (3), Autosomal recessive; Ulna and fibula, absence of, with severe limb deficiency, 276820 (3), Autosomal recessive
<i>WRAP53</i>	612661	Dyskeratosis congenita, autosomal recessive 3, 613988 (3), Autosomal recessive
<i>WRN</i>	604611	Werner syndrome, 277700 (3), Autosomal recessive
<i>WT1</i>	607102	Mesothelioma, somatic, 156240 (3); Wilms tumor, type 1, 194070 (3), Autosomal dominant, Somatic mutation; Frasier syndrome, 136680 (3), Autosomal dominant, Somatic mutation; Denys-Drash syndrome, 194080 (3), Autosomal dominant, Somatic mutation; Meacham syndrome, 608978 (3); Nephrotic syndrome, type 4, 256370 (3), Autosomal dominant
<i>WWC1</i>	610533	[Memory, enhanced, QTL], 615602 (3)
<i>WWOX</i>	605131	Spinocerebellar ataxia, autosomal recessive 12, 614322 (3), Autosomal recessive; Epileptic encephalopathy, early infantile, 28, 616211 (3), Autosomal recessive; Esophageal squamous cell carcinoma, somatic, 133239 (3)
<i>XBP1</i>	194355	{Major affective disorder-7, susceptibility to}, 612371 (3)
<i>XDH</i>	607633	Xanthinuria, type I, 278300 (3), Autosomal recessive
<i>XG</i>	300879/314700	-/[Blood group, XG system] (3)
<i>XIAP</i>	300079	Lymphoproliferative syndrome, X-linked, 2, 300635 (3), X-linked recessive
<i>XK</i>	314850	McLeod syndrome with or without chronic granulomatous disease, 300842 (3), X-linked
<i>XPA</i>	611153	Xeroderma pigmentosum, group A, 278700 (3), Autosomal recessive
<i>XPC</i>	613208	Xeroderma pigmentosum, group C, 278720 (3), Autosomal recessive

<i>XPNPEP2</i>	300145	{Angioedema induced by ACE inhibitors, susceptibility to}, 300909 (3)
<i>XPNPEP3</i>	613553	Nephronophthisis-like nephropathy 1, 613159 (3), Autosomal recessive
<i>XPR1</i>	605237	Basal ganglia calcification, idiopathic, 6, 616413 (3), Autosomal dominant
<i>XRCC1</i>	194360	?Spinocerebellar ataxia, autosomal recessive 26, 617633 (3), Autosomal recessive
<i>XRCC2</i>	600375	?Fanconi anemia, complementation group U, 617247 (3), Autosomal recessive
<i>XRCC3</i>	600675	{Melanoma, cutaneous malignant, 6}, 613972 (3); {Breast cancer, susceptibility to}, 114480 (3), Autosomal dominant, Somatic mutation
<i>XRCC4</i>	194363	Short stature, microcephaly, and endocrine dysfunction, 616541 (3), Autosomal recessive
<i>XYLT1</i>	608124	{Pseudoxanthoma elasticum, modifier of severity of}, 264800 (3), Autosomal recessive; Desbuquois dysplasia 2, 615777 (3), Autosomal recessive
<i>XYLT2</i>	608125	{Pseudoxanthoma elasticum, modifier of severity of}, 264800 (3), Autosomal recessive; Spondyloocular syndrome, 605822 (3), Autosomal recessive
<i>YAP1</i>	606608	Coloboma, ocular, with or without hearing impairment, cleft lip/palate, and/or mental retardation, 120433 (3), Autosomal dominant
<i>YARS1 (YARS)</i>	603623	Charcot-Marie-Tooth disease, dominant intermediate C, 608323 (3), Autosomal dominant
<i>YARS2</i>	610957	Myopathy, lactic acidosis, and sideroblastic anemia 2, 613561 (3), Autosomal recessive
<i>YEATS2</i>	613373	?Epilepsy, myoclonic, familial adult, 4, 615127 (3), Autosomal dominant
<i>YME1L1</i>	607472	?Optic atrophy 11, 617302 (3), Autosomal recessive
<i>YWHAG</i>	605356	Epileptic encephalopathy, early infantile, 56, 617665 (3), Autosomal dominant
<i>YY1</i>	600013	Gabriele-de Vries syndrome, 617557 (3), Autosomal dominant
<i>YY1AP1</i>	607860	Grange syndrome, 602531 (3), Autosomal recessive
<i>ZAP70</i>	176947	Autoimmune disease, multisystem, infantile-onset, 2, 617006 (3), Autosomal recessive; Immunodeficiency 48, 269840 (3), Autosomal recessive
<i>ZBTB11</i>	618181	Intellectual developmental disorder, autosomal recessive 69, 618383 (3), Autosomal recessive
<i>ZBTB16</i>	176797	Skeletal defects, genital hypoplasia, and mental retardation, 612447 (3), Autosomal recessive; Leukemia, acute promyelocytic, PL2F/RARA type (3)
<i>ZBTB18</i>	608433	Mental retardation, autosomal dominant 22, 612337 (3), Autosomal dominant
<i>ZBTB20</i>	606025	Primrose syndrome, 259050 (3), Autosomal dominant

<i>ZBTB24</i>	614064	Immunodeficiency-centromeric instability-facial anomalies syndrome 2, 614069 (3), Autosomal recessive
<i>ZBTB42</i>	613915	?Lethal congenital contracture syndrome 6, 616248 (3), Autosomal recessive
<i>ZC3H14</i>	613279	Mental retardation, autosomal recessive 56, 617125 (3), Autosomal recessive
<i>ZC4H2</i>	300897	Wieacker-Wolff syndrome, 314580 (3), X-linked recessive; Wieacker-Wolff syndrome, female-restricted, 301041 (3), X-linked dominant
<i>ZCCHC8</i>	616381	?Pulmonary fibrosis and/or bone marrow failure, telomere-related, 5, 618674 (3), Autosomal dominant
<i>ZDHH15</i>	300576/300577	-/Mental retardation, X-linked 91, 300577 (4), X-linked dominant
<i>ZDHH9</i>	300646	Mental retardation, X-linked syndromic, Raymond type, 300799 (3), X-linked
<i>ZEB1</i>	189909	Corneal dystrophy, posterior polymorphous, 3, 609141 (3); Corneal dystrophy, Fuchs endothelial, 6, 613270 (3)
<i>ZEB2</i>	605802	Mowat-Wilson syndrome, 235730 (3), Autosomal dominant
<i>ZFAT</i>	610931	{Autoimmune thyroid disease, susceptibility to, 3}, 608175 (3)
<i>ZFHX2</i>	617828	?Marsili syndrome, 147430 (3), Autosomal dominant
<i>ZFHX3</i>	104155	Prostate cancer, somatic, 176807 (3)
<i>ZFHX4</i>	606940	?Ptosis, congenital, 178300 (2), Autosomal dominant
<i>ZFP57</i>	612192	Diabetes mellitus, transient neonatal, 1, 601410 (3), Autosomal dominant
<i>ZFPM2</i>	603693	46XY sex reversal 9, 616067 (3), Autosomal dominant; Diaphragmatic hernia 3, 610187 (3); Tetralogy of Fallot, 187500 (3), Autosomal dominant
<i>ZFYVE26</i>	612012	Spastic paraplegia 15, autosomal recessive, 270700 (3), Autosomal recessive
<i>ZFYVE27</i>	610243	Spastic paraplegia 33, autosomal dominant, 610244 (3), Autosomal dominant
<i>ZIC1</i>	600470	Structural brain anomalies with impaired intellectual development and craniosynostosis, 618736 (3), Autosomal dominant; ?Craniosynostosis 6, 616602 (3), Autosomal dominant
<i>ZIC2</i>	603073	Holoprosencephaly 5, 609637 (3), Autosomal dominant
<i>ZIC3</i>	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
<i>ZMIZ1</i>	607159	Neurodevelopmental disorder with dysmorphic facies and distal skeletal anomalies, 618659 (3), Autosomal dominant
<i>ZMPSTE24</i>	606480	Restrictive dermopathy, lethal, 275210 (3), Autosomal recessive; Mandibuloacral dysplasia with type B lipodystrophy, 608612 (3), Autosomal recessive
<i>ZMYND10</i>	607070	Ciliary dyskinesia, primary, 22, 615444 (3), Autosomal recessive
<i>ZMYND11</i>	608668	Mental retardation, autosomal dominant 30, 616083 (3), Autosomal dominant

ZMYND15	614312	?Spermatogenic failure 14, 615842 (3), Autosomal recessive
ZNF141	194648	?Polydactyly, postaxial, type A6, 615226 (3), Autosomal recessive
ZNF142	604083	Neurodevelopmental disorder with impaired speech and hyperkinetic movements, 618425 (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
ZNF341	618269	Hyper-IgE recurrent infection syndrome 3, autosomal recessive, 618282 (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
ZNF41	314995/300848	-/Mental retardation, X-linked 89, 300848 (2), X-linked dominant
ZNF423	604557	Nephronophthisis 14, 614844 (3), Autosomal recessive, Autosomal dominant; Joubert syndrome 19, 614844 (3), Autosomal recessive, Autosomal dominant
ZNF462	617371	Weiss-Kruszka syndrome, 618619 (3), Autosomal dominant
ZNF469	612078	Brittle cornea syndrome 1, 229200 (3), Autosomal recessive
ZNF513	613598	?Retinitis pigmentosa 58, 613617 (3), Autosomal recessive
ZNF644	614159	Myopia 21, autosomal dominant, 614167 (3), Autosomal dominant
ZNF674	300573/300851	-/Mental retardation, X-linked 92, 300851 (2), X-linked recessive
ZNF687	610568	Paget disease of bone 6, 616833 (3), Autosomal dominant
ZNF711	314990	Mental retardation, X-linked 97, 300803 (3), X-linked
ZNF750	610226	Seborrhea-like dermatitis with psoriasiform elements, 610227 (3)
ZNF81	314998/300498	-/Mental retardation,X-linked 45, 300498 (2), X-linked
ZNHIT3	604500	PEHO syndrome, 260565 (3), Autosomal recessive
ZP1	195000	Oocyte maturation defect 1, 615774 (3), Autosomal recessive
ZP2	182888	Oocyte maturation defect 6, 618353 (3), Autosomal recessive
ZP3	182889	Oocyte maturation defect 3, 617712 (3), Autosomal dominant
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: Sep 01, 2020

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.