|  |  |  |
| --- | --- | --- |
| **Mendeliome panel** | | |
| **versie** | v4 (4542 genen) | Centrum voor Medische Genetica Gent |
|  |  |  |
|  |  |  |
| **Gene** | **OMIM gene ID** | **Associated phenotype, OMIM phenotype ID, phenotype mapping key and inheritance pattern** |
| *A2ML1* | 610627 | {Otitis media, susceptibility to}, 166760 (3), Autosomal dominant |
| *A4GALT* | 607922 | [Blood group, P1Pk system, P(2) phenotype], 111400 (3); NOR polyagglutination syndrome, 111400 (3); [Blood group, P1Pk system, p phenotype], 111400 (3) |
| *AAAS* | 605378 | Achalasia-addisonianism-alacrimia syndrome, 231550 (3), Autosomal recessive |
| *AAGAB* | 614888 | Keratoderma, palmoplantar, punctate type IA, 148600 (3), Autosomal dominant |
| *AARS1 (AARS)* | 601065 | Developmental and epileptic encephalopathy 29, 616339 (3), Autosomal recessive; Charcot-Marie-Tooth disease, axonal, type 2N, 613287 (3), Autosomal dominant |
| *AARS2* | 612035 | Leukoencephalopathy, progressive, with ovarian failure, 615889 (3), Autosomal recessive; Combined oxidative phosphorylation deficiency 8, 614096 (3), Autosomal recessive |
| *AASS* | 605113 | Hyperlysinemia, 238700 (3), Autosomal recessive |
| *ABAT* | 137150 | GABA-transaminase deficiency, 613163 (3), Autosomal recessive |
| *ABCA1* | 600046 | Tangier disease, 205400 (3), Autosomal recessive; HDL deficiency, familial, 1, 604091 (3) |
| *ABCA12* | 607800 | Ichthyosis, congenital, autosomal recessive 4B (harlequin), 242500 (3), Autosomal recessive; Ichthyosis, congenital, autosomal recessive 4A, 601277 (3), Autosomal recessive |
| *ABCA2* | 600047 | Intellectual developmental disorder with poor growth and with or without seizures or ataxia, 618808 (3), Autosomal recessive |
| *ABCA3* | 601615 | Surfactant metabolism dysfunction, pulmonary, 3, 610921 (3), Autosomal recessive |
| *ABCA4* | 601691 | Retinal dystrophy, early-onset severe, 248200 (3), Autosomal recessive; Retinitis pigmentosa 19, 601718 (3), Autosomal recessive; {Macular degeneration, age-related, 2}, 153800 (3), Autosomal dominant; Cone-rod dystrophy 3, 604116 (3); Fundus flavimaculatus, 248200 (3), Autosomal recessive; Stargardt disease 1, 248200 (3), Autosomal recessive |
| *ABCA5* | 612503 | ?Hypertrichosis, congenital generalized, with gingival hyperplasia, 135400 (3), Autosomal recessive |
| *ABCA7* | 605414 | {Alzheimer disease 9, susceptibility to}, 608907 (3), Autosomal dominant |
| *ABCB1* | 171050 | {Inflammatory bowel disease 13}, 612244 (3); {Colchicine resistance}, 120080 (3) |
| *ABCB11* | 603201 | Cholestasis, benign recurrent intrahepatic, 2, 605479 (3), Autosomal recessive; Cholestasis, progressive familial intrahepatic 2, 601847 (3), Autosomal recessive |
| *ABCB4* | 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 |
| *ABCB6* | 605452 | Microphthalmia, isolated, with coloboma 7, 614497 (3), Autosomal dominant; Dyschromatosis universalis hereditaria 3, 615402 (3), Autosomal dominant; [Blood group, Langereis system], 111600 (3); Pseudohyperkalemia, familial, 2, due to red cell leak, 609153 (3), Autosomal dominant |
| *ABCB7* | 300135 | Anemia, sideroblastic, with ataxia, 301310 (3), X-linked recessive |
| *ABCC1* | 158343 | ?Deafness, autosomal dominant 77, 618915 (3), Autosomal dominant |
| *ABCC11* | 607040 | [Axillary odor, variation in], 117800 (3), Autosomal dominant; [Earwax, wet/dry], 117800 (3), Autosomal dominant; [Colostrum secretion, variation in], 117800 (3), Autosomal dominant |
| *ABCC2* | 601107 | Dubin-Johnson syndrome, 237500 (3), Autosomal recessive |
| *ABCC6* | 603234 | Pseudoxanthoma elasticum, 264800 (3), Autosomal recessive; Arterial calcification, generalized, of infancy, 2, 614473 (3), Autosomal recessive; Pseudoxanthoma elasticum, forme fruste, 177850 (3), Autosomal dominant |
| *ABCC8* | 600509 | Diabetes mellitus, permanent neonatal 3, with or without neurologic features, 618857 (3), Autosomal recessive, Autosomal dominant; Diabetes mellitus, transient neonatal 2, 610374 (3); Diabetes mellitus, noninsulin-dependent, 125853 (3), Autosomal dominant; Hypoglycemia of infancy, leucine-sensitive, 240800 (3), Autosomal dominant; Hyperinsulinemic hypoglycemia, familial, 1, 256450 (3), Autosomal recessive, Autosomal dominant |
| *ABCC9* | 601439 | Cardiomyopathy, dilated, 1O, 608569 (3), Autosomal dominant; Hypertrichotic osteochondrodysplasia, 239850 (3), Autosomal dominant; ?Atrial fibrillation, familial, 12, 614050 (3), Autosomal dominant |
| *ABCD1* | 300371 | Adrenoleukodystrophy, 300100 (3), X-linked recessive; Adrenomyeloneuropathy, adult, 300100 (3), X-linked recessive |
| *ABCD3* | 170995 | ?Bile acid synthesis defect, congenital, 5, 616278 (3), Autosomal recessive |
| *ABCD4* | 603214 | Methylmalonic aciduria and homocystinuria, cblJ type, 614857 (3), Autosomal recessive |
| *ABCG2* | 603756 | [Junior blood group system], 614490 (3); [Uric acid concentration, serum, QTL1], 138900 (3), ?Autosomal dominant |
| *ABCG5* | 605459 | Sitosterolemia 2, 618666 (3), Autosomal recessive |
| *ABCG8* | 605460 | Sitosterolemia 1, 210250 (3), Autosomal recessive; {Gallbladder disease 4}, 611465 (3) |
| *ABHD12* | 613599 | Polyneuropathy, hearing loss, ataxia, retinitis pigmentosa, and cataract, 612674 (3), Autosomal recessive |
| *ABHD5* | 604780 | Chanarin-Dorfman syndrome, 275630 (3), Autosomal recessive |
| *ABL1* | 189980 | Leukemia, Philadelphia chromosome-positive, resistant to imatinib, 608232 (3), Somatic mutation; Congenital heart defects and skeletal malformations syndrome, 617602 (3), Autosomal dominant |
| *ABO* | 110300 | [Blood group, ABO system], 616093 (3) |
| *ACACA* | 200350 | Acetyl-CoA carboxylase deficiency, 613933 (1), Autosomal recessive |
| *ACAD8* | 604773 | Isobutyryl-CoA dehydrogenase deficiency, 611283 (3), Autosomal recessive |
| *ACAD9* | 611103 | Mitochondrial complex I deficiency, nuclear type 20, 611126 (3), Autosomal recessive |
| *ACADM* | 607008 | Acyl-CoA dehydrogenase, medium chain, deficiency of, 201450 (3), Autosomal recessive |
| *ACADS* | 606885 | Acyl-CoA dehydrogenase, short-chain, deficiency of, 201470 (3), Autosomal recessive |
| *ACADSB* | 600301 | 2-methylbutyrylglycinuria, 610006 (3), Autosomal recessive |
| *ACADVL* | 609575 | VLCAD deficiency, 201475 (3), Autosomal recessive |
| *ACAN* | 155760 | ?Spondyloepiphyseal dysplasia, Kimberley type, 608361 (3), Autosomal dominant; 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 |
| *ACAT1* | 607809 | Alpha-methylacetoacetic aciduria, 203750 (3), Autosomal recessive |
| *ACAT2* | 100678 | ?ACAT2 deficiency, 614055 (1), Isolated cases |
| *ACBD5* | 616618 | Retinal dystrophy with leukodystrophy, 618863 (3), Autosomal recessive |
| *ACD* | 609377 | ?Dyskeratosis congenita, autosomal recessive 7, 616553 (3), Autosomal recessive, Autosomal dominant; ?Dyskeratosis congenita, autosomal dominant 6, 616553 (3), Autosomal recessive, Autosomal dominant |
| *ACE* | 106180 | {Stroke, hemorrhagic}, 614519 (3); 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) |
| *ACER3* | 617036 | ?Leukodystrophy, progressive, early childhood-onset, 617762 (3), Autosomal recessive |
| *ACHE* | 100740 | [Blood group, Yt system], 112100 (3) |
| *ACKR1* | 613665 | [Blood group, Duffy system], 110700 (3), Autosomal recessive, Autosomal dominant; [White blood cell count QTL], 611862 (3), Autosomal recessive; {Malaria, vivax, protection against}, 611162 (3) |
| *ACKR3* | 610376 | ?Oculomotor-abducens synkinesis, 619215 (3), Autosomal recessive |
| *ACO2* | 100850 | ?Optic atrophy 9, 616289 (3), Autosomal recessive; Infantile cerebellar-retinal degeneration, 614559 (3), Autosomal recessive |
| *ACOX1* | 609751 | Mitchell syndrome, 618960 (3), Autosomal dominant; Peroxisomal acyl-CoA oxidase deficiency, 264470 (3), Autosomal recessive |
| *ACOX2* | 601641 | Bile acid synthesis defect, congenital, 6, 617308 (3), Autosomal recessive |
| *ACP2* | 171650 | ?Lysosomal acid phosphatase deficiency, 200950 (1), Autosomal recessive |
| *ACP4* | 606362 | Amelogenesis imperfecta, type IJ, 617297 (3), Autosomal recessive |
| *ACP5* | 171640 | Spondyloenchondrodysplasia with immune dysregulation, 607944 (3), Autosomal recessive |
| *ACR* | 102480 | ?Male infertility due to acrosin deficiency, 102480 (2) |
| *ACSF3* | 614245 | Combined malonic and methylmalonic aciduria, 614265 (3) |
| *ACSL4* | 300157 | Mental retardation, X-linked 63, 300387 (3), X-linked dominant |
| *ACSL6* | 604443 | Myelodysplastic syndrome (3); Myelogenous leukemia, acute (3) |
| *ACSM3* | 145505 | {?Hypertension, essential} (1) |
| *ACTA1* | 102610 | ?Myopathy, scapulohumeroperoneal, 616852 (3), Autosomal dominant; Nemaline myopathy 3, autosomal dominant or recessive, 161800 (3), Autosomal recessive, Autosomal dominant; Myopathy, actin, congenital, with excess of thin myofilaments, 161800 (3), Autosomal recessive, Autosomal dominant; Myopathy, actin, congenital, with cores, 161800 (3), Autosomal recessive, Autosomal dominant; Myopathy, congenital, with fiber-type disproportion 1, 255310 (3), Autosomal recessive, Autosomal dominant |
| *ACTA2* | 102620 | Multisystemic smooth muscle dysfunction syndrome, 613834 (3), Autosomal dominant; Aortic aneurysm, familial thoracic 6, 611788 (3), Autosomal dominant; Moyamoya disease 5, 614042 (3) |
| *ACTB* | 102630 | Baraitser-Winter syndrome 1, 243310 (3), Autosomal dominant; ?Dystonia, juvenile-onset, 607371 (3), Autosomal dominant |
| *ACTC1* | 102540 | Left ventricular noncompaction 4, 613424 (3), Autosomal dominant; Cardiomyopathy, hypertrophic, 11, 612098 (3), Autosomal dominant; Atrial septal defect 5, 612794 (3), Autosomal dominant; Cardiomyopathy, dilated, 1R, 613424 (3), Autosomal dominant |
| *ACTG1* | 102560 | Deafness, autosomal dominant 20/26, 604717 (3), Autosomal dominant; Baraitser-Winter syndrome 2, 614583 (3), Autosomal dominant |
| *ACTG2* | 102545 | Megacystis-microcolon-intestinal hypoperistalsis syndrome 5, 619431 (3), Autosomal dominant; Visceral myopathy 1, 155310 (3), Autosomal dominant |
| *ACTL6B* | 612458 | Developmental and epileptic encephalopathy 76, 618468 (3), Autosomal recessive; Intellectual developmental disorder with severe speech and ambulation defects, 618470 (3), Autosomal dominant |
| *ACTL9* | 619251 | Spermatogenic failure 53, 619258 (3) |
| *ACTN1* | 102575 | Bleeding disorder, platelet-type, 15, 615193 (3), Autosomal dominant |
| *ACTN2* | 102573 | Myopathy, distal, 6, adult onset, 618655 (3), Autosomal dominant; Cardiomyopathy, hypertrophic, 23, with or without LVNC, 612158 (3), Autosomal dominant; Cardiomyopathy, dilated, 1AA, with or without LVNC, 612158 (3), Autosomal dominant; Myopathy, congenital with structured cores and Z-line abnormalities, 618654 (3), Autosomal dominant |
| *ACTN3* | 102574 | [Sprinting performance], 617749 (3); [Alpha-actinin-3 deficiency], 617749 (3) |
| *ACTN4* | 604638 | Glomerulosclerosis, focal segmental, 1, 603278 (3), Autosomal dominant |
| *ACVR1* | 102576 | Fibrodysplasia ossificans progressiva, 135100 (3), Autosomal dominant |
| *ACVR1B* | 601300 | Pancreatic cancer, somatic (3) |
| *ACVR2B* | 602730 | Heterotaxy, visceral, 4, autosomal, 613751 (3) |
| *ACVRL1* | 601284 | Telangiectasia, hereditary hemorrhagic, type 2, 600376 (3), Autosomal dominant |
| *ACY1* | 104620 | Aminoacylase 1 deficiency, 609924 (3), Autosomal recessive |
| *ADA* | 608958 | Adenosine deaminase deficiency, partial, 102700 (3), Autosomal recessive, Somatic mosaicism; Severe combined immunodeficiency due to ADA deficiency, 102700 (3), Autosomal recessive, Somatic mosaicism |
| *ADA2* | 607575 | Sneddon syndrome, 182410 (3), Autosomal recessive; Vasculitis, autoinflammation, immunodeficiency, and hematologic defects syndrome, 615688 (3), Autosomal recessive |
| *ADAM10* | 602192 | {Alzheimer disease 18, susceptibility to}, 615590 (3); Reticulate acropigmentation of Kitamura, 615537 (3), Autosomal dominant |
| *ADAM17* | 603639 | ?Inflammatory skin and bowel disease, neonatal, 1, 614328 (3), Autosomal recessive |
| *ADAM22* | 603709 | Developmental and epileptic encephalopathy 61, 617933 (3), Autosomal recessive |
| *ADAM9* | 602713 | Cone-rod dystrophy 9, 612775 (3), Autosomal recessive |
| *ADAMTS10* | 608990 | Weill-Marchesani syndrome 1, recessive, 277600 (3), Autosomal recessive |
| *ADAMTS13* | 604134 | Thrombotic thrombocytopenic purpura, hereditary, 274150 (3), Autosomal recessive |
| *ADAMTS17* | 607511 | Weill-Marchesani 4 syndrome, recessive, 613195 (3), Autosomal recessive |
| *ADAMTS18* | 607512 | Microcornea, myopic chorioretinal atrophy, and telecanthus, 615458 (3), Autosomal recessive |
| *ADAMTS2* | 604539 | Ehlers-Danlos syndrome, dermatosparaxis type, 225410 (3), Autosomal recessive |
| *ADAMTS3* | 605011 | Hennekam lymphangiectasia-lymphedema syndrome 3, 618154 (3), Autosomal recessive |
| *ADAMTSL2* | 612277 | Geleophysic dysplasia 1, 231050 (3), Autosomal recessive |
| *ADAMTSL4* | 610113 | Ectopia lentis et pupillae, 225200 (3), Autosomal recessive; Ectopia lentis, isolated, autosomal recessive, 225100 (3), Autosomal recessive |
| *ADAR* | 146920 | Dyschromatosis symmetrica hereditaria, 127400 (3), Autosomal dominant; Aicardi-Goutieres syndrome 6, 615010 (3), Autosomal recessive |
| *ADARB1* | 601218 | Neurodevelopmental disorder with hypotonia, microcephaly, and seizures, 618862 (3), Autosomal recessive |
| *ADAT3* | 615302 | Mental retardation, autosomal recessive 36, 615286 (3), Autosomal recessive |
| *ADCY1* | 103072 | ?Deafness, autosomal recessive 44, 610154 (3), Autosomal recessive |
| *ADCY10* | 605205 | {Hypercalciuria, absorptive, susceptibility to}, 143870 (3), Autosomal dominant |
| *ADCY3* | 600291 | {Obesity, susceptibility to, BMIQ19}, 617885 (3), Autosomal recessive |
| *ADCY5* | 600293 | Dyskinesia, familial, with facial myokymia, 606703 (3), Autosomal dominant |
| *ADCY6* | 600294 | Lethal congenital contracture syndrome 8, 616287 (3), Autosomal recessive |
| *ADD1* | 102680 | {Hypertension, essential, salt-sensitive}, 145500 (3), Multifactorial |
| *ADD3* | 601568 | Cerebral palsy, spastic quadriplegic, 3, 617008 (3), Autosomal recessive |
| *ADGRE2* | 606100 | Vibratory urticaria, 125630 (3), Autosomal dominant |
| *ADGRG1* | 604110 | Polymicrogyria, bilateral frontoparietal, 606854 (3), Autosomal recessive; Polymicrogyria, bilateral perisylvian, 615752 (3) |
| *ADGRG2* | 300572 | Congenital bilateral absence of vas deferens, X-linked, 300985 (3), X-linked |
| *ADGRG6* | 612243 | Lethal congenital contracture syndrome 9, 616503 (3), Autosomal recessive |
| *ADGRV1* | 602851 | Usher syndrome, type 2C, 605472 (3), Digenic dominant, Autosomal recessive; Usher syndrome, type 2C, GPR98/PDZD7 digenic, 605472 (3), Digenic dominant, Autosomal recessive; ?Febrile seizures, familial, 4, 604352 (3), Autosomal dominant |
| *ADH1B* | 103720 | {Aerodigestive tract cancer, squamous cell, alcohol-related, protection against}, 103780 (3), Multifactorial; {Alcohol dependence, protection against}, 103780 (3), Multifactorial |
| *ADH1C* | 103730 | {Alcohol dependence, protection against}, 103780 (3), Multifactorial; {Parkinson disease, susceptibility to}, 168600 (3), Multifactorial, Autosomal dominant |
| *ADH5* | 103710 | AMED syndrome, digenic, 619151 (3), Digenic recessive |
| *ADIPOQ* | 605441 | Adiponectin deficiency, 612556 (3) |
| *ADK* | 102750 | Hypermethioninemia due to adenosine kinase deficiency, 614300 (3), Autosomal recessive |
| *ADNP* | 611386 | Helsmoortel-van der Aa syndrome, 615873 (3), Autosomal dominant |
| *ADPRS (ADPRHL2)* | 610624 | Neurodegeneration, childhood-onset, stress-induced, with variable ataxia and seizures, 618170 (3), Autosomal recessive |
| *ADRA2C* | 104250 | {Congestive heart failure and beta-blocker response, modifier of} (3) |
| *ADRB1* | 109630 | [Short sleep, familial natural, 2], 618591 (3), Autosomal dominant; [Resting heart rate], 607276 (3) |
| *ADRB2* | 109690 | Beta-2-adrenoreceptor agonist, reduced response to (3); {Obesity, susceptibility to}, 601665 (3), Multifactorial, Autosomal recessive, Autosomal dominant; {Asthma, nocturnal, susceptibility to}, 600807 (3), Autosomal dominant |
| *ADRB3* | 109691 | {Obesity, susceptibility to}, 601665 (3), Multifactorial, Autosomal recessive, Autosomal dominant |
| *ADSL* | 608222 | Adenylosuccinase deficiency, 103050 (3), Autosomal recessive |
| *ADSS1 (ADSSL1)* | 612498 | Myopathy, distal, 5, 617030 (3), Autosomal recessive |
| *AEBP1* | 602981 | Ehlers-Danlos syndrome, classic-like, 2, 618000 (3), Autosomal recessive |
| *AFF2* | 300806 | Intellectual developmental disorder, X-linked 109, 309548 (3), X-linked recessive |
| *AFF3* | 601464 | KINSSHIP syndrome, 619297 (3), Autosomal dominant |
| *AFF4* | 604417 | CHOPS syndrome, 616368 (3), Autosomal dominant |
| *AFG3L2* | 604581 | Spastic ataxia 5, autosomal recessive, 614487 (3), Autosomal recessive; Optic atrophy 12, 618977 (3), Autosomal dominant; Spinocerebellar ataxia 28, 610246 (3), Autosomal dominant |
| *AFP* | 104150 | [Hereditary persistence of alpha-fetoprotein], 615970 (3), Autosomal dominant; Alpha-fetoprotein deficiency, 615969 (3), Autosomal recessive |
| *AGA* | 613228 | Aspartylglucosaminuria, 208400 (3), Autosomal recessive |
| *AGBL1* | 615496 | Corneal dystrophy, Fuchs endothelial, 8, 615523 (3), Autosomal dominant |
| *AGBL5* | 615900 | Retinitis pigmentosa 75, 617023 (3), Autosomal recessive |
| *AGK* | 610345 | Cataract 38, autosomal recessive, 614691 (3), Autosomal recessive; Sengers syndrome, 212350 (3), Autosomal recessive |
| *AGL* | 610860 | Glycogen storage disease IIIa, 232400 (3), Autosomal recessive; Glycogen storage disease IIIb, 232400 (3), Autosomal recessive |
| *AGO2* | 606229 | Lessel-Kreienkamp syndrome, 619149 (3), Autosomal dominant |
| *AGPAT2* | 603100 | Lipodystrophy, congenital generalized, type 1, 608594 (3), Autosomal recessive |
| *AGPS* | 603051 | Rhizomelic chondrodysplasia punctata, type 3, 600121 (3), Autosomal recessive |
| *AGRN* | 103320 | Myasthenic syndrome, congenital, 8, with pre- and postsynaptic defects, 615120 (3), Autosomal recessive |
| *AGRP* | 602311 | {Leanness, inherited}, 601665 (3), Multifactorial, Autosomal recessive, Autosomal dominant; {Obesity, late-onset}, 601665 (3), Multifactorial, Autosomal recessive, Autosomal dominant |
| *AGT* | 106150 | Renal tubular dysgenesis, 267430 (3), Autosomal recessive; {Preeclampsia, susceptibility to} (3); {Hypertension, essential, susceptibility to}, 145500 (3), Multifactorial |
| *AGTPBP1* | 606830 | Neurodegeneration, childhood-onset, with cerebellar atrophy, 618276 (3), Autosomal recessive |
| *AGTR1* | 106165 | {Hypertension, essential}, 145500 (3), Multifactorial; Renal tubular dysgenesis, 267430 (3), Autosomal recessive |
| *AGXT* | 604285 | Hyperoxaluria, primary, type 1, 259900 (3), Autosomal recessive |
| *AGXT2* | 612471 | [Beta-aminoisobutyric acid, urinary excretion of], 210100 (3), Autosomal recessive |
| *AHCY* | 180960 | Hypermethioninemia with deficiency of S-adenosylhomocysteine hydrolase, 613752 (3), Autosomal recessive |
| *AHDC1* | 615790 | Xia-Gibbs syndrome, 615829 (3), Autosomal dominant |
| *AHI1* | 608894 | Joubert syndrome 3, 608629 (3), Autosomal recessive |
| *AHR* | 600253 | ?Retinitis pigmentosa 85, 618345 (3), Autosomal recessive |
| *AHSG* | 138680 | ?Alopecia-mental retardation syndrome 1, 203650 (3), Autosomal recessive |
| *AICDA* | 605257 | Immunodeficiency with hyper-IgM, type 2, 605258 (3), Autosomal recessive |
| *AIFM1* | 300169 | Combined oxidative phosphorylation deficiency 6, 300816 (3), X-linked recessive; Cowchock syndrome, 310490 (3), X-linked recessive; Spondyloepimetaphyseal dysplasia, X-linked, with hypomyelinating leukodystrophy, 300232 (3), X-linked recessive; Deafness, X-linked 5, 300614 (3), X-linked recessive |
| *AIMP1* | 603605 | Leukodystrophy, hypomyelinating, 3, 260600 (3), Autosomal recessive |
| *AIMP2* | 600859 | Leukodystrophy, hypomyelinating, 17, 618006 (3), Autosomal recessive |
| *AIP* | 605555 | Pituitary adenoma 1, multiple types, 102200 (3), Somatic mutation, Autosomal dominant; Pituitary adenoma predisposition, 102200 (3), Somatic mutation, Autosomal dominant |
| *AIPL1* | 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 |
| *AIRE* | 607358 | Autoimmune polyendocrinopathy syndrome , type I, with or without reversible metaphyseal dysplasia, 240300 (3), Autosomal recessive, Autosomal dominant |
| *AK1* | 103000 | Hemolytic anemia due to adenylate kinase deficiency, 612631 (3), Autosomal recessive |
| *AK2* | 103020 | Reticular dysgenesis, 267500 (3), Autosomal recessive |
| *AK7* | 615364 | ?Spermatogenic failure 27, 617965 (3), Autosomal recessive |
| *AKAP10* | 604694 | {Cardiac conduction defect, susceptibility to}, 115080 (3), Autosomal dominant |
| *AKAP9* | 604001 | ?Long QT syndrome 11, 611820 (3), Autosomal dominant |
| *AKR1C2* | 600450 | 46XY sex reversal 8, 614279 (3), Autosomal recessive |
| *AKR1C4* | 600451 | {46XY sex reversal 8, modifier of}, 614279 (3), Autosomal recessive |
| *AKR1D1* | 604741 | Bile acid synthesis defect, congenital, 2, 235555 (3), Autosomal recessive |
| *AKT1* | 164730 | Breast cancer, somatic, 114480 (3); Cowden syndrome 6, 615109 (3); Colorectal cancer, somatic, 114500 (3); Proteus syndrome, somatic, 176920 (3); Ovarian cancer, somatic, 167000 (3) |
| *AKT2* | 164731 | Diabetes mellitus, type II, 125853 (3), Autosomal dominant; Hypoinsulinemic hypoglycemia with hemihypertrophy, 240900 (3), Autosomal dominant |
| *AKT3* | 611223 | Megalencephaly-polymicrogyria-polydactyly-hydrocephalus syndrome 2, 615937 (3), Autosomal dominant |
| *ALAD* | 125270 | Porphyria, acute hepatic, 612740 (3), Autosomal recessive; {Lead poisoning, susceptibility to}, 612740 (3), Autosomal recessive |
| *ALAS2* | 301300 | Anemia, sideroblastic, 1, 300751 (3), X-linked recessive; Protoporphyria, erythropoietic, X-linked, 300752 (3), X-linked |
| *ALB* | 103600 | ?[Dysalbuminemic hypertriiodothyroninemia], 615999 (3), Autosomal recessive, Autosomal dominant; Analbuminemia, 616000 (3), Autosomal recessive; [Dysalbuminemic hyperthyroxinemia], 615999 (3), Autosomal recessive, Autosomal dominant |
| *ALDH18A1* | 138250 | Spastic paraplegia 9A, autosomal dominant, 601162 (3), Autosomal dominant; Cutis laxa, autosomal recessive, type IIIA, 219150 (3), Autosomal recessive; Spastic paraplegia 9B, autosomal recessive, 616586 (3), Autosomal recessive; Cutis laxa, autosomal dominant 3, 616603 (3), Autosomal dominant |
| *ALDH1A3* | 600463 | Microphthalmia, isolated 8, 615113 (3), Autosomal recessive |
| *ALDH2* | 100650 | {Esophageal cancer, alcohol-related, susceptibility to} (3); {Sublingual nitroglycerin, susceptibility to poor response to} (3); Alcohol sensitivity, acute, 610251 (3), Autosomal dominant; {Hangover, susceptibility to}, 610251 (3), Autosomal dominant |
| *ALDH3A2* | 609523 | Sjogren-Larsson syndrome, 270200 (3), Autosomal recessive |
| *ALDH4A1* | 606811 | Hyperprolinemia, type II, 239510 (3), Autosomal recessive |
| *ALDH5A1* | 610045 | Succinic semialdehyde dehydrogenase deficiency, 271980 (3), Autosomal recessive |
| *ALDH6A1* | 603178 | Methylmalonate semialdehyde dehydrogenase deficiency, 614105 (3), Autosomal recessive |
| *ALDH7A1* | 107323 | Epilepsy, pyridoxine-dependent, 266100 (3), Autosomal recessive |
| *ALDOA* | 103850 | Glycogen storage disease XII, 611881 (3), Autosomal recessive |
| *ALDOB* | 612724 | Fructose intolerance, hereditary, 229600 (3), Autosomal recessive |
| *ALG1* | 605907 | Congenital disorder of glycosylation, type Ik, 608540 (3), Autosomal recessive |
| *ALG10B* | 603313 | {Long QT syndrome, acquired, reduced susceptibility to}, 613688 (3), Autosomal dominant |
| *ALG11* | 613666 | Congenital disorder of glycosylation, type Ip, 613661 (3), Autosomal recessive |
| *ALG12* | 607144 | Congenital disorder of glycosylation, type Ig, 607143 (3), Autosomal recessive |
| *ALG13* | 300776 | ?Congenital disorder of glycosylation, type Is, 300884 (3), X-linked; Developmental and epileptic encephalopathy 36, 300884 (3), X-linked |
| *ALG14* | 612866 | Intellectual developmental disorder with epilepsy, behavioral abnormalities, and coarse facies, 619031 (3); Myopathy, epilepsy, and progressive cerebral atrophy, 619036 (3), Autosomal recessive; ?Myasthenic syndrome, congenital, 15, without tubular aggregates, 616227 (3), Autosomal recessive |
| *ALG2* | 607905 | ?Congenital disorder of glycosylation, type Ii, 607906 (3), Autosomal recessive; Myasthenic syndrome, congenital, 14, with tubular aggregates, 616228 (3), Autosomal recessive |
| *ALG3* | 608750 | Congenital disorder of glycosylation, type Id, 601110 (3), Autosomal recessive |
| *ALG6* | 604566 | Congenital disorder of glycosylation, type Ic, 603147 (3), Autosomal recessive |
| *ALG8* | 608103 | Congenital disorder of glycosylation, type Ih, 608104 (3), Autosomal recessive; Polycystic liver disease 3 with or without kidney cysts, 617874 (3), Autosomal dominant |
| *ALG9* | 606941 | Gillessen-Kaesbach-Nishimura syndrome, 263210 (3), Autosomal recessive; Congenital disorder of glycosylation, type Il, 608776 (3), Autosomal recessive |
| *ALK* | 105590 | {Neuroblastoma, susceptibility to, 3}, 613014 (3) |
| *ALKBH8* | 613306 | Intellectual developmental disorder, autosomal recessive 71, 618504 (3), Autosomal recessive |
| *ALMS1* | 606844 | Alstrom syndrome, 203800 (3), Autosomal recessive |
| *ALOX12B* | 603741 | Ichthyosis, congenital, autosomal recessive 2, 242100 (3), Autosomal recessive |
| *ALOX5* | 152390 | {Atherosclerosis, susceptibility to} (3); {Asthma, diminished response to antileukotriene treatment in}, 600807 (3), Autosomal dominant |
| *ALOX5AP* | 603700 | {Stroke, susceptibility to}, 601367 (3), Multifactorial |
| *ALOXE3* | 607206 | Ichthyosis, congenital, autosomal recessive 3, 606545 (3), Autosomal recessive |
| *ALPK1* | 607347 | ROSAH syndrome, 614979 (3), Autosomal dominant |
| *ALPK3* | 617608 | Cardiomyopathy, familial hypertrophic 27, 618052 (3), Autosomal recessive |
| *ALPL* | 171760 | Odontohypophosphatasia, 146300 (3), Autosomal recessive, Autosomal dominant; Hypophosphatasia, infantile, 241500 (3), Autosomal recessive; Hypophosphatasia, childhood, 241510 (3), Autosomal recessive; Hypophosphatasia, adult, 146300 (3), Autosomal recessive, Autosomal dominant |
| *ALS2* | 606352 | Primary lateral sclerosis, juvenile, 606353 (3), Autosomal recessive; Spastic paralysis, infantile onset ascending, 607225 (3), Autosomal recessive; Amyotrophic lateral sclerosis 2, juvenile, 205100 (3), Autosomal recessive |
| *ALX1* | 601527 | Frontonasal dysplasia 3, 613456 (3), Autosomal recessive |
| *ALX3* | 606014 | Frontonasal dysplasia 1, 136760 (3), Autosomal recessive |
| *ALX4* | 605420 | Parietal foramina 2, 609597 (3), Autosomal dominant; {Craniosynostosis 5, susceptibility to}, 615529 (3), Autosomal dominant; Frontonasal dysplasia 2, 613451 (3), Autosomal recessive |
| *AMACR* | 604489 | Alpha-methylacyl-CoA racemase deficiency, 614307 (3), Autosomal recessive; Bile acid synthesis defect, congenital, 4, 214950 (3), Autosomal recessive |
| *AMBN* | 601259 | Amelogenesis imperfecta, type IF, 616270 (3), Autosomal recessive |
| *AMELX* | 300391 | Amelogenesis imperfecta, type 1E, 301200 (3), X-linked dominant |
| *AMER1* | 300647 | Osteopathia striata with cranial sclerosis, 300373 (3), X-linked dominant |
| *AMH* | 600957 | Persistent Mullerian duct syndrome, type I, 261550 (3), Autosomal recessive |
| *AMHR2* | 600956 | Persistent Mullerian duct syndrome, type II, 261550 (3), Autosomal recessive |
| *AMMECR1* | 300195 | Midface hypoplasia, hearing impairment, elliptocytosis, and nephrocalcinosis, 300990 (3), X-linked recessive |
| *AMN* | 605799 | Imerslund-Grasbeck syndrome 2, 618882 (3), Autosomal recessive |
| *AMPD1* | 102770 | Myopathy due to myoadenylate deaminase deficiency, 615511 (3), Autosomal recessive |
| *AMPD2* | 102771 | ?Spastic paraplegia 63, 615686 (3), Autosomal recessive; Pontocerebellar hypoplasia, type 9, 615809 (3), Autosomal recessive |
| *AMPD3* | 102772 | [AMP deaminase deficiency, erythrocytic], 612874 (3), Autosomal recessive |
| *AMT* | 238310 | Glycine encephalopathy, 605899 (3), Autosomal recessive |
| *AMTN* | 610912 | ?Amelogenesis imperfecta, type IIIB, 617607 (3), Autosomal dominant |
| *ANAPC1* | 608473 | Rothmund-Thomson syndrome, type 1, 618625 (3), Autosomal recessive |
| *ANG* | 105850 | Amyotrophic lateral sclerosis 9, 611895 (3) |
| *ANGPT1* | 601667 | ?Angioedema, hereditary, 5, 619361 (3), Autosomal dominant |
| *ANGPT2* | 601922 | Lymphatic malformation 10, 619369 (3), Autosomal dominant |
| *ANGPTL3* | 604774 | Hypobetalipoproteinemia, familial, 2, 605019 (3), Autosomal recessive |
| *ANGPTL4* | 605910 | Plasma triglyceride level QTL, low, 615881 (3), Autosomal dominant |
| *ANK1* | 612641 | Spherocytosis, type 1, 182900 (3), Autosomal recessive, Autosomal dominant |
| *ANK2* | 106410 | Long QT syndrome 4, 600919 (3), Autosomal dominant; Cardiac arrhythmia, ankyrin-B-related, 600919 (3), Autosomal dominant |
| *ANK3* | 600465 | Mental retardation, autosomal recessive, 37, 615493 (3), Autosomal recessive |
| *ANKH* | 605145 | Chondrocalcinosis 2, 118600 (3), Autosomal dominant; Craniometaphyseal dysplasia, 123000 (3), Autosomal dominant |
| *ANKLE2* | 616062 | Microcephaly 16, primary, autosomal recessive, 616681 (3), Autosomal recessive |
| *ANKRD11* | 611192 | KBG syndrome, 148050 (3), Autosomal dominant |
| *ANKRD26* | 610855 | Thrombocytopenia 2, 188000 (3), Autosomal dominant |
| *ANKS6* | 615370 | Nephronophthisis 16, 615382 (3), Autosomal recessive |
| *ANLN* | 616027 | Focal segmental glomerulosclerosis 8, 616032 (3), Autosomal dominant |
| *ANO10* | 613726 | Spinocerebellar ataxia, autosomal recessive 10, 613728 (3), Autosomal recessive |
| *ANO3* | 610110 | Dystonia 24, 615034 (3), Autosomal dominant |
| *ANO5* | 608662 | Muscular dystrophy, limb-girdle, autosomal recessive 12, 611307 (3), Autosomal recessive; Miyoshi muscular dystrophy 3, 613319 (3), Autosomal recessive; Gnathodiaphyseal dysplasia, 166260 (3), Autosomal dominant |
| *ANO6* | 608663 | Scott syndrome, 262890 (3), Autosomal recessive |
| *ANOS1* | 300836 | Hypogonadotropic hypogonadism 1 with or without anosmia (Kallmann syndrome 1), 308700 (3), X-linked recessive |
| *ANTXR1* | 606410 | GAPO syndrome, 230740 (3), Autosomal recessive; {?Hemangioma, capillary infantile, susceptibility to}, 602089 (3), Autosomal dominant |
| *ANTXR2* | 608041 | Hyaline fibromatosis syndrome, 228600 (3), Autosomal recessive |
| *ANXA11* | 602572 | Amyotrophic lateral sclerosis 23, 617839 (3), Autosomal dominant |
| *ANXA5* | 131230 | {Pregnancy loss, recurrent, susceptibility to, 3}, 614391 (3), Autosomal dominant |
| *AP1B1* | 600157 | Keratitis-ichthyosis-deafness syndrome, autosomal recessive, 242150 (3), Autosomal recessive |
| *AP1S1* | 603531 | MEDNIK syndrome, 609313 (3), Autosomal recessive |
| *AP1S2* | 300629 | Mental retardation, X-linked syndromic 5, 304340 (3), X-linked recessive |
| *AP1S3* | 615781 | {Psoriasis 15, pustular, susceptibility to}, 616106 (3), Autosomal dominant |
| *AP2M1* | 601024 | Intellectual developmental disorder 60 with seizures, 618587 (3), Autosomal dominant |
| *AP2S1* | 602242 | Hypocalciuric hypercalcemia, type III, 600740 (3), Autosomal dominant |
| *AP3B1* | 603401 | Hermansky-Pudlak syndrome 2, 608233 (3), Autosomal recessive |
| *AP3B2* | 602166 | Developmental and epileptic encephalopathy 48, 617276 (3), Autosomal recessive |
| *AP3D1* | 607246 | ?Hermansky-Pudlak syndrome 10, 617050 (3), Autosomal recessive |
| *AP4B1* | 607245 | Spastic paraplegia 47, autosomal recessive, 614066 (3), Autosomal recessive |
| *AP4E1* | 607244 | Stuttering, familial persistent, 1, 184450 (3), Autosomal dominant; Spastic paraplegia 51, autosomal recessive, 613744 (3), Autosomal recessive |
| *AP4M1* | 602296 | Spastic paraplegia 50, autosomal recessive, 612936 (3), Autosomal recessive |
| *AP4S1* | 607243 | Spastic paraplegia 52, autosomal recessive, 614067 (3), Autosomal recessive |
| *AP5Z1* | 613653 | Spastic paraplegia 48, autosomal recessive, 613647 (3), Autosomal recessive |
| *APC* | 611731 | Colorectal cancer, somatic, 114500 (3); Brain tumor-polyposis syndrome 2, 175100 (3), Autosomal dominant; Desmoid disease, hereditary, 135290 (3), Autosomal dominant; Adenoma, periampullary, somatic, 175100 (3); Hepatoblastoma, somatic, 114550 (3); Gastric cancer, somatic, 613659 (3); Gastric adenocarcinoma and proximal polyposis of the stomach, 619182 (3), Autosomal dominant; Gardner syndrome, 175100 (3), Autosomal dominant; Adenomatous polyposis coli, 175100 (3), Autosomal dominant |
| *APC2* | 612034 | Cortical dysplasia, complex, with other brain malformations 10, 618677 (3), Autosomal recessive; ?Sotos syndrome 3, 617169 (3), Autosomal recessive |
| *APCDD1* | 607479 | Hypotrichosis 1, 605389 (3), Autosomal dominant |
| *APCS* | 104770 | {?Amyloidosis, secondary, susceptibility to} (1) |
| *APOA1* | 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) |
| *APOA2* | 107670 | Apolipoprotein A-II deficiency (3); {Hypercholesterolemia, familial, modifier of}, 143890 (3), Autosomal recessive, Autosomal dominant |
| *APOA5* | 606368 | Hyperchylomicronemia, late-onset, 144650 (3), Autosomal dominant; {Hypertriglyceridemia, susceptibility to}, 145750 (3), Autosomal dominant |
| *APOB* | 107730 | Hypercholesterolemia, familial, 2, 144010 (3), Autosomal dominant; Hypobetalipoproteinemia, 615558 (3), Autosomal recessive |
| *APOC2* | 608083 | Hyperlipoproteinemia, type Ib, 207750 (3), Autosomal recessive |
| *APOC3* | 107720 | Apolipoprotein C-III deficiency, 614028 (3) |
| *APOE* | 107741 | Alzheimer disease 2, 104310 (3), Autosomal dominant; Sea-blue histiocyte disease, 269600 (3), Autosomal recessive; {?Alzheimer disease, protection against, due to APOE3-Christchurch}, 607822 (3), Autosomal dominant; {Coronary artery disease, severe, susceptibility to}, 617347 (3); Lipoprotein glomerulopathy, 611771 (3); {?Macular degeneration, age-related}, 603075 (3), Autosomal dominant; Hyperlipoproteinemia, type III, 617347 (3) |
| *APOL1* | 603743 | {End-stage renal disease, nondiabetic, susceptibility to}, 612551 (3); {Glomerulosclerosis, focal segmental, 4, susceptibility to}, 612551 (3) |
| *APOL2* | 607252 | {Schizophrenia}, 181500 (1), Autosomal dominant |
| *APOL4* | 607254 | {Schizophrenia}, 181500 (1), Autosomal dominant |
| *APP* | 104760 | Cerebral amyloid angiopathy, Dutch, Italian, Iowa, Flemish, Arctic variants, 605714 (3), Autosomal dominant; Alzheimer disease 1, familial, 104300 (3), Autosomal dominant |
| *APPL1* | 604299 | {Maturity-onset diabetes of the young, type 14}, 616511 (3), Autosomal dominant |
| *APRT* | 102600 | Adenine phosphoribosyltransferase deficiency, 614723 (3), Autosomal recessive |
| *APTX* | 606350 | Ataxia, early-onset, with oculomotor apraxia and hypoalbuminemia, 208920 (3), Autosomal recessive |
| *AQP1* | 107776 | [Aquaporin-1 deficiency], 110450 (3); [Blood group, Colton], 110450 (3) |
| *AQP2* | 107777 | Diabetes insipidus, nephrogenic, 2, 125800 (3), Autosomal recessive, Autosomal dominant |
| *AQP3* | 600170 | [Blood group GIL], 607457 (3) |
| *AQP5* | 600442 | Palmoplantar keratoderma, Bothnian type, 600231 (3), Autosomal dominant |
| *AQP7* | 602974 | [Glycerol quantitative trait locus], 614411 (3), Autosomal recessive |
| *AR* | 313700 | Androgen insensitivity, partial, with or without breast cancer, 312300 (3), X-linked recessive; {Prostate cancer, susceptibility to}, 176807 (3), Somatic mutation, Autosomal dominant; Androgen insensitivity, 300068 (3), X-linked recessive; Spinal and bulbar muscular atrophy of Kennedy, 313200 (3), X-linked recessive; Hypospadias 1, X-linked, 300633 (3), X-linked recessive |
| *ARCN1* | 600820 | Short stature, rhizomelic, with microcephaly, micrognathia, and developmental delay, 617164 (3), Autosomal dominant |
| *ARF1* | 103180 | Periventricular nodular heterotopia 8, 618185 (3), Autosomal dominant |
| *ARFGEF2* | 605371 | Periventricular heterotopia with microcephaly, 608097 (3), Autosomal recessive |
| *ARG1* | 608313 | Argininemia, 207800 (3), Autosomal recessive |
| *ARHGAP26* | 605370 | Leukemia, juvenile myelomonocytic, somatic, 607785 (3) |
| *ARHGAP31* | 610911 | Adams-Oliver syndrome 1, 100300 (3), Autosomal dominant |
| *ARHGDIA* | 601925 | Nephrotic syndrome, type 8, 615244 (3), Autosomal recessive |
| *ARHGEF1* | 601855 | ?Immunodeficiency 62, 618459 (3), Autosomal recessive |
| *ARHGEF10* | 608136 | ?Slowed nerve conduction velocity, AD, 608236 (3), Autosomal dominant |
| *ARHGEF18* | 616432 | Retinitis pigmentosa 78, 617433 (3), Autosomal recessive |
| *ARHGEF2* | 607560 | ?Neurodevelopmental disorder with midbrain and hindbrain malformations, 617523 (3), Autosomal recessive |
| *ARHGEF6* | 300267/300436 | -/Mental retardation, X-linked 46, 300436 (2), X-linked recessive |
| *ARHGEF9* | 300429 | Developmental and epileptic encephalopathy 8, 300607 (3), X-linked |
| *ARID1A* | 603024 | Coffin-Siris syndrome 2, 614607 (3), Autosomal dominant |
| *ARID1B* | 614556 | Coffin-Siris syndrome 1, 135900 (3), Autosomal dominant |
| *ARID2* | 609539 | Coffin-Siris syndrome 6, 617808 (3), Autosomal dominant |
| *ARL13B* | 608922 | Joubert syndrome 8, 612291 (3), Autosomal recessive |
| *ARL2* | 601175 | ?Microcornea, rod-cone dystrophy, cataract, and posterior staphyloma 1, 619082 (3), Autosomal dominant |
| *ARL2BP* | 615407 | Retinitis pigmentosa with or without situs inversus, 615434 (3), Autosomal recessive |
| *ARL3* | 604695 | Retinitis pigmentosa 83, 618173 (3), Autosomal dominant; Joubert syndrome 35, 618161 (3), Autosomal recessive |
| *ARL6* | 608845 | Retinitis pigmentosa 55, 613575 (3), Autosomal recessive; {Bardet-Biedl syndrome 1, modifier of}, 209900 (3), Autosomal recessive, Digenic recessive; Bardet-Biedl syndrome 3, 600151 (3), Autosomal recessive |
| *ARL6IP1* | 607669 | ?Spastic paraplegia 61, autosomal recessive, 615685 (3), Autosomal recessive |
| *ARMC2* | 618424 | Spermatogenic failure 38, 618433 (3), Autosomal recessive |
| *ARMC5* | 615549 | ACTH-independent macronodular adrenal hyperplasia 2, 615954 (3), Somatic mutation, Autosomal dominant |
| *ARMC9* | 617612 | Joubert syndrome 30, 617622 (3), Autosomal recessive |
| *ARMS2* | 611313 | {Macular degeneration, age-related, 8}, 613778 (3) |
| *ARNT2* | 606036 | ?Webb-Dattani syndrome, 615926 (3), Autosomal recessive |
| *ARPC1B* | 604223 | Immunodeficiency 71 with inflammatory disease and congenital thrombocytopenia, 617718 (3), Autosomal recessive |
| *ARR3* | 301770 | Myopia 26, X-linked, female-limited, 301010 (3), X-linked |
| *ARSA* | 607574 | Metachromatic leukodystrophy, 250100 (3), Autosomal recessive |
| *ARSB* | 611542 | Mucopolysaccharidosis type VI (Maroteaux-Lamy), 253200 (3), Autosomal recessive |
| *ARSG* | 610008 | Usher syndrome, type IV, 618144 (3), Autosomal recessive |
| *ARSL (ARSE)* | 300180 | Chondrodysplasia punctata, X-linked recessive, 302950 (3), X-linked recessive |
| *ART4* | 110600 | [Blood group, Dombrock], 616060 (3) |
| *ARV1* | 611647 | Developmental and epileptic encephalopathy 38, 617020 (3), Autosomal recessive |
| *ARX* | 300382 | Proud syndrome, 300004 (3), X-linked; Hydranencephaly with abnormal genitalia, 300215 (3), X-linked; Partington syndrome, 309510 (3), X-linked recessive; Developmental and epileptic encephalopathy 1, 308350 (3), X-linked recessive; Lissencephaly, X-linked 2, 300215 (3), X-linked; Mental retardation, X-linked 29 and others, 300419 (3), X-linked recessive |
| *ASAH1* | 613468 | Spinal muscular atrophy with progressive myoclonic epilepsy, 159950 (3), Autosomal recessive; Farber lipogranulomatosis, 228000 (3), Autosomal recessive |
| *ASB10* | 615054 | Glaucoma 1, open angle, F, 603383 (3), Autosomal dominant |
| *ASCC1* | 614215 | Spinal muscular atrophy with congenital bone fractures 2, 616867 (3), Autosomal recessive; Barrett esophagus/esophageal adenocarcinoma, 614266 (3) |
| *ASCL1* | 100790 | Haddad syndrome, 209880 (3), Autosomal dominant; Central hypoventilation syndrome, congenital, 209880 (3), Autosomal dominant |
| *ASH1L* | 607999 | Mental retardation, autosomal dominant 52, 617796 (3), Autosomal dominant |
| *ASIP* | 600201 | [Skin/hair/eye pigmentation 9, brown/nonbrown eyes], 611742 (3); [Skin/hair/eye pigmentation 9, dark/light hair], 611742 (3) |
| *ASL* | 608310 | Argininosuccinic aciduria, 207900 (3), Autosomal recessive |
| *ASNS* | 108370 | Asparagine synthetase deficiency, 615574 (3), Autosomal recessive |
| *ASPA* | 608034 | Canavan disease, 271900 (3), Autosomal recessive |
| *ASPH* | 600582 | Traboulsi syndrome, 601552 (3), Autosomal recessive |
| *ASPM* | 605481 | Microcephaly 5, primary, autosomal recessive, 608716 (3), Autosomal recessive |
| *ASPN* | 608135 | {Lumbar disc degeneration}, 603932 (3); {Osteoarthritis susceptibility 3}, 607850 (3), Autosomal dominant |
| *ASPRV1* | 611765 | Ichthyosis, lamellar, autosomal dominant, 146750 (3), Autosomal dominant |
| *ASPSCR1* | 606236 | Alveolar soft-part sarcoma, 606243 (3) |
| *ASS1* | 603470 | Citrullinemia, 215700 (3), Autosomal recessive |
| *ASXL1* | 612990 | Myelodysplastic syndrome, somatic, 614286 (3); Bohring-Opitz syndrome, 605039 (3), Autosomal dominant |
| *ASXL2* | 612991 | Shashi-Pena syndrome, 617190 (3), Autosomal dominant |
| *ASXL3* | 615115 | Bainbridge-Ropers syndrome, 615485 (3), Autosomal dominant |
| *ATAD1* | 614452 | Hyperekplexia 4, 618011 (3), Autosomal recessive |
| *ATAD3A* | 612316 | Harel-Yoon syndrome, 617183 (3), Autosomal recessive, Autosomal dominant; Pontocerebellar hypoplasia, hypotonia, and respiratory insufficiency syndrome, neonatal lethal, 618810 (3), Autosomal recessive |
| *ATCAY* | 608179 | Ataxia, cerebellar, Cayman type, 601238 (3), Autosomal recessive |
| *ATF6* | 605537 | Achromatopsia 7, 616517 (3), Autosomal recessive |
| *ATG16L1* | 610767 | {Inflammatory bowel disease (Crohn disease) 10}, 611081 (3) |
| *ATG5* | 604261 | ?Spinocerebellar ataxia, autosomal recessive 25, 617584 (3), Autosomal recessive |
| *ATG7* | 608760 | Spinocerebellar ataxia, autosomal recessive 31, 619422 (3), Autosomal recessive |
| *ATIC* | 601731 | AICA-ribosiduria due to ATIC deficiency, 608688 (3), Autosomal recessive |
| *ATL1* | 606439 | Spastic paraplegia 3A, autosomal dominant, 182600 (3), Autosomal dominant; Neuropathy, hereditary sensory, type ID, 613708 (3), Autosomal dominant |
| *ATL3* | 609369 | Neuropathy, hereditary sensory, type IF, 615632 (3), Autosomal dominant |
| *ATM* | 607585 | Lymphoma, B-cell non-Hodgkin, somatic (3); Ataxia-telangiectasia, 208900 (3), Autosomal recessive; {Breast cancer, susceptibility to}, 114480 (3), Somatic mutation, Autosomal dominant; T-cell prolymphocytic leukemia, somatic (3); Lymphoma, mantle cell, somatic (3) |
| *ATN1* | 607462 | Dentatorubral-pallidoluysian atrophy, 125370 (3), Autosomal dominant; Congenital hypotonia, epilepsy, developmental delay, and digital anomalies, 618494 (3), Autosomal dominant |
| *ATOH7* | 609875 | Persistent hyperplastic primary vitreous, autosomal recessive, 221900 (3), Autosomal recessive |
| *ATP11C* | 300516 | ?Hemolytic anemia, congenital, X-linked, 301015 (3), X-linked recessive |
| *ATP13A2* | 610513 | Spastic paraplegia 78, autosomal recessive, 617225 (3), Autosomal recessive; Kufor-Rakeb syndrome, 606693 (3), Autosomal recessive |
| *ATP1A1* | 182310 | Hypomagnesemia, seizures, and mental retardation 2, 618314 (3), Autosomal dominant; Charcot-Marie-Tooth disease, axonal, type 2DD, 618036 (3), Autosomal dominant |
| *ATP1A2* | 182340 | Alternating hemiplegia of childhood 1, 104290 (3), Autosomal dominant; Migraine, familial basilar, 602481 (3), Autosomal dominant; Migraine, familial hemiplegic, 2, 602481 (3), Autosomal dominant |
| *ATP1A3* | 182350 | Alternating hemiplegia of childhood 2, 614820 (3), Autosomal dominant; Dystonia-12, 128235 (3), Autosomal dominant; CAPOS syndrome, 601338 (3), Autosomal dominant |
| *ATP1B1* | 182330 | [Blood pressure regulation QTL], 145500 (2), Multifactorial |
| *ATP2A1* | 108730 | Brody myopathy, 601003 (3), Autosomal recessive |
| *ATP2A2* | 108740 | Acrokeratosis verruciformis, 101900 (3), Autosomal dominant; Darier disease, 124200 (3), Autosomal dominant |
| *ATP2B2* | 108733 | {Deafness, autosomal recessive 12, modifier of}, 601386 (3), Autosomal recessive |
| *ATP2B3* | 300014 | ?Spinocerebellar ataxia, X-linked 1, 302500 (3), X-linked recessive |
| *ATP2C1* | 604384 | Hailey-Hailey disease, 169600 (3), Autosomal dominant |
| *ATP5F1A* | 164360 | ?Mitochondrial complex V (ATP synthase) deficiency, nuclear type 4, 615228 (3), Autosomal recessive; ?Combined oxidative phosphorylation deficiency 22, 616045 (3), Autosomal recessive |
| *ATP5F1D* | 603150 | Mitochondrial complex V (ATP synthase) deficiency, 618120 (3), Autosomal recessive |
| *ATP5F1E* | 606153 | ?Mitochondrial complex V (ATP synthase) deficiency, nuclear type 3, 614053 (3), Autosomal recessive |
| *ATP5MK (ATP5MD)* | 615204 | Mitochondrial complex V (ATP synthase) deficiency, nuclear type 6, 618683 (3), Autosomal recessive |
| *ATP6AP1* | 300197 | Immunodeficiency 47, 300972 (3), X-linked recessive |
| *ATP6AP2* | 300556 | ?Parkinsonism with spasticity, X-linked, 300911 (3), X-linked recessive; Congenital disorder of glycosylation, type IIr, 301045 (3), X-linked recessive; Mental retardation, X-linked, syndromic, Hedera type, 300423 (3), X-linked recessive |
| *ATP6V0A2* | 611716 | Wrinkly skin syndrome, 278250 (3), Autosomal recessive; Cutis laxa, autosomal recessive, type IIA, 219200 (3), Autosomal recessive |
| *ATP6V0A4* | 605239 | Distal renal tubular acidosis 3, with or without sensorineural hearing loss, 602722 (3), Autosomal recessive |
| *ATP6V1A* | 607027 | Cutis laxa, autosomal recessive, type IID, 617403 (3), Autosomal recessive; Developmental and epileptic encephalopathy 93, 618012 (3), Autosomal dominant |
| *ATP6V1B1* | 192132 | Distal renal tubular acidosis 2 with progressive sensorineural hearing loss, 267300 (3), Autosomal recessive |
| *ATP6V1B2* | 606939 | Zimmermann-Laband syndrome 2, 616455 (3), Autosomal dominant; Deafness, congenital, with onychodystrophy, autosomal dominant, 124480 (3), Autosomal dominant |
| *ATP6V1E1* | 108746 | Cutis laxa, autosomal recessive, type IIC, 617402 (3), Autosomal recessive |
| *ATP7A* | 300011 | Occipital horn syndrome, 304150 (3), X-linked recessive; Spinal muscular atrophy, distal, X-linked 3, 300489 (3), X-linked recessive; Menkes disease, 309400 (3), X-linked recessive |
| *ATP7B* | 606882 | Wilson disease, 277900 (3), Autosomal recessive |
| *ATP8A2* | 605870 | ?Cerebellar ataxia, mental retardation, and dysequilibrium syndrome 4, 615268 (3), Autosomal recessive |
| *ATP8B1* | 602397 | Cholestasis, progressive familial intrahepatic 1, 211600 (3), Autosomal recessive; Cholestasis, intrahepatic, of pregnancy, 1, 147480 (3), Autosomal dominant; Cholestasis, benign recurrent intrahepatic, 243300 (3), Autosomal recessive |
| *ATPAF2* | 608918 | ?Mitochondrial complex V (ATP synthase) deficiency, nuclear type 1, 604273 (3), Autosomal recessive |
| *ATR* | 601215 | Seckel syndrome 1, 210600 (3), Autosomal recessive; ?Cutaneous telangiectasia and cancer syndrome, familial, 614564 (3), Autosomal dominant |
| *ATRX* | 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 |
| *ATXN1* | 601556 | Spinocerebellar ataxia 1, 164400 (3), Autosomal dominant |
| *ATXN10* | 611150 | Spinocerebellar ataxia 10, 603516 (3), Autosomal dominant |
| *ATXN2* | 601517 | {Amyotrophic lateral sclerosis, susceptibility to, 13}, 183090 (3), Autosomal dominant; Spinocerebellar ataxia 2, 183090 (3), Autosomal dominant; {Parkinson disease, late-onset, susceptibility to}, 168600 (3), Multifactorial, Autosomal dominant |
| *ATXN3* | 607047 | Machado-Joseph disease, 109150 (3), Autosomal dominant |
| *ATXN7* | 607640 | Spinocerebellar ataxia 7, 164500 (3), Autosomal dominant |
| *AUH* | 600529 | 3-methylglutaconic aciduria, type I, 250950 (3), Autosomal recessive |
| *AURKA* | 603072 | {Colon cancer, susceptibility to}, 114500 (3), Somatic mutation, Autosomal dominant |
| *AURKC* | 603495 | Spermatogenic failure 5, 243060 (3), Autosomal recessive |
| *AUTS2* | 607270 | Mental retardation, autosomal dominant 26, 615834 (3), Autosomal dominant |
| *AVIL* | 613397 | Nephrotic syndrome, type 21, 618594 (3), Autosomal recessive |
| *AVP* | 192340 | Diabetes insipidus, neurohypophyseal, 125700 (3), Autosomal dominant |
| *AVPR2* | 300538 | Diabetes insipidus, nephrogenic, 1, 304800 (3), X-linked recessive; Nephrogenic syndrome of inappropriate antidiuresis, 300539 (3), X-linked recessive |
| *AXIN1* | 603816 | Hepatocellular carcinoma, somatic, 114550 (3); ?Caudal duplication anomaly, 607864 (3) |
| *AXIN2* | 604025 | Colorectal cancer, somatic, 114500 (3); Oligodontia-colorectal cancer syndrome, 608615 (3), Autosomal dominant |
| *B2M* | 109700 | ?Amyloidosis, familial visceral, 105200 (3), Autosomal dominant; Immunodeficiency 43, 241600 (3), Autosomal recessive |
| *B3GALNT1* | 603094 | [Blood group, P1PK system, P(k) phenotype], 111400 (3); [Blood group, globoside system], 615021 (3) |
| *B3GALNT2* | 610194 | Muscular dystrophy-dystroglycanopathy (congenital with brain and eye anomalies, type A, 11, 615181 (3), Autosomal recessive |
| *B3GALT6* | 615291 | Ehlers-Danlos syndrome, spondylodysplastic type, 2, 615349 (3), Autosomal recessive; Spondyloepimetaphyseal dysplasia with joint laxity, type 1, with or without fractures, 271640 (3), Autosomal recessive; Al-Gazali syndrome, 609465 (3) |
| *B3GAT3* | 606374 | Multiple joint dislocations, short stature, craniofacial dysmorphism, with or without congenital heart defects, 245600 (3), Autosomal recessive |
| *B3GLCT* | 610308 | Peters-plus syndrome, 261540 (3), Autosomal recessive |
| *B4GALNT1* | 601873 | Spastic paraplegia 26, autosomal recessive, 609195 (3), Autosomal recessive |
| *B4GALT1* | 137060 | Congenital disorder of glycosylation, type IId, 607091 (3), Autosomal recessive |
| *B4GALT7* | 604327 | Ehlers-Danlos syndrome, spondylodysplastic type, 1, 130070 (3), Autosomal recessive |
| *B4GAT1* | 605517 | Muscular dystrophy-dystroglycanopathy (congenital with brain and eye anomalies), type A, 13, 615287 (3), Autosomal recessive |
| *B9D1* | 614144 | ?Meckel syndrome 9, 614209 (3), Autosomal recessive; Joubert syndrome 27, 617120 (3), Autosomal recessive |
| *B9D2* | 611951 | ?Meckel syndrome 10, 614175 (3), Autosomal recessive; Joubert syndrome 34, 614175 (3), Autosomal recessive |
| *BAAT* | 602938 | Hypercholanemia, familial, 607748 (3), Autosomal recessive; Bile acid conjugation defect 1, 619232 (3), Autosomal recessive |
| *BACH2* | 605394 | Immunodeficiency 60, 618394 (3), Autosomal dominant |
| *BAG3* | 603883 | Cardiomyopathy, dilated, 1HH, 613881 (3), Autosomal dominant; Myopathy, myofibrillar, 6, 612954 (3), Autosomal dominant |
| *BANF1* | 603811 | Nestor-Guillermo progeria syndrome, 614008 (3), Autosomal recessive |
| *BAP1* | 603089 | Tumor predisposition syndrome, 614327 (3), Autosomal dominant |
| *BARD1* | 601593 | {Breast cancer, susceptibility to}, 114480 (3), Somatic mutation, Autosomal dominant |
| *BAX* | 600040 | Colorectal cancer, somatic, 114500 (3); T-cell acute lymphoblastic leukemia, somatic, 613065 (3) |
| *BBIP1* | 613605 | ?Bardet-Biedl syndrome 18, 615995 (3), Autosomal recessive |
| *BBS1* | 209901 | Bardet-Biedl syndrome 1, 209900 (3), Autosomal recessive, Digenic recessive |
| *BBS10* | 610148 | Bardet-Biedl syndrome 10, 615987 (3), Autosomal recessive |
| *BBS12* | 610683 | Bardet-Biedl syndrome 12, 615989 (3), Autosomal recessive |
| *BBS2* | 606151 | Retinitis pigmentosa 74, 616562 (3), Autosomal recessive; Bardet-Biedl syndrome 2, 615981 (3), Autosomal recessive |
| *BBS4* | 600374 | Bardet-Biedl syndrome 4, 615982 (3), Autosomal recessive |
| *BBS5* | 603650 | Bardet-Biedl syndrome 5, 615983 (3), Autosomal recessive |
| *BBS7* | 607590 | Bardet-Biedl syndrome 7, 615984 (3), Autosomal recessive |
| *BBS9* | 607968 | Bardet-Biedl syndrome 9, 615986 (3), Autosomal recessive |
| *BCAM* | 612773 | [Blood group, Lutheran system], 111200 (3); [Blood group, Auberger system], 111200 (3); [Blood group, Lutheran null], 247420 (3), Autosomal recessive |
| *BCAP31* | 300398 | Deafness, dystonia, and cerebral hypomyelination, 300475 (3), X-linked recessive |
| *BCAT2* | 113530 | ?Hypervalinemia or hyperleucine-isoleucinemia, 618850 (3), Autosomal recessive |
| *BCHE* | 177400 | Butyrylcholinesterase deficiency, 617936 (3), Autosomal recessive; {Apnea, postanesthetic, susceptibility to, due to BCHE deficiency}, 617936 (3), Autosomal recessive |
| *BCKDHA* | 608348 | Maple syrup urine disease, type Ia, 248600 (3), Autosomal recessive |
| *BCKDHB* | 248611 | Maple syrup urine disease, type Ib, 248600 (3), Autosomal recessive |
| *BCKDK* | 614901 | Branched-chain ketoacid dehydrogenase kinase deficiency, 614923 (3) |
| *BCL10* | 603517 | {Lymphoma, follicular, somatic}, 605027 (3); ?Immunodeficiency 37, 616098 (3), Autosomal recessive; {Sezary syndrome, somatic} (3); {Male germ cell tumor, somatic}, 273300 (3); Lymphoma, MALT, somatic, 137245 (3); {Mesothelioma, somatic}, 156240 (3) |
| *BCL11A* | 606557 | Dias-Logan syndrome, 617101 (3), Autosomal dominant |
| *BCL11B* | 606558 | Intellectual developmental disorder with dysmorphic facies, speech delay, and T-cell abnormalities, 618092 (3), Autosomal dominant; Immunodeficiency 49, 617237 (3), Autosomal dominant |
| *BCL2* | 151430 | Leukemia/lymphoma, B-cell, 2 (3) |
| *BCL3* | 109560 | Leukemia/lymphoma, B-cell, 3, 109560 (2) |
| *BCL7A* | 601406 | B-cell non-Hodgkin lymphoma, high-grade (3) |
| *BCO1* | 605748 | ?Hypercarotenemia and vitamin A deficiency, autosomal dominant, 115300 (3), Autosomal dominant |
| *BCOR* | 300485 | Microphthalmia, syndromic 2, 300166 (3), X-linked dominant |
| *BCORL1* | 300688 | Shukla-Vernon syndrome, 301029 (3), X-linked recessive |
| *BCR* | 151410 | Leukemia, chronic myeloid, Philadelphia chromosome positive, somatic, 608232 (4); Leukemia, acute lymphocytic, Philadelphia chromosome positive, somatic, 613065 (4) |
| *BCS1L* | 603647 | GRACILE syndrome, 603358 (3), Autosomal recessive; Mitochondrial complex III deficiency, nuclear type 1, 124000 (3), Autosomal recessive; Bjornstad syndrome, 262000 (3), Autosomal recessive |
| *BDP1* | 607012 | ?Deafness, autosomal recessive 112, 618257 (3), Autosomal recessive |
| *BEAN1* | 612051 | Spinocerebellar ataxia 31, 117210 (3), Autosomal dominant |
| *BEST1* | 607854 | Macular dystrophy, vitelliform, 2, 153700 (3), Autosomal dominant; ?Microcornea, rod-cone dystrophy, cataract, and posterior staphyloma 2, 193220 (3), Autosomal dominant; Retinitis pigmentosa-50, 613194 (3); Retinitis pigmentosa, concentric, 613194 (3); Vitreoretinochoroidopathy, 193220 (3), Autosomal dominant; Bestrophinopathy, autosomal recessive, 611809 (3) |
| *BFSP1* | 603307 | Cataract 33, multiple types, 611391 (3), Autosomal recessive, Autosomal dominant |
| *BFSP2* | 603212 | Cataract 12, multiple types, 611597 (3), Autosomal dominant |
| *BGN* | 301870 | Meester-Loeys syndrome, 300989 (3), X-linked; Spondyloepimetaphyseal dysplasia, X-linked, 300106 (3), X-linked recessive |
| *BHLHA9* | 615416 | ?Camptosynpolydactyly, complex, 607539 (3), Autosomal recessive; Syndactyly, mesoaxial synostotic, with phalangeal reduction, 609432 (3), Autosomal recessive |
| *BHLHE41* | 606200 | [Short sleep, familial natural, 1], 612975 (3), Autosomal dominant |
| *BICC1* | 614295 | {Renal dysplasia, cystic, susceptibility to}, 601331 (3), Autosomal dominant |
| *BICD2* | 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 |
| *BICRA* | 605690 | Coffin-Siris syndrome 12, 619325 (3), Autosomal dominant |
| *BIN1* | 601248 | Centronuclear myopathy 2, 255200 (3), Autosomal recessive |
| *BLK* | 191305 | Maturity-onset diabetes of the young, type 11, 613375 (3), Autosomal dominant |
| *BLM* | 604610 | Bloom syndrome, 210900 (3), Autosomal recessive |
| *BLNK* | 604515 | ?Agammaglobulinemia 4, 613502 (3), Autosomal recessive |
| *BLOC1S3* | 609762 | Hermansky-Pudlak syndrome 8, 614077 (3), Autosomal recessive |
| *BLOC1S5* | 607289 | Hermansky-Pudlak syndrome 11, 619172 (3), Autosomal recessive |
| *BLOC1S6* | 604310 | ?Hermansky-Pudlak syndrome 9, 614171 (3), Autosomal recessive |
| *BLVRA* | 109750 | Hyperbiliverdinemia, 614156 (3), Autosomal recessive, Autosomal dominant |
| *BMP1* | 112264 | Osteogenesis imperfecta, type XIII, 614856 (3), Autosomal recessive |
| *BMP15* | 300247 | Premature ovarian failure 4, 300510 (3), X-linked; Ovarian dysgenesis 2, 300510 (3), X-linked |
| *BMP2* | 112261 | Short stature, facial dysmorphism, and skeletal anomalies with or without cardiac anomalies 1, 617877 (3), Autosomal dominant; Brachydactyly, type A2, 112600 (3), Autosomal dominant; {HFE hemochromatosis, modifier of}, 235200 (3), Autosomal recessive |
| *BMP4* | 112262 | Orofacial cleft 11, 600625 (3); Microphthalmia, syndromic 6, 607932 (3), Autosomal dominant |
| *BMPER* | 608699 | Diaphanospondylodysostosis, 608022 (3), Autosomal recessive |
| *BMPR1A* | 601299 | Polyposis syndrome, hereditary mixed, 2, 610069 (3); Polyposis, juvenile intestinal, 174900 (3), Autosomal dominant |
| *BMPR1B* | 603248 | Acromesomelic dysplasia, Demirhan type, 609441 (3), Autosomal recessive; Brachydactyly, type A2, 112600 (3), Autosomal dominant; Brachydactyly, type A1, D, 616849 (3), Autosomal dominant |
| *BMPR2* | 600799 | Pulmonary hypertension, familial primary, 1, with or without HHT, 178600 (3), Autosomal dominant; Pulmonary hypertension, primary, fenfluramine or dexfenfluramine-associated, 178600 (3), Autosomal dominant; Pulmonary venoocclusive disease 1, 265450 (3), Autosomal dominant |
| *BMS1* | 611448 | ?Aplasia cutis congenita, nonsyndromic, 107600 (3), Autosomal dominant |
| *BNC1* | 601930 | ?Premature ovarian failure 16, 618723 (3), Autosomal dominant |
| *BNC2* | 608669 | Lower urinary tract obstruction, congenital, 618612 (3), Autosomal dominant |
| *BOLA3* | 613183 | Multiple mitochondrial dysfunctions syndrome 2 with hyperglycinemia, 614299 (3), Autosomal recessive |
| *BPGM* | 613896 | Erythrocytosis, familial, 8, 222800 (3), Autosomal recessive |
| *BPNT2 (IMPAD1)* | 614010 | Chondrodysplasia with joint dislocations, GPAPP type, 614078 (3), Autosomal recessive |
| *BPTF* | 601819 | Neurodevelopmental disorder with dysmorphic facies and distal limb anomalies, 617755 (3), Autosomal dominant |
| *BRAF* | 164757 | Melanoma, malignant, somatic, 155600 (3); LEOPARD syndrome 3, 613707 (3), Autosomal dominant; Cardiofaciocutaneous syndrome, 115150 (3), Autosomal dominant; Adenocarcinoma of lung, somatic, 211980 (3); Noonan syndrome 7, 613706 (3), Autosomal dominant; Colorectal cancer, somatic, 114500 (3); Nonsmall cell lung cancer, somatic, 211980 (3) |
| *BRAT1* | 614506 | Neurodevelopmental disorder with cerebellar atrophy and with or without seizures, 618056 (3), Autosomal recessive; Rigidity and multifocal seizure syndrome, lethal neonatal, 614498 (3), Autosomal recessive |
| *BRCA1* | 113705 | Fanconi anemia, complementation group S, 617883 (3), Autosomal recessive; {Breast-ovarian cancer, familial, 1}, 604370 (3), Multifactorial, Autosomal dominant; {Pancreatic cancer, susceptibility to, 4}, 614320 (3) |
| *BRCA2* | 600185 | Fanconi anemia, complementation group D1, 605724 (3), Autosomal recessive; {Glioblastoma 3}, 613029 (3), Autosomal recessive; {Medulloblastoma}, 155255 (3), Somatic mutation, Autosomal recessive, Autosomal dominant; {Prostate cancer}, 176807 (3), Somatic mutation, Autosomal dominant; {Breast-ovarian cancer, familial, 2}, 612555 (3), Autosomal dominant; {Breast cancer, male, susceptibility to}, 114480 (3), Somatic mutation, Autosomal dominant; {Pancreatic cancer 2}, 613347 (3); Wilms tumor, 194070 (3), Somatic mutation, Autosomal dominant |
| *BRDT* | 602144 | ?Spermatogenic failure 21, 617644 (3), Autosomal recessive |
| *BRF1* | 604902 | Cerebellofaciodental syndrome, 616202 (3), Autosomal recessive |
| *BRIP1* | 605882 | Fanconi anemia, complementation group J, 609054 (3); {Breast cancer, early-onset, susceptibility to}, 114480 (3), Somatic mutation, Autosomal dominant |
| *BRPF1* | 602410 | Intellectual developmental disorder with dysmorphic facies and ptosis, 617333 (3), Autosomal dominant |
| *BRWD3* | 300553 | Mental retardation, X-linked 93, 300659 (3), X-linked recessive |
| *BSCL2* | 606158 | Lipodystrophy, congenital generalized, type 2, 269700 (3), Autosomal recessive; Neuropathy, distal hereditary motor, type VC, 619112 (3), Autosomal dominant; Silver spastic paraplegia syndrome, 270685 (3), Autosomal dominant; Encephalopathy, progressive, with or without lipodystrophy, 615924 (3), Autosomal recessive |
| *BSG* | 109480 | [Blood group, OK], 111380 (3) |
| *BSND* | 606412 | Sensorineural deafness with mild renal dysfunction, 602522 (3), Autosomal recessive; Bartter syndrome, type 4a, 602522 (3), Autosomal recessive |
| *BTD* | 609019 | Biotinidase deficiency, 253260 (3), Autosomal recessive |
| *BTG4* | 605673 | Oocyte maturation defect 8, 619009 (3), Autosomal recessive |
| *BTK* | 300300 | Agammaglobulinemia, X-linked 1, 300755 (3), X-linked recessive; Isolated growth hormone deficiency, type III, with agammaglobulinemia, 307200 (3), X-linked recessive |
| *BTNL2* | 606000 | {Sarcoidosis, susceptibility to, 2}, 612387 (3), Autosomal dominant |
| *BUB1* | 602452 | Colorectal cancer with chromosomal instability, somatic, 114500 (3) |
| *BUB1B* | 602860 | Colorectal cancer, somatic, 114500 (3); [Premature chromatid separation trait], 176430 (3), Autosomal dominant; Mosaic variegated aneuploidy syndrome 1, 257300 (3), Autosomal recessive |
| *BVES* | 604577 | Muscular dystrophy, limb-girdle, autosomal recessive 25, 616812 (3), Autosomal recessive |
| *C11orf80* | 616109 | Hydatidiform mole, recurrent, 4, 618432 (3), Autosomal recessive |
| *C12orf4* | 616082 | Mental retardation, autosomal recessive 66, 618221 (3), Autosomal recessive |
| *C12orf57* | 615140 | Temtamy syndrome, 218340 (3), Autosomal recessive |
| *C14orf39* | 617307 | Spermatogenic failure 52, 619202 (3), Autosomal recessive; ?Premature ovarian failure 18, 619203 (3) |
| *C19orf12* | 614297 | Neurodegeneration with brain iron accumulation 4, 614298 (3), Autosomal recessive, Autosomal dominant; ?Spastic paraplegia 43, autosomal recessive, 615043 (3), Autosomal recessive |
| *C1GALT1C1* | 300611 | Tn polyagglutination syndrome, somatic, 300622 (3) |
| *C1QA* | 120550 | C1q deficiency, 613652 (3), Autosomal recessive |
| *C1QB* | 120570 | C1q deficiency, 613652 (3), Autosomal recessive |
| *C1QBP* | 601269 | Combined oxidative phosphorylation deficiency 33, 617713 (3), Autosomal recessive |
| *C1QC* | 120575 | C1q deficiency, 613652 (3), Autosomal recessive |
| *C1QTNF5* | 608752 | Retinal degeneration, late-onset, autosomal dominant, 605670 (3), Autosomal dominant |
| *C1R* | 613785 | Ehlers-Danlos syndrome, periodontal type, 1, 130080 (3), Autosomal dominant |
| *C1S* | 120580 | C1s deficiency, 613783 (3); Ehlers-Danlos syndrome, periodontal type, 2, 617174 (3), Autosomal dominant |
| *C2* | 613927 | C2 deficiency, 217000 (3), Autosomal recessive; {Macular degeneration, age-related, 14, reduced risk of}, 615489 (3), Digenic dominant |
| *C2CD3* | 615944 | Orofaciodigital syndrome XIV, 615948 (3), Autosomal recessive |
| *C2orf69* | 619219 | Combined oxidative phosphorylation deficiency 53, 619423 (3), Autosomal recessive |
| *C3* | 120700 | C3 deficiency, 613779 (3), Autosomal recessive; {Hemolytic uremic syndrome, atypical, susceptibility to, 5}, 612925 (3), Autosomal dominant; {Macular degeneration, age-related, 9}, 611378 (3) |
| *C4A* | 120810 | [Blood group, Rodgers], 614374 (3); C4a deficiency, 614380 (3), Autosomal recessive |
| *C4B* | 120820 | C4B deficiency, 614379 (3) |
| *C5* | 120900 | C5 deficiency, 609536 (3), Autosomal recessive; [Eculizumab, poor response to], 615749 (3), Autosomal dominant |
| *C6* | 217050 | C6 deficiency, 612446 (3); Combined C6/C7 deficiency (3) |
| *C7* | 217070 | C7 deficiency, 610102 (3) |
| *C8A* | 120950 | C8 deficiency, type I, 613790 (3), Autosomal recessive |
| *C8B* | 120960 | C8 deficiency, type II, 613789 (3), Autosomal recessive |
| *C9* | 120940 | C9 deficiency, 613825 (3); {Macular degeneration, age-related, 15, susceptibility to}, 615591 (3) |
| *C9orf72* | 614260 | Frontotemporal dementia and/or amyotrophic lateral sclerosis 1, 105550 (3), Autosomal dominant |
| *CA12* | 603263 | Hyperchlorhidrosis, isolated, 143860 (3), Autosomal recessive |
| *CA2* | 611492 | Osteopetrosis, autosomal recessive 3, with renal tubular acidosis, 259730 (3), Autosomal recessive |
| *CA5A* | 114761 | Hyperammonemia due to carbonic anhydrase VA deficiency, 615751 (3), Autosomal recessive |
| *CA8* | 114815 | Cerebellar ataxia and mental retardation with or without quadrupedal locomotion 3, 613227 (3), Autosomal recessive |
| *CABP2* | 607314 | Deafness, autosomal recessive 93, 614899 (3), Autosomal recessive |
| *CABP4* | 608965 | Cone-rod synaptic disorder, congenital nonprogressive, 610427 (3), Autosomal recessive |
| *CACNA1A* | 601011 | Developemental and epileptic encephalopathy 42, 617106 (3), Autosomal dominant; Spinocerebellar ataxia 6, 183086 (3), Autosomal dominant; Episodic ataxia, type 2, 108500 (3), Autosomal dominant; Migraine, familial hemiplegic, 1, with progressive cerebellar ataxia, 141500 (3), Autosomal dominant; Migraine, familial hemiplegic, 1, 141500 (3), Autosomal dominant |
| *CACNA1B* | 601012 | Neurodevelopmental disorder with seizures and nonepileptic hyperkinetic movements, 618497 (3), Autosomal recessive |
| *CACNA1C* | 114205 | Timothy syndrome, 601005 (3), Autosomal dominant; Long QT syndrome 8, 618447 (3); Brugada syndrome 3, 611875 (3) |
| *CACNA1D* | 114206 | Primary aldosteronism, seizures, and neurologic abnormalities, 615474 (3), Autosomal dominant; Sinoatrial node dysfunction and deafness, 614896 (3), Autosomal recessive |
| *CACNA1E* | 601013 | Developmental and epileptic encephalopathy 69, 618285 (3), Autosomal dominant |
| *CACNA1F* | 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 |
| *CACNA1G* | 604065 | Spinocerebellar ataxia 42, 616795 (3), Autosomal dominant; Spinocerebellar ataxia 42, early-onset, severe, with neurodevelopmental deficits, 618087 (3), Autosomal dominant |
| *CACNA1H* | 607904 | {Epilepsy, childhood absence, susceptibility to, 6}, 611942 (3); Hyperaldosteronism, familial, type IV, 617027 (3), Autosomal dominant; {Epilepsy, idiopathic generalized, susceptibility to, 6}, 611942 (3) |
| *CACNA1S* | 114208 | {Thyrotoxic periodic paralysis, susceptibility to, 1}, 188580 (3), Autosomal dominant; Hypokalemic periodic paralysis, type 1, 170400 (3), Autosomal dominant; {Malignant hyperthermia susceptibility 5}, 601887 (3), Autosomal dominant |
| *CACNA2D1* | 114204/154276 | -/{Malignant hyperthermia susceptibility 3}, 154276 (2), Autosomal dominant |
| *CACNA2D2* | 607082 | Cerebellar atrophy with seizures and variable developmental delay, 618501 (3), Autosomal recessive |
| *CACNA2D4* | 608171 | Retinal cone dystrophy 4, 610478 (3), Autosomal recessive |
| *CACNB2* | 600003 | Brugada syndrome 4, 611876 (3) |
| *CACNB4* | 601949 | {Epilepsy, juvenile myoclonic, susceptibility to, 6}, 607682 (3), Autosomal dominant; Episodic ataxia, type 5, 613855 (3), Autosomal dominant; {Epilepsy, idiopathic generalized, susceptibility to, 9}, 607682 (3), Autosomal dominant |
| *CACNG2* | 602911 | ?Mental retardation, autosomal dominant 10, 614256 (3), Autosomal dominant |
| *CAD* | 114010 | Developmental and epileptic encephalopathy 50, 616457 (3), Autosomal recessive |
| *CALCR* | 114131 | {Osteoporosis, postmenopausal, susceptibility}, 166710 (3), Autosomal dominant |
| *CALCRL* | 114190 | ?Lymphatic malformation 8, 618773 (3), Autosomal recessive |
| *CALM1* | 114180 | Ventricular tachycardia, catecholaminergic polymorphic, 4, 614916 (3), Autosomal dominant; Long QT syndrome 14, 616247 (3), Autosomal dominant |
| *CALM2* | 114182 | Long QT syndrome 15, 616249 (3), Autosomal dominant |
| *CALM3* | 114183 | Long QT syndrome 16, 618782 (3), Autosomal dominant; ?Ventricular tachycardia, catecholaminergic polymorphic 6, 618782 (3), Autosomal dominant |
| *CALR* | 109091 | Myelofibrosis, somatic, 254450 (3); Thrombocythemia, somatic, 187950 (3) |
| *CAMK2A* | 114078 | Mental retardation, autosomal dominant 53, 617798 (3), Autosomal dominant; ?Mental retardation, autosomal recessive 63, 618095 (3), Autosomal recessive |
| *CAMK2B* | 607707 | Mental retardation, autosomal dominant 54, 617799 (3), Autosomal dominant |
| *CAMK2G* | 602123 | Mental retardation, autosomal dominant 59, 618522 (3), Autosomal dominant |
| *CAMTA1* | 611501 | Cerebellar ataxia, nonprogressive, with mental retardation, 614756 (3), Autosomal dominant |
| *CANT1* | 613165 | Desbuquois dysplasia 1, 251450 (3), Autosomal recessive; Epiphyseal dysplasia, multiple, 7, 617719 (3), Autosomal recessive |
| *CAPN1* | 114220 | Spastic paraplegia 76, autosomal recessive, 616907 (3), Autosomal recessive |
| *CAPN10* | 605286 | {Diabetes mellitus, noninsulin-dependent 1}, 601283 (3) |
| *CAPN15* | 603267 | Oculogastrointestinal neurodevelopmental syndrome, 619318 (3), Autosomal recessive |
| *CAPN3* | 114240 | Muscular dystrophy, limb-girdle, autosomal recessive 1, 253600 (3), Autosomal recessive; Muscular dystrophy, limb-girdle, autosomal dominant 4, 618129 (3), Autosomal dominant |
| *CAPN5* | 602537 | Vitreoretinopathy, neovascular inflammatory, 193235 (3), Autosomal dominant |
| *CARD11* | 607210 | B-cell expansion with NFKB and T-cell anergy, 616452 (3), Autosomal dominant; Immunodeficiency 11B with atopic dermatitis, 617638 (3), Autosomal dominant; Immunodeficiency 11A, 615206 (3), Autosomal recessive |
| *CARD14* | 607211 | Psoriasis 2, 602723 (3), Autosomal dominant; Pityriasis rubra pilaris, 173200 (3), Autosomal dominant |
| *CARD8* | 609051 | ?Inflammatory bowel disease (Crohn disease) 30, 619079 (3), Autosomal dominant |
| *CARD9* | 607212 | Candidiasis, familial, 2, autosomal recessive, 212050 (3), Autosomal recessive |
| *CARMIL2* | 610859 | Immunodeficiency 58, 618131 (3), Autosomal recessive |
| *CARS1 (CARS)* | 123859 | Microcephaly, developmental delay, and brittle hair syndrome, 618891 (3), Autosomal recessive |
| *CARS2* | 612800 | Combined oxidative phosphorylation deficiency 27, 616672 (3), Autosomal recessive |
| *CARTPT* | 602606 | {?Obesity, susceptibility to}, 601665 (3), Multifactorial, Autosomal recessive, Autosomal dominant |
| *CASK* | 300172 | Mental retardation and microcephaly with pontine and cerebellar hypoplasia, 300749 (3), X-linked dominant; Mental retardation, with or without nystagmus, 300422 (3); FG syndrome 4, 300422 (3) |
| *CASP10* | 601762 | Autoimmune lymphoproliferative syndrome, type II, 603909 (3), Autosomal dominant; Gastric cancer, somatic, 613659 (3); Lymphoma, non-Hodgkin, somatic, 605027 (3) |
| *CASP12* | 608633 | {Sepsis, susceptibility to} (3) |
| *CASP14* | 605848 | Ichthyosis, congenital, autosomal recessive 12, 617320 (3), Autosomal recessive |
| *CASP8* | 601763 | {Breast cancer, protection against}, 114480 (3), Somatic mutation, Autosomal dominant; Hepatocellular carcinoma, somatic, 114550 (3); ?Autoimmune lymphoproliferative syndrome, type IIB, 607271 (3), Autosomal recessive; {Lung cancer, protection against}, 211980 (3), Somatic mutation, Autosomal dominant |
| *CASQ1* | 114250 | Myopathy, vacuolar, with CASQ1 aggregates, 616231 (3), Autosomal dominant |
| *CASQ2* | 114251 | Ventricular tachycardia, catecholaminergic polymorphic, 2, 611938 (3), Autosomal recessive |
| *CASR* | 601199 | Hypocalcemia, autosomal dominant, with Bartter syndrome, 601198 (3), Autosomal dominant; {Epilepsy idiopathic generalized, susceptibility to, 8}, 612899 (3); Hyperparathyroidism, neonatal, 239200 (3), Autosomal recessive, Autosomal dominant; Hypocalcemia, autosomal dominant, 601198 (3), Autosomal dominant; Hypocalciuric hypercalcemia, type I, 145980 (3), Autosomal dominant |
| *CAST* | 114090 | Peeling skin with leukonychia, acral punctate keratoses, cheilitis, and knuckle pads, 616295 (3), Autosomal recessive |
| *CAT* | 115500 | Acatalasemia, 614097 (3) |
| *CATIP* | 619387 | ?Spermatogenic failure 54, 619379 (3), Autosomal recessive |
| *CATSPER1* | 606389 | Spermatogenic failure 7, 612997 (3), Autosomal recessive |
| *CAV1* | 601047 | ?Lipodystrophy, congenital generalized, type 3, 612526 (3), Autosomal recessive; Pulmonary hypertension, primary, 3, 615343 (3), Autosomal dominant; Lipodystrophy, familial partial, type 7, 606721 (3), Autosomal dominant |
| *CAV3* | 601253 | Myopathy, distal, Tateyama type, 614321 (3), Autosomal dominant; Creatine phosphokinase, elevated serum, 123320 (3), Autosomal dominant; Cardiomyopathy, familial hypertrophic, 192600 (3), Digenic dominant, Autosomal dominant; Rippling muscle disease 2, 606072 (3), Autosomal dominant; Long QT syndrome 9, 611818 (3), Autosomal dominant |
| *CAVIN1* | 603198 | Lipodystrophy, congenital generalized, type 4, 613327 (3), Autosomal recessive |
| *CBFB* | 121360 | Myeloid leukemia, acute, M4/M4Eo subtype, somatic, 601626 (1) |
| *CBL* | 165360 | Noonan syndrome-like disorder with or without juvenile myelomonocytic leukemia, 613563 (3), Autosomal dominant; ?Juvenile myelomonocytic leukemia, 607785 (3), Somatic mutation, Autosomal dominant |
| *CBLIF (GIF)* | 609342 | Intrinsic factor deficiency, 261000 (3), Autosomal recessive |
| *CBS* | 613381 | Thrombosis, hyperhomocysteinemic, 236200 (3), Autosomal recessive; Homocystinuria, B6-responsive and nonresponsive types, 236200 (3), Autosomal recessive |
| *CBX2* | 602770 | ?46XY sex reversal 5, 613080 (3), Autosomal recessive |
| *CC2D1A* | 610055 | Mental retardation, autosomal recessive 3, 608443 (3), Autosomal recessive |
| *CC2D2A* | 612013 | COACH syndrome 2, 619111 (3); Meckel syndrome 6, 612284 (3), Autosomal recessive; Joubert syndrome 9, 612285 (3), Autosomal recessive |
| *CCBE1* | 612753 | Hennekam lymphangiectasia-lymphedema syndrome 1, 235510 (3), Autosomal recessive |
| *CCDC103* | 614677 | Ciliary dyskinesia, primary, 17, 614679 (3), Autosomal recessive |
| *CCDC115* | 613734 | Congenital disorder of glycosylation, type IIo, 616828 (3), Autosomal recessive |
| *CCDC174* | 616735 | Hypotonia, infantile, with psychomotor retardation, 616816 (3), Autosomal recessive |
| *CCDC22* | 300859 | Ritscher-Schinzel syndrome 2, 300963 (3), X-linked recessive |
| *CCDC28B* | 610162 | {Bardet-Biedl syndrome 1, modifier of}, 209900 (3), Autosomal recessive, Digenic recessive |
| *CCDC32* | 618941 | Cardiofacioneurodevelopmental syndrome, 619123 (3) |
| *CCDC39* | 613798 | Ciliary dyskinesia, primary, 14, 613807 (3), Autosomal recessive |
| *CCDC40* | 613799 | Ciliary dyskinesia, primary, 15, 613808 (3), Autosomal recessive |
| *CCDC47* | 618260 | Trichohepatoneurodevelopmental syndrome, 618268 (3), Autosomal recessive |
| *CCDC50* | 611051 | ?Deafness, autosomal dominant 44, 607453 (3), Autosomal dominant |
| *CCDC65* | 611088 | Ciliary dyskinesia, primary, 27, 615504 (3), Autosomal recessive |
| *CCDC78* | 614666 | ?Centronuclear myopathy 4, 614807 (3), Autosomal dominant |
| *CCDC8* | 614145 | 3-M syndrome 3, 614205 (3), Autosomal recessive |
| *CCDC88A* | 609736 | ?PEHO syndrome-like, 617507 (3), Autosomal recessive |
| *CCDC88C* | 611204 | ?Spinocerebellar ataxia 40, 616053 (3), Autosomal dominant; Hydrocephalus, congenital, 1, 236600 (3), Autosomal recessive |
| *CCL11* | 601156 | {Asthma, susceptibility to}, 600807 (3), Autosomal dominant; {HIV1, resistance to}, 609423 (3) |
| *CCL2* | 158105 | {Mycobacterium tuberculosis, susceptibility to}, 607948 (3); {HIV-1, resistance to}, 609423 (3); {Coronary artery disease, modifier of} (3); {Spina bifida, susceptibility to}, 182940 (3), Autosomal dominant |
| *CCL3* | 182283 | {HIV infection, resistance to}, 609423 (2) |
| *CCL3L1* | 601395 | {HIV/AIDS, susceptibility to}, 609423 (3) |
| *CCL5* | 187011 | {HIV-1 disease, rapid progression of}, 609423 (3); {HIV-1 disease, delayed progression of}, 609423 (3) |
| *CCM2* | 607929 | Cerebral cavernous malformations-2, 603284 (3), Autosomal dominant |
| *CCN6 (WISP3)* | 603400 | Progressive pseudorheumatoid dysplasia, 208230 (3), Autosomal recessive |
| *CCND1* | 168461 | {von Hippel-Lindau syndrome, modifier of}, 193300 (3), Autosomal dominant; {Colorectal cancer, susceptibility to}, 114500 (3), Somatic mutation, Autosomal dominant; {Multiple myeloma, susceptibility to}, 254500 (3), Somatic mutation |
| *CCND2* | 123833 | Megalencephaly-polymicrogyria-polydactyly-hydrocephalus syndrome 3, 615938 (3), Autosomal dominant |
| *CCNF* | 600227 | Frontotemporal dementia and/or amyotrophic lateral sclerosis 5, 619141 (3) |
| *CCNK* | 603544 | ?Intellectual developmental disorder with hypertelorism and distinctive facies, 618147 (3), Autosomal dominant |
| *CCNO* | 607752 | Ciliary dyskinesia, primary, 29, 615872 (3), Autosomal recessive |
| *CCNQ* | 300708 | STAR syndrome, 300707 (3), X-linked dominant |
| *CCR2* | 601267 | {HIV infection, susceptibility/resistance to} (3) |
| *CCR5* | 601373 | {HIV infection, susceptibility/resistance to} (3); {Diabetes mellitus, insulin-dependent, 22}, 612522 (3); {Hepatitis C virus, resistance to}, 609532 (3); {West nile virus, susceptibility to}, 610379 (3) |
| *CCT5* | 610150 | Neuropathy, hereditary sensory, with spastic paraplegia, 256840 (3), Autosomal recessive |
| *CD151* | 602243 | [Blood group, Raph], 179620 (3); Nephropathy with pretibial epidermolysis bullosa and deafness, 609057 (3) |
| *CD164* | 603356 | ?Deafness, autosomal dominant 66, 616969 (3), Autosomal dominant |
| *CD19* | 107265 | Immunodeficiency, common variable, 3, 613493 (3), Autosomal recessive |
| *CD207* | 604862 | [?Birbeck granule deficiency], 613393 (3) |
| *CD209* | 604672 | {HIV type 1, susceptibility to}, 609423 (3); {Mycobacterium tuberculosis, susceptibility to}, 607948 (3); {Dengue fever, protection against}, 614371 (3) |
| *CD244* | 605554 | {Rheumatoid arthritis, susceptibility to}, 180300 (3) |
| *CD247* | 186780 | ?Immunodeficiency 25, 610163 (3), Autosomal recessive |
| *CD27* | 186711 | Lymphoproliferative syndrome 2, 615122 (3), Autosomal recessive |
| *CD2AP* | 604241 | Glomerulosclerosis, focal segmental, 3, 607832 (3) |
| *CD320* | 606475 | Methylmalonic aciduria, transient, due to transcobalamin receptor defect, 613646 (3) |
| *CD36* | 173510 | Platelet glycoprotein IV deficiency, 608404 (3), Autosomal recessive; {Coronary heart disease, susceptibility to, 7}, 610938 (3); {Malaria, cerebral, susceptibility to}, 611162 (3); {Malaria, cerebral, reduced risk of}, 611162 (3) |
| *CD3D* | 186790 | Immunodeficiency 19, 615617 (3), Autosomal recessive |
| *CD3E* | 186830 | Immunodeficiency 18, 615615 (3), Autosomal recessive; Immunodeficiency 18, SCID variant, 615615 (3), Autosomal recessive |
| *CD3G* | 186740 | Immunodeficiency 17, CD3 gamma deficient, 615607 (3), Autosomal recessive |
| *CD4* | 186940 | Immunodeficiency 79, 619238 (3), Autosomal recessive; OKT4 epitope deficiency, 613949 (3) |
| *CD40* | 109535 | Immunodeficiency with hyper-IgM, type 3, 606843 (3), Autosomal recessive |
| *CD40LG* | 300386 | Immunodeficiency, X-linked, with hyper-IgM, 308230 (3), X-linked recessive |
| *CD44* | 107269 | [Blood group, Indian system], 609027 (3) |
| *CD46* | 120920 | {Hemolytic uremic syndrome, atypical, susceptibility to, 2}, 612922 (3), Autosomal recessive, Autosomal dominant |
| *CD55* | 125240 | [Blood group Cromer], 613793 (3), Autosomal recessive; Complement hyperactivation, angiopathic thrombosis, and protein-losing enteropathy, 226300 (3), Autosomal recessive |
| *CD59* | 107271 | Hemolytic anemia, CD59-mediated, with or without immune-mediated polyneuropathy, 612300 (3), Autosomal recessive |
| *CD70* | 602840 | Lymphoproliferative syndrome 3, 618261 (3), Autosomal recessive |
| *CD79A* | 112205 | Agammaglobulinemia 3, 613501 (3), Autosomal recessive |
| *CD79B* | 147245 | Agammaglobulinemia 6, 612692 (3), Autosomal recessive |
| *CD81* | 186845 | Immunodeficiency, common variable, 6, 613496 (3), Autosomal recessive |
| *CD8A* | 186910 | CD8 deficiency, familial, 608957 (3), Autosomal recessive |
| *CD96* | 606037 | C syndrome, 211750 (3), Autosomal dominant |
| *CDAN1* | 607465 | Dyserythropoietic anemia, congenital, type Ia, 224120 (3), Autosomal recessive |
| *CDC14A* | 603504 | Deafness, autosomal recessive 32, with or without immotile sperm, 608653 (3), Autosomal recessive |
| *CDC40* | 605585 | ?Pontocerebellar hypoplasia, type 15, 619302 (3), Autosomal recessive |
| *CDC42* | 116952 | Takenouchi-Kosaki syndrome, 616737 (3), Autosomal dominant |
| *CDC45* | 603465 | Meier-Gorlin syndrome 7, 617063 (3), Autosomal recessive |
| *CDC6* | 602627 | ?Meier-Gorlin syndrome 5, 613805 (3), Autosomal recessive |
| *CDC73* | 607393 | Hyperparathyroidism, familial primary, 145000 (3), Autosomal dominant; Parathyroid adenoma with cystic changes, 145001 (3), Autosomal dominant; Parathyroid carcinoma, 608266 (3); Hyperparathyroidism-jaw tumor syndrome, 145001 (3), Autosomal dominant |
| *CDCA7* | 609937 | Immunodeficiency-centromeric instability-facial anomalies syndrome 3, 616910 (3), Autosomal recessive |
| *CDH1* | 192090 | Ovarian cancer, somatic, 167000 (3); Blepharocheilodontic syndrome 1, 119580 (3), Autosomal dominant; Endometrial carcinoma, somatic, 608089 (3); {Breast cancer, lobular}, 114480 (3), Somatic mutation, Autosomal dominant; Gastric cancer, hereditary diffuse, with or without cleft lip and/or palate, 137215 (3), Autosomal dominant; {Prostate cancer, susceptibility to}, 176807 (3), Somatic mutation, Autosomal dominant |
| *CDH11* | 600023 | Elsahy-Waters syndrome, 211380 (3), Autosomal recessive |
| *CDH15* | 114019 | Mental retardation, autosomal dominant 3, 612580 (3) |
| *CDH2* | 114020 | Arrhythmogenic right ventricular dysplasia, familial, 14, 618920 (3), Autosomal dominant; Agenesis of corpus callosum, cardiac, ocular, and genital syndrome, 618929 (3), Autosomal dominant |
| *CDH23* | 605516 | Usher syndrome, type 1D, 601067 (3), Autosomal recessive, Digenic recessive; {Pituitary adenoma 5, multiple types}, 617540 (3), Autosomal dominant; Usher syndrome, type 1D/F digenic, 601067 (3), Autosomal recessive, Digenic recessive; Deafness, autosomal recessive 12, 601386 (3), Autosomal recessive |
| *CDH3* | 114021 | Hypotrichosis, congenital, with juvenile macular dystrophy, 601553 (3), Autosomal recessive; Ectodermal dysplasia, ectrodactyly, and macular dystrophy, 225280 (3), Autosomal recessive |
| *CDHR1* | 609502 | Cone-rod dystrophy 15, 613660 (3), Autosomal recessive; Retinitis pigmentosa 65, 613660 (3), Autosomal recessive |
| *CDIN1 (C15orf41)* | 615626 | Dyserythropoietic anemia, congenital, type Ib, 615631 (3), Autosomal recessive |
| *CDK10* | 603464 | Al Kaissi syndrome, 617694 (3), Autosomal recessive |
| *CDK13* | 603309 | Congenital heart defects, dysmorphic facial features, and intellectual developmental disorder, 617360 (3), Autosomal dominant |
| *CDK19* | 614720 | Developmental and epileptic encephalopathy 87, 618916 (3), Autosomal dominant |
| *CDK4* | 123829 | {Melanoma, cutaneous malignant, 3}, 609048 (3), Autosomal dominant |
| *CDK5* | 123831 | ?Lissencephaly 7 with cerebellar hypoplasia, 616342 (3), Autosomal recessive |
| *CDK5RAP2* | 608201 | Microcephaly 3, primary, autosomal recessive, 604804 (3), Autosomal recessive |
| *CDK6* | 603368 | ?Microcephaly 12, primary, autosomal recessive, 616080 (3), Autosomal recessive |
| *CDK8* | 603184 | Intellectual developmental disorder with hypotonia and behavioral abnormalities, 618748 (3), Autosomal dominant |
| *CDKL5* | 300203 | Developmental and epileptic encephalopathy 2, 300672 (3), X-linked dominant |
| *CDKN1B* | 600778 | Multiple endocrine neoplasia, type IV, 610755 (3), Autosomal dominant |
| *CDKN1C* | 600856 | IMAGE syndrome, 614732 (3), Autosomal dominant; Beckwith-Wiedemann syndrome, 130650 (3), Autosomal dominant |
| *CDKN2A* | 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 |
| *CDON* | 608707 | Holoprosencephaly 11, 614226 (3), Autosomal dominant |
| *CDSN* | 602593 | Hypotrichosis 2, 146520 (3), Autosomal dominant; Peeling skin syndrome 1, 270300 (3), Autosomal recessive |
| *CDT1* | 605525 | Meier-Gorlin syndrome 4, 613804 (3), Autosomal recessive |
| *CEACAM16* | 614591 | Deafness, autosomal dominant 4B, 614614 (3), Autosomal dominant; Deafness, autosomal recessive 113, 618410 (3), Autosomal recessive |
| *CEBPA* | 116897 | Leukemia, acute myeloid, somatic, 601626 (3); ?Leukemia, acute myeloid, 601626 (3), Somatic mutation, Autosomal dominant |
| *CEBPE* | 600749 | Specific granule deficiency, 245480 (3), Autosomal recessive |
| *CEL* | 114840 | Maturity-onset diabetes of the young, type VIII, 609812 (3), Autosomal dominant |
| *CELA2A* | 609443 | Abdominal obesity-metabolic syndrome 4, 618620 (3), Autosomal dominant |
| *CELSR1* | 604523 | Lymphatic malformation 9, 619319 (3), Autosomal dominant |
| *CENPE* | 117143 | ?Microcephaly 13, primary, autosomal recessive, 616051 (3), Autosomal recessive |
| *CENPF* | 600236 | Stromme syndrome, 243605 (3), Autosomal recessive |
| *CENPJ* | 609279 | Microcephaly 6, primary, autosomal recessive, 608393 (3), Autosomal recessive; ?Seckel syndrome 4, 613676 (3), Autosomal recessive |
| *CENPT* | 611510 | ?Short stature and microcephaly with genital anomalies, 618702 (3), Autosomal recessive |
| *CEP104* | 616690 | Joubert syndrome 25, 616781 (3), Autosomal recessive |
| *CEP112* | 618980 | Spermatogenic failure 44, 619044 (3), Autosomal recessive |
| *CEP120* | 613446 | Short-rib thoracic dysplasia 13 with or without polydactyly, 616300 (3), Autosomal recessive; Joubert syndrome 31, 617761 (3), Autosomal recessive |
| *CEP135* | 611423 | Microcephaly 8, primary, autosomal recessive, 614673 (3), Autosomal recessive |
| *CEP152* | 613529 | Microcephaly 9, primary, autosomal recessive, 614852 (3), Autosomal recessive; Seckel syndrome 5, 613823 (3), Autosomal recessive |
| *CEP164* | 614848 | Nephronophthisis 15, 614845 (3), Autosomal recessive |
| *CEP19* | 615586 | Morbid obesity and spermatogenic failure, 615703 (3), Autosomal recessive |
| *CEP250* | 609689 | Cone-rod dystrophy and hearing loss 2, 618358 (3), Autosomal recessive |
| *CEP290* | 610142 | Leber congenital amaurosis 10, 611755 (3); Joubert syndrome 5, 610188 (3), Autosomal recessive; Senior-Loken syndrome 6, 610189 (3), Autosomal recessive; ?Bardet-Biedl syndrome 14, 615991 (3), Autosomal recessive; Meckel syndrome 4, 611134 (3), Autosomal recessive |
| *CEP41* | 610523 | Joubert syndrome 15, 614464 (3), Autosomal recessive |
| *CEP43 (FGFR1OP)* | 605392 | Myeloproliferative disorder, 605392 (2) |
| *CEP55* | 610000 | Multinucleated neurons, anhydramnios, renal dysplasia, cerebellar hypoplasia, and hydranencephaly, 236500 (3), Autosomal recessive |
| *CEP57* | 607951 | Mosaic variegated aneuploidy syndrome 2, 614114 (3), Autosomal recessive |
| *CEP63* | 614724 | ?Seckel syndrome 6, 614728 (3), Autosomal recessive |
| *CEP78* | 617110 | Cone-rod dystrophy and hearing loss, 617236 (3), Autosomal recessive |
| *CEP83* | 615847 | Nephronophthisis 18, 615862 (3), Autosomal recessive |
| *CEP85L* | 618865 | Lissencephaly 10, 618873 (3), Autosomal dominant |
| *CERKL* | 608381 | Retinitis pigmentosa 26, 608380 (3) |
| *CERS1* | 606919 | ?Epilepsy, progressive myoclonic, 8, 616230 (3), Autosomal recessive |
| *CERS3* | 615276 | Ichthyosis, congenital, autosomal recessive 9, 615023 (3), Autosomal recessive |
| *CERT1 (COL4A3BP)* | 604677 | Mental retardation, autosomal dominant 34, 616351 (3), Autosomal dominant |
| *CES1* | 114835 | Drug metabolism, altered, CES1-related, 618057 (3), Autosomal dominant |
| *CETP* | 118470 | [High density lipoprotein cholesterol level QTL 10], 143470 (3), Autosomal dominant; Hyperalphalipoproteinemia, 143470 (3), Autosomal dominant |
| *CFAP251 (WDR66)* | 618146 | Spermatogenic failure 33, 618152 (3), Autosomal recessive |
| *CFAP298* | 615494 | Ciliary dyskinesia, primary, 26, 615500 (3), Autosomal recessive |
| *CFAP300* | 618058 | Ciliary dyskinesia, primary, 38, 618063 (3), Autosomal recessive |
| *CFAP410* | 603191 | Retinal dystrophy with macular staphyloma, 617547 (3), Autosomal recessive; Spondylometaphyseal dysplasia, axial, 602271 (3), Autosomal recessive |
| *CFAP418 (C8orf37)* | 614477 | Retinitis pigmentosa 64, 614500 (3), Autosomal recessive; Cone-rod dystrophy 16, 614500 (3), Autosomal recessive; Bardet-Biedl syndrome 21, 617406 (3), Autosomal recessive |
| *CFAP43* | 617558 | Hydrocephalus, normal pressure, 1, 236690 (3), Autosomal dominant; Spermatogenic failure 19, 617592 (3), Autosomal recessive |
| *CFAP44* | 617559 | ?Spermatogenic failure 20, 617593 (3), Autosomal recessive |
| *CFAP47* | 301057 | Spermatogenic failure, X-linked, 3, 301059 (3), X-linked recessive |
| *CFAP53* | 614759 | Heterotaxy, visceral, 6, autosomal recessive, 614779 (3), Autosomal recessive |
| *CFAP58* | 619129 | Spermatogenic failure 49, 619144 (3) |
| *CFAP65* | 614270 | Spermatogenic failure 40, 618664 (3), Autosomal recessive |
| *CFAP69* | 617949 | Spermatogenic failure 24, 617959 (3), Autosomal recessive |
| *CFAP70* | 618661 | ?Spermatogenic failure 41, 618670 (3), Autosomal recessive |
| *CFAP91 (MAATS1)* | 609910 | Spermatogenic failure 51, 619177 (3), Autosomal recessive |
| *CFB* | 138470 | ?Complement factor B deficiency, 615561 (3), Autosomal recessive; {Hemolytic uremic syndrome, atypical, susceptibility to, 4}, 612924 (3), Autosomal dominant; {Macular degeneration, age-related, 14, reduced risk of}, 615489 (3), Digenic dominant |
| *CFC1* | 605194 | Heterotaxy, visceral, 2, autosomal, 605376 (3), Autosomal dominant |
| *CFD* | 134350 | Complement factor D deficiency, 613912 (3), Autosomal recessive |
| *CFH* | 134370 | {Macular degeneration, age-related, 4}, 610698 (3); Basal laminar drusen, 126700 (3), Autosomal dominant; Complement factor H deficiency, 609814 (3), Autosomal recessive, Autosomal dominant; {Hemolytic uremic syndrome, atypical, susceptibility to, 1}, 235400 (3), Autosomal recessive, Autosomal dominant |
| *CFHR1* | 134371 | {Macular degeneration, age-related, reduced risk of}, 603075 (3), Autosomal dominant; {Hemolytic uremic syndrome, atypical, susceptibility to}, 235400 (3), Autosomal recessive, Autosomal dominant |
| *CFHR3* | 605336 | {Macular degeneration, age-related, reduced risk of}, 603075 (3), Autosomal dominant; {Hemolytic uremic syndrome, atypical, susceptibility to}, 235400 (3), Autosomal recessive, Autosomal dominant |
| *CFHR5* | 608593 | Nephropathy due to CFHR5 deficiency, 614809 (3), Autosomal dominant |
| *CFI* | 217030 | {Hemolytic uremic syndrome, atypical, susceptibility to, 3}, 612923 (3), Autosomal dominant; {Macular degeneration, age-related, 13, susceptibility to}, 615439 (3), Autosomal dominant; Complement factor I deficiency, 610984 (3), Autosomal recessive |
| *CFL2* | 601443 | Nemaline myopathy 7, autosomal recessive, 610687 (3), Autosomal recessive |
| *CFP* | 300383 | Properdin deficiency, X-linked, 312060 (3), X-linked recessive |
| *CFTR* | 602421 | Cystic fibrosis, 219700 (3), Autosomal recessive; Sweat chloride elevation without CF (3); Congenital bilateral absence of vas deferens, 277180 (3), Autosomal recessive; {Pancreatitis, hereditary}, 167800 (3), Autosomal dominant; {Bronchiectasis with or without elevated sweat chloride 1, modifier of}, 211400 (3), Autosomal dominant; {Hypertrypsinemia, neonatal} (3) |
| *CHAMP1* | 616327 | Mental retardation, autosomal dominant 40, 616579 (3), Autosomal dominant |
| *CHAT* | 118490 | Myasthenic syndrome, congenital, 6, presynaptic, 254210 (3), Autosomal recessive |
| *CHCHD10* | 615903 | ?Myopathy, isolated mitochondrial, autosomal dominant, 616209 (3), Autosomal dominant; Spinal muscular atrophy, Jokela type, 615048 (3), Autosomal dominant; Frontotemporal dementia and/or amyotrophic lateral sclerosis 2, 615911 (3), Autosomal dominant |
| *CHCHD2* | 616244 | Parkinson disease 22, autosomal dominant, 616710 (3), Autosomal dominant |
| *CHD1* | 602118 | Pilarowski-Bjornsson syndrome, 617682 (3), Autosomal dominant |
| *CHD2* | 602119 | Develomental and epileptic encephalopathy 94, 615369 (3), Autosomal dominant |
| *CHD3* | 602120 | Snijders Blok-Campeau syndrome, 618205 (3), Autosomal dominant |
| *CHD4* | 603277 | Sifrim-Hitz-Weiss syndrome, 617159 (3), Autosomal dominant |
| *CHD7* | 608892 | Hypogonadotropic hypogonadism 5 with or without anosmia, 612370 (3), Autosomal dominant; CHARGE syndrome, 214800 (3), Autosomal dominant |
| *CHD8* | 610528 | {Autism, susceptibility to, 18}, 615032 (3), Autosomal dominant |
| *CHEK2* | 604373 | {Colorectal cancer, susceptibility to}, 114500 (3), Somatic mutation, Autosomal dominant; Osteosarcoma, somatic, 259500 (3); {Breast cancer, susceptibility to}, 114480 (3), Somatic mutation, Autosomal dominant; {Prostate cancer, familial, susceptibility to}, 176807 (3), Somatic mutation, Autosomal dominant; Li-Fraumeni syndrome 2, 609265 (3) |
| *CHI3L1* | 601525 | {Asthma-related traits, susceptibility to, 7}, 611960 (3); {Schizophrenia, susceptibility to}, 181500 (3), Autosomal dominant |
| *CHIC2* | 604332 | {Leukemia, acute myeloid}, 601626 (3), Somatic mutation, Autosomal dominant |
| *CHIT1* | 600031 | [Chitotriosidase deficiency], 614122 (3), Autosomal recessive |
| *CHKB* | 612395 | Muscular dystrophy, congenital, megaconial type, 602541 (3), Autosomal recessive |
| *CHM* | 300390 | Choroideremia, 303100 (3), X-linked dominant |
| *CHMP1A* | 164010 | Pontocerebellar hypoplasia, type 8, 614961 (3), Autosomal recessive |
| *CHMP2B* | 609512 | Frontotemporal dementia and/or amyotrophic lateral sclerosis 7, 600795 (3), Autosomal dominant |
| *CHMP4B* | 610897 | Cataract 31, multiple types, 605387 (3), Autosomal dominant |
| *CHN1* | 118423 | Duane retraction syndrome 2, 604356 (3), Autosomal dominant |
| *CHP1* | 606988 | ?Spastic ataxia 9, autosomal recessive, 618438 (3), Autosomal recessive |
| *CHRDL1* | 300350 | Megalocornea 1, X-linked, 309300 (3), X-linked recessive |
| *CHRM3* | 118494 | Prune belly syndrome, 100100 (3), Autosomal recessive |
| *CHRNA1* | 100690 | Myasthenic syndrome, congenital, 1B, fast-channel, 608930 (3), Autosomal recessive, Autosomal dominant; Myasthenic syndrome, congenital, 1A, slow-channel, 601462 (3), Autosomal dominant; Multiple pterygium syndrome, lethal type, 253290 (3), Autosomal recessive |
| *CHRNA2* | 118502 | Epilepsy, nocturnal frontal lobe, type 4, 610353 (3), Autosomal dominant |
| *CHRNA3* | 118503 | {Lung cancer susceptibility 2}, 612052 (3); Bladder dysfunction, autonomic, with impaired pupillary reflex and secondary CAKUT, 191800 (3), Autosomal recessive |
| *CHRNA4* | 118504 | {Nicotine addiction, susceptibility to}, 188890 (3); Epilepsy, nocturnal frontal lobe, 1, 600513 (3), Autosomal dominant |
| *CHRNA5* | 118505 | {Nicotine dependence, susceptibility to}, 612052 (3); {Lung cancer susceptibility 2}, 612052 (3) |
| *CHRNB1* | 100710 | ?Myasthenic syndrome, congenital, 2C, associated with acetylcholine receptor deficiency, 616314 (3), Autosomal recessive; Myasthenic syndrome, congenital, 2A, slow-channel, 616313 (3), Autosomal dominant |
| *CHRNB2* | 118507 | Epilepsy, nocturnal frontal lobe, 3, 605375 (3) |
| *CHRND* | 100720 | ?Myasthenic syndrome, congenital, 3C, associated with acetylcholine receptor deficiency, 616323 (3), Autosomal recessive; Multiple pterygium syndrome, lethal type, 253290 (3), Autosomal recessive; Myasthenic syndrome, congenital, 3B, fast-channel, 616322 (3), Autosomal recessive; ?Myasthenic syndrome, congenital, 3A, slow-channel, 616321 (3), Autosomal dominant |
| *CHRNE* | 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 |
| *CHRNG* | 100730 | Multiple pterygium syndrome, lethal type, 253290 (3), Autosomal recessive; Escobar syndrome, 265000 (3), Autosomal recessive |
| *CHST11* | 610128 | ?Osteochondrodysplasia, brachydactyly, and overlapping malformed digits, 618167 (3), Autosomal recessive |
| *CHST14* | 608429 | Ehlers-Danlos syndrome, musculocontractural type 1, 601776 (3), Autosomal recessive |
| *CHST3* | 603799 | Spondyloepiphyseal dysplasia with congenital joint dislocations, 143095 (3), Autosomal recessive |
| *CHST6* | 605294 | Macular corneal dystrophy, 217800 (3), Autosomal recessive |
| *CHST8* | 610190 | ?Peeling skin syndrome 3, 616265 (3), Autosomal recessive |
| *CHSY1* | 608183 | Temtamy preaxial brachydactyly syndrome, 605282 (3), Autosomal recessive |
| *CHUK* | 600664 | ?Popliteal pterygium syndrome, Bartsocas-Papas type 2, 619339 (3); Cocoon syndrome, 613630 (3) |
| *CIB1* | 602293 | Epidermodysplasia verruciformis 3, 618267 (3), Autosomal recessive |
| *CIB2* | 605564 | Deafness, autosomal recessive 48, 609439 (3), Autosomal recessive; Usher syndrome, type IJ, 614869 (3), Autosomal recessive |
| *CIBAR1 (FAM92A)* | 617273 | ?Polydactyly, postaxial, type A9, 618219 (3), Autosomal recessive |
| *CIC* | 612082 | Mental retardation, autosomal dominant 45, 617600 (3), Autosomal dominant |
| *CIDEC* | 612120 | ?Lipodystrophy, familial partial, type 5, 615238 (3), Autosomal recessive |
| *CIITA* | 600005 | {Rheumatoid arthritis, susceptibility to}, 180300 (3); Bare lymphocyte syndrome, type II, complementation group A, 209920 (3), Autosomal recessive |
| *CILK1 (ICK)* | 612325 | {Epilepsy, juvenile myoclonic, susceptibility to, 10}, 617924 (3), Autosomal dominant; Endocrine-cerebroosteodysplasia, 612651 (3), Autosomal recessive |
| *CILP* | 603489 | {Lumbar disc disease, susceptibility to}, 603932 (3) |
| *CISD2* | 611507 | Wolfram syndrome 2, 604928 (3), Autosomal recessive |
| *CISH* | 602441 | {Malaria, susceptibility to}, 611162 (3); {Bacteremia, susceptibility to}, 614383 (3); {Tuberculosis, susceptibility to}, 607948 (3) |
| *CIT* | 605629 | Microcephaly 17, primary, autosomal recessive, 617090 (3), Autosomal recessive |
| *CITED2* | 602937 | Atrial septal defect 8, 614433 (3), Autosomal dominant; Ventricular septal defect 2, 614431 (3), Autosomal dominant |
| *CKAP2L* | 616174 | Filippi syndrome, 272440 (3), Autosomal recessive |
| *CLCC1* | 617539 | Retinitis pigmentosa 32, 609913 (3), Autosomal recessive |
| *CLCF1* | 607672 | Cold-induced sweating syndrome 2, 610313 (3), Autosomal recessive |
| *CLCN1* | 118425 | Myotonia levior, recessive (3); Myotonia congenita, recessive, 255700 (3), Autosomal recessive; Myotonia congenita, dominant, 160800 (3), Autosomal dominant |
| *CLCN2* | 600570 | Leukoencephalopathy with ataxia, 615651 (3), Autosomal recessive; Hyperaldosteronism, familial, type II, 605635 (3), Autosomal dominant; {Epilepsy, juvenile myoclonic, susceptibility to, 8}, 607628 (3), Autosomal dominant; {Epilepsy, juvenile absence, susceptibility to, 2}, 607628 (3), Autosomal dominant; {Epilepsy, idiopathic generalized, susceptibility to, 11}, 607628 (3), Autosomal dominant |
| *CLCN4* | 302910 | Raynaud-Claes syndrome, 300114 (3), X-linked dominant |
| *CLCN5* | 300008 | Proteinuria, low molecular weight, with hypercalciuric nephrocalcinosis, 308990 (3), X-linked recessive; Hypophosphatemic rickets, 300554 (3), X-linked recessive; Dent disease 1, 300009 (3), X-linked recessive; Nephrolithiasis, type I, 310468 (3), X-linked recessive |
| *CLCN6* | 602726 | Neurodegeneration, childhood-onset, hypotonia, respiratory insufficiency and brain imaging abnormalities, 619173 (3), Autosomal dominant |
| *CLCN7* | 602727 | Hypopigmentation, organomegaly, and delayed myelination and development, 618541 (3), Autosomal dominant; Osteopetrosis, autosomal recessive 4, 611490 (3), Autosomal recessive; Osteopetrosis, autosomal dominant 2, 166600 (3), Autosomal dominant |
| *CLCNKA* | 602024 | Bartter syndrome, type 4b, digenic, 613090 (3), Digenic recessive |
| *CLCNKB* | 602023 | Bartter syndrome, type 3, 607364 (3), Autosomal recessive; Bartter syndrome, type 4b, digenic, 613090 (3), Digenic recessive |
| *CLDN1* | 603718 | Ichthyosis, leukocyte vacuoles, alopecia, and sclerosing cholangitis, 607626 (3), Autosomal recessive |
| *CLDN10* | 617579 | HELIX syndrome, 617671 (3), Autosomal recessive |
| *CLDN11* | 601326 | Leukodystrophy, hypomyelinating, 22, 619328 (3), Autosomal dominant |
| *CLDN14* | 605608 | Deafness, autosomal recessive 29, 614035 (3), Autosomal recessive |
| *CLDN16* | 603959 | Hypomagnesemia 3, renal, 248250 (3), Autosomal recessive |
| *CLDN19* | 610036 | Hypomagnesemia 5, renal, with ocular involvement, 248190 (3), Autosomal recessive |
| *CLDN2* | 300520 | ?Azoospermia, obstructive, with nephrolithiasis, 301060 (3), X-linked recessive |
| *CLDN9* | 615799 | ?Deafness, autosomal recessive 116, 619093 (3), Autosomal recessive |
| *CLEC1A* | 606782 | {Aspergillosis, susceptibility to}, 614079 (3) |
| *CLEC7A* | 606264 | Candidiasis, familial, 4, autosomal recessive, 613108 (3), Autosomal recessive; {Aspergillosis, susceptibility to}, 614079 (3) |
| *CLIC2* | 300138 | ?Mental retardation, X-linked, syndromic 32, 300886 (3), X-linked recessive |
| *CLIC5* | 607293 | ?Deafness, autosomal recessive 103, 616042 (3), Autosomal recessive |
| *CLMP* | 611693 | Congenital short bowel syndrome, 615237 (3), Autosomal recessive |
| *CLN3* | 607042 | Ceroid lipofuscinosis, neuronal, 3, 204200 (3), Autosomal recessive |
| *CLN5* | 608102 | Ceroid lipofuscinosis, neuronal, 5, 256731 (3), Autosomal recessive |
| *CLN6* | 606725 | Ceroid lipofuscinosis, neuronal, Kufs type, adult onset, 204300 (3), Autosomal recessive; Ceroid lipofuscinosis, neuronal, 6, 601780 (3), Autosomal recessive |
| *CLN8* | 607837 | Ceroid lipofuscinosis, neuronal, 8, Northern epilepsy variant, 610003 (3), Autosomal recessive; Ceroid lipofuscinosis, neuronal, 8, 600143 (3), Autosomal recessive |
| *CLP1* | 608757 | Pontocerebellar hypoplasia, type 10, 615803 (3), Autosomal recessive |
| *CLPB* | 616254 | 3-methylglutaconic aciduria, type VII, with cataracts, neurologic involvement and neutropenia, 616271 (3), Autosomal recessive |
| *CLPP* | 601119 | Perrault syndrome 3, 614129 (3), Autosomal recessive |
| *CLPX* | 615611 | ?Protoporphyria, erythropoietic, 2, 618015 (3), Autosomal dominant |
| *CLRN1* | 606397 | Usher syndrome, type 3A, 276902 (3), Autosomal recessive; Retinitis pigmentosa 61, 614180 (3) |
| *CLRN2* | 618988 | ?Deafness, autosomal recessive 117, 619174 (3), Autosomal recessive |
| *CLTC* | 118955 | Mental retardation, autosomal dominant 56, 617854 (3), Autosomal dominant |
| *CNBP* | 116955 | Myotonic dystrophy 2, 602668 (3), Autosomal dominant |
| *CNGA1* | 123825 | Retinitis pigmentosa 49, 613756 (3) |
| *CNGA3* | 600053 | Achromatopsia 2, 216900 (3), Autosomal recessive |
| *CNGB1* | 600724 | Retinitis pigmentosa 45, 613767 (3), Autosomal recessive |
| *CNGB3* | 605080 | Achromatopsia 3, 262300 (3), Autosomal recessive |
| *CNKSR2* | 300724 | Mental retardation, X-linked, syndromic, Houge type, 301008 (3), X-linked |
| *CNNM2* | 607803 | Hypomagnesemia 6, renal, 613882 (3), Autosomal dominant; Hypomagnesemia, seizures, and mental retardation, 616418 (3), Autosomal recessive, Autosomal dominant |
| *CNNM4* | 607805 | Jalili syndrome, 217080 (3), Autosomal recessive |
| *CNOT1* | 604917 | Vissers-Bodmer syndrome, 619033 (3), Autosomal dominant; Holoprosencephaly 12, with or without pancreatic agenesis, 618500 (3), Autosomal dominant |
| *CNOT2* | 604909 | Intellectual developmental disorder with nasal speech, dysmorphic facies, and variable skeletal anomalies, 618608 (3), Autosomal dominant |
| *CNOT3* | 604910 | Intellectual developmental disorder with speech delay, autism, and dysmorphic facies, 618672 (3), Autosomal dominant |
| *CNP* | 123830 | ?Leukodystrophy, hypomyelinating, 20, 619071 (3), Autosomal recessive |
| *CNPY3* | 610774 | Developmental and epileptic encephalopathy 60, 617929 (3), Autosomal recessive |
| *CNTN1* | 600016 | ?Myopathy, congenital, Compton-North, 612540 (3), Autosomal recessive |
| *CNTN2* | 190197 | ?Epilepsy, myoclonic, familial adult, 5, 615400 (3), Autosomal recessive |
| *CNTNAP1* | 602346 | Lethal congenital contracture syndrome 7, 616286 (3), Autosomal recessive; Hypomyelinating neuropathy, congenital, 3, 618186 (3), Autosomal recessive |
| *CNTNAP2* | 604569 | Pitt-Hopkins like syndrome 1, 610042 (3), Autosomal recessive; Cortical dysplasia-focal epilepsy syndrome, 610042 (3), Autosomal recessive; {Autism susceptibility 15}, 612100 (3) |
| *COA3* | 614775 | ?Mitochondrial complex IV deficiency, nuclear type 14, 619058 (3), Autosomal recessive |
| *COA5* | 613920 | ?Mitochondrial complex IV, deficiency, nuclear type 9, 616500 (3), Autosomal recessive |
| *COA6* | 614772 | Mitochondrial complex IV deficiency, nuclear type 13, 616501 (3), Autosomal recessive |
| *COA7* | 615623 | Spinocerebellar ataxia, autosomal recessive, with axonal neuropathy 3, 618387 (3), Autosomal recessive |
| *COA8 (APOPT1)* | 616003 | Mitochondrial complex IV deficiency, nuclear type 17, 619061 (3), Autosomal recessive |
| *COASY* | 609855 | Pontocerebellar hypoplasia, type 12, 618266 (3), Autosomal recessive; Neurodegeneration with brain iron accumulation 6, 615643 (3), Autosomal recessive |
| *COCH* | 603196 | Deafness, autosomal dominant 9, 601369 (3), Autosomal dominant; ?Deafness, autosomal recessive 110, 618094 (3), Autosomal recessive |
| *COG1* | 606973 | Congenital disorder of glycosylation, type IIg, 611209 (3), Autosomal recessive |
| *COG2* | 606974 | ?Congenital disorder of glycosylation, type IIq, 617395 (3), Autosomal recessive |
| *COG4* | 606976 | Congenital disorder of glycosylation, type IIj, 613489 (3), Autosomal recessive; Saul-Wilson syndrome, 618150 (3), Autosomal dominant |
| *COG5* | 606821 | Congenital disorder of glycosylation, type IIi, 613612 (3), Autosomal recessive |
| *COG6* | 606977 | Shaheen syndrome, 615328 (3), Autosomal recessive; Congenital disorder of glycosylation, type IIl, 614576 (3), Autosomal recessive |
| *COG7* | 606978 | Congenital disorder of glycosylation, type IIe, 608779 (3), Autosomal recessive |
| *COG8* | 606979 | Congenital disorder of glycosylation, type IIh, 611182 (3) |
| *COL10A1* | 120110 | Metaphyseal chondrodysplasia, Schmid type, 156500 (3), Autosomal dominant |
| *COL11A1* | 120280 | Fibrochondrogenesis 1, 228520 (3), Autosomal recessive; 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) |
| *COL11A2* | 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 |
| *COL12A1* | 120320 | Bethlem myopathy 2, 616471 (3), Autosomal dominant; ?Ullrich congenital muscular dystrophy 2, 616470 (3), Autosomal recessive |
| *COL13A1* | 120350 | Myasthenic syndrome, congenital, 19, 616720 (3), Autosomal recessive |
| *COL17A1* | 113811 | Epithelial recurrent erosion dystrophy, 122400 (3), Autosomal dominant; Epidermolysis bullosa, junctional, localisata variant, 226650 (3), Autosomal recessive; Epidermolysis bullosa, junctional, non-Herlitz type, 226650 (3), Autosomal recessive |
| *COL18A1* | 120328 | Knobloch syndrome, type 1, 267750 (3), Autosomal recessive; Glaucoma, primary closed-angle, 618880 (3), Autosomal dominant |
| *COL1A1* | 120150 | Osteogenesis imperfecta, type II, 166210 (3), Autosomal dominant; Caffey disease, 114000 (3), Autosomal dominant; Ehlers-Danlos syndrome, arthrochalasia type, 1, 130060 (3), Autosomal dominant; Osteogenesis imperfecta, type I, 166200 (3), Autosomal dominant; {Bone mineral density variation QTL, osteoporosis}, 166710 (3), Autosomal dominant; Combined osteogenesis imperfecta and Ehlers-Danlos syndrome 1, 619115 (3), Autosomal dominant; Osteogenesis imperfecta, type IV, 166220 (3), Autosomal dominant; Osteogenesis imperfecta, type III, 259420 (3), Autosomal dominant |
| *COL1A2* | 120160 | Osteogenesis imperfecta, type III, 259420 (3), Autosomal dominant; {Osteoporosis, postmenopausal}, 166710 (3), Autosomal dominant; Ehlers-Danlos syndrome, arthrochalasia type, 2, 617821 (3), Autosomal dominant; Combined osteogenesis imperfecta and Ehlers-Danlos syndrome 2, 619120 (3), Autosomal dominant; Ehlers-Danlos syndrome, cardiac valvular type, 225320 (3), Autosomal recessive; Osteogenesis imperfecta, type IV, 166220 (3), Autosomal dominant; Osteogenesis imperfecta, type II, 166210 (3), Autosomal dominant |
| *COL25A1* | 610004 | Fibrosis of extraocular muscles, congenital, 5, 616219 (3), Autosomal recessive |
| *COL27A1* | 608461 | Steel syndrome, 615155 (3), Autosomal recessive |
| *COL2A1* | 120140 | ?Vitreoretinopathy with phalangeal epiphyseal dysplasia, 619248 (3), Autosomal dominant; Czech dysplasia, 609162 (3), Autosomal dominant; Achondrogenesis, type II or hypochondrogenesis, 200610 (3), Autosomal dominant; Spondyloperipheral dysplasia, 271700 (3), Autosomal dominant; SMED Strudwick type, 184250 (3), Autosomal dominant; Stickler sydrome, type I, nonsyndromic ocular, 609508 (3), Autosomal dominant; ?Epiphyseal dysplasia, multiple, with myopia and deafness, 132450 (3), Autosomal dominant; SED congenita, 183900 (3), Autosomal dominant; Kniest dysplasia, 156550 (3), Autosomal dominant; Osteoarthritis with mild chondrodysplasia, 604864 (3), Autosomal dominant; Stickler syndrome, type I, 108300 (3), Autosomal dominant; Platyspondylic skeletal dysplasia, Torrance type, 151210 (3), Autosomal dominant; Spondyloepiphyseal dysplasia, Stanescu type, 616583 (3), Autosomal dominant; Avascular necrosis of the femoral head, 608805 (3), Autosomal dominant; Legg-Calve-Perthes disease, 150600 (3), Autosomal dominant |
| *COL3A1* | 120180 | Ehlers-Danlos syndrome, vascular type, 130050 (3), Autosomal dominant; Polymicrogyria with or without vascular-type EDS, 618343 (3), Autosomal recessive |
| *COL4A1* | 120130 | ?Retinal arteries, tortuosity of, 180000 (3), Autosomal dominant; {Hemorrhage, intracerebral, susceptibility to}, 614519 (3); Angiopathy, hereditary, with nephropathy, aneurysms, and muscle cramps, 611773 (3), Autosomal dominant; Microangiopathy and leukoencephalopathy, pontine, autosomal dominant, 618564 (3), Autosomal dominant; Brain small vessel disease with or without ocular anomalies, 175780 (3), Autosomal dominant |
| *COL4A2* | 120090 | Brain small vessel disease 2, 614483 (3), Autosomal dominant; {Hemorrhage, intracerebral, susceptibility to}, 614519 (3) |
| *COL4A3* | 120070 | Hematuria, benign familial, 141200 (3), Autosomal dominant; Alport syndrome 3, autosomal dominant, 104200 (3), Autosomal dominant; Alport syndrome 2, autosomal recessive, 203780 (3), Autosomal recessive |
| *COL4A4* | 120131 | Hematuria, familial benign, 141200 (3), Autosomal dominant; Alport syndrome 2, autosomal recessive, 203780 (3), Autosomal recessive |
| *COL4A5* | 303630 | Alport syndrome 1, X-linked, 301050 (3), X-linked dominant |
| *COL4A6* | 303631 | ?Deafness, X-linked 6, 300914 (3), X-linked recessive |
| *COL5A1* | 120215 | Ehlers-Danlos syndrome, classic type, 1, 130000 (3), Autosomal dominant; Fibromuscular dysplasia, multifocal, 619329 (3), Autosomal dominant |
| *COL5A2* | 120190 | Ehlers-Danlos syndrome, classic type, 2, 130010 (3), Autosomal dominant |
| *COL6A1* | 120220 | Bethlem myopathy 1, 158810 (3), Autosomal recessive, Autosomal dominant; Ullrich congenital muscular dystrophy 1, 254090 (3), Autosomal recessive, Autosomal dominant |
| *COL6A2* | 120240 | Bethlem myopathy 1, 158810 (3), Autosomal recessive, Autosomal dominant; ?Myosclerosis, congenital, 255600 (3), Autosomal recessive; Ullrich congenital muscular dystrophy 1, 254090 (3), Autosomal recessive, Autosomal dominant |
| *COL6A3* | 120250 | Ullrich congenital muscular dystrophy 1, 254090 (3), Autosomal recessive, Autosomal dominant; Dystonia 27, 616411 (3), Autosomal recessive; Bethlem myopathy 1, 158810 (3), Autosomal recessive, Autosomal dominant |
| *COL7A1* | 120120 | EBD, localisata variant (3); Epidermolysis bullosa, pretibial, 131850 (3), Autosomal recessive, Autosomal dominant; Transient bullous of the newborn, 131705 (3), Autosomal recessive, Autosomal dominant; EBD, Bart type, 132000 (3), Autosomal dominant; Epidermolysis bullosa dystrophica, AD, 131750 (3), Autosomal dominant; Epidermolysis bullosa pruriginosa, 604129 (3), Autosomal recessive, Autosomal dominant; EBD inversa, 226600 (3), Autosomal recessive; Epidermolysis bullosa dystrophica, AR, 226600 (3), Autosomal recessive; Toenail dystrophy, isolated, 607523 (3), Autosomal dominant |
| *COL8A2* | 120252 | Corneal dystrophy, posterior polymorphous 2, 609140 (3), Autosomal dominant; Corneal dystrophy, Fuchs endothelial, 1, 136800 (3), Autosomal dominant |
| *COL9A1* | 120210 | Stickler syndrome, type IV, 614134 (3); ?Epiphyseal dysplasia, multiple, 6, 614135 (3), Autosomal dominant |
| *COL9A2* | 120260 | Epiphyseal dysplasia, multiple, 2, 600204 (3), Autosomal dominant; ?Stickler syndrome, type V, 614284 (3), Autosomal recessive |
| *COL9A3* | 120270 | {Intervertebral disc disease, susceptibility to}, 603932 (3); Epiphyseal dysplasia, multiple, 3, with or without myopathy, 600969 (3), Autosomal dominant |
| *COLEC10* | 607620 | 3MC syndrome 3, 248340 (3), Autosomal recessive |
| *COLEC11* | 612502 | 3MC syndrome 2, 265050 (3), Autosomal recessive |
| *COLGALT1* | 617531 | Brain small vessel disease 3, 618360 (3), Autosomal recessive |
| *COLQ* | 603033 | Myasthenic syndrome, congenital, 5, 603034 (3), Autosomal recessive |
| *COMP* | 600310 | Pseudoachondroplasia, 177170 (3), Autosomal dominant; Carpal tunnel syndrome 2, 619161 (3), Autosomal dominant; Epiphyseal dysplasia, multiple, 1, 132400 (3), Autosomal dominant |
| *COMT* | 116790 | {Schizophrenia, susceptibility to}, 181500 (3), Autosomal dominant; {Panic disorder, susceptibility to}, 167870 (3), ?Autosomal dominant |
| *COPA* | 601924 | {Autoimmune interstitial lung, joint, and kidney disease}, 616414 (3), Autosomal dominant |
| *COPB1* | 600959 | Baralle-Macken syndrome, 619255 (3), Autosomal recessive |
| *COPB2* | 606990 | ?Microcephaly 19, primary, autosomal recessive, 617800 (3), Autosomal recessive |
| *COQ2* | 609825 | {Multiple system atrophy, susceptibility to}, 146500 (3), Autosomal recessive, Autosomal dominant; Coenzyme Q10 deficiency, primary, 1, 607426 (3), Autosomal recessive |
| *COQ4* | 612898 | Coenzyme Q10 deficiency, primary, 7, 616276 (3), Autosomal recessive |
| *COQ5* | 616359 | ?Coenzyme Q10 deficiency, primary, 9, 619028 (3), Autosomal recessive |
| *COQ6* | 614647 | Coenzyme Q10 deficiency, primary, 6, 614650 (3), Autosomal recessive |
| *COQ7* | 601683 | ?Coenzyme Q10 deficiency, primary, 8, 616733 (3), Autosomal recessive |
| *COQ8A* | 606980 | Coenzyme Q10 deficiency, primary, 4, 612016 (3), Autosomal recessive |
| *COQ8B* | 615567 | Nephrotic syndrome, type 9, 615573 (3), Autosomal recessive |
| *COQ9* | 612837 | Coenzyme Q10 deficiency, primary, 5, 614654 (3), Autosomal recessive |
| *CORIN* | 605236 | Preeclampsia/eclampsia 5, 614595 (3) |
| *CORO1A* | 605000 | Immunodeficiency 8, 615401 (3), Autosomal recessive |
| *COX10* | 602125 | Mitochondrial complex IV deficiency, nuclear type 3, 619046 (3) |
| *COX14* | 614478 | ?Mitochondrial complex IV deficiency, nuclear type 10, 619053 (3), Autosomal recessive |
| *COX15* | 603646 | Mitochondrial complex IV deficiency, nuclear type 6, 615119 (3), Autosomal recessive |
| *COX16* | 618064 | Mitochondrial complex IV deficiency, nuclear type 22, 619355 (3), Autosomal recessive |
| *COX20* | 614698 | Mitochondrial complex IV deficiency, nuclear type 11, 619054 (3), Autosomal recessive |
| *COX4I1* | 123864 | Mitochondrial complex IV deficiency, nuclear type 16, 619060 (3), Autosomal recessive |
| *COX4I2* | 607976 | Exocrine pancreatic insufficiency, dyserythropoietic anemia, and calvarial hyperostosis, 612714 (3), Autosomal recessive |
| *COX5A* | 603773 | ?Mitochondrial complex IV deficiency, nuclear type 20, 619064 (3), Autosomal recessive |
| *COX6A1* | 602072 | Charcot-Marie-Tooth disease, recessive intermediate D, 616039 (3), Autosomal recessive |
| *COX6A2* | 602009 | Mitochondrial complex IV deficiency, nuclear type 18, 619062 (3), Autosomal recessive |
| *COX6B1* | 124089 | Mitochondrial complex IV deficiency, nuclear type 7, 619051 (3), Autosomal recessive |
| *COX7B* | 300885 | Linear skin defects with multiple congenital anomalies 2, 300887 (3), X-linked dominant |
| *COX8A* | 123870 | ?Mitochondrial complex IV deficiency, nuclear type 15, 619059 (3), Autosomal recessive |
| *CP* | 117700 | Cerebellar ataxia, 604290 (3), Autosomal recessive; [Hypoceruloplasminemia, hereditary], 604290 (3), Autosomal recessive; Hemosiderosis, systemic, due to aceruloplasminemia, 604290 (3), Autosomal recessive |
| *CPA6* | 609562 | Febrile seizures, familial, 11, 614418 (3), Autosomal recessive; Epilepsy, familial temporal lobe, 5, 614417 (3), Autosomal recessive, Autosomal dominant |
| *CPAMD8* | 608841 | Anterior segment dysgenesis 8, 617319 (3), Autosomal recessive |
| *CPE* | 114855 | Intellectual developmental disorder and hypogonadotropic hypogonadism, 619326 (3), Autosomal recessive |
| *CPLANE1* | 614571 | Orofaciodigital syndrome VI, 277170 (3), Autosomal recessive; Joubert syndrome 17, 614615 (3), Autosomal recessive |
| *CPLX1* | 605032 | Developmental and epileptic encephalopathy 63, 617976 (3), Autosomal recessive |
| *CPN1* | 603103 | Carboxypeptidase N deficiency, 212070 (3), Autosomal recessive |
| *CPOX* | 612732 | Coproporphyria, 121300 (3), Autosomal recessive, Autosomal dominant; Harderoporphyria, 618892 (3), Autosomal recessive |
| *CPS1* | 608307 | Carbamoylphosphate synthetase I deficiency, 237300 (3), Autosomal recessive; {Pulmonary hypertension, neonatal, susceptibility to}, 615371 (3) |
| *CPSF1* | 606027 | Myopia 27, 618827 (3), Autosomal dominant |
| *CPT1A* | 600528 | CPT deficiency, hepatic, type IA, 255120 (3), Autosomal recessive |
| *CPT1C* | 608846 | ?Spastic paraplegia 73, autosomal dominant, 616282 (3), Autosomal dominant |
| *CPT2* | 600650 | {Encephalopathy, acute, infection-induced, 4, susceptibility to}, 614212 (3), Autosomal recessive, Autosomal dominant; CPT II deficiency, infantile, 600649 (3), Autosomal recessive; CPT II deficiency, lethal neonatal, 608836 (3), Autosomal recessive; CPT II deficiency, myopathic, stress-induced, 255110 (3), Autosomal recessive, Autosomal dominant |
| *CR1* | 120620 | [Blood group, Knops system], 607486 (3); {Malaria, severe, resistance to}, 611162 (3) |
| *CR2* | 120650 | {Systemic lupus erythematosus, susceptibility to, 9}, 610927 (3); Immunodeficiency, common variable, 7, 614699 (3), Autosomal recessive |
| *CRADD* | 603454 | Mental retardation, autosomal recessive 34, with variant lissencephaly, 614499 (3), Autosomal recessive |
| *CRAT* | 600184 | ?Neurodegeneration with brain iron accumulation 8, 617917 (3), Autosomal recessive |
| *CRB1* | 604210 | Leber congenital amaurosis 8, 613835 (3), Autosomal recessive; Retinitis pigmentosa-12, 600105 (3), Autosomal recessive; Pigmented paravenous chorioretinal atrophy, 172870 (3), Autosomal dominant |
| *CRB2* | 609720 | Focal segmental glomerulosclerosis 9, 616220 (3), Autosomal recessive; Ventriculomegaly with cystic kidney disease, 219730 (3), Autosomal recessive |
| *CRBN* | 609262 | Mental retardation, autosomal recessive 2, 607417 (3), Autosomal recessive |
| *CREB1* | 123810 | Histiocytoma, angiomatoid fibrous, somatic, 612160 (3) |
| *CREB3L1* | 616215 | Osteogenesis imperfecta, type XVI, 616229 (3), Autosomal recessive |
| *CREB3L3* | 611998 | Hypertriglyceridemia 2, 619324 (3), Autosomal dominant |
| *CREBBP* | 600140 | Menke-Hennekam syndrome 1, 618332 (3), Autosomal dominant; Rubinstein-Taybi syndrome 1, 180849 (3), Autosomal dominant |
| *CRELD1* | 607170 | Atrioventricular septal defect, partial, with heterotaxy syndrome, 606217 (3), Autosomal dominant; {Atrioventricular septal defect, susceptibility to, 2}, 606217 (3), Autosomal dominant |
| *CRIPT* | 604594 | Short stature with microcephaly and distinctive facies, 615789 (3), Autosomal recessive |
| *CRLF1* | 604237 | Cold-induced sweating syndrome 1, 272430 (3), Autosomal recessive |
| *CRPPA (ISPD)* | 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 |
| *CRTAP* | 605497 | Osteogenesis imperfecta, type VII, 610682 (3), Autosomal recessive |
| *CRTC1* | 607536 | Mucoepidermoid salivary gland carcinoma (3) |
| *CRX* | 602225 | Leber congenital amaurosis 7, 613829 (3); Cone-rod retinal dystrophy-2, 120970 (3), Autosomal dominant |
| *CRY1* | 601933 | {Delayed sleep phase disorder, susceptibility to}, 614163 (3), Autosomal dominant |
| *CRYAA* | 123580 | Cataract 9, multiple types, 604219 (3), Autosomal recessive, Autosomal dominant |
| *CRYAB* | 123590 | Myopathy, myofibrillar, fatal infantile hypertonic, alpha-B crystallin-related, 613869 (3), Autosomal recessive; Myopathy, myofibrillar, 2, 608810 (3), Autosomal dominant; Cataract 16, multiple types, 613763 (3), Autosomal recessive, Autosomal dominant; Cardiomyopathy, dilated, 1II, 615184 (3), Autosomal dominant |
| *CRYBA1* | 123610 | Cataract 10, multiple types, 600881 (3), Autosomal dominant |
| *CRYBA2* | 600836 | ?Cataract 42, 115900 (3), Autosomal dominant |
| *CRYBA4* | 123631 | Cataract 23, 610425 (3) |
| *CRYBB1* | 600929 | Cataract 17, multiple types, 611544 (3), Autosomal recessive, Autosomal dominant |
| *CRYBB2* | 123620 | Cataract 3, multiple types, 601547 (3), Autosomal dominant |
| *CRYBB3* | 123630 | Cataract 22, 609741 (3), Autosomal recessive, Autosomal dominant |
| *CRYGB* | 123670 | Cataract 39, multiple types, autosomal dominant, 615188 (3), Autosomal dominant |
| *CRYGC* | 123680 | Cataract 2, multiple types, 604307 (3), Autosomal dominant |
| *CRYGD* | 123690 | Cataract 4, multiple types, 115700 (3), Autosomal dominant |
| *CRYGS* | 123730 | Cataract 20, multiple types, 116100 (3), Autosomal dominant |
| *CRYM* | 123740 | Deafness, autosomal dominant 40, 616357 (3), Autosomal dominant |
| *CSF1R* | 164770 | Brain abnormalities, neurodegeneration, and dysosteosclerosis, 618476 (3), Autosomal recessive; Leukoencephalopathy, diffuse hereditary, with spheroids, 221820 (3), Autosomal dominant |
| *CSF2RA* | 306250 | Surfactant metabolism dysfunction, pulmonary, 4, 300770 (3) |
| *CSF2RB* | 138981 | Surfactant metabolism dysfunction, pulmonary, 5, 614370 (3), Autosomal recessive |
| *CSF3R* | 138971 | Neutropenia, severe congenital, 7, autosomal recessive, 617014 (3), Autosomal recessive |
| *CSGALNACT1* | 616615 | Skeletal dysplasia, mild, with joint laxity and advanced bone age, 618870 (3), Autosomal recessive |
| *CSH1* | 150200 | [Placental lactogen deficiency] (1) |
| *CSNK1D* | 600864 | Advanced sleep-phase syndrome, familial, 2, 615224 (3), Autosomal dominant |
| *CSNK2A1* | 115440 | Okur-Chung neurodevelopmental syndrome, 617062 (3), Autosomal dominant |
| *CSNK2B* | 115441 | Poirier-Bienvenu neurodevelopmental syndrome, 618732 (3), Autosomal dominant |
| *CSPP1* | 611654 | Joubert syndrome 21, 615636 (3), Autosomal recessive |
| *CSRP3* | 600824 | ?Cardiomyopathy, dilated, 1M, 607482 (3); Cardiomyopathy, hypertrophic, 12, 612124 (3), Autosomal dominant |
| *CST3* | 604312 | {Macular degeneration, age-related, 11}, 611953 (3); Cerebral amyloid angiopathy, 105150 (3), Autosomal dominant |
| *CST6* | 601891 | ?Ectodermal dysplasia 15, hypohidrotic/hair type, 618535 (3), Autosomal recessive |
| *CSTA* | 184600 | Peeling skin syndrome 4, 607936 (3), Autosomal recessive |
| *CSTB* | 601145 | Epilepsy, progressive myoclonic 1A (Unverricht and Lundborg), 254800 (3), Autosomal recessive |
| *CTBP1* | 602618 | Hypotonia, ataxia, developmental delay, and tooth enamel defect syndrome, 617915 (3), Autosomal dominant |
| *CTC1* | 613129 | Cerebroretinal microangiopathy with calcifications and cysts, 612199 (3), Autosomal recessive |
| *CTCF* | 604167 | Mental retardation, autosomal dominant 21, 615502 (3), Autosomal dominant |
| *CTDP1* | 604927 | Congenital cataracts, facial dysmorphism, and neuropathy, 604168 (3), Autosomal recessive |
| *CTH* | 607657 | Cystathioninuria, 219500 (3), Autosomal recessive |
| *CTHRC1* | 610635 | Barrett esophagus/esophageal adenocarcinoma, 614266 (3) |
| *CTLA4* | 123890 | Autoimmune lymphoproliferative syndrome, type V, 616100 (3), Autosomal dominant; {Diabetes mellitus, insulin-dependent, 12}, 601388 (3); {Celiac disease, susceptibility to, 3}, 609755 (3); {Hashimoto thyroiditis}, 140300 (3), Autosomal dominant; {Systemic lupus erythematosus, susceptibility to}, 152700 (3), Autosomal dominant |
| *CTNNA1* | 116805 | Macular dystrophy, patterned, 2, 608970 (3), Autosomal dominant |
| *CTNNA2* | 114025 | Cortical dysplasia, complex, with other brain malformations 9, 618174 (3), Autosomal recessive |
| *CTNNA3* | 607667 | Arrhythmogenic right ventricular dysplasia, familial, 13, 615616 (3), Autosomal dominant |
| *CTNNB1* | 116806 | Exudative vitreoretinopathy 7, 617572 (3), Autosomal dominant; Pilomatricoma, somatic, 132600 (3); Colorectal cancer, somatic, 114500 (3); Neurodevelopmental disorder with spastic diplegia and visual defects, 615075 (3), Autosomal dominant; Medulloblastoma, somatic, 155255 (3); Ovarian cancer, somatic, 167000 (3); Hepatocellular carcinoma, somatic, 114550 (3) |
| *CTNND1* | 601045 | Blepharocheilodontic syndrome 2, 617681 (3), Autosomal dominant |
| *CTNS* | 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 |
| *CTPS1* | 123860 | Immunodeficiency 24, 615897 (3), Autosomal recessive |
| *CTRC* | 601405 | {Pancreatitis, chronic, susceptibility to}, 167800 (3), Autosomal dominant |
| *CTSA* | 613111 | Galactosialidosis, 256540 (3), Autosomal recessive |
| *CTSB* | 116810 | Keratolytic winter erythema, 148370 (4), Autosomal dominant |
| *CTSC* | 602365 | Periodontitis 1, juvenile, 170650 (3), Autosomal recessive; Haim-Munk syndrome, 245010 (3), Autosomal recessive; Papillon-Lefevre syndrome, 245000 (3), Autosomal recessive |
| *CTSD* | 116840 | Ceroid lipofuscinosis, neuronal, 10, 610127 (3), Autosomal recessive |
| *CTSF* | 603539 | Ceroid lipofuscinosis, neuronal, 13, Kufs type, 615362 (3), Autosomal recessive |
| *CTSK* | 601105 | Pycnodysostosis, 265800 (3), Autosomal recessive |
| *CTU2* | 617057 | Microcephaly, facial dysmorphism, renal agenesis, and ambiguous genitalia syndrome, 618142 (3), Autosomal recessive |
| *CUBN* | 602997 | [Proteinuria, chronic benign], 618884 (3), Autosomal recessive; Imerslund-Grasbeck syndrome 1, 261100 (3), Autosomal recessive |
| *CUL3* | 603136 | Neurodevelopmental disorder with or without autism or seizures, 619239 (3), Autosomal dominant; Pseudohypoaldosteronism, type IIE, 614496 (3), Autosomal dominant |
| *CUL4B* | 300304 | Mental retardation, X-linked, syndromic 15 (Cabezas type), 300354 (3), X-linked recessive |
| *CUL7* | 609577 | 3-M syndrome 1, 273750 (3), Autosomal recessive |
| *CUX1* | 116896 | Global developmental delay with or without impaired intellectual development, 618330 (3), Autosomal dominant |
| *CUX2* | 610648 | Developmental and epileptic encephalopathy 67, 618141 (3), Autosomal dominant |
| *CWC27* | 617170 | Retinitis pigmentosa with or without skeletal anomalies, 250410 (3), Autosomal recessive |
| *CWF19L1* | 616120 | Spinocerebellar ataxia, autosomal recessive 17, 616127 (3), Autosomal recessive |
| *CX3CR1* | 601470 | {Rapid progression to AIDS from HIV1 infection}, 609423 (3); {Macular degeneration, age-related, 12}, 613784 (3); {Coronary artery disease, resistance to}, 607339 (3) |
| *CXCL12* | 600835 | {AIDS, resistance to}, 609423 (3) |
| *CXCR1* | 146929 | {AIDS, slow progression to}, 609423 (3) |
| *CXCR2* | 146928 | ?WHIM syndrome 2, 619407 (3) |
| *CXCR4* | 162643 | WHIM syndrome 1, 193670 (3), Autosomal dominant; Myelokathexis, isolated, 193670 (3), Autosomal dominant |
| *CYB561* | 600019 | Orthostatic hypotension 2, 618182 (3), Autosomal recessive |
| *CYB5A* | 613218 | Methemoglobinemia and ambiguous genitalia, 250790 (3), Autosomal recessive |
| *CYB5R3* | 613213 | Methemoglobinemia, type I, 250800 (3), Autosomal recessive; Methemoglobinemia, type II, 250800 (3), Autosomal recessive |
| *CYBA* | 608508 | Chronic granulomatous disease 4, autosomal recessive, 233690 (3), Autosomal recessive |
| *CYBB* | 300481 | Immunodeficiency 34, mycobacteriosis, X-linked, 300645 (3), X-linked recessive; Chronic granulomatous disease, X-linked, 306400 (3), X-linked recessive |
| *CYBC1* | 618334 | Chronic granulomatous disease 5, autosomal recessive, 618935 (3), Autosomal recessive |
| *CYC1* | 123980 | Mitochondrial complex III deficiency, nuclear type 6, 615453 (3), Autosomal recessive |
| *CYCS* | 123970 | Thrombocytopenia 4, 612004 (3), Autosomal dominant |
| *CYFIP2* | 606323 | Developmental and epileptic encephalopathy 65, 618008 (3), Autosomal dominant |
| *CYLD* | 605018 | Brooke-Spiegler syndrome, 605041 (3), Autosomal dominant; Cylindromatosis, familial, 132700 (3), Autosomal dominant; Trichoepithelioma, multiple familial, 1, 601606 (3), Autosomal dominant; ?Frontotemporal dementia and/or amyotrophic lateral sclerosis 8, 619132 (3) |
| *CYP11A1* | 118485 | Adrenal insufficiency, congenital, with 46XY sex reversal, partial or complete, 613743 (3) |
| *CYP11B1* | 610613 | Aldosteronism, glucocorticoid-remediable, 103900 (3), Autosomal dominant; Adrenal hyperplasia, congenital, due to 11-beta-hydroxylase deficiency, 202010 (3), Autosomal recessive |
| *CYP11B2* | 124080 | Hypoaldosteronism, congenital, due to CMO I deficiency, 203400 (3), Autosomal recessive; Aldosterone to renin ratio raised (3); {Low renin hypertension, susceptibility to} (3); Hypoaldosteronism, congenital, due to CMO II deficiency, 610600 (3), Autosomal recessive |
| *CYP17A1* | 609300 | 17,20-lyase deficiency, isolated, 202110 (3), Autosomal recessive; 17-alpha-hydroxylase/17,20-lyase deficiency, 202110 (3), Autosomal recessive |
| *CYP19A1* | 107910 | Aromatase deficiency, 613546 (3); Aromatase excess syndrome, 139300 (3), Autosomal dominant |
| *CYP1B1* | 601771 | Glaucoma 3A, primary open angle, congenital, juvenile, or adult onset, 231300 (3), Autosomal recessive; Anterior segment dysgenesis 6, multiple subtypes, 617315 (3), Autosomal recessive |
| *CYP21A2* | 613815 | Hyperandrogenism, nonclassic type, due to 21-hydroxylase deficiency, 201910 (3), Autosomal recessive; Adrenal hyperplasia, congenital, due to 21-hydroxylase deficiency, 201910 (3), Autosomal recessive |
| *CYP24A1* | 126065 | Hypercalcemia, infantile, 1, 143880 (3), Autosomal recessive |
| *CYP26B1* | 605207 | Craniosynostosis with radiohumeral fusions and other skeletal and craniofacial anomalies, 614416 (3) |
| *CYP26C1* | 608428 | Focal facial dermal dysplasia 4, 614974 (3), Autosomal recessive |
| *CYP27A1* | 606530 | Cerebrotendinous xanthomatosis, 213700 (3), Autosomal recessive |
| *CYP27B1* | 609506 | Vitamin D-dependent rickets, type I, 264700 (3), Autosomal recessive |
| *CYP2A6* | 122720 | {Lung cancer, resistance to}, 211980 (3), Somatic mutation, Autosomal dominant; Coumarin resistance, 122700 (3), Autosomal dominant; {Nicotine addiction, protection from}, 188890 (3) |
| *CYP2B6* | 123930 | {Efavirenz central nervous system toxicity, susceptibility to}, 614546 (3); Efavirenz, poor metabolism of, 614546 (3) |
| *CYP2C19* | 124020 | Proguanil poor metabolizer, 609535 (3), Autosomal recessive; Mephenytoin poor metabolizer, 609535 (3), Autosomal recessive; Clopidogrel, impaired responsiveness to, 609535 (3), Autosomal recessive; Omeprazole poor metabolizer, 609535 (3), Autosomal recessive |
| *CYP2C8* | 601129 | {Drug metabolism, altered, CYP2C8-related}, 618018 (3) |
| *CYP2C9* | 601130 | Warfarin sensitivity, 122700 (3), Autosomal dominant; Tolbutamide poor metabolizer (3) |
| *CYP2D6* | 124030 | {Codeine sensitivity}, 608902 (3), Autosomal recessive; {Debrisoquine sensitivity}, 608902 (3), Autosomal recessive |
| *CYP2R1* | 608713 | Rickets due to defect in vitamin D 25-hydroxylation deficiency, 600081 (3), Autosomal recessive |
| *CYP2U1* | 610670 | Spastic paraplegia 56, autosomal recessive, 615030 (3), Autosomal recessive |
| *CYP3A4* | 124010 | Vitamin D-dependent rickets, type 3, 619073 (3), Autosomal dominant |
| *CYP3A5* | 605325 | {Hypertension, salt-sensitive essential, susceptibility to}, 145500 (3), Multifactorial |
| *CYP4F22* | 611495 | Ichthyosis, congenital, autosomal recessive 5, 604777 (3), Autosomal recessive |
| *CYP4V2* | 608614 | Bietti crystalline corneoretinal dystrophy, 210370 (3), Autosomal recessive |
| *CYP7B1* | 603711 | Spastic paraplegia 5A, autosomal recessive, 270800 (3), Autosomal recessive; Bile acid synthesis defect, congenital, 3, 613812 (3), Autosomal recessive |
| *D2HGDH* | 609186 | D-2-hydroxyglutaric aciduria, 600721 (3), Autosomal recessive |
| *DAAM2* | 606627 | Nephrotic syndrome, type 24, 619263 (3), Autosomal recessive |
| *DAB1* | 603448 | Spinocerebellar ataxia 37, 615945 (3), Autosomal dominant |
| *DACT1* | 607861 | ?Townes-Brocks syndrome 2, 617466 (3), Autosomal dominant |
| *DAG1* | 128239 | Muscular dystrophy-dystroglycanopathy (congenital with brain and eye anomalies), type A, 9, 616538 (3), Autosomal recessive; Muscular dystrophy-dystroglycanopathy (limb-girdle), type C, 9, 613818 (3), Autosomal recessive |
| *DALRD3* | 618904 | ?Developmental and epileptic encephalopathy 86, 618910 (3), Autosomal recessive |
| *DAOA* | 607408 | {Schizophrenia}, 181500 (2), Autosomal dominant |
| *DARS1 (DARS)* | 603084 | Hypomyelination with brainstem and spinal cord involvement and leg spasticity, 615281 (3), Autosomal recessive |
| *DARS2* | 610956 | Leukoencephalopathy with brain stem and spinal cord involvement and lactate elevation, 611105 (3), Autosomal recessive |
| *DAZL* | 601486 | {Spermatogenic failure, susceptibility to} (3) |
| *DBH* | 609312 | Orthostatic hypotension 1, due to DBH deficiency, 223360 (3), Autosomal recessive |
| *DBR1* | 607024 | {Encephalitis, acute, infection (viral)-induced, susceptibility to, 11}, 619441 (3), Autosomal recessive |
| *DBT* | 248610 | Maple syrup urine disease, type II, 248600 (3), Autosomal recessive |
| *DCAF17* | 612515 | Woodhouse-Sakati syndrome, 241080 (3), Autosomal recessive |
| *DCAF8* | 615820 | ?Giant axonal neuropathy 2, autosomal dominant, 610100 (3), Autosomal dominant |
| *DCC* | 120470 | Mirror movements 1 and/or agenesis of the corpus callosum, 157600 (3), Autosomal dominant; Esophageal carcinoma, somatic, 133239 (3); Colorectal cancer, somatic, 114500 (3); Gaze palsy, familial horizontal, with progressive scoliosis, 2, 617542 (3), Autosomal recessive |
| *DCDC2* | 605755 | Nephronophthisis 19, 616217 (3), Autosomal recessive; ?Deafness, autosomal recessive 66, 610212 (3), Autosomal recessive; Sclerosing cholangitis, neonatal, 617394 (3), Autosomal recessive |
| *DCHS1* | 603057 | Mitral valve prolapse 2, 607829 (3), Autosomal dominant; Van Maldergem syndrome 1, 601390 (3), Autosomal recessive |
| *DCLRE1C* | 605988 | Severe combined immunodeficiency, Athabascan type, 602450 (3), Autosomal recessive; Omenn syndrome, 603554 (3), Autosomal recessive |
| *DCN* | 125255 | Corneal dystrophy, congenital stromal, 610048 (3), Autosomal dominant |
| *DCPS* | 610534 | Al-Raqad syndrome, 616459 (3), Autosomal recessive |
| *DCT* | 191275 | Oculocutaneous albinism, type VIII, 619165 (3), Autosomal recessive |
| *DCTN1* | 601143 | Neuronopathy, distal hereditary motor, type VIIB, 607641 (3), Autosomal dominant; Perry syndrome, 168605 (3), Autosomal dominant; {Amyotrophic lateral sclerosis, susceptibility to}, 105400 (3), Autosomal recessive, Autosomal dominant |
| *DCX* | 300121 | Subcortical laminal heterotopia, X-linked, 300067 (3), X-linked; Lissencephaly, X-linked, 300067 (3), X-linked |
| *DCXR* | 608347 | [Pentosuria], 260800 (3), Autosomal recessive |
| *DDB1* | 600045 | White-Kernohan syndrome, 619426 (3), Autosomal dominant |
| *DDB2* | 600811 | Xeroderma pigmentosum, group E, DDB-negative subtype, 278740 (3), Autosomal recessive |
| *DDC* | 107930 | Aromatic L-amino acid decarboxylase deficiency, 608643 (3), Autosomal recessive |
| *DDHD1* | 614603 | Spastic paraplegia 28, autosomal recessive, 609340 (3), Autosomal recessive |
| *DDHD2* | 615003 | Spastic paraplegia 54, autosomal recessive, 615033 (3), Autosomal recessive |
| *DDOST* | 602202 | ?Congenital disorder of glycosylation, type Ir, 614507 (3), Autosomal recessive |
| *DDR2* | 191311 | Warburg-Cinotti syndrome, 618175 (3), Autosomal dominant; Spondylometaepiphyseal dysplasia, short limb-hand type, 271665 (3), Autosomal recessive |
| *DDRGK1* | 616177 | Spondyloepimetaphyseal dysplasia, Shohat type, 602557 (3), Autosomal recessive |
| *DDX11* | 601150 | Warsaw breakage syndrome, 613398 (3), Autosomal recessive |
| *DDX3X* | 300160 | Intellectual developmental disorder, X-linked, syndrome, Snijders Blok type, 300958 (3), X-linked dominant, X-linked recessive |
| *DDX41* | 608170 | {Myeloproliferative/lymphoproliferative neoplasms, familial (multiple types), susceptibility to}, 616871 (3), Autosomal dominant |
| *DDX58* | 609631 | Singleton-Merten syndrome 2, 616298 (3), Autosomal dominant |
| *DDX59* | 615464 | Orofaciodigital syndrome V, 174300 (3), Autosomal recessive |
| *DDX6* | 600326 | Intellectual developmental disorder with impaired language and dysmorphic facies, 618653 (3), Autosomal dominant |
| *DEAF1* | 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 |
| *DEGS1* | 615843 | Leukodystrophy, hypomyelinating, 18, 618404 (3), Autosomal recessive |
| *DEK* | 125264 | Leukemia, acute nonlymphocytic, 125264 (2) |
| *DENND5A* | 617278 | Developmental and epileptic encephalopathy 49, 617281 (3), Autosomal recessive |
| *DEPDC5* | 614191 | Epilepsy, familial focal, with variable foci 1, 604364 (3), Autosomal dominant |
| *DES* | 125660 | Scapuloperoneal syndrome, neurogenic, Kaeser type, 181400 (3), Autosomal dominant; Cardiomyopathy, dilated, 1I, 604765 (3), Autosomal dominant; Myopathy, myofibrillar, 1, 601419 (3), Autosomal recessive, Autosomal dominant |
| *DGAT1* | 604900 | ?Diarrhea 7, protein-losing enteropathy type, 615863 (3), Autosomal recessive |
| *DGKE* | 601440 | {Hemolytic uremic syndrome, atypical, susceptibility to, 7}, 615008 (3), Autosomal recessive; Nephrotic syndrome, type 7, 615008 (3), Autosomal recessive |
| *DGUOK* | 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 |
| *DHCR24* | 606418 | Desmosterolosis, 602398 (3), Autosomal recessive |
| *DHCR7* | 602858 | Smith-Lemli-Opitz syndrome, 270400 (3), Autosomal recessive |
| *DHDDS* | 608172 | Developmental delay and seizures with or without movement abnormalities, 617836 (3), Autosomal dominant; ?Congenital disorder of glycosylation, type 1bb, 613861 (3), Autosomal recessive; Retinitis pigmentosa 59, 613861 (3), Autosomal recessive |
| *DHFR* | 126060 | Megaloblastic anemia due to dihydrofolate reductase deficiency, 613839 (3), Autosomal recessive |
| *DHH* | 605423 | 46XY gonadal dysgenesis with minifascicular neuropathy, 607080 (3), Autosomal recessive; 46XY sex reversal 7, 233420 (3), Autosomal recessive |
| *DHODH* | 126064 | Miller syndrome, 263750 (3), Autosomal recessive |
| *DHPS* | 600944 | Neurodevelopmental disorder with seizures and speech and walking impairment, 618480 (3), Autosomal recessive |
| *DHTKD1* | 614984 | ?Charcot-Marie-Tooth disease, axonal, type 2Q, 615025 (3), Autosomal dominant; Alpha-aminoadipic and alpha-ketoadipic aciduria, 204750 (3), Autosomal recessive |
| *DHX16* | 603405 | Neuromuscular disease and ocular or auditory anomalies with or without seizures, 618733 (3), Autosomal dominant |
| *DHX30* | 616423 | Neurodevelopmental disorder with severe motor impairment and absent language, 617804 (3), Autosomal dominant |
| *DHX37* | 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 |
| *DHX38* | 605584 | Retinitis pigmentosa 84, 618220 (3), Autosomal recessive |
| *DIABLO* | 605219 | Deafness, autosomal dominant 64, 614152 (3), Autosomal dominant |
| *DIAPH1* | 602121 | Deafness, autosomal dominant 1, with or without thrombocytopenia, 124900 (3), Autosomal dominant; Seizures, cortical blindness, microcephaly syndrome, 616632 (3), Autosomal recessive |
| *DIAPH2* | 300108 | ?Premature ovarian failure 2A, 300511 (3), X-linked dominant |
| *DIAPH3* | 614567 | Auditory neuropathy, autosomal dominant, 1, 609129 (3), Autosomal dominant |
| *DICER1* | 606241 | Pleuropulmonary blastoma, 601200 (3), Autosomal dominant; Goiter, multinodular 1, with or without Sertoli-Leydig cell tumors, 138800 (3), Autosomal dominant; GLOW syndrome, somatic mosaic, 618272 (3); Rhabdomyosarcoma, embryonal, 2, 180295 (3) |
| *DIP2B* | 611379 | Mental retardation, FRA12A type, 136630 (3), Autosomal dominant |
| *DIS3L2* | 614184 | Perlman syndrome, 267000 (3), Autosomal recessive |
| *DISC1* | 605210 | {Schizophrenia 9, susceptibility to}, 604906 (3) |
| *DKC1* | 300126 | Dyskeratosis congenita, X-linked, 305000 (3), X-linked recessive |
| *DLAT* | 608770 | Pyruvate dehydrogenase E2 deficiency, 245348 (3), Autosomal recessive |
| *DLC1* | 604258 | Colorectal cancer, somatic, 114500 (3) |
| *DLD* | 238331 | Dihydrolipoamide dehydrogenase deficiency, 246900 (3), Autosomal recessive |
| *DLG3* | 300189 | Mental retardation, X-linked 90, 300850 (3), X-linked recessive |
| *DLG4* | 602887 | Intellectual developmental disorder 62, 618793 (3), Autosomal dominant |
| *DLL1* | 606582 | Neurodevelopmental disorder with nonspecific brain abnormalities and with or without seizures, 618709 (3), Autosomal dominant |
| *DLL3* | 602768 | Spondylocostal dysostosis 1, autosomal recessive, 277300 (3), Autosomal recessive |
| *DLL4* | 605185 | Adams-Oliver syndrome 6, 616589 (3), Autosomal dominant |
| *DLST* | 126063 | Paragangliomas 7, 618475 (3), Autosomal dominant |
| *DLX3* | 600525 | Trichodontoosseous syndrome, 190320 (3), Autosomal dominant; Amelogenesis imperfecta, type IV, 104510 (3), Autosomal dominant |
| *DLX4* | 601911 | ?Orofacial cleft 15, 616788 (3), Autosomal dominant |
| *DLX5* | 600028 | Split-hand/foot malformation 1, 183600 (3), Autosomal dominant; ?Split-hand/foot malformation 1 with sensorineural hearing loss, 220600 (3), Autosomal recessive |
| *DMD* | 300377 | Becker muscular dystrophy, 300376 (3), X-linked recessive; Cardiomyopathy, dilated, 3B, 302045 (3), X-linked; Duchenne muscular dystrophy, 310200 (3), X-linked recessive |
| *DMGDH* | 605849 | Dimethylglycine dehydrogenase deficiency, 605850 (3), Autosomal recessive |
| *DMP1* | 600980 | Hypophosphatemic rickets, AR, 241520 (3), Autosomal recessive |
| *DMPK* | 605377 | Myotonic dystrophy 1, 160900 (3), Autosomal dominant |
| *DMXL2* | 612186 | Developmental and epileptic encephalopathy 81, 618663 (3), Autosomal recessive; ?Deafness, autosomal dominant 71, 617605 (3), Autosomal dominant; ?Polyendocrine-polyneuropathy syndrome, 616113 (3), Autosomal recessive |
| *DNA2* | 601810 | ?Seckel syndrome 8, 615807 (3), Autosomal recessive; Progressive external ophthalmoplegia with mitochondrial DNA deletions, autosomal dominant 6, 615156 (3), Autosomal dominant |
| *DNAAF1* | 613190 | Ciliary dyskinesia, primary, 13, 613193 (3), Autosomal recessive |
| *DNAAF11 (LRRC6)* | 614930 | Ciliary dyskinesia, primary, 19, 614935 (3), Autosomal recessive |
| *DNAAF2* | 612517 | Ciliary dyskinesia, primary, 10, 612518 (3) |
| *DNAAF3* | 614566 | Ciliary dyskinesia, primary, 2, 606763 (3), Autosomal recessive |
| *DNAAF4* | 608706 | {Dyslexia, susceptibility to, 1}, 127700 (3), Autosomal dominant; Ciliary dyskinesia, primary, 25, 615482 (3), Autosomal recessive |
| *DNAAF5* | 614864 | Ciliary dyskinesia, primary, 18, 614874 (3), Autosomal recessive |
| *DNAAF6 (PIH1D3)* | 300933 | Ciliary dyskinesia, primary, 36, X-linked, 300991 (3), X-linked recessive |
| *DNAH1* | 603332 | Spermatogenic failure 18, 617576 (3), Autosomal recessive; ?Ciliary dyskinesia, primary, 37, 617577 (3), Autosomal recessive |
| *DNAH11* | 603339 | Ciliary dyskinesia, primary, 7, with or without situs inversus, 611884 (3), Autosomal recessive |
| *DNAH17* | 610063 | Spermatogenic failure 39, 618643 (3), Autosomal recessive |
| *DNAH2* | 603333 | Spermatogenic failure 45, 619094 (3), Autosomal recessive |
| *DNAH5* | 603335 | Ciliary dyskinesia, primary, 3, with or without situs inversus, 608644 (3) |
| *DNAH8* | 603337 | Spermatogenic failure 46, 619095 (3), Autosomal recessive |
| *DNAH9* | 603330 | Ciliary dyskinesia, primary, 40, 618300 (3), Autosomal recessive |
| *DNAI1* | 604366 | Ciliary dyskinesia, primary, 1, with or without situs inversus, 244400 (3), Autosomal recessive |
| *DNAI2* | 605483 | Ciliary dyskinesia, primary, 9, with or without situs inversus, 612444 (3), Autosomal recessive |
| *DNAJB11* | 611341 | Polycystic kidney disease 6 with or without polycystic liver disease, 618061 (3), Autosomal dominant |
| *DNAJB13* | 610263 | Ciliary dyskinesia, primary, 34, 617091 (3), Autosomal recessive |
| *DNAJB2* | 604139 | Spinal muscular atrophy, distal, autosomal recessive, 5, 614881 (3), Autosomal recessive |
| *DNAJB6* | 611332 | Muscular dystrophy, limb-girdle, autosomal dominant 1, 603511 (3), Autosomal dominant |
| *DNAJC12* | 606060 | Hyperphenylalaninemia, mild, non-BH4-deficient, 617384 (3), Autosomal recessive |
| *DNAJC19* | 608977 | 3-methylglutaconic aciduria, type V, 610198 (3), Autosomal recessive |
| *DNAJC21* | 617048 | Bone marrow failure syndrome 3, 617052 (3), Autosomal recessive |
| *DNAJC3* | 601184 | ?Ataxia, combined cerebellar and peripheral, with hearing loss and diabetes mellitus, 616192 (3), Autosomal recessive |
| *DNAJC30* | 618202 | Leber hereditary optic neuropathy, autosomal recessive, 619382 (3), Autosomal recessive |
| *DNAJC5* | 611203 | Ceroid lipofuscinosis, neuronal, 4, Parry type, 162350 (3), Autosomal dominant |
| *DNAJC6* | 608375 | Parkinson disease 19a, juvenile-onset, 615528 (3), Autosomal recessive; Parkinson disease 19b, early-onset, 615528 (3), Autosomal recessive |
| *DNAL1* | 610062 | Ciliary dyskinesia, primary, 16, 614017 (3), Autosomal recessive |
| *DNAL4* | 610565 | ?Mirror movements 3, 616059 (3), Autosomal recessive |
| *DNASE1* | 125505 | {Systemic lupus erythematosus, susceptibility to}, 152700 (3), Autosomal dominant |
| *DNASE1L3* | 602244 | Systemic lupus erythematosus 16, 614420 (3), Autosomal recessive |
| *DNM1* | 602377 | Developmental and epileptic encephalopathy 31, 616346 (3), Autosomal dominant |
| *DNM1L* | 603850 | Optic atrophy 5, 610708 (3), Autosomal dominant; Encephalopathy, lethal, due to defective mitochondrial peroxisomal fission 1, 614388 (3), Autosomal recessive, Autosomal dominant |
| *DNM2* | 602378 | Centronuclear myopathy 1, 160150 (3), Autosomal dominant; Charcot-Marie-Tooth disease, axonal type 2M, 606482 (3), Autosomal dominant; Charcot-Marie-Tooth disease, dominant intermediate B, 606482 (3), Autosomal dominant; Lethal congenital contracture syndrome 5, 615368 (3), Autosomal recessive |
| *DNMBP* | 611282 | Cataract 48, 618415 (3), Autosomal recessive |
| *DNMT1* | 126375 | Neuropathy, hereditary sensory, type IE, 614116 (3), Autosomal dominant; Cerebellar ataxia, deafness, and narcolepsy, autosomal dominant, 604121 (3), Autosomal dominant |
| *DNMT3A* | 602769 | Tatton-Brown-Rahman syndrome, 615879 (3), Autosomal dominant; Acute myeloid leukemia, somatic, 601626 (3); Heyn-Sproul-Jackson syndrome, 618724 (3), Autosomal dominant |
| *DNMT3B* | 602900 | Immunodeficiency-centromeric instability-facial anomalies syndrome 1, 242860 (3), Autosomal recessive |
| *DOCK2* | 603122 | Immunodeficiency 40, 616433 (3), Autosomal recessive |
| *DOCK3* | 603123 | Neurodevelopmental disorder with impaired intellectual development, hypotonia, and ataxia, 618292 (3), Autosomal recessive |
| *DOCK6* | 614194 | Adams-Oliver syndrome 2, 614219 (3), Autosomal recessive |
| *DOCK7* | 615730 | Developmental and epileptic encephalopathy 23, 615859 (3), Autosomal recessive |
| *DOCK8* | 611432 | Hyper-IgE recurrent infection syndrome, autosomal recessive, 243700 (3), Autosomal recessive |
| *DOK7* | 610285 | Fetal akinesia deformation sequence 3, 618389 (3), Autosomal recessive; Myasthenic syndrome, congenital, 10, 254300 (3), Autosomal recessive |
| *DOLK* | 610746 | Congenital disorder of glycosylation, type Im, 610768 (3), Autosomal recessive |
| *DONSON* | 611428 | Microcephaly, short stature, and limb abnormalities, 617604 (3), Autosomal recessive; Microcephaly-micromelia syndrome, 251230 (3), Autosomal recessive |
| *DPAGT1* | 191350 | Myasthenic syndrome, congenital, 13, with tubular aggregates, 614750 (3), Autosomal recessive; Congenital disorder of glycosylation, type Ij, 608093 (3), Autosomal recessive |
| *DPF2* | 601671 | Coffin-Siris syndrome 7, 618027 (3), Autosomal dominant |
| *DPH1* | 603527 | Developmental delay with short stature, dysmorphic facial features, and sparse hair, 616901 (3), Autosomal recessive |
| *DPM1* | 603503 | Congenital disorder of glycosylation, type Ie, 608799 (3), Autosomal recessive |
| *DPM2* | 603564 | Congenital disorder of glycosylation, type Iu, 615042 (3), Autosomal recessive |
| *DPM3* | 605951 | ?Muscular dystrophy-dystroglycanopathy (congenital with impaired intellectual development), type B, 15, 618992 (3), Autosomal recessive; Muscular dystrophy-dystroglycanopathy (limb-girdle), type C, 15, 612937 (3), Autosomal recessive |
| *DPP6* | 126141 | Mental retardation, autosomal dominant 33, 616311 (3); {Ventricular fibrillation, paroxysmal familial, 2}, 612956 (3), Autosomal dominant |
| *DPY19L2* | 613893 | Spermatogenic failure 9, 613958 (3), Autosomal recessive |
| *DPYD* | 612779 | Dihydropyrimidine dehydrogenase deficiency, 274270 (3), Autosomal recessive; 5-fluorouracil toxicity, 274270 (3), Autosomal recessive |
| *DPYS* | 613326 | Dihydropyrimidinuria, 222748 (3), Autosomal recessive |
| *DPYSL5* | 608383 | Ritscher-Schinzel syndrome 4, 619435 (3), Autosomal dominant |
| *DRAM2* | 613360 | Cone-rod dystrophy 21, 616502 (3), Autosomal recessive |
| *DRC1* | 615288 | Ciliary dyskinesia, primary, 21, 615294 (3), Autosomal recessive |
| *DRD3* | 126451 | {Essential tremor, hereditary, 1}, 190300 (3), Autosomal dominant; {Schizophrenia, susceptibility to}, 181500 (3), Autosomal dominant |
| *DRD4* | 126452 | {Attention deficit-hyperactivity disorder}, 143465 (3), Autosomal dominant; Autonomic nervous system dysfunction (3) |
| *DRD5* | 126453 | {Blepharospasm, primary benign}, 606798 (3), Autosomal dominant; {Attention deficit-hyperactivity disorder, susceptibility to}, 143465 (3), Autosomal dominant |
| *DSC2* | 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 |
| *DSC3* | 600271 | Hypotrichosis and recurrent skin vesicles, 613102 (3), Autosomal recessive |
| *DSE* | 605942 | Ehlers-Danlos syndrome, musculocontractural type 2, 615539 (3), Autosomal recessive |
| *DSG1* | 125670 | Keratosis palmoplantaris striata I, AD, 148700 (3), Autosomal dominant; Erythroderma, congenital, with palmoplantar keratoderma, hypotrichosis, and hyper IgE, 615508 (3), Autosomal recessive |
| *DSG2* | 125671 | Cardiomyopathy, dilated, 1BB, 612877 (3); Arrhythmogenic right ventricular dysplasia 10, 610193 (3), Autosomal dominant |
| *DSG3* | 169615 | Blistering, acantholytic, of oral and laryngeal mucosa, 619226 (3), Autosomal recessive |
| *DSG4* | 607892 | Hypotrichosis 6, 607903 (3), Autosomal recessive |
| *DSP* | 125647 | Arrhythmogenic right ventricular dysplasia 8, 607450 (3), Autosomal dominant; Skin fragility-woolly hair syndrome, 607655 (3), Autosomal recessive; Epidermolysis bullosa, lethal acantholytic, 609638 (3), Autosomal recessive; Keratosis palmoplantaris striata II, 612908 (3), Autosomal dominant; Dilated cardiomyopathy with woolly hair, keratoderma, and tooth agenesis, 615821 (3), Autosomal dominant; Cardiomyopathy, dilated, with woolly hair and keratoderma, 605676 (3), Autosomal recessive |
| *DSPP* | 125485 | Dentinogenesis imperfecta, Shields type III, 125500 (3), Autosomal dominant; Dentinogenesis imperfecta, Shields type II, 125490 (3), Autosomal dominant; Dentin dysplasia, type II, 125420 (3), Autosomal dominant; Deafness, autosomal dominant 39, with dentinogenesis, 605594 (3), Autosomal dominant |
| *DST* | 113810 | Epidermolysis bullosa simplex, autosomal recessive 2, 615425 (3), Autosomal recessive; ?Neuropathy, hereditary sensory and autonomic, type VI, 614653 (3), Autosomal recessive |
| *DSTYK* | 612666 | Congenital anomalies of kidney and urinary tract 1, 610805 (3), Autosomal dominant; Spastic paraplegia 23, 270750 (3), Autosomal recessive |
| *DTNA* | 601239 | Left ventricular noncompaction 1, with or without congenital heart defects, 604169 (3), Autosomal dominant |
| *DTNBP1* | 607145 | Hermansky-Pudlak syndrome 7, 614076 (3), Autosomal recessive |
| *DUOX2* | 606759 | Thyroid dyshormonogenesis 6, 607200 (3), Autosomal recessive |
| *DUOXA2* | 612772 | Thyroid dyshormonogenesis 5, 274900 (3), Autosomal recessive |
| *DUSP6* | 602748 | Hypogonadotropic hypogonadism 19 with or without anosmia, 615269 (3), Autosomal dominant |
| *DVL1* | 601365 | Robinow syndrome, autosomal dominant 2, 616331 (3), Autosomal dominant |
| *DVL3* | 601368 | Robinow syndrome, autosomal dominant 3, 616894 (3), Autosomal dominant |
| *DYM* | 607461 | Smith-McCort dysplasia, 607326 (3), Autosomal recessive; Dyggve-Melchior-Clausen disease, 223800 (3), Autosomal recessive |
| *DYNC1H1* | 600112 | Spinal muscular atrophy, lower extremity-predominant 1, AD, 158600 (3), Autosomal dominant; Charcot-Marie-Tooth disease, axonal, type 20, 614228 (3), Autosomal dominant; Mental retardation, autosomal dominant 13, 614563 (3), Autosomal dominant |
| *DYNC1I2* | 603331 | Neurodevelopmental disorder with microcephaly and structural brain anomalies, 618492 (3), Autosomal recessive |
| *DYNC2H1* | 603297 | Short-rib thoracic dysplasia 3 with or without polydactyly, 613091 (3), Autosomal recessive, Digenic recessive |
| *DYNC2I1 (WDR60)* | 615462 | Short-rib thoracic dysplasia 8 with or without polydactyly, 615503 (3), Autosomal recessive |
| *DYNC2I2 (WDR34)* | 613363 | Short-rib thoracic dysplasia 11 with or without polydactyly, 615633 (3), Autosomal recessive |
| *DYNC2LI1* | 617083 | Short-rib thoracic dysplasia 15 with polydactyly, 617088 (3), Autosomal recessive |
| *DYNLT2B (TCTEX1D2)* | 617353 | Short-rib thoracic dysplasia 17 with or without polydactyly, 617405 (3), Autosomal recessive |
| *DYRK1A* | 600855 | Mental retardation, autosomal dominant 7, 614104 (3), Autosomal dominant |
| *DYRK1B* | 604556 | Abdominal obesity-metabolic syndrome 3, 615812 (3), Autosomal dominant |
| *DYSF* | 603009 | Muscular dystrophy, limb-girdle, autosomal recessive 2, 253601 (3), Autosomal recessive; Miyoshi muscular dystrophy 1, 254130 (3), Autosomal recessive; Myopathy, distal, with anterior tibial onset, 606768 (3), Autosomal recessive |
| *DZIP1* | 608671 | Spermatogenic failure 47, 619102 (3), Autosomal recessive; ?Mitral valve prolapse 3, 610840 (3), Autosomal dominant |
| *DZIP1L* | 617570 | Polycystic kidney disease 5, 617610 (3), Autosomal recessive |
| *EARS2* | 612799 | Combined oxidative phosphorylation deficiency 12, 614924 (3), Autosomal recessive |
| *EBF3* | 607407 | Hypotonia, ataxia, and delayed development syndrome, 617330 (3), Autosomal dominant |
| *EBP* | 300205 | MEND syndrome, 300960 (3), X-linked recessive; Chondrodysplasia punctata, X-linked dominant, 302960 (3), X-linked dominant |
| *ECE1* | 600423 | {Hypertension, essential, susceptibility to}, 145500 (3), Multifactorial; ?Hirschsprung disease, cardiac defects, and autonomic dysfunction, 613870 (3), Autosomal dominant |
| *ECEL1* | 605896 | Arthrogryposis, distal, type 5D, 615065 (3), Autosomal recessive |
| *ECHS1* | 602292 | Mitochondrial short-chain enoyl-CoA hydratase 1 deficiency, 616277 (3), Autosomal recessive |
| *ECM1* | 602201 | Urbach-Wiethe disease, 247100 (3), Autosomal recessive |
| *EDA* | 300451 | Tooth agenesis, selective, X-linked 1, 313500 (3), X-linked dominant; Ectodermal dysplasia 1, hypohidrotic, X-linked, 305100 (3), X-linked recessive |
| *EDAR* | 604095 | [Hair morphology 1, hair thickness], 612630 (3); 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 |
| *EDARADD* | 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 |
| *EDC3* | 609842 | ?Mental retardation, autosomal recessive 50, 616460 (3), Autosomal recessive |
| *EDN1* | 131240 | {High density lipoprotein cholesterol level QTL 7} (3); Question mark ears, isolated, 612798 (3), Autosomal dominant; Auriculocondylar syndrome 3, 615706 (3), Autosomal recessive |
| *EDN3* | 131242 | Waardenburg syndrome, type 4B, 613265 (3), Autosomal recessive, Autosomal dominant; {Hirschsprung disease, susceptibility to, 4}, 613712 (3), Autosomal dominant; Central hypoventilation syndrome, congenital, 209880 (3), Autosomal dominant |
| *EDNRA* | 131243 | {Migraine, resistance to}, 157300 (3), Autosomal dominant; Mandibulofacial dysostosis with alopecia, 616367 (3), Autosomal dominant |
| *EDNRB* | 131244 | {Hirschsprung disease, susceptibility to, 2}, 600155 (3), Autosomal dominant; ABCD syndrome, 600501 (3), Autosomal recessive; Waardenburg syndrome, type 4A, 277580 (3), Autosomal recessive, Autosomal dominant |
| *EED* | 605984 | Cohen-Gibson syndrome, 617561 (3), Autosomal dominant |
| *EEF1A2* | 602959 | Mental retardation, autosomal dominant 38, 616393 (3), Autosomal dominant; Developmental and epileptic encephalopathy 33, 616409 (3), Autosomal dominant |
| *EEF2* | 130610 | ?Spinocerebellar ataxia 26, 609306 (3), Autosomal dominant |
| *EFEMP1* | 601548 | Doyne honeycomb degeneration of retina, 126600 (3), Autosomal dominant |
| *EFEMP2* | 604633 | Cutis laxa, autosomal recessive, type IB, 614437 (3), Autosomal recessive |
| *EFHC1* | 608815 | {Epilepsy, juvenile absence, susceptibility to, 1}, 607631 (3), Autosomal dominant; {Myoclonic epilepsy, juvenile, susceptibility to, 1}, 254770 (3), Autosomal dominant |
| *EFL1* | 617538 | Shwachman-Diamond syndrome 2, 617941 (3), Autosomal recessive |
| *EFNB1* | 300035 | Craniofrontonasal dysplasia, 304110 (3), X-linked dominant |
| *EFTUD2* | 603892 | Mandibulofacial dysostosis, Guion-Almeida type, 610536 (3), Autosomal dominant |
| *EGF* | 131530 | Hypomagnesemia 4, renal, 611718 (3) |
| *EGFR* | 131550 | ?Inflammatory skin and bowel disease, neonatal, 2, 616069 (3), Autosomal recessive; Nonsmall cell lung cancer, response to tyrosine kinase inhibitor in, 211980 (3), Somatic mutation, Autosomal dominant; Adenocarcinoma of lung, response to tyrosine kinase inhibitor in, 211980 (3), Somatic mutation, Autosomal dominant; {Nonsmall cell lung cancer, susceptibility to}, 211980 (3), Somatic mutation, Autosomal dominant |
| *EGLN1* | 606425 | Erythrocytosis, familial, 3, 609820 (3), Autosomal dominant; [Hemoglobin, high altitude adaptation], 609070 (3), Autosomal dominant |
| *EGR2* | 129010 | Dejerine-Sottas disease, 145900 (3), Autosomal recessive, Autosomal dominant; Charcot-Marie-Tooth disease, type 1D, 607678 (3), Autosomal dominant; Hypomyelinating neuropathy, congenital, 1, 605253 (3), Autosomal recessive, Autosomal dominant |
| *EHBP1* | 609922 | {Prostate cancer, hereditary, 12}, 611868 (3) |
| *EHHADH* | 607037 | ?Fanconi renotubular syndrome 3, 615605 (3), Autosomal dominant |
| *EHMT1* | 607001 | Kleefstra syndrome 1, 610253 (3), Autosomal dominant |
| *EIF2AK1* | 613635 | ?Leukoencephalopathy, motor delay, spasticity, and dysarthria syndrome, 618878 (3) |
| *EIF2AK2* | 176871 | Leukoencephalopathy, developmental delay, and episodic neurologic regression syndrome, 618877 (3), Autosomal dominant |
| *EIF2AK3* | 604032 | Wolcott-Rallison syndrome, 226980 (3), Autosomal recessive |
| *EIF2AK4* | 609280 | Pulmonary venoocclusive disease 2, 234810 (3), Autosomal recessive |
| *EIF2B1* | 606686 | Leukoencephalopathy with vanishing white matter, 603896 (3), Autosomal recessive |
| *EIF2B2* | 606454 | Leukoencephalopathy with vanishing white matter, 603896 (3), Autosomal recessive; Ovarioleukodystrophy, 603896 (3), Autosomal recessive |
| *EIF2B3* | 606273 | Leukoencephalopathy with vanishing white matter, 603896 (3), Autosomal recessive |
| *EIF2B4* | 606687 | Ovarioleukodystrophy, 603896 (3), Autosomal recessive; Leukoencephalopathy with vanishing white matter, 603896 (3), Autosomal recessive |
| *EIF2B5* | 603945 | Ovarioleukodystrophy, 603896 (3), Autosomal recessive; Leukoencephalopathy with vanishing white matter, 603896 (3), Autosomal recessive |
| *EIF2S3* | 300161 | MEHMO syndrome, 300148 (3), X-linked recessive |
| *EIF3F* | 603914 | Mental retardation, autosomal recessive 67, 618295 (3), Autosomal recessive |
| *EIF4A3* | 608546 | Robin sequence with cleft mandible and limb anomalies, 268305 (3), Autosomal recessive |
| *EIF4E* | 133440 | {Autism, susceptibility to, 19}, 615091 (3) |
| *EIF4G1* | 600495 | {Parkinson disease 18}, 614251 (3), Autosomal dominant |
| *EIF5A* | 600187 | Faundes-Banka syndrome, 619376 (3), Autosomal dominant |
| *ELAC2* | 605367 | {Prostate cancer, hereditary, 2, susceptibility to}, 614731 (3); Combined oxidative phosphorylation deficiency 17, 615440 (3), Autosomal recessive |
| *ELANE* | 130130 | Neutropenia, cyclic, 162800 (3), Autosomal dominant; Neutropenia, severe congenital 1, autosomal dominant, 202700 (3), Autosomal dominant |
| *ELMO2* | 606421 | Vascular malformation, primary intraosseous, 606893 (3), Autosomal recessive |
| *ELMOD3* | 615427 | ?Deafness, autosomal recessive 88, 615429 (3), Autosomal recessive |
| *ELN* | 130160 | Cutis laxa, autosomal dominant, 123700 (3), Autosomal dominant; Supravalvar aortic stenosis, 185500 (3), Autosomal dominant |
| *ELOVL1* | 611813 | Ichthyotic keratoderma, spasticity, hypomyelination, and dysmorphic facies, 618527 (3), Autosomal dominant |
| *ELOVL4* | 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 |
| *ELOVL5* | 611805 | Spinocerebellar ataxia 38, 615957 (3), Autosomal dominant |
| *ELP1* | 603722 | Dysautonomia, familial, 223900 (3), Autosomal recessive |
| *ELP2* | 616054 | Mental retardation, autosomal recessive 58, 617270 (3), Autosomal recessive |
| *ELP4* | 606985 | ?Aniridia 2, 617141 (3), Autosomal dominant |
| *EMC1* | 616846 | Cerebellar atrophy, visual impairment, and psychomotor retardation, 616875 (3), Autosomal recessive |
| *EMC10* | 614545 | Neurodevelopmental disorder with dysmorphic facies and variable seizures, 619264 (3), Autosomal recessive |
| *EMD* | 300384 | Emery-Dreifuss muscular dystrophy 1, X-linked, 310300 (3), X-linked recessive |
| *EMG1* | 611531 | Bowen-Conradi syndrome, 211180 (3), Autosomal recessive |
| *EML1* | 602033 | Band heterotopia, 600348 (3), Autosomal recessive |
| *EMP2* | 602334 | Nephrotic syndrome, type 10, 615861 (3), Autosomal recessive |
| *EMX2* | 600035 | Schizencephaly, 269160 (3) |
| *EN1* | 131290 | ?ENDOVE syndrome, limb-brain type, 619218 (3), Autosomal recessive |
| *ENAM* | 606585 | Amelogenesis imperfecta, type IC, 204650 (3), Autosomal recessive; Amelogenesis imperfecta, type IB, 104500 (3), Autosomal dominant |
| *ENG* | 131195 | Telangiectasia, hereditary hemorrhagic, type 1, 187300 (3), Autosomal dominant |
| *ENO3* | 131370 | ?Glycogen storage disease XIII, 612932 (3), Autosomal recessive |
| *ENPP1* | 173335 | {Obesity, susceptibility to}, 601665 (3), Multifactorial, Autosomal recessive, Autosomal dominant; Hypophosphatemic rickets, autosomal recessive, 2, 613312 (3), Autosomal recessive; {Diabetes mellitus, non-insulin-dependent, susceptibility to}, 125853 (3), Autosomal dominant; Arterial calcification, generalized, of infancy, 1, 208000 (3), Autosomal recessive; Cole disease, 615522 (3), Autosomal dominant |
| *ENTPD1* | 601752 | Spastic paraplegia 64, autosomal recessive, 615683 (3), Autosomal recessive |
| *EOGT* | 614789 | Adams-Oliver syndrome 4, 615297 (3), Autosomal recessive |
| *EP300* | 602700 | Menke-Hennekam syndrome 2, 618333 (3), Autosomal dominant; Colorectal cancer, somatic, 114500 (3); Rubinstein-Taybi syndrome 2, 613684 (3), Autosomal dominant |
| *EPAS1* | 603349 | Erythrocytosis, familial, 4, 611783 (3), Autosomal dominant |
| *EPB41* | 130500 | Elliptocytosis-1, 611804 (3), Autosomal recessive, Autosomal dominant |
| *EPB41L1* | 602879 | ?Intellectual developmental disorder, autosomal dominant 11, 614257 (3), Autosomal dominant |
| *EPB42* | 177070 | Spherocytosis, type 5, 612690 (3) |
| *EPCAM* | 185535 | Colorectal cancer, hereditary nonpolyposis, type 8, 613244 (3); Diarrhea 5, with tufting enteropathy, congenital, 613217 (3), Autosomal recessive |
| *EPG5* | 615068 | Vici syndrome, 242840 (3), Autosomal recessive |
| *EPHA2* | 176946 | Cataract 6, multiple types, 116600 (3), Autosomal dominant |
| *EPHB2* | 600997 | ?Bleeding disorder, platelet-type, 22, 618462 (3), Autosomal recessive; {Prostate cancer/brain cancer susceptibility, somatic}, 603688 (3) |
| *EPHB4* | 600011 | Capillary malformation-arteriovenous malformation 2, 618196 (3), Autosomal dominant; Lymphatic malformation 7, 617300 (3), Autosomal dominant |
| *EPHX2* | 132811 | {Hypercholesterolemia, familial, due to LDLR defect, modifier of}, 143890 (3), Autosomal recessive, Autosomal dominant |
| *EPM2A* | 607566 | Epilepsy, progressive myoclonic 2A (Lafora), 254780 (3), Autosomal recessive |
| *EPO* | 133170 | {Microvascular complications of diabetes 2}, 612623 (3); Erythrocytosis, familial, 5, 617907 (3), Autosomal dominant; ?Diamond-Blackfan anemia-like, 617911 (3), Autosomal recessive |
| *EPOR* | 133171 | [Erythrocytosis, familial, 1], 133100 (3), Autosomal dominant |
| *EPRS1 (EPRS)* | 138295 | Leukodystrophy, hypomyelinating, 15, 617951 (3), Autosomal recessive |
| *EPS8* | 600206 | ?Deafness, autosomal recessive 102, 615974 (3), Autosomal recessive |
| *EPS8L2* | 614988 | Deafness autosomal recessive 106, 617637 (3), Autosomal recessive |
| *EPS8L3* | 614989 | ?Hypotrichosis 5, 612841 (3), Autosomal dominant |
| *EPX* | 131399 | [Eosinophil peroxidase deficiency], 261500 (3), Autosomal recessive |
| *ERAL1* | 607435 | Perrault syndrome 6, 617565 (3), Autosomal recessive |
| *ERBB2* | 164870 | Gastric cancer, somatic, 613659 (3); Adenocarcinoma of lung, somatic, 211980 (3); Ovarian cancer, somatic (3); Glioblastoma, somatic, 137800 (3) |
| *ERBB3* | 190151 | ?Lethal congenital contractural syndrome 2, 607598 (3), Autosomal recessive; {?Erythroleukemia, familial, susceptibility to}, 133180 (3), Autosomal dominant |
| *ERBB4* | 600543 | Amyotrophic lateral sclerosis 19, 615515 (3), Autosomal dominant |
| *ERCC1* | 126380 | Cerebrooculofacioskeletal syndrome 4, 610758 (3), Autosomal recessive |
| *ERCC2* | 126340 | Xeroderma pigmentosum, group D, 278730 (3), Autosomal recessive; Trichothiodystrophy 1, photosensitive, 601675 (3), Autosomal recessive; ?Cerebrooculofacioskeletal syndrome 2, 610756 (3), Autosomal recessive |
| *ERCC3* | 133510 | Trichothiodystrophy 2, photosensitive, 616390 (3), Autosomal recessive; Xeroderma pigmentosum, group B, 610651 (3), Autosomal recessive |
| *ERCC4* | 133520 | Xeroderma pigmentosum, type F/Cockayne syndrome, 278760 (3), Autosomal recessive; XFE progeroid syndrome, 610965 (3), Autosomal recessive; Xeroderma pigmentosum, group F, 278760 (3), Autosomal recessive; Fanconi anemia, complementation group Q, 615272 (3), Autosomal recessive |
| *ERCC5* | 133530 | Xeroderma pigmentosum, group G, 278780 (3), Autosomal recessive; Cerebrooculofacioskeletal syndrome 3, 616570 (3), Autosomal recessive; Xeroderma pigmentosum, group G/Cockayne syndrome, 278780 (3), Autosomal recessive |
| *ERCC6* | 609413 | UV-sensitive syndrome 1, 600630 (3), Autosomal recessive; Cerebrooculofacioskeletal syndrome 1, 214150 (3), Autosomal recessive; Cockayne syndrome, type B, 133540 (3), Autosomal recessive; De Sanctis-Cacchione syndrome, 278800 (3), Autosomal recessive; {Macular degeneration, age-related, susceptibility to, 5}, 613761 (3); Premature ovarian failure 11, 616946 (3), Autosomal dominant; {Lung cancer, susceptibility to}, 211980 (3), Somatic mutation, Autosomal dominant |
| *ERCC6L2* | 615667 | Bone marrow failure syndrome 2, 615715 (3), Autosomal recessive |
| *ERCC8* | 609412 | UV-sensitive syndrome 2, 614621 (3), Autosomal recessive; Cockayne syndrome, type A, 216400 (3), Autosomal recessive |
| *ERF* | 611888 | Craniosynostosis 4, 600775 (3), Autosomal dominant; Chitayat syndrome, 617180 (3), Autosomal dominant |
| *ERGIC1* | 617946 | ?Arthrogryposis multiplex congenita 2, neurogenic type, 208100 (3), Autosomal recessive |
| *ERLIN1* | 611604 | Spastic paraplegia 62, 615681 (3), Autosomal recessive |
| *ERLIN2* | 611605 | Spastic paraplegia 18, autosomal recessive, 611225 (3), Autosomal recessive |
| *ERMAP* | 609017 | [Blood group, Scianna system], 111750 (3); [Blood group, Radin], 111620 (3) |
| *ERMARD* | 615532 | ?Periventricular nodular heterotopia 6, 615544 (3), Autosomal dominant |
| *ESCO2* | 609353 | Juberg-Hayward syndrome, 216100 (3), Autosomal recessive; Roberts-SC phocomelia syndrome, 268300 (3), Autosomal recessive |
| *ESPN* | 606351 | Deafness, neurosensory, without vestibular involvement, autosomal dominant, 609006 (3), Autosomal recessive; Deafness, autosomal recessive 36, 609006 (3), Autosomal recessive; ?Usher syndrome, type 1M, 618632 (3), Autosomal recessive |
| *ESR1* | 133430 | Breast cancer, somatic, 114480 (3); {Migraine, susceptibility to}, 157300 (3), Autosomal dominant; Estrogen resistance, 615363 (3), Autosomal recessive; {Myocardial infarction, susceptibility to}, 608446 (3) |
| *ESR2* | 601663 | ?Ovarian dysgenesis 8, 618187 (3), Autosomal dominant |
| *ESRP1* | 612959 | ?Deafness, autosomal recessive 109, 618013 (3), Autosomal recessive |
| *ESRRB* | 602167 | Deafness, autosomal recessive 35, 608565 (3), Autosomal recessive |
| *ETFA* | 608053 | Glutaric acidemia IIA, 231680 (3), Autosomal recessive |
| *ETFB* | 130410 | Glutaric acidemia IIB, 231680 (3), Autosomal recessive |
| *ETFDH* | 231675 | Glutaric acidemia IIC, 231680 (3), Autosomal recessive |
| *ETHE1* | 608451 | Ethylmalonic encephalopathy, 602473 (3), Autosomal recessive |
| *ETV6* | 600618 | Thrombocytopenia 5, 616216 (3), Autosomal dominant; Leukemia, acute myeloid, somatic, 601626 (3) |
| *EVC* | 604831 | Ellis-van Creveld syndrome, 225500 (3), Autosomal recessive; ?Weyers acrofacial dysostosis, 193530 (3), Autosomal dominant |
| *EVC2* | 607261 | Ellis-van Creveld syndrome, 225500 (3), Autosomal recessive; Weyers acrofacial dysostosis, 193530 (3), Autosomal dominant |
| *EWSR1* | 133450 | Neuroepithelioma, 612219 (3); Ewing sarcoma, 612219 (3) |
| *EXOC2* | 615329 | Neurodevelopmental disorder with dysmorphic facies and cerebellar hypoplasia, 619306 (3), Autosomal recessive |
| *EXOC3L2* | 616927 | Dandy-Walker malformation; Meckel-Gruber-like syndrome. PMID: 30327448 |
| *EXOC6B* | 607880 | Spondyloepimetaphyseal dysplasia with joint laxity, type 3, 618395 (3), Autosomal recessive |
| *EXOC7* | 608163 | Neurodevelopmental disorder with seizures and brain atrophy, 619072 (3) |
| *EXOC8* | 615283 | ?Neurodevelopmental disorder with microcephaly, seizures, and brain atrophy, 619076 (3), Autosomal recessive |
| *EXOSC1* | 606493 | ?Pontocerebellar hypoplasia, type 1F, 619304 (3), Autosomal recessive |
| *EXOSC2* | 602238 | Short stature, hearing loss, retinitis pigmentosa, and distinctive facies, 617763 (3), Autosomal recessive |
| *EXOSC3* | 606489 | Pontocerebellar hypoplasia, type 1B, 614678 (3), Autosomal recessive |
| *EXOSC8* | 606019 | Pontocerebellar hypoplasia, type 1C, 616081 (3), Autosomal recessive |
| *EXOSC9* | 606180 | Pontocerebellar hypoplasia, type 1D, 618065 (3), Autosomal recessive |
| *EXPH5* | 612878 | Epidermolysis bullosa, nonspecific, autosomal recessive, 615028 (3), Autosomal recessive |
| *EXT1* | 608177 | Exostoses, multiple, type 1, 133700 (3), Autosomal dominant; Chondrosarcoma, 215300 (3), Somatic mutation |
| *EXT2* | 608210 | Seizures, scoliosis, and macrocephaly syndrome, 616682 (3), Autosomal recessive; Exostoses, multiple, type 2, 133701 (3), Autosomal dominant |
| *EXTL3* | 605744 | Immunoskeletal dysplasia with neurodevelopmental abnormalities, 617425 (3), Autosomal recessive |
| *EYA1* | 601653 | Branchiootic syndrome 1, 602588 (3), Autosomal dominant; Branchiootorenal syndrome 1, with or without cataracts, 113650 (3), Autosomal dominant; Anterior segment anomalies with or without cataract, 602588 (3), Autosomal dominant; ?Otofaciocervical syndrome, 166780 (3), Autosomal dominant |
| *EYA4* | 603550 | ?Cardiomyopathy, dilated, 1J, 605362 (3), Autosomal dominant; Deafness, autosomal dominant 10, 601316 (3), Autosomal dominant |
| *EYS* | 612424 | Retinitis pigmentosa 25, 602772 (3), Autosomal recessive |
| *EZH2* | 601573 | Weaver syndrome, 277590 (3), Autosomal dominant |
| *F10* | 613872 | Factor X deficiency, 227600 (3), Autosomal recessive |
| *F11* | 264900 | Factor XI deficiency, autosomal dominant, 612416 (3); Factor XI deficiency, autosomal recessive, 612416 (3) |
| *F12* | 610619 | Angioedema, hereditary, 3, 610618 (3), Autosomal dominant; Factor XII deficiency, 234000 (3), Autosomal recessive |
| *F13A1* | 134570 | Factor XIIIA deficiency, 613225 (3), Autosomal recessive; {Myocardial infarction, protection against}, 608446 (3); {Venous thrombosis, protection against}, 188050 (3), Autosomal dominant |
| *F13B* | 134580 | Factor XIIIB deficiency, 613235 (3), Autosomal recessive |
| *F2* | 176930 | Hypoprothrombinemia, 613679 (3), Autosomal recessive; {Pregnancy loss, recurrent, susceptibility to, 2}, 614390 (3), Autosomal dominant; Dysprothrombinemia, 613679 (3), Autosomal recessive; {Stroke, ischemic, susceptibility to}, 601367 (3), Multifactorial; Thrombophilia due to thrombin defect, 188050 (3), Autosomal dominant |
| *F5* | 612309 | {Pregnancy loss, recurrent, susceptibility to, 1}, 614389 (3), Autosomal dominant; {Thrombophilia, susceptibility to, due to factor V Leiden}, 188055 (3), Autosomal dominant; {Budd-Chiari syndrome}, 600880 (3), Autosomal recessive; Thrombophilia due to activated protein C resistance, 188055 (3), Autosomal dominant; {Stroke, ischemic, susceptibility to}, 601367 (3), Multifactorial; Factor V deficiency, 227400 (3), Autosomal recessive |
| *F7* | 613878 | {Myocardial infarction, decreased susceptibility to}, 608446 (3); Factor VII deficiency, 227500 (3), Autosomal recessive |
| *F8* | 300841 | Hemophilia A, 306700 (3), X-linked recessive |
| *F9* | 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 |
| *FA2H* | 611026 | Spastic paraplegia 35, autosomal recessive, 612319 (3), Autosomal recessive |
| *FAAH* | 602935 | {Drug addiction, susceptibility to}, 606581 (3) |
| *FADD* | 602457 | Infections, recurrent, with encephalopathy, hepatic dysfunction, and cardiovascular malformations, 613759 (3), Autosomal recessive |
| *FAH* | 613871 | Tyrosinemia, type I, 276700 (3), Autosomal recessive |
| *FAM111A* | 615292 | Kenny-Caffey syndrome, type 2, 127000 (3), Autosomal dominant; Gracile bone dysplasia, 602361 (3), Autosomal dominant |
| *FAM111B* | 615584 | Poikiloderma, hereditary fibrosing, with tendon contractures, myopathy, and pulmonary fibrosis, 615704 (3), Autosomal dominant |
| *FAM126A* | 610531 | Leukodystrophy, hypomyelinating, 5, 610532 (3), Autosomal recessive |
| *FAM149B1* | 618413 | Joubert syndrome 36, 618763 (3), Autosomal recessive |
| *FAM161A* | 613596 | Retinitis pigmentosa 28, 606068 (3) |
| *FAM20A* | 611062 | Amelogenesis imperfecta, type IG (enamel-renal syndrome), 204690 (3), Autosomal recessive |
| *FAM20C* | 611061 | Raine syndrome, 259775 (3), Autosomal recessive |
| *FAM50A* | 300453 | Intellectual developmental disorder, X-linked, syndromic, Armfield type, 300261 (3), X-linked recessive |
| *FAM83H* | 611927 | Amelogenesis imperfecta, type IIIA, 130900 (3), Autosomal dominant |
| *FAN1* | 613534 | Interstitial nephritis, karyomegalic, 614817 (3), Autosomal recessive |
| *FANCA* | 607139 | Fanconi anemia, complementation group A, 227650 (3), Autosomal recessive |
| *FANCB* | 300515 | Fanconi anemia, complementation group B, 300514 (3), X-linked recessive |
| *FANCC* | 613899 | Fanconi anemia, complementation group C, 227645 (3), Autosomal recessive |
| *FANCD2* | 613984 | Fanconi anemia, complementation group D2, 227646 (3), Autosomal recessive |
| *FANCE* | 613976 | Fanconi anemia, complementation group E, 600901 (3), Autosomal recessive |
| *FANCF* | 613897 | Fanconi anemia, complementation group F, 603467 (3), Autosomal recessive |
| *FANCG* | 602956 | Fanconi anemia, complementation group G, 614082 (3), Autosomal recessive |
| *FANCI* | 611360 | Fanconi anemia, complementation group I, 609053 (3), Autosomal recessive |
| *FANCL* | 608111 | Fanconi anemia, complementation group L, 614083 (3), Autosomal recessive |
| *FANCM* | 609644 | ?Premature ovarian failure 15, 618096 (3), Autosomal recessive; Spermatogenic failure 28, 618086 (3), Autosomal recessive |
| *FAR1* | 616107 | Peroxisomal fatty acyl-CoA reductase 1 disorder, 616154 (3), Autosomal recessive; Cataracts, spastic paraparesis, and speech delay, 619338 (3), Autosomal dominant |
| *FARS2* | 611592 | Combined oxidative phosphorylation deficiency 14, 614946 (3), Autosomal recessive; Spastic paraplegia 77, autosomal recessive, 617046 (3), Autosomal recessive |
| *FARSA* | 602918 | ?Rajab interstitial lung disease with brain calcifications 2, 619013 (3), Autosomal recessive |
| *FARSB* | 609690 | Rajab interstitial lung disease with brain calcifications 1, 613658 (3), Autosomal recessive |
| *FAS* | 134637 | Squamous cell carcinoma, burn scar-related, somatic (3); Autoimmune lymphoproliferative syndrome, type IA, 601859 (3), Autosomal dominant; {Autoimmune lymphoproliferative syndrome}, 601859 (3), Autosomal dominant |
| *FASLG* | 134638 | Autoimmune lymphoproliferative syndrome, type IB, 601859 (3), Autosomal dominant; {Lung cancer, susceptibility to}, 211980 (3), Somatic mutation, Autosomal dominant |
| *FASTKD2* | 612322 | Combined oxidative phosphorylation deficiency 44, 618855 (3), Autosomal recessive |
| *FAT2* | 604269 | Spinocerebellar ataxia 45, 617769 (3), Autosomal dominant |
| *FAT4* | 612411 | Van Maldergem syndrome 2, 615546 (3), Autosomal recessive; Hennekam lymphangiectasia-lymphedema syndrome 2, 616006 (3), Autosomal recessive |
| *FBLN1* | 135820 | Synpolydactyly, 3/3'4, associated with metacarpal and metatarsal synostoses, 608180 (4), Autosomal dominant |
| *FBLN5* | 604580 | Cutis laxa, autosomal recessive, type IA, 219100 (3), Autosomal recessive; Macular degeneration, age-related, 3, 608895 (3), Autosomal dominant; Neuropathy, hereditary, with or without age-related macular degeneration, 608895 (3), Autosomal dominant; ?Cutis laxa, autosomal dominant 2, 614434 (3), Autosomal dominant |
| *FBN1* | 134797 | Geleophysic dysplasia 2, 614185 (3), Autosomal dominant; Weill-Marchesani syndrome 2, dominant, 608328 (3), Autosomal dominant; Ectopia lentis, familial, 129600 (3), Autosomal dominant; MASS syndrome, 604308 (3), Autosomal dominant; Marfan lipodystrophy syndrome, 616914 (3), Autosomal dominant; Acromicric dysplasia, 102370 (3), Autosomal dominant; Marfan syndrome, 154700 (3), Autosomal dominant; Stiff skin syndrome, 184900 (3), Autosomal dominant |
| *FBN2* | 612570 | Macular degeneration, early-onset, 616118 (3), Autosomal dominant; Contractural arachnodactyly, congenital, 121050 (3), Autosomal dominant |
| *FBP1* | 611570 | Fructose-1,6-bisphosphatase deficiency, 229700 (3), Autosomal recessive |
| *FBXL3* | 605653 | Intellectual developmental disorder with short stature, facial anomalies, and speech defects, 606220 (3), Autosomal recessive |
| *FBXL4* | 605654 | Mitochondrial DNA depletion syndrome 13 (encephalomyopathic type), 615471 (3), Autosomal recessive |
| *FBXO11* | 607871 | Intellectual developmental disorder with dysmorphic facies and behavioral abnormalities, 618089 (3), Autosomal dominant |
| *FBXO31* | 609102 | ?Mental retardation, autosomal recessive 45, 615979 (3), Autosomal recessive |
| *FBXO38* | 608533 | Neuronopathy, distal hereditary motor, type IID, 615575 (3), Autosomal dominant |
| *FBXO7* | 605648 | Parkinson disease 15, autosomal recessive, 260300 (3), Autosomal recessive |
| *FBXW11* | 605651 | Neurodevelopmental, jaw, eye, and digital syndrome, 618914 (3), Autosomal dominant |
| *FBXW4* | 608071/246560 | -/Split-hand/foot malformation 3, gene duplication syndrome, 246560 (4), Autosomal dominant |
| *FCGR1A* | 146760 | [IgG receptor I, phagocytic, familial deficiency of] (3) |
| *FCGR2A* | 146790 | {Malaria, severe, susceptibility to}, 611162 (3); {Pseudomonas aeruginosa, susceptibility to chronic infection by, in cystic fibrosis}, 219700 (3), Autosomal recessive; {Lupus nephritis, susceptibility to}, 152700 (3), Autosomal dominant |
| *FCGR2B* | 604590 | {Systemic lupus erythematosus, susceptibility to}, 152700 (3), Autosomal dominant; {Malaria, resistance to}, 611162 (3) |
| *FCGR3A* | 146740 | Immunodeficiency 20, 615707 (3), Autosomal recessive |
| *FCHO1* | 613437 | Immunodeficiency 76, 619164 (3), Autosomal recessive |
| *FCN3* | 604973 | Immunodeficiency due to ficolin 3 deficiency, 613860 (3), Autosomal recessive |
| *FCSK (FUK)* | 608675 | Congenital disorder of glycosylation with defective fucosylation 2, 618324 (3), Autosomal recessive |
| *FDFT1* | 184420 | Squalene synthase deficiency, 618156 (3), Autosomal recessive |
| *FDPS* | 134629 | Porokeratosis 9, multiple types, 616631 (3), Autosomal dominant |
| *FDX2* | 614585 | Mitochondrial myopathy, episodic, with optic atrophy and reversible leukoencephalopathy, 251900 (3), Autosomal recessive |
| *FDXR* | 103270 | Auditory neuropathy and optic atrophy, 617717 (3), Autosomal recessive |
| *FECH* | 612386 | Protoporphyria, erythropoietic, 1, 177000 (3), Autosomal recessive |
| *FERMT1* | 607900 | Kindler syndrome, 173650 (3), Autosomal recessive |
| *FERMT3* | 607901 | Leukocyte adhesion deficiency, type III, 612840 (3), Autosomal recessive |
| *FEZF1* | 613301 | Hypogonadotropic hypogonadism 22, with or without anosmia, 616030 (3), Autosomal recessive |
| *FFAR4* | 609044 | {Obesity, susceptibility to}, 607514 (3) |
| *FGA* | 134820 | Hypodysfibrinogenemia, congenital, 616004 (3); Dysfibrinogenemia, congenital, 616004 (3); Amyloidosis, familial visceral, 105200 (3), Autosomal dominant; Afibrinogenemia, congenital, 202400 (3), Autosomal recessive |
| *FGB* | 134830 | Hypofibrinogenemia, congenital, 202400 (3), Autosomal recessive; Dysfibrinogenemia, congenital, 616004 (3); Afibrinogenemia, congenital, 202400 (3), Autosomal recessive |
| *FGD1* | 300546 | Mental retardation, X-linked syndromic 16, 305400 (3), X-linked recessive; Aarskog-Scott syndrome, 305400 (3), X-linked recessive |
| *FGD4* | 611104 | Charcot-Marie-Tooth disease, type 4H, 609311 (3), Autosomal recessive |
| *FGF10* | 602115 | Aplasia of lacrimal and salivary glands, 180920 (3), Autosomal dominant; LADD syndrome, 149730 (3), Autosomal dominant |
| *FGF12* | 601513 | Developmental and epileptic encephalopathy 47, 617166 (3), Autosomal dominant |
| *FGF13* | 300070 | Developmental and epileptic encephalopathy 90, 301058 (3), X-linked dominant, X-linked recessive |
| *FGF14* | 601515 | Spinocerebellar ataxia 27, 609307 (3), Autosomal dominant |
| *FGF16* | 300827 | Metacarpal 4-5 fusion, 309630 (3), X-linked recessive |
| *FGF17* | 603725 | Hypogonadotropic hypogonadism 20 with or without anosmia, 615270 (3), Autosomal dominant |
| *FGF20* | 605558 | ?Renal hypodysplasia/aplasia 2, 615721 (3), Autosomal recessive |
| *FGF23* | 605380 | Tumoral calcinosis, hyperphosphatemic, familial, 2, 617993 (3), Autosomal recessive; Hypophosphatemic rickets, autosomal dominant, 193100 (3), Autosomal dominant |
| *FGF3* | 164950 | Deafness, congenital with inner ear agenesis, microtia, and microdontia, 610706 (3), Autosomal recessive |
| *FGF5* | 165190 | Trichomegaly, 190330 (3), Autosomal recessive |
| *FGF8* | 600483 | Hypogonadotropic hypogonadism 6 with or without anosmia, 612702 (3), Autosomal dominant |
| *FGF9* | 600921 | Multiple synostoses syndrome 3, 612961 (3), Autosomal dominant |
| *FGFR1* | 136350 | Pfeiffer syndrome, 101600 (3), Autosomal dominant; Hypogonadotropic hypogonadism 2 with or without anosmia, 147950 (3), Autosomal dominant; Jackson-Weiss syndrome, 123150 (3), Autosomal dominant; Hartsfield syndrome, 615465 (3), Autosomal dominant; Trigonocephaly 1, 190440 (3), Autosomal dominant; Osteoglophonic dysplasia, 166250 (3), Autosomal dominant; Encephalocraniocutaneous lipomatosis, somatic mosaic, 613001 (3) |
| *FGFR2* | 176943 | Bent bone dysplasia syndrome, 614592 (3), Autosomal dominant; LADD syndrome, 149730 (3), Autosomal dominant; Scaphocephaly, maxillary retrusion, and mental retardation, 609579 (3); Antley-Bixler syndrome without genital anomalies or disordered steroidogenesis, 207410 (3), Autosomal dominant; Scaphocephaly and Axenfeld-Rieger anomaly (3); Jackson-Weiss syndrome, 123150 (3), Autosomal dominant; Gastric cancer, somatic, 613659 (3); Craniofacial-skeletal-dermatologic dysplasia, 101600 (3), Autosomal dominant; Apert syndrome, 101200 (3), Autosomal dominant; Pfeiffer syndrome, 101600 (3), Autosomal dominant; Craniosynostosis, nonspecific (3); Beare-Stevenson cutis gyrata syndrome, 123790 (3), Autosomal dominant; Crouzon syndrome, 123500 (3), Autosomal dominant; Saethre-Chotzen syndrome, 101400 (3), Autosomal dominant |
| *FGFR3* | 134934 | Muenke syndrome, 602849 (3), Autosomal dominant; SADDAN, 616482 (3), Autosomal dominant; Hypochondroplasia, 146000 (3), Autosomal dominant; LADD syndrome, 149730 (3), Autosomal dominant; Thanatophoric dysplasia, type II, 187601 (3), Autosomal dominant; Nevus, epidermal, somatic, 162900 (3); CATSHL syndrome, 610474 (3), Autosomal recessive, Autosomal dominant; Thanatophoric dysplasia, type I, 187600 (3), Autosomal dominant; Spermatocytic seminoma, somatic, 273300 (3); Bladder cancer, somatic, 109800 (3); Achondroplasia, 100800 (3), Autosomal dominant; Cervical cancer, somatic, 603956 (3); Colorectal cancer, somatic, 114500 (3); Crouzon syndrome with acanthosis nigricans, 612247 (3), Autosomal dominant |
| *FGFR4* | 134935 | {Cancer progression/metastasis} (3) |
| *FGG* | 134850 | Dysfibrinogenemia, congenital, 616004 (3); Hypodysfibrinogenemia, 616004 (3); Hypofibrinogenemia, congenital, 202400 (3), Autosomal recessive; Afibrinogenemia, congenital, 202400 (3), Autosomal recessive |
| *FH* | 136850 | Leiomyomatosis and renal cell cancer, 150800 (3), Autosomal dominant; Fumarase deficiency, 606812 (3), Autosomal recessive |
| *FHL1* | 300163 | Myopathy, X-linked, with postural muscle atrophy, 300696 (3), X-linked recessive; Emery-Dreifuss muscular dystrophy 6, X-linked, 300696 (3), X-linked recessive; ?Uruguay faciocardiomusculoskeletal syndrome, 300280 (3), X-linked recessive; 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; Reducing body myopathy, X-linked 1a, severe, infantile or early childhood onset, 300717 (3), X-linked dominant |
| *FHOD3* | 609691 | Cardiomyopathy, familial hypertrophic, 28, 619402 (3), Autosomal dominant |
| *FIBP* | 608296 | Thauvin-Robinet-Faivre syndrome, 617107 (3), Autosomal recessive |
| *FIG4* | 609390 | Yunis-Varon syndrome, 216340 (3), Autosomal recessive; ?Polymicrogyria, bilateral temporooccipital, 612691 (3), Autosomal recessive; Amyotrophic lateral sclerosis 11, 612577 (3), Autosomal dominant; Charcot-Marie-Tooth disease, type 4J, 611228 (3), Autosomal recessive |
| *FIGLA* | 608697 | Premature ovarian failure 6, 612310 (3), Autosomal dominant |
| *FITM2* | 612029 | Siddiqi syndrome, 618635 (3), Autosomal recessive |
| *FKBP10* | 607063 | Osteogenesis imperfecta, type XI, 610968 (3), Autosomal recessive; Bruck syndrome 1, 259450 (3), Autosomal recessive |
| *FKBP14* | 614505 | Ehlers-Danlos syndrome, kyphoscoliotic type, 2, 614557 (3), Autosomal recessive |
| *FKBP5* | 602623 | {Major depressive disorder and accelerated response to antidepressant drug treatment}, 608516 (3) |
| *FKRP* | 606596 | Muscular dystrophy-dystroglycanopathy (congenital with or without mental retardation), type B, 5, 606612 (3), Autosomal recessive; Muscular dystrophy-dystroglycanopathy (limb-girdle), type C, 5, 607155 (3), Autosomal recessive; Muscular dystrophy-dystroglycanopathy (congenital with brain and eye anomalies), type A, 5, 613153 (3), Autosomal recessive |
| *FKTN* | 607440 | Muscular dystrophy-dystroglycanopathy (limb-girdle), type C, 4, 611588 (3), Autosomal recessive; Muscular dystrophy-dystroglycanopathy (congenital with brain and eye anomalies), type A, 4, 253800 (3), Autosomal recessive; Muscular dystrophy-dystroglycanopathy (congenital without mental retardation), type B, 4, 613152 (3), Autosomal recessive; Cardiomyopathy, dilated, 1X, 611615 (3), Autosomal recessive |
| *FLAD1* | 610595 | Lipid storage myopathy due to flavin adenine dinucleotide synthetase deficiency, 255100 (3), Autosomal recessive |
| *FLCN* | 607273 | Birt-Hogg-Dube syndrome, 135150 (3), Autosomal dominant; Colorectal cancer, somatic, 114500 (3); Pneumothorax, primary spontaneous, 173600 (3), Autosomal dominant; Renal carcinoma, chromophobe, somatic, 144700 (3) |
| *FLG* | 135940 | Ichthyosis vulgaris, 146700 (3), Autosomal recessive, Autosomal dominant; {Dermatitis, atopic, susceptibility to, 2}, 605803 (3) |
| *FLG2* | 616284 | Peeling skin syndrome 6, 618084 (3), Autosomal recessive |
| *FLI1* | 193067 | Bleeding disorder, platelet-type, 21, 617443 (3), Autosomal recessive, Autosomal dominant |
| *FLNA* | 300017 | Otopalatodigital syndrome, type II, 304120 (3), X-linked dominant; Intestinal pseudoobstruction, neuronal, 300048 (3), X-linked recessive; Cardiac valvular dysplasia, X-linked, 314400 (3), X-linked; ?FG syndrome 2, 300321 (3), X-linked; Melnick-Needles syndrome, 309350 (3), X-linked dominant; Terminal osseous dysplasia, 300244 (3), X-linked dominant; Congenital short bowel syndrome, 300048 (3), X-linked recessive; Otopalatodigital syndrome, type I, 311300 (3), X-linked dominant; Heterotopia, periventricular, 1, 300049 (3), X-linked dominant; Frontometaphyseal dysplasia 1, 305620 (3), X-linked recessive |
| *FLNB* | 603381 | Larsen syndrome, 150250 (3), Autosomal dominant; Atelosteogenesis, type I, 108720 (3), Autosomal dominant; Atelosteogenesis, type III, 108721 (3), Autosomal dominant; Spondylocarpotarsal synostosis syndrome, 272460 (3), Autosomal recessive; Boomerang dysplasia, 112310 (3), Autosomal dominant |
| *FLNC* | 102565 | Cardiomyopathy, familial hypertrophic, 26, 617047 (3), Autosomal dominant; Cardiomyopathy, familial restrictive 5, 617047 (3), Autosomal dominant; Myopathy, distal, 4, 614065 (3), Autosomal dominant; Myopathy, myofibrillar, 5, 609524 (3), Autosomal dominant |
| *FLRT3* | 604808 | Hypogonadotropic hypogonadism 21 with anosmia, 615271 (3), Autosomal dominant |
| *FLT3* | 136351 | Leukemia, acute lymphoblastic, somatic, 613065 (3); Leukemia, acute myeloid, reduced survival in, somatic, 601626 (3); Leukemia, acute myeloid, somatic, 601626 (3) |
| *FLT4* | 136352 | Hemangioma, capillary infantile, somatic, 602089 (3); Lymphatic malformation 1, 153100 (3), Autosomal dominant; Congenital heart defects, multiple types, 7, 618780 (3), Autosomal dominant |
| *FLVCR1* | 609144 | Ataxia, posterior column, with retinitis pigmentosa, 609033 (3), Autosomal recessive |
| *FLVCR2* | 610865 | Proliferative vasculopathy and hydranencephaly-hydrocephaly syndrome, 225790 (3), Autosomal recessive |
| *FMN2* | 606373 | Mental retardation, autosomal recessive 47, 616193 (3), Autosomal recessive |
| *FMO3* | 136132 | Trimethylaminuria, 602079 (3), Autosomal recessive |
| *FMR1* | 309550 | Fragile X tremor/ataxia syndrome, 300623 (3), X-linked dominant; Fragile X syndrome, 300624 (3), X-linked dominant; Premature ovarian failure 1, 311360 (3), X-linked |
| *FN1* | 135600 | Spondylometaphyseal dysplasia, corner fracture type, 184255 (3), Autosomal dominant; Glomerulopathy with fibronectin deposits 2, 601894 (3), Autosomal dominant |
| *FOLR1* | 136430 | Neurodegeneration due to cerebral folate transport deficiency, 613068 (3), Autosomal recessive |
| *FOXC1* | 601090 | Axenfeld-Rieger syndrome, type 3, 602482 (3), Autosomal dominant; Anterior segment dysgenesis 3, multiple subtypes, 601631 (3), Autosomal dominant |
| *FOXC2* | 602402 | Lymphedema-distichiasis syndrome, 153400 (3), Autosomal dominant; Lymphedema-distichiasis syndrome with renal disease and diabetes mellitus, 153400 (3), Autosomal dominant |
| *FOXD3* | 611539 | {Autoimmune disease, susceptibility to, 1}, 607836 (3), Autosomal dominant |
| *FOXE1* | 602617 | Bamforth-Lazarus syndrome, 241850 (3), Autosomal recessive; {Thyroid cancer, nonmedullary, 4}, 616534 (3), Autosomal dominant |
| *FOXE3* | 601094 | Anterior segment dysgenesis 2, multiple subtypes, 610256 (3), Autosomal recessive; {Aortic aneurysm, familial thoracic 11, susceptibility to}, 617349 (3), Autosomal dominant; Cataract 34, multiple types, 612968 (3) |
| *FOXF1* | 601089 | Alveolar capillary dysplasia with misalignment of pulmonary veins, 265380 (3), Autosomal dominant |
| *FOXG1* | 164874 | Rett syndrome, congenital variant, 613454 (3), Autosomal dominant |
| *FOXI1* | 601093 | Enlarged vestibular aqueduct, 600791 (3), Autosomal recessive |
| *FOXJ1* | 602291 | Ciliary dyskinesia, primary, 43, 618699 (3), Autosomal dominant |
| *FOXL2* | 605597 | Blepharophimosis, epicanthus inversus, and ptosis, type 2, 110100 (3), Autosomal recessive, Autosomal dominant; Blepharophimosis, epicanthus inversus, and ptosis, type 1, 110100 (3), Autosomal recessive, Autosomal dominant; Premature ovarian failure 3, 608996 (3), Autosomal dominant |
| *FOXN1* | 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 |
| *FOXO1* | 136533 | Rhabdomyosarcoma, alveolar, 268220 (3), Somatic mutation |
| *FOXP1* | 605515 | Mental retardation with language impairment and with or without autistic features, 613670 (3), Autosomal dominant |
| *FOXP2* | 605317 | Speech-language disorder-1, 602081 (3), Autosomal dominant |
| *FOXP3* | 300292 | Immunodysregulation, polyendocrinopathy, and enteropathy, X-linked, 304790 (3), X-linked recessive |
| *FOXRED1* | 613622 | Mitochondrial complex I deficiency, nuclear type 19, 618241 (3), Autosomal recessive |
| *FRAS1* | 607830 | Fraser syndrome 1, 219000 (3), Autosomal recessive |
| *FREM1* | 608944 | Manitoba oculotrichoanal syndrome, 248450 (3), Autosomal recessive; Bifid nose with or without anorectal and renal anomalies, 608980 (3); Trigonocephaly 2, 614485 (3), Autosomal dominant |
| *FREM2* | 608945 | Fraser syndrome 2, 617666 (3), Autosomal recessive; Cryptophthalmos, unilateral or bilateral, isolated, 123570 (3), Autosomal recessive |
| *FRMD4A* | 616305 | ?Corpus callosum, agenesis of, with facial anomalies and cerebellar ataxia, 616819 (3), Autosomal recessive |
| *FRMD7* | 300628 | Nystagmus, infantile periodic alternating, X-linked, 310700 (3), X-linked; Nystagmus 1, congenital, X-linked, 310700 (3), X-linked |
| *FRMPD4* | 300838 | Mental retardation, X-linked 104, 300983 (3), X-linked |
| *FRRS1L* | 604574 | Developmental and epileptic encephalopathy 37, 616981 (3), Autosomal recessive |
| *FRZB* | 605083 | {Osteoarthritis susceptibility 1}, 165720 (3), Multifactorial |
| *FSCN2* | 607643 | Retinitis pigmentosa 30, 607921 (3) |
| *FSHB* | 136530 | Hypogonadotropic hypogonadism 24 without anosmia, 229070 (3), Autosomal recessive |
| *FSHR* | 136435 | Ovarian response to FSH stimulation, 276400 (3), Autosomal recessive; Ovarian hyperstimulation syndrome, 608115 (3), Autosomal dominant; Ovarian dysgenesis 1, 233300 (3), Autosomal recessive |
| *FSIP2* | 615796 | Spermatogenic failure 34, 618153 (3), Autosomal recessive |
| *FTCD* | 606806 | Glutamate formiminotransferase deficiency, 229100 (3), Autosomal recessive |
| *FTH1* | 134770 | ?Hemochromatosis, type 5, 615517 (3), Autosomal dominant |
| *FTL* | 134790 | Hyperferritinemia-cataract syndrome, 600886 (3), Autosomal dominant; L-ferritin deficiency, dominant and recessive, 615604 (3), Autosomal recessive, Autosomal dominant; Neurodegeneration with brain iron accumulation 3, 606159 (3), Autosomal dominant |
| *FTO* | 610966 | Growth retardation, developmental delay, facial dysmorphism, 612938 (3), Autosomal recessive; {Obesity, susceptibility to, BMIQ14}, 612460 (3), Autosomal recessive |
| *FTSJ1* | 300499 | Mental retardation, X-linked 9/44, 309549 (3), X-linked recessive |
| *FUCA1* | 612280 | Fucosidosis, 230000 (3), Autosomal recessive |
| *FUS* | 137070 | Amyotrophic lateral sclerosis 6, with or without frontotemporal dementia, 608030 (3); Essential tremor, hereditary, 4, 614782 (3), Autosomal dominant |
| *FUT1* | 211100 | [Bombay phenotype], 616754 (3), Autosomal recessive |
| *FUT2* | 182100 | {Norwalk virus infection, resistance to} (3); {Vitamin B12 plasma level QTL1}, 612542 (3); [Bombay phenotype, digenic], 616754 (3), Autosomal recessive |
| *FUT3* | 111100 | [Blood group, Lewis], 618983 (3) |
| *FUT6* | 136836 | [Fucosyltransferase 6 deficiency], 613852 (3) |
| *FUT8* | 602589 | Congenital disorder of glycosylation with defective fucosylation 1, 618005 (3), Autosomal recessive |
| *FUZ* | 610622 | {Neural tube defects, susceptibility to}, 182940 (3), Autosomal dominant |
| *FXN* | 606829 | Friedreich ataxia with retained reflexes, 229300 (3), Autosomal recessive; Friedreich ataxia, 229300 (3), Autosomal recessive |
| *FXR1* | 600819 | ?Myopathy, congenital, with respiratory insufficiency and bone fractures, 618822 (3), Autosomal recessive; ?Myopathy, congenital proximal, with minicore lesions, 618823 (3), Autosomal recessive |
| *FXYD2* | 601814 | Hypomagnesemia 2, renal, 154020 (3), Autosomal dominant |
| *FYB1* | 602731 | Thrombocytopenia 3, 273900 (3), Autosomal recessive |
| *FYCO1* | 607182 | Cataract 18, autosomal recessive, 610019 (3), Autosomal recessive |
| *FZD2* | 600667 | Omodysplasia 2, 164745 (3), Autosomal dominant |
| *FZD4* | 604579 | Retinopathy of prematurity, 133780 (3), Autosomal dominant; Exudative vitreoretinopathy 1, 133780 (3), Autosomal dominant |
| *FZD5* | 601723 | Autosomal Dominant Coloboma. PMID: 33633439, 26908622 |
| *FZD6* | 603409 | Nail disorder, nonsyndromic congenital, 1, 161050 (3), Autosomal recessive |
| *G6PC1 (G6PC)* | 613742 | Glycogen storage disease Ia, 232200 (3), Autosomal recessive |
| *G6PC3* | 611045 | Dursun syndrome, 612541 (3), Autosomal recessive; Neutropenia, severe congenital 4, autosomal recessive, 612541 (3), Autosomal recessive |
| *G6PD* | 305900 | Hemolytic anemia, G6PD deficient (favism), 300908 (3), X-linked dominant; {Resistance to malaria due to G6PD deficiency}, 611162 (3) |
| *GAA* | 606800 | Glycogen storage disease II, 232300 (3), Autosomal recessive |
| *GAB1* | 604439 | ?Deafness, autosomal recessive 26, 605428 (3), Autosomal recessive |
| *GABBR2* | 607340 | {Nicotine dependence, protection against}, 188890 (3); {Nicotine dependence, susceptibility to}, 188890 (3); Developmental and epileptic encephalopathy 59, 617904 (3), Autosomal dominant; Neurodevelopmental disorder with poor language and loss of hand skills, 617903 (3), Autosomal dominant |
| *GABRA1* | 137160 | {Epilepsy, juvenile myoclonic, susceptibility to, 5}, 611136 (3); Developmental and epileptic encephalopathy 19, 615744 (3), Autosomal dominant; {Epilepsy, childhood absence, susceptibility to, 4}, 611136 (3) |
| *GABRA2* | 137140 | Developmental and epileptic encephalopathy 78, 618557 (3), Autosomal dominant; {Alcohol dependence, susceptibility to}, 103780 (3), Multifactorial |
| *GABRA5* | 137142 | Developmental and epileptic encephalopathy 79, 618559 (3), Autosomal dominant |
| *GABRB1* | 137190 | Developmental and epileptic encephalopathy 45, 617153 (3), Autosomal dominant |
| *GABRB2* | 600232 | Developmental and epileptic encephalopathy 92, 617829 (3), Autosomal dominant |
| *GABRB3* | 137192 | {Epilepsy, childhood absence, susceptibility to, 5}, 612269 (3); Developmental and epileptic encephalopathy 43, 617113 (3), Autosomal dominant |
| *GABRD* | 137163 | {Epilepsy, idiopathic generalized, 10}, 613060 (3), Autosomal dominant; {Epilepsy, generalized, with febrile seizures plus, type 5, susceptibility to}, 613060 (3), Autosomal dominant; {Epilepsy, juvenile myoclonic, susceptibility to}, 613060 (3), Autosomal dominant |
| *GABRG2* | 137164 | Developmental and epileptic encephalopathy 74, 618396 (3), Autosomal dominant; Epilepsy, generalized, with febrile seizures plus, type 3, 607681 (3), Autosomal dominant; Febrile seizures, familial, 8, 607681 (3), Autosomal dominant |
| *GAD1* | 605363 | Developmental and epileptic encephalopathy 89, 619124 (3), Autosomal recessive; ?Cerebral palsy, spastic quadriplegic, 1, 603513 (3), Autosomal recessive |
| *GAL* | 137035 | ?Epilepsy, familial temporal lobe, 8, 616461 (3), Autosomal dominant |
| *GALC* | 606890 | Krabbe disease, 245200 (3), Autosomal recessive |
| *GALE* | 606953 | Galactose epimerase deficiency, 230350 (3), Autosomal recessive |
| *GALK1* | 604313 | Galactokinase deficiency with cataracts, 230200 (3), Autosomal recessive |
| *GALM* | 137030 | Galactosemia IV, 618881 (3), Autosomal recessive |
| *GALNS* | 612222 | Mucopolysaccharidosis IVA, 253000 (3), Autosomal recessive |
| *GALNT12* | 610290 | {Colorectal cancer, susceptibility to, 1}, 608812 (3) |
| *GALNT2* | 602274 | Congenital disorder of glycosylation, type IIt, 618885 (3), Autosomal recessive |
| *GALNT3* | 601756 | Tumoral calcinosis, hyperphosphatemic, familial, 1, 211900 (3), Autosomal recessive |
| *GALT* | 606999 | Galactosemia, 230400 (3), Autosomal recessive |
| *GAMT* | 601240 | Cerebral creatine deficiency syndrome 2, 612736 (3), Autosomal recessive |
| *GAN* | 605379 | Giant axonal neuropathy-1, 256850 (3), Autosomal recessive |
| *GANAB* | 104160 | Polycystic kidney disease 3, 600666 (3), Autosomal dominant |
| *GARS1 (GARS)* | 600287 | Spinal muscular atrophy, infantile, James type, 619042 (3), Autosomal dominant; Neuronopathy, distal hereditary motor, type VA, 600794 (3), Autosomal dominant; Charcot-Marie-Tooth disease, type 2D, 601472 (3), Autosomal dominant |
| *GAS2L2* | 611398 | ?Ciliary dyskinesia, primary, 41, 618449 (3), Autosomal recessive |
| *GAS8* | 605178 | Ciliary dyskinesia, primary, 33, 616726 (3), Autosomal recessive |
| *GATA1* | 305371 | Leukemia, megakaryoblastic, with or without Down syndrome, somatic, 190685 (3); Thrombocytopenia, X-linked, with or without dyserythropoietic anemia, 300367 (3), X-linked recessive; Anemia, X-linked, with/without neutropenia and/or platelet abnormalities, 300835 (3), X-linked recessive; Thrombocytopenia with beta-thalassemia, X-linked, 314050 (3), X-linked recessive |
| *GATA2* | 137295 | {Leukemia, acute myeloid, susceptibility to}, 601626 (3), Somatic mutation, Autosomal dominant; Emberger syndrome, 614038 (3), Autosomal dominant; Immunodeficiency 21, 614172 (3), Autosomal dominant; {Myelodysplastic syndrome, susceptibility to}, 614286 (3) |
| *GATA3* | 131320 | Hypoparathyroidism, sensorineural deafness, and renal dysplasia, 146255 (3), Autosomal dominant |
| *GATA4* | 600576 | Tetralogy of Fallot, 187500 (3), Autosomal dominant; Atrial septal defect 2, 607941 (3), Autosomal dominant; Ventricular septal defect 1, 614429 (3), Autosomal dominant; Atrioventricular septal defect 4, 614430 (3), Autosomal dominant; ?Testicular anomalies with or without congenital heart disease, 615542 (3), Autosomal dominant |
| *GATA5* | 611496 | Congenital heart defects, multiple types, 5, 617912 (3), Autosomal recessive, Autosomal dominant |
| *GATA6* | 601656 | Atrial septal defect 9, 614475 (3), Autosomal dominant; Persistent truncus arteriosus, 217095 (3); Pancreatic agenesis and congenital heart defects, 600001 (3), Autosomal dominant; Atrioventricular septal defect 5, 614474 (3), Autosomal dominant; Tetralogy of Fallot, 187500 (3), Autosomal dominant |
| *GATAD1* | 614518 | ?Cardiomyopathy, dilated, 2B, 614672 (3), Autosomal recessive |
| *GATAD2B* | 614998 | GAND syndrome, 615074 (3), Autosomal dominant |
| *GATB* | 603645 | ?Combined oxidative phosphorylation deficiency 41, 618838 (3), Autosomal recessive |
| *GATC* | 617210 | Combined oxidative phosphorylation deficiency 42, 618839 (3), Autosomal recessive |
| *GATM* | 602360 | Cerebral creatine deficiency syndrome 3, 612718 (3), Autosomal recessive; Fanconi renotubular syndrome 1, 134600 (3), Autosomal dominant |
| *GBA* | 606463 | {Lewy body dementia, susceptibility to}, 127750 (3), Autosomal dominant; Gaucher disease, type II, 230900 (3), Autosomal recessive; Gaucher disease, type IIIC, 231005 (3), Autosomal recessive; Gaucher disease, type III, 231000 (3), Autosomal recessive; Gaucher disease, type I, 230800 (3), Autosomal recessive; Gaucher disease, perinatal lethal, 608013 (3), Autosomal recessive; {Parkinson disease, late-onset, susceptibility to}, 168600 (3), Multifactorial, Autosomal dominant |
| *GBA2* | 609471 | Spastic paraplegia 46, autosomal recessive, 614409 (3), Autosomal recessive |
| *GBE1* | 607839 | Glycogen storage disease IV, 232500 (3), Autosomal recessive; Polyglucosan body disease, adult form, 263570 (3), Autosomal recessive |
| *GCDH* | 608801 | Glutaricaciduria, type I, 231670 (3), Autosomal recessive |
| *GCGR* | 138033 | Mahvash disease, 619290 (3), Autosomal recessive |
| *GCH1* | 600225 | Dystonia, DOPA-responsive, with or without hyperphenylalaninemia, 128230 (3), Autosomal recessive, Autosomal dominant; Hyperphenylalaninemia, BH4-deficient, B, 233910 (3), Autosomal recessive |
| *GCK* | 138079 | MODY, type II, 125851 (3), Autosomal dominant; Diabetes mellitus, permanent neonatal 1, 606176 (3), Autosomal recessive; Hyperinsulinemic hypoglycemia, familial, 3, 602485 (3), Autosomal dominant; Diabetes mellitus, noninsulin-dependent, late onset, 125853 (3), Autosomal dominant |
| *GCKR* | 600842 | [Fasting plasma glucose level QTL 5], 613463 (3) |
| *GCLC* | 606857 | {Myocardial infarction, susceptibility to}, 608446 (3); Hemolytic anemia due to gamma-glutamylcysteine synthetase deficiency, 230450 (3), Autosomal recessive |
| *GCLM* | 601176 | {Myocardial infarction, susceptibility to}, 608446 (3) |
| *GCM2* | 603716 | Hypoparathyroidism, familial isolated 2, 618883 (3), Autosomal recessive, Autosomal dominant; Hyperparathyroidism 4, 617343 (3), Autosomal dominant |
| *GCNT2* | 600429 | [Blood group, Ii], 110800 (3), Autosomal dominant; Adult i phenotype without cataract, 110800 (3), Autosomal dominant; Cataract 13 with adult i phenotype, 116700 (3), Autosomal recessive |
| *GCSH* | 238330 | ?Glycine encephalopathy, 605899 (3), Autosomal recessive |
| *GDAP1* | 606598 | Charcot-Marie-Tooth disease, axonal, with vocal cord paresis, 607706 (3), Autosomal recessive; Charcot-Marie-Tooth disease, recessive intermediate, A, 608340 (3), Autosomal recessive; Charcot-Marie-Tooth disease, axonal, type 2K, 607831 (3), Autosomal recessive, Autosomal dominant; Charcot-Marie-Tooth disease, type 4A, 214400 (3), Autosomal recessive |
| *GDAP2* | 618128 | Spinocerebellar ataxia, autosomal recessive 27, 618369 (3), Autosomal recessive |
| *GDF1* | 602880 | Congenital heart defects, multiple types, 6, 613854 (3), Autosomal dominant; Right atrial isomerism (Ivemark), 208530 (3), Autosomal recessive |
| *GDF11* | 603936 | ?Vertebral hypersegmentation and orofacial anomalies, 619122 (3), Autosomal dominant |
| *GDF2* | 605120 | Telangiectasia, hereditary hemorrhagic, type 5, 615506 (3), Autosomal dominant |
| *GDF3* | 606522 | Klippel-Feil syndrome 3, autosomal dominant, 613702 (3); Microphthalmia with coloboma 6, 613703 (3), Autosomal dominant; Microphthalmia, isolated 7, 613704 (3), Autosomal dominant |
| *GDF5* | 601146 | Du Pan syndrome, 228900 (3), Autosomal recessive; Multiple synostoses syndrome 2, 610017 (3), Autosomal dominant; Symphalangism, proximal, 1B, 615298 (3), Autosomal dominant; ?Acromesomelic dysplasia, Hunter-Thompson type, 201250 (3), Autosomal recessive; Brachydactyly, type A2, 112600 (3), Autosomal dominant; Brachydactyly, type C, 113100 (3), Autosomal dominant; Chondrodysplasia, Grebe type, 200700 (3), Autosomal recessive; {Osteoarthritis-5}, 612400 (3); Brachydactyly, type A1, C, 615072 (3), Autosomal recessive, Autosomal dominant |
| *GDF6* | 601147 | Microphthalmia with coloboma 6, digenic, 613703 (3), Autosomal dominant; Microphthalmia, isolated 4, 613094 (3); Leber congenital amaurosis 17, 615360 (3), Autosomal recessive; Multiple synostoses syndrome 4, 617898 (3), Autosomal dominant; Klippel-Feil syndrome 1, autosomal dominant, 118100 (3), Autosomal dominant |
| *GDF9* | 601918 | ?Premature ovarian failure 14, 618014 (3), Autosomal recessive |
| *GDI1* | 300104 | Mental retardation, X-linked 41, 300849 (3), X-linked dominant |
| *GDNF* | 600837 | Central hypoventilation syndrome, 209880 (3), Autosomal dominant; {Pheochromocytoma, modifier of}, 171300 (3), Autosomal dominant; {Hirschsprung disease, susceptibility to, 3}, 613711 (3), Autosomal dominant |
| *GEMIN4* | 606969 | Neurodevelopmental disorder with microcephaly, cataracts, and renal abnormalities, 617913 (3), Autosomal recessive |
| *GEMIN5* | 607005 | Neurodevelopmental disorder with cerebellar atrophy and motor dysfunction, 619333 (3), Autosomal recessive |
| *GFAP* | 137780 | Alexander disease, 203450 (3), Autosomal dominant |
| *GFER* | 600924 | Myopathy, mitochondrial progressive, with congenital cataract and developmental delay, 613076 (3), Autosomal recessive |
| *GFI1* | 600871 | ?Neutropenia, nonimmune chronic idiopathic, of adults, 607847 (3), Autosomal dominant; Neutropenia, severe congenital 2, autosomal dominant, 613107 (3), Autosomal dominant |
| *GFI1B* | 604383 | Bleeding disorder, platelet-type, 17, 187900 (3), Autosomal recessive, Autosomal dominant |
| *GFM1* | 606639 | Combined oxidative phosphorylation deficiency 1, 609060 (3), Autosomal recessive |
| *GFM2* | 606544 | Combined oxidative phosphorylation deficiency 39, 618397 (3), Autosomal recessive |
| *GFPT1* | 138292 | Myasthenia, congenital, 12, with tubular aggregates, 610542 (3), Autosomal recessive |
| *GGCX* | 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) |
| *GGT1* | 612346 | ?Glutathioninuria, 231950 (3), Autosomal recessive |
| *GGT2* | 137181 | [Gamma-glutamyltransferase, familial high serum], 137181 (2) |
| *GH1* | 139250 | Kowarski syndrome, 262650 (3), Autosomal recessive; Growth hormone deficiency, isolated, type II, 173100 (3), Autosomal dominant; Growth hormone deficiency, isolated, type IB, 612781 (3); Growth hormone deficiency, isolated, type IA, 262400 (3), Autosomal recessive |
| *GHR* | 600946 | Laron dwarfism, 262500 (3), Autosomal recessive; Increased responsiveness to growth hormone, 604271 (3), Autosomal dominant; Growth hormone insensitivity, partial, 604271 (3), Autosomal dominant; {Hypercholesterolemia, familial, modifier of}, 143890 (3), Autosomal recessive, Autosomal dominant |
| *GHRH* | 139190 | Gigantism due to GHRF hypersecretion (1); ?Isolated growth hormone deficiency due to defect in GHRF (1) |
| *GHRHR* | 139191 | Growth hormone deficiency, isolated, type IV, 618157 (3), Autosomal recessive |
| *GHRL* | 605353 | {Obesity, susceptibility to}, 601665 (3), Multifactorial, Autosomal recessive, Autosomal dominant |
| *GHSR* | 601898 | Growth hormone deficiency, isolated partial, 615925 (3), Autosomal recessive, Autosomal dominant |
| *GIGYF2* | 612003 | {Parkinson disease 11}, 607688 (3) |
| *GINS1* | 610608 | Immunodeficiency 55, 617827 (3), Autosomal recessive |
| *GIPC1* | 605072 | Oculopharyngodistal myopathy 2, 618940 (3), Autosomal dominant |
| *GIPC3* | 608792 | Deafness, autosomal recessive 15, 601869 (3), Autosomal recessive |
| *GJA1* | 121014 | Erythrokeratodermia variabilis et progressiva 3, 617525 (3), Autosomal dominant; Craniometaphyseal dysplasia, autosomal recessive, 218400 (3), Autosomal recessive; Oculodentodigital dysplasia, 164200 (3), Autosomal dominant; Hypoplastic left heart syndrome 1, 241550 (3), Autosomal recessive; Palmoplantar keratoderma with congenital alopecia, 104100 (3), Autosomal dominant; Syndactyly, type III, 186100 (3), Autosomal dominant; Oculodentodigital dysplasia, autosomal recessive, 257850 (3), Autosomal recessive; Atrioventricular septal defect 3, 600309 (3), Autosomal dominant |
| *GJA3* | 121015 | Cataract 14, multiple types, 601885 (3), Autosomal dominant |
| *GJA5* | 121013 | Atrial fibrillation, familial, 11, 614049 (3), Autosomal dominant; Atrial standstill, digenic (GJA5/SCN5A), 108770 (3), Autosomal dominant |
| *GJA8* | 600897 | Cataract 1, multiple types, 116200 (3), Autosomal dominant |
| *GJB1* | 304040 | Charcot-Marie-Tooth neuropathy, X-linked dominant, 1, 302800 (3), X-linked dominant |
| *GJB2* | 121011 | Keratoderma, palmoplantar, with deafness, 148350 (3), Autosomal dominant; Deafness, autosomal recessive 1A, 220290 (3), Digenic dominant, Autosomal recessive; Deafness, autosomal dominant 3A, 601544 (3), Autosomal dominant; Hystrix-like ichthyosis with deafness, 602540 (3), Autosomal dominant; Bart-Pumphrey syndrome, 149200 (3), Autosomal dominant; Keratitis-ichthyosis-deafness syndrome, 148210 (3), Autosomal dominant; Vohwinkel syndrome, 124500 (3), Autosomal dominant |
| *GJB3* | 603324 | Deafness, digenic, GJB2/GJB3, 220290 (3), Digenic dominant, Autosomal recessive; Deafness, autosomal recessive (3); Deafness, autosomal dominant 2B, 612644 (3), Autosomal dominant; Erythrokeratodermia variabilis et progressiva 1, 133200 (3), Autosomal recessive, Autosomal dominant; Deafness, autosomal dominant, with peripheral neuropathy (3) |
| *GJB4* | 605425 | Erythrokeratodermia variabilis et progressiva 2, 617524 (3), Autosomal dominant |
| *GJB6* | 604418 | Ectodermal dysplasia 2, Clouston type, 129500 (3), Autosomal dominant; Deafness, autosomal dominant 3B, 612643 (3), Autosomal dominant; Deafness, autosomal recessive 1B, 612645 (3), Autosomal recessive; Deafness, digenic GJB2/GJB6, 220290 (3), Digenic dominant, Autosomal recessive |
| *GJC2* | 608803 | Lymphatic malformation 3, 613480 (3), Autosomal dominant; Spastic paraplegia 44, autosomal recessive, 613206 (3), Autosomal recessive; Leukodystrophy, hypomyelinating, 2, 608804 (3), Autosomal recessive |
| *GK* | 300474 | Glycerol kinase deficiency, 307030 (3), X-linked recessive |
| *GLA* | 300644 | Fabry disease, cardiac variant, 301500 (3), X-linked; Fabry disease, 301500 (3), X-linked |
| *GLB1* | 611458 | GM1-gangliosidosis, type I, 230500 (3), Autosomal recessive; GM1-gangliosidosis, type III, 230650 (3), Autosomal recessive; Mucopolysaccharidosis type IVB (Morquio), 253010 (3), Autosomal recessive; GM1-gangliosidosis, type II, 230600 (3), Autosomal recessive |
| *GLCCI1* | 614283 | {Glucocorticoid therapy, response to}, 614400 (3) |
| *GLDC* | 238300 | Glycine encephalopathy, 605899 (3), Autosomal recessive |
| *GLDN* | 608603 | Lethal congenital contracture syndrome 11, 617194 (3), Autosomal recessive |
| *GLE1* | 603371 | Lethal congenital contracture syndrome 1, 253310 (3), Autosomal recessive; Congenital arthrogryposis with anterior horn cell disease, 611890 (3), Autosomal recessive |
| *GLI1* | 165220 | Polydactyly, preaxial I, 174400 (3), Autosomal recessive; Polydactyly, postaxial, type A8, 618123 (3), Autosomal recessive |
| *GLI2* | 165230 | Culler-Jones syndrome, 615849 (3), Autosomal dominant; Holoprosencephaly 9, 610829 (3), Autosomal dominant |
| *GLI3* | 165240 | Greig cephalopolysyndactyly syndrome, 175700 (3), Autosomal dominant; Polydactyly, postaxial, types A1 and B, 174200 (3), Autosomal dominant; Pallister-Hall syndrome, 146510 (3), Autosomal dominant; Polydactyly, preaxial, type IV, 174700 (3), Autosomal dominant |
| *GLIS2* | 608539 | Nephronophthisis 7, 611498 (3) |
| *GLIS3* | 610192 | Diabetes mellitus, neonatal, with congenital hypothyroidism, 610199 (3), Autosomal recessive |
| *GLMN* | 601749 | Glomuvenous malformations, 138000 (3), Autosomal dominant |
| *GLRA1* | 138491 | Hyperekplexia 1, 149400 (3), Autosomal recessive, Autosomal dominant |
| *GLRB* | 138492 | Hyperekplexia 2, 614619 (3), Autosomal recessive |
| *GLRX5* | 609588 | Anemia, sideroblastic, 3, pyridoxine-refractory, 616860 (3), Autosomal recessive; Spasticity, childhood-onset, with hyperglycinemia, 616859 (3), Autosomal recessive |
| *GLS* | 138280 | Global developmental delay, progressive ataxia, and elevated glutamine, 618412 (3), Autosomal recessive; ?Infantile cataract, skin abnormalities, glutamate excess, and impaired intellectual development, 618339 (3), Autosomal dominant; Developmental and epileptic encephalopathy 71, 618328 (3), Autosomal recessive |
| *GLUD1* | 138130 | Hyperinsulinism-hyperammonemia syndrome, 606762 (3), Autosomal dominant |
| *GLUD2* | 300144 | {Parkinson disease, age of onset, modifier}, 168600 (3), Multifactorial, Autosomal dominant |
| *GLUL* | 138290 | Glutamine deficiency, congenital, 610015 (3), Autosomal recessive |
| *GLYCTK* | 610516 | D-glyceric aciduria, 220120 (3), Autosomal recessive |
| *GM2A* | 613109 | GM2-gangliosidosis, AB variant, 272750 (3), Autosomal recessive |
| *GMNN* | 602842 | Meier-Gorlin syndrome 6, 616835 (3), Autosomal dominant |
| *GMPPA* | 615495 | Alacrima, achalasia, and mental retardation syndrome, 615510 (3), Autosomal recessive |
| *GMPPB* | 615320 | Muscular dystrophy-dystroglycanopathy (limb-girdle), type C, 14, 615352 (3), Autosomal recessive; Muscular dystrophy-dystroglycanopathy (congenital with mental retardation), type B, 14, 615351 (3), Autosomal recessive; Muscular dystrophy-dystroglycanopathy (congenital with brain and eye anomalies), type A, 14, 615350 (3), Autosomal recessive |
| *GNA11* | 139313 | Hypocalciuric hypercalcemia, type II, 145981 (3), Autosomal dominant; Hypocalcemia, autosomal dominant 2, 615361 (3), Autosomal dominant |
| *GNAI1* | 139310 | GNAI1-related Neurodevelopmental disorder PMID: 33473207 |
| *GNAI2* | 139360 | Ventricular tachycardia, idiopathic, 192605 (3), Autosomal dominant; Pituitary adenoma, ACTH-secreting, somatic (3) |
| *GNAI3* | 139370 | Auriculocondylar syndrome 1, 602483 (3), Autosomal dominant |
| *GNAL* | 139312 | Dystonia 25, 615073 (3), Autosomal dominant |
| *GNAO1* | 139311 | Developmental and epileptic encephalopathy 17, 615473 (3), Autosomal dominant; Neurodevelopmental disorder with involuntary movements, 617493 (3), Autosomal dominant |
| *GNAQ* | 600998 | Capillary malformations, congenital, 1, somatic, mosaic, 163000 (3); Sturge-Weber syndrome, somatic, mosaic, 185300 (3) |
| *GNAS* | 139320 | ACTH-independent macronodular adrenal hyperplasia, 219080 (3), Somatic mutation; Pituitary adenoma 3, multiple types, somatic, 617686 (3); Pseudohypoparathyroidism Ic, 612462 (3), Autosomal dominant; Pseudohypoparathyroidism Ia, 103580 (3), Autosomal dominant; Osseous heteroplasia, progressive, 166350 (3), Autosomal dominant; Pseudohypoparathyroidism Ib, 603233 (3), Autosomal dominant; McCune-Albright syndrome, somatic, mosaic, 174800 (3); Pseudopseudohypoparathyroidism, 612463 (3), Autosomal dominant |
| *GNAT1* | 139330 | Night blindness, congenital stationary, autosomal dominant 3, 610444 (3), Autosomal dominant; Night blindness, congenital stationary, type 1G, 616389 (3), Autosomal recessive |
| *GNAT2* | 139340 | Achromatopsia 4, 613856 (3) |
| *GNB1* | 139380 | Myelodysplastic syndrome, somatic, 614286 (3); Leukemia, acute lymphoblastic, somatic, 613065 (3); Mental retardation, autosomal dominant 42, 616973 (3), Autosomal dominant |
| *GNB3* | 139130 | Night blindness, congenital stationary, type 1H, 617024 (3), Autosomal recessive; {Hypertension, essential, susceptibility to}, 145500 (3), Multifactorial |
| *GNB4* | 610863 | Charcot-Marie-Tooth disease, dominant intermediate F, 615185 (3), Autosomal dominant |
| *GNB5* | 604447 | Language delay and ADHD/cognitive impairment with or without cardiac arrhythmia, 617182 (3), Autosomal recessive; Intellectual developmental disorder with cardiac arrhythmia, 617173 (3), Autosomal recessive |
| *GNE* | 603824 | Sialuria, 269921 (3), Autosomal dominant; Nonaka myopathy, 605820 (3), Autosomal recessive |
| *GNMT* | 606628 | Glycine N-methyltransferase deficiency, 606664 (3), Autosomal recessive |
| *GNPAT* | 602744 | Rhizomelic chondrodysplasia punctata, type 2, 222765 (3), Autosomal recessive |
| *GNPTAB* | 607840 | Mucolipidosis III alpha/beta, 252600 (3), Autosomal recessive; Mucolipidosis II alpha/beta, 252500 (3), Autosomal recessive |
| *GNPTG* | 607838 | Mucolipidosis III gamma, 252605 (3), Autosomal recessive |
| *GNRH1* | 152760 | ?Hypogonadotropic hypogonadism 12 with or without anosmia, 614841 (3), Autosomal recessive |
| *GNRHR* | 138850 | Hypogonadotropic hypogonadism 7 without anosmia, 146110 (3), Autosomal recessive |
| *GNS* | 607664 | Mucopolysaccharidosis type IIID, 252940 (3), Autosomal recessive |
| *GORAB* | 607983 | Geroderma osteodysplasticum, 231070 (3), Autosomal recessive |
| *GOSR2* | 604027 | Epilepsy, progressive myoclonic 6, 614018 (3), Autosomal recessive |
| *GOT1* | 138180 | Aspartate aminotransferase, serum level of, QTL1, 614419 (3) |
| *GOT2* | 138150 | Developmental and epileptic encephalopathy 82, 618721 (3), Autosomal recessive |
| *GP1BA* | 606672 | Bernard-Soulier syndrome, type A1 (recessive), 231200 (3), Autosomal recessive; Bernard-Soulier syndrome, type A2 (dominant), 153670 (3), Autosomal dominant; von Willebrand disease, platelet-type, 177820 (3), Autosomal dominant; {Nonarteritic anterior ischemic optic neuropathy, susceptibility to}, 258660 (3), Autosomal recessive |
| *GP1BB* | 138720 | Giant platelet disorder, isolated, 231200 (3), Autosomal recessive; Bernard-Soulier syndrome, type B, 231200 (3), Autosomal recessive |
| *GP6* | 605546 | Bleeding disorder, platelet-type, 11, 614201 (3), Autosomal recessive |
| *GP9* | 173515 | Bernard-Soulier syndrome, type C, 231200 (3), Autosomal recessive |
| *GPAA1* | 603048 | Glycosylphosphatidylinositol biosynthesis defect 15, 617810 (3), Autosomal recessive |
| *GPC3* | 300037 | Wilms tumor, somatic, 194070 (3); Simpson-Golabi-Behmel syndrome, type 1, 312870 (3), X-linked recessive |
| *GPC4* | 300168 | Keipert syndrome, 301026 (3), X-linked recessive |
| *GPC6* | 604404 | Omodysplasia 1, 258315 (3), Autosomal recessive |
| *GPD1* | 138420 | Hypertriglyceridemia, transient infantile, 614480 (3), Autosomal recessive |
| *GPD1L* | 611778 | Brugada syndrome 2, 611777 (3) |
| *GPD2* | 138430 | {Type 2 diabetes mellitus, susceptibility to}, 125853 (3), Autosomal dominant |
| *GPHN* | 603930 | Molybdenum cofactor deficiency C, 615501 (3), Autosomal recessive |
| *GPI* | 172400 | Hemolytic anemia, nonspherocytic, due to glucose phosphate isomerase deficiency, 613470 (3), Autosomal recessive |
| *GPIHBP1* | 612757 | Hyperlipoproteinemia, type 1D, 615947 (3), Autosomal recessive |
| *GPNMB* | 604368 | Amyloidosis, primary localized cutaneous, 3, 617920 (3), Autosomal recessive |
| *GPR101* | 300393 | Pituitary adenoma 2, GH-secreting, 300943 (3), X-linked |
| *GPR143* | 300808 | Ocular albinism, type I, Nettleship-Falls type, 300500 (3), X-linked; Nystagmus 6, congenital, X-linked, 300814 (3), X-linked recessive |
| *GPR179* | 614515 | Night blindness, congenital stationary (complete), 1E, autosomal recessive, 614565 (3), Autosomal recessive |
| *GPR68* | 601404 | Amelogenesis imperfecta, hypomaturation type, IIA6, 617217 (3), Autosomal recessive |
| *GPR88* | 607468 | ?Chorea, childhood-onset, with psychomotor retardation, 616939 (3), Autosomal recessive |
| *GPRASP2* | 300969 | ?Deafness, X-linked 7, 301018 (3), X-linked recessive |
| *GPSM2* | 609245 | Chudley-McCullough syndrome, 604213 (3), Autosomal recessive |
| *GPT2* | 138210 | Neurodevelopmental disorder with microcephaly and spastic paraplegia, 616281 (3), Autosomal recessive |
| *GPX1* | 138320 | Hemolytic anemia due to glutathione peroxidase deficiency, 614164 (1), Autosomal recessive |
| *GPX4* | 138322 | Spondylometaphyseal dysplasia, Sedaghatian type, 250220 (3), Autosomal recessive |
| *GRAP* | 604330 | Deafness, autosomal recessive 114, 618456 (3), Autosomal recessive |
| *GREB1L* | 617782 | Deafness, autosomal dominant 80, 619274 (3), Autosomal dominant; Renal hypodysplasia/aplasia 3, 617805 (3), Autosomal dominant |
| *GREM2* | 608832 | Tooth agenesis, selective, 9, 617275 (3), Autosomal dominant |
| *GRHL2* | 608576 | Deafness, autosomal dominant 28, 608641 (3), Autosomal dominant; Ectodermal dysplasia/short stature syndrome, 616029 (3), Autosomal recessive; Corneal dystrophy, posterior polymorphous, 4, 618031 (3), Autosomal dominant |
| *GRHL3* | 608317 | Van der Woude syndrome 2, 606713 (3), Autosomal dominant |
| *GRHPR* | 604296 | Hyperoxaluria, primary, type II, 260000 (3), Autosomal recessive |
| *GRIA2* | 138247 | Neurodevelopmental disorder with language impairment and behavioral abnormalities, 618917 (3), Autosomal dominant |
| *GRIA3* | 305915 | Intellectual developmental disorder, X-linked, syndromic, Wu type, 300699 (3), X-linked recessive |
| *GRIA4* | 138246 | Neurodevelopmental disorder with or without seizures and gait abnormalities, 617864 (3), Autosomal dominant |
| *GRID2* | 602368 | Spinocerebellar ataxia, autosomal recessive 18, 616204 (3), Autosomal recessive |
| *GRIK2* | 138244 | Mental retardation, autosomal recessive, 6, 611092 (3), Autosomal recessive |
| *GRIN1* | 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 |
| *GRIN2A* | 138253 | Epilepsy, focal, with speech disorder and with or without mental retardation, 245570 (3), Autosomal dominant |
| *GRIN2B* | 138252 | Developmental and epileptic encephalopathy 27, 616139 (3), Autosomal dominant; Intellectual developmental disorder, autosomal dominant 6, with or without seizures, 613970 (3), Autosomal dominant |
| *GRIN2D* | 602717 | Developmental and epileptic encephalopathy 46, 617162 (3), Autosomal dominant |
| *GRIP1* | 604597 | Fraser syndrome 3, 617667 (3), Autosomal recessive |
| *GRK1* | 180381 | Oguchi disease-2, 613411 (3) |
| *GRM1* | 604473 | Spinocerebellar ataxia, autosomal recessive 13, 614831 (3), Autosomal recessive; Spinocerebellar ataxia 44, 617691 (3), Autosomal dominant |
| *GRM6* | 604096 | Night blindness, congenital stationary (complete), 1B, autosomal recessive, 257270 (3), Autosomal recessive |
| *GRM7* | 604101 | Neurodevelopmental disorder with seizures, hypotonia, and brain abnormalities, 618922 (3), Autosomal recessive |
| *GRN* | 138945 | Aphasia, primary progressive, 607485 (3), Autosomal dominant; Frontotemporal lobar degeneration with ubiquitin-positive inclusions, 607485 (3), Autosomal dominant; Ceroid lipofuscinosis, neuronal, 11, 614706 (3), Autosomal recessive |
| *GRXCR1* | 613283 | Deafness, autosomal recessive 25, 613285 (3), Autosomal recessive |
| *GRXCR2* | 615762 | ?Deafness, autosomal recessive 101, 615837 (3), Autosomal recessive |
| *GSC* | 138890 | Short stature, auditory canal atresia, mandibular hypoplasia, skeletal abnormalities, 602471 (3), Autosomal recessive |
| *GSDME* | 608798 | Deafness, autosomal dominant 5, 600994 (3), Autosomal dominant |
| *GSN* | 137350 | Amyloidosis, Finnish type, 105120 (3), Autosomal dominant |
| *GSR* | 138300 | Hemolytic anemia due to glutathione reductase deficiency, 618660 (3), Autosomal recessive |
| *GSS* | 601002 | Hemolytic anemia due to glutathione synthetase deficiency, 231900 (3), Autosomal recessive; Glutathione synthetase deficiency, 266130 (3), Autosomal recessive |
| *GSTZ1* | 603758 | [Maleylacetoacetate isomerase deficiency], 617596 (3), Autosomal recessive |
| *GSX2* | 616253 | Diencephalic-mesencephalic junction dysplasia syndrome 2, 618646 (3), Autosomal recessive |
| *GTF2E2* | 189964 | Trichothiodystrophy 6, nonphotosensitive, 616943 (3), Autosomal recessive |
| *GTF2H5* | 608780 | Trichothiodystrophy 3, photosensitive, 616395 (3) |
| *GTPBP2* | 607434 | Jaberi-Elahi syndrome, 617988 (3), Autosomal recessive |
| *GTPBP3* | 608536 | Combined oxidative phosphorylation deficiency 23, 616198 (3), Autosomal recessive |
| *GUCA1A* | 600364 | Cone-rod dystrophy 14, 602093 (3), Autosomal dominant; Cone dystrophy-3, 602093 (3), Autosomal dominant |
| *GUCA1B* | 602275 | Retinitis pigmentosa 48, 613827 (3) |
| *GUCY1A1* | 139396 | Moyamoya 6 with achalasia, 615750 (3), Autosomal recessive |
| *GUCY2C* | 601330 | Diarrhea 6, 614616 (3), Autosomal dominant; Meconium ileus, 614665 (3), Autosomal recessive |
| *GUCY2D* | 600179 | Cone-rod dystrophy 6, 601777 (3), Autosomal recessive, Autosomal dominant; ?Choroidal dystrophy, central areolar 1, 215500 (3), Autosomal dominant; Leber congenital amaurosis 1, 204000 (3), Autosomal recessive; Night blindness, congenital stationary, type 1I, 618555 (3), Autosomal recessive |
| *GUF1* | 617064 | ?Developmental and epileptic encephalopathy 40, 617065 (3), Autosomal recessive |
| *GUSB* | 611499 | Mucopolysaccharidosis VII, 253220 (3), Autosomal recessive |
| *GYG1* | 603942 | ?Glycogen storage disease XV, 613507 (3), Autosomal recessive; Polyglucosan body myopathy 2, 616199 (3), Autosomal recessive |
| *GYPA* | 617922 | {Malaria, resistance to}, 611162 (3); [Blood group, MNSs system], 111300 (3) |
| *GYPB* | 617923 | [Blood group, Ss], 111740 (3); {Malaria, resistance to}, 611162 (3) |
| *GYPC* | 110750 | [Blood group, Gerbich], 616089 (3); {Malaria, resistance to}, 611162 (3) |
| *GYS1* | 138570 | Glycogen storage disease 0, muscle, 611556 (3), Autosomal recessive |
| *GYS2* | 138571 | Glycogen storage disease 0, liver, 240600 (3), Autosomal recessive |
| *GZF1* | 613842 | Joint laxity, short stature, and myopia, 617662 (3), Autosomal recessive |
| *H1-4 (HIST1H1E)* | 142220 | Rahman syndrome, 617537 (3), Autosomal dominant |
| *H4C3 (HIST1H4C)* | 602827 | HIST1H4C-associated neurodevelopmental disorder PMID:  31804630 |
| *H6PD* | 138090 | Cortisone reductase deficiency 1, 604931 (3), Autosomal recessive |
| *HAAO* | 604521 | Vertebral, cardiac, renal, and limb defects syndrome 1, 617660 (3), Autosomal recessive |
| *HABP2* | 603924 | {?Thyroid cancer, nonmedullary, 5}, 616535 (3), Autosomal dominant; {Venous thromboembolism, susceptibility to}, 188050 (3), Autosomal dominant |
| *HACE1* | 610876 | Spastic paraplegia and psychomotor retardation with or without seizures, 616756 (3), Autosomal recessive |
| *HADH* | 601609 | Hyperinsulinemic hypoglycemia, familial, 4, 609975 (3), Autosomal recessive; 3-hydroxyacyl-CoA dehydrogenase deficiency, 231530 (3), Autosomal recessive |
| *HADHA* | 600890 | HELLP syndrome, maternal, of pregnancy, 609016 (3), Autosomal recessive; Mitochondrial trifunctional protein deficiency, 609015 (3), Autosomal recessive; LCHAD deficiency, 609016 (3), Autosomal recessive; Fatty liver, acute, of pregnancy, 609016 (3), Autosomal recessive |
| *HADHB* | 143450 | Trifunctional protein deficiency, 609015 (3), Autosomal recessive |
| *HAGH* | 138760 | [Glyoxalase II deficiency], 614033 (1), Autosomal dominant |
| *HAL* | 609457 | [Histidinemia], 235800 (3), Autosomal recessive, Autosomal dominant |
| *HAMP* | 606464 | Hemochromatosis, type 2B, 613313 (3), Autosomal recessive |
| *HARS1 (HARS)* | 142810 | Charcot-Marie-Tooth disease, axonal, type 2W, 616625 (3), Autosomal dominant; Usher syndrome type 3B, 614504 (3), Autosomal recessive |
| *HARS2* | 600783 | Perrault syndrome 2, 614926 (3), Autosomal recessive |
| *HAVCR2* | 606652 | T-cell lymphoma, subcutaneous panniculitis-like, 618398 (3), Autosomal recessive |
| *HAX1* | 605998 | Neutropenia, severe congenital 3, autosomal recessive, 610738 (3), Autosomal recessive |
| *HBA1* | 141800 | Hemoglobin H disease, nondeletional, 613978 (3); Thalassemias, alpha-, 604131 (3); Heinz body anemias, alpha-, 140700 (3), Autosomal dominant; Methemoglobinemia, alpha type, 617973 (3), Autosomal dominant; Erythrocytosis 7, 617981 (3), Autosomal dominant |
| *HBA2* | 141850 | Heinz body anemia, 140700 (3), Autosomal dominant; Erythrocytosis 7, 617981 (3), Autosomal dominant; Thalassemia, alpha-, 604131 (3); Hemoglobin H disease, deletional and nondeletional, 613978 (3) |
| *HBB* | 141900 | Methemoglobinemia, beta type, 617971 (3), Autosomal dominant; Thalassemia-beta, dominant inclusion-body, 603902 (3); Sickle cell anemia, 603903 (3), Autosomal recessive; Thalassemia, beta, 613985 (3); Delta-beta thalassemia, 141749 (3), Autosomal dominant; {Malaria, resistance to}, 611162 (3); Hereditary persistence of fetal hemoglobin, 141749 (3), Autosomal dominant; Heinz body anemia, 140700 (3), Autosomal dominant; Erythrocytosis 6, 617980 (3), Autosomal dominant |
| *HBD* | 142000 | Thalassemia due to Hb Lepore (3); Thalassemia, delta- (3) |
| *HBEGF* | 126150 | {Diphtheria, susceptibility to} (1) |
| *HBG1* | 142200 | Fetal hemoglobin quantitative trait locus 1, 141749 (3), Autosomal dominant |
| *HBG2* | 142250 | Fetal hemoglobin quantitative trait locus 1, 141749 (3), Autosomal dominant; Cyanosis, transient neonatal, 613977 (3), Autosomal dominant |
| *HCCS* | 300056 | Linear skin defects with multiple congenital anomalies 1, 309801 (3), X-linked dominant |
| *HCFC1* | 300019 | Mental retardation, X-linked 3 (methylmalonic acidemia and homocysteinemia, cblX type ), 309541 (3), X-linked recessive |
| *HCN1* | 602780 | Developmental and epileptic encephalopathy 24, 615871 (3), Autosomal dominant; Generalized epilepsy with febrile seizures plus, type 10, 618482 (3), Autosomal dominant |
| *HCN4* | 605206 | Sick sinus syndrome 2, 163800 (3), Autosomal dominant; Brugada syndrome 8, 613123 (3) |
| *HCRT* | 602358 | ?Narcolepsy 1, 161400 (3), Autosomal dominant |
| *HDAC4* | 605314/600430 | -/Chromosome 2q37 deletion syndrome, 600430 (4), Autosomal dominant |
| *HDAC6* | 300272 | ?Chondrodysplasia with platyspondyly, distinctive brachydactyly, hydrocephaly, and microphthalmia, 300863 (3), X-linked dominant |
| *HDAC8* | 300269 | Cornelia de Lange syndrome 5, 300882 (3), X-linked dominant |
| *HDC* | 142704 | {Gilles de la Tourette syndrome, susceptibility to}, 137580 (3), Autosomal dominant |
| *HECW2* | 617245 | Neurodevelopmental disorder with hypotonia, seizures, and absent language, 617268 (3), Autosomal dominant |
| *HELLS* | 603946 | Immunodeficiency-centromeric instability-facial anomalies syndrome 4, 616911 (3), Autosomal recessive |
| *HEPACAM* | 611642 | Megalencephalic leukoencephalopathy with subcortical cysts 2A, 613925 (3), Autosomal recessive; Megalencephalic leukoencephalopathy with subcortical cysts 2B, remitting, with or without mental retardation, 613926 (3), Autosomal dominant |
| *HEPHL1* | 618455 | ?Abnormal hair, joint laxity, and developmental delay, 261990 (3), Autosomal recessive |
| *HERC1* | 605109 | Macrocephaly, dysmorphic facies, and psychomotor retardation, 617011 (3), Autosomal recessive |
| *HERC2* | 605837 | Mental retardation, autosomal recessive 38, 615516 (3), Autosomal recessive; [Skin/hair/eye pigmentation 1, blond/brown hair], 227220 (3), Autosomal recessive; [Skin/hair/eye pigmentation 1, blue/nonblue eyes], 227220 (3), Autosomal recessive |
| *HES7* | 608059 | Spondylocostal dysostosis 4, autosomal recessive, 613686 (3), Autosomal recessive |
| *HESX1* | 601802 | Pituitary hormone deficiency, combined, 5, 182230 (3), Autosomal recessive, Autosomal dominant; Septooptic dysplasia, 182230 (3), Autosomal recessive, Autosomal dominant; Growth hormone deficiency with pituitary anomalies, 182230 (3), Autosomal recessive, Autosomal dominant |
| *HEXA* | 606869 | [Hex A pseudodeficiency], 272800 (3), Autosomal recessive; GM2-gangliosidosis, several forms, 272800 (3), Autosomal recessive; Tay-Sachs disease, 272800 (3), Autosomal recessive |
| *HEXB* | 606873 | Sandhoff disease, infantile, juvenile, and adult forms, 268800 (3), Autosomal recessive |
| *HFE* | 613609 | {Porphyria variegata, susceptibility to}, 176200 (3), Autosomal dominant; {Microvascular complications of diabetes 7}, 612635 (3); Hemochromatosis, 235200 (3), Autosomal recessive; {Alzheimer disease, susceptibility to}, 104300 (3), Autosomal dominant; [Transferrin serum level QTL2], 614193 (3); {Porphyria cutanea tarda, susceptibility to}, 176100 (3), Autosomal recessive, Autosomal dominant |
| *HFM1* | 615684 | Premature ovarian failure 9, 615724 (3), Autosomal recessive |
| *HGD* | 607474 | Alkaptonuria, 203500 (3), Autosomal recessive |
| *HGF* | 142409 | Deafness, autosomal recessive 39, 608265 (3), Autosomal recessive |
| *HGSNAT* | 610453 | Mucopolysaccharidosis type IIIC (Sanfilippo C), 252930 (3), Autosomal recessive; Retinitis pigmentosa 73, 616544 (3), Autosomal recessive |
| *HHAT* | 605743 | Nivelon-Nivelon-Mabille syndrome, 600092 (3), Autosomal recessive |
| *HIBCH* | 610690 | 3-hydroxyisobutryl-CoA hydrolase deficiency, 250620 (3), Autosomal recessive |
| *HIKESHI* | 614908 | Leukodystrophy, hypomyelinating, 13, 616881 (3), Autosomal recessive |
| *HINT1* | 601314 | Neuromyotonia and axonal neuropathy, autosomal recessive, 137200 (3), Autosomal recessive |
| *HIVEP2* | 143054 | Mental retardation, autosomal dominant 43, 616977 (3), Autosomal dominant |
| *HJV* | 608374 | Hemochromatosis, type 2A, 602390 (3), Autosomal recessive |
| *HK1* | 142600 | Retinitis pigmentosa 79, 617460 (3), Autosomal dominant; Neuropathy, hereditary motor and sensory, Russe type, 605285 (3), Autosomal recessive; Neurodevelopmental disorder with visual defects and brain anomalies, 618547 (3), Autosomal dominant; Hemolytic anemia due to hexokinase deficiency, 235700 (3), Autosomal recessive |
| *HLA-A* | 142800 | {Hypersensitivity syndrome, carbamazepine-induced, susceptibility to}, 608579 (3) |
| *HLA-B* | 142830 | {Synovitis, chronic, susceptibility to} (3); {Abacavir hypersensitivity, susceptibility to} (3); {Spondyloarthropathy, susceptibility to, 1}, 106300 (3), Multifactorial; {Stevens-Johnson syndrome, susceptibility to}, 608579 (3); {Drug-induced liver injury due to flucloxacillin} (3); {Toxic epidermal necrolysis, susceptibility to}, 608579 (3) |
| *HLA-C* | 142840 | {Psoriasis susceptibility 1}, 177900 (3), Multifactorial; {HIV-1 viremia, susceptibility to}, 609423 (3) |
| *HLA-DPB1* | 142858 | {Beryllium disease, chronic, susceptibility to} (3) |
| *HLA-DQA1* | 146880 | {Celiac disease, susceptibility to}, 212750 (3), Multifactorial, Autosomal recessive |
| *HLA-DQB1* | 604305 | {Celiac disease, susceptibility to}, 212750 (3), Multifactorial, Autosomal recessive; {Multiple sclerosis, susceptibility to, 1}, 126200 (3), Multifactorial; {Creutzfeldt-Jakob disease, variant, resistance to}, 123400 (3), Autosomal dominant |
| *HLA-DRB1* | 142857 | {Multiple sclerosis, susceptibility to, 1}, 126200 (3), Multifactorial; {Sarcoidosis, susceptibility to, 1}, 181000 (3), Autosomal dominant |
| *HLA-G* | 142871 | {Asthma, susceptibility to}, 600807 (2), Autosomal dominant |
| *HLCS* | 609018 | Holocarboxylase synthetase deficiency, 253270 (3), Autosomal recessive |
| *HMBS* | 609806 | Porphyria, acute intermittent, nonerythroid variant, 176000 (3), Autosomal dominant; Porphyria, acute intermittent, 176000 (3), Autosomal dominant |
| *HMCN1* | 608548 | {Macular degeneration, age-related, 1}, 603075 (3), Autosomal dominant |
| *HMGA1* | 600701 | {Type 2 diabetes mellitus, susceptibility to}, 125853 (3), Autosomal dominant |
| *HMGA2* | 600698 | Silver-Russell syndrome 5, 618908 (3), Autosomal dominant |
| *HMGB3* | 300193 | ?Microphthalmia, syndromic 13, 300915 (3), X-linked |
| *HMGCL* | 613898 | HMG-CoA lyase deficiency, 246450 (3), Autosomal recessive |
| *HMGCR* | 142910 | [Statins, attenuated cholesterol lowering by] (3); [Low density lipoprotein cholesterol level QTL 3] (3) |
| *HMGCS2* | 600234 | HMG-CoA synthase-2 deficiency, 605911 (3), Autosomal recessive |
| *HMMR* | 600936 | {Breast cancer, susceptibility to}, 114480 (3), Somatic mutation, Autosomal dominant |
| *HMOX1* | 141250 | Heme oxygenase-1 deficiency, 614034 (3); {Pulmonary disease, chronic obstructive, susceptibility to}, 606963 (3) |
| *HMX1* | 142992 | Oculoauricular syndrome, 612109 (3), Autosomal recessive |
| *HNF1A* | 142410 | Hepatic adenoma, somatic, 142330 (3); Diabetes mellitus, insulin-dependent, 20, 612520 (3); {Diabetes mellitus, noninsulin-dependent, 2}, 125853 (3), Autosomal dominant; MODY, type III, 600496 (3), Autosomal dominant; {Diabetes mellitus, insulin-dependent}, 222100 (3), Autosomal recessive; Renal cell carcinoma, 144700 (3) |
| *HNF1B* | 189907 | Type 2 diabetes mellitus, 125853 (3), Autosomal dominant; Renal cysts and diabetes syndrome, 137920 (3), Autosomal dominant; {Renal cell carcinoma}, 144700 (3) |
| *HNF4A* | 600281 | Fanconi renotubular syndrome 4, with maturity-onset diabetes of the young, 616026 (3), Autosomal dominant; {Diabetes mellitus, noninsulin-dependent}, 125853 (3), Autosomal dominant; MODY, type I, 125850 (3), Autosomal dominant |
| *HNMT* | 605238 | Mental retardation, autosomal recessive 51, 616739 (3), Autosomal recessive; {Asthma, susceptibility to}, 600807 (3), Autosomal dominant |
| *HNRNPA1* | 164017 | ?Inclusion body myopathy with early-onset Paget disease without frontotemporal dementia 3, 615424 (3), Autosomal dominant; Amyotrophic lateral sclerosis 20, 615426 (3), Autosomal dominant |
| *HNRNPA2B1* | 600124 | ?Inclusion body myopathy with early-onset Paget disease with or without frontotemporal dementia 2, 615422 (3) |
| *HNRNPDL* | 607137 | Muscular dystrophy, limb-girdle, autosomal dominant 3, 609115 (3), Autosomal dominant |
| *HNRNPH2* | 300610 | Mental retardation, X-linked, syndromic, Bain type, 300986 (3), X-linked dominant |
| *HNRNPK* | 600712 | Au-Kline syndrome, 616580 (3), Autosomal dominant |
| *HNRNPU* | 602869 | Developmental and epileptic encephalopathy 54, 617391 (3), Autosomal dominant |
| *HOGA1* | 613597 | Hyperoxaluria, primary, type III, 613616 (3), Autosomal recessive |
| *HOMER2* | 604799 | ?Deafness, autosomal dominant 68, 616707 (3), Autosomal dominant |
| *HOXA1* | 142955 | Bosley-Salih-Alorainy syndrome, 601536 (3), Autosomal recessive; Athabaskan brainstem dysgenesis syndrome, 601536 (3), Autosomal recessive |
| *HOXA11* | 142958 | Radioulnar synostosis with amegakaryocytic thrombocytopenia 1, 605432 (3), Autosomal dominant |
| *HOXA13* | 142959 | Hand-foot-uterus syndrome, 140000 (3), Autosomal dominant; ?Guttmacher syndrome, 176305 (3), Autosomal dominant |
| *HOXA2* | 604685 | Microtia with or without hearing impairment (AD), 612290 (3), Autosomal recessive, Autosomal dominant; ?Microtia, hearing impairment, and cleft palate (AR), 612290 (3), Autosomal recessive, Autosomal dominant |
| *HOXB1* | 142968 | Facial paresis, hereditary congenital, 3, 614744 (3), Autosomal recessive |
| *HOXB13* | 604607 | {Prostate cancer, hereditary, 9}, 610997 (3) |
| *HOXC13* | 142976 | Ectodermal dysplasia 9, hair/nail type, 614931 (3), Autosomal recessive |
| *HOXD10* | 142984 | Vertical talus, congenital, 192950 (3), Autosomal dominant; Charcot-Marie-Tooth disease, foot deformity of, 192950 (3), Autosomal dominant |
| *HOXD13* | 142989 | Syndactyly, type V, 186300 (3), Autosomal dominant; Synpolydactyly 1, 186000 (3), Autosomal dominant; Brachydactyly, type E, 113300 (3), Autosomal dominant; Brachydactyly, type D, 113200 (3), Autosomal dominant; ?Brachydactyly-syndactyly syndrome, 610713 (3) |
| *HP* | 140100 | [Anhaptoglobinemia], 614081 (3); [Hypohaptoglobinemia], 614081 (3) |
| *HPCA* | 142622 | Dystonia 2, torsion, autosomal recessive, 224500 (3), Autosomal recessive |
| *HPD* | 609695 | Hawkinsinuria, 140350 (3), Autosomal dominant; Tyrosinemia, type III, 276710 (3), Autosomal recessive |
| *HPDL* | 618994 | Neurodevelopmental disorder with progressive spasticity and brain white matter abnormalities, 619026 (3), Autosomal recessive; Spastic paraplegia 83, autosomal recessive, 619027 (3), Autosomal recessive |
| *HPGD* | 601688 | ?Digital clubbing, isolated congenital, 119900 (3), Autosomal recessive; Hypertrophic osteoarthropathy, primary, autosomal recessive 1, 259100 (3), Autosomal recessive; Cranioosteoarthropathy, 259100 (3), Autosomal recessive |
| *HPRT1* | 308000 | Hyperuricemia, HRPT-related, 300323 (3), X-linked recessive; Lesch-Nyhan syndrome, 300322 (3), X-linked recessive |
| *HPS1* | 604982 | Hermansky-Pudlak syndrome 1, 203300 (3), Autosomal recessive |
| *HPS3* | 606118 | Hermansky-Pudlak syndrome 3, 614072 (3), Autosomal recessive |
| *HPS4* | 606682 | Hermansky-Pudlak syndrome 4, 614073 (3), Autosomal recessive |
| *HPS5* | 607521 | Hermansky-Pudlak syndrome 5, 614074 (3), Autosomal recessive |
| *HPS6* | 607522 | Hermansky-Pudlak syndrome 6, 614075 (3), Autosomal recessive |
| *HPSE2* | 613469 | Urofacial syndrome 1, 236730 (3), Autosomal recessive |
| *HR* | 602302 | Atrichia with papular lesions, 209500 (3), Autosomal recessive; Alopecia universalis, 203655 (3), Autosomal recessive |
| *HRAS* | 190020 | Bladder cancer, somatic, 109800 (3); Thyroid carcinoma, follicular, somatic, 188470 (3); Congenital myopathy with excess of muscle spindles, 218040 (3), Autosomal dominant; Nevus sebaceous or woolly hair nevus, somatic, 162900 (3); Schimmelpenning-Feuerstein-Mims syndrome, somatic mosaic, 163200 (3); Spitz nevus or nevus spilus, somatic, 137550 (3); Costello syndrome, 218040 (3), Autosomal dominant |
| *HRG* | 142640 | Thrombophilia due to HRG deficiency, 613116 (3), Autosomal dominant |
| *HS2ST1* | 604844 | Neurofacioskeletal syndrome with or without renal agenesis, 619194 (3), Autosomal recessive |
| *HS3ST6* | 619210 | ?Angioedema, hereditary, 8, 619367 (3), Autosomal dominant |
| *HS6ST1* | 604846 | {Hypogonadotropic hypogonadism 15 with or without anosmia}, 614880 (3), Autosomal dominant |
| *HS6ST2* | 300545 | ?Paganini-Miozzo syndrome, 301025 (3), X-linked recessive |
| *HSD11B1* | 600713 | Cortisone reductase deficiency 2, 614662 (3), Autosomal dominant |
| *HSD11B2* | 614232 | Apparent mineralocorticoid excess, 218030 (3), Autosomal recessive |
| *HSD17B10* | 300256 | HSD10 mitochondrial disease, 300438 (3), X-linked dominant |
| *HSD17B3* | 605573 | Pseudohermaphroditism, male, with gynecomastia, 264300 (3), Autosomal recessive |
| *HSD17B4* | 601860 | D-bifunctional protein deficiency, 261515 (3), Autosomal recessive; Perrault syndrome 1, 233400 (3), Autosomal recessive |
| *HSD3B2* | 613890 | Adrenal hyperplasia, congenital, due to 3-beta-hydroxysteroid dehydrogenase 2 deficiency, 201810 (3), Autosomal recessive |
| *HSD3B7* | 607764 | Bile acid synthesis defect, congenital, 1, 607765 (3), Autosomal recessive |
| *HSF2BP* | 604554 | Premature ovarian failure 19, 619245 (3), Autosomal recessive |
| *HSF4* | 602438 | Cataract 5, multiple types, 116800 (3), Autosomal dominant |
| *HSPA9* | 600548 | Even-plus syndrome, 616854 (3), Autosomal recessive; Anemia, sideroblastic, 4, 182170 (3), Autosomal dominant |
| *HSPB1* | 602195 | Neuronopathy, distal hereditary motor, type IIB, 608634 (3), Autosomal dominant; Charcot-Marie-Tooth disease, axonal, type 2F, 606595 (3), Autosomal dominant |
| *HSPB3* | 604624 | ?Neuronopathy, distal hereditary motor, type IIC, 613376 (3), Autosomal dominant |
| *HSPB8* | 608014 | Neuronopathy, distal hereditary motor, type IIA, 158590 (3), Autosomal dominant; Charcot-Marie-Tooth disease, axonal, type 2L, 608673 (3), Autosomal dominant |
| *HSPD1* | 118190 | Spastic paraplegia 13, autosomal dominant, 605280 (3), Autosomal dominant; Leukodystrophy, hypomyelinating, 4, 612233 (3), Autosomal recessive |
| *HSPG2* | 142461 | Dyssegmental dysplasia, Silverman-Handmaker type, 224410 (3), Autosomal recessive; Schwartz-Jampel syndrome, type 1, 255800 (3), Autosomal recessive |
| *HTR1A* | 109760 | Periodic fever, menstrual cycle dependent, 614674 (3), Autosomal dominant |
| *HTR2A* | 182135 | {Alcohol dependence, susceptibility to}, 103780 (3), Multifactorial; {Anorexia nervosa, susceptibility to}, 606788 (3); {Major depressive disorder, response to citalopram therapy in}, 608516 (3); {Seasonal affective disorder, susceptibility to}, 608516 (3); {Obsessive-compulsive disorder, susceptibility to}, 164230 (3), Autosomal dominant; {Schizophrenia, susceptibility to}, 181500 (3), Autosomal dominant |
| *HTRA1* | 602194 | {Macular degeneration, age-related, neovascular type}, 610149 (3); {Macular degeneration, age-related, 7}, 610149 (3); CARASIL syndrome, 600142 (3), Autosomal recessive; Cerebral arteriopathy, autosomal dominant, with subcortical infarcts and leukoencephalopathy, type 2, 616779 (3), Autosomal dominant |
| *HTRA2* | 606441 | {Parkinson disease 13}, 610297 (3); 3-methylglutaconic aciduria, type VIII, 617248 (3), Autosomal recessive |
| *HTT* | 613004 | Lopes-Maciel-Rodan syndrome, 617435 (3), Autosomal recessive; Huntington disease, 143100 (3), Autosomal dominant |
| *HUWE1* | 300697 | Mental retardation, X-linked syndromic, Turner type, 309590 (3), X-linked |
| *HYAL1* | 607071 | ?Mucopolysaccharidosis type IX, 601492 (3), Autosomal recessive |
| *HYDIN* | 610812 | Ciliary dyskinesia, primary, 5, 608647 (3), Autosomal recessive |
| *HYLS1* | 610693 | Hydrolethalus syndrome, 236680 (3), Autosomal recessive |
| *HYOU1* | 601746 | ?Immunodeficiency 59 and hypoglycemia, 233600 (3), Autosomal recessive |
| *IARS1 (IARS)* | 600709 | Growth retardation, impaired intellectual development, hypotonia, and hepatopathy, 617093 (3), Autosomal recessive |
| *IARS2* | 612801 | ?Cataracts, growth hormone deficiency, sensory neuropathy, sensorineural hearing loss, and skeletal dysplasia, 616007 (3), Autosomal recessive |
| *IBA57* | 615316 | Multiple mitochondrial dysfunctions syndrome 3, 615330 (3), Autosomal recessive; ?Spastic paraplegia 74, autosomal recessive, 616451 (3), Autosomal recessive |
| *ICAM1* | 147840 | {Malaria, cerebral, susceptibility to}, 611162 (3) |
| *ICAM4* | 614088 | [Blood group, Landsteiner-Wiener], 111250 (3) |
| *ICOS* | 604558 | Immunodeficiency, common variable, 1, 607594 (3), Autosomal recessive |
| *IDH1* | 147700 | {Glioma, susceptibility to, somatic}, 137800 (3) |
| *IDH2* | 147650 | D-2-hydroxyglutaric aciduria 2, 613657 (3) |
| *IDH3A* | 601149 | Retinitis pigmentosa 90, 619007 (3), Autosomal recessive |
| *IDH3B* | 604526 | Retinitis pigmentosa 46, 612572 (3), Autosomal recessive |
| *IDS* | 300823 | Mucopolysaccharidosis II, 309900 (3), X-linked recessive |
| *IDUA* | 252800 | Mucopolysaccharidosis Is, 607016 (3), Autosomal recessive; Mucopolysaccharidosis Ih/s, 607015 (3), Autosomal recessive; Mucopolysaccharidosis Ih, 607014 (3), Autosomal recessive |
| *IER3IP1* | 609382 | Microcephaly, epilepsy, and diabetes syndrome, 614231 (3), Autosomal recessive |
| *IFIH1* | 606951 | Aicardi-Goutieres syndrome 7, 615846 (3), Autosomal dominant; Singleton-Merten syndrome 1, 182250 (3), Autosomal dominant |
| *IFITM3* | 605579 | {Influenza, severe, susceptibility to}, 614680 (3) |
| *IFITM5* | 614757 | Osteogenesis imperfecta, type V, 610967 (3), Autosomal dominant |
| *IFNA1* | 147660 | Interferon, alpha, deficiency (1) |
| *IFNAR2* | 602376 | {Hepatitis B virus, susceptibility to}, 610424 (3); ?Immunodeficiency 45, 616669 (3), Autosomal recessive |
| *IFNG* | 147570 | {Hepatitis C virus, response to therapy of}, 609532 (3); {TSC2 angiomyolipomas, renal, modifier of}, 613254 (3), Autosomal dominant; {Aplastic anemia}, 609135 (3); ?Immunodeficiency 69, mycobacteriosis, 618963 (3), Autosomal recessive; {Tuberculosis, protection against}, 607948 (3); {AIDS, rapid progression to}, 609423 (3) |
| *IFNGR1* | 107470 | {H. pylori infection, susceptibility to}, 600263 (3); Immunodeficiency 27A, mycobacteriosis, AR, 209950 (3), Autosomal recessive; Immunodeficiency 27B, mycobacteriosis, AD, 615978 (3), Autosomal dominant; {Tuberculosis infection, protection against}, 607948 (3); {Tuberculosis, susceptibility to}, 607948 (3); {Hepatitis B virus infection, susceptibility to}, 610424 (3) |
| *IFNGR2* | 147569 | Immunodeficiency 28, mycobacteriosis, 614889 (3), Autosomal recessive |
| *IFNL3* | 607402 | {Hepatitis C virus infection, response to therapy of}, 609532 (3) |
| *IFT122* | 606045 | Cranioectodermal dysplasia 1, 218330 (3), Autosomal recessive |
| *IFT140* | 614620 | Short-rib thoracic dysplasia 9 with or without polydactyly, 266920 (3), Autosomal recessive; Retinitis pigmentosa 80, 617781 (3), Autosomal recessive |
| *IFT172* | 607386 | Retinitis pigmentosa 71, 616394 (3), Autosomal recessive; Short-rib thoracic dysplasia 10 with or without polydactyly, 615630 (3), Autosomal recessive |
| *IFT27* | 615870 | Bardet-Biedl syndrome 19, 615996 (3), Autosomal recessive |
| *IFT43* | 614068 | ?Cranioectodermal dysplasia 3, 614099 (3), Autosomal recessive; ?Retinitis pigmentosa 81, 617871 (3), Autosomal recessive; Short-rib thoracic dysplasia 18 with polydactyly, 617866 (3), Autosomal recessive |
| *IFT52* | 617094 | Short-rib thoracic dysplasia 16 with or without polydactyly, 617102 (3), Autosomal recessive |
| *IFT57* | 606621 | ?Orofaciodigital syndrome XVIII, 617927 (3), Autosomal recessive |
| *IFT74* | 608040 | ?Bardet-Biedl syndrome 20, 617119 (3), Autosomal recessive |
| *IFT80* | 611177 | Short-rib thoracic dysplasia 2 with or without polydactyly, 611263 (3), Autosomal recessive |
| *IFT81* | 605489 | Short-rib thoracic dysplasia 19 with or without polydactyly, 617895 (3), Autosomal recessive |
| *IGBP1* | 300139 | Corpus callosum, agenesis of, with mental retardation, ocular coloboma and micrognathia, 300472 (3), X-linked recessive |
| *IGF1* | 147440 | Growth retardation with deafness and mental retardation due to IGF1 deficiency, 608747 (3), Autosomal recessive |
| *IGF1R* | 147370 | Insulin-like growth factor I, resistance to, 270450 (3), Autosomal recessive, Autosomal dominant |
| *IGF2* | 147470 | Silver-Russell syndrome 3, 616489 (3), Autosomal dominant |
| *IGF2BP2* | 608289 | {Diabetes mellitus, noninsulin-dependent, susceptibility to}, 125853 (3), Autosomal dominant |
| *IGF2R* | 147280 | Hepatocellular carcinoma, somatic, 114550 (3) |
| *IGFALS* | 601489 | Acid-labile subunit, deficiency of, 615961 (3), Autosomal recessive |
| *IGFBP7* | 602867 | Retinal arterial macroaneurysm with supravalvular pulmonic stenosis, 614224 (3), Autosomal recessive |
| *IGHG2* | 147110 | IgG2 deficiency, selective (3) |
| *IGHM* | 147020 | Agammaglobulinemia 1, 601495 (3), Autosomal recessive |
| *IGHMBP2* | 600502 | Neuronopathy, distal hereditary motor, type VI, 604320 (3), Autosomal recessive; Charcot-Marie-Tooth disease, axonal, type 2S, 616155 (3), Autosomal recessive |
| *IGKC* | 147200 | Kappa light chain deficiency, 614102 (3), Autosomal recessive |
| *IGLL1* | 146770 | Agammaglobulinemia 2, 613500 (3), Autosomal recessive |
| *IGSF1* | 300137 | Hypothyroidism, central, and testicular enlargement, 300888 (3), X-linked recessive |
| *IGSF3* | 603491 | ?Lacrimal duct defect, 149700 (3), Autosomal recessive |
| *IHH* | 600726 | Acrocapitofemoral dysplasia, 607778 (3), Autosomal recessive; Brachydactyly, type A1, 112500 (3), Autosomal dominant |
| *IKBKB* | 603258 | Immunodeficiency 15B, 615592 (3), Autosomal recessive; Immunodeficiency 15A, 618204 (3), Autosomal dominant |
| *IKBKG* | 300248 | Incontinentia pigmenti, 308300 (3), X-linked dominant; Ectodermal dysplasia and immunodeficiency 1, 300291 (3), X-linked recessive; Immunodeficiency 33, 300636 (3), X-linked recessive |
| *IKZF1* | 603023 | Immunodeficiency, common variable, 13, 616873 (3), Autosomal dominant |
| *IKZF5* | 606238 | Thrombocytopenia, autosomal dominant, 7, 619130 (3) |
| *IL10* | 124092 | {Rheumatoid arthritis, progression of}, 180300 (3); {Graft-versus-host disease, protection against}, 614395 (3); {HIV-1, susceptibility to}, 609423 (3) |
| *IL10RA* | 146933 | Inflammatory bowel disease 28, early onset, autosomal recessive, 613148 (3), Autosomal recessive |
| *IL10RB* | 123889 | {Hepatitis B virus, susceptibility to}, 610424 (3); Inflammatory bowel disease 25, early onset, autosomal recessive, 612567 (3), Autosomal recessive |
| *IL11RA* | 600939 | Craniosynostosis and dental anomalies, 614188 (3), Autosomal recessive |
| *IL12B* | 161561 | Immunodeficiency 29, mycobacteriosis, 614890 (3), Autosomal recessive |
| *IL12RB1* | 601604 | Immunodeficiency 30, 614891 (3), Autosomal recessive |
| *IL13* | 147683 | {Asthma, susceptibility to}, 600807 (3), Autosomal dominant; {Allergic rhinitis, susceptibility to}, 607154 (3) |
| *IL17F* | 606496 | ?Candidiasis, familial, 6, autosomal dominant, 613956 (3) |
| *IL17RA* | 605461 | Immunodeficiency 51, 613953 (3), Autosomal recessive |
| *IL17RC* | 610925 | Candidiasis, familial, 9, 616445 (3), Autosomal recessive |
| *IL17RD* | 606807 | Hypogonadotropic hypogonadism 18 with or without anosmia, 615267 (3), Digenic dominant, Autosomal recessive, Autosomal dominant |
| *IL18BP* | 604113 | {?Hepatitis, fulminant viral, susceptibility to}, 618549 (3), Autosomal recessive |
| *IL1B* | 147720 | {Gastric cancer risk after H. pylori infection}, 137215 (3), Autosomal dominant |
| *IL1RAPL1* | 300206 | Mental retardation, X-linked 21/34, 300143 (3), X-linked recessive |
| *IL1RN* | 147679 | {Gastric cancer risk after H. pylori infection}, 137215 (3), Autosomal dominant; {Microvascular complications of diabetes 4}, 612628 (3); Interleukin 1 receptor antagonist deficiency, 612852 (3), Autosomal recessive |
| *IL21* | 605384 | ?Immunodeficiency, common variable, 11, 615767 (3), Autosomal recessive |
| *IL21R* | 605383 | [IgE, elevated level of], 147050 (3), Autosomal dominant; Immunodeficiency 56, 615207 (3), Autosomal recessive |
| *IL23R* | 607562 | {Inflammatory bowel disease 17, protection against}, 612261 (3); {Psoriasis, protection against}, 605606 (3) |
| *IL2RA* | 147730 | Immunodeficiency 41 with lymphoproliferation and autoimmunity, 606367 (3), Autosomal recessive; {Diabetes, mellitus, insulin-dependent, susceptibility to, 10}, 601942 (3) |
| *IL2RB* | 146710 | Immunodeficiency 63 with lymphoproliferation and autoimmunity, 618495 (3), Autosomal recessive |
| *IL2RG* | 308380 | Combined immunodeficiency, X-linked, moderate, 312863 (3), X-linked recessive; Severe combined immunodeficiency, X-linked, 300400 (3), X-linked recessive |
| *IL31RA* | 609510 | ?Amyloidosis, primary localized cutaneous, 2, 613955 (3), Autosomal dominant |
| *IL36RN* | 605507 | Psoriasis 14, pustular, 614204 (3), Autosomal recessive |
| *IL37* | 605510 | ?Inflammatory bowel disease (infantile ulcerative colitis) 31, 619398 (3), Autosomal recessive |
| *IL4R* | 147781 | {AIDS, slow progression to}, 609423 (3); {Atopy, susceptibility to}, 147050 (3), Autosomal dominant |
| *IL6* | 147620 | {Type 2 diabetes mellitus}, 125853 (3), Autosomal dominant; {Rheumatoid arthritis, systemic juvenile}, 604302 (3); {Intracranial hemorrhage in brain cerebrovascular malformations, susceptibility to}, 108010 (3), Somatic mutation; {Type 1 diabetes mellitus}, 222100 (3), Autosomal recessive; {Crohn disease-associated growth failure}, 266600 (3), Multifactorial; {Kaposi sarcoma, susceptibility to}, 148000 (3), Autosomal dominant |
| *IL6R* | 147880 | [Interleukin 6, serum level of, QTL], 614752 (3); Hyper-IgE recurrent infection syndrome 5, autosomal recessive, 618944 (3), Autosomal recessive; [Interleukin-6 receptor, soluble, serum level of, QTL], 614689 (3) |
| *IL6ST* | 600694 | Hyper-IgE recurrent infection syndrome 4, autosomal recessive, 618523 (3), Autosomal recessive |
| *IL7* | 146660 | {?Epidermodysplasia verruciformis, susceptibility to, 5}, 618309 (3), Autosomal recessive |
| *IL7R* | 146661 | Severe combined immunodeficiency, T-cell negative, B-cell/natural killer cell-positive type, 608971 (3), Autosomal recessive |
| *ILDR1* | 609739 | Deafness, autosomal recessive 42, 609646 (3), Autosomal recessive |
| *IMPA1* | 602064 | Mental retardation, autosomal recessive 59, 617323 (3), Autosomal recessive |
| *IMPDH1* | 146690 | Retinitis pigmentosa 10, 180105 (3), Autosomal dominant; Leber congenital amaurosis 11, 613837 (3), Autosomal dominant |
| *IMPDH2* | 146691 | [IMPDH2 enzyme activity, variation in], 617995 (3) |
| *IMPG1* | 602870 | Macular dystrophy, vitelliform, 4, 616151 (3), Autosomal dominant |
| *IMPG2* | 607056 | Retinitis pigmentosa 56, 613581 (3), Autosomal recessive; Macular dystrophy, vitelliform, 5, 616152 (3), Autosomal dominant |
| *INAVA* | 618051 | {Inflammatory bowel disease 29}, 618077 (3), Autosomal dominant |
| *INF2* | 610982 | Glomerulosclerosis, focal segmental, 5, 613237 (3); Charcot-Marie-Tooth disease, dominant intermediate E, 614455 (3), Autosomal dominant |
| *ING1* | 601566 | Squamous cell carcinoma, head and neck, somatic, 275355 (3) |
| *INPP5E* | 613037 | Joubert syndrome 1, 213300 (3), Autosomal recessive; Mental retardation, truncal obesity, retinal dystrophy, and micropenis, 610156 (3), Autosomal recessive |
| *INPP5K* | 607875 | Muscular dystrophy, congenital, with cataracts and intellectual disability, 617404 (3), Autosomal recessive |
| *INPPL1* | 600829 | Opsismodysplasia, 258480 (3), Autosomal recessive |
| *INS* | 176730 | Diabetes mellitus, insulin-dependent, 2, 125852 (3), Autosomal dominant; Maturity-onset diabetes of the young, type 10, 613370 (3), Autosomal dominant; Hyperproinsulinemia, 616214 (3), Autosomal dominant; Diabetes mellitus, permanent neonatal 4, 618858 (3), Autosomal recessive, Autosomal dominant |
| *INSL3* | 146738 | Cryptorchidism, 219050 (3), Autosomal dominant |
| *INSR* | 147670 | Rabson-Mendenhall syndrome, 262190 (3), Autosomal recessive; Leprechaunism, 246200 (3), Autosomal recessive; Diabetes mellitus, insulin-resistant, with acanthosis nigricans, 610549 (3); Hyperinsulinemic hypoglycemia, familial, 5, 609968 (3), Autosomal dominant |
| *INTS1* | 611345 | Neurodevelopmental disorder with cataracts, poor growth, and dysmorphic facies, 618571 (3), Autosomal recessive |
| *INTS8* | 611351 | ?Neurodevelopmental disorder with cerebellar hypoplasia and spasticity, 618572 (3), Autosomal recessive |
| *INTU* | 610621 | ?Orofaciodigital syndrome XVII, 617926 (3), Autosomal recessive; ?Short-rib thoracic dysplasia 20 with polydactyly, 617925 (3), Autosomal recessive |
| *INVS* | 243305 | Nephronophthisis 2, infantile, 602088 (3), Autosomal recessive |
| *IQCB1* | 609237 | Senior-Loken syndrome 5, 609254 (3), Autosomal recessive |
| *IQCE* | 617631 | Polydactyly, postaxial, type A7, 617642 (3), Autosomal recessive |
| *IQSEC1* | 610166 | Intellectual developmental disorder with short stature and behavioral abnormalities, 618687 (3), Autosomal recessive |
| *IQSEC2* | 300522 | Mental retardation, X-linked 1/78, 309530 (3), X-linked dominant |
| *IRAK3* | 604459 | {Asthma susceptibility 5}, 611064 (3) |
| *IRAK4* | 606883 | Immunodeficiency 67, 607676 (3), Autosomal recessive |
| *IREB2* | 147582 | Neurodegeneration, early-onset, with choreoathetoid movements and microcytic anemia, 618451 (3), Autosomal recessive |
| *IRF1* | 147575 | Nonsmall cell lung cancer, somatic, 211980 (3); Myelodysplastic syndrome, preleukemic (3); Gastric cancer, somatic, 613659 (3); Myelogenous leukemia, acute (3) |
| *IRF2BP2* | 615332 | ?Immunodeficiency, common variable, 14, 617765 (3), Autosomal dominant |
| *IRF2BPL* | 611720 | Neurodevelopmental disorder with regression, abnormal movements, loss of speech, and seizures, 618088 (3), Autosomal dominant |
| *IRF3* | 603734 | {Encephalopathy, acute, infection-induced (herpes-specific), susceptibility to, 7}, 616532 (3), Autosomal dominant |
| *IRF4* | 601900 | [Skin/hair/eye pigmentation, variation in, 8], 611724 (3) |
| *IRF5* | 607218 | {Inflammatory bowel disease 14}, 612245 (3); {Systemic lupus erythematosus, susceptibility to, 10}, 612251 (3) |
| *IRF6* | 607199 | {Orofacial cleft 6}, 608864 (3), Autosomal dominant; Popliteal pterygium syndrome 1, 119500 (3), Autosomal dominant; van der Woude syndrome, 119300 (3), Autosomal dominant |
| *IRF7* | 605047 | ?Immunodeficiency 39, 616345 (3), Autosomal recessive |
| *IRF8* | 601565 | Immunodeficiency 32A, mycobacteriosis, autosomal dominant, 614893 (3), Autosomal dominant; Immunodeficiency 32B, monocyte and dendritic cell deficiency, autosomal recessive, 226990 (3), Autosomal recessive |
| *IRF9* | 147574 | Immunodeficiency 65, susceptibility to viral infections, 618648 (3), Autosomal recessive |
| *IRGM* | 608212 | {Mycobacterium tuberculosis, protection against}, 607948 (3); {Inflammatory bowel disease (Crohn disease) 19}, 612278 (3) |
| *IRS1* | 147545 | {Type 2 diabetes mellitus, susceptibility to}, 125853 (3), Autosomal dominant; {Coronary artery disease, susceptibility to} (3) |
| *IRS2* | 600797 | {Diabetes mellitus, noninsulin-dependent}, 125853 (3), Autosomal dominant |
| *IRS4* | 300904 | Hypothyroidism, congenital, nongoitrous, 9, 301035 (3), X-linked recessive |
| *IRX5* | 606195 | Hamamy syndrome, 611174 (3), Autosomal recessive |
| *ISCA1* | 611006 | Multiple mitochondrial dysfunctions syndrome 5, 617613 (3), Autosomal recessive |
| *ISCA2* | 615317 | Multiple mitochondrial dysfunctions syndrome 4, 616370 (3), Autosomal recessive |
| *ISCU* | 611911 | Myopathy with lactic acidosis, hereditary, 255125 (3), Autosomal recessive |
| *ISG15* | 147571 | Immunodeficiency 38, 616126 (3), Autosomal recessive |
| *ITCH* | 606409 | Autoimmune disease, multisystem, with facial dysmorphism, 613385 (3), Autosomal recessive |
| *ITGA2B* | 607759 | Thrombocytopenia, neonatal alloimmune, BAK antigen related (3); Glanzmann thrombasthenia 1, 273800 (3), Autosomal recessive; Bleeding disorder, platelet-type, 16, autosomal dominant, 187800 (3), Autosomal dominant |
| *ITGA3* | 605025 | Interstitial lung disease, nephrotic syndrome, and epidermolysis bullosa, congenital, 614748 (3), Autosomal recessive |
| *ITGA6* | 147556 | Epidermolysis bullosa, junctional, with pyloric stenosis, 226730 (3), Autosomal recessive |
| *ITGA7* | 600536 | Muscular dystrophy, congenital, due to ITGA7 deficiency, 613204 (3), Autosomal recessive |
| *ITGA8* | 604063 | Renal hypodysplasia/aplasia 1, 191830 (3), Autosomal recessive |
| *ITGB2* | 600065 | Leukocyte adhesion deficiency, 116920 (3), Autosomal recessive |
| *ITGB3* | 173470 | Bleeding disorder, platelet-type, 24, autosomal dominant, 619271 (3); Thrombocytopenia, neonatal alloimmune (3); Purpura, posttransfusion (3); {Myocardial infarction, susceptibility to}, 608446 (3); Glanzmann thrombasthenia 2, 619267 (3) |
| *ITGB4* | 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 |
| *ITGB6* | 147558 | Amelogenesis imperfecta, type IH, 616221 (3), Autosomal recessive |
| *ITK* | 186973 | Lymphoproliferative syndrome 1, 613011 (3), Autosomal recessive |
| *ITM2B* | 603904 | ?Retinal dystrophy with inner retinal dysfunction and ganglion cell abnormalities, 616079 (3), Autosomal dominant; Dementia, familial British, 176500 (3), Autosomal dominant; Dementia, familial Danish, 117300 (3), Autosomal dominant |
| *ITPA* | 147520 | [Inosine triphosphatase deficiency], 613850 (3); Developmental and epileptic encephalopathy 35, 616647 (3), Autosomal recessive |
| *ITPR1* | 147265 | Gillespie syndrome, 206700 (3), Autosomal recessive, Autosomal dominant; Spinocerebellar ataxia 29, congenital nonprogressive, 117360 (3), Autosomal dominant; Spinocerebellar ataxia 15, 606658 (3), Autosomal dominant |
| *ITPR2* | 600144 | ?Anhidrosis, isolated, with normal sweat glands, 106190 (3), Autosomal recessive |
| *ITPR3* | 147267 | {Diabetes, type 1, susceptibility to}, 222100 (2), Autosomal recessive |
| *IVD* | 607036 | Isovaleric acidemia, 243500 (3), Autosomal recessive |
| *IVNS1ABP* | 609209 | Immunodeficiency 70, 618969 (3), Autosomal dominant |
| *IYD* | 612025 | Thyroid dyshormonogenesis 4, 274800 (3), Autosomal recessive |
| *JAG1* | 601920 | ?Deafness, congenital heart defects, and posterior embryotoxon, 617992 (3), Autosomal dominant; Alagille syndrome 1, 118450 (3), Autosomal dominant; Tetralogy of Fallot, 187500 (3), Autosomal dominant |
| *JAGN1* | 616012 | Neutropenia, severe congenital, 6, autosomal recessive, 616022 (3), Autosomal recessive |
| *JAK1* | 147795 | Autoinflammation, immune dysregulation, and eosinophilia, 618999 (3), Autosomal dominant |
| *JAK2* | 147796 | {Budd-Chiari syndrome, somatic}, 600880 (3); Myelofibrosis, somatic, 254450 (3); Erythrocytosis, somatic, 133100 (3); Leukemia, acute myeloid, somatic, 601626 (3); Thrombocythemia 3, 614521 (3), Somatic mutation, Autosomal dominant; Polycythemia vera, somatic, 263300 (3) |
| *JAK3* | 600173 | SCID, autosomal recessive, T-negative/B-positive type, 600802 (3), Autosomal recessive |
| *JAM2* | 606870 | Basal ganglia calcification, idiopathic, 8, autosomal recessive, 618824 (3), Autosomal recessive |
| *JAM3* | 606871 | Hemorrhagic destruction of the brain, subependymal calcification, and cataracts, 613730 (3), Autosomal recessive |
| *JPH1* | 605266 | ?Charcot-Marie-Tooth disease, axonal, autosomal dominant, type 2K, 607831 (3), Autosomal recessive, Autosomal dominant |
| *JPH2* | 605267 | Cardiomyopathy, hypertrophic, 17, 613873 (3), Autosomal dominant |
| *JPH3* | 605268 | Huntington disease-like 2, 606438 (3), Autosomal dominant |
| *JUP* | 173325 | Naxos disease, 601214 (3), Autosomal recessive; ?Arrhythmogenic right ventricular dysplasia 12, 611528 (3), Autosomal dominant |
| *KANK1* | 607704 | Cerebral palsy, spastic quadriplegic, 2, 612900 (3) |
| *KANK2* | 614610 | Nephrotic syndrome, type 16, 617783 (3), Autosomal recessive; Palmoplantar keratoderma and woolly hair, 616099 (3), Autosomal recessive |
| *KANSL1* | 612452 | Koolen-De Vries syndrome, 610443 (3), Autosomal dominant |
| *KARS1 (KARS)* | 601421 | Deafness, autosomal recessive 89, 613916 (3), Autosomal recessive; Leukoencephalopathy, progressive, infantile-onset, with or without deafness, 619147 (3), Autosomal recessive; ?Charcot-Marie-Tooth disease, recessive intermediate, B, 613641 (3), Autosomal recessive; Deafness, congenital, and adult-onset progressive leukoencephalopathy, 619196 (3), Autosomal recessive |
| *KAT5* | 601409 | Neurodevelopmental disorder wtih dysmorphic facies, sleep disturbance, and brain abnormalities, 619103 (3) |
| *KAT6A* | 601408 | Arboleda-Tham syndrome, 616268 (3), Autosomal dominant |
| *KAT6B* | 605880 | SBBYSS syndrome, 603736 (3), Autosomal dominant; Genitopatellar syndrome, 606170 (3), Autosomal dominant |
| *KAT8* | 609912 | Li-Ghorgani-Weisz-Hubshman syndrome, 618974 (3), Autosomal dominant |
| *KATNB1* | 602703 | Lissencephaly 6, with microcephaly, 616212 (3), Autosomal recessive |
| *KATNIP (KIAA0556)* | 616650 | Joubert syndrome 26, 616784 (3), Autosomal recessive |
| *KBTBD13* | 613727 | Nemaline myopathy 6, autosomal dominant, 609273 (3), Autosomal dominant |
| *KCNA1* | 176260 | Episodic ataxia/myokymia syndrome, 160120 (3), Autosomal dominant |
| *KCNA2* | 176262 | Developmental and epileptic encephalopathy 32, 616366 (3), Autosomal dominant |
| *KCNA4* | 176266 | Microcephaly, cataracts, impaired intellectual development, and dystonia with abnormal striatum, 618284 (3), Autosomal recessive |
| *KCNA5* | 176267 | Atrial fibrillation, familial, 7, 612240 (3), Autosomal dominant |
| *KCNB1* | 600397 | Developmental and epileptic encephalopathy 26, 616056 (3), Autosomal dominant |
| *KCNC1* | 176258 | Epilepsy, progressive myoclonic 7, 616187 (3), Autosomal dominant |
| *KCNC3* | 176264 | Spinocerebellar ataxia 13, 605259 (3), Autosomal dominant |
| *KCND3* | 605411 | Spinocerebellar ataxia 19, 607346 (3), Autosomal dominant; Brugada syndrome 9, 616399 (3), Autosomal dominant |
| *KCNE1* | 176261 | Jervell and Lange-Nielsen syndrome 2, 612347 (3), Autosomal recessive; Long QT syndrome 5, 613695 (3), Autosomal dominant |
| *KCNE2* | 603796 | Long QT syndrome 6, 613693 (3), Autosomal dominant; Atrial fibrillation, familial, 4, 611493 (3) |
| *KCNE3* | 604433 | ?Brugada syndrome 6, 613119 (3) |
| *KCNH1* | 603305 | Zimmermann-Laband syndrome 1, 135500 (3), Autosomal dominant; Temple-Baraitser syndrome, 611816 (3), Autosomal dominant |
| *KCNH2* | 152427 | Short QT syndrome 1, 609620 (3); Long QT syndrome 2, 613688 (3), Autosomal dominant; {Long QT syndrome 2, acquired, susceptibility to}, 613688 (3), Autosomal dominant |
| *KCNJ1* | 600359 | Bartter syndrome, type 2, 241200 (3), Autosomal recessive |
| *KCNJ10* | 602208 | Enlarged vestibular aqueduct, digenic, 600791 (3), Autosomal recessive; SESAME syndrome, 612780 (3), Autosomal recessive |
| *KCNJ11* | 600937 | Diabetes, permanent neonatal 2, with or without neurologic features, 618856 (3), Autosomal dominant; {Diabetes mellitus, type 2, susceptibility to}, 125853 (3), Autosomal dominant; Maturity-onset diabetes of the young, type 13, 616329 (3), Autosomal dominant; Diabetes mellitus, transient neonatal 3, 610582 (3), Autosomal dominant; Hyperinsulinemic hypoglycemia, familial, 2, 601820 (3), Autosomal recessive, Autosomal dominant |
| *KCNJ13* | 603208 | Snowflake vitreoretinal degeneration, 193230 (3), Autosomal dominant; Leber congenital amaurosis 16, 614186 (3), Autosomal recessive |
| *KCNJ16* | 605722 | Hypokalemic tubulopathy and deafness, 619406 (3), Autosomal recessive |
| *KCNJ18* | 613236 | {Thyrotoxic periodic paralysis, susceptibility to, 2}, 613239 (3), Autosomal dominant |
| *KCNJ2* | 600681 | Atrial fibrillation, familial, 9, 613980 (3), Autosomal dominant; Andersen syndrome, 170390 (3), Autosomal dominant; Short QT syndrome 3, 609622 (3) |
| *KCNJ5* | 600734 | Long QT syndrome 13, 613485 (3), Autosomal dominant; Hyperaldosteronism, familial, type III, 613677 (3), Autosomal dominant |
| *KCNJ6* | 600877 | Keppen-Lubinsky syndrome, 614098 (3), Autosomal dominant |
| *KCNK18* | 613655 | {Migraine, with or without aura, susceptibility to, 13}, 613656 (3), Autosomal dominant |
| *KCNK3* | 603220 | Pulmonary hypertension, primary, 4, 615344 (3), Autosomal dominant |
| *KCNK4* | 605720 | Facial dysmorphism, hypertrichosis, epilepsy, intellectual/developmental delay, and gingival overgrowth syndrome, 618381 (3), Autosomal dominant |
| *KCNK9* | 605874 | Birk-Barel syndrome, 612292 (3) |
| *KCNMA1* | 600150 | {Epilepsy, idiopathic generalized, susceptibility to, 16}, 618596 (3), Autosomal dominant; Paroxysmal nonkinesigenic dyskinesia, 3, with or without generalized epilepsy, 609446 (3), Autosomal dominant; Cerebellar atrophy, developmental delay, and seizures, 617643 (3), Autosomal recessive; Liang-Wang syndrome, 618729 (3), Autosomal dominant |
| *KCNMB1* | 603951 | {Hypertension, diastolic, resistance to}, 608622 (3), Autosomal dominant |
| *KCNN3* | 602983 | Zimmermann-Laband syndrome 3, 618658 (3), Autosomal dominant |
| *KCNN4* | 602754 | Dehydrated hereditary stomatocytosis 2, 616689 (3), Autosomal dominant |
| *KCNQ1* | 607542 | Short QT syndrome 2, 609621 (3), Autosomal dominant; Atrial fibrillation, familial, 3, 607554 (3), Autosomal dominant; Long QT syndrome 1, 192500 (3), Autosomal dominant; {Long QT syndrome 1, acquired, susceptibility to}, 192500 (3), Autosomal dominant; Jervell and Lange-Nielsen syndrome, 220400 (3), Autosomal recessive |
| *KCNQ2* | 602235 | Developmental and epileptic encephalopathy 7, 613720 (3), Autosomal dominant; Seizures, benign neonatal, 1, 121200 (3), Autosomal dominant; Myokymia, 121200 (3), Autosomal dominant |
| *KCNQ3* | 602232 | Seizures, benign neonatal, 2, 121201 (3), Autosomal dominant |
| *KCNQ4* | 603537 | Deafness, autosomal dominant 2A, 600101 (3), Autosomal dominant |
| *KCNQ5* | 607357 | Mental retardation, autosomal dominant 46, 617601 (3), Autosomal dominant |
| *KCNT1* | 608167 | Developmental and epileptic encephalopathy 14, 614959 (3), Autosomal dominant; Epilepsy nocturnal frontal lobe, 5, 615005 (3), Autosomal dominant |
| *KCNT2* | 610044 | Developmental and epileptic encephalopathy 57, 617771 (3), Autosomal dominant |
| *KCNV2* | 607604 | Retinal cone dystrophy 3B, 610356 (3), Autosomal recessive |
| *KCTD1* | 613420 | Scalp-ear-nipple syndrome, 181270 (3), Autosomal dominant |
| *KCTD17* | 616386 | Dystonia 26, myoclonic, 616398 (3), Autosomal dominant |
| *KCTD7* | 611725 | Epilepsy, progressive myoclonic 3, with or without intracellular inclusions, 611726 (3), Autosomal recessive |
| *KDELR2* | 609024 | Osteogenesis imperfecta 21, 619131 (3), Autosomal recessive |
| *KDF1* | 616758 | ?Ectodermal dysplasia 12, hypohidrotic/hair/tooth/nail type, 617337 (3), Autosomal dominant |
| *KDM1A* | 609132 | Cleft palate, psychomotor retardation, and distinctive facial features, 616728 (3), Autosomal dominant |
| *KDM3B* | 609373 | Diets-Jongmans syndrome, 618846 (3), Autosomal dominant |
| *KDM4B* | 609765 | Intellectual developmental disorder, autosomal dominant 65, 619320 (3), Autosomal dominant |
| *KDM5B* | 605393 | Mental retardation, autosomal recessive 65, 618109 (3), Autosomal recessive |
| *KDM5C* | 314690 | Mental retardation, X-linked, syndromic, Claes-Jensen type, 300534 (3), X-linked recessive |
| *KDM6A* | 300128 | Kabuki syndrome 2, 300867 (3), X-linked dominant |
| *KDM6B* | 611577 | Neurodevelopmental disorder with coarse facies and mild distal skeletal abnormalities, 618505 (3), Autosomal dominant |
| *KDR* | 191306 | {Hemangioma, capillary infantile, susceptibility to}, 602089 (3), Autosomal dominant; Hemangioma, capillary infantile, somatic, 602089 (3) |
| *KDSR* | 136440 | Erythrokeratodermia variabilis et progressiva 4, 617526 (3), Autosomal recessive |
| *KEL* | 613883 | [Blood group, Kell], 110900 (3) |
| *KERA* | 603288 | Cornea plana 2, autosomal recessive, 217300 (3), Autosomal recessive |
| *KHDC3L* | 611687 | Hydatidiform mole, recurrent, 2, 614293 (3), Autosomal recessive |
| *KHK* | 614058 | [Fructosuria], 229800 (3), Autosomal recessive |
| *KIAA0586* | 610178 | Short-rib thoracic dysplasia 14 with polydactyly, 616546 (3), Autosomal recessive; Joubert syndrome 23, 616490 (3), Autosomal recessive |
| *KIAA0753* | 617112 | ?Orofaciodigital syndrome XV, 617127 (3), Autosomal recessive |
| *KIAA0825* | 617266 | Polydactyly, postaxial, type A10, 618498 (3), Autosomal recessive |
| *KIAA1109* | 611565 | Alkuraya-Kucinskas syndrome, 617822 (3), Autosomal recessive |
| *KIAA1549* | 613344 | Retinitis pigmentosa 86, 618613 (3), Autosomal recessive |
| *KIDINS220* | 615759 | Spastic paraplegia, intellectual disability, nystagmus, and obesity, 617296 (3), Autosomal dominant |
| *KIF11* | 148760 | Microcephaly with or without chorioretinopathy, lymphedema, or mental retardation, 152950 (3), Autosomal dominant |
| *KIF14* | 611279 | Microcephaly 20, primary, autosomal recessive, 617914 (3), Autosomal recessive; ?Meckel syndrome 12, 616258 (3), Autosomal recessive |
| *KIF1A* | 601255 | NESCAV syndrome, 614255 (3), Autosomal dominant; Neuropathy, hereditary sensory, type IIC, 614213 (3), Autosomal recessive; Spastic paraplegia 30, autosomal dominant, 610357 (3), Autosomal recessive, Autosomal dominant; Spastic paraplegia 30, autosomal recessive, 610357 (3), Autosomal recessive, Autosomal dominant |
| *KIF1B* | 605995 | Pheochromocytoma, 171300 (3), Autosomal dominant; {Neuroblastoma, susceptibility to, 1}, 256700 (3), Somatic mutation, Autosomal dominant; Charcot-Marie-Tooth disease, type 2A1, 118210 (3), Autosomal dominant |
| *KIF1C* | 603060 | Spastic ataxia 2, autosomal recessive, 611302 (3), Autosomal recessive |
| *KIF20A* | 605664 | ?Cardiomyopathy, familial restrictive, 6, 619433 (3), Autosomal recessive |
| *KIF21A* | 608283 | Fibrosis of extraocular muscles, congenital, 3B, 135700 (3), Autosomal dominant; Fibrosis of extraocular muscles, congenital, 1, 135700 (3), Autosomal dominant |
| *KIF22* | 603213 | Spondyloepimetaphyseal dysplasia with joint laxity, type 2, 603546 (3), Autosomal dominant |
| *KIF2A* | 602591 | Cortical dysplasia, complex, with other brain malformations 3, 615411 (3), Autosomal dominant |
| *KIF3B* | 603754 | Retinitis pigmentosa 89, 618955 (3), Autosomal dominant |
| *KIF4A* | 300521 | ?Mental retardation, X-linked 100, 300923 (3), X-linked recessive |
| *KIF5A* | 602821 | Myoclonus, intractable, neonatal, 617235 (3), Autosomal dominant; {Amyotrophic lateral sclerosis, susceptibility to, 25}, 617921 (3), Autosomal dominant; Spastic paraplegia 10, autosomal dominant, 604187 (3), Autosomal dominant |
| *KIF5C* | 604593 | Cortical dysplasia, complex, with other brain malformations 2, 615282 (3), Autosomal dominant |
| *KIF7* | 611254 | Joubert syndrome 12, 200990 (3), Autosomal recessive; Acrocallosal syndrome, 200990 (3), Autosomal recessive; ?Hydrolethalus syndrome 2, 614120 (3), Autosomal recessive; ?Al-Gazali-Bakalinova syndrome, 607131 (3), Autosomal recessive |
| *KIFBP (KIF1BP)* | 609367 | Goldberg-Shprintzen megacolon syndrome, 609460 (3), Autosomal recessive |
| *KIR3DL1* | 604946 | {AIDS, delayed/rapid progression to}, 609423 (3) |
| *KIRREL1* | 607428 | Nephrotic syndrome, type 23, 619201 (3), Autosomal recessive |
| *KISS1* | 603286 | ?Hypogonadotropic hypogonadism 13 with or without anosmia, 614842 (3), Autosomal recessive |
| *KISS1R* | 604161 | Hypogonadotropic hypogonadism 8 with or without anosmia, 614837 (3), Autosomal recessive; ?Precocious puberty, central, 1, 176400 (3), Autosomal dominant |
| *KIT* | 164920 | Gastrointestinal stromal tumor, familial, 606764 (3), Isolated cases, Autosomal dominant; Mastocytosis, cutaneous, 154800 (3), Autosomal dominant; Piebaldism, 172800 (3), Autosomal dominant; Germ cell tumors, somatic, 273300 (3); Mastocytosis, systemic, somatic, 154800 (3); Leukemia, acute myeloid, somatic, 601626 (3) |
| *KITLG* | 184745 | Hyperpigmentation with or without hypopigmentation, 145250 (3), Autosomal dominant; Deafness, autosomal dominant 69, unilateral or asymmetric, 616697 (3), Autosomal dominant; [Skin/hair/eye pigmentation 7, blond/brown hair], 611664 (3) |
| *KIZ* | 615757 | Retinitis pigmentosa 69, 615780 (3), Autosomal recessive |
| *KL* | 604824 | ?Tumoral calcinosis, hyperphosphatemic, familial, 3, 617994 (3), Autosomal recessive |
| *KLC2* | 611729 | Spastic paraplegia, optic atrophy, and neuropathy, 609541 (3), Autosomal recessive |
| *KLF1* | 600599 | Blood group--Lutheran inhibitor, 111150 (3); Dyserythropoietic anemia, congenital, type IV, 613673 (3), Autosomal dominant; [Hereditary persistence of fetal hemoglobin], 613566 (3) |
| *KLF11* | 603301 | Maturity-onset diabetes of the young, type VII, 610508 (3) |
| *KLF6* | 602053 | Gastric cancer, somatic, 613659 (3); Prostate cancer, somatic, 176807 (3) |
| *KLHDC8B* | 613169 | {Hodgkin lymphoma, susceptibility to}, 236000 (3), Autosomal recessive |
| *KLHL10* | 608778 | Spermatogenic failure 11, 615081 (3), Autosomal dominant |
| *KLHL15* | 300980 | Mental retardation, X-linked 103, 300982 (3), X-linked recessive |
| *KLHL24* | 611295 | Epidermolysis bullosa simplex, generalized, with scarring and hair loss, 617294 (3), Autosomal dominant |
| *KLHL3* | 605775 | Pseudohypoaldosteronism, type IID, 614495 (3), Autosomal recessive, Autosomal dominant |
| *KLHL40* | 615340 | Nemaline myopathy 8, autosomal recessive, 615348 (3), Autosomal recessive |
| *KLHL41* | 607701 | Nemaline myopathy 9, 615731 (3), Autosomal recessive |
| *KLHL7* | 611119 | Retinitis pigmentosa 42, 612943 (3), Autosomal dominant; PERCHING syndrome, 617055 (3), Autosomal recessive |
| *KLK1* | 147910 | [Kallikrein, decreased urinary activity of], 615953 (3) |
| *KLK4* | 603767 | Amelogenesis imperfecta, type IIA1, 204700 (3), Autosomal recessive |
| *KLKB1* | 229000 | Fletcher factor (prekallikrein) deficiency, 612423 (3), Autosomal recessive |
| *KLLN* | 612105 | Cowden syndrome 4, 615107 (3) |
| *KMT2A* | 159555 | Wiedemann-Steiner syndrome, 605130 (3), Autosomal dominant |
| *KMT2B* | 606834 | Dystonia 28, childhood-onset, 617284 (3), Autosomal dominant |
| *KMT2C* | 606833 | Kleefstra syndrome 2, 617768 (3), Autosomal dominant |
| *KMT2D* | 602113 | Kabuki syndrome 1, 147920 (3), Autosomal dominant |
| *KMT2E* | 608444 | O'Donnell-Luria-Rodan syndrome, 618512 (3), Autosomal dominant |
| *KMT5B* | 610881 | Mental retardation, autosomal dominant 51, 617788 (3), Autosomal dominant |
| *KNG1* | 612358 | [Kininogen deficiency], 228960 (3), Autosomal recessive; Angioedema, hereditary, 6, 619363 (3), Autosomal dominant; [High molecular weight kininogen deficiency], 228960 (3), Autosomal recessive |
| *KNL1* | 609173 | Microcephaly 4, primary, autosomal recessive, 604321 (3), Autosomal recessive |
| *KNSTRN* | 614718 | ?Roifman-Chitayat syndrome, digenic, 613328 (3), Digenic recessive |
| *KPTN* | 615620 | Mental retardation, autosomal recessive 41, 615637 (3), Autosomal recessive |
| *KRAS* | 190070 | Gastric cancer, somatic, 137215 (3); Oculoectodermal syndrome, somatic, 600268 (3); Breast cancer, somatic, 114480 (3); Noonan syndrome 3, 609942 (3), Autosomal dominant; RAS-associated autoimmune leukoproliferative disorder, 614470 (3), Autosomal dominant; Arteriovenous malformation of the brain, somatic, 108010 (3); Lung cancer, somatic, 211980 (3); Pancreatic carcinoma, somatic, 260350 (3); Leukemia, acute myeloid, somatic, 601626 (3); Schimmelpenning-Feuerstein-Mims syndrome, somatic mosaic, 163200 (3); Cardiofaciocutaneous syndrome 2, 615278 (3), Autosomal dominant; Bladder cancer, somatic, 109800 (3) |
| *KREMEN1* | 609898 | Ectodermal dysplasia 13, hair/tooth type, 617392 (3), Autosomal recessive |
| *KRIT1* | 604214 | Hyperkeratotic cutaneous capillary-venous malformations associated with cerebral capillary malformations, 116860 (3), Autosomal dominant; Cerebral cavernous malformations-1, 116860 (3), Autosomal dominant; Cavernous malformations of CNS and retina, 116860 (3), Autosomal dominant |
| *KRT1* | 139350 | Ichthyosis, cyclic, with epidermolytic hyperkeratosis, 607602 (3), Autosomal dominant; Epidermolytic hyperkeratosis, 113800 (3), Autosomal recessive, Autosomal dominant; Palmoplantar keratoderma, nonepidermolytic, 600962 (3), Autosomal dominant; Keratosis palmoplantaris striata III, 607654 (3); Palmoplantar keratoderma, epidermolytic, 144200 (3), Autosomal dominant; Ichthyosis histrix, Curth-Macklin type, 146590 (3), Autosomal dominant |
| *KRT10* | 148080 | Epidermolytic hyperkeratosis, 113800 (3), Autosomal recessive, Autosomal dominant; Ichthyosis with confetti, 609165 (3), Autosomal dominant; Ichthyosis, cyclic, with epidermolytic hyperkeratosis, 607602 (3), Autosomal dominant |
| *KRT12* | 601687 | Meesmann corneal dystrophy 1, 122100 (3), Autosomal dominant |
| *KRT13* | 148065 | White sponge nevus 2, 615785 (3), Autosomal dominant |
| *KRT14* | 148066 | Epidermolysis bullosa simplex, recessive 1, 601001 (3), Autosomal recessive; Epidermolysis bullosa simplex, Weber-Cockayne type, 131800 (3), Autosomal dominant; Dermatopathia pigmentosa reticularis, 125595 (3), Autosomal dominant; Epidermolysis bullosa simplex, Koebner type, 131900 (3), Autosomal dominant; Naegeli-Franceschetti-Jadassohn syndrome, 161000 (3), Autosomal dominant; Epidermolysis bullosa simplex, Dowling-Meara type, 131760 (3), Autosomal dominant |
| *KRT16* | 148067 | Palmoplantar keratoderma, nonepidermolytic, focal, 613000 (3), Autosomal dominant; Pachyonychia congenita 1, 167200 (3), Autosomal dominant |
| *KRT17* | 148069 | Steatocystoma multiplex, 184500 (3), Autosomal dominant; Pachyonychia congenita 2, 167210 (3), Autosomal dominant |
| *KRT18* | 148070 | Cirrhosis, cryptogenic, 215600 (3), Autosomal recessive; {Cirrhosis, noncryptogenic, susceptibility to}, 215600 (3), Autosomal recessive |
| *KRT2* | 600194 | Ichthyosis bullosa of Siemens, 146800 (3), Autosomal dominant |
| *KRT25* | 616646 | Woolly hair, autosomal recessive 3, 616760 (3), Autosomal recessive |
| *KRT3* | 148043 | Meesmann corneal dystrophy 2, 618767 (3), Autosomal dominant |
| *KRT4* | 123940 | White sponge nevus 1, 193900 (3), Autosomal dominant |
| *KRT5* | 148040 | Dowling-Degos disease 1, 179850 (3), Autosomal dominant; Epidermolysis bullosa simplex-MP, 131960 (3), Autosomal dominant; Epidermolysis bullosa simplex, Koebner type, 131900 (3), Autosomal dominant; Epidermolysis bullosa simplex-MCR, 609352 (3); Epidermolysis bullosa simplex, Weber-Cockayne type, 131800 (3), Autosomal dominant; Epidermolysis bullosa simplex, recessive 1, 601001 (3), Autosomal recessive; Epidermolysis bullosa simplex, Dowling-Meara type, 131760 (3), Autosomal dominant |
| *KRT6A* | 148041 | Pachyonychia congenita 3, 615726 (3), Autosomal dominant |
| *KRT6B* | 148042 | Pachyonychia congenita 4, 615728 (3), Autosomal dominant |
| *KRT6C* | 612315 | Palmoplantar keratoderma, nonepidermolytic, focal or diffuse, 615735 (3), Autosomal dominant |
| *KRT71* | 608245 | ?Hypotrichosis 13, 615896 (3), Autosomal dominant |
| *KRT74* | 608248 | Woolly hair, autosomal dominant, 194300 (3), Autosomal dominant; ?Hypotrichosis 3, 613981 (3), Autosomal dominant; ?Ectodermal dysplasia 7, hair/nail type, 614929 (3), Autosomal recessive |
| *KRT75* | 609025 | {Pseudofolliculitis barbae, susceptibility to}, 612318 (3) |
| *KRT8* | 148060 | {Cirrhosis, noncryptogenic, susceptibility to}, 215600 (3), Autosomal recessive; Cirrhosis, cryptogenic, 215600 (3), Autosomal recessive |
| *KRT81* | 602153 | Monilethrix, 158000 (3), Autosomal dominant |
| *KRT83* | 602765 | Monilethrix, 158000 (3), Autosomal dominant; Erythrokeratodermia variabilis et progressiva 5, 617756 (3), Autosomal recessive |
| *KRT85* | 602767 | Ectodermal dysplasia 4, hair/nail type, 602032 (3), Autosomal recessive |
| *KRT86* | 601928 | Monilethrix, 158000 (3), Autosomal dominant |
| *KRT9* | 607606 | Palmoplantar keratoderma, epidermolytic, 144200 (3), Autosomal dominant |
| *KY* | 605739 | Myopathy, myofibrillar, 7, 617114 (3), Autosomal recessive |
| *KYNU* | 605197 | ?Hydroxykynureninuria, 236800 (3), Autosomal recessive; Vertebral, cardiac, renal, and limb defects syndrome 2, 617661 (3), Autosomal recessive |
| *L1CAM* | 308840 | MASA syndrome, 303350 (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 with Hirschsprung disease, 307000 (3), X-linked recessive; Hydrocephalus due to aqueductal stenosis, 307000 (3), X-linked recessive |
| *L2HGDH* | 609584 | L-2-hydroxyglutaric aciduria, 236792 (3), Autosomal recessive |
| *LACC1* | 613409 | Juvenile arthritis, 618795 (3), Autosomal recessive |
| *LAGE3* | 300060 | Galloway-Mowat syndrome 2, X-linked, 301006 (3), X-linked recessive |
| *LAMA1* | 150320 | Poretti-Boltshauser syndrome, 615960 (3), Autosomal recessive |
| *LAMA2* | 156225 | Muscular dystrophy, limb-girdle, autosomal recessive 23, 618138 (3), Autosomal recessive; Muscular dystrophy, congenital, merosin deficient or partially deficient, 607855 (3), Autosomal recessive |
| *LAMA3* | 600805 | Laryngoonychocutaneous syndrome, 245660 (3), Autosomal recessive; Epidermolysis bullosa, junctional, Herlitz type, 226700 (3), Autosomal recessive; Epidermolysis bullosa, generalized atrophic benign, 226650 (3), Autosomal recessive |
| *LAMA4* | 600133 | Cardiomyopathy, dilated, 1JJ, 615235 (3), Autosomal dominant |
| *LAMB1* | 150240 | Lissencephaly 5, 615191 (3), Autosomal recessive |
| *LAMB2* | 150325 | Nephrotic syndrome, type 5, with or without ocular abnormalities, 614199 (3); Pierson syndrome, 609049 (3), Autosomal recessive |
| *LAMB3* | 150310 | Epidermolysis bullosa, junctional, non-Herlitz type, 226650 (3), Autosomal recessive; Epidermolysis bullosa, junctional, Herlitz type, 226700 (3), Autosomal recessive; Amelogenesis imperfecta, type IA, 104530 (3), Autosomal dominant |
| *LAMC2* | 150292 | Epidermolysis bullosa, junctional, non-Herlitz type, 226650 (3), Autosomal recessive; Epidermolysis bullosa, junctional, Herlitz type, 226700 (3), Autosomal recessive |
| *LAMC3* | 604349 | Cortical malformations, occipital, 614115 (3), Autosomal recessive |
| *LAMP2* | 309060 | Danon disease, 300257 (3), X-linked dominant |
| *LAMTOR2* | 610389 | :Immunodeficiency due to defect in MAPBP-interacting protein, 610798 (3), Autosomal recessive |
| *LARGE1* | 603590 | Muscular dystrophy-dystroglycanopathy (congenital with 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 |
| *LARP7* | 612026 | Alazami syndrome, 615071 (3), Autosomal recessive |
| *LARS1 (LARS)* | 151350 | ?Infantile liver failure syndrome 1, 615438 (3), Autosomal recessive |
| *LARS2* | 604544 | Perrault syndrome 4, 615300 (3), Autosomal recessive; Hydrops, lactic acidosis, and sideroblastic anemia, 617021 (3), Autosomal recessive |
| *LAS1L* | 300964 | Wilson-Turner syndrome, 309585 (3), X-linked recessive |
| *LAT* | 602354 | Immunodeficiency 52, 617514 (3), Autosomal recessive |
| *LBR* | 600024 | Pelger-Huet anomaly, 169400 (3), Autosomal dominant; Pelger-Huet anomaly with mild skeletal anomalies, 618019 (3); ?Reynolds syndrome, 613471 (3), Autosomal dominant; Greenberg skeletal dysplasia, 215140 (3), Autosomal recessive |
| *LCA5* | 611408 | Leber congenital amaurosis 5, 604537 (3), Autosomal recessive |
| *LCAT* | 606967 | Fish-eye disease, 136120 (3), Autosomal recessive; Norum disease, 245900 (3), Autosomal recessive |
| *LCK* | 153390 | ?Immunodeficiency 22, 615758 (3), Autosomal recessive |
| *LCP2* | 601603 | ?Immunodeficiency 81, 619374 (3), Autosomal recessive |
| *LCT* | 603202 | Lactase deficiency, congenital, 223000 (3), Autosomal recessive |
| *LDB3* | 605906 | Left ventricular noncompaction 3, 601493 (3), Autosomal dominant; Cardiomyopathy, hypertrophic, 24, 601493 (3), Autosomal dominant; Myopathy, myofibrillar, 4, 609452 (3), Autosomal dominant; Cardiomyopathy, dilated, 1C, with or without LVNC, 601493 (3), Autosomal dominant |
| *LDHA* | 150000 | Glycogen storage disease XI, 612933 (3), Autosomal recessive |
| *LDHB* | 150100 | [Lactate dehydrogenase-B deficiency], 614128 (3) |
| *LDHD* | 607490 | D-lactic aciduria with susceptibility to gout, 245450 (3), Autosomal recessive |
| *LDLR* | 606945 | LDL cholesterol level QTL2, 143890 (3), Autosomal recessive, Autosomal dominant; Hypercholesterolemia, familial, 1, 143890 (3), Autosomal recessive, Autosomal dominant |
| *LDLRAP1* | 605747 | Hypercholesterolemia, familial, 4, 603813 (3), Autosomal recessive |
| *LEF1* | 153245 | Sebaceous tumors, somatic (3) |
| *LEMD2* | 616312 | Marbach-Rustad progeroid syndrome, 619322 (3), Autosomal dominant; Cataract 46, juvenile-onset, 212500 (3), Autosomal recessive |
| *LEMD3* | 607844 | Buschke-Ollendorff syndrome, 166700 (3), Autosomal dominant; Osteopoikilosis with or without melorheostosis, 166700 (3), Autosomal dominant |
| *LEP* | 164160 | Obesity, morbid, due to leptin deficiency, 614962 (3), Autosomal recessive |
| *LEPR* | 601007 | Obesity, morbid, due to leptin receptor deficiency, 614963 (3), Autosomal recessive |
| *LFNG* | 602576 | Spondylocostal dysostosis 3, autosomal recessive, 609813 (3), Autosomal recessive |
| *LGALS2* | 150571 | {Myocardial infarction, susceptibility to}, 608446 (3) |
| *LGI1* | 604619 | Epilepsy, familial temporal lobe, 1, 600512 (3), Autosomal dominant |
| *LGI4* | 608303 | Arthrogryposis multiplex congenita 1, neurogenic, with myelin defect, 617468 (3), Autosomal recessive |
| *LGR4* | 606666 | {Bone mineral density, low, susceptibility to}, 615311 (3) |
| *LHB* | 152780 | Hypogonadotropic hypogonadism 23 with or without anosmia, 228300 (3), Autosomal recessive |
| *LHCGR* | 152790 | Leydig cell adenoma, somatic, with precocious puberty, 176410 (3); Leydig cell hypoplasia with pseudohermaphroditism, 238320 (3), Autosomal recessive; Leydig cell hypoplasia with hypergonadotropic hypogonadism, 238320 (3), Autosomal recessive; Luteinizing hormone resistance, female, 238320 (3), Autosomal recessive; Precocious puberty, male, 176410 (3), Autosomal dominant |
| *LHFPL5* | 609427 | Deafness, autosomal recessive 67, 610265 (3), Autosomal recessive |
| *LHX3* | 600577 | Pituitary hormone deficiency, combined, 3, 221750 (3), Autosomal recessive |
| *LHX4* | 602146 | Pituitary hormone deficiency, combined, 4, 262700 (3), Autosomal dominant |
| *LIAS* | 607031 | Hyperglycinemia, lactic acidosis, and seizures, 614462 (3), Autosomal recessive |
| *LIFR* | 151443 | Stuve-Wiedemann syndrome/Schwartz-Jampel type 2 syndrome, 601559 (3), Autosomal recessive |
| *LIG4* | 601837 | LIG4 syndrome, 606593 (3), Autosomal recessive; {Multiple myeloma, resistance to}, 254500 (3), Somatic mutation |
| *LIM2* | 154045 | Cataract 19, multiple types, 615277 (3), Autosomal recessive |
| *LIMA1* | 608364 | [Low density lipoprotein cholesterol level QTL 8], 618079 (3) |
| *LIMS2* | 607908 | ?Muscular dystrophy, autosomal recessive, with cardiomyopathy and triangular tongue, 616827 (3), Autosomal recessive |
| *LINGO1* | 609791 | Mental retardation, autosomal recessive 64, 618103 (3), Autosomal recessive |
| *LINS1* | 610350 | Mental retardation, autosomal recessive 27, 614340 (3), Autosomal recessive |
| *LIPA* | 613497 | Wolman disease, 278000 (3), Autosomal recessive; Cholesteryl ester storage disease, 278000 (3), Autosomal recessive |
| *LIPC* | 151670 | {Diabetes mellitus, noninsulin-dependent}, 125853 (3), Autosomal dominant; Hepatic lipase deficiency, 614025 (3), Autosomal recessive; [High density lipoprotein cholesterol level QTL 12], 612797 (3) |
| *LIPE* | 151750 | Lipodystrophy, familial partial, type 6, 615980 (3), Autosomal recessive |
| *LIPH* | 607365 | Hypotrichosis 7, 604379 (3), Autosomal recessive; Woolly hair, autosomal recessive 2 with or without hypotrichosis, 604379 (3), Autosomal recessive |
| *LIPN* | 613924 | Ichthyosis, congenital, autosomal recessive 8, 613943 (3), Autosomal recessive |
| *LIPT1* | 610284 | Lipoyltransferase 1 deficiency, 616299 (3), Autosomal recessive |
| *LIPT2* | 617659 | Encephalopathy, neonatal severe, with lactic acidosis and brain abnormalities, 617668 (3), Autosomal recessive |
| *LITAF* | 603795 | Charcot-Marie-Tooth disease, type 1C, 601098 (3), Autosomal dominant |
| *LMAN1* | 601567 | Combined factor V and VIII deficiency, 227300 (3), Autosomal recessive |
| *LMAN2L* | 609552 | ?Mental retardation, autosomal recessive, 52, 616887 (3), Autosomal recessive |
| *LMBR1* | 605522 | Triphalangeal thumb, type I, 174500 (3), Autosomal dominant; Syndactyly, type IV, 186200 (3), Autosomal dominant; 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; Acheiropody, 200500 (3), Autosomal recessive; Triphalangeal thumb-polysyndactyly syndrome, 174500 (3), Autosomal dominant |
| *LMBRD1* | 612625 | Methylmalonic aciduria and homocystinuria, cblF type, 277380 (3), Autosomal recessive |
| *LMF1* | 611761 | Lipase deficiency, combined, 246650 (3), Autosomal recessive |
| *LMNA* | 150330 | Mandibuloacral dysplasia, 248370 (3), Autosomal recessive; Heart-hand syndrome, Slovenian type, 610140 (3), Autosomal dominant; Cardiomyopathy, dilated, 1A, 115200 (3), Autosomal dominant; Restrictive dermopathy, lethal, 275210 (3), Autosomal recessive; Emery-Dreifuss muscular dystrophy 3, autosomal recessive, 616516 (3), Autosomal recessive; Charcot-Marie-Tooth disease, type 2B1, 605588 (3), Autosomal recessive; Emery-Dreifuss muscular dystrophy 2, autosomal dominant, 181350 (3), Autosomal dominant; Hutchinson-Gilford progeria, 176670 (3), Autosomal dominant; Lipodystrophy, familial partial, type 2, 151660 (3), Autosomal dominant; Muscular dystrophy, congenital, 613205 (3), Autosomal dominant; Malouf syndrome, 212112 (3), Autosomal dominant |
| *LMNB1* | 150340 | Leukodystrophy, adult-onset, autosomal dominant, 169500 (3), Autosomal dominant; Microcephaly 26, primary, autosomal dominant, 619179 (3), Autosomal dominant |
| *LMNB2* | 150341 | Microcephaly 27, primary, autosomal dominant, 619180 (3); ?Epilepsy, progressive myoclonic, 9, 616540 (3), Autosomal recessive; {Lipodystrophy, partial, acquired, susceptibility to}, 608709 (3), Autosomal dominant |
| *LMO1* | 186921 | Leukemia, T-cell acute lymphoblastic, 186921 (2) |
| *LMO2* | 180385 | Leukemia, acute T-cell, 180385 (2) |
| *LMOD1* | 602715 | ?Megacystis-microcolon-intestinal hypoperistalsis syndrome 3, 619362 (3), Autosomal recessive |
| *LMOD3* | 616112 | Nemaline myopathy 10, 616165 (3), Autosomal recessive |
| *LMX1A* | 600298 | Deafness, autosomal dominant 7, 601412 (3), Autosomal dominant |
| *LMX1B* | 602575 | Focal segmental glomerulosclerosis 10, 256020 (3), Autosomal dominant; Nail-patella syndrome, 161200 (3), Autosomal dominant |
| *LNPK* | 610236 | Neurodevelopmental disorder with epilepsy and hypoplasia of the corpus callosum, 618090 (3), Autosomal recessive |
| *LONP1* | 605490 | CODAS syndrome, 600373 (3), Autosomal recessive |
| *LORICRIN (LOR)* | 152445 | Vohwinkel syndrome with ichthyosis, 604117 (3), Autosomal dominant |
| *LOX* | 153455 | Aortic aneurysm, familial thoracic 10, 617168 (3), Autosomal dominant |
| *LOXHD1* | 613072 | Deafness, autosomal recessive 77, 613079 (3), Autosomal recessive |
| *LOXL1* | 153456 | {Exfoliation syndrome, susceptibility to}, 177650 (3), Autosomal dominant |
| *LPA* | 152200 | [LPA deficiency, congenital], 618807 (3), Autosomal dominant; {Coronary artery disease, susceptibility to}, 618807 (3), Autosomal dominant |
| *LPAR6* | 609239 | Hypotrichosis 8, 278150 (3), Autosomal recessive; Woolly hair, autosomal recessive 1, with or without hypotrichosis, 278150 (3), Autosomal recessive |
| *LPIN1* | 605518 | Myoglobinuria, acute recurrent, autosomal recessive, 268200 (3), Autosomal recessive |
| *LPIN2* | 605519 | Majeed syndrome, 609628 (3) |
| *LPL* | 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 |
| *LPP* | 600700 | Lipoma (3); Leukemia, acute myeloid, 601626 (3), Somatic mutation, Autosomal dominant |
| *LRAT* | 604863 | Leber congenital amaurosis 14, 613341 (3), Autosomal recessive; Retinal dystrophy, early-onset severe, 613341 (3), Autosomal recessive; Retinitis pigmentosa, juvenile, 613341 (3), Autosomal recessive |
| *LRBA* | 606453 | Immunodeficiency, common variable, 8, with autoimmunity, 614700 (3), Autosomal recessive |
| *LRIG2* | 608869 | Urofacial syndrome 2, 615112 (3), Autosomal recessive |
| *LRIT3* | 615004 | Night blindness, congenital stationary (complete), 1F, autosomal recessive, 615058 (3), Autosomal recessive |
| *LRMDA* | 614537 | Albinism, oculocutaneous, type VII, 615179 (3), Autosomal recessive |
| *LRP1* | 107770 | ?Keratosis pilaris atrophicans, 604093 (3), Autosomal recessive |
| *LRP12* | 618299 | Oculopharyngodistal myopathy 1, 164310 (3), Autosomal dominant |
| *LRP2* | 600073 | Donnai-Barrow syndrome, 222448 (3), Autosomal recessive |
| *LRP4* | 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 |
| *LRP5* | 603506 | Osteopetrosis, autosomal dominant 1, 607634 (3), Autosomal dominant; [Bone mineral density variability 1], 601884 (3), Autosomal dominant; {Osteoporosis}, 166710 (3), 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; Exudative vitreoretinopathy 4, 601813 (3), Autosomal recessive, Autosomal dominant; van Buchem disease, type 2, 607636 (3) |
| *LRP6* | 603507 | {Coronary artery disease, autosomal dominant, 2}, 610947 (3), Autosomal dominant; Tooth agenesis, selective, 7, 616724 (3), Autosomal dominant |
| *LRP8* | 602600 | {Myocardial infarction, susceptibility to}, 608446 (3) |
| *LRPAP1* | 104225 | Myopia 23, autosomal recessive, 615431 (3), Autosomal recessive |
| *LRPPRC* | 607544 | Mitochondrial complex IV deficiency, nuclear type 5, (French-Canadian), 220111 (3), Autosomal recessive |
| *LRRC32* | 137207 | Cleft palate, proliferative retinopathy, and developmental delay, 619074 (3), Autosomal recessive |
| *LRRC56* | 618227 | Ciliary dyskinesia, primary, 39, 618254 (3), Autosomal recessive |
| *LRRC8A* | 608360 | ?Agammaglobulinemia 5, 613506 (3), Autosomal dominant |
| *LRRK2* | 609007 | {Parkinson disease 8}, 607060 (3), Autosomal dominant |
| *LRSAM1* | 610933 | Charcot-Marie-Tooth disease, axonal, type 2P, 614436 (3), Autosomal recessive, Autosomal dominant |
| *LRTOMT* | 612414 | Deafness, autosomal recessive 63, 611451 (3), Autosomal recessive |
| *LSS* | 600909 | Hypotrichosis 14, 618275 (3), Autosomal recessive; Cataract 44, 616509 (3), Autosomal recessive; Alopecia-mental retardation syndrome 4, 618840 (3), Autosomal recessive |
| *LTA* | 153440 | {Psoriatic arthritis, susceptibility to}, 607507 (3); {Myocardial infarction, susceptibility to}, 608446 (3); {Leprosy, susceptibility to, 4}, 610988 (3) |
| *LTBP1* | 150390 | Cutis laxa, autosomal recessive, type IIE, 619451 (3), Autosomal recessive |
| *LTBP2* | 602091 | Glaucoma 3, primary congenital, D, 613086 (3); Microspherophakia and/or megalocornea, with ectopia lentis and with or without secondary glaucoma, 251750 (3), Autosomal recessive; ?Weill-Marchesani syndrome 3, recessive, 614819 (3), Autosomal recessive |
| *LTBP3* | 602090 | Dental anomalies and short stature, 601216 (3), Autosomal recessive; Geleophysic dysplasia 3, 617809 (3), Autosomal dominant |
| *LTBP4* | 604710 | Cutis laxa, autosomal recessive, type IC, 613177 (3), Autosomal recessive |
| *LTC4S* | 246530 | Leukotriene C4 synthase deficiency, 614037 (1), Autosomal recessive |
| *LYL1* | 151440 | Leukemia, T-cell acute lymphoblastoid, 151440 (2) |
| *LYRM4* | 613311 | ?Combined oxidative phosphorylation deficiency 19, 615595 (3), Autosomal recessive |
| *LYRM7* | 615831 | Mitochondrial complex III deficiency, nuclear type 8, 615838 (3), Autosomal recessive |
| *LYST* | 606897 | Chediak-Higashi syndrome, 214500 (3), Autosomal recessive |
| *LYZ* | 153450 | Amyloidosis, renal, 105200 (3), Autosomal dominant |
| *LZTFL1* | 606568 | Bardet-Biedl syndrome 17, 615994 (3), Autosomal recessive |
| *LZTR1* | 600574 | Noonan syndrome 2, 605275 (3), Autosomal recessive; Noonan syndrome 10, 616564 (3), Autosomal dominant; {Schwannomatosis-2, susceptibility to}, 615670 (3), Autosomal dominant |
| *LZTS1* | 606551 | Esophageal squamous cell carcinoma, somatic, 133239 (3) |
| *M1AP* | 619098 | Spermatogenic failure 48, 619108 (3), Autosomal recessive |
| *MAB21L1* | 601280 | Cerebellar, ocular, craniofacial, and genital syndrome, 618479 (3), Autosomal recessive |
| *MAB21L2* | 604357 | Microphthalmia/coloboma and skeletal dysplasia syndrome, 615877 (3), Autosomal recessive, Autosomal dominant |
| *MACF1* | 608271 | Lissencephaly 9 with complex brainstem malformation, 618325 (3), Autosomal dominant |
| *MAD1L1* | 602686 | Prostate cancer, somatic, 176807 (3); Lymphoma, somatic (3) |
| *MAD2L2* | 604094 | ?Fanconi anemia, complementation group V, 617243 (3), Autosomal recessive |
| *MADD* | 603584 | Neurodevelopmental disorder with dysmorphic facies, impaired speech and hypotonia, 619005 (3), Autosomal recessive; DEEAH syndrome, 619004 (3), Autosomal recessive |
| *MAF* | 177075 | Cataract 21, multiple types, 610202 (3), Autosomal dominant; Ayme-Gripp syndrome, 601088 (3), Autosomal dominant |
| *MAFA* | 610303 | Insulinomatosis and diabetes mellitus, 147630 (3), Autosomal dominant |
| *MAFB* | 608968 | Duane retraction syndrome 3, 617041 (3), Autosomal dominant; Multicentric carpotarsal osteolysis syndrome, 166300 (3), Autosomal dominant |
| *MAG* | 159460 | Spastic paraplegia 75, autosomal recessive, 616680 (3), Autosomal recessive |
| *MAGED2* | 300470 | Bartter syndrome, type 5, antenatal, transient, 300971 (3), X-linked recessive |
| *MAGEL2* | 605283 | Schaaf-Yang syndrome, 615547 (3), Autosomal dominant |
| *MAGI2* | 606382 | Nephrotic syndrome, type 15, 617609 (3), Autosomal recessive |
| *MAGT1* | 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 |
| *MAK* | 154235 | Retinitis pigmentosa 62, 614181 (3), Autosomal recessive |
| *MALT1* | 604860 | Immunodeficiency 12, 615468 (3), Autosomal recessive |
| *MAML2* | 607537 | Mucoepidermoid salivary gland carcinoma (3) |
| *MAMLD1* | 300120 | Hypospadias 2, X-linked, 300758 (3), X-linked recessive |
| *MAN1B1* | 604346 | Rafiq syndrome, 614202 (3), Autosomal recessive |
| *MAN2B1* | 609458 | Mannosidosis, alpha-, types I and II, 248500 (3), Autosomal recessive |
| *MANBA* | 609489 | Mannosidosis, beta, 248510 (3), Autosomal recessive |
| *MAOA* | 309850 | {Antisocial behavior}, 300615 (3), X-linked recessive; Brunner syndrome, 300615 (3), X-linked recessive |
| *MAP1B* | 157129 | Periventricular nodular heterotopia 9, 618918 (3), Autosomal dominant |
| *MAP2K1* | 176872 | Cardiofaciocutaneous syndrome 3, 615279 (3), Autosomal dominant; Melorheostosis, isolated, somatic mosaic, 155950 (3) |
| *MAP2K2* | 601263 | Cardiofaciocutaneous syndrome 4, 615280 (3), Autosomal dominant |
| *MAP3K1* | 600982 | 46XY sex reversal 6, 613762 (3), Autosomal dominant |
| *MAP3K20* | 609479 | Centronuclear myopathy 6 with fiber-type disproportion, 617760 (3), Autosomal recessive; Split-foot malformation with mesoaxial polydactyly, 616890 (3), Autosomal recessive |
| *MAP3K7* | 602614 | Frontometaphyseal dysplasia 2, 617137 (3), Autosomal dominant; Cardiospondylocarpofacial syndrome, 157800 (3), Autosomal dominant |
| *MAP3K8* | 191195 | Lung cancer, somatic, 211980 (3) |
| *MAPK1* | 176948 | Noonan syndrome 13, 619087 (3), Autosomal dominant |
| *MAPK8IP1* | 604641 | {Diabetes mellitus, noninsulin-dependent}, 125853 (3), Autosomal dominant |
| *MAPK8IP3* | 605431 | Neurodevelopmental disorder with or without variable brain abnormalities, 618443 (3), Autosomal dominant |
| *MAPKAPK3* | 602130 | ?Macular dystrophy, patterned, 3, 617111 (3), Autosomal dominant |
| *MAPKBP1* | 616786 | Nephronophthisis 20, 617271 (3), Autosomal recessive |
| *MAPRE2* | 605789 | Symmetric circumferential skin creases, congenital, 2, 616734 (3), Autosomal dominant |
| *MAPT* | 157140 | Supranuclear palsy, progressive, 601104 (3), Autosomal dominant; Supranuclear palsy, progressive atypical, 260540 (3), Autosomal recessive; Dementia, frontotemporal, with or without parkinsonism, 600274 (3), Autosomal dominant; {Parkinson disease, susceptibility to}, 168600 (3), Multifactorial, Autosomal dominant; Pick disease, 172700 (3), Autosomal dominant |
| *MARCHF6 (MARCH6)* | 613297 | Epilepsy, familial adult myoclonic, 3, 613608 (3), Autosomal dominant |
| *MARK3* | 602678 | ?Visual impairment and progressive phthisis bulbi, 618283 (3), Autosomal recessive |
| *MARS1 (MARS)* | 156560 | Interstitial lung and liver disease, 615486 (3), Autosomal recessive; Charcot-Marie-Tooth disease, axonal, type 2U, 616280 (3), Autosomal dominant |
| *MARS2* | 609728 | ?Combined oxidative phosphorylation deficiency 25, 616430 (3), Autosomal recessive; Spastic ataxia 3, autosomal recessive, 611390 (3), Autosomal recessive |
| *MARVELD2* | 610572 | Deafness, autosomal recessive 49, 610153 (3), Autosomal recessive |
| *MASP1* | 600521 | 3MC syndrome 1, 257920 (3), Autosomal recessive |
| *MASP2* | 605102 | MASP2 deficiency, 613791 (3), Autosomal recessive |
| *MAST1* | 612256 | Mega-corpus-callosum syndrome with cerebellar hypoplasia and cortical malformations, 618273 (3), Autosomal dominant |
| *MAT1A* | 610550 | Hypermethioninemia, persistent, autosomal dominant, due to methionine adenosyltransferase I/III deficiency, 250850 (3), Autosomal recessive, Autosomal dominant; Methionine adenosyltransferase deficiency, autosomal recessive, 250850 (3), Autosomal recessive, Autosomal dominant |
| *MATN3* | 602109 | {Osteoarthritis susceptibility 2}, 140600 (3), Autosomal dominant; Spondyloepimetaphyseal dysplasia, Borochowitz-Cormier-Daire type, 608728 (3), Autosomal recessive; Epiphyseal dysplasia, multiple, 5, 607078 (3), Autosomal dominant |
| *MATR3* | 164015 | Amyotrophic lateral sclerosis 21, 606070 (3), Autosomal dominant |
| *MAX* | 154950 | {Pheochromocytoma, susceptibility to}, 171300 (3), Autosomal dominant |
| *MBD5* | 611472 | Mental retardation, autosomal dominant 1, 156200 (3), Autosomal dominant |
| *MBL2* | 154545 | {Chronic infections, due to MBL deficiency}, 614372 (3), Autosomal dominant |
| *MBOAT7* | 606048 | Mental retardation, autosomal recessive 57, 617188 (3), Autosomal recessive |
| *MBTPS1* | 603355 | ?Spondyloepiphyseal dysplasia, Kondo-Fu type, 618392 (3), Autosomal recessive |
| *MBTPS2* | 300294 | Keratosis follicularis spinulosa decalvans, X-linked, 308800 (3), X-linked recessive; Osteogenesis imperfecta, type XIX, 301014 (3), X-linked recessive; IFAP syndrome with or without BRESHECK syndrome, 308205 (3), X-linked recessive; ?Olmsted syndrome, X-linked, 300918 (3), X-linked recessive |
| *MC1R* | 155555 | [Analgesia from kappa-opioid receptor agonist, female-specific], 613098 (3); [Skin/hair/eye pigmentation 2, red hair/fair skin], 266300 (3), Autosomal recessive; [Skin/hair/eye pigmentation 2, blond hair/fair skin], 266300 (3), Autosomal recessive; {Melanoma, cutaneous malignant, 5}, 613099 (3); {Albinism, oculocutaneous, type II, modifier of}, 203200 (3), Autosomal recessive; {UV-induced skin damage}, 266300 (3), Autosomal recessive |
| *MC2R* | 607397 | Glucocorticoid deficiency, due to ACTH unresponsiveness, 202200 (3), Autosomal recessive |
| *MC3R* | 155540 | {Obesity, severe, susceptibility to, BMIQ9}, 602025 (3) |
| *MC4R* | 155541 | Obesity (BMIQ20), 618406 (3), Autosomal recessive, Autosomal dominant; {Obesity, resistance to (BMIQ20)}, 618406 (3), Autosomal recessive, Autosomal dominant |
| *MCC* | 159350 | Colorectal cancer, somatic, 114500 (3) |
| *MCCC1* | 609010 | 3-Methylcrotonyl-CoA carboxylase 1 deficiency, 210200 (3), Autosomal recessive |
| *MCCC2* | 609014 | 3-Methylcrotonyl-CoA carboxylase 2 deficiency, 210210 (3), Autosomal recessive |
| *MCEE* | 608419 | Methylmalonyl-CoA epimerase deficiency, 251120 (3), Autosomal recessive |
| *MCFD2* | 607788 | Factor V and factor VIII, combined deficiency of, 613625 (3) |
| *MCIDAS* | 614086 | Ciliary dyskinesia, primary, 42, 618695 (3), Autosomal recessive |
| *MCM10* | 609357 | Immunodeficiency 80 with or without cardiomyopathy, 619313 (3), Autosomal recessive |
| *MCM2* | 116945 | ?Deafness, autosomal dominant 70, 616968 (3), Autosomal dominant |
| *MCM3AP* | 603294 | Peripheral neuropathy, autosomal recessive, with or without impaired intellectual development, 618124 (3), Autosomal recessive |
| *MCM4* | 602638 | Immunodeficiency 54, 609981 (3), Autosomal recessive |
| *MCM5* | 602696 | ?Meier-Gorlin syndrome 8, 617564 (3), Autosomal recessive |
| *MCM6* | 601806 | Lactase persistence/nonpersistence, 223100 (3), Autosomal dominant |
| *MCM8* | 608187 | ?Premature ovarian failure 10, 612885 (3), Autosomal recessive |
| *MCM9* | 610098 | Ovarian dysgenesis 4, 616185 (3), Autosomal recessive |
| *MCOLN1* | 605248 | Mucolipidosis IV, 252650 (3), Autosomal recessive |
| *MCPH1* | 607117 | Microcephaly 1, primary, autosomal recessive, 251200 (3), Autosomal recessive |
| *MDH1* | 154200 | ?Developmental and epileptic encephalopathy 88, 618959 (3), Autosomal recessive |
| *MDH2* | 154100 | Developmental and epileptic encephalopathy 51, 617339 (3), Autosomal recessive |
| *MDM2* | 164785 | {Accelerated tumor formation, susceptibility to}, 614401 (3), Autosomal dominant; ?Lessel-Kubisch syndrome, 618681 (3), Autosomal recessive |
| *MDM4* | 602704 | ?Bone marrow failure syndrome 6, 618849 (3), Autosomal dominant |
| *MECOM* | 165215 | Radioulnar synostosis with amegakaryocytic thrombocytopenia 2, 616738 (3), Autosomal dominant |
| *MECP2* | 300005 | Rett syndrome, atypical, 312750 (3), X-linked dominant; Encephalopathy, neonatal severe, 300673 (3), X-linked recessive; Mental retardation, X-linked, syndromic 13, 300055 (3), X-linked recessive; {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; Mental retardation, X-linked syndromic, Lubs type, 300260 (3), X-linked recessive |
| *MECR* | 608205 | Dystonia, childhood-onset, with optic atrophy and basal ganglia abnormalities, 617282 (3), Autosomal recessive |
| *MED12* | 300188 | Lujan-Fryns syndrome, 309520 (3), X-linked recessive; Ohdo syndrome, X-linked, 300895 (3), X-linked recessive; Opitz-Kaveggia syndrome, 305450 (3), X-linked recessive |
| *MED12L* | 611318 | Nizon-Isidor syndrome, 618872 (3), Autosomal dominant |
| *MED13* | 603808 | Intellectual developmental disorder 61, 618009 (3), Autosomal dominant |
| *MED13L* | 608771 | Impaired intellectual development and distinctive facial features with or without cardiac defects, 616789 (3), Autosomal dominant; Transposition of the great arteries, dextro-looped 1, 608808 (3), Autosomal dominant |
| *MED17* | 603810 | Microcephaly, postnatal progressive, with seizures and brain atrophy, 613668 (3), Autosomal recessive |
| *MED23* | 605042 | Mental retardation, autosomal recessive 18, 614249 (3), Autosomal recessive |
| *MED25* | 610197 | Basel-Vanagait-Smirin-Yosef syndrome, 616449 (3), Autosomal recessive |
| *MED27* | 605044 | Neurodevelopmental disorder with spasticity, cataracts, and cerebellar hypoplasia, 619286 (3), Autosomal recessive |
| *MEF2A* | 600660 | {Coronary artery disease, autosomal dominant, 1}, 608320 (3), Autosomal dominant |
| *MEF2C* | 600662 | Mental retardation, stereotypic movements, epilepsy, and/or cerebral malformations, 613443 (3), Autosomal dominant; Chromosome 5q14.3 deletion syndrome, 613443 (4), Autosomal dominant |
| *MEFV* | 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 |
| *MEGF10* | 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 |
| *MEGF8* | 604267 | Carpenter syndrome 2, 614976 (3), Autosomal recessive |
| *MEI1* | 608797 | Hydatidiform mole, recurrent, 3, 618431 (3), Autosomal recessive |
| *MEIOB* | 617670 | ?Spermatogenic failure 22, 617706 (3), Autosomal recessive |
| *MEIS2* | 601740 | Cleft palate, cardiac defects, and mental retardation, 600987 (3), Autosomal dominant |
| *MEN1* | 613733 | Lipoma, somatic (3); Angiofibroma, somatic (3); Multiple endocrine neoplasia 1, 131100 (3), Autosomal dominant; Carcinoid tumor of lung (3); Adrenal adenoma, somatic (3); Parathyroid adenoma, somatic (3) |
| *MEOX1* | 600147 | Klippel-Feil syndrome 2, 214300 (3), Autosomal recessive |
| *MERTK* | 604705 | Retinitis pigmentosa 38, 613862 (3), Autosomal recessive |
| *MESD* | 607783 | Osteogenesis imperfecta, type XX, 618644 (3), Autosomal recessive |
| *MESP2* | 605195 | Spondylocostal dysostosis 2, autosomal recessive, 608681 (3), Autosomal recessive |
| *MET* | 164860 | Renal cell carcinoma, papillary, 1, familial and somatic, 605074 (3); Hepatocellular carcinoma, childhood type, somatic, 114550 (3); {Osteofibrous dysplasia, susceptibility to}, 607278 (3), Autosomal dominant; ?Deafness, autosomal recessive 97, 616705 (3), Autosomal recessive |
| *METTL13 (EEF1AKNMT)* | 617987 | ?{Deafness, autosomal recessive 26, modifier of}, 605429 (3), Autosomal dominant |
| *METTL23* | 615262 | Mental retardation, autosomal recessive 44, 615942 (3), Autosomal recessive |
| *METTL5* | 618628 | Intellectual developmental disorder, autosomal recessive 72, 618665 (3), Autosomal recessive |
| *MFAP5* | 601103 | Aortic aneurysm, familial thoracic 9, 616166 (3), Autosomal dominant |
| *MFF* | 614785 | Encephalopathy due to defective mitochondrial and peroxisomal fission 2, 617086 (3), Autosomal recessive |
| *MFHAS1* | 605352 | Malignant fibrous histiocytoma, 605352 (2) |
| *MFN2* | 608507 | Charcot-Marie-Tooth disease, axonal, type 2A2A, 609260 (3), Autosomal dominant; Charcot-Marie-Tooth disease, axonal, type 2A2B, 617087 (3), Autosomal recessive; Hereditary motor and sensory neuropathy VIA, 601152 (3), Autosomal dominant |
| *MFRP* | 606227 | Microphthalmia, isolated 5, 611040 (3), Autosomal recessive; Nanophthalmos 2, 609549 (3) |
| *MFSD2A* | 614397 | Neurodevelopmental disorder with progressive microcephaly, spasticity, and brain abnormalities, 616486 (3), Autosomal recessive |
| *MFSD8* | 611124 | Macular dystrophy with central cone involvement, 616170 (3), Autosomal recessive; Ceroid lipofuscinosis, neuronal, 7, 610951 (3), Autosomal recessive |
| *MGAT2* | 602616 | Congenital disorder of glycosylation, type IIa, 212066 (3), Autosomal recessive |
| *MGME1* | 615076 | Mitochondrial DNA depletion syndrome 11, 615084 (3), Autosomal recessive |
| *MGP* | 154870 | Keutel syndrome, 245150 (3), Autosomal recessive |
| *MIA3* | 613455 | ?Ondontochondrodysplasia 2 with hearing loss and diabetes, 619269 (3), Autosomal recessive |
| *MIB1* | 608677 | Left ventricular noncompaction 7, 615092 (3), Autosomal dominant |
| *MICOS13 (C19orf70)* | 616658 | Combined oxidative phosphorylation deficiency 37, 618329 (3), Autosomal recessive |
| *MICU1* | 605084 | Myopathy with extrapyramidal signs, 615673 (3), Autosomal recessive |
| *MID1* | 300552 | Opitz GBBB syndrome, type I, 300000 (3), X-linked recessive |
| *MID2* | 300204 | ?Mental retardation, X-linked 101, 300928 (3), X-linked recessive |
| *MIEF2* | 615498 | ?Combined oxidative phosphorylation deficiency 49, 619024 (3), Autosomal recessive |
| *MIF* | 153620 | {Rheumatoid arthritis, systemic juvenile, susceptibility to}, 604302 (3) |
| *MINPP1* | 605391 | {Thyroid carcinoma, follicular}, 188470 (3), Somatic mutation, Autosomal dominant |
| *MIP* | 154050 | Cataract 15, multiple types, 615274 (3), Autosomal dominant |
| *MIPEP* | 602241 | Combined oxidative phosphorylation deficiency 31, 617228 (3), Autosomal recessive |
| *MIR204* | 610942 | ?Retinal dystrophy and iris coloboma with or without cataract, 616722 (3), Autosomal dominant |
| *MIR2861* | 613405 | [Bone mineral density QTL 15], 613418 (3), Autosomal recessive, Autosomal dominant |
| *MITF* | 156845 | Waardenburg syndrome, type 2A, 193510 (3), Autosomal dominant; {Melanoma, cutaneous malignant, susceptibility to, 8}, 614456 (3); Tietz albinism-deafness syndrome, 103500 (3), Autosomal dominant; Waardenburg syndrome/ocular albinism, digenic, 103470 (3); COMMAD syndrome, 617306 (3), Autosomal recessive |
| *MKKS* | 604896 | McKusick-Kaufman syndrome, 236700 (3), Autosomal recessive; Bardet-Biedl syndrome 6, 605231 (3), Autosomal recessive |
| *MKRN3* | 603856 | Precocious puberty, central, 2, 615346 (3), Autosomal dominant |
| *MKS1* | 609883 | Bardet-Biedl syndrome 13, 615990 (3), Autosomal recessive; Meckel syndrome 1, 249000 (3), Autosomal recessive; Joubert syndrome 28, 617121 (3), Autosomal recessive |
| *MLC1* | 605908 | Megalencephalic leukoencephalopathy with subcortical cysts, 604004 (3), Autosomal recessive |
| *MLH1* | 120436 | Colorectal cancer, hereditary nonpolyposis, type 2, 609310 (3); Muir-Torre syndrome, 158320 (3), Autosomal dominant; Mismatch repair cancer syndrome 1, 276300 (3), Autosomal recessive |
| *MLH3* | 604395 | {Endometrial cancer, susceptibility to}, 608089 (3), Somatic mutation, Autosomal dominant; Colorectal cancer, somatic, 114500 (3); Colorectal cancer, hereditary nonpolyposis, type 7, 614385 (3) |
| *MLLT10* | 602409 | Leukemia, acute myeloid, 601626 (3), Somatic mutation, Autosomal dominant |
| *MLPH* | 606526 | Griscelli syndrome, type 3, 609227 (3), Autosomal recessive |
| *MLYCD* | 606761 | Malonyl-CoA decarboxylase deficiency, 248360 (3), Autosomal recessive |
| *MMAA* | 607481 | Methylmalonic aciduria, vitamin B12-responsive, cblA type, 251100 (3), Autosomal recessive |
| *MMAB* | 607568 | Methylmalonic aciduria, vitamin B12-responsive, cblB type, 251110 (3), Autosomal recessive |
| *MMACHC* | 609831 | Methylmalonic aciduria and homocystinuria, cblC type, 277400 (3), Autosomal recessive |
| *MMADHC* | 611935 | Methylmalonic aciduria, cblD type, variant 2, 277410 (3), Autosomal recessive; Methylmalonic aciduria and homocystinuria, cblD type, 277410 (3), Autosomal recessive; Homocystinuria, cblD type, variant 1, 277410 (3), Autosomal recessive |
| *MME* | 120520 | ?Spinocerebellar ataxia 43, 617018 (3), Autosomal dominant; Charcot-Marie-Tooth disease, axonal, type 2T, 617017 (3), Autosomal recessive, Autosomal dominant |
| *MMP1* | 120353 | COPD, rate of decline of lung function in, 606963 (3); {Epidermolysis bullosa dystrophica, autosomal recessive, modifier of}, 226600 (3), Autosomal recessive |
| *MMP13* | 600108 | ?Spondyloepimetaphyseal dysplasia, Missouri type, 602111 (3), Autosomal dominant; Metaphyseal anadysplasia 1, 602111 (3), Autosomal dominant; Metaphyseal dysplasia, Spahr type, 250400 (3), Autosomal recessive |
| *MMP14* | 600754 | ?Winchester syndrome, 277950 (3) |
| *MMP19* | 601807 | Cavitary optic disc anomalies, 611543 (3), Autosomal dominant |
| *MMP2* | 120360 | Multicentric osteolysis, nodulosis, and arthropathy, 259600 (3), Autosomal recessive |
| *MMP20* | 604629 | Amelogenesis imperfecta, type IIA2, 612529 (3), Autosomal recessive |
| *MMP21* | 608416 | Heterotaxy, visceral, 7, autosomal, 616749 (3), Autosomal recessive |
| *MMP3* | 185250 | {Coronary heart disease, susceptibility to, 6}, 614466 (3) |
| *MMP9* | 120361 | Metaphyseal anadysplasia 2, 613073 (3) |
| *MMUT (MUT)* | 609058 | Methylmalonic aciduria, mut(0) type, 251000 (3), Autosomal recessive |
| *MN1* | 156100 | CEBALID syndrome, 618774 (3), Autosomal dominant; Meningioma, 607174 (3), Autosomal dominant |
| *MNS1* | 610766 | Heterotaxy, visceral, 9, autosomal, with male infertility, 618948 (3), Autosomal recessive |
| *MNX1* | 142994 | Currarino syndrome, 176450 (3), Autosomal dominant |
| *MOCOS* | 613274 | Xanthinuria, type II, 603592 (3), Autosomal recessive |
| *MOCS1* | 603707 | Molybdenum cofactor deficiency A, 252150 (3), Autosomal recessive |
| *MOCS2* | 603708 | Molybdenum cofactor deficiency B, 252160 (3), Autosomal recessive |
| *MOG* | 159465 | ?Narcolepsy 7, 614250 (3), Autosomal dominant |
| *MOGS* | 601336 | Congenital disorder of glycosylation, type IIb, 606056 (3), Autosomal recessive |
| *MORC2* | 616661 | Charcot-Marie-Tooth disease, axonal, type 2Z, 616688 (3), Autosomal dominant; Developmental delay, impaired growth, dysmorphic facies, and axonal neuropathy, 619090 (3), Autosomal dominant |
| *MPC1* | 614738 | Mitochondrial pyruvate carrier deficiency, 614741 (3), Autosomal recessive |
| *MPDU1* | 604041 | Congenital disorder of glycosylation, type If, 609180 (3), Autosomal recessive |
| *MPDZ* | 603785 | Hydrocephalus, congenital, 2, with or without brain or eye anomalies, 615219 (3), Autosomal recessive |
| *MPEG1* | 610390 | Immunodeficiency 77, 619223 (3), Autosomal dominant |
| *MPI* | 154550 | Congenital disorder of glycosylation, type Ib, 602579 (3), Autosomal recessive |
| *MPIG6B* | 606520 | ?Thrombocytopenia, anemia, and myelofibrosis, 617441 (3), Autosomal recessive |
| *MPL* | 159530 | Myelofibrosis with myeloid metaplasia, somatic, 254450 (3); Thrombocythemia 2, 601977 (3), Somatic mutation, Autosomal dominant; Thrombocytopenia, congenital amegakaryocytic, 604498 (3), Autosomal recessive |
| *MPLKIP* | 609188 | Trichothiodystrophy 4, nonphotosensitive, 234050 (3), Autosomal recessive |
| *MPO* | 606989 | {Alzheimer disease, susceptibility to}, 104300 (3), Autosomal dominant; Myeloperoxidase deficiency, 254600 (3), Autosomal recessive; {Lung cancer, protection against, in smokers} (3) |
| *MPV17* | 137960 | Charcot-Marie-Tooth disease, axonal, type 2EE, 618400 (3), Autosomal recessive; Mitochondrial DNA depletion syndrome 6 (hepatocerebral type), 256810 (3), Autosomal recessive |
| *MPZ* | 159440 | Charcot-Marie-Tooth disease, type 2I, 607677 (3), Autosomal dominant; Dejerine-Sottas disease, 145900 (3), Autosomal recessive, Autosomal dominant; Charcot-Marie-Tooth disease, type 1B, 118200 (3), Autosomal dominant; Roussy-Levy syndrome, 180800 (3), Autosomal dominant; Charcot-Marie-Tooth disease, dominant intermediate D, 607791 (3), Autosomal dominant; Hypomyelinating neuropathy, congenital, 2, 618184 (3), Autosomal dominant; Charcot-Marie-Tooth disease, type 2J, 607736 (3), Autosomal dominant |
| *MPZL2* | 604873 | Deafness, autosomal recessive 111, 618145 (3), Autosomal recessive |
| *MRAP* | 609196 | Glucocorticoid deficiency 2, 607398 (3), Autosomal recessive |
| *MRAP2* | 615410 | {?Obesity, susceptibility to, BMIQ18}, 615457 (3), Autosomal dominant |
| *MRAS* | 608435 | Noonan syndrome 11, 618499 (3), Autosomal dominant |
| *MRE11* | 600814 | Ataxia-telangiectasia-like disorder 1, 604391 (3), Autosomal recessive |
| *MRM2* | 606906 | ?Mitochondrial DNA depletion syndrome 17, 618567 (3), Autosomal recessive |
| *MRPL12* | 602375 | ?Combined oxidative phosphorylation deficiency 45, 618951 (3), Autosomal recessive |
| *MRPL3* | 607118 | Combined oxidative phosphorylation deficiency 9, 614582 (3), Autosomal recessive |
| *MRPL44* | 611849 | ?Combined oxidative phosphorylation deficiency 16, 615395 (3), Autosomal recessive |
| *MRPS14* | 611978 | ?Combined oxidative phosphorylation deficiency 38, 618378 (3), Autosomal recessive |
| *MRPS16* | 609204 | Combined oxidative phosphorylation deficiency 2, 610498 (3), Autosomal recessive |
| *MRPS2* | 611971 | Combined oxidative phosphorylation deficiency 36, 617950 (3), Autosomal recessive |
| *MRPS22* | 605810 | Ovarian dysgenesis 7, 618117 (3), Autosomal recessive; Combined oxidative phosphorylation deficiency 5, 611719 (3), Autosomal recessive |
| *MRPS23* | 611985 | ?Combined oxidative phosphorylation defiency 46, 618952 (3), Autosomal recessive |
| *MRPS25* | 611987 | ?Combined oxidative phosphorylation deficiency 50, 619025 (3), Autosomal recessive |
| *MRPS28* | 611990 | ?Combined oxidative phosphorylation deficiency 47, 618958 (3), Autosomal recessive |
| *MRPS34* | 611994 | Combined oxidative phosphorylation deficiency 32, 617664 (3), Autosomal recessive |
| *MRPS7* | 611974 | ?Combined oxidative phosphorylation deficiency 34, 617872 (3), Autosomal recessive |
| *MRTFA* | 606078 | ?Immunodeficiency 66, 618847 (3), Autosomal recessive |
| *MS4A1* | 112210 | ?Immunodeficiency, common variable, 5, 613495 (3), Autosomal recessive |
| *MS4A2* | 147138 | {Atopy, susceptibility to}, 147050 (3), Autosomal dominant |
| *MSH2* | 609309 | Muir-Torre syndrome, 158320 (3), Autosomal dominant; Colorectal cancer, hereditary nonpolyposis, type 1, 120435 (3), Autosomal dominant; Mismatch repair cancer syndrome 2, 619096 (3), Autosomal recessive |
| *MSH3* | 600887 | Familial adenomatous polyposis 4, 617100 (3), Autosomal recessive; Endometrial carcinoma, somatic, 608089 (3) |
| *MSH5* | 603382 | ?Premature ovarian failure 13, 617442 (3), Autosomal recessive |
| *MSH6* | 600678 | Colorectal cancer, hereditary nonpolyposis, type 5, 614350 (3), Autosomal dominant; Mismatch repair cancer syndrome 3, 619097 (3), Autosomal recessive; {Endometrial cancer, familial}, 608089 (3), Somatic mutation, Autosomal dominant |
| *MSL3* | 300609 | Basilicata-Akhtar syndrome, 301032 (3), X-linked dominant |
| *MSMB* | 157145 | {Prostate cancer, hereditary, 13}, 611928 (3) |
| *MSMO1* | 607545 | Microcephaly, congenital cataract, and psoriasiform dermatitis, 616834 (3), Autosomal recessive |
| *MSN* | 309845 | Immunodeficiency 50, 300988 (3), X-linked recessive |
| *MSR1* | 153622 | Barrett esophagus/esophageal adenocarcinoma, 614266 (3) |
| *MSRB3* | 613719 | Deafness, autosomal recessive 74, 613718 (3), Autosomal recessive |
| *MST1R* | 600168 | {Nasopharyngeal carcinoma, susceptibility to, 3}, 617075 (3), Autosomal dominant |
| *MSTN* | 601788 | Muscle hypertrophy, 614160 (3) |
| *MSTO1* | 617619 | Myopathy, mitochondrial, and ataxia, 617675 (3), Autosomal recessive, Autosomal dominant |
| *MSX1* | 142983 | Tooth agenesis, selective, 1, with or without orofacial cleft, 106600 (3), Autosomal dominant; Ectodermal dysplasia 3, Witkop type, 189500 (3), Autosomal dominant; Orofacial cleft 5, 608874 (3), Autosomal dominant |
| *MSX2* | 123101 | Parietal foramina with cleidocranial dysplasia, 168550 (3), Autosomal dominant; Craniosynostosis 2, 604757 (3), Autosomal dominant; Parietal foramina 1, 168500 (3), Autosomal dominant |
| *MTAP* | 156540 | Diaphyseal medullary stenosis with malignant fibrous histiocytoma, 112250 (3), Autosomal dominant |
| *MTFMT* | 611766 | Combined oxidative phosphorylation deficiency 15, 614947 (3), Autosomal recessive; Mitochondrial complex I deficiency, nuclear type 27, 618248 (3), Autosomal recessive |
| *MTHFD1* | 172460 | {Neural tube defects, folate-sensitive, susceptibility to}, 601634 (3), Autosomal recessive; Combined immunodeficiency and megaloblastic anemia with or without hyperhomocysteinemia, 617780 (3), Autosomal recessive |
| *MTHFR* | 607093 | {Vascular disease, susceptibility to} (3); Homocystinuria due to MTHFR deficiency, 236250 (3), Autosomal recessive; {Thromboembolism, susceptibility to}, 188050 (3), Autosomal dominant; {Schizophrenia, susceptibility to}, 181500 (3), Autosomal dominant; {Neural tube defects, susceptibility to}, 601634 (3), Autosomal recessive |
| *MTHFS* | 604197 | Neurodevelopmental disorder with microcephaly, epilepsy, and hypomyelination, 618367 (3), Autosomal recessive |
| *MTM1* | 300415 | Myotubular myopathy, X-linked, 310400 (3), X-linked recessive |
| *MTMR14* | 611089 | {Centronuclear myopathy, autosomal, modifier of}, 160150 (3), Autosomal dominant |
| *MTMR2* | 603557 | Charcot-Marie-Tooth disease, type 4B1, 601382 (3), Autosomal recessive |
| *MTNR1B* | 600804 | {Diabetes mellitus, type 2, susceptibility to}, 125853 (3), Autosomal dominant |
| *MTO1* | 614667 | Combined oxidative phosphorylation deficiency 10, 614702 (3), Autosomal recessive |
| *MTOR* | 601231 | Focal cortical dysplasia, type II, somatic, 607341 (3); Smith-Kingsmore syndrome, 616638 (3), Autosomal dominant |
| *MTPAP* | 613669 | ?Spastic ataxia 4, autosomal recessive, 613672 (3), Autosomal recessive |
| *MTR* | 156570 | {Neural tube defects, folate-sensitive, susceptibility to}, 601634 (3), Autosomal recessive; Homocystinuria-megaloblastic anemia, cblG complementation type, 250940 (3), Autosomal recessive |
| *MTRFR (C12orf65)* | 613541 | Spastic paraplegia 55, autosomal recessive, 615035 (3), Autosomal recessive; Combined oxidative phosphorylation deficiency 7, 613559 (3), Autosomal recessive |
| *MTRR* | 602568 | Homocystinuria-megaloblastic anemia, cbl E type, 236270 (3), Autosomal recessive; {Neural tube defects, folate-sensitive, susceptibility to}, 601634 (3), Autosomal recessive |
| *MTTP* | 157147 | {Metabolic syndrome, protection against}, 605552 (3), Autosomal dominant; Abetalipoproteinemia, 200100 (3), Autosomal recessive |
| *MTX2* | 608555 | Mandibuloacral dysplasia progeroid syndrome, 619127 (3), Autosomal recessive |
| *MUC1* | 158340 | Tubulointerstitial kidney disease, autosomal dominant, 2, 174000 (3), Autosomal dominant |
| *MUC5B* | 600770 | {Pulmonary fibrosis, idiopathic, susceptibility to}, 178500 (3), Autosomal dominant |
| *MUC7* | 158375 | {Asthma, protection against}, 600807 (3), Autosomal dominant |
| *MUCL3* | 613928/604809 | -/Panbronchiolitis, diffuse, 604809 (2), Multifactorial |
| *MUSK* | 601296 | Fetal akinesia deformation sequence 1, 208150 (3), Autosomal recessive; Myasthenic syndrome, congenital, 9, associated with acetylcholine receptor deficiency, 616325 (3), Autosomal recessive |
| *MUTYH* | 604933 | Adenomas, multiple colorectal, 608456 (3), Autosomal recessive; Gastric cancer, somatic, 613659 (3) |
| *MVD* | 603236 | Porokeratosis 7, multiple types, 614714 (3), Autosomal dominant |
| *MVK* | 251170 | Hyper-IgD syndrome, 260920 (3), Autosomal recessive; Porokeratosis 3, multiple types, 175900 (3), Autosomal dominant; Mevalonic aciduria, 610377 (3), Autosomal recessive |
| *MVP* | 605088/157700 | -/Mitral valve prolapse, myxomatous 1, 157700 (2), Autosomal dominant |
| *MXI1* | 600020 | Prostate cancer, somatic, 176807 (3); Neurofibrosarcoma, somatic (3) |
| *MYB* | 189990 | {T-cell acute lymphoblastic leukemia} (3) |
| *MYBPC1* | 160794 | Myopathy, congenital, with tremor, 618524 (3), Autosomal dominant; Lethal congenital contracture syndrome 4, 614915 (3), Autosomal recessive; Arthrogryposis, distal, type 1B, 614335 (3), Autosomal dominant |
| *MYBPC3* | 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 |
| *MYC* | 190080 | Burkitt lymphoma, somatic, 113970 (3) |
| *MYCN* | 164840 | Feingold syndrome 1, 164280 (3), Autosomal dominant |
| *MYD88* | 602170 | Macroglobulinemia, Waldenstrom, somatic, 153600 (3); Immunodeficiency 68, 612260 (3), Autosomal recessive |
| *MYF5* | 159990 | Ophthalmoplegia, external, with rib and vertebral anomalies, 618155 (3), Autosomal recessive |
| *MYH10* | 160776 | MYH10-related Multiple congenital anomalies; Bilateral ventriculomegaly;aqueductal stenosis PMID: 25003005, 25356899, 24825879 |
| *MYH11* | 160745 | Megacystis-microcolon-intestinal hypoperistalsis syndrome 2, 619351 (3), Autosomal recessive; Aortic aneurysm, familial thoracic 4, 132900 (3), Autosomal dominant; Visceral myopathy 2, 619350 (3), Autosomal dominant |
| *MYH14* | 608568 | ?Peripheral neuropathy, myopathy, hoarseness, and hearing loss, 614369 (3), Autosomal dominant; Deafness, autosomal dominant 4A, 600652 (3), Autosomal dominant |
| *MYH2* | 160740 | Proximal myopathy and ophthalmoplegia, 605637 (3), Autosomal recessive, Autosomal dominant |
| *MYH3* | 160720 | Contractures, pterygia, and spondylocarpostarsal fusion syndrome 1A, 178110 (3), Autosomal dominant; Contractures, pterygia, and spondylocarpotarsal 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 |
| *MYH6* | 160710 | {Sick sinus syndrome 3}, 614090 (3); Atrial septal defect 3, 614089 (3); Cardiomyopathy, dilated, 1EE, 613252 (3), Autosomal dominant; Cardiomyopathy, hypertrophic, 14, 613251 (3), Autosomal dominant |
| *MYH7* | 160760 | Laing distal myopathy, 160500 (3), Autosomal dominant; Cardiomyopathy, hypertrophic, 1, 192600 (3), Digenic dominant, Autosomal dominant; Left ventricular noncompaction 5, 613426 (3), Autosomal dominant; Cardiomyopathy, dilated, 1S, 613426 (3), Autosomal dominant; Scapuloperoneal syndrome, myopathic type, 181430 (3), Autosomal dominant; Myopathy, myosin storage, autosomal dominant, 608358 (3), Autosomal dominant; Myopathy, myosin storage, autosomal recessive, 255160 (3), Autosomal recessive |
| *MYH8* | 160741 | Carney complex variant, 608837 (3); Trismus-pseudocamptodactyly syndrome, 158300 (3), Autosomal dominant |
| *MYH9* | 160775 | Macrothrombocytopenia and granulocyte inclusions with or without nephritis or sensorineural hearing loss, 155100 (3), Autosomal dominant; Deafness, autosomal dominant 17, 603622 (3), Autosomal dominant |
| *MYL1* | 160780 | Myopathy, congenital, with fast-twitch (type II) fiber atrophy, 618414 (3), Autosomal recessive |
| *MYL2* | 160781 | Cardiomyopathy, hypertrophic, 10, 608758 (3), Autosomal dominant |
| *MYL3* | 160790 | Cardiomyopathy, hypertrophic, 8, 608751 (3), Autosomal recessive, Autosomal dominant |
| *MYL4* | 160770 | ?Atrial fibrillation, familial, 18, 617280 (3), Autosomal dominant |
| *MYL9* | 609905 | ?Megacystis-microcolon-intestinal hypoperistalsis syndrome 4, 619365 (3), Autosomal recessive |
| *MYLK* | 600922 | Megacystis-microcolon-intestinal hypoperistalsis syndrome 1, 249210 (3), Autosomal recessive; Aortic aneurysm, familial thoracic 7, 613780 (3), Autosomal dominant |
| *MYLK2* | 606566 | Cardiomyopathy, hypertrophic, 1, digenic, 192600 (3), Digenic dominant, Autosomal dominant |
| *MYLPF* | 617378 | Arthrogryposis, distal, type 1C, 619110 (3), Autosomal recessive, Autosomal dominant |
| *MYMK* | 615345 | Carey-Fineman-Ziter syndrome, 254940 (3), Autosomal recessive |
| *MYO15A* | 602666 | Deafness, autosomal recessive 3, 600316 (3), Autosomal recessive |
| *MYO18B* | 607295 | Klippel-Feil syndrome 4, autosomal recessive, with myopathy and facial dysmorphism, 616549 (3), Autosomal recessive |
| *MYO1A* | 601478/607841 | -/Deafness, autosomal dominant 48, 607841 (2), Autosomal dominant |
| *MYO1E* | 601479 | Glomerulosclerosis, focal segmental, 6, 614131 (3), Autosomal recessive |
| *MYO3A* | 606808 | Deafness, autosomal recessive 30, 607101 (3), Autosomal recessive |
| *MYO5A* | 160777 | Griscelli syndrome, type 1, 214450 (3), Autosomal recessive |
| *MYO5B* | 606540 | Diarrhea 2, with microvillus atrophy, 251850 (3), Autosomal recessive |
| *MYO6* | 600970 | Deafness, autosomal dominant 22, with hypertrophic cardiomyopathy, 606346 (3), Autosomal dominant; Deafness, autosomal dominant 22, 606346 (3), Autosomal dominant; Deafness, autosomal recessive 37, 607821 (3), Autosomal recessive |
| *MYO7A* | 276903 | Deafness, autosomal recessive 2, 600060 (3), Autosomal recessive; Usher syndrome, type 1B, 276900 (3), Autosomal recessive; Deafness, autosomal dominant 11, 601317 (3), Autosomal dominant |
| *MYO9A* | 604875 | Myasthenic syndrome, congenital, 24, presynaptic, 618198 (3), Autosomal recessive |
| *MYO9B* | 602129 | {Celiac disease, susceptibility to, 4}, 609753 (3) |
| *MYOC* | 601652 | Glaucoma 1A, primary open angle, 137750 (3), Autosomal dominant |
| *MYOCD* | 606127 | Megabladder, congenital, 618719 (3), Autosomal dominant |
| *MYOD1* | 159970 | Myopathy, congenital, with diaphragmatic defects, respiratory insufficiency, and dysmorphic facies, 618975 (3), Autosomal recessive |
| *MYOF* | 604603 | ?Angioedema, hereditary, 7, 619366 (3), Autosomal dominant |
| *MYORG* | 618255 | Basal ganglia calcification, idiopathic, 7, autosomal recessive, 618317 (3), Autosomal recessive |
| *MYOT* | 604103 | Myopathy, myofibrillar, 3, 609200 (3), Autosomal dominant; Myopathy, spheroid body, 182920 (3), Autosomal dominant |
| *MYOZ2* | 605602 | Cardiomyopathy, hypertrophic, 16, 613838 (3), Autosomal dominant |
| *MYPN* | 608517 | Cardiomyopathy, hypertrophic, 22, 615248 (3), Autosomal dominant; Cardiomyopathy, familial restrictive, 4, 615248 (3), Autosomal dominant; Cardiomyopathy, dilated, 1KK, 615248 (3), Autosomal dominant; Nemaline myopathy 11, autosomal recessive, 617336 (3), Autosomal recessive |
| *MYRF* | 608329 | Encephalitis/encephalopathy, mild, with reversible myelin vacuolization, 618113 (3), Autosomal dominant; Cardiac-urogenital syndrome, 618280 (3), Autosomal dominant |
| *MYSM1* | 612176 | Bone marrow failure syndrome 4, 618116 (3), Autosomal recessive |
| *MYT1* | 600379 | Oculo-auriculo-vertebral spectrum (OAVS);OAVS/Goldenhar syndrome PMID: 28612832, 27358179, 32871052 |
| *MYT1L* | 613084 | Mental retardation, autosomal dominant 39, 616521 (3), Autosomal dominant |
| *NAA10* | 300013 | Microphthalmia, syndromic 1, 309800 (3), X-linked; Ogden syndrome, 300855 (3), X-linked dominant, X-linked recessive |
| *NAA15* | 608000 | Intellectual developmental disorder, autosomal dominant 50, with behavioral abnormalities, 617787 (3), Autosomal dominant |
| *NACC1* | 610672 | Neurodevelopmental disorder with epilepsy, cataracts, feeding difficulties, and delayed brain myelination, 617393 (3), Autosomal dominant |
| *NADK2* | 615787 | 2,4-dienoyl-CoA reductase deficiency, 616034 (3), Autosomal recessive |
| *NADSYN1* | 608285 | Vertebral, cardiac, renal, and limb defects syndrome 3, 618845 (3), Autosomal recessive |
| *NAGA* | 104170 | Schindler disease, type I, 609241 (3), Autosomal recessive; Kanzaki disease, 609242 (3), Autosomal recessive; Schindler disease, type III, 609241 (3), Autosomal recessive |
| *NAGLU* | 609701 | ?Charcot-Marie-Tooth disease, axonal, type 2V, 616491 (3), Autosomal dominant; Mucopolysaccharidosis type IIIB (Sanfilippo B), 252920 (3), Autosomal recessive |
| *NAGS* | 608300 | N-acetylglutamate synthase deficiency, 237310 (3), Autosomal recessive |
| *NALCN* | 611549 | Congenital contractures of the limbs and face, hypotonia, and developmental delay, 616266 (3), Autosomal dominant; Hypotonia, infantile, with psychomotor retardation and characteristic facies 1, 615419 (3), Autosomal recessive |
| *NANOS1* | 608226 | Spermatogenic failure 12, 615413 (3), Autosomal dominant |
| *NANS* | 605202 | Spondyloepimetaphyseal dysplasia, Camera-Genevieve type, 610442 (3), Autosomal recessive |
| *NARS1 (NARS)* | 108410 | Neurodevelopmental disorder with microcephaly, impaired language, epilepsy, and gait abnormalities, autosomal dominant, 619092 (3); Neurodevelopmental disorder with microcephaly, impaired language, and gait abnormalities, autosomal recessive, 619091 (3), Autosomal recessive |
| *NARS2* | 612803 | Combined oxidative phosphorylation deficiency 24, 616239 (3), Autosomal recessive; ?Deafness, autosomal recessive 94, 618434 (3), Autosomal recessive |
| *NAT2* | 612182 | [Acetylation, slow], 243400 (3), Autosomal recessive |
| *NAT8L* | 610647 | ?N-acetylaspartate deficiency, 614063 (3), Autosomal recessive |
| *NAXD* | 615910 | Encephalopathy, progressive, early-onset, with brain edema and/or leukoencephalopathy, 2, 618321 (3), Autosomal recessive |
| *NAXE* | 608862 | Encephalopathy, progressive, early-onset, with brain edema and/or leukoencephalopathy, 617186 (3), Autosomal recessive |
| *NBAS* | 608025 | Short stature, optic nerve atrophy, and Pelger-Huet anomaly, 614800 (3), Autosomal recessive; Infantile liver failure syndrome 2, 616483 (3), Autosomal recessive |
| *NBEA* | 604889 | Neurodevelopmental disorder with or without early-onset generalized epilepsy, 619157 (3), Autosomal dominant |
| *NBEAL2* | 614169 | Gray platelet syndrome, 139090 (3), Autosomal recessive |
| *NBN* | 602667 | Leukemia, acute lymphoblastic, 613065 (3); Aplastic anemia, 609135 (3); Nijmegen breakage syndrome, 251260 (3), Autosomal recessive |
| *NCAPD2* | 615638 | ?Microcephaly 21, primary, autosomal recessive, 617983 (3), Autosomal recessive |
| *NCAPD3* | 609276 | Microcephaly 22, primary, autosomal recessive, 617984 (3), Autosomal recessive |
| *NCAPG2* | 608532 | Khan-Khan-Katsanis syndrome, 618460 (3), Autosomal recessive |
| *NCAPH* | 602332 | ?Microcephaly 23, primary, autosomal recessive, 617985 (3), Autosomal recessive |
| *NCDN* | 608458 | Neurodevelopmental disorder with infantile epileptic spasms, 619373 (3), Autosomal dominant |
| *NCF1* | 608512 | Chronic granulomatous disease 1, autosomal recessive, 233700 (3), Autosomal recessive |
| *NCF2* | 608515 | Chronic granulomatous disease 2, autosomal recessive, 233710 (3), Autosomal recessive |
| *NCF4* | 601488 | Chronic granulomatous disease 3, autosomal recessive, 613960 (3), Autosomal recessive |
| *NCKAP1L* | 141180 | Immunodeficiency 72 with autoinflammation, 618982 (3), Autosomal recessive |
| *NCR3* | 611550 | {Malaria, mild, susceptibility to}, 609148 (3) |
| *NCSTN* | 605254 | Acne inversa, familial, 1, 142690 (3), Autosomal dominant |
| *NDE1* | 609449 | Lissencephaly 4 (with microcephaly), 614019 (3), Autosomal recessive; ?Microhydranencephaly, 605013 (3), Autosomal recessive |
| *NDN* | 602117 | Prader-Willi syndrome, 176270 (3), Autosomal dominant |
| *NDNF* | 616506 | Hypogonadotropic hypogonadism 25 with anosmia, 618841 (3), Autosomal dominant |
| *NDP* | 300658 | Exudative vitreoretinopathy 2, X-linked, 305390 (3), X-linked dominant, X-linked recessive; Norrie disease, 310600 (3), X-linked recessive |
| *NDRG1* | 605262 | Charcot-Marie-Tooth disease, type 4D, 601455 (3), Autosomal recessive |
| *NDST1* | 600853 | Mental retardation, autosomal recessive 46, 616116 (3), Autosomal recessive |
| *NDUFA1* | 300078 | Mitochondrial complex I deficiency, nuclear type 12, 301020 (3), X-linked recessive |
| *NDUFA10* | 603835 | Mitochondrial complex I deficiency, nuclear type 22, 618243 (3), Autosomal recessive |
| *NDUFA11* | 612638 | Mitochondrial complex I deficiency, nuclear type 14, 618236 (3), Autosomal recessive |
| *NDUFA12* | 614530 | ?Mitochondrial complex I deficiency, nuclear type 23, 618244 (3), Autosomal recessive |
| *NDUFA13* | 609435 | {Thyroid carcinoma, Hurthle cell}, 607464 (3); ?Mitochondrial complex I deficiency, nuclear type 28, 618249 (3), Autosomal recessive |
| *NDUFA2* | 602137 | Mitochondrial complex I deficiency, nuclear type 13, 618235 (3), Autosomal recessive |
| *NDUFA4* | 603833 | ?Mitochondrial complex IV deficiency, nuclear type 21, 619065 (3), Autosomal recessive |
| *NDUFA6* | 602138 | Mitochondrial complex I deficiency, nuclear type 33, 618253 (3), Autosomal recessive |
| *NDUFA8* | 603359 | Mitochondrial complex I deficiency, nuclear type 37, 619272 (3), Autosomal recessive |
| *NDUFA9* | 603834 | Mitochondrial complex I deficiency, nuclear type 26, 618247 (3), Autosomal recessive |
| *NDUFAF1* | 606934 | Mitochondrial complex I deficiency, nuclear type 11, 618234 (3), Autosomal recessive |
| *NDUFAF2* | 609653 | Mitochondrial complex I deficiency, nuclear type 10, 618233 (3), Autosomal recessive |
| *NDUFAF3* | 612911 | Mitochondrial complex I deficiency, nuclear type 18, 618240 (3), Autosomal recessive |
| *NDUFAF4* | 611776 | Mitochondrial complex I deficiency, nuclear type 15, 618237 (3), Autosomal recessive |
| *NDUFAF5* | 612360 | Mitochondrial complex I deficiency, nuclear type 16, 618238 (3), Autosomal recessive |
| *NDUFAF6* | 612392 | Mitochondrial complex I deficiency, nuclear type 17, 618239 (3), Autosomal recessive; Fanconi renotubular syndrome 5, 618913 (3), Autosomal recessive |
| *NDUFAF8* | 618461 | Mitochondrial complex I deficiency, nuclear type 34, 618776 (3), Autosomal recessive |
| *NDUFB10* | 603843 | ?Mitochondrial complex I deficiency, nuclear type 35, 619003 (3), Autosomal recessive |
| *NDUFB11* | 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 |
| *NDUFB3* | 603839 | Mitochondrial complex I deficiency, nuclear type 25, 618246 (3), Autosomal recessive |
| *NDUFB8* | 602140 | Mitochondrial complex I deficiency, nuclear type 32, 618252 (3), Autosomal recessive |
| *NDUFB9* | 601445 | ?Mitochondrial complex I deficiency, nuclear type 24, 618245 (3), Autosomal recessive |
| *NDUFC2* | 603845 | Mitochondrial complex I deficiency, nuclear type 36, 619170 (3), Autosomal recessive |
| *NDUFS1* | 157655 | Mitochondrial complex I deficiency, nuclear type 5, 618226 (3), Autosomal recessive |
| *NDUFS2* | 602985 | Mitochondrial complex I deficiency, nuclear type 6, 618228 (3), Autosomal recessive |
| *NDUFS3* | 603846 | Mitochondrial complex I deficiency, nuclear type 8, 618230 (3), Autosomal recessive |
| *NDUFS4* | 602694 | Mitochondrial complex I deficiency, nuclear type 1, 252010 (3), Autosomal recessive |
| *NDUFS6* | 603848 | Mitochondrial complex I deficiency, nuclear type 9, 618232 (3), Autosomal recessive |
| *NDUFS7* | 601825 | Mitochondrial complex I deficiency, nuclear type 3, 618224 (3), Autosomal recessive |
| *NDUFS8* | 602141 | Mitochondrial complex I deficiency, nuclear type 2, 618222 (3), Autosomal recessive |
| *NDUFV1* | 161015 | Mitochondrial complex I deficiency, nuclear type 4, 618225 (3), Autosomal recessive |
| *NDUFV2* | 600532 | Mitochondrial complex I deficiency, nuclear type 7, 618229 (3), Autosomal recessive |
| *NEB* | 161650 | Nemaline myopathy 2, autosomal recessive, 256030 (3), Autosomal recessive; Arthrogryposis multiplex congenita 6, 619334 (3), Autosomal recessive |
| *NECAP1* | 611623 | Developmental and epileptic encephalopathy 21, 615833 (3), Autosomal recessive |
| *NECTIN1* | 600644 | Cleft lip/palate-ectodermal dysplasia syndrome, 225060 (3), Autosomal recessive; Orofacial cleft 7, 225060 (3), Autosomal recessive |
| *NECTIN4* | 609607 | Ectodermal dysplasia-syndactyly syndrome 1, 613573 (3), Autosomal recessive |
| *NEDD4L* | 606384 | Periventricular nodular heterotopia 7, 617201 (3), Autosomal dominant |
| *NEFH* | 162230 | ?{Amyotrophic lateral sclerosis, susceptibility to}, 105400 (3), Autosomal recessive, Autosomal dominant; Charcot-Marie-Tooth disease, axonal, type 2CC, 616924 (3), Autosomal dominant |
| *NEFL* | 162280 | Charcot-Marie-Tooth disease, 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 |
| *NEK1* | 604588 | Short-rib thoracic dysplasia 6 with or without polydactyly, 263520 (3), Autosomal recessive, Digenic recessive; {Amyotrophic lateral sclerosis, susceptibility to, 24}, 617892 (3), Autosomal dominant |
| *NEK10* | 618726 | Ciliary dyskinesia, primary, 44, 618781 (3), Autosomal recessive |
| *NEK2* | 604043 | ?Retinitis pigmentosa 67, 615565 (3), Autosomal recessive |
| *NEK8* | 609799 | Renal-hepatic-pancreatic dysplasia 2, 615415 (3), Autosomal recessive; ?Nephronophthisis 9, 613824 (3) |
| *NEK9* | 609798 | ?Arthrogryposis, Perthes disease, and upward gaze palsy, 614262 (3), Autosomal recessive; Nevus comedonicus, somatic, 617025 (3); Lethal congenital contracture syndrome 10, 617022 (3), Autosomal recessive |
| *NEMF* | 608378 | Intellectual developmental disorder with speech delay and axonal peripheral neuropathy, 619099 (3), Autosomal recessive |
| *NEPRO* | 617089 | Anauxetic dysplasia 3, 618853 (3), Autosomal recessive |
| *NEU1* | 608272 | Sialidosis, type II, 256550 (3), Autosomal recessive; Sialidosis, type I, 256550 (3), Autosomal recessive |
| *NEUROD1* | 601724 | {Type 2 diabetes mellitus, susceptibility to}, 125853 (3), Autosomal dominant; Maturity-onset diabetes of the young 6, 606394 (3) |
| *NEUROD2* | 601725 | Developmental and epileptic encephalopathy 72, 618374 (3), Autosomal dominant |
| *NEUROG3* | 604882 | Diarrhea 4, malabsorptive, congenital, 610370 (3), Autosomal recessive |
| *NEXMIF* | 300524 | Mental retardation, X-linked 98, 300912 (3), X-linked dominant |
| *NEXN* | 613121 | Cardiomyopathy, dilated, 1CC, 613122 (3), Autosomal dominant; Cardiomyopathy, hypertrophic, 20, 613876 (3), Autosomal dominant |
| *NF1* | 613113 | Watson syndrome, 193520 (3), Autosomal dominant; Leukemia, juvenile myelomonocytic, 607785 (3), Somatic mutation, Autosomal dominant; Neurofibromatosis, familial spinal, 162210 (3), Autosomal dominant; Neurofibromatosis, type 1, 162200 (3), Autosomal dominant; Neurofibromatosis-Noonan syndrome, 601321 (3), Autosomal dominant |
| *NF2* | 607379 | Neurofibromatosis, type 2, 101000 (3), Autosomal dominant; Meningioma, NF2-related, somatic, 607174 (3); Schwannomatosis, somatic, 162091 (3) |
| *NFASC* | 609145 | Neurodevelopmental disorder with central and peripheral motor dysfunction, 618356 (3), Autosomal recessive |
| *NFE2L2* | 600492 | Immunodeficiency, developmental delay, and hypohomocysteinemia, 617744 (3), Autosomal dominant |
| *NFIA* | 600727 | Brain malformations with or without urinary tract defects, 613735 (3), Autosomal dominant |
| *NFIB* | 600728 | Macrocephaly, acquired, with impaired intellectual development, 618286 (3), Autosomal dominant |
| *NFIX* | 164005 | Marshall-Smith syndrome, 602535 (3), Autosomal dominant; Sotos syndrome 2, 614753 (3), Autosomal dominant |
| *NFKB1* | 164011 | Immunodeficiency, common variable, 12, 616576 (3), Autosomal dominant |
| *NFKB2* | 164012 | Immunodeficiency, common variable, 10, 615577 (3), Autosomal dominant |
| *NFKBIA* | 164008 | Ectodermal dysplasia and immunodeficiency 2, 612132 (3), Autosomal dominant |
| *NFKBIL1* | 601022 | {Rheumatoid arthritis, susceptibility to}, 180300 (3) |
| *NFS1* | 603485 | Combined oxidative phosphorylation deficiency 52, 619386 (3), Autosomal recessive |
| *NFU1* | 608100 | Multiple mitochondrial dysfunctions syndrome 1, 605711 (3), Autosomal recessive |
| *NGF* | 162030 | Neuropathy, hereditary sensory and autonomic, type V, 608654 (3), Autosomal recessive |
| *NGLY1* | 610661 | Congenital disorder of deglycosylation, 615273 (3), Autosomal recessive |
| *NHEJ1* | 611290 | Severe combined immunodeficiency with microcephaly, growth retardation, and sensitivity to ionizing radiation, 611291 (3) |
| *NHLRC1* | 608072 | Epilepsy, progressive myoclonic 2B (Lafora), 254780 (3), Autosomal recessive |
| *NHLRC2* | 618277 | FINCA syndrome, 618278 (3), Autosomal recessive |
| *NHP2* | 606470 | Dyskeratosis congenita, autosomal recessive 2, 613987 (3), Autosomal recessive |
| *NHS* | 300457 | Cataract 40, X-linked, 302200 (3), X-linked; Nance-Horan syndrome, 302350 (3), X-linked dominant |
| *NIN* | 608684 | ?Seckel syndrome 7, 614851 (3), Autosomal recessive |
| *NIPA1* | 608145 | Spastic paraplegia 6, autosomal dominant, 600363 (3), Autosomal dominant |
| *NIPAL4* | 609383 | Ichthyosis, congenital, autosomal recessive 6, 612281 (3), Autosomal recessive |
| *NIPBL* | 608667 | Cornelia de Lange syndrome 1, 122470 (3), Autosomal dominant |
| *NKAP* | 300766 | Intellectual developmental disorder, X-linked, syndromic, Hackman-Di Donato type, 301039 (3), X-linked recessive |
| *NKX2-1* | 600635 | Chorea, hereditary benign, 118700 (3), Autosomal dominant; {Thyroid cancer, nonmedullary, 1}, 188550 (3), Autosomal dominant; Choreoathetosis, hypothyroidism, and neonatal respiratory distress, 610978 (3), Autosomal dominant |
| *NKX2-5* | 600584 | Hypoplastic left heart syndrome 2, 614435 (3), Autosomal dominant; Tetralogy of Fallot, 187500 (3), Autosomal dominant; Hypothyroidism, congenital nongoitrous, 5, 225250 (3), Autosomal dominant; Conotruncal heart malformations, variable, 217095 (3); Ventricular septal defect 3, 614432 (3), Autosomal dominant; Atrial septal defect 7, with or without AV conduction defects, 108900 (3), Autosomal dominant |
| *NKX2-6* | 611770 | Persistent truncus arteriosus, 217095 (3); Conotruncal heart malformations, 217095 (3) |
| *NKX3-2* | 602183 | Spondylo-megaepiphyseal-metaphyseal dysplasia, 613330 (3), Autosomal recessive |
| *NKX6-2* | 605955 | Spastic ataxia 8, autosomal recessive, with hypomyelinating leukodystrophy, 617560 (3), Autosomal recessive |
| *NLGN1* | 600568 | {Autism, susceptibility to, 20}, 618830 (3), Autosomal dominant |
| *NLGN3* | 300336 | {Asperger syndrome susceptibility, X-linked 1}, 300494 (3), Multifactorial, Isolated cases, X-linked; {Autism susceptibility, X-linked 1}, 300425 (3), X-linked |
| *NLGN4X* | 300427 | {Asperger syndrome susceptibility, X-linked 2}, 300497 (3), X-linked; Mental retardation, X-linked, 300495 (3), Multifactorial, Isolated cases, X-linked; {Autism susceptibility, X-linked 2}, 300495 (3), Multifactorial, Isolated cases, X-linked |
| *NLRC4* | 606831 | ?Familial cold autoinflammatory syndrome 4, 616115 (3), Autosomal dominant; Autoinflammation with infantile enterocolitis, 616050 (3), Autosomal dominant |
| *NLRP1* | 606636 | {Vitiligo-associated multiple autoimmune disease susceptibility 1}, 606579 (3); ?Respiratory papillomatosis, juvenile recurrent, congenital, 618803 (3), Autosomal recessive; Autoinflammation with arthritis and dyskeratosis, 617388 (3), Autosomal recessive, Autosomal dominant; Palmoplantar carcinoma, multiple self-healing, 615225 (3), Autosomal dominant |
| *NLRP12* | 609648 | Familial cold autoinflammatory syndrome 2, 611762 (3), Autosomal dominant |
| *NLRP3* | 606416 | CINCA syndrome, 607115 (3), Autosomal dominant; Familial cold inflammatory syndrome 1, 120100 (3), Autosomal dominant; Keratoendothelitis fugax hereditaria, 148200 (3), Autosomal dominant; Deafness, autosomal dominant 34, with or without inflammation, 617772 (3), Autosomal dominant; Muckle-Wells syndrome, 191900 (3), Autosomal dominant |
| *NLRP7* | 609661 | Hydatidiform mole, recurrent, 1, 231090 (3), Autosomal recessive |
| *NME8* | 607421 | Ciliary dyskinesia, primary, 6, 610852 (3), Autosomal recessive |
| *NMNAT1* | 608700 | Spondyloepiphyseal dysplasia, sensorineural hearing loss, intellectual developmental disorder, and Leber congenital amaurosis, 619260 (3), Autosomal recessive; Leber congenital amaurosis 9, 608553 (3), Autosomal recessive |
| *NMNAT2* | 608701 | Neuropathy PMID: 31132363, 33248200 |
| *NNMT* | 600008 | Homocysteine plasma level, 600008 (2) |
| *NNT* | 607878 | Glucocorticoid deficiency 4, with or without mineralocorticoid deficiency, 614736 (3), Autosomal recessive |
| *NOBOX* | 610934 | Premature ovarian failure 5, 611548 (3), Autosomal dominant |
| *NOD2* | 605956 | Blau syndrome, 186580 (3), Autosomal dominant; {Yao syndrome}, 617321 (3), Multifactorial; {Inflammatory bowel disease 1, Crohn disease}, 266600 (3), Multifactorial |
| *NODAL* | 601265 | Heterotaxy, visceral, 5, 270100 (3), Autosomal dominant |
| *NOG* | 602991 | Symphalangism, proximal, 1A, 185800 (3), Autosomal dominant; Brachydactyly, type B2, 611377 (3), Autosomal dominant; Stapes ankylosis with broad thumbs and toes, 184460 (3), Autosomal dominant; Tarsal-carpal coalition syndrome, 186570 (3), Autosomal dominant; Multiple synostoses syndrome 1, 186500 (3), Autosomal dominant |
| *NOL3* | 605235 | ?Myoclonus, familial, 1, 614937 (3), Autosomal dominant |
| *NONO* | 300084 | Mental retardation, X-linked, syndromic 34, 300967 (3), X-linked |
| *NOP10* | 606471 | Dyskeratosis congenita, autosomal recessive 1, 224230 (3), Autosomal recessive |
| *NOP56* | 614154 | Spinocerebellar ataxia 36, 614153 (3), Autosomal dominant |
| *NOS1AP* | 605551 | Nephrotic syndrome, type 22, 619155 (3), Autosomal recessive |
| *NOS2* | 163730 | {Malaria, resistance to}, 611162 (3) |
| *NOS3* | 163729 | {Coronary artery spasm 1, susceptibility to} (3); {Hypertension, susceptibility to}, 145500 (3), Multifactorial; {Placental abruption} (3); {Alzheimer disease, late-onset, susceptibility to}, 104300 (3), Autosomal dominant; {Hypertension, pregnancy-induced}, 189800 (3), Autosomal dominant; {Ischemic stroke, susceptibility to}, 601367 (3), Multifactorial |
| *NOTCH1* | 190198 | Adams-Oliver syndrome 5, 616028 (3), Autosomal dominant; Aortic valve disease 1, 109730 (3), Autosomal dominant |
| *NOTCH2* | 600275 | Alagille syndrome 2, 610205 (3), Autosomal dominant; Hajdu-Cheney syndrome, 102500 (3), Autosomal dominant |
| *NOTCH3* | 600276 | Lateral meningocele syndrome, 130720 (3), Autosomal dominant; ?Myofibromatosis, infantile 2, 615293 (3), Autosomal dominant; Cerebral arteriopathy with subcortical infarcts and leukoencephalopathy 1, 125310 (3), Autosomal dominant |
| *NOVA2* | 601991 | Neurodevelopmental disorder with or without autistic features and/or structural brain abnormalities, 618859 (3), Autosomal dominant |
| *NPC1* | 607623 | Niemann-Pick disease, type C1, 257220 (3), Autosomal recessive; Niemann-Pick disease, type D, 257220 (3), Autosomal recessive |
| *NPC1L1* | 608010 | [Ezetimibe, nonresponse to], 617966 (3); [Low density lipoprotein cholesterol level QTL 7], 617966 (3) |
| *NPC2* | 601015 | Niemann-pick disease, type C2, 607625 (3), Autosomal recessive |
| *NPHP1* | 607100 | Joubert syndrome 4, 609583 (3), Autosomal recessive; Nephronophthisis 1, juvenile, 256100 (3), Autosomal recessive; Senior-Loken syndrome-1, 266900 (3), Autosomal recessive |
| *NPHP3* | 608002 | Nephronophthisis 3, 604387 (3), Autosomal recessive; Renal-hepatic-pancreatic dysplasia 1, 208540 (3), Autosomal recessive; Meckel syndrome 7, 267010 (3), Autosomal recessive |
| *NPHP4* | 607215 | Senior-Loken syndrome 4, 606996 (3), Autosomal recessive; Nephronophthisis 4, 606966 (3), Autosomal recessive |
| *NPHS1* | 602716 | Nephrotic syndrome, type 1, 256300 (3), Autosomal recessive |
| *NPHS2* | 604766 | Nephrotic syndrome, type 2, 600995 (3), Autosomal recessive |
| *NPM1* | 164040 | Leukemia, acute myeloid, somatic, 601626 (3) |
| *NPPA* | 108780 | Atrial standstill 2, 615745 (3), Autosomal recessive; Atrial fibrillation, familial, 6, 612201 (3), Autosomal dominant |
| *NPR2* | 108961 | Acromesomelic dysplasia, Maroteaux type, 602875 (3), Autosomal recessive; Epiphyseal chondrodysplasia, Miura type, 615923 (3), Autosomal dominant; Short stature with nonspecific skeletal abnormalities, 616255 (3), Autosomal dominant |
| *NPR3* | 108962 | ?Hypertension, salt-resistant (1) |
| *NPRL2* | 607072 | Epilepsy, familial focal, with variable foci 2, 617116 (3), Autosomal dominant |
| *NPRL3* | 600928 | Epilepsy, familial focal, with variable foci 3, 617118 (3), Autosomal dominant |
| *NPSR1* | 608595 | {Asthma, susceptibility to, 2}, 608584 (3) |
| *NQO1* | 125860 | {Breast cancer, poor survival after chemotherapy for} (3); {Leukemia, post-chemotherapy, susceptibility to} (3); {Benzene toxicity, susceptibility to} (3) |
| *NQO2* | 160998 | {?Breast cancer susceptibility}, 114480 (1), Somatic mutation, Autosomal dominant |
| *NR0B1* | 300473 | Adrenal hypoplasia, congenital, 300200 (3), X-linked recessive; 46XY sex reversal 2, dosage-sensitive, 300018 (3), X-linked |
| *NR0B2* | 604630 | Obesity, mild, early-onset, 601665 (3), Multifactorial, Autosomal recessive, Autosomal dominant |
| *NR1H4* | 603826 | Cholestasis, progressive familial intrahepatic, 5, 617049 (3), Autosomal recessive |
| *NR2E3* | 604485 | Retinitis pigmentosa 37, 611131 (3), Autosomal recessive, Autosomal dominant; Enhanced S-cone syndrome, 268100 (3), Autosomal recessive |
| *NR2F1* | 132890 | Bosch-Boonstra-Schaaf optic atrophy syndrome, 615722 (3), Autosomal dominant |
| *NR2F2* | 107773 | 46,XX sex reversal 5, 618901 (3), Autosomal dominant; Congenital heart defects, multiple types, 4, 615779 (3), Autosomal dominant |
| *NR3C1* | 138040 | Glucocorticoid resistance, 615962 (3), Autosomal dominant |
| *NR3C2* | 600983 | Pseudohypoaldosteronism type I, autosomal dominant, 177735 (3), Autosomal dominant; Hypertension, early-onset, autosomal dominant, with exacerbation in pregnancy, 605115 (3) |
| *NR4A3* | 600542 | Chondrosarcoma, extraskeletal myxoid, 612237 (3) |
| *NR5A1* | 184757 | 46, XX sex reversal 4, 617480 (3), Autosomal dominant; Premature ovarian failure 7, 612964 (3), Autosomal dominant; 46XY sex reversal 3, 612965 (3), Autosomal dominant; Adrenocortical insufficiency, 612964 (3), Autosomal dominant; Spermatogenic failure 8, 613957 (3), Autosomal dominant |
| *NRAS* | 164790 | Noonan syndrome 6, 613224 (3), Autosomal dominant; ?RAS-associated autoimmune lymphoproliferative syndrome type IV, somatic, 614470 (3); Melanocytic nevus syndrome, congenital, somatic, 137550 (3); Epidermal nevus, somatic, 162900 (3); Schimmelpenning-Feuerstein-Mims syndrome, somatic mosaic, 163200 (3); Thyroid carcinoma, follicular, somatic, 188470 (3); Neurocutaneous melanosis, somatic, 249400 (3); Colorectal cancer, somatic, 114500 (3) |
| *NRG1* | 142445 | {?Schizophrenia, susceptibility to}, 603013 (1) |
| *NRIP1* | 602490 | ?Congenital anomalies of kidney and urinary tract 3, 618270 (3), Autosomal dominant |
| *NRL* | 162080 | Retinitis pigmentosa 27, 613750 (3), Autosomal dominant; Retinal degeneration, autosomal recessive, clumped pigment type (3) |
| *NRROS* | 615322 | Seizures, early-onset, with neurodegeneration and brain calcification, 618875 (3), Autosomal recessive |
| *NRXN1* | 600565 | Pitt-Hopkins-like syndrome 2, 614325 (3), Autosomal recessive; {Schizophrenia, susceptibility to, 17}, 614332 (3) |
| *NSD1* | 606681 | Sotos syndrome 1, 117550 (3), Autosomal dominant |
| *NSDHL* | 300275 | CK syndrome, 300831 (3), X-linked recessive; CHILD syndrome, 308050 (3), X-linked dominant |
| *NSF* | 601633 | Developmental and epileptic encephalopathy 96, 619340 (3), Autosomal dominant |
| *NSMCE2* | 617246 | Seckel syndrome 10, 617253 (3), Autosomal recessive |
| *NSMCE3* | 608243 | Lung disease, immunodeficiency, and chromosome breakage syndrome, 617241 (3), Autosomal recessive |
| *NSMF* | 608137 | Hypogonadotropic hypogonadism 9 with or without anosmia, 614838 (3), Autosomal dominant |
| *NSUN2* | 610916 | Mental retardation, autosomal recessive 5, 611091 (3), Autosomal recessive |
| *NSUN3* | 617491 | Combined oxidative phosphorylation deficiency 48, 619012 (3) |
| *NT5C2* | 600417 | Spastic paraplegia 45, autosomal recessive, 613162 (3), Autosomal recessive |
| *NT5C3A* | 606224 | Anemia, hemolytic, due to UMPH1 deficiency, 266120 (3), Autosomal recessive |
| *NT5E* | 129190 | Calcification of joints and arteries, 211800 (3), Autosomal recessive |
| *NTF4* | 162662 | Glaucoma 1, open angle, 1O, 613100 (3) |
| *NTHL1* | 602656 | Familial adenomatous polyposis 3, 616415 (3), Autosomal recessive |
| *NTN1* | 601614 | Mirror movements 4, 618264 (3), Autosomal dominant |
| *NTNG2* | 618689 | Neurodevelopmental disorder with behavioral abnormalities, absent speech, and hypotonia, 618718 (3), Autosomal recessive |
| *NTRK1* | 191315 | Insensitivity to pain, congenital, with anhidrosis, 256800 (3), Autosomal recessive |
| *NTRK2* | 600456 | Developmental and epileptic encephalopathy 58, 617830 (3), Autosomal dominant; Obesity, hyperphagia, and developmental delay, 613886 (3), Autosomal dominant |
| *NUAK2* | 608131 | ?Anencephaly 2, 619452 (3), Autosomal recessive |
| *NUBPL* | 613621 | Mitochondrial complex I deficiency, nuclear type 21, 618242 (3), Autosomal recessive |
| *NUDT15* | 615792 | {Thiopurines, poor metabolism of, 2}, 616903 (3), Autosomal dominant |
| *NUMA1* | 164009 | Leukemia, acute promyelocytic, somatic, 612376 (3) |
| *NUP107* | 607617 | ?Ovarian dysgenesis 6, 618078 (3), Autosomal recessive; Galloway-Mowat syndrome 7, 618348 (3), Autosomal recessive; Nephrotic syndrome, type 11, 616730 (3), Autosomal recessive |
| *NUP133* | 607613 | ?Galloway-Mowat syndrome 8, 618349 (3), Autosomal recessive; Nephrotic syndrome, type 18, 618177 (3), Autosomal recessive |
| *NUP155* | 606694 | ?Atrial fibrillation 15, 615770 (3), Autosomal recessive |
| *NUP160* | 607614 | ?Nephrotic syndrome, type 19, 618178 (3), Autosomal recessive |
| *NUP188* | 615587 | Sandestig-Stefanova syndrome, 618804 (3), Autosomal recessive |
| *NUP205* | 614352 | ?Nephrotic syndrome, type 13, 616893 (3), Autosomal recessive |
| *NUP214* | 114350 | Leukemia, T-cell acute lymphoblastic, somatic, 613065 (3); Leukemia, acute myeloid, somatic, 601626 (3); {Encephalopathy, acute, infection-induced, susceptibility to, 9}, 618426 (3), Autosomal recessive |
| *NUP37* | 609264 | ?Microcephaly 24, primary, autosomal recessive, 618179 (3), Autosomal recessive |
| *NUP62* | 605815 | Striatonigral degeneration, infantile, 271930 (3), Autosomal recessive |
| *NUP85* | 170285 | Nephrotic syndrome, type 17, 618176 (3), Autosomal recessive |
| *NUP88* | 602552 | Fetal akinesia deformation sequence 4, 618393 (3), Autosomal recessive |
| *NUP93* | 614351 | Nephrotic syndrome, type 12, 616892 (3), Autosomal recessive |
| *NUS1* | 610463 | Mental retardation, autosomal dominant 55, with seizures, 617831 (3), Autosomal dominant; ?Congenital disorder of glycosylation, type 1aa, 617082 (3), Autosomal recessive |
| *NXN* | 612895 | Robinow syndrome, autosomal recessive 2, 618529 (3), Autosomal recessive |
| *NYX* | 300278 | Night blindness, congenital stationary (complete), 1A, X-linked, 310500 (3), X-linked recessive |
| *OAT* | 613349 | Gyrate atrophy of choroid and retina with or without ornithinemia, 258870 (3), Autosomal recessive |
| *OBSL1* | 610991 | 3-M syndrome 2, 612921 (3), Autosomal recessive |
| *OCA2* | 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, brown oculocutaneous, 203200 (3), Autosomal recessive; Albinism, oculocutaneous, type II, 203200 (3), Autosomal recessive |
| *OCLN* | 602876 | Pseudo-TORCH syndrome 1, 251290 (3), Autosomal recessive |
| *OCRL* | 300535 | Dent disease 2, 300555 (3), X-linked recessive; Lowe syndrome, 309000 (3), X-linked recessive |
| *ODAD1 (CCDC114)* | 615038 | Ciliary dyskinesia, primary, 20, 615067 (3), Autosomal recessive |
| *ODAD2 (ARMC4)* | 615408 | Ciliary dyskinesia, primary, 23, 615451 (3), Autosomal recessive |
| *ODAD3 (CCDC151)* | 615956 | Ciliary dyskinesia, primary, 30, 616037 (3), Autosomal recessive |
| *ODAD4 (TTC25)* | 617095 | Ciliary dyskinesia, primary, 35, 617092 (3), Autosomal recessive |
| *ODAPH* | 614829 | Amelogenesis imperfecta, type IIA4, 614832 (3), Autosomal recessive |
| *ODC1* | 165640 | Bachmann-Bupp syndrome, 619075 (3), Autosomal dominant |
| *OFD1* | 300170 | Simpson-Golabi-Behmel syndrome, type 2, 300209 (3), X-linked recessive; ?Retinitis pigmentosa 23, 300424 (3), X-linked recessive; Orofaciodigital syndrome I, 311200 (3), X-linked dominant; Joubert syndrome 10, 300804 (3), X-linked recessive |
| *OGDH* | 613022 | Alpha-ketoglutarate dehydrogenase deficiency, 203740 (1), Autosomal recessive |
| *OGG1* | 601982 | Renal cell carcinoma, clear cell, somatic, 144700 (3) |
| *OGT* | 300255 | Mental retardation, X-linked 106, 300997 (3), X-linked recessive |
| *OLR1* | 602601 | {Myocardial infarction, susceptibility to}, 608446 (3) |
| *OPA1* | 605290 | Optic atrophy plus syndrome, 125250 (3), Autosomal dominant; {Glaucoma, normal tension, susceptibility to}, 606657 (3); Optic atrophy 1, 165500 (3), Autosomal dominant; Behr syndrome, 210000 (3), Autosomal recessive; ?Mitochondrial DNA depletion syndrome 14 (encephalocardiomyopathic type), 616896 (3), Autosomal recessive |
| *OPA3* | 606580 | 3-methylglutaconic aciduria, type III, 258501 (3), Autosomal recessive; Optic atrophy 3 with cataract, 165300 (3), Autosomal dominant |
| *OPCML* | 600632 | Ovarian cancer, somatic, 167000 (3) |
| *OPHN1* | 300127 | Mental retardation, X-linked, with cerebellar hypoplasia and distinctive facial appearance, 300486 (3), X-linked recessive |
| *OPLAH* | 614243 | 5-oxoprolinase deficiency, 260005 (3), Autosomal recessive, Autosomal dominant |
| *OPN1LW* | 300822 | Blue cone monochromacy, 303700 (3), X-linked recessive; Colorblindness, protan, 303900 (3), X-linked |
| *OPN1MW* | 300821 | Colorblindness, deutan, 303800 (3), X-linked; Blue cone monochromacy, 303700 (3), X-linked recessive |
| *OPN1SW* | 613522 | Colorblindness, tritan, 190900 (3), Autosomal dominant |
| *OPTN* | 602432 | Glaucoma 1, open angle, E, 137760 (3), Autosomal dominant; Amyotrophic lateral sclerosis 12 with or without frontotemporal dementia, 613435 (3); {Glaucoma, normal tension, susceptibility to}, 606657 (3) |
| *OR2J3* | 615016 | [C3HEX, ability to smell], 615082 (3), Autosomal dominant |
| *ORAI1* | 610277 | Immunodeficiency 9, 612782 (3), Autosomal recessive; Myopathy, tubular aggregate, 2, 615883 (3), Autosomal dominant |
| *ORC1* | 601902 | Meier-Gorlin syndrome 1, 224690 (3), Autosomal recessive |
| *ORC4* | 603056 | Meier-Gorlin syndrome 2, 613800 (3), Autosomal recessive |
| *ORC6* | 607213 | Meier-Gorlin syndrome 3, 613803 (3), Autosomal recessive |
| *OSBPL2* | 606731 | Deafness, autosomal dominant 67, 616340 (3), Autosomal dominant |
| *OSGEP* | 610107 | Galloway-Mowat syndrome 3, 617729 (3), Autosomal recessive |
| *OSMR* | 601743 | Amyloidosis, primary localized cutaneous, 1, 105250 (3), Autosomal dominant |
| *OSTM1* | 607649 | Osteopetrosis, autosomal recessive 5, 259720 (3), Autosomal recessive |
| *OTC* | 300461 | Ornithine transcarbamylase deficiency, 311250 (3), X-linked |
| *OTOA* | 607038 | Deafness, autosomal recessive 22, 607039 (3), Autosomal recessive |
| *OTOF* | 603681 | Auditory neuropathy, autosomal recessive, 1, 601071 (3), Autosomal recessive; Deafness, autosomal recessive 9, 601071 (3), Autosomal recessive |
| *OTOG* | 604487 | Deafness, autosomal recessive 18B, 614945 (3), Autosomal recessive |
| *OTOGL* | 614925 | Deafness, autosomal recessive 84B, 614944 (3), Autosomal recessive |
| *OTUD5* | 300713 | Multiple congenital anomalies-neurodevelopmental syndrome, X-linked, 301056 (3), X-linked recessive |
| *OTUD6B* | 612021 | Intellectual developmental disorder with dysmorphic facies, seizures, and distal limb anomalies, 617452 (3), Autosomal recessive |
| *OTULIN* | 615712 | Autoinflammation, panniculitis, and dermatosis syndrome, 617099 (3), Autosomal recessive |
| *OTX2* | 600037 | Retinal dystrophy, early-onset, with or without pituitary dysfunction, 610125 (3), Autosomal dominant; Pituitary hormone deficiency, combined, 6, 613986 (3), Autosomal dominant; Microphthalmia, syndromic 5, 610125 (3), Autosomal dominant |
| *OVOL2* | 616441 | Corneal dystrophy, posterior polymorphous, 1, 122000 (3), Autosomal dominant |
| *OXCT1* | 601424 | Succinyl CoA:3-oxoacid CoA transferase deficiency, 245050 (3), Autosomal recessive |
| *OXR1* | 605609 | Cerebellar hypoplasia/atrophy, epilepsy, and global developmental delay, 213000 (3), Autosomal recessive |
| *P2RX2* | 600844 | Deafness, autosomal dominant 41, 608224 (3), Autosomal dominant |
| *P2RY12* | 600515 | Bleeding disorder, platelet-type, 8, 609821 (3), Autosomal recessive |
| *P3H1* | 610339 | Osteogenesis imperfecta, type VIII, 610915 (3), Autosomal recessive |
| *P3H2* | 610341 | Myopia, high, with cataract and vitreoretinal degeneration, 614292 (3), Autosomal recessive |
| *P4HA2* | 600608 | Myopia 25, autosomal dominant, 617238 (3), Autosomal dominant |
| *P4HB* | 176790 | Cole-Carpenter syndrome 1, 112240 (3), Autosomal dominant |
| *P4HTM* | 614584 | Hypotonia, hypoventilation, impaired intellectual development, dysautonomia, epilepsy, and eye abnormalities, 618493 (3), Autosomal recessive |
| *PABPN1* | 602279 | Oculopharyngeal muscular dystrophy, 164300 (3), Autosomal dominant |
| *PACS1* | 607492 | Schuurs-Hoeijmakers syndrome, 615009 (3), Autosomal dominant |
| *PACS2* | 610423 | Developmental and epileptic encephalopathy 66, 618067 (3), Autosomal dominant |
| *PADI3* | 606755 | Uncombable hair syndrome, 191480 (3), Autosomal recessive |
| *PADI6* | 610363 | Preimplantation embryonic lethality 2, 617234 (3), Autosomal recessive |
| *PAFAH1B1* | 601545 | Subcortical laminar heterotopia, 607432 (3), Autosomal dominant; Lissencephaly 1, 607432 (3), Autosomal dominant |
| *PAH* | 612349 | [Hyperphenylalaninemia, non-PKU mild], 261600 (3), Autosomal recessive; Phenylketonuria, 261600 (3), Autosomal recessive |
| *PAK1* | 602590 | Intellectual developmental disorder with macrocephaly, seizures, and speech delay, 618158 (3), Autosomal dominant |
| *PAK3* | 300142 | Mental retardation, X-linked 30/47, 300558 (3), X-linked recessive |
| *PALB2* | 610355 | {Breast cancer, susceptibility to}, 114480 (3), Somatic mutation, Autosomal dominant; {Pancreatic cancer, susceptibility to, 3}, 613348 (3); Fanconi anemia, complementation group N, 610832 (3) |
| *PALLD* | 608092 | {Pancreatic cancer, susceptibility to, 1}, 606856 (3), Autosomal dominant |
| *PAM16* | 614336 | Spondylometaphyseal dysplasia, Megarbane-Dagher-Melike type, 613320 (3), Autosomal recessive |
| *PANK2* | 606157 | HARP syndrome, 607236 (3), Autosomal recessive; Neurodegeneration with brain iron accumulation 1, 234200 (3), Autosomal recessive |
| *PANX1* | 608420 | Oocyte maturation defect 7, 618550 (3), Autosomal dominant |
| *PAPSS2* | 603005 | Brachyolmia 4 with mild epiphyseal and metaphyseal changes, 612847 (3), Autosomal recessive |
| *PARK7* | 602533 | Parkinson disease 7, autosomal recessive early-onset, 606324 (3), Autosomal recessive |
| *PARN* | 604212 | Dyskeratosis congenita, autosomal recessive 6, 616353 (3), Autosomal recessive; Pulmonary fibrosis and/or bone marrow failure, telomere-related, 4, 616371 (3), Autosomal dominant |
| *PARS2* | 612036 | Developmental and epileptic encephalopathy 75, 618437 (3), Autosomal recessive |
| *PATL2* | 614661 | Oocyte maturation defect 4, 617743 (3), Autosomal recessive |
| *PAX1* | 167411 | Otofaciocervical syndrome 2, 615560 (3), Autosomal recessive |
| *PAX2* | 167409 | Glomerulosclerosis, focal segmental, 7, 616002 (3), Autosomal dominant; Papillorenal syndrome, 120330 (3), Autosomal dominant |
| *PAX3* | 606597 | Craniofacial-deafness-hand syndrome, 122880 (3), Autosomal dominant; Waardenburg syndrome, type 3, 148820 (3), Autosomal recessive, Autosomal dominant; Waardenburg syndrome, type 1, 193500 (3), Autosomal dominant; Rhabdomyosarcoma 2, alveolar, 268220 (3), Somatic mutation |
| *PAX4* | 167413 | {Diabetes mellitus, ketosis-prone, susceptibility to}, 612227 (3), Autosomal recessive, Autosomal dominant; Maturity-onset diabetes of the young, type IX, 612225 (3); Diabetes mellitus, type 2, 125853 (3), Autosomal dominant |
| *PAX5* | 167414 | {Leukemia, acute lymphoblastic, susceptibility to, 3}, 615545 (3) |
| *PAX6* | 607108 | Optic nerve hypoplasia, 165550 (3), Autosomal dominant; Cataract with late-onset corneal dystrophy, 106210 (3), Autosomal dominant; ?Coloboma, ocular, 120200 (3), Autosomal dominant; ?Coloboma of optic nerve, 120430 (3), Autosomal dominant; Aniridia, 106210 (3), Autosomal dominant; Anterior segment dysgenesis 5, multiple subtypes, 604229 (3), Autosomal dominant; ?Morning glory disc anomaly, 120430 (3), Autosomal dominant; Foveal hypoplasia 1, 136520 (3), Autosomal dominant; Keratitis, 148190 (3), Autosomal dominant |
| *PAX7* | 167410 | Rhabdomyosarcoma 2, alveolar, 268220 (3), Somatic mutation; Myopathy, congenital, progressive, with scoliosis, 618578 (3), Autosomal recessive |
| *PAX8* | 167415 | Hypothyroidism, congenital, due to thyroid dysgenesis or hypoplasia, 218700 (3), Autosomal dominant |
| *PAX9* | 167416 | Tooth agenesis, selective, 3, 604625 (3), Autosomal dominant |
| *PBRM1* | 606083 | ?Renal cell carcinoma, clear cell, 144700 (3) |
| *PBX1* | 176310 | Congenital anomalies of kidney and urinary tract syndrome with or without hearing loss, abnormal ears, or developmental delay, 617641 (3), Autosomal dominant |
| *PC* | 608786 | Pyruvate carboxylase deficiency, 266150 (3), Autosomal recessive |
| *PCARE* | 613425 | Retinitis pigmentosa 54, 613428 (3) |
| *PCBD1* | 126090 | Hyperphenylalaninemia, BH4-deficient, D, 264070 (3), Autosomal recessive |
| *PCCA* | 232000 | Propionicacidemia, 606054 (3), Autosomal recessive |
| *PCCB* | 232050 | Propionicacidemia, 606054 (3), Autosomal recessive |
| *PCDH12* | 605622 | Diencephalic-mesencephalic junction dysplasia syndrome 1, 251280 (3), Autosomal recessive |
| *PCDH15* | 605514 | Usher syndrome, type 1D/F digenic, 601067 (3), Autosomal recessive, Digenic recessive; Deafness, autosomal recessive 23, 609533 (3), Autosomal recessive; Usher syndrome, type 1F, 602083 (3), Autosomal recessive |
| *PCDH19* | 300460 | Developmental and epileptic encephalopathy 9, 300088 (3), X-linked |
| *PCGF2* | 600346 | Turnpenny-Fry syndrome, 618371 (3), Autosomal dominant |
| *PCK1* | 614168 | ?Phosphoenolpyruvate carboxykinase deficiency, cytosolic, 261680 (3), Autosomal recessive |
| *PCK2* | 614095 | PEPCK deficiency, mitochondrial, 261650 (1), Autosomal recessive |
| *PCLO* | 604918 | ?Pontocerebellar hypoplasia, type 3, 608027 (3), Autosomal recessive |
| *PCNA* | 176740 | ?Ataxia-telangiectasia-like disorder 2, 615919 (3), Autosomal recessive |
| *PCNT* | 605925 | Microcephalic osteodysplastic primordial dwarfism, type II, 210720 (3), Autosomal recessive |
| *PCSK1* | 162150 | Obesity with impaired prohormone processing, 600955 (3), Autosomal recessive; {Obesity, susceptibility to, BMIQ12}, 612362 (3) |
| *PCSK9* | 607786 | {Low density lipoprotein cholesterol level QTL 1}, 603776 (3), Autosomal dominant; Hypercholesterolemia, familial, 3, 603776 (3), Autosomal dominant |
| *PCYT1A* | 123695 | Spondylometaphyseal dysplasia with cone-rod dystrophy, 608940 (3), Autosomal recessive |
| *PCYT2* | 602679 | Spastic paraplegia 82, autosomal recessive, 618770 (3), Autosomal recessive |
| *PDCD1* | 600244 | {Multiple sclerosis, disease progression, modifier of}, 126200 (3), Multifactorial; {Systemic lupus erythematosus, susceptibility to, 2}, 605218 (3) |
| *PDCD10* | 609118 | Cerebral cavernous malformations 3, 603285 (3), Autosomal dominant |
| *PDE10A* | 610652 | Striatal degeneration, autosomal dominant, 616922 (3), Autosomal dominant; Dyskinesia, limb and orofacial, infantile-onset, 616921 (3), Autosomal recessive |
| *PDE11A* | 604961 | Pigmented nodular adrenocortical disease, primary, 2, 610475 (3), Autosomal dominant |
| *PDE1C* | 602987 | ?Deafness, autosomal dominant 74, 618140 (3), Autosomal dominant |
| *PDE2A* | 602658 | Intellectual developmental disorder with paroxysmal dyskinesia or seizures, 619150 (3), Autosomal recessive |
| *PDE3A* | 123805 | Hypertension and brachydactyly syndrome, 112410 (3), Autosomal dominant |
| *PDE4D* | 600129 | Acrodysostosis 2, with or without hormone resistance, 614613 (3), Autosomal dominant |
| *PDE6A* | 180071 | Retinitis pigmentosa 43, 613810 (3) |
| *PDE6B* | 180072 | Retinitis pigmentosa-40, 613801 (3), Autosomal recessive; Night blindness, congenital stationary, autosomal dominant 2, 163500 (3), Autosomal dominant |
| *PDE6C* | 600827 | Cone dystrophy 4, 613093 (3), Autosomal recessive |
| *PDE6D* | 602676 | Joubert syndrome 22, 615665 (3), Autosomal recessive |
| *PDE6G* | 180073 | Retinitis pigmentosa 57, 613582 (3), Autosomal recessive |
| *PDE6H* | 601190 | Retinal cone dystrophy 3, 610024 (3), Autosomal recessive, Autosomal dominant; Achromatopsia 6, 610024 (3), Autosomal recessive, Autosomal dominant |
| *PDE8B* | 603390 | Pigmented nodular adrenocortical disease, primary, 3, 614190 (3); Striatal degeneration, autosomal dominant, 609161 (3), Autosomal dominant |
| *PDGFB* | 190040 | Meningioma, SIS-related, 607174 (3), Autosomal dominant; Basal ganglia calcification, idiopathic, 5, 615483 (3), Autosomal dominant; Dermatofibrosarcoma protuberans, 607907 (3) |
| *PDGFRA* | 173490 | Gastrointestinal stromal tumor/GIST-plus syndrome, somatic or familial, 175510 (3); Hypereosinophilic syndrome, idiopathic, resistant to imatinib, 607685 (3), Somatic mutation, Isolated cases |
| *PDGFRB* | 173410 | Premature aging syndrome, Penttinen type, 601812 (3), Autosomal dominant; Kosaki overgrowth syndrome, 616592 (3), Autosomal dominant; Myofibromatosis, infantile, 1, 228550 (3), Autosomal dominant; Basal ganglia calcification, idiopathic, 4, 615007 (3), Autosomal dominant; Myeloproliferative disorder with eosinophilia, 131440 (4), Autosomal dominant |
| *PDGFRL* | 604584 | Hepatocellular cancer, somatic, 114550 (3); Colorectal cancer, somatic, 114500 (3) |
| *PDHA1* | 300502 | Pyruvate dehydrogenase E1-alpha deficiency, 312170 (3), X-linked dominant |
| *PDHB* | 179060 | Pyruvate dehydrogenase E1-beta deficiency, 614111 (3), Autosomal recessive |
| *PDHX* | 608769 | Lacticacidemia due to PDX1 deficiency, 245349 (3), Autosomal recessive |
| *PDK3* | 300906 | ?Charcot-Marie-Tooth disease, X-linked dominant, 6, 300905 (3), X-linked dominant |
| *PDLIM4* | 603422 | {Osteoporosis, susceptibility to}, 166710 (3), Autosomal dominant |
| *PDP1* | 605993 | Pyruvate dehydrogenase phosphatase deficiency, 608782 (3), Autosomal recessive |
| *PDSS1* | 607429 | Coenzyme Q10 deficiency, primary, 2, 614651 (3), Autosomal recessive |
| *PDSS2* | 610564 | Coenzyme Q10 deficiency, primary, 3, 614652 (3), Autosomal recessive |
| *PDX1* | 600733 | {Diabetes mellitus, type II, susceptibility to}, 125853 (3), Autosomal dominant; Pancreatic agenesis 1, 260370 (3), Autosomal recessive; MODY, type IV, 606392 (3) |
| *PDXK* | 179020 | Neuropathy, hereditary motor and sensory, type VIC, with optic atrophy, 618511 (3), Autosomal recessive |
| *PDYN* | 131340 | Spinocerebellar ataxia 23, 610245 (3), Autosomal dominant |
| *PDZD7* | 612971 | Deafness, autosomal recessive 57, 618003 (3), Autosomal recessive; {Retinal disease in Usher syndrome type IIA, modifier of}, 276901 (3), Autosomal recessive; Usher syndrome, type IIC, GPR98/PDZD7 digenic, 605472 (3), Digenic dominant, Autosomal recessive |
| *PEPD* | 613230 | Prolidase deficiency, 170100 (3), Autosomal recessive |
| *PER2* | 603426 | ?Advanced sleep phase syndrome, familial, 1, 604348 (3), Autosomal dominant |
| *PER3* | 603427 | ?Advanced sleep phase syndrome, familial, 3, 616882 (3), Autosomal dominant |
| *PERCC1 (AL032819.3)* | 618656 | Diarrhea 11, malabsorptive, congenital, 618662 (3), Autosomal recessive |
| *PERP* | 609301 | Erythrokeratodermia variabilis et progressiva 7, 619209 (3), Autosomal recessive; Olmsted syndrome 2, 619208 (3), Autosomal dominant |
| *PET100* | 614770 | Mitochondrial complex IV deficiency, nuclear type 12, 619055 (3), Autosomal recessive |
| *PET117* | 614771 | ?Mitochondrial complex IV deficiency, nuclear type 19, 619063 (3), Autosomal recessive |
| *PEX1* | 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 |
| *PEX10* | 602859 | Peroxisome biogenesis disorder 6A (Zellweger), 614870 (3), Autosomal recessive; Peroxisome biogenesis disorder 6B, 614871 (3), Autosomal recessive |
| *PEX11B* | 603867 | Peroxisome biogenesis disorder 14B, 614920 (3), Autosomal recessive |
| *PEX12* | 601758 | Peroxisome biogenesis disorder 3B, 266510 (3), Autosomal recessive; Peroxisome biogenesis disorder 3A (Zellweger), 614859 (3), Autosomal recessive |
| *PEX13* | 601789 | Peroxisome biogenesis disorder 11A (Zellweger), 614883 (3), Autosomal recessive; Peroxisome biogenesis disorder 11B, 614885 (3), Autosomal recessive |
| *PEX14* | 601791 | Peroxisome biogenesis disorder 13A (Zellweger), 614887 (3), Autosomal recessive |
| *PEX16* | 603360 | Peroxisome biogenesis disorder 8B, 614877 (3), Autosomal recessive; Peroxisome biogenesis disorder 8A (Zellweger), 614876 (3), Autosomal recessive |
| *PEX19* | 600279 | Peroxisome biogenesis disorder 12A (Zellweger), 614886 (3), Autosomal recessive |
| *PEX2* | 170993 | Peroxisome biogenesis disorder 5A (Zellweger), 614866 (3), Autosomal recessive; Peroxisome biogenesis disorder 5B, 614867 (3), Autosomal recessive |
| *PEX26* | 608666 | Peroxisome biogenesis disorder 7B, 614873 (3), Autosomal recessive; Peroxisome biogenesis disorder 7A (Zellweger), 614872 (3), Autosomal recessive |
| *PEX3* | 603164 | Peroxisome biogenesis disorder 10A (Zellweger), 614882 (3), Autosomal recessive; ?Peroxisome biogenesis disorder 10B, 617370 (3), Autosomal recessive |
| *PEX5* | 600414 | Peroxisome biogenesis disorder 2B, 202370 (3), Autosomal recessive; Peroxisome biogenesis disorder 2A (Zellweger), 214110 (3), Autosomal recessive; Rhizomelic chondrodysplasia punctata, type 5, 616716 (3), Autosomal recessive |
| *PEX6* | 601498 | Peroxisome biogenesis disorder 4B, 614863 (3), Autosomal recessive, Autosomal dominant; Peroxisome biogenesis disorder 4A (Zellweger), 614862 (3), Autosomal recessive; Heimler syndrome 2, 616617 (3), Autosomal recessive |
| *PEX7* | 601757 | Rhizomelic chondrodysplasia punctata, type 1, 215100 (3), Autosomal recessive; Peroxisome biogenesis disorder 9B, 614879 (3), Autosomal recessive |
| *PFKL* | 171860 | Hemolytic anemia due to phosphofructokinase deficiency (1) |
| *PFKM* | 610681 | Glycogen storage disease VII, 232800 (3), Autosomal recessive |
| *PFN1* | 176610 | Amyotrophic lateral sclerosis 18, 614808 (3) |
| *PGAM2* | 612931 | Glycogen storage disease X, 261670 (3), Autosomal recessive |
| *PGAP1* | 611655 | Neurodevelopmental disorder with dysmorphic features, spasticity, and brain abnormalities, 615802 (3), Autosomal recessive |
| *PGAP2* | 615187 | Hyperphosphatasia with mental retardation syndrome 3, 614207 (3), Autosomal recessive |
| *PGAP3* | 611801 | Hyperphosphatasia with mental retardation syndrome 4, 615716 (3), Autosomal recessive |
| *PGK1* | 311800 | Phosphoglycerate kinase 1 deficiency, 300653 (3), X-linked recessive |
| *PGM1* | 171900 | Congenital disorder of glycosylation, type It, 614921 (3), Autosomal recessive |
| *PGM3* | 172100 | Immunodeficiency 23, 615816 (3), Autosomal recessive |
| *PGR* | 607311 | ?Progesterone resistance, 264080 (2), Autosomal recessive |
| *PHACTR1* | 608723 | Developmental and epileptic encephalopathy 70, 618298 (3), Autosomal dominant |
| *PHB* | 176705 | {Breast cancer, susceptibility to}, 114480 (3), Somatic mutation, Autosomal dominant |
| *PHC1* | 602978 | ?Microcephaly 11, primary, autosomal recessive, 615414 (3), Autosomal recessive |
| *PHEX* | 300550 | Hypophosphatemic rickets, X-linked dominant, 307800 (3), X-linked dominant |
| *PHF21A* | 608325 | Intellectual developmental disorder with behavioral abnormalities and craniofacial dysmorphism with or without seizures, 618725 (3), Autosomal dominant |
| *PHF6* | 300414 | Borjeson-Forssman-Lehmann syndrome, 301900 (3), X-linked recessive |
| *PHF8* | 300560 | Mental retardation syndrome, X-linked, Siderius type, 300263 (3), X-linked recessive |
| *PHGDH* | 606879 | Neu-Laxova syndrome 1, 256520 (3), Autosomal recessive; Phosphoglycerate dehydrogenase deficiency, 601815 (3), Autosomal recessive |
| *PHIP* | 612870 | Chung-Jansen syndrome, 617991 (3), Autosomal dominant |
| *PHKA1* | 311870 | Muscle glycogenosis, 300559 (3), X-linked recessive |
| *PHKA2* | 300798 | Glycogen storage disease, type IXa2, 306000 (3), X-linked recessive; Glycogen storage disease, type IXa1, 306000 (3), X-linked recessive |
| *PHKB* | 172490 | Phosphorylase kinase deficiency of liver and muscle, autosomal recessive, 261750 (3), Autosomal recessive |
| *PHKG2* | 172471 | Glycogen storage disease IXc, 613027 (3), Autosomal recessive |
| *PHOX2A* | 602753 | Fibrosis of extraocular muscles, congenital, 2, 602078 (3), Autosomal recessive |
| *PHOX2B* | 603851 | {Neuroblastoma, susceptibility to, 2}, 613013 (3); Neuroblastoma with Hirschsprung disease, 613013 (3); Central hypoventilation syndrome, congenital, with or without Hirschsprung disease, 209880 (3), Autosomal dominant |
| *PHYH* | 602026 | Refsum disease, 266500 (3), Autosomal recessive |
| *PHYKPL* | 614683 | [?Phosphohydroxylysinuria], 615011 (3) |
| *PI4KA* | 600286 | Polymicrogyria, perisylvian, with cerebellar hypoplasia and arthrogryposis, 616531 (3), Autosomal recessive |
| *PIBF1* | 607532 | Joubert syndrome 33, 617767 (3), Autosomal recessive |
| *PICALM* | 603025 | Leukemia, acute myeloid, somatic, 601626 (3) |
| *PIEZO1* | 611184 | Lymphatic malformation 6, 616843 (3), Autosomal recessive; Dehydrated hereditary stomatocytosis with or without pseudohyperkalemia and/or perinatal edema, 194380 (3), Autosomal dominant |
| *PIEZO2* | 613629 | Arthrogryposis, distal, type 5, 108145 (3), Autosomal dominant; Arthrogryposis, distal, with impaired proprioception and touch, 617146 (3), Autosomal recessive; Arthrogryposis, distal, type 3, 114300 (3), Autosomal dominant; ?Marden-Walker syndrome, 248700 (3), Autosomal dominant |
| *PIGA* | 311770 | Paroxysmal nocturnal hemoglobinuria, somatic, 300818 (3); Multiple congenital anomalies-hypotonia-seizures syndrome 2, 300868 (3), X-linked recessive |
| *PIGB* | 604122 | Developmental and epileptic encephalopathy 80, 618580 (3), Autosomal recessive |
| *PIGC* | 601730 | Glycosylphosphatidylinositol biosynthesis defect 16, 617816 (3), Autosomal recessive |
| *PIGF* | 600153 | Onychodystrophy, osteodystrophy, impaired intellectual development, and seizures syndrome, 619356 (3), Autosomal recessive |
| *PIGG* | 616918 | Mental retardation, autosomal recessive 53, 616917 (3), Autosomal recessive |
| *PIGH* | 600154 | Glycosylphosphatidylinositol biosynthesis defect 17, 618010 (3), Autosomal recessive |
| *PIGK* | 605087 | Neurodevelopmental disorder with hypotonia and cerebellar atrophy, with or without seizures, 618879 (3), Autosomal recessive |
| *PIGL* | 605947 | CHIME syndrome, 280000 (3), Autosomal recessive |
| *PIGM* | 610273 | Glycosylphosphatidylinositol deficiency, 610293 (3), Autosomal recessive |
| *PIGN* | 606097 | Multiple congenital anomalies-hypotonia-seizures syndrome 1, 614080 (3), Autosomal recessive |
| *PIGO* | 614730 | Hyperphosphatasia with mental retardation syndrome 2, 614749 (3), Autosomal recessive |
| *PIGP* | 605938 | Developmental and epileptic encephalopathy 55, 617599 (3), Autosomal recessive |
| *PIGQ* | 605754 | Developmental and epileptic encephalopathy 77, 618548 (3), Autosomal recessive |
| *PIGS* | 610271 | Developmental and epileptic encephalopathy 95, 618143 (3), Autosomal recessive |
| *PIGT* | 610272 | ?Paroxysmal nocturnal hemoglobinuria 2, 615399 (3), Somatic mutation, Autosomal dominant; Multiple congenital anomalies-hypotonia-seizures syndrome 3, 615398 (3), Autosomal recessive |
| *PIGU* | 608528 | Neurodevelopmental disorder with brain anomalies, seizures, and scoliosis, 618590 (3), Autosomal recessive |
| *PIGV* | 610274 | Hyperphosphatasia with mental retardation syndrome 1, 239300 (3), Autosomal recessive |
| *PIGW* | 610275 | Glycosylphosphatidylinositol biosynthesis defect 11, 616025 (3), Autosomal recessive |
| *PIGY* | 610662 | Hyperphosphatasia with mental retardation syndrome 6, 616809 (3), Autosomal recessive |
| *PIK3C2A* | 603601 | Oculoskeletodental syndrome, 618440 (3), Autosomal recessive |
| *PIK3CA* | 171834 | CLOVE syndrome, somatic, 612918 (3); Hepatocellular carcinoma, somatic, 114550 (3); Breast cancer, somatic, 114480 (3); Ovarian cancer, somatic, 167000 (3); Colorectal cancer, somatic, 114500 (3); Macrodactyly, somatic, 155500 (3); CLAPO syndrome, somatic, 613089 (3); Keratosis, seborrheic, somatic, 182000 (3); Nevus, epidermal, somatic, 162900 (3); Gastric cancer, somatic, 613659 (3); Nonsmall cell lung cancer, somatic, 211980 (3); Megalencephaly-capillary malformation-polymicrogyria syndrome, somatic, 602501 (3); Cowden syndrome 5, 615108 (3) |
| *PIK3CD* | 602839 | Immunodeficiency 14A, autosomal dominant, 615513 (3), Autosomal dominant; Immunodeficiency 14B, autosomal recessive, 619281 (3), Autosomal recessive; ?Roifman-Chitayat syndrome, digenic, 613328 (3), Digenic recessive |
| *PIK3R1* | 171833 | Immunodeficiency 36, 616005 (3), Autosomal dominant; ?Agammaglobulinemia 7, autosomal recessive, 615214 (3), Autosomal recessive; SHORT syndrome, 269880 (3), Autosomal dominant |
| *PIK3R2* | 603157 | Megalencephaly-polymicrogyria-polydactyly-hydrocephalus syndrome 1, 603387 (3), Autosomal dominant |
| *PIK3R5* | 611317 | Ataxia-oculomotor apraxia 3, 615217 (3), Autosomal recessive |
| *PIKFYVE* | 609414 | Corneal fleck dystrophy, 121850 (3), Autosomal dominant |
| *PINK1* | 608309 | Parkinson disease 6, early onset, 605909 (3), Autosomal recessive |
| *PIP5K1C* | 606102 | Lethal congenital contractural syndrome 3, 611369 (3), Autosomal recessive |
| *PISD* | 612770 | Liberfarb syndrome, 618889 (3), Autosomal recessive |
| *PITPNM3* | 608921 | Cone-rod dystrophy 5, 600977 (3), Autosomal dominant |
| *PITRM1* | 618211 | Spinocerebellar ataxia, autosomal recessive 30, 619405 (3), Autosomal recessive |
| *PITX1* | 602149 | Liebenberg syndrome, 186550 (4), Autosomal dominant; Clubfoot, congenital, with or without deficiency of long bones and/or mirror-image polydactyly, 119800 (3), Autosomal dominant |
| *PITX2* | 601542 | Ring dermoid of cornea, 180550 (3), Autosomal dominant; Axenfeld-Rieger syndrome, type 1, 180500 (3), Autosomal dominant; Anterior segment dysgenesis 4, 137600 (3), Autosomal dominant |
| *PITX3* | 602669 | Cataract 11, multiple types, 610623 (3), Autosomal recessive, Autosomal dominant; Anterior segment dysgenesis 1, multiple subtypes, 107250 (3), Autosomal dominant; Cataract 11, syndromic, autosomal recessive, 610623 (3), Autosomal recessive, Autosomal dominant |
| *PJVK* | 610219 | Deafness, autosomal recessive 59, 610220 (3), Autosomal recessive |
| *PKD1* | 601313 | Polycystic kidney disease 1, 173900 (3), Autosomal dominant |
| *PKD1L1* | 609721 | Heterotaxy, visceral, 8, autosomal, 617205 (3), Autosomal recessive |
| *PKD2* | 173910 | Polycystic kidney disease 2, 613095 (3), Autosomal dominant |
| *PKDCC* | 614150 | Rhizomelic limb shortening with dysmorphic features, 618821 (3), Autosomal recessive |
| *PKHD1* | 606702 | Polycystic kidney disease 4, with or without hepatic disease, 263200 (3), Autosomal recessive |
| *PKLR* | 609712 | Adenosine triphosphate, elevated, of erythrocytes, 102900 (3), Autosomal dominant; Pyruvate kinase deficiency, 266200 (3), Autosomal recessive |
| *PKP1* | 601975 | Ectodermal dysplasia/skin fragility syndrome, 604536 (3), Autosomal recessive |
| *PKP2* | 602861 | Arrhythmogenic right ventricular dysplasia 9, 609040 (3), Autosomal dominant |
| *PLA2G2A* | 172411 | {?Colorectal cancer, susceptibility to}, 114500 (3), Somatic mutation, Autosomal dominant |
| *PLA2G4A* | 600522 | Gastrointestinal ulceration, recurrent, with dysfunctional platelets, 618372 (3), Autosomal recessive |
| *PLA2G5* | 601192 | [Fleck retina, familial benign], 228980 (3), Autosomal recessive |
| *PLA2G6* | 603604 | Parkinson disease 14, autosomal recessive, 612953 (3), Autosomal recessive; Neurodegeneration with brain iron accumulation 2B, 610217 (3), Autosomal recessive; Infantile neuroaxonal dystrophy 1, 256600 (3), Autosomal recessive |
| *PLA2G7* | 601690 | Platelet-activating factor acetylhydrolase deficiency, 614278 (3), Autosomal recessive; {Asthma, susceptibility to}, 600807 (3), Autosomal dominant; {Atopy, susceptibility to}, 147050 (3), Autosomal dominant |
| *PLAA* | 603873 | Neurodevelopmental disorder with progressive microcephaly, spasticity, and brain anomalies, 617527 (3), Autosomal recessive |
| *PLAG1* | 603026 | Adenomas, salivary gland pleomorphic, somatic, 181030 (3); Silver-Russell syndrome 4, 618907 (3), Autosomal dominant |
| *PLAT* | 173370 | Thrombophilia, familial, due to decreased release of PLAT, 612348 (1); Hyperfibrinolysis, familial, due to increased release of PLAT, 612348 (1) |
| *PLAU* | 191840 | Quebec platelet disorder, 601709 (3), Autosomal dominant; {Alzheimer disease, late-onset, susceptibility to}, 104300 (3), Autosomal dominant |
| *PLCB1* | 607120 | Developmental and epileptic encephalopathy 12, 613722 (3), Autosomal recessive |
| *PLCB2* | 604114 | Platelet PLC beta-2 deficiency (1) |
| *PLCB3* | 600230 | Spondylometaphyseal dysplasia with corneal dystrophy, 618961 (3), Autosomal recessive |
| *PLCB4* | 600810 | Auriculocondylar syndrome 2, 614669 (3), Autosomal recessive, Autosomal dominant |
| *PLCD1* | 602142 | Nail disorder, nonsyndromic congenital, 3, (leukonychia), 151600 (3), Autosomal recessive, Autosomal dominant |
| *PLCE1* | 608414 | Nephrotic syndrome, type 3, 610725 (3), Autosomal recessive |
| *PLCG2* | 600220 | Autoinflammation, antibody deficiency, and immune dysregulation syndrome, 614878 (3), Autosomal dominant; Familial cold autoinflammatory syndrome 3, 614468 (3), Autosomal dominant |
| *PLCZ1* | 608075 | Spermatogenic failure 17, 617214 (3), Autosomal recessive |
| *PLD1* | 602382 | Cardiac valvular defect, developmental, 212093 (3), Autosomal recessive |
| *PLD3* | 615698 | ?Spinocerebellar ataxia 46, 617770 (3), Autosomal dominant |
| *PLEC* | 601282 | ?Epidermolysis bullosa simplex with nail dystrophy, 616487 (3), Autosomal recessive; Epidermolysis bullosa simplex, Ogna type, 131950 (3), Autosomal dominant; Epidermolysis bullosa simplex with pyloric atresia, 612138 (3), Autosomal recessive; Muscular dystrophy, limb-girdle, autosomal recessive 17, 613723 (3), Autosomal recessive; Epidermolysis bullosa simplex with muscular dystrophy, 226670 (3), Autosomal recessive |
| *PLEKHG2* | 611893 | Leukodystrophy and acquired microcephaly with or without dystonia, 616763 (3), Autosomal recessive |
| *PLEKHG4* | 609526/600223 | -/Spinocerebellar ataxia 4, 600223 (2), Autosomal dominant |
| *PLEKHG5* | 611101 | Spinal muscular atrophy, distal, autosomal recessive, 4, 611067 (3), Autosomal recessive; Charcot-Marie-Tooth disease, recessive intermediate C, 615376 (3), Autosomal recessive |
| *PLEKHM1* | 611466 | ?Osteopetrosis, autosomal recessive 6, 611497 (3), Autosomal recessive; Osteopetrosis, autosomal dominant 3, 618107 (3), Autosomal dominant |
| *PLG* | 173350 | Dysplasminogenemia, 217090 (3), Autosomal recessive; Angioedema, hereditary, 4, 619360 (3), Autosomal dominant; Plasminogen deficiency, type I, 217090 (3), Autosomal recessive |
| *PLIN1* | 170290 | Lipodystrophy, familial partial, type 4, 613877 (3), Autosomal dominant |
| *PLK4* | 605031 | Microcephaly and chorioretinopathy, autosomal recessive, 2, 616171 (3), Autosomal recessive |
| *PLN* | 172405 | Cardiomyopathy, dilated, 1P, 609909 (3); Cardiomyopathy, hypertrophic, 18, 613874 (3), Autosomal dominant |
| *PLOD1* | 153454 | Ehlers-Danlos syndrome, kyphoscoliotic type, 1, 225400 (3), Autosomal recessive |
| *PLOD2* | 601865 | Bruck syndrome 2, 609220 (3), Autosomal recessive |
| *PLOD3* | 603066 | Lysyl hydroxylase 3 deficiency, 612394 (3), Autosomal recessive |
| *PLP1* | 300401 | Pelizaeus-Merzbacher disease, 312080 (3), X-linked recessive; Spastic paraplegia 2, X-linked, 312920 (3), X-linked recessive |
| *PLPBP* | 604436 | Epilepsy, early-onset, vitamin B6-dependent, 617290 (3), Autosomal recessive |
| *PLS1* | 602734 | Deafness, autosomal dominant 76, 618787 (3), Autosomal dominant |
| *PLS3* | 300131 | Bone mineral density QTL18, osteoporosis, 300910 (3), X-linked dominant |
| *PLTP* | 172425 | [High density lipoprotein cholesterol level QTL 9] (3) |
| *PLVAP* | 607647 | Diarrhea 10, protein-losing enteropathy type, 618183 (3), Autosomal recessive |
| *PMFBP1* | 618085 | Spermatogenic failure 31, 618112 (3), Autosomal recessive |
| *PML* | 102578 | Leukemia, acute promyelocytic, PML/RARA type (3) |
| *PMM2* | 601785 | Congenital disorder of glycosylation, type Ia, 212065 (3), Autosomal recessive |
| *PMP2* | 170715 | Charcot-Marie-Tooth disease, demyelinating, type 1G, 618279 (3), Autosomal dominant |
| *PMP22* | 601097 | Charcot-Marie-Tooth disease, type 1A, 118220 (3), Autosomal dominant; Roussy-Levy syndrome, 180800 (3), Autosomal dominant; Charcot-Marie-Tooth disease, type 1E, 118300 (3), Autosomal dominant; ?Neuropathy, inflammatory demyelinating, 139393 (3), ?Autosomal dominant; Neuropathy, recurrent, with pressure palsies, 162500 (3), Autosomal dominant; Dejerine-Sottas disease, 145900 (3), Autosomal recessive, Autosomal dominant |
| *PMPCA* | 613036 | Spinocerebellar ataxia, autosomal recessive 2, 213200 (3), Autosomal recessive |
| *PMPCB* | 603131 | Multiple mitochondrial dysfunctions syndrome 6, 617954 (3), Autosomal recessive |
| *PMS2* | 600259 | Colorectal cancer, hereditary nonpolyposis, type 4, 614337 (3); Mismatch repair cancer syndrome 4, 619101 (3), Autosomal recessive |
| *PMVK* | 607622 | Porokeratosis 1, multiple types, 175800 (3), Autosomal dominant |
| *PNKD* | 609023 | Paroxysmal nonkinesigenic dyskinesia 1, 118800 (3), Autosomal dominant |
| *PNKP* | 605610 | ?Charcot-Marie-Tooth disease, type 2B2, 605589 (3), Autosomal recessive; Ataxia-oculomotor apraxia 4, 616267 (3), Autosomal recessive; Microcephaly, seizures, and developmental delay, 613402 (3), Autosomal recessive |
| *PNLIP* | 246600 | ?Pancreatic lipase deficiency, 614338 (3), Autosomal recessive |
| *PNP* | 164050 | Immunodeficiency due to purine nucleoside phosphorylase deficiency, 613179 (3), Autosomal recessive |
| *PNPLA1* | 612121 | Ichthyosis, congenital, autosomal recessive 10, 615024 (3), Autosomal recessive |
| *PNPLA2* | 609059 | Neutral lipid storage disease with myopathy, 610717 (3), Autosomal recessive |
| *PNPLA6* | 603197 | Spastic paraplegia 39, autosomal recessive, 612020 (3), Autosomal recessive; Oliver-McFarlane syndrome, 275400 (3), Autosomal recessive; ?Laurence-Moon syndrome, 245800 (3), Autosomal recessive; Boucher-Neuhauser syndrome, 215470 (3), Autosomal recessive |
| *PNPLA8* | 612123 | ?Mitochondrial myopathy with lactic acidosis, 251950 (3), Autosomal recessive |
| *PNPO* | 603287 | Pyridoxamine 5'-phosphate oxidase deficiency, 610090 (3), Autosomal recessive |
| *PNPT1* | 610316 | Deafness, autosomal recessive 70, 614934 (3), Autosomal recessive; Combined oxidative phosphorylation deficiency 13, 614932 (3), Autosomal recessive |
| *POC1A* | 614783 | Short stature, onychodysplasia, facial dysmorphism, and hypotrichosis, 614813 (3), Autosomal recessive |
| *POC1B* | 614784 | Cone-rod dystrophy 20, 615973 (3), Autosomal recessive |
| *POF1B* | 300603 | ?Premature ovarian failure 2B, 300604 (3), X-linked recessive |
| *POFUT1* | 607491 | Dowling-Degos disease 2, 615327 (3), Autosomal dominant |
| *POGLUT1* | 615618 | Dowling-Degos disease 4, 615696 (3), Autosomal dominant; ?Muscular dystrophy, limb-girdle, autosomal recessive 21, 617232 (3), Autosomal recessive |
| *POGZ* | 614787 | White-Sutton syndrome, 616364 (3), Autosomal dominant |
| *POLA1* | 312040 | Pigmentary disorder, reticulate, with systemic manifestations, X-linked, 301220 (3), X-linked recessive; Van Esch-O'Driscoll syndrome, 301030 (3), X-linked recessive |
| *POLD1* | 174761 | Mandibular hypoplasia, deafness, progeroid features, and lipodystrophy syndrome, 615381 (3), Autosomal dominant; {Colorectal cancer, susceptibility to, 10}, 612591 (3), Autosomal dominant |
| *POLE* | 174762 | {Colorectal cancer, susceptibility to, 12}, 615083 (3), Autosomal dominant; FILS syndrome, 615139 (3), Autosomal recessive; IMAGE-I syndrome, 618336 (3), Autosomal recessive |
| *POLG* | 174763 | Mitochondrial recessive ataxia syndrome (includes SANDO and SCAE), 607459 (3), Autosomal recessive; Mitochondrial DNA depletion syndrome 4B (MNGIE type), 613662 (3), Autosomal recessive; Mitochondrial DNA depletion syndrome 4A (Alpers type), 203700 (3), Autosomal recessive; Progressive external ophthalmoplegia, autosomal dominant 1, 157640 (3), Autosomal dominant; Progressive external ophthalmoplegia, autosomal recessive 1, 258450 (3), Autosomal recessive |
| *POLG2* | 604983 | Progressive external ophthalmoplegia with mitochondrial DNA deletions, autosomal dominant 4, 610131 (3), Autosomal dominant; ?Mitochondrial DNA depletion syndrome 16 (hepatic type), 618528 (3), Autosomal recessive; ?Mitochondrial DNA depletion syndrome 16B (neuroophthalmic type), 619425 (3), Autosomal recessive |
| *POLH* | 603968 | Xeroderma pigmentosum, variant type, 278750 (3), Autosomal recessive |
| *POLR1A* | 616404 | Acrofacial dysostosis, Cincinnati type, 616462 (3), Autosomal dominant |
| *POLR1B* | 602000 | Treacher-Collins syndrome 4, 618939 (3), Autosomal dominant |
| *POLR1C* | 610060 | Leukodystrophy, hypomyelinating, 11, 616494 (3), Autosomal recessive; Treacher Collins syndrome 3, 248390 (3), Autosomal recessive |
| *POLR1D* | 613715 | Treacher Collins syndrome 2, 613717 (3), Autosomal recessive, Autosomal dominant |
| *POLR2A* | 180660 | Neurodevelopmental disorder with hypotonia and variable intellectual and behavioral abnormalities, 618603 (3), Autosomal dominant |
| *POLR3A* | 614258 | Wiedemann-Rautenstrauch syndrome, 264090 (3), Autosomal recessive; Leukodystrophy, hypomyelinating, 7, with or without oligodontia and/or hypogonadotropic hypogonadism, 607694 (3), Autosomal recessive |
| *POLR3B* | 614366 | Leukodystrophy, hypomyelinating, 8, with or without oligodontia and/or hypogonadotropic hypogonadism, 614381 (3), Autosomal recessive |
| *POLR3GL* | 617457 | Short stature, oligodontia, dysmorphic facies, and motor delay, 619234 (3), Autosomal recessive |
| *POLR3K* | 606007 | Leukodystrophy, hypomyelinating, 21, 619310 (3), Autosomal recessive |
| *POMC* | 176830 | {Obesity, early-onset, susceptibility to}, 601665 (3), Multifactorial, Autosomal recessive, Autosomal dominant; Obesity, adrenal insufficiency, and red hair due to POMC deficiency, 609734 (3), Autosomal recessive |
| *POMGNT1* | 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 |
| *POMGNT2* | 614828 | Muscular dystrophy-dystroglycanopathy (limb-girdle) type C, 8, 618135 (3), Autosomal recessive; Muscular dystrophy-dystroglycanopathy (congenital with brain and eye anomalies, type A, 8, 614830 (3), Autosomal recessive |
| *POMK* | 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 |
| *POMP* | 613386 | Proteasome-associated autoinflammatory syndrome 2, 618048 (3), Autosomal dominant; Keratosis linearis with ichthyosis congenita and sclerosing keratoderma, 601952 (3), Autosomal recessive |
| *POMT1* | 607423 | Muscular dystrophy-dystroglycanopathy (congenital with brain and eye anomalies), type A, 1, 236670 (3), Autosomal recessive; Muscular dystrophy-dystroglycanopathy (limb-girdle), type C, 1, 609308 (3), Autosomal recessive; Muscular dystrophy-dystroglycanopathy (congenital with mental retardation), type B, 1, 613155 (3), Autosomal recessive |
| *POMT2* | 607439 | Muscular dystrophy-dystroglycanopathy (limb-girdle), type C, 2, 613158 (3), Autosomal recessive; Muscular dystrophy-dystroglycanopathy (congenital with mental retardation), type B, 2, 613156 (3), Autosomal recessive; Muscular dystrophy-dystroglycanopathy (congenital with brain and eye anomalies), type A, 2, 613150 (3), Autosomal recessive |
| *PON1* | 168820 | {Organophosphate poisoning, sensitivity to} (3); {Coronary artery spasm 2, susceptibility to (3); {Coronary artery disease, susceptibility to} (3); {Microvascular complications of diabetes 5}, 612633 (3) |
| *PON2* | 602447 | {Coronary artery disease, susceptibility to} (3) |
| *POP1* | 602486 | Anauxetic dysplasia 2, 617396 (3), Autosomal recessive |
| *POPDC3* | 605824 | Muscular dystrophy, limb-girdle, autosomal recessive 26, 618848 (3), Autosomal recessive |
| *POR* | 124015 | Antley-Bixler syndrome with genital anomalies and disordered steroidogenesis, 201750 (3), Autosomal recessive; Disordered steroidogenesis due to cytochrome P450 oxidoreductase, 613571 (3) |
| *PORCN* | 300651 | Focal dermal hypoplasia, 305600 (3), X-linked dominant |
| *POT1* | 606478 | {Glioma susceptibility 9}, 616568 (3), Autosomal dominant; {Melanoma, cutaneous malignant, susceptibility to, 10}, 615848 (3), Autosomal dominant |
| *POU1F1* | 173110 | Pituitary hormone deficiency, combined, 1, 613038 (3), Autosomal recessive, Autosomal dominant |
| *POU3F3* | 602480 | Snijders Blok-Fisher syndrome, 618604 (3), Autosomal dominant |
| *POU3F4* | 300039 | Deafness, X-linked 2, 304400 (3), X-linked recessive |
| *POU4F1* | 601632 | Ataxia, intention tremor, and hypotonia syndrome, childhood-onset, 619352 (3), Autosomal dominant |
| *POU4F3* | 602460 | Deafness, autosomal dominant 15, 602459 (3), Autosomal dominant |
| *POU6F2* | 609062 | {Wilms tumor susceptibility-5}, 601583 (3), Somatic mutation, Autosomal dominant |
| *PPA2* | 609988 | ?Sudden cardiac failure, alcohol-induced, 617223 (3), Autosomal recessive; Sudden cardiac failure, infantile, 617222 (3), Autosomal recessive |
| *PPARA* | 170998 | {Hyperapobetalipoproteinemia, susceptibility to} (3) |
| *PPARG* | 601487 | {Diabetes, type 2}, 125853 (3), Autosomal dominant; Insulin resistance, severe, digenic, 604367 (3), Autosomal dominant; Lipodystrophy, familial partial, type 3, 604367 (3), Autosomal dominant; [Obesity, resistance to] (3); Obesity, severe, 601665 (3), Multifactorial, Autosomal recessive, Autosomal dominant; Carotid intimal medial thickness 1, 609338 (3) |
| *PPCS* | 609853 | Cardiomyopathy, dilated, 2C, 618189 (3), Autosomal recessive |
| *PPIB* | 123841 | Osteogenesis imperfecta, type IX, 259440 (3), Autosomal recessive |
| *PPIL1* | 601301 | Pontocerebellar hypoplasia, type 14, 619301 (3), Autosomal recessive |
| *PPIP5K2* | 611648 | Deafness, autosomal recessive 100, 618422 (3), Autosomal recessive |
| *PPM1D* | 605100 | Breast cancer, somatic, 114480 (3); Jansen de Vries syndrome, 617450 (3), Autosomal dominant |
| *PPM1K* | 611065 | ?Maple syrup urine disease, mild variant, 615135 (3) |
| *PPOX* | 600923 | Porphyria variegata, 176200 (3), Autosomal dominant |
| *PPP1CB* | 600590 | Noonan syndrome-like disorder with loose anagen hair 2, 617506 (3), Autosomal dominant |
| *PPP1R12A* | 602021 | Genitourinary and/or/brain malformation syndrome, 618820 (3), Autosomal dominant |
| *PPP1R15B* | 613257 | Microcephaly, short stature, and impaired glucose metabolism 2, 616817 (3), Autosomal recessive |
| *PPP1R17* | 604088 | {Hypercholesterolemia, susceptibility to}, 143890 (3), Autosomal recessive, Autosomal dominant |
| *PPP1R21* | 618159 | Neurodevelopmental disorder with hypotonia, facial dysmorphism, and brain abnormalities, 619383 (3), Autosomal recessive |
| *PPP1R3A* | 600917 | Insulin resistance, severe, digenic, 125853 (3), Autosomal dominant |
| *PPP2CA* | 176915 | Neurodevelopmental disorder and language delay with or without structural brain abnormalities, 618354 (3), Autosomal dominant |
| *PPP2R1A* | 605983 | Mental retardation, autosomal dominant 36, 616362 (3), Autosomal dominant |
| *PPP2R1B* | 603113 | Lung cancer, somatic, 211980 (3) |
| *PPP2R2B* | 604325 | Spinocerebellar ataxia 12, 604326 (3), Autosomal dominant |
| *PPP2R3C* | 615902 | Gonadal dysgenesis, dysmorphic facies, retinal dystrophy, and myopathy, 618419 (3), Autosomal recessive; Spermatogenic failure 36, 618420 (3), Autosomal dominant |
| *PPP2R5D* | 601646 | Mental retardation, autosomal dominant 35, 616355 (3), Autosomal dominant |
| *PPP3CA* | 114105 | Arthrogryposis, cleft palate, craniosynostosis, and impaired intellectual development, 618265 (3), Autosomal dominant; Developmental and epileptic encephalopathy 91, 617711 (3), Autosomal dominant |
| *PPT1* | 600722 | Ceroid lipofuscinosis, neuronal, 1, 256730 (3), Autosomal recessive |
| *PQBP1* | 300463 | Renpenning syndrome, 309500 (3), X-linked recessive |
| *PRCC* | 179755 | Renal cell carcinoma, papillary, 605074 (3) |
| *PRCD* | 610598 | Retinitis pigmentosa 36, 610599 (3) |
| *PRDM12* | 616458 | Neuropathy, hereditary sensory and autonomic, type VIII, 616488 (3), Autosomal recessive |
| *PRDM16* | 605557 | Left ventricular noncompaction 8, 615373 (3), Autosomal dominant; Cardiomyopathy, dilated, 1LL, 615373 (3), Autosomal dominant |
| *PRDM5* | 614161 | Brittle cornea syndrome 2, 614170 (3), Autosomal recessive |
| *PRDM6* | 616982 | Patent ductus arteriosus 3, 617039 (3), Autosomal dominant |
| *PRDM8* | 616639 | ?Epilepsy, progressive myoclonic, 10, 616640 (3), Autosomal recessive |
| *PRDX1* | 176763 | Methylmalonic aciduria and homocystinuria, cblC type, digenic, 277400 (3), Autosomal recessive |
| *PREPL* | 609557 | Myasthenic syndrome, congenital, 22, 616224 (3), Autosomal recessive |
| *PRF1* | 170280 | Hemophagocytic lymphohistiocytosis, familial, 2, 603553 (3), Autosomal recessive; Aplastic anemia, 609135 (3); Lymphoma, non-Hodgkin, 605027 (3) |
| *PRG4* | 604283 | Camptodactyly-arthropathy-coxa vara-pericarditis syndrome, 208250 (3), Autosomal recessive |
| *PRICKLE1* | 608500 | Epilepsy, progressive myoclonic 1B, 612437 (3), Autosomal recessive |
| *PRICKLE3* | 300111 | {Leber hereditary optic neuropathy, modifier of}, 308905 (3), X-linked dominant |
| *PRIMPOL* | 615421 | Myopia 22, autosomal dominant, 615420 (3), Autosomal dominant |
| *PRKACA* | 601639 | Cushing syndrome, ACTH-independent adrenal, somatic, 615830 (3); Cardioacrofacial dysplasia 1, 619142 (3), Autosomal dominant |
| *PRKACB* | 176892 | Cardioacrofacial dysplasia 2, 619143 (3) |
| *PRKACG* | 176893 | ?Bleeding disorder, platelet-type, 19, 616176 (3), Autosomal recessive |
| *PRKAG2* | 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 |
| *PRKAG3* | 604976 | [Skeletal muscle glycogen content and metabolism QTL], 619030 (3), Autosomal recessive |
| *PRKAR1A* | 188830 | Pigmented nodular adrenocortical disease, primary, 1, 610489 (3), Autosomal dominant; Acrodysostosis 1, with or without hormone resistance, 101800 (3), Autosomal dominant; Adrenocortical tumor, somatic (3); Carney complex, type 1, 160980 (3), Autosomal dominant; Myxoma, intracardiac, 255960 (3), Autosomal dominant |
| *PRKCA* | 176960 | Pituitary tumor, invasive (3) |
| *PRKCD* | 176977 | Autoimmune lymphoproliferative syndrome, type III, 615559 (3), Autosomal recessive |
| *PRKCG* | 176980 | Spinocerebellar ataxia 14, 605361 (3), Autosomal dominant |
| *PRKCH* | 605437 | {Cerebral infarction, susceptibility to}, 601367 (3), Multifactorial |
| *PRKCSH* | 177060 | Polycystic liver disease 1, 174050 (3), Autosomal dominant |
| *PRKD1* | 605435 | Congenital heart defects and ectodermal dysplasia, 617364 (3), Autosomal dominant |
| *PRKDC* | 600899 | Immunodeficiency 26, with or without neurologic abnormalities, 615966 (3), Autosomal recessive |
| *PRKG1* | 176894 | Aortic aneurysm, familial thoracic 8, 615436 (3), Autosomal dominant |
| *PRKN* | 602544 | Adenocarcinoma of lung, somatic, 211980 (3); Parkinson disease, juvenile, type 2, 600116 (3), Autosomal recessive; Ovarian cancer, somatic, 167000 (3) |
| *PRKRA* | 603424 | Dystonia 16, 612067 (3), Autosomal recessive |
| *PRLR* | 176761 | Multiple fibroadenomas of the breast, 615554 (3), Autosomal dominant; Hyperprolactinemia, 615555 (3), Autosomal recessive, Autosomal dominant |
| *PRMT7* | 610087 | Short stature, brachydactyly, intellectual developmental disability, and seizures, 617157 (3), Autosomal recessive |
| *PRNP* | 176640 | Spongiform encephalopathy with neuropsychiatric features, 606688 (3), Autosomal dominant; Gerstmann-Straussler disease, 137440 (3), Autosomal dominant; Huntington disease-like 1, 603218 (3), Autosomal dominant; Insomnia, fatal familial, 600072 (3), Autosomal dominant; {Kuru, susceptibility to}, 245300 (3); Cerebral amyloid angiopathy, PRNP-related, 137440 (3), Autosomal dominant; Creutzfeldt-Jakob disease, 123400 (3), Autosomal dominant |
| *PROC* | 612283 | Thrombophilia due to protein C deficiency, autosomal recessive, 612304 (3), Autosomal recessive; Thrombophilia due to protein C deficiency, autosomal dominant, 176860 (3), Autosomal dominant |
| *PRODH* | 606810 | {Schizophrenia, susceptibility to, 4}, 600850 (3), Autosomal dominant; Hyperprolinemia, type I, 239500 (3), Autosomal recessive |
| *PROK2* | 607002 | Hypogonadotropic hypogonadism 4 with or without anosmia, 610628 (3), Autosomal dominant |
| *PROKR2* | 607123 | Hypogonadotropic hypogonadism 3 with or without anosmia, 244200 (3), Autosomal dominant |
| *PROM1* | 604365 | Macular dystrophy, retinal, 2, 608051 (3), Autosomal dominant; Retinitis pigmentosa 41, 612095 (3), Autosomal recessive; Stargardt disease 4, 603786 (3), Autosomal dominant; Cone-rod dystrophy 12, 612657 (3), Autosomal recessive, Autosomal dominant |
| *PROP1* | 601538 | Pituitary hormone deficiency, combined, 2, 262600 (3), Autosomal recessive |
| *PROS1* | 176880 | Thrombophilia due to protein S deficiency, autosomal dominant, 612336 (3), Autosomal dominant; Thrombophilia due to protein S deficiency, autosomal recessive, 614514 (3), Autosomal recessive |
| *PROZ* | 176895 | [Protein Z deficiency], 614024 (3) |
| *PRPF3* | 607301 | Retinitis pigmentosa 18, 601414 (3), Autosomal dominant |
| *PRPF31* | 606419 | Retinitis pigmentosa 11, 600138 (3), Autosomal dominant |
| *PRPF4* | 607795 | Retinitis pigmentosa 70, 615922 (3), Autosomal dominant |
| *PRPF6* | 613979 | Retinitis pigmentosa 60, 613983 (3), Autosomal dominant |
| *PRPF8* | 607300 | Retinitis pigmentosa 13, 600059 (3), Autosomal dominant |
| *PRPH* | 170710 | {Amyotrophic lateral sclerosis, susceptibility to}, 105400 (3), Autosomal recessive, Autosomal dominant |
| *PRPH2* | 179605 | Macular dystrophy, patterned, 1, 169150 (3), Autosomal dominant; Choroidal dystrophy, central areolar 2, 613105 (3), Autosomal dominant; Retinitis punctata albescens, 136880 (3), Autosomal recessive, Autosomal dominant; Leber congenital amaurosis 18, 608133 (3), Digenic dominant, Autosomal recessive, Autosomal dominant; Macular dystrophy, vitelliform, 3, 608161 (3), Autosomal dominant; Retinitis pigmentosa 7 and digenic form, 608133 (3), Digenic dominant, Autosomal recessive, Autosomal dominant |
| *PRPS1* | 311850 | Arts syndrome, 301835 (3), X-linked recessive; Phosphoribosylpyrophosphate synthetase superactivity, 300661 (3), X-linked recessive; Charcot-Marie-Tooth disease, X-linked recessive, 5, 311070 (3), X-linked recessive; Deafness, X-linked 1, 304500 (3), X-linked; Gout, PRPS-related, 300661 (3), X-linked recessive |
| *PRRT2* | 614386 | Convulsions, familial infantile, with paroxysmal choreoathetosis, 602066 (3), Autosomal dominant; Seizures, benign familial infantile, 2, 605751 (3), Autosomal dominant; Episodic kinesigenic dyskinesia 1, 128200 (3), Autosomal dominant |
| *PRRX1* | 167420 | Agnathia-otocephaly complex, 202650 (3), Autosomal recessive, Autosomal dominant |
| *PRSS1* | 276000 | Pancreatitis, hereditary, 167800 (3), Autosomal dominant |
| *PRSS12* | 606709 | Mental retardation, autosomal recessive 1, 249500 (3), Autosomal recessive |
| *PRSS2* | 601564 | {Pancreatitis, chronic, protection against}, 167800 (3), Autosomal dominant |
| *PRSS56* | 613858 | Microphthalmia, isolated 6, 613517 (3), Autosomal recessive |
| *PRUNE1* | 617413 | Neurodevelopmental disorder with microcephaly, hypotonia, and variable brain anomalies, 617481 (3), Autosomal recessive |
| *PRX* | 605725 | Charcot-Marie-Tooth disease, type 4F, 614895 (3), Autosomal recessive; Dejerine-Sottas disease, 145900 (3), Autosomal recessive, Autosomal dominant |
| *PSAP* | 176801 | Combined SAP deficiency, 611721 (3), Autosomal recessive; Krabbe disease, atypical, 611722 (3), Autosomal recessive; Metachromatic leukodystrophy due to SAP-b deficiency, 249900 (3), Autosomal recessive; Gaucher disease, atypical, 610539 (3) |
| *PSAT1* | 610936 | Neu-Laxova syndrome 2, 616038 (3), Autosomal recessive; ?Phosphoserine aminotransferase deficiency, 610992 (3), Autosomal recessive |
| *PSEN1* | 104311 | Pick disease, 172700 (3), Autosomal dominant; Alzheimer disease, type 3, with spastic paraparesis and apraxia, 607822 (3), Autosomal dominant; Dementia, frontotemporal, 600274 (3), Autosomal dominant; ?Acne inversa, familial, 3, 613737 (3), Autosomal dominant; Cardiomyopathy, dilated, 1U, 613694 (3), Autosomal dominant; Alzheimer disease, type 3, with spastic paraparesis and unusual plaques, 607822 (3), Autosomal dominant; Alzheimer disease, type 3, 607822 (3), Autosomal dominant |
| *PSEN2* | 600759 | Alzheimer disease-4, 606889 (3), Autosomal dominant; Cardiomyopathy, dilated, 1V, 613697 (3), Autosomal dominant |
| *PSENEN* | 607632 | Acne inversa, familial, 2, with or without Dowling-Degos disease, 613736 (3), Autosomal dominant |
| *PSMA6* | 602855 | {Myocardial infarction, susceptibility to}, 608446 (3) |
| *PSMB10* | 176847 | Proteasome-associated autoinflammatory syndrome 5, 619175 (3), Autosomal recessive |
| *PSMB4* | 602177 | ?Proteasome-associated autoinflammatory syndrome 3 and digenic forms, 617591 (3), Autosomal recessive |
| *PSMB8* | 177046 | Proteasome-associated autoinflammatory syndrome 1 and digenic forms, 256040 (3), Autosomal recessive |
| *PSMB9* | 177045 | ?Proteasome-associated autoinflammatory syndrome 3, digenic, 617591 (3), Autosomal recessive |
| *PSMC3* | 186852 | ?Deafness, cataract, impaired intellectual development, and polyneuropathy, 619354 (3), Autosomal recessive |
| *PSMC3IP* | 608665 | Ovarian dysgenesis 3, 614324 (3), Autosomal recessive |
| *PSMD12* | 604450 | Stankiewicz-Isidor syndrome, 617516 (3), Autosomal dominant |
| *PSMG2* | 609702 | ?Proteasome-associated autoinflammatory syndrome 4, 619183 (3), Autosomal recessive |
| *PSPH* | 172480 | Phosphoserine phosphatase deficiency, 614023 (3), Autosomal recessive |
| *PSTPIP1* | 606347 | Pyogenic sterile arthritis, pyoderma gangrenosum, and acne, 604416 (3), Autosomal dominant |
| *PTCD3* | 614918 | ?Combined oxidative phosphorylation deficiency 51, 619057 (3), Autosomal recessive |
| *PTCH1* | 601309 | Basal cell carcinoma, somatic, 605462 (3); Holoprosencephaly 7, 610828 (3), Autosomal dominant; Basal cell nevus syndrome, 109400 (3), Autosomal dominant |
| *PTCH2* | 603673 | Medulloblastoma, somatic, 155255 (3); Basal cell nevus syndrome, 109400 (3), Autosomal dominant; Basal cell carcinoma, somatic, 605462 (3) |
| *PTCHD1* | 300828 | {Autism, susceptibility to, X-linked 4}, 300830 (3), X-linked recessive |
| *PTDSS1* | 612792 | Lenz-Majewski hyperostotic dwarfism, 151050 (3), Autosomal dominant |
| *PTEN* | 601728 | {Glioma susceptibility 2}, 613028 (3); {Meningioma}, 607174 (3), Autosomal dominant; Lhermitte-Duclos syndrome, 158350 (3), Autosomal dominant; Cowden syndrome 1, 158350 (3), Autosomal dominant; Prostate cancer, somatic, 176807 (3); Macrocephaly/autism syndrome, 605309 (3), Autosomal dominant |
| *PTF1A* | 607194 | Pancreatic and cerebellar agenesis, 609069 (3), Autosomal recessive; Pancreatic agenesis 2, 615935 (3), Autosomal recessive |
| *PTGDR* | 604687 | {Asthma, susceptibility to, 1}, 607277 (3) |
| *PTGER2* | 176804 | {Asthma, aspirin-induced, susceptibility to}, 208550 (3), Autosomal recessive |
| *PTGIS* | 601699 | Hypertension, essential, 145500 (3), Multifactorial |
| *PTH* | 168450 | Hypoparathyroidism, familial isolated 1, 146200 (3), Autosomal recessive, Autosomal dominant |
| *PTH1R* | 168468 | Metaphyseal chondrodysplasia, Murk Jansen type, 156400 (3), Autosomal dominant; Eiken syndrome, 600002 (3), Autosomal recessive; Failure of tooth eruption, primary, 125350 (3), Autosomal dominant; Chondrodysplasia, Blomstrand type, 215045 (3), Autosomal recessive |
| *PTHLH* | 168470 | Brachydactyly, type E2, 613382 (3), Autosomal dominant |
| *PTPN1* | 176885 | {Insulin resistance, susceptibility to}, 125853 (3), Autosomal dominant |
| *PTPN11* | 176876 | Noonan syndrome 1, 163950 (3), Autosomal dominant; LEOPARD syndrome 1, 151100 (3), Autosomal dominant; Metachondromatosis, 156250 (3), Autosomal dominant; Leukemia, juvenile myelomonocytic, somatic, 607785 (3) |
| *PTPN12* | 600079 | Colon cancer, somatic, 114500 (3) |
| *PTPN14* | 603155 | Choanal atresia and lymphedema, 613611 (3), Autosomal recessive |
| *PTPN22* | 600716 | {Rheumatoid arthritis, susceptibility to}, 180300 (3); {Systemic lupus erythematosus susceptibility to}, 152700 (3), Autosomal dominant; {Diabetes, type 1, susceptibility to}, 222100 (3), Autosomal recessive |
| *PTPN23* | 606584 | Neurodevelopmental disorder and structural brain anomalies with or without seizures and spasticity, 618890 (3), Autosomal recessive |
| *PTPRC* | 151460 | Severe combined immunodeficiency, T cell-negative, B-cell/natural killer-cell positive, 608971 (3), Autosomal recessive; {Hepatitis C virus, susceptibility to}, 609532 (3) |
| *PTPRF* | 179590 | ?Breasts and/or nipples, aplasia or hypoplasia of, 2, 616001 (3), Autosomal recessive |
| *PTPRJ* | 600925 | Colon cancer, somatic, 114500 (3) |
| *PTPRO* | 600579 | Nephrotic syndrome, type 6, 614196 (3), Autosomal recessive |
| *PTPRQ* | 603317 | Deafness, autosomal dominant 73, 617663 (3), Autosomal dominant; Deafness, autosomal recessive 84A, 613391 (3), Autosomal recessive |
| *PTRH2* | 608625 | Infantile-onset multisystem neurologic, endocrine, and pancreatic disease, 616263 (3), Autosomal recessive |
| *PTS* | 612719 | Hyperphenylalaninemia, BH4-deficient, A, 261640 (3), Autosomal recessive |
| *PUF60* | 604819 | Verheij syndrome, 615583 (3), Autosomal dominant |
| *PUM1* | 607204 | Spinocerebellar ataxia 47, 617931 (3), Autosomal dominant |
| *PURA* | 600473 | Mental retardation, autosomal dominant 31, 616158 (3), Autosomal dominant |
| *PUS1* | 608109 | Myopathy, lactic acidosis, and sideroblastic anemia 1, 600462 (3), Autosomal recessive |
| *PUS3* | 616283 | Neurodevelopmental disorder with microcephaly and gray sclerae, 617051 (3), Autosomal recessive |
| *PUS7* | 616261 | Intellectual developmental disorder with abnormal behavior, microcephaly, and short stature, 618342 (3), Autosomal recessive |
| *PXDN* | 605158 | Anterior segment dysgenesis 7, with sclerocornea, 269400 (3), Autosomal recessive |
| *PYCR1* | 179035 | Cutis laxa, autosomal recessive, type IIIB, 614438 (3); Cutis laxa, autosomal recessive, type IIB, 612940 (3), Autosomal recessive |
| *PYCR2* | 616406 | Leukodystrophy, hypomyelinating, 10, 616420 (3), Autosomal recessive |
| *PYGL* | 613741 | Glycogen storage disease VI, 232700 (3), Autosomal recessive |
| *PYGM* | 608455 | McArdle disease, 232600 (3), Autosomal recessive |
| *PYROXD1* | 617220 | Myopathy, myofibrillar, 8, 617258 (3), Autosomal recessive |
| *QARS1 (QARS)* | 603727 | Microcephaly, progressive, seizures, and cerebral and cerebellar atrophy, 615760 (3), Autosomal recessive |
| *QDPR* | 612676 | Hyperphenylalaninemia, BH4-deficient, C, 261630 (3), Autosomal recessive |
| *QRICH1* | 617387 | Ververi-Brady syndrome, 617982 (3), Autosomal dominant |
| *QRICH2* | 618304 | Spermatogenic failure 35, 618341 (3), Autosomal recessive |
| *QRSL1* | 617209 | Combined oxidative phosphorylation deficiency 40, 618835 (3), Autosomal recessive |
| *RAB11B* | 604198 | Neurodevelopmental disorder with ataxic gait, absent speech, and decreased cortical white matter, 617807 (3), Autosomal dominant |
| *RAB18* | 602207 | Warburg micro syndrome 3, 614222 (3), Autosomal recessive |
| *RAB23* | 606144 | Carpenter syndrome, 201000 (3), Autosomal recessive |
| *RAB27A* | 603868 | Griscelli syndrome, type 2, 607624 (3), Autosomal recessive |
| *RAB28* | 612994 | Cone-rod dystrophy 18, 615374 (3), Autosomal recessive |
| *RAB33B* | 605950 | Smith-McCort dysplasia 2, 615222 (3), Autosomal recessive |
| *RAB39B* | 300774 | Waisman syndrome, 311510 (3), X-linked recessive; Mental retardation, X-linked 72, 300271 (3), X-linked recessive |
| *RAB3GAP1* | 602536 | Martsolf syndrome 2, 619420 (3), Autosomal recessive; Warburg micro syndrome 1, 600118 (3), Autosomal recessive |
| *RAB3GAP2* | 609275 | Martsolf syndrome 1, 212720 (3), Autosomal recessive; Warburg micro syndrome 2, 614225 (3), Autosomal recessive |
| *RAB7A* | 602298 | Charcot-Marie-Tooth disease, type 2B, 600882 (3), Autosomal dominant |
| *RABL3* | 618542 | {?Pancreatic cancer, susceptibility to, 5}, 618680 (3), Autosomal dominant |
| *RAC1* | 602048 | Mental retardation, autosomal dominant 48, 617751 (3), Autosomal dominant |
| *RAC2* | 602049 | Immunodeficiency 73A with defective neutrophil chemotaxix and leukocytosis, 608203 (3), Autosomal dominant; ?Immunodeficiency 73C with defective neutrophil chemotaxis and hypogammaglobulinemia, 618987 (3); Immunodeficiency 73B with defective neutrophil chemotaxis and lymphopenia, 618986 (3) |
| *RAC3* | 602050 | Neurodevelopmental disorder with structural brain anomalies and dysmorphic facies, 618577 (3), Autosomal dominant |
| *RAD21* | 606462 | Cornelia de Lange syndrome 4, 614701 (3), Autosomal dominant; ?Mungan syndrome, 611376 (3), Autosomal recessive |
| *RAD50* | 604040 | Nijmegen breakage syndrome-like disorder, 613078 (3), Autosomal recessive |
| *RAD51* | 179617 | Mirror movements 2, 614508 (3), Autosomal dominant; {Breast cancer, susceptibility to}, 114480 (3), Somatic mutation, Autosomal dominant; Fanconi anemia, complementation group R, 617244 (3), Autosomal dominant |
| *RAD51C* | 602774 | {Breast-ovarian cancer, familial, susceptibility to, 3}, 613399 (3); Fanconi anemia, complementation group O, 613390 (3), Autosomal recessive |
| *RAD51D* | 602954 | {Breast-ovarian cancer, familial, susceptibility to, 4}, 614291 (3) |
| *RAD54B* | 604289 | Colon cancer, somatic, 114500 (3); Lymphoma, non-Hodgkin, somatic, 605027 (3) |
| *RAD54L* | 603615 | {Breast cancer, invasive ductal}, 114480 (3), Somatic mutation, Autosomal dominant; Adenocarcinoma, colonic, somatic (3); Lymphoma, non-Hodgkin, somatic, 605027 (3) |
| *RAF1* | 164760 | Cardiomyopathy, dilated, 1NN, 615916 (3), Autosomal dominant; Noonan syndrome 5, 611553 (3), Autosomal dominant; LEOPARD syndrome 2, 611554 (3) |
| *RAG1* | 179615 | Omenn syndrome, 603554 (3), Autosomal recessive; Severe combined immunodeficiency, B cell-negative, 601457 (3), Autosomal recessive; Combined cellular and humoral immune defects with granulomas, 233650 (3), Autosomal recessive; Alpha/beta T-cell lymphopenia with gamma/delta T-cell expansion, severe cytomegalovirus infection, and autoimmunity, 609889 (3) |
| *RAG2* | 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 |
| *RAI1* | 607642 | Smith-Magenis syndrome, 182290 (3), Isolated cases, Autosomal dominant |
| *RALA* | 179550 | Hiatt-Neu-Cooper neurodevelopmental syndrome, 619311 (3), Autosomal dominant |
| *RALGAPA1* | 608884 | Neurodevelopmental disorder with hypotonia, neonatal respiratory insufficiency, and thermodysregulation, 618797 (3), Autosomal recessive |
| *RANBP2* | 601181 | {Encephalopathy, acute, infection-induced, 3, susceptibility to}, 608033 (3), Autosomal dominant |
| *RAP1GDS1* | 179502 | Lymphocytic leukemia, acute T-cell (3) |
| *RAPGEF2* | 609530 | ?Epilepsy, familial adult myoclonic, 7, 618075 (3), Autosomal dominant |
| *RAPSN* | 601592 | Fetal akinesia deformation sequence 2, 618388 (3), Autosomal recessive; Myasthenic syndrome, congenital, 11, associated with acetylcholine receptor deficiency, 616326 (3), Autosomal recessive |
| *RARA* | 180240 | Leukemia, acute promyelocytic, 612376 (1) |
| *RARB* | 180220 | Microphthalmia, syndromic 12, 615524 (3), Autosomal recessive, Autosomal dominant |
| *RARS1 (RARS)* | 107820 | Leukodystrophy, hypomyelinating, 9, 616140 (3), Autosomal recessive |
| *RARS2* | 611524 | Pontocerebellar hypoplasia, type 6, 611523 (3), Autosomal recessive |
| *RASA1* | 139150 | Capillary malformation-arteriovenous malformation 1, 608354 (3), Autosomal dominant; Basal cell carcinoma, somatic, 605462 (3) |
| *RASGRP1* | 603962 | Immunodeficiency 64, 618534 (3), Autosomal recessive |
| *RASGRP2* | 605577 | ?Bleeding disorder, platelet-type, 18, 615888 (3), Autosomal recessive |
| *RAX* | 601881 | Microphthalmia, isolated 3, 611038 (3), Autosomal recessive |
| *RAX2* | 610362 | Cone-rod dystrophy 11, 610381 (3), Autosomal dominant; ?Macular degeneration, age-related, 6, 613757 (3) |
| *RB1* | 614041 | Small cell cancer of the lung, somatic, 182280 (3); Bladder cancer, somatic, 109800 (3); Retinoblastoma, trilateral, 180200 (3), Somatic mutation, Autosomal dominant; Osteosarcoma, somatic, 259500 (3); Retinoblastoma, 180200 (3), Somatic mutation, Autosomal dominant |
| *RB1CC1* | 606837 | Breast cancer, somatic, 114480 (3) |
| *RBBP8* | 604124 | Seckel syndrome 2, 606744 (3), Autosomal recessive; Jawad syndrome, 251255 (3), Autosomal recessive; Pancreatic carcinoma, somatic (3) |
| *RBCK1* | 610924 | Polyglucosan body myopathy 1 with or without immunodeficiency, 615895 (3), Autosomal recessive |
| *RBM10* | 300080 | TARP syndrome, 311900 (3), X-linked recessive |
| *RBM12* | 607179 | {Schizophrenia 19, susceptibility to}, 617629 (3), Autosomal dominant |
| *RBM15* | 606077 | Megakaryoblastic leukemia, acute, 606077 (2) |
| *RBM20* | 613171 | Cardiomyopathy, dilated, 1DD, 613172 (3), Autosomal dominant |
| *RBM28* | 612074 | ?Alopecia, neurologic defects, and endocrinopathy syndrome, 612079 (3), Autosomal recessive |
| *RBM8A* | 605313 | Thrombocytopenia-absent radius syndrome, 274000 (3), Autosomal recessive |
| *RBMX* | 300199 | ?Mental retardation, X-linked, syndromic 11, Shashi type, 300238 (3), X-linked recessive |
| *RBP3* | 180290 | ?Retinitis pigmentosa 66, 615233 (3), Autosomal recessive |
| *RBP4* | 180250 | Microphthalmia, isolated, with coloboma 10, 616428 (3), Autosomal dominant; Retinal dystrophy, iris coloboma, and comedogenic acne syndrome, 615147 (3), Autosomal recessive |
| *RBPJ* | 147183 | Adams-Oliver syndrome 3, 614814 (3), Autosomal dominant |
| *RC3H1* | 609424 | ?Immune dysregulation and systemic hyperinflammation syndrome, 618998 (3) |
| *RCBTB1* | 607867 | Retinal dystrophy with or without extraocular anomalies, 617175 (3), Autosomal recessive |
| *RD3* | 180040 | Leber congenital amaurosis 12, 610612 (3), Autosomal recessive |
| *RDH11* | 607849 | ?Retinal dystrophy, juvenile cataracts, and short stature syndrome, 616108 (3), Autosomal recessive |
| *RDH12* | 608830 | Leber congenital amaurosis 13, 612712 (3), Autosomal recessive, Autosomal dominant |
| *RDH5* | 601617 | Fundus albipunctatus, 136880 (3), Autosomal recessive, Autosomal dominant |
| *RDX* | 179410 | Deafness, autosomal recessive 24, 611022 (3), Autosomal recessive |
| *REC114* | 618421 | Oocyte maturation defect 10, 619176 (3), Autosomal recessive |
| *RECQL4* | 603780 | Baller-Gerold syndrome, 218600 (3), Autosomal recessive; Rothmund-Thomson syndrome, type 2, 268400 (3), Autosomal recessive; RAPADILINO syndrome, 266280 (3), Autosomal recessive |
| *REEP1* | 609139 | ?Neuronopathy, distal hereditary motor, type VB, 614751 (3), Autosomal dominant; Spastic paraplegia 31, autosomal dominant, 610250 (3), Autosomal dominant |
| *REEP2* | 609347 | ?Spastic paraplegia 72, autosomal dominant, 615625 (3), Autosomal recessive, Autosomal dominant; ?Spastic paraplegia 72, autosomal recessive, 615625 (3), Autosomal recessive, Autosomal dominant |
| *REEP6* | 609346 | Retinitis pigmentosa 77, 617304 (3), Autosomal recessive |
| *RELA* | 164014 | ?Mucocutaneous ulceration, chronic, 618287 (3), Autosomal dominant |
| *RELB* | 604758 | ?Immunodeficiency 53, 617585 (3), Autosomal recessive |
| *RELN* | 600514 | {Epilepsy, familial temporal lobe, 7}, 616436 (3), Autosomal dominant; Lissencephaly 2 (Norman-Roberts type), 257320 (3), Autosomal recessive |
| *RELT* | 611211 | Amelogenesis imperfecta, type IIIC, 618386 (3), Autosomal recessive |
| *REN* | 179820 | Renal tubular dysgenesis, 267430 (3), Autosomal recessive; [Hyperproreninemia] (3); Tubulointerstitial kidney disease, autosomal dominant, 4, 613092 (3), Autosomal dominant |
| *REPS1* | 614825 | ?Neurodegeneration with brain iron accumulation 7, 617916 (3), Autosomal recessive |
| *RERE* | 605226 | Neurodevelopmental disorder with or without anomalies of the brain, eye, or heart, 616975 (3), Autosomal dominant |
| *REST* | 600571 | ?Deafness, autosomal dominant 27, 612431 (3), Autosomal dominant; {Wilms tumor 6, susceptibility to}, 616806 (3), Autosomal dominant; Fibromatosis, gingival, 5, 617626 (3), Autosomal dominant |
| *RET* | 164761 | {Hirschsprung disease, susceptibility to, 1}, 142623 (3), Autosomal dominant; Multiple endocrine neoplasia IIA, 171400 (3), Autosomal dominant; Central hypoventilation syndrome, congenital, 209880 (3), Autosomal dominant; {Hirschsprung disease, protection against}, 142623 (3), Autosomal dominant; Medullary thyroid carcinoma, 155240 (3), Autosomal dominant; Pheochromocytoma, 171300 (3), Autosomal dominant; Multiple endocrine neoplasia IIB, 162300 (3), Autosomal dominant |
| *RETN* | 605565 | {Hypertension, insulin resistance-related, susceptibility to}, 125853 (3), Autosomal dominant; {Diabetes mellitus, noninsulin-dependent, susceptibility to}, 125853 (3), Autosomal dominant |
| *RETREG1* | 613114 | Neuropathy, hereditary sensory and autonomic, type IIB, 613115 (3), Autosomal recessive |
| *RFC1* | 102579 | Cerebellar ataxia, neuropathy, and vestibular areflexia syndrome, 614575 (3), Autosomal recessive |
| *RFT1* | 611908 | Congenital disorder of glycosylation, type In, 612015 (3), Autosomal recessive |
| *RFWD3* | 614151 | ?Fanconi anemia, complementation group W, 617784 (3), Autosomal recessive |
| *RFX5* | 601863 | Bare lymphocyte syndrome, type II, complementation group C, 209920 (3), Autosomal recessive; Bare lymphocyte syndrome, type II, complementation group E, 209920 (3), Autosomal recessive |
| *RFX6* | 612659 | Mitchell-Riley syndrome, 615710 (3), Autosomal recessive |
| *RFXANK* | 603200 | MHC class II deficiency, complementation group B, 209920 (3), Autosomal recessive |
| *RFXAP* | 601861 | Bare lymphocyte syndrome, type II, complementation group D, 209920 (3), Autosomal recessive |
| *RGR* | 600342 | Retinitis pigmentosa 44, 613769 (3) |
| *RGS5* | 603276 | [Blood pressure regulation QTL], 145500 (2), Multifactorial |
| *RGS9* | 604067 | Bradyopsia, 608415 (3) |
| *RGS9BP* | 607814 | Bradyopsia, 608415 (3) |
| *RHAG* | 180297 | Overhydrated hereditary stomatocytosis, 185000 (3), Autosomal dominant; Anemia, hemolytic, Rh-null, regulator type, 268150 (3), Autosomal dominant |
| *RHBDF2* | 614404 | Tylosis with esophageal cancer, 148500 (3), Autosomal dominant |
| *RHCE* | 111700 | Rh-null disease, amorph type, 617970 (3); [Blood group, Rhesus] (3) |
| *RHD* | 111680 | [Rh-negative blood type] (3) |
| *RHO* | 180380 | Night blindness, congenital stationary, autosomal dominant 1, 610445 (3); Retinitis pigmentosa 4, autosomal dominant or recessive, 613731 (3), Autosomal recessive, Autosomal dominant; Retinitis punctata albescens, 136880 (3), Autosomal recessive, Autosomal dominant |
| *RHOA* | 165390 | Ectodermal dysplasia with facial dysmorphism and acral, ocular, and brain anomalies, somatic mosaic, 618727 (3) |
| *RHOBTB2* | 607352 | Developmental and epileptic encephalopathy 64, 618004 (3), Autosomal dominant |
| *RHOH* | 602037 | {?Epidermodysplasia verruciformis, susceptibility to, 4}, 618307 (3), Autosomal recessive |
| *RIC1* | 610354 | CATIFA syndrome, 618761 (3), Autosomal recessive |
| *RIMS1* | 606629 | Cone-rod dystrophy 7, 603649 (3), Autosomal dominant |
| *RIMS2* | 606630 | Cone-rod synaptic disorder syndrome, congenital nonprogressive, 618970 (3), Autosomal recessive |
| *RIN2* | 610222 | Macrocephaly, alopecia, cutis laxa, and scoliosis, 613075 (3), Autosomal recessive |
| *RINT1* | 610089 | Infantile liver failure syndrome 3, 618641 (3), Autosomal recessive |
| *RIPK1* | 603453 | Immunodeficiency 57 with autoinflammation, 618108 (3), Autosomal recessive; Autoinflammation with episodic fever and lymphadenopathy, 618852 (3), Autosomal dominant |
| *RIPK4* | 605706 | CHAND syndrome, 214350 (3), Autosomal recessive; Popliteal pterygium syndrome, Bartsocas-Papas type 1, 263650 (3), Autosomal recessive |
| *RIPOR2* | 611410 | ?Deafness, autosomal recessive 104, 616515 (3), Autosomal recessive |
| *RIPPLY2* | 609891 | ?Spondylocostal dysostosis 6, 616566 (3), Autosomal recessive |
| *RIT1* | 609591 | Noonan syndrome 8, 615355 (3), Autosomal dominant |
| *RLBP1* | 180090 | Bothnia retinal dystrophy, 607475 (3), Autosomal recessive; Newfoundland rod-cone dystrophy, 607476 (3); Retinitis punctata albescens, 136880 (3), Autosomal recessive, Autosomal dominant; Fundus albipunctatus, 136880 (3), Autosomal recessive, Autosomal dominant |
| *RLIM* | 300379 | Tonne-Kalscheuer syndrome, 300978 (3), X-linked |
| *RMND1* | 614917 | Combined oxidative phosphorylation deficiency 11, 614922 (3), Autosomal recessive |
| *RNASEH1* | 604123 | Progressive external ophthalmoplegia with mitochondrial DNA deletions, autosomal recessive 2, 616479 (3), Autosomal recessive |
| *RNASEH2A* | 606034 | Aicardi-Goutieres syndrome 4, 610333 (3), Autosomal recessive |
| *RNASEH2B* | 610326 | Aicardi-Goutieres syndrome 2, 610181 (3), Autosomal recessive |
| *RNASEH2C* | 610330 | Aicardi-Goutieres syndrome 3, 610329 (3), Autosomal recessive |
| *RNASEL* | 180435 | Prostate cancer 1, 601518 (3), Autosomal dominant |
| *RNASET2* | 612944 | Leukoencephalopathy, cystic, without megalencephaly, 612951 (3), Autosomal recessive |
| *RNF113A* | 300951 | Trichothiodystrophy 5, nonphotosensitive, 300953 (3), X-linked dominant |
| *RNF114* | 612451/612950 | -/{Psoriasis susceptibility 12}, 612950 (2) |
| *RNF125* | 610432 | Tenorio syndrome, 616260 (3), Autosomal dominant |
| *RNF13* | 609247 | Developmental and epileptic encephalopathy 73, 618379 (3), Autosomal dominant |
| *RNF139* | 603046 | Renal cell carcinoma, 144700 (3) |
| *RNF168* | 612688 | RIDDLE syndrome, 611943 (3), Autosomal recessive |
| *RNF170* | 614649 | Ataxia, sensory, 1, autosomal dominant, 608984 (3), Autosomal dominant |
| *RNF212* | 612041 | Recombination rate QTL 1, 612042 (3) |
| *RNF213* | 613768 | {Moyamoya disease 2, susceptibility to}, 607151 (3), Autosomal recessive, Autosomal dominant |
| *RNF216* | 609948 | Cerebellar ataxia and hypogonadotropic hypogonadism, 212840 (3), Autosomal recessive |
| *RNF43* | 612482 | Sessile serrated polyposis cancer syndrome, 617108 (3), Autosomal dominant |
| *RNF6* | 604242 | Esophageal carcinoma, somatic, 133239 (3) |
| *RNPC3* | 618016 | ?Growth hormone deficiency, isolated, type V, 618160 (3), Autosomal recessive |
| *RNU4ATAC* | 601428 | Roifman syndrome, 616651 (3), Autosomal recessive; Lowry-Wood syndrome, 226960 (3), Autosomal recessive; Microcephalic osteodysplastic primordial dwarfism, type I, 210710 (3), Autosomal recessive |
| *ROBO1* | 602430 | Tetralogy of Fallot and septal defects. PMID: 30692597, 31448886, 30530901 |
| *ROBO2* | 602431 | Vesicoureteral reflux 2, 610878 (3), Autosomal dominant |
| *ROBO3* | 608630 | Gaze palsy, familial horizontal, with progressive scoliosis, 1, 607313 (3), Autosomal recessive |
| *ROBO4* | 607528 | Aortic valve disease 3, 618496 (3), Autosomal dominant |
| *ROGDI* | 614574 | Kohlschutter-Tonz syndrome, 226750 (3), Autosomal recessive |
| *ROM1* | 180721 | Retinitis pigmentosa 7, digenic form, 608133 (3), Digenic dominant, Autosomal recessive, Autosomal dominant |
| *ROR1* | 602336 | ?Deafness, autosomal recessive 108, 617654 (3), Autosomal recessive |
| *ROR2* | 602337 | Brachydactyly, type B1, 113000 (3), Autosomal dominant; Robinow syndrome, autosomal recessive, 268310 (3), Autosomal recessive |
| *RORA* | 600825 | Intellectual developmental disorder with or without epilepsy or cerebellar ataxia, 618060 (3), Autosomal dominant |
| *RORB* | 601972 | {Epilepsy, idiopathic generalized, susceptibility to, 15}, 618357 (3), Autosomal dominant |
| *RORC* | 602943 | Immunodeficiency 42, 616622 (3), Autosomal recessive |
| *RP1* | 603937 | Retinitis pigmentosa 1, 180100 (3), Autosomal recessive, Autosomal dominant |
| *RP1L1* | 608581 | Occult macular dystrophy, 613587 (3), Autosomal dominant; Retinitis pigmentosa 88, 618826 (3), Autosomal recessive |
| *RP2* | 300757 | Retinitis pigmentosa 2, 312600 (3), X-linked |
| *RP9* | 607331 | ?Retinitis pigmentosa 9, 180104 (3), Autosomal dominant |
| *RPE65* | 180069 | Retinitis pigmentosa 20, 613794 (3), Autosomal recessive; Retinitis pigmentosa 87 with choroidal involvement, 618697 (3), Autosomal dominant; Leber congenital amaurosis 2, 204100 (3), Autosomal recessive |
| *RPGR* | 312610 | Retinitis pigmentosa, X-linked, and sinorespiratory infections, with or without deafness, 300455 (3); Cone-rod dystrophy, X-linked, 1, 304020 (3), X-linked; Retinitis pigmentosa 3, 300029 (3); Macular degeneration, X-linked atrophic, 300834 (3), X-linked recessive |
| *RPGRIP1* | 605446 | Cone-rod dystrophy 13, 608194 (3); Leber congenital amaurosis 6, 613826 (3), Autosomal recessive |
| *RPGRIP1L* | 610937 | Joubert syndrome 7, 611560 (3), Autosomal recessive; Meckel syndrome 5, 611561 (3), Autosomal recessive; ?COACH syndrome 3, 619113 (3) |
| *RPIA* | 180430 | Ribose 5-phosphate isomerase deficiency, 608611 (3), Autosomal recessive |
| *RPL10* | 312173 | Mental retardation, X-linked, syndromic, 35, 300998 (3), X-linked recessive; {Autism, susceptibility to, X-linked 5}, 300847 (3) |
| *RPL11* | 604175 | Diamond-Blackfan anemia 7, 612562 (3), Autosomal dominant |
| *RPL13* | 113703 | Spondyloepimetaphyseal dysplasia, Isidor-Toutain type, 618728 (3), Autosomal dominant |
| *RPL15* | 604174 | ?Diamond-Blackfan anemia 12, 615550 (3), Autosomal dominant |
| *RPL18* | 604179 | ?Diamond-Blackfan anemia 18, 618310 (3), Autosomal dominant |
| *RPL21* | 603636 | Hypotrichosis 12, 615885 (3), Autosomal dominant |
| *RPL26* | 603704 | ?Diamond-Blackfan anemia 11, 614900 (3), Autosomal dominant |
| *RPL27* | 607526 | ?Diamond-Blackfan anemia 16, 617408 (3), Autosomal dominant |
| *RPL35* | 618315 | ?Diamond-Blackfan anemia 19, 618312 (3), Autosomal dominant |
| *RPL35A* | 180468 | Diamond-Blackfan anemia 5, 612528 (3), Autosomal dominant |
| *RPL3L* | 617416 | Cardiomyopathy, dilated, 2D, 619371 (3), Autosomal recessive |
| *RPL5* | 603634 | Diamond-Blackfan anemia 6, 612561 (3), Autosomal dominant |
| *RPS10* | 603632 | Diamond-Blackfan anemia 9, 613308 (3), Autosomal dominant |
| *RPS14* | 130620 | Macrocytic anemia, refractory, due to 5q deletion, somatic, 153550 (3) |
| *RPS15A* | 603674 | ?Diamond-Blackfan anemia 20, 618313 (3), Autosomal dominant |
| *RPS17* | 180472 | Diamond-Blackfan anemia 4, 612527 (3), Autosomal dominant |
| *RPS19* | 603474 | Diamond-Blackfan anemia 1, 105650 (3), Autosomal dominant |
| *RPS23* | 603683 | Brachycephaly, trichomegaly, and developmental delay, 617412 (3), Autosomal dominant |
| *RPS24* | 602412 | Diamond-blackfan anemia 3, 610629 (3), Autosomal dominant |
| *RPS26* | 603701 | Diamond-Blackfan anemia 10, 613309 (3), Autosomal dominant |
| *RPS27* | 603702 | ?Diamond-Blackfan anemia 17, 617409 (3), Autosomal dominant |
| *RPS28* | 603685 | Diamond Blackfan anemia 15 with mandibulofacial dysostosis, 606164 (3), Autosomal dominant |
| *RPS29* | 603633 | Diamond-Blackfan anemia 13, 615909 (3), Autosomal dominant |
| *RPS6KA3* | 300075 | Mental retardation, X-linked 19, 300844 (3), X-linked dominant; Coffin-Lowry syndrome, 303600 (3), X-linked dominant |
| *RPS7* | 603658 | Diamond-Blackfan anemia 8, 612563 (3), Autosomal dominant |
| *RPSA* | 150370 | Asplenia, isolated congenital, 271400 (3), Autosomal dominant |
| *RRAS2* | 600098 | Ovarian carcinoma (3); Noonan syndrome 12, 618624 (3), Autosomal dominant |
| *RRM2B* | 604712 | Mitochondrial DNA depletion syndrome 8B (MNGIE type), 612075 (3), Autosomal recessive; Mitochondrial DNA depletion syndrome 8A (encephalomyopathic type with renal tubulopathy), 612075 (3), Autosomal recessive; Progressive external ophthalmoplegia with mitochondrial DNA deletions, autosomal dominant 5, 613077 (3), Autosomal dominant |
| *RS1* | 300839 | Retinoschisis, 312700 (3), X-linked recessive |
| *RSPH1* | 609314 | Ciliary dyskinesia, primary, 24, 615481 (3), Autosomal recessive |
| *RSPH3* | 615876 | Ciliary dyskinesia, primary, 32, 616481 (3), Autosomal recessive |
| *RSPH4A* | 612647 | Ciliary dyskinesia, primary, 11, 612649 (3) |
| *RSPH9* | 612648 | Ciliary dyskinesia, primary, 12, 612650 (3) |
| *RSPO1* | 609595 | Palmoplantar hyperkeratosis and true hermaphroditism, 610644 (3), Autosomal recessive; Palmoplantar hyperkeratosis with squamous cell carcinoma of skin and sex reversal, 610644 (3), Autosomal recessive |
| *RSPO2* | 610575 | ?Humerofemoral hypoplasia with radiotibial ray deficiency, 618022 (3), Autosomal recessive; Tetraamelia syndrome 2, 618021 (3), Autosomal recessive |
| *RSPO4* | 610573 | Anonychia congenita, 206800 (3), Autosomal recessive |
| *RSPRY1* | 616585 | Spondyloepimetaphyseal dysplasia, Faden-Alkuraya type, 616723 (3), Autosomal recessive |
| *RSRC1* | 613352 | Intellectual developmental disorder, autosomal recessive 70, 618402 (3), Autosomal recessive |
| *RTEL1* | 608833 | 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; Dyskeratosis congenita, autosomal recessive 5, 615190 (3), Autosomal recessive, Autosomal dominant |
| *RTN2* | 603183 | Spastic paraplegia 12, autosomal dominant, 604805 (3), Autosomal dominant |
| *RTN4IP1* | 610502 | Optic atrophy 10 with or without ataxia, mental retardation, and seizures, 616732 (3), Autosomal recessive |
| *RTN4R* | 605566 | {Schizophrenia, susceptibility to}, 181500 (3), Autosomal dominant |
| *RTTN* | 610436 | Microcephaly, short stature, and polymicrogyria with seizures, 614833 (3), Autosomal recessive |
| *RUBCN* | 613516 | Spinocerebellar ataxia, autosomal recessive 15, 615705 (3), Autosomal recessive |
| *RUNX1* | 151385 | Platelet disorder, familial, with associated myeloid malignancy, 601399 (3), Autosomal dominant; Leukemia, acute myeloid, 601626 (3), Somatic mutation, Autosomal dominant |
| *RUNX2* | 600211 | Metaphyseal dysplasia with maxillary hypoplasia with or without brachydactyly, 156510 (3), Autosomal dominant; Cleidocranial dysplasia, forme fruste, with brachydactyly, 119600 (3), Autosomal dominant; Cleidocranial dysplasia, forme fruste, dental anomalies only, 119600 (3), Autosomal dominant; Cleidocranial dysplasia, 119600 (3), Autosomal dominant |
| *RUSC2* | 611053 | Mental retardation, autosomal recessive 61, 617773 (3), Autosomal recessive |
| *RXYLT1* | 605862 | Muscular dystrophy-dystroglycanopathy (congenital with brain and eye anomalies), type A, 10, 615041 (3), Autosomal recessive |
| *RYR1* | 180901 | Neuromuscular disease, congenital, with uniform type 1 fiber, 117000 (3), Autosomal recessive, Autosomal dominant; 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 |
| *RYR2* | 180902 | Ventricular tachycardia, catecholaminergic polymorphic, 1, 604772 (3), Autosomal dominant; Ventricular arrythmias due to cardiac ryanodine receptor calcium release deficiency syndrome, 115000 (3), Autosomal dominant; Arrhythmogenic right ventricular dysplasia 2, 600996 (3), Autosomal dominant |
| *S1PR2* | 605111 | Deafness, autosomal recessive 68, 610419 (3), Autosomal recessive |
| *SACS* | 604490 | Spastic ataxia, Charlevoix-Saguenay type, 270550 (3), Autosomal recessive |
| *SAG* | 181031 | Retinitis pigmentosa 47, 613758 (3); Oguchi disease-1, 258100 (3), Autosomal recessive |
| *SALL1* | 602218 | Townes-Brocks syndrome 1, 107480 (3), Autosomal dominant; Townes-Brocks branchiootorenal-like syndrome, 107480 (3), Autosomal dominant |
| *SALL2* | 602219 | ?Coloboma, ocular, autosomal recessive, 216820 (3), Autosomal recessive |
| *SALL4* | 607343 | ?IVIC syndrome, 147750 (3), Autosomal dominant; Duane-radial ray syndrome, 607323 (3), Autosomal dominant |
| *SAMD12* | 618073 | Epilepsy, familial adult myoclonic, 1, 601068 (3), Autosomal dominant |
| *SAMD9* | 610456 | Tumoral calcinosis, familial, normophosphatemic, 610455 (3), Autosomal recessive; Monosomy 7 myelodysplasia and leukemia syndrome 2, 619041 (3), Autosomal dominant; MIRAGE syndrome, 617053 (3), Autosomal dominant |
| *SAMD9L* | 611170 | Ataxia-pancytopenia syndrome, 159550 (3), Autosomal dominant; Monosomy 7 myelodysplasia and leukemia syndrome 1, 252270 (3), Autosomal dominant |
| *SAMHD1* | 606754 | ?Chilblain lupus 2, 614415 (3), Autosomal dominant; Aicardi-Goutieres syndrome 5, 612952 (3), Autosomal recessive |
| *SAR1B* | 607690 | Chylomicron retention disease, 246700 (3), Autosomal recessive |
| *SARDH* | 604455 | [Sarcosinemia], 268900 (3), Autosomal recessive |
| *SARS1 (SARS)* | 607529 | ?Neurodevelopmental disorder with microcephaly, ataxia, and seizures, 617709 (3), Autosomal recessive |
| *SARS2* | 612804 | Hyperuricemia, pulmonary hypertension, renal failure, and alkalosis, 613845 (3), Autosomal recessive |
| *SASH1* | 607955 | Dyschromatosis universalis hereditaria 1, 127500 (3), Autosomal dominant; ?Cancer, alopecia, pigment dyscrasia, onychodystrophy, and keratoderma, 618373 (3), Autosomal recessive |
| *SASS6* | 609321 | ?Microcephaly 14, primary, autosomal recessive, 616402 (3), Autosomal recessive |
| *SATB1* | 602075 | Kohlschutter-Tonz syndrome-like, 619229 (3), Autosomal dominant; Developmental delay with dysmorphic facies and dental anomalies, 619228 (3), Autosomal dominant |
| *SATB2* | 608148 | Glass syndrome, 612313 (3), Autosomal dominant |
| *SBDS* | 607444 | {Aplastic anemia, susceptibility to}, 609135 (3); Shwachman-Diamond syndrome, 260400 (3), Autosomal recessive |
| *SBF1* | 603560 | Charcot-Marie-Tooth disease, type 4B3, 615284 (3), Autosomal recessive |
| *SBF2* | 607697 | Charcot-Marie-Tooth disease, type 4B2, 604563 (3), Autosomal recessive |
| *SC5D* | 602286 | Lathosterolosis, 607330 (3), Autosomal recessive |
| *SCAPER* | 611611 | Intellectual developmental disorder and retinitis pigmentosa, 618195 (3), Autosomal recessive |
| *SCARB1* | 601040 | [High density lipoprotein cholesterol level QTL6], 610762 (3) |
| *SCARB2* | 602257 | Epilepsy, progressive myoclonic 4, with or without renal failure, 254900 (3), Autosomal recessive |
| *SCARF2* | 613619 | Van den Ende-Gupta syndrome, 600920 (3), Autosomal recessive |
| *SCD5* | 608370 | ?Deafness, autosomal dominant 79, 619086 (3), Autosomal dominant |
| *SCGB3A2* | 606531 | {Asthma, susceptibility to}, 600807 (3), Autosomal dominant |
| *SCN10A* | 604427 | Episodic pain syndrome, familial, 2, 615551 (3), Autosomal dominant |
| *SCN11A* | 604385 | Episodic pain syndrome, familial, 3, 615552 (3), Autosomal dominant; Neuropathy, hereditary sensory and autonomic, type VII, 615548 (3), Autosomal dominant |
| *SCN1A* | 182389 | Developmental and epileptic encephalopathy 6B, non-Dravet, 619317 (3), Autosomal dominant; Migraine, familial hemiplegic, 3, 609634 (3), Autosomal dominant; Dravet syndrome, 607208 (3), Autosomal dominant; Febrile seizures, familial, 3A, 604403 (3), Autosomal dominant; Epilepsy, generalized, with febrile seizures plus, type 2, 604403 (3), Autosomal dominant |
| *SCN1B* | 600235 | Epilepsy, generalized, with febrile seizures plus, type 1, 604233 (3), Autosomal dominant; Developmental and epileptic encephalopathy 52, 617350 (3), Autosomal recessive; Cardiac conduction defect, nonspecific, 612838 (3); Atrial fibrillation, familial, 13, 615377 (3), Autosomal dominant; Brugada syndrome 5, 612838 (3) |
| *SCN2A* | 182390 | Seizures, benign familial infantile, 3, 607745 (3), Autosomal dominant; Developmental and epileptic encephalopathy 11, 613721 (3), Autosomal dominant; Episodic ataxia, type 9, 618924 (3), Autosomal dominant |
| *SCN2B* | 601327 | Atrial fibrillation, familial, 14, 615378 (3), Autosomal dominant |
| *SCN3A* | 182391 | Epilepsy, familial focal, with variable foci 4, 617935 (3), Autosomal dominant; Developmental and epileptic encephalopathy 62, 617938 (3), Autosomal dominant |
| *SCN3B* | 608214 | Atrial fibrillation, familial, 16, 613120 (3), Autosomal dominant; Brugada syndrome 7, 613120 (3), Autosomal dominant |
| *SCN4A* | 603967 | Paramyotonia congenita, 168300 (3), Autosomal dominant; Hypokalemic periodic paralysis, type 2, 613345 (3), Autosomal dominant; Myotonia congenita, atypical, acetazolamide-responsive, 608390 (3), Autosomal dominant; Myasthenic syndrome, congenital, 16, 614198 (3), Autosomal recessive; Hyperkalemic periodic paralysis, type 2, 170500 (3), Autosomal dominant |
| *SCN4B* | 608256 | Atrial fibrillation, familial, 17, 611819 (3), Autosomal dominant; Long QT syndrome 10, 611819 (3), Autosomal dominant |
| *SCN5A* | 600163 | Ventricular fibrillation, familial, 1, 603829 (3); Heart block, progressive, type IA, 113900 (3), Autosomal dominant; Cardiomyopathy, dilated, 1E, 601154 (3), Autosomal dominant; Heart block, nonprogressive, 113900 (3), Autosomal dominant; Long QT syndrome 3, 603830 (3), Autosomal dominant; Sick sinus syndrome 1, 608567 (3), Autosomal recessive; Brugada syndrome 1, 601144 (3), Autosomal dominant; Atrial fibrillation, familial, 10, 614022 (3), Autosomal dominant; {Sudden infant death syndrome, susceptibility to}, 272120 (3), Autosomal recessive |
| *SCN8A* | 600702 | ?Myoclonus, familial, 2, 618364 (3), Autosomal dominant; Seizures, benign familial infantile, 5, 617080 (3), Autosomal dominant; Cognitive impairment with or without cerebellar ataxia, 614306 (3), Autosomal dominant; Developmental and epileptic encephalopathy 13, 614558 (3), Autosomal dominant |
| *SCN9A* | 603415 | Erythermalgia, primary, 133020 (3), Autosomal dominant; Insensitivity to pain, congenital, 243000 (3), Autosomal recessive; Small fiber neuropathy, 133020 (3), Autosomal dominant; Paroxysmal extreme pain disorder, 167400 (3), Autosomal dominant; Neuropathy, hereditary sensory and autonomic, type IID, 243000 (3), Autosomal recessive |
| *SCNN1A* | 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 |
| *SCNN1B* | 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 |
| *SCNN1G* | 600761 | Bronchiectasis with or without elevated sweat chloride 3, 613071 (3), Autosomal dominant; Pseudohypoaldosteronism, type I, 264350 (3), Autosomal recessive; Liddle syndrome 2, 618114 (3), Autosomal dominant |
| *SCO1* | 603644 | Mitochondrial complex IV deficiency, nuclear type 4, 619048 (3), Autosomal recessive |
| *SCO2* | 604272 | Myopia 6, 608908 (3), Autosomal dominant; Mitochondrial complex IV deficiency, nuclear type 2, 604377 (3), Autosomal recessive |
| *SCP2* | 184755 | ?Leukoencephalopathy with dystonia and motor neuropathy, 613724 (3), Autosomal recessive |
| *SCUBE3* | 614708 | Short stature, facial dysmorphism, and skeletal anomalies with or without cardiac anomalies, 619184 (3), Autosomal recessive |
| *SCYL1* | 607982 | Spinocerebellar ataxia, autosomal recessive 21, 616719 (3), Autosomal recessive |
| *SCYL2* | 616365 | Arthrogryposis multiplex congenita 4, neurogenic, with agenesis of the corpus callosum, 618766 (3), Autosomal recessive |
| *SDC3* | 186357 | {Obesity, association with}, 601665 (3), Multifactorial, Autosomal recessive, Autosomal dominant |
| *SDCCAG8* | 613524 | Senior-Loken syndrome 7, 613615 (3); Bardet-Biedl syndrome 16, 615993 (3), Autosomal recessive |
| *SDHA* | 600857 | Cardiomyopathy, dilated, 1GG, 613642 (3), Autosomal recessive; Mitochondrial complex II deficiency, nuclear type 1, 252011 (3), Autosomal recessive; Neurodegeneration with ataxia and late-onset optic atrophy, 619259 (3), Autosomal dominant; Paragangliomas 5, 614165 (3), Autosomal dominant |
| *SDHAF1* | 612848 | Mitochondrial complex II deficiency, nuclear type 2, 619166 (3), Autosomal recessive |
| *SDHAF2* | 613019 | Paragangliomas 2, 601650 (3), Autosomal dominant |
| *SDHB* | 185470 | Paragangliomas 4, 115310 (3), Autosomal dominant; Mitochondrial complex II deficiency, nuclear type 4, 619224 (3), Autosomal recessive; Gastrointestinal stromal tumor, 606764 (3), Isolated cases, Autosomal dominant; Pheochromocytoma, 171300 (3), Autosomal dominant; Paraganglioma and gastric stromal sarcoma, 606864 (3) |
| *SDHC* | 602413 | Paragangliomas 3, 605373 (3), Autosomal dominant; Paraganglioma and gastric stromal sarcoma, 606864 (3); Gastrointestinal stromal tumor, 606764 (3), Isolated cases, Autosomal dominant |
| *SDHD* | 602690 | Paragangliomas 1, with or without deafness, 168000 (3), Autosomal dominant; Paraganglioma and gastric stromal sarcoma, 606864 (3); Mitochondrial complex II deficiency, nuclear type 3, 619167 (3), Autosomal recessive; Pheochromocytoma, 171300 (3), Autosomal dominant |
| *SDR9C7* | 609769 | Ichthyosis, congenital, autosomal recessive 13, 617574 (3), Autosomal recessive |
| *SEC23A* | 610511 | Craniolenticulosutural dysplasia, 607812 (3), Autosomal recessive |
| *SEC23B* | 610512 | ?Cowden syndrome 7, 616858 (3), Autosomal dominant; Dyserythropoietic anemia, congenital, type II, 224100 (3), Autosomal recessive |
| *SEC24D* | 607186 | Cole-Carpenter syndrome 2, 616294 (3), Autosomal recessive |
| *SEC31A* | 610257 | ?Neurodevelopmental disorder with spastic quadriplegia, optic atrophy, seizures, and structural brain anomalies, 618651 (3), Autosomal recessive |
| *SEC61A1* | 609213 | Tubulointerstitial kidney disease, autosomal dominant, 5, 617056 (3), Autosomal dominant |
| *SEC63* | 608648 | Polycystic liver disease 2, 617004 (3), Autosomal dominant |
| *SECISBP2* | 607693 | Thyroid hormone metabolism, abnormal, 609698 (3), Autosomal recessive |
| *SELENBP1* | 604188 | Extraoral halitosis due to MTO deficiency, 618148 (3), Autosomal recessive |
| *SELENOI* | 607915 | Spastic paraplegia 81, autosomal recessive, 618768 (3), Autosomal recessive |
| *SELENON* | 606210 | Myopathy, congenital, with fiber-type disproportion, 255310 (3), Autosomal recessive, Autosomal dominant; Muscular dystrophy, rigid spine, 1, 602771 (3), Autosomal recessive |
| *SEMA3A* | 603961 | {Hypogonadotropic hypogonadism 16 with or without anosmia}, 614897 (3), Autosomal dominant |
| *SEMA3E* | 608166 | ?CHARGE syndrome, 214800 (3), Autosomal dominant |
| *SEMA4A* | 607292 | Retinitis pigmentosa 35, 610282 (3), Autosomal recessive, Autosomal dominant; Cone-rod dystrophy 10, 610283 (3), Autosomal recessive |
| *SEMA6B* | 608873 | Epilepsy, progressive myoclonic, 11, 618876 (3), Autosomal dominant |
| *SEMA7A* | 607961 | [Blood group, John-Milton-Hagen system], 614745 (3) |
| *SEPSECS* | 613009 | Pontocerebellar hypoplasia type 2D, 613811 (3), Autosomal recessive |
| *SEPTIN12 (SEPT12)* | 611562 | Spermatogenic failure 10, 614822 (3), Autosomal dominant |
| *SEPTIN9 (SEPT9)* | 604061 | Amyotrophy, hereditary neuralgic, 162100 (3), Autosomal dominant |
| *SERAC1* | 614725 | 3-methylglutaconic aciduria with deafness, encephalopathy, and Leigh-like syndrome, 614739 (3), Autosomal recessive |
| *SERPINA1* | 107400 | Hemorrhagic diathesis due to antithrombin Pittsburgh, 613490 (3), Autosomal recessive; Emphysema due to AAT deficiency, 613490 (3), Autosomal recessive; Emphysema-cirrhosis, due to AAT deficiency, 613490 (3), Autosomal recessive |
| *SERPINA3* | 107280 | Alpha-1-antichymotrypsin deficiency (3); Cerebrovascular disease, occlusive (3) |
| *SERPINA6* | 122500 | Corticosteroid-binding globulin deficiency, 611489 (3), Autosomal recessive, Autosomal dominant |
| *SERPINA7* | 314200 | [Thyroxine-binding globulin QTL], 300932 (3), X-linked |
| *SERPINB6* | 173321 | ?Deafness, autosomal recessive 91, 613453 (3), Autosomal recessive |
| *SERPINB7* | 603357 | Palmoplantar keratoderma, Nagashima type, 615598 (3), Autosomal recessive |
| *SERPINB8* | 601697 | Peeling skin syndrome 5, 617115 (3), Autosomal recessive |
| *SERPINC1* | 107300 | Thrombophilia due to antithrombin III deficiency, 613118 (3), Autosomal recessive, Autosomal dominant |
| *SERPIND1* | 142360 | Thrombophilia due to heparin cofactor II deficiency, 612356 (3), Autosomal dominant |
| *SERPINE1* | 173360 | Plasminogen activator inhibitor-1 deficiency, 613329 (3), Autosomal recessive, Autosomal dominant; {Transcription of plasminogen activator inhibitor, modulator of} (3) |
| *SERPINF1* | 172860 | Osteogenesis imperfecta, type VI, 613982 (3), Autosomal recessive |
| *SERPINF2* | 613168 | Alpha-2-plasmin inhibitor deficiency, 262850 (3), Autosomal recessive |
| *SERPING1* | 606860 | Angioedema, hereditary, 1 and 2, 106100 (3), Autosomal recessive, Autosomal dominant; Complement component 4, partial deficiency of, 120790 (3), Autosomal dominant |
| *SERPINH1* | 600943 | {Preterm premature rupture of the membranes, susceptibility to}, 610504 (3); Osteogenesis imperfecta, type X, 613848 (3), Autosomal recessive |
| *SERPINI1* | 602445 | Encephalopathy, familial, with neuroserpin inclusion bodies, 604218 (3), Autosomal dominant |
| *SET* | 600960 | Mental retardation, autosomal dominant 58, 618106 (3), Autosomal dominant |
| *SETBP1* | 611060 | Schinzel-Giedion midface retraction syndrome, 269150 (3), Autosomal dominant; Mental retardation, autosomal dominant 29, 616078 (3), Autosomal dominant |
| *SETD1A* | 611052 | Epilepsy, early-onset, with or without developmental delay, 618832 (3), Autosomal dominant; Neurodevelopmental disorder with speech impairment and dysmorphic facies, 619056 (3), Autosomal dominant |
| *SETD1B* | 611055 | Intellectual developmental disorder with seizures and language delay, 619000 (3), Autosomal dominant |
| *SETD2* | 612778 | Luscan-Lumish syndrome, 616831 (3), Autosomal dominant |
| *SETD5* | 615743 | Mental retardation, autosomal dominant 23, 615761 (3), Autosomal dominant |
| *SETX* | 608465 | Spinocerebellar ataxia, autosomal recessive, with axonal neuropathy 2, 606002 (3), Autosomal recessive; Amyotrophic lateral sclerosis 4, juvenile, 602433 (3), Autosomal dominant |
| *SF3B1* | 605590 | Myelodysplastic syndrome, somatic, 614286 (3) |
| *SF3B4* | 605593 | Acrofacial dysostosis 1, Nager type, 154400 (3), Autosomal dominant |
| *SFRP4* | 606570 | Pyle disease, 265900 (3), Autosomal recessive |
| *SFTPA2* | 178642 | Pulmonary fibrosis, idiopathic, 178500 (3), Autosomal dominant |
| *SFTPB* | 178640 | Surfactant metabolism dysfunction, pulmonary, 1, 265120 (3), Autosomal recessive |
| *SFTPC* | 178620 | Surfactant metabolism dysfunction, pulmonary, 2, 610913 (3), Autosomal dominant |
| *SFXN4* | 615564 | Combined oxidative phosphorylation deficiency 18, 615578 (3), Autosomal recessive |
| *SGCA* | 600119 | Muscular dystrophy, limb-girdle, autosomal recessive 3, 608099 (3), Autosomal recessive |
| *SGCB* | 600900 | Muscular dystrophy, limb-girdle, autosomal recessive 4, 604286 (3), Autosomal recessive |
| *SGCD* | 601411 | Cardiomyopathy, dilated, 1L, 606685 (3); Muscular dystrophy, limb-girdle, autosomal recessive 6, 601287 (3), Autosomal recessive |
| *SGCE* | 604149 | Dystonia-11, myoclonic, 159900 (3), Autosomal dominant |
| *SGCG* | 608896 | Muscular dystrophy, limb-girdle, autosomal recessive 5, 253700 (3), Autosomal recessive |
| *SGMS2* | 611574 | Calvarial doughnut lesions with bone fragility with or without spondylometaphyseal dysplasia, 126550 (3), Autosomal dominant |
| *SGO1* | 609168 | Chronic atrial and intestinal dysrhythmia, 616201 (3), Autosomal recessive |
| *SGPL1* | 603729 | Nephrotic syndrome, type 14, 617575 (3), Autosomal recessive |
| *SGSH* | 605270 | Mucopolysaccharidosis type IIIA (Sanfilippo A), 252900 (3), Autosomal recessive |
| *SH2B3* | 605093 | Thrombocythemia, somatic, 187950 (3); Myelofibrosis, somatic, 254450 (3); Erythrocytosis, somatic, 133100 (3) |
| *SH2D1A* | 300490 | Lymphoproliferative syndrome, X-linked, 1, 308240 (3), X-linked recessive |
| *SH3BP2* | 602104 | Cherubism, 118400 (3), Autosomal dominant |
| *SH3GL1* | 601768 | Leukemia, acute myeloid, 601626 (1), Somatic mutation, Autosomal dominant |
| *SH3KBP1* | 300374 | ?Immunodeficiency 61, 300310 (3), X-linked recessive |
| *SH3PXD2B* | 613293 | Frank-ter Haar syndrome, 249420 (3), Autosomal recessive |
| *SH3TC2* | 608206 | Charcot-Marie-Tooth disease, type 4C, 601596 (3), Autosomal recessive; Mononeuropathy of the median nerve, mild, 613353 (3), Autosomal dominant |
| *SHANK1* | 604999 | Spectrum of NDD. PMID: 34113010 |
| *SHANK2* | 603290 | {Autism susceptibility 17}, 613436 (3) |
| *SHANK3* | 606230 | Phelan-McDermid syndrome, 606232 (3), Autosomal dominant; {Schizophrenia 15}, 613950 (3), Autosomal dominant |
| *SHH* | 600725 | Microphthalmia with coloboma 5, 611638 (3), Autosomal dominant; Schizencephaly, 269160 (3); Single median maxillary central incisor, 147250 (3), Autosomal dominant; Holoprosencephaly 3, 142945 (3), Autosomal dominant |
| *SHMT2* | 138450 | Neurodevelopmental disorder with cardiomyopathy, spasticity, and brain abnormalities, 619121 (3), Autosomal recessive |
| *SHOC2* | 602775 | Noonan syndrome-like with loose anagen hair 1, 607721 (3), Autosomal dominant |
| *SHOX* | 400020 | Short stature, idiopathic familial, 300582 (3); Langer mesomelic dysplasia, 249700 (3), Pseudoautosomal recessive; Leri-Weill dyschondrosteosis, 127300 (3), Pseudoautosomal dominant |
| *SHPK* | 605060 | [Sedoheptulokinase deficiency], 617213 (3), Autosomal recessive |
| *SHROOM4* | 300579 | Stocco dos Santos X-linked mental retardation syndrome, 300434 (3), X-linked |
| *SI* | 609845 | Sucrase-isomaltase deficiency, congenital, 222900 (3), Autosomal recessive |
| *SIAE* | 610079 | {Autoimmune disease, susceptibility to, 6}, 613551 (3) |
| *SIAH1* | 602212 | Buratti-Harel syndrome, 619314 (3), Autosomal dominant |
| *SIGMAR1* | 601978 | ?Spinal muscular atrophy, distal, autosomal recessive, 2, 605726 (3), Autosomal recessive; ?Amyotrophic lateral sclerosis 16, juvenile, 614373 (3), Autosomal recessive |
| *SIK1* | 605705 | Developmental and epileptic encephalopathy 30, 616341 (3), Autosomal dominant |
| *SIK3* | 614776 | ?Spondyloepimetaphyseal dysplasia, Krakow type, 618162 (3), Autosomal recessive |
| *SIL1* | 608005 | Marinesco-Sjogren syndrome, 248800 (3), Autosomal recessive |
| *SIM1* | 603128 | Early onset obesity and hypopituitarism. PMID: 33434169 |
| *SIN3A* | 607776 | Witteveen-Kolk syndrome, 613406 (3), Autosomal dominant |
| *SIPA1L3* | 616655 | ?Cataract 45, 616851 (3), Autosomal recessive |
| *SIX1* | 601205 | Deafness, autosomal dominant 23, 605192 (3), Autosomal dominant; Branchiootic syndrome 3, 608389 (3), Autosomal dominant |
| *SIX3* | 603714 | Schizencephaly, 269160 (3); Holoprosencephaly 2, 157170 (3), Autosomal dominant |
| *SIX5* | 600963 | Branchiootorenal syndrome 2, 610896 (3) |
| *SIX6* | 606326 | Optic disc anomalies with retinal and/or macular dystrophy, 212550 (3), Autosomal recessive |
| *SKI* | 164780 | Shprintzen-Goldberg syndrome, 182212 (3), Autosomal dominant |
| *SKIV2L* | 600478 | Trichohepatoenteric syndrome 2, 614602 (3), Autosomal recessive |
| *SLC10A1* | 182396 | Hypercholanemia, familial 2, 619256 (3), Autosomal recessive |
| *SLC10A2* | 601295 | Bile acid malabsorption, primary, 613291 (3), Autosomal recessive |
| *SLC10A7* | 611459 | Short stature, amelogenesis imperfecta, and skeletal dysplasia with scoliosis, 618363 (3), Autosomal recessive |
| *SLC11A1* | 600266 | {Mycobacterium tuberculosis, susceptibility to infection by}, 607948 (3); {Buruli ulcer, susceptibility to}, 610446 (3) |
| *SLC11A2* | 600523 | Anemia, hypochromic microcytic, with iron overload 1, 206100 (3), Autosomal recessive |
| *SLC12A1* | 600839 | Bartter syndrome, type 1, 601678 (3), Autosomal recessive |
| *SLC12A2* | 600840 | Kilquist syndrome, 619080 (3), Autosomal recessive; Delpire-McNeill syndrome, 619083 (3), Autosomal dominant; Deafness, autosomal dominant 78, 619081 (3), Autosomal dominant |
| *SLC12A3* | 600968 | Gitelman syndrome, 263800 (3), Autosomal recessive |
| *SLC12A5* | 606726 | {Epilepsy, idiopathic generalized, susceptibility to, 14}, 616685 (3), Autosomal dominant; Developmental and epileptic encephalopathy 34, 616645 (3), Autosomal recessive |
| *SLC12A6* | 604878 | Agenesis of the corpus callosum with peripheral neuropathy, 218000 (3), Autosomal recessive |
| *SLC13A3* | 606411 | Leukoencephalopathy, acute reversible, with increased urinary alpha-ketoglutarate, 618384 (3), Autosomal recessive |
| *SLC13A5* | 608305 | Developmental and epileptic encephalopathy 25, with amelogenesis imperfecta, 615905 (3), Autosomal recessive |
| *SLC14A1* | 613868 | [Blood group, Kidd], 111000 (3) |
| *SLC16A1* | 600682 | Hyperinsulinemic hypoglycemia, familial, 7, 610021 (3), Autosomal dominant; Erythrocyte lactate transporter defect, 245340 (3), Autosomal dominant; Monocarboxylate transporter 1 deficiency, 616095 (3), Autosomal recessive, Autosomal dominant |
| *SLC16A12* | 611910 | Cataract 47, juvenile, with microcornea, 612018 (3), Autosomal dominant |
| *SLC16A2* | 300095 | Allan-Herndon-Dudley syndrome, 300523 (3), X-linked |
| *SLC17A3* | 611034 | [Uric acid concentration, serum, QTL4], 612671 (3), Autosomal dominant; {Gout susceptibility 4}, 612671 (3), Autosomal dominant |
| *SLC17A5* | 604322 | Salla disease, 604369 (3), Autosomal recessive; Sialic acid storage disorder, infantile, 269920 (3), Autosomal recessive |
| *SLC17A8* | 607557 | Deafness, autosomal dominant 25, 605583 (3), Autosomal dominant |
| *SLC17A9* | 612107 | Porokeratosis 8, disseminated superficial actinic type, 616063 (3), Autosomal dominant |
| *SLC18A2* | 193001 | ?Parkinsonism-dystonia, infantile, 2, 618049 (3), Autosomal recessive |
| *SLC18A3* | 600336 | Myasthenic syndrome, congenital, 21, presynaptic, 617239 (3), Autosomal recessive |
| *SLC19A1* | 600424 | ?Megaloblastic anemia, folate-responsive, 601775 (3), Autosomal recessive |
| *SLC19A2* | 603941 | Thiamine-responsive megaloblastic anemia syndrome, 249270 (3), Autosomal recessive |
| *SLC19A3* | 606152 | Thiamine metabolism dysfunction syndrome 2 (biotin- or thiamine-responsive encephalopathy type 2), 607483 (3), Autosomal recessive |
| *SLC1A1* | 133550 | Dicarboxylic aminoaciduria, 222730 (3), Autosomal recessive; {?Schizophrenia susceptibility 18}, 615232 (3) |
| *SLC1A2* | 600300 | Developmental and epileptic encephalopathy 41, 617105 (3), Autosomal dominant |
| *SLC1A3* | 600111 | Episodic ataxia, type 6, 612656 (3), Autosomal dominant |
| *SLC1A4* | 600229 | Spastic tetraplegia, thin corpus callosum, and progressive microcephaly, 616657 (3), Autosomal recessive |
| *SLC20A2* | 158378 | Basal ganglia calcification, idiopathic, 1, 213600 (3), Autosomal dominant |
| *SLC22A12* | 607096 | Hypouricemia, renal, 220150 (3), Autosomal recessive |
| *SLC22A18* | 602631 | Breast cancer, somatic, 114480 (3); Lung cancer, somatic, 211980 (3); Rhabdomyosarcoma, somatic, 268210 (3) |
| *SLC22A4* | 604190 | {Rheumatoid arthritis, susceptibility to}, 180300 (3) |
| *SLC22A5* | 603377 | Carnitine deficiency, systemic primary, 212140 (3), Autosomal recessive |
| *SLC24A1* | 603617 | Night blindness, congenital stationary (complete), 1D, autosomal recessive, 613830 (3), Autosomal recessive |
| *SLC24A4* | 609840 | [Skin/hair/eye pigmentation 6, blond/brown hair], 210750 (3), Autosomal recessive; Amelogenesis imperfecta, type IIA5, 615887 (3), Autosomal recessive; [Skin/hair/eye pigmentation 6, blue/green eyes], 210750 (3), Autosomal recessive |
| *SLC24A5* | 609802 | [Skin/hair/eye pigmentation 4, fair/dark skin], 113750 (3), Autosomal recessive; Albinism, oculocutaneous, type VI, 113750 (3), Autosomal recessive |
| *SLC25A1* | 190315 | Combined D-2- and L-2-hydroxyglutaric aciduria, 615182 (3), Autosomal recessive; Myasthenic syndrome, congenital, 23, presynaptic, 618197 (3), Autosomal recessive |
| *SLC25A10* | 606794 | ?Mitochondrial DNA depletion syndrome 19, 618972 (3), Autosomal recessive |
| *SLC25A11* | 604165 | Paragangliomas 6, 618464 (3), Autosomal dominant |
| *SLC25A12* | 603667 | Developmental and epileptic encephalopathy 39, 612949 (3), Autosomal recessive |
| *SLC25A13* | 603859 | Citrullinemia, type II, neonatal-onset, 605814 (3), Autosomal recessive; Citrullinemia, adult-onset type II, 603471 (3), Autosomal recessive |
| *SLC25A15* | 603861 | Hyperornithinemia-hyperammonemia-homocitrullinemia syndrome, 238970 (3), Autosomal recessive |
| *SLC25A19* | 606521 | Microcephaly, Amish type, 607196 (3), Autosomal recessive; Thiamine metabolism dysfunction syndrome 4 (progressive polyneuropathy type), 613710 (3), Autosomal recessive |
| *SLC25A20* | 613698 | Carnitine-acylcarnitine translocase deficiency, 212138 (3), Autosomal recessive |
| *SLC25A21* | 607571 | ?Mitochondrial DNA depletion syndrome 18, 618811 (3), Autosomal recessive |
| *SLC25A22* | 609302 | Developmental and epileptic encephalopathy 3, 609304 (3), Autosomal recessive |
| *SLC25A24* | 608744 | Fontaine progeroid syndrome, 612289 (3), Autosomal dominant |
| *SLC25A26* | 611037 | Combined oxidative phosphorylation deficiency 28, 616794 (3), Autosomal recessive |
| *SLC25A3* | 600370 | Mitochondrial phosphate carrier deficiency, 610773 (3), Autosomal recessive |
| *SLC25A32* | 138480 | ?Exercise intolerance, riboflavin-responsive, 616839 (3), Autosomal recessive |
| *SLC25A38* | 610819 | Anemia, sideroblastic, 2, pyridoxine-refractory, 205950 (3), Autosomal recessive |
| *SLC25A4* | 103220 | Mitochondrial DNA depletion syndrome 12B (cardiomyopathic type) AR, 615418 (3), Autosomal recessive; Progressive external ophthalmoplegia with mitochondrial DNA deletions, autosomal dominant 2, 609283 (3), Autosomal dominant; Mitochondrial DNA depletion syndrome 12A (cardiomyopathic type) AD, 617184 (3), Autosomal dominant |
| *SLC25A42* | 610823 | Metabolic crises, recurrent, with variable encephalomyopathic features and neurologic regression, 618416 (3), Autosomal recessive |
| *SLC25A46* | 610826 | Neuropathy, hereditary motor and sensory, type VIB, 616505 (3), Autosomal recessive; Pontocerebellar hypoplasia, type 1E, 619303 (3), Autosomal recessive |
| *SLC26A1* | 610130 | ?Nephrolithiasis, calcium oxalate, 167030 (3), Autosomal recessive |
| *SLC26A2* | 606718 | Epiphyseal dysplasia, multiple, 4, 226900 (3), Autosomal recessive; De la Chapelle dysplasia, 256050 (3), Autosomal recessive; Diastrophic dysplasia, 222600 (3), Autosomal recessive; Diastrophic dysplasia, broad bone-platyspondylic variant, 222600 (3), Autosomal recessive; Achondrogenesis Ib, 600972 (3), Autosomal recessive; Atelosteogenesis, type II, 256050 (3), Autosomal recessive |
| *SLC26A3* | 126650 | Diarrhea 1, secretory chloride, congenital, 214700 (3), Autosomal recessive |
| *SLC26A4* | 605646 | Deafness, autosomal recessive 4, with enlarged vestibular aqueduct, 600791 (3), Autosomal recessive; Pendred syndrome, 274600 (3), Autosomal recessive |
| *SLC26A5* | 604943 | ?Deafness, autosomal recessive 61, 613865 (3), Autosomal recessive |
| *SLC26A8* | 608480 | Spermatogenic failure 3, 606766 (3), Autosomal dominant |
| *SLC27A4* | 604194 | Ichthyosis prematurity syndrome, 608649 (3), Autosomal recessive |
| *SLC28A1* | 606207 | [Uridine-cytidineuria], 618477 (3), Autosomal recessive |
| *SLC29A3* | 612373 | Histiocytosis-lymphadenopathy plus syndrome, 602782 (3), Autosomal recessive |
| *SLC2A1* | 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; {Epilepsy, idiopathic generalized, susceptibility to, 12}, 614847 (3), Autosomal dominant; GLUT1 deficiency syndrome 2, childhood onset, 612126 (3), Autosomal dominant |
| *SLC2A10* | 606145 | Arterial tortuosity syndrome, 208050 (3), Autosomal recessive |
| *SLC2A2* | 138160 | Fanconi-Bickel syndrome, 227810 (3), Autosomal recessive; {Diabetes mellitus, noninsulin-dependent}, 125853 (3), Autosomal dominant |
| *SLC2A9* | 606142 | {Uric acid concentration, serum, QTL 2}, 612076 (3), Autosomal recessive, Autosomal dominant; Hypouricemia, renal, 2, 612076 (3), Autosomal recessive, Autosomal dominant |
| *SLC30A10* | 611146 | Hypermanganesemia with dystonia 1, 613280 (3), Autosomal recessive |
| *SLC30A2* | 609617 | Zinc deficiency, transient neonatal, 608118 (3), Autosomal dominant |
| *SLC30A8* | 611145 | {Diabetes mellitus, noninsulin-dependent, susceptibility to}, 125853 (3), Autosomal dominant |
| *SLC30A9* | 604604 | ?Birk-Landau-Perez syndrome, 617595 (3), Autosomal recessive |
| *SLC33A1* | 603690 | Spastic paraplegia 42, autosomal dominant, 612539 (3), Autosomal dominant; Congenital cataracts, hearing loss, and neurodegeneration, 614482 (3), Autosomal recessive |
| *SLC34A1* | 182309 | ?Fanconi renotubular syndrome 2, 613388 (3), Autosomal recessive; Hypercalcemia, infantile, 2, 616963 (3), Autosomal recessive; Nephrolithiasis/osteoporosis, hypophosphatemic, 1, 612286 (3), Autosomal dominant |
| *SLC34A2* | 604217 | Pulmonary alveolar microlithiasis, 265100 (3), Autosomal recessive |
| *SLC34A3* | 609826 | Hypophosphatemic rickets with hypercalciuria, 241530 (3), Autosomal recessive |
| *SLC35A1* | 605634 | Congenital disorder of glycosylation, type IIf, 603585 (3), Autosomal recessive |
| *SLC35A2* | 314375 | Congenital disorder of glycosylation, type IIm, 300896 (3), X-linked dominant, Somatic mosaicism |
| *SLC35A3* | 605632 | ?Arthrogryposis, mental retardation, and seizures, 615553 (3), Autosomal recessive |
| *SLC35C1* | 605881 | Congenital disorder of glycosylation, type IIc, 266265 (3), Autosomal recessive |
| *SLC35D1* | 610804 | Schneckenbecken dysplasia, 269250 (3), Autosomal recessive |
| *SLC36A2* | 608331 | Iminoglycinuria, digenic, 242600 (3), Autosomal recessive, Digenic recessive; Hyperglycinuria, 138500 (3), Autosomal dominant |
| *SLC37A4* | 602671 | Glycogen storage disease Ib, 232220 (3), Autosomal recessive; Glycogen storage disease Ic, 232240 (3), Autosomal recessive |
| *SLC38A8* | 615585 | Foveal hypoplasia 2, with or without optic nerve misrouting and/or anterior segment dysgenesis, 609218 (3), Autosomal recessive |
| *SLC39A13* | 608735 | Ehlers-Danlos syndrome, spondylodysplastic type, 3, 612350 (3), Autosomal recessive |
| *SLC39A14* | 608736 | ?Hyperostosis cranalis interna, 144755 (3), Autosomal dominant; Hypermanganesemia with dystonia 2, 617013 (3), Autosomal recessive |
| *SLC39A4* | 607059 | Acrodermatitis enteropathica, 201100 (3), Autosomal recessive |
| *SLC39A5* | 608730 | Myopia 24, autosomal dominant, 615946 (3), Autosomal dominant |
| *SLC39A8* | 608732 | Congenital disorder of glycosylation, type IIn, 616721 (3), Autosomal recessive |
| *SLC3A1* | 104614 | Cystinuria, 220100 (3), Autosomal recessive, Autosomal dominant |
| *SLC40A1* | 604653 | Hemochromatosis, type 4, 606069 (3), Autosomal dominant |
| *SLC44A1* | 606105 | Neurodegeneration, childhood-onset, with ataxia, tremor, optic atrophy, and cognitive decline, 618868 (3), Autosomal recessive |
| *SLC44A4* | 606107 | ?Deafness, autosomal dominant 72, 617606 (3), Autosomal dominant |
| *SLC45A1* | 605763 | Intellectual developmental disorder with neuropsychiatric features, 617532 (3), Autosomal recessive |
| *SLC45A2* | 606202 | [Skin/hair/eye pigmentation 5, dark/light eyes], 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/fair skin], 227240 (3), Autosomal recessive |
| *SLC46A1* | 611672 | Folate malabsorption, hereditary, 229050 (3), Autosomal recessive |
| *SLC4A1* | 109270 | [Blood group, Swann], 601550 (3); [Blood group, Wright], 112050 (3); [Malaria, resistance to], 611162 (3); Distal renal tubular acidosis 1, 179800 (3), Autosomal dominant; [Blood group, Waldner], 112010 (3); Spherocytosis, type 4, 612653 (3), Autosomal dominant; [Blood group, Froese], 601551 (3); Distal renal tubular acidosis 4 with hemolytic anemia, 611590 (3), Autosomal recessive; Cryohydrocytosis, 185020 (3), Autosomal dominant; Ovalocytosis, SA type, 166900 (3), Autosomal dominant; [Blood group, Diego], 110500 (3) |
| *SLC4A11* | 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 |
| *SLC4A4* | 603345 | Renal tubular acidosis, proximal, with ocular abnormalities, 604278 (3), Autosomal recessive |
| *SLC52A1* | 607883 | Riboflavin deficiency, 615026 (3), Autosomal dominant |
| *SLC52A2* | 607882 | Brown-Vialetto-Van Laere syndrome 2, 614707 (3), Autosomal recessive |
| *SLC52A3* | 613350 | ?Fazio-Londe disease, 211500 (3), Autosomal recessive; Brown-Vialetto-Van Laere syndrome 1, 211530 (3), Autosomal recessive |
| *SLC5A1* | 182380 | Glucose/galactose malabsorption, 606824 (3), Autosomal recessive |
| *SLC5A2* | 182381 | Renal glucosuria, 233100 (3), Autosomal recessive, Autosomal dominant |
| *SLC5A5* | 601843 | Thyroid dyshormonogenesis 1, 274400 (3), Autosomal recessive |
| *SLC5A6* | 604024 | Neurodegeneration, infantile-onset, biotin-responsive, 618973 (3), Autosomal recessive |
| *SLC5A7* | 608761 | Neuronopathy, distal hereditary motor, type VIIA, 158580 (3), Autosomal dominant; Myasthenic syndrome, congenital, 20, presynaptic, 617143 (3), Autosomal recessive |
| *SLC6A1* | 137165 | Myoclonic-atonic epilepsy, 616421 (3), Autosomal dominant |
| *SLC6A17* | 610299 | Mental retardation, autosomal recessive 48, 616269 (3), Autosomal recessive |
| *SLC6A19* | 608893 | Iminoglycinuria, digenic, 242600 (3), Autosomal recessive, Digenic recessive; Hartnup disorder, 234500 (3), Autosomal recessive; Hyperglycinuria, 138500 (3), Autosomal dominant |
| *SLC6A2* | 163970 | ?Orthostatic intolerance, 604715 (3) |
| *SLC6A20* | 605616 | Iminoglycinuria, digenic, 242600 (3), Autosomal recessive, Digenic recessive; Hyperglycinuria, 138500 (3), Autosomal dominant |
| *SLC6A3* | 126455 | Parkinsonism-dystonia, infantile, 1, 613135 (3), Autosomal recessive; {Nicotine dependence, protection against}, 188890 (3) |
| *SLC6A4* | 182138 | {Obsessive-compulsive disorder}, 164230 (3), Autosomal dominant; {Anxiety-related personality traits}, 607834 (3) |
| *SLC6A5* | 604159 | Hyperekplexia 3, 614618 (3), Autosomal recessive, Autosomal dominant |
| *SLC6A6* | 186854 | Hypotaurinemic retinal degeneration and cardiomyopathy, 145350 (3), Autosomal recessive |
| *SLC6A8* | 300036 | Cerebral creatine deficiency syndrome 1, 300352 (3), X-linked recessive |
| *SLC6A9* | 601019 | Glycine encephalopathy with normal serum glycine, 617301 (3), Autosomal recessive |
| *SLC7A14* | 615720 | Retinitis pigmentosa 68, 615725 (3), Autosomal recessive |
| *SLC7A6OS* | 619192 | Epilepsy, progressive myoclonic, 12, 619191 (3), Autosomal recessive |
| *SLC7A7* | 603593 | Lysinuric protein intolerance, 222700 (3), Autosomal recessive |
| *SLC7A9* | 604144 | Cystinuria, 220100 (3), Autosomal recessive, Autosomal dominant |
| *SLC9A1* | 107310 | Lichtenstein-Knorr syndrome, 616291 (3), Autosomal recessive |
| *SLC9A3* | 182307 | Diarrhea 8, secretory sodium, congenital, 616868 (3), Autosomal recessive |
| *SLC9A3R1* | 604990 | Nephrolithiasis/osteoporosis, hypophosphatemic, 2, 612287 (3), Autosomal dominant |
| *SLC9A6* | 300231 | Mental retardation, X-linked syndromic, Christianson type, 300243 (3), X-linked |
| *SLC9A7* | 300368 | Intellectual developmental disorder, X-linked 108, 301024 (3), X-linked recessive |
| *SLC9A9* | 608396 | {?Autism susceptibility 16}, 613410 (3) |
| *SLCO1B1* | 604843 | Hyperbilirubinemia, Rotor type, digenic, 237450 (3), Digenic recessive |
| *SLCO1B3* | 605495 | Hyperbilirubinemia, Rotor type, digenic, 237450 (3), Digenic recessive |
| *SLCO2A1* | 601460 | Hypertrophic osteoarthropathy, primary, autosomal dominant, 167100 (3), Autosomal dominant; Hypertrophic osteoarthropathy, primary, autosomal recessive 2, 614441 (3), Autosomal recessive |
| *SLFN14* | 614958 | Bleeding disorder, platelet-type, 20, 616913 (3), Autosomal dominant |
| *SLITRK1* | 609678 | Tourette syndrome, 137580 (3), Autosomal dominant; ?Trichotillomania, 613229 (3), Multifactorial, Autosomal dominant |
| *SLITRK6* | 609681 | Deafness and myopia, 221200 (3), Autosomal recessive |
| *SLURP1* | 606119 | Meleda disease, 248300 (3), Autosomal recessive |
| *SLX4* | 613278 | Fanconi anemia, complementation group P, 613951 (3), Autosomal recessive |
| *SMAD3* | 603109 | Loeys-Dietz syndrome 3, 613795 (3), Autosomal dominant |
| *SMAD4* | 600993 | Pancreatic cancer, somatic, 260350 (3); Myhre syndrome, 139210 (3), Autosomal dominant; Polyposis, juvenile intestinal, 174900 (3), Autosomal dominant; Juvenile polyposis/hereditary hemorrhagic telangiectasia syndrome, 175050 (3), Autosomal dominant |
| *SMAD6* | 602931 | Aortic valve disease 2, 614823 (3), Autosomal dominant; {Radioulnar synostosis, nonsyndromic}, 179300 (3), Autosomal dominant; {Craniosynostosis 7, susceptibility to}, 617439 (3), Autosomal dominant |
| *SMAD7* | 602932 | {Colorectal cancer, susceptibility to, 3}, 612229 (3) |
| *SMAD9* | 603295 | Pulmonary hypertension, primary, 2, 615342 (3), Autosomal dominant |
| *SMARCA2* | 600014 | Nicolaides-Baraitser syndrome, 601358 (3), Autosomal dominant; Blepharophimosis-impaired intellectual development syndrome, 619293 (3), Autosomal dominant |
| *SMARCA4* | 603254 | Coffin-Siris syndrome 4, 614609 (3), Autosomal dominant; {Rhabdoid tumor predisposition syndrome 2}, 613325 (3), Autosomal dominant |
| *SMARCAD1* | 612761 | Basan syndrome, 129200 (3), Autosomal dominant; Huriez syndrome, 181600 (3), Autosomal dominant; Adermatoglyphia, 136000 (3), Autosomal dominant |
| *SMARCAL1* | 606622 | Schimke immunoosseous dysplasia, 242900 (3), Autosomal recessive |
| *SMARCB1* | 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 |
| *SMARCC2* | 601734 | Coffin-Siris syndrome 8, 618362 (3), Autosomal dominant |
| *SMARCD1* | 601735 | Coffin-Siris syndrome 11, 618779 (3), Autosomal dominant |
| *SMARCD2* | 601736 | Specific granule deficiency 2, 617475 (3), Autosomal recessive |
| *SMARCE1* | 603111 | {Meningioma, familial, susceptibility to}, 607174 (3), Autosomal dominant; Coffin-Siris syndrome 5, 616938 (3), Autosomal dominant |
| *SMC1A* | 300040 | Cornelia de Lange syndrome 2, 300590 (3), X-linked dominant; Developmental and epileptic encephalopathy 85, with or without midline brain defects, 301044 (3), X-linked dominant |
| *SMC3* | 606062 | Cornelia de Lange syndrome 3, 610759 (3), Autosomal dominant |
| *SMCHD1* | 614982 | Bosma arhinia microphthalmia syndrome, 603457 (3), Autosomal dominant; Fascioscapulohumeral muscular dystrophy 2, digenic, 158901 (3), Digenic dominant |
| *SMG8* | 613175 | Alzahrani-Kuwahara syndrome, 619268 (3), Autosomal recessive |
| *SMG9* | 613176 | Heart and brain malformation syndrome, 616920 (3), Autosomal recessive |
| *SMIM1* | 615242 | [Blood group, Vel system], 615264 (3), Autosomal recessive |
| *SMN1* | 600354 | Spinal muscular atrophy-2, 253550 (3), Autosomal recessive; Spinal muscular atrophy-4, 271150 (3), Autosomal recessive; Spinal muscular atrophy-3, 253400 (3), Autosomal recessive; Spinal muscular atrophy-1, 253300 (3), Autosomal recessive |
| *SMN2* | 601627 | {Spinal muscular atrophy, type III, modifier of}, 253400 (3), Autosomal recessive |
| *SMO* | 601500 | Pallister-Hall-like syndrome, 241800 (3), Autosomal recessive; Basal cell carcinoma, somatic, 605462 (3); Curry-Jones syndrome, somatic mosaic, 601707 (3) |
| *SMOC1* | 608488 | Microphthalmia with limb anomalies, 206920 (3), Autosomal recessive |
| *SMOC2* | 607223 | Dentin dysplasia, type I, with microdontia and misshapen teeth, 125400 (3), Autosomal recessive |
| *SMPD1* | 607608 | Niemann-Pick disease, type B, 607616 (3), Autosomal recessive; Niemann-Pick disease, type A, 257200 (3), Autosomal recessive |
| *SMPD4* | 610457 | Neurodevelopmental disorder with microcephaly, arthrogryposis, and structural brain anomalies, 618622 (3), Autosomal recessive |
| *SMPX* | 300226 | Deafness, X-linked 4, 300066 (3), X-linked dominant |
| *SMS* | 300105 | Mental retardation, X-linked, Snyder-Robinson type, 309583 (3), X-linked recessive |
| *SNAI2* | 602150 | Waardenburg syndrome, type 2D, 608890 (3), Autosomal recessive; Piebaldism, 172800 (3), Autosomal dominant |
| *SNAP25* | 600322 | ?Myasthenic syndrome, congenital, 18, 616330 (3), Autosomal dominant |
| *SNAP29* | 604202 | Cerebral dysgenesis, neuropathy, ichthyosis, and palmoplantar keratoderma syndrome, 609528 (3), Autosomal recessive |
| *SNCA* | 163890 | Dementia, Lewy body, 127750 (3), Autosomal dominant; Parkinson disease 1, 168601 (3), Autosomal dominant; Parkinson disease 4, 605543 (3), Autosomal dominant |
| *SNCB* | 602569 | Dementia, Lewy body, 127750 (3), Autosomal dominant |
| *SNIP1* | 608241 | Psychomotor retardation, epilepsy, and craniofacial dysmorphism, 614501 (3), Autosomal recessive |
| *SNORD118* | 616663 | Leukoencephalopathy, brain calcifications, and cysts, 614561 (3), Autosomal recessive |
| *SNRNP200* | 601664 | Retinitis pigmentosa 33, 610359 (3), Autosomal dominant |
| *SNRPB* | 182282 | Cerebrocostomandibular syndrome, 117650 (3), Autosomal dominant |
| *SNRPE* | 128260 | Hypotrichosis 11, 615059 (3), Autosomal dominant |
| *SNRPN* | 182279 | Prader-Willi syndrome, 176270 (3), Autosomal dominant |
| *SNTA1* | 601017 | Long QT syndrome 12, 612955 (3), Autosomal dominant |
| *SNX10* | 614780 | Osteopetrosis, autosomal recessive 8, 615085 (3), Autosomal recessive |
| *SNX14* | 616105 | Spinocerebellar ataxia, autosomal recessive 20, 616354 (3), Autosomal recessive |
| *SOBP* | 613667 | Mental retardation, anterior maxillary protrusion, and strabismus, 613671 (3), Autosomal recessive |
| *SOCS1* | 603597 | Autoinflammatory syndrome, familial, with or without immunodeficiency, 619375 (3), Autosomal dominant |
| *SOD1* | 147450 | Spastic tetraplegia and axial hypotonia, progressive, 618598 (3), Autosomal recessive; Amyotrophic lateral sclerosis 1, 105400 (3), Autosomal recessive, Autosomal dominant |
| *SOD2* | 147460 | {Microvascular complications of diabetes 6}, 612634 (3) |
| *SOD3* | 185490 | [Superoxide dismutase, elevated extracellular] (3) |
| *SOHLH1* | 610224 | Ovarian dysgenesis 5, 617690 (3), Autosomal recessive; Spermatogenic failure 32, 618115 (3), Autosomal dominant |
| *SON* | 182465 | ZTTK syndrome, 617140 (3), Autosomal dominant |
| *SORD* | 182500 | Sorbitol dehydrogenase deficiency with peripheral neuropathy, 618912 (3), Autosomal recessive |
| *SORT1* | 602458 | [Low density lipoprotein cholesterol level QTL6], 613589 (3), Autosomal dominant |
| *SOS1* | 182530 | Noonan syndrome 4, 610733 (3), Autosomal dominant; ?Fibromatosis, gingival, 1, 135300 (3), Autosomal dominant |
| *SOS2* | 601247 | Noonan syndrome 9, 616559 (3), Autosomal dominant |
| *SOST* | 605740 | Sclerosteosis 1, 269500 (3), Autosomal recessive; Craniodiaphyseal dysplasia, autosomal dominant, 122860 (3), Autosomal dominant |
| *SOX10* | 602229 | Waardenburg syndrome, type 4C, 613266 (3), Autosomal dominant; PCWH syndrome, 609136 (3), Autosomal dominant; Waardenburg syndrome, type 2E, with or without neurologic involvement, 611584 (3), Autosomal dominant |
| *SOX11* | 600898 | Coffin-Siris syndrome 9, 615866 (3), Autosomal dominant |
| *SOX17* | 610928 | Vesicoureteral reflux 3, 613674 (3), Autosomal dominant |
| *SOX18* | 601618 | Hypotrichosis-lymphedema-telangiectasia syndrome, 607823 (3), Autosomal recessive; Hypotrichosis-lymphedema-telangiectasia-renal defect syndrome, 137940 (3), Autosomal dominant |
| *SOX2* | 184429 | Optic nerve hypoplasia and abnormalities of the central nervous system, 206900 (3), Autosomal dominant; Microphthalmia, syndromic 3, 206900 (3), Autosomal dominant |
| *SOX3* | 313430 | Mental retardation, X-linked, with isolated growth hormone deficiency, 300123 (3); Panhypopituitarism, X-linked, 312000 (3), X-linked |
| *SOX4* | 184430 | Coffin-Siris syndrome 10, 618506 (3), Autosomal dominant |
| *SOX5* | 604975 | Lamb-Shaffer syndrome, 616803 (3), Autosomal dominant |
| *SOX6* | 607257 | Tolchin-Le Caignec syndrome, 618971 (3), Autosomal dominant |
| *SOX9* | 608160 | Campomelic dysplasia with autosomal sex reversal, 114290 (3), Autosomal dominant; Acampomelic campomelic dysplasia, 114290 (3), Autosomal dominant; Campomelic dysplasia, 114290 (3), Autosomal dominant |
| *SP110* | 604457 | {Mycobacterium tuberculosis, susceptibility to}, 607948 (3); Hepatic venoocclusive disease with immunodeficiency, 235550 (3), Autosomal recessive |
| *SP7* | 606633 | Osteogenesis imperfecta, type XII, 613849 (3), Autosomal recessive |
| *SPAG1* | 603395 | Ciliary dyskinesia, primary, 28, 615505 (3), Autosomal recessive |
| *SPAG17* | 616554 | ?Spermatogenic failure 55, 619380 (3), Autosomal recessive |
| *SPARC* | 182120 | Osteogenesis imperfecta, type XVII, 616507 (3), Autosomal recessive |
| *SPART* | 607111 | Troyer syndrome, 275900 (3), Autosomal recessive |
| *SPAST* | 604277 | Spastic paraplegia 4, autosomal dominant, 182601 (3), Autosomal dominant |
| *SPATA16* | 609856 | ?Spermatogenic failure 6, 102530 (3), Autosomal recessive |
| *SPATA5* | 613940 | Epilepsy, hearing loss, and mental retardation syndrome, 616577 (3), Autosomal recessive |
| *SPATA7* | 609868 | Retinitis pigmentosa, juvenile, autosomal recessive, 604232 (3); Leber congenital amaurosis 3, 604232 (3) |
| *SPECC1L* | 614140 | Opitz GBBB syndrome, type II, 145410 (3), Autosomal dominant; Teebi hypertelorism syndrome, 145420 (3), Autosomal dominant; ?Facial clefting, oblique, 1, 600251 (3), Autosomal dominant |
| *SPEF2* | 610172 | Spermatogenic failure 43, 618751 (3), Autosomal recessive |
| *SPEG* | 615950 | Centronuclear myopathy 5, 615959 (3), Autosomal recessive |
| *SPEN* | 613484 | Radio-Tartaglia syndrome, 619312 (3), Autosomal dominant |
| *SPG11* | 610844 | Amyotrophic lateral sclerosis 5, juvenile, 602099 (3), Autosomal recessive; Charcot-Marie-Tooth disease, axonal, type 2X, 616668 (3), Autosomal recessive; Spastic paraplegia 11, autosomal recessive, 604360 (3), Autosomal recessive |
| *SPG21* | 608181 | Mast syndrome, 248900 (3), Autosomal recessive |
| *SPG7* | 602783 | Spastic paraplegia 7, autosomal recessive, 607259 (3), Autosomal recessive, Autosomal dominant |
| *SPINK1* | 167790 | Tropical calcific pancreatitis, 608189 (3), Autosomal recessive, Autosomal dominant; Pancreatitis, hereditary, 167800 (3), Autosomal dominant; {Fibrocalculous pancreatic diabetes, susceptibility to}, 608189 (3), Autosomal recessive, Autosomal dominant |
| *SPINK2* | 605753 | ?Spermatogenic failure 29, 618091 (3), Autosomal recessive |
| *SPINK5* | 605010 | Netherton syndrome, 256500 (3), Autosomal recessive |
| *SPINT2* | 605124 | Diarrhea 3, secretory sodium, congenital, syndromic, 270420 (3), Autosomal recessive |
| *SPNS2* | 612584 | ?Deafness, autosomal recessive 115, 618457 (3), Autosomal recessive |
| *SPOP* | 602650 | Nabais Sa-de Vries syndrome, type 1, 618828 (3), Autosomal dominant; Nabais Sa-de Vries syndrome, type 2, 618829 (3), Autosomal dominant |
| *SPR* | 182125 | Dystonia, dopa-responsive, due to sepiapterin reductase deficiency, 612716 (3), Autosomal recessive, ?Autosomal dominant |
| *SPRED1* | 609291 | Legius syndrome, 611431 (3), Autosomal dominant |
| *SPRTN* | 616086 | Ruijs-Aalfs syndrome, 616200 (3), Autosomal recessive |
| *SPRY2* | 602466 | {?IgA nephropathy, susceptibility to, 3}, 616818 (3), Autosomal dominant |
| *SPRY4* | 607984 | Hypogonadotropic hypogonadism 17 with or without anosmia, 615266 (3), Autosomal dominant |
| *SPTA1* | 182860 | Spherocytosis, type 3, 270970 (3), Autosomal recessive; Elliptocytosis-2, 130600 (3), Autosomal dominant; Pyropoikilocytosis, 266140 (3), Autosomal recessive |
| *SPTAN1* | 182810 | Developmental and epileptic encephalopathy 5, 613477 (3), Autosomal dominant |
| *SPTB* | 182870 | Anemia, neonatal hemolytic, fatal or near-fatal, 617948 (3); Elliptocytosis-3, 617948 (3); Spherocytosis, type 2, 616649 (3), Autosomal dominant |
| *SPTBN2* | 604985 | Spinocerebellar ataxia 5, 600224 (3), Autosomal dominant; Spinocerebellar ataxia, autosomal recessive 14, 615386 (3), Autosomal recessive |
| *SPTBN4* | 606214 | Neurodevelopmental disorder with hypotonia, neuropathy, and deafness, 617519 (3), Autosomal recessive |
| *SPTLC1* | 605712 | Neuropathy, hereditary sensory and autonomic, type IA, 162400 (3), Autosomal dominant |
| *SPTLC2* | 605713 | Neuropathy, hereditary sensory and autonomic, type IC, 613640 (3), Autosomal dominant |
| *SQOR* | 617658 | Sulfide:quinone oxidoreductase deficiency, 619221 (3), Autosomal recessive |
| *SQSTM1* | 601530 | Neurodegeneration with ataxia, dystonia, and gaze palsy, childhood-onset, 617145 (3), Autosomal recessive; Frontotemporal dementia and/or amyotrophic lateral sclerosis 3, 616437 (3), Autosomal dominant; Myopathy, distal, with rimmed vacuoles, 617158 (3), Autosomal dominant; Paget disease of bone 3, 167250 (3), Autosomal dominant |
| *SRC* | 190090 | ?Thrombocytopenia 6, 616937 (3), Autosomal dominant; Colon cancer, advanced, somatic, 114500 (3) |
| *SRCAP* | 611421 | Floating-Harbor syndrome, 136140 (3), Autosomal dominant |
| *SRD5A2* | 607306 | Pseudovaginal perineoscrotal hypospadias, 264600 (3), Autosomal recessive |
| *SRD5A3* | 611715 | Kahrizi syndrome, 612713 (3), Autosomal recessive; Congenital disorder of glycosylation, type Iq, 612379 (3), Autosomal recessive |
| *SREBF1* | 184756 | Ichthyosis, follicular, with atrichia and photophobia syndrome 2, 619016 (3), Autosomal dominant; Mucoepithelial dysplasia, hereditary, 158310 (3), Autosomal dominant |
| *SRGAP1* | 606523 | {Thyroid cancer, nonmedullary, 2}, 188470 (3), Somatic mutation, Autosomal dominant |
| *SRP54* | 604857 | Neutropenia, severe congenital, 8, autosomal dominant, 618752 (3), Autosomal dominant |
| *SRP72* | 602122 | Bone marrow failure syndrome 1, 614675 (3), Autosomal dominant |
| *SRPX2* | 300642 | ?Rolandic epilepsy, mental retardation, and speech dyspraxia, 300643 (3) |
| *SRY* | 480000 | 46XY sex reversal 1, 400044 (3), Y-linked; 46XX sex reversal 1, 400045 (4), X-linked dominant |
| *SS18* | 600192 | Sarcoma, synovial (1) |
| *SSBP1* | 600439 | Optic atrophy 13 with retinal and foveal abnormalities, 165510 (3), Autosomal dominant |
| *SSR4* | 300090 | Congenital disorder of glycosylation, type Iy, 300934 (3), X-linked recessive |
| *SSX1* | 312820 | ?Sarcoma, synovial, 300813 (3) |
| *SSX2* | 300192 | ?Sarcoma, synovial, 300813 (3) |
| *ST14* | 606797 | Ichthyosis, congenital, autosomal recessive 11, 602400 (3), Autosomal recessive |
| *ST3GAL3* | 606494 | Developmental and epileptic encephalopathy 15, 615006 (3), Autosomal recessive; Intellectual developmental disorder, autosomal recessive 12, 611090 (3), Autosomal recessive |
| *ST3GAL5* | 604402 | Salt and pepper developmental regression syndrome, 609056 (3), Autosomal recessive |
| *STAC3* | 615521 | Myopathy, congenital, Baily-Bloch, 255995 (3), Autosomal recessive |
| *STAG1* | 604358 | Mental retardation, autosomal dominant 47, 617635 (3), Autosomal dominant |
| *STAG2* | 300826 | Holoprosencephaly 13, X-linked, 301043 (3), X-linked dominant, X-linked recessive; Mullegama-Klein-Martinez syndrome, 301022 (3), X-linked |
| *STAG3* | 608489 | Premature ovarian failure 8, 615723 (3), Autosomal recessive |
| *STAMBP* | 606247 | Microcephaly-capillary malformation syndrome, 614261 (3), Autosomal recessive |
| *STAR* | 600617 | Lipoid adrenal hyperplasia, 201710 (3), Autosomal recessive |
| *STARD7* | 616712 | Epilepsy, familial adult myoclonic, 2, 607876 (3), Autosomal dominant |
| *STAT1* | 600555 | Immunodeficiency 31C, chronic mucocutaneous candidiasis, 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 |
| *STAT2* | 600556 | Pseudo-TORCH syndrome 3, 618886 (3), Autosomal recessive; Immunodeficiency 44, 616636 (3), Autosomal recessive |
| *STAT3* | 102582 | Hyper-IgE recurrent infection syndrome, 147060 (3), Autosomal dominant; Autoimmune disease, multisystem, infantile-onset, 1, 615952 (3), Autosomal dominant |
| *STAT4* | 600558 | {Systemic lupus erythematosus, susceptibility to, 11}, 612253 (3) |
| *STAT5B* | 604260 | Growth hormone insensitivity with immune dysregulation 1, autosomal recessive, 245590 (3), Autosomal recessive; Growth hormone insensitivity with immune dysregulation 2, autosomal dominant, 618985 (3), Autosomal dominant; Leukemia, acute promyelocytic, somatic, 102578 (3) |
| *STEAP3* | 609671 | ?Anemia, hypochromic microcytic, with iron overload 2, 615234 (3), Autosomal dominant |
| *STEEP1 (CXorf56)* | 301012 | ?Mental retardation, X-linked 107, 301013 (3), X-linked |
| *STIL* | 181590 | Microcephaly 7, primary, autosomal recessive, 612703 (3), Autosomal recessive |
| *STIM1* | 605921 | Myopathy, tubular aggregate, 1, 160565 (3), Autosomal dominant; Stormorken syndrome, 185070 (3), Autosomal dominant; Immunodeficiency 10, 612783 (3), Autosomal recessive |
| *STING1 (TMEM173)* | 612374 | STING-associated vasculopathy, infantile-onset, 615934 (3), Autosomal dominant |
| *STK11* | 602216 | Melanoma, malignant, somatic, 155600 (3); Pancreatic cancer, somatic, 260350 (3); Peutz-Jeghers syndrome, 175200 (3), Autosomal dominant; Testicular tumor, somatic, 273300 (3) |
| *STK36* | 607652 | ?Ciliary dyskinesia, primary, 46, 619436 (3), Autosomal recessive |
| *STK4* | 604965 | T-cell immunodeficiency, recurrent infections, autoimmunity, and cardiac malformations, 614868 (3) |
| *STN1* | 613128 | Cerebroretinal microangiopathy with calcifications and cysts 2, 617341 (3), Autosomal recessive |
| *STOX1* | 609397 | Preeclampsia/eclampsia 4, 609404 (3) |
| *STRA6* | 610745 | Microphthalmia, syndromic 9, 601186 (3), Autosomal recessive; Microphthalmia, isolated, with coloboma 8, 601186 (3), Autosomal recessive |
| *STRADA* | 608626 | Polyhydramnios, megalencephaly, and symptomatic epilepsy, 611087 (3), Autosomal recessive |
| *STRC* | 606440 | Deafness, autosomal recessive 16, 603720 (3), Autosomal recessive |
| *STS* | 300747 | Ichthyosis, X-linked, 308100 (3), X-linked recessive |
| *STT3A* | 601134 | Congenital disorder of glycosylation, type Iw, 615596 (3), Autosomal recessive |
| *STT3B* | 608605 | ?Congenital disorder of glycosylation, type Ix, 615597 (3), Autosomal recessive |
| *STUB1* | 607207 | ?Spinocerebellar ataxia 48, 618093 (3), Autosomal dominant; Spinocerebellar ataxia, autosomal recessive 16, 615768 (3), Autosomal recessive |
| *STX11* | 605014 | Hemophagocytic lymphohistiocytosis, familial, 4, 603552 (3), Autosomal recessive |
| *STX16* | 603666 | Pseudohypoparathyroidism, type IB, 603233 (3), Autosomal dominant |
| *STX1B* | 601485 | Generalized epilepsy with febrile seizures plus, type 9, 616172 (3), Autosomal dominant |
| *STX3* | 600876 | Retinal dystrophy and microvillus inclusion disease, 619446 (3), Autosomal recessive; Diarrhea 12, with microvillus atrophy, 619445 (3), Autosomal recessive |
| *STXBP1* | 602926 | Developmental and epileptic encephalopathy 4, 612164 (3), Autosomal dominant |
| *STXBP2* | 601717 | Hemophagocytic lymphohistiocytosis, familial, 5, with or without microvillus inclusion disease, 613101 (3) |
| *SUCLA2* | 603921 | Mitochondrial DNA depletion syndrome 5 (encephalomyopathic with or without methylmalonic aciduria), 612073 (3), Autosomal recessive |
| *SUCLG1* | 611224 | Mitochondrial DNA depletion syndrome 9 (encephalomyopathic type with methylmalonic aciduria), 245400 (3), Autosomal recessive |
| *SUFU* | 607035 | {Meningioma, familial, susceptibility to}, 607174 (3), Autosomal dominant; Joubert syndrome 32, 617757 (3), Autosomal recessive; Medulloblastoma, desmoplastic, 155255 (3), Somatic mutation, Autosomal recessive, Autosomal dominant; Basal cell nevus syndrome, 109400 (3), Autosomal dominant |
| *SUGCT* | 609187 | Glutaric aciduria III, 231690 (3), Autosomal recessive |
| *SULT2B1* | 604125 | Ichthyosis, congenital, autosomal recessive 14, 617571 (3), Autosomal recessive |
| *SUMF1* | 607939 | Multiple sulfatase deficiency, 272200 (3), Autosomal recessive |
| *SUMO1* | 601912 | ?Orofacial cleft 10, 613705 (3), Isolated cases |
| *SUMO4* | 608829 | {Diabetes mellitus, insulin-dependent, 5}, 600320 (3) |
| *SUN5* | 613942 | Spermatogenic failure 16, 617187 (3), Autosomal recessive |
| *SUOX* | 606887 | Sulfite oxidase deficiency, 272300 (3), Autosomal recessive |
| *SURF1* | 185620 | Charcot-Marie-Tooth disease, type 4K, 616684 (3), Autosomal recessive; Mitochondrial complex IV deficiency, nuclear type 1, 220110 (3), Autosomal recessive |
| *SUZ12* | 606245 | Imagawa-Matsumoto syndrome, 618786 (3), Autosomal dominant |
| *SVBP* | 617853 | Neurodevelopmental disorder with ataxia, hypotonia, and microcephaly, 618569 (3), Autosomal recessive |
| *SVIL* | 604126 | Myofibrillar myopathy 10, 619040 (3), Autosomal recessive |
| *SYCE1* | 611486 | ?Spermatogenic failure 15, 616950 (3), Autosomal recessive; ?Premature ovarian failure 12, 616947 (3), Autosomal recessive |
| *SYCP2* | 604105 | Spermatogenic failure 1, 258150 (3), Autosomal recessive |
| *SYCP3* | 604759 | Pregnancy loss, recurrent, 4, 270960 (3), Autosomal dominant; Spermatogenic failure 4, 270960 (3), Autosomal dominant |
| *SYK* | 600085 | Immunodeficiency 82 with systemic inflammation, 619381 (3), Autosomal dominant |
| *SYN1* | 313440 | Intellectual developmental disorder, X-linked 50, 300115 (3), X-linked; Epilepsy, X-linked, with variable learning disabilities and behavior disorders, 300491 (3), X-linked dominant, X-linked recessive |
| *SYN2* | 600755 | {Schizophrenia, susceptibility to}, 181500 (3), Autosomal dominant |
| *SYNE1* | 608441 | Arthrogryposis multiplex congenita 3, myogenic type, 618484 (3), Autosomal recessive; Emery-Dreifuss muscular dystrophy 4, autosomal dominant, 612998 (3), Autosomal dominant; Spinocerebellar ataxia, autosomal recessive 8, 610743 (3), Autosomal recessive |
| *SYNE2* | 608442 | Emery-Dreifuss muscular dystrophy 5, autosomal dominant, 612999 (3), Autosomal dominant |
| *SYNE4* | 615535 | Deafness, autosomal recessive 76, 615540 (3), Autosomal recessive |
| *SYNGAP1* | 603384 | Mental retardation, autosomal dominant 5, 612621 (3), Autosomal dominant |
| *SYNJ1* | 604297 | Parkinson disease 20, early-onset, 615530 (3), Autosomal recessive; Developmental and epileptic encephalopathy 53, 617389 (3), Autosomal recessive |
| *SYP* | 313475 | Mental retardation, X-linked 96, 300802 (3), X-linked recessive |
| *SYT1* | 185605 | Baker-Gordon syndrome, 618218 (3), Autosomal dominant |
| *SYT14* | 610949 | ?Spinocerebellar ataxia, autosomal recessive 11, 614229 (3), Autosomal recessive |
| *SYT2* | 600104 | Myasthenic syndrome, congenital, 7, presynaptic, 616040 (3), Autosomal dominant |
| *SZT2* | 615463 | Developmental and epileptic encephalopathy 18, 615476 (3), Autosomal recessive |
| *TAB2* | 605101 | Congenital heart defects, nonsyndromic, 2, 614980 (3), Autosomal dominant |
| *TAC3* | 162330 | Hypogonadotropic hypogonadism 10 with or without anosmia, 614839 (3), Autosomal recessive |
| *TACO1* | 612958 | Mitochondrial complex IV deficiency, nuclear type 8, 619052 (3), Autosomal recessive |
| *TACR3* | 162332 | Hypogonadotropic hypogonadism 11 with or without anosmia, 614840 (3), Autosomal recessive |
| *TACSTD2* | 137290 | Corneal dystrophy, gelatinous drop-like, 204870 (3), Autosomal recessive |
| *TAF1* | 313650 | Mental retardation, X-linked, syndromic 33, 300966 (3), X-linked recessive; Dystonia-Parkinsonism, X-linked, 314250 (3), X-linked recessive |
| *TAF13* | 600774 | Mental retardation, autosomal recessive 60, 617432 (3), Autosomal recessive |
| *TAF15* | 601574 | Chondrosarcoma, extraskeletal myxoid, 612237 (1) |
| *TAF2* | 604912 | Mental retardation, autosomal recessive 40, 615599 (3), Autosomal recessive |
| *TAF4B* | 601689 | ?Spermatogenic failure 13, 615841 (3), Autosomal recessive |
| *TAF6* | 602955 | Alazami-Yuan syndrome, 617126 (3), Autosomal recessive |
| *TAFAZZIN (TAZ)* | 300394 | Barth syndrome, 302060 (3), X-linked recessive |
| *TAGAP* | 609667/612521 | -/{Diabetes mellitus, insulin-dependent, 21}, 612521 (2) |
| *TAL1* | 187040 | Leukemia, T-cell acute lymphocytic, somatic, 613065 (3) |
| *TAL2* | 186855 | Leukemia, T-cell acute lymphocytic, somatic, 613065 (3) |
| *TALDO1* | 602063 | Transaldolase deficiency, 606003 (3), Autosomal recessive |
| *TANC2* | 615047 | Intellectual developmental disorder with autistic features and language delay, with or without seizures, 618906 (3), Autosomal dominant |
| *TANGO2* | 616830 | Metabolic encephalomyopathic crises, recurrent, with rhabdomyolysis, cardiac arrhythmias, and neurodegeneration, 616878 (3), Autosomal recessive |
| *TAP1* | 170260 | Bare lymphocyte syndrome, type I, 604571 (3), Autosomal recessive |
| *TAP2* | 170261 | Bare lymphocyte syndrome, type I, due to TAP2 deficiency, 604571 (3), Autosomal recessive |
| *TAPBP* | 601962 | Bare lymphocyte syndrome, type I, 604571 (3), Autosomal recessive |
| *TAPT1* | 612758 | Osteochondrodysplasia, complex lethal, Symoens-Barnes-Gistelinck type, 616897 (3), Autosomal recessive |
| *TARDBP* | 605078 | Frontotemporal lobar degeneration, TARDBP-related, 612069 (3), Autosomal dominant; Amyotrophic lateral sclerosis 10, with or without FTD, 612069 (3), Autosomal dominant |
| *TARS1 (TARS)* | 187790 | Trichothiodystrophy 7, nonphotosensitive, 618546 (3), Autosomal recessive |
| *TARS2* | 612805 | ?Combined oxidative phosphorylation deficiency 21, 615918 (3), Autosomal recessive |
| *TAS2R16* | 604867 | [Beta-glycopyranoside tasting], (3) {Alcohol dependence, susceptibility to}, 617956 (3), Autosomal dominant |
| *TAS2R38* | 607751 | [Phenylthiocarbamide tasting], 171200 (3), Autosomal dominant |
| *TASP1* | 608270 | Suleiman-El-Hattab syndrome, 618950 (3), Autosomal recessive |
| *TAT* | 613018 | Tyrosinemia, type II, 276600 (3), Autosomal recessive |
| *TBC1D20* | 611663 | Warburg micro syndrome 4, 615663 (3), Autosomal recessive |
| *TBC1D23* | 617687 | Pontocerebellar hypoplasia, type 11, 617695 (3), Autosomal recessive |
| *TBC1D24* | 613577 | Deafness, autosomal recessive 86, 614617 (3), Autosomal recessive; Epilepsy, rolandic, with proxysmal exercise-induce dystonia and writer's cramp, 608105 (3), Autosomal recessive; Myoclonic epilepsy, infantile, familial, 605021 (3), Autosomal recessive; Deafness, autosomal dominant 65, 616044 (3), Autosomal dominant; Developmental and epileptic encephalopathy 16, 615338 (3), Autosomal recessive; DOORS syndrome, 220500 (3), Autosomal recessive |
| *TBC1D2B* | 619152 | Neurodevelopmental disorder with seizures and gingival overgrowth, 619323 (3), Autosomal recessive |
| *TBC1D4* | 612465 | {Diabetes mellitus, noninsulin-dependent, 5}, 616087 (3) |
| *TBC1D7* | 612655 | Macrocephaly/megalencephaly syndrome, autosomal recessive, 248000 (3), Autosomal recessive |
| *TBC1D8B* | 301027 | Nephrotic syndrome, type 20, 301028 (3), X-linked |
| *TBCD* | 604649 | Encephalopathy, progressive, early-onset, with brain atrophy and thin corpus callosum, 617193 (3), Autosomal recessive |
| *TBCE* | 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 |
| *TBCK* | 616899 | Hypotonia, infantile, with psychomotor retardation and characteristic facies 3, 616900 (3), Autosomal recessive |
| *TBK1* | 604834 | {Encephalopathy, acute, infection-induced (herpes-specific), susceptibility to, 8}, 617900 (3), Autosomal dominant; Frontotemporal dementia and/or amyotrophic lateral sclerosis 4, 616439 (3), Autosomal dominant |
| *TBL1X* | 300196 | Hypothyroidism, congenital, nongoitrous, 8, 301033 (3), X-linked |
| *TBL1XR1* | 608628 | Pierpont syndrome, 602342 (3), Autosomal dominant; Mental retardation, autosomal dominant 41, 616944 (3), Autosomal dominant |
| *TBL1Y* | 400033 | ?Deafness, Y-linked 2, 400047 (3), Y-linked |
| *TBP* | 600075 | Spinocerebellar ataxia 17, 607136 (3), Autosomal dominant; {Parkinson disease, susceptibility to}, 168600 (3), Multifactorial, Autosomal dominant |
| *TBR1* | 604616 | Intellectual developmental disorder with autism and speech delay, 606053 (3), Autosomal dominant |
| *TBX1* | 602054 | Tetralogy of Fallot, 187500 (3), Autosomal dominant; DiGeorge syndrome, 188400 (3), Autosomal dominant; Conotruncal anomaly face syndrome, 217095 (3); Velocardiofacial syndrome, 192430 (3), Autosomal dominant |
| *TBX15* | 604127 | Cousin syndrome, 260660 (3), Autosomal recessive |
| *TBX18* | 604613 | Congenital anomalies of kidney and urinary tract 2, 143400 (3), Autosomal dominant |
| *TBX19* | 604614 | Adrenocorticotropic hormone deficiency, 201400 (3), Autosomal recessive |
| *TBX2* | 600747 | Vertebral anomalies and variable endocrine and T-cell dysfunction, 618223 (3), Autosomal dominant |
| *TBX20* | 606061 | Atrial septal defect 4, 611363 (3) |
| *TBX21* | 604895 | Asthma and nasal polyps, 208550 (3), Autosomal recessive; {Asthma, aspirin-induced, susceptibility to}, 208550 (3), Autosomal recessive |
| *TBX22* | 300307 | Cleft palate with ankyloglossia, 303400 (3), X-linked; ?Abruzzo-Erickson syndrome, 302905 (3), X-linked |
| *TBX3* | 601621 | Ulnar-mammary syndrome, 181450 (3), Autosomal dominant |
| *TBX4* | 601719 | Ischiocoxopodopatellar syndrome with or without pulmonary arterial hypertension, 147891 (3), Autosomal dominant; Amelia, posterior, with pelvic and pulmonary hypoplasia syndrome, 601360 (3), Autosomal recessive |
| *TBX5* | 601620 | Holt-Oram syndrome, 142900 (3), Autosomal dominant |
| *TBX6* | 602427 | Spondylocostal dysostosis 5, 122600 (3), Autosomal recessive, Autosomal dominant |
| *TBXA2R* | 188070 | {Bleeding disorder, platelet-type, 13, susceptibility to}, 614009 (3), Autosomal dominant |
| *TBXAS1* | 274180 | Ghosal hematodiaphyseal syndrome, 231095 (3), Autosomal recessive |
| *TBXT* | 601397 | Sacral agenesis with vertebral anomalies, 615709 (3), Autosomal recessive; {Neural tube defects, susceptibility to}, 182940 (3), Autosomal dominant |
| *TCAP* | 604488 | Cardiomyopathy, hypertrophic, 25, 607487 (3), Autosomal dominant; Muscular dystrophy, limb-girdle, autosomal recessive 7, 601954 (3), Autosomal recessive |
| *TCF12* | 600480 | Craniosynostosis 3, 615314 (3), Autosomal dominant |
| *TCF20* | 603107 | Developmental delay with variable intellectual impairment and behavioral abnormalities, 618430 (3), Autosomal dominant |
| *TCF3* | 147141 | Agammaglobulinemia 8, autosomal dominant, 616941 (3), Autosomal dominant |
| *TCF4* | 602272 | Pitt-Hopkins syndrome, 610954 (3), Autosomal dominant; Corneal dystrophy, Fuchs endothelial, 3, 613267 (3), Autosomal dominant |
| *TCF7L2* | 602228 | {Diabetes mellitus, type 2, susceptibility to}, 125853 (3), Autosomal dominant |
| *TCHH* | 190370 | ?Uncombable hair syndrome 3, 617252 (3), Autosomal recessive |
| *TCIRG1* | 604592 | Osteopetrosis, autosomal recessive 1, 259700 (3), Autosomal recessive |
| *TCL1A* | 186960 | Leukemia/lymphoma, T-cell, 186960 (2) |
| *TCL1B* | 603769 | Leukemia/lymphoma, T-cell, 603769 (2) |
| *TCN2* | 613441 | Transcobalamin II deficiency, 275350 (3), Autosomal recessive |
| *TCOF1* | 606847 | Treacher Collins syndrome 1, 154500 (3), Autosomal dominant |
| *TCTN1* | 609863 | Joubert syndrome 13, 614173 (3), Autosomal recessive |
| *TCTN2* | 613846 | Joubert syndrome 24, 616654 (3), Autosomal recessive; ?Meckel syndrome 8, 613885 (3), Autosomal recessive |
| *TCTN3* | 613847 | Joubert syndrome 18, 614815 (3), Autosomal recessive; Orofaciodigital syndrome IV, 258860 (3), Autosomal recessive |
| *TDGF1* | 187395 | Forebrain defects (3) |
| *TDO2* | 191070 | [?Hypertryptophanemia], 600627 (3), Autosomal recessive |
| *TDP1* | 607198 | ?Spinocerebellar ataxia, autosomal recessive, with axonal neuropathy 1, 607250 (3), Autosomal recessive |
| *TDP2* | 605764 | Spinocerebellar ataxia, autosomal recessive 23, 616949 (3), Autosomal recessive |
| *TDRD7* | 611258 | Cataract 36, 613887 (3), Autosomal recessive |
| *TDRD9* | 617963 | ?Spermatogenic failure 30, 618110 (3), Autosomal recessive |
| *TEAD1* | 189967 | Sveinsson chorioretinal atrophy, 108985 (3), Autosomal dominant |
| *TECPR2* | 615000 | Spastic paraplegia 49, autosomal recessive, 615031 (3), Autosomal recessive |
| *TECR* | 610057 | Mental retardation, autosomal recessive 14, 614020 (3), Autosomal recessive |
| *TECRL* | 617242 | Ventricular tachycardia, catecholaminergic polymorphic, 3, 614021 (3), Autosomal recessive |
| *TECTA* | 602574 | Deafness, autosomal dominant 8/12, 601543 (3), Autosomal dominant; Deafness, autosomal recessive 21, 603629 (3), Autosomal recessive |
| *TEK* | 600221 | Venous malformations, multiple cutaneous and mucosal, 600195 (3), Autosomal dominant; Glaucoma 3, primary congenital, E, 617272 (3), Autosomal dominant |
| *TELO2* | 611140 | You-Hoover-Fong syndrome, 616954 (3), Autosomal recessive |
| *TENM3* | 610083 | Microphthalmia, syndromic 15, 615145 (3), Autosomal recessive; ?Microphthalmia, isolated, with coloboma 9, 615145 (3), Autosomal recessive |
| *TENM4* | 610084 | Essential tremor, hereditary, 5, 616736 (3), Autosomal dominant |
| *TENT5A* | 611357 | Osteogenesis imperfecta, type XVIII, 617952 (3), Autosomal recessive |
| *TERT* | 187270 | {Pulmonary fibrosis and/or bone marrow failure, telomere-related, 1}, 614742 (3), Autosomal dominant; {Melanoma, cutaneous malignant, 9}, 615134 (3); {Dyskeratosis congenita, autosomal dominant 2}, 613989 (3), Autosomal recessive, Autosomal dominant; {Dyskeratosis congenita, autosomal recessive 4}, 613989 (3), Autosomal recessive, Autosomal dominant; {Leukemia, acute myeloid}, 601626 (3), Somatic mutation, Autosomal dominant |
| *TET2* | 612839 | Myelodysplastic syndrome, somatic, 614286 (3); Immunodeficiency 75, 619126 (3), Autosomal recessive |
| *TET3* | 613555 | Beck-Fahrner syndrome, 618798 (3), Autosomal recessive, Autosomal dominant |
| *TEX11* | 300311 | Spermatogenic failure, X-linked, 2, 309120 (3), X-linked recessive |
| *TEX14* | 605792 | Spermatogenic failure 23, 617707 (3), Autosomal recessive |
| *TEX15* | 605795 | Spermatogenic failure 25, 617960 (3), Autosomal recessive |
| *TF* | 190000 | Atransferrinemia, 209300 (3), Autosomal recessive |
| *TFAM* | 600438 | ?Mitochondrial DNA depletion syndrome 15 (hepatocerebral type), 617156 (3), Autosomal recessive |
| *TFAP2A* | 107580 | Branchiooculofacial syndrome, 113620 (3), Autosomal dominant |
| *TFAP2B* | 601601 | Patent ductus arteriosus 2, 617035 (3), Autosomal dominant; Char syndrome, 169100 (3), Autosomal dominant |
| *TFE3* | 314310 | Renal cell carcinoma, papillary, 1, 300854 (3) |
| *TFG* | 602498 | ?Spastic paraplegia 57, autosomal recessive, 615658 (3), Autosomal recessive; Hereditary motor and sensory neuropathy, Okinawa type, 604484 (3), Autosomal dominant |
| *TFR2* | 604720 | Hemochromatosis, type 3, 604250 (3), Autosomal recessive |
| *TFRC* | 190010 | Immunodeficiency 46, 616740 (3), Autosomal recessive |
| *TG* | 188450 | {Autoimmune thyroid disease, susceptibility to, 3}, 608175 (3); Thyroid dyshormonogenesis 3, 274700 (3), Autosomal recessive |
| *TGDS* | 616146 | Catel-Manzke syndrome, 616145 (3), Autosomal recessive |
| *TGFB1* | 190180 | Inflammatory bowel disease, immunodeficiency, and encephalopathy, 618213 (3), Autosomal recessive; Camurati-Engelmann disease, 131300 (3), Autosomal dominant; {Cystic fibrosis lung disease, modifier of}, 219700 (3), Autosomal recessive |
| *TGFB2* | 190220 | Loeys-Dietz syndrome 4, 614816 (3), Autosomal dominant |
| *TGFB3* | 190230 | Arrhythmogenic right ventricular dysplasia 1, 107970 (3), Autosomal dominant; Loeys-Dietz syndrome 5, 615582 (3), Autosomal dominant |
| *TGFBI* | 601692 | Corneal dystrophy, Avellino type, 607541 (3), Autosomal dominant; Corneal dystrophy, Reis-Bucklers type, 608470 (3); Corneal dystrophy, Thiel-Behnke type, 602082 (3), Autosomal dominant; Corneal dystrophy, Groenouw type I, 121900 (3), Autosomal dominant; Corneal dystrophy, epithelial basement membrane, 121820 (3), Autosomal dominant; Corneal dystrophy, lattice type I, 122200 (3), Autosomal dominant; Corneal dystrophy, lattice type IIIA, 608471 (3), Autosomal dominant |
| *TGFBR1* | 190181 | {Multiple self-healing squamous epithelioma, susceptibility to}, 132800 (3), Autosomal dominant; Loeys-Dietz syndrome 1, 609192 (3), Autosomal dominant |
| *TGFBR2* | 190182 | Loeys-Dietz syndrome 2, 610168 (3), Autosomal dominant; Colorectal cancer, hereditary nonpolyposis, type 6, 614331 (3); Esophageal cancer, somatic, 133239 (3) |
| *TGIF1* | 602630 | Holoprosencephaly 4, 142946 (3), Autosomal dominant |
| *TGM1* | 190195 | Ichthyosis, congenital, autosomal recessive 1, 242300 (3), Autosomal recessive |
| *TGM3* | 600238 | ?Uncombable hair syndrome 2, 617251 (3), Autosomal recessive |
| *TGM5* | 603805 | Peeling skin syndrome 2, 609796 (3), Autosomal recessive |
| *TGM6* | 613900 | Spinocerebellar ataxia 35, 613908 (3), Autosomal dominant |
| *TH* | 191290 | Segawa syndrome, recessive, 605407 (3), Autosomal recessive |
| *THAP1* | 609520 | Dystonia 6, torsion, 602629 (3), Autosomal dominant |
| *THBD* | 188040 | Thrombophilia due to thrombomodulin defect, 614486 (3); {Hemolytic uremic syndrome, atypical, susceptibility to, 6}, 612926 (3), Autosomal dominant |
| *THBS2* | 188061 | {Lumbar disc herniation, susceptibility to}, 603932 (3) |
| *THG1L* | 618802 | Spinocerebellar ataxia, autosomal recessive 28, 618800 (3), Autosomal recessive |
| *THOC2* | 300395 | Mental retardation, X-linked 12/35, 300957 (3), X-linked recessive |
| *THOC6* | 615403 | Beaulieu-Boycott-Innes syndrome, 613680 (3), Autosomal recessive |
| *THPO* | 600044 | Thrombocythemia 1, 187950 (3), Autosomal dominant |
| *THRA* | 190120 | Hypothyroidism, congenital, nongoitrous, 6, 614450 (3), Autosomal dominant |
| *THRB* | 190160 | Thyroid hormone resistance, autosomal recessive, 274300 (3), Autosomal recessive; Thyroid hormone resistance, 188570 (3), Autosomal dominant; Thyroid hormone resistance, selective pituitary, 145650 (3), Autosomal dominant |
| *THSD1* | 616821 | ?Aneurysm, intracranial berry, 12, 618734 (3), Autosomal dominant |
| *TIA1* | 603518 | Welander distal myopathy, 604454 (3), Autosomal recessive, Autosomal dominant; Amyotrophic lateral sclerosis 26 with or without frontotemporal dementia, 619133 (3) |
| *TICAM1* | 607601 | {Encephalopathy, acute, infection-induced (herpes-specific), susceptibility to, 6}, 614850 (3), Autosomal recessive, Autosomal dominant |
| *TIE1* | 600222 | Lymphatic malformation 11, 619401 (3), Autosomal dominant |
| *TIMM22* | 607251 | ?Combined oxidative phosphorylation deficiency 43, 618851 (3), Autosomal recessive |
| *TIMM50* | 607381 | 3-methylglutaconic aciduria, type IX, 617698 (3), Autosomal recessive |
| *TIMM8A* | 300356 | Mohr-Tranebjaerg syndrome, 304700 (3), X-linked recessive |
| *TIMMDC1* | 615534 | Mitochondrial complex I deficiency, nuclear type 31, 618251 (3), Autosomal recessive |
| *TIMP3* | 188826 | Sorsby fundus dystrophy, 136900 (3), Autosomal dominant |
| *TINF2* | 604319 | Dyskeratosis congenita, autosomal dominant 3, 613990 (3), Autosomal dominant; Revesz syndrome, 268130 (3), Autosomal dominant |
| *TIRAP* | 606252 | {Malaria, protection against}, 611162 (3); {Tuberculosis, protection against}, 607948 (3); {Bacteremia, protection against}, 614382 (3) |
| *TJP2* | 607709 | Hypercholanemia, familial 1, 607748 (3), Autosomal recessive; Cholestasis, progressive familial intrahepatic 4, 615878 (3), Autosomal recessive |
| *TK2* | 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 |
| *TKFC* | 615844 | Triokinase and FMN cyclase deficiency syndrome, 618805 (3), Autosomal recessive |
| *TKT* | 606781 | Short stature, developmental delay, and congenital heart defects, 617044 (3), Autosomal recessive |
| *TLE6* | 612399 | Preimplantation embryonic lethality, 616814 (3), Autosomal recessive |
| *TLK2* | 608439 | Mental retardation, autosomal dominant 57, 618050 (3), Autosomal dominant |
| *TLL1* | 606742 | Atrial septal defect 6, 613087 (3), Autosomal dominant |
| *TLR1* | 601194 | {Leprosy, susceptibility to, 5}, 613223 (3); {Leprosy, protection against}, 613223 (3) |
| *TLR2* | 603028 | {Colorectal cancer, susceptibility to}, 114500 (3), Somatic mutation, Autosomal dominant; {Leprosy, susceptibility to}, 246300 (3), Autosomal dominant; {Mycobacterium tuberculosis, susceptibility to}, 607948 (3) |
| *TLR3* | 603029 | {HIV1 infection, resistance to}, 609423 (3); {Immunodeficiency 83, susceptibility to viral infections}, 613002 (3), Autosomal recessive, Autosomal dominant |
| *TLR4* | 603030/611488 | -/Macular degeneration, age-related, 10, 611488 (2) |
| *TLR5* | 603031 | {Melioidosis, susceptibility to}, 615557 (3), Autosomal dominant; {Systemic lupus erythematosus, susceptibility to, 1}, 601744 (3); {Systemic lupus erythematosus, resistance to}, 601744 (3); {Legionnaire disease, susceptibility to}, 608556 (3) |
| *TLR7* | 300365 | Immunodeficiency 74, COVID19-related, X-linked, 301051 (3), X-linked recessive |
| *TM4SF20* | 615404 | {Specific language impairment 5}, 615432 (3), Autosomal dominant |
| *TMC1* | 606706 | Deafness, autosomal dominant 36, 606705 (3), Autosomal dominant; Deafness, autosomal recessive 7, 600974 (3), Autosomal recessive |
| *TMC6* | 605828 | Epidermodysplasia verruciformis, 226400 (3), Autosomal recessive |
| *TMC8* | 605829 | Epidermodysplasia verruciformis 2, 618231 (3), Autosomal recessive |
| *TMCO1* | 614123 | Craniofacial dysmorphism, skeletal anomalies, and mental retardation syndrome, 213980 (3), Autosomal recessive |
| *TMEM106B* | 613413 | Leukodystrophy, hypomyelinating, 16, 617964 (3), Autosomal dominant |
| *TMEM107* | 616183 | Orofaciodigital syndrome XVI, 617563 (3), Autosomal recessive; Meckel syndrome 13, 617562 (3), Autosomal recessive; ?Joubert syndrome 29, 617562 (3), Autosomal recessive |
| *TMEM126A* | 612988 | Optic atrophy 7, 612989 (3), Autosomal recessive |
| *TMEM126B* | 615533 | Mitochondrial complex I deficiency, nuclear type 29, 618250 (3), Autosomal recessive |
| *TMEM127* | 613403 | {Pheochromocytoma, susceptibility to}, 171300 (3), Autosomal dominant |
| *TMEM132E* | 616178 | Deafness, autosomal recessive 99, 618481 (3), Autosomal recessive |
| *TMEM138* | 614459 | Joubert syndrome 16, 614465 (3), Autosomal recessive |
| *TMEM165* | 614726 | Congenital disorder of glycosylation, type IIk, 614727 (3), Autosomal recessive |
| *TMEM199* | 616815 | Congenital disorder of glycosylation, type IIp, 616829 (3), Autosomal recessive |
| *TMEM216* | 613277 | Joubert syndrome 2, 608091 (3), Autosomal recessive; Meckel syndrome 2, 603194 (3), Autosomal recessive |
| *TMEM231* | 614949 | Joubert syndrome 20, 614970 (3), Autosomal recessive; Meckel syndrome 11, 615397 (3), Autosomal recessive |
| *TMEM237* | 614423 | Joubert syndrome 14, 614424 (3), Autosomal recessive |
| *TMEM240* | 616101 | Spinocerebellar ataxia 21, 607454 (3), Autosomal dominant |
| *TMEM251* | 619332 | Dysostosis multiplex, Ain-Naz type, 619345 (3), Autosomal recessive |
| *TMEM260* | 617449 | Structural heart defects and renal anomalies syndrome, 617478 (3), Autosomal recessive |
| *TMEM38B* | 611236 | Osteogenesis imperfecta, type XIV, 615066 (3) |
| *TMEM43* | 612048 | Arrhythmogenic right ventricular dysplasia 5, 604400 (3), Autosomal dominant; Emery-Dreifuss muscular dystrophy 7, AD, 614302 (3), Autosomal dominant |
| *TMEM63A* | 618685 | Leukodystrophy, hypomyelinating, 19, transient infantile, 618688 (3), Autosomal dominant |
| *TMEM67* | 609884 | Nephronophthisis 11, 613550 (3), Autosomal recessive; {Bardet-Biedl syndrome 14, modifier of}, 615991 (3), Autosomal recessive; Joubert syndrome 6, 610688 (3), Autosomal recessive; Meckel syndrome 3, 607361 (3), Autosomal recessive; ?RHYNS syndrome, 602152 (3), Autosomal recessive; COACH syndrome 1, 216360 (3), Autosomal recessive |
| *TMEM70* | 612418 | Mitochondrial complex V (ATP synthase) deficiency, nuclear type 2, 614052 (3), Autosomal recessive |
| *TMEM94* | 618163 | Intellectual developmental disorder with cardiac defects and dysmorphic facies, 618316 (3), Autosomal recessive |
| *TMEM98* | 615949 | Nanophthalmos 4, 615972 (3), Autosomal dominant |
| *TMIE* | 607237 | Deafness, autosomal recessive 6, 600971 (3), Autosomal recessive |
| *TMLHE* | 300777 | {Autism, susceptibility to, X-linked 6}, 300872 (3), X-linked recessive |
| *TMPRSS15* | 606635 | Enterokinase deficiency, 226200 (3), Autosomal recessive |
| *TMPRSS3* | 605511 | Deafness, autosomal recessive 8/10, 601072 (3), Autosomal recessive |
| *TMPRSS6* | 609862 | Iron-refractory iron deficiency anemia, 206200 (3), Autosomal recessive |
| *TMTC3* | 617218 | Lissencephaly 8, 617255 (3), Autosomal recessive |
| *TMX2* | 616715 | Neurodevelopmental disorder with microcephaly, cortical malformations, and spasticity, 618730 (3), Autosomal recessive |
| *TNC* | 187380 | Deafness, autosomal dominant 56, 615629 (3), Autosomal dominant |
| *TNF* | 191160 | {Migraine without aura, susceptibility to}, 157300 (3), Autosomal dominant; {Dementia, vascular, susceptibility to} (3); {Asthma, susceptibility to}, 600807 (3), Autosomal dominant; {Septic shock, susceptibility to} (3); {Malaria, cerebral, susceptibility to}, 611162 (3) |
| *TNFAIP3* | 191163 | Autoinflammatory syndrome, familial, Behcet-like, 616744 (3), Autosomal dominant |
| *TNFRSF10B* | 603612 | Squamous cell carcinoma, head and neck, 275355 (3), Autosomal recessive |
| *TNFRSF11A* | 603499 | Osteopetrosis, autosomal recessive 7, 612301 (3), Autosomal recessive; {Paget disease of bone 2, early-onset}, 602080 (3), Autosomal dominant; Osteolysis, familial expansile, 174810 (3), Autosomal dominant |
| *TNFRSF11B* | 602643 | Paget disease of bone 5, juvenile-onset, 239000 (3), Autosomal recessive |
| *TNFRSF13B* | 604907 | Immunodeficiency, common variable, 2, 240500 (3), Autosomal recessive, Autosomal dominant; Immunoglobulin A deficiency 2, 609529 (3) |
| *TNFRSF13C* | 606269 | Immunodeficiency, common variable, 4, 613494 (3), Autosomal recessive |
| *TNFRSF1A* | 191190 | {Multiple sclerosis, susceptibility to, 5}, 614810 (3); Periodic fever, familial, 142680 (3), Autosomal dominant |
| *TNFRSF4* | 600315 | ?Immunodeficiency 16, 615593 (3), Autosomal recessive |
| *TNFSF11* | 602642 | Osteopetrosis, autosomal recessive 2, 259710 (3), Autosomal recessive |
| *TNFSF4* | 603594 | {Myocardial infarction, susceptibility to}, 608446 (3) |
| *TNIK* | 610005 | Mental retardation, autosomal recessive 54, 617028 (3), Autosomal recessive |
| *TNNC1* | 191040 | Cardiomyopathy, dilated, 1Z, 611879 (3); Cardiomyopathy, hypertrophic, 13, 613243 (3), Autosomal dominant |
| *TNNI2* | 191043 | Arthrogryposis, distal, type 2B1, 601680 (3), Autosomal dominant |
| *TNNI3* | 191044 | ?Cardiomyopathy, dilated, 2A, 611880 (3), Autosomal recessive; Cardiomyopathy, hypertrophic, 7, 613690 (3), Autosomal dominant; Cardiomyopathy, familial restrictive, 1, 115210 (3), Autosomal dominant; Cardiomyopathy, dilated, 1FF, 613286 (3) |
| *TNNI3K* | 613932 | Cardiac conduction disease with or without dilated cardiomyopathy, 616117 (3), Autosomal dominant |
| *TNNT1* | 191041 | Nemaline myopathy 5, Amish type, 605355 (3), Autosomal recessive |
| *TNNT2* | 191045 | Cardiomyopathy, dilated, 1D, 601494 (3), Autosomal dominant; Cardiomyopathy, hypertrophic, 2, 115195 (3), Autosomal dominant; Cardiomyopathy, familial restrictive, 3, 612422 (3), Autosomal dominant; Left ventricular noncompaction 6, 601494 (3), Autosomal dominant |
| *TNNT3* | 600692 | Arthrogryposis, distal, type 2B2, 618435 (3), Autosomal dominant |
| *TNPO3* | 610032 | Muscular dystrophy, limb-girdle, autosomal dominant 2, 608423 (3), Autosomal dominant |
| *TNRC6A* | 610739 | ?Epilepsy, familial adult myoclonic, 6, 618074 (3), Autosomal dominant |
| *TNRC6B* | 610740 | Global developmental delay with speech and behavioral abnormalities, 619243 (3), Autosomal dominant |
| *TNXB* | 600985 | Ehlers-Danlos syndrome, classic-like, 1, 606408 (3), Autosomal recessive; Vesicoureteral reflux 8, 615963 (3), Autosomal dominant |
| *TOE1* | 613931 | Pontocerebellar hypoplasia, type 7, 614969 (3), Autosomal recessive |
| *TOGARAM1* | 617618 | Joubert syndrome 37, 619185 (3), Autosomal recessive |
| *TONSL* | 604546 | Spondyloepimetaphyseal dysplasia, sponastrime type, 271510 (3), Autosomal recessive |
| *TOP1* | 126420 | DNA topoisomerase I, camptothecin-resistant (3) |
| *TOP2A* | 126430 | DNA topoisomerase II, resistance to inhibition of, by amsacrine (3) |
| *TOP3A* | 601243 | ?Progressive external ophthalmoplegia with mitochondrial DNA deletions, autosomal recessive 5, 618098 (3), Autosomal recessive; Microcephaly, growth restriction, and increased sister chromatid exchange 2, 618097 (3), Autosomal recessive |
| *TOPORS* | 609507 | Retinitis pigmentosa 31, 609923 (3) |
| *TOR1A* | 605204 | {Dystonia-1, modifier of} (3); Arthrogryposis multiplex congenita 5, 618947 (3), Autosomal recessive; Dystonia-1, torsion, 128100 (3), Autosomal dominant |
| *TOR1AIP1* | 614512 | ?Muscular dystrophy, autosomal recessive, with rigid spine and distal joint contractures, 617072 (3), Autosomal recessive |
| *TP53* | 191170 | {Basal cell carcinoma 7}, 614740 (3), Autosomal dominant; {Adrenocortical carcinoma, pediatric}, 202300 (3), Autosomal dominant; Hepatocellular carcinoma, somatic, 114550 (3); Breast cancer, somatic, 114480 (3); Li-Fraumeni syndrome, 151623 (3), Autosomal dominant; Pancreatic cancer, somatic, 260350 (3); Nasopharyngeal carcinoma, somatic, 607107 (3); {Osteosarcoma}, 259500 (3), Somatic mutation; {Choroid plexus papilloma}, 260500 (3), Autosomal dominant; {Colorectal cancer}, 114500 (3), Somatic mutation, Autosomal dominant; {Glioma susceptibility 1}, 137800 (3), Somatic mutation, Autosomal dominant; Bone marrow failure syndrome 5, 618165 (3), Autosomal dominant |
| *TP53RK* | 608679 | Galloway-Mowat syndrome 4, 617730 (3), Autosomal recessive |
| *TP63* | 603273 | Ectrodactyly, ectodermal dysplasia, and cleft lip/palate syndrome 3, 604292 (3), Autosomal dominant; Hay-Wells syndrome, 106260 (3), Autosomal dominant; Split-hand/foot malformation 4, 605289 (3), Autosomal dominant; Orofacial cleft 8, 618149 (3); Rapp-Hodgkin syndrome, 129400 (3), Autosomal dominant; ADULT syndrome, 103285 (3), Autosomal dominant; Limb-mammary syndrome, 603543 (3), Autosomal dominant |
| *TPCN2* | 612163 | [Skin/hair/eye pigmentation 10, blond/brown hair], 612267 (3) |
| *TPH2* | 607478 | {Unipolar depression, susceptibility to}, 608516 (3); {Attention deficit-hyperactivity disorder, susceptibility to, 7}, 613003 (3) |
| *TPI1* | 190450 | Hemolytic anemia due to triosephosphate isomerase deficiency, 615512 (3), Autosomal recessive |
| *TPK1* | 606370 | Thiamine metabolism dysfunction syndrome 5 (episodic encephalopathy type), 614458 (3), Autosomal recessive |
| *TPM1* | 191010 | Left ventricular noncompaction 9, 611878 (3), Autosomal dominant; Cardiomyopathy, hypertrophic, 3, 115196 (3), Autosomal dominant; Cardiomyopathy, dilated, 1Y, 611878 (3), Autosomal dominant |
| *TPM2* | 190990 | Arthrogryposis, distal, type 2B4, 108120 (3), Autosomal dominant; Arthrogryposis, distal, type 1A, 108120 (3), Autosomal dominant; Nemaline myopathy 4, autosomal dominant, 609285 (3), Autosomal dominant; CAP myopathy 2, 609285 (3), Autosomal dominant |
| *TPM3* | 191030 | CAP myopathy 1, 609284 (3), Autosomal recessive, Autosomal dominant; Myopathy, congenital, with fiber-type disproportion, 255310 (3), Autosomal recessive, Autosomal dominant; Nemaline myopathy 1, autosomal dominant or recessive, 609284 (3), Autosomal recessive, Autosomal dominant |
| *TPMT* | 187680 | {Thiopurines, poor metabolism of, 1}, 610460 (3), Autosomal recessive |
| *TPO* | 606765 | Thyroid dyshormonogenesis 2A, 274500 (3), Autosomal recessive |
| *TPP1* | 607998 | Ceroid lipofuscinosis, neuronal, 2, 204500 (3), Autosomal recessive; Spinocerebellar ataxia, autosomal recessive 7, 609270 (3), Autosomal recessive |
| *TPP2* | 190470 | Immunodeficiency 78 with autoimmunity and developmental delay, 619220 (3), Autosomal recessive |
| *TPRKB* | 608680 | Galloway-Mowat syndrome 5, 617731 (3), Autosomal recessive |
| *TPRN* | 613354 | Deafness, autosomal recessive 79, 613307 (3), Autosomal recessive |
| *TRAC* | 186880 | Immunodeficiency 7, TCR-alpha/beta deficient, 615387 (3), Autosomal recessive |
| *TRAF3* | 601896 | {?Encephalopathy, acute, infection-induced (herpes-specific), susceptibility to, 5}, 614849 (3) |
| *TRAF3IP1* | 607380 | Senior-Loken syndrome 9, 616629 (3), Autosomal recessive |
| *TRAF3IP2* | 607043 | ?Candidiasis, familial, 8, 615527 (3), Autosomal recessive; {Psoriasis susceptibility 13}, 614070 (3) |
| *TRAF7* | 606692 | Cardiac, facial, and digital anomalies with developmental delay, 618164 (3), Autosomal dominant |
| *TRAIP* | 605958 | Seckel syndrome 9, 616777 (3), Autosomal recessive |
| *TRAK1* | 608112 | Developmental and epileptic encephalopathy 68, 618201 (3), Autosomal recessive |
| *TRAPPC11* | 614138 | Muscular dystrophy, limb-girdle, autosomal recessive 18, 615356 (3), Autosomal recessive |
| *TRAPPC12* | 614139 | Encephalopathy, progressive, early-onset, with brain atrophy and spasticity, 617669 (3), Autosomal recessive |
| *TRAPPC14 (C7orf43)* | 618350 | ?Microcephaly 25, primary, autosomal recessive, 618351 (3), Autosomal recessive |
| *TRAPPC2* | 300202 | Spondyloepiphyseal dysplasia tarda, 313400 (3), X-linked recessive |
| *TRAPPC2L* | 610970 | Encephalopathy, progressive, early-onset, with episodic rhabdomyolysis, 618331 (3), Autosomal recessive |
| *TRAPPC4* | 610971 | Neurodevelopmental disorder with epilepsy, spasticity, and brain atrophy, 618741 (3), Autosomal recessive |
| *TRAPPC6B* | 610397 | Neurodevelopmental disorder with microcephaly, epilepsy, and brain atrophy, 617862 (3), Autosomal recessive |
| *TRAPPC9* | 611966 | Mental retardation, autosomal recessive 13, 613192 (3), Autosomal recessive |
| *TRDN* | 603283 | Ventricular tachycardia, catecholaminergic polymorphic, 5, with or without muscle weakness, 615441 (3), Autosomal recessive |
| *TREH* | 275360 | Trehalase deficiency, 612119 (3), Autosomal recessive |
| *TREM2* | 605086 | Polycystic lipomembranous osteodysplasia with sclerosing leukoencephalopathy 2, 618193 (3), Autosomal recessive |
| *TREX1* | 606609 | Vasculopathy, retinal, with cerebral leukoencephalopathy and systemic manifestations, 192315 (3), Autosomal dominant; Aicardi-Goutieres syndrome 1, dominant and recessive, 225750 (3), Autosomal recessive, Autosomal dominant; {Systemic lupus erythematosus, susceptibility to}, 152700 (3), Autosomal dominant; Chilblain lupus, 610448 (3), Autosomal dominant |
| *TRH* | 613879 | Thyrotropin-releasing hormone deficiency, 275120 (1), Autosomal recessive |
| *TRHR* | 188545 | Hypothyroidism, congenital, nongoitrous, 7, 618573 (3), Autosomal recessive |
| *TRIM2* | 614141 | Charcot-Marie-Tooth disease, type 2R, 615490 (3), Autosomal recessive |
| *TRIM32* | 602290 | ?Bardet-Biedl syndrome 11, 615988 (3), Autosomal recessive; Muscular dystrophy, limb-girdle, autosomal recessive 8, 254110 (3), Autosomal recessive |
| *TRIM36* | 609317 | ?Anencephaly 1, 206500 (3), Autosomal recessive |
| *TRIM37* | 605073 | Mulibrey nanism, 253250 (3), Autosomal recessive |
| *TRIM44* | 612298 | ?Aniridia 3, 617142 (3), Autosomal dominant |
| *TRIM71* | 618570 | Hydrocephalus, congenital communicating, 1, 618667 (3), Autosomal dominant |
| *TRIO* | 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 |
| *TRIOBP* | 609761 | Deafness, autosomal recessive 28, 609823 (3), Autosomal recessive |
| *TRIP11* | 604505 | Odontochondrodysplasia 1, 184260 (3), Autosomal recessive; Achondrogenesis, type IA, 200600 (3), Autosomal recessive |
| *TRIP12* | 604506 | Mental retardation, autosomal dominant 49, 617752 (3), Autosomal dominant |
| *TRIP13* | 604507 | Oocyte maturation defect 9, 619011 (3), Autosomal recessive; Mosaic variegated aneuploidy syndrome 3, 617598 (3), Autosomal recessive |
| *TRIP4* | 604501 | ?Muscular dystrophy, congenital, Davignon-Chauveau type, 617066 (3), Autosomal recessive; Spinal muscular atrophy with congenital bone fractures 1, 616866 (3), Autosomal recessive |
| *TRIT1* | 617840 | Combined oxidative phosphorylation deficiency 35, 617873 (3), Autosomal recessive |
| *TRMT1* | 611669 | Mental retardation, autosomal recessive 68, 618302 (3), Autosomal recessive |
| *TRMT10A* | 616013 | Microcephaly, short stature, and impaired glucose metabolism 1, 616033 (3), Autosomal recessive |
| *TRMT10C* | 615423 | Combined oxidative phosphorylation deficiency 30, 616974 (3), Autosomal recessive |
| *TRMT5* | 611023 | Combined oxidative phosphorylation deficiency 26, 616539 (3), Autosomal recessive |
| *TRMU* | 610230 | {Deafness, mitochondrial, modifier of}, 580000 (3), Mitochondrial; Liver failure, transient infantile, 613070 (3), Autosomal recessive |
| *TRNT1* | 612907 | Sideroblastic anemia with B-cell immunodeficiency, periodic fevers, and developmental delay, 616084 (3), Autosomal recessive; Retinitis pigmentosa and erythrocytic microcytosis, 616959 (3), Autosomal recessive |
| *TRPA1* | 604775 | ?Episodic pain syndrome, familial, 1, 615040 (3), Autosomal dominant |
| *TRPC3* | 602345 | ?Spinocerebellar ataxia 41, 616410 (3), Autosomal dominant |
| *TRPC6* | 603652 | Glomerulosclerosis, focal segmental, 2, 603965 (3), Autosomal dominant |
| *TRPM1* | 603576 | Night blindness, congenital stationary (complete), 1C, autosomal recessive, 613216 (3) |
| *TRPM4* | 606936 | Progressive familial heart block, type IB, 604559 (3), Autosomal dominant; Erythrokeratodermia veriabilis et progressiva 6, 618531 (3), Autosomal dominant |
| *TRPM6* | 607009 | Hypomagnesemia 1, intestinal, 602014 (3), Autosomal recessive |
| *TRPM7* | 605692 | {Amyotrophic lateral sclerosis-parkinsonism/dementia complex, susceptibility to}, 105500 (3), Autosomal dominant |
| *TRPS1* | 604386 | Trichorhinophalangeal syndrome, type III, 190351 (3), Autosomal dominant; Trichorhinophalangeal syndrome, type I, 190350 (3), Autosomal dominant |
| *TRPV3* | 607066 | ?Palmoplantar keratoderma, nonepidermolytic, focal 2, 616400 (3), Autosomal dominant; Olmsted syndrome 1, 614594 (3), Autosomal dominant |
| *TRPV4* | 605427 | Spondylometaphyseal dysplasia, Kozlowski type, 184252 (3), Autosomal dominant; Digital arthropathy-brachydactyly, familial, 606835 (3), Autosomal dominant; [Sodium serum level QTL 1], 613508 (3); SED, Maroteaux type, 184095 (3), Autosomal dominant; Metatropic dysplasia, 156530 (3), Autosomal dominant; Scapuloperoneal spinal muscular atrophy, 181405 (3), Autosomal dominant; Hereditary motor and sensory neuropathy, type IIc, 606071 (3), Autosomal dominant; ?Avascular necrosis of femoral head, primary, 2, 617383 (3), Autosomal dominant; Neuronopathy, distal hereditary motor, type VIII, 600175 (3), Autosomal dominant; Parastremmatic dwarfism, 168400 (3), Autosomal dominant; Brachyolmia type 3, 113500 (3), Autosomal dominant |
| *TRPV6* | 606680 | Hyperparathyroidism, transient neonatal, 618188 (3), Autosomal recessive |
| *TRRAP* | 603015 | ?Deafness, autosomal dominant 75, 618778 (3), Autosomal dominant; Developmental delay with or without dysmorphic facies and autism, 618454 (3), Autosomal dominant |
| *TSC1* | 605284 | Focal cortical dysplasia, type II, somatic, 607341 (3); Tuberous sclerosis-1, 191100 (3), Autosomal dominant; Lymphangioleiomyomatosis, 606690 (3) |
| *TSC2* | 191092 | Lymphangioleiomyomatosis, somatic, 606690 (3); ?Focal cortical dysplasia, type II, somatic, 607341 (3); Tuberous sclerosis-2, 613254 (3), Autosomal dominant |
| *TSEN15* | 608756 | Pontocerebellar hypoplasia, type 2F, 617026 (3), Autosomal recessive |
| *TSEN2* | 608753 | Pontocerebellar hypoplasia type 2B, 612389 (3), Autosomal recessive |
| *TSEN34* | 608754 | ?Pontocerebellar hypoplasia type 2C, 612390 (3), Autosomal recessive |
| *TSEN54* | 608755 | Pontocerebellar hypoplasia type 2A, 277470 (3), Autosomal recessive; Pontocerebellar hypoplasia type 4, 225753 (3), Autosomal recessive; ?Pontocerebellar hypoplasia type 5, 610204 (3), Autosomal recessive |
| *TSFM* | 604723 | Combined oxidative phosphorylation deficiency 3, 610505 (3), Autosomal recessive |
| *TSGA10* | 607166 | ?Spermatogenic failure 26, 617961 (3), Autosomal recessive |
| *TSHB* | 188540 | Hypothyroidism, congenital, nongoitrous 4, 275100 (3), Autosomal recessive |
| *TSHR* | 603372 | Hyperthyroidism, familial gestational, 603373 (3), Autosomal dominant; Hyperthyroidism, nonautoimmune, 609152 (3), Autosomal dominant; Thyroid adenoma, hyperfunctioning, somatic (3); Thyroid carcinoma with thyrotoxicosis (3); Hypothyroidism, congenital, nongoitrous, 1, 275200 (3), Autosomal recessive |
| *TSHZ1* | 614427 | Aural atresia, congenital, 607842 (3), Autosomal dominant |
| *TSPAN12* | 613138 | Exudative vitreoretinopathy 5, 613310 (3), Autosomal dominant |
| *TSPAN7* | 300096 | Mental retardation, X-linked 58, 300210 (3), X-linked recessive |
| *TSPEAR* | 612920 | ?Deafness, autosomal recessive 98, 614861 (3), Autosomal recessive; Ectodermal dysplasia 14, hair/tooth type with or without hypohidrosis, 618180 (3), Autosomal recessive |
| *TSPYL1* | 604714 | Sudden infant death with dysgenesis of the testes syndrome, 608800 (3), Autosomal recessive |
| *TSR2* | 300945 | ?Diamond-Blackfan anemia 14 with mandibulofacial dysostosis, 300946 (3), X-linked recessive |
| *TTBK2* | 611695 | Spinocerebellar ataxia 11, 604432 (3), Autosomal dominant |
| *TTC12* | 610732 | Ciliary dyskinesia, primary, 45, 618801 (3), Autosomal recessive |
| *TTC19* | 613814 | Mitochondrial complex III deficiency, nuclear type 2, 615157 (3), Autosomal recessive |
| *TTC21A* | 611430 | Spermatogenic failure 37, 618429 (3), Autosomal recessive |
| *TTC21B* | 612014 | Short-rib thoracic dysplasia 4 with or without polydactyly, 613819 (3), Autosomal recessive; Nephronophthisis 12, 613820 (3), Autosomal recessive, Autosomal dominant |
| *TTC29* | 618735 | Spermatogenic failure 42, 618745 (3), Autosomal recessive |
| *TTC37* | 614589 | Trichohepatoenteric syndrome 1, 222470 (3), Autosomal recessive |
| *TTC5* | 619014 | Neurodevelopmental disorder with cerebral atrophy and variable facial dysmorphism, 619244 (3), Autosomal recessive |
| *TTC7A* | 609332 | Gastrointestinal defects and immunodeficiency syndrome, 243150 (3), Autosomal recessive |
| *TTC8* | 608132 | Bardet-Biedl syndrome 8, 615985 (3), Autosomal recessive; ?Retinitis pigmentosa 51, 613464 (3), Autosomal recessive |
| *TTI2* | 614426 | Mental retardation, autosomal recessive 39, 615541 (3), Autosomal recessive |
| *TTLL5* | 612268 | Cone-rod dystrophy 19, 615860 (3), Autosomal recessive |
| *TTN* | 188840 | Muscular dystrophy, limb-girdle, autosomal recessive 10, 608807 (3), Autosomal recessive; Cardiomyopathy, familial hypertrophic, 9, 613765 (3), Autosomal dominant; Tibial muscular dystrophy, tardive, 600334 (3), Autosomal dominant; Salih myopathy, 611705 (3), Autosomal recessive; Cardiomyopathy, dilated, 1G, 604145 (3); Myopathy, myofibrillar, 9, with early respiratory failure, 603689 (3), Autosomal dominant |
| *TTPA* | 600415 | Ataxia with isolated vitamin E deficiency, 277460 (3), Autosomal recessive |
| *TTR* | 176300 | Amyloidosis, hereditary, transthyretin-related, 105210 (3), Autosomal dominant; Carpal tunnel syndrome, familial, 115430 (3), Autosomal dominant; [Dystransthyretinemic hyperthyroxinemia], 145680 (3), Autosomal dominant |
| *TUB* | 601197 | ?Retinal dystrophy and obesity, 616188 (3), Autosomal recessive |
| *TUBA1A* | 602529 | Lissencephaly 3, 611603 (3), Autosomal dominant |
| *TUBA3D* | 617878 | Keratoconus 9, 617928 (3), Autosomal dominant |
| *TUBA4A* | 191110 | Amyotrophic lateral sclerosis 22 with or without frontotemporal dementia, 616208 (3), Autosomal dominant |
| *TUBB* | 191130 | Symmetric circumferential skin creases, congenital, 1, 156610 (3), Autosomal dominant; Cortical dysplasia, complex, with other brain malformations 6, 615771 (3), Autosomal dominant |
| *TUBB1* | 612901 | Macrothrombocytopenia, autosomal dominant, TUBB1-related, 613112 (3), Autosomal dominant |
| *TUBB2A* | 615101 | Cortical dysplasia, complex, with other brain malformations 5, 615763 (3), Autosomal dominant |
| *TUBB2B* | 612850 | Cortical dysplasia, complex, with other brain malformations 7, 610031 (3), Autosomal dominant |
| *TUBB3* | 602661 | Fibrosis of extraocular muscles, congenital, 3A, 600638 (3), Autosomal dominant; Cortical dysplasia, complex, with other brain malformations 1, 614039 (3), Autosomal dominant |
| *TUBB4A* | 602662 | Dystonia 4, torsion, autosomal dominant, 128101 (3), Autosomal dominant; Leukodystrophy, hypomyelinating, 6, 612438 (3), Autosomal dominant |
| *TUBB4B* | 602660 | Leber congenital amaurosis with early-onset deafness, 617879 (3), Autosomal dominant |
| *TUBB6* | 615103 | ?Facial palsy, congenitla, with ptosis and velopharyngeal dysfunction, 617732 (3), Autosomal dominant |
| *TUBB8* | 616768 | Oocyte maturation defect 2, 616780 (3), Autosomal recessive, Autosomal dominant |
| *TUBG1* | 191135 | Cortical dysplasia, complex, with other brain malformations 4, 615412 (3), Autosomal dominant |
| *TUBGCP2* | 617817 | Pachygyria, microcephaly, developmental delay, and dysmorphic facies, with or without seizures, 618737 (3), Autosomal recessive |
| *TUBGCP4* | 609610 | Microcephaly and chorioretinopathy, autosomal recessive, 3, 616335 (3), Autosomal recessive |
| *TUBGCP6* | 610053 | Microcephaly and chorioretinopathy, autosomal recessive, 1, 251270 (3), Autosomal recessive |
| *TUFM* | 602389 | Combined oxidative phosphorylation deficiency 4, 610678 (3), Autosomal recessive |
| *TULP1* | 602280 | Leber congenital amaurosis 15, 613843 (3), Autosomal recessive; Retinitis pigmentosa 14, 600132 (3), Autosomal recessive |
| *TUSC3* | 601385 | Mental retardation, autosomal recessive 7, 611093 (3), Autosomal recessive |
| *TWIST1* | 601622 | Craniosynostosis 1, 123100 (3), Autosomal dominant; Robinow-Sorauf syndrome, 180750 (3), Autosomal dominant; Sweeney-Cox syndrome, 617746 (3), Autosomal dominant; Saethre-Chotzen syndrome with or without eyelid anomalies, 101400 (3), Autosomal dominant |
| *TWIST2* | 607556 | Ablepharon-macrostomia syndrome, 200110 (3), Autosomal dominant; Barber-Say syndrome, 209885 (3), Autosomal dominant; Focal facial dermal dysplasia 3, Setleis type, 227260 (3), Autosomal recessive |
| *TWNK* | 606075 | Mitochondrial DNA depletion syndrome 7 (hepatocerebral type), 271245 (3), Autosomal recessive; Progressive external ophthalmoplegia with mitochondrial DNA deletions, autosomal dominant 3, 609286 (3), Autosomal dominant; Perrault syndrome 5, 616138 (3), Autosomal recessive |
| *TXN2* | 609063 | ?Combined oxidative phosphorylation deficiency 29, 616811 (3), Autosomal recessive |
| *TXNL4A* | 611595 | Burn-McKeown syndrome, 608572 (3), Autosomal recessive |
| *TXNRD2* | 606448 | ?Glucocorticoid deficiency 5, 617825 (3), Autosomal recessive |
| *TYK2* | 176941 | Immunodeficiency 35, 611521 (3), Autosomal recessive |
| *TYMP* | 131222 | Mitochondrial DNA depletion syndrome 1 (MNGIE type), 603041 (3), Autosomal recessive |
| *TYR* | 606933 | [Skin/hair/eye pigmentation 3, light/dark/freckling skin], 601800 (3), Autosomal dominant; [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 IB, 606952 (3), Autosomal recessive; Waardenburg syndrome/albinism, digenic, 103470 (3); Albinism, oculocutaneous, type IA, 203100 (3), Autosomal recessive |
| *TYROBP* | 604142 | Polycystic lipomembranous osteodysplasia with sclerosing leukoencephalopathy 1, 221770 (3), Autosomal recessive |
| *TYRP1* | 115501 | [Skin/hair/eye pigmentation, variation in, 11 (Melanesian blond hair)], 612271 (3); Albinism, oculocutaneous, type III, 203290 (3), Autosomal recessive |
| *UBA1* | 314370 | Spinal muscular atrophy, X-linked 2, infantile, 301830 (3), X-linked recessive; VEXAS syndrome, somatic, 301054 (3) |
| *UBA5* | 610552 | ?Spinocerebellar ataxia, autosomal recessive 24, 617133 (3), Autosomal recessive; Developmental and epileptic encephalopathy 44, 617132 (3), Autosomal recessive |
| *UBAP1* | 609787 | Spastic paraplegia 80, autosomal dominant, 618418 (3), Autosomal dominant |
| *UBE2A* | 312180 | Mental retardation, X-linked syndromic, Nascimento-type, 300860 (3), X-linked recessive |
| *UBE2T* | 610538 | Fanconi anemia, complementation group T, 616435 (3), Autosomal recessive |
| *UBE3A* | 601623 | Angelman syndrome, 105830 (3), Autosomal dominant |
| *UBE3B* | 608047 | Kaufman oculocerebrofacial syndrome, 244450 (3), Autosomal recessive |
| *UBIAD1* | 611632 | Corneal dystrophy, Schnyder type, 121800 (3), Autosomal dominant |
| *UBQLN2* | 300264 | Amyotrophic lateral sclerosis 15, with or without frontotemporal dementia, 300857 (3), X-linked dominant |
| *UBR1* | 605981 | Johanson-Blizzard syndrome, 243800 (3), Autosomal recessive |
| *UBR7* | 613816 | Li-Campeau syndrome, 619189 (3), Autosomal recessive |
| *UBTF* | 600673 | Neurodegeneration, childhood-onset, with brain atrophy, 617672 (3), Autosomal dominant |
| *UCHL1* | 191342 | Spastic paraplegia 79, autosomal recessive, 615491 (3), Autosomal recessive; {?Parkinson disease 5, susceptibility to}, 613643 (3), Autosomal dominant |
| *UCP2* | 601693 | {Obesity, susceptibility to, BMIQ4}, 607447 (3) |
| *UCP3* | 602044 | {Obesity, severe, and type II diabetes}, 601665 (3), Multifactorial, Autosomal recessive, Autosomal dominant |
| *UFC1* | 610554 | Neurodevelopmental disorder with spasticity and poor growth, 618076 (3), Autosomal recessive |
| *UFM1* | 610553 | Leukodystrophy, hypomyelinating, 14, 617899 (3), Autosomal recessive |
| *UFSP2* | 611482 | ?Hip dysplasia, Beukes type, 142669 (3), Autosomal dominant; ?Spondyloepimetaphyseal dysplasia, Di Rocco type, 617974 (3), Autosomal dominant |
| *UGDH* | 603370 | Developmental and epileptic encephalopathy 84, 618792 (3), Autosomal recessive |
| *UGP2* | 191760 | Developmental and epileptic encephalopathy 83, 618744 (3), Autosomal recessive |
| *UGT1A1* | 191740 | Crigler-Najjar syndrome, type I, 218800 (3), Autosomal recessive; [Bilirubin, serum level of, QTL1], 601816 (3); Hyperbilirubinemia, familial transient neonatal, 237900 (3), Autosomal recessive; Crigler-Najjar syndrome, type II, 606785 (3), Autosomal recessive; [Gilbert syndrome], 143500 (3), Autosomal recessive |
| *UGT2B17* | 601903 | {Bone mineral density QTL 12, osteoporosis}, 612560 (3) |
| *UMOD* | 191845 | Tubulointerstitial kidney disease, autosomal dominant, 1, 162000 (3), Autosomal dominant |
| *UMPS* | 613891 | Orotic aciduria, 258900 (3), Autosomal recessive |
| *UNC119* | 604011 | ?Immunodeficiency 13, 615518 (3), Autosomal dominant; ?Cone-rod dystrophy (3) |
| *UNC13D* | 608897 | Hemophagocytic lymphohistiocytosis, familial, 3, 608898 (3), Autosomal recessive |
| *UNC45A* | 611219 | Osteootohepatoenteric syndrome, 619377 (3), Autosomal recessive |
| *UNC45B* | 611220 | ?Cataract 43, 616279 (3), Autosomal dominant; Myofibrillar myopathy 11, 619178 (3), Autosomal recessive |
| *UNC80* | 612636 | Hypotonia, infantile, with psychomotor retardation and characteristic facies 2, 616801 (3), Autosomal recessive |
| *UNC93B1* | 608204 | {Encephalopathy, acute, infection-induced (herpes-specific), susceptibility to, 1}, 610551 (3) |
| *UNG* | 191525 | Immunodeficiency with hyper IgM, type 5, 608106 (3), Autosomal recessive |
| *UPB1* | 606673 | Beta-ureidopropionase deficiency, 613161 (3), Autosomal recessive |
| *UPF3B* | 300298 | Mental retardation, X-linked, syndromic 14, 300676 (3), X-linked recessive |
| *UQCC2* | 614461 | Mitochondrial complex III deficiency, nuclear type 7, 615824 (3), Autosomal recessive |
| *UQCC3* | 616097 | ?Mitochondrial complex III deficiency, nuclear type 9, 616111 (3), Autosomal recessive |
| *UQCRB* | 191330 | Mitochondrial complex III deficiency, nuclear type 3, 615158 (3), Autosomal recessive |
| *UQCRC1* | 191328 | Parkinsonism with polyneuropathy, 619279 (3), Autosomal dominant |
| *UQCRC2* | 191329 | Mitochondrial complex III deficiency, nuclear type 5, 615160 (3), Autosomal recessive |
| *UQCRFS1* | 191327 | Mitochondrial complex III deficiency, nuclear type 10, 618775 (3), Autosomal recessive |
| *UQCRQ* | 612080 | Mitochondrial complex III deficiency, nuclear type 4, 615159 (3), Autosomal recessive |
| *UROC1* | 613012 | ?Urocanase deficiency, 276880 (3), Autosomal recessive |
| *UROD* | 613521 | Porphyria, hepatoerythropoietic, 176100 (3), Autosomal recessive, Autosomal dominant; Porphyria cutanea tarda, 176100 (3), Autosomal recessive, Autosomal dominant |
| *UROS* | 606938 | Porphyria, congenital erythropoietic, 263700 (3), Autosomal recessive |
| *USB1* | 613276 | Poikiloderma with neutropenia, 604173 (3), Autosomal recessive |
| *USF1* | 191523 | {Hyperlipidemia, familial combined, susceptibility to}, 602491 (3) |
| *USH1C* | 605242 | Usher syndrome, type 1C, 276904 (3), Autosomal recessive; Deafness, autosomal recessive 18A, 602092 (3), Autosomal recessive |
| *USH1G* | 607696 | Usher syndrome, type 1G, 606943 (3), Autosomal recessive |
| *USH2A* | 608400 | Usher syndrome, type 2A, 276901 (3), Autosomal recessive; Retinitis pigmentosa 39, 613809 (3) |
| *USP18* | 607057 | Pseudo-TORCH syndrome 2, 617397 (3), Autosomal recessive |
| *USP27X* | 300975 | Mental retardation, X-linked 105, 300984 (3), X-linked recessive |
| *USP45* | 618439 | ?Leber congenital amaurosis 19, 618513 (3), Autosomal recessive |
| *USP7* | 602519 | Hao-Fountain syndrome, 616863 (3), Autosomal dominant |
| *USP8* | 603158 | Pituitary adenoma 4, ACTH-secreting, somatic, 219090 (3) |
| *USP9X* | 300072 | Mental retardation, X-linked 99, syndromic, female-restricted, 300968 (3), X-linked dominant; Mental retardation, X-linked 99, 300919 (3), X-linked recessive |
| *USP9Y* | 400005 | Spermatogenic failure, Y-linked, 2, 415000 (3), Y-linked |
| *UTP4* | 607456/604901 | -/North American Indian childhood cirrhosis, 604901 (2) |
| *UVSSA* | 614632 | UV-sensitive syndrome 3, 614640 (3), Autosomal recessive |
| *VAC14* | 604632 | Striatonigral degeneration, childhood-onset, 617054 (3), Autosomal recessive |
| *VAMP1* | 185880 | Myasthenic syndrome, congenital, 25, 618323 (3), Autosomal recessive; Spastic ataxia 1, autosomal dominant, 108600 (3), Autosomal dominant |
| *VAMP2* | 185881 | Neurodevelopmental disorder with hypotonia and autistic features with or without hyperkinetic movements, 618760 (3), Autosomal dominant |
| *VANGL1* | 610132 | {Neural tube defects, susceptibility to}, 182940 (3), Autosomal dominant; Caudal regression syndrome, 600145 (3), Autosomal dominant |
| *VANGL2* | 600533 | Neural tube defects, 182940 (3), Autosomal dominant |
| *VAPB* | 605704 | Spinal muscular atrophy, late-onset, Finkel type, 182980 (3), Autosomal dominant; Amyotrophic lateral sclerosis 8, 608627 (3), Autosomal dominant |
| *VARS1 (VARS)* | 192150 | Neurodevelopmental disorder with microcephaly, seizures, and cortical atrophy, 617802 (3), Autosomal recessive |
| *VARS2* | 612802 | Combined oxidative phosphorylation deficiency 20, 615917 (3), Autosomal recessive |
| *VAX1* | 604294 | ?Microphthalmia, syndromic 11, 614402 (3), Autosomal recessive |
| *VCAN* | 118661 | Wagner syndrome 1, 143200 (3), Autosomal dominant |
| *VCL* | 193065 | Cardiomyopathy, dilated, 1W, 611407 (3); Cardiomyopathy, hypertrophic, 15, 613255 (3), Autosomal dominant |
| *VCP* | 601023 | Frontotemporal dementia and/or amyotrophic lateral sclerosis 6, 613954 (3); Charcot-Marie-Tooth disease, type 2Y, 616687 (3), Autosomal dominant; Inclusion body myopathy with early-onset Paget disease and frontotemporal dementia 1, 167320 (3), Autosomal dominant |
| *VDR* | 601769 | Rickets, vitamin D-resistant, type IIA, 277440 (3), Autosomal recessive |
| *VEGFA* | 192240 | {Microvascular complications of diabetes 1}, 603933 (3) |
| *VEGFC* | 601528 | Lymphatic malformation 4, 615907 (3), Autosomal dominant |
| *VHL* | 608537 | Hemangioblastoma, cerebellar, somatic (3); Erythrocytosis, familial, 2, 263400 (3), Autosomal recessive; von Hippel-Lindau syndrome, 193300 (3), Autosomal dominant; Renal cell carcinoma, somatic, 144700 (3); Pheochromocytoma, 171300 (3), Autosomal dominant |
| *VIM* | 193060 | Cataract 30, pulverulent, 116300 (3), Autosomal dominant |
| *VIPAS39* | 613401 | Arthrogryposis, renal dysfunction, and cholestasis 2, 613404 (3), Autosomal recessive |
| *VKORC1* | 608547 | Vitamin K-dependent clotting factors, combined deficiency of, 2, 607473 (3); Warfarin resistance, 122700 (3), Autosomal dominant |
| *VLDLR* | 192977 | Cerebellar hypoplasia and mental retardation with or without quadrupedal locomotion 1, 224050 (3), Autosomal recessive |
| *VMA21* | 300913 | Myopathy, X-linked, with excessive autophagy, 310440 (3), X-linked recessive |
| *VNN1* | 603570 | [High density lipoprotein cholesterol level QTL 8] (3) |
| *VPS11* | 608549 | Leukodystrophy, hypomyelinating, 12, 616683 (3), Autosomal recessive |
| *VPS13A* | 605978 | Choreoacanthocytosis, 200150 (3), Autosomal recessive |
| *VPS13B* | 607817 | Cohen syndrome, 216550 (3), Autosomal recessive |
| *VPS13C* | 608879 | Parkinson disease 23, autosomal recessive, early onset, 616840 (3), Autosomal recessive |
| *VPS13D* | 608877 | Spinocerebellar ataxia, autosomal recessive 4, 607317 (3), Autosomal recessive |
| *VPS16* | 608550 | Dystonia 30, 619291 (3), Autosomal dominant |
| *VPS33A* | 610034 | Mucopolysaccharidosis-plus syndrome, 617303 (3), Autosomal recessive |
| *VPS33B* | 608552 | Arthrogryposis, renal dysfunction, and cholestasis 1, 208085 (3), Autosomal recessive |
| *VPS35* | 601501 | {Parkinson disease 17}, 614203 (3), Autosomal dominant |
| *VPS35L* | 618981 | Ritscher-Schinzel syndrome 3, 619135 (3) |
| *VPS37A* | 609927 | Spastic paraplegia 53, autosomal recessive, 614898 (3), Autosomal recessive |
| *VPS41* | 605485 | Spinocerebellar ataxia, autosomal recessive 29, 619389 (3), Autosomal recessive |
| *VPS45* | 610035 | Neutropenia, severe congenital, 5, autosomal recessive, 615285 (3), Autosomal recessive |
| *VPS4A* | 609982 | CIMDAG syndrome, 619273 (3), Autosomal dominant |
| *VPS51* | 615738 | Pontocerebellar hypoplasia, type 13, 618606 (3), Autosomal recessive |
| *VPS53* | 615850 | Pontocerebellar hypoplasia, type 2E, 615851 (3), Autosomal recessive |
| *VRK1* | 602168 | Pontocerebellar hypoplasia type 1A, 607596 (3), Autosomal recessive |
| *VSX1* | 605020 | ?Craniofacial anomalies and anterior segment dysgenesis syndrome, 614195 (3); Keratoconus 1, 148300 (3), Autosomal dominant |
| *VSX2* | 142993 | Microphthalmia, isolated 2, 610093 (3); Microphthalmia with coloboma 3, 610092 (3) |
| *VWA1* | 611901 | Neuropathy, hereditary motor, with myopathic features, 619216 (3), Autosomal recessive |
| *VWA3B* | 614884 | ?Spinocerebellar ataxia, autosomal recessive 22, 616948 (3), Autosomal recessive |
| *VWF* | 613160 | von Willebrand disease, type 1, 193400 (3), Autosomal dominant; von Willebrand disease, types 2A, 2B, 2M, and 2N, 613554 (3), Autosomal recessive, Autosomal dominant; von Willebrand disease, type 3, 277480 (3), Autosomal recessive |
| *WAC* | 615049 | Desanto-Shinawi syndrome, 616708 (3), Autosomal dominant |
| *WARS1 (WARS)* | 191050 | Neuronopathy, distal hereditary motor, type IX, 617721 (3), Autosomal dominant |
| *WARS2* | 604733 | Neurodevelopmental disorder, mitochondrial, with abnormal movements and lactic acidosis, with or without seizures, 617710 (3), Autosomal recessive |
| *WAS* | 300392 | Wiskott-Aldrich syndrome, 301000 (3), X-linked recessive; Neutropenia, severe congenital, X-linked, 300299 (3), X-linked recessive; Thrombocytopenia, X-linked, intermittent, 313900 (3), X-linked recessive; Thrombocytopenia, X-linked, 313900 (3), X-linked recessive |
| *WASF1* | 605035 | Neurodevelopmental disorder with absent language and variable seizures, 618707 (3), Autosomal dominant |
| *WASHC4* | 615748 | ?Mental retardation, autosomal recessive 43, 615817 (3), Autosomal recessive |
| *WASHC5* | 610657 | Ritscher-Schinzel syndrome 1, 220210 (3), Autosomal recessive; Spastic paraplegia 8, autosomal dominant, 603563 (3), Autosomal dominant |
| *WBP11* | 618083 | Vertebral, cardiac, tracheoesophageal, renal, and limb defects, 619227 (3), Autosomal dominant |
| *WBP2* | 606962 | Deafness, autosomal recessive 107, 617639 (3), Autosomal recessive |
| *WDFY3* | 617485 | ?Microcephaly 18, primary, autosomal dominant, 617520 (3), Autosomal dominant |
| *WDPCP* | 613580 | ?Bardet-Biedl syndrome 15, 615992 (3), Autosomal recessive; Congenital heart defects, hamartomas of tongue, and polysyndactyly, 217085 (3), Autosomal recessive |
| *WDR1* | 604734 | Periodic fever, immunodeficiency, and thrombocytopenia syndrome, 150550 (3), Autosomal recessive |
| *WDR11* | 606417 | Hypogonadotropic hypogonadism 14 with or without anosmia, 614858 (3), Autosomal dominant |
| *WDR19* | 608151 | Nephronophthisis 13, 614377 (3), Autosomal recessive; Senior-Loken syndrome 8, 616307 (3), Autosomal recessive; ?Short-rib thoracic dysplasia 5 with or without polydactyly, 614376 (3), Autosomal recessive; ?Cranioectodermal dysplasia 4, 614378 (3), Autosomal recessive |
| *WDR26* | 617424 | Skraban-Deardorff syndrome, 617616 (3), Autosomal dominant |
| *WDR35* | 613602 | Short-rib thoracic dysplasia 7 with or without polydactyly, 614091 (3), Autosomal recessive; Cranioectodermal dysplasia 2, 613610 (3), Autosomal recessive |
| *WDR36* | 609669 | Glaucoma 1, open angle, G, 609887 (3) |
| *WDR37* | 618586 | Neurooculocardiogenitourinary syndrome, 618652 (3), Autosomal dominant |
| *WDR4* | 605924 | Galloway-Mowat syndrome 6, 618347 (3), Autosomal recessive; Microcephaly, growth deficiency, seizures, and brain malformations, 618346 (3), Autosomal recessive |
| *WDR45* | 300526 | Neurodegeneration with brain iron accumulation 5, 300894 (3), X-linked dominant |
| *WDR45B* | 609226 | Neurodevelopmental disorder with spastic quadriplegia and brain abnormalities with or without seizures, 617977 (3), Autosomal recessive |
| *WDR62* | 613583 | Microcephaly 2, primary, autosomal recessive, with or without cortical malformations, 604317 (3), Autosomal recessive |
| *WDR72* | 613214 | Amelogenesis imperfecta, type IIA3, 613211 (3), Autosomal recessive |
| *WDR73* | 616144 | Galloway-Mowat syndrome 1, 251300 (3), Autosomal recessive |
| *WDR81* | 614218 | Cerebellar ataxia, mental retardation, and dysequilibrium syndrome 2, 610185 (3), Autosomal recessive; Hydrocephalus, congenital, 3, with brain anomalies, 617967 (3), Autosomal recessive |
| *WEE2* | 614084 | Oocyte maturation defect 5, 617996 (3), Autosomal recessive |
| *WFS1* | 606201 | Deafness, autosomal dominant 6/14/38, 600965 (3), Autosomal dominant; ?Cataract 41, 116400 (3), Autosomal dominant; Wolfram-like syndrome, autosomal dominant, 614296 (3), Autosomal dominant; {Diabetes mellitus, noninsulin-dependent, association with}, 125853 (3), Autosomal dominant; Wolfram syndrome 1, 222300 (3), Autosomal recessive |
| *WHRN* | 607928 | Deafness, autosomal recessive 31, 607084 (3), Autosomal recessive; Usher syndrome, type 2D, 611383 (3), Autosomal recessive |
| *WIPF1* | 602357 | Wiskott-Aldrich syndrome 2, 614493 (3), Autosomal recessive |
| *WIPI2* | 609225 | ?Intellectual developmental disorder with short stature and variable skeletal anomalies, 618453 (3), Autosomal recessive |
| *WNK1* | 605232 | Neuropathy, hereditary sensory and autonomic, type II, 201300 (3), Autosomal recessive; Pseudohypoaldosteronism, type IIC, 614492 (3), Autosomal dominant |
| *WNK4* | 601844 | Pseudohypoaldosteronism, type IIB, 614491 (3), Autosomal dominant |
| *WNT1* | 164820 | {Osteoporosis, early-onset, susceptibility to, autosomal dominant}, 615221 (3); Osteogenesis imperfecta, type XV, 615220 (3), Autosomal recessive |
| *WNT10A* | 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 |
| *WNT10B* | 601906 | Tooth agenesis, selective, 8, 617073 (3), Autosomal dominant; Split-hand/foot malformation 6, 225300 (3), Autosomal recessive |
| *WNT2B* | 601968 | Diarrhea 9, 618168 (3), Autosomal recessive |
| *WNT3* | 165330 | ?Tetra-amelia syndrome 1, 273395 (3), Autosomal recessive |
| *WNT4* | 603490 | ?SERKAL syndrome, 611812 (3), Autosomal recessive; Mullerian aplasia and hyperandrogenism, 158330 (3), Autosomal dominant |
| *WNT5A* | 164975 | Robinow syndrome, autosomal dominant 1, 180700 (3), Autosomal dominant |
| *WNT7A* | 601570 | Fuhrmann syndrome, 228930 (3), Autosomal recessive; Ulna and fibula, absence of, with severe limb deficiency, 276820 (3), Autosomal recessive |
| *WRAP53* | 612661 | Dyskeratosis congenita, autosomal recessive 3, 613988 (3), Autosomal recessive |
| *WRN* | 604611 | Werner syndrome, 277700 (3), Autosomal recessive |
| *WT1* | 607102 | Mesothelioma, somatic, 156240 (3); Meacham syndrome, 608978 (3); Frasier syndrome, 136680 (3), Somatic mutation, Autosomal dominant; Nephrotic syndrome, type 4, 256370 (3), Autosomal dominant; Denys-Drash syndrome, 194080 (3), Somatic mutation, Autosomal dominant; Wilms tumor, type 1, 194070 (3), Somatic mutation, Autosomal dominant |
| *WWC1* | 610533 | [Memory, enhanced, QTL], 615602 (3) |
| *WWOX* | 605131 | Esophageal squamous cell carcinoma, somatic, 133239 (3); Developmental and epileptic encephalopathy 28, 616211 (3), Autosomal recessive; Spinocerebellar ataxia, autosomal recessive 12, 614322 (3), Autosomal recessive |
| *XBP1* | 194355 | {Major affective disorder-7, susceptibility to}, 612371 (3) |
| *XDH* | 607633 | Xanthinuria, type I, 278300 (3), Autosomal recessive |
| *XG* | 300879/314705 | -/[XG blood group system, Xg(a-) phenotype, 314700 (3) |
| *XIAP* | 300079 | Lymphoproliferative syndrome, X-linked, 2, 300635 (3), X-linked recessive |
| *XK* | 314850 | McLeod syndrome with or without chronic granulomatous disease, 300842 (3), X-linked |
| *XPA* | 611153 | Xeroderma pigmentosum, group A, 278700 (3), Autosomal recessive |
| *XPC* | 613208 | Xeroderma pigmentosum, group C, 278720 (3), Autosomal recessive |
| *XPNPEP2* | 300145 | {Angioedema induced by ACE inhibitors, susceptibility to}, 300909 (3) |
| *XPNPEP3* | 613553 | Nephronophthisis-like nephropathy 1, 613159 (3), Autosomal recessive |
| *XPR1* | 605237 | Basal ganglia calcification, idiopathic, 6, 616413 (3), Autosomal dominant |
| *XRCC1* | 194360 | ?Spinocerebellar ataxia, autosomal recessive 26, 617633 (3), Autosomal recessive |
| *XRCC2* | 600375 | Spermatogenic failure, 619145 (3); ?Premature ovarian failure 17, 619146 (3); ?Fanconi anemia, complementation group U, 617247 (3), Autosomal recessive |
| *XRCC3* | 600675 | {Breast cancer, susceptibility to}, 114480 (3), Somatic mutation, Autosomal dominant; {Melanoma, cutaneous malignant, 6}, 613972 (3) |
| *XRCC4* | 194363 | Short stature, microcephaly, and endocrine dysfunction, 616541 (3), Autosomal recessive |
| *XYLT1* | 608124 | Desbuquois dysplasia 2, 615777 (3), Autosomal recessive; {Pseudoxanthoma elasticum, modifier of severity of}, 264800 (3), Autosomal recessive |
| *XYLT2* | 608125 | {Pseudoxanthoma elasticum, modifier of severity of}, 264800 (3), Autosomal recessive; Spondyloocular syndrome, 605822 (3), Autosomal recessive |
| *YAP1* | 606608 | Coloboma, ocular, with or without hearing impairment, cleft lip/palate, and/or mental retardation, 120433 (3), Autosomal dominant |
| *YARS1 (YARS)* | 603623 | Infantile-onset multisystem neurologic, endocrine, and pancreatic disease 2, 619418 (3), Autosomal recessive; Charcot-Marie-Tooth disease, dominant intermediate C, 608323 (3), Autosomal dominant |
| *YARS2* | 610957 | Myopathy, lactic acidosis, and sideroblastic anemia 2, 613561 (3), Autosomal recessive |
| *YEATS2* | 613373 | ?Epilepsy, myoclonic, familial adult, 4, 615127 (3), Autosomal dominant |
| *YIF1B* | 619109 | Kaya-Barakat-Masson syndrome, 619125 (3), Autosomal recessive |
| *YIPF5* | 611483 | Microcephaly, epilepsy, and diabetes syndrome 2, 619278 (3), Autosomal recessive |
| *YME1L1* | 607472 | ?Optic atrophy 11, 617302 (3), Autosomal recessive |
| *YWHAG* | 605356 | Developmental and epileptic encephalopathy 56, 617665 (3), Autosomal dominant |
| *YY1* | 600013 | Gabriele-de Vries syndrome, 617557 (3), Autosomal dominant |
| *YY1AP1* | 607860 | Grange syndrome, 602531 (3), Autosomal recessive |
| *ZAP70* | 176947 | Immunodeficiency 48, 269840 (3), Autosomal recessive; Autoimmune disease, multisystem, infantile-onset, 2, 617006 (3), Autosomal recessive |
| *ZBTB11* | 618181 | Intellectual developmental disorder, autosomal recessive 69, 618383 (3), Autosomal recessive |
| *ZBTB16* | 176797 | Leukemia, acute promyelocytic, PL2F/RARA type (3); Skeletal defects, genital hypoplasia, and mental retardation, 612447 (3), Autosomal recessive |
| *ZBTB18* | 608433 | Mental retardation, autosomal dominant 22, 612337 (3), Autosomal dominant |
| *ZBTB20* | 606025 | Primrose syndrome, 259050 (3), Autosomal dominant |
| *ZBTB24* | 614064 | Immunodeficiency-centromeric instability-facial anomalies syndrome 2, 614069 (3), Autosomal recessive |
| *ZBTB42* | 613915 | ?Lethal congenital contracture syndrome 6, 616248 (3), Autosomal recessive |
| *ZC3H14* | 613279 | Mental retardation, autosomal recessive 56, 617125 (3), Autosomal recessive |
| *ZC4H2* | 300897 | Wieacker-Wolff syndrome, 314580 (3), X-linked recessive; Wieacker-Wolff syndrome, female-restricted, 301041 (3), X-linked dominant |
| *ZCCHC8* | 616381 | ?Pulmonary fibrosis and/or bone marrow failure, telomere-related, 5, 618674 (3), Autosomal dominant |
| *ZDHHC15* | 300576/300577 | -/Mental retardation, X-linked 91, 300577 (4), X-linked dominant |
| *ZDHHC9* | 300646 | Mental retardation, X-linked syndromic, Raymond type, 300799 (3), X-linked |
| *ZEB1* | 189909 | Corneal dystrophy, posterior polymorphous, 3, 609141 (3); Corneal dystrophy, Fuchs endothelial, 6, 613270 (3) |
| *ZEB2* | 605802 | Mowat-Wilson syndrome, 235730 (3), Autosomal dominant |
| *ZFAT* | 610931 | {Autoimmune thyroid disease, susceptibility to, 3}, 608175 (3) |
| *ZFHX2* | 617828 | ?Marsili syndrome, 147430 (3), Autosomal dominant |
| *ZFHX3* | 104155 | Prostate cancer, somatic, 176807 (3) |
| *ZFHX4* | 606940 | ?Ptosis, congenital, 178300 (2), Autosomal dominant |
| *ZFP57* | 612192 | Diabetes mellitus, transient neonatal 1, 601410 (3), Autosomal dominant |
| *ZFPM2* | 603693 | Diaphragmatic hernia 3, 610187 (3); 46XY sex reversal 9, 616067 (3), Autosomal dominant; Tetralogy of Fallot, 187500 (3), Autosomal dominant |
| *ZFYVE26* | 612012 | Spastic paraplegia 15, autosomal recessive, 270700 (3), Autosomal recessive |
| *ZFYVE27* | 610243 | Spastic paraplegia 33, autosomal dominant, 610244 (3), Autosomal dominant |
| *ZIC1* | 600470 | ?Craniosynostosis 6, 616602 (3), Autosomal dominant; Structural brain anomalies with impaired intellectual development and craniosynostosis, 618736 (3), Autosomal dominant |
| *ZIC2* | 603073 | Holoprosencephaly 5, 609637 (3), Autosomal dominant |
| *ZIC3* | 300265 | Congenital heart defects, nonsyndromic, 1, X-linked, 306955 (3), X-linked recessive; Heterotaxy, visceral, 1, X-linked, 306955 (3), X-linked recessive; VACTERL association, X-linked, 314390 (3), X-linked recessive |
| *ZMIZ1* | 607159 | Neurodevelopmental disorder with dysmorphic facies and distal skeletal anomalies, 618659 (3), Autosomal dominant |
| *ZMPSTE24* | 606480 | Mandibuloacral dysplasia with type B lipodystrophy, 608612 (3), Autosomal recessive; Restrictive dermopathy, lethal, 275210 (3), Autosomal recessive |
| *ZMYND10* | 607070 | Ciliary dyskinesia, primary, 22, 615444 (3), Autosomal recessive |
| *ZMYND11* | 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 |
| *ZNF292* | 616213 | Intellectual developmental disorder, autosomal dominant 64, 619188 (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 | Retinitis pigmentosa 72, 616469 (3), Autosomal recessive; ?Exudative vitreoretinopathy 6, 616468 (3), Autosomal dominant |
| *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 |
| *ZPR1* | 603901 | ?Growth restriction, hypoplastic kidneys, alopecia, and distinctive facies, 619321 (3), Autosomal recessive |
| *ZSWIM6* | 615951 | Neurodevelopmental disorder with movement abnormalities, abnormal gait, and autistic features, 617865 (3), Autosomal dominant; Acromelic frontonasal dysostosis, 603671 (3), Autosomal dominant |

|  |  |  |  |  |  |  |  |  |  |  |  |  |  |
| --- | --- | --- | --- | --- | --- | --- | --- | --- | --- | --- | --- | --- | --- |
| |  | | --- | | Gene symbols used are according to the HGNC guidelines. For some genes a previously HGNC-approved symbol is in brackets. | | Each Phenotype is followed by its MIM number, phenotype mapping key and inheritance pattern. | | OMIM release used for OMIM disease identifiers and descriptions: July 26, 2021 | |  | | 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. | |