ECS_mandatory_optional panel

versie	V1 (1124 genen)	Centrum voor Medische Genetica Gent

Gene	OMIM gene ID	Associated phenotype, OMIM phenotype ID, phenotype mapping key and inheritance pattern
AAAS	605378	Achalasia-addisonianism-alacrimia syndrome, 231550 (3), Autosomal recessive
AARS2	612035	Combined oxidative phosphorylation deficiency 8, 614096 (3), Autosomal recessive; Leukoencephalopathy, progressive, with ovarian failure, 615889 (3), Autosomal recessive
ABAT	137150	GABA-transaminase deficiency, 613163 (3), Autosomal recessive
ABCA1	600046	HDL deficiency, familial, 1, 604091 (3); Tangier disease, 205400 (3), Autosomal recessive
ABCA12	607800	Ichthyosis, congenital, autosomal recessive 4A, 601277 (3), Autosomal recessive; Ichthyosis, congenital, autosomal recessive 4B (harlequin), 242500 (3), Autosomal recessive
ABCA3	601615	Surfactant metabolism dysfunction, pulmonary, 3, 610921 (3), Autosomal recessive
ABCA4	601691	Cone-rod dystrophy 3, 604116 (3); Fundus flavimaculatus, 248200 (3), Autosomal recessive; {Macular degeneration, age-related, 2}, 153800 (3), Autosomal dominant; Retinal dystrophy, early-onset severe, 248200 (3), Autosomal recessive; Retinitis pigmentosa 19, 601718 (3), Autosomal recessive; Stargardt disease 1, 248200 (3), Autosomal recessive
ABCB11	603201	Cholestasis, benign recurrent intrahepatic, 2, 605479 (3), Autosomal recessive; Cholestasis, progressive familial intrahepatic 2, 601847 (3), Autosomal recessive
ABCB4	171060	Cholestasis, intrahepatic, of pregnancy, 3, 614972 (3), Autosomal recessive, Autosomal dominant; Cholestasis, progressive familial intrahepatic 3, 602347 (3), Autosomal recessive; Gallbladder disease 1, 600803 (3), Autosomal recessive, Autosomal dominant
ABCC2	601107	Dubin-Johnson syndrome, 237500 (3), Autosomal recessive
ABCC6	603234	Arterial calcification, generalized, of infancy, 2, 614473 (3), Autosomal recessive; Pseudoxanthoma elasticum, 264800 (3), Autosomal recessive; Pseudoxanthoma elasticum, forme fruste, 177850 (3), Autosomal dominant
ABCC8	600509	Diabetes mellitus, noninsulin-dependent, 125853 (3), Autosomal dominant; Diabetes mellitus, permanent neonatal, 606176 (3), Autosomal recessive, Autosomal dominant; Diabetes mellitus, transient neonatal 2, 610374 (3); Hyperinsulinemic hypoglycemia, familial, 1, 256450 (3), Autosomal recessive, Autosomal dominant;

		Hypoglycemia of infancy, leucine-sensitive, 240800 (3), Autosomal dominant
ABCD3	170995	?Bile acid synthesis defect, congenital, 5, 616278 (3), Autosomal recessive
ABCG5	605459	Sitosterolemia, 210250 (3), Autosomal recessive
ABCG8	605460	{Gallbladder disease 4}, 611465 (3); Sitosterolemia, 210250 (3), Autosomal recessive
ABHD12	613599	Polyneuropathy, hearing loss, ataxia, retinitis pigmentosa, and cataract, 612674 (3), Autosomal recessive
ABHD5	604780	Chanarin-Dorfman syndrome, 275630 (3), Autosomal recessive
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
ACAT1	607809	Alpha-methylacetoacetic aciduria, 203750 (3), Autosomal recessive
ACE	106180	[Angiotensin I-converting enzyme, benign serum increase] (3); {Microvascular complications of diabetes 3}, 612624 (3); {Myocardial infarction, susceptibility to} (3); Renal tubular dysgenesis, 267430 (3), Autosomal recessive; {SARS, progression of} (3); {Stroke, hemorrhagic}, 614519 (3)
ACO2	100850	Infantile cerebellar-retinal degeneration, 614559 (3), Autosomal recessive; ?Optic atrophy 9, 616289 (3), Autosomal recessive
ACOX1	609751	Peroxisomal acyl-CoA oxidase deficiency, 264470 (3), Autosomal recessive
ACP5	171640	Spondyloenchondrodysplasia with immune dysregulation, 607944 (3), Autosomal recessive
ACSF3	614245	Combined malonic and methylmalonic aciduria, 614265 (3)
ACTA1	102610	Myopathy, actin, congenital, with cores, 161800 (3), Autosomal recessive, Autosomal dominant; Myopathy, actin, congenital, with excess of thin myofilaments, 161800 (3), Autosomal recessive, Autosomal dominant; Myopathy, congenital, with fiber-type disproportion 1, 255310 (3), Autosomal recessive, Autosomal dominant; ?Myopathy, scapulohumeroperoneal, 616852 (3), Autosomal dominant; Nemaline myopathy 3, autosomal dominant or recessive, 161800 (3), Autosomal recessive, Autosomal dominant
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
ADAMTS10	608990	Weill-Marchesani syndrome 1, recessive, 277600 (3), Autosomal recessive
ADAMTS13	604134	Thrombotic thrombocytopenic purpura, familial, 274150 (3), Autosomal recessive
ADAMTS17	607511	Weill-Marchesani 4 syndrome, recessive, 613195 (3), Autosomal recessive
ADAMTS2	604539	Ehlers-Danlos syndrome, dermatosparaxis type, 225410 (3), Autosomal recessive
ADAMTSL2	612277	Geleophysic dysplasia 1, 231050 (3), Autosomal recessive
ADAR	146920	Aicardi-Goutieres syndrome 6, 615010 (3), Autosomal recessive; Dyschromatosis symmetrica hereditaria, 127400 (3), Autosomal dominant
ADGRG1	604110	Polymicrogyria, bilateral frontoparietal, 606854 (3), Autosomal recessive; Polymicrogyria, bilateral perisylvian, 615752 (3)
ADGRV1	602851	?Febrile seizures, familial, 4, 604352 (3), Autosomal dominant; Usher syndrome, type 2C, 605472 (3), Autosomal recessive; Usher syndrome, type 2C, GPR98/PDZD7 digenic, 605472 (3), Autosomal recessive
ADSL	608222	Adenylosuccinase deficiency, 103050 (3), Autosomal recessive
AFG3L2	604581	Spastic ataxia 5, autosomal recessive, 614487 (3), Autosomal recessive; Spinocerebellar ataxia 28, 610246 (3), Autosomal dominant
AGA	613228	Aspartylglucosaminuria, 208400 (3), Autosomal recessive
AGK	610345	Cataract 38, autosomal recessive, 614691 (3), Autosomal recessive; Sengers syndrome, 212350 (3), Autosomal recessive
AGL	610860	Glycogen storage disease IIIa, 232400 (3), Autosomal recessive; Glycogen storage disease IIIb, 232400 (3), Autosomal recessive
AGPAT2	603100	Lipodystrophy, congenital generalized, type 1, 608594 (3), Autosomal recessive
AGPS	603051	Rhizomelic chondrodysplasia punctata, type 3, 600121 (3), Autosomal recessive
AGT	106150	{Hypertension, essential, susceptibility to}, 145500 (3), Multifactorial; {Preeclampsia, susceptibility to} (3); Renal tubular dysgenesis, 267430 (3), Autosomal recessive
AGTR1	106165	{Hypertension, essential}, 145500 (3), Multifactorial; Renal tubular dysgenesis, 267430 (3), Autosomal recessive
AGXT	604285	Hyperoxaluria, primary, type 1, 259900 (3), Autosomal recessive
AHCY	180960	Hypermethioninemia with deficiency of S-adenosylhomocysteine hydrolase, 613752 (3), Autosomal recessive
AHI1	608894	Joubert syndrome 3, 608629 (3), Autosomal recessive
AICDA	605257	Immunodeficiency with hyper-IgM, type 2, 605258 (3), Autosomal recessive

AIMP1	603605	Leukodystrophy, hypomyelinating, 3, 260600 (3), Autosomal recessive
AIPL1	604392	Cone-rod dystrophy, 604393 (3), Autosomal recessive; Leber congenital amaurosis 4, 604393 (3), Autosomal recessive; Retinitis pigmentosa, juvenile, 604393 (3), Autosomal recessive
AIRE	607358	Autoimmune polyendocrinopathy syndrome , type I, with or without reversible metaphyseal dysplasia, 240300 (3), Autosomal recessive, Autosomal dominant
AK1	103000	Hemolytic anemia due to adenylate kinase deficiency, 612631 (3), Autosomal recessive
AK2	103020	Reticular dysgenesis, 267500 (3), Autosomal recessive
AKR1C2	600450	46XY sex reversal 8, 614279 (3), Autosomal recessive
ALAD	125270	{Lead poisoning, susceptibility to}, 612740 (3), Autosomal recessive; Porphyria, acute hepatic, 612740 (3), Autosomal recessive
ALDH18A1	138250	Cutis laxa, autosomal dominant 3, 616603 (3), Autosomal dominant; Cutis laxa, autosomal recessive, type IIIA, 219150 (3), Autosomal recessive; Spastic paraplegia 9A, autosomal dominant, 601162 (3), Autosomal dominant; Spastic paraplegia 9B, autosomal recessive, 616586 (3), Autosomal recessive
ALDH1A3	600463	Microphthalmia, isolated 8, 615113 (3), Autosomal recessive
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
ALDH7A1	107323	Epilepsy, pyridoxine-dependent, 266100 (3), Autosomal recessive
ALDOB	612724	Fructose intolerance, hereditary, 229600 (3), Autosomal recessive
ALG1	605907	Congenital disorder of glycosylation, type Ik, 608540 (3), Autosomal recessive
ALG11	613666	Congenital disorder of glycosylation, type Ip, 613661 (3), Autosomal recessive
ALG12	607144	Congenital disorder of glycosylation, type Ig, 607143 (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	Congenital disorder of glycosylation, type Il, 608776 (3), Autosomal recessive; Gillessen-Kaesbach-Nishimura syndrome, 263210 (3), Autosomal recessive
ALMS1	606844	Alstrom syndrome, 203800 (3), Autosomal recessive

ALOX12B	603741	Ichthyosis, congenital, autosomal recessive 2, 242100 (3), Autosomal recessive
ALPL	171760	Hypophosphatasia, adult, 146300 (3), Autosomal recessive, Autosomal dominant; Hypophosphatasia, childhood, 241510 (3), Autosomal recessive; Hypophosphatasia, infantile, 241500 (3), Autosomal recessive; Odontohypophosphatasia, 146300 (3), Autosomal recessive, Autosomal dominant
ALS2	606352	Amyotrophic lateral sclerosis 2, juvenile, 205100 (3), Autosomal recessive; Primary lateral sclerosis, juvenile, 606353 (3), Autosomal recessive; Spastic paralysis, infantile onset ascending, 607225 (3), Autosomal recessive
ALX3	606014	Frontonasal dysplasia 1, 136760 (3), Autosomal recessive
ALX4	605420	{Craniosynostosis 5, susceptibility to}, 615529 (3), Autosomal dominant; Frontonasal dysplasia 2, 613451 (3), Autosomal recessive; Parietal foramina 2, 609597 (3), Autosomal dominant
AMACR	604489	Alpha-methylacyl-CoA racemase deficiency, 614307 (3), Autosomal recessive; Bile acid synthesis defect, congenital, 4, 214950 (3), Autosomal recessive
AMH	600957	Persistent Mullerian duct syndrome, type I, 261550 (3), Autosomal recessive
AMHR2	600956	Persistent Mullerian duct syndrome, type II, 261550 (3), Autosomal recessive
AMN	605799	Megaloblastic anemia-1, Norwegian type, 261100 (3), Autosomal recessive
AMPD1	102770	Myopathy due to myoadenylate deaminase deficiency, 615511 (3), Autosomal recessive
AMT	238310	Glycine encephalopathy, 605899 (3), Autosomal recessive
ANO10	613726	Spinocerebellar ataxia, autosomal recessive 10, 613728 (3), Autosomal recessive
ANO5	608662	Gnathodiaphyseal dysplasia, 166260 (3), Autosomal dominant; Miyoshi muscular dystrophy 3, 613319 (3), Autosomal recessive; Muscular dystrophy, limb-girdle, autosomal recessive 12, 611307 (3), Autosomal recessive
ANTXR2	608041	Hyaline fibromatosis syndrome, 228600 (3), Autosomal recessive
AP3B1	603401	Hermansky-Pudlak syndrome 2, 608233 (3), Autosomal recessive
AP4B1	607245	Spastic paraplegia 47, autosomal recessive, 614066 (3), Autosomal recessive
AP4E1	607244	Spastic paraplegia 51, autosomal recessive, 613744 (3), Autosomal recessive; Stuttering, familial persistent, 1, 184450 (3), Autosomal dominant
APOB	107730	Hypercholesterolemia, familial, 2, 144010 (3), Autosomal dominant; Hypobetalipoproteinemia, 615558 (3), Autosomal recessive
APOC2	608083	Hyperlipoproteinemia, type Ib, 207750 (3), Autosomal recessive
APRT	102600	Adenine phosphoribosyltransferase deficiency, 614723 (3), Autosomal recessive

APTX	606350	Ataxia, early-onset, with oculomotor apraxia and hypoalbuminemia, 208920 (3), Autosomal recessive
AQP2	107777	Diabetes insipidus, nephrogenic, 125800 (3), Autosomal recessive, Autosomal dominant
ARFGEF2	605371	Periventricular heterotopia with microcephaly, 608097 (3), Autosomal recessive
ARG1	608313	Argininemia, 207800 (3), Autosomal recessive
ARL13B	608922	Joubert syndrome 8, 612291 (3), Autosomal recessive
ARL6	608845	{Bardet-Biedl syndrome 1, modifier of}, 209900 (3), Autosomal recessive, Digenic recessive; Bardet-Biedl syndrome 3, 600151 (3), Autosomal recessive; ?Retinitis pigmentosa 55, 613575 (3)
ARSA	607574	Metachromatic leukodystrophy, 250100 (3), Autosomal recessive
ARSB	611542	Mucopolysaccharidosis type VI (Maroteaux-Lamy), 253200 (3), Autosomal recessive
ASAH1	613468	Farber lipogranulomatosis, 228000 (3), Autosomal recessive; Spinal muscular atrophy with progressive myoclonic epilepsy, 159950 (3), Autosomal recessive
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
ASPM	605481	Microcephaly 5, primary, autosomal recessive, 608716 (3), Autosomal recessive
ASS1	603470	Citrullinemia, 215700 (3), Autosomal recessive
ATF6	605537	Achromatopsia 7, 616517 (3), Autosomal recessive
ATIC	601731	AICA-ribosiduria due to ATIC deficiency, 608688 (3), Autosomal recessive
ATM	607585	Ataxia-telangiectasia, 208900 (3), Autosomal recessive; {Breast cancer, susceptibility to}, 114480 (3), Autosomal dominant, Somatic mutation; Lymphoma, B-cell non-Hodgkin, somatic (3); Lymphoma, mantle cell, somatic (3); T-cell prolymphocytic leukemia, somatic (3)
ATP13A2	610513	Kufor-Rakeb syndrome, 606693 (3), Autosomal recessive; Spastic paraplegia 78, autosomal recessive, 617225 (3), Autosomal recessive
ATP6V0A2	611716	Cutis laxa, autosomal recessive, type IIA, 219200 (3), Autosomal recessive; Wrinkly skin syndrome, 278250 (3), Autosomal recessive
ATP6V0A4	605239	Renal tubular acidosis, distal, autosomal recessive, 602722 (3)
ATP6V1B1	192132	Renal tubular acidosis with deafness, 267300 (3), Autosomal recessive
ATP7B	606882	Wilson disease, 277900 (3), Autosomal recessive
ATP8A2	605870	?Cerebellar ataxia, mental retardation, and dysequilibrium syndrome 4, 615268 (3), Autosomal recessive
ATP8B1	602397	Cholestasis, benign recurrent intrahepatic, 243300 (3), Autosomal recessive; Cholestasis, intrahepatic, of pregnancy, 1, 147480 (3), Autosomal dominant; Cholestasis, progressive familial intrahepatic 1, 211600 (3), Autosomal recessive

ATR	601215	?Cutaneous telangiectasia and cancer syndrome, familial, 614564 (3), Autosomal dominant; Seckel syndrome 1, 210600 (3), Autosomal recessive
AUH	600529	3-methylglutaconic aciduria, type I, 250950 (3), Autosomal recessive
B3GALNT2	610194	Muscular dystrophy-dystroglycanopathy (congenital with brain and eye anomalies, type A, 11, 615181 (3), Autosomal recessive
B3GALT6	615291	Ehlers-Danlos syndrome, spondylodysplastic type, 2, 615349 (3), Autosomal recessive; Spondyloepimetaphyseal dysplasia with joint laxity, type 1, with or without fractures, 271640 (3), Autosomal recessive
B3GAT3	606374	Multiple joint dislocations, short stature, craniofacial dysmorphism, with or without congenital heart defects, 245600 (3), Autosomal recessive
B3GLCT	610308	Peters-plus syndrome, 261540 (3), Autosomal recessive
B4GALT1	137060	Congenital disorder of glycosylation, type IId, 607091 (3), Autosomal recessive
B4GAT1	605517	Muscular dystrophy-dystroglycanopathy (congenital with brain and eye anomalies), type A, 13, 615287 (3), Autosomal recessive
B9D1	614144	Joubert syndrome 27, 617120 (3), Autosomal recessive; ?Meckel syndrome 9, 614209 (3), Autosomal recessive
B9D2	611951	Joubert syndrome 34, 614175 (3), Autosomal recessive; ?Meckel syndrome 10, 614175 (3), Autosomal recessive
BBIP1	613605	?Bardet-Biedl syndrome 18, 615995 (3), Autosomal recessive
BBS1	209901	Bardet-Biedl syndrome 1, 209900 (3), Autosomal recessive, Digenic recessive
BBS10	610148	Bardet-Biedl syndrome 10, 615987 (3), Autosomal recessive
BBS12	610683	Bardet-Biedl syndrome 12, 615989 (3), Autosomal recessive
BBS2	606151	Bardet-Biedl syndrome 2, 615981 (3), Autosomal recessive; Retinitis pigmentosa 74, 616562 (3), Autosomal recessive
BBS4	600374	Bardet-Biedl syndrome 4, 615982 (3), Autosomal recessive
BBS5	603650	Bardet-Biedl syndrome 5, 615983 (3), Autosomal recessive
BBS7	607590	Bardet-Biedl syndrome 7, 615984 (3), Autosomal recessive
BBS9	607968	Bardet-Biedl syndrome 9, 615986 (3), Autosomal recessive
BCKDHA	608348	Maple syrup urine disease, type Ia, 248600 (3), Autosomal recessive
BCKDHB	248611	Maple syrup urine disease, type Ib, 248600 (3), Autosomal recessive
BCS1L	603647	Bjornstad syndrome, 262000 (3), Autosomal recessive; GRACILE syndrome, 603358 (3), Autosomal recessive; Leigh syndrome, 256000 (3), Autosomal recessive, Mitochondrial; Mitochondrial complex III deficiency, nuclear type 1, 124000 (3), Autosomal recessive
BEST1	607854	Bestrophinopathy, autosomal recessive, 611809 (3); Macular dystrophy, vitelliform, 2, 153700 (3), Autosomal dominant; Microcornea, rod-cone dystrophy, cataract, and posterior staphyloma, 193220 (3), Autosomal dominant; Retinitis pigmentosa, concentric, 613194 (3); Retinitis pigmentosa-50, 613194 (3); Vitreoretinochoroidopathy, 193220 (3), Autosomal dominant
BIN1	601248	Centronuclear myopathy 2, 255200 (3), Autosomal recessive

BLM	604610	Bloom syndrome, 210900 (3), Autosomal recessive
BLOC1S6	604310	?Hermansky-pudlak syndrome 9, 614171 (3), Autosomal recessive
BMP1	112264	Osteogenesis imperfecta, type XIII, 614856 (3), Autosomal recessive
BMP2	112261	Brachydactyly, type A2, 112600 (3), Autosomal dominant; {HFE hemochromatosis, modifier of}, 235200 (3), Autosomal recessive; Short stature, facial dysmorphism, and skeletal anomalies with or without cardiac anomalies, 617877 (3), Autosomal dominant
BMPER	608699	Diaphanospondylodysostosis, 608022 (3), Autosomal recessive
BMPR1B	603248	Acromesomelic dysplasia, Demirhan type, 609441 (3), Autosomal recessive; Brachydactyly, type A1, D, 616849 (3), Autosomal dominant; Brachydactyly, type A2, 112600 (3), Autosomal dominant
BRCA2	600185	{Breast cancer, male, susceptibility to}, 114480 (3), Autosomal dominant, Somatic mutation; {Breast-ovarian cancer, familial, 2}, 612555 (3), Autosomal dominant; Fanconi anemia, complementation group D1, 605724 (3), Autosomal recessive; {Glioblastoma 3}, 613029 (3), Autosomal recessive; {Medulloblastoma}, 155255 (3), Autosomal recessive, Autosomal dominant; {Pancreatic cancer 2}, 613347 (3); {Prostate cancer}, 176807 (3), Autosomal dominant, Somatic mutation; Wilms tumor, 194070 (3), Autosomal dominant, Somatic mutation
BRIP1	605882	{Breast cancer, early-onset, susceptibility to}, 114480 (3), Autosomal dominant, Somatic mutation; Fanconi anemia, complementation group J, 609054 (3)
BSCL2	606158	Encephalopathy, progressive, with or without lipodystrophy, 615924 (3), Autosomal recessive; Lipodystrophy, congenital generalized, type 2, 269700 (3), Autosomal recessive; Neuropathy, distal hereditary motor, type VA, 600794 (3), Autosomal dominant; Silver spastic paraplegia syndrome, 270685 (3), Autosomal dominant
BSND	606412	Bartter syndrome, type 4a, 602522 (3), Autosomal recessive; Sensorineural deafness with mild renal dysfunction, 602522 (3), Autosomal recessive
BTD	609019	Biotinidase deficiency, 253260 (3), Autosomal recessive
BUB1B	602860	Colorectal cancer, somatic, 114500 (3); Mosaic variegated aneuploidy syndrome 1, 257300 (3), Autosomal recessive; [Premature chromatid separation trait], 176430 (3), Autosomal dominant
C12orf57	615140	Temtamy syndrome, 218340 (3), Autosomal recessive
C12orf65	613541	Combined oxidative phosphorylation deficiency 7, 613559 (3), Autosomal recessive; Spastic paraplegia 55, autosomal recessive, 615035 (3), Autosomal recessive
C19orf12	614297	Neurodegeneration with brain iron accumulation 4, 614298 (3), Autosomal recessive, Autosomal dominant; ?Spastic paraplegia 43, autosomal recessive, 615043 (3), Autosomal recessive
C1QTNF5	608752	Retinal degeneration, late-onset, autosomal dominant, 605670 (3), Autosomal dominant

C8B	120960	C8 deficiency, type II, 613789 (3), Autosomal recessive
CA2	611492	Osteopetrosis, autosomal recessive 3, with renal tubular acidosis, 259730 (3), Autosomal recessive
CANT1	613165	Desbuquois dysplasia 1, 251450 (3), Autosomal recessive; Epiphyseal dysplasia, multiple, 7, 617719 (3), Autosomal recessive
CAPN3	114240	Muscular dystrophy, limb-girdle, autosomal dominant 4, 618129 (3), Autosomal dominant; Muscular dystrophy, limb-girdle, autosomal recessive 1, 253600 (3), Autosomal recessive
CASQ2	114251	Ventricular tachycardia, catecholaminergic polymorphic, 2, 611938 (3), Autosomal recessive
CASR	601199	{Epilepsy idiopathic generalized, susceptibility to, 8}, 612899 (3); Hyperparathyroidism, neonatal, 239200 (3), Autosomal recessive, Autosomal dominant; Hypocalcemia, autosomal dominant, 601198 (3), Autosomal dominant; Hypocalcemia, autosomal dominant, with Bartter syndrome, 601198 (3), Autosomal dominant; Hypocalciuric hypercalcemia, type I, 145980 (3), Autosomal dominant
CAV3	601253	Cardiomyopathy, familial hypertrophic, 192600 (3), Autosomal dominant; Creatine phosphokinase, elevated serum, 123320 (3), Autosomal dominant; Long QT syndrome 9, 611818 (3), Autosomal dominant; Myopathy, distal, Tateyama type, 614321 (3), Autosomal dominant; Rippling muscle disease 2, 606072 (3), Autosomal dominant
CBS	613381	Homocystinuria, B6-responsive and nonresponsive types, 236200 (3), Autosomal recessive; Thrombosis, hyperhomocysteinemic, 236200 (3), Autosomal recessive
CC2D2A	612013	COACH syndrome, 216360 (3), Autosomal recessive; Joubert syndrome 9, 612285 (3), Autosomal recessive; Meckel syndrome 6, 612284 (3), Autosomal recessive
CCBE1	612753	Hennekam lymphangiectasia-lymphedema syndrome 1, 235510 (3), Autosomal recessive
CCDC28B	610162	{Bardet-Biedl syndrome 1, modifier of}, 209900 (3), Autosomal recessive, Digenic recessive
CCDC39	613798	Ciliary dyskinesia, primary, 14, 613807 (3)
CCDC40	613799	Ciliary dyskinesia, primary, 15, 613808 (3)
CCN6 (WISP3)	603400	Arthropathy, progressive pseudorheumatoid, of childhood, 208230 (3), Autosomal recessive; Spondyloepiphyseal dysplasia tarda with progressive arthropathy, 208230 (3), Autosomal recessive
CD19	107265	Immunodeficiency, common variable, 3, 613493 (3), Autosomal recessive
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
CD96	606037	C syndrome, 211750 (3), Autosomal dominant

CDAN1	607465	Dyserythropoietic anemia, congenital, type Ia, 224120 (3), Autosomal recessive
CDC45	603465	Meier-Gorlin syndrome 7, 617063 (3), Autosomal recessive
CDC6	602627	?Meier-Gorlin syndrome 5, 613805 (3), Autosomal recessive
CDH23	605516	Deafness, autosomal recessive 12, 601386 (3), Autosomal recessive; {Pituitary adenoma 5, multiple types}, 617540 (3), Autosomal dominant; Usher syndrome, type 1D, 601067 (3), Autosomal recessive, Digenic recessive; Usher syndrome, type 1D/F digenic, 601067 (3), Autosomal recessive, Digenic recessive
CDK5RAP2	608201	Microcephaly 3, primary, autosomal recessive, 604804 (3), Autosomal recessive
CDT1	605525	Meier-Gorlin syndrome 4, 613804 (3), Autosomal recessive
CENPJ	609279	Microcephaly 6, primary, autosomal recessive, 608393 (3), Autosomal recessive; ?Seckel syndrome 4, 613676 (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
CEP290	610142	?Bardet-Biedl syndrome 14, 615991 (3), Autosomal recessive; Joubert syndrome 5, 610188 (3), Autosomal recessive; Leber congenital amaurosis 10, 611755 (3); Meckel syndrome 4, 611134 (3), Autosomal recessive; Senior-Loken syndrome 6, 610189 (3), Autosomal recessive
CEP41	610523	Joubert syndrome 15, 614464 (3), Autosomal recessive
CEP57	607951	Mosaic variegated aneuploidy syndrome 2, 614114 (3), Autosomal recessive
CERKL	608381	Retinitis pigmentosa 26, 608380 (3)
CFTR	602421	{Bronchiectasis with or without elevated sweat chloride 1, modifier of}, 211400 (3), Autosomal dominant; Congenital bilateral absence of vas deferens, 277180 (3), Autosomal recessive; Cystic fibrosis, 219700 (3), Autosomal recessive; {Hypertrypsinemia, neonatal} (3); {Pancreatitis, hereditary}, 167800 (3), Autosomal dominant; Sweat chloride elevation without CF (3)
CHAT	118490	Myasthenic syndrome, congenital, 6, presynaptic, 254210 (3), Autosomal recessive
CHMP1A	164010	Pontocerebellar hypoplasia, type 8, 614961 (3), Autosomal recessive
CHRNA1	100690	Multiple pterygium syndrome, lethal type, 253290 (3), Autosomal recessive; Myasthenic syndrome, congenital, 1A, slow-channel, 601462 (3), Autosomal dominant; Myasthenic syndrome, congenital, 1B, fast-channel, 608930 (3), Autosomal recessive, Autosomal dominant
CHRNB1	100710	Myasthenic syndrome, congenital, 2A, slow-channel, 616313 (3), Autosomal dominant; ?Myasthenic syndrome, congenital, 2C, associated with acetylcholine receptor deficiency, 616314 (3), Autosomal recessive

CHRND	100720	Multiple pterygium syndrome, lethal type, 253290 (3), Autosomal recessive; ?Myasthenic syndrome, congenital, 3A, slow-channel, 616321 (3), Autosomal dominant; Myasthenic syndrome, congenital, 3B, fast-channel, 616322 (3), Autosomal recessive; ?Myasthenic syndrome, congenital, 3C, associated with acetylcholine receptor deficiency, 616323 (3), Autosomal recessive
CHRNE	100725	Myasthenic syndrome, congenital, 4A, slow-channel, 605809 (3), Autosomal recessive, Autosomal dominant; Myasthenic syndrome, congenital, 4B, fast-channel, 616324 (3), Autosomal recessive; Myasthenic syndrome, congenital, 4C, associated with acetylcholine receptor deficiency, 608931 (3), Autosomal recessive
CHRNG	100730	Escobar syndrome, 265000 (3), Autosomal recessive; Multiple pterygium syndrome, lethal type, 253290 (3), Autosomal recessive
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
CHSY1	608183	Temtamy preaxial brachydactyly syndrome, 605282 (3), Autosomal recessive
CIITA	600005	Bare lymphocyte syndrome, type II, complementation group A, 209920 (3), Autosomal recessive; {Rheumatoid arthritis, susceptibility to}, 180300 (3)
CILK1 (ICK)	612325	Endocrine-cerebroosteodysplasia, 612651 (3), Autosomal recessive; {Epilepsy, juvenile myoclonic, susceptibility to, 10}, 617924 (3), Autosomal dominant
CKAP2L	616174	Filippi syndrome, 272440 (3), Autosomal recessive
CLCN1	118425	Myotonia congenita, dominant, 160800 (3), Autosomal dominant; Myotonia congenita, recessive, 255700 (3), Autosomal recessive; Myotonia levior, recessive (3)
CLCN2	600570	{Epilepsy, idiopathic generalized, susceptibility to, 11}, 607628 (3), Autosomal dominant; {Epilepsy, juvenile absence, susceptibility to, 2}, 607628 (3), Autosomal dominant; {Epilepsy, juvenile myoclonic, susceptibility to, 8}, 607628 (3), Autosomal dominant; Hyperaldosteronism, familial, type II, 605635 (3), Autosomal dominant; Leukoencephalopathy with ataxia, 615651 (3), Autosomal recessive
CLCN7	602727	Hypopigmentation, organomegaly, and delayed myelination and development, 618541 (3), Autosomal dominant; Osteopetrosis, autosomal dominant 2, 166600 (3), Autosomal dominant; Osteopetrosis, autosomal recessive 4, 611490 (3), Autosomal recessive
CLDN1	603718	Ichthyosis, leukocyte vacuoles, alopecia, and sclerosing cholangitis, 607626 (3), Autosomal recessive
CLDN14	605608	Deafness, autosomal recessive 29, 614035 (3), Autosomal recessive
CLDN16	603959	Hypomagnesemia 3, renal, 248250 (3), Autosomal recessive

CLDN19	610036	Hypomagnesemia 5, renal, with ocular involvement, 248190 (3), Autosomal recessive
CLN3	607042	Ceroid lipofuscinosis, neuronal, 3, 204200 (3), Autosomal recessive
CLN5	608102	Ceroid lipofuscinosis, neuronal, 5, 256731 (3), Autosomal recessive
CLN6	606725	Ceroid lipofuscinosis, neuronal, 6, 601780 (3), Autosomal recessive; Ceroid lipofuscinosis, neuronal, Kufs type, adult onset, 204300 (3), Autosomal recessive
CLN8	607837	Ceroid lipofuscinosis, neuronal, 8, 600143 (3), Autosomal recessive; Ceroid lipofuscinosis, neuronal, 8, Northern epilepsy variant, 610003 (3), Autosomal recessive
CLRN1	606397	Retinitis pigmentosa 61, 614180 (3); Usher syndrome, type 3A, 276902 (3), Autosomal recessive
CNGA1	123825	Retinitis pigmentosa 49, 613756 (3)
CNGA3	600053	Achromatopsia 2, 216900 (3), Autosomal recessive
CNGB1	600724	Retinitis pigmentosa 45, 613767 (3), Autosomal recessive
CNGB3	605080	Achromatopsia 3, 262300 (3), Autosomal recessive; Macular degeneration, juvenile, 248200 (3), Autosomal recessive
CNNM4	607805	Jalili syndrome, 217080 (3), Autosomal recessive
CNTN1	600016	?Myopathy, congenital, Compton-North, 612540 (3), Autosomal recessive
CNTNAP2	604569	{Autism susceptibility 15}, 612100 (3); Cortical dysplasia-focal epilepsy syndrome, 610042 (3), Autosomal recessive; Pitt-Hopkins like syndrome 1, 610042 (3), Autosomal recessive
COG1	606973	Congenital disorder of glycosylation, type IIg, 611209 (3), Autosomal recessive
COG4	606976	Congenital disorder of glycosylation, type IIj, 613489 (3), Autosomal recessive; Saul-Wilson syndrome, 618150 (3), Autosomal dominant
COG7	606978	Congenital disorder of glycosylation, type IIe, 608779 (3), Autosomal recessive
COG8	606979	Congenital disorder of glycosylation, type IIh, 611182 (3)
COL11A1	120280	?Deafness, autosomal dominant 37, 618533 (3); Fibrochondrogenesis 1, 228520 (3), Autosomal recessive; {Lumbar disc herniation, susceptibility to}, 603932 (3); Marshall syndrome, 154780 (3), Autosomal dominant; Stickler syndrome, type II, 604841 (3), Autosomal dominant
COL11A2	120290	Deafness, autosomal dominant 13, 601868 (3), Autosomal dominant; Deafness, autosomal recessive 53, 609706 (3), Autosomal recessive; Fibrochondrogenesis 2, 614524 (3), Autosomal recessive, Autosomal dominant; Otospondylomegaepiphyseal dysplasia, autosomal dominant, 184840 (3), Autosomal dominant; Otospondylomegaepiphyseal dysplasia, autosomal recessive, 215150 (3), Autosomal recessive

COL17A1	113811	Epidermolysis bullosa, junctional, localisata variant, 226650 (3), Autosomal recessive; Epidermolysis bullosa, junctional, non-Herlitz type, 226650 (3), Autosomal recessive; Epithelial recurrent erosion dystrophy, 122400 (3), Autosomal dominant
COL18A1	120328	Knobloch syndrome, type 1, 267750 (3), Autosomal recessive
COL1A2	120160	Ehlers-Danlos syndrome, arthrochalasia type, 2, 617821 (3), Autosomal dominant; Ehlers-Danlos syndrome, cardiac valvular type, 225320 (3), Autosomal recessive; Osteogenesis imperfecta, type II, 166210 (3), Autosomal dominant; Osteogenesis imperfecta, type III, 259420 (3), Autosomal dominant; Osteogenesis imperfecta, type IV, 166220 (3), Autosomal dominant; {Osteoporosis, postmenopausal}, 166710 (3), Autosomal dominant
COL27A1	608461	Steel syndrome, 615155 (3), Autosomal recessive
COL2A1	120140	Achondrogenesis, type II or hypochondrogenesis, 200610 (3), Autosomal dominant; Avascular necrosis of the femoral head, 608805 (3), Autosomal dominant; Czech dysplasia, 609162 (3), Autosomal dominant; Epiphyseal dysplasia, multiple, with myopia and deafness, 132450 (3), Autosomal dominant; Kniest dysplasia, 156550 (3), Autosomal dominant; Legg-Calve-Perthes disease, 150600 (3), Autosomal dominant; Osteoarthritis with mild chondrodysplasia, 604864 (3), Autosomal dominant; Platyspondylic skeletal dysplasia, Torrance type, 151210 (3), Autosomal dominant; SED congenita, 183900 (3), Autosomal dominant; SMED Strudwick type, 184250 (3), Autosomal dominant; Spondyloepiphyseal dysplasia, Stanescu type, 616583 (3), Autosomal dominant; Spondyloperipheral dysplasia, 271700 (3), Autosomal dominant; Stickler sydrome, type I, nonsyndromic ocular, 609508 (3), Autosomal dominant; Stickler syndrome, type I, 108300 (3), Autosomal dominant; Vitreoretinopathy with phalangeal epiphyseal dysplasia (3)
COL4A3	120070	Alport syndrome 2, autosomal recessive, 203780 (3), Autosomal recessive; Alport syndrome 3, autosomal dominant, 104200 (3), Autosomal dominant; Hematuria, benign familial, 141200 (3), Autosomal dominant
COL4A4	120131	Alport syndrome 2, autosomal recessive, 203780 (3), Autosomal recessive; Hematuria, familial benign, 141200 (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	Bethlem myopathy 1, 158810 (3), Autosomal recessive, Autosomal dominant; Dystonia 27, 616411 (3), Autosomal recessive; Ullrich congenital muscular dystrophy 1, 254090 (3), Autosomal recessive, Autosomal dominant
COL7A1	120120	EBD inversa, 226600 (3), Autosomal recessive; EBD, Bart type, 132000 (3), Autosomal dominant; EBD, localisata variant (3); Epidermolysis bullosa dystrophica, AD, 131750 (3), Autosomal dominant; Epidermolysis bullosa dystrophica, AR, 226600 (3), Autosomal recessive; Epidermolysis bullosa pruriginosa, 604129 (3), Autosomal recessive, Autosomal dominant; Epidermolysis bullosa, pretibial, 131850 (3), Autosomal recessive, Autosomal dominant; Toenail dystrophy, isolated, 607523 (3), Autosomal dominant; Transient bullous of the newborn, 131705 (3), Autosomal recessive, Autosomal dominant
COL9A2	120260	Epiphyseal dysplasia, multiple, 2, 600204 (3), Autosomal dominant; ?Stickler syndrome, type V, 614284 (3), Autosomal recessive
COLEC11	612502	3MC syndrome 2, 265050 (3), Autosomal recessive
COLQ	603033	Myasthenic syndrome, congenital, 5, 603034 (3), Autosomal recessive
COQ2	609825	Coenzyme Q10 deficiency, primary, 1, 607426 (3), Autosomal recessive; {Multiple system atrophy, susceptibility to}, 146500 (3), Autosomal recessive, Autosomal dominant
COQ4	612898	Coenzyme Q10 deficiency, primary, 7, 616276 (3), Autosomal recessive
COQ8A	606980	Coenzyme Q10 deficiency, primary, 4, 612016 (3), Autosomal recessive
COQ9	612837	Coenzyme Q10 deficiency, primary, 5, 614654 (3), Autosomal recessive
COX10	602125	Leigh syndrome due to mitochondrial COX4 deficiency, 256000 (3), Autosomal recessive, Mitochondrial; Mitochondrial complex IV deficiency, 220110 (3), Autosomal recessive, Mitochondrial
COX15	603646	Cardioencephalomyopathy, fatal infantile, due to cytochrome c oxidase deficiency 2, 615119 (3), Autosomal recessive; Leigh syndrome due to cytochrome c oxidase deficiency, 256000 (3), Autosomal recessive, Mitochondrial
COX6B1	124089	Mitochondrial complex IV deficiency, 220110 (3), Autosomal recessive, Mitochondrial
CP	117700	Cerebellar ataxia, 604290 (3), Autosomal recessive; Hemosiderosis, systemic, due to aceruloplasminemia, 604290 (3), Autosomal recessive; [Hypoceruloplasminemia, hereditary], 604290 (3), Autosomal recessive
CPLANE1	614571	Joubert syndrome 17, 614615 (3), Autosomal recessive; Orofaciodigital syndrome VI, 277170 (3), Autosomal recessive
CPS1	608307	Carbamoylphosphate synthetase I deficiency, 237300 (3), Autosomal recessive; {Pulmonary hypertension, neonatal, susceptibility to}, 615371 (3)

CPT1A	600528	CPT deficiency, hepatic, type IA, 255120 (3), Autosomal recessive
CPT2	600650	CPT II deficiency, infantile, 600649 (3), Autosomal recessive; CPT II deficiency, lethal neonatal, 608836 (3), Autosomal recessive; CPT II deficiency, myopathic, stress-induced, 255110 (3), Autosomal recessive, Autosomal dominant; {Encephalopathy, acute, infection-induced, 4, susceptibility to}, 614212 (3), Autosomal recessive, Autosomal dominant
CRB1	604210	Leber congenital amaurosis 8, 613835 (3), Autosomal recessive; Pigmented paravenous chorioretinal atrophy, 172870 (3), Autosomal dominant; Retinitis pigmentosa-12, 600105 (3), Autosomal recessive
CRLF1	604237	Cold-induced sweating syndrome 1, 272430 (3), Autosomal recessive
CRPPA (ISPD)	614631	Muscular dystrophy-dystroglycanopathy (congenital with brain and eye anomalies), type A, 7, 614643 (3), Autosomal recessive; Muscular dystrophy-dystroglycanopathy (limb-girdle), type C, 7, 616052 (3), Autosomal recessive
CRTAP	605497	Osteogenesis imperfecta, type VII, 610682 (3), Autosomal recessive
CRYAB	123590	Cardiomyopathy, dilated, 1II, 615184 (3), Autosomal dominant; Cataract 16, multiple types, 613763 (3), Autosomal recessive, Autosomal dominant; Myopathy, myofibrillar, 2, 608810 (3), Autosomal dominant; Myopathy, myofibrillar, fatal infantile hypertonic, alpha-B crystallin-related, 613869 (3), Autosomal recessive
CSPP1	611654	Joubert syndrome 21, 615636 (3), Autosomal recessive
CSTB	601145	Epilepsy, progressive myoclonic 1A (Unverricht and Lundborg), 254800 (3), Autosomal recessive
CTC1	613129	Cerebroretinal microangiopathy with calcifications and cysts, 612199 (3), Autosomal recessive
CTH	607657	Cystathioninuria, 219500 (3), Autosomal recessive; Homocysteine, total plasma, elevated (3)
CTNS	606272	Cystinosis, atypical nephropathic, 219800 (3), Autosomal recessive; Cystinosis, late-onset juvenile or adolescent nephropathic, 219900 (3), Autosomal recessive; Cystinosis, nephropathic, 219800 (3), Autosomal recessive; Cystinosis, ocular nonnephropathic, 219750 (3), Autosomal recessive
CTSA	613111	Galactosialidosis, 256540 (3), Autosomal recessive
CTSC	602365	Haim-Munk syndrome, 245010 (3), Autosomal recessive; Papillon-Lefevre syndrome, 245000 (3), Autosomal recessive; Periodontitis 1, juvenile, 170650 (3), Autosomal recessive
CTSD	116840	Ceroid lipofuscinosis, neuronal, 10, 610127 (3), Autosomal recessive
CTSK	601105	Pycnodysostosis, 265800 (3), Autosomal recessive
CUBN	602997	Megaloblastic anemia-1, Finnish type, 261100 (3), Autosomal recessive
CUL7	609577	3-M syndrome 1, 273750 (3), Autosomal recessive
CYB5R3	613213	Methemoglobinemia, type I, 250800 (3), Autosomal recessive; Methemoglobinemia, type II, 250800 (3), Autosomal recessive

CYBA	608508	Chronic granulomatous disease, autosomal, due to deficiency of CYBA, 233690 (3), Autosomal recessive
CYP11A1	118485	Adrenal insufficiency, congenital, with 46XY sex reversal, partial or complete, 613743 (3)
CYP11B1	610613	Adrenal hyperplasia, congenital, due to 11-beta-hydroxylase deficiency, 202010 (3), Autosomal recessive; Aldosteronism, glucocorticoid-remediable, 103900 (3), Autosomal dominant
CYP11B2	124080	Aldosterone to renin ratio raised (3); Hypoaldosteronism, congenital, due to CMO I deficiency, 203400 (3), Autosomal recessive; Hypoaldosteronism, congenital, due to CMO II deficiency, 610600 (3), Autosomal recessive; {Low renin hypertension, susceptibility to} (3)
CYP17A1	609300	17-alpha-hydroxylase/17,20-lyase deficiency, 202110 (3), Autosomal recessive; 17,20-lyase deficiency, isolated, 202110 (3), Autosomal recessive
CYP19A1	107910	Aromatase deficiency, 613546 (3); Aromatase excess syndrome, 139300 (3), Autosomal dominant
CYP1B1	601771	Anterior segment dysgenesis 6, multiple subtypes, 617315 (3); Glaucoma 3A, primary open angle, congenital, juvenile, or adult onset, 231300 (3), Autosomal recessive
CYP21A2	613815	Adrenal hyperplasia, congenital, due to 21-hydroxylase deficiency, 201910 (3), Autosomal recessive; Hyperandrogenism, nonclassic type, due to 21-hydroxylase deficiency, 201910 (3), Autosomal recessive
CYP24A1	126065	Hypercalcemia, infantile, 1, 143880 (3), Autosomal recessive
CYP27A1	606530	Cerebrotendinous xanthomatosis, 213700 (3), Autosomal recessive
CYP27B1	609506	Vitamin D-dependent rickets, type I, 264700 (3), Autosomal recessive
CYP2U1	610670	Spastic paraplegia 56, autosomal recessive, 615030 (3), Autosomal recessive
CYP4V2	608614	Bietti crystalline corneoretinal dystrophy, 210370 (3), Autosomal recessive
CYP7B1	603711	Bile acid synthesis defect, congenital, 3, 613812 (3), Autosomal recessive; Spastic paraplegia 5A, autosomal recessive, 270800 (3), Autosomal recessive
D2HGDH	609186	D-2-hydroxyglutaric aciduria, 600721 (3), Autosomal recessive
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
DBH	609312	Orthostatic hypotension 1, due to DBH deficiency, 223360 (3), Autosomal recessive
DBT	248610	Maple syrup urine disease, type II, 248600 (3), Autosomal recessive
DCHS1	603057	Mitral valve prolapse 2, 607829 (3), Autosomal dominant; Van Maldergem syndrome 1, 601390 (3), Autosomal recessive

DCLRE1C	605988	Omenn syndrome, 603554 (3), Autosomal recessive; Severe combined immunodeficiency, Athabascan type, 602450 (3), Autosomal recessive
DDB2	600811	Xeroderma pigmentosum, group E, DDB-negative subtype, 278740 (3), Autosomal recessive
DDC	107930	Aromatic L-amino acid decarboxylase deficiency, 608643 (3), Autosomal recessive
DDHD2	615003	Spastic paraplegia 54, autosomal recessive, 615033 (3), Autosomal recessive
DDX59	615464	Orofaciodigital syndrome V, 174300 (3), Autosomal recessive
DES	125660	Cardiomyopathy, dilated, 1I, 604765 (3); Myopathy, myofibrillar, 1, 601419 (3), Autosomal recessive, Autosomal dominant; Scapuloperoneal syndrome, neurogenic, Kaeser type, 181400 (3), Autosomal dominant
DGUOK	601465	Mitochondrial DNA depletion syndrome 3 (hepatocerebral type), 251880 (3), Autosomal recessive; Portal hypertension, noncirrhotic, 617068 (3), Autosomal recessive; Progressive external ophthalmoplegia with mitochondrial DNA deletions, autosomal recessive 4, 617070 (3), Autosomal recessive
DHCR24	606418	Desmosterolosis, 602398 (3), Autosomal recessive
DHCR7	602858	Smith-Lemli-Opitz syndrome, 270400 (3), Autosomal recessive
DHDDS	608172	?Congenital disorder of glycosylation, type 1bb, 613861 (3), Autosomal recessive; Developmental delay and seizures with or without movement abnormalities, 617836 (3), Autosomal dominant; Retinitis pigmentosa 59, 613861 (3), Autosomal recessive
DHH	605423	46XY partial gonadal dysgenesis, with minifascicular neuropathy, 607080 (3); 46XY sex reversal 7, 233420 (3), Autosomal recessive
DHODH	126064	Miller syndrome, 263750 (3), Autosomal recessive
DIS3L2	614184	Perlman syndrome, 267000 (3), Autosomal recessive
DLD	238331	Dihydrolipoamide dehydrogenase deficiency, 246900 (3), Autosomal recessive
DLL3	602768	Spondylocostal dysostosis 1, autosomal recessive, 277300 (3), Autosomal recessive
DLX5	600028	?Split-hand/foot malformation 1 with sensorineural hearing loss, 220600 (3), Autosomal recessive
DMP1	600980	Hypophosphatemic rickets, AR, 241520 (3), Autosomal recessive
DNAAF1	613190	Ciliary dyskinesia, primary, 13, 613193 (3), Autosomal recessive
DNAH11	603339	Ciliary dyskinesia, primary, 7, with or without situs inversus, 611884 (3), Autosomal recessive
DNAH5	603335	Ciliary dyskinesia, primary, 3, with or without situs inversus, 608644 (3)
DNAI1	604366	Ciliary dyskinesia, primary, 1, with or without situs inversus, 244400 (3), Autosomal recessive
DNAI2	605483	Ciliary dyskinesia, primary, 9, with or without situs inversus, 612444 (3)

DNAJC19	608977	3-methylglutaconic aciduria, type V, 610198 (3), Autosomal recessive
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
DNMT3B	602900	Immunodeficiency-centromeric instability-facial anomalies syndrome 1, 242860 (3), Autosomal recessive
DOCK6	614194	Adams-Oliver syndrome 2, 614219 (3), Autosomal recessive
DOCK8	611432	Hyper-IgE recurrent infection syndrome, autosomal recessive, 243700 (3), Autosomal recessive
DOK7	610285	?Fetal akinesia deformation sequence 3, 618389 (3); Myasthenic syndrome, congenital, 10, 254300 (3), Autosomal recessive
DOLK	610746	Congenital disorder of glycosylation, type Im, 610768 (3), Autosomal recessive
DPAGT1	191350	Congenital disorder of glycosylation, type Ij, 608093 (3), Autosomal recessive; Myasthenic syndrome, congenital, 13, with tubular aggregates, 614750 (3), Autosomal recessive
DPM1	603503	Congenital disorder of glycosylation, type Ie, 608799 (3), Autosomal recessive
DPYD	612779	Dihydropyrimidine dehydrogenase deficiency, 274270 (3), Autosomal recessive; 5-fluorouracil toxicity, 274270 (3), Autosomal recessive
DSC2	125645	Arrhythmogenic right ventricular dysplasia 11, 610476 (3), Autosomal recessive, Autosomal dominant; Arrhythmogenic right ventricular dysplasia 11 with mild palmoplantar keratoderma and woolly hair, 610476 (3), Autosomal recessive, Autosomal dominant
DSG1	125670	Erythroderma, congenital, with palmoplantar keratoderma, hypotrichosis, and hyper IgE, 615508 (3), Autosomal recessive; Keratosis palmoplantaris striata I, AD, 148700 (3), Autosomal dominant
DSP	125647	Arrhythmogenic right ventricular dysplasia 8, 607450 (3), Autosomal dominant; Cardiomyopathy, dilated, with woolly hair and keratoderma, 605676 (3), Autosomal recessive; Dilated cardiomyopathy with woolly hair, keratoderma, and tooth agenesis, 615821 (3), Autosomal dominant; Epidermolysis bullosa, lethal acantholytic, 609638 (3), Autosomal recessive; Keratosis palmoplantaris striata II, 612908 (3), Autosomal dominant; Skin fragility-woolly hair syndrome, 607655 (3), Autosomal recessive
DUOX2	606759	Thyroid dyshormonogenesis 6, 607200 (3), Autosomal recessive
DUOXA2	612772	Thyroid dyshormonogenesis 5, 274900 (3), Autosomal recessive
DYM	607461	Dyggve-Melchior-Clausen disease, 223800 (3), Autosomal recessive; Smith-McCort dysplasia, 607326 (3), Autosomal recessive
DYNC2H1	603297	Short-rib thoracic dysplasia 3 with or without polydactyly, 613091 (3), Autosomal recessive, Digenic recessive

DYSF	603009	Miyoshi muscular dystrophy 1, 254130 (3), Autosomal recessive; Muscular dystrophy, limb-girdle, autosomal recessive 2, 253601 (3), Autosomal recessive; Myopathy, distal, with anterior tibial onset, 606768 (3), Autosomal recessive
EARS2	612799	Combined oxidative phosphorylation deficiency 12, 614924 (3), Autosomal recessive
ECEL1	605896	Arthrogryposis, distal, type 5D, 615065 (3), Autosomal recessive
ECM1	602201	Urbach-Wiethe disease, 247100 (3), Autosomal recessive
EDAR	604095	Ectodermal dysplasia 10A, hypohidrotic/hair/nail type, autosomal dominant, 129490 (3), Autosomal dominant; Ectodermal dysplasia 10B, hypohidrotic/hair/tooth type, autosomal recessive, 224900 (3), Autosomal recessive; [Hair morphology 1, hair thickness], 612630 (3)
EDARADD	606603	Ectodermal dysplasia 11A, hypohidrotic/hair/tooth type, autosomal dominant, 614940 (3), Autosomal dominant; Ectodermal dysplasia 11B, hypohidrotic/hair/tooth type, autosomal recessive, 614941 (3), Autosomal recessive
EDN3	131242	Central hypoventilation syndrome, congenital, 209880 (3), Autosomal dominant; {Hirschsprung disease, susceptibility to, 4}, 613712 (3), Autosomal dominant; Waardenburg syndrome, type 4B, 613265 (3), Autosomal recessive, Autosomal dominant
EDNRB	131244	ABCD syndrome, 600501 (3), Autosomal recessive; {Hirschsprung disease, susceptibility to, 2}, 600155 (3), Autosomal dominant; Waardenburg syndrome, type 4A, 277580 (3), Autosomal recessive, Autosomal dominant
EFEMP2	604633	Cutis laxa, autosomal recessive, type IB, 614437 (3), Autosomal recessive
EGR2	129010	Charcot-Marie-Tooth disease, type 1D, 607678 (3), Autosomal dominant; Dejerine-Sottas disease, 145900 (3), Autosomal recessive, Autosomal dominant; Hypomyelinating neuropathy, congenital, 1, 605253 (3), Autosomal recessive, Autosomal dominant
EIF2AK3	604032	Wolcott-Rallison syndrome, 226980 (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	Leukoencephalopathy with vanishing white matter, 603896 (3), Autosomal recessive; Ovarioleukodystrophy, 603896 (3), Autosomal recessive
EIF2B5	603945	Leukoencephalopathy with vanishing white matter, 603896 (3), Autosomal recessive; Ovarioleukodystrophy, 603896 (3), Autosomal recessive

EIF4A3	608546	Robin sequence with cleft mandible and limb anomalies, 268305 (3), Autosomal recessive
ELAC2	605367	Combined oxidative phosphorylation deficiency 17, 615440 (3), Autosomal recessive; {Prostate cancer, hereditary, 2, susceptibility to}, 614731 (3)
ELP1	603722	Dysautonomia, familial, 223900 (3), Autosomal recessive
EMG1	611531	Bowen-Conradi syndrome, 211180 (3), Autosomal recessive
ENPP1	173335	Arterial calcification, generalized, of infancy, 1, 208000 (3), Autosomal recessive; Cole disease, 615522 (3), Autosomal dominant; {Diabetes mellitus, non-insulin-dependent, susceptibility to}, 125853 (3), Autosomal dominant; Hypophosphatemic rickets, autosomal recessive, 2, 613312 (3), Autosomal recessive; {Obesity, susceptibility to}, 601665 (3), Autosomal recessive, Autosomal dominant, Multifactorial
EOGT	614789	Adams-Oliver syndrome 4, 615297 (3), Autosomal recessive
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
EPM2A	607566	Epilepsy, progressive myoclonic 2A (Lafora), 254780 (3), Autosomal recessive
ERBB3	190151	{?Erythroleukemia, familial, susceptibility to}, 133180 (3), Autosomal dominant; ?Lethal congenital contractural syndrome 2, 607598 (3), Autosomal recessive
ERCC1	126380	Cerebrooculofacioskeletal syndrome 4, 610758 (3), Autosomal recessive
ERCC2	126340	?Cerebrooculofacioskeletal syndrome 2, 610756 (3), Autosomal recessive; Trichothiodystrophy 1, photosensitive, 601675 (3), Autosomal recessive; Xeroderma pigmentosum, group D, 278730 (3), Autosomal recessive
ERCC3	133510	Trichothiodystrophy 2, photosensitive, 616390 (3), Autosomal recessive; Xeroderma pigmentosum, group B, 610651 (3), Autosomal recessive
ERCC4	133520	Fanconi anemia, complementation group Q, 615272 (3), Autosomal recessive; XFE progeroid syndrome, 610965 (3), Autosomal recessive; Xeroderma pigmentosum, group F, 278760 (3), Autosomal recessive; Xeroderma pigmentosum, type F/Cockayne syndrome, 278760 (3), Autosomal recessive
ERCC5	133530	Cerebrooculofacioskeletal syndrome 3, 616570 (3), Autosomal recessive; Xeroderma pigmentosum, group G, 278780 (3), Autosomal recessive; Xeroderma pigmentosum, group G/Cockayne syndrome, 278780 (3), Autosomal recessive

ERCC6	609413	Cerebrooculofacioskeletal syndrome 1, 214150 (3), Autosomal recessive; Cockayne syndrome, type B, 133540 (3), Autosomal recessive; De Sanctis-Cacchione syndrome, 278800 (3), Autosomal recessive; {Lung cancer, susceptibility to}, 211980 (3), Autosomal dominant, Somatic mutation; {Macular degeneration, age-related, susceptibility to, 5}, 613761 (3); Premature ovarian failure 11, 616946 (3), Autosomal dominant; UV-sensitive syndrome 1, 600630 (3), Autosomal recessive
ERCC8	609412	Cockayne syndrome, type A, 216400 (3), Autosomal recessive; UV-sensitive syndrome 2, 614621 (3), Autosomal recessive
ERLIN2	611605	Spastic paraplegia 18, autosomal recessive, 611225 (3), Autosomal recessive
ESCO2	609353	Roberts syndrome, 268300 (3), Autosomal recessive; SC phocomelia syndrome, 269000 (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
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
EXOSC3	606489	Pontocerebellar hypoplasia, type 1B, 614678 (3), Autosomal recessive
EYS	612424	Retinitis pigmentosa 25, 602772 (3), Autosomal recessive
F11	264900	Factor XI deficiency, autosomal dominant, 612416 (3); Factor XI deficiency, autosomal recessive, 612416 (3)
F13A1	134570	Factor XIIIA deficiency, 613225 (3), Autosomal recessive; {Myocardial infarction, protection against}, 608446 (3); {Venous thrombosis, protection against}, 188050 (3), Autosomal dominant
F2	176930	Dysprothrombinemia, 613679 (3), Autosomal recessive; Hypoprothrombinemia, 613679 (3), Autosomal recessive; {Pregnancy loss, recurrent, susceptibility to, 2}, 614390 (3), Autosomal dominant; {Stroke, ischemic, susceptibility to}, 601367 (3), Multifactorial; Thrombophilia due to thrombin defect, 188050 (3), Autosomal dominant
F5	612309	{Budd-Chiari syndrome}, 600880 (3), Autosomal recessive; Factor V deficiency, 227400 (3), Autosomal recessive; {Pregnancy loss, recurrent, susceptibility to, 1}, 614389 (3), Autosomal dominant; {Stroke, ischemic, susceptibility to}, 601367 (3), Multifactorial; Thrombophilia due to activated protein C resistance, 188055 (3), Autosomal dominant; {Thrombophilia, susceptibility to, due to factor V Leiden}, 188055 (3), Autosomal dominant
F7	613878	Factor VII deficiency, 227500 (3), Autosomal recessive; {Myocardial infarction, decreased susceptibility to}, 608446 (3)

FA2H	611026	Spastic paraplegia 35, autosomal recessive, 612319 (3), Autosomal recessive
FAH	613871	Tyrosinemia, type I, 276700 (3), Autosomal recessive
FAM126A	610531	Leukodystrophy, hypomyelinating, 5, 610532 (3), Autosomal recessive
FAM161A	613596	Retinitis pigmentosa 28, 606068 (3)
FAM20C	611061	Raine syndrome, 259775 (3), Autosomal recessive
FANCA	607139	Fanconi anemia, complementation group A, 227650 (3), Autosomal recessive
FANCC	613899	Fanconi anemia, complementation group C, 227645 (3), Autosomal recessive
FANCD2	613984	Fanconi anemia, complementation group D2, 227646 (3), Autosomal recessive
FANCE	613976	Fanconi anemia, complementation group E, 600901 (3), Autosomal recessive
FANCF	613897	Fanconi anemia, complementation group F, 603467 (3)
FANCG	602956	Fanconi anemia, complementation group G, 614082 (3)
FANCI	611360	Fanconi anemia, complementation group I, 609053 (3), Autosomal recessive
FANCL	608111	Fanconi anemia, complementation group L, 614083 (3), Autosomal recessive
FARS2	611592	Combined oxidative phosphorylation deficiency 14, 614946 (3), Autosomal recessive; Spastic paraplegia 77, autosomal recessive, 617046 (3), Autosomal recessive
FAS	134637	Autoimmune lymphoproliferative syndrome, type IA, 601859 (3), Autosomal dominant; {Autoimmune lymphoproliferative syndrome}, 601859 (3), Autosomal dominant; Squamous cell carcinoma, burn scar-related, somatic (3)
FASTKD2	612322	?Mitochondrial complex IV deficiency, 220110 (3), Autosomal recessive, Mitochondrial
FAT4	612411	Hennekam lymphangiectasia-lymphedema syndrome 2, 616006 (3), Autosomal recessive; Van Maldergem syndrome 2, 615546 (3), Autosomal recessive
FBLN5	604580	Cutis laxa, autosomal dominant 2, 614434 (3), Autosomal dominant; 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
FBXL4	605654	Mitochondrial DNA depletion syndrome 13 (encephalomyopathic type), 615471 (3), Autosomal recessive
FECH	612386	Protoporphyria, erythropoietic, 1, 177000 (3), Autosomal recessive
FERMT3	607901	Leukocyte adhesion deficiency, type III, 612840 (3), Autosomal recessive

FGA	134820	Afibrinogenemia, congenital, 202400 (3), Autosomal recessive; Amyloidosis, familial visceral, 105200 (3), Autosomal dominant; Dysfibrinogenemia, congenital, 616004 (3); Hypodysfibrinogenemia, congenital, 616004 (3)
FGB	134830	Afibrinogenemia, congenital, 202400 (3), Autosomal recessive; Dysfibrinogenemia, congenital, 616004 (3); Hypofibrinogenemia, congenital, 202400 (3), Autosomal recessive
FGD4	611104	Charcot-Marie-Tooth disease, type 4H, 609311 (3), Autosomal recessive
FGF3	164950	Deafness, congenital with inner ear agenesis, microtia, and microdontia, 610706 (3), Autosomal recessive
FGFR2	176943	Antley-Bixler syndrome without genital anomalies or disordered steroidogenesis, 207410 (3), Autosomal dominant; Apert syndrome, 101200 (3), Autosomal dominant; Beare-Stevenson cutis gyrata syndrome, 123790 (3), Autosomal dominant; Bent bone dysplasia syndrome, 614592 (3), Autosomal dominant; Craniofacial-skeletal-dermatologic dysplasia, 101600 (3), Autosomal dominant; Craniosynostosis, nonspecific (3); Crouzon syndrome, 123500 (3), Autosomal dominant; Gastric cancer, somatic, 613659 (3); Jackson-Weiss syndrome, 123150 (3), Autosomal dominant; LADD syndrome, 149730 (3), Autosomal dominant; Pfeiffer syndrome, 101600 (3), Autosomal dominant; Saethre-Chotzen syndrome, 101400 (3), Autosomal dominant; Scaphocephaly and Axenfeld-Rieger anomaly (3); Scaphocephaly, maxillary retrusion, and mental retardation, 609579 (3)
FGFR3	134934	Achondroplasia, 100800 (3), Autosomal dominant; Bladder cancer, somatic, 109800 (3); CATSHL syndrome, 610474 (3), Autosomal recessive, Autosomal dominant; Cervical cancer, somatic, 603956 (3); Colorectal cancer, somatic, 114500 (3); Crouzon syndrome with acanthosis nigricans, 612247 (3), Autosomal dominant; Hypochondroplasia, 146000 (3), Autosomal dominant; LADD syndrome, 149730 (3), Autosomal dominant; Muenke syndrome, 602849 (3), Autosomal dominant; Nevus, epidermal, somatic, 162900 (3); SADDAN, 616482 (3), Autosomal dominant; Spermatocytic seminoma, somatic, 273300 (3); Thanatophoric dysplasia, type I, 187600 (3), Autosomal dominant; Thanatophoric dysplasia, type II, 187601 (3), Autosomal dominant
FGG	134850	Afibrinogenemia, congenital, 202400 (3), Autosomal recessive; Dysfibrinogenemia, congenital, 616004 (3); Hypodysfibrinogenemia, 616004 (3); Hypofibrinogenemia, congenital, 202400 (3), Autosomal recessive
FH	136850	Fumarase deficiency, 606812 (3), Autosomal recessive; Leiomyomatosis and renal cell cancer, 150800 (3), Autosomal dominant

FIG4	609390	Amyotrophic lateral sclerosis 11, 612577 (3), Autosomal dominant; Charcot-Marie-Tooth disease, type 4J, 611228 (3), Autosomal recessive; ?Polymicrogyria, bilateral temporooccipital, 612691 (3), Autosomal recessive; Yunis-Varon syndrome, 216340 (3), Autosomal recessive
FKBP14	614505	Ehlers-Danlos syndrome, kyphoscoliotic type, 2, 614557 (3), Autosomal recessive
FKRP	606596	Muscular dystrophy-dystroglycanopathy (congenital with brain and eye anomalies), type A, 5, 613153 (3), Autosomal recessive; 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
FKTN	607440	Cardiomyopathy, dilated, 1X, 611615 (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; Muscular dystrophy-dystroglycanopathy (limb-girdle), type C, 4, 611588 (3), Autosomal recessive
FLNB	603381	Atelosteogenesis, type I, 108720 (3), Autosomal dominant; Atelosteogenesis, type III, 108721 (3), Autosomal dominant; Boomerang dysplasia, 112310 (3), Autosomal dominant; Larsen syndrome, 150250 (3), Autosomal dominant; Spondylocarpotarsal synostosis syndrome, 272460 (3), Autosomal recessive
FLVCR2	610865	Proliferative vasculopathy and hydranencephaly-hydrocephaly syndrome, 225790 (3), Autosomal recessive
FMO3	136132	Trimethylaminuria, 602079 (3), Autosomal recessive
FOLR1	136430	Neurodegeneration due to cerebral folate transport deficiency, 613068 (3), Autosomal recessive
FOXE1	602617	Bamforth-Lazarus syndrome, 241850 (3), Autosomal recessive; {Thyroid cancer, nonmedullary, 4}, 616534 (3), Autosomal dominant
FOXN1	600838	T-cell immunodeficiency, congenital alopecia, and nail dystrophy, 601705 (3), Autosomal 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	Bifid nose with or without anorectal and renal anomalies, 608980 (3); Manitoba oculotrichoanal syndrome, 248450 (3), Autosomal recessive; Trigonocephaly 2, 614485 (3), Autosomal dominant
FREM2	608945	Cryptophthalmos, unilateral or bilateral, isolated, 123570 (3), Autosomal recessive; Fraser syndrome 2, 617666 (3), Autosomal recessive
FSHR	136435	Ovarian dysgenesis 1, 233300 (3), Autosomal recessive; Ovarian hyperstimulation syndrome, 608115 (3), Autosomal dominant;

		Ovarian response to FSH stimulation, 276400 (3), Autosomal recessive
FTCD	606806	Glutamate formiminotransferase deficiency, 229100 (3), Autosomal recessive
FTO	610966	Growth retardation, developmental delay, facial dysmorphism, 612938 (3), Autosomal recessive; {Obesity, susceptibility to, BMIQ14}, 612460 (3), Autosomal recessive
FUCA1	612280	Fucosidosis, 230000 (3), Autosomal recessive
G6PC	613742	Glycogen storage disease Ia, 232200 (3), Autosomal recessive
G6PC3	611045	Dursun syndrome, 612541 (3), Autosomal recessive; Neutropenia, severe congenital 4, autosomal recessive, 612541 (3), Autosomal recessive
GAA	606800	Glycogen storage disease II, 232300 (3), Autosomal recessive
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
GALNS	612222	Mucopolysaccharidosis IVA, 253000 (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
GATM	602360	Cerebral creatine deficiency syndrome 3, 612718 (3), Autosomal recessive
GBA	606463	Gaucher disease, perinatal lethal, 608013 (3), Autosomal recessive; Gaucher disease, type I, 230800 (3), Autosomal recessive; Gaucher disease, type II, 230900 (3), Autosomal recessive; Gaucher disease, type III, 231000 (3), Autosomal recessive; Gaucher disease, type IIIC, 231005 (3), Autosomal recessive; {Lewy body dementia, susceptibility to}, 127750 (3), Autosomal dominant; {Parkinson disease, late-onset, susceptibility to}, 168600 (3), Autosomal dominant, Multifactorial
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
GCH1	600225	Dystonia, DOPA-responsive, with or without hyperphenylalaninemia, 128230 (3), Autosomal recessive, Autosomal dominant; Hyperphenylalaninemia, BH4-deficient, B, 233910 (3), Autosomal recessive
GCSH	238330	?Glycine encephalopathy, 605899 (3), Autosomal recessive

GDAP1	606598	Charcot-Marie-Tooth disease, axonal, type 2K, 607831 (3), Autosomal recessive, Autosomal dominant; Charcot-Marie-Tooth disease, axonal, with vocal cord paresis, 607706 (3), Autosomal recessive; Charcot-Marie-Tooth disease, recessive intermediate, A, 608340 (3), Autosomal recessive; Charcot-Marie-Tooth disease, type 4A, 214400 (3), Autosomal recessive
GDF1	602880	Congenital heart defects, multiple types, 6, 613854 (3), Autosomal dominant; Right atrial isomerism (Ivemark), 208530 (3), Autosomal recessive
GDF5	601146	?Acromesomelic dysplasia, Hunter-Thompson type, 201250 (3), Autosomal recessive; Brachydactyly, type A1, C, 615072 (3), Autosomal recessive, Autosomal dominant; Brachydactyly, type A2, 112600 (3), Autosomal dominant; Brachydactyly, type C, 113100 (3), Autosomal dominant; Chondrodysplasia, Grebe type, 200700 (3), Autosomal recessive; Du Pan syndrome, 228900 (3), Autosomal recessive; Multiple synostoses syndrome 2, 610017 (3), Autosomal dominant; {Osteoarthritis-5}, 612400 (3); Symphalangism, proximal, 1B, 615298 (3)
GDF6	601147	Klippel-Feil syndrome 1, autosomal dominant, 118100 (3), Autosomal dominant; Leber congenital amaurosis 17, 615360 (3), Autosomal recessive; Microphthalmia with coloboma 6, digenic, 613703 (3), Autosomal dominant; Microphthalmia, isolated 4, 613094 (3); Multiple synostoses syndrome 4, 617898 (3), Autosomal dominant
GFM1	606639	Combined oxidative phosphorylation deficiency 1, 609060 (3), Autosomal recessive
GH1	139250	Growth hormone deficiency, isolated, type IA, 262400 (3), Autosomal recessive; Growth hormone deficiency, isolated, type IB, 612781 (3); Growth hormone deficiency, isolated, type II, 173100 (3), Autosomal dominant; Kowarski syndrome, 262650 (3), Autosomal recessive
GHR	600946	Growth hormone insensitivity, partial, 604271 (3), Autosomal dominant; {Hypercholesterolemia, familial, modifier of}, 143890 (3), Autosomal dominant; Increased responsiveness to growth hormone, 604271 (3), Autosomal dominant; Laron dwarfism, 262500 (3), Autosomal recessive
GIPC3	608792	Deafness, autosomal recessive 15, 601869 (3), Autosomal recessive
GJA1	121014	Atrioventricular septal defect 3, 600309 (3), Autosomal dominant; Craniometaphyseal dysplasia, autosomal recessive, 218400 (3), Autosomal recessive; Erythrokeratodermia variabilis et progressiva 3, 617525 (3), Autosomal dominant; Hypoplastic left heart syndrome 1, 241550 (3), Autosomal recessive; Oculodentodigital dysplasia, 164200 (3), Autosomal dominant; Oculodentodigital dysplasia, autosomal recessive, 257850 (3), Autosomal recessive; Palmoplantar keratoderma with congenital alopecia, 104100 (3), Autosomal dominant; Syndactyly, type III, 186100 (3), Autosomal dominant

GJB2	121011	Bart-Pumphrey syndrome, 149200 (3), Autosomal dominant; Deafness, autosomal dominant 3A, 601544 (3), Autosomal dominant; Deafness, autosomal recessive 1A, 220290 (3), Autosomal recessive; Hystrix-like ichthyosis with deafness, 602540 (3), Autosomal dominant; Keratitis-ichthyosis-deafness syndrome, 148210 (3), Autosomal dominant; Keratoderma, palmoplantar, with deafness, 148350 (3), Autosomal dominant; Vohwinkel syndrome, 124500 (3), Autosomal dominant
GJB6	604418	Deafness, autosomal dominant 3B, 612643 (3), Autosomal dominant; Deafness, autosomal recessive 1B, 612645 (3), Autosomal recessive; Deafness, digenic GJB2/GJB6, 220290 (3), Autosomal recessive; Ectodermal dysplasia 2, Clouston type, 129500 (3), Autosomal dominant
GJC2	608803	Leukodystrophy, hypomyelinating, 2, 608804 (3), Autosomal recessive; Lymphatic malformation 3, 613480 (3), Autosomal dominant; Spastic paraplegia 44, autosomal recessive, 613206 (3), Autosomal recessive
GLB1	611458	GM1-gangliosidosis, type I, 230500 (3), Autosomal recessive; GM1-gangliosidosis, type II, 230600 (3), Autosomal recessive; GM1-gangliosidosis, type III, 230650 (3), Autosomal recessive; Mucopolysaccharidosis type IVB (Morquio), 253010 (3), Autosomal recessive
GLDC	238300	Glycine encephalopathy, 605899 (3), Autosomal recessive
GLE1	603371	Congenital arthrogryposis with anterior horn cell disease, 611890 (3), Autosomal recessive; Lethal congenital contracture syndrome 1, 253310 (3), Autosomal recessive
GLIS3	610192	Diabetes mellitus, neonatal, with congenital hypothyroidism, 610199 (3), Autosomal recessive
GLRA1	138491	Hyperekplexia 1, 149400 (3), Autosomal recessive, Autosomal dominant
GLUL	138290	Glutamine deficiency, congenital, 610015 (3), Autosomal recessive
GMPPB	615320	Muscular dystrophy-dystroglycanopathy (congenital with brain and eye anomalies), type A, 14, 615350 (3), Autosomal recessive; Muscular dystrophy-dystroglycanopathy (congenital with mental retardation), type B, 14, 615351 (3), Autosomal recessive; Muscular dystrophy-dystroglycanopathy (limb-girdle), type C, 14, 615352 (3), Autosomal recessive
GNAS	139320	ACTH-independent macronodular adrenal hyperplasia, 219080 (3), Somatic mutation; McCune-Albright syndrome, somatic, mosaic, 174800 (3); Osseous heteroplasia, progressive, 166350 (3), Autosomal dominant; Pituitary adenoma 3, multiple types, somatic, 617686 (3); Pseudohypoparathyroidism Ia, 103580 (3), Autosomal dominant; Pseudohypoparathyroidism Ib, 603233 (3), Autosomal dominant; Pseudohypoparathyroidism Ic, 612462 (3), Autosomal dominant; Pseudopseudohypoparathyroidism, 612463 (3), Autosomal dominant
GNAT2	139340	Achromatopsia 4, 613856 (3)

GNE	603824	Nonaka myopathy, 605820 (3), Autosomal recessive; Sialuria, 269921 (3), Autosomal dominant
GNMT	606628	Glycine N-methyltransferase deficiency, 606664 (3), Autosomal recessive
GNPAT	602744	Rhizomelic chondrodysplasia punctata, type 2, 222765 (3), Autosomal recessive
GNPTAB	607840	Mucolipidosis II alpha/beta, 252500 (3), Autosomal recessive; Mucolipidosis III alpha/beta, 252600 (3), Autosomal recessive
GNPTG	607838	Mucolipidosis III gamma, 252605 (3), Autosomal recessive
GNRHR	138850	Hypogonadotropic hypogonadism 7 without anosmia, 146110 (3), Autosomal recessive
GNS	607664	Mucopolysaccharidosis type IIID, 252940 (3), Autosomal recessive
GP1BA	606672	Bernard-Soulier syndrome, type A1 (recessive), 231200 (3), Autosomal recessive; Bernard-Soulier syndrome, type A2 (dominant), 153670 (3), Autosomal dominant; {Nonarteritic anterior ischemic optic neuropathy, susceptibility to}, 258660 (3), Autosomal recessive; von Willebrand disease, platelet-type, 177820 (3), Autosomal dominant
GP1BB	138720	Bernard-Soulier syndrome, type B, 231200 (3), Autosomal recessive; Giant platelet disorder, isolated, 231200 (3), Autosomal recessive
GP9	173515	Bernard-Soulier syndrome, type C, 231200 (3), Autosomal recessive
GPC6	604404	Omodysplasia 1, 258315 (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
GPSM2	609245	Chudley-McCullough syndrome, 604213 (3), Autosomal recessive
GRHPR	604296	Hyperoxaluria, primary, type II, 260000 (3), Autosomal recessive
GRIP1	604597	Fraser syndrome 3, 617667 (3), Autosomal recessive
GRM1	604473	Spinocerebellar ataxia 44, 617691 (3), Autosomal dominant; Spinocerebellar ataxia, autosomal recessive 13, 614831 (3), Autosomal recessive
GRN	138945	Aphasia, primary progressive, 607485 (3), Autosomal dominant; Ceroid lipofuscinosis, neuronal, 11, 614706 (3), Autosomal recessive; Frontotemporal lobar degeneration with ubiquitin-positive inclusions, 607485 (3), Autosomal dominant
GSS	601002	Glutathione synthetase deficiency, 266130 (3), Autosomal recessive; Hemolytic anemia due to glutathione synthetase deficiency, 231900 (3), Autosomal recessive
GTF2H5	608780	Trichothiodystrophy 3, photosensitive, 616395 (3)
GUCY2D	600179	?Choroidal dystrophy, central areolar 1, 215500 (3), Autosomal dominant; Cone-rod dystrophy 6, 601777 (3), Autosomal recessive, Autosomal dominant; Leber congenital amaurosis 1, 204000 (3), Autosomal recessive; Night blindness, congenital stationary, type 1I, 618555 (3), Autosomal recessive
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
GYS2	138571	Glycogen storage disease 0, liver, 240600 (3), Autosomal recessive
H6PD	138090	Cortisone reductase deficiency 1, 604931 (3), Autosomal recessive
HADH	601609	3-hydroxyacyl-CoA dehydrogenase deficiency, 231530 (3), Autosomal recessive; Hyperinsulinemic hypoglycemia, familial, 4, 609975 (3), Autosomal recessive
HADHA	600890	Fatty liver, acute, of pregnancy, 609016 (3), Autosomal recessive; HELLP syndrome, maternal, of pregnancy, 609016 (3), Autosomal recessive; LCHAD deficiency, 609016 (3), Autosomal recessive; Trifunctional protein deficiency, 609015 (3), Autosomal recessive
HADHB	143450	Trifunctional protein deficiency, 609015 (3), Autosomal recessive
HAL	609457	[Histidinemia], 235800 (3), Autosomal recessive, Autosomal dominant
HAMP	606464	Hemochromatosis, type 2B, 613313 (3), Autosomal recessive
HAX1	605998	Neutropenia, severe congenital 3, autosomal recessive, 610738 (3), Autosomal recessive
HBA1	141800	Erythrocytosis, 7, 617981 (3); Heinz body anemias, alpha-, 140700 (3), Autosomal dominant; Hemoglobin H disease, nondeletional, 613978 (3); Methemoglobinemia, alpha type, 617973 (3); Thalassemias, alpha-, 604131 (3)
HBA2	141850	Erythrocytosis 7, 617981 (3); Heinz body anemia, 140700 (3), Autosomal dominant; Hemoglobin H disease, deletional and nondeletional, 613978 (3); Thalassemia, alpha-, 604131 (3)
HBB	141900	Delta-beta thalassemia, 141749 (3), Autosomal dominant; Erythrocytosis 6, 617980 (3); Heinz body anemia, 140700 (3), Autosomal dominant; Hereditary persistence of fetal hemoglobin, 141749 (3), Autosomal dominant; {Malaria, resistance to}, 611162 (3); Methmoglobinemia, beta type, 617971 (3); Sickle cell anemia, 603903 (3), Autosomal recessive; Thalassemia, beta, 613985 (3); Thalassemia-beta, dominant inclusion-body, 603902 (3)
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
HES7	608059	Spondylocostal dysostosis 4, autosomal recessive, 613686 (3), Autosomal recessive
HESX1	601802	Growth hormone deficiency with pituitary anomalies, 182230 (3), Autosomal recessive, Autosomal dominant; Pituitary hormone deficiency, combined, 5, 182230 (3), Autosomal recessive, Autosomal dominant; Septooptic dysplasia, 182230 (3), Autosomal recessive, Autosomal dominant
HEXA	606869	GM2-gangliosidosis, several forms, 272800 (3), Autosomal recessive; [Hex A pseudodeficiency], 272800 (3), Autosomal recessive; Tay-Sachs disease, 272800 (3), Autosomal recessive

HEXB	606873	Sandhoff disease, infantile, juvenile, and adult forms, 268800 (3), Autosomal recessive
HFE	613609	{Alzheimer disease, susceptibility to}, 104300 (3), Autosomal dominant; Hemochromatosis, 235200 (3), Autosomal recessive; {Microvascular complications of diabetes 7}, 612635 (3); {Porphyria cutanea tarda, susceptibility to}, 176100 (3), Autosomal recessive, Autosomal dominant; {Porphyria variegata, susceptibility to}, 176200 (3), Autosomal dominant; [Transferrin serum level QTL2], 614193 (3)
HGD	607474	Alkaptonuria, 203500 (3), Autosomal recessive
HGSNAT	610453	Mucopolysaccharidosis type IIIC (Sanfilippo C), 252930 (3), Autosomal recessive; Retinitis pigmentosa 73, 616544 (3), Autosomal recessive
HIBCH	610690	3-hydroxyisobutryl-CoA hydrolase deficiency, 250620 (3), Autosomal recessive
HJV	608374	Hemochromatosis, type 2A, 602390 (3), Autosomal recessive
HLCS	609018	Holocarboxylase synthetase deficiency, 253270 (3), Autosomal recessive
HMGCL	613898	HMG-CoA lyase deficiency, 246450 (3), Autosomal recessive
HMGCS2	600234	HMG-CoA synthase-2 deficiency, 605911 (3), Autosomal recessive
HOGA1	613597	Hyperoxaluria, primary, type III, 613616 (3)
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
HPD	609695	Hawkinsinuria, 140350 (3), Autosomal dominant; Tyrosinemia, type III, 276710 (3), Autosomal recessive
HPGD	601688	Cranioosteoarthropathy, 259100 (3), Autosomal recessive; Digital clubbing, isolated congenital, 119900 (3), Autosomal recessive; Hypertrophic osteoarthropathy, primary, autosomal recessive 1, 259100 (3), Autosomal recessive
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
HPS6	607522	Hermansky-Pudlak syndrome 6, 614075 (3), Autosomal recessive
HSD11B2	614232	Apparent mineralocorticoid excess, 218030 (3), Autosomal recessive
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

HSPG2	142461	Dyssegmental dysplasia, Silverman-Handmaker type, 224410 (3), Autosomal recessive; Schwartz-Jampel syndrome, type 1, 255800 (3), Autosomal recessive
HTRA1	602194	CARASIL syndrome, 600142 (3), Autosomal recessive; Cerebral arteriopathy, autosomal dominant, with subcortical infarcts and leukoencephalopathy, type 2, 616779 (3), Autosomal dominant; {Macular degeneration, age-related, 7}, 610149 (3); {Macular degeneration, age-related, neovascular type}, 610149 (3)
HYAL1	607071	?Mucopolysaccharidosis type IX, 601492 (3), Autosomal recessive
HYLS1	610693	Hydrolethalus syndrome, 236680 (3), Autosomal recessive
IBA57	615316	Multiple mitochondrial dysfunctions syndrome 3, 615330 (3), Autosomal recessive; ?Spastic paraplegia 74, autosomal recessive, 616451 (3), Autosomal recessive
IDUA	252800	Mucopolysaccharidosis Ih, 607014 (3), Autosomal recessive; Mucopolysaccharidosis Ih/s, 607015 (3), Autosomal recessive; Mucopolysaccharidosis Is, 607016 (3), Autosomal recessive
IER3IP1	609382	Microcephaly, epilepsy, and diabetes syndrome, 614231 (3), Autosomal recessive
IFNGR1	107470	{H. pylori infection, susceptibility to}, 600263 (3); {Hepatitis B virus infection, susceptibility to}, 610424 (3); Immunodeficiency 27A, mycobacteriosis, AR, 209950 (3), Autosomal recessive; Immunodeficiency 27B, mycobacteriosis, AD, 615978 (3), Autosomal dominant; {Tuberculosis infection, protection against}, 607948 (3); {Tuberculosis, susceptibility to}, 607948 (3)
IFNGR2	147569	Immunodeficiency 28, mycobacteriosis, 614889 (3), Autosomal recessive
IFT122	606045	Cranioectodermal dysplasia 1, 218330 (3), Autosomal recessive
IFT140	614620	Retinitis pigmentosa 80, 617781 (3), Autosomal recessive; Short-rib thoracic dysplasia 9 with or without polydactyly, 266920 (3), Autosomal recessive
IFT172	607386	Retinitis pigmentosa 71, 616394 (3), Autosomal recessive; Short-rib thoracic dysplasia 10 with or without polydactyly, 615630 (3), Autosomal recessive
IFT27	615870	?Bardet-Biedl syndrome 19, 615996 (3), Autosomal recessive
IFT80	611177	Short-rib thoracic dysplasia 2 with or without polydactyly, 611263 (3), Autosomal recessive
IGF1	147440	Growth retardation with deafness and mental retardation due to IGF1 deficiency, 608747 (3), Autosomal recessive
IGHMBP2	600502	Charcot-Marie-Tooth disease, axonal, type 2S, 616155 (3), Autosomal recessive; Neuronopathy, distal hereditary motor, type VI, 604320 (3), Autosomal recessive
IHH	600726	Acrocapitofemoral dysplasia, 607778 (3), Autosomal recessive; Brachydactyly, type A1, 112500 (3), Autosomal dominant
IL12B	161561	Immunodeficiency 29, mycobacteriosis, 614890 (3), Autosomal recessive

IL12RB1	601604	Immunodeficiency 30, 614891 (3), Autosomal recessive
IL1RN	147679	{Gastric cancer risk after H. pylori infection}, 137215 (3), Autosomal dominant; Interleukin 1 receptor antagonist deficiency, 612852 (3), Autosomal recessive; {Microvascular complications of diabetes 4}, 612628 (3)
IL36RN	605507	Psoriasis 14, pustular, 614204 (3), Autosomal recessive
IL7R	146661	Severe combined immunodeficiency, T-cell negative, B-cell/natural killer cell-positive type, 608971 (3), Autosomal recessive
IMPG2	607056	Macular dystrophy, vitelliform, 5, 616152 (3), Autosomal dominant; Retinitis pigmentosa 56, 613581 (3), Autosomal recessive
INPP5E	613037	Joubert syndrome 1, 213300 (3), Autosomal recessive; Mental retardation, truncal obesity, retinal dystrophy, and micropenis, 610156 (3), Autosomal recessive
INSR	147670	Diabetes mellitus, insulin-resistant, with acanthosis nigricans, 610549 (3); Hyperinsulinemic hypoglycemia, familial, 5, 609968 (3), Autosomal dominant; Leprechaunism, 246200 (3), Autosomal recessive; Rabson-Mendenhall syndrome, 262190 (3), Autosomal recessive
INVS	243305	Nephronophthisis 2, infantile, 602088 (3), Autosomal recessive
IQCB1	609237	Senior-Loken syndrome 5, 609254 (3), Autosomal recessive
ITGA2B	607759	Bleeding disorder, platelet-type, 16, autosomal dominant, 187800 (3), Autosomal dominant; Glanzmann thrombasthenia, 273800 (3), Autosomal recessive; Thrombocytopenia, neonatal alloimmune, BAK antigen related (3)
ITGA6	147556	Epidermolysis bullosa, junctional, with pyloric stenosis, 226730 (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, 16, autosomal dominant, 187800 (3), Autosomal dominant; Glanzmann thrombasthenia, 273800 (3), Autosomal recessive; {Myocardial infarction, susceptibility to}, 608446 (3); Purpura, posttransfusion (3); Thrombocytopenia, neonatal alloimmune (3)
ITGB4	147557	Epidermolysis bullosa of hands and feet, 131800 (3), Autosomal dominant; Epidermolysis bullosa, junctional, non-Herlitz type, 226650 (3), Autosomal recessive; Epidermolysis bullosa, junctional, with pyloric atresia, 226730 (3), Autosomal recessive
IVD	607036	Isovaleric acidemia, 243500 (3), Autosomal recessive
IYD	612025	Thyroid dyshormonogenesis 4, 274800 (3), Autosomal recessive
JAK3	600173	SCID, autosomal recessive, T-negative/B-positive type, 600802 (3), Autosomal recessive
JAM3	606871	Hemorrhagic destruction of the brain, subependymal calcification, and cataracts, 613730 (3), Autosomal recessive
JUP	173325	Arrhythmogenic right ventricular dysplasia 12, 611528 (3), Autosomal dominant; Naxos disease, 601214 (3), Autosomal recessive

KARS1 (KARS)	601421	?Charcot-Marie-Tooth disease, recessive intermediate, B, 613641 (3), Autosomal recessive; Deafness, autosomal recessive 89, 613916 (3), Autosomal recessive
KCNJ1	600359	Bartter syndrome, type 2, 241200 (3), Autosomal recessive
KCNJ11	600937	Diabetes mellitus, transient neonatal, 3, 610582 (3), Autosomal dominant; {Diabetes mellitus, type 2, susceptibility to}, 125853 (3), Autosomal dominant; Diabetes, permanent neonatal, with or without neurologic features, 606176 (3), Autosomal recessive, Autosomal dominant; Hyperinsulinemic hypoglycemia, familial, 2, 601820 (3), Autosomal recessive; Maturity-onset diabetes of the young, type 13, 616329 (3), Autosomal dominant
KCNJ13	603208	Leber congenital amaurosis 16, 614186 (3), Autosomal recessive; Snowflake vitreoretinal degeneration, 193230 (3), Autosomal dominant
KCNQ1	607542	Atrial fibrillation, familial, 3, 607554 (3), Autosomal dominant; Jervell and Lange-Nielsen syndrome, 220400 (3), Autosomal recessive; Long QT syndrome 1, 192500 (3), Autosomal dominant; {Long QT syndrome 1, acquired, susceptibility to}, 192500 (3), Autosomal dominant; Short QT syndrome 2, 609621 (3), Autosomal dominant
KCNV2	607604	Retinal cone dystrophy 3B, 610356 (3), Autosomal recessive
KCTD7	611725	Epilepsy, progressive myoclonic 3, with or without intracellular inclusions, 611726 (3), Autosomal recessive
KHDC3L	611687	Hydatidiform mole, recurrent, 2, 614293 (3), Autosomal recessive
KIF14	611279	?Meckel syndrome 12, 616258 (3), Autosomal recessive; Microcephaly 20, primary, autosomal recessive, 617914 (3), Autosomal recessive
KIF1A	601255	Mental retardation, autosomal dominant 9, 614255 (3), Autosomal dominant; Neuropathy, hereditary sensory, type IIC, 614213 (3), Autosomal recessive; Spastic paraplegia 30, autosomal recessive, 610357 (3), Autosomal recessive
KIF1BP	No OMIM gene	No OMIM phenotype
KIF7	611254	Acrocallosal syndrome, 200990 (3), Autosomal recessive; ?Al-Gazali-Bakalinova syndrome, 607131 (3), Autosomal recessive; ?Hydrolethalus syndrome 2, 614120 (3), Autosomal recessive; Joubert syndrome 12, 200990 (3), Autosomal recessive
KISS1R	604161	Hypogonadotropic hypogonadism 8 with or without anosmia, 614837 (3), Autosomal recessive; ?Precocious puberty, central, 1, 176400 (3), Autosomal dominant
KRT14	148066	Dermatopathia pigmentosa reticularis, 125595 (3), Autosomal dominant; Epidermolysis bullosa simplex, Dowling-Meara type, 131760 (3), Autosomal dominant; Epidermolysis bullosa simplex, Koebner type, 131900 (3), Autosomal dominant; Epidermolysis bullosa simplex, Weber-Cockayne type, 131800 (3), Autosomal dominant; Epidermolysis bullosa simplex, recessive 1, 601001 (3),

		Autosomal recessive; Naegeli-Franceschetti-Jadassohn syndrome, 161000 (3), Autosomal dominant
KRT18	148070	Cirrhosis, cryptogenic, 215600 (3), Autosomal recessive; {Cirrhosis, noncryptogenic, susceptibility to}, 215600 (3), Autosomal recessive
KRT5	148040	Dowling-Degos disease 1, 179850 (3), Autosomal dominant; Epidermolysis bullosa simplex, Dowling-Meara type, 131760 (3), Autosomal dominant; Epidermolysis bullosa simplex, Koebner type, 131900 (3), Autosomal dominant; Epidermolysis bullosa simplex, Weber-Cockayne type, 131800 (3), Autosomal dominant; Epidermolysis bullosa simplex, recessive 1, 601001 (3), Autosomal recessive; Epidermolysis bullosa simplex-MCR, 609352 (3); Epidermolysis bullosa simplex-MP, 131960 (3), Autosomal dominant
KRT8	148060	Cirrhosis, cryptogenic, 215600 (3), Autosomal recessive; {Cirrhosis, noncryptogenic, susceptibility to}, 215600 (3), Autosomal recessive
L2HGDH	609584	L-2-hydroxyglutaric aciduria, 236792 (3), Autosomal recessive
LAMA1	150320	Poretti-Boltshauser syndrome, 615960 (3), Autosomal recessive
LAMA2	156225	Muscular dystrophy, congenital, merosin deficient or partially deficient, 607855 (3), Autosomal recessive; Muscular dystrophy, limb-girdle, autosomal recessive 23, 618138 (3), Autosomal recessive
LAMA3	600805	Epidermolysis bullosa, generalized atrophic benign, 226650 (3), Autosomal recessive; Epidermolysis bullosa, junctional, Herlitz type, 226700 (3), Autosomal recessive; Laryngoonychocutaneous syndrome, 245660 (3), Autosomal recessive
LAMB1	150240	Lissencephaly 5, 615191 (3), Autosomal recessive
LAMB2	150325	Nephrotic syndrome, type 5, with or without ocular abnormalities, 614199 (3); Pierson syndrome, 609049 (3), Autosomal recessive
LAMB3	150310	Amelogenesis imperfecta, type IA, 104530 (3), Autosomal dominant; Epidermolysis bullosa, junctional, Herlitz type, 226700 (3), Autosomal recessive; Epidermolysis bullosa, junctional, non-Herlitz type, 226650 (3), Autosomal recessive
LAMC2	150292	Epidermolysis bullosa, junctional, Herlitz type, 226700 (3), Autosomal recessive; Epidermolysis bullosa, junctional, non-Herlitz type, 226650 (3), Autosomal recessive
LAMC3	604349	Cortical malformations, occipital, 614115 (3), Autosomal recessive
LARGE1	603590	Muscular dystrophy-dystroglycanopathy (congenital with brain and eye anomalies), type A, 6, 613154 (3), Autosomal recessive; Muscular dystrophy-dystroglycanopathy (congenital with mental retardation), type B, 6, 608840 (3), Autosomal recessive

LARS2	604544	?Hydrops, lactic acidosis, and sideroblastic anemia, 617021 (3), Autosomal recessive; Perrault syndrome 4, 615300 (3), Autosomal recessive
LBR	600024	Greenberg skeletal dysplasia, 215140 (3), Autosomal recessive; Pelger-Huet anomaly, 169400 (3), Autosomal dominant; Pelger-Huet anomaly with mild skeletal anomalies, 618019 (3); ?Reynolds syndrome, 613471 (3), Autosomal dominant
LCA5	611408	Leber congenital amaurosis 5, 604537 (3), Autosomal recessive
LCAT	606967	Fish-eye disease, 136120 (3), Autosomal recessive; Norum disease, 245900 (3), Autosomal recessive
LDLR	606945	Hypercholesterolemia, familial, 1, 143890 (3), Autosomal dominant; LDL cholesterol level QTL2, 143890 (3), Autosomal dominant
LDLRAP1	605747	Hypercholesterolemia, familial, 4, 603813 (3), Autosomal recessive
LHB	152780	Hypogonadotropic hypogonadism 23 with or without anosmia, 228300 (3), Autosomal recessive
LHCGR	152790	Leydig cell adenoma, somatic, with precocious puberty, 176410 (3); Leydig cell hypoplasia with hypergonadotropic hypogonadism, 238320 (3), Autosomal recessive; Leydig cell hypoplasia with pseudohermaphroditism, 238320 (3), Autosomal recessive; Luteinizing hormone resistance, female, 238320 (3), Autosomal recessive; Precocious puberty, male, 176410 (3), Autosomal dominant
LHX3	600577	Pituitary hormone deficiency, combined, 3, 221750 (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
LIPA	613497	Cholesteryl ester storage disease, 278000 (3), Autosomal recessive; Wolman disease, 278000 (3), Autosomal recessive
LMBR1	605522	Acheiropody, 200500 (3), Autosomal recessive; Hypoplastic or aplastic tibia with polydactyly, 188740 (3), Autosomal dominant; Laurin-Sandrow syndrome, 135750 (3), Autosomal dominant; Polydactyly, preaxial type II, 174500 (3), Autosomal dominant; Syndactyly, type IV, 186200 (3), Autosomal dominant; Triphalangeal thumb, type I, 174500 (3), Autosomal dominant; Triphalangeal thumb-polysyndactyly syndrome, 174500 (3), Autosomal dominant

LMNA	150330	Cardiomyopathy, dilated, 1A, 115200 (3), Autosomal dominant; Charcot-Marie-Tooth disease, type 2B1, 605588 (3), Autosomal recessive; Emery-Dreifuss muscular dystrophy 2, autosomal dominant, 181350 (3), Autosomal dominant; Emery-Dreifuss muscular dystrophy 3, autosomal recessive, 616516 (3), Autosomal recessive; Heart-hand syndrome, Slovenian type, 610140 (3), Autosomal dominant; Hutchinson-Gilford progeria, 176670 (3), Autosomal recessive, Autosomal dominant; Lipodystrophy, familial partial, type 2, 151660 (3), Autosomal dominant; Malouf syndrome, 212112 (3), Autosomal dominant; Mandibuloacral dysplasia, 248370 (3), Autosomal recessive; Muscular dystrophy, congenital, 613205 (3), Autosomal dominant; Restrictive dermopathy, lethal, 275210 (3), Autosomal recessive
LOXHD1	613072	Deafness, autosomal recessive 77, 613079 (3), Autosomal recessive
LPIN2	605519	Majeed syndrome, 609628 (3)
LPL	609708	Combined hyperlipidemia, familial, 144250 (3), Autosomal dominant; [High density lipoprotein cholesterol level QTL 11], 238600 (3), Autosomal recessive; Lipoprotein lipase deficiency, 238600 (3), Autosomal recessive
LRAT	604863	Leber congenital amaurosis 14, 613341 (3), Autosomal recessive; Retinal dystrophy, early-onset severe, 613341 (3), Autosomal recessive; Retinitis pigmentosa, juvenile, 613341 (3), Autosomal recessive
LRP2	600073	Donnai-Barrow syndrome, 222448 (3), Autosomal recessive
LRP4	604270	Cenani-Lenz syndactyly syndrome, 212780 (3), Autosomal recessive; ?Myasthenic syndrome, congenital, 17, 616304 (3), Autosomal recessive; Sclerosteosis 2, 614305 (3), Autosomal recessive, Autosomal dominant
LRP5	603506	[Bone mineral density variability 1], 601884 (3), Autosomal dominant; Exudative vitreoretinopathy 4, 601813 (3), Autosomal recessive, Autosomal dominant; Hyperostosis, endosteal, 144750 (3), Autosomal dominant; Osteopetrosis, autosomal dominant 1, 607634 (3), Autosomal dominant; Osteoporosis-pseudoglioma syndrome, 259770 (3), Autosomal recessive; {Osteoporosis}, 166710 (3), Autosomal dominant; Osteosclerosis, 144750 (3), Autosomal dominant; Polycystic liver disease 4 with or without kidney cysts, 617875 (3), Autosomal dominant; van Buchem disease, type 2, 607636 (3), Autosomal dominant
LRPPRC	607544	Leigh syndrome, French-Canadian type, 220111 (3), Autosomal recessive
LRRC6	614930	Ciliary dyskinesia, primary, 19, 614935 (3), Autosomal recessive
LRTOMT	612414	Deafness, autosomal recessive 63, 611451 (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

LTBP4	604710	Cutis laxa, autosomal recessive, type IC, 613177 (3), Autosomal recessive
LYST	606897	Chediak-Higashi syndrome, 214500 (3), Autosomal recessive
LZTFL1	606568	Bardet-Biedl syndrome 17, 615994 (3), Autosomal recessive
MAK	154235	Retinitis pigmentosa 62, 614181 (3), Autosomal recessive
MAN1B1	604346	Mental retardation, autosomal recessive 15, 614202 (3), Autosomal recessive
MAN2B1	609458	Mannosidosis, alpha-, types I and II, 248500 (3), Autosomal recessive
MAPT	157140	Dementia, frontotemporal, with or without parkinsonism, 600274 (3), Autosomal dominant; {Parkinson disease, susceptibility to}, 168600 (3), Autosomal dominant, Multifactorial; Pick disease, 172700 (3), Autosomal dominant; Supranuclear palsy, progressive, 601104 (3), Autosomal dominant; Supranuclear palsy, progressive atypical, 260540 (3), Autosomal recessive
MASP1	600521	3MC syndrome 1, 257920 (3), Autosomal recessive
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
MC2R	607397	Glucocorticoid deficiency, due to ACTH unresponsiveness, 202200 (3), Autosomal recessive
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
MCOLN1	605248	Mucolipidosis IV, 252650 (3), Autosomal recessive
MCPH1	607117	Microcephaly 1, primary, autosomal recessive, 251200 (3), Autosomal recessive
MED17	603810	Microcephaly, postnatal progressive, with seizures and brain atrophy, 613668 (3), Autosomal recessive
MEFV	608107	Familial Mediterranean fever, AD, 134610 (3), Autosomal dominant; Familial Mediterranean fever, AR, 249100 (3), Autosomal recessive
MEGF10	612453	Myopathy, areflexia, respiratory distress, and dysphagia, early-onset, 614399 (3), Autosomal recessive; Myopathy, areflexia, respiratory distress, and dysphagia, early-onset, mild variant, 614399 (3), Autosomal recessive
MEOX1	600147	Klippel-Feil syndrome 2, 214300 (3), Autosomal recessive
MERTK	604705	Retinitis pigmentosa 38, 613862 (3), Autosomal recessive
MESP2	605195	Spondylocostal dysostosis 2, autosomal recessive, 608681 (3), Autosomal recessive

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
MFSD8	611124	Ceroid lipofuscinosis, neuronal, 7, 610951 (3), Autosomal recessive; Macular dystrophy with central cone involvement, 616170 (3), Autosomal recessive
MGAT2	602616	Congenital disorder of glycosylation, type IIa, 212066 (3), Autosomal recessive
MGP	154870	Keutel syndrome, 245150 (3), Autosomal recessive
MKKS	604896	Bardet-Biedl syndrome 6, 605231 (3), Autosomal recessive; McKusick-Kaufman syndrome, 236700 (3), Autosomal recessive
MKS1	609883	Bardet-Biedl syndrome 13, 615990 (3), Autosomal recessive; Joubert syndrome 28, 617121 (3), Autosomal recessive; Meckel syndrome 1, 249000 (3), Autosomal recessive
MLC1	605908	Megalencephalic leukoencephalopathy with subcortical cysts, 604004 (3), Autosomal recessive
MLH1	120436	Colorectal cancer, hereditary nonpolyposis, type 2, 609310 (3); Mismatch repair cancer syndrome, 276300 (3), Autosomal recessive; Muir-Torre syndrome, 158320 (3), Autosomal dominant
MMAA	607481	Methylmalonic aciduria, vitamin B12-responsive, 251100 (3), Autosomal recessive
MMAB	607568	Methylmalonic aciduria, vitamin B12-responsive, due to defect in synthesis of adenosylcobalamin, cblB complementation type, 251110 (3), Autosomal recessive
MMACHC	609831	Methylmalonic aciduria and homocystinuria, cblC type, 277400 (3), Autosomal recessive
MMADHC	611935	Homocystinuria, cblD type, variant 1, 277410 (3), Autosomal recessive; Methylmalonic aciduria and homocystinuria, cblD type, 277410 (3), Autosomal recessive; Methylmalonic aciduria, cblD type, variant 2, 277410 (3), Autosomal recessive
MMP2	120360	Multicentric osteolysis, nodulosis, and arthropathy, 259600 (3), Autosomal recessive
MMUT (MUT)	609058	Methylmalonic aciduria, mut(0) type, 251000 (3), Autosomal recessive
MOCS1	603707	Molybdenum cofactor deficiency A, 252150 (3), Autosomal recessive
MOCS2	603708	Molybdenum cofactor deficiency B, 252160 (3), Autosomal recessive
MOGS	601336	Congenital disorder of glycosylation, type IIb, 606056 (3), Autosomal recessive
MPDU1	604041	Congenital disorder of glycosylation, type If, 609180 (3), Autosomal recessive
MPI	154550	Congenital disorder of glycosylation, type Ib, 602579 (3), Autosomal recessive

MPL	159530	Myelofibrosis with myeloid metaplasia, somatic, 254450 (3); Thrombocythemia 2, 601977 (3), Autosomal dominant, Somatic mutation; Thrombocytopenia, congenital amegakaryocytic, 604498 (3), Autosomal recessive
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, dominant intermediate D, 607791 (3), Autosomal dominant; Charcot-Marie-Tooth disease, type 1B, 118200 (3), Autosomal dominant; Charcot-Marie-Tooth disease, type 2I, 607677 (3), Autosomal dominant; Charcot-Marie-Tooth disease, type 2J, 607736 (3), Autosomal dominant; Dejerine-Sottas disease, 145900 (3), Autosomal recessive, Autosomal dominant; Hypomyelinating neuropathy, congenital, 2, 618184 (3), Autosomal dominant; Roussy-Levy syndrome, 180800 (3), Autosomal dominant
MRE11	600814	Ataxia-telangiectasia-like disorder 1, 604391 (3), Autosomal recessive
MRPS16	609204	Combined oxidative phosphorylation deficiency 2, 610498 (3), Autosomal recessive
MRPS22	605810	Combined oxidative phosphorylation deficiency 5, 611719 (3), Autosomal recessive; Ovarian dysgenesis 7, 618117 (3), Autosomal recessive
MSH2	609309	Colorectal cancer, hereditary nonpolyposis, type 1, 120435 (3), Autosomal dominant; Mismatch repair cancer syndrome, 276300 (3), Autosomal recessive; Muir-Torre syndrome, 158320 (3), Autosomal dominant
MSH6	600678	Colorectal cancer, hereditary nonpolyposis, type 5, 614350 (3), Autosomal dominant; {Endometrial cancer, familial}, 608089 (3), Autosomal dominant, Somatic mutation; Mismatch repair cancer syndrome, 276300 (3), Autosomal recessive
MTFMT	611766	Combined oxidative phosphorylation deficiency 15, 614947 (3), Autosomal recessive; Mitochondrial complex I deficiency, nuclear type 27, 618248 (3), Autosomal recessive
MTHFR	607093	Homocystinuria due to MTHFR deficiency, 236250 (3), Autosomal recessive; {Neural tube defects, susceptibility to}, 601634 (3), Autosomal recessive; {Schizophrenia, susceptibility to}, 181500 (3), Autosomal dominant; {Thromboembolism, susceptibility to}, 188050 (3), Autosomal dominant; {Vascular disease, susceptibility to} (3)
MTR	156570	Homocystinuria-megaloblastic anemia, cblG complementation type, 250940 (3), Autosomal recessive; {Neural tube defects, folate-sensitive, susceptibility to}, 601634 (3), Autosomal recessive
MTRR	602568	Homocystinuria-megaloblastic anemia, cbl E type, 236270 (3), Autosomal recessive; {Neural tube defects, folate-sensitive, susceptibility to}, 601634 (3), Autosomal recessive
MTTP	157147	Abetalipoproteinemia, 200100 (3), Autosomal recessive; {Metabolic syndrome, protection against}, 605552 (3), Autosomal dominant

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; Colorectal adenomatous polyposis, autosomal recessive, with pilomatricomas, 132600 (3), Somatic mutation; Gastric cancer, somatic, 613659 (3)
MVK	251170	Hyper-IgD syndrome, 260920 (3), Autosomal recessive; Mevalonic aciduria, 610377 (3), Autosomal recessive; Porokeratosis 3, multiple types, 175900 (3), Autosomal dominant
MYD88	602170	Macroglobulinemia, Waldenstrom, somatic, 153600 (3); Pyogenic bacterial infections, recurrent, due to MYD88 deficiency, 612260 (3)
MYH2	160740	Proximal myopathy and ophthalmoplegia, 605637 (3), Autosomal recessive, Autosomal dominant
MYH7	160760	Cardiomyopathy, dilated, 1S, 613426 (3), Autosomal dominant; Cardiomyopathy, hypertrophic, 1, 192600 (3), Autosomal dominant; Laing distal myopathy, 160500 (3), Autosomal dominant; Left ventricular noncompaction 5, 613426 (3), Autosomal dominant; Myopathy, myosin storage, autosomal dominant, 608358 (3), Autosomal dominant; Myopathy, myosin storage, autosomal recessive, 255160 (3), Autosomal recessive; Scapuloperoneal syndrome, myopathic type, 181430 (3), Autosomal dominant
MYO15A	602666	Deafness, autosomal recessive 3, 600316 (3), Autosomal recessive
MYO5A	160777	Griscelli syndrome, type 1, 214450 (3), Autosomal recessive
MYO5B	606540	Microvillus inclusion disease, 251850 (3), Autosomal recessive
MYO6	600970	Deafness, autosomal dominant 22, 606346 (3), Autosomal dominant; Deafness, autosomal dominant 22, with hypertrophic cardiomyopathy, 606346 (3), Autosomal dominant; Deafness, autosomal recessive 37, 607821 (3), Autosomal recessive
MYO7A	276903	Deafness, autosomal dominant 11, 601317 (3), Autosomal dominant; Deafness, autosomal recessive 2, 600060 (3), Autosomal recessive; Usher syndrome, type 1B, 276900 (3), Autosomal recessive
NAGA	104170	Kanzaki disease, 609242 (3), Autosomal recessive; Schindler disease, type I, 609241 (3), Autosomal recessive; Schindler disease, type III, 609241 (3), Autosomal recessive
NAGLU	609701	?Charcot-Marie-Tooth disease, axonal, type 2V, 616491 (3), Autosomal dominant; Mucopolysaccharidosis type IIIB (Sanfilippo B), 252920 (3), Autosomal recessive
NAGS	608300	N-acetylglutamate synthase deficiency, 237310 (3), Autosomal recessive
NBAS	608025	Infantile liver failure syndrome 2, 616483 (3), Autosomal recessive; Short stature, optic nerve atrophy, and Pelger-Huet anomaly, 614800 (3), Autosomal recessive
NBEAL2	614169	Gray platelet syndrome, 139090 (3), Autosomal recessive

NBN	602667	Aplastic anemia, 609135 (3); Leukemia, acute lymphoblastic, 613065 (3); Nijmegen breakage syndrome, 251260 (3), Autosomal recessive
NCF2	608515	Chronic granulomatous disease due to deficiency of NCF-2, 233710 (3), Autosomal recessive
NDE1	609449	Lissencephaly 4 (with microcephaly), 614019 (3), Autosomal recessive; ?Microhydranencephaly, 605013 (3), Autosomal recessive
NDRG1	605262	Charcot-Marie-Tooth disease, type 4D, 601455 (3), Autosomal recessive
NDUFAF2	609653	Mitochondrial complex I deficiency, nuclear type 10, 618233 (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
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
NECTIN1	600644	Cleft lip/palate-ectodermal dysplasia syndrome, 225060 (3), Autosomal recessive; Orofacial cleft 7, 225060 (3), Autosomal recessive
NEFL	162280	Charcot-Marie-Tooth disease, dominant intermediate G, 617882 (3), Autosomal dominant; Charcot-Marie-Tooth disease, type 1F, 607734 (3), Autosomal recessive, Autosomal dominant; Charcot-Marie-Tooth disease, type 2E, 607684 (3), Autosomal dominant
NEK1	604588	{Amyotrophic lateral sclerosis, susceptibility to, 24}, 617892 (3), Autosomal dominant; Short-rib thoracic dysplasia 6 with or without polydactyly, 263520 (3), Autosomal recessive, Digenic recessive
NEU1	608272	Sialidosis, type I, 256550 (3), Autosomal recessive; Sialidosis, type II, 256550 (3), Autosomal recessive
NEUROG3	604882	Diarrhea 4, malabsorptive, congenital, 610370 (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
NIN	608684	?Seckel syndrome 7, 614851 (3), Autosomal recessive
NKX2-1	600635	Chorea, hereditary benign, 118700 (3), Autosomal dominant; Choreoathetosis, hypothyroidism, and neonatal respiratory distress, 610978 (3), Autosomal dominant; {Thyroid cancer, nonmedullary, 1}, 188550 (3), Autosomal dominant
NKX2-5	600584	Atrial septal defect 7, with or without AV conduction defects, 108900 (3), Autosomal dominant; Conotruncal heart malformations, variable, 217095 (3); Hypoplastic left heart syndrome 2, 614435 (3), Autosomal dominant; Hypothyroidism, congenital nongoitrous, 5, 225250 (3), Autosomal dominant; Tetralogy of Fallot, 187500 (3), Autosomal dominant; Ventricular septal defect 3, 614432 (3), Autosomal dominant
NLRP7	609661	Hydatidiform mole, recurrent, 1, 231090 (3), Autosomal recessive
NMNAT1	608700	Leber congenital amaurosis 9, 608553 (3), Autosomal recessive
NPC1	607623	Niemann-Pick disease, type C1, 257220 (3), Autosomal recessive; Niemann-Pick disease, type D, 257220 (3), Autosomal recessive
NPC2	601015	Niemann-pick disease, type C2, 607625 (3), Autosomal recessive
NPHP1	607100	Joubert syndrome 4, 609583 (3), Autosomal recessive; Nephronophthisis 1, juvenile, 256100 (3), Autosomal recessive; Senior-Loken syndrome-1, 266900 (3), Autosomal recessive
NPHP3	608002	Meckel syndrome 7, 267010 (3), Autosomal recessive; Nephronophthisis 3, 604387 (3), Autosomal recessive; Renal-hepatic-pancreatic dysplasia 1, 208540 (3), Autosomal recessive
NPHP4	607215	Nephronophthisis 4, 606966 (3), Autosomal recessive; Senior-Loken syndrome 4, 606996 (3), Autosomal recessive
NPHS1	602716	Nephrotic syndrome, type 1, 256300 (3), Autosomal recessive
NPHS2	604766	Nephrotic syndrome, type 2, 600995 (3), Autosomal recessive
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
NR2E3	604485	Enhanced S-cone syndrome, 268100 (3), Autosomal recessive; Retinitis pigmentosa 37, 611131 (3), Autosomal recessive, Autosomal dominant
NSUN2	610916	Mental retardation, autosomal recessive 5, 611091 (3), Autosomal recessive
NT5C2	600417	Spastic paraplegia 45, autosomal recessive, 613162 (3), Autosomal recessive
NT5C3A	606224	Anemia, hemolytic, due to UMPH1 deficiency, 266120 (3), Autosomal recessive
NTHL1	602656	Familial adenomatous polyposis 3, 616415 (3), Autosomal recessive

NTRK1	191315	Insensitivity to pain, congenital, with anhidrosis, 256800 (3), Autosomal recessive; Medullary thyroid carcinoma, familial, 155240 (3), Autosomal dominant
NUBPL	613621	Mitochondrial complex I deficiency, nuclear type 21, 618242 (3), Autosomal recessive
NUP62	605815	Striatonigral degeneration, infantile, 271930 (3), Autosomal recessive
OAT	613349	Gyrate atrophy of choroid and retina with or without ornithinemia, 258870 (3), Autosomal recessive
OCA2	611409	Albinism, brown oculocutaneous, 203200 (3), Autosomal recessive; Albinism, oculocutaneous, type II, 203200 (3), Autosomal recessive; [Skin/hair/eye pigmentation 1, blond/brown hair], 227220 (3), Autosomal recessive; [Skin/hair/eye pigmentation 1, blue/nonblue eyes], 227220 (3), Autosomal recessive
OCLN	602876	Pseudo-TORCH syndrome 1, 251290 (3), Autosomal recessive
OPA3	606580	3-methylglutaconic aciduria, type III, 258501 (3), Autosomal recessive; Optic atrophy 3 with cataract, 165300 (3), Autosomal dominant
OPTN	602432	Amyotrophic lateral sclerosis 12, 613435 (3); Glaucoma 1, open angle, E, 137760 (3), Autosomal dominant; {Glaucoma, normal tension, susceptibility to}, 606657 (3)
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
OSTM1	607649	Osteopetrosis, autosomal recessive 5, 259720 (3), Autosomal recessive
OTOA	607038	Deafness, autosomal recessive 22, 607039 (3), Autosomal recessive
OTOF	603681	Auditory neuropathy, autosomal recessive, 1, 601071 (3), Autosomal recessive; Deafness, autosomal recessive 9, 601071 (3), Autosomal recessive
OXCT1	601424	Succinyl CoA:3-oxoacid CoA transferase deficiency, 245050 (3)
P3H1	610339	Osteogenesis imperfecta, type VIII, 610915 (3), Autosomal recessive
PAH	612349	[Hyperphenylalaninemia, non-PKU mild], 261600 (3), Autosomal recessive; Phenylketonuria, 261600 (3), Autosomal recessive
PANK2	606157	HARP syndrome, 607236 (3), Autosomal recessive; Neurodegeneration with brain iron accumulation 1, 234200 (3), Autosomal recessive
PAPSS2	603005	Brachyolmia 4 with mild epiphyseal and metaphyseal changes, 612847 (3), Autosomal recessive
PAX3	606597	Craniofacial-deafness-hand syndrome, 122880 (3), Autosomal dominant; Rhabdomyosarcoma 2, alveolar, 268220 (3), Somatic mutation; Waardenburg syndrome, type 1, 193500 (3), Autosomal dominant; Waardenburg syndrome, type 3, 148820 (3), Autosomal recessive, Autosomal dominant

PAX8	167415	Hypothyroidism, congenital, due to thyroid dysgenesis or hypoplasia, 218700 (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
PCDH15	605514	Deafness, autosomal recessive 23, 609533 (3), Autosomal recessive; Usher syndrome, type 1D/F digenic, 601067 (3), Autosomal recessive, Digenic recessive; Usher syndrome, type 1F, 602083 (3), Autosomal recessive
PCNT	605925	Microcephalic osteodysplastic primordial dwarfism, type II, 210720 (3), Autosomal recessive
PDE6A	180071	Retinitis pigmentosa 43, 613810 (3)
PDE6B	180072	Night blindness, congenital stationary, autosomal dominant 2, 163500 (3), Autosomal dominant; Retinitis pigmentosa-40, 613801 (3), Autosomal recessive
PDE6C	600827	Cone dystrophy 4, 613093 (3), Autosomal recessive
PDE6D	602676	?Joubert syndrome 22, 615665 (3), Autosomal recessive
PDE6H	601190	Achromatopsia 6, 610024 (3), Autosomal recessive, Autosomal dominant; Retinal cone dystrophy 3, 610024 (3), Autosomal recessive, Autosomal dominant
PDHB	179060	Pyruvate dehydrogenase E1-beta deficiency, 614111 (3)
PDHX	608769	Lacticacidemia due to PDX1 deficiency, 245349 (3), Autosomal recessive
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; MODY, type IV, 606392 (3); Pancreatic agenesis 1, 260370 (3), Autosomal recessive
PEPD	613230	Prolidase deficiency, 170100 (3), Autosomal recessive
PEX1	602136	Heimler syndrome 1, 234580 (3), Autosomal recessive; Peroxisome biogenesis disorder 1A (Zellweger), 214100 (3), Autosomal recessive; Peroxisome biogenesis disorder 1B (NALD/IRD), 601539 (3), Autosomal recessive
PEX10	602859	Peroxisome biogenesis disorder 6A (Zellweger), 614870 (3), Autosomal recessive; Peroxisome biogenesis disorder 6B, 614871 (3), Autosomal recessive
PEX11B	603867	?Peroxisome biogenesis disorder 14B, 614920 (3), Autosomal recessive

PEX12	601758	Peroxisome biogenesis disorder 3A (Zellweger), 614859 (3), Autosomal recessive; Peroxisome biogenesis disorder 3B, 266510 (3), Autosomal recessive
PEX13	601789	Peroxisome biogenesis disorder 11A (Zellweger), 614883 (3), Autosomal recessive; Peroxisome biogenesis disorder 11B, 614885 (3), Autosomal recessive
PEX14	601791	Peroxisome biogenesis disorder 13A (Zellweger), 614887 (3), Autosomal recessive
PEX16	603360	Peroxisome biogenesis disorder 8A (Zellweger), 614876 (3), Autosomal recessive; Peroxisome biogenesis disorder 8B, 614877 (3), Autosomal recessive
PEX19	600279	Peroxisome biogenesis disorder 12A (Zellweger), 614886 (3), Autosomal recessive
PEX2	170993	Peroxisome biogenesis disorder 5A (Zellweger), 614866 (3), Autosomal recessive; Peroxisome biogenesis disorder 5B, 614867 (3), Autosomal recessive
PEX26	608666	Peroxisome biogenesis disorder 7A (Zellweger), 614872 (3), Autosomal recessive; Peroxisome biogenesis disorder 7B, 614873 (3), Autosomal recessive
PEX3	603164	Peroxisome biogenesis disorder 10A (Zellweger), 614882 (3), Autosomal recessive; ?Peroxisome biogenesis disorder 10B, 617370 (3), Autosomal recessive
PEX5	600414	Peroxisome biogenesis disorder 2A (Zellweger), 214110 (3), Autosomal recessive; Peroxisome biogenesis disorder 2B, 202370 (3), Autosomal recessive; Rhizomelic chondrodysplasia punctata, type 5, 616716 (3), Autosomal recessive
PEX6	601498	Heimler syndrome 2, 616617 (3), Autosomal recessive; Peroxisome biogenesis disorder 4A (Zellweger), 614862 (3), Autosomal recessive; Peroxisome biogenesis disorder 4B, 614863 (3), Autosomal recessive, Autosomal dominant
PEX7	601757	Peroxisome biogenesis disorder 9B, 614879 (3), Autosomal recessive; Rhizomelic chondrodysplasia punctata, type 1, 215100 (3), Autosomal recessive
PFKM	610681	Glycogen storage disease VII, 232800 (3), Autosomal recessive
PGM1	171900	Congenital disorder of glycosylation, type It, 614921 (3), Autosomal recessive
PHGDH	606879	Neu-Laxova syndrome 1, 256520 (3), Autosomal recessive; Phosphoglycerate dehydrogenase deficiency, 601815 (3), Autosomal recessive
PHKB	172490	Phosphorylase kinase deficiency of liver and muscle, autosomal recessive, 261750 (3), Autosomal recessive
PHYH	602026	Refsum disease, 266500 (3), Autosomal recessive

PIEZO2	613629	Arthrogryposis, distal, type 3, 114300 (3), Autosomal dominant; Arthrogryposis, distal, type 5, 108145 (3), Autosomal dominant; Arthrogryposis, distal, with impaired proprioception and touch, 617146 (3), Autosomal recessive; ?Marden-Walker syndrome, 248700 (3), Autosomal dominant
PIGL	605947	CHIME syndrome, 280000 (3), Autosomal recessive
PIGV	610274	Hyperphosphatasia with mental retardation syndrome 1, 239300 (3), Autosomal recessive
PINK1	608309	Parkinson disease 6, early onset, 605909 (3), Autosomal recessive
PIP5K1C	606102	Lethal congenital contractural syndrome 3, 611369 (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
PLA2G6	603604	Infantile neuroaxonal dystrophy 1, 256600 (3), Autosomal recessive; Neurodegeneration with brain iron accumulation 2B, 610217 (3), Autosomal recessive; Parkinson disease 14, autosomal recessive, 612953 (3), Autosomal recessive
PLCB4	600810	Auriculocondylar syndrome 2, 614669 (3), Autosomal recessive, Autosomal dominant
PLCE1	608414	Nephrotic syndrome, type 3, 610725 (3), Autosomal recessive
PLEC	601282	Epidermolysis bullosa simplex with muscular dystrophy, 226670 (3), Autosomal recessive; ?Epidermolysis bullosa simplex with nail dystrophy, 616487 (3), Autosomal recessive; Epidermolysis bullosa simplex with pyloric atresia, 612138 (3), Autosomal recessive; Epidermolysis bullosa simplex, Ogna type, 131950 (3), Autosomal dominant; Muscular dystrophy, limb-girdle, autosomal recessive 17, 613723 (3), Autosomal recessive
PLEKHG5	611101	Charcot-Marie-Tooth disease, recessive intermediate C, 615376 (3), Autosomal recessive; Spinal muscular atrophy, distal, autosomal recessive, 4, 611067 (3), Autosomal recessive
PLG	173350	Dysplasminogenemia, 217090 (3), Autosomal recessive; Plasminogen deficiency, type I, 217090 (3), Autosomal recessive
PLK4	605031	Microcephaly and chorioretinopathy, autosomal recessive, 2, 616171 (3), Autosomal recessive
PLOD1	153454	Ehlers-Danlos syndrome, kyphoscoliotic type, 1, 225400 (3), Autosomal recessive
PLOD3	603066	Lysyl hydroxylase 3 deficiency, 612394 (3), Autosomal recessive
PMM2	601785	Congenital disorder of glycosylation, type Ia, 212065 (3), Autosomal recessive

PMP22	601097	Charcot-Marie-Tooth disease, type 1A, 118220 (3), Autosomal dominant; Charcot-Marie-Tooth disease, type 1E, 118300 (3), Autosomal dominant; Dejerine-Sottas disease, 145900 (3), Autosomal recessive, Autosomal dominant; ?Neuropathy, inflammatory demyelinating, 139393 (3), ?Autosomal dominant; Neuropathy, recurrent, with pressure palsies, 162500 (3), Autosomal dominant; Roussy-Levy syndrome, 180800 (3), Autosomal dominant
PMS2	600259	Colorectal cancer, hereditary nonpolyposis, type 4, 614337 (3); Mismatch repair cancer syndrome, 276300 (3), Autosomal recessive
PNKP	605610	Ataxia-oculomotor apraxia 4, 616267 (3), Autosomal recessive; Microcephaly, seizures, and developmental delay, 613402 (3), Autosomal recessive
PNP	164050	Immunodeficiency due to purine nucleoside phosphorylase deficiency, 613179 (3), Autosomal recessive
PNPLA2	609059	Neutral lipid storage disease with myopathy, 610717 (3), Autosomal recessive
PNPLA6	603197	Boucher-Neuhauser syndrome, 215470 (3), Autosomal recessive; ?Laurence-Moon syndrome, 245800 (3), Autosomal recessive; Oliver-McFarlane syndrome, 275400 (3), Autosomal recessive; Spastic paraplegia 39, autosomal recessive, 612020 (3), Autosomal recessive
PNPO	603287	Pyridoxamine 5'-phosphate oxidase deficiency, 610090 (3), Autosomal recessive
POLG	174763	Mitochondrial DNA depletion syndrome 4A (Alpers type), 203700 (3), Autosomal recessive; Mitochondrial DNA depletion syndrome 4B (MNGIE type), 613662 (3), Autosomal recessive; Mitochondrial recessive ataxia syndrome (includes SANDO and SCAE), 607459 (3), Autosomal recessive; Progressive external ophthalmoplegia, autosomal dominant 1, 157640 (3), Autosomal dominant; Progressive external ophthalmoplegia, autosomal recessive 1, 258450 (3), Autosomal recessive
POLH	603968	Xeroderma pigmentosum, variant type, 278750 (3), Autosomal recessive
POLR1C	610060	Leukodystrophy, hypomyelinating, 11, 616494 (3), Autosomal recessive; Treacher Collins syndrome 3, 248390 (3), Autosomal recessive
POLR3A	614258	Leukodystrophy, hypomyelinating, 7, with or without oligodontia and/or hypogonadotropic hypogonadism, 607694 (3), Autosomal recessive; Wiedemann-Rautenstrauch syndrome, 264090 (3), Autosomal recessive
POLR3B	614366	Leukodystrophy, hypomyelinating, 8, with or without oligodontia and/or hypogonadotropic hypogonadism, 614381 (3), Autosomal recessive
POMC	176830	Obesity, adrenal insufficiency, and red hair due to POMC deficiency, 609734 (3), Autosomal recessive; {Obesity, early-onset, susceptibility to}, 601665 (3), Autosomal recessive, Autosomal dominant, Multifactorial

POMGNT1	606822	Muscular dystrophy-dystroglycanopathy (congenital with brain and eye anomalies), type A, 3, 253280 (3), Autosomal recessive; Muscular dystrophy-dystroglycanopathy (congenital with mental retardation), type B, 3, 613151 (3), Autosomal recessive; Muscular dystrophy-dystroglycanopathy (limb-girdle), type C, 3, 613157 (3), Autosomal recessive; Retinitis pigmentosa 76, 617123 (3), Autosomal recessive
POMGNT2	614828	Muscular dystrophy-dystroglycanopathy (congenital with brain and eye anomalies, type A, 8, 614830 (3), Autosomal recessive; Muscular dystrophy-dystroglycanopathy (limb-girdle) type C, 8, 618135 (3), Autosomal recessive
POMK	615247	Muscular dystrophy-dystroglycanopathy (congenital with brain and eye anomalies), type A, 12, 615249 (3), Autosomal recessive; ?Muscular dystrophy-dystroglycanopathy (limb-girdle), type C, 12, 616094 (3), Autosomal recessive
POMT1	607423	Muscular dystrophy-dystroglycanopathy (congenital with brain and eye anomalies), type A, 1, 236670 (3), Autosomal recessive; Muscular dystrophy-dystroglycanopathy (congenital with mental retardation), type B, 1, 613155 (3), Autosomal recessive; Muscular dystrophy-dystroglycanopathy (limb-girdle), type C, 1, 609308 (3), Autosomal recessive
POMT2	607439	Muscular dystrophy-dystroglycanopathy (congenital with brain and eye anomalies), type A, 2, 613150 (3), Autosomal recessive; Muscular dystrophy-dystroglycanopathy (congenital with mental retardation), type B, 2, 613156 (3), Autosomal recessive; Muscular dystrophy-dystroglycanopathy (limb-girdle), type C, 2, 613158 (3), Autosomal recessive
POR	124015	Antley-Bixler syndrome with genital anomalies and disordered steroidogenesis, 201750 (3), Autosomal recessive; Disordered steroidogenesis due to cytochrome P450 oxidoreductase, 613571 (3)
POU1F1	173110	Pituitary hormone deficiency, combined, 1, 613038 (3), Autosomal recessive, Autosomal dominant
PPIB	123841	Osteogenesis imperfecta, type IX, 259440 (3), Autosomal recessive
PPT1	600722	Ceroid lipofuscinosis, neuronal, 1, 256730 (3), Autosomal recessive
PRDM5	614161	Brittle cornea syndrome 2, 614170 (3), Autosomal recessive
PREPL	609557	Myasthenic syndrome, congenital, 22, 616224 (3), Autosomal recessive
PRF1	170280	Aplastic anemia, 609135 (3); Hemophagocytic lymphohistiocytosis, familial, 2, 603553 (3), Autosomal recessive; Lymphoma, non-Hodgkin, 605027 (3)
PRG4	604283	Camptodactyly-arthropathy-coxa vara-pericarditis syndrome, 208250 (3), Autosomal recessive
PRKN	602544	Adenocarcinoma of lung, somatic, 211980 (3); Ovarian cancer, somatic, 167000 (3); Parkinson disease, juvenile, type 2, 600116 (3), Autosomal recessive

PROC	612283	Thrombophilia due to protein C deficiency, autosomal dominant, 176860 (3), Autosomal dominant; Thrombophilia due to protein C deficiency, autosomal recessive, 612304 (3), Autosomal recessive
PRODH	606810	Hyperprolinemia, type I, 239500 (3), Autosomal recessive; {Schizophrenia, susceptibility to, 4}, 600850 (3), Autosomal dominant
PROM1	604365	Cone-rod dystrophy 12, 612657 (3); Macular dystrophy, retinal, 2, 608051 (3), Autosomal dominant; Retinitis pigmentosa 41, 612095 (3), Autosomal recessive; Stargardt disease 4, 603786 (3)
PROP1	601538	Pituitary hormone deficiency, combined, 2, 262600 (3), Autosomal recessive
PROS1	176880	Thrombophilia due to protein S deficiency, autosomal dominant, 612336 (3), Autosomal dominant; Thrombophilia due to protein S deficiency, autosomal recessive, 614514 (3), Autosomal recessive
PRPH2	179605	Choroidal dystrophy, central areolar 2, 613105 (3), Autosomal dominant; Leber congenital amaurosis 18, 608133 (3), Autosomal recessive, Autosomal dominant; Macular dystrophy, patterned, 1, 169150 (3), Autosomal dominant; Macular dystrophy, vitelliform, 3, 608161 (3), Autosomal dominant; Retinitis pigmentosa 7 and digenic form, 608133 (3), Autosomal recessive, Autosomal dominant; Retinitis punctata albescens, 136880 (3), Autosomal recessive, Autosomal dominant
PRRX1	167420	Agnathia-otocephaly complex, 202650 (3), Autosomal recessive, Autosomal dominant
PRSS56	613858	Microphthalmia, isolated 6, 613517 (3), Autosomal recessive
PRX	605725	Charcot-Marie-Tooth disease, type 4F, 614895 (3), Autosomal recessive; Dejerine-Sottas disease, 145900 (3), Autosomal recessive, Autosomal dominant
PSAP	176801	Combined SAP deficiency, 611721 (3), Autosomal recessive; Gaucher disease, atypical, 610539 (3); Krabbe disease, atypical, 611722 (3), Autosomal recessive; Metachromatic leukodystrophy due to SAP-b deficiency, 249900 (3), Autosomal recessive
PSAT1	610936	Neu-Laxova syndrome 2, 616038 (3), Autosomal recessive; ?Phosphoserine aminotransferase deficiency, 610992 (3), Autosomal recessive
PTEN	601728	Cowden syndrome 1, 158350 (3), Autosomal dominant; {Glioma susceptibility 2}, 613028 (3); Lhermitte-Duclos syndrome, 158350 (3), Autosomal dominant; Macrocephaly/autism syndrome, 605309 (3), Autosomal dominant; {Meningioma}, 607174 (3), Autosomal dominant; Prostate cancer, somatic, 176807 (3)
PTH1R	168468	Chondrodysplasia, Blomstrand type, 215045 (3), Autosomal recessive; Eiken syndrome, 600002 (3), Autosomal recessive; Failure of tooth eruption, primary, 125350 (3), Autosomal dominant; Metaphyseal chondrodysplasia, Murk Jansen type, 156400 (3), Autosomal dominant
PTS	612719	Hyperphenylalaninemia, BH4-deficient, A, 261640 (3), Autosomal recessive

PUS1	608109	Myopathy, lactic acidosis, and sideroblastic anemia 1, 600462 (3), Autosomal recessive
PYCR1	179035	Cutis laxa, autosomal recessive, type IIB, 612940 (3), Autosomal recessive; Cutis laxa, autosomal recessive, type IIIB, 614438 (3)
PYGL	613741	Glycogen storage disease VI, 232700 (3), Autosomal recessive
PYGM	608455	McArdle disease, 232600 (3), Autosomal recessive
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
RAB3GAP1	602536	Warburg micro syndrome 1, 600118 (3), Autosomal recessive
RAB3GAP2	609275	Martsolf syndrome, 212720 (3), Autosomal recessive; Warburg micro syndrome 2, 614225 (3), Autosomal recessive
RAD51C	602774	{Breast-ovarian cancer, familial, susceptibility to, 3}, 613399 (3); Fanconi anemia, complementation group O, 613390 (3), Autosomal recessive
RAG1	179615	Alpha/beta T-cell lymphopenia with gamma/delta T-cell expansion, severe cytomegalovirus infection, and autoimmunity, 609889 (3); Combined cellular and humoral immune defects with granulomas, 233650 (3), Autosomal recessive; Omenn syndrome, 603554 (3), Autosomal recessive; Severe combined immunodeficiency, B cell-negative, 601457 (3), Autosomal recessive
RAG2	179616	Combined cellular and humoral immune defects with granulomas, 233650 (3), Autosomal recessive; Omenn syndrome, 603554 (3), Autosomal recessive; Severe combined immunodeficiency, B cell-negative, 601457 (3), Autosomal recessive
RAPSN	601592	Fetal akinesia deformation sequence 2, 618388 (3); Myasthenic syndrome, congenital, 11, associated with acetylcholine receptor deficiency, 616326 (3), Autosomal recessive
RARS2	611524	Pontocerebellar hypoplasia, type 6, 611523 (3), Autosomal recessive
RAX	601881	Microphthalmia, isolated 3, 611038 (3), Autosomal recessive
RBM8A	605313	Thrombocytopenia-absent radius syndrome, 274000 (3), Autosomal recessive
RD3	180040	Leber congenital amaurosis 12, 610612 (3), Autosomal recessive
RDH12	608830	Leber congenital amaurosis 13, 612712 (3), Autosomal recessive, Autosomal dominant
RECQL4	603780	Baller-Gerold syndrome, 218600 (3), Autosomal recessive; RAPADILINO syndrome, 266280 (3), Autosomal recessive; Rothmund-Thomson syndrome, 268400 (3), Autosomal recessive
RELN	600514	{Epilepsy, familial temporal lobe, 7}, 616436 (3), Autosomal dominant; Lissencephaly 2 (Norman-Roberts type), 257320 (3), Autosomal recessive
REN	179820	[Hyperproreninemia] (3); Hyperuricemic nephropathy, familial juvenile 2, 613092 (3), Autosomal dominant; Renal tubular dysgenesis, 267430 (3), Autosomal recessive

RFT1	611908	Congenital disorder of glycosylation, type In, 612015 (3), Autosomal recessive
RFX6	612659	Mitchell-Riley syndrome, 615710 (3), Autosomal recessive
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
RIPK4	605706	CHAND syndrome, 214350 (3), Autosomal recessive; Popliteal pterygium syndrome, Bartsocas-Papas type, 263650 (3), Autosomal recessive
RLBP1	180090	Bothnia retinal dystrophy, 607475 (3), Autosomal recessive; Fundus albipunctatus, 136880 (3), Autosomal recessive, Autosomal dominant; Newfoundland rod-cone dystrophy, 607476 (3); Retinitis punctata albescens, 136880 (3), Autosomal recessive, Autosomal dominant
RMND1	614917	Combined oxidative phosphorylation deficiency 11, 614922 (3), Autosomal recessive
RMRP	157660	Anauxetic dysplasia 1, 607095 (3), Autosomal recessive; Cartilage-hair hypoplasia, 250250 (3), Autosomal recessive; Metaphyseal dysplasia without hypotrichosis, 250460 (3), Autosomal recessive
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
RNU4ATAC	601428	Microcephalic osteodysplastic primordial dwarfism, type I, 210710 (3), Autosomal recessive; Roifman syndrome, 616651 (3), Autosomal recessive
ROGDI	614574	Kohlschutter-Tonz syndrome, 226750 (3), Autosomal recessive
ROR2	602337	Brachydactyly, type B1, 113000 (3), Autosomal dominant; Robinow syndrome, autosomal recessive, 268310 (3), Autosomal recessive
RP1	603937	Retinitis pigmentosa 1, 180100 (3), Autosomal recessive, Autosomal dominant
RPE65	180069	Leber congenital amaurosis 2, 204100 (3), Autosomal recessive; Retinitis pigmentosa 20, 613794 (3), Autosomal recessive
RPGRIP1	605446	Cone-rod dystrophy 13, 608194 (3); Leber congenital amaurosis 6, 613826 (3), Autosomal recessive
RPGRIP1L	610937	COACH syndrome, 216360 (3), Autosomal recessive; Joubert syndrome 7, 611560 (3), Autosomal recessive; Meckel syndrome 5, 611561 (3), Autosomal recessive
RRM2B	604712	Mitochondrial DNA depletion syndrome 8A (encephalomyopathic type with renal tubulopathy), 612075 (3), Autosomal recessive; Mitochondrial DNA depletion syndrome 8B (MNGIE type), 612075 (3), Autosomal recessive; Progressive external ophthalmoplegia with mitochondrial DNA deletions, autosomal dominant 5, 613077 (3), Autosomal dominant
RSPH4A	612647	Ciliary dyskinesia, primary, 11, 612649 (3)

RTEL1	608833	Dyskeratosis congenita, autosomal dominant 4, 615190 (3), Autosomal recessive, Autosomal dominant; Dyskeratosis congenita, autosomal recessive 5, 615190 (3), Autosomal recessive, Autosomal dominant; Pulmonary fibrosis and/or bone marrow failure, telomere-related, 3, 616373 (3), Autosomal dominant
RTTN	610436	Microcephaly, short stature, and polymicrogyria with seizures, 614833 (3), Autosomal recessive
RXYLT1	605862	Muscular dystrophy-dystroglycanopathy (congenital with brain and eye anomalies), type A, 10, 615041 (3), Autosomal recessive
RYR1	180901	Central core disease, 117000 (3), Autosomal recessive, Autosomal dominant; King-Denborough syndrome, 145600 (3), Autosomal dominant; {Malignant hyperthermia susceptibility 1}, 145600 (3), Autosomal dominant; Minicore myopathy with external ophthalmoplegia, 255320 (3), Autosomal recessive; Neuromuscular disease, congenital, with uniform type 1 fiber, 117000 (3), Autosomal recessive, Autosomal dominant
SACS	604490	Spastic ataxia, Charlevoix-Saguenay type, 270550 (3), Autosomal recessive
SAMHD1	606754	Aicardi-Goutieres syndrome 5, 612952 (3), Autosomal recessive; ?Chilblain lupus 2, 614415 (3), Autosomal dominant
SBDS	607444	{Aplastic anemia, susceptibility to}, 609135 (3); Shwachman-Diamond syndrome, 260400 (3), Autosomal recessive
SC5D	602286	Lathosterolosis, 607330 (3), Autosomal recessive
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
SCN4A	603967	Hyperkalemic periodic paralysis, type 2, 170500 (3), Autosomal dominant; Hypokalemic periodic paralysis, type 2, 613345 (3), Autosomal dominant; Myasthenic syndrome, congenital, 16, 614198 (3), Autosomal recessive; Myotonia congenita, atypical, acetazolamide-responsive, 608390 (3), Autosomal dominant; Paramyotonia congenita, 168300 (3), Autosomal dominant
SCN5A	600163	Atrial fibrillation, familial, 10, 614022 (3), Autosomal dominant; Brugada syndrome 1, 601144 (3), Autosomal dominant; Cardiomyopathy, dilated, 1E, 601154 (3), Autosomal dominant; Heart block, nonprogressive, 113900 (3), Autosomal dominant; Heart block, progressive, type IA, 113900 (3), Autosomal dominant; Long QT syndrome-3, 603830 (3), Autosomal dominant; Sick sinus syndrome 1, 608567 (3), Autosomal recessive; {Sudden infant death syndrome, susceptibility to}, 272120 (3), Autosomal recessive; Ventricular fibrillation, familial, 1, 603829 (3)

SCN9A	603415	{Dravet syndrome, modifier of}, 607208 (3), Autosomal dominant; Epilepsy, generalized, with febrile seizures plus, type 7, 613863 (3), Autosomal dominant; Erythermalgia, primary, 133020 (3), Autosomal dominant; Febrile seizures, familial, 3B, 613863 (3), Autosomal dominant; HSAN2D, autosomal recessive, 243000 (3), Autosomal recessive; Insensitivity to pain, congenital, 243000 (3), Autosomal recessive; Paroxysmal extreme pain disorder, 167400 (3), Autosomal dominant; Small fiber neuropathy, 133020 (3), Autosomal dominant
SCNN1A	600228	Bronchiectasis with or without elevated sweat chloride 2, 613021 (3), Autosomal dominant; ?Liddle syndrome 3, 618126 (3), Autosomal dominant; Pseudohypoaldosteronism, type I, 264350 (3), Autosomal recessive
SCNN1B	600760	Bronchiectasis with or without elevated sweat chloride 1, 211400 (3), Autosomal dominant; Liddle syndrome 1, 177200 (3), Autosomal dominant; Pseudohypoaldosteronism, type I, 264350 (3), Autosomal recessive
SCNN1G	600761	Bronchiectasis with or without elevated sweat chloride 3, 613071 (3), Autosomal dominant; Liddle syndrome 2, 618114 (3), Autosomal dominant; Pseudohypoaldosteronism, type I, 264350 (3), Autosomal recessive
SCO1	603644	Mitochondrial complex IV deficiency, 220110 (3), Autosomal recessive, Mitochondrial
SCO2	604272	Cardioencephalomyopathy, fatal infantile, due to cytochrome c oxidase deficiency 1, 604377 (3), Autosomal recessive; Myopia 6, 608908 (3), Autosomal dominant
SDCCAG8	613524	Bardet-Biedl syndrome 16, 615993 (3), Autosomal recessive; Senior-Loken syndrome 7, 613615 (3)
SDHA	600857	Cardiomyopathy, dilated, 1GG, 613642 (3); Leigh syndrome, 256000 (3), Autosomal recessive, Mitochondrial; Mitochondrial respiratory chain complex II deficiency, 252011 (3), Autosomal recessive; Paragangliomas 5, 614165 (3), Autosomal dominant
SDHD	602690	Mitochondrial complex II deficiency, 252011 (3), Autosomal recessive; Paraganglioma and gastric stromal sarcoma, 606864 (3); Paragangliomas 1, with or without deafness, 168000 (3), Autosomal dominant; Pheochromocytoma, 171300 (3), Autosomal dominant
SEC23A	610511	Craniolenticulosutural dysplasia, 607812 (3), Autosomal recessive
SEC23B	610512	?Cowden syndrome 7, 616858 (3), Autosomal dominant; Dyserythropoietic anemia, congenital, type II, 224100 (3), Autosomal recessive
SELENON	606210	Muscular dystrophy, rigid spine, 1, 602771 (3), Autosomal recessive; Myopathy, congenital, with fiber-type disproportion, 255310 (3), Autosomal recessive, Autosomal dominant
SEPSECS	613009	Pontocerebellar hypoplasia type 2D, 613811 (3), Autosomal recessive

SERPINA1	107400	Emphysema due to AAT deficiency, 613490 (3), Autosomal recessive; Emphysema-cirrhosis, due to AAT deficiency, 613490 (3), Autosomal recessive; Hemorrhagic diathesis due to antithrombin Pittsburgh, 613490 (3), Autosomal recessive; {Pulmonary disease, chronic obstructive, susceptibility to}, 606963 (1)
SERPINF1	172860	Osteogenesis imperfecta, type VI, 613982 (3)
SETX	608465	Amyotrophic lateral sclerosis 4, juvenile, 602433 (3), Autosomal dominant; Spinocerebellar ataxia, autosomal recessive, with axonal neuropathy 2, 606002 (3), Autosomal recessive
SFTPB	178640	Surfactant metabolism dysfunction, pulmonary, 1, 265120 (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
SGCG	608896	Muscular dystrophy, limb-girdle, autosomal recessive 5, 253700 (3), Autosomal 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
SI	609845	Sucrase-isomaltase deficiency, congenital, 222900 (3), Autosomal recessive
SIL1	608005	Marinesco-Sjogren syndrome, 248800 (3), Autosomal recessive
SIX6	606326	Optic disc anomalies with retinal and/or macular dystrophy, 212550 (3), Autosomal recessive
SLC12A1	600839	Bartter syndrome, type 1, 601678 (3), Autosomal recessive
SLC12A3	600968	Gitelman syndrome, 263800 (3), Autosomal recessive
SLC12A6	604878	Agenesis of the corpus callosum with peripheral neuropathy, 218000 (3), Autosomal recessive
SLC16A1	600682	Erythrocyte lactate transporter defect, 245340 (3), Autosomal dominant; Hyperinsulinemic hypoglycemia, familial, 7, 610021 (3), Autosomal dominant; Monocarboxylate transporter 1 deficiency, 616095 (3), Autosomal recessive, Autosomal dominant
SLC17A5	604322	Salla disease, 604369 (3), Autosomal recessive; Sialic acid storage disorder, infantile, 269920 (3), Autosomal recessive
SLC19A3	606152	Thiamine metabolism dysfunction syndrome 2 (biotin- or thiamine-responsive encephalopathy type 2), 607483 (3), Autosomal recessive
SLC22A12	607096	Hypouricemia, renal, 220150 (3), Autosomal recessive
SLC22A5	603377	Carnitine deficiency, systemic primary, 212140 (3), Autosomal recessive

SLC25A13	603859	Citrullinemia, adult-onset type II, 603471 (3), Autosomal recessive; Citrullinemia, type II, neonatal-onset, 605814 (3), Autosomal recessive
SLC25A15	603861	Hyperornithinemia-hyperammonemia-homocitrullinemia syndrome, 238970 (3), Autosomal recessive
SLC25A19	606521	Microcephaly, Amish type, 607196 (3), Autosomal recessive; Thiamine metabolism dysfunction syndrome 4 (progressive polyneuropathy type), 613710 (3), Autosomal recessive
SLC25A20	613698	Carnitine-acylcarnitine translocase deficiency, 212138 (3), Autosomal recessive
SLC25A22	609302	Epileptic encephalopathy, early infantile, 3, 609304 (3), Autosomal recessive
SLC25A4	103220	Mitochondrial DNA depletion syndrome 12A (cardiomyopathic type) AD, 617184 (3), Autosomal dominant; Mitochondrial DNA depletion syndrome 12B (cardiomyopathic type) AR, 615418 (3), Autosomal recessive; Progressive external ophthalmoplegia with mitochondrial DNA deletions, autosomal dominant 2, 609283 (3), Autosomal dominant
SLC26A2	606718	Achondrogenesis Ib, 600972 (3), Autosomal recessive; Atelosteogenesis, type II, 256050 (3), Autosomal recessive; De la Chapelle dysplasia, 256050 (3), Autosomal recessive; Diastrophic dysplasia, 222600 (3), Autosomal recessive; Diastrophic dysplasia, broad bone-platyspondylic variant, 222600 (3), Autosomal recessive; Epiphyseal dysplasia, multiple, 4, 226900 (3), Autosomal recessive
SLC26A3	126650	Diarrhea 1, secretory chloride, congenital, 214700 (3), Autosomal recessive
SLC26A4	605646	Deafness, autosomal recessive 4, with enlarged vestibular aqueduct, 600791 (3), Autosomal recessive; Pendred syndrome, 274600 (3), Autosomal recessive
SLC27A4	604194	Ichthyosis prematurity syndrome, 608649 (3)
SLC2A1	138140	Dystonia 9, 601042 (3), Autosomal dominant; {Epilepsy, idiopathic generalized, susceptibility to, 12}, 614847 (3), Autosomal dominant; GLUT1 deficiency syndrome 1, infantile onset, severe, 606777 (3), Autosomal recessive, Autosomal dominant; GLUT1 deficiency syndrome 2, childhood onset, 612126 (3), Autosomal dominant; Stomatin-deficient cryohydrocytosis with neurologic defects, 608885 (3), Autosomal dominant
SLC2A10	606145	Arterial tortuosity syndrome, 208050 (3), Autosomal recessive
SLC2A2	138160	{Diabetes mellitus, noninsulin-dependent}, 125853 (3), Autosomal dominant; Fanconi-Bickel syndrome, 227810 (3), Autosomal recessive
SLC2A9	606142	Hypouricemia, renal, 2, 612076 (3), Autosomal recessive, Autosomal dominant; {Uric acid concentration, serum, QTL 2}, 612076 (3), Autosomal recessive, Autosomal dominant
SLC30A10	611146	Hypermanganesemia with dystonia 1, 613280 (3), Autosomal recessive

SLC34A2	604217	Pulmonary alveolar microlithiasis, 265100 (3), Autosomal recessive
SLC35A1	605634	Congenital disorder of glycosylation, type IIf, 603585 (3), Autosomal recessive
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
SLC37A4	602671	Glycogen storage disease Ib, 232220 (3), Autosomal recessive; Glycogen storage disease Ic, 232240 (3), Autosomal recessive
SLC39A4	607059	Acrodermatitis enteropathica, 201100 (3), Autosomal recessive
SLC3A1	104614	Cystinuria, 220100 (3), Autosomal recessive, Autosomal dominant
SLC45A2	606202	Albinism, oculocutaneous, type IV, 606574 (3); [Skin/hair/eye pigmentation 5, black/nonblack hair], 227240 (3), Autosomal recessive; [Skin/hair/eye pigmentation 5, dark/fair skin], 227240 (3), Autosomal recessive; [Skin/hair/eye pigmentation 5, dark/light eyes], 227240 (3), Autosomal recessive
SLC46A1	611672	Folate malabsorption, hereditary, 229050 (3), Autosomal recessive
SLC4A1	109270	[Blood group, Diego], 110500 (3); [Blood group, Froese], 601551 (3); [Blood group, Swann], 601550 (3); [Blood group, Waldner], 112010 (3); [Blood group, Wright], 112050 (3); Cryohydrocytosis, 185020 (3), Autosomal dominant; [Malaria, resistance to], 611162 (3); Ovalocytosis, SA type, 166900 (3), Autosomal dominant; Renal tubular acidosis, distal, AD, 179800 (3), Autosomal dominant; Renal tubular acidosis, distal, AR, 611590 (3), Autosomal recessive; Spherocytosis, type 4, 612653 (3), Autosomal dominant
SLC4A11	610206	Corneal dystrophy, Fuchs endothelial, 4, 613268 (3); Corneal endothelial dystrophy and perceptive deafness, 217400 (3), Autosomal recessive; Corneal endothelial dystrophy, autosomal recessive, 217700 (3), Autosomal recessive
SLC52A3	613350	Brown-Vialetto-Van Laere syndrome 1, 211530 (3), Autosomal recessive; ?Fazio-Londe disease, 211500 (3), Autosomal recessive
SLC5A5	601843	Thyroid dyshormonogenesis 1, 274400 (3), Autosomal recessive
SLC6A3	126455	{Nicotine dependence, protection against}, 188890 (3); Parkinsonism-dystonia, infantile, 1, 613135 (3), Autosomal recessive
SLC6A5	604159	Hyperekplexia 3, 614618 (3), Autosomal recessive, Autosomal dominant
SLC7A7	603593	Lysinuric protein intolerance, 222700 (3), Autosomal recessive
SLC7A9	604144	Cystinuria, 220100 (3), Autosomal recessive, Autosomal dominant
SLX4	613278	Fanconi anemia, complementation group P, 613951 (3), Autosomal recessive
SMARCAL1	606622	Schimke immunoosseous dysplasia, 242900 (3), Autosomal recessive

SMN1	600354	Spinal muscular atrophy-1, 253300 (3), Autosomal recessive; Spinal muscular atrophy-2, 253550 (3), Autosomal recessive; Spinal muscular atrophy-3, 253400 (3), Autosomal recessive; Spinal muscular atrophy-4, 271150 (3), Autosomal recessive
SMN2	601627	{Spinal muscular atrophy, type III, modifier of}, 253400 (3), Autosomal recessive
SMOC1	608488	Microphthalmia with limb anomalies, 206920 (3), Autosomal recessive
SMPD1	607608	Niemann-Pick disease, type A, 257200 (3), Autosomal recessive; Niemann-Pick disease, type B, 607616 (3), Autosomal recessive
SNAP29	604202	Cerebral dysgenesis, neuropathy, ichthyosis, and palmoplantar keratoderma syndrome, 609528 (3), Autosomal recessive
SNIP1	608241	Psychomotor retardation, epilepsy, and craniofacial dysmorphism, 614501 (3), Autosomal recessive
SOD1	147450	Amyotrophic lateral sclerosis 1, 105400 (3), Autosomal recessive, Autosomal dominant; Spastic tetraplegia and axial hypotonia, progressive, 618598 (3)
SOX10	602229	PCWH syndrome, 609136 (3), Autosomal dominant; Waardenburg syndrome, type 2E, with or without neurologic involvement, 611584 (3), Autosomal dominant; Waardenburg syndrome, type 4C, 613266 (3), Autosomal dominant
SP110	604457	Hepatic venoocclusive disease with immunodeficiency, 235550 (3), Autosomal recessive; {Mycobacterium tuberculosis, susceptibility to}, 607948 (3)
SPATA7	609868	Leber congenital amaurosis 3, 604232 (3); Retinitis pigmentosa, juvenile, autosomal recessive, 604232 (3)
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
SPG7	602783	Spastic paraplegia 7, autosomal recessive, 607259 (3), Autosomal recessive, Autosomal dominant
SPINT2	605124	Diarrhea 3, secretory sodium, congenital, syndromic, 270420 (3), Autosomal recessive
SPR	182125	Dystonia, dopa-responsive, due to sepiapterin reductase deficiency, 612716 (3), ?Autosomal dominant, Autosomal recessive
SPTBN2	604985	Spinocerebellar ataxia 5, 600224 (3), Autosomal dominant; Spinocerebellar ataxia, autosomal recessive 14, 615386 (3), Autosomal recessive
SRD5A2	607306	Pseudovaginal perineoscrotal hypospadias, 264600 (3), Autosomal recessive
SRD5A3	611715	Congenital disorder of glycosylation, type Iq, 612379 (3), Autosomal recessive; Kahrizi syndrome, 612713 (3), Autosomal recessive

ST3GAL3	606494	?Epileptic encephalopathy, early infantile, 15, 615006 (3), Autosomal recessive; Mental retardation, 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
STAMBP	606247	Microcephaly-capillary malformation syndrome, 614261 (3), Autosomal recessive
STAR	600617	Lipoid adrenal hyperplasia, 201710 (3), Autosomal recessive
STAT1	600555	Immunodeficiency 31A, mycobacteriosis, autosomal dominant, 614892 (3), Autosomal dominant; Immunodeficiency 31B, mycobacterial and viral infections, autosomal recessive, 613796 (3), Autosomal recessive; Immunodeficiency 31C, autosomal dominant, 614162 (3), Autosomal dominant
STIL	181590	Microcephaly 7, primary, autosomal recessive, 612703 (3), Autosomal recessive
STIM1	605921	Immunodeficiency 10, 612783 (3), Autosomal recessive; Myopathy, tubular aggregate, 1, 160565 (3), Autosomal dominant; Stormorken syndrome, 185070 (3), Autosomal dominant
STRA6	610745	Microphthalmia, isolated, with coloboma 8, 601186 (3), Autosomal recessive; Microphthalmia, syndromic 9, 601186 (3), Autosomal recessive
STX11	605014	Hemophagocytic lymphohistiocytosis, familial, 4, 603552 (3), Autosomal recessive
STXBP2	601717	Hemophagocytic lymphohistiocytosis, familial, 5, 613101 (3)
SUCLA2	603921	Mitochondrial DNA depletion syndrome 5 (encephalomyopathic with or without methylmalonic aciduria), 612073 (3), Autosomal recessive
SUCLG1	611224	Mitochondrial DNA depletion syndrome 9 (encephalomyopathic type with methylmalonic aciduria), 245400 (3), Autosomal recessive
SUMF1	607939	Multiple sulfatase deficiency, 272200 (3), Autosomal recessive
SUOX	606887	Sulfite oxidase deficiency, 272300 (3), Autosomal recessive
SURF1	185620	Charcot-Marie-Tooth disease, type 4K, 616684 (3), Autosomal recessive; Leigh syndrome, due to COX IV deficiency, 256000 (3), Autosomal recessive, Mitochondrial
SYNE1	608441	Arthrogryposis multiplex congenita, 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
TACR3	162332	Hypogonadotropic hypogonadism 11 with or without anosmia, 614840 (3), Autosomal recessive
TACSTD2	137290	Corneal dystrophy, gelatinous drop-like, 204870 (3), Autosomal recessive
TAF2	604912	Mental retardation, autosomal recessive 40, 615599 (3), Autosomal recessive

TAT	613018	Tyrosinemia, type II, 276600 (3), Autosomal recessive
TBC1D20	611663	Warburg micro syndrome 4, 615663 (3), Autosomal recessive
TBC1D24	613577	DOORS syndrome, 220500 (3), Autosomal recessive; Deafness , autosomal recessive 86, 614617 (3), Autosomal recessive; Deafness, autosomal dominant 65, 616044 (3), Autosomal dominant; Epilepsy, rolandic, with proxysmal exercise-induce dystonia and writer's cramp, 608105 (3), Autosomal recessive; Epileptic encephalopathy, early infantile, 16, 615338 (3), Autosomal recessive; Myoclonic epilepsy, infantile, familial, 605021 (3), Autosomal recessive
TBCE	604934	Encephalopathy, progressive, with amyotrophy and optic atrophy, 617207 (3), Autosomal recessive; Hypoparathyroidism-retardation-dysmorphism syndrome, 241410 (3), Autosomal recessive; Kenny-Caffey syndrome, type 1, 244460 (3), Autosomal recessive
TBX15	604127	Cousin syndrome, 260660 (3), Autosomal recessive
TBX6	602427	Spondylocostal dysostosis 5, 122600 (3), Autosomal recessive, Autosomal dominant
TCAP	604488	Cardiomyopathy, hypertrophic, 25, 607487 (3), Autosomal dominant; Muscular dystrophy, limb-girdle, autosomal recessive 7, 601954 (3), Autosomal recessive
TCIRG1	604592	Osteopetrosis, autosomal recessive 1, 259700 (3), Autosomal recessive
TCTN1	609863	Joubert syndrome 13, 614173 (3), Autosomal recessive
TCTN2	613846	Joubert syndrome 24, 616654 (3), Autosomal recessive; ?Meckel syndrome 8, 613885 (3), Autosomal recessive
TCTN3	613847	Joubert syndrome 18, 614815 (3), Autosomal recessive; Orofaciodigital syndrome IV, 258860 (3), Autosomal recessive
TECPR2	615000	Spastic paraplegia 49, autosomal recessive, 615031 (3), Autosomal recessive
TECTA	602574	Deafness, autosomal dominant 8/12, 601543 (3), Autosomal dominant; Deafness, autosomal recessive 21, 603629 (3), Autosomal recessive
TERT	187270	{Dyskeratosis congenita, autosomal dominant 2}, 613989 (3), Autosomal recessive, Autosomal dominant; {Dyskeratosis congenita, autosomal recessive 4}, 613989 (3), Autosomal recessive, Autosomal dominant; {Leukemia, acute myeloid}, 601626 (3), Autosomal dominant, Somatic mutation; {Melanoma, cutaneous malignant, 9}, 615134 (3); {Pulmonary fibrosis and/or bone marrow failure, telomere-related, 1}, 614742 (3), Autosomal dominant
TFR2	604720	Hemochromatosis, type 3, 604250 (3), Autosomal recessive
TG	188450	{Autoimmune thyroid disease, susceptibility to, 3}, 608175 (3); Thyroid dyshormonogenesis 3, 274700 (3), Autosomal recessive
TGM1	190195	Ichthyosis, congenital, autosomal recessive 1, 242300 (3), Autosomal recessive
TH	191290	Segawa syndrome, recessive, 605407 (3), Autosomal recessive

TJP2	607709	Cholestasis, progressive familial intrahepatic 4, 615878 (3), Autosomal recessive; Hypercholanemia, familial, 607748 (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
TMC1	606706	Deafness, autosomal dominant 36, 606705 (3), Autosomal dominant; Deafness, autosomal recessive 7, 600974 (3), Autosomal recessive
TMCO1	614123	Craniofacial dysmorphism, skeletal anomalies, and mental retardation syndrome, 213980 (3), Autosomal recessive
TMEM138	614459	Joubert syndrome 16, 614465 (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
TMEM67	609884	{Bardet-Biedl syndrome 14, modifier of}, 615991 (3), Autosomal recessive; COACH syndrome, 216360 (3), Autosomal recessive; Joubert syndrome 6, 610688 (3), Autosomal recessive; Meckel syndrome 3, 607361 (3), Autosomal recessive; Nephronophthisis 11, 613550 (3), Autosomal recessive; ?RHYNS syndrome, 602152 (3), Autosomal recessive
TMEM70	612418	Mitochondrial complex V (ATP synthase) deficiency, nuclear type 2, 614052 (3), Autosomal recessive
TMIE	607237	Deafness, autosomal recessive 6, 600971 (3), Autosomal recessive
TMPRSS3	605511	Deafness, autosomal recessive 8/10, 601072 (3), Autosomal recessive
TNFRSF11A	603499	Osteolysis, familial expansile, 174810 (3), Autosomal dominant; Osteopetrosis, autosomal recessive 7, 612301 (3), Autosomal recessive; {Paget disease of bone 2, early-onset}, 602080 (3), Autosomal dominant
TNFRSF11B	602643	Paget disease of bone 5, juvenile-onset, 239000 (3), Autosomal recessive
TNFRSF13B	604907	Immunodeficiency, common variable, 2, 240500 (3), Autosomal recessive, Autosomal dominant; Immunoglobulin A deficiency 2, 609529 (3)
TNNT1	191041	Nemaline myopathy 5, Amish type, 605355 (3), Autosomal recessive
TNXB	600985	Ehlers-Danlos syndrome, classic-like, 1, 606408 (3), Autosomal recessive; Vesicoureteral reflux 8, 615963 (3), Autosomal dominant
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

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
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
TRAPPC9	611966	Mental retardation, autosomal recessive 13, 613192 (3), Autosomal recessive
TREM2	605086	Polycystic lipomembranous osteodysplasia with sclerosing leukoencephalopathy 2, 618193 (3)
TREX1	606609	Aicardi-Goutieres syndrome 1, dominant and recessive, 225750 (3), Autosomal recessive, Autosomal dominant; Chilblain lupus, 610448 (3), Autosomal dominant; {Systemic lupus erythematosus, susceptibility to}, 152700 (3), Autosomal dominant; Vasculopathy, retinal, with cerebral leukodystrophy, 192315 (3), Autosomal dominant
TRIM32	602290	?Bardet-Biedl syndrome 11, 615988 (3), Autosomal recessive; Muscular dystrophy, limb-girdle, autosomal recessive 8, 254110 (3), Autosomal recessive
TRIM37	605073	Mulibrey nanism, 253250 (3), Autosomal recessive
TRIOBP	609761	Deafness, autosomal recessive 28, 609823 (3), Autosomal recessive
TRIP11	604505	Achondrogenesis, type IA, 200600 (3), Autosomal recessive; Osteochondrodysplasia, 184260 (3), Autosomal recessive
TRMU	610230	{Deafness, mitochondrial, modifier of}, 580000 (3), Mitochondrial; Liver failure, transient infantile, 613070 (3), Autosomal recessive
TRPM6	607009	Hypomagnesemia 1, intestinal, 602014 (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
TSHB	188540	Hypothyroidism, congenital, nongoitrous 4, 275100 (3), Autosomal recessive
TSHR	603372	Hyperthyroidism, familial gestational, 603373 (3); Hyperthyroidism, nonautoimmune, 609152 (3), Autosomal dominant; Hypothyroidism, congenital, nongoitrous, 1, 275200 (3), Autosomal recessive; Thyroid adenoma, hyperfunctioning, somatic (3); Thyroid carcinoma with thyrotoxicosis (3)

TSPYL1	604714	Sudden infant death with dysgenesis of the testes syndrome, 608800 (3), Autosomal recessive
TTC19	613814	Mitochondrial complex III deficiency, nuclear type 2, 615157 (3), Autosomal recessive
TTC21B	612014	Nephronophthisis 12, 613820 (3), Autosomal recessive, Autosomal dominant; Short-rib thoracic dysplasia 4 with or without polydactyly, 613819 (3), Autosomal recessive
TTC8	608132	Bardet-Biedl syndrome 8, 615985 (3), Autosomal recessive; ?Retinitis pigmentosa 51, 613464 (3), Autosomal recessive
TTPA	600415	Ataxia with isolated vitamin E deficiency, 277460 (3), Autosomal recessive
TUBA8	605742	Cortical dysplasia, complex, with other brain malformations 8, 613180 (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
TWNK	606075	Mitochondrial DNA depletion syndrome 7 (hepatocerebral type), 271245 (3), Autosomal recessive; Perrault syndrome 5, 616138 (3), Autosomal recessive; Progressive external ophthalmoplegia with mitochondrial DNA deletions, autosomal dominant 3, 609286 (3), Autosomal dominant
TYK2	176941	Immunodeficiency 35, 611521 (3), Autosomal recessive
TYMP	131222	Mitochondrial DNA depletion syndrome 1 (MNGIE type), 603041 (3), Autosomal recessive
TYR	606933	Albinism, oculocutaneous, type IA, 203100 (3), Autosomal recessive; Albinism, oculocutaneous, type IB, 606952 (3); {Melanoma, cutaneous malignant, susceptibility to, 8}, 601800 (3), Autosomal dominant; [Skin/hair/eye pigmentation 3, blue/green eyes], 601800 (3), Autosomal dominant; [Skin/hair/eye pigmentation 3, light/dark/freckling skin], 601800 (3), Autosomal dominant; Waardenburg syndrome/albinism, digenic, 103470 (3), Autosomal dominant
TYROBP	604142	Polycystic lipomembranous osteodysplasia with sclerosing leukoencephalopathy 1, 221770 (3), Autosomal recessive
UBE3B	608047	Kaufman oculocerebrofacial syndrome, 244450 (3), Autosomal recessive
UBR1	605981	Johanson-Blizzard syndrome, 243800 (3), Autosomal recessive

UGT1A1	191740	[Bilirubin, serum level of, QTL1], 601816 (3); Crigler-Najjar syndrome, type I, 218800 (3), Autosomal recessive; Crigler-Najjar syndrome, type II, 606785 (3), Autosomal recessive; [Gilbert syndrome], 143500 (3), Autosomal recessive; Hyperbilirubinemia, familial transient neonatal, 237900 (3), Autosomal recessive
UNC13D	608897	Hemophagocytic lymphohistiocytosis, familial, 3, 608898 (3)
UQCRB	191330	Mitochondrial complex III deficiency, nuclear type 3, 615158 (3), Autosomal recessive
UQCRQ	612080	Mitochondrial complex III deficiency, nuclear type 4, 615159 (3), Autosomal recessive
UROS	606938	Porphyria, congenital erythropoietic, 263700 (3), Autosomal recessive
USH1C	605242	Deafness, autosomal recessive 18A, 602092 (3), Autosomal recessive; Usher syndrome, type 1C, 276904 (3), Autosomal recessive
USH1G	607696	Usher syndrome, type 1G, 606943 (3), Autosomal recessive
USH2A	608400	Retinitis pigmentosa 39, 613809 (3); Usher syndrome, type 2A, 276901 (3), Autosomal recessive
VAX1	604294	?Microphthalmia, syndromic 11, 614402 (3), Autosomal recessive
VDR	601769	?Osteoporosis, involutional, 166710 (1), Autosomal dominant; Rickets, vitamin D-resistant, type IIA, 277440 (3), Autosomal recessive
VHL	608537	Erythrocytosis, familial, 2, 263400 (3), Autosomal recessive; Hemangioblastoma, cerebellar, somatic (3); Pheochromocytoma, 171300 (3), Autosomal dominant; Renal cell carcinoma, somatic, 144700 (3); von Hippel-Lindau syndrome, 193300 (3), Autosomal dominant
VIPAS39	613401	Arthrogryposis, renal dysfunction, and cholestasis 2, 613404 (3), Autosomal recessive
VLDLR	192977	Cerebellar hypoplasia and mental retardation with or without quadrupedal locomotion 1, 224050 (3), Autosomal recessive
VPS13A	605978	Choreoacanthocytosis, 200150 (3), Autosomal recessive
VPS13B	607817	Cohen syndrome, 216550 (3), Autosomal recessive
VPS33B	608552	Arthrogryposis, renal dysfunction, and cholestasis 1, 208085 (3), Autosomal recessive
VPS45	610035	Neutropenia, severe congenital, 5, autosomal recessive, 615285 (3), Autosomal recessive
VRK1	602168	Pontocerebellar hypoplasia type 1A, 607596 (3), Autosomal recessive
VSX2	142993	Microphthalmia with coloboma 3, 610092 (3); Microphthalmia, isolated 2, 610093 (3)
VWF	613160	von Willebrand disease, type 1, 193400 (3), Autosomal dominant; von Willebrand disease, types 2A, 2B, 2M, and 2N, 613554 (3), Autosomal recessive, Autosomal dominant; von Willibrand disease, type 3, 277480 (3), Autosomal recessive

WASHC5	610657	Ritscher-Schinzel syndrome 1, 220210 (3), Autosomal recessive; Spastic paraplegia 8, autosomal dominant, 603563 (3), Autosomal dominant
WDPCP	613580	?Bardet-Biedl syndrome 15, 615992 (3), Autosomal recessive; ?Congenital heart defects, hamartomas of tongue, and polysyndactyly, 217085 (3), Autosomal recessive
WDR19	608151	?Cranioectodermal dysplasia 4, 614378 (3), Autosomal recessive; Nephronophthisis 13, 614377 (3), Autosomal recessive; Senior-Loken syndrome 8, 616307 (3), Autosomal recessive; ?Short-rib thoracic dysplasia 5 with or without polydactyly, 614376 (3), Autosomal recessive
WDR34	613363	Short-rib thoracic dysplasia 11 with or without polydactyly, 615633 (3), Autosomal recessive
WDR35	613602	Cranioectodermal dysplasia 2, 613610 (3), Autosomal recessive; Short-rib thoracic dysplasia 7 with or without polydactyly, 614091 (3), Autosomal recessive
WDR60	615462	Short-rib thoracic dysplasia 8 with or without polydactyly, 615503 (3), Autosomal recessive
WDR62	613583	Microcephaly 2, primary, autosomal recessive, with or without cortical malformations, 604317 (3), Autosomal recessive
WDR81	614218	Cerebellar ataxia, mental retardation, and dysequilibrium syndrome 2, 610185 (3), Autosomal recessive; Hydrocephalus, congenital, 3, with brain anomalies, 617967 (3), Autosomal recessive
WFS1	606201	?Cataract 41, 116400 (3), Autosomal dominant; Deafness, autosomal dominant 6/14/38, 600965 (3), Autosomal dominant; {Diabetes mellitus, noninsulin-dependent, association with}, 125853 (3), Autosomal dominant; Wolfram syndrome 1, 222300 (3), Autosomal recessive; Wolfram-like syndrome, autosomal dominant, 614296 (3), Autosomal dominant
WHRN	607928	Deafness, autosomal recessive 31, 607084 (3), Autosomal recessive; Usher syndrome, type 2D, 611383 (3), Autosomal recessive
WNK1	605232	Neuropathy, hereditary sensory and autonomic, type II, 201300 (3), Autosomal recessive; Pseudohypoaldosteronism, type IIC, 614492 (3), Autosomal dominant
WNT10A	606268	Odontoonychodermal dysplasia, 257980 (3), Autosomal recessive; Schopf-Schulz-Passarge syndrome, 224750 (3), Autosomal recessive; Tooth agenesis, selective, 4, 150400 (3), Autosomal recessive, Autosomal dominant
WNT10B	601906	Split-hand/foot malformation 6, 225300 (3), Autosomal recessive; Tooth agenesis, selective, 8, 617073 (3), Autosomal dominant
WNT3	165330	?Tetra-amelia syndrome 1, 273395 (3), Autosomal recessive
WNT4	603490	Mullerian aplasia and hyperandrogenism, 158330 (3), Autosomal dominant; ?SERKAL syndrome, 611812 (3), Autosomal recessive
WNT7A	601570	Fuhrmann syndrome, 228930 (3), Autosomal recessive; Ulna and fibula, absence of, with severe limb deficiency, 276820 (3), Autosomal recessive

WRN	604611	Werner syndrome, 277700 (3), Autosomal recessive
WWOX	605131	Epileptic encephalopathy, early infantile, 28, 616211 (3), Autosomal recessive; Esophageal squamous cell carcinoma, somatic, 133239 (3); Spinocerebellar ataxia, autosomal recessive 12, 614322 (3), Autosomal recessive
XPA	611153	Xeroderma pigmentosum, group A, 278700 (3), Autosomal recessive
XPC	613208	Xeroderma pigmentosum, group C, 278720 (3), Autosomal recessive
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
YARS2	610957	Myopathy, lactic acidosis, and sideroblastic anemia 2, 613561 (3), Autosomal recessive
ZAP70	176947	Autoimmune disease, multisystem, infantile-onset, 2, 617006 (3), Autosomal recessive; Immunodeficiency 48, 269840 (3), Autosomal recessive
ZFYVE26	612012	Spastic paraplegia 15, autosomal recessive, 270700 (3), Autosomal recessive
ZMPSTE24	606480	Mandibuloacral dysplasia with type B lipodystrophy, 608612 (3), Autosomal recessive; Restrictive dermopathy, lethal, 275210 (3), Autosomal recessive
ZNF423	604557	Joubert syndrome 19, 614844 (3), Autosomal recessive, Autosomal dominant; Nephronophthisis 14, 614844 (3), Autosomal recessive, Autosomal dominant
ZNF469	612078	Brittle cornea syndrome 1, 229200 (3), Autosomal recessive

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: Sept 30, 2019

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.