CGT Exome v3.1.2

gene	OMIM gene	chrom	Previous symbol	OMIM phenotype	Phenotype	MOI
AAAS	605378	12		231550	Triple-A syndrome (achalasia-addisonianism-alacrimia)	Autosomal recessive
AARS1	601065	16	AARS	616339	Epileptic encephalopathy, early infantile, type 29	Autosomal recessive
AARS2	612035	6		614096; 615889	Combined oxidative phosphorylation deficiency 8; Leukoencephalopathy, progressive, with ovarian failure	Autosomal recessive
AASS	605113	7		238700; 268700	Hyperlysinemia, type 1 and type 2	Autosomal recessive
ABAT	137150	16		613163	GABA-transaminase deficiency	Autosomal recessive
ABCA1	600046	9		205400	Tangier disease	Autosomal recessive
ABCA12	607800	2		601277; 242500	Ichthyosis, congenital, autosomal recessive, type 4A; ICAR, type 4B (harlequin)	Autosomal recessive
ABCA3	601615	16		610921	Surfactant metabolism dysfunction, pulmonary, type 3	Autosomal recessive
ABCA4	601691	1		248200; 604116	Stargardt disease type 1; Cone-rod dystrophy type 3	Autosomal recessive
ABCB11	603201	2		605479; 601847	Cholestasis, benign recurrent intrahepatic, type 2; Cholestasis, progressive familial intrahepatic, type 2	Autosomal recessive
ABCB4	171060	7		602347	Cholestasis, progressive familial intrahepatic, type 3	Autosomal recessive
ABCC2	601107	10		237500	Dubin-Johnson syndrome	Autosomal recessive
ABCC6	603234	16		264800; 614473	Pseudoxanthoma elasticum; Generalized arterial calcification of infancy, type 2	Autosomal recessive
ABCC8	600509	11		256450*; 606176*	Hyperinsulinemic hypoglycemia, type 1 (congenital hyperinsulinism); Permanent neonatal diabetes mellitus (PNDM)	Autosomal recessive*
ABCD1	300371	X		300100	Adrenoleukodystrophy	X-linked
ABCD4	603214	14		614857	Methylmalonic aciduria and homocystinuria, cblJ type	Autosomal recessive
ABCG5	605459	2		210250	Sitosterolemia	Autosomal recessive
ABCG8	605460	2		210250	Sitosterolemia	Autosomal recessive
ABHD12	613599	20		612674	PHARC syndrome (polyneuropathy, hearing loss, ataxia, retinitis pigmentosa and cataract)	Autosomal recessive
ABHD5	604780	3		275630	Chanarin-Dorfman syndrome	Autosomal recessive
ACAD8	604773	11		611283	Isobutyryl-CoA dehydrogenase deficiency	Autosomal recessive
ACAD9	611103	3		611126	Acyl-CoA dehydrogenase 9 deficiency (mitochondrial complex I deficiency, nuclear, type 20)	Autosomal recessive
ACADM	607008	1		201450	Medium-chain acyl-CoA dehydrogenase deficiency	Autosomal recessive
ACADS	606885	12		201470	Short-chain acyl-CoA dehydrogenase deficiency	Autosomal recessive
ACADSB	600301	10		610006	Short/branched-chain acyl-CoA dehydrogenase deficiency	Autosomal recessive
ACADVL	609575	17		201475	Very long-chain acyl-CoA dehydrogenase (VLCAD) deficiency	Autosomal recessive
ACAT1	607809	11		203750	Alpha-methylacetoacetic aciduria (3-ketothiolase deficiency)	Autosomal recessive
ACE	106180	17		267430	Renal tubular dysgenesis	Autosomal recessive
ACO2	100850	22		614559	Infantile cerebellar-retinal degeneration	Autosomal recessive
ACOX1	609751	17		264470	Peroxisomal acyl-CoA oxidase deficiency	Autosomal recessive
ACOX2	601641	3		617308	Bile acid synthesis defect, congenital, type 6	Autosomal recessive
ACP5	171640	19		607944	Spondyloenchondrodysplasia with immune dysregulation	Autosomal recessive
ACSF3	614245	16		614265	Combined malonic and methylmalonic aciduria	Autosomal recessive
ACTA1	102610	1		161800*; 255310*	Nemaline myopathy 3; Congenital fiber-type disproportion myopathy 1	Autosomal recessive*
ACY1	104620	3		609924	Aminoacylase 1 deficiency	Autosomal recessive
ADA	608958	20		102700	Severe combined immunodeficiency due to adenosine deaminase deficiency (ADA)	Autosomal recessive
ADA2	607575	22	CECR1	615688	Vasculitis, autoinflammation, immunodeficiency, and hematologic defects syndrome	Autosomal recessive
ADAM9	602713	8		612775	Cone-rod dystrophy 9	Autosomal recessive
ADAMTS10	608990	19		277600	Weill-Marchesani syndrome, type 1, recessive	Autosomal recessive
ADAMTS13	604134	9		274150	Thrombotic thrombocytopenic purpura, familial (Schulman-Upshaw syndrome)	Autosomal recessive
ADAMTS17	607511	15		613195	Weill-Marchesani syndrome, type 4, recessive	Autosomal recessive
ADAMTS18	607512	16		615458	Microcornea, myopic chorioretinal atrophy, and telecanthus	Autosomal recessive
ADAMTS2	604539	5		225410	Ehlers-Danlos syndrome, dermatosparaxis type	Autosomal recessive
ADAMTSL2	612277	9		231050	Geleophysic dysplasia type 1	Autosomal recessive
ADAMTSL4	610113	1		225200; 225100	Ectopia lentis et pupillae; Ectopia lentis, isolated, type 2	Autosomal recessive
ADAR	146920	1		615010	Aicardi-Goutieres syndrome, type 6	Autosomal recessive
ADAT3	615302	19		615286	Mental retardation, autosomal recessive 36	Autosomal recessive
ADGRG1	604110	16	GPR56	606854	Polymicrogyria, bilateral frontoparietal	Autosomal recessive
ADGRG6	612243	6	GPR126	616503	Lethal congenital contracture syndrome 9	Autosomal recessive
ADGRV1	602851	5	GPR98	605472	Usher syndrome, type 2C	Autosomal recessive, digenic inheritance (PDZD7 gene)

ADK	102750	10		614300	Hypermethioninemia due to adenosine kinase deficiency	Autosomal recessive
ADSL	608222	22		103050	Adenylosuccinase deficiency	Autosomal recessive
ADSS1	612498	14	ADSSL1	617030	Myopathy, distal, 5	Autosomal recessive
AFG3L2	604581	18		614487	Spastic ataxia, type 5, autosomal recessive	Autosomal recessive
AFP	104150	4		615969	Alpha-fetoprotein deficiency	Autosomal recessive
AGA	613228	4		208400	Aspartylglucosaminuria (glycosylasparaginase deficiency)	Autosomal recessive
AGBL5	615900	2		617023	Retinitis pigmentosa 75	Autosomal recessive
AGK	610345	7		614691; 212350	Cataract 38; Sengers syndrome	Autosomal recessive
AGL	610860	1		232400	Glycogen storage disease, type 3	Autosomal recessive
AGPAT2	603100	9		608594	Congenital generalized lipodystrophy (Berardinelli-Seip syndrome)	Autosomal recessive
AGPS	603051	2		600121	Rhizomelic chondrodysplasia punctata, type 3	Autosomal recessive
AGRN	103320	1		615120	Myasthenic syndrome, congenital, type 8	Autosomal recessive
AGT	106150	1		267430	Renal tubular dysgenesis	Autosomal recessive
AGTR1	106165	3		267430	Renal tubular dysgenesis	Autosomal recessive
AGXT	604285	2		259900	Hyperoxaluria, primary, type 1	Autosomal recessive
AHCY	180960	20		613752	Hypermethioninemia with deficiency of S-adenosylhomocysteine hydrolase	Autosomal recessive
AHI1	608894	6		608629	Joubert syndrome, type 3	Autosomal recessive
AICDA	605257	12		605258	Immunodeficiency with hyper-IgM, type 2	Autosomal recessive
AIMP1	603605	4		260600	Leukodystrophy, hypomyelinating, type 3	Autosomal recessive
AIMP2	600859	7		618006	Leukodystrophy, hypomyelinating, type 17	Autosomal recessive
AIPL1	604392	17		604393	Leber congenital amaurosis, type 4	Autosomal recessive
AIRE	607358	21		240300*	Autoimmune polyendocrinopathy syndrome, type 1	Autosomal recessive*
AK1	103000	9		612631	Hemolytic anemia due to adenylate kinase deficiency	Autosomal recessive
AK2	103020	1		267500	Reticular dysgenesis	Autosomal recessive
AKR1C2	600450	10		614279	46,XY disorder of sex development due to testicular 17,20-desmolase deficiency	Autosomal recessive
AKR1D1	604741	7		235555	Bile acid synthesis defect, congenital, type 2	Autosomal recessive
ALAD	125270	9		612740	Porphyria, acute hepatic	Autosomal recessive
ALB	103600	4		616000	Analbuminemia	Autosomal recessive
ALDH18A1	138250	10		616586; 219150	Spastic paraplegia, type 9B, autosomal recessive; Cutis laxa, type 3A (De Barsy syndrome)	Autosomal recessive
ALDH1A3	600463	15		615113	Microphthalmia, isolated 8	Autosomal recessive
ALDH3A2	609523	17		270200	Sjogren-Larsson syndrome	Autosomal recessive
ALDH4A1	606811	1		239510	Hyperprolinemia, type 2	Autosomal recessive
ALDH5A1	610045	6		271980	Succinic semialdehyde dehydrogenase deficiency	Autosomal recessive
ALDH6A1	603178	14		614105	Methylmalonate semialdehyde dehydrogenase deficiency	Autosomal recessive
ALDH7A1	107323	5		266100	Epilepsy, pyridoxine-dependent	Autosomal recessive
ALDOA	103850	16		611881	Glycogen storage disease type 12	Autosomal recessive
ALDOB	612724	9		229600	Fructose intolerance, hereditary	Autosomal recessive
ALG1	605907	16		608540	Congenital disorder of glycosylation, type 1K	Autosomal recessive
ALG11	613666	13		613661	Congenital disorder of glycosylation, type 1P	Autosomal recessive
ALG12	607144	22		607143	Congenital disorder of glycosylation, type 1G	Autosomal recessive
ALG2	607905	9		616228	Myasthenic syndrome, congenital, type 14, with tubular aggregates	Autosomal recessive
ALG3	608750	3		601110	Congenital disorder of glycosylation, type 1D	Autosomal recessive
ALG6	604566	1		603147	Congenital disorder of glycosylation, type 1C	Autosomal recessive
ALG8	608103	11		608104	Congenital disorder of glycosylation, type 1H	Autosomal recessive
ALG9	606941	11		608776; 263210	Congenital disorder of glycosylation, type 1L; Gillessen-Kaesbach-Nishimura syndrome	Autosomal recessive
ALMS1	606844	2		203800	Alstr”m syndrome	Autosomal recessive
ALOX12B	603741	17		242100	Ichthyosis, congenital, autosomal recessive, type 2	Autosomal recessive
ALOXE3	607206	17		606545	Ichthyosis, congenital, autosomal recessive, type 3	Autosomal recessive
ALPK3	617608	15		618052	Cardiomyopathy, familial hypertrophic, type 27	Autosomal recessive
ALPL	171760	1		241500; 241510	Hypophosphatasia, infantile/childhood	Autosomal recessive
ALS2	606352	2		205100; 606353; 607225	Amyotrophic lateral sclerosis, type 2, juvenile; Primary lateral sclerosis, juvenile; Spastic paralysis, infantile onset ascending	Autosomal recessive
ALX1	601527	12		613456	Frontonasal dysplasia, type 3	Autosomal recessive
ALX3	606014	1		136760	Frontonasal dysplasia, type 1	Autosomal recessive
ALX4	605420	11		613451	Frontonasal dysplasia, type 2	Autosomal recessive

AMACR	604489	5		214950; 614307	Bile acid synthesis defect, congenital, type 4; Alpha-methylacyl-CoA racemase deficiency	Autosomal recessive
AMBN	601259	4		616270	Amelogenesis imperfecta, type IF	Autosomal recessive
AMH	600957	19		261550	Persistent Mullerian duct syndrome, type 1	Autosomal recessive
AMHR2	600956	12		261550	Persistent Mullerian duct syndrome, type II	Autosomal recessive
AMN	605799	14		261100	Megaloblastic anemia 1 (Imerslund-Grasbeck syndrome)	Autosomal recessive
AMPD1	102770	1		615511	Myopathy due to myoadenylate deaminase deficiency	Autosomal recessive
AMPD2	102771	1		615809	Pontocerebellar hypoplasia, type 9	Autosomal recessive
AMT	238310	3		605899	Glycine encephalopathy	Autosomal recessive
ANGPTL3	604774	1		605019	Hypobetalipoproteinemia, familial, type 2	Autosomal recessive
ANKS6	615370	9		615382	Nephronophthisis 16	Autosomal recessive
ANO10	613726	3		613728	Spinocerebellar ataxia, autosomal recessive, type 10	Autosomal recessive
ANO5	608662	11		611307	Limb-girdle muscular dystrophy, type 12 (LGMD R12)	Autosomal recessive
ANTXR1	606410	2		230740	GAPO syndrome	Autosomal recessive
ANTXR2	608041	4		228600	Hyaline fibromatosis syndrome	Autosomal recessive
AP1S1	603531	7		609313	MEDNIK syndrome	Autosomal recessive
AP1S2	300629	X		304340	Mental retardation, X-linked, syndromic, type 5 (Pettigrew syndrome)	X-linked
AP3B1	603401	5		608233	Hermansky-Pudlak syndrome, type 2	Autosomal recessive
AP3B2	602166	15		617276	Epileptic encephalopathy, early infantile, type 48	Autosomal recessive
AP3D1	607246	19		617050	?Hermansky-Pudlak syndrome, type 10	Autosomal recessive
AP4B1	607245	1		614066	Spastic paraplegia, type 47, autosomal recessive	Autosomal recessive
AP4E1	607244	15		613744	Spastic paraplegia, type 51, autosomal recessive	Autosomal recessive
AP4M1	602296	7		612936	Spastic paraplegia, type 50, autosomal recessive	Autosomal recessive
AP4S1	607243	14		614067	Spastic paraplegia, type 52, autosomal recessive	Autosomal recessive
AP5Z1	613653	7		613647	Spastic paraplegia, type 48, autosomal recessive	Autosomal recessive
APOC2	608083	19		207750	Hyperlipoproteinemia, type 1B	Autosomal recessive
APOE	107741	19		269600	Sea-blue histiocyte disease	Autosomal recessive
APRT	102600	16		614723	Adenine phosphoribosyltransferase deficiency	Autosomal recessive
APTX	606350	9		208920	Ataxia, early-onset, with oculomotor apraxia and hypoalbuminemia	Autosomal recessive
AQP2	107777	12		125800*	Diabetes insipidus, nephrogenic, type 2	Autosomal recessive*
AR	313700	X		300068	Androgen insensitivity syndrome, complete	X-linked
ARFGEF2	605371	20		608097	Periventricular heterotopia with microcephaly	Autosomal recessive
ARG1	608313	6		207800	Argininemia (arginase deficiency)	Autosomal recessive
ARHGDIA	601925	17		615244	Nephrotic syndrome, type 8	Autosomal recessive
ARHGEF18	616432	19		617433	Retinitis pigmentosa 78	Autosomal recessive
ARL13B	608922	3		612291	Joubert syndrome type 8	Autosomal recessive
ARL2BP	615407	16		615434	Retinitis pigmentosa with or without situs inversus	Autosomal recessive
ARL6	608845	3		600151	Bardet-Biedl syndrome, type 3	Autosomal recessive
ARMC9	617612	2		617622	Joubert syndrome 30	Autosomal recessive
ARPC1B	604223	7		617718	Immunodeficiency, type 71, with inflammatory disease and congenital thrombocytopenia	Autosomal recessive
ARSA	607574	22		250100	Metachromatic leukodystrophy	Autosomal recessive
ARSB	611542	5		253200	Mucopolysaccharidosis, type 6 (Maroteaux-Lamy syndrome)	Autosomal recessive
ARSL	300180	X	ARSE	302950	Chondrodysplasia punctata, brachytelephalangic	X-linked
ARV1	611647	1		617020	Epileptic encephalopathy, early infantile, 38	Autosomal recessive
ARX	300382	X		308350; 300215; 309510	Epileptic encephalopathy, early infantile, type 1; ARX-related developmental disorders	X-linked
ASAH1	613468	8		228000; 159950	Farber lipogranulomatosis; Spinal muscular atrophy with progressive myoclonic epilepsy	Autosomal recessive
ASL	608310	7		207900	Argininosuccinic aciduria	Autosomal recessive
ASNS	108370	7		615574	Asparagine synthetase deficiency	Autosomal recessive
ASPA	608034	17		271900	Canavan disease	Autosomal recessive
ASPH	600582	8		601552	Traboulsi syndrome	Autosomal recessive
ASPM	605481	1		608716	Primary microcephaly type 5, autosomal recessive	Autosomal recessive
ASS1	603470	9		215700	Citrullinemia, type 1	Autosomal recessive
ATAD1	614452	10		618011	Hyperekplexia 4	Autosomal recessive
ATF6	605537	1		616517	Achromatopsia, type 7	Autosomal recessive
ATIC	601731	2		608688	AICA-ribosiduria due to ATIC deficiency	Autosomal recessive
ATM	607585	11		208900	Ataxia-telangiectasia	Autosomal recessive

ATOH7	609875	10		221900	Persistent hyperplastic primary vitreous, autosomal recessive	Autosomal recessive
ATP13A2	610513	1		606693; 617225	Kufor-Rakeb syndrome; Spastic paraplegia, type 78, autosomal recessive	Autosomal recessive
ATP2A1	108730	16		601003	Brody myopathy	Autosomal recessive
ATP6V0A2	611716	12		219200; 278250	Cutis laxa, autosomal recessive, type 2A; Wrinkly skin syndrome	Autosomal recessive
ATP6V0A4	605239	7		602722	Renal tubular acidosis, distal, autosomal recessive	Autosomal recessive
ATP6V1A	607027	3		617403	Cutis laxa, autosomal recessive, type 2D	Autosomal recessive
ATP6V1B1	192132	2		267300	Renal tubular acidosis with deafness	Autosomal recessive
ATP6V1E1	108746	22		617402	Cutis laxa, autosomal recessive, type 2C	Autosomal recessive
ATP7A	300011	X		309400; 304150	Menkes disease; Occipital horn syndrome	X-linked
ATP7B	606882	13		277900	Wilson disease	Autosomal recessive
ATP8B1	602397	18		211600; 243300	Cholestasis, progressive familial intrahepatic, type 1; Cholestasis, benign recurrent intrahepatic, type 1	Autosomal recessive
ATR	601215	3		210600	Seckel syndrome, type 1	Autosomal recessive
ATRX	300032	X		309580; 301040	Mental retardation-hypotonic facies syndrome, X-linked; Alpha-thalassemia/mental retardation syndrome	X-linked
AUH	600529	9		250950	3-methylglutaconic aciduria, type 1	Autosomal recessive
AURKC	603495	19		243060	Spermatogenic failure, type 5	Autosomal recessive
AVIL	613397	12		618594	Nephrotic syndrome, type 21	Autosomal recessive
B2M	109700	15		241600	Immunodeficiency, type 43	Autosomal recessive
B3GALNT2	610194	1		615181	Muscular dystrophy-dystroglycanopathy (congenital with brain and eye anomalies, type A, 11	Autosomal recessive
B3GALT6	615291	1		615349	Ehlers-Danlos syndrome, spondylodysplastic type, 2	Autosomal recessive
B3GAT3	606374	11		245600	Multiple joint dislocations, short stature, craniofacial dysmorphism, with or without congenital heart defects	Autosomal recessive
B3GLCT	610308	13	B3GALTL	261540	Peters-plus syndrome	Autosomal recessive
B4GALNT1	601873	12		609195	Spastic paraplegia, type 26, autosomal recessive	Autosomal recessive
B4GALT1	137060	9		607091	Congenital disorder of glycosylation, type 2D	Autosomal recessive
B4GALT7	604327	5		130070	Ehlers-Danlos syndrome, spondylodysplastic, type 1	Autosomal recessive
B4GAT1	605517	11	B3GNT1	615287	Muscular dystrophy-dystroglycanopathy (congenital with brain and eye anomalies), type A, 13	Autosomal recessive
B9D1	614144	17		617120; 614209	Joubert syndrome, type 27; ?Meckel syndrome 9	Autosomal recessive
B9D2	611951	19		614175; 614175	Joubert syndrome, type 34; ?Meckel syndrome, type 10	Autosomal recessive
BBS1	209901	11		209900	Bardet-Biedl syndrome, type 1	Autosomal recessive
BBS10	610148	12		615987	Bardet-Biedl syndrome, type 10	Autosomal recessive
BBS12	610683	4		615989	Bardet-Biedl syndrome, type 12	Autosomal recessive
BBS2	606151	16		615981	Bardet-Biedl syndrome, type 2	Autosomal recessive
BBS4	600374	15		615982	Bardet-Biedl syndrome, type 4	Autosomal recessive
BBS5	603650	2		615983	Bardet-Biedl syndrome, type 5	Autosomal recessive
BBS7	607590	4		615984	Bardet-Biedl syndrome, type 7	Autosomal recessive
BBS9	607968	7		615986	Bardet-Biedl syndrome, type 9	Autosomal recessive
BCAT2	113530	19		618850	?Hypervalinemia or hyperleucine-isoleucinemia	Autosomal recessive
BCHE	177400	3		617936	Butyrylcholinesterase deficiency	Autosomal recessive
BCKDHA	608348	19		248600	Maple syrup urine disease, type 1A	Autosomal recessive
BCKDHB	248611	6		248600	Maple syrup urine disease, type 1B	Autosomal recessive
BCKDK	614901	16		614923	Branched-chain ketoacid dehydrogenase kinase deficiency	Autosomal recessive
BCL10	603517	1		616098	?Immunodeficiency, type 37	Autosomal recessive
BCS1L	603647	2		256000	BCS1L-related disorders, including Leigh syndrome	Autosomal recessive
BEST1	607854	11		611809	Bestrophinopathy, AR	Autosomal recessive
BFSP1	603307	20		611391*	Cataract 33, multiple types	Autosomal recessive*
BHLHA9	615416	17		609432	Syndactyly, mesoaxial synostotic, with phalangeal reduction	Autosomal recessive
BIN1	601248	2		255200	Centronuclear myopathy, type 2	Autosomal recessive
BLM	604610	15		210900	Bloom syndrome	Autosomal recessive
BLNK	604515	10		613502	?Agammaglobulinemia 4	Autosomal recessive
BLOC1S3	609762	19		614077	Hermansky-Pudlak syndrome, type 8	Autosomal recessive
BLOC1S6	604310	15	PLDN	614171	?Hermansky-Pudlak syndrome, type 9	Autosomal recessive
BLVRA	109750	7		614156*	Hyperbiliverdinemia	Autosomal recessive*
BMP1	112264	8		614856	Osteogenesis imperfecta, type 13	Autosomal recessive
BMPER	608699	7		608022	Diaphanospondylodysostosis	Autosomal recessive
BMPR1B	603248	4		609441	Acromesomelic dysplasia, Demirhan type	Autosomal recessive
BOLA3	613183	2		614299	Multiple mitochondrial dysfunctions syndrome 2 with hyperglycinemia	Autosomal recessive

BPGM	613896	7		222800	Erythrocytosis due to bisphosphoglycerate mutase deficiency	Autosomal recessive
BPNT2	614010	8	IMPAD1	614078	Chondrodysplasia with joint dislocations, GPAPP type	Autosomal recessive
BRAT1	614506	7		614498; 618056	Rigidity and multifocal seizure syndrome, lethal neonatal; Neurodevelopmental disorder with cerebellar atrophy and with or without seizures	Autosomal recessive
BRF1	604902	14		616202	Cerebellofaciodental syndrome	Autosomal recessive
BRIP1	605882	17		609054	Fanconi anemia, complementation group J	Autosomal recessive
BRWD3	300553	X		300659	Mental retardation, X-linked, type 93	X-linked
BSCL2	606158	11		269700; 615924	Congenital generalized lipodystrophy, type 2; Encephalopathy, progressive, with or without lipodystrophy	Autosomal recessive
BSND	606412	1		602522	Bartter syndrome, type 4A	Autosomal recessive
BTD	609019	3		253260	Biotinidase deficiency	Autosomal recessive
BTK	300300	X		300755	Agammaglobulinemia X-linked, type 1	X-linked
BUB1B	602860	15		257300	Mosaic variegated aneuploidy syndrome 1	Autosomal recessive
C12orf57	615140	12		218340	Temtamy syndrome	Autosomal recessive
C12orf65	613541	12		613559	Combined oxidative phosphorylation deficiency 7; Spastic paraplegia, type 55, autosomal recessive	Autosomal recessive
C19orf12	614297	19		614298*	Neurodegeneration with brain iron accumulation, type 4	Autosomal recessive*
C1QA	120550	1		613652	C1q deficiency	Autosomal recessive
C1QB	120570	1		613652	C1q deficiency	Autosomal recessive
C1QBP	601269	17		617713	Combined oxidative phosphorylation deficiency 33	Autosomal recessive
C1QC	120575	1		613652	C1q deficiency	Autosomal recessive
C1S	120580	12		613783	C1s deficiency	Autosomal recessive
C2	613927	6		217000	C2 deficiency	Autosomal recessive
C2CD3	615944	11		615948	Orofaciodigital syndrome, type 14	Autosomal recessive
C3	120700	19		613779	Complement component 3 deficiency	Autosomal recessive
C5	120900	9		609536	Complement component 5 deficiency	Autosomal recessive
C6	217050	5		612446	Complement component 6 deficiency	Autosomal recessive
C7	217070	5		610102	Complement component 7 deficiency	Autosomal recessive
C8B	120960	1		613789	Complement component 8 deficiency, type 2	Autosomal recessive
C8orf37	614477	8		617406; 614500	Bardet-Biedl syndrome, type 21; Cone-rod dystrophy 16 and Retintis pigmentosa 64	Autosomal recessive
CA12	603263	15		143860	Hyperchlorhidrosis, isolated	Autosomal recessive
CA2	611492	8		259730	Osteopetrosis with renal tubular acidosis (osteopetrosis, autosomal recessive, type 3)	Autosomal recessive
CA5A	114761	16		615751	Hyperammonemia due to carbonic anhydrase VA deficiency	Autosomal recessive
CA8	114815	8		613227	Cerebellar ataxia and mental retardation with or without quadrupedal locomotion 3	Autosomal recessive
CABP2	607314	11		614899	Deafness, autosomal recessive, type 93	Autosomal recessive
CABP4	608965	11		610427	Congenital stationary night blindness, type 2B	Autosomal recessive
CACNA1D	114206	3		614896	Sinoatrial node dysfunction and deafness	Autosomal recessive
CACNA2D4	608171	12		610478	Retinal cone dystrophy 4	Autosomal recessive
CAD	114010	2		616457	Epileptic encephalopathy, early infantile, 50	Autosomal recessive
CALCRL	114190	2		618773	?Lymphatic malformation 8	Autosomal recessive
CANT1	613165	17		251450; 617719	Desbuquois dysplasia, type 1; Epiphyseal dysplasia, multiple, type 7	Autosomal recessive
CAPN1	114220	11		616907	Spastic paraplegia, type 76, autosomal recessive	Autosomal recessive
CAPN3	114240	15		253600	Limb-girdle muscular dystrophy, type 1 (LGMD R1)	Autosomal recessive
CARD11	607210	7		615206	Immunodeficiency, type 11A	Autosomal recessive
CARD9	607212	9		212050	Candidiasis, familial, type 2, autosomal recessive	Autosomal recessive
CARS2	612800	13		616672	Combined oxidative phosphorylation deficiency 27	Autosomal recessive
CASQ2	114251	1		611938	Ventricular tachycardia, catecholaminergic polymorphic, type 2	Autosomal recessive
CASR	601199	3		239200*	Hyperparathyroidism, neonatal	Autosomal recessive*
CAST	114090	5		616295	Peeling skin with leukonychia, acral punctate keratoses, cheilitis, and knuckle pads	Autosomal recessive
CAT	115500	11		614097	Acatalasemia	Autosomal recessive
CATSPER1	606389	11		612997	Spermatogenic failure, type 7	Autosomal recessive
CAVIN1	603198	17	PTRF	613327	Lipodystrophy, congenital generalized, type 4	Autosomal recessive
CBLIF	609342	11	GIF	261000	Intrinsic factor deficiency	Autosomal recessive
CBS	613381	21		236200	Homocystinuria due to cystathionine beta-synthase	Autosomal recessive
CC2D1A	610055	19		608443	Mental retardation, autosomal recessive, type 3	Autosomal recessive
CC2D2A	612013	4		612285; 612284	Joubert syndrome, type 9; Meckel syndrome, type 6	Autosomal recessive
CCBE1	612753	18		235510	Hennekam lymphangiectasia-lymphedema syndrome, type 1	Autosomal recessive

CCDC103	614677	17		614679	Ciliary dyskinesia, primary, type 17	Autosomal recessive
CCDC115	613734	2		616828	Congenital disorder of glycosylation, type IIo	Autosomal recessive
CCDC174	616735	3		616816	Hypotonia, infantile, with psychomotor retardation	Autosomal recessive
CCDC39	613798	3		613807	Ciliary dyskinesia, primary, type 14	Autosomal recessive
CCDC40	613799	17		613808	Ciliary dyskinesia, primary, type 15	Autosomal recessive
CCDC65	611088	12		615504	Ciliary dyskinesia, primary, type 27	Autosomal recessive
CCDC8	614145	19		614205	3M syndrome 3	Autosomal recessive
CCDC88C	611204	14		236600	Hydrocephalus, congenital, type 1	Autosomal recessive
CCN6	603400	6	WISP3	208230	Arthropathy, progressive pseudorheumatoid, of childhood	Autosomal recessive
CCNO	607752	5		615872	Ciliary dyskinesia, primary, type 29	Autosomal recessive
CD19	107265	16		613493	Immunodeficiency, common variable, type 3	Autosomal recessive
CD247	186780	1		610163	?Immunodeficiency, type 25	Autosomal recessive
CD27	186711	12		615122	Lymphoproliferative syndrome 2	Autosomal recessive
CD2AP	604241	6		607832*	Glomerulosclerosis, focal segmental, type 3, susceptibility to	Autosomal recessive*
CD320	606475	19		613646	Methylmalonic aciduria, transient, due to transcobalamin receptor defect	Autosomal recessive
CD36	173510	7		608404	Platelet glycoprotein 4 deficiency	Autosomal recessive
CD3D	186790	11		615617	Immunodeficiency, type 19	Autosomal recessive
CD3E	186830	11		615615	Immunodeficiency, type 18	Autosomal recessive
CD3G	186740	11		615607	Immunodeficiency, type 17, CD3 gamma deficient	Autosomal recessive
CD40	109535	20		606843	Immunodeficiency with hyper-IgM, type 3	Autosomal recessive
CD40LG	300386	X		308230	Hyper-IgM syndrome, type 1 (immunodeficiency, X-linked, with hyper-IgM, type 1)	X-linked
CD55	125240	1		226300	Complement hyperactivation, angiopathic thrombosis, and protein-losing enteropathy (CHAPLE)	Autosomal recessive
CD59	107271	11		612300	CD59 deficiency	Autosomal recessive
CD79A	112205	19		613501	Agammaglobulinemia 3	Autosomal recessive
CD79B	147245	17		612692	Agammaglobulinemia 6	Autosomal recessive
CD81	186845	11		613496	Immunodeficiency, common variable, type 6	Autosomal recessive
CD8A	186910	2		608957	CD8 deficiency, familial	Autosomal recessive
CDAN1	607465	15		224120	Dyserythropoietic anemia, congenital, type 1A	Autosomal recessive
CDC14A	603504	1		616958	Deafness, autosomal recessive, type 105	Autosomal recessive
CDC45	603465	22		617063	Meier-Gorlin syndrome 7	Autosomal recessive
CDCA7	609937	2		616910	Immunodeficiency-centromeric instability-facial anomalies syndrome 3	Autosomal recessive
CDH11	600023	16		211380	Elsahy-Waters syndrome	Autosomal recessive
CDH23	605516	10		601386; 601067	Deafness, autosomal recessive, type 12; Usher syndrome, type 1D	Autosomal recessive
CDH3	114021	16		225280	Ectodermal dysplasia, ectrodactyly, and macular dystrophy	Autosomal recessive
CDHR1	609502	10		613660	Cone-rod dystrophy, type 15	Autosomal recessive
CDIN1	615626	15	C15orf41	615631	Dyserythropoietic anemia, congenital, type Ib	Autosomal recessive
CDK10	603464	16		617694	Al Kaissi syndrome	Autosomal recessive
CDK5RAP2	608201	9		604804	Primary microcephaly type 3, autosomal recessive	Autosomal recessive
CDSN	602593	6		270300	Peeling skin syndrome 1	Autosomal recessive
CDT1	605525	16		613804	Meier-Gorlin syndrome, type 4	Autosomal recessive
CEBPE	600749	14		245480	Specific granule deficiency	Autosomal recessive
CENPF	600236	1		243605	Stromme syndrome	Autosomal recessive
CENPJ	609279	13		608393	Primary microcephaly type 6, autosomal recessive	Autosomal recessive
CEP104	616690	1		616781	Joubert syndrome 25	Autosomal recessive
CEP120	613446	5		616300	Short-rib thoracic dysplasia 13 with or without polydactyly	Autosomal recessive
CEP135	611423	4		614673	Microcephaly 8, primary, autosomal recessive	Autosomal recessive
CEP152	613529	15		614852	Primary microcephaly type 9, autosomal recessive	Autosomal recessive
CEP164	614848	11		614845	Nephronophthisis 15	Autosomal recessive
CEP19	615586	3		615703	Morbid obesity and spermatogenic failure	Autosomal recessive
CEP290	610142	12		611134; 610188; 611755	Meckel syndrome, type 4; Joubert syndrome, type 5; Leber congenital amaurosis, type 10	Autosomal recessive
CEP41	610523	7		614464	Joubert syndrome, type 15	Autosomal recessive
CEP55	610000	10		236500	Multinucleated neurons, anhydramnios, renal dysplasia, cerebellar hypoplasia, and hydranencephaly	Autosomal recessive
CEP57	607951	11		614114	Mosaic variegated aneuploidy syndrome 2	Autosomal recessive
CEP78	617110	9		617236	Cone-rod dystrophy and hearing loss	Autosomal recessive
CEP83	615847	12	CCDC41	615862	Nephronophthisis 18	Autosomal recessive

CERKL	608381	2		608380	Retinitis pigmentosa, type 26	Autosomal recessive
CERS3	615276	15		615023	Ichthyosis, congenital, autosomal recessive 9	Autosomal recessive
CFAP43	617558	10	WDR96	617592	Spermatogenic failure, type 19	Autosomal recessive
CFAP53	614759	18	CCDC11	614779	Heterotaxy, visceral, 6, autosomal recessive	Autosomal recessive
CFD	134350	19		613912	Complement factor D deficiency	Autosomal recessive
CFH	134370	1		609814	Complement factor H deficiency	Autosomal recessive
CFI	217030	4		610984	Complement factor I deficiency	Autosomal recessive
CFL2	601443	14		610687	Nemaline myopathy, type 7, autosomal recessive	Autosomal recessive
CFTR	602421	7		219700	Cystic fibrosis	Autosomal recessive
CHAT	118490	10		254210	Myasthenic syndrome, congenital, type 6, presynaptic	Autosomal recessive
CHKB	612395	22		602541	Muscular dystrophy, congenital, megaconial type	Autosomal recessive
CHM	300390	X		303100	Choroideremia	X-linked
CHMP1A	164010	16		614961	Pontocerebellar hypoplasia, type 8	Autosomal recessive
CHRNA1	100690	2		253290	Multiple pterygium syndrome, lethal type	Autosomal recessive
CHRNB1	100710	17		616314	?Myasthenic syndrome, congenital, 2C, associated with acetylcholine receptor deficiency	Autosomal recessive
CHRND	100720	2		616322; 253290	Myasthenic syndrome, congenital, type 3B, fast-channel; Multiple pterygium syndrome, lethal type	Autosomal recessive
CHRNE	100725	17		616324; 608931	Myasthenic syndrome, congenital, type 4B, fast-channel; Myasthenic syndrome, congenital, type 4C, associated with acetylcholine receptor deficiency	Autosomal recessive
CHRNG	100730	2		265000; 253290	Multiple pterygium syndrome (MPS), Escobar type; MPS, lethal type	Autosomal recessive
CHST14	608429	15		601776	Ehlers-Danlos syndrome, musculocontractural, type 1	Autosomal recessive
CHST3	603799	10		143095	Spondyloepiphyseal dysplasia with congenital joint dislocations	Autosomal recessive
CHST6	605294	16		217800	Macular corneal dystrophy	Autosomal recessive
CHSY1	608183	15		605282	Temtamy preaxial brachydactyly syndrome	Autosomal recessive
CHUK	600664	10		613630	Cocoon syndrome	Autosomal recessive
CIB2	605564	15		609439;614869	Deafness, autosomal recessive, type 48; Usher syndrome, type 1J	Autosomal recessive
CIITA	600005	16		209920	Bare lymphocyte syndrome, type 2, complementation group A	Autosomal recessive
CILK1	612325	6	ICK	612651	Endocrine-cerebroosteodysplasia	Autosomal recessive
CISD2	611507	4		604928	Wolfram syndrome 2	Autosomal recessive
CIT	605629	12		617090	Microcephaly 17, primary, autosomal recessive	Autosomal recessive
CKAP2L	616174	2		272440	Filippi syndrome	Autosomal recessive
CLCF1	607672	11		610313	Cold-induced sweating syndrome 2	Autosomal recessive
CLCN1	118425	7		255700	Myotonia congenita, recessive	Autosomal recessive
CLCN2	600570	3		615651	Leukoencephalopathy with ataxia	Autosomal recessive
CLCN7	602727	16		611490	Osteopetrosis, autosomal recessive type 4	Autosomal recessive
CLCNKA	602024	1		613090	Bartter syndrome, type 4B, digenic	Digenic inheritance (CLCNKB gene)
CLCNKB	602023	1		607364; 613090	Bartter syndrome, type 3; Bartter syndrome, type 4B, digenic	Autosomal recessive; Digenic inheritance (CLCNKA gene)
CLDN1	603718	3		607626	Ichthyosis, leukocyte vacuoles, alopecia, and sclerosing cholangitis	Autosomal recessive
CLDN10	617579	13		617671	HELIX syndrome	Autosomal recessive
CLDN14	605608	21		614035	Deafness type 29, autosomal recessive	Autosomal recessive
CLDN16	603959	3		248250	Hypomagnesemia, type 3, renal	Autosomal recessive
CLDN19	610036	1		248190	Rena hypomagnesemia type 5, with ocular involvement	Autosomal recessive
CLMP	611693	11		615237	Congenital short bowel syndrome	Autosomal recessive
CLN3	607042	16		204200	Ceroid lipofuscinosis, neuronal, type 3	Autosomal recessive
CLN5	608102	13		256731	Ceroid lipofuscinosis, neuronal, type 5	Autosomal recessive
CLN6	606725	15		601780	Ceroid lipofuscinosis, neuronal, type 6	Autosomal recessive
CLN8	607837	8		600143	Ceroid lipofuscinosis, neuronal, type 8	Autosomal recessive
CLP1	608757	11		615803	Pontocerebellar hypoplasia, type 10	Autosomal recessive
CLPB	616254	11		616271	3-methylglutaconic aciduria, type 7, with cataracts, neurologic involvement and neutropenia	autosomal recessive
CLPP	601119	19		614129	Perrault syndrome 3	Autosomal recessive
CLRN1	606397	3		276902	Usher syndrome, type 3A	Autosomal recessive
CNGA1	123825	4		613756	Retinitis pigmentosa type 49	Autosomal recessive
CNGA3	600053	2		216900	Achromatopsia, type 2	Autosomal recessive
CNGB1	600724	16		613767	Retinitis pigmentosa type 45	Autosomal recessive

CNGB3	605080	8		262300	Achromatopsia, type 3	Autosomal recessive
CNNM2	607803	10		616418*	Hypomagnesemia, seizures, and mental retardation	Autosomal recessive*
CNNM4	607805	2		217080	Jalili syndrome	Autosomal recessive
CNPY3	610774	6		617929	Epileptic encephalopathy, early infantile, type 60	Autosomal recessive
CNTNAP1	602346	17		616286	Lethal congenital contracture syndrome 7	Autosomal recessive
CNTNAP2	604569	7		610042	Pitt-Hopkins like syndrome 1	Autosomal recessive
COA6	614772	1		616501	Cardioencephalomyopathy, fatal infantile, due to cytochrome c oxidase deficiency 4	Autosomal recessive
COA8	616003	14	APOPT1	619061	Mitochondrial complex IV deficiency, nuclear type 17	Autosomal recessive
COASY	609855	17		615643	Neurodegeneration with brain iron accumulation 6	Autosomal recessive
COG1	606973	17		611209	Congenital disorder of glycosylation, type IIg	Autosomal recessive
COG4	606976	16		613489	Congenital disorder of glycosylation, type 2J	Autosomal recessive
COG5	606821	7		613612	Congenital disorder of glycosylation, type 2I	Autosomal recessive
COG6	606977	13		614576; 615328	Congenital disorder of glycosylation, type 2L; Shaheen syndrome	Autosomal recessive
COG7	606978	16		608779	Congenital disorder of glycosylation, type 2E	Autosomal recessive
COG8	606979	16		611182	Congenital disorder of glycosylation, type 2H	Autosomal recessive
COL11A1	120280	1		228520	Fibrochondrogenesis type 1	Autosomal recessive
COL11A2	120290	6		215150	Otospondylomegaepiphyseal dysplasia, autosomal recessive	Autosomal recessive
COL13A1	120350	10		616720	Myasthenic syndrome, congenital, 19	Autosomal recessive
COL17A1	113811	10		226650	Epidermolysis bullosa, junctional, non-Herlitz type	Autosomal recessive
COL18A1	120328	21		267750	Knobloch syndrome, type 1	Autosomal recessive
COL1A2	120160	7		225320	Ehlers-Danlos syndrome, cardiac valvular type	Autosomal recessive
COL25A1	610004	4		616219	Fibrosis of extraocular muscles, congenital, type 5	Autosomal recessive
COL27A1	608461	9		615155	Steel syndrome	Autosomal recessive
COL4A3	120070	2		203780	Alport syndrome, autosomal recessive, type 2	Autosomal recessive
COL4A4	120131	2		203780	Alport syndrome, autosomal recessive, type 2	Autosomal recessive
COL4A5	303630	X		301050	Alport syndrome, X-linked	X-linked
COL6A1	120220	21		254090*	Ullrich congenital muscular dystrophy, type 1 (Limb-girdle muscular dystrophy, type 22 [LGMD R22])	Autosomal recessive*
COL6A2	120240	21		254090*	Ullrich congenital muscular dystrophy, type 1 (Limb-girdle muscular dystrophy, type 22 [LGMD R22])	Autosomal recessive*
COL6A3	120250	2		254090*	Ullrich congenital muscular dystrophy, type 1 (Limb-girdle muscular dystrophy, type 22 [LGMD R22])	Autosomal recessive*
COL7A1	120120	3		226600; 604129*; 131850*	Dystrophic epidermolysis bullosa (DEB), Hallopeau-Siemens (HS) type and non-HS type; DEB pruriginosa; DEB pretibial	Autosomal recessive; Autosomal recessive*; Autosomal recessive*
COL9A1	120210	6		614134	Stickler syndrome, type 4	Autosomal recessive
COL9A2	120260	1		614284	?Stickler syndrome, type V	Autosomal recessive
COLEC10	607620	8		248340	3MC syndrome 3	Autosomal recessive
COLEC11	612502	2		265050	3MC syndrome 2	Autosomal recessive
COLQ	603033	3		603034	Myasthenic syndrome, congenital, type 5	Autosomal recessive
COQ2	609825	4		607426	Primary coenzyme Q10 deficiency, type 1	Autosomal recessive
COQ4	612898	9		616276	Coenzyme Q10 deficiency, primary, type 7	Autosomal recessive
COQ6	614647	14		614650	Coenzyme Q10 deficiency, primary, type 6	Autosomal recessive
COQ8A	606980	1	ADCK3	612016	Primary coenzyme Q10 deficiency, type 4	Autosomal recessive
COQ8B	615567	19	ADCK4	615573	Nephrotic syndrome, type 9	Autosomal recessive
COQ9	612837	16		614654	Coenzyme Q10 deficiency, primary, type 5	Autosomal recessive
CORO1A	605000	16		615401	Immunodeficiency, type 8	Autosomal recessive
COX10	602125	17		619046	Mitochondrial complex IV deficiency, nuclear type 3	Autosomal recessive
COX15	603646	10		615119; 256000	Cardioencephalomyopathy, fatal infantile, due to cytochrome c oxidase deficiency, type 2; Leigh syndrome due to cytochrome c oxidase deficiency	Autosomal recessive
COX20	614698	1		619054	Mitochondrial complex IV deficiency, nuclear type 11	Autosomal recessive
COX6B1	124089	19		619051	Mitochondrial complex IV deficiency, nuclear type 7	Autosomal recessive
CP	117700	3		604290	Aceruloplasminemia	Autosomal recessive
CPA6	609562	8		614418	Febrile seizures, familial, type 11	Autosomal recessive
CPAMD8	608841	19		617319	Anterior segment dysgenesis, type 8	Autosomal recessive
CPLANE1	614571	5	C5orf42	614615	Joubert syndrome 17	Autosomal recessive
CPLX1	605032	4		617976	Epileptic encephalopathy, early infantile, 63	Autosomal recessive
CPS1	608307	2		237300	Carbamoylphosphate synthetase 1 deficiency	Autosomal recessive

CPT1A	600528	11		255120	Carnitine palmitoyltransferase type 1A deficiency, hepatic	Autosomal recessive
CPT2	600650	1		608836; 600649	Carnitine palmitoyltransferase type 2 deficiency, lethal neonatal; Carnitine palmitoyltransferase type 2 deficiency, infantile	Autosomal recessive
CR2	120650	1		614699	Immunodeficiency, common variable, type 7	Autosomal recessive
CRADD	603454	12		614499	Mental retardation, autosomal recessive, type 34, with variant lissencephaly	Autosomal recessive
CRB1	604210	1		600105; 613835	Retinitis pigmentosa, type 12; Leber congenital amaurosis, type 8	Autosomal recessive
CRB2	609720	9		219730	Ventriculomegaly with cystic kidney disease	Autosomal recessive
CRBN	609262	3		607417	Mental retardation, autosomal recessive, type 2	Autosomal recessive
CRIPT	604594	2		615789	Short stature with microcephaly and distinctive facies	Autosomal recessive
CRLF1	604237	19		272430	Cold-induced sweating syndrome type 1	Autosomal recessive
CRPPA	614631	7	ISPD	614643; 616052	Muscular dystrophy-dystroglycanopathy, type A7; Muscular dystrophy-dystroglycanopathy, type C7	Autosomal recessive
CRTAP	605497	3		610682	Osteogenesis imperfecta, type 7	Autosomal recessive
CRYAA	123580	21		604219*	Cataract 9, multiple types	Autosomal recessive*
CRYAB	123590	11		613869; 613763*	Myopathy, myofibrillar, fatal infantile hypertonic, alpha-B crystallin-related; Cataract 16, multiple types	Autosomal recessive; Autosomal recessive*
CRYBB1	600929	22		611544*	Cataract 17	Autosomal recessive*
CRYBB3	123630	22		609741	Cataract 22	Autosomal recessive
CSF2RB	138981	22		614370	Surfactant metabolism dysfunction, pulmonary, type 5	Autosomal recessive
CSF3R	138971	1		617014	Neutropenia, severe congenital, type 7, autosomal recessive	Autosomal recessive
CSPP1	611654	8		615636	Joubert syndrome 21	Autosomal recessive
CSTA	184600	3		607936	Peeling skin syndrome, type 4	Autosomal recessive
CSTB	601145	21		254800	Epilepsy, progressive myoclonic type 1A (Unverricht and Lundborg)	Autosomal recessive
CTC1	613129	17		612199	Cerebroretinal microangiopathy with calcifications and cysts	Autosomal recessive
CTH	607657	1		219500	Cystathioninuria	Autosomal recessive
CTNS	606272	17		219800	Nephropathic cystinosis	Autosomal recessive
CTPS1	123860	1		615897	Immunodeficiency, type 24	Autosomal recessive
CTSA	613111	20		256540	Galactosialidosis	Autosomal recessive
CTSC	602365	11		245010; 245000	Haim-Munk syndrome; Papillon-Lefevre syndrome	Autosomal recessive
CTSD	116840	11		610127	Ceroid lipofuscinosis, neuronal, type 10	Autosomal recessive
CTSF	603539	11		615362	Ceroid lipofuscinosis, neuronal, type 13 (Kufs type)	Autosomal recessive
CTSK	601105	1		265800	Pycnodysostosis	Autosomal recessive
CUBN	602997	10		261100	Megaloblastic anemia 1 (Imerslund-Grasbeck syndrome)	Autosomal recessive
CUL4B	300304	X		300354	Mental retardation, X-linked, syndromic, type 15 (Cabezas type)	X-linked
CUL7	609577	6		273750	3M syndrome 1	Autosomal recessive
CWC27	617170	5		250410	Retinitis pigmentosa with or without skeletal anomalies	Autosomal recessive
CWF19L1	616120	10		616127	Spinocerebellar ataxia, autosomal recessive, type 17	Autosomal recessive
CYB5A	613218	18		250790	46,XY disorder of sex development due to isolated 17,20-lyase deficiency	Autosomal recessive
CYB5R3	613213	22		250800	Methemoglobinemia, type 1; Methemoglobinemia, type 2	Autosomal recessive
CYBA	608508	16		233690	Chronic granulomatous disease, type 4	Autosomal recessive
CYBB	300481	X		306400	Chronic granulomatous disease, X-linked	X-linked
CYC1	123980	8		615453	Mitochondrial complex III deficiency, nuclear type 6	Autosomal recessive
CYP11A1	118485	15		613743	46,XY disorder of sex development-adrenal insufficiency due to CYP11A1 deficiency	Autosomal recessive
CYP11B1	610613	8		202010	Adrenal hyperplasia, congenital, due to 11-beta-hydroxylase deficiency	Autosomal recessive
CYP11B2	124080	8		203400	Hypoaldosteronism, congenital, due to CMO I deficiency	Autosomal recessive
CYP17A1	609300	10		202110	17 alpha(?)-hydroxylase/17,20-lyase deficiency	Autosomal recessive
CYP19A1	107910	15		613546	Aromatase deficiency	Autosomal recessive
CYP1B1	601771	2		231300	Glaucoma, primary congenital, type 3A	Autosomal recessive
CYP21A2	613815	6		201910	Congenital adrenal hyperplasia due to 21-hydroxylase deficiency	Autosomal recessive
CYP24A1	126065	20		143880	Hypercalcemia, infantile, type 1	Autosomal recessive
CYP26B1	605207	2		614416	Craniosynostosis with radiohumeral fusions and other skeletal and craniofacial anomalies	Autosomal recessive
CYP26C1	608428	10		614974	Focal facial dermal dysplasia 4	Autosomal recessive
CYP27A1	606530	2		213700	Cerebrotendinous xanthomatosis	Autosomal recessive
CYP27B1	609506	12		264700	Vitamin D-dependent rickets, type 1	Autosomal recessive
CYP2R1	608713	11		600081	Rickets due to defect in vitamin D 25-hydroxylation	Autosomal recessive
CYP2U1	610670	4		615030	Spastic paraplegia, type 56, autosomal recessive	Autosomal recessive

CYP4F22	611495	19		604777	Ichthyosis, congenital, autosomal recessive, type 5	Autosomal recessive
CYP4V2	608614	4		210370	Bietti crystalline corneoretinal dystrophy	Autosomal recessive
CYP7B1	603711	8		270800	Spastic paraplegia, type 5A, autosomal recessive	Autosomal recessive
D2HGDH	609186	2		600721	D-2-hydroxyglutaric aciduria	Autosomal recessive
DAG1	128239	3		616538; 613818	Muscular dystrophy-dystroglycanopathy type A9; Muscular dystrophy-dystroglycanopathy type C9	Autosomal recessive
DARS1	603084	2	DARS	615281	Hypomyelination with brainstem and spinal cord involvement and leg spasticity	Autosomal recessive
DARS2	610956	1		611105	Leukoencephalopathy with brain stem and spinal cord involvement and lactate elevation	Autosomal recessive
DBH	609312	9		223360	Dopamine beta-hydroxylase deficiency	Autosomal recessive
DBT	248610	1		248600	Maple syrup urine disease, type 2	Autosomal recessive
DCAF17	612515	2		241080	Woodhouse-Sakati syndrome	Autosomal recessive
DCC	120470	18		617542	Gaze palsy, familial horizontal, with progressive scoliosis, type 2	Autosomal recessive
DCDC2	605755	6		617394; 616217	Sclerosing cholangitis, neonatal; Nephronophthisis 19	Autosomal recessive
DCHS1	603057	11		601390	Van Maldergem syndrome 1	Autosomal recessive
DCLRE1C	605988	10		603554; 602450	Omenn syndrome; Severe combined immunodeficiency, Athabascan type	Autosomal recessive
DCPS	610534	11		616459	Al-Raqad syndrome	Autosomal recessive
DCX	300121	X		300067	Lissencephaly, X-linked, type 1	X-linked
DDB2	600811	11		278740	Xeroderma pigmentosum, complementation group E	Autosomal recessive
DDC	107930	7		608643	Aromatic L-amino acid decarboxylase deficiency	Autosomal recessive
DDHD1	614603	14		609340	Spastic paraplegia, type 28, autosomal recessive	Autosomal recessive
DDHD2	615003	8		615033	Spastic paraplegia, type 54, autosomal recessive	Autosomal recessive
DDR2	191311	1		271665	Spondylometaepiphyseal dysplasia, short limb-hand type	Autosomal recessive
DDRGK1	616177	20		602557	Spondyloepimetaphyseal dysplasia, Shohat type	Autosomal recessive
DDX11	601150	12		613398	Warsaw breakage syndrome	Autosomal recessive
DDX59	615464	1		174300	Orofaciodigital syndrome V	Autosomal recessive
DENND5A	617278	11		617281	Epileptic encephalopathy, early infantile, 49	Autosomal recessive
DES	125660	2		601419*	Myopathy, myofibrillar, type 1	Autosomal recessive*
DGAT1	604900	8		615863	?Diarrhea 7, protein-losing enteropathy type	Autosomal recessive
DGKE	601440	17		615008	Nephrotic syndrome, type 7	Autosomal recessive
DGUOK	601465	2		251880	DGUOK-related mitochondrial DNA depletion syndrome	Autosomal recessive
DHCR24	606418	1		602398	Desmosterolosis	Autosomal recessive
DHCR7	602858	11		270400	Smith-Lemli-Opitz syndrome	Autosomal recessive
DHDDS	608172	1		613861	Retinitis pigmentosa, type 59	Autosomal recessive
DHFR	126060	5		613839	Megaloblastic anemia due to dihydrofolate reductase deficiency	Autosomal recessive
DHH	605423	12		233420	46,XY complete gonadal dysgenesis	Autosomal recessive
DHODH	126064	16		263750	Miller syndrome	Autosomal recessive
DHPS	600944	19		618480	Neurodevelopmental disorder with seizures and speech and walking impairment	Autosomal recessive
DHTKD1	614984	10		204750	2-aminoadipic 2-oxoadipic aciduria	Autosomal recessive
DIAPH1	602121	5		616632	Seizures, cortical blindness, microcephaly syndrome	Autosomal recessive
DIS3L2	614184	2		267000	Perlman syndrome	Autosomal recessive
DKC1	300126	X		305000	Dyskeratosis congenita, X-linked	X-linked
DLAT	608770	11		245348	Pyruvate dehydrogenase E2 deficiency	Autosomal recessive
DLD	238331	7		246900	Dihydrolipoamide dehydrogenase deficiency	Autosomal recessive
DLG3	300189	X		300850	Mental retardation, X-linked, type 90	X-linked
DLL3	602768	19		277300	Spondylocostal dysostosis type 1	Autosomal recessive
DMD	300377	X		310200; 300376	Duchenne/Becker muscular dystrophy	X-linked
DMGDH	605849	5		605850	Dimethylglycine dehydrogenase deficiency	Autosomal recessive
DMP1	600980	4		241520	Hypophosphatemic rickets, autosomal recessive	Autosomal recessive
DMXL2	612186	15		618663	Developmental and epileptic encephalopathy, type 81	Autosomal recessive
DNAAF1	613190	16		613193	Ciliary dyskinesia, primary, type 13	Autosomal recessive
DNAAF2	612517	14		612518	Ciliary dyskinesia, primary, type 10	Autosomal recessive
DNAAF3	614566	19		606763	Ciliary dyskinesia, primary, type 2	Autosomal recessive
DNAAF4	608706	15	DYX1C1	615482	Ciliary dyskinesia, primary, type 25	Autosomal recessive
DNAAF5	614864	7	HEATR2	614874	Ciliary dyskinesia, primary, type 18	Autosomal recessive
DNAH1	603332	3		617576	Spermatogenic failure, type 18	Autosomal recessive
DNAH11	603339	7		611884	Ciliary dyskinesia, primary, type 7, with or without situs inversus	Autosomal recessive

DNAH5	603335	5		608644	Ciliary dyskinesia, primary, type 3, with or without situs inversus	Autosomal recessive
DNAH9	603330	17		618300	Ciliary dyskinesia, primary, type 40	Autosomal recessive
DNAI1	604366	9		244400	Ciliary dyskinesia, primary, type 1, with or without situs inversus	Autosomal recessive
DNAI2	605483	17		612444	Ciliary dyskinesia, primary, type 9, with or without situs inversus	Autosomal recessive
DNAJB13	610263	11		617091	Ciliary dyskinesia, primary, type 34	Autosomal recessive
DNAJB2	604139	2		614881	Spinal muscular atrophy, distal, autosomal recessive, type 5	Autosomal recessive
DNAJC12	606060	10		617384	Hyperphenylalaninemia, mild, non-BH4-deficient	Autosomal recessive
DNAJC19	608977	3		610198	3-methylglutaconic aciduria, type 5	Autosomal recessive
DNAJC21	617048	5		617052	Bone marrow failure syndrome, type 3	Autosomal recessive
DNAJC6	608375	1		615528	Parkinson disease, type 19A, juvenile-onset; Parkinson disease, type 19B, early-onset	Autosomal recessive
DNAL1	610062	14		614017	Ciliary dyskinesia, primary, type 16	Autosomal recessive
DNASE1L3	602244	3		614420	Systemic lupus erythematosus 16	Autosomal recessive
DNM1L	603850	12		614388*	Encephalopathy due to defective mitochondrial and peroxisomal fission, type 1	Autosomal recessive*
DNM2	602378	19		615368	Lethal congenital contracture syndrome, type 5	Autosomal recessive
DNMT3B	602900	20		242860	Immunodeficiency-centromeric instability-facial anomalies syndrome, type 1	Autosomal recessive
DOCK2	603122	5		616433	Immunodeficiency, type 40	Autosomal recessive
DOCK6	614194	19		614219	Adams-Oliver syndrome 2	Autosomal recessive
DOCK7	615730	1		615859	Epileptic encephalopathy, early infantile, 23	Autosomal recessive
DOCK8	611432	9		243700	Hyper-IgE recurrent infection syndrome, autosomal recessive	Autosomal recessive
DOK7	610285	4		618389; 254300	Fetal akinesia deformation sequence, type 3; Myasthenic syndrome, congenital, type 10	Autosomal recessive
DOLK	610746	9		610768	Congenital disorder of glycosylation, type 1M	Autosomal recessive
DONSON	611428	21		617604	Microcephaly, short stature, and limb abnormalities	Autosomal recessive
DPAGT1	191350	11		608093; 614750	Congenital disorder of glycosylation, type 1J; Myasthenic syndrome, congenital, type 13	Autosomal recessive
DPH1	603527	17		616901	Developmental delay with short stature, dysmorphic features, and sparse hair	Autosomal recessive
DPM1	603503	20		608799	Congenital disorder of glycosylation, type 1E	Autosomal recessive
DPM2	603564	9		615042	Congenital disorder of glycosylation, type Iu	Autosomal recessive
DPM3	605951	1		612937	Congenital disorder of glycosylation, type Io	Autosomal recessive
DPY19L2	613893	12		613958	Spermatogenic failure, type 9	Autosomal recessive
DPYD	612779	1		274270	Dihydropyrimidine dehydrogenase deficiency	Autosomal recessive
DPYS	613326	8		222748	Dihydropyrimidinuria	Autosomal recessive
DRAM2	613360	1		616502	Cone-rod dystrophy 21	Autosomal recessive
DRC1	615288	2		615294	Ciliary dyskinesia, primary, type 21	Autosomal recessive
DSG1	125670	18		615508	Erythroderma, congenital, with palmoplantar keratoderma, hypotrichosis, and hyper IgE	Autosomal recessive
DSG4	607892	18		607903	Hypotrichosis, type 6	Autosomal recessive
DSP	125647	6		605676; 609638	Cardiomyopathy, dilated, with woolly hair and keratoderma; Epidermolysis bullosa, lethal acantholytic	Autosomal recessive
DST	113810	6		615425	Epidermolysis bullosa simplex, autosomal recessive, type 2	Autosomal recessive
DSTYK	612666	1		270750	Spastic paraplegia, type 23, autosomal recessive	Autosomal recessive
DTNBP1	607145	6		614076	Hermansky-Pudlak syndrome, type 7	Autosomal recessive
DUOX2	606759	15		607200	Thyroid dyshormonogenesis, type 6	Autosomal recessive
DUOXA2	612772	15		274900	Thyroid dyshormonogenesis, type 5	Autosomal recessive
DYM	607461	18		607326; 223800	Smith-McCort dysplasia; Dyggve-Melchior-Clausen disease	Autosomal recessive
DYNC2H1	603297	11		613091	Short-rib thoracic dysplasia, type 3, with or without polydactyly	Autosomal recessive
DYNC2I1	615462	7	WDR60	615503	Short-rib thoracic dysplasia 8 with or without polydactyly	Autosomal recessive
DYNC2I2	613363	9	WDR34	615633	Short-rib thoracic dysplasia 11 with or without polydactyly	Autosomal recessive
DYNC2LI1	617083	2		617088	Short-rib thoracic dysplasia 15 with polydactyly	Autosomal recessive
DYNLT2B	617353	3	TCTEX1D2	617405	Short-rib thoracic dysplasia 17 with or without polydactyly	Autosomal recessive
DYSF	603009	2		254130; 253601	Miyoshi muscular dystrophy, type 1; Limb-girdle muscular dystrophy, type 2 (LGMD R2)	Autosomal recessive
DZIP1L	617570	3		617610	Polycystic kidney disease 5	Autosomal recessive
EARS2	612799	16		614924	Combined oxidative phosphorylation deficiency 12	Autosomal recessive
ECEL1	605896	2		615065	Arthrogryposis, distal, type 5D	Autosomal recessive
ECHS1	602292	10		616277	Mitochondrial short-chain enoyl-CoA hydratase 1 deficiency	Autosomal recessive
ECM1	602201	1		247100	Urbach-Wiethe disease	Autosomal recessive
EDA	300451	X		305100	Ectodermal dysplasia, type 1, hypohidrotic, X-linked	X-linked
EDAR	604095	2		224900	Ectodermal dysplasia 10B, hypohidrotic/hair/tooth type	Autosomal recessive
EDARADD	606603	1		614941	Ectodermal dysplasia 11B, hypohidrotic/hair/tooth type	Autosomal recessive

EDN1	131240	6		615706	Auriculocondylar syndrome, type 3	Autosomal recessive
EDN3	131242	20		613265	Waardenburg syndrome, type 4B	Autosomal recessive
EDNRB	131244	13		600501	ABCD syndrome	Autosomal recessive
EFEMP2	604633	11		614437	Cutis laxa, autosomal recessive, type 1B	Autosomal recessive
EFL1	617538	15	EFTUD1	617941	Shwachman-Diamond syndrome 2	Autosomal recessive
EGFR	131550	7		616069	?Inflammatory skin and bowel disease, neonatal, 2	Autosomal recessive
EGR2	129010	10		145900*	Dejerine-Sottas disease	Autosomal recessive*
EIF2AK3	604032	2		226980	Wolcott-Rallison syndrome	Autosomal recessive
EIF2AK4	609280	15		234810	Pulmonary venoocclusive disease 2	Autosomal recessive
EIF2B1	606686	12		603896	Leukoencephalopathy with vanishing white matter	Autosomal recessive
EIF2B2	606454	14		603896	Leukoencephalopathy with vanishing white matter	Autosomal recessive
EIF2B3	606273	1		603896	Leukoencephalopathy with vanishing white matter	Autosomal recessive
EIF2B4	606687	2		603896	Leukoencephalopathy with vanishing white matter	Autosomal recessive
EIF2B5	603945	3		603896	Leukoencephalopathy with vanishing white matter	Autosomal recessive
EIF4A3	608546	17		268305	Robin sequence with cleft mandible and limb anomalies	Autosomal recessive
ELAC2	605367	17		615440	Combined oxidative phosphorylation deficiency 17	Autosomal recessive
ELMO2	606421	20		606893	Vascular malformation, primary intraosseous	Autosomal recessive
ELOVL4	605512	6		614457	Ichthyosis, spastic quadriplegia, and mental retardation	Autosomal recessive
ELP1	603722	9	IKBKAP	223900	Familial dysautonomia	Autosomal recessive
ELP2	616054	18		617270	Mental retardation, autosomal recessive, type 58	Autosomal recessive
EMC1	616846	1		616875	Cerebellar atrophy, visual impairment, and psychomotor retardation	Autosomal recessive
EMD	300384	X		310300	Emery-Dreifuss muscular dystrophy, type 1, X-linked	X-linked
EML1	602033	14		600348	Band heterotopia	Autosomal recessive
EMP2	602334	16		615861	Nephrotic syndrome, type 10	Autosomal recessive
ENAM	606585	4		204650	Amelogenesis imperfecta, type 1C	Autosomal recessive
ENO3	131370	17		612932	?Glycogen storage disease XIII	Autosomal recessive
ENPP1	173335	6		208000	Arterial calcification, generalized, of infancy, type 1	Autosomal recessive
ENTPD1	601752	10		615683	Spastic paraplegia, type 64, autosomal recessive	Autosomal recessive
EOGT	614789	3		615297	Adams-Oliver syndrome 4	Autosomal recessive
EPB41	130500	1		611804*	Elliptocytosis, type 1	Autosomal recessive*
EPB42	177070	15		612690	Spherocytosis, type 5	Autosomal recessive
EPCAM	185535	2		613217	Diarrhea 5, with tufting enteropathy, congenital	Autosomal recessive
EPG5	615068	18		242840	Vici syndrome	Autosomal recessive
EPM2A	607566	6		254780	Epilepsy, progressive myoclonic, type 2A (Lafora)	Autosomal recessive
EPRS1	138295	1	EPRS	617951	Leukodystrophy, hypomyelinating, type 15	Autosomal recessive
EPS8L2	614988	11		617637	Deafness autosomal recessive, type 106	Autosomal recessive
ERAL1	607435	17		617565	Perrault syndrome 6	Autosomal recessive
ERBB3	190151	12		607598	Lethal congenital contractural syndrome, type 2	Autosomal recessive
ERCC1	126380	19		610758	Cerebrooculofacioskeletal syndrome, type 4	Autosomal recessive
ERCC2	126340	19		601675	Trichothiodystrophy, type 1	Autosomal recessive
ERCC3	133510	2		616390	Trichothiodystrophy, type 2	Autosomal recessive
ERCC4	133520	16		615272	Fanconi anemia, complementation group Q	Autosomal recessive
ERCC5	133530	13		616570	Cerebrooculofacioskeletal syndrome, type 3	Autosomal recessive
ERCC6	609413	10		133540; 214150	Cockayne syndrome, type B; Cerebrooculofacioskeletal syndrome, type 1	Autosomal recessive
ERCC6L2	615667	9		615715	Bone marrow failure syndrome, type 2	Autosomal recessive
ERCC8	609412	5		216400	Cockayne syndrome, type A	Autosomal recessive
ERLIN1	611604	10		615681	Spastic paraplegia, type 62, autosomal recessive	Autosomal recessive
ERLIN2	611605	8		611225	Spastic paraplegia, type 18, autosomal recessive	Autosomal recessive
ESCO2	609353	8		268300	Roberts syndrome	Autosomal recessive
ESPN	606351	1		609006	Deafness, autosomal recessive, type 36	Autosomal recessive
ESR1	133430	6		615363	Estrogen resistance	Autosomal recessive
ESRRB	602167	14		608565	Deafness, autosomal recessive, type 35	Autosomal recessive
ETFA	608053	15		231680	Glutaric acidemia, type 2A	Autosomal recessive
ETFB	130410	19		231680	Glutaric acidemia, type 2B	Autosomal recessive
ETFDH	231675	4		231680	Glutaric acidemia, type 2C	Autosomal recessive

ETHE1	608451	19	602473	Ethylmalonic encephalopathy	Autosomal recessive
EVC	604831	4	225500	Ellis-van Creveld syndrome	Autosomal recessive
EVC2	607261	4	225500	Ellis-van Creveld syndrome	Autosomal recessive
EXOSC3	606489	9	614678	Pontocerebellar hypoplasia, type 1B	Autosomal recessive
EXPH5	612878	11	615028	Epidermolysis bullosa, nonspecific, autosomal recessive	Autosomal recessive
EXT1	608177	8	215300	Chondrosarcoma	Autosomal recessive
EXTL3	605744	8	617425	Immunoskeletal dysplasia with neurodevelopmental abnormalities	Autosomal recessive
EYS	612424	6	602772	Retinitis pigmentosa, type 25	Autosomal recessive
F10	613872	13	227600	Factor X deficiency	Autosomal recessive
F11	264900	4	612416*	Factor XI deficiency	Autosomal recessive*
F13A1	134570	6	613225	Factor XIIIA deficiency	Autosomal recessive
F13B	134580	1	613235	Factor XIIIB deficiency	Autosomal recessive
F2	176930	11	613679	Prothrombin deficiency	Autosomal recessive
F5	612309	1	227400	Factor V deficiency	Autosomal recessive
F7	613878	13	227500	Factor VII deficiency	Autosomal recessive
F8	300841	X	306700	Hemophilia A	X-linked
F9	300746	X	306900	Hemophilia B	X-linked
FA2H	611026	16	612319	Spastic paraplegia, type 35, autosomal recessive	Autosomal recessive
FADD	602457	11	613759	Infections, recurrent, with encephalopathy, hepatic dysfunction, and cardiovascular malformations	Autosomal recessive
FAH	613871	15	276700	Tyrosinemia, type 1	Autosomal recessive
FAM126A	610531	7	610532	Leukodystrophy, hypomyelinating, type 5	Autosomal recessive
FAM161A	613596	2	606068	Retinitis pigmentosa, type 28	Autosomal recessive
FAM20A	611062	17	204690	Amelogenesis imperfecta, type 1G (Enamel-renal syndrome)	Autosomal recessive
FAM20C	611061	7	259775	Raine syndrome	Autosomal recessive
FAN1	613534	15	614817	Interstitial nephritis, karyomegalic	Autosomal recessive
FANCA	607139	16	227650	Fanconi anemia, complementation group A	Autosomal recessive
FANCC	613899	9	227645	Fanconi anemia, complementation group C	Autosomal recessive
FANCD2	613984	3	227646	Fanconi anemia, complementation group D2	Autosomal recessive
FANCE	613976	6	600901	Fanconi anemia, complementation group E	Autosomal recessive
FANCF	613897	11	603467	Fanconi anemia, complementation group F	Autosomal recessive
FANCG	602956	9	614082	Fanconi anemia, complementation group G	Autosomal recessive
FANCI	611360	15	609053	Fanconi anemia, complementation group I	Autosomal recessive
FANCL	608111	2	614083	Fanconi anemia, complementation group L	Autosomal recessive
FANCM	609644	14	618086	Spermatogenic failure, type 28	Autosomal recessive
FAR1	616107	11	616154	Peroxisomal fatty acyl-CoA reductase 1 disorder	Autosomal recessive
FARS2	611592	6	614946; 617046	Combined oxidative phosphorylation deficiency 14; Spastic paraplegia, type 77, autosomal recessive	Autosomal recessive
FASTKD2	612322	2	618855	Combined oxidative phosphorylation deficiency 44	Autosomal recessive
FAT4	612411	4	616006	Hennekam lymphangiectasia-lymphedema syndrome 2	Autosomal recessive
FBLN5	604580	14	219100	Cutis laxa, autosomal recessive, type 1A	Autosomal recessive
FBP1	611570	9	229700	Fructose-1,6-bisphosphatase deficiency	Autosomal recessive
FBXL4	605654	6	615471	Mitochondrial DNA depletion syndrome 13 (encephalomyopathic type)	Autosomal recessive
FBXO7	605648	22	260300	Parkinson disease, type 15, autosomal recessive	Autosomal recessive
FDXR	103270	17	617717	Auditory neuropathy and optic atrophy	Autosomal recessive
FECH	612386	18	177000	Protoporphyria, erythropoietic, autosomal recessive	Autosomal recessive
FERMT1	607900	20	173650	Kindler syndrome	Autosomal recessive
FERMT3	607901	11	612840	Leukocyte adhesion deficiency, type 3	Autosomal recessive
FEZF1	613301	7	616030	Hypogonadotropic hypogonadism type 22, with or without anosmia	Autosomal recessive
FGA	134820	4	202400	Afibrinogenemia, congenital	Autosomal recessive
FGB	134830	4	202400	Congenital afibrinogenemia	Autosomal recessive
FGD1	300546	X	305400	Aarskog-Scott syndrome; Mental retardation, X-linked syndromic, type 16	X-linked
FGD4	611104	12	609311	Charcot-Marie-Tooth disease, type 4H	Autosomal recessive
FGF23	605380	12	617993	Tumoral calcinosis, hyperphosphatemic, familial, type 2	Autosomal recessive
FGF3	164950	11	610706	Deafness, congenital with inner ear agenesis, microtia, and microdontia	Autosomal recessive
FGG	134850	4	202400	Afibrinogenemia, congenital; Hypofibrinogenemia, congenital	Autosomal recessive
FH	136850	1	606812	Fumarase deficiency	Autosomal recessive

FIBP	608296	11	617107	Thauvin-Robinet-Faivre syndrome	Autosomal recessive
FIG4	609390	6	611228; 216340	Charcot-Marie-Tooth disease, type 4J; Yunis-Varon syndrome	Autosomal recessive
FKBP10	607063	17	259450	Bruck syndrome 1	Autosomal recessive
FKBP14	614505	7	614557	Ehlers-Danlos syndrome, kyphoscoliotic type, 2	Autosomal recessive
FKRP	606596	19	613153; 606612; 607155	Muscular dystrophy-dystroglycanopathy, type 5A (Walker-Warburg syndrome); Type 5B; Type 5C (limb-girdle muscular dystrophy, type 9 [LGMDR9])	Autosomal recessive
FKTN	607440	9	253800; 613152; 611588	Muscular dystrophy-dystroglycanopathy, type 4A (Walker-Warburg syndrome); Type 4B; Type 4C (limb-girdle muscular dystrophy, type 13 [LGMD R13])	Autosomal recessive
FLAD1	610595	1	255100	Lipid storage myopathy due to flavin adenine dinucleotide synthetase deficiency	Autosomal recessive
FLG	135940	1	146700	Ichthyosis vulgaris	Autosomal recessive*
FLI1	193067	11	617443*	Bleeding disorder, platelet-type, type 21	Autosomal recessive*
FLNB	603381	3	272460	Spondylocarpotarsal synostosis syndrome	Autosomal recessive
FLVCR1	609144	1	609033	Posterior column ataxia-retinitis pigmentosa syndrome	Autosomal recessive
FLVCR2	610865	14	225790	Proliferative vasculopathy and hydranencephaly-hydrocephaly syndrome	Autosomal recessive
FMN2	606373	1	616193	Mental retardation, autosomal recessive, type 47	Autosomal recessive
FMO3	136132	1	602079	Trimethylaminuria	Autosomal recessive
FMR1	309550	X	300624	Fragile X syndrome	X-linked
FOLR1	136430	11	613068	Neurodegeneration due to cerebral folate transport deficiency	Autosomal recessive
FOXE1	602617	9	241850	Bamforth-Lazarus syndrome	Autosomal recessive
FOXE3	601094	1	610256	Anterior segment dysgenesis, type 2, multiple subtypes	Autosomal recessive
FOXN1	600838	17	601705	T-cell immunodeficiency, congenital alopecia and nail dystrophy	Autosomal recessive
FOXRED1	613622	11	618241	Mitochondrial complex I deficiency, nuclear type 19	Autosomal recessive
FRAS1	607830	4	219000	Fraser syndrome, type 1	Autosomal recessive
FREM1	608944	9	248450	Manitoba oculotrichoanal syndrome	Autosomal recessive
FREM2	608945	13	617666	Fraser syndrome, type 2	Autosomal recessive
FRRS1L	604574	9	616981	Epileptic encephalopathy, early infantile, 37	Autosomal recessive
FSHB	136530	11	229070	Hypogonadotropic hypogonadism, type 24, without anosmia	Autosomal recessive
FSHR	136435	2	233300	Ovarian dysgenesis 1	Autosomal recessive
FTCD	606806	21	229100	Glutamate formiminotransferase deficiency	Autosomal recessive
FTL	134790	19	615604*	L-ferritin deficiency	Autosomal recessive*
FTO	610966	16	612938	Growth retardation, developmental delay, facial dysmorphism	Autosomal recessive
FTSJ1	300499	X	309549	Mental retardation, X-linked 44	X-linked
FUCA1	612280	1	230000	Fucosidosis	Autosomal recessive
FUT8	602589	14	618005	Congenital disorder of glycosylation with defective fucosylation, type 1	Autosomal recessive
FYCO1	607182	3	610019	Cataract 18	Autosomal recessive
FZD6	603409	8	614157	Nail disorder, nonsyndromic congenital, type 10 (claw-shaped nails)	Autosomal recessive
G6PC	613742	17	232200	Glycogen storage disease, type 1A	Autosomal recessive
G6PC3	611045	17	612541	Dursun syndrome	Autosomal recessive
G6PD	305900	X	300908	Hemolytic anemia, G6PD deficient (favism)	X-linked
GAA	606800	17	232300	Glycogen storage disease, type 2	Autosomal recessive
GALC	606890	14	245200	Krabbe disease	Autosomal recessive
GALE	606953	1	230350	Galactose epimerase deficiency	Autosomal recessive
GALK1	604313	17	230200	Galactokinase deficiency with cataracts	Autosomal recessive
GALNS	612222	16	253000	Mucopolysaccharidosis, type 4A	Autosomal recessive
GALNT3	601756	2	211900	Tumoral calcinosis, hyperphosphatemic, familial, type 1	Autosomal recessive
GALT	606999	9	230400	Galactosemia	Autosomal recessive
GAMT	601240	19	612736	Cerebral creatine deficiency syndrome, type 2	Autosomal recessive
GAN	605379	16	256850	Giant axonal neuropathy, type 1	Autosomal recessive
GAS8	605178	16	616726	Ciliary dyskinesia, primary, type 33	Autosomal recessive
GATM	602360	15	612718	Cerebral creatine deficiency syndrome, type 3	Autosomal recessive
GBA	606463	1	230800	Gaucher disease	Autosomal recessive
GBA2	609471	9	614409	Spastic paraplegia, type 46, autosomal recessive	Autosomal recessive
GBE1	607839	3	232500	Glycogen storage disease, type 4	Autosomal recessive
GCDH	608801	19	231670	Glutaricaciduria, type 1	Autosomal recessive
GCH1	600225	14	233910	Hyperphenylalaninemia, BH4-deficient, type B	Autosomal recessive

GCK	138079	7	606176*	Permanent neonatal diabetes mellitus (PNDM)	Autosomal recessive*
GCM2	603716	6	618883	Hypoparathyroidism, familial isolated (FIH) 2	Autosomal recessive
GCNT2	600429	6	116700	Cataract 13, with adult i phenotype	Autosomal recessive
GCSH	238330	16	605899	?Glycine encephalopathy	Autosomal recessive
GDAP1	606598	8	608340	Charcot-Marie-Tooth disease, recessive intermediate, type A	Autosomal recessive
GDF1	602880	19	208530	Right atrial isomerism (Ivemark syndrome)	Autosomal recessive
GDF5	601146	20	200700	Chondrodysplasia, Grebe type	Autosomal recessive
GDF6	601147	8	615360	Leber congenital amaurosis, type 17	Autosomal recessive
GFER	600924	16	613076	Myopathy, mitochondrial progressive, with congenital cataract, hearing loss, and developmental delay	Autosomal recessive
GFM1	606639	3	609060	Combined oxidative phosphorylation deficiency, type 1	Autosomal recessive
GFPT1	138292	2	610542	Myasthenia, congenital, type 12, with tubular aggregates	Autosomal recessive
GGCX	137167	2	277450	Vitamin K-dependent clotting factors, combined deficiency of, type 1	Autosomal recessive
GH1	139250	17	262400; 262650	Growth hormone deficiency, isolated, type 1A; Kowarski syndrome	Autosomal recessive
GHR	600946	5	262500	Laron dwarfism	Autosomal recessive
GHRHR	139191	7	612781	Growth hormone deficiency, isolated, type 1B	Autosomal recessive
GHSR	601898	3	615925	Growth hormone deficiency, isolated partial	Autosomal recessive
GINS1	610608	20	617827	Immunodeficiency, type 55	Autosomal recessive
GIPC3	608792	19	601869	Deafness, autosomal recessive, type 15	Autosomal recessive
GJA1	121014	6	218400	Craniometaphyseal dysplasia, autosomal recessive	Autosomal recessive
GJB1	304040	X	302800	Charcot-Marie-Tooth neuropathy, X-linked dominant, type 1	X-linked
GJB2	121011	13	220290	Deafness, autosomal recessive, type 1A; Deafness, digenic, GJB2/GJB6	Autosomal recessive; Digenic inheritance (GJB6 gene)
GJB6	604418	13	612645; 220290	Deafness, autosomal recessive, type 1B; Deafness, digenic GJB2/GJB6	Autosomal recessive; Digenic inheritance (GJB2 gene)
GJC2	608803	1	613206	Spastic paraplegia, type 44, autosomal recessive	Autosomal recessive
GLA	300644	X	301500	Fabry disease	X-linked
GLB1	611458	3	230500, 230600, 230650; 253010	GM1-gangliosidosis, types 1-3; Mucopolysaccharidosis, type 4B (Morquio)	Autosomal recessive
GLDC	238300	9	605899	Glycine encephalopathy	Autosomal recessive
GLDN	608603	15	617194	Lethal congenital contracture syndrome 11	Autosomal recessive
GLE1	603371	9	253310; 611890	Lethal congenital contracture syndrome, type 1; Congenital arthrogryposis with anterior horn cell disease	Autosomal recessive
GLIS2	608539	16	611498	Nephronophthisis, type 7	Autosomal recessive
GLIS3	610192	9	610199	Diabetes mellitus, neonatal, with congenital hypothyroidism	Autosomal recessive
GLRA1	138491	5	149400*	Hyperekplexia, type 1	Autosomal recessive*
GLRB	138492	4	614619	Hyperekplexia, type 2	Autosomal recessive
GLRX5	609588	14	616860; 616859	Anemia, sideroblastic, type 3, pyridoxine-refractory; Spasticity, childhood-onset, with hyperglycinemia	Autosomal recessive
GLUL	138290	1	610015	Glutamine deficiency, congenital	Autosomal recessive
GLYCTK	610516	3	220120	D-glyceric aciduria	Autosomal recessive
GM2A	613109	5	272750	GM2-gangliosidosis, AB variant	Autosomal recessive
GMPPA	615495	2	615510	Alacrima, achalasia, and mental retardation syndrome	Autosomal recessive
GMPPB	615320	3	615351	Muscular dystrophy-dystroglycanopathy 14	Autosomal recessive
GNAT1	139330	3	616389	Night blindness, congenital stationary, type 1G	Autosomal recessive
GNAT2	139340	1	613856	Achromatopsia, type 4	Autosomal recessive
GNB5	604447	15	617173; 617182	Intellectual developmental disorder with cardiac arrhythmia; Language delay and ADHD/cognitive impairment with or without cardiac arrhythmia	Autosomal recessive
GNE	603824	9	605820	Inclusion body myopathy, type 2 (Nonaka myopathy)	Autosomal recessive
GNMT	606628	6	606664	Glycine N-methyltransferase deficiency	Autosomal recessive
GNPAT	602744	1	222765	Rhizomelic chondrodysplasia punctata, type 2	Autosomal recessive
GNPTAB	607840	12	252500; 252600	Mucolipidosis 2 alpha/beta; Mucolipidosis 3 alpha/beta	Autosomal recessive
GNPTG	607838	16	252605	Mucolipidosis III gamma	Autosomal recessive
GNRHR	138850	4	146110	Hypogonadotropic hypogonadism, type 7, without anosmia	Autosomal recessive
GNS	607664	12	252940	Mucopolysaccharidosis, type 3D (Sanfilippo syndrome D)	Autosomal recessive
GORAB	607983	1	231070	Geroderma osteodysplasticum	Autosomal recessive
GOSR2	604027	17	614018	Epilepsy, progressive myoclonic, type 6	Autosomal recessive
GOT2	138150	16	618721	Epileptic encephalopathy, early infantile, 82	Autosomal recessive

GP1BA	606672	17		231200	Bernard-Soulier syndrome, type A1	Autosomal recessive
GP1BB	138720	22		231200	Bernard-Soulier syndrome, type B	Autosomal recessive
GP6	605546	19		614201	Bleeding disorder, platelet-type, type 11	Autosomal recessive
GP9	173515	3		231200	Bernard-Soulier syndrome, type C	Autosomal recessive
GPAA1	603048	8		617810	Glycosylphosphatidylinositol biosynthesis defect 15	Autosomal recessive
GPC6	604404	13		258315	Omodysplasia, type 1	Autosomal recessive
GPD1	138420	12		614480	Hypertriglyceridemia, transient infantile	Autosomal recessive
GPHN	603930	14		615501	Molybdenum cofactor deficiency C	Autosomal recessive
GPI	172400	19		613470	Hemolytic anemia, nonspherocytic, due to glucose phosphate isomerase deficiency	Autosomal recessive
GPIHBP1	612757	8		615947	Hyperlipoproteinemia, type 1D	Autosomal recessive
GPR143	300808	X		300500	Ocular albinism, type 1 (Nettleship-Falls type)	X-linked
GPR179	614515	17		614565	Night blindness, congenital stationary (complete), type 1E, autosomal recessive	Autosomal recessive
GPR68	601404	14		617217	Amelogenesis imperfecta, type 2A6 (hypomaturation type)	Autosomal recessive
GPSM2	609245	1		604213	Chudley-McCullough syndrome	Autosomal recessive
GPT2	138210	16		616281	Mental retardation, autosomal recessive 49	Autosomal recessive
GPX4	138322	19		250220	Spondylometaphyseal dysplasia, Sedaghatian type	Autosomal recessive
GRHL2	608576	8		616029	Ectodermal dysplasia/short stature syndrome	Autosomal recessive
GRHPR	604296	9		260000	Hyperoxaluria, primary, type 2	Autosomal recessive
GRID2	602368	4		616204	Spinocerebellar ataxia, autosomal recessive, type 18	Autosomal recessive
GRIK2	138244	6		611092	Mental retardation, autosomal recessive, type, 6	Autosomal recessive
GRIN1	138249	9		617820	Neurodevelopmental disorder with or without hyperkinetic movements and seizures, autosomal recessive	Autosomal recessive
GRIP1	604597	12		617667	Fraser syndrome 3	Autosomal recessive
GRK1	180381	13		613411	Oguchi disease-2	Autosomal recessive
GRM1	604473	6		614831	Spinocerebellar ataxia, autosomal recessive, type 13	Autosomal recessive
GRM6	604096	5		257270	Night blindness, congenital stationary (complete), type 1B, autosomal recessive	Autosomal recessive
GRN	138945	17		614706	Ceroid lipofuscinosis, neuronal, type 11	Autosomal recessive
GRXCR1	613283	4		613285	Deafness, autosomal recessive, type 25	Autosomal recessive
GSC	138890	14		602471	Short stature, auditory canal atresia, mandibular hypoplasia, skeletal abnormalities	Autosomal recessive
GSS	601002	20		266130	Glutathione synthetase deficiency	Autosomal recessive
GTF2H5	608780	6		616395	Trichothiodystrophy, type 3, photosensitive	Autosomal recessive
GTPBP2	607434	6		617988	Jaberi-Elahi syndrome	Autosomal recessive
GTPBP3	608536	19		616198	Combined oxidative phosphorylation deficiency 23	Autosomal recessive
GUCY2C	601330	12		614665	Meconium ileus	Autosomal recessive
GUCY2D	600179	17		204000	Leber congenital amaurosis, type 1	Autosomal recessive
GUF1	617064	4		617065	?Epileptic encephalopathy, early infantile, 40	Autosomal recessive
GUSB	611499	7		253220	Mucopolysaccharidosis, type 7	Autosomal recessive
GYG1	603942	3		616199	Polyglucosan body myopathy, type 2	Autosomal recessive
GYS1	138570	19		611556	Glycogen storage disease, type 0, muscle	Autosomal recessive
GYS2	138571	12		240600	Glycogen storage disease, type 0, liver	Autosomal recessive
GZF1	613842	20		617662	Joint laxity, short stature, and myopia	Autosomal recessive
H6PD	138090	1		604931	Cortisone reductase deficiency 1	Autosomal recessive
HAAO	604521	2		617660	Vertebral, cardiac, renal, and limb defects syndrome 1	Autosomal recessive
HACE1	610876	6		616756	Spastic paraplegia and psychomotor retardation with or without seizures	Autosomal recessive
HADH	601609	4		231530	3-hydroxyacyl-CoA dehydrogenase deficiency	Autosomal recessive
HADHA	600890	2		609016; 609015	Long-chain 3-hydroxyl-CoA dehydrogenase (LCHAD) deficiency; Mitochondrial trifunctional protein deficiency	Autosomal recessive
HADHB	143450	2		609015	Mitochondrial trifunctional protein deficiency	Autosomal recessive
HAMP	606464	19		613313	Hemochromatosis, type 2B	Autosomal recessive
HARS1	142810	5	HARS	614504	Usher syndrome, type 3B	Autosomal recessive
HAX1	605998	1		610738	Neutropenia, severe congenital, type 3, autosomal recessive	Autosomal recessive
HBA1	141800	16		604131	Thalassemia, alpha-	Autosomal recessive
HBA2	141850	16		604131	Thalassemia, alpha-	Autosomal recessive
HBB	141900	11		603903	HBB-related hemoglobinopathy	Autosomal recessive
HCFC1	300019	X		309541	Mental retardation, X-linked 3 (methylmalonic acidemia and homocysteinemia, cblX type)	X-linked
HELLS	603946	10		616911	Immunodeficiency-centromeric instability-facial anomalies syndrome 4	Autosomal recessive
HEPACAM	611642	11		613925	Megalencephalic leukoencephalopathy with subcortical cysts 2A	Autosomal recessive

HERC1	605109	15		617011	Macrocephaly, dysmorphic facies, and psychomotor retardation	Autosomal recessive
HERC2	605837	15		615516	Mental retardation, autosomal recessive, type 38	Autosomal recessive
HES7	608059	17		613686	Spondylocostal dysostosis, type 4, autosomal recessive	Autosomal recessive
HESX1	601802	3		182230	Growth hormone deficiency with pituitary anomalies	Autosomal recessive
HEXA	606869	15		272800	Tay-Sachs disease	Autosomal recessive
HEXB	606873	5		268800	Sandhoff disease, infantile, juvenile, and adult forms	Autosomal recessive
HFM1	615684	1		615724	Premature ovarian failure 9	Autosomal recessive
HGD	607474	3		203500	Alkaptonuria	Autosomal recessive
HGF	142409	7	DFNB39	608265	Deafness, autosomal recessive, type 39	Autosomal recessive
HGSNAT	610453	8		252930	Mucopolysaccharidosis type 3C (Sanfilippo syndrome C)	Autosomal recessive
HIBCH	610690	2		250620	3-hydroxyisobutryl-CoA hydrolase deficiency	Autosomal recessive
HIKESHI	614908	11	C11orf73	616881	Leukodystrophy, hypomyelinating, type 13	Autosomal recessive
HINT1	601314	5		137200	Neuromyotonia and axonal neuropathy, autosomal recessive	Autosomal recessive
HJV	608374	1	HFE2	602390	Hemochromatosis, type 2A	Autosomal recessive
HK1	142600	10		605285	Charcot-Marie-Tooth disease, type 4G	Autosomal recessive
HLCS	609018	21		253270	Holocarboxylase synthetase deficiency	Autosomal recessive
HMGCL	613898	1		246450	HMG-CoA lyase deficiency	Autosomal recessive
HMGCS2	600234	1		605911	HMG-CoA synthase-2 deficiency	Autosomal recessive
HMOX1	141250	22		614034	Heme oxygenase-1 deficiency	Autosomal recessive
HMX1	142992	4		612109	Oculoauricular syndrome	Autosomal recessive
HNMT	605238	2		616739	Mental retardation, autosomal recessive, type 51	Autosomal recessive
HOGA1	613597	10		613616	Hyperoxaluria, primary, type 3	Autosomal recessive
HOXA1	142955	7		601536	Athabaskan brainstem dysgenesis syndrome	Autosomal recessive
HOXB1	142968	17		614744	Facial paresis, hereditary congenital, 3	Autosomal recessive
HOXC13	142976	12		614931	Ectodermal dysplasia 9, hair/nail type	Autosomal recessive
HPCA	142622	1		224500	Dystonia 2, torsion, autosomal recessive	Autosomal recessive
HPD	609695	12		276710	Tyrosinemia, type 3	Autosomal recessive
HPGD	601688	4		259100	Hypertrophic osteoarthropathy, primary, type 1 (pachydermoperiostosis)	Autosomal recessive
HPRT1	308000	X		300322	Lesch-Nyhan syndrome	X-linked
HPS1	604982	10		203300	Hermansky-Pudlak syndrome, type 1	Autosomal recessive
HPS3	606118	3		614072	Hermansky-Pudlak syndrome, type 3	Autosomal recessive
HPS4	606682	22		614073	Hermansky-Pudlak syndrome, type 4	Autosomal recessive
HPS5	607521	11		614074	Hermansky-Pudlak syndrome, type 5	Autosomal recessive
HPS6	607522	10		614075	Hermansky-Pudlak syndrome, type 6	Autosomal recessive
HPSE2	613469	10		236730	Urofacial syndrome, type 1	Autosomal recessive
HR	602302	8		203655; 209500	Alopecia universalis; Atrichia with papular lesions	Autosomal recessive
HSD11B2	614232	16		218030	Apparent mineralocorticoid excess	Autosomal recessive
HSD17B10	300256	X		300438	HSD10 mitochondrial disease	X-linked
HSD17B3	605573	9		264300	46,XY disorder of sex development due to 17-beta-hydroxysteroid dehydrogenase 3 deficiency	Autosomal recessive
HSD17B4	601860	5		261515	D-bifunctional protein deficiency	Autosomal recessive
HSD3B2	613890	1		201810	Adrenal hyperplasia, congenital, due to 3-beta-hydroxysteroid dehydrogenase 2 deficiency	Autosomal recessive
HSD3B7	607764	16		607765	Bile acid synthesis defect, congenital, type 1	Autosomal recessive
HSPA9	600548	5		616854	Even-plus syndrome	Autosomal recessive
HSPD1	118190	2		612233	Leukodystrophy, hypomyelinating, type 4	Autosomal recessive
HSPG2	142461	1		224410	Dyssegmental dysplasia, Silverman-Handmaker type	Autosomal recessive
HTRA1	602194	10		600142	CARASIL syndrome	Autosomal recessive
HTRA2	606441	2		617248	3-methylglutaconic aciduria, type 8	Autosomal recessive
HYAL1	607071	3		601492	?Mucopolysaccharidosis, type 9	Autosomal recessive
HYDIN	610812	16		608647	Ciliary dyskinesia, primary, type 5	Autosomal recessive
HYLS1	610693	11		236680	Hydrolethalus syndrome	Autosomal recessive
IARS1	600709	9	IARS	617093	Growth retardation, intellectual developmental disorder, hypotonia, and hepatopathy	Autosomal recessive
IBA57	615316	1		615330	Multiple mitochondrial dysfunctions syndrome 3	Autosomal recessive
ICOS	604558	2		607594	Immunodeficiency, common variable, 1	Autosomal recessive
IDH3B	604526	20		612572	Retinitis pigmentosa, type 46	Autosomal recessive
IDS	300823	X		309900	Mucopolysaccharidosis, type 2	X-linked

IDUA	252800	4	607014; 607015; 607016	Mucopolysaccharidosis type 1	Autosomal recessive
IER3IP1	609382	18	614231	Microcephaly, epilepsy, and diabetes syndrome	Autosomal recessive
IFNGR1	107470	6	209950	Immunodeficiency, type 27A, mycobacteriosis	Autosomal recessive
IFNGR2	147569	21	614889	Immunodeficiency, type 28, mycobacteriosis	Autosomal recessive
IFT122	606045	3	218330	Cranioectodermal dysplasia 1	Autosomal recessive
IFT140	614620	16	617781; 266920	Retinitis pigmentosa, type 80; Short-rib thoracic dysplasia 9 with or without polydactyly	Autosomal recessive
IFT172	607386	2	615630	Short-rib thoracic dysplasia 10 with or without polydactyly	Autosomal recessive
IFT43	614068	14	617866	Short-rib thoracic dysplasia 18 with polydactyly	Autosomal recessive
IFT52	617094	20	617102	Short-rib thoracic dysplasia 16 with or without polydactyly	Autosomal recessive
IFT80	611177	3	611263	Short-rib thoracic dysplasia, type 2, with or without polydactyly	Autosomal recessive
IFT81	605489	12	617895	Short-rib thoracic dysplasia 19 with or without polydactyly	Autosomal recessive
IGF1	147440	12	608747	Growth retardation with deafness and mental retardation due to IGF1 deficiency	Autosomal recessive
IGF1R	147370	15	270450*	Insulin-like growth factor I, resistance to	Autosomal recessive*
IGFALS	601489	16	615961	Acid-labile subunit deficiency	Autosomal recessive
IGFBP7	602867	4	614224	Retinal arterial macroaneurysm with supravalvular pulmonic stenosis	Autosomal recessive
IGHMBP2	600502	11	616155	Charcot-Marie-Tooth disease, axonal, type 2S	Autosomal recessive
IGLL1	146770	22	613500	Agammaglobulinemia 2	Autosomal recessive
IHH	600726	2	607778	Acrocapitofemoral dysplasia	Autosomal recessive
IKBKB	603258	8	615592	Immunodeficiency, type 15	Autosomal recessive
IL10RA	146933	11	613148	Inflammatory bowel disease, type 28, early onset, autosomal recessive	Autosomal recessive
IL10RB	123889	21	612567	Inflammatory bowel disease, type 25, early onset, autosomal recessive	Autosomal recessive
IL11RA	600939	9	614188	Craniosynostosis and dental anomalies	Autosomal recessive
IL12B	161561	5	614890	Immunodeficiency, type 29, mycobacteriosis	Autosomal recessive
IL12RB1	601604	19	614891	Immunodeficiency, type 30	Autosomal recessive
IL17RA	605461	22	613953	Immunodeficiency, type 51	Autosomal recessive
IL17RC	610925	3	616445	Candidiasis, familial, 9	Autosomal recessive
IL1RAPL1	300206	X	300143	Mental retardation, X-linked, type 21/34	X-linked
IL1RN	147679	2	612852	Sterile multifocal osteomyelitis with periostitis and pustulosis	Autosomal recessive
IL21R	605383	16	615207	Immunodeficiency, type 56	Autosomal recessive
IL2RA	147730	10	606367	Immunodeficiency, type 41, with lymphoproliferation and autoimmunity	Autosomal recessive
IL2RG	308380	X	300400	Severe combined immunodeficiency, X-linked	X-linked
IL36RN	605507	2	614204	Psoriasis, type 14, pustular	Autosomal recessive
IL7R	146661	5	608971	Severe combined immunodeficiency, T-cell negative, B-cell/natural killer cell-positive type	Autosomal recessive
ILDR1	609739	3	609646	Deafness, autosomal recessive, type 42	Autosomal recessive
IMPA1	602064	8	617323	Mental retardation, autosomal recessive 59	Autosomal recessive
IMPG2	607056	3	613581	Retinitis pigmentosa, type 56	Autosomal recessive
INPP5E	613037	9	213300	Joubert syndrome, type 1	Autosomal recessive
INPP5K	607875	17	617404	Muscular dystrophy, congenital, with cataracts and intellectual disability	Autosomal recessive
INPPL1	600829	11	258480	Opsismodysplasia	Autosomal recessive
INS	176730	11	606176*	Permanent neonatal diabetes mellitus (PNDM)	Autosomal recessive*
INSR	147670	19	610549	Diabetes mellitus, insulin-resistant, with acanthosis nigricans, type A	Autosomal recessive
INTS1	611345	7	618571	Neurodevelopmental disorder with cataracts, poor growth, and dysmorphic facies	Autosomal recessive
INVS	243305	9	602088	Nephronophthisis, type 2, infantile	Autosomal recessive
IQCB1	609237	3	609254	Senior-Loken syndrome, type 5	Autosomal recessive
IQCE	617631	7	617642	Polydactyly, postaxial, type A7	Autosomal recessive
IRAK4	606883	12	607676	Immunodeficiency, type 67 (IRAK4 deficiency)	Autosomal recessive
IRF8	601565	16	614894	Immunodeficiency, type 32B, monocyte and dendritic cell deficiency	Autosomal recessive
IRX5	606195	16	611174	Hamamy syndrome	Autosomal recessive
ISCA1	611006	9	617613	Multiple mitochondrial dysfunctions syndrome 5	Autosomal recessive
ISCA2	615317	14	616370	Multiple mitochondrial dysfunctions syndrome 4	Autosomal recessive
ISCU	611911	12	255125	Myopathy with lactic acidosis, hereditary	Autosomal recessive
ISG15	147571	1	616126	Immunodeficiency, type 38	Autosomal recessive
ITCH	606409	20	613385	Autoimmune disease, multisystem, with facial dysmorphism	Autosomal recessive
ITGA2B	607759	17	273800	Glanzmann thrombasthenia	Autosomal recessive
ITGA3	605025	17	614748	Interstitial lung disease, nephrotic syndrome, and epidermolysis bullosa, congenital	Autosomal recessive

ITGA6	147556	2		226730	Epidermolysis bullosa, junctional, with pyloric stenosis	Autosomal recessive
ITGA7	600536	12		613204	Muscular dystrophy, congenital, due to ITGA7 deficiency	Autosomal recessive
ITGA8	604063	10		191830	Renal hypodysplasia/aplasia 1	Autosomal recessive
ITGB2	600065	21		116920	Leukocyte adhesion deficiency	Autosomal recessive
ITGB3	173470	17		273800	Glanzmann thrombasthenia	Autosomal recessive
ITGB4	147557	17		226730	Epidermolysis bullosa, junctional, with pyloric atresia	Autosomal recessive
ITGB6	147558	2		616221	Amelogenesis imperfecta, type 1H	Autosomal recessive
ITK	186973	5		613011	Lymphoproliferative syndrome 1	Autosomal recessive
ITPA	147520	20		616647	Epileptic encephalopathy, early infantile, type 35	Autosomal recessive
ITPR1	147265	3		206700*	Gillespie syndrome	Autosomal recessive*
IVD	607036	15		243500	Isovaleric acidemia	Autosomal recessive
IYD	612025	6		274800	Thyroid dyshormonogenesis, type 4	Autosomal recessive
JAGN1	616012	3		616022	Neutropenia, severe congenital, 6, autosomal recessive	Autosomal recessive
JAK3	600173	19		600802	Severe Combined Immunodeficiency, autosomal recessive, T-negative/B-positive type	Autosomal recessive
JAM3	606871	11		613730	Hemorrhagic destruction of the brain, subependymal calcification, and cataracts	Autosomal recessive
JUP	173325	17		601214	Naxos disease	Autosomal recessive
KANK2	614610	19		617783	Nephrotic syndrome, type 16	Autosomal recessive
KARS1	601421	16	KARS	613916	Deafness, autosomal recessive, type 89	Autosomal recessive
KATNB1	602703	16		616212	Lissencephaly 6, with microcephaly	Autosomal recessive
KATNIP	616650	16	KIAA0556	616784	Joubert syndrome 26	Autosomal recessive
KCNE1	176261	21		612347	Jervell and Lange-Nielsen syndrome 2	Autosomal recessive
KCNJ1	600359	11		241200	Bartter syndrome, type 2	Autosomal recessive
KCNJ10	602208	1		612780	SESAME syndrome	Autosomal recessive
KCNJ11	600937	11		601820; 606176*	Hyperinsulinemic hypoglycemia, type 2 (congenital hyperinsulinism); Permanent neonatal diabetes mellitus (PNDM)	Autosomal recessive; Autosomal recessive*
KCNJ13	603208	2		614186	Leber congenital amaurosis, type 16	Autosomal recessive
KCNV2	607604	9		610356	Retinal cone dystrophy, type 3B	Autosomal recessive
KCTD7	611725	7		611726	Epilepsy, progressive myoclonic, type 3, with or without intracellular inclusions	Autosomal recessive
KDM5C	314690	X		300534	Mental retardation, X-linked, syndromic, Claes-Jensen type	X-linked
KDSR	136440	18		617526	Erythrokeratodermia variabilis et progressiva 4	Autosomal recessive
KERA	603288	12		217300	Cornea plana 2, autosomal recessive	Autosomal recessive
KHDC3L	611687	6		614293	Hydatidiform mole, recurrent, type 2	Autosomal recessive
KIAA0586	610178	14		616546	Short-rib thoracic dysplasia 14 with polydactyly	Autosomal recessive
KIAA0753	617112	17		617127	?Orofaciodigital syndrome, type 15	Autosomal recessive
KIAA1109	611565	4		617822	Alkuraya-Kucinskas syndrome	Autosomal recessive
KIAA1549	613344	7		618613	Retinitis pigmentosa, type 86	Autosomal recessive
KIF14	611279	1		617914; 616258	Microcephaly 20, primary, autosomal recessive; ?Meckel syndrome 12	Autosomal recessive
KIF1A	601255	2		614213; 610357	Neuropathy, hereditary sensory, type 2C; Spastic paraplegia, type 30, autosomal recessive	Autosomal recessive
KIF1C	603060	17		611302	Spastic ataxia 2, autosomal recessive	Autosomal recessive
KIF7	611254	15		200990	Acrocallosal syndrome; Joubert syndrome, type 12	Autosomal recessive
KIFBP	609367	10	KIF1BP; KIAA1279	609460	Goldberg-Shprintzen megacolon syndrome	Autosomal recessive
KISS1R	604161	19		614837	Hypogonadotropic hypogonadism, type 8, with or without anosmia	Autosomal recessive
KIZ	615757	20		615780	Retinitis pigmentosa 69	Autosomal recessive
KLHL3	605775	5		614495	Pseudohypoaldosteronism, type 2D	Autosomal recessive
KLHL40	615340	3		615348	Nemaline myopathy 8, autosomal recessive	Autosomal recessive
KLHL41	607701	2		615731	Nemaline myopathy 9	Autosomal recessive
KLHL7	611119	7		617055	Cold-induced sweating syndrome 3	Autosomal recessive
KLK4	603767	19		204700	Amelogenesis imperfecta, type 2A1 (hypomaturation type)	Autosomal recessive
KLKB1	229000	4		612423	Fletcher factor (prekallikrein) deficiency	Autosomal recessive
KNL1	609173	15	CASC5	604321	Microcephaly 4, primary, autosomal recessive	Autosomal recessive
KPTN	615620	19		615637	Mental retardation, autosomal recessive 41	Autosomal recessive
KREMEN1	609898	22		617392	Ectodermal dysplasia 13, hair/tooth type	Autosomal recessive
KRT10	148080	17		113800*	Epidermolytic hyperkeratosis	Autosomal recessive*
KRT14	148066	17		601001	Epidermolysis bullosa simplex, autosomal recessive, type 1	Autosomal recessive
KRT25	616646	17		616760	Woolly hair, autosomal recessive 3	Autosomal recessive

KRT5	148040	12		601001	Epidermolysis bullosa simplex, autosomal recessive, type 1	Autosomal recessive
KRT85	602767	12		602032	Ectodermal dysplasia 4, hair/nail type	Autosomal recessive
KY	605739	3		617114	Myopathy, myofibrillar, type 7	Autosomal recessive
KYNU	605197	2		617661	Vertebral, cardiac, renal, and limb defects syndrome, type 2	Autosomal recessive
L1CAM	308840	X		307000; 303350; 304100	L1 Syndrome	X-linked
L2HGDH	609584	14		236792	L-2-hydroxyglutaric aciduria	Autosomal recessive
LAMA1	150320	18		615960	Poretti-Boltshauser syndrome	Autosomal recessive
LAMA2	156225	6		607855; 618138	LAMA2-related muscular dystrophy	Autosomal recessive
LAMA3	600805	18		226700; 226650	Junctional epidermolysis bullosa (JEB) Herlitz type; JEB non-Herlitz type	Autosomal recessive
LAMB1	150240	7		615191	Lissencephaly, type 5	Autosomal recessive
LAMB2	150325	3		609049; 614199	Pierson syndrome; Nephrotic syndrome, type 5, with or without ocular abnormalities	Autosomal recessive
LAMB3	150310	1		226700; 226650	Junctional epidermolysis bullosa (JEB) Herlitz type; JEB non-Herlitz type	Autosomal recessive
LAMC2	150292	1		226700; 226650	Junctional epidermolysis bullosa (JEB) Herlitz type; JEB non-Herlitz type	Autosomal recessive
LAMC3	604349	9		614115	Cortical malformations, occipital	Autosomal recessive
LARGE1	603590	22	LARGE	613154; 608840	Muscular dystrophy-dystroglycanopathy, type 6A and 6B	Autosomal recessive
LARP7	612026	4		615071	Alazami syndrome	Autosomal recessive
LARS1	151350	5	LARS	615438	?Infantile liver failure syndrome 1 (ILFS1)	Autosomal recessive
LARS2	604544	3		615300	Perrault syndrome, type 4	Autosomal recessive
LAT	602354	16		617514	Immunodeficiency, type 52	Autosomal recessive
LBR	600024	1		215140	Greenberg skeletal dysplasia	Autosomal recessive
LCA5	611408	6		604537	Leber congenital amaurosis, type 5	Autosomal recessive
LCAT	606967	16		245900; 136120	Familial LCAT deficiency; Fish-eye disease	Autosomal recessive
LCK	153390	1		615758	?Immunodeficiency, type 22	Autosomal recessive
LCT	603202	2		223000	Lactase deficiency, congenital	Autosomal recessive
LDHA	150000	11		612933	Glycogen storage disease type 11	Autosomal recessive
LDLRAP1	605747	1		603813	Hypercholesterolemia, familial, autosomal recessive	Autosomal recessive
LEMD2	616312	6		212500	Cataract 46, juvenile-onset	Autosomal recessive
LEP	164160	7		614962	Obesity, morbid, due to leptin deficiency	Autosomal recessive
LEPR	601007	1		614963	Obesity, morbid, due to leptin receptor deficiency	Autosomal recessive
LGI4	608303	19		617468	Arthrogryposis multiplex congenita, neurogenic, with myelin defect	Autosomal recessive
LHB	152780	19		228300	Hypogonadotropic hypogonadism, type 23, with or without anosmia	Autosomal recessive
LHCGR	152790	2		238320	Leydig cell hypoplasia	Autosomal recessive
LHFPL5	609427	6		610265	Deafness, autosomal recessive, type 67	Autosomal recessive
LHX3	600577	9		221750	Pituitary hormone deficiency, combined, type 3	Autosomal recessive
LIAS	607031	4		614462	Hyperglycinemia, lactic acidosis, and seizures	Autosomal recessive
LIFR	151443	5		601559	Stuve-Wiedemann syndrome / Schwartz-Jampel type 2 syndrome	Autosomal recessive
LIG4	601837	13		606593	LIG4 syndrome	Autosomal recessive
LIM2	154045	19		615277	Cataract 19, multiple types	Autosomal recessive
LINS1	610350	15	LINS	614340	Mental retardation, autosomal recessive, type 27	Autosomal recessive
LIPA	613497	10		278000	Lysosomal acid lipase deficiency	Autosomal recessive
LIPE	151750	19		615980	Lipodystrophy, familial partial, type 6	Autosomal recessive
LIPH	607365	3		604379	Hypotrichosis, type 7 or woolly hair, autosomal recessive, type 2, with or without hypotrichosis	Autosomal recessive
LIPN	613924	10		613943	Ichthyosis, congenital, autosomal recessive 8	Autosomal recessive
LIPT1	610284	2		616299	Lipoyltransferase 1 deficiency	Autosomal recessive
LIPT2	617659	11		617668	Encephalopathy, neonatal severe, with lactic acidosis and brain abnormalities	Autosomal recessive
LMAN1	601567	18		227300	Combined deficiency of factor V and factor VIII, type 1	Autosomal recessive
LMBRD1	612625	6		277380	Methylmalonic aciduria and homocystinuria, cblF type	Autosomal recessive
LMF1	611761	16		246650	Lipase deficiency, combined	Autosomal recessive
LMNA	150330	1		605588; 616516	LMNA-related disorders, autosomal recessive	Autosomal recessive
LMOD3	616112	3		616165	Nemaline myopathy 10	Autosomal recessive
LONP1	605490	19		600373	CODAS syndrome	Autosomal recessive
LOXHD1	613072	18		613079	Deafness, autosomal recessive, type 77	Autosomal recessive
LPAR6	609239	13		278150	Hypotrichosis, type 8 or woolly hair, autosomal recessive, type 1, with or without hypotrichosis	Autosomal recessive
LPIN1	605518	2		268200	Myoglobinuria, acute recurrent, autosomal recessive	Autosomal recessive
LPIN2	605519	18		609628	Majeed syndrome	Autosomal recessive

LPL	609708	8		238600	Lipoprotein lipase deficiency	Autosomal recessive
LRAT	604863	4		613341	Leber congenital amaurosis type 14	Autosomal recessive
LRBA	606453	4		614700	Immunodeficiency, common variable, 8, with autoimmunity	Autosomal recessive
LRIG2	608869	1		615112	Urofacial syndrome 2	Autosomal recessive
LRIT3	615004	4		615058	Night blindness, congenital stationary (complete), 1F, autosomal recessive	Autosomal recessive
LRMDA	614537	10	C10orf11	615179	Albinism, oculocutaneous, type 7	Autosomal recessive
LRP2	600073	2		222448	Donnai-Barrow syndrome	Autosomal recessive
LRP4	604270	11		212780	Cenani-Lenz syndactyly syndrome	Autosomal recessive
LRP5	603506	11		259770	Osteoporosis-pseudoglioma syndrome	Autosomal recessive
LRPAP1	104225	4		615431	Myopia, type 23, autosomal recessive	Autosomal recessive
LRPPRC	607544	2		220111	Leigh syndrome, French-Canadian type	Autosomal recessive
LRRC6	614930	8		614935	Ciliary dyskinesia, primary, type 19	Autosomal recessive
LRSAM1	610933	9		614436	Charcot-Marie-Tooth disease, axonal, type 2P	Autosomal recessive
LRTOMT	612414	11		611451	Deafness, autosomal recessive, type 63	Autosomal recessive
LSS	600909	21		616509	Cataract 44	Autosomal recessive
LTBP2	602091	14		251750	Microspherophakia and/or megalocornea, with ectopia lentis and with or without secondary glaucoma	Autosomal recessive
LTBP3	602090	11		601216	Dental anomalies and short stature	Autosomal recessive
LTBP4	604710	19		613177	Cutis laxa, autosomal recessive, type 1C	Autosomal recessive
LYRM7	615831	5		615838	Mitochondrial complex III deficiency, nuclear type 8	Autosomal recessive
LYST	606897	1		214500	Chediak-Higashi syndrome	Autosomal recessive
LZTFL1	606568	3		615994	Bardet-Biedl syndrome, type 17	Autosomal recessive
LZTR1	600574	22		605275	Noonan syndrome, type 2	Autosomal recessive
MAG	159460	19		616680	Spastic paraplegia, type 75, autosomal recessive	Autosomal recessive
MAGI2	606382	7		617609	Nephrotic syndrome, type 15	Autosomal recessive
MAK	154235	6		614181	Retinitis pigmentosa type 62	Autosomal recessive
MALT1	604860	18		615468	Immunodeficiency, type 12	Autosomal recessive
MAN1B1	604346	9		614202	Mental retardation, autosomal recessive, type 15	Autosomal recessive
MAN2B1	609458	19		248500	Alpha-mannosidosis	Autosomal recessive
MANBA	609489	4		248510	Mannosidosis, beta	Autosomal recessive
MAP3K20	609479	2	ZAK	617760	Centronuclear myopathy, type 6, with fiber-type disproportion	Autosomal recessive
MAPKBP1	616786	15		617271	Nephronophthisis 20	Autosomal recessive
MAPT	157140	17		260540	Supranuclear palsy, progressive atypical (parkinsonism syndrome)	Autosomal recessive
MARS1	156560	12	MARS	615486	Interstitial lung and liver disease	Autosomal recessive
MARS2	609728	2		611390	Spastic ataxia, type 3, autosomal recessive	Autosomal recessive
MARVELD2	610572	5		610153	Deafness, autosomal recessive, type 49	Autosomal recessive
MASP1	600521	3		257920	3MC syndrome 1	Autosomal recessive
MAT1A	610550	10		250850	Methionine adenosyltransferase deficiency, autosomal recessive	Autosomal recessive
MATN3	602109	2		608728	?Spondyloepimetaphyseal dysplasia	Autosomal recessive
MBOAT7	606048	19		617188	Mental retardation, autosomal recessive 57	Autosomal recessive
MC2R	607397	18		202200	Glucocorticoid deficiency, due to ACTH unresponsiveness	Autosomal recessive
MCCC1	609010	3		210200	3-Methylcrotonyl-CoA carboxylase deficiency, type 1	Autosomal recessive
MCCC2	609014	5		210210	3-Methylcrotonyl-CoA carboxylase deficiency, type 2	Autosomal recessive
MCEE	608419	2		251120	Methylmalonyl-CoA epimerase deficiency	Autosomal recessive
MCFD2	607788	2		613625	Combined deficiency of factor V and factor VIII, type 2	Autosomal recessive
MCIDAS	614086	5		618695	Ciliary dyskinesia, primary, type 42	Autosomal recessive
MCM3AP	603294	21		618124	Peripheral neuropathy, autosomal recessive, with or without impaired intellectual development	Autosomal recessive
MCM4	602638	8		609981	Immunodeficiency, type 54	Autosomal recessive
MCM9	610098	6		616185	Ovarian dysgenesis 4	Autosomal recessive
MCOLN1	605248	19		252650	Mucolipidosis type 4	Autosomal recessive
MCPH1	607117	8		251200	Microcephaly type 1, primary, autosomal recessive	Autosomal recessive
MDH2	154100	7		617339	Epileptic encephalopathy, early infantile, 51	Autosomal recessive
MECP2	300005	X		300673;312750	Encephalopathy, neonatal severe; Rett syndrome	X-linked
MECR	608205	1		617282	Dystonia, childhood-onset, with optic atrophy and basal ganglia abnormalities	Autosomal recessive
MED17	603810	11		613668	Microcephaly, postnatal progressive, with seizures and brain atrophy	Autosomal recessive
MED23	605042	6		614249	Mental retardation, autosomal recessive, type 18	Autosomal recessive

MED25	610197	19		616449	Basel-Vanagait-Smirin-Yosef syndrome	Autosomal recessive
MEFV	608107	16		249100	Familial Mediterranean fever	Autosomal recessive
MEGF10	612453	5		614399	Myopathy, areflexia, respiratory distress, and dysphagia, early-onset	Autosomal recessive
MEGF8	604267	19		614976	Carpenter syndrome, type 2	Autosomal recessive
MEOX1	600147	17		214300	Klippel-Feil syndrome 2	Autosomal recessive
MERTK	604705	2		613862	Retinitis pigmentosa type 38	Autosomal recessive
MESP2	605195	15		608681	Spondylocostal dysostosis, type 2, autosomal recessive	Autosomal recessive
METTL23	615262	17		615942	Mental retardation, autosomal recessive 44	Autosomal recessive
MFF	614785	2		617086	Encephalopathy due to defective mitochondrial and peroxisomal fission, type 2	Autosomal recessive
MFN2	608507	1		617087	Charcot-Marie-Tooth disease, axonal, type 2A2B	Autosomal recessive
MFRP	606227	11		611040	Microphthalmia, isolated type 5	Autosomal recessive
MFSD2A	614397	1		616486	Microcephaly 15, primary, autosomal recessive	Autosomal recessive
MFSD8	611124	4		610951	Ceroid lipofuscinosis, neuronal, type 7	Autosomal recessive
MGAT2	602616	14		212066	Congenital disorder of glycosylation, type 2a	Autosomal recessive
MGME1	615076	20		615084	Mitochondrial DNA depletion syndrome 11	Autosomal recessive
MGP	154870	12		245150	Keutel syndrome	Autosomal recessive
MICU1	605084	10		615673	Myopathy with extrapyramidal signs	Autosomal recessive
MIPEP	602241	13		617228	Combined oxidative phosphorylation deficiency 31	Autosomal recessive
MITF	156845	3		617306	COMMAD syndrome	Autosomal recessive
MKKS	604896	20		605231	Bardet-Biedl syndrome type 6	Autosomal recessive
MKS1	609883	17		615990; 249000; 617121	Bardet-Biedl syndrome type 13; Meckel syndrome, type 1; Joubert syndrome, type 28	Autosomal recessive
MLC1	605908	22		604004	Megalencephalic leukoencephalopathy with subcortical cysts	Autosomal recessive
MLPH	606526	2		609227	Griscelli syndrome, type 3	Autosomal recessive
MLYCD	606761	16		248360	Malonyl-CoA decarboxylase deficiency	Autosomal recessive
MMAA	607481	4		251100	Methylmalonic aciduria, vitamin B12-responsive	Autosomal recessive
MMAB	607568	12		251110	Methylmalonic aciduria, vitamin B12-responsive, type cblB	Autosomal recessive
MMACHC	609831	1		277400	Methylmalonic aciduria and homocystinuria, cblC type	Autosomal recessive, digenic inheritance (PRDX1 gene)
MMADHC	611935	2		277410	Homocystinuria, cblD type, variant 1	Autosomal recessive
MME	120520	3		617017*	Charcot-Marie-Tooth disease, axonal, type 2T	Autosomal recessive*
MMP13	600108	11		250400	Metaphyseal dysplasia, Spahr type	Autosomal recessive
MMP2	120360	16		259600	Multicentric osteolysis, nodulosis, and arthropathy (MONA)	Autosomal recessive
MMP20	604629	11		612529	Amelogenesis imperfecta, type 2A2 (hypomaturation type)	Autosomal recessive
MMP21	608416	10		616749	Heterotaxy, visceral, 7, autosomal	Autosomal recessive
MMUT	609058	6	MUT	251000	Methylmalonic aciduria, mut(0) type	Autosomal recessive
MOCOS	613274	18		603592	Xanthinuria, type 2	Autosomal recessive
MOCS1	603707	6		252150	Molybdenum cofactor deficiency A	Autosomal recessive
MOCS2	603708	5		252160	Molybdenum cofactor deficiency B	Autosomal recessive
MOGS	601336	2		606056	Congenital disorder of glycosylation, type 2B	Autosomal recessive
MPC1	614738	6		614741	Mitochondrial pyruvate carrier deficiency	Autosomal recessive
MPDU1	604041	17		609180	Congenital disorder of glycosylation, type 1F	Autosomal recessive
MPDZ	603785	9		615219	Hydrocephalus, congenital, type 2, with or without brain or eye anomalies	Autosomal recessive
MPI	154550	15		602579	Congenital disorder of glycosylation, type 1B	Autosomal recessive
MPIG6B	606520	6	C6orf25	617441	Thrombocytopenia, anemia, and myelofibrosis	Autosomal recessive
MPL	159530	1		604498	Thrombocytopenia, congenital amegakaryocytic	Autosomal recessive
MPLKIP	609188	7		234050	Trichothiodystrophy, type 4, nonphotosensitive	Autosomal recessive
MPO	606989	17		254600	Myeloperoxidase deficiency	Autosomal recessive
MPV17	137960	2		256810; 618400	Mitochondrial DNA depletion syndrome type 6 (hepatocerebral); Charcot-Marie-Tooth disease, axonal, type 2EE	Autosomal recessive
MPZ	159440	1		145900*	Dejerine-Sottas disease	Autosomal recessive*
MRAP	609196	21		607398	Glucocorticoid deficiency, type 2	Autosomal recessive
MRE11	600814	11	MRE11A	604391	Ataxia-telangiectasia-like disorder 1	Autosomal recessive
MRPS16	609204	10		610498	Combined oxidative phosphorylation deficiency 2	Autosomal recessive
MRPS22	605810	3		611719	Combined oxidative phosphorylation deficiency type 5	Autosomal recessive
MRPS34	611994	16		617664	Combined oxidative phosphorylation deficiency 32	Autosomal recessive
MSH3	600887	5		617100	Familial adenomatous polyposis, type 4	Autosomal recessive

MSMO1	607545	4		616834	Microcephaly, congenital cataract, and psoriasiform dermatitis	Autosomal recessive
MSRB3	613719	12		613718	Deafness, autosomal recessive, type 74	Autosomal recessive
MSTO1	617619	1		617675*	Myopathy, mitochondrial, and ataxia	Autosomal recessive*
MTFMT	611766	15		614947	Combined oxidative phosphorylation deficiency 15	Autosomal recessive
MTHFD1	172460	14		617780	Combined immunodeficiency and megaloblastic anemia with or without hyperhomocysteinemia	Autosomal recessive
MTHFR	607093	1		236250	Homocystinuria due to MTHFR deficiency	Autosomal recessive
MTM1	300415	X		310400	Myotubular myopathy, X-linked	X-linked
MTMR2	603557	11		601382	Charcot-Marie-Tooth disease, type 4B1	Autosomal recessive
MTO1	614667	6		614702	Combined oxidative phosphorylation deficiency 10	Autosomal recessive
MTR	156570	1		250940	Homocystinuria-megaloblastic anemia, cblG complementation type	Autosomal recessive
MTRR	602568	5		236270	Homocystinuria-megaloblastic anemia, cbl E type	Autosomal recessive
MTTP	157147	4		200100	Abetalipoproteinemia	Autosomal recessive
MUSK	601296	9		208150; 616325	Fetal akinesia deformation sequence, type 1; Myasthenic syndrome, congenital, type 9, associated with acetylcholine receptor deficiency	Autosomal recessive
MUTYH	604933	1		608456	Adenomas, multiple colorectal	Autosomal recessive
MVK	251170	12		610377	Mevalonic aciduria	Autosomal recessive
MYBPC1	160794	12		614915	Lethal congenital contracture syndrome, type 4	Autosomal recessive
MYD88	602170	3		612260	Immunodeficiency, type 68	Autosomal recessive
MYH2	160740	17		605637	Proximal myopathy and ophthalmoplegia	Autosomal recessive
MYMK	615345	9	TMEM8C	254940	Carey-Fineman-Ziter syndrome	Autosomal recessive
MYO15A	602666	17		600316	Deafness, autosomal recessive, type 3	Autosomal recessive
MYO18B	607295	22		616549	Klippel-Feil syndrome, type 4, autosomal recessive, with myopathy and facial dysmorphism	Autosomal recessive
MYO1E	601479	15		614131	Glomerulosclerosis, focal segmental, 6	Autosomal recessive
MYO3A	606808	10		607101	Deafness, autosomal recessive, type 30	Autosomal recessive
MYO5A	160777	15		214450	Griscelli syndrome, type 1	Autosomal recessive
MYO5B	606540	18		251850	Microvillus inclusion disease	Autosomal recessive
MYO6	600970	6		607821	Deafness, autosomal recessive, type 37	Autosomal recessive
MYO7A	276903	11		276900; 600060	Usher syndrome, type 1B; Deafness, autosomal recessive, type 2	Autosomal recessive
MYPN	608517	10		617336	Nemaline myopathy, type 11, autosomal recessive	Autosomal recessive
NADK2	615787	5		616034	2,4-dienoyl-CoA reductase deficiency	Autosomal recessive
NAGA	104170	22		609241	Schindler disease, type I	Autosomal recessive
NAGLU	609701	17		252920	Mucopolysaccharidosis, type 3B (Sanfilippo B)	Autosomal recessive
NAGS	608300	17		237310	N-acetylglutamate synthase deficiency	Autosomal recessive
NALCN	611549	13		615419	Hypotonia, infantile, with psychomotor retardation and characteristic facies 1	Autosomal recessive
NANS	605202	9		610442	Spondyloepimetaphyseal dysplasia, Camera-Genevieve type	Autosomal recessive
NARS2	612803	11		616239	Combined oxidative phosphorylation deficiency 24	Autosomal recessive
NAXE	608862	1	APOA1BP	617186	Encephalopathy, progressive, early-onset, with brain edema and/or leukoencephalopathy	Autosomal recessive
NBAS	608025	2		616483; 614800	Infantile liver failure syndrome, type 2; Short stature, optic nerve atrophy, and Pelger-Huet anomaly	Autosomal recessive
NBEAL2	614169	3		139090	Gray platelet syndrome	Autosomal recessive
NBN	602667	8		251260	Nijmegen breakage syndrome	Autosomal recessive
NCAPD3	609276	11		617984	Microcephaly 22, primary, autosomal recessive	Autosomal recessive
NCF1	608512	7		233700	Chronic granulomatous disease, type 1	Autosomal recessive
NCF2	608515	1		233710	Chronic granulomatous disease, type 2	Autosomal recessive
NCF4	601488	22		613960	Chronic granulomatous disease, type 3	Autosomal recessive
NDE1	609449	16		614019	Lissencephaly, type 4 (with microcephaly)	Autosomal recessive
NDP	300658	X		310600	Norrie disease	X-linked
NDRG1	605262	8		601455	Charcot-Marie-Tooth disease, type 4D	Autosomal recessive
NDST1	600853	5		616116	Mental retardation, autosomal recessive, type 46	Autosomal recessive
NDUFA10	603835	2		618243	Mitochondrial complex I deficiency, nuclear type 22	Autosomal recessive
NDUFA11	612638	19		618236	Mitochondrial complex I deficiency, nuclear type 14	Autosomal recessive
NDUFA12	614530	12		618244	?Mitochondrial complex I deficiency, nuclear type 23	Autosomal recessive
NDUFA2	602137	5		618235	Mitochondrial complex I deficiency, nuclear type 13	Autosomal recessive
NDUFA9	603834	12		618247	Mitochondrial complex I deficiency, nuclear type 26	Autosomal recessive
NDUFAF1	606934	15		618234	Mitochondrial complex I deficiency, nuclear type 11	Autosomal recessive
NDUFAF2	609653	5		618233	Mitochondrial complex I deficiency, nuclear type 10	Autosomal recessive

NDUFAF3	612911	3		618240	Mitochondrial complex I deficiency, nuclear type 18	Autosomal recessive
NDUFAF5	612360	20		618238	Mitochondrial complex I deficiency, nuclear type 16	Autosomal recessive
NDUFAF6	612392	8		618239	Mitochondrial complex I deficiency, nuclear type 17	Autosomal recessive
NDUFB3	603839	2		618246	Mitochondrial complex I deficiency, nuclear type 25	Autosomal recessive
NDUFB9	601445	8		618245	Mitochondrial complex I deficiency, nuclear type 24	Autosomal recessive
NDUFS1	157655	2		618226	Mitochondrial complex I deficiency, nuclear type 5	Autosomal recessive
NDUFS2	602985	1		618228	Mitochondrial complex I deficiency, nuclear type 6	Autosomal recessive
NDUFS3	603846	11		618230	Mitochondrial complex I deficiency, nuclear type 8	Autosomal recessive
NDUFS4	602694	5		252010	Mitochondrial complex I deficiency, nuclear type 1	Autosomal recessive
NDUFS6	603848	5		618232	Mitochondrial complex I deficiency, nuclear type 9	Autosomal recessive
NDUFS7	601825	19		618224	Mitochondrial complex I deficiency, nuclear type 3	Autosomal recessive
NDUFS8	602141	11		618222	Mitochondrial complex I deficiency, nuclear type 2	Autosomal recessive
NDUFV1	161015	11		618225	Mitochondrial complex I deficiency, nuclear type 4	Autosomal recessive
NDUFV2	600532	18		618229	Mitochondrial complex I deficiency, nuclear type 7	Autosomal recessive
NEB	161650	2		256030	Nemaline myopathy type 2	Autosomal recessive
NECTIN1	600644	11	PVRL1	225060	Cleft lip/palate-ectodermal dysplasia syndrome; Orofacial cleft 7	Autosomal recessive
NECTIN4	609607	1	PVRL4	613573	Ectodermal dysplasia-syndactyly syndrome, type 1	Autosomal recessive
NEFL	162280	8		607734	Charcot-Marie-Tooth disease, type 1F	Autosomal recessive
NEK1	604588	4		263520	Short-rib thoracic dysplasia, type 6, with or without polydactyly	Autosomal recessive
NEK8	609799	17		615415	Renal-hepatic-pancreatic dysplasia, type 2	Autosomal recessive
NEK9	609798	14		617022	Lethal congenital contracture syndrome 10	Autosomal recessive
NEU1	608272	6		256550	Sialidosis, type 1 and type 2	Autosomal recessive
NEUROG3	604882	10		610370	Diarrhea 4, malabsorptive, congenital	Autosomal recessive
NFU1	608100	2		605711	Multiple mitochondrial dysfunctions syndrome 1	Autosomal recessive
NGF	162030	1		608654	Neuropathy, hereditary sensory and autonomic, type 5	Autosomal recessive
NGLY1	610661	3		615273	Congenital disorder of deglycosylation	Autosomal recessive
NHEJ1	611290	2		611291	Severe combined immunodeficiency with microcephaly, growth retardation, and sensitivity to ionizing radiation	Autosomal recessive
NHLRC1	608072	6		254780	Epilepsy, progressive myoclonic, type 2B (Lafora)	Autosomal recessive
NHP2	606470	5		613987	Dyskeratosis congenita, autosomal recessive type 2	Autosomal recessive
NIN	608684	14		614851	Seckel syndrome, type 7	Autosomal recessive
NIPAL4	609383	5		612281	Ichthyosis, congenital, autosomal recessive, type 6	Autosomal recessive
NKX2-6	611770	8		217095	Conotruncal heart malformations	Autosomal recessive
NKX3-2	602183	4		613330	Spondylo-megaepiphyseal-metaphyseal dysplasia	Autosomal recessive
NKX6-2	605955	10		617560	Spastic ataxia 8, autosomal recessive, with hypomyelinating leukodystrophy	Autosomal recessive
NLRP1	606636	17		617388*	Autoinflammation with arthritis and dyskeratosis	Autosomal recessive*
NLRP7	609661	19		231090	Hydatidiform mole, recurrent, type 1	Autosomal recessive
NME8	607421	7		610852	Ciliary dyskinesia, primary, type 6	Autosomal recessive
NMNAT1	608700	1		608553	Leber congenital amaurosis type 9	Autosomal recessive
NNT	607878	5		614736	Glucocorticoid deficiency 4, with or without mineralocorticoid deficiency	Autosomal recessive
NOP10	606471	15		224230	Dyskeratosis congenita, autosomal recessive type 1	Autosomal recessive
NPC1	607623	18		257220	Niemann-Pick disease, type C1	Autosomal recessive
NPC2	601015	14		607625	Niemann-pick disease, type C2	Autosomal recessive
NPHP1	607100	2		609583	Joubert syndrome type 4	Autosomal recessive
NPHP3	608002	3		267010	Meckel syndrome type 7	Autosomal recessive
NPHP4	607215	1		606966	Nephronophthisis type 4	Autosomal recessive
NPHS1	602716	19		256300	Nephrotic syndrome, type 1	Autosomal recessive
NPHS2	604766	1		600995	Nephrotic syndrome, type 2	Autosomal recessive
NPR2	108961	9		602875	Acromesomelic dysplasia, Maroteaux type	Autosomal recessive
NR0B1	300473	X		300200	Adrenal hypoplasia, congenital	X-linked
NR1H4	603826	12		617049	Cholestasis, progressive familial intrahepatic, type 5	Autosomal recessive
NR2E3	604485	15		268100; 611131*	Enhanced S-cone syndrome (Goldmann-Favre); Retinitis pigmentosa, type 37	Autosomal recessive; Autosomal recessive*
NRL	162080	14		613750*	Retinal degeneration, autosomal recessive, clumped pigment type	Autosomal recessive*
NRXN1	600565	2		614325	Pitt-Hopkins-like syndrome, type 2	Autosomal recessive
NSMCE2	617246	8		617253	Seckel syndrome, type 10	Autosomal recessive

NSUN2	610916	5		611091	Mental retardation, autosomal recessive, type 5	Autosomal recessive
NT5C2	600417	10		613162	Spastic paraplegia, type 45, autosomal recessive	Autosomal recessive
NT5C3A	606224	7		266120	Anemia, hemolytic, due to UMPH1 deficiency	Autosomal recessive
NT5E	129190	6		211800	Calcification of joints and arteries	Autosomal recessive
NTHL1	602656	16		616415	Familial adenomatous polyposis, type 3	Autosomal recessive
NTRK1	191315	1		256800	Insensitivity to pain, congenital, with anhidrosis	Autosomal recessive
NUBPL	613621	14		618242	Mitochondrial complex I deficiency, nuclear type 21	Autosomal recessive
NUP107	607617	12		616730	Nephrotic syndrome, type 11	Autosomal recessive
NUP62	605815	19		271930	Striatonigral degeneration, infantile	Autosomal recessive
NUP93	614351	16		616892	Nephrotic syndrome, type 12	Autosomal recessive
OAT	613349	10		258870	Gyrate atrophy of choroid and retina	Autosomal recessive
OBSL1	610991	2		612921	3M syndrome 2	Autosomal recessive
OCA2	611409	15		203200	Oculocutaneous albinism type 2	Autosomal recessive
OCLN	602876	5		251290	Pseudo-TORCH syndrome, type 1	Autosomal recessive
OCRL	300535	X		309000; 300555	Lowe Syndrome; Dent disease type 2	X-linked
ODAD1	615038	19	CCDC114	615067	Ciliary dyskinesia, primary, type 20	Autosomal recessive
ODAD2	615408	10	ARMC4	615451	Ciliary dyskinesia, primary, type 23	Autosomal recessive
ODAD3	615956	19	CCDC151	616037	Ciliary dyskinesia, primary, type 30	Autosomal recessive
OPA1	605290	3		210000	Behr syndrome	Autosomal recessive
OPA3	606580	19		258501	3-methylglutaconic aciduria, type 3	Autosomal recessive
OPHN1	300127	X		300486	Mental retardation, X-linked, with cerebellar hypoplasia and distinctive facial appearance	X-linked
OPTN	602432	10		613435	Amyotrophic lateral sclerosis, type 12	Autosomal recessive
ORAI1	610277	12		612782	Immunodeficiency, type 9	Autosomal recessive
ORC1	601902	1		224690	Meier-Gorlin syndrome, type 1	Autosomal recessive
ORC4	603056	2		613800	Meier-Gorlin syndrome, type 2	Autosomal recessive
ORC6	607213	16		613803	Meier-Gorlin syndrome, type 3	Autosomal recessive
OSGEP	610107	14		617729	Galloway-Mowat syndrome 3	Autosomal recessive
OSTM1	607649	6		259720	Osteopetrosis, autosomal recessive type 5	Autosomal recessive
OTC	300461	X		311250	Ornithine transcarbamylase deficiency	X-linked
OTOA	607038	16		607039	Deafness, autosomal recessive, type 22	Autosomal recessive
OTOF	603681	2		601071	Deafness, autosomal recessive, type 9	Autosomal recessive
OTOG	604487	11		614945	Deafness, autosomal recessive, type 18B	Autosomal recessive
OTOGL	614925	12		614944	Deafness, autosomal recessive, type 84B	Autosomal recessive
OTUD6B	612021	8		617452	Intellectual developmental disorder with dysmorphic facies, seizures, and distal limb anomalies	Autosomal recessive
OTULIN	615712	5	FAM105B	617099	Autoinflammation, panniculitis, and dermatosis syndrome	Autosomal recessive
OXCT1	601424	5		245050	Succinyl CoA:3-oxoacid CoA transferase deficiency	Autosomal recessive
P2RY12	600515	3		609821	Bleeding disorder, platelet-type, type 8	Autosomal recessive
P3H1	610339	1	LEPRE1	610915	Osteogenesis imperfecta, type 8	Autosomal recessive
P3H2	610341	3	LEPREL1	614292	Myopia, high, with cataract and vitreoretinal degeneration	Autosomal recessive
PADI3	606755	1		191480	Uncombable hair syndrome	Autosomal recessive
PADI6	610363	1		617234	Preimplantation embryonic lethality 2	Autosomal recessive
PAH	612349	12		261600	Phenylketonuria	Autosomal recessive
PAK3	300142	X		300558	Mental retardation, X-linked, type 30	X-linked
PALB2	610355	16		610832	Fanconi anemia, complementation group N	Autosomal recessive
PAM16	614336	16		613320	Spondylometaphyseal dysplasia, Megarbane-Dagher-Melike type	Autosomal recessive
PANK2	606157	20		234200	Neurodegeneration with brain iron accumulation type 1	Autosomal recessive
PAPSS2	603005	10		612847	Brachyolmia, type 4, with mild epiphyseal and metaphyseal changes	Autosomal recessive
PARK7	602533	1		606324	Parkinson disease, type 7, autosomal recessive, early-onset	Autosomal recessive
PARN	604212	16		616353	Dyskeratosis congenita, autosomal recessive 6	Autosomal recessive
PATL2	614661	15		617743	Oocyte maturation defect 4	Autosomal recessive
PAX7	167410	1		268220	Rhabdomyosarcoma 2, alveolar	Autosomal recessive
PC	608786	11		266150	Pyruvate carboxylase deficiency	Autosomal recessive
PCARE	613425	2	C2orf71	613428	Retinitis pigmentosa, type 54	Autosomal recessive
PCBD1	126090	10		264070	Hyperphenylalaninemia, BH4-deficient, type D	Autosomal recessive
PCCA	232000	13		606054	Propionic acidemia	Autosomal recessive

PCCB	232050	3	606054	Propionic acidemia	Autosomal recessive
PCDH12	605622	5	251280	Microcephaly, seizures, spasticity, and brain calcification	Autosomal recessive
PCDH15	605514	10	609533; 601067	Deafness, autosomal recessive, type 23; Usher syndrome, type 1D/F digenic	Autosomal recessive
PCK2	614095	14	261650	PEPCK deficiency, mitochondrial	Autosomal recessive
PCNT	605925	21	210720	Microcephalic osteodysplastic primordial dwarfism, type 2	Autosomal recessive
PCSK1	162150	5	600955	Obesity with impaired prohormone processing	Autosomal recessive
PCYT1A	123695	3	608940	Spondylometaphyseal dysplasia with cone-rod dystrophy	Autosomal recessive
PDE10A	610652	6	616921	Dyskinesia, limb and orofacial, infantile-onset	Autosomal recessive
PDE6A	180071	5	613810	Retinitis pigmentosa type 43	Autosomal recessive
PDE6B	180072	4	613801	Retinitis pigmentosa type 40	Autosomal recessive
PDE6C	600827	10	613093	Cone dystrophy type 4	Autosomal recessive
PDE6G	180073	17	613582	Retinitis pigmentosa type 57	Autosomal recessive
PDE6H	601190	12	610024*	Retinal cone dystrophy 3 and achromatopsia 6	Autosomal recessive*
PDHA1	300502	X	312170	Pyruvate dehydrogenase E1-alpha deficiency	X-linked
PDHB	179060	3	614111	Pyruvate dehydrogenase E1-beta deficiency	Autosomal recessive
PDHX	608769	11	245349	Lacticacidemia due to PDX1 deficiency	Autosomal recessive
PDP1	605993	8	608782	Pyruvate dehydrogenase phosphatase deficiency	Autosomal recessive
PDSS1	607429	10	614651	Coenzyme Q10 deficiency, primary, type 2	Autosomal recessive
PDSS2	610564	6	614652	Coenzyme Q10 deficiency, primary, type 3	Autosomal recessive
PDX1	600733	13	260370	Pancreatic agenesis type 1	Autosomal recessive
PDXK	179020	21	618511	Neuropathy, hereditary motor and sensory, type VIC, with optic atrophy	Autosomal recessive
PDZD7	612971	10	618003; 605472	Deafness, autosomal recessive, type 57; Usher syndrome, type 2C, digenic	Autosomal recessive; Digenic inheritance (ADGRV1 gene)
PEPD	613230	19	170100	Prolidase deficiency	Autosomal recessive
PET100	614770	19	619055	Mitochondrial complex IV deficiency, nuclear type 12	Autosomal recessive
PEX1	602136	7	234580	Heimler syndrome type 1	Autosomal recessive
PEX10	602859	1	614870; 614871	Peroxisome biogenesis disorder, type 6A (Zellweger syndrome); Peroxisome biogenesis disorder, type 6B	Autosomal recessive
PEX11B	603867	1	614920	?Peroxisome biogenesis disorder 14B	Autosomal recessive
PEX12	601758	17	614859	Peroxisome biogenesis disorder type 3A (Zellweger)	Autosomal recessive
PEX13	601789	2	614883; 614885	Peroxisome biogenesis disorder, type 11A (Zellweger syndrome); Peroxisome biogenesis disorder, type 11B	Autosomal recessive
PEX14	601791	1	614887	Peroxisome biogenesis disorder, type 13A (Zellweger syndrome)	Autosomal recessive
PEX16	603360	11	614876; 614877	Peroxisome biogenesis disorder, type 8A (Zellweger syndrome); Peroxisome biogenesis disorder, type 8B	Autosomal recessive
PEX19	600279	1	614886	Peroxisome biogenesis disorder, type 12A (Zellweger syndrome)	Autosomal recessive
PEX2	170993	8	614866	Peroxisome biogenesis disorder type 5A (Zellweger)	Autosomal recessive
PEX26	608666	22	614872	Peroxisome biogenesis disorder type 7A (Zellweger)	Autosomal recessive
PEX3	603164	6	614882	Peroxisome biogenesis disorder, type 10A (Zellweger syndrome)	Autosomal recessive
PEX5	600414	12	214110	Peroxisome biogenesis disorder type 2A (Zellweger)	Autosomal recessive
PEX6	601498	6	614862; 616617*; 614863	Peroxisome biogenesis disorder, type 4A (Zellweger syndrome); Peroxisome biogenesis disorder, type 4B; Heimler syndrome 2	Autosomal recessive; Autosomal recessive*; Autosomal recessive
PEX7	601757	6	215100	Rhizomelic chondrodysplasia punctata, type 1	Autosomal recessive
PFKM	610681	12	232800	Glycogen storage disease, type 7	Autosomal recessive
PGAM2	612931	7	261670	Glycogen storage disease X	Autosomal recessive
PGAP1	611655	2	615802	Mental retardation, autosomal recessive 42	Autosomal recessive
PGAP2	615187	11	614207	Hyperphosphatasia with mental retardation syndrome 3	Autosomal recessive
PGAP3	611801	17	615716	Hyperphosphatasia with mental retardation syndrome 4	Autosomal recessive
PGK1	311800	X	300653	Phosphoglycerate kinase 1 deficiency	X-linked
PGM1	171900	1	614921	Congenital disorder of glycosylation, type 1t	Autosomal recessive
PGM3	172100	6	615816	Immunodeficiency, type 23	Autosomal recessive
PHF8	300560	X	300263	Mental retardation syndrome, X-linked, Siderius type	X-linked
PHGDH	606879	1	256520; 601815	Neu-Laxova syndrome, type 1; Phosphoglycerate dehydrogenase deficiency	Autosomal recessive
PHKB	172490	16	261750	Glycogen storage disease, type 9B	Autosomal recessive
PHKG2	172471	16	613027	Glycogen storage disease type 9c	Autosomal recessive
PHOX2A	602753	11	602078	Fibrosis of extraocular muscles, congenital, 2	Autosomal recessive
PHYH	602026	10	266500	Refsum disease	Autosomal recessive

PI4KA	600286	22		616531	Polymicrogyria, perisylvian, with cerebellar hypoplasia and arthrogryposis	Autosomal recessive
PIBF1	607532	13		617767	Joubert syndrome 33	Autosomal recessive
PIEZO1	611184	16		616843	Lymphedema, hereditary, type 3	Autosomal recessive
PIEZO2	613629	18		617146	Arthrogryposis, distal, with impaired proprioception and touch	Autosomal recessive
PIGC	601730	1		617816	Glycosylphosphatidylinositol biosynthesis defect 16	Autosomal recessive
PIGG	616918	4		616917	Mental retardation, autosomal recessive 53	Autosomal recessive
PIGL	605947	17		280000	Zunich neuroectodermal syndrome	Autosomal recessive
PIGM	610273	1		610293	Glycosylphosphatidylinositol deficiency	Autosomal recessive
PIGN	606097	18		614080	Multiple congenital anomalies-hypotonia-seizures syndrome, type 1	Autosomal recessive
PIGO	614730	9		614749	Hyperphosphatasia with mental retardation syndrome 2	Autosomal recessive
PIGT	610272	20		615398	Multiple congenital anomalies-hypotonia-seizures syndrome 3	Autosomal recessive
PIGV	610274	1		239300	Hyperphosphatasia with mental retardation syndrome 1	Autosomal recessive
PIGW	610275	17		616025	Glycosylphosphatidylinositol biosynthesis defect 11	Autosomal recessive
PIGY	610662	4		616809	Hyperphosphatasia with mental retardation syndrome 6	Autosomal recessive
PINK1	608309	1		605909	Parkinson disease, type 6, early onset	Autosomal recessive
PIP5K1C	606102	19		611369	Lethal congenital contractural syndrome, type 3	Autosomal recessive
PJVK	610219	2	DFNB59	610220	Deafness, autosomal recessive, type 59	Autosomal recessive
PKD1L1	609721	7		617205	Heterotaxy, visceral, 8, autosomal	Autosomal recessive
PKHD1	606702	6		263200	Polycystic kidney disease type 4	Autosomal recessive
PKLR	609712	1		266200	Pyruvate kinase deficiency	Autosomal recessive
PKP1	601975	1		604536	Ectodermal dysplasia/skin fragility syndrome	Autosomal recessive
PLA2G6	603604	22		256600	Infantile neuroaxonal dystrophy type 1	Autosomal recessive
PLAA	603873	9		617527	Neurodevelopmental disorder with progressive microcephaly, spasticity, and brain anomalies	Autosomal recessive
PLCB1	607120	20		613722	Epileptic encephalopathy, early infantile, type 12	Autosomal recessive
PLCB4	600810	20		614669*	Auriculocondylar syndrome, type 2	Autosomal recessive*
PLCD1	602142	3		151600	Nail disorder, nonsyndromic congenital, type 3 (leukonychia)	Autosomal recessive
PLCE1	608414	10		610725	Nephrotic syndrome, type 3	Autosomal recessive
PLD1	602382	3		212093	Cardiac valvular defect, developmental	Autosomal recessive
PLEC	601282	8		226670	Epidermolysis bullosa simplex with muscular dystrophy	Autosomal recessive
PLEKHG5	611101	1		615376	Charcot-Marie-Tooth disease, recessive intermediate, type C	Autosomal recessive
PLG	173350	6		217090	Plasminogen deficiency, type I	Autosomal recessive
PLK4	605031	4		616171	Microcephaly and chorioretinopathy, autosomal recessive, 2	Autosomal recessive
PLOD1	153454	1		225400	Ehlers-Danlos syndrome, kyphoscoliotic type, 1	Autosomal recessive
PLOD2	601865	3		609220	Bruck syndrome 2	Autosomal recessive
PLOD3	603066	7		612394	Lysyl hydroxylase 3 deficiency	Autosomal recessive
PLP1	300401	X		312080	Pelizaeus-Merzbacher disease	X-linked
PLPBP	604436	8	PROSC	617290	Epilepsy, early-onset, vitamin B6-dependent	Autosomal recessive
PMM2	601785	16		212065	Congenital disorder of glycosylation, type 1A	Autosomal recessive
PMP22	601097	17		145900*	Dejerine-Sottas disease	Autosomal recessive*
PMPCA	613036	9		213200	Spinocerebellar ataxia, autosomal recessive, type 2	Autosomal recessive
PMPCB	603131	7		617954	Multiple mitochondrial dysfunctions syndrome 6	Autosomal recessive
PNKP	605610	19		616267; 613402	Ataxia-oculomotor apraxia, type 4; Microcephaly, seizures, and developmental delay	Autosomal recessive
PNP	164050	14		613179	Immunodeficiency due to purine nucleoside phosphorylase deficiency	Autosomal recessive
PNPLA1	612121	6		615024	Ichthyosis, congenital, autosomal recessive, type 10	Autosomal recessive
PNPLA2	609059	11		610717	Neutral lipid storage disease with myopathy	Autosomal recessive
PNPLA6	603197	19		215470; 275400; 612020	Boucher-Neuhauser syndrome; Oliver-McFarlane syndrome; Spastic paraplegia, type 39, autosomal recessive	Autosomal recessive
PNPO	603287	17		610090	Pyridoxamine 5'-phosphate oxidase deficiency	Autosomal recessive
PNPT1	610316	2		614932	Combined oxidative phosphorylation deficiency 13	Autosomal recessive
POC1A	614783	3		614813	Short stature, onychodysplasia, facial dysmorphism, and hypotrichosis	Autosomal recessive
POC1B	614784	12		615973	Cone-rod dystrophy 20	Autosomal recessive
POLE	174762	12		615139	FILS syndrome	Autosomal recessive
POLG	174763	15		203700; 613662; 607459	POLG-related disorders	Autosomal recessive
POLH	603968	6		278750	Xeroderma pigmentosum, variant type	Autosomal recessive
POLR1C	610060	6		616494; 248390	Leukodystrophy, hypomyelinating, type 11; Treacher Collins syndrome 3	Autosomal recessive
POLR1D	613715	13		613717*	Treacher Collins syndrome, type 2	Autosomal recessive*

POLR3A	614258	10		607694	Leukodystrophy, hypomyelinating, type 7	Autosomal recessive
POLR3B	614366	12		614381	Leukodystrophy, hypomyelinating, type 8	Autosomal recessive
POMC	176830	2		609734	Obesity, adrenal insufficiency, and red hair due to POMC deficiency	Autosomal recessive
POMGNT1	606822	1		253280; 613151; 613157	Muscular dystrophy-dystroglycanopathy, type 3A (Walker-Warburg syndrome); Type 3B; Type 3C (limb-girdle muscular dystrophy, type 15 [LGMDR15])	Autosomal recessive
POMGNT2	614828	3		614830; 618135	Muscular dystrophy-dystroglycanopathy, type 8A (Walker-Warburg syndrome); Type 8C (limb-girdle muscular dystrophy, type 24 [LGMD R24])	Autosomal recessive
POMK	615247	8		615249	Muscular dystrophy-dystroglycanopathy (congenital with brain and eye anomalies), type A, 12	Autosomal recessive
POMP	613386	13		601952	Keratosis linearis with ichthyosis congenita and sclerosing keratoderma	Autosomal recessive
POMT1	607423	9		236670; 613155; 609308	Muscular dystrophy-dystroglycanopathy, type 1A (Walker-Warburg syndrome); Type 1B; Type 1C (limb-girdle muscular dystrophy, type 11 [LGMD R11])	Autosomal recessive
POMT2	607439	14		613150; 613156; 613158	Muscular dystrophy-dystroglycanopathy, type 2A (Walker-Warburg syndrome); Type 2B; Type 2C (limb-girdle muscular dystrophy, type 14 [LGMD R14])	Autosomal recessive
POP1	602486	8		617396	Anauxetic dysplasia, type 2	Autosomal recessive
POR	124015	7		201750	Antley-Bixler syndrome with genital anomalies and disordered steroidogenesis	Autosomal recessive
POU1F1	173110	3		613038*	Pituitary hormone deficiency, combined, type 1	Autosomal recessive*
POU3F4	300039	X		304400	Deafness, X-linked, type 2	X-linked
PPA2	609988	4		617222	Sudden cardiac failure, infantile	Autosomal recessive
PPIB	123841	15		259440	Osteogenesis imperfecta, type 9	Autosomal recessive
PPM1K	611065	4		615135	?Maple syrup urine disease, mild variant	Autosomal recessive
PPP1R15B	613257	1		616817	Microcephaly, short stature, and impaired glucose metabolism 2	Autosomal recessive
PPT1	600722	1		256730	Ceroid lipofuscinosis, neuronal, type 1	Autosomal recessive
PQBP1	300463	X		309500	Renpenning syndrome	X-linked
PRCD	610598	17		610599	Retinitis pigmentosa, type 36	Autosomal recessive
PRDM12	616458	9		616488	Neuropathy, hereditary sensory and autonomic, type VIII	Autosomal recessive
PRDM5	614161	4		614170	Brittle cornea syndrome, type 2	Autosomal recessive
PRDX1	176763	1		277400	Methylmalonic aciduria and homocystinuria, cblC type, digenic	Autosomal recessive, digenic inheritance (MMACHC gene)
PREPL	609557	2		616224	Myasthenic syndrome, congenital, type 22	Autosomal recessive
PRF1	170280	10		603553	Hemophagocytic lymphohistiocytosis, familial, type 2	Autosomal recessive
PRG4	604283	1		208250	Camptodactyly-arthropathy-coxa vara-pericarditis syndrome	Autosomal recessive
PRICKLE1	608500	12		612437	Epilepsy, progressive myoclonic, type 1B	Autosomal recessive
PRKCD	176977	3		615559	Autoimmune lymphoproliferative syndrome, type 3	Autosomal recessive
PRKN	602544	6	PARK2	600116	Parkinson disease, type 2, juvenile	Autosomal recessive
PRKRA	603424	2		612067	Dystonia, type 16	Autosomal recessive
PRMT7	610087	16		617157	Short stature, brachydactyly, intellectual developmental disability, and seizures	Autosomal recessive
PROC	612283	2		612304	Thrombophilia due to protein C deficiency, autosomal recessive	Autosomal recessive
PRODH	606810	22		239500	Hyperprolinemia, type 1	Autosomal recessive
PROM1	604365	4		612095	Retinitis pigmentosa, type 41	Autosomal recessive
PROP1	601538	5		262600	Pituitary hormone deficiency, combined, type 2	Autosomal recessive
PROS1	176880	3		614514	Thrombophilia due to protein S deficiency, autosomal recessive	Autosomal recessive
PRPH2	179605	6		608133*; 136880*	Leber congenital amaurosis 18; Retinitis punctata albescens	Autosomal recessive*
PRPS1	311850	X		300661; 304500; 311070; 301835	PRPS1-related disoders	X-linked
PRRX1	167420	1		202650*	Agnathia-otocephaly complex	Autosomal recessive*
PRSS1	276000	7		614044	Trypsinogen deficiency	Autosomal recessive
PRSS12	606709	4		249500	Mental retardation, autosomal recessive, type 1	Autosomal recessive
PRSS56	613858	2		613517	Microphthalmia, isolated, type 6	Autosomal recessive
PRUNE1	617413	1	PRUNE	617481	Neurodevelopmental disorder with microcephaly, hypotonia, and variable brain anomalies	Autosomal recessive
PRX	605725	19		614895	Charcot-Marie-Tooth disease, type 4F	Autosomal recessive
PSAP	176801	10		611721	Combined SAP deficiency	Autosomal recessive
PSAT1	610936	9		616038	Neu-Laxova syndrome, type 2	Autosomal recessive
PSMB8	177046	6		256040	Autoinflammation, lipodystrophy, and dermatosis syndrome	Autosomal recessive
PSMC3IP	608665	17		614324	Ovarian dysgenesis 3	Autosomal recessive
PSPH	172480	7		614023	Phosphoserine phosphatase deficiency	Autosomal recessive

PTF1A	607194	10		615935	Pancreatic agenesis 2	Autosomal recessive
PTH	168450	11		146200*	Hypoparathyroidism, familial isolated, type 1	Autosomal recessive*
PTH1R	168468	3		215045; 600002	Chondrodysplasia, Blomstrand type; Eiken syndrome	Autosomal recessive
PTPN23	606584	3		618890	Neurodevelopmental disorder and structural brain anomalies with or without seizures and spasticity	Autosomal recessive
PTPRC	151460	1		608971	Severe combined immunodeficiency, T cell-negative, B-cell/natural killer-cell positive	Autosomal recessive
PTPRO	600579	12		614196	Nephrotic syndrome, type 6	Autosomal recessive
PTPRQ	603317	12		613391	Deafness, autosomal recessive, type 84A	Autosomal recessive
PTRH2	608625	17		616263	Infantile-onset multisystem neurologic, endocrine, and pancreatic disease	Autosomal recessive
PTS	612719	11		261640	Hyperphenylalaninemia, BH4-deficient, type A	Autosomal recessive
PUS1	608109	12		600462	Myopathy, lactic acidosis, and sideroblastic anemia, type 1	Autosomal recessive
PXDN	605158	2		269400	Anterior segment dysgenesis, type 7, with sclerocornea	Autosomal recessive
PYCR1	179035	17		612940	Cutis laxa, autosomal recessive, type 2B	Autosomal recessive
PYCR2	616406	1		616420	Leukodystrophy, hypomyelinating, type 10	Autosomal recessive
PYGL	613741	14		232700	Glycogen storage disease, type 6	Autosomal recessive
PYGM	608455	11		232600	McArdle disease	Autosomal recessive
PYROXD1	617220	12		617258	Myopathy, myofibrillar, type 8	Autosomal recessive
QARS1	603727	3	QARS	615760	Microcephaly, progressive, seizures, and cerebral and cerebellar atrophy	Autosomal recessive
QDPR	612676	4		261630	Hyperphenylalaninemia, BH4-deficient, type C	Autosomal recessive
RAB18	602207	10		614222	Warburg micro syndrome, type 3	Autosomal recessive
RAB23	606144	6		201000	Carpenter syndrome	Autosomal recessive
RAB27A	603868	15		607624	Griscelli syndrome, type 2	Autosomal recessive
RAB28	612994	4		615374	Cone-rod dystrophy 18	Autosomal recessive
RAB33B	605950	4		615222	Smith-McCort dysplasia 2	Autosomal recessive
RAB3GAP1	602536	2		600118	Warburg micro syndrome, type 1	Autosomal recessive
RAB3GAP2	609275	1		212720	Martsolf syndrome	Autosomal recessive
RAD50	604040	5		613078	Nijmegen breakage syndrome-like disorder	Autosomal recessive
RAD51C	602774	17		613390	Fanconi anemia, complementation group O	Autosomal recessive
RAG1	179615	11		603554; 601457	Omenn syndrome; Severe combined immunodeficiency, B cell-negative	Autosomal recessive
RAG2	179616	11		603554; 601457	Omenn syndrome; Severe combined immunodeficiency, B cell-negative	Autosomal recessive
RAPSN	601592	11		208150; 616326	Fetal akinesia deformation sequence, type 2; Myasthenic syndrome, congenital, type 11, associated with AChR deficiency	Autosomal recessive
RARB	180220	3		615524	Microphthalmia, syndromic 12	Autosomal recessive
RARS1	107820	5	RARS	616140	Leukodystrophy, hypomyelinating, type 9	Autosomal recessive
RARS2	611524	6		611523	Pontocerebellar hypoplasia, type 6	Autosomal recessive
RASGRP1	603962	15		618534	Immunodeficiency, type 64	Autosomal recessive
RAX	601881	18		611038	Isolated microphthalmia, type 3	Autosomal recessive
RBBP8	604124	18		251255; 606744	Jawad syndrome; Seckel syndrome, type 2	Autosomal recessive
RBCK1	610924	20		615895	Polyglucosan body myopathy 1 with or without immunodeficiency	Autosomal recessive
RBM8A	605313	1		274000	Thrombocytopenia-absent radius syndrome	Autosomal recessive
RBP3	180290	10		615233	?Retinitis pigmentosa 66	Autosomal recessive
RBP4	180250	10		615147	Retinal dystrophy, iris coloboma, and comedogenic acne syndrome	Autosomal recessive
RCBTB1	607867	13		617175	Retinal dystrophy with or without extraocular anomalies	Autosomal recessive
RD3	180040	1		610612	Leber congenital amaurosis, type 12	Autosomal recessive
RDH12	608830	14		612712	Leber congenital amaurosis, type 13	Autosomal recessive
RDH5	601617	12		136880*	Fundus albipunctatus	Autosomal recessive*
RDX	179410	11		611022	Deafness, autosomal recessive, type 24	Autosomal recessive
RECQL4	603780	8		218600; 266280; 268400	Baller-Gerold syndrome; RAPADILINO syndrome; Rothmund-Thomson syndrome	Autosomal recessive
REEP6	609346	19		617304	Retinitis pigmentosa 77	Autosomal recessive
RELN	600514	7		257320	Lissencephaly 2 (Norman-Roberts type)	Autosomal recessive
REN	179820	1		267430	Renal tubular dysgenesis	Autosomal recessive
RETREG1	613114	5	FAM134B	613115	Neuropathy, hereditary sensory and autonomic, type 2B	Autosomal recessive
RFT1	611908	3		612015	Congenital disorder of glycosylation, type In	Autosomal recessive
RFX5	601863	1		209920	Bare lymphocyte syndrome, type 2	Autosomal recessive
RFX6	612659	6		615710	Mitchell-Riley syndrome	Autosomal recessive
RFXANK	603200	19		209920	Bare lymphocyte syndrome, type 2, complementation group B	Autosomal recessive

RFXAP	601861	13		209920	Bare lymphocyte syndrome, type 2	Autosomal recessive
RGR	600342	10		613769	Retinitis pigmentosa, type 44	Autosomal recessive
RHO	180380	3		613731*; 136880*	Retinitis pigmentosa, type 4; Retinitis punctata albescens	Autosomal recessive*
RIN2	610222	20		613075	Macs syndrome	Autosomal recessive
RIPK4	605706	21		263650	Popliteal pterygium syndrome, Bartsocas-Papas type	Autosomal recessive
RIPOR2	611410	6	C6orf32	616515	Deafness, autosomal recessive, type 104	Autosomal recessive
RLBP1	180090	15		607475; 136880*	Bothnia retinal dystrophy; Fundus albipunctatus	Autosomal recessive; Autosomal recessive*
RMND1	614917	6		614922	Combined oxidative phosphorylation deficiency 11	Autosomal recessive
RMRP	157660	9	CHH	607095	Anauxetic dysplasia, type 1	Autosomal recessive
RNASEH1	604123	2		616479	Progressive external ophthalmoplegia with mitochondrial DNA deletions, autosomal recessive 2	Autosomal recessive
RNASEH2A	606034	19		610333	Aicardi-Goutieres syndrome, type 4	Autosomal recessive
RNASEH2B	610326	13		610181	Aicardi-Goutieres syndrome, type 2	Autosomal recessive
RNASEH2C	610330	11		610329	Aicardi-Goutieres syndrome, type 3	Autosomal recessive
RNASET2	612944	6		612951	Leukoencephalopathy, cystic, without megalencephaly	Autosomal recessive
RNF168	612688	3		611943	RIDDLE syndrome	Autosomal recessive
RNF216	609948	7		212840	Gordon Holmes syndrome	Autosomal recessive
RNU4ATAC	601428	2		210710	Microcephalic osteodysplastic primordial dwarfism, type I	Autosomal recessive
ROBO3	608630	11		607313	Gaze palsy, familial horizontal, with progressive scoliosis, type 1	Autosomal recessive
ROGDI	614574	16		226750	Kohlschutter-Tonz syndrome	Autosomal recessive
ROM1	180721	11		608133	Retinitis pigmentosa, type 7, digenic	Autosomal recessive
ROR2	602337	9		268310	Robinow syndrome, autosomal recessive	Autosomal recessive
RORC	602943	1		616622	Immunodeficiency, type 42	Autosomal recessive
RP1	603937	8		180100	Retinitis pigmentosa, type 1	Autosomal recessive
RP2	300757	X		312600	Retinitis pigmentosa, type 2, X-linked	X-linked
RPE65	180069	1		204100; 613794	RPE65-related Leberÿcongenitalÿamaurosis/early-onset severe retinal dystrophyÿ	Autosomal recessive
RPGR	312610	X		300029; 304020	Retinitis pigmentosa, type 3, X-linked; Cone-rod dystrophy, X-linked, 1	X-linked
RPGRIP1	605446	14		613826	Leber congenital amaurosis, type 6	Autosomal recessive
RPGRIP1L	610937	16		611560; 611561; 619113	Joubert syndrome, type 7; Meckel syndrome, type 5; COACH syndrome	Autosomal recessive
RRM2B	604712	8		612075	Mitochondrial DNA depletion syndrome, type 8A (encephalomyopathic type with renal tubulopathy) and type 8B (MNGIE type)	Autosomal recessive
RS1	300839	X		312700	Retinoschisis	X-linked
RSPH1	609314	21		615481	Ciliary dyskinesia, primary, type 24	Autosomal recessive
RSPH3	615876	6		616481	Ciliary dyskinesia, primary, type 32	Autosomal recessive
RSPH4A	612647	6		612649	Ciliary dyskinesia, primary, type 11	Autosomal recessive
RSPH9	612648	6		612650	Ciliary dyskinesia, primary, type 12	Autosomal recessive
RSPO4	610573	20		206800	Anonychia congenita	Autosomal recessive
RSPRY1	616585	16		616723	Spondyloepimetaphyseal dysplasia, Faden-Alkuraya type	Autosomal recessive
RTEL1	608833	20		615190*	Dyskeratosis congenita, autosomal recessive type 5	Autosomal recessive*
RTN4IP1	610502	6		616732	Optic atrophy 10 with or without ataxia, mental retardation, and seizures	Autosomal recessive
RTTN	610436	18		614833	Microcephaly, short stature, and polymicrogyria with seizures	Autosomal recessive
RUSC2	611053	9		617773	Mental retardation, autosomal recessive 61	Autosomal recessive
RXYLT1	605862	12	TMEM5	615041	Muscular dystrophy-dystroglycanopathy (congenital with brain and eye anomalies), type A, 10	Autosomal recessive
RYR1	180901	19		255320	Minicore myopathy with external ophthalmoplegia	Autosomal recessive
S1PR2	605111	19		610419	Deafness, autosomal recessive, type 68	Autosomal recessive
SACS	604490	13		270550	Spastic ataxia, Charlevoix-Saguenay, type	Autosomal recessive
SAG	181031	2		258100	Oguchi disease, type 1	Autosomal recessive
SAMD9	610456	7		610455	Tumoral calcinosis, familial, normophosphatemic	Autosomal recessive
SAMHD1	606754	20		612952	Aicardi-Goutieres syndrome, type 5	Autosomal recessive
SAR1B	607690	5		246700	Chylomicron retention disease	Autosomal recessive
SARS2	612804	19		613845	Hyperuricemia, pulmonary hypertension, renal failure, and alkalosis	Autosomal recessive
SBDS	607444	7		260400	Shwachman-Diamond syndrome	Autosomal recessive
SBF1	603560	22		615284	Charcot-Marie-Tooth disease, type 4B3	Autosomal recessive
SBF2	607697	11		604563	Charcot-Marie-Tooth disease, type 4B2	Autosomal recessive
SC5D	602286	11	SC5DL	607330	Lathosterolosis	Autosomal recessive

SCARB2	602257	4		254900	Epilepsy, progressive myoclonic, type 4, with or without renal failure	Autosomal recessive
SCARF2	613619	22		600920	Van den Ende-Gupta syndrome	Autosomal recessive
SCN1B	600235	19		617350	Epileptic encephalopathy, early infantile, type 52	Autosomal recessive
SCN4A	603967	17		614198	Myasthenic syndrome, congenital, type 16	Autosomal recessive
SCN9A	603415	2		243000	Indifference to pain and autosomal recessive hereditary sensory neuropathy type 2D	Autosomal recessive
SCNN1A	600228	12		264350	Pseudohypoaldosteronism, type 1	Autosomal recessive
SCNN1B	600760	16		264350	Pseudohypoaldosteronism, type 1	Autosomal recessive
SCNN1G	600761	16		264350	Pseudohypoaldosteronism, type 1	Autosomal recessive
SCO1	603644	17		619048	Mitochondrial complex IV deficiency, nuclear type 4	Autosomal recessive
SCO2	604272	22		604377	Cardioencephalomyopathy, fatal infantile, due to cytochrome c oxidase deficiency, type 1	Autosomal recessive
SCYL1	607982	11		616719	Spinocerebellar ataxia, autosomal recessive, type 21	Autosomal recessive
SDCCAG8	613524	1		615993	Bardet-Biedl syndrome, type 16	Autosomal recessive
SDHA	600857	5		252011; 256000	Mitochondrial respiratory chain complex II deficiency; Leigh syndrome	Autosomal recessive
SDHAF1	612848	19		252011	Mitochondrial complex II deficiency	Autosomal recessive
SDR9C7	609769	12		617574	Ichthyosis, congenital, autosomal recessive 13	Autosomal recessive
SEC23A	610511	14		607812	Craniolenticulosutural dysplasia	Autosomal recessive
SEC23B	610512	20		224100	Dyserythropoietic anemia, congenital, type 2	Autosomal recessive
SEC24D	607186	4		616294	Cole-Carpenter syndrome 2	Autosomal recessive
SECISBP2	607693	9		609698	Thyroid hormone metabolism, abnormal	Autosomal recessive
SELENON	606210	1	SEPN1	602771	Muscular dystrophy, rigid spine, type 1	Autosomal recessive
SEMA4A	607292	1		610283; 610282	Cone-rod dystrophy, type 10; Retinitis pigmentosa, type 35	Autosomal recessive
SEPSECS	613009	4		613811	Pontocerebellar hypoplasia, type 2D	Autosomal recessive
SERAC1	614725	6		614739	3-methylglutaconic aciduria with deafness, encephalopathy, and Leigh-like syndrome (MEGDEL)	Autosomal recessive
SERPINA1	107400	14		613490	Alpha-1 antitrypsin deficiency	Autosomal recessive
SERPINB7	603357	18		615598	Palmoplantar keratoderma, Nagashima type	Autosomal recessive
SERPINB8	601697	18		617115	Peeling skin syndrome 5	Autosomal recessive
SERPINC1	107300	1		613118*	Thrombophilia due to antithrombin III deficiency	Autosomal recessive*
SERPINE1	173360	7		613329*	Plasminogen activator inhibitor-1 deficiency	Autosomal recessive*
SERPINF1	172860	17		613982	Osteogenesis imperfecta, type 6	Autosomal recessive
SERPINF2	613168	17		262850	Alpha-2-plasmin inhibitor deficiency	Autosomal recessive
SERPING1	606860	11		106100*	Angioedema, hereditary, types 1 and 2	Autosomal recessive*
SERPINH1	600943	11		613848	Osteogenesis imperfecta, type 10	Autosomal recessive
SETX	608465	9		606002	Spinocerebellar ataxia, autosomal recessive, type 1	Autosomal recessive
SFRP4	606570	7		265900	Pyle disease	Autosomal recessive
SFTPB	178640	2		265120	Surfactant metabolism dysfunction, pulmonary, type 1	Autosomal recessive
SFXN4	615564	10		615578	Combined oxidative phosphorylation deficiency 18	Autosomal recessive
SGCA	600119	17		608099	Limb-girdle muscular dystrophy, type 3 (LGMD R3)	Autosomal recessive
SGCB	600900	4		604286	Limb-girdle muscular dystrophy, type 4 (LGMD R4)	Autosomal recessive
SGCD	601411	5		601287	Limb-girdle muscular dystrophy, type 6 (LGMD R6)	Autosomal recessive
SGCG	608896	13		253700	Limb-girdle muscular dystrophy, type 5 (LGMD R5)	Autosomal recessive
SGPL1	603729	10		617575	Nephrotic syndrome, type 14	Autosomal recessive
SGSH	605270	17		252900	Mucopolysaccharidosis, type 3A (Sanfilippo A)	Autosomal recessive
SH2D1A	300490	X		308240	Lymphoproliferative syndrome, X-linked, type 1	X-linked
SH3PXD2B	613293	5		249420	Frank-ter Haar syndrome	Autosomal recessive
SH3TC2	608206	5		601596	Charcot-Marie-Tooth disease, type 4C	Autosomal recessive
SI	609845	3		222900	Sucrase-isomaltase deficiency, congenital	Autosomal recessive
SIL1	608005	5		248800	Marinesco-Sjogren syndrome	Autosomal recessive
SIX6	606326	14		212550	Optic disc anomalies with retinal and/or macular dystrophy	Autosomal recessive
SKIV2L	600478	6		614602	Trichohepatoenteric syndrome, type 2 (diarrhea, syndromic)	Autosomal recessive
SLC10A2	601295	13		613291	Bile acid malabsorption, primary	Autosomal recessive
SLC11A2	600523	12		206100	Anemia, hypochromic microcytic, with iron overload 1	Autosomal recessive
SLC12A1	600839	15		601678	Bartter syndrome, type 1	Autosomal recessive
SLC12A3	600968	16		263800	Gitelman syndrome	Autosomal recessive
SLC12A5	606726	20		616645	Epileptic encephalopathy, early infantile, 34	Autosomal recessive
SLC12A6	604878	15		218000	Agenesis of the corpus callosum with peripheral neuropathy	Autosomal recessive

SLC13A5	608305	17	615905	Epileptic encephalopathy, early infantile, 25	Autosomal recessive
SLC16A1	600682	1	616095*	Monocarboxylate transporter 1 deficiency	Autosomal recessive*
SLC16A2	300095	X	300523	Allan-Herndon-Dudley syndrome	X-linked
SLC17A5	604322	6	604369	Salla disease	Autosomal recessive
SLC18A3	600336	10	617239	Myasthenic syndrome, congenital, 21, presynaptic	Autosomal recessive
SLC19A2	603941	1	249270	Thiamine-responsive megaloblastic anemia syndrome	Autosomal recessive
SLC19A3	606152	2	607483	Thiamine metabolism dysfunction syndrome, type 2 (biotin- or thiamine-responsive encephalopathy type)	Autosomal recessive
SLC1A1	133550	9	222730	Dicarboxylic aminoaciduria	Autosomal recessive
SLC1A4	600229	2	616657	Spastic tetraplegia, thin corpus callosum, and progressive microcephaly	Autosomal recessive
SLC22A12	607096	11	220150	Hypouricemia, renal	Autosomal recessive
SLC22A5	603377	5	212140	Carnitine deficiency, systemic primary	Autosomal recessive
SLC24A1	603617	15	613830	Night blindness, congenital stationary (complete), type 1D, autosomal recessive	Autosomal recessive
SLC24A4	609840	14	615887	Amelogenesis imperfecta, type IIA5	Autosomal recessive
SLC24A5	609802	15	113750	Albinism, oculocutaneous, type 6	Autosomal recessive
SLC25A1	190315	22	615182	Combined D-2- and L-2-hydroxyglutaric aciduria	Autosomal recessive
SLC25A12	603667	2	612949	Epileptic encephalopathy, early infantile, type 39	Autosomal recessive
SLC25A13	603859	7	605814; 603471	Citrullinemia, type 2, neonatal-onset; Citrullinemia, type 2, adult-onset	Autosomal recessive
SLC25A15	603861	13	238970	Hyperornithinemia-hyperammonemia-homocitrullinemia syndrome	Autosomal recessive
SLC25A19	606521	17	607196; 613710	Microcephaly, Amish type; Thiamine metabolism dysfunction syndrome 4 (progressive polyneuropathy type)	Autosomal recessive
SLC25A20	613698	3	212138	Carnitine-acylcarnitine translocase deficiency	Autosomal recessive
SLC25A22	609302	11	609304	Epileptic encephalopathy, early infantile, type 3	Autosomal recessive
SLC25A26	611037	3	616794	Combined oxidative phosphorylation deficiency 28	Autosomal recessive
SLC25A3	600370	12	610773	Mitochondrial phosphate carrier deficiency	Autosomal recessive
SLC25A38	610819	3	205950	Anemia, sideroblastic, type 2, pyridoxine-refractory	Autosomal recessive
SLC25A4	103220	4	615418	Mitochondrial DNA depletion syndrome, type 12B (cardiomyopathic type) AR	Autosomal recessive
SLC25A46	610826	5	616505	Neuropathy, hereditary motor and sensory, type VIB	Autosomal recessive
SLC26A2	606718	5	600972	Achondrogenesis, type 1B (diastrophic dysplasia)	Autosomal recessive
SLC26A3	126650	7	214700	Diarrhea 1, secretory chloride, congenital	Autosomal recessive
SLC26A4	605646	7	600791; 274600	Deafness, autosomal recessive, type 4; Pendred syndrome	Autosomal recessive
SLC26A5	604943	7	613865	?Deafness, autosomal recessive, type 61	Autosomal recessive
SLC27A4	604194	9	608649	Ichthyosis prematurity syndrome	Autosomal recessive
SLC29A3	612373	10	602782	Histiocytosis-lymphadenopathy plus syndrome	Autosomal recessive
SLC2A1	138140	1	606777*	GLUT1 deficiency syndrome 1, infantile onset, severe	Autosomal recessive*
SLC2A10	606145	20	208050	Arterial tortuosity syndrome	Autosomal recessive
SLC2A2	138160	3	227810	Fanconi-Bickel syndrome	Autosomal recessive
SLC2A9	606142	4	612076*	Hypouricemia, renal, type 2	Autosomal recessive*
SLC30A10	611146	1	613280	Hypermanganesemia with dystonia, type 1	Autosomal recessive
SLC33A1	603690	3	614482	Congenital cataracts, hearing loss, and neurodegeneration	Autosomal recessive
SLC34A1	182309	5	616963	Hypercalcemia, infantile, type 2	Autosomal recessive
SLC34A2	604217	4	265100	Pulmonary alveolar microlithiasis	Autosomal recessive
SLC34A3	609826	9	241530	Hypophosphatemic rickets with hypercalciuria	Autosomal recessive
SLC35A1	605634	6	603585	Congenital disorder of glycosylation, type 2F	Autosomal recessive
SLC35A3	605632	1	615553	?Arthrogryposis, mental retardation, and seizures	Autosomal recessive
SLC35C1	605881	11	266265	Congenital disorder of glycosylation, type 2C	Autosomal recessive
SLC35D1	610804	1	269250	Schneckenbecken dysplasia	Autosomal recessive
SLC37A4	602671	11	232220	Glycogen storage disease, type 1B	Autosomal recessive
SLC38A8	615585	16	609218	Foveal hypoplasia 2, with or without optic nerve misrouting and/or anterior segment dysgenesis	Autosomal recessive
SLC39A13	608735	11	612350	Ehlers-Danlos syndrome, spondylodysplastic type, 3	Autosomal recessive
SLC39A14	608736	8	617013	Hypermanganesemia with dystonia 2	Autosomal recessive
SLC39A4	607059	8	201100	Acrodermatitis enteropathica	Autosomal recessive
SLC39A8	608732	4	616721	Congenital disorder of glycosylation, type IIn	Autosomal recessive
SLC3A1	104614	2	220100*	Cystinuria	Autosomal recessive*
SLC45A1	605763	1	617532	Intellectual developmental disorder with neuropsychiatric features	Autosomal recessive
SLC45A2	606202	5	606574	Albinism, oculocutaneous, type 4	Autosomal recessive
SLC46A1	611672	17	229050	Folate malabsorption, hereditary	Autosomal recessive

SLC4A1	109270	17		611590	Distal renal tubular acidosis	Autosomal recessive
SLC4A11	610206	20		217700	Corneal endothelial dystrophy, autosomal recessive	Autosomal recessive
SLC4A4	603345	4		604278	Renal tubular acidosis, proximal, with ocular abnormalities	Autosomal recessive
SLC52A2	607882	8		614707	Brown-Vialetto-Van Laere syndrome, type 2	Autosomal recessive
SLC52A3	613350	20		211530	Brown-Vialetto-Van Laere syndrome, type 1	Autosomal recessive
SLC5A1	182380	22		606824	Glucose/galactose malabsorption	Autosomal recessive
SLC5A2	182381	16		233100*	Renal glucosuria	Autosomal recessive*
SLC5A5	601843	19		274400	Thyroid dyshormonogenesis, type 1	Autosomal recessive
SLC5A7	608761	2		617143	Myasthenic syndrome, congenital, type 20, presynaptic	Autosomal recessive
SLC6A17	610299	1		616269	Mental retardation, autosomal recessive 48	Autosomal recessive
SLC6A19	608893	5		234500	Hartnup disorder	Autosomal recessive
SLC6A3	126455	5		613135	Parkinsonism-dystonia, infantile	Autosomal recessive
SLC6A5	604159	11		614618*	Hyperekplexia, type 3	Autosomal recessive*
SLC6A8	300036	X		300352	Cerebral creatine deficiency syndrome, type 1	X-linked
SLC6A9	601019	1		617301	Glycine encephalopathy with normal serum glycine	Autosomal recessive
SLC7A14	615720	3		615725	Retinitis pigmentosa 68	Autosomal recessive
SLC7A7	603593	14		222700	Lysinuric protein intolerance	Autosomal recessive
SLC7A9	604144	19		220100*	Cystinuria	Autosomal recessive*
SLC9A3	182307	5		616868	Diarrhea 8, secretory sodium, congenital	Autosomal recessive
SLCO2A1	601460	3		614441	Hypertrophic osteoarthropathy, primary, autosomal recessive, type 2	Autosomal recessive
SLITRK6	609681	13		221200	Deafness and myopia	Autosomal recessive
SLURP1	606119	8		248300	Meleda disease	Autosomal recessive
SLX4	613278	16		613951	Fanconi anemia, complementation group P	Autosomal recessive
SMARCAL1	606622	2		242900	Schimke immunoosseous dysplasia	Autosomal recessive
SMARCD2	601736	17		617475	Specific granule deficiency 2	Autosomal recessive
SMG9	613176	19		616920	Heart and brain malformation syndrome	Autosomal recessive
SMN1	600354	5		253300	Spinal muscular atrophy	Autosomal recessive
SMOC1	608488	14		206920	Microphthalmia. with limb anomalies	Autosomal recessive
SMOC2	607223	6		125400	Dentin dysplasia, type 1, with microdontia and misshapen teeth	Autosomal recessive
SMPD1	607608	11		257200; 607616	Niemann-Pick disease, type A; Niemann-Pick disease, type B	Autosomal recessive
SNAP29	604202	22		609528	Cerebral dysgenesis, neuropathy, ichthyosis, and palmoplantar keratoderma syndrome	Autosomal recessive
SNX10	614780	7		615085	Osteopetrosis, autosomal recessive, type 8	Autosomal recessive
SNX14	616105	6		616354	Spinocerebellar ataxia, autosomal recessive, type 20	Autosomal recessive
SOBP	613667	6		613671	Mental retardation, anterior maxillary protrusion, and strabismus	Autosomal recessive
SOD1	147450	21		618598; 105400*	Spastic tetraplegia and axial hypotonia, progressive; Amyotrophic lateral sclerosis, type 1	Autosomal recessive; Autosomal recessive*
SOHLH1	610224	9		617690	Ovarian dysgenesis 5	Autosomal recessive
SOST	605740	17		269500; 239100	Sclerosteosis, type 1; Van Buchem disease	Autosomal recessive
SOX18	601618	20		607823	Hypotrichosis-lymphedema-telangiectasia syndrome	Autosomal recessive
SP110	604457	2		235550	Hepatic venoocclusive disease with immunodeficiency	Autosomal recessive
SP7	606633	12		613849	Osteogenesis imperfecta, type XII	Autosomal recessive
SPAG1	603395	8		615505	Ciliary dyskinesia, primary, type 28	Autosomal recessive
SPARC	182120	5		616507	Osteogenesis imperfecta, type XVII	Autosomal recessive
SPART	607111	13	SPG20	275900	Spactic paraplegia, type 20, autosomal recessive	Autosomal recessive
SPATA5	613940	4		616577	Epilepsy, hearing loss, and mental retardation syndrome	Autosomal recessive
SPATA7	609868	14		604232	Leber congenital amaurosis, type 3	Autosomal recessive
SPEG	615950	2		615959	Centronuclear myopathy, type 5	Autosomal recessive
SPG11	610844	15		602099	Amyotrophic lateral sclerosis, type 5, juvenile	Autosomal recessive
SPG21	608181	15		248900	Mast syndrome	Autosomal recessive
SPG7	602783	16		607259	Spastic paraplegia, type 7, autosomal recessive	Autosomal recessive
SPINK1	167790	5		608189*	Tropical calcific pancreatitis	Autosomal recessive*
SPINK5	605010	5		256500	Netherton syndrome	Autosomal recessive
SPINT2	605124	19		270420	Diarrhea 3, secretory sodium, congenital, syndromic	Autosomal recessive
SPR	182125	2		612716*	Dystonia, dopa-responsive, due to sepiapterin reductase deficiency	Autosomal recessive*
SPRTN	616086	1		616200	Ruijs-Aalfs syndrome	Autosomal recessive

SPTA1	182860	1	266140; 270970	Pyropoikilocytosis; Apherocytosis, type 3	Autosomal recessive
SPTBN2	604985	11	615386	Spinocerebellar ataxia, autosomal recessive, type 14	Autosomal recessive
SPTBN4	606214	19	617519	Neurodevelopmental disorder with hypotonia, neuropathy, and deafness	Autosomal recessive
SQSTM1	601530	5	617145	Neurodegeneration with ataxia, dystonia, and gaze palsy, childhood-onset	Autosomal recessive
SRD5A2	607306	2	264600	46,XY disorder of sex development due to 5-alpha-reductase 2 deficiency (pseudovaginal perineoscrotal hypospadias)	Autosomal recessive
SRD5A3	611715	4	612379; 612713	Congenital disorder of glycosylation, type 1Q; Kahrizi syndrome	Autosomal recessive
ST14	606797	11	602400	Ichthyosis, congenital, autosomal recessive, type 11	Autosomal recessive
ST3GAL3	606494	1	611090	Mental retardation, autosomal recessive 12	Autosomal recessive
ST3GAL5	604402	2	609056	Salt and pepper developmental regression syndrome	Autosomal recessive
STAC3	615521	12	255995	Native American myopathy	Autosomal recessive
STAG3	608489	7	615723	Premature ovarian failure 8	Autosomal recessive
STAMBP	606247	2	614261	Microcephaly-capillary malformation syndrome	Autosomal recessive
STAR	600617	8	201710	Lipoid adrenal hyperplasia	Autosomal recessive
STAT1	600555	2	613796	Immunodeficiency, type 31B, mycobacterial and viral infections	Autosomal recessive
STAT2	600556	12	616636	Immunodeficiency, type 44	Autosomal recessive
STAT5B	604260	17	245590	Laron syndrome with immunodeficiency	Autosomal recessive
STIL	181590	1	612703	Microcephaly, type 7, primary, autosomal recessive	Autosomal recessive
STIM1	605921	11	612783	Immunodeficiency, type 10	Autosomal recessive
STK4	604965	20	614868	T-cell immunodeficiency, recurrent infections, autoimmunity, and cardiac malformations	Autosomal recessive
STRA6	610745	15	601186	Microphthalmia, isolated, with coloboma, type 8	Autosomal recessive
STRADA	608626	17	611087	Polyhydramnios, megalencephaly, and symptomatic epilepsy	Autosomal recessive
STRC	606440	15	603720	Deafness, autosomal recessive, type 16	Autosomal recessive
STUB1	607207	16	615768	Spinocerebellar ataxia, autosomal recessive, type 16	Autosomal recessive
STX11	605014	6	603552	Hemophagocytic lymphohistiocytosis, familial, type 4	Autosomal recessive
STXBP2	601717	19	613101	Hemophagocytic lymphohistiocytosis, familial, type 5	Autosomal recessive
SUCLA2	603921	13	612073	Mitochondrial DNA depletion syndrome, type 5 (encephalomyopathic with or without methylmalonic aciduria)	Autosomal recessive
SUCLG1	611224	2	245400	Mitochondrial DNA depletion syndrome, type 9 (encephalomyopathic, type with methylmalonic aciduria)	Autosomal recessive
SUFU	607035	10	617757	Joubert syndrome, type 32	Autosomal recessive
SUGCT	609187	7	231690	Glutaric aciduria, type 3	Autosomal recessive
SULT2B1	604125	19	617571	Ichthyosis, congenital, autosomal recessive, type 14	Autosomal recessive
SUMF1	607939	3	272200	Multiple sulfatase deficiency	Autosomal recessive
SUN5	613942	20	617187	Spermatogenic failure, type 16	Autosomal recessive
SUOX	606887	12	272300	Sulfite oxidase deficiency	Autosomal recessive
SURF1	185620	9	616684; 256000	Charcot-Marie-Tooth disease, type 4K; Leigh syndrome, due to COX IV deficiency	Autosomal recessive
SYN1	313440	X	300491	Epilepsy, X-linked, with variable learning disabilities and behavior disorders	X-linked
SYNE1	608441	6	610743	Spinocerebellar ataxia, autosomal recessive, type 8	Autosomal recessive
SYNE4	615535	19	615540	Deafness, autosomal recessive, type 76	Autosomal recessive
SYNJ1	604297	21	617389	Epileptic encephalopathy, early infantile, 53	Autosomal recessive
SYT14	610949	1	614229	?Spinocerebellar ataxia, autosomal recessive, type 11	Autosomal recessive
SZT2	615463	1	615476	Epileptic encephalopathy, early infantile, 18	Autosomal recessive
TAC3	162330	12	614839	Hypogonadotropic hypogonadism, type 10, with or without anosmia	Autosomal recessive
TACO1	612958	17	619052	Mitochondrial complex IV deficiency, nuclear type 8	Autosomal recessive
TACR3	162332	4	614840	Hypogonadotropic hypogonadism, type 11, with or without anosmia	Autosomal recessive
TACSTD2	137290	1	204870	Corneal dystrophy, gelatinous drop-like	Autosomal recessive
TAF13	600774	1	617432	Mental retardation, autosomal recessive 60	Autosomal recessive
TAF2	604912	8	615599	Mental retardation, autosomal recessive 40	Autosomal recessive
TAF6	602955	7	617126	Alazami-Yuan syndrome	Autosomal recessive
TALDO1	602063	11	606003	Transaldolase deficiency	Autosomal recessive
TANGO2	616830	22	616878	Metabolic encephalomyopathic crises, recurrent, with rhabdomyolysis, cardiac arrhythmias, and neurodegeneration	Autosomal recessive
TAP1	170260	6	604571	Bare lymphocyte syndrome, type 1	Autosomal recessive
TAP2	170261	6	604571	Bare lymphocyte syndrome, type 1, due to TAP2 deficiency	Autosomal recessive
TAPBP	601962	6	604571	Bare lymphocyte syndrome, type 1	Autosomal recessive
TAPT1	612758	4	616897	Osteochondrodysplasia, complex lethal, Symoens-Barnes-Gistelinck type	Autosomal recessive
TAT	613018	16	276600	Tyrosinemia, type 2	Autosomal recessive
TBC1D20	611663	20	615663	Warburg micro syndrome 4	Autosomal recessive

TBC1D23	617687	3	617695	Pontocerebellar hypoplasia, type 11	Autosomal recessive
TBC1D24	613577	16	220500; 615338; 614617	DOORS (deafness, onychodystrophy, osteodystrophy, mental retardation, and seizures) syndrome; Epileptic encephalopathy, early infantile, type 16; Deafness, autosomal recessive, type 86	Autosomal recessive
TBC1D7	612655	6	248000	Macrocephaly/megalencephaly syndrome, autosomal recessive	Autosomal recessive
TBCD	604649	17	617193	Encephalopathy, progressive, early-onset, with brain atrophy and thin corpus callosum	Autosomal recessive
TBCE	604934	1	617207; 241410; 244460	Encephalopathy, progressive, with amyotrophy and optic atrophy; Hypoparathyroidism-retardation-dysmorphism syndrome; Kenny-Caffey syndrome, type 1	Autosomal recessive
TBCK	616899	4	616900	Hypotonia, infantile, with psychomotor retardation and characteristic facies 3	Autosomal recessive
TBX15	604127	1	260660	Cousin syndrome	Autosomal recessive
TBX19	604614	1	201400	Congenital isolated adrenocorticotropic hormone deficiency	Autosomal recessive
TBXAS1	274180	7	231095	Ghosal syndrome	Autosomal recessive
TCAP	604488	17	601954	Limb-girdle muscular dystrophy, type 7 (LGMD R7)	Autosomal recessive
TCIRG1	604592	11	259700	Osteopetrosis, autosomal recessive, type 1	Autosomal recessive
TCN2	613441	22	275350	Transcobalamin II deficiency	Autosomal recessive
TCTN1	609863	12	614173	Joubert syndrome, type 13	Autosomal recessive
TCTN2	613846	12	616654; 613885	Joubert syndrome, type 24; ?Meckel syndrome, type 8	Autosomal recessive
TCTN3	613847	10	614815	Joubert syndrome 18	Autosomal recessive
TDP1	607198	14	607250	?Spinocerebellar ataxia, autosomal recessive with axonal neuropathy	Autosomal recessive
TDP2	605764	6	616949	Spinocerebellar ataxia, autosomal recessive, type 23	Autosomal recessive
TDRD7	611258	9	613887	Cataract 36	Autosomal recessive
TECPR2	615000	14	615031	Spastic paraplegia, type 49, autosomal recessive	Autosomal recessive
TECR	610057	19	614020	Mental retardation, autosomal recessive, type 14	Autosomal recessive
TECRL	617242	4	614021	Ventricular tachycardia, catecholaminergic polymorphic, 3	Autosomal recessive
TECTA	602574	11	603629	Deafness, autosomal recessive, type 21	Autosomal recessive
TELO2	611140	16	616954	You-Hoover-Fong syndrome	Autosomal recessive
TENM3	610083	4	615145	Microphthalmia, isolated, with coloboma 9	Autosomal recessive
TERT	187270	5	613989	Dyskeratosis congenita, autosomal recessive, type 4	Autosomal recessive
TEX15	605795	8	617960	Spermatogenic failure, type 25	Autosomal recessive
TF	190000	3	209300	Atransferrinemia	Autosomal recessive
TFR2	604720	7	604250	Hemochromatosis, type 3	Autosomal recessive
TFRC	190010	3	616740	Immunodeficiency, type 46	Autosomal recessive
TG	188450	8	274700	Thyroid dyshormonogenesis, type 3	Autosomal recessive
TGDS	616146	13	616145	Catel-Manzke syndrome	Autosomal recessive
TGM1	190195	14	242300	Ichthyosis, congenital, autosomal recessive, type 1	Autosomal recessive
TGM5	603805	15	609796	Peeling skin syndrome, type 2	Autosomal recessive
TH	191290	11	605407	Segawa syndrome, recessive	Autosomal recessive
THOC2	300395	X	300957	Mental retardation, X-linked 12	X-linked
THOC6	615403	16	613680	Beaulieu-Boycott-Innes syndrome	Autosomal recessive
THRB	190160	3	274300	Thyroid hormone resistance, autosomal recessive	Autosomal recessive
TIMM50	607381	19	617698	3-methylglutaconic aciduria, type 9	Autosomal recessive
TIMMDC1	615534	3	618251	Mitochondrial complex I deficiency, nuclear type 31	Autosomal recessive
TJP2	607709	9	615878	Cholestasis, progressive familial intrahepatic, type 4	Autosomal recessive
TK2	188250	16	609560	Mitochondrial DNA depletion syndrome , type 2 (myopathic type)	Autosomal recessive
TKT	606781	3	617044	Short stature, developmental delay, and congenital heart defects	Autosomal recessive
TLE6	612399	19	616814	Preimplantation embryonic lethality	Autosomal recessive
TMC1	606706	9	600974	Deafness, autosomal recessive, type 7	Autosomal recessive
TMC6	605828	17	226400	Epidermodysplasia verruciformis	Autosomal recessive
TMC8	605829	17	226400	Epidermodysplasia verruciformis	Autosomal recessive
TMCO1	614123	1	213980	Craniofacial dysmorphism, skeletal anomalies, and mental retardation syndrome	Autosomal recessive
TMEM107	616183	17	617562; 617563	Meckel syndrome, type 13; Orofaciodigital syndrome, type 16	Autosomal recessive
TMEM126A	612988	11	612989	Optic atrophy 7	Autosomal recessive
TMEM126B	615533	11	618250	Mitochondrial complex I deficiency, nuclear type 29	Autosomal recessive
TMEM138	614459	11	614465	Joubert syndrome 16	Autosomal recessive
TMEM165	614726	4	614727	Congenital disorder of glycosylation, type 2K	Autosomal recessive
TMEM199	616815	17	616829	Congenital disorder of glycosylation, type 2P	Autosomal recessive

TMEM216	613277	11	608091; 603194	Joubert syndrome, type 2; Meckel syndrome, type 2	Autosomal recessive
TMEM231	614949	16	614970; 615397	Joubert syndrome, type 20; Meckel syndrome,type 11	Autosomal recessive
TMEM237	614423	2	614424	Joubert syndrome, type 14	Autosomal recessive
TMEM260	617449	14	617478	Structural heart defects and renal anomalies syndrome	Autosomal recessive
TMEM67	609884	8	610688; 607361; 216360	Joubert syndrome, type 6; Meckel syndrome, type 3; COACH syndrome	Autosomal recessive
TMEM70	612418	8	614052	Mitochondrial complex V (ATP synthase) deficiency, nuclear type 2	Autosomal recessive
TMIE	607237	3	600971	Deafness, autosomal recessive, type 6	Autosomal recessive
TMPRSS15	606635	21	226200	Enterokinase deficiency	Autosomal recessive
TMPRSS3	605511	21	601072	Deafness, autosomal recessive, type 8/10	Autosomal recessive
TMPRSS6	609862	22	206200	Iron-refractory iron deficiency anemia	Autosomal recessive
TMTC3	617218	12	617255	Lissencephaly 8	Autosomal recessive
TNFRSF11A	603499	18	612301	Osteopetrosis, autosomal recessive, type 7	Autosomal recessive
TNFRSF11B	602643	8	239000	Paget disease of bone, type 5, juvenile-onset	Autosomal recessive
TNFRSF13B	604907	17	240500	Immunodeficiency, common variable, type 2	Autosomal recessive
TNFSF11	602642	13	259710	Osteopetrosis, autosomal recessive, type 2	Autosomal recessive
TNIK	610005	3	617028	Mental retardation, autosomal recessive 54	Autosomal recessive
TNNT1	191041	19	605355	Nemaline myopathy , type 5, Amish type	Autosomal recessive
TNXB	600985	6	606408	Ehlers-Danlos syndrome, classic-like	Autosomal recessive
TOE1	613931	1	614969	Pontocerebellar hypoplasia, type 7	Autosomal recessive
TOP3A	601243	17	618097	Microcephaly, growth restriction, and increased sister chromatid exchange 2	Autosomal recessive
TP53RK	608679	20	617730	Galloway-Mowat syndrome 4	Autosomal recessive
TPI1	190450	12	615512	Hemolytic anemia due to triosephosphate isomerase deficiency	Autosomal recessive
TPK1	606370	7	614458	Episodic encephalopathy due to thiamine pyrophosphokinase deficiency	Autosomal recessive
TPM3	191030	1	609284*; 255310*	Nemaline myopathy, type 1; Congenital fiber-type disproportion myopathy	Autosomal recessive*
TPO	606765	2	274500	Thyroid dyshormonogenesis, type 2A	Autosomal recessive
TPP1	607998	11	204500; 609270	Ceroid lipofuscinosis, neuronal, type 2; Spinocerebellar ataxia, autosomal recessive, type 7	Autosomal recessive
TPRN	613354	9	613307	Deafness, autosomal recessive, type 79	Autosomal recessive
TRAF3IP1	607380	2	616629	Senior-Loken syndrome, type 9	Autosomal recessive
TRAIP	605958	3	616777	Seckel syndrome, type 9	Autosomal recessive
TRAPPC11	614138	4	615356	Limb-girdle muscular dystrophy, type 18 (LGMD R18)	Autosomal recessive
TRAPPC12	614139	2	617669	Encephalopathy, progressive, early-onset, with brain atrophy and spasticity	Autosomal recessive
TRAPPC6B	610397	14	617862	Neurodevelopmental disorder with microcephaly, epilepsy, and brain atrophy	Autosomal recessive
TRAPPC9	611966	8	613192	Mental retardation, autosomal recessive, type 13	Autosomal recessive
TRDN	603283	6	615441	Ventricular tachycardia, catecholaminergic polymorphic, type 5, with or without muscle weakness	Autosomal recessive
TREM2	605086	6	221770	Nasu-Hakola disease	Autosomal recessive
TREX1	606609	3	225750	Aicardi-Goutieres syndrome, type 1	Autosomal recessive
TRHR	188545	8	618573	Hypothyroidism, congenital, nongoitrous, type 7	Autosomal recessive
TRIM2	614141	4	615490	Charcot-Marie-Tooth disease, type 2R	Autosomal recessive
TRIM32	602290	9	254110	Limb-girdle muscular dystrophy, type 8 (LGMD R8)	Autosomal recessive
TRIM37	605073	17	253250	Mulibrey nanism	Autosomal recessive
TRIOBP	609761	22	609823	Deafness, autosomal recessive, type 28	Autosomal recessive
TRIP11	604505	14	200600	Achondrogenesis, type 1A	Autosomal recessive
TRIP13	604507	5	617598	Mosaic variegated aneuploidy syndrome 3	Autosomal recessive
TRIP4	604501	15	616866	Spinal muscular atrophy with congenital bone fractures 1	Autosomal recessive
TRIT1	617840	1	617873	Combined oxidative phosphorylation deficiency 35	Autosomal recessive
TRMT10A	616013	4	616033	Microcephaly, short stature, and impaired glucose metabolism 1	Autosomal recessive
TRMT10C	615423	3	616974	Combined oxidative phosphorylation deficiency 30	Autosomal recessive
TRMT5	611023	14	616539	Combined oxidative phosphorylation deficiency 26	Autosomal recessive
TRMU	610230	22	613070	Liver failure, transient infantile	Autosomal recessive
TRNT1	612907	3	616959	Retinitis pigmentosa and erythrocytic microcytosis	Autosomal recessive
TRPM1	603576	15	613216	Night blindness, congenital stationary (complete), type 1C, autosomal recessive	Autosomal recessive
TRPM6	607009	9	602014	Familial hypomagnesemia with secondary hypocalcemia	Autosomal recessive
TRPV6	606680	7	618188	Hyperparathyroidism, transient neonatal	Autosomal recessive
TSEN15	608756	1	617026	Pontocerebellar hypoplasia, type 2F	Autosomal recessive
TSEN2	608753	3	612389	Pontocerebellar hypoplasia, type 2B	Autosomal recessive

TSEN34	608754	19		612390	Pontocerebellar hypoplasia type 2C	Autosomal recessive
TSEN54	608755	17		277470; 225753	Pontocerebellar hypoplasia, type 2A; Pontocerebellar hypoplasia, type 4	Autosomal recessive
TSFM	604723	12		610505	Combined oxidative phosphorylation deficiency, type 3	Autosomal recessive
TSHB	188540	1		275100	Hypothyroidism, congenital, nongoitrous, type 4	Autosomal recessive
TSHR	603372	14		275200	Hypothyroidism, congenital, nongoitrous, type 1	Autosomal recessive
TTC19	613814	17		615157	Mitochondrial complex III deficiency, nuclear type 2	Autosomal recessive
TTC21B	612014	2		613819	Short-rib thoracic dysplasia, type 4, with or without polydactyly	Autosomal recessive
TTC37	614589	5		222470	Trichohepatoenteric syndrome, type 1 (diarrhea, syndromic)	Autosomal recessive
TTC7A	609332	2		243150	Gastrointestinal defects and immunodeficiency syndrome	Autosomal recessive
TTC8	608132	14		615985	Bardet-Biedl syndrome, type 8	Autosomal recessive
TTI2	614426	8		615541	Mental retardation, autosomal recessive, type 39	Autosomal recessive
TTLL5	612268	14		615860	Cone-rod dystrophy 19	Autosomal recessive
TTN	188840	2		608807; 611705	Limb-girdle muscular dystrophy type 10 (LGMDR10); Early-onset myopathy with fatal cardiomyopathy (Salih myopathy)	Autosomal recessive
TTPA	600415	8		277460	Ataxia with isolated vitamin E deficiency	Autosomal recessive
TUBA8	605742	22		613180	Cortical dysplasia, complex, with other brain malformations, type 8	Autosomal recessive
TUBGCP4	609610	15		616335	Microcephaly and chorioretinopathy, autosomal recessive, type 3	Autosomal recessive
TUBGCP6	610053	22		251270	Microcephaly and chorioretinopathy, autosomal recessive, type 1	Autosomal recessive
TUFM	602389	16		610678	Combined oxidative phosphorylation deficiency 4	Autosomal recessive
TULP1	602280	6		613843	Leber congenital amaurosis, type 15	Autosomal recessive
TUSC3	601385	8		611093	Mental retardation, autosomal recessive, type 7	Autosomal recessive
TWIST2	607556	2		227260	Focal facial dermal dysplasia, type 3 (Setleis type)	Autosomal recessive
TWNK	606075	10	C10orf2	271245; 616138	Mitochondrial DNA depletion syndrome, type 7 (hepatocerebral type); Perrault syndrome type 5	Autosomal recessive
TXNL4A	611595	18		608572	Burn-McKeown syndrome	Autosomal recessive
TYK2	176941	19		611521	Immunodeficiency, type 35	Autosomal recessive
TYMP	131222	22		603041	Mitochondrial DNA depletion syndrome, type 1 (MNGIE type)	Autosomal recessive
TYR	606933	11		203100; 606952	Oculocutaneous albinism (OCA) type 1A; OCA type 1B	Autosomal recessive
TYROBP	604142	19		221770	Polycystic lipomembranous osteodysplasia with sclerosing leukoencephalopathy, type 1 (Nasu-Hakola disease)	Autosomal recessive
TYRP1	115501	9		203290	Albinism, oculocutaneous, type 3	Autosomal recessive
UBA5	610552	3		617132	Epileptic encephalopathy, early infantile, 44	Autosomal recessive
UBE2T	610538	1		616435	Fanconi anemia, complementation group T	Autosomal recessive
UBE3A	601623	15		105830	Angelman syndrome	Autosomal dominant
UBE3B	608047	12		244450	Kaufman oculocerebrofacial syndrome	Autosomal recessive
UBR1	605981	15		243800	Johanson-Blizzard syndrome	Autosomal recessive
UCHL1	191342	4		615491	Spastic paraplegia, type 79, autosomal recessive	Autosomal recessive
UFM1	610553	13		617899	Leukodystrophy, hypomyelinating, type 14	Autosomal recessive
UGT1A1	191740	2		606785; 218800	Crigler-Najjar syndrome, type 1; Crigler-Najjar syndrome, type 2	Autosomal recessive
UMPS	613891	3		258900	Orotic aciduria	Autosomal recessive
UNC13D	608897	17		608898	Hemophagocytic lymphohistiocytosis, familial, type 3	Autosomal recessive
UNC80	612636	2		616801	Hypotonia, infantile, with psychomotor retardation and characteristic facies, type 2	Autosomal recessive
UNG	191525	12		608106	Immunodeficiency with hyper IgM, type 5	Autosomal recessive
UPB1	606673	22		613161	Beta-ureidopropionase deficiency	Autosomal recessive
UPF3B	300298	X		300676	Mental retardation, X-linked, syndromic, type 14	X-linked
UQCRB	191330	8		615158	Mitochondrial complex III deficiency, nuclear, type 3	Autosomal recessive
UQCRC2	191329	16		615160	Mitochondrial complex III deficiency, nuclear type 5	Autosomal recessive
UQCRQ	612080	5		615159	Mitochondrial complex III deficiency, nuclear, type 4	Autosomal recessive
UROD	613521	1		176100	Porphyria cutanea tarda	Autosomal recessive
UROS	606938	10		263700	Porphyria, congenital erythropoietic	Autosomal recessive
USB1	613276	16		604173	Poikiloderma with neutropenia	Autosomal recessive
USH1C	605242	11		276904; 602092	Usher syndrome, type 1C; Deafness, autosomal recessive, type 18A	Autosomal recessive
USH1G	607696	17		606943	Usher syndrome, type 1G	Autosomal recessive
USH2A	608400	1		276901	Usher syndrome, type 2A	Autosomal recessive
USP18	607057	22		617397	Pseudo-TORCH syndrome 2	Autosomal recessive
UVSSA	614632	4		614640	UV-sensitive syndrome, type 3	Autosomal recessive
VAC14	604632	16		617054	Striatonigral degeneration, childhood-onset	Autosomal recessive
VARS1	192150	6	VARS	617802	Neurodevelopmental disorder with microcephaly, seizures, and cortical atrophy	Autosomal recessive

VARS2	612802	6		615917	Combined oxidative phosphorylation deficiency 20	Autosomal recessive
VDR	601769	12		277440	Rickets, vitamin D-resistant, type 2A	Autosomal recessive
VIPAS39	613401	14	VIPAR	613404	Arthrogryposis, renal dysfunction and cholestasis, type 2	Autosomal recessive
VKORC1	608547	16		607473	Vitamin K-dependent clotting factors, combined deficiency of, type 2	Autosomal recessive
VLDLR	192977	9		224050	Cerebellar hypoplasia and mental retardation with or without quadrupedal locomotion, type 1	Autosomal recessive
VPS13A	605978	9		200150	Choreoacanthocytosis	Autosomal recessive
VPS13B	607817	8		216550	Cohen syndrome	Autosomal recessive
VPS13C	608879	15		616840	Parkinson disease 23, autosomal recessive, early onset	Autosomal recessive
VPS33B	608552	15		208085	Arthrogryposis, renal dysfunction and cholestasis, type 1	Autosomal recessive
VPS37A	609927	8		614898	Spastic paraplegia, type 53, autosomal recessive	Autosomal recessive
VPS45	610035	1		615285	Neutropenia, severe congenital, type 5	Autosomal recessive
VPS53	615850	17		615851	Pontocerebellar hypoplasia, type 2E	Autosomal recessive
VRK1	602168	14		607596	Pontocerebellar hypoplasia, type 1A	Autosomal recessive
VSX2	142993	14		610092; 610093	Microphthalmia with coloboma 3; Isolated microphthalmia 2	Autosomal recessive
VWF	613160	12		277480	von Willibrand disease, type 3	Autosomal recessive
WARS2	604733	1		617710	Neurodevelopmental disorder, mitochondrial, with abnormal movements and lactic acidosis, with or without seizures	Autosomal recessive
WAS	300392	X		301000; 313900	Wiskott-Aldrich syndrome; Thrombocytopenia, X-linked	X-linked
WASHC4	615748	12	KIAA1033	615817	?Mental retardation, autosomal recessive, type 43	Autosomal recessive
WASHC5	610657	8	KIAA0196	220210	Ritscher-Schinzel syndrome, type 1	Autosomal recessive
WDR19	608151	4		614377; 616307	Nephronophthisis, type 13; Senior-Loken syndrome, type 8	Autosomal recessive
WDR35	613602	2		613610	Cranioectodermal dysplasia 2	Autosomal recessive
WDR45B	609226	17		617977	Neurodevelopmental disorder with spastic quadriplegia and brain abnormalities with or without seizures	Autosomal recessive
WDR62	613583	19		604317	Microcephaly, type 2, primary, autosomal recessive, with or without cortical malformations	Autosomal recessive
WDR72	613214	15		613211	Amelogenesis imperfecta, type 2A3 (hypomaturation type)	Autosomal recessive
WDR73	616144	15		251300	Galloway-Mowat syndrome 1	Autosomal recessive
WDR81	614218	17		610185	Cerebellar ataxia, mental retardation, and dysequilibrium syndrome, type 2	Autosomal recessive
WEE2	614084	7		617996	Oocyte maturation defect 5	Autosomal recessive
WFS1	606201	4		222300	Wolfram syndrome, type 1	Autosomal recessive
WHRN	607928	9	DFNB31	611383; 607084	Usher syndrome, type 2D; Deafness, autosomal recessive, type 31	Autosomal recessive
WIPF1	602357	2		614493	?Wiskott-Aldrich syndrome 2	Autosomal recessive
WNK1	605232	12		201300	Neuropathy, hereditary sensory and autonomic, type 2	Autosomal recessive
WNT1	164820	12		615220	Osteogenesis imperfecta, type XV	Autosomal recessive
WNT10A	606268	2		257980	Odontoonychodermal dysplasia	Autosomal recessive
WNT10B	601906	12		225300	Split-hand/foot malformation, type 6	Autosomal recessive
WNT3	165330	17		273395	?Tetra-amelia syndrome	Autosomal recessive
WNT7A	601570	3		228930	Fuhrmann syndrome	Autosomal recessive
WRAP53	612661	17		613988	Dyskeratosis congenita, autosomal recessive, type 3	Autosomal recessive
WRN	604611	8		277700	Werner syndrome	Autosomal recessive
WWOX	605131	16		616211; 614322	Epileptic encephalopathy, early infantile, type 28; Spinocerebellar ataxia, autosomal recessive, type 12	Autosomal recessive
XDH	607633	2		278300	Xanthinuria, type 1	Autosomal recessive
XPA	611153	9		278700	Xeroderma pigmentosum, group A	Autosomal recessive
XPC	613208	3		278720	Xeroderma pigmentosum, group C	Autosomal recessive
XPNPEP3	613553	22		613159	Nephronophthisis-like nephropathy, type 1	Autosomal recessive
XRCC4	194363	5		616541	Short stature, microcephaly, and endocrine dysfunction	Autosomal recessive
XYLT1	608124	16		615777	Desbuquois dysplasia, type 2	Autosomal recessive
XYLT2	608125	17		605822	Spondyloocular syndrome	Autosomal recessive
YARS2	610957	12		613561	Myopathy, lactic acidosis, and sideroblastic anemia, type 2	Autosomal recessive
YY1AP1	607860	1		602531	Grange syndrome	Autosomal recessive
ZAP70	176947	2		617006; 269840	Autoimmune disease, multisystem, infantile-onset, type 2; Immunodeficiency, type 48	Autosomal recessive
ZBTB16	176797	11		612447	Skeletal defects, genital hypoplasia, and mental retardation	Autosomal recessive
ZBTB24	614064	6		614069	Immunodeficiency-centromeric instability-facial anomalies syndrome, type 2	Autosomal recessive
ZC3H14	613279	14		617125	Mental retardation, autosomal recessive, type 56	Autosomal recessive
ZDHHC9	300646	X		300799	Mental retardation, X-linked syndromic, Raymond type	X-linked
ZFYVE26	612012	14		270700	Spastic paraplegia, type 15, autosomal recessive	Autosomal recessive
ZMPSTE24	606480	1		608612	Mandibuloacral dysplasia with, type B lipodystrophy	Autosomal recessive

ZMYND10	607070	3	615444	Ciliary dyskinesia, primary, type 22	Autosomal recessive
ZNF408	616454	11	616469	Retinitis pigmentosa, type 72	Autosomal recessive
ZNF423	604557	16	614844	Joubert syndrome, type 19	Autosomal recessive
ZNF469	612078	16	229200	Brittle cornea syndrome, type 1	Autosomal recessive
ZNF711	314990	X	300803	Mental retardation, X-linked, type 97	X-linked
ZNHIT3	604500	17	260565	PEHO syndrome	Autosomal recessive
ZP1	195000	11	615774	Oocyte maturation defect, type 1	Autosomal recessive