Igenomix
PART OF RITHOLIS GROUP

CGT Exome v5.4.5

Chromosome	Gene symbol	OMIM (gene)	Disease name (phenotype)	Inheritance
12	AAAS	605378	Triple-A syndrome (achalasia-addisonianism-alacrimia)	Autosomal recessive
16	AARS1	601065	Epileptic encephalopathy, early infantile, type 29	Autosomal recessive
6	AARS2	612035	Combined oxidative phosphorylation deficiency 8; Leukoencephalopathy, progressive, with ovarian failure	Autosomal recessive
7	AASS	605113	Hyperlysinemia, type 1 and type 2	Autosomal recessive
16	ABAT	137150	GABA-transaminase deficiency	Autosomal recessive
9	ABCA1	600046	Tangier disease	Autosomal recessive
2	ABCA12	607800	Ichthyosis, congenital, autosomal recessive, type 4A; ICAR, type 4B (harlequin)	Autosomal recessive
16	ABCA3	601615	Surfactant metabolism dysfunction, pulmonary, type 3	Autosomal recessive
1	ABCA4	601691	Stargardt disease 1; Retinitis pigmentosa 19; Cone-rod dystrophy 3	Autosomal recessive
2	ABCB11	603201	Cholestasis, benign recurrent intrahepatic, type 2; Cholestasis, progressive familial intrahepatic, type 2	Autosomal recessive
7	ABCB4	171060	Cholestasis, progressive familial intrahepatic, type 3	Autosomal recessive
10	ABCC2	601107	Dubin-Johnson syndrome	Autosomal recessive
16	ABCC6	603234	Pseudoxanthoma elasticum; Generalized arterial calcification of infancy, type 2	Autosomal recessive
11	ABCC8	600509	Hyperinsulinemic hypoglycemia, type 1 (congenital hyperinsulinism); Permanent neonatal diabetes mellitus (PNDM)	Autosomal recessive*
X	ABCD1	300371	Adrenoleukodystrophy	X-linked
14	ABCD4	603214	Methylmalonic aciduria and homocystinuria, cblJ type	Autosomal recessive
2	ABCG5	605459	Sitosterolemia 2	Autosomal recessive
2	ABCG8	605460	Sitosterolemia 1	Autosomal recessive
20	ABHD12	613599	PHARC syndrome (polyneuropathy, hearing loss, ataxia, retinitis pigmentosa and cataract)	Autosomal recessive
3	ABHD5	604780	Chanarin-Dorfman syndrome	Autosomal recessive
11	ACAD8	604773	Isobutyryl-CoA dehydrogenase deficiency	Autosomal recessive
3	ACAD9	611103	Acyl-CoA dehydrogenase 9 deficiency (mitochondrial complex I deficiency, nuclear, type 20)	Autosomal recessive
1	ACADM	607008	Medium-chain acyl-CoA dehydrogenase deficiency	Autosomal recessive
12	ACADS	606885	Short-chain acyl-CoA dehydrogenase deficiency	Autosomal recessive
10	ACADSB	600301	Short/branched-chain acyl-CoA dehydrogenase deficiency	Autosomal recessive
17	ACADVL	609575	Very long-chain acyl-CoA dehydrogenase (VLCAD) deficiency	Autosomal recessive
11	ACAT1	607809	Alpha-methylacetoacetic aciduria (3-ketothiolase deficiency)	Autosomal recessive
17	ACE	106180	Renal tubular dysgenesis	Autosomal recessive
22	ACO2	100850	Infantile cerebellar-retinal degeneration	Autosomal recessive
17	ACOX1	609751	Peroxisomal acyl-CoA oxidase deficiency	Autosomal recessive
3	ACOX2	601641	Bile acid synthesis defect, congenital, type 6	Autosomal recessive
19	ACP5	171640	Spondyloenchondrodysplasia with immune dysregulation	Autosomal recessive
16	ACSF3	614245	Combined malonic and methylmalonic aciduria	Autosomal recessive
1	ACTA1	102610	Nemaline myopathy 3; Congenital fiber-type disproportion myopathy 1	Autosomal recessive*
3	ACY1	104620	Aminoacylase 1 deficiency	Autosomal recessive
20	ADA	608958	Severe combined immunodeficiency due to adenosine deaminase deficiency (ADA)	Autosomal recessive
22	ADA2	607575	Vasculitis, autoinflammation, immunodeficiency, and hematologic defects syndrome; Sneddon syndrome	Autosomal recessive
8	ADAM9	602713	Cone-rod dystrophy 9	Autosomal recessive
19	ADAMTS10	608990	Weill-Marchesani syndrome, type 1, recessive	Autosomal recessive
9	ADAMTS13	604134	Thrombotic thrombocytopenic purpura, familial (Schulman-Upshaw syndrome)	Autosomal recessive
15	ADAMTS17	607511	Weill-Marchesani syndrome, type 4, recessive	Autosomal recessive
16	ADAMTS18	607512	Microcornea, myopic chorioretinal atrophy, and telecanthus	Autosomal recessive
5	ADAMTS2	604539	Ehlers-Danlos syndrome, dermatosparaxis type	Autosomal recessive
9	ADAMTSL2	612277	Geleophysic dysplasia type 1	Autosomal recessive
1	ADAMTSL4	610113	Ectopia lentis et pupillae; Ectopia lentis, isolated, type 2	Autosomal recessive
1	ADAR	146920	Aicardi-Goutieres syndrome, type 6	Autosomal recessive

19	ADAT3	615302	Mental retardation, autosomal recessive 36	Autosomal recessive
16	ADGRG1	604110	Polymicrogyria, bilateral frontoparietal	Autosomal recessive
6	ADGRG6	612243	Lethal congenital contracture syndrome 9	Autosomal recessive
5	ADGRV1	602851	Usher syndrome, type 2C	Autosomal recessive; Digenic inheritance (PDZD7 gene)
10	ADK	102750	Hypermethioninemia due to adenosine kinase deficiency	Autosomal recessive
22	ADSL	608222	Adenylosuccinase deficiency	Autosomal recessive
14	ADSS1	612498	Myopathy, distal, 5	Autosomal recessive
18	AFG3L2	604581	Spastic ataxia, type 5, autosomal recessive	Autosomal recessive
4	AFP	104150	Alpha-fetoprotein deficiency	Autosomal recessive
4	AGA	613228	Aspartylglucosaminuria (glycosylasparaginase deficiency)	Autosomal recessive
2	AGBL5	615900	Retinitis pigmentosa 75	Autosomal recessive
7	AGK	610345	Cataract 38; Sengers syndrome	Autosomal recessive
1	AGL	610860	Glycogen storage disease, type 3	Autosomal recessive
9	AGPAT2	603100	Congenital generalized lipodystrophy (Berardinelli-Seip syndrome)	Autosomal recessive
2	AGPS	603051	Rhizomelic chondrodysplasia punctata, type 3	Autosomal recessive
1	AGRN	103320	Myasthenic syndrome, congenital, type 8	Autosomal recessive
1	AGT	106150	Renal tubular dysgenesis	Autosomal recessive
3	AGTR1	106165	Renal tubular dysgenesis	Autosomal recessive
2	AGXT	604285	Hyperoxaluria, primary, type 1	Autosomal recessive
20	AHCY	180960	Hypermethioninemia with deficiency of S-adenosylhomocysteine hydrolase	Autosomal recessive
6	AHI1	608894	Joubert syndrome, type 3	Autosomal recessive
12	AICDA	605257	Immunodeficiency with hyper-IgM, type 2	Autosomal recessive
4	AIMP1	603605	Leukodystrophy, hypomyelinating, type 3	Autosomal recessive
7	AIMP2	600859	Leukodystrophy, hypomyelinating, type 17	Autosomal recessive
17	AIPL1	604392	Leber congenital amaurosis, type 4	Autosomal recessive
21	AIRE	607358	Autoimmune polyendocrinopathy syndrome, type 1	Autosomal recessive*
9	AK1	103000	Hemolytic anemia due to adenylate kinase deficiency	Autosomal recessive
1	AK2	103020	Reticular dysgenesis	Autosomal recessive
10	AKR1C2	600450	46,XY disorder of sex development due to testicular 17,20-desmolase deficiency	Autosomal recessive
7	AKR1D1	604741	Bile acid synthesis defect, congenital, type 2	Autosomal recessive
9	ALAD	125270	Porphyria, acute hepatic	Autosomal recessive
4	ALB	103600	Analbuminemia	Autosomal recessive
10	ALDH18A1	138250	Spastic paraplegia, type 9B, autosomal recessive; Cutis laxa, type 3A (De Barsy syndrome)	Autosomal recessive
15	ALDH1A3	600463	Microphthalmia, isolated 8	Autosomal recessive
17	ALDH3A2	609523	Sjogren-Larsson syndrome	Autosomal recessive
1	ALDH4A1	606811	Hyperprolinemia, type 2	Autosomal recessive
6	ALDH5A1	610045	Succinic semialdehyde dehydrogenase deficiency	Autosomal recessive
14	ALDH6A1	603178	Methylmalonate semialdehyde dehydrogenase deficiency	Autosomal recessive
5	ALDH7A1	107323	Epilepsy, pyridoxine-dependent	Autosomal recessive
16	ALDOA	103850	Glycogen storage disease type 12	Autosomal recessive
9	ALDOB	612724	Fructose intolerance, hereditary	Autosomal recessive
16	ALG1	605907	Congenital disorder of glycosylation, type 1K	Autosomal recessive
13	ALG11	613666	Congenital disorder of glycosylation, type 1P	Autosomal recessive
22	ALG12	607144	Congenital disorder of glycosylation, type 1G	Autosomal recessive
9	ALG2	607905	Myasthenic syndrome, congenital, type 14, with tubular aggregates	Autosomal recessive
3	ALG3	608750	Congenital disorder of glycosylation, type 1D	Autosomal recessive
1	ALG6	604566	Congenital disorder of glycosylation, type 1C	Autosomal recessive
11	ALG8	608103	Congenital disorder of glycosylation, type 1H	Autosomal recessive
			Congenital disorder of glycosylation, type 1L; Gillessen-Kaesbach-Nishimura	

11	ALG9	606941	Congenital disorder of glycosylation, type 1L, Ollessen-Kjaesbach-Nishimura syndrome	Autosomal recessive
2	ALMS1	606844	Alström syndrome	Autosomal recessive
17	ALOX12B	603741	Ichthyosis, congenital, autosomal recessive, type 2	Autosomal recessive
17	ALOXE3	607206	Ichthyosis, congenital, autosomal recessive, type 3	Autosomal recessive
15	ALPK3	617608	Cardiomyopathy, familial hypertrophic, type 27	Autosomal recessive
1	ALPL	171760	ALPL-related conditions	Autosomal recessive
2	ALS2	606352	Amyotrophic lateral sclerosis, type 2, juvenile; Primary lateral sclerosis, juvenile; Spastic paralysis, infantile onset ascending	Autosomal recessive
12	ALX1	601527	Frontonasal dysplasia, type 3	Autosomal recessive
1	ALX3	606014	Frontonasal dysplasia, type 1	Autosomal recessive
11	ALX4	605420	Frontonasal dysplasia, type 2	Autosomal recessive
5	AMACR	604489	Bile acid synthesis defect, congenital, type 4; Alpha-methylacyl-CoA racemase deficiency	Autosomal recessive
4	AMBN	601259	Amelogenesis imperfecta, type IF	Autosomal recessive
19	AMH	600957	Persistent Mullerian duct syndrome, type 1	Autosomal recessive
12	AMHR2	600956	Persistent Mullerian duct syndrome, type II	Autosomal recessive
14	AMN	605799	Megaloblastic anemia 1 (Imerslund-Grasbeck syndrome)	Autosomal recessive
1	AMPD1	102770	Myopathy due to myoadenylate deaminase deficiency	Autosomal recessive
1	AMPD2	102771	Pontocerebellar hypoplasia, type 9	Autosomal recessive
3	AMT	238310	Glycine encephalopathy	Autosomal recessive
1	ANGPTL3	604774	Hypobetalipoproteinemia, familial, type 2	Autosomal recessive
9	ANKS6	615370	Nephronophthisis 16	Autosomal recessive
3	ANO10	613726	Spinocerebellar ataxia, autosomal recessive, type 10	Autosomal recessive
11	ANO5	608662	Limb-girdle muscular dystrophy, type 12 (LGMD R12)	Autosomal recessive
2	ANTXR1	606410	GAPO syndrome	Autosomal recessive
4	ANTXR2	608041	Hyaline fibromatosis syndrome	Autosomal recessive
7	AP1S1	603531	MEDNIK syndrome	Autosomal recessive
X	AP1S2	300629	Mental retardation, X-linked, syndromic, type 5 (Pettigrew syndrome)	X-linked
5	AP3B1	603401	Hermansky-Pudlak syndrome, type 2	Autosomal recessive
15	AP3B2	602166	Epileptic encephalopathy, early infantile, type 48	Autosomal recessive
19	AP3D1	607246	Hermansky-Pudlak syndrome, type 10	Autosomal recessive
1	AP4B1	607245	Spastic paraplegia, type 47, autosomal recessive	Autosomal recessive
15	AP4E1	607244	Spastic paraplegia, type 51, autosomal recessive	Autosomal recessive
7	AP4M1	602296	Spastic paraplegia, type 50, autosomal recessive	Autosomal recessive
14	AP4S1	607243	Spastic paraplegia, type 52, autosomal recessive	Autosomal recessive
7	AP5Z1	613653	Spastic paraplegia, type 48, autosomal recessive	Autosomal recessive
19	APOC2	608083	Hyperlipoproteinemia, type 1B	Autosomal recessive
19	APOE	107741	Sea-blue histiocyte disease	Autosomal recessive
16	APRT	102600	Adenine phosphoribosyltransferase deficiency	Autosomal recessive
9	APTX	606350	Ataxia, early-onset, with oculomotor apraxia and hypoalbuminemia	Autosomal recessive
12	AQP2	107777	Diabetes insipidus, nephrogenic, type 2	Autosomal recessive*
X	AR	313700	Androgen insensitivity syndrome	X-linked
20	ARFGEF2	605371	Periventricular heterotopia with microcephaly	Autosomal recessive
6	ARG1	608313	Argininemia (arginase deficiency)	Autosomal recessive
17	ARHGDIA	601925	Nephrotic syndrome, type 8	Autosomal recessive
19	ARHGEF18	616432	Retinitis pigmentosa 78	Autosomal recessive
3	ARL13B	608922	Joubert syndrome type 8	Autosomal recessive
16	ARL2BP	615407	Retinitis pigmentosa with or without situs inversus	Autosomal recessive
3	ARL6	608845	Bardet-Biedl syndrome, type 3	Autosomal recessive
2	ARMC9	617612	Joubert syndrome 30	Autosomal recessive
7	ARPC1B	604223	Immunodeficiency, type 71, with inflammatory disease and congenital thrombocytopenia	Autosomal recessive

22	ARSA	607574	Metachromatic leukodystrophy	Autosomal recessive
5	ARSB	611542	Mucopolysaccharidosis, type 6 (Maroteaux-Lamy syndrome)	Autosomal recessive
X	ARSL	300180	Chondrodysplasia punctata, brachytelephalangic	X-linked
1	ARV1	611647	Epileptic encephalopathy, early infantile, 38	Autosomal recessive
X	ARX	300382	Epileptic encephalopathy, early infantile, type 1; ARX-related developmental disorders	X-linked
8	ASAH1	613468	Farber lipogranulomatosis; Spinal muscular atrophy with progressive myoclonic epilepsy	Autosomal recessive
7	ASL	608310	Argininosuccinic aciduria	Autosomal recessive
7	ASNS	108370	Asparagine synthetase deficiency	Autosomal recessive
17	ASPA	608034	Canavan disease	Autosomal recessive
8	ASPH	600582	Traboulsi syndrome	Autosomal recessive
1	ASPM	605481	Primary microcephaly type 5, autosomal recessive	Autosomal recessive
9	ASS1	603470	Citrullinemia, type 1	Autosomal recessive
10	ATAD1	614452	Hyperekplexia 4	Autosomal recessive
1	ATF6	605537	Achromatopsia, type 7	Autosomal recessive
2	ATIC	601731	AICA-ribosiduria due to ATIC deficiency	Autosomal recessive
11	ATM	607585	ATM-related conditions	Autosomal recessive
10	ATOH7	609875	Persistent hyperplastic primary vitreous, autosomal recessive	Autosomal recessive
1	ATP13A2	610513	Kufor-Rakeb syndrome; Spastic paraplegia, type 78, autosomal recessive	Autosomal recessive
16	ATP2A1	108730	Brody myopathy	Autosomal recessive
12	ATP6V0A2	611716	Cutis laxa, autosomal recessive, type 2A; Wrinkly skin syndrome	Autosomal recessive
7	ATP6V0A4	605239	Renal tubular acidosis, distal, autosomal recessive	Autosomal recessive
3	ATP6V1A	607027	Cutis laxa, autosomal recessive, type 2D	Autosomal recessive
2	ATP6V1B1	192132	Renal tubular acidosis with deafness	Autosomal recessive
22	ATP6V1E1	108746	Cutis laxa, autosomal recessive, type 2C	Autosomal recessive
X	ATP7A	300011	Menkes disease; Occipital horn syndrome	X-linked
13	ATP7B	606882	Wilson disease	Autosomal recessive
18	ATP8B1	602397	Cholestasis, progressive familial intrahepatic, type 1; Cholestasis, benign recurrent intrahepatic, type 1	Autosomal recessive
3	ATR	601215	Seckel syndrome, type 1	Autosomal recessive
X	ATRX	300032	Mental retardation-hypotonic facies syndrome, X-linked; Alpha-thalassemia/mental retardation syndrome	X-linked
9	AUH	600529	3-methylglutaconic aciduria, type 1	Autosomal recessive
19	AURKC	603495	Spermatogenic failure, type 5	Autosomal recessive
12	AVIL	613397	Nephrotic syndrome, type 21	Autosomal recessive
15	B2M	109700	Immunodeficiency, type 43	Autosomal recessive
1	B3GALNT2	610194	Muscular dystrophy-dystroglycanopathy (congenital with brain and eye anomalies, type A, 11	Autosomal recessive
1	B3GALT6	615291	Ehlers-Danlos syndrome, spondylodysplastic type, 2	Autosomal recessive
11	B3GAT3	606374	Multiple joint dislocations, short stature, craniofacial dysmorphism, with or without congenital heart defects	Autosomal recessive
13	B3GLCT	610308	Peters-plus syndrome	Autosomal recessive
12	B4GALNT1	601873	Spastic paraplegia, type 26, autosomal recessive	Autosomal recessive
9	B4GALT1	137060	Congenital disorder of glycosylation, type 2D	Autosomal recessive
5	B4GALT7	604327	Ehlers-Danlos syndrome, spondylodysplastic, type 1	Autosomal recessive
11	B4GAT1	605517	Muscular dystrophy-dystroglycanopathy (congenital with brain and eye anomalies), type A, 13	Autosomal recessive
17	B9D1	614144	Joubert syndrome, type 27; ?Meckel syndrome 9	Autosomal recessive
19	B9D2	611951	Joubert syndrome, type 34; ?Meckel syndrome, type 10	Autosomal recessive
11	BBS1	209901	Bardet-Biedl syndrome, type 1	Autosomal recessive
12	BBS10	610148	Bardet-Biedl syndrome, type 10	Autosomal recessive
4	BBS12	610683	Bardet-Biedl syndrome, type 12	Autosomal recessive
16	BBS2	606151	Bardet-Biedl syndrome, type 2	Autosomal recessive
15	BBS4	600374	Bardet-Biedl syndrome, type 4	Autosomal recessive

2	BBS5	603650	Bardet-Biedl syndrome, type 5	Autosomal recessive
4	BBS7	607590	Bardet-Biedl syndrome, type 7	Autosomal recessive
7	BBS9	607968	Bardet-Biedl syndrome, type 9	Autosomal recessive
19	BCAT2	113530	?Hypervalinemia or hyperleucine-isoleucinemia	Autosomal recessive
3	BCHE	177400	Butyrylcholinesterase deficiency	Autosomal recessive
19	BCKDHA	608348	Maple syrup urine disease, type 1A	Autosomal recessive
6	BCKDHB	248611	Maple syrup urine disease, type 1B	Autosomal recessive
16	BCKDK	614901	Branched-chain ketoacid dehydrogenase kinase deficiency	Autosomal recessive
1	BCL10	603517	?Immunodeficiency, type 37	Autosomal recessive
2	BCS1L	603647	Mitochondrial complex III deficiency nuclear type 1; GRACILE syndrome; Bjornstad syndrome	Autosomal recessive
11	BEST1	607854	Bestrophinopathy, AR	Autosomal recessive
20	BFSP1	603307	Cataract 33, multiple types	Autosomal recessive*
17	BHLHA9	615416	Syndactyly, mesoaxial synostotic, with phalangeal reduction	Autosomal recessive
2	BIN1	601248	Centronuclear myopathy, type 2	Autosomal recessive
15	BLM	604610	Bloom syndrome	Autosomal recessive
10	BLNK	604515	?Agammaglobulinemia 4	Autosomal recessive
19	BLOC1S3	609762	Hermansky-Pudlak syndrome, type 8	Autosomal recessive
15	BLOC1S6	604310	?Hermansky-Pudlak syndrome, type 9	Autosomal recessive
4	BLTP1	611565	Alkuraya-Kucinskas syndrome	Autosomal recessive
7	BLVRA	109750	Hyperbiliverdinemia	Autosomal recessive*
8	BMP1	112264	Osteogenesis imperfecta, type 13	Autosomal recessive
7	BMPER	608699	Diaphanospondylodysostosis	Autosomal recessive
4	BMPR1B	603248	Acromesomelic dysplasia, Demirhan type	Autosomal recessive
2	BOLA3	613183	Multiple mitochondrial dysfunctions syndrome 2 with hyperglycinemia	Autosomal recessive
7	BPGM	613896	Erythrocytosis due to bisphosphoglycerate mutase deficiency	Autosomal recessive
8	BPNT2	614010	Chondrodysplasia with joint dislocations, GPAPP type	Autosomal recessive
7	BRAT1	614506	Rigidity and multifocal seizure syndrome, lethal neonatal; Neurodevelopmental disorder with cerebellar atrophy and with or without seizures	Autosomal recessive
14	BRF1	604902	Cerebellofaciodental syndrome	Autosomal recessive
17	BRIP1	605882	Fanconi anemia, complementation group J	Autosomal recessive
X	BRWD3	300553	Mental retardation, X-linked, type 93	X-linked
11	BSCL2	606158	Congenital generalized lipodystrophy, type 2; Encephalopathy, progressive, with or without lipodystrophy	Autosomal recessive
1	BSND	606412	Bartter syndrome, type 4A	Autosomal recessive
3	BTD	609019	Biotinidase deficiency	Autosomal recessive
X	BTK	300300	Agammaglobulinemia X-linked, type 1	X-linked
15	BUB1B	602860	Mosaic variegated aneuploidy syndrome 1	Autosomal recessive
12	C12orf57	615140	Temtamy syndrome	Autosomal recessive
19	C19orf12	614297	Neurodegeneration with brain iron accumulation, type 4	Autosomal recessive*
1	C1QA	120550	C1q deficiency	Autosomal recessive
1	C1QB	120570	C1q deficiency	Autosomal recessive
17	C1QBP	601269	Combined oxidative phosphorylation deficiency 33	Autosomal recessive
1	C1QC	120575	C1q deficiency	Autosomal recessive
12	C1S	120580	C1s deficiency	Autosomal recessive
6	C2	613927	C2 deficiency	Autosomal recessive
11	C2CD3	615944	Orofaciodigital syndrome, type 14	Autosomal recessive
19	C3	120700	Complement component 3 deficiency	Autosomal recessive
9	C5	120900	Complement component 5 deficiency	Autosomal recessive
5	C6	217050	Complement component 6 deficiency	Autosomal recessive
5	C7	217070	Complement component 7 deficiency	Autosomal recessive
1	C8B	120960	Complement component 8 deficiency, type 2	Autosomal recessive

15	CA12	603263	Hyperchlorhidrosis, isolated	Autosomal recessive
8	CA2	611492	Osteopetrosis with renal tubular acidosis (osteopetrosis, autosomal recessive, type 3)	Autosomal recessive
16	CA5A	114761	Hyperammonemia due to carbonic anhydrase VA deficiency	Autosomal recessive
8	CA8	114815	Cerebellar ataxia and mental retardation with or without quadrupedal locomotion 3	Autosomal recessive
11	CABP2	607314	Deafness, autosomal recessive, type 93	Autosomal recessive
11	CABP4	608965	Congenital stationary night blindness, type 2B	Autosomal recessive
3	CACNA1D	114206	Sinoatrial node dysfunction and deafness	Autosomal recessive
12	CACNA2D4	608171	Retinal cone dystrophy 4	Autosomal recessive
2	CAD	114010	Epileptic encephalopathy, early infantile, 50	Autosomal recessive
2	CALCRL	114190	?Lymphatic malformation 8	Autosomal recessive
17	CANT1	613165	Desbuquois dysplasia, type 1; Epiphyseal dysplasia, multiple, type 7	Autosomal recessive
11	CAPN1	114220	Spastic paraplegia, type 76, autosomal recessive	Autosomal recessive
15	CAPN3	114240	Limb-girdle muscular dystrophy, type 1 (LGMD R1)	Autosomal recessive
7	CARD11	607210	Immunodeficiency, type 11A	Autosomal recessive
9	CARD9	607212	Candidiasis, familial, type 2, autosomal recessive	Autosomal recessive
13	CARS2	612800	Combined oxidative phosphorylation deficiency 27	Autosomal recessive
1	CASQ2	114251	Ventricular tachycardia, catecholaminergic polymorphic, type 2	Autosomal recessive
3	CASR	601199	Hyperparathyroidism, neonatal	Autosomal recessive*
5	CAST	114090	Peeling skin with leukonychia, acral punctate keratoses, cheilitis, and knuckle pads	Autosomal recessive
11	CAT	115500	Acatalasemia	Autosomal recessive
11	CATSPER1	606389	Spermatogenic failure, type 7	Autosomal recessive
17	CAVIN1	603198	Lipodystrophy, congenital generalized, type 4	Autosomal recessive
11	CBLIF	609342	Intrinsic factor deficiency	Autosomal recessive
21	CBS	613381	Homocystinuria due to cystathionine beta-synthase	Autosomal recessive
19	CC2D1A	610055	Mental retardation, autosomal recessive, type 3	Autosomal recessive
4	CC2D2A	612013	Joubert syndrome, type 9; Meckel syndrome, type 6; COACH syndrome, 2	Autosomal recessive
18	CCBE1	612753	Hennekam lymphangiectasia-lymphedema syndrome, type 1	Autosomal recessive
17	CCDC103	614677	Ciliary dyskinesia, primary, type 17	Autosomal recessive
2	CCDC115	613734	Congenital disorder of glycosylation, type IIo	Autosomal recessive
3	CCDC174	616735	Hypotonia, infantile, with psychomotor retardation	Autosomal recessive
3	CCDC39	613798	Ciliary dyskinesia, primary, type 14	Autosomal recessive
17	CCDC40	613799	Ciliary dyskinesia, primary, type 15	Autosomal recessive
12	CCDC65	611088	Ciliary dyskinesia, primary, type 27	Autosomal recessive
19	CCDC8	614145	3M syndrome 3	Autosomal recessive
14	CCDC88C	611204	Hydrocephalus, congenital, type 1	Autosomal recessive
6	CCN6	603400	Progressive pseudorheumatoid dysplasia	Autosomal recessive
5	CCNO	607752	Ciliary dyskinesia, primary, type 29	Autosomal recessive
16	CD19	107265	Immunodeficiency, common variable, type 3	Autosomal recessive
1	CD247	186780	?Immunodeficiency, type 25	Autosomal recessive
12	CD27	186711	Lymphoproliferative syndrome 2	Autosomal recessive
6	CD2AP	604241	Glomerulosclerosis, focal segmental, type 3, susceptibility to	Autosomal recessive*
19	CD320	606475	Methylmalonic aciduria, transient, due to transcobalamin receptor defect	Autosomal recessive
7	CD36	173510	Platelet glycoprotein 4 deficiency	Autosomal recessive
11	CD3D	186790	Immunodeficiency, type 19	Autosomal recessive
11	CD3E	186830	Immunodeficiency, type 18	Autosomal recessive
11	CD3G	186740	Immunodeficiency, type 17, CD3 gamma deficient	Autosomal recessive
20	CD40	109535	Immunodeficiency with hyper-IgM, type 3	Autosomal recessive
X	CD40LG	300386	Hyper-IgM syndrome, type 1 (immunodeficiency, X-linked, with hyper-IgM, type 1)	X-linked
1	CD55	125240	Complement hyperactivation, angiopathic thrombosis, and protein-losing	Autosomal recessive

1	CD55	125240	enteropathy (CHAPLE)	Autosomal recessive
11	CD59	107271	CD59 deficiency	Autosomal recessive
19	CD79A	112205	Agammaglobulinemia 3	Autosomal recessive
17	CD79B	147245	Agammaglobulinemia 6	Autosomal recessive
11	CD81	186845	Immunodeficiency, common variable, type 6	Autosomal recessive
2	CD8A	186910	CD8 deficiency, familial	Autosomal recessive
15	CDAN1	607465	Dyserythropoietic anemia, congenital, type 1A	Autosomal recessive
1	CDC14A	603504	Deafness, autosomal recessive, type 105	Autosomal recessive
22	CDC45	603465	Meier-Gorlin syndrome 7	Autosomal recessive
2	CDCA7	609937	Immunodeficiency-centromeric instability-facial anomalies syndrome 3	Autosomal recessive
16	CDH11	600023	Elsahy-Waters syndrome	Autosomal recessive
10	CDH23	605516	Deafness, autosomal recessive, type 12; Usher syndrome, type 1D	Autosomal recessive
16	CDH3	114021	Ectodermal dysplasia, ectrodactyly, and macular dystrophy; Hypotrichosis, congenital, with juvenile macular dystrophy	Autosomal recessive
10	CDHR1	609502	Cone-rod dystrophy, type 15	Autosomal recessive
15	CDIN1	615626	Dyserythropoietic anemia, congenital, type Ib	Autosomal recessive
16	CDK10	603464	Al Kaissi syndrome	Autosomal recessive
9	CDK5RAP2	608201	Primary microcephaly type 3, autosomal recessive	Autosomal recessive
6	CDSN	602593	Peeling skin syndrome 1	Autosomal recessive
16	CDT1	605525	Meier-Gorlin syndrome, type 4	Autosomal recessive
14	CEBPE	600749	Specific granule deficiency	Autosomal recessive
1	CENPF	600236	Stromme syndrome	Autosomal recessive
13	CENPJ	609279	Primary microcephaly type 6, autosomal recessive	Autosomal recessive
1	CEP104	616690	Joubert syndrome 25	Autosomal recessive
5	CEP120	613446	Short-rib thoracic dysplasia 13 with or without polydactyly	Autosomal recessive
4	CEP135	611423	Microcephaly 8, primary, autosomal recessive	Autosomal recessive
15	CEP152	613529	Primary microcephaly type 9, autosomal recessive	Autosomal recessive
11	CEP164	614848	Nephronophthisis 15	Autosomal recessive
3	CEP19	615586	Morbid obesity and spermatogenic failure	Autosomal recessive
12	CEP290	610142	Meckel syndrome, type 4; Joubert syndrome, type 5; Leber congenital amaurosis, type 10	Autosomal recessive
7	CEP41	610523	Joubert syndrome, type 15	Autosomal recessive
10	CEP55	610000	Multinucleated neurons, anhydramnios, renal dysplasia, cerebellar hypoplasia, and hydranencephaly	Autosomal recessive
11	CEP57	607951	Mosaic variegated aneuploidy syndrome 2	Autosomal recessive
9	CEP78	617110	Cone-rod dystrophy and hearing loss	Autosomal recessive
12	CEP83	615847	Nephronophthisis 18	Autosomal recessive
2	CERKL	608381	Retinitis pigmentosa, type 26	Autosomal recessive
15	CERS3	615276	Ichthyosis, congenital, autosomal recessive 9	Autosomal recessive
8	CFAP418	614477	Bardet-Biedl syndrome, type 21; Cone-rod dystrophy 16 and Retintis pigmentosa 64; Ciliary dyskinesia, primary, 26	Autosomal recessive
10	CFAP43	617558	Spermatogenic failure, type 19	Autosomal recessive
18	CFAP53	614759	Heterotaxy, visceral, 6, autosomal recessive	Autosomal recessive
19	CFD	134350	Complement factor D deficiency	Autosomal recessive
1	CFH	134370	Complement factor H deficiency	Autosomal recessive
4	CFI	217030	Complement factor I deficiency	Autosomal recessive
14	CFL2	601443	Nemaline myopathy, type 7, autosomal recessive	Autosomal recessive
7	CFTR	602421	Cystic fibrosis	Autosomal recessive
10	CHAT	118490	Myasthenic syndrome, congenital, type 6, presynaptic	Autosomal recessive
22	CHKB	612395	Muscular dystrophy, congenital, megaconial type	Autosomal recessive
X	CHM	300390	Choroideremia	X-linked
16	CHMP1A	164010	Pontocerebellar hypoplasia, type 8	Autosomal recessive
2	CHRNA1	100690	Multiple pterygium syndrome, lethal type	Autosomal recessive

17	CHRNB1	100710	?Myasthenic syndrome, congenital, 2C, associated with acetylcholine receptor deficiency	Autosomal recessive
2	CHRND	100720	Myasthenic syndrome, congenital, type 3B, fast-channel; Multiple pterygium syndrome, lethal type	Autosomal recessive
17	CHRNE	100725	Myasthenic syndrome, congenital, type 4B, fast-channel; Myasthenic syndrome, congenital, type 4C, associated with acetylcholine receptor deficiency	Autosomal recessive
2	CHRNG	100730	Multiple pterygium syndrome (MPS), Escobar type; MPS, lethal type	Autosomal recessive
15	CHST14	608429	Ehlers-Danlos syndrome, musculocontractural, type 1	Autosomal recessive
10	CHST3	603799	Spondyloepiphyseal dysplasia with congenital joint dislocations	Autosomal recessive
16	CHST6	605294	Macular corneal dystrophy	Autosomal recessive
15	CHSY1	608183	Temtamy preaxial brachydactyly syndrome	Autosomal recessive
10	CHUK	600664	Cocoon syndrome	Autosomal recessive
15	CIB2	605564	Deafness, autosomal recessive, type 48; Usher syndrome, type 1J	Autosomal recessive
16	CIITA	600005	Bare lymphocyte syndrome, type 2, complementation group A	Autosomal recessive
6	CILK1	612325	Endocrine-cerebroosteodysplasia	Autosomal recessive
4	CISD2	611507	Wolfram syndrome 2	Autosomal recessive
12	CIT	605629	Microcephaly 17, primary, autosomal recessive	Autosomal recessive
2	CKAP2L	616174	Filippi syndrome	Autosomal recessive
11	CLCF1	607672	Cold-induced sweating syndrome 2	Autosomal recessive
7	CLCN1	118425	Myotonia congenita, recessive	Autosomal recessive
3	CLCN2	600570	Leukoencephalopathy with ataxia	Autosomal recessive
16	CLCN7	602727	Osteopetrosis, autosomal recessive type 4	Autosomal recessive
1	CLCNKA	602024	Bartter syndrome, type 4B, digenic	Digenic inheritance (CLCNKB gene)
1	CLCNKB	602023	Bartter syndrome, type 3; Bartter syndrome, type 4B, digenic	Autosomal recessive; Digenic inheritance (CLCNKA gene)
3	CLDN1	603718	Ichthyosis, leukocyte vacuoles, alopecia, and sclerosing cholangitis	Autosomal recessive
13	CLDN10	617579	HELIX syndrome	Autosomal recessive
21	CLDN14	605608	Deafness type 29, autosomal recessive	Autosomal recessive
3	CLDN16	603959	Hypomagnesemia, type 3, renal	Autosomal recessive
1	CLDN19	610036	Rena hypomagnesemia type 5, with ocular involvement	Autosomal recessive
11	CLMP	611693	Congenital short bowel syndrome	Autosomal recessive
16	CLN3	607042	Ceroid lipofuscinosis, neuronal, type 3	Autosomal recessive
13	CLN5	608102	Ceroid lipofuscinosis, neuronal, type 5	Autosomal recessive
15	CLN6	606725	Ceroid lipofuscinosis, neuronal, type 6	Autosomal recessive
8	CLN8	607837	Ceroid lipofuscinosis, neuronal, type 8	Autosomal recessive
11	CLP1	608757	Pontocerebellar hypoplasia, type 10	Autosomal recessive
11	CLPB	616254	3-methylglutaconic aciduria, type 7, with cataracts, neurologic involvement and neutropenia	Autosomal recessive
19	CLPP	601119	Perrault syndrome 3	Autosomal recessive
3	CLRN1	606397	Usher syndrome, type 3A	Autosomal recessive
4	CNGA1	123825	Retinitis pigmentosa type 49	Autosomal recessive
2	CNGA3	600053	Achromatopsia, type 2	Autosomal recessive
16	CNGB1	600724	Retinitis pigmentosa type 45	Autosomal recessive
8	CNGB3	605080	Achromatopsia, type 3	Autosomal recessive
10	CNNM2	607803	Hypomagnesemia, seizures, and mental retardation	Autosomal recessive*
2	CNNM4	607805	Jalili syndrome	Autosomal recessive
6	CNPY3	610774	Epileptic encephalopathy, early infantile, type 60	Autosomal recessive
17	CNTNAP1	602346	Lethal congenital contracture syndrome 7	Autosomal recessive
7	CNTNAP2	604569	Pitt-Hopkins like syndrome 1	Autosomal recessive
1	COA6	614772	Cardioencephalomyopathy, fatal infantile, due to cytochrome c oxidase deficiency 4	Autosomal recessive
14	COA8	616003	Mitochondrial complex IV deficiency, nuclear type 17	Autosomal recessive
17	COASY	609855	Neurodegeneration with brain iron accumulation 6	Autosomal recessive
17	COG1	606973	Congenital disorder of glycosylation, type IIg	Autosomal recessive
16	COG4	606976	Congenital disorder of glycosylation, type 2J	Autosomal recessive

7	COG5	606821	Congenital disorder of glycosylation, type 2I	Autosomal recessive
13	COG6	606977	Congenital disorder of glycosylation, type 2L; Shaheen syndrome	Autosomal recessive
16	COG7	606978	Congenital disorder of glycosylation, type 2E	Autosomal recessive
16	COG8	606979	Congenital disorder of glycosylation, type 2H	Autosomal recessive
1	COL11A1	120280	Fibrochondrogenesis type 1	Autosomal recessive
6	COL11A2	120290	Otospondylomegaepiphyseal dysplasia, autosomal recessive	Autosomal recessive
10	COL13A1	120350	Myasthenic syndrome, congenital, 19	Autosomal recessive
10	COL17A1	113811	Epidermolysis bullosa, junctional, non-Herlitz type	Autosomal recessive
21	COL18A1	120328	Knobloch syndrome, type 1	Autosomal recessive
7	COL1A2	120160	Ehlers-Danlos syndrome, cardiac valvular type	Autosomal recessive
4	COL25A1	610004	Fibrosis of extraocular muscles, congenital, type 5	Autosomal recessive
9	COL27A1	608461	Steel syndrome	Autosomal recessive
2	COL4A3	120070	Alport syndrome, autosomal recessive, type 2	Autosomal recessive; Autosomal dominant
2	COL4A4	120131	Alport syndrome, autosomal recessive, type 2	Autosomal recessive; Autosomal dominant
X	COL4A5	303630	Alport syndrome, X-linked	X-linked
21	COL6A1	120220	Ullrich congenital muscular dystrophy, type 1 (Limb-girdle muscular dystrophy, type 22 [LGMD R22])	Autosomal recessive*
21	COL6A2	120240	Ullrich congenital muscular dystrophy 1; Bethlem myopathy-1; Myosclerosis	Autosomal recessive
2	COL6A3	120250	Bethlem myopathy 1; Ullrich congenital muscular dystrophy 1; Dystonia 27	Autosomal recessive*;Autosomal recessive*;Autosomal recessive
3	COL7A1	120120	Dystrophic epidermolysis bullosa (DEB), Hallopeau-Siemens (HS) type and non-HS type; DEB pruriginosa; DEB pretibial	Autosomal recessive; Autosomal recessive*; Autosomal recessive*
6	COL9A1	120210	Stickler syndrome, type 4	Autosomal recessive
1	COL9A2	120260	?Stickler syndrome, type V	Autosomal recessive
8	COLEC10	607620	3MC syndrome 3	Autosomal recessive
2	COLEC11	612502	3MC syndrome 2	Autosomal recessive
3	COLQ	603033	Myasthenic syndrome, congenital, type 5	Autosomal recessive
4	COQ2	609825	Primary coenzyme Q10 deficiency, type 1	Autosomal recessive
9	COQ4	612898	Coenzyme Q10 deficiency, primary, type 7	Autosomal recessive
14	COQ6	614647	Coenzyme Q10 deficiency, primary, type 6	Autosomal recessive
1	COQ8A	606980	Primary coenzyme Q10 deficiency, type 4	Autosomal recessive
19	COQ8B	615567	Nephrotic syndrome, type 9	Autosomal recessive
16	COQ9	612837	Coenzyme Q10 deficiency, primary, type 5	Autosomal recessive
16	CORO1A	605000	Immunodeficiency, type 8	Autosomal recessive
17	COX10	602125	Mitochondrial complex IV deficiency, nuclear type 3	Autosomal recessive
10	COX15	603646	Cardioencephalomyopathy, fatal infantile, due to cytochrome c oxidase deficiency, type 2; Leigh syndrome due to cytochrome c oxidase deficiency	Autosomal recessive
1	COX20	614698	Mitochondrial complex IV deficiency, nuclear type 11	Autosomal recessive
19	COX6B1	124089	Mitochondrial complex IV deficiency, nuclear type 7	Autosomal recessive
3	CP	117700	Aceruloplasminemia	Autosomal recessive
8	CPA6	609562	Febrile seizures, familial, type 11	Autosomal recessive
19	CPAMD8	608841	Anterior segment dysgenesis, type 8	Autosomal recessive
5	CPLANE1	614571	Joubert syndrome 17	Autosomal recessive
4	CPLX1	605032	Epileptic encephalopathy, early infantile, 63	Autosomal recessive
2	CPS1	608307	Carbamoylphosphate synthetase 1 deficiency	Autosomal recessive
11	CPT1A	600528	Carnitine palmitoyltransferase type 1A deficiency, hepatic	Autosomal recessive
1	CPT2	600650	Carnitine palmitoyltransferase type 2 deficiency, lethal neonatal; Carnitine palmitoyltransferase type 2 deficiency, infantile	Autosomal recessive
1	CR2	120650	Immunodeficiency, common variable, type 7	Autosomal recessive
12	CRADD	603454	Mental retardation, autosomal recessive, type 34, with variant lissencephaly	Autosomal recessive
1	CRB1	604210	Retinitis pigmentosa, type 12; Leber congenital amaurosis, type 8	Autosomal recessive
9	CRB2	609720	Ventriculomegaly with cystic kidney disease	Autosomal recessive
3	CRBN	609262	Mental retardation, autosomal recessive, type 2	Autosomal recessive

2	CRIPT	604594	Short stature with microcephaly and distinctive facies	Autosomal recessive
19	CRLF1	604237	Cold-induced sweating syndrome type 1	Autosomal recessive
7	CRPPA	614631	Muscular dystrophy-dystroglycanopathy, type A7; Muscular dystrophy-dystroglycanopathy, type C7	Autosomal recessive
3	CRTAP	605497	Osteogenesis imperfecta, type 7	Autosomal recessive
21	CRYAA	123580	Cataract 9, multiple types	Autosomal recessive*
11	CRYAB	123590	Myopathy, myofibrillar, fatal infantile hypertonic, alpha-B crystallin-related; Cataract 16, multiple types	Autosomal recessive; Autosomal recessive*
22	CRYBB1	600929	Cataract 17	Autosomal recessive*
22	CRYBB3	123630	Cataract 22	Autosomal recessive
22	CSF2RB	138981	Surfactant metabolism dysfunction, pulmonary, type 5	Autosomal recessive
1	CSF3R	138971	Neutropenia, severe congenital, type 7, autosomal recessive	Autosomal recessive
8	CSPP1	611654	Joubert syndrome 21	Autosomal recessive
3	CSTA	184600	Peeling skin syndrome, type 4	Autosomal recessive
21	CSTB	601145	Epilepsy, progressive myoclonic type 1A (Unverricht and Lundborg)	Autosomal recessive
17	CTC1	613129	Cerebroretinal microangiopathy with calcifications and cysts	Autosomal recessive
1	CTH	607657	Cystathioninuria	Autosomal recessive
17	CTNS	606272	Nephropathic cystinosis	Autosomal recessive
1	CTPS1	123860	Immunodeficiency, type 24	Autosomal recessive
20	CTSA	613111	Galactosialidosis	Autosomal recessive
11	CTSC	602365	Haim-Munk syndrome; Papillon-Lefevre syndrome	Autosomal recessive
11	CTSD	116840	Ceroid lipofuscinosis, neuronal, type 10	Autosomal recessive
11	CTSF	603539	Ceroid lipofuscinosis, neuronal, type 13 (Kufs type)	Autosomal recessive
1	CTSK	601105	Pycnodysostosis	Autosomal recessive
10	CUBN	602997	Megaloblastic anemia 1 (Imerslund-Grasbeck syndrome)	Autosomal recessive
X	CUL4B	300304	Mental retardation, X-linked, syndromic, type 15 (Cabezas type)	X-linked
6	CUL7	609577	3M syndrome 1	Autosomal recessive
5	CWC27	617170	Retinitis pigmentosa with or without skeletal anomalies	Autosomal recessive
10	CWF19L1	616120	Spinocerebellar ataxia, autosomal recessive, type 17	Autosomal recessive
18	CYB5A	613218	46,XY disorder of sex development due to isolated 17,20-lyase deficiency	Autosomal recessive
22	CYB5R3	613213	Methemoglobinemia, type 1; Methemoglobinemia, type 2	Autosomal recessive
16	CYBA	608508	Chronic granulomatous disease, type 4	Autosomal recessive
X	CYBB	300481	Chronic granulomatous disease, X-linked	X-linked
8	CYC1	123980	Mitochondrial complex III deficiency, nuclear type 6	Autosomal recessive
15	CYP11A1	118485	46,XY disorder of sex development-adrenal insufficiency due to CYP11A1 deficiency	Autosomal recessive
8	CYP11B1	610613	Adrenal hyperplasia, congenital, due to 11-beta-hydroxylase deficiency	Autosomal recessive
8	CYP11B2	124080	Hypoaldosteronism, congenital, due to CMO I deficiency	Autosomal recessive
10	CYP17A1	609300	17 alpha(?)-hydroxylase/17,20-lyase deficiency	Autosomal recessive
15	CYP19A1	107910	Aromatase deficiency	Autosomal recessive
2	CYP1B1	601771	Glaucoma, primary congenital, type 3A	Autosomal recessive
6	CYP21A2	613815	Congenital adrenal hyperplasia due to 21-hydroxylase deficiency	Autosomal recessive
20	CYP24A1	126065	Hypercalcemia, infantile, type 1	Autosomal recessive
2	CYP26B1	605207	Craniosynostosis with radiohumeral fusions and other skeletal and craniofacial anomalies	Autosomal recessive
10	CYP26C1	608428	Focal facial dermal dysplasia 4	digenic inheritance (CLCNKB gene)
2	CYP27A1	606530	Cerebrotendinous xanthomatosis	Autosomal recessive
12	CYP27B1	609506	Vitamin D-dependent rickets, type 1	Autosomal recessive
11	CYP2R1	608713	Rickets due to defect in vitamin D 25-hydroxylation	Autosomal recessive
4	CYP2U1	610670	Spastic paraplegia, type 56, autosomal recessive	Autosomal recessive
19	CYP4F22	611495	Ichthyosis, congenital, autosomal recessive, type 5	Autosomal recessive
4	CYP4V2	608614	Bietti crystalline corneoretinal dystrophy	Autosomal recessive
8	CYP7B1	603711	Spastic paraplegia, type 5A, autosomal recessive	Autosomal recessive

2	D2HGDH	609186	D-2-hydroxyglutaric aciduria	Autosomal recessive
3	DAG1	128239	Muscular dystrophy-dystroglycanopathy type A9; Muscular dystrophy-dystroglycanopathy type C9	Autosomal recessive
2	DARS1	603084	Hypomyelination with brainstem and spinal cord involvement and leg spasticity	Autosomal recessive
1	DARS2	610956	Leukoencephalopathy with brain stem and spinal cord involvement and lactate elevation	Autosomal recessive
9	DBH	609312	Dopamine beta-hydroxylase deficiency	Autosomal recessive
1	DBT	248610	Maple syrup urine disease, type 2	Autosomal recessive
2	DCAF17	612515	Woodhouse-Sakati syndrome	Autosomal recessive
18	DCC	120470	Gaze palsy, familial horizontal, with progressive scoliosis, type 2	Autosomal recessive
6	DCDC2	605755	Sclerosing cholangitis, neonatal; Nephronophthisis 19	Autosomal recessive
11	DCHS1	603057	Van Maldergem syndrome 1	Autosomal recessive
10	DCLRE1C	605988	Omenn syndrome; Severe combined immunodeficiency, Athabascan type	Autosomal recessive
11	DCPS	610534	Al-Raqad syndrome	Autosomal recessive
X	DCX	300121	Lissencephaly, X-linked, type 1	X-linked
11	DDB2	600811	Xeroderma pigmentosum, complementation group E	Autosomal recessive
7	DDC	107930	Aromatic L-amino acid decarboxylase deficiency	Autosomal recessive
14	DDHD1	614603	Spastic paraplegia, type 28, autosomal recessive	Autosomal recessive
8	DDHD2	615003	Spastic paraplegia, type 54, autosomal recessive	Autosomal recessive
1	DDR2	191311	Spondylometaepiphyseal dysplasia, short limb-hand type	Autosomal recessive
20	DDRGK1	616177	Spondyloepimetaphyseal dysplasia, Shohat type	Autosomal recessive
12	DDX11	601150	Warsaw breakage syndrome	Autosomal recessive
1	DDX59	615464	Orofaciodigital syndrome V	Autosomal recessive
11	DENND5A	617278	Epileptic encephalopathy, early infantile, 49	Autosomal recessive
2	DES	125660	Myopathy, myofibrillar, type 1	Autosomal recessive*; Autosomal dominant
8	DGAT1	604900	?Diarrhea 7, protein-losing enteropathy type	Autosomal recessive
17	DGKE	601440	Nephrotic syndrome, type 7	Autosomal recessive
2	DGUOK	601465	DGUOK-related mitochondrial DNA depletion syndrome	Autosomal recessive
1	DHCR24	606418	Desmosterolosis	Autosomal recessive
11	DHCR7	602858	Smith-Lemli-Opitz syndrome	Autosomal recessive
1	DHDDS	608172	Retinitis pigmentosa, type 59	Autosomal recessive
5	DHFR	126060	Megaloblastic anemia due to dihydrofolate reductase deficiency	Autosomal recessive
12	DHH	605423	46,XY complete gonadal dysgenesis	Autosomal recessive
16	DHODH	126064	Miller syndrome	Autosomal recessive
19	DHPS	600944	Neurodevelopmental disorder with seizures and speech and walking impairment	Autosomal recessive
10	DHTKD1	614984	2-aminoadipic 2-oxoadipic aciduria	Autosomal recessive
5	DIAPH1	602121	Seizures, cortical blindness, microcephaly syndrome	Autosomal recessive
2	DIS3L2	614184	Perlman syndrome	Autosomal recessive
X	DKC1	300126	Dyskeratosis congenita, X-linked	X-linked
11	DLAT	608770	Pyruvate dehydrogenase E2 deficiency	Autosomal recessive
7	DLD	238331	Dihydrolipoamide dehydrogenase deficiency	Autosomal recessive
X	DLG3	300189	Mental retardation, X-linked, type 90	X-linked
19	DLL3	602768	Spondylocostal dysostosis type 1	Autosomal recessive
X	DMD	300377	DMD-related conditions	X-linked
5	DMGDH	605849	Dimethylglycine dehydrogenase deficiency	Autosomal recessive
4	DMP1	600980	Hypophosphatemic rickets, autosomal recessive	Autosomal recessive
15	DMXL2	612186	Developmental and epileptic encephalopathy, type 81	Autosomal recessive
16	DNAAF1	613190	Ciliary dyskinesia, primary, type 13	Autosomal recessive
8	DNAAF11	614930	Ciliary dyskinesia, primary, type 19	Autosomal recessive
14	DNAAF2	612517	Ciliary dyskinesia, primary, type 10	Autosomal recessive

19	DNAAF3	614566	Ciliary dyskinesia, primary, type 2	Autosomal recessive
15	DNAAF4	608706	Ciliary dyskinesia, primary, type 25	Autosomal recessive
7	DNAAF5	614864	Ciliary dyskinesia, primary, type 18	Autosomal recessive
3	DNAH1	603332	Spermatogenic failure, type 18	Autosomal recessive
7	DNAH11	603339	Ciliary dyskinesia, primary, type 7, with or without situs inversus	Autosomal recessive
5	DNAH5	603335	Ciliary dyskinesia, primary, type 3, with or without situs inversus	Autosomal recessive
17	DNAH9	603330	Ciliary dyskinesia, primary, type 40	Autosomal recessive
9	DNAI1	604366	Ciliary dyskinesia, primary, type 1, with or without situs inversus	Autosomal recessive
17	DNAI2	605483	Ciliary dyskinesia, primary, type 9, with or without situs inversus	Autosomal recessive
11	DNAJB13	610263	Ciliary dyskinesia, primary, type 34	Autosomal recessive
2	DNAJB2	604139	Spinal muscular atrophy, distal, autosomal recessive, type 5	Autosomal recessive
10	DNAJC12	606060	Hyperphenylalaninemia, mild, non-BH4-deficient	Autosomal recessive
3	DNAJC19	608977	3-methylglutaconic aciduria, type 5	Autosomal recessive
5	DNAJC21	617048	Bone marrow failure syndrome, type 3	Autosomal recessive
1	DNAJC6	608375	Parkinson disease, type 19A, juvenile-onset; Parkinson disease, type 19B, early-onset	Autosomal recessive
14	DNAL1	610062	Ciliary dyskinesia, primary, type 16	Autosomal recessive
3	DNASE1L3	602244	Systemic lupus erythematosus 16	Autosomal recessive
12	DNM1L	603850	Encephalopathy due to defective mitochondrial and peroxisomal fission, type 1	Autosomal recessive*
19	DNM2	602378	Lethal congenital contracture syndrome, type 5	Autosomal recessive
20	DNMT3B	602900	Immunodeficiency-centromeric instability-facial anomalies syndrome, type 1	Autosomal recessive
5	DOCK2	603122	Immunodeficiency, type 40	Autosomal recessive
19	DOCK6	614194	Adams-Oliver syndrome 2	Autosomal recessive
1	DOCK7	615730	Epileptic encephalopathy, early infantile, 23	Autosomal recessive
9	DOCK8	611432	Hyper-IgE recurrent infection syndrome, autosomal recessive	Autosomal recessive
4	DOK7	610285	Fetal akinesia deformation sequence, type 3; Myasthenic syndrome, congenital, type 10	Autosomal recessive
9	DOLK	610746	Congenital disorder of glycosylation, type 1M	Autosomal recessive
21	DONSON	611428	Microcephaly, short stature, and limb abnormalities	Autosomal recessive
11	DPAGT1	191350	Congenital disorder of glycosylation, type 1J; Myasthenic syndrome, congenital, type 13	Autosomal recessive
17	DPH1	603527	Developmental delay with short stature, dysmorphic features, and sparse hair	Autosomal recessive
20	DPM1	603503	Congenital disorder of glycosylation, type 1E	Autosomal recessive
9	DPM2	603564	Congenital disorder of glycosylation, type Iu	Autosomal recessive
1	DPM3	605951	Congenital disorder of glycosylation, type Io	Autosomal recessive
12	DPY19L2	613893	Spermatogenic failure, type 9	Autosomal recessive
1	DPYD	612779	Dihydropyrimidine dehydrogenase deficiency	Autosomal recessive
8	DPYS	613326	Dihydropyrimidinuria	Autosomal recessive
1	DRAM2	613360	Cone-rod dystrophy 21	Autosomal recessive
2	DRC1	615288	Ciliary dyskinesia, primary, type 21	Autosomal recessive
18	DSG1	125670	Erythroderma, congenital, with palmoplantar keratoderma, hypotrichosis, and hyper IgE	Autosomal recessive
18	DSG4	607892	Hypotrichosis, type 6	Autosomal recessive
6	DSP	125647	Cardiomyopathy, dilated, with woolly hair and keratoderma; Epidermolysis bullosa, lethal acantholytic	Autosomal recessive
6	DST	113810	Neuropathy, hereditary sensory and autonomic, type VI;Epidermolysis bullosa simplex 3, localized or generalized intermediate, with bp230 deficiency	Autosomal recessive
1	DSTYK	612666	Spastic paraplegia, type 23, autosomal recessive	Autosomal recessive
6	DTNBP1	607145	Hermansky-Pudlak syndrome, type 7	Autosomal recessive
15	DUOX2	606759	Thyroid dyshormonogenesis, type 6	Autosomal recessive
15	DUOXA2	612772	Thyroid dyshormonogenesis, type 5	Autosomal recessive
18	DYM	607461	Smith-McCort dysplasia; Dyggve-Melchior-Clausen disease	Autosomal recessive
11	DYNC2H1	603297	Short-rib thoracic dysplasia, type 3, with or without polydactyly	Autosomal recessive
7	DYNC2I1	615462	Short-rib thoracic dysplasia 8 with or without polydactyly	Autosomal recessive
9	DYNC2I2	613363	Short-rib thoracic dysplasia 11 with or without polydactyly	Autosomal recessive

2	DYNC2LI1	617083	Short-rib thoracic dysplasia 15 with polydactyly	Autosomal recessive
3	DYNLT2B	617353	Short-rib thoracic dysplasia 17 with or without polydactyly	Autosomal recessive
2	DYSF	603009	Miyoshi muscular dystrophy, type 1; Limb-girdle muscular dystrophy, type 2 (LGMD R2)	Autosomal recessive
3	DZIP1L	617570	Polycystic kidney disease 5	Autosomal recessive
16	EARS2	612799	Combined oxidative phosphorylation deficiency 12	Autosomal recessive
2	ECEL1	605896	Arthrogryposis, distal, type 5D	Autosomal recessive
10	ECHS1	602292	Mitochondrial short-chain enoyl-CoA hydratase 1 deficiency	Autosomal recessive
1	ECM1	602201	Urbach-Wiethe disease	Autosomal recessive
X	EDA	300451	Ectodermal dysplasia, type 1, hypohidrotic, X-linked	X-linked
2	EDAR	604095	Ectodermal dysplasia 10B, hypohidrotic/hair/tooth type	Autosomal recessive
1	EDARADD	606603	Ectodermal dysplasia 11B, hypohidrotic/hair/tooth type	Autosomal recessive
6	EDN1	131240	Auriculocondylar syndrome, type 3	Autosomal recessive
20	EDN3	131242	Waardenburg syndrome, type 4B	Autosomal recessive
13	EDNRB	131244	ABCD syndrome	Autosomal recessive
11	EFEMP2	604633	Cutis laxa, autosomal recessive, type 1B	Autosomal recessive
15	EFL1	617538	Shwachman-Diamond syndrome 2	Autosomal recessive
7	EGFR	131550	?Inflammatory skin and bowel disease, neonatal, 2	Autosomal recessive
10	EGR2	129010	Dejerine-Sottas disease	Autosomal recessive*
2	EIF2AK3	604032	Wolcott-Rallison syndrome	Autosomal recessive
15	EIF2AK4	609280	Pulmonary venoocclusive disease 2	Autosomal recessive
12	EIF2B1	606686	Leukoencephalopathy with vanishing white matter (VWM)	Autosomal recessive
14	EIF2B2	606454	Leukoencephalopathy with vanishing white matter (VWM)	Autosomal recessive
1	EIF2B3	606273	Leukoencephalopathy with vanishing white matter (VWM)	Autosomal recessive
2	EIF2B4	606687	Leukoencephalopathy with vanishing white matter (VWM)	Autosomal recessive
3	EIF2B5	603945	Leukoencephalopathy with vanishing white matter (VWM)	Autosomal recessive
17	EIF4A3	608546	Robin sequence with cleft mandible and limb anomalies	Autosomal recessive
17	ELAC2	605367	Combined oxidative phosphorylation deficiency 17	Autosomal recessive
20	ELMO2	606421	Vascular malformation, primary intraosseous	Autosomal recessive
6	ELOVL4	605512	Ichthyosis, spastic quadriplegia, and mental retardation	Autosomal recessive
9	ELP1	603722	Familial dysautonomia	Autosomal recessive
18	ELP2	616054	Mental retardation, autosomal recessive, type 58	Autosomal recessive
1	EMC1	616846	Cerebellar atrophy, visual impairment, and psychomotor retardation	Autosomal recessive
X	EMD	300384	Emery-Dreifuss muscular dystrophy, type 1, X-linked	X-linked
14	EML1	602033	Band heterotopia	Autosomal recessive
16	EMP2	602334	Nephrotic syndrome, type 10	Autosomal recessive
4	ENAM	606585	Amelogenesis imperfecta, type 1C	Autosomal recessive
17	ENO3	131370	?Glycogen storage disease XIII	Autosomal recessive
6	ENPP1	173335	Arterial calcification, generalized, of infancy, type 1	Autosomal recessive
10	ENTPD1	601752	Spastic paraplegia, type 64, autosomal recessive	Autosomal recessive
3	EOGT	614789	Adams-Oliver syndrome 4	Autosomal recessive
1	EPB41	130500	Elliptocytosis, type 1	Autosomal recessive*
15	EPB42	177070	Spherocytosis, type 5	Autosomal recessive
2	EPCAM	185535	EPCAM-related conditions	Autosomal recessive
18	EPG5	615068	Vici syndrome	Autosomal recessive
6	EPM2A	607566	Epilepsy, progressive myoclonic, type 2A (Lafora)	Autosomal recessive
1	EPRS1	138295	Leukodystrophy, hypomyelinating, type 15	Autosomal recessive
11	EPS8L2	614988	Deafness autosomal recessive, type 106	Autosomal recessive
17	ERAL1	607435	Perrault syndrome 6	Autosomal recessive

12	ERBB3	190151	Lethal congenital contractural syndrome, type 2	Autosomal recessive
19	ERCC1	126380	Cerebrooculofacioskeletal syndrome, type 4	Autosomal recessive
19	ERCC2	126340	Trichothiodystrophy, type 1; Xeroderma pigmentosum, group D	Autosomal recessive
2	ERCC3	133510	Trichothiodystrophy, type 2	Autosomal recessive
16	ERCC4	133520	Fanconi anemia, complementation group Q	Autosomal recessive
13	ERCC5	133530	Cerebrooculofacioskeletal syndrome 3; Xeroderma pigmentosum, group G;Xeroderma pigmentosum, group G/Cockayne syndrome	Autosomal recessive
10	ERCC6	609413	Cockayne syndrome, type B; Cerebrooculofacioskeletal syndrome, type 1	Autosomal recessive
9	ERCC6L2	615667	Bone marrow failure syndrome, type 2	Autosomal recessive
5	ERCC8	609412	Cockayne syndrome, type A	Autosomal recessive
10	ERLIN1	611604	Spastic paraplegia, type 62, autosomal recessive	Autosomal recessive
8	ERLIN2	611605	Spastic paraplegia, type 18, autosomal recessive	Autosomal recessive
8	ESCO2	609353	Roberts syndrome	Autosomal recessive
1	ESPN	606351	Deafness, autosomal recessive, type 36	Autosomal recessive
6	ESR1	133430	Estrogen resistance	Autosomal recessive
14	ESRRB	602167	Deafness, autosomal recessive, type 35	Autosomal recessive
15	ETFA	608053	Glutaric acidemia, type 2A	Autosomal recessive
19	ETFB	130410	Glutaric acidemia, type 2B	Autosomal recessive
4	ETFDH	231675	Glutaric acidemia, type 2C	Autosomal recessive
19	ETHE1	608451	Ethylmalonic encephalopathy	Autosomal recessive
4	EVC	604831	Ellis-van Creveld syndrome	Autosomal recessive
4	EVC2	607261	Ellis-van Creveld syndrome	Autosomal recessive
9	EXOSC3	606489	Pontocerebellar hypoplasia, type 1B	Autosomal recessive
11	EXPH5	612878	Epidermolysis bullosa, nonspecific, autosomal recessive	Autosomal recessive
8	EXTL3	605744	Immunoskeletal dysplasia with neurodevelopmental abnormalities	Autosomal recessive
6	EYS	612424	Retinitis pigmentosa, type 25	Autosomal recessive
13	F10	613872	Factor X deficiency	Autosomal recessive
4	F11	264900	Factor XI deficiency	Autosomal recessive*
6	F13A1	134570	Factor XIIIA deficiency	Autosomal recessive
1	F13B	134580	Factor XIIIB deficiency	Autosomal recessive
11	F2	176930	Prothrombin deficiency	Autosomal recessive
1	F5	612309	Factor V deficiency	Autosomal recessive
13	F7	613878	Factor VII deficiency	Autosomal recessive
X	F8	300841	Hemophilia A	X-linked
X	F9	300746	Hemophilia B	X-linked
16	FA2H	611026	Spastic paraplegia, type 35, autosomal recessive	Autosomal recessive
11	FADD	602457	Infections, recurrent, with encephalopathy, hepatic dysfunction, and cardiovascular malformations	Autosomal recessive
15	FAH	613871	Tyrosinemia, type 1	Autosomal recessive
2	FAM161A	613596	Retinitis pigmentosa, type 28	Autosomal recessive
17	FAM20A	611062	Amelogenesis imperfecta, type 1G (Enamel-renal syndrome)	Autosomal recessive
7	FAM20C	611061	Raine syndrome	Autosomal recessive
15	FAN1	613534	Interstitial nephritis, karyomegalic	Autosomal recessive
16	FANCA	607139	Fanconi anemia, complementation group A	Autosomal recessive
9	FANCC	613899	Fanconi anemia, complementation group C	Autosomal recessive
3	FANCD2	613984	Fanconi anemia, complementation group D2	Autosomal recessive
6	FANCE	613976	Fanconi anemia, complementation group E	Autosomal recessive
11	FANCF	613897	Fanconi anemia, complementation group F	Autosomal recessive
9	FANCG	602956	Fanconi anemia, complementation group G	Autosomal recessive
15	FANCI	611360	Fanconi anemia, complementation group I	Autosomal recessive
2	FANCL	608111	Fanconi anemia, complementation group L	Autosomal recessive

14	FANCM	609644	Spermatogenic failure, type 28; ?Premature ovarian failure 15	Autosomal recessive
11	FAR1	616107	Peroxisomal fatty acyl-CoA reductase 1 disorder	Autosomal recessive
6	FARS2	611592	Combined oxidative phosphorylation deficiency 14; Spastic paraplegia, type 77, autosomal recessive	Autosomal recessive
2	FASTKD2	612322	Combined oxidative phosphorylation deficiency 44	Autosomal recessive
4	FAT4	612411	Hennekam lymphangiectasia-lymphedema syndrome 2	Autosomal recessive
14	FBLN5	604580	Cutis laxa, autosomal recessive, type 1A	Autosomal recessive
9	FBP1	611570	Fructose-1,6-bisphosphatase deficiency	Autosomal recessive
6	FBXL4	605654	Mitochondrial DNA depletion syndrome 13 (encephalomyopathic type)	Autosomal recessive
22	FBXO7	605648	Parkinson disease, type 15, autosomal recessive	Autosomal recessive
17	FDXR	103270	Auditory neuropathy and optic atrophy	Autosomal recessive
18	FECH	612386	Protoporphyria, erythropoietic, autosomal recessive	Autosomal recessive
20	FERMT1	607900	Kindler syndrome	Autosomal recessive
11	FERMT3	607901	Leukocyte adhesion deficiency, type 3	Autosomal recessive
7	FEZF1	613301	Hypogonadotropic hypogonadism type 22, with or without anosmia	Autosomal recessive
4	FGA	134820	Afibrinogenemia, congenital	Autosomal recessive
4	FGB	134830	Congenital afibrinogenemia	Autosomal recessive
X	FGD1	300546	Aarskog-Scott syndrome; Mental retardation, X-linked syndromic, type 16	X-linked
12	FGD4	611104	Charcot-Marie-Tooth disease, type 4H	Autosomal recessive
12	FGF23	605380	Tumoral calcinosis, hyperphosphatemic, familial, type 2	Autosomal recessive
11	FGF3	164950	Deafness, congenital with inner ear agenesis, microtia, and microdontia	Autosomal recessive
4	FGG	134850	Afibrinogenemia, congenital; Hypofibrinogenemia, congenital	Autosomal recessive
1	FH	136850	Fumarase deficiency	Autosomal recessive
11	FIBP	608296	Thauvin-Robinet-Faivre syndrome	Autosomal recessive
6	FIG4	609390	Charcot-Marie-Tooth disease, type 4J; Yunis-Varon syndrome	Autosomal recessive
17	FKBP10	607063	Bruck syndrome 1	Autosomal recessive
7	FKBP14	614505	Ehlers-Danlos syndrome, kyphoscoliotic type, 2	Autosomal recessive
19	FKRP	606596	Muscular dystrophy-dystroglycanopathy, type 5A (Walker-Warburg syndrome); Type 5B; Type 5C (limb-girdle muscular dystrophy, type 9 [LGMDR9])	Autosomal recessive
9	FKTN	607440	Muscular dystrophy-dystroglycanopathy, type 4A (Walker-Warburg syndrome); Type 4B; Type 4C (limb-girdle muscular dystrophy, type 13 [LGMD R13])	Autosomal recessive
1	FLAD1	610595	Lipid storage myopathy due to flavin adenine dinucleotide synthetase deficiency	Autosomal recessive
1	FLG	135940	Ichthyosis vulgaris	Autosomal recessive*
11	FLI1	193067	Bleeding disorder, platelet-type, type 21	Autosomal recessive*
3	FLNB	603381	Spondylocarpotarsal synostosis syndrome	Autosomal recessive
1	FLVCR1	609144	Posterior column ataxia-retinitis pigmentosa syndrome	Autosomal recessive
14	FLVCR2	610865	Proliferative vasculopathy and hydranencephaly-hydrocephaly syndrome	Autosomal recessive
1	FMN2	606373	Mental retardation, autosomal recessive, type 47	Autosomal recessive
1	FMO3	136132	Trimethylaminuria	Autosomal recessive
X	FMR1	309550	FMR1-related conditions	X-linked
11	FOLR1	136430	Neurodegeneration due to cerebral folate transport deficiency	Autosomal recessive
9	FOXE1	602617	Bamforth-Lazarus syndrome	Autosomal recessive
1	FOXE3	601094	Anterior segment dysgenesis, type 2, multiple subtypes	Autosomal recessive
17	FOXN1	600838	T-cell immunodeficiency, congenital alopecia and nail dystrophy	Autosomal recessive
11	FOXRED1	613622	Mitochondrial complex I deficiency, nuclear type 19	Autosomal recessive
4	FRAS1	607830	Fraser syndrome, type 1	Autosomal recessive
9	FREM1	608944	Manitoba oculotrichoanal syndrome	Autosomal recessive
13	FREM2	608945	Fraser syndrome, type 2	Autosomal recessive
9	FRRS1L	604574	Epileptic encephalopathy, early infantile, 37	Autosomal recessive
11	FSHB	136530	Hypogonadotropic hypogonadism, type 24, without anosmia	Autosomal recessive
2	FSHR	136435	Ovarian dysgenesis 1;Ovarian hyperstimulation syndrome;Ovarian response to FSH stimulation	Autosomal recessive; Autosomal dominant; Autosomal recessive

21	FTCD	606806	Glutamate formiminotransferase deficiency	Autosomal recessive
19	FTL	134790	L-ferritin deficiency	Autosomal recessive*
16	FTO	610966	Growth retardation, developmental delay, facial dysmorphism	Autosomal recessive
X	FTSJ1	300499	Mental retardation, X-linked 44	X-linked
1	FUCA1	612280	Fucosidosis	Autosomal recessive
14	FUT8	602589	Congenital disorder of glycosylation with defective fucosylation, type 1	Autosomal recessive
9	FXN	606829	Friedreich ataxia	Autosomal recessive
3	FYCO1	607182	Cataract 18	Autosomal recessive
8	FZD6	603409	Nail disorder, nonsyndromic congenital, type 10 (claw-shaped nails)	Autosomal recessive
17	G6PC1	613742	Glycogen storage disease, type 1A	Autosomal recessive
17	G6PC3	611045	Dursun syndrome	Autosomal recessive
X	G6PD	305900	G6PD deficiency	X-linked
17	GAA	606800	Glycogen storage disease, type 2	Autosomal recessive
14	GALC	606890	Krabbe disease	Autosomal recessive
1	GALE	606953	Galactose epimerase deficiency	Autosomal recessive
17	GALK1	604313	Galactokinase deficiency with cataracts	Autosomal recessive
16	GALNS	612222	Mucopolysaccharidosis, type 4A	Autosomal recessive
2	GALNT3	601756	Tumoral calcinosis, hyperphosphatemic, familial, type 1	Autosomal recessive
9	GALT	606999	Galactosemia	Autosomal recessive
19	GAMT	601240	Cerebral creatine deficiency syndrome, type 2	Autosomal recessive
16	GAN	605379	Giant axonal neuropathy, type 1	Autosomal recessive
16	GAS8	605178	Ciliary dyskinesia, primary, type 33	Autosomal recessive
15	GATM	602360	Cerebral creatine deficiency syndrome, type 3	Autosomal recessive
1	GBA1	606463	Gaucher disease	Autosomal recessive
9	GBA2	609471	Spastic paraplegia, type 46, autosomal recessive	Autosomal recessive
3	GBE1	607839	Glycogen storage disease, type 4	Autosomal recessive
19	GCDH	608801	Glutaricaciduria, type 1	Autosomal recessive
14	GCH1	600225	Hyperphenylalaninemia, BH4-deficient, type B	Autosomal recessive
7	GCK	138079	Permanent neonatal diabetes mellitus (PNDM)	Autosomal recessive*
6	GCM2	603716	Hypoparathyroidism, familial isolated (FIH) 2	Autosomal recessive
6	GCNT2	600429	Cataract 13, with adult i phenotype	Autosomal recessive
16	GCSH	238330	Multiple mitochondrial dysfunctions syndrome 7	Autosomal recessive
8	GDAP1	606598	Charcot-Marie-Tooth disease, recessive intermediate, type A	Autosomal recessive
19	GDF1	602880	Right atrial isomerism (Ivemark syndrome)	Autosomal recessive
20	GDF5	601146	Chondrodysplasia, Grebe type	Autosomal recessive
8	GDF6	601147	Leber congenital amaurosis, type 17	Autosomal recessive
16	GFER	600924	Myopathy, mitochondrial progressive, with congenital cataract, hearing loss, and developmental delay	Autosomal recessive
3	GFM1	606639	Combined oxidative phosphorylation deficiency, type 1	Autosomal recessive
2	GFPT1	138292	Myasthenia, congenital, type 12, with tubular aggregates	Autosomal recessive
2	GGCX	137167	Vitamin K-dependent clotting factors, combined deficiency of, type 1	Autosomal recessive
17	GH1	139250	Growth hormone deficiency, isolated, type 1A; Kowarski syndrome	Autosomal recessive
5	GHR	600946	Laron dwarfism	Autosomal recessive
7	GHRHR	139191	Growth hormone deficiency, isolated, type 1B	Autosomal recessive
3	GHSR	601898	Growth hormone deficiency, isolated partial	Autosomal recessive
20	GINS1	610608	Immunodeficiency, type 55	Autosomal recessive
19	GIPC3	608792	Deafness, autosomal recessive, type 15	Autosomal recessive
6	GJA1	121014	Craniometaphyseal dysplasia, autosomal recessive	Autosomal recessive
X	GJB1	304040	Charcot-Marie-Tooth neuropathy, X-linked dominant, type 1	X-linked
13	GJB2	121011	Deafness, autosomal recessive, type 1A; Deafness, digenic, GJB2/GJB6	Autosomal recessive; Digenic inheritance (GJB6 gene)

13	GJB6	604418	Deafness, autosomal recessive, type 1B; Deafness, digenic GJB2/GJB6	Autosomal recessive; Digenic inheritance (GJB2 gene)
1	GJC2	608803	Spastic paraplegia, type 44, autosomal recessive	Autosomal recessive
X	GLA	300644	Fabry disease	X-linked
3	GLB1	611458	GM1-gangliosidosis, types 1-3; Mucopolysaccharidosis, type 4B (Morquio)	Autosomal recessive
9	GLDC	238300	Glycine encephalopathy	Autosomal recessive
15	GLDN	608603	Lethal congenital contracture syndrome 11	Autosomal recessive
9	GLE1	603371	Lethal congenital contracture syndrome, type 1; Congenital arthrogryposis with anterior horn cell disease	Autosomal recessive
16	GLIS2	608539	Nephronophthisis, type 7	Autosomal recessive
9	GLIS3	610192	Diabetes mellitus, neonatal, with congenital hypothyroidism	Autosomal recessive
5	GLRA1	138491	Hyperekplexia, type 1	Autosomal recessive*
4	GLRB	138492	Hyperekplexia, type 2	Autosomal recessive
14	GLRX5	609588	Anemia, sideroblastic, type 3, pyridoxine-refractory; Spasticity, childhood-onset, with hyperglycinemia	Autosomal recessive
1	GLUL	138290	Glutamine deficiency, congenital	Autosomal recessive
3	GLYCTK	610516	D-glyceric aciduria	Autosomal recessive
5	GM2A	613109	GM2-gangliosidosis, AB variant	Autosomal recessive
2	GMPPA	615495	Alacrima, achalasia, and mental retardation syndrome	Autosomal recessive
3	GMPPB	615320	Muscular dystrophy-dystroglycanopathy 14	Autosomal recessive
3	GNAT1	139330	Night blindness, congenital stationary, type 1G	Autosomal recessive
1	GNAT2	139340	Achromatopsia, type 4	Autosomal recessive
15	GNB5	604447	Intellectual developmental disorder with cardiac arrhythmia; Language delay and ADHD/cognitive impairment with or without cardiac arrhythmia	Autosomal recessive
9	GNE	603824	Inclusion body myopathy, type 2 (Nonaka myopathy)	Autosomal recessive
6	GNMT	606628	Glycine N-methyltransferase deficiency	Autosomal recessive
1	GNPAT	602744	Rhizomelic chondrodysplasia punctata, type 2	Autosomal recessive
12	GNPTAB	607840	Mucolipidosis 2 alpha/beta; Mucolipidosis 3 alpha/beta	Autosomal recessive
16	GNPTG	607838	Mucolipidosis III gamma	Autosomal recessive
4	GNRHR	138850	Hypogonadotropic hypogonadism, type 7, without anosmia	Autosomal recessive
12	GNS	607664	Mucopolysaccharidosis, type 3D (Sanfilippo syndrome D)	Autosomal recessive
1	GORAB	607983	Geroderma osteodysplasticum	Autosomal recessive
17	GOSR2	604027	Epilepsy, progressive myoclonic, type 6	Autosomal recessive
16	GOT2	138150	Epileptic encephalopathy, early infantile, 82	Autosomal recessive
17	GP1BA	606672	Bernard-Soulier syndrome, type A1	Autosomal recessive
22	GP1BB	138720	Bernard-Soulier syndrome, type B	Autosomal recessive
19	GP6	605546	Bleeding disorder, platelet-type, type 11	Autosomal recessive
3	GP9	173515	Bernard-Soulier syndrome, type C	Autosomal recessive
8	GPAA1	603048	Glycosylphosphatidylinositol biosynthesis defect 15	Autosomal recessive
13	GPC6	604404	Omodysplasia, type 1	Autosomal recessive
12	GPD1	138420	Hypertriglyceridemia, transient infantile	Autosomal recessive
14	GPHN	603930	Molybdenum cofactor deficiency C	Autosomal recessive
19	GPI	172400	Hemolytic anemia, nonspherocytic, due to glucose phosphate isomerase deficiency	Autosomal recessive
8	GPIHBP1	612757	Hyperlipoproteinemia, type 1D	Autosomal recessive
X	GPR143	300808	Ocular albinism, type 1 (Nettleship-Falls type)	X-linked
17	GPR179	614515	Night blindness, congenital stationary (complete), type 1E, autosomal recessive	Autosomal recessive
14	GPR68	601404	Amelogenesis imperfecta, type 2A6 (hypomaturation type)	Autosomal recessive
1	GPSM2	609245	Chudley-McCullough syndrome	Autosomal recessive
16	GPT2	138210	Mental retardation, autosomal recessive 49	Autosomal recessive
19	GPX4	138322	Spondylometaphyseal dysplasia, Sedaghatian type	Autosomal recessive
8	GRHL2	608576	Ectodermal dysplasia/short stature syndrome	Autosomal recessive
9	GRHPR	604296	Hyperoxaluria, primary, type 2	Autosomal recessive

4	GRID2	602368	Spinocerebellar ataxia, autosomal recessive, type 18	Autosomal recessive
6	GRIK2	138244	Mental retardation, autosomal recessive, type, 6	Autosomal recessive
9	GRIN1	138249	Neurodevelopmental disorder with or without hyperkinetic movements and seizures, autosomal recessive	Autosomal recessive
12	GRIP1	604597	Fraser syndrome 3	Autosomal recessive
13	GRK1	180381	Oguchi disease-2	Autosomal recessive
6	GRM1	604473	Spinocerebellar ataxia, autosomal recessive, type 13	Autosomal recessive
5	GRM6	604096	Night blindness, congenital stationary (complete), type 1B, autosomal recessive	Autosomal recessive
17	GRN	138945	Ceroid lipofuscinosis, neuronal, type 11	Autosomal recessive
4	GRXCR1	613283	Deafness, autosomal recessive, type 25	Autosomal recessive
14	GSC	138890	Short stature, auditory canal atresia, mandibular hypoplasia, skeletal abnormalities	Autosomal recessive
20	GSS	601002	Glutathione synthetase deficiency	Autosomal recessive
6	GTF2H5	608780	Trichothiodystrophy, type 3, photosensitive	Autosomal recessive
6	GTPBP2	607434	Jaberi-Elahi syndrome	Autosomal recessive
19	GTPBP3	608536	Combined oxidative phosphorylation deficiency 23	Autosomal recessive
12	GUCY2C	601330	Meconium ileus	Autosomal recessive
17	GUCY2D	600179	Leber congenital amaurosis, type 1	Autosomal recessive
4	GUF1	617064	?Epileptic encephalopathy, early infantile, 40	Autosomal recessive
7	GUSB	611499	Mucopolysaccharidosis, type 7	Autosomal recessive
3	GYG1	603942	Polyglucosan body myopathy, type 2	Autosomal recessive
19	GYS1	138570	Glycogen storage disease, type 0, muscle	Autosomal recessive
12	GYS2	138571	Glycogen storage disease, type 0, liver	Autosomal recessive
20	GZF1	613842	Joint laxity, short stature, and myopia	Autosomal recessive
1	H6PD	138090	Cortisone reductase deficiency 1	Autosomal recessive
2	HAAO	604521	Vertebral, cardiac, renal, and limb defects syndrome 1	Autosomal recessive
6	HACE1	610876	Spastic paraplegia and psychomotor retardation with or without seizures	Autosomal recessive
4	HADH	601609	3-hydroxyacyl-CoA dehydrogenase deficiency	Autosomal recessive
2	HADHA	600890	Long-chain 3-hydroxyl-CoA dehydrogenase (LCHAD) deficiency; Mitochondrial trifunctional protein deficiency	Autosomal recessive
2	HADHB	143450	Mitochondrial trifunctional protein deficiency	Autosomal recessive
19	HAMP	606464	Hemochromatosis, type 2B	Autosomal recessive
5	HARS1	142810	Usher syndrome, type 3B	Autosomal recessive
1	HAX1	605998	Neutropenia, severe congenital, type 3, autosomal recessive	Autosomal recessive
16	HBA1	141800	Alpha thalassemia	Autosomal recessive
16	HBA2	141850	Alpha thalassemia	Autosomal recessive
11	HBB	141900	HBB-related hemoglobinopathies	Autosomal recessive
X	HCFC1	300019	Mental retardation, X-linked 3 (methylmalonic acidemia and homocysteinemia, cblX type)	X-linked
10	HELLS	603946	Immunodeficiency-centromeric instability-facial anomalies syndrome 4	Autosomal recessive
11	HEPACAM	611642	Megalencephalic leukoencephalopathy with subcortical cysts 2A	Autosomal recessive
15	HERC1	605109	Macrocephaly, dysmorphic facies, and psychomotor retardation	Autosomal recessive
15	HERC2	605837	Mental retardation, autosomal recessive, type 38	Autosomal recessive
17	HES7	608059	Spondylocostal dysostosis, type 4, autosomal recessive	Autosomal recessive
3	HESX1	601802	Growth hormone deficiency with pituitary anomalies	Autosomal recessive
15	HEXA	606869	Tay-Sachs disease	Autosomal recessive
5	HEXB	606873	Sandhoff disease, infantile, juvenile, and adult forms	Autosomal recessive
6	HFE	613609	Hemochromatosis, type 1	Autosomal recessive
1	HFM1	615684	Premature ovarian failure 9	Autosomal recessive
3	HGD	607474	Alkaptonuria	Autosomal recessive
7	HGF	142409	Deafness, autosomal recessive, type 39	Autosomal recessive
8	HGSNAT	610453	Mucopolysaccharidosis type 3C (Sanfilippo syndrome C)	Autosomal recessive
2	HIBCH	610690	3-hydroxyisobutryl-CoA hydrolase deficiency	Autosomal recessive

11	HIKESHI	614908	Leukodystrophy, hypomyelinating, type 13	Autosomal recessive
5	HINT1	601314	Neuromyotonia and axonal neuropathy, autosomal recessive	Autosomal recessive
1	HJV	608374	Hemochromatosis, type 2A	Autosomal recessive
10	HK1	142600	Charcot-Marie-Tooth disease, type 4G	Autosomal recessive
21	HLCS	609018	Holocarboxylase synthetase deficiency	Autosomal recessive
1	HMGCL	613898	HMG-CoA lyase deficiency	Autosomal recessive
1	HMGCS2	600234	HMG-CoA synthase-2 deficiency	Autosomal recessive
22	HMOX1	141250	Heme oxygenase-1 deficiency	Autosomal recessive
4	HMX1	142992	Oculoauricular syndrome	Autosomal recessive
2	HNMT	605238	Mental retardation, autosomal recessive, type 51	Autosomal recessive
10	HOGA1	613597	Hyperoxaluria, primary, type 3	Autosomal recessive
7	HOXA1	142955	Athabaskan brainstem dysgenesis syndrome	Autosomal recessive
17	HOXB1	142968	Facial paresis, hereditary congenital, 3	Autosomal recessive
12	HOXC13	142976	Ectodermal dysplasia 9, hair/nail type	Autosomal recessive
1	HPCA	142622	Dystonia 2, torsion, autosomal recessive	Autosomal recessive
12	HPD	609695	Tyrosinemia, type 3	Autosomal recessive
4	HPGD	601688	Hypertrophic osteoarthropathy, primary, type 1 (pachydermoperiostosis)	Autosomal recessive
X	HPRT1	308000	Lesch-Nyhan syndrome	X-linked
10	HPS1	604982	Hermansky-Pudlak syndrome, type 1	Autosomal recessive
3	HPS3	606118	Hermansky-Pudlak syndrome, type 3	Autosomal recessive
22	HPS4	606682	Hermansky-Pudlak syndrome, type 4	Autosomal recessive
11	HPS5	607521	Hermansky-Pudlak syndrome, type 5	Autosomal recessive
10	HPS6	607522	Hermansky-Pudlak syndrome, type 6	Autosomal recessive
10	HPSE2	613469	Urofacial syndrome, type 1	Autosomal recessive
8	HR	602302	Alopecia universalis; Atrichia with papular lesions	Autosomal recessive
16	HSD11B2	614232	Apparent mineralocorticoid excess	Autosomal recessive
X	HSD17B10	300256	HSD10 mitochondrial disease	X-linked
9	HSD17B3	605573	46,XY disorder of sex development due to 17-beta-hydroxysteroid dehydrogenase 3 deficiency	Autosomal recessive
5	HSD17B4	601860	D-bifunctional protein deficiency	Autosomal recessive
1	HSD3B2	613890	Adrenal hyperplasia, congenital, due to 3-beta-hydroxysteroid dehydrogenase 2 deficiency	Autosomal recessive
16	HSD3B7	607764	Bile acid synthesis defect, congenital, type 1	Autosomal recessive
5	HSPA9	600548	Even-plus syndrome	Autosomal recessive
2	HSPD1	118190	Leukodystrophy, hypomyelinating, type 4	Autosomal recessive
1	HSPG2	142461	Schwartz-Jampel syndrome, type 1; Dyssegmental dysplasia, Silverman-Handmaker type	Autosomal recessive
10	HTRA1	602194	CARASIL syndrome	Autosomal recessive
2	HTRA2	606441	3-methylglutaconic aciduria, type 8	Autosomal recessive
3	HYAL1	607071	?Mucopolysaccharidosis, type 9	Autosomal recessive
7	HYC11	610531	Leukodystrophy, hypomyelinating, type 5	Autosomal recessive
16	HYDIN	610812	Ciliary dyskinesia, primary, type 5	Autosomal recessive
11	HYLS1	610693	Hydrolethalus syndrome	Autosomal recessive
9	IARS1	600709	Growth retardation, intellectual developmental disorder, hypotonia, and hepatopathy	Autosomal recessive
1	IBA57	615316	Multiple mitochondrial dysfunctions syndrome 3	Autosomal recessive
2	ICOS	604558	Immunodeficiency, common variable, 1	Autosomal recessive
20	IDH3B	604526	Retinitis pigmentosa, type 46	Autosomal recessive
X	IDS	300823	Mucopolysaccharidosis, type 2	X-linked
4	IDUA	252800	Mucopolysaccharidosis type 1	Autosomal recessive
18	IER3IP1	609382	Microcephaly, epilepsy, and diabetes syndrome	Autosomal recessive
6	IFNGR1	107470	Immunodeficiency, type 27A, mycobacteriosis	Autosomal recessive
21	IFNGR2	147569	Immunodeficiency, type 28, mycobacteriosis	Autosomal recessive

21	IFNGR2	147569	Immunodeficiency, type 28, mycobacteriosis	Autosomal recessive
3	IFT122	606045	Cranioectodermal dysplasia 1	Autosomal recessive
16	IFT140	614620	Retinitis pigmentosa, type 80; Short-rib thoracic dysplasia 9 with or without polydactyly	Autosomal recessive
2	IFT172	607386	Short-rib thoracic dysplasia 10 with or without polydactyly	Autosomal recessive
14	IFT43	614068	Short-rib thoracic dysplasia 18 with polydactyly	Autosomal recessive
20	IFT52	617094	Short-rib thoracic dysplasia 16 with or without polydactyly	Autosomal recessive
3	IFT80	611177	Short-rib thoracic dysplasia, type 2, with or without polydactyly	Autosomal recessive
12	IFT81	605489	Short-rib thoracic dysplasia 19 with or without polydactyly	Autosomal recessive
12	IGF1	147440	Growth retardation with deafness and mental retardation due to IGF1 deficiency	Autosomal recessive
15	IGF1R	147370	Insulin-like growth factor I, resistance to	Autosomal recessive*
16	IGFALS	601489	Acid-labile subunit deficiency	Autosomal recessive
4	IGFBP7	602867	Retinal arterial macroaneurysm with supravalvular pulmonic stenosis	Autosomal recessive
11	IGHMBP2	600502	Charcot-Marie-Tooth disease, axonal, type 2S	Autosomal recessive
22	IGLL1	146770	Agammaglobulinemia 2	Autosomal recessive
2	IHH	600726	Acrocapitofemoral dysplasia	Autosomal recessive
8	IKBKB	603258	Immunodeficiency, type 15	Autosomal recessive
11	IL10RA	146933	Inflammatory bowel disease, type 28, early onset, autosomal recessive	Autosomal recessive
21	IL10RB	123889	Inflammatory bowel disease, type 25, early onset, autosomal recessive	Autosomal recessive
9	IL11RA	600939	Craniosynostosis and dental anomalies	Autosomal recessive
5	IL12B	161561	Immunodeficiency, type 29, mycobacteriosis	Autosomal recessive
19	IL12RB1	601604	Immunodeficiency, type 30	Autosomal recessive
22	IL17RA	605461	Immunodeficiency, type 51	Autosomal recessive
3	IL17RC	610925	Candidiasis, familial, 9	Autosomal recessive
X	IL1RAPL1	300206	Mental retardation, X-linked, type 21/34	X-linked
2	IL1RN	147679	Sterile multifocal osteomyelitis with periostitis and pustulosis	Autosomal recessive
16	IL21R	605383	Immunodeficiency, type 56	Autosomal recessive
10	IL2RA	147730	Immunodeficiency, type 41, with lymphoproliferation and autoimmunity	Autosomal recessive
X	IL2RG	308380	Severe combined immunodeficiency, X-linked	X-linked
2	IL36RN	605507	Psoriasis, type 14, pustular	Autosomal recessive
5	IL7R	146661	Severe combined immunodeficiency, T-cell negative, B-cell/natural killer cell-positive type	Autosomal recessive
3	ILDR1	609739	Deafness, autosomal recessive, type 42	Autosomal recessive
8	IMPA1	602064	Mental retardation, autosomal recessive 59	Autosomal recessive
3	IMPG2	607056	Retinitis pigmentosa, type 56	Autosomal recessive
9	INPP5E	613037	Joubert syndrome, type 1	Autosomal recessive
17	INPP5K	607875	Muscular dystrophy, congenital, with cataracts and intellectual disability	Autosomal recessive
11	INPPL1	600829	Opsismodysplasia	Autosomal recessive
11	INS	176730	Permanent neonatal diabetes mellitus (PNDM)	Autosomal recessive*
19	INSR	147670	Diabetes mellitus, insulin-resistant, with acanthosis nigricans, type A	Autosomal recessive
7	INTS1	611345	Neurodevelopmental disorder with cataracts, poor growth, and dysmorphic facies	Autosomal recessive
9	INVS	243305	Nephronophthisis, type 2, infantile	Autosomal recessive
3	IQCB1	609237	Senior-Loken syndrome, type 5	Autosomal recessive
7	IQCE	617631	Polydactyly, postaxial, type A7	Autosomal recessive
12	IRAK4	606883	Immunodeficiency, type 67 (IRAK4 deficiency)	Autosomal recessive
16	IRF8	601565	Immunodeficiency, type 32B, monocyte and dendritic cell deficiency	Autosomal recessive
16	IRX5	606195	Hamamy syndrome	Autosomal recessive
9	ISCA1	611006	Multiple mitochondrial dysfunctions syndrome 5	Autosomal recessive
14	ISCA2	615317	Multiple mitochondrial dysfunctions syndrome 4	Autosomal recessive
12	ISCU	611911	Myopathy with lactic acidosis, hereditary	Autosomal recessive
1	ISG15	147571	Immunodeficiency, type 38	Autosomal recessive

20	ITCH	606409	Autoimmune disease, multisystem, with facial dysmorphism	Autosomal recessive
17	ITGA2B	607759	Glanzmann thrombasthenia	Autosomal recessive
17	ITGA3	605025	Interstitial lung disease, nephrotic syndrome, and epidermolysis bullosa, congenital	Autosomal recessive
2	ITGA6	147556	Epidermolysis bullosa, junctional, with pyloric stenosis	Autosomal recessive
12	ITGA7	600536	Muscular dystrophy, congenital, due to ITGA7 deficiency	Autosomal recessive
10	ITGA8	604063	Renal hypodysplasia/aplasia 1	Autosomal recessive
21	ITGB2	600065	Leukocyte adhesion deficiency	Autosomal recessive
17	ITGB3	173470	Glanzmann thrombasthenia	Autosomal recessive
17	ITGB4	147557	Epidermolysis bullosa, junctional, with pyloric atresia	Autosomal recessive
2	ITGB6	147558	Amelogenesis imperfecta, type 1H	Autosomal recessive
5	ITK	186973	Lymphoproliferative syndrome 1	Autosomal recessive
20	ITPA	147520	Epileptic encephalopathy, early infantile, type 35	Autosomal recessive
3	ITPR1	147265	Gillespie syndrome	Autosomal recessive*
15	IVD	607036	Isovaleric acidemia	Autosomal recessive
6	IYD	612025	Thyroid dyshormonogenesis, type 4	Autosomal recessive
3	JAGN1	616012	Neutropenia, severe congenital, 6, autosomal recessive	Autosomal recessive
19	JAK3	600173	Severe Combined Immunodeficiency, autosomal recessive, T-negative/B-positive type	Autosomal recessive
11	JAM3	606871	Hemorrhagic destruction of the brain, subependymal calcification, and cataracts	Autosomal recessive
17	JUP	173325	Naxos disease	Autosomal recessive
19	KANK2	614610	Nephrotic syndrome, type 16	Autosomal recessive
16	KARS1	601421	Deafness, autosomal recessive, type 89	Autosomal recessive
16	KATNB1	602703	Lissencephaly 6, with microcephaly	Autosomal recessive
16	KATNIP	616650	Joubert syndrome 26	Autosomal recessive
21	KCNE1	176261	Jervell and Lange-Nielsen syndrome 2	Autosomal recessive
11	KCNJ1	600359	Bartter syndrome, type 2	Autosomal recessive
1	KCNJ10	602208	SESAME syndrome	Autosomal recessive
11	KCNJ11	600937	Hyperinsulinemic hypoglycemia, type 2 (congenital hyperinsulinism); Permanent neonatal diabetes mellitus (PNDM)	Autosomal recessive; Autosomal recessive*
2	KCNJ13	603208	Leber congenital amaurosis, type 16	Autosomal recessive
9	KCNV2	607604	Retinal cone dystrophy, type 3B	Autosomal recessive
7	KCTD7	611725	Epilepsy, progressive myoclonic, type 3, with or without intracellular inclusions	Autosomal recessive
X	KDM5C	314690	Mental retardation, X-linked, syndromic, Claes-Jensen type	X-linked
18	KDSR	136440	Erythrokeratodermia variabilis et progressiva 4	Autosomal recessive
12	KERA	603288	Cornea plana 2, autosomal recessive	Autosomal recessive
6	KHDC3L	611687	Hydatidiform mole, recurrent, type 2	Autosomal recessive
14	KIAA0586	610178	Short-rib thoracic dysplasia 14 with polydactyly	Autosomal recessive
17	KIAA0753	617112	?Orofaciodigital syndrome, type 15	Autosomal recessive
7	KIAA1549	613344	Retinitis pigmentosa, type 86	Autosomal recessive
1	KIF14	611279	Microcephaly 20, primary, autosomal recessive; ?Meckel syndrome 12	Autosomal recessive
2	KIF1A	601255	Neuropathy, hereditary sensory, type 2C; Spastic paraplegia, type 30, autosomal recessive	Autosomal recessive
17	KIF1C	603060	Spastic ataxia 2, autosomal recessive	Autosomal recessive
15	KIF7	611254	Acrocallosal syndrome; Joubert syndrome, type 12	Autosomal recessive
10	KIFBP	609367	Goldberg-Shprintzen megacolon syndrome	Autosomal recessive
19	KISS1R	604161	Hypogonadotropic hypogonadism, type 8, with or without anosmia	Autosomal recessive
20	KIZ	615757	Retinitis pigmentosa 69	Autosomal recessive
5	KLHL3	605775	Pseudohypoaldosteronism, type 2D	Autosomal recessive
3	KLHL40	615340	Nemaline myopathy 8, autosomal recessive	Autosomal recessive
2	KLHL41	607701	Nemaline myopathy 9	Autosomal recessive
7	KLHL7	611119	Cold-induced sweating syndrome 3	Autosomal recessive
19	KLK4	603767	Amelogenesis imperfecta, type 2A1 (hypomaturation type)	Autosomal recessive

4	KLKB1	229000	Fletcher factor (prekallikrein) deficiency	Autosomal recessive
15	KNL1	609173	Microcephaly 4, primary, autosomal recessive	Autosomal recessive
19	KPTN	615620	Mental retardation, autosomal recessive 41	Autosomal recessive
22	KREMEN1	609898	Ectodermal dysplasia 13, hair/tooth type	Autosomal recessive
17	KRT10	148080	Epidermolytic hyperkeratosis	Autosomal recessive*
17	KRT14	148066	Epidermolysis bullosa simplex, autosomal recessive, type 1	Autosomal recessive
17	KRT25	616646	Woolly hair, autosomal recessive 3	Autosomal recessive
12	KRT5	148040	Epidermolysis bullosa simplex, autosomal recessive, type 1	Autosomal recessive
12	KRT85	602767	Ectodermal dysplasia 4, hair/nail type	Autosomal recessive
3	KY	605739	Myopathy, myofibrillar, type 7	Autosomal recessive
2	KYNU	605197	Vertebral, cardiac, renal, and limb defects syndrome, type 2	Autosomal recessive
X	L1CAM	308840	L1 Syndrome	X-linked
14	L2HGDH	609584	L-2-hydroxyglutaric aciduria	Autosomal recessive
18	LAMA1	150320	Poretti-Boltshauser syndrome	Autosomal recessive
6	LAMA2	156225	LAMA2-related muscular dystrophy	Autosomal recessive
18	LAMA3	600805	Junctional epidermolysis bullosa (JEB) Herlitz type; JEB non-Herlitz type	Autosomal recessive
7	LAMB1	150240	Lissencephaly, type 5	Autosomal recessive
3	LAMB2	150325	Pierson syndrome; Nephrotic syndrome, type 5, with or without ocular abnormalities	Autosomal recessive; Autosomal recessive
1	LAMB3	150310	Junctional epidermolysis bullosa (JEB) Herlitz type; JEB non-Herlitz type	Autosomal recessive
1	LAMC2	150292	Junctional epidermolysis bullosa (JEB) Herlitz type; JEB non-Herlitz type	Autosomal recessive
9	LAMC3	604349	Cortical malformations, occipital	Autosomal recessive
22	LARGE1	603590	Muscular dystrophy-dystroglycanopathy, type 6A and 6B	Autosomal recessive
4	LARP7	612026	Alazami syndrome	Autosomal recessive
5	LARS1	151350	?Infantile liver failure syndrome 1 (ILFS1)	Autosomal recessive
3	LARS2	604544	Perrault syndrome, type 4	Autosomal recessive
16	LAT	602354	Immunodeficiency, type 52	Autosomal recessive
1	LBR	600024	Greenberg skeletal dysplasia	Autosomal recessive
6	LCA5	611408	Leber congenital amaurosis, type 5	Autosomal recessive
16	LCAT	606967	Familial LCAT deficiency; Fish-eye disease	Autosomal recessive
1	LCK	153390	?Immunodeficiency, type 22	Autosomal recessive
2	LCT	603202	Lactase deficiency, congenital	Autosomal recessive
11	LDHA	150000	Glycogen storage disease type 11	Autosomal recessive
19	LDLR	606945	Hypercholesterolemia, familial, type 1	Autosomal recessive; Autosomal dominant
1	LDLRAP1	605747	Hypercholesterolemia, familial, autosomal recessive	Autosomal recessive
6	LEMD2	616312	Cataract 46, juvenile-onset	Autosomal recessive
7	LEP	164160	Obesity, morbid, due to leptin deficiency	Autosomal recessive
1	LEPR	601007	Obesity, morbid, due to leptin receptor deficiency	Autosomal recessive
19	LGI4	608303	Arthrogryposis multiplex congenita, neurogenic, with myelin defect	Autosomal recessive
19	LHB	152780	Hypogonadotropic hypogonadism, type 23, with or without anosmia	Autosomal recessive
2	LHCGR	152790	Leydig cell hypoplasia	Autosomal recessive
6	LHFPL5	609427	Deafness, autosomal recessive, type 67	Autosomal recessive
9	LHX3	600577	Pituitary hormone deficiency, combined, type 3	Autosomal recessive
4	LIAS	607031	Hyperglycinemia, lactic acidosis, and seizures	Autosomal recessive
5	LIFR	151443	Stuve-Wiedemann syndrome / Schwartz-Jampel type 2 syndrome	Autosomal recessive
13	LIG4	601837	LIG4 syndrome	Autosomal recessive
19	LIM2	154045	Cataract 19, multiple types	Autosomal recessive
15	LINS1	610350	Mental retardation, autosomal recessive, type 27	Autosomal recessive
10	LIPA	613497	Lysosomal acid lipase deficiency	Autosomal recessive

19	LIPE	151750	Lipodystrophy, familial partial, type 6	Autosomal recessive
3	LIPH	607365	Hypotrichosis, type 7 or woolly hair, autosomal recessive, type 2, with or without hypotrichosis	Autosomal recessive
10	LIPN	613924	Ichthyosis, congenital, autosomal recessive 8	Autosomal recessive
2	LIPT1	610284	Lipoyltransferase 1 deficiency	Autosomal recessive
11	LIPT2	617659	Encephalopathy, neonatal severe, with lactic acidosis and brain abnormalities	Autosomal recessive
18	LMAN1	601567	Combined deficiency of factor V and factor VIII, type 1	Autosomal recessive
6	LMBRD1	612625	Methylmalonic aciduria and homocystinuria, cblF type	Autosomal recessive
16	LMF1	611761	Lipase deficiency, combined	Autosomal recessive
3	LMOD3	616112	Nemaline myopathy 10	Autosomal recessive
19	LONP1	605490	CODAS syndrome	Autosomal recessive
18	LOXHD1	613072	Deafness, autosomal recessive, type 77	Autosomal recessive
13	LPAR6	609239	Hypotrichosis, type 8 or woolly hair, autosomal recessive, type 1, with or without hypotrichosis	Autosomal recessive
2	LPIN1	605518	Myoglobinuria, acute recurrent, autosomal recessive	Autosomal recessive
18	LPIN2	605519	Majeed syndrome	Autosomal recessive
8	LPL	609708	Lipoprotein lipase deficiency	Autosomal recessive
4	LRAT	604863	Leber congenital amaurosis type 14	Autosomal recessive
4	LRBA	606453	Immunodeficiency, common variable, 8, with autoimmunity	Autosomal recessive
1	LRIG2	608869	Urofacial syndrome 2	Autosomal recessive
4	LRIT3	615004	Night blindness, congenital stationary (complete), 1F, autosomal recessive	Autosomal recessive
10	LRMDA	614537	Albinism, oculocutaneous, type 7	Autosomal recessive
2	LRP2	600073	Donnai-Barrow syndrome	Autosomal recessive
11	LRP4	604270	Cenani-Lenz syndactyly syndrome	Autosomal recessive
11	LRP5	603506	Osteoporosis-pseudoglioma syndrome	Autosomal recessive
4	LRPAP1	104225	Myopia, type 23, autosomal recessive	Autosomal recessive
2	LRPPRC	607544	Leigh syndrome, French-Canadian type	Autosomal recessive
9	LRSAM1	610933	Charcot-Marie-Tooth disease, axonal, type 2P	Autosomal recessive
11	LRTOMT	612414	Deafness, autosomal recessive, type 63	Autosomal recessive
21	LSS	600909	Alopecia-intellectual disability syndrome 4; Cataract 44; Hypotrichosis 14	Autosomal recessive
14	LTBP2	602091	Microspherophakia and/or megalocornea, with ectopia lentis and with or without secondary glaucoma	Autosomal recessive
11	LTBP3	602090	Dental anomalies and short stature	Autosomal recessive
19	LTBP4	604710	Cutis laxa, autosomal recessive, type 1C	Autosomal recessive
5	LYRM7	615831	Mitochondrial complex III deficiency, nuclear type 8	Autosomal recessive
1	LYST	606897	Chediak-Higashi syndrome	Autosomal recessive
3	LZTFL1	606568	Bardet-Biedl syndrome, type 17	Autosomal recessive
22	LZTR1	600574	Noonan syndrome, type 2	Autosomal recessive
19	MAG	159460	Spastic paraplegia, type 75, autosomal recessive	Autosomal recessive
7	MAGI2	606382	Nephrotic syndrome, type 15	Autosomal recessive
6	MAK	154235	Retinitis pigmentosa type 62	Autosomal recessive
18	MALT1	604860	Immunodeficiency, type 12	Autosomal recessive
9	MAN1B1	604346	Mental retardation, autosomal recessive, type 15	Autosomal recessive
19	MAN2B1	609458	Alpha-mannosidosis	Autosomal recessive
4	MANBA	609489	Mannosidosis, beta	Autosomal recessive
2	MAP3K20	609479	Centronuclear myopathy, type 6, with fiber-type disproportion	Autosomal recessive
15	MAPKBP1	616786	Nephronophthisis 20	Autosomal recessive
17	MAPT	157140	Supranuclear palsy, progressive atypical (parkinsonism syndrome)	Autosomal recessive
12	MARS1	156560	Interstitial lung and liver disease	Autosomal recessive
2	MARS2	609728	Spastic ataxia, type 3, autosomal recessive	Autosomal recessive
5	MARVELD2	610572	Deafness, autosomal recessive, type 49	Autosomal recessive
3	MASP1	600521	3MC syndrome 1	Autosomal recessive

10	MAT1A	610550	Methionine adenosyltransferase deficiency, autosomal recessive	Autosomal recessive
2	MATN3	602109	?Spondyloepimetaphyseal dysplasia	Autosomal recessive
19	MBOAT7	606048	Mental retardation, autosomal recessive 57	Autosomal recessive
18	MC2R	607397	Glucocorticoid deficiency, due to ACTH unresponsiveness	Autosomal recessive
3	MCCC1	609010	3-Methylcrotonyl-CoA carboxylase deficiency, type 1	Autosomal recessive
5	MCCC2	609014	3-Methylcrotonyl-CoA carboxylase deficiency, type 2	Autosomal recessive
2	MCEE	608419	Methylmalonyl-CoA epimerase deficiency	Autosomal recessive
2	MCFD2	607788	Combined deficiency of factor V and factor VIII, type 2	Autosomal recessive
5	MCIDAS	614086	Ciliary dyskinesia, primary, type 42	Autosomal recessive
21	MCM3AP	603294	Peripheral neuropathy, autosomal recessive, with or without impaired intellectual development	Autosomal recessive
8	MCM4	602638	Immunodeficiency, type 54	Autosomal recessive
6	MCM9	610098	Ovarian dysgenesis 4	Autosomal recessive
19	MCOLN1	605248	Mucolipidosis type 4	Autosomal recessive
8	MCPH1	607117	Microcephaly type 1, primary, autosomal recessive	Autosomal recessive
7	MDH2	154100	Epileptic encephalopathy, early infantile, 51	Autosomal recessive
X	MECP2	300005	Encephalopathy, neonatal severe; Rett syndrome	X-linked
1	MECR	608205	Dystonia, childhood-onset, with optic atrophy and basal ganglia abnormalities	Autosomal recessive
11	MED17	603810	Microcephaly, postnatal progressive, with seizures and brain atrophy	Autosomal recessive
6	MED23	605042	Mental retardation, autosomal recessive, type 18	Autosomal recessive
19	MED25	610197	Basel-Vanagait-Smirin-Yosef syndrome	Autosomal recessive
16	MEFV	608107	Familial Mediterranean fever	Autosomal recessive
5	MEGF10	612453	Myopathy, areflexia, respiratory distress, and dysphagia, early-onset	Autosomal recessive
19	MEGF8	604267	Carpenter syndrome, type 2	Autosomal recessive
17	MEOX1	600147	Klippel-Feil syndrome 2	Autosomal recessive
2	MERTK	604705	Retinitis pigmentosa type 38	Autosomal recessive
15	MESP2	605195	Spondylocostal dysostosis, type 2, autosomal recessive	Autosomal recessive
17	METTL23	615262	Mental retardation, autosomal recessive 44	Autosomal recessive
2	MFF	614785	Encephalopathy due to defective mitochondrial and peroxisomal fission, type 2	Autosomal recessive
1	MFN2	608507	Charcot-Marie-Tooth disease, axonal, type 2A2B	Autosomal recessive
11	MFRP	606227	Microphthalmia, isolated type 5	Autosomal recessive
1	MFSD2A	614397	Microcephaly 15, primary, autosomal recessive	Autosomal recessive
4	MFSD8	611124	Ceroid lipofuscinosis, neuronal, type 7	Autosomal recessive
14	MGAT2	602616	Congenital disorder of glycosylation, type 2a	Autosomal recessive
20	MGME1	615076	Mitochondrial DNA depletion syndrome 11	Autosomal recessive
12	MGP	154870	Keutel syndrome	Autosomal recessive
10	MICU1	605084	Myopathy with extrapyramidal signs	Autosomal recessive
X	MID1	300552	Opitz GBBB syndrome, type 1	X-linked
13	MIPEP	602241	Combined oxidative phosphorylation deficiency 31	Autosomal recessive
3	MITF	156845	COMMAD syndrome	Autosomal recessive
20	MKKS	604896	Bardet-Biedl syndrome type 6	Autosomal recessive
17	MKS1	609883	Bardet-Biedl syndrome type 13; Meckel syndrome, type 1; Joubert syndrome, type 28	Autosomal recessive
22	MLC1	605908	Megalencephalic leukoencephalopathy with subcortical cysts	Autosomal recessive
2	MLPH	606526	Griscelli syndrome, type 3	Autosomal recessive
16	MLYCD	606761	Malonyl-CoA decarboxylase deficiency	Autosomal recessive
4	MMAA	607481	Methylmalonic aciduria, vitamin B12-responsive	Autosomal recessive
12	MMAB	607568	Methylmalonic aciduria, vitamin B12-responsive, type cblB	Autosomal recessive
1	MMACHC	609831	Methylmalonic aciduria and homocystinuria, cblC type	Autosomal recessive; digenic inheritance (PRDX1 gene)
2	MMADHC	611935	Homocystinuria, cblD type, variant 1	Autosomal recessive

3	MME	120520	Charcot-Marie-Tooth disease, axonal, type 2T	Autosomal recessive*
11	MMP13	600108	Metaphyseal dysplasia, Spahr type	Autosomal recessive
16	MMP2	120360	Multicentric osteolysis, nodulosis, and arthropathy (MONA)	Autosomal recessive
11	MMP20	604629	Amelogenesis imperfecta, type 2A2 (hypomaturation type)	Autosomal recessive
10	MMP21	608416	Heterotaxy, visceral, 7, autosomal	Autosomal recessive
6	MMUT	609058	Methylmalonic aciduria, mut(0) type	Autosomal recessive
18	MOCOS	613274	Xanthinuria, type 2	Autosomal recessive
6	MOCS1	603707	Molybdenum cofactor deficiency A	Autosomal recessive
5	MOCS2	603708	Molybdenum cofactor deficiency B	Autosomal recessive
2	MOGS	601336	Congenital disorder of glycosylation, type 2B	Autosomal recessive
6	MPC1	614738	Mitochondrial pyruvate carrier deficiency	Autosomal recessive
17	MPDU1	604041	Congenital disorder of glycosylation, type 1F	Autosomal recessive
9	MPDZ	603785	Hydrocephalus, congenital, type 2, with or without brain or eye anomalies	Autosomal recessive
15	MPI	154550	Congenital disorder of glycosylation, type 1B	Autosomal recessive
6	MPIG6B	606520	Thrombocytopenia, anemia, and myelofibrosis	Autosomal recessive
1	MPL	159530	Thrombocytopenia, congenital amegakaryocytic	Autosomal recessive
7	MPLKIP	609188	Trichothiodystrophy, type 4, nonphotosensitive	Autosomal recessive
17	MPO	606989	Myeloperoxidase deficiency	Autosomal recessive
2	MPV17	137960	Mitochondrial DNA depletion syndrome type 6 (hepatocerebral); Charcot-Marie-Tooth disease, axonal, type 2EE	Autosomal recessive
1	MPZ	159440	Dejerine-Sottas disease	Autosomal recessive*
21	MRAP	609196	Glucocorticoid deficiency, type 2	Autosomal recessive
11	MRE11	600814	Ataxia-telangiectasia-like disorder 1	Autosomal recessive
10	MRPS16	609204	Combined oxidative phosphorylation deficiency 2	Autosomal recessive
3	MRPS22	605810	Combined oxidative phosphorylation deficiency type 5	Autosomal recessive
16	MRPS34	611994	Combined oxidative phosphorylation deficiency 32	Autosomal recessive
5	MSH3	600887	Familial adenomatous polyposis, type 4	Autosomal recessive
4	MSMO1	607545	Microcephaly, congenital cataract, and psoriasiform dermatitis	Autosomal recessive
12	MSRB3	613719	Deafness, autosomal recessive, type 74	Autosomal recessive
1	MSTO1	617619	Myopathy, mitochondrial, and ataxia	Autosomal recessive*
15	MTFMT	611766	Combined oxidative phosphorylation deficiency 15	Autosomal recessive
14	MTHFD1	172460	Combined immunodeficiency and megaloblastic anemia with or without hyperhomocysteinemia	Autosomal recessive
1	MTHFR	607093	Homocystinuria due to MTHFR deficiency	Autosomal recessive
X	MTM1	300415	Myotubular myopathy, X-linked	X-linked
11	MTMR2	603557	Charcot-Marie-Tooth disease, type 4B1	Autosomal recessive
6	MTO1	614667	Combined oxidative phosphorylation deficiency 10	Autosomal recessive
1	MTR	156570	Homocystinuria-megaloblastic anemia, cblG complementation type	Autosomal recessive
12	MTRFR	613541	Combined oxidative phosphorylation deficiency 7; Spastic paraplegia, type 55, autosomal recessive	Autosomal recessive
5	MTRR	602568	Homocystinuria-megaloblastic anemia, cbl E type	Autosomal recessive
4	MTTP	157147	Abetalipoproteinemia	Autosomal recessive
9	MUSK	601296	Fetal akinesia deformation sequence, type 1; Myasthenic syndrome, congenital, type 9, associated with acetylcholine receptor deficiency	Autosomal recessive
1	MUTYH	604933	Adenomas, multiple colorectal	Autosomal recessive
12	MVK	251170	Mevalonic aciduria	Autosomal recessive
12	MYBPC1	160794	Lethal congenital contracture syndrome, type 4	Autosomal recessive
3	MYD88	602170	Immunodeficiency, type 68	Autosomal recessive
17	MYH2	160740	Proximal myopathy and ophthalmoplegia	Autosomal recessive
9	MYMK	615345	Carey-Fineman-Ziter syndrome	Autosomal recessive
17	MYO15A	602666	Deafness, autosomal recessive, type 3	Autosomal recessive
22	MYO18B	607295	Klippel-Feil syndrome, type 4, autosomal recessive, with myopathy and facial dysmorphism	Autosomal recessive
15	MYO1E	601479	Glomerulosclerosis, focal segmental, 6	Autosomal recessive

10	MYO3A	606808	Deafness, autosomal recessive, type 30	Autosomal recessive
15	MYO5A	160777	Griscelli syndrome, type 1	Autosomal recessive
18	MYO5B	606540	Microvillus inclusion disease	Autosomal recessive
6	MYO6	600970	Deafness, autosomal recessive, type 37	Autosomal recessive
11	MYO7A	276903	Usher syndrome, type 1B; Deafness, autosomal recessive, type 2	Autosomal recessive
10	MYPN	608517	Nemaline myopathy, type 11, autosomal recessive	Autosomal recessive
2	NADK2	615787	2,4-dienoyl-CoA reductase deficiency	Autosomal recessive
22	NAGA	104170	Schindler disease, type I; Schindler disease, type III; Kanzaki disease	Autosomal recessive
17	NAGLU	609701	Mucopolysaccharidosis, type 3B (Sanfilippo B)	Autosomal recessive
17	NAGS	608300	N-acetylglutamate synthase deficiency	Autosomal recessive
13	NALCN	611549	Hypotonia, infantile, with psychomotor retardation and characteristic facies 1	Autosomal recessive
9	NANS	605202	Spondyloepimetaphyseal dysplasia, Camera-Genevieve type	Autosomal recessive
11	NARS2	612803	Combined oxidative phosphorylation deficiency 24	Autosomal recessive
1	NAXE	608862	Encephalopathy, progressive, early-onset, with brain edema and/or leukoencephalopathy	Autosomal recessive
2	NBAS	608025	Infantile liver failure syndrome, type 2; Short stature, optic nerve atrophy, and Pelger-Huet anomaly	Autosomal recessive
3	NBEAL2	614169	Gray platelet syndrome	Autosomal recessive
8	NBN	602667	Nijmegen breakage syndrome	Autosomal recessive
11	NCAPD3	609276	Microcephaly 22, primary, autosomal recessive	Autosomal recessive
7	NCF1	608512	Chronic granulomatous disease, type 1	Autosomal recessive
1	NCF2	608515	Chronic granulomatous disease, type 2	Autosomal recessive
22	NCF4	601488	Chronic granulomatous disease, type 3	Autosomal recessive
16	NDE1	609449	Lissencephaly, type 4 (with microcephaly)	Autosomal recessive
X	NDP	300658	Norrie disease	X-linked
8	NDRG1	605262	Charcot-Marie-Tooth disease, type 4D	Autosomal recessive
5	NDST1	600853	Mental retardation, autosomal recessive, type 46	Autosomal recessive
2	NDUFA10	603835	Mitochondrial complex I deficiency, nuclear type 22	Autosomal recessive
19	NDUFA11	612638	Mitochondrial complex I deficiency, nuclear type 14	Autosomal recessive
12	NDUFA12	614530	?Mitochondrial complex I deficiency, nuclear type 23	Autosomal recessive
5	NDUFA2	602137	Mitochondrial complex I deficiency, nuclear type 13	Autosomal recessive
12	NDUFA9	603834	Mitochondrial complex I deficiency, nuclear type 26	Autosomal recessive
15	NDUFAF1	606934	Mitochondrial complex I deficiency, nuclear type 11	Autosomal recessive
5	NDUFAF2	609653	Mitochondrial complex I deficiency, nuclear type 10	Autosomal recessive
3	NDUFAF3	612911	Mitochondrial complex I deficiency, nuclear type 18	Autosomal recessive
20	NDUFAF5	612360	Mitochondrial complex I deficiency, nuclear type 16	Autosomal recessive
8	NDUFAF6	612392	Mitochondrial complex I deficiency, nuclear type 17	Autosomal recessive
2	NDUFB3	603839	Mitochondrial complex I deficiency, nuclear type 25	Autosomal recessive
8	NDUFB9	601445	Mitochondrial complex I deficiency, nuclear type 24	Autosomal recessive
2	NDUFS1	157655	Mitochondrial complex I deficiency, nuclear type 5	Autosomal recessive
1	NDUFS2	602985	Mitochondrial complex I deficiency, nuclear type 6	Autosomal recessive
11	NDUFS3	603846	Mitochondrial complex I deficiency, nuclear type 8	Autosomal recessive
5	NDUFS4	602694	Mitochondrial complex I deficiency, nuclear type 1	Autosomal recessive
5	NDUFS6	603848	Mitochondrial complex I deficiency, nuclear type 9	Autosomal recessive
19	NDUFS7	601825	Mitochondrial complex I deficiency, nuclear type 3	Autosomal recessive
11	NDUFS8	602141	Mitochondrial complex I deficiency, nuclear type 2	Autosomal recessive
11	NDUFV1	161015	Mitochondrial complex I deficiency, nuclear type 4	Autosomal recessive
18	NDUFV2	600532	Mitochondrial complex I deficiency, nuclear type 7	Autosomal recessive
2	NEB	161650	Nemaline myopathy type 2	Autosomal recessive
11	NECTIN1	600644	Cleft lip/palate-ectodermal dysplasia syndrome; Orofacial cleft 7	Autosomal recessive

1	NECTIN4	609607	Ectodermal dysplasia-syndactyly syndrome, type 1	Autosomal recessive
8	NEFL	162280	Charcot-Marie-Tooth disease, type 1F	Autosomal recessive
4	NEK1	604588	Short-rib thoracic dysplasia, type 6, with or without polydactyly	Autosomal recessive
17	NEK8	609799	Renal-hepatic-pancreatic dysplasia, type 2	Autosomal recessive
14	NEK9	609798	Lethal congenital contracture syndrome 10	Autosomal recessive
6	NEU1	608272	Sialidosis, type 1 and type 2	Autosomal recessive
10	NEUROG3	604882	Diarrhea 4, malabsorptive, congenital	Autosomal recessive
2	NFU1	608100	Multiple mitochondrial dysfunctions syndrome 1	Autosomal recessive
1	NGF	162030	Neuropathy, hereditary sensory and autonomic, type 5	Autosomal recessive
3	NGLY1	610661	Congenital disorder of deglycosylation	Autosomal recessive
2	NHEJ1	611290	Severe combined immunodeficiency with microcephaly, growth retardation, and sensitivity to ionizing radiation	Autosomal recessive
6	NHLRC1	608072	Epilepsy, progressive myoclonic, type 2B (Lafora)	Autosomal recessive
5	NHP2	606470	Dyskeratosis congenita, autosomal recessive type 2	Autosomal recessive
14	NIN	608684	Seckel syndrome, type 7	Autosomal recessive
5	NIPAL4	609383	Ichthyosis, congenital, autosomal recessive, type 6	Autosomal recessive
8	NKX2-6	611770	Conotruncal heart malformations	Autosomal recessive
4	NKX3-2	602183	Spondylo-megaepiphyseal-metaphyseal dysplasia	Autosomal recessive
10	NKX6-2	605955	Spastic ataxia 8, autosomal recessive, with hypomyelinating leukodystrophy	Autosomal recessive
17	NLRP1	606636	Autoinflammation with arthritis and dyskeratosis	Autosomal recessive*
19	NLRP7	609661	Hydatidiform mole, recurrent, type 1	Autosomal recessive
7	NME8	607421	Ciliary dyskinesia, primary, type 6	Autosomal recessive
1	NMNAT1	608700	Leber congenital amaurosis 9; Spondyloepiphyseal dysplasia, sensorineural hearing loss, intellectual developmental disorder, and Leber congenital amaurosis	Autosomal recessive
5	NNT	607878	Glucocorticoid deficiency 4, with or without mineralocorticoid deficiency	Autosomal recessive
15	NOP10	606471	Dyskeratosis congenita, autosomal recessive type 1	Autosomal recessive
18	NPC1	607623	Niemann-Pick disease, type C1	Autosomal recessive
14	NPC2	601015	Niemann-pick disease, type C2	Autosomal recessive
2	NPHP1	607100	Joubert syndrome type 4	Autosomal recessive
3	NPHP3	608002	Meckel syndrome type 7	Autosomal recessive
1	NPHP4	607215	Nephronophthisis type 4	Autosomal recessive
19	NPHS1	602716	Nephrotic syndrome, type 1	Autosomal recessive
1	NPHS2	604766	Nephrotic syndrome, type 2	Autosomal recessive
9	NPR2	108961	Acromesomelic dysplasia, Maroteaux type	Autosomal recessive
X	NR0B1	300473	Adrenal hypoplasia, congenital	X-linked
12	NR1H4	603826	Cholestasis, progressive familial intrahepatic, type 5	Autosomal recessive
15	NR2E3	604485	Enhanced S-cone syndrome (Goldmann-Favre); Retinitis pigmentosa, type 37	Autosomal recessive; Autosomal recessive*
14	NRL	162080	Retinal degeneration, autosomal recessive, clumped pigment type	Autosomal recessive*
2	NRXN1	600565	Pitt-Hopkins-like syndrome, type 2	Autosomal recessive
8	NSMCE2	617246	Seckel syndrome, type 10	Autosomal recessive
5	NSUN2	610916	Mental retardation, autosomal recessive, type 5	Autosomal recessive
10	NT5C2	600417	Spastic paraplegia, type 45, autosomal recessive	Autosomal recessive
7	NT5C3A	606224	Anemia, hemolytic, due to UMPH1 deficiency	Autosomal recessive
6	NT5E	129190	Calcification of joints and arteries	Autosomal recessive
16	NTHL1	602656	Familial adenomatous polyposis, type 3	Autosomal recessive
1	NTRK1	191315	Insensitivity to pain, congenital, with anhidrosis	Autosomal recessive
14	NUBPL	613621	Mitochondrial complex I deficiency, nuclear type 21	Autosomal recessive
12	NUP107	607617	Nephrotic syndrome, type 11	Autosomal recessive
19	NUP62	605815	Striatonigral degeneration, infantile	Autosomal recessive
16	NUP93	614351	Nephrotic syndrome, type 12	Autosomal recessive
10	OAT	613349	Gyrate atrophy of choroid and retina	Autosomal recessive

2	OBSL1	610991	3M syndrome 2	Autosomal recessive
15	OCA2	611409	Oculocutaneous albinism type 2	Autosomal recessive
5	OCLN	602876	Pseudo-TORCH syndrome, type 1	Autosomal recessive
X	OCRL	300535	Lowe Syndrome; Dent disease type 2	X-linked
19	ODAD1	615038	Ciliary dyskinesia, primary, type 20	Autosomal recessive
10	ODAD2	615408	Ciliary dyskinesia, primary, type 23	Autosomal recessive
19	ODAD3	615956	Ciliary dyskinesia, primary, type 30	Autosomal recessive
3	OPA1	605290	Behr syndrome	Autosomal recessive
19	OPA3	606580	3-methylglutaconic aciduria, type 3	Autosomal recessive
X	OPHN1	300127	Mental retardation, X-linked, with cerebellar hypoplasia and distinctive facial appearance	X-linked
10	OPTN	602432	Amyotrophic lateral sclerosis, type 12	Autosomal recessive
12	ORAI1	610277	Immunodeficiency, type 9	Autosomal recessive
1	ORC1	601902	Meier-Gorlin syndrome, type 1	Autosomal recessive
2	ORC4	603056	Meier-Gorlin syndrome, type 2	Autosomal recessive
16	ORC6	607213	Meier-Gorlin syndrome, type 3	Autosomal recessive
14	OSGEP	610107	Galloway-Mowat syndrome 3	Autosomal recessive
6	OSTM1	607649	Osteopetrosis, autosomal recessive type 5	Autosomal recessive
X	OTC	300461	Ornithine transcarbamylase deficiency	X-linked
16	OTOA	607038	Deafness, autosomal recessive, type 22	Autosomal recessive
2	OTOF	603681	Deafness, autosomal recessive, type 9	Autosomal recessive
11	OTOG	604487	Deafness, autosomal recessive, type 18B	Autosomal recessive
12	OTOGL	614925	Deafness, autosomal recessive, type 84B	Autosomal recessive
8	OTUD6B	612021	Intellectual developmental disorder with dysmorphic facies, seizures, and distal limb anomalies	Autosomal recessive
5	OTULIN	615712	Autoinflammation, panniculitis, and dermatosis syndrome	Autosomal recessive
5	OXCT1	601424	Succinyl CoA:3-oxoacid CoA transferase deficiency	Autosomal recessive
3	P2RY12	600515	Bleeding disorder, platelet-type, type 8	Autosomal recessive
1	P3H1	610339	Osteogenesis imperfecta, type 8	Autosomal recessive
3	P3H2	610341	Myopia, high, with cataract and vitreoretinal degeneration	Autosomal recessive
1	PADI6	610363	Preimplantation embryonic lethality 2	Autosomal recessive
12	PAH	612349	Phenylketonuria	Autosomal recessive
X	PAK3	300142	Mental retardation, X-linked, type 30	X-linked
16	PALB2	610355	PALB2-related conditions	Autosomal recessive
16	PAM16	614336	Spondylometaphyseal dysplasia, Megarbane-Dagher-Melike type	Autosomal recessive
20	PANK2	606157	Neurodegeneration with brain iron accumulation type 1	Autosomal recessive
10	PAPSS2	603005	Brachyolmia, type 4, with mild epiphyseal and metaphyseal changes	Autosomal recessive
1	PARK7	602533	Parkinson disease, type 7, autosomal recessive, early-onset	Autosomal recessive
16	PARN	604212	Dyskeratosis congenita, autosomal recessive 6	Autosomal recessive
15	PATL2	614661	Oocyte maturation defect 4	Autosomal recessive
1	PAX7	167410	Rhabdomyosarcoma 2, alveolar	Autosomal recessive
11	PC	608786	Pyruvate carboxylase deficiency	Autosomal recessive
2	PCARE	613425	Retinitis pigmentosa, type 54	Autosomal recessive
10	PCBD1	126090	Hyperphenylalaninemia, BH4-deficient, type D	Autosomal recessive
13	PCCA	232000	Propionic acidemia	Autosomal recessive
3	PCCB	232050	Propionic acidemia	Autosomal recessive
5	PCDH12	605622	Microcephaly, seizures, spasticity, and brain calcification	Autosomal recessive
10	PCDH15	605514	Deafness, autosomal recessive, type 23; Usher syndrome, type 1D/F digenic	Autosomal recessive
14	PCK2	614095	PEPCK deficiency, mitochondrial	Autosomal recessive
21	PCNT	605925	Microcephalic osteodysplastic primordial dwarfism, type 2	Autosomal recessive

5	PCSK1	162150	Obesity with impaired prohormone processing	Autosomal recessive
3	PCYT1A	123695	Spondylometaphyseal dysplasia with cone-rod dystrophy	Autosomal recessive
6	PDE10A	610652	Dyskinesia, limb and orofacial, infantile-onset	Autosomal recessive
5	PDE6A	180071	Retinitis pigmentosa type 43	Autosomal recessive
4	PDE6B	180072	Retinitis pigmentosa type 40	Autosomal recessive
10	PDE6C	600827	Cone dystrophy type 4	Autosomal recessive
17	PDE6G	180073	Retinitis pigmentosa type 57	Autosomal recessive
12	PDE6H	601190	Retinal cone dystrophy 3 and achromatopsia 6	Autosomal recessive*
X	PDHA1	300502	Pyruvate dehydrogenase E1-alpha deficiency	X-linked
3	PDHB	179060	Pyruvate dehydrogenase E1-beta deficiency	Autosomal recessive
11	PDHX	608769	Lacticacidemia due to PDX1 deficiency	Autosomal recessive
8	PDP1	605993	Pyruvate dehydrogenase phosphatase deficiency	Autosomal recessive
10	PDSS1	607429	Coenzyme Q10 deficiency, primary, type 2	Autosomal recessive
6	PDSS2	610564	Coenzyme Q10 deficiency, primary, type 3	Autosomal recessive
13	PDX1	600733	Pancreatic agenesis type 1	Autosomal recessive
21	PDXK	179020	Neuropathy, hereditary motor and sensory, type VIC, with optic atrophy	Autosomal recessive
10	PDZD7	612971	Deafness, autosomal recessive, type 57; Usher syndrome, type 2C, digenic	Autosomal recessive; Digenic inheritance (ADGRV1 gene)
19	PEPD	613230	Prolidase deficiency	Autosomal recessive
19	PET100	614770	Mitochondrial complex IV deficiency, nuclear type 12	Autosomal recessive
7	PEX1	602136	Heimler syndrome 1; Peroxisome biogenesis disorder 1A (Zellweger); Peroxisome biogenesis disorder 1B (NALD/IRD)	Autosomal recessive
1	PEX10	602859	Peroxisome biogenesis disorder, type 6A (Zellweger syndrome); Peroxisome biogenesis disorder, type 6B	Autosomal recessive
1	PEX11B	603867	?Peroxisome biogenesis disorder 14B	Autosomal recessive
17	PEX12	601758	Peroxisome biogenesis disorder type 3A (Zellweger)	Autosomal recessive
2	PEX13	601789	Peroxisome biogenesis disorder, type 11A (Zellweger syndrome); Peroxisome biogenesis disorder, type 11B	Autosomal recessive
1	PEX14	601791	Peroxisome biogenesis disorder, type 13A (Zellweger syndrome)	Autosomal recessive
11	PEX16	603360	Peroxisome biogenesis disorder, type 8A (Zellweger syndrome); Peroxisome biogenesis disorder, type 8B	Autosomal recessive
1	PEX19	600279	Peroxisome biogenesis disorder, type 12A (Zellweger syndrome)	Autosomal recessive
8	PEX2	170993	Peroxisome biogenesis disorder type 5A (Zellweger)	Autosomal recessive
22	PEX26	608666	Peroxisome biogenesis disorder type 7A (Zellweger)	Autosomal recessive
6	PEX3	603164	Peroxisome biogenesis disorder, type 10A (Zellweger syndrome)	Autosomal recessive
12	PEX5	600414	Peroxisome biogenesis disorder type 2A (Zellweger)	Autosomal recessive
6	PEX6	601498	Peroxisome biogenesis disorder, type 4A (Zellweger syndrome); Peroxisome biogenesis disorder, type 4B; Heimler syndrome 2	Autosomal recessive; Autosomal recessive*; Autosomal recessive
6	PEX7	601757	Rhizomelic chondrodysplasia punctata, type 1	Autosomal recessive
12	PFKM	610681	Glycogen storage disease, type 7	Autosomal recessive
7	PGAM2	612931	Glycogen storage disease X	Autosomal recessive
2	PGAP1	611655	Mental retardation, autosomal recessive 42	Autosomal recessive
11	PGAP2	615187	Hyperphosphatasia with mental retardation syndrome 3	Autosomal recessive
17	PGAP3	611801	Hyperphosphatasia with mental retardation syndrome 4	Autosomal recessive
X	PGK1	311800	Phosphoglycerate kinase 1 deficiency	X-linked
1	PGM1	171900	Congenital disorder of glycosylation, type 1t	Autosomal recessive
6	PGM3	172100	Immunodeficiency, type 23	Autosomal recessive
X	PHF8	300560	Mental retardation syndrome, X-linked, Siderius type	X-linked
1	PHGDH	606879	Neu-Laxova syndrome, type 1; Phosphoglycerate dehydrogenase deficiency	Autosomal recessive
16	PHKB	172490	Glycogen storage disease, type 9B	Autosomal recessive
16	PHKG2	172471	Glycogen storage disease type 9c	Autosomal recessive
11	PHOX2A	602753	Fibrosis of extraocular muscles, congenital, 2	Autosomal recessive
10	PHYH	602026	Refsum disease	Autosomal recessive
22	PI4KA	600286	Polymicrogyria, perisylvian, with cerebellar hypoplasia and arthrogryposis	Autosomal recessive
13	PIBF1	607532	Joubert syndrome 33	Autosomal recessive

16	PIEZO1	611184	Lymphedema, hereditary, type 3	Autosomal recessive
18	PIEZO2	613629	Arthrogryposis, distal, with impaired proprioception and touch	Autosomal recessive
1	PIGC	601730	Glycosylphosphatidylinositol biosynthesis defect 16	Autosomal recessive
4	PIGG	616918	Mental retardation, autosomal recessive 53	Autosomal recessive
17	PIGL	605947	Zunich neuroectodermal syndrome	Autosomal recessive
1	PIGM	610273	Glycosylphosphatidylinositol deficiency	Autosomal recessive
18	PIGN	606097	Multiple congenital anomalies-hypotonia-seizures syndrome, type 1	Autosomal recessive
9	PIGO	614730	Hyperphosphatasia with mental retardation syndrome 2	Autosomal recessive
20	PIGT	610272	Multiple congenital anomalies-hypotonia-seizures syndrome 3	Autosomal recessive
1	PIGV	610274	Hyperphosphatasia with mental retardation syndrome 1	Autosomal recessive
17	PIGW	610275	Glycosylphosphatidylinositol biosynthesis defect 11	Autosomal recessive
4	PIGY	610662	Hyperphosphatasia with mental retardation syndrome 6	Autosomal recessive
1	PINK1	608309	Parkinson disease, type 6, early onset	Autosomal recessive
19	PIP5K1C	606102	Lethal congenital contractural syndrome, type 3	Autosomal recessive
2	PJVK	610219	Deafness, autosomal recessive, type 59	Autosomal recessive
7	PKD1L1	609721	Heterotaxy, visceral, 8, autosomal	Autosomal recessive
6	PKHD1	606702	Polycystic kidney disease type 4	Autosomal recessive
1	PKLR	609712	Pyruvate kinase deficiency	Autosomal recessive
1	PKP1	601975	Ectodermal dysplasia/skin fragility syndrome	Autosomal recessive
22	PLA2G6	603604	Infantile neuroaxonal dystrophy type 1	Autosomal recessive
9	PLAA	603873	Neurodevelopmental disorder with progressive microcephaly, spasticity, and brain anomalies	Autosomal recessive
20	PLCB1	607120	Epileptic encephalopathy, early infantile, type 12	Autosomal recessive
20	PLCB4	600810	Auriculocondylar syndrome, type 2	Autosomal recessive*
3	PLCD1	602142	Nail disorder, nonsyndromic congenital, type 3 (leukonychia)	Autosomal recessive
10	PLCE1	608414	Nephrotic syndrome, type 3	Autosomal recessive
3	PLD1	602382	Cardiac valvular defect, developmental	Autosomal recessive
8	PLEC	601282	Epidermolysis bullosa simplex with muscular dystrophy	Autosomal recessive
1	PLEKHG5	611101	Charcot-Marie-Tooth disease, recessive intermediate, type C	Autosomal recessive
6	PLG	173350	Plasminogen deficiency, type I	Autosomal recessive
4	PLK4	605031	Microcephaly and chorioretinopathy, autosomal recessive, 2	Autosomal recessive
1	PLOD1	153454	Ehlers-Danlos syndrome, kyphoscoliotic type, 1	Autosomal recessive
3	PLOD2	601865	Bruck syndrome 2	Autosomal recessive
7	PLOD3	603066	Lysyl hydroxylase 3 deficiency	Autosomal recessive
X	PLP1	300401	Pelizaeus-Merzbacher disease	X-linked
8	PLPBP	604436	Epilepsy, early-onset, vitamin B6-dependent	Autosomal recessive
16	PMM2	601785	Congenital disorder of glycosylation, type 1A	Autosomal recessive
17	PMP22	601097	Dejerine-Sottas disease	Autosomal recessive*
9	PMPCA	613036	Spinocerebellar ataxia, autosomal recessive, type 2	Autosomal recessive
7	PMPCB	603131	Multiple mitochondrial dysfunctions syndrome 6	Autosomal recessive
19	PNKP	605610	Microcephaly, seizures, and developmental delay; Ataxia-oculomotor apraxia 4; ? Charcot-Marie-Tooth disease, type 2B2	Autosomal recessive
14	PNP	164050	Immunodeficiency due to purine nucleoside phosphorylase deficiency	Autosomal recessive
6	PNPLA1	612121	Ichthyosis, congenital, autosomal recessive, type 10	Autosomal recessive
11	PNPLA2	609059	Neutral lipid storage disease with myopathy	Autosomal recessive
19	PNPLA6	603197	Boucher-Neuhauser syndrome; Oliver-McFarlane syndrome; Spastic paraplegia, type 39, autosomal recessive	Autosomal recessive
17	PNPO	603287	Pyridoxamine 5'-phosphate oxidase deficiency	Autosomal recessive
2	PNPT1	610316	Combined oxidative phosphorylation deficiency 13	Autosomal recessive
3	POC1A	614783	Short stature, onychodysplasia, facial dysmorphism, and hypotrichosis	Autosomal recessive
12	POC1B	614784	Cone-rod dystrophy 20	Autosomal recessive

12	POLE	174762	FILS syndrome	Autosomal recessive
15	POLG	174763	POLG-related disorders	Autosomal recessive
6	POLH	603968	Xeroderma pigmentosum, variant type	Autosomal recessive
6	POLR1C	610060	Leukodystrophy, hypomyelinating, type 11; Treacher Collins syndrome 3	Autosomal recessive
13	POLR1D	613715	Treacher Collins syndrome, type 2	Autosomal recessive*
10	POLR3A	614258	Leukodystrophy, hypomyelinating, type 7	Autosomal recessive
12	POLR3B	614366	Leukodystrophy, hypomyelinating, type 8	Autosomal recessive
2	POMC	176830	Obesity, adrenal insufficiency, and red hair due to POMC deficiency	Autosomal recessive
1	POMGNT1	606822	Muscular dystrophy-dystroglycanopathy, type 3A (Walker-Warburg syndrome); Type 3B; Type 3C (limb-girdle muscular dystrophy, type 15 [LGMDR15])	Autosomal recessive
3	POMGNT2	614828	Muscular dystrophy-dystroglycanopathy, type 8A (Walker-Warburg syndrome); Type 8C (limb-girdle muscular dystrophy, type 24 [LGMD R24])	Autosomal recessive
8	POMK	615247	Muscular dystrophy-dystroglycanopathy (congenital with brain and eye anomalies), type A, 12	Autosomal recessive
13	POMP	613386	Keratosis linearis with ichthyosis congenita and sclerosing keratoderma	Autosomal recessive
9	POMT1	607423	Muscular dystrophy-dystroglycanopathy, type 1A (Walker-Warburg syndrome); Type 1B; Type 1C (limb-girdle muscular dystrophy, type 11 [LGMD R11])	Autosomal recessive
14	POMT2	607439	Muscular dystrophy-dystroglycanopathy, type 2A (Walker-Warburg syndrome); Type 2B; Type 2C (limb-girdle muscular dystrophy, type 14 [LGMD R14])	Autosomal recessive
8	POP1	602486	Anauxetic dysplasia, type 2	Autosomal recessive
7	POR	124015	Antley-Bixler syndrome with genital anomalies and disordered steroidogenesis	Autosomal recessive
3	POU1F1	173110	Pituitary hormone deficiency, combined, type 1	Autosomal recessive*
X	POU3F4	300039	Deafness, X-linked, type 2	X-linked
4	PPA2	609988	Sudden cardiac failure, infantile	Autosomal recessive
15	PPIB	123841	Osteogenesis imperfecta, type 9	Autosomal recessive
4	PPM1K	611065	?Maple syrup urine disease, mild variant	Autosomal recessive
1	PPP1R15B	613257	Microcephaly, short stature, and impaired glucose metabolism 2	Autosomal recessive
1	PPT1	600722	Ceroid lipofuscinosis, neuronal, type 1	Autosomal recessive
X	PQBP1	300463	Renpenning syndrome	X-linked
17	PRCD	610598	Retinitis pigmentosa, type 36	Autosomal recessive
9	PRDM12	616458	Neuropathy, hereditary sensory and autonomic, type VIII	Autosomal recessive
4	PRDM5	614161	Brittle cornea syndrome, type 2	Autosomal recessive
1	PRDX1	176763	Methylmalonic aciduria and homocystinuria, cblC type, digenic	Autosomal recessive; Digenic inheritance (MMACHC gene)
2	PREPL	609557	Myasthenic syndrome, congenital, type 22	Autosomal recessive
10	PRF1	170280	Hemophagocytic lymphohistiocytosis, familial, type 2	Autosomal recessive
1	PRG4	604283	Camptodactyly-arthropathy-coxa vara-pericarditis syndrome	Autosomal recessive
12	PRICKLE1	608500	Epilepsy, progressive myoclonic, type 1B	Autosomal recessive
3	PRKCD	176977	Autoimmune lymphoproliferative syndrome, type 3	Autosomal recessive
6	PRKN	602544	Parkinson disease, type 2, juvenile	Autosomal recessive
2	PRKRA	603424	Dystonia, type 16	Autosomal recessive
16	PRMT7	610087	Short stature, brachydactyly, intellectual developmental disability, and seizures	Autosomal recessive
2	PROC	612283	Thrombophilia due to protein C deficiency, autosomal recessive	Autosomal recessive
22	PRODH	606810	Hyperprolinemia, type 1	Autosomal recessive
4	PROM1	604365	Retinitis pigmentosa, type 41	Autosomal recessive
5	PROP1	601538	Pituitary hormone deficiency, combined, type 2	Autosomal recessive
3	PROS1	176880	Thrombophilia due to protein S deficiency, autosomal recessive	Autosomal recessive
6	PRPH2	179605	Leber congenital amaurosis 18; Retinitis punctata albescens	Autosomal recessive*
X	PRPS1	311850	PRPS1-related disoders	X-linked
1	PRRX1	167420	Agnathia-otocephaly complex	Autosomal recessive*
4	PRSS12	606709	Mental retardation, autosomal recessive, type 1	Autosomal recessive
2	PRSS56	613858	Microphthalmia, isolated, type 6	Autosomal recessive
1	PRUNE1	617413	Neurodevelopmental disorder with microcephaly, hypotonia, and variable brain anomalies	Autosomal recessive
19	PRX	605725	Charcot-Marie-Tooth disease, type 4F	Autosomal recessive
10	PSAP	176801	Combined SAP deficiency	Autosomal recessive

9	PSAT1	610936	Neu-Laxova syndrome, type 2	Autosomal recessive
6	PSMB8	177046	Autoinflammation, lipodystrophy, and dermatosis syndrome	Autosomal recessive
17	PSMC3IP	608665	Ovarian dysgenesis 3	Autosomal recessive
7	PSPH	172480	Phosphoserine phosphatase deficiency	Autosomal recessive
10	PTF1A	607194	Pancreatic agenesis 2	Autosomal recessive
11	PTH	168450	Hypoparathyroidism, familial isolated, type 1	Autosomal recessive*
3	PTH1R	168468	Chondrodysplasia, Blomstrand type; Eiken syndrome	Autosomal recessive
3	PTPN23	606584	Neurodevelopmental disorder and structural brain anomalies with or without seizures and spasticity	Autosomal recessive
1	PTPRC	151460	Severe combined immunodeficiency, T cell-negative, B-cell/natural killer-cell positive	Autosomal recessive
12	PTPRO	600579	Nephrotic syndrome, type 6	Autosomal recessive
12	PTPRQ	603317	Deafness, autosomal recessive, type 84A	Autosomal recessive
17	PTRH2	608625	Infantile-onset multisystem neurologic, endocrine, and pancreatic disease	Autosomal recessive
11	PTS	612719	Hyperphenylalaninemia, BH4-deficient, type A	Autosomal recessive
12	PUS1	608109	Myopathy, lactic acidosis, and sideroblastic anemia, type 1	Autosomal recessive
2	PXDN	605158	Anterior segment dysgenesis, type 7, with sclerocornea	Autosomal recessive
17	PYCR1	179035	Cutis laxa, autosomal recessive, type 2B	Autosomal recessive
1	PYCR2	616406	Leukodystrophy, hypomyelinating, type 10	Autosomal recessive
14	PYGL	613741	Glycogen storage disease, type 6	Autosomal recessive
11	PYGM	608455	McArdle disease	Autosomal recessive
12	PYROXD1	617220	Myopathy, myofibrillar, type 8	Autosomal recessive
3	QARS1	603727	Microcephaly, progressive, seizures, and cerebral and cerebellar atrophy	Autosomal recessive
4	QDPR	612676	Hyperphenylalaninemia, BH4-deficient, type C	Autosomal recessive
10	RAB18	602207	Warburg micro syndrome, type 3	Autosomal recessive
6	RAB23	606144	Carpenter syndrome	Autosomal recessive
15	RAB27A	603868	Griscelli syndrome, type 2	Autosomal recessive
4	RAB28	612994	Cone-rod dystrophy 18	Autosomal recessive
4	RAB33B	605950	Smith-McCort dysplasia 2	Autosomal recessive
2	RAB3GAP1	602536	Warburg micro syndrome, type 1	Autosomal recessive
1	RAB3GAP2	609275	Martsolf syndrome 1; Warburg micro syndrome 2	Autosomal recessive
5	RAD50	604040	Nijmegen breakage syndrome-like disorder	Autosomal recessive
17	RAD51C	602774	RAD51C-related conditions	Autosomal recessive
11	RAG1	179615	Omenn syndrome; Severe combined immunodeficiency, B cell-negative	Autosomal recessive
11	RAG2	179616	Omenn syndrome; Severe combined immunodeficiency, B cell-negative	Autosomal recessive
11	RAPSN	601592	Fetal akinesia deformation sequence, type 2; Myasthenic syndrome, congenital, type 11, associated with AChR deficiency	Autosomal recessive
3	RARB	180220	Microphthalmia, syndromic 12	Autosomal recessive
5	RARS1	107820	Leukodystrophy, hypomyelinating, type 9	Autosomal recessive
6	RARS2	611524	Pontocerebellar hypoplasia, type 6	Autosomal recessive
15	RASGRP1	603962	Immunodeficiency, type 64	Autosomal recessive
18	RAX	601881	Isolated microphthalmia, type 3	Autosomal recessive
18	RBBP8	604124	Jawad syndrome; Seckel syndrome, type 2	Autosomal recessive
20	RBCK1	610924	Polyglucosan body myopathy 1 with or without immunodeficiency	Autosomal recessive
1	RBM8A	605313	Thrombocytopenia-absent radius syndrome	Autosomal recessive
10	RBP3	180290	?Retinitis pigmentosa 66	Autosomal recessive
10	RBP4	180250	Retinal dystrophy, iris coloboma, and comedogenic acne syndrome	Autosomal recessive
13	RCBTB1	607867	Retinal dystrophy with or without extraocular anomalies	Autosomal recessive
1	RD3	180040	Leber congenital amaurosis, type 12	Autosomal recessive
14	RDH12	608830	Leber congenital amaurosis, type 13	Autosomal recessive
12	RDH5	601617	Fundus albipunctatus	Autosomal recessive*

11	RDX	179410	Deafness, autosomal recessive, type 24	Autosomal recessive
8	RECQL4	603780	Baller-Gerold syndrome; RAPADILINO syndrome; Rothmund-Thomson syndrome	Autosomal recessive
19	REEP6	609346	Retinitis pigmentosa 77	Autosomal recessive
7	RELN	600514	Lissencephaly 2 (Norman-Roberts type)	Autosomal recessive
1	REN	179820	Renal tubular dysgenesis	Autosomal recessive
5	RETREG1	613114	Neuropathy, hereditary sensory and autonomic, type 2B	Autosomal recessive
3	RFT1	611908	Congenital disorder of glycosylation, type In	Autosomal recessive
1	RFX5	601863	Bare lymphocyte syndrome, type 2	Autosomal recessive
6	RFX6	612659	Mitchell-Riley syndrome	Autosomal recessive
19	RFXANK	603200	Bare lymphocyte syndrome, type 2, complementation group B	Autosomal recessive
13	RFXAP	601861	Bare lymphocyte syndrome, type 2	Autosomal recessive
3	RHO	180380	Retinitis pigmentosa, type 4; Retinitis punctata albescens	Autosomal recessive*
20	RIN2	610222	Macs syndrome	Autosomal recessive
21	RIPK4	605706	Popliteal pterygium syndrome, Bartsocas-Papas type	Autosomal recessive
6	RIPOR2	611410	Deafness, autosomal recessive, type 104	Autosomal recessive
15	RLBP1	180090	Bothnia retinal dystrophy; Fundus albipunctatus	Autosomal recessive; Autosomal recessive*
6	RMND1	614917	Combined oxidative phosphorylation deficiency 11	Autosomal recessive
9	RMRP	157660	Anauxetic dysplasia, type 1	Autosomal recessive
2	RNASEH1	604123	Progressive external ophthalmoplegia with mitochondrial DNA deletions, autosomal recessive 2	Autosomal recessive
19	RNASEH2A	606034	Aicardi-Goutieres syndrome, type 4	Autosomal recessive
13	RNASEH2B	610326	Aicardi-Goutieres syndrome, type 2	Autosomal recessive
11	RNASEH2C	610330	Aicardi-Goutieres syndrome, type 3	Autosomal recessive
6	RNASET2	612944	Leukoencephalopathy, cystic, without megalencephaly	Autosomal recessive
3	RNF168	612688	RIDDLE syndrome	Autosomal recessive
7	RNF216	609948	Gordon Holmes syndrome	Autosomal recessive
11	ROBO3	608630	Gaze palsy, familial horizontal, with progressive scoliosis, type 1	Autosomal recessive
16	ROGDI	614574	Kohlschutter-Tonz syndrome	Autosomal recessive
11	ROM1	180721	Retinitis pigmentosa, type 7, digenic	Autosomal recessive
9	ROR2	602337	Robinow syndrome, autosomal recessive	Autosomal recessive
1	RORC	602943	Immunodeficiency, type 42	Autosomal recessive
8	RP1	603937	Retinitis pigmentosa, type 1	Autosomal recessive
X	RP2	300757	Retinitis pigmentosa, type 2, X-linked	X-linked
1	RPE65	180069	RPE65-related Leber congenital amaurosis/early-onset severe retinal dystrophy	Autosomal recessive
X	RPGR	312610	Retinitis pigmentosa, type 3, X-linked; Cone-rod dystrophy, X-linked, 1	X-linked
14	RPGRIP1	605446	Leber congenital amaurosis, type 6	Autosomal recessive
16	RPGRIP1L	610937	Joubert syndrome, type 7; Meckel syndrome, type 5; COACH syndrome	Autosomal recessive
8	RRM2B	604712	Mitochondrial DNA depletion syndrome, type 8A (encephalomyopathic type with renal tubulopathy) and type 8B (MNGIE type)	Autosomal recessive
X	RS1	300839	Retinoschisis	X-linked
21	RSPH1	609314	Ciliary dyskinesia, primary, type 24	Autosomal recessive
6	RSPH3	615876	Ciliary dyskinesia, primary, type 32	Autosomal recessive
6	RSPH4A	612647	Ciliary dyskinesia, primary, type 11	Autosomal recessive
6	RSPH9	612648	Ciliary dyskinesia, primary, type 12	Autosomal recessive
20	RSPO4	610573	Anonychia congenita	Autosomal recessive
16	RSPRY1	616585	Spondyloepimetaphyseal dysplasia, Faden-Alkuraya type	Autosomal recessive
20	RTEL1	608833	Dyskeratosis congenita, autosomal recessive type 5	Autosomal recessive*
6	RTN4IP1	610502	Optic atrophy 10 with or without ataxia, mental retardation, and seizures	Autosomal recessive
18	RTTN	610436	Microcephaly, short stature, and polymicrogyria with seizures	Autosomal recessive
9	RUSC2	611053	Mental retardation, autosomal recessive 61	Autosomal recessive
12	RXYLT1	605862	Muscular dystrophy-dystroglycanopathy (congenital with brain and eye anomalies), type A, 10	Autosomal recessive

19	RYR1	180901	Malignant hyperthermia susceptibility, type 1	Autosomal dominant
19	S1PR2	605111	Deafness, autosomal recessive, type 68	Autosomal recessive
13	SACS	604490	Spastic ataxia, Charlevoix-Saguenay, type	Autosomal recessive
2	SAG	181031	Oguchi disease, type 1	Autosomal recessive
7	SAMD9	610456	Tumoral calcinosis, familial, normophosphatemic	Autosomal recessive
20	SAMHD1	606754	Aicardi-Goutieres syndrome, type 5	Autosomal recessive
5	SAR1B	607690	Chylomicron retention disease	Autosomal recessive
19	SARS2	612804	Hyperuricemia, pulmonary hypertension, renal failure, and alkalosis	Autosomal recessive
7	SBDS	607444	Shwachman-Diamond syndrome	Autosomal recessive
22	SBF1	603560	Charcot-Marie-Tooth disease, type 4B3	Autosomal recessive
11	SBF2	607697	Charcot-Marie-Tooth disease, type 4B2	Autosomal recessive
11	SC5D	602286	Lathosterolosis	Autosomal recessive
4	SCARB2	602257	Epilepsy, progressive myoclonic, type 4, with or without renal failure	Autosomal recessive
22	SCARF2	613619	Van den Ende-Gupta syndrome	Autosomal recessive
19	SCN1B	600235	Epileptic encephalopathy, early infantile, type 52	Autosomal recessive
17	SCN4A	603967	Myasthenic syndrome, congenital, type 16	Autosomal recessive
2	SCN9A	603415	Indifference to pain and autosomal recessive hereditary sensory neuropathy type 2D	Autosomal recessive
12	SCNN1A	600228	Pseudohypoaldosteronism, type 1	Autosomal recessive
16	SCNN1B	600760	Pseudohypoaldosteronism, type 1	Autosomal recessive
16	SCNN1G	600761	Pseudohypoaldosteronism, type 1	Autosomal recessive
17	SCO1	603644	Mitochondrial complex IV deficiency, nuclear type 4	Autosomal recessive
22	SCO2	604272	Cardioencephalomyopathy, fatal infantile, due to cytochrome c oxidase deficiency, type 1	Autosomal recessive
11	SCYL1	607982	Spinocerebellar ataxia, autosomal recessive, type 21	Autosomal recessive
1	SDCCAG8	613524	Bardet-Biedl syndrome, type 16	Autosomal recessive
5	SDHA	600857	Mitochondrial respiratory chain complex II deficiency; Leigh syndrome	Autosomal recessive
19	SDHAF1	612848	Mitochondrial complex II deficiency	Autosomal recessive
12	SDR9C7	609769	Ichthyosis, congenital, autosomal recessive 13	Autosomal recessive
14	SEC23A	610511	Craniolenticulosutural dysplasia	Autosomal recessive
20	SEC23B	610512	Dyserythropoietic anemia, congenital, type 2	Autosomal recessive
4	SEC24D	607186	Cole-Carpenter syndrome 2	Autosomal recessive
9	SECISBP2	607693	Thyroid hormone metabolism, abnormal	Autosomal recessive
1	SELENON	606210	Muscular dystrophy, rigid spine, type 1	Autosomal recessive
1	SEMA4A	607292	Cone-rod dystrophy, type 10; Retinitis pigmentosa, type 35	Autosomal recessive
4	SEPSECS	613009	Pontocerebellar hypoplasia, type 2D	Autosomal recessive
6	SERAC1	614725	3-methylglutaconic aciduria with deafness, encephalopathy, and Leigh-like syndrome (MEGDEL)	Autosomal recessive
14	SERPINA1	107400	Alpha-1 antitrypsin deficiency	Autosomal recessive
18	SERPINB7	603357	Palmoplantar keratoderma, Nagashima type	Autosomal recessive
18	SERPINB8	601697	Peeling skin syndrome 5	Autosomal recessive
1	SERPINC1	107300	Thrombophilia due to antithrombin III deficiency	Autosomal recessive*
7	SERPINE1	173360	Plasminogen activator inhibitor-1 deficiency	Autosomal recessive*
17	SERPINF1	172860	Osteogenesis imperfecta, type 6	Autosomal recessive
17	SERPINF2	613168	Alpha-2-plasmin inhibitor deficiency	Autosomal recessive
11	SERPING1	606860	Angioedema, hereditary, types 1 and 2	Autosomal recessive*
11	SERPINH1	600943	Osteogenesis imperfecta, type 10	Autosomal recessive
9	SETX	608465	Spinocerebellar ataxia, autosomal recessive, type 1	Autosomal recessive
7	SFRP4	606570	Pyle disease	Autosomal recessive
2	SFTPB	178640	Surfactant metabolism dysfunction, pulmonary, type 1	Autosomal recessive
10	SFXN4	615564	Combined oxidative phosphorylation deficiency 18	Autosomal recessive

17	SGCA	600119	Limb-girdle muscular dystrophy, type 3 (LGMD R3)	Autosomal recessive
4	SGCB	600900	Limb-girdle muscular dystrophy, type 4 (LGMD R4)	Autosomal recessive
5	SGCD	601411	Limb-girdle muscular dystrophy, type 6 (LGMD R6)	Autosomal recessive
13	SGCG	608896	Limb-girdle muscular dystrophy, type 5 (LGMD R5)	Autosomal recessive
10	SGPL1	603729	Nephrotic syndrome, type 14	Autosomal recessive
17	SGSH	605270	Mucopolysaccharidosis, type 3A (Sanfilippo A)	Autosomal recessive
X	SH2D1A	300490	Lymphoproliferative syndrome, X-linked, type 1	X-linked
5	SH3PXD2B	613293	Frank-ter Haar syndrome	Autosomal recessive
5	SH3TC2	608206	Charcot-Marie-Tooth disease, type 4C	Autosomal recessive
3	SI	609845	Sucrase-isomaltase deficiency, congenital	Autosomal recessive
5	SIL1	608005	Marinesco-Sjogren syndrome	Autosomal recessive
14	SIX6	606326	Optic disc anomalies with retinal and/or macular dystrophy	Autosomal recessive
6	SKIC2	600478	Trichohepatoenteric syndrome, type 2 (diarrhea, syndromic)	Autosomal recessive
5	SKIC3	614589	Trichohepatoenteric syndrome, type 1 (diarrhea, syndromic)	Autosomal recessive
13	SLC10A2	601295	Bile acid malabsorption, primary	Autosomal recessive
12	SLC11A2	600523	Anemia, hypochromic microcytic, with iron overload 1	Autosomal recessive
15	SLC12A1	600839	Bartter syndrome, type 1	Autosomal recessive
16	SLC12A3	600968	Gitelman syndrome	Autosomal recessive
20	SLC12A5	606726	Epileptic encephalopathy, early infantile, 34	Autosomal recessive
15	SLC12A6	604878	Agenesis of the corpus callosum with peripheral neuropathy	Autosomal recessive
17	SLC13A5	608305	Epileptic encephalopathy, early infantile, 25	Autosomal recessive
1	SLC16A1	600682	Monocarboxylate transporter 1 deficiency	Autosomal recessive*
X	SLC16A2	300095	Allan-Herndon-Dudley syndrome	X-linked
6	SLC17A5	604322	Salla disease	Autosomal recessive
10	SLC18A3	600336	Myasthenic syndrome, congenital, 21, presynaptic	Autosomal recessive
1	SLC19A2	603941	Thiamine-responsive megaloblastic anemia syndrome	Autosomal recessive
2	SLC19A3	606152	Thiamine metabolism dysfunction syndrome, type 2 (biotin- or thiamine-responsive encephalopathy type)	Autosomal recessive
9	SLC1A1	133550	Dicarboxylic aminoaciduria	Autosomal recessive
2	SLC1A4	600229	Spastic tetraplegia, thin corpus callosum, and progressive microcephaly	Autosomal recessive
11	SLC22A12	607096	Hypouricemia, renal	Autosomal recessive
5	SLC22A5	603377	Carnitine deficiency, systemic primary	Autosomal recessive
15	SLC24A1	603617	Night blindness, congenital stationary (complete), type 1D, autosomal recessive	Autosomal recessive
14	SLC24A4	609840	Amelogenesis imperfecta, type IIA5	Autosomal recessive
15	SLC24A5	609802	Albinism, oculocutaneous, type 6	Autosomal recessive
22	SLC25A1	190315	Combined D-2- and L-2-hydroxyglutaric aciduria	Autosomal recessive
2	SLC25A12	603667	Epileptic encephalopathy, early infantile, type 39	Autosomal recessive
7	SLC25A13	603859	Citrullinemia, type 2, neonatal-onset; Citrullinemia, type 2, adult-onset	Autosomal recessive
13	SLC25A15	603861	Hyperornithinemia-hyperammonemia-homocitrullinemia syndrome	Autosomal recessive
17	SLC25A19	606521	Microcephaly, Amish type; Thiamine metabolism dysfunction syndrome 4 (progressive polyneuropathy type)	Autosomal recessive
3	SLC25A20	613698	Carnitine-acylcarnitine translocase deficiency	Autosomal recessive
11	SLC25A22	609302	Epileptic encephalopathy, early infantile, type 3	Autosomal recessive
3	SLC25A26	611037	Combined oxidative phosphorylation deficiency 28	Autosomal recessive
12	SLC25A3	600370	Mitochondrial phosphate carrier deficiency	Autosomal recessive
3	SLC25A38	610819	Anemia, sideroblastic, type 2, pyridoxine-refractory	Autosomal recessive
4	SLC25A4	103220	Mitochondrial DNA depletion syndrome, type 12B (cardiomyopathic type) AR	Autosomal recessive
5	SLC25A46	610826	Neuropathy, hereditary motor and sensory, type VIB	Autosomal recessive
5	SLC26A2	606718	Achondrogenesis Ib; Atelosteogenesis, type II;De la Chapelle dysplasia; Diastrophic dysplasia;Diastrophic dysplasia, broad bone-platyspondylic variant;Epiphyseal dysplasia, multiple, 4	Autosomal recessive
7	SLC26A3	126650	Diarrhea 1, secretory chloride, congenital	Autosomal recessive
7	SLC26A4	605646	Deafness, autosomal recessive, type 4; Pendred syndrome	Autosomal recessive

7	SLC26A5	604943	?Deafness, autosomal recessive, type 61	Autosomal recessive
9	SLC27A4	604194	Ichthyosis prematurity syndrome	Autosomal recessive
10	SLC29A3	612373	Histiocytosis-lymphadenopathy plus syndrome	Autosomal recessive
1	SLC2A1	138140	GLUT1 deficiency syndrome 1, infantile onset, severe	Autosomal recessive*
20	SLC2A10	606145	Arterial tortuosity syndrome	Autosomal recessive
3	SLC2A2	138160	Fanconi-Bickel syndrome	Autosomal recessive
4	SLC2A9	606142	Hypouricemia, renal, type 2	Autosomal recessive*
1	SLC30A10	611146	Hypermanganesemia with dystonia, type 1	Autosomal recessive
3	SLC33A1	603690	Congenital cataracts, hearing loss, and neurodegeneration	Autosomal recessive
5	SLC34A1	182309	Hypercalcemia, infantile, type 2	Autosomal recessive
4	SLC34A2	604217	Pulmonary alveolar microlithiasis	Autosomal recessive
9	SLC34A3	609826	Hypophosphatemic rickets with hypercalciuria	Autosomal recessive
6	SLC35A1	605634	Congenital disorder of glycosylation, type 2F	Autosomal recessive
1	SLC35A3	605632	Arthrogryposis, impaired intellectual development, and seizures	Autosomal recessive
11	SLC35C1	605881	Congenital disorder of glycosylation, type 2C	Autosomal recessive
1	SLC35D1	610804	Schneckenbecken dysplasia	Autosomal recessive
11	SLC37A4	602671	Glycogen storage disease, type 1B	Autosomal recessive
16	SLC38A8	615585	Foveal hypoplasia 2, with or without optic nerve misrouting and/or anterior segment dysgenesis	Autosomal recessive
11	SLC39A13	608735	Ehlers-Danlos syndrome, spondylodysplastic type, 3	Autosomal recessive
8	SLC39A14	608736	Hypermanganesemia with dystonia 2	Autosomal recessive
8	SLC39A4	607059	Acrodermatitis enteropathica	Autosomal recessive
4	SLC39A8	608732	Congenital disorder of glycosylation, type IIn	Autosomal recessive
2	SLC3A1	104614	Cystinuria	Autosomal recessive*
1	SLC45A1	605763	Intellectual developmental disorder with neuropsychiatric features	Autosomal recessive
5	SLC45A2	606202	Albinism, oculocutaneous, type 4	Autosomal recessive
17	SLC46A1	611672	Folate malabsorption, hereditary	Autosomal recessive
17	SLC4A1	109270	Distal renal tubular acidosis	Autosomal recessive
20	SLC4A11	610206	Corneal endothelial dystrophy, autosomal recessive	Autosomal recessive
4	SLC4A4	603345	Renal tubular acidosis, proximal, with ocular abnormalities	Autosomal recessive
8	SLC52A2	607882	Brown-Vialetto-Van Laere syndrome, type 2	Autosomal recessive
20	SLC52A3	613350	Brown-Vialetto-Van Laere syndrome, type 1	Autosomal recessive
22	SLC5A1	182380	Glucose/galactose malabsorption	Autosomal recessive
16	SLC5A2	182381	Renal glucosuria	Autosomal recessive*
19	SLC5A5	601843	Thyroid dyshormonogenesis, type 1	Autosomal recessive
2	SLC5A7	608761	Myasthenic syndrome, congenital, type 20, presynaptic	Autosomal recessive
1	SLC6A17	610299	Mental retardation, autosomal recessive 48	Autosomal recessive
5	SLC6A19	608893	Hartnup disorder	Autosomal recessive
5	SLC6A3	126455	Parkinsonism-dystonia, infantile	Autosomal recessive
11	SLC6A5	604159	Hyperekplexia, type 3	Autosomal recessive*
X	SLC6A8	300036	Cerebral creatine deficiency syndrome, type 1	X-linked
1	SLC6A9	601019	Glycine encephalopathy with normal serum glycine	Autosomal recessive
3	SLC7A14	615720	Retinitis pigmentosa 68	Autosomal recessive
14	SLC7A7	603593	Lysinuric protein intolerance	Autosomal recessive
19	SLC7A9	604144	Cystinuria	Autosomal recessive*
5	SLC9A3	182307	Diarrhea 8, secretory sodium, congenital	Autosomal recessive
3	SLCO2A1	601460	Hypertrophic osteoarthropathy, primary, autosomal recessive, type 2	Autosomal recessive
13	SLITRK6	609681	Deafness and myopia	Autosomal recessive
8	SLURP1	606119	Meleda disease	Autosomal recessive

16	SLX4	613278	Fanconi anemia, complementation group P	Autosomal recessive
2	SMARCAL1	606622	Schimke immunoosseous dysplasia	Autosomal recessive
17	SMARCD2	601736	Specific granule deficiency 2	Autosomal recessive
19	SMG9	613176	Heart and brain malformation syndrome	Autosomal recessive
5	SMN1	600354	Spinal muscular atrophy	Autosomal recessive
14	SMOC1	608488	Microphthalmia. with limb anomalies	Autosomal recessive
6	SMOC2	607223	Dentin dysplasia, type 1, with microdontia and misshapen teeth	Autosomal recessive
11	SMPD1	607608	Niemann-Pick disease, type A; Niemann-Pick disease, type B	Autosomal recessive
22	SNAP29	604202	Cerebral dysgenesis, neuropathy, ichthyosis, and palmoplantar keratoderma syndrome	Autosomal recessive
7	SNX10	614780	Osteopetrosis, autosomal recessive, type 8	Autosomal recessive
6	SNX14	616105	Spinocerebellar ataxia, autosomal recessive, type 20	Autosomal recessive
6	SOBP	613667	Mental retardation, anterior maxillary protrusion, and strabismus	Autosomal recessive
21	SOD1	147450	Spastic tetraplegia and axial hypotonia, progressive; Amyotrophic lateral sclerosis, type 1	Autosomal recessive; Autosomal recessive*
9	SOHLH1	610224	Ovarian dysgenesis 5	Autosomal recessive
17	SOST	605740	Sclerosteosis, type 1; Van Buchem disease	Autosomal recessive
20	SOX18	601618	Hypotrichosis-lymphedema-telangiectasia syndrome	Autosomal recessive
2	SP110	604457	Hepatic venoocclusive disease with immunodeficiency	Autosomal recessive
12	SP7	606633	Osteogenesis imperfecta, type XII	Autosomal recessive
8	SPAG1	603395	Ciliary dyskinesia, primary, type 28	Autosomal recessive
5	SPARC	182120	Osteogenesis imperfecta, type XVII	Autosomal recessive
13	SPART	607111	Spactic paraplegia, type 20, autosomal recessive	Autosomal recessive
4	SPATA5	613940	Epilepsy, hearing loss, and mental retardation syndrome	Autosomal recessive
14	SPATA7	609868	Leber congenital amaurosis, type 3	Autosomal recessive
2	SPEG	615950	Centronuclear myopathy, type 5	Autosomal recessive
15	SPG11	610844	Amyotrophic lateral sclerosis 5, juvenile; Charcot-Marie-Tooth disease, axonal, type 2X; Spastic paraplegia 11	Autosomal recessive
15	SPG21	608181	Mast syndrome	Autosomal recessive
16	SPG7	602783	Spastic paraplegia, type 7, autosomal recessive	Autosomal recessive
5	SPINK1	167790	Tropical calcific pancreatitis	Autosomal recessive*
5	SPINK5	605010	Netherton syndrome	Autosomal recessive
19	SPINT2	605124	Diarrhea 3, secretory sodium, congenital, syndromic	Autosomal recessive
2	SPR	182125	Dystonia, dopa-responsive, due to sepiapterin reductase deficiency	Autosomal recessive*
1	SPRTN	616086	Ruijs-Aalfs syndrome	Autosomal recessive
1	SPTA1	182860	Pyropoikilocytosis; Spherocytosis, type 3	Autosomal recessive
11	SPTBN2	604985	Spinocerebellar ataxia, autosomal recessive, type 14	Autosomal recessive
19	SPTBN4	606214	Neurodevelopmental disorder with hypotonia, neuropathy, and deafness	Autosomal recessive
5	SQSTM1	601530	Neurodegeneration with ataxia, dystonia, and gaze palsy, childhood-onset	Autosomal recessive
2	SRD5A2	607306	46,XY disorder of sex development due to 5-alpha-reductase 2 deficiency (pseudovaginal perineoscrotal hypospadias)	Autosomal recessive
4	SRD5A3	611715	Congenital disorder of glycosylation, type 1Q; Kahrizi syndrome	Autosomal recessive
11	ST14	606797	Ichthyosis, congenital, autosomal recessive, type 11	Autosomal recessive
1	ST3GAL3	606494	Mental retardation, autosomal recessive 12	Autosomal recessive
2	ST3GAL5	604402	Salt and pepper developmental regression syndrome	Autosomal recessive
12	STAC3	615521	Native American myopathy	Autosomal recessive
7	STAG3	608489	Premature ovarian failure, type 8; Spermatogenic failure 61	Autosomal recessive
2	STAMBP	606247	Microcephaly-capillary malformation syndrome	Autosomal recessive
8	STAR	600617	Lipoid adrenal hyperplasia	Autosomal recessive
2	STAT1	600555	Immunodeficiency, type 31B, mycobacterial and viral infections	Autosomal recessive
12	STAT2	600556	Immunodeficiency, type 44	Autosomal recessive
17	STAT5B	604260	Laron syndrome with immunodeficiency	Autosomal recessive
1	STIL	181590	Microcephaly, type 7, primary, autosomal recessive	Autosomal recessive

11	STIM1	605921	Immunodeficiency, type 10	Autosomal recessive
20	STK4	604965	T-cell immunodeficiency, recurrent infections, autoimmunity, and cardiac malformations	Autosomal recessive
15	STRA6	610745	Microphthalmia, isolated, with coloboma, type 8	Autosomal recessive
17	STRADA	608626	Polyhydramnios, megalencephaly, and symptomatic epilepsy	Autosomal recessive
15	STRC	606440	Deafness, autosomal recessive, type 16	Autosomal recessive
16	STUB1	607207	Spinocerebellar ataxia, autosomal recessive, type 16	Autosomal recessive
6	STX11	605014	Hemophagocytic lymphohistiocytosis, familial, type 4	Autosomal recessive
19	STXBP2	601717	Hemophagocytic lymphohistiocytosis, familial, type 5	Autosomal recessive
13	SUCLA2	603921	Mitochondrial DNA depletion syndrome, type 5 (encephalomyopathic with or without methylmalonic aciduria)	Autosomal recessive
2	SUCLG1	611224	Mitochondrial DNA depletion syndrome, type 9 (encephalomyopathic, type with methylmalonic aciduria)	Autosomal recessive
10	SUFU	607035	Joubert syndrome, type 32	Autosomal recessive
7	SUGCT	609187	Glutaric aciduria, type 3	Autosomal recessive
19	SULT2B1	604125	Ichthyosis, congenital, autosomal recessive, type 14	Autosomal recessive
3	SUMF1	607939	Multiple sulfatase deficiency	Autosomal recessive
20	SUN5	613942	Spermatogenic failure, type 16	Autosomal recessive
12	SUOX	606887	Sulfite oxidase deficiency	Autosomal recessive
9	SURF1	185620	Mitochondrial complex IV deficiency, nuclear type 1; Charcot-Marie-Tooth disease, type 4K	Autosomal recessive
X	SYN1	313440	Epilepsy, X-linked, with variable learning disabilities and behavior disorders	X-linked
6	SYNE1	608441	Spinocerebellar ataxia, autosomal recessive, type 8	Autosomal recessive
19	SYNE4	615535	Deafness, autosomal recessive, type 76	Autosomal recessive
21	SYNJ1	604297	Epileptic encephalopathy, early infantile, 53	Autosomal recessive
1	SYT14	610949	?Spinocerebellar ataxia, autosomal recessive, type 11	Autosomal recessive
1	SZT2	615463	Epileptic encephalopathy, early infantile, 18	Autosomal recessive
12	TAC3	162330	Hypogonadotropic hypogonadism, type 10, with or without anosmia	Autosomal recessive
17	TACO1	612958	Mitochondrial complex IV deficiency, nuclear type 8	Autosomal recessive
4	TACR3	162332	Hypogonadotropic hypogonadism, type 11, with or without anosmia	Autosomal recessive
1	TACSTD2	137290	Corneal dystrophy, gelatinous drop-like	Autosomal recessive
1	TAF13	600774	Mental retardation, autosomal recessive 60	Autosomal recessive
8	TAF2	604912	Mental retardation, autosomal recessive 40	Autosomal recessive
7	TAF6	602955	Alazami-Yuan syndrome	Autosomal recessive
11	TALDO1	602063	Transaldolase deficiency	Autosomal recessive
22	TANGO2	616830	Metabolic encephalomyopathic crises, recurrent, with rhabdomyolysis, cardiac arrhythmias, and neurodegeneration	Autosomal recessive
6	TAP1	170260	Bare lymphocyte syndrome, type 1	Autosomal recessive
6	TAP2	170261	Bare lymphocyte syndrome, type 1, due to TAP2 deficiency	Autosomal recessive
6	TAPBP	601962	Bare lymphocyte syndrome, type 1	Autosomal recessive
4	TAPT1	612758	Osteochondrodysplasia, complex lethal, Symoens-Barnes-Gistelinck type	Autosomal recessive
16	TAT	613018	Tyrosinemia, type 2	Autosomal recessive
20	TBC1D20	611663	Warburg micro syndrome 4	Autosomal recessive
3	TBC1D23	617687	Pontocerebellar hypoplasia, type 11	Autosomal recessive
16	TBC1D24	613577	DOORS (deafness, onychodystrophy, osteodystrophy, mental retardation, and seizures) syndrome; Epileptic encephalopathy, early infantile, type 16; Deafness, autosomal recessive, type 86	Autosomal recessive
6	TBC1D7	612655	Macrocephaly/megalencephaly syndrome, autosomal recessive	Autosomal recessive
17	TBCD	604649	Encephalopathy, progressive, early-onset, with brain atrophy and thin corpus callosum	Autosomal recessive
1	TBCE	604934	Encephalopathy, progressive, with amyotrophy and optic atrophy; Hypoparathyroidism-retardation-dysmorphism syndrome; Kenny-Caffey syndrome, type 1	Autosomal recessive
4	TBCK	616899	Hypotonia, infantile, with psychomotor retardation and characteristic facies 3	Autosomal recessive
1	TBX15	604127	Cousin syndrome	Autosomal recessive
1	TBX19	604614	Congenital isolated adrenocorticotropic hormone deficiency	Autosomal recessive
7	TBXAS1	274180	Ghosal syndrome	Autosomal recessive
17	TCAP	604488	Limb-girdle muscular dystrophy, type 7 (LGMD R7)	Autosomal recessive

11	TCIRG1	604592	Osteopetrosis, autosomal recessive, type 1	Autosomal recessive
22	TCN2	613441	Transcobalamin II deficiency	Autosomal recessive
12	TCTN1	609863	Joubert syndrome, type 13	Autosomal recessive
12	TCTN2	613846	Joubert syndrome, type 24; ?Meckel syndrome, type 8	Autosomal recessive
10	TCTN3	613847	Joubert syndrome 18; Orofaciodigital syndrome IV	Autosomal recessive
14	TDP1	607198	?Spinocerebellar ataxia, autosomal recessive with axonal neuropathy	Autosomal recessive
6	TDP2	605764	Spinocerebellar ataxia, autosomal recessive, type 23	Autosomal recessive
9	TDRD7	611258	Cataract 36	Autosomal recessive
14	TECPR2	615000	Spastic paraplegia, type 49, autosomal recessive	Autosomal recessive
19	TECR	610057	Mental retardation, autosomal recessive, type 14	Autosomal recessive
4	TECRL	617242	Ventricular tachycardia, catecholaminergic polymorphic, 3	Autosomal recessive
11	TECTA	602574	Deafness, autosomal recessive, type 21	Autosomal recessive
16	TELO2	611140	You-Hoover-Fong syndrome	Autosomal recessive
4	TENM3	610083	Microphthalmia, isolated, with coloboma 9	Autosomal recessive
5	TERT	187270	Dyskeratosis congenita, autosomal recessive, type 4	Autosomal recessive
8	TEX15	605795	Spermatogenic failure, type 25	Autosomal recessive
3	TF	190000	Atransferrinemia	Autosomal recessive
7	TFR2	604720	Hemochromatosis, type 3	Autosomal recessive
3	TFRC	190010	Immunodeficiency, type 46	Autosomal recessive
8	TG	188450	Thyroid dyshormonogenesis, type 3	Autosomal recessive
13	TGDS	616146	Catel-Manzke syndrome	Autosomal recessive
14	TGM1	190195	Ichthyosis, congenital, autosomal recessive, type 1	Autosomal recessive
15	TGM5	603805	Peeling skin syndrome, type 2	Autosomal recessive
11	TH	191290	Segawa syndrome, recessive	Autosomal recessive
X	THOC2	300395	Mental retardation, X-linked 12	X-linked
16	THOC6	615403	Beaulieu-Boycott-Innes syndrome	Autosomal recessive
3	THRB	190160	Thyroid hormone resistance, autosomal recessive	Autosomal recessive
19	TIMM50	607381	3-methylglutaconic aciduria, type 9	Autosomal recessive
3	TIMMDC1	615534	Mitochondrial complex I deficiency, nuclear type 31	Autosomal recessive
9	TJP2	607709	Cholestasis, progressive familial intrahepatic 4; Hypercholanemia, familial 1	Autosomal recessive
16	TK2	188250	Mitochondrial DNA depletion syndrome , type 2 (myopathic type)	Autosomal recessive
3	TKT	606781	Short stature, developmental delay, and congenital heart defects	Autosomal recessive
19	TLE6	612399	Preimplantation embryonic lethality	Autosomal recessive
9	TMC1	606706	Deafness, autosomal recessive, type 7	Autosomal recessive
17	TMC6	605828	Epidermodysplasia verruciformis	Autosomal recessive
17	TMC8	605829	Epidermodysplasia verruciformis	Autosomal recessive
1	TMCO1	614123	Craniofacial dysmorphism, skeletal anomalies, and mental retardation syndrome	Autosomal recessive
17	TMEM107	616183	Meckel syndrome, type 13; Orofaciodigital syndrome, type 16	Autosomal recessive
11	TMEM126A	612988	Optic atrophy 7	Autosomal recessive
11	TMEM126B	615533	Mitochondrial complex I deficiency, nuclear type 29	Autosomal recessive
11	TMEM138	614459	Joubert syndrome 16	Autosomal recessive
4	TMEM165	614726	Congenital disorder of glycosylation, type 2K	Autosomal recessive
17	TMEM199	616815	Congenital disorder of glycosylation, type 2P	Autosomal recessive
11	TMEM216	613277	Joubert syndrome, type 2; Meckel syndrome, type 2	Autosomal recessive
16	TMEM231	614949	Joubert syndrome, type 20; Meckel syndrome,type 11	Autosomal recessive
2	TMEM237	614423	Joubert syndrome, type 14	Autosomal recessive
14	TMEM260	617449	Structural heart defects and renal anomalies syndrome	Autosomal recessive
8	TMEM67	609884	Meckel syndrome 3; COACH syndrome 1; Joubert syndrome 6; Nephronophthisis 11	Autosomal recessive
8	TMEM70	612418	Mitochondrial complex V (ATP synthase) deficiency, nuclear type 2	Autosomal recessive

3	TMIE	607237	Deafness, autosomal recessive, type 6	Autosomal recessive
21	TMPRSS15	606635	Enterokinase deficiency	Autosomal recessive
21	TMPRSS3	605511	Deafness, autosomal recessive, type 8/10	Autosomal recessive
22	TMPRSS6	609862	Iron-refractory iron deficiency anemia	Autosomal recessive
12	TMTC3	617218	Lissencephaly 8	Autosomal recessive
18	TNFRSF11A	603499	Osteopetrosis, autosomal recessive, type 7	Autosomal recessive
8	TNFRSF11B	602643	Paget disease of bone, type 5, juvenile-onset	Autosomal recessive
17	TNFRSF13B	604907	Immunodeficiency, common variable, type 2	Autosomal recessive
13	TNFSF11	602642	Osteopetrosis, autosomal recessive, type 2	Autosomal recessive
3	TNIK	610005	Mental retardation, autosomal recessive 54	Autosomal recessive
19	TNNT1	191041	Nemaline myopathy , type 5, Amish type	Autosomal recessive
6	TNXB	600985	Ehlers-Danlos syndrome, classic-like	Autosomal recessive
1	TOE1	613931	Pontocerebellar hypoplasia, type 7	Autosomal recessive
17	TOP3A	601243	Microcephaly, growth restriction, and increased sister chromatid exchange 2	Autosomal recessive
20	TP53RK	608679	Galloway-Mowat syndrome 4	Autosomal recessive
12	TPI1	190450	Hemolytic anemia due to triosephosphate isomerase deficiency	Autosomal recessive
7	TPK1	606370	Episodic encephalopathy due to thiamine pyrophosphokinase deficiency	Autosomal recessive
1	TPM3	191030	Nemaline myopathy, type 1; Congenital fiber-type disproportion myopathy	Autosomal recessive*
2	TPO	606765	Thyroid dyshormonogenesis, type 2A	Autosomal recessive
11	TPP1	607998	Ceroid lipofuscinosis, neuronal, type 2; Spinocerebellar ataxia, autosomal recessive, type 7	Autosomal recessive
9	TPRN	613354	Deafness, autosomal recessive, type 79	Autosomal recessive
2	TRAF3IP1	607380	Senior-Loken syndrome, type 9	Autosomal recessive
3	TRAIP	605958	Seckel syndrome, type 9	Autosomal recessive
4	TRAPPC11	614138	Limb-girdle muscular dystrophy, type 18 (LGMD R18)	Autosomal recessive
2	TRAPPC12	614139	Encephalopathy, progressive, early-onset, with brain atrophy and spasticity	Autosomal recessive
14	TRAPPC6B	610397	Neurodevelopmental disorder with microcephaly, epilepsy, and brain atrophy	Autosomal recessive
8	TRAPPC9	611966	Mental retardation, autosomal recessive, type 13	Autosomal recessive
6	TRDN	603283	Ventricular tachycardia, catecholaminergic polymorphic, type 5, with or without muscle weakness	Autosomal recessive
6	TREM2	605086	Nasu-Hakola disease	Autosomal recessive
3	TREX1	606609	Aicardi-Goutieres syndrome, type 1	Autosomal recessive
8	TRHR	188545	Hypothyroidism, congenital, nongoitrous, type 7	Autosomal recessive
4	TRIM2	614141	Charcot-Marie-Tooth disease, type 2R	Autosomal recessive
9	TRIM32	602290	Limb-girdle muscular dystrophy, type 8 (LGMD R8)	Autosomal recessive
17	TRIM37	605073	Mulibrey nanism	Autosomal recessive
22	TRIOBP	609761	Deafness, autosomal recessive, type 28	Autosomal recessive
14	TRIP11	604505	Achondrogenesis, type 1A	Autosomal recessive
5	TRIP13	604507	Mosaic variegated aneuploidy syndrome 3	Autosomal recessive
15	TRIP4	604501	Spinal muscular atrophy with congenital bone fractures 1	Autosomal recessive
1	TRIT1	617840	Combined oxidative phosphorylation deficiency 35	Autosomal recessive
4	TRMT10A	616013	Microcephaly, short stature, and impaired glucose metabolism 1	Autosomal recessive
3	TRMT10C	615423	Combined oxidative phosphorylation deficiency 30	Autosomal recessive
14	TRMT5	611023	Combined oxidative phosphorylation deficiency 26	Autosomal recessive
22	TRMU	610230	Liver failure, transient infantile	Autosomal recessive
3	TRNT1	612907	Retinitis pigmentosa and erythrocytic microcytosis	Autosomal recessive
15	TRPM1	603576	Night blindness, congenital stationary (complete), type 1C, autosomal recessive	Autosomal recessive
9	TRPM6	607009	Familial hypomagnesemia with secondary hypocalcemia	Autosomal recessive
7	TRPV6	606680	Hyperparathyroidism, transient neonatal	Autosomal recessive
1	TSEN15	608756	Pontocerebellar hypoplasia, type 2F	Autosomal recessive

3	TSEN2	608753	Pontocerebellar hypoplasia, type 2B	Autosomal recessive
19	TSEN34	608754	Pontocerebellar hypoplasia type 2C	Autosomal recessive
17	TSEN54	608755	Pontocerebellar hypoplasia, type 2A; Pontocerebellar hypoplasia, type 4	Autosomal recessive
12	TSFM	604723	Combined oxidative phosphorylation deficiency, type 3	Autosomal recessive
1	TSHB	188540	Hypothyroidism, congenital, nongoitrous, type 4	Autosomal recessive
14	TSHR	603372	Hypothyroidism, congenital, nongoitrous, type 1	Autosomal recessive
17	TTC19	613814	Mitochondrial complex III deficiency, nuclear type 2	Autosomal recessive
2	TTC21B	612014	Short-rib thoracic dysplasia, type 4, with or without polydactyly	Autosomal recessive
2	TTC7A	609332	Gastrointestinal defects and immunodeficiency syndrome	Autosomal recessive
14	TTC8	608132	Bardet-Biedl syndrome, type 8	Autosomal recessive
8	TTI2	614426	Mental retardation, autosomal recessive, type 39	Autosomal recessive
14	TTLL5	612268	Cone-rod dystrophy 19	Autosomal recessive
2	TTN	188840	Limb-girdle muscular dystrophy type 10 (LGMDR10); Early-onset myopathy with fatal cardiomyopathy (Salih myopathy)	Autosomal recessive
8	TTPA	600415	Ataxia with isolated vitamin E deficiency	Autosomal recessive
22	TUBA8	605742	Cortical dysplasia, complex, with other brain malformations, type 8	Autosomal recessive
15	TUBGCP4	609610	Microcephaly and chorioretinopathy, autosomal recessive, type 3	Autosomal recessive
22	TUBGCP6	610053	Microcephaly and chorioretinopathy, autosomal recessive, type 1	Autosomal recessive
16	TUFM	602389	Combined oxidative phosphorylation deficiency 4	Autosomal recessive
6	TULP1	602280	Retinitis pigmentosa 14; Leber congenital amaurosis 15	Autosomal recessive
8	TUSC3	601385	Mental retardation, autosomal recessive, type 7	Autosomal recessive
2	TWIST2	607556	Focal facial dermal dysplasia, type 3 (Setleis type)	Autosomal recessive
10	TWNK	606075	Mitochondrial DNA depletion syndrome, type 7 (hepatocerebral type); Perrault syndrome type 5	Autosomal recessive
18	TXNL4A	611595	Burn-McKeown syndrome	Autosomal recessive
19	TYK2	176941	Immunodeficiency, type 35	Autosomal recessive
22	TYMP	131222	Mitochondrial DNA depletion syndrome, type 1 (MNGIE type)	Autosomal recessive
11	TYR	606933	Oculocutaneous albinism (OCA) type 1A; OCA type 1B	Autosomal recessive
19	TYROBP	604142	Polycystic lipomembranous osteodysplasia with sclerosing leukoencephalopathy, type 1 (Nasu-Hakola disease)	Autosomal recessive
9	TYRP1	115501	Albinism, oculocutaneous, type 3	Autosomal recessive
3	UBA5	610552	Epileptic encephalopathy, early infantile, 44	Autosomal recessive
1	UBE2T	610538	Fanconi anemia, complementation group T	Autosomal recessive
15	UBE3A	601623	Angelman syndrome	Autosomal dominant*
12	UBE3B	608047	Kaufman oculocerebrofacial syndrome	Autosomal recessive
15	UBR1	605981	Johanson-Blizzard syndrome	Autosomal recessive
4	UCHL1	191342	Spastic paraplegia, type 79, autosomal recessive	Autosomal recessive
13	UFM1	610553	Leukodystrophy, hypomyelinating, type 14	Autosomal recessive
2	UGT1A1	191740	Crigler-Najjar syndrome, type 1; Crigler-Najjar syndrome, type 2	Autosomal recessive
3	UMPS	613891	Orotic aciduria	Autosomal recessive
17	UNC13D	608897	Hemophagocytic lymphohistiocytosis, familial, type 3	Autosomal recessive
2	UNC80	612636	Hypotonia, infantile, with psychomotor retardation and characteristic facies, type 2	Autosomal recessive
12	UNG	191525	Immunodeficiency with hyper IgM, type 5	Autosomal recessive
22	UPB1	606673	Beta-ureidopropionase deficiency	Autosomal recessive
X	UPF3B	300298	Mental retardation, X-linked, syndromic, type 14	X-linked
8	UQCRB	191330	Mitochondrial complex III deficiency, nuclear, type 3	Autosomal recessive
16	UQCRC2	191329	Mitochondrial complex III deficiency, nuclear type 5	Autosomal recessive
5	UQCRQ	612080	Mitochondrial complex III deficiency, nuclear, type 4	Autosomal recessive
1	UROD	613521	Porphyria cutanea tarda	Autosomal recessive
10	UROS	606938	Porphyria, congenital erythropoietic	Autosomal recessive
16	USB1	613276	Poikiloderma with neutropenia	Autosomal recessive
11	USH1C	605242	Usher syndrome, type 1C; Deafness, autosomal recessive, type 18A	Autosomal recessive

17	USH1G	607696	Usher syndrome, type 1G	Autosomal recessive
1	USH2A	608400	Usher syndrome, type 2A; Retinitis pigmentosa 39	Autosomal recessive
22	USP18	607057	Pseudo-TORCH syndrome 2	Autosomal recessive
4	UVSSA	614632	UV-sensitive syndrome, type 3	Autosomal recessive
16	VAC14	604632	Striatonigral degeneration, childhood-onset	Autosomal recessive
6	VARS1	192150	Neurodevelopmental disorder with microcephaly, seizures, and cortical atrophy	Autosomal recessive
6	VARS2	612802	Combined oxidative phosphorylation deficiency 20	Autosomal recessive
12	VDR	601769	Rickets, vitamin D-resistant, type 2A	Autosomal recessive
14	VIPAS39	613401	Arthrogryposis, renal dysfunction and cholestasis, type 2	Autosomal recessive
16	VKORC1	608547	Vitamin K-dependent clotting factors, combined deficiency of, type 2	Autosomal recessive
9	VLDLR	192977	Cerebellar hypoplasia and mental retardation with or without quadrupedal locomotion, type 1	Autosomal recessive
9	VPS13A	605978	Choreoacanthocytosis	Autosomal recessive
8	VPS13B	607817	Cohen syndrome	Autosomal recessive
15	VPS13C	608879	Parkinson disease 23, autosomal recessive, early onset	Autosomal recessive
15	VPS33B	608552	Arthrogryposis, renal dysfunction and cholestasis, type 1	Autosomal recessive
8	VPS37A	609927	Spastic paraplegia, type 53, autosomal recessive	Autosomal recessive
1	VPS45	610035	Neutropenia, severe congenital, type 5	Autosomal recessive
17	VPS53	615850	Pontocerebellar hypoplasia, type 2E	Autosomal recessive
14	VRK1	602168	Pontocerebellar hypoplasia, type 1A	Autosomal recessive
14	VSX2	142993	Microphthalmia with coloboma 3; Isolated microphthalmia 2	Autosomal recessive
12	VWF	613160	von Willibrand disease, type 3	Autosomal recessive
1	WARS2	604733	Neurodevelopmental disorder, mitochondrial, with abnormal movements and lactic acidosis, with or without seizures	Autosomal recessive
X	WAS	300392	Wiskott-Aldrich syndrome; Thrombocytopenia, X-linked	X-linked
12	WASHC4	615748	?Mental retardation, autosomal recessive, type 43	Autosomal recessive
8	WASHC5	610657	Ritscher-Schinzel syndrome, type 1	Autosomal recessive
4	WDR19	608151	Nephronophthisis, type 13; Senior-Loken syndrome, type 8	Autosomal recessive
2	WDR35	613602	Cranioectodermal dysplasia 2	Autosomal recessive
17	WDR45B	609226	Neurodevelopmental disorder with spastic quadriplegia and brain abnormalities with or without seizures	Autosomal recessive
19	WDR62	613583	Microcephaly, type 2, primary, autosomal recessive, with or without cortical malformations	Autosomal recessive
15	WDR72	613214	Amelogenesis imperfecta, type 2A3 (hypomaturation type)	Autosomal recessive
15	WDR73	616144	Galloway-Mowat syndrome 1	Autosomal recessive
17	WDR81	614218	Cerebellar ataxia, mental retardation, and dysequilibrium syndrome, type 2	Autosomal recessive
7	WEE2	614084	Oocyte maturation defect 5	Autosomal recessive
4	WFS1	606201	Wolfram syndrome, type 1	Autosomal recessive
9	WHRN	607928	Usher syndrome, type 2D; Deafness, autosomal recessive, type 31	Autosomal recessive
2	WIPF1	602357	?Wiskott-Aldrich syndrome 2	Autosomal recessive
12	WNK1	605232	Neuropathy, hereditary sensory and autonomic, type 2	Autosomal recessive
12	WNT1	164820	Osteogenesis imperfecta, type XV	Autosomal recessive
2	WNT10A	606268	WNT10A-related conditions	Autosomal recessive
12	WNT10B	601906	Split-hand/foot malformation, type 6	Autosomal recessive
17	WNT3	165330	?Tetra-amelia syndrome	Autosomal recessive
3	WNT7A	601570	Fuhrmann syndrome	Autosomal recessive
17	WRAP53	612661	Dyskeratosis congenita, autosomal recessive, type 3	Autosomal recessive
8	WRN	604611	Werner syndrome	Autosomal recessive
16	WWOX	605131	Epileptic encephalopathy, early infantile, type 28; Spinocerebellar ataxia, autosomal recessive, type 12	Autosomal recessive
2	XDH	607633	Xanthinuria, type 1	Autosomal recessive
9	XPA	611153	Xeroderma pigmentosum, group A	Autosomal recessive
3	XPC	613208	Xeroderma pigmentosum, group C	Autosomal recessive

22	XPNPEP3	613553	Nephronophthisis-like nephropathy, type 1	Autosomal recessive
5	XRCC4	194363	Short stature, microcephaly, and endocrine dysfunction	Autosomal recessive
16	XYLT1	608124	Desbuquois dysplasia, type 2	Autosomal recessive
17	XYLT2	608125	Spondyloocular syndrome	Autosomal recessive
12	YARS2	610957	Myopathy, lactic acidosis, and sideroblastic anemia, type 2	Autosomal recessive
1	YY1AP1	607860	Grange syndrome	Autosomal recessive
2	ZAP70	176947	Autoimmune disease, multisystem, infantile-onset, type 2; Immunodeficiency, type 48	Autosomal recessive
11	ZBTB16	176797	Skeletal defects, genital hypoplasia, and mental retardation	Autosomal recessive
6	ZBTB24	614064	Immunodeficiency-centromeric instability-facial anomalies syndrome, type 2	Autosomal recessive
14	ZC3H14	613279	Mental retardation, autosomal recessive, type 56	Autosomal recessive
X	ZDHHC9	300646	Mental retardation, X-linked syndromic, Raymond type	X-linked
14	ZFYVE26	612012	Spastic paraplegia, type 15, autosomal recessive	Autosomal recessive
1	ZMPSTE24	606480	Mandibuloacral dysplasia with, type B lipodystrophy	Autosomal recessive
3	ZMYND10	607070	Ciliary dyskinesia, primary, type 22	Autosomal recessive
11	ZNF408	616454	Retinitis pigmentosa, type 72	Autosomal recessive
16	ZNF423	604557	Joubert syndrome, type 19	Autosomal recessive
16	ZNF469	612078	Brittle cornea syndrome, type 1	Autosomal recessive
X	ZNF711	314990	Mental retardation, X-linked, type 97	X-linked
17	ZNHIT3	604500	PEHO syndrome	Autosomal recessive
11	ZP1	195000	Oocyte maturation defect, type 1	Autosomal recessive