CGT Exome v1.0

chrom	OMIM (gene)	Gene	*Previous symbol*	OMIM (phen)	DISEASE	MOI
12	605378	AAAS		231550	Triple-A syndrome (achalasia-addisonianism-alacrimia)	Autosomal recessive
16	601065	AARS1	AARS	616339	Epileptic encephalopathy, early infantile, type 29	Autosomal recessive
6	612035	AARS2		614096; 615889	Combined oxidative phosphorylation deficiency 8; Leukoencephalopathy, progressive, with ovarian failure	Autosomal recessive
7	605113	AASS		238700; 268700	Hyperlysinemia, type 1 and type 2	Autosomal recessive
16	137150	ABAT		613163	GABA-transaminase deficiency	Autosomal recessive
9	600046	ABCA1		205400	Tangier disease	Autosomal recessive
2	607800	ABCA12		601277; 242500	Ichthyosis, congenital, autosomal recessive, type 4A; ICAR, type 4B (harlequin)	Autosomal recessive
16	601615	ABCA3		610921	Surfactant metabolism dysfunction, pulmonary, type 3	Autosomal recessive
1	601691	ABCA4		248200; 604116	Stargardt disease type 1; Cone-rod dystrophy type 3	Autosomal recessive
2	603201	ABCB11		605479; 601847	Cholestasis, benign recurrent intrahepatic, type 2; Cholestasis, progressive familial intrahepatic, type 2	Autosomal recessive
7	171060	ABCB4		602347	Cholestasis, progressive familial intrahepatic, type 3	Autosomal recessive
X	300135	ABCB7		301310	X-linked sideroblastic anemia and ataxia (XLSA/A)	X-linked
16	603234	ABCC6		264800; 614473	Pseudoxanthoma elasticum; Generalized arterial calcification of infancy, type 2	Autosomal recessive
11	600509	ABCC8		256450*; 606176*	Hyperinsulinemic hypoglycemia, type 1 (congenital hyperinsulinism); Permanent neonatal diabetes mellitus (PNDM)	Autosomal recessive*
X	300371	ABCD1		300100	Adrenoleukodystrophy	X-linked
14	603214	ABCD4		614857	Methylmalonic aciduria and homocystinuria, cblJ type	Autosomal recessive
2	605459	ABCG5		210250	Sitosterolemia	Autosomal recessive
2	605460	ABCG8		210250	Sitosterolemia	Autosomal recessive
20	613599	ABHD12		612674	PHARC syndrome (polyneuropathy, hearing loss, ataxia, retinitis pigmentosa and cataract)	Autosomal recessive
3	604780	ABHD5		275630	Chanarin-Dorfman syndrome	Autosomal recessive
11	604773	ACAD8		611283	Isobutyryl-CoA dehydrogenase deficiency	Autosomal recessive
3	611103	ACAD9		611126	Acyl-CoA dehydrogenase 9 deficiency (mitochondrial complex I deficiency, nuclear, type 20)	Autosomal recessive
1	607008	ACADM		201450	Medium-chain acyl-CoA dehydrogenase deficiency	Autosomal recessive
12	606885	ACADS		201470	Short-chain acyl-CoA dehydrogenase deficiency	Autosomal recessive
10	600301	ACADSB		610006	Short/branched-chain acyl-CoA dehydrogenase deficiency	Autosomal recessive
17	609575	ACADVL		201475	Very long-chain acyl-CoA dehydrogenase (VLCAD) deficiency	Autosomal recessive
11	607809	ACAT1		203750	Alpha-methylacetoacetic aciduria (3-ketothiolase deficiency)	Autosomal recessive
17	106180	ACE		267430	Renal tubular dysgenesis	Autosomal recessive
22	100850	ACO2		614559	Infantile cerebellar-retinal degeneration	Autosomal recessive
17	609751	ACOX1		264470	Peroxisomal acyl-CoA oxidase deficiency	Autosomal recessive
19	171640	ACP5		607944	Spondyloenchondrodysplasia with immune dysregulation	Autosomal recessive
16	614245	ACSF3		614265	Combined malonic and methylmalonic aciduria	Autosomal recessive
X	300157	ACSL4		300387	Mental retardation, X-linked, type 63	X-linked
1	102610	ACTA1		161800*; 255310*	Nemaline myopathy 3; Congenital fiber-type disproportion myopathy 1	Autosomal recessive*
3	104620	ACY1		609924	Aminoacylase 1 deficiency	Autosomal recessive
20	608958	ADA		102700	Severe combined immunodeficiency due to adenosine deaminase deficiency (ADA)	Autosomal recessive
8	602713	ADAM9		612775	Cone-rod dystrophy 9	Autosomal recessive
19	608990	ADAMTS10		277600	Weill-Marchesani syndrome, type 1, recessive	Autosomal recessive

9	604134	ADAMTS13		274150	Thrombotic thrombocytopenic purpura, familial (Schulman-Upshaw syndrome)	Autosomal recessive
15	607511	ADAMTS17		613195	Weill-Marchesani syndrome, type 4, recessive	Autosomal recessive
16	607512	ADAMTS18		615458	Microcornea, myopic chorioretinal atrophy, and telecanthus	Autosomal recessive
5	604539	ADAMTS2		225410	Ehlers-Danlos syndrome, dermatosparaxis type	Autosomal recessive
9	612277	ADAMTSL2		231050	Geleophysic dysplasia type 1	Autosomal recessive
1	610113	ADAMTSL4		225200; 225100	Ectopia lentis et pupillae; Ectopia lentis, isolated, type 2	Autosomal recessive
1	146920	ADAR		615010	Aicardi-Goutieres syndrome, type 6	Autosomal recessive
16	604110	ADGRG1	GPR56	606854	Polymicrogyria, bilateral frontoparietal	Autosomal recessive
5	602851	ADGRV1	GPR98	605472	Usher syndrome, type 2C	Autosomal recessive, digenic inheritance (PDZD7 gene)
10	102750	ADK		614300	Hypermethioninemia due to adenosine kinase deficiency	Autosomal recessive
22	608222	ADSL		103050	Adenylosuccinase deficiency	Autosomal recessive
X	300806	AFF2		309548	Mental retardation, X-linked, FRAXE type	X-linked
18	604581	AFG3L2		614487	Spastic ataxia, type 5, autosomal recessive	Autosomal recessive
4	613228	AGA		208400	Aspartylglucosaminuria (glycosylasparaginase deficiency)	Autosomal recessive
7	610345	AGK		614691; 212350	Cataract 38; Sengers syndrome	Autosomal recessive
1	610860	AGL		232400	Glycogen storage disease, type 3	Autosomal recessive
9	603100	AGPAT2		608594	Congenital generalized lipodystrophy (Berardinelli-Seip syndrome)	Autosomal recessive
2	603051	AGPS		600121	Rhizomelic chondrodysplasia punctata, type 3	Autosomal recessive
1	103320	AGRN		615120	Myasthenic syndrome, congenital, type 8	Autosomal recessive
1	106150	AGT		267430	Renal tubular dysgenesis	Autosomal recessive
3	106165	AGTR1		267430	Renal tubular dysgenesis	Autosomal recessive
2	604285	AGXT		259900	Hyperoxaluria, primary, type 1	Autosomal recessive
20	180960	AHCY		613752	Hypermethioninemia with deficiency of S-adenosylhomocysteine hydrolase	Autosomal recessive
6	608894	AHI1		608629	Joubert syndrome, type 3	Autosomal recessive
12	605257	AICDA		605258	Immunodeficiency with hyper-IgM, type 2	Autosomal recessive
X	300169	AIFM1		310490; 300614	Cowchock syndrome; Deafness, X-linked, type 5	X-linked
4	603605	AIMP1		260600	Leukodystrophy, hypomyelinating, type 3	Autosomal recessive
17	604392	AIPL1		604393	Leber congenital amaurosis, type 4	Autosomal recessive
21	607358	AIRE		240300*	Autoimmune polyendocrinopathy syndrome, type 1	Autosomal recessive*
9	103000	AK1		612631	Hemolytic anemia due to adenylate kinase deficiency	Autosomal recessive
1	103020	AK2		267500	Reticular dysgenesis	Autosomal recessive
10	600450	AKR1C2		614279	46,XY disorder of sex development due to testicular 17,20-desmolase deficiency	Autosomal recessive
7	604741	AKR1D1		235555	Bile acid synthesis defect, congenital, type 2	Autosomal recessive
9	125270	ALAD		612740	Porphyria, acute hepatic	Autosomal recessive
X	301300	ALAS2		300751	X-linked sideroblastic anemia, type 1 (XLSA or SIDBA1)	X-linked
10	138250	ALDH18A1		616586; 219150	Spastic paraplegia, type 9B, autosomal recessive; Cutis laxa, type 3A (De Barsy syndrome)	Autosomal recessive
17	609523	ALDH3A2		270200	Sjogren-Larsson syndrome	Autosomal recessive
1	606811	ALDH4A1		239510	Hyperprolinemia, type 2	Autosomal recessive
6	610045	ALDH5A1		271980	Succinic semialdehyde dehydrogenase deficiency	Autosomal recessive
14	603178	ALDH6A1		614105	Methylmalonate semialdehyde dehydrogenase deficiency	Autosomal recessive

5	107323	ALDH7A1		266100	Epilepsy, pyridoxine-dependent	Autosomal recessive
16	103850	ALDOA		611881	Glycogen storage disease type 12	Autosomal recessive
9	612724	ALDOB		229600	Fructose intolerance, hereditary	Autosomal recessive
16	605907	ALG1		608540	Congenital disorder of glycosylation, type 1K	Autosomal recessive
13	613666	ALG11		613661	Congenital disorder of glycosylation, type 1P	Autosomal recessive
22	607144	ALG12		607143	Congenital disorder of glycosylation, type 1G	Autosomal recessive
X	300776	ALG13		300884	Developmental and epileptic encephalopathy, type 36	X-linked
9	607905	ALG2		616228	Myasthenic syndrome, congenital, type 14, with tubular aggregates	Autosomal recessive
1	604566	ALG6		603147	Congenital disorder of glycosylation, type 1C	Autosomal recessive
11	608103	ALG8		608104	Congenital disorder of glycosylation, type 1H	Autosomal recessive
11	606941	ALG9		608776; 263210	Congenital disorder of glycosylation, type 1L; Gillessen-Kaesbach-Nishimura syndrome	Autosomal recessive
2	606844	ALMS1		203800	Alström syndrome	Autosomal recessive
17	603741	ALOX12B		242100	Ichthyosis, congenital, autosomal recessive, type 2	Autosomal recessive
17	607206	ALOXE3		606545	Ichthyosis, congenital, autosomal recessive, type 3	Autosomal recessive
1	171760	ALPL		241500; 241510	Hypophosphatasia, infantile/childhood	Autosomal recessive
2	606352	ALS2		205100; 606353; 607225	Amyotrophic lateral sclerosis, type 2, juvenile; Primary lateral sclerosis, juvenile; Spastic paralysis, infantile onset ascending	Autosomal recessive
12	601527	ALX1		613456	Frontonasal dysplasia, type 3	Autosomal recessive
1	606014	ALX3		136760	Frontonasal dysplasia, type 1	Autosomal recessive
11	605420	ALX4		613451	Frontonasal dysplasia, type 2	Autosomal recessive
5	604489	AMACR		214950; 614307	Bile acid synthesis defect, congenital, type 4; Alpha-methylacyl-CoA racemase deficiency	Autosomal recessive
X	300391	AMELX		301200	Amelogenesis imperfecta, type 1E (hypomaturation type)	X-linked
X	300647	AMER1		300373	Osteopathia striata with cranial sclerosis	X-linked
19	600957	AMH		261550	Persistent Mullerian duct syndrome, type 1	Autosomal recessive
14	605799	AMN		261100	Megaloblastic anemia 1 (Imerslund-Grasbeck syndrome)	Autosomal recessive
1	102770	AMPD1		615511	Myopathy due to myoadenylate deaminase deficiency	Autosomal recessive
3	238310	AMT		605899	Glycine encephalopathy	Autosomal recessive
1	604774	ANGPTL3		605019	Hypobetalipoproteinemia, familial, type 2	Autosomal recessive
3	613726	ANO10		613728	Spinocerebellar ataxia, autosomal recessive, type 10	Autosomal recessive
11	608662	ANO5		611307	Limb-girdle muscular dystrophy, type 12 (LGMD R12)	Autosomal recessive
X	300836	ANOS1	KAL1	308700	Hypogonadotropic hypogonadism, type 1, with or without anosmia (Kallmann syndrome 1)	X-linked
2	606410	ANTXR1		230740	GAPO syndrome	Autosomal recessive
4	608041	ANTXR2		228600	Hyaline fibromatosis syndrome	Autosomal recessive
7	603531	AP1S1		609313	MEDNIK syndrome	Autosomal recessive
X	300629	AP1S2		304340	Mental retardation, X-linked, syndromic, type 5 (Pettigrew syndrome)	X-linked
5	603401	AP3B1		608233	Hermansky-Pudlak syndrome, type 2	Autosomal recessive
15	602166	AP3B2		617276	Epileptic encephalopathy, early infantile, type 48	Autosomal recessive
1	607245	AP4B1		614066	Spastic paraplegia, type 47, autosomal recessive	Autosomal recessive
7	602296	AP4M1		612936	Spastic paraplegia, type 50, autosomal recessive	Autosomal recessive
14	607243	AP4S1		614067	Spastic paraplegia, type 52, autosomal recessive	Autosomal recessive
7	613653	AP5Z1		613647	Spastic paraplegia, type 48, autosomal recessive	Autosomal recessive
19	608083	APOC2		207750	Hyperlipoproteinemia, type 1B	Autosomal recessive
19	107741	APOE		269600	Sea-blue histiocyte disease	Autosomal recessive
16	102600	APRT		614723	Adenine phosphoribosyltransferase deficiency	Autosomal recessive

9	606350	APTX		208920	Ataxia, early-onset, with oculomotor apraxia and hypoalbuminemia	Autosomal recessive
12	107777	AQP2		125800*	Diabetes insipidus, nephrogenic, type 2	Autosomal recessive*
X	313700	AR		300068	Androgen insensitivity syndrome, complete	X-linked
20	605371	ARFGEF2		608097	Periventricular heterotopia with microcephaly	Autosomal recessive
6	608313	ARG1		207800	Argininemia (arginase deficiency)	Autosomal recessive
X	300429	ARHGEF9		300607	Developmental and epileptic encephalopathy, type 8	X-linked
3	608922	ARL13B		612291	Joubert syndrome type 8	Autosomal recessive
3	608845	ARL6		600151	Bardet-Biedl syndrome, type 3	Autosomal recessive
22	607574	ARSA		250100	Metachromatic leukodystrophy	Autosomal recessive
5	611542	ARSB		253200	Mucopolysaccharidosis, type 6 (Maroteaux-Lamy syndrome)	Autosomal recessive
X	300180	ARSL	ARSE	302950	Chondrodysplasia punctata, brachytelephalangic	X-linked
X	300382	ARX		308350; 300215; 309510	Epileptic encephalopathy, early infantile, type 1; ARX-related developmental disorders	X-linked
8	613468	ASAH1		228000; 159950	Farber lipogranulomatosis; Spinal muscular atrophy with progressive myoclonic epilepsy	Autosomal recessive
7	608310	ASL		207900	Argininosuccinic aciduria	Autosomal recessive
7	108370	ASNS		615574	Asparagine synthetase deficiency	Autosomal recessive
17	608034	ASPA		271900	Canavan disease	Autosomal recessive
1	605481	ASPM		608716	Primary microcephaly type 5, autosomal recessive	Autosomal recessive
9	603470	ASS1		215700	Citrullinemia, type 1	Autosomal recessive
1	605537	ATF6		616517	Achromatopsia, type 7	Autosomal recessive
2	601731	ATIC		608688	AICA-ribosiduria due to ATIC deficiency	Autosomal recessive
11	607585	ATM		208900	Ataxia-telangiectasia	Autosomal recessive
10	609875	ATOH7		221900	Persistent hyperplastic primary vitreous, autosomal recessive	Autosomal recessive
1	610513	ATP13A2		606693; 617225	Kufor-Rakeb syndrome; Spastic paraplegia, type 78, autosomal recessive	Autosomal recessive
16	108730	ATP2A1		601003	Brody myopathy	Autosomal recessive
12	611716	ATP6V0A2		219200; 278250	Cutis laxa, autosomal recessive, type 2A; Wrinkly skin syndrome	Autosomal recessive
7	605239	ATP6V0A4		602722	Renal tubular acidosis, distal, autosomal recessive	Autosomal recessive
2	192132	ATP6V1B1		267300	Renal tubular acidosis with deafness	Autosomal recessive
X	300011	ATP7A		309400; 304150	Menkes disease; Occipital horn syndrome	X-linked
13	606882	ATP7B		277900	Wilson disease	Autosomal recessive
18	602397	ATP8B1		211600; 243300	Cholestasis, progressive familial intrahepatic, type 1; Cholestasis, benign recurrent intrahepatic, type 1	Autosomal recessive
3	601215	ATR		210600	Seckel syndrome, type 1	Autosomal recessive
X	300504	ATRX		309580; 301040	Mental retardation-hypotonic facies syndrome, X-linked; Alpha-thalassemia/mental retardation syndrome	X-linked
9	600529	AUH		250950	3-methylglutaconic aciduria, type 1	Autosomal recessive
19	603495	AURKC		243060	Spermatogenic failure, type 5	Autosomal recessive
X	300538	AVPR2		304800; 300539	Diabetes insipidus, nephrogenic, type 1; Nephrogenic syndrome of inappropriate antidiuresis (NSIAD)	X-linked
15	109700	B2M		241600	Immunodeficiency, type 43	Autosomal recessive
11	606374	B3GAT3		245600	Multiple joint dislocations, short stature, craniofacial dysmorphism, with or without congenital heart defects	Autosomal recessive
13	610308	B3GLCT	B3GALTL	261540	Peters-plus syndrome	Autosomal recessive
9	137060	B4GALT1		607091	Congenital disorder of glycosylation, type 2D	Autosomal recessive
5	604327	B4GALT7		130070	Ehlers-Danlos syndrome, spondylodysplastic, type 1	Autosomal recessive
17	614144	B9D1		617120; 614209	Joubert syndrome, type 27; ?Meckel syndrome 9	Autosomal recessive
19	611951	B9D2		614175; 614175	Joubert syndrome, type 34; ?Meckel syndrome, type 10	Autosomal recessive
11	209901	BBS1		209900	Bardet-Biedl syndrome, type 1	Autosomal recessive

12	610148	BBS10		615987	Bardet-Biedl syndrome, type 10	Autosomal recessive
4	610683	BBS12		615989	Bardet-Biedl syndrome, type 12	Autosomal recessive
16	606151	BBS2		615981	Bardet-Biedl syndrome, type 2	Autosomal recessive
15	600374	BBS4		615982	Bardet-Biedl syndrome, type 4	Autosomal recessive
2	603650	BBS5		615983	Bardet-Biedl syndrome, type 5	Autosomal recessive
4	607590	BBS7		615984	Bardet-Biedl syndrome, type 7	Autosomal recessive
7	607968	BBS9		615986	Bardet-Biedl syndrome, type 9	Autosomal recessive
X	300398	BCAP31		300475	Deafness, dystonia, and cerebral hypomyelination	X-linked
19	608348	BCKDHA		248600	Maple syrup urine disease, type 1A	Autosomal recessive
6	248611	BCKDHB		248600	Maple syrup urine disease, type 1B	Autosomal recessive
X	300485	BCOR		300166	Microphthalmia, syndromic, type 2	X-linked
2	603647	BCS1L		256000	BCS1L-related disorders, including Leigh syndrome	Autosomal recessive
11	607854	BEST1		611809	Bestrophinopathy, AR	Autosomal recessive
17	615416	BHLHA9		609432	Syndactyly, mesoaxial synostotic, with phalangeal reduction	Autosomal recessive
2	601248	BIN1		255200	Centronuclear myopathy, type 2	Autosomal recessive
15	604610	BLM		210900	Bloom syndrome	Autosomal recessive
7	109750	BLVRA		614156*	Hyperbiliverdinemia	Autosomal recessive*
8	112264	BMP1		614856	Osteogenesis imperfecta, type 13	Autosomal recessive
X	300247	BMP15		300510	Ovarian dysgenesis 2	X-linked
7	608699	BMPER		608022	Diaphanospondylodysostosis	Autosomal recessive
4	603248	BMPR1B		609441	Acromesomelic dysplasia, Demirhan type	Autosomal recessive
2	613183	BOLA3		614299	Multiple mitochondrial dysfunctions syndrome 2 with hyperglycinemia	Autosomal recessive
7	613896	BPGM		222800	Erythrocytosis due to bisphosphoglycerate mutase deficiency	Autosomal recessive
8	614010	BPNT2	IMPAD1	614078	Chondrodysplasia with joint dislocations, GPAPP type	Autosomal recessive
7	614506	BRAT1		614498; 618056	Rigidity and multifocal seizure syndrome, lethal neonatal; Neurodevelopmental disorder with cerebellar atrophy and with or without seizures	Autosomal recessive
17	605882	BRIP1		609054	Fanconi anemia, complementation group J	Autosomal recessive
X	300553	BRWD3		300659	Mental retardation, X-linked, type 93	X-linked
11	606158	BSCL2		269700; 615924	Congenital generalized lipodystrophy, type 2; Encephalopathy, progressive, with or without lipodystrophy	Autosomal recessive
1	606412	BSND		602522	Bartter syndrome, type 4A	Autosomal recessive
3	609019	BTD		253260	Biotinidase deficiency	Autosomal recessive
X	300300	BTK		300755	Agammaglobulinemia X-linked, type 1	X-linked
15	602860	BUB1B		257300	Mosaic variegated aneuploidy syndrome 1	Autosomal recessive
12	615140	C12orf57		218340	Temtamy syndrome	Autosomal recessive
12	613541	C12orf65		613559	Combined oxidative phosphorylation deficiency 7; Spastic paraplegia, type 55, autosomal recessive	Autosomal recessive
19	614297	C19orf12		614298*	Neurodegeneration with brain iron accumulation, type 4	Autosomal recessive*
1	120550	C1QA		613652	C1q deficiency	Autosomal recessive
1	120570	C1QB		613652	C1q deficiency	Autosomal recessive
1	120575	C1QC		613652	C1q deficiency	Autosomal recessive
12	120580	C1S		613783	C1s deficiency	Autosomal recessive
19	120700	C3		613779	Complement component 3 deficiency	Autosomal recessive
9	120900	C5		609536	Complement component 5 deficiency	Autosomal recessive
5	217070	C7		610102	Complement component 7 deficiency	Autosomal recessive
1	120960	C8B		613789	Complement component 8 deficiency, type 2	Autosomal recessive

8	614477	C8orf37		617406; 614500	Bardet-Biedl syndrome, type 21; Cone-rod dystrophy 16 and Retintis pigmentosa 64	Autosomal recessive
15	603263	CA12		143860	Hyperchlorhidrosis, isolated	Autosomal recessive
8	611492	CA2		259730	Osteopetrosis with renal tubular acidosis (osteopetrosis, autosomal recessive, type 3)	Autosomal recessive
11	608965	CABP4		610427	Congenital stationary night blindness, type 2B	Autosomal recessive
3	114206	CACNA1D		614896	Sinoatrial node dysfunction and deafness	Autosomal recessive
X	300110	CACNA1F		300476; 300071; 300600	Cone-rod dystrophy, X-linked, type 3; Night blindness, congenital stationary, type 2A; Aland Island eye disease	X-linked
17	613165	CANT1		251450; 617719	Desbuquois dysplasia, type 1; Epiphyseal dysplasia, multiple, type 7	Autosomal recessive
15	114240	CAPN3		253600	Limb-girdle muscular dystrophy, type 1 (LGMD R1)	Autosomal recessive
9	607212	CARD9		212050	Candidiasis, familial, type 2, autosomal recessive	Autosomal recessive
X	300172	CASK		300749	Mental retardation, X-linked, syndromic, Najm type	X-linked
1	114251	CASQ2		611938	Ventricular tachycardia, catecholaminergic polymorphic, type 2	Autosomal recessive
3	601199	CASR		239200*	Hyperparathyroidism, neonatal	Autosomal recessive*
5	114090	CAST		616295	Peeling skin with leukonychia, acral punctate keratoses, cheilitis, and knuckle pads	Autosomal recessive
11	606389	CATSPER1		612997	Spermatogenic failure, type 7	Autosomal recessive
17	603198	CAVIN1	PTRF	613327	Lipodystrophy, congenital generalized, type 4	Autosomal recessive
11	609342	CBLIF	GIF	261000	Intrinsic factor deficiency	Autosomal recessive
21	613381	CBS		236200	Homocystinuria due to cystathionine beta-synthase	Autosomal recessive
19	610055	CC2D1A		608443	Mental retardation, autosomal recessive, type 3	Autosomal recessive
4	612013	CC2D2A		612285; 612284	Joubert syndrome, type 9; Meckel syndrome, type 6	Autosomal recessive
18	612753	CCBE1		235510	Hennekam lymphangiectasia-lymphedema syndrome, type 1	Autosomal recessive
17	614677	CCDC103		614679	Ciliary dyskinesia, primary, type 17	Autosomal recessive
3	613798	CCDC39		613807	Ciliary dyskinesia, primary, type 14	Autosomal recessive
17	613799	CCDC40		613808	Ciliary dyskinesia, primary, type 15	Autosomal recessive
14	611204	CCDC88C		236600	Hydrocephalus, congenital, type 1	Autosomal recessive
6	603400	CCN6	WISP3	208230	Arthropathy, progressive pseudorheumatoid, of childhood	Autosomal recessive
16	107265	CD19		613493	Immunodeficiency, common variable, type 3	Autosomal recessive
12	186711	CD27		615122	Lymphoproliferative syndrome 2	Autosomal recessive
11	186790	CD3D		615617	Immunodeficiency, type 19	Autosomal recessive
11	186830	CD3E		615615	Immunodeficiency, type 18	Autosomal recessive
11	186740	CD3G		615607	Immunodeficiency, type 17, CD3 gamma deficient	Autosomal recessive
20	109535	CD40		606843	Immunodeficiency with hyper-IgM, type 3	Autosomal recessive
X	300386	CD40LG		308230	Hyper-IgM syndrome, type 1 (immunodeficiency, X-linked, with hyper-IgM, type 1)	X-linked
1	125240	CD55		226300	Complement hyperactivation, angiopathic thrombosis, and protein-losing enteropathy (CHAPLE)	Autosomal recessive
11	107271	CD59		612300	CD59 deficiency	Autosomal recessive
19	112205	CD79A		613501	Agammaglobulinemia 3	Autosomal recessive
17	147245	CD79B		612692	Agammaglobulinemia 6	Autosomal recessive
11	186845	CD81		613496	Immunodeficiency, common variable, type 6	Autosomal recessive
2	186910	CD8A		608957	CD8 deficiency, familial	Autosomal recessive
15	607465	CDAN1		224120	Dyserythropoietic anemia, congenital, type 1A	Autosomal recessive
10	605516	CDH23		601386; 601067	Deafness, autosomal recessive, type 12; Usher syndrome, type 1D	Autosomal recessive
16	114021	CDH3		225280	Ectodermal dysplasia, ectrodactyly, and macular dystrophy	Autosomal recessive
10	609502	CDHR1		613660	Cone-rod dystrophy, type 15	Autosomal recessive
9	608201	CDK5RAP2		604804	Primary microcephaly type 3, autosomal recessive	Autosomal recessive

X	300203	CDKL5		300672	Developmental and epileptic encephalopathy, type 2	X-linked
16	605525	CDT1		613804	Meier-Gorlin syndrome, type 4	Autosomal recessive
13	609279	CENPJ		608393	Primary microcephaly type 6, autosomal recessive	Autosomal recessive
4	611423	CEP135		614673	Microcephaly 8, primary, autosomal recessive	Autosomal recessive
15	613529	CEP152		614852	Primary microcephaly type 9, autosomal recessive	Autosomal recessive
12	610142	CEP290		611134; 610188; 611755	Meckel syndrome, type 4; Joubert syndrome, type 5; Leber congenital amaurosis, type 10	Autosomal recessive
7	610523	CEP41		614464	Joubert syndrome, type 15	Autosomal recessive
11	607951	CEP57		614114	Mosaic variegated aneuploidy syndrome 2	Autosomal recessive
2	608381	CERKL		608380	Retinitis pigmentosa, type 26	Autosomal recessive
19	134350	CFD		613912	Complement factor D deficiency	Autosomal recessive
1	134370	CFH		609814	Complement factor H deficiency	Autosomal recessive
4	217030	CFI		610984	Complement factor I deficiency	Autosomal recessive
14	601443	CFL2		610687	Nemaline myopathy, type 7, autosomal recessive	Autosomal recessive
X	300383	CFP		312060	Properdin deficiency, X-linked	X-linked
7	602421	CFTR		219700	Cystic fibrosis	Autosomal recessive
10	118490	CHAT		254210	Myasthenic syndrome, congenital, type 6, presynaptic	Autosomal recessive
22	612395	CHKB		602541	Muscular dystrophy, congenital, megaconial type	Autosomal recessive
X	300390	CHM		303100	Choroideremia	X-linked
X	300350	CHRDL1		309300	Megalocornea 1, X-linked	X-linked
2	100720	CHRND		616322; 253290	Myasthenic syndrome, congenital, type 3B, fast-channel; Multiple pterygium syndrome, lethal type	Autosomal recessive
17	100725	CHRNE		616324; 608931	Myasthenic syndrome, congenital, type 4B, fast-channel; Myasthenic syndrome, congenital, type 4C, associated with acetylcholine receptor deficiency	Autosomal recessive
2	100730	CHRNG		265000; 253290	Multiple pterygium syndrome (MPS), Escobar type; MPS, lethal type	Autosomal recessive
15	608429	CHST14		601776	Ehlers-Danlos syndrome, musculocontractural, type 1	Autosomal recessive
10	603799	CHST3		143095	Spondyloepiphyseal dysplasia with congenital joint dislocations	Autosomal recessive
16	605294	CHST6		217800	Macular corneal dystrophy	Autosomal recessive
15	608183	CHSY1		605282	Temtamy preaxial brachydactyly syndrome	Autosomal recessive
10	600664	CHUK		613630	Cocoon syndrome	Autosomal recessive
15	605564	CIB2		609439;614869	Deafness, autosomal recessive, type 48; Usher syndrome, type 1J	Autosomal recessive
16	600005	CIITA		209920	Bare lymphocyte syndrome, type 2, complementation group A	Autosomal recessive
6	612325	CILK1	ICK	612651	Endocrine-cerebroosteodysplasia	Autosomal recessive
4	611507	CISD2		604928	Wolfram syndrome 2	Autosomal recessive
11	607672	CLCF1		610313	Cold-induced sweating syndrome 2	Autosomal recessive
7	118425	CLCN1		255700	Myotonia congenita, recessive	Autosomal recessive
3	600570	CLCN2		615651	Leukoencephalopathy with ataxia	Autosomal recessive
X	300008	CLCN5		300009; 300554	Dent disease; Hypophosphatemic rickets	X-linked
16	602727	CLCN7		611490	Osteopetrosis, autosomal recessive type 4	Autosomal recessive
1	602024	CLCNKA		613090	Bartter syndrome, type 4B, digenic	Digenic inheritance (CLCNKB gene)
1	602023	CLCNKB		607364; 613090	Bartter syndrome, type 3; Bartter syndrome, type 4B, digenic	Autosomal recessive; Digenic inheritance (CLCNKA gene)
3	603718	CLDN1		607626	Ichthyosis, leukocyte vacuoles, alopecia, and sclerosing cholangitis	Autosomal recessive
21	605608	CLDN14		614035	Deafness type 29, autosomal recessive	Autosomal recessive
3	603959	CLDN16		248250	Hypomagnesemia, type 3, renal	Autosomal recessive

1	610036	CLDN19		248190	Rena hypomagnesemia type 5, with ocular involvement	Autosomal recessive
11	611693	CLMP		615237	Congenital short bowel syndrome	Autosomal recessive
16	607042	CLN3		204200	Ceroid lipofuscinosis, neuronal, type 3	Autosomal recessive
13	608102	CLN5		256731	Ceroid lipofuscinosis, neuronal, type 5	Autosomal recessive
15	606725	CLN6		601780	Ceroid lipofuscinosis, neuronal, type 6	Autosomal recessive
8	607837	CLN8		600143	Ceroid lipofuscinosis, neuronal, type 8	Autosomal recessive
3	606397	CLRN1		276902	Usher syndrome, type 3A	Autosomal recessive
4	123825	CNGA1		613756	Retinitis pigmentosa type 49	Autosomal recessive
2	600053	CNGA3		216900	Achromatopsia, type 2	Autosomal recessive
16	600724	CNGB1		613767	Retinitis pigmentosa type 45	Autosomal recessive
8	605080	CNGB3		262300	Achromatopsia, type 3	Autosomal recessive
X	300724	CNKSR2		301008	Mental retardation, X-linked, syndromic, Houge type	X-linked
10	607803	CNNM2		616418*	Hypomagnesemia, seizures, and mental retardation	Autosomal recessive*
2	607805	CNNM4		217080	Jalili syndrome	Autosomal recessive
6	610774	CNPY3		617929	Epileptic encephalopathy, early infantile, type 60	Autosomal recessive
7	604569	CNTNAP2		610042	Pitt-Hopkins like syndrome 1	Autosomal recessive
16	606976	COG4		613489	Congenital disorder of glycosylation, type 2J	Autosomal recessive
7	606821	COG5		613612	Congenital disorder of glycosylation, type 2I	Autosomal recessive
13	606977	COG6		614576; 615328	Congenital disorder of glycosylation, type 2L; Shaheen syndrome	Autosomal recessive
16	606978	COG7		608779	Congenital disorder of glycosylation, type 2E	Autosomal recessive
16	606979	COG8		611182	Congenital disorder of glycosylation, type 2H	Autosomal recessive
1	120280	COL11A1		228520	Fibrochondrogenesis type 1	Autosomal recessive
10	113811	COL17A1		226650	Epidermolysis bullosa, junctional, non-Herlitz type	Autosomal recessive
21	120328	COL18A1		267750	Knobloch syndrome, type 1	Autosomal recessive
4	610004	COL25A1		616219	Fibrosis of extraocular muscles, congenital, type 5	Autosomal recessive
2	120070	COL4A3		203780	Alport syndrome, autosomal recessive, type 2	Autosomal recessive
2	120131	COL4A4		203780	Alport syndrome, autosomal recessive, type 2	Autosomal recessive
X	303630	COL4A5		301050	Alport syndrome, X-linked	X-linked
21	120220	COL6A1		254090*	Ullrich congenital muscular dystrophy, type 1 (Limb-girdle muscular dystrophy, type 22 [LGMD R22])	Autosomal recessive*
21	120240	COL6A2		254090*	Ullrich congenital muscular dystrophy, type 1 (Limb-girdle muscular dystrophy, type 22 [LGMD R22])	Autosomal recessive*
2	120250	COL6A3		254090*	Ullrich congenital muscular dystrophy, type 1 (Limb-girdle muscular dystrophy, type 22 [LGMD R22])	Autosomal recessive*
3	120120	COL7A1		226600; 604129*; 131850*	Dystrophic epidermolysis bullosa (DEB), Hallopeau-Siemens (HS) type and non-HS type; DEB pruriginosa; DEB pretibial	Autosomal recessive; Autosomal recessive*; Autosomal recessive*
6	120210	COL9A1		614134	Stickler syndrome, type 4	Autosomal recessive
2	612502	COLEC11		265050	3MC syndrome 2	Autosomal recessive
3	603033	COLQ		603034	Myasthenic syndrome, congenital, type 5	Autosomal recessive
4	609825	COQ2		607426	Primary coenzyme Q10 deficiency, type 1	Autosomal recessive
9	612898	COQ4		616276	Coenzyme Q10 deficiency, primary, type 7	Autosomal recessive
14	614647	COQ6		614650	Coenzyme Q10 deficiency, primary, type 6	Autosomal recessive
1	606980	COQ8A	ADCK3	612016	Primary coenzyme Q10 deficiency, type 4	Autosomal recessive
16	612837	COQ9		614654	Coenzyme Q10 deficiency, primary, type 5	Autosomal recessive
16	605000	CORO1A		615401	Immunodeficiency, type 8	Autosomal recessive
17	602125	COX10		619046	Mitochondrial complex IV deficiency, nuclear type 3	Autosomal recessive

10	603646	COX15		615119; 256000	Cardioencephalomyopathy, fatal infantile, due to cytochrome c oxidase deficiency, type 2; Leigh syndrome due to cytochrome c oxidase deficiency	Autosomal recessive
3	117700	CP		604290	Aceruloplasminemia	Autosomal recessive
8	609562	CPA6		614418	Febrile seizures, familial, type 11	Autosomal recessive
2	608307	CPS1		237300	Carbamoylphosphate synthetase 1 deficiency	Autosomal recessive
11	600528	CPT1A		255120	Carnitine palmitoyltransferase type 1A deficiency, hepatic	Autosomal recessive
1	600650	CPT2		608836; 600649	Carnitine palmitoyltransferase type 2 deficiency, lethal neonatal; Carnitine palmitoyltransferase type 2 deficiency, infantile	Autosomal recessive
1	120650	CR2		614699	Immunodeficiency, common variable, type 7	Autosomal recessive
12	603454	CRADD		614499	Mental retardation, autosomal recessive, type 34, with variant lissencephaly	Autosomal recessive
1	604210	CRB1		600105; 613835	Retinitis pigmentosa, type 12; Leber congenital amaurosis, type 8	Autosomal recessive
3	609262	CRBN		607417	Mental retardation, autosomal recessive, type 2	Autosomal recessive
19	604237	CRLF1		272430	Cold-induced sweating syndrome type 1	Autosomal recessive
7	614631	CRPPA	ISPD	614643; 616052	Muscular dystrophy-dystroglycanopathy, type A7; Muscular dystrophy-dystroglycanopathy, type C7	Autosomal recessive
3	605497	CRTAP		610682	Osteogenesis imperfecta, type 7	Autosomal recessive
21	123580	CRYAA		604219*	Cataract 9, multiple types	Autosomal recessive*
11	123590	CRYAB		613869; 613763*	Myopathy, myofibrillar, fatal infantile hypertonic, alpha-B crystallin-related; Cataract 16, multiple types	Autosomal recessive; Autosomal recessive*
22	600929	CRYBB1		611544*	Cataract 17	Autosomal recessive*
22	123630	CRYBB3		609741	Cataract 22	Autosomal recessive
22	138981	CSF2RB		614370	Surfactant metabolism dysfunction, pulmonary, type 5	Autosomal recessive
1	138971	CSF3R		617014	Neutropenia, severe congenital, type 7, autosomal recessive	Autosomal recessive
3	184600	CSTA		607936	Peeling skin syndrome, type 4	Autosomal recessive
21	601145	CSTB		254800	Epilepsy, progressive myoclonic type 1A (Unverricht and Lundborg)	Autosomal recessive
17	613129	CTC1		612199	Cerebroretinal microangiopathy with calcifications and cysts	Autosomal recessive
1	607657	CTH		219500	Cystathioninuria	Autosomal recessive
17	606272	CTNS		219800	Nephropathic cystinosis	Autosomal recessive
20	613111	CTSA		256540	Galactosialidosis	Autosomal recessive
11	602365	CTSC		245010; 245000	Haim-Munk syndrome; Papillon-Lefevre syndrome	Autosomal recessive
11	116840	CTSD		610127	Ceroid lipofuscinosis, neuronal, type 10	Autosomal recessive
1	601105	CTSK		265800	Pycnodysostosis	Autosomal recessive
10	602997	CUBN		261100	Megaloblastic anemia 1 (Imerslund-Grasbeck syndrome)	Autosomal recessive
X	300304	CUL4B		300354	Mental retardation, X-linked, syndromic, type 15 (Cabezas type)	X-linked
6	609577	CUL7		273750	3M syndrome 1	Autosomal recessive
18	613218	CYB5A		250790	46,XY disorder of sex development due to isolated 17,20-lyase deficiency	Autosomal recessive
22	613213	CYB5R3		250800	Methemoglobinemia, type 1; Methemoglobinemia, type 2	Autosomal recessive
16	608508	CYBA		233690	Chronic granulomatous disease, type 4	Autosomal recessive
X	300481	CYBB		306400	Chronic granulomatous disease, X-linked	X-linked
15	118485	CYP11A1		613743	46,XY disorder of sex development-adrenal insufficiency due to CYP11A1 deficiency	Autosomal recessive
8	610613	CYP11B1		202010	Adrenal hyperplasia, congenital, due to 11-beta-hydroxylase deficiency	Autosomal recessive
8	124080	CYP11B2		203400	Hypoaldosteronism, congenital, due to CMO I deficiency	Autosomal recessive
10	609300	CYP17A1		202110	17 alpha(α)-hydroxylase/17,20-lyase deficiency	Autosomal recessive
15	107910	CYP19A1		613546	Aromatase deficiency	Autosomal recessive
2	601771	CYP1B1		231300	Glaucoma, primary congenital, type 3A	Autosomal recessive
6	613815	CYP21A2		201910	Congenital adrenal hyperplasia due to 21-hydroxylase deficiency	Autosomal recessive

20	126065	CYP24A1		143880	Hypercalcemia, infantile, type 1	Autosomal recessive
2	606530	CYP27A1		213700	Cerebrotendinous xanthomatosis	Autosomal recessive
12	609506	CYP27B1		264700	Vitamin D-dependent rickets, type 1	Autosomal recessive
19	611495	CYP4F22		604777	Ichthyosis, congenital, autosomal recessive, type 5	Autosomal recessive
4	608614	CYP4V2		210370	Bietti crystalline corneoretinal dystrophy	Autosomal recessive
8	603711	CYP7B1		270800	Spastic paraplegia, type 5A, autosomal recessive	Autosomal recessive
2	609186	D2HGDH		600721	D-2-hydroxyglutaric aciduria	Autosomal recessive
3	128239	DAG1		616538; 613818	Muscular dystrophy-dystroglycanopathy type A9; Muscular dystrophy-dystroglycanopathy type C9	Autosomal recessive
1	610956	DARS2		611105	Leukoencephalopathy with brain stem and spinal cord involvement and lactate elevation	Autosomal recessive
9	609312	DBH		223360	Dopamine beta-hydroxylase deficiency	Autosomal recessive
1	248610	DBT		248600	Maple syrup urine disease, type 2	Autosomal recessive
2	612515	DCAF17		241080	Woodhouse-Sakati syndrome	Autosomal recessive
18	120470	DCC		617542	Gaze palsy, familial horizontal, with progressive scoliosis, type 2	Autosomal recessive
6	605755	DCDC2		617394; 616217	Sclerosing cholangitis, neonatal; Nephronophthisis 19	Autosomal recessive
10	605988	DCLRE1C		603554; 602450	Omenn syndrome; Severe combined immunodeficiency, Athabascan type	Autosomal recessive
X	300121	DCX		300067	Lissencephaly, X-linked, type 1	X-linked
11	600811	DDB2		278740	Xeroderma pigmentosum, complementation group E	Autosomal recessive
7	107930	DDC		608643	Aromatic L-amino acid decarboxylase deficiency	Autosomal recessive
1	191311	DDR2		271665	Spondylometaepiphyseal dysplasia, short limb-hand type	Autosomal recessive
12	601150	DDX11		613398	Warsaw breakage syndrome	Autosomal recessive
X	300160	DDX3X		300958	Mental retardation, X-linked, type 102	X-linked
2	125660	DES		601419*	Myopathy, myofibrillar, type 1	Autosomal recessive*
2	601465	DGUOK		251880	DGUOK-related mitochondrial DNA depletion syndrome	Autosomal recessive
1	606418	DHCR24		602398	Desmosterolosis	Autosomal recessive
11	602858	DHCR7		270400	Smith-Lemli-Opitz syndrome	Autosomal recessive
1	608172	DHDDS		613861	Retinitis pigmentosa, type 59	Autosomal recessive
5	126060	DHFR		613839	Megaloblastic anemia due to dihydrofolate reductase deficiency	Autosomal recessive
12	605423	DHH		233420	46,XY complete gonadal dysgenesis	Autosomal recessive
16	126064	DHODH		263750	Miller syndrome	Autosomal recessive
5	602121	DIAPH1		616632	Seizures, cortical blindness, microcephaly syndrome	Autosomal recessive
2	614184	DIS3L2		267000	Perlman syndrome	Autosomal recessive
X	300126	DKC1		305000	Dyskeratosis congenita, X-linked	X-linked
11	608770	DLAT		245348	Pyruvate dehydrogenase E2 deficiency	Autosomal recessive
7	238331	DLD		246900	Dihydrolipoamide dehydrogenase deficiency	Autosomal recessive
X	300189	DLG3		300850	Mental retardation, X-linked, type 90	X-linked
19	602768	DLL3		277300	Spondylocostal dysostosis type 1	Autosomal recessive
X	300377	DMD		310200; 300376	Duchenne/Becker muscular dystrophy	X-linked
4	600980	DMP1		241520	Hypophosphatemic rickets, autosomal recessive	Autosomal recessive
16	613190	DNAAF1		613193	Ciliary dyskinesia, primary, type 13	Autosomal recessive
14	612517	DNAAF2		612518	Ciliary dyskinesia, primary, type 10	Autosomal recessive
19	614566	DNAAF3		606763	Ciliary dyskinesia, primary, type 2	Autosomal recessive
15	608706	DNAAF4	DYX1C1	615482	Ciliary dyskinesia, primary, type 25	Autosomal recessive
7	614864	DNAAF5	HEATR2	614874	Ciliary dyskinesia, primary, type 18	Autosomal recessive
7	603339	DNAH11		611884	Ciliary dyskinesia, primary, type 7, with or without situs inversus	Autosomal recessive

5	603335	DNAH5	608644	Ciliary dyskinesia, primary, type 3, with or without situs inversus	Autosomal recessive
9	604366	DNAI1	244400	Ciliary dyskinesia, primary, type 1, with or without situs inversus	Autosomal recessive
17	605483	DNAI2	612444	Ciliary dyskinesia, primary, type 9, with or without situs inversus	Autosomal recessive
2	604139	DNAJB2	614881	Spinal muscular atrophy, distal, autosomal recessive, type 5	Autosomal recessive
3	608977	DNAJC19	610198	3-methylglutaconic aciduria, type 5	Autosomal recessive
1	608375	DNAJC6	615528	Parkinson disease, type 19A, juvenile-onset; Parkinson disease, type 19B, early-onset	Autosomal recessive
14	610062	DNAL1	614017	Ciliary dyskinesia, primary, type 16	Autosomal recessive
12	603850	DNM1L	614388*	Encephalopathy due to defective mitochondrial and peroxisomal fission, type 1	Autosomal recessive*
19	602378	DNM2	615368	Lethal congenital contracture syndrome, type 5	Autosomal recessive
20	602900	DNMT3B	242860	Immunodeficiency-centromeric instability-facial anomalies syndrome, type 1	Autosomal recessive
19	614194	DOCK6	614219	Adams-Oliver syndrome 2	Autosomal recessive
9	611432	DOCK8	243700	Hyper-IgE recurrent infection syndrome, autosomal recessive	Autosomal recessive
4	610285	DOK7	618389; 254300	Fetal akinesia deformation sequence, type 3; Myasthenic syndrome, congenital, type 10	Autosomal recessive
9	610746	DOLK	610768	Congenital disorder of glycosylation, type 1M	Autosomal recessive
11	191350	DPAGT1	608093; 614750	Congenital disorder of glycosylation, type 1J; Myasthenic syndrome, congenital, type 13	Autosomal recessive
20	603503	DPM1	608799	Congenital disorder of glycosylation, type 1E	Autosomal recessive
12	613893	DPY19L2	613958	Spermatogenic failure, type 9	Autosomal recessive
1	612779	DPYD	274270	Dihydropyrimidine dehydrogenase deficiency	Autosomal recessive
8	613326	DPYS	222748	Dihydropyrimidinuria	Autosomal recessive
18	125670	DSG1	615508	Erythroderma, congenital, with palmoplantar keratoderma, hypotrichosis, and hyper IgE	Autosomal recessive
18	607892	DSG4	607903	Hypotrichosis, type 6	Autosomal recessive
6	125647	DSP	605676; 609638	Cardiomyopathy, dilated, with woolly hair and keratoderma; Epidermolysis bullosa, lethal acantholytic	Autosomal recessive
6	113810	DST	615425	Epidermolysis bullosa simplex, autosomal recessive, type 2	Autosomal recessive
6	607145	DTNBP1	614076	Hermansky-Pudlak syndrome, type 7	Autosomal recessive
15	606759	DUOX2	607200	Thyroid dyshormonogenesis, type 6	Autosomal recessive
15	612772	DUOXA2	274900	Thyroid dyshormonogenesis, type 5	Autosomal recessive
18	607461	DYM	607326; 223800	Smith-McCort dysplasia; Dyggve-Melchior-Clausen disease	Autosomal recessive
11	603297	DYNC2H1	613091	Short-rib thoracic dysplasia, type 3, with or without polydactyly	Autosomal recessive
2	603009	DYSF	254130; 253601	Miyoshi muscular dystrophy, type 1; Limb-girdle muscular dystrophy, type 2 (LGMD R2)	Autosomal recessive
16	612799	EARS2	614924	Combined oxidative phosphorylation deficiency 12	Autosomal recessive
X	300205	EBP	300960; 302960	MEND syndrome; Chondrodysplasia punctata	X-linked
1	602201	ECM1	247100	Urbach-Wiethe disease	Autosomal recessive
X	300451	EDA	305100	Ectodermal dysplasia, type 1, hypohidrotic, X-linked	X-linked
2	604095	EDAR	224900	Ectodermal dysplasia 10B, hypohidrotic/hair/tooth type	Autosomal recessive
1	606603	EDARADD	614941	Ectodermal dysplasia 11B, hypohidrotic/hair/tooth type	Autosomal recessive
6	131240	EDN1	615706	Auriculocondylar syndrome, type 3	Autosomal recessive
20	131242	EDN3	613265	Waardenburg syndrome, type 4B	Autosomal recessive
13	131244	EDNRB	600501	ABCD syndrome	Autosomal recessive
11	604633	EFEMP2	614437	Cutis laxa, autosomal recessive, type 1B	Autosomal recessive
X	300035	EFNB1	304110	Craniofrontonasal dysplasia	X-linked
7	131550	EGFR	616069	?Inflammatory skin and bowel disease, neonatal, 2	Autosomal recessive
10	129010	EGR2	145900*	Dejerine-Sottas disease	Autosomal recessive*
2	604032	EIF2AK3	226980	Wolcott-Rallison syndrome	Autosomal recessive
14	606454	EIF2B2	603896	Leukoencephalopathy with vanishing white matter	Autosomal recessive

1	606273	EIF2B3		603896	Leukoencephalopathy with vanishing white matter	Autosomal recessive
2	606687	EIF2B4		603896	Leukoencephalopathy with vanishing white matter	Autosomal recessive
3	603945	EIF2B5		603896	Leukoencephalopathy with vanishing white matter	Autosomal recessive
17	605367	ELAC2		615440	Combined oxidative phosphorylation deficiency 17	Autosomal recessive
6	605512	ELOVL4		614457	Ichthyosis, spastic quadriplegia, and mental retardation	Autosomal recessive
9	603722	ELP1	IKBKAP	223900	Familial dysautonomia	Autosomal recessive
18	616054	ELP2		617270	Mental retardation, autosomal recessive, type 58	Autosomal recessive
X	300384	EMD		310300	Emery-Dreifuss muscular dystrophy, type 1, X-linked	X-linked
4	606585	ENAM		204650	Amelogenesis imperfecta, type 1C	Autosomal recessive
6	173335	ENPP1		208000	Arterial calcification, generalized, of infancy, type 1	Autosomal recessive
10	601752	ENTPD1		615683	Spastic paraplegia, type 64, autosomal recessive	Autosomal recessive
1	130500	EPB41		611804*	Elliptocytosis, type 1	Autosomal recessive*
15	177070	EPB42		612690	Spherocytosis, type 5	Autosomal recessive
2	185535	EPCAM		613217	Diarrhea 5, with tufting enteropathy, congenital	Autosomal recessive
6	607566	EPM2A		254780	Epilepsy, progressive myoclonic, type 2A (Lafora)	Autosomal recessive
12	190151	ERBB3		607598	Lethal congenital contractural syndrome, type 2	Autosomal recessive
19	126380	ERCC1		610758	Cerebrooculofacioskeletal syndrome, type 4	Autosomal recessive
19	126340	ERCC2		601675	Trichothiodystrophy, type 1	Autosomal recessive
2	133510	ERCC3		616390	Trichothiodystrophy, type 2	Autosomal recessive
13	133530	ERCC5		616570	Cerebrooculofacioskeletal syndrome, type 3	Autosomal recessive
10	609413	ERCC6		133540; 214150	Cockayne syndrome, type B; Cerebrooculofacioskeletal syndrome, type 1	Autosomal recessive
5	609412	ERCC8		216400	Cockayne syndrome, type A	Autosomal recessive
8	611605	ERLIN2		611225	Spastic paraplegia, type 18, autosomal recessive	Autosomal recessive
8	609353	ESCO2		268300	Roberts syndrome	Autosomal recessive
1	606351	ESPN		609006	Deafness, autosomal recessive, type 36	Autosomal recessive
6	133430	ESR1		615363	Estrogen resistance	Autosomal recessive
14	602167	ESRRB		608565	Deafness, autosomal recessive, type 35	Autosomal recessive
15	608053	ETFA		231680	Glutaric acidemia, type 2A	Autosomal recessive
19	130410	ETFB		231680	Glutaric acidemia, type 2B	Autosomal recessive
4	231675	ETFDH		231680	Glutaric acidemia, type 2C	Autosomal recessive
19	608451	ETHE1		602473	Ethylmalonic encephalopathy	Autosomal recessive
4	604831	EVC		225500	Ellis-van Creveld syndrome	Autosomal recessive
4	607261	EVC2		225500	Ellis-van Creveld syndrome	Autosomal recessive
9	606489	EXOSC3		614678	Pontocerebellar hypoplasia, type 1B	Autosomal recessive
8	608177	EXT1		215300	Chondrosarcoma	Autosomal recessive
8	605744	EXTL3		617425	Immunoskeletal dysplasia with neurodevelopmental abnormalities	Autosomal recessive
6	612424	EYS		602772	Retinitis pigmentosa, type 25	Autosomal recessive
13	613872	F10		227600	Factor X deficiency	Autosomal recessive
6	134570	F13A1		613225	Factor XIIIA deficiency	Autosomal recessive
1	134580	F13B		613235	Factor XIIIB deficiency	Autosomal recessive
11	176930	F2		613679	Prothrombin deficiency	Autosomal recessive
13	613878	F7		227500	Factor VII deficiency	Autosomal recessive
X	300841	F8		306700	Hemophilia A	X-linked
X	300746	F9		306900	Hemophilia B	X-linked

16	611026	FA2H	612319	Spastic paraplegia, type 35, autosomal recessive	Autosomal recessive
15	613871	FAH	276700	Tyrosinemia, type 1	Autosomal recessive
7	610531	FAM126A	610532	Leukodystrophy, hypomyelinating, type 5	Autosomal recessive
2	613596	FAM161A	606068	Retinitis pigmentosa, type 28	Autosomal recessive
17	611062	FAM20A	204690	Amelogenesis imperfecta, type 1G (Enamel-renal syndrome)	Autosomal recessive
7	611061	FAM20C	259775	Raine syndrome	Autosomal recessive
16	607139	FANCA	227650	Fanconi anemia, complementation group A	Autosomal recessive
X	300515	FANCB	300514	Fanconi anemia, complementation group B	X-linked
9	613899	FANCC	227645	Fanconi anemia, complementation group C	Autosomal recessive
3	613984	FANCD2	227646	Fanconi anemia, complementation group D2	Autosomal recessive
6	613976	FANCE	600901	Fanconi anemia, complementation group E	Autosomal recessive
11	613897	FANCF	603467	Fanconi anemia, complementation group F	Autosomal recessive
9	602956	FANCG	614082	Fanconi anemia, complementation group G	Autosomal recessive
15	611360	FANCI	609053	Fanconi anemia, complementation group I	Autosomal recessive
2	608111	FANCL	614083	Fanconi anemia, complementation group L	Autosomal recessive
6	611592	FARS2	614946; 617046	Combined oxidative phosphorylation deficiency 14; Spastic paraplegia, type 77, autosomal recessive	Autosomal recessive
14	604580	FBLN5	219100	Cutis laxa, autosomal recessive, type 1A	Autosomal recessive
9	611570	FBP1	229700	Fructose-1,6-bisphosphatase deficiency	Autosomal recessive
22	605648	FBXO7	260300	Parkinson disease, type 15, autosomal recessive	Autosomal recessive
18	612386	FECH	177000	Protoporphyria, erythropoietic, autosomal recessive	Autosomal recessive
20	607900	FERMT1	173650	Kindler syndrome	Autosomal recessive
11	607901	FERMT3	612840	Leukocyte adhesion deficiency, type 3	Autosomal recessive
4	134820	FGA	202400	Afibrinogenemia, congenital	Autosomal recessive
4	134830	FGB	202400	Congenital afibrinogenemia	Autosomal recessive
X	300546	FGD1	305400	Aarskog-Scott syndrome; Mental retardation, X-linked syndromic, type 16	X-linked
12	611104	FGD4	609311	Charcot-Marie-Tooth disease, type 4H	Autosomal recessive
12	605380	FGF23	617993	Tumoral calcinosis, hyperphosphatemic, familial, type 2	Autosomal recessive
11	164950	FGF3	610706	Deafness, congenital with inner ear agenesis, microtia, and microdontia	Autosomal recessive
4	134850	FGG	202400	Afibrinogenemia, congenital; Hypofibrinogenemia, congenital	Autosomal recessive
1	136850	FH	606812	Fumarase deficiency	Autosomal recessive
X	300163	FHL1	300696	Emery-Dreifuss muscular dystrophy, type 6, X-linked	X-linked
6	609390	FIG4	611228; 216340	Charcot-Marie-Tooth disease, type 4J; Yunis-Varon syndrome	Autosomal recessive
17	607063	FKBP10	259450	Bruck syndrome 1	Autosomal recessive
19	606596	FKRP	613153; 606612; 607155	Muscular dystrophy-dystroglycanopathy, type 5A (Walker-Warburg syndrome); Type 5B; Type 5C (limb-girdle muscular dystrophy, type 9 [LGMDR9])	Autosomal recessive
9	607440	FKTN	253800; 613152; 611588	Muscular dystrophy-dystroglycanopathy, type 4A (Walker-Warburg syndrome); Type 4B; Type 4C (limb-girdle muscular dystrophy, type 13 [LGMD R13])	Autosomal recessive
11	193067	FLI1	617443*	Bleeding disorder, platelet-type, type 21	Autosomal recessive*
X	300017	FLNA	305620	FLNA-related disorders	X-linked
3	603381	FLNB	272460	Spondylocarpotarsal synostosis syndrome	Autosomal recessive
1	609144	FLVCR1	609033	Posterior column ataxia-retinitis pigmentosa syndrome	Autosomal recessive
14	610865	FLVCR2	225790	Proliferative vasculopathy and hydranencephaly-hydrocephaly syndrome	Autosomal recessive
1	606373	FMN2	616193	Mental retardation, autosomal recessive, type 47	Autosomal recessive
1	136132	FMO3	602079	Trimethylaminuria	Autosomal recessive
X	309550	FMR1	300624	Fragile X syndrome	X-linked

11	136430	FOLR1	613068	Neurodegeneration due to cerebral folate transport deficiency	Autosomal recessive
9	602617	FOXE1	241850	Bamforth-Lazarus syndrome	Autosomal recessive
1	601094	FOXE3	610256	Anterior segment dysgenesis, type 2, multiple subtypes	Autosomal recessive
17	600838	FOXN1	601705	T-cell immunodeficiency, congenital alopecia and nail dystrophy	Autosomal recessive
X	300292	FOXP3	304790	Immunodysregulation, polyendocrinopathy, and enteropathy, X-linked	X-linked
11	613622	FOXRED1	618241	Mitochondrial complex I deficiency, nuclear type 19	Autosomal recessive
4	607830	FRAS1	219000	Fraser syndrome, type 1	Autosomal recessive
9	608944	FREM1	248450	Manitoba oculotrichoanal syndrome	Autosomal recessive
13	608945	FREM2	617666	Fraser syndrome, type 2	Autosomal recessive
X	300628	FRMD7	310700	Nystagmus 1, congenital, X-linked	X-linked
X	300838	FRMPD4	300983	Mental retardation, X-linked, type 104	X-linked
11	136530	FSHB	229070	Hypogonadotropic hypogonadism, type 24, without anosmia	Autosomal recessive
2	136435	FSHR	233300	Ovarian dysgenesis 1	Autosomal recessive
21	606806	FTCD	229100	Glutamate formiminotransferase deficiency	Autosomal recessive
19	134790	FTL	615604*	L-ferritin deficiency	Autosomal recessive*
16	610966	FTO	612938	Growth retardation, developmental delay, facial dysmorphism	Autosomal recessive
1	612280	FUCA1	230000	Fucosidosis	Autosomal recessive
14	602589	FUT8	618005	Congenital disorder of glycosylation with defective fucosylation, type 1	Autosomal recessive
9	606829	FXN	229300	Friedreich ataxia	Autosomal recessive
3	607182	FYCO1	610019	Cataract 18	Autosomal recessive
8	603409	FZD6	614157	Nail disorder, nonsyndromic congenital, type 10 (claw-shaped nails)	Autosomal recessive
17	613742	G6PC	232200	Glycogen storage disease, type 1A	Autosomal recessive
17	611045	G6PC3	612541	Dursun syndrome	Autosomal recessive
X	305900	G6PD	300908	Hemolytic anemia, G6PD deficient (favism)	X-linked
17	606800	GAA	232300	Glycogen storage disease, type 2	Autosomal recessive
14	606890	GALC	245200	Krabbe disease	Autosomal recessive
1	606953	GALE	230350	Galactose epimerase deficiency	Autosomal recessive
17	604313	GALK1	230200	Galactokinase deficiency with cataracts	Autosomal recessive
16	612222	GALNS	253000	Mucopolysaccharidosis, type 4A	Autosomal recessive
2	601756	GALNT3	211900	Tumoral calcinosis, hyperphosphatemic, familial, type 1	Autosomal recessive
9	606999	GALT	230400	Galactosemia	Autosomal recessive
19	601240	GAMT	612736	Cerebral creatine deficiency syndrome, type 2	Autosomal recessive
16	605379	GAN	256850	Giant axonal neuropathy, type 1	Autosomal recessive
X	305371	GATA1	300835; 314050; 300367	Anemia, X-linked, with/without neutropenia and/or platelet abnormalities; Thrombocytopenia with beta-thalassemia, X-linked; Thrombocytopenia, X-linked, with or without dyserythropoietic anemia	X-linked
15	602360	GATM	612718	Cerebral creatine deficiency syndrome, type 3	Autosomal recessive
1	606463	GBA	230800	Gaucher disease	Autosomal recessive
3	607839	GBE1	232500	Glycogen storage disease, type 4	Autosomal recessive
19	608801	GCDH	231670	Glutaricaciduria, type 1	Autosomal recessive
14	600225	GCH1	233910	Hyperphenylalaninemia, BH4-deficient, type B	Autosomal recessive
7	138079	GCK	606176*	Permanent neonatal diabetes mellitus (PNDM)	Autosomal recessive*
6	600429	GCNT2	116700	Cataract 13, with adult i phenotype	Autosomal recessive
8	606598	GDAP1	608340	Charcot-Marie-Tooth disease, recessive intermediate, type A	Autosomal recessive
19	602880	GDF1	208530	Right atrial isomerism (Ivemark syndrome)	Autosomal recessive

20	601146	GDF5	200700	Chondrodysplasia, Grebe type	Autosomal recessive
8	601147	GDF6	615360	Leber congenital amaurosis, type 17	Autosomal recessive
X	300104	GDI1	300849	Mental retardation, X-linked, type 41	X-linked
16	600924	GFER	613076	Myopathy, mitochondrial progressive, with congenital cataract, hearing loss, and developmental delay	Autosomal recessive
3	606639	GFM1	609060	Combined oxidative phosphorylation deficiency, type 1	Autosomal recessive
2	138292	GFPT1	610542	Myasthenia, congenital, type 12, with tubular aggregates	Autosomal recessive
2	137167	GGCX	277450	Vitamin K-dependent clotting factors, combined deficiency of, type 1	Autosomal recessive
17	139250	GH1	262400; 262650	Growth hormone deficiency, isolated, type 1A; Kowarski syndrome	Autosomal recessive
7	139191	GHRHR	612781	Growth hormone deficiency, isolated, type 1B	Autosomal recessive
3	601898	GHSR	615925	Growth hormone deficiency, isolated partial	Autosomal recessive
19	608792	GIPC3	601869	Deafness, autosomal recessive, type 15	Autosomal recessive
6	121014	GJA1	218400	Craniometaphyseal dysplasia, autosomal recessive	Autosomal recessive
X	304040	GJB1	302800	Charcot-Marie-Tooth neuropathy, X-linked dominant, type 1	X-linked
13	121011	GJB2	220290	Deafness, autosomal recessive, type 1A; Deafness, digenic, GJB2/GJB6	Autosomal recessive; Digenic inheritance (GJB6 gene)
13	604418	GJB6	612645; 220290	Deafness, autosomal recessive, type 1B; Deafness, digenic GJB2/GJB6	Autosomal recessive; Digenic inheritance (GJB2 gene)
1	608803	GJC2	613206	Spastic paraplegia, type 44, autosomal recessive	Autosomal recessive
X	300474	GK	307030	Glycerol kinase deficiency	X-linked
X	300644	GLA	301500	Fabry disease	X-linked
3	611458	GLB1	230500, 230600, 230650; 253010	GM1-gangliosidosis, types 1-3; Mucopolysaccharidosis, type 4B (Morquio)	Autosomal recessive
9	238300	GLDC	605899	Glycine encephalopathy	Autosomal recessive
9	603371	GLE1	253310; 611890	Lethal congenital contracture syndrome, type 1; Congenital arthrogryposis with anterior horn cell disease	Autosomal recessive
16	608539	GLIS2	611498	Nephronophthisis, type 7	Autosomal recessive
9	610192	GLIS3	610199	Diabetes mellitus, neonatal, with congenital hypothyroidism	Autosomal recessive
5	138491	GLRA1	149400*	Hyperekplexia, type 1	Autosomal recessive*
4	138492	GLRB	614619	Hyperekplexia, type 2	Autosomal recessive
14	609588	GLRX5	616860; 616859	Anemia, sideroblastic, type 3, pyridoxine-refractory; Spasticity, childhood-onset, with hyperglycinemia	Autosomal recessive
1	138290	GLUL	610015	Glutamine deficiency, congenital	Autosomal recessive
3	610516	GLYCTK	220120	D-glyceric aciduria	Autosomal recessive
5	613109	GM2A	272750	GM2-gangliosidosis, AB variant	Autosomal recessive
3	139330	GNAT1	616389	Night blindness, congenital stationary, type 1G	Autosomal recessive
1	139340	GNAT2	613856	Achromatopsia, type 4	Autosomal recessive
15	604447	GNB5	617173; 617182	Intellectual developmental disorder with cardiac arrhythmia; Language delay and ADHD/cognitive impairment with or without cardiac arrhythmia	Autosomal recessive
9	603824	GNE	605820	Inclusion body myopathy, type 2 (Nonaka myopathy)	Autosomal recessive
6	606628	GNMT	606664	Glycine N-methyltransferase deficiency	Autosomal recessive
1	602744	GNPAT	222765	Rhizomelic chondrodysplasia punctata, type 2	Autosomal recessive
12	607840	GNPTAB	252500; 252600	Mucolipidosis 2 alpha/beta; Mucolipidosis 3 alpha/beta	Autosomal recessive
16	607838	GNPTG	252605	Mucolipidosis III gamma	Autosomal recessive
4	138850	GNRHR	146110	Hypogonadotropic hypogonadism, type 7, without anosmia	Autosomal recessive
12	607664	GNS	252940	Mucopolysaccharidosis, type 3D (Sanfilippo syndrome D)	Autosomal recessive
1	607983	GORAB	231070	Geroderma osteodysplasticum	Autosomal recessive

17	604027	GOSR2		614018	Epilepsy, progressive myoclonic, type 6	Autosomal recessive
17	606672	GP1BA		231200	Bernard-Soulier syndrome, type A1	Autosomal recessive
22	138720	GP1BB		231200	Bernard-Soulier syndrome, type B	Autosomal recessive
19	605546	GP6		614201	Bleeding disorder, platelet-type, type 11	Autosomal recessive
3	173515	GP9		231200	Bernard-Soulier syndrome, type C	Autosomal recessive
X	300037	GPC3		312870	Simpson-Golabi-Behmel syndrome, type 1	X-linked
13	604404	GPC6		258315	Omodysplasia, type 1	Autosomal recessive
12	138420	GPD1		614480	Hypertriglyceridemia, transient infantile	Autosomal recessive
14	603930	GPHN		615501	Molybdenum cofactor deficiency C	Autosomal recessive
19	172400	GPI		613470	Hemolytic anemia, nonspherocytic, due to glucose phosphate isomerase deficiency	Autosomal recessive
8	612757	GPIHBP1		615947	Hyperlipoproteinemia, type 1D	Autosomal recessive
X	300808	GPR143		300500	Ocular albinism, type 1 (Nettleship-Falls type)	X-linked
17	614515	GPR179		614565	Night blindness, congenital stationary (complete), type 1E, autosomal recessive	Autosomal recessive
14	601404	GPR68		617217	Amelogenesis imperfecta, type 2A6 (hypomaturation type)	Autosomal recessive
1	609245	GPSM2		604213	Chudley-McCullough syndrome	Autosomal recessive
19	138322	GPX4		250220	Spondylometaphyseal dysplasia, Sedaghatian type	Autosomal recessive
8	608576	GRHL2		616029	Ectodermal dysplasia/short stature syndrome	Autosomal recessive
9	604296	GRHPR		260000	Hyperoxaluria, primary, type 2	Autosomal recessive
X	305915	GRIA3		300699	Mental retardation, X-linked, type 94	X-linked
6	138244	GRIK2		611092	Mental retardation, autosomal recessive, type, 6	Autosomal recessive
9	138249	GRIN1		617820	Neurodevelopmental disorder with or without hyperkinetic movements and seizures, autosomal recessive	Autosomal recessive
12	604597	GRIP1		617667	Fraser syndrome 3	Autosomal recessive
6	604473	GRM1		614831	Spinocerebellar ataxia, autosomal recessive, type 13	Autosomal recessive
5	604096	GRM6		257270	Night blindness, congenital stationary (complete), type 1B, autosomal recessive	Autosomal recessive
17	138945	GRN		614706	Ceroid lipofuscinosis, neuronal, type 11	Autosomal recessive
4	613283	GRXCR1		613285	Deafness, autosomal recessive, type 25	Autosomal recessive
14	138890	GSC		602471	Short stature, auditory canal atresia, mandibular hypoplasia, skeletal abnormalities	Autosomal recessive
20	601002	GSS		266130	Glutathione synthetase deficiency	Autosomal recessive
6	608780	GTF2H5		616395	Trichothiodystrophy, type 3, photosensitive	Autosomal recessive
12	601330	GUCY2C		614665	Meconium ileus	Autosomal recessive
17	600179	GUCY2D		204000	Leber congenital amaurosis, type 1	Autosomal recessive
7	611499	GUSB		253220	Mucopolysaccharidosis, type 7	Autosomal recessive
3	603942	GYG1		616199	Polyglucosan body myopathy, type 2	Autosomal recessive
19	138570	GYS1		611556	Glycogen storage disease, type 0, muscle	Autosomal recessive
12	138571	GYS2		240600	Glycogen storage disease, type 0, liver	Autosomal recessive
1	138090	H6PD		604931	Cortisone reductase deficiency 1	Autosomal recessive
6	610876	HACE1		616756	Spastic paraplegia and psychomotor retardation with or without seizures	Autosomal recessive
4	601609	HADH		231530	3-hydroxyacyl-CoA dehydrogenase deficiency	Autosomal recessive
2	600890	HADHA		609016; 609015	Long-chain 3-hydroxyl-CoA dehydrogenase (LCHAD) deficiency; Mitochondrial trifunctional protein deficiency	Autosomal recessive
2	143450	HADHB		609015	Mitochondrial trifunctional protein deficiency	Autosomal recessive
19	606464	HAMP		613313	Hemochromatosis, type 2B	Autosomal recessive
5	142810	HARS1	HARS	614504	Usher syndrome, type 3B	Autosomal recessive
1	605998	HAX1		610738	Neutropenia, severe congenital, type 3, autosomal recessive	Autosomal recessive

16	141800	HBA1		604131	Thalassemia, alpha-	Autosomal recessive
16	141850	HBA2		604131	Thalassemia, alpha-	Autosomal recessive
11	141900	HBB		603903	HBB-related hemoglobinopathy	Autosomal recessive
X	300056	HCCS		309801	Linear skin defects with multiple congenital anomalies, type 1	X-linked
X	300269	HDAC8		300882	Cornelia de Lange syndrome 5	X-linked
11	611642	HEPACAM		613925	Megalencephalic leukoencephalopathy with subcortical cysts 2A	Autosomal recessive
15	605837	HERC2		615516	Mental retardation, autosomal recessive, type 38	Autosomal recessive
17	608059	HES7		613686	Spondylocostal dysostosis, type 4, autosomal recessive	Autosomal recessive
3	601802	HESX1		182230	Growth hormone deficiency with pituitary anomalies	Autosomal recessive
15	606869	HEXA		272800	Tay-Sachs disease	Autosomal recessive
5	606873	HEXB		268800	Sandhoff disease, infantile, juvenile, and adult forms	Autosomal recessive
3	607474	HGD		203500	Alkaptonuria	Autosomal recessive
7	142409	HGF	DFNB39	608265	Deafness, autosomal recessive, type 39	Autosomal recessive
8	610453	HGSNAT		252930	Mucopolysaccharidosis type 3C (Sanfilippo syndrome C)	Autosomal recessive
2	610690	HIBCH		250620	3-hydroxyisobutryl-CoA hydrolase deficiency	Autosomal recessive
10	142600	HK1		605285	Charcot-Marie-Tooth disease, type 4G	Autosomal recessive
21	609018	HLCS		253270	Holocarboxylase synthetase deficiency	Autosomal recessive
1	613898	HMGCL		246450	HMG-CoA lyase deficiency	Autosomal recessive
1	600234	HMGCS2		605911	HMG-CoA synthase-2 deficiency	Autosomal recessive
4	142992	HMX1		612109	Oculoauricular syndrome	Autosomal recessive
2	605238	HNMT		616739	Mental retardation, autosomal recessive, type 51	Autosomal recessive
10	613597	HOGA1		613616	Hyperoxaluria, primary, type 3	Autosomal recessive
7	142955	HOXA1		601536	Athabaskan brainstem dysgenesis syndrome	Autosomal recessive
12	609695	HPD		276710	Tyrosinemia, type 3	Autosomal recessive
4	601688	HPGD		259100	Hypertrophic osteoarthropathy, primary, type 1 (pachydermoperiostosis)	Autosomal recessive
X	308000	HPRT1		300322	Lesch-Nyhan syndrome	X-linked
10	604982	HPS1		203300	Hermansky-Pudlak syndrome, type 1	Autosomal recessive
3	606118	HPS3		614072	Hermansky-Pudlak syndrome, type 3	Autosomal recessive
22	606682	HPS4		614073	Hermansky-Pudlak syndrome, type 4	Autosomal recessive
11	607521	HPS5		614074	Hermansky-Pudlak syndrome, type 5	Autosomal recessive
10	607522	HPS6		614075	Hermansky-Pudlak syndrome, type 6	Autosomal recessive
10	613469	HPSE2		236730	Urofacial syndrome, type 1	Autosomal recessive
8	602302	HR		203655; 209500	Alopecia universalis; Atrichia with papular lesions	Autosomal recessive
16	614232	HSD11B2		218030	Apparent mineralocorticoid excess	Autosomal recessive
X	300256	HSD17B10		300438	HSD10 mitochondrial disease	X-linked
9	605573	HSD17B3		264300	46,XY disorder of sex development due to 17-beta-hydroxysteroid dehydrogenase 3 deficiency	Autosomal recessive
5	601860	HSD17B4		261515	D-bifunctional protein deficiency	Autosomal recessive
1	613890	HSD3B2		201810	Adrenal hyperplasia, congenital, due to 3-beta-hydroxysteroid dehydrogenase 2 deficiency	Autosomal recessive
16	607764	HSD3B7		607765	Bile acid synthesis defect, congenital, type 1	Autosomal recessive
5	600548	HSPA9		616854	Even-plus syndrome	Autosomal recessive
2	118190	HSPD1		612233	Leukodystrophy, hypomyelinating, type 4	Autosomal recessive
1	142461	HSPG2		224410	Dyssegmental dysplasia, Silverman-Handmaker type	Autosomal recessive
10	602194	HTRA1		600142	CARASIL syndrome	Autosomal recessive
2	606441	HTRA2		617248	3-methylglutaconic aciduria, type 8	Autosomal recessive

X	300697	HUWE1	300706	Mental retardation, X-linked syndromic, Turner type	X-linked
11	610693	HYLS1	236680	Hydrolethalus syndrome	Autosomal recessive
20	604526	IDH3B	612572	Retinitis pigmentosa, type 46	Autosomal recessive
X	300823	IDS	309900	Mucopolysaccharidosis, type 2	X-linked
4	252800	IDUA	607014; 607015; 607016	Mucopolysaccharidosis type 1	Autosomal recessive
18	609382	IER3IP1	614231	Microcephaly, epilepsy, and diabetes syndrome	Autosomal recessive
6	107470	IFNGR1	209950	Immunodeficiency, type 27A, mycobacteriosis	Autosomal recessive
21	147569	IFNGR2	614889	Immunodeficiency, type 28, mycobacteriosis	Autosomal recessive
3	606045	IFT122	218330	Cranioectodermal dysplasia 1	Autosomal recessive
16	614620	IFT140	617781; 266920	Retinitis pigmentosa, type 80; Short-rib thoracic dysplasia 9 with or without polydactyly	Autosomal recessive
3	611177	IFT80	611263	Short-rib thoracic dysplasia, type 2, with or without polydactyly	Autosomal recessive
12	147440	IGF1	608747	Growth retardation with deafness and mental retardation due to IGF1 deficiency	Autosomal recessive
15	147370	IGF1R	270450*	Insulin-like growth factor I, resistance to	Autosomal recessive*
16	601489	IGFALS	615961	Acid-labile subunit deficiency	Autosomal recessive
4	602867	IGFBP7	614224	Retinal arterial macroaneurysm with supravalvular pulmonic stenosis	Autosomal recessive
11	600502	IGHMBP2	616155	Charcot-Marie-Tooth disease, axonal, type 2S	Autosomal recessive
22	146770	IGLL1	613500	Agammaglobulinemia 2	Autosomal recessive
2	600726	IHH	607778	Acrocapitofemoral dysplasia	Autosomal recessive
X	300248	IKBKG	300636	Immunodeficiency, type 33	X-linked
11	146933	IL10RA	613148	Inflammatory bowel disease, type 28, early onset, autosomal recessive	Autosomal recessive
21	123889	IL10RB	612567	Inflammatory bowel disease, type 25, early onset, autosomal recessive	Autosomal recessive
9	600939	IL11RA	614188	Craniosynostosis and dental anomalies	Autosomal recessive
5	161561	IL12B	614890	Immunodeficiency, type 29, mycobacteriosis	Autosomal recessive
19	601604	IL12RB1	614891	Immunodeficiency, type 30	Autosomal recessive
22	605461	IL17RA	613953	Immunodeficiency, type 51	Autosomal recessive
X	300206	IL1RAPL1	300143	Mental retardation, X-linked, type 21/34	X-linked
2	147679	IL1RN	612852	Sterile multifocal osteomyelitis with periostitis and pustulosis	Autosomal recessive
16	605383	IL21R	615207	Immunodeficiency, type 56	Autosomal recessive
10	147730	IL2RA	606367	Immunodeficiency, type 41, with lymphoproliferation and autoimmunity	Autosomal recessive
X	308380	IL2RG	300400	Severe combined immunodeficiency, X-linked	X-linked
2	605507	IL36RN	614204	Psoriasis, type 14, pustular	Autosomal recessive
5	146661	IL7R	608971	Severe combined immunodeficiency, T-cell negative, B-cell/natural killer cell-positive type	Autosomal recessive
3	609739	ILDR1	609646	Deafness, autosomal recessive, type 42	Autosomal recessive
3	607056	IMPG2	613581	Retinitis pigmentosa, type 56	Autosomal recessive
9	613037	INPP5E	213300	Joubert syndrome, type 1	Autosomal recessive
11	176730	INS	606176*	Permanent neonatal diabetes mellitus (PNDM)	Autosomal recessive*
19	147670	INSR	610549	Diabetes mellitus, insulin-resistant, with acanthosis nigricans, type A	Autosomal recessive
9	243305	INVS	602088	Nephronophthisis, type 2, infantile	Autosomal recessive
3	609237	IQCB1	609254	Senior-Loken syndrome, type 5	Autosomal recessive
X	300522	IQSEC2	309530	Mental retardation, X-linked, type 1/78	X-linked
12	606883	IRAK4	607676	Immunodeficiency, type 67 (IRAK4 deficiency)	Autosomal recessive
16	601565	IRF8	614894	Immunodeficiency, type 32B, monocyte and dendritic cell deficiency	Autosomal recessive
16	606195	IRX5	611174	Hamamy syndrome	Autosomal recessive
12	611911	ISCU	255125	Myopathy with lactic acidosis, hereditary	Autosomal recessive

20	606409	ITCH		613385	Autoimmune disease, multisystem, with facial dysmorphism	Autosomal recessive
17	607759	ITGA2B		273800	Glanzmann thrombasthenia	Autosomal recessive
17	605025	ITGA3		614748	Interstitial lung disease, nephrotic syndrome, and epidermolysis bullosa, congenital	Autosomal recessive
2	147556	ITGA6		226730	Epidermolysis bullosa, junctional, with pyloric stenosis	Autosomal recessive
12	600536	ITGA7		613204	Muscular dystrophy, congenital, due to ITGA7 deficiency	Autosomal recessive
21	600065	ITGB2		116920	Leukocyte adhesion deficiency	Autosomal recessive
17	173470	ITGB3		273800	Glanzmann thrombasthenia	Autosomal recessive
17	147557	ITGB4		226730	Epidermolysis bullosa, junctional, with pyloric atresia	Autosomal recessive
5	186973	ITK		613011	Lymphoproliferative syndrome 1	Autosomal recessive
20	147520	ITPA		616647	Epileptic encephalopathy, early infantile, type 35	Autosomal recessive
3	147265	ITPR1		206700*	Gillespie syndrome	Autosomal recessive*
15	607036	IVD		243500	Isovaleric acidemia	Autosomal recessive
6	612025	IYD		274800	Thyroid dyshormonogenesis, type 4	Autosomal recessive
19	600173	JAK3		600802	Severe Combined Immunodeficiency, autosomal recessive, T-negative/B-positive type	Autosomal recessive
11	606871	JAM3		613730	Hemorrhagic destruction of the brain, subependymal calcification, and cataracts	Autosomal recessive
17	173325	JUP		601214	Naxos disease	Autosomal recessive
16	601421	KARS1	KARS	613916	Deafness, autosomal recessive, type 89	Autosomal recessive
21	176261	KCNE1		612347	Jervell and Lange-Nielsen syndrome 2	Autosomal recessive
11	600359	KCNJ1		241200	Bartter syndrome, type 2	Autosomal recessive
1	602208	KCNJ10		612780	SESAME syndrome	Autosomal recessive
11	600937	KCNJ11		601820; 606176*	Hyperinsulinemic hypoglycemia, type 2 (congenital hyperinsulinism); Permanent neonatal diabetes mellitus (PNDM)	Autosomal recessive; Autosomal recessive*
2	603208	KCNJ13		614186	Leber congenital amaurosis, type 16	Autosomal recessive
9	607604	KCNV2		610356	Retinal cone dystrophy, type 3B	Autosomal recessive
7	611725	KCTD7		611726	Epilepsy, progressive myoclonic, type 3, with or without intracellular inclusions	Autosomal recessive
X	314690	KDM5C		300534	Mental retardation, X-linked, syndromic, Claes-Jensen type	X-linked
X	300128	KDM6A		300867	Kabuki syndrome, type 2	X-linked
12	603288	KERA		217300	Cornea plana 2, autosomal recessive	Autosomal recessive
6	611687	KHDC3L		614293	Hydatidiform mole, recurrent, type 2	Autosomal recessive
2	601255	KIF1A		614213; 610357	Neuropathy, hereditary sensory, type 2C; Spastic paraplegia, type 30, autosomal recessive	Autosomal recessive
15	611254	KIF7		200990	Acrocallosal syndrome; Joubert syndrome, type 12	Autosomal recessive
10	609367	KIFBP	KIF1BP; KIAA1279	609460	Goldberg-Shprintzen megacolon syndrome	Autosomal recessive
19	604161	KISS1R		614837	Hypogonadotropic hypogonadism, type 8, with or without anosmia	Autosomal recessive
5	605775	KLHL3		614495	Pseudohypoaldosteronism, type 2D	Autosomal recessive
7	611119	KLHL7		617055	Cold-induced sweating syndrome 3	Autosomal recessive
19	603767	KLK4		204700	Amelogenesis imperfecta, type 2A1 (hypomaturation type)	Autosomal recessive
4	229000	KLKB1		612423	Fletcher factor (prekallikrein) deficiency	Autosomal recessive
15	609173	KNL1	CASC5	604321	Microcephaly 4, primary, autosomal recessive	Autosomal recessive
17	148080	KRT10		113800*	Epidermolytic hyperkeratosis	Autosomal recessive*
17	148066	KRT14		601001	Epidermolysis bullosa simplex, autosomal recessive, type 1	Autosomal recessive
12	148040	KRT5		601001	Epidermolysis bullosa simplex, autosomal recessive, type 1	Autosomal recessive
2	605197	KYNU		617661	Vertebral, cardiac, renal, and limb defects syndrome, type 2	Autosomal recessive
X	308840	L1CAM		307000; 303350; 304100	L1 Syndrome	X-linked
14	609584	L2HGDH		236792	L-2-hydroxyglutaric aciduria	Autosomal recessive

18	150320	LAMA1		615960	Poretti-Boltshauser syndrome	Autosomal recessive
6	156225	LAMA2		607855; 618138	LAMA2-related muscular dystrophy	Autosomal recessive
18	600805	LAMA3		226700; 226650	Junctional epidermolysis bullosa (JEB) Herlitz type; JEB non-Herlitz type	Autosomal recessive
7	150240	LAMB1		615191	Lissencephaly, type 5	Autosomal recessive
3	150325	LAMB2		609049; 614199	Pierson syndrome; Nephrotic syndrome, type 5, with or without ocular abnormalities	Autosomal recessive
1	150310	LAMB3		226700; 226650	Junctional epidermolysis bullosa (JEB) Herlitz type; JEB non-Herlitz type	Autosomal recessive
1	150292	LAMC2		226700; 226650	Junctional epidermolysis bullosa (JEB) Herlitz type; JEB non-Herlitz type	Autosomal recessive
9	604349	LAMC3		614115	Cortical malformations, occipital	Autosomal recessive
X	309060	LAMP2		300257	Danon disease	X-linked
22	603590	LARGE1	LARGE	613154; 608840	Muscular dystrophy-dystroglycanopathy, type 6A and 6B	Autosomal recessive
3	604544	LARS2		615300	Perrault syndrome, type 4	Autosomal recessive
1	600024	LBR		215140	Greenberg skeletal dysplasia	Autosomal recessive
6	611408	LCA5		604537	Leber congenital amaurosis, type 5	Autosomal recessive
16	606967	LCAT		245900; 136120	Familial LCAT deficiency; Fish-eye disease	Autosomal recessive
2	603202	LCT		223000	Lactase deficiency, congenital	Autosomal recessive
11	150000	LDHA		612933	Glycogen storage disease type 11	Autosomal recessive
1	605747	LDLRAP1		603813	Hypercholesterolemia, familial, autosomal recessive	Autosomal recessive
7	164160	LEP		614962	Obesity, morbid, due to leptin deficiency	Autosomal recessive
1	601007	LEPR		614963	Obesity, morbid, due to leptin receptor deficiency	Autosomal recessive
19	152780	LHB		228300	Hypogonadotropic hypogonadism, type 23, with or without anosmia	Autosomal recessive
2	152790	LHCGR		238320	Leydig cell hypoplasia	Autosomal recessive
6	609427	LHFPL5		610265	Deafness, autosomal recessive, type 67	Autosomal recessive
9	600577	LHX3		221750	Pituitary hormone deficiency, combined, type 3	Autosomal recessive
4	607031	LIAS		614462	Hyperglycinemia, lactic acidosis, and seizures	Autosomal recessive
5	151443	LIFR		601559	Stuve-Wiedemann syndrome / Schwartz-Jampel type 2 syndrome	Autosomal recessive
13	601837	LIG4		606593	LIG4 syndrome	Autosomal recessive
15	610350	LINS1	LINS	614340	Mental retardation, autosomal recessive, type 27	Autosomal recessive
10	613497	LIPA		278000	Lysosomal acid lipase deficiency	Autosomal recessive
19	151750	LIPE		615980	Lipodystrophy, familial partial, type 6	Autosomal recessive
3	607365	LIPH		604379	Hypotrichosis, type 7 or woolly hair, autosomal recessive, type 2, with or without hypotrichosis	Autosomal recessive
18	601567	LMAN1		227300	Combined deficiency of factor V and factor VIII, type 1	Autosomal recessive
6	612625	LMBRD1		277380	Methylmalonic aciduria and homocystinuria, cblF type	Autosomal recessive
16	611761	LMF1		246650	Lipase deficiency, combined	Autosomal recessive
1	150330	LMNA		605588; 616516	LMNA-related disorders, autosomal recessive	Autosomal recessive
18	613072	LOXHD1		613079	Deafness, autosomal recessive, type 77	Autosomal recessive
13	609239	LPAR6		278150	Hypotrichosis, type 8 or woolly hair, autosomal recessive, type 1, with or without hypotrichosis	Autosomal recessive
2	605518	LPIN1		268200	Myoglobinuria, acute recurrent, autosomal recessive	Autosomal recessive
18	605519	LPIN2		609628	Majeed syndrome	Autosomal recessive
8	609708	LPL		238600	Lipoprotein lipase deficiency	Autosomal recessive
4	604863	LRAT		613341	Leber congenital amaurosis type 14	Autosomal recessive
10	614537	LRMDA	C10orf11	615179	Albinism, oculocutaneous, type 7	Autosomal recessive
2	600073	LRP2		222448	Donnai-Barrow syndrome	Autosomal recessive
11	604270	LRP4		212780	Cenani-Lenz syndactyly syndrome	Autosomal recessive
11	603506	LRP5		259770	Osteoporosis-pseudoglioma syndrome	Autosomal recessive

4	104225	LRPAP1	615431	Myopia, type 23, autosomal recessive	Autosomal recessive
2	607544	LRPPRC	220111	Leigh syndrome, French-Canadian type	Autosomal recessive
8	614930	LRRC6	614935	Ciliary dyskinesia, primary, type 19	Autosomal recessive
9	610933	LRSAM1	614436	Charcot-Marie-Tooth disease, axonal, type 2P	Autosomal recessive
11	612414	LRTOMT	611451	Deafness, autosomal recessive, type 63	Autosomal recessive
14	602091	LTBP2	251750	Microspherophakia and/or megalocornea, with ectopia lentis and with or without secondary glaucoma	Autosomal recessive
11	602090	LTBP3	601216	Dental anomalies and short stature	Autosomal recessive
19	604710	LTBP4	613177	Cutis laxa, autosomal recessive, type 1C	Autosomal recessive
1	606897	LYST	214500	Chediak-Higashi syndrome	Autosomal recessive
3	606568	LZTFL1	615994	Bardet-Biedl syndrome, type 17	Autosomal recessive
7	606382	MAGI2	617609	Nephrotic syndrome, type 15	Autosomal recessive
X	300715	MAGT1	300853	Immunodeficiency, X-linked, with magnesium defect, Epstein-Barr virus infection and neoplasia	X-linked
6	154235	MAK	614181	Retinitis pigmentosa type 62	Autosomal recessive
X	300120	MAMLD1	300758	Hypospadias 2, X-linked	X-linked
9	604346	MAN1B1	614202	Mental retardation, autosomal recessive, type 15	Autosomal recessive
19	609458	MAN2B1	248500	Alpha-mannosidosis	Autosomal recessive
4	609489	MANBA	248510	Mannosidosis, beta	Autosomal recessive
X	309850	MAOA	300615	Brunner syndrome	X-linked
17	157140	MAPT	260540	Supranuclear palsy, progressive atypical (parkinsonism syndrome)	Autosomal recessive
2	609728	MARS2	611390	Spastic ataxia, type 3, autosomal recessive	Autosomal recessive
5	610572	MARVELD2	610153	Deafness, autosomal recessive, type 49	Autosomal recessive
3	600521	MASP1	257920	3MC syndrome 1	Autosomal recessive
10	610550	MAT1A	250850	Methionine adenosyltransferase deficiency, autosomal recessive	Autosomal recessive
X	300294	MBTPS2	308205; 301014	IFAP/BRESHECK syndrome; Osteogenesis imperfecta, type 19	X-linked
18	607397	MC2R	202200	Glucocorticoid deficiency, due to ACTH unresponsiveness	Autosomal recessive
3	609010	MCCC1	210200	3-Methylcrotonyl-CoA carboxylase deficiency, type 1	Autosomal recessive
5	609014	MCCC2	210210	3-Methylcrotonyl-CoA carboxylase deficiency, type 2	Autosomal recessive
2	608419	MCEE	251120	Methylmalonyl-CoA epimerase deficiency	Autosomal recessive
2	607788	MCFD2	613625	Combined deficiency of factor V and factor VIII, type 2	Autosomal recessive
21	603294	MCM3AP	618124	Peripheral neuropathy, autosomal recessive, with or without impaired intellectual development	Autosomal recessive
19	605248	MCOLN1	252650	Mucolipidosis type 4	Autosomal recessive
8	607117	MCPH1	251200	Microcephaly type 1, primary, autosomal recessive	Autosomal recessive
X	300005	MECP2	300673;312750	Encephalopathy, neonatal severe; Rett syndrome	X-linked
X	300188	MED12	309520	Lujan-Fryns syndrome	X-linked
6	605042	MED23	614249	Mental retardation, autosomal recessive, type 18	Autosomal recessive
19	610197	MED25	616449	Basel-Vanagait-Smirin-Yosef syndrome	Autosomal recessive
16	608107	MEFV	249100	Familial Mediterranean fever	Autosomal recessive
5	612453	MEGF10	614399	Myopathy, areflexia, respiratory distress, and dysphagia, early-onset	Autosomal recessive
2	604705	MERTK	613862	Retinitis pigmentosa type 38	Autosomal recessive
15	605195	MESP2	608681	Spondylocostal dysostosis, type 2, autosomal recessive	Autosomal recessive
2	614785	MFF	617086	Encephalopathy due to defective mitochondrial and peroxisomal fission, type 2	Autosomal recessive
1	608507	MFN2	617087	Charcot-Marie-Tooth disease, axonal, type 2A2B	Autosomal recessive
11	606227	MFRP	611040	Microphthalmia, isolated type 5	Autosomal recessive

1	614397	MFSD2A		616486	Microcephaly 15, primary, autosomal recessive	Autosomal recessive
4	611124	MFSD8		610951	Ceroid lipofuscinosis, neuronal, type 7	Autosomal recessive
14	602616	MGAT2		212066	Congenital disorder of glycosylation, type 2a	Autosomal recessive
12	154870	MGP		245150	Keutel syndrome	Autosomal recessive
X	300552	MID1		300000	Opitz GBBB syndrome, type 1	X-linked
3	156845	MITF		617306	COMMAD syndrome	Autosomal recessive
20	604896	MKKS		605231	Bardet-Biedl syndrome type 6	Autosomal recessive
17	609883	MKS1		615990; 249000; 617121	Bardet-Biedl syndrome type 13; Meckel syndrome, type 1; Joubert syndrome, type 28	Autosomal recessive
22	605908	MLC1		604004	Megalencephalic leukoencephalopathy with subcortical cysts	Autosomal recessive
2	606526	MLPH		609227	Griscelli syndrome, type 3	Autosomal recessive
16	606761	MLYCD		248360	Malonyl-CoA decarboxylase deficiency	Autosomal recessive
4	607481	MMAA		251100	Methylmalonic aciduria, vitamin B12-responsive	Autosomal recessive
12	607568	MMAB		251110	Methylmalonic aciduria, vitamin B12-responsive, type cblB	Autosomal recessive
1	609831	MMACHC		277400	Methylmalonic aciduria and homocystinuria, cblC type	Autosomal recessive, digenic inheritance (PRDX1 gene)
2	611935	MMADHC		277410	Homocystinuria, cblD type, variant 1	Autosomal recessive
3	120520	MME		617017*	Charcot-Marie-Tooth disease, axonal, type 2T	Autosomal recessive*
11	600108	MMP13		250400	Metaphyseal dysplasia, Spahr type	Autosomal recessive
16	120360	MMP2		259600	Multicentric osteolysis, nodulosis, and arthropathy (MONA)	Autosomal recessive
11	604629	MMP20		612529	Amelogenesis imperfecta, type 2A2 (hypomaturation type)	Autosomal recessive
6	609058	MMUT	MUT	251000	Methylmalonic aciduria, mut(0) type	Autosomal recessive
18	613274	MOCOS		603592	Xanthinuria, type 2	Autosomal recessive
6	603707	MOCS1		252150	Molybdenum cofactor deficiency A	Autosomal recessive
5	603708	MOCS2		252160	Molybdenum cofactor deficiency B	Autosomal recessive
2	601336	MOGS		606056	Congenital disorder of glycosylation, type 2B	Autosomal recessive
17	604041	MPDU1		609180	Congenital disorder of glycosylation, type 1F	Autosomal recessive
9	603785	MPDZ		615219	Hydrocephalus, congenital, type 2, with or without brain or eye anomalies	Autosomal recessive
15	154550	MPI		602579	Congenital disorder of glycosylation, type 1B	Autosomal recessive
1	159530	MPL		604498	Thrombocytopenia, congenital amegakaryocytic	Autosomal recessive
7	609188	MPLKIP		234050	Trichothiodystrophy, type 4, nonphotosensitive	Autosomal recessive
17	606989	MPO		254600	Myeloperoxidase deficiency	Autosomal recessive
2	137960	MPV17		256810; 618400	Mitochondrial DNA depletion syndrome type 6 (hepatocerebral); Charcot-Marie-Tooth disease, axonal, type 2EE	Autosomal recessive
21	609196	MRAP		607398	Glucocorticoid deficiency, type 2	Autosomal recessive
11	600814	MRE11	MRE11A	604391	Ataxia-telangiectasia-like disorder 1	Autosomal recessive
10	609204	MRPS16		610498	Combined oxidative phosphorylation deficiency 2	Autosomal recessive
3	605810	MRPS22		611719	Combined oxidative phosphorylation deficiency type 5	Autosomal recessive
5	600887	MSH3		617100	Familial adenomatous polyposis, type 4	Autosomal recessive
4	607545	MSMO1		616834	Microcephaly, congenital cataract, and psoriasiform dermatitis	Autosomal recessive
12	613719	MSRB3		613718	Deafness, autosomal recessive, type 74	Autosomal recessive
15	611766	MTFMT		614947	Combined oxidative phosphorylation deficiency 15	Autosomal recessive
14	172460	MTHFD1		617780	Combined immunodeficiency and megaloblastic anemia with or without hyperhomocysteinemia	Autosomal recessive
X	300415	MTM1		310400	Myotubular myopathy, X-linked	X-linked
11	603557	MTMR2		601382	Charcot-Marie-Tooth disease, type 4B1	Autosomal recessive

6	614667	MTO1	614702	Combined oxidative phosphorylation deficiency 10	Autosomal recessive
1	156570	MTR	250940	Homocystinuria-megaloblastic anemia, cblG complementation type	Autosomal recessive
5	602568	MTRR	236270	Homocystinuria-megaloblastic anemia, cbl E type	Autosomal recessive
4	157147	MTTP	200100	Abetalipoproteinemia	Autosomal recessive
9	601296	MUSK	208150; 616325	Fetal akinesia deformation sequence, type 1; Myasthenic syndrome, congenital, type 9, associated with acetylcholine receptor deficiency	Autosomal recessive
12	251170	MVK	610377	Mevalonic aciduria	Autosomal recessive
12	160794	MYBPC1	614915	Lethal congenital contracture syndrome, type 4	Autosomal recessive
3	602170	MYD88	612260	Immunodeficiency, type 68	Autosomal recessive
17	160740	MYH2	605637	Proximal myopathy and ophthalmoplegia	Autosomal recessive
17	602666	MYO15A	600316	Deafness, autosomal recessive, type 3	Autosomal recessive
22	607295	MYO18B	616549	Klippel-Feil syndrome, type 4, autosomal recessive, with myopathy and facial dysmorphism	Autosomal recessive
10	606808	MYO3A	607101	Deafness, autosomal recessive, type 30	Autosomal recessive
15	160777	MYO5A	214450	Griscelli syndrome, type 1	Autosomal recessive
18	606540	MYO5B	251850	Microvillus inclusion disease	Autosomal recessive
11	276903	MYO7A	276900; 600060	Usher syndrome, type 1B; Deafness, autosomal recessive, type 2	Autosomal recessive
10	608517	MYPN	617336	Nemaline myopathy, type 11, autosomal recessive	Autosomal recessive
X	300013	NAA10	300855	Ogden syndrome	X-linked
22	104170	NAGA	609241	Schindler disease, type I	Autosomal recessive
17	609701	NAGLU	252920	Mucopolysaccharidosis, type 3B (Sanfilippo B)	Autosomal recessive
17	608300	NAGS	237310	N-acetylglutamate synthase deficiency	Autosomal recessive
11	612803	NARS2	616239	Combined oxidative phosphorylation deficiency 24	Autosomal recessive
2	608025	NBAS	616483; 614800	Infantile liver failure syndrome, type 2; Short stature, optic nerve atrophy, and Pelger-Huet anomaly	Autosomal recessive
3	614169	NBEAL2	139090	Gray platelet syndrome	Autosomal recessive
8	602667	NBN	251260	Nijmegen breakage syndrome	Autosomal recessive
7	608512	NCF1	233700	Chronic granulomatous disease, type 1	Autosomal recessive
1	608515	NCF2	233710	Chronic granulomatous disease, type 2	Autosomal recessive
16	609449	NDE1	614019	Lissencephaly, type 4 (with microcephaly)	Autosomal recessive
X	300658	NDP	310600	Norrie disease	X-linked
8	605262	NDRG1	601455	Charcot-Marie-Tooth disease, type 4D	Autosomal recessive
5	600853	NDST1	616116	Mental retardation, autosomal recessive, type 46	Autosomal recessive
2	603835	NDUFA10	618243	Mitochondrial complex I deficiency, nuclear type 22	Autosomal recessive
19	612638	NDUFA11	618236	Mitochondrial complex I deficiency, nuclear type 14	Autosomal recessive
12	614530	NDUFA12	618244	?Mitochondrial complex I deficiency, nuclear type 23	Autosomal recessive
15	606934	NDUFAF1	618234	Mitochondrial complex I deficiency, nuclear type 11	Autosomal recessive
5	609653	NDUFAF2	618233	Mitochondrial complex I deficiency, nuclear type 10	Autosomal recessive
3	612911	NDUFAF3	618240	Mitochondrial complex I deficiency, nuclear type 18	Autosomal recessive
20	612360	NDUFAF5	618238	Mitochondrial complex I deficiency, nuclear type 16	Autosomal recessive
8	612392	NDUFAF6	618239	Mitochondrial complex I deficiency, nuclear type 17	Autosomal recessive
2	603839	NDUFB3	618246	Mitochondrial complex I deficiency, nuclear type 25	Autosomal recessive
2	157655	NDUFS1	618226	Mitochondrial complex I deficiency, nuclear type 5	Autosomal recessive
1	602985	NDUFS2	618228	Mitochondrial complex I deficiency, nuclear type 6	Autosomal recessive
11	603846	NDUFS3	618230	Mitochondrial complex I deficiency, nuclear type 8	Autosomal recessive
5	602694	NDUFS4	252010	Mitochondrial complex I deficiency, nuclear type 1	Autosomal recessive

5	603848	NDUFS6		618232	Mitochondrial complex I deficiency, nuclear type 9	Autosomal recessive
19	601825	NDUFS7		618224	Mitochondrial complex I deficiency, nuclear type 3	Autosomal recessive
11	602141	NDUFS8		618222	Mitochondrial complex I deficiency, nuclear type 2	Autosomal recessive
11	161015	NDUFV1		618225	Mitochondrial complex I deficiency, nuclear type 4	Autosomal recessive
2	161650	NEB		256030	Nemaline myopathy type 2	Autosomal recessive
11	600644	NECTIN1	PVRL1	225060	Cleft lip/palate-ectodermal dysplasia syndrome; Orofacial cleft 7	Autosomal recessive
1	609607	NECTIN4	PVRL4	613573	Ectodermal dysplasia-syndactyly syndrome, type 1	Autosomal recessive
8	162280	NEFL		607734	Charcot-Marie-Tooth disease, type 1F	Autosomal recessive
4	604588	NEK1		263520	Short-rib thoracic dysplasia, type 6, with or without polydactyly	Autosomal recessive
17	609799	NEK8		615415	Renal-hepatic-pancreatic dysplasia, type 2	Autosomal recessive
6	608272	NEU1		256550	Sialidosis, type 1 and type 2	Autosomal recessive
10	604882	NEUROG3		610370	Diarrhea 4, malabsorptive, congenital	Autosomal recessive
2	608100	NFU1		605711	Multiple mitochondrial dysfunctions syndrome 1	Autosomal recessive
1	162030	NGF		608654	Neuropathy, hereditary sensory and autonomic, type 5	Autosomal recessive
2	611290	NHEJ1		611291	Severe combined immunodeficiency with microcephaly, growth retardation, and sensitivity to ionizing radiation	Autosomal recessive
6	608072	NHLRC1		254780	Epilepsy, progressive myoclonic, type 2B (Lafora)	Autosomal recessive
5	606470	NHP2		613987	Dyskeratosis congenita, autosomal recessive type 2	Autosomal recessive
X	300457	NHS		302200	Cataract 40, X-linked	X-linked
5	609383	NIPAL4		612281	Ichthyosis, congenital, autosomal recessive, type 6	Autosomal recessive
8	611770	NKX2-6		217095	Conotruncal heart malformations	Autosomal recessive
X	300427	NLGN4X		300495	Mental retardation, X-linked	X-linked, Multifactorial, Isolated cases
17	606636	NLRP1		617388*	Autoinflammation with arthritis and dyskeratosis	Autosomal recessive*
19	609661	NLRP7		231090	Hydatidiform mole, recurrent, type 1	Autosomal recessive
7	607421	NME8		610852	Ciliary dyskinesia, primary, type 6	Autosomal recessive
1	608700	NMNAT1		608553	Leber congenital amaurosis type 9	Autosomal recessive
15	606471	NOP10		224230	Dyskeratosis congenita, autosomal recessive type 1	Autosomal recessive
18	607623	NPC1		257220	Niemann-Pick disease, type C1	Autosomal recessive
14	601015	NPC2		607625	Niemann-pick disease, type C2	Autosomal recessive
2	607100	NPHP1		609583	Joubert syndrome type 4	Autosomal recessive
3	608002	NPHP3		267010	Meckel syndrome type 7	Autosomal recessive
1	607215	NPHP4		606966	Nephronophthisis type 4	Autosomal recessive
19	602716	NPHS1		256300	Nephrotic syndrome, type 1	Autosomal recessive
1	604766	NPHS2		600995	Nephrotic syndrome, type 2	Autosomal recessive
9	108961	NPR2		602875	Acromesomelic dysplasia, Maroteaux type	Autosomal recessive
X	300473	NR0B1		300200	Adrenal hypoplasia, congenital	X-linked
12	603826	NR1H4		617049	Cholestasis, progressive familial intrahepatic, type 5	Autosomal recessive
15	604485	NR2E3		268100; 611131*	Enhanced S-cone syndrome (Goldmann-Favre); Retinitis pigmentosa, type 37	Autosomal recessive; Autosomal recessive*
14	162080	NRL		613750*	Retinal degeneration, autosomal recessive, clumped pigment type	Autosomal recessive*
2	600565	NRXN1		614325	Pitt-Hopkins-like syndrome, type 2	Autosomal recessive
X	300275	NSDHL		308050; 300831	CHILD syndrome	X-linked
5	610916	NSUN2		611091	Mental retardation, autosomal recessive, type 5	Autosomal recessive

7	606224	NT5C3A		266120	Anemia, hemolytic, due to UMPH1 deficiency	Autosomal recessive
6	129190	NT5E		211800	Calcification of joints and arteries	Autosomal recessive
16	602656	NTHL1		616415	Familial adenomatous polyposis, type 3	Autosomal recessive
1	191315	NTRK1		256800	Insensitivity to pain, congenital, with anhidrosis	Autosomal recessive
14	613621	NUBPL		618242	Mitochondrial complex I deficiency, nuclear type 21	Autosomal recessive
19	605815	NUP62		271930	Striatonigral degeneration, infantile	Autosomal recessive
X	300278	NYX		310500	Night blindness, congenital stationary (complete), type 1A, X-linked	X-linked
10	613349	OAT		258870	Gyrate atrophy of choroid and retina	Autosomal recessive
2	610991	OBSL1		612921	3M syndrome 2	Autosomal recessive
15	611409	OCA2		203200	Oculocutaneous albinism type 2	Autosomal recessive
5	602876	OCLN		251290	Pseudo-TORCH syndrome, type 1	Autosomal recessive
X	300535	OCRL		309000; 300555	Lowe Syndrome; Dent disease type 2	X-linked
X	300170	OFD1		311200; 300209; 300804	Orofaciodigital syndrome, type 1; Simpson-Golabi-Behmel syndrome, type 2; Joubert syndrome, type 10	X-linked
3	605290	OPA1		210000	Behr syndrome	Autosomal recessive
19	606580	OPA3		258501	3-methylglutaconic aciduria, type 3	Autosomal recessive
X	300127	OPHN1		300486	Mental retardation, X-linked, with cerebellar hypoplasia and distinctive facial appearance	X-linked
10	602432	OPTN		613435	Amyotrophic lateral sclerosis, type 12	Autosomal recessive
1	601902	ORC1		224690	Meier-Gorlin syndrome, type 1	Autosomal recessive
2	603056	ORC4		613800	Meier-Gorlin syndrome, type 2	Autosomal recessive
16	607213	ORC6		613803	Meier-Gorlin syndrome, type 3	Autosomal recessive
6	607649	OSTM1		259720	Osteopetrosis, autosomal recessive type 5	Autosomal recessive
X	300461	OTC		311250	Ornithine transcarbamylase deficiency	X-linked
16	607038	OTOA		607039	Deafness, autosomal recessive, type 22	Autosomal recessive
2	603681	OTOF		601071	Deafness, autosomal recessive, type 9	Autosomal recessive
5	601424	OXCT1		245050	Succinyl CoA:3-oxoacid CoA transferase deficiency	Autosomal recessive
3	600515	P2RY12		609821	Bleeding disorder, platelet-type, type 8	Autosomal recessive
1	610339	P3H1	LEPRE1	610915	Osteogenesis imperfecta, type 8	Autosomal recessive
3	610341	P3H2	LEPREL1	614292	Myopia, high, with cataract and vitreoretinal degeneration	Autosomal recessive
12	612349	PAH		261600	Phenylketonuria	Autosomal recessive
X	300142	PAK3		300558	Mental retardation, X-linked, type 30	X-linked
20	606157	PANK2		234200	Neurodegeneration with brain iron accumulation type 1	Autosomal recessive
10	603005	PAPSS2		612847	Brachyolmia, type 4, with mild epiphyseal and metaphyseal changes	Autosomal recessive
1	602533	PARK7		606324	Parkinson disease, type 7, autosomal recessive, early-onset	Autosomal recessive
11	608786	PC		266150	Pyruvate carboxylase deficiency	Autosomal recessive
10	126090	PCBD1		264070	Hyperphenylalaninemia, BH4-deficient, type D	Autosomal recessive
13	232000	PCCA		606054	Propionic acidemia	Autosomal recessive
3	232050	PCCB		606054	Propionic acidemia	Autosomal recessive
10	605514	PCDH15		609533; 601067	Deafness, autosomal recessive, type 23; Usher syndrome, type 1D/F digenic	Autosomal recessive
X	300460	PCDH19		300088	Epileptic encephalopathy, early infantile, type 9	X-linked
21	605925	PCNT		210720	Microcephalic osteodysplastic primordial dwarfism, type 2	Autosomal recessive
5	162150	PCSK1		600955	Obesity with impaired prohormone processing	Autosomal recessive
6	610652	PDE10A		616921	Dyskinesia, limb and orofacial, infantile-onset	Autosomal recessive
5	180071	PDE6A		613810	Retinitis pigmentosa type 43	Autosomal recessive
4	180072	PDE6B		613801	Retinitis pigmentosa type 40	Autosomal recessive

10	600827	PDE6C	613093	Cone dystrophy type 4	Autosomal recessive
17	180073	PDE6G	613582	Retinitis pigmentosa type 57	Autosomal recessive
12	601190	PDE6H	610024*	Retinal cone dystrophy 3 and achromatopsia 6	Autosomal recessive*
X	300502	PDHA1	312170	Pyruvate dehydrogenase E1-alpha deficiency	X-linked
3	179060	PDHB	614111	Pyruvate dehydrogenase E1-beta deficiency	Autosomal recessive
11	608769	PDHX	245349	Lacticacidemia due to PDX1 deficiency	Autosomal recessive
8	605993	PDP1	608782	Pyruvate dehydrogenase phosphatase deficiency	Autosomal recessive
10	607429	PDSS1	614651	Coenzyme Q10 deficiency, primary, type 2	Autosomal recessive
6	610564	PDSS2	614652	Coenzyme Q10 deficiency, primary, type 3	Autosomal recessive
13	600733	PDX1	260370	Pancreatic agenesis type 1	Autosomal recessive
10	612971	PDZD7	618003; 605472	Deafness, autosomal recessive, type 57; Usher syndrome, type 2C, digenic	Autosomal recessive; Digenic inheritance (ADGRV1 gene)
19	613230	PEPD	170100	Prolidase deficiency	Autosomal recessive
7	602136	PEX1	234580	Heimler syndrome type 1	Autosomal recessive
1	602859	PEX10	614870; 614871	Peroxisome biogenesis disorder, type 6A (Zellweger syndrome); Peroxisome biogenesis disorder, type 6B	Autosomal recessive
17	601758	PEX12	614859	Peroxisome biogenesis disorder type 3A (Zellweger)	Autosomal recessive
2	601789	PEX13	614883; 614885	Peroxisome biogenesis disorder, type 11A (Zellweger syndrome); Peroxisome biogenesis disorder, type 11B	Autosomal recessive
1	601791	PEX14	614887	Peroxisome biogenesis disorder, type 13A (Zellweger syndrome)	Autosomal recessive
11	603360	PEX16	614876; 614877	Peroxisome biogenesis disorder, type 8A (Zellweger syndrome); Peroxisome biogenesis disorder, type 8B	Autosomal recessive
1	600279	PEX19	614886	Peroxisome biogenesis disorder, type 12A (Zellweger syndrome)	Autosomal recessive
8	170993	PEX2	614866	Peroxisome biogenesis disorder type 5A (Zellweger)	Autosomal recessive
22	608666	PEX26	614872	Peroxisome biogenesis disorder type 7A (Zellweger)	Autosomal recessive
6	603164	PEX3	614882	Peroxisome biogenesis disorder, type 10A (Zellweger syndrome)	Autosomal recessive
12	600414	PEX5	214110	Peroxisome biogenesis disorder type 2A (Zellweger)	Autosomal recessive
6	601498	PEX6	614862; 616617*; 614863	Peroxisome biogenesis disorder, type 4A (Zellweger syndrome); Peroxisome biogenesis disorder, type 4B; Heimler syndrome 2	Autosomal recessive; Autosomal recessive*; Autosomal recessive
6	601757	PEX7	215100	Rhizomelic chondrodysplasia punctata, type 1	Autosomal recessive
12	610681	PFKM	232800	Glycogen storage disease, type 7	Autosomal recessive
X	311800	PGK1	300653	Phosphoglycerate kinase 1 deficiency	X-linked
1	171900	PGM1	614921	Congenital disorder of glycosylation, type 1t	Autosomal recessive
X	300550	PHEX	307800	Hypophosphatemic rickets, X-linked dominant	X-linked
X	300414	PHF6	301900	Borjeson-Forssman-Lehmann syndrome	X-linked
X	300560	PHF8	300263	Mental retardation syndrome, X-linked, Siderius type	X-linked
1	606879	PHGDH	256520; 601815	Neu-Laxova syndrome, type 1; Phosphoglycerate dehydrogenase deficiency	Autosomal recessive
X	311870	PHKA1	300559	Glycogen storage disease, type 9D	X-linked
X	300798	PHKA2	306000	Glycogen storage disease, type 9A1 and type 9A2	X-linked
16	172490	PHKB	261750	Glycogen storage disease, type 9B	Autosomal recessive
16	172471	PHKG2	613027	Glycogen storage disease type 9c	Autosomal recessive
10	602026	PHYH	266500	Refsum disease	Autosomal recessive
16	611184	PIEZO1	616843	Lymphedema, hereditary, type 3	Autosomal recessive
18	613629	PIEZO2	617146	Arthrogryposis, distal, with impaired proprioception and touch	Autosomal recessive
X	311770	PIGA	300868	Multiple congenital anomalies-hypotonia-seizures syndrome, type 2	X-linked

17	605947	PIGL		280000	Zunich neuroectodermal syndrome	Autosomal recessive
18	606097	PIGN		614080	Multiple congenital anomalies-hypotonia-seizures syndrome, type 1	Autosomal recessive
9	614730	PIGO		614749	Hyperphosphatasia with mental retardation syndrome 2	Autosomal recessive
1	608309	PINK1		605909	Parkinson disease, type 6, early onset	Autosomal recessive
19	606102	PIP5K1C		611369	Lethal congenital contractural syndrome, type 3	Autosomal recessive
2	610219	PJVK	DFNB59	610220	Deafness, autosomal recessive, type 59	Autosomal recessive
6	606702	PKHD1		263200	Polycystic kidney disease type 4	Autosomal recessive
1	609712	PKLR		266200	Pyruvate kinase deficiency	Autosomal recessive
1	601975	PKP1		604536	Ectodermal dysplasia/skin fragility syndrome	Autosomal recessive
22	603604	PLA2G6		256600	Infantile neuroaxonal dystrophy type 1	Autosomal recessive
20	607120	PLCB1		613722	Epileptic encephalopathy, early infantile, type 12	Autosomal recessive
20	600810	PLCB4		614669*	Auriculocondylar syndrome, type 2	Autosomal recessive*
3	602142	PLCD1		151600	Nail disorder, nonsyndromic congenital, type 3 (leukonychia)	Autosomal recessive
10	608414	PLCE1		610725	Nephrotic syndrome, type 3	Autosomal recessive
8	601282	PLEC		226670	Epidermolysis bullosa simplex with muscular dystrophy	Autosomal recessive
1	611101	PLEKHG5		615376	Charcot-Marie-Tooth disease, recessive intermediate, type C	Autosomal recessive
6	173350	PLG		217090	Plasminogen deficiency, type I	Autosomal recessive
1	153454	PLOD1		225400	Ehlers-Danlos syndrome, kyphoscoliotic type, 1	Autosomal recessive
3	601865	PLOD2		609220	Bruck syndrome 2	Autosomal recessive
7	603066	PLOD3		612394	Lysyl hydroxylase 3 deficiency	Autosomal recessive
X	300401	PLP1		312080	Pelizaeus-Merzbacher disease	X-linked
16	601785	PMM2		212065	Congenital disorder of glycosylation, type 1A	Autosomal recessive
17	601097	PMP22		145900*	Dejerine-Sottas disease	Autosomal recessive*
19	605610	PNKP		616267; 613402	Ataxia-oculomotor apraxia, type 4; Microcephaly, seizures, and developmental delay	Autosomal recessive
14	164050	PNP		613179	Immunodeficiency due to purine nucleoside phosphorylase deficiency	Autosomal recessive
6	612121	PNPLA1		615024	Ichthyosis, congenital, autosomal recessive, type 10	Autosomal recessive
11	609059	PNPLA2		610717	Neutral lipid storage disease with myopathy	Autosomal recessive
19	603197	PNPLA6		215470; 275400; 612020	Boucher-Neuhauser syndrome; Oliver-McFarlane syndrome; Spastic paraplegia, type 39, autosomal recessive	Autosomal recessive
17	603287	PNPO		610090	Pyridoxamine 5'-phosphate oxidase deficiency	Autosomal recessive
15	174763	POLG		203700; 613662; 607459	POLG-related disorders	Autosomal recessive
6	603968	POLH		278750	Xeroderma pigmentosum, variant type	Autosomal recessive
6	610060	POLR1C		616494; 248390	Leukodystrophy, hypomyelinating, type 11; Treacher Collins syndrome 3	Autosomal recessive
13	613715	POLR1D		613717*	Treacher Collins syndrome, type 2	Autosomal recessive*
10	614258	POLR3A		607694	Leukodystrophy, hypomyelinating, type 7	Autosomal recessive
12	614366	POLR3B		614381	Leukodystrophy, hypomyelinating, type 8	Autosomal recessive
2	176830	POMC		609734	Obesity, adrenal insufficiency, and red hair due to POMC deficiency	Autosomal recessive
1	606822	POMGNT1		253280; 613151; 613157	Muscular dystrophy-dystroglycanopathy, type 3A (Walker-Warburg syndrome); Type 3B; Type 3C (limb-girdle muscular dystrophy, type 15 [LGMDR15])	Autosomal recessive
3	614828	POMGNT2		614830; 618135	Muscular dystrophy-dystroglycanopathy, type 8A (Walker-Warburg syndrome); Type 8C (limb-girdle muscular dystrophy, type 24 [LGMD R24])	Autosomal recessive
13	613386	POMP		601952	Keratosis linearis with ichthyosis congenita and sclerosing keratoderma	Autosomal recessive
9	607423	POMT1		236670; 613155; 609308	Muscular dystrophy-dystroglycanopathy, type 1A (Walker-Warburg syndrome); Type 1B; Type 1C (limb-girdle muscular dystrophy, type 11 [LGMD R11])	Autosomal recessive
14	607439	POMT2		613150; 613156; 613158	Muscular dystrophy-dystroglycanopathy, type 2A (Walker-Warburg syndrome); Type 2B; Type 2C (limb-girdle muscular dystrophy, type 14 [LGMD R14])	Autosomal recessive

8	602486	POP1		617396	Anauxetic dysplasia, type 2	Autosomal recessive
7	124015	POR		201750	Antley-Bixler syndrome with genital anomalies and disordered steroidogenesis	Autosomal recessive
X	300651	PORCN		305600	Focal dermal hypoplasia	X-linked
3	173110	POU1F1		613038*	Pituitary hormone deficiency, combined, type 1	Autosomal recessive*
X	300039	POU3F4		304400	Deafness, X-linked, type 2	X-linked
15	123841	PPIB		259440	Osteogenesis imperfecta, type 9	Autosomal recessive
1	600722	PPT1		256730	Ceroid lipofuscinosis, neuronal, type 1	Autosomal recessive
X	300463	PQBP1		309500	Renpenning syndrome	X-linked
17	610598	PRCD		610599	Retinitis pigmentosa, type 36	Autosomal recessive
4	614161	PRDM5		614170	Brittle cornea syndrome, type 2	Autosomal recessive
2	609557	PREPL		616224	Myasthenic syndrome, congenital, type 22	Autosomal recessive
10	170280	PRF1		603553	Hemophagocytic lymphohistiocytosis, familial, type 2	Autosomal recessive
1	604283	PRG4		208250	Camptodactyly-arthropathy-coxa vara-pericarditis syndrome	Autosomal recessive
12	608500	PRICKLE1		612437	Epilepsy, progressive myoclonic, type 1B	Autosomal recessive
6	602544	PRKN	PARK2	600116	Parkinson disease, type 2, juvenile	Autosomal recessive
2	603424	PRKRA		612067	Dystonia, type 16	Autosomal recessive
16	610087	PRMT7		617157	Short stature, brachydactyly, intellectual developmental disability, and seizures	Autosomal recessive
2	612283	PROC		612304	Thrombophilia due to protein C deficiency, autosomal recessive	Autosomal recessive
22	606810	PRODH		239500	Hyperprolinemia, type 1	Autosomal recessive
4	604365	PROM1		612095	Retinitis pigmentosa, type 41	Autosomal recessive
5	601538	PROP1		262600	Pituitary hormone deficiency, combined, type 2	Autosomal recessive
3	176880	PROS1		614514	Thrombophilia due to protein S deficiency, autosomal recessive	Autosomal recessive
6	179605	PRPH2		608133*; 136880*	Leber congenital amaurosis 18; Retinitis punctata albescens	Autosomal recessive*
X	311850	PRPS1		300661; 304500; 311070; 301835	PRPS1-related disoders	X-linked
1	167420	PRRX1		202650*	Agnathia-otocephaly complex	Autosomal recessive*
7	276000	PRSS1		614044	Trypsinogen deficiency	Autosomal recessive
4	606709	PRSS12		249500	Mental retardation, autosomal recessive, type 1	Autosomal recessive
2	613858	PRSS56		613517	Microphthalmia, isolated, type 6	Autosomal recessive
19	605725	PRX		614895	Charcot-Marie-Tooth disease, type 4F	Autosomal recessive
10	176801	PSAP		611721	Combined SAP deficiency	Autosomal recessive
9	610936	PSAT1		616038	Neu-Laxova syndrome, type 2	Autosomal recessive
6	177046	PSMB8		256040	Autoinflammation, lipodystrophy, and dermatosis syndrome	Autosomal recessive
7	172480	PSPH		614023	Phosphoserine phosphatase deficiency	Autosomal recessive
11	168450	PTH		146200*	Hypoparathyroidism, familial isolated, type 1	Autosomal recessive*
3	168468	PTH1R		215045; 600002	Chondrodysplasia, Blomstrand type; Eiken syndrome	Autosomal recessive
1	151460	PTPRC		608971	Severe combined immunodeficiency, T cell-negative, B-cell/natural killer-cell positive	Autosomal recessive
12	603317	PTPRQ		613391	Deafness, autosomal recessive, type 84A	Autosomal recessive
11	612719	PTS		261640	Hyperphenylalaninemia, BH4-deficient, type A	Autosomal recessive
12	608109	PUS1		600462	Myopathy, lactic acidosis, and sideroblastic anemia, type 1	Autosomal recessive
2	605158	PXDN		269400	Anterior segment dysgenesis, type 7, with sclerocornea	Autosomal recessive
17	179035	PYCR1		612940	Cutis laxa, autosomal recessive, type 2B	Autosomal recessive
14	613741	PYGL		232700	Glycogen storage disease, type 6	Autosomal recessive
11	608455	PYGM		232600	McArdle disease	Autosomal recessive

4	612676	QDPR		261630	Hyperphenylalaninemia, BH4-deficient, type C	Autosomal recessive
10	602207	RAB18		614222	Warburg micro syndrome, type 3	Autosomal recessive
6	606144	RAB23		201000	Carpenter syndrome	Autosomal recessive
15	603868	RAB27A		607624	Griscelli syndrome, type 2	Autosomal recessive
4	612994	RAB28		615374	Cone-rod dystrophy 18	Autosomal recessive
X	300774	RAB39B		300271; 311510	Mental retardation, X-linked, type 72; Waisman syndrome	X-linked
2	602536	RAB3GAP1		600118	Warburg micro syndrome, type 1	Autosomal recessive
1	609275	RAB3GAP2		212720	Martsolf syndrome	Autosomal recessive
11	179615	RAG1		603554; 601457	Omenn syndrome; Severe combined immunodeficiency, B cell-negative	Autosomal recessive
11	179616	RAG2		603554; 601457	Omenn syndrome; Severe combined immunodeficiency, B cell-negative	Autosomal recessive
11	601592	RAPSN		208150; 616326	Fetal akinesia deformation sequence, type 2; Myasthenic syndrome, congenital, type 11, associated with AChR deficiency	Autosomal recessive
6	611524	RARS2		611523	Pontocerebellar hypoplasia, type 6	Autosomal recessive
18	601881	RAX		611038	Isolated microphthalmia, type 3	Autosomal recessive
18	604124	RBBP8		251255; 606744	Jawad syndrome; Seckel syndrome, type 2	Autosomal recessive
X	300080	RBM10		311900	TARP syndrome	X-linked
1	605313	RBM8A		274000	Thrombocytopenia-absent radius syndrome	Autosomal recessive
10	180250	RBP4		615147	Retinal dystrophy, iris coloboma, and comedogenic acne syndrome	Autosomal recessive
1	180040	RD3		610612	Leber congenital amaurosis, type 12	Autosomal recessive
14	608830	RDH12		612712	Leber congenital amaurosis, type 13	Autosomal recessive
12	601617	RDH5		136880*	Fundus albipunctatus	Autosomal recessive*
11	179410	RDX		611022	Deafness, autosomal recessive, type 24	Autosomal recessive
8	603780	RECQL4		218600; 266280; 268400	Baller-Gerold syndrome; RAPADILINO syndrome; Rothmund-Thomson syndrome	Autosomal recessive
7	600514	RELN		257320	Lissencephaly 2 (Norman-Roberts type)	Autosomal recessive
1	179820	REN		267430	Renal tubular dysgenesis	Autosomal recessive
5	613114	RETREG1	FAM134B	613115	Neuropathy, hereditary sensory and autonomic, type 2B	Autosomal recessive
6	612659	RFX6		615710	Mitchell-Riley syndrome	Autosomal recessive
19	603200	RFXANK		209920	Bare lymphocyte syndrome, type 2, complementation group B	Autosomal recessive
10	600342	RGR		613769	Retinitis pigmentosa, type 44	Autosomal recessive
3	180380	RHO		613731*; 136880*	Retinitis pigmentosa, type 4; Retinitis punctata albescens	Autosomal recessive*
20	610222	RIN2		613075	Macs syndrome	Autosomal recessive
21	605706	RIPK4		263650	Popliteal pterygium syndrome, Bartsocas-Papas type	Autosomal recessive
15	180090	RLBP1		607475; 136880*	Bothnia retinal dystrophy; Fundus albipunctatus	Autosomal recessive; Autosomal recessive*
9	157660	RMRP	CHH	607095	Anauxetic dysplasia, type 1	Autosomal recessive
19	606034	RNASEH2A		610333	Aicardi-Goutieres syndrome, type 4	Autosomal recessive
13	610326	RNASEH2B		610181	Aicardi-Goutieres syndrome, type 2	Autosomal recessive
11	610330	RNASEH2C		610329	Aicardi-Goutieres syndrome, type 3	Autosomal recessive
3	612688	RNF168		611943	RIDDLE syndrome	Autosomal recessive
11	608630	ROBO3		607313	Gaze palsy, familial horizontal, with progressive scoliosis, type 1	Autosomal recessive
16	614574	ROGDI		226750	Kohlschutter-Tonz syndrome	Autosomal recessive
11	180721	ROM1		608133	Retinitis pigmentosa, type 7, digenic	Autosomal recessive
9	602337	ROR2		268310	Robinow syndrome, autosomal recessive	Autosomal recessive
8	603937	RP1		180100	Retinitis pigmentosa, type 1	Autosomal recessive
X	300757	RP2		312600	Retinitis pigmentosa, type 2, X-linked	X-linked

1	180069	RPE65		204100; 613794	RPE65-related Leber congenital amaurosis/early-onset severe retinal dystrophy	Autosomal recessive
X	312610	RPGR		300029; 304020	Retinitis pigmentosa, type 3, X-linked; Cone-rod dystrophy, X-linked, 1	X-linked
14	605446	RPGRIP1		613826	Leber congenital amaurosis, type 6	Autosomal recessive
16	610937	RPGRIP1L		611560; 611561; 619113	Joubert syndrome, type 7; Meckel syndrome, type 5; COACH syndrome	Autosomal recessive
X	312173	RPL10		300998	Mental retardation, X-linked, syndromic, type 35	X-linked
X	300075	RPS6KA3		300844	Mental retardation, X-linked, type 19	X-linked
8	604712	RRM2B		612075	Mitochondrial DNA depletion syndrome, type 8A (encephalomyopathic type with renal tubulopathy) and type 8B (MNGIE type)	Autosomal recessive
X	300839	RS1		312700	Retinoschisis	X-linked
6	612647	RSPH4A		612649	Ciliary dyskinesia, primary, type 11	Autosomal recessive
6	612648	RSPH9		612650	Ciliary dyskinesia, primary, type 12	Autosomal recessive
20	610573	RSPO4		206800	Anonychia congenita	Autosomal recessive
20	608833	RTEL1		615190*	Dyskeratosis congenita, autosomal recessive type 5	Autosomal recessive*
19	180901	RYR1		255320	Minicore myopathy with external ophthalmoplegia	Autosomal recessive
13	604490	SACS		270550	Spastic ataxia, Charlevoix-Saguenay, type	Autosomal recessive
2	181031	SAG		258100	Oguchi disease, type 1	Autosomal recessive
20	606754	SAMHD1		612952	Aicardi-Goutieres syndrome, type 5	Autosomal recessive
5	607690	SAR1B		246700	Chylomicron retention disease	Autosomal recessive
19	612804	SARS2		613845	Hyperuricemia, pulmonary hypertension, renal failure, and alkalosis	Autosomal recessive
7	607444	SBDS		260400	Shwachman-Diamond syndrome	Autosomal recessive
11	607697	SBF2		604563	Charcot-Marie-Tooth disease, type 4B2	Autosomal recessive
11	602286	SC5D	SC5DL	607330	Lathosterolosis	Autosomal recessive
4	602257	SCARB2		254900	Epilepsy, progressive myoclonic, type 4, with or without renal failure	Autosomal recessive
22	613619	SCARF2		600920	Van den Ende-Gupta syndrome	Autosomal recessive
19	600235	SCN1B		617350	Epileptic encephalopathy, early infantile, type 52	Autosomal recessive
17	603967	SCN4A		614198	Myasthenic syndrome, congenital, type 16	Autosomal recessive
2	603415	SCN9A		243000	Indifference to pain and autosomal recessive hereditary sensory neuropathy type 2D	Autosomal recessive
12	600228	SCNN1A		264350	Pseudohypoaldosteronism, type 1	Autosomal recessive
16	600760	SCNN1B		264350	Pseudohypoaldosteronism, type 1	Autosomal recessive
16	600761	SCNN1G		264350	Pseudohypoaldosteronism, type 1	Autosomal recessive
17	603644	SCO1		619048	Mitochondrial complex IV deficiency, nuclear type 4	Autosomal recessive
22	604272	SCO2		604377	Cardioencephalomyopathy, fatal infantile, due to cytochrome c oxidase deficiency, type 1	Autosomal recessive
1	613524	SDCCAG8		615993	Bardet-Biedl syndrome, type 16	Autosomal recessive
5	600857	SDHA		252011; 256000	Mitochondrial respiratory chain complex II deficiency; Leigh syndrome	Autosomal recessive
19	612848	SDHAF1		252011	Mitochondrial complex II deficiency	Autosomal recessive
14	610511	SEC23A		607812	Craniolenticulosutural dysplasia	Autosomal recessive
20	610512	SEC23B		224100	Dyserythropoietic anemia, congenital, type 2	Autosomal recessive
9	607693	SECISBP2		609698	Thyroid hormone metabolism, abnormal	Autosomal recessive
1	606210	SELENON	SEPN1	602771	Muscular dystrophy, rigid spine, type 1	Autosomal recessive
1	607292	SEMA4A		610283; 610282	Cone-rod dystrophy, type 10; Retinitis pigmentosa, type 35	Autosomal recessive
4	613009	SEPSECS		613811	Pontocerebellar hypoplasia, type 2D	Autosomal recessive
1	107300	SERPINC1		613118*	Thrombophilia due to antithrombin III deficiency	Autosomal recessive*
7	173360	SERPINE1		613329*	Plasminogen activator inhibitor-1 deficiency	Autosomal recessive*
17	172860	SERPINF1		613982	Osteogenesis imperfecta, type 6	Autosomal recessive

11	606860	SERPING1	106100*	Angioedema, hereditary, types 1 and 2	Autosomal recessive*
11	600943	SERPINH1	613848	Osteogenesis imperfecta, type 10	Autosomal recessive
9	608465	SETX	606002	Spinocerebellar ataxia, autosomal recessive, type 1	Autosomal recessive
2	178640	SFTPB	265120	Surfactant metabolism dysfunction, pulmonary, type 1	Autosomal recessive
17	600119	SGCA	608099	Limb-girdle muscular dystrophy, type 3 (LGMD R3)	Autosomal recessive
4	600900	SGCB	604286	Limb-girdle muscular dystrophy, type 4 (LGMD R4)	Autosomal recessive
5	601411	SGCD	601287	Limb-girdle muscular dystrophy, type 6 (LGMD R6)	Autosomal recessive
13	608896	SGCG	253700	Limb-girdle muscular dystrophy, type 5 (LGMD R5)	Autosomal recessive
17	605270	SGSH	252900	Mucopolysaccharidosis, type 3A (Sanfilippo A)	Autosomal recessive
X	300490	SH2D1A	308240	Lymphoproliferative syndrome, X-linked, type 1	X-linked
5	613293	SH3PXD2B	249420	Frank-ter Haar syndrome	Autosomal recessive
5	608206	SH3TC2	601596	Charcot-Marie-Tooth disease, type 4C	Autosomal recessive
X	300579	SHROOM4	300434	Stocco dos Santos X-linked mental retardation syndrome	X-linked
3	609845	SI	222900	Sucrase-isomaltase deficiency, congenital	Autosomal recessive
5	608005	SIL1	248800	Marinesco-Sjogren syndrome	Autosomal recessive
14	606326	SIX6	212550	Optic disc anomalies with retinal and/or macular dystrophy	Autosomal recessive
6	600478	SKIV2L	614602	Trichohepatoenteric syndrome, type 2 (diarrhea, syndromic)	Autosomal recessive
13	601295	SLC10A2	613291	Bile acid malabsorption, primary	Autosomal recessive
15	600839	SLC12A1	601678	Bartter syndrome, type 1	Autosomal recessive
16	600968	SLC12A3	263800	Gitelman syndrome	Autosomal recessive
15	604878	SLC12A6	218000	Agenesis of the corpus callosum with peripheral neuropathy	Autosomal recessive
1	600682	SLC16A1	616095*	Monocarboxylate transporter 1 deficiency	Autosomal recessive*
X	300095	SLC16A2	300523	Allan-Herndon-Dudley syndrome	X-linked
6	604322	SLC17A5	604369	Salla disease	Autosomal recessive
1	603941	SLC19A2	249270	Thiamine-responsive megaloblastic anemia syndrome	Autosomal recessive
2	606152	SLC19A3	607483	Thiamine metabolism dysfunction syndrome, type 2 (biotin- or thiamine-responsive encephalopathy type)	Autosomal recessive
11	607096	SLC22A12	220150	Hypouricemia, renal	Autosomal recessive
5	603377	SLC22A5	212140	Carnitine deficiency, systemic primary	Autosomal recessive
15	603617	SLC24A1	613830	Night blindness, congenital stationary (complete), type 1D, autosomal recessive	Autosomal recessive
15	609802	SLC24A5	113750	Albinism, oculocutaneous, type 6	Autosomal recessive
2	603667	SLC25A12	612949	Epileptic encephalopathy, early infantile, type 39	Autosomal recessive
7	603859	SLC25A13	605814; 603471	Citrullinemia, type 2, neonatal-onset; Citrullinemia, type 2, adult-onset	Autosomal recessive
13	603861	SLC25A15	238970	Hyperornithinemia-hyperammonemia-homocitrullinemia syndrome	Autosomal recessive
17	606521	SLC25A19	607196; 613710	Microcephaly, Amish type; Thiamine metabolism dysfunction syndrome 4 (progressive polyneuropathy type)	Autosomal recessive
3	613698	SLC25A20	212138	Carnitine-acylcarnitine translocase deficiency	Autosomal recessive
11	609302	SLC25A22	609304	Epileptic encephalopathy, early infantile, type 3	Autosomal recessive
12	600370	SLC25A3	610773	Mitochondrial phosphate carrier deficiency	Autosomal recessive
3	610819	SLC25A38	205950	Anemia, sideroblastic, type 2, pyridoxine-refractory	Autosomal recessive
4	103220	SLC25A4	615418	Mitochondrial DNA depletion syndrome, type 12B (cardiomyopathic type) AR	Autosomal recessive
5	606718	SLC26A2	600972	Achondrogenesis, type 1B (diastrophic dysplasia)	Autosomal recessive
7	126650	SLC26A3	214700	Diarrhea 1, secretory chloride, congenital	Autosomal recessive
7	605646	SLC26A4	600791; 274600	Deafness, autosomal recessive, type 4; Pendred syndrome	Autosomal recessive
9	604194	SLC27A4	608649	Ichthyosis prematurity syndrome	Autosomal recessive

10	612373	SLC29A3	602782	Histiocytosis-lymphadenopathy plus syndrome	Autosomal recessive
1	138140	SLC2A1	606777*	GLUT1 deficiency syndrome 1, infantile onset, severe	Autosomal recessive*
20	606145	SLC2A10	208050	Arterial tortuosity syndrome	Autosomal recessive
3	138160	SLC2A2	227810	Fanconi-Bickel syndrome	Autosomal recessive
4	606142	SLC2A9	612076*	Hypouricemia, renal, type 2	Autosomal recessive*
1	611146	SLC30A10	613280	Hypermanganesemia with dystonia, type 1	Autosomal recessive
3	603690	SLC33A1	614482	Congenital cataracts, hearing loss, and neurodegeneration	Autosomal recessive
5	182309	SLC34A1	616963	Hypercalcemia, infantile, type 2	Autosomal recessive
4	604217	SLC34A2	265100	Pulmonary alveolar microlithiasis	Autosomal recessive
9	609826	SLC34A3	241530	Hypophosphatemic rickets with hypercalciuria	Autosomal recessive
6	605634	SLC35A1	603585	Congenital disorder of glycosylation, type 2F	Autosomal recessive
1	605632	SLC35A3	615553	?Arthrogryposis, mental retardation, and seizures	Autosomal recessive
11	605881	SLC35C1	266265	Congenital disorder of glycosylation, type 2C	Autosomal recessive
1	610804	SLC35D1	269250	Schneckenbecken dysplasia	Autosomal recessive
11	602671	SLC37A4	232220	Glycogen storage disease, type 1B	Autosomal recessive
8	607059	SLC39A4	201100	Acrodermatitis enteropathica	Autosomal recessive
2	104614	SLC3A1	220100*	Cystinuria	Autosomal recessive*
5	606202	SLC45A2	606574	Albinism, oculocutaneous, type 4	Autosomal recessive
17	611672	SLC46A1	229050	Folate malabsorption, hereditary	Autosomal recessive
17	109270	SLC4A1	611590	Distal renal tubular acidosis	Autosomal recessive
20	610206	SLC4A11	217700	Corneal endothelial dystrophy, autosomal recessive	Autosomal recessive
4	603345	SLC4A4	604278	Renal tubular acidosis, proximal, with ocular abnormalities	Autosomal recessive
8	607882	SLC52A2	614707	Brown-Vialetto-Van Laere syndrome, type 2	Autosomal recessive
20	613350	SLC52A3	211530	Brown-Vialetto-Van Laere syndrome, type 1	Autosomal recessive
16	182381	SLC5A2	233100*	Renal glucosuria	Autosomal recessive*
19	601843	SLC5A5	274400	Thyroid dyshormonogenesis, type 1	Autosomal recessive
2	608761	SLC5A7	617143	Myasthenic syndrome, congenital, type 20, presynaptic	Autosomal recessive
5	608893	SLC6A19	234500	Hartnup disorder	Autosomal recessive
5	126455	SLC6A3	613135	Parkinsonism-dystonia, infantile	Autosomal recessive
11	604159	SLC6A5	614618*	Hyperekplexia, type 3	Autosomal recessive*
X	300036	SLC6A8	300352	Cerebral creatine deficiency syndrome, type 1	X-linked
14	603593	SLC7A7	222700	Lysinuric protein intolerance	Autosomal recessive
19	604144	SLC7A9	220100*	Cystinuria	Autosomal recessive*
X	300231	SLC9A6	300243	Mental retardation, X-linked syndromic, Christianson type	X-linked
3	601460	SLCO2A1	614441	Hypertrophic osteoarthropathy, primary, autosomal recessive, type 2	Autosomal recessive
8	606119	SLURP1	248300	Meleda disease	Autosomal recessive
2	606622	SMARCAL1	242900	Schimke immunoosseous dysplasia	Autosomal recessive
X	300040	SMC1A	300590	Cornelia de Lange syndrome 2	X-linked
5	600354	SMN1	253300	Spinal muscular atrophy	Autosomal recessive
14	608488	SMOC1	206920	Microphthalmia. with limb anomalies	Autosomal recessive
6	607223	SMOC2	125400	Dentin dysplasia, type 1, with microdontia and misshapen teeth	Autosomal recessive
11	607608	SMPD1	257200; 607616	Niemann-Pick disease, type A; Niemann-Pick disease, type B	Autosomal recessive
X	300226	SMPX	300066	Deafness, X-linked, type 4	X-linked
X	300105	SMS	309583	Mental retardation, X-linked, Snyder-Robinson type	X-linked

22	604202	SNAP29		609528	Cerebral dysgenesis, neuropathy, ichthyosis, and palmoplantar keratoderma syndrome	Autosomal recessive
7	614780	SNX10		615085	Osteopetrosis, autosomal recessive, type 8	Autosomal recessive
21	147450	SOD1		618598; 105400*	Spastic tetraplegia and axial hypotonia, progressive; Amyotrophic lateral sclerosis, type 1	Autosomal recessive; Autosomal recessive*
9	610224	SOHLH1		617690	Ovarian dysgenesis 5	Autosomal recessive
17	605740	SOST		269500; 239100	Sclerosteosis, type 1; Van Buchem disease	Autosomal recessive
20	601618	SOX18		607823	Hypotrichosis-lymphedema-telangiectasia syndrome	Autosomal recessive
2	604457	SP110		235550	Hepatic venoocclusive disease with immunodeficiency	Autosomal recessive
13	607111	SPART	SPG20	275900	Spactic paraplegia, type 20, autosomal recessive	Autosomal recessive
14	609868	SPATA7		604232	Leber congenital amaurosis, type 3	Autosomal recessive
15	610844	SPG11		602099	Amyotrophic lateral sclerosis, type 5, juvenile	Autosomal recessive
15	608181	SPG21		248900	Mast syndrome	Autosomal recessive
16	602783	SPG7		607259	Spastic paraplegia, type 7, autosomal recessive	Autosomal recessive
5	167790	SPINK1		608189*	Tropical calcific pancreatitis	Autosomal recessive*
5	605010	SPINK5		256500	Netherton syndrome	Autosomal recessive
19	605124	SPINT2		270420	Diarrhea 3, secretory sodium, congenital, syndromic	Autosomal recessive
2	182125	SPR		612716*	Dystonia, dopa-responsive, due to sepiapterin reductase deficiency	Autosomal recessive*
1	182860	SPTA1		266140; 270970	Pyropoikilocytosis; Apherocytosis, type 3	Autosomal recessive
11	604985	SPTBN2		615386	Spinocerebellar ataxia, autosomal recessive, type 14	Autosomal recessive
5	601530	SQSTM1		617145	Neurodegeneration with ataxia, dystonia, and gaze palsy, childhood-onset	Autosomal recessive
2	607306	SRD5A2		264600	46,XY disorder of sex development due to 5-alpha-reductase 2 deficiency (pseudovaginal perineoscrotal hypospadias)	Autosomal recessive
4	611715	SRD5A3		612379; 612713	Congenital disorder of glycosylation, type 1Q; Kahrizi syndrome	Autosomal recessive
11	606797	ST14		602400	Ichthyosis, congenital, autosomal recessive, type 11	Autosomal recessive
2	604402	ST3GAL5		609056	Salt and pepper developmental regression syndrome	Autosomal recessive
8	600617	STAR		201710	Lipoid adrenal hyperplasia	Autosomal recessive
2	600555	STAT1		613796	Immunodeficiency, type 31B, mycobacterial and viral infections	Autosomal recessive
17	604260	STAT5B		245590	Laron syndrome with immunodeficiency	Autosomal recessive
1	181590	STIL		612703	Microcephaly, type 7, primary, autosomal recessive	Autosomal recessive
11	605921	STIM1		612783	Immunodeficiency, type 10	Autosomal recessive
20	604965	STK4		614868	T-cell immunodeficiency, recurrent infections, autoimmunity, and cardiac malformations	Autosomal recessive
15	610745	STRA6		601186	Microphthalmia, isolated, with coloboma, type 8	Autosomal recessive
17	608626	STRADA		611087	Polyhydramnios, megalencephaly, and symptomatic epilepsy	Autosomal recessive
15	606440	STRC		603720	Deafness, autosomal recessive, type 16	Autosomal recessive
X	300747	STS		308100	Ichthyosis, X-linked	X-linked
6	605014	STX11		603552	Hemophagocytic lymphohistiocytosis, familial, type 4	Autosomal recessive
19	601717	STXBP2		613101	Hemophagocytic lymphohistiocytosis, familial, type 5	Autosomal recessive
13	603921	SUCLA2		612073	Mitochondrial DNA depletion syndrome, type 5 (encephalomyopathic with or without methylmalonic aciduria)	Autosomal recessive
2	611224	SUCLG1		245400	Mitochondrial DNA depletion syndrome, type 9 (encephalomyopathic, type with methylmalonic aciduria)	Autosomal recessive
10	607035	SUFU		617757	Joubert syndrome, type 32	Autosomal recessive
7	609187	SUGCT		231690	Glutaric aciduria, type 3	Autosomal recessive
19	604125	SULT2B1		617571	Ichthyosis, congenital, autosomal recessive, type 14	Autosomal recessive
3	607939	SUMF1		272200	Multiple sulfatase deficiency	Autosomal recessive
12	606887	SUOX		272300	Sulfite oxidase deficiency	Autosomal recessive

9	185620	SURF1	616684; 256000	Charcot-Marie-Tooth disease, type 4K; Leigh syndrome, due to COX IV deficiency	Autosomal recessive
X	313440	SYN1	300491	Epilepsy, X-linked, with variable learning disabilities and behavior disorders	X-linked
6	608441	SYNE1	610743	Spinocerebellar ataxia, autosomal recessive, type 8	Autosomal recessive
1	610949	SYT14	614229	?Spinocerebellar ataxia, autosomal recessive, type 11	Autosomal recessive
12	162330	TAC3	614839	Hypogonadotropic hypogonadism, type 10, with or without anosmia	Autosomal recessive
17	612958	TACO1	619052	Mitochondrial complex IV deficiency, nuclear type 8	Autosomal recessive
4	162332	TACR3	614840	Hypogonadotropic hypogonadism, type 11, with or without anosmia	Autosomal recessive
1	137290	TACSTD2	204870	Corneal dystrophy, gelatinous drop-like	Autosomal recessive
11	602063	TALDO1	606003	Transaldolase deficiency	Autosomal recessive
16	613018	TAT	276600	Tyrosinemia, type 2	Autosomal recessive
X	300394	TAZ	302060	3-methylglutaconic aciduria, type 2 (Barth syndrome)	X-linked
3	617687	TBC1D23	617695	Pontocerebellar hypoplasia, type 11	Autosomal recessive
16	613577	TBC1D24	220500; 615338; 614617	DOORS (deafness, onychodystrophy, osteodystrophy, mental retardation, and seizures) syndrome; Epileptic encephalopathy, early infantile, type 16; Deafness, autosomal recessive, type 86	Autosomal recessive
1	604934	TBCE	617207; 241410; 244460	Encephalopathy, progressive, with amyotrophy and optic atrophy; Hypoparathyroidism-retardation-dysmorphism syndrome; Kenny-Caffey syndrome, type 1	Autosomal recessive
1	604614	TBX19	201400	Congenital isolated adrenocorticotropic hormone deficiency	Autosomal recessive
X	300307	TBX22	303400	Cleft palate with ankyloglossia	X-linked
7	274180	TBXAS1	231095	Ghosal syndrome	Autosomal recessive
17	604488	TCAP	601954	Limb-girdle muscular dystrophy, type 7 (LGMD R7)	Autosomal recessive
11	604592	TCIRG1	259700	Osteopetrosis, autosomal recessive, type 1	Autosomal recessive
22	613441	TCN2	275350	Transcobalamin II deficiency	Autosomal recessive
12	609863	TCTN1	614173	Joubert syndrome, type 13	Autosomal recessive
12	613846	TCTN2	616654; 613885	Joubert syndrome, type 24; ?Meckel syndrome, type 8	Autosomal recessive
14	607198	TDP1	607250	?Spinocerebellar ataxia, autosomal recessive with axonal neuropathy	Autosomal recessive
9	611258	TDRD7	613887	Cataract 36	Autosomal recessive
19	610057	TECR	614020	Mental retardation, autosomal recessive, type 14	Autosomal recessive
11	602574	TECTA	603629	Deafness, autosomal recessive, type 21	Autosomal recessive
5	187270	TERT	613989	Dyskeratosis congenita, autosomal recessive, type 4	Autosomal recessive
3	190000	TF	209300	Atransferrinemia	Autosomal recessive
7	604720	TFR2	604250	Hemochromatosis, type 3	Autosomal recessive
8	188450	TG	274700	Thyroid dyshormonogenesis, type 3	Autosomal recessive
14	190195	TGM1	242300	Ichthyosis, congenital, autosomal recessive, type 1	Autosomal recessive
15	603805	TGM5	609796	Peeling skin syndrome, type 2	Autosomal recessive
11	191290	TH	605407	Segawa syndrome, recessive	Autosomal recessive
3	190160	THRB	274300	Thyroid hormone resistance, autosomal recessive	Autosomal recessive
X	300356	TIMM8A	304700	Mohr-Tranebjaerg syndrome	X-linked
9	607709	TJP2	615878	Cholestasis, progressive familial intrahepatic, type 4	Autosomal recessive
16	188250	TK2	609560	Mitochondrial DNA depletion syndrome , type 2 (myopathic type)	Autosomal recessive
9	606706	TMC1	600974	Deafness, autosomal recessive, type 7	Autosomal recessive
17	605828	TMC6	226400	Epidermodysplasia verruciformis	Autosomal recessive
17	605829	TMC8	226400	Epidermodysplasia verruciformis	Autosomal recessive
1	614123	TMCO1	213980	Craniofacial dysmorphism, skeletal anomalies, and mental retardation syndrome	Autosomal recessive
11	612988	TMEM126A	612989	Optic atrophy 7	Autosomal recessive

11	613277	TMEM216	608091; 603194	Joubert syndrome, type 2; Meckel syndrome, type 2	Autosomal recessive
2	614423	TMEM237	614424	Joubert syndrome, type 14	Autosomal recessive
8	609884	TMEM67	610688; 607361; 216360	Joubert syndrome, type 6; Meckel syndrome, type 3; COACH syndrome	Autosomal recessive
8	612418	TMEM70	614052	Mitochondrial complex V (ATP synthase) deficiency, nuclear type 2	Autosomal recessive
3	607237	TMIE	600971	Deafness, autosomal recessive, type 6	Autosomal recessive
21	606635	TMPRSS15	226200	Enterokinase deficiency	Autosomal recessive
21	605511	TMPRSS3	601072	Deafness, autosomal recessive, type 8/10	Autosomal recessive
22	609862	TMPRSS6	206200	Iron-refractory iron deficiency anemia	Autosomal recessive
18	603499	TNFRSF11A	612301	Osteopetrosis, autosomal recessive, type 7	Autosomal recessive
8	602643	TNFRSF11B	239000	Paget disease of bone, type 5, juvenile-onset	Autosomal recessive
17	604907	TNFRSF13B	240500	Immunodeficiency, common variable, type 2	Autosomal recessive
13	602642	TNFSF11	259710	Osteopetrosis, autosomal recessive, type 2	Autosomal recessive
19	191041	TNNT1	605355	Nemaline myopathy , type 5, Amish type	Autosomal recessive
6	600985	TNXB	606408	Ehlers-Danlos syndrome, classic-like	Autosomal recessive
12	190450	TPI1	615512	Hemolytic anemia due to triosephosphate isomerase deficiency	Autosomal recessive
7	606370	TPK1	614458	Episodic encephalopathy due to thiamine pyrophosphokinase deficiency	Autosomal recessive
1	191030	TPM3	609284*; 255310*	Nemaline myopathy, type 1; Congenital fiber-type disproportion myopathy	Autosomal recessive*
2	606765	TPO	274500	Thyroid dyshormonogenesis, type 2A	Autosomal recessive
11	607998	TPP1	204500; 609270	Ceroid lipofuscinosis, neuronal, type 2; Spinocerebellar ataxia, autosomal recessive, type 7	Autosomal recessive
9	613354	TPRN	613307	Deafness, autosomal recessive, type 79	Autosomal recessive
2	607380	TRAF3IP1	616629	Senior-Loken syndrome, type 9	Autosomal recessive
X	300202	TRAPPC2	313400	Spondyloepiphyseal dysplasia tarda	X-linked
8	611966	TRAPPC9	613192	Mental retardation, autosomal recessive, type 13	Autosomal recessive
6	603283	TRDN	615441	Ventricular tachycardia, catecholaminergic polymorphic, type 5, with or without muscle weakness	Autosomal recessive
6	605086	TREM2	221770	Nasu-Hakola disease	Autosomal recessive
3	606609	TREX1	225750	Aicardi-Goutieres syndrome, type 1	Autosomal recessive
9	602290	TRIM32	254110	Limb-girdle muscular dystrophy, type 8 (LGMD R8)	Autosomal recessive
17	605073	TRIM37	253250	Mulibrey nanism	Autosomal recessive
22	609761	TRIOBP	609823	Deafness, autosomal recessive, type 28	Autosomal recessive
14	604505	TRIP11	200600	Achondrogenesis, type 1A	Autosomal recessive
22	610230	TRMU	613070	Liver failure, transient infantile	Autosomal recessive
15	603576	TRPM1	613216	Night blindness, congenital stationary (complete), type 1C, autosomal recessive	Autosomal recessive
9	607009	TRPM6	602014	Familial hypomagnesemia with secondary hypocalcemia	Autosomal recessive
3	608753	TSEN2	612389	Pontocerebellar hypoplasia, type 2B	Autosomal recessive
17	608755	TSEN54	277470; 225753	Pontocerebellar hypoplasia, type 2A; Pontocerebellar hypoplasia, type 4	Autosomal recessive
12	604723	TSFM	610505	Combined oxidative phosphorylation deficiency, type 3	Autosomal recessive
1	188540	TSHB	275100	Hypothyroidism, congenital, nongoitrous, type 4	Autosomal recessive
14	603372	TSHR	275200	Hypothyroidism, congenital, nongoitrous, type 1	Autosomal recessive
X	300096	TSPAN7	300210	Mental retardation, X-linked, type 58	X-linked
17	613814	TTC19	615157	Mitochondrial complex III deficiency, nuclear type 2	Autosomal recessive
2	612014	TTC21B	613819	Short-rib thoracic dysplasia, type 4, with or without polydactyly	Autosomal recessive
5	614589	TTC37	222470	Trichohepatoenteric syndrome, type 1 (diarrhea, syndromic)	Autosomal recessive

14	608132	TTC8		615985	Bardet-Biedl syndrome, type 8	Autosomal recessive
8	614426	TTI2		615541	Mental retardation, autosomal recessive, type 39	Autosomal recessive
2	188840	TTN		608807; 611705	Limb-girdle muscular dystrophy type 10 (LGMDR10); Early-onset myopathy with fatal cardiomyopathy (Salih myopathy)	Autosomal recessive
8	600415	TTPA		277460	Ataxia with isolated vitamin E deficiency	Autosomal recessive
22	605742	TUBA8		613180	Cortical dysplasia, complex, with other brain malformations, type 8	Autosomal recessive
15	609610	TUBGCP4		616335	Microcephaly and chorioretinopathy, autosomal recessive, type 3	Autosomal recessive
22	610053	TUBGCP6		251270	Microcephaly and chorioretinopathy, autosomal recessive, type 1	Autosomal recessive
16	602389	TUFM		610678	Combined oxidative phosphorylation deficiency 4	Autosomal recessive
6	602280	TULP1		613843	Leber congenital amaurosis, type 15	Autosomal recessive
8	601385	TUSC3		611093	Mental retardation, autosomal recessive, type 7	Autosomal recessive
2	607556	TWIST2		227260	Focal facial dermal dysplasia, type 3 (Setleis type)	Autosomal recessive
10	606075	TWNK	C10orf2	271245; 616138	Mitochondrial DNA depletion syndrome, type 7 (hepatocerebral type); Perrault syndrome type 5	Autosomal recessive
19	176941	TYK2		611521	Immunodeficiency, type 35	Autosomal recessive
22	131222	TYMP		603041	Mitochondrial DNA depletion syndrome, type 1 (MNGIE type)	Autosomal recessive
11	606933	TYR		203100; 606952	Oculocutaneous albinism (OCA) type 1A; OCA type 1B	Autosomal recessive
19	604142	TYROBP		221770	Polycystic lipomembranous osteodysplasia with sclerosing leukoencephalopathy, type 1 (Nasu-Hakola disease)	Autosomal recessive
9	115501	TYRP1		203290	Albinism, oculocutaneous, type 3	Autosomal recessive
X	312180	UBE2A		300860	Mental retardation, X-linked syndromic, Nascimento-type	X-linked
X	300264	UBQLN2		300857	Amyotrophic lateral sclerosis, type 15, with or without frontotemporal dementia	X-linked
15	605981	UBR1		243800	Johanson-Blizzard syndrome	Autosomal recessive
4	191342	UCHL1		615491	Spastic paraplegia, type 79, autosomal recessive	Autosomal recessive
2	191740	UGT1A1		606785; 218800	Crigler-Najjar syndrome, type 1; Crigler-Najjar syndrome, type 2	Autosomal recessive
3	613891	UMPS		258900	Orotic aciduria	Autosomal recessive
17	608897	UNC13D		608898	Hemophagocytic lymphohistiocytosis, familial, type 3	Autosomal recessive
2	612636	UNC80		616801	Hypotonia, infantile, with psychomotor retardation and characteristic facies, type 2	Autosomal recessive
12	191525	UNG		608106	Immunodeficiency with hyper IgM, type 5	Autosomal recessive
22	606673	UPB1		613161	Beta-ureidopropionase deficiency	Autosomal recessive
X	300298	UPF3B		300676	Mental retardation, X-linked, syndromic, type 14	X-linked
8	191330	UQCRB		615158	Mitochondrial complex III deficiency, nuclear, type 3	Autosomal recessive
5	612080	UQCRQ		615159	Mitochondrial complex III deficiency, nuclear, type 4	Autosomal recessive
1	613521	UROD		176100	Porphyria cutanea tarda	Autosomal recessive
10	606938	UROS		263700	Porphyria, congenital erythropoietic	Autosomal recessive
16	613276	USB1		604173	Poikiloderma with neutropenia	Autosomal recessive
11	605242	USH1C		276904; 602092	Usher syndrome, type 1C; Deafness, autosomal recessive, type 18A	Autosomal recessive
17	607696	USH1G		606943	Usher syndrome, type 1G	Autosomal recessive
1	608400	USH2A		276901	Usher syndrome, type 2A	Autosomal recessive
X	300072	USP9X		300919; 300968	Mental retardation, X-linked, type 99; Mental retardation, X-linked, type 99, syndromic, female-restricted	X-linked
4	614632	UVSSA		614640	UV-sensitive syndrome, type 3	Autosomal recessive
12	601769	VDR		277440	Rickets, vitamin D-resistant, type 2A	Autosomal recessive
14	613401	VIPAS39	VIPAR	613404	Arthrogryposis, renal dysfunction and cholestasis, type 2	Autosomal recessive
16	608547	VKORC1		607473	Vitamin K-dependent clotting factors, combined deficiency of, type 2	Autosomal recessive
9	192977	VLDLR		224050	Cerebellar hypoplasia and mental retardation with or without quadrupedal locomotion, type 1	Autosomal recessive
X	300913	VMA21		310440	Myopathy, X-linked, with excessive autophagy	X-linked

9	605978	VPS13A		200150	Choreoacanthocytosis	Autosomal recessive
8	607817	VPS13B		216550	Cohen syndrome	Autosomal recessive
15	608552	VPS33B		208085	Arthrogryposis, renal dysfunction and cholestasis, type 1	Autosomal recessive
14	602168	VRK1		607596	Pontocerebellar hypoplasia, type 1A	Autosomal recessive
14	142993	VSX2		610092; 610093	Microphthalmia with coloboma 3; Isolated microphthalmia 2	Autosomal recessive
12	613160	VWF		277480	von Willibrand disease, type 3	Autosomal recessive
X	300392	WAS		301000; 313900	Wiskott-Aldrich syndrome; Thrombocytopenia, X-linked	X-linked
12	615748	WASHC4	KIAA1033	615817	?Mental retardation, autosomal recessive, type 43	Autosomal recessive
8	610657	WASHC5	KIAA0196	220210	Ritscher-Schinzel syndrome, type 1	Autosomal recessive
4	608151	WDR19		614377; 616307	Nephronophthisis, type 13; Senior-Loken syndrome, type 8	Autosomal recessive
2	613602	WDR35		613610	Cranioectodermal dysplasia 2	Autosomal recessive
X	300526	WDR45		300894	Neurodegeneration with brain iron accumulation, type 5	X-linked
17	609226	WDR45B		617977	Neurodevelopmental disorder with spastic quadriplegia and brain abnormalities with or without seizures	Autosomal recessive
19	613583	WDR62		604317	Microcephaly, type 2, primary, autosomal recessive, with or without cortical malformations	Autosomal recessive
15	613214	WDR72		613211	Amelogenesis imperfecta, type 2A3 (hypomaturation type)	Autosomal recessive
17	614218	WDR81		610185	Cerebellar ataxia, mental retardation, and dysequilibrium syndrome, type 2	Autosomal recessive
4	606201	WFS1		222300	Wolfram syndrome, type 1	Autosomal recessive
9	607928	WHRN	DFNB31	611383; 607084	Usher syndrome, type 2D; Deafness, autosomal recessive, type 31	Autosomal recessive
12	605232	WNK1		201300	Neuropathy, hereditary sensory and autonomic, type 2	Autosomal recessive
2	606268	WNT10A		257980	Odontoonychodermal dysplasia	Autosomal recessive
12	601906	WNT10B		225300	Split-hand/foot malformation, type 6	Autosomal recessive
3	601570	WNT7A		228930	Fuhrmann syndrome	Autosomal recessive
17	612661	WRAP53		613988	Dyskeratosis congenita, autosomal recessive, type 3	Autosomal recessive
8	604611	WRN		277700	Werner syndrome	Autosomal recessive
16	605131	WWOX		616211; 614322	Epileptic encephalopathy, early infantile, type 28; Spinocerebellar ataxia, autosomal recessive, type 12	Autosomal recessive
2	607633	XDH		278300	Xanthinuria, type 1	Autosomal recessive
X	300079	XIAP		300635	Lymphoproliferative syndrome, X-linked, 2	X-linked
X	314850	XK		300842	McLeod syndrome with or without chronic granulomatous disease	X-linked
9	611153	XPA		278700	Xeroderma pigmentosum, group A	Autosomal recessive
3	613208	XPC		278720	Xeroderma pigmentosum, group C	Autosomal recessive
22	613553	XPNPEP3		613159	Nephronophthisis-like nephropathy, type 1	Autosomal recessive
5	194363	XRCC4		616541	Short stature, microcephaly, and endocrine dysfunction	Autosomal recessive
16	608124	XYLT1		615777	Desbuquois dysplasia, type 2	Autosomal recessive
17	608125	XYLT2		605822	Spondyloocular syndrome	Autosomal recessive
12	610957	YARS2		613561	Myopathy, lactic acidosis, and sideroblastic anemia, type 2	Autosomal recessive
1	607860	YY1AP1		602531	Grange syndrome	Autosomal recessive
2	176947	ZAP70		617006; 269840	Autoimmune disease, multisystem, infantile-onset, type 2; Immunodeficiency, type 48	Autosomal recessive
11	176797	ZBTB16		612447	Skeletal defects, genital hypoplasia, and mental retardation	Autosomal recessive
6	614064	ZBTB24		614069	Immunodeficiency-centromeric instability-facial anomalies syndrome, type 2	Autosomal recessive
14	613279	ZC3H14		617125	Mental retardation, autosomal recessive, type 56	Autosomal recessive
X	300646	ZDHHC9		300799	Mental retardation, X-linked syndromic, Raymond type	X-linked
14	612012	ZFYVE26		270700	Spastic paraplegia, type 15, autosomal recessive	Autosomal recessive
X	300265	ZIC3		306955; 314390	Heterotaxy, visceral, 1, X-linked; X-linked VACTERL syndrome with or without hydrocephalus	X-linked
1	606480	ZMPSTE24		608612	Mandibuloacral dysplasia with, type B lipodystrophy	Autosomal recessive

16	612078	ZNF469	229200	Brittle cornea syndrome, type 1	Autosomal recessive
X	314990	ZNF711	300803	Mental retardation, X-linked, type 97	X-linked