Igenomix
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CGT Plus v5.4.8

Chromosome	Gene symbol	OMIM (gene)	Disease name (phenotype)	Inheritance
12	AAAS	605378	Triple-A syndrome (achalasia-addisonianism-alacrimia)	Autosomal recessive
2	ABCA12	607800	Ichthyosis, congenital, autosomal recessive, type 4A; ICAR, type 4B (harlequin)	Autosomal recessive
16	ABCA3	601615	Surfactant metabolism dysfunction, pulmonary, type 3	Autosomal recessive
1	ABCA4	601691	Stargardt disease 1; Retinitis pigmentosa 19; Cone-rod dystrophy 3	Autosomal recessive
2	ABCB11	603201	Cholestasis, benign recurrent intrahepatic, type 2; Cholestasis, progressive familial intrahepatic, type 2	Autosomal recessive
11	ABCC8	600509	Hyperinsulinemic hypoglycemia, type 1 (congenital hyperinsulinism); Permanent neonatal diabetes mellitus (PNDM)	Autosomal recessive*
X	ABCD1	300371	Adrenoleukodystrophy	X-linked
14	ABCD4	603214	Methylmalonic aciduria and homocystinuria, cblJ type	Autosomal recessive
11	ACAD8	604773	Isobutyryl-CoA dehydrogenase deficiency	Autosomal recessive
3	ACAD9	611103	Acyl-CoA dehydrogenase 9 deficiency (mitochondrial complex I deficiency, nuclear, type 20)	Autosomal recessive
1	ACADM	607008	Medium-chain acyl-CoA dehydrogenase deficiency	Autosomal recessive
12	ACADS	606885	Short-chain acyl-CoA dehydrogenase deficiency	Autosomal recessive
10	ACADSB	600301	Short/branched-chain acyl-CoA dehydrogenase deficiency	Autosomal recessive
17	ACADVL	609575	Very long-chain acyl-CoA dehydrogenase (VLCAD) deficiency	Autosomal recessive
11	ACAT1	607809	Alpha-methylacetoacetic aciduria (3-ketothiolase deficiency)	Autosomal recessive
17	ACOX1	609751	Peroxisomal acyl-CoA oxidase deficiency	Autosomal recessive
16	ACSF3	614245	Combined malonic and methylmalonic aciduria	Autosomal recessive
20	ADA	608958	Severe combined immunodeficiency due to adenosine deaminase deficiency (ADA)	Autosomal recessive
5	ADAMTS2	604539	Ehlers-Danlos syndrome, dermatosparaxis type	Autosomal recessive
16	ADGRG1	604110	Polymicrogyria, bilateral frontoparietal	Autosomal recessive
5	ADGRV1	602851	Usher syndrome, type 2C	Autosomal recessive; Digenic inheritance (PDZD7 gene)
10	ADK	102750	Hypermethioninemia due to adenosine kinase deficiency	Autosomal recessive
4	AGA	613228	Aspartylglucosaminuria (glycosylasparaginase deficiency)	Autosomal recessive
1	AGL	610860	Glycogen storage disease, type 3	Autosomal recessive
2	AGPS	603051	Rhizomelic chondrodysplasia punctata, type 3	Autosomal recessive
2	AGXT	604285	Hyperoxaluria, primary, type 1	Autosomal recessive
20	AHCY	180960	Hypermethioninemia with deficiency of S-adenosylhomocysteine hydrolase	Autosomal recessive
6	AHI1	608894	Joubert syndrome, type 3	Autosomal recessive
17	AIPL1	604392	Leber congenital amaurosis, type 4	Autosomal recessive
21	AIRE	607358	Autoimmune polyendocrinopathy syndrome, type 1	Autosomal recessive*
17	ALDH3A2	609523	Sjogren-Larsson syndrome	Autosomal recessive
1	ALDH4A1	606811	Hyperprolinemia, type 2	Autosomal recessive
9	ALDOB	612724	Fructose intolerance, hereditary	Autosomal recessive
16	ALG1	605907	Congenital disorder of glycosylation, type 1K	Autosomal recessive
1	ALG6	604566	Congenital disorder of glycosylation, type 1C	Autosomal recessive
2	ALMS1	606844	Alström syndrome	Autosomal recessive
1	ALPL	171760	ALPL-related conditions	Autosomal recessive
3	AMT	238310	Glycine encephalopathy	Autosomal recessive
3	ANO10	613726	Spinocerebellar ataxia, autosomal recessive, type 10	Autosomal recessive
X	AP1S2	300629	Mental retardation, X-linked, syndromic, type 5 (Pettigrew syndrome)	X-linked
12	AQP2	107777	Diabetes insipidus, nephrogenic, type 2	Autosomal recessive*
X	AR	313700	Androgen insensitivity syndrome	X-linked
6	ARG1	608313	Argininemia (arginase deficiency)	Autosomal recessive
3	ARL13B	608922	Joubert syndrome type 8	Autosomal recessive
22	ARSA	607574	Metachromatic leukodystrophy	Autosomal recessive
5	ARSB	611542	Mucopolysaccharidosis, type 6 (Maroteaux-Lamy syndrome)	Autosomal recessive

X	ARSL	300180	Chondrodysplasia punctata, brachytelephalangic	X-linked
X	ARX	300382	Epileptic encephalopathy, early infantile, type 1; ARX-related developmental disorders	X-linked
7	ASL	608310	Argininosuccinic aciduria	Autosomal recessive
7	ASNS	108370	Asparagine synthetase deficiency	Autosomal recessive
17	ASPA	608034	Canavan disease	Autosomal recessive
9	ASS1	603470	Citrullinemia, type 1	Autosomal recessive
11	ATM	607585	ATM-related conditions	Autosomal recessive
2	ATP6V1B1	192132	Renal tubular acidosis with deafness	Autosomal recessive
X	ATP7A	300011	Menkes disease; Occipital horn syndrome	X-linked
13	ATP7B	606882	Wilson disease	Autosomal recessive
18	ATP8B1	602397	Cholestasis, progressive familial intrahepatic, type 1; Cholestasis, benign recurrent intrahepatic, type 1	Autosomal recessive
X	ATRX	300032	Mental retardation-hypotonic facies syndrome, X-linked; Alpha-thalassemia/mental retardation syndrome	X-linked
9	AUH	600529	3-methylglutaconic aciduria, type 1	Autosomal recessive
9	B4GALT1	137060	Congenital disorder of glycosylation, type 2D	Autosomal recessive
11	BBS1	209901	Bardet-Biedl syndrome, type 1	Autosomal recessive
12	BBS10	610148	Bardet-Biedl syndrome, type 10	Autosomal recessive
4	BBS12	610683	Bardet-Biedl syndrome, type 12	Autosomal recessive
16	BBS2	606151	Bardet-Biedl syndrome, type 2	Autosomal recessive
3	BCHE	177400	Butyrylcholinesterase deficiency	Autosomal recessive
19	BCKDHA	608348	Maple syrup urine disease, type 1A	Autosomal recessive
6	BCKDHB	248611	Maple syrup urine disease, type 1B	Autosomal recessive
2	BCS1L	603647	Mitochondrial complex III deficiency nuclear type 1; GRACILE syndrome; Bjornstad syndrome	Autosomal recessive
15	BLM	604610	Bloom syndrome	Autosomal recessive
X	BRWD3	300553	Mental retardation, X-linked, type 93	X-linked
1	BSND	606412	Bartter syndrome, type 4A	Autosomal recessive
3	BTD	609019	Biotinidase deficiency	Autosomal recessive
X	BTK	300300	Agammaglobulinemia X-linked, type 1	X-linked
8	CA2	611492	Osteopetrosis with renal tubular acidosis (osteopetrosis, autosomal recessive, type 3)	Autosomal recessive
15	CAPN3	114240	Limb-girdle muscular dystrophy, type 1 (LGMD R1)	Autosomal recessive
1	CASQ2	114251	Ventricular tachycardia, catecholaminergic polymorphic, type 2	Autosomal recessive
21	CBS	613381	Homocystinuria due to cystathionine beta-synthase	Autosomal recessive
4	CC2D2A	612013	Joubert syndrome, type 9; Meckel syndrome, type 6; COACH syndrome, 2	Autosomal recessive
14	CCDC88C	611204	Hydrocephalus, congenital, type 1	Autosomal recessive
6	CCN6	603400	Progressive pseudorheumatoid dysplasia	Autosomal recessive
X	CD40LG	300386	Hyper-IgM syndrome, type 1 (immunodeficiency, X-linked, with hyper-IgM, type 1)	X-linked
10	CDH23	605516	Deafness, autosomal recessive, type 12; Usher syndrome, type 1D	Autosomal recessive
12	CEP290	610142	Meckel syndrome, type 4; Joubert syndrome, type 5; Leber congenital amaurosis, type 10	Autosomal recessive
2	CERKL	608381	Retinitis pigmentosa, type 26	Autosomal recessive
7	CFTR	602421	Cystic fibrosis	Autosomal recessive
10	CHAT	118490	Myasthenic syndrome, congenital, type 6, presynaptic	Autosomal recessive
X	CHM	300390	Choroideremia	X-linked
17	CHRNE	100725	Myasthenic syndrome, congenital, type 4B, fast-channel; Myasthenic syndrome, congenital, type 4C, associated with acetylcholine receptor deficiency	Autosomal recessive
2	CHRNG	100730	Multiple pterygium syndrome (MPS), Escobar type; MPS, lethal type	Autosomal recessive
16	CHST6	605294	Macular corneal dystrophy	Autosomal recessive
16	CIITA	600005	Bare lymphocyte syndrome, type 2, complementation group A	Autosomal recessive
7	CLCN1	118425	Myotonia congenita, recessive	Autosomal recessive
16	CLN3	607042	Ceroid lipofuscinosis, neuronal, type 3	Autosomal recessive
13	CLN5	608102	Ceroid lipofuscinosis, neuronal, type 5	Autosomal recessive

15	CLN6	606725	Ceroid lipofuscinosis, neuronal, type 6	Autosomal recessive
8	CLN8	607837	Ceroid lipofuscinosis, neuronal, type 8	Autosomal recessive
3	CLRN1	606397	Usher syndrome, type 3A	Autosomal recessive
4	CNGA1	123825	Retinitis pigmentosa type 49	Autosomal recessive
16	CNGB1	600724	Retinitis pigmentosa type 45	Autosomal recessive
8	CNGB3	605080	Achromatopsia, type 3	Autosomal recessive
9	COL27A1	608461	Steel syndrome	Autosomal recessive
2	COL4A3	120070	Alport syndrome, autosomal recessive, type 2	Autosomal recessive; Autosomal dominant
2	COL4A4	120131	Alport syndrome, autosomal recessive, type 2	Autosomal recessive; Autosomal dominant
X	COL4A5	303630	Alport syndrome, X-linked	X-linked
3	COL7A1	120120	Dystrophic epidermolysis bullosa (DEB), Hallopeau-Siemens (HS) type and non-HS type; DEB pruriginosa; DEB pretibial	Autosomal recessive; Autosomal recessive*; Autosomal recessive*
3	COLQ	603033	Myasthenic syndrome, congenital, type 5	Autosomal recessive
10	COX15	603646	Cardioencephalomyopathy, fatal infantile, due to cytochrome c oxidase deficiency, type 2; Leigh syndrome due to cytochrome c oxidase deficiency	Autosomal recessive
2	CPS1	608307	Carbamoylphosphate synthetase 1 deficiency	Autosomal recessive
11	CPT1A	600528	Carnitine palmitoyltransferase type 1A deficiency, hepatic	Autosomal recessive
1	CPT2	600650	Carnitine palmitoyltransferase type 2 deficiency, lethal neonatal; Carnitine palmitoyltransferase type 2 deficiency, infantile	Autosomal recessive
1	CRB1	604210	Retinitis pigmentosa, type 12; Leber congenital amaurosis, type 8	Autosomal recessive
3	CRTAP	605497	Osteogenesis imperfecta, type 7	Autosomal recessive
1	CTH	607657	Cystathioninuria	Autosomal recessive
17	CTNS	606272	Nephropathic cystinosis	Autosomal recessive
20	CTSA	613111	Galactosialidosis	Autosomal recessive
11	CTSC	602365	Haim-Munk syndrome; Papillon-Lefevre syndrome	Autosomal recessive
11	CTSD	116840	Ceroid lipofuscinosis, neuronal, type 10	Autosomal recessive
1	CTSK	601105	Pycnodysostosis	Autosomal recessive
X	CUL4B	300304	Mental retardation, X-linked, syndromic, type 15 (Cabezas type)	X-linked
16	CYBA	608508	Chronic granulomatous disease, type 4	Autosomal recessive
X	CYBB	300481	Chronic granulomatous disease, X-linked	X-linked
15	CYP11A1	118485	46,XY disorder of sex development-adrenal insufficiency due to CYP11A1 deficiency	Autosomal recessive
8	CYP11B1	610613	Adrenal hyperplasia, congenital, due to 11-beta-hydroxylase deficiency	Autosomal recessive
8	CYP11B2	124080	Hypoaldosteronism, congenital, due to CMO I deficiency	Autosomal recessive
10	CYP17A1	609300	17 alpha(?)-hydroxylase/17,20-lyase deficiency	Autosomal recessive
15	CYP19A1	107910	Aromatase deficiency	Autosomal recessive
2	CYP1B1	601771	Glaucoma, primary congenital, type 3A	Autosomal recessive
6	CYP21A2	613815	Congenital adrenal hyperplasia due to 21-hydroxylase deficiency	Autosomal recessive
2	CYP27A1	606530	Cerebrotendinous xanthomatosis	Autosomal recessive
12	CYP27B1	609506	Vitamin D-dependent rickets, type 1	Autosomal recessive
1	DBT	248610	Maple syrup urine disease, type 2	Autosomal recessive
10	DCLRE1C	605988	Omenn syndrome; Severe combined immunodeficiency, Athabascan type	Autosomal recessive
X	DCX	300121	Lissencephaly, X-linked, type 1	X-linked
11	DDB2	600811	Xeroderma pigmentosum, complementation group E	Autosomal recessive
11	DHCR7	602858	Smith-Lemli-Opitz syndrome	Autosomal recessive
1	DHDDS	608172	Retinitis pigmentosa, type 59	Autosomal recessive
X	DKC1	300126	Dyskeratosis congenita, X-linked	X-linked
7	DLD	238331	Dihydrolipoamide dehydrogenase deficiency	Autosomal recessive
X	DLG3	300189	Mental retardation, X-linked, type 90	X-linked
X	DMD	300377	DMD-related conditions	X-linked
5	DNAH5	603335	Ciliary dyskinesia, primary, type 3, with or without situs inversus	Autosomal recessive
9	DNAI1	604366	Ciliary dyskinesia, primary, type 1, with or without situs inversus	Autosomal recessive
17	DNAI2	605483	Ciliary dyskinesia, primary, type 9, with or without situs inversus	Autosomal recessive

10	DNAJC12	606060	Hyperphenylalaninemia, mild, non-BH4-deficient	Autosomal recessive
4	DOK7	610285	Fetal akinesia deformation sequence, type 3; Myasthenic syndrome, congenital, type 10	Autosomal recessive
9	DOLK	610746	Congenital disorder of glycosylation, type 1M	Autosomal recessive
11	DPAGT1	191350	Congenital disorder of glycosylation, type 1J; Myasthenic syndrome, congenital, type 13	Autosomal recessive
20	DPM1	603503	Congenital disorder of glycosylation, type 1E	Autosomal recessive
1	DPYD	612779	Dihydropyrimidine dehydrogenase deficiency	Autosomal recessive
15	DUOX2	606759	Thyroid dyshormonogenesis, type 6	Autosomal recessive
15	DUOXA2	612772	Thyroid dyshormonogenesis, type 5	Autosomal recessive
11	DYNC2H1	603297	Short-rib thoracic dysplasia, type 3, with or without polydactyly	Autosomal recessive
2	DYSF	603009	Miyoshi muscular dystrophy, type 1; Limb-girdle muscular dystrophy, type 2 (LGMD R2)	Autosomal recessive
X	EDA	300451	Ectodermal dysplasia, type 1, hypohidrotic, X-linked	X-linked
2	EDAR	604095	Ectodermal dysplasia 10B, hypohidrotic/hair/tooth type	Autosomal recessive
2	EIF2AK3	604032	Wolcott-Rallison syndrome	Autosomal recessive
3	EIF2B5	603945	Leukoencephalopathy with vanishing white matter (VWM)	Autosomal recessive
9	ELP1	603722	Familial dysautonomia	Autosomal recessive
X	EMD	300384	Emery-Dreifuss muscular dystrophy, type 1, X-linked	X-linked
19	ERCC2	126340	Trichothiodystrophy, type 1; Xeroderma pigmentosum, group D	Autosomal recessive
2	ERCC3	133510	Trichothiodystrophy, type 2	Autosomal recessive
13	ERCC5	133530	Cerebrooculofacioskeletal syndrome 3; Xeroderma pigmentosum, group G;Xeroderma pigmentosum, group G/Cockayne syndrome	Autosomal recessive
10	ERCC6	609413	Cockayne syndrome, type B; Cerebrooculofacioskeletal syndrome, type 1	Autosomal recessive
5	ERCC8	609412	Cockayne syndrome, type A	Autosomal recessive
8	ESCO2	609353	Roberts syndrome	Autosomal recessive
15	ETFA	608053	Glutaric acidemia, type 2A	Autosomal recessive
19	ETFB	130410	Glutaric acidemia, type 2B	Autosomal recessive
4	ETFDH	231675	Glutaric acidemia, type 2C	Autosomal recessive
19	ETHE1	608451	Ethylmalonic encephalopathy	Autosomal recessive
4	EVC	604831	Ellis-van Creveld syndrome	Autosomal recessive
4	EVC2	607261	Ellis-van Creveld syndrome	Autosomal recessive
9	EXOSC3	606489	Pontocerebellar hypoplasia, type 1B	Autosomal recessive
6	EYS	612424	Retinitis pigmentosa, type 25	Autosomal recessive
4	F11	264900	Factor XI deficiency	Autosomal recessive*
11	F2	176930	Prothrombin deficiency	Autosomal recessive
1	F5	612309	Factor V deficiency	Autosomal recessive
X	F8	300841	Hemophilia A	X-linked
X	F9	300746	Hemophilia B	X-linked
15	FAH	613871	Tyrosinemia, type 1	Autosomal recessive
2	FAM161A	613596	Retinitis pigmentosa, type 28	Autosomal recessive
7	FAM20C	611061	Raine syndrome	Autosomal recessive
16	FANCA	607139	Fanconi anemia, complementation group A	Autosomal recessive
9	FANCC	613899	Fanconi anemia, complementation group C	Autosomal recessive
9	FANCG	602956	Fanconi anemia, complementation group G	Autosomal recessive
X	FGD1	300546	Aarskog-Scott syndrome; Mental retardation, X-linked syndromic, type 16	X-linked
1	FH	136850	Fumarase deficiency	Autosomal recessive
19	FKRP	606596	Muscular dystrophy-dystroglycanopathy, type 5A (Walker-Warburg syndrome); Type 5B; Type 5C (limb-girdle muscular dystrophy, type 9 [LGMDR9])	Autosomal recessive
9	FKTN	607440	Muscular dystrophy-dystroglycanopathy, type 4A (Walker-Warburg syndrome); Type 4B; Type 4C (limb-girdle muscular dystrophy, type 13 [LGMD R13])	Autosomal recessive
1	FMO3	136132	Trimethylaminuria	Autosomal recessive
X	FMR1	309550	FMR1-related conditions	X-linked
11	FOXRED1	613622	Mitochondrial complex I deficiency, nuclear type 19	Autosomal recessive

4	FRAS1	607830	Fraser syndrome, type 1	Autosomal recessive
21	FTCD	606806	Glutamate formiminotransferase deficiency	Autosomal recessive
X	FTSJ1	300499	Mental retardation, X-linked 44	X-linked
1	FUCA1	612280	Fucosidosis	Autosomal recessive
9	FXN	606829	Friedreich ataxia	Autosomal recessive
17	G6PC1	613742	Glycogen storage disease, type 1A	Autosomal recessive
17	G6PC3	611045	Dursun syndrome	Autosomal recessive
X	G6PD	305900	G6PD deficiency	X-linked
17	GAA	606800	Glycogen storage disease, type 2	Autosomal recessive
14	GALC	606890	Krabbe disease	Autosomal recessive
1	GALE	606953	Galactose epimerase deficiency	Autosomal recessive
17	GALK1	604313	Galactokinase deficiency with cataracts	Autosomal recessive
16	GALNS	612222	Mucopolysaccharidosis, type 4A	Autosomal recessive
9	GALT	606999	Galactosemia	Autosomal recessive
19	GAMT	601240	Cerebral creatine deficiency syndrome, type 2	Autosomal recessive
1	GBA1	606463	Gaucher disease	Autosomal recessive
3	GBE1	607839	Glycogen storage disease, type 4	Autosomal recessive
19	GCDH	608801	Glutaricaciduria, type 1	Autosomal recessive
14	GCH1	600225	Hyperphenylalaninemia, BH4-deficient, type B	Autosomal recessive
16	GCSH	238330	Multiple mitochondrial dysfunctions syndrome 7	Autosomal recessive
8	GDAP1	606598	Charcot-Marie-Tooth disease, recessive intermediate, type A	Autosomal recessive
20	GDF5	601146	Chondrodysplasia, Grebe type	Autosomal recessive
3	GFM1	606639	Combined oxidative phosphorylation deficiency, type 1	Autosomal recessive
7	GHRHR	139191	Growth hormone deficiency, isolated, type 1B	Autosomal recessive
X	GJB1	304040	Charcot-Marie-Tooth neuropathy, X-linked dominant, type 1	X-linked
13	GJB2	121011	Deafness, autosomal recessive, type 1A; Deafness, digenic, GJB2/GJB6	Autosomal recessive; Digenic inheritance (GJB6 gene)
13	GJB6	604418	Deafness, autosomal recessive, type 1B; Deafness, digenic GJB2/GJB6	Autosomal recessive; Digenic inheritance (GJB2 gene)
X	GLA	300644	Fabry disease	X-linked
3	GLB1	611458	GM1-gangliosidosis, types 1-3; Mucopolysaccharidosis, type 4B (Morquio)	Autosomal recessive
9	GLDC	238300	Glycine encephalopathy	Autosomal recessive
9	GLE1	603371	Lethal congenital contracture syndrome, type 1; Congenital arthrogryposis with anterior horn cell disease	Autosomal recessive
9	GNE	603824	Inclusion body myopathy, type 2 (Nonaka myopathy)	Autosomal recessive
6	GNMT	606628	Glycine N-methyltransferase deficiency	Autosomal recessive
12	GNPTAB	607840	Mucolipidosis 2 alpha/beta; Mucolipidosis 3 alpha/beta	Autosomal recessive
16	GNPTG	607838	Mucolipidosis III gamma	Autosomal recessive
4	GNRHR	138850	Hypogonadotropic hypogonadism, type 7, without anosmia	Autosomal recessive
12	GNS	607664	Mucopolysaccharidosis, type 3D (Sanfilippo syndrome D)	Autosomal recessive
17	GP1BA	606672	Bernard-Soulier syndrome, type A1	Autosomal recessive
22	GP1BB	138720	Bernard-Soulier syndrome, type B	Autosomal recessive
3	GP9	173515	Bernard-Soulier syndrome, type C	Autosomal recessive
X	GPR143	300808	Ocular albinism, type 1 (Nettleship-Falls type)	X-linked
9	GRHPR	604296	Hyperoxaluria, primary, type 2	Autosomal recessive
12	GRIP1	604597	Fraser syndrome 3	Autosomal recessive
20	GSS	601002	Glutathione synthetase deficiency	Autosomal recessive
17	GUCY2D	600179	Leber congenital amaurosis, type 1	Autosomal recessive
7	GUSB	611499	Mucopolysaccharidosis, type 7	Autosomal recessive
4	HADH	601609	3-hydroxyacyl-CoA dehydrogenase deficiency	Autosomal recessive
2	HADHA	600890	Long-chain 3-hydroxyl-CoA dehydrogenase (LCHAD) deficiency; Mitochondrial trifunctional protein deficiency	Autosomal recessive
2	HADHB	143450	Mitochondrial trifunctional protein deficiency	Autosomal recessive

1	HAX1	605998	Neutropenia, severe congenital, type 3, autosomal recessive	Autosomal recessive
16	HBA1	141800	Alpha thalassemia	Autosomal recessive
16	HBA2	141850	Alpha thalassemia	Autosomal recessive
11	HBB	141900	HBB-related hemoglobinopathies	Autosomal recessive
X	HCFC1	300019	Mental retardation, X-linked 3 (methylmalonic acidemia and homocysteinemia, cblX type)	X-linked
15	HEXA	606869	Tay-Sachs disease	Autosomal recessive
5	HEXB	606873	Sandhoff disease, infantile, juvenile, and adult forms	Autosomal recessive
6	HFE	613609	Hemochromatosis, type 1	Autosomal recessive
3	HGD	607474	Alkaptonuria	Autosomal recessive
8	HGSNAT	610453	Mucopolysaccharidosis type 3C (Sanfilippo syndrome C)	Autosomal recessive
1	HJV	608374	Hemochromatosis, type 2A	Autosomal recessive
21	HLCS	609018	Holocarboxylase synthetase deficiency	Autosomal recessive
1	HMGCL	613898	HMG-CoA lyase deficiency	Autosomal recessive
22	HMOX1	141250	Heme oxygenase-1 deficiency	Autosomal recessive
10	HOGA1	613597	Hyperoxaluria, primary, type 3	Autosomal recessive
12	HPD	609695	Tyrosinemia, type 3	Autosomal recessive
X	HPRT1	308000	Lesch-Nyhan syndrome	X-linked
10	HPS1	604982	Hermansky-Pudlak syndrome, type 1	Autosomal recessive
3	HPS3	606118	Hermansky-Pudlak syndrome, type 3	Autosomal recessive
X	HSD17B10	300256	HSD10 mitochondrial disease	X-linked
9	HSD17B3	605573	46,XY disorder of sex development due to 17-beta-hydroxysteroid dehydrogenase 3 deficiency	Autosomal recessive
5	HSD17B4	601860	D-bifunctional protein deficiency	Autosomal recessive
1	HSD3B2	613890	Adrenal hyperplasia, congenital, due to 3-beta-hydroxysteroid dehydrogenase 2 deficiency	Autosomal recessive
1	HSPG2	142461	Schwartz-Jampel syndrome, type 1; Dyssegmental dysplasia, Silverman-Handmaker type	Autosomal recessive
3	HYAL1	607071	?Mucopolysaccharidosis, type 9	Autosomal recessive
11	HYLS1	610693	Hydrolethalus syndrome	Autosomal recessive
20	IDH3B	604526	Retinitis pigmentosa, type 46	Autosomal recessive
X	IDS	300823	Mucopolysaccharidosis, type 2	X-linked
4	IDUA	252800	Mucopolysaccharidosis type 1	Autosomal recessive
11	IGHMBP2	600502	Charcot-Marie-Tooth disease, axonal, type 2S	Autosomal recessive
X	IL1RAPL1	300206	Mental retardation, X-linked, type 21/34	X-linked
X	IL2RG	308380	Severe combined immunodeficiency, X-linked	X-linked
15	IVD	607036	Isovaleric acidemia	Autosomal recessive
6	IYD	612025	Thyroid dyshormonogenesis, type 4	Autosomal recessive
19	JAK3	600173	Severe Combined Immunodeficiency, autosomal recessive, T-negative/B-positive type	Autosomal recessive
11	KCNJ11	600937	Hyperinsulinemic hypoglycemia, type 2 (congenital hyperinsulinism); Permanent neonatal diabetes mellitus (PNDM)	Autosomal recessive; Autosomal recessive*
X	KDM5C	314690	Mental retardation, X-linked, syndromic, Claes-Jensen type	X-linked
X	L1CAM	308840	L1 Syndrome	X-linked
6	LAMA2	156225	LAMA2-related muscular dystrophy	Autosomal recessive
18	LAMA3	600805	Junctional epidermolysis bullosa (JEB) Herlitz type; JEB non-Herlitz type	Autosomal recessive
1	LAMB3	150310	Junctional epidermolysis bullosa (JEB) Herlitz type; JEB non-Herlitz type	Autosomal recessive
1	LAMC2	150292	Junctional epidermolysis bullosa (JEB) Herlitz type; JEB non-Herlitz type	Autosomal recessive
22	LARGE1	603590	Muscular dystrophy-dystroglycanopathy, type 6A and 6B	Autosomal recessive
6	LCA5	611408	Leber congenital amaurosis, type 5	Autosomal recessive
19	LDLR	606945	Hypercholesterolemia, familial, type 1	Autosomal recessive; Autosomal dominant
1	LDLRAP1	605747	Hypercholesterolemia, familial, autosomal recessive	Autosomal recessive
2	LHCGR	152790	Leydig cell hypoplasia	Autosomal recessive
9	LHX3	600577	Pituitary hormone deficiency, combined, type 3	Autosomal recessive

5	LIFR	151443	Stuve-Wiedemann syndrome / Schwartz-Jampel type 2 syndrome	Autosomal recessive
10	LIPA	613497	Lysosomal acid lipase deficiency	Autosomal recessive
3	LIPH	607365	Hypotrichosis, type 7 or woolly hair, autosomal recessive, type 2, with or without hypotrichosis	Autosomal recessive
6	LMBRD1	612625	Methylmalonic aciduria and homocystinuria, cblF type	Autosomal recessive
18	LOXHD1	613072	Deafness, autosomal recessive, type 77	Autosomal recessive
8	LPL	609708	Lipoprotein lipase deficiency	Autosomal recessive
2	LRP2	600073	Donnai-Barrow syndrome	Autosomal recessive
2	LRPPRC	607544	Leigh syndrome, French-Canadian type	Autosomal recessive
1	LYST	606897	Chediak-Higashi syndrome	Autosomal recessive
19	MAN2B1	609458	Alpha-mannosidosis	Autosomal recessive
4	MANBA	609489	Mannosidosis, beta	Autosomal recessive
10	MAT1A	610550	Methionine adenosyltransferase deficiency, autosomal recessive	Autosomal recessive
3	MCCC1	609010	3-Methylcrotonyl-CoA carboxylase deficiency, type 1	Autosomal recessive
5	MCCC2	609014	3-Methylcrotonyl-CoA carboxylase deficiency, type 2	Autosomal recessive
2	MCEE	608419	Methylmalonyl-CoA epimerase deficiency	Autosomal recessive
19	MCOLN1	605248	Mucolipidosis type 4	Autosomal recessive
8	MCPH1	607117	Microcephaly type 1, primary, autosomal recessive	Autosomal recessive
X	MECP2	300005	Encephalopathy, neonatal severe; Rett syndrome	X-linked
11	MED17	603810	Microcephaly, postnatal progressive, with seizures and brain atrophy	Autosomal recessive
16	MEFV	608107	Familial Mediterranean fever	Autosomal recessive
15	MESP2	605195	Spondylocostal dysostosis, type 2, autosomal recessive	Autosomal recessive
4	MFSD8	611124	Ceroid lipofuscinosis, neuronal, type 7	Autosomal recessive
X	MID1	300552	Opitz GBBB syndrome, type 1	X-linked
17	MKS1	609883	Bardet-Biedl syndrome type 13; Meckel syndrome, type 1; Joubert syndrome, type 28	Autosomal recessive
22	MLC1	605908	Megalencephalic leukoencephalopathy with subcortical cysts	Autosomal recessive
16	MLYCD	606761	Malonyl-CoA decarboxylase deficiency	Autosomal recessive
4	MMAA	607481	Methylmalonic aciduria, vitamin B12-responsive	Autosomal recessive
12	MMAB	607568	Methylmalonic aciduria, vitamin B12-responsive, type cblB	Autosomal recessive
1	MMACHC	609831	Methylmalonic aciduria and homocystinuria, cblC type	Autosomal recessive; digenic inheritance (PRDX1 gene)
2	MMADHC	611935	Homocystinuria, cblD type, variant 1	Autosomal recessive
6	MMUT	609058	Methylmalonic aciduria, mut(0) type	Autosomal recessive
2	MOGS	601336	Congenital disorder of glycosylation, type 2B	Autosomal recessive
15	MPI	154550	Congenital disorder of glycosylation, type 1B	Autosomal recessive
1	MPL	159530	Thrombocytopenia, congenital amegakaryocytic	Autosomal recessive
2	MPV17	137960	Mitochondrial DNA depletion syndrome type 6 (hepatocerebral); Charcot-Marie-Tooth disease, axonal, type 2EE	Autosomal recessive
1	MTHFR	607093	Homocystinuria due to MTHFR deficiency	Autosomal recessive
X	MTM1	300415	Myotubular myopathy, X-linked	X-linked
11	MTMR2	603557	Charcot-Marie-Tooth disease, type 4B1	Autosomal recessive
1	MTR	156570	Homocystinuria-megaloblastic anemia, cblG complementation type	Autosomal recessive
5	MTRR	602568	Homocystinuria-megaloblastic anemia, cbl E type	Autosomal recessive
4	MTTP	157147	Abetalipoproteinemia	Autosomal recessive
12	MVK	251170	Mevalonic aciduria	Autosomal recessive
17	MYO15A	602666	Deafness, autosomal recessive, type 3	Autosomal recessive
11	MYO7A	276903	Usher syndrome, type 1B; Deafness, autosomal recessive, type 2	Autosomal recessive
2	NADK2	615787	2,4-dienoyl-CoA reductase deficiency	Autosomal recessive
22	NAGA	104170	Schindler disease, type I; Schindler disease, type III; Kanzaki disease	Autosomal recessive
17	NAGLU	609701	Mucopolysaccharidosis, type 3B (Sanfilippo B)	Autosomal recessive
17	NAGS	608300	N-acetylglutamate synthase deficiency	Autosomal recessive
8	NBN	602667	Nijmegen breakage syndrome	Autosomal recessive

7	NCF1	608512	Chronic granulomatous disease, type 1	Autosomal recessive
1	NCF2	608515	Chronic granulomatous disease, type 2	Autosomal recessive
X	NDP	300658	Norrie disease	X-linked
8	NDRG1	605262	Charcot-Marie-Tooth disease, type 4D	Autosomal recessive
5	NDUFAF2	609653	Mitochondrial complex I deficiency, nuclear type 10	Autosomal recessive
20	NDUFAF5	612360	Mitochondrial complex I deficiency, nuclear type 16	Autosomal recessive
5	NDUFS4	602694	Mitochondrial complex I deficiency, nuclear type 1	Autosomal recessive
5	NDUFS6	603848	Mitochondrial complex I deficiency, nuclear type 9	Autosomal recessive
19	NDUFS7	601825	Mitochondrial complex I deficiency, nuclear type 3	Autosomal recessive
11	NDUFV1	161015	Mitochondrial complex I deficiency, nuclear type 4	Autosomal recessive
2	NEB	161650	Nemaline myopathy type 2	Autosomal recessive
6	NEU1	608272	Sialidosis, type 1 and type 2	Autosomal recessive
5	NHP2	606470	Dyskeratosis congenita, autosomal recessive type 2	Autosomal recessive
19	NLRP7	609661	Hydatidiform mole, recurrent, type 1	Autosomal recessive
15	NOP10	606471	Dyskeratosis congenita, autosomal recessive type 1	Autosomal recessive
18	NPC1	607623	Niemann-Pick disease, type C1	Autosomal recessive
14	NPC2	601015	Niemann-pick disease, type C2	Autosomal recessive
2	NPHP1	607100	Joubert syndrome type 4	Autosomal recessive
19	NPHS1	602716	Nephrotic syndrome, type 1	Autosomal recessive
1	NPHS2	604766	Nephrotic syndrome, type 2	Autosomal recessive
X	NR0B1	300473	Adrenal hypoplasia, congenital	X-linked
15	NR2E3	604485	Enhanced S-cone syndrome (Goldmann-Favre); Retinitis pigmentosa, type 37	Autosomal recessive; Autosomal recessive*
1	NTRK1	191315	Insensitivity to pain, congenital, with anhidrosis	Autosomal recessive
10	OAT	613349	Gyrate atrophy of choroid and retina	Autosomal recessive
15	OCA2	611409	Oculocutaneous albinism type 2	Autosomal recessive
X	OCRL	300535	Lowe Syndrome; Dent disease type 2	X-linked
19	OPA3	606580	3-methylglutaconic aciduria, type 3	Autosomal recessive
X	OPHN1	300127	Mental retardation, X-linked, with cerebellar hypoplasia and distinctive facial appearance	X-linked
6	OSTM1	607649	Osteopetrosis, autosomal recessive type 5	Autosomal recessive
X	OTC	300461	Ornithine transcarbamylase deficiency	X-linked
2	OTOF	603681	Deafness, autosomal recessive, type 9	Autosomal recessive
1	P3H1	610339	Osteogenesis imperfecta, type 8	Autosomal recessive
12	PAH	612349	Phenylketonuria	Autosomal recessive
X	PAK3	300142	Mental retardation, X-linked, type 30	X-linked
20	PANK2	606157	Neurodegeneration with brain iron accumulation type 1	Autosomal recessive
11	PC	608786	Pyruvate carboxylase deficiency	Autosomal recessive
10	PCBD1	126090	Hyperphenylalaninemia, BH4-deficient, type D	Autosomal recessive
13	PCCA	232000	Propionic acidemia	Autosomal recessive
3	PCCB	232050	Propionic acidemia	Autosomal recessive
10	PCDH15	605514	Deafness, autosomal recessive, type 23; Usher syndrome, type 1D/F digenic	Autosomal recessive
5	PDE6A	180071	Retinitis pigmentosa type 43	Autosomal recessive
X	PDHA1	300502	Pyruvate dehydrogenase E1-alpha deficiency	X-linked
3	PDHB	179060	Pyruvate dehydrogenase E1-beta deficiency	Autosomal recessive
7	PEX1	602136	Heimler syndrome 1; Peroxisome biogenesis disorder 1A (Zellweger); Peroxisome biogenesis disorder 1B (NALD/IRD)	Autosomal recessive
1	PEX10	602859	Peroxisome biogenesis disorder, type 6A (Zellweger syndrome); Peroxisome biogenesis disorder, type 6B	Autosomal recessive
17	PEX12	601758	Peroxisome biogenesis disorder type 3A (Zellweger)	Autosomal recessive
8	PEX2	170993	Peroxisome biogenesis disorder type 5A (Zellweger)	Autosomal recessive
22	PEX26	608666	Peroxisome biogenesis disorder type 7A (Zellweger)	Autosomal recessive

12	PEX5	600414	Peroxisome biogenesis disorder type 2A (Zellweger)	Autosomal recessive
6	PEX6	601498	Peroxisome biogenesis disorder, type 4A (Zellweger syndrome); Peroxisome biogenesis disorder, type 4B; Heimler syndrome 2	Autosomal recessive; Autosomal recessive*; Autosomal recessive
6	PEX7	601757	Rhizomelic chondrodysplasia punctata, type 1	Autosomal recessive
12	PFKM	610681	Glycogen storage disease, type 7	Autosomal recessive
X	PGK1	311800	Phosphoglycerate kinase 1 deficiency	X-linked
X	PHF8	300560	Mental retardation syndrome, X-linked, Siderius type	X-linked
1	PHGDH	606879	Neu-Laxova syndrome, type 1; Phosphoglycerate dehydrogenase deficiency	Autosomal recessive
6	PKHD1	606702	Polycystic kidney disease type 4	Autosomal recessive
22	PLA2G6	603604	Infantile neuroaxonal dystrophy type 1	Autosomal recessive
1	PLOD1	153454	Ehlers-Danlos syndrome, kyphoscoliotic type, 1	Autosomal recessive
X	PLP1	300401	Pelizaeus-Merzbacher disease	X-linked
16	PMM2	601785	Congenital disorder of glycosylation, type 1A	Autosomal recessive
17	PNPO	603287	Pyridoxamine 5'-phosphate oxidase deficiency	Autosomal recessive
15	POLG	174763	POLG-related disorders	Autosomal recessive
6	POLR1C	610060	Leukodystrophy, hypomyelinating, type 11; Treacher Collins syndrome 3	Autosomal recessive
1	POMGNT1	606822	Muscular dystrophy-dystroglycanopathy, type 3A (Walker-Warburg syndrome); Type 3B; Type 3C (limb-girdle muscular dystrophy, type 15 [LGMDR15])	Autosomal recessive
9	POMT1	607423	Muscular dystrophy-dystroglycanopathy, type 1A (Walker-Warburg syndrome); Type 1B; Type 1C (limb-girdle muscular dystrophy, type 11 [LGMD R11])	Autosomal recessive
14	POMT2	607439	Muscular dystrophy-dystroglycanopathy, type 2A (Walker-Warburg syndrome); Type 2B; Type 2C (limb-girdle muscular dystrophy, type 14 [LGMD R14])	Autosomal recessive
7	POR	124015	Antley-Bixler syndrome with genital anomalies and disordered steroidogenesis	Autosomal recessive
3	POU1F1	173110	Pituitary hormone deficiency, combined, type 1	Autosomal recessive*
X	POU3F4	300039	Deafness, X-linked, type 2	X-linked
4	PPM1K	611065	?Maple syrup urine disease, mild variant	Autosomal recessive
1	PPT1	600722	Ceroid lipofuscinosis, neuronal, type 1	Autosomal recessive
X	PQBP1	300463	Renpenning syndrome	X-linked
1	PRDX1	176763	Methylmalonic aciduria and homocystinuria, cblC type, digenic	Autosomal recessive; Digenic inheritance (MMACHC gene)
10	PRF1	170280	Hemophagocytic lymphohistiocytosis, familial, type 2	Autosomal recessive
22	PRODH	606810	Hyperprolinemia, type 1	Autosomal recessive
5	PROP1	601538	Pituitary hormone deficiency, combined, type 2	Autosomal recessive
X	PRPS1	311850	PRPS1-related disoders	X-linked
10	PSAP	176801	Combined SAP deficiency	Autosomal recessive
11	PTS	612719	Hyperphenylalaninemia, BH4-deficient, type A	Autosomal recessive
12	PUS1	608109	Myopathy, lactic acidosis, and sideroblastic anemia, type 1	Autosomal recessive
11	PYGM	608455	McArdle disease	Autosomal recessive
4	QDPR	612676	Hyperphenylalaninemia, BH4-deficient, type C	Autosomal recessive
6	RAB23	606144	Carpenter syndrome	Autosomal recessive
11	RAG1	179615	Omenn syndrome; Severe combined immunodeficiency, B cell-negative	Autosomal recessive
11	RAG2	179616	Omenn syndrome; Severe combined immunodeficiency, B cell-negative	Autosomal recessive
11	RAPSN	601592	Fetal akinesia deformation sequence, type 2; Myasthenic syndrome, congenital, type 11, associated with AChR deficiency	Autosomal recessive
6	RARS2	611524	Pontocerebellar hypoplasia, type 6	Autosomal recessive
18	RAX	601881	Isolated microphthalmia, type 3	Autosomal recessive
14	RDH12	608830	Leber congenital amaurosis, type 13	Autosomal recessive
9	RMRP	157660	Anauxetic dysplasia, type 1	Autosomal recessive
13	RNASEH2B	610326	Aicardi-Goutieres syndrome, type 2	Autosomal recessive
11	RNASEH2C	610330	Aicardi-Goutieres syndrome, type 3	Autosomal recessive
X	RP2	300757	Retinitis pigmentosa, type 2, X-linked	X-linked
1	RPE65	180069	RPE65-related Leber congenital amaurosis/early-onset severe retinal dystrophy	Autosomal recessive
X	RPGR	312610	Retinitis pigmentosa, type 3, X-linked; Cone-rod dystrophy, X-linked, 1	X-linked
16	RPGRIP1L	610937	Joubert syndrome, type 7; Meckel syndrome, type 5; COACH syndrome	Autosomal recessive
X	RS1	300839	Retinoschisis	X-linked

20	RTEL1	608833	Dyskeratosis congenita, autosomal recessive type 5	Autosomal recessive*
13	SACS	604490	Spastic ataxia, Charlevoix-Saguenay, type	Autosomal recessive
2	SAG	181031	Oguchi disease, type 1	Autosomal recessive
20	SAMHD1	606754	Aicardi-Goutieres syndrome, type 5	Autosomal recessive
7	SBDS	607444	Shwachman-Diamond syndrome	Autosomal recessive
22	SCO2	604272	Cardioencephalomyopathy, fatal infantile, due to cytochrome c oxidase deficiency, type 1	Autosomal recessive
4	SEPSECS	613009	Pontocerebellar hypoplasia, type 2D	Autosomal recessive
14	SERPINA1	107400	Alpha-1 antitrypsin deficiency	Autosomal recessive
17	SGCA	600119	Limb-girdle muscular dystrophy, type 3 (LGMD R3)	Autosomal recessive
4	SGCB	600900	Limb-girdle muscular dystrophy, type 4 (LGMD R4)	Autosomal recessive
5	SGCD	601411	Limb-girdle muscular dystrophy, type 6 (LGMD R6)	Autosomal recessive
13	SGCG	608896	Limb-girdle muscular dystrophy, type 5 (LGMD R5)	Autosomal recessive
17	SGSH	605270	Mucopolysaccharidosis, type 3A (Sanfilippo A)	Autosomal recessive
X	SH2D1A	300490	Lymphoproliferative syndrome, X-linked, type 1	X-linked
5	SH3TC2	608206	Charcot-Marie-Tooth disease, type 4C	Autosomal recessive
5	SKIC3	614589	Trichohepatoenteric syndrome, type 1 (diarrhea, syndromic)	Autosomal recessive
16	SLC12A3	600968	Gitelman syndrome	Autosomal recessive
15	SLC12A6	604878	Agenesis of the corpus callosum with peripheral neuropathy	Autosomal recessive
X	SLC16A2	300095	Allan-Herndon-Dudley syndrome	X-linked
6	SLC17A5	604322	Salla disease	Autosomal recessive
1	SLC19A2	603941	Thiamine-responsive megaloblastic anemia syndrome	Autosomal recessive
2	SLC19A3	606152	Thiamine metabolism dysfunction syndrome, type 2 (biotin- or thiamine-responsive encephalopathy type)	Autosomal recessive
5	SLC22A5	603377	Carnitine deficiency, systemic primary	Autosomal recessive
7	SLC25A13	603859	Citrullinemia, type 2, neonatal-onset; Citrullinemia, type 2, adult-onset	Autosomal recessive
13	SLC25A15	603861	Hyperornithinemia-hyperammonemia-homocitrullinemia syndrome	Autosomal recessive
3	SLC25A20	613698	Carnitine-acylcarnitine translocase deficiency	Autosomal recessive
5	SLC26A2	606718	Achondrogenesis Ib; Atelosteogenesis, type II;De la Chapelle dysplasia; Diastrophic dysplasia;Diastrophic dysplasia, broad bone-platyspondylic variant;Epiphyseal dysplasia, multiple, 4	Autosomal recessive
7	SLC26A3	126650	Diarrhea 1, secretory chloride, congenital	Autosomal recessive
7	SLC26A4	605646	Deafness, autosomal recessive, type 4; Pendred syndrome	Autosomal recessive
6	SLC35A1	605634	Congenital disorder of glycosylation, type 2F	Autosomal recessive
1	SLC35A3	605632	Arthrogryposis, impaired intellectual development, and seizures	Autosomal recessive
11	SLC35C1	605881	Congenital disorder of glycosylation, type 2C	Autosomal recessive
1	SLC35D1	610804	Schneckenbecken dysplasia	Autosomal recessive
11	SLC37A4	602671	Glycogen storage disease, type 1B	Autosomal recessive
8	SLC39A4	607059	Acrodermatitis enteropathica	Autosomal recessive
2	SLC3A1	104614	Cystinuria	Autosomal recessive*
5	SLC45A2	606202	Albinism, oculocutaneous, type 4	Autosomal recessive
17	SLC46A1	611672	Folate malabsorption, hereditary	Autosomal recessive
20	SLC4A11	610206	Corneal endothelial dystrophy, autosomal recessive	Autosomal recessive
19	SLC5A5	601843	Thyroid dyshormonogenesis, type 1	Autosomal recessive
5	SLC6A19	608893	Hartnup disorder	Autosomal recessive
X	SLC6A8	300036	Cerebral creatine deficiency syndrome, type 1	X-linked
14	SLC7A7	603593	Lysinuric protein intolerance	Autosomal recessive
19	SLC7A9	604144	Cystinuria	Autosomal recessive*
2	SMARCAL1	606622	Schimke immunoosseous dysplasia	Autosomal recessive
5	SMN1	600354	Spinal muscular atrophy	Autosomal recessive
11	SMPD1	607608	Niemann-Pick disease, type A; Niemann-Pick disease, type B	Autosomal recessive
15	SPG11	610844	Amyotrophic lateral sclerosis 5, juvenile; Charcot-Marie-Tooth disease, axonal, type 2X; Spastic paraplegia 11	Autosomal recessive

16	SPG7	602783	Spastic paraplegia, type 7, autosomal recessive	Autosomal recessive
2	SRD5A2	607306	46,XY disorder of sex development due to 5-alpha-reductase 2 deficiency (pseudovaginal perineoscrotal hypospadias)	Autosomal recessive
2	ST3GAL5	604402	Salt and pepper developmental regression syndrome	Autosomal recessive
8	STAR	600617	Lipoid adrenal hyperplasia	Autosomal recessive
3	SUMF1	607939	Multiple sulfatase deficiency	Autosomal recessive
9	SURF1	185620	Mitochondrial complex IV deficiency, nuclear type 1; Charcot-Marie-Tooth disease, type 4K	Autosomal recessive
X	SYN1	313440	Epilepsy, X-linked, with variable learning disabilities and behavior disorders	X-linked
16	TAT	613018	Tyrosinemia, type 2	Autosomal recessive
11	TCIRG1	604592	Osteopetrosis, autosomal recessive, type 1	Autosomal recessive
14	TECPR2	615000	Spastic paraplegia, type 49, autosomal recessive	Autosomal recessive
3	TF	190000	Atransferrinemia	Autosomal recessive
7	TFR2	604720	Hemochromatosis, type 3	Autosomal recessive
8	TG	188450	Thyroid dyshormonogenesis, type 3	Autosomal recessive
14	TGM1	190195	Ichthyosis, congenital, autosomal recessive, type 1	Autosomal recessive
11	TH	191290	Segawa syndrome, recessive	Autosomal recessive
X	THOC2	300395	Mental retardation, X-linked 12	X-linked
11	TMEM216	613277	Joubert syndrome, type 2; Meckel syndrome, type 2	Autosomal recessive
8	TMEM67	609884	Meckel syndrome 3; COACH syndrome 1; Joubert syndrome 6; Nephronophthisis 11	Autosomal recessive
21	TMPRSS3	605511	Deafness, autosomal recessive, type 8/10	Autosomal recessive
6	TNXB	600985	Ehlers-Danlos syndrome, classic-like	Autosomal recessive
2	TPO	606765	Thyroid dyshormonogenesis, type 2A	Autosomal recessive
11	TPP1	607998	Ceroid lipofuscinosis, neuronal, type 2; Spinocerebellar ataxia, autosomal recessive, type 7	Autosomal recessive
6	TRDN	603283	Ventricular tachycardia, catecholaminergic polymorphic, type 5, with or without muscle weakness	Autosomal recessive
3	TREX1	606609	Aicardi-Goutieres syndrome, type 1	Autosomal recessive
9	TRIM32	602290	Limb-girdle muscular dystrophy, type 8 (LGMD R8)	Autosomal recessive
17	TRIM37	605073	Mulibrey nanism	Autosomal recessive
22	TRMU	610230	Liver failure, transient infantile	Autosomal recessive
17	TSEN54	608755	Pontocerebellar hypoplasia, type 2A; Pontocerebellar hypoplasia, type 4	Autosomal recessive
12	TSFM	604723	Combined oxidative phosphorylation deficiency, type 3	Autosomal recessive
1	TSHB	188540	Hypothyroidism, congenital, nongoitrous, type 4	Autosomal recessive
14	TSHR	603372	Hypothyroidism, congenital, nongoitrous, type 1	Autosomal recessive
8	TTPA	600415	Ataxia with isolated vitamin E deficiency	Autosomal recessive
22	TYMP	131222	Mitochondrial DNA depletion syndrome, type 1 (MNGIE type)	Autosomal recessive
11	TYR	606933	Oculocutaneous albinism (OCA) type 1A; OCA type 1B	Autosomal recessive
9	TYRP1	115501	Albinism, oculocutaneous, type 3	Autosomal recessive
15	UBE3A	601623	Angelman syndrome	Autosomal dominant*
2	UGT1A1	191740	Crigler-Najjar syndrome, type 1; Crigler-Najjar syndrome, type 2	Autosomal recessive
17	UNC13D	608897	Hemophagocytic lymphohistiocytosis, familial, type 3	Autosomal recessive
X	UPF3B	300298	Mental retardation, X-linked, syndromic, type 14	X-linked
11	USH1C	605242	Usher syndrome, type 1C; Deafness, autosomal recessive, type 18A	Autosomal recessive
17	USH1G	607696	Usher syndrome, type 1G	Autosomal recessive
1	USH2A	608400	Usher syndrome, type 2A; Retinitis pigmentosa 39	Autosomal recessive
9	VPS13A	605978	Choreoacanthocytosis	Autosomal recessive
8	VPS13B	607817	Cohen syndrome	Autosomal recessive
1	VPS45	610035	Neutropenia, severe congenital, type 5	Autosomal recessive
17	VPS53	615850	Pontocerebellar hypoplasia, type 2E	Autosomal recessive
14	VRK1	602168	Pontocerebellar hypoplasia, type 1A	Autosomal recessive
14	VSX2	142993	Microphthalmia with coloboma 3; Isolated microphthalmia 2	Autosomal recessive
X	WAS	300392	Wiskott-Aldrich syndrome; Thrombocytopenia, X-linked	X-linked

9	WHRN	607928	Usher syndrome, type 2D; Deafness, autosomal recessive, type 31	Autosomal recessive
2	WNT10A	606268	WNT10A-related conditions	Autosomal recessive
8	WRN	604611	Werner syndrome	Autosomal recessive
9	XPA	611153	Xeroderma pigmentosum, group A	Autosomal recessive
3	XPC	613208	Xeroderma pigmentosum, group C	Autosomal recessive
X	ZDHHC9	300646	Mental retardation, X-linked syndromic, Raymond type	X-linked
14	ZFYVE26	612012	Spastic paraplegia, type 15, autosomal recessive	Autosomal recessive
X	ZNF711	314990	Mental retardation, X-linked, type 97	X-linked