3-Hydroxy-3-methylglutaryl-CoA lyase deficiency	HMGCL	AR
3-Ketothiolase deficiency	ACAT 1	AR
3-Methylcrotonyl-CoAcarboxylase 1 deficiency (3-MCC deficiency)	MCCC1	AR
3-Methylcrotonyl-CoAcarboxylase 2 deficiency (3-MCC deficiency)	MCCC2	AR
Abetalipoproteinemia	MTTP	AR
Achondrogenesis, type IB; Atelosteogenesis II; Diastrophic dysplasia; Multiple epiphyseal dysplasia	SLC26 A2	AR
Achromatopsia	CNGB3	AR
Acrodermatitis enteropathica	SLC 39A4	AR
Acyl-CoA dehydrogenase-9 (ACAD9) Deficiency	ACAD9	AR
Adenosine deaminase deficiency	ADA	AR
Adrenal insufficiency, congenital, with 46,XY sex reversal, partial or complete	CYP 11 A1	AR
Adrenoleukodystrophy, X-linked	ABCD1	XL
Aicardi-Goutieres syndrome	SAMHD1	AR
Aicardi-Goutieres syndrome 2	RNASEH2B	AR
Albinism, oculocutaneous, type IA; Albinism, oculocutaneous, type IB	TYR	AR
Albinism, oculocutaneous, type II; Albinism, brown oculocutaneous; Skin/hair/eye pigmentation 1	OCA2	AR
Alkaptonuria	HGD	AR
Allan-Herndon-Dudley syndrome	SLC16 A2	XL
Alpha thalassemia	HBA1	AR
Alpha thalassemia	HBA2	AR
Alpha thalassemia X-linked intellectual disability syndrome	ATRX	XL
Alpha-1 antitrypsin deficiency	SERPINA1	AR
Alpha-mannosidosis	MAN2B1	AR
Alport syndrome, COL4A3- related	COL 4 A3	AR
Alport syndrome, COL4A4- related	COL 4 A4	AR
Alport syndrome, COL4A5- related	COL 4 A5	XL
Alstrom syndrome	ALMS1	AR
Anauxetic dysplasia; Cartilage-hair hypoplasia; Metaphyseal dysplasia without hypotrichosis	RMRP	AR
Andermann syndrome	SLC12 A6	AR
Arginase deficiency	ARG1	AR
Argininosuccinate lyase deficiency	ASL	AR
Aromatase deficiency	CYP19A1	AR
Arthrogryposis, mental retardation, and seizures	SLC35A3	AR
Arts syndrome; Rosenberg- Chutorian syndrome; Phosphoribosylpyrophosphate synthetase superactivity; Non- syndromic hearing loss, PRPS1- r	PRPS1	XL
Asparagine synthetase deficiency	ASNS	AR
Aspartylglucosaminuria	AGA	AR
Ataxia with isolated vitamin E deficiency	TTPA	AR
Ataxia-telangiectasia	ATM	AR
Atransferrinemia	TF	AR
Autoimmune polyendocrinopathy syndrome type I	AIRE	AR
Autosomal Recessive Spastic Ataxia of Charlevoix-Saguenay	SACS	AR
Bardet-Biedl syndrome 14; Joubert syndrome 5; Leber congenital amaurosis 10; Meckel syndrome 4; Senior-Løken syndrome 6	CEP290	AR
Bardet-Biedl syndrome 2; Retinitis Pigmentosa 74	BBS2	AR
Bardet-Biedl syndrome type 1	BBS1	AR
Bardet-Biedl syndrome type 10	BBS10	AR
Bardet-Biedl syndrome type 12	BBS12	AR
Bare lymphocyte syndrome, type II	CIITA	AR
Bartter syndrome	BSND	AR
Bernard-Soulier syndrome type A1	GP1BA	AR
Bernard-Soulier syndrome type C	GP9	AR
Bilateral frontoparietal polymicrogyria	ADGRG1	AR
Biotinidase deficiency	BTD	AR
Björnstad syndrome; GRACILE syndrome; Mitochondrial complex III deficiency	BCS1L	AR
Bloom syndrome	BLM	AR
Butyrlcholinesterase deficiency	BCHE	AR
Canavan disease	ASPA	AR
Carbamoylphosphate synthetase I deficiency	CPS1	AR
Carnitine palmitoyltransferase IA deficiency	CPT1A	AR
Carnitine palmitoyltransferase II deficiency	CPT2	AR
Carnitine-acylcarnitine translocase deficiency	SLC25A20	AR
Carpenter syndrome	RAB23	AR
Catecholaminergic polymorphic ventricular tachycardia, CASQ2- related	CASQ2	AR
Catecholaminergic polymorphic ventricular tachycardia, TRDN- related	TRDN	AR
Cerebrotendinous xanthomatosis	CYP27A1	AR
Charcot-Marie-Tooth disease, GDAP1-related	GDAP1	AR
Charcot-Marie-Tooth disease, SH3TC2-related	SH3TC2	AR
Charcot-Marie-Tooth disease, SURF1-related	SURF1	AR
Charcot-Marie-Tooth disease, type 4B1	MTMR2	AR
Charcot-Marie-Tooth disease, type 4D	NDRG1	AR
Charcot-Marie-Tooth disease, X-linked type 1	GJB1	XL
Chediak-Higashi syndrome	LYST	AR
Childhood-onset severe retinal dystrophy, AIPL1-related	AIPL1	AR
Chondrodysplasia punctata type 1, X-linked	ARSE	XL
Choreoacanthocytosis	VPS13A	AR
Choroideremia	CHM	XL
Chronic granulomatous disease	CYBA	AR
Chronic granulomatous disease, X-linked	CYBB	XL
Citrin deficiency	SLC25A13	AR
Citrullinemia	ASS1	AR
Cockayne syndrome type A	ERCC8	AR
Cockayne syndrome type B;De Sanctis-Cacchione syndrome	ERCC6	AR

Cohen syndrome	VPS13B	AR
Combined malonic and methylmalonic aciduria	ACSF3	AR
Combined oxidative phosphorylation deficiency, GFM1-related	GFM1	AR
Combined oxidative phosphorylation deficiency, TSFM-related	TSFM	AR
Combined pituitary hormone deficiency 2	PROP1	AR
Combined pituitary hormone deficiency 3	LHX3	AR
Congenital adrenal hyperplasia due to 11-beta-hydroxylase deficiency	CYP11B1	AR
Congenital adrenal hyperplasia due to 17-alpha-hydroxylase deficiency	CYP17A1	AR
Congenital adrenal hyperplasia due to 21-hydroxylase deficiency	CYP21A2	AR
Congenital adrenal hyperplasia due to 3-beta-hydroxysteroid dehydrogenase 2 deficiency	HSD3B2	AR
Congenital adrenal hypoplasia, X-linked	NR0B1	XL
Congenital amegakaryocytic thrombocytopenia	MPL	AR
Congenital disorder of glycosylation type 1a	PMM2	AR
Congenital disorder of glycosylation type Ib	MPI	AR
Congenital disorder of glycosylation type Ic	ALG6	AR
Congenital hydrocephalus 1	CCDC88C	AR
Congenital hyperinsulinism; Permanent neonatal diabetes mellitus	KCNJ11	AR
Congenital hypothyroidism, TSHB-related	TSHB	AR
Congenital ichthyosis	TGM1	AR
Congenital insensitivity to pain with anhidrosis	NTRK1	AR
Congenital myasthenic syndrome, CHRNE-related	CHRNE	AR
Congenital myasthenic syndrome, RAPSN-related; Fetal akinesia deformation sequence	RAPSN	AR
Congenital nephrotic syndrome, type 1	NPHS1	AR
Congenital nephrotic syndrome, type 2	NPHS2	AR
Congenital secretory chloride diarrhea	SLC26A3	AR
Congenital hypothyroidism, DUOX2-related	DUOX2	AR
Congenital hypothyroidism, DUOXA2-related	DUOXA2	AR
Corneal endothelial dystrophy	SLC4A11	AR
Corticosterone methyloxidase deficiency	CYP11B2	AR
Costeff syndrome	OPA3	AR
Creatine deficiency syndrome	SLC6A8	XL
Crigler-Najjar syndrome	UGT1A1	AR
Cystic fibrosis	CFTR	AR
Cystinosis	CTNS	AR
D-bifunctional protein deficiency	HSD17B4	AR
Dent disease 2; Lowe syndrome	OCRL	XL
Dihydrolipoamide dehydrogenase deficiency	DLD	AR
Dihydropyrimidine dehydrogenase deficiency	DPYD	AR
Donnai-Barrow syndrome; Facioоculoacousticorenal syndrome	LRP2	AR
Duchenne muscular dystrophy	DMD	XL
Dyskeratosis congenita type 5	RTEL1	AR
Dystrophic epidermolysis bullosa	COL7A1	AR
Ehlers-Danlos syndrome with kyphoscoliosis, PLOD1-related	PLOD1	AR
Ehlers-Danlos syndrome, autosomal recessive, due to tenascin X deficiency	TNXB	AR
Ehlers-Danlos syndrome, Dermatosparaxis type VIIC	ADAMTS2	AR
Ellis-van Creveld syndrome, EVC-related; Weyers acrofacial dysostosis, EVC-related	EVC	AR
Ellis-van Creveld syndrome, EVC2-related; Weyers acrodental dysostosis, EVC2-related	EVC2	AR
Emery-Dreifuss muscular dystrophy	EMD	XL
Enhanced S-cone syndrome; Retinitis pigmentosa 37	NR2E3	AR
Ethylmalonic encephalopathy	ETHE1	AR
Fabry disease	GLA	XL
Factor V deficiency	F5	AR
Factor XI deficiency	F11	AR
Familial dysautonomia	ELP1 (IKBKAP)	AR
Familial hypercholesterolemia	LDLRAP1	AR
Familial hyperinsulinism, ABCC8- related	ABCC8	AR
Familial lipoprotein lipase deficiency	LPL	AR
Familial Mediterranean fever	MEFV	AR
Fanconi anemia group A	FANCA	AR
Fanconi anemia group C	FANCC	AR
Fanconi anemia group G	FANCG	AR
Fragile X syndrome	FMR1	XL
Fraser syndrome	GRIP1	AR
Friedreich ataxia	FXN	AR
Fumarase deficiency	FH	AR
Galactokinase deficiency	GALK1	AR
Galactose epimerase deficiency	GALE	AR
Galactosemia	GALT	AR
Gaucher disease	GBA	AR
Gitelman syndrome	SLC12A3	AR
Glucose-6-phosphate dehydrogenase deficiency	G6PD	XL
Glutamate formiminotransferase deficiency	FTCD	AR
Glutaric aciduria IIA	ETFA	AR
Glutaric aciduria IIB	ETFB	AR
Glutaric aciduria IIC	ETFDH	AR
Glutaric aciduria, type I	GCDH	AR
Glycine encephalopathy, AMT- related	AMT	AR
Glycine encephalopathy, GLDC- related	GLDC	AR
Glycogen storage disease IV	GBE1	AR
Glycogen storage disease type III	AGL	AR
Glycogen storage disease type V	PYGM	AR

Glycogen storage disease VII	PFKM	AR
Glycogen Storage disease, type 1a	G6PC	AR
Glycogen storage disease, type Ib	SLC37A4	AR
Guanidinoacetate methyltransferase deficiency	GAMT	AR
Gyrate atrophy of choroid and retina	OAT	AR
Hartnup disorder	SLC6A19	AR
Hemochromatosis, HFE-related	HFE	AR
Hemochromatosis, type 2A	HJV (HFE2)	AR
Hemochromatosis, type 3	TFR2	AR
Hemophagocytic lymphohistiocytosis, familial, 2	PRF1	AR
Hemophilia A	F8	XL
Hemophilia B	F9	XL
Hepatocerebral mitochondrial DNA depletion syndrome, MPV17-related	MPV17	AR
Hereditary folate malabsorption	SLC46A1	AR
Hereditary fructose intolerance	ALDOB	AR
Hermansky-Pudlak syndrome 1	HPS1	AR
Hermansky-Pudlak syndrome 3	HPS3	AR
Holocarboxylase synthetase deficiency	HLCS	AR
Homocystinuria due to cystathionine beta-synthase deficiency	CBS	AR
Homocystinuria-megaloblastic anemia, cobalamin E type	MTRR	AR
Homocystinuria, MTHFR-related	MTHFR	AR
Hydrolethalus syndrome	HYLS1	AR
Hyper IgM syndrome, X-linked	CD40LG	XL
Hyperimmunoglobulinemia D syndrome	MVK	AR
Hypermethioninemia due to adenosine kinase deficiency	ADK	AR
Hypermethioninemia due to deficiency ofS-adenosylhomocysteine hydrolase	AHCY	AR
Hyperornithinemia- hyperammonemia- homocitrullinemia syndrome (Triple H syndrome)	SLC25A15	AR
Hyperprolinemia type II	ALDH4A1	AR
Hypogonadotropic hypogonadism, GNRHR-related	GNRHR	AR
Hypohidrotic ectodermal dysplasia	EDA	XL
Hypophosphatasia	ALPL	AR
Inclusion body myopathy type 2 (Nonaka myopathy)	GNE	AR
Infantile neuroaxonal dystrophy	PLA2G6	AR
Isovaleric acidemia	IVD	AR
Joubert syndrome 2; Meckel syndrome 2	TMEM216	AR
Joubert syndrome 28; Meckel syndrome 1; Bardet-Biedlsyndrome 13	MKS1	AR
Joubert syndrome 4; Senior-Løken syndrome 1; Nephronophthisis	NPHP1	AR
Joubert syndrome 9; Meckel syndrome 6; COACH syndrome	CC2D2A	AR
Joubert syndrome, AHI1-related	AHI1	AR
Joubert syndrome, ARL13B- related	ARL13B	AR
Junctional epidermolysis bullosa,LAMA3-related; Laryngo-onycho- cutaneous syndrome	LAMA3	AR
Junctional epidermolysis bullosa, LAMB3-related	LAMB3	AR
Junctional epidermolysis bullosa, LAMC2-related	LAMC2	AR
Juvenile retinoschisis, X-linked	RS1	XL
Krabbe disease	GALC	AR
L1 syndrome	L1CAM	XL
Leber congenital amaurosis 2; Retinitis pigmentosa 20	RPE65	AR
Leber congenital amaurosis 5	LCA5	AR
Leber congenital amaurosis 8; Retinitis pigmentosa 12	CRB1	AR
Leber congenital amaurosis type 13	RDH12	AR
Leigh syndrome with Complex IV deficiency	LRPPRC	AR
Lethal congenital contracture syndrome 1	GLE1	AR
Leukoencephalopathy with vanishing white matter	EIF2B5	AR
Limb-girdle muscular dystrophy type 2A	CAPN3	AR
Limb-girdle muscular dystrophy type 2B	DYSF	AR
Limb-girdle muscular dystrophy, type 2C	SGCG	AR
Limb-girdle muscular dystrophy, type 2D	SGCA	AR
Limb-girdle muscular dystrophy, type 2E	SGCB	AR
Limb-girdle muscular dystrophy, type 2F	SGCD	AR
Limb-girdle muscular dystrophy, type 2H; Bardet-Biedl syndrome 11	TRIM32	AR
Lipoid congenital adrenal hyperplasia	STAR	AR
Lissencephaly, X-linked	DCX	XL
Liver failure, acute infantile	TRMU	AR
Long-chain 3-hydroxyacyl- CoA dehydrogenase (LCHAD) deficiency; Trifunctional protein deficiency	HADHA	AR
Lujan-Fryns syndrome, UPF3B- related	UPF3B	XL
Lujan-Fryns syndrome, ZDHHC9- related	ZDHHC9	XL
Lysinuric protein intolerance	SLC7A7	AR
Lysosomal acid lipase deficiency	LIPA	AR
Macular corneal dystrophy, CHST6-related	CHST6	AR
Maple syrup urine disease type Ia	BCKDHA	AR
Maple syrup urine disease type Ib	BCKDHB	AR
Maple syrup urine disease, type II	DBT	AR
Meckel syndrome 5; Joubert syndrome 7; COACH syndrome	RPGRIP1L	AR
Medium-chain acyl-CoA dehydrogenase (MCAD) deficiency	ACADM	AR
Megalencephalic leukoencephalopathy with subcortical cysts	MLC1	AR
Menkes disease	ATP7A	XL
Mental retardation, X-linked, associated with fragile site FRAXE	AFF2	XL
Metachromatic leukodystrophy	ARSA	AR
Metachromatic leukodystrophy due to saposin-b deficiency	PSAP	AR
Methylmalonic acidemia, MUT- related	MUT	AR
Methylmalonic aciduria and homocystinuria, cblC type	MMACHC	AR

Methylmalonic aciduria and homocystinuria, cblD type	MMADHC	AR
Methylmalonic aciduria and homocystinuria, cblF type	LMBRD1	AR
Methylmalonic aciduria and homocystinuria, cblJ type	ABCD4	AR
Methylmalonic aciduria, cblA type	MMAA	AR
Methylmalonic aciduria, cblB type	MMAB	AR
Methylmalonyl-CoA epimerase deficiency	MCEE	AR
Microcephaly, primary autosomal recessive, 1	MCPH1	AR
Microphthalmia with or without coloboma	VSX2	AR
Microphthalmia, isolated 3	RAX	AR
Mitochondrial complex I deficiency (Leigh syndrome), NDUFAF5-related	NDUFAF5	AR
Mitochondrial complex I deficiency (Leigh syndrome), NDUFS6-related	NDUFS6	AR
Mitochondrial complex IV deficiency, nuclear type 2	SCO2	AR
Mitochondrial myopathy and sideroblastic anemia 1	PUS1	AR
Mitochondrial neurogastrointestinal encephalopathy (MNGIE) disease	TYMP	AR
Mucolipidosis III alpha/beta; Mucolipidosis II alpha/beta	GNPTAB	AR
Mucolipidosis III gamma	GNPTG	AR
Mucolipidosis IV	MCOLN1	AR
Mucopolysaccharidosis IIIA (Sanfilippo syndrome A)	SGSH	AR
Mucopolysaccharidosis IIID (Sanfilippo syndrome D)	GNS	AR
Mucopolysaccharidosis IVA (Morquio syndrome A)	GALNS	AR
Mucopolysaccharidosis type II (Hunter syndrome)	IDS	XL
Mucopolysaccharidosis type IIIB (Sanfilippo syndrome B)	NAGLU	AR
Mucopolysaccharidosis type IIIC (Sanfilippo syndrome C)	HGSNAT	AR
Mucopolysaccharidosis type IVB (Morquio syndrome B); GM1- gangliosidosis	GLB1	AR
Mucopolysaccharidosis type IX	HYAL1	AR
Mucopolysaccharidosis type VI (Maroteaux-Lamy syndrome)	ARSB	AR
Mucopolysaccharidosis type VII	GUSB	AR
Mucopolysaccharidosis, type I (Hurler syndrome)	IDUA	AR
Multiple pterygium syndrome	CHRNG	AR
Multiple sulfatase deficiency	SUMF1	AR
Muscular dystrophy- dystroglycanopathy, FKRP-related	FKRP	AR
Muscular dystrophy- dystroglycanopathy, FKTN- related; Fukuyama congenital muscular dystrophy	FKTN	AR
Muscular dystrophy- dystroglycanopathy, POMT1- related	POMT1	AR
Muscular dystrophy- dystroglycanopathy, POMT2- related	POMT2	AR
Muscular dystrophy- dystroglycanopathy; Retinitis pigmentosa 76	POMGNT1	AR
Muscular dystrophy, LAMA2- related	LAMA2	AR
Myotonia congenita, autosomal dominant; Myotonia congenita, autosomal recessive, Myotonia levior	CLCN1	AR
Myotubular myopathy, X-linked	MTM1	XL
N-acetylglutamate synthase deficiency	NAGS	AR
Nemaline myopathy	NEB	AR
Nephrogenic diabetes insipidus	AQP2	AR
Neuronal ceroid lipofuscinosis, CLN3-related	CLN3	AR
Neuronal ceroid lipofuscinosis, CLN5-related	CLN5	AR
Neuronal ceroid lipofuscinosis, CLN6-related	CLN6	AR
Neuronal ceroid lipofuscinosis, CLN8-related	CLN8	AR
Neuronal ceroid lipofuscinosis, MFSD8-related	MFSD8	AR
Neuronal ceroid lipofuscinosis, PPT1-related	PPT1	AR
Neuronal ceroid lipofuscinosis, TPP1-related	TPP1	AR
Niemann-Pick disease, type A/B	SMPD1	AR
Niemann-Pick disease, type C1	NPC1	AR
Niemann-Pick disease, type C2	NPC2	AR
Nijmegen breakage syndrome	NBN	AR
Non-syndromic hearing loss, MYO7A-related; Usher syndrome, type 1B	MYO7A	AR
Non-syndromic hearing loss, PCDH15-related; Usher syndrome, type 1F	PCDH15	AR/Digenic
Non-syndromic hearing loss, USH1C-related; Usher syndrome, type IC	USH1C	AR
Nonsyndromic hearing loss, GJB2- related	GJB2	AR
Nonsyndromic hearing loss, GJB6- related	GJB6	AR
Nonsyndromic hearing loss, GJB6- related	CRYL1	AR
Nonsyndromic hearing loss, LOXHD1-related	LOXHD1	AR
Nonsyndromic hearing loss, OTOF-related	OTOF	AR
Norrie disease	NDP	XL
Omenn syndrome, RAG1-related	RAG1	AR
Omenn syndrome, RAG2-related	RAG2	AR
Opitz GBBB syndrome, type I	MID1	XL
Ornithine transcarbamylase deficiency	OTC	XL
Osteogenesis imperfecta, type VIII	P3H1	AR
Osteopetrosis, TCIRG1-related	TCIRG1	AR
Pantothenate kinase-associated neurodegeneration	PANK2	AR
Pendred syndrome	SLC26A4	AR
Peroxisomal acyl-CoA oxidase deficiency	ACOX1	AR
Phenylalanine hydroxylase deficiency (Phenylketonuria)	PAH	AR
Phosphoglycerate dehydrogenase deficiency	PHGDH	AR
Phosphoglycerate kinase 1 deficiency	PGK1	AR
Polycystic kidney disease, PKHD1- related	PKHD1	AR
Pompe disease	GAA	AR
Pontocerebellar hypoplasia type 1A	VRK1	AR
Pontocerebellar hypoplasia type 1B	EXOSC3	AR
Pontocerebellar hypoplasia type 6	RARS2	AR
Pontocerebellar hypoplasia, type 2D	SEPSECS	AR
Postnatal progressive microcephaly with seizures and brain atrophy	MED17	AR
Primary ciliary dyskinesia, DNAH5-related	DNAH5	AR

Primary ciliary dyskinesia, DNAI1- related	DNAI1	AR
Primary ciliary dyskinesia, DNAI2- related	DNAI2	AR
Primary ciliary dyskinesia, DNAL1- related	DNAL1	AR
Primary ciliary dyskinesia, type 14	CCDC39	AR
Primary ciliary dyskinesia, type 17	CCDC103	AR
Primary ciliary dyskinesia, type 30	CCDC151	AR
Primary congenital glaucoma	CYP1B1	AR
Primary hyperoxaluria type 1	AGXT	AR
Primary Hyperoxaluria type II	GRHPR	AR
Primary hyperoxaluria type III	HOGA1	AR
Progressive external ophthalmoplegia; Alpers- Huttenlocher syndrome; Ataxia neuropathy spectrum; Myocerebrohepatopathy syndrome	POLG	AR
Progressive Familial Intrahepatic Cholestasis	ABCB11	AR
Propionic acidemia, PCCA-related	PCCA	AR
Propionic acidemia, PCCB-related	PCCB	AR
Prothrombin-related conditions	F2	AR
Pycnodysostosis	CTSK	AR
Pyruvate carboxylase deficiency	PC	AR
Pyruvate dehydrogenase E1-alpha deficiency	PDHA1	XL
Pyruvate dehydrogenase E1-beta deficiency	PDHB	AR
Renal tubular acidosis with deafness	ATP6V1B1	AR
Renpenning syndrome	PQBP1	XL
Retinitis pigmentosa 25	EYS	AR
Retinitis pigmentosa 26	CERKL	AR
Retinitis pigmentosa 28	FAM161A	AR
Retinitis pigmentosa 59	DHDDS	AR
Retinitis Pigmentosa, CNGA1- related	CNGA1	AR
Retinitis Pigmentosa, CNGB1- related	CNGB1	AR
Retinitis pigmentosa, IDH3B- related	IDH3B	AR
Retinitis pigmentosa, PDE6A- related	PDE6A	AR
Rhizomelic chondrodysplasia punctata, type 1	PEX7	AR
Rhizomelic chondrodysplasia punctata, type 3	AGPS	AR
Roberts syndrome	ESCO2	AR
Sandhoff disease	HEXB	AR
Schimke immunoosseous dysplasia	SMARCAL1	AR
Schindler disease type I; Schindler disease type III	NAGA	AR
Schopf-Schulz-Passarge syndrome; Odontoonychodermal dysplasia	WNT10A	AR
Segawa syndrome	TH	AR
Severe combined immunodeficiency with sensitivity to ionizing radiation	DCLRE1C	AR
Severe combined immunodeficiency, JAK3-related	JAK3	AR
Severe combined immunodeficiency, X-linked	IL2RG	XL
Severe Congenital Neutropenia, HAX1-related	HAX1	AR
Severe congenital neutropenia, VPS45-related	VPS45	AR
Short branched chain acyl- CoA dehydrogenase (SBCAD) deficiency	ACADSB	AR
Short-chain acyl-coA dehydrogenase (SCAD) Deficiency	ACADS	AR
Short-rib thoracic dysplasia 3 with or without polydactyly	DYNC2H1	AR
Sialic acid storage disorder	SLC17A5	AR
Sickle cell disease; Beta thalassemia	HBB	AR
Sjögren-Larsson syndrome	ALDH3A2	AR
Smith-Lemli-Opitz syndrome	DHCR7	AR
Spastic paraplegia 15	ZFYVE26	AR
Spastic paraplegia 2, X-linked (SPG2)	PLP1	XL
Spastic paraplegia 49	TECPR2	AR
Spastic paraplegia type 7	SPG7	AR
SPG11-related Neuromuscular Disorders	SPG11	AR
Spinal muscular atrophy	SMN1	AR
Spinocerebellar ataxia, autosomal recessive 10	ANO10	AR
Spondylocostal dysostosis	MESP2	AR
Steel syndrome	COL27A1	AR
Stuve-Wiedemann syndrome	LIFR	AR
Surfactant metabolism dysfunction, pulmonary, 3; Interstitial lung disease	ABCA3	AR
Systemic primary carnitine deficiency	SLC22A5	AR
Tay-Sachs disease	HEXA	AR
Tetrahydrobiopterin deficiency	PTS	AR
Tetrahydrobiopterin deficiency, PCBD1-related	PCBD1	AR
Tetrahydrobiopterin deficiency, QDPR-related	QDPR	AR
Thiamine metabolism dysfunction syndrome 2 (biotin or thiamine responsive)	SLC19A3	AR
Thyroid dyshormonogenesis, IYD-related	IYD	AR
Thyroid dyshormonogenesis, SLC5A5-related	SLC5A5	AR
Thyroid dyshormonogenesis, TG-related	TG	AR
Thyroid dyshormonogenesis, TPO-related	TPO	AR
Treacher Collins syndrome, POLR1C-related	POLR1C	AR
Trichohepatoenteric syndrome	TTC37	AR
Trichothiodystrophy 1, photosensitive; Xeroderma pigmentosum, group D	ERCC2	AR
Trimethylaminuria	FMO3	AR
Tyrosinemia, type 1	FAH	AR
Tyrosinemia, type II	TAT	AR
Usher syndrome type 2D	WHRN	AR
Usher syndrome type IG	USH1G	AR
Usher syndrome, type 1D	CDH23	AR/Digenic
Usher syndrome, type 2A	USH2A	AR
Usher syndrome, type 3A	CLRN1	AR

Very long-chain acyl-CoA dehydrogenase (VLCAD) deficiency	ACADVL	AR
Vitamin D-dependent rickets, type 1A	CYP27B1	AR
Wilson disease	ATP7B	AR
Wiskott-Aldrich syndrome; Thrombocytopenia, X-linked; Severe Congenital Neutropenia, WAS-related	WAS	XL
Wolcott-Rallison syndrome	EIF2AK3	AR
X-linked Aarskog-Scott syndrome	FGD1	XL
X-linked epilepsy with variable learning disabilities	SYN1	XL
X-linked hearing loss, POU3F4- related	POU3F4	XL
X-linked intellectual disability with cerebellar hypoplasia and distinctive facial appearance	OPHN1	XL
X-linked Intellectual disability, AP1S2-related	AP1S2	XL
X-linked intellectual disability, ARX-related	ARX	XL
X-linked intellectual disability, BRWD3-related	BRWD3	XL
X-linked intellectual disability, CUL4B-related	CUL4B	XL
X-linked intellectual disability, DLG3-related	DLG3	XL
X-linked intellectual disability, FTSJ1-related	FTSJ1	XL
X-linked intellectual disability, IL1RAPL1-related	IL1RAPL1	XL
X-linked intellectual disability, KDM5C-related	KDM5C	XL
X-linked intellectual disability, PAK3-related	PAK3	XL
X-linked intellectual disability, Siderius type	PHF8	XL
X-linked Intellectual disability, THOC2-related	THOC2	XL
X-linked intellectual disability, ZNF711-related	ZNF711	XL
X-linked Ocular albinism, GPR143- related	GPR143	XL
X-linked Retinitis pigmentosa, RP2-related	RP2	XL
X-linked Retinitis pigmentosa, RPGR-related	RPGR	XL
Xeroderma pigmentosum, group A	XPA	AR
Xeroderma pigmentosum, group C	XPC	AR
Zellweger syndrome, PEX1-related	PEX1	AR
Zellweger syndrome, PEX10-related	PEX10	AR
Zellweger syndrome, PEX12-related	PEX12	AR
Zellweger syndrome, PEX2-related	PEX2	AR
Zellweger syndrome, PEX6-related	PEX6	AR