virtusGENETICS
A MEMBER OF VIRTUS HEALTH

Individual Expanded Carrier Screen (Fulgent)

CLICK HERE TO VIEW
270 Genes Excluding X-linked Genes
700+ Genes
700+ Genes Excluding X-linked Genes

Virtus 342 Genes

GENE	CONDITION	INHERITANCE
ABCB11	Progressive familial intrahepatic cholestasis	AR
ABCC8	Familial hyperinsulinism	AR
ABCD1	Adrenoleukodystrophy, X-linked	XL
ACAD9	Acyl-CoA dehydrogenase-9 (ACAD9) deficiency	AR
ACADM	Medium-chain acyl-CoA dehydrogenase (MCAD) deficiency	AR
ACADVL	Very long-chain acyl-CoA dehydrogenase (VLCAD) deficiency	AR
ACAT1	3-ketothiolase deficiency	AR
ACOX1	Peroxisomal acyl-CoA oxidase deficiency	AR
ACSF3	Combined malonic and methylmalonic aciduria	AR
ADA	Adenosine deaminase deficiency	AR
ADAMTS2	Ehlers-Danlos syndrome, dermatosparaxis type	AR
ADGRG1	Bilateral frontoparietal polymicrogyria	AR
AFF2	Fragile XE syndrome	XL
AGA	Aspartylglucosaminuria	AR
AGL	Glycogen storage disease type III	AR
AGPS	Rhizomelic chondrodysplasia punctata, type 3	AR
AGXT	Primary hyperoxaluria type 1	AR
AIRE	Autoimmune polyendocrinopathy syndrome type I	AR
ALDH3A2	Sjogren-Larsson syndrome	AR
ALDOB	Hereditary fructose intolerance	AR
ALG6	Congenital disorder of glycosylation type Ic	AR
ALMS1	Alstrom syndrome	AR
ALPL	Hypophosphatasia	AR
AMT	Glycine encephalopathy	AR
AP1S2	X-linked Intellectual disability, AP1S2-related	XL
AQP2	Nephrogenic diabetes insipidus	AR
AR	Androgen insensitivity syndrome	XL

AR: autosomal recessive XL: X-linked

GENE	CONDITION	INHERITANCE
ARG1	Arginase deficiency	AR
ARSA	Metachromatic leukodystrophy	AR
ARSB	Mucopolysaccharidosis type VI (Maroteaux-Lamy syndrome)	AR
ARSE	Chondrodysplasia punctata type 1, X-linked	XL
ARX	X-linked intellectual disability, ARX-related	XL
ASL	Argininosuccinate lyase deficiency	AR
ASNS	Asparagine synthetase deficiency	AR
ASPA	Canavan disease	AR
ASS1	Citrullinemia	AR
ATM	Ataxia-telangiectasia	AR
ATP6V1B1	Renal tubular acidosis with deafness	AR
ATP7A	Menkes disease	XL
ATP7B	Wilson disease	AR
ATRX	Alpha thalassemia X-linked intellectual disability syndrome	XL
AVPR2	Nephrogenic diabetes insipidus	XL
BBS1	Bardet-Biedl syndrome type 1	AR
BBS10	Bardet-Biedl syndrome type 10	AR
BBS12	Bardet-Biedl syndrome type 12	AR
BBS2	BBS2-related ciliopathies	AR
BCKDHA	Maple syrup urine disease type Ia	AR
BCKDHB	Maple syrup urine disease type Ib	AR
BCS1L	Mitochondrial complex III deficiency	AR
BLM	Bloom syndrome	AR
BRWD3	X-linked intellectual disability, BRWD3-related	XL
BSND	Bartter syndrome	AR
BTK	X-linked agammaglobulinemia	XL
CAPN3	Limb-girdle muscular dystrophy type 2A	AR
CBS	Homocystinuria due to cystathionine beta-synthase deficiency	AR
CD40LG	Hyper IgM syndrome, X-linked	XL
CDH23	Usher syndrome, type 1D	AR
CEP290	CEP290-related Ciliopathies	AR
CERKL	Retinitis pigmentosa 26	AR
CFTR	Cystic Fibrosis	AR
CHM	Choroideremia	XL
CHRNE	Congenital myasthenic syndrome	AR
CIITA	Bare lymphocyte syndrome, type II	AR
CLCN5	Dent disease	XL

AR: autosomal recessive XL: X-linked

GENE	CONDITION	INHERITANCE
CLN3	Neuronal ceroid lipofuscinosis	AR
CLN5	Neuronal ceroid lipofuscinosis 5	AR
CLN6	Neuronal ceroid lipofuscinosis, CLN6-related	AR
CLN8	Neuronal ceroid lipofuscinosis, CLN8-related	AR
CLRN1	Usher syndrome, type 3A	AR
CNGB3	Achromatopsia	AR
COL27A1	Steel syndrome	AR
COL4A3	Alport syndrome, COL4A3-related	AR
COL4A4	Alport syndrome, COL4A4-related	AR
COL4A5	Alport syndrome, COL4A5-related	XL
COL7A1	Dystrophic epidermolysis bullosa	AR
CPS1	Carbamoylphosphate synthetase I deficiency	AR
CPT1A	Carnitine palmitoyltransferase IA deficiency	AR
CPT2	Carnitine palmitoyltransferase II deficiency	AR
CRB1	CRB1-related retinopathy	AR
CRYL1	GJB6-CRYL1 related nonsyndromic hearing loss	AR, Digenic
CTNS	Cystinosis	AR
CTSK	Pycnodysostosis	AR
CUL4B	X-linked intellectual disability, CUL4B-related	XL
CYBA	Chronic granulomatous disease	AR
CYBB	Chronic granulomatous disease, X-linked	XL
CYP11B1	Congenital adrenal hyperplasia due to 11-beta-hydroxylase deficiency	AR
CYP11B2	Corticosterone methyloxidase deficiency	AR
CYP17A1	Congenital adrenal hyperplasia due to 17-alpha-hydroxylase deficiency	AR
CYP19A1	Aromatase deficiency	AR
CYP21A2	Congenital adrenal hyperplasia due to 21-hydroxylase deficiency	AR
CYP27A1	Cerebrotendinous xanthomatosis	AR
DBT	Maple syrup urine disease, type II	AR
DCLRE1C	Severe combined immunodeficiency with sensitivity to ionizing radiation	AR
DCX	Lissencephaly, X-linked	XL
DHCR7	Smith-Lemli-Opitz syndrome	AR
DHDDS	Retinitis pigmentosa 59	AR
DKC1	X-linked dyskeratosis congenita	XL
DLD	Dihydrolipoamide dehydrogenase deficiency	AR
DLG3	X-linked intellectual disability, DLG3-related	XL
DMD	Dystrophinopathies	XL
DNAH5	Primary ciliary dyskinesia, DNAH5-related	AR

AR: autosomal recessive XL: X-linked

GENE	CONDITION	INHERITANCE
DNAI1	Primary ciliary dyskinesia, DNAI1-related	AR
DNAI2	Primary ciliary dyskinesia, DNAI2-related	AR
DYSF	Limb-girdle muscular dystrophy type 2B	AR
EDA	Hypohidrotic ectodermal dysplasia	XL
EIF2B5	Leukoencephalopathy with vanishing white matter	AR
ELP1	Familial Dysautonomia	AR
EMD	Emery-Dreifuss muscular dystrophy	XL
ERCC6	ERCC6-related disorders	AR
ERCC8	Cockayne syndrome type A	AR
ESCO2	Roberts syndrome	AR
ETFA	Glutaric aciduria IIA	AR
ETFDH	Glutaric aciduria IIC	AR
ETHE1	Ethylmalonic encephalopathy	AR
EVC	EVC-related bone growth disorders	AR
EVC2	EVC2-related bone growth disorders	AR
EYS	Retinitis pigmentosa 25	AR
F8	Hemophilia A	XL
F9	Hemophilia B	XL
FAH	Tyrosinemia, type 1	AR
FAM161A	Retinitis pigmentosa 28	AR
FANCA	Fanconi anemia group A	AR
FANCB	Fanconi anemia group B	XL
FANCC	Fanconi anemia group C	AR
FANCG	Fanconi anemia group G	AR
FGD1	X-linked Aarskog-Scott syndrome	XL
FH	Fumarase deficiency	AR
FHL1	FHL1-related neuromuscular disorders	XL
FKRP	FKRP Alpha-dystroglycanopathies	AR
FKTN	FKTN Alpha-dystroglycanopathies	AR
FMR1	Fragile X Syndrome Intermediate Allele	XL
FOXP3	IPEX syndrome	XL
FTSJ1	X-linked intellectual disability, FTSJ1-related	XL
G6PC	Glycogen storage disease, type 1a	AR
GAA	Pompe disease	AR
GALC	Krabbe disease	AR
GALK1	Galactokinase deficiency	AR
GALT	Galactosemia	AR

AR: autosomal recessive XL: X-linked

GENE	CONDITION	INHERITANCE
GAMT	Guanidinoacetate methyltransferase deficiency	AR
GBA	Gaucher disease	AR
GBE1	Glycogen storage disease IV	AR
GCDH	Glutaric aciduria, type I	AR
GFM1	Combined oxidative phosphorylation deficiency, GFM1-related	AR
GJB1	Charcot-Marie-Tooth disease, X-linked type 1	XL
GJB2	Nonsyndromic hearing loss 1A	AR
GJB6	GJB6-CRYL1 related nonsyndromic hearing loss	AR, Digenic
GLA	Fabry disease	XL
GLB1	GLB1-related gangliosidoses	AR
GLDC	Glycine encephalopathy, GLDC-related	AR
GLE1	Lethal congenital contracture syndrome 1	AR
GNE	Inclusion body myopathy type 2 (Nonaka myopathy)	AR
GNPTAB	Mucolipidosis II & III	AR
GNPTG	Mucolipidosis III gamma	AR
GNS	Mucopolysaccharidosis IIID (Sanfilippo syndrome D)	AR
GPR143	X-linked Ocular albinism, GPR143-related	XL
GRHPR	Primary hyperoxaluria type II	AR
HADHA	Trifunctional protein deficiency	AR
HAX1	Severe congenital neutropenia, HAX1-related	AR
HBA1	Alpha thalassemia	AR
HBA2	Alpha thalassemia	AR
HBB	Sickle cell disease	AR
HCFC1	Methylmalonic acidemia with homocystinuria, type cblX	XL
HEXA	Tay-Sachs disease	AR
HEXB	Sandhoff disease	AR
HGSNAT	Mucopolysaccharidosis type IIIC (Sanfilippo syndrome C)	AR
HJV	Hemochromatosis, type 2A	AR
HLCS	Holocarboxylase synthetase deficiency	AR
HMGCL	3-hydroxy-3-methylglutaryl-CoA lyase deficiency	AR
HOGA1	Primary hyperoxaluria type III	AR
HPS1	Hermansky-Pudlak syndrome 1	AR
HPS3	Hermansky-Pudlak syndrome 3	AR
HSD17B10	HSD10 mitochondrial disease	XL
HSD17B4	D-bifunctional protein deficiency	AR
HSD3B2	Congenital adrenal hyperplasia due to 3-beta-hydroxysteroid dehydrogenase 2 deficiency	AR

AR: autosomal recessive XL: X-linked

GENE	CONDITION	INHERITANCE
HYAL1	Mucopolysaccharidosis type IX	AR
HYLS1	Hydrolethalus syndrome	AR
IDS	Mucopolysaccharidosis type II (Hunter syndrome)	XL
IDUA	Mucopolysaccharidosis, type I (Hurler syndrome)	AR
IGSF1	X-linked central hypothyroidism and testicular enlargement	XL
IL1RAPL1	X-linked intellectual disability, IL1RAPL1-related	XL
IL2RG	X-linked severe combined immunodeficiency	XL
IVD	Isovaleric Acidemia	AR
KCNJ11	KCNJ11-related hyperinsulinism	AR
KDM5C	X-linked intellectual disability, KDM5C-related	XL
L1CAM	L1 syndrome	XL
LAMA2	Muscular dystrophy, LAMA2-related	AR
LAMA3	Junctional epidermolysis bullosa 2	AR
LAMB3	Junctional epidermolysis bullosa, LAMB3-related	AR
LAMC2	Junctional epidermolysis bullosa, LAMC2-related	AR
LCA5	Leber congenital amaurosis 5	AR
LDLR	Familial Hypercholesterolemia	AR
LDLRAP1	Familial Hypercholesterolemia	AR
LHX3	Combined pituitary hormone deficiency 3	AR
LIFR	Stuve-Wiedemann syndrome	AR
LIPA	Lysosomal acid lipase deficiency	AR
LOXHD1	Nonsyndromic hearing loss 77	AR
LPL	Familial lipoprotein lipase deficiency	AR
LRPPRC	Leigh syndrome with Complex IV deficiency	AR
MAN2B1	Alpha-Mannosidosis	AR
MCOLN1	Mucolipidosis IV	AR
MED17	Postnatal Progressive Microcephaly with Seizures and Brain Atrophy	AR
MESP2	Spondylocostal dysostosis	AR
MFSD8	Neuronal ceroid lipofuscinosis, MFSD8-related	AR
MID1	Opitz GBBB syndrome, type I	XL
MKS1	MKS1-related ciliopathies	AR
MLC1	Megalencephalic leukoencephalopathy with subcortical cysts	AR
MMAA	Methylmalonic aciduria, cblA type	AR
MMAB	Methylmalonic aciduria, cblB type	AR
MMACHC	Methylmalonic aciduria and homocystinuria, cblC type	AR
MMADHC	Methylmalonic aciduria and homocystinuria, cblD type	AR
MPI	Congenital disorder of glycosylation type Ib	AR

AR: autosomal recessive XL: X-linked

GENE	CONDITION	INHERITANCE
MPL	Congenital amegakaryocytic thrombocytopenia	AR
MPV17	Hepatocerebral mitochondrial DNA depletion syndrome, MPV17-related	AR
MTHFR	Homocystinuria, MTHFR-related	AR
MTM1	Myotubular myopathy, X-linked	XL
MTRR	Homocystinuria-megaloblastic anemia, cobalamin E type	AR
MTTP	Abetalipoproteinemia	AR
MUT	Methylmalonic aciduria, methylmalonyl-CoA mutase deficiency	AR
MYO7A	MYO7A-related disorders	AR
NAGLU	Mucopolysaccharidosis type IIIB (Sanfilippo syndrome B)	AR
NAGS	N-acetylglutamate synthase deficiency	AR
NBN	Nijmegen breakage syndrome	AR
NDP	Norrie disease	XL
NDRG1	Charcot-Marie-Tooth disease, type 4D	AR
NDUFAF5	Mitochondrial complex I deficiency (Leigh syndrome)	AR
NDUFS6	Mitochondrial complex I deficiency (Leigh syndrome)	AR
NEB	Nemaline myopathy	AR
NONO	X-linked intellectual disability syndrome 34	XL
NPC1	Niemann-Pick disease, type C1	AR
NPC2	Niemann-Pick disease, type C2	AR
NPHS1	Congenital nephrotic syndrome, type 1	AR
NPHS2	Congenital nephrotic syndrome, type 2	AR
NR0B1	Congenital adrenal hypoplasia, X-linked	XL
NR2E3	NR2E3-related retinal dystrophies	AR
NTRK1	Congenital insensitivity to pain with anhidrosis	AR
OAT	Gyrate atrophy of choroid and retina	AR
OCRL	OCRL-related disoders	XL
OPA3	Costeff syndrome	AR
OPHN1	X-linked intellectual disability-cerebellar hypoplasia syndrome	XL
OTC	Ornithine transcarbamylase deficiency	XL
PAH	Phenylalanine Hydroxylase deficiency (Phenylketonuria)	AR
PAK3	X-linked intellectual disability, PAK3-related	XL
PC	Pyruvate carboxylase deficiency	AR
PCCA	Propionic acidemia, PCCA-related	AR
PCCB	Propionic acidemia, PCCB-related	AR
PCDH15	PCDH15-related sensory loss	AR
PDHA1	Pyruvate dehydrogenase E1-alpha deficiency	XL
PDHB	Pyruvate dehydrogenase E1-beta deficiency	AR

AR: autosomal recessive XL: X-linked

GENE	CONDITION	INHERITANCE
PEX1	Zellweger syndrome, PEX1-related	AR
PEX10	Zellweger syndrome, PEX10-related	AR
PEX12	Zellweger syndrome, PEX12-related	AR
PEX2	Zellweger syndrome, PEX2-related	AR
PEX6	Zellweger syndrome, PEX6-related	AR
PEX7	Rhizomelic chondrodysplasia punctata, type 1	AR
PFKM	Glycogen storage disease VII	AR
PGK1	Phosphoglycerate kinase 1 deficiency	XL
PHF8	X-linked intellectual disability, Siderius type	XL
PHGDH	Phosphoglycerate dehydrogenase deficiency	AR
PHKA1	Glycogen storage disease type IXd	XL
PHKA2	Glycogen storage disease type IXa	XL
PKHD1	Polycystic kidney disease, PKHD1-related	AR
PLP1	PLP1-related disorders	XL
PMM2	Congenital disorder of glycosylation type 1a	AR
POMGNT1	POMGNT1 Alpha-dystroglycanopathies	AR
POU3F4	X-linked hearing loss, POU3F4-related	XL
PPT1	Neuronal ceroid lipofuscinosis, PPT1-related	AR
PQBP1	Renpenning syndrome	XL
PROP1	Combined pituitary hormone deficiency 2	AR
PRPS1	PRPS1-related disorders	XL
PSAP	Metachromatic leukodystrophy due to saposin-b deficiency	AR
PTS	Tetrahydrobiopterin deficiency	AR
PUS1	Mitochondrial myopathy and sideroblastic anemia 1	AR
PYGM	Glycogen storage disease type V	AR
RAB23	Carpenter syndrome	AR
RAG2	Omenn syndrome, RAG2-related	AR
RAPSN	RAPSN-associated acetylcholine receptor deficiency	AR
RARS2	Pontocerebellar hypoplasia type 6	AR
RDH12	Leber congenital amaurosis type 13	AR
RMRP	Cartilage-Hair Hypoplasia Anauxetic Dysplasia Spectrum Disorder	AR
RP2	X-linked Retinitis pigmentosa, RP2-related	XL
RPE65	RPE65-related retinopathy	AR
RPGR	X-linked Retinitis pigmentosa, RPGR-related	XL
RPGRIP1L	RPGRIP1L-related ciliopathies	AR
RS1	Juvenile retinoschisis, X-linked	XL
RTEL1	Dyskeratosis congenita type 5	AR

AR: autosomal recessive XL: X-linked

GENE	CONDITION	INHERITANCE
SACS	Autosomal recessive spastic ataxia of Charlevoix-Saguenay	AR
SAMHD1	Aicardi-Goutieres syndrome	AR
SEPSECS	Pontocerebellar hypoplasia type 2D	AR
SGCA	Limb-girdle muscular dystrophy, type 2D	AR
SGCB	Limb-girdle muscular dystrophy, type 2E	AR
SGCG	Limb-girdle muscular dystrophy, type 2C	AR
SGSH	Mucopolysaccharidosis IIIA (Sanfilippo syndrome A)	AR
SLC12A3	Gitelman syndrome	AR
SLC12A6	Andermann syndrome	AR
SLC16A2	Allan-Herndon-Dudley syndrome	XL
SLC17A5	Sialic acid storage disorder	AR
SLC22A5	Systemic primary carnitine deficiency	AR
SLC25A13	Citrin deficiency	AR
SLC25A15	Hyperornithinemia-hyperammonemia-homocitrullinemia syndrome (Triple H syndrome)	AR
SLC26A2	SLC26A2-related disorders	AR
SLC26A4	Pendred syndrome	AR
SLC35A3	Arthrogryposis, intellectual disability, and seizures	AR
SLC37A4	Glycogen storage disease, type Ib	AR
SLC39A4	Acrodermatitis enteropathica	AR
SLC4A11	Corneal endothelial dystrophy	AR
SLC6A8	Creatine deficiency syndrome	XL
SLC7A7	Lysinuric protein intolerance	AR
SMARCAL1	Schimke immunoosseous dysplasia	AR
SMN1	Spinal muscular atrophy	AR
SMPD1	Niemann-Pick disease, type A/B	AR
STAR	Lipoid congenital adrenal hyperplasia	AR
SUMF1	Multiple sulfatase deficiency	AR
SYN1	X-linked epilepsy with variable learning disabilities	XL
TAT	Tyrosinemia, type II	AR
TAZ	Barth syndrome	XL
TCIRG1	Osteopetrosis 1	AR
TECPR2	Spastic paraplegia 49	AR
TFR2	Hemochromatosis, type 3	AR
TGM1	Congenital ichthyosis	AR
TH	Segawa syndrome	AR
THOC2	X-linked Intellectual disability, THOC2-related	XL
TMEM216	TMEM216-related ciliopathies	AR

AR: autosomal recessive XL: X-linked

GENE	CONDITION	INHERITANCE
TPP1	Neuronal ceroid lipofuscinosis, TPP1-related	AR
TRMU	Liver failure, acute infantile	AR
TSFM	Combined oxidative phosphorylation deficiency, TSFM-related	AR
TTPA	Ataxia with isolated vitamin E deficiency	AR
TYMP	Mitochondrial neurogastrointestinal encephalopathy (MNGIE) disease	AR
UPF3B	Lujan-Fryns syndrome, UPF3B-related	XL
USH1C	USH1C-related disorders	AR
USH2A	Usher syndrome, type 2A	AR
VPS13A	Choreoacanthocytosis	AR
VPS13B	Cohen syndrome	AR
VPS45	Severe congenital neutropenia, VPS45-related	AR
VRK1	Pontocerebellar hypoplasia type 1A	AR
VSX2	Microphthalmia with or without coloboma	AR
WAS	WAS-related hematopoietic disorder	XL
WNT10A	WNT10A-related ectodermal dysplasias	AR
XPA	Xeroderma pigmentosum, group A	AR
XPC	Xeroderma pigmentosum, group C	AR
ZDHHC9	X-linked intellectual disability, ZDHHC9-related	XL
ZFYVE26	Spastic paraplegia 15	AR
ZNF711	X-linked intellectual disability, ZNF711-related	XL

AR: autosomal recessive XL: X-linked

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700+ Genes Excluding X-linked Genes

Virtus 270 Genes Excluding X-linked Genes

GENE	CONDITION	INHERITANCE
ABCB11	Progressive familial intrahepatic cholestasis	AR
ABCC8	Familial hyperinsulinism	AR
ACAD9	Acyl-CoA dehydrogenase-9 (ACAD9) deficiency	AR
ACADM	Medium-chain acyl-CoA dehydrogenase (MCAD) deficiency	AR
ACADVL	Very long-chain acyl-CoA dehydrogenase (VLCAD) deficiency	AR
ACAT1	3-ketothiolase deficiency	AR
ACOX1	Peroxisomal acyl-CoA oxidase deficiency	AR
ACSF3	Combined malonic and methylmalonic aciduria	AR
ADA	Adenosine deaminase deficiency	AR
ADAMTS2	Ehlers-Danlos syndrome, dermatosparaxis type	AR
ADGRG1	Bilateral frontoparietal polymicrogyria	AR
AGA	Aspartylglucosaminuria	AR
AGL	Glycogen storage disease type III	AR
AGPS	Rhizomelic chondrodysplasia punctata, type 3	AR
AGXT	Primary hyperoxaluria type 1	AR
AIRE	Autoimmune polyendocrinopathy syndrome type I	AR
ALDH3A2	Sjogren-Larsson syndrome	AR
ALDOB	Hereditary fructose intolerance	AR
ALG6	Congenital disorder of glycosylation type Ic	AR
ALMS1	Alstrom syndrome	AR
ALPL	Hypophosphatasia	AR
AMT	Glycine encephalopathy	AR
AQP2	Nephrogenic diabetes insipidus	AR
ARG1	Arginase deficiency	AR
ARSA	Metachromatic leukodystrophy	AR
ARSB	Mucopolysaccharidosis type VI (Maroteaux-Lamy syndrome)	AR
ASL	Argininosuccinate lyase deficiency	AR
ASNS	Asparagine synthetase deficiency	AR
ASPA	Canavan disease	AR
ASS1	Citrullinemia	AR
ATM	Ataxia-telangiectasia	AR
ATP6V1B1	Renal tubular acidosis with deafness	AR
ATP7B	Wilson disease	AR

AR: autosomal recessive XL: X-linked

GENE	CONDITION	INHERITANCE
BBS1	Bardet-Biedl syndrome type 1	AR
BBS10	Bardet-Biedl syndrome type 10	AR
BBS12	Bardet-Biedl syndrome type 12	AR
BBS2	BBS2-related ciliopathies	AR
BCKDHA	Maple syrup urine disease type Ia	AR
BCKDHB	Maple syrup urine disease type Ib	AR
BCS1L	Mitochondrial complex III deficiency	AR
BLM	Bloom syndrome	AR
BSND	Bartter syndrome	AR
CAPN3	Limb-girdle muscular dystrophy type 2A	AR
CBS	Homocystinuria due to cystathionine beta-synthase deficiency	AR
CDH23	Usher syndrome, type 1D	AR
CEP290	CEP290-related Ciliopathies	AR
CERKL	Retinitis pigmentosa 26	AR
CFTR	Cystic Fibrosis	AR
CHRNE	Congenital myasthenic syndrome	AR
CIITA	Bare lymphocyte syndrome, type II	AR
CLN3	Neuronal ceroid lipofuscinosis	AR
CLN5	Neuronal ceroid lipofuscinosis 5	AR
CLN6	Neuronal ceroid lipofuscinosis, CLN6-related	AR
CLN8	Neuronal ceroid lipofuscinosis, CLN8-related	AR
CLRN1	Usher syndrome, type 3A	AR
CNGB3	Achromatopsia	AR
COL27A1	Steel syndrome	AR
COL4A3	Alport syndrome, COL4A3-related	AR
COL4A4	Alport syndrome, COL4A4-related	AR
COL7A1	Dystrophic epidermolysis bullosa	AR
CPS1	Carbamoylphosphate synthetase I deficiency	AR
CPT1A	Carnitine palmitoyltransferase IA deficiency	AR
CPT2	Carnitine palmitoyltransferase II deficiency	AR
CRB1	CRB1-related retinopathy	AR
CRYL1	GJB6-CRYL1 related nonsyndromic hearing loss	AR, Digenic
CTNS	Cystinosis	AR
CTSK	Pycnodysostosis	AR
CYBA	Chronic granulomatous disease	AR
CYP11B1	Congenital adrenal hyperplasia due to 11-beta-hydroxylase deficiency	AR
CYP11B2	Corticosterone methyloxidase deficiency	AR

AR: autosomal recessive XL: X-linked

GENE	CONDITION	INHERITANCE
CYP17A1	Congenital adrenal hyperplasia due to 17-alpha-hydroxylase deficiency	AR
CYP19A1	Aromatase deficiency	AR
CYP21A2	Congenital adrenal hyperplasia due to 21-hydroxylase deficiency	AR
CYP27A1	Cerebrotendinous xanthomatosis	AR
DBT	Maple syrup urine disease, type II	AR
DCLRE1C	Severe combined immunodeficiency with sensitivity to ionizing radiation	AR
DHCR7	Smith-Lemli-Opitz syndrome	AR
DHDDS	Retinitis pigmentosa 59	AR
DLD	Dihydrolipoamide dehydrogenase deficiency	AR
DNAH5	Primary ciliary dyskinesia, DNAH5-related	AR
DNAI1	Primary ciliary dyskinesia, DNAI1-related	AR
DNAI2	Primary ciliary dyskinesia, DNAI2-related	AR
DYSF	Limb-girdle muscular dystrophy type 2B	AR
EIF2B5	Leukoencephalopathy with vanishing white matter	AR
ELP1	Familial Dysautonomia	AR
ERCC6	ERCC6-related disorders	AR
ERCC8	Cockayne syndrome type A	AR
ESCO2	Roberts syndrome	AR
ETFA	Glutaric aciduria IIA	AR
ETFDH	Glutaric aciduria IIC	AR
ETHE1	Ethylmalonic encephalopathy	AR
EVC	EVC-related bone growth disorders	AR
EVC2	EVC2-related bone growth disorders	AR
EYS	Retinitis pigmentosa 25	AR
FAH	Tyrosinemia, type 1	AR
FAM161A	Retinitis pigmentosa 28	AR
FANCA	Fanconi anemia group A	AR
FANCC	Fanconi anemia group C	AR
FANCG	Fanconi anemia group G	AR
FH	Fumarase deficiency	AR
FKRP	FKRP Alpha-dystroglycanopathies	AR
FKTN	FKTN Alpha-dystroglycanopathies	AR
G6PC	Glycogen storage disease, type 1a	AR
GAA	Pompe disease	AR
GALC	Krabbe disease	AR
GALK1	Galactokinase deficiency	AR
GALT	Galactosemia	AR

AR: autosomal recessive XL: X-linked

GENE	CONDITION	INHERITANCE
GAMT	Guanidinoacetate methyltransferase deficiency	AR
GBA	Gaucher disease	AR
GBE1	Glycogen storage disease IV	AR
GCDH	Glutaric aciduria, type I	AR
GFM1	Combined oxidative phosphorylation deficiency, GFM1-related	AR
GJB2	Nonsyndromic hearing loss 1A	AR
GJB6	GJB6-CRYL1 related nonsyndromic hearing loss	AR, Digenic
GLB1	GLB1-related gangliosidoses	AR
GLDC	Glycine encephalopathy, GLDC-related	AR
GLE1	Lethal congenital contracture syndrome 1	AR
GNE	Inclusion body myopathy type 2 (Nonaka myopathy)	AR
GNPTAB	Mucolipidosis II & III	AR
GNPTG	Mucolipidosis III gamma	AR
GNS	Mucopolysaccharidosis IIID (Sanfilippo syndrome D)	AR
GRHPR	Primary hyperoxaluria type II	AR
HADHA	Trifunctional protein deficiency	AR
HAX1	Severe congenital neutropenia, HAX1-related	AR
HBA1	Alpha thalassemia	AR
HBA2	Alpha thalassemia	AR
HBB	Sickle cell disease	AR
HEXA	Tay-Sachs disease	AR
HEXB	Sandhoff disease	AR
HGSNAT	Mucopolysaccharidosis type IIIC (Sanfilippo syndrome C)	AR
HJV	Hemochromatosis, type 2A	AR
HLCS	Holocarboxylase synthetase deficiency	AR
HMGCL	3-hydroxy-3-methylglutaryl-CoA lyase deficiency	AR
HOGA1	Primary hyperoxaluria type III	AR
HPS1	Hermansky-Pudlak syndrome 1	AR
HPS3	Hermansky-Pudlak syndrome 3	AR
HSD17B4	D-bifunctional protein deficiency	AR
HSD3B2	Congenital adrenal hyperplasia due to 3-beta-hydroxysteroid dehydrogenase 2 deficiency	AR
HYAL1	Mucopolysaccharidosis type IX	AR
HYLS1	Hydrolethalus syndrome	AR
IDUA	Mucopolysaccharidosis, type I (Hurler syndrome)	AR
IVD	Isovaleric Acidemia	AR
KCNJ11	KCNJ11-related hyperinsulinism	AR

GENE	CONDITION	INHERITANCE
LAMA2	Muscular dystrophy, LAMA2-related	AR
LAMA3	Junctional epidermolysis bullosa 2	AR
LAMB3	Junctional epidermolysis bullosa, LAMB3-related	AR
LAMC2	Junctional epidermolysis bullosa, LAMC2-related	AR
LCA5	Leber congenital amaurosis 5	AR
LDLR	Familial Hypercholesterolemia	AR
LDLRAP1	Familial Hypercholesterolemia	AR
LHX3	Combined pituitary hormone deficiency 3	AR
LIFR	Stuve-Wiedemann syndrome	AR
LIPA	Lysosomal acid lipase deficiency	AR
LOXHD1	Nonsyndromic hearing loss 77	AR
LPL	Familial lipoprotein lipase deficiency	AR
LRPPRC	Leigh syndrome with Complex IV deficiency	AR
MAN2B1	Alpha-Mannosidosis	AR
MCOLN1	Mucolipidosis IV	AR
MED17	Postnatal Progressive Microcephaly with Seizures and Brain Atrophy	AR
MESP2	Spondylocostal dysostosis	AR
MFSD8	Neuronal ceroid lipofuscinosis, MFSD8-related	AR
MKS1	MKS1-related ciliopathies	AR
MLC1	Megalencephalic leukoencephalopathy with subcortical cysts	AR
MMAA	Methylmalonic aciduria, cblA type	AR
MMAB	Methylmalonic aciduria, cblB type	AR
MMACHC	Methylmalonic aciduria and homocystinuria, cblC type	AR
MMADHC	Methylmalonic aciduria and homocystinuria, cblD type	AR
MPI	Congenital disorder of glycosylation type Ib	AR
MPL	Congenital amegakaryocytic thrombocytopenia	AR
MPV17	Hepatocerebral mitochondrial DNA depletion syndrome, MPV17-related	AR
MTHFR	Homocystinuria, MTHFR-related	AR
MTRR	Homocystinuria-megaloblastic anemia, cobalamin E type	AR
MTTP	Abetalipoproteinemia	AR
MUT	Methylmalonic aciduria, methylmalonyl-CoA mutase deficiency	AR
MYO7A	MYO7A-related disorders	AR
NAGLU	Mucopolysaccharidosis type IIIB (Sanfilippo syndrome B)	AR
NAGS	N-acetylglutamate synthase deficiency	AR
NBN	Nijmegen breakage syndrome	AR
NDRG1	Charcot-Marie-Tooth disease, type 4D	AR
NDUFAF5	Mitochondrial complex I deficiency (Leigh syndrome)	AR

AR: autosomal recessive XL: X-linked

GENE	CONDITION	INHERITANCE
NDUFS6	Mitochondrial complex I deficiency (Leigh syndrome)	AR
NEB	Nemaline myopathy	AR
NPC1	Niemann-Pick disease, type C1	AR
NPC2	Niemann-Pick disease, type C2	AR
NPHS1	Congenital nephrotic syndrome, type 1	AR
NPHS2	Congenital nephrotic syndrome, type 2	AR
NR2E3	NR2E3-related retinal dystrophies	AR
NTRK1	Congenital insensitivity to pain with anhidrosis	AR
OAT	Gyrate atrophy of choroid and retina	AR
OPA3	Costeff syndrome	AR
PAH	Phenylalanine Hydroxylase deficiency (Phenylketonuria)	AR
PC	Pyruvate carboxylase deficiency	AR
PCCA	Propionic acidemia, PCCA-related	AR
PCCB	Propionic acidemia, PCCB-related	AR
PCDH15	PCDH15-related sensory loss	AR
PDHB	Pyruvate dehydrogenase E1-beta deficiency	AR
PEX1	Zellweger syndrome, PEX1-related	AR
PEX10	Zellweger syndrome, PEX10-related	AR
PEX12	Zellweger syndrome, PEX12-related	AR
PEX2	Zellweger syndrome, PEX2-related	AR
PEX6	Zellweger syndrome, PEX6-related	AR
PEX7	Rhizomelic chondrodysplasia punctata, type 1	AR
PFKM	Glycogen storage disease VII	AR
PHGDH	Phosphoglycerate dehydrogenase deficiency	AR
PKHD1	Polycystic kidney disease, PKHD1-related	AR
PMM2	Congenital disorder of glycosylation type 1a	AR
POMGNT1	POMGNT1 Alpha-dystroglycanopathies	AR
PPT1	Neuronal ceroid lipofuscinosis, PPT1-related	AR
PROP1	Combined pituitary hormone deficiency 2	AR
PSAP	Metachromatic leukodystrophy due to saposin-b deficiency	AR
PTS	Tetrahydrobiopterin deficiency	AR
PUS1	Mitochondrial myopathy and sideroblastic anemia 1	AR
PYGM	Glycogen storage disease type V	AR
RAB23	Carpenter syndrome	AR
RAG2	Omenn syndrome, RAG2-related	AR
RAPSN	RAPSN-associated acetylcholine receptor deficiency	AR
RARS2	Pontocerebellar hypoplasia type 6	AR

AR: autosomal recessive XL: X-linked

GENE	CONDITION	INHERITANCE
RDH12	Leber congenital amaurosis type 13	AR
RMRP	Cartilage-Hair Hypoplasia Anauxetic Dysplasia Spectrum Disorder	AR
RPE65	RPE65-related retinopathy	AR
RPGRIP1L	RPGRIP1L-related ciliopathies	AR
RTEL1	Dyskeratosis congenita type 5	AR
SACS	Autosomal recessive spastic ataxia of Charlevoix-Saguenay	AR
SAMHD1	Aicardi-Goutieres syndrome	AR
SEPSECS	Pontocerebellar hypoplasia type 2D	AR
SGCA	Limb-girdle muscular dystrophy, type 2D	AR
SGCB	Limb-girdle muscular dystrophy, type 2E	AR
SGCG	Limb-girdle muscular dystrophy, type 2C	AR
SGSH	Mucopolysaccharidosis IIIA (Sanfilippo syndrome A)	AR
SLC12A3	Gitelman syndrome	AR
SLC12A6	Andermann syndrome	AR
SLC17A5	Sialic acid storage disorder	AR
SLC22A5	Systemic primary carnitine deficiency	AR
SLC25A13	Citrin deficiency	AR
SLC25A15	Hyperornithinemia-hyperammonemia-homocitrullinemia syndrome (Triple H syndrome)	AR
SLC26A2	SLC26A2-related disorders	AR
SLC26A4	Pendred syndrome	AR
SLC35A3	Arthrogryposis, intellectual disability, and seizures	AR
SLC37A4	Glycogen storage disease, type Ib	AR
SLC39A4	Acrodermatitis enteropathica	AR
SLC4A11	Corneal endothelial dystrophy	AR
SLC7A7	Lysinuric protein intolerance	AR
SMARCAL1	Schimke immunoosseous dysplasia	AR
SMN1	Spinal muscular atrophy	AR
SMPD1	Niemann-Pick disease, type A/B	AR
STAR	Lipoid congenital adrenal hyperplasia	AR
SUMF1	Multiple sulfatase deficiency	AR
TAT	Tyrosinemia, type II	AR
TCIRG1	Osteopetrosis 1	AR
TECPR2	Spastic paraplegia 49	AR
TFR2	Hemochromatosis, type 3	AR
TGM1	Congenital ichthyosis	AR
TH	Segawa syndrome	AR
TMEM216	TMEM216-related ciliopathies	AR

AR: autosomal recessive XL: X-linked

GENE	CONDITION	INHERITANCE
TPP1	Neuronal ceroid lipofuscinosis, TPP1-related	AR
TRMU	Liver failure, acute infantile	AR
TSFM	Combined oxidative phosphorylation deficiency, TSFM-related	AR
TTPA	Ataxia with isolated vitamin E deficiency	AR
TYMP	Mitochondrial neurogastrointestinal encephalopathy (MNGIE) disease	AR
USH1C	USH1C-related disorders	AR
USH2A	Usher syndrome, type 2A	AR
VPS13A	Choreoacanthocytosis	AR
VPS13B	Cohen syndrome	AR
VPS45	Severe congenital neutropenia, VPS45-related	AR
VRK1	Pontocerebellar hypoplasia type 1A	AR
VSX2	Microphthalmia with or without coloboma	AR
WNT10A	WNT10A-related ectodermal dysplasias	AR
XPA	Xeroderma pigmentosum, group A	AR
XPC	Xeroderma pigmentosum, group C	AR
ZFYVE26	Spastic paraplegia 15	AR

CLICK HERE TO VIEW
342 Genes
270 Genes Excluding X-linked Genes
700+ Genes Excluding X-linked Genes

Virtus 700+ Genes

GENE	CONDITION	INHERITANCE
ABCB11	Progressive familial intrahepatic cholestasis	AR
AAAS	Achalasia-addisonianism-alacrimia syndrome	AR
ABCA12	Congenital ichthyosis, ABCA12-related	AR
ABCA3	Surfactant metabolism dysfunction, pulmonary 3	AR
ABCA4	Stargardt disease	AR
ABCB4	Progressive familial intrahepatic cholestasis	AR
ABCC8	Familial hyperinsulinism	AR
ABCD1	Adrenoleukodystrophy, X-linked	XL
ABCD4	Methylmalonic aciduria and homocystinuria, cblJ type	AR
ACAD9	Acyl-CoA dehydrogenase-9 (ACAD9) deficiency	AR
ACADM	Medium-chain acyl-CoA dehydrogenase (MCAD) deficiency	AR
ACADS	Short-chain acyl-coA dehydrogenase (SCAD) deficiency	AR
ACADSB	Short branched chain acyl-CoA dehydrogenase (SBCAD) deficiency	AR
ACADVL	Very long-chain acyl-CoA dehydrogenase (VLCAD) deficiency	AR
ACAT1	3-ketothiolase deficiency	AR
ACOX1	Peroxisomal acyl-CoA oxidase deficiency	AR
ACSF3	Combined malonic and methylmalonic aciduria	AR
ADA	Adenosine deaminase deficiency	AR
ADAMTS2	Ehlers-Danlos syndrome, dermatosparaxis type	AR
ADGRG1	Bilateral frontoparietal polymicrogyria	AR
ADGRV1	Usher syndrome, type IIC	AR
ADK	Hypermethioninemia due to adenosine kinase deficiency	AR
AFF2	Fragile XE syndrome	XL
AGA	Aspartylglucosaminuria	AR
AGL	Glycogen storage disease type III	AR
AGPAT2	Congenital generalized lipodystrophy, type 1	AR
AGPS	Rhizomelic chondrodysplasia punctata, type 3	AR
AGXT	Primary hyperoxaluria type 1	AR
AHCY	Hypermethioninemia due to deficiency of S-adenosylhomocysteine hydrolase	AR
AHI1	Joubert syndrome, AHI1-related	AR
AIMP1	Hypomyelinating leukodystrophy 3	AR
AIPL1	Childhood-onset severe retinal dystrophy, AIPL1-related	AR
AIRE	Autoimmune polyendocrinopathy syndrome type I	AR

AR: autosomal recessive XL: X-linked

GENE	CONDITION	INHERITANCE
AK2	Reticular dysgenesis	AR
AKR1D1	Congenital Bile Acid Synthesis Defect 2	AR
ALDH3A2	Sjogren-Larsson syndrome	AR
ALDH4A1	Hyperprolinemia type II	AR
ALDH7A1	Pyridoxine-dependent epilepsy	AR
ALDOB	Hereditary fructose intolerance	AR
ALG1	Congenital disorder of glycosylation type Ik	AR
ALG12	Congenital disorder of glycosylation type Ig	AR
ALG3	Congenital disorder of glycosylation type Id	AR
ALG6	Congenital disorder of glycosylation type Ic	AR
ALMS1	Alstrom syndrome	AR
ALOX12B	Autosomal recessive, congenital, ichthyosis 2	AR
ALOXE3	Congenital ichthyosiform erythroderma	AR
ALPL	Hypophosphatasia	AR
AMH	Persistent mullerian duct syndrome, type I	AR
AMHR2	Persistent mullerian duct syndrome, type II	AR
AMN	Megaloblastic anemia 1	AR
AMPD2	Pontocerebellar hypoplasia type 9	AR
AMT	Glycine encephalopathy	AR
ANO10	Spinocerebellar ataxia 10	AR
ANO5	Limb girdle muscular dystrophy, type 2L	AR
ANTXR2	Hyaline fibromatosis syndrome	AR
AP1S1	MEDNIK syndrome	AR
AP1S2	X-linked Intellectual disability, AP1S2-related	XL
AP3B1	Hermansky-Pudlak syndrome 2	AR
AP3D1	Hermansky-Pudlak syndrome 10	AR
APOPT1	Mitochondrial complex IV deficiency	AR
AQP2	Nephrogenic diabetes insipidus	AR
AR	Androgen insensitivity syndrome	XL
ARG1	Arginase deficiency	AR
ARL13B	Joubert syndrome, ARL13B-related	AR
ARL6	ARL6-related disorders	AR
ARSA	Metachromatic leukodystrophy	AR
ARSB	Mucopolysaccharidosis type VI (Maroteaux-Lamy syndrome)	AR
ARSE	Chondrodysplasia punctata type 1, X-linked	XL
ARX	X-linked intellectual disability, ARX-related	XL
ASL	Argininosuccinate lyase deficiency	AR

AR: autosomal recessive XL: X-linked

GENE	CONDITION	INHERITANCE
ASNS	Asparagine synthetase deficiency	AR
ASPA	Canavan disease	AR
ASS1	Citrullinemia	AR
ATM	Ataxia-telangiectasia	AR
ATP13A2	Kufor-Rakeb syndrome	AR
ATP6V0A2	Cutis laxa, type IIA	AR
ATP6V0A4	Renal tubular acidosis	AR
ATP6V1B1	Renal tubular acidosis with deafness	AR
ATP6V1E1	Cutis laxa, type IIC	AR
ATP7A	Menkes disease	XL
ATP7B	Wilson disease	AR
ATP8B1	Progressive familial intrahepatic cholestasis	AR
ATRX	Alpha thalassemia X-linked intellectual disability syndrome	XL
AVPR2	Nephrogenic diabetes insipidus	XL
B9D1	Joubert syndrome 27	AR
B9D2	Meckel syndrome 10	AR
BBS1	Bardet-Biedl syndrome type 1	AR
BBS10	Bardet-Biedl syndrome type 10	AR
BBS12	Bardet-Biedl syndrome type 12	AR
BBS2	BBS2-related ciliopathies	AR
BBS4	Bardet-Biedl syndrome 4	AR
BBS5	Bardet-Biedl syndrome 5	AR
BBS7	Bardet-Biedl syndrome 7	AR
BBS9	Bardet-Biedl syndrome 9	AR
BCHE	Butyrylcholinesterase deficiency	AR
BCKDHA	Maple syrup urine disease type Ia	AR
BCKDHB	Maple syrup urine disease type Ib	AR
BCS1L	Mitochondrial complex III deficiency	AR
BLM	Bloom syndrome	AR
BLOC1S3	Hermansky-Pudlak syndrome 8	AR
BLOC1S6	Hermansky-Pudlak syndrome 9	AR
BMP1	Osteogenesis imperfecta, type XIII	AR
BMPER	Diaphanospondylodysostosis	AR
BRIP1	Fanconi anemia group J	AR
BRWD3	X-linked intellectual disability, BRWD3-related	XL
BSND	Bartter syndrome	AR
BTD	Biotinidase deficiency	AR

AR: autosomal recessive XL: X-linked

GENE	CONDITION	INHERITANCE
BTK	X-linked agammaglobulinemia	XL
C19orf12	Mitochondrial membrane protein-associated neurodegeneration	AR
C8orf37	Bardet-Biedl Syndrome 21	AR
CAD	Early Infantile Epileptic Encephalopathy 50	AR
CANT1	Desbuquois dysplasia 1	AR
CAPN3	Limb-girdle muscular dystrophy type 2A	AR
CASP14	Congenital Ichthyosis 12	AR
CASQ2	Catecholaminergic polymorphic ventricular tachycardia	AR
CASR	Neonatal hyperparathyroidism	AR
CAVIN1	Congenital Generalized Lipodystrophy 4	AR
CBS	Homocystinuria due to cystathionine beta-synthase deficiency	AR
CC2D1A	Autosomal recessive intellectual developmental disorder 3	AR
CC2D2A	Joubert syndrome 9	AR
CCDC103	Primary ciliary dyskinesia, type 17	AR
CCDC151	Primary ciliary dyskinesia, type 30	AR
CCDC39	Primary ciliary dyskinesia, type 14	AR
CCDC8	3-M Syndrome	AR
CCDC88C	Congenital hydrocephalus 1	AR
CD247	Severe Combined Immunodeficiency	AR
CD3D	Severe Combined Immunodeficiency	AR
CD3E	Severe Combined Immunodeficiency	AR
CD3G	Severe Combined Immunodeficiency	AR
CD40LG	Hyper IgM syndrome, X-linked	XL
CD59	CD59 deficiency	AR
CD8A	Familial CD8 Deficiency	AR
CDAN1	Dyserythropoietic congenital anemia, type Ia	AR
CDCA7	Immunodeficiency-centromeric instability-facial anomalies syndrome 3	AR
CDH23	Usher syndrome, type 1D	AR
CEP104	Joubert syndrome 25	AR
CEP152	CEP152-related disorders	AR
CEP290	CEP290-related Ciliopathies	AR
CERKL	Retinitis pigmentosa 26	AR
CERS3	Congenital ichthyosis 9	AR
CFTR	Cystic Fibrosis	AR
CHAT	Congenital myasthenic syndrome 6	AR
CHM	Choroideremia	XL
CHMP1A	Pontocerebellar hypoplasia type 8	AR

AR: autosomal recessive XL: X-linked

GENE	CONDITION	INHERITANCE
CHRNE	Congenital myasthenic syndrome	AR
CHRNG	Multiple pterygium syndrome	AR
CHST6	Macular corneal dystrophy, CHST6-related	AR
CIB2	Nonsyndromic hearing loss 48	AR
CIITA	Bare lymphocyte syndrome, type II	AR
CLCF1	Crisponi cold-induced sweating syndrome 2	AR
CLCN1	Autosomal recessive congenital myotonia	AR
CLCN5	Dent disease	XL
CLCNKB	Bartter syndrome	AR
CLN3	Neuronal ceroid lipofuscinosis	AR
CLN5	Neuronal ceroid lipofuscinosis 5	AR
CLN6	Neuronal ceroid lipofuscinosis, CLN6-related	AR
CLN8	Neuronal ceroid lipofuscinosis, CLN8-related	AR
CLP1	Pontocerebellar hypoplasia type 10	AR
CLRN1	Usher syndrome, type 3A	AR
CNGA1	Retinitis Pigmentosa, CNGA1-related	AR
CNGA3	CNGA3-related retinopathy	AR
CNGB1	Retinitis Pigmentosa, CNGB1-related	AR
CNGB3	Achromatopsia	AR
CNTNAP2	Cortical dysplasia-focal epilepsy syndrome	AR
COASY	Pontocerebellar hypoplasia type 12	AR
COL11A2	COL11A2-related disorders	AR
COL17A1	Junctional epidermolysis bullosa	AR
COL27A1	Steel syndrome	AR
COL4A3	Alport syndrome, COL4A3-related	AR
COL4A4	Alport syndrome, COL4A4-related	AR
COL4A5	Alport syndrome, COL4A5-related	XL
COL7A1	Dystrophic epidermolysis bullosa	AR
COLQ	Congenital myasthenic syndrome 5	AR
COQ4	Primary Coenzyme Q10 deficiency 7	AR
CORO1A	Immunodeficiency 8	AR
COX10	Mitochondrial complex IV deficiency	AR
COX15	Mitochondrial complex IV deficiency	AR
COX20	Mitochondrial complex IV deficiency	AR
COX6B1	Mitochondrial complex IV deficiency	AR
CP	Aceruloplasminemia	AR
CPLANE1	Joubert syndrome 17	AR

AR: autosomal recessive XL: X-linked

GENE	CONDITION	INHERITANCE
CPS1	Carbamoylphosphate synthetase I deficiency	AR
CPT1A	Carnitine palmitoyltransferase IA deficiency	AR
CPT2	Carnitine palmitoyltransferase II deficiency	AR
CRADD	Intellectual developmental disorder with variant lissencephaly	AR
CRB1	CRB1-related retinopathy	AR
CRLF1	Crisponi cold-induced sweating syndrome 1	AR
CRTAP	Osteogenesis imperfecta, type VII	AR
CRYL1	GJB6-CRYL1 related nonsyndromic hearing loss	AR, Digenic
CTC1	Cerebroretinal microangiopathy with calcifications and cysts 1	AR
CTNS	Cystinosis	AR
CTSA	Galactosialidosis	AR
CTSC	Papillon-Lefevre syndrome	AR
CTSD	Neuronal ceroid lipofuscinosis, CTSD-related	AR
CTSF	Neuronal ceroid lipofuscinosis 13	AR
CTSK	Pycnodysostosis	AR
CUL4B	X-linked intellectual disability, CUL4B-related	XL
CUL7	Three M syndrome 1	AR
CWC27	Retinitis pigmentosa with or without skeletal anomalies	AR
CYBA	Chronic granulomatous disease	AR
CYBB	Chronic granulomatous disease, X-linked	XL
CYP11A1	Congenital adrenal insufficiency	AR
CYP11B1	Congenital adrenal hyperplasia due to 11-beta-hydroxylase deficiency	AR
CYP11B2	Corticosterone methyloxidase deficiency	AR
CYP17A1	Congenital adrenal hyperplasia due to 17-alpha-hydroxylase deficiency	AR
CYP19A1	Aromatase deficiency	AR
CYP1B1	Primary congenital glaucoma	AR
CYP21A2	Congenital adrenal hyperplasia due to 21-hydroxylase deficiency	AR
CYP27A1	Cerebrotendinous xanthomatosis	AR
CYP27B1	Vitamin Dâ€"dependent rickets, type 1	AR
CYP4F22	Congenital ichthyosis 5	AR
CYP7B1	Congenital bile acid synthesis defect 3	AR
DBT	Maple syrup urine disease, type II	AR
DCAF17	Woodhouse-Sakati syndrome	AR
DCLRE1C	Severe combined immunodeficiency with sensitivity to ionizing radiation	AR
DCX	Lissencephaly, X-linked	XL
DDB2	Xeroderma pigmentosum, group E	AR
DDC	Aromatic l-amino acid decarboxylase deficiency	AR

AR: autosomal recessive XL: X-linked

GENE	CONDITION	INHERITANCE
DDR2	Spondylometaepiphyseal dysplasia	AR
DDX11	Warsaw breakage syndrome	AR
DGUOK	Mitochondrial DNA depletion syndrome 3	AR
DHCR24	Desmosterolosis	AR
DHCR7	Smith-Lemli-Opitz syndrome	AR
DHDDS	Retinitis pigmentosa 59	AR
DKC1	X-linked dyskeratosis congenita	XL
DLAT	Pyruvate dehydrogenase E2 deficiency	AR
DLD	Dihydrolipoamide dehydrogenase deficiency	AR
DLG3	X-linked intellectual disability, DLG3-related	XL
DLL3	Spondylocostal dysostosis 1	AR
DMD	Dystrophinopathies	XL
DNAH5	Primary ciliary dyskinesia, DNAH5-related	AR
DNAI1	Primary ciliary dyskinesia, DNAI1-related	AR
DNAI2	Primary ciliary dyskinesia, DNAI2-related	AR
DNAL1	Primary ciliary dyskinesia, DNAL1-related	AR
DNMT3B	ICF Syndrome	AR
DOCK8	Hyper-IgE syndrome due to DOCK8 defiency	AR
DOK7	Congenital myasthenic syndrome, DOK7-related	AR
DOLK	Congenital disorder of glycosylation type Im	AR
DPYD	Dihydropyrimidine dehydrogenase deficiency	AR
DTNBP1	Hermansky-Pudlak syndrome 7	AR
DUOX2	Congenital hypothyroidism, DUOX2-related	AR
DUOXA2	Congenital hypothyroidism, DUOXA2-related	AR
DYNC2H1	Short-rib thoracic dysplasia 3 with or without polydactyly	AR
DYSF	Limb-girdle muscular dystrophy type 2B	AR
EDA	Hypohidrotic ectodermal dysplasia	XL
EFEMP2	Cutis laxa, type 1B	AR
EIF2AK3	Wolcott-Rallison Syndrome	AR
EIF2B1	Leukoencephalopathy with vanishing white matter	AR
EIF2B2	Leukoencephalopathy with vanishing white matter	AR
EIF2B3	Leukoencephalopathy with vanishing white matter	AR
EIF2B4	Leukoencephalopathy with vanishing white matter	AR
EIF2B5	Leukoencephalopathy with vanishing white matter	AR
ELP1	Familial Dysautonomia	AR
EMD	Emery-Dreifuss muscular dystrophy	XL
EPB42	Spherocytosis, type 5	AR

AR: autosomal recessive XL: X-linked

GENE	CONDITION	INHERITANCE
ERBB3	Familial visceral neuropathy type 1	AR
ERCC2	ERCC2-related disorders	AR
ERCC3	ERCC3-related photosensitivity	AR
ERCC4	ERCC4-related disorders	AR
ERCC5	Xeroderma Pigmentosa, group G	AR
ERCC6	ERCC6-related disorders	AR
ERCC8	Cockayne syndrome type A	AR
ESCO2	Roberts syndrome	AR
ETFA	Glutaric aciduria IIA	AR
ETFB	Glutaric aciduria IIB	AR
ETFDH	Glutaric aciduria IIC	AR
ETHE1	Ethylmalonic encephalopathy	AR
EVC	EVC-related bone growth disorders	AR
EVC2	EVC2-related bone growth disorders	AR
EXOSC3	Pontocerebellar hypoplasia type 1B	AR
EYS	Retinitis pigmentosa 25	AR
F2	Prothrombin-related conditions	AR
F7	Factor VII deficiency	AR
F8	Hemophilia A	XL
F9	Hemophilia B	XL
FA2H	Spastic paraplegia type 35	AR
FAH	Tyrosinemia, type 1	AR
FAM126A	Hypomyelinating leukodystropy type 5	AR
FAM161A	Retinitis pigmentosa 28	AR
FANCA	Fanconi anemia group A	AR
FANCB	Fanconi anemia group B	XL
FANCC	Fanconi anemia group C	AR
FANCD2	Fanconi anemia, group D2	AR
FANCE	Fanconi anemia, group E	AR
FANCF	Fanconi anemia, group F	AR
FANCG	Fanconi anemia group G	AR
FANCI	Fanconi anemia, group I	AR
FANCL	Fanconi anemia, group L	AR
FBP1	Fructose-1,6-bisphosphatase deficiency	AR
FBXL4	Mitochondrial DNA depletion syndrome 13	AR
FGD1	X-linked Aarskog-Scott syndrome	XL
FH	Fumarase deficiency	AR

AR: autosomal recessive XL: X-linked

GENE	CONDITION	INHERITANCE
FHL1	FHL1-related neuromuscular disorders	XL
FKBP10	Osteogenesis imperfecta type XI	AR
FKRP	FKRP Alpha-dystroglycanopathies	AR
FKTN	FKTN Alpha-dystroglycanopathies	AR
FMO3	Trimethylaminuria	AR
FMR1	Fragile X Syndrome Intermediate Allele	XL
FOLR1	Cerebral folate deficiency	AR
FOXN1	T-cell immunodeficiency with thymic aplasia	AR
FOXP3	IPEX syndrome	XL
FOXRED1	Mitochondrial complex I deficiency	AR
FRAS1	Fraser syndrome	AR
FREM2	Fraser syndrome	AR
FTCD	Glutamate formiminotransferase deficiency	AR
FTSJ1	X-linked intellectual disability, FTSJ1-related	XL
FUCA1	Fucosidosis	AR
FXN	Friedreich ataxia	AR
G6PC	Glycogen storage disease, type 1a	AR
G6PC3	Severe congenital neutropenia 4	AR
GAA	Pompe disease	AR
GALC	Krabbe disease	AR
GALE	Galactose epimerase deficiency	AR
GALK1	Galactokinase deficiency	AR
GALNS	Mucopolysaccharidosis IVA (Morquio syndrome A)	AR
GALNT3	Familial hyperphosphatemic tumoral calcinosis	AR
GALT	Galactosemia	AR
GAMT	Guanidinoacetate methyltransferase deficiency	AR
GATM	Cerebral creatine deficiency syndrome 3	AR
GBA	Gaucher disease	AR
GBE1	Glycogen storage disease IV	AR
GCDH	Glutaric aciduria, type I	AR
GDAP1	Charcot-Marie-Tooth disease, GDAP1-related	AR
GDF5	Du Pan Syndrome	AR
GFM1	Combined oxidative phosphorylation deficiency, GFM1-related	AR
GFPT1	Congenital myasthenic syndrome 12	AR
GHR	Growth hormone insensitivity syndrome	AR
GHRHR	Isolated growth hormone deficiency, type 1B	AR
GJB1	Charcot-Marie-Tooth disease, X-linked type 1	XL

AR: autosomal recessive XL: X-linked

GENE	CONDITION	INHERITANCE
GJB2	Nonsyndromic hearing loss 1A	AR
GJB6	GJB6-CRYL1 related nonsyndromic hearing loss	AR, Digenic
GLA	Fabry disease	XL
GLB1	GLB1-related gangliosidoses	AR
GLDC	Glycine encephalopathy, GLDC-related	AR
GLE1	Lethal congenital contracture syndrome 1	AR
GNE	Inclusion body myopathy type 2 (Nonaka myopathy)	AR
GNPAT	Rhizomelic chondrodysplasia punctata, type 2	AR
GNPTAB	Mucolipidosis II & III	AR
GNPTG	Mucolipidosis III gamma	AR
GNRHR	Hypogonadotropic hypogonadism, GNRHR-related	AR
GNS	Mucopolysaccharidosis IIID (Sanfilippo syndrome D)	AR
GORAB	Geroderma osteodysplasticum	AR
GPR143	X-linked Ocular albinism, GPR143-related	XL
GRHPR	Primary hyperoxaluria type II	AR
GRIP1	Fraser syndrome	AR
GSS	Glutathione synthetase deficiency	AR
GUCY2D	Leber congenital amaurosis 1	AR
GUSB	Mucopolysaccharidosis type VII	AR
GYS2	Glycogen storage disease, type 0, liver	AR
HADH	Familial hyperinsulinemic hypoglycemia 4	AR
HADHA	Trifunctional protein deficiency	AR
HADHB	Trifunctional protein deficiency	AR
HAMP	Hemochromatosis, type 2B	AR
HAX1	Severe congenital neutropenia, HAX1-related	AR
HBA1	Alpha thalassemia	AR
HBB	Sickle cell disease	AR
HCFC1	Methylmalonic acidemia with homocystinuria, type cblX	XL
HELLS	Immunodeficiency, Centromeric region instability, Facial anomalies syndrome	AR
HEXA	Tay-Sachs disease	AR
HEXB	Sandhoff disease	AR
HGSNAT	Mucopolysaccharidosis type IIIC (Sanfilippo syndrome C)	AR
HINT1	Neuromyotonia and axonal neuropathy	AR
HJV	Hemochromatosis, type 2A	AR
HLCS	Holocarboxylase synthetase deficiency	AR
HMGCL	3-hydroxy-3-methylglutaryl-CoA lyase deficiency	AR
HMGCS2	3-hydroxy-3-methylglutaryl-CoA synthase 2 deficiency	AR

AR: autosomal recessive XL: X-linked

GENE	CONDITION	INHERITANCE
HOGA1	Primary hyperoxaluria type III	AR
HPD	Tyrosinemia type III	AR
HPS1	Hermansky-Pudlak syndrome 1	AR
HPS3	Hermansky-Pudlak syndrome 3	AR
HPS4	Hermansky-Pudlak syndrome 4	AR
HPS5	Hermansky-Pudlak syndrome 5	AR
HPS6	Hermansky-Pudlak syndrome 6	AR
HSD17B10	HSD10 mitochondrial disease	XL
HSD17B3	17-Beta-Hydroxysteroid Dehydrogenase Deficiency	AR
HSD17B4	D-bifunctional protein deficiency	AR
HSD3B2	Congenital adrenal hyperplasia due to 3-beta-hydroxysteroid dehydrogenase 2 deficiency	AR
HSD3B7	Congenital bile acid synthesis defect 1	AR
HYAL1	Mucopolysaccharidosis type IX	AR
HYLS1	Hydrolethalus syndrome	AR
IDH3B	Retinitis pigmentosa, IDH3B-related	AR
IDS	Mucopolysaccharidosis type II (Hunter syndrome)	XL
IDUA	Mucopolysaccharidosis, type I (Hurler syndrome)	AR
IFT140	IFT140-related disorders	AR
IGHMBP2	IGHMBP2-related neuropathies	AR
IGSF1	X-linked central hypothyroidism and testicular enlargement	XL
IKBKB	Immunodeficiency 15B	AR
IL1RAPL1	X-linked intellectual disability, IL1RAPL1-related	XL
IL2RA	Immunodeficiency due to CD25 deficiency	AR
IL2RG	X-linked severe combined immunodeficiency	XL
IL7R	Severe Combined Immunodeficiency 104	AR
INPP5E	Joubert syndrome 1	AR
INVS	Nephronophthisis 2	AR
ITGA2B	Glanzmann thrombasthenia	AR
ITGA6	Junctional epidermolysis bullosa	AR
ITGB3	Glanzmann thrombasthenia	AR
ITGB4	Junctional epidermolysis bullosa	AR
ITPA	Developmental and epileptic encephalopathy 35	AR
IVD	Isovaleric Acidemia	AR
IYD	Thyroid dyshormonogenesis, IYD-related	AR
JAK3	Severe combined immunodeficiency, JAK3-related	AR
KCNJ1	Bartter syndrome	AR

AR: autosomal recessive XL: X-linked

GENE	CONDITION	INHERITANCE
KCNJ11	KCNJ11-related hyperinsulinism	AR
KCTD7	Progressive myoclonic epilepsy type 3	AR
KDM5C	X-linked intellectual disability, KDM5C-related	XL
KIF14	Primary Autosomal Recessive Microcephaly 20	AR
L1CAM	L1 syndrome	XL
LAMA2	Muscular dystrophy, LAMA2-related	AR
LAMA3	Junctional epidermolysis bullosa 2	AR
LAMB3	Junctional epidermolysis bullosa, LAMB3-related	AR
LAMC2	Junctional epidermolysis bullosa, LAMC2-related	AR
LARS	Infantile liver failure syndrome 1	AR
LCA5	Leber congenital amaurosis 5	AR
LCK	Immunodeficiency 22	AR
LDLR	Familial Hypercholesterolemia	AR
LDLRAP1	Familial Hypercholesterolemia	AR
LHCGR	Leydig cell hypoplasia	AR
LHX3	Combined pituitary hormone deficiency 3	AR
LIFR	Stuve-Wiedemann syndrome	AR
LIG4	LIG4 syndrome	AR
LIPA	Lysosomal acid lipase deficiency	AR
LIPN	Congenital Ichthyosis 8	AR
LMAN1	Combined factor V and VIII deficiency	AR
LMBRD1	Methylmalonic aciduria and homocystinuria, cblF type	AR
LOXHD1	Nonsyndromic hearing loss 77	AR
LPAR6	Hypotrichosis 8	AR
LPL	Familial lipoprotein lipase deficiency	AR
LRAT	Leber congenital amaurosis 14	AR
LRP2	Donnaiâ€"Barrow syndrome	AR
LRPPRC	Leigh syndrome with Complex IV deficiency	AR
LTBP4	Cutis laxa with severe pulmonary, gastrointestinal, and urinary abnormalities	AR
LYST	Chediak-Higashi syndrome	AR
MAK	Retinitis Pigmentosa 62	AR
MALT1	Immunodeficiency 12	AR
MAN2B1	Alpha-Mannosidosis	AR
MANBA	Beta-Mannosidosis	AR
MAT1A	Methionine adenosyltransferase deficiency	AR
MCEE	Methylmalonyl-CoA epimerase deficiency	AR
MCOLN1	Mucolipidosis IV	AR

AR: autosomal recessive XL: X-linked

GENE	CONDITION	INHERITANCE
MCPH1	Primary microcephaly 1, recessive	AR
MED17	Postnatal Progressive Microcephaly with Seizures and Brain Atrophy	AR
MEGF8	Carpenter syndrome 2	AR
MESP2	Spondylocostal dysostosis	AR
MFSD8	Neuronal ceroid lipofuscinosis, MFSD8-related	AR
MID1	Opitz GBBB syndrome, type I	XL
MKKS	Bardet-Biedl syndrome 6	AR
MKS1	MKS1-related ciliopathies	AR
MLC1	Megalencephalic leukoencephalopathy with subcortical cysts	AR
MLYCD	Malonyl-CoA decarboxylase deficiency	AR
MMAA	Methylmalonic aciduria, cblA type	AR
MMAB	Methylmalonic aciduria, cblB type	AR
MMACHC	Methylmalonic aciduria and homocystinuria, cblC type	AR
MMADHC	Methylmalonic aciduria and homocystinuria, cblD type	AR
MPI	Congenital disorder of glycosylation type Ib	AR
MPL	Congenital amegakaryocytic thrombocytopenia	AR
MPV17	Hepatocerebral mitochondrial DNA depletion syndrome, MPV17-related	AR
MRE11	Ataxia-Telangiectasia-Like Disorder 1	AR
MTHFD1	Combined immunodeficiency and megaloblastic anemia	AR
MTHFR	Homocystinuria, MTHFR-related	AR
MTM1	Myotubular myopathy, X-linked	XL
MTMR2	Charcot-Marie-Tooth disease, type 4B1	AR
MTR	Methylcobalamin deficiency, type cblG	AR
MTRR	Homocystinuria-megaloblastic anemia, cobalamin E type	AR
MTTP	Abetalipoproteinemia	AR
MUT	Methylmalonic aciduria, methylmalonyl-CoA mutase deficiency	AR
MVK	Mevalonate kinase deficiency	AR
MYO15A	Nonsyndromic hearing loss, MYO15A-related	AR
MYO7A	MYO7A-related disorders	AR
NAGA	Schindler disease types 1 and 3	AR
NAGLU	Mucopolysaccharidosis type IIIB (Sanfilippo syndrome B)	AR
NAGS	N-acetylglutamate synthase deficiency	AR
NBAS	SOPH syndrome	AR
NBEAL2	Gray platelet syndrome	AR
NBN	Nijmegen breakage syndrome	AR
NCF2	Chronic granulomatous disease 2	AR
NCF4	Chronic granulomatous disease 4	AR

AR: autosomal recessive XL: X-linked

GENE	CONDITION	INHERITANCE
NDP	Norrie disease	XL
NDRG1	Charcot-Marie-Tooth disease, type 4D	AR
NDUFA11	Mitochondrial complex I deficiency	AR
NDUFAF2	Mitochondrial complex I deficiency	AR
NDUFAF5	Mitochondrial complex I deficiency (Leigh syndrome)	AR
NDUFS4	Mitochondrial complex I deficiency	AR
NDUFS6	Mitochondrial complex I deficiency (Leigh syndrome)	AR
NDUFS7	Mitochondrial complex I deficiency	AR
NDUFV1	Mitochondrial complex I deficiency, nuclear type 4	AR
NEB	Nemaline myopathy	AR
NEU1	Sialidosis, type I and II	AR
NGLY1	Congenital disorder of deglycosylation	AR
NHEJ1	Severe combined immunodeficiency with microcephaly, growth retardation, and sensitivity to ionizing radiation	AR
NIPAL4	Autosomal Recessive Congenital Ichthyosis 6	AR
NONO	X-linked intellectual disability syndrome 34	XL
NPC1	Niemann-Pick disease, type C1	AR
NPC2	Niemann-Pick disease, type C2	AR
NPHP1	NPHP1-related ciliopathies	AR
NPHP3	NPHP3-related ciliopathies	AR
NPHS1	Congenital nephrotic syndrome, type 1	AR
NPHS2	Congenital nephrotic syndrome, type 2	AR
NR0B1	Congenital adrenal hypoplasia, X-linked	XL
NR2E3	NR2E3-related retinal dystrophies	AR
NTRK1	Congenital insensitivity to pain with anhidrosis	AR
OAT	Gyrate atrophy of choroid and retina	AR
OBSL1	3M syndrome 2	AR
OCA2	Oculocutaneous albinism type II	AR
OCRL	OCRL-related disoders	XL
OPA3	Costeff syndrome	AR
OPHN1	X-linked intellectual disability-cerebellar hypoplasia syndrome	XL
OSTM1	Osteopetrosis 5	AR
OTC	Ornithine transcarbamylase deficiency	XL
OTOF	Nonsyndromic hearing loss, OTOF-related	AR
P3H1	Osteogenesis imperfecta, type VIII	AR
PAH	Phenylalanine Hydroxylase deficiency (Phenylketonuria)	AR
PAK3	X-linked intellectual disability, PAK3-related	XL

AR: autosomal recessive XL: X-linked

GENE	CONDITION	INHERITANCE
PANK2	Pantothenate kinase-associated neurodegeneration	AR
PC	Pyruvate carboxylase deficiency	AR
PCBD1	Tetrahydrobiopterin deficiency, PCBD1-related	AR
PCCA	Propionic acidemia, PCCA-related	AR
PCCB	Propionic acidemia, PCCB-related	AR
PCDH15	PCDH15-related sensory loss	AR
PCNT	Microcephalic osteodysplastic primordial dwarfism, type II	AR
PDE6A	Retinitis pigmentosa, PDE6A-related	AR
PDHA1	Pyruvate dehydrogenase E1-alpha deficiency	XL
PDHB	Pyruvate dehydrogenase E1-beta deficiency	AR
PDHX	Pyruvate dehydrogenase E3-binding protein deficiency	AR
PDP1	Pyruvate dehydrogenase phosphatase deficiency	AR
PEPD	Prolidase deficiency	AR
PET100	Mitochondrial complex IV deficiency	AR
PEX1	Zellweger syndrome, PEX1-related	AR
PEX10	Zellweger syndrome, PEX10-related	AR
PEX11B	Zellweger spectrum disorder	AR
PEX12	Zellweger syndrome, PEX12-related	AR
PEX13	Zellweger spectrum disorder	AR
PEX14	Zellweger spectrum disorder	AR
PEX16	Zellweger spectrum disorder	AR
PEX19	Zellweger spectrum disorder	AR
PEX2	Zellweger syndrome, PEX2-related	AR
PEX26	Zellweger syndrome	AR
PEX3	Zellweger spectrum disorder	AR
PEX5	Zellweger spectrum disorder	AR
PEX6	Zellweger syndrome, PEX6-related	AR
PEX7	Rhizomelic chondrodysplasia punctata, type 1	AR
PFKM	Glycogen storage disease VII	AR
PGK1	Phosphoglycerate kinase 1 deficiency	XL
PGM3	Immunodeficiency 23	AR
PHF8	X-linked intellectual disability, Siderius type	XL
PHGDH	Phosphoglycerate dehydrogenase deficiency	AR
PHKA1	Glycogen storage disease type IXd	XL
PHKA2	Glycogen storage disease type IXa	XL
PHKB	Glycogen storage disease type IXb	AR
PHKG2	Glycogen storage disease type IXc	AR

AR: autosomal recessive XL: X-linked

GENE	CONDITION	INHERITANCE
PHYH	Refsum disease	AR
PIGN	Multiple congenital anomalies hypotonia seizures syndrome 1	AR
PIP5K1C	Lethal congenital contractural syndrome 3	AR
PJVK	Nonsyndromic hearing loss 59	AR
PKHD1	Polycystic kidney disease, PKHD1-related	AR
PLA2G6	Infantile neuroaxonal dystrophy	AR
PLEKHG5	PLEKHG5-related motor neuropathies	AR
PLOD1	Ehlers-Danlos syndrome with kyphoscoliosis, PLOD1-related	AR
PLOD2	Bruck syndrome 2	AR
PLP1	PLP1-related disorders	XL
PMM2	Congenital disorder of glycosylation type 1a	AR
PNP	Purine nucleoside phosphorylase deficiency	AR
PNPLA1	Autosomal recessive congenital ichthyosis 10	AR
PNPO	Pyridoxamine 5'-phosphate oxidase deficiency	AR
POC1A	Short stature, onychodysplasia, facial dysmorphism, and hypotrichosis syndrome	AR
POLG	POLG-related disorders	AR
POLH	Xeroderma pigmentosum	AR
POLR1C	POLR1C-related disorders	AR
POMGNT1	POMGNT1 Alpha-dystroglycanopathies	AR
POMT1	POMT1 Alpha-dystroglycanopathies	AR
POMT2	POMT2 Alpha-dystroglycanopathies	AR
POR	Antley-Bixler syndrome	AR
POU1F1	Combined pituitary hormone deficiency	AR
POU3F4	X-linked hearing loss, POU3F4-related	XL
PPIB	Osteogenesis imperfecta, type IX	AR
PPT1	Neuronal ceroid lipofuscinosis, PPT1-related	AR
PQBP1	Renpenning syndrome	XL
PRCD	Retinitis pigmentosa 36	AR
PRDM5	Brittle cornea syndrome 2	AR
PRF1	Hemophagocytic lymphohistiocytosis, familial, 2	AR
PRICKLE1	Progressive myoclonic epilepsy, type 1B	AR
PRKDC	PRKDC-related immunodeficiency	AR
PROP1	Combined pituitary hormone deficiency 2	AR
PRPS1	PRPS1-related disorders	XL
PSAP	Metachromatic leukodystrophy due to saposin-b deficiency	AR
PTPRC	PTPRC related-severe combined immunodeficiency	AR
PTS	Tetrahydrobiopterin deficiency	AR

AR: autosomal recessive XL: X-linked

GENE	CONDITION	INHERITANCE
PUS1	Mitochondrial myopathy and sideroblastic anemia 1	AR
PYCR1	Cutis laxa type IIB and type IIIB	AR
PYGL	Glycogen storage disease VI	AR
PYGM	Glycogen storage disease type V	AR
QDPR	Tetrahydrobiopterin deficiency, QDPR-related	AR
RAB23	Carpenter syndrome	AR
RAG1	Omenn syndrome, RAG1-related	AR
RAG2	Omenn syndrome, RAG2-related	AR
RAPSN	RAPSN-associated acetylcholine receptor deficiency	AR
RARS2	Pontocerebellar hypoplasia type 6	AR
RAX	Microphthalmia, isolated 3	AR
RD3	Leber congenital amaurosis 12	AR
RDH12	Leber congenital amaurosis type 13	AR
RDH5	Fundus albipunctatus	AR
RFX5	Bare lymphocyte syndrome type II	AR
RFXANK	MHC class II deficiency	AR
RFXAP	Bare lymphocyte syndrome type II	AR
RHAG	Rh Deficiency syndrome	AR
RLBP1	Retinal dystrophy, RLBP1-related	AR
RMRP	Cartilage-Hair Hypoplasia Anauxetic Dysplasia Spectrum Disorder	AR
RNASEH2A	Aicardi-Goutieres syndrome 4	AR
RNASEH2B	Aicardi Goutieres syndrome 2	AR
RNASEH2C	Aicardi-Goutieres syndrome 3	AR
ROGDI	Kohlschutter-Tonz syndrome	AR
RP2	X-linked Retinitis pigmentosa, RP2-related	XL
RPE65	RPE65-related retinopathy	AR
RPGR	X-linked Retinitis pigmentosa, RPGR-related	XL
RPGRIP1	Leber congenital amaurosis and Cone-rod dystrophy	AR
RPGRIP1L	RPGRIP1L-related ciliopathies	AR
RS1	Juvenile retinoschisis, X-linked	XL
RSPH9	Primary ciliary dyskinesia 12	AR
RTEL1	Dyskeratosis congenita type 5	AR
SACS	Autosomal recessive spastic ataxia of Charlevoix-Saguenay	AR
SAG	Retinitis pigmentosa 47	AR
SAMD9	Normophosphatemic Familial Tumoral Calcinosis	AR
SAMHD1	Aicardi-Goutieres syndrome	AR
SARS2	Hyperuricemia, pulmonary hypertension, renal failure, and alkalosis syndrome	AR

AR: autosomal recessive XL: X-linked

GENE	CONDITION	INHERITANCE
SBDS	Shwachman-Diamond syndrome	AR
SCO1	Mitochondrial complex IV deficiency	AR
SCO2	Mitochondrial complex IV deficiency	AR
SDCCAG8	Bardet-Biedl syndrome and Senior-Loken syndrome	AR
SDR9C7	Autosomal recessive congenital ichthyosis	AR
SEC23B	Congenital dyserythropoietic anemia, type II	AR
SELENON	Rigid spine muscular dystrophy	AR
SEPSECS	Pontocerebellar hypoplasia type 2D	AR
SERPINF1	Osteogenesis imperfecta, type VI	AR
SGCA	Limb-girdle muscular dystrophy, type 2D	AR
SGCB	Limb-girdle muscular dystrophy, type 2E	AR
SGCD	Limb-girdle muscular dystrophy, type 2F	AR
SGCG	Limb-girdle muscular dystrophy, type 2C	AR
SGSH	Mucopolysaccharidosis IIIA (Sanfilippo syndrome A)	AR
SH3TC2	Charcot-Marie-Tooth disease, SH3TC2-related	AR
SKIV2L	Trichohepatoenteric syndrome 2	AR
SLC12A1	Bartter syndrome	AR
SLC12A3	Gitelman syndrome	AR
SLC12A6	Andermann syndrome	AR
SLC16A2	Allan-Herndon-Dudley syndrome	XL
SLC17A5	Sialic acid storage disorder	AR
SLC19A2	Thiamine-responsive megaloblastic anemia syndrome	AR
SLC19A3	Biotin-responsive basal ganglia disease	AR
SLC1A4	Spastic tetraplegia, thin corpus callosum, and progressive microcephaly syndrome	AR
SLC22A5	Systemic primary carnitine deficiency	AR
SLC25A13	Citrin deficiency	AR
SLC25A15	Hyperornithinemia-hyperammonemia-homocitrullinemia syndrome (Triple H syndrome)	AR
SLC25A20	Carnitine-acylcarnitine translocase deficiency	AR
SLC26A2	SLC26A2-related disorders	AR
SLC26A3	Congenital secretory chloride diarrhea	AR
SLC26A4	Pendred syndrome	AR
SLC27A4	Ichthyosis prematurity syndrome	AR
SLC2A10	Arterial tortuosity syndrome	AR
SLC2A2	Fanconi-Bickel syndrome	AR
SLC34A3	Hereditary hypophosphatemic rickets with hypercalciuria	AR
SLC35A3	Arthrogryposis, intellectual disability, and seizures	AR
SLC37A4	Glycogen storage disease, type Ib	AR

AR: autosomal recessive XL: X-linked

GENE	CONDITION	INHERITANCE
SLC39A4	Acrodermatitis enteropathica	AR
SLC3A1	Cystinuria, type I	AR
SLC45A2	Oculocutaneous albinism, type IV	AR
SLC46A1	Hereditary folate malabsorption	AR
SLC4A1	Distal Renal Tubular Acidosis	AR
SLC4A11	Corneal endothelial dystrophy	AR
SLC5A5	Thyroid dyshormonogenesis, SLC5A5-related	AR
SLC6A19	Hartnup disorder	AR
SLC6A8	Creatine deficiency syndrome	XL
SLC7A7	Lysinuric protein intolerance	AR
SLC7A9	Cystinuria, non-type I	AR
SMARCAL1	Schimke immunoosseous dysplasia	AR
SMN1	Spinal muscular atrophy	AR
SMPD1	Niemann-Pick disease, type A/B	AR
SNAP29	Cerebral dysgenesis, neuropathy, ichthyosis, and palmoplantar keratoderma syndrome	AR
SNX10	Osteopetrosis 8	AR
SP110	Hepatic venoocclusive disease with immunodeficiency	AR
SPATA7	Leber congenital amaurosis (LCA) and juvenile retinitis pigmentosa (RP)	AR
SPG11	SPG11-related Neuromuscular Disorders	AR
SPG21	Mast syndrome	AR
SPG7	Spastic paraplegia type 7	AR
SPINK5	Netherton syndrome	AR
SPR	Sepiapterin Reductase Deficiency	AR
SRD5A2	5-alpha reductase deficiency	AR
ST3GAL5	Salt and pepper developmental regression syndrome	AR
STAR	Lipoid congenital adrenal hyperplasia	AR
STK4	Combined immunodeficiency due to STK4 deficiency	AR
STX11	Familial hemophagocytic lymphohistiocytosis	AR
STXBP2	Familial hemophagocytic lymphohistiocytosis	AR
SUCLA2	Mitochondrial DNA depletion syndrome 5	AR
SUMF1	Multiple sulfatase deficiency	AR
SUOX	Sulfite oxidase deficiency	AR
SURF1	Leigh syndrome, SURF1-related	AR
SYN1	X-linked epilepsy with variable learning disabilities	XL
SYNE4	Autosomal recessive deafness 76	AR
TAT	Tyrosinemia, type II	AR
TAZ	Barth syndrome	XL

AR: autosomal recessive XL: X-linked

GENE	CONDITION	INHERITANCE
TBCE	Hypoparathyroidism-retardation-dysmorphism syndrome	AR
TBX19	Adrenocorticotropic hormone deficiency	AR
TCIRG1	Osteopetrosis 1	AR
TCTN1	Joubert syndrome 13	AR
TCTN2	TCTN2-related ciliopathies	AR
TCTN3	Joubert syndrome 18	AR
TECPR2	Spastic paraplegia 49	AR
TERT	Dyskeratosis congenita type 4	AR
TF	Atransferrinemia	AR
TFR2	Hemochromatosis, type 3	AR
TG	Thyroid dyshormonogenesis, TG-related	AR
TGM1	Congenital ichthyosis	AR
TH	Segawa syndrome	AR
THOC2	X-linked Intellectual disability, THOC2-related	XL
TK2	Mitochondrial DNA depletion syndrome 2	AR
TMC1	Nonsyndromic hearing loss 7	AR
TMEM138	Joubert syndrome 16	AR
TMEM216	TMEM216-related ciliopathies	AR
TMEM231	Joubert syndrome 20	AR
TMEM237	Joubert syndrome 14	AR
TMEM38B	Osteogenesis imperfecta, type XIV	AR
TMEM67	COACH syndrome	AR
TMEM70	Mitochondrial complex V deficiency type 2	AR
TMPRSS3	Nonsyndromic hearing loss, TMPRSS3-related	AR
TNFSF11	Osteopetrosis 2	AR
TNXB	Ehlers-Danlos-like syndrome due to tenascin-X deficiency	AR
TPO	Thyroid dyshormonogenesis, TPO-related	AR
TPP1	Neuronal ceroid lipofuscinosis, TPP1-related	AR
TRAPPC11	Limb-girdle muscular dystrophy 18	AR
TRDN	Catecholaminergic polymorphic ventricular tachycardia	AR
TREX1	Aicardi-Goutieres syndrome 1	AR
TRHR	Generalized thyrotropin-releasing hormone resistance	AR
TRIM32	TRIM32-related disorders	AR
TRIM37	Mulibrey nanism	AR
TRMU	Liver failure, acute infantile	AR
TRPM6	Hypomagnesemia 1	AR
TSEN2	Pontocerebellar hypoplasia type 2B	AR

AR: autosomal recessive XL: X-linked

GENE	CONDITION	INHERITANCE
TSEN34	Pontocerebellar hypoplasia type 2C	AR
TSEN54	Pontocerebellar hypoplasia type 2A	AR
TSFM	Combined oxidative phosphorylation deficiency, TSFM-related	AR
TSHB	Congenital hypothyroidism, TSHB-related	AR
TSHR	Congenital hypothyroidism, TSHR-related	AR
TTC37	Trichohepatoenteric syndrome	AR
TTC7A	Gastrointestinal defects and immunodeficiency syndrome	AR
TTC8	Bardet-Biedl syndrome 8	AR
TTPA	Ataxia with isolated vitamin E deficiency	AR
TULP1	TULP1-related retinal disorders	AR
TYMP	Mitochondrial neurogastrointestinal encephalopathy (MNGIE) disease	AR
TYR	Oculocutaneous albinism types 1A and 1B	AR
TYRP1	Oculocutaneous albinism, type III	AR
UGT1A1	Crigler-Najjar syndrome	AR
UNC13D	Familial hemophagocytic lymphohistiocytosis type 3	AR
UPF3B	Lujan-Fryns syndrome, UPF3B-related	XL
USH1C	USH1C-related disorders	AR
USH1G	Usher syndrome type IG	AR
USH2A	Usher syndrome, type 2A	AR
VDR	Vitamin D-dependent rickets, type 2A	AR
VLDLR	Cerebellar ataxia, mental retardation, and dysequilibrium syndrome 1	AR
VPS13A	Choreoacanthocytosis	AR
VPS13B	Cohen syndrome	AR
VPS45	Severe congenital neutropenia, VPS45-related	AR
VPS53	Pontocerebellar hypoplasia type 2E	AR
VRK1	Pontocerebellar hypoplasia type 1A	AR
VSX2	Microphthalmia with or without coloboma	AR
WAS	WAS-related hematopoietic disorder	XL
WHRN	Usher syndrome type 2D	AR
WISP3	Progressive pseudorheumatoid dysplasia	AR
WNT1	Osteogenesis imperfecta type 15	AR
WNT10A	WNT10A-related ectodermal dysplasias	AR
WRN	Werner syndrome	AR
XPA	Xeroderma pigmentosum, group A	AR
XPC	Xeroderma pigmentosum, group C	AR
ZAP70	ZAP70-related Immunodeficiency	AR
ZBTB24	Immunodeficiency-centromeric instability-facial anomalies syndrome 2	AR

AR: autosomal recessive XL: X-linked

GENE	CONDITION	INHERITANCE
ZDHHC9	X-linked intellectual disability, ZDHHC9-related	XL
ZFYVE26	Spastic paraplegia 15	AR
ZNF469	Brittle cornea syndrome 1	AR
ZNF711	X-linked intellectual disability, ZNF711-related	XL
HBA2	Alpha thalassemia	AR

CLICK HERE TO VIEW
342 Genes
270 Genes Excluding X-linked Genes
700+ Genes

Virtus 700+ Genes Excluding X-linked Genes

GENE	CONDITION	INHERITANCE
ABCB11	Progressive familial intrahepatic cholestasis	AR
AAAS	Achalasia-addisonianism-alacrimia syndrome	AR
ABCA12	Congenital ichthyosis, ABCA12-related	AR
ABCA3	Surfactant metabolism dysfunction, pulmonary 3	AR
ABCA4	Stargardt disease	AR
ABCB4	Progressive familial intrahepatic cholestasis	AR
ABCC8	Familial hyperinsulinism	AR
ABCD4	Methylmalonic aciduria and homocystinuria, cblJ type	AR
ACAD9	Acyl-CoA dehydrogenase-9 (ACAD9) deficiency	AR
ACADM	Medium-chain acyl-CoA dehydrogenase (MCAD) deficiency	AR
ACADS	Short-chain acyl-coA dehydrogenase (SCAD) deficiency	AR
ACADSB	Short branched chain acyl-CoA dehydrogenase (SBCAD) deficiency	AR
ACADVL	Very long-chain acyl-CoA dehydrogenase (VLCAD) deficiency	AR
ACAT1	3-ketothiolase deficiency	AR
ACOX1	Peroxisomal acyl-CoA oxidase deficiency	AR
ACSF3	Combined malonic and methylmalonic aciduria	AR
ADA	Adenosine deaminase deficiency	AR
ADAMTS2	Ehlers-Danlos syndrome, dermatosparaxis type	AR
ADGRG1	Bilateral frontoparietal polymicrogyria	AR
ADGRV1	Usher syndrome, type IIC	AR
ADK	Hypermethioninemia due to adenosine kinase deficiency	AR
AGA	Aspartylglucosaminuria	AR
AGL	Glycogen storage disease type III	AR
AGPAT2	Congenital generalized lipodystrophy, type 1	AR
AGPS	Rhizomelic chondrodysplasia punctata, type 3	AR
AGXT	Primary hyperoxaluria type 1	AR
AHCY	Hypermethioninemia due to deficiency of S-adenosylhomocysteine hydrolase	AR
AHI1	Joubert syndrome, AHI1-related	AR
AIMP1	Hypomyelinating leukodystrophy 3	AR
AIPL1	Childhood-onset severe retinal dystrophy, AIPL1-related	AR
AIRE	Autoimmune polyendocrinopathy syndrome type I	AR
AK2	Reticular dysgenesis	AR
AKR1D1	Congenital Bile Acid Synthesis Defect 2	AR

AR: autosomal recessive XL: X-linked

GENE	CONDITION	INHERITANCE
ALDH3A2	Sjogren-Larsson syndrome	AR
ALDH4A1	Hyperprolinemia type II	AR
ALDH7A1	Pyridoxine-dependent epilepsy	AR
ALDOB	Hereditary fructose intolerance	AR
ALG1	Congenital disorder of glycosylation type Ik	AR
ALG12	Congenital disorder of glycosylation type Ig	AR
ALG3	Congenital disorder of glycosylation type Id	AR
ALG6	Congenital disorder of glycosylation type Ic	AR
ALMS1	Alstrom syndrome	AR
ALOX12B	Autosomal recessive, congenital, ichthyosis 2	AR
ALOXE3	Congenital ichthyosiform erythroderma	AR
ALPL	Hypophosphatasia	AR
AMH	Persistent mullerian duct syndrome, type I	AR
AMHR2	Persistent mullerian duct syndrome, type II	AR
AMN	Megaloblastic anemia 1	AR
AMPD2	Pontocerebellar hypoplasia type 9	AR
AMT	Glycine encephalopathy	AR
ANO10	Spinocerebellar ataxia 10	AR
ANO5	Limb girdle muscular dystrophy, type 2L	AR
ANTXR2	Hyaline fibromatosis syndrome	AR
AP1S1	MEDNIK syndrome	AR
AP3B1	Hermansky-Pudlak syndrome 2	AR
AP3D1	Hermansky-Pudlak syndrome 10	AR
APOPT1	Mitochondrial complex IV deficiency	AR
AQP2	Nephrogenic diabetes insipidus	AR
ARG1	Arginase deficiency	AR
ARL13B	Joubert syndrome, ARL13B-related	AR
ARL6	ARL6-related disorders	AR
ARSA	Metachromatic leukodystrophy	AR
ARSB	Mucopolysaccharidosis type VI (Maroteaux-Lamy syndrome)	AR
ASL	Argininosuccinate lyase deficiency	AR
ASNS	Asparagine synthetase deficiency	AR
ASPA	Canavan disease	AR
ASS1	Citrullinemia	AR
ATM	Ataxia-telangiectasia	AR
ATP13A2	Kufor-Rakeb syndrome	AR
ATP6V0A2	Cutis laxa, type IIA	AR

AR: autosomal recessive XL: X-linked

GENE	CONDITION	INHERITANCE
ATP6V0A4	Renal tubular acidosis	AR
ATP6V1B1	Renal tubular acidosis with deafness	AR
ATP6V1E1	Cutis laxa, type IIC	AR
ATP7B	Wilson disease	AR
ATP8B1	Progressive familial intrahepatic cholestasis	AR
B9D1	Joubert syndrome 27	AR
B9D2	Meckel syndrome 10	AR
BBS1	Bardet-Biedl syndrome type 1	AR
BBS10	Bardet-Biedl syndrome type 10	AR
BBS12	Bardet-Biedl syndrome type 12	AR
BBS2	BBS2-related ciliopathies	AR
BBS4	Bardet-Biedl syndrome 4	AR
BBS5	Bardet-Biedl syndrome 5	AR
BBS7	Bardet-Biedl syndrome 7	AR
BBS9	Bardet-Biedl syndrome 9	AR
BCHE	Butyrylcholinesterase deficiency	AR
BCKDHA	Maple syrup urine disease type Ia	AR
BCKDHB	Maple syrup urine disease type Ib	AR
BCS1L	Mitochondrial complex III deficiency	AR
BLM	Bloom syndrome	AR
BLOC1S3	Hermansky-Pudlak syndrome 8	AR
BLOC1S6	Hermansky-Pudlak syndrome 9	AR
BMP1	Osteogenesis imperfecta, type XIII	AR
BMPER	Diaphanospondylodysostosis	AR
BRIP1	Fanconi anemia group J	AR
BSND	Bartter syndrome	AR
BTD	Biotinidase deficiency	AR
C19orf12	Mitochondrial membrane protein-associated neurodegeneration	AR
C8orf37	Bardet-Biedl Syndrome 21	AR
CAD	Early Infantile Epileptic Encephalopathy 50	AR
CANT1	Desbuquois dysplasia 1	AR
CAPN3	Limb-girdle muscular dystrophy type 2A	AR
CASP14	Congenital Ichthyosis 12	AR
CASQ2	Catecholaminergic polymorphic ventricular tachycardia	AR
CASR	Neonatal hyperparathyroidism	AR
CAVIN1	Congenital Generalized Lipodystrophy 4	AR
CBS	Homocystinuria due to cystathionine beta-synthase deficiency	AR

GENE	CONDITION	INHERITANCE
CC2D1A	Autosomal recessive intellectual developmental disorder 3	AR
CC2D2A	Joubert syndrome 9	AR
CCDC103	Primary ciliary dyskinesia, type 17	AR
CCDC151	Primary ciliary dyskinesia, type 30	AR
CCDC39	Primary ciliary dyskinesia, type 14	AR
CCDC8	3-M Syndrome	AR
CCDC88C	Congenital hydrocephalus 1	AR
CD247	Severe Combined Immunodeficiency	AR
CD3D	Severe Combined Immunodeficiency	AR
CD3E	Severe Combined Immunodeficiency	AR
CD3G	Severe Combined Immunodeficiency	AR
CD59	CD59 deficiency	AR
CD8A	Familial CD8 Deficiency	AR
CDAN1	Dyserythropoietic congenital anemia, type Ia	AR
CDCA7	Immunodeficiency-centromeric instability-facial anomalies syndrome 3	AR
CDH23	Usher syndrome, type 1D	AR
CEP104	Joubert syndrome 25	AR
CEP152	CEP152-related disorders	AR
CEP290	CEP290-related Ciliopathies	AR
CERKL	Retinitis pigmentosa 26	AR
CERS3	Congenital ichthyosis 9	AR
CFTR	Cystic Fibrosis	AR
CHAT	Congenital myasthenic syndrome 6	AR
CHMP1A	Pontocerebellar hypoplasia type 8	AR
CHRNE	Congenital myasthenic syndrome	AR
CHRNG	Multiple pterygium syndrome	AR
CHST6	Macular corneal dystrophy, CHST6-related	AR
CIB2	Nonsyndromic hearing loss 48	AR
CIITA	Bare lymphocyte syndrome, type II	AR
CLCF1	Crisponi cold-induced sweating syndrome 2	AR
CLCN1	Autosomal recessive congenital myotonia	AR
CLCNKB	Bartter syndrome	AR
CLN3	Neuronal ceroid lipofuscinosis	AR
CLN5	Neuronal ceroid lipofuscinosis 5	AR
CLN6	Neuronal ceroid lipofuscinosis, CLN6-related	AR
CLN8	Neuronal ceroid lipofuscinosis, CLN8-related	AR
CLP1	Pontocerebellar hypoplasia type 10	AR

GENE	CONDITION	INHERITANCE
CLRN1	Usher syndrome, type 3A	AR
CNGA1	Retinitis Pigmentosa, CNGA1-related	AR
CNGA3	CNGA3-related retinopathy	AR
CNGB1	Retinitis Pigmentosa, CNGB1-related	AR
CNGB3	Achromatopsia	AR
CNTNAP2	Cortical dysplasia-focal epilepsy syndrome	AR
COASY	Pontocerebellar hypoplasia type 12	AR
COL11A2	COL11A2-related disorders	AR
COL17A1	Junctional epidermolysis bullosa	AR
COL27A1	Steel syndrome	AR
COL4A3	Alport syndrome, COL4A3-related	AR
COL4A4	Alport syndrome, COL4A4-related	AR
COL7A1	Dystrophic epidermolysis bullosa	AR
COLQ	Congenital myasthenic syndrome 5	AR
COQ4	Primary Coenzyme Q10 deficiency 7	AR
CORO1A	Immunodeficiency 8	AR
COX10	Mitochondrial complex IV deficiency	AR
COX15	Mitochondrial complex IV deficiency	AR
COX20	Mitochondrial complex IV deficiency	AR
COX6B1	Mitochondrial complex IV deficiency	AR
CP	Aceruloplasminemia	AR
CPLANE1	Joubert syndrome 17	AR
CPS1	Carbamoylphosphate synthetase I deficiency	AR
CPT1A	Carnitine palmitoyltransferase IA deficiency	AR
CPT2	Carnitine palmitoyltransferase II deficiency	AR
CRADD	Intellectual developmental disorder with variant lissencephaly	AR
CRB1	CRB1-related retinopathy	AR
CRLF1	Crisponi cold-induced sweating syndrome 1	AR
CRTAP	Osteogenesis imperfecta, type VII	AR
CRYL1	GJB6-CRYL1 related nonsyndromic hearing loss	AR, Digenic
CTC1	Cerebroretinal microangiopathy with calcifications and cysts 1	AR
CTNS	Cystinosis	AR
CTSA	Galactosialidosis	AR
CTSC	Papillon-Lefevre syndrome	AR
CTSD	Neuronal ceroid lipofuscinosis, CTSD-related	AR
CTSF	Neuronal ceroid lipofuscinosis 13	AR
CTSK	Pycnodysostosis	AR

AR: autosomal recessive XL: X-linked

GENE	CONDITION	INHERITANCE
CUL7	Three M syndrome 1	AR
CWC27	Retinitis pigmentosa with or without skeletal anomalies	AR
CYBA	Chronic granulomatous disease	AR
CYP11A1	Congenital adrenal insufficiency	AR
CYP11B1	Congenital adrenal hyperplasia due to 11-beta-hydroxylase deficiency	AR
CYP11B2	Corticosterone methyloxidase deficiency	AR
CYP17A1	Congenital adrenal hyperplasia due to 17-alpha-hydroxylase deficiency	AR
CYP19A1	Aromatase deficiency	AR
CYP1B1	Primary congenital glaucoma	AR
CYP21A2	Congenital adrenal hyperplasia due to 21-hydroxylase deficiency	AR
CYP27A1	Cerebrotendinous xanthomatosis	AR
CYP27B1	Vitamin Dâ€"dependent rickets, type 1	AR
CYP4F22	Congenital ichthyosis 5	AR
CYP7B1	Congenital bile acid synthesis defect 3	AR
DBT	Maple syrup urine disease, type II	AR
DCAF17	Woodhouse-Sakati syndrome	AR
DCLRE1C	Severe combined immunodeficiency with sensitivity to ionizing radiation	AR
DDB2	Xeroderma pigmentosum, group E	AR
DDC	Aromatic l-amino acid decarboxylase deficiency	AR
DDR2	Spondylometaepiphyseal dysplasia	AR
DDX11	Warsaw breakage syndrome	AR
DGUOK	Mitochondrial DNA depletion syndrome 3	AR
DHCR24	Desmosterolosis	AR
DHCR7	Smith-Lemli-Opitz syndrome	AR
DHDDS	Retinitis pigmentosa 59	AR
DLAT	Pyruvate dehydrogenase E2 deficiency	AR
DLD	Dihydrolipoamide dehydrogenase deficiency	AR
DLL3	Spondylocostal dysostosis 1	AR
DNAH5	Primary ciliary dyskinesia, DNAH5-related	AR
DNAI1	Primary ciliary dyskinesia, DNAI1-related	AR
DNAI2	Primary ciliary dyskinesia, DNAI2-related	AR
DNAL1	Primary ciliary dyskinesia, DNAL1-related	AR
DNMT3B	ICF Syndrome	AR
DOCK8	Hyper-IgE syndrome due to DOCK8 defiency	AR
DOK7	Congenital myasthenic syndrome, DOK7-related	AR
DOLK	Congenital disorder of glycosylation type Im	AR
DPYD	Dihydropyrimidine dehydrogenase deficiency	AR

AR: autosomal recessive XL: X-linked

GENE	CONDITION	INHERITANCE
DTNBP1	Hermansky-Pudlak syndrome 7	AR
DUOX2	Congenital hypothyroidism, DUOX2-related	AR
DUOXA2	Congenital hypothyroidism, DUOXA2-related	AR
DYNC2H1	Short-rib thoracic dysplasia 3 with or without polydactyly	AR
DYSF	Limb-girdle muscular dystrophy type 2B	AR
EFEMP2	Cutis laxa, type 1B	AR
EIF2AK3	Wolcott-Rallison Syndrome	AR
EIF2B1	Leukoencephalopathy with vanishing white matter	AR
EIF2B2	Leukoencephalopathy with vanishing white matter	AR
EIF2B3	Leukoencephalopathy with vanishing white matter	AR
EIF2B4	Leukoencephalopathy with vanishing white matter	AR
EIF2B5	Leukoencephalopathy with vanishing white matter	AR
ELP1	Familial Dysautonomia	AR
EPB42	Spherocytosis, type 5	AR
ERBB3	Familial visceral neuropathy type 1	AR
ERCC2	ERCC2-related disorders	AR
ERCC3	ERCC3-related photosensitivity	AR
ERCC4	ERCC4-related disorders	AR
ERCC5	Xeroderma Pigmentosa, group G	AR
ERCC6	ERCC6-related disorders	AR
ERCC8	Cockayne syndrome type A	AR
ESCO2	Roberts syndrome	AR
ETFA	Glutaric aciduria IIA	AR
ETFB	Glutaric aciduria IIB	AR
ETFDH	Glutaric aciduria IIC	AR
ETHE1	Ethylmalonic encephalopathy	AR
EVC	EVC-related bone growth disorders	AR
EVC2	EVC2-related bone growth disorders	AR
EXOSC3	Pontocerebellar hypoplasia type 1B	AR
EYS	Retinitis pigmentosa 25	AR
F2	Prothrombin-related conditions	AR
F7	Factor VII deficiency	AR
FA2H	Spastic paraplegia type 35	AR
FAH	Tyrosinemia, type 1	AR
FAM126A	Hypomyelinating leukodystropy type 5	AR
FAM161A	Retinitis pigmentosa 28	AR
FANCA	Fanconi anemia group A	AR

AR: autosomal recessive XL: X-linked

GENE	CONDITION	INHERITANCE
FANCC	Fanconi anemia group C	AR
FANCD2	Fanconi anemia, group D2	AR
FANCE	Fanconi anemia, group E	AR
FANCF	Fanconi anemia, group F	AR
FANCG	Fanconi anemia group G	AR
FANCI	Fanconi anemia, group I	AR
FANCL	Fanconi anemia, group L	AR
FBP1	Fructose-1,6-bisphosphatase deficiency	AR
FBXL4	Mitochondrial DNA depletion syndrome 13	AR
FH	Fumarase deficiency	AR
FKBP10	Osteogenesis imperfecta type XI	AR
FKRP	FKRP Alpha-dystroglycanopathies	AR
FKTN	FKTN Alpha-dystroglycanopathies	AR
FMO3	Trimethylaminuria	AR
FOLR1	Cerebral folate deficiency	AR
FOXN1	T-cell immunodeficiency with thymic aplasia	AR
FOXRED1	Mitochondrial complex I deficiency	AR
FRAS1	Fraser syndrome	AR
FREM2	Fraser syndrome	AR
FTCD	Glutamate formiminotransferase deficiency	AR
FUCA1	Fucosidosis	AR
FXN	Friedreich ataxia	AR
G6PC	Glycogen storage disease, type 1a	AR
G6PC3	Severe congenital neutropenia 4	AR
GAA	Pompe disease	AR
GALC	Krabbe disease	AR
GALE	Galactose epimerase deficiency	AR
GALK1	Galactokinase deficiency	AR
GALNS	Mucopolysaccharidosis IVA (Morquio syndrome A)	AR
GALNT3	Familial hyperphosphatemic tumoral calcinosis	AR
GALT	Galactosemia	AR
GAMT	Guanidinoacetate methyltransferase deficiency	AR
GATM	Cerebral creatine deficiency syndrome 3	AR
GBA	Gaucher disease	AR
GBE1	Glycogen storage disease IV	AR
GCDH	Glutaric aciduria, type I	AR
GDAP1	Charcot-Marie-Tooth disease, GDAP1-related	AR

AR: autosomal recessive XL: X-linked

GENE	CONDITION	INHERITANCE
GDF5	Du Pan Syndrome	AR
GFM1	Combined oxidative phosphorylation deficiency, GFM1-related	AR
GFPT1	Congenital myasthenic syndrome 12	AR
GHR	Growth hormone insensitivity syndrome	AR
GHRHR	Isolated growth hormone deficiency, type 1B	AR
GJB2	Nonsyndromic hearing loss 1A	AR
GJB6	GJB6-CRYL1 related nonsyndromic hearing loss	AR, Digenic
GLB1	GLB1-related gangliosidoses	AR
GLDC	Glycine encephalopathy, GLDC-related	AR
GLE1	Lethal congenital contracture syndrome 1	AR
GNE	Inclusion body myopathy type 2 (Nonaka myopathy)	AR
GNPAT	Rhizomelic chondrodysplasia punctata, type 2	AR
GNPTAB	Mucolipidosis II & III	AR
GNPTG	Mucolipidosis III gamma	AR
GNRHR	Hypogonadotropic hypogonadism, GNRHR-related	AR
GNS	Mucopolysaccharidosis IIID (Sanfilippo syndrome D)	AR
GORAB	Geroderma osteodysplasticum	AR
GRHPR	Primary hyperoxaluria type II	AR
GRIP1	Fraser syndrome	AR
GSS	Glutathione synthetase deficiency	AR
GUCY2D	Leber congenital amaurosis 1	AR
GUSB	Mucopolysaccharidosis type VII	AR
GYS2	Glycogen storage disease, type 0, liver	AR
HADH	Familial hyperinsulinemic hypoglycemia 4	AR
HADHA	Trifunctional protein deficiency	AR
HADHB	Trifunctional protein deficiency	AR
HAMP	Hemochromatosis, type 2B	AR
HAX1	Severe congenital neutropenia, HAX1-related	AR
HBA1	Alpha thalassemia	AR
HBB	Sickle cell disease	AR
HELLS	Immunodeficiency, Centromeric region instability, Facial anomalies syndrome	AR
HEXA	Tay-Sachs disease	AR
HEXB	Sandhoff disease	AR
HGSNAT	Mucopolysaccharidosis type IIIC (Sanfilippo syndrome C)	AR
HINT1	Neuromyotonia and axonal neuropathy	AR
HJV	Hemochromatosis, type 2A	AR
HLCS	Holocarboxylase synthetase deficiency	AR

AR: autosomal recessive XL: X-linked

GENE	CONDITION	INHERITANCE
HMGCL	3-hydroxy-3-methylglutaryl-CoA lyase deficiency	AR
HMGCS2	3-hydroxy-3-methylglutaryl-CoA synthase 2 deficiency	AR
HOGA1	Primary hyperoxaluria type III	AR
HPD	Tyrosinemia type III	AR
HPS1	Hermansky-Pudlak syndrome 1	AR
HPS3	Hermansky-Pudlak syndrome 3	AR
HPS4	Hermansky-Pudlak syndrome 4	AR
HPS5	Hermansky-Pudlak syndrome 5	AR
HPS6	Hermansky-Pudlak syndrome 6	AR
HSD17B3	17-Beta-Hydroxysteroid Dehydrogenase Deficiency	AR
HSD17B4	D-bifunctional protein deficiency	AR
HSD3B2	Congenital adrenal hyperplasia due to 3-beta-hydroxysteroid dehydrogenase 2 deficiency	AR
HSD3B7	Congenital bile acid synthesis defect 1	AR
HYAL1	Mucopolysaccharidosis type IX	AR
HYLS1	Hydrolethalus syndrome	AR
IDH3B	Retinitis pigmentosa, IDH3B-related	AR
IDUA	Mucopolysaccharidosis, type I (Hurler syndrome)	AR
IFT140	IFT140-related disorders	AR
IGHMBP2	IGHMBP2-related neuropathies	AR
IKBKB	Immunodeficiency 15B	AR
IL2RA	Immunodeficiency due to CD25 deficiency	AR
IL7R	Severe Combined Immunodeficiency 104	AR
INPP5E	Joubert syndrome 1	AR
INVS	Nephronophthisis 2	AR
ITGA2B	Glanzmann thrombasthenia	AR
ITGA6	Junctional epidermolysis bullosa	AR
ITGB3	Glanzmann thrombasthenia	AR
ITGB4	Junctional epidermolysis bullosa	AR
ITPA	Developmental and epileptic encephalopathy 35	AR
IVD	Isovaleric Acidemia	AR
IYD	Thyroid dyshormonogenesis, IYD-related	AR
JAK3	Severe combined immunodeficiency, JAK3-related	AR
KCNJ1	Bartter syndrome	AR
KCNJ11	KCNJ11-related hyperinsulinism	AR
KCTD7	Progressive myoclonic epilepsy type 3	AR
KIF14	Primary Autosomal Recessive Microcephaly 20	AR

GENE	CONDITION	INHERITANCE
LAMA2	Muscular dystrophy, LAMA2-related	AR
LAMA3	Junctional epidermolysis bullosa 2	AR
LAMB3	Junctional epidermolysis bullosa, LAMB3-related	AR
LAMC2	Junctional epidermolysis bullosa, LAMC2-related	AR
LARS	Infantile liver failure syndrome 1	AR
LCA5	Leber congenital amaurosis 5	AR
LCK	Immunodeficiency 22	AR
LDLR	Familial Hypercholesterolemia	AR
LDLRAP1	Familial Hypercholesterolemia	AR
LHCGR	Leydig cell hypoplasia	AR
LHX3	Combined pituitary hormone deficiency 3	AR
LIFR	Stuve-Wiedemann syndrome	AR
LIG4	LIG4 syndrome	AR
LIPA	Lysosomal acid lipase deficiency	AR
LIPN	Congenital Ichthyosis 8	AR
LMAN1	Combined factor V and VIII deficiency	AR
LMBRD1	Methylmalonic aciduria and homocystinuria, cblF type	AR
LOXHD1	Nonsyndromic hearing loss 77	AR
LPAR6	Hypotrichosis 8	AR
LPL	Familial lipoprotein lipase deficiency	AR
LRAT	Leber congenital amaurosis 14	AR
LRP2	Donnaiâ€"Barrow syndrome	AR
LRPPRC	Leigh syndrome with Complex IV deficiency	AR
LTBP4	Cutis laxa with severe pulmonary, gastrointestinal, and urinary abnormalities	AR
LYST	Chediak-Higashi syndrome	AR
MAK	Retinitis Pigmentosa 62	AR
MALT1	Immunodeficiency 12	AR
MAN2B1	Alpha-Mannosidosis	AR
MANBA	Beta-Mannosidosis	AR
MAT1A	Methionine adenosyltransferase deficiency	AR
MCEE	Methylmalonyl-CoA epimerase deficiency	AR
MCOLN1	Mucolipidosis IV	AR
MCPH1	Primary microcephaly 1, recessive	AR
MED17	Postnatal Progressive Microcephaly with Seizures and Brain Atrophy	AR
MEGF8	Carpenter syndrome 2	AR
MESP2	Spondylocostal dysostosis	AR
MFSD8	Neuronal ceroid lipofuscinosis, MFSD8-related	AR

GENE	CONDITION	INHERITANCE
MKKS	Bardet-Biedl syndrome 6	AR
MKS1	MKS1-related ciliopathies	AR
MLC1	Megalencephalic leukoencephalopathy with subcortical cysts	AR
MLYCD	Malonyl-CoA decarboxylase deficiency	AR
MMAA	Methylmalonic aciduria, cblA type	AR
MMAB	Methylmalonic aciduria, cblB type	AR
MMACHC	Methylmalonic aciduria and homocystinuria, cblC type	AR
MMADHC	Methylmalonic aciduria and homocystinuria, cblD type	AR
MPI	Congenital disorder of glycosylation type Ib	AR
MPL	Congenital amegakaryocytic thrombocytopenia	AR
MPV17	Hepatocerebral mitochondrial DNA depletion syndrome, MPV17-related	AR
MRE11	Ataxia-Telangiectasia-Like Disorder 1	AR
MTHFD1	Combined immunodeficiency and megaloblastic anemia	AR
MTHFR	Homocystinuria, MTHFR-related	AR
MTMR2	Charcot-Marie-Tooth disease, type 4B1	AR
MTR	Methylcobalamin deficiency, type cblG	AR
MTRR	Homocystinuria-megaloblastic anemia, cobalamin E type	AR
MTTP	Abetalipoproteinemia	AR
MUT	Methylmalonic aciduria, methylmalonyl-CoA mutase deficiency	AR
MVK	Mevalonate kinase deficiency	AR
MYO15A	Nonsyndromic hearing loss, MYO15A-related	AR
MYO7A	MYO7A-related disorders	AR
NAGA	Schindler disease types 1 and 3	AR
NAGLU	Mucopolysaccharidosis type IIIB (Sanfilippo syndrome B)	AR
NAGS	N-acetylglutamate synthase deficiency	AR
NBAS	SOPH syndrome	AR
NBEAL2	Gray platelet syndrome	AR
NBN	Nijmegen breakage syndrome	AR
NCF2	Chronic granulomatous disease 2	AR
NCF4	Chronic granulomatous disease 4	AR
NDRG1	Charcot-Marie-Tooth disease, type 4D	AR
NDUFA11	Mitochondrial complex I deficiency	AR
NDUFAF2	Mitochondrial complex I deficiency	AR
NDUFAF5	Mitochondrial complex I deficiency (Leigh syndrome)	AR
NDUFS4	Mitochondrial complex I deficiency	AR
NDUFS6	Mitochondrial complex I deficiency (Leigh syndrome)	AR
NDUFS7	Mitochondrial complex I deficiency	AR

AR: autosomal recessive XL: X-linked

GENE	CONDITION	INHERITANCE
NDUFV1	Mitochondrial complex I deficiency, nuclear type 4	AR
NEB	Nemaline myopathy	AR
NEU1	Sialidosis, type I and II	AR
NGLY1	Congenital disorder of deglycosylation	AR
NHEJ1	Severe combined immunodeficiency with microcephaly, growth retardation, and sensitivity to ionizing radiation	AR
NIPAL4	Autosomal Recessive Congenital Ichthyosis 6	AR
NPC1	Niemann-Pick disease, type C1	AR
NPC2	Niemann-Pick disease, type C2	AR
NPHP1	NPHP1-related ciliopathies	AR
NPHP3	NPHP3-related ciliopathies	AR
NPHS1	Congenital nephrotic syndrome, type 1	AR
NPHS2	Congenital nephrotic syndrome, type 2	AR
NR2E3	NR2E3-related retinal dystrophies	AR
NTRK1	Congenital insensitivity to pain with anhidrosis	AR
OAT	Gyrate atrophy of choroid and retina	AR
OBSL1	3M syndrome 2	AR
OCA2	Oculocutaneous albinism type II	AR
OPA3	Costeff syndrome	AR
OSTM1	Osteopetrosis 5	AR
OTOF	Nonsyndromic hearing loss, OTOF-related	AR
P3H1	Osteogenesis imperfecta, type VIII	AR
PAH	Phenylalanine Hydroxylase deficiency (Phenylketonuria)	AR
PANK2	Pantothenate kinase-associated neurodegeneration	AR
PC	Pyruvate carboxylase deficiency	AR
PCBD1	Tetrahydrobiopterin deficiency, PCBD1-related	AR
PCCA	Propionic acidemia, PCCA-related	AR
PCCB	Propionic acidemia, PCCB-related	AR
PCDH15	PCDH15-related sensory loss	AR
PCNT	Microcephalic osteodysplastic primordial dwarfism, type II	AR
PDE6A	Retinitis pigmentosa, PDE6A-related	AR
PDHB	Pyruvate dehydrogenase E1-beta deficiency	AR
PDHX	Pyruvate dehydrogenase E3-binding protein deficiency	AR
PDP1	Pyruvate dehydrogenase phosphatase deficiency	AR
PEPD	Prolidase deficiency	AR
PET100	Mitochondrial complex IV deficiency	AR
PEX1	Zellweger syndrome, PEX1-related	AR

GENE	CONDITION	INHERITANCE
PEX10	Zellweger syndrome, PEX10-related	AR
PEX11B	Zellweger spectrum disorder	AR
PEX12	Zellweger syndrome, PEX12-related	AR
PEX13	Zellweger spectrum disorder	AR
PEX14	Zellweger spectrum disorder	AR
PEX16	Zellweger spectrum disorder	AR
PEX19	Zellweger spectrum disorder	AR
PEX2	Zellweger syndrome, PEX2-related	AR
PEX26	Zellweger syndrome	AR
PEX3	Zellweger spectrum disorder	AR
PEX5	Zellweger spectrum disorder	AR
PEX6	Zellweger syndrome, PEX6-related	AR
PEX7	Rhizomelic chondrodysplasia punctata, type 1	AR
PFKM	Glycogen storage disease VII	AR
PGM3	Immunodeficiency 23	AR
PHGDH	Phosphoglycerate dehydrogenase deficiency	AR
PHKB	Glycogen storage disease type IXb	AR
PHKG2	Glycogen storage disease type IXc	AR
PHYH	Refsum disease	AR
PIGN	Multiple congenital anomalies hypotonia seizures syndrome 1	AR
PIP5K1C	Lethal congenital contractural syndrome 3	AR
PJVK	Nonsyndromic hearing loss 59	AR
PKHD1	Polycystic kidney disease, PKHD1-related	AR
PLA2G6	Infantile neuroaxonal dystrophy	AR
PLEKHG5	PLEKHG5-related motor neuropathies	AR
PLOD1	Ehlers-Danlos syndrome with kyphoscoliosis, PLOD1-related	AR
PLOD2	Bruck syndrome 2	AR
PMM2	Congenital disorder of glycosylation type 1a	AR
PNP	Purine nucleoside phosphorylase deficiency	AR
PNPLA1	Autosomal recessive congenital ichthyosis 10	AR
PNPO	Pyridoxamine 5'-phosphate oxidase deficiency	AR
POC1A	Short stature, onychodysplasia, facial dysmorphism, and hypotrichosis syndrome	AR
POLG	POLG-related disorders	AR
POLH	Xeroderma pigmentosum	AR
POLR1C	POLR1C-related disorders	AR
POMGNT1	POMGNT1 Alpha-dystroglycanopathies	AR
POMT1	POMT1 Alpha-dystroglycanopathies	AR

AR: autosomal recessive XL: X-linked

GENE	CONDITION	INHERITANCE
POMT2	POMT2 Alpha-dystroglycanopathies	AR
POR	Antley-Bixler syndrome	AR
POU1F1	Combined pituitary hormone deficiency	AR
PPIB	Osteogenesis imperfecta, type IX	AR
PPT1	Neuronal ceroid lipofuscinosis, PPT1-related	AR
PRCD	Retinitis pigmentosa 36	AR
PRDM5	Brittle cornea syndrome 2	AR
PRF1	Hemophagocytic lymphohistiocytosis, familial, 2	AR
PRICKLE1	Progressive myoclonic epilepsy, type 1B	AR
PRKDC	PRKDC-related immunodeficiency	AR
PROP1	Combined pituitary hormone deficiency 2	AR
PSAP	Metachromatic leukodystrophy due to saposin-b deficiency	AR
PTPRC	PTPRC related-severe combined immunodeficiency	AR
PTS	Tetrahydrobiopterin deficiency	AR
PUS1	Mitochondrial myopathy and sideroblastic anemia 1	AR
PYCR1	Cutis laxa type IIB and type IIIB	AR
PYGL	Glycogen storage disease VI	AR
PYGM	Glycogen storage disease type V	AR
QDPR	Tetrahydrobiopterin deficiency, QDPR-related	AR
RAB23	Carpenter syndrome	AR
RAG1	Omenn syndrome, RAG1-related	AR
RAG2	Omenn syndrome, RAG2-related	AR
RAPSN	RAPSN-associated acetylcholine receptor deficiency	AR
RARS2	Pontocerebellar hypoplasia type 6	AR
RAX	Microphthalmia, isolated 3	AR
RD3	Leber congenital amaurosis 12	AR
RDH12	Leber congenital amaurosis type 13	AR
RDH5	Fundus albipunctatus	AR
RFX5	Bare lymphocyte syndrome type II	AR
RFXANK	MHC class II deficiency	AR
RFXAP	Bare lymphocyte syndrome type II	AR
RHAG	Rh Deficiency syndrome	AR
RLBP1	Retinal dystrophy, RLBP1-related	AR
RMRP	Cartilage-Hair Hypoplasia Anauxetic Dysplasia Spectrum Disorder	AR
RNASEH2A	Aicardi-Goutieres syndrome 4	AR
RNASEH2B	Aicardi Goutieres syndrome 2	AR
RNASEH2C	Aicardi-Goutieres syndrome 3	AR

AR: autosomal recessive XL: X-linked

GENE	CONDITION	INHERITANCE
ROGDI	Kohlschutter-Tonz syndrome	AR
RPE65	RPE65-related retinopathy	AR
RPGRIP1	Leber congenital amaurosis and Cone-rod dystrophy	AR
RPGRIP1L	RPGRIP1L-related ciliopathies	AR
RSPH9	Primary ciliary dyskinesia 12	AR
RTEL1	Dyskeratosis congenita type 5	AR
SACS	Autosomal recessive spastic ataxia of Charlevoix-Saguenay	AR
SAG	Retinitis pigmentosa 47	AR
SAMD9	Normophosphatemic Familial Tumoral Calcinosis	AR
SAMHD1	Aicardi-Goutieres syndrome	AR
SARS2	Hyperuricemia, pulmonary hypertension, renal failure, and alkalosis syndrome	AR
SBDS	Shwachman-Diamond syndrome	AR
SCO1	Mitochondrial complex IV deficiency	AR
SCO2	Mitochondrial complex IV deficiency	AR
SDCCAG8	Bardet-Biedl syndrome and Senior-Loken syndrome	AR
SDR9C7	Autosomal recessive congenital ichthyosis	AR
SEC23B	Congenital dyserythropoietic anemia, type II	AR
SELENON	Rigid spine muscular dystrophy	AR
SEPSECS	Pontocerebellar hypoplasia type 2D	AR
SERPINF1	Osteogenesis imperfecta, type VI	AR
SGCA	Limb-girdle muscular dystrophy, type 2D	AR
SGCB	Limb-girdle muscular dystrophy, type 2E	AR
SGCD	Limb-girdle muscular dystrophy, type 2F	AR
SGCG	Limb-girdle muscular dystrophy, type 2C	AR
SGSH	Mucopolysaccharidosis IIIA (Sanfilippo syndrome A)	AR
SH3TC2	Charcot-Marie-Tooth disease, SH3TC2-related	AR
SKIV2L	Trichohepatoenteric syndrome 2	AR
SLC12A1	Bartter syndrome	AR
SLC12A3	Gitelman syndrome	AR
SLC12A6	Andermann syndrome	AR
SLC17A5	Sialic acid storage disorder	AR
SLC19A2	Thiamine-responsive megaloblastic anemia syndrome	AR
SLC19A3	Biotin-responsive basal ganglia disease	AR
SLC1A4	Spastic tetraplegia, thin corpus callosum, and progressive microcephaly syndrome	AR
SLC22A5	Systemic primary carnitine deficiency	AR
SLC25A13	Citrin deficiency	AR
SLC25A15	Hyperornithinemia-hyperammonemia-homocitrullinemia syndrome (Triple H syndrome)	AR

AR: autosomal recessive XL: X-linked

GENE	CONDITION	INHERITANCE
SLC25A20	Carnitine-acylcarnitine translocase deficiency	AR
SLC26A2	SLC26A2-related disorders	AR
SLC26A3	Congenital secretory chloride diarrhea	AR
SLC26A4	Pendred syndrome	AR
SLC27A4	Ichthyosis prematurity syndrome	AR
SLC2A10	Arterial tortuosity syndrome	AR
SLC2A2	Fanconi-Bickel syndrome	AR
SLC34A3	Hereditary hypophosphatemic rickets with hypercalciuria	AR
SLC35A3	Arthrogryposis, intellectual disability, and seizures	AR
SLC37A4	Glycogen storage disease, type Ib	AR
SLC39A4	Acrodermatitis enteropathica	AR
SLC3A1	Cystinuria, type I	AR
SLC45A2	Oculocutaneous albinism, type IV	AR
SLC46A1	Hereditary folate malabsorption	AR
SLC4A1	Distal Renal Tubular Acidosis	AR
SLC4A11	Corneal endothelial dystrophy	AR
SLC5A5	Thyroid dyshormonogenesis, SLC5A5-related	AR
SLC6A19	Hartnup disorder	AR
SLC7A7	Lysinuric protein intolerance	AR
SLC7A9	Cystinuria, non-type I	AR
SMARCAL1	Schimke immunoosseous dysplasia	AR
SMN1	Spinal muscular atrophy	AR
SMPD1	Niemann-Pick disease, type A/B	AR
SNAP29	Cerebral dysgenesis, neuropathy, ichthyosis, and palmoplantar keratoderma syndrome	AR
SNX10	Osteopetrosis 8	AR
SP110	Hepatic venoocclusive disease with immunodeficiency	AR
SPATA7	Leber congenital amaurosis (LCA) and juvenile retinitis pigmentosa (RP)	AR
SPG11	SPG11-related Neuromuscular Disorders	AR
SPG21	Mast syndrome	AR
SPG7	Spastic paraplegia type 7	AR
SPINK5	Netherton syndrome	AR
SPR	Sepiapterin Reductase Deficiency	AR
SRD5A2	5-alpha reductase deficiency	AR
ST3GAL5	Salt and pepper developmental regression syndrome	AR
STAR	Lipoid congenital adrenal hyperplasia	AR
STK4	Combined immunodeficiency due to STK4 deficiency	AR
STX11	Familial hemophagocytic lymphohistiocytosis	AR

AR: autosomal recessive XL: X-linked

GENE	CONDITION	INHERITANCE
STXBP2	Familial hemophagocytic lymphohistiocytosis	AR
SUCLA2	Mitochondrial DNA depletion syndrome 5	AR
SUMF1	Multiple sulfatase deficiency	AR
SUOX	Sulfite oxidase deficiency	AR
SURF1	Leigh syndrome, SURF1-related	AR
SYNE4	Autosomal recessive deafness 76	AR
TAT	Tyrosinemia, type II	AR
TBCE	Hypoparathyroidism-retardation-dysmorphism syndrome	AR
TBX19	Adrenocorticotropic hormone deficiency	AR
TCIRG1	Osteopetrosis 1	AR
TCTN1	Joubert syndrome 13	AR
TCTN2	TCTN2-related ciliopathies	AR
TCTN3	Joubert syndrome 18	AR
TECPR2	Spastic paraplegia 49	AR
TERT	Dyskeratosis congenita type 4	AR
TF	Atransferrinemia	AR
TFR2	Hemochromatosis, type 3	AR
TG	Thyroid dyshormonogenesis, TG-related	AR
TGM1	Congenital ichthyosis	AR
TH	Segawa syndrome	AR
TK2	Mitochondrial DNA depletion syndrome 2	AR
TMC1	Nonsyndromic hearing loss 7	AR
TMEM138	Joubert syndrome 16	AR
TMEM216	TMEM216-related ciliopathies	AR
TMEM231	Joubert syndrome 20	AR
TMEM237	Joubert syndrome 14	AR
TMEM38B	Osteogenesis imperfecta, type XIV	AR
TMEM67	COACH syndrome	AR
TMEM70	Mitochondrial complex V deficiency type 2	AR
TMPRSS3	Nonsyndromic hearing loss, TMPRSS3-related	AR
TNFSF11	Osteopetrosis 2	AR
TNXB	Ehlers-Danlos-like syndrome due to tenascin-X deficiency	AR
TPO	Thyroid dyshormonogenesis, TPO-related	AR
TPP1	Neuronal ceroid lipofuscinosis, TPP1-related	AR
TRAPPC11	Limb-girdle muscular dystrophy 18	AR
TRDN	Catecholaminergic polymorphic ventricular tachycardia	AR
TREX1	Aicardi-Goutieres syndrome 1	AR

AR: autosomal recessive XL: X-linked

GENE	CONDITION	INHERITANCE
TRHR	Generalized thyrotropin-releasing hormone resistance	AR
TRIM32	TRIM32-related disorders	AR
TRIM37	Mulibrey nanism	AR
TRMU	Liver failure, acute infantile	AR
TRPM6	Hypomagnesemia 1	AR
TSEN2	Pontocerebellar hypoplasia type 2B	AR
TSEN34	Pontocerebellar hypoplasia type 2C	AR
TSEN54	Pontocerebellar hypoplasia type 2A	AR
TSFM	Combined oxidative phosphorylation deficiency, TSFM-related	AR
TSHB	Congenital hypothyroidism, TSHB-related	AR
TSHR	Congenital hypothyroidism, TSHR-related	AR
TTC37	Trichohepatoenteric syndrome	AR
TTC7A	Gastrointestinal defects and immunodeficiency syndrome	AR
TTC8	Bardet-Biedl syndrome 8	AR
TTPA	Ataxia with isolated vitamin E deficiency	AR
TULP1	TULP1-related retinal disorders	AR
TYMP	Mitochondrial neurogastrointestinal encephalopathy (MNGIE) disease	AR
TYR	Oculocutaneous albinism types 1A and 1B	AR
TYRP1	Oculocutaneous albinism, type III	AR
UGT1A1	Crigler-Najjar syndrome	AR
UNC13D	Familial hemophagocytic lymphohistiocytosis type 3	AR
USH1C	USH1C-related disorders	AR
USH1G	Usher syndrome type IG	AR
USH2A	Usher syndrome, type 2A	AR
VDR	Vitamin D-dependent rickets, type 2A	AR
VLDLR	Cerebellar ataxia, mental retardation, and dysequilibrium syndrome 1	AR
VPS13A	Choreoacanthocytosis	AR
VPS13B	Cohen syndrome	AR
VPS45	Severe congenital neutropenia, VPS45-related	AR
VPS53	Pontocerebellar hypoplasia type 2E	AR
VRK1	Pontocerebellar hypoplasia type 1A	AR
VSX2	Microphthalmia with or without coloboma	AR
WHRN	Usher syndrome type 2D	AR
WISP3	Progressive pseudorheumatoid dysplasia	AR
WNT1	Osteogenesis imperfecta type 15	AR
WNT10A	WNT10A-related ectodermal dysplasias	AR
WRN	Werner syndrome	AR

AR: autosomal recessive XL: X-linked

GENE	CONDITION	INHERITANCE
XPA	Xeroderma pigmentosum, group A	AR
XPC	Xeroderma pigmentosum, group C	AR
ZAP70	ZAP70-related Immunodeficiency	AR
ZBTB24	Immunodeficiency-centromeric instability-facial anomalies syndrome 2	AR
ZFYVE26	Spastic paraplegia 15	AR
ZNF469	Brittle cornea syndrome 1	AR
HBA2	Alpha thalassemia	AR

AR: autosomal recessive XL: X-linked