VirtusGenetics
A MEMBER OF VIRTUS HEALTH

Expanded carrier screen in males

268 genes (without X-linked disorders)

CLICK HERE TO VIEW BY GENE CLICK HERE TO VIEW OPTIONAL ADD-ON GENES

ORDERED BY DISEASE	GENE	INHERITANCE
3-hydroxy-3-methylglutarayl-CoA (HMG-CoA) lyase deficiency	HMGCL	AR
ABCC8-related disorders	ABCC8	AR
Abetalipoproteinemia	MTTP	AR
ACAD9 deficiency	ACAD9	AR
Achromatopsia (CNGB3-related)	CNGB3	AR
Acrodermatitis enteropathica	SLC39A4	AR
Adenosine deaminase deficiency	ADA	AR
Aicardi-Goutieres syndrome (SAMHD1-related)	SAMHD1	AR
Aldosterone synthase deficiency	CYP11B2	AR
Alpha-mannosidosis	MAN2B1	AR
Alpha-thalassemia	HBA1/HBA2	AR
Alport Syndrome (COL4A3-related)	COL4A3	AR
Alport Syndrome (COL4A4-related)	COL4A4	AR
Alström syndrome	ALMS1	AR
Andermann syndrome	SLC12A6	AR
Arginase deficiency	ARG1	AR
Argininosuccinic aciduria	ASL	AR
Aromatase deficiency	CYP19A1	AR
Asparagine synthetase deficiency	ASNS	AR
Aspartylglucosaminuria	AGA	AR
Ataxia with vitamin E deficiency	TTPA	AR
Ataxia-telangiectasia	ATM	AR
Autoimmune polyendocrinopathy with candidiasis and ectodermal dysplasia	AIRE	AR
Autosomal recessive deafness 77	LOXHD1	AR
Autosomal recessive spastic ataxia of Charlevoix-Saguenay (ARSACS)	SACS	AR
Bardet-Biedl syndrome (BBS10-related)	BBS10	AR
Bardet-Biedl syndrome (BBS12-related)	BBS12	AR
Bartter syndrome type 4A	BSND	AR
BBS1-related disorders	BBS1	AR
BBS2-related disorders	BBS2	AR
Beta-ketothiolase deficiency	ACAT1	AR
Bloom syndrome	BLM	AR
Canavan disease	ASPA	AR

ORDERED BY DISEASE	GENE	INHERITANCE
Carbamoylphosphate synthetase I deficiency	CPS1	AR
Carnitine palmitoyltransferase I deficiency	CPT1A	AR
Carnitine palmitoyltransferase II deficiency	CPT2	AR
Carpenter syndrome (RAB23-related)	RAB23	AR
Cartilage-hair hypoplasia-anauxetic dysplasia spectrum disorders	RMRP	AR
Cerebrotendinous xanthomatosis	CYP27A1	AR
CFTR-related disorders (including cystic fibrosis)	CFTR	AR
Charcot-Marie-Tooth disease (NDRG1-related)	NDRG1	AR
Chorea-acanthocytosis	VPS13A	AR
Chronic granulomatous disease (CYBA-related)	CYBA	AR
Citrin deficiency	SLC25A13	AR
Citrullinemia type 1	ASS1	AR
Cockayne syndrome type A	ERCC8	AR
Cockayne syndrome type B	ERCC6	AR
Cohen syndrome	VPS13B	AR
Combined malonic and methylmalonic aciduria (ACSF3-related)	ACSF3	AR
Combined oxidative phosphorylation deficiency (GFM1-related)	GFM1	AR
Combined oxidative phosphorylation deficiency (TSFM-related)	TSFM	AR
Combined pituitary hormone deficiency (LHX3-related)	LHX3	AR
Combined pituitary hormone deficiency (PROP1-related)	PROP1	AR
Congenital adrenal hyperplasia due to 11-beta-hydroxylase-deficiency	CYP11B1	AR
Congenital adrenal hyperplasia due to 21-hydroxylase deficiency	CYP21A2	AR
Congenital adrenal hyperplasia due to 3-beta-hydroxysteroid dehydrogenase type II deficiency	HSD3B2	AR
Congenital amegakaryocytic thrombocytopenia	MPL	AR
Congenital disorder of glycosylation (ALG6-related)	ALG6	AR
Congenital disorder of glycosylation (MPI-related)	MPI	AR
Congenital disorder of glycosylation (PMM2-related)	PMM2	AR
Congenital ichthyosis (TGM1-related)	TGM1	AR
Congenital insensitivity to pain with anhidrosis	NTRK1	AR
Congenital myasthenic syndrome (CHRNE-related)	CHRNE	AR
Corneal dystrophy and perceptive deafness	SLC4A11	AR
CYP17A1-related disorders	CYP17A1	AR
Cystinosis	CTNS	AR
DHDDS-related disorders	DHDDS	AR
Dihydrolipoamide dehydrogenase deficiency (DLD)	DLD	AR
Dysferlinopathy	DYSF	AR
Dystrophic epidermolysis bullosa (COL7A1-related)	COL7A1	AR
Ehlers-Danlos syndrome, dermatosparaxis type	ADAMTS2	AR
Ellis-van Creveld syndrome (EVC-related)	EVC	AR
Ellis-van Creveld syndrome (EVC2-related)	EVC2	AR

ORDERED BY DISEASE	GENE	INHERITANCE
Enhanced S-cone syndrome/ retinitis pigmentosa 37	NR2E3	AR
Ethylmalonic encephalopathy	ETHE1	AR
Familial dysautonomia	ELP1	AR
Familial hypercholesterolemia (LDLR-related)	LDLR	AD
Familial hypercholesterolemia (LDLRAP1-related)	LDLRAP1	AR
Fanconi anemia type A	FANCA	AR
Fanconi anemia type C	FANCC	AR
Fanconi anemia type G	FANCG	AR
FKRP-related disorders	FKRP	AR
FKTN-related disorders	FKTN	AR
Fumarate hydratase deficiency	FH	AR
Galactokinase deficiency galactosemia	GALK1	AR
Galactosemia (GALT-related)	GALT	AR
Gaucher disease	GBA	AR
Gitelman syndrome (SLC12A3-related)	SLC12A3	AR
GJB2-related DFNB1 nonsyndromic hearing loss and deafness	GJB2	AR
GLE1-related disorders	GLE1	AR
Glutaric acidemia type I	GCDH	AR
Glutaric acidemia type IIA	ETFA	AR
Glutaric acidemia type IIC	ETFDH	AR
Glycine encephalopathy (AMT-related)	AMT	AR
Glycine encephalopathy (GLDC-related)	GLDC	AR
Glycogen storage disease type Ia	G6PC	AR
Glycogen storage disease type Ib	SLC37A4	AR
Glycogen storage disease type II (Pompe disease)	GAA	AR
Glycogen storage disease type III	AGL	AR
Glycogen storage disease type IV/ adult polyglucosan body disease	GBE1	AR
Glycogen storage disease type V	PYGM	AR
Glycogen storage disease type VII	PFKM	AR
GRACILE syndrome/ BCS1L-related disorders	BCS1L	AR
Guanidinoacetate methyltransferase deficiency	GAMT	AR
HBB-related hemoglobinopathies	HBB	AR
Hereditary fructose intolerance	ALDOB	AR
Hereditary hemochromatosis type 2 (HJV-related)	HJV	AR
Hereditary hemochromatosis type 3	TFR2	AR
Hermansky-Pudlak syndrome type 1	HPS1	AR
Hermansky-Pudlak syndrome type 3	HPS3	AR
Holocarboxylase synthetase deficiency	HLCS	AR
Homocystinuria due to CBS deficiency	CBS	AR
Homocystinuria due to MTHFR deficiency	MTHFR	AR
Homocystinuria, cobalamin E type	MTRR	AR

ORDERED BY DISEASE	GENE	INHERITANCE
HSD17B4-related disorders	HSD17B4	AR
Hydrolethalus syndrome type 1	HYLS1	AR
Hyperornithinemia-hyperammonemia-homocitrullinuria (HHH) syndrome	SLC25A15	AR
Hypophosphatasia	ALPL	AR
Inclusion body myopathy 2	GNE	AR
Isovaleric acidemia	IVD	AR
Joubert syndrome 2/ TMEM216-related disorders	TMEM216	AR
Junctional epidermolysis bullosa (LAMB3-related)	LAMB3	AR
Junctional epidermolysis bullosa (LAMC2-related)	LAMC2	AR
KCNJ11-related disorders	KCNJ11	AR
Krabbe disease	GALC	AR
LAMA2-related muscular dystrophy	LAMA2	AR
LAMA3-related disorders	LAMA3	AR
Leber congenital amaurosis 10/ CEP290-related disorders	CEP290	AR
Leber congenital amaurosis 13	RDH12	AR
Leber congenital amaurosis 5	LCA5	AR
Leber congenital amaurosis 8/ CRB1-related disorders	CRB1	AR
Leigh syndrome, French Canadian type	LRPPRC	AR
Leukoencephalopathy with vanishing white matter (EIF2B5-related)	EIF2B5	AR
Limb-girdle muscular dystrophy type 2A (calpainopathy)	CAPN3	AR
Limb-girdle muscular dystrophy type 2C	SGCG	AR
Limb-girdle muscular dystrophy type 2D	SGCA	AR
Limb-girdle muscular dystrophy type 2E	SGCB	AR
Lipoid congenital adrenal hyperplasia	STAR	AR
Lipoprotein lipase deficiency	LPL	AR
Long chain 3-hydroxyacyl-CoA dehydrogenase (LCHAD) deficiency	HADHA	AR
Lysinuric protein intolerance	SLC7A7	AR
Lysosomal acid lipase deficiency	LIPA	AR
Major histocompatibility complex class II deficiency (CIITA-related)	CIITA	AR
Maple syrup urine disease (MSUD) type 1A	BCKDHA	AR
Maple syrup urine disease (MSUD) type 1B	BCKDHB	AR
Maple syrup urine disease (MSUD) type 2	DBT	AR
Medium chain acyl-CoA dehydrogenase (MCAD) deficiency	ACADM	AR
Megalencephalic leukoencephalopathy with subcortical cysts type 1	MLC1	AR
Metachromatic leukodystrophy (ARSA-related)	ARSA	AR
Methylmalonic acidemia (MMAA-related)	MMAA	AR
Methylmalonic acidemia (MMAB-related)	MMAB	AR
Methylmalonic acidemia (MUT-related)	MUT	AR
Methylmalonic acidemia with homocystinuria, cobalamin C type	MMACHC	AR
Methylmalonic acidemia with homocystinuria, cobalamin D type	MMADHC	AR
Microphthalmia /clinical anophthalmia (VSX2-related)	VSX2	AR

ORDERED BY DISEASE	GENE	INHERITANCE
Mitochondrial complex I deficiency/ Leigh syndrome (NDUFAF5-related)	NDUFAF5	AR
Mitochondrial complex I deficiency/ Leigh syndrome (NDUFS6- related)	NDUFS6	AR
Mitochondrial DNA depletion syndrome (MPV17-related)	MPV17	AR
Mitochondrial myopathy and sideroblastic anemia 1	PUS1	AR
Mitochondrial neurogastrointestinal encephalopathy (MNGIE) disease	TYMP	AR
MKS1-related disorders	MKS1	AR
Mucolipidosis type II/III (GNPTAB-related)	GNPTAB	AR
Mucolipidosis type III (GNPTG-related)	GNPTG	AR
Mucolipidosis type IV	MCOLN1	AR
Mucopolysaccharidosis type I	IDUA	AR
Mucopolysaccharidosis type IIIA (Sanfilippo A syndrome)	SGSH	AR
Mucopolysaccharidosis type IIIB (Sanfilippo B syndrome)	NAGLU	AR
Mucopolysaccharidosis type IIIC (Sanfilippo C syndrome)/ retinitis pigmentosa 73	HGSNAT	AR
Mucopolysaccharidosis type IIID (Sanfilippo D syndrome)	GNS	AR
Mucopolysaccharidosis type IVB (Morquio B syndrome)/ GM1 gangliosidosis	GLB1	AR
Mucopolysaccharidosis type IX	HYAL1	AR
Mucopolysaccharidosis type VI (Maroteaux-Lamy syndrome)	ARSB	AR
Multiple sulfatase deficiency	SUMF1	AR
N-Acetylglutamate synthase deficiency	NAGS	AR
Nemaline myopathy 2	NEB	AR
Nephrogenic diabetes insipidus (AQP2-related)	AQP2	AR
Nephrotic syndrome/ congenital Finnish nephrosis (NPHS1-related)	NPHS1	AR
Nephrotic syndrome/steroid-resistant nephrotic syndrome (NPHS2-related)	NPHS2	AR
Neuronal ceroid lipofuscinosis (TPP1-related)	TPP1	AR
Neuronal ceroid-lipofuscinosis (CLN3-related)	CLN3	AR
Neuronal ceroid-lipofuscinosis (CLN5-related)	CLN5	AR
Neuronal ceroid-lipofuscinosis (CLN6-related)	CLN6	AR
Neuronal ceroid-lipofuscinosis (MFSD8-related)	MFSD8	AR
Neuronal ceroid-lipofuscinosis (PPT1-related)	PPT1	AR
Neuronal ceroid-lipofuscinosis/ Northern epilepsy (CLN8-related)	CLN8	AR
Niemann-Pick disease type A/B	SMPD1	AR
Niemann-Pick disease type C (NPC1-related)	NPC1	AR
Niemann-Pick disease type C (NPC2-related)	NPC2	AR
Nijmegen breakage syndrome	NBN	AR
OPA3-related conditions	OPA3	AR
Ornithine aminotransferase deficiency	OAT	AR
Osteopetrosis (TCIRG1-related)	TCIRG1	AR
Pendred syndrome	SLC26A4	AR
Peroxisomal acyl-CoA oxidase deficiency	ACOX1	AR
Phenylalanine hydroxylase deficiency (including Phenylketonuria (PKU))	PAH	AR
Phosphoglycerate dehydrogenase deficiency/ Neu-Laxova syndrome type 1	PHGDH	AR

ORDERED BY DISEASE	GENE	INHERITANCE
Polycystic kidney disease (PKHD1-related)	PKHD1	AR
Polymicrogyria (ADGRG1-related)	ADGRG1	AR
POMGNT1-related disorders	POMGNT1	AR
Pontocerebellar hypoplasia (RARS2-related)	RARS2	AR
Pontocerebellar hypoplasia (SEPSECS-related)	SEPSECS	AR
Postnatal progressive microcephaly with seizures and brain atrophy/ Infantile cerebral and cerebellar atrophy (MED17-related)	MED17	AR
Primary carnitine deficiency	SLC22A5	AR
Primary Ciliary Dyskinesia (DNAH5-related)	DNAH5	AR
Primary Ciliary Dyskinesia (DNAI1-related)	DNAI1	AR
Primary Ciliary Dyskinesia (DNAI2-related)	DNAI2	AR
Primary hyperoxaluria type 1	AGXT	AR
Primary hyperoxaluria type 2	GRHPR	AR
Primary hyperoxaluria type 3	HOGA1	AR
Progressive familial intrahepatic cholestasis type 2	ABCB11	AR
Propionic acidemia (PCCA-related)	PCCA	AR
Propionic acidemia (PCCB-related)	PCCB	AR
PSAP-related disorders	PSAP	AR
Pycnodysostosis	CTSK	AR
Pyruvate carboxylase deficiency	PC	AR
Pyruvate dehydrogenase complex deficiency (PDHB-related)	PDHB	AR
RAPSN-related disorders	RAPSN	AR
Renal tubular acidosis with deafness (ATP6V1B1-related)	ATP6V1B1	AR
Retinitis pigmentosa 25	EYS	AR
Retinitis pigmentosa 26	CERKL	AR
Retinitis Pigmentosa 28	FAM161A	AR
Rhizomelic chondrodysplasia punctata type 1/ Refsum disease (PEX7-related)	PEX7	AR
Rhizomelic chondrodysplasia punctata type 3	AGPS	AR
Roberts syndrome	ESCO2	AR
RPE65-related disorders	RPE65	AR
RPGRIP1L-related disorders	RPGRIP1L	AR
RTEL-1-related disorders	RTEL1	AR
Sandhoff disease	HEXB	AR
Schimke immuno-osseous dysplasia	SMARCAL1	AR
Severe combined immune deficiency (DCLRE1C-related)	DCLRE1C	AR
Severe combined immunodeficiency (RAG2-related)	RAG2	AR
Severe congenital neutropenia due to VPS45-deficiency	VPS45	AR
Severe congenital neutropenia type 3	HAX1	AR
Sialic acid storage disorders	SLC17A5	AR
Sjögren-Larsson syndrome	ALDH3A2	AR
SLC26A2-related disorders	SLC26A2	AR

ORDERED BY DISEASE	GENE	INHERITANCE
SLC35A3-related disorders	SLC35A3	AR
Smith-Lemli-Opitz syndrome	DHCR7	AR
Spastic paraplegia type 15	ZFYVE26	AR
Spastic paraplegia type 49	TECPR2	AR
Spinal muscular atrophy	SMN1	AR
Spondylothoracic dysostosis	MESP2	AR
Steel Syndrome	COL27A1	AR
Stüve-Wiedemann syndrome	LIFR	AR
Tay-Sachs disease/ hexosaminidase A deficiency	HEXA	AR
Tetrahydrobiopterin deficiency (PTS-related)	PTS	AR
Transient infantile liver failure	TRMU	AR
Tyrosine hydroxylase deficiency	TH	AR
Tyrosinemia type I	FAH	AR
Tyrosinemia type II	TAT	AR
Usher syndrome type IB/ MYO7A-related disorders	MYO7A	AR
Usher syndrome type IC/ USH1C-related disorders	USH1C	AR
Usher syndrome type ID	CDH23	AR
Usher syndrome type IF/ PCDH15-related disorders	PCDH15	AR
Usher syndrome type IIA/ USH2A-related disorders	USH2A	AR
Usher syndrome type IIIA	CLRN1	AR
Very long-chain acyl-CoA dehydrogenase (VLCAD) deficiency	ACADVL	AR
VRK1-related disorders	VRK1	AR
Wilson disease	ATP7B	AR
WNT10A-related disorders	WNT10A	AR
Xeroderma pigmentosum complementation group A	XPA	AR
Xeroderma pigmentosum complementation group C	XPC	AR
Zellweger spectrum disorder (PEX1-related)	PEX1	AR
Zellweger spectrum disorder (PEX10-related)	PEX10	AR
Zellweger spectrum disorder (PEX12-related)	PEX12	AR
Zellweger spectrum disorder (PEX2-related)	PEX2	AR
Zellweger spectrum disorder (PEX6-related)	PEX6	AR

ORDERED BY GENE

GENE	DISEASE	INHERITANCE
ABCB11	Progressive familial intrahepatic cholestasis type 2	AR
ABCC8	ABCC8-related disorders	AR
ACAD9	ACAD9 deficiency	AR
ACADM	Medium chain acyl-CoA dehydrogenase (MCAD) deficiency	AR
ACADVL	Very long-chain acyl-CoA dehydrogenase (VLCAD) deficiency	AR
ACAT1	Beta-ketothiolase deficiency	AR
ACOX1	Peroxisomal acyl-CoA oxidase deficiency	AR
ACSF3	Combined malonic and methylmalonic aciduria (ACSF3-related)	AR
ADA	Adenosine deaminase deficiency	AR
ADAMTS2	Ehlers-Danlos syndrome, dermatosparaxis type	AR
ADGRG1	Polymicrogyria (ADGRG1-related)	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 with candidiasis and ectodermal dysplasia	AR
ALDH3A2	Sjögren-Larsson syndrome	AR
ALDOB	Hereditary fructose intolerance	AR
ALG6	Congenital disorder of glycosylation (ALG6-related)	AR
ALMS1	Alström syndrome	AR
ALPL	Hypophosphatasia	AR
AMT	Glycine encephalopathy (AMT-related)	AR
AQP2	Nephrogenic diabetes insipidus (AQP2-related)	AR
ARG1	Arginase deficiency	AR
ARSA	Metachromatic leukodystrophy (ARSA-related)	AR
ARSB	Mucopolysaccharidosis type VI (Maroteaux-Lamy syndrome)	AR
ASL	Argininosuccinic aciduria	AR
ASNS	Asparagine synthetase deficiency	AR
ASPA	Canavan disease	AR
ASS1	Citrullinemia type 1	AR
ATM	Ataxia-telangiectasia	AR
ATP6V1B1	Renal tubular acidosis with deafness (ATP6V1B1-related)	AR
ATP7B	Wilson disease	AR
BBS1	BBS1-related disorders	AR
BBS10	Bardet-Biedl syndrome (BBS10-related)	AR
BBS12	Bardet-Biedl syndrome (BBS12-related)	AR
BBS2	BBS2-related disorders	AR
BCKDHA	Maple syrup urine disease (MSUD) type 1A	AR
BCKDHB	Maple syrup urine disease (MSUD) type 1B	AR
BCS1L	GRACILE syndrome/ BCS1L-related disorders	AR

GENE	DISEASE	INHERITANCE
BLM	Bloom syndrome	AR
BSND	Bartter syndrome type 4A	AR
CAPN3	Limb-girdle muscular dystrophy type 2A (calpainopathy)	AR
CBS	Homocystinuria due to CBS deficiency	AR
CDH23	Usher syndrome type ID	AR
CEP290	Leber congenital amaurosis 10/ CEP290-related disorders	AR
CERKL	Retinitis pigmentosa 26	AR
CFTR	CFTR-related disorders (including cystic fibrosis)	AR
CHRNE	Congenital myasthenic syndrome (CHRNE-related)	AR
CIITA	Major histocompatibility complex class II deficiency (CIITA-related)	AR
CLN3	Neuronal ceroid-lipofuscinosis (CLN3-related)	AR
CLN5	Neuronal ceroid-lipofuscinosis (CLN5-related)	AR
CLN6	Neuronal ceroid-lipofuscinosis (CLN6-related)	AR
CLN8	Neuronal ceroid-lipofuscinosis/ Northern epilepsy (CLN8-related)	AR
CLRN1	Usher syndrome type IIIA	AR
CNGB3	Achromatopsia (CNGB3-related)	AR
COL27A1	Steel Syndrome	AR
COL4A3	Alport Syndrome (COL4A3-related)	AR
COL4A4	Alport Syndrome (COL4A4-related)	AR
COL7A1	Dystrophic epidermolysis bullosa (COL7A1-related)	AR
CPS1	Carbamoylphosphate synthetase I deficiency	AR
CPT1A	Carnitine palmitoyltransferase I deficiency	AR
CPT2	Carnitine palmitoyltransferase II deficiency	AR
CRB1	Leber congenital amaurosis 8/ CRB1-related disorders	AR
CTNS	Cystinosis	AR
CTSK	Pycnodysostosis	AR
CYBA	Chronic granulomatous disease (CYBA-related)	AR
CYP11B1	Congenital adrenal hyperplasia due to 11-beta-hydroxylase-deficiency	AR
CYP11B2	Aldosterone synthase deficiency	AR
CYP17A1	CYP17A1-related disorders	AR
CYP19A1	Aromatase deficiency	AR
CYP21A2	Congenital adrenal hyperplasia due to 21-hydroxylase deficiency	AR
CYP27A1	Cerebrotendinous xanthomatosis	AR
DBT	Maple syrup urine disease (MSUD) type 2	AR
DCLRE1C	Severe combined immune deficiency (DCLRE1C-related)	AR
DHCR7	Smith-Lemli-Opitz syndrome	AR
DHDDS	DHDDS-related disorders	AR
DLD	Dihydrolipoamide dehydrogenase deficiency (DLD)	AR
DNAH5	Primary Ciliary Dyskinesia (DNAH5-related)	AR
DNAI1	Primary Ciliary Dyskinesia (DNAI1-related)	AR
DNAI2	Primary Ciliary Dyskinesia (DNAI2-related)	AR

GENE	DISEASE	INHERITANCE
DYSF	Dysferlinopathy	AR
EIF2B5	Leukoencephalopathy with vanishing white matter (EIF2B5-related)	AR
ELP1	Familial dysautonomia	AR
ERCC6	Cockayne syndrome type B	AR
ERCC8	Cockayne syndrome type A	AR
ESCO2	Roberts syndrome	AR
ETFA	Glutaric acidemia type IIA	AR
ETFDH	Glutaric acidemia type IIC	AR
ETHE1	Ethylmalonic encephalopathy	AR
EVC	Ellis-van Creveld syndrome (EVC-related)	AR
EVC2	Ellis-van Creveld syndrome (EVC2-related)	AR
EYS	Retinitis pigmentosa 25	AR
FAH	Tyrosinemia type I	AR
FAM161A	Retinitis Pigmentosa 28	AR
FANCA	Fanconi anemia type A	AR
FANCC	Fanconi anemia type C	AR
FANCG	Fanconi anemia type G	AR
FH	Fumarate hydratase deficiency	AR
FKRP	FKRP-related disorders	AR
FKTN	FKTN-related disorders	AR
G6PC	Glycogen storage disease type Ia	AR
GAA	Glycogen storage disease type II (Pompe disease)	AR
GALC	Krabbe disease	AR
GALK1	Galactokinase deficiency galactosemia	AR
GALT	Galactosemia (GALT-related)	AR
GAMT	Guanidinoacetate methyltransferase deficiency	AR
GBA	Gaucher disease	AR
GBE1	Glycogen storage disease type IV/ adult polyglucosan body disease	AR
GCDH	Glutaric acidemia type I	AR
GFM1	Combined oxidative phosphorylation deficiency (GFM1-related)	AR
GJB2	GJB2-related DFNB1 nonsyndromic hearing loss and deafness	AR
GLB1	Mucopolysaccharidosis type IVB (Morquio B syndrome)/ GM1 gangliosidosis	AR
GLDC	Glycine encephalopathy (GLDC-related)	AR
GLE1	GLE1-related disorders	AR
GNE	Inclusion body myopathy 2	AR
GNPTAB	Mucolipidosis type II/III (GNPTAB-related)	AR
GNPTG	Mucolipidosis type III (GNPTG-related)	AR
GNS	Mucopolysaccharidosis type IIID (Sanfilippo D syndrome)	AR
GRHPR	Primary hyperoxaluria type 2	AR
HADHA	Long chain 3-hydroxyacyl-CoA dehydrogenase (LCHAD) deficiency	AR
HAX1	Severe congenital neutropenia type 3	AR

GENE	DISEASE	INHERITANCE
HBA1/HBA2	Alpha-thalassemia	AR
HBB	HBB-related hemoglobinopathies	AR
HEXA	Tay-Sachs disease/ hexosaminidase A deficiency	AR
HEXB	Sandhoff disease	AR
HGSNAT	Mucopolysaccharidosis type IIIC (Sanfilippo C syndrome)/ retinitis pigmentosa 73	AR
HJV	Hereditary hemochromatosis type 2 (HJV-related)	AR
HLCS	Holocarboxylase synthetase deficiency	AR
HMGCL	3-hydroxy-3-methylglutaryl-CoA (HMG-CoA) lyase deficiency	AR
HOGA1	Primary hyperoxaluria type 3	AR
HPS1	Hermansky-Pudlak syndrome type 1	AR
HPS3	Hermansky-Pudlak syndrome type 3	AR
HSD17B4	HSD17B4-related disorders	AR
HSD3B2	Congenital adrenal hyperplasia due to 3-beta-hydroxysteroid dehydrogenase type II deficiency	AR
HYAL1	Mucopolysaccharidosis type IX	AR
HYLS1	Hydrolethalus syndrome type 1	AR
IDUA	Mucopolysaccharidosis type I	AR
IVD	Isovaleric acidemia	AR
KCNJ11	KCNJ11-related disorders	AR
LAMA2	LAMA2-related muscular dystrophy	AR
LAMA3	LAMA3-related disorders	AR
LAMB3	Junctional epidermolysis bullosa (LAMB3-related)	AR
LAMC2	Junctional epidermolysis bullosa (LAMC2-related)	AR
LCA5	Leber congenital amaurosis 5	AR
LDLR	Familial hypercholesterolemia (LDLR-related)	AD
LDLRAP1	Familial hypercholesterolemia (LDLRAP1-related)	AR
LHX3	Combined pituitary hormone deficiency (LHX3-related)	AR
LIFR	Stüve-Wiedemann syndrome	AR
LIPA	Lysosomal acid lipase deficiency	AR
LOXHD1	Autosomal recessive deafness 77	AR
LPL	Lipoprotein lipase deficiency	AR
LRPPRC	Leigh syndrome, French Canadian type	AR
MAN2B1	Alpha-mannosidosis	AR
MCOLN1	Mucolipidosis type IV	AR
MED17	Postnatal progressive microcephaly with seizures and brain atrophy/ Infantile cerebral and cerebellar atrophy (MED17-related)	AR
MESP2	Spondylothoracic dysostosis	AR
MFSD8	Neuronal ceroid-lipofuscinosis (MFSD8-related)	AR
MKS1	MKS1-related disorders	AR
MLC1	Megalencephalic leukoencephalopathy with subcortical cysts type 1	AR
MMAA	Methylmalonic acidemia (MMAA-related)	AR
MMAB	Methylmalonic acidemia (MMAB-related)	AR

GENE	DISEASE	INHERITANCE
MMACHC	Methylmalonic acidemia with homocystinuria, cobalamin C type	AR
MMADHC	Methylmalonic acidemia with homocystinuria, cobalamin D type	AR
MPI	Congenital disorder of glycosylation (MPI-related)	AR
MPL	Congenital amegakaryocytic thrombocytopenia	AR
MPV17	Mitochondrial DNA depletion syndrome (MPV17-related)	AR
MTHFR	Homocystinuria due to MTHFR deficiency	AR
MTRR	Homocystinuria, cobalamin E type	AR
MTTP	Abetalipoproteinemia	AR
MUT	Methylmalonic acidemia (MUT-related)	AR
MYO7A	Usher syndrome type IB/ MYO7A-related disorders	AR
NAGLU	Mucopolysaccharidosis type IIIB (Sanfilippo B syndrome)	AR
NAGS	N-Acetylglutamate synthase deficiency	AR
NBN	Nijmegen breakage syndrome	AR
NDRG1	Charcot-Marie-Tooth disease (NDRG1-related)	AR
NDUFAF5	Mitochondrial complex I deficiency/ Leigh syndrome (NDUFAF5-related)	AR
NDUFS6	Mitochondrial complex I deficiency/ Leigh syndrome (NDUFS6- related)	AR
NEB	Nemaline myopathy 2	AR
NPC1	Niemann-Pick disease type C (NPC1-related)	AR
NPC2	Niemann-Pick disease type C (NPC2-related)	AR
NPHS1	Nephrotic syndrome/ congenital Finnish nephrosis (NPHS1-related)	AR
NPHS2	Nephrotic syndrome/steroid-resistant nephrotic syndrome (NPHS2-related)	AR
NR2E3	Enhanced S-cone syndrome/ retinitis pigmentosa 37	AR
NTRK1	Congenital insensitivity to pain with anhidrosis	AR
OAT	Ornithine aminotransferase deficiency	AR
OPA3	OPA3-related conditions	AR
PAH	Phenylalanine hydroxylase deficiency (including Phenylketonuria (PKU))	AR
PC	Pyruvate carboxylase deficiency	AR
PCCA	Propionic acidemia (PCCA-related)	AR
PCCB	Propionic acidemia (PCCB-related)	AR
PCDH15	Usher syndrome type IF/ PCDH15-related disorders	AR
PDHB	Pyruvate dehydrogenase complex deficiency (PDHB-related)	AR
PEX1	Zellweger spectrum disorder (PEX1-related)	AR
PEX10	Zellweger spectrum disorder (PEX10-related)	AR
PEX12	Zellweger spectrum disorder (PEX12-related)	AR
PEX2	Zellweger spectrum disorder (PEX2-related)	AR
PEX6	Zellweger spectrum disorder (PEX6-related)	AR
PEX7	Rhizomelic chondrodysplasia punctata type 1/ Refsum disease (PEX7-related)	AR
PFKM	Glycogen storage disease type VII	AR
PHGDH	Phosphoglycerate dehydrogenase deficiency/ Neu-Laxova syndrome type 1	AR
PKHD1	Polycystic kidney disease (PKHD1-related)	AR
PMM2	Congenital disorder of glycosylation (PMM2-related)	AR

GENE	DISEASE	INHERITANCE
POMGNT1	POMGNT1-related disorders	AR
PPT1	Neuronal ceroid-lipofuscinosis (PPT1-related)	AR
PROP1	Combined pituitary hormone deficiency (PROP1-related)	AR
PSAP	PSAP-related disorders	AR
PTS	Tetrahydrobiopterin deficiency (PTS-related)	AR
PUS1	Mitochondrial myopathy and sideroblastic anemia 1	AR
PYGM	Glycogen storage disease type V	AR
RAB23	Carpenter syndrome (RAB23-related)	AR
RAG2	Severe combined immunodeficiency (RAG2-related)	AR
RAPSN	RAPSN-related disorders	AR
RARS2	Pontocerebellar hypoplasia (RARS2-related)	AR
RDH12	Leber congenital amaurosis 13	AR
RMRP	Cartilage-hair hypoplasia-anauxetic dysplasia spectrum disorders	AR
RPE65	RPE65-related disorders	AR
RPGRIP1L	RPGRIP1L-related disorders	AR
RTEL1	RTEL-1-related disorders	AR
SACS	Autosomal recessive spastic ataxia of Charlevoix-Saguenay (ARSACS)	AR
SAMHD1	Aicardi-Goutieres syndrome (SAMHD1-related)	AR
SEPSECS	Pontocerebellar hypoplasia (SEPSECS-related)	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 type IIIA (Sanfilippo A syndrome)	AR
SLC12A3	Gitelman syndrome (SLC12A3-related)	AR
SLC12A6	Andermann syndrome	AR
SLC17A5	Sialic acid storage disorders	AR
SLC22A5	Primary carnitine deficiency	AR
SLC25A13	Citrin deficiency	AR
SLC25A15	Hyperornithinemia-hyperammonemia-homocitrullinuria (HHH) syndrome	AR
SLC26A2	SLC26A2-related disorders	AR
SLC26A4	Pendred syndrome	AR
SLC35A3	SLC35A3-related disorders	AR
SLC37A4	Glycogen storage disease type Ib	AR
SLC39A4	Acrodermatitis enteropathica	AR
SLC4A11	Corneal dystrophy and perceptive deafness	AR
SLC7A7	Lysinuric protein intolerance	AR
SMARCAL1	Schimke immuno-osseous 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

GENE	DISEASE	INHERITANCE
TAT	Tyrosinemia type II	AR
TCIRG1	Osteopetrosis (TCIRG1-related)	AR
TECPR2	Spastic paraplegia type 49	AR
TFR2	Hereditary hemochromatosis type 3	AR
TGM1	Congenital ichthyosis (TGM1-related)	AR
TH	Tyrosine hydroxylase deficiency	AR
TMEM216	Joubert syndrome 2/ TMEM216-related disorders	AR
TPP1	Neuronal ceroid lipofuscinosis (TPP1-related)	AR
TRMU	Transient infantile liver failure	AR
TSFM	Combined oxidative phosphorylation deficiency (TSFM-related)	AR
TTPA	Ataxia with vitamin E deficiency	AR
TYMP	Mitochondrial neurogastrointestinal encephalopathy (MNGIE) disease	AR
USH1C	Usher syndrome type IC/ USH1C-related disorders	AR
USH2A	Usher syndrome type IIA/ USH2A-related disorders	AR
VPS13A	Chorea-acanthocytosis	AR
VPS13B	Cohen syndrome	AR
VPS45	Severe congenital neutropenia due to VPS45-deficiency	AR
VRK1	VRK1-related disorders	AR
VSX2	Microphthalmia /clinical anophthalmia (VSX2-related)	AR
WNT10A	WNT10A-related disorders	AR
XPA	Xeroderma pigmentosum complementation group A	AR
XPC	Xeroderma pigmentosum complementation group C	AR
ZFYVE26	Spastic paraplegia type 15	AR

Add-on genes (without X-linked disorders)

GENE	DISEASE	INHERITANCE
BTD	Biotinidase deficiency	AR
F11	Factor XI deficiency (Hemophilia C)	AR
F2	Prothrombin-related thrombophilia	AD
F5	Factor V Leiden thrombophilia	AD
GP1BA	Bernard-Soulier syndrome (GP1BA-related)	AR
GP9	Bernard-Soulier syndrome (GP9-related)	AR
HFE	Hereditary hemochromatosis (HFE-related)	AR
HGD	Alkaptonuria	AR
MCCC1	3-methylcrotonyl-CoA carboxylase (3-MCC) deficiency (MCC1-related)	AR
MCCC2	3-methylcrotonyl-CoA carboxylase (3-MCC) deficiency (MCC2-related)	AR
MEFV	Familial mediterranean fever	AR
SERPINA1	Alpha-1 antitrypsin deficiency	AR