DISORDER	GENE
3-hydroxy-3-methylglutarayl-CoA (HMG-CoA) lyase deficiency	HMGCL
3-methylcrotonyl-CoA carboxylase (3-MCC) deficiency (MCC1-related)	MCCC1
3-methylcrotonyl-CoA carboxylase (3-MCC) deficiency (MCC2-related)	MCCC2
ABCC8-related disorders	ABCC8
Abetalipoproteinemia	MTTP
ACAD9 deficiency	ACAD9
Achromatopsia (CNGB3-related)	CNGB3
Acrodermatitis enteropathica	SLC39A4
Adenosine deaminase deficiency	ADA
Aicardi-Goutieres syndrome (SAMHD1-related)	SAMHD1
Aldosterone synthase deficiency	CYP11B2
Alkaptonuria	HGD
Alpha-1 antitrypsin deficiency	SERPINA1
Alpha-mannosidosis	MAN2B1
Alpha-thalassemia	HBA1/HBA2
Alpha-thalassemia X-linked intellectual disability syndrome	ATRX

DISORDER	GENE
Alport Syndrome (COL4A3-related)	COL4A3
Alport Syndrome (COL4A4-related)	COL4A4
Alport Syndrome, X-linked (COL4A5-related)	COL4A5
Alström syndrome	ALMS1
Andermann syndrome	SLC12A6
Arginase deficiency	ARG1
Argininosuccinic aciduria	ASL
Aromatase deficiency	CYP19A1
Asparagine synthetase deficiency	ASNS
Aspartylglucosaminuria	AGA
Ataxia with vitamin E deficiency	TTPA
Ataxia-telangiectasia	ATM
Autoimmune polyendocrinopathy with candidiasis and ectodermal dysplasia	AIRE
Autosomal recessive deafness 77	LOXHD1
Autosomal recessive spastic ataxia of Charlevoix-Saguenay (ARSACS)	SACS
Bardet-Biedl syndrome (BBS10-related)	BBS10

DISORDER	GENE
Bardet-Biedl syndrome (BBS12-related)	BBS12
Bartter syndrome type 4A	BSND
BBS1-related disorders	BBS1
BBS2-related disorders	BBS2
Bernard-Soulier syndrome (GP1BA-related)	GP1BA
Bernard-Soulier syndrome (GP9-related)	GP9
Beta-ketothiolase deficiency	ACAT1
Biotinidase deficiency	BTD
Bloom syndrome	BLM
Canavan disease	ASPA
Carbamoylphosphate synthetase I deficiency	CPS1
Carnitine palmitoyltransferase I deficiency	CPT1A
Carnitine palmitoyltransferase II deficiency	CPT2
Carpenter syndrome (RAB23-related)	RAB23
Cartilage-hair hypoplasia-anauxetic dysplasia spectrum disorders	RMRP
Cerebrotendinous xanthomatosis	CYP27A1

DISORDER	GENE
CFTR-related disorders (including cystic fibrosis)	CFTR
Charcot-Marie-Tooth disease (NDRG1-related)	NDRG1
Charcot-Marie-Tooth disease, X-linked (GJB1-related)	GJB1
Chorea-acanthocytosis	VPS13A
Choroideremia	CHM
Chronic granulomatous disease (CYBA-related)	CYBA
Chronic granulomatous disease (CYBB-related)	CYBB
Citrin deficiency	SLC25A13
Citrullinemia type 1	ASS1
Cockayne syndrome type A	ERCC8
Cockayne syndrome type B	ERCC6
Cohen syndrome	VPS13B
Combined malonic and methylmalonic aciduria (ACSF3-related)	ACSF3
Combined oxidative phosphorylation deficiency (GFM1-related)	GFM1
Combined oxidative phosphorylation deficiency (TSFM-related)	TSFM
Combined pituitary hormone deficiency (LHX3-related)	LHX3

DISORDER	GENE
Combined pituitary hormone deficiency (PROP1-related)	PROP1
Congenital adrenal hyperplasia due to 11-beta-hydroxylase-deficiency	CYP11B1
Congenital adrenal hyperplasia due to 21-hydroxylase deficiency	CYP21A2
Congenital adrenal hyperplasia due to 3-beta-hydroxysteroid dehydrogenase type II deficiency	HSD3B2
Congenital amegakaryocytic thrombocytopenia	MPL
Congenital disorder of glycosylation (ALG6-related)	ALG6
Congenital disorder of glycosylation (MPI-related)	MPI
Congenital disorder of glycosylation (PMM2-related)	PMM2
Congenital ichthyosis (TGM1-related)	TGM1
Congenital insensitivity to pain with anhidrosis	NTRK1
Congenital myasthenic syndrome (CHRNE-related)	CHRNE
Corneal dystrophy and perceptive deafness	SLC4A11
CYP17A1-related disorders	CYP17A1
Cystinosis	CTNS
DHDDS-related disorders	DHDDS
Dihydrolipoamide dehydrogenase deficiency (DLD)	DLD

DISORDER	GENE
DMD-related dystrophinopathy	DMD
Dysferlinopathy	DYSF
Dystrophic epidermolysis bullosa (COL7A1-related)	COL7A1
Ehlers-Danlos syndrome, dermatosparaxis type	ADAMTS2
Ellis-van Creveld syndrome (EVC2-related)	EVC2
Ellis-van Creveld syndrome (EVC-related)	EVC
Emery-Dreifuss muscular dystrophy (EMD-related)	EMD
Enhanced S-cone syndrome/ retinitis pigmentosa 37	NR2E3
Ethylmalonic encephalopathy	ETHE1
Fabry disease	GLA
Factor IX deficiency (Hemophilia B)	F9
Factor V Leiden thrombophilia	F5
Factor XI deficiency (Hemophilia C)	F11
Familial dysautonomia	ELP1
Familial hypercholesterolemia (LDLRAP1-related)	LDLRAP1
Familial hypercholesterolemia (LDLR-related)	LDLR

DISORDER	GENE
Familial mediterranean fever	MEFV
Fanconi anemia type A	FANCA
Fanconi anemia type C	FANCC
Fanconi anemia type G	FANCG
FKRP-related disorders	FKRP
FKTN-related disorders	FKTN
Fragile X syndrome	FMR1
Fumarate hydratase deficiency	FH
Galactokinase deficiency galactosemia	GALK1
Galactosemia (GALT-related)	GALT
Gaucher disease	GBA
Gitelman syndrome (SLC12A3-related)	SLC12A3
GJB2-related DFNB1 nonsyndromic hearing loss and deafness	GJB2
GLE1-related disorders	GLE1
Glucose-6-phosphate dehydrogenase (G6PD) deficiency	G6PD
Glutaric acidemia type I	GCDH

DISORDER	GENE
Glutaric acidemia type IIA	ETFA
Glutaric acidemia type IIC	ETFDH
Glycine encephalopathy (AMT-related)	AMT
Glycine encephalopathy (GLDC-related)	GLDC
Glycogen storage disease type Ia	G6PC
Glycogen storage disease type Ib	SLC37A4
Glycogen storage disease type II (Pompe disease)	GAA
Glycogen storage disease type IV/ adult polyglucosan body disease	GBE1
Glycogen storage disease type V	PYGM
Glycogen storage disease type III	AGL
Glycogen storage disease type VII	PFKM
GRACILE syndrome/ BCS1L-related disorders	BCS1L
Guanidinoacetate methyltransferase deficiency	GAMT
HBB-related hemoglobinopathies	HBB
Hereditary fructose intolerance	ALDOB
Hereditary hemochromatosis (HFE-related)	HFE

DISORDER	GENE
Hereditary hemochromatosis type 2 (HJV-related)	HJV
Hereditary hemochromatosis type 3	TFR2
Hermansky-Pudlak syndrome type 1	HPS1
Hermansky-Pudlak syndrome type 3	HPS3
Holocarboxylase synthetase deficiency	HLCS
Homocystinuria due to CBS deficiency	CBS
Homocystinuria due to MTHFR deficiency	MTHFR
Homocystinuria, cobalamin E type	MTRR
HSD17B4-related disorders	HSD17B4
Hydrolethalus syndrome type 1	HYLS1
Hyperornithinemia-hyperammonemia-homocitrullinuria (HHH) syndrome	SLC25A15
Hypohidrotic ectodermal dysplasia (EDA-related)	EDA
Hypophosphatasia	ALPL
Inclusion body myopathy 2	GNE
Isovaleric acidemia	IVD
Joubert syndrome 2/ TMEM216-related disorders	TMEM216

DISORDER	GENE
Junctional epidermolysis bullosa (LAMB3-related)	LAMB3
Junctional epidermolysis bullosa (LAMC2-related)	LAMC2
KCNJ11-related disorders	KCNJ11
Krabbe disease	GALC
LAMA2-related muscular dystrophy	LAMA2
LAMA3-related disorders	LAMA3
Leber congenital amaurosis 10/ CEP290-related disorders	CEP290
Leber congenital amaurosis 13	RDH12
Leber congenital amaurosis 5	LCA5
Leber congenital amaurosis 8/ CRB1-related disorders	CRB1
Leigh syndrome, French Canadian type	LRPPRC
Leukoencephalopathy with vanishing white matter (EIF2B5-related)	EIF2B5
Limb-girdle muscular dystrophy type 2A (calpainopathy)	CAPN3
Limb-girdle muscular dystrophy type 2C	SGCG
Limb-girdle muscular dystrophy type 2D	SGCA
Limb-girdle muscular dystrophy type 2E	SGCB

DISORDER	GENE
Lipoid congenital adrenal hyperplasia	STAR
Lipoprotein lipase deficiency	LPL
Long chain 3-hydroxyacyl-CoA dehydrogenase (LCHAD) deficiency	HADHA
Lysinuric protein intolerance	SLC7A7
Lysosomal acid lipase deficiency	LIPA
Major histocompatibility complex class II deficiency (CIITA-related)	CIITA
Maple syrup urine disease (MSUD) type 1A	BCKDHA
Maple syrup urine disease (MSUD) type 1B	BCKDHB
Maple syrup urine disease (MSUD) type 2	DBT
Medium chain acyl-CoA dehydrogenase (MCAD) deficiency	ACADM
Megalencephalic leukoencephalopathy with subcortical cysts type 1	MLC1
Menkes disease/ ATP7A-related disorders	ATP7A
Metachromatic leukodystrophy (ARSA-related)	ARSA
Methylmalonic acidemia (MMAA-related)	MMAA
Methylmalonic acidemia (MMAB-related)	MMAB
Methylmalonic acidemia (MUT-related)	MUT

DISORDER	GENE
Methylmalonic acidemia with homocystinuria, cobalamin C type	MMACHC
Methylmalonic acidemia with homocystinuria, cobalamin D type	MMADHC
Microphthalmia /clinical anophthalmia (VSX2-related)	VSX2
Mitochondrial complex I deficiency/ Leigh syndrome (NDUFAF5-related)	NDUFAF5
Mitochondrial complex I deficiency/ Leigh syndrome (NDUFS6- related)	NDUFS6
Mitochondrial myopathy and sideroblastic anemia 1	PUS1
Mitochondrial neurogastrointestinal encephalopathy (MNGIE) disease	TYMP
Mitochondrial DNA depletion syndrome (MPV17-related)	MPV17
MKS1-related disorders	MKS1
Mucolipidosis type II/III (GNPTAB-related)	GNPTAB
Mucolipidosis type IV	MCOLN1
Mucolipidosis type III (GNPTG-related)	GNPTG
Mucopolysaccharidosis type I	IDUA
Mucopolysaccharidosis type II (Hunter syndrome)	IDS
Mucopolysaccharidosis type IX	HYAL1
Mucopolysaccharidosis type VI (Maroteaux-Lamy syndrome)	ARSB

DISORDER	GENE
Mucopolysaccharidosis type IIIA (Sanfilippo A syndrome)	SGSH
Mucopolysaccharidosis type IIIB (Sanfilippo B syndrome)	NAGLU
Mucopolysaccharidosis type IIIC (Sanfilippo C syndrome)/ retinitis pigmentosa 73	HGSNAT
Mucopolysaccharidosis type IIID (Sanfilippo D syndrome)	GNS
Mucopolysaccharidosis type IVB (Morquio B syndrome)/ GM1 gangliosidosis	GLB1
Multiple sulfatase deficiency	SUMF1
N-Acetylglutamate synthase deficiency	NAGS
Nemaline myopathy 2	NEB
Nephrogenic diabetes insipidus (AQP2-related)	AQP2
Nephrotic syndrome/ congenital Finnish nephrosis (NPHS1-related)	NPHS1
Nephrotic syndrome/steroid-resistant nephrotic syndrome (NPHS2-related)	NPHS2
Neuronal ceroid lipofuscinosis (TPP1-related)	TPP1
Neuronal ceroid-lipofuscinosis (CLN3-related)	CLN3
Neuronal ceroid-lipofuscinosis (CLN5-related)	CLN5
Neuronal ceroid-lipofuscinosis (CLN6-related)	CLN6
Neuronal ceroid-lipofuscinosis (MFSD8-related)	MFSD8

DISORDER	GENE
Neuronal ceroid-lipofuscinosis (PPT1-related)	PPT1
Neuronal ceroid-lipofuscinosis/ Northern epilepsy (CLN8-related)	CLN8
Niemann-Pick disease type A/B	SMPD1
Niemann-Pick disease type C (NPC1-related)	NPC1
Niemann-Pick disease type C (NPC2-related)	NPC2
Nijmegen breakage syndrome	NBN
OPA3-related conditions	OPA3
Ornithine aminotransferase deficiency	OAT
Ornithine transcarbamylase (OTC) deficiency	OTC
Osteopetrosis (TCIRG1-related)	TCIRG1
Pendred syndrome	SLC26A4
Peroxisomal acyl-CoA oxidase deficiency	ACOX1
Phenylalanine hydroxylase deficiency (including Phenylketonuria (PKU))	PAH
Phosphoglycerate dehydrogenase deficiency/ Neu-Laxova syndrome type 1	PHGDH
Polycystic kidney disease (PKHD1-related)	PKHD1
Polymicrogyria (ADGRG1-related)	ADGRG1

DISORDER	GENE
POMGNT1-related disorders	POMGNT1
Pontocerebellar hypoplasia (RARS2-related)	RARS2
Pontocerebellar hypoplasia (SEPSECS-related)	SEPSECS
Postnatal progressive microcephaly with seizures and brain atrophy/ Infantile cerebral and cerebellar atrophy (MED17-related)	MED17
Primary carnitine deficiency	SLC22A5
Primary Ciliary Dyskinesia (DNAH5-related)	DNAH5
Primary Ciliary Dyskinesia (DNAI1-related)	DNAI1
Primary Ciliary Dyskinesia (DNAI2-related)	DNAI2
Primary hyperoxaluria type 1	AGXT
Primary hyperoxaluria type 2	GRHPR
Primary hyperoxaluria type 3	HOGA1
Progressive familial intrahepatic cholestasis type 2	ABCB11
Propionic acidemia (PCCA-related)	PCCA
Propionic acidemia (PCCB-related)	PCCB
Prothrombin-related thrombophilia	F2
PRPS1-related disorders	PRPS1

DISORDER	GENE
PSAP-related disorders	PSAP
Pycnodysostosis	CTSK
Pyruvate carboxylase deficiency	PC
Pyruvate dehydrogenase complex deficiency (PDHA1-related)	PDHA1
Pyruvate dehydrogenase complex deficiency (PDHB-related)	PDHB
RAPSN-related disorders	RAPSN
Renal tubular acidosis with deafness (ATP6V1B1-related)	ATP6V1B1
Retinitis pigmentosa 25	EYS
Retinitis pigmentosa 26	CERKL
Retinitis Pigmentosa 28	FAM161A
Rhizomelic chondrodysplasia punctata type 1/ Refsum disease (PEX7-related)	PEX7
Rhizomelic chondrodysplasia punctata type 3	AGPS
Roberts syndrome	ESCO2
RPE65-related disorders	RPE65
RPGRIP1L-related disorders	RPGRIP1L
RTEL-1-related disorders	RTEL1

DISORDER	GENE
Sandhoff disease	HEXB
Schimke immuno-osseous dysplasia	SMARCAL1
Severe combined immune deficiency (DCLRE1C-related)	DCLRE1C
Severe combined immunodeficiency (RAG2-related)	RAG2
Severe congenital neutropenia due to VPS45-deficiency	VPS45
Severe congenital neutropenia type 3	HAX1
Sialic acid storage disorders	SLC17A5
Sjögren-Larsson syndrome	ALDH3A2
SLC26A2-related disorders	SLC26A2
SLC35A3-related disorders	SLC35A3
Smith-Lemli-Opitz syndrome	DHCR7
Spastic paraplegia type 15	ZFYVE26
Spastic paraplegia type 49	TECPR2
Spinal muscular atrophy	SMN1
Spondylothoracic dysostosis	MESP2
Steel Syndrome	COL27A1

DISORDER	GENE
Stüve-Wiedemann syndrome	LIFR
Tay-Sachs disease/ hexosaminidase A deficiency	HEXA
Tetrahydrobiopterin deficiency (PTS-related)	PTS
Transient infantile liver failure	TRMU
Tyrosine hydroxylase deficiency	TH
Tyrosinemia type I	FAH
Tyrosinemia type II	TAT
Usher syndrome type IB/ MYO7A-related disorders	MYO7A
Usher syndrome type IC/ USH1C-related disorders	USH1C
Usher syndrome type ID	CDH23
Usher syndrome type IF/ PCDH15-related disorders	PCDH15
Usher syndrome type IIA/ USH2A-related disorders	USH2A
Usher syndrome type IIIA	CLRN1
Very long-chain acyl-CoA dehydrogenase (VLCAD) deficiency	ACADVL
VRK1-related disorders	VRK1
Wilson disease	ATP7B

DISORDER	GENE
WNT10A-related disorders	WNT10A
Xeroderma pigmentosum complementation group A	XPA
Xeroderma pigmentosum complementation group C	XPC
X-linked adrenoleukodystrophy	ABCD1
X-linked creatine transporter deficiency	SLC6A8
X-linked juvenile retinoschisis	RS1
X-linked myotubular myopathy	MTM1
X-linked severe combined immunodeficiency (X-SCID)	IL2RG
Zellweger spectrum disorder (PEX10-related)	PEX10
Zellweger spectrum disorder (PEX12-related)	PEX12
Zellweger spectrum disorder (PEX1-related)	PEX1
Zellweger spectrum disorder (PEX2-related)	PEX2
Zellweger spectrum disorder (PEX6-related)	PEX6