DISORDER	GENE
11-beta-hydroxylase-deficient congenital adrenal hyperplasia	CYP11B1
17-alpha-hydroxylase-deficient congenital adrenal hyperplasia	CYP17A1
3-beta-hydroxysteroid dehydrogenase type II deficiency	
(Congenital adrenal hyperplasia)	HSD3B2
3-hydroxy-3-methylglutarayl-CoA (HMG-CoA) lyase deficiency	HMGCL
3-methylcrotonyl-CoA carboxylase (3-MCC) deficiency (MCCC1-related)	MCCC1
3-methylcrotonyl-CoA carboxylase (3-MCC) deficiency (MCCC2-related)	MCCC2
3-methylglutaconic aciduria type III (Costeff optic atrophy)	OPA3
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
Alpha-1 antitrypsin deficiency	SERPINA1
Alpha-mannosidosis	MAN2B1
Alpha-thalassemia	HBA1/HBA2
Alpha-thalassemia X-linked intellectual disability syndrome	ATRX
Alkaptonuria	HGD
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 qATM	
Autoimmune polyendocrinopathy with candidiasis and ectodermal dysplasia	AIRE
Autosomal recessive deafness 77 (DFNB77)	LOXHD1
Autosomal recessive spastic ataxia of Charlevoix-Saguenay (ARSACS)	SACS
Bardet-Biedl syndrome (BBS1-related)	BBS1
Bardet-Biedl syndrome (BBS10-related)	BBS10
Bardet-Biedl syndrome (BBS12-related)	BBS12
Bardet-Biedl syndrome (BBS2-related)	BBS2
Bartter syndrome type IV (BSND-related)	BSND
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
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
Combined pituitary hormone deficiency (PROP1-related)	PROP1
Combined SAP Deficiency	PSAP
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
Congenital myasthenic syndrome (RAPSN-related)	RAPSN
Congenital neutropenia (HAX1-related)	HAX1
Corneal dystrophy and perceptive deafness	SLC4A11
Cystic fibrosis/ CFTR-related disorders	CFTR
Cystinosis	CTNS
D-bifunctional protein deficiency	HSD17B4
DHDDS-related disorders	
(including Congenital disorder of glycoslylation/ Retinitis pigmentosa 59)	DHDDS
Dihydrolipoamide dehydrogenase deficiency (DLD)	DLD
DMD-related dystrophinopathy	
(Including Duchenne/Becker muscular dystrophy and Dilated cardiomyopathy)	DMD
Dystrophic epidermolysis bullosa (COL7A1-related)	COL7A1
Ehlers-Danlos syndrome, dermatosparaxis type	ADAMTS2
Ellis-van Creveld syndrome (EVC-related)	EVC
Ellis-van Creveld syndrome (EVC2-related)	EVC2
Emery-Dreifuss muscular dystrophy (EMD-related)	EMD
Enhanced S-cone syndrome/ Retinitis pigmentosa 37	NR2E3
Ethylmalonic encephalopathy	ETHE1

Fabry disease GLA	
Factor V Leiden thrombophilia	F5
Factor IX deficiency (Hemophilia B)	F9
Factor XI deficiency (Hemophilia C)	F11
Familial dysautonomia	ELP1
Familial hypercholesterolemia (LDLR-related)	LDLR
Familial hypercholesterolemia (LDLRAP1-related)	LDLRAP1
Familial hyperinsulinism (ABCC8-related)	ABCC8
Familial hyperinsulinism (KCNJ11-related)	KCNJ11
Fanconi anemia type A	FANCA
Fanconi anemia type C	FANCC
Fanconi anemia type G	FANCG
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
Glutaric acidemia type I	GCDH
Glutaric acidemia type II (ETFA-related)	ETFA
Glutaric acidemia type II (ETFDH-related)	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 III	AGL
Glycogen storage disease type IV/ Adult polyglucosan body disease	GBE1
Glycogen storage disease type V	PYGM
Glycogen storage disease type VII	PFKM
GRACILE syndrome/ BCS1L-related disorders	

(including Mitochondrial complex III deficiency,	
Bjornstad syndrome, Leigh syndrome)	BCS1L
Guanidinoacetate methyltransferase deficiency GAMT	
HBB-related hemoglobinopathies	
(including Beta-thalassemia and Sickle cell disease)	HBB
Hereditary fructose intolerance	ALDOB
Hereditary hemochromatosis (HJV-related)	HJV
Hereditary hemochromatosis (TFR2-related)	TFR2
Hermansky-Pudlak syndrome (HPS1-related)	HPS1
Hermansky-Pudlak syndrome (HPS3-related)	HPS3
Holocarboxylase synthetase deficiency	HLCS
Homocystinuria (CBS-related)	CBS
Homocystinuria due to MTHFR deficiency	MTHFR
Homocystinuria, cobalamin E type	MTRR
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
Junctional epidermolysis bullosa (LAMA3-related)	LAMA3
Junctional epidermolysis bullosa (LAMB3-related)	LAMB3
Junctional epidermolysis bullosa (LAMC2-related)	LAMC2
Krabbe disease GALC	
LAMA2-related muscular dystrophy	LAMA2
Leber congenital amaurosis 10/ CEP290-related disorders	CEP290
Leber congenital amaurosis 13	RDH12
Leber congenital amaurosis 2	RPE65
Leber congenital amaurosis 5	LCA5
Leber congenital amaurosis 8/ CRB1-related disorders	CRB1
Leigh syndrome, French Canadian type	LRPPRC

Lethal congenital contracture syndrome 1 /	
Lethal arthrogryposis with anterior horn cell disease	GLE1
Leukoencephalopathy with vanishing white matter (EIF2B5-related)	EIF2B5
Limb-girdle muscular dystrophy type 2A (calpainopathy)	CAPN3
Limb-girdle muscular dystrophy type 2B (dysferlinopathy)	DYSF
Limb-girdle muscular dystrophy type 2C	SGCG
Limb-girdle muscular dystrophy type 2D	SGCA
Limb-girdle muscular dystrophy type 2E	SGCB
Lipoid congenital adrenal hyperplasia (STAR-related)	STAR
Lipoprotein lipase deficiency	LPL
Long chain 3-hydroxyacyl-CoA dehydrogenase (LCHAD) deficiency	HADHA
Lysinuric protein intolerance	SLC7A7
Lysosomal acid lipase deficiency	
(includes Wolman disease and Cholesterol ester storage disease)	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	
(including Occipital horn syndrome and Distal hereditary motor neuropathy)	ATP7A
Metachromatic leukodystrophy (ARSA-related)	ARSA
Methylmalonic acidemia (MMAA-related)	MMAA
Methylmalonic acidemia (MMAB-related)	MMAB
Methylmalonic acidemia (MUT-related)	MUT
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 DNA depletion syndrome (MPV17-related)	MPV17

Mitochondrial myopathy and sideroblastic anemia 1	PUS1
Mitochondrial neurogastrointestinal encephalopathy (MNGIE) disease	TYMP
MKS1-related disorders	MKS1
Mucolipidosis type II/III (GNPTAB-related)	GNPTAB
Mucolipidosis type III (GNPTG-related)	GNPTG
Mucolipidosis type IV	MCOLN1
Mucopolysaccharidosis type I	
(includes Hurler, Hurler-Scheie, and Scheie syndromes)	IDUA
Mucopolysaccharidosis type II (Hunter syndrome)	IDS
Mucopolysaccharidosis type IIIA (Sanfilippo A syndrome)	SGSH
Mucopolysaccharidosis type IIIB	NAGLU
Mucopolysaccharidosis type IIIC (Sanfilippo syndrome)/ Retinitis pigmentosa 73	HGSNAT
Mucopolysaccharidosis type IIID (Sanfilippo syndrome)	GNS
Mucopolysaccharidosis type IVB (Morquio B syndrome)/ GM1 gangliosidosis	GLB1
Mucopolysaccharidosis type IX	HYAL1
Mucopolysaccharidosis type VI (Maroteaux-Lamy syndrome)	ARSB
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
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
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	PHGDH
Polycystic kidney disease (PKHD1-related)	PKHD1
Polymicrogyria (ADGRG1-related)	ADGRG1
POMGNT1-related disorders (including Muscle eye brain disease)	POMGNT1
Pontocerebellar hypoplasia (RARS2-related)	RARS2
Pontocerebellar hypoplasia (SEPSECS-related)	SEPSECS
Pontocerebellar hypoplasia (VRK1-related)	VRK1
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
PRPS1-related disorders	
(including Charcot-Marie-Tooth disease type 5 and Arts syndrome)	PRPS1
Pycnodysostosis	CTSK
Pyruvate carboxylase deficiency	PC
Pyruvate dehydrogenase deficiency (PDHA1-related)	PDHA1
Pyruvate dehydrogenase deficiency (PDHB-related)	PDHB

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
RPGRIP1L-related disorders	
(including Joubert syndrome 7, COACH syndrome and Meckel syndrome 5)	RPGRIP1L
RTEL-1-related disorders (including Dyskeratosis congenita)	RTEL1
Sandhoff disease	HEXB
Schimke immuno-osseous dysplasia	SMARCAL1
Severe combined immune deficiency (DCLRE1C-related)	DCLRE1C
Severe combined immunodeficiency/ Omenn syndrome (RAG2-related)	RAG2
Severe congenital neutropenia (VPS45-related)	VPS45
Sialic acid storage disorders	SLC17A5
Sjögren-Larsson syndrome	ALDH3A2
SLC26A2-related disorders	
(including Diatrophic dysplasia, Atelosteogenesis type 2,	
Achondrogenesis type 1B/ Multiple metaphyseal dysplasia)	SLC26A2
SLC35A3-related disorder	SLC35A3
Smith-Lemli-Opitz syndrome	DHCR7
Spastic paraplegia type 15	ZFYVE26
Spastic paraplegia type 49	TECPR2
Spinal muscular atrophySMN1	
Spondylothoracic dysostosis	MESP2
Steel Syndrome	COL27A1
Stüve-Wiedemann syndrome	LIFR
Tay-Sachs disease/ Hexosaminidase A deficiency	HEXA
Tetrahydrobiopterin deficiency (PTS-related)	PTS
Transient infantile liver failure (TRMU-related)	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
Walker-Warburg syndrome/ FKRP-related disorders	FKRP
Walker-Warburg syndrome/ FKTN-related disorders	FKTN
Wilson disease	ATP7B
WNT10A-related disorders	
(including Odonto-onycho-dermal dysplasia and	
Schopf-Schulz-Passarge syndrome)	WNT10A
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
Xeroderma pigmentosum complementation group A	XPA
Xeroderma pigmentosum complementation group C	XPC
Zellweger spectrum disorder (PEX1-related)	PEX1
Zellweger spectrum disorder (PEX10-related)	PEX10
Zellweger spectrum disorder (PEX12-related)	PEX12
Zellweger spectrum disorder (PEX2-related)	PEX2
Zellweger spectrum disorder (PEX6-related)	PEX6