3-hydroxy-3-methylglutarayl-CoA (HMG-CoA) lyase deficiency
3-methylcrotonyl-CoA carboxylase (3-MCC) deficiency (MCC1-related)
3-methylcrotonyl-CoA carboxylase (3-MCC) deficiency (MCC2-related)
ABCC8-related disorders
Abetalipoproteinemia
ACAD9 deficiency
Achromatopsia (CNGB3-related)
Acrodermatitis enteropathica
Adenosine deaminase deficiency
Aicardi-Goutieres syndrome (SAMHD1-related)
Aldosterone synthase deficiency
Alkaptonuria
Alpha-1 antitrypsin deficiency
Alpha-mannosidosis
Alpha-thalassemia
Alpha-thalassemia X-linked intellectual disability syndrome
Alport Syndrome (COL4A3-related)
Alport Syndrome (COL4A4-related)
Alport Syndrome, X-linked (COL4A5-related)
Alström syndrome
Andermann syndrome
Arginase deficiency
Argininosuccinic aciduria
Aromatase deficiency
Asparagine synthetase deficiency
Aspartylglucosaminuria
Ataxia with vitamin E deficiency
Ataxia-telangiectasia
Autoimmune polyendocrinopathy with candidiasis and ectodermal dysplasia
Autosomal recessive deafness 77
Autosomal recessive spastic ataxia of Charlevoix-Saguenay (ARSACS)

Bardet-Biedl syndrome (BBS10-related)
Bardet-Biedl syndrome (BBS12-related)
Bartter syndrome type 4A
BBS1-related disorders
BBS2-related disorders
Bernard-Soulier syndrome (GP1BA-related)
Bernard-Soulier syndrome (GP9-related)
Beta-ketothiolase deficiency
Biotinidase deficiency
Bloom syndrome
Canavan disease
Carbamoylphosphate synthetase I deficiency
Carnitine palmitoyltransferase I deficiency
Carnitine palmitoyltransferase II deficiency
Carpenter syndrome (RAB23-related)
Cartilage-hair hypoplasia-anauxetic dysplasia spectrum disorders
Cerebrotendinous xanthomatosis
CFTR-related disorders (including cystic fibrosis)
Charcot-Marie-Tooth disease (NDRG1-related)
Charcot-Marie-Tooth disease, X-linked (GJB1-related)
Chorea-acanthocytosis
Choroideremia
Chronic granulomatous disease (CYBA-related)
Chronic granulomatous disease (CYBB-related)
Citrin deficiency
Citrullinemia type 1
Cockayne syndrome type A
Cockayne syndrome type B
Cohen syndrome
Combined malonic and methylmalonic aciduria (ACSF3-related)
Combined oxidative phosphorylation deficiency (GFM1-related)
Combined oxidative phosphorylation deficiency (TSFM-related)
Combined pituitary hormone deficiency (LHX3-related)

Combined pituitary hormone deficiency (PROP1-related)
Congenital adrenal hyperplasia due to 11-beta-hydroxylase-deficiency
Congenital adrenal hyperplasia due to 21-hydroxylase deficiency
Congenital adrenal hyperplasia due to 3-beta-hydroxysteroid dehydrogenase type II deficiency
Congenital amegakaryocytic thrombocytopenia
Congenital disorder of glycosylation (ALG6-related)
Congenital disorder of glycosylation (MPI-related)
Congenital disorder of glycosylation (PMM2-related)
Congenital ichthyosis (TGM1-related)
Congenital insensitivity to pain with anhidrosis
Congenital myasthenic syndrome (CHRNE-related)
Corneal dystrophy and perceptive deafness
CYP17A1-related disorders
Cystinosis
DHDDS-related disorders
Dihydrolipoamide dehydrogenase deficiency (DLD)
DMD-related dystrophinopathy
Dysferlinopathy
Dystrophic epidermolysis bullosa (COL7A1-related)
Ehlers-Danlos syndrome, dermatosparaxis type
Ellis-van Creveld syndrome (EVC2-related)
Ellis-van Creveld syndrome (EVC-related)
Emery-Dreifuss muscular dystrophy (EMD-related)
Enhanced S-cone syndrome/ retinitis pigmentosa 37
Ethylmalonic encephalopathy
Fabry disease
Factor IX deficiency (Hemophilia B)
Factor V Leiden thrombophilia
Factor XI deficiency (Hemophilia C)
Familial dysautonomia
Familial hypercholesterolemia (LDLRAP1-related)
Familial hypercholesterolemia (LDLR-related)

Familial mediterranean fever
Fanconi anemia type A
Fanconi anemia type C
Fanconi anemia type G
FKRP-related disorders
FKTN-related disorders
Fragile X syndrome
Fumarate hydratase deficiency
Galactokinase deficiency galactosemia
Galactosemia (GALT-related)
Gaucher disease
Gitelman syndrome (SLC12A3-related)
GJB2-related DFNB1 nonsyndromic hearing loss and deafness
GLE1-related disorders
Glucose-6-phosphate dehydrogenase (G6PD) deficiency
Glutaric acidemia type I
Glutaric acidemia type IIA
Glutaric acidemia type IIC
Glycine encephalopathy (AMT-related)
Glycine encephalopathy (GLDC-related)
Glycogen storage disease type Ia
Glycogen storage disease type Ib
Glycogen storage disease type II (Pompe disease)
Glycogen storage disease type IV/ adult polyglucosan body disease
Glycogen storage disease type V
Glycogen storage disease type III
Glycogen storage disease type VII
GRACILE syndrome/ BCS1L-related disorders
Guanidinoacetate methyltransferase deficiency
HBB-related hemoglobinopathies
Hereditary fructose intolerance
Hereditary hemochromatosis (HFE-related)
Hereditary hemochromatosis type 2 (HJV-related)

Hereditary hemochromatosis type 3
Hermansky-Pudlak syndrome type 1
Hermansky-Pudlak syndrome type 3
Holocarboxylase synthetase deficiency
Homocystinuria due to CBS deficiency
Homocystinuria due to MTHFR deficiency
Homocystinuria, cobalamin E type
HSD17B4-related disorders
Hydrolethalus syndrome type 1
Hyperornithinemia-hyperammonemia-homocitrullinuria (HHH) syndrome
Hypohidrotic ectodermal dysplasia (EDA-related)
Hypophosphatasia
Inclusion body myopathy 2
Isovaleric acidemia
Joubert syndrome 2/ TMEM216-related disorders
Junctional epidermolysis bullosa (LAMB3-related)
Junctional epidermolysis bullosa (LAMC2-related)
KCNJ11-related disorders
Krabbe disease
LAMA2-related muscular dystrophy
LAMA3-related disorders
Leber congenital amaurosis 10/ CEP290-related disorders
Leber congenital amaurosis 13
Leber congenital amaurosis 5
Leber congenital amaurosis 8/ CRB1-related disorders
Leigh syndrome, French Canadian type
Leukoencephalopathy with vanishing white matter (EIF2B5-related)
Limb-girdle muscular dystrophy type 2A (calpainopathy)
Limb-girdle muscular dystrophy type 2C
Limb-girdle muscular dystrophy type 2D
Limb-girdle muscular dystrophy type 2E
Lipoid congenital adrenal hyperplasia
Lipoprotein lipase deficiency

Long chain 3-hydroxyacyl-CoA dehydrogenase (LCHAD) deficiency
Lysinuric protein intolerance
Lysosomal acid lipase deficiency
Major histocompatibility complex class II deficiency (CIITA-related)
Maple syrup urine disease (MSUD) type 1A
Maple syrup urine disease (MSUD) type 1B
Maple syrup urine disease (MSUD) type 2
Medium chain acyl-CoA dehydrogenase (MCAD) deficiency
Megalencephalic leukoencephalopathy with subcortical cysts type 1
Menkes disease/ ATP7A-related disorders
Metachromatic leukodystrophy (ARSA-related)
Methylmalonic acidemia (MMAA-related)
Methylmalonic acidemia (MMAB-related)
Methylmalonic acidemia (MUT-related)
Methylmalonic acidemia with homocystinuria, cobalamin C type
Methylmalonic acidemia with homocystinuria, cobalamin D type
Microphthalmia /clinical anophthalmia (VSX2-related)
Mitochondrial complex I deficiency/ Leigh syndrome (NDUFAF5-related)
Mitochondrial complex I deficiency/ Leigh syndrome (NDUFS6-related)
Mitochondrial myopathy and sideroblastic anemia 1
Mitochondrial neurogastrointestinal encephalopathy (MNGIE) disease
Mitochondrial DNA depletion syndrome (MPV17-related)
MKS1-related disorders
Mucolipidosis type II/III (GNPTAB-related)
Mucolipidosis type IV
Mucolipidosis type III (GNPTG-related)
Mucopolysaccharidosis type I
Mucopolysaccharidosis type II (Hunter syndrome)
Mucopolysaccharidosis type IX
Mucopolysaccharidosis type VI (Maroteaux-Lamy syndrome)
Mucopolysaccharidosis type IIIA (Sanfilippo A syndrome)

Mucopolysaccharidosis type IIIB (Sanfilippo B syndrome)
Mucopolysaccharidosis type IIIC (Sanfilippo C syndrome)/ retinitis pigmentosa 73
Mucopolysaccharidosis type IIID (Sanfilippo D syndrome)
Mucopolysaccharidosis type IVB (Morquio B syndrome)/ GM1 gangliosidosis
Multiple sulfatase deficiency
N-Acetylglutamate synthase deficiency
Nemaline myopathy 2
Nephrogenic diabetes insipidus (AQP2-related)
Nephrotic syndrome/ congenital Finnish nephrosis (NPHS1-related)
Nephrotic syndrome/steroid-resistant nephrotic syndrome (NPHS2-related)
Neuronal ceroid lipofuscinosis (TPP1-related)
Neuronal ceroid-lipofuscinosis (CLN3-related)
Neuronal ceroid-lipofuscinosis (CLN5-related)
Neuronal ceroid-lipofuscinosis (CLN6-related)
Neuronal ceroid-lipofuscinosis (MFSD8-related)
Neuronal ceroid-lipofuscinosis (PPT1-related)
Neuronal ceroid-lipofuscinosis/ Northern epilepsy (CLN8-related)
Niemann-Pick disease type A/B
Niemann-Pick disease type C (NPC1-related)
Niemann-Pick disease type C (NPC2-related)
Nijmegen breakage syndrome
OPA3-related conditions
Ornithine aminotransferase deficiency
Ornithine transcarbamylase (OTC) deficiency
Osteopetrosis (TCIRG1-related)
Pendred syndrome
Peroxisomal acyl-CoA oxidase deficiency
Phenylalanine hydroxylase deficiency (including Phenylketonuria (PKU))
Phosphoglycerate dehydrogenase deficiency/ Neu-Laxova syndrome type 1
Polycystic kidney disease (PKHD1-related)
Polymicrogyria (ADGRG1-related)

POMGNT1-related disorders
Pontocerebellar hypoplasia (RARS2-related)
Pontocerebellar hypoplasia (SEPSECS-related)
Postnatal progressive microcephaly with seizures and brain atrophy/ Infantile cerebral and cerebellar atrophy (MED17-related)
Primary carnitine deficiency
Primary Ciliary Dyskinesia (DNAH5-related)
Primary Ciliary Dyskinesia (DNAI1-related)
Primary Ciliary Dyskinesia (DNAI2-related)
Primary hyperoxaluria type 1
Primary hyperoxaluria type 2
Primary hyperoxaluria type 3
Progressive familial intrahepatic cholestasis type 2
Propionic acidemia (PCCA-related)
Propionic acidemia (PCCB-related)
Prothrombin-related thrombophilia
PRPS1-related disorders
PSAP-related disorders
Pycnodysostosis
Pyruvate carboxylase deficiency
Pyruvate dehydrogenase complex deficiency (PDHA1-related)
Pyruvate dehydrogenase complex deficiency (PDHB-related)
RAPSN-related disorders
Renal tubular acidosis with deafness (ATP6V1B1-related)
Retinitis pigmentosa 25
Retinitis pigmentosa 26
Retinitis Pigmentosa 28
Rhizomelic chondrodysplasia punctata type 1/ Refsum disease (PEX7-related)
Rhizomelic chondrodysplasia punctata type 3
Roberts syndrome
RPE65-related disorders
RPGRIP1L-related disorders
RTEL-1-related disorders

Sandhoff disease
Schimke immuno-osseous dysplasia
Severe combined immune deficiency (DCLRE1C-related)
Severe combined immunodeficiency (RAG2-related)
Severe congenital neutropenia due to VPS45-deficiency
Severe congenital neutropenia type 3
Sialic acid storage disorders
Sjögren-Larsson syndrome
SLC26A2-related disorders
SLC35A3-related disorders
Smith-Lemli-Opitz syndrome
Spastic paraplegia type 15
Spastic paraplegia type 49
Spinal muscular atrophy
Spondylothoracic dysostosis
Steel Syndrome
Stüve-Wiedemann syndrome
Tay-Sachs disease/ hexosaminidase A deficiency
Tetrahydrobiopterin deficiency (PTS-related)
Transient infantile liver failure
Tyrosine hydroxylase deficiency
Tyrosinemia type I
Tyrosinemia type II
Usher syndrome type IB/ MYO7A-related disorders
Usher syndrome type IC/ USH1C-related disorders
Usher syndrome type ID
Usher syndrome type IF/ PCDH15-related disorders
Usher syndrome type IIA/ USH2A-related disorders
Usher syndrome type IIIA
Very long-chain acyl-CoA dehydrogenase (VLCAD) deficiency
VRK1-related disorders
Wilson disease
WNT10A-related disorders