Gene	Disease
ABCA3	Surfactant metabolism dysfunction, pulmonary, type 3
ABCC8	Hyperinsulinemic hypoglycemia, type 1 (congenital hyperinsulinism); Permanent neonatal diabetes mellitus (PNDM)
ABCD1	Adrenoleukodystrophy
ACADM	Medium-chain acyl-CoA dehydrogenase deficiency
ACADVL	Very long-chain acyl-CoA dehydrogenase (VLCAD) deficiency
ACAT1	Alpha-methylacetoacetic aciduria (3-ketothiolase deficiency)
AFF2	Intellectual developmental disorder, X-linked 109
AGA	Aspartylglucosaminuria (glycosylasparaginase deficiency)
AGXT	Hyperoxaluria, primary, type 1
AHI1	Joubert syndrome, type 3
AIRE	Autoimmune polyendocrinopathy syndrome, type 1
ALDOB	Fructose intolerance, hereditary
ALPL	Hypophosphatasia, infantile/childhood
ANO10	Spinocerebellar ataxia, autosomal recessive, type 10
AR	Androgen insensitivity syndrome, complete
ARSA	Metachromatic leukodystrophy
ARX	Epileptic encephalopathy, early infantile, type 1; ARX-related developmental disorders
ASL	Argininosuccinic aciduria
ASPA	Canavan disease
ATP7B	Wilson disease
BBS1	Bardet-Biedl syndrome, type 1
BBS2	Bardet-Biedl syndrome, type 2
BCKDHB	Maple syrup urine disease, type 1B
BTD	Biotinidase deficiency
CAPN3	Limb-girdle muscular dystrophy, type 1 (LGMD R1)
CBS	Homocystinuria due to cystathionine beta-synthase
CC2D2A	Joubert syndrome, type 9; Meckel syndrome, type 6; COACH syndrome, 2
CCDC88C	Hydrocephalus, congenital, type 1
CEP290	Meckel syndrome, type 4; Joubert syndrome, type 5; Leber congenital amaurosis, type 10
CFTR	Cystic fibrosis
CHRNE	Myasthenic syndrome, congenital, type 4B, fast-channel; Myasthenic syndrome, congenital, type 4C, associated with acetylcholine receptor deficiency

Gene	Disease
CLCN1	Myotonia congenita, recessive
CNGB3	Achromatopsia, type 3
COL4A3	Alport syndrome, autosomal recessive, type 2
COL4A4	Alport syndrome, autosomal recessive, type 2
COL7A1	Dystrophic epidermolysis bullosa (DEB), Hallopeau-Siemens (HS) type and non-HS type; DEB pruriginosa; DEB pretibial
CPT2	Carnitine palmitoyltransferase type 2 deficiency, lethal neonatal; Carnitine palmitoyltransferase type 2 deficiency, infantile
CRB1	Retinitis pigmentosa, type 12; Leber congenital amaurosis, type 8
CYP11A1	46,XY disorder of sex development-adrenal insufficiency due to CYP11A1 deficiency
CYP21A2	Congenital adrenal hyperplasia due to 21-hydroxylase deficiency
CYP27A1	Cerebrotendinous xanthomatosis
CYP27B1	Vitamin D-dependent rickets, type 1
DCLRE1C	Omenn syndrome; Severe combined immunodeficiency, Athabascan type
DHCR7	Smith-Lemli-Opitz syndrome
DHDDS	Retinitis pigmentosa, type 59
DMD	Duchenne/Becker muscular dystrophy
DYNC2H1	Short-rib thoracic dysplasia, type 3 with or without polydactyly
ERCC2	Trichothiodystrophy, type 1
EVC2	Ellis-van Creveld syndrome
F8	Hemophilia A
F9	Hemophilia B
FAH	Tyrosinemia, type 1
FANCC	Fanconi anemia, complementation group C
FKRP	Muscular dystrophy-dystroglycanopathy, type 5A (Walker-Warburg syndrome); Type 5B; Type 5C (limb-girdle muscular dystrophy, type 9 [LGMDR9])
FKTN	Muscular dystrophy-dystroglycanopathy, type 4A (Walker-Warburg syndrome); Type 4B; Type 4C (limb-girdle muscular dystrophy, type 13 [LGMD R13])
FMO3	Trimethylaminuria
FMR1	Fragile X syndrome
G6PC	Glycogen storage disease, type 1A
G6PD	Hemolytic anemia, G6PD deficient (favism)
GAA	Glycogen storage disease, type 2
GALNS	Mucopolysaccharidosis, type 4A

Gene	Disease
GALT	Galactosemia
GBA	Gaucher Disease
GBE1	Glycogen storage disease, type 4
GJB2	Deafness, autosomal recessive, type 1A; Deafness, digenic, GJB2/GJB6
GJB6	Deafness, autosomal recessive, type 1B; Deafness, digenic GJB2/GJB6
GLA	Fabry disease
GNPTAB	Mucolipidosis 2 alpha/beta; Mucolipidosis 3 alpha/beta
GNRHR	Hypogonadotropic hypogonadism, type 7 without anosmia
GRIP1	Fraser syndrome 3
HBA1	Alpha-thalassemia
HBA2	Alpha-thalassemia
HBB	Beta-thalassemia, Sickle cell anemia and other HBB-related hemoglobinopathies
HEXA	Tay-Sachs disease
HGSNAT	Mucopolysaccharidosis type 3C (Sanfilippo syndrome C)
IDUA	Mucopolysaccharidosis type 1
L1CAM	L1 Syndrome
LRP2	Donnai-Barrow syndrome
MCCC2	3-Methylcrotonyl-CoA carboxylase deficiency, type 2
MCOLN1	Mucolipidosis type 4
MCPH1	Microcephaly type 1 primary, autosomal recessive
MEFV	Familial Mediterranean fever
MID1	Opitz GBBB syndrome, type 1
MLC1	Megalencephalic leukoencephalopathy with subcortical cysts
MMACHC	Methylmalonic aciduria and homocystinuria, cblC type
MMUT	Methylmalonic aciduria, mut(0) type
MVK	Mevalonic aciduria
MYO7A	Usher syndrome, type 1B; Deafness, autosomal recessive, type 2
NAGA	Schindler disease, type I
NPHS1	Nephrotic syndrome, type 1
NR0B1	Adrenal hypoplasia, congenital
OCA2	Oculocutaneous albinism type 2
OTC	Ornithine transcarbamylase deficiency
OTOF	Deafness, autosomal recessive, type 9
PAH	Phenylketonuria
PCDH15	Deafness, autosomal recessive, type 23; Usher syndrome, type 1D/F digenic

Gene	Disease
PKHD1	Polycystic kidney disease type 4
PLP1	Pelizaeus-Merzbacher disease
PMM2	Congenital disorder of glycosylation, type 1A
POLG	POLG-related disorders
PRF1	Hemophagocytic lymphohistiocytosis, familial, type 2
RARS2	Pontocerebellar hypoplasia, type 6
RNASEH2B	Aicardi-Goutieres syndrome, type 2
RPGR	Retinitis pigmentosa, type 3 X-linked; Cone-rod dystrophy, X-linked, 1
RS1	Retinoschisis
SAG	Oguchi disease, type 1
SCO2	Mitochondrial complex IV deficiency, nuclear type 2
SERPINA1	Alpha-1 antitrypsin deficiency
SLC19A3	Thiamine metabolism dysfunction syndrome, type 2 (biotin- or thiamine-responsive encephalopathy type)
SLC26A2	Achondrogenesis, type 1B (diastrophic dysplasia)
SLC26A4	Deafness, autosomal recessive, type 4; Pendred syndrome
SLC37A4	Glycogen storage disease, type 1B
SLC6A8	Cerebral creatine deficiency syndrome, type 1
SMN1	Spinal muscular atrophy
SMPD1	Niemann-Pick disease, type A; Niemann-Pick disease, type B
SPG7	Spastic paraplegia, type 7 autosomal recessive
TF	Atransferrinemia
TMEM216	Joubert syndrome, type 2; Meckel syndrome, type 2
TMPRSS3	Deafness, autosomal recessive, type 45573
TNXB	Ehlers-Danlos syndrome, classic-like
TSHR	Hypothyroidism, congenital, nongoitrous, type 1
TYR	Oculocutaneous albinism (OCA) type 1A; OCA type 1B
USH2A	Usher syndrome, type 2A
XPC	Xeroderma pigmentosum, group C