CGT Exome version 2.0

Gene	Disease
AAAS	Triple-A syndrome (achalasia-addisonianism-alacrimia)
AARS	Epileptic encephalopathy, early infantile, type 29
AARS2	Combined oxidative phosphorylation deficiency 8; Leukoencephalopathy, progressive, with ovarian failure
AASS	Hyperlysinemia, type 1 and type 2
ABAT	GABA-transaminase deficiency
ABCA1	Tangier disease
ABCA12	Ichthyosis, congenital, autosomal recessive, type 4A; ICAR, type 4B (harlequin)
ABCA3	Surfactant metabolism dysfunction, pulmonary, type 3
ABCA4	Stargardt disease type 1; Cone-rod dystrophy type 3
ABCB11	Cholestasis, benign recurrent intrahepatic, type 2; Cholestasis, progressive familial intrahepatic, type 2
ABCB4	Cholestasis, progressive familial intrahepatic, type 3
ABCB7	X-linked sideroblastic anemia and ataxia (XLSA/A)
ABCC6	Arterial calcification, generalized, of infancy, type 2
ABCC8	Hyperinsulinemic hypoglycemia, type 1 (congenital hyperinsulinism); Permanent neonatal diabetes mellitus (PNDM)
ABCD1	Adrenoleukodystrophy
ABCD4	Methylmalonic aciduria and homocystinuria, cblJ type
ABCG5	Sitosterolemia
ABCG8	Sitosterolemia
ABHD12	PHARC syndrome (polyneuropathy, hearing loss, ataxia, retinitis pigmentosa and cataract)
ABHD5	Chanarin-Dorfman syndrome
ACAD8	Isobutyryl-CoA dehydrogenase deficiency
ACAD9	Acyl-CoA dehydrogenase 9 deficiency (mitochondrial complex I deficiency, nuclear, type 20)
ACADM	Medium-chain acyl-CoA dehydrogenase deficiency
ACADS	Short-chain acyl-CoA dehydrogenase deficiency
ACADSB	Short/branched-chain acyl-CoA dehydrogenase deficiency
ACADVL	Very long-chain acyl-CoA dehydrogenase deficiency
ACAT1	Alpha-methylacetoacetic aciduria (3-ketothiolase deficiency)
ACE	Renal tubular dysgenesis
ACO2	Infantile cerebellar-retinal degeneration
ACOX1	Peroxisomal acyl-CoA oxidase deficiency
ACP5	Spondyloenchondrodysplasia with immune dysregulation

ACSF3	Combined malonic and methylmalonic aciduria
ACSL4	Mental retardation, X-linked, type 63
ACTA1	Nemaline myopathy 3; Congenital fiber-type disproportion myopathy 1
ACY1	Aminoacylase 1 deficiency
ADA	Severe combined immunodeficiency due to adenosine deaminase deficiency (ADA)
ADAM9	Cone-rod dystrophy 9
ADAMTS10	Weill-Marchesani syndrome, type 1, recessive
ADAMTS13	Thrombotic thrombocytopenic purpura, familial (Schulman-Upshaw syndrome)
ADAMTS17	Weill-Marchesani syndrome, type 4, recessive
ADAMTS18	Microcornea, myopic chorioretinal atrophy, and telecanthus
ADAMTS2	Ehlers-Danlos syndrome, dermatosparaxis type
ADAMTSL2	Geleophysic dysplasia type 1
ADAMTSL4	Ectopia lentis et pupillae; Ectopia lentis, isolated
ADAR	Aicardi-Goutieres syndrome 6
ADGRG1	Polymicrogyria, bilateral frontoparietal
ADGRV1	Usher syndrome, type 2C
ADK	Hypermethioninemia due to adenosine kinase deficiency
ADSL	Adenylosuccinase deficiency
AFF2	Mental retardation, X-linked, FRAXE type
AFG3L2	Spastic ataxia, type 5, autosomal recessive
AGA	Aspartylglucosaminuria (glycosylasparaginase deficiency)
AGK	Cataract 38, autosomal recessive; Sengers syndrome
AGL	Glycogen storage disease, type 3
AGPAT2	Congenital generalized lipodystrophy (Berardinelli-Seip syndrome)
AGPS	Rhizomelic chondrodysplasia punctata, type 3
AGRN	Myasthenic syndrome, congenital, type 8
AGT	Renal tubular dysgenesis
AGTR1	Renal tubular dysgenesis
AGXT	Hyperoxaluria, primary, type 1
AHCY	Hypermethioninemia with deficiency of S-adenosylhomocysteine hydrolase
AHI1	Joubert syndrome, type 3
AICDA	Immunodeficiency with hyper-IgM, type 2
AIFM1	Cowchock syndrome; Deafness, X-linked, type 5
AIMP1	Leukodystrophy, hypomyelinating, type 3
AIPL1	Leber congenital amaurosis type 4

AIRE	Autoimmune polyendocrinopathy syndrome type 1
AK1	Hemolytic anemia due to adenylate kinase deficiency
AK2	Reticular dysgenesis
AKR1C2	46,XY disorder of sex development due to testicular 17,20-desmolase deficiency
AKR1D1	Bile acid synthesis defect, congenital, type 2
ALAD	Porphyria, acute hepatic
ALAS2	X-linked sideroblastic anemia, type 1 (XLSA or SIDBA1)
ALDH18A1	Spastic paraplegia, type 9B, autosomal recessive; De Barsy syndrome
ALDH3A2	Sjogren-Larsson syndrome
ALDH4A1	Hyperprolinemia, type 2
ALDH5A1	Succinic semialdehyde dehydrogenase deficiency
ALDH6A1	Methylmalonate semialdehyde dehydrogenase deficiency
ALDH7A1	Epilepsy, pyridoxine-dependent
ALDOA	Glycogen storage disease type 12
ALDOB	Fructose intolerance, hereditary
ALG1	Congenital disorder of glycosylation, type 1K
ALG11	Congenital disorder of glycosylation, type 1P
ALG12	Congenital disorder of glycosylation, type 1G
ALG13	Epileptic encephalopathy, early infantile, type 36
ALG2	Myasthenic syndrome, congenital, type 14, with tubular aggregates
ALG6	Congenital disorder of glycosylation, type 1C
ALG8	Congenital disorder of glycosylation, type 1H
ALG9	Congenital disorder of glycosylation, type 1L; Gillessen-Kaesbach-Nishimura syndrome
ALMS1	Alstr?m syndrome
ALOX12B	Ichthyosis, congenital, autosomal recessive, type 2
ALOXE3	Ichthyosis, congenital, autosomal recessive, type 3
ALPL	Hypophosphatasia, childhood/infantile
ALS2	Amyotrophic lateral sclerosis, type 2, juvenile; Primary lateral sclerosis, juvenile; Spastic paralysis, infantile onset ascending
ALX1	Frontonasal dysplasia, type 3
ALX3	Frontonasal dysplasia, type 1
ALX4	Frontonasal dysplasia, type 2
AMACR	Bile acid synthesis defect, congenital, type 4; Alpha-methylacyl-CoA racemase deficiency
AMELX	Amelogenesis imperfecta, type 1E
AMER1	Osteopathia striata with cranial sclerosis
AMH	Persistent Mullerian duct syndrome, type 1

AMN	Megaloblastic anemia 1 (Imerslund-Grasbeck syndrome)
AMPD1	Myopathy due to myoadenylate deaminase deficiency
AMT	Glycine encephalopathy
ANGPTL3	Hypobetalipoproteinemia, familial, type 2
ANO10	Spinocerebellar ataxia, autosomal recessive, type 10
ANO5	Limb-girdle muscular dystrophy, type 12 (LGMD R12)
ANOS1	Hypogonadotropic hypogonadism, type 1, with or without anosmia (Kallmann syndrome 1)
ANTXR1	GAPO syndrome
ANTXR2	Hyaline fibromatosis syndrome
AP1S1	MEDNIK syndrome
AP1S2	Mental retardation, X-linked, syndromic, type 5 (Pettigrew syndrome)
AP3B1	Hermansky-Pudlak syndrome, type 2
AP3B2	Epileptic encephalopathy, early infantile, type 48
AP4B1	Spastic paraplegia, type 47, autosomal recessive
AP4M1	Spastic paraplegia, type 50, autosomal recessive
AP4S1	Spastic paraplegia, type 52, autosomal recessive
AP5Z1	Spastic paraplegia, type 48, autosomal recessive
APOC2	Hyperlipoproteinemia, type 1B
APOE	Sea-blue histiocyte disease
APRT	Adenine phosphoribosyltransferase deficiency
APTX	Ataxia, early-onset, with oculomotor apraxia and hypoalbuminemia
AQP2	Diabetes insipidus, nephrogenic, type 2
AR	Androgen insensitivity syndrome, complete
ARFGEF2	Periventricular heterotopia with microcephaly
ARG1	Argininemia (arginase deficiency)
ARHGEF9	Epileptic encephalopathy, early infantile, type 8
ARL13B	Joubert syndrome type 8
ARL6	Bardet-Biedl syndrome, type 3
ARSA	Metachromatic leukodystrophy
ARSB	Mucopolysaccharidosis, type 6 (Maroteaux-Lamy syndrome)
ARSE	Chondrodysplasia punctata, brachytelephalangic
ARX	Epileptic encephalopathy, early infantile, type 1; ARX-related developmental disorders
ASAH1	Farber lipogranulomatosis; Spinal muscular atrophy with progressive myoclonic epilepsy
ASL	Argininosuccinic aciduria
ASNS	Asparagine synthetase deficiency

ASPA	Canavan disease
ASPM	Primary microcephaly type 5, autosomal recessive
ASS1	Citrullinemia, type 1
ATF6	Achromatopsia 7
ATIC	AICA-ribosiduria due to ATIC deficiency
ATM	Ataxia-telangiectasia
ATOH7	Persistent hyperplastic primary vitreous, autosomal recessive
ATP13A2	Kufor-Rakeb syndrome; Spastic paraplegia, type 78, autosomal recessive
ATP2A1	Brody myopathy
ATP6V0A2	Cutis laxa, autosomal recessive, type 2A; Wrinkly skin syndrome
ATP6V0A4	Renal tubular acidosis, distal, autosomal recessive
ATP6V1B1	Renal tubular acidosis with deafness
ATP7A	Menkes disease; Occipital horn syndrome
ATP7B	Wilson disease
ATP8B1	Cholestasis, progressive familial intrahepatic, type 1; Cholestasis, benign recurrent intrahepatic, type 1
ATR	Seckel syndrome type 1
ATRX	Mental retardation-hypotonic facies syndrome, X-linked
AUH	3-methylglutaconic aciduria, type 1
AURKC	Male infertility spermatogenic failure, type 5
AVPR2	Diabetes insipidus, nephrogenic, type 1; Nephrogenic syndrome of inappropriate antidiuresis (NSIAD)
B2M	Immunodeficiency, type 43
B3GAT3	Multiple joint dislocations, short stature, craniofacial dysmorphism, with or without congenital heart defects
B3GLCT	Peters-plus syndrome
B4GALT1	Congenital disorder of glycosylation, type 2D
B4GALT7	Ehlers-Danlos syndrome, spondylodysplastic, type 1
B9D1	Joubert syndrome, type 27
B9D2	Joubert syndrome type 34; Meckel syndrome type 10
BBS1	Bardet-Biedl syndrome, type 1
BBS10	Bardet-Biedl syndrome, type 10
BBS12	Bardet-Biedl syndrome, type 12
BBS2	Bardet-Biedl syndrome, type 2
BBS4	Bardet-Biedl syndrome, type 4
BBS5	Bardet-Biedl syndrome, type 5
BBS7	Bardet-Biedl syndrome, type 7
BBS9	Bardet-Biedl syndrome, type 9

BCAP31	Deafness, dystonia, and cerebral hypomyelination
BCKDHA	Maple syrup urine disease, type 1A
BCKDHB	Maple syrup urine disease, type 1B
BCOR	Microphthalmia, syndromic, type 2
BCS1L	BCS1L-related disorders, including Leigh syndrome
BEST1	Bestrophinopathy, AR
BHLHA9	Syndactyly, mesoaxial synostotic, with phalangeal reduction
BIN1	Centronuclear myopathy, type 2
BLM	Bloom syndrome
BLVRA	Hyperbiliverdinemia
BMP1	Osteogenesis imperfecta, type 13
BMP15	Ovarian dysgenesis 2
BMPER	Diaphanospondylodysostosis
BMPR1B	Acromesomelic dysplasia, Demirhan type
BOLA3	Multiple mitochondrial dysfunctions syndrome 2 with hyperglycinemia
BPGM	Erythrocytosis due to bisphosphoglycerate mutase deficiency
BRAT1	Rigidity and multifocal seizure syndrome, lethal neonatal; Neurodevelopmental disorder with cerebellar atrophy and with or without seizures
BRIP1	Fanconi anemia, complementation group J
BRWD3	Mental retardation, X-linked, type 93
BSCL2	Congenital generalized lipodystrophy, type 2; Encephalopathy, progressive, with or without lipodystrophy
BSND	Bartter syndrome, type 4A
BTD	Biotinidase deficiency
BTK	Agammaglobulinemia X-linked, type 1
BUB1B	Mosaic variegated aneuploidy syndrome 1
C12orf57	Temtamy syndrome
C12orf65	Combined oxidative phosphorylation deficiency 7; Spastic paraplegia, type 55, autosomal recessive
C19orf12	Neurodegeneration with brain iron accumulation, type 4
C1QA	C1q deficiency
C1QB	C1q deficiency
C1QC	C1q deficiency
C1S	C1s deficiency
C3	Complement component 3 deficiency
C5	Complement component 5 deficiency
C7	Complement component 7 deficiency
C8B	Complement component 8 deficiency, type 2

C8orf37	Bardet-Biedl syndrome, type 21; Cone-rod dystrophy 16 and Retintis pigmentosa 64
CA12	Hyperchlorhidrosis, isolated
CA2	Osteopetrosis with renal tubular acidosis (osteopetrosis, autosomal recessive, type 3)
CABP4	Congenital stationary night blindness, type 2B
CACNA1D	Sinoatrial node dysfunction and deafness
CACNA1F	Cone-rod dystrophy, X-linked, type 3; Night blindness, congenital stationary, type 2A; Aland Island eye disease
CANT1	Desbuquois dysplasia, type 1; Epiphyseal dysplasia, multiple, type 7
CAPN3	Limb-girdle muscular dystrophy, type 1 (LGMD R1)
CARD9	Candidiasis, familial, type 2, autosomal recessive
CASK	Mental retardation, X-linked, syndromic, Najm type
CASQ2	Ventricular tachycardia, catecholaminergic polymorphic, type 2
CASR	Hyperparathyroidism, neonatal
CAST	Peeling skin with leukonychia, acral punctate keratoses, cheilitis, and knuckle pads
CATSPER1	Male infertility spermatogenic failure, type 7
CAVIN1	Lipodystrophy, congenital generalized, type 4
CBS	Homocystinuria due to cystathionine beta-synthase
CC2D1A	Mental retardation, autosomal recessive, type 3
CC2D2A	Joubert syndrome type 9
CCBE1	Hennekam lymphangiectasia-lymphedema syndrome, type 1
CCDC103	Ciliary dyskinesia, primary, type 17
CCDC39	Ciliary dyskinesia, primary, type 14
CCDC40	Ciliary dyskinesia, primary, type 15
CCDC88C	Hydrocephalus, congenital, type 1
CD19	Immunodeficiency, common variable, type 3
CD27	Lymphoproliferative syndrome 2
CD3D	Immunodeficiency, type 19
CD3E	Immunodeficiency, type 18
CD3G	Immunodeficiency, type 17, CD3 gamma deficient
CD40	Immunodeficiency with hyper-IgM, type 3
CD40LG	Hyper-IgM syndrome, type 1 (immunodeficiency, X-linked, with hyper-IgM, type 1)
CD55	Complement hyperactivation, angiopathic thrombosis, and protein-losing enteropathy (CHAPLE)
CD59	CD59 Deficiency
CD79A	Agammaglobulinemia 3
CD79B	Agammaglobulinemia 6
CD81	Immunodeficiency, common variable, type 6

CD8A	CD8 deficiency, familial
CDAN1	Dyserythropoietic anemia, congenital, type 1A
CDH23	Deafness, autosomal recessive, type 12; Usher syndrome, type 1D
CDH3	Ectodermal dysplasia, ectrodactyly, and macular dystrophy
CDHR1	Cone-rod dystrophy, type 15
CDK5RAP2	Primary microcephaly type 3, autosomal recessive
CDKL5	Epileptic encephalopathy, early infantile, type 2
CDT1	Meier-Gorlin syndrome, type 4
CENPJ	Primary microcephaly type 6, autosomal recessive
CEP135	Microcephaly 8, primary, autosomal recessive
CEP152	Primary microcephaly type 9, autosomal recessive
CEP290	Meckel syndrome, type 4; Joubert syndrome, type 5; Leber congenital amaurosis, type 10
CEP41	Joubert syndrome, type 15
CEP57	Mosaic variegated aneuploidy syndrome 2
CERKL	Retinitis pigmentosa, type 26
CFD	Complement factor D deficiency
CFH	Complement factor H deficiency
CFI	Complement factor I deficiency
CFL2	Nemaline myopathy, type 7, autosomal recessive
CFP	Properdin deficiency, X-linked
CFTR	Cystic fibrosis
CHAT	Myasthenic syndrome, congenital, type 6, presynaptic
CHKB	Muscular dystrophy, congenital, megaconial type
CHM	Choroideremia
CHRDL1	Megalocornea 1, X-linked
CHRND	Myasthenic syndrome, congenital, type 3B, fast-channel; Multiple pterygium syndrome, lethal type
CHRNE	Myasthenic syndrome, congenital, type 4B, fast-channel; Myasthenic syndrome, congenital, type 4C, associated with acetylcholine receptor deficiency
CHRNG	Multiple pterygium syndrome (MPS), Escobar type; MPS, lethal type
CHST14	Ehlers-Danlos syndrome, musculocontractural, type 1
CHST3	Spondyloepiphyseal dysplasia with congenital joint dislocations
CHST6	Macular corneal dystrophy
CHSY1	Temtamy preaxial brachydactyly syndrome
CHUK	Cocoon syndrome
CIB2	Deafness, autosomal recessive, type 48; Usher syndrome, type 1J
CIITA	Bare lymphocyte syndrome, type 2, complementation group A

CISD2	Wolfram syndrome 2
CLCF1	Cold-induced sweating syndrome 2
CLCN1	Myotonia congenita, recessive
CLCN2	Leukoencephalopathy with ataxia
CLCN5	Dent disease; Hypophosphatemic rickets
CLCN7	Osteopetrosis, autosomal recessive type 4
CLCNKA	Bartter syndrome, type 4B, digenic
CLCNKB	Bartter syndrome, type 3
CLDN1	Ichthyosis, leukocyte vacuoles, alopecia, and sclerosing cholangitis
CLDN14	Deafness type 29, autosomal recessive
CLDN16	Hypomagnesemia, type 3, renal
CLDN19	Rena hypomagnesemia type 5, with ocular involvement
CLMP	Congenital short bowel syndrome
CLN3	Ceroid lipofuscinosis, neuronal, type 3
CLN5	Ceroid lipofuscinosis, neuronal, type 5
CLN6	Ceroid lipofuscinosis, neuronal, type 6
CLN8	Ceroid lipofuscinosis, neuronal, type 8
CLRN1	Usher syndrome, type 3A
CNGA1	Retinitis pigmentosa type 49
CNGA3	Achromatopsia 2
CNGB1	Retinitis pigmentosa type 45
CNGB3	Achromatopsia, type 3
CNKSR2	Mental retardation, X-linked, syndromic, Houge type
CNNM2	Hypomagnesemia, seizures, and mental retardation
CNNM4	Jalili syndrome
CNPY3	Epileptic encephalopathy, early infantile, type 60
CNTNAP2	Pitt-Hopkins like syndrome 1
COG4	Congenital disorder of glycosylation, type 2J
COG5	Congenital disorder of glycosylation, type 2I
COG6	Congenital disorder of glycosylation, type 2L; Shaheen syndrome
COG7	Congenital disorder of glycosylation, type 2E
COG8	Congenital disorder of glycosylation, type 2H
COL11A1	Fibrochondrogenesis type 1
COL17A1	Epidermolysis bullosa, junctional, non-Herlitz type
COL18A1	Knobloch syndrome, type 1

COL25A1	Fibrosis of extraocular muscles, congenital, type 5
COL4A3	Alport syndrome, autosomal recessive, type 2
COL4A4	Alport syndrome, autosomal recessive, type 2
COL4A5	Alport syndrome, X-linked
COL6A1	Ullrich congenital muscular dystrophy, type 1; Bethlem myopathy 1
COL6A2	Ullrich congenital muscular dystrophy, type 1; Bethlem myopathy 1
COL6A3	Dystonia, type 27
COL7A1	Dystrophic epidermolysis bullosa (DEB), Hallopeau-Siemens (HS) type and non-HS type; DEB pruriginosa; DEB pretibial
COL9A1	Stickler syndrome, type 4
COLEC11	3MC syndrome, type 2
COLQ	Myasthenic syndrome, congenital, type 5
COQ2	Primary coenzyme Q10 deficiency, type 1
COQ4	Coenzyme Q10 deficiency, primary, type 7
COQ6	Coenzyme Q10 deficiency, primary, type 6
COQ8A	Primary coenzyme Q10 deficiency, type 4
COQ9	Coenzyme Q10 deficiency, primary, type 5
CORO1A	Immunodeficiency, type 8
COX10	Mitochondrial complex IV deficiency; Leigh syndrome due to mitochondrial COX4 deficiency
COX15	Cardioencephalomyopathy, fatal infantile, due to cytochrome c oxidase deficiency, type 2; Leigh syndrome due to cytochrome c oxidase deficiency
CP	Aceruloplasminemia
CPA6	Febrile seizures, familial, type 11
CPS1	Carbamoylphosphate synthetase 1 deficiency
CPT1A	Carnitine palmitoyltransferase type 1A deficiency, hepatic
CPT2	Carnitine palmitoyltransferase type 2 deficiency, lethal neonatal; Carnitine palmitoyltransferase type 2 deficiency, infantile
CR2	Immunodeficiency, common variable, type 7
CRADD	Mental retardation, autosomal recessive, type 34, with variant lissencephaly
CRB1	Retinitis pigmentosa, type 12; Leber congenital amaurosis, type 8
CRBN	Mental retardation, autosomal recessive, type 2
CRLF1	Cold-induced sweating syndrome type 1
CRTAP	Osteogenesis imperfecta, type 7
CRYAA	Cataract 9, multiple types
CRYAB	Myopathy, myofibrillar, fatal infantile hypertonic, alpha-B crystallin-related; Cataract 16, multiple types
CRYBB1	Cataract 17, multiple types
CRYBB3	Cataract 22
CSF2RB	Surfactant metabolism dysfunction, pulmonary, type 5

CSF3R	Neutropenia, severe congenital, type 7, autosomal recessive
CSTA	Peeling skin syndrome, type 4
CSTB	Epilepsy, progressive myoclonic type 1A (Unverricht and Lundborg)
CTC1	Cerebroretinal microangiopathy with calcifications and cysts
CTH	Cystathioninuria
CTNS	Nephropathic cystinosis
CTSA	Galactosialidosis
CTSC	Haim-Munk syndrome; Papillon-Lefevre syndrome
CTSD	Ceroid lipofuscinosis, neuronal, type 10
CTSK	Pycnodysostosis
CUBN	Megaloblastic anemia 1 (Imerslund-Grasbeck syndrome)
CUL4B	Mental retardation, X-linked, syndromic, type 15 (Cabezas type)
CUL7	3-M syndrome 1
CYB5A	46,XY disorder of sex development due to isolated 17,20-lyase deficiency
CYB5R3	Methemoglobinemia, type 1; Methemoglobinemia, type 2
CYBA	Chronic granulomatous disease, autosomal recessive, due to deficiency of CYBA
CYBB	Chronic granulomatous disease, X-linked
CYP11A1	46,XY disorder of sex development-adrenal insufficiency due to CYP11A1 deficiency
CYP11B1	Adrenal hyperplasia, congenital, due to 11-beta-hydroxylase deficiency
CYP11B2	Hypoaldosteronism, congenital, due to CMO I deficiency
CYP17A1	17 alpha(?)-hydroxylase/17,20-lyase deficiency
CYP19A1	Aromatase deficiency
CYP1B1	Glaucoma, primary congenital, type 3A
CYP21A2	Congenital adrenal hyperplasia due to 21-hydroxylase deficiency
CYP24A1	Hypercalcemia, infantile, type 1
CYP27A1	Cerebrotendinous xanthomatosis
CYP27B1	Vitamin D-dependent rickets, type 1
CYP4F22	Ichthyosis, congenital, autosomal recessive, type 5
CYP4V2	Bietti crystalline corneoretinal dystrophy
CYP7B1	Spastic paraplegia type 5A, autosomal recessive
D2HGDH	D-2-hydroxyglutaric aciduria
DAG1	Muscular dystrophy-dystroglycanopathy type A9; Muscular dystrophy-dystroglycanopathy type C9
DARS2	Leukoencephalopathy with brain stem and spinal cord involvement and lactate elevation
DBH	Dopamine beta-hydroxylase deficiency
DBT	Maple syrup urine disease, type 2

DCAF17	Woodhouse-Sakati syndrome
DCC	Gaze palsy, familial horizontal, with progressive scoliosis, type 2
DCDC2	Sclerosing cholangitis, neonatal; Nephronophthisis 19
DCLRE1C	Omenn syndrome; Severe combined immunodeficiency, Athabascan type
DCX	Lissencephaly, X-linked
DDB2	Xeroderma pigmentosum, complementation group E
DDC	Aromatic L-amino acid decarboxylase deficiency
DDR2	Spondylometaepiphyseal dysplasia, short limb-hand type
DDX11	Warsaw breakage syndrome
DDX3X	Mental retardation, X-linked, type 102
DES	Myopathy, myofibrillar, type 1
DGUOK	DGUOK-related mitochondrial DNA depletion syndrome
DHCR24	Desmosterolosis
DHCR7	Smith-Lemli-Opitz syndrome
DHDDS	Retinitis pigmentosa, type 59
DHFR	Megaloblastic anemia due to dihydrofolate reductase deficiency
DHH	46,XY complete gonadal dysgenesis
DHODH	Miller syndrome
DIAPH1	Seizures, cortical blindness, microcephaly syndrome
DIS3L2	Perlman syndrome
DKC1	Dyskeratosis congenita, X-linked
DLAT	Pyruvate dehydrogenase E2 deficiency
DLD	Dihydrolipoamide dehydrogenase deficiency
DLG3	Mental retardation, X-linked, type 90
DLL3	Spondylocostal dysostosis type 1
DMD	Duchenne/Becker muscular dystrophy
DMP1	Hypophosphatemic rickets, autosomal recessive
DNAAF1	Ciliary dyskinesia, primary, type 13
DNAAF2	Ciliary dyskinesia, primary, type 10
DNAAF3	Ciliary dyskinesia, primary, type 2
DNAAF4	Ciliary dyskinesia, primary, type 25
DNAAF5	Ciliary dyskinesia, primary, type 18
DNAH11	Ciliary dyskinesia, primary, type 7, with or without situs inversus
DNAH5	Ciliary dyskinesia, primary, type 3, with or without situs inversus
DNAI1	Ciliary dyskinesia, primary, type 1, with or without situs inversus

DNAI2	Ciliary dyskinesia, primary, type 9, with or without situs inversus
DNAJB2	Spinal muscular atrophy, distal, autosomal recessive, type 5
DNAJC19	3-methylglutaconic aciduria, type 5
DNAJC6	Parkinson disease, type 19A, juvenile-onset; Parkinson disease, type 19B, early-onset
DNAL1	Ciliary dyskinesia, primary, type 16
DNM1L	Encephalopathy due to defective mitochondrial and peroxisomal fission, type 1
DNM2	Lethal congenital contracture syndrome, type 5
DNMT3B	Immunodeficiency-centromeric instability-facial anomalies syndrome, type 1
DOCK6	Adams-Oliver syndrome 2
DOCK8	Hyper-IgE recurrent infection syndrome, autosomal recessive
DOK7	Fetal akinesia deformation sequence, type 3; Myasthenic syndrome, congenital, type 10
DOLK	Congenital disorder of glycosylation, type 1M
DPAGT1	Congenital disorder of glycosylation, type 1J; Myasthenic syndrome, congenital, type 13
DPM1	Congenital disorder of glycosylation, type 1E
DPY19L2	Male infertility spermatogenic failure, type 9
DPYD	Dihydropyrimidine dehydrogenase deficiency
DPYS	Dihydropyrimidinuria
DSG1	Erythroderma, congenital, with palmoplantar keratoderma, hypotrichosis, and hyper IgE
DSG4	Hypotrichosis, type 6
DSP	Cardiomyopathy, dilated, with woolly hair and keratoderma; Epidermolysis bullosa, lethal acantholytic
DST	Epidermolysis bullosa simplex, autosomal recessive, type 2
DTNBP1	Hermansky-Pudlak syndrome 7
DUOX2	Thyroid dyshormonogenesis, type 6
DUOXA2	Thyroid dyshormonogenesis, type 5
DYM	Smith-McCort dysplasia; Dyggve-Melchior-Clausen disease
DYNC2H1	Short-rib thoracic dysplasia, type 3, with or without polydactyly
DYSF	Miyoshi muscular dystrophy, type 1; Limb-girdle muscular dystrophy, type 2 (LGMD R2)
EARS2	Combined oxidative phosphorylation deficiency 12
EBP	MEND syndrome; Chondrodysplasia punctata
ECM1	Urbach-Wiethe disease
EDA	Ectodermal dysplasia, type 1, hypohidrotic, X-linked
EDAR	Ectodermal dysplasia 10B, hypohidrotic/hair/tooth type
EDARADD	Ectodermal dysplasia 11B, hypohidrotic/hair/tooth type
EDN1	Auriculocondylar syndrome, type 3
EDN3	Waardenburg syndrome, type 4B

EDNRB	ABCD syndrome
EFEMP2	Cutis laxa, autosomal recessive, type 1B
EFNB1	Craniofrontonasal dysplasia
EGFR	?Inflammatory skin and bowel disease, neonatal, 2
EGR2	Dejerine-Sottas disease
EIF2AK3	Wolcott-Rallison syndrome
EIF2B2	Leukoencephalopathy with vanishing white matter
EIF2B3	Leukoencephalopathy with vanishing white matter
EIF2B4	Leukoencephalopathy with vanishing white matter
EIF2B5	Leukoencephalopathy with vanishing white matter
ELAC2	Combined oxidative phosphorylation deficiency 17
ELOVL4	Ichthyosis, spastic quadriplegia, and mental retardation
ELP1	Dysautonomia, familial
ELP2	Mental retardation, autosomal recessive, type 58
EMD	Emery-Dreifuss muscular dystrophy, type 1, X-linked
ENAM	Amelogenesis imperfecta, type 1C
ENPP1	Arterial calcification, generalized, of infancy, type 1
ENTPD1	Spastic paraplegia, type 64, autosomal recessive
EPB41	Elliptocytosis, type 1
EPB42	Spherocytosis, type 5
EPCAM	Congenital tufting enteropathy
EPM2A	Epilepsy, progressive myoclonic, type 2A (Lafora)
ERBB3	Lethal congenital contractural syndrome, type 2
ERCC1	Cerebrooculofacioskeletal syndrome, type 4
ERCC2	Trichothiodystrophy, type 1
ERCC3	Trichothiodystrophy, type 2
ERCC5	Cerebrooculofacioskeletal syndrome, type 3
ERCC6	Cockayne syndrome, type B; Cerebrooculofacioskeletal syndrome, type 1
ERCC8	Cockayne syndrome, type A
ERLIN2	Spastic paraplegia, type 18, autosomal recessive
ESCO2	Roberts syndrome
ESPN	Deafness, autosomal recessive, type 36
ESR1	Estrogen resistance
ESRRB	Deafness, autosomal recessive, type 35
ETFA	Glutaric acidemia, type 2A

ETFB	Glutaric acidemia, type 2B
ETFDH	Glutaric acidemia, type 2C
ETHE1	Ethylmalonic encephalopathy
EVC	Ellis-van Creveld syndrome
EVC2	Ellis-van Creveld syndrome
EXOSC3	Pontocerebellar hypoplasia, type 1B
EXT1	Chondrosarcoma
EXTL3	Immunoskeletal dysplasia with neurodevelopmental abnormalities
EYS	Retinitis pigmentosa, type 25
F10	Factor X deficiency
F13A1	Factor XIIIA deficiency
F13B	Factor XIIIB deficiency
F2	Prothrombin deficiency
F7	Factor VII deficiency
F8	Hemophilia A
F9	Hemophilia B
FA2H	Spastic paraplegia, type 35, autosomal recessive
FAH	Tyrosinemia, type 1
FAM126A	Hypomyelinating leukodystrophy, type 5
FAM161A	Retinitis pigmentosa, type 28
FAM20A	Amelogenesis imperfecta, type 1G (Enamel-renal syndrome)
FAM20C	Raine syndrome
FANCA	Fanconi anemia, complementation group A
FANCB	Fanconi anemia, complementation group B
FANCC	Fanconi anemia, complementation group C
FANCD2	Fanconi anemia, complementation group D2
FANCE	Fanconi anemia, complementation group E
FANCF	Fanconi anemia, complementation group F
FANCG	Fanconi anemia, complementation group G
FANCI	Fanconi anemia, complementation group I
FANCL	Fanconi anemia, complementation group L
FARS2	Combined oxidative phosphorylation deficiency 14; Spastic paraplegia, type 77, autosomal recessive
FBLN5	Cutis laxa, autosomal recessive, type 1A
FBP1	Fructose-1,6-bisphosphatase deficiency
FBXO7	Parkinson disease, type 15, autosomal recessive

FECH	Protoporphyria, erythropoietic, autosomal recessive
FERMT1	Kindler syndrome
FERMT3	Leukocyte adhesion deficiency, type 3
FGA	Afibrinogenemia, congenital
FGB	Congenital afibrinogenemia
FGD1	Aarskog-Scott syndrome; Mental retardation, X-linked syndromic, type 16
FGD4	Charcot-Marie-Tooth disease, type 4H
FGF23	Tumoral calcinosis, hyperphosphatemic, familial, type 2
FGF3	Deafness, congenital with inner ear agenesis, microtia, and microdontia
FGG	Afibrinogenemia, congenital; Hypofibrinogenemia, congenital
FH	Fumarase deficiency
FHL1	Emery-Dreifuss muscular dystrophy, type 6, X-linked
FIG4	Charcot-Marie-Tooth disease, type 4J; Yunis-Varon syndrome
FKBP10	Bruck syndrome 1
FKRP	Muscular dystrophy-dystroglycanopathy, type 5A, 5B and 5C
FKTN	Muscular dystrophy-dystroglycanopathy, type 4A (Walker-Warburg syndrome); Type 4B; Type 4C (limb-girdle muscular dystrophy, type 13 [LGMD R13])
FLI1	Bleeding disorder, platelet-type, type 21
FLNA	FLNA-related disorders
FLNB	Spondylocarpotarsal synostosis syndrome
FLVCR1	Posterior column ataxia-retinitis pigmentosa syndrome
FLVCR2	Proliferative vasculopathy and hydranencephaly-hydrocephaly syndrome
FMN2	Mental retardation, autosomal recessive, type 47
FMO3	Trimethylaminuria
FMR1	Fragile X syndrome
FOLR1	Neurodegeneration due to cerebral folate transport deficiency
FOXE1	Bamforth-Lazarus syndrome
FOXE3	Anterior segment dysgenesis, type 2, multiple subtypes
FOXN1	T-cell immunodeficiency, congenital alopecia and nail dystrophy
FOXP3	Immunodysregulation, polyendocrinopathy, and enteropathy, X-linked
FOXRED1	Mitochondrial complex I deficiency, nuclear type 19
FRAS1	Fraser syndrome, type 1
FREM1	Manitoba oculotrichoanal syndrome
FREM2	Fraser syndrome, type 2
FRMD7	Nystagmus 1, congenital, X-linked; Nystagmus, infantile periodic alternating, X-linked
FRMPD4	Mental retardation, X-linked, type 104

FSHB	Hypogonadotropic hypogonadism, type 24, without anosmia
FSHR	Ovarian dysgenesis 1
FTCD	Glutamate formiminotransferase deficiency
FTL	L-ferritin deficiency
FTO	Growth retardation, developmental delay, facial dysmorphism
FUCA1	Fucosidosis
FUT8	Congenital disorder of glycosylation with defective fucosylation, type 1
FXN	Friedreich ataxia
FYCO1	Cataract 18, autosomal recessive
FZD6	Nail disorder, nonsyndromic congenital, type 10 (claw-shaped nails)
G6PC	Glycogen storage disease, type 1A
G6PC3	Dursun syndrome
G6PD	Hemolytic anemia, G6PD deficient (favism)
GAA	Glycogen storage disease, type 2
GALC	Krabbe disease
GALE	Galactose epimerase deficiency
GALK1	Galactokinase deficiency with cataracts
GALNS	Mucopolysaccharidosis, type 4A
GALNT3	Tumoral calcinosis, hyperphosphatemic, familial, type 1
GALT	Galactosemia
GAMT	Cerebral creatine deficiency syndrome type 2
GAN	Giant axonal neuropathy, type 1
GATA1	Anemia, X-linked, with/without neutropenia and/or platelet abnormalities; Thrombocytopenia with beta-thalassemia, X-linked; Thrombocytopenia, X-linked, with or without dyserythropoietic anemia
GATM	Cerebral creatine deficiency syndrome, type 3
GBA	Gaucher disease
GBE1	Glycogen storage disease, type 4
GCDH	Glutaricaciduria, type 1
GCH1	Hyperphenylalaninemia, BH4-deficient, type B
GCK	Permanent neonatal diabetes mellitus (PNDM)
GCNT2	Cataract 13, with adult i phenotype
GDAP1	Charcot-Marie-Tooth disease, recessive intermediate, type A
GDF1	Right atrial isomerism (Ivemark syndrome)
GDF5	Chondrodysplasia, Grebe type
GDF6	Leber congenital amaurosis 17

GDI1	Mental retardation, X-linked, type 41
GFER	Myopathy, mitochondrial progressive, with congenital cataract, hearing loss, and developmental delay
GFM1	Combined oxidative phosphorylation deficiency, type 1
GFPT1	Myasthenia, congenital, type 12, with tubular aggregates
GGCX	Vitamin K-dependent clotting factors, combined deficiency of, type 1
GH1	Growth hormone deficiency, isolated, type 1A; Kowarski syndrome
GHRHR	Growth hormone deficiency, isolated, type 1B
GHSR	Growth hormone deficiency, isolated partial
GIF	Intrinsic factor deficiency
GIPC3	Deafness, autosomal recessive, type 15
GJA1	Craniometaphyseal dysplasia, autosomal recessive
GJB1	Charcot-Marie-Tooth neuropathy, X-linked dominant, type 1
GJB2	Deafness, autosomal recessive, type 1A
GJB6	Deafness, autosomal recessive, type 1B
GJC2	Spastic paraplegia, type 44, autosomal recessive
GK	Glycerol kinase deficiency
GLA	Fabry disease
GLB1	GM1-gangliosidosis, types 1-3; Mucopolysaccharidosis, type 4B (Morquio)
GLDC	Glycine encephalopathy
GLE1	Lethal congenital contracture syndrome, type 1; Congenital arthrogryposis with anterior horn cell disease
GLIS2	Nephronophthisis, type 7
GLIS3	Diabetes mellitus, neonatal, with congenital hypothyroidism
GLRA1	Hyperekplexia, type 1
GLRB	Hyperekplexia, type 2
GLRX5	Anemia, sideroblastic, type 3, pyridoxine-refractory; Spasticity, childhood-onset, with hyperglycinemia
GLUL	Glutamine deficiency, congenital
GLYCTK	D-glyceric aciduria
GM2A	GM2-gangliosidosis, AB variant
GNAT1	Night blindness, congenital stationary, type 1G
GNAT2	Achromatopsia 4
GNB5	Intellectual developmental disorder with cardiac arrhythmia; Language delay and ADHD/cognitive impairment with or without cardiac arrhythmia
GNE	Inclusion body myopathy, type 2 (Nonaka myopathy)
GNMT	Glycine N-methyltransferase deficiency
GNPAT	Rhizomelic chondrodysplasia punctata, type 2
GNPTAB	Mucolipidosis 2 alpha/beta; Mucolipidosis 3 alpha/beta

GNPTG	Mucolipidosis III gamma
GNRHR	Hypogonadotropic hypogonadism, type 7, without anosmia
GNS	Mucopolysaccharidosis, type 3D (Sanfilippo syndrome D)
GORAB	Geroderma osteodysplasticum
GOSR2	Epilepsy, progressive myoclonic, type 6
GP1BA	Bernard-Soulier syndrome, type A1
GP1BB	Bernard-Soulier syndrome, type B
GP6	Bleeding disorder, platelet-type, type 11
GP9	Bernard-Soulier syndrome, type C
GPC3	Simpson-Golabi-Behmel syndrome, type 1
GPC6	Omodysplasia, type 1
GPD1	Hypertriglyceridemia, transient infantile
GPHN	Molybdenum cofactor deficiency C
GPI	Hemolytic anemia, nonspherocytic, due to glucose phosphate isomerase deficiency
GPIHBP1	Hyperlipoproteinemia, type 1D
GPR143	Ocular albinism, type 1 (Nettleship-Falls type)
GPR179	Night blindness, congenital stationary (complete), type 1E, autosomal recessive
GPR68	Amelogenesis imperfecta, type 2A6 (hypomaturation type)
GPSM2	Chudley-McCullough syndrome
GPX4	Spondylometaphyseal dysplasia, Sedaghatian type
GRHL2	Ectodermal dysplasia/short stature syndrome
GRHPR	Hyperoxaluria, primary, type 2
GRIA3	Mental retardation, X-linked, type 94
GRIK2	Mental retardation, autosomal recessive, type, 6
GRIN1	Neurodevelopmental disorder with or without hyperkinetic movements and seizures, autosomal recessive
GRIP1	Fraser syndrome 3
GRM1	Spinocerebellar ataxia, autosomal recessive, type 13
GRM6	Night blindness, congenital stationary (complete), type 1B, autosomal recessive
GRN	Ceroid lipofuscinosis, neuronal, 11
GRXCR1	Deafness, autosomal recessive, type 25
GSC	Short stature, auditory canal atresia, mandibular hypoplasia, skeletal abnormalities
GSS	Glutathione synthetase deficiency
GTF2H5	Trichothiodystrophy, type 3, photosensitive
GUCY2C	Meconium ileus
GUCY2D	Leber congenital amaurosis, type 1

GUSB	Mucopolysaccharidosis, type 7
GYG1	Polyglucosan body myopathy, type 2
GYS1	Glycogen storage disease, type 0, muscle
GYS2	Glycogen storage disease, type 0, liver
H6PD	Cortisone reductase deficiency 1
HACE1	Spastic paraplegia and psychomotor retardation with or without seizures
HADH	3-hydroxyacyl-CoA dehydrogenase deficiency
HADHA	LCHAD deficiency
HADHB	Trifunctional protein deficiency
HAMP	Hemochromatosis, type 2B
HARS	Usher syndrome, type 3B
HAX1	Neutropenia, severe congenital, type 3, autosomal recessive
HBA1	Thalassemia, alpha-
HBA2	Thalassemia, alpha-
HBB	HBB-related hemoglobinopathy
HCCS	Linear skin defects with multiple congenital anomalies, type 1
HDAC8	Cornelia de Lange syndrome 5
HEPACAM	Megalencephalic leukoencephalopathy with subcortical cysts 2A
HERC2	Mental retardation, autosomal recessive, type 38
HES7	Spondylocostal dysostosis, type 4, autosomal recessive
HESX1	Growth hormone deficiency with pituitary anomalies
HEXA	Tay-Sachs disease
HEXB	Sandhoff disease, infantile, juvenile, and adult forms
HGD	Alkaptonuria
HGF	Deafness, autosomal recessive, type 39
HGSNAT	Mucopolysaccharidosis type 3C (Sanfilippo syndrome C)
HIBCH	3-hydroxyisobutryl-CoA hydrolase deficiency
HK1	Charcot-Marie-Tooth disease, type 4G
HLCS	Holocarboxylase synthetase deficiency
HMGCL	HMG-CoA lyase deficiency
HMGCS2	HMG-CoA synthase-2 deficiency
HMX1	Oculoauricular syndrome
HNMT	Mental retardation, autosomal recessive, type 51
HOGA1	Hyperoxaluria, primary, type 3
HOXA1	Athabaskan brainstem dysgenesis syndrome

HPD	Tyrosinemia, type 3
HPGD	Hypertrophic osteoarthropathy, primary, type 1 (pachydermoperiostosis)
HPRT1	Lesch-Nyhan syndrome
HPS1	Hermansky-Pudlak syndrome, type 1
HPS3	Hermansky-Pudlak syndrome 3
HPS4	Hermansky-Pudlak syndrome 4
HPS5	Hermansky-Pudlak syndrome 5
HPS6	Hermansky-Pudlak syndrome 6
HPSE2	Urofacial syndrome, type 1
HR	Alopecia universalis; Atrichia with papular lesions
HSD11B2	Apparent mineralocorticoid excess
HSD17B10	HSD10 mitochondrial disease
HSD17B3	46,XY disorder of sex development due to 17-beta-hydroxysteroid dehydrogenase 3 deficiency
HSD17B4	D-bifunctional protein deficiency
HSD3B2	Adrenal hyperplasia, congenital, due to 3-beta-hydroxysteroid dehydrogenase 2 deficiency
HSD3B7	Bile acid synthesis defect, congenital, type 1
HSPA9	Even-plus syndrome
HSPD1	Leukodystrophy, hypomyelinating, type 4
HSPG2	Dyssegmental dysplasia, Silverman-Handmaker type
HTRA1	CARASIL syndrome
HTRA2	3-methylglutaconic aciduria, type 8
HUWE1	Mental retardation, X-linked syndromic, Turner type
HYLS1	Hydrolethalus syndrome
ICK	Endocrine-cerebroosteodysplasia
IDH3B	Retinitis pigmentosa, type 46
IDS	Mucopolysaccharidosis, type 2
IDUA	Mucopolysaccharidosis, type 1H; Mucopolysaccharidosis, type 1H/S; Mucopolysaccharidosis, type 1S
IER3IP1	Microcephaly, epilepsy, and diabetes syndrome
IFNGR1	Immunodeficiency, type 27A, mycobacteriosis
IFNGR2	Immunodeficiency, type 28, mycobacteriosis
IFT122	Cranioectodermal dysplasia 1
IFT140	Retinitis pigmentosa, type 80; Short-rib thoracic dysplasia 9 with or without polydactyly
IFT80	Short-rib thoracic dysplasia, type 2, with or without polydactyly
IGF1	Growth retardation with deafness and mental retardation due to IGF1 deficiency
IGF1R	Insulin-like growth factor I, resistance to

IGFALS	Acid-labile subunit deficiency
IGFBP7	Retinal arterial macroaneurysm with supravalvular pulmonic stenosis
IGHMBP2	Charcot-Marie-Tooth disease, axonal, type 2S
IGLL1	Agammaglobulinemia 2
IHH	Acrocapitofemoral dysplasia
IKBKG	Immunodeficiency, type 33
IL10RA	Inflammatory bowel disease, type 28, early onset, autosomal recessive
IL10RB	Inflammatory bowel disease, type 25, early onset, autosomal recessive
IL11RA	Craniosynostosis and dental anomalies
IL12B	Immunodeficiency, type 29, mycobacteriosis
IL12RB1	Immunodeficiency, type 30
IL17RA	Immunodeficiency, type 51
IL1RAPL1	Mental retardation, X-linked, type 21/34
IL1RN	Sterile multifocal osteomyelitis with periostitis and pustulosis
IL21R	Immunodeficiency, type 56
IL2RA	Immunodeficiency, type 41 with lymphoproliferation and autoimmunity
IL2RG	Severe combined immunodeficiency, X-linked
IL36RN	Psoriasis, type 14, pustular
IL7R	Severe combined immunodeficiency, T-cell negative, B-cell/natural killer cell-positive type
ILDR1	Deafness, autosomal recessive, type 42
IMPAD1	Chondrodysplasia with joint dislocations, GPAPP type
IMPG2	Retinitis pigmentosa, type 56
INPP5E	Joubert syndrome, type 1
INS	Permanent neonatal diabetes mellitus (PNDM)
INSR	Diabetes mellitus, insulin-resistant, with acanthosis nigricans, type A
INVS	Nephronophthisis, type 2, infantile
IQCB1	Senior-Loken syndrome, type 5
IQSEC2	Mental retardation, X-linked, type 1/78
IRAK4	IRAK4 deficiency
IRF8	Immunodeficiency, type 32B, monocyte and dendritic cell deficiency, autosomal recessive
IRX5	Hamamy syndrome
ISCU	Myopathy with lactic acidosis, hereditary
ISPD	Muscular dystrophy-dystroglycanopathy, type A7; Muscular dystrophy-dystroglycanopathy, type C7
ITCH	Autoimmune disease, multisystem, with facial dysmorphism
ITGA2B	Glanzmann thrombasthenia

ITGA3	Interstitial lung disease, nephrotic syndrome, and epidermolysis bullosa, congenital
ITGA6	Epidermolysis bullosa, junctional, with pyloric stenosis
ITGA7	Muscular dystrophy, congenital, due to ITGA7 deficiency
ITGB2	Leukocyte adhesion deficiency
ITGB3	Glanzmann thrombasthenia
ITGB4	Epidermolysis bullosa, junctional, with pyloric atresia
ITK	Lymphoproliferative syndrome 1
ITPA	Epileptic encephalopathy, early infantile, type 35
ITPR1	Gillespie syndrome
IVD	Isovaleric acidemia
IYD	Thyroid dyshormonogenesis, type 4
JAK3	Severe Combined Immunodeficiency, autosomal recessive, T-negative/B-positive type
JAM3	Hemorrhagic destruction of the brain, subependymal calcification, and cataracts
JUP	Naxos disease
KARS	Deafness, autosomal recessive, type 89
KCNE1	Jervell and Lange-Nielsen syndrome 2
KCNJ1	Bartter syndrome, type 2
KCNJ10	SESAME syndrome
KCNJ11	Hyperinsulinemic hypoglycemia, type 2 (congenital hyperinsulinism); Permanent neonatal diabetes mellitus (PNDM)
KCNJ13	Leber congenital amaurosis, type 16
KCNV2	Retinal cone dystrophy, type 3B
KCTD7	Epilepsy, progressive myoclonic, type 3, with or without intracellular inclusions
KDM5C	Mental retardation, X-linked, syndromic, Claes-Jensen type
KDM6A	Kabuki syndrome, type 2
KERA	Cornea plana 2, autosomal recessive
KHDC3L	Hydatidiform mole, recurrent, type 2
KIF1A	Neuropathy, hereditary sensory, type 2C; Spastic paraplegia, type 30, autosomal recessive
KIF1BP	Goldberg-Shprintzen megacolon syndrome
KIF7	Acrocallosal syndrome; Joubert syndrome, type 12
KISS1R	Hypogonadotropic hypogonadism, type 8, with or without anosmia
KLHL3	Pseudohypoaldosteronism, type 2D
KLHL7	Cold-induced sweating syndrome 3
KLK4	Amelogenesis imperfecta, type 2A1
KLKB1	Fletcher factor (prekallikrein) deficiency
KNL1	Microcephaly 4, primary, autosomal recessive

KRT10	Epidermolytic hyperkeratosis
KRT14	Epidermolysis bullosa simplex, autosomal recessive, type 1
KRT5	Epidermolysis bullosa simplex, autosomal recessive, type 1
KYNU	Vertebral, cardiac, renal, and limb defects syndrome, type 2
L1CAM	L1 Syndrome
L2HGDH	L-2-hydroxyglutaric aciduria
LAMA1	Poretti-Boltshauser syndrome
LAMA2	LAMA2-related muscular dystrophy
LAMA3	Junctional epidermolysis bullosa (JEB) Herlitz type; JEB non-Herlitz type
LAMB1	Lissencephaly, type 5
LAMB2	Pierson syndrome; Nephrotic syndrome, type 5, with or without ocular abnormalities
LAMB3	Junctional epidermolysis bullosa (JEB) Herlitz type; JEB non-Herlitz type
LAMC2	Junctional epidermolysis bullosa (JEB) Herlitz type; JEB non-Herlitz type
LAMC3	Cortical malformations, occipital
LAMP2	Danon disease
LARGE1	Muscular dystrophy-dystroglycanopathy, type 6A and 6B
LARS2	Perrault syndrome, type 4
LBR	Greenberg skeletal dysplasia
LCA5	Leber congenital amaurosis, type 5
LCAT	Familial LCAT deficiency; Fish-eye disease
LCT	Lactase deficiency, congenital
LDHA	Glycogen storage disease type 11
LDLRAP1	Hypercholesterolemia, familial, autosomal recessive
LEP	Obesity, morbid, due to leptin deficiency
LEPR	Obesity, morbid, due to leptin receptor deficiency
LHB	Hypogonadotropic hypogonadism, type 23, with or without anosmia
LHCGR	Leydig cell hypoplasia
LHFPL5	Deafness, autosomal recessive type 67
LHX3	Pituitary hormone deficiency, combined, type 3
LIAS	Hyperglycinemia, lactic acidosis, and seizures
LIFR	Stuve-Wiedemann syndrome / Schwartz-Jampel type 2 syndrome
LIG4	LIG4 syndrome
LINS1	Mental retardation, autosomal recessive, type 27
LIPA	Lysosomal acid lipase deficiency
LIPE	Lipodystrophy, familial partial, type 6

LIPH	Hypotrichosis, type 7 or woolly hair, autosomal recessive, type 2, with or without hypotrichosis
LMAN1	Combined deficiency of factor V and factor VIII, type 1
LMBRD1	Methylmalonic aciduria and homocystinuria, cblF type
LMF1	Lipase deficiency, combined
LMNA	LMNA-related disorders, autosomal recessive
LOXHD1	Deafness, autosomal recessive type 77
LPAR6	Hypotrichosis, type 8 or woolly hair, autosomal recessive, type 1, with or without hypotrichosis
LPIN1	Myoglobinuria, acute recurrent, autosomal recessive
LPIN2	Majeed syndrome
LPL	Lipoprotein lipase deficiency
LRAT	Leber congenital amaurosis type 14
LRMDA	Albinism, oculocutaneous, type 7
LRP2	Donnai-Barrow syndrome
LRP4	Cenani-Lenz syndactyly syndrome
LRP5	Osteoporosis-pseudoglioma syndrome
LRPAP1	Myopia, type 23, autosomal recessive
LRPPRC	Leigh syndrome, French-Canadian type
LRRC6	Ciliary dyskinesia, primary, type 19
LRSAM1	Charcot-Marie-Tooth disease, axonal, type 2P
LRTOMT	Deafness, autosomal recessive type 63
LTBP2	Microspherophakia and/or megalocornea, with ectopia lentis and with or without secondary glaucoma
LTBP3	Dental anomalies and short stature
LTBP4	Cutis laxa, autosomal recessive, type 1C
LYST	Chediak-Higashi syndrome
LZTFL1	Bardet-Biedl syndrome, type 17
MAGI2	Nephrotic syndrome, type 15
MAGT1	Immunodeficiency, X-linked, with magnesium defect, Epstein-Barr virus infection and neoplasia
MAK	Retinitis pigmentosa type 62
MAMLD1	Hypospadias 2, X-linked
MAN1B1	Mental retardation, autosomal recessive, type 15
MAN2B1	Mannosidosis, alpha-, types I and II
MANBA	Mannosidosis, beta
MAOA	Brunner syndrome
MAPT	Supranuclear palsy, progressive atypical (parkinsonism syndrome)
MARS2	Spastic ataxia, type 3, autosomal recessive

MARVELD2	Deafness, autosomal recessive type 49
MASP1	3MC syndrome 1
MAT1A	Methionine adenosyltransferase deficiency, autosomal recessive
MBTPS2	IFAP/BRESHECK syndrome; Osteogenesis imperfecta, type 19
MC2R	Glucocorticoid deficiency, due to ACTH unresponsiveness
MCCC1	3-Methylcrotonyl-CoA carboxylase type 1 deficiency
MCCC2	3-Methylcrotonyl-CoA carboxylase type 2, deficiency
MCEE	Methylmalonyl-CoA epimerase deficiency
MCFD2	Combined deficiency of factor V and factor VIII, type 2
MCM3AP	Peripheral neuropathy, autosomal recessive, with or without impaired intellectual development
MCOLN1	Mucolipidosis type 4
MCPH1	Microcephaly type 1, primary, autosomal recessive
MECP2	Encephalopathy, neonatal severe
MED12	Lujan-Fryns syndrome
MED23	Mental retardation, autosomal recessive, type 18
MED25	Basel-Vanagait-Smirin-Yosef syndrome
MEFV	Familial Mediterranean fever
MEGF10	Myopathy, areflexia, respiratory distress, and dysphagia, early-onset
MERTK	Retinitis pigmentosa type 38
MESP2	Spondylocostal dysostosis, type 2, autosomal recessive
MFF	Encephalopathy due to defective mitochondrial and peroxisomal fission, type 2
MFN2	Charcot-Marie-Tooth disease, axonal, type 2A2B
MFRP	Microphthalmia, isolated type 5
MFSD2A	Microcephaly 15, primary, autosomal recessive
MFSD8	Ceroid lipofuscinosis, neuronal, type 7
MGAT2	Congenital disorder of glycosylation, type 2a
MGP	Keutel syndrome
MID1	Opitz GBBB syndrome, type 1
MITF	COMMAD syndrome
MKKS	Bardet-Biedl syndrome type 6
MKS1	Bardet-Biedl syndrome type 13; Meckel syndrome, type 1
MLC1	Megalencephalic leukoencephalopathy with subcortical cysts
MLPH	Griscelli syndrome, type 3
MLYCD	Malonyl-CoA decarboxylase deficiency
MMAA	Methylmalonic aciduria, vitamin B12-responsive

MMAB	Methylmalonic aciduria, vitamin B12-responsive, type cblB
MMACHC	Methylmalonic aciduria and homocystinuria, cblC type
MMADHC	Homocystinuria, cblD type, variant 1
MME	Charcot-Marie-Tooth disease, axonal, type 2T
MMP13	Metaphyseal dysplasia, Spahr type
MMP2	Multicentric osteolysis, nodulosis, and arthropathy
MMP20	Amelogenesis imperfecta, type 2A2
MMUT	Methylmalonic aciduria, mut(0) type
MOCOS	Xanthinuria, type 2
MOCS1	Molybdenum cofactor deficiency A
MOCS2	Molybdenum cofactor deficiency B
MOGS	Congenital disorder of glycosylation, type 2B
MPDU1	Congenital disorder of glycosylation, type 1F
MPDZ	Hydrocephalus, congenital, type 2, with or without brain or eye anomalies
MPI	Congenital disorder of glycosylation, type 1b
MPL	Thrombocytopenia, congenital amegakaryocytic
MPLKIP	Trichothiodystrophy, type 4, nonphotosensitive
MPO	Myeloperoxidase deficiency
MPV17	Mitochondrial DNA depletion syndrome type 6 (hepatocerebral); Charcot-Marie-Tooth disease, axonal, type 2EE
MRAP	Glucocorticoid deficiency, type 2
MRE11	Ataxia-telangiectasia-like disorder 1
MRPS16	Combined oxidative phosphorylation deficiency 2
MRPS22	Combined oxidative phosphorylation deficiency type 5
MSH3	Familial adenomatous polyposis, type 4
MSMO1	Microcephaly, congenital cataract, and psoriasiform dermatitis
MSRB3	Deafness, autosomal recessive, type 74
MTFMT	Combined oxidative phosphorylation deficiency 15
MTHFD1	Combined immunodeficiency and megaloblastic anemia with or without hyperhomocysteinemia
MTM1	Myotubular myopathy, X-linked
MTMR2	Charcot-Marie-Tooth disease, type 4B1
MTO1	Combined oxidative phosphorylation deficiency 10
MTR	Homocystinuria-megaloblastic anemia, cblG complementation type
MTRR	Homocystinuria-megaloblastic anemia, cbl E type
MTTP	Abetalipoproteinemia
MUSK	Fetal akinesia deformation sequence, type 1; Myasthenic syndrome, congenital, type 9, associated with acetylcholine receptor deficiency

MVK	Mevalonic aciduria
MYBPC1	Lethal congenital contracture syndrome, type 4
MYD88	Pyogenic bacterial infections, recurrent, due to MYD88 deficiency
MYH2	Proximal myopathy and ophthalmoplegia
MYO15A	Deafness, autosomal recessive type 3
MYO18B	Klippel-Feil syndrome, type 4, autosomal recessive, with myopathy and facial dysmorphism
MYO3A	Deafness, autosomal recessive type 30
MYO5A	Griscelli syndrome, type 1
MYO5B	Microvillus inclusion disease
MYO7A	Usher syndrome, type 1B; Deafness, autosomal recessive, type 2
MYPN	Nemaline myopathy, type 11, autosomal recessive
NAA10	Ogden syndrome
NAGA	Schindler disease, type I
NAGLU	Mucopolysaccharidosis, type 3B (Sanfilippo B)
NAGS	N-acetylglutamate synthase deficiency
NARS2	Combined oxidative phosphorylation deficiency 24
NBAS	Infantile liver failure syndrome, type 2; Short stature, optic nerve atrophy, and Pelger-Huet anomaly
NBEAL2	Gray platelet syndrome
NBN	Nijmegen breakage syndrome
NCF1	Chronic granulomatous disease due to deficiency of NCF-1
NCF2	Chronic granulomatous disease due to deficiency of NCF-2
NDE1	Lissencephaly, type 4 (with microcephaly)
NDP	Norrie disease
NDRG1	Charcot-Marie-Tooth disease, type 4D
NDST1	Mental retardation, autosomal recessive, type 46
NDUFA10	Mitochondrial complex I deficiency, nuclear type 22
NDUFA11	Mitochondrial complex I deficiency, nuclear type 14
NDUFA12	?Mitochondrial complex I deficiency, nuclear type 23
NDUFAF1	Mitochondrial complex I deficiency, nuclear type 11
NDUFAF2	Mitochondrial complex I deficiency, nuclear type 10
NDUFAF3	Mitochondrial complex I deficiency, nuclear type 18
NDUFAF5	Mitochondrial complex I deficiency, nuclear type 16
NDUFAF6	Mitochondrial complex I deficiency, nuclear type 17
NDUFB3	Mitochondrial complex I deficiency, nuclear type 25
NDUFS1	Mitochondrial complex I deficiency, nuclear type 5

NDUFS2	Mitochondrial complex I deficiency, nuclear type 6
NDUFS3	Mitochondrial complex I deficiency, nuclear type 8
NDUFS4	Mitochondrial complex I deficiency, nuclear type 1
NDUFS6	Mitochondrial complex I deficiency, nuclear type 9
NDUFS7	Mitochondrial complex I deficiency, nuclear type 3
NDUFS8	Mitochondrial complex I deficiency, nuclear type 2
NDUFV1	Mitochondrial complex I deficiency, nuclear type 4
NEB	Nemaline myopathy type 2, autosomal recessive
NECTIN1	Cleft lip/palate-ectodermal dysplasia syndrome; Orofacial cleft 7
NECTIN4	Ectodermal dysplasia-syndactyly syndrome, type 1
NEFL	Charcot-Marie-Tooth disease, type 1F
NEK1	Short-rib thoracic dysplasia, type 6, with or without polydactyly
NEK8	Renal-hepatic-pancreatic dysplasia, type 2
NEU1	Sialidosis, type 1 and type 2
NEUROG3	Diarrhea type 4, malabsorptive, congenital
NFU1	Multiple mitochondrial dysfunctions syndrome 1
NGF	Neuropathy, hereditary sensory and autonomic, type 5
NHEJ1	Severe combined immunodeficiency with microcephaly, growth retardation, and sensitivity to ionizing radiation
NHLRC1	Epilepsy, progressive myoclonic, type 2B (Lafora)
NHP2	Dyskeratosis congenita, autosomal recessive type 2
NHS	Cataract 40, X-linked
NIPAL4	Ichthyosis, congenital, autosomal recessive, type 6
NKX2-6	Conotruncal heart malformations
NLGN4X	Mental retardation, X-linked
NLRP1	Autoinflammation with arthritis and dyskeratosis
NLRP7	Hydatidiform mole, recurrent, type 1
NME8	Ciliary dyskinesia, primary, type 6
NMNAT1	Leber congenital amaurosis type 9
NOP10	Dyskeratosis congenita, autosomal recessive type 1
NPC1	Niemann-Pick disease, type C1
NPC2	Niemann-pick disease, type C2
NPHP1	Joubert syndrome type 4
NPHP3	Meckel syndrome type 7
NPHP4	Nephronophthisis type 4
NPHS1	Nephrotic syndrome, type 1

NPHS2	Nephrotic syndrome, type 2
NPR2	Acromesomelic dysplasia, Maroteaux type
NR0B1	Adrenal hypoplasia, congenital
NR1H4	Cholestasis, progressive familial intrahepatic, type 5
NR2E3	Enhanced S-cone syndrome (Goldmann-Favre); Retinitis pigmentosa, type 37
NRL	Retinal degeneration, autosomal recessive, clumped pigment type
NRXN1	Pitt-Hopkins-like syndrome, type 2
NSDHL	CHILD syndrome
NSUN2	Mental retardation, autosomal recessive, type 5
NT5C3A	Anemia, hemolytic, due to UMPH1 deficiency
NT5E	Calcification of joints and arteries
NTHL1	Familial adenomatous polyposis, type 3
NTRK1	Insensitivity to pain, congenital, with anhidrosis
NUBPL	Mitochondrial complex I deficiency, nuclear type 21
NUP62	Striatonigral degeneration, infantile
NYX	Night blindness, congenital stationary (complete), type 1A, X-linked
OAT	Gyrate atrophy of choroid and retina
OBSL1	3-M syndrome 2
OCA2	Oculocutaneous albinism type 2
OCLN	Pseudo-TORCH syndrome, type 1
OCRL	Lowe Syndrome; Dent disease type 2
OFD1	Joubert syndrome type 10; Orofaciodigital syndrome type 1
OPA1	Behr syndrome
OPA3	3-methylglutaconic aciduria, type 3
OPHN1	Mental retardation, X-linked, with cerebellar hypoplasia and distinctive facial appearance
OPTN	Amyotrophic lateral sclerosis, type 12
ORC1	Meier-Gorlin syndrome, type 1
ORC4	Meier-Gorlin syndrome, type 2
ORC6	Meier-Gorlin syndrome, type 3
OSTM1	Osteopetrosis, autosomal recessive type 5
OTC	Ornithine transcarbamylase deficiency
OTOA	Deafness, autosomal recessive type 22
OTOF	Auditory neuropathy, autosomal recessive, type 1
OXCT1	Succinyl CoA:3-oxoacid CoA transferase deficiency
P2RY12	Bleeding disorder, platelet-type, type 8

P3H1	Osteogenesis imperfecta, type 8
P3H2	Myopia, high, with cataract and vitreoretinal degeneration
PAH	Phenylketonuria
PAK3	Mental retardation, X-linked, type 30
PANK2	Neurodegeneration with brain iron accumulation type 1
PAPSS2	Brachyolmia, type 4, with mild epiphyseal and metaphyseal changes
PARK7	Parkinson disease, type 7, autosomal recessive, early-onset
PC	Pyruvate carboxylase deficiency
PCBD1	Hyperphenylalaninemia, BH4-deficient, type D
PCCA	Propionic acidemia
PCCB	Propionic acidemia
PCDH15	Deafness, autosomal recessive type 23; Usher syndrome, type 1D/F digenic
PCDH19	Epileptic encephalopathy, early infantile, type 9
PCNT	Microcephalic osteodysplastic primordial dwarfism, type 2
PCSK1	Obesity with impaired prohormone processing
PDE10A	Dyskinesia, limb and orofacial, infantile-onset
PDE6A	Retinitis pigmentosa type 43
PDE6B	Retinitis pigmentosa type 40
PDE6C	Cone dystrophy type 4
PDE6G	Retinitis pigmentosa type 57
PDE6H	Retinal cone dystrophy 3
PDHA1	Pyruvate dehydrogenase E1-alpha deficiency
PDHB	Pyruvate dehydrogenase E1-beta deficiency
PDHX	Lacticacidemia due to PDX1 deficiency
PDP1	Pyruvate dehydrogenase phosphatase deficiency
PDSS1	Coenzyme Q10 deficiency, primary, type 2
PDSS2	Coenzyme Q10 deficiency, primary, type 3
PDX1	Pancreatic agenesis type 1
PDZD7	Usher syndrome, type 2C, GPR98/PDZD7 digenic
PEPD	Prolidase deficiency
PEX1	Heimler syndrome type 1
PEX10	Peroxisome biogenesis disorder, type 6A (Zellweger syndrome); Peroxisome biogenesis disorder, type 6B
PEX12	Peroxisome biogenesis disorder type 3A (Zellweger)
PEX13	Peroxisome biogenesis disorder, type 11A (Zellweger syndrome); Peroxisome biogenesis disorder, type 11B
PEX14	Peroxisome biogenesis disorder, type 13A (Zellweger syndrome)

PEX16	Peroxisome biogenesis disorder, type 8A (Zellweger syndrome); Peroxisome biogenesis disorder, type 8B
PEX19	Peroxisome biogenesis disorder, type 12A (Zellweger syndrome)
PEX2	Peroxisome biogenesis disorder type 5A (Zellweger)
PEX26	Peroxisome biogenesis disorder type 7A (Zellweger)
PEX3	Peroxisome biogenesis disorder, type 10A (Zellweger syndrome)
PEX5	Peroxisome biogenesis disorder type 2A (Zellweger)
PEX6	Peroxisome biogenesis disorder, type 4A (Zellweger syndrome); Peroxisome biogenesis disorder, type 4B; Heimler syndrome 2
PEX7	Rhizomelic chondrodysplasia punctata, type 1
PFKM	Glycogen storage disease, type 7
PGK1	Phosphoglycerate kinase 1 deficiency
PGM1	Congenital disorder of glycosylation, type 1t
PHEX	Hypophosphatemic rickets, X-linked dominant
PHF6	Borjeson-Forssman-Lehmann syndrome
PHF8	Mental retardation syndrome, X-linked, Siderius type
PHGDH	Neu-Laxova syndrome, type 1; Phosphoglycerate dehydrogenase deficiency
PHKA1	Glycogen storage disease, type 9D
PHKA2	Glycogen storage disease, type 9A1 and type 9A2
PHKB	Glycogen storage disease, type 9B
PHKG2	Glycogen storage disease type 9c
PHYH	Refsum disease
PIEZO1	Lymphedema, hereditary, type 3
PIEZO2	Arthrogryposis, distal, with impaired proprioception and touch
PIGA	Multiple congenital anomalies-hypotonia-seizures syndrome, type 2
PIGL	Zunich neuroectodermal syndrome
PIGN	Multiple congenital anomalies-hypotonia-seizures syndrome, type 1
PIGO	Hyperphosphatasia with mental retardation syndrome 2
PINK1	Parkinson disease, type 6, early onset
PIP5K1C	Lethal congenital contractural syndrome, type 3
PJVK	Deafness, autosomal recessive type 59
PKHD1	Polycystic kidney disease type 4
PKLR	Pyruvate kinase deficiency
PKP1	Ectodermal dysplasia/skin fragility syndrome
PLA2G6	Infantile neuroaxonal dystrophy type 1
PLCB1	Epileptic encephalopathy, early infantile, type 12
PLCB4	Auriculocondylar syndrome, type 2

PLCD1	Nail disorder, nonsyndromic congenital, type 3 (leukonychia)
PLCE1	Nephrotic syndrome, type 3
PLEC	Epidermolysis bullosa simplex with muscular dystrophy
PLEKHG5	Charcot-Marie-Tooth disease, recessive intermediate C
PLG	Plasminogen deficiency, type I
PLOD1	Ehlers-Danlos syndrome, kyphoscoliotic type, 1
PLOD2	Bruck syndrome 2
PLOD3	Lysyl hydroxylase 3 deficiency
PLP1	Pelizaeus-Merzbacher disease
PMM2	Congenital disorder of glycosylation, type 1A
PMP22	Dejerine-Sottas disease
PNKP	Ataxia-oculomotor apraxia, type 4; Microcephaly, seizures, and developmental delay
PNP	Immunodeficiency due to purine nucleoside phosphorylase deficiency
PNPLA1	Ichthyosis, congenital, autosomal recessive, type 10
PNPLA2	Neutral lipid storage disease with myopathy
PNPLA6	Boucher-Neuhauser syndrome; Oliver-McFarlane syndrome; Spastic paraplegia, type 39, autosomal recessive
PNPO	Pyridoxamine 5'-phosphate oxidase deficiency
POLG	POLG-related disorders
POLH	Xeroderma pigmentosum, variant type
POLR1C	Leukodystrophy, hypomyelinating, type 11; Treacher Collins syndrome 3
POLR1D	Treacher Collins syndrome, type 2
POLR3A	Leukodystrophy, hypomyelinating, type 7
POLR3B	Leukodystrophy, hypomyelinating, type 8
POMC	Obesity, adrenal insufficiency, and red hair due to POMC deficiency
POMGNT1	Muscular dystrophy-dystroglycanopathy, type 3A (Walker-Warburg syndrome); Type 3B; Type 3C (limb-girdle muscular dystrophy, type 15 [LGMDR15])
POMGNT2	Muscular dystrophy-dystroglycanopathy, type 8A (Walker-Warburg syndrome); Type 8C (limb-girdle muscular dystrophy, type 24 [LGMD R24])
POMP	Keratosis linearis with ichthyosis congenita and sclerosing keratoderma
POMT1	Muscular dystrophy-dystroglycanopathy, type 1A (Walker-Warburg syndrome); Type 1B; Type 1C (limb-girdle muscular dystrophy, type 11 [LGMD R11])
POMT2	Muscular dystrophy-dystroglycanopathy, type 2A (Walker-Warburg syndrome); Type 2B; Type 2C (limb-girdle muscular dystrophy, type 14 [LGMD R14])
POP1	Anauxetic dysplasia, type 2
POR	Antley-Bixler syndrome with genital anomalies and disordered steroidogenesis
PORCN	Focal dermal hypoplasia
POU1F1	Pituitary hormone deficiency, combined, type 1
POU3F4	Deafness, X-linked type 2 (DFNX2)
PPIB	Osteogenesis imperfecta, type 9

PPT1	Ceroid lipofuscinosis, neuronal, type 1
PQBP1	Renpenning syndrome
PRCD	Retinitis pigmentosa, type 36
PRDM5	Brittle cornea syndrome, type 2
PREPL	Myasthenic syndrome, congenital, type 22
PRF1	Hemophagocytic lymphohistiocytosis, familial, type 2
PRG4	Camptodactyly-arthropathy-coxa vara-pericarditis syndrome
PRICKLE1	Epilepsy, progressive myoclonic, type 1B
PRKN	Parkinson disease, type 2, juvenile
PRKRA	Dystonia, type 16
PRMT7	Short stature, brachydactyly, intellectual developmental disability, and seizures
PROC	Thrombophilia due to protein C deficiency, autosomal recessive
PRODH	Hyperprolinemia, type 1
PROM1	Retinitis pigmentosa, type 41
PROP1	Pituitary hormone deficiency, combined, type 2
PROS1	Thrombophilia due to protein S deficiency, autosomal recessive
PRPH2	Leber congenital amaurosis 18; Retinitis punctata albescens
PRPS1	Deafness, X-linked 1 (DFNX1); Arts syndrome; PRPS1-related disorders
PRRX1	Agnathia-otocephaly complex
PRSS1	Trypsinogen deficiency
PRSS12	Mental retardation, autosomal recessive, type 1
PRSS56	Microphthalmia, isolated, type 6
PRX	Charcot-Marie-Tooth disease, type 4F
PSAP	Combined SAP deficiency
PSAT1	Neu-Laxova syndrome, type 2
PSMB8	Autoinflammation, lipodystrophy, and dermatosis syndrome
PSPH	Phosphoserine phosphatase deficiency
PTH	Hypoparathyroidism, autosomal recessive
PTH1R	Chondrodysplasia, Blomstrand type; Eiken syndrome
PTPRC	Severe combined immunodeficiency, T cell-negative, B-cell/natural killer-cell positive
PTPRQ	Deafness, autosomal recessive, type 84A
PTS	Hyperphenylalaninemia, BH4-deficient, type A
PUS1	Myopathy, lactic acidosis, and sideroblastic anemia, type 1
PXDN	Anterior segment dysgenesis, type 7, with sclerocornea
PYCR1	Cutis laxa, autosomal recessive, type 2B

PYGL	Glycogen storage disease, type 6
PYGM	McArdle disease
QDPR	Hyperphenylalaninemia, BH4-deficient, type C
RAB18	Warburg micro syndrome, type 3
RAB23	Carpenter syndrome
RAB27A	Griscelli syndrome, type 2
RAB28	Cone-rod dystrophy 18
RAB39B	Mental retardation, X-linked, type 72; Waisman syndrome
RAB3GAP1	Warburg micro syndrome, type 1
RAB3GAP2	Martsolf syndrome
RAG1	Omenn syndrome; Severe combined immunodeficiency, B cell-negative
RAG2	Omenn syndrome; Severe combined immunodeficiency, B cell-negative
RAPSN	Fetal akinesia deformation sequence; Myasthenic syndrome, congenital, 11, associated with AChR deficiency
RARS2	Pontocerebellar hypoplasia, type 6
RAX	Isolated microphthalmia, type 3
RBBP8	Jawad syndrome; Seckel syndrome, type 2
RBM10	TARP syndrome
RBM8A	Thrombocytopenia-absent radius syndrome
RBP4	Retinal dystrophy, iris coloboma, and comedogenic acne syndrome
RD3	Leber congenital amaurosis 12
RDH12	Leber congenital amaurosis, type 13
RDH5	Fundus albipunctatus
RDX	Deafness, autosomal recessive, type 24
RECQL4	Baller-Gerold syndrome; RAPADILINO syndrome; Rothmund-Thomson syndrome
RELN	Lissencephaly 2 (Norman-Roberts type)
REN	Renal tubular dysgenesis
RETREG1	Neuropathy, hereditary sensory and autonomic, type 2B
RFX6	Mitchell-Riley syndrome
RFXANK	Bare lymphocyte syndrome, type 2, complementation group B
RGR	Retinitis pigmentosa, type 44
RHO	Retinitis pigmentosa, type 4, autosomal recessive; Fundus albipunctatus
RIN2	Macs syndrome
RIPK4	Popliteal pterygium syndrome, Bartsocas-Papas type
RLBP1	Bothnia retinal dystrophy; Fundus albipunctatus
RMRP	Anauxetic dysplasia, type 1

RNASEH2A	Aicardi-Goutieres syndrome 4
RNASEH2B	Aicardi-Goutieres syndrome 2
RNASEH2C	Aicardi-Goutieres syndrome 3
RNF168	RIDDLE syndrome
ROBO3	Gaze palsy, familial horizontal, with progressive scoliosis, type 1
ROGDI	Kohlschutter-Tonz syndrome
ROM1	Retinitis pigmentosa, type 7, digenic
ROR2	Robinow syndrome, autosomal recessive
RP1	Retinitis pigmentosa, type 1
RP2	Retinitis pigmentosa, type 2, X-linked
RPE65	Leber congenital amaurosis, type 2
RPGR	Retinitis pigmentosa, type 3, X-linked
RPGRIP1	Leber congenital amaurosis 6
RPGRIP1L	Joubert syndrome, type 7; Meckel syndrome, type 5; COACH syndrome
RPL10	Mental retardation, X-linked, syndromic, type 35
RPS6KA3	Mental retardation, X-linked, type 19
RRM2B	Mitochondrial DNA depletion syndrome, type 8A (encephalomyopathic type with renal tubulopathy) and type 8B (MNGIE type)
RS1	Retinoschisis
RSPH4A	Ciliary dyskinesia, primary, type 11
RSPH9	Ciliary dyskinesia, primary, type 12
RSPO4	Anonychia congenita
RTEL1	Dyskeratosis congenita, autosomal recessive 5
RYR1	Minicore myopathy with external ophthalmoplegia
SACS	Spastic ataxia, Charlevoix-Saguenay, type
SAG	Oguchi disease, type 1
SAMHD1	Aicardi-Goutieres syndrome 5
SAR1B	Chylomicron retention disease
SARS2	Hyperuricemia, pulmonary hypertension, renal failure, and alkalosis
SBDS	Shwachman-Diamond syndrome
SBF2	Charcot-Marie-Tooth disease, type 4B2
SC5D	Lathosterolosis
SCARB2	Epilepsy, progressive myoclonic, type 4, with or without renal failure
SCARF2	Van den Ende-Gupta syndrome
SCN1B	Epileptic encephalopathy, early infantile, type 52
SCN4A	Myasthenic syndrome, congenital, type 16

SCN9A	Indifference to pain and autosomal recessive hereditary sensory neuropathy type 2D
SCNN1A	Pseudohypoaldosteronism, type 1
SCNN1B	Pseudohypoaldosteronism, type 1
SCNN1G	Pseudohypoaldosteronism, type 1
SCO1	Mitochondrial complex IV deficiency
SCO2	Cardioencephalomyopathy, fatal infantile, due to cytochrome c oxidase deficiency, type 1
SDCCAG8	Bardet-Biedl syndrome, type 16
SDHA	Mitochondrial respiratory chain complex II deficiency; Leigh syndrome
SDHAF1	Mitochondrial complex II deficiency
SEC23A	Craniolenticulosutural dysplasia
SEC23B	Dyserythropoietic anemia, congenital, type 2
SECISBP2	Thyroid hormone metabolism, abnormal
SELENON	Muscular dystrophy, rigid spine, type 1
SEMA4A	Cone-rod dystrophy, type 10; Retinitis pigmentosa, type 35
SEPSECS	Pontocerebellar hypoplasia, type 2D
SERPINC1	Thrombophilia due to antithrombin III deficiency
SERPINE1	Plasminogen activator inhibitor-1 deficiency
SERPINF1	Osteogenesis imperfecta, type 6
SERPING1	Angioedema, hereditary, types 1 and 2
SERPINH1	Osteogenesis imperfecta, type 10
SETX	Spinocerebellar ataxia, autosomal recessive, type 1
SFTPB	Surfactant metabolism dysfunction, pulmonary, type 1
SGCA	Limb-girdle muscular dystrophy, type 3 (LGMD R3)
SGCB	Limb-girdle muscular dystrophy, type 4 (LGMD R4)
SGCD	Limb-girdle muscular dystrophy, type 6 (LGMD R6)
SGCG	Limb-girdle muscular dystrophy, type 5 (LGMD R5)
SGSH	Mucopolysaccharidosis, type 3A (Sanfilippo A)
SH2D1A	Lymphoproliferative syndrome, X-linked, type 1
SH3PXD2B	Frank-ter Haar syndrome
SH3TC2	Charcot-Marie-Tooth disease, type 4C
SHOX	Langer mesomelic dysplasia
SHROOM4	Stocco dos Santos X-linked mental retardation syndrome
SI	Sucrase-isomaltase deficiency, congenital
SIL1	Marinesco-Sjogren syndrome
SIX6	Optic disc anomalies with retinal and/or macular dystrophy

SKIV2L	Trichohepatoenteric syndrome, type 2 (diarrhea, syndromic)
SLC10A2	Bile acid malabsorption, primary
SLC12A1	Bartter syndrome, type 1
SLC12A3	Gitelman syndrome
SLC12A6	Agenesis of the corpus callosum with peripheral neuropathy
SLC16A1	Monocarboxylate transporter 1 deficiency
SLC16A2	Allan-Herndon-Dudley syndrome
SLC17A5	Salla disease
SLC19A2	Thiamine-responsive megaloblastic anemia syndrome
SLC19A3	Thiamine metabolism dysfunction syndrome, type 2 (biotin- or thiamine-responsive encephalopathy type)
SLC22A12	Hypouricemia, renal
SLC22A5	Carnitine deficiency, systemic primary
SLC24A1	Night blindness, congenital stationary (complete), type 1D, autosomal recessive
SLC24A5	Albinism, oculocutaneous, type 6
SLC25A12	Epileptic encephalopathy, early infantile, type 39
SLC25A13	Citrullinemia, adult-onset, type 2
SLC25A15	Hyperornithinemia-hyperammonemia-homocitrullinemia syndrome
SLC25A19	Microcephaly, Amish type; Thiamine metabolism dysfunction syndrome 4 (progressive polyneuropathy type)
SLC25A20	Carnitine-acylcarnitine translocase deficiency
SLC25A22	Epileptic encephalopathy, early infantile, type 3
SLC25A3	Mitochondrial phosphate carrier deficiency
SLC25A38	Anemia, sideroblastic, type 2, pyridoxine-refractory
SLC25A4	Mitochondrial DNA depletion syndrome, type 12B (cardiomyopathic type) AR
SLC26A2	Achondrogenesis, type 1B (diastrophic dysplasia)
SLC26A3	Diarrhea 1, secretory chloride, congenital
SLC26A4	Deafness, autosomal recessive, type 4; Pendred syndrome
SLC27A4	Ichthyosis prematurity syndrome
SLC29A3	Histiocytosis-lymphadenopathy plus syndrome
SLC2A1	GLUT1 deficiency syndrome 1, infantile onset, severe
SLC2A10	Arterial tortuosity syndrome
SLC2A2	Fanconi-Bickel syndrome
SLC2A9	Hypouricemia, renal, type 2
SLC30A10	Hypermanganesemia with dystonia, type 1
SLC33A1	Congenital cataracts, hearing loss, and neurodegeneration
SLC34A1	Hypercalcemia, infantile, type 2

SLC34A2	Pulmonary alveolar microlithiasis
SLC34A3	Hypophosphatemic rickets with hypercalciuria
SLC35A1	Congenital disorder of glycosylation, type 2F
SLC35A3	?Arthrogryposis, mental retardation, and seizures
SLC35C1	Congenital disorder of glycosylation, type 2C
SLC35D1	Schneckenbecken dysplasia
SLC37A4	Glycogen storage disease, type 1b
SLC39A4	Acrodermatitis enteropathica
SLC3A1	Cystinuria
SLC45A2	Albinism, oculocutaneous, type 4
SLC46A1	Folate malabsorption, hereditary
SLC4A1	Distal renal tubular acidosis (dRTA) with hemolytic anemia
SLC4A11	Corneal endothelial dystrophy, autosomal recessive
SLC4A4	Renal tubular acidosis, proximal, with ocular abnormalities
SLC52A2	Brown-Vialetto-Van Laere syndrome, type 2
SLC52A3	Brown-Vialetto-Van Laere syndrome, type 1
SLC5A2	Renal glucosuria
SLC5A5	Thyroid dyshormonogenesis, type 1
SLC5A7	Myasthenic syndrome, congenital, type 20, presynaptic
SLC6A19	Hartnup disorder
SLC6A3	Parkinsonism-dystonia, infantile
SLC6A5	Hyperekplexia, type 3
SLC6A8	Cerebral creatine deficiency syndrome, type 1
SLC7A7	Lysinuric protein intolerance
SLC7A9	Cystinuria
SLC9A6	Mental retardation, X-linked syndromic, Christianson type
SLCO2A1	Hypertrophic osteoarthropathy, primary, autosomal recessive, type 2
SLURP1	Meleda disease
SMARCAL1	Schimke immunoosseous dysplasia
SMC1A	Cornelia de Lange syndrome 2
SMN1	Spinal muscular atrophy
SMOC1	Microphthalmia. with limb anomalies
SMOC2	Dentin dysplasia, type 1, with microdontia and misshapen teeth
SMPD1	Niemann-Pick disease, type A; Niemann-Pick disease, type B
SMPX	Deafness, X-linked, type 4

SMS	Mental retardation, X-linked, Snyder-Robinson type
SNAP29	Cerebral dysgenesis, neuropathy, ichthyosis, and palmoplantar keratoderma syndrome
SNX10	Osteopetrosis, autosomal recessive, type 8
SOD1	Spastic tetraplegia and axial hypotonia, progressive; Amyotrophic lateral sclerosis, type 1
SOHLH1	Ovarian dysgenesis 5
SOST	Sclerosteosis, type 1; Van Buchem disease
SOX18	Hypotrichosis-lymphedema-telangiectasia syndrome
SP110	Hepatic venoocclusive disease with immunodeficiency
SPART	Spactic paraplegia, type 20, autosomal recessive
SPATA7	Leber congenital amaurosis 3
SPG11	Amyotrophic lateral sclerosis, type 5, juvenile
SPG21	Mast syndrome
SPG7	Spastic paraplegia, type 7, autosomal recessive
SPINK1	Tropical calcific pancreatitis
SPINK5	Netherton syndrome
SPINT2	Diarrhea, type 3, secretory sodium, congenital, syndromic
SPR	Dystonia, dopa-responsive, due to sepiapterin reductase deficiency
SPTA1	Pyropoikilocytosis; Apherocytosis, type 3
SPTBN2	Spinocerebellar ataxia, autosomal recessive, type 14
SQSTM1	Neurodegeneration with ataxia, dystonia, and gaze palsy, childhood-onset
SRD5A2	46,XY disorder of sex development due to 5-alpha-reductase 2 deficiency (pseudovaginal perineoscrotal hypospadias)
SRD5A3	Congenital disorder of glycosylation, type 1Q; Kahrizi syndrome
ST14	Ichthyosis, congenital, autosomal recessive, type 11
ST3GAL5	Salt and pepper developmental regression syndrome
STAR	Lipoid adrenal hyperplasia
STAT1	Immunodeficiency, type 31B, mycobacterial and viral infections, autosomal recessive
STAT5B	Laron syndrome with immunodeficiency
STIL	Microcephaly, type 7, primary, autosomal recessive
STIM1	Immunodeficiency, type 10
STK4	T-cell immunodeficiency, recurrent infections, autoimmunity, and cardiac malformations
STRA6	Microphthalmia, isolated, with coloboma, type 8
STRADA	Polyhydramnios, megalencephaly, and symptomatic epilepsy
STRC	Deafness, autosomal recessive, type 16
STS	Ichthyosis, X-linked
STX11	Hemophagocytic lymphohistiocytosis, familial, type 4

STXBP2	Hemophagocytic lymphohistiocytosis, familial, type 5
SUCLA2	Mitochondrial DNA depletion syndrome, type 5 (encephalomyopathic with or without methylmalonic aciduria)
SUCLG1	Mitochondrial DNA depletion syndrome, type 9 (encephalomyopathic, type with methylmalonic aciduria)
SUFU	Joubert syndrome, type 32
SUGCT	Glutaric aciduria, type 3
SULT2B1	Ichthyosis, congenital, autosomal recessive, type 14
SUMF1	Multiple sulfatase deficiency
SUOX	Sulfite oxidase deficiency
SURF1	Charcot-Marie-Tooth disease, type 4K; Leigh syndrome, due to COX IV deficiency
SYN1	Epilepsy, X-linked, with variable learning disabilities and behavior disorders
SYNE1	Spinocerebellar ataxia, autosomal recessive, type 8
SYT14	Spinocerebellar ataxia, autosomal recessive, type 11
TAC3	Hypogonadotropic hypogonadism, type 10, with or without anosmia
TACO1	Mitochondrial complex IV deficiency
TACR3	Hypogonadotropic hypogonadism, type 11, with or without anosmia
TACSTD2	Corneal dystrophy, gelatinous drop-like
TALDO1	Transaldolase deficiency
TAT	Tyrosinemia, type 2
TAZ	3-methylglutaconic aciduria, type 2 (Barth syndrome)
TBC1D23	Pontocerebellar hypoplasia, type 11
TBC1D24	DOORS (deafness, onychodystrophy, osteodystrophy, mental retardation, and seizures) syndrome; Epileptic encephalopathy, early infantile, type 16; Deafness, autosomal recessive, type 86
TBCE	Encephalopathy, progressive, with amyotrophy and optic atrophy; Hypoparathyroidism-retardation-dysmorphism syndrome; Kenny-Caffey syndrome, type 1
TBX19	Congenital isolated adrenocorticotropic hormone deficiency
TBX22	Cleft palate with ankyloglossia
TBXAS1	Ghosal syndrome
TCAP	Limb-girdle muscular dystrophy, type 7 (LGMD R7)
TCIRG1	Osteopetrosis, autosomal recessive, type 1
TCN2	Transcobalamin II deficiency
TCTN1	Joubert syndrome, type 13
TCTN2	Joubert syndrome, type 24
TDP1	Spinocerebellar ataxia, autosomal recessive with axonal neuropathy
TDRD7	Cataract 36
TECR	Mental retardation, autosomal recessive, type 14
TECTA	Deafness, autosomal recessive, type 21

TERT	Dyskeratosis congenita, autosomal recessive, type 4
TF	Atransferrinemia
TFR2	Hemochromatosis, type 3
TG	Thyroid dyshormonogenesis, type 3
TGM1	Ichthyosis, congenital, autosomal recessive, type 1
TGM5	Peeling skin syndrome, type 2
TH	Segawa syndrome, recessive
THRB	Thyroid hormone resistance, autosomal recessive
TIMM8A	Mohr-Tranebjaerg syndrome
TJP2	Cholestasis, progressive familial intrahepatic, type 4
TK2	Mitochondrial DNA depletion syndrome , type 2 (myopathic type)
TMC1	Deafness, autosomal recessive, type 7
TMC6	Epidermodysplasia verruciformis
TMC8	Epidermodysplasia verruciformis
TMCO1	Craniofacial dysmorphism, skeletal anomalies, and mental retardation syndrome
TMEM126A	Optic atrophy 7
TMEM216	Joubert syndrome, type 2; Meckel syndrome, type 2
TMEM237	Joubert syndrome, type 14
TMEM67	Joubert syndrome, type 6; Meckel syndrome, type 3; COACH syndrome
TMEM70	Mitochondrial complex V (ATP synthase) deficiency, nuclear type 2
TMIE	Deafness, autosomal recessive, type 6
TMPRSS15	Enterokinase deficiency
TMPRSS3	Deafness, autosomal recessive, type 8/10
TMPRSS6	Iron-refractory iron deficiency anemia
TNFRSF11A	Osteopetrosis, autosomal recessive, type 7
TNFRSF11B	Paget disease of bone, type 5, juvenile-onset
TNFRSF13B	Immunodeficiency, common variable, type 2
TNFSF11	Osteopetrosis, autosomal recessive, type 2
TNNT1	Nemaline myopathy , type 5, Amish type
TNXB	Ehlers-Danlos syndrome, classic-like
TPI1	Hemolytic anemia due to triosephosphate isomerase deficiency
TPK1	Episodic encephalopathy due to thiamine pyrophosphokinase deficiency
TPM3	Nemaline myopathy, type 1; Congenital fiber-type disproportion myopathy
TPO	Thyroid dyshormonogenesis, type 2A
TPP1	Ceroid lipofuscinosis, neuronal, type 2

TPRN	Deafness, autosomal recessive, type 79
TRAF3IP1	Senior-Loken syndrome, type 9
TRAPPC2	Spondyloepiphyseal dysplasia tarda
TRAPPC9	Mental retardation, autosomal recessive, type 13
TRDN	Ventricular tachycardia, catecholaminergic polymorphic, type 5, with or without muscle weakness
TREM2	Nasu-Hakola disease
TREX1	Aicardi-Goutieres syndrome, type 1
TRIM32	Limb-girdle muscular dystrophy, type 8 (LGMD R8)
TRIM37	Mulibrey nanism
TRIOBP	Deafness, autosomal recessive, type 28
TRIP11	Achondrogenesis, type 1A
TRMU	Liver failure, transient infantile
TRPM1	Night blindness, congenital stationary (complete), type 1C, autosomal recessive
TRPM6	Familial hypomagnesemia with secondary hypocalcemia
TSEN2	Pontocerebellar hypoplasia, type 2B
TSEN54	Pontocerebellar hypoplasia, type 2A; Pontocerebellar hypoplasia, type 4
TSFM	Combined oxidative phosphorylation deficiency, type 3
TSHB	Hypothyroidism, congenital, nongoitrous, type 4
TSHR	Hypothyroidism, congenital, nongoitrous, type 1
TSPAN7	Mental retardation, X-linked, type 58
TTC19	Mitochondrial complex III deficiency, nuclear type 2
TTC21B	Short-rib thoracic dysplasia, type 4, with or without polydactyly
TTC37	Trichohepatoenteric syndrome, type 1 (diarrhea, syndromic)
TTC8	Bardet-Biedl syndrome, type 8
TTI2	Mental retardation, autosomal recessive, type 39
TTN	Limb-girdle muscular dystrophy type 10 (LGMDR10); Early-onset myopathy with fatal cardiomyopathy
TTPA	Ataxia with isolated vitamin E deficiency
TUBA8	Cortical dysplasia, complex, with other brain malformations, type 8
TUBGCP4	Microcephaly and chorioretinopathy, autosomal recessive, type 3
TUBGCP6	Microcephaly and chorioretinopathy, autosomal recessive, type 1
TUFM	Combined oxidative phosphorylation deficiency 4
TULP1	Leber congenital amaurosis, type 15
TUSC3	Mental retardation, autosomal recessive, type 7
TWIST2	Focal facial dermal dysplasia, type 3 (Setleis type)
TWNK	Mitochondrial DNA depletion syndrome, type 7 (hepatocerebral type); Perrault syndrome type 5

TYK2	Immunodeficiency, type 35
TYMP	Mitochondrial DNA depletion syndrome, type 1 (MNGIE type)
TYR	Oculocutaneous albinism (OCA) type 1A; OCA type 1B
TYROBP	Polycystic lipomembranous osteodysplasia with sclerosing leukoencephalopathy, type 1 (Nasu-Hakola disease)
TYRP1	Albinism, oculocutaneous, type 3
UBE2A	Mental retardation, X-linked syndromic, Nascimento-type
UBQLN2	Amyotrophic lateral sclerosis, type 15, with or without frontotemporal dementia
UBR1	Johanson-Blizzard syndrome
UCHL1	Spastic paraplegia, type 79, autosomal recessive
UGT1A1	Crigler-Najjar syndrome, type 1; Crigler-Najjar syndrome, type 2
UMPS	Orotic aciduria
UNC13D	Hemophagocytic lymphohistiocytosis, familial, type 3
UNC80	Hypotonia, infantile, with psychomotor retardation and characteristic facies, type 2
UNG	Immunodeficiency with hyper IgM, type 5
UPB1	Beta-ureidopropionase deficiency
UPF3B	Mental retardation, X-linked, syndromic, type 14
UQCRB	Mitochondrial complex III deficiency, nuclear, type 3
UQCRQ	Mitochondrial complex III deficiency, nuclear, type 4
UROD	Porphyria cutanea tarda
UROS	Porphyria, congenital erythropoietic
USB1	Poikiloderma with neutropenia
USH1C	Usher syndrome, type 1C; Deafness, autosomal recessive, type 18A
USH1G	Usher syndrome, type 1G
USH2A	Usher syndrome, type 2A
USP9X	Mental retardation, X-linked, type 99; Mental retardation, X-linked, type 99, syndromic, female-restricted
UVSSA	UV-sensitive syndrome, type 3
VDR	Rickets, vitamin D-resistant, type 2A
VIPAS39	Arthrogryposis, renal dysfunction, and cholestasis 2
VKORC1	Vitamin K-dependent clotting factors, combined deficiency of, type 2
VLDLR	Cerebellar hypoplasia and mental retardation with or without quadrupedal locomotion, type 1
VMA21	Myopathy, X-linked, with excessive autophagy
VPS13A	Choreoacanthocytosis
VPS13B	Cohen syndrome
VPS33B	Arthrogryposis, renal dysfunction and cholestasis, type 1
VRK1	Pontocerebellar hypoplasia, type 1A

VSX2	Microphthalmia, isolated, with coloboma, type 3
VWF	von Willibrand disease, type 3
WAS	Wiskott-Aldrich syndrome; Thrombocytopenia, X-linked
WASHC4	?Mental retardation, autosomal recessive, type 43
WASHC5	Ritscher-Schinzel syndrome, type 1
WDR19	Nephronophthisis, type 13; Senior-Loken syndrome, type 8
WDR35	Cranioectodermal dysplasia 2
WDR45	Neurodegeneration with brain iron accumulation, type 5
WDR45B	Neurodevelopmental disorder with spastic quadriplegia and brain abnormalities with or without seizures
WDR62	Microcephaly, type 2, primary, autosomal recessive, with or without cortical malformations
WDR72	Amelogenesis imperfecta, type 2A3
WDR81	Cerebellar ataxia, mental retardation, and dysequilibrium syndrome, type 2
WFS1	Wolfram syndrome, type 1
WHRN	Usher syndrome, type 2D; Deafness, autosomal recessive, type 31
WISP3	Arthropathy, progressive pseudorheumatoid, of childhood
WNK1	Neuropathy, hereditary sensory and autonomic, type 2
WNT10A	Odontoonychodermal dysplasia
WNT10B	Split-hand/foot malformation, type 6
WNT7A	Fuhrmann syndrome
WRAP53	Dyskeratosis congenita, autosomal recessive, type 3
WRN	Werner syndrome
WWOX	Epileptic encephalopathy, early infantile, type 28; Spinocerebellar ataxia, autosomal recessive, type 12
XDH	Xanthinuria, type 1
XIAP	Lymphoproliferative syndrome, X-linked, 2
XK	McLeod syndrome with or without chronic granulomatous disease
XPA	Xeroderma pigmentosum, group A
XPC	Xeroderma pigmentosum, group C
XPNPEP3	Nephronophthisis-like nephropathy, type 1
XRCC4	Short stature, microcephaly, and endocrine dysfunction
XYLT1	Desbuquois dysplasia, type 2
XYLT2	Spondyloocular syndrome
YARS2	Myopathy, lactic acidosis, and sideroblastic anemia, type 2
YY1AP1	Grange syndrome
ZAP70	Autoimmune disease, multisystem, infantile-onset, type 2; Immunodeficiency, type 48
ZBTB16	Skeletal defects, genital hypoplasia, and mental retardation

ZBTB24	Immunodeficiency-centromeric instability-facial anomalies syndrome, type 2
ZC3H14	Mental retardation, autosomal recessive, type 56
ZDHHC9	Mental retardation, X-linked syndromic, Raymond type
ZFYVE26	Spastic paraplegia, type 15, autosomal recessive
ZIC3	Heterotaxy, visceral, 1, X-linked; X-linked VACTERL syndrome with or without hydrocephalus
ZMPSTE24	Mandibuloacral dysplasia with, type B lipodystrophy
ZNF469	Brittle cornea syndrome, type 1
ZNF711	Mental retardation, X-linked, type 97