CGT Plus version 3.0.1

Gene	Disease
ABCA12	Ichthyosis, congenital, autosomal recessive, type 4A; ICAR, type 4B (harlequin)
ABCB11	Cholestasis, benign recurrent intrahepatic, type 2; Cholestasis, progressive familial intrahepatic, type 2
ABCC8	Hyperinsulinemic hypoglycemia, type 1 (congenital hyperinsulinism); Permanent neonatal diabetes mellitus (PNDM)
ABCD1	Adrenoleukodystrophy
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
ACADVL	Very long-chain acyl-CoA dehydrogenase deficiency
ACAT1	Alpha-methylacetoacetic aciduria (3-ketothiolase deficiency)
ACOX1	Peroxisomal acyl-CoA oxidase deficiency
ADA	Severe combined immunodeficiency due to adenosine deaminase deficiency (ADA)
ADAMTS2	Ehlers-Danlos syndrome, dermatosparaxis type
AGA	Aspartylglucosaminuria (glycosylasparaginase deficiency)
AGL	Glycogen storage disease, type 3
AGPS	Rhizomelic chondrodysplasia punctata, type 3
AGXT	Hyperoxaluria, primary, type 1
AHI1	Joubert syndrome, type 3
AIRE	Autoimmune polyendocrinopathy syndrome type 1
ALDH3A2	Sjogren-Larsson syndrome
ALDOB	Fructose intolerance, hereditary
ALG1	Congenital disorder of glycosylation, type 1K
ALG6	Congenital disorder of glycosylation, type 1C
ALMS1	Alstr?m syndrome
ALPL	Hypophosphatasia, childhood/infantile
AMT	Glycine encephalopathy
AR	Androgen insensitivity syndrome, complete
ARG1	Argininemia (arginase deficiency)
ARSA	Metachromatic leukodystrophy
ARSB	Mucopolysaccharidosis, type 6 (Maroteaux-Lamy syndrome)
ARSE	Chondrodysplasia punctata, brachytelephalangic
ASL	Argininosuccinic aciduria

ASPA	Canavan disease
ASS1	Citrullinemia, type 1
ATM	Ataxia-telangiectasia
ATP7A	Menkes disease; Occipital horn syndrome
ATP7B	Wilson disease
ATP8B1	Cholestasis, progressive familial intrahepatic, type 1; Cholestasis, benign recurrent intrahepatic, type 1
B4GALT1	Congenital disorder of glycosylation, type 2D
BBS1	Bardet-Biedl syndrome, type 1
BBS10	Bardet-Biedl syndrome, type 10
BBS2	Bardet-Biedl syndrome, type 2
BCKDHA	Maple syrup urine disease, type 1A
BCKDHB	Maple syrup urine disease, type 1B
BCS1L	BCS1L-related disorders, including Leigh syndrome
BLM	Bloom syndrome
BSND	Bartter syndrome, type 4A
BTD	Biotinidase deficiency
BTK	Agammaglobulinemia X-linked, type 1
CA2	Osteopetrosis with renal tubular acidosis (osteopetrosis, autosomal recessive, type 3)
CAPN3	Limb-girdle muscular dystrophy, type 1 (LGMD R1)
CBS	Homocystinuria due to cystathionine beta-synthase
CD40LG	Hyper-IgM syndrome, type 1 (immunodeficiency, X-linked, with hyper-IgM, type 1)
CDH23	Deafness, autosomal recessive, type 12; Usher syndrome, type 1D
CEP290	Meckel syndrome, type 4; Joubert syndrome, type 5; Leber congenital amaurosis, type 10
CERKL	Retinitis pigmentosa, type 26
CFTR	Cystic fibrosis
CHAT	Myasthenic syndrome, congenital, type 6, presynaptic
CHM	Choroideremia
CHRNE	Myasthenic syndrome, congenital, type 4B, fast-channel; Myasthenic syndrome, congenital, type 4C, associated with acetylcholine receptor deficiency
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
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
COLQ	Myasthenic syndrome, congenital, type 5
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
CRB1	Retinitis pigmentosa, type 12; Leber congenital amaurosis, type 8
CRTAP	Osteogenesis imperfecta, type 7
CTNS	Nephropathic cystinosis
CTSD	Ceroid lipofuscinosis, neuronal, type 10
CTSK	Pycnodysostosis
CYBA	Chronic granulomatous disease, autosomal recessive, due to deficiency of CYBA
CYBB	Chronic granulomatous disease, X-linked
CYP1B1	Glaucoma, primary congenital, type 3A
CYP27A1	Cerebrotendinous xanthomatosis
DBT	Maple syrup urine disease, type 2
DCLRE1C	Omenn syndrome; Severe combined immunodeficiency, Athabascan type
DDB2	Xeroderma pigmentosum, complementation group E
DHCR7	Smith-Lemli-Opitz syndrome
DHDDS	Retinitis pigmentosa, type 59
DLD	Dihydrolipoamide dehydrogenase deficiency
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
DPYD	Dihydropyrimidine dehydrogenase deficiency
DYSF	Miyoshi muscular dystrophy, type 1; Limb-girdle muscular dystrophy, type 2 (LGMD R2)
EDA	Ectodermal dysplasia, type 1, hypohidrotic, X-linked
EIF2B5	Leukoencephalopathy with vanishing white matter
EMD	Emery-Dreifuss muscular dystrophy, type 1, X-linked
ERCC2	Trichothiodystrophy, type 1
ERCC3	Trichothiodystrophy, type 2
ERCC5	Cerebrooculofacioskeletal syndrome, type 3
ESCO2	Roberts syndrome

ETFA	Glutaric acidemia, type 2A
ETFB	Glutaric acidemia, type 2B
ETFDH	Glutaric acidemia, type 2C
ETHE1	Ethylmalonic encephalopathy
EYS	Retinitis pigmentosa, type 25
F8	Hemophilia A
F9	Hemophilia B
FAH	Tyrosinemia, type 1
FAM20C	Raine syndrome
FANCA	Fanconi anemia, complementation group A
FANCC	Fanconi anemia, complementation group C
FANCG	Fanconi anemia, complementation group G
FH	Fumarase deficiency
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])
FMR1	Fragile X syndrome
FRAS1	Fraser syndrome, type 1
G6PC	Glycogen storage disease, type 1A
G6PC3	Dursun syndrome
G6PD	Hemolytic anemia, G6PD deficient (favism)
GAA	Glycogen storage disease, type 2
GALC	Krabbe disease
GALK1	Galactokinase deficiency with cataracts
GALNS	Mucopolysaccharidosis, type 4A
GALT	Galactosemia
GAMT	Cerebral creatine deficiency syndrome type 2
GBA	Gaucher disease
GBE1	Glycogen storage disease, type 4
GCDH	Glutaricaciduria, type 1
GCH1	Hyperphenylalaninemia, BH4-deficient, type B
GFM1	Combined oxidative phosphorylation deficiency, type 1
GJB2	Deafness, autosomal recessive, type 1A
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
GNE	Inclusion body myopathy, type 2 (Nonaka myopathy)
GNPTAB	Mucolipidosis 2 alpha/beta; Mucolipidosis 3 alpha/beta
GNS	Mucopolysaccharidosis, type 3D (Sanfilippo syndrome D)
GPR143	Ocular albinism, type 1 (Nettleship-Falls type)
GRHPR	Hyperoxaluria, primary, type 2
GUSB	Mucopolysaccharidosis, type 7
HADHA	LCHAD deficiency
HADHB	Trifunctional protein deficiency
HAX1	Neutropenia, severe congenital, type 3, autosomal recessive
HBA1	Thalassemia, alpha-
HBA2	Thalassemia, alpha-
HBB	HBB-related hemoglobinopathy
HEXA	Tay-Sachs disease
HEXB	Sandhoff disease, infantile, juvenile, and adult forms
HGD	Alkaptonuria
HGSNAT	Mucopolysaccharidosis type 3C (Sanfilippo syndrome C)
HLCS	Holocarboxylase synthetase deficiency
HMGCL	HMG-CoA lyase deficiency
HPS1	Hermansky-Pudlak syndrome, type 1
HPS3	Hermansky-Pudlak syndrome 3
HSD17B4	D-bifunctional protein deficiency
HSPG2	Dyssegmental dysplasia, Silverman-Handmaker type
HYLS1	Hydrolethalus syndrome
IDS	Mucopolysaccharidosis, type 2
IDUA	Mucopolysaccharidosis, type 1H; Mucopolysaccharidosis, type 1H/S; Mucopolysaccharidosis, type 1S
IGHMBP2	Charcot-Marie-Tooth disease, axonal, type 2S
IL2RG	Severe combined immunodeficiency, X-linked
IVD	Isovaleric acidemia
KCNJ11	Hyperinsulinemic hypoglycemia, type 2 (congenital hyperinsulinism); Permanent neonatal diabetes mellitus (PNDM)
L1CAM	L1 Syndrome
LAMA2	LAMA2-related muscular dystrophy
LAMA3	Junctional epidermolysis bullosa (JEB) Herlitz type; JEB non-Herlitz type
LAMB3	Junctional epidermolysis bullosa (JEB) Herlitz type; JEB non-Herlitz type

LAMC2	Junctional epidermolysis bullosa (JEB) Herlitz type; JEB non-Herlitz type
LARGE1	Muscular dystrophy-dystroglycanopathy, type 6A and 6B
LHX3	Pituitary hormone deficiency, combined, type 3
LIFR	Stuve-Wiedemann syndrome / Schwartz-Jampel type 2 syndrome
LIPA	Lysosomal acid lipase deficiency
LOXHD1	Deafness, autosomal recessive type 77
LRPPRC	Leigh syndrome, French-Canadian type
LYST	Chediak-Higashi syndrome
MAN2B1	Mannosidosis, alpha-, types I and II
MCCC2	3-Methylcrotonyl-CoA carboxylase type 2, deficiency
MCOLN1	Mucolipidosis type 4
MECP2	Encephalopathy, neonatal severe
MEFV	Familial Mediterranean fever
MFSD8	Ceroid lipofuscinosis, neuronal, type 7
MKS1	Bardet-Biedl syndrome type 13; Meckel syndrome, type 1
MLC1	Megalencephalic leukoencephalopathy with subcortical cysts
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
MMUT	Methylmalonic aciduria, mut(0) type
MOGS	Congenital disorder of glycosylation, type 2B
MPI	Congenital disorder of glycosylation, type 1b
MPL	Thrombocytopenia, congenital amegakaryocytic
MPV17	Mitochondrial DNA depletion syndrome type 6 (hepatocerebral); Charcot-Marie-Tooth disease, axonal, type 2EE
MTM1	Myotubular myopathy, X-linked
MTR	Homocystinuria-megaloblastic anemia, cblG complementation type
MTRR	Homocystinuria-megaloblastic anemia, cbl E type
MTTP	Abetalipoproteinemia
MYO7A	Usher syndrome, type 1B; Deafness, autosomal recessive, type 2
NAGLU	Mucopolysaccharidosis, type 3B (Sanfilippo B)
NAGS	N-acetylglutamate synthase deficiency
NBN	Nijmegen breakage syndrome
NCF1	Chronic granulomatous disease due to deficiency of NCF-1
NCF2	Chronic granulomatous disease due to deficiency of NCF-2

NDRG1	Charcot-Marie-Tooth disease, type 4D
NEB	Nemaline myopathy type 2, autosomal recessive
NHP2	Dyskeratosis congenita, autosomal recessive type 2
NOP10	Dyskeratosis congenita, autosomal recessive type 1
NPC1	Niemann-Pick disease, type C1
NPC2	Niemann-pick disease, type C2
NPHS1	Nephrotic syndrome, type 1
NPHS2	Nephrotic syndrome, type 2
NR0B1	Adrenal hypoplasia, congenital
NR2E3	Enhanced S-cone syndrome (Goldmann-Favre); Retinitis pigmentosa, type 37
NTRK1	Insensitivity to pain, congenital, with anhidrosis
OAT	Gyrate atrophy of choroid and retina
OCA2	Oculocutaneous albinism type 2
OCRL	Lowe Syndrome; Dent disease type 2
OPA3	3-methylglutaconic aciduria, type 3
OSTM1	Osteopetrosis, autosomal recessive type 5
OTC	Ornithine transcarbamylase deficiency
P3H1	Osteogenesis imperfecta, type 8
PAH	Phenylketonuria
PC	Pyruvate carboxylase deficiency
PCCA	Propionic acidemia
PCCB	Propionic acidemia
PCDH15	Deafness, autosomal recessive type 23; Usher syndrome, type 1D/F digenic
PDHA1	Pyruvate dehydrogenase E1-alpha deficiency
PEX1	Heimler syndrome type 1
PEX2	Peroxisome biogenesis disorder type 5A (Zellweger)
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
PHGDH	Neu-Laxova syndrome, type 1; Phosphoglycerate dehydrogenase deficiency
PKHD1	Polycystic kidney disease type 4
PLA2G6	Infantile neuroaxonal dystrophy type 1
PLOD1	Ehlers-Danlos syndrome, kyphoscoliotic type, 1
PMM2	Congenital disorder of glycosylation, type 1A
PNPO	Pyridoxamine 5'-phosphate oxidase deficiency

POLG	POLG-related disorders
POMGNT1	Muscular dystrophy-dystroglycanopathy, type 3A (Walker-Warburg syndrome); Type 3B; Type 3C (limb-girdle muscular dystrophy, type 15 [LGMDR15])
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])
POU1F1	Pituitary hormone deficiency, combined, type 1
PPT1	Ceroid lipofuscinosis, neuronal, type 1
PRF1	Hemophagocytic lymphohistiocytosis, familial, type 2
PROP1	Pituitary hormone deficiency, combined, type 2
PSAP	Combined SAP deficiency
PTS	Hyperphenylalaninemia, BH4-deficient, type A
PYGM	McArdle disease
QDPR	Hyperphenylalaninemia, BH4-deficient, type C
RAB23	Carpenter 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
RDH12	Leber congenital amaurosis, type 13
RPE65	Leber congenital amaurosis, type 2
RPGRIP1L	Joubert syndrome, type 7; Meckel syndrome, type 5; COACH syndrome
RTEL1	Dyskeratosis congenita, autosomal recessive 5
SACS	Spastic ataxia, Charlevoix-Saguenay, type
SBDS	Shwachman-Diamond syndrome
SGCA	Limb-girdle muscular dystrophy, type 3 (LGMD R3)
SGCB	Limb-girdle muscular dystrophy, type 4 (LGMD R4)
SGCG	Limb-girdle muscular dystrophy, type 5 (LGMD R5)
SGSH	Mucopolysaccharidosis, type 3A (Sanfilippo A)
SH2D1A	Lymphoproliferative syndrome, X-linked, type 1
SLC12A3	Gitelman syndrome
SLC12A6	Agenesis of the corpus callosum with peripheral neuropathy
SLC17A5	Salla disease
SLC22A5	Carnitine deficiency, systemic primary
SLC25A13	Citrullinemia, adult-onset, type 2

SLC25A15	Hyperornithinemia-hyperammonemia-homocitrullinemia syndrome
SLC26A2	Achondrogenesis, type 1B (diastrophic dysplasia)
SLC26A4	Deafness, autosomal recessive, type 4; Pendred syndrome
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
SLC45A2	Albinism, oculocutaneous, type 4
SLC4A11	Corneal endothelial dystrophy, autosomal recessive
SLC6A8	Cerebral creatine deficiency syndrome, type 1
SLC7A7	Lysinuric protein intolerance
SMN1	Spinal muscular atrophy
SMPD1	Niemann-Pick disease, type A; Niemann-Pick disease, type B
STAR	Lipoid adrenal hyperplasia
SUMF1	Multiple sulfatase deficiency
TCIRG1	Osteopetrosis, autosomal recessive, type 1
TFR2	Hemochromatosis, type 3
TGM1	Ichthyosis, congenital, autosomal recessive, type 1
TH	Segawa syndrome, recessive
TMEM216	Joubert syndrome, type 2; Meckel syndrome, type 2
TPP1	Ceroid lipofuscinosis, neuronal, type 2
TSEN54	Pontocerebellar hypoplasia, type 2A; Pontocerebellar hypoplasia, type 4
TSFM	Combined oxidative phosphorylation deficiency, type 3
TTPA	Ataxia with isolated vitamin E deficiency
TYR	Oculocutaneous albinism (OCA) type 1A; OCA type 1B
UGT1A1	Crigler-Najjar syndrome, type 1; Crigler-Najjar syndrome, type 2
UNC13D	Hemophagocytic lymphohistiocytosis, familial, type 3
USH1C	Usher syndrome, type 1C; Deafness, autosomal recessive, type 18A
USH1G	Usher syndrome, type 1G
USH2A	Usher syndrome, type 2A
VPS13A	Choreoacanthocytosis
WAS	Wiskott-Aldrich syndrome; Thrombocytopenia, X-linked
WNT10A	Odontoonychodermal dysplasia
XPA	Xeroderma pigmentosum, group A