Complete list of condition and genes associated

Condition	Gene
Achromatopsia	CNGB3
Alpha-1 Antitrypsin Deficiency	SERPINA1
Alpha-Mannosidosis	MAN2B1
Alpha-thalassemia	HBA1
Alpha-thalassemia	HBA2
Andermann Syndrome	SLC12A6
Argininosuccinate Lyase Deficiency	ASL
Aspartylglycosaminuria	AGA
Ataxia With Vitamin E Deficiency	TTPA
Ataxia-Telangiectasia	ATM
Autosomal Recessive Spastic Ataxia of Charlevoix-Saguenay (ARSACS)	SACS
Bardet-Biedl Syndrome, Type 1	BBS1
Bardet-Biedl Syndrome, Type 10	BBS10
Bernard-Soulier Syndrome, Type B	GP1BB
Bernard-Soulier Syndrome,Type C	GP9
Beta-Hemoglobinopathies (including Sickle Cell and Beta-Thalassemia)	HBB
Beta-Ketothiolase Deficiency	ACAT1
Biotinidase Deficiency	BTD
Bloom Syndrome	BLM
Canavan Disease	ASPA
Carnitine Deficiency, Primary	SLC22A5
Carnitine Palmitoyltransferase IA Deficiency	CPT1A
Carnitine Palmitoyltransferase II Deficiency	CPT2
Cartilage-Hair Hypoplasia	RMRP
Choroideremia	CHM
Citrullinemia Type 1	ASS1

Cohen Syndrome	VPS13B
Combined Pituitary Hormone Deficiency	PROP1
Congenital Adrenal Hyperplasia (CAH)	CYP21A2
Congenital Disorder of Glycosylation Type Ia	PMM2
Congenital Disorder of Glycosylation Type Ib	MPI
Costeff Optic Atrophy Syndrome	OPA3
Cystic Fibrosis	CFTR
Cystinosis	CTNS
D-Bifunctional Protein Deficiency	HSD17B4
Dihydrolipoamide Dehydrogenase Deficiency (a.k.a Maple Syrup Urine Disease Type 3)	DLD
Dihydropyrimidine Dehydrogenase Deficiency	DPYD
Duchenne/Becker Muscular Dystrophy	DMD
Fabry Disease	GLA
Factor XI Deficiency	F11
Familial Dysautonomia	IKBKAP
Familial Mediterranean Fever	MEFV
Fanconi Anemia Type C	FANCC
Finnish Nephrosis (a.k.a Nephrotic Syndrome Type 1)	NPHS1
Fumarase Deficiency	FH
Galactosemia	GALT
Gaucher Disease	GBA
Glaucoma, Primary Congenital	CYP1B1
Glucose-6-Phosphate Dehydrogenase Deficiency	G6PD
Glutaric Acidemia Type 1	GCDH
Glycogen Storage Disease Type Ia (von Gierke)	G6PC
Glycogen Storage Disease Type Ib (von Gierke)	SLC37A4
Glycogen Storage Disease Type III (Cori/Forbes)	AGL
Glycogen Storage Disease Type V (McArdle)	PYGM
GRACILE Syndrome	BCS1L
Growth Hormone Deficiency, Isolated	GHRHR

Hearing Loss, Non-syndromic (a.k.a Connexin 26)	GJB2
Hearing Loss, Non-syndromic (a.k.a Connexin 30)	GJB6
Heme Oxygenase 1 Deficiency	HMOX1
Hemochromatosis	HFE
Hemophilia B	F9
Hereditary Fructose Intolerance	ALDOB
Herlitz Junctional Epidermolysis Bullosa, LAMA3-Related	LAMA3
Herlitz Junctional Epidermolysis Bullosa, LAMB3-Related	LAMB3
Herlitz Junctional Epidermolysis Bullosa, LAMC2-Related	LAMC2
Homocystinuria, CBS-deficient	CBS
Hydatidiform Mole, Recurrent	NLRP7
Hyperinsulinism	ABCC8
Hyperoxaluria, Primary Type 1	AGXT
Hyperoxaluria, Primary Type 2	GRHPR
Hypohidrotic Ectodermal Dysplasia	EDAR
Hypophosphatasia	ALPL
Inclusion Body Myopathy 2	GNE
Isovaleric Acidemia	IVD
Joubert Syndrome 2	TMEM216
Krabbe Disease	GALC
Limb-Girdle Muscular Dystrophy Type 2A	CAPN3
Limb-Girdle Muscular Dystrophy Type 2C	SGCG
Limb-Girdle Muscular Dystrophy Type 2D	SGCA
Limb-Girdle Muscular Dystrophy Type 2E	SGCB
Long Chain 3-Hydroxyacyl-CoA Dehydrogenase Deficiency	HADHA
Maple Syrup Urine Disease Type 1A	BCKDHA
Maple Syrup Urine Disease Type 1B	BCKDHB
Maple Syrup Urine Disease Type 2	DBT
Medium Chain Acyl-CoA Dehydrogenase Deficiency	ACADM
Megalencephalic Leukoencephalopathy with Subcortical Cysts	MLC1

Megaloblastic Anemia Syndrome	SLC19A2
Metachromatic Leukodystrophy	ARSA
Methlymalonic Acidemia, cblA Type	MMAA
Methlymalonic Acidemia, cblB-Type	MMAB
Methlymalonic Acidemia, mut-Type	MUT
Methylmalonic Acidemia and Homocystinuria, cblC Type	MMACHC
Mucolipidosis Type II/IIIA	GNPTAB
Mucolipidosis Type IV	MCOLN1
Mucopolysaccharidosis Type I (Hurler)	IDUA
Mucopolysaccharidosis Type II (Hunter)	IDS
Mucopolysaccharidosis Type IIIA (Sanfilippo A)	SGSH
Mucopolysaccharidosis Type IIIB (Sanfilippo B)	NAGLU
Mucopolysaccharidosis Type IVA (Morquio A)	GALNS
Mucopolysaccharidosis Type IVB (Morquio B)	GLB1
Mucopolysaccharidosis Type VI (Maroteaux-Lamy)	ARSB
Mucopolysaccharidosis Type VII (Sly)	GUSB
Muscle-Eye-Brain Disease	POMGNT1
Nemaline Myopathy	NEB
Nephrotic Syndrome Type 2	NPHS2
Neuronal Ceroid Lipofuscinosis Type 1	PPT1
Neuronal Ceroid Lipofuscinosis Type 2	TPP1
Neuronal Ceroid Lipofuscinosis Type 3	CLN3
Neuronal Ceroid Lipofuscinosis Type 5	CLN5
Neuronal Ceroid Lipofuscinosis Type 8 (a.k.a Northern Epilepsy)	CLN8
Niemann-Pick Disease Type A & B (a.k.a. Acid Sphingomyelinase Deficiency)	SMPD1
Niemann-Pick Disease Type C1	NPC1
Niemann-Pick Disease Type C2	NPC2
Nijmegen Breakage Syndrome	NBN
Oculocutaneous Albinism Type 1	TYR

Ornithine Transcarbamlyase Deficiency	OTC
Pantothenate Kinase-associated Neurodegeneration	PANK2
Papillon-Lefevre Syndrome (also Haim-Munk Syndrome)	CTSC
Pendred Syndrome	SLC26A4
Phenylalanine Hydroxylase Deficiency (PKU)	PAH
Polycystic Kidney Disease, Autosomal Recessive	PKHD1
Polyglandular Autoimmune Syndrome Type 1	AIRE
Pompe Disease (a.k.a Glycogen Storage Disease Type 2 or Acid Maltase Deficiency)	GAA
Progressive Pseudorheumatoid Dysplasia	WISP3
Pycnodysostosis	CTSK
Retinoschisis, Juvenile	RS1
Rett Syndrome	MECP2
Rhizomelic Chondrodysplasia Punctata Type 1	PEX7
Salla Disease (a.k.a Sialic Acid Storage Disease)	SLC17A5
Sandhoff Disease	HEXB
Segawa Syndrome	TH
Short Chain Acyl-CoA Dehydrogenase Deficiency	ACADS
Sjogren-Larsson Syndrome	ALDH3A2
Skeletal Dysplasias, SLC26A2-related	SLC26A2
Smith-Lemli-Opitz Syndrome	DHCR7
Tay Sachs Disease (a.k.a. Hexosaminidase A Deficiency)	HEXA
Tricho-Hepato-Enteric Syndrome	TTC37
Tyrosinemia Type I	FAH
Usher Syndrome Type 1F	PCDH15
Usher Syndrome Type 3	CLRN1
Very Long Chain Acyl-CoA Dehydrogenase Deficiency	ACADVL
Walker- Warburg Syndrome, Type 4	FKTN
Werner Syndrome	WRN
Wilson Disease	ATP7B

Woolly Hair/Hypotrichosis Syndrome	LIPH
Zellweger Spectrum Disorder Type 1 (a.k.a Infantile Refsum Disease)	PEX1