CONGENITAL DISORDERS OF GLYCOSYLATION GENE TABLE

Note: This panel can be ordered from the Mitochondrial/Metabolic test requisition form.

Gene Name	OMIM #	Associated Disease(s)/Phenotypes	Inheritance Pattern
ALG1	605907	Congenital disorder of glycosylation, type Ik; ALG1-CDG (CDG-Ik)	Autosomal Recessive
ALG11	613666	Congenital disorder of glycosylation, type Ip; ALG11-CDG (CDG-Ip)	Autosomal Recessive
ALG12	607144	Congenital disorder of glycosylation, type Ig; ALG12-CDG (CDG-Ig)	Autosomal Recessive
ALG13	300776	Congenital disorder of glycosylation, type Is; ALG13-CDG	X-Linked
ALG14	612866	Congenital myasthenic syndrome 15	Autosomal Recessive
ALG2	607905	Congenital disorder of glycosylation, type Ii; ALG2-CDG (CDG-Ii)	Autosomal Recessive
ALG3	608750	Congenital disorder of glycosylation, type Id; ALG3-CDG (CDG-Id)	Autosomal Recessive
ALG6	604566	Congenital disorder of glycosylation, type Ic; ALG6-CDG (CDG-Ic)	Autosomal Recessive
ALG8	608103	Congenital disorder of glycosylation, type Ih; ALG8-CDG (CDG-Ih)	Autosomal Recessive
ALG9	606941	Congenital disorder of glycosylation, type Il; ALG9-CDG (CDG-IL)	Autosomal Recessive
ATP6AP1	300197	Immunodeficiency 47	X-Linked
ATP6V0A2	611716	Autosomal recessive cutis laxa type IIA; Wrinkly skin syndrome	Autosomal Recessive
B3GALNT2	610194	Muscular dystrophy-dystroglycanopathy (congenital with brain and eye anomalies, type A, 11	Autosomal Recessive
B3GALT6	615291	Ehlers-Danlos syndrome, progeroid type, 2; Spondyloepimetaphyseal dysplasia with joint laxity, type 1, with or without fractures	Autosomal Recessive
B3GALTL	610308	Peters-plus syndrome	Autosomal Recessive
B3GAT3	606374	Multiple joint dislocations, short stature, craniofacial dysmorphism, with or without congenital heart defects	Autosomal Recessive
B4GALNT1	601873	Autosomal recessive spastic paraplegia 26	Autosomal Recessive
B4GALT1	137060	Congenital disorder of glycosylation, type IId; B4GALT1-CDG (CDG-IId)	Autosomal Recessive
B4GALT7	604327	Ehlers-Danlos syndrome with short stature and limb anomalies	Autosomal Recessive
B4GAT1	605517	Muscular dystrophy-dystroglycanopathy (congenital with brain and eye anomalies), type A, 13	Autosomal Recessive
CCDC115	613734	Congenital disorder of glycosylation, type IIo; CCDC115-CDG (CDG-IIo)	Autosomal Recessive
CHST14	608429	Ehlers-Danlos syndrome, musculocontractural type 1	Autosomal Recessive
CHST3	603799	Spondyloepiphyseal dysplasia with congenital joint dislocations	Autosomal Recessive
CHST6	605294	Macular corneal dystrophy	Autosomal Recessive
CHSY1	608183	Temtamy preaxial brachydactyly syndrome	Autosomal Recessive
COG1	606973	Congenital disorder of glycosylation, type IIg; COG1-CDG (CDG-IIg)	Autosomal Recessive
COG2	606974	Congenital disorder of glycosylation, type IIq; COG2-CDG (CDG-IIq)	Autosomal Recessive
COG4	606976	Congenital disorder of glycosylation, type IIj; COG4-CDG (CDG-IIj)	Autosomal Recessive
COG5	606821	Congenital disorder of glycosylation, type IIi; COG5-CDG (CDG-IIi)	Autosomal Recessive
COG6	606977	Congenital disorder of glycosylation, type III; COG6-CDG (CDG-IIL)	Autosomal Recessive
COG7	606978	Congenital disorder of glycosylation, type IIe; COG7-CDG (CDG-IIe)	Autosomal Recessive
COG8	606979	Congenital disorder of glycosylation, type IIh; COG8-CDG (CDG-IIh)	Autosomal Recessive
DDOST	602202	Congenital disorder of glycosylation, type Ir; DDOST-CDG (CDG-Ir)	Autosomal Recessive
DHDDS	608172	Retinitis pigmentosa 59; DHDDS-CDG	Autosomal Recessive
DOLK	610746	Congenital disorder of glycosylation, type Im; DOLK-CDG (CDG-Im)	Autosomal Recessive

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DPAGT1	191350	Congenital disorder of glycosylation, type Ij; DPAGT1-CDG (CDG-Ij)	Autosomal Recessive
DPM1	603503	Congenital disorder of glycosylation, type Ie; DPM1-CDG (CDG-Ie)	Autosomal Recessive
DPM2	603564	Congenital disorder of glycosylation, type Iu; DPM2-CDG	Autosomal Recessive
DPM3	605951	Congenital disorder of glycosylation, type Io; DPM3-CDG (CDG-Io)	Autosomal Recessive
DSE	605942	Ehlers-Danlos syndrome, musculocontractural type 2	Autosomal Recessive
EOGT	614789	Adams-Oliver syndrome 4	Autosomal Recessive
EXT1	608177	Multiple exostoses type 1	Autosomal Dominant
EXT2	608210	Multiple exostoses type 2	Autosomal Dominant
FKRP	606596	Muscular dystrophy-dystroglycanopathy	Autosomal Recessive
FKTN	607440	Muscular dystrophy-dystroglycanopathy	Autosomal Recessive
FUT8	602589	Polyhydramnios, intrauterine growth retardation, shortened limbs, microcephaly, seizures, hyptonia, respiratory distress, feeding difficulties[1]	Autosomal Recessive
G6PC3	611045	Dursun syndrome; Autosomal recessive, severe congenital neutropenial 4	Autosomal Recessive
GALNT3	601756	Familial hyperphosphatemic tumoral calcinosis	Autosomal Recessive
GFPT1	138292	Congenital myasthenia 12	Autosomal Recessive
GMPPA	615495	Alacrima, achalasia, and mental retardation syndrome; GMPPA-CDG	Autosomal Recessive
GMPPB	615320	Muscular dystrophy-dystroglycanopathy	Autosomal Recessive
GNE	603824	Nonaka myopathy; Sialuria	Autosomal Recessive/ Autosomal Dominant
ISPD	614631	Muscular dystrophy-dystroglycanopathy	Autosomal Recessive
LARGE	114220	Autosomal recessive spastic paraplegia 76	Autosomal Recessive
LFNG	602576	Autosomal recessive spondylocostal dysostosis 3	Autosomal Recessive
MAN1B1	604346	Autosomal recessive mental retardation 15; MAN1B1-CDG	Autosomal Recessive
MGAT2	602616	Congenital disorder of glycosylation, type IIa; MGAT2-CDG (CDG-IIa)	Autosomal Recessive
MOGS	601336	Congenital disorder of glycosylation, type IIb; MOGS-CDG (CDG-IIb)	Autosomal Recessive
MPDU1	604041	Congenital disorder of glycosylation, type If; MPDU1-CDG (CDG-If)	Autosomal Recessive
MPI	154550	Congenital disorder of glycosylation, type Ib; MPI-CDG (CDG-Ib)	Autosomal Recessive
NGLY1	610661	Congenital disorder of deglycosylation	Autosomal Recessive
PAPSS2	603005	Brachyolmia 4 with mild epiphyseal and metaphyseal changes	Autosomal Recessive
PGAP1	611655	Autosomal recessive mental retardation 42	Autosomal Recessive
PGAP2	615187	Hyperphosphatasia with mental retardation syndrome 3	Autosomal Recessive
PGAP3	611801	Hyperphosphatasia with mental retardation syndrome 4	Autosomal Recessive
PGM1	171900	Congenital disorder of glycosylation, type It; PGM1-CDG (CDG-It)	Autosomal Recessive
PGM3	172100	Immunodeficiency 23; PGM3-CDG	Autosomal Recessive
PIGA	311770	Multiple congenital anomalies-hypotonia-seizures syndrome 2	X-Linked
PIGL	605947	CHIME syndrome	Autosomal Recessive
PIGM	610273	Glycosylphosphatidylinositol deficiency	Autosomal Recessive

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PIGN	606097	Multiple congenital anomalies-hypotonia-seizures syndrome 1	Autosomal Recessive
PIGO	614730	Hyperphosphatasia with mental retardation syndrome 2	Autosomal Recessive
PIGT	610272	Multiple congenital anomalies-hypotonia-seizures syndrome 3	Autosomal Recessive
PIGV	610274	Hyperphosphatasia with mental retardation syndrome 1	Autosomal Recessive
PIGW	610275	Hyperphosphatasia with mental retardation syndrome 5	Autosomal Recessive
PIGY	610662	Hyperphosphatasia with mental retardation syndrome 6	Autosomal Recessive
PMM2	601785	Congenital disorder of glycosylation, type Ia; PMM2-CDG (CDG-Ia)	Autosomal Recessive
POFUT1	607491	Dowling-Degos disease 2	Autosomal Dominant
POGLUT1	615618	Muscular dystrophy, limb-girdle, type 2Z; Dowling-Degos disease 4	Autosomal Recessive/ Autosomal Dominant
POMGNT1	606822	Muscular dystrophy-dystroglycanopathy	Autosomal Recessive
POMGNT2	614828	Muscular dystrophy-dystroglycanopathy	Autosomal Recessive
POMK	615247	Muscular dystrophy-dystroglycanopathy	Autosomal Recessive
POMT1	607423	Muscular dystrophy-dystroglycanopathy	Autosomal Recessive
POMT2	607439	Muscular dystrophy-dystroglycanopathy	Autosomal Recessive
RFT1	611908	Congenital disorder of glycosylation, type In; RFT1-CDG (CDG-In)	Autosomal Recessive
RPN2	180490	Congenital disorder of glycosylation 1	Autosomal Recessive
SEC23A	610511	Craniolenticulosutural dysplasia	Autosomal Recessive
SEC23B	610512	Cowden syndrome 7; Dyserythropoietic anemia, congenital, type II	Autosomal Dominant/ Autosomal Recessive
SLC26A2	606718	Diastrophic dysplasia	Autosomal Recessive
SLC35A1	605634	Congenital disorder of glycosylation, type IIf; SLC35A1-CDG (CDG-IIf)	Autosomal Recessive
SLC35A2	314375	Congenital disorder of glycosylation, type IIm; SLC35A2-CDG (CDG-IIm)	X-Linked
SLC35A3	605632	Arthrogryposis, mental retardation, and seizures	Autosomal Recessive
SLC35C1	605881	Congenital disorder of glycosylation, type IIc; SLC35C1-CDG (CDG-IIc)	Autosomal Recessive
SLC35D1	610804	Schneckenbecken dysplasia	Autosomal Recessive
SLC39A8	608732	Congenital disorder of glycosylation, type IIn; SLC39A8-CDG (CDG-IIn)	Autosomal Recessive
SRD5A3	611715	Congenital disorder of glycosylation, type Iq; SRD5A3-CDG (CDG-Iq)	Autosomal Recessive
SSR4	300090	Congenital disorder of glycosylation, type Iy; SSR4-CDG (CDG-Iy)	X-Linked
ST3GAL3	606494	Epileptic encephalopathy, early infantile, 15; Mental retardation, autosomal recessive 12	Autosomal Recessive
ST3GAL5	604402	Salt and pepper developmental regression syndrome	Autosomal Recessive
STT3A	601134	Congenital disorder of glycosylation, type Iw; STT3A-CDG (CDG-Iw)	Autosomal Recessive
STT3B	608605	Congenital disorder of glycosylation, type Ix; STT3B-CDG (CDG-Ix)	Autosomal Recessive
TMEM165	614726	Congenital disorder of glycosylation, type IIk; TMEM165-CDG (CDG-IIk)	Autosomal Recessive
TMEM199	616815	Congenital disorder of glycosylation, type IIp; TMEM199-CDG (CDG-IIp)	Autosomal Recessive
TMEM5	605862	Muscular dystrophy-dystroglycanopathy	Autosomal Recessive
TRAPPC11	614138	Muscular dystrophy, limb-girdle, type 2S	Autosomal Recessive

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TRIP11	604505	Achondrogenesis, type IA	Autosomal Recessive
TUSC3	601385	Mental retardation, autosomal recessive 7	Autosomal Recessive
XYLT1	608124	Desbuquois dysplasia 2	Autosomal Recessive

Reference:

1. Ng et al., (2018) Am J Hum Genet (ahead of print)

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