Gene-CDG (IEMbase hyperlink)	Name	Alternative names	Gene	Inheritance	OMIM	PMID	Comments	Diagnostic biomarkers	Cardiovascular	Dental	Dermatological	Digestive	Dysmorphic	Ear	Endocrine	Eye	Genitourinary	Hair	Hematological	Immunological	Muscular	Neurologic	Psychiatric	Renal	Respiratory	Skeletal	Short stature
1. Disorders of monosaccharide synthesis and interconversion																											
PMM2-CDG	Phosphomannomutase 2 deficiency	Jaeken syndrome	*PMM2*	AR	601785	PMID: 9140401	prim. N-Gly affected	Sialotransferrins (S)	X		X	X	X		X	X	X					X	X	X		X	
MPI-CDG	Phosphomannose isomerase deficiency	Saguenay-Lac Saint-Jean syndrome	*MPI*	AR	154550, 602579	PMID: 9525984	prim. N-Gly affected	Sialotransferrins (S), Fatty acids and ketones (P,U), Routine tests				X			X				X			X					
MAN2B2-CDG	Core-specific lysosomal alpha-1,6-mannosidase deficiency		*MAN2B2*	AR	618899	PMID: 31775018	Recycling	Sialotransferrins (S), C-reactive protein (P), CRP (P), IgE (S)				X	X			X	X		X	X		X			X	X	
FCSK-CDG	Fucokinase deficiency		*FCSK*	AR	608675, 618324	PMID: 30503518		DNA														X					
G6PC3-CDG	Ubiquitous glucose-6-phosphatase deficiency	Dursun syndrome	*G6PC3*	AR	612541	PMID: 21385794		DNA, Abnormal neutrophil N-glycans	X		X	X	X				X		X			X				X	
GFPT1-CDG	Glutamine:fructose-6-phosphate transaminase deficiency	Congenital myasthenic syndrome type 12	*GFPT1*	AR	138292, 610542	PMID: 21310273		DNA								X					X	X					
GNE-CDG(ar)	UDP-N-acetylglucosamine-2-epimerase/N-acetylmannosamine kinase deficiency	Nonaka myopathy; GNE myopathy	*GNE*	AR	603824, 600737, 605820	PMID: 11528398		DNA, Creatine kinase (P)	X												X				X		
GNE-CDG(ad)	UDP-N-acetylglucosamine-2-epimerase/N-acetylmannosamine kinase superactivity	Sialuria, French type	*GNE*	AD	603824, 600737, 605820	PMID: 10330343		N-Acetylneuraminic acid (U), Sialic acid, free (U)				X	X									X					
GNPNAT1-CDG	Glucosamine-6-phosphate N-acetyltransferase 1 deficiency	Rhizomelic skeletal dysplasia GNPNAT1	*GNPNAT1*	AR	616510	PMID: 32591345		DNA					X									X				X	X
NANS-CDG	N-acetylneuraminic acid-9-phosphate synthase deficiency	Spondyloepimetaphyseal dysplasia, Camera-Genevieve type	*NANS*	AR	605202, 610442	PMID: 27213289		N-acetyl-D-mannosamine (P,U)					X					X					X				X	X
NPL-CDG	N-acetylneuraminate pyruvate lyase deficiency	Sialic acid aldolase deficiency	*NPL*	AR	611412, 611412	PMID: 30568043		Sialic acid, free (U)	X					X								X				X	
PGM1-CDG	Phosphoglucomutase 1 deficiency	Glycogen storage disease type 14	*PGM1*	AR	171900, 614921	PMID: 22492991		Sialotransferrins (S), N-glycans (S), Creatine kinase (P), Fatty acids and ketones (P,U)	X			X	X		X							X	X					X
PGM3-CDG	Phosphoglucomutase 3 deficiency	Immunodeficiency-23	*PGM3*	AR	172100, 615816	PMID: 24589341, PMID: 24698316		N-and O-glycans (S), IgE (S)					X						X	X		X				X	X
ALDOB-CDG	Aldolase B deficiency	Hereditary fructose intolerance	*ALDOB*	AR	229600	PMID: 3383242		Sialotransferrins (S), Uric acid (P), Glycerol (U), Coagulation factors (P), ASAT/ALAT (P)				X												X			
GALE-CDG	Uridine diphosphate galactose-4-epimerase deficiency	Galactosemia type 3	*GALE*	AR	230350	PMID: 9326324		Sialotransferrins (S), N-glycans (S), Galactose (P,U), Galactitol (U), Galactokinase (RBC)				X				X							X				
GALK1-CDG	Galactokinase deficiency	Galactosemia type 2	*GALK1*	AR	230200	PMID: 7670469		Galactose (P,U), Galactitol (U), Galactokinase (Ec)								X											
GALM-CDG	Galactose mutarotase deficiency	Galactosemia type 4	*GALM*	AR	137030	PMID: 30451973		Galactose (P,U), Galactose-1-phosphate (Ec)								X											
GALT-CDG	Galactose-1-phosphate uridyltransferase deficiency	Classic galactosemia; Galactosemia type 1	*GALT*	AR	230400	PMID: 2011574		Sialotransferrins (S), N-glycans (S), Galactose (P,U), Galactitol (U), Coagulation factors (P)				X			X	X	X		X	X		X	X				
2. Disorders of nucleotide sugar synthesis and transport																											
CAD-CDG	CAD trifunctional protein deficiency	Epileptic encephalopathy, early infantile, 50 CAD-CDG	*CAD*	AR	616457	PMID: 25678555	*De novo* biosynthesis of uridine needed for multiple pathways.	DNA											X			X					
GMPPA-CDG	GDP-mannose pyrophosphorylase subunit A deficiency	Alacrima, achalasia, and mental retardation syndrome	*GMPPA*	AR	615495, 615510	PMID: 24035193		DNA	X			X	X	X	X	X						X					
GMPPB-CDG	GDP-mannose pyrophosphorylase subunit B deficiency	Muscular dystrophy-dystroglycanopathy	*GMPPB*	AR	615320, 615350, 615351, 615352	PMID: 23768512		Hypoglycosylation of α-dystroglycan (MU), Creatine kinase (P)								X					X	X					
GFUS-CDG	GDP-L-fucose synthase deficiency		*GFUS*	AR	137020	PMID: 34468083		DNA, Abnormal fucosylation (S,WBC,PLT,FB)				X										X					X
UGDH-CDG	UDP-glucose dehydrogenase deficiency	Early infantile epileptic encephalopathy type 84	*UGDH*	AR	603370, 618792	PMID: 32001716		DNA				X	X					X			X	X				
UGP2-CDG	UDP-glucose pyrophosphorylase 2 deficiency	Early infantile epileptic encephalopathy type 83	*UGP2*	AR	191760, 618744	PMID: 31820119		DNA					X			X				X		X				
SLC35A1-CDG	CMP-sialic acid transporter deficiency	CMP-sialic acid transporter deficiency	*SLC35A1*	AR	605634, 603585	PMID: 15576474, PMID: 23873973		Sialotransferrins (S), N-glycans (S)											X			X	X				
SLC35A2-CDG	Golgi UDP-galactose transporter deficiency	Early infantile epileptic encephalopathy 22	*SLC35A2*	XL	314375	PMID: 23561849		Sialotransferrins (S), N-glycans (S)				X	X			X				X		X					
SLC35A3-CDG	UDP-N-acetylglucosamine transporter deficiency	Arthrogryposis, impaired intellectual development, and seizures	*SLC35A3*	AR	605632/ 615553	PMID: 24031089		N-glycans (S)					X									X	X			X	
SLC35C1-CDG	GDP-fucose transporter deficiency	Leukocyte adhesion deficiency syndrome type II	*SLC35C1*	AR	605881/ 266265	PMID: 11326279		N-glycans (S), Neutrophil motility and rolling, Sialyl-Lewis on neutrophils		X			X						X	X		X					X
SLC35D1-CDG	UDP-glucuronic acid-UDP-N-acetylgalactosamine dual transporter deficiency	Schneckenbecken dysplasia	*SLC35D1*	AR	610804/ 269250	PMID: 17952091		DNA				X	X													X	
SLC17A5-CDG	Sialin deficiency	Salla disease; Sialuria, Finnish type	*SLC17A5*	AR	269920	PMID: 10581036		N-Acetylneuraminic acid (U), Sialic acid, free (U)				X	X		X	X						X				X	
SLC37A4-CDG(ad)			*SLC37A4*	AD	619525	PMID: 33964207		Sialotransferrins (S), N-glycans (S), Coagulation factors (P), GGT (P)		X		X	X		X				X	X				X	X	X	X
SLC37A4-CDG(ar)	Glucose-6-phosphate transporter deficiency	Glycogen strorage disease Ib/Ic	*SLC37A4*	AR	232220, 232240	PMID: 21385794		Glycogen (LI), Biotinidase (P), 1,5-Anhydroglucitol-6-phosphate (P,U), Routine tests	X			X							X						X	X	
TGDS-CDG	TDP-D-glucose 4,6-dehydrogenase deficiency	Catel-Manzke syndrome	*TGDS*	AR	616145	PMID: 25480037		DNA	X				X												X	X
3. Disorders of N-linked protein glycosylation																											
PMM2-CDG	Phosphomannomutase 2 deficiency	Jaeken syndrome	*PMM2*	AR	601785	PMID: 9140401	Multiple, CDG-Ia	Sialotransferrins (S)	X		X	X	X		X	X	X					X	X	X		X	
MPI-CDG	Phosphomannose isomerase deficiency	Saguenay-Lac Saint-Jean syndrome	*MPI*	AR	154550, 602579	PMID: 9525984	Multiple, CDG-Ib	Sialotransferrins (S), Routine tests, Fatty acids and ketones (P,U)				X			X				X			X					
DPAGT1-CDG	UDP-GlcNAc:Dol-P-GlcNac-P transferase deficiency	Myasthenic syndrome, congenital, 13, with tubular aggregates	*DPAGT1*	AR	191350, 608093	PMID: 12872255		Sialotransferrins (S), Antithrombin III (P)				X	X			X					X	X					
ALG13-CDG	UDP-N-acetylglucosamine transferase catalytic subunit deficiency	Developmental and epileptic encephalopathy 36	*ALG13*	XL	300776, 300884	PMID: 22492991		DNA				X	X			X						X					
ALG14-CDG	UDP-GlcNAc:dolichol pyrophosphate N-acetylglucosamine transferase deficiency	Myasthenic syndrome, congenital, 15, without tubular aggregates	*ALG14*	AR	616227;612866	PMID: 23404334		DNA													X	X			X		

Gene-CDG (IEMbase hyperlink)	Name	Alternative names	Gene	Inheritance	OMIM	PMID	Comments	Diagnostic biomarkers	Cardiovascular	Dental	Dermatological	Digestive	Dysmorphic	Ear	Endocrine	Eye	Genitourinary	Hair	Hematological	Immunological	Muscular	Neurologic	Psychiatric	Renal	Respiratory	Skeletal	Short stature
ALG1-CDG	Mannosyltransferase 1 deficiency		*ALG1*	AR	605907, 608540	PMID: 14709599, PMID: 14973778, PMID: 14973782		Sialotransferrins (S), Dolichol-linked GlcNAc2 (S), Protein C (S), Xeno-tetrasaccharide (S, FB)	X			X	X			X			X		X	X		X	X	X	
ALG2-CDG	Mannosyltransferase 2 deficiency	Myasthenic syndrome, congenital, 14, with tubular aggregates	*ALG2*	AR	607905; 607906	PMID: 12684507, PMID: 23404334		Sialotransferrins (S), Dolichol-linked Man1/2 GlcNAc2 (S), Factor XI (B)								X						X					
ALG11-CDG	Mannosyltransferase 4-5 deficiency		*ALG11*	AR	613666, 613661	PMID: 20080937		Sialotransferrins (S), Dolichol-linked Man3/4GlcNAc2 (S), Prolactin (P), Factor XI (B), Antithrombin III (P)			X	X	X	X		X			X			X			X	X	
RFT1-CDG	Flippase of Man5GlcNAc2-PP-Dol deficiency	Lipid-linked oligosaccharide flippase deficiency	*RFT1*	AR	611908, 612015	PMID: 18313027		Sialotransferrins (S), Dolichol-linked Man5GlcNAc2 (S), Prolactin (P), Factor XI (B), Antithrombin III (P)				X	X	X		X			X			X					
ALG3-CDG	Mannosyltransferase 6 deficiency		*ALG3*	AR	608750, 601110	PMID: 10581255		Sialotransferrins (S), Antithrombin III (P), Apolipoprotein B (S), Protein S (S), Glucose (P)	X			X	X			X						X				X	
ALG9-CDG(ar)	Mannosyltransferase 7-9 deficiency	Gillessen-Kaesbach-Nishimura syndrome	*ALG9*	AR	606941, 608776	PMID: 15148656		Sialotransferrins (S), Lipid panel (S), Albumin (S), Factor XI (B)	X			X	X			X						X		X		X	
ALG9-CDG(ad)	Mannosyltransferase 7-9 deficiency	Polycystic liver disease	*ALG9*	AD	606941	PMID: 31395617		DNA				X															
ALG12-CDG	Mannosyltransferase 8 deficiency		*ALG12*	AR	607144, 607143	PMID: 11983712, PMID: 12217961		Sialotransferrins (S), ASAT/ALAT (P), Glucose (S), Lipid panel (S),IGF BP3, Antithrombin III (P)	X		X	X	X	X	X		X			X			X				X	
ALG5-CDG	Dolichyl-phosphate beta-glucosyltransferase deficiency	Atypical polycystic kidney disease and interstitial fibrosis	*ALG5*	AD	604565	PMID: 35896117		DNA																X			
ALG6-CDG	Glucosyltransferase 1 deficiency		*ALG6*	AR	604566, 603147	PMID: 10359825		Sialotransferrins (S), ASAT/ALAT (P), Factor XI (B)								X						X	X			X	
ALG8-CDG(ar)	Glucosyltransferase 2 deficiency		*ALG8*	AR	608103, 608104	PMID: 12480927		Sialotransferrins (S), Albumin (S), Factor IX and XII (B), Antithrombin III (P), CK (P)	X			X	X		X	X	X		X			X		X		X	
ALG8-CDG(ad)		Polycystic liver disease type 3	*ALG8*	AD	608103, 608104	PMID: 12480927		DNA				X															
ALG10-CDG	ALG10 alpha-1,2-glucosyltransferase deficiency		*ALG10*	AR	618355	PMID: 33798445		DNA														X				X	
DDOST-CDG	Subunit DDOST (OST complex) deficiency		*DDOST*	AR	602202, 614507	PMID: 22305527		Sialotransferrins (S), Factor XI (B), Proteins C/S (S)				X		X		X						X				X	
TUSC3-CDG	Oligosaccharyltransferase subunit tusc 3 deficiency	Intellectual developmental disorder, autosomal recessive 7	*TUSC3*	AR	601385, 611093	PMID: 18452889, PMID: 18455129		DNA					X									X				X	X
STT3A-CDG(ar)	Subunit STT3A (OST complex) deficiency		*STT3A*	AR	615596	PMID: 23842455		Sialotransferrins (S)				X										X				X	
STT3A-CDG(ad)	Subunit STT3A (OST complex) deficiency		*STT3A*	AD	601134	PMID: 34653363		Sialotransferrins (S)					X			X					X	X				X	X
STT3B-CDG	Subunit STT3B (OST complex) deficiency		*STT3B*	AR	608605, 615597	PMID: 23842455		Sialotransferrins (S)				X				X	X		X			X			X		
OSTC-CDG	Oligosaccharyltransferase complex deficiency		*OSTC*	AR	619023	PMID: 32267060		Sialotransferrins (S)	X				X			X						X				X	
MAGT1-CDG	Magnesium transporter 1 deficiency	Immunodeficiency, XL, with magnesium defect, Epstein-Barr virus infection and neoplasia	*MAGT1*	XL	300715, 300716;300853	PMID: 18455129		Sialotransferrins (S)												X		X					
SEC63-CDG	SEC63 homolog, protein translocation regulator deficiency	Polycystic liver disease 2	*SEC63*	AD	617004	PMID: 15133510		DNA				X															
SSR3-CDG	Translocon associated protein complex subunit SSR3 deficiency		*SSR3*	AR	606213	PMID: 30945312		Sialotransferrins (S)				X	X	X		X	X			X		X			X	X	
SSR4-CDG	Signal sequence receptor 4 protein (TRAP complex) deficiency		*SSR4*	XL	300090	PMID: 24218363		Sialotransferrins (S)			X		X			X	X					X				X	
LYSET-CDG	Lysosomal enzyme trafficking factor TMEM251 deficiency	Dysostosis multiplex, Ain-Naz type	*LYSET*	AR	619332	PMID: 33252156		Alpha-iduronidase, Iduronate-2-sulfatase (WBC)					X									X				X	X
TREX1-CDG	3' Repair exonuclease 1 deficiency (AG1) deficiency	Retinal vasculopathy with cerebral leukodystrophy	*TREX1*	AR	225750	PMID: 26320659		ASAT/ALAT (P), Blood count (B), Interferon-alpha (B), Pterins (CSF), C26:0 lysophosphatidylcholine	X		X	X				X				X		X	X	X				
MOGS-CDG	Glucosidase 1 deficiency		*MOGS*	AR	601336, 606056	PMID: 10788335		Sialotransferrins (S), N-glycans (S), ASAT/ALAT (P), Oligosaccharide (U)				X	X			X	X	X				X			X	X	
GANAB-CDG	Alpha glucosidase II deficiency	Polycyctic kidney disease 3	*GANAB*	AD	104160/ 600666	PMID: 27259053		DNA				X												X			
PRKCSH-CDG	α-1,3-glucosidase II subunit β deficiency	Polycystic liver disease type 1	*PRKCSH*	AD	177060/ 174050	PMID: 12529853, PMID: 12577059		DNA				X												X			
EDEM3-CDG	Endoplasmic reticulum degradation-enhancing alpha-mannosidase-like protein 3 deficiency		*EDEM3*	AR	610214/ 619493	PMID: 34143952	Recycling	DNA, N-glycans (S, FB)					X			X						X				X	
MAN1B1-CDG	Mannosyl-oligosaccharide alpha-1,2-mannosidase deficiency	Mental retardation, AR 15	*MAN1B1*	AR	604346, 614202	PMID: 21763484		Sialotransferrins (S), N-glycans (S, FB)					X			X						X	X				
MAN2B2-CDG	Core-specific lysosomal alpha-1,6-mannosidase deficiency		*MAN2B2*	AR	618899	PMID: 31775018	Recycling	Sialotransferrins (S), CK (P), IgE (S)				X	X			X	X		X	X		X			X	X	
MGAT2-CDG	N-acetylglucosaminyltransferase 2 deficiency		*MGAT2*	AR	602616, 212066	PMID: 8808595		Sialotransferrins (S), ASAT/ALAT (P), CK (P), Factor IX and XII (B), Antithrombin III (P), N-glycans (S, FB)	X	X		X	X	X	X	X	X	X	X	X	X	X			X	X	X
B4GALT1-CDG	Beta-1,4-galactosyltransferase 1 deficiency		*B4GALT1*	AR	137060, 607091	PMID: 11901181		Sialotransferrins (S), Antithrombin (B), Factor XI (B), N-glycans (S, FB)				X	X			X			X			X					
FUT8-CDG	α-1,6-fucosyltransferase deficiency		*FUT8*	AR	602589/ 618005	PMID: 29304374		N-glycans missing core fucose (S, FB), Glucose (P), Blood count, DNA				X	X		X	X		X			X	X		X	X	X
MAN2A2-CDG	Alpha-mannosidase IIA deficiency		*MAN2A2*	AR	600988	PMID: 36357165		DNA		X			X					X				X	X			X	
GNPTAB-CDG	UDP-N-acetylglucosamine-1-phosphotransferase subunit alpha/beta deficiency	Mucolipidosis II and mucolipidosis III alpha/beta	*GNPTAB*	AR	252500	PMID: 6461005	Lysosomal enzyme targeting	N-glycans (S), Enzyme activity (S), Oligosaccharides (U)	X	X	X	X	X	X					X			X				X	
GNPTG-CDG	UDP-N-acetylglucosamine-1-phosphotransferase subunit gamma deficiency	Mucolipidosis III gamma; Pseudo-hurler polydystrophy	*GNPTG*	AR	252605	PMID: 10712439		N-glycans (S), Enzyme activity (S), Oligosaccharides (U)					X	X		X						X				X	
4. Disorders of O-linked protein glycosylation																											
4.1 Disorders of O-mannosylation																											
POMT1-CDG	O-Mannosyltransferase 1 deficiency	Muscular dystrophy-dystroglycanopathy type A1, -type B1, -type C1	*POMT1*	AR	607423, 236670;613555;609308	PMID: 12369018		Matriglycan-specific monoclonal antibody, CK (P), DNA					X			X					X	X					

Gene-CDG (IEMbase hyperlink)	Name	Alternative names	Gene	Inheritance	OMIM	PMID	Comments	Diagnostic biomarkers	Cardiovascular	Dental	Dermatological	Digestive	Dysmorphic	Ear	Endocrine	Eye	Genitourinary	Hair	Hematological	Immunological	Muscular	Neurologic	Psychiatric	Renal	Respiratory	Skeletal	Short stature
POMT2-CDG	O-Mannosyltransferase 2 deficiency	Muscular dystrophy-dystroglycanopathy type A2, -type B2, -type C2	*POMT2*	AR	607439, 613150;613156;613158	PMID: 15894594		Matriglycan-specific monoclonal antibody, CK (P), DNA					x			x					x	x					
POMGNT1-CDG	O-Mannose beta-1,2-N-acetylglucosaminyltransferase deficiency	Muscular dystrophy-dystroglycanopathy type A3, -type B3, -type C3	*POMGNT1*	AR	606822, 253280, 613151; 613157	PMID: 11709191		Matriglycan-specific monoclonal antibody, CK (P), DNA					x			x					x	x					
POMGNT2-CDG	O-mannose β-1,4-N-acetylglucosaminyltransferase deficiency	Muscular dystrophy-dystroglycanopathy (limb-girdle) type C, 8	*POMGNT2*	AR	614828, 614830; 618135	PMID: 22958903		Matriglycan-specific monoclonal antibody, CK (P), DNA					x			x					x	x					
B3GALNT2-CDG	β-1,3-galactosaminyltransferase 2 deficiency	Muscular dystrophy-dystroglycanopathy (congenital with brain and eye anomalies, type A, 11	*B3GALNT2*	AR	610194, 615181	PMID: 23453667		Matriglycan-specific monoclonal antibody, CK (P), DNA					x			x					x	x	x				
POMK-CDG	O-mannose kinase deficiency	Muscular dystrophy-dystroglycanopathy (congenital with brain and eye anomalies), type A, 12	*POMK*	AR	615247, 616094, 615249	PMID: 23929950, PMID: 23519211		Matriglycan-specific monoclonal antibody, CK (P), DNA								x					x	x					
CRPPA-CDG	2-C-methyl-D-erythritol 4-phosphate cytidylyltransferase deficiency	Muscular dystrophy-dystroglycanopathy type A7 and C7;Walker-Warburg syndrome	*CRPPA*	AR	614631/ 614643;616052	PMID: 22522420, PMID: 22522421		Matriglycan-specific monoclonal antibody, CK (P), DNA	x				x		x	x					x	x					x
FKTN-CDG	Fukutin deficiency	Muscular dystrophy-dystroglycanopathy (limb-girdle), type C, 4 FKTN-CDG C	*FKTN*	AR	607440/ 611588	PMID: 9690476		Matriglycan-specific monoclonal antibody, CK (P), DNA	x				x			x					x	x			x		x
FKRP-CDG	Fukutin-related protein deficiency	Muscular dystrophy-dystroglycanopathy (limb-girdle), type C, 5	*FKRP*	AR	606596	PMID: 11592034		Matriglycan-specific monoclonal antibody, CK (P), DNA	x				x			x					x	x					
RXYLT1-CDG	Ribitol β1,4-xylosyltransferase deficiency	Muscular dystrophy-dystroglycanopathy (congenital with brain and eye anomalies), type A, 10; Cobblestone lissencephaly	*RXYLT1*	AR	605862/ 615041	PMID: 23217329		Matriglycan-specific monoclonal antibody, CK (P), DNA							x	x					x	x					
B4GAT1-CDG	β-1,4-glucuronyltransferase 1 deficiency	Walker-Warburg syndrome (WWS); Muscle-eye-brain disease (MEB)	*B4GAT1*	AR	605517/ 615287	PMID: 23359570		Matriglycan-specific monoclonal antibody, CK (P), DNA								x	x				x	x		x			
LARGE1-CDG	β-1,3-glucuronyltransferase-α-1,3-xylosytransferase deficiency	Muscular dystrophy-dystroglycanopathy (congenital with brain and eye anomalies), type A, 6 or (congenital with impaired intellectual development), type B, 6	*LARGE1*	AR	603590/ 613154;608840	PMID: 12966029		Matriglycan-specific monoclonal antibody, CK (P), DNA								x					x	x				x	
TMEM260-CDG	Transmembrane protein 260 deficiency	Structural heart defects and renal anomalies syndrome	*TMEM260*	AR	617478	PMID: 28318500		Creatinine (U), DNA	x					x								x		x			
TMTC3-CDG	Transmembrane and tetratricopeptide repeat domains-containing protein 3 deficiency	Lissencephaly 8	*TMTC3*	AR	617255	PMID: 27773428		DNA					x			x						x	x			
4.2 Disorders of O-GalNAc																											
C1GALT1C-CDG	Core 1 β-1,3-galactosyltransferase chaperone deficiency	Tn polyagglutination syndrome or Hemolytic uremic syndrome, atypical, 8, with rhizomelic short stature	*C1GALT1C1*	XL	300611/ 300622	PMID: 16251947, PMID: 37216524		Blood count, DNA											x	x						
GALNT2-CDG	UDP-N-acetyl-alpha-D-galactosamine:polypeptide N-acetylgalactosaminyltransferase 2 deficiency		*GALNT2*	AR	602274/ 618885	PMID: 32293671		HDL cholesterol (P), DNA					x		x							x	x				
GALNT3-CDG	Polypeptide N-acetylgalactosaminyltransferase 3 deficiency	Hyperphosphatemic familial tumoral calcinosis	*GALNT3*	AR	601756/ 211900	PMID: 15133511		Phosphate (P), DNA			x															x	
GALNT14-CDG	Polypeptide N-acetylgalactosaminyltransferase 14 deficiency		*GALNT14*	AR	608225	PMID: 26036949		DNA								x										
4.3 Disorders of O-GlcNAc																											
EOGT-CDG	EGF domain-specific O-linked N-acetylglucosamine transferase deficiency	Adams-Oliver syndrome type 4	*EOGT*	AR	614789/ 615297	PMID: 23522784		DNA	x		x		x									x				x	
OGT-CDG	O-linked N-acetylglucosamine transferase deficiency	XL mental retardation type 106	*OGT*	XL	300255/ 300997	PMID: 28302723, PMID: 28584052		DNA					x			x	x					x				x	
4.4 Disorders of O-Glc																											
POGLUT1-CDG(ar)	Autosomal recessive endoplasmic reticulum O-glucosyltransferase deficiency	Muscular dystrophy, limb-girdle, autosomal recessive 21	*POGLUT1*	AR	615618/ 615696	PMID: 27807076		DNA													x						
POGLUT1-CDG(ad)	Autosomal dominant endoplasmic reticulum O-glucosyltransferase deficiency	Dowling-Degos disease type 4	*POGLUT1*	AD	615618/ 615696	PMID: 24387993		DNA			x																
4.5 Disorders of O-Gal																											
COLGALT1-CDG	Collagen beta(1-O)galactosyltransferase 1 deficiency	Brain small vessel disease type 3	*COLGALT1*	AR	618360	PMID:30412317	adds Galactose to hydroxylysine, so it is O-linked	DNA														x					
4.6 Disorders of O-fucosylation																											
POFUT1-CDG(ar)	Autosomal recessive O-fucosyltransferase deficiency		*POFUT1*	AR	607491	PMID: 29452367		DNA	x													x				x	
POFUT1-CDG(ad)	Autosomal dominant O-fucosyltransferase deficiency	Dowling-Degos disease type 2	*POFUT1*	AD	607491	PMID: 23684010		DNA			x					x											
LFNG-CDG	O-Fucose-specific beta-1,3-N-acetylglucosaminyltransferase deficiency	Spondylocostal dysostosis type 3	*LFNG*	AR	602576/ 609813	PMID: 16385447		DNA																		x	
B3GLCT-CDG	Beta-1,3-glucosyltransferase	Peters plus syndrome	*B3GLCT*	AR	610308, 261540	PMID: 16909395		DNA	x			x	x	x	x		x					x		x		x	
4.7 Disorders of glycosaminoglycan synthesis and O-xylosylation																											
XYLT1-CDG	Xylosyltransferase 1 deficiency	Desbuquois dysplasia 2	*XYLT1*	AR	608124/ 615777	PMID: 23982343		DNA					x			x		x								x	x
XYLT2-CDG	Xylosyltransferase 2 deficiency	Spondyloocular syndrome	*XYLT2*	AR	608125/ 605822	PMID: 26027496		DNA	x					x		x						x				x	x
B4GALT7-CDG	Beta-1,4-galactosyltransferase 7 deficiency	Ehlers-Danlos syndrome with short stature and limb anomalies	*B4GALT7*	AR	604327/ 130070	PMID: 2106134		DNA		x	x		x			x						x				x	x
FAM20B-CDG	Glycosaminoglycan xylosylkinase deficiency	Neonatal short limb dysplasia FAM20B	*FAM20B*	AR	611063	PMID: 30847897		DNA					x		x							x			x	x	x
B3GALT6-CDG	β-1,3-galactosyltransferase 6 deficiency	Spondyloepimetaphyseal dysplasia with joint laxity type 1; Progeroid Ehlers-Danlos syndrome type 2	*B3GALT6*	AR	615291/ 271640;615349	PMID: 23664117		DNA	x		x		x			x						x				x	x
B3GAT3-CDG	β-1,3-glucuronyltransferase 3 deficiency	Larsen-like syndrome	*B3GAT3*	AR	606374/ 245600	PMID: 21763480		DNA	x				x									x				x	x
EXTL3-CDG	Exostosin-like glycosyltransferase 3 deficiency	Immunoskeletal dysplasia with neurodevelopmental abnormalities	*EXTL3*	AR	605744/ 617425	PMID: 28132690, PMID: 28148688, PMID: 28331220		DNA				x								x		x				x	x
EXT1-CDG	Exostosin glycosyltransferase 1 deficiency	Multiple cartilaginous exostoses type 1	*EXT1*	AD	608177/ 133700	PMID: 7550340		DNA																		x	
EXT2-CDG(ad)	Exostosin glycosyltransferase 2 (AD) deficiency	Multiple hereditary exostoses type 2	*EXT2*	AD	608210/ 133701	PMID: 7550340		DNA					x									x				x	
EXT2-CDG(ar)	Exostosin glycosyltransferase 2 (AR) deficiency	Seizures, scoliosis, and macrocephaly syndrome	*EXT2*	AR	608210	PMID: 26246518		DNA	x	x		x	x					x				x	x			x	

Gene-CDG (IEMbase hyperlink)	Name	Alternative names	Gene	Inheritance	OMIM	PMID	Comments	Diagnostic biomarkers	Cardiovascular	Dental	Dermatological	Digestive	Dysmorphic	Ear	Endocrine	Eye	Genitourinary	Hair	Hematological	Immunological	Muscular	Neurologic	Psychiatric	Renal	Respiratory	Skeletal	Short stature
NDST1-CDG	Heparan sulfate N-deacetylase-N-sulfotransferase 1 deficiency	AR mental retardation type 46	*NDST1*	AR	600853/ 616116	PMID: 25125150		DNA														x	x		x		
HS2ST1-CDG	Heparan sulfate 2-O-sulfotransferase 1 deficiency	Neurofacioskeletal syndrome with or without renal agenesis	*HS2ST1*	AR	619194	PMID: 33159882		DNA					x								x	x		x		x	x
HS6ST1-CDG	Heparan sulfate 6-O-sulfate transferase 1 deficiency	Hypogonadotropic hypogonadism type 15 with or without anosmia	*HS6ST1*	AD	604846/614880	PMID: 21700882		DNA					x	x	x							x					
HS6ST2-CDG	Heparan sulfate 6-O-sulfotransferase 2 deficiency	Paganini-Miozzo syndrome; X-linked intellectual disability	*HS6ST2*	XL	301025	PMID: 30471091		DNA					x			x						x					
CSGALNACT1-CDG	Chondroitin sulfate N-acetylgalactosaminyltransferase 1 deficiency	Skeletal dysplasia, mild, with joint laxity and advanced bone age	*CSGALNACT1*	AR	616615	PMID: 27599773		DNA	x				x									x				x	x
CHSY1-CDG	Chondroitin sulfate synthase 1 deficiency	Tentamy preaxial brachydactyly syndrome	*CHSY1*	AR	608183/ 605282	PMID: 21129728		DNA						x		x						x				x	
DSE-CDG	Dermatan sulfate epimerase deficiency	Ehlers-Danlos syndrome musculocontractural type 2	*DSE*	AR	605942/ 615539	PMID: 23704329		DNA				x	x						x			x				x	x
CHST11-CDG	Chondroitin 4-sulfotransferase 1 deficiency		*CHST11*	AR	610128	PMID: 26436107		DNA																		x	
CHST14-CDG	Dermatan 4-sulfotransferase 1 deficiency	Ehlers-Danlos syndrome musculocontractural type 1	*CHST14*	AR	608429/ 601776	PMID: 20004762, PMID: 20533528		DNA				x	x			x			x		x					x	x
CHST3-CDG	Chondroitin 6-sulfotransferase deficiency	Autosomal recessive Larsen syndrome; spondyloepiphyseal dysplasia Omani type; humerospinal dysostosis	*CHST3*	AR	603799/ 143095	PMID: 15215498		DNA	x					x												x	x
CHST6-CDG	Corneal N-acetylglucosamine 6-O-sulfotransferase deficiency	Macular corneal dystrophy	*CHST6*	AR	605294/ 217800	PMID: 11017086		DNA								x											
CANT1-CDG	UDP-galactose nucleotidase deficiency	Desbuquois dysplasia type 1; multiple epiphyseal dysplasia type 7	*CANT1*	AR	613165/ 617719	PMID: 19853239		DNA					x			x						x				x	x
SLC26A2-CDG	Sulfate transporter deficiency	Achondrogenesis type 1; atelosteogenesis type 2; diastrophic dysplasia; multiple epiphyseal dysplasia type 4	*SLC26A2*	AR	606718/ 226900;222600;256050;6009	PMID: 8528239		DNA					x	x								x			x	x	x
PAPSS2-CDG	Phosphoadenosine 5'-phosphosulfate synthetase 2 deficiency	Spondyloepimetaphyseal dysplasia, Pakistani type	*PAPSS2*	AR	603005/ 612847	PMID: 9771708		Steroids (P), DNA				x			x			x								x	x
BPNT2-CDG	Golgi-resident phosphoadenosine phosphate phosphatase deficiency	Chondrodysplasia with joint dislocations, gPAPP type	*BPNT2*	AR	614010/ 614078	PMID: 21549340		DNA					x													x	x
SLC10A7-CDG	Solute carrier family 10 member 7 deficiency	Short stature, amelogenesis imperfecta, and skeletal dysplasia with scoliosis	*SLC10A7*	AR	611459/ 618363	PMID: 30082715, PMID: 29878199		Sialotransferrins (S), N-glycans (S)																			x
SLC35B2-CDG	Phosphoadenosine 5'-phosphosulfate transporter deficiency	Leukodystrophy, hypomyelinating, 26, with chondrodysplasia	*SLC35B2*	AR	620269	PMID: 35325049		DNA					x		x						x	x				x	
5. Disorders of lipid glycosylation																											
5.1 Disorders of glycosylphosphatidylinositol biosynthesis																											
C18ORF32-CDG	Chromosome 18 open reading frame 32 deficiency	Neurodevelopmental disorder with hypotonia and contractures due to C18orf32 loss-of-function; Glycosylphosphatidylinositol biosynthesis defect 25	*C18ORF32*	AR	619985	PMID: 35107634		Alkaline phosphatase (P), DNA					x			x		x				x				x	
PIGA-CDG	Phosphatidylinositol glycan anchor biosynthesis class A protein deficiency	Multiple congenital anomalies-hypotonia-seizures syndrome type 2; GPI biosynthesis defect type 4; Early infantile epileptic encephalopathy type 20	*PIGA*	XL	311770, 300868;300818	PMID: 8500164, PMID: 22305531		GPI-anchored proteins (WBC,FB), ALP (P),				x	x									x					
PIGC-CDG	Glycosylphosphatidylinositol glycan anchor biosynthesis C protein deficiency	Developmental disability, severe intellectual disability, and drug-responsive epilepsy; GPI biosynthesis defect type 16; autosomal recessive intellectual disability type 62	*PIGC*	AR	601730, 615716	PMID: 27694521		Flow cytometry of GPI markers (PLT)					x									x					
PIGQ-CDG	Phosphatidylinositol glycan anchor biosynthesis class Q protein deficiency	Multiple congenital anomalies-hypotonia-seizures syndrome 4	*PIGQ*	AR	605754	PMID: 24463883		Flow cytometry of GPI markers (PLT), ALP (P), DNA,								x						x					
PIGH-CDG	Phosphatidylinositol glycan anchor biosynthesis class H protein deficiency	Glycosylphosphatidylinositol biosynthesis defect 17	*PIGH*	AR	600154, 618010	PMID:29573052, PMID:29603516		Triglycerides (S), ALP (P), DNA														x	x				
PIGP-CDG	Phosphatidylinositol glycan anchor biosynthesis class P protein deficiency	GPI biosynthesis defect type 14; Early infantile epileptic encephalopathy type 55	*PIGP*	AR	605938, 617599	PMID: 28334793		Flow cytometry of GPI markers (PLT)														x					
PIGY-CDG	Phosphatidylinositol glycan anchor biosynthesis class Y, deficiency	Hyperphosphatasia with intellectual disability type 6; GPI biosynthesis defect type 12	*PIGY*	AR	610662, 239300	PMID: 26293662		DNA, ALP (P)					x									x				x	
PIGL-CDG	Phosphatidylinositol glycan anchor biosynthesis class L protein deficiency	CHIME syndrome; GPI biosynthesis defect type 5	*PIGL*	AR	605947, 280000	PMID: 22444671		GPI-anchored proteins (Lc, F, ALP (P),	x		x			x		x						x					
PIGW-CDG	Glycosylphosphatidylinositol glycan anchor biosynthesis W protein deficiency	Hyperphosphatasia with intellectual disability type 5; GPI biosynthesis defect type 11	*PIGW*	AR	610275, 616025	PMID: 24367057		Flow cytometry of GPI markers (PLT)														x					
PIGM-CDG	Phosphatidylinositol glycan anchor biosynthesis class M, deficiency	Glycosylphosphatidylinositol deficiency; GPI biosynthesis defect type 1	*PIGM*	AR	610273, 610293	PMID: 16767100		Flow cytometry of GPI markers (PLT)				x							x			x					
ARV1-CDG	ARV1 homolog deficiency	Developmental and epileptic encephalopathy 38	*ARV1*	AR	617020	PMID: 32165008		Flow cytometry of GPI markers (PLT)								x					x	x					
PIGV-CDG	Phosphatidylinositol glycan anchor biosynthesis class V protein deficiency	Hyperphosphatasia with mental retardation type 1; GPI biosynthesis defect type 2	*PIGV*	AR	610274, 239300	PMID: 20802478		GPI-anchored proteins (WBC,FB), ALP (P)				x	x	x								x				x	
PIGN-CDG	Phosphatidylinositol glycan anchor biosynthesis class N protein deficiency	Multiple congenital anomalies-hypotonia-seizures syndrome type 1; GPI biosynthesis defect type 3	*PIGN*	AR	606097, 614080	PMID: 21493957		GPI-anchored proteins (WBC,FB), ALP (P)	x				x									x			x	x	
PIGB-CDG	Phosphatidylinositol glycan anchor biosynthesis class B protein deficiency	Developmental and epileptic encephalopathy type 80	*PIGB*	AR	604122	PMID: 31256876		GPI-anchored proteins (WBC,FB), ALP (P)				x		x		x						x				x	
PIGO-CDG	Phosphatidylinositol glycan anchor biosynthesis class O protein deficiency	Hyperphosphatasia with mental retardation type 2; GPI biosynthesis defect type 6	*PIGO*	AR	614730, 614749	PMID: 22683086		GPI-anchored proteins (WBC,FB), ALP (P)	x			x	x	x								x				x	
PIGF-CDG	Phosphatidylinositol glycan anchor biosynthesis class F protein deficiency	Onychodystrophy, osteodystrophy, impaired intellectual development and seizures syndrome; GPI biosynthesis defect type 24	*PIGF*	AR	600153, 619356	PMID: 33386993		Flow cytometry of GPI markers (PLT)				x		x								x				x	
PIGG-CDG	Glycosylphosphatidylinositol glycan anchor biosynthesis G protein deficiency	Mental retardation, AR 53; GPI biosynthesis defect 13	*PIGG*	AR	616918, 616917	PMID: 26996948		DNA														x					
PIGT-CDG	Phosphatidylinositol glycan anchor biosynthesis class T protein deficiency	Multiple congenital anomalies-hypotonia-seizures syndrome type 3; GPI biosynthesis defect type 7	*PIGT*	AR	610272, 615398	PMID: 23636107		GPI-anchored proteins (WBC,FB), ALP (P)	x				x			x			x	x		x				x	
PIGS-CDG	Phosphatidylinositol glycan anchor biosynthesis class S protein deficiency	Developmental and epileptic encephalopathy type 95	*PIGS*	AR	610271, 618143	PMID: 30269814		Flow cytometry of GPI markers (PLT)					x			x					x	x				x	
PIGU-CDG	Phosphatidylinositol glycan anchor biosynthesis class U protein deficiency	Neurodevelopmental disorder with brain anomalies, seizures, and scoliosis	*PIGU*	AR	608528, 618590	PMID: 31353022		Flow cytometry of GPI markers (PLT)					x			x						x				x	
PIGK-CDG	Phosphatidylinositol glycan anchor biosynthesis class K protein deficiency	Neurodevelopmental disorder with hypotonia and cerebellar atrophy, with or without seizures	*PIGK*	AR	605087, 618879	PMID: 32220290		Flow cytometry of GPI markers (PLT)					x		x	x		x				x					

Gene-CDG (IEMbase hyperlink)	Name	Alternative names	Gene	Inheritance	OMIM	PMID	Comments	Diagnostic biomarkers	Cardiovascular	Dental	Dermatological	Digestive	Dysmorphic	Ear	Endocrine	Eye	Genitourinary	Hair	Hematological	Immunological	Muscular	Neurologic	Psychiatric	Renal	Respiratory	Skeletal	Short stature
GPAA1-CDG	Glycosylphosphatidylinositol anchor attachment protein 1 deficiency	GPI biosynthesis defect type 15	*GPAA1*	AR	603048, 617810	PMID: 29100095		Flow cytometry of GPI markers (PLT)								X						X				X	
PGAP1-CDG	GPI deacylase deficiency	Autosomal recessive intellectual disability type 42; GPI biosynthesis defect type 9	*PGAP1*	AR	611655, 615716	PMID: 24784135		DNA					X									X			X	X	
PGAP3-CDG	PER1-like domain-containing protein 1 deficiency	Hyperphosphatasia with intellectual disability type 4; GPI biosynthesis defect type 1	*PGAP3*	AR	611801	PMID: 24439110		GPI-anchored proteins (WBC,FB), ALP (P)					X									X					
PGAP2-CDG	Post-GPI attachment to proteins 2 deficiency	Hyperphosphatasia with intellectual disability type 3; GPI biosynthesis defect type 8	*PGAP2*	AR	615187, 614207	PMID: 23561846, PMID: 23561847		ALP (P)						X								X					
5.2 Disorders of glycosphingolipid synthesis																											
ST3GAL5-CDG	Lactosylceramide alpha-2,3-sialyltransferase deficiency	GM3 synthase deficiency; Amish infantile epilepsy syndrome; salt and pepper developmental regression syndrome	*ST3GAL5*	AR	604402/ 609056	PMID: 15502825		Lactosylceramide (P), GM3 ganglioside (P)			X			X		X						X					
B4GALNT1-CDG	GM2-GD2 synthase deficiency	GM2/GD2 synthase deficiency; autosomal recessive spastic paraplegia type 26	*B4GALNT1*	AR	601873/ 609195	PMID: 23746551		GM2/GM3 gangliosides (FB)							X	X						X				X	
ST3GAL3-CDG	GD1a-GT1b synthase deficiency	Mental retardation, AR 12; Epileptic encephalopathy, early infantile, 15	*ST3GAL3*	AR	606494/ 611090	PMID: 21907012, PMID: 23252400		DNA														X					
A4GALT-CDG	GB3 synthase deficiency	GB3 synthase deficiency; NOR polyagglutination syndrome	*A4GALT*	AD	607922/ 111400	PMID: 22965229		DNA												X							
6. Disorders of vesicular trafficking																											
COG1-CDG	Conserved oligomeric Golgi complex subunit 1 deficiency		*COG1*	AR	606973/ 611209	PMID: 16537452, PMID: 19008299		Sialotransferrins (S)	X			X	X						X			X					X
COG2-CDG	Conserved oligomeric Golgi complex subunit 2 deficiency		*COG2*	AR	606974	PMID: 24784932		Sialotransferrins (S), Ceruloplasmin (S), Copper (S)				X	X		X				X			X					
COG3-CDG	Conserved oligomeric Golgi complex subunit 3 deficiency		*COG3*	AR	606975	PMID: 37711075		Sialotransferrins (S)				X	X			X						X					
COG4-CDG(ar)	Conserved oligomeric Golgi complex subunit 4 deficiency		*COG4*	AR	606976/ 613489	PMID: 19494034		Sialotransferrins (S)														X					
COG4-CDG(ad)	Saul-Wilson syndrome	Saul-Wilson syndrome; COG4-SWS	*COG4*	AD	606976	PMID: 30290151		DNA					X			X						X			X	X
COG5-CDG	Conserved oligomeric Golgi complex subunit 5 deficiency		*COG5*	AR	606821/613612	PMID: 19690088		Sialotransferrins (S)			X	X		X		X	X					X			X	X
COG6-CDG	Conserved oligomeric Golgi complex subunit 6 deficiency	Shaheen syndrome	*COG6*	AR	606977/ 614576	PMID: 20605848		Sialotransferrins (S), ASAT/ALAT (P), CK (P), Lactate (P), Vitamins A, D, E, K (S)		X	X	X	X		X				X	X		X					
COG7-CDG	Conserved oligomeric Golgi complex subunit 7 deficiency		*COG7*	AR	606978/ 608779	PMID: 15107842		Sialotransferrins (S), ASAT/ALAT (P), CK (P), Glucose (S), Total/direct bilirubin (S)	X		X	X	X				X			X		X		X			
COG8-CDG	Conserved oligomeric Golgi complex subunit 8 deficiency		*COG8*	AR	606979/ 611182	PMID: 17331980, PMID: 17220172		Sialotransferrins (S), Factor XI (B), Protein C (S)								X				X		X				X	X
STX5-CDG	Syntaxin-5 deficiency		*STX5*	AR	620454	PMID: 34711829		Sialotransferrins (S), N-glycans (S), Alkaline phosphatase (P), IGF1 (P), Insulin (P), Coagulation factors (P), Ammonia (P)	X			X			X							X		X		X		
CAMLG-CDG	Calcium-modulating cyclophilin ligand deficiency		*CAMLG*	AR	601118	PMID: 35262690		Sialotransferrins (S)													X	X			X		
GET3-CDG	Guided entry of tail-anchored proteins factor 3 deficiency		*GET3*	AR	601913	PMID: 31461301		Sialotransferrins (S)	X			X										X			X		
GET4-CDG	Guided entry of tail-anchored proteins factor 4 deficiency		*GET4*	AR	612056	PMID: 32395830		Sialotransferrins (S), N-Acetyl aspartate (CNS)					X					X				X				X	
JAGN1-CDG	Jagunal 1 deficiency	Severe congenital neutropenia type 6	*JAGN1*	AR	616022	PMID: 25129144, PMID: 25129145		Abnormal neutrophil N-glycans											X	X							
TRAPPC11-CDG	Trafficking protein particle complex, subunit 11 deficiency	Muscular dystrophy, limb-girdle, type 2S	*TRAPPC11*	AR	615356	PMID: 23830518, PMID: 27707803		CK (P), DNA				X	X								X	X				X	
TRAPPC9-CDG	Trafficking protein particle complex 9 deficiency	Mental retardation, AR 13	*TRAPPC9*	AR	613192	PMID: 35042660		DNA					X					X				X				X	
SEC23B-CDG	COPII component SEC23B deficiency	Congenital dyserythropoietic anemia type 2	*SEC23B*	AR	224100	PMID: 19561605		Bilirubin (P), Multinucleated erythroblasts (bone marrow)	X			X			X				X								
TRIP11-CDG	Thyroid hormone receptor coactivator complex deficiency	Achondrogenesis type IA, Houston-Harris Type; odontochondrodysplasia	*TRIP11*	AR	200600	PMID: 20089971		DNA																		X	
VPS13B-CDG	Vacuolar protein sorting 13B deficiency	Pepper syndrome; Cohen syndrome	*VPS13B*	AR	216550	PMID: 24334764		N-glycans (S)								X			X							X	
GOSR2-CDG	Golgi SNAP receptor complex member 2 deficiency	Epilepsy, progressive myoclonic 6	*GOSR2*	AR	614018	PMID: 21549339		DNA														X					
GM130-CDG	Golgin A2 deficiency	Developmental delay with hypotonia, myopathy, and brain abnormalities	*GM130*	AR	602580	PMID: 26742501		CK (P), DNA					X								X	X					
VPS51-CDG	VPS51 subunit of GARP complex deficiency	Pontocerebellar hypoplasia, type 13	*VPS51*	AR	615738	PMID: 30624672, PMID: 31207318		DNA, N and O-glycans		X			X			X						X				X	
VPS53-CDG	VPS53 subunit of GARP complex deficiency	Pontocerebellar hypoplasia, type 2E	*VPS53*	AR	615850	PMID: 24577744		DNA														X	X			X	X
7. Disorders of multiple glycosylation pathways																											
7.1 Disorders of dolichol metabolism																											
DHDDS-CDG(ar)	Autosomal recessive Dehydrodolichyl diphosphate synthase deficiency	Retinitis pigmentosa 59	*DHDDS*	AR	613861; 608172; 617836	PMID: 21295282, PMID: 21295283		DNA								X	X					X		X			
DHDDS-CDG(ad)	Autosomal dominant Dehydrodolichyl diphosphate synthase deficiency	Developmental delay and seizures with or without movement abnormalities	*DHDDS*	AD	613861; 608172; 617836	PMID: 34382076		DNA														X	X				
NUS1-CDG(ar)	Autosomal recessive Nogo-B receptor deficiency		*NUS1*	AR	610463/ 617082	PMID: 25066056		DNA								X						X				X	
NUS1-CDG(ad)	Autosomal dominant Nogo-B receptor deficiency	Intellectual developmental disorder, autosomal dominant 55, with seizures	*NUS1*	AD	610463/ 617082	PMID: 29100083		DNA														X					
SRD5A3-CDG	Steroid 5 alpha-reductase 3 deficiency	Ocular coloboma with ichthyosis, brain malformations and endocrine abnormalities	*SRD5A3*	AR	611715/ 612379	PMID: 20637498		Sialotransferrins (S), Antithrombin III (P), Protein S (S)				X		X		X	X					X	X			X	
DOLK-CDG	Dolichol kinase deficiency		*DOLK*	AR	610746/ 610768	PMID: 17273964		Sialotransferrins (S), Lipid-linked oligosaccharide (F)	X			X		X		X	X		X				X				

Gene-CDG (IEMbase hyperlink)	Name	Alternative names	Gene	Inheritance	OMIM	PMID	Comments	Diagnostic biomarkers	Cardiovascular	Dental	Dermatological	Digestive	Dysmorphic	Ear	Endocrine	Eye	Genitourinary	Hair	Hematological	Immunological	Muscular	Neurologic	Psychiatric	Renal	Respiratory	Skeletal	Short stature
DPM1-CDG	GDP-Man:Dol-P mannosyltransferase subunit 1 deficiency		*DPM1*	AR	603503/ 608799	PMID: 10642597, PMID: 10642602		Sialotransferrins (S), Factor XI (B)				X				X						X					
DPM2-CDG	Dolichol-P-mannose synthase-2 deficiency		*DPM2*	AR	603564/ 615042	PMID: 23109149		Sialotransferrins (S), ASAT/ALAT (P), CK (P)				X	X			X				X	X	X			X	X	
DPM3-CDG	GDP-Man:Dol-P mannosyltransferase 3 deficiency		*DPM3*	AR	605951/ 612937	PMID: 19576565		Sialotransferrins (S), Dolichol-P-mannose (S), Factor XI (B), ASAT/ALAT (P), CK (P)	X				X									X	X				X	
MPDU1-CDG	Dol-P-Man utilization 1 deficiency		*MPDU1*	AR	604041/ 609180	PMID: 11733556, PMID: 11733564		Sialotransferrins (S), Antithrombin III (P)				X	X		X	X						X				X	
7.2 Disorders of Golgi homeostasis																											
ATP6V0A2-CDG	Lysosomal H(+)-transporting ATPase V0 subunit A2 deficiency	Cutis laxa, AR, type IIA; wrinkly skin syndrome	*ATP6V0A2*	AR	611716/ 219200;278250	PMID: 18157129		Sialotransferrins (S), ASAT/ALAT (P), N and O-glycans (S)				X		X		X	X						X			X	
ATP6V1A-CDG	V-ATPase A subunit 1 deficiency	Cutis laxa, AR, type IID	*ATP6V1A*	AR	607027/ 617403	PMID: 28065471		Sialotransferrins (S), Lipid panel (S)	X			X		X			X						X	X			X	
ATP6V1E1-CDG	ATPase subunit E, ATP6E deficiency	Cutis laxa, AR, type IIC	*ATP6V1E1*	AR	108746/ 617402	PMID: 28065471		Sialotransferrins (S)	X			X		X			X						X	X			X	
ATP6AP1-CDG	Accessory subunit 1 of the vacuolar-ATPase protein deficiency	Immunodeficiency 47 and hepatopathy with or without neurologic features	*ATP6AP1*	XL	300197/ 300972	PMID: 27231034		Sialotransferrins (S), Ceruloplasmin (S), Copper (S), IgG (S), N and O-glycans (S)				X	X							X	X		X					
ATP6AP2-CDG	ATPase, H+ transporting, lysosomal, accessory protein 2 deficiency	XL mental retardation, Hedera type	*ATP6AP2*	XL	300556/ 300423	PMID: 29127204		Sialotransferrins (S), ASAT/ALAT (P), IgG (S), Factor XI (B), N and O-glycans (S)				X	X	X				X					X					
TMEM199-CDG	Transmembrane protein 199 deficiency		*TMEM199*	AR	616815/ 616829	PMID: 26833330		Sialotransferrins (S), ALP (P), Ceruloplasmin (S), N and O-glycans (S)				X															
CCDC115-CDG	Coiled-coil domain-containing protein 115 deficiency		*CCDC115*	AR	613734/ 616828	PMID: 26833332		Sialotransferrins (S), ALP (P), Ceruloplasmin (S), Cholesterol (S), N and O-glycans (S)				X	X			X							X	X				
VMA21-CDG	Vacuolar ATPase assembly factor VMA21 deficiency	Myopathy, XL, with excessive autophagy	*VMA21*	XL	300913/ 310440	PMID: 32145091		Sialotransferrins (S), N and O-glycans (S), ASAT/ALAT (P), CK (P)													X						
SLC39A8-CDG	Solute carrier family 39 (Zn transporter) deficiency		*SLC39A8*	AR	616721	PMID: 26637978, PMID: 26637979		Sialotransferrins (S), Zinc (S, U), Manganese (B), Lactate (P)								X				X		X				X	X
TMEM165-CDG	TMEM165 (TPARL) protein deficiency		*TMEM165*	AR	614726/ 614727	PMID: 22683087		Sialotransferrins (S), ASAT/ALAT (P), CK (P), N-glycans (S)				X	X		X					X	X	X				X	
SLC9A7-CDG	Sodium/hydrogen exchanger 7 deficiency	Nonsyndromic intellectual disability due to SLC9A7 de novo variant	*SLC9A7*	XL	300368	PMID: 30335141		DNA													X	X				X	
SLC37A4-CDG(ad)	Glucose-6-phosphate transporter deficiency		*SLC37A4*	AD	602671/ 619525	PMID: 33964207		ASAT/ALAT (P), CK (P), Lactate (P), Glucose (P), Biotinidase (P)	X			X	X						X						X	X	
8. Disorders of Glycoprotein/Glycan Degradation																											
NGLY1-CDDG	N-glycanase 1 deficiency		*NGLY1*	AR	610661/ 615273	PMID: 22581936		Oligosaccharides (U), GlcNAc-Asn (U)				X	X		X	X						X				X	
MAN2C1-CDG	Mannosidase, alpha, class 2C, member 1 deficiency		*MAN2C1*	AR	619775	PMID: 35045343		DNA				X	X			X						X	X				