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ERRATUM

Erratum to: What is new in CDG?

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Due to a typesetting error, part of Table 2 is missing in the pdf file. The online version of Table 2 is correct.

The original article was corrected.

The online version of the original article can be found under doi:10.1007/s10545-017-0050-6

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Table 2 List of all CDG reported up to date according to their cellular location and the different pathways. The asterisk (*) refers to recently reported CDG which are highlighted in this review. The CDG for which there is a causative treatment are highlighted in yellow

Table 2.1 Defects localized in the cytosol

NAME	CLINICALLY AFFECTED ORGANS AND TISSUES	DEFECTIVE PROTEIN	TF-IEF	MOI
Protein N-glycosylation disorders				
Synthesis of monosaccharides				
GMPPA-CDG	Autonomic nerve fibers of distal oesophagus (achalasia) and lacrimal glands (alacrimia), neurons (brain, hearing system, visual system)	Guanosine diphosphate mannose pyrophosphorylase A		
GMPPB-CDG	Brain, skeletal muscles, eyes, heart	Guanosine diphosphate mannose pyrophosphorylase B		
MPI-CDG	Intestine, liver	Mannose 6-phosphate isomerase	Type 1	
PMM2-CDG	Nervous system, fat tissue, and nearly all other organs	Phosphomannomutase 2	Type 1	
Protein O-glycosylation disorders				
Defects in O-N-acetylglucosaminylglycan synthesis				
EOGT-CDG	Skin (aplasia cutis congenita), skeleton (terminal transverse limb defect)	EGF domain-specific O-GlcNAc transferase		
Defects in multiple and other glycosylation pathways				
Defects in dolichol synthesis				
DHDDS-CDG* (retinitis pigmentosa 59)	Retina	Dehydrodolichyl diphosphate	Type 1	
Defects in monosaccharide synthesis				
CAD-CDG*	Brain, blood cells	Carbamoyl-phosphate synthetase 2 (CPS2) and aspartate transcarbamylase (ATCase) activities of the trifunctional enzyme CAD		
GFPT1-CDG (limb girdle congenital myasthenic syndrome)	Neuromuscular junction, skeletal muscles	Glutamine:fructose 6-phosphate amidotransferase 1		
GNE-CDG (hereditary inclusion body myopathy)	Skeletal muscles (with sparing of quadriceps muscles), rarely cardiac muscles	UDP-GlcNAc 2-epimerase/Man-NAc kinase		
NANS-CDG*	Brain, skeleton	N-acetylneuraminic acid synthase		
PGM1-CDG	Uvula (palate, lips), heart, liver, muscles, endocrine organs	Phosphoglucomutase 1	Type 2	
PGM3-CDG*	Brain, immune system, skeleton	Phosphoglucomutase 3		
Defects in nucleotide-sugar synthesis				
CPS2-CDG	Brain, intestine, kidneys, erythrocytes	Carbamylphosphate synthetase 2 deficiency		

Table 2.2 Defects localized in the ER

NAME	CLINICALLY AFFECTED ORGANS AND TISSUES	DEFECTIVE PROTEIN	TF-IEF	MOI
Protein N-glycosylation disorders				
ALG1-CDG	Brain, and variable involvement of eyes, heart, liver, beta cells, kidneys, gonads	Mannosyltransferase 1	Type 1	
ALG2-CDG	Brain, eyes, skeletal muscles, neuromuscular junction (congenital myasthenic syndrome)	Mannosyltransferase 2	Type 1	
ALG3-CDG	Brain, skeleton	Mannosyltransferase 6	Type 1	
ALG6-CDG	Brain, and variable involvement of eyes, gastrointestinal system, liver, heart and skeleton	Glucosyltransferase 1	Type 1	
ALG8-CDG	Brain, and variable involvement of eyes, skin, liver and intestine	Glucosyltransferase 2	Type 1	
ALG9-CDG*	Brain, liver, kidneys, and variable involvement of adipose tissue, heart, skeleton, intestine	Mannosyltransferase 7/9	Type 1	
ALG11-CDG	Brain, hearing system	Mannosyltransferase 4/5	Type 1	
ALG12-CDG	Brain, skeleton, heart, genitalia and immune system	Mannosyltransferase 8	Type 1	
ALG13-CDG	Brain, eyes, liver	UDP-GlcNAc:Dol-P-GlcNAc-P transferase	Type 1	X-linked
ALG14-CDG	Neuromuscular junction (congenital myasthenic syndrome)	UDP-GlcNAc:Dol-PP-GlcNAc transferase	Type 1	
DDOST-CDG	Brain, eyes, liver	Oligosaccharyltransferase subunit DDOST	Type 1	
DPAGT1-CDG	Brain, neuromuscular junction (congenital myasthenic syndrome)	UDP-GlcNAc: Dol-P-GlcNAc-P	Type 1	
MOGS-CDG	Brain, skeleton, immune system	Mannosyl-oligosaccharide glycosidase (glucosidase 1)		
GANAB-CDG*	Liver, kidneys (polycystic)	Glucosidase II subunit α		AD
PRKCSH-CDG	Liver, kidneys (polycystic)	Glucosidase II subunit β		AD

RFT1-CDG	Brain, hearing system	Flippase of Man5GlcNAc2-PP-Dol	Type 1	
STT3A-CDG	Brain, gastrointestinal tract	Oligosaccharyltransferase subunit STT3A	Type 1	
STT3B-CDG	Brain, optic nerve, gastrointestinal tract	Oligosaccharyltransferase subunit STT3B	Type 1 (mild)	
SSR3-CDG	Brain, lungs, gastrointestinal system	Signal sequence receptor 3 of TRAP complex	Type 1 (disialo increase only)	
SSR4-CDG*	Brain, respiratory system, skeleton	Signal sequence receptor 4 of TRAP complex	Type 1 (mild)	X-linked
TUSC3-CDG	Brain (non-syndromic autosomal recessive mental disability)	Oligosaccharyltransferase subunit TUSC3	Type 1	
Protein O-glycosylation disorders				
Defect in O-xylosyl/N-acetylgalactosaminylglycan synthesis				
SLC35D1-CDG (Schneckenbecken dysplasia)	Skeleton (generalized; radiographic snail-like configuration of iliac bones)(stillborn or lethal in the neonatal period)	Solute carrier family 35 (UDP-glucuronic acid/UDP-N-acetylgalactosamine dual transporter) member D1		
Defect in O-glucosylglycan synthesis				
POGLUT1-CDG*	Skin (progressive reticular hyper- and hypopigmentation)	Protein O-glucosyltransferase 1		AD
Defects of lipid glycosylation and of glycosylphosphatidylinositol synthesis				
Defects in glycosylphosphatidylinositol synthesis				
PIGA-CDG*	Brain, heart, liver, kidneys, skin	UDP-GlcNAc:phosphatidylinositol N-acetylglucosaminyltransferase subunit		X-linked
PIGC-CDG*	Brain	UDP-GlcNAc:phosphatidylinositol N-acetylglucosaminyltransferase subunit		
PIGG-CDG*	Brain	Glycosylphosphatidylinositol ethanolamine phosphate transferase 2		
PIGL-CDG (CHIME syndrome)	Brain, eyes, hearing system, heart, skin	GlcNAc-phosphatidylinositol de-acetylase		
PIGM-CDG	Brain, hepatic veins	Dol-P-Man:phosphatidylinositol mannosyltransferase 1		
PIGN-CDG	Brain, skeleton (including palate, fingers), cardiovascular system, kidneys	Glycosylphosphatidylinositol ethanolamine phosphate transferase 1		
PIGO-CDG	Brain, lips, fingers, toes, anus/rectum, hearing system, cardiovascular system	Glycosylphosphatidylinositol ethanolaminephosphate transferase 3		
PIGQ-CDG	Brain	UDP-GlcNAc:phosphatidylinositol N-acetylglucosaminyltransferase subunit		
PIGT-CDG	Brain, eyes, heart, kidneys, skeleton	PIGT transamidase subunit		
PIGV-CDG	Brain, fingers, toes, and less frequent involvement of lips, palate, anus/rectum, hearing system	Dol-P-Man:phosphatidylinositol mannosyltransferase 2		
PIGW-CDG*	Brain, skeleton	Phosphatidylinositol acylase		
PIGY-CDG*	Brain, fingers, toes	UDP-GlcNAc:phosphatidylinositol N-acetylglucosaminyltransferase subunit		
PGAP1-CDG	Brain	Phosphatidylinositol deacylase		
Defects in multiple and other glycosylation pathways				
Defects in dolichol synthesis				
DOLK-CDG	Brain, heart, skin	Dolichol kinase	Type 1	
NUS1-CDG*	Brain, eyes (bilateral macular lesions), skeleton	Nogo-B receptor (subunit of cis-prenyltransferase)	Type 1	
SRD5A3-CDG	Brain, eyes, heart, skin, joints	Steroid 5 α-reductase 3	Type 1	
Defects in dolichol utilization/recycling				
DPM1-CDG	Brain, eyes, skeletal muscles	GDP-Man:Dol-P-mannosyltransferase 1 (Dol-P-Man synthase 1)	Type 1	
DPM2-CDG	Brain, skeletal muscles	GDP-Man:Dol-P-mannosyltransferase 2 (Dol-P-Man synthase 2)	Type 1	
DPM3-CDG	Skeletal and cardiac muscles	GDP-Man:Dol-P-mannosyltransferase 3 (Dol-P-Man synthase 3)	Type 1	
MPDU1-CDG	Brain, eyes, skin	Man-P-Dol utilization 1	Type 1	
Defects in the v-ATPase complex				
ATP6AP1-CDG*	Brain, B cells, liver (muscles, hearing system)	Accessory protein Ac45 of the V-ATPase	Type 2	X-linked
Other defects				
TRAPPC11-CDG*	Muscles (limb girdle muscular dystrophy, type 2S)	Trafficking protein particle complex (TRAPPIII), subunit 11	NA.	

Table 2.3 Defects localized in the Golgi				
NAME	CLINICALLY AFFECTED ORGANS AND TISSUES	DEFECTIVE PROTEIN	TF-IEF	MOI
Protein N-glycosylation disorders				
MAN1B1-CDG	Brain, cranial skeleton, fat tissue	Golgi α1-2 mannosidase 1	Type 1	
MGAT2-CDG	Brain, skeleton, intestine, immune system	N-Acetylglucosaminyltransferase 2	Type 2	
Protein O-glycosylation disorders				
Defect in O-xylosylglycan synthesis				
B4GALT7-CDG	Brain, skeleton (short stature, bowing of extremities), articulations (hyperlaxity, dislocations), skin (premature aging phenotype)	B-1,4-galactosyltransferase 7		
B3GALT6-CDG	Skeleton (spondyloepimetaphyseal dysplasia with bone fragility, severe kyphoscoliose), joints, skin (fragility, delayed wound healing)	B-1,3-galactosyltransferase 6		
B3GAT3-CDG	Brain, aorta, heart, skeleton, joints, skin, teeth	B-1,3-glucuronyltransferase 3		
CHSY1-CDG (Tentamy preaxial brachydactyly syndrome)	Brain, teeth, skeleton (particularly brachydactyly), hearing system	Chondroitin β-1,4-N-acetylgalactosaminyltransferase 1 (chondroitin synthase 1)		
EXT1-CDG (multiple cartilaginous exostoses)	Cartilage (osteochondromas of the ends of long bones)	Exostosin 1		AD
EXT2-CDG* (multiple cartilaginous exostoses)	Cartilage (osteochondromas of the ends of long bones)	Exostosin 2		AD
XYLT1-CDG*	Brain, skeleton (short stature, advanced bone age), articulations (joint laxity), fat	Xylosyltransferase 1		
XYLT2-CDG*	Brain, eyes, heart, hearing system, bones	Xylosyltransferase 2		
Defect in O-N-acetylgalactosaminalglycan synthesis				
GALNT3-CDG (familial hyperphosphatemic tumoral calcinosis)	Subcutaneous tissue (painful calcified masses)	UDP-N-acetyl-α-D-galactosamine:polypeptide N-acetylgalactosaminyltransferase 3		
Defects in O-fucosylglycan synthesis				
LFNG-CDG (spondylocostal dysostosis type 3)	Axial skeleton, associated muscles	O-fucose-specific β-1,3-N-acetylglucosaminyltransferase		
POFUT1-CDG (Dowling-Degos disease 2)	Skin (progressive reticular hyper- and hypopigmentation)	Protein O-fucosyltransferase 1		AD
Defects of lipid glycosylation and of glycosylphosphatidylinositol synthesis				
Defects in lipid glycosylation				
B4GALNT1-CDG (spastic paraplegia 26, autosomal recessive)	Brain, peripheral nerves (spastic paraplegia), gonads	B-1,4-N-acetylgalactosaminyltransferase 1 (GM2 synthase)		
ST3GAL5-CDG (Amish infantile epilepsy; salt and pepper syndrome)	Brain, hearing system, skin	Lactosylceramide α-2,3-sialyltransferase (GM3 synthase)		
Defects in glycosylphosphatidylinositol synthesis				
PGAP2-CDG	Brain	Phosphatidylinositol glycerol acylase		
PGAP3-CDG*	Brain, skeleton	Phosphatidylinositol glycerol deacylase		
Defects in multiple and other glycosylation pathways				
Defects in glycosyltransferases				
B4GALT1-CDG	Face (dysmorphism), eyes (myopia)	B-1,4-galactosyltransferase	Type 2	
ST3GAL3-CDG	Brain	B-galactoside α-2,3-sialyltransferase 3		
Defects in nucleotide-sugar transporters				
SLC35A1-CDG	Brain, heart, kidneys, platelets	CMP-sialic acid transporter	Type 2	
SLC35A2-CDG	Brain, eyes, gastrointestinal system, skeleton	UDP-galactose transporter	Type 2	X-linked
SLC35A3-CDG	Brain, skeleton	UDP-GlcNAc transporter		
SLC35C1-CDG	Brain, cranial skeleton, neutrophils	GDP-fucose transporter		
Defects in the COG complex				
COG1-CDG	Brain, skeleton	COG component 1	Type 2	
COG2-CDG	Brain, liver	COG component 2	Type 2	
COG4-CDG	Brain, face	COG component 4	Type 2	

COG5-CDG	Brain, hearing system, vision, liver, bladder	COG component 5	Type 2	
COG6-CDG	Brain, gastrointestinal system including liver, immune system	COG component 6	Type 2	
COG7-CDG	Brain, skeleton, skin, gastrointestinal system including liver, heart	COG component 7	Type 2	
COG8-CDG	Brain, eyes, peripheral nervous system	COG component 8	Type 2	
Defects in the v-ATPase complex				
ATP6V0A2-CDG (autosomal recessive cutis laxa type II; wrinkly skin syndrome)	Skin (cutis laxa becoming less obvious with age), brain (mental development mostly normal), eyes, neuromuscular system, skeleton	V0 subunit A2 of V-ATPase	Type 2	
Other defects				
TMEM165-CDG	Brain, skeleton (particularly cartilage), joints, heart, liver, kidneys	Transmembrane protein 165	Type 2	
VPS13B-CDG* (Cohen syndrome)	Brain, eyes (chorioretinal dystrophy with myopia), joints, immune system (neutropenia), fat tissue	Vacuolar protein sorting-associated protein 13B		

Table 2.4 Defects localized in the ERGIC

NAME	CLINICALLY AFFECTED ORGANS AND TISSUES	DEFECTIVE PROTEIN	TF-IEF	MOI
Defects in multiple and other glycosylation pathways				
Defects in COPII				
SEC23B-CDG (congenital dyserythropoietic anemia type II)	Red cell lineage (secondary involvement of heart, liver, beta cells)	COPII component SEC23B		
Other defects				
CCDC115-CDG*	Liver, spleen, brain	Coiled-coil domain containing 115	Type 2	
TMEM199 -CDG*	Liver	Transmembrane protein 199	Type 2	

Table 2.5 Defects localized at the plasma membrane

NAME	CLINICALLY AFFECTED ORGANS AND TISSUES	DEFECTIVE PROTEIN	TF-IEF	MOI
Defects in multiple and other glycosylation pathways				
SLC39A8-CDG*	Brain, skeleton, immune system	Manganese and zinc transporter	Type 2	

Table 2.6 Defects localized at the sarcolemma membrane

NAME	CLINICALLY AFFECTED ORGANS AND TISSUES	DEFECTIVE PROTEIN	TF-IEF	MOI
Defects in O-mannosylglycan synthesis				
B3GALNT2-CDG	Brain, eyes, skeletal muscles	B-1,3-N-acetylgalactosaminyltransferase 2		
FKTN-CDG*	Brain, eyes, skeletal muscles	Ribitol-5-phosphate transferase		
FKRP-CDG*	Brain, eyes, skeletal muscles	Ribitol-5-phosphate transferase		
ISPD-CDG*	Brain, eyes, skeletal muscles	Isoprenoid synthase domain-containing protein (CDP-ribitol synthase)		
LARGE-CDG	Brain, eyes, skeletal muscles	Acetylglucosaminyltransferase-like protein		
POMGNT1-CDG (muscle-eye-brain disease, isolated RP)	Brain, eyes, skeletal muscles	Protein O-mannose β-1,2-N-acetylglucosaminyltransferase 1		
POMT1-CDG (cerebro-ocular dysplasia-muscular dystrophy syndrome)	Brain, eyes, skeletal muscles, heart	Protein O-mannosyltransferase 1		
POMT2-CDG (cerebro-ocular dysplasia-muscular dystrophy syndrome)	Brain, eyes, skeletal muscles	Protein O-mannosyltransferase 2		
TMEM5-CDG*	Brain, eyes, skeletal muscles, gonades	O-mannosylation β-1,4-xylosyltransferase		

The pdf file has been corrected. We apologize for this error.