

CDG – an update

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As of 2011, we know more than 3000 inherited diseases. Among these genetic entities, congenital disorders of glycosylation is one of the most rapidly growing groups, with actually about 50 subtypes known, positioning CDG in the focus of attention of both clinicians and basic scientists (Table 1).

In this special issue on CDG, we provide an overview on CDG, on the diagnostic approach to CDG, and on diagnostic techniques. We also offer a novel tool for disease rating and progression, discuss animal models, and try to bring more insight in the genetic and biochemical background of some disorders of N-linked glycosylation. We provide an overview of metabolic cutis laxa syndromes, several of which are due to Golgi defects, novel case reports on COG defects, and a review on the genetic and clinical features of exostosis multiplex.

Although the current issue is devoted mostly to CDG-I defects, we do expect many new genes and gene defects

to be discovered in the CDG-II patient group particularly in the secretory pathway and the complex Golgi trafficking process.

We hope that this issue will stimulate further collaboration between clinicians, biochemists, geneticists and other specialists involved in this fascinating area of metabolic diseases, full of surprises. Many young scientists have been inspired by the scientific and clinical discoveries of Professor Jaak Jaeken. A unique feature of our issue is a special contribution article written by different glycobiologists and friends of Jaak, as a tribute to his work.

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Table 1 Congenital disorders of glycosylation: disorders according to the new nomenclature (previous nomenclature in brackets)

Defects of protein N-glycosylation	Defects of protein O-glycosylation	Defects of glycosphingolipid and GPI-anchor glycosylation	Defects of multiple glycosylation and other pathways
PMM2-CDG (CDG-Ia)	EXT1/EXT2-CDG	SIAT9-CDG	DPM1-CDG (CDG-Ie)
MPI-CDG (CDG-Ib)	B4GALT7-CDG	PIGM-CDG	DPM3-CDG (CDG-Io)
ALG6-CDG (CDG-Ic)	GALNT3-CDG	PIGV-CDG	MPDU1-CDG (CDG-If)
ALG3-CDG (CDG-Id)	SLC35D1-CDG		GNE-CDG
ALG12-CDG (CDG-Ig)	POMGNT1-CDG		B4GALT1-CDG (CDG-IIId)
ALG8-CDG (CDG-Ih)	SCDO3-CDG		SLC35A1-CDG (CDG-IIIf)
ALG2-CDG (CDG-Ii)	B3GALTL-CDG		SLC35C1-CDG (CDG-IIe)
DPAGT1-CDG (CDG-Ij)			DK1-CDG (CDG-Im)
ALG1-CDG (CDG-Ik)			SRD5A3-CDG (CDG-Iq)
ALG9-CDG (CDG-II)			COG-CDG (COG1, COG4, COG5, COG6, COG7, COG8)
RFT1-CDG (CDG-In)			ATP6V0A2-CDG
ALG11-CDG (CDG-Ip)			SEC23B-CDG
TUSC3-CDG			
MAGT1-CDG			
MGAT2-CDG (CDG-IIa)			
GCS1-CDG (CDG-IIb)			