What is new in CDG?

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Abstract

Congenital disorders of glycosylation (CDG) are one group among the disorders of glycosylation. The latter comprise defects associated with hypoglycosylation but also defects with hyperglycosylation. Genetic diseases with hypoglycosylation can be divided in primary congenital disorders of glycosylation (CDG) and in genetic diseases causing secondary hypoglycosylation. This review covers the human CDG highlights from the last 3 years (2014-2016) following a summary of the actual status of CDG. It expands on 23 novel CDG namely defects in SLC39A8, CAD, NANS, PGM3, SSR4, POGLUT1, NUS1, GANAB, PIGY, PIGW, PIGC, PIGG, PGAP1, PGAP3, VPS13B, CCDC115, TMEM199, ATP6AP1, ATP6V1A, ATP6V1E1, TRAPPC11, XYLT1 and XYLT2. Besides, it discusses novel phenotypes of known CDG (DHDDS-CDG, ALG9-CDG, EXT2-CDG, PIGA-CDG, PIGN-CDG), the elucidation of putative glycosyltransferase disorders as O-mannosylglycan synthesis disorders (TMEM5-CDG, ISPD-CDG, FKTN-CDG, FKRP-CDG), a novel CDG mechanism, advances in diagnosis, pathogenesis, treatment and finally an updated list of the 104 known CDG.

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