Pediatric Neurology Part III: Chapter 179. Congenital disorders of glycosylation (Handbook of Clinical Neurology)

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Congenital disorders of glycosylation (CDG) are genetic diseases due to defects in the synthesis or the attachment of the glycan moiety of glycoproteins and glycolipids. They can be divided into four groups: disorders of protein N-glycosylation, disorders of protein O-glycosylation, disorders of lipid glycosylation, and disorders of other glycosylation pathways and of multiple glycosylation pathways. Of the more than 40 reported CDG, some 80% are neurological or have an important neurological component. By far the most common neurological CDG is phosphomannomutase 2 deficiency. Isoelectrofocusing of serum transferrin, the most widely used screening test, picks up only CDG associated with sialic acid deficiency of N-linked glycans. Predominant neurological signs and symptoms are psychomotor retardation, epilepsy, hypotonia, hyporeflexia, strabismus, retinitis pigmentosa, polyneuropathy, myopathy, and cerebellar hypotrophy/hypoplasia. All known neurological CDG have an autosomal recessive inheritance except for IAP-CDG, an X-linked pure mental retardation syndrome. No curative or effective treatment is available for neurological CDG. Since at least 1% of the genome is involved in glycosylation, it is likely that the large majority of CDG is yet to be discovered. In 2008, a novel nomenclature was introduced using the gene symbol followed by -CDG, e.g., CDG-Ia becomes PMM2-CDG. CDG should be looked for in any unexplained neurological syndrome.

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