Two argentinean siblings with CDG-IX: A novel type of congenital disorder of glycosylation?

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Resumen

Congenital disorders of glycosylation (CDG) aregenetic diseases caused by abnormal protein and lipidglycosylation. In this chapter, we report the clinical, biochemical,and molecular findings in two siblings with anunidentified CDG (CDG-Ix). They are the first and thethird child of healthy consanguineous Argentinean parents.Patient 1 is now a 11-year-old girl, and patient 2 died at theage of 4 months. Their clinical picture involved liver dysfunctionin the neonatal period, psychomotor retardation,microcephaly, seizures, axial hypotonia, feeding difficulties,and hepatomegaly. Patient 1 also developed strabismusand cataract. They showed a type 1 pattern of serum sialotransferrin.Enzymatic analysis for phosphomannomutaseand phosphomannoseisomerase in leukocytes and fibroblastsexcluded PMM2-CDG and MPI-CDG. Lipid-linkedoligosaccharide (LLO) analysis showed a normal profile.Therefore, this result could point to a deficiency inthe dolichol metabolism. In this context, ALG8-CDG,DPAGT1-CDG, and SRD5A3-CDG were analyzed and nodefects were identified. In conclusion, we could not identifythe genetic deficiency in these patients yet. Further studiesare underway to identify the basic defect in them, takinginto account the new CDG types that have been recentlydescribed.

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