P-031 - THREE CASES OF PMM2-CDG: CLINICAL, BIOCHEMICAL, NEURORADIOLOGICAL AND MUTATIONS, DESCRIPTION.

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INTRODUCTION: PMM2-CDG (MIM#212065) or CDG-Ia is the most common type of congenital disorders of glycosylation (CDG), caused by mutations in PMM2 gene. Is an autosomal recessive condition. Estimated incidence of up to 1/20,000. Major clinical features of disease include neurologic involvement, including cerebellar hypotrophy, hypotonia, strabismus, epilepsy, failure to thrive, feeding difficulties and developmental delay. Physical exam can be notable for abnormal fat distribution with suprapubic or buttock fat pads and nipple inversion. Infections and fevers may trigger stroke-like episodes. Isoelectric focusing (IEF) of serum transferrin is the standard test for diagnosing CDG due to N-glycosylation. PMM2-CDG shows type I CDG pattern. The confirmatory diagnosis requires molecular study


CONCLUSIONS: The clinical phenotype of PMM2-CDG is very suggestive. It is important to be aware when a patient with development delay is been studied. At present there is no specific treatment but the diagnosis is important because the genetic counseling of the family.