P-029 - WEST SYNDROME BY VARIANT IN ALG13 GENE ASSOCIATED WITH CDGIS WITH NORMAL PATTERN OF GLYCOSYLATED SERUM TRANSFERRIN ISOFORMS.

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\textbf{INTRODUCTION:} Congenital Disorders of Glycosylation (CDG) are new and rapidly expanding neurometabolic disorders with multisystem involvement, broad phenotypic manifestations, and variable severity. The majority results from a defect of one of the steps involved with protein or lipid N-glycosylation pathway. Mutations in the ALG13 gene, which is located on the X-chromosome, are associated with CDG type Is and have been reported in 10 patients approximately until now. \textbf{OBJECTIVE:} we report a female patient with a rare metabolic disease: CDG type Is. We discuss clinical, biochemical and molecular findings. 

\textbf{METHODS:} retrospective medical record review of a patient diagnosed with CDG Is.

\textbf{RESULTS:} this case is about a female patient, 5yo, second child of non-consanguineous parents. She had poor visual contact, developmental delay, and spasms in flexion from the first months of life. Epilepsy was refractory to multiple drugs including ACTH. Seizures frequency decreased with ketogenic diet. Cerebral MRI showed hypoplasia of the corpus callosum and increased subarachnoid spaces. Metabolic screening including glycosylation studies by transferrin isoelectric focusing resulted normal. The ratio of the CSF glucose/plasma glucose was 0.35 (normal 0.65±0.1) so molecular analysis was performed for SLC2A1 gene resulting normal. She underwent exome sequencing and was diagnosed with a heterozygous pathogenic variant in ALG13 gene related to patient´s clinical phenotype.

\textbf{DISCUSSION:} The history and clinical findings identified are typical of CDGIs. The majority of patients with CDG type Is have epileptic encephalopathy, especially West syndrome. The CSF glucose/plasma glucose ratio has not previously been systematically studied and favorable response to the ketogenic diet has not been described in epileptic encephalopathy due to mutation in the ALG13 gene. Since glycosylation studies are normal in patients with this mutation, the diagnosis of CDGIs can be missed if genetic studies are not performed.