O-012 - OUR EXPERIENCE WITH GLYCOSYLATION DISORDERS AFTER STARTING WITH SCREENING AND MOLECULAR STUDIES AT THE NATIONAL NEWBORN SCREENING LABORATORY

Audicio P [1], Méndez S [1], González G [2], Cerisola A [2], Queijo C [1], Lemes A [1]

(1) Laboratorio de Pesquisa Neonatal. Instituto de la Seguridad Social-B.P.S. (2) Catedra de Neuropediatria. Universidad de La Republica. Montevideo. Uruguay. lemesa@adinet.com.uy

INTRODUCTION: Congenital disorders of glycosylation (CDG) are a group of genetic diseases caused by hypoglycosylation of proteins and lipids. Nearly 70 inborn errors of metabolism have been described due to congenital defects of glycosylation, present as clinical syndromes, affecting multiple systems and impacting nearly every organ. Symptoms of CDG are widely variable, but some are common for several CDG types, such as psychomotor retardation, failure to thrive, coagulopathies, dysmorphic features, seizures and stroke-like episodes. Isoelectrofocusing of transferrin (TIEF) is the screening test for N-glycosylation defects. Based on the TIEF pattern, protein N-glycosylation disorders have been subdivide into two groups: CDG-I caused by defects in the assembly of glycans and their attachment to proteins; and CDG-II caused by defect in the processing of the glycans. The most frequent CDG type, over 85% of cases, is PMM2-CDG. Confirmation of clinical and biochemical diagnosis of CDG-I relies on enzymatic studies followed by molecular analysis to detect underlying gene defects. We think that in our country, CDGs remain under or misdiagnosed. ***OBJECTIVE:*** Present our experience with CDG diagnosis after we started with TIEF at the NBS laboratory. ***MATERIALS AND METHODS:*** On November 2017, TIEF assay was started in our NBS laboratory and the neuropediatric and pediatric community was informed. Our laboratory has molecular area, so, sequencing of PMM2 gene of patients with CDG type I pattern were performed. ***RESULTS:*** Since November 2017 to December 2018 there have been performed 49 TIEF studies, 4 CDG type I patterns has been identified and 2 CDG Type II. PMM2-CDG were confirm in 3 of the 4 patients with CDG type I pattern with known pathogenic variant in both alleles. ***CONCLUSIONS:*** Congenital disorders of glycosylation is part of a wide group of inborn errors of metabolism. It is a challenge to confirm the diagnosis but it is very important due to it frequency and the possibility to give genetic counseling to the family. In Uruguay, the awareness of pediatric and neuropediatric community and the introduction of TIEF and molecular study of PMM2 gene, have allowed to identify and confirm the diagnosis 3 patients in a short time.