P-027 - ISOELECTRIC FOCUS IN OF SERUM TRANSFERRIN FOR INVESTIGATION OF CONGENITAL DISEASE OF GLYCOSILATION: ANALYSIS OF THE EXPERIENCE OF TEN YEARS OF A BRAZILIAN REFERENCE CENTER

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INTRODUCTION: Congenital disorders of glycosylation (CDG) are genetic diseases of the glycoprotein, glycan and glycolipid metabolism, most of which are autosomal recessive. Most CDGs occur by defects in N-glycosylation or O-glycosylation pathways. Currently, there are over 100 CDGs reported. The clinical aspects range from prenatal symptoms (hydrops or ascites) to congenital malformations and neuropsychomotor developmental delay. The screening of CDGs involving N-glycosylation is most commonly isoelectric focusing (IEF) of serum transferring. In most cases, confirmation of diagnosis of CDGs involves genetic tests that are still difficult to access through the Unified Health System in Brazil. OBJECTIVES: To characterize the cases screened for CDG by IEF in the Inborn Errors of Metabolism Laboratory (IEML) of the Medical Genetics Service at Hospital de Clínicas de Porto Alegre, Brazil. METHODOLOGY: Observational, cross-sectional, retrospective study based on the review of laboratory records of patients investigated by the IEML from 2008-2017. RESULTS: From 2008 to 2017, 1546 patients were screened by IEF; of these, 51 (3%; mean age = 24 months, IQ 25-75 = 11-57; male = 27) presented an altered pattern of IEF (median = 5 ± 2.8 cases / year). It was possible to obtain data on the diagnosis conclusion for 14 patients. Ten of them were false-positives (classical galactosemia = 4; hereditary fructose intolerance = 4; peroxisomal diseases = 2). The others were diagnosed with PMM2-CDG (n = 2), MPDU1-CDG (n = 1), and SLC35A2-CDG (n = 1). When cases with altered pattern were compared to those with normal patterns, we observed a higher prevalence of altered cases occurred when the investigation was performed in the age group of 11mo-3y. The occurrence of inverted nipples and hepatopathy increases the probability of occurring alterations in IEF by 11x, 4.6x, respectively. CONCLUSIONS: Our data suggest that investigation of cases with suspicion of CDG is complex, and it is exacerbated by the occurrence of false positives in IEF and lack of access to diagnostic tests. The presence of inverted nipples and hepatopathy should suggest the need for investigation using IEF.