P-226 - GLUCOSE-6-PHOSPHATE DEHYDROGENASE DEFICIENCY AND NEWBORN SCREENING – 6 YEARS EXPERIENCE IN THE FEDERAL DISTRICT PUBLIC HEALTH SYSTEM

Thomas JV, Almeida TLL, Silvestre ACO, Medeiros LCA, Toledo LBB, Araújo VGB, Santos J, Gameleira KPD, Adjuto GMAF

Unidade de Genética - Hospital de Apoio de Brasília. Distrito Federal – Brasil. triagem.ce.df@gmail.com

INTRODUCTION: The Glucose-6-Phosphate Dehydrogenase (G6PD) deficiency is a widespread inherited enzyme deficiency that causes neonatal hyperbilirubinemia and hemolytic anemia. The early diagnosis allows parents orientation and prevents negatives outcomes. In Brazil, the Newborn Screening Nacional Program offers to the whole country a six diseases newborn screening test which does not include the G6PD deficiency. The Federal District, Brazil’s capital, has its own laws and provides to the population a newborn screening test for 30 diseases including the G6PD deficiency. OBJECTIVE: Presents the Federal District’s experience in 6 years of newborn screening for G6PD deficiency. METHODS: Data were collected from the software that runs the neonatal screening laboratory. RESULTS: From January 2012 to December 2018, 309.346 newborns were screened by fluorimetry (normal reference range: > 22 U/dl Hb). All the positive samples were confirmed by a second one, until the 30th day of life. The disease’s incidence is 3% of the population. There is also a whole blood third sample, measured by UV kinetic method (normal reference range: > 6,7 U/g Hb) at the age of 2 years old to assure the diagnosis. The diagnosis was confirmed in 96,5 % of these samples. All the parents receive the proper orientations at an educational lecture when they are informed about the clinical characteristics of the disease and learn to avoid the anemia hemolytic triggers. The patients follow up is carried out by the Primary Care where the physicians were trained to identify the signals and symptoms of the hemolytic anemia. If one of them goes to hemolytic anemia, the physicians from Primary Care refer them to the pediatric hematologist. CONCLUSION: As long as G6PD deficiency is a very common disease, it would be a great challenge to provide individual assistance to each committed child in a public health system with limited resources. But the early diagnosis, the parent's group orientations, the primary care asymptomatic patients follow up and the pediatric hematologist support, enabled the provision of adequate and qualified care to these children.