P-007 - REPORT OF TWO CASES OF MUTATION IN THE PHOSPHORYLASE KINASE (PHKA2) GENE IN CARTAGENA DE INDIAS, COLOMBIA

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**INTRODUCTION:** The glycogen storage disease (GSD) type IX is a rare disease of variable clinical severity that mainly affects the liver tissue. Individuals with hepatic phosphorylase b-kinase (PHK) deficiency due to mutation in the PHKA2 gene (GSD IXa) may present hepatomegaly with elevated serum transaminases, ketotic hypoglycemia, hyperlipidemia, and poor growth with considerable variation in clinical severity. **OBJECTIVE:** to identify and describe the different clinical manifestations of two brothers with the same type of mutation in the PHKA2 gene (GSD IXa), which represent the first two cases described in Colombia. **MATERIALS AND METHODS:** Two children were studied, from the city of Cartagena, who according to the symptoms, findings to the physical, clinical and biochemical examinations performed in the Biochemistry Laboratory of the University of Cartagena, had high suspicion that they had some glycogenosis. In addition, genetic studies were carried out at the Center for the Diagnosis of Molecular Diseases (CEDEM) in Madrid-Spain, through massive sequencing analysis, bioinformatic analysis, bioinformatic analysis of mutations and confirmation by sequencing of Sanger. **RESULTS:** we present the cases of two siblings, who according to the symptomatology, clinical, biochemical and genetic tests carried out, get to diagnose and confirm that they suffer from GDSIXa with mutation c.919-2A> G in hemicigosis of the PHKA2 gene, the which represents a new variant of this gene. **CONCLUSIONS:** the development of the so-called next generation sequencing technologies (NGS), such as those used in this study, is currently the method of choice to confirm the diagnosis of GSD, avoiding the use of a test invasive as the liver biopsy.