P-011 - GLYCOGEN STORAGE DISEASE TYPE XI WITH AN UNREPORTED VARIANT IN SLC2A2: CASE REPORT

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INTRODUCTION: Glycogen storage disease type XI or Fanconi-Bickel Syndrome, is a rare disease whose incidence is unknown; due to a pathogenic variant in SLC2A2 (3q26.2-q27) with an autosomal recessive inheritance pattern, finding a deficiency in GLUT2 transporters. A wide spectrum of pathogenic variants in SLC2A2 has been reported, however, they have not been associated with any specific population. It is characterized by hepatorenal glycogen accumulation, proximal renal tubular dysfunction and impaired glucose and galactose usage. Clinical manifestations appear from the first months of life, including growth retardation, polyuria, rickets, and subsequently hepatosplenomegaly. We present a case of Glycogen storage type XI with a compound heterozygous variant of SLC2A2.

CASE REPORT: Male patient, known by the Genetics Department since 2 years old; product of the second pregnancy of non-consanguineous parents. Important family history: paternal aunt with breast cancer, two uncles and paternal grandfather with Diabetes Mellitus. Full-term pregnancy ended via normal vaginal delivery; weight and height suitable for gestational age. In the first year of life, growth retardation and development delay were identified, as chronic gastroenteritis, prominent abdomen due to hepatomegaly, hypoglycemia and recurrent infections. At physical examination the patient was found hypotonic, dolichocephalic, with prominent forehead, oval face, prominent cheeks, “doll face”, long eyelashes and hepatomegaly. Neonatal screening results, sweat electrolyte tests, and brain TC Scan, within normal parameters. Sequencing analysis of G6PC, does not report pathogenic variants. At 3 years old, hypokalemia and hypocalcemia was present, as well as metabolic acidosis. With the suspicion of Fanconi syndrome, sequencing of the SLC2A2 gene in peripheral blood sample was requested.

RESULTS: Two heterozygous variants were identified: c.609_612delTCAG (p.S203Rfs*47) in exon 5, probably a pathogenic variant not reported previously. The second identified variant is c.1250C> T (p.P417L) in exon 10, described in 1999 as a missense variant in homozygous state.

CONCLUSIONS: This work presents a case of Glycogen storage type XI, in heterozygous compound state, whose probably pathogenic unreported variant (p.S203Rfs * 47) is known to generate a reading frame shift and a stop codon, translating a truncated SLC2A2 protein.