P-005 - CLINICAL PRESENTATION OF GLYCOGEN STORAGE DISEASE IN A REFERENCE CENTER IN SÃO PAULO, BRAZIL.

Gomes JVB, Curiati MA, Oliveira RB, Frangipani BJ, Mendes CSC, Rand MH, Martins AM

Reference Center in Inborn Errors of Metabolism, Federal University of São Paulo. São Paulo – Brazil. gomes.jvb@gmail.com

INTRODUCTION: Glycogen Storage Diseases (GSD) are inherited metabolic disorders related to the inadequate functioning of enzymes or transporters involved in glycogen synthesis and degradation. The overall incidence is estimated at 1: 20,000 – 43,000 live births. GSDs can be divided into those with hepatic impairment, which manifests as hypoglycemia and fasting intolerance, and those with neuromuscular involvement. Some types may exhibit both manifestations. The severity of the disease ranges from mild disorders with normal lifespan to fatal cases if untreated. The treatment is based on specific diet aimed at euglycemia and prevention of metabolic disorders and complications of the disease.

OBJECTIVES: To describe the first clinical and laboratory features of patients with GSD who are followed at the Reference Center in Inborn Errors of Metabolism.

MATERIALS AND METHODS: Retrospective evaluation of physical medical records.

RESULTS: Among the 28 patients diagnosed with GSD, 12 (43%) were classified as type I and 11 (39%) as type III. Eighteen (64%) were male and 11 (39%) were children of consanguineous marriage. The median age at onset of symptoms, diagnosis, first consultation and initiation of treatment was 3 months, 19 months, 28 months and 32 months, respectively. Clinical features that led to the diagnosis were hepatomegaly (n=27), failure to thrive (n=16), neuropsychomotor development delay (n=10) and seizures (n=9). The first laboratory tests showed hypoglycaemia (n=20; mean = 41 mg/dL), hypercholesterolemia (n=22; mean = 245 mg/dL), hypertriglyceridemia (n=26; mean = 537 mg/dL), increased aspartate aminotransferase (n=24; mean = 221 U/L), increased alanine aminotransferase (n=22; mean = 129 U/L), hyperlactatemia (n=18; mean = 61 mg/dL) and metabolic acidosis (n=14). Hepatic biopsy was performed in 46% of patients and 11% underwent molecular testing. Nowadays, the median age of the patients is 19 years old and only one of them has died.

CONCLUSIONS: GSD type I is the most prevalent in our service, just as hepatomegaly is the most common manifestation. The knowledge of the clinical and laboratory characteristics of GSD’s patients is fundamental for the pediatrician. Early diagnosis, establishment of specific diet and periodic monitoring are essential to prevent complications of the disease.