P-021 - A CASE SERIES OF FATTY ACID OXIDATION DISORDERS FROM A TERTIARY TEACHING HOSPITAL IN SOUTHERN BRAZIL

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INTRODUCTION: Mitochondrial fatty acid β-oxidation disorders (FAOD) are a heterogeneous group of defects in fatty acid transport and mitochondrial β-oxidation, affecting energy homeostasis with multi-system involvement, including life-threatening manifestations. FAOD are inherited as autosomal recessive disorders and have a variable presentation, with either neonatal or later onset.

OBJECTIVE: To characterize phenotypically a series of patients with FAOD managed at an outpatient clinic in a tertiary teaching Hospital of Southern Brazil.

METHODS: Retrospective observational case series study including eight patients with clinical, biochemical and/or molecular diagnosis of FAOD. RESULTS: Five female and three male individuals were evaluated, with mean age 11.07 years (three months to 39 years) and mean age of clinical presentation 22.12 months (18 hours to 14 years). The diagnosis were Long chain acyl-coA dehydrogenase deficiency (n=2); Multiple acyl-coA dehydrogenase deficiency (n=2); Carnitine palmitoyltransferase II deficiency (n=2); Carnitine palmitoyltransferase I deficiency (n=1); and Very long chain acyl-coA dehydrogenase deficiency (n=1). Mean age at diagnosis was 8 months (20 days to 36 years). Diagnosis was molecularly confirmed in five patients. Hypoglycemia was the initial symptom for 87.5% (n=7) patients. Other observed signs and symptoms included hypotonia (6/8); hepatomegaly (3/8); seizures (3/8); developmental delay (2/8); feeding problems (1/8); dehydration (1/8); intellectual disability (1/8); unspecific dysmorphisms (1/8); cardiomyopathy and arrhythmia (1/8). One patient diagnosed at age 36 presented lower limbs weakness and pain, rhabdomyolysis, hypoglycemia and deep vein thrombosis. Three patients had a sibling with previous history of unexplained death and one had consanguineous parents. CONCLUSION AND DISCUSSION: This descriptive study emphasizes the multisystemic, severe and life-threatening character of FAOD, requiring integrated and multidisciplinary management. Although this is the most prevalent disorder in other series, no case of Medium-chain acyl-CoA dehydrogenase (MCAD) deficiency was observed. Newborn screening for such diseases is not widely available, leading to late access to reference centers. Greater awareness of this disorder among assistants should aid their search for FAOD in cases of hypoglycemia, hypotonia, hepatic and cardiac manifestations that might otherwise be improperly managed. Larger studies should be done to better understand those symptoms in Brazilian patients, as well as unified ones on a national basis.