P-180 - OTC DEFICIENCY. DESCRIPTION OF 7 PATIENTS OF ONE CENTER IN MONTEVIDEO.

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INTRODUCTION: Ornithine transcarbamylase (OTC) is the urea cycle enzyme that catalyzes the formation of citrulline. The OTC gene is located on the X chromosome. OTC deficiency, is the most frequent hereditary defect of ureagenesis. The estimated incidence varies in different countries. Hemizygotes with complete OTC deficiency, present acute hyperammonemia in the first week of life, while that with partial deficiency have a late presentation. The severity of OTC symptoms in women ranges from asymptomatic to almost as severe as in affected males. The clinical manifestations are caused by the toxic effects of hyperammonemia and glutamine in brain. Male and female may present lethargy, vomiting, behavioral abnormalities, cerebral edema, coma, and death. Biochemical abnormalities include increased plasma ammonia, glutamine and decrease citrulline, and increased urinary orotic acid. Carrier identification is important because genetic counseling. Chronic treatment includes ammonia scavengers in combination with limited protein diet and arginine or citrulline supplementation.

OBJECTIVE: To present our patients with OTC deficiency. MATERIALS AND METHODS: We perform description of clinical, biochemical and molecular findings of patients with OTC deficiency followed in our center between January 2008 and December 2018. RESULTS: The clinical phenotypes were: 1 hemizygotes with late-onset disease, 5 symptomatic and one asymptomatic heterozygous. The age of onset was below 6 years the heterozygous and at 18 months the male. All symptomatic patients had had more than one admission before the diagnosis. Clinical manifestations varied between cases, with one or more of the following signs and symptoms: drowsiness, lethargy, encephalopathy, seizures, vomiting, behavior alterations, and irritability. Biochemical findings: elevated plasma ammonia and glutamine, low citrulline and elevated urinary orotic acid. Genetic analysis, showed 7 different mutations Regarding disease progression after start treatment, one adolescent female required admission because diet transgression several times, one was pregnant without symptoms during pregnancy or at or after delivery, the male had one symptomatic hyperammoniemic episode after start treatment but admission was not required. CONCLUSIONS: Suspicion of OTC, early confirmatory diagnosis, and prompt initiation of treatment are essential to avoid new hyperammonemic episode that could cause irreversible neurological damage and or death and for the adequate family genetic counseling.