

P-020 - CLINICAL, TREATMENT AND OUTCOME FEATURES IN 7 ARGENTINEAN PATIENTS WITH VERY LONG CHAIN ACYL COA DEHYDROGENASE DEFICIENCY.

Armani GR, Garcia J, Eiroa HD

Hospital Juan Pedro Garrahan. Buenos Aires - Argentina.

INTRODUCTION: Very long chain acyl CoA dehydrogenase (VLCAD) deficiency (OMIM#201475) is an autosomal recessive disorder of fatty acid oxidation, and the second most common disorder of fatty acid oxidation all over the world. Despite VLCAD deficiency is included in many newborn screening (NBS) programs worldwide, Argentina does not include it in its NBS and therefore does not have a true incidence of this disorder. **OBJECTIVE:** The aim of this study is to analyze the clinical, biochemical, treatment and outcome features of 7 patients in Argentina, which is the first and largest existing description of Argentinean patients so far. **METHODS:** We conducted a retrospective review of all health records of patients with VLCAD deficiency diagnosed and treated at our center, since the beginning of our metabolic service in 2008. **RESULTS:** Severe early onset is the most frequent phenotype in our cohort, 71% of patients had a variable degree of hypertrophic cardiomyopathy which recovered after treatment, 71% presented hypotonia and 57% hepatomegaly at diagnosis. Family history of sudden death was positive in 28% of patients and only one of our patients died before diagnosis. In accordance with the WHO's body mass index, 4 of 6 remaining children present obesity as a complication of diet and low physical activity and 1 has overweight. Based on a suggested score of severity proposed by Dr Diekman we could not find a correlation between the highest scores and the severity of the disease (understanding "severity" as the number of admissions even if the patient was following a treatment). One particular patient has a severe disease evolution with 22 hospitalizations of which 17 were without a trigger. All, except one patient, were under a high MCT dietary treatment plus cornstarch. **CONCLUSION:** This study reports the clinical, treatment and outcome features of patients with VLCAD deficiency over the last 10 years. The paucity of the current published data about Argentinian patients makes it very difficult to estimate the incidence of VLCAD in the country. This is further aggravated by the lack of NBS and by the lack of knowledge of this disease by medical community in general.