P-120 - MAPLE SYRUP URINE DISEASE IN CHILE: CHARACTERISTICS OF DIAGNOSIS AND TREATMENT IN 45 CHILEAN PATIENTS IN FOLLOW-UP AT INTA, UNIVERSITY OF CHILE


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INTRODUCTION: Maple syrup urine disease (MSUD) is an autosomal recessive inherited metabolic disease caused by deficient activity of the branched-chain α-keto acid dehydrogenase (BCKD) enzymatic complex, that catalyzes the last reaction in the catabolic route of branched chain amino acids (leucine, valine and isoleucine). The disorder varies in severity and the clinical spectrum is quite broad with five recognized clinical variants. The treatment consists in dietary restriction of BCAAs and close metabolic monitoring. OBJECTIVE: Characterize MSUD Chilean patients in follow-up at INTA. METHODS: Retrospective study with 45 MSUD patients in follow-up. RESULTS: 45 patients were included, 40/45 patients showed classic presentation, only 8/45 diagnosed before the onset of clinical manifestations. The average diagnosis age was 67 ± 231 days. Debut level average of offending amino acids were: Leucine 1118 ± 579 umol/L, Valine: 550 ± 598 umol/L and Isoleucine 454± 458 umol/L. The age of patients was between 9-18 years old (22/45) and the majority of patients in follow up were females (27/45). Regarding nutritional treatment, patients received Kcals: 1515 ± 458 kcal/day, Prot special formula: 2,0 ± g/kg/day, representing 97% of the daily protein intake. All patients received supplementation: L-Valine: 277±221mg/kg/day; L-Isoleucine 281±244mg/kg/day; Tiamine: 50 mg/day; L-Carnitine: 45±15 mg/wt/d. The nutritional assessment showed a majority of eutrophic patients 29/45, overweight and obesity 12/45 and only 4/45 under weight. Children under and over 5 years old showed good metabolic follow up: Leucine level: 254± 67 and 298± 136 umol/L, respectively. Most of our patients (35/45) have some psychomotor or neurodevelopmental delay. During the follow up there were 8/45 patients who died between 10 and 17 years old, due to causes non-related with uncompensated metabolic disease. CONCLUSION: The existence of a newborn screening program is essential to allow the detection of patients in pre-clinical stage and decrease the severity of neuropsychological delay and lethality associated to MSUD. Our patients presented satisfactory adherence to nutritional treatment, laboratory and clinical follow up. Nevertheless adherence to treatment does not seem to be directly related to a good neurological outcome and multiple factors may intervene in the neurological delay that patients show.