P-170 - NUTRITIONAL MANAGEMENT AND OUTCOMES OF TYROSINEMIA TYPE I PATIENTS IN A PEDIATRIC REFERENCE CENTER IN ARGENTINA

Carmona NM, Lavorgna SL, Cresta AS, Levy L, Bindi V

Hospital de Pediatría “Juan.P. Garrahan”. Buenos Aires-Argentina

i: Hereditary tyrosinemia type I (HT1) is an autosomal recessive disorder caused by deficiency of fumarylacetoacetase (FAH), the last enzyme of tyrosine degradation. The treatment consists of inhibition of the formation of toxic metabolites by nitisinone and reduction of tyrosine levels through dietary treatment. ii: To report the dietary treatment and experience of HT1 patients in follow up at Garrahan Hospital. iii: A review of 6 patients (4 female and 2 male) with diagnosis of HT1 by classic biochemical and clinical characteristics in follow up. Plasmatic amino-acids(AA), alpha-fetoprotein, succinylacetone were monitored every 6 months as well as liver and kidney ultrasound and magnetic resonance imaging for the detection of nodules. Anthropometric measurements (weight, length, BMI) and dietary intake (3 days’ food record) were analyzed at last control data. Detailed dietary intake of phenylalanine plus tyrosine (phe+tyr) and protein content from phe+tyr free amino acid formula (AA formula) were analyzed. iv: Median age at diagnosis: 3.5 months; 2 patients were diagnosed after sibling death. Normal growth was achieved (3 overweight/obese). Actual median age: 8.9 years (1.6-16) All patients received AA formula, phe+tyr restricted diet and administration of nitisinone. Dietary intake analysis: Phe+tyr: 655 ± 274 mg/day, protein from AA formula: 1.3 ±0.3 g/kg 16% (n=1) of the patients presented normal plasmatic tyrosine levels while 83% (n=5) still had high levels at last control. Alpha-fetoprotein levels remained stable in 5 patients, except 1 who started treatment last year v: Most patients have acceptable metabolic control and up to present have not presented complications. It is difficult to maintain plasmatic tyrosine levels within normal range, proving especially challenging in adolescent period when compliance usually decreases. In order to ensure best care of these patients it is important to schedule regular appointments with them, their family and the multidisciplinary metabolic team.