P-172 - EXPERIENCE IN MANAGEMENT OF HEREDITARY TYROSINEMIA TYPE 1 IN A REFERENCE HOSPITAL OF PERU

Morales-Acosta MA¹, Dueñas-Roque MM¹, Bonilla-Suárez C¹, Protzel-Pinedo A¹

(1) Hospital Nacional Edgardo Rebagliati Martins – EsSalud, Lima, Peru.

INTRODUCTION: Tyrosinemia type 1 (HT1) is an inborn error of tyrosine catabolism caused by the defective activity of fumarylacetoacetate hydrolase with progressive liver disease, renal tubular dysfunction and peripheral neuropathy. The estimated birth incidence is 1: 100,000 worldwide. Treatment with oral nitisinone has notably improved prognosis and life expectancy. OBJECTIVES: To describe 2 cases of tyrosinemia type 1 successfully treated in Peru. MATERIALS AND METHODS: We present two cases of HT1 managed at Rebagliati Hospital of Lima, Peru. RESULTS: Case 1: A 11-month infant, first child of healthy non-consanguineous parents. Born by cesarean section due to fetal macrosomia (4334g). Appropriate psychomotor development until sixth month. Since seventh month he had recurrent hypoglycemia, hypotonia and failure to thrive. It was noticed hepatomegaly, humeral fracture, rickets, and motor delay. It was found markedly high alpha-fetoproteins (AFP) (148, 912 ng/ml), altered liver profile, increased plasma tyrosine and methionine and high urine succinylacetone (423 ug/mg creatinin). At 13th month, it was started treatment with oral nitisinone at dose 1 mg/kg, protein-restricted diet and a medical formula. It was observed improvement of growth and psychomotor development and urinary succinylacetone got normal.AFP values decreased but did not reach normal ranges. It was performed liver transplantation due advanced cirrhosis and suboptimal response to treatment. Case 2: A 10-month infant , fourth child from consanguineous parents, with positive family history of HT1 in two older sibs, one died from severe liver failure, and the other was underwent to a liver transplantation. He was born by normal delivery, weighted 3750g,had normal psychomotor development. He was referred because of hepatomegaly, AFP 185,000 ng / ml; high plasma tyrosine and elevated urine succinylacetone levels (39.7 mg/g UCr). Ultrasound reported chronic liver disease. At 11 months old, it was started oral nitisinone at dose 1mg/kg, protein-restricted diet and medical formula. Improvement of hepatomegaly and alpha-fetoprotein level was observed. Currently he continues on treatment with favorable evolution. CONCLUSIONS: This is the first experience of management of HT1 in Peru. Following an adequate treatment, patients diagnosed with HT1 who were referred to the Rebagliati Hospital of Lima Peru have been successfully recovered.