P-157 - PHENYLKETONURIA IN CHILE; ADHERENCE TO TREATMENT IN 130 PKU PATIENTS DIAGNOSED DURING THE NEWBORN PERIOD


Laboratorio de Genética y Enfermedades Metabólicas, Instituto de Nutrición y Tecnología de los Alimentos (INTA), Dr Fernando Monckeberg Barros, Universidad de Chile, Santiago-Chile. felipe.penaloza@inta.uchile.cl.

INTRODUCTION: Phenylketonuria (PKU) is caused by a deficiency in the enzyme phenylalanine hydroxylase (PAH). Treatment consists of a phenylalanine (Phe) restricted diet and an amino acid formula without Phe (AAF). OBJECTIVE: To analyze the results of the clinical follow-up of 130 PKU of neonatal diagnosis in active control. METHODOLOGY: A retrospective, cross-sectional study was performed in 130 PKU patients. Phe blood levels were analyzed according to treatment protocol from 1992 to 2007 where good metabolic control (GMC) was defined as: Phe<10 mg/dL; protocol of 2007 and 2014 which considered GMC: Phe<8 mg/dL, and the current protocol since 2014 that defines GMC: Phe< 6 mg/dL. In addition, dietary intake (Phe, calories, proteins and minerals), intellectual quotient (IQ) and nutritional status were evaluated. RESULTS: The age of diagnosis (dg) was 16.8 ± 8 days of life, Phe level at dg was 16.2 ±7.9 mg/dL. Patients are currently between 4 months to 26 years old. 91/130 PKU maintain GMC according to protocol, 30/130 maintains regular metabolic control and 9/130 has poor metabolic control. PKU patients >3 years of age (n = 68) with good metabolic control had a total IQ at 101.6 ±15.3, unlike PKU >3 years (n = 9) with poor metabolic control, who registered an IQ of 72.4 ±16.0. Phe intake was 400.15 ±275.95 mg/day, protein intake 2.3 ±0.41 g/kg/day, 89.4% of which comes from AAF. The dietary analysis found a caloric, calcium, zinc and iron intake consistent with the RDI. The nutritional status according to WHO 2002 standards, found that 61.2% were eutrophic, 18.5% were overweight, 15.5% were obese and 4.7% were at risk of malnutrition. CONCLUSIONS: Good metabolic control, long-term integral follow-up and the constant provision of the AAF are essential variables to achieve growth and development in normal ranges.