EVALUATION OF THE SERIES OF ADULTS WITH INBORN METABOLIC DISEASES FOLLOWED IN ENDOCRINOLOGY IN ANDALUSIA (SPAIN)

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OBJECTIVES

The transition of the sanitary attention from the paediatric age to the adult is a particularly vulnerable period in the patients with metabolic congenital diseases. In Andalusia two adults’ units exist in the Hospitals of reference of Seville and Malaga for the follow-up of the patients with inborn metabolic diseases. Our aim in this study was to evaluate the current series of attended patients.

MATERIAL AND METHODS

We evaluated all patients transferred to the adult units since 2008. The clinical records of pediatric cases and current records of patients were analyzed retrospectively.

RESULTS

149 adults with metabolic disorders (98 in Seville and 51 in Malaga) are evaluated. Aged between 14 and 65 years. 76 women and 73 men.

HPA benigna (1), Fructosuria (4), galactosuria (4), Glucogenosis: Ia (1), Ib (1), II (2), trimethylaminuria (2), Tyrosinemia tipo 1 (1), Tyrosinemia tipo 2 (2), Aciduria 3OH 3 metilglutaríca (2), propioníacidemia (1), metilmalonic acidemia (1), metilmalonic acidemia with homocystinuria acidemia (1), Acidemia methylglutácono (1), maple syrup urine disease (1), classical Homocystinuria (déficit CBS) (4), Déficit MTHFR (3), alcaptonuria (2), OTO deficiency (1), déficit succinyl CoA (1), beta fatty acid oxidation deficiency (CPT1): (4), carnitine transporter deficiency (4), Methylcrotonoilcínuricía: (2), Xanthomatosis cerebrotendinosus: (2), hipercarnitoniemia-Hyperinsulinism syndrome (1), adrenoleukodystrophy (2)

CONCLUSIONS

The majority pathology in our series is phenylketonuria. Although most patients come from Pediatric follow-up, patients in adulthood have been diagnosed as a result of neonatal screening.

The multidisciplinary, comprehensive, coordinated and individualized treatment is the guarantee for optimum care and quality of life in these patients.

It is a challenge for endocrinology training and knowledge of these diseases, enabling the care for these patients in specialist clinics.