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

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OBJETIVES

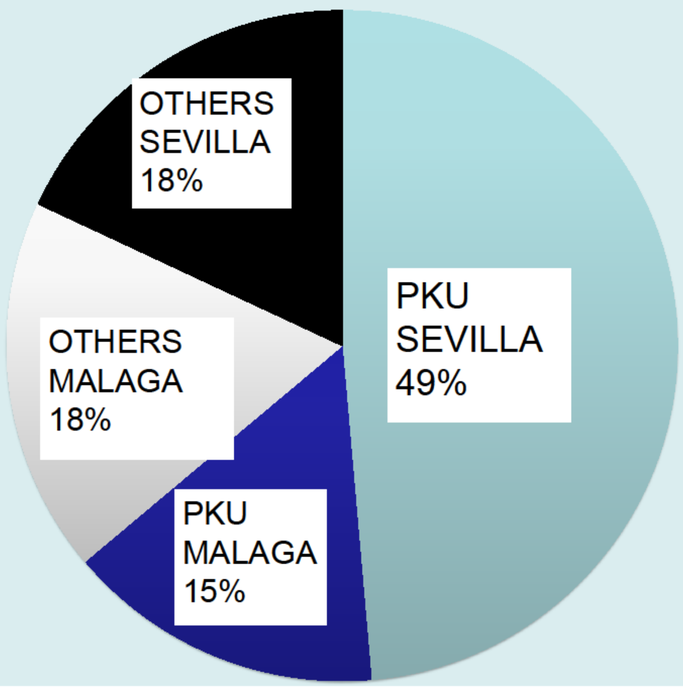
The transition of the sanitary attention from the paediatric age to the adult is a particularly vulnerable period in the patients with metabolic congenital diseases. In Andalusia two adults' units exist in the Hospitals of reference of Seville and Malaga for the follow-up of the patients with inmborn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

MATERIAL AND METHODS

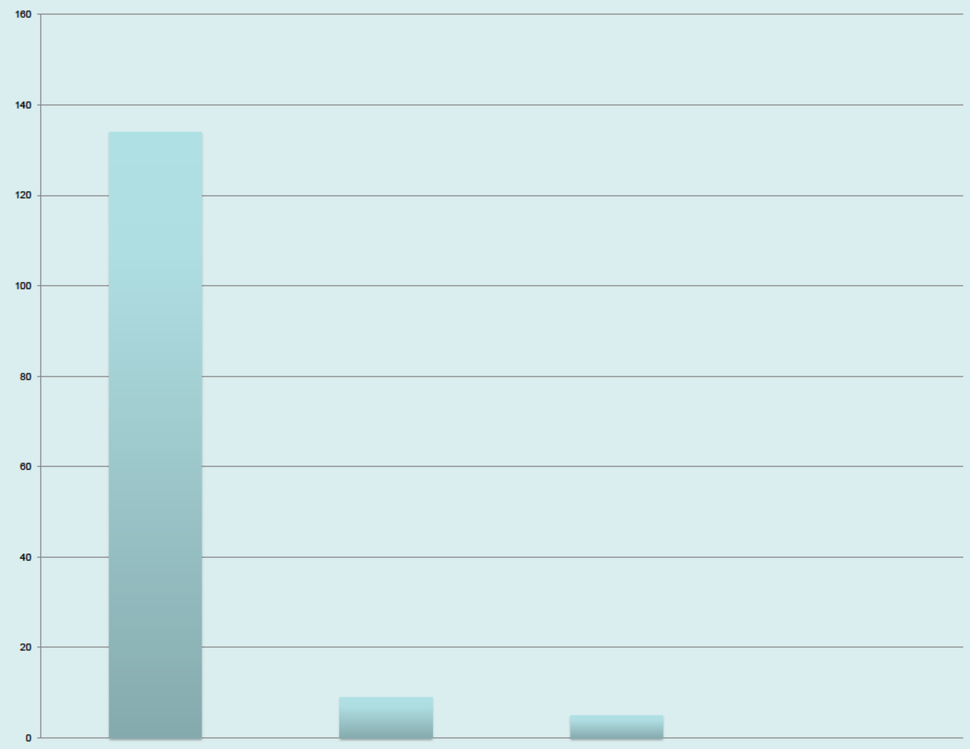
We evaluated all patients transferred to the adult units since 2008. The clinical records of pediatric cases and current records of patients were analyzed retrospectively

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), Fructosemia (4), galactosemia (4), Glucogenosis: Ia (1), Ib(1), III (2), trimethylaminuria (2), Tyrosinemia tipo 1 (1), Tyrosinemia tipo 2 (2), Aciduria 3OH 3 metilglutárica (2), propionic acidemia (1), metilmalonic acidemia (1), metilmalonic with homocystinuria acidemia (1), Acidemia methylglutaconic (1), maple syrup urine disease (1), classical Homocystinuria (déficit CBS): (4), Déficit MTHFR: (3), alcaptonuria (2), OTC deficiency (1), déficit succinyl CoA (1), beta fatty acid oxidation deficiency (CPT1): (4), carnitine transporter deficiency (4), Methylcrotonilglycinuria: (2), Xanthomatosis cerebrotendinous : (2), hiperamoniemic-Hyperinsulinism syndrome (1), adrenoleukodystrophy (2)



Transtition 135 patinets

Adult DX 9 patients*

Adult DX from neonatal screening 5 patients **

* 1 alcaptonuria 1 adrenoleukodystrophy, 2 metylmalonic acidemia (MMA): deficit CBLC and CBLA, 2 cerebrotendinous xanthomatosis, 3 deficit MTHFR

**3 carnitine transporter deficiency and 2 methylcrotonylglycinuria

17 pregnancies in patients with metabolic disorders (12 PKU, 2 homocystinuria, 2 PA ,1 MMA

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.

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