P-158 - EVOLUTION OF PKU ADULT PATIENTS DETECTED BY NEONATAL SCREENING

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INTRODUCTION: Dietary treatment is the mainstay of therapy for phenylketonuria (PKU), but adherence to low-phenylalanine diet decreases from adolescence. Newborn Screening (NBS) programs in Argentina started in the 90’s and our hospital has been a reference center for PKU. OBJECTIVES: Characterize a population of PKU patients older than 16yrs detected by NBS who were followed-up since then by the same professional’s team. Understand their nutritional habits, difficulties to follow the treatment and knowledge about PKU through a survey. Describe their treatment adherence and socio-cultural level, through medical records. MATERIALS AND METHODS: Over a 6 months period (Jul/2018-Jan/2019), 38 adult PKU patients were recruited to the survey. Patient’s inclusion criteria were as follows: age $\geq$16yrs, confirmed diagnosis of PKU from NBS and being on follow-up. A survey consisting in a set of multiple-choice questions and an open section for comments was designed and distributed by e-mail or during consultations. RESULTS: 38 PKU patients aged 16-33yrs answered the survey and were included in the study. 76% of patients did not have knowledge about their phenylalanine intake. 87% required amino acid (AA) mixture, but only 9% correctly took the supplements and 68% consumed limited quantities of low-protein food. 80% reported difficulties with treatment, mainly with adherence to the diet outside home and phenylalanine monitoring compliance; however, 58% considered they did the diet correctly. Around half of patients showed poor treatment adherence. Socio-cultural, educational level and work opportunities were similar to the average population. CONCLUSION: Despite the relatively small sample size, it reflects the outcomes of our PKU adult patients detected through NBS. Adherence to nutritional treatment was unsatisfactory, affecting all the social classes. Diet outside home and regular phenylalanine monitoring were their main difficulties. Patients did not consider the inappropriate frequency of AA mixture and low-protein food intake as a problem. A high number of patients seem to have a poor perception and knowledge about their disease. Considering these results, we assume the need to implement a simplified diet, increase the multi-media material supply to patients, work in an early transition to health self-care, and create a local system of social working groups.