

Demographic status of patients with classic and non-classic PKU since the beginning of screening (2006) in Ardabil province

Abstract

Background: Studies have been conducted to determine the characteristics of patients with phenylketonuria in the country, but such a study has not been conducted in our region, and there is no documented information about the characteristics of patients with phenylketonuria.

Aim: To evaluate the demographic status of patients with classic and non-classic PKU since the beginning of screening in Ardabil province.

Materials and methods: All phenylketonuria patients identified since 2006 in Ardabil province, which were 70 people, were included in this study. Information related to gender, age, place of residence, type of phenylketonuria, consanguineous marriage, age and weight at birth, regular follow-up of the patient, level of disease control, history of seizures, presence of behavioral disorder, presence of mental retardation, and presence of an affected sibling was determined for all patients and recorded in the checklist.

Results: Out of 70 examined patients, 41 (58.6%) were boys and 29 (41.4%) were girls. Most of the patients lived in the Ardabil (51.4%), Parsabad (18.6%), and Meshkinshahr (10%). There was mental retardation in 8 patients (11.4%), attention deficit hyperactivity disorder in 13 patients (18.6%), and a history of seizures in 5 patients (7.1%). The disease was classic in 62 people (88.6%) and non-classic in 8 people (11.4%). Patients with classic and non-classic phenylketonuria had no significant difference in terms of demographic variables ($P>0.05$).

Conclusion: The incidence of phenylketonuria disease has been higher than the national average in Ardabil province since 2005 (21.8 vs. 16.5 per 100,000 births). Consanguineous marriage was present in 60% of patients. Attention deficit hyperactivity disorder was present in almost one fifth and mental retardation in one tenth of patients. Most of the patients (88.6%) were suffering from classic phenylketonuria and the patients suffering from classic and non-classic types had no significant difference in terms of demographic variables.

Keywords: Phenylketonuria, demographic characteristics, Ardabil.