Comparison of the phenylketonuria phenotypes in Qazvin province before and after neonatal screening until 2017

Fatemeh Saffari[1], Camelia Taherkhani[2], Neda Esmailzadehha[3], Ali Homaei[4]

[1] Associate Professor of Pediatric Endocrinology, Children Growth Research Center, Qazvin University of Medical Sciences, Qazvin, Iran
[2] General Practitioner, Children Growth Research Center, Qazvin University of Medical Sciences, Qazvin, Iran;
[3] Metabolic Diseases Research Center, Qazvin University of Medical Sciences, Qazvin, Iran
[4] PhD Student of Epidemiology, School of Health, Iran University of Medical Sciences, Tehran, Iran
[5] Student of Medicine, School of Medicine, Shahid Beheshti University of Medical Sciences, Tehran, Iran

Introduction & Objectives

Phenylketonuria (PKU) is an autosomal recessive disorder that primarily affects the brain. Patients are at risk for intellectual disability, developmental disorder, hyperactivity, seizure, autism, and so on. The aim of this study was to compare the PKU phenotypes in Qazvin province, Iran before and after neonatal screening until 2017.

Methods

All children with PKU (61 patients) in Qazvin province, Iran who had been diagnosed before and after neonatal screening until 2017 were examined. Data were analyzed using descriptive statistics and Chi-square test.

Results

Of 61, 31 (50.8%) were female. Patients were among 2.5 months to 18 years old. Of 61 patients, 7 (11.5%), 33 (54.1%), and 21 (34.4%) had malignant, classic, and HPA form of PKU, respectively. 23 (37.7%) were identified in neonatal screening and 38 (62.3%) were diagnosed before screening by clinical findings. The incidence rate of PKU was one in 4858 live births in Qazvin province. The most frequent clinical manifestations were delay in motor (57.4%) and language (54.1%) development, seizure (36.1%), restless (34.4%), hyperactivity (29.5%), eczematous rash (29.5%), and severe mental retardation (8.2%). All clinical manifestations in patients identified after neonatal screening was significantly lower than patients diagnosed before screening ($P< 0.001$).

Conclusions

Intellectual disorders, developmental delay, and seizure were the most frequent phenotypes in patients with PKU in Qazvin Province. Neonatal screening is necessary to prevent brain damage in patients with PKU.

References

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