

Title: Epidemiology of phenylketonuria disease in Jordan: Medical and nutritional challenges

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Background: Phenylketonuria (PKU) is the most frequent inborn error in amino acid metabolism caused by a deficiency of the Phenylalanine Hydroxylase enzyme (PAH). If PKU is left untreated, high concentrations of Phenylalanine (Phe) accumulate in the blood, leading to severe brain dysfunction, neurodevelopmental, behavioral and psychological problems. Data concerning the epidemiology of PKU in Jordan are limited. The main objectives of our study were to determine the prevalence of PKU in Jordan, analyze the PKU phenotypes and identify major challenges in providing dietary management to PKU patients.

Methods: Data were collected utilizing the medical records of PKU patients attending the PKU clinic at the Ministry of Health in Amman, Jordan, between 2008 and 2021.

Results: The total number of patients diagnosed with PKU was 294. The prevalence of PKU was estimated to be 1/5263. Most patients were Jordanians (90.8%) and 9.2% were non-Jordanians. More than half of the patients (56%) were diagnosed through the National Newborn Screening (NBS) program. Regarding the phenotypes of PKU, 46.6% had moderate PKU, whereas 42.9% had the classic type of PKU and only 8 (2.7%) had cofactor Tetrahydrobiopterin (BH4) deficiency (atypical PKU). According to the age of diagnosis, 66% of patients were diagnosed more than 30 days post-birth. Consanguinity was found in 87.4% of patients and the majority of patients, 218 (74.2%), had first-degree consanguinity. The most common complication was mental retardation (31%). Most patients were committed to dietary management (83%) and developed fewer complications.

Conclusion: In our study, we demonstrated the importance of the NBS program in the early identification and diagnosis of new PKU cases which allows the initiation of treatment and dietary management to prevent severe complications of PKU in Jordan.

Keywords: Jordan, Dietary management, National Newborn Screening (NBS) program, Phenylketonuria (PKU).

Biography

Safwan Dababneh is affiliated to Ministry of Health, Jordan.