Heel Blood Screen Awareness: Diagnose and Prevent Phenylketonuria

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Introduction: Phenylketonuria (PKU) is the most prevalent inherited metabolic disorder affecting amino acid metabolism in Turkey. Characterized by a deficiency of the liver enzyme phenylalanine hydroxylase (PAH), deficiency of PAH leads to the accumulation of phenylalanine in body tissues , resulting in severe neurodevelopmental consequences if not diagnosed and treated early. Timely newborn screening, particularly with the heel-prick test, plays a critical role in early detection and treatment.

Case: We present a 2-month-old male infant born at home to consanguineous parents with inadequate prenatal follow-up. The patient exhibited lethargy, hypotonia, eczematous rash, and a characteristic musty body odor. Neurological examination revealed hyporeflexia. Laboratory findings showed a plasma phenylalanine level >1200 µmol/L (normal <120 µmol/L), and EEG indicated epileptiform activity. Newborn screening had not been performed. The patient was diagnosed with phenylketonuria and started on a phenylalanine-restricted diet alongside antiepileptic treatment. At 6 months, seizures were controlled, phenylalanine levels remained within the therapeutic range, but mild developmental delay was present.

Discussion: The accumulation of phenylalanine disrupts myelin formation and causes neurotoxicity, highlighting the importance of early diagnosis and dietary management. Although infants appear normal at birth, neurological deterioration progresses rapidly without treatment. This case underscores the risks associated with lack of newborn screening and the heightened incidence of PKU in populations with high consanguinity rates.

Conclusion: In addition to the heel blood test being able to diagnose phenylketonuria, it is a public health duty to perform these screenings because it also allows the diagnosis of some other newborn diseases. Raising awareness, especially in rural or underserved populations, is essential to prevent irreversible neurodevelopmental damage associated with late diagnosis.

Keywords: phenylketonuria, newborn screening, heel-prick test, consanguinity, phenylalanine hydroxylase deficiency