Late-diagnosed phenylketonuria in an eight-year-old boy with dyslexia and attention-deficit hyperactivity disorder.

Yıldız Y, Dursun A, Tokatlı A, Coşkun T, Sivri HS.

Turk J Pediatr. 2016;58(1):94-96.

Abstract

Phenylketonuria, previously a common cause of severe intellectual disability, is a metabolic disorder now promptly diagnosed and effectively treated thanks to newborn screening programs. Here, we report a male patient presenting with dyslexia and attention-deficit hyperactivity disorder, who was diagnosed with mild phenylketonuria at eight years of age. Earlier recognition and treatment before the establishment of irreversible brain damage would have resulted in better neurobehavioural outcomes. Classical phenylketonuria and milder phenotypes of phenylalanine hydroxylase deficiency need to be considered in the differential diagnosis of all cognitive and behavioural problems of unknown cause.