البحث الثالث

Relationship between time at diagnosis and clinical manifestations of phenylketonuria in a sample of Egyptian children.

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Abstract

Background: The aim of this study was to explore the relationship between age at the time of diagnosis of pediatric phenylketonuria (PKU) and clinical manifestations in a sample of Egyptian children.

Method: This retrospective cohort included children diagnosed as PKU. The collected data included the time, age, and phenylalanine (Phe) level at diagnosis, sex, serial plasma Phe levels, neurological and behavioral problems, developmental quotient, and anthropometric measurements.

Results: Sixty-six patients were included. Compared with early diagnosed patients, late cases had significantly lower median Phe levels ($P<0.001$) and higher rates of neurological complications ($P<0.001$) and behavioral problems ($P<0.001$). Compared with compliant patients, noncompliance was significantly associated with late diagnosis ($P=0.001$), neurological complications ($P=0.001$), and hyperactivity ($P=0.017$).

Conclusion: Stature and head circumference were not significantly affected by time of diagnosis or compliance with treatment. Early diagnosis and compliance with dietary treatment have a potential impact on neurological and behavioral problems in pediatric PKU patients

Keywords: behavioral problems, compliance, dietary restriction, growth, neurological complications, phenylketonuria