

Attention-deficit hyperactivity disorder in Brazilian patients with phenylketonuria

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Abstract

Recent studies have shown that patients with phenylketonuria (PKU), even with the early diagnosis and continuous treatment, may have symptoms of attention-deficit hyperactivity disorder (ADHD) and that the prevalence of ADHD in this population would be higher than in the general population. This study aims to determine the prevalence of ADHD in a sample of PKU patients from Southern Brazil. Patients were prospectively assessed by clinical interviews, neurological examination, and application of the MTA-SNAP-IV scales for patients aged 5-17 years and the Adult Self-Report Scale for patients over 17 years. Thirty-one patients (mean age = 17.4; early diagnosis = 27) were followed. Patients with ADHD and younger than 17 years had a median Phe in the last 6 months of life higher than those without the diagnosis of ADHD (ADHD patients = 617.1 $\mu\text{mol/L}$, no-ADHD patients 393.2 $\mu\text{mol/L}$, and $p = 0.03$). There was a predominantly hyperactive/impulsivity clinical presentation of ADHD ($n = 4/5$ patients), which differs from that reported elsewhere in the literature. Future studies are essential to better define the clinical presentation of ADHD in these patients and further elucidate its pathophysiology.