NEUROLOGICAL MANIFESTATIONS OF PYRUVATE DEHYDROGENASE COMPLEX DEFICIENCY

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Introduction

Pyruvate dehydrogenase complex deficiency (PDHc) is a rare metabolic disorder characterized by a spectrum of clinical manifestations, with an extremely heterogeneous severity and biochemical abnormalities.

Aim and Methods

Characterization of children/adolescents with PDHc followed in metabolic and neuropediatric consult, through analysis of clinical processes. Variables: gender, age at diagnosis, clinical manifestations, diagnosis and treatment.

Results

Five cases with PDHc deficiency were followed. Mean age at diagnosis was 37 months and the average time between the first consultation and diagnosis of 14.8 months. Most common neurological manifestations were delayed psychomotor development (5), behavioral changes(4), language disorders(3), hypotonia (3) and movement disorder (2 cases, one with dystonia and pyramidal syndrome and another with paroxysmal dystonia). There were no cases of epilepsy.

Average values of serum lactate and pyruvate were 3.13 + 1.07 mmol/L and 276.6 + 176.23 mmol/L, respectively. The main change in brain magnetic resonance was the abnormality of the corpus callosum (2). Average enzymatic activity of PDHc was 30.5%. Molecular study was performed in one case, showing E1α gene mutation.All patients began therapy with thiamine and ketogenic diet.

Conclusion

Neurological manifestations were present in all cases, reflecting the critical role of PDHc in brain development. Absence of epilepsy may be explained by the small number of cases or the early institution of ketogenic diet. We emphasize the need for diagnostic suspicion in the presence of early-onset neurological disease, especially if associated with structural brain abnormalities or unexplained lactic acidosis, given the potential severity of the disease.

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