Pediatric Neurology Part III: Chapter 169. Disorders of pyruvate metabolism (Handbook of Clinical Neurology)

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Pyruvate dehydrogenase and pyruvate carboxylase deficiency are the most common disorders in pyruvate metabolism. Diagnosis is made by enzymatic and DNA analysis after basic biochemical tests in plasma, urine, and CSF. Pyruvate dehydrogenase has three main subunits, an additional E3-binding protein and two complex regulatory enzymes. Most frequent are deficiencies in PDH-E1α. There is a spectrum of clinical presentations in E1α deficiency, ranging in boys from severe neonatal lactic acidosis, Leigh encephalopathy, to later onset of neurological disease such as intermittent ataxia or dystonia. Females tend to have a more uniform presentation resembling nonprogressive cerebral palsy. Neuroradiological abnormalities such as corpus callosum agenesis are seen more frequently in girls, basal ganglia and midbrain disturbances in boys. Deficiencies in the other subunits have also been described, but in a smaller number of patients. Pyruvate carboxylase deficiency has three clinical phenotypes. The infantile type is characterized mainly by severe developmental delay, failure to thrive, and seizures. The second type is characterized by neonatal onset of severe lactic acidosis with rigidity and hypokinesia. A third form is rarer with intermittent episodes of lactic acidosis and ketoacidosis. Neuroradiological findings such as cystic periventricular leukomalacia have been described.

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