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Pyruvate dehydrogenase deficiency

INSERM

Source

INSERM. (1999). *Orphanet: an online rare disease and orphan drug data base. Pyruvate dehydrogenase deficiency. ORPHA:765*

Pyruvate dehydrogenase deficiency (PDHD) is a rare neurometabolic disorder characterized by a wide range of clinical signs with metabolic and neurological components of varying severity. Manifestations range from often fatal, severe, neonatal lactic acidosis to later-onset neurological disorders. Six subtypes related to the affected subunit of the PDH complex have been recognized with significant clinical overlap: PDHD due to E1-alpha, E1-beta, E2 and E3 deficiency, PDHD due to E3-binding protein deficiency, and PDH phosphatase deficiency (see these terms).