Opis choroby *

Definicja

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).

Dane

Klasyfikacja
Choroba

Synonimy
PDH
Niedobór kompleksu dehydrogenazy pirogronianowej
PDH
PDHC
PDHC
Pyruvate dehydrogenase complex deficiency

Kod ORPHA
765

Kod OMIM
614111

Kod ICD10
E74.4

Kod ICD11
5C53.02

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*Źródło

orphanet