

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
pirogrońianowej

PDH

PDHC

PDHC

Pyruvate dehydrogenase complex deficiency

Kod ORPHA

765

Kod OMIM

614111

Kod ICD10

E74.4

Kod ICD11

5C53.02

[*Źródło](#)

orphanet