

## 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

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#### [\\*Źródło](#)

orphanet