

Opis choroby *

Definicja

Pyruvate dehydrogenase phosphatase deficiency is a very rare subtype of pyruvate dehydrogenase deficiency (PDHD, see this term) characterized by lactic acidemia in the neonatal period.

Dane

Klasyfikacja

Podtyp kliniczny

Synonimy

PDH phosphatase deficiency

Niedobór fosfatazy PDH

Kod ORPHA

79246

Kod OMIM

608782

Kod ICD10

E74.4

Kod ICD11

5C53.02

[*Źródło](#)

orphanet