

Opis choroby *

Definicja

Pyruvate dehydrogenase E3 deficiency is a very rare subtype of pyruvate dehydrogenase deficiency (PDHD, see this term) characterized by either early-onset lactic acidosis and delayed development, later-onset neurological dysfunction or liver disease.

Dane

Klasyfikacja

Podtyp kliniczny

Synonimy

DLD deficiency

Choroba syropu klonowego z deficytem E3

Niedobór dehydrogenazy dihydrolipoamidowej

Niedobór DLD

Dihydrolipoamide dehydrogenase deficiency

E3-deficient maple syrup urine disease

Kod ORPHA

2394

Kod OMIM

246900

Kod ICD10

E74.4

Kod ICD11

5C53.02

*Źródło

orphanet