Opis choroby *

Definicja

A very rare form of pyruvate dehydrogenase deficiency (PDHD) characterized by variable lactic acidosis and neurological dysfunction, mainly appearing during childhood.

Dane

Klasyfikacja
Podtyp kliniczny

Synonimy
Dihydrolipoamide acetyltransferase component of pyruvate dehydrogenase complex deficiency
Dihydrolipoamide acetyltransferase component of pyruvate dehydrogenase complex deficiency
Dihydrolipoyllysine-residue acetyltransferase component of pyruvate dehydrogenase complex deficiency
Niedobór dehydrogenazy pirogronianowej kompleksu komponentu E2
Dihydrolipoyllysine-residue acetyltransferase component of pyruvate dehydrogenase complex deficiency
Pyruvate dehydrogenase complex component E2 deficiency

Kod ORPHA
79244

Kod OMIM
245348

Kod ICD10
E74.4

Kod ICD11
5C53.02

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*Źródło

orphanet