

## 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