

## Opis choroby \*

### Definicja

A rare, genetic neurological disorder defined by early-onset of neurologic symptoms, biphasic clinical course, unique MRI features (incl. extensive, symmetrical, deep white matter abnormalities), and increased lactate in body fluids. The severe form is characterized by delayed psychomotor development, seizures, early-onset hypotonia, and persistently increased lactate levels. The mild form usually presents with irritability, psychomotor regression after six months of age, and temporary high lactate levels, with overall clinical improvement from the second year onward.

### Dane

#### Klasyfikacja

Choroba

#### Synonimy

COXPD12

LTBL

COXPD12

Złożony defekt fosforylacji oksydacyjnej typu 12

Combined oxidative phosphorylation defect type

12

LTBL

#### Kod ORPHA

314051

#### Kod OMIM

614924

#### Kod ICD10

E88.8

#### Kod ICD11

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#### \*Źródło

orphanet