

## Opis choroby \*

### Definicja

A rare mitochondrial oxidative phosphorylation disorder characterized by a variable clinical phenotype including infantile onset of epileptic encephalopathy, hypotonia, global developmental delay, failure to thrive, complex movement disorder, and liver involvement, as well as childhood onset of severe myoclonus epilepsy, cognitive decline, progressive hearing and visual impairment, and progressive tetraparesis. Serum lactate may be increased, and brain imaging shows variable atrophy and white matter abnormalities.

### Dane

#### Klasyfikacja

Choroba

#### Synonimy

COXPD27

COXPD27

#### Kod ORPHA

477774

#### Kod OMIM

616672

#### Kod ICD10

E88.8

#### Kod ICD11

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#### \*Źródło

orphanet