

## Opis choroby \*

### Definicja

A rare, inherited mitochondrial disorder due to a defect in mitochondrial protein synthesis characterized by intrauterine growth retardation, metabolic decompensation with recurrent vomiting, persistent severe lactic acidosis, encephalopathy, seizures, failure to thrive, severe global developmental delay, poor eye contact, severe muscular hypotonia or axial hypotonia with limb hypertonia, hepatomegaly and/or liver dysfunction and/or liver failure, leading to fatal outcome in severe cases. Neuroimaging abnormalities may include corpus callosum thinning, leukodystrophy, delayed myelination and basal ganglia involvement.

### Dane

#### Klasyfikacja

Choroba

#### Synonimy

Hepatoencephalopathy due to COXPD1

Encefalopatia wątrobowa z powodu COXPD1

#### Kod ORPHA

137681

#### Kod OMIM

609060

#### Kod ICD10

E88.8

#### Kod ICD11

5C53.23

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#### \*Źródło

orphanet