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

---

*Źródło

orphanet