Opis choroby *

Definicja

Combined oxidative phosphorylation defect type 2 is a rare mitochondrial disorder due to a defect in mitochondrial protein synthesis characterized by severe intrauterine growth retardation, neonatal limb edema and redundant skin on the neck (hydrops), developmental brain defects (corpus callosum agenesis, ventriculomegaly), brachydactyly, dysmorphic facial features with low set ears, severe intractable neonatal lactic acidosis with lethargy, hypotonia, absent spontaneous movements and fatal outcome. Markedly decreased activity of complex I, II + III and IV in muscle and liver have been determined.

Dane

Klasyfikacja
Choroba

Synonimy
COXPD2
COXPD2

Kod ORPHA
254920

Kod OMIM
610498

Kod ICD10
E88.8

Kod ICD11
5C53.23

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*Źródło

orphanet