

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

*Źródło

orphanet