

# Złożony defekt fosforylacji oksydacyjnej typu 2

## Kod Orpha: 254920 Kod OMIM: 610498

### 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

### Rozszerzony opis choroby

Brak opisu rozszerzonego dla tej choroby. Opracowanie w toku.