

## Opis choroby \*

### Definicja

Combined oxidative phosphorylation defect type 25 is a rare mitochondrial oxidative phosphorylation disorder with decreased respiratory complex I and IV enzyme activities, characterized by hypotonia, global developmental delay, neonatal onset of progressive pectus carinatum without other skeletal abnormalities, poor growth, sensorineural hearing loss, dysmorphic features and brain abnormalities such as cerebral atrophy, quadriventricular dilatation and thin corpus callosum posteriorly.

### Dane

#### Klasyfikacja

Choroba

#### Synonimy

COXPD25

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#### Kod ORPHA

447954

#### Kod OMIM

616430

#### Kod ICD10

E88.8

#### Kod ICD11

5C53.23

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#### \*Źródło

orphanet