

## Opis choroby \*

### Definicja

Combined oxidative phosphorylation deficiency type 3 is an extremely rare clinically heterogenous disorder described in about 5 patients to date. Clinical signs included hypotonia, lactic acidosis, and hepatic insufficiency, with progressive encephalomyopathy or hypertrophic cardiomyopathy.

### Dane

#### Klasyfikacja

Choroba

#### Synonimy

Fatal mitochondrial disease due to COXPD3

Śmiertelna choroba mitochondrialna

spowodowana COXPD3

#### Kod ORPHA

168566

#### Kod OMIM

610505

#### Kod ICD10

E88.8

#### Kod ICD11

5C53.23

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#### \*Źródło

orphanet