

## Opis choroby \*

### Definicja

Severe X-linked mitochondrial encephalomyopathy is an extremely rare mitochondrial respiratory chain disease resulting in a neurodegenerative disorder characterized by psychomotor delay, hypotonia, areflexia, muscle weakness and wasting in the two patients reported to date.

### Dane

#### Klasyfikacja

Choroba

#### Synonimy

Mitochondrial encephalomyopathy due to COXPD6

Encefalopatia mitochondrialna spowodowana COXPD6

Encefalomiopatia mitochondrialna spowodowana złożonym defektem fosforylacji oksydacyjnej 6

Mitochondrial encephalomyopathy due to combined oxidative phosphorylation defect 6

#### Kod ORPHA

238329

#### Kod OMIM

300816

#### Kod ICD10

E88.8

#### Kod ICD11

-

---

#### \*Źródło

orphanet