

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