

Złożony defekt fosforylacji oksydacyjnej typu 15

Kod Orpha: 319524 Kod OMIM: 614947

Opis choroby *

Definicja

Combined oxidative phosphorylation defect type 15 is a rare mitochondrial disease due to a defect in mitochondrial protein synthesis characterized by onset in infancy or early childhood of muscular hypotonia, gait ataxia, mild bilateral pyramidal tract signs, developmental delay (affecting mostly speech and coordination) and subsequent intellectual disability. Short stature, obesity, microcephaly, strabismus, nystagmus, reduced visual acuity, lactic acidosis, and a brain neuropathology consistent with Leigh syndrome are also reported.

Dane

Klasyfikacja

Choroba

Synonimy

COXPD15

COXPD15

Kod ORPHA

319524

Kod OMIM

614947

Kod ICD10

E88.8

Kod ICD11

5C53.23

[*Źródło](#)

orphanet

Rozszerzony opis choroby

Brak opisu rozszerzonego dla tej choroby. Opracowanie w toku.