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

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*Źródło

orphanet