Opis choroby *

Definicja

Combined oxidative phosphorylation defect type 13 is a rare mitochondrial disease due to a defect in mitochondrial protein synthesis characterized by normal early development followed by the sudden onset in infancy of poor feeding, dysphagia, truncal (followed by global) hypotonia, motor regression, abnormal movements (i.e. severe dystonia of limbs, choreoathetosis, facial dyskinesias) and reduced tendon reflexes. The disease course is severe but nonprogressive.

Dane

Klasyfikacja
Choroba

Synonimy
COXPD13
COXPD13

Kod ORPHA
319514

Kod OMIM
614932

Kod ICD10
E88.8

Kod ICD11
5C53.23

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*Źródło

orphanet