## 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