Opis choroby *

Definicja

Combined oxidative phosphorylation defect type 7 is a rare mitochondrial disease due to a defect in mitochondrial protein synthesis characterized by a variable phenotype that includes onset in infancy or early childhood of failure to thrive and psychomotor regression (after initial normal development), as well as ocular manifestations (such as ptosis, nystagmus, optic atrophy, ophthalmoplegia and reduced vision). Additional manifestations include bulbar paresis with facial weakness, hypotonia, difficulty chewing, dysphagia, mild dysarthria, ataxia, global muscle atrophy, and areflexia. It has a relatively slow disease progression with patients often living into the third decade of life.

Dane

Klasyfikacja
Choroba

Synonimy
COXPD7
COXPD7
Severe C12ORF65-related COXPD
Severe C12ORF65-related combined oxidative phosphorylation defect

Kod ORPHA
254930

Kod OMIM
613559

Kod ICD10
E88.8

Kod ICD11
5C53.23

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*Źródło

orphanet