Opis choroby *

Definicja

A rare mitochondrial disease characterized by a variable clinical phenotype ranging from fetal hydrops and postnatal hypotonia, bradycardia, and respiratory failure, resulting in death in the neonatal period, to infantile onset of episodes of acute cardiopulmonary failure associated with severe lactic acidosis, and slowly progressive muscle weakness. Muscle biopsy shows reduced activity of mitochondrial complexes I, III, and IV.

Dane

Klasyfikacja
Choroba

Synonimy
COXPD28
COXPD28
Złożony defekt fosforylacji oksydacyjnej typu 28
Combined oxidative phosphorylation defect type 28

Kod ORPHA
466784

Kod OMIM
616794

Kod ICD10
E88.8

Kod ICD11
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*Źródło

orphanet