

Opis choroby *

Definicja

A rare mitochondrial oxidative phosphorylation disorder characterized by neonatal onset of hypotonia, feeding difficulties, deafness, and early fatal respiratory failure. Cardiac and liver involvement has been reported. Serum lactate is increased, and metabolic studies show decreased activity of mitochondrial respiratory complexes I and IV in skeletal muscle.

Dane

Klasyfikacja

Choroba

Synonimy

COXPD30

COXPD30

Kod ORPHA

478042

Kod OMIM

616974

Kod ICD10

E88.8

Kod ICD11

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*Źródło

orphanet