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