Opis choroby *

Definicja

A rare, genetic, mitochondrial oxidative phosphorylation disorder characterized by a potentially life-threatening, severe myopathy manifesting in the neonatal to early infantile period, followed by marked, spontaneous improvement of muscular function by early childhood. Associated biochemical findings include lactic acidosis and a transient, marked decrease in respiratory chain activity.

Dane

Klasyfikacja
Choroba

Synonimy
Benign COX deficiency
Dziecięcy odwracalny Niedobór oksydazy cytochromu c
Łagodny Niedobór COX
Miopatia mitochondrialna z odwracalnym niedoborem COX
Miopatia mitochondrialna z odwracalnym niedoborem kompleksu IV
Infantile reversible cytochrome C oxidase deficiency myopathy
Mitochondrial myopathy with reversible COX deficiency
Mitochondrial myopathy with reversible complex IV deficiency
Reversible infantile cytochrome C oxidase deficiency
Reversible infantile respiratory chain deficiency

Kod ORPHA
254864

Kod OMIM
500009

Kod ICD10
G71.3

Kod ICD11
8C73.Y

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*Źródło

orphanet