

## 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](#)

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