Opis choroby *

Definicja

A rare mitochondrial oxidative phosphorylation disorder characterized by a highly variable clinical phenotype, including a benign infantile mitochondrial type affecting mainly the skeletal muscle, a lethal infantile mitochondrial myopathy linked to severe metabolic acidosis and mitochondrial dysfunction in skeletal muscle and often also in heart, Leigh syndrome, which causes severe, early-onset, progressive, and fatal encephalopathy, and French-Canadian type Leigh syndrome, which affects mostly the skeletal muscle, but also brain and liver.

Dane

Klasyfikacja
Choroba

Synonimy
Isolated COX deficiency
Izolowany Niedobór COX
Izolowany Niedobór kompleksu IV mitochondrialnego łańcucha oddechowego
Isolated mitochondrial respiratory chain complex IV deficiency

Kod ORPHA
254905

Kod OMIM
619052

Kod ICD10
E88.8

Kod ICD11
5C53.2Y

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*Źródło

orphanet