

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

[*Źródło](#)

orphanet