Opis choroby *

Definicja

Isolated complex I deficiency is a rare inborn error of metabolism due to mutations in nuclear or mitochondrial genes encoding subunits or assembly factors of the human mitochondrial complex I (NADH: ubiquinone oxidoreductase) and is characterized by a wide range of manifestations including marked and often fatal lactic acidosis, cardiomyopathy, leukoencephalopathy, pure myopathy and hepatopathy with tubulopathy. Among the numerous clinical phenotypes observed are Leigh syndrome, Leber hereditary optic neuropathy and MELAS syndrome (see these terms).

Dane

Klasyfikacja
Choroba

Synonimy
Isolated NADH-CoQ reductase deficiency
Izolowany Niedobór kompleksu I mitochondrialnego łańcucha oddechowego
Izolowany Niedobór reduktazy NADH-koenzym Q
Izolowany Niedobór reduktazy NADH-ubikwinon
Isolated NADH-coenzyme Q reductase deficiency
Isolated NADH-ubiquinone reductase deficiency
Isolated mitochondrial respiratory chain complex I deficiency

Kod ORPHA
2609

Kod OMIM
618242

Kod ICD10
G71.3

Kod ICD11
5C53.2Y

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*Źródło

orphanet