

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

*Źródło

orphanet