Opis choroby *

Definicja

A rare, mitochondrial oxidative phosphorylation disorder characterized by a highly variable phenotype. The severe, multisystemic disease involves brain, heart, muscles, liver, kidneys, and eyes and results in death in infancy. Mildly affected individuals have only isolated cardiac or muscle involvement in the adulthood. Histochemical and biochemical analysis reveals a global reduction of succinate dehydrogenase activity.

Dane

Klasyfikacja
Choroba

Synonimy
Isolated mitochondrial respiratory chain complex II deficiency
Izolowany Niedobór kompleksu II mitochondrialnego łańcucha oddechowego
Izolowany Niedobór reduktazy sukcynylo-koenzymu Q
Izolowany Niedobór reduktazy sukcynylo-ubichinionu
Isolated succinate dehydrogenase deficiency
Isolated succinate-coenzyme Q reductase deficiency
Isolated succinate-ubiquinone reductase deficiency

Kod ORPHA
3208

Kod OMIM
252011

Kod ICD10
G71.3

Kod ICD11
5C53.2Y

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*Źródło

orphanet