

Opis choroby *

Definicja

Mitochondrial encephalo-cardio-myopathy due to *TMEM70* mutation is characterized by early neonatal onset of hypotonia, hypertrophic cardiomyopathy and apneic spells within hours after birth accompanied by lactic acidosis, hyperammonemia and 3-methylglutaconic aciduria.

Dane

Klasyfikacja

Choroba

Synonimy

Mitochondrial encephalo-cardio-myopathy due to F1Fo ATPase deficiency
Mitochondrialna encefalo-kardio-miopatia z powodu niedoboru F1Fo ATP-azy
Mitochondrialna encefalo-kardio-miopatia z powodu niedoboru kompleksu V mitochondrialnego łańcucha oddechowego
Mitochondrialna encefalo-kardio-miopatia z powodu niedoboru syntazy ATP
Mitochondrial encephalo-cardio-myopathy due to isolated ATP synthase deficiency
Mitochondrial encephalo-cardio-myopathy due to isolated mitochondrial respiratory chain complex V deficiency

Kod ORPHA

1194

Kod OMIM

614052

Kod ICD10

G71.3

Kod ICD11

5C53.2Y

*Źródło

orphanet