Opis choroby *

Definicja

Mitochondrial encephalo-cardio-myopathy due to TMEM70 mutation is characterized by early neonatal onset of hypotonia, hypetrophic 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

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*Źródło

orphanet