## Opis choroby *

Definicja

A rare mitochondrial oxidative phosphorylation disorder characterized by a spectrum of three main clinical phenotypes comprising a severe neonatal phenotype with early fatal lactic acidosis, a more protracted course with early-onset developmental delay, motor weakness, extrapyramidal signs, and with or without epilepsy, and a phenotype with normal early development and Parkinson-like symptoms starting around the age of one year. Additional, variably reported, signs and symptoms include cardiomyopathy, optic anomalies, hepatosplenomegaly, and abnormal brain MRI findings, among others. Deficiencies in mitochondrial oxidative phosphorylation enzymes are inconsistent.

Dane

**Klasyfikacja**
Choroba

Synonimy
Mitochondrial tryptophanyl-tRNA synthetase deficiency
Mitochondrial tryptophanyl-tRNA synthetase deficiency

**Kod ORPHA**
572798

**Kod OMIM**
617710

**Kod ICD10**
E88.8

**Kod ICD11**
5C53.2Y

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*Źródło

orphanet