

## Opis choroby \*

### Definicja

A rare mitochondrial DNA depletion syndrome characterized by congenital or early-onset lactic acidosis, hypotonia, and severe global developmental delay with feeding difficulties and failure to thrive. It is frequently associated with variable dysmorphic facial features. Additional manifestations include seizures, movement disorders, and cardiac and ophthalmologic anomalies, among others. Brain imaging may show generalized atrophy and white matter abnormalities.

### Dane

#### Klasyfikacja

Choroba

#### Synonimy

mtDNA depletion syndrome,  
encephalomyopathic form with variable  
craniofacial anomalies  
Zespół deplecji mtDNA, postać  
encefalomiopatyczna ze zróżnicowanymi  
wadami twarzoczaszki

#### Kod ORPHA

369897

#### Kod OMIM

615471

#### Kod ICD10

E88.8

#### Kod ICD11

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#### \*Źródło

orphanet