

## Opis choroby \*

### Definicja

A rare mitochondrial disease characterized by signs and symptoms within a phenotypic and metabolic spectrum that includes global developmental delay, hypotonia, intellectual disability, optic atrophy, axonal neuropathy, hypertrophic cardiomyopathy, lactic acidosis, and increased excretion of Krebs cycle intermediates. Other variable features are spasticity, seizures, ataxia, congenital cataract, and dysmorphic facial features. Age of onset is in the neonatal period or infancy.

### Dane

#### Klasyfikacja

Choroba

#### Synonimy

Harel-Yoon syndrome

Zespół Harel i Yoona

#### Kod ORPHA

496790

#### Kod OMIM

617183

#### Kod ICD10

E88.8

#### Kod ICD11

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#### \*Źródło

orphanet