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