Opis choroby *

Definicja

A rare mitochondrial DNA depletion syndrome characterized by neonatal or infantile onset of hypotonia, failure to thrive, global developmental delay, and persistent lactic acidosis. The disease course is variable and ranges from intractable diarrhea and respiratory failure with fatal outcome in early infancy to a milder phenotype with survival into childhood. Additional reported features include sensorineural hearing loss, microcephaly, seizures, pigmentary retinopathy, and renal tubulopathy.

Dane

Klasyfikacja
Choroba

Synonimy
mtDNA depletion syndrome, encephalomyopathic form with renal tubulopathy
Zespół deplecji mtDNA, postać encefalomiopatyczna z tubulopatią

Kod ORPHA
255235

Kod OMIM
612075

Kod ICD10
G31.8

Kod ICD11
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*Źródło

orphanet