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