Opis choroby *

Definicja

A rare mitochondrial DNA depletion syndrome characterized by neonatal or infantile onset of global developmental delay, hypotonia, failure to thrive, progressive neurologic decline, sensorineural deafness, and movement disorder. Seizures, external ophthalmoplegia, polyneuropathy, cardiomyopathy, and renal tubular dysfunction have also been reported. Brain imaging may show T2-weighted hyperintensities in the basal ganglia, and laboratory examination may reveal lactic acidosis and mild methylmalonic aciduria.

Dane

Klasyfikacja
Choroba

Synonimy
Booth-Haworth-Dilling syndrome
Mitochondrialna encefalomiopatia - aminoacidopatia
Zespół Bootha, Hawortha i Dillinga
Zespół deplecji mtDNA, forma z encefalomiopatią i kwasicą metylomalonową
Mitochondrial encephalomyopathy-aminoacidopathy syndrome
mtDNA depletion syndrome, encephalomyopathic form with methylmalonic aciduria

Kod ORPHA
1933

Kod OMIM
612073

Kod ICD10
G71.3

Kod ICD11
5C53.20

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*Źródło

orphanet