

## 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