Opis choroby *

Definicja

A form of mitochondrial DNA depletion syndrome that displays a broad phenotypic spectrum but that is most often characterized by hypotonia, proximal muscle weakness, facial and bulbar weakness and failure to thrive.

Dane

Klasyfikacja
Choroba

Synonimy
mtDNA depletion syndrome, myopathic form
Zespół deplecji mtDNA, postać miopatyczna

Kod ORPHA
254875

Kod OMIM
609560

Kod ICD10
G71.3

Kod ICD11
5C53.20

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*Źródło

orphanet