

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

*Źródło

orphanet