

## Opis choroby \*

### Definicja

A rare autosomal dominant hereditary axonal motor and sensory neuropathy characterized by early onset of generalized hypotonia and weakness, or later onset of distal lower limb muscle weakness and atrophy, cramps, and sensory impairment. Weakness and atrophy progress in an asymmetric fashion to involve also the proximal and upper limbs in the course of the disease. Additional features are pyramidal signs like increased muscle tone and extensor plantar reflexes, as well as learning difficulties.

### Dane

#### Klasyfikacja

Choroba

#### Synonimy

Autosomal dominant Charcot-Marie-Tooth disease type 2 due to MORC2 mutation  
Autosomalna dominująca choroba Charcota, Mariego i Tootha typu 2 spowodowana mutacją MORC2  
CMT2Z  
CMT2Z

#### Kod ORPHA

466768

#### Kod OMIM

616688

#### Kod ICD10

G60.0

#### Kod ICD11

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#### \*Źródło

orphanet