

## Opis choroby \*

### Definicja

A rare, axonal hereditary motor and sensory neuropathy characterized by progressive distal muscle weakness and atrophy of variable onset and severity. Patients present with postural instability, gait and running difficulties, decreased deep tendon reflexes, foot deformities, fine motor impairment, and distal sensory impairment. Dysarthria, dysphagia, and mild cognitive and behavioral abnormalities have also been reported.

### Dane

#### Klasyfikacja

Choroba

#### Synonimy

Autosomal dominant Charcot-Marie-Tooth disease type 2 due to VCP mutation  
CMT2 spowodowana mutacją VCP  
CMT2 due to VCP mutation  
CMT2Y

#### Kod ORPHA

435387

#### Kod OMIM

616687

#### Kod ICD10

G60.0

#### Kod ICD11

8C20.1

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#### \*Źródło

orphanet