

## Opis choroby \*

### Definicja

Charcot-Marie-Tooth disease type 4C (CMT4C) is a subtype of Charcot-Marie-Tooth type 4 characterized by childhood or adolescent-onset of a relatively mild, demyelinating sensorimotor neuropathy that contrasts with a severe, rapidly progressing, early-onset scoliosis, and the typical CMT phenotype (i.e. distal muscle weakness and atrophy, sensory loss, and often foot deformity). A wide spectrum of nerve conduction velocities are observed and cranial nerve involvement and kyphoscoliosis have also been reported.

### Dane

#### Klasyfikacja

Choroba

#### Synonimy

CMT4C

CMT4C

#### Kod ORPHA

99949

#### Kod OMIM

601596

#### Kod ICD10

G60.0

#### Kod ICD11

8C20.0

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#### \*Źródło

orphanet