

# **Choroba Charcota, Mariego i Tootha typu 4C**

## **Kod Orpha: 99949 Kod OMIM: 601596**

### **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

### **Rozszerzony opis choroby**

Brak opisu rozszerzonego dla tej choroby. Opracowanie w toku.