

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

*Źródło

orphanet