

Autosomalna dominująca choroba Charcota, Mariego i Tootha typu 2L

Kod Orpha: 99945 Kod OMIM: 608673

Opis choroby *

Definicja

A form of axonal Charcot-Marie-Tooth disease, a peripheral sensorimotor neuropathy. In the single family reported to date, CMT2L onset is between 15 and 33 years. Patients present with a symmetric distal weakness of legs and occasionally of the hands, absent or reduced tendon reflexes, distal legs sensory loss and frequently a pes cavus. Progression is slow.

Dane

Klasyfikacja

Choroba

Synonimy

CMT2L

CMT2L

Kod ORPHA

99945

Kod OMIM

608673

Kod ICD10

G60.0

Kod ICD11

8C20.1

[*Źródło](#)

orphanet

Rozszerzony opis choroby

Brak opisu rozszerzonego dla tej choroby. Opracowanie w toku.