

Opis choroby *

Definicja

X-linked Charcot-Marie-Tooth disease type 1 is a rare, genetic, peripheral sensorimotor neuropathy characterized by an X-linked dominant inheritance pattern and the childhood-onset (within the first decade in males) of progressive, distal, moderate to severe muscle weakness and atrophy in lower extremities and intrinsic hand muscles, pes cavus, bilateral foot drop, reduced or absent tendon reflexes, as well as mild to moderate sensory impairment in lower extremities. Females tend to have milder manifestations or may be asymptomatic. Sensorineural deafness and central nervous system involvement have also been reported.

Dane

Klasyfikacja

Choroba

Synonimy

CMT1X

CMT1X

CMTX1

CMTX1

Kod ORPHA

101075

Kod OMIM

302800

Kod ICD10

G60.0

Kod ICD11

8C20.0

*Źródło

orphanet