

Choroba Charcota, Mariego i Tootha typu 3 sprzężona z chromosomem X

Kod Orpha: 101077 Kod OMIM: 302802

Opis choroby *

Definicja

X-linked Charcot-Marie-Tooth disease type 3 is a rare, genetic, peripheral sensorimotor neuropathy characterized by an X-linked recessive inheritance pattern and the childhood- to adolescent-onset of progressive, distal muscle weakness and atrophy (beginning in the lower extremities and then affecting the upper extremities), as well as distal, pansenory loss in the upper and lower extremities, pes cavus, and absent or reduced distal tendon reflexes. Pain and paresthesia are frequently the initial sensory symptoms. Spastic paraparesis (manifested by clasp-knife sign, hyperactive deep-tendon reflexes, and Babinski sign) has also been reported.

Dane

Klasyfikacja

Choroba

Synonimy

CMT3X

CMT3X

CMTX3

CMTX3

Kod ORPHA

101077

Kod OMIM

302802

Kod ICD10

G60.0

Kod ICD11

LD90.Y

[*Źródło](#)

orphanet

Rozszerzony opis choroby

Brak opisu rozszerzonego dla tej choroby. Opracowanie w toku.

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