Choroba Charcota, Mariego i Tootha typu 4 sprzężona z chromosomem X
Kod Orpha: 101078 Kod OMIM: 310490

Opis choroby *

Definicja
X-linked Charcot-Marie-Tooth disease type 4 is a rare, genetic, axonal, peripheral sensorimotor neuropathy characterized by an X-linked recessive inheritance pattern and the neonatal- to early childhood-onset of severe, slowly progressive, distal muscle weakness and atrophy (in particular of the peroneal group), as well as sensory impairment (with the lower extremities being more affected than the upper extremities), pes cavus, areflexia and hammertoes. Sensorineural hearing loss and cognitive impairment may also be associated. Females are asymptomatic and do not display the phenotype.

Dane

Klasyfikacja
Choroba

Synonimy
CMT4X
CMT4X
CMTX4
Zespół Cowchocka
CMTX4
Cowchock syndrome

Kod ORPHA
101078

Kod OMIM
310490

Kod ICD10
G60.0

Kod ICD11
LD90.Y

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*Źródło

orphanet

Rozszerzony opis choroby

Brak opisu rozszerzonego dla tej choroby. Opracowanie w toku.