

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

[*Źródło](#)

orphanet