

Choroba Charcota, Mariego i Tootha typu 4 sprzęzona 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	Synonimy
Choroba	CMT4X
	CMT4X
	CMTX4
	Zespół Cowchocka
	CMTX4
	Cowchock syndrome

Kod ORPHA	Kod OMIM	Kod ICD10
101078	310490	G60.0

Kod ICD11
LD90.Y

[* Źródło](#)

[orphanet](http://www.orphanet.org)

Rozszerzony opis choroby

Brak opisu rozszerzonego dla tej choroby. Opracowanie w toku.

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