

Opis choroby *

Definicja

X-linked Charcot-Marie-Tooth disease type 6 is a rare, genetic, principally axonal, peripheral sensorimotor neuropathy characterized by an X-linked dominant inheritance pattern and the childhood-onset of slowly progressive, moderate to severe, distal muscle weakness and atrophy of the lower extremities, as well as distal, panmodal sensory abnormalities, bilateral foot deformities (pes cavus, clawed toes), absent ankle reflexes and gait abnormalities (steppage gait). Females are usually asymptomatic or only present mild manifestations (mild postural hand tremor, mild wasting of hand intrinsic muscles).

Dane

Klasyfikacja

Choroba

Synonimy

CMT6X

CMT6X

CMTX6

CMTX6

Kod ORPHA

352675

Kod OMIM

300905

Kod ICD10

G60.0

Kod ICD11

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*Źródło

orphanet