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, pansensory 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

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*Źródło

orphanet