Opis choroby *

Definicja

X-linked Charcot-Marie-Tooth disease type 2 is a rare, genetic, peripheral sensorimotor neuropathy characterized by an X-linked recessive inheritance pattern and the infantile- to childhood-onset of progressive, distal muscle weakness and atrophy (more prominent in the lower extremities than in the upper extremities), pes cavus, and absent tendon reflexes. Sensory impairment and intellectual disability has been reported in some individuals.

Dane

Klasyfikacja
Choroba

Synonimy
CMTX2
CMT2X
CMTX2

Kod ORPHA
101076

Kod OMIM
302801

Kod ICD10
G60.0

Kod ICD11
LD90.Y

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*Źródło

orphanet