

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

*Źródło

orphanet