Opis choroby *

Definicja

A rare autosomal recessive axonal hereditary motor and sensory neuropathy characterized by childhood to adult onset of slowly progressive, sometimes asymmetric distal muscle weakness and atrophy, as well as sensory impairment, predominantly of the lower limbs. Additional common features include pes cavus, kyphoscoliosis, ankle contractures, tremor, or urogenital dysfunction. Fasciculations and proximal involvement may be seen in some cases. Patients usually remain ambulatory.

Dane

Klasyfikacja
Choroba

Synonimy
ARCMT2X
ARCMT2X
Autosomalna recesywna choroba Charcota, Mariego i Tootha typu 2 spowodowana SPG11
CMT2X
Autosomal recessive Charcot-Marie-Tooth disease type 2 due to SPG11 mutation
CMT2X

Kod ORPHA
466775

Kod OMIM
616668

Kod ICD10
G60.0

Kod ICD11
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*Źródło

orphanet