Opis choroby *

Definicja

A rare autosomal dominant hereditary axonal motor and sensory neuropathy characterized by early onset of generalized hypotonia and weakness, or later onset of distal lower limb muscle weakness and atrophy, cramps, and sensory impairment. Weakness and atrophy progress in an asymmetric fashion to involve also the proximal and upper limbs in the course of the disease. Additional features are pyramidal signs like increased muscle tone and extensor plantar reflexes, as well as learning difficulties.

Dane

Klasyfikacja
Choroba

Synonimy
Autosomal dominant Charcot-Marie-Tooth disease type 2 due to MORC2 mutation
Autosomalna dominująca choroba Charcota, Mariego i Tootha typu 2 spowodowana mutacją MORC2
CMT2Z
CMT2Z

Kod ORPHA
466768

Kod OMIM
616688

Kod ICD10
G60.0

Kod ICD11
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*Źródło

orphanet