Opis choroby *

Definicja

A rare predominantly axonal hereditary motor and sensory neuropathy characterized by a broad phenotypic spectrum of slowly progressive signs and symptoms mainly affecting the lower limbs. Most patients present with gait difficulties and distal sensory impairment, while some may lack sensory symptoms altogether. Pes cavus is frequently reported. Age of onset is also highly variable, ranging from childhood to late adulthood.

Dane

Klasyfikacja
Choroba

Synonimy
Autosomal dominant Charcot-Marie-Tooth disease type 2 due to HARS mutation
Autosomalna dominująca choroba Charcota, Mariego i Toothа typu 2 z powodu mutacji w HARS
CMT2W

Kod ORPHA
488333

Kod OMIM
616625

Kod ICD10
G60.0

Kod ICD11
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*Źródło

orphanet