

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 Tootha 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