Opis choroby *

Definicja

A rare, autosomal dominant, hereditary, demyelinating motor and sensory neuropathy which may present either as a classic Charcot-Marie-Tooth disease phenotype with distal motor weakness and wasting, gait difficulties, parethesias, decreased vibration and pain sensation, or as a milder, predominantly sensory form with transient paresthesias, decreased sensation and distal pain in upper or lower limbs, without significant motor weakness. Pes cavus is a common feature, and additional symptoms may include hand tremor and decreased or absent deep tendon reflexes.

Dane

Klasyfikacja
Choroba

Synonimy
CMT1C
CMT1C

Kod ORPHA
101083

Kod OMIM
601098

Kod ICD10
G60.0

Kod ICD11
8C20.0

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*Źródło

orphanet