Opis choroby *

Definicja

A rare, axonal hereditary motor and sensory neuropathy characterized by progressive distal muscle weakness and atrophy of variable onset and severity. Patients present with postural instability, gait and running difficulties, decreased deep tendon reflexes, foot deformities, fine motor impairment, and distal sensory impairment. Dysarthria, dysphagia, and mild cognitive and behavioral abnormalities have also been reported.

Dane

Klasyfikacja
Choroba

Synonimy
Autosomal dominant Charcot-Marie-Tooth disease type 2 due to VCP mutation
CMT2 spowodowana mutacją VCP
CMT2 due to VCP mutation
CMT2Y

Kod ORPHA
435387

Kod OMIM
616687

Kod ICD10
G60.0

Kod ICD11
8C20.1

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*Źródło

orphanet