Opis choroby *

Definicja

A rare autosomal dominant hereditary demyelinating motor and sensory neuropathy characterized by progressive distal muscle weakness and atrophy, distal sensory impairment, and decreased or absent reflexes in the affected limbs, with an onset in the first or second decade of life. Median motor nerve conduction velocities are typically less than 38 m/s. Patients often have foot deformities. Sural nerve biopsy shows decrease in myelinated fibers, myelin abnormalities, and onion bulb formation. Fatty replacement of muscle tissue predominantly affects the anterior and lateral compartment of the lower legs.

Dane

Klasyfikacja
Choroba

Synonimy
PMP2-related CMT1
CMT1 zależna od PMP2
Dziedziczna neuropatia Charcota, Mariego i Toothа typu 1 zależna od PMP2
Neuropatia Charcota, Mariego i Toothа typu 1 zależna od PMP2
PMP2-related Charcot-Marie-Tooth neuropathy type 1
PMP2-related hereditary motor and sensory neuropathy type 1

Kod ORPHA
476394

Kod OMIM
618279

Kod ICD10
G60.0

Kod ICD11
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*Źródło

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