Opis choroby *

Definicja

A subtype of autosomal dominant Charcot-Marie-Tooth disease type 2, characterized by late adult-onset (50-60 years of age) of slowly progressive, axonal, peripheral sensorimotor neuropathy resulting in distal upper limb and proximal and distal lower limb muscle weakness and atrophy, in conjunction with distal, panmodal sensory impairment in upper and lower limbs. Tendon reflexes are reduced and nerve conduction velocities range from reduced to absent. Neuropathic pain has also been associated.

Dane

Klasyfikacja
Choroba

Synonimy
Autosomal dominant Charcot-Marie-Tooth disease type 2 due to MARS mutation
Autosomalna dominująca choroba Charcota, Mariego i Tootha typu 2 z powodu mutacji MARS
CMT2U
CMT2U

Kod ORPHA
397735

Kod OMIM
616280

Kod ICD10
G60.0

Kod ICD11
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*Źródło

orphanet