Opis choroby *

Definicja

A rare autosomal recessive axonal hereditary motor and sensory neuropathy characterized by adult onset of slowly progressive distal muscle weakness and atrophy, sensory impairment, and decreased or absent deep tendon reflexes predominantly in the lower extremities. Patients present gait disturbances but remain ambulatory. Mild involvement of the upper limbs may be seen.

Dane

Klasyfikacja
Choroba

Synonimy
AR-CMT2T
Choroba Charcota, Mariego i Tootha aksonalna, autosomalna recesywna, typu 2T
Autosomal recessive axonal Charcot-Marie-Tooth disease type 2T
CMT2T

Kod ORPHA
495274

Kod OMIM
617017

Kod ICD10
G60.0

Kod ICD11
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*Źródło

orphanet