

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