Opis choroby *

Definicja

A rare autosomal recessive axonal hereditary motor and sensory neuropathy characterized by adolescent or adult onset of slowly progressive muscle weakness and atrophy of the distal lower limbs progressing to involve also the upper limbs and proximal muscles, and sensory impairment. Patients present gait disturbances and loss of reflexes, at later stages loss of ambulation, dysarthria, dysphagia, facial weakness, and impairment of respiratory muscles requiring assisted ventilation.

Dane

Klasyfikacja
Choroba

Synonimy
DNAJB2-related CMT2
AR-CMT2T
Autosomalna recesywna aksonalna choroba Charcota, Mariego i Tootha typu 2T
CMT2T
Choroba Charcot-Marie-Tooth typu 2 związana z DNAJB2

Kod ORPHA
443950

Kod OMIM
614881

Kod ICD10
G60.0

Kod ICD11
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*Źródło

orphanet