

Opis choroby *

Definicja

A rare hereditary motor and sensory neuropathy characterized by intermediate motor median nerve conduction velocities (usually between 25 and 45 m/s) and signs of both demyelination and axonal degeneration in nerve biopsies. It presents with usual clinical features of Charcot-Marie-Tooth disease (progressive muscle weakness and atrophy of the distal extremities, distal sensory loss, reduced or absent deep tendon reflexes, and feet deformities) in the first to second decade of life with steady progression until the fourth decade, severe progression and stabilization afterwards.

Dane

Klasyfikacja

Choroba

Synonimy

CMTDIA

CMTDIA

Kod ORPHA

100043

Kod OMIM

606483

Kod ICD10

G60.0

Kod ICD11

8C20.2

*Źródło

orphanet