

Opis choroby *

Definicja

Charcot-Marie-Tooth disease type 4B3 (CMT4B3) is a subtype of Charcot-Marie-Tooth type 4 characterized by a childhood onset of slowly progressing, demyelinating sensorimotor neuropathy, focally folded myelin sheaths in nerve biopsy, reduced nerve conduction velocities (less than 38 m/s), and the typical CMT phenotype (i.e. distal muscle weakness and atrophy, and sensory loss).

Dane

Klasyfikacja

Choroba

Synonimy

CMT4B3

Choroba Charcota, Mariego i Tootha z ogniskowym pofałdowaniem mieliny

CMT4B3

Charcot-Marie-Tooth disease with focally folded myelin

Kod ORPHA

363981

Kod OMIM

615284

Kod ICD10

G60.0

Kod ICD11

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*Źródło

orphanet