

Opis choroby *

Definicja

Charcot-Marie-Tooth disease type 4F (CMT4F) is a severe, demyelinating subtype of Charcot-Marie-Tooth disease type 4 characterized by the childhood onset of a slowly-progressing typical CMT phenotype (i.e. distal muscle weakness and atrophy, as well as pes cavus) that presents severe sensory loss (frequently with sensory ataxia), moderately to severely reduced motor nerve conduction velocities and almost invariable absence of sensory nerve action potentials, and delayed motor milestones.

Dane

Klasyfikacja

Choroba

Synonimy

CMT4F

CMT4F

Kod ORPHA

99952

Kod OMIM

614895

Kod ICD10

G60.0

Kod ICD11

8C20.0

*Źródło

orphanet