

Opis choroby *

Definicja

Charcot-Marie-Tooth disease type 4B2 (CMT4B2) is a subtype of Charcot-Marie-Tooth type 4 characterized by a severe, early childhood-onset of demyelinating sensorimotor neuropathy, early-onset glaucoma, focally folded myelin sheaths in the peripheral nerves, severely reduced nerve conduction velocities, and the typical CMT phenotype (i.e. distal muscle weakness and atrophy, sensory loss, and frequent pes cavus). Severe visual impairment leading to visual loss has also been reported.

Dane

Klasyfikacja

Choroba

Synonimy

CMT4B2

CMT4B2

Kod ORPHA

99956

Kod OMIM

604563

Kod ICD10

G60.0

Kod ICD11

8C20.0

*Źródło

orphanet