Opis choroby *

Definicja

Charcot-Marie-Tooth disease type 4D (CMT4D) is a subtype of Charcot-Marie-Tooth disease type 4 characterized by a childhood-onset of severe, progressive, demyelinating sensorimotor neuropathy manifesting with distal muscle weakness and atrophy, sensorineural hearing impairment leading to deafness (usually in third decade), severely reduced nerve conduction velocities, and skeletal, especially foot, deformities. Tongue atrophy has also been reported.

Dane

Klasyfikacja
Choroba

Synonimy
CMT4D
CMT4D
Dziedziczna neuropatia ruchowa i czuciowa, typ Loma
HMSN, typ Loma
HMSN-Lom
HMSN, Lom type
HMSN-Lom
Hereditary motor and sensory neuropathy, Lom type

Kod ORPHA
99950

Kod OMIM
601455

Kod ICD10
G60.0

Kod ICD11
8C20.0

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*Źródło

orphanet