Opis choroby *

Definicja

A rare form of X-linked Charcot-Marie-Tooth disease, a peripheral sensorimotor neuropathy, characterized by infancy- to childhood-onset of: 1) progressive distal muscle weakness and atrophy (first appearing and more prominent in the lower extremities than the upper) which usually manifests with foot drop and gait disturbance, 2) bilateral, profound, prelingual sensorineural hearing loss and 3) progressive optic neuropathy.

Dane

Klasyfikacja
Choroba

Synonimy
CMT5X
CMT5X
CMTX5
CMTX5

Kod ORPHA
99014

Kod OMIM
311070

Kod ICD10
G60.0

Kod ICD11
LD90.Y

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*Źródło

orphanet