

## 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