

Opis choroby *

Definicja

A form of axonal Charcot-Marie-Tooth disease, a peripheral motor and sensory neuropathy, characterized by congenital ptosis and early cataract associated to a mildly progressive peripheral neuropathy of variable onset from birth to the 6th decade, pes cavus, reduced to absent ankles tendon reflexes and sometimes neutropenia.

Dane

Klasyfikacja

Choroba

Synonimy

CMT2M

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Kod ORPHA

228179

Kod OMIM

606482

Kod ICD10

G60.0

Kod ICD11

8C20.1

*Źródło

orphanet