

Opis choroby *

Definicja

A form of axonal Charcot-Marie-Tooth disease, a peripheral sensorimotor neuropathy, with onset in the first to 6th decade with a gait anomaly and a leg weakness that reaches the arms secondarily. Tendon reflexes are reduced or absent and, after years, all patients have a pes cavus. Other signs may be present, including hearing loss and postural tremor.

Dane

Klasyfikacja

Choroba

Synonimy

CMT2E

CMT2E

Kod ORPHA

99939

Kod OMIM

607684

Kod ICD10

G60.0

Kod ICD11

8C20.1

*Źródło

orphanet