

Opis choroby *

Definicja

A rare, axonal hereditary motor and sensory neuropathy characterized by adult onset of recurrent pain in legs with or without cramps, progressive loss of deep tendon reflexes and vibration sense, paresthesias in the feet and later in the hands. Patients often experience sleep disturbances and mild sensory ataxia.

Dane

Klasyfikacja

Choroba

Synonimy

Autosomal dominant Charcot-Marie-Tooth disease type 2 due to NAGLU mutation

Autosomalna dominująca choroba Charcota, Mariego i Tootha typu 2 spowodowana mutacją NAGLU

CMT2V

Dziedziczna bolesna polineuropatia aksonalna o początku w wieku dorosłym

CMT2V

Hereditary adult-onset painful axonal polyneuropathy

Kod ORPHA

447964

Kod OMIM

616491

Kod ICD10

G60.0

Kod ICD11

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*Źródło

orphanet