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 Toothа 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