Opis choroby *

Definicja

A rare peripheral neuropathy characterized by slowly progressive axonal, motor greater than sensory, polyneuropathy combined with neuromytonia (including spontaneous muscular activity at rest (myokymia), impaired muscle relaxation (pseudomyotonia), and contractures of hands and feet) and neuromyotonic or myokymic discharges on needle EMG. It presents with distal lower limb weakness with gait impairment, muscle stiffness, fasciculations and cramps in hands and legs worsened by cold, decreased to absent tendon reflexes, intrinsic hand muscle atrophy and, variably, mild distal sensory impairment.

Dane

Klasyfikacja
Choroba

Synonimy
ARAN-NM
ARAN-NM
ARCMT2-NM
Autosomalna recesywna choroba Charcota, Mariego i Tootha typu 2 z neuromiotonią
ARCMT2-NM
Autosomal recessive Charcot-Marie-Tooth disease type 2 with neuromyotonia

Kod ORPHA
324442

Kod OMIM
137200

Kod ICD10
G60.0

Kod ICD11
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*Źródło

orphanet