Opis choroby *

Definicja

A rare autosomal dominant hereditary axonal motor and sensory neuropathy characterized by predominantly distal weakness and muscle atrophy, decreased or absent tendon reflexes, and reduced vibratory sensation in the lower and upper extremities. Pes cavus develops in many patients. Additional symptoms like ataxia, tremor, or swallowing difficulties have been reported. Patients usually remain ambulatory even late in the disease. Age of onset ranges from childhood to adulthood, with earlier onset tending to be associated with a more severe disease phenotype.

Dane

Klasyfikacja
Choroba

Synonimy
ATP1A1-related CMT2
CMT2DD
ATP1A1-związana z CMT2
Autosomalna dominująca choroba Charcota, Mariego i Tootha typu 2 związana z ATP1A1
ATP1A1-related autosomal dominant Charcot-Marie-Tooth disease type 2
CMT2DD

Kod ORPHA
521414

Kod OMIM
618036

Kod ICD10
G60.0

Kod ICD11
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*Źródło

orphanet