

## 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

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