

## Opis choroby \*

### Definicja

A rare form of axonal peripheral sensorimotor neuropathy characterized by classical CMT2 signs and symptoms (progressive weakness and atrophy of distal limb muscles, mild sensory deficits of position, vibration and pain/temperature, pes cavus, and symmetrically absent or reduced muscle and sensory action potentials with relatively preserved nerve conduction velocities in neurophysiological studies) as well as pyramidal tract involvement (spasticity, hyperreflexia). Spasticity and pain may be the presenting symptoms.

### Dane

#### Klasyfikacja

Choroba

#### Synonimy

CMT2 due to KIF5A mutation  
CMT z powodu mutacji KIF5A

#### Kod ORPHA

324611

#### Kod OMIM

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#### Kod ICD10

G60.0

#### Kod ICD11

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#### \*Źródło

orphanet