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