Opis choroby *

Definicja

A rare subtype of axonal hereditary motor and sensory neuropathy characterized by progressive distal muscle weakness and atrophy of both the lower and upper limbs, absent or reduced deep tendon reflexes, mild sensory loss, foot drop, and pes cavus leading eventually to wheelchair dependance. Some patients present with early hypotonia and delayed motor development. Scoliosis and variable autonomic disturbances may be associated.

Dane

Klasyfikacja
Choroba

Synonimy
CMT2S
CMT2S

Kod ORPHA
443073

Kod OMIM
616155

Kod ICD10
G60.0

Kod ICD11
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*Źródło

orphanet