Opis choroby *

Definicja

A rare autosomal dominant distal hereditary motor neuropathy disease characterized by muscle weakness and wasting predominantly affecting the hands, in particular the thenar and first dorsal interosseus muscles, and/or marked foot deformity and gait disturbance. Sensation is normal, although reduced response to vibration has been described. The disease is slowly progressive with an age of onset within the first few decades of life.

Dane

Klasyfikacja
Choroba

Synonimy
Distal HMN V
dHMN5
Dystalna HMN V
Dystalny rdzeniowy zanik mięśni typu 5
Dystalna dziedziczna neuropatia ruchowa typu V
Distal hereditary motor neuropathy type V
Distal spinal muscular atrophy type 5
dHMN5

Kod ORPHA
139536

Kod OMIM
619112

Kod ICD10
G12.2

Kod ICD11
8B61.4

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*Źródło

orphanet