Opis choroby *

Definicja

A rare, genetic, neuromuscular disease characterized by progressive, symmetrical, moderate to severe, distal muscle weakness and atrophy, without sensory involvement, first affecting the lower limbs (towards the end of the first decade) and then involving (within two years) the upper extremities. Patients typically develop foot drop, pes varus, hammer toes and claw hands. Pyramidal tract signs (such as brisk knee reflexes and positive Babinski sign) with absent ankle reflexes are initially associated but regress as disease stabilizes (~10 years after onset).

Dane

Klasyfikacja
Choroba

Synonimy
Autosomal recessive distal spinal muscular atrophy type 2
Autosomalny recesywny dystalny rdzeniowy zanik mięśni typu 2
dHMNJ
dHMNJ

Kod ORPHA
139552

Kod OMIM
605726

Kod ICD10
G12.2

Kod ICD11
8B61.4

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*Źródło

orphanet