

## 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  
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#### Kod ORPHA

139552

#### Kod OMIM

605726

#### Kod ICD10

G12.2

#### Kod ICD11

8B61.4

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#### [\\*Źródło](#)

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