

## Opis choroby \*

### Definicja

A rare, genetic, neuromuscular disease characterized by proximal muscle weakness with an early involvement of foot and hand muscles following normal motor development in early childhood, a rapidly progressive disease course leading to generalized areflexic tetraplegia with contractures, severe scoliosis, hyperlordosis, and progressive respiratory insufficiency leading to assisted ventilation. Cranial nerve functions are normal and tongue wasting and fasciculations are absent. Milder phenotype with a moderate generalized weakness and slower disease progress was reported.

### Dane

#### Klasyfikacja

Choroba

#### Synonimy

Autosomal recessive distal spinal muscular atrophy type 4  
Autosomalny recesywny dystalny rdzeniowy zanik mięśni typu 4  
dSMA4  
Dystalny rdzeniowy zanik mięśni typu 4  
Distal spinal muscular atrophy type 4  
dSMA4

#### Kod ORPHA

206580

#### Kod OMIM

611067

#### Kod ICD10

G12.2

#### Kod ICD11

8B61.4

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#### \*Źródło

orphanet