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

---

*Źródło

orphanet