

Opis choroby *

Definicja

Distal spinal muscular atrophy type 3 is a rare neuromuscular disease characterized by progressive muscular weakness and atrophy predominantly affecting distal parts of limbs, later involvement of proximal and trunk muscles with marked hyperlordosis and late diaphragmatic dysfunction.

Dane

Klasyfikacja

Choroba

Synonimy

Autosomal recessive distal spinal muscular atrophy type 3

Autosomalny recesywny dystalny rdzeniowy zanik mięśni typu 3

dHMN3 and dHMN4

dSMA3

Dystalna dziedziczna neuropatia ruchowa typu 3 i typu 4

Distal hereditary motor neuropathy type 3 and type 4

dHMN3 and dHMN4

dSMA3

Kod ORPHA

139547

Kod OMIM

607088

Kod ICD10

G12.2

Kod ICD11

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*Źródło

orphanet