

Opis choroby *

Definicja

A rare, slowly progressive genetic peripheral neuropathy characterized by distal atrophy and weakness affecting the upper limbs (with a predilection for the thenar eminence) and subsequently the lower limbs, associated with uni- or bilateral vocal cord paresis leading to hoarse voice and breathing difficulties, and facial weakness.

Dane

Klasyfikacja

Choroba

Synonimy

Distal spinal muscular atrophy with vocal cord paralysis
dHMN7
Dystalny rdzeniowy zanik mięśni z porażeniem strun głosowych
dHMN7

Kod ORPHA

139589

Kod OMIM

607641

Kod ICD10

G12.2

Kod ICD11

8B61.4

*Źródło

orphanet