Opis choroby *

Definicja

A rare, genetic proximal spinal muscular atrophy characterized by degeneration of alpha motor neurons in the anterior horns of the spinal cord and lower brain stem manifesting with onset of progressive proximal muscle weakness (legs greater than arms) between 18 months and adulthood. Motor development is heterogeneous but walking is typically acquired.

Dane

Klasyfikacja
Podtyp kliniczny

Synonimy
Juvenile spinal muscular atrophy
Choroba Kugelberga i Welandera
Młodzieńczy rdzeniowy zanik mięśni
SMA typu 3
SMA typu III
SMA3
SMA-III
Kugelberg-Welander disease
SMA type 3
SMA type III
SMA-III
SMA3

Kod ORPHA
83419

Kod OMIM
253400

Kod ICD10
G12.1

Kod ICD11
8B61.2

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*Źródło

orphanet