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 severe and progressive muscle weakness in the first 6 months of life and presenting with severe, generalized hypotonia and weakness,. Dysphagia and respiratory impairment may also be present at presentation or appear at a later stage. Classically, before the advent of recent therapies, type 1 patients never achieved sitting without support.

Dane

Klasyfikacja
Podtyp kliniczny

Synonimy
Infantile spinal muscular atrophy
Choroba Werdniga i Hoffmanna
Dziecięcy rdzeniowy zanik mięśni
SMA typu 1
SMA typu I
SMA1
SMA-I
Infantile-onset spinal muscular atrophy
SMA type 1
SMA type I
SMA-I
SMA1
Werdnig-Hoffmann disease

Kod ORPHA
83330

Kod OMIM
253300

Kod ICD10
G12.0

Kod ICD11
8B61.0

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*Źródło

orphanet