Opis choroby *

Definicja

A rare, genetic motor neuron disease characterized by predominantly motor axonal peripheral neuropathy manifesting with progressive scapuloperoneal muscular atrophy and weakness, laryngeal palsy, congenital absence of muscles, and, in some, skeletal abnormalities.

Dane

Klasyfikacja
Choroba

Synonimy
Neurogenic scapuloperoneal amyotrophy, New England type
Neurogenna amiotrofia łopatkowo-strzałkowa, typ N England
Neuronopatia łopatkowo-strzałkowa
SPSMA
SPSMA
Scapuloperoneal neuronopathy

Kod ORPHA
431255

Kod OMIM
181405

Kod ICD10
G12.1

Kod ICD11
8B61.4

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*Źródło

orphanet