Opis choroby *

Definicja

A rare, genetic, motor neuron disease characterized by adulthood-onset of slowly progressive, proximal muscular weakness with fasciculations, amyotrophy, cramps, and absent/hypoactive reflexes, without bulbar or pyramidal involvement.

Dane

Klasyfikacja
Choroba

Synonimy
Autosomal dominant adult-onset proximal SMA
Choroba Finkela
Autosomal dominant late-onset spinal muscular atrophy, Finkel type
Finkel disease
SMAFK

Kod ORPHA
209335

Kod OMIM
182980

Kod ICD10
G12.1

Kod ICD11
8B61.Y

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*Źródło

orphanet