Opis choroby *

Definicja

A rare, genetic, motor neuron disease characterized by slowly progressive, predominantly proximal, muscular weakness and atrophy which typically manifests with muscle cramps, fasciculations, decreased/absent deep tendon reflexes, hand tremor, and elevated serum creatine kinase at onset and later associates with reduced walking ability and impaired vibration sensation.

Dane

Klasyfikacja
Choroba

Synonimy
LOSMoN
Late-onset spinal motor neuronopathy
SMAJ
Spinal muscular atrophy, Jokela type

Kod ORPHA
276435

Kod OMIM
615048

Kod ICD10
G12.1

Kod ICD11
8B60.Y

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*Źródło

orphanet