

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

*Źródło

orphanet