Opis choroby *

Definicja

A form of hereditary spastic ataxia characterized by an onset usually in adulthood (but ranging from 10-72 years) of progressive bilateral lower limb weakness and spasticity and sometimes predominant cerebellar ataxia. In addition to frequent sphincter dysfunction and decreased vibratory sense at the ankles, manifestations may include optical neuropathy, nystagmus, blepharoptosis, ophthalmoplegia, decreased hearing, scoliosis, pes cavus, motor and sensory neuropathy, muscle atrophy, parkinsonism, and dystonia.

Dane

Klasyfikacja
Choroba

Synonimy
SPG7
SPG7

Kod ORPHA
99013

Kod OMIM
607259

Kod ICD10
G11.4

Kod ICD11
8B44.01

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*Źródło

orphanet