

Opis choroby *

Definicja

A rare, pure or complex form of hereditary spastic paraplegia usually characterized by a pure phenotype of a slowly progressive spastic paraplegia associated with urinary incontinence with an onset in mid- to late-adulthood. A complex phenotype, with the additional findings of cognitive impairment, sensorimotor polyneuropathy, ataxia, parkinsonism, and dystonia as well as thin corpus callosum and white matter lesions (seen on brain and spine magnetic resonance imaging), has also been reported.

Dane

Klasyfikacja

Choroba

Synonimy

SPG48

SPG48

Kod ORPHA

306511

Kod OMIM

613647

Kod ICD10

G11.4

Kod ICD11

-

*Źródło

orphanet