

Opis choroby *

Definicja

A rare, hereditary spastic paraplegia that can present as either a pure or complex phenotype. The pure form is characterized by lower limb spasticity, hyperreflexia and extensor plantar responses, presenting in childhood or adolescence. The complex form is characterized by the association with additional manifestations including peripheral neuropathy with upper limb muscle atrophy, moderate intellectual disability and parkinsonism. Deafness and retinitis pigmentosa have also been reported.

Dane

Klasyfikacja

Choroba

Synonimy

SPG10

SPG10

Kod ORPHA

100991

Kod OMIM

604187

Kod ICD10

G11.4

Kod ICD11

8B44.00

*Źródło

orphanet