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

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*Źródło

orphanet