Opis choroby *

Definicja

A rare, pure or complex form of hereditary spastic paraplegia typically characterized by presentation in late adolescence or early adulthood as a pure phenotype of lower limb spasticity with hyperreflexia and extensor plantar responses, as well as mild bladder disturbances and pes cavus. Rarely, it can present as a complex phenotype with additional manifestations including epilepsy, variable peripheral neuropathy and/or memory impairment.

Dane

Klasyfikacja
Choroba

Synonimy
SPG6
Autosomalna dominująca rodzinna paraplegia spastyczna typu 3
SPG6

Kod ORPHA
100988

Kod OMIM
600363

Kod ICD10
G11.4

Kod ICD11
8B44.00

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*Źródło

orphanet