Opis choroby *

Definicja

A rare type of hereditary spastic paraplegia usually characterized by a pure phenotype of proximal weakness of the lower extremities with spastic gait and brisk reflexes, with a bimodal age of onset of either childhood or adulthood (>30 years). In some cases, it can present as a complex phenotype with additional associated manifestations including peripheral neuropathy, bulbar palsy (with dysarthria and dysphagia), distal amyotrophy, and impaired distal vibration sense.

Dane

Klasyfikacja
Choroba

Synonimy
SPG31
SPG31

Kod ORPHA
101011

Kod OMIM
610250

Kod ICD10
G11.4

Kod ICD11
8B44.00

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*Źródło

orphanet