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

*Źródło

orphanet