

Autosomalna recesywna paraplegia spastyczna typu 31

Kod Orpha: 101011 Kod OMIM: 610250

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

Rozszerzony opis choroby

Brak opisu rozszerzonego dla tej choroby. Opracowanie w toku.

Dostępna na stronie www.orphanet.pl