Opis choroby *

Definicja

A rare, pure or complex form of hereditary spastic paraplegia characterized by either a pure spastic paraplegia phenotype, usually presenting in the first or second decade of life, with spastic lower extremities, unsteady spastic gait, hyperreflexia and extensor plantar responses, or as a complicated phenotype with the additional manifestations of distal wasting, saccadic ocular movements, mild cerebellar ataxia and mild, distal, axonal neuropathy.

Dane

Klasyfikacja
Choroba

Synonimy
SPG30
SPG30

Kod ORPHA
101010

Kod OMIM
610357

Kod ICD10
G11.4

Kod ICD11
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*Źródło

orphanet