Opis choroby *

Definicja

A rare, pure or complex form of hereditary spastic paraplegia, with variable phenotype, typically characterized by childhood-onset of minimally progressive, bilateral, mainly symmetric lower limb spasticity and weakness, associated with pes cavus, scoliosis, sphincter disturbances and/or urinary bladder hyperactivity. Rare additional associated manifestations may include mild intellectual disability, axonal motor neuropathy, and seizures.

Dane

Klasyfikacja
Choroba

Synonimy
Strümpell disease
Choroba Strümpella

Kod ORPHA
100984

Kod OMIM
182600

Kod ICD10
G11.4

Kod ICD11
8B44.00

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*Źródło

orphanet