Opis choroby *

Definicja

A rare form of hereditary spastic paraplegia with high intrafamilial clinical variability, characterized in most cases as a pure phenotype with an adult onset (mainly the 3rd to 5th decade of life, but that can present at any age) of progressive gait impairment due to bilateral lower-limb spasticity and weakness as well as very mild proximal weakness and urinary urgency. In some cases, a complex phenotype is also reported with additional manifestations including cognitive impairment, cerebellar ataxia, epilepsy and neuropathy. A faster disease progression is noted in patients with a later age of onset.

Dane

Klasyfikacja
Choroba

Synonimy
SPG4
SPG4

Kod ORPHA
100985

Kod OMIM
182601

Kod ICD10
G11.4

Kod ICD11
8B44.00

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*Źródło

orphanet