Opis choroby *

Definicja

Autosomal recessive spastic paraplegia type 5A is a form of hereditary spastic paraplegia characterized by either a pure phenotype of slowly progressive spastic paraplegia of the lower extremities with bladder dysfunction and pes cavus or a complex presentation with additional manifestations including cerebellar signs, nystagmus, distal or generalized muscle atrophy and cognitive impairment. Age of onset is highly variable, ranging from early childhood to adulthood. White matter hyperintensity and cerebellar and spinal cord atrophy may be noted, on brain magnetic resonance imaging, in some patients.

Dane

Klasyfikacja
Choroba

Synonimy
SPG5A
SPG5A

Kod ORPHA
100986

Kod OMIM
270800

Kod ICD10
G11.4

Kod ICD11
8B44.01

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*Źródło

orphanet