Opis choroby *

Definicja

Autosomal recessive spastic paraplegia type 44 (SPG44) is a very rare, complex form of hereditary spastic paraplegia characterized by a late-onset, slowly progressive spastic paraplegia associated with mild ataxia and dysarthria, upper extremity involvement (i.e. loss of finger dexterity, dysmetria), and mild cognitive impairment, without the presence of nystagmus. A hypomyelinating leukodystrophy and thin corpus callosum is observed in all cases and psychomotor development is normal or near normal. SPG44 is caused by mutations in the GJC2 gene (1q41-q42) encoding the gap junction gamma-2 protein.

Dane

Klasyfikacja
Choroba

Synonimy
SPG44
SPG44

Kod ORPHA
320401

Kod OMIM
613206

Kod ICD10
G11.4

Kod ICD11
8B44.01

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*Źródło

orphanet