

## 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