

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

*Źródło

orphanet