Opis choroby *

Definicja

Autosomal recessive spastic paraplegia type 26 (SPG26) is a rare, complex type of hereditary spastic paraplegia characterized by the onset in childhood/adolescence (ages 2-19) of progressive spastic paraplegia associated mainly with mild to moderate cognitive impairment and developmental delay, cerebellar ataxia, dysarthria, and peripheral neuropathy. Less commonly reported manifestations include skeletal abnormalities (i.e. pes cavus, scoliosis), dyskinesia, dystonia, cataracts, cerebellar signs (i.e. saccadic dysfunction, nystagmus, dysmetria), bladder disturbances, and behavioral problems. SPG26 is caused by mutations in the B4GALNT1 gene (12q13.3), encoding Beta-1, 4 N-acetylgalactosaminyltransferase 1.

Dane

Klasyfikacja
Choroba

Synonimy
GM2 synthase deficiency
Niedobór syntazy GM2
SPG26
SPG26

Kod ORPHA
101006

Kod OMIM
609195

Kod ICD10
G11.4

Kod ICD11
8B44.01

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*Źródło

orphanet