Opis choroby *

Definicja

Autosomal recessive spastic paraplegia type 32 (SPG32) is a rare, complex type of hereditary spastic paraplegia characterized by a slowly progressive spastic paraplegia (with walking difficulties appearing at onset at 6-7 years of age) associated with mild intellectual disability. Brain imaging reveals thin corpus callosum, cortical and cerebellar atrophy, and pontine dysraphia. The SPG32 phenotype has been mapped to a locus on chromosome 14q12-q21.

Dane

Klasyfikacja
Choroba

Synonimy
SPG32
SPG32

Kod ORPHA
171622

Kod OMIM
611252

Kod ICD10
G11.4

Kod ICD11
8B44.01

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*Źródło

orphanet