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 Synonimy Choroba SPG32 SPG32

 Kod ORPHA
 Kod OMIM
 Kod ICD10

 171622
 611252
 G11.4

Kod ICD11 8B44.01

*Źródło

orphanet