Autosomalna recesywna paraplegia spastyczna typu 46
Kod Orpha: 320391 Kod OMIM: 614409

Opis choroby *

Definicja
Autosomal recessive spastic paraplegia type 46 (SPG46) is a rare, complex type of hereditary spastic paraplegia characterized by an onset, in infancy or childhood, of the typical signs of spastic paraplegia (i.e. spastic gait and weakness of the lower limbs) associated with a variety of additional manifestations including upper limb spasticity and weakness, pseudobulbar dysarthria, bladder dysfunction, cerebellar ataxia, cataracts, and cognitive impairment that can progress to dementia. Brain imaging may show thinning of the corpus callosum and mild atrophy of the cerebrum and cerebellum. SPG46 is due to mutations in the GBA2 gene (9p13.2) encoding non-lysosomal glucosylceramidase.

Dane

Klasyfikacja
Choroba

Synonimy
SPG46
SPG46

Kod ORPHA
320391

Kod OMIM
614409

Kod ICD10
G11.4

Kod ICD11
8B44.01

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*Źródło

orphanet

Rozszerzony opis choroby

Brak opisu rozszerzonego dla tej choroby. Opracowanie w toku.

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