

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

*Źródło

orphanet