

Opis choroby *

Definicja

Autosomal recessive spastic paraplegia type 21 is a complex type of hereditary spastic paraplegia characterized by an onset in adolescence or adulthood of slowly progressive spastic paraparesis associated with the additional manifestations of apraxia, cognitive and speech decline (leading to dementia and akinetic mutism in some cases), personality disturbances and extrapyramidal (e.g. oromandibular dyskinesia, rigidity) and cerebellar (i.e. dysdiadochokinesia and incoordination) signs. Subtle abnormalities (e.g. developmental delays) may be noted earlier in childhood. A thin corpus callosum and white matter abnormalities are equally reported on magnetic resonance imaging.

Dane

Klasyfikacja

Choroba

Synonimy

Mast syndrome

SPG21

Zespół Mast

SPG21

Kod ORPHA

101001

Kod OMIM

248900

Kod ICD10

G11.4

Kod ICD11

8B44.01

*Źródło

orphanet