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

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*Źródło

orphanet