DISEASE:
Autosomal recessive spastic paraplegia type 21

NAME:	Autosomal recessive spastic paraplegia type 21
DESCRIPTION:	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.
ORPHACODE:	101001
SYNONYMS:	Mast syndrome SPG21
XREF(S):	Orphanet OMIM ICD-10
ANALYTE(S):	SPG21
CREATED:	13 May 2019 - 01:02
CHANGED:	22 Jun 2023 - 16:14

RELATED CONTENT

Related Genetic Tests

- Hereditary Spastic Paraplegia (gene panel)
- Spastic Paraplegia (gene panel)

Related Laboratories

- Centre de Génétique-Institut de Pathologie et de Génétique (IPG)
- Centrum Medische Genetica - UZ Antwerpen

Related Analytes

- SPG21 abhydrolase domain containing, maspardin

Related Gene Panels

- Hereditary Spastic Paraplegia & ataxia (genepanel) - UZA
- Spastic Paraplegia (89 genes) - IPG