

DISEASE:
Autosomal dominant spastic paraplegia type 6

NAME:	Autosomal dominant spastic paraplegia type 6
DESCRIPTION:	A rare, pure or complex form of hereditary spastic paraplegia typically characterized by presentation in late adolescence or early adulthood as a pure phenotype of lower limb spasticity with hyperreflexia and extensor plantar responses, as well as mild bladder disturbances and pes cavus. Rarely, it can present as a complex phenotype with additional manifestations including epilepsy, variable peripheral neuropathy and/or memory impairment.
ORPHACODE:	100988
SYNONYMS:	SPG6
XREF(S):	Orphanet OMIM MeSH ICD-10
ANALYTE(S):	NIPA1
CREATED:	13 May 2019 - 01:02
CHANGED:	22 Jun 2023 - 16:14

RELATED CONTENT

Related Genetic Tests

- Spastic Paraplegia (gene panel)

Related Laboratories

- Centre de Génétique-Institut de Pathologie et de Génétique (IPG)

Related Analytes

- NIPA magnesium transporter 1

Related Gene Panels

- Spastic Paraplegia (89 genes) - IPG

Source URL: <http://gentest.healthdata.be/disease/3716>