DISEASE:
Autosomal recessive spastic paraplegia type 53

NAME:	Autosomal recessive spastic paraplegia type 53
DESCRIPTION:	Autosomal recessive spastic paraplegia type 53 (SPG53) is a very rare, complex type of hereditary spastic paraplegia characterized by early-onset spastic paraplegia (with spasticity in the lower extremities that progresses to the upper extremities) associated with developmental and motor delay, mild to moderate cognitive and speech delay, skeletal dysmorphism (e.g. kyphosis and pectus), hypertrichosis and mildly impaired vibration sense. SPG53 is due to mutations in the VPS37A gene (8p22) encoding vacuolar protein sorting-associated protein 37A.
ORPHACODE:	319199
SYNONYMS:	SPG53
XREF(S):	Orphanet ICD-10 OMIM
ANALYTE(S):	VPS37A
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RELATED CONTENT

Related Genetic Tests

- Spastic Paraplegia (gene panel)

Related Laboratories

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

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- VPS37A subunit of ESCRT-I

Related Gene Panels

- Spastic Paraplegia (89 genes) - IPG

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