DISEASE:
Autosomal spastic paraplegia type 18

NAME:	Autosomal spastic paraplegia type 18
DESCRIPTION:	Autosomal spastic paraplegia type 18 (SPG18) is a rare, complex type of hereditary spastic paraplegia characterized by progressive spastic paraplegia (presenting in early childhood) associated with delayed motor development, severe intellectual disability and joint contractures. A thin corpus callosum is equally noted on brain magnetic resonance imaging. SPG18 is caused by a mutation in the ERLIN2 gene (8p11.2) encoding the protein, Erlin-2.
ORPHACODE:	209951
SYNONYMS:	SPG18
XREF(S):	Orphanet ICD-10 OMIM
ANALYTE(S):	ERLIN2
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

- ER lipid raft associated 2

Related Gene Panels

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