DISEASE:
Autosomal recessive spastic paraplegia type 26

NAME:	Autosomal recessive spastic paraplegia type 26
DESCRIPTION:	Autosomal recessive spastic paraplegia type 26 (SPG26) is a rare, complex type of hereditary spastic paraplegia characterized by the onset in childhood/adolescence (ages 2-19) of progressive spastic paraplegia associated mainly with mild to moderate cognitive impairment and developmental delay, cerebellar ataxia, dysarthria, and peripheral neuropathy. Less commonly reported manifestations include skeletal abnormalities (i.e. pes cavus, scoliosis), dyskinesia, dystonia, cataracts, cerebellar signs (i.e. saccadic dysfunction, nystagmus, dysmetria), bladder disturbances, and behavioral problems. SPG26 is caused by mutations in the B4GALNT1 gene (12q13.3), encoding Beta-1, 4 N-acetylgalactosaminyltransferase 1.
ORPHACODE:	101006
SYNONYMS:	GM2 synthase deficiency SPG26
XREF(S):	Orphanet ICD-10 MeSH OMIM
ANALYTE(S):	B4GALNT1
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- Centre de Génétique-Institut de Pathologie et de Génétique (IPG)
- Centrum Medische Genetica - UZ Antwerpen

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- beta-1,4-N-acetyl-galactosaminyltransferase 1

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