DISEASE:
Autosomal recessive spastic paraplegia type 46

NAME:	Autosomal recessive spastic paraplegia type 46
DESCRIPTION:	Autosomal recessive spastic paraplegia type 46 (SPG46) is a rare, complex type of hereditary spastic paraplegia characterized by an onset, in infancy or childhood, of the typical signs of spastic paraplegia (i.e. spastic gait and weakness of the lower limbs) associated with a variety of additional manifestations including upper limb spasticity and weakness, pseudobulbar dysarthria, bladder dysfunction, cerebellar ataxia, cataracts, and cognitive impairment that can progress to dementia. Brain imaging may show thinning of the corpus callosum and mild atrophy of the cerebrum and cerebellum. SPG46 is due to mutations in the GBA2 gene (9p13.2) encoding non-lysosomal glucosylceramidase.
ORPHACODE:	320391
SYNONYMS:	SPG46
XREF(S):	Orphanet OMIM ICD-10
ANALYTE(S):	GBA2
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