DISEASE:
Autosomal recessive spastic paraplegia type 39

NAME:	Autosomal recessive spastic paraplegia type 39
DESCRIPTION:	A rare autosomal recessive complex spastic paraplegia characterized by upper motor neuron involvement and peripheral neuropathy with an onset between childhood and early adulthood. Patients present with progressive spasticity, hyperreflexia, and distal upper and lower muscle wasting. Reduced cognitive functioning and cerebellar ataxia have also been reported. MR imaging may reveal cerebellar and/or spinal cord atrophy.
ORPHACODE:	139480
SYNONYMS:	SPG39 Spastic paraplegia due to NTE mutation Spastic paraplegia due to neuropathy target esterase mutation
XREF(S):	Orphanet ICD-10 OMIM
ANALYTE(S):	PNPLA6
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- Centre de Génétique-Institut de Pathologie et de Génétique (IPG)
- Centrum Medische Genetica - UZ Antwerpen

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