DISEASE:
Autosomal recessive spastic paraplegia type 55

NAME:	Autosomal recessive spastic paraplegia type 55
DESCRIPTION:	Autosomal recessive spastic paraplegia type 55 (SPG 55) is a rare, complex type of hereditary spastic paraplegia characterized by childhood onset of progressive spastic paraplegia associated with optic atrophy (with reduced visual acuity and central scotoma), ophthalmoplegia, reduced upper-extremity strength and dexterity, muscular atrophy in the lower extremities, and sensorimotor neuropathy. SPG55 is caused by mutations in the C12ORF65 gene (12q24.31) encoding probable peptide chain release factor C12orf65, mitochondrial.
ORPHACODE:	320375
SYNONYMS:	SPG55
XREF(S):	Orphanet ICD-10 OMIM
ANALYTE(S):	MTRFR
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RELATED CONTENT

Related Genetic Tests

- Charcot-Marie-Tooth (other than type 1A) (gene panel, IPN panel)
- Spastic Paraplegia (gene panel)

Related Laboratories

- Centre de Génétique-Institut de Pathologie et de Génétique (IPG)
- Centrum Menselijke Erfelijkheid - KUL

Related Analytes

- mitochondrial translation release factor in rescue

Related Gene Panels

- Inherited Peripheral Neuropathies gene panel (139 genes) - KUL
- Spastic Paraplegia (89 genes) - IPG