DISEASE:
Autosomal recessive spastic paraplegia type 64

NAME:	Autosomal recessive spastic paraplegia type 64
DESCRIPTION:	Autosomal recessive spastic paraplegia type 64 is an extremely rare and complex form of hereditary spastic paraplegia (see this term), reported in only 4 patients from 2 families to date, characterized by spastic paraplegia (presenting between the ages of 1 to 4 years with abnormal gait) associated with microcephaly, amyotrophy, cerebellar signs (e.g. dysarthria) aggressiveness, delayed puberty and mild to moderate intellectual disability. SPG64 is due to mutations in the ENTPD1 gene (10q24.1), encoding ectonucleoside triphosphate diphosphohydrolase 1.
ORPHACODE:	401810
SYNONYMS:	SPG64
XREF(S):	Orphanet ICD-10 OMIM
ANALYTE(S):	ENTPD1
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RELATED CONTENT

Related Genetic Tests

- Spastic Paraplegia (gene panel)

Related Laboratories

- Centre de Génétique-Institut de Pathologie et de Génétique (IPG)

Related Analytes

- ectonucleoside triphosphate diphosphohydrolase 1

Related Gene Panels

- Spastic Paraplegia (89 genes) - IPG