DISEASE:
Severe intellectual disability and progressive spastic paraplegia

NAME:	Severe intellectual disability and progressive spastic paraplegia
DESCRIPTION:	Severe intellectual disability and progressive spastic paraplegia is a rare complex spastic paraplegia characterized by an early onset hypotonia that progresses to spasticity, global developmental delay, severe intellectual disability and speech impairment, microcephaly, short stature and dysmorphic features. Patients often become non-ambulatory, and some develop seizures and stereotypic laughter.
ORPHACODE:	280763
SYNONYMS:	AP4 deficiency syndrome
XREF(S):	Orphanet ICD-10 OMIM OMIM OMIM OMIM
ANALYTE(S):	AP4M1 AP4E1 AP4B1 AP4S1
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RELATED CONTENT

Related Genetic Tests

- Hereditary Spastic Paraplegia (gene panel)
- Neurodegeneration with Brain Iron Accumulation (gene panel)
- Spastic Paraplegia (gene panel)

Related Laboratories

- Centre de Génétique-Institut de Pathologie et de Génétique (IPG)
- Centrum Medische Genetica - UZ Antwerpen
- Centrum Medische Genetica - UZ Gent

Related Analytes

- adaptor related protein complex 4 subunit beta 1
- adaptor related protein complex 4 subunit epsilon 1
- adaptor related protein complex 4 subunit mu 1
- adaptor related protein complex 4 subunit sigma 1

Related Gene Panels

- Hereditary Spastic Paraplegia & ataxia (genepanel) - UZA
- Neurodegeneration with Brain Iron Accumulation (NBIA) - UGent
- Spastic Paraplegia (89 genes) - IPG
