DISEASE:
Autosomal dominant spastic paraplegia type 9A

NAME:	Autosomal dominant spastic paraplegia type 9A
DESCRIPTION:	A rare complex hereditary spastic paraplegia characterized by juvenile to adult onset of slowly progressive spasticity mainly affecting the lower limbs, associated with spastic dysarthria and motor neuropathy. Additional manifestations include congenital bilateral cataract, gastroesophageal reflux, persistent vomiting, mild cerebellar signs, pes cavus, and occasionally short stature, among others.
ORPHACODE:	447753
SYNONYMS:	AD-SPG9A Cataracts-motor neuropathy-short stature-skeletal anomalies syndrome Spastic paraparesis-amyopathy-cataracts-gastroesophageal reflux syndrome
XREF(S):	Orphanet ICD-10 OMIM
ANALYTE(S):	ALDH18A1 FAR1
CREATED:	13 May 2019 - 01:02
CHANGED:	22 Jun 2023 - 16:14

Source URL: http://gentest.healthdata.be/disease/2489

RELATED CONTENT

Related Genetic Tests

- Cataract (gene panel)
- Hereditary Spastic Paraplegia (gene panel)

Related Laboratories

- Centrum Medische Genetica - UZ Antwerpen
- Centrum Medische Genetica - UZ Gent

Related Analytes

- aldehyde dehydrogenase 18 family member A1
- fatty acyl-CoA reductase 1

Related Gene Panels

- Cataract - UGent
- Hereditary Spastic Paraplegia & ataxia (genepanel) - UZA
- test