DISEASE:
Spinocerebellar ataxia with axonal neuropathy type 2

NAME:	Spinocerebellar ataxia with axonal neuropathy type 2
DESCRIPTION:	A rare autosomal recessive cerebellar ataxia (ARCA), characterized by progressive cerebellar ataxia associated with frequent oculomotor apraxia, severe neuropathy and an elevated serum alpha-fetoprotein (AFP) level.
ORPHACODE:	64753
SYNONYMS:	AOA2 Ataxia-oculomotor apraxia type 2 SCAN 2 SCAR1
XREF(S):	Orphanet OMIM OMIM ICD-10
ANALYTE(S):	SETX PIK3R5
CREATED:	13 May 2019 - 01:02
CHANGED:	22 Jun 2023 - 16:14

RELATED CONTENT

Related Genetic Tests

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

Related Laboratories

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

Related Analytes

- phosphoinositide-3-kinase regulatory subunit 5
- senataxin

Related Gene Panels

- Hereditary Spastic Paraplegia & ataxia (genepanel) - UZA
- Neurodegeneration (99 genes) - IPG
- Spastic Paraplegia (89 genes) - IPG