DISEASE:
Spinocerebellar ataxia type 7

NAME:	Spinocerebellar ataxia type 7
DESCRIPTION:	An autosomal dominant cerebellar ataxia type II that is characterized by progressive ataxia, motor system abnormalities, dysarthria, dysphagia and retinal degeneration leading to progressive blindness.
ORPHACODE:	94147
SYNONYMS:	Ataxia with pigmentary retinopathy Cerebellar syndrome-pigmentary maculopathy syndrome SCA7
XREF(S):	Orphanet OMIM ICD-10
ANALYTE(S):	ATXN7
CREATED:	13 May 2019 - 01:02
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RELATED CONTENT

Related Genetic Tests

- Spinocerebellar ataxia (SCA) types 1, 2, 3, 6, 7 - CAG repeat expansion
- Spinocerebellar ataxia (types 1, 2, 3, 6, 7) - CAG repeat expansion
- Spinocerebellar ataxia (types 1, 2, 3, 6, 7) - CAG repeat expansion
- Spinocerebellar ataxia (types 1, 2, 3, 6, 7, 17) - CAG repeat expansion

Related Laboratories

- Centre de Génétique-Institut de Pathologie et de Génétique (IPG)
- Centrum Medische Genetica - UZ Antwerpen
- Centrum Medische Genetica - UZ Brussel VUB
- Centrum Menselijke Erfelijkheid - KUL

Related Analytes

- ataxin 7

Related Gene Panels

- Spinocerebellar ataxia (types 1, 2, 3, 6, 7) (5 genes) - KUL