DISEASE:
Spinocerebellar ataxia type 12

NAME:	Spinocerebellar ataxia type 12
DESCRIPTION:	Spinocerebellar ataxia type 12 (SCA12) is a very rare subtype of type I autosomal dominant cerebellar ataxia (ADCA type I; see this term). It is characterized by the presence of action tremor associated with relatively mild cerebellar ataxia. Associated pyramidal and extrapyramidal signs and dementia have been reported.
ORPHACODE:	98762
SYNONYMS:	SCA12
XREF(S):	Orphanet ICD-10 OMIM
ANALYTE(S):	PPP2R2B
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RELATED CONTENT

Related Genetic Tests

- Spinocerebellar ataxia (SCA) types 8, 10, 12, 17 - repeat expansion
- Spinocerebellar ataxia (type 8, 17) + Dentatorubral pallidoluysian atrophy - repeat expansion

Related Laboratories

- Centrum Medische Genetica - UZ Antwerpen
- Centrum Medische Genetica - UZ Brussel VUB

Related Analytes

- protein phosphatase 2 regulatory subunit Bbeta

Related Gene Panels

- Spinocerebellar ataxia (type 8, 10, 12, 17) (4 genes) - UZA
- Spinocerebellar ataxia (type 8, 17 + ATN1) (5 genes) - VUB