Spinocerebellar Ataxia Type 1 / 2 / 3 / 6 / 7 / 17 (SCA1 / 2 / 3 / 6 / 7 / 17) – OMIM 164400 / 183090 / 109150 / 183086 / 164500 / 607136

Background

Ataxia means 'absence of order'; parts of the nervous system that normally control co-ordination and balance are affected. Ataxia is the principal symptom of a group of neurological disorders called the cerebellar ataxias, most of which are progressive. The spinocerebellar ataxias (SCAs) are autosomal dominant so only one faulty gene is required from one parent to cause the condition. They are characterised by slowly progressive incoordination of gait and often associated with poor coordination of hands, speech and eye movement. About 30 different genetic loci have been mapped for autosomal dominant cerebellar ataxia (ADCAs) and genes have been characterized for about half of these. Most commonly offered diagnostic tests are for SCA types 1, 2, 3, 6, 7 & 17. For these ataxias, different gene mutations cause different types of SCAs - the symptoms are very similar but the age of onset and the rate of progression vary for each type. Each of these is a polyglutamine disease caused by expansion of a CAG repeat in the coding region of the genes involved.

Recommended Clinical Referral Criteria

- SCA affects mainly the central nervous system; patients exhibit a cerebellar syndrome, including gait and limb ataxia, limb dysarthria, and impaired eye movements.
- Autosomal dominant inheritance

Molecular Analysis

Mutation screen: Fragment length analysis of the [CAG]n repeat expansion of the following genes by PCR and triplet primed PCR where appropriate - *ATXN1* (SCA1), *ATXN2* (SCA2), *ATXN3* (SCA3), *CACNA1A* (SCA6), *ATXN7* (SCA7), *TBP* (SCA17); each test detects >99% of affected individuals.
Family follow-up: Testing for known familial mutations in the SCA genes (listed above)

Test (Price available on request)	TAT (calendar days)
SCA1, 2, 3, 6, 7 and/or 17 gene expansion analysis	42
Predictive single gene analysis (any of the above)	28

Contact Details

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Sample Requirements

Blood – 5ml in EDTA (1ml neonates/infants);
Please contact lab prior to sending a prenatal sample.
Please label samples with three identifiers and date of collection.
All samples must be accompanied by request form
Consent for testing & DNA storage is assumed when request for test received

Links

Orphanet
http://www.orpha.net/
OMIM
http://www.omim.org/
National Genomic Test Directory
https://www.england.nhs.uk/wp-content/uploads/2018/08/rare-and-inherited-disease-eligibility-criteria-v4.pdf

Support
Ataxia UK
http://www.ataxia.org.uk/