

Spinocerebellar Ataxia types 1, 2, 3, 6, 7 DNA Tests

Background

The dominant cerebellar ataxias are a clinically and genetically heterogeneous group of disorders that cannot be differentiated reliably from each other on a clinical basis. DNA testing is highly sensitive and specific and provides a definitive diagnosis for an estimated 50-60% of Caucasian patients with findings of dominant cerebellar ataxia. Patient DNA is analyzed to determine the number of CAG trinucleotide repeats located within each allele of the five causative genes; an abnormally large number of CAG repeats is diagnostic for the disease.

Disorder Name	Estimated % of Dominant Cerebellar Ataxias	Gene Name
Spinocerebellar ataxia 1	5 -15	<i>ATXN1</i>
Spinocerebellar ataxia 2	10-15	<i>ATXN2</i>
Spinocerebellar ataxia 3	20-30	<i>ATXN3</i>
Spinocerebellar ataxia 6	1-15	<i>CACNA1A</i>
Spinocerebellar ataxia 7	1-5	<i>ATXN7</i>

Notes: Frequencies of SCAs vary significantly in the Japanese and perhaps in other populations. Abnormal-sized *ATXN7* alleles have been detected in ataxia patients without retinal degeneration. A DNA test for Friedreich ataxia, an autosomal recessive disorder, can be ordered separately.

Indications for Testing

- Symptomatic testing for patients with ataxia and a family history of ataxia
- Differential diagnosis for isolated cases of unexplained progressive ataxia (expect a relatively low positive rate)
- Presymptomatic testing for at-risk family members with appropriate genetic counseling
- Prenatal testing for an at-risk fetus

Ordering

Two categories of tests are available:

- **Spinocerebellar Ataxia Panel** includes testing for all five types of SCA.
- **Spinocerebellar Ataxia** single test is offered to test for a single specific type of SCA.

1. Obtain blood sample.
2. Fill out a Clinical Lab Request - Genetics for each patient.
(available at <http://depts.washington.edu/labweb/Divisions/MolDiag/MolDiagGen/index.htm>).
-Request either the "Spinocerebellar Ataxia Panel" or Spinocerebellar Ataxia, single test.
3. Provide items needed for test interpretation:
 - Reason for testing (e.g., confirm diagnosis; rule out)
 - Clinical history and pedigree / family information
4. Call Laboratory Medicine Community Services (206) 598-6066 to arrange the best method of shipment.

Sample Requirements and Specimen Handling

Whole blood - EDTA (purple top) - 5 mL.

Samples should be received within 72 hours of collection. Samples may be refrigerated until shipped.

For prenatal diagnosis specimens, consult the laboratory.

Heparin (green top) tubes are not acceptable.

Test Frequency and Reporting

Test results usually within 1-2 weeks of specimen receipt. A written interpretative report is provided.

References

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