MAYO CLINIC LABORATORIES

NEW TEST

Notification Date: October 4, 2021
Effective Date: November 2, 2021

Spinocerebellar Ataxia Type 1, 2, 3, 6, or 7, Repeat Expansion Analysis, Varies

Test ID: SCARA

Useful for:

Molecular confirmation of clinically suspected spinocerebellar ataxia type 1, 2, 3, 6, or 7

Reflex Tests:

Test ID	Reporting Name	Available Separately	Always Performed
CULFB	Fibroblast Culture for Genetic Test	Yes	No
CULAF	Amniotic Fluid Culture/Genetic Test	Yes	No
MATCC	Maternal Cell Contamination, B	Yes	No
G204	ATXN1 (SCA 1) Gene Analysis	No (Bill Only)	No
G205	ATXN2 (SCA 2) Gene Analysis	No (Bill Only)	No
G206	ATXN3 (SCA 3) Gene Analysis	No (Bill Only)	No
G207	ATXN7 (SCA 7) Gene Analysis	No (Bill Only)	No
G208	CACNA1A (SCA 6) Gene Analysis	No (Bill Only)	No

Methods:

Polymerase Chain Reaction (PCR)

Reference Values:

- **SPINOCEREBELLAR ATAXIA TYPE 1**

Normal alleles: <36 CAG repeats
Normal alleles with CAT interruptions: 36-43 repeats
Intermediate alleles without CAT interruptions: 36-37 repeats
Uncertain significance: 38 repeats
Expanded alleles: >38 CAG repeats

- **SPINOCEREBELLAR ATAXIA TYPE 2**

Normal alleles: <32 repeats
Uncertain significance: 31 homozygous and 32 repeats
Reduced penetrance: 33-34 repeats
Expanded alleles: >34 repeats

- **SPINOCEREBELLAR ATAXIA TYPE 3**

Normal alleles: <45 repeats
Intermediate alleles: 45-59 repeats
Expanded alleles: >59 repeats

- **SPINOCEREBELLAR ATAXIA TYPE 6**

Normal alleles: <19 repeats
Intermediate alleles: 19 heterozygous repeats

Uncertain significance: 19 homozygous repeats
Expanded alleles: >19 repeats

- **SPINOCEREBELLAR ATAXIA TYPE 7**

Normal alleles: <19 repeats
Uncertain significance: 19-27 repeats
Intermediate alleles: 28-33 repeats
Reduced penetrance: 34-36 repeats
Expanded alleles: >36 repeats

***An interpretive report will be provided.**

Specimen Requirements:

Patient Preparation: A previous bone marrow transplant from an allogenic donor will interfere with testing. Call 800-533-1710 for instructions for testing patients who have received a bone marrow transplant.

Submit only 1 of the following specimens:

Specimen Type: Whole blood
Preferred: Lavender top (EDTA) or yellow top (ACD)
Acceptable: Any anticoagulant
Specimen Volume: 3 mL
Collection Instructions:
1. Invert several times to mix blood.
2. Send specimen in original tube. **Do not** aliquot.

Specimen Stability Information: Ambient (preferred) 96 hours/Refrigerated
Minimum Volume: 0.5 mL

Due to the complexity of prenatal testing, consultation with the laboratory is required for all prenatal testing. Prenatal specimens can be sent Monday through Thursday and **must be received by 5 p.m. CST on Friday** in order to be processed appropriately. All prenatal specimens must be accompanied by a maternal blood specimen. Order MATCC / Maternal Cell Contamination, Molecular Analysis, Varies on the maternal specimen.

Specimen Type: Amniotic fluid
Container/Tube: Amniotic fluid container
Specimen Volume: 20 mL
Specimen Stability Information: Refrigerated (preferred)/Ambient

Specimen Type: Chorionic villi
Container/Tube: 15-mL tube containing 15 mL of transport media
Specimen Volume: 20 mg
Specimen Stability Information: Refrigerated

Specimen Type: Confluent cultured cells
Container/Tube: T-25 flask
Specimen Volume: 2 Flasks
Collection Instructions: Submit confluent cultured cells from another laboratory.
Specimen Stability Information: Ambient (preferred)/Refrigerated

Specimen Stability Information:

Specimen Type	Temperature	Time
Varies	Varies	

Cautions:

- For predictive testing, it is important to first document the presence of a cytosine-adenine-guanine (CAG)-repeat expansion in an affected family member to confirm that the repeat expansion is the underlying mechanism of disease in the family.
- It is strongly recommended that patients undergoing predictive testing receive genetic counseling both prior to testing and after results are available.
- Test results should be interpreted in the context of clinical findings, family history, and other laboratory data. Errors in the interpretation of results may occur if information given is inaccurate or incomplete.
- Due to somatic mosaicism, repeat size identified in the peripheral blood specimen may not reflect the repeat size in untested tissues (eg, central nervous system). In addition, a negative result does not rule out the presence of a variant in the mosaic state that may be present but below the limit of detection of this assay (approximately 10%).
- Rare sequence variants immediately downstream of the spinocerebellar ataxia (SCA) repeat regions may interfere with genotype results but are not expected to affect repeat-primed peaks.
- Rare undocumented alterations (ie, polymorphisms) in polymerase-chain reaction primer binding regions may lead to false-negative results.

CPT Code:

88233-Fibroblast Culture (if appropriate)
88235-Amniotic Fluid Culture (if appropriate)
88240-Cryopreservation (if appropriate)
81265-Maternal Cell Contamination (if appropriate)
81178 (if appropriate)
81179 (if appropriate)
81180 (if appropriate)
81181 (if appropriate)
81184 (if appropriate)

Day(s) Performed: Monday & Wednesday **Report Available:** 5 to 11 days

Note:

The following referral test codes will become obsolete:

Test Name	Test ID	Referral Lab Code	Referral Lab
SCA1 (ATXN1) Repeat Expansion	FSCA1	371	Athena Diagnostics
SCA3 (MJC/ATXN3) Repeat Expansion	FSCA3	105	Athena Diagnostics
SCA 6 (CACNA1A) Repeat Expansion	FSCA6	373	Athena Diagnostics
SCA7 (ATXN7) Repeat Expansion	ZW127	677	Athena Diagnostics

Questions

Contact Michelle Raths, Laboratory Technologist Resource Coordinator at 800-533-1710.