MAYO CLINIC LABORATORIES

# Test Definition: SCARA

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

**Reporting Title:** SCA 1,2,3,6, or 7 Repeat Analysis
**Performing Location:** Mayo Clinic Laboratories - Rochester Main Campus

## Ordering Guidance:

This test is **not a gene panel** for all types of spinocerebellar ataxia (SCA). If individual findings are not specific for one type of SCA, panel analysis is available and includes testing for SCA1, 2, 3, 6, and 7; order SCAP / Spinocerebellar Ataxia Repeat Expansion Panel, Varies.

This test and SCAP should not be ordered concurrently.

## Shipping Instructions:

Specimen preferred to arrive within 96 hours of collection.

## Necessary Information:

**The type of spinocerebellar ataxia (SCA) to be assessed (SCA1, 2, 3, 6, or 7) is required**. This information must be provided for testing to be performed.

## Specimen Requirements:

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

**Submit only 1 of the following specimens:**

**Specimen Type:** Whole blood
**Container/Tube:**
**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 whole blood specimen in original tube. **Do not aliquot**.
**Specimen Stability Information:** Ambient (preferred) 4 days/Refrigerated

**Prenatal Specimens**
**Due to its complexity, consultation with the laboratory is required for all prenatal testing;** call 800-533-1710 to speak to a genetic counselor.

**Specimen Type:** Amniotic fluid
**Container/Tube:** Amniotic fluid container
**Specimen Volume:** 20 mL
**Specimen Stability Information:** Refrigerated (preferred)/Ambient
**Additional information:**
1. A separate culture charge will be assessed under CULAF / Culture for Genetic Testing, Amniotic Fluid.
**2. 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:** Chorionic villi
**Container/Tube:** 15-mL tube containing 15 mL of transport media
**Specimen Volume:** 20 mg
**Specimen Stability Information:** Refrigerated
**Additional Information:**
1. A separate culture charge will be assessed under CULFB / Fibroblast Culture for Biochemical or Molecular Testing.
**2. All prenatal specimens must be accompanied by a maternal blood specimen;** order MATCC / Maternal Cell Contamination, Molecular Analysis, Varies on the maternal specimen.

**Acceptable:**
**Specimen Type:** Confluent cultured cells
**Container/Tube:** T-25 flask
**Specimen Volume:** 2 Full flasks
**Collection Instructions:** Submit confluent cultured cells from another laboratory.
**Specimen Stability Information:** Ambient (preferred)/Refrigerated
**Additional Information: All prenatal specimens must be accompanied by a maternal blood specimen;** order MATCC / Maternal Cell Contamination, Molecular Analysis, Varies on the maternal specimen.

## Forms:

1. **New York Clients-Informed consent is required.** Document on the request form or electronic order that a copy is on file. The following documents are available:
-Informed Consent for Genetic Testing (T576)
-Informed Consent for Genetic Testing-Spanish (T826)
2. Molecular Genetics: Neurology Patient Information
3. If not ordering electronically, complete, print, and send a Neurology Specialty Testing Client Test Request (T732) with the specimen.

| Specimen Type | Temperature | Time | Special Container |
|---|---|---|---|
| Varies | Varies | | |

## Ask at Order Entry (AOE) Questions:

| Test ID | Question ID | Description | Type | Reportable |
|---|---|---|---|---|
| SCARA | MG323 | Test Code:<br>• SCA1<br>• SCA2<br>• SCA3<br>• SCA6<br>• SCA7 | Answer List | Yes |

## Result Codes:

| Result ID | Reporting Name | Type | Unit | LOINC® |
|---|---|---|---|---|
| 609700 | Result Summary | Alphanumeric | | 21769-5 |
| MG323 | Test Code | Alphanumeric | | 21768-7 |
| 609701 | Result | Alphanumeric | | 36911-6 |
| 609702 | Interpretation | Alphanumeric | | 69047-9 |
| 609703 | Reason for Referral | Alphanumeric | | 42349-1 |
| 609704 | Specimen | Alphanumeric | | 31208-2 |
| 609705 | Source | Alphanumeric | | 31208-2 |
| 609706 | Method | Alphanumeric | | 85069-3 |
| 609707 | Disclaimer | Alphanumeric | | 62364-5 |
| 609708 | Released By | Alphanumeric | | 18771-6 |

**LOINC® and CPT codes are provided by the performing laboratory.**

### Supplemental Report:

No

### CPT Code Information:

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)

### Reflex Tests:

| Test Id | Reporting Name | CPT Units | CPT Code | Always Performed | Available Separately |
|---|---|---|---|---|---|
| CULFB | Fibroblast Culture for Genetic Test | 1 | 88233 | No | Yes |
| CULAF | Amniotic Fluid Culture/Genetic Test | 1 | 88235 | No | Yes |
| MATCC | Maternal Cell Contamination, B | 1 | 81265 | No | Yes |
| G204 | ATXN1 (SCA 1) Gene Analysis | 1 | 81178 | No | No, (Bill Only) |
| G205 | ATXN2 (SCA 2) Gene Analysis | 1 | 81179 | No | No, (Bill Only) |
| G206 | ATXN3 (SCA 3) Gene Analysis | 1 | 81180 | No | No, (Bill Only) |
| G207 | ATXN7 (SCA 7) Gene Analysis | 1 | 81181 | No | No, (Bill Only) |
| G208 | CACNA1A (SCA 6) Gene Analysis | 1 | 81184 | No | No, (Bill Only) |
| _STR1 | Comp Analysis using STR (Bill only) | 1 | 81265 | No | No, (Bill only) |
| _STR2 | Add'l comp analysis w/STR (Bill Only) | 1 | 81266 | No | No, (Bill only) |

**Result Codes for Reflex Tests:**

| Test ID | Result ID | Reporting Name | Type | Unit | LOINC® |
|---|---|---|---|---|---|
| CULAF | 52304 | Result Summary | Alphanumeric | | 50397-9 |
| CULAF | 52306 | Interpretation | Alphanumeric | | 69965-2 |
| CULAF | 52305 | Result | Alphanumeric | | 82939-0 |
| CULAF | CG767 | Reason for Referral | Alphanumeric | | 42349-1 |
| CULAF | 52307 | Specimen | Alphanumeric | | 31208-2 |
| CULAF | 52308 | Source | Alphanumeric | | 31208-2 |
| CULAF | 52309 | Method | Alphanumeric | | 85069-3 |
| CULAF | 54641 | Additional Information | Alphanumeric | | 48767-8 |
| CULAF | 52310 | Released By | Alphanumeric | | 18771-6 |
| CULFB | 52327 | Result Summary | Alphanumeric | | 50397-9 |
| CULFB | 52329 | Interpretation | Alphanumeric | | 69965-2 |
| CULFB | 52328 | Result | Alphanumeric | | 82939-0 |
| CULFB | CG770 | Reason for Referral | Alphanumeric | | 42349-1 |
| CULFB | CG899 | Specimen | Alphanumeric | | 31208-2 |
| CULFB | 52331 | Source | Alphanumeric | | 31208-2 |
| CULFB | 52332 | Method | Alphanumeric | | 85069-3 |
| CULFB | 54625 | Additional Information | Alphanumeric | | 48767-8 |
| CULFB | 52333 | Released By | Alphanumeric | | 18771-6 |
| MATCC | 53285 | Result Summary | Alphanumeric | | 50397-9 |
| MATCC | 53286 | Result | Alphanumeric | | 40704-9 |
| MATCC | 53287 | Interpretation | Alphanumeric | | 69047-9 |
| MATCC | 53288 | Reason for referral | Alphanumeric | | 42349-1 |
| MATCC | 53289 | Specimen | Alphanumeric | | 31208-2 |
| MATCC | 53290 | Source | Alphanumeric | | 31208-2 |
| MATCC | 53291 | Released By | Alphanumeric | | 18771-6 |
| MATCC | 55150 | Method | Alphanumeric | | 85069-3 |

**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 without CAT interruptions: >38 CAG repeats
Expanded alleles with CAT interruptions: >43 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.