
Reporting Title: SCA 1,2,3,6, or 7 Repeat Analysis**Performing Location:** Rochester**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.

Specimen Minimum Volume:

Amniotic fluid: 10 mL

Blood: 0.5 mL

Chorionic villi: 5 mg

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: • SCA1 • SCA2 • SCA3 • SCA6 • SCA7	Answer List	Yes
CULFB	CG770	Reason for Referral	Plain Text	No
CULFB	CG899	Specimen	Plain Text	No
CULAF	CG767	Reason for Referral	Plain Text	No

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	Orderable Separately
CULFB	Fibroblast Culture for Genetic Test			No	Yes
CULAF	Amniotic Fluid Culture/Genetic Test			No	Yes
MATCC	Maternal Cell Contamination, B			No	Yes
_STR1	Comp Analysis using STR (Bill only)			No	No (Bill only)
_STR2	Add'l comp analysis w/STR (Bill Only)			No	No (Bill only)
G204	ATXN1 (SCA 1) Gene Analysis			No	No (Bill Only)
G205	ATXN2 (SCA 2) Gene Analysis			No	No (Bill Only)
G206	ATXN3 (SCA 3) Gene Analysis			No	No (Bill Only)
G207	ATXN7 (SCA 7) Gene Analysis			No	No (Bill Only)
G208	CACNA1A (SCA 6) Gene Analysis			No	No (Bill Only)

Result Codes for Reflex Tests:

Test ID	Result ID	Reporting Name	Type	Unit	LOINC®
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

Test ID	Result ID	Reporting Name	Type	Unit	LOINC®
CULFB	52332	Method	Alphanumeric		85069-3
CULFB	54625	Additional Information	Alphanumeric		48767-8
CULFB	52333	Released By	Alphanumeric		18771-6
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
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	55150	Method	Alphanumeric		85069-3
MATCC	53291	Released By	Alphanumeric		18771-6

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.