MAYO CLINIC LABORATORIES

NEW TEST

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

Spinocerebellar Ataxia Repeat Expansion Panel, Varies

Test ID: SCAP

Useful for:

Molecular confirmation of clinically suspected spinocerebellar ataxia

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.
Specimen:	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.
Minimum Volume:	0.5 mL

Specimen Stability Information:

Specimen Type	Temperature	Time
Varies	Ambient (preferred)	
	Frozen	
	Refrigerated	

*Specimen preferred to arrive within 96 hours of collection

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:

81178
81179
81180
81181
81184

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

Note:

The following referral test code will become obsolete.

Test Name	Test ID	Referral Lab Code	Referral Lab
Spinocerebellar Ataxia Panel	ZW278	SCAPN	University of Washington Medical Center (UWCI-Clinical Immunology)

Questions

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