

# **Test Definition: SCAP**

Spinocerebellar Ataxia Repeat Expansion Panel, Varies

Reporting Title: Spinocerebellar Ataxia Panel

Performing Location: Rochester

### Ordering Guidance:

If the patient has a family history or clinical symptoms specific to a particular type of spinocerebellar ataxia, order SCARA / Spinocerebellar Ataxia Type 1, 2, 3, 6, or 7, Repeat Expansion Analysis, Varies instead of this test.

This test and SCARA should not be ordered concurrently.

## **Shipping Instructions:**

Specimen preferred to arrive within 96 hours of collection.

# **Specimen Requirements:**

Specimen Type: Whole blood

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.

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 Minimum Volume:**

0.5 mL

### 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	Ambient (preferred)		



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	Frozen	
	Refrigerated	

## **Result Codes:**

Result ID	Reporting Name	Туре	Unit	LOINC®
609506	Result Summary	Alphanumeric		21769-5
609507	Result	Alphanumeric		36911-6
609508	Interpretation	Alphanumeric		69047-9
609509	Additional Information	Alphanumeric		48767-8
609510	Specimen	Alphanumeric		31208-2
609511	Source	Alphanumeric		31208-2
609512	Method	Alphanumeric		85069-3
609513	Disclaimer	Alphanumeric		62364-5
609514	Released By	Alphanumeric		18771-6

LOINC and CPT codes are provided by the performing laboratory.

# **Supplemental Report:**

No

## **CPT Code Information:**

81178

81179

81180

81181

81184

81479 (if appropriate for government payers)

## **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



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#### 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.