

Reporting Title: Spinocerebellar Ataxia Panel
Performing Location: Rochester

Ordering Guidance:

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.**

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)		
	Refrigerated		
	Frozen		

Result Codes:

Result ID	Reporting Name	Type	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
609512	Method	Alphanumeric		85069-3

609513	Disclaimer	Alphanumeric		62364-5
609514	Released By	Alphanumeric		18771-6
609511	Source	Alphanumeric		31208-2

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

- 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.