

Test Definition: SBULB

Spinobulbar Muscular Atrophy (Kennedy Disease), Molecular Analysis, Varies

Reporting Title: Spinobulbar Musc Atrophy, Kennedy's

Performing Location: Rochester

Shipping Instructions:

Specimen preferred to arrive within 96 hours of draw.

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 Type: Whole blood

Container/Tube:

Preferred: Lavender top (EDTA) or yellow top (ACD)

Acceptable: Any anticoagulant Specimen Volume: 3 mL Collection Instructions:

Invert several times to mix blood.
Send specimen in original tube.

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 in Special Instructions:
- -Informed Consent for Genetic Testing (T576)
- -Informed Consent for Genetic Testing-Spanish (T826)
- 2. Molecular Genetics: Neurology Patient Information in Special Instructions
- 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)				
	Frozen				
	Refrigerated				



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Result Codes:

Result ID	Reporting Name	Туре	Unit	LOINC®
53341	Result Summary	Alphanumeric		50397-9
53342	Result	Alphanumeric		82939-0
53343	Interpretation	Alphanumeric		69047-9
53344	Reason for Referral	Alphanumeric		42349-1
53345	Specimen	Alphanumeric		31208-2
53346	Source	Alphanumeric		31208-2
53348	Released By	Alphanumeric		18771-6

LOINC and CPT codes are provided by the performing laboratory.

Supplemental Report:

No

CPT Code Information:

81204-AR (androgen receptor)(eg, spinal and bulba muscular atrophy, Kennedy disease, X chromosome inactivation) gene analysis; characterization of alleles (eg, expanded size or methylation status)

Reference Values:

Normal alleles: 11-34 CAG repeats Abnormal alleles: 36-62 CAG repeats

An interpretive report will be provided.