

---

**Reporting Title:** CSTB, Repeat Expansion Analysis**Performing Location:** Rochester**Ordering Guidance:**

This test only detects dodecamer repeat expansions. If testing for both dodecamer repeat expansions and other CSTB variants is requested, order a custom gene panel for the CSTB gene. For more information see CGPH / Custom Gene Panel, Hereditary, Next-Generation Sequencing, Varies.

**Shipping Instructions:**

Specimen preferred to arrive within 96 hours of collection.

**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:**

1. Invert several times to mix blood.
2. Send whole blood specimen in original tube. Do not aliquot.

**Specimen Stability Information:** Ambient (preferred)/Refrigerated

**Forms:**

1. New York Clients-Informed consent is required. Please 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)		
	Frozen		
	Refrigerated		

**Result Codes:**

Result ID	Reporting Name	Type	Unit	LOINC®
616516	Result Summary	Alphanumeric		50397-9
616517	Result	Alphanumeric		82939-0
616518	Interpretation	Alphanumeric		69047-9
616519	Reason for Referral	Alphanumeric		42349-1
616520	Specimen	Alphanumeric		31208-2
616521	Method	Alphanumeric		85069-3
616522	Source	Alphanumeric		31208-2
616523	Released By	Alphanumeric		18771-6

LOINC and CPT codes are provided by the performing laboratory.

**Supplemental Report:**

No

**CPT Code Information:**

81188

**Reference Values:**

Normal: <5 dodecamer repeats

Repeat Size of Uncertain Significance: 5-29 dodecamer repeats

Full Penetrance Expansion: >29 dodecamer repeats

An interpretive report will be provided.