

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.