MAYO CLINIC LABORATORIES

NEW TEST

Notification Date: May 2, 2022
Effective Date: June 2, 2022

CSTB Gene, Repeat Expansion Analysis, Varies

Test ID: CSTB

Useful for:

- Molecular confirmation of clinically suspected *CSTB*-related progressive myoclonic epilepsy
- Identifying full penetrance dodecamer repeat expansions within *CSTB* known to cause *CSTB*-related progressive myoclonic epilepsy, allowing for predictive testing of at-risk family members
- Impacting patient treatment and management through the identification of a specific underlying etiology for epilepsy (eg, directing appropriate use of anti-epileptic drugs and other treatment modalities)

Methods:

Polymerase Chain Reaction (PCR)

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.

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
Minimum Volume:	1 mL

Note:
Specimen preferred to arrive within 96 hours of collection.

Specimen Stability Information:

Specimen Type	Temperature	Time	Special Container
Varies	Ambient (preferred)		
	Frozen		
	Refrigerated		

Cautions:
- For predictive testing, it is important to first document the molecular etiology of disease in an affected family member to confirm that a *CSTB* repeat expansion is the underlying mechanism of disease in the family. Specifically, this assay will not detect nonrepeat expansion variants and progressive myoclonic epilepsy may be caused by variants in other genes.
- It is recommended that patients undergoing predictive testing receive genetic counseling both prior to testing and after results are available.
- Test results should be interpreted in the context of clinical findings, family history, and other laboratory data. Errors in test interpretation may occur if the provided information is inaccurate or incomplete.
- Rare variants (ie, polymorphisms) exist which could lead to false-negative results.
- Bone marrow transplants from allogenic donors will interfere with testing. Call Mayo Clinic Laboratories for instructions for testing patients who have received a bone marrow transplant.

CPT Code:
81188

Day(s) Performed: Varies **Report Available:** 28 to 42 days

Questions
Contact Michelle Raths, Laboratory Technologist Resource Coordinator at 800-533-1710.