

Patient ID 321	Patient Name TESTRNV, IMPLEMENTATION	Birth Date 1968-02-19	Sex M	Age 54
Order Number X100383124	Client Order Number X100383124	Ordering Physician 321	Report Notes	
Account Information C7028846 DLMP Rochester		Collected 11 Mar 2022 08:45		

CSTB, Repeat Expansion Analysis

Result Summary

MCR

NEGATIVE

Result

MCR

Number of dodecamer repeats: 2 (Normal) and 2 (Normal)

Interpretation

1 MCR

This result decreases the likelihood that this individual has CSTB-related progressive myoclonic epilepsy. A diagnosis of CSTB-related progressive myoclonic epilepsy cannot be completely excluded as other variants within the CSTB gene are not detected by this method.

If a diagnosis is still suspected, testing the CSTB gene via alternative methodologies can be used to identify variants not detected by this assay. Please contact the Genomics Laboratory at 1-800-533-1710 with questions about these tests.

A genetic consultation may be of benefit.

ADDITIONAL INFORMATION

Test results should be interpreted in the context of clinical findings, family history, and other laboratory data. Misinterpretation of results may occur if the information provided is inaccurate or incomplete.

Rare polymorphisms exist that could lead to false-negative or false-positive results. If results obtained do not match the clinical

findings, additional testing should be considered.

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.

Reason for Referral

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Test for the presence of a repeat expansion in the CSTB gene.

Specimen

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WB Whole Blood

Method

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PCR-based methods were utilized to detect CCC-CGC-CCC-GCG dodecamer repeat expansions within CSTB.

Normal: <5

Repeat Size of Uncertain Significance: 5-29

Full Penetrance Expansion: >29

Released By

MCR

Cherisse A. Marcou, Ph.D.

Received: 15 Mar 2022 07:59

Reported: 21 Mar 2022 16:00

Laboratory Notes

- 1 This test was developed and its performance characteristics determined by Mayo Clinic in a manner consistent with CLIA requirements. This test has not been cleared or approved by the U.S. Food and Drug Administration.

Performing Site Legend

Code	Laboratory	Address	Lab Director	CLIA Certificate
MCR	Mayo Clinic Laboratories - Rochester Main Campus	200 First Street SW, Rochester, MN 55905	Nikola Baumann Ph.D.	24D0404292