

Patient ID 321	Patient Name TESTRNV, IMPLEMENTATION	Birth Date 1968-02-19	Sex M	Age 54
Order Number X100383794	Client Order Number X100383794	Ordering Physician UNKNOWN, PROVIDER	Report Notes	
Account Information C7028846 DLMP Rochester		Collected 24 Mar 2022 10:00		

CSTB, Repeat Expansion Analysis

Result Summary

MCR

POSITIVE

Result

MCR

Number of dodecamer repeats: 60 (Full Penetrance) and 60 (Full Penetrance)

Interpretation

1 MCR

This result is consistent with a molecular diagnosis of CSTB-related progressive myoclonic epilepsy.

As full penetrance repeat expansion alleles were detected in this individual, genetic testing of at-risk family members could be considered. If appropriate, genetic testing should be offered to this individual's reproductive partner to further clarify the risk of having a child affected with CSTB-related progressive myoclonic epilepsy.

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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Evaluation for a diagnosis of CSTB-related progressive myoclonic epilepsy. 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: 25 Mar 2022 10:12

Reported: 12 Apr 2022 12:21

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