MAYO CLINIC LABORATORIES

1-800-533-1710

FMTT

Familial Mutation, Targeted Testing, Varies

Patient ID	Patient Name		Birth Date	Sex	Age
321	FMTT, VALIDATION LABELS		1973-05-01	F	50
Order Number	**Client Order Number**	**Ordering Physician**	**Report Notes**		
X100454353	X100454353	TESTING			
Account Information		**Collected**			
C7028846 DLMP Rochester		13 Feb 2024 07:00			

Familial Mutation, Targeted Testing

Result Summary

MCR

Pathogenic Variant Detected

Result

MCR

The following heterozygous variant was detected:
Gene (Transcript): PLN (NM_002667.4)
Amino Acid change: p.R14del (Arg14del)
DNA change: c.40_42del (g.118880124_118880126del)
Classification: Pathogenic

Interpretation

1 MCR

The c.40_42del (p.Arg14del) variant in the PLN gene was previously detected in a family member and is classified by our laboratory as established pathogenic. The PLN gene encodes phospholamban, a protein found as a pentamer in the sarcoplasmic reticulum membrane which controls cellular calcium levels based on phosphorylation. Pathogenic variants in this gene are associated with autosomal dominant forms of hypertrophic cardiomyopathy, dilated cardiomyopathy, and arrhythmogenic right ventricular cardiomyopathy, also referred to collectively as intrinsic cardiomyopathy.

The c.40_42del (p.Arg14del) deletion in the PLN gene was detected and is an established pathogenic variant.

This result is supportive of a diagnosis of PLN-associated cardiomyopathy for this individual. Clinical correlation is recommended.

This assay does not exclude the presence of other variants in the gene tested. Accurate result interpretation is dependent upon correct information provided to the laboratory, including clinical and family history, and biological relationships among family members.

Consultation with a genetics professional is recommended for interpretation of this result and to determine whether reproductive risk assessment and familial testing may be of benefit to this family. Genetic testing for family members is available by ordering FMTT / Familial Mutation, Targeted Testing for the specific variant(s) detected. Please contact the laboratory at 1–800–533–1710 or the online test catalog at www.mayocliniclabs.com for information about FMTT.

ADDITIONAL INFORMATION

CLINICAL CORRELATIONS

To discuss the availability of further testing options or for assistance in the interpretation of these results, Mayo Clinic Laboratory genetic counselors can be contacted at 1–800–533–1710.

TECHNICAL LIMITATIONS

In rare cases, false negative or false positive results may occur. Given these limitations, negative results do not rule out the diagnosis of a genetic disorder. If a specific clinical disorder is suspected, evaluation by alternative methods can be considered.

If the variant(s) tested here was/were previously detected at an outside laboratory (in a family member) or in an alternate specimen, and a positive control sample was not provided, the possibility of a false negative result should be considered. Providing a control sample would confirm the laboratory's ability to detect this variant and reduces the likelihood of a false negative result for the analysis performed.

Additionally, low level mosaic variants and/or germline mosaicism may not be detected.

RECLASSIFICATION OF VARIANTS POLICY

For information regarding the laboratory's policy for reclassification of variants, please see Test ID FMTT at www.mayocliniclabs.com

VARIANT EVALUATION

Variant curation is performed using published ACMG-AMP recommendations as a guideline. Other gene-specific guidelines may also be considered. Variants classified as benign or likely benign are not reported.

Results from in silico evaluation tools may change over time and should be interpreted with caution and professional clinical judgment.

For Test Environment Only

Performing Site Legend

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

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Specimen

MCR

WB Whole Blood

Method

MCR

DNA sequence analysis was used to test for the presence of the following variant:

PLN (NM_002667.5) g.118880124_118880126del, c.40_42del, p.Arg14del (p.R14del)

Released By

MCR

Ann Marie Moyer, M.D., Ph.D.

Received: 13 Feb 2024 08:55

Reported: 21 Feb 2024 13:27

Test Environment
ETBM Template

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.

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