MAYO CLINIC LABORATORIES

1-800-533-1710

GFDZ

FTCD Gene, Full Gene Analysis

Patient ID	Patient Name		Birth Date	Gender	Age
SA00000802	SAMPLEREPDMGLM, VLD20150713A0062		1981-01-01	M	34
Order Number	Client Order Number	Ordering Physician	Report Notes		
SA00000802	SA00000802	CLIENT, CLIENT			
Account Information		Collected			
C7028846 DLMP Rochester		12 Jul 2015 14:48			

Result Summary

MCR

Positive

Result

MCR

The following heterozygous alteration was identified:
Amino Acid change: p.Q113X (Gln113X)
DNA change: c.337C>T (g.47572851)
Classification: LIKELY PATHOGENIC

Interpretation

1 MCR

The c.337C>T (p.Q113X) alteration results in a stop codon and is therefore predicted to be pathogenic.

This result indicates that this individual is a carrier of glutamate formiminotransferase deficiency. This assumes that this individual does not have biochemical evidence suggestive of glutamate formiminotransferase deficiency.

Since a mutation has been identified, genetic testing of at risk family members could be considered. Mutation specific testing is available at Mayo Medical Laboratories by ordering FMTT / Familial Mutation, Targeted Testing. Please contact the Molecular Genetics Laboratory at 1–800–533–1710 with questions about this test.

If appropriate, genetic testing should be offered to this individual's reproductive partner to further clarify their risks of having a child with glutamate formiminotransferase deficiency.

A genetic consultation may be of benefit.

Unless reported or predicted to cause disease, alterations found deep in the intron or alterations that do not result in an amino acid substitution are not reported. These and common polymorphisms identified for this patient are available upon request.

ADDITIONAL INFORMATION

Bi-directional sequence analysis was performed to test for the presence of mutations in all coding regions and intron/exon boundaries of the FTCD gene (GenBank accession number NM_206965; build GRCh37 (hg19)). An online research opportunity called GenomeConnect (genomeconnect.org), a project of ClinGen, is available for the recipient of this genetic test. This patient registry collects de-identified genetic and health information to advance the knowledge of genetic variants. Mayo Clinic is a collaborator of ClinGen. This may not be applicable for all tests.

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.

Multiple in-silico evaluation tools may have been used to assist in the interpretation of these results. Of note, the sensitivity and specificity of these tools for the determination of pathogenicity is currently unvalidated.

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

Released By MCR

EMILY LAUER

Received: 13 Jul 2015 20:20 **Reported:** 24 Jul 2015 14:06

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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