

| | | | | |
|---|---|---|--------------------|------------------|
| Patient ID SA00000960 | Patient Name SAMPLEREPDMGLM, VLD20150713A0221 | Birth Date 1981-01-01 | Gender M | Age 34 |
| Order Number SA00000960 | Client Order Number SA00000960 | Ordering Physician CLIENT, CLIENT | Report Notes | |
| Account Information C7028846 DLMP Rochester | | Collected 12 Jul 2015 18:02 | | |

Result Summary

MCR

Negative

Result

MCR

No reportable variants were identified.

Interpretation

1 MCR

This result decreases the likelihood but does not rule out the diagnosis of glutamate formiminotransferase deficiency for this individual. We predict that there are individuals with a diagnosis of glutamate formiminotransferase deficiency who have pathogenic mutations that are not detectable by the method described (e.g. large deletions/duplications, promoter mutations, or deep intronic mutations). Additionally, this assay does not rule out the presence of mutations in other genes associated with metabolic disease.

This result should be interpreted in the context of clinical findings, family history, and other laboratory testing (e.g. measurement of urine formiminotransferase (FIGLU)).

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.

Specimen

MCR

WB Whole Blood

Released By

MCR

EMILY LAUER

Received: 13 Jul 2015 20:26

Reported: 24 Jul 2015 14:31

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 |



| | | | | |
|---|---|---|--------------------|------------------|
| Patient ID SA00000960 | Patient Name SAMPLEREPDMGLM, VLD20150713A0221 | Birth Date 1981-01-01 | Gender M | Age 34 |
| Order Number SA00000960 | Client Order Number SA00000960 | Ordering Physician CLIENT, CLIENT | Report Notes | |
| Account Information C7028846 DLMP Rochester | | Collected 12 Jul 2015 18:02 | | |

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.

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 |