MAYO CLINIC
Mayo Medical Laboratories

# Laboratory Service Report 1-800-533-1710

| Patient Name | Patient ID | Age | Gender | Order # |
|---|---|---|---|---|
| SAMPLEREPORT,FECHS | SA00046704 | 45 | F | SA00046704 |

| Ordering Phys | | DOB |
|---|---|---|
| | | 06/10/1966 |

| Client Order # | Account Information | Report Notes |
|---|---|---|
| SA00046704 | C7028846-DLMP ROCHESTER<br>3050 SUPERIOR DRIVE<br>ROCHESTER,MN 55901 | |
| **Collected**<br>05/20/2012 | | |
| **Printed**<br>09/15/2012 12:25 | | |

| Test | Flag | Results | Unit | Reference Value | Perform Site* |
|---|---|---|---|---|---|
| **FECH Gene, Full Gene Analysis** | | REPORTED 07/13/2012 10:07 | | | |
| Specimen | | Blood | | | MCR |
| Specimen ID | | 1038198 | | | MCR |
| Order Date | | 22 May 2012 15:01 | | | MCR |
| Reason For Referral | | | | | MCR |
| Patient reported to have features suggestive of erythropoietic protoporphyria (EPP). Test for the presence of mutations in the FECH gene. | | | | | |
| Method | | | | | MCR |
| Bi-directional sequence analysis was performed to test for the presence of mutations in all coding regions and intron/exon boundaries of the FECH gene. Mutation nomenclature is based on GenBank accession number; NM 000140.3. | | | | | |
| Result | | | | | MCR |
| A mutation was NOT detected. | | | | | |
| Interpretation | | | | | MCR |

This result decreases the likelihood of a diagnosis of EPP due to ferrochelatase deficiency. However, we predict that some individuals who have a diagnosis of EPP may have mutations in the FECH gene that are not identifiable by the method described (e.g. large deletions/duplications, promoter mutations, or deep intronic mutations).

This assay does not rule out the presence of mutations in other genes that are associated with EPP, such as ALAS2. This result should be interpreted in light of clinical findings, family history, and other laboratory tests, such as Porphyrins Evaluation, Erythrocytes (Mayo Test ID PEE/88886).

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.

CAUTIONS:
Test results should be interpreted in 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

***Performing Site Legend on Last Page of Report***



* Report times for Mayo performed tests are CST/CDT

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| or 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 Medical Laboratories for instructions for testing patients who have received a bone marrow transplant. | | | | | |
| Laboratory developed test. | | | | | |
| Extraction Performed? | | Yes | | | MCR |
| Reviewed By<br>Melody Elizabeth Kimball | | | | | MCR |
| Release Date | | 13 Jul 2012 10:01 | | | MCR |

* Performing Site:

| | | |
|---|---|---|
| MCR | Mayo Clinic Laboratories - Rochester Main Campus<br>200 First St SW Rochester, MN 55905 | Lab Director: |