MAYO CLINIC
Mayo Medical Laboratories

Laboratory Service Report 1-800-533-1710

Patient Name	Patient ID	Age	Gender	Order #
SAMPLEREPORT,FECHK A	SA00058759	46	F	SA00058759
Ordering Phys CLIENT,CLIENT				**DOB** 06/10/1966
Client Order # SA00058759	**Account Information**			**Report Notes**
Collected 06/04/2013 00:00	C7028846-DLMP Rochester 3050 Superior Drive Rochester, MN 55901			
Printed 06/18/2013 09:15				

Test	Flag	Results	Unit	Reference Value	Perform Site*
FECH Gene, Known Mutation					
RECEIVED: 06/04/2013 17:36 **REPORTED:** 06/10/2013 15:17					
Specimen		Blood			MCR
Specimen ID		1062041			MCR
Order Date		05 Jun 2013 08:43			MCR
Reason For Referral					MCR
Family history of erythropoietic protoporphyria (EPP). Test for the presence of mutations in the FECH gene.					
Method					MCR
DNA sequence analysis was used to test for the presence of the p.C406Y (c.1217G>A) alteration in exon 11 of the FECH gene (GenBank accession number; NM 000140.3). Analysis for this specific alteration was performed because it has been identified in a family member.					
Result					MCR
The following heterozygous sequence change was detected: Exon: 11 DNA change: c.1217G>A Amino Acid change: p.C406Y (Cys406Tyr) Classification: DELETERIOUS					
Interpretation					MCR
This alteration is a known deleterious mutation. The presence of one mutation indicates that this individual is a carrier of EPP. This interpretation assumes that this individual is clinically healthy and is not affected with EPP. 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). This assay does not rule out the presence of other disease causing mutations within this gene or within other genes that are associated with porphyria. Errors in the diagnosis or pedigree provided to us, including non paternity, may lead to an erroneous interpretation of test results. Since a mutation in the FECH gene has been identified, testing of other at risk family members is possible. 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. CAUTIONS: Rare polymorphisms exist that could lead to false negative					

Performing Site Legend on Last Page of Report

Patient Name	Collection Date and Time	Report Status
SAMPLEREPORT,FECHK A	06/04/2013 00:00	Final
		>> Continued on Next Page >>

* 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.					
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.					
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 Emily Christine Lauer					MCR
Release Date		10 Jun 2013 15:15			MCR

* Performing Site:

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

Patient Name	Collection Date and Time	Report Status
SAMPLEREPORT,FECHK A	06/04/2013 00:00	Final
Page 2 of 2		** End of Report **

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