MAYO CLINIC
Mayo Medical Laboratories

Laboratory Service Report 1-800-533-1710

Patient Name	Patient ID	Age	Gender	Order #
SAMPLEREOPRT,FECHS	SA00046707	45	F	SA00046707
Ordering Phys				**DOB** 06/10/1966
Client Order # SA00046707	**Account Information**			**Report Notes**
Collected 05/20/2012	C7028846-DLMP ROCHESTER 3050 SUPERIOR DRIVE ROCHESTER,MN 55901			
Printed 09/15/2012 12:26				

Test	Flag	Results	Unit	Reference Value	Perform Site*
FECH Gene, Full Gene Analysis			REPORTED 07/13/2012 10:07		
Specimen		Blood			MCR
Specimen ID		1038199			MCR
Order Date		22 May 2012 15:02			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

The following heterozygous intronic sequence change was detected:
Intron: 3
DNA change: c.315-48T>C (IVS3-48T>C)
This variant is associated with reduced gene expression.

The following heterozygous deletion was detected:
Exon: 8
DNA change: c.901_902delTG
Amino Acid change: p.W301AfsX23 (Trp301Alafs23)
Classification: DELETERIOUS

Interpretation MCR

The 315-48T>C variant is associated with reduced FECH gene expression.

The c.901_902delTG alteration is a known deleterious mutation.

This finding is consistent with a diagnosis of erythropoietic protoporphyria (EPP).

Since a mutation has been identified, testing of at risk family members is possible. Mutation specific testing is available at Mayo Medical Laboratories by ordering FECHK/60372. Please contact the Molecular Genetics Laboratory at 1-800-533-1710 with questions about this test.

A genetic consultation may be of benefit.

Performing Site Legend on Last Page of Report

* Report times for Mayo performed tests are CST/CDT

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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 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					MCR
Melody Elizabeth Kimball					
Release Date		13 Jul 2012 10:01			MCR

* Performing Site:

MCR	Mayo Clinic Laboratories - Rochester Main Campus 200 First St SW Rochester, MN 55905	Lab Director: Franklin R. Cockerill, III, M.D.

TEST SITE