

Patient Name SAMPLEREPORT,FECHK	Patient ID SA00046689	Age 45	Gender F	Order # SA00046689
Ordering Phys				DOB 06/10/1966
Client Order # SA00046689	Account Information			Report Notes
Collected 05/20/2012	C7028846-DLMP ROCHESTER 3050 SUPERIOR DRIVE ROCHESTER,MN 55901			
Printed 09/15/2012 12:21				

Test	Flag	Results	Unit	Reference Value	Perform Site*
FECH Gene, Known Mutation			REPORTED 07/13/2012 10:05		
Specimen		Blood			MCR
Specimen ID		1038195			MCR
Order Date		21 May 2012 14:59			MCR
Reason For Referral		Family history of erythropoietic protoporphyria (EPP). Test for the presence of mutations in the FECH gene.			MCR
Method		DNA sequence analysis was used to test for the presence of the c.315-48T>C and c.901_902delTG alterations in intron 3 and exon 8, respectively, of the FECH gene (GenBank accession number; NM 000140.3). Analysis for these specific alterations was performed because they have been identified in a family member.			MCR
Result		The c.315-48T>C and c.901_902delTG alterations were NOT detected.			MCR
Interpretation		Absence of the mutation(s) previously identified in an affected family member indicates that this individual is at no greater risk than someone in the general population for developing symptoms related to EPP.			MCR
		This assay does not rule out the presence of other disease causing mutations within this gene or 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.			
		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 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.			

Performing Site Legend on Last Page of Report

Patient Name SAMPLEREPORT,FECHK	Collection Date and Time 05/20/2012	Report Status Final
Page 1 of 2		>> Continued on Next Page >>

* Report times for Mayo performed tests are CST/CDT

Patient Name SAMPLEREP,FECHK	Patient ID SA00046689	Age 45	Gender F	Order # SA00046689
Ordering Phys				DOB 06/10/1966
Client Order # SA00046689	Account Information C7028846-DLMP ROCHESTER 3050 SUPERIOR DRIVE ROCHESTER,MN 55901			Report Notes
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Test	Flag	Results	Unit	Reference Value	Perform Site*
<p>Misinterpretation of results may occur if the information provided is inaccurate or incomplete.</p> <p>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.</p> <p>Laboratory developed test.</p>					
Extraction Performed?		Yes			MCR
Reviewed By Melody Elizabeth Kimball					MCR
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:
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Patient Name SAMPLEREP,FECHK	Collection Date and Time 05/20/2012	Report Status Final
Page 2 of 2		** End of Report **

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