MAYO CLINIC
Mayo Medical Laboratories

Laboratory Service Report 1-800-533-1710

Patient Name	Patient ID	Age	Gender	Order #
SAMPLE REPORTS,PPOXK	SA00054582	4D	F	SA00054582
Ordering Phys CLIENT,CLIENT				**DOB** 03/03/2013
Client Order # SA00054582	**Account Information**			**Report Notes**
Collected 03/07/2013	C7028846-DLMP Rochester 3050 Superior Drive Rochester, MN 55901			
Printed 03/26/2013 14:06				

Test	Flag	Results	Unit	Reference Value	Perform Site*
PPOX Gene, Known Mutation			REPORTED 03/08/2013 09:38		
Reason for Referral					MCR
Family history of fibrinogen alpha chain (FGA)-related familial visceral amyloidosis. Test for the presence of a mutation in the FGA gene.					
Result					MCR
The p.Q435X mutation was NOT detected.					
Interpretation					MCR
This result does not provide evidence for a diagnosis of variegate porphyria. This interpretation assumes that the mutation listed above was previously identified in an affected family member.					
This assay does not rule out the presence of other mutations in this gene or in 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. 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.					
Method					MCR

Performing Site Legend on Last Page of Report

* Report times for Mayo performed tests are CST/CDT

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A sequence analysis was used to test for the presence of the c.1303C>T (p.Q435X) mutation in exon 12 of the PPOX gene. Testing was performed for this specific mutation because it was previously identified in an affected family member of this individual. Mutation nomenclature is based on GenBank accession number; NM_000309.3.					
Specimen		Blood			MCR
Reviewed By		Devin Oglesbee PhD			MCR
Release Date		08 Mar 2013 09:36			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.