MAYO CLINIC
Mayo Medical Laboratories

Laboratory Service Report 1-800-533-1710

Patient Name	Patient ID	Age	Gender	Order #
SAMPLE REPORTS,PPOXK	SA00054583	4D	M	SA00054583
Ordering Phys CLIENT,CLIENT				**DOB** 03/03/2013
Client Order # SA00054583	**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 following heterozygous sequence change was detected: Exon: 12 DNA change: c.1303C>T Amino acid change: p.Q435X (Gln435X) Classification: DELETERIOUS					
Interpretation					MCR
The p.Q435X alteration is a known deleterious mutation. This result is consistent with a diagnosis of variegate porphyria. Appropriate prophylactic and screening procedures should be considered. A genetic consultation may be of benefit. Since a mutation has been identified, testing of at risk family members is possible. 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.					

Performing Site Legend on Last Page of Report

* Report times for Mayo performed tests are CST/CDT

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Test	Flag	Results	Unit	Reference Value	Perform Site*
Laboratory developed test.					
Method		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.			MCR
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:

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