MAYO CLINIC
Mayo Medical Laboratories

Laboratory Service Report 1-800-533-1710

Patient Name	Patient ID	Age	Gender	Order #
SAMPLEREPORT,CPOXK	SA00049981	11	M	SA00049981
Ordering Phys				**DOB** 06/15/2001
Client Order # SA00049981	**Account Information**			**Report Notes**
Collected 10/23/2012	C7028846-DLMP ROCHESTER 3050 SUPERIOR DRIVE ROCHESTER,MN 55901			
Printed 11/07/2012 16:39				

Test	Flag	Results	Unit	Reference Value	Perform Site*
CPOX Gene, Known Mutation		REPORTED 10/23/2012 15:22			
Reason For Referral					MCR
Family history of hereditary coproporphyria (HCP). Test for the presence of a mutation in the CPOX gene.					
Result					MCR
The p.Q306X mutation was NOT detected.					
Interpretation					MCR
This result does not provide evidence for a diagnosis of hereditary coproporphyria (HCP). 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
DNA sequence analysis was used to test for the presence of					

Performing Site Legend on Last Page of Report

* Report times for Mayo performed tests are CST/CDT

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the c.916C>T (p.Q306X) mutation in exon 4 of the CPOX 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_000097.5.					
Specimen		Blood			MCR
Reviewed By		Devin Oglesbee PhD			MCR
Release Date		23 Oct 2012 15:19			MCR

* Performing Site:

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