Patient Name	Patient ID	Age	Gender	Order #
SAMPLEREPORT,CPOXS	SA00049983	6	M	SA00049983
Ordering Phys				**DOB** 06/15/2006
Client Order # SA00049983	**Account Information**			**Report Notes**
Collected 10/23/2012	C7028846-DLMP ROCHESTER 3050 SUPERIOR DRIVE ROCHESTER,MN 55901			
Printed 11/07/2012 16:32				

Test	Flag	Results	Unit	Reference Value	Perform Site*
CPOX Gene, Full Gene Analysis		REPORTED 10/23/2012 15:22			
Reason For Referral					MCR
Possible diagnosis of hereditary coproporphyria (HCP). Test for the presence of a mutation in the CPOX gene.					
Result					MCR
A mutation was NOT detected.					
Interpretation					MCR

This result decreases the likelihood but does not rule out a diagnosis of hereditary coproporphyria (HCP). We predict that some individuals who have a diagnosis of HCP may have a mutation that is not identified by the methods described (e.g. large deletions/duplications, promoter mutations, or deep intronic mutations). Additionally, the clinical phenotype that is observed in this individual may be due to a disease causing mutation in other genes (i.e. HMBS and PPOX). This assay does not rule out the presence of disease causing mutations in other genes that are associated with porphyria.

This result should be interpreted in the context of clinical findings, family history, and other laboratory testing (i.e. porphobilinogen deaminase and urine porphobilinogen).

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

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Test	Flag	Results	Unit	Reference Value	Perform Site*
marrow transplant. Laboratory developed test.					
Method		Bi-directional sequence analysis was performed to test for the presence of a mutation in all coding regions and intron/exon boundaries of the CPOX gene. Mutation nomenclature is based on GenBank accession number; NM_000097.5.			MCR
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: