

Laboratory Service Report

1-800-533-1710

Patient Name SAMPLEREPORT, CPOXS	Patient ID SA00049984	Age 5	Gender M	Order # SA00049984
Ordering Phys	C/ 1000 1000 1			DOB 06/15/2007
Client Order # SA00049984	Account Information			Report Notes
Collected 10/23/2012	3050 SUPERIOR DRI	C7028846-DLMP ROCHESTER 3050 SUPERIOR DRIVE		
Printed 11/07/2012 16:33	ROCHESTER,MN 559	901		

Reference Perform
Test Flag Results Unit Value Site*

CPOX Gene, Full Gene Analysis

REPORTED 10/23/2012 15:22

MCR

MCR

MCR

Reason For Referral

Possible diagnosis of hereditary coproporphyria (HCP). Test

for the presence of a mutation in the CPOX gene. Result

The following heterozygous sequence change was detected:

Exon: 4

DNA change: c.916C>T

Amino acid change: p.Q306X (Gln306X)

Classification: DELETERIOUS

Interpretation

This alteration is a known deleterious mutation.

This result is consistent with a diagnosis of hereditary coproporphyria (HCP).

Since a mutation has been identified, testing of at risk family members is possible. Mutation-specific testing for HCP is available at Mayo Medical Laboratories by ordering CPOXK/61264 CPOX Gene, Known Mutation. Please contact the Molecular Genetics Laboratory at 1-800-533-1710 with questions about this test.

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

Performing Site Legend on Last Page of Report

Patient Name	Collection Date and Time	Report Status
SAMPLEREPORT, CPOXS	10/23/2012	Final
Page 1 of 2		>> Continued on Next Page >>

^{*} Report times for Mayo performed tests are CST/CDT



Laboratory Service Report

1-800-533-1710

Patient Name SAMPLEREPORT,CPOXS	Patient ID SA00049984	Age 5	Gender M	Order # SA00049984
Ordering Phys		1		DOB 06/15/2007
Client Order # SA00049984	Account Information			Report Notes
Collected 10/23/2012	3050 SUPERIOR DRI	C7028846-DLMP ROCHESTER 3050 SUPERIOR DRIVE		
Printed 11/07/2012 16:33	ROCHESTER,MN 559	901		

Test	Flag Results	Unit	Reference Value	Perform Site*
marrow transplant.				
the presence of a mutatic intron/exon boundaries of nomenclature is based on	nalysis was performed to test on in all coding regions and the CPOX gene. Mutation GenBank accession number;	for		MCR
NM_000097.5. Specimen Reviewed By Release Date	Blood Devin Oglesbee 23 Oct 2012 15:			MCR MCR MCR

* Performing Site:

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

Patient Name	Collection Date and Time	Report Status
SAMPLEREPORT, CPOXS	10/23/2012	Final
Page 2 of 2		** End of Report **

^{*} Report times for Mayo performed tests are CST/CDT