MAYO CLINIC
Mayo Medical Laboratories

Laboratory Service Report 1-800-533-1710

Patient Name	Patient ID	Age	Gender	Order #
SAMPLEREPORT,PPAN	SA00054560	47	M	SA00054560
Ordering Phys CLIENT,CLIENT				**DOB** 06/15/1965
Client Order # SA00054560	**Account Information**			**Report Notes**
Collected 03/07/2013	C7028846-DLMP Rochester 3050 Superior Drive Rochester, MN 55901			
Printed 03/26/2013 14:05				

Test	Flag	Results	Unit	Reference Value	Perform Site*
Acute Porphyria, Multi-Gene Panel			REPORTED 03/07/2013 09:04		
Reason For Referral					MCR

Test for the presence of mutations in the genes included in the Acute Porphyria Panel.

Result MCR

The following heterozygous sequence change was detected:
Exon: 14
DNA change: c.849G>A
Amino acid change: p.W283X (Trp283X)
Classification: DELETERIOUS

Interpretation MCR

This alteration is a known deleterious mutation.

This result is consistent with a diagnosis of acute porphyria due to HMBS deficiency (acute intermittent porphyria). Appropriate prophylactic and screening procedures should be considered.

Since a mutation has been identified, testing of at risk family members is possible. Mutation-specific testing is available at Mayo Medical Laboratories by ordering HMBSK/61217 HMBS 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

Performing Site Legend on Last Page of Report

* Report times for Mayo performed tests are CST/CDT

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with testing. Call Mayo Medical Laboratories for instructions for testing patients who have received a bone marrow transplant.					
Laboratory developed test.					
Method					MCR
Bi-directional sequence analysis was performed to test for the presence of a mutation in all coding regions and intron/exon boundaries of the HMBS, CPOX, and PPOX genes. Mutation nomenclature is based on GenBank accession numbers NM_000190.3 (HMBS); NM_000097.5 (CPOX); and NM_000309.3 (PPOX).					
Specimen		Blood			MCR
Reviewed By		Devin Oglesbee PhD			MCR
Release Date		07 Mar 2013 09:03			MCR

* Performing Site:

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