MAYO CLINIC
Mayo Medical Laboratories

Laboratory Service Report 1-800-533-1710

Patient Name	Patient ID	Age	Gender	Order #
TESTINGRNV,REPORTS	SA00051508	1D	M	SA00051508
Ordering Phys CLIENT,CLIENT				**DOB** 12/03/2012
Client Order # SA00051508	**Account Information**			**Report Notes**
Collected 12/04/2012	C7028846-DLMP ROCHESTER 3050 SUPERIOR DRIVE ROCHESTER,MN 55901			
Printed 01/04/2013 14:59				

Test	Flag	Results	Unit	Reference Value	Perform Site*
GRHPR Gene, Known Mutation			REPORTED 12/05/2012 09:05		
Reason for Referral					MCR
Family history of primary hyperoxaluria type 2 (PH2). Test for the presence of familial mutations within the GRHPR gene.					
Result					MCR
The following homozygous sequence change was detected: Exon: 2 DNA change: c.103delG Amino acid change: p.D35TfsX11 (Asp35ThrfsX11) Classification: DELETERIOUS					
Interpretation					MCR
The c.103delG alteration is a known deleterious mutation. This result is consistent with a diagnosis of primary hyperoxaluria type 2 (PH2). 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 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. 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. 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 the c.103delG alteration in exon 2 of the GRHPR gene.					

Performing Site Legend on Last Page of Report

* Report times for Mayo performed tests are CST/CDT

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Testing was performed for this specific alteration because it was previously identified in an affected family member of this individual. Mutation nomenclature is based on GenBank accession number; NM_012203.1.					
Specimen		Blood			MCR
Reviewed By		D Brian Dawson PhD			MCR
Release Date		05 Dec 2012 09:04			MCR

* Performing Site:

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

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