MAYO CLINIC
Mayo Medical Laboratories

Laboratory Service Report 1-800-533-1710

Patient Name	Patient ID	Age	Gender	Order #
TESTINGRNV,REPORTS	SA00051505	1D	M	SA00051505
Ordering Phys				**DOB**
CLIENT,CLIENT				12/03/2012
Client Order #	**Account Information**			**Report Notes**
SA00051505				
Collected	C7028846-DLMP ROCHESTER			
12/04/2012	3050 SUPERIOR DRIVE			
Printed	ROCHESTER,MN 55901			
01/04/2013 15:06				

Test	Flag	Results	Unit	Reference Value	Perform Site*
GRHPR Gene, Full Gene Analysis			REPORTED 12/05/2012 09:03		
Reason for Referral					MCR
Possible diagnosis of primary hyperoxaluria type 2 (PH2). Test for the presence of mutations in the GRHPR gene.					
Result					MCR
A mutation was NOT detected.					
Interpretation					MCR

This result decreases the likelihood but does not rule out the diagnosis of primary hyperoxaluria type 2 (PH2). We predict that a small percentage of individuals who have a diagnosis of PH2 have mutations that are not detectable by the methods described (e.g. promoter mutations or intronic mutations).

Measurement of urinary oxalate in a timed, 24 hour urine collection (Mayo unit code #HYOX/86213 Hyperoxaluria Panel, Urine) is recommended to provide further evidence for a diagnosis of 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.

Performing Site Legend on Last Page of Report

* Report times for Mayo performed tests are CST/CDT

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12/04/2012	3050 SUPERIOR DRIVE			
Printed	ROCHESTER,MN 55901			
01/04/2013 15:07				

Test	Flag	Results	Unit	Reference Value	Perform Site*
Method					MCR
Bi-directional sequence analysis was performed to test for the presence of mutations in all coding regions and intron/exon boundaries of the GRHPR gene. Gene dosage analysis (MLPA) was used to test for the presence of large deletions and duplications in this gene. 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 09:01			MCR

* Performing Site:

MCR	Mayo Clinic Laboratories - Rochester Main Campus 200 First St SW Rochester, MN 55905	Lab Director: Franklin R. Cockerill, III, M.D.

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