MAYO CLINIC
Mayo Medical Laboratories

Laboratory Service Report 1-800-533-1710

Patient Name	Patient ID	Age	Gender	Order #
SAMPLEREPORT,AGXMS	SA00045504	45	F	SA00045504
Ordering Phys				**DOB** 06/10/1966
Client Order # SA00045504	**Account Information**			**Report Notes**
Collected 04/28/2012 18:13	C7028846-DLMP ROCHESTER 3050 SUPERIOR DRIVE ROCHESTER,MN 55901			
Printed 04/30/2012 15:06				

Test	Flag	Results	Unit	Reference Value	Perform Site*
AGXT Gene, Full Gene Analysis			REPORTED 04/30/2012 10:43		
Specimen		Blood			MCR
Specimen ID		1038085			MCR
Order Date		30 Apr 2012 10:41			MCR
Reason For Referral					MCR
Not Provided. Test for the presence of mutations in the AGXT gene.					
Method					MCR
DNA sequence analysis was performed to test for the presence of mutations in all 11 exons of the AGXT gene. Gene dosage analysis (MLPA) was used to test for the presence of large deletions and duplications within these genes. Mutation nomenclature is based on GenBank accession number NM_000030.2.					
Result					MCR
A mutation was NOT detected.					
Interpretation					MCR

These results decrease the likelihood but do not rule out the diagnosis of primary hyperoxaluria type 1 (PH1) for this individual. We predict that a small percentage of individuals who have a diagnosis of PH1 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 #86213 Hyperoxaluria Panel, Urine) is recommended to provide further evidence for a diagnosis of PH1.

A genetic consultation may be of benefit.

A list of common polymorphisms identified for this patient is 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.

Performing Site Legend on Last Page of Report

* Report times for Mayo performed tests are CST/CDT

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Test	Flag	Results	Unit	Reference Value	Perform Site*
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.					
Extraction Performed?		Yes			MCR
MLPA Performed?		Yes.			MCR
See #89918, AGXT Large Deletion/Duplication, MLPA, for billing information.					
Reviewed By		Heather Lynn Owen			MCR
Release Date		30 Apr 2012 10:42			MCR

* Performing Site:

MCR	Mayo Clinic Dpt of Lab Med & Pathology 200 First St SW Rochester, MN 55905	Lab Director: Franklin R. Cockerill, III, M.D.

Patient Name	Collection Date and Time	Report Status
SAMPLEREPORT,AGXMS	04/28/2012 18:13	Final
Page 2 of 2		** End of Report **

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