MAYO CLINIC
Mayo Medical Laboratories

Laboratory Service Report 1-800-533-1710

Patient Name	Patient ID	Age	Gender	Order #
SAMPLEREPORT,AGXT	SA00045546	45	F	SA00045546
Ordering Phys				**DOB** 06/10/1966
Client Order # SA00045546	**Account Information**			**Report Notes**
Collected 05/01/2012	C7028846-DLMP ROCHESTER 3050 SUPERIOR DRIVE ROCHESTER,MN 55901			
Printed 05/03/2012 13:32				

Test	Flag	Results	Unit	Reference Value	Perform Site*
AGXT Mutation Analysis (G170R)			REPORTED 05/02/2012 08:48		
Specimen		Blood			MCR
Specimen ID		1038101			MCR
Order Date		02 May 2012 08:45			MCR
Reason For Referral					MCR

Patient has diagnosis of hyperoxaluria type 1. Analyze specimen for the presence of the G170R mutation within the alanine:glyoxylate aminotransferase (AGXT) gene.

Method MCR

A PCR-based assay was used to examine for the presence of the 630G>A mutation within the alanine:glyoxylate aminotransferase (AGXT) gene.

Result MCR

One copy of the G170R mutation within the alanine:glyoxylate aminotransferase (AGXT) gene was detected.

Interpretation MCR

One copy of the G170R mutation was identified. Primary hyperoxaluria type 1 (PH1) is an autosomal recessive disorder. Therefore, this result does not exclude the diagnosis of primary hyperoxaluria, type 1 (PH 1) since other disease causing mutations may be present.

Heterozygosity for the G170R mutation within the alanine:glyoxylate aminotransferase (AGXT) gene has been associated with favorable response to pyridoxine treatment in affected individuals.

Biochemical testing including urine glycolate and glycerate (MML test code 86213) may provide useful information.

As a mutation has been identified for this individual, carrier testing of other family members is possible. A genetic consultation may be of benefit.

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.					
Reviewed By: Melody Elizabeth Kimball					MCR
Release Date		02 May 2012 08:46			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,AGXT	05/01/2012	Final
		** End of Report **

* Report times for Mayo performed tests are CST/CDT