MAYO CLINIC
Mayo Medical Laboratories

Laboratory Service Report 1-800-533-1710

Patient Name	Patient ID	Age	Gender	Order #
SAMPLEREPORT,AGXT	SA00045507	45	F	SA00045507
Ordering Phys				**DOB** 06/10/1966
Client Order # SA00045507	**Account Information**			**Report Notes**
Collected 04/28/2012 18:20	C7028846-DLMP ROCHESTER 3050 SUPERIOR DRIVE ROCHESTER,MN 55901			
Printed 04/30/2012 15:22				

Test	Flag	Results	Unit	Reference Value	Perform Site*
AGXT Mutation Analysis (G170R)			REPORTED 04/30/2012 10:50		
Specimen		Blood			MCR
Specimen ID		1038088			MCR
Order Date		30 Apr 2012 10:46			MCR
Reason For Referral					MCR

Patient reported to have features 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

The G170R mutation within the alanine:glyoxylate aminotransferase (AGXT) gene was not detected.

Interpretation MCR

The G170R mutation was not identified. This result does not exclude the diagnosis of primary hyperoxaluria, type 1 (PH 1) since other disease causing mutations may be present. If this individual is known to be affected, he/she is not predicted to show significant clinical improvement after pyridoxine administration. However, a trial for pyridoxine-treatment responsiveness should be performed since a small proportion of individuals without the G170R mutation may benefit from pyridoxine.

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

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.

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*
Laboratory developed test.					
Reviewed By:		Heather Lynn Owen			MCR
Release Date		30 Apr 2012 10:48			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,AGXT	04/28/2012 18:20	Final
		** End of Report **

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