MAYO CLINIC
Mayo Medical Laboratories

Laboratory Service Report 1-800-533-1710

Patient Name	Patient ID	Age	Gender	Order #
TESTINGRNV,LYZMS	SA00048143	18	M	SA00048143
Ordering Phys				**DOB** 06/15/1994
Client Order # SA00048143	**Account Information**			**Report Notes**
Collected 07/31/2012	C7028846-DLMP ROCHESTER 3050 SUPERIOR DRIVE ROCHESTER,MN 55901			
Printed 08/30/2012 15:25				

Test	Flag	Results	Unit	Reference Value	Perform Site*
LYZ Gene, Full Gene Analysis			REPORTED 08/08/2012 08:24		
Specimen		Blood			MCR
Specimen ID		1038494			MCR
Order Date		31 Jul 2012 14:55			MCR
Reason For Referral					MCR

Possible diagnosis of lysozyme(LYZ)-related familial visceral amyloidosis. Test for the presence of a mutation in the LYZ gene.

Method MCR

Bi-directional sequence analysis was performed to test for the presence of a mutation in all coding regions and intron/exon boundaries of the LYZ gene. Mutation nomenclature is based on GenBank accession number; NM.000239

Result MCR

The following sequence change was detected:
Exon: 4
DNA change: c. 388T>C
Amino acid change: p.W130R (Trp130Arg)
Legacy nomenclature: p.W112R (Trp112Arg)
Classification: DELETERIOUS

Interpretation MCR

This alteration is a known deleterious mutation.

This result is consistent with a diagnosis of LYZ-related familial visceral amyloidosis.

Since a mutation has been identified, testing of at risk family members is possible. Mutation-specific testing for LYZ-related familial amyloidosis is available at Mayo Medical Laboratories by ordering LYZKM/60720 LYZ Gene, Known Mutation. Please contact the Molecular Genetics Laboratory at 1-800-533-1710 with questions about this test.

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:
Rare polymorphisms exist that could lead to false negative

Performing Site Legend on Last Page of Report

* Report times for Mayo performed tests are CST/CDT

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or positive results. If results obtained do not match the clinical findings, additional testing should be considered.					
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.					
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					MCR
W Edward Highsmith Jr., PhD					
Release Date		08 Aug 2012 08:21			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.