MAYO CLINIC
Mayo Medical Laboratories

Laboratory Service Report 1-800-533-1710

Patient Name	Patient ID	Age	Gender	Order #
SAMPLEREPORT,AMYL	SA00046650	45	F	SA00046650
Ordering Phys				**DOB** 06/10/1966
Client Order # SA00046650	**Account Information**			**Report Notes**
Collected 05/20/2012	C7028846-DLMP ROCHESTER 3050 SUPERIOR DRIVE ROCHESTER,MN 55901			
Printed 09/14/2012 15:10				

Test	Flag	Results	Unit	Reference Value	Perform Site*
Familial Amyloidosis, DNA Sequence			REPORTED 07/13/2012 10:01		
Specimen		Blood			MCR
Specimen ID		1038183			MCR
Order Date		21 May 2012 13:18			MCR
Reason For Referral					MCR

Possible diagnosis of transthyretin (TTR)-associated familial amyloidosis. Test for the presence of a mutation in the TTR 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 TTR gene. Mutation nomenclature is based on GenBank accession number NM 000371.

Result — MCR

The following heterozygous sequence change was detected:
Exon: 4
DNA change: c.424G>A
Amino acid change: p. V122I (Val122Ile)
Classification: DELETERIOUS

Interpretation — MCR

This alteration is a known deleterious mutation.

This result is consistent with a diagnosis of TTR associated familial amyloidosis.

Since a mutation has been identified, testing of at risk family members is possible. Mutation-specific testing for TTR-associated familial amyloidosis is available at Mayo Medical Laboratories by ordering AMYKM/83705 Familial Amyloidosis 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 or positive results. If results obtained do not match the

Performing Site Legend on Last Page of Report

* Report times for Mayo performed tests are CST/CDT

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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
Melody Elizabeth Kimball

Release Date 13 Jul 2012 09:56 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.