MAYO CLINIC
Mayo Medical Laboratories

Laboratory Service Report 1-800-533-1710

Patient Name	Patient ID	Age	Gender	Order #
TESTINGRNV,GSNMS	SA00048173	25	M	SA00048173
Ordering Phys				**DOB** 06/15/1987
Client Order # SA00048173	**Account Information**			**Report Notes**
Collected 08/01/2012	C7028846-DLMP ROCHESTER 3050 SUPERIOR DRIVE ROCHESTER,MN 55901			
Printed 08/30/2012 15:18				

Test	Flag	Results	Unit	Reference Value	Perform Site*
GSN Gene, Full Gene Analysis			REPORTED 08/08/2012 08:22		
Specimen		Blood			MCR
Specimen ID		1038508			MCR
Order Date		01 Aug 2012 09:37			MCR
Reason For Referral					MCR

Possible diagnosis of amyloidosis V. Test for the presence of a mutation in the gelsolin (GSN) 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 GSN gene. Mutation nomenclature is based on GenBank accession number; NM_000177.4.

Result MCR

The following sequence change was detected:
Exon: 4
DNA change: c.640G>T
Amino acid change: p.D214Y (Asp214Tyr)
Legacy nomenclature: p. D187Y (Asp187Tyr)
Classification: DELETERIOUS

Interpretation MCR

This alteration is a known deleterious mutation.

This result is consistent with a diagnosis of amyloidosis V.

Since a mutation has been identified, testing of at risk family members is possible. Mutation-specific testing for amyloidosis V is available at Mayo Medical Laboratories by ordering GSNKM/60718 GSN 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 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
W Edward Highsmith Jr., PhD					
Release Date		08 Aug 2012 08:19			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.