MAYO CLINIC
Mayo Medical Laboratories

Laboratory Service Report 1-800-533-1710

Patient Name	Patient ID	Age	Gender	Order #
TESTINGRNV,GSNKM	SA00048146	14	M	SA00048146
Ordering Phys				**DOB** 06/15/1998
Client Order # SA00048146	**Account Information**			**Report Notes**
Collected 07/31/2012	C7028846-DLMP ROCHESTER 3050 SUPERIOR DRIVE ROCHESTER,MN 55901			
Printed 08/30/2012 15:20				

Test	Flag	Results	Unit	Reference Value	Perform Site*
GSN Gene, Known Mutation			REPORTED 08/08/2012 08:22		
Specimen		Blood			MCR
Specimen ID		1038496			MCR
Order Date		31 Jul 2012 14:56			MCR
Reason For Referral					MCR

Family history of amyloidosis V. Test for the presence of a mutation in the Gelsolin (GSN) gene.

Method MCR

DNA sequence analysis was used to test for the presence of the c.640G>T (p.D214Y) mutation in exon 4 of the GSN gene. Testing was performed for this specific mutation because it was previously identified in an affected family member of this individual. 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.

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 clinical findings, additional testing should be considered.

Test results should be interpreted in context of clinical

Performing Site Legend on Last Page of Report

* Report times for Mayo performed tests are CST/CDT

Patient Name	Patient ID	Age	Gender	Order #
TESTINGRNV,GSNKM	SA00048146	14	M	SA00048146
Ordering Phys				**DOB** 06/15/1998
Client Order # SA00048146	**Account Information**			**Report Notes**
Collected 07/31/2012	C7028846-DLMP ROCHESTER 3050 SUPERIOR DRIVE ROCHESTER,MN 55901			
Printed 08/30/2012 15:20				

Test	Flag	Results	Unit	Reference Value	Perform Site*
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 W Edward Highsmith Jr., PhD					MCR
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.

* Report times for Mayo performed tests are CST/CDT