MAYO CLINIC
Mayo Medical Laboratories

Laboratory Service Report 1-800-533-1710

Patient Name	Patient ID	Age	Gender	Order #
TESTINGRNV,REPORTS	SA00049025	11D	M	SA00049025
Ordering Phys				**DOB** 09/03/2012
Client Order # SA00049025	**Account Information**			**Report Notes**
Collected 09/14/2012	C7028846-DLMP ROCHESTER 3050 SUPERIOR DRIVE ROCHESTER,MN 55901			
Printed 10/05/2012 16:46				

Test	Flag	Results	Unit	Reference Value	Perform Site*
SEPT9 Gene, Large Del/Dup			REPORTED 09/14/2012 14:26		
Specimen		Blood			MCR
Specimen ID		1038801			MCR
Order Date		14 Sep 2012 14:22			MCR
Result					MCR
Analysis has been completed. Refer to the SEPT9 Gene, Known Mutation for results and interpretation.					
Review By:		Victoria Marie Waugh			MCR
Release Date		14 Sep 2012 14:25			MCR
SEPT9 Gene, Known Mutation			REPORTED 09/14/2012 14:57		
Specimen		Blood			MCR
Specimen ID		1038801			MCR
Order Date		14 Sep 2012 14:22			MCR
Reason for Referral					MCR
Family history of hereditary neuralgic amyotrophy (HNA). Test for the presence of a mutation in the SEPT9 gene.					
Method					MCR
DNA sequence analysis was used to test for the presence of the p.R88W (c.262C>T) alteration in exon 2 of the SEPT9 gene. Testing was performed for this specific alteration because it was identified in a family member. Mutation nomenclature is based on GenBank accession number; NM_006640.4.					
Result					MCR
The R88W mutation was NOT detected.					
Interpretation					MCR

Absence of the mutation previously identified in an affected family member indicates that this individual is NOT at increased risk for developing HNA.

This assay does not rule out the presence of other mutations in this gene or other genes associated with HNA. Errors in the diagnosis or pedigree provided to us, including non paternity, may lead to an erroneous interpretation of test results.

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

Performing Site Legend on Last Page of Report

* Report times for Mayo performed tests are CST/CDT

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available upon request.

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.

Laboratory developed test.

Review By:	Victoria Marie Waugh	MCR
Release Date	14 Sep 2012 14:55	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.