MAYO CLINIC
Mayo Medical Laboratories

Laboratory Service Report 1-800-533-1710

Patient Name	Patient ID	Age	Gender	Order #
TESTINGRNV,REPORTS	SA00049024	11D	F	SA00049024
Ordering Phys				**DOB** 09/03/2012
Client Order # SA00049024	**Account Information**			**Report Notes**
Collected 09/14/2012	C7028846-DLMP ROCHESTER 3050 SUPERIOR DRIVE ROCHESTER,MN 55901			
Printed 10/05/2012 15:44				

Test	Flag	Results	Unit	Reference Value	Perform Site*
SEPT9 Gene, Mutation Screen			REPORTED 09/14/2012 14:49		
Specimen		Blood			MCR
Specimen ID		1038800			MCR
Order Date		14 Sep 2012 14:21			MCR
Reason for Referral					MCR

Possible diagnosis of hereditary neuralgic amyotrophy (HNA).
Test for the presence of a mutation in the SEPT9 gene.

Method MCR

Bi-directional sequence analysis was used to test for the presence of a mutation in the 5-prime untranslated region (5'UTR) and all coding regions and intron/exon boundaries of the SEPT9 gene. Gene dosage analysis (MLPA) was used to test for the presence of large deletions and duplications. Mutation nomenclature is based on GenBank accession number; NM_006640.4.

Result MCR

The following sequence change was detected:
Exon: 2
DNA change: c.262C>T
Amino acid change: p.R88W (Arg88Trp)
Classification: DELETERIOUS

Interpretation MCR

This alteration is a known deleterious mutation.

This result is consistent with a diagnosis of HNA.

Since a mutation has been identified, testing of at risk family members is possible. Mutation-specific testing for HNA is available at Mayo Medical Laboratories by ordering SEPTK/61101 SEPT9 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:
Test results should be interpreted in context of clinical findings, family history, and other laboratory data.

Performing Site Legend on Last Page of Report

* Report times for Mayo performed tests are CST/CDT

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Test	Flag	Results	Unit	Reference Value	Perform Site*
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 14:46			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.

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