

Patient Name TESTINGRVN,REPORTS	Patient ID SA00049026	Age 11D	Gender M	Order # SA00049026
Ordering Phys				DOB 09/03/2012
Client Order # SA00049026	Account Information			Report Notes
Collected 09/14/2012	C7028846-DLMP ROCHESTER 3050 SUPERIOR DRIVE ROCHESTER,MN 55901			
Printed 10/05/2012 16:47				

Test	Flag	Results	Unit	Reference Value	Perform Site*
SEPT9 Gene, Known Mutation			REPORTED	09/14/2012 15:01	
Specimen		Blood			MCR
Specimen ID		1038802			MCR
Order Date		14 Sep 2012 14:23			MCR
Reason for Referral		Family history of hereditary neuralgic amyotrophy (HNA). Test for the presence of a mutation in the SEPT9 gene.			MCR
Method		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.			MCR
Result		The following sequence change was detected: Exon: 2 DNA change: c.262C>T Amino acid change: p.R88W (Arg88Trp) Classification: DELETERIOUS			MCR
Interpretation		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. 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. Misinterpretation of results may occur if the information provided is inaccurate or incomplete. Rare polymorphisms exist that could lead to false negative			MCR

Performing Site Legend on Last Page of Report

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* Report times for Mayo performed tests are CST/CDT

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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 15:00	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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