

<b>Patient Name</b> SAMPLEREP,HEXKM	<b>Patient ID</b> SA00046747	<b>Age</b> 45	<b>Gender</b> F	<b>Order #</b> SA00046747
<b>Ordering Phys</b>				<b>DOB</b> 06/10/1966
<b>Client Order #</b> SA00046747	<b>Account Information</b>			<b>Report Notes</b>
<b>Collected</b> 05/21/2012	C7028846-DLMP ROCHESTER 3050 SUPERIOR DRIVE ROCHESTER,MN 55901			
<b>Printed</b> 09/15/2012 13:07				

Test	Flag	Results	Unit	Reference Value	Perform Site*
<b>HEXA Gene, Known Mutation</b>				REPORTED 07/13/2012 10:11	
Specimen		Blood			MCR
Specimen ID		1038208			MCR
Order Date		22 May 2012 15:05			MCR
Reason For Referral		Family history of Tay-Sachs disease (TSD). Test for the presence of familial alterations in the HEXA gene.			MCR
Method		DNA sequence analysis was used to test for the presence of the and p.G269S (c.805G>A) and c.1073+1G>A alterations in exon 7 and intron 9, respectively, in the HEXA gene (GenBank accession number; NM_000520.4). Analysis for these specific alterations was performed because they were identified in a family member.			MCR
Result		The p.G269S and c.1073+1G>A alterations were NOT detected.			MCR
Interpretation		Absence of the mutation(s) previously identified in an affected family member indicates that this individual is at no greater risk than someone in the general population to be a carrier of TSD.			MCR
		This assay does not rule out the presence of other disease causing mutations in this gene or other genes associated with metabolic disease. Errors in the diagnosis or pedigree provided to us, including non paternity, may lead to an erroneous interpretation of the 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 available upon request.			
		<b>CAUTIONS:</b> 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			

\*\*\*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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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.					
Extraction Performed?		Yes			MCR
Reviewed By		Melody Elizabeth Kimball			MCR
Release Date		13 Jul 2012 10:04			MCR

\* Performing Site:

MCR	Mayo Clinic Laboratories - Rochester Main Campus 200 First St SW Rochester, MN 55905	Lab Director:
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