

Patient Name SAMPLEREP,HEXMS	Patient ID SA00046785	Age 45	Gender F	Order # SA00046785
Ordering Phys				DOB 06/10/1966
Client Order # SA00046785	Account Information C7028846-DLMP ROCHESTER 3050 SUPERIOR DRIVE ROCHESTER,MN 55901			Report Notes
Collected 05/24/2012				
Printed 09/15/2012 14:37				

Test	Flag	Results	Unit	Reference Value	Perform Site*
HEXA Gene, Full Gene Analysis				REPORTED 07/13/2012 10:11	
Specimen		Blood			MCR
Specimen ID		1038224			MCR
Order Date		29 May 2012 08:39			MCR
Reason For Referral		Patient reported to have features suggestive of Tay-Sachs disease (TSD). Test for the presence of mutations in the HEXA gene.			MCR
Method		Bi-directional sequence analysis was performed to test for the presence of mutations in all coding regions and intron/exon boundaries of the HEXA gene. Mutation nomenclature is based on GenBank accession number; NM_000520.4.			MCR
Result		A mutation was NOT detected.			MCR
Interpretation		This result decreases the likelihood but does not completely exclude a diagnosis of TSD. We predict that there may be disease causing mutations in the HEXA gene not identifiable by the method described (e.g., large deletions/duplications, promoter mutations, or deep intronic mutations). This assay does not rule out the presence of disease causing mutations in other genes associated with metabolic disease. This result should be interpreted in the context of clinical findings, family history, and other laboratory testing (e.g. hexosaminidase A analysis in leukocytes). 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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<p>or positive results. If results obtained do not match the clinical findings, additional testing should be considered.</p> <p>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.</p> <p>Laboratory developed test.</p>					
Extraction Performed?		Yes			MCR
Reviewed By Melody Elizabeth Kimball					MCR
Release Date		13 Jul 2012 10:05			MCR

* Performing Site:

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