

1-800-533-1710

PATIENT NAME TESTING, GAU		PATIENT NUMBER L3MRNG9156453		AGE 20Y	SEX M	ACCESSION # G9156453
ORDERING PHYSICIAN			CLIENT ORDER #		ACCOUNT # LIAISONS	
COLLECTION 09/22/10 10:50 A	RECEIVED 09/22/10 10:50 A	REPORT PRINTED 11/22/10 03:08 P		SPECIMEN INFORMATION DATE OF BIRTH:		
DATE TIME	DATE TIME	DATE TIME				
Test Client Attn: Mayo Liaisons 200 First Street SW Rochester, MN 55905 507-284-8202						

TEST REQUESTED	HI	LO	REF RANGE	PERFORM SITE *
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Gaucher Disease, Mutation Analysis

Specimen	Blood	MCR
Specimen ID	1034719	MCR
Order Date	22 Sep 2010 13:01	MCR
Reason For Referral		MCR

Not Provided. Test for the presence of a mutation within the beta-glucosidase gene.

Method		MCR
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The multiplex PCR-based assay from TM Bioscience is used to test for the following mutations within the beta-glucosidase gene: N370S, IVS2(+1)G>A, 84G>GG, R496H, L444P, delta55bp V394L, and D409H.

Result		MCR
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None of the listed mutations were detected.

Interpretation		MCR
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Interpretation of results depends on reason for referral and ethnic background. For carrier testing and no family history of Gaucher disease, an individual of Ashkenazi Jewish ancestry has a 1/341 risk to be a carrier of a mutation not identified by this assay. This risk calculation is based on a carrier frequency of 1/18 and a mutation detection rate of 95% in the Ashkenazi Jewish population.

For individuals of non Ashkenazi Jewish ancestry, up to 60% of carriers of Gaucher disease are predicted to have a mutation detected by this assay

For testing to rule out a diagnosis of Gaucher disease, these results neither prove nor exclude the diagnosis. Less than 1% of Gaucher patients of Ashkenazi Jewish ancestry and 16% of patients of non-Ashkenazi Jewish ancestry are expected to have two mutations not identified by this assay. These values are based on a mutation detection rate of 95%

* Perform Site Legend on last page of report

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for Ashkenazi Jewish ancestry and 60% for individuals of non Ashkenazi Jewish ancestry.

Beta glucosidase enzyme analysis may provide useful diagnostic information.

CAUTIONS:

This test was developed and its performance characteristics determined by Laboratory Medicine and Pathology, Mayo Clinic. This test has not been cleared or approved by the U.S. Food and Drug Administration.

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 allogeneic donors will interfere with testing. Call Mayo Medical Laboratories for instructions for testing patients who have received a bone marrow transplant.

Reviewed By:

Heather Lynn Owen

MCR

Release Date

22 Sep 2010 13:19

MCR

* PERFORMING SITE

MCR	Mayo Clinic Dpt of Lab Med & Pathology 200 First Street SW Rochester, MN 55905	Lab Director: Franklin R. Cockerill, III, M.D.
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