

Ga

Laboratory Service Report

1-800-533-1710

Patient Name	Patient ID	Age	Gender	Order #
TESTINGRNV,GBAMS	SA00005806	8	F	SA00005806
Ordering Phys				DOB
				05/24/2004
Client Order #	Account Information			Report Notes
SA00005806				
Collected	C7028846-DLMP ROCHESTER			
05/30/2012	3050 SUPERIOR DRIVE			
Printed	ROCHESTER,MN 559	001		
05/31/2012 10:28				

Test	Flag	Results	Unit	Reference Value	Perform Site*
aucher Disease, Full Gene Analysi	s		REPORTED 0	5/30/2012 08:46	
Specimen		Blood			MCR
Specimen ID		963443			MCR
Order Date		30 May 2012 08:24			MCR
Reason For Referral					MCR
Not provided. Test for the	e presence of m	mutations in the GBA			
gene.					
Method					MCR
DNA sequence analysis was		_			
mutation in all 11 exons of					
nomenclature is based on G	enBank accession	on number; NM_			
000157.2.					
Result					MCR
A mutation was NOT detected	a.				MOD
Interpretation	likalibaad but	door not someletely			MCR
This result decreases the exclude carrier or affected					
predict that there may be					
identifiable by the method		_			
dolotions/duplications pr					

deletions/duplications, promoter mutations, or deep intronic ${\tt mutations)}\,.$ This assay does not rule out the presence of disease causing mutations in other genes associated with metabolic disease.

If testing was performed to rule out a diagnosis of Gaucher disease, this result should be interpreted in the context of clinical findings, family history, and other laboratory testing (e.g. beta-glucosidase activity level in leukocytes, blood spots, or fibroblasts). If the patient has a family history, referral for genetic counseling is recommended.

A genetic consultation may be of benefit.

A list of common polymorphisms identified for this patient is available upon request.

CAUTTONS:

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

Performing Site Legend on Last Page of Report

Patient Name	Collection Date and Time	Report Status
TESTINGRNV,GBAMS	05/30/2012	Final
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^{*} Report times for Mayo performed tests are CST/CDT



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05/31/2012 10:28				

30 May 2012 08:44

Test Flag Results Unit Value Site*

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. Reviewed By Melody Elizabeth Trimble Release Date

MCR

MCR

* Performing Site:

MCR Mayo Clinic Dpt of Lab Med & Pathology
200 First St SW Rochester, MN 55905

Lab Director: Franklin R. Cockerill, III, M.D.

Patient Name	Collection Date and Time	Report Status	
TESTINGRNV,GBAMS	05/30/2012	Final	
Page 2 of 2		** End of Report **	

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