

#### **Laboratory Service Report**

### 1-800-533-1710

Patient Name TESTINGRNV,REPORTS	Patient ID SA00064005	Age 29D	Gender F	<b>Order #</b> SA00064005	
Ordering Phys CLIENT, CLIENT				<b>DOB</b> 10/01/2013	
Client Order # SA00064005	Account Information			Report Notes	
<b>Collected</b> 10/30/2013 00:00	C7028846-DLMP Roch SDSC 2 - Client Suppo				
Printed 11/21/2013 14:23	Rochester, MN 55901	Rochester, MN 55901			

Test Flag Results Unit Value Site\*

MPSVI, Known Mutation

**RECEIVED:** 10/30/2013 14:59 **REPORTED:** 10/30/2013 16:15

Reason for Referral

MCR

Family history of mucopolysaccharidosis type VI (Maroteaux-Lamy syndrome). Test for the presence of familial alterations in the ARSB gene.

Result

MCR

The p.W322X alteration was NOT detected.

Interpretation

MCR

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 develop symptoms related to mucopolysaccharidosis type VI (MPS-VI), also known as Maroteaux-Lamy syndrome.

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 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.

### 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 or positive results. If results obtained do not match the clinical findings, additional testing should be considered. Method

DNA sequence analysis was performed to test for the presence of the p.W322X (c.966G>A) alteration in exon 5 in the ARSB gene. Testing was performed for this specific alteration because it was previously in a family member of this individual. Mutation nomenclature is based on GenBank

MCR

\*\*\*Performing Site Legend on Last Page of Report\*\*\*

Patient Name	Collection Date and Time	Report Status
TESTINGRNV,REPORTS	10/30/2013 00:00	Final
Page 1 of 2		>> Continued on Next Page >>

<sup>\*</sup> Report times for Mayo performed tests are CST/CDT



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Client Order # SA00064005	Account Information			Report Notes
<b>Collected</b> 10/30/2013 00:00	SDSC 2 - Client Suppo	C7028846-DLMP Rochester SDSC 2 - Client Support		
Printed 11/21/2013 14:23	Rochester, MN 55901			

Test	Flag	Results	Unit	Reference Value	Perform Site*
accession number NM_000046.3. Specimen Reviewed By Dimitar Gavrilov MD, PhD		Blood			MCR MCR
Release Date		30 Oct 2013 16:12			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.

Patient Name	Collection Date and Time	Report Status
TESTINGRNV,REPORTS	10/30/2013 00:00	Final
Page 2 of 2		** End of Report **

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