MAYO CLINIC
Mayo Medical Laboratories

Laboratory Service Report 1-800-533-1710

Patient Name	Patient ID	Age	Gender	Order #
TESTINGRNV,REPORTS	SA00064007	29D	F	SA00064007
Ordering Phys CLIENT,CLIENT				**DOB** 10/01/2013
Client Order # SA00064007	**Account Information**			**Report Notes**
Collected 10/30/2013 13:28	C7028846-DLMP Rochester SDSC 2 - Client Support Rochester, MN 55901			
Printed 11/21/2013 14:24				

Test	Flag	Results	Unit	Reference Value	Perform 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 following homozygous sequence change was detected:
Exon: 5
DNA change: c.966G>A
Amino Acid change: p.W322X (Trp322X)
Classification: DELETERIOUS

Interpretation MCR

The p.W322X alteration is a known deleterious mutation.

This result is consistent with the diagnosis of mucopolysaccharidosis type VI (MPS-VI), also known as Maroteaux-Lamy syndrome.

Since mutations have been identified in the ARSB gene, genetic testing of at risk family members is possible.

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.

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.

Performing Site Legend on Last Page of Report

* Report times for Mayo performed tests are CST/CDT

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Test	Flag	Results	Unit	Reference Value	Perform Site*
Laboratory developed test.					
Method					MCR
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 accession number NM_000046.3.					
Specimen		Blood			MCR
Reviewed By					MCR
Dimitar Gavrilov MD, PhD					
Release Date		30 Oct 2013 16:13			MCR

* Performing Site:

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