

#### **Laboratory Service Report**

### 1-800-533-1710

Patient Name TESTINGRNV,REPORTS	Patient ID SA00062306	Age 9D	Gender F	<b>Order #</b> SA00062306	
Ordering Phys CLIENT,CLIENT				<b>DOB</b> 09/02/2013	
Client Order # SA00062306	Account Information			Report Notes	
<b>Collected</b> 09/11/2013 00:00	SDSC 2 - Client Suppor	C7028846-DLMP Rochester SDSC 2 - Client Support			
<b>Printed</b> 10/03/2013 09:11	Rochester, MN 55901				

Reference Perform Test Flag Results Unit Value Site\*

SUMF1 Gene, Full Gene Analysis

**RECEIVED:** 09/11/2013 11:59 **REPORTED:** 09/16/2013 10:08

Reason for Referral

MCR Possible diagnosis of multiple sulfatase deficiency (MSD).

Test for the presence of mutations in the SUMF1 gene.

Result

MCR A mutation was NOT detected.

Interpretation MCR This result decreases the likelihood but does not rule out

the diagnosis of multiple sulfatase deficiency (MSD). We predict that a small percentage of individuals who have a diagnosis of MSD may have mutations that are not identified by the methods 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 that are associated with sulfatase deficiencies.

This result should be interpreted in the context of clinical findings, family history, and other laboratory testing (e.g. urine sulfatide analysis, iduronate sulfatase and arylsulfatase A and B enzyme levels).

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

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

Patient Name	Collection Date and Time	Report Status
TESTINGRNV,REPORTS	09/11/2013 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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Test	Flag Results	Unit	Reference Value	Perform Site*
marrow transplant.				
the presence of mutations	analysis was performed to test s in all coding regions and t the SUMF1 gene. Mutation	for		MCR
Specimen Reviewed By Release Date	Blood Devin Oglesbee 16 Sep 2013 10:			MCR MCR MCR

### \* Performing Site:

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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	09/11/2013 00:00	Final
Page 2 of 2		** End of Report **

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