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

Test	Flag	Results	Unit	Reference Value	Perform 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

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Test	Flag	Results	Unit	Reference Value	Perform Site*
marrow transplant. Laboratory developed test.					
Method		Bi-directional sequence analysis was performed to test for the presence of mutations in all coding regions and intron/exon boundaries of the SUMF1 gene. Mutation nomenclature is based on GenBank accession number NM_182760.3.			MCR
Specimen		Blood			MCR
Reviewed By		Devin Oglesbee PhD			MCR
Release Date		16 Sep 2013 10:05			MCR

* Performing Site:

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