Patient Name	Patient ID	Age	Gender	Order #
TESTINGRNV,REPORTS	SA00062309	9D	M	SA00062309
Ordering Phys CLIENT,CLIENT				**DOB** 09/02/2013
Client Order # SA00062309	**Account Information**			**Report Notes**
Collected 09/11/2013 00:00	C7028846-DLMP Rochester SDSC 2 - Client Support Rochester, MN 55901			
Printed 10/03/2013 09:15				

Test	Flag	Results	Unit	Reference Value	Perform Site*

SUMF1 Gene, Known Mutation
RECEIVED: 09/11/2013 12:00 **REPORTED:** 09/16/2013 10:09

Reason for Referral MCR

Family history of multiple sulfatase deficiency (MSD). Test for the presence of mutations in the SUMF1 gene.

Result MCR

The following heterozygous sequence change was detected:
Exon: 8
DNA change: c.979C>T
Amino Acid change: p.R327X (Arg327X)
Classification: DELETERIOUS

Interpretation MCR

This alteration is a known deleterious mutation.

This result indicates that this individual is a carrier of multiple sulfatase deficiency (MSD). This interpretation assumes this individual is healthy and not clinically affected with MSD.

Since a mutation has been identified, testing of at risk family members is possible. If appropriate, genetic testing should be offered to this individual's reproductive partner to clarify their risk of having a child with MSD.

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

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instructions for testing patients who have received a bone marrow transplant. Laboratory developed test.					
Method					MCR
DNA sequence analysis was performed to test for the presence of the p.R327X (c.979C>T) alteration in exon 8 of the SUMF1 gene. Testing was performed for this specific alteration because it was previously identified in a family member of this individual. Mutation nomenclature is based on GenBank accession number NM_182760.3.					
Specimen		Blood			MCR
Reviewed By		Devin Oglesbee PhD			MCR
Release Date		16 Sep 2013 10:06			MCR

* Performing Site:

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