

Patient ID <b>SA00000884</b>	Patient Name <b>SAMPLEREPDMGLM, VLD20150713A0145</b>	Birth Date <b>1981-01-01</b>	Gender <b>M</b>	Age <b>34</b>
Order Number <b>SA00000884</b>	Client Order Number <b>SA00000884</b>	Ordering Physician <b>CLIENT, CLIENT</b>	Report Notes	
Account Information <b>C7028846 DLMP Rochester</b>		Collected <b>12 Jul 2015 16:23</b>		

## Result Summary

MCR

### Positive

## Result

MCR

The following heterozygous alteration was identified:

**Amino Acid change:** p.S155P (Ser155Pro)

**DNA change:** c.463T>C (g.4491006)

**Classification:** PATHOGENIC

## Interpretation

ⓘ MCR

The c.463T>C (p.S155P) alteration is a known pathogenic mutation.

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

Since a mutation has been identified, genetic testing of at risk family members could be considered. Mutation specific testing is available at Mayo Medical Laboratories by ordering FMTT / Familial Mutation, Targeted Testing. Please contact the MOlecular Genetics Laboratory at 1-800-533-1710 with questions about this test.

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.

### ADDITIONAL INFORMATION

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 (GenBank accession number NM\_182760; build GRCh37 (hg19)). An online research opportunity called GenomeConnect (genomeconnect.org), a project of ClinGen, is available for the recipient of this genetic test. This patient registry collects de-identified genetic and health information to advance the knowledge of genetic variants. Mayo Clinic is a collaborator of ClinGen. This may not be applicable for all tests.

Test results should be interpreted in the 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 false-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 Clinic Laboratories for instructions for testing patients who have received a bone marrow transplant.

Multiple in-silico evaluation tools may have been used to assist in the interpretation of these results. Of note, the sensitivity and specificity of these tools for the determination of pathogenicity is currently unvalidated.

### Performing Site Legend

Code	Laboratory	Address	Lab Director	CLIA Certificate
MCR	Mayo Clinic Laboratories - Rochester Main Campus	200 First Street SW, Rochester, MN 55905	William G. Morice M.D. Ph.D	24D0404292



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**Specimen**

WB Whole Blood

MCR

**Released By**

EMILY LAUER

MCR

**Received:** 13 Jul 2015 20:23

**Reported:** 24 Jul 2015 16:43

Test Environment  
ETBM Template

**Laboratory Notes**

- 1 This test was developed and its performance characteristics determined by Mayo Clinic in a manner consistent with CLIA requirements. This test has not been cleared or approved by the U.S. Food and Drug Administration.

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