Patient Name	Patient ID	Age	Gender	Order #
TESTINGRNV,LABELS	SA00057598	20D	F	SA00057598
Ordering Phys CLIENT,CLIENT				**DOB** 05/01/2013
Client Order # SA00057598	**Account Information**			**Report Notes**
Collected 05/21/2013 00:00	C7028846-DLMP Rochester 3050 Superior Drive Rochester, MN 55901			
Printed 06/25/2013 13:44				

Test	Flag	Results	Unit	Reference Value	Perform Site*

Hunter Syndrome, Known Mutation
RECEIVED: **REPORTED:** 05/24/2013 08:50

Reason for Referral MCR

Family history of mucopolysaccharidosis type II (Hunter syndrome). Test for the presence of the familial alteration in the IDS gene.

Result MCR

The following heterozygous sequence change was detected:
Exon: 9
DNA change: c.1591C>T
Amino Acid change: p.Q531X (Gln531X)
Classification: DELETERIOUS

Interpretation MCR

This alteration is a known deleterious mutation.

The presence of a mutation previously identified in a family member indicates that this individual is a carrier of mucopolysaccharidosis type II (MPS-II), also known as Hunter syndrome. Studies suggest that on rare occasions female carriers of IDS mutations may be at risk for developing symptoms associated with MPS-II.

Since a mutation has been identified in the IDS 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:
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.

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.

Bone marrow transplants from allogenic donors will interfere

Performing Site Legend on Last Page of Report

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Test	Flag	Results	Unit	Reference Value	Perform Site*
with testing. Call Mayo Medical Laboratories for instructions for testing patients who have received a bone marrow transplant.					
Laboratory developed test.					
Method					MCR
DNA sequence analysis was used to test for the presence of the p.Q531X (c.1591C>T) alteration in exon 9 of the IDS 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_000202.5.					
Specimen		Blood			MCR
Reviewed By		Devin Oglesbee PhD			MCR
Release Date		24 May 2013 08:48			MCR

* Performing Site:

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