Patient Name	Patient ID	Age	Gender	Order #
TESTINGRNV,REPORTS	SA00057594	20D	F	SA00057594
Ordering Phys CLIENT,CLIENT				**DOB** 05/01/2013
Client Order # SA00057594	**Account Information**			**Report Notes**
Collected 05/21/2013 00:00	C7028846-DLMP Rochester 3050 Superior Drive Rochester, MN 55901			
Printed 06/25/2013 14:29				

Test	Flag	Results	Unit	Reference Value	Perform Site*

Hunter Syndrome, Full Gene Analysis
RECEIVED: **REPORTED:** 05/24/2013 08:50

Reason for Referral MCR

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

Result MCR

A mutation was NOT detected.

Interpretation MCR

This result decreases the likelihood but does not rule out carrier status for mucopolysaccharidosis type II (MPS-II), also known as Hunter syndrome. We predict that there may be disease causing mutations in the IDS gene that are not identified by the method 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 associated with metabolic disease.

If testing was performed to rule out a diagnosis of MPS-II, this result should be interpreted in the context of clinical findings, family history, and other laboratory testing (e.g. iduronate 2-sulfatase analysis in fibroblasts).

It is often helpful to test the affected individual or an obligate carrier. Identification of the mutation in this family would allow for more accurate risk assessment of at risk individuals.

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

Performing Site Legend on Last Page of Report

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Test	Flag	Results	Unit	Reference Value	Perform Site*
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 marrow transplant.					
Laboratory developed test.					
Method					MCR
Bi-directional sequence analysis was performed to test for the presence of mutations in all coding regions and intron/exon boundaries of the IDS gene. Additionally, a PCR-based assay was utilized to test for the presence of rearrangements between the IDS gene and pseudogene, IDSP1. 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: