MAYO CLINIC
Mayo Medical Laboratories

Laboratory Service Report 1-800-533-1710

Patient Name	Patient ID	Age	Gender	Order #
TESTINGRNV,REPORTS	SA00057595	20D	M	SA00057595
Ordering Phys CLIENT,CLIENT				**DOB** 05/01/2013
Client Order # SA00057595	**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

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

Result MCR

The following hemizygous 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.

This result is consistent with a diagnosis of mucopolysaccharidosis type II (MPS-II), also known as Hunter syndrome.

Since a mutation has been identified, testing of at risk family members is possible. Mutation-specific testing for MPS-II is available at Mayo Medical Laboratories by ordering HUNTK/61496 Hunter Syndrome, Known Mutation. Please contact the Molecular Genetics Laboratory at 1-800-533-1710 with questions about this test.

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.

Performing Site Legend on Last Page of Report

* Report times for Mayo performed tests are CST/CDT

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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: Franklin R. Cockerill, III, M.D.