

Laboratory Service Report

1-800-533-1710

| Patient Name | Patient ID | Age | Gender | Order # |
|-----------------------------------|--|-----|--------|-----------------------|
| TESTINGRNV,REPORTS | SA00057597 | 20D | F | SA00057597 |
| Ordering Phys CLIENT,CLIENT | | | | DOB 05/01/2013 |
| Client Order # SA00057597 | Account Information | | | Report Notes |
| Collected 05/21/2013 00:00 | C7028846-DLMP Rochester 3050 Superior Drive | | | |
| Printed 06/25/2013 13:38 | Rochester, MN 55901 | | | |

Test Flag Results Unit Value Site*

Hunter Syndrome, Known Mutation

ECEIVED: 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 p.Q531X alteration was NOT detected.

Interpretation

MCR

Absence of the mutation previously identified in an affected family member indicates that this individual is at no greater risk than someone in the general population to be a carrier of or affected with mucopolysaccharidosis type II, also known as Hunter syndrome.

This assay does not rule out the presence of other disease causing mutations in this gene or other genes associated with metabolic disease. Errors in the diagnosis or pedigree provided to us may lead to an erroneous interpretation of test results.

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 with testing. Call Mayo Medical Laboratories for instructions for testing patients who have received a bone marrow transplant.

Performing Site Legend on Last Page of Report

| Patient Name | Collection Date and Time | Report Status |
|--------------------|--------------------------|------------------------------|
| TESTINGRNV,REPORTS | 05/21/2013 00:00 | Final |
| Page 1 of 2 | | >> Continued on Next Page >> |



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|--------------------------|------------------------------------|------|--------------------|------------------|
| Laboratory developed tes | st. | | | |
| Method | | | | MCR |
| DNA sequence analysis wa | as used to test for the presence | of | | |
| ± ~ ' | alteration in exon 9 of the IDS | | | |
| 3 3 1 | ormed for this specific alteration | | | |
| - | y identified in a family member | | | |
| | on nomenclature is based on GenB | ank | | |
| accession number NM_0002 | | | | |
| Specimen | Blood | | | MCR |
| Reviewed By | Devin Oglesbee Ph | nD | | MCR |
| Release Date | 24 May 2013 08:48 | 3 | | MCR |

* Performing Site:

| MCR | Mayo Clinic Laboratories - Rochester Main Campus | Lab Director: Franklin R. Cockerill, III, M.D. |
|-----|--|--|
| 1 - | 200 First St SW Rochester MN 55905 | |

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| TESTINGRNV,REPORTS | 05/21/2013 00:00 | Final |
| Page 2 of 2 | | ** End of Report ** |