MAYO CLINIC
Mayo Medical Laboratories

# Laboratory Service Report 1-800-533-1710

| Patient Name | Patient ID | Age | Gender | Order # |
|---|---|---|---|---|
| TESTINGRNV,REPORTS | SA00053769 | 9D | M | SA00053769 |
| **Ordering Phys** CLIENT,CLIENT | | | | **DOB** 02/04/2013 |
| **Client Order #** SA00053769 | **Account Information** | | | **Report Notes** |
| **Collected** 02/13/2013 | C7028846-DLMP Rochester<br>3050 Superior Drive<br>Rochester, MN 55901 | | | |
| **Printed** 03/06/2013 15:38 | | | | |

| Test | Flag | Results | Unit | Reference Value | Perform Site* |
|---|---|---|---|---|---|
| **Hurler Syndrome, Full Gene Analysis** | | | REPORTED 02/14/2013 11:23 | | |
| Reason for Referral | | | | | MCR |
| Patient reported to have features suggestive of mucopolysaccharidosis type I (Hurler/Scheie). Test for the presence of mutations in the IDUA gene. | | | | | |
| Result | | | | | MCR |
| The following homozygous sequence change was detected:<br>Exon: 9<br>DNA change: c.1205G>A<br>Amino Acid change: p.W402X (Trp402X)<br>Classification: DELETERIOUS | | | | | |
| Interpretation | | | | | MCR |
| The p.W402X alteration is a known deleterious mutation.<br><br>This result is consistent with a diagnosis of mucopolysaccharidosis type I (MPS-I).<br><br>MPS-I can be categorized into Hurler syndrome, Scheie syndrome, and Hurler-Scheie syndrome. Correlation of these results with biochemical and clinical findings is recommended.<br><br>Since mutations have been identified, testing of at risk family members is possible. Mutation-specific testing for MPS-I is available at Mayo Medical Laboratories by ordering HURLK/61482 Hurler Syndrome, Known Mutation. Please contact the Molecular Genetics Laboratory at 1-800-533-1710 with questions about this test.<br><br>A genetic consultation may be of benefit.<br><br>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.<br><br>CAUTIONS:<br>Test results should be interpreted in the context of clinical findings, family history, and other laboratory data. Errors in our interpretation of results may occur if information given is inaccurate or incomplete.<br><br>Rare polymorphisms exist that could lead to false-negative | | | | | |

***Performing Site Legend on Last Page of Report***



* Report times for Mayo performed tests are CST/CDT

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| or false-positive results. If results obtained do not match the clinical findings, additional testing should be considered. | | | | | |
| A previous bone marrow transplant from an allogenic donor 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 IDUA gene. Mutation nomenclature is based on GenBank accession number NM_000203.3. | | | | | |
| Specimen | | Blood | | | MCR |
| Reviewed By | | | | | MCR |
| Dimitar Gavrilov MD, PhD | | | | | |
| Release Date | | 14 Feb 2013 11:20 | | | MCR |

* Performing Site:

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