MAYO CLINIC
Mayo Medical Laboratories

# Laboratory Service Report 1-800-533-1710

| Patient Name | Patient ID | Age | Gender | Order # |
|---|---|---|---|---|
| SAMPLEREPORT,MUGS A | M0002174 | 40 | F | M0002174 |
| **Ordering Phys** | | | | **DOB** 09/28/1971 |
| **Client Order #** M0002174 | **Account Information** | | | **Report Notes** |
| **Collected** 06/27/2012 01:51 | C7028846-DLMP ROCHESTER<br>3050 SUPERIOR DRIVE<br>ROCHESTER,MN 55901 | | | |
| **Printed** 02/20/2013 16:52 | | | | |

| Test | Flag | Results | Unit | Reference Value | Perform Site* |
|---|---|---|---|---|---|
| | | | REPORTED 02/20/2013 16:19 | | |
| **Hexosaminidase A (MUGS), S** | L | 0.00 | U/L | | MCR |

In this sample, the activity of hexosaminidase A is reduced. This result indicates this patient is affected with Tay-Sachs disease (OMIM 272800). If previous hexosaminidase A testing has been performed in leukocytes and/or serum and was within normal limits, this may indicate that this individual is affected with the B1 variant of Tay-Sachs disease (Tutor JC. Braz J Med Biol Res. 2004 Jun: 37(6):777-83). Consider submitting a new blood sample for molecular genetic sequencing analysis (MML 89278) to confirm the diagnosis and to facilitate testing of at-risk family members. A genetics consultation is recommended. In addition, the National Tay-Sachs and Allied Diseases Association (1-800-906-8723; http://www.ntsad.org/) provides information and support services for families and health care professionals. Please contact the Biochemical Genetics consultant or genetic counselor on call (1-800-533-1710) if you have any questions.
-- REFERENCE VALUE --
1.23-2.59 (Normal)
1.16-1.22 (Intermediate)
0.58-1.15 (Carrier)
Fluorometric Enzyme Assay

* Performing Site:

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

| Patient Name | Collection Date and Time | Report Status |
|---|---|---|
| SAMPLEREPORT,MUGS A | 06/27/2012 01:51 | Final |
| Page 1 of 1 | | ** End of Report ** |

* Report times for Mayo performed tests are CST/CDT

TEST SITE