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|-------------------------------------------------------|---------------------------------------------|---------------------------------------|--------------------|------------------|
| Patient ID<br><b>M0002174</b>                         | Patient Name<br><b>SAMPLEREPORT, MUGS A</b> | Birth Date<br><b>1971-09-28</b>       | Gender<br><b>F</b> | Age<br><b>40</b> |
| Order Number<br><b>M0002174</b>                       | Client Order Number<br><b>M0002174</b>      | Ordering Physician                    | Report Notes       |                  |
| Account Information<br><b>C7028846 DLMP Rochester</b> |                                             | Collected<br><b>27 Jun 2012 01:51</b> |                    |                  |

Hexosaminidase A (MUGS), S

MCR



0.00 U/L

Low

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)

ADDITIONAL INFORMATION

Fluorometric Enzyme Assay

Received: 27 Jun 2012 01:51

Reported: 20 Feb 2013 16:19

Performing Site Legend

| Code | Laboratory                                       | Address                                  | Lab Director                | CLIA Certificate |
|------|--------------------------------------------------|------------------------------------------|-----------------------------|------------------|
| MCR  | Mayo Clinic Laboratories - Rochester Main Campus | 200 First Street SW, Rochester, MN 55905 | William G. Morice M.D. Ph.D | 24D0404292       |