



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

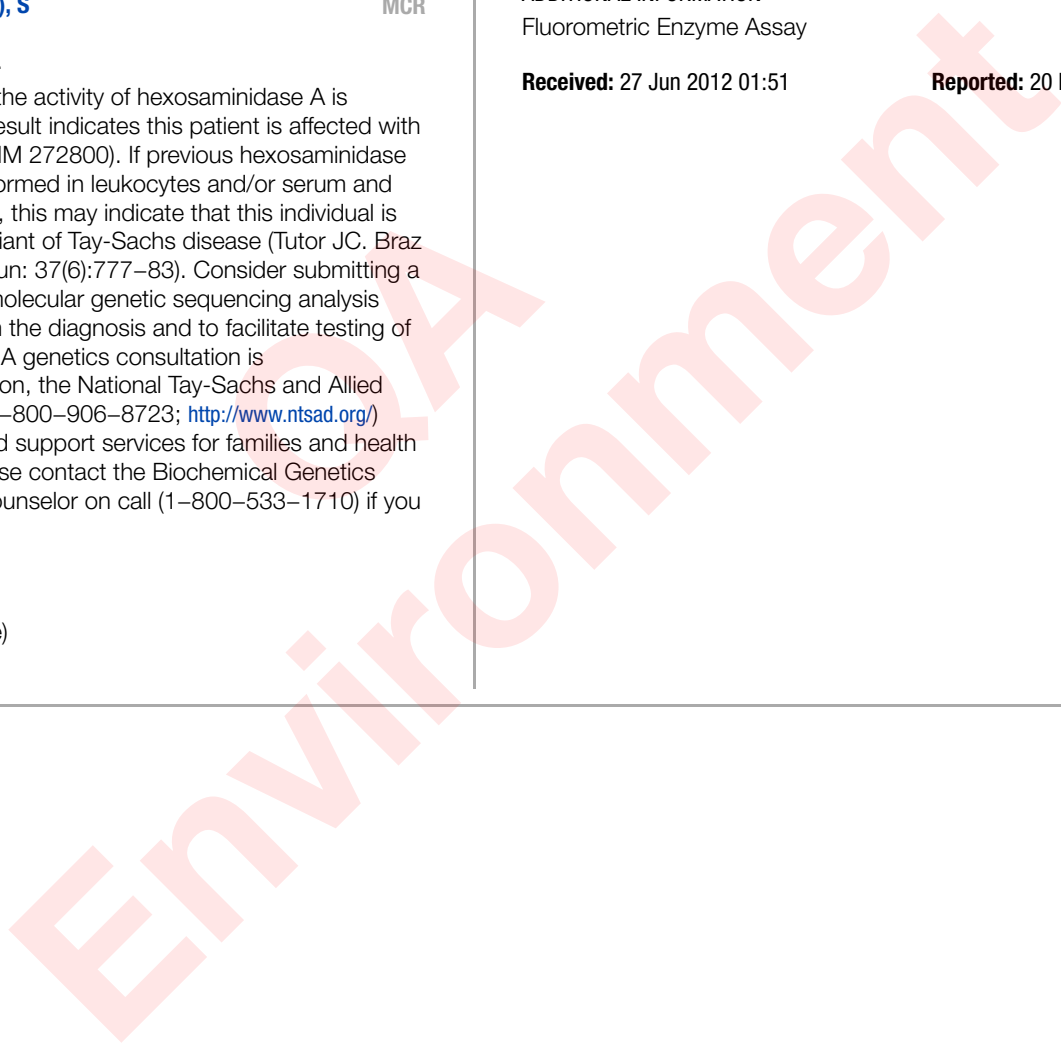
- 1.23-2.59 (Normal)
- 1.16-1.22 (Intermediate)
- 0.58-1.15 (Carrier)

ADDITIONAL INFORMATION

Fluorometric Enzyme Assay

**Received:** 27 Jun 2012 01:51

**Reported:** 20 Feb 2013 16:19



**Performing Site Legend**

Code	Laboratory	Address
MCR	Mayo Clinic Dept. of Lab Med and Pathology	200 First Street SW, Rochester, MN 55905