

1-800-533-1710

PATIENT NAME			PATIENT NU	MBER		AGE	SEX	ACCESSION #
TESTING, 8775			L3MRNG91	L3MRNG9165440		34	F	G9165440
ORDERING PHYSICIAN			CLIENT ORD	CLIENT ORDER #				ACCOUNT # LIAISONS
COLLECTION	RECEIVED		REPORT PR	INTED	SPECIMEN INFORMA	TION		
02/25/11 09:39 A	02/25/11 0	9:39 A	03/10/11	02:34 P	DATE OF BIRTH:			
DATE TIME	DATE	TIME	DATE	TIME				
Test Client								
Attn: Mayo Liaisons								
200 First Street SW								
Rochester, MN 55905								
507-284-8202								

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TEST REQUESTED LO REF RANGE PERFORM SITE*

Hexosaminidase A and Total, WBC

REPORTED: 02/25/11 10:50 A

Hexosaminidase A and

19.7

U/g Prot

MCR

Total, WBC

-- EXPECTED VALUES --

16.4-36.2 U/g of cellular protein

Percent A 67 % MML Comment

63-75 MCR MCR

In this sample, total hexosaminidase activity and percent hexosaminidase A are normal. These results indicate this individual is NOT affected with Tay-Sachs disease (OMIN 272800) and suggest that this individual is NOT a carrier for Tay-Sachs disease or Sandhoff disease. However, please note that a small percentage (<0.5%) of carriers may exhibit normal hexosaminidase A activity and will not be detected by this method (Triggs-Raine et al. NEJM. 1990; 323:6-12). In addition, patients with or carriers of the AB variant (OMIM 272750) or late-onset Tay-Sachs disease (B1 variant OMIM 272800) will not be detected. If this patient shows symptoms of later-onset Tay-Sachs disease (>=2 years), consider ordering Hexosaminidase A (MUGS), Serum (MML 80350) to rule out the B1 variant. Please contact the Biochemical Genetics consultant or genetic counselor on call (1-800-533-1710) to discuss remaining residual risk or questions you may have.

* PERFORMING SITE

MCR Mayo Clinic Dpt of Lab Med & Pathology
Lab Director: Franklin R. Cockerill, III, M.D.
200 First Street SW Rochester, MN 55905

PATIENT NAME	ORDER STATUS	COLLECTION DATE AND TIME
TESTING, 8775	Final	02/25/11 09:39 A