



Hexosaminidase A and Total, Leukocytes/
Molecular Reflex

Patient ID SA00035505	Patient Name TESTINGRNV, NAGRNORMAL	Birth Date 1971-09-12	Gender F	Age 41
Order Number SA00035505	Client Order Number SA00035505	Ordering Physician CLIENT,CLIENT	Report Notes	
Account Information C7028846 DLMP Rochester		Collected 20 Feb 2013 00:00		

Hexosaminidase A and Tot, WBC/Mole

Hexosaminidase A and Total, WBC

16.4 nmol/min/mg

MCR

Reference Value
16.4–36.2

Percent A

63 %

MCR

Reference Value
63–75

MML Comment

MCR

In this sample, total hexosaminidase activity and percent hexosaminidase A are normal. Automatic reflex to molecular genetic testing will NOT be performed. If desired, please call MML within one month to add Tay-Sachs Disease, Mutation Analysis, HEXA (MML 82588).

These results indicate this individual is NOT affected with Tay-Sachs disease (OMIM 272800) and suggest that this individual is

NOT a carrier for Tay-Sachs 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 (≥ 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) if you have any questions.

Reviewed by

MCR

JANICE HELGESON

Received: 21 Feb 2013 13:12

Reported: 21 Feb 2013 13:38

Performing Site Legend

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