



Hexosaminidase A and Total Hexosaminidase, Leukocytes

Patient ID SA00036093	Patient Name TESTINGRNV, NAGWNORMAL	Birth Date 1985-02-11	Gender F	Age 28
Order Number SA00036093	Client Order Number SA00036093	Ordering Physician CLIENT,CLIENT	Report Notes	
Account Information C7028846 DLMP Rochester		Collected 24 Feb 2013 00:00		

Hexosaminidase A and Total, WBC

Hexosaminidase Total, WBC

36.2 nmol/min/mg

MCR
Reference Value
16.4–36.2

Hexosaminidase Percent A, WBC

75 %

MCR
Reference Value
63–75

Interpretation (NAGW)

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 (OMIM 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) if you have any questions.

ADDITIONAL INFORMATION

Heat Inactivation, Fluorometric

Reviewed By

MCR

JANICE HELGESON

Received: 25 Feb 2013 07:49

Reported: 25 Feb 2013 09:29

Performing Site Legend

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