



Hexosaminidase A and Total
Hexosaminidase, Serum

Patient ID SA00052550	Patient Name TESTINGRNV, REPORTNORM	Birth Date 1990-02-02	Gender M	Age 22
Order Number SA00052550	Client Order Number SA00052550	Ordering Physician Client, Client	Report Notes	
Account Information C7028846 DLMP Rochester		Collected 07 Jan 2013 00:00		

Hexosaminidase A and Total, S

Hexosaminidase Total, S

23.8 nmol/min/mL

MCR
Reference Value
10.4–23.8

Hexosaminidase Percent A, S

80 %

MCR
Reference Value
56–80

Interpretation (NAGS)

In this sample, total hexosaminidase activity and percent hexosaminidase A are normal. These results indicate that 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. If not already ordered, this test can be added on to the existing sample by calling MML within one month.

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

JANICE HELGESON

MCR

Received: 08 Jan 2013 13:40

Reported: 08 Jan 2013 14:27

Performing Site Legend

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