

Laboratory Service Report

1-800-533-1710

Patient Name TESTINGRNV,REPORTNORM	Patient ID SA00052550	Age 22	Gender M	Order # SA00052550	
Ordering Phys CLIENT,CLIENT				DOB 02/02/1990	
Client Order # SA00052550	Account Information			Report Notes	
Collected 01/07/2013	C7028846-DLMP ROO 3050 SUPERIOR DRI	VE			
Printed 01/21/2013 14:09	ROCHESTER,MN 559	901			

Test	Flag	Results	Unit	Reference Value	Perform Site*
Hexosaminidase A and Total, S			REPORTED 01/0	8/2013 14:27	
Hexosaminidase Total, S		23.8	nmol/min/mL	10.4-23.8	MCR
Hexosaminidase Percent A, S		80	%	56-80	MCR
Interpretation (NAGS)					MCR

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.

Heat Inactivation, Fluorometric

Reviewed By JANICE HELGESON MCR

* Performing Site:

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MCR	Mayo Clinic Laboratories - Rochester Main Campus 200 First St SW Rochester, MN 55905	Lab Director: Franklin R. Cockerill, III, M.D.

	Report Status Final
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^{*} Report times for Mayo performed tests are CST/CDT