

Laboratory Service Report

1-800-533-1710

Patient Name TESTINGRNV,REPORTNORM	Patient ID SA00052547	Age 27	Gender F	Order # SA00052547
Ordering Phys CLIENT,CLIENT			•	DOB 02/11/1985
Client Order # SA00052547	Account Information	Account Information		Report Notes
Collected 01/07/2013	C7028846-DLMP RO 3050 SUPERIOR DRI			
Printed 01/21/2013 14:15	ROCHESTER,MN 55	901		

Test	Flag Results	Unit	Reference Value	Perform Site*
Hexosaminidase A and Total, WBC		REPORTED 01/	08/2013 13:57	
Hexosaminidase Total, WBC	36.2	nmol/min/mg	16.4-36.2	MCR
Hexosaminidase Percent A, WBC	75	%	63-75	MCR
Interpretation (NAGW)				MCR
In this sample, total hexosaminio	dase activity and	percent		
hexosaminidase A are normal. The	se results indica	te this		
individual is NOT affected with 7	Tay-Sachs disease	(OMIM		
272800) and suggest that this inc	dividual is NOT a	carrier		
for Tay-Sachs disease or Sandhoff	disease. Howeve	r, 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.

Heat Inactivation, Fluorometric

Reviewed By JANICE HELGESON MCR

* Performing Site:

MCR	Mayo Clinic Laboratories - Rochester Main Campus 200 First St SW Rochester, MN 55905	Lab Director:	

Patient Name Collection Date and Time		Report Status	
TESTINGRNV,REPORTNORM	01/07/2013	Final	
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^{*} Report times for Mayo performed tests are CST/CDT