

Patient Name TESTINGRNV,REPORTABNORM	Patient ID SA00052548	Age 3	Gender M	Order # SA00052548
Ordering Phys CLIENT,CLIENT				DOB 07/18/2009
Client Order # SA00052548	Account Information			Report Notes
Collected 01/07/2013	C7028846-DLMP ROCHESTER 3050 SUPERIOR DRIVE ROCHESTER,MN 55901			
Printed 01/21/2013 14:16				

Test	Flag	Results	Unit	Reference Value	Perform Site*
Hexosaminidase A and Total, WBC					
Hexosaminidase Total, WBC	L	19.0	nmol/min/mg	>=20	MCR
Hexosaminidase Percent A, WBC	L	19	%	20-80	MCR
Interpretation (NAGW) These results indicate that this patient is affected with Tay-Sachs disease (OMIM 272800). The percentage of hexosaminidase A activity is reduced. Consider submitting a new blood sample for molecular genetic analysis (MML 82588) to confirm the diagnosis and to facilitate testing of at-risk family members. A genetics consultation is recommended. In addition, the National Tay-Sachs and Allied Diseases Association (1-800-906-8723; http://www.ntsad.org/) provides information and support services for families and health care professionals. 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:
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Patient Name TESTINGRNV,REPORTABNORM	Collection Date and Time 01/07/2013	Report Status Final
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* Report times for Mayo performed tests are CST/CDT