





Hexosaminidase A and Total
Hexosaminidase, Serum

Patient ID SA00052554	Patient Name TESTINGRNV, REPORTABNORMAL	Birth Date 2008-12-25	Gender M	Age 4
Order Number SA00052554	Client Order Number SA00052554	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

 **19.0 nmol/min/mL**


MCR

Reference Value
≥20

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.

Hexosaminidase Percent A, S

 **19 %**


MCR

Reference Value
20-90

ADDITIONAL INFORMATION

Heat Inactivation, Fluorometric

Interpretation (NAGS)

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

MCR

Reviewed By

JANICE HELGESON

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Received: 08 Jan 2013 13:41

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