

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

Low

MCR

Reference Value
≥20

Hexosaminidase Percent A, S


19 %

Low

MCR

Reference Value
20–90

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

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.

ADDITIONAL INFORMATION

Heat Inactivation, Fluorometric

Reviewed By

JANICE HELGESON

MCR

Received: 08 Jan 2013 13:41

Reported: 08 Jan 2013 14:27

Performing Site Legend

Code	Laboratory	Address	Lab Director	CLIA Certificate
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