Lab Dept:	**Chemistry**
Test Name:	**HEXOSAMINIDASE A & TOTAL HEXOSAMINIDASE, SERUM**

General Information

Lab Order Codes:	HEXAT
Synonyms:	B-N-Acetylglucosaminidase; GM2 Gangliosidosis; Sandhoff Carrier Screening; Tay Sachs Carrier Screening
CPT Codes:	83080 x2 – b-Hexosaminidase, each assay
Test Includes:	Total Hexosaminidase level reported in nmol/min/mL, Hexoaminidase A reported as a % of total.

Logistics

Test Indications:	Diagnosing Tay-Sachs disease, carriers of Tay-Sachs, Sandhoff's disease, and carriers of Sandhoff's disease.
Lab Testing Sections:	Chemistry - Sendouts
Referred to:	Mayo Medical Laboratories (MML Test: NAGS)
Phone Numbers:	MIN Lab: 612-813-6280 STP Lab: 651-220-6550
Test Availability:	Daily, 24 hours
Turnaround Time:	8 – 15 days, performed on Tuesdays
Special Instructions:	For females of child-bearing age: If females wish screening for carrier status of hexoaminidase A deficiency, they must be tested prior to pregnancy. Serum assay results are often ambiguous on pregnant females and will not be run.

Specimen

Specimen Type:	Blood
Container:	Red top tube
Draw Volume:	3 mL (Minimum: 1.5 mL) blood

Processed Volume: 1 mL (Minimum: 0.5 mL) serum

Collection: Routine venipuncture

Special Processing: Lab Staff: Centrifuge specimen. Remove serum aliquot and place into a screw-capped plastic vial. Ship and store at frozen temperatures.

Patient Preparation: None

Sample Rejection: Warm specimens; samples from pregnant females; mislabeled or unlabeled specimens

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Interpretive

Reference Range:

Hexosaminidase A and Total, Serum (nmol/min/mL)	
≤15 years:	≥20 nmol/min/mL
≥16 years:	10.4 – 23.8 nmol/min/mL
Percent A (%)	
≤15 years:	20 – 90%
≥16 years:	56 – 80%
Interpretation is provided with report.	

Critical Values: N/A

Limitations: This test cannot be performed on pregnant females.

GM2 activator deficiency (CM2-gangliosidosis, AB variant) is a rare disorder with clinical features similar to Tay-Sachs and Sandhoff diseases; however, levels of both hexosaminidase A and B are normal. GM2 activator deficiency cannot be identified through testing offered at Mayo Medical Laboratories.

Methodology: Heat inactivation, Fluorometric, Automated

References: Mayo Medical Laboratories July 2013

Updates: 7/18/2013: Note change in units change for Hexoaminidase A, previously reported as U/L. Also reference ranges have been amended for age groups.
1/27/2016: CPT update