
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: SST (Gold, marble or red) 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

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](#) December 2017

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
 12/12/2017: Collection container update.