**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 |

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<th>Percent A (%)</th>
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<td>≤15 years:</td>
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<td>≥16 years:</td>
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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](https://www.mayoclinic.org) 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.