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

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| 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 nm | ol/min/mL | | |
| | ≥16 years: | 10.4 – 23.8 nmol/min/mL | | | |
| | Percent A (%) | | | | |
| | ≤15 years: 20 ≥16 years: 56 | | 20 – 90% | | |
| | | | 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. | | | | |
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| 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. | | | | |