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**Reporting Title:** Hexosaminidase A and Total, S**Performing Location:** Rochester**Ordering Guidance:**

Testing for Tay-Sachs Disease and Sandhoff Disease

The following tests are available for diagnostic and carrier testing for Tay-Sachs and Sandhoff diseases.

NAGR / Hexosaminidase A and Total, Leukocytes/Molecular Reflex, Whole Blood:

- This is the recommended test for carrier testing for Tay-Sachs disease and Sandhoff disease.
- Testing begins with hexosaminidase A and total enzyme analysis. If the results are consistent with an affected or carrier for Tay-Sachs disease or Sandhoff disease, next generation sequencing to detect single nucleotide and copy number variants for *HEXA* or *HEXB*, respectively, will automatically be performed on the original specimen.
- This test is appropriate for males and pregnant or nonpregnant females.

NAGW / Hexosaminidase A and Total Hexosaminidase, Leukocytes:

- This test can be used for diagnosis and carrier testing for Tay-Sachs disease or Sandhoff disease.
- Results for hexosaminidase A and total enzyme analysis are reported with recommendations for additional testing when appropriate. All follow-up testing must be ordered separately on new specimens.
- This test is appropriate for males and pregnant or nonpregnant females.

NAGS / Hexosaminidase A and Total Hexosaminidase, Serum:

- This test can be used for diagnosis and carrier testing for Tay-Sachs disease or Sandhoff disease.
- Results for hexosaminidase A and total enzyme analysis are reported with recommendations for additional testing when appropriate.
- If results indicate normal, indeterminate, or carrier status and the suspicion of Tay-Sachs disease remains high, MUGS / Hexosaminidase A, Serum for Tay-Sachs disease (B1 variant) can typically be added and performed on the same specimen.
- With the exception of MUGS, all follow-up testing must be ordered separately on new specimens.
- This test is **not appropriate for pregnant females** or women receiving hormonal contraception. This test is appropriate for males and nonpregnant females.
- This test is particularly useful when it is difficult to obtain enough blood to perform leukocyte testing (NAGR or NAGW), as may be the case with infants.

MUGS / Hexosaminidase A, Serum:

- This is the recommended test for diagnosis and carrier testing for the B1 variant of Tay-Sachs disease. This test will not detect Sandhoff disease.
- This test should not be ordered as a first-line test.** Rather, this test should be ordered when the NAGR, NAGW, NAGS indicate normal, indeterminate, or carrier results and the suspicion of Tay-Sachs disease remains high. In most cases, this test can be performed on the original specimen collected for NAGS.

**Specimen Requirements:****Supplies:** Sarstedt Aliquot Tube, 5 mL (T914)**Collection Container/Tube:****Preferred:** Serum gel**Acceptable:** Red top

**Submission Container/Tube:** Plastic vial  
**Specimen Volume:** 1 mL  
**Collection Instructions:** Centrifuge and aliquot serum into plastic vial.

Forms:

1. **New York Clients-Informed consent is required.** Document on the request form or electronic order that a copy is on file. The following documents are available:  
-[Informed Consent for Genetic Testing](#) (T576)  
-[Informed Consent for Genetic Testing-Spanish](#) (T826)
2. [Biochemical Genetics Patient Information](#) (T602)
3. If not ordering electronically, complete, print, and send a [Biochemical Genetics Test Request](#) (T798) with the specimen.

| Specimen Type | Temperature        | Time    | Special Container |
|---------------|--------------------|---------|-------------------|
| Serum         | Refrigerated       | 7 days  |                   |
|               | Frozen (preferred) | 30 days |                   |

Result Codes:

| Result ID | Reporting Name              | Type         | Unit        | LOINC®  |
|-----------|-----------------------------|--------------|-------------|---------|
| 27612     | Hexosaminidase Total, S     | Numeric      | nmol/min/mL | 1956-2  |
| 27613     | Hexosaminidase Percent A, S | Numeric      | %           | 12914-8 |
| 27216     | Interpretation (NAGS)       | Alphanumeric |             | 59462-2 |
| 27218     | Reviewed By                 | Alphanumeric |             | 18771-6 |

LOINC® and CPT codes are provided by the performing laboratory.

Supplemental Report:

No

CPT Code Information:

83080 x 2

Reference Values:

Hexosaminidase total  
< or =15 years: > or =20 nmol/min/mL  
> or =16 years: 10.4-23.8 nmol/min/mL

Hexosaminidase percent A  
< or =15 years: 20-90%  
> or =16 years: 56-80%