

Test Definition: MUGS

Hexosaminidase A, Serum

Reporting Title: Hexosaminidase A (MUGS), 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, or 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.

Necessary Information:

Healthcare professional name and phone number are required.

Specimen Requirements:

Patient Preparation: Patient should be fasting for 4 hours.

Supplies: Sarstedt Aliquot Tube, 5 mL (T914)



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Hexosaminidase A, Serum

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. <u>Biochemical Genetics Patient Information</u> (T602)
- 3. If not ordering electronically, complete, print, and send a <u>Biochemical Genetics Test Request</u> (T798) with the specimen.

Specimen Type	Temperature	Time	Special Container	
Serum	Refrigerated	5 days		
	Frozen (preferred)	365 days		

Result Codes:

Result ID	Reporting Name	Туре	Unit	LOINC®
80350	Hexosaminidase A (MUGS), S	Numeric	U/L	2643-5

 $\ensuremath{\mathsf{LOINC}}\xspace^{\ensuremath{\mathsf{B}}}$ and CPT codes are provided by the performing laboratory.

Supplemental Report:

No

CPT Code Information:

83080

Reference Values:

1.23-2.59 U/L (Normal)

1.16-1.22 U/L (Indeterminate)

0.58-1.15 U/L (Carrier)