MAYO CLINIC LABORATORIES

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:

Physician's name and phone number are required.

Specimen Requirements:

Patient Preparation: Patient should be fasting for 4 hours.
Collection Container/Tube:

MAYO CLINIC LABORATORIES

Test Definition: MUGS

Hexosaminidase A, Serum

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	Frozen (preferred)	365 days	
	Refrigerated	5 days	

Result Codes:

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

LOINC® 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)