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):
-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:

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.

Specimen Minimum Volume:
0.5 mL

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)	30 days	
	Refrigerated	7 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%