



Reporting Title: Hexosaminidase A and Tot, WBC/Mole

Performing Location: Rochester

Ordering Guidance:

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.

Shipping Instructions:

For optimal isolation of leukocytes, it is recommended the specimen arrive refrigerated within 6 days of collection to be stabilized. Collect specimen Monday through Thursday only and not the day before a holiday. Specimen should be collected and packaged as close to shipping time as possible.

Specimen Requirements:

Container/Tube:

Preferred: Yellow top (ACD solution B)

Acceptable: Yellow top (ACD solution A)

Specimen Volume: 6 mL

Collection Instructions: Send whole blood specimen in original tube. Do not aliquot.

Specimen Minimum Volume:

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
Whole Blood ACD	Refrigerated (preferred)	6 days	YELLOW TOP/ACD
	Ambient	6 days	YELLOW TOP/ACD

Ask at Order Entry (AOE) Questions:

Test ID	Question ID	Description	Type	Reportable
CGPH	MG135	Gene List ID	Plain Text	No

Result Codes:

Result ID	Reporting Name	Type	Unit	LOINC®
8775	Hexosaminidase Total, WBC Also used by tests: NAGW	Alphanumeric	nmol/min/mg	24075-4
2294	Hexosaminidase Percent A, WBC Also used by tests: NAGW	Alphanumeric	%	23825-3



Result ID	Reporting Name	Type	Unit	LOINC®
2284	Interpretation (NAGW) Also used by tests: NAGW	Alphanumeric		59462-2
35029	Reviewed By Also used by tests: NAGW	Alphanumeric		18771-6

LOINC and CPT codes are provided by the performing laboratory.

Supplemental Report:

No

CPT Code Information:

83080 x2

81406 (if appropriate)

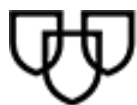
81479 (if appropriate)

Reflex Tests:

Test ID	Reporting Name	CPT Units	CPT Code	Always Performed	Orderable Separately
CGPH	Custom Gene Panel, Hereditary			No	Yes
G145	Hereditary Custom Gene Panel Tier 1			No	No (Bill Only)
G146	Hereditary Custom Gene Panel Tier 2			No	No (Bill Only)
G147	Hereditary Custom Gene Panel Tier 3			No	No (Bill Only)
G148	Hereditary Custom Gene Panel Tier 4			No	No (Bill Only)
G149	Hereditary Custom Gene Panel Tier 5			No	No (Bill Only)
G150	Hereditary Custom Gene Panel Tier 6			No	No (Bill Only)
HEXBZ	HEXB Gene, Full Gene Analysis			No	Yes
CULFB	Fibroblast Culture for Genetic Test			No	Yes

**Result Codes for Reflex Tests:**

Test ID	Result ID	Reporting Name	Type	Unit	LOINC®
CGPH	MG135	Gene List ID	Alphanumeric		48018-6
CGPH	610422	Test Description	Alphanumeric		62364-5
CGPH	606046	Specimen	Alphanumeric		31208-2
CGPH	606047	Source	Alphanumeric		31208-2
CGPH	606040	Result Summary	Alphanumeric		50397-9
CGPH	606041	Result	Alphanumeric		82939-0
CGPH	606042	Interpretation	Alphanumeric		69047-9
CGPH	620157	Additional Results	Alphanumeric		82939-0
CGPH	610423	Resources	Alphanumeric		99622-3
CGPH	606043	Additional Information	Alphanumeric		48767-8
CGPH	606044	Method	Alphanumeric		85069-3
CGPH	610424	Genes Analyzed	Alphanumeric		48018-6
CGPH	606045	Disclaimer	Alphanumeric		62364-5
CGPH	606048	Released By	Alphanumeric		18771-6
HEXBZ	608716	Test Description	Alphanumeric		62364-5
HEXBZ	608717	Specimen	Alphanumeric		31208-2
HEXBZ	608718	Source	Alphanumeric		31208-2
HEXBZ	608719	Result Summary	Alphanumeric		50397-9
HEXBZ	608720	Result	Alphanumeric		82939-0
HEXBZ	608721	Interpretation	Alphanumeric		69047-9
HEXBZ	608722	Resources	Alphanumeric		99622-3
HEXBZ	608723	Additional Information	Alphanumeric		48767-8
HEXBZ	608724	Method	Alphanumeric		85069-3
HEXBZ	608725	Genes Analyzed	Alphanumeric		48018-6
HEXBZ	608726	Disclaimer	Alphanumeric		62364-5
HEXBZ	608727	Released By	Alphanumeric		18771-6
CULFB	52327	Result Summary	Alphanumeric		50397-9
CULFB	52329	Interpretation	Alphanumeric		69965-2
CULFB	52328	Result	Alphanumeric		82939-0



Test ID	Result ID	Reporting Name	Type	Unit	LOINC®
CULFB	CG770	Reason for Referral	Alphanumeric		42349-1
CULFB	CG899	Specimen	Alphanumeric		31208-2
CULFB	52331	Source	Alphanumeric		31208-2
CULFB	52332	Method	Alphanumeric		85069-3
CULFB	54625	Additional Information	Alphanumeric		48767-8
CULFB	52333	Released By	Alphanumeric		18771-6

Reference Values:

HEXOSAMINIDASE TOTAL

< or =15 years: > or =20 nmol/min/mg
> or =16 years: 16.4-36.2 nmol/min/mg

HEXOSAMINIDASE PERCENT A

< or =15 years: 20-80% of total
> or =16 years: 63-75% of total