Test Definition: NAGR

Hexosaminidase A and Tot, WBC/Mole

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 specimen in original tube. **Do not aliquot**.

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 an Inborn Errors of Metabolism 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

Result Codes:

Result ID	Reporting Name	Type	Unit	LOINC
8775	Hexosaminidase Total, WBC	Alphanumeric	nmol/min/mg	24075-4
2294	Hexosaminidase Percent A, WBC	Alphanumeric	%	23825-3
2284	Interpretation (NAGW)	Alphanumeric		59462-2
35029	Reviewed By	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	Available Separately
CGPH	Custom Gene Panel, Hereditary	1	CPT c	No	Yes
HEXBZ	HEXB Gene, Full Gene Analysis	1	81479	No	Yes

Result Codes for Reflex Tests:

Test ID	Result ID	Reporting Name	Type	Unit	LOINC

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
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	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

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