MAYO CLINIC LABORATORIES

Test Definition: LGB3S

Globotriaosylsphingosine, Serum

Overview

Useful For
Diagnosis and monitoring of Fabry disease

Genetics Test Information
This test is used to diagnose and monitor patients with Fabry disease.

Testing Algorithm
The following algorithms are available:
-Fabry Disease: Newborn Screen-Positive Follow-up
-Fabry Disease Diagnostic Testing Algorithm

Special Instructions
- Fabry Disease Diagnostic Testing Algorithm
- Fabry Disease: Newborn Screen-Positive Follow-up
- Biochemical Genetics Patient Information

Method Name
Liquid Chromatography Tandem Mass Spectrometry (LC-MS/MS)

NY State Available
Yes

Specimen

Specimen Type
Serum

Ordering Guidance
This test **should not be used** to determine carrier status. Order GLA / Fabry Disease, *GLA* Gene Sequencing with Deletion/Duplication, Varies for carrier testing.

Necessary Information
1. Patient's age is required.
2. Reason for testing is required.

Specimen Required
Supplies: Sarstedt Aliquot Tube, 5 mL (T914)
Collection Container/Tube:
Preferred: Serum gel

Acceptable: Red top
Submission Container/Tube: Plastic vial
Specimen Volume: 1 mL serum
Collection Instructions: Centrifuge and aliquot serum into a plastic vial.

Forms

1. Biochemical Genetics Patient Information (T602)
2. If not ordering electronically, complete, print, and send a Biochemical Genetics Test Request (T798) with the specimen.

Specimen Minimum Volume

Serum: 0.5 mL

Reject Due To

Gross hemolysis	OK
Gross lipemia	Reject
Gross icterus	OK

Specimen Stability Information

Specimen Type	Temperature	Time	Special Container
Serum	Frozen (preferred)	90 days	
	Refrigerated	48 hours	

Clinical & Interpretive

Clinical Information

Fabry disease is an X-linked lysosomal storage disorder caused by a deficiency of the enzyme alpha-galactosidase A (alpha-Gal A). Reduced enzyme activity results in accumulation of glycosphingolipids in the lysosomes throughout the body, in particular, the kidney, heart, and brain. Severity and onset of symptoms are dependent on the residual enzyme activity. Symptoms may include acroparesthesias (pain crises), multiple angiokeratomas, reduced or absent sweating, corneal opacity, renal insufficiency leading to end-stage kidney disease, and cardiac and cerebrovascular disease. There are renal and cardiac variant forms of Fabry disease that may be underdiagnosed. Female patients with Fabry disease can have clinical presentations ranging from asymptomatic to severely affected, and they may have alpha-Gal A activity in the normal range.

Unless irreversible damage has already occurred, treatment with enzyme replacement therapy leads to significant clinical improvement in affected individuals. For this reason, early diagnosis and treatment are desirable. In a few US states, early detection of Fabry disease through newborn screening has been implemented.

Absent or reduced alpha-Gal A in blood spots, leukocytes (AGAW / Alpha-Galactosidase, Leukocytes), or serum (AGAS / Alpha-Galactosidase, Serum) can indicate a diagnosis of classic or variant Fabry disease. Molecular sequence analysis of

the *GLA* gene (GLA / Fabry Disease, *GLA* Gene Sequencing with Deletion/Duplication, Varies) allows for detection of the disease-causing variant. Molecular genetic testing is the recommended diagnostic test for female patients as alpha-galactosidase activity may be in the normal range even in those who are symptomatic.

The glycosphingolipid, globotriaosylsphingosine (LGb3), may be elevated in symptomatic patients and supports a diagnosis of Fabry disease. It may also be helpful as a tool for monitoring disease progression as well as determining treatment response in known patients. In addition, measurement of LGb3, may provide additional diagnostic information in the evaluation of uncertain cases, such as in asymptomatic heterozygous female patients, individuals with novel *GLA* variants of unclear clinical significance, as well as asymptomatic patients identified by family screening.

Reference Values

< or =1.0 ng/mL

Interpretation

Elevation of globotriaosylsphingosine is diagnostic for Fabry disease.

Cautions

Carrier detection using globotriaosylsphingosine (LGb3) is unreliable.

Some patients with Fabry disease may, and all individuals with pseudodeficiency of alpha-galactosidase enzyme, have normal concentrations of LGb3.

Clinical Reference

1. Aerts JM, Groener JE, Kuiper S, et al. Elevated globotriaosylsphingosine is a hallmark of Fabry disease. Proc Natl Acad Sci USA. 2008;105(8)2812-2817
2. Mehta A, Hughes DA. Fabry disease. In: Adam MP, Feldman J, Mirzaa GM, et al, eds. GeneReviews [Internet].University of Washington, Seattle; 2002. Updated April 11, 2024. Accessed October 13, 2025. Available at www.ncbi.nlm.nih.gov/books/NBK1292/
3. Laney DA, Bennett RL, Clarke V, et al. Fabry disease practice guidelines: recommendations of the National Society of Genetic Counselors. J Genet Couns. 2013;22(5):555-564
4. Laney DA, Peck DS, Atherton AM, et al. Fabry disease in infancy and early childhood: a systematic literature review. Genet Med. 2015;17(5):323-330
5. Weidemann F, Beer M, Kralewski M, Siwy J, Kampmann C. Early detection of organ involvement in Fabry disease by biomarker assessment in conjunction with LGE cardiac MRI: results from the SOPHIA study. Mol Genet Metab. 2019;126(2):169-182

Performance

Method Description

Internal standard is added to the serum. Globotriaosylsphingosine (LGb3) is extracted from the serum prior to injection onto a liquid chromatography tandem mass spectrometry (LC-MS/MS) system. Following chromatographic isolation, the concentration is measured by MS/MS analysis in the selected reaction monitoring positive mode. The ratio of extracted peak area to internal standard is utilized to calculate the concentration of LGb3 in the sample.(Unpublished Mayo method)

Test Definition: LGB3S

Globotriaosylsphingosine, Serum

PDF Report
No

Day(s) Performed
Wednesday

Report Available
3 to 16 days

Specimen Retention Time
1 month

Performing Laboratory Location
Mayo Clinic Laboratories - Rochester Main Campus

Fees & Codes

Fees
- Authorized users can sign in to Test Prices for detailed fee information.
- Clients without access to Test Prices can contact Customer Service 24 hours a day, seven days a week.
- Prospective clients should contact their account representative. For assistance, contact Customer Service.

Test Classification
This test was developed and its performance characteristics determined by Mayo Clinic in a manner consistent with CLIA requirements. It has not been cleared or approved by the US Food and Drug Administration.

CPT Code Information
82542

LOINC® Information

Test ID	Test Order Name	Order LOINC® Value
LGB3S	Lyso-GB3, S	90234-6

Result ID	Test Result Name	Result LOINC® Value
BG708	Reason for Referral	42349-1
65532	Lyso-GB3, S	90234-6
113176	Interpretation (LGB3S)	59462-2
113177	Reviewed By	18771-6