Test Definition: LGBBS

Globotriaosylsphingosine, Blood Spot

Overview

Useful For

Screening of patients with Fabry disease using dried blood spots when a serum specimen is not available

This test **should not be used for** newborn screening followup.

Special Instructions

- Biochemical Genetics Patient Information
- Blood Spot Collection Card-Spanish Instructions
- Blood Spot Collection Card-Chinese Instructions
- Blood Spot Collection Instructions

Method Name

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

NY State Available

Yes

Specimen

Specimen Type

Whole blood

Ordering Guidance

Serum is the recommended specimen type for diagnosing and monitoring patients with Fabry Disease. For more information see LGB3S / Globotriaosylsphingosine, Serum.

Specimen Required

Supplies:

-Card-Blood Spot Collection (Filter Paper) (T493)

-Card Postmortem Screening (Filter Paper) (T525)

Container/Tube:

Preferred: Blood Spot Collection Card (Filter Paper)

Acceptable: Whatman Protein Saver 903 filter paper, PerkinElmer 226 filter paper, Munktell filter paper, Postmortem Screening Card or collected with EDTA, sodium heparin, lithium heparin, or ACD B-containing devices

Specimen Volume: 2 Blood spots

Collection Instructions:

1. An alternative blood collection option for a patient older than 1 year is a fingerstick. For detailed instructions, see How to Collect a Dried Blood Spot Sample.

2. Let blood dry completely on filter paper at ambient temperature in a horizontal position for a minimum of 3 hours.

3. At least 1 spot should be complete,(ie, unpunched).
4. Do not expose specimen to heat or direct sunlight.
5. Do not stack wet specimens.
6. Keep specimen dry.

Additional Information:

1. For collection instructions, see Blood Spot Collection Instructions.
2. For collection instructions in Spanish, see Blood Spot Collection Card-Spanish Instructions (T777).
3. For collection instructions in Chinese, see Blood Spot Collection Card-Chinese Instructions (T800).

Forms

Biochemical Genetics Patient Information (T602)

Specimen Minimum Volume

1 Blood spot

Reject Due To

Blood spot specimen that shows serum rings or has multiple layers	Reject
Insufficient specimen	Reject

Specimen Stability Information

Specimen Type	Temperature	Time	Special Container
Whole blood	Refrigerated (preferred)	10 days	FILTER PAPER
	Ambient	10 days	FILTER PAPER
	Frozen	59 days	FILTER PAPER

Clinical & Interpretive

Clinical Information

Fabry disease is an X-linked recessive 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, particularly in 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, kidney insufficiency leading to kidney failure, and cardiac and cerebrovascular disease. There are renal and cardiac variant forms of Fabry disease that may be underdiagnosed. Female patients who are heterozygous for Fabry disease can have clinical presentations ranging from asymptomatic to severely affected, and they may have alpha-GAL A activity in the normal range. The estimated incidence varies from 1 in 3000 infants detected via newborn screening to 1 in 10,000 male patients diagnosed after onset of symptoms.

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

Measurement of alpha-GAL A in leukocytes (AGAW / Alpha-Galactosidase, Leukocytes), serum (AGAS / Alpha-Galactosidase, Serum), or blood spots (PLSD / Lysosomal and Peroxisomal Disorders Screen, Blood Spot) can reliably diagnose classic or variant Fabry disease in male patients. Molecular genetic testing is the recommended diagnostic test for female patients as alpha-GAL A activity may be in the normal range in an affected female patient. Molecular sequence analysis of the *GLA* gene allows for detection of the disease-causing variant in both male and female patients, order GLA / Fabry Disease, *GLA* Gene Sequencing with Deletion/Duplication, Varies.

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

Cutoff: < or =0.034 nmol/mL

Interpretation

An elevation of globotriaosylsphingosine is indicative of Fabry disease, however a normal result does not rule out Fabry disease.

Cautions

Some patients with late-onset Fabry disease may have normal concentrations of globotriaosylsphingosine.

Clinical Reference

1. Vardarli I, Rischpler C, Herrmann K, Weidemann F. Diagnosis and screening of patients with Fabry disease. Ther Clin Risk Manag. 2020;16:551-558. doi:10.2147/TCRM.S247814
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 3, 2025. Available at www.ncbi.nlm.nih.gov/books/NBK1292/
3. Nowak A, Mechtler T, Kasper DC, Desnick RJ. Correlation of Lyso-Gb3 levels in dried blood spots and sera from patients with classic and later-onset Fabry disease. Mol Genet Metab. 2017;121(4):320-324. doi:10.1016/j.ymgme.2017.06.006
4. Johnson B, Mascher H, Mascher D, et al. Analysis of lyso-globotriaosylsphingosine in dried blood spots. Ann Lab Med. 2013;33(4):274-278. doi:10.3343/alm.2013.33.4.274

Performance

Method Description

A 3-mm dried blood spot is extracted with internal standard. The extract is subjected to liquid chromatography tandem

mass spectrometry (LC-MS/MS) analysis. The MS/MS is operated in the multiple reaction monitoring positive mode to follow the precursor to product species transitions for each analyte and internal standard. The ratio of the extracted peak areas to internal standard is determined by LC-MS/MS is used to calculate the concentration of in the sample.(Unpublished Mayo method)

PDF Report
No

Day(s) Performed
Tuesday

Report Available
3 to 9 days

Specimen Retention Time
Normal result: 2 months; Abnormal result: Indefinitely

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
LGBBS	Globotriaosylsphingosine, BS	92754-1

Result ID	Test Result Name	Result LOINC® Value
BA4368	Interpretation (LGBBS)	59462-2
BA4367	Globotriaosylsphingosine	92754-1
BA4369	Reviewed By	18771-6