Overview

Useful For
This test is **not intended for** newborn screening followup.

Diagnosing and monitoring of patients with Fabry disease when a serum specimen is not available

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

NY State Available
Yes

Specimen

Specimen Type
Whole blood

Advisory Information
Serum is the recommended specimen type for monitoring patients with Fabry disease. For more information see LGB3S / Globotriaosylsphingosine, Serum.

Specimen Required

Container/Tube:
- **Preferred:** Lavender top (EDTA)
- **Acceptable:** Sodium heparin, lithium heparin, ACD B

Specimen Volume: 1 mL

Forms
If not ordering electronically, complete, print, and send an Inborn Errors of Metabolism Test Request (T798) with the specimen.

Specimen Minimum Volume
0.25 mL

Reject Due To

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<tbody>
<tr>
<td>Hemolysis</td>
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<td>Lipemia</td>
<td>Mild OK; Gross OK</td>
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<tr>
<td>Icterus</td>
<td>Mild OK; Gross OK</td>
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Specimen Stability Information
**Clinical and 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, 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 renal disease, and cardiac and cerebrovascular disease. There are renal and cardiac variant forms of Fabry disease that may be underdiagnosed. Heterozygous females of 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 males diagnosed after onset of symptoms.

Unless irreversible damage has already occurred, treatment with enzyme replacement therapy (ERT) 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 (AGA / Alpha-Galactosidase, Leukocytes), serum (AGAS / Alpha-Galactosidase, Serum), or blood spots (AGABS / Alpha-Galactosidase, Blood Spot) can reliably diagnose classic or variant Fabry disease in males. Molecular genetic testing is the recommended diagnostic test for females as alpha-Gal A may be in the normal range in an affected female patient. Molecular analysis of the GLA gene (FABRZ / Fabry Disease, Full Gene Analysis) allows for detection of the disease-causing mutation in males and females.

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 or globotriaosylsphingosine (LGb3), may provide additional diagnostic information in the evaluation of uncertain cases, such as in asymptomatic heterozygous females, individuals with novel GLA variants of unclear clinical significance, as well as asymptomatic patients identified by family screening.

**Reference Values**

Cutoff: \(<\) or \(\leq\) 0.034 nmol/mL

**Interpretation**

An elevation of globotriaosylsphingosine (LGb3) is suggestive of Fabry disease.

**Cautions**

Some patients with Fabry disease may have normal concentrations of globotriaosylsphingosine (LGb3).

**Clinical Reference**


Test Definition: LGBWB
Globotriaosylsphingosine, B


Performance

Method Description
A 3-mm dried blood spot (DBS) 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 (MRM) 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) and Time(s) Test Performed
Samples received Monday through Saturday; 4 p.m.
Sunday 1 p.m. will be spotted same day.
Testing performed Tuesday and Thursday 8 a.m.

Analytic Time
2 days (not reported on Saturday or Sunday)

Maximum Laboratory Time
8 days

Specimen Retention Time
Whole blood: 7 days Dried Blood Spot: Normals: 1 year; Abnormal: Indefinitely

Performing Laboratory Location
Rochester

Fees and 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 Regional Manager. 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. This test has not been cleared or approved by the U.S. Food and Drug Administration.
Test Definition: LGBWB
Globotriaosylsphingosine, B

CPT Code Information
82542

LOINC® Information

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