Overview

Useful For
Second-tier test when newborn screening results with reduced beta-glucosidase (GBA) activity are identified
Diagnosis and monitoring of patients with Gaucher disease
Documentation of an elevated glucopsychosine (glucosylsphingosine: lyso-GL1) level supports the biochemical diagnosis of Gaucher disease
Monitoring a patient's response to treatment

This test is not useful for identifying carriers of GBA mutations.

Highlights
Gaucher disease is an autosomal recessive lysosomal storage disorder caused by deficient beta-glucosidase activity.
There are 3 described types of Gaucher disease with varying clinical presentations and age of onset from a perinatal lethal disorder to an asymptomatic type.
Glucopsychosine (glucosylsphingosine: lyso-GL1) is elevated in symptomatic patients and supports a diagnosis of Gaucher disease.

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

NY State Available
Yes

Specimen

Specimen Type
Plasma

Specimen Required

Collection Container/Tube:

Preferred: Lavender top (EDTA)
Acceptable: Green top (sodium heparin, lithium heparin) or yellow top (ACD B)

Submission Container/Tube: Plastic vial

Specimen Volume: 0.3 mL

Collection Instructions:

1. Centrifuge at 4 degrees C, if possible
2. Aliquot plasma being careful to ensure no buffy coat is transferred with the plasma then freeze.

**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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<td>Gross lipemia</td>
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<td>Gross icterus</td>
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**Specimen Stability Information**

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<tr>
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**Clinical and Interpretive**

**Clinical Information**

Gaucher disease is an autosomal recessive lysosomal storage disorder caused by a deficiency of the enzyme, beta-glucosidase. Beta-glucosidase facilitates the lysosomal degradation of glucosylceramide (glucocerebroside) and glucopsychosine (glucosylsphingosine: lyso-GL1). Gaucher disease is caused by mutations in the GBA gene. There are 3 described types of Gaucher disease with varying clinical presentations and age of onset from a perinatal lethal disorder to an asymptomatic type. Features of all types of Gaucher disease include hepatosplenomegaly and hematological abnormalities.

Gaucher disease type I is the most common, representing more than 90% of cases. It is generally characterized by bone disease, hepatosplenomegaly, anemia and thrombocytopenia, coagulation abnormalities, lung disease, but no central nervous system (CNS) involvement. Gaucher disease types II and III are characterized by the presence of primary neurologic disease. In addition, Type II typically presents with limited psychomotor development, hepatosplenomegaly, and lung disease, resulting in death usually between 2 and 4 years of age. Individuals with Gaucher disease type III may present prior to 2 years of age, but the progression is not as rapid and patients may survive into the third and fourth decade. Additional subtypes of Gaucher disease include a perinatal lethal form associated with skin abnormalities and nonimmune hydrops fetalis, and a cardiovascular form presenting with calcification of the aortic and mitral valves, mild splenomegaly, corneal opacities, and gaze impairment.

Treatment is available in the form of enzyme replacement therapy and substrate reduction therapy for types I and III. These treatment options have generally made bone marrow transplantation obsolete. Currently, only supportive therapy is available for type II because of the inability of enzyme provided by replacement therapy to cross the blood-brain barrier.

The incidence of Gaucher disease type I ranges from 1 in 30,000 to 1 in 100,000 in the general population but is much more frequent among Ashkenazi Jews with an incidence of approximately 1 in 900. Types II and III both have an incidence of approximately 1 in 100,000 in the general population.
A diagnostic workup for Gaucher disease may demonstrate the characteristic finding of Gaucher cells on bone marrow examination, other hematologic abnormalities, and hepatosplenomegaly. The diagnosis can be confirmed by the demonstration of reduced or absent acid beta-glucosidase activity in leukocytes (BGL / Beta-Glucosidase, Leukocytes), or dried blood spots (PLSD / Lysosomal and Peroxisomal Storage Disorders Screen, Blood Spot) and molecular genetic analysis of the GBA gene (GAUP / Gaucher Disease, Mutation Analysis, GBA; or GBAZ / Gaucher Disease, Full Gene Analysis). Lyso-GL1 is elevated in symptomatic patients and supports a diagnosis of Gaucher disease. It may also be helpful in determining treatment response.

**Reference Values**

**GLUCOPSYCHOSINE**

Cutoff: \(< or =\) 0.003 nmol/mL

**Interpretation**

An elevation of glucopsychosine (glucosylsphingosine: lyso-GL1) is indicative of Gaucher disease.

**Cautions**

No significant cautionary statements.

**Clinical Reference**


**Performance**

**Method Description**

An internal standard is added to an aliquot of plasma, which is then subjected to protein precipitation. Following centrifugation, the supernatant 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)
Test Definition: GPSYP
Glucopsychosine, P

PDF Report
No

Day(s) and Time(s) Test Performed
Tuesday; 8 a.m.

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

Maximum Laboratory Time
8 days

Specimen Retention Time
2 months

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.

CPT Code Information
82542

LOINC® Information

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