Test Definition: GPSYW

Glucopsychosine, B

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* variants.

Genetics Test Information

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.

Testing Algorithm

The following are available in Special Instructions:

-Newborn Screen Follow-up for Gaucher Disease

-Newborn Screening Act Sheet Gaucher Disease: Decreased Acid Beta-Glucosidase

Special Instructions

- Biochemical Genetics Patient Information
- Newborn Screening Act Sheet Gaucher Disease: Decreased Acid Beta-Glucosidase
- Newborn Screen Follow-up for Gaucher Disease

Method Name

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

NY State Available

Yes

Specimen

Specimen Type

Whole blood

Specimen Required

Container/Tube:

Preferred: Lavender top (EDTA)

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

Specimen Volume: 1 mL

Forms

1. Biochemical Genetics Patient Information (T602) in Special Instructions.

2. 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

Gross hemolysis	OK
Gross lipemia	OK
Gross icterus	OK

Specimen Stability Information

Specimen Type	Temperature	Time	Special Container
Whole blood	Refrigerated (preferred)	72 hours	
	Ambient	48 hours	

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 variants 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 form, 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,* Varies; or GBAZ / Gaucher Disease, Full Gene Analysis, Varies). 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

Cutoff: < or =0.040 nmol/mL

Interpretation

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

Cautions

No significant cautionary statements

Clinical Reference

1. Orvisky E, Park J, LaMarca M, et al: Glucosylsphingosine accumulation in tissues from patients with Gaucher disease: correlation with phenotype and genotype. Mol Genet Metab 2002;76:262-270

2. Pastores GM, Hughes DA: Gaucher Disease. In GeneReviews Edited by RA Pagon, MP Adam, HH Ardinger, et al: University of Washington, Seattle, 1993-2019. 2000 Jul 27 (Updated 2018 Jun 21). Accessed May 2019. Available at www.ncbi.nlm.nih.gov/books/NBK1269/

3. Dekker N, Dussen L, Hollak C, et al: Elevated plasma glucosylsphingosine in Gaucher disease: relation to phenotype, storage cell markers, and therapeutic response. Blood 2011;118(16):118-127

4. Kaplan P, Baris H, De Meirleir L, et al: Revised recommendations for the management of Gaucher disease in children. Eur J Pediatr 2013;172:447-458

5. Grabowski GA, Petsko GA, Kolodny EH, et al: Gaucher disease. In The Online Matabolic and Molecular Bases of Inherited Disease. Edited by D Valle, AL Beaudet, B Vogelstein, et al. New York, McGraw-Hill. Accessed August 11, 2017. Available at http://ommbid.mhmedical.com/content.aspx?bookid=971&sectionid=62643884

6. Murugeasan V, Chuan WL, Liu J, et al: Glycosylsphingosine is a key biomarker of Gaucher disease. Am J Hematol 2016;91(11)1082-1089

Performance

Method Description

Whole blood is spotted on filter paper and allowed to dry. 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 prepared same day.

Testing performed Tuesday; 8 a.m

Analytic Time

2 days

Maximum Laboratory Time

8 days

Specimen Retention Time

Whole blood: 7 days; Dried Blood Spot: Normal results: 1 year; Abnormal results: 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.

CPT Code Information

82542

LOINC® Information

Test ID	Test Order Name	Order LOINC Value
GPSYW	Glucopsychosine, B	92751-7

Result ID	Test Result Name	Result LOINC Value
BA4357	Interpretation (GPSYW)	59462-2
BA4356	Glucopsychosine	92751-7
BA4358	Reviewed By	18771-6