Overview

Useful For
Diagnosis of Gaucher disease

This test is not intended for carrier detection.

Genetics Test Information
This test provides diagnostic testing for patients with clinical signs and symptoms suspicious for Gaucher disease.

Enzyme testing is included in the diagnostic workup for infants following a positive newborn screen result for 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

- Informed Consent for Genetic Testing
- Biochemical Genetics Patient Information
- Newborn Screening Act Sheet Gaucher Disease: Decreased Acid Beta-Glucosidase
- Newborn Screen Follow-up for Gaucher Disease
- Informed Consent for Genetic Testing (Spanish)

Method Name
Flow Injection Analysis-Tandem Mass Spectrometry (FIA-MS/MS)

NY State Available
Yes

Specimen

Specimen Type
Whole Blood ACD

Advisory Information
This test is preferred for diagnostic testing; however results are not reflective of carrier status. For carrier detection; order GAUP / Gaucher Disease, Mutation Analysis, GBA, Varies or GBAZ / Gaucher Disease, Full Gene Analysis, Varies. Call 800-533-1710 to discuss testing options.

Shipping Instructions
For optimal isolation of leukocytes, it is recommended the specimen arrive refrigerated within 96 hours of collection to be stabilized. Collect specimen Monday through Thursday only and not the day before a holiday. Specimen should be collected and packaged as close to shipping time as possible.

Specimen Required
Container/Tube:

Preferred: Yellow top (ACD solution B)

Acceptable: Yellow top (ACD solution A) or lavender top (EDTA)

Specimen Volume: 6 mL

Collection Instructions: Send specimen in original tube. Do not transfer blood to other containers.

Forms

1. **New York Clients-Informed consent is required.** Document on the request form or electronic order that a copy is on file. The following documents are available in Special Instructions:
   - *Informed Consent for Genetic Testing* (T576)
   - *Informed Consent for Genetic Testing-Spanish* (T826)

2. *Biochemical Genetics Patient Information* (T602) in Special Instructions

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

Specimen Minimum Volume

2 mL

Reject Due To

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Specimen Stability Information

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<th>Temperature</th>
<th>Time</th>
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<tr>
<td>Whole Blood ACD</td>
<td>Refrigerated (preferred)</td>
<td>4 days</td>
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Clinical and Interpretive

Clinical Information

Gaucher disease is an autosomal recessive lysosomal storage disorder caused by reduced or absent acid beta-glucosidase (glucocerebrosidase) enzyme activity. Absent or reduced activity of this enzyme results in accumulation of glucosylceramide (glucocerebroside) and glucosylsphingosine (glucosylsphingosine) in the lysosomes and interferes with the normal functioning of cells.

Clinical features and severity of symptoms are widely variable within Gaucher disease, but in general, the disorder is characterized by abnormal blood parameters such as decreased red blood cells (anemia) and/or platelets (thrombocytopenia), bone disease, and hepatosplenomegaly. Individuals with more severe types of Gaucher disease may have central nervous system (CNS) involvement. There are 3 clinical subtypes of the disorder that vary with
respect to age of onset and clinical presentation. Type 1 is the most common type, representing 95% of all cases, and is generally characterized by bone disease, hepatosplenomegaly, anemia and thrombocytopenia, coagulation abnormalities, lung disease, and no CNS involvement. Type 2 typically has a very severe progression with onset in the first 2 years of life including neurologic disease, hepatosplenomegaly, and lung disease, with death usually between 2 and 4 years due to lung failure. Individuals with type 3 may have onset prior to 2 years of age, but the progression is not as severe and they may survive into the third and fourth decade. Finally, there is 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, and corneal opacities.

Treatment is available in the form of enzyme replacement therapy (ERT), substrate reduction therapy, and chaperone therapy for types 1 and 3. Individuals with type 3 may benefit from bone marrow transplantation. Currently, only supportive therapy is available for type 2.

The incidence of type 1 ranges from 1 in 20,000 to 200,000 in the general population, but it is much more frequent among Ashkenazi Jewish population with an incidence between 1 in 400 and 900. Types 2 and 3 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. Significantly reduced or absent enzyme activity of acid beta-gluco-4sidosidase is diagnostic. Additionally, the biomarker, glucocerebroside, is elevated in symptomatic patients and supports a diagnosis of Gaucher disease (see GPSY / Glucocerebroside, Blood Spot; GPSYP / Glucocerebroside, Plasma; GPSYW / Glucocerebroside, Blood). A targeted variant panel may allow for detection of disease-causing variants in affected patients (GAUP / Gaucher Disease, Mutation Analysis, GBA, Varies). In addition, full sequencing of the GBA gene allows for detection of disease-causing variants in affected patients in whom a targeted variant panel identifies no variants or only a single variant (GBAZ / Gaucher Disease, Full Gene Analysis, Varies).

Reference Values

> or =3.53 nmol/hour/mg protein

An interpretative report will be provided.

Note: Results from this assay do not reflect carrier status because of individual variation of beta-glucosidase enzyme levels.

Interpretation

Individuals affected with Gaucher disease will have enzyme levels less than 3.53 nmol/h/mg protein. In our experience some carriers will also have less than 3.53 nmol/h/mg protein activity.

Cautions

Enzyme levels may be normal in individuals receiving enzyme replacement therapy.

Clinical Reference


**Performance**

**Method Description**

The specimens are incubated with a mix of substrate and internal standard for acid sphingomyelinase (ASM), beta-glucocerebrosidase (ABG), alpha-glucosidase (GAA), alpha-galactosidase (GLA), galactocerebrosidase (GALC) and alpha-L-iduronidase (IDUA). The sample is then purified by liquid-liquid extraction. The extract is evaporated and reconstituted before analysis by tandem mass spectrometry. (Unpublished Mayo method)

**PDF Report**

No

**Day(s) and Time(s) Test Performed**

Specimens are processed Monday through Sunday.

Assay is performed: Varies

**Analytic Time**

5 days

**Maximum Laboratory Time**

10 days

**Specimen Retention Time**

WBC homogenate; 1 month

**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**

82963

**LOINC® Information**
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