Overview

Useful For

Confirmation of a suspected clinical diagnosis of Gaucher disease

Carrier testing for individuals of Ashkenazi Jewish ancestry or who have a family history of Gaucher disease

Prenatal diagnosis of Gaucher disease in at-risk pregnancies

Genetics Test Information

Mutations tested for include N370S, IVS2(+1)G->A, 84GG, R496H, L444P, delta55bp, V394L, and D409H.

Reflex Tests

Test ID	Reporting Name	Available Separately	Always Performed
CULFB	Fibroblast Culture for Genetic Test	Yes	No
CULAF	Amniotic Fluid Culture/Genetic Test	Yes	No
MATCC	Maternal Cell Contamination, B	Yes	No

Testing Algorithm

For prenatal specimens only: If amniotic fluid (non-confluent cultured cells) is received, amniotic fluid culture/genetic test will be added and charged separately. If chorionic villus specimen (non-confluent cultured cells) is received, fibroblast culture for genetic test will be added and charged separately. For any prenatal specimen that is received, maternal cell contamination studies will be added.

See Newborn Screen Follow-up for Gaucher Disease in Special Instructions.

For more information, see Newborn Screening Act Sheet Gaucher Disease: Decreased Acid Beta-Glucosidase in Special Instructions.

Special Instructions

- Molecular Genetics: Congenital Inherited Diseases Patient Information
- Informed Consent for Genetic Testing
- 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

Polymerase Chain Reaction (PCR)

NY State Available

Yes

Specimen

Specimen Type

Varies

Additional Testing Requirements

All prenatal specimens **must be** accompanied by a maternal blood specimen.

-Order MATCC / Maternal Cell Contamination, Molecular Analysis on the maternal specimen.

Shipping Instructions

Specimen preferred to arrive within 96 hours of collection.

Prenatal specimens can be sent Monday through Thursday and **must be received by 5 p.m. CST on Friday** in order to be processed appropriately.

Specimen Required

Patient Preparation: A previous bone marrow transplant from an allogenic donor will interfere with testing. Call 800-533-1710 for instructions for testing patients who have received a bone marrow transplant.

Submit only 1 of the following specimens:

Specimen Type: Whole blood

Container/Tube:

Preferred: Lavender top (EDTA) or yellow top (ACD)

Acceptable: Any anticoagulant

Specimen Volume: 2.6 mL

Collection Instructions:

1. Invert several times to mix blood.

2. Send specimenin original tube.

Specimen Stability Information: Ambient (preferred)/Refrigerated/Frozen

Prenatal Specimen

Due to the complexity of prenatal testing, consultation with the laboratory is required for all prenatal testing.

Specimen Type: Amniotic fluid

Container/Tube: Amniotic fluid container

Specimen Volume: 20 mL

Specimen Stability Information: Refrigerated (preferred)/Ambient

Specimen Type: Chorionic villi

Container/Tube: 15-mL tube containing 15 mL of transport media

Specimen Volume: 20 mg

Specimen Stability Information: Refrigerated

Acceptable:

Specimen Type: Confluent cultured cells

Container/Tube: T-25 flask

Specimen Volume: 2 flasks

Collection Instructions: Submit confluent cultured cells from another laboratory.

Specimen Stability Information: Ambient (preferred)/Refrigerated

Forms

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)

Specimen Minimum Volume

Blood: 0.5 mL
Amniotic Fluid: 10 mL
Chorionic Villi: 5 mg

Reject Due To

All specimens will be evaluated by Mayo Clinic Laboratories for test suitability.

Specimen Stability Information

Specimen Type	Temperature	Time	Special Container
Varies	Varies		

Clinical and Interpretive

Clinical Information

Gaucher disease is a relatively rare lysosomal storage disorder resulting from a deficiency of beta-glucocerebrosidase. Mutations within the beta-glucocerebrosidase gene (*GBA*) cause the clinical manifestations of Gaucher disease. There are 3 major types of Gaucher disease: nonneuropathic (type 1), acute neuropathic (type 2), and subacute neuropathic (type 3). Type 1 Gaucher disease occurs most frequently and is the presentation commonly found among Ashkenazi Jewish patients. The carrier rate of Gaucher disease in the Ashkenazi Jewish population is 1 in 18.

Type 1 disease does not involve nervous system dysfunction; patients display anemia, low blood platelet levels, massively enlarged livers and spleens, lung infiltration, and extensive skeletal disease. The clinical variability in type 1 disease is large, with some patients exhibiting severe disease and others very mild disease.

Eight *GBA* mutations, including the N370S mutation found most commonly in the Ashkenazi Jewish population, are included in this test: delta 55bp, V394L, N370S, IVS2+1, 84GG, R496H, L444P, and D409H. This testing panel provides a 95% detection rate for the Ashkenazi Jewish population and up to a 60% detection rate for the non-Ashkenazi Jewish population. Alternatively, full gene sequencing is available to evaluate for mutations in all coding regions and exon/intron boundaries of the *GBA* gene by ordering GBAZ / Gaucher Disease, Full Gene Analysis.

Reference Values

An interpretive report will be provided.

Interpretation

An interpretive report will be provided.

Cautions

This assay will not detect all of the mutations that cause Gaucher disease. Therefore, the absence of a detectable mutation does not rule out the possibility that an individual is a carrier of or affected with this disease.

Test results should be interpreted in the context of clinical findings, family history, and other laboratory data. Errors in our interpretation of results may occur if information given is inaccurate or incomplete.

Rare polymorphisms exist that could lead to false-negative or false-positive results. If results obtained do not match the clinical findings, additional testing should be considered.

In rare cases, DNA alterations of undetermined significance may be identified.

Clinical Reference

1. Beutler E, Grabowski GA: Glucosylceramide lipidoses: Chapter 146: Gaucher disease. In The Metabolic Basis of Inherited Disease. Edited by CR Scriver, AL Beaudet, WS Sly, D Valle. New York, McGraw-Hill Book Company, 1994

2. Gaucher Disease, Current Issues in Diagnosis and Treatment. Technology Assessment Conference Program and Abstracts, National Institutes of Health, Bethesda, MD, February 27-March 1, 1995

3. Charrow J, Andersson HC, Kaplan P, et al: The Gaucher Registry: Demographics and disease characteristics of 1,698 patients with Gaucher Disease. Arch Int Med 2000;160:2835-2843

4. Gross SJ, Pletcher BA, Monaghan KG: Carrier screening individuals of Ashkenazi Jewish descent. Genet Med 2008;10(1):54-56

Performance

Method Description

A laboratory-developed multiple PCR-based assay is used to detect the following mutations in the *GBA* gene: 84G->GG, IVS2(+1)G->A, N370S, delta 55bp, V394L, D409H, L444P, and R496H.(Fulton R, McDade R, Smith P, et al: Advanced multiplexed analysis with the FlowMetrix system. Clin Chem 1997;43:1749-1756; Ye F, Li MS, Taylor JD, et al: Fluorescent microsphere-based readout technology for multiplexed human single nucleotide polymorphism analysis and bacterial identification. Hum Mutat 2001;Apr;17[4]:305-316)

PDF Report

No

Day(s) and Time(s) Test Performed

Tuesday; 10 a.m.

Analytic Time

9 days

Maximum Laboratory Time

12 days

Specimen Retention Time

Whole Blood: 2 weeks (if available) Extracted DNA: 3 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

81251-*GBA (glucosidase, beta, acid)* (eg, Gaucher disease) gene analysis, common variants (eg, N370S, 84GG, L444P, IVS2+IG>A)

Fibroblast Culture for Genetic Test

88233-Tissue culture, skin or solid tissue biopsy (if appropriate)

88240-Cryopreservation (if appropriate)

Amniotic Fluid Culture/Genetic Test

88235-Tissue culture for amniotic fluid (if appropriate)

88240-Cryopreservation (if appropriate)

Maternal Cell Contamination, B

81265-Comparative analysis using Short Tandem Repeat (STR) markers; patient and comparative specimen (eg, pre-transplant recipient and donor germline testing, post-transplant non-hematopoietic recipient germline [eg, buccal swab or other germline tissue sample] and donor testing, twin zygosity testing or maternal cell contamination of fetal cells (if appropriate)

LOINC® Information

Test ID	Test Order Name	Order LOINC Value
GAUP	Gaucher Disease, Mutation Analysis	35693-1

Result ID	Test Result Name	Result LOINC Value
53164	Result Summary	50397-9
53165	Result	82939-0
53166	Interpretation	69047-9
52428	Additional Information	48767-8
53167	Reason for Referral	42349-1
53168	Specimen	31208-2
53169	Source	31208-2
53170	Released By	18771-6