

## **Gaucher Disease, Full Gene Analysis**

Test ID: GBAMS

**USEFUL FOR:**

- Confirmation of a diagnosis of Gaucher disease
- Carrier screening in cases where there is a family history of Gaucher disease, but an affected individual is not available for testing or disease-causing mutations have not been identified

**METHODOLOGY:** Polymerase Chain Reaction (PCR) Followed by DNA Sequence Analysis

**REFERENCE VALUES:** An interpretive report will be provided.

**SPECIMEN REQUIREMENTS:** Specimen must arrive within 96 hours of collection.

**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:** 3 mL  
**Specimen Stability Information:** Ambient (preferred)/Refrigerated
- **Specimen Type:** Cultured fibroblasts  
**Container/Tube:** T-75 or T-25 flask  
**Specimen Volume:** 1 full T-75 or 2 full T-25 flasks  
**Specimen Stability Information:** Ambient (preferred)/Refrigerated <24 hours
- **Specimen Type:** Skin biopsy  
**Container/Tube:** Sterile container with any standard cell culture media (eg, minimal essential media, RPMI 1640). The solution should be supplemented with 1% penicillin and streptomycin. Tubes can be supplied upon request (Eagle's minimum essential medium with 1% penicillin and streptomycin [Supply T115]).  
**Specimen Volume:** 4-mm punch  
**Specimen Stability Information:** Refrigerated (preferred)/Ambient

**Acceptable:**

- **Specimen Type:** Blood spot  
**Container/Tube:** Whatman Protein Saver 903 Paper  
**Specimen Volume:** 5 blood spots  
**Specimen Stability Information:** Ambient (preferred)/Refrigerated

**CAUTIONS:**

- A small percentage of individuals who are carriers or have a diagnosis of Gaucher disease may have a mutation that is not identified by this method (eg, large genomic deletions, promoter mutations). The absence of a mutation, therefore, does not eliminate the possibility of positive carrier status or the diagnosis of Gaucher disease. For carrier testing, it is important to first document the presence of a *GBA* gene mutation in an affected family member.
- In some cases, DNA alterations of undetermined significance may be identified.
- 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.
- A previous bone marrow transplant from an allogenic donor will interfere with testing. Call Mayo Medical Laboratories for instructions for testing patients who have received a bone marrow transplant.
- 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.
- This is **not** the preferred genetic test for carrier screening or diagnosis in individuals of Ashkenazi Jewish ancestry. For these situations, order ZW185 - Ashkenazi Jewish GBA only Ambry test #1804GBA

**LIST FEE:** \$1,250.00

- For skin biopsy specimens, the following test will be added at an additional charge:  
\$261.10 for #80333 Fibroblast Culture for Genetic Testing  
\$1511.10=Total List Fee

**CPT CODE:**

- 81251 GBA (glucosidase, beta, acid (eg, Gaucher disease) gene analysis, common variants

For non participating payers

- 83891- Isolation or extraction of highly purified nucleic acid
- 83892- Enzymatic digestion
- 83894- Separation by gel electrophoresis
- 83898- Amplification, target, each nucleic acid sequence
- 83909 x 22- Separation and identification by high-resolution technique
- 83912- Interpretation and report

Fibroblast Culture for Genetic Testing

- 88233-Tissue culture, skin or solid tissue biopsy (if appropriate)
- 88240-Cryopreservation (if appropriate)

**DAY(S) SET UP:** Friday; 2 PM

**ANALYTIC TIME:** 5 days

QUESTIONS: Contact your Mayo Medical Laboratories' Regional Manager or Marvin H. Anderson, Jr., MML Laboratory Technologist Resource Coordinator  
Telephone: 800-533-1710