

## Gaucher Disease: Beta-Glucosidase Enzyme Activity, Leukocytes

**Test Code:** LG

**Turnaround time:** 7 days - 10 days

**CPT Codes:** 84155 x1, 82963 x1

### Condition Description

Gaucher disease is the most common lysosomal storage disease, affecting approximately 1 in 40,000 people in the general population and 1 in 450 in the Ashkenazi (Eastern European) Jewish population. The condition is inherited in an autosomal recessive pattern. Individuals with Gaucher disease lack sufficient activity of the enzyme glucocerebrosidase (GBA), which leads to accumulation of glucocerebroside (glucosylceramide) in cells of monocyte/macrophage lineage. These Gaucher cells most often accumulate in the spleen, liver and bone marrow. Symptoms of the disease vary from mild to severe and may appear at any age, from infancy to adulthood. Common symptoms include hepatosplenomegaly, anemia, thrombocytopenia, bone pain and fractures and excessive fatigue. There are 3 forms of Gaucher disease. Type I is the most common form and does not involve the nervous system. Types II and III present with neurological involvement in infancy and in early childhood, respectively.

Diagnosis of Gaucher disease is based on beta-glucosidase enzyme analysis on peripheral blood leukocytes. Enzyme activity is very low in affected individuals. Enzyme analysis is not reliable for detection of gene carriers. Carrier testing may be accomplished through molecular analysis of the GBA gene (see Gaucher mutation analysis).

For further information about Gaucher disease, please call the Emory Lysosomal Storage Disease Center at (404) 778-8565 or (800) 200-1524. For general questions, call EGL Genetics at 470-378-2200.

[Click here](#) for the GeneReviews summary on this condition.

Visit [www.ThinkGenetic.com](http://www.ThinkGenetic.com) for patient-friendly information on [Gaucher disease](#).

### Indications

This test is indicated for individuals with:

- Clinical symptoms of Gaucher disease
- Testing of an affected individual's siblings
- Testing of the children of confirmed carrier parents

### Methodology

Fluorometric Enzyme Assay using artificial 4-MU substrate. Beta-glucosidase activity is quantitated in peripheral blood leukocytes to confirm a diagnosis of Gaucher disease.

### Specimen Requirements

**Submit only 1 of the following specimen types**

**Type: Whole Blood (Sodium Heparin)**

**Specimen Requirements:**

Sodium Heparin (Green Top)  
3-5 ml

**Specimen Collection and Shipping:**

Ship sample at room temperature for receipt at EGL within 24 hours of collection. Do not refrigerate or freeze. Not accepted on Saturday. (Late Friday collections may be stored at room temperature over the weekend for Monday receipt.)

### Related Tests

- [Gaucher Mutation Analysis \(GU\)](#) is a molecular test used for detection of carriers. Identification of two GBA mutations in an at-risk individual may be diagnostic of Gaucher disease.
- Biomarker Monitoring Panel (GM) which includes ACE, CHIT0, TRAP, is used for monitoring progression or response to treatment.
- Prenatal testing may be available to couples who are confirmed carriers of Gaucher disease. Please contact the laboratory genetic counselor to determine the availability of prenatal testing.