

Gaucher Disease: Chitotriosidase (CHITO) Enzyme Activity, Serum

Test Code: CZ

Turnaround time: 7 days - 10 days

CPT Codes: 82657 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.

Biomarker analysis is used to monitor of disease progression or response to treatment in children and adults with Gaucher disease. Elevated levels of chitotriosidase (CHITO) reflect excess lipid storage in Gaucher disease. The levels usually decrease and remain stable with adequate enzyme replacement therapy. Results of these biochemical marker studies should be correlated with clinical findings and other assessments of disease involvement.

For further information about Gaucher disease, please call the Emory Lysosomal Storage Disease Center at (404) 778-8565 or (800) 200-1524. To order shipping kits or test request forms, call EGL Genetics at 470-378-2200.

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

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

Indications

This test is indicated for monitoring patients with a diagnosis of Gaucher disease. Elevated levels of chitotriosidase reflect excess lipid storage in Gaucher disease. The levels usually decrease and remain stable with adequate enzyme replacement therapy. Result should be correlated with clinical findings and other assessments of disease involvement.

Methodology

Fluorometric enzyme activity assay using artificial 4-MU substrate for Chitotriosidase (CHITO).

Reference Range

Normal range: < 78.5 nmol/hr/ml

Specimen Requirements

Submit only 1 of the following specimen types

Type: Whole Blood (No additive)

Specimen Requirements:

No Additive (Red Top) or SST (Serum Separator Tube - no additives)

1-2 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

Type: Serum

Specimen Requirements:

Clean container without additives

1-2 ml

Spin down, transfer, and ship frozen.

Specimen Collection and Shipping:

Ship frozen sample on dry ice with overnight delivery.

Related Tests

- [Biomarker Monitoring Panel \(GM\)](#) which includes ACE, CHITO, TRAP, is used for monitoring progression or response to treatment.
- [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.
- [Beta-Glucosidase Enzyme Analysis \(LG\)](#) is a biochemical test used for definitive diagnosis of Gaucher disease.

- 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.