Gaucher Disease: Biomarker Panel (ACE, CHITO, TRAP) Enzyme Activities, Serum

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 activity levels of angiotensin converting enzyme (ACE), chitotriosidase (CHITO) and tartrate resistant acid phosphatase (TRAP) may 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.

Indications

This test is indicated for monitoring patients with a diagnosis of Gaucher disease.

Elevated activity levels of angiotensin converting enzyme (ACE), chitotriosidase (CHITO) and tartrate resistant acid phosphatase (TRAP) may 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.

Methodology

Fluorometric enzyme activity assay using artificial substrate for Angiotensin Converting Enzyme (ACE), artificial 4-MU substrate for Chitotriosidase (CHITO), and enzymatic reaction detected by spectrophotometric assay to measure Tartrate-resistant acid phosphatase (TRAP) activity.

Reference Range

ACE: Normal range: 32.8 - 107.9 IU/L (70.3 +/- 37.6, n=23), Median: 68.9 IU/L.  
CHITO: Normal range: < 78.5 nmol/hr/ml  
TRAP: Normal range: 0.28-9.84 IU/L (5.1 +/-4.8, n=23), Median: 4.6 IU/L.

We noted that a small portion of the samples we tested have relatively low chitotriosidase activity. It was recently reported (Rodrigues M, et al., 2010) that >11% of the Mexican population have chitotriosidase deficiency, while < 6% was reported in other populations. This means the low end of the reference range previously used by our laboratory is now determined inaccurate due to the possibility of a mixed population in our control group. The reference range of our chitotriosidase activity has therefore been updated to < 78.5 nmol/hr/ml to reflect this newly published information.

Specimen Requirements

Submit only 1 of the following specimen types

* Preferred specimen type: Serum

Type: Serum

Specimen Requirements:

In a no additive (red top) or SST tube: 2-5 ml

Centrifuge to separate serum and freeze.

Specimen Collection and Shipping: Ship frozen sample on dry ice with overnight delivery.

Type: Whole Blood

Specimen Requirements:

Disclaimer: This information is confidential and subject to change without notice. It may not be reproduced in whole or part unless authorized in writing by an authorized EGL representative.
In a no additive (red top) or SST tube: 2-5 ml

Fresh sample (unseparated whole blood) only accepted if received at EGL within 24 hours of collection.

Specimen Collection and Shipping: Ship sample at room temperature for receipt at EGL within 24 hours of collection. Do not refrigerate or freeze.

**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.
- **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.