Gaucher Disease

Gene Tested: *GBA*

Also Known As:

- ß-glucosidase deficiency
- Acid beta-glucosidase deficiency
- Glucosylceramidase deficiency

Disorder:

Gaucher disease is the most common lysosomal storage disease and is caused by decreased activity of ß-glucosidase. Gaucher disease is inherited as an autosomal recessive disorder. It is estimated that about 1 in 40,000 to 1 in 60,000 people are affected with Gaucher disease type 1 worldwide. Gaucher disease occurs more frequently in the Ashkenazi Jewish population at a rate of 1 in 450 to 1 in 1000 individuals. Types 2 and 3 diseases occur in all populations at a rate of about 1 in 100,000.

Non-neuronopathic Gaucher disease, or Gaucher disease Type 1, describes Gaucher disease that does not involve the central nervous system. Symptoms may begin in infancy, childhood, or adulthood and often include hepatosplenomegaly, anemia, thrombocytopenia, and skeletal manifestations, such as bone pain, Erlenmeyer flask deformity, and avascular necrosis. Symptoms are highly variable among individuals.

Neuronopathic Gaucher disease, or Gaucher disease Types 2 and 3, includes the variants of Gaucher disease which affect both the body and brain. Type 2 disease (acute) usually begins early in infancy, progresses quickly, and usually results in death by 2-3 years of age. Type 3 disease (chronic) usually causes symptoms in later infancy or childhood and has a slower progression with prolonged survival.

The diagnosis of Gaucher disease is confirmed by measuring decreased ß-glucosidase activity in leukocytes or other tissues. Molecular determination of the specific *GBA* mutations is recommended in all patients.

Indications:

- Confirmation of diagnosis in a symptomatic individual
- Presymptomatic testing of at-risk relatives
- Carrier identification in individuals with a family history of Gaucher disease
- Prenatal diagnosis of an at-risk fetus, after confirmation of mutations in the parents (by prior arrangement only)

Specimen:

Blood samples should be collected in EDTA (lavender topped) tubes. A minimum of 5 mLs on adult or 3 mLs on child is required for analysis. Blood collected on Friday may be stored in refrigerator until Monday for overnight shipment. If sending DNA you should send at LEAST 10 mg, preferably more.

For other tissue requirements please call 1-800-344-2462, extension 4474.

Testing Methodology:

PCR-based sequencing of all eleven exons and exon/intron boundaries of the *GBA* gene.

Test Sensitivity:

PCR-based sequencing detects two mutations in >99% of patients with Gaucher disease. The sensitivity of DNA sequencing is over 99% for the detection of nucleotide base changes, small deletions and insertions in the regions analyzed. Multiple exon deletions and insertions may not be identified by this methodology but are rare in Gaucher disease. *GBA* is the only gene associated with Gaucher disease.

Turn-Around Time:

Reports are routinely available within 21 days of sample receipt. Abnormal results will be called to the referring physician. All reports will be faxed to the referring physician.

Cincinnati Children's®

Human Genetics

Molecular Genetics Laboratory
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CPT Codes:

- GBA Full Gene Sequencing 81479
- Ashkenazi Panel 81251
- Family Mutation Studies 81479

Please call 1-866-450-4198 for pricing, insurance preauthorization, or with any billing questions.

Results: Each test report includes a detailed interpretation of the genetic findings, the clinical significance of the result, and specific recommendations for clinical management and additional testing, if warranted. Results will be reported to the referring physician or health care provider as specified on the test requisition form.

Additional information and test requisitions are available at: www.cchmc.org/molecular-genetics

Shipping Instructions:

Please enclose **test requisition** with sample.
All information must be completed before sample can be processed.

Place samples in styrofoam mailer and ship at room temperature by overnight Federal Express to arrive Monday through Friday

Ship to:

Cytogenetics and Molecular Genetics Laboratories
3333 Burnet Avenue NRB 1042
Cincinnati, OH 45229
513-636-4474