Cincinnati Children's

Division of Human Genetics

Gaucher Disease

Gene Tested: *GBA*

Molecular Genetics Laboratory
CLIA#: 36D0656333
Phone: (513) 636-4474
Fax: (513) 636-4373
Email: moleculargenetics@cchmc.org

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

Helping you fit the pieces together

Shipping Instructions

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

- All samples must be clearly labeled with the patient's name and birth date and initialed by the phlebotomist.
- All samples should be wrapped, cushioned against breakage and sealed in a plastic bag before placement in the shipping container.
- All samples should be sent at room temperature. Do not allow to freeze.
- Samples should be sent by overnight carrier to arrive Monday through Friday. Do not ship samples on Friday.

Ship to:
Molecular Genetics Laboratory
3333 Burnet Avenue NRB 1042
Cincinnati, OH 45229

Also known as:

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

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.

METHODOLOGY:

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

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 three weeks of sample receipt. Abnormal results will be called to the referring physician. All reports will be faxed to the referring physician.

COST:

Please call 1-866-450-4198 for prices or with any billing questions.

CPT CODES:

GBA Full Gene Sequencing	81479
Ashkenazi Panel	81251
Family Mutation Studies	81403

Updated 12/2011