**Enzyme testing – Blood Test**

The most accurate diagnosis of Gaucher disease is made based on the results of a blood test that measures the level of glucocerebrosidase enzyme activity. In people with Gaucher disease, the amount of enzyme activity is much lower than normal. Enzyme testing is not reliable for detecting Gaucher carriers, since some carriers have a glucocerebrosidase level in the normal range.

**Targeted Mutation Analysis Blood Test**

A few of the known mutations in the gene that codes for glucocerebrosidase are more common in certain ethnic groups. Testing for some of the more common mutations in the Ashkenazi Jewish population, where Gaucher disease occurs more frequently and the carrier frequency is ~ 1 in 15, identifies the majority of affected and carrier individuals. Targeted mutation analysis is also useful for non-Jewish individuals to determine genotype or carrier status although the detection rate is lower in this population.

Listed below are testing centers that conduct enzyme assay and targeted mutation analysis for Gaucher disease within the United States.*  This list was compiled as of May 2008.

<table>
<thead>
<tr>
<th>Laboratory</th>
<th>Ship-To Address</th>
</tr>
</thead>
<tbody>
<tr>
<td>Baylor College of Medicine Medical Genetics Laboratories <a href="http://www.bcmgeneticlabs.org">http://www.bcmgeneticlabs.org</a> Phone: 1-800-411-GENE (4363) Fax: 713-798-6584 E-mail: <a href="mailto:geneticetest@bcm.edu">geneticetest@bcm.edu</a></td>
<td>Baylor College of Medicine Medical Genetics Laboratories 2450 Holcombe Grand Blvd. – Receiving Dock Houston, TX 77021-2024</td>
</tr>
<tr>
<td>Children’s Hospital and Regional Medical Center Biochemical and Molecular Genetics Laboratories <a href="http://www.seattlechildrens.org/geneticslab">http://www.seattlechildrens.org/geneticslab</a> Phone: 206-987-2102 E-mail: <a href="mailto:lisa.snyderman-king@seattlechildrens.org">lisa.snyderman-king@seattlechildrens.org</a></td>
<td>Children’s Hospital and Regional Medical Center Laboratory, A-4901 4800 Sand Point Way NE Seattle, WA 98105</td>
</tr>
<tr>
<td>Emory University, Department of Human Genetics Emory Genetics Laboratory <a href="http://www.geneticslab.emory.edu">www.geneticslab.emory.edu</a> Phone: 1-800-366-1502 Fax: 404-778-8559 E-mail: <a href="mailto:lab@genetics.emory.edu">lab@genetics.emory.edu</a></td>
<td>Emory Genetics Laboratory 2165 N. Decatur Road Decatur, GA 30033</td>
</tr>
<tr>
<td>Genzyme Genetics Molecular Diagnostic Laboratory <a href="http://www.genzymegenetics.com">www.genzymegenetics.com</a> Phone: 800-848-4436 Fax: 505-438-2270</td>
<td>Genzyme Genetics 2000 Vivigen Way Santa Fe, NM 87505</td>
</tr>
<tr>
<td>Mayo Clinic College of Medicine Biochemical Genetics Laboratory <a href="http://www.mayomedicallaboratories.com">www.mayomedicallaboratories.com</a> Phone: 507-266-8158 Fax: 507-266-2888 E-mail: <a href="mailto:Biochemicalgenetics@mayo.edu">Biochemicalgenetics@mayo.edu</a></td>
<td>Mayo Medical Laboratories 3050 Superior Drive NW Rochester, MN 55901-1995</td>
</tr>
<tr>
<td>Mount Sinai School of Medicine Biochemical and Molecular Diagnostic Laboratories <a href="http://www.mssm.edu/gaucher">www.mssm.edu/gaucher</a> Phone: 212-241-0432 Fax: 212-241-0139</td>
<td>Mount Sinai School of Medicine Genetics and Genomic Sciences Atran Laboratory Building Room AB2-32 1428 Madison Avenue New York, NY 10029 Atrn: Genetic Testing</td>
</tr>
<tr>
<td>New York University School of Medicine NYU Neurogenetics Laboratory Phone: 212-263-8344 Fax: 212-263-1018</td>
<td>NYU Neurogenetics Laboratory 400 East 34 th St, RR213 New York, NY 10016</td>
</tr>
</tbody>
</table>

Contact the laboratories listed above for information on obtaining test requisitions, sample requirements, and turnaround time. For additional information about laboratories that offer diagnostic testing for Gaucher Disease, visit [www.genetests.org](http://www.genetests.org)

*This listing includes only laboratories that have agreed to be listed by Genzyme and is not intended to be exhaustive. It is for informational purposes only, and no endorsement of or representations regarding the services offered are either intended or implied. Please note that, while Genzyme has endeavored to obtain information that is current as of the time of publication, it makes no representation as to accuracy, and physicians are directed to the individual laboratories for the specific details of the services provided.*
Gaucher disease (pronounced go-shay) is the most common lysosomal storage disorder. It is caused by a deficiency in the enzyme glucocerebrosidase. Insufficient enzyme activity in Gaucher patients results in progressive accumulation of glucocerebroside in the macrophages. Clinical symptoms arise due to the displacement of normal cells by lipid-engorged Gaucher cells. Accumulation occurs in organs throughout the body, typically the liver, spleen, and bone marrow.

Gaucher disease is multisystemic and its symptomatology is extremely heterogeneous. Gaucher disease may progress and result in pathological features that may be difficult or impossible to reverse.

**Key Facts**

- Gaucher disease is not gender specific
- Can be diagnosed with blood tests
- Diagnosed at any age
- Gaucher disease is more prevalent than Tay-Sachs
- Gaucher disease is treatable

**Hematologic**

- Anemia/thrombocytopenia

**Visceral**

- Hepatosplenomegaly

**Skeletol**

- Bone pain/bone crises
- Growth retardation
- Avascular necrosis
- Pathologic fractures
- Osteopenia

The definitive diagnosis of Gaucher disease is determination of deficient glucocerebrosidase enzyme activity in either leukocytes or cultured skin fibroblasts.

**Genetics**

- Autosomal recessive
- Panethnic, high prevalence in persons of Ashkenazi Jewish ancestry
- More than 200 mutations have been identified

**Gaucher Disease Inheritance**

When both parents are carriers of the disease-causing genetic mutation, there is a 25% chance with each pregnancy of having a child affected with Gaucher disease, a 50% chance of having a child who carries the gene mutation, and a 25% chance of having a child who does not carry the mutation. Males and females have an equal chance of being affected.

**For more information about Gaucher disease, go to:**

- [gaucherregistry.com](http://www.gaucherregistry.com) for managing and monitoring guidelines
- [genetests.org](http://www.genetests.org) for testing information
- [gauchercare.com](http://www.gauchercare.com) for more information on testing and other resources, or call: Genzyme Medical Information: 1-800-745-4447, option 2

References: