

Diagnostic Lab List

Updated May 2008

Gaucher Disease

Labs Offering Diagnostic Testing for Gaucher Disease, both enzyme assay and targeted mutation analysis

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.

Laboratory	Ship-To Address
Baylor College of Medicine Medical Genetics Laboratories http://www.bcmgeneticlabs.org Phone: 1-800-411-GENE (4363) Fax: 713-798-6584 E-mail: geneticstest@bcm.edu	Baylor College of Medicine Medical Genetics Laboratories 2450 Holcombe Grand Blvd. - Receiving Dock Houston, TX 77021-2024
Children's Hospital and Regional Medical Center Biochemical and Molecular Genetics Laboratories http://www.seattlechildrens.org/geneticslab Phone: 206-987-2102 E-mail: lisa.sniderman-king@seattlechildrens.org	Children's Hospital and Regional Medical Center Laboratory, A-6901 4800 Sand Point Way NE Seattle, WA 98105
Emory University, Department of Human Genetics Emory Genetics Laboratory www.geneticslab.emory.edu Phone: 1-800-366-1502 Fax: 404-778-8559 E-mail: lab@genetics.emory.edu	Emory Genetics Laboratory 2165 N. Decatur Road Decatur, GA 30033
Genzyme Genetics Molecular Diagnostic Laboratory www.genzymegenetics.com Phone: 800-848-4436 Fax: 505-438-2270	Genzyme Genetics 2000 Vivigen Way Santa Fe, NM 87505
Mayo Clinic College of Medicine Biochemical Genetics Laboratory www.mayomedicallaboratories.com Phone: 507-266-8158 Fax: 507-266-2888 E-mail: Biochemicalgenetics@mayo.edu	Mayo Medical Laboratories 3050 Superior Drive NW Rochester, MN 55901-1995
Mount Sinai School of Medicine Biochemical and Molecular Diagnostic Laboratories www.mssm.edu/gaucher Phone: 212-241-0432 Fax: 212-241-0139	Mount Sinai School of Medicine Genetics and Genomic Sciences Atran Laboratory Building Room AB2-32 1428 Madison Avenue New York, NY 10029 Attn: Genetic Testing
New York University School of Medicine NYU Neurogenetics Laboratory Phone: 212-263-8344 Fax: 212-263-1018	NYU Neurogenetics Laboratory 400 East 34 th St , RR213 New York, NY 10016

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

*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.