

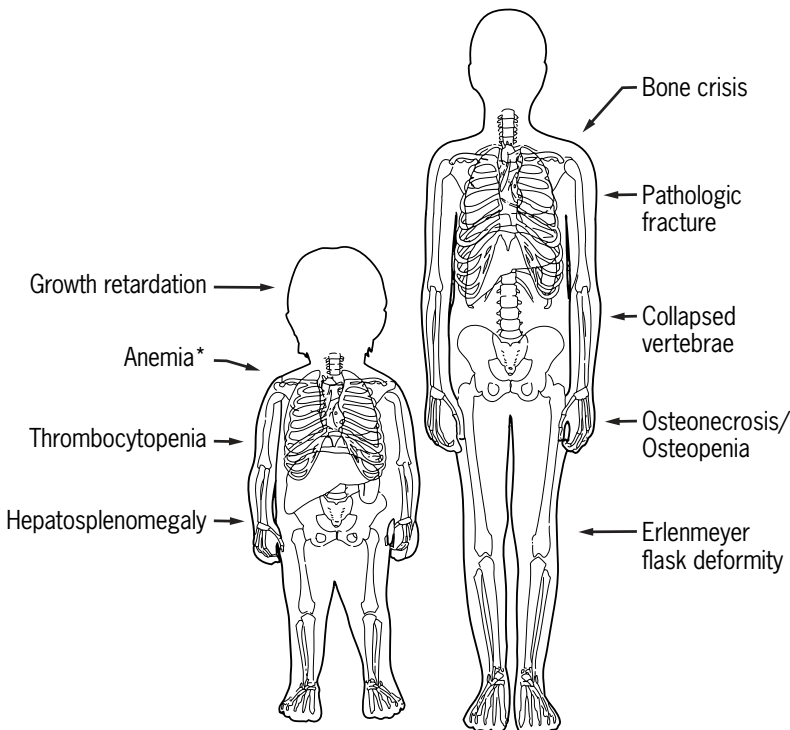
Does your patient have type 1 Gaucher disease?

A combination of symptoms may help distinguish type 1 Gaucher disease— an unpredictably progressive, debilitating, and potentially life-threatening disorder from more common conditions.

Type 1 Gaucher disease is the most common lysosomal storage disorder and is caused by a deficiency of the enzyme glucocerebrosidase, leading to the accumulation of a fatty substance called glucocerebroside in multiple organs. The disease affects individuals of any ethnicity (~1:50,000+) but may be as high as ~1:850 among individuals of Ashkenazi descent (Eastern European).^{1,2}

Clinical onset may occur at any age, but approximately 66% of patients have disease manifestation in childhood.³ Children may experience irreversible disease if treatment is not initiated at the earliest signs and symptoms.⁴

Most Common Symptoms at Time of Diagnosis^{4,5}

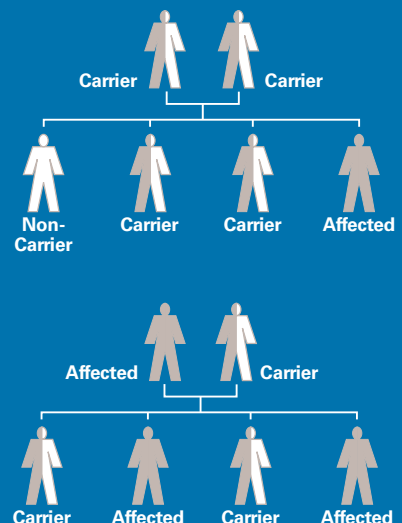


*Hgb levels <11 g/dL for males 12 years and older; <10 g/dL for females 12 years and older; <9.5 g/dL for children ages 2 to <12 years; <8.5 g/dL for children ages 6 months to <2 years; <9.1 g/dL for children younger than 6 months of age.²

Questions to Ask or Observe

1. Is there an enlarged abdomen?
Yes No
2. Is patient usually lethargic?
Yes No
3. Does patient bruise easily?
Yes No
4. Does patient experience prolonged bleeding?
Yes No
5. Does patient complain of bone pain?
Yes No
6. Does patient have a history of broken bones?
Yes No
7. Are you aware of anyone in the family who was diagnosed with Gaucher disease?
Yes No

Likelihood that a child will be a carrier of, or affected by, type 1 Gaucher disease



Take Action

Don't ignore the warning signs

Clinical suspicion

- Finding of a unique sign or symptom
- Presentation of a cluster of common signs and symptoms

Urgent referral

- Refer your patient to a geneticist or metabolic specialist

Definitive diagnosis

Enzyme assay is the gold standard. In general, a reading of <30% of normal activity confirms the diagnosis, although this may vary by laboratory.⁶

For more information, please contact Genzyme Medical Information at 1-800-745-4447, option 2.

Laboratory

Baylor College of Medicine
Medical Genetics Laboratories
www.bcm.edu/geneticlabs
Phone: 800-411-GENE (4363)
Fax: 713-798-6584
E-mail: genectest@bcm.edu

Children's Hospital and Regional
Medical Center
Biochemical and Molecular
Genetics Laboratories
www.seattlechildrens.org/
geneticslab
Phone: 206-987-2102
E-mail: lisa.sniderman-king@
seattlechildrens.org

Emory University
Dept of Human Genetics
Emory Genetics Laboratory
www.geneticslab.emory.edu
Phone: 800-366-1502
Fax: 404-778-8559
E-mail: lab@genetics.emory.edu

Genzyme Genetics
Molecular Diagnostic Laboratory
www.genzymegenetics.com
Phone: 800-848-4436
Fax: 505-438-2270

Mayo Clinic College of Medicine
Biochemical Genetics Laboratory
www.mayomedicallaboratories.com
Phone: 507-266-8158
Fax: 507-266-2888
E-mail: biochemicalgenetics@
mayo.edu

Ship-to Address

Baylor College of Medicine
Medical Genetics Laboratories
Grand Blvd – Receiving Dock
2450 Holcombe Blvd
Houston, TX 77021-2024

Children's Hospital and
Regional Medical Center
Laboratory, A-6901
4800 Sand Point Way NE
Seattle, WA 98105

Emory Genetics Laboratory
2165 N Decatur Rd
Decatur, GA 30033

Genzyme Genetics
2000 Vivigen Way
Santa Fe, NM 87505

Mayo Medical Laboratories
3050 Superior Dr NW
Rochester, MN 55901-1995

Laboratory

Mount Sinai School of Medicine
Biochemical and Molecular
Diagnostics
www.mssm.edu/gaucher
Phone: 212-241-6927
Fax: 212-860-3316
E-mail: jane.robinowitz@mssm.edu

New York University School
of Medicine
NYU Neurogenetics Laboratory
Phone: 212-263-8344
Fax: 212-263-1018
E-mail: paula.torres@nyumc.org

Ship-to Address

Mount Sinai School
of Medicine
Genetics and Genomic
Sciences
Atran Laboratory Building
Rm AB2-32
1428 Madison Ave
New York, NY 10029
Attn: Genetic Testing

NYU Neurogenetics
Laboratory
400 East 34th St, RR213
New York, NY 10016
Attn: Paula Torres

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.

References: 1. National Library of Medicine Genetics Home Reference. Gaucher's disease. <http://ghr.nlm.nih.gov/condition=gaucherdisease>. Accessed July 7, 2009. 2. Beutler E, Grabowski GA. Chapter 146: Gaucher Disease. Scriver's Online Metabolic & Molecular Bases of Inherited Disease. http://www.ommbid.com/OMMBID/the_online_metabolic_and_molecular_bases_of_inherited_disease/b/abstract/part16/ch146. Accessed July 7, 2009. 3. Charrow J, Andersson HC, Kaplan P, et al. The Gaucher Registry: demographics and disease characteristics of 1698 patients with Gaucher disease. *Arch Intern Med*. 2000;160(18):2835-2843. 4. Kaplan P, Andersson HC, Kacena KA, Yee JD. The clinical and demographic characteristics of nonneuronopathic Gaucher disease in 887 children at diagnosis. *Arch Pediatr Adolesc Med*. 2006;160(6):603-608. 5. Wenstrup RJ, Roca-Espiau M, Weinreb NJ, Bembi B. Skeletal aspects of Gaucher disease: a review. *Br J Radiol*. 2002;75(suppl 1):A2-A12. 6. Aerts JMFG, Boot RG, Renkema GH, et al. Molecular and biochemical abnormalities of Gaucher disease: chitotriosidase, a newly identified biochemical marker. *Semin Hematol*. 1995;32(suppl 1):10-13.

