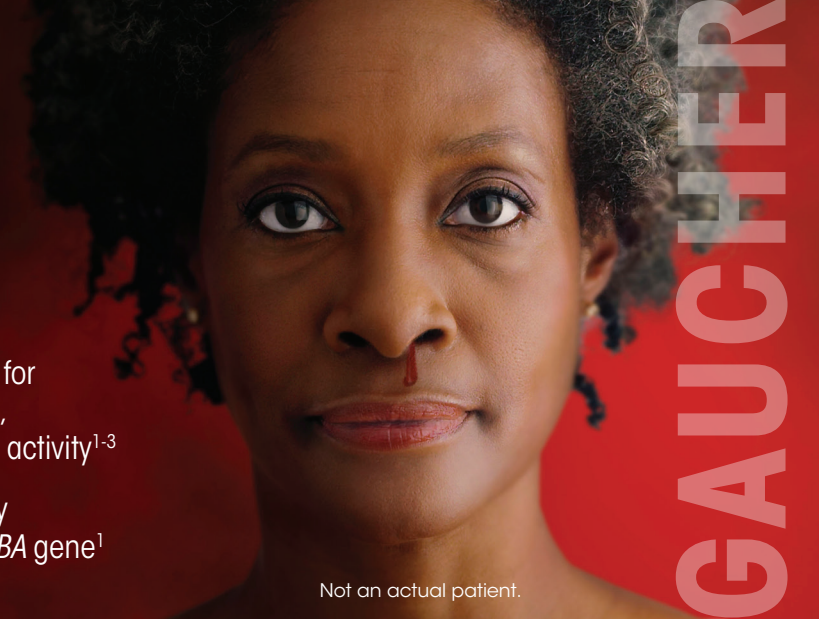
Diagnostic Testing for Gaucher Disease



Not an actual patient.

Suspect Gaucher disease? Test to know

- Enzyme assay is the standard, recommended method for establishing a definitive diagnosis of Gaucher disease, which is demonstrated by deficiency of ß-glucosidase activity[1-3]
- Genetic testing can also diagnose Gaucher disease by demonstrating 2 pathogenic variants *in trans* in the *GBA* gene[1]

This simple diagnostic algorithm may help you know when to test for Gaucher disease in the general population[1]

Splenomegaly and/or thrombocytopenia?

YES

Hematologic malignancies?

NO

Gaucher disease?

PERFORM GAUCHER ENZYME TEST*†

Blood-based enzyme assay (acid beta-glucosidase) is the **gold standard** for definitive diagnosis of Gaucher disease[1]

In patients of Ashkenazi Jewish ancestry, it is prudent to test for Gaucher disease as a first-line investigation in patients presenting with splenomegaly and cytopenia[1]

› **Bone marrow biopsy is not required to diagnose Gaucher disease** because although bone marrow biopsy specimens can help rule out hematologic malignancies, they are not reliable for establishing the presence of Gaucher cells[1]

Commercial lab testing, coding, and obtaining reimbursement are the responsibility of the provider submitting a claim for the item or service.

To see a listing of labs where Gaucher enzyme tests and DNA tests can be performed, please see reverse side.

*Acid ß-glucosidase enzyme activity assays.
†Algorithm is modified from original reference.

Testing Options for Gaucher Disease

Some laboratories offering diagnostic testing for Gaucher disease are listed below. There may be other diagnostic testing appropriate for your patient, and this is not an endorsement of any specific lab. Other testing options can be found at www.concertgenetics.com or www.ncbi.nlm.nih.gov/gtr. Consult each laboratory for a full range of options. Content is current at time of publication and tests may not be available in all states. Please call laboratory to confirm test availability, sample shipping information, and all other logistics. Sanofi does not review or control the content of non–Sanofi websites. This listing does not constitute an endorsement by Sanofi of information provided by any other organizations.

Lab	Available Testing	Test Code	Sample Requirements	Kits	Avg TAT	Mobile Blood Draw	Billing	Contact
ARUP Laboratories	Enzyme	2014459	WB: 3 ml ACD (yellow), EDTA (lavender), or sodium heparin (green) tube	No	3-10 d	No	Inst	P: 800-522-2787 E: clientservices@aruplab.com W: www.aruplab.com
	Sequencing	3001648	WB: 3 ml ACD (yellow), EDTA (lavender) tube		2-3 wks			
Centogene	Enzyme	N/A	WB: 5 ml EDTA (lavender) tube; DBS card: 10 circles	Blood, DBS, Saliva	7 d	Yes	Inst, Self-pay, Ins	P: 617-580-2102 E: customer.support-US@centogene.com W: www.centogene.com
	Sequencing (+/- Del/Dup)		WB: 1 ml EDTA (lavender) tube; DBS card: 10 circles; Saliva, buccal swab		15 d			
	Lyso-GL-1		WB: 1 ml EDTA (lavender) tube; DBS card: 10 circles		7 d			
Greenwood Genetic Center	Enzyme	N/A	WB: 5-10 ml heparin (green) tube; DBS card: 3 circles	Blood, DBS, Saliva	2 wks	No	Inst, Self-pay, Ins (SC only)	P: 800-473-9411 E: labgc@ggc.org W: www.ggc.org
	Sequencing		WB: 5-6 ml EDTA (lavender) tube; DBS card: 3 circles; Saliva		3 wks			
Labcorp Women's Health	Enzyme	451780	WB: 2 x 10 ml EDTA (lavender) tube (peds 1 x 10 ml)	Blood, Buccal	3-13 d	Yes	Inst, Ins, Self-pay	Labcorp Customers: P: 800-345-4363 W: www.labcorp.com Labcorp Women's Health Customers: E: https://womenshealth.labcorp.com/contact-us W: https://womenshealth.labcorp.com/
	Sequencing	451910	WB: 3-7 ml EDTA (lavender) or ACD (yellow) tube; Buccal Note: For sequencing done via "Inheritest Gene-Specific Sequencing," provider can select *GBA*		14-21 d			
The Lantern Project (Performed at PerkinElmer Genomics)	Enzyme	N/A	WB: 2-10 ml heparin (green) tube (volume varies with age); DBS card: 3 circles	Blood, DBS, Saliva	3 d	Yes	No charge*	P: 866-354-2910 E: genomics@perkinelmer.com W: www.LanternProjectDx.com
	Sequencing		WB: 2-10 ml EDTA (lavender) tube (volume varies with age); DBS card: 3 circles; Saliva		3 wks			
	Lyso-GL-1		WB: 2-10 ml EDTA (lavender) tube (volume varies with age); DBS card: 3 circles		3 d			
Mayo Clinic Laboratories	Enzyme	BGL	WB: 6 ml ACD (yellow) tube	DBS, Saliva	5-10 d	Yes	Inst, Ins (can be billed in some cases but account required)	P: 800-533-1710 E: mcl@mayo.edu W: www.mayocliniclabs.com
	Sequencing	GBAZ	WB: 3 ml EDTA (lavender) or ACD (yellow) tube; DBS card: 2-5 circles		14-20 d			
	Lyso-GL-1	GPSY, GPSYP, or GPSYW	WB: 1 ml EDTA (lavender), ACD B (yellow), or sodium heparin (green); Plasma: 0.3 ml; DBS card: 2 circles		2-8 d			
Seattle Children's Hospital	Enzyme	LAB2840	WB: 10 ml ACD (yellow) or sodium heparin (green) tube	DBS, Saliva	7-10 d	No	Inst, Self-pay, Ins	P: 206-987-2617 E: labGC@seattlechildrens.org W: https://seattlechildrenslab.testcatalog.org
	Sequencing	LAB1850	WB: 1-3 ml EDTA (lavender) or ACD (yellow) tube		2-3 wks			
Sema4	Enzyme	N/A	WB: 5-10 ml heparin (green) tube	Blood, Saliva	7 d	Yes	Inst, Self-pay, Ins	P: 800-298-6470 E: clientservices@sema4.com W: www.sema4.com
	Sequencing		WB: 2 x 5-10 ml ACD (yellow) OR 2 x 5-10 ml EDTA (lavender); Saliva		14 d			
	Lyso-GL-1		WB: 1-2 ml EDTA (lavender) or heparin (green) tube; Frozen plasma: 0.5-1 ml		5 d			

*__Testing is performed at no charge. Local charges may apply for sample collection, processing, or shipping.__

ACD=acid citrate; Avg TAT=average turnaround time; d=days; DBS=dried blood spot; Del=deletion; Dup=duplication; EDTA=ethylenediaminetetraacetic acid; GPSY=glucopsychosine; Ins=insurance; Inst=institution; N/A=not applicable; peds=pediatric patients; WB=whole blood; wks=weeks.

References: 1. Mistry PK, Cappellini MD, Lukina E, et al. A reappraisal of Gaucher disease-diagnosis and disease management algorithms. *Am J Hematol.* 2011;86(1):110-115. doi:10.1002/ajh.21888 **2.** 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. doi:10.1001/archinte.160.18.2835 **3.** Pastores GM, Hughes DA. Gaucher disease. In: Adam MP, Ardinger HH, Pagon RA, et al, eds. *GeneReviews®*. Seattle (WA): University of Washington, Seattle. Published July 27, 2000. Updated June 21, 2018. Accessed April 27, 2022. https://www.ncbi.nlm.nih.gov/books/NBK1269

MAT-US-2019959_v2.0_05/2022