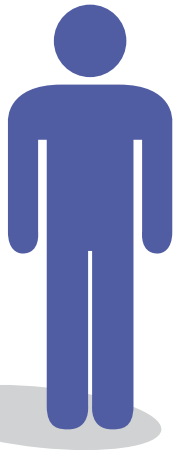


## DIAGNOSTIC TESTING FOR GAUCHER DISEASE

### Suspect Gaucher disease? Test to know

- Enzyme assay is the standard, recommended method for establishing a confirmatory diagnosis of Gaucher disease, which is demonstrated by deficiency of  $\beta$ -glucosidase activity<sup>1-3</sup>
- Molecular testing (DNA) can be used to confirm the type of Gaucher disease as well as carrier status<sup>1</sup>

This simple diagnostic algorithm may help you know when to test for **Gaucher disease**<sup>1</sup>



**PERFORM GAUCHER ENZYME TEST**

### THESE OTHER TESTING METHODS ARE TYPICALLY NOT NECESSARY FOR DIAGNOSIS

- Demonstration of two pathogenic alleles in the *GBA* gene can provide additional diagnostic confirmation, but is not generally used for diagnosis in lieu of biochemical testing<sup>1</sup>
- Bone marrow biopsy is not recommended to diagnose Gaucher disease because although bone marrow biopsy specimens can rule out hematologic malignancies, they are not reliable for establishing the presence of Gaucher cells<sup>1</sup>

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.

## Testing Options for Gaucher Disease

Some of the laboratories offering diagnostic testing for Gaucher type 1 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](http://www.concertgenetics.com) or [www.ncbi.nlm.nih.gov/gtr](http://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 Genzyme does not review or control the content of non-Sanofi Genzyme websites. This listing does not constitute an endorsement by Sanofi Genzyme 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: <a href="mailto:clientservices@aruplab.com">clientservices@aruplab.com</a> W: <a href="http://www.aruplab.com">www.aruplab.com</a>
	Sequencing		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: <a href="mailto:customer.support-US@centogene.com">customer.support-US@centogene.com</a> W: <a href="http://www.centogene.com">www.centogene.com</a>
	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			
EGL Genetics/ Eurofins	Enzyme	LG	WB: 3-5 ml heparin (green) tube	Blood, DBS, Saliva	7-10 d	No	Inst, Self-pay, Ins	P: 855-831-7447 E: <a href="mailto:eglics@egl-eurofins.com">eglics@egl-eurofins.com</a> W: <a href="http://www.egl-eurofins.com">www.egl-eurofins.com</a>
	Sequencing	AK	WB: 2-10 ml EDTA (lavender) tube (volume varies with age); Saliva per Oragene kit		6 wks			
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: <a href="mailto:labgc@ggc.org">labgc@ggc.org</a> W: <a href="http://www.ggc.org">www.ggc.org</a>
	Sequencing		WB: 5-7 ml EDTA (lavender) tube; DBS card: 3 circles; Saliva		3 wks			
LabCorp/ Integrated Genetics	Enzyme - LabCorp	451780	WB: 2 x 10 ml EDTA (lavender) tube (peds 1 x 10 ml)  WB: 3-7 ml EDTA (lavender) or ACD (yellow) tube; Buccal Note: For sequencing done via "Inheritest Gene-Specific Sequencing" provider can select GBA gene	Blood, Buccal	7-13 d	Yes	Inst, Ins, Self-pay	LabCorp Customers: P: 800-345-4363 W: <a href="http://www.labcorp.com">www.labcorp.com</a>  Integrated Customers: P: 800-848-4436 E: <a href="mailto:askIGclientservices@integratedgenetics.com">askIGclientservices@integratedgenetics.com</a> W: <a href="http://www.integratedgenetics.com">www.integratedgenetics.com</a>
	Enzyme - Integrated	370						
	Sequencing - LabCorp	511048						
	Sequencing - Integrated	451910			9-15 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: <a href="mailto:genomics@perkinelmer.com">genomics@perkinelmer.com</a> W: <a href="http://www.LanternProjectDx.com">www.LanternProjectDx.com</a>
	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: <a href="mailto:mcl@mayo.edu">mcl@mayo.edu</a> W: <a href="http://www.mayocliniclabs.com">www.mayocliniclabs.com</a>
	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			
National Gaucher Foundation Diagnostic Testing	Enzyme	N/A	Contact the National Gaucher Foundation at <a href="http://www.gaucherdisease.org/intakeform">www.gaucherdisease.org/intakeform</a>			Yes	No charge*	W: <a href="http://www.gaucherdisease.org/intakeform">www.gaucherdisease.org/intakeform</a>
	Sequencing							
Quest Diagnostics	Enzyme	38195	WB: 6 ml ACD (yellow) tube	Blood	9 d	No	Inst, Self-pay	P: 866-697-8378 W: <a href="http://www.questdiagnostics.com">www.questdiagnostics.com</a>
Seattle Children's Hospital	Enzyme	B Glu Gau	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: <a href="mailto:labGC@seattlechildrens.org">labGC@seattlechildrens.org</a> W: <a href="https://seattlechildrenslab.testcatalog.org">https://seattlechildrenslab.testcatalog.org</a>
	Sequencing	GAUCHSEQ	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: <a href="mailto:clientservices@sema4.com">clientservices@sema4.com</a> W: <a href="http://www.sema4.com">www.sema4.com</a>
	Sequencing		WB: 5-10 ml ACD (yellow) AND 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.

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

References: 1. Mistry PK, et al. *Am J Hematol*. 2011;86:110-115. 2. Charrow J, et al. *Arch Intern Med*. 2000;160:2835-2843. 3. Pastores GM, Hughes DA. 2000. GeneReviews. Seattle, WA; University of Washington, Seattle; 2013. <http://www.ncbi.nlm.nih.gov/books/NBK1269>. Accessed August 31, 2018.