

Suspect Gaucher Disease? Get tested.

Information for you and your health care professional

Once Gaucher disease is suspected, testing is straightforward. It's called a Gaucher enzyme test, and it's just a simple blood test. Your doctor can collect a blood sample and send it to a laboratory for testing, often at little or no cost to you.

Talk to your doctor about getting tested:

- If you are experiencing signs and symptoms of Gaucher disease, such as:



BONE PAIN



FREQUENT NOSEBLEEDS



FATIGUE



BRUISING EASILY

- If you are of Ashkenazi Jewish descent
- If someone in your family has been diagnosed with Gaucher disease

When your doctor may suspect Gaucher disease



OR



AND



If you have an enlarged spleen

(called splenomegaly)

- The spleen may swell up to 15 times normal size
- The abdomen may protrude, appearing overweight or pregnant

Your blood platelet count is low

(called thrombocytopenia)

- Lowered ability to form clots
- Increased tendency for bruising and bleeding
- Frequent heavy nosebleeds and bleeding gums

Your doctor rules out any blood cancer or disease

(called hematologic malignancies)

THEN

Your doctor will probably want to perform a simple blood test for Gaucher disease

Enzyme testing is the gold standard for diagnosing Gaucher disease.

In healthy individuals, the test shows white blood cells that contain normal enzyme activity; in individuals with Gaucher disease, enzyme activity is much lower. Your doctor can explain more about how this simple test helps diagnose Gaucher disease.

Testing information for you and your health care professional

Labs across the United States offer diagnostic tests for Gaucher disease. Testing sample requirements depend on the type of test your physician may perform and can include: whole blood, dried blood spot, saliva and buccal swab. Below are some available options, but this is not an all-inclusive list.

LAB	TESTS	TIMING	FREE SERVICE	CONTACT
ARUP Laboratories	Enzyme	3-10 days	No	1-800-522-2787 clientservices@aruplab.com www.aruplab.com
Centogene	Enzyme	7 days	No	1-617-580-2102 www.centogene.com
	Sequencing	15 days		
	Lyso-GL-1 Biomarker	7 days		
Children's Hospital of Philadelphia (CHOP)	Enzyme	2-3 weeks	No	1-215-590-3394 metabolicclab@email.chop.edu www.chop.edu
Duke University Hospital	Enzyme	15 days	No	1-919-549-0445 https://testcatalog.duke.edu
EGL Genetics/Eurofins	Enzyme	7-10 days	No	1-855-831-7447 eglcs@egl-eurofins.com www.egl-eurofins.com
	Sequencing	6 weeks		
Greenwood Genetic Center	Enzyme	2 weeks	No	1-800-473-9411 labgc@ggc.org www.ggc.org
	Sequencing	3 weeks		
LabCorp/Integrated Genetics	Enzyme	7-14 days	No	LabCorp Customers: 1-888-522-2677 www.labcorp.com Integrated Customers: 1-800-848-4436
	Sequencing	9-15 days		
The Lantern Project (Performed at PerkinElmer Genomics)	Enzyme	3 days	Yes*	1-866-354-2910 genomics@perkinelmer.com www.lanternprojectdx.com
	Sequencing	3 weeks		
Mayo Clinic Laboratories	Enzyme	8-15 days	No	1-800-533-1710 mcl@mayo.edu www.mayocliniclabs.com
	Sequencing	14-20 days		
	Lyso-GL-1 Biomarker	2-8 days		
National Gaucher Foundation Diagnostic Testing Initiative	Enzyme	N/A	Yes*	www.gaucherdisease.org/intakeform
	Sequencing			
Quest Diagnostics	Enzyme	9 days	No	1-866-697-8378 www.questdiagnostics.com
Seattle Children's Hospital	Enzyme	7-10 days	No	1-206-987-2617 labGC@seattlechildrens.org https://seattlechildrenslab.testcatalog.org
	Sequencing	2-3 weeks		
Sema4	Enzyme	7 days	No	1-800-298-6470 clientservices@sema4.com www.sema4.com
	Sequencing	14 days		
	Lyso-GL-1 Biomarker	5 days		

*Testing is performed at no charge, but local charges may apply for sample collection, processing, or shipping.

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