

Suspect Gaucher disease type 1?

Make history for your family

Test for Gaucher disease type 1 — A guide for you and your healthcare team

Gaucher disease type 1 (GD1) is a rare disease that is passed down through families. Some people can't feel the symptoms of GD1, and it is often misdiagnosed. However, GD1 is a lifelong disease that can get worse over time. Confirming a diagnosis of GD1 as soon as possible is the best way to start managing GD1 and to help potentially avoid severe, irreversible damage.

GD1 is an **inherited, autosomal recessive** disorder

Inherited

The genetic variant is passed from parents to their children

Autosomal

Affects both males and females

Recessive

The genetic variant must be passed down from both parents for the child to be affected by GD1

Test early, treat early with family screening

When someone tests positive for GD1, there is a high likelihood that other family members are at risk for the disease as well.

GD1 can damage organs and bones, even when it is not causing pain

The signs and symptoms of GD1 are caused by the buildup of a compound called GL-1. GL-1 buildup happens in bones and various organs throughout the body.



Enlarged spleen



Enlarged liver



Blood diseases



Bone diseases

Treatment can help manage certain symptoms and can lower the risk of permanent complications

Confirm with enzyme testing

Enzyme blood tests can detect abnormalities that can tell your doctor you have GD1. This type of test is considered the gold standard for diagnosing GD1.

Learn more with genetic testing

Genetic testing can sometimes provide additional details about the disease and can determine which other family members are at risk for GD1. Gaucher disease can also be diagnosed by genetic testing.

Testing information for your healthcare team

Below is a list of labs in the United States that perform diagnostic tests for GD1. *You can share this list with your healthcare team to help start the testing process.

Testing sample requirements depend on the type of test your physician chooses. Your doctor may request a sample of your blood and/or saliva, or an oral swab.

*This is not an exhaustive list of labs offering these services nor an endorsement of any one lab. Other testing options can be found at www.concertgenetics.com or www.ncbi.nlm.nih.gov/gtr. Content is current at time of printing 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. These listings do not constitute an endorsement by Sanofi of information provided by any other organization.

Diagnostic Lab	Enzyme testing	Genetic testing	Sponsored†	Contact (for healthcare professionals only)
[Lab name]	[Y/N]	[Y/N]	[Y/N]	Phone: [XXX-XXX-XXXX] Email: [labname@web.com] Website: [labname.com]
[ARUP Laboratories]	[✓]	[✓]	[—]	Phone: [800-522-2787] Email: [clientservices@aruplab.com] Website: [www.aruplab.com]
[Centogene]	[✓]	[✓]	[—]	Phone: [617-580-2102] Email: [customer.support-US@centogene.com] Website: [www.centogene.com]
[Greenwood Genetic Center]	[✓]	[✓]	[—]	Phone: [800-442-4363] Email: [labgc@ggc.org] Website: [www.ggc.org]
[LabCorp]	[✓]	[✓]	[—]	Phone: [800-345-4363] Website: [www.labcorp.com]
[Mayo Clinic Laboratories]	[✓]	[✓]	[—]	Phone: [800-533-1710] Email: [mcl@mayo.edu] Website: [www.mayocliniclabs.com]
[Revvity Omics (including The Lantern Project)]	[✓]	[✓]	[✓]	Phone: [866-354-2910] Email: [genomics@revvity.com] Website: [www.LanternProjectDx.com] Website: [www.revvity.com]

The above is a selection of laboratories offering Gaucher disease enzyme assays (acid beta-glucosidase) or *GBA1* sequencing.

†Testing is performed at no charge; local charges may apply for sample collection, or shipping.