National
FABRY DISEASE
Foundation

www.fabrydisease.org
info@fabrydisease.org
800-651-9131

GENETIC TESTING IN THE U.S.

Programs are available to make genetic testing for Fabry disease accessible and available free of charge.

To qualify for free testing and individual must:

- Live in the United States
- Have a US healthcare provider (HCP) to order your testing
- Have one of the following: a family history of Fabry disease, symptoms that are suggestive of Fabry disease, or an abnormal enzyme screening test.

INVITAE

Questions?
800-436-3037

Detect Lysosomal Storage Diseases Program

GLA gene analysis (Test code: “Individual Genes” “GLA”)
https://www.invitae.com/us/sponsored-testing/detect-lsds?tab=for-your-patients

Your doctor may choose to test for Fabry disease only (as above), or as part of a larger LSD panel.

PREVENTION GENETICS part of EX

Questions?
800-200-1524

Free Fabry Diagnostic Testing and Education Project Sponsor: AAKP

GLA gene analysis (Test code: 7681)(email: fabry.testing@emory.edu)
https://aakp.org/center-for-patient-research-and-education/fabry-disease/

revvity

Questions?
800-762-4000

The Lantern Project Sponsor: Sanofi

GLA gene analysis (Test code: SAN007)
https://www.revvity.com/category/the-lantern-project

Options for giving a sample may include:

1. A traditional blood draw
2. Dried blood spot (with a finger prick)
3. Saliva (swab or oral rinse)

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HOW ARE PEOPLE DIAGNOSED WITH FABRY DISEASE?

An individual can be suspected to have Fabry disease based on signs and symptoms, but a diagnosis cannot be confirmed without an enzyme or genetic test.

Enzyme = alpha-galactosidase A (test = alpha-galactosidase A enzyme analysis)

- testing is typically performed on a blood sample or a dried blood spot
- the test tells us the percentage of normal enzyme found in the blood
- in MALES abnormally low enzyme levels are able to confirm Fabry disease
- in FEMALES, low enzyme levels are often found but the levels can also be normal. A woman almost always needs genetic testing to confirm the diagnosis.

Gene = *GLA* (test = *GLA* gene analysis or, *GLA* gene sequencing)

- testing is performed on a blood or saliva sample
- genetic testing looks for any variations in the gene's spelling that are abnormal enough to be called a pathogenic variant (aka mutation)
- gene sequencing reads through the spelling of the Fabry disease gene (*GLA)*
- not all *GLA* changes are found by sequencing, some variants are large and require additional analysis for deletions and duplications.

Many individuals with Fabry disease will consult with a **genetic counselor** to help understand what their genetic testing results mean for themselves and their family members.

For those with a positive or uncertain result, consultation with a **Fabry specialist** is recommended. For a list of specialists please visit FabryDisease.org.