Testing Options
for Fabry Disease

Daisy and Viviana,
Fabry patients

SANOFI GENZYME

WHY GET TESTED FOR FABRY DISEASE?

Fabry disease is inherited. If one family member is diagnosed with the disease, others are likely to be affected as well. If you know Fabry disease runs in your family, here are some reasons to consider getting tested:

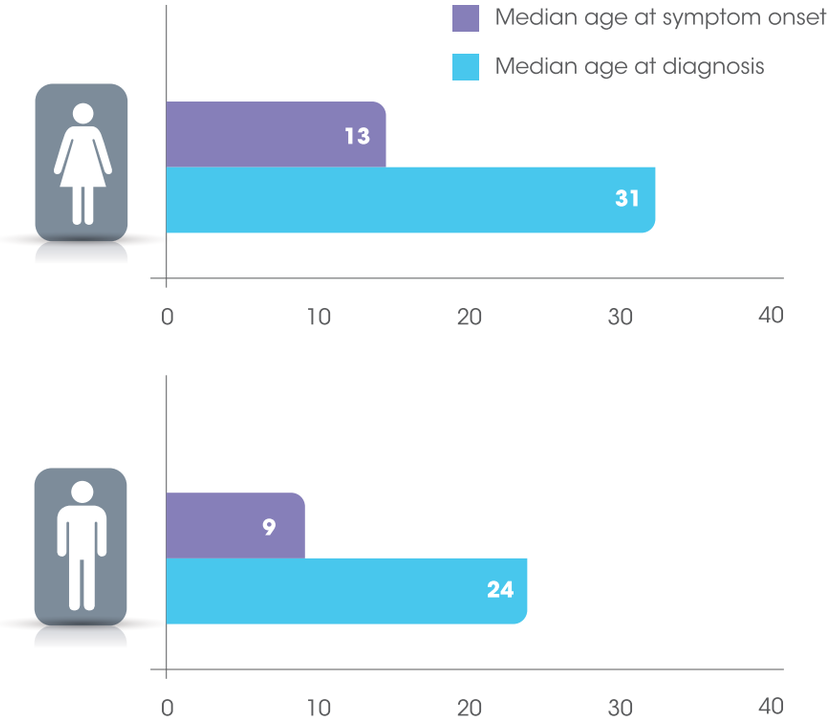
- Reduce the diagnostic delay, because Fabry disease is progressive, meaning it can get worse over time
- Eliminate uncertainty
- Help make sense of previously unexplained symptoms
- The earlier Fabry disease is diagnosed, the earlier disease management can begin

George, a Fabry patient

Understanding the Diagnostic Journey

Prior to a diagnosis of Fabry disease, individuals may experience many years of suffering and frustration while potentially receiving unnecessary medical treatments due to misdiagnoses. Diagnosis of Fabry disease may be delayed by many years from when symptoms first appear. Many people see a number of different specialists before they get an accurate diagnosis, including:

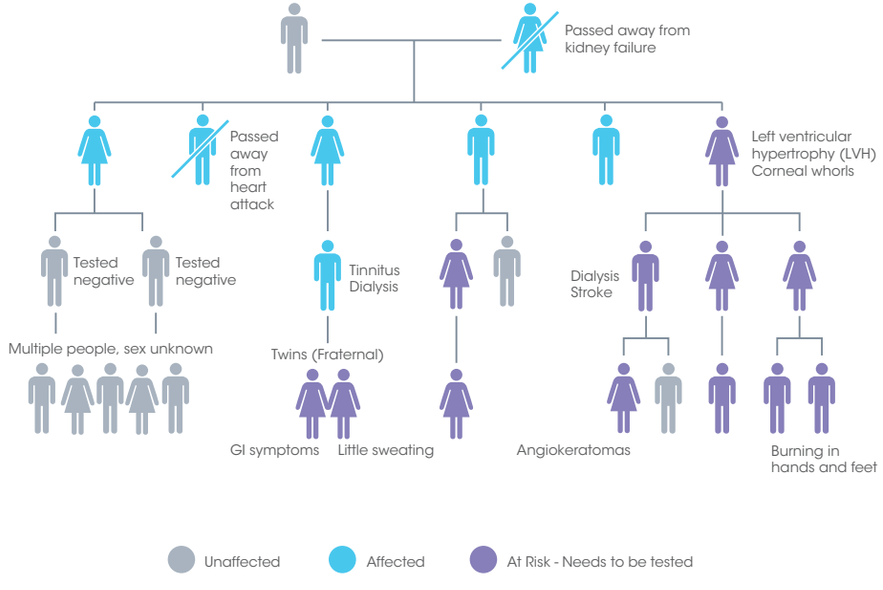
- Nephrologist for kidney problems
- Cardiologist for heart problems
- Neurologist for cerebrovascular problems, such as stroke
- Doctors for pain or gastrointestinal (GI) problems



The First Step: Creating a Medical Family Tree

When one member of a family is diagnosed with Fabry disease, a medical family tree can help identify others who may be at risk.

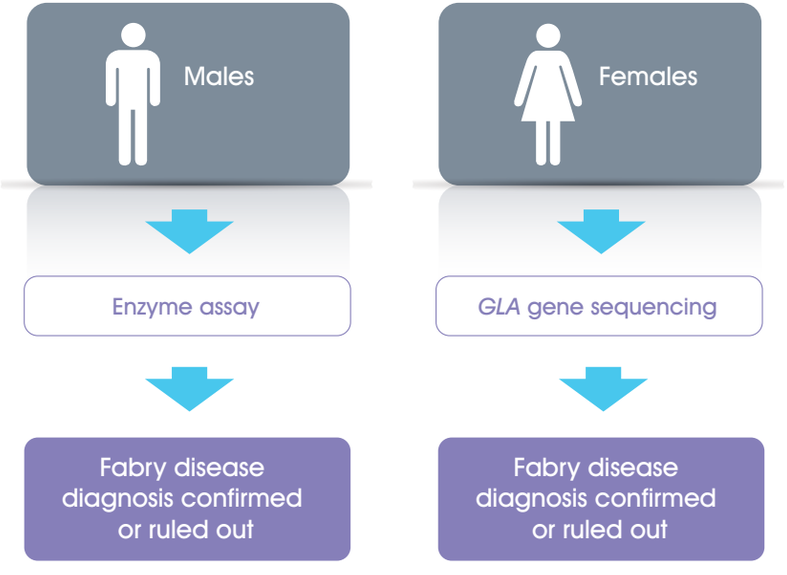
In this example, if the male on the lower right gets tested and learns that he has Fabry disease, it could help explain why his grandmother had left ventricular hypertrophy (enlarged left chamber of the heart). It is important to remember, though, that the absence of early signs and symptoms does not mean that a person is not affected. The disease can progress silently. Someone who inherits the altered gene is at risk for later complications, including heart disease, kidney disease and stroke.



A Sanofi Genzyme Patient Education Liaison can assist in creating a medical family tree.

About Testing

- Fabry disease can be confirmed using a blood or saliva sample
- Many genetic labs around the country are able to analyze blood or saliva samples to diagnose Fabry disease
- You can simply have your blood or saliva sample sent to the lab; some doctors' offices are able to help with the blood draw, or you may go to a special blood draw center
- If you choose to be tested, you could have results in two months or less
- A genetic counselor can help before, during or after testing



- Males with Fabry disease usually have a very low amount of an enzyme called alpha-galactosidase
- A test called an enzyme assay is usually enough to confirm or rule out a diagnosis; it can be done with a blood sample
- Females may have either low or near normal enzyme levels, so they need to have gene sequencing, which can be done using a blood or saliva sample
- Gene sequencing can determine the family mutation, which is useful for screening other family members; diagnosed males may choose to have gene sequencing done for this reason

Testing Options

Labs across the United States offer diagnostic tests for Fabry disease, and some provide the service free of charge. Below are some options available to you and your physician. This is not an all inclusive list. For additional testing centers that perform diagnostic testing, please visit https://www.ncbi.nlm.nih.gov/gtr/

Facility	Free Service*	Timing	Contact Information for Your Doctor
Duke University	No	Up to 6 weeks	919-549-0445
Fabry Diagnostic Project	Yes, except urine GL-3 test	Up to 6 weeks	800-200-1524 fabry.testing@emory.edu
GeneDx	No	Up to 5 weeks	888-729-1206 genedx.com
Greenwood Diagnostics Lab	No	2-3 weeks	800-473-9411
Integrated Genetics	No	Up to 2 weeks	800-848-4436 integratedgenetics.com
LabCorp	No	Up to 4 weeks	800-533-0567 labcorp.com
Mayo Medical Laboratories	No	Up to 4 weeks	800-533-1710
Sema4 (Mount Sinai)	Yes	Up to 8 weeks	212-659-6779

*In free testing programs, patients may be responsible for nominal costs such as the cost of the blood draw and overnight shipping to the lab.

This list of testing programs and/or laboratories is provided for informational use only. It should not be considered a recommendation or endorsement of any company, test or service for Fabry disease. Physicians are directed to contact the individual testing programs and/or laboratories for information on obtaining test requisitions, sample requirements, turnaround time and the specific details of the services provided. For commercial lab testing, appropriate coding and obtaining reimbursement is the responsibility of the provider submitting a claim for the item or service.

Stephanie and Sarah, Fabry patients

Sanofi Genzyme Offers Support Services for You and Your Family

For more than 30 years, Sanofi Genzyme has been committed to helping meet the needs of people who are living with rare disorders like Fabry disease. Our commitment extends to those who have Fabry disease and their families. We offer a wide spectrum of services, all personalized and tailored to you through your Case Manager.

Contact a Sanofi Genzyme Case Manager:

1-800-745-4447 (option 3)
Monday–Friday 8am–6pm EST

Connect with us online:

genzymesupportservices.com

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