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:

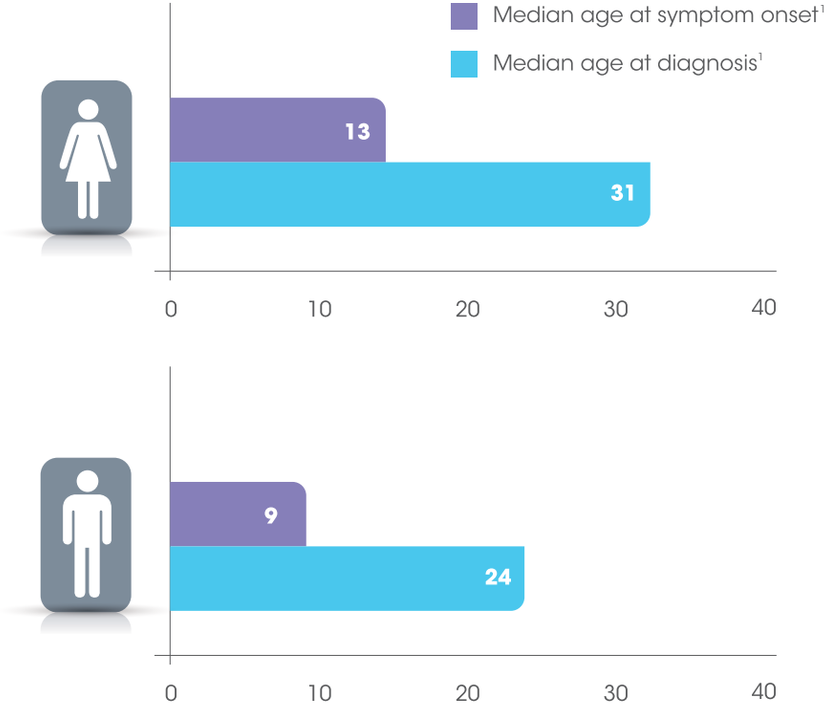
- Reduce the diagnostic delay. Fabry disease is progressive, which means it can get worse over time
- Eliminate uncertainty
- Help make sense of previously unexplained symptoms
- The earlier Fabry disease is diagnosed, the earlier disease management can begin

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:

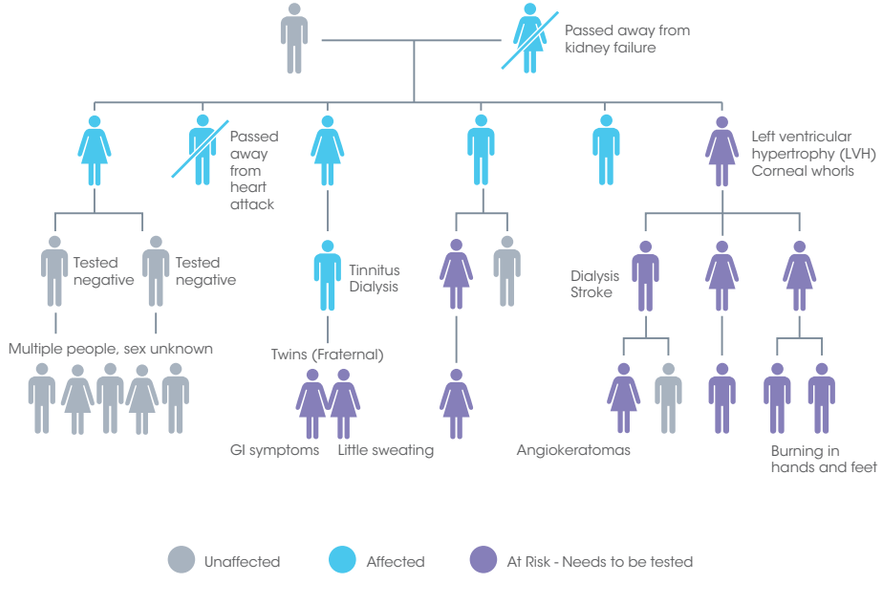
- Nephrologist for kidney problems
- Cardiologist for heart problems
- Neurologist for cerebrovascular problems, such as stroke
- Doctors for pain or gastrointestinal (GI) problems



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.

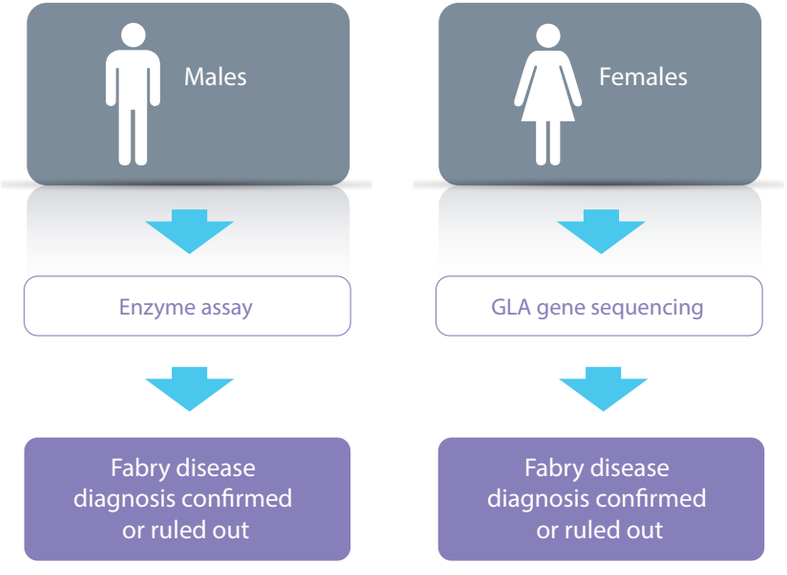
In this example, if the male on the lower right gets tested and learns that he has Fabry disease, it could help explain why his grandmother had left ventricular hypertrophy (enlarged left chamber of the heart). It is important to remember, though, that the absence of early signs and symptoms does not 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 CareConnectPSS Patient Education Liaison is available to 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

Some of the laboratories offering diagnostic testing for Fabry disease are listed below. There may be other diagnostic testing appropriate for you, and this is not an endorsement of any specific lab. Other testing options can be found at www.concertgenetics.com or 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.

Facility	Free Service*	Timing	Contact Information for Your Doctor
Centogene	No	Up to 5 weeks	617-580-2102 www.centogene.com
Duke University	No	Up to 3 weeks	919-613-8400 https://testcatalog.duke.edu
Fabry Diagnostic Project	Yes	Up to 4 weeks	404-778-8570 www.genetics.emory.edu
Greenwood Diagnostics Lab	No	Up to 6 weeks	800-473-9411 www.ggc.org
LabCorp/ Integrated Genetics	No	Up to 5 weeks	**LabCorp Customers:** 800-345-4363 www.labcorp.com **Integrated Genetics Customers:** 800-848-4436 www.integratedgenetics.com
The Lantern Project (performed at Perkin Elmer Genomics)	Yes	Up to 3 weeks	866-354-2910 www.LanternProjectDx.com
Mayo Clinic Laboratories	No	Up to 4 weeks	800-533-1710 www.mayocliniclabs.com
Mount Sinai International Center for Fabry Disease	Yes	Up to 3 weeks	866-322-7963 icahn.mssm.edu/research/fabry
Sema4	No	Up to 3 weeks	800-298-6470 www.sema4.com

*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.

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 CareConnectPSS Case Manager:

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

Connect with us online:

careconnectpss.com

1. Wilcox WR et al. Mol Genet Metab 2008;93:112-28.

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