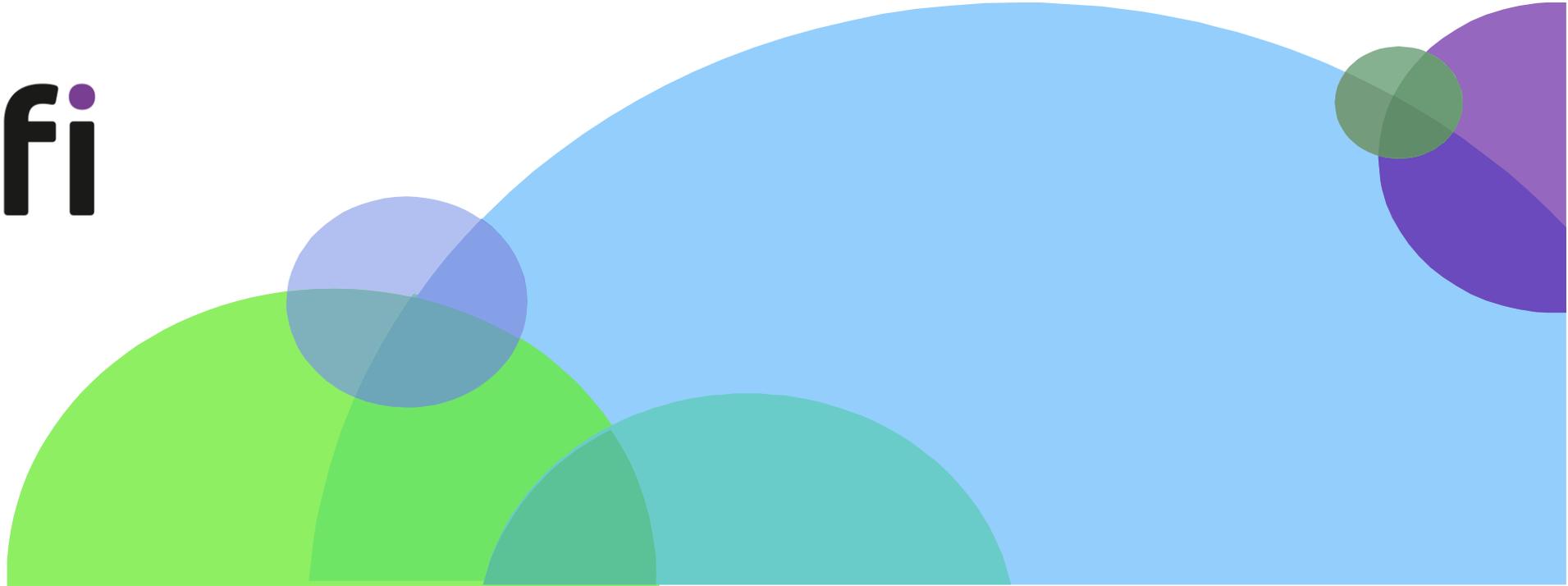


Testing Options for Fabry Disease

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A decorative graphic at the bottom of the slide consists of several overlapping semi-circles. From left to right, there is a bright green semi-circle, a light blue semi-circle overlapping it, a teal semi-circle overlapping the light blue one, a large light blue semi-circle overlapping the teal one, and a purple semi-circle overlapping the large light blue one. There are also smaller overlapping circles in shades of purple and green.

Sanofi does not provide medical advice, diagnosis, or treatment. The health information contained herein is provided for **general educational purposes only**. Your healthcare professional is the best source of information regarding your health. Please consult your healthcare professional if you have any questions about your health or treatment.

Intended for U.S. Residents Only

Reasons to Consider Testing for Fabry Disease

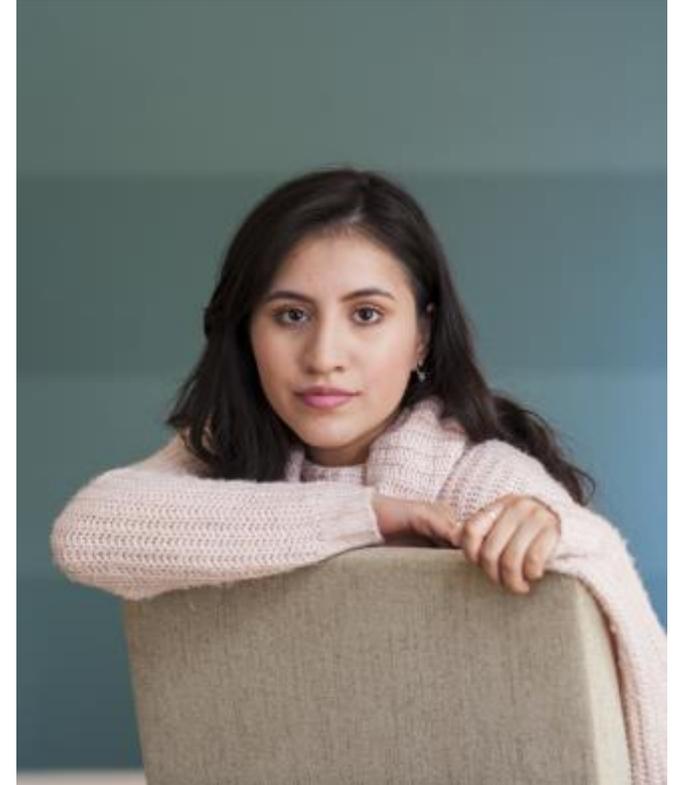
1. Your family member was diagnosed with Fabry disease
 - Fabry disease is inherited, so when one family member is diagnosed, others are likely to be affected¹
2. A medical family tree suggests you may be at risk for Fabry disease²
 - Diagnosis for some patients can be delayed for an average of 15 years after symptoms first appear; knowing there is Fabry disease in your family may help shorten the time to diagnosis^{1,3,4}



Avery, Fabry patient

Reasons to Consider Testing for Fabry Disease

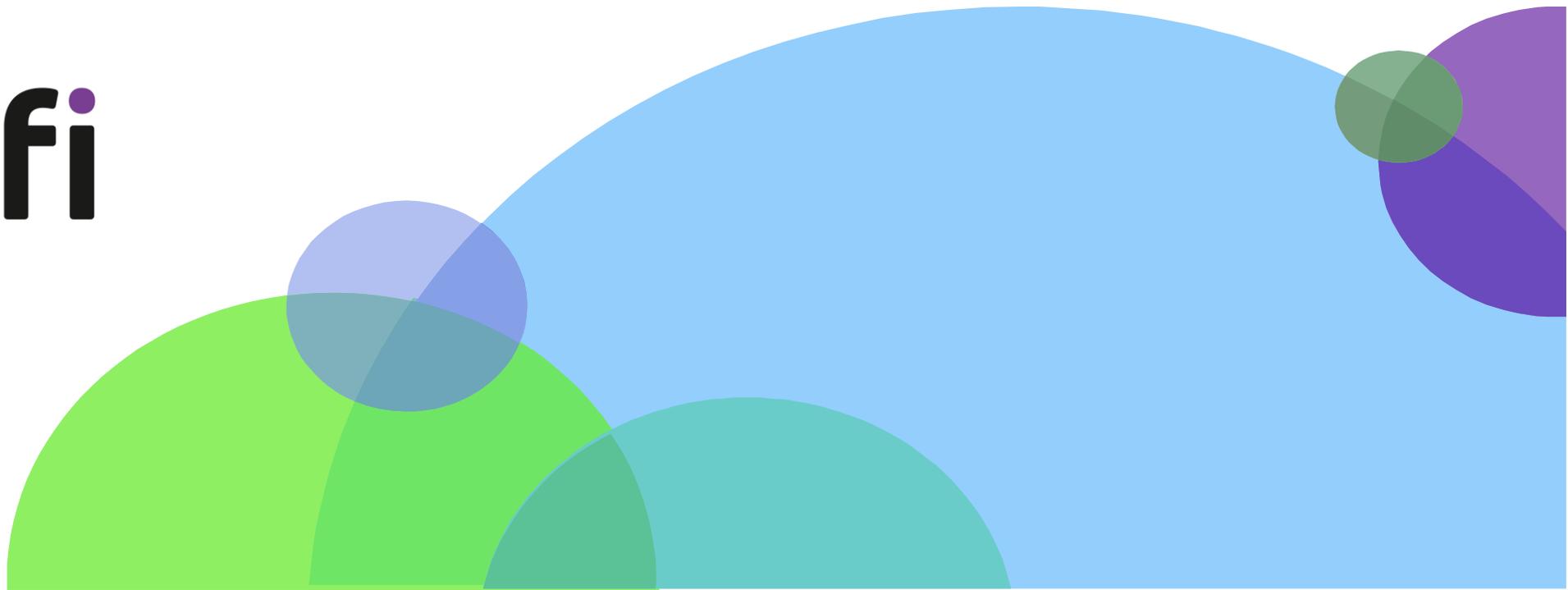
3. Confirm or rule out a diagnosis to help:
 - Decrease uncertainty and bring peace of mind
 - Make sense of previously unexplained symptoms
 - Advocate for the care you need and support your family members^{1,2}
4. The earlier that family members at risk for Fabry disease are diagnosed, the sooner they can explore options for disease management³
5. Diagnosis does not need to be invasive and can be completed with a single blood sample or in some cases a saliva sample⁴



Viviana, Fabry patient

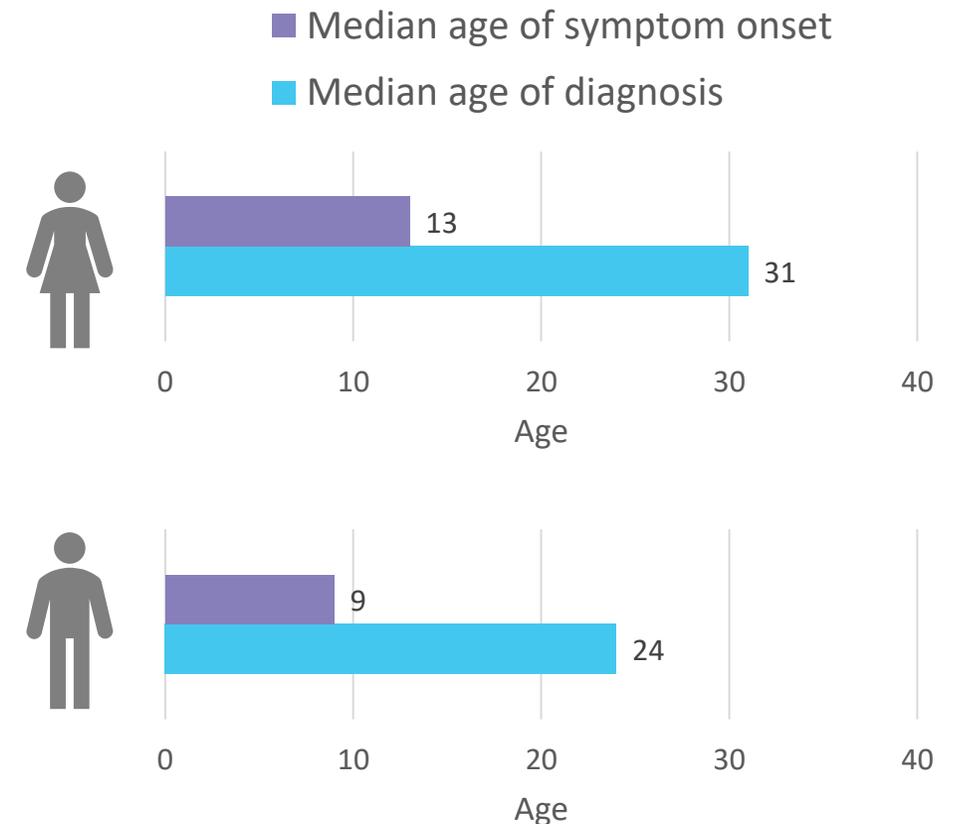
Fabry Diagnosis

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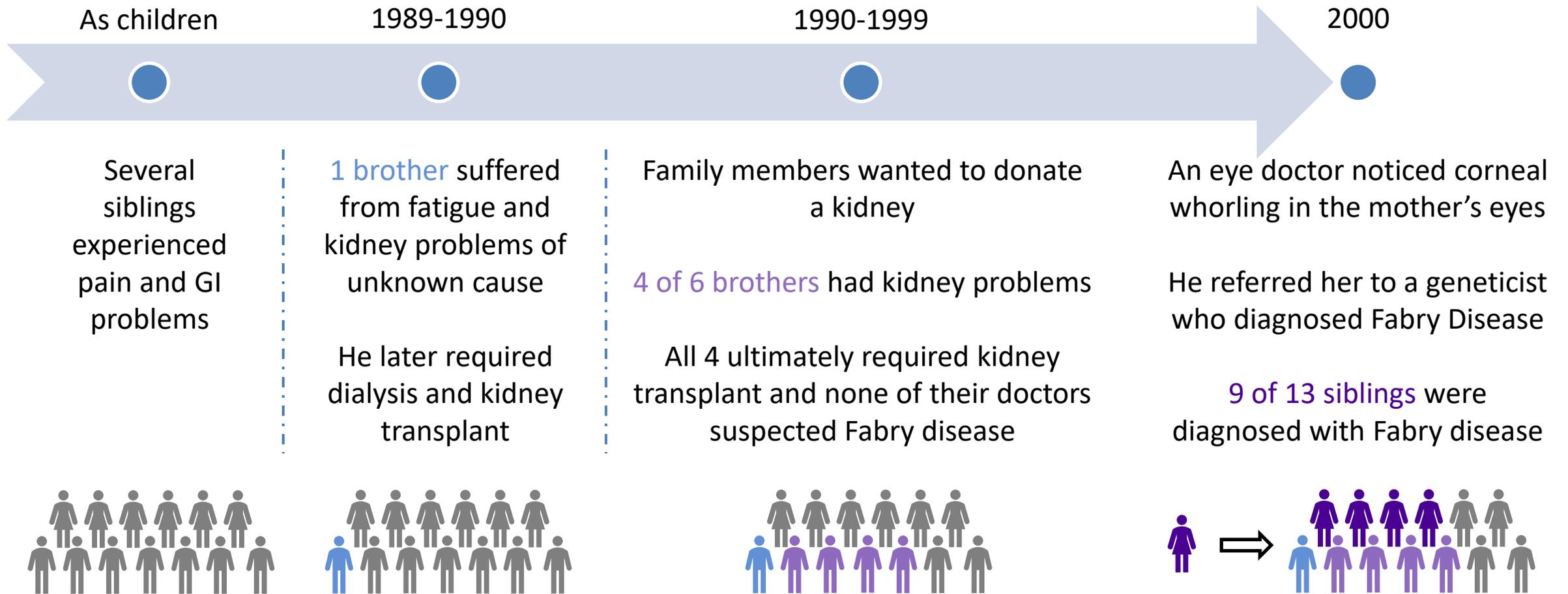


Understanding the Diagnostic Journey

- Prior to a diagnosis, individuals may experience many years of suffering and frustration while experiencing unexplained medical symptoms¹
- A diagnosis of Fabry disease takes an average of 15 years from when symptoms first appear²
- Many people see a number of different specialists before they get an accurate diagnosis³, including:
 - Nephrologist for kidney problems
 - Cardiologist for heart problems
 - Neurologist for stroke
 - Potentially several doctors for pain or GI problems



Path to Diagnosis: One Family's Story*



Confirming or Ruling Out a Diagnosis Can...

- Eliminate uncertainty which may provide additional options^{1,2}
 - Example: a child who is diagnosed may be eligible for an individualized education plan (IEP) or a 504 plan that lists accommodations that must be made at school to help manage symptoms like pain, GI problems or temperature sensitivity

*“My son used to get upset at school because gym class was so hard for him. After he was diagnosed, we were able to work with the school to create a plan that lets him participate at his own pace.” -
Quote from a Fabry patient*



Ed, Fabry patient

Confirming or Ruling Out a Diagnosis Can...

- Make sense of previously unexplained symptoms^{1,2}
 - Many people recognize the pattern of pain, GI problems and lack of sweating in childhood

“As soon as I read a description of Fabry disease, I thought, ‘I have this.’ The pain in the hands and feet, the stomach problems, I recognized all of it.” – Quote from a Fabry patient



Kathy, Fabry patient

Confirming or Ruling Out a Diagnosis Can...

- Empower you to advocate for the care you need^{1,2,3}
 - Planning for the future, monitoring symptoms, and talking to family members are all important actions you can take

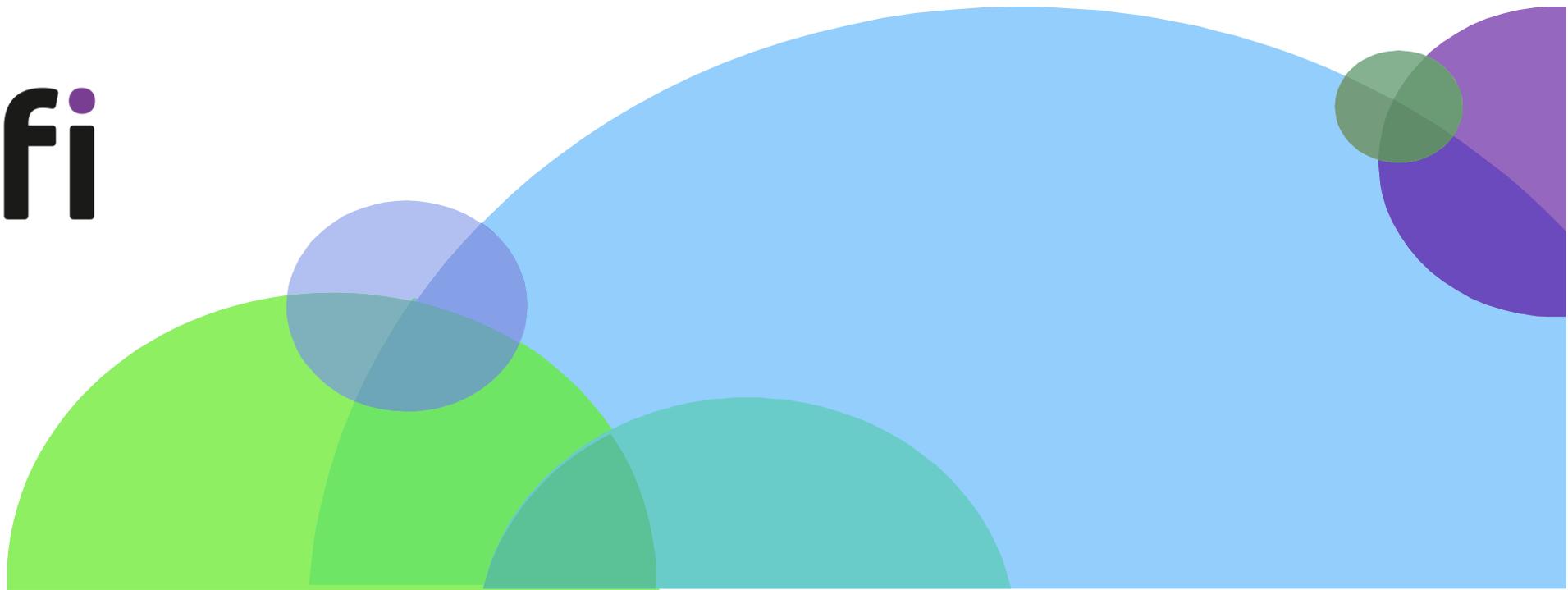
*“That first diagnosis really made a difference for our family. We all got together to discuss it, and now most of us have been tested.” -
Quote from a Fabry patient*



Daisy, Fabry patient

Fabry Testing

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About Testing

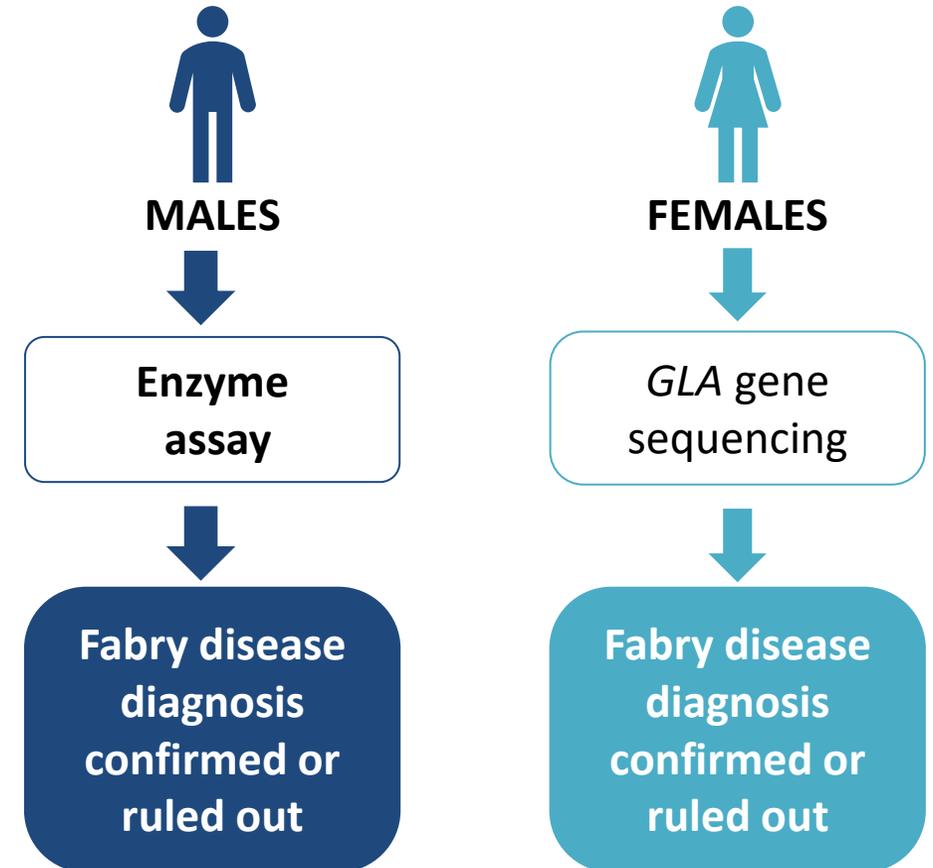
- Fabry disease can be confirmed through a blood or saliva test^{1,2}
- Many genetic labs around the country are able to analyze blood or saliva samples to diagnose Fabry disease
 - Talk to your doctor about ways to send your blood or saliva sample to labs
- Some doctors' offices are able to help with the blood draw while other times you may need to go to a special blood draw center; in both cases the sample is then sent to the lab to be analyzed
- Choosing to be tested may help avoid further diagnostic delays
- A genetic counselor can help before, during, and/or after testing³



Xanadu, Fabry patient

Testing for Fabry Disease^{1,2}

- Males with Fabry disease have a reduced level of an enzyme called alpha-Galactosidase A
- In males, 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 low or near normal enzyme levels, so gene sequencing is required to confirm the diagnosis. Gene sequencing can be done with a blood or saliva sample.
- Gene sequencing can determine the family gene variant, which is useful for screening other family members; diagnosed males may choose to have gene sequencing done for this reason



Labs That Offer Fabry Testing

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 www.icahn.mssm.edu/research/fabry
Sema4	No	Up to 3 weeks	800-298-6470 www.sema4.com

Some of the laboratories offering diagnostic testing for Fabry disease are listed above. There may be other diagnostic testing available to 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 does not review or control the content of non-Sanofi websites.

This listing does not constitute an endorsement by Sanofi of information provided by any other organizations.

*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.

Steps You Can Take After a Diagnosis

- If you are affected by Fabry disease:
 - Consider sharing a diagnosis of Fabry disease with friends or family if you are comfortable doing so
 - Find and establish care with specialists who are familiar with the multi-organ involvement of Fabry disease (for instance, the need for regular assessments for the heart, brain, kidneys, and ears)
 - Make plans for how to monitor your disease^{1,2}
 - Find support through national organizations, online groups, and meetings³



Luke, Fabry patient

Steps You Can Take After Testing

- If you are not affected by Fabry disease:
 - Recognize that no one has control over their genetics
 - Many people experience relief when they learn they are unaffected but at the same time feel bad for other family members who are affected. This is very typical.
 - Fabry disease is a family disease and even those without the gene variant can be impacted by family members who have Fabry disease
 - Unaffected family members can help advocate for their family members with Fabry disease¹



George, Fabry patient

Disease Management Options

- The earlier that family members at-risk for Fabry disease are diagnosed, the sooner they can explore symptom management options¹
- **Ask your doctor about what disease management options are right for you**

Symptom	Specialist	Examples of Ways to Manage
 Pain	Primary care, pain specialist	<ul style="list-style-type: none"> • Various over-the-counter or prescription pain medications • Avoid triggers such as cold or heat exposure, stress, or certain foods²
 Nausea Diarrhea Abdominal pain	Primary care, gastroenterologist	<ul style="list-style-type: none"> • Eat small meals • Avoid greasy or spicy foods • Over-the-counter or prescription medications to reduce nausea or aid digestion^{3,6}
 Kidney problems	Nephrologist	<ul style="list-style-type: none"> • Low protein, low sodium diet^{4,6} • Prescription medications such as angiotensin-converting enzyme (ACE) or angiotensin receptor blockers (ARBs)^{5,6}
 Heart problems	Cardiologist	<ul style="list-style-type: none"> • Heart-healthy diet • Manage blood pressure and cholesterol through certain medications⁶
 Cerebrovascular problems	Neurologist	<ul style="list-style-type: none"> • Blood thinners⁷

Summary^{1,2,3}

- Once someone in your family is diagnosed with Fabry disease, others could be identified as at risk for inheriting the condition
- Testing for Fabry disease can be done via blood, or in some cases saliva, and sent to labs across the country
- Confirming whether or not you are affected by Fabry disease gives you information to make appropriate health decisions for yourself and your family
- The earlier you confirm a diagnosis of Fabry disease the sooner you can start discussing medical recommendations with your doctor



Fabry Patients from L to R:
Viviana, Avery, Luke,
Daisy, Ed, and Xanadu