# TESTING FOR FABRY DISEASE

## may help you and your family

Fabry disease is a genetic disease with highly variable signs and symptoms. These signs and symptoms may be experienced from early childhood or starting later, in adulthood. That's why early genetic testing is so important. Sometimes Fabry disease can be undiagnosed or misdiagnosed. However, genetic testing can help identify the correct disease for the appropriate management. Anyone with a known family history of Fabry disease should encourage family members to speak with a doctor or genetic counselor.

Diagnosis in males can be made by testing for enzyme deficiencies, but further GLA gene testing must be conducted to confirm the diagnosis of Fabry disease and determine the specific genetic variant. However, females must have GLA gene testing for a diagnosis and to identify the specific genetic mutation.

Testing can be accomplished by a buccal (inside of cheek) mouth swab at home, by your healthcare provider, or lab; or by a blood test at a lab or genetic testing center.

**The reasons for testing and diagnosis include:**

- Identifying family members who may be affected
- Enabling earlier testing for family members
- Providing help to best manage the disease

Once a diagnosis is made, genetic counseling is encouraged.

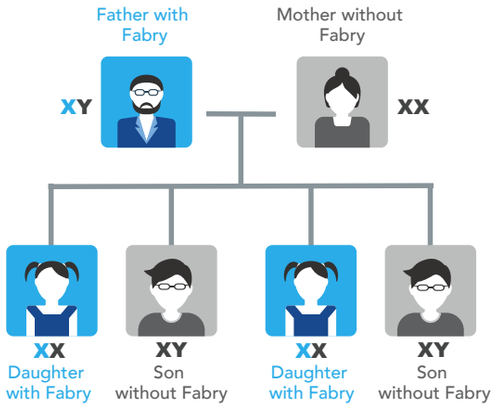
## How Fabry disease gets passed from parent to child

- A female with Fabry disease may have inherited it from either parent
- A male with Fabry disease could have inherited it only from his mother
- A father with Fabry disease will not pass it to his sons, but all of his daughters will be affected
- A mother with Fabry disease has a 50% chance of passing it to her children

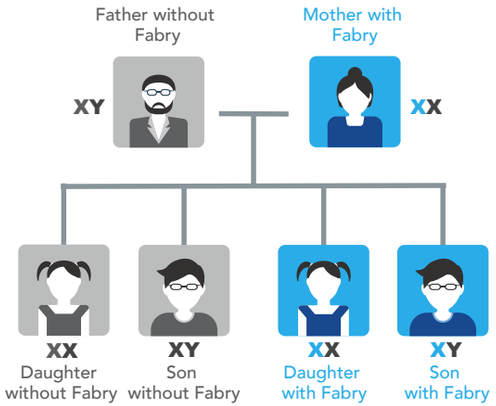
## Hereditary

Two factors determine inheritance of Fabry disease—which parent contributes the mutated X chromosome and whether the child is a boy or a girl.

**An affected father (XY) will pass the mutation to all of his daughters, but none of his sons.**




**An affected mother (XX) has a 50% chance of passing the mutation to any of her children.**




## Testing Locations

Below is a list of Fabry genetic testing locations. Please discuss with your healthcare provider what the best option is for you.

**Duke University Health System**
Kit available – Yes
Free service – No
Contact – 919-684-3604
https://www.dukehealth.org/treatments/genetic-disorders/lysosomal-disorders

**Fabry Diagnostic Testing & Education Project sponsored by the American Association of Kidney Patients (AAKP)**
Kit available – Yes
Free service – Yes
Contact – 404-778-8518 or 800-200-1524
http://genetics.emory.edu/patient-care/lysosomal-storage-disease-center/lab-testing.html

**GeneDx**
Kit available – Yes
Free service – No
Contact – 888-729-1206 or 301-519-2100
https://www.genedx.com/test-catalog/disorders/fabry-disease/

**Integrated Genetics**
Kit available – No
Free service – No
Contact – 800-848-4436
https://www.integratedgenetics.com

**LabCorp**
Kit available – Yes
Free service – No
Contact – 888-522-2677
https://www.labcorp.com/test-menu/search

**Mayo Clinic Laboratories**
Kit available – Yes
Free service – No
Contact – 480-301-8000
https://www.mayocliniclabs.com

**Icahn School of Medicine at Mount Sinai**
Kit available – No
Free service – Yes
Contact – 866-322-7963
https://icahn.mssm.edu/research/fabry

**Quest Diagnostics**
Kit available - Yes
Free Service – No
Contact – 866-697-8378
https://questdiagnostics.com

**The Lantern Project from Sanofi Genzyme and PerkinElmer**
Must request kit via mail order
Kit available - Yes
Free service - Yes
Contact – 866-354-2910
https://www.perkinelmergenomics.com/fabry-disease-lantern/

Please note: Some programs are free and some may have a cost. Genetic testing may or may not be covered by your insurance. Contact your insurer for coverage details. A doctor's note may be required; please contact your healthcare provider for testing authorization.

This is not an exhaustive list of testing centers, and Amicus does not recommend any particular center. For a list of additional centers, please consult with your healthcare provider.

## Medicare and Medicaid Coverage

If you have Medicare or Medicaid you may be covered for testing.

- Medicaid coverage differs from state to state and may require prior approval before testing, depending on where you live
- Medicare does not offer prior authorization, so coverage should be determined prior to testing

YOU MAY BE ELIGIBLE FOR

# FINANCIAL HELP

Assistance funds may be available for people living with Fabry disease with unaffordable medical expenses (i.e., co-payment, premium, genetic testing, ancillary services, infusion, nursing services, travel, and/or concomitant medications). These funds may help reduce stress and facilitate access to important treatments.

## Patient Services Incorporated (PSI)

The nonprofit PSI provides premium, copayment, ancillary, travel, and infusion/nursing assistance to the eligible members of the Fabry community.
Contact – 800-366-7741 or 804-521-7906
https://www.patientservicesinc.org

## The Assistance Fund (TAF)

The Assistance Fund breaks down financial barriers to proper treatment.
Contact – 855-253-9223 or 855-845-3663
https://tafcares.org

## Patient Access Network (PAN) Foundation

The PAN Foundation helps people with life-threatening, chronic and rare diseases with the out-of-pocket costs for their prescribed medications.
Contact – 866-316-7263
www.panfoundation.org

Please contact individual assistance funds to obtain specific Fabry information, since they may not be accepting new Fabry patients and funds are not always available.

Please feel free to contact us at:
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