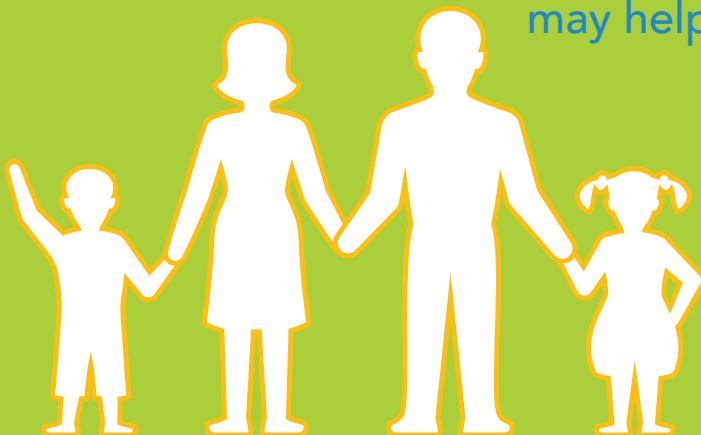




TESTING FOR FABRY DISEASE

may help you and your family



Fabry disease is a rare 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 galactosidase alpha (*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 variant.

Testing can be accomplished by a buccal (inside of cheek) mouth swab at home, by your health-care 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.



Amicus Therapeutics has developed this educational resource in collaboration with the rare disease community and thought leaders.

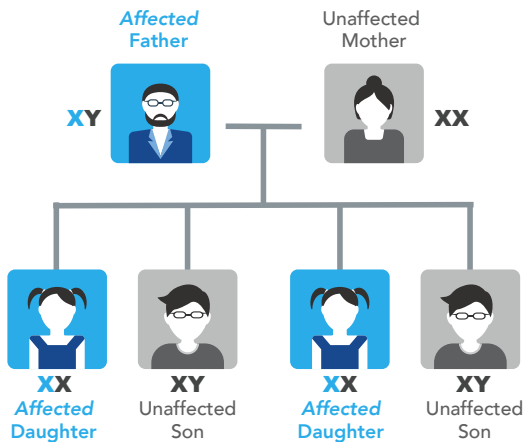
How does Fabry disease affect families?

- 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

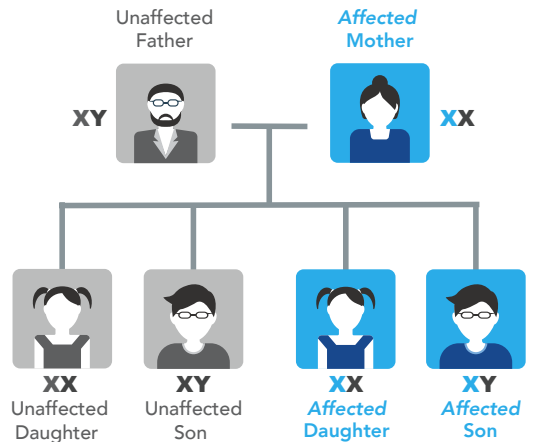
Hereditary

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

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



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



Testing Options

There are many options available for genetic testing and your health-care provider can help determine which may be the best for you.

Duke University Health System

Kit available – Yes
Free service – No
Contact – 855-855-6484
<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
<https://med.emory.edu/departments/human-genetics/patient-care/lysosomal-peroxisomal/index.html>

GeneDx

Kit available – Yes
Free service – No
Contact – 888-729-1206 or 301-519-2100
<https://www.genedx.com/test-catalog/available-tests/lysosomal-disorder-panel/>

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/test-catalog/Fees+and+Codes/35415>

Icahn School of Medicine at Mount Sinai

Kit available – No
Free service – Yes
Contact – 866-322-7963
<https://icahn.mssm.edu/research/fabry>

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/index.html>

Invitae

Kit available – Yes
Free service – Yes
Contact - 800-436-3037
<https://www.invitae.com/en/detectLSDs/>

Greenwood Genetic Center

Kit available – Yes
Free service – No
Contact – 888-442-4363
<https://www.ggc.org/test-finder-item/fabry-disease-gla-sequencing?A=SearchResult&SearchID=12430711&ObjectID=5273447&ObjectType=35>

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 health-care 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 health-care provider.

Medicare and Medicaid Coverage

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

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

YOU MAY BE ELIGIBLE FOR FINANCIAL HELP



Assistance funds may be available for people living with Fabry disease with unaffordable medical expenses (ie, premium, copayment, 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 services, travel, infusion/nursing assistance and travel to 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 helps to break 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 out-of-pocket costs for their prescribed medications.

Contact – 866-316-7263
www.panfoundation.org

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



Please discuss any medical questions with a health-care professional (HCP).

If you would like to provide feedback on this educational resource or would like additional information, please contact:
patientadvocacy@amicusrx.com.

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