

FABRY DISEASE

INFORMATION FOR PATIENTS AND FAMILIES

With the tremendous advances in our understanding of genetic diseases and the promise of advanced therapies, the need to help Fabry patients and families understand their family history has never been greater.

Fabry Support & Information Group is working to increase awareness of the importance for Fabry patients and their families of knowing their family tree.

This handout reviews key information about how Fabry disease may affect you or a loved one, and provides information about resources that can help you complete your own family tree.

"The Surgeon General has launched an initiative to encourage all American families to learn more about their family health histories..."

With a copy of your family health history, you and a health care professional can individualize your care to prevent and screen for conditions for which you may be at higher risk."

— US Surgeon General's Family Health Initiative - My Family Health Portrait

About Fabry Disease

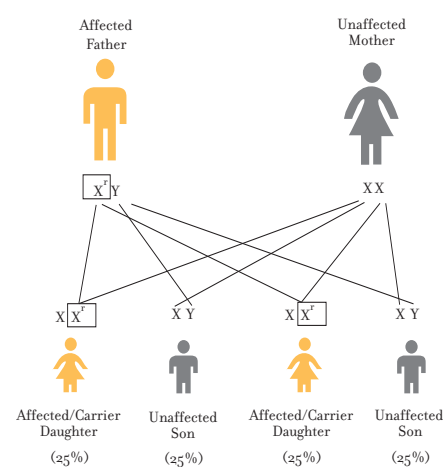
Fabry disease is a rare, inherited disorder caused by a faulty gene. When people inherit this faulty gene, their bodies cannot produce enough of an important enzyme called alpha-galactosidase A, or alpha-GAL.

Alpha-GAL is needed to break down a fatty substance called globotriaosylceramide, or GL-3. Without alpha-GAL, GL-3 cannot be broken down. Instead, it accumulates in the cells. The most commonly affected cells are in blood vessels and tissues of the kidney, heart, skin, and brain. The buildup of GL-3 in these cells can eventually lead to life-threatening problems.

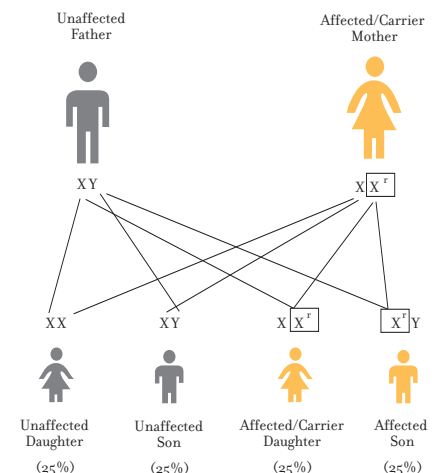
How Fabry Disease is Inherited

The gene that causes Fabry disease is located on the X chromosome. Females have two X chromosomes. Males have one X and one Y. Females inherit an X from each parent, while males inherit an X from their mothers and a Y from their fathers.

If a male inherits an X chromosome with a faulty alpha-GAL gene, he will produce little or no alpha-GAL, and will develop symptoms of Fabry disease. If a female inherits the Fabry gene, she may experience no Fabry related symptoms or may have mild to sometimes severe symptoms.



Fabry Disease Inheritance Pattern
Affected Father



Fabry Disease Inheritance Pattern
Affected Mother (Carrier)

Resources

To learn more about Fabry disease, below is a list of resources. These resources also contain information about how to fill out your family tree.

Fabry Support & Information Group (FSIG)
www.fabry.org

Genzyme's Medical Information
800-745-4447 (option 2)

National Society of Genetic Counselors
www.nsgc.org

Fabry Community (a Genzyme web site)
www.fabrycommunity.com

These listings are provided as additional information for patients with Fabry disease. The web pages and their content are maintained by the organizations listed above. With the exception of their own respective web sites, FSIG and Genzyme do not endorse any particular organization or the content contained on those web sites.

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Diagnosing Fabry Disease

Because Fabry disease is rare and causes a wide variety of symptoms, its diagnosis can be difficult and confused with that of other diseases. People may have the disease for a long time before it is accurately diagnosed. This is a concern because the longer a person has Fabry disease, the more damage is likely to occur. The earlier Fabry disease is diagnosed, the earlier doctors can begin to manage the disease and try to prevent further health problems.

The Importance of Family Trees

One way to increase the likelihood of early diagnosis is to understand who is at risk for developing Fabry disease. When one person in a family has the disease, others may also be at risk.

A family medical tree is a record of health and illness within a family. It resembles the family tree you might draw to record your genealogy, except that it also includes medical information. It can help you understand how Fabry disease has affected relatives, both living and deceased, as well as how the gene may affect future generations.

What You Can Do

To help families outline their medical histories, the U.S. Surgeon General has initiated the Family Health Portrait (available at www.hhs.gov/familyhistory/). In this Family Health Portrait, you can fill out health information about yourself, your immediate and extended family, and family members from past generations. You may want to seek the assistance of a health care provider or a medical genetic counselor to complete this historical account.

