Facts About Fabry Disease

What is Fabry disease?

Fabry disease is an inherited disorder caused by an altered gene. A person who inherits this gene is unable to produce an enzyme called alpha-galactosidase A, or alpha-GAL, in the amounts the body needs. Alpha-GAL is supposed to break down a fatty acid called globotriaosylceramide, or GL-3. Since a person with Fabry disease does not have enough alpha-GAL, the GL-3 is not broken down, but instead builds up in the cells.

What are the major symptoms of Fabry disease?

The build-up of GL-3 in the cells causes symptoms that change over time. Early symptoms are often mistaken for other, more common conditions. This is one of the reasons Fabry disease is hard to diagnose. Males with the disease will often have most of the symptoms listed here. Females can have symptoms that range from mild to severe, but they may get severe symptoms later in life, even if they didn't have the earlier, milder ones.

Signs & Symptoms

	Childhood	Adolescence	Adulthood
Episodic pain crises	●	●	●
Neuropathic pain	●	●	●
Hypohidrosis/anhidrosis (inability to sweat)	●	●	●
Corneal opacities (distinct pattern)	●	●	●
Recurrent fever	●	●	●
Heat and cold intolerance	●	●	●
Psychosocial manifestations	●	●	●
Gastrointestinal distress	●	●	●
Proteinuria		●	●
Angiokeratomas (red skin spots)		●	●
Fatigue		●	●
Renal insufficiency (kidney problems)			●
Neurological complications			●
Cerebrovascular disease (TIA or stroke)			●
Cardiac dysfunction (heart disease)			●
Hearing loss and tinnitus			●

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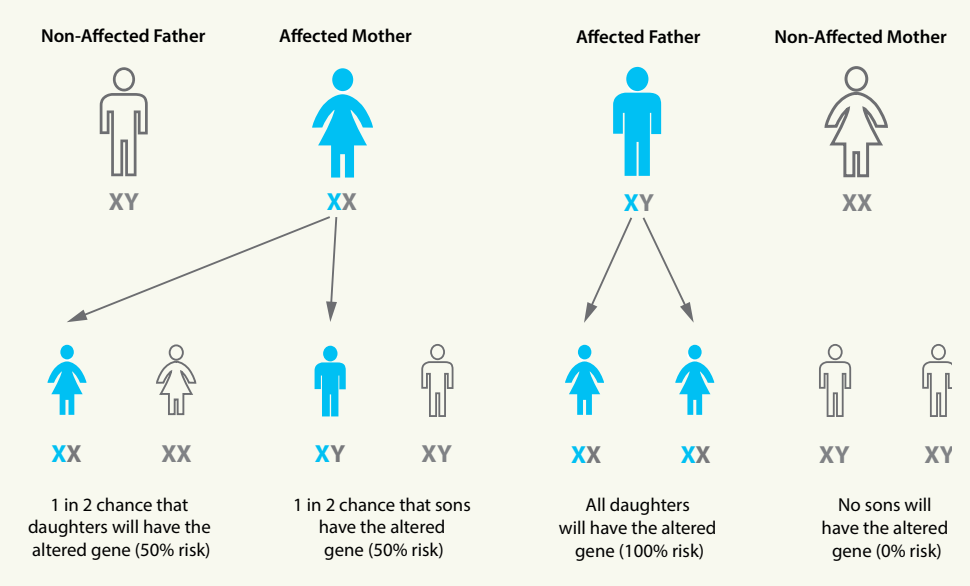
How is Fabry disease inherited?

The gene that causes Fabry disease is located on the X chromosome. Men have one X and one Y chromosome, while women have two X chromosomes. This means that men pass the gene to all of their daughters and not to their sons, while women have a 50% chance of passing it on with each pregnancy.



How is Fabry disease diagnosed?

Fabry disease can be diagnosed with a blood test. For males, a lab will perform a test called an enzyme assay on the blood sample. For women, they perform a DNA test on a blood sample. Your doctor can help you arrange to get tested.

What happens if I am diagnosed with Fabry disease?

If you are diagnosed with Fabry disease, your doctor may refer you to a geneticist. A geneticist has expertise in genetic disorders like Fabry disease. He or she can help you better understand the disease and determine who else in your family may have inherited the altered gene. Because Fabry disease can affect different organ systems such as your heart, kidneys, brain and nervous system, you may see a team of specialists. Your geneticist or Sanofi Genzyme Patient Education Liaison can help you understand who else in your family may be at risk and give you suggestions for reaching out to them.

Where else can I get information?

There are a number of organizations that can provide information and support to people living with Fabry disease. They include:

Sanofi Genzyme Fabry Community
fabrycommunity.com

Sanofi Genzyme Patient & Caregiver Portal
genzymesupportservices.com

FSIG – Fabry Support & Information Group*
fabry.org

National Fabry Disease Foundation*
fabrydisease.org

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www.genzymesupportservices.com

*Sanofi Genzyme does not review or control the content of non-Sanofi Genzyme websites. These listings do not constitute an endorsement by Sanofi Genzyme of information provided by any other organizations.

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