A VISUAL GUIDE TO UNDERSTANDING FABRY DISEASE

INFORMATION FOR FAMILIES LIVING WITH FABRY DISEASE
How does Fabry disease affect families?[^7]

Fabry disease is an X-linked disorder. It is caused by a disease-causing variant of a gene (called the GLA gene) that is located on the X chromosome. This gene variant can be passed down by either parent. In rare cases, Fabry disease can be caused by a gene variant that occurs spontaneously in a child and is not present in either parent (called a de novo variant or de novo mutation).

**How Fathers Pass Down Fabry Disease**

Men have one X chromosome and one Y chromosome. A father with Fabry disease passes his disease-causing variant of the GLA gene to all of his daughters, because daughters inherit their father’s only X chromosome. An affected father cannot pass the gene variant to his sons, because sons inherit a Y chromosome from their fathers.

**How Mothers Pass Down Fabry Disease**

Women have two X chromosomes. A mother who has a disease-causing variant of the GLA gene on one of her two X chromosomes has a 50% chance of passing down Fabry disease to each of her children. Since males have only one X chromosome, if they inherit the disease-causing gene variant, they will develop Fabry disease. Because females have two X chromosomes, a daughter who inherits one copy of the disease-causing gene variant may have a normal copy of the GLA gene on her other X chromosome. This may allow her body to produce some normal a-Gal A. Affected daughters may have more variable symptoms than affected sons for this reason.[^7]

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**What is Fabry disease?**

Fabry disease is a rare genetic disorder. People who have lysosomal disorders may have problems making specific lysosomal enzymes or other proteins.

**What causes Fabry disease?**

DNA is inherited through genes that are passed down from the person’s mother and father. Sometimes, gene variants (also called mutations) occur in the DNA code of a particular gene that can change the way the gene functions. Everyone has information called DNA coded into his or her cells.

**How does Fabry disease affect the body?**

The signs and symptoms of Fabry disease tend to gradually worsen over time. However, it’s important to be aware that everyone experiences Fabry disease differently. Some signs and symptoms may be more or less severe in some people compared with others. Also, not everyone will experience all of the signs and symptoms listed below.

**How is Fabry disease treated?**

People who are diagnosed with Fabry disease may be referred to a team of specialized health-care providers, such as nephrologists, cardiologists, genetic counselors, and other specialists.

Further testing may be done, including blood and urine tests, imaging tests such as computed tomography or CT, scans, and evaluations of sight and hearing.

A personalized treatment plan is created, which may include:

- Treatments that help manage the specific signs, symptoms, and complications the person is experiencing
- Enzyme replacement therapy (ERT)
- For people who have certain gene variants, a type of treatment called chaperone therapy

Research is currently being done on potential new treatments for Fabry disease. To find out more about ongoing research trials, search for Fabry disease on clinicaltrials.gov, or talk to your health-care provider.

**Other resources that may be helpful are listed below.**

- Fabry International Network
- Fabry Support & Information Group
- The National Fabry Disease Foundation
- Society for Mucopoly saccharide Diseases (UK)
- Global Genes
Why are gene variants important?

More than 1000 different gene variants have been identified that can cause Fabry disease.10

The specific gene variant a person has may help predict when symptoms appear, what kind of symptoms appear, and how bad the symptoms may become.5,7

It’s important for individuals and families with Fabry to know which gene variant they have.

References