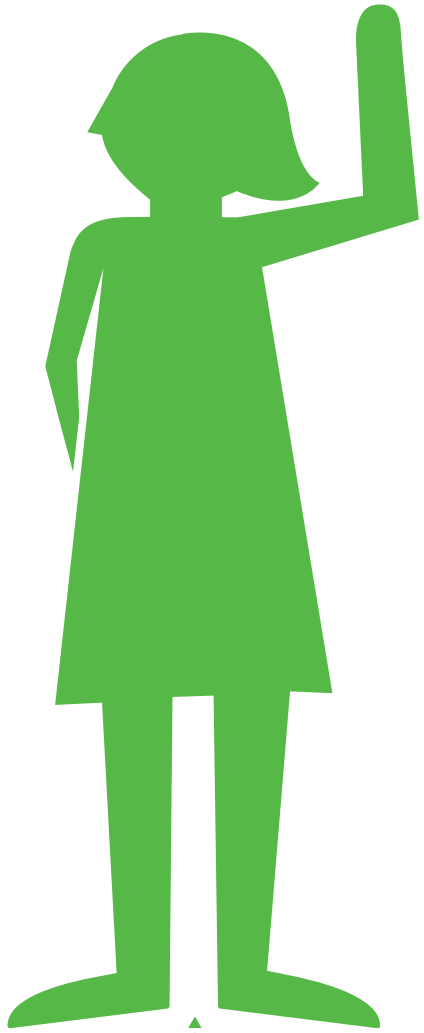
WOMEN AND FABRY DISEASE

An evolving understanding

Fabry disease is caused by certain changes in the DNA that are called "variants." It is an X-linked genetic (inherited) disorder, which means that the variants cause it to happen in a gene that's located on the X chromosome. Fabry disease can be passed down by either parent.[1]

Thousands of women around the world have gene variants that are known to cause Fabry disease. Women who have these variants were once considered by medical professionals to be asymptomatic carriers of Fabry disease—meaning that while they could pass the condition to their children, they experienced no (or few, and very mild) signs and symptoms of the disease themselves.[1]

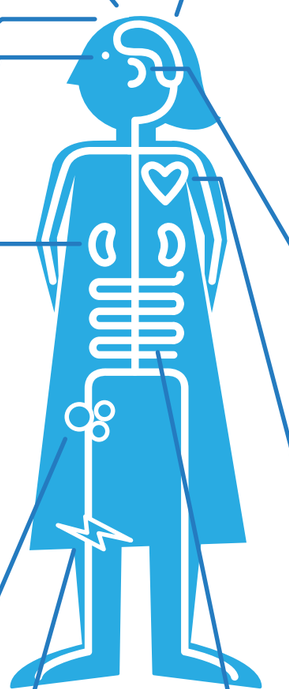


Because of this misconception, health-care providers (HCPs) have sometimes dismissed concerns women have expressed about the effects of Fabry disease on their health. Some HCPs also have not taken any signs and symptoms they report seriously. Understandably, this has led to frustration among many women who live with the disease.[2-4]

However, medical research has now confirmed that most women who have gene variants that cause Fabry disease *do* experience signs and symptoms of the disorder. These signs and symptoms vary from woman to woman, and they can range from mild to very severe.[2,5-9] Ongoing research is continuing to improve awareness and understanding among HCPs of how Fabry disease can affect women.[5]

This brochure provides information about researchers' current understanding of how Fabry disease can affect health and quality of life in women. It's designed to offer support to women who are living with the disease, and to encourage and empower them to learn more about their condition.

Women with Fabry disease may have a wide range of signs and symptoms



DIZZINESS (VERTIGO)[7]
- Experienced by approximately 25% to 85%
- Average age of onset: 30s

FATIGUE[9]
- Experienced by approximately 59%

CORNEAL WHORLING AND/OR FABRY CATARACTS[7]
- Experienced by approximately 36%
- Average age of onset: 30s

EFFECTS ON THE KIDNEYS[7]
- Experienced by approximately 21% to 55%
- Average age of onset: 30s

SPOTS ON THE SKIN (ANGIOKERATOMAS)[7]
- Experienced by approximately 13% to 55%
- Average age of onset: 30s

PAIN[1,7,10]
- Experienced by 67%, most commonly in the hands and feet
- Average age of onset: Teens

STROKE[7]
- Experienced by approximately 6%
- Average age of onset: 50s

TRANSIENT ISCHEMIC ATTACK (TIA)[7]
- Experienced by approximately 7%
- Average age of onset: 40s

RINGING IN THE EARS (TINNITUS)[7]
- Experienced by approximately 27%
- Average age of onset: 30s

HEARING LOSS[9]
- Experienced by approximately 37%

EFFECTS ON THE HEART[7]
- Experienced by approximately 5% to 65%
- Average age of onset: 30s

GASTROINTESTINAL (GI) ISSUES[7]
- Experienced by approximately 11% to 60%
- Average age of onset: 20s

Women with Fabry disease may experience all, a few, or none of these signs and symptoms. These signs and symptoms may also vary in severity among those who do have them.

For many women, Fabry disease also reduces quality of life

In a study that evaluated health-related quality of life in 44 women with Fabry disease compared with the general female population, researchers found that women who had Fabry disease had poorer impressions of many aspects of their physical, mental, and social functioning, especially their vitality, physical functioning, and overall health. The researchers also found that almost two-thirds of the women with Fabry disease reported symptoms of depression and more than one-third reported anxiety.[9]

In a study that included 202 women with Fabry disease, researchers found that self-reported quality of life in women who had Fabry disease was not only significantly worse than in healthy women, but that it was similar to—or sometimes even worse than—quality of life among women who had multiple sclerosis or rheumatoid arthritis (both chronic diseases that are known to have a major impact on people's lives).[8]

In a study of the impact of pain on the lives of 367 people with Fabry disease, researchers found that pain intensity and frequency were similar in women and men; they also found that for both women and men, increasing levels of pain were associated with lower quality of life in every aspect of life that the study evaluated (including mood, relationships, work, sleep, and life enjoyment).[10]

People with Fabry disease also may experience reduced self-esteem and feelings of anger, grief, and hopelessness related to their condition, as well as feelings of guilt or blame toward family members because of the inherited nature of the disease.[11]

Every woman's experience is different—and important!

Exactly why there's so much variation in signs and symptoms among women who have Fabry disease is not yet known. However, some scientists believe that among other possible factors, it may have something to do with a process called X inactivation.[1,4]

During fetal development, one of the two X chromosomes in each cell of a woman's body is inactivated, or shut off. This process occurs randomly—either the normal X chromosome or the X chromosome that has the variant that causes Fabry disease can be inactivated in any given cell.

Which specific cells—and also how many cells overall—have the normal X chromosome inactivated vs the X chromosome that has the variant may have an impact on which of a woman's organs are affected, as well as how severely they are affected. This, in turn, can influence the woman's experience of Fabry disease.[9,12]

Regardless of the signs and symptoms she may be experiencing, every woman who has Fabry disease should speak with her health-care provider about monitoring the effects of the disease regularly. Fabry disease can cause serious complications over time in both men and women.[9,12]

Women who live with Fabry disease face unique challenges

Fabry disease often manifests differently in women than in men, and its signs and symptoms also tend to vary significantly among different women who have the condition. Also, some HCPs are still not fully aware of how serious the effects of Fabry disease can be in women. All this can contribute to delays in diagnosis: although women with Fabry disease may begin to notice its effects while still in their teens, many are not diagnosed for more than 15 years after they first experience symptoms.[5,9]

Additionally, because women with Fabry disease may have multiple roles and responsibilities, living with the disease may be especially overwhelming. For example, some women may manage their household and work outside the home, while also serving as caregivers for others with Fabry disease, including children, siblings, and/or parents.

For many women, the combined physical, emotional, and social effects of Fabry disease can add up to a heavy burden. With all the difficulties and demands they may face, some may find it difficult to devote time and energy to recognizing and addressing their own needs. But as our understanding of how the disease affects women has evolved, the tremendous importance of attention and care for the health and well-being of women with Fabry disease has become clear. By advocating for themselves and prioritizing their own care, women with Fabry disease may be able to live better lives—and also become better able to support those who depend on them.

It can be helpful—and empowering—to learn more about Fabry disease and connect with other women who are affected by it. To find out more about Fabry disease and the Fabry community, talk to an HCP, and consider visiting www.fabrynetwork.org/members/ to find a local member organizations.

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Global Patient & Professional Advocacy

Amicus Therapeutics, Inc.
1 Cedar Brook Drive
Cranbury, NJ 08512
USA

Amicus Therapeutics UK LTD
One Globeside
Fieldhouse Lane
Marlow SL7 1HZ
United Kingdom