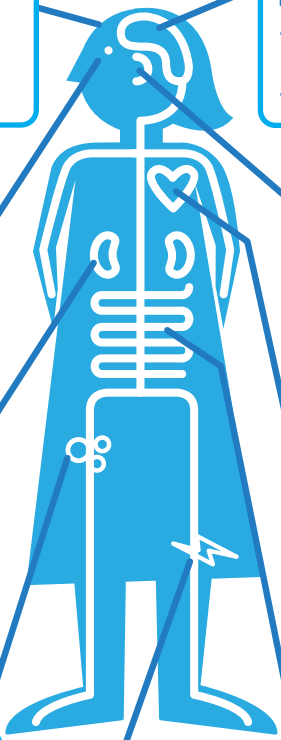
WOMEN & FABRY

This figure shows some of the more common effects of Fabry in women



DIZZINESS (VERTIGO) [3]
- Experienced by approximately 25% to 85%
- Average age of onset: 30s

FATIGUE [5]
- Experienced by approximately 59%

STROKE [3]
- Experienced by approximately 6%
- Average age of onset: 50s

MINI-STROKE (TIA) [3]
- Experienced by approximately 7%
- Average age of onset: 40s

CORNEAL WHORLING AND/OR FABRY CATARACTS [3]
- Experienced by approximately 36%
- Average age of onset: 30s

RINGING IN THE EARS (TINNITUS) [3]
- Experienced by approximately 27%
- Average age of onset: 30s

HEARING LOSS [5]
- Experienced by approximately 37%

EFFECTS ON THE KIDNEYS [3]
- Experienced by approximately 21% to 55%
- Average age of onset: 30s

EFFECTS ON THE HEART [3]
- Experienced by approximately 5% to 65%
- Average age of onset: 30s

SPOTS ON THE SKIN (ANGIOKERATOMAS) [3]
- Experienced by approximately 13% to 55%
- Average age of onset: 30s

PAIN [3]
- Experienced by approximately 53% to 90%
- Average age of onset: Teens

GASTROINTESTINAL (GI) ISSUES [3]
- Experienced by approximately 11% to 60%
- Average age of onset: 20s

Women with Fabry may experience all or just some of these effects to different extents, or they might never experience any of them.

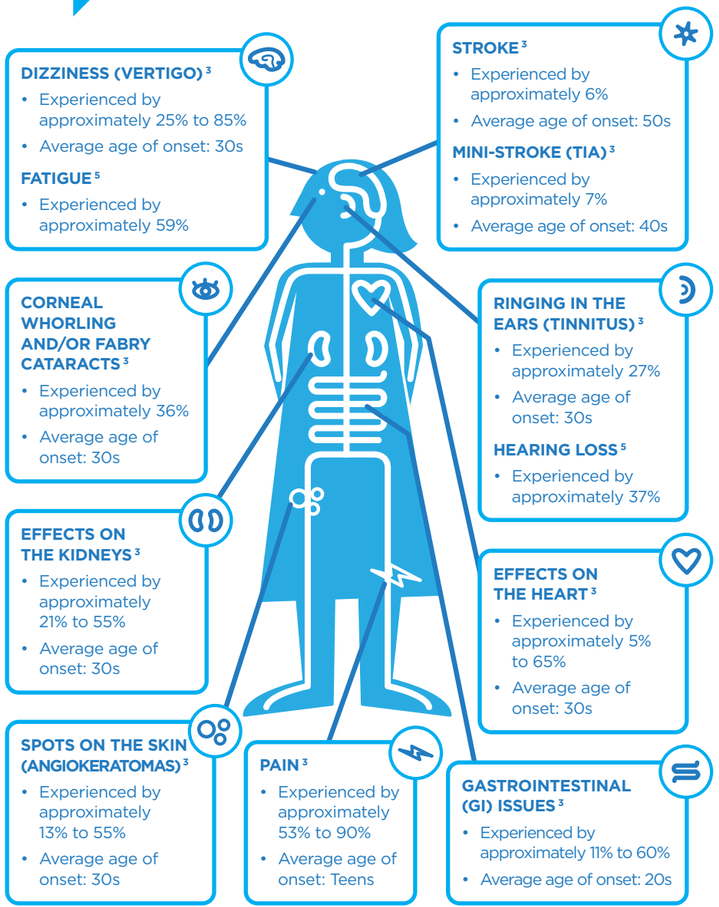
Women and Fabry

Fabry disease is an "X-linked" genetic (inherited) disorder. The disease is caused by a change in the DNA called a "mutation." This mutation happens on the X chromosome, and can be passed down by either a man or a woman.

Thousands of women around the world have Fabry disease; however, as recently as 2001, women with the mutation that causes Fabry were considered by medical professionals to be "asymptomatic carriers" of the disease, meaning they could pass on Fabry without actually having any signs or symptoms of the disease.[1] We now know that most women who have the mutation in the gene *do* experience the effects of Fabry,[2-5] sometimes as severely as affected men who have the mutation.

Women may first become aware of the effects of Fabry in their teens, but many are not diagnosed for at least 10 years after this first awareness.[3] Symptoms may range from mild to moderate to serious and debilitating, and researchers are still studying how Fabry affects women.

This figure shows some of the more common effects of Fabry in women



Women with Fabry may experience all or just some of these effects to different extents, or they might never experience any of them.

Scientists are not completely certain why there is so much variation, but some believe that it has something to do with "X inactivation." In each cell in a woman's body, there are two X chromosomes. Randomly during fetal development, one of the two X chromosomes in each cell is inactivated (this could be the normal X chromosome, or the X chromosome with the mutation associated with Fabry). This process is referred to as X inactivation. This random selection of the X chromosome may explain the variation of Fabry symptoms in women.[3] That is, the location and number of cells with an active X-chromosome with a mutation, can affect which organs are involved and the severity of a woman's experience of Fabry. Regardless of the experience of each woman with Fabry, all women with the disease should speak with their healthcare provider (HCP) about regular monitoring, since there can be significant effects of the disease.[5]

More than medical

In addition to physical signs and symptoms of the disease, many women living with Fabry have decreased quality of life.[4,5]

In one study where researchers looked at responses of women with Fabry and women without Fabry to a health-related quality-of-life survey, they found that pain was associated with lower mood and less enjoyment of life. Many women had poorer impressions of their own vitality, social functioning, and overall mental health than women who did not have Fabry. In this study, researchers found depression in almost two-thirds of women with Fabry and anxiety in more than one-third.[5]

Another study, in which 202 women with Fabry completed questionnaires to determine their quality of life, researchers found that women with Fabry had a self-reported quality of life similar to, and sometimes even worse than, that of people with multiple sclerosis (MS) and rheumatoid arthritis (RA)—other chronic diseases commonly understood to have a major impact on people's lives.[4]

Of course, we cannot forget that women often don't just think of themselves and how they are doing or feeling. Many women have multiple roles: they may be working, managing a home, and caring for others with Fabry, including children, siblings, and/or parents. This could be overwhelming for anyone! Finally, we also know that people with Fabry may have feelings of isolation, fear, and guilt for having and possibly passing the disease on to their children.

Despite having all of these physical and psychological effects, some women still find that not all HCPs believe their experiences are serious.[6]

Even though researchers agree that women are not "just carriers," as people used to believe, some HCPs still need to be convinced. This can cause serious frustration and even mistrust of doctors or other HCPs among women with Fabry.[6,7]

All of these things together—the physical, emotional, and social effects of Fabry—can make it difficult for women to find the time and the energy to take care of themselves and focus on their own health and well-being. It is this combined impact that is exactly what makes it so important. If women with Fabry are to live the best lives possible and continue to support those who count on them, they must make the time to recognise and address their own needs.

For those looking to learn as much as possible about Fabry and its effects, please refer to your local patient organisation or talk to your healthcare professional.

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ONE GLOBESIDE, FIELDHOUSE LANE,
MARLOW, BUCKINGHAMSHIRE, SL7 1HZ

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