

Women and Fabry

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

Thousands of women around the world have Fabry disease. However, in the past, women with the mutation that causes Fabry have been considered to be 'asymptomatic carriers' of the disease, meaning they could pass on Fabry without actually having any signs or symptoms of the disease. We now know that most women who have the mutation in the gene do experience some of the effects of Fabry, sometimes as severely as 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. Symptoms may range from mild to debilitating, and researchers are still studying how Fabry affects women.



If you want to learn more about Fabry and its effects, contact your local patient organisation or talk to your healthcare professional.

Information in this guide came from the following sources.

KD MacDermot, A Holmes, AH Miners, Anderson-Fabry disease: clinical manifestations and impact of disease in a cohort of 60 obligate carrier females, *J Med Genet*, 2001

MG Bouwman, SM Rombach, E Schenck, and others, Prevalence of symptoms in female Fabry disease patients: a case-control survey, *J Inherit Metab Dis*, 2012

PB Deegan, F Böhner, M Barba, DA Hughes, M Beck, Fabry disease in females: clinical characteristics and effects of enzyme replacement therapy, In *Fabry Disease: Perspectives From 5 Years of FOS*. (A Mehta, M Beck, G Sunder-Plassmann), Oxford PharmaGenesis, 2006

NJ Street, MS Yi, LA Bailey, RJ Hopkin, Comparison of health-related quality of life between heterozygous women with Fabry disease, a healthy control population, and patients with other chronic disease, *Genet Med*, 2006

RY Wang, A Lellis, J Mirocha, WR Wilcox, Heterozygous Fabry women are not just carriers, but have a significant burden of disease and impaired quality of life, *Genet Med*, 2007

AL Gibas, R Klatt, J Johnson, JT Clarke, J Katz, Disease rarity, carrier status, and gender: a triple disadvantage for women with Fabry disease, *J Genet Couns*, 2008

AL Gibas, R Klatt, J Johnson, JTR Clarke, J Katz, A survey of the pain experienced by males and females with Fabry disease, *Pain Res Manage*, 2006

© Amicus Therapeutics UK Ltd
1 Globeside
Fieldhouse Lane
Marlow
Buckinghamshire
SL7 1HZ

A company registered in England
and Wales (registration number 05541527)

NP-NN-UK-00020722

Date of preparation: July 2022



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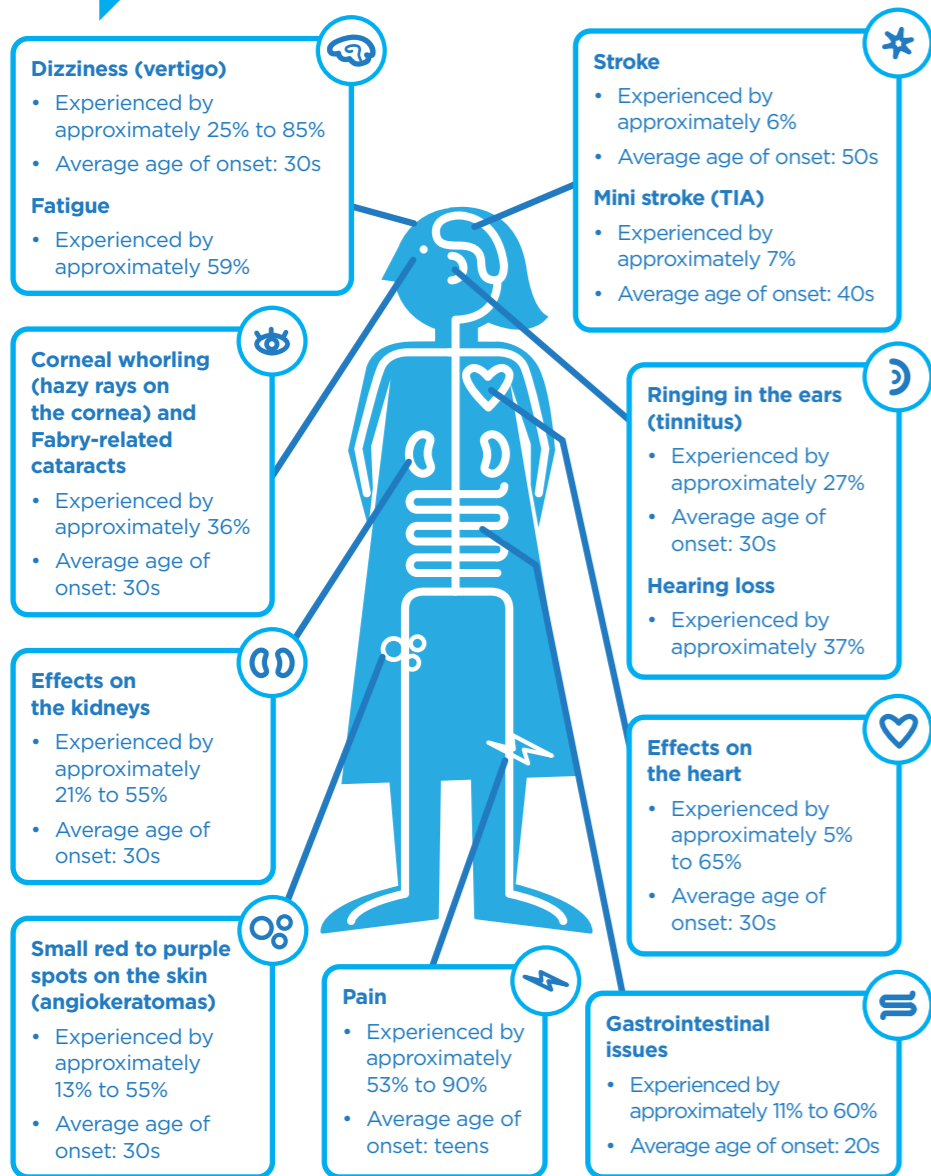


Plain English Campaign's Crystal Mark for clarity does not apply to the design of this guide.

This guide has been developed and funded by Amicus Therapeutics UK Ltd.



The diagram below shows some of the more common effects of Fabry in women.



Women with Fabry may experience all or 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 in the effects felt between women. Some believe that it has something to do with 'X inactivation'. In each cell in a woman's body, there are two X chromosomes. While a female foetus is developing in the womb, one of the chromosomes is inactivated. This could be the normal X chromosome or the X chromosome with the mutation associated with Fabry. This random selection of the X chromosome may explain the variation of Fabry symptoms between women. 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 to their healthcare professional about regular monitoring, as there can be significant effects of the disease.

More than physical

As well as the physical signs and symptoms of the disease, many women living with Fabry have a reduced quality of life compared with women without Fabry.

In one study, responses that women with Fabry and women without Fabry gave in a quality-of-life survey showed that pain was associated with lower mood and less enjoyment of life. Many women with Fabry had poorer impressions of their own vitality, social functioning and overall mental health than women without Fabry. In this study, researchers found depression in almost two-thirds of women with Fabry, and anxiety in more than one-third.

In another study, in which 202 Canadian women with Fabry filled in questionnaires on their quality of life, researchers found that women with Fabry had a quality of life similar to, and sometimes even worse than, that of people with multiple sclerosis and rheumatoid arthritis - other long-term diseases commonly understood to have a major effect on people's lives.

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

Despite all of these physical and psychological effects, some women still find that not all healthcare professionals take them seriously.

Even though researchers agree that women are not just carriers, as people used to believe, some healthcare professionals still need better education on Fabry. This can cause serious frustration and even mistrust of doctors or other healthcare professionals.

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 to focus on their own health and well-being. 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 meet their own needs.