MPS Society

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Fabry Females: Your Voices – Our Insights

Community Summary from the Webinar | Tuesday 7 April

On Tuesday 7 April, the Fabry International Network hosted the webinar *Fabry Females: Your Voices – Our Insights*, featuring Dr Hopkin.

The session shared and discussed findings from a global survey of women living with Fabry disease, conducted between October 2025 and February 2026.

This summary highlights key themes from that webinar for our Fabry community.

About the Survey

The survey gathered responses from 88 women living with Fabry disease, aged 15 to 78, reflecting experiences across a wide range of life stages. Participants came from many countries around the world.

The UK was the most significantly represented country, with 32 respondents**,** reflecting strong engagement from our UK Fabry community. This was largely due to the survey being shared widely during a period of active discussion around women's health and Fabry disease.

We would like to sincerely thank our UK members for their engagement and openness. Your participation has been incredibly valuable and provides important insight into what it means to live with Fabry disease as a woman in the UK.

Key Themes Discussed During the Webinar

A clear message from the webinar was that diagnostic delay remains very common for women with Fabry disease.

A majority of women reported experiencing symptoms for many years before diagnosis. For many, diagnosis occurred following family screening, the diagnosis of a relative, or after a serious health event, rather than through early recognition of symptoms.

Women described living with a broad and complex range of symptoms. Fatigue was most commonly highlighted as having a major impact on daily life, alongside pain (particularly in the hands and feet), heat and cold intolerance, gastrointestinal issues, hearing problems, and cognitive difficulties such as brain fog. These symptoms were described as significantly affecting work, education, relationships, and quality of life.

A particularly strong theme was the impact of hormones and life stages.

Many women described symptom onset or worsening during puberty, with further changes during menstrual cycles, pregnancy, perimenopause, menopause, and as they got older. Despite this, only a minority of women reported that hormonal health had been meaningfully discussed as part of their Fabry care. As a result, many women seek advice outside specialist Fabry services, including from GPs, gynaecologists, endocrinologists, or patient organisations – but struggled with clinicians from other specialities not having enough Fabry knowledge to support them.

Challenges related to access to treatment and ongoing care were also commonly discussed. A significant minority of women reported not being on Fabry-specific treatment, often because symptoms were not considered severe enough or because eligibility criteria focused on organ damage rather than daily impact. Issues around continuity of care and lack of understanding outside specialist centres were frequently raised.

Beyond physical symptoms, the webinar highlighted the emotional and psychological burden of living with Fabry disease. Many women spoke about anxiety, depression, the impact on work and careers, strained relationships, and the long-term effects of feeling dismissed or not believed by healthcare professionals.

What Women Want to See Change

Women's voices during the webinar were clear and consistent. They want:

- Recognition that women are patients, not carriers
- Acknowledgement that Fabry disease can be just as severe in women
- More holistic care that includes pain, fatigue, mental health, and hormonal and reproductive health
- Better awareness across the healthcare system

- More research focused specifically on Fabry disease in women and across different life stages

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Thank You to Our Community

We are extremely grateful to everyone who contributed to the survey and attended or engaged with the webinar - particularly members of the UK Fabry community. Your willingness to share experiences has given us a much deeper understanding of the realities of living with Fabry disease as a woman in the UK.

These insights will support ongoing advocacy, awareness-raising, and conversations with clinicians, researchers, and organisations such as the MPS Society, helping to ensure women's experiences are better recognised, understood, and addressed.