Self-advocacy in treatment decisions

When considering whether to start, continue or stop treatment, be it enzyme replacement therapy (ERT), chaperone therapy or another form of supportive care, it is important to ensure that decisions are based on sound clinical reasoning, the latest evidence and your individual circumstances.

This guide outlines how to advocate effectively for yourself or a loved one during these conversations, so that care remains person-centred, informed, and collaborative.

Treatment decisions are nuanced and individual

Starting or stopping treatment is rarely a simple yes or no decision. Fabry disease is a complex, multisystem condition and the role of treatment will vary over time.

There may be circumstances where not starting treatment is appropriate; particularly if an individual does not currently meet criteria for therapy based on national or international guidelines, such as those used in the UK. In early stages of the condition, active monitoring and symptom management may be the most suitable approach.

Equally, there may come a point where continuing treatment offers limited additional benefit, especially in advanced disease or when unrelated health conditions predominate.

Importantly, choosing not to initiate or continue therapy does not equate to giving up. Rather, it may reflect a clinically reasoned decision to focus care where it will have the most meaningful impact.

Treatment decisions should always be:

- Individualised to the person's clinical profile and life circumstances
- Made jointly, with time and space for discussion between the patient, their family and the clinical team
- Guided by the latest evidence and expert recommendations

It is perfectly reasonable to ask for more information, to express uncertainty, or to request time to reflect before coming to a decision.

Monitoring response to therapy

If you are receiving treatment, it's helpful to keep a record of how it is impacting your health. This can provide useful context in clinical reviews and support decisions about ongoing care.

MPS Society Alison Wilson, Support & Advocacy Officer, August 2025

Consider noting:

- Any changes in symptoms such as pain, fatigue, gastrointestinal issues, or cognitive function
- Results from objective tests (kidney function, cardiac monitoring, MRI findings, etc.)
- Any improvements in day-to-day functioning or quality of life - be as specific as possible
- What happened when treatment was delayed, missed or paused

This sort of information helps the clinical team understand how therapy is affecting you in real terms and can support future decisions about its appropriateness.

Understanding treatment criteria and guidelines

In the UK, access to Fabry-specific therapies is guided by national criteria designed to ensure that treatment is used where it is most likely to provide benefit.

It may be that, based on your current health status and test results, you do not yet meet the criteria for starting therapy. This does not mean your symptoms or concerns are being dismissed; rather, it reflects the importance of using treatment at the right time and for the right reasons.

Being aware of the treatment guidelines and the rationale behind them can help you participate more confidently in discussions with your care team.

If a decision is made not to start or to stop treatment

If you are advised that treatment is not currently indicated, you are entitled to a clear explanation.

You can reasonably ask for:

- A written summary of the decision and rationale
- Clarification of the clinical evidence or guidelines underpinning it
- A plan for ongoing monitoring and review
- The circumstances under which treatment might be reconsidered

These are complex decisions and in many cases they are reviewed periodically as your health evolves.

Joined-up care and the importance of multidisciplinary review

Fabry disease affects multiple systems, including the heart, kidneys, nervous system and, at times, mental health. A well-coordinated approach to care is essential. If your care feels fragmented or symptoms are not fully explained, it may be appropriate to request:

- A multidisciplinary team (MDT) review
- Improved communication between specialists (e.g. neurology, nephrology, cardiology)
- Consideration of non-Fabry-related health issues as part of your overall care

Written requests can sometimes be helpful in ensuring these steps are taken forward.

Advocating for someone who cannot speak for themselves

If you are supporting someone who lacks capacity to make healthcare decisions:

Clarify your role in decision-making (parent, carer, legal deputy)

Ensure their values, preferences, and prior wishes are part of the conversation

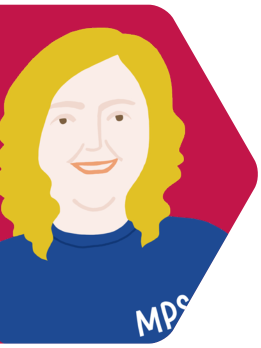
Ask for a best interests meeting

Seeking a second opinion

It is entirely appropriate to request a second opinion if you feel uncertain or if a decision does not sit well with you. **Options include:**

- Asking for another clinician's review within the same centre
- Requesting referral to an alternative Fabry specialist centre for further assessment

Second opinions are a routine part of good medical practice and can help ensure that care is aligned with your goals and needs.



Support from the MPS Society

The MPS Society is here to support you. We can help with:

- Understanding your rights and treatment options
- Preparing for appointments or writing letters
- Navigating access to treatment or requesting reviews
- Connecting you with other families, resources and peer support

You don't need to navigate this alone.

Communicating effectively

How you express your concerns can influence how they are received. If you have questions or specific topics you'd like to raise, writing them down ahead of your appointment can be helpful. **Suggested prompts:**

Can we review whether I currently meet the criteria for treatment

What are the risks and benefits of starting or continuing treatment in my case

If treatment isn't appropriate right now, how often will this be reviewed

Could a second opinion be helpful in confirming the best course of action

A final thought

Self-advocacy does not mean demanding a particular treatment, it means being actively involved in decisions about your care. Your perspective matters, it should be heard and respected.

MPS Society Alison Wilson, Support & Advocacy Officer, August 2025