

Living Well with Fabry

A SHARED DECISION-MAKING TOOLKIT

A toolkit developed by people with Fabry and Fabry specialists to support you in discussions about your Fabry care.



This toolkit has been funded by Chiesi Limited and developed in collaboration with Rare Disease Research Partners.

Job code: IE-ELF-2400021 Date of preparation July 2024
Copyright© 2024 Chiesi Limited. All rights reserved.



Living well with Fabry toolkit

Preparing for your appointment

Page 3 Talking about your symptoms

Page 4 Keeping track between appointments

Page 6 How your symptoms are affecting you

Talking about your symptoms

Page 7 Talking about pain
Page 8 Your notes: Pain

Page 9 Talking about stomach and digestive issues
Page 10 Your notes: Stomach and digestive issues

Page 11 Talking about fatigue
Page 12 Your notes: Fatigue

Page 13 Your appointment notes

Making choices

Page 14 Making choices about your care

Who is the toolkit for?

This toolkit is for anyone living with Fabry.

How can this toolkit help you?

This toolkit has been developed in collaboration with people living with Fabry and Fabry specialists and can help you talk about some of the symptoms that people have told us impact them the most. It combines their insights to help you to:

- Feel confident about raising and discussing your health concerns
- Ask the questions you need to about your Fabry care
- Be involved in managing and making decisions about your care in the way you want to be

How to use this toolkit

You can use this toolkit to help you prepare for your next appointment with your Fabry specialists.

'Think it's a great way of getting people thinking about what to mention in next meeting, and how to explain it'

- A person living with Fabry

'A very helpful tool to organise ideas and prompt questions'

- A person living with Fabry

Important note: This toolkit is intended to help you prepare for discussions with your Fabry specialists. This toolkit does not provide medical advice, always seek the advice of your Fabry specialists with any questions or concerns you have regarding your medical condition, care or treatment.



Talking about your symptoms

Addressing some concerns you might have.

'I'm not a complainer, I will just put up with it'

'It's just my Fabry'

'I'm not sure if it's due to my Fabry'

'Some doctors have not taken my concerns seriously in the past'

Understanding Fabry can be difficult because it is rare and has a wide range of symptoms, not all of which will appear in everyone. In the past doctors might not have been able to address your concerns, but your Fabry specialists are experts in supporting people living with Fabry and understand how it can impact people's lives.

'I have always had this symptom'

If something is making your day-to-day life more difficult, it's worth speaking to your Fabry specialists about it.

'I know my doctor is busy, I don't want to bother them'

Your health and well-being is your Fabry specialists' priority. This toolkit can help you to get the most out of your time with your Fabry specialist.

'There isn't enough time to discuss all my concerns'

Many people find that taking some time to prepare for medical appointments helps them to get the best out of their time with their Fabry specialists. It might help to think about what is affecting you the most. This could be done before your appointment or with your Fabry specialists.

'There is probably nothing that can be done'

Even if you think there is nothing that can help, make sure you talk to your Fabry specialist. They may be able to suggest other ways to deal with the symptoms that you are struggling with.

'There are some things I don't want to talk about'

Whilst there are some health concerns such as digestive or sexual health issues that can be uncomfortable to discuss, it is important to remember that your Fabry specialists are used to talking about these things. Everything you tell your Fabry specialists is confidential, try letting them know you are feeling nervous or embarrassed using phrases like – 'this is embarrassing for me to speak about...'

'I'm not sure if its due to my Fabry'

It can be difficult to know which symptoms are caused by Fabry or not, so remember that if you are ever in doubt you can get in touch with your Fabry specialist team who will be able to help you decide who would be the best person to talk to about your concerns.

Keeping track between appointments

You may only see your Fabry specialist once a year so it can be difficult to remember everything you want to talk to them about. This section of the toolkit is a place to keep notes between appointments.

Fill out this table before your appointment to help you and your Fabry specialist to see if and how your symptoms have changed since your last visit.



'If you have a serious problem related to your condition, please reach out to your Fabry team. They are there to help you. No need to wait until your next visit.'

A Fabry specialist working in a UK specialist centre



Symptom	Things I would like to discuss	Date

Notes: Things my relatives or caregivers would like my specialist to know.



A large writing area consisting of two columns of horizontal lines. Each column contains 15 lines, providing space for handwritten notes.

How your symptoms are affecting you

It can be difficult to describe how your symptoms are affecting you. This section of the toolkit can help you and your Fabry specialist to talk about the things that are most important to you.

I am feeling isolated

It is affecting my work or education

I can't get a good nights sleep

It is difficult to look after myself or my family

I am struggling to focus/ concentrate

I can't enjoy my social or leisure activities

Fill out this form to help you think about how your symptoms are affecting you before you discuss your care with your specialist.



How I am managing...

Think about how your symptoms are affecting your ability to manage your everyday life. Is there anything that you wanted to do over the last few weeks or months but couldn't because of your symptoms?

What are my priorities for managing my symptoms?

This could be things like, 'I want to get back to work', or 'I want to get a good night's sleep'.

Talking about pain

Pain is often invisible to others which makes it hard to measure or talk about. Before you speak to your Fabry specialist, it may be useful to spend some time thinking about how your pain is affecting you, and the questions you want to ask.

‘If you have Fabry pain, do discuss this with your specialist team. Your specialist will be able to advise on medications and liaise with your local hospital doctors or GP* for ongoing care.’

A Fabry specialist working in a UK specialist centre

*GP – General Practitioner



My pain concerns

My pain

- I don't know if my pain is related to Fabry
- My pain is getting worse
- I have noticed a change in my pain

The way I am feeling

- I feel that people don't understand my pain
- I am worried about how I will manage my pain in the future
- My pain is making me feel anxious or depressed
- My pain is stopping me doing the things I want to do



Questions to ask about pain

Questions about my pain

- Is there anything else which could be causing my pain?
- Are there other ways to manage my pain?
- Is there anyone else I should be seeing to help me manage my pain?


Questions about the way I am feeling

- What help is available to me for getting back to work or leisure activities?
- What is available to help me deal with negative feelings?
- How can I explain to people about the effect my pain has on me?
- What can help me manage my pain?
- How can I connect with other people who are going through the same thing?

Talking about stomach & digestive issues

Stomach and digestive issues can make daily life more difficult, but many people feel uncomfortable or embarrassed talking about these topics.

If you are concerned about managing these symptoms, this form may help you to discuss this with your Fabry specialist and get the information and advice you need.



'Many patients with Fabry disease have symptoms that resemble irritable bowel syndrome with diarrhoea and/or constipation with bloating and sometimes pain. Try to understand if there are any foods which trigger the symptoms and avoid these. Some patients find benefit from the FODMAP diet. Ask your doctor to speak with a dietician about this. If you have a change in your bowel symptoms or notice blood or mucus in the stool then please make an appointment to discuss with your GP urgently.'

A Fabry specialist working in a UK specialist centre



My stomach and digestive concerns

My stomach and digestive issues

- I need to know if my symptoms are due to Fabry
- My symptoms are getting worse
- My symptoms are becoming more frequent

The way I am feeling

- My symptoms are making it difficult to eat normally
- My symptoms make me anxious about going out
- My symptoms are embarrassing and it's affecting my social life



Questions to ask about stomach and digestive

Questions about my symptoms

- Is there something specific that triggers my symptoms?
- Are there any changes I can make to my diet or other treatments that can alleviate my symptoms?
- Should I be referred to a specialist?

Questions about the way I am feeling

- How can I talk to my friends and family about my symptoms without feeling embarrassed?
- Are there support groups for people with chronic stomach and digestive issues I could join?
- Is there anything available to help me with the way I am feeling?

My notes: about stomach
& digestive issues

My questions: about
stomach & digestive issues



Talking about fatigue

Living with fatigue can impact you both physically and mentally.

If you are concerned about managing fatigue, this form may help you to discuss this with your Fabry specialists and get the information and advice you need.



My fatigue concerns



Questions to ask about fatigue

‘Fatigue is a common symptom of Fabry disease as well as other conditions. You can try to manage this by pacing yourself and trying to get nutritious regular meals, good exercise and hydration and good sleep. If you experience a new change in symptoms of fatigue please mention this to your specialist’

A Fabry specialist working in a UK specialist centre



My fatigue

- I feel tired all the time, even after resting
- My energy levels are very low
- I want to understand if my fatigue is related to Fabry

The way I am feeling

- Fatigue is preventing me from enjoying my hobbies and activities
- I'm finding it hard to concentrate or stay motivated
- I'm concerned about my ability to work or study
- I feel judged by others because fatigue limits what I can do

Questions about my symptoms

- Could there be another reason for my constant tiredness?
- What strategies can help me cope with fatigue?
- Who else can support me in managing my fatigue?

Questions about the way I am feeling

- How can I help others to understand how fatigue affects me? My family? My friends? My employer?
- What resources are available to help me with how I am feeling?
- How can I connect with others who experience similar fatigue?

My appointment notes

I had this conversation with _____

Date of conversation _____

Use this page to keep a record of your discussions with your Fabry specialist. You might want to use this space to record things like:

- The answers to my questions
- Test results
- Next steps
- My specialist's recommendations



Have I got a choice to make?

Pages 14-16 can help you gather the information you need.

My next appointment is on: _____ With: _____

Making choices about your care

Fill out these forms to help you think about what you need to make a choice about your Fabry care.



Remember! You can take as much time as you need to make choices about your treatment and care. Many people find it helpful to take some time to discuss their choices with friends or family. If you need more time to talk to your Fabry specialist, you may be able to communicate with them by phone or email or ask to schedule another appointment.

Is there anything which would help you understand your choices?

- I would like information written down
- I need things explained in plain language
- I want time to consider my options
- I need adjustments to make information accessible to me

What role do you want to play in making choices about your care?

- I want to have full control
- I want to share the decision with

- I want to decide myself after hearing the views of

- I want someone else to make the decision

If you need more information about your options, ask...

- 'Where can I find more information about the benefits and risks of these options?'
- 'I want to make sure I understand, could you try explaining that in another way?' or 'can I just check that I have understood what you have said?'
- 'How will my daily activities be impacted by these options?'

If you need more support and advice to make a choice, ask...

- 'Who else can I speak with to help me decide what will work best for me?'
- 'Can I bring my friend/family member to my next appointment?'

Making choices about your care

Before making a choice about your Fabry care, make sure you get the answer to these questions:

- What are my choices?
- What is good and bad about each choice?

Fill out this form to help you think about your options when making a choice about your Fabry care.



I had this conversation with _____

Date of conversation _____

What choice needs to be made? _____

Option 1

Option 2

What is the option?

Possible benefits

Possible risks

	Option 1	Option 2
<p>What would this mean for me?</p> <p>Think about how much each of the benefits and risks of each options matter to you.</p>		
<p>Which option does your specialist recommend? Why?</p>		
<p>Which option do you prefer? Why?</p>		

If you have decided what to do next, ask...

'Is there anything I need to do or know about the choice I have made?'

'How can I stay on top of new updates about Fabry disease management?'

'Who else do I need to share details of my decision with?'

'When do I next need to come and see you?'

Remember! Whatever you choose now, even if you have decided to do nothing, it does not mean that you cannot change your mind at a later stage. Your specialist will understand that your choices might change.

Where can I get more information and support?



MPS Society

transforming lives through support, research & awareness

The Society for Mucopolysaccharide Diseases (MPS Society) is the only registered UK charity providing professional support to individuals and families affected by MPS, Fabry and related lysosomal storage diseases throughout the UK.

The MPS Society has developed into the leading provider of information and support for MPS, Fabry and related lysosomal storage diseases, with a mission to transform lives through specialist knowledge, support and advocacy and research.

Visit <https://mpssociety.org.uk/> to learn more

Society for Mucopolysaccharide Diseases

MPS House, Repton Place, White Lion Road, Amersham Buckinghamshire, HP7 9LP

0345 389 9901

mps@mpssociety.org.uk

Rare Disease Research Partners

MPS House, Repton Place, White Lion Road

Amersham, Buckinghamshire, HP7 9LP, UK

t: +44 (0) 345 260 1087

w: rd-rp.com

MPS Commercial is a Private Limited Company Registered No 08621283. MPS Commercial trades as Rare Disease Research Partners and is a wholly owned, not for profit subsidiary of the Society for Mucopolysaccharide Diseases (the MPS Society), Registered Charity in England and Wales No 1143472. Registered as a Charity in Scotland No. SCO41012. Rare Disease Research Partners social objectives are to reinvest any surplus to support the mission of the MPS Society to transform the lives of patients through specialist knowledge, support, advocacy and research.