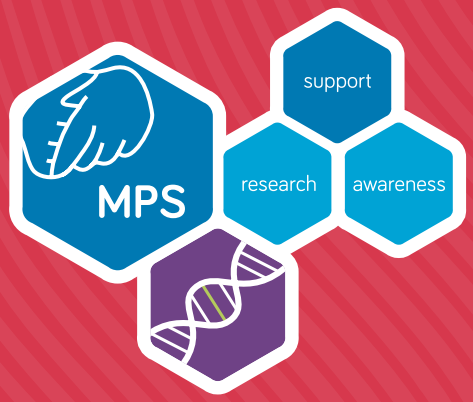

My Fabry Treatment

A guide to empower and inform Fabry patients on their treatment journey

SUPPORT | RESEARCH | AWARENESS

Society for Mucopolysaccharide Diseases

mpssociety.org.uk

Contents

Disclaimer

The information contained in this booklet is intended as a guide to help Fabry patients make an informed choice about Fabry disease-specific treatment and how to prepare for check-up appointments at their specialist treating centre. This guide does not provide medical advice, always seek the advice of your specialist doctor with any questions you may have regarding your medical condition.

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Introduction

This guide is for patients with a lysosomal storage disorder called Fabry disease. It explains the structure of care you may receive at your specialist treating centre.

If you have recently been diagnosed with Fabry disease, you can use this guide to understand how to make an informed choice about your treatment by getting the right information from your specialist doctor. You can also use this booklet to prepare for your check-ups by recording relevant symptoms and test results to track your disease progress.

Fabry disease

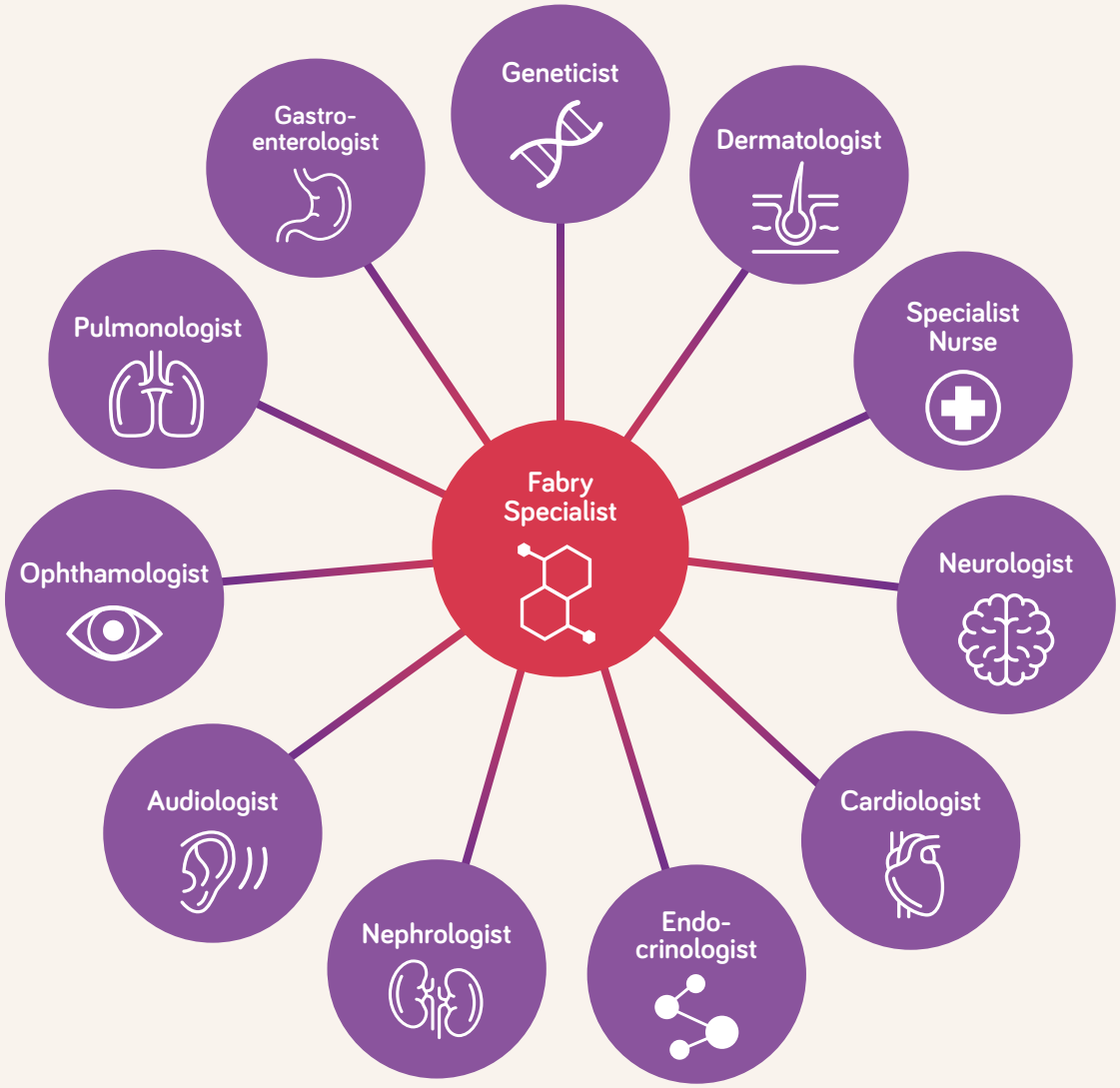
In the course of normal life there is a continuous recycling process in the body which consists of building new materials and breaking down old ones ready for disposal. This process requires a series of biochemical tools called enzymes. The enzyme alpha-galactosidase A (alpha-GAL A) is essential in breaking down the fatty acid globotriaosylceramide (also known as GL3 or Gb3).

People with Fabry cannot make enough alpha-GAL A, which leads to a build up of Gb3 within the cells of the body causing progressive damage.

A detailed information leaflet on Fabry disease can be accessed on the MPS Society website.[1]

Fabry care

A number of hospitals in the UK specialise in the care of patients with Fabry. Your care team at your specialist treating centre may consist of doctors and nurses from different specialities.



Fabry Specialist

Your Fabry specialist will be a specialist in diagnosing and treating people with a range of metabolic disorders like Fabry. They will work closely with a number of other doctors to monitor your disease progress and ensure you receive the right treatment.

Audiologist

As Fabry progresses, you may notice loss of hearing over certain frequencies, a ringing in the ears (called tinnitus) or vertigo. An audiologist or hearing specialist will keep an eye on your hearing and be able to recommend hearing aids should you need them.[2]

Cardiologist

Over time, Gb3 builds up in the blood vessels. This leads to an increased risk of stroke and progressive heart disease. Your cardiologist will check you regularly for heart problems and talk you through any treatments such as medication or pacemakers if the need arises.[2]

Dermatologist

Angiokeratomas are small, raised dark spots on the skin that arise from damage to the blood vessels. A dermatologist is a skin specialist that will be well equipped to manage these symptoms that may cause pain or be unsightly.[2]

Gastroenterologist

As Fabry can also impact on your digestive tract, you may develop digestive problems such as diarrhoea, nausea, vomiting, constipation, bloating or pain after eating. A gastroenterologist is a gut specialist who will be able to monitor your digestive health and prescribe treatments.[2]

Geneticist

A geneticist can provide genetic counselling about the risk of inheriting and passing on the disease. They can help you understand the risk of having a child with Fabry and can discuss options that may be available to minimize that risk.[2]

Nephrologist

Fabry can damage the kidneys and lead to chronic kidney disease[2]. A nephrologist is a kidney specialist that will monitor the health of your kidneys and watch for signs of kidney disease.

Neurologist

The build-up of Gb3 can affect the functioning of the central nervous system that includes the brain and spinal cord. Your neurologist will monitor any damage to your nervous system and recommend treatments.[2]

Ophthalmologist

Gb3 can accumulate in the cornea of the eye, which is the clear part of the front of the eye. This may cause brown, yellow, or grey streaks in this part leading to eyesight problems. Blood-filled cysts may also develop in the cornea of the eye as Fabry progresses. Your ophthalmologist will closely monitor your eye health and vision for any signs of problems, and will be able to prescribe treatments such as eyedrops for painful or dry eyes.[2]

Pulmonologist

You may also see a lung specialist, also called a pulmonologist, who can assess and treat breathing difficulties or causes of wheezing if you experience these.[2]

Specialist nurse

A specialist nurse has a unique relationship with each Fabry patient and their wider family to help them cope with their condition. They are responsible for administering and monitoring enzyme replacement therapy and may train patients who wish to self-administer at home.[3]

Treatment for Fabry

Fabry is a progressive disorder, and though there is currently no cure, the aim of treatment is to delay its progression or stabilise the systems it affects. If you have a new diagnosis of Fabry, you might not be automatically started on treatment. Your Fabry specialist will assess your condition and discuss with you if starting treatment is an option.[2] Two types of Fabry-specific treatment are currently available: **enzyme replacement therapy** and **chaperone therapy**.

Enzyme Replacement Therapy

This treatment replaces your missing enzyme and is delivered through intravenous infusions once every 2 weeks.[4, 5]

Enzyme replacement therapy infusions can be administered at home after several successful infusions at your specialist treating centre.

Enzyme replacement therapy is suitable for both children over 7 years of age and adults.

There are two different enzyme replacement therapies currently available. You can find information leaflets about these here:

Information on Fabrazyme®
https://www.medicines.org.uk/emc/files/pil.2419.pdf

Information on Replagal®
https://www.medicines.org.uk/emc/files/pil.9186.pdf

Chaperone Therapy

Another type of treatment available is chaperone therapy.

This type of treatment helps to stabilise the faulty enzyme and helps it to be transported to where it is needed inside the cells. It is taken as a capsule every other day at the same time of the day.

Chaperone therapy is suitable for patients aged 12 or over.

This treatment is only appropriate for those with specific types of mutations of the GLA gene. It will not work for patients with other types of mutations.

You can read more about this treatment here:

Information on Galafold®
https://www.medicines.org.uk/emc/files/pil.10934.pdf

When can treatment start?

Your Fabry specialist follows a set of guidelines that determine who Fabry-specific treatment is suitable for, when it should be started and when it may need to stop.[7]

Adults

Male patients with certain types of mutation termed “classical variants” may be considered for treatment at diagnosis.[7]

For adult females and males with ‘later onset’ disease, Fabry-specific treatment will be considered if one or more of the following are present:[7]

- Chronic kidney disease
- Persistent levels of increased protein in the urine
- Heart disease
- Uncontrolled pain or gastrointestinal problems that significantly interfere with quality of life

If only pain or gastrointestinal problems are present, then it is usual to be referred to a pain or gastrointestinal specialist for a trial of conventional treatment for these symptoms before Fabry-specific treatment is considered. Similarly, in females where only increased protein in the urine (proteinuria) is present, anti-proteinuria treatment would be tried first.[7]

Children

For children under 16 years of age, treatment for Fabry should be considered when one or more of the following Fabry-related symptoms are present:[7]

- Fabry-related kidney disease
- Fabry-related heart disease
- Painful tingling, pricking or burning feeling in the hands or feet (acroparesthesia)
- Unexplained gastrointestinal symptoms affecting quality of life

Before considering Fabry treatment for gastrointestinal symptoms in children, other common causes such as food allergies, coeliac disease or infections should be ruled out.[7]

When is Fabry treatment not an option?

According to the current guidelines, Fabry-specific treatment will not be started in the following cases:[7]

- Fabry patients that are too severely affected to benefit from Fabry-specific treatment
- Those with another life-threatening illness where the outcome is unlikely to be improved by Fabry-specific treatment
- Some patients with end stage kidney failure requiring dialysis

These patients will receive the appropriate supportive care to manage their symptoms.

Making an informed choice about treatment

When discussing treatment with your Fabry specialist it is important to understand what treatment will involve and how it may affect you and your everyday life. Your Fabry specialist will explain all the options available to you. Before starting treatment, a full discussion of the expected outcomes of treatment and the possibility of stopping treatment if the disease continues to progress should be had.[7]

Specialist centres may have access to clinical trials investigating new therapies for Fabry disease.

It may be helpful to think about what questions to ask your doctor before your appointment. Finding the answers may help your understanding and confidence in making a treatment choice together with your healthcare team.

We have included some suggestions here and there is also space for you to add your own questions and to note down the answers.

Suggested questions to ask your Fabry specialist

Questions on treatment

- Do I need treatment?
- Which treatment is suitable for me (enzyme replacement therapy or chaperone therapy)?
- Why is this treatment suitable for me?
- Can I swap to an alternative treatment if one doesn't suit me?
- How long do I take my treatment for?
- Will I be permanently on treatment?
- Are there any risks or side effects of treatment?
- How often will I need to come to hospital for check-ups?
- Who do I see if I have problems?
- What does treatment do to my disease progression?
- How long after starting treatment will I see improvement in my symptoms?

Questions on enzyme replacement therapy

- What will happen at my enzyme replacement therapy infusions?
- Where will I receive my infusions?
- How long do infusions take?
- What happens after the infusion?
- Will the treatment always be administered by a nurse or can I learn to administer the treatment myself?
- How can I schedule infusions around work, childcare, school or my other commitments?
- What is the difference between the available enzyme replacement therapies?

Questions on oral therapy

- How do I obtain the tablet therapy?
- How often do I take the tablets?
- Does it matter when I take them each day?
- Do I take them with or without food?

Your questions

Notes

My hospital appointments

Before starting treatment, you will have a number of tests and once treatment starts you will have regular check-ups. It is important that you attend all check-ups so that your specialist doctor can monitor your health. The tables below show which assessments and tests may be carried out at your first appointment, and at your check-ups during treatment.

Tests and check-ups

Adults

Those on treatment will have a review in person at their specialist centre at least once a year, with an additional review every 6 months in person or over the telephone as appropriate. The assessments may include the following:[7]

	TYPE OF ASSESSMENT	HOW OFTEN			
		Before treatment starts	Every 6 months	Every year	Every 3–5 years
	**Medical history** A detailed history of all past and current health conditions and medications	✓	✓	✓	
	**Clinical examination** Your doctor will carry out some tests to review your current health	✓	✓	✓	
	**Family pedigree** A pedigree analysis is a collection of your family members' medical history for up to three generations. This can help to identify family members with Fabry	✓			
	**Pain assessment** Pain will be measured using a questionnaire	✓		✓	
	**Quality of life assessment** A quality of life questionnaire will be completed with you	✓		✓	

	TYPE OF ASSESSMENT	HOW OFTEN			
		Before treatment starts	Every 6 months	Every year	Every 3–5 years
	**Heart function** Electrocardiogram (ECG): to assess the rhythm of the heart, measured through electrodes placed on the skin Echocardiogram: to look at the blood flow in the heart and in the nearby blood vessels, using an ultrasound device Cardiac Magnetic Resonance Imaging (MRI), to produce detailed images of the structures within and around the heart[8]	✓		✓	
	**Brain function** Brain Magnetic Resonance Imaging (MRI), for adults only: to evaluate the health of the brain	✓			✓
	**Blood tests** Collection of blood samples to measure: • Levels of all blood cell types and check for anaemia • The health of the liver and kidneys	✓	✓	✓	
	**Urine test** Collection of a urine sample to assess the health of the kidneys and how well they work	✓	✓	✓	
	**Eye tests**	✓			✓
	**Hearing tests**	✓			✓
	**Other tests as needed** Your specialist may wish to perform additional blood tests to monitor your Fabry	✓	✓	✓	

Children

Children under ten years of age will visit their specialist centre as often as their Fabry specialist feels is required based on their disease and mutation type. From the age of ten, children will be reviewed every year:[7]

- Under 5 years: reviewed as needed
- Between 5–10 years: reviewed every 1–3 years as needed
- Age 10–16 years: reviewed every year

The assessments carried out at these appointments from age five onwards are outlined in the table below. Some assessments are only done in older children as shown.[7]

	TYPE OF ASSESSMENT	AGE OF CHILD		
		5–16 years	10 years and over only	14 years and over only
	**Clinical review** Your doctor will carry out some tests to review your current health	✓		
	**Pain assessment** Pain will be measured using a questionnaire	✓		
	**Quality of life assessment** A quality of life questionnaire will be completed with you	✓		
	**Heart function** Electrocardiogram (ECG): to assess the rhythm of the heart, measured through electrodes placed on the skin Echocardiogram: to look at the blood flow in the heart and in the nearby blood vessels, using an ultrasound device	✓		
	Magnetic Resonance Imaging (MRI) of the heart may be conducted in older children			✓

	TYPE OF ASSESSMENT	AGE OF CHILD		
		5–16 years	10 years and over only	14 years and over only
	**Brain function** Brain Magnetic Resonance Imaging (MRI) to evaluate the health of the brain – **only if necessary**		✓	
	**Blood test** Collection of a blood sample to measure lyso-Gb3, an indicator of Fabry disease activity	✓		
	Additional blood test in older children to measure kidney function (calculated GFR)		✓	
	**Urine test** Collection of a urine sample to assess the health of the kidneys and how well they work	✓		
	**Additional kidney function test** A test known as a measured GFR every 3 years in older children			✓
	**Eye tests**	✓		
	**Hearing tests**	✓		
	**Growth and development**	✓		
	**School attendance**	✓		

Monitoring your Fabry and treatment

Your Fabry specialist will monitor your symptoms and quality of life during your treatment. In adults, treatment may be stopped if you develop another serious illness that impacts the benefits of continued Fabry-specific treatment, you experience intolerable side effects, or if treatment is not effective in slowing disease progression. It may also be stopped if you are not able to follow the treatment schedule.[7]

If your treatment stops because of a life-threatening illness, your care will be reviewed so that you receive the support you need.[7]

If your treatment is stopped because you were unable to stick to your treatment schedule, you will be reviewed by your specialist centre every 6 months, and if appropriate, treatment will be restarted as soon as possible.[7]

For children, treatment may need to stop if there are severe life-threatening reactions from infusions, another life-limiting illness is present or if end-stage kidney failure develops.[7]

Preparing for your check-ups

To get the most out of your scheduled clinic appointments, you may find it helpful to make a list of symptoms or problems you have had, points you want to talk about and questions you want to ask.

You may want to take a family member or a caregiver along with you to help you with discussing your Fabry, asking questions and making a note of the answers you get.

Keeping a record of your symptoms

To help your specialist to understand how your Fabry is affecting you, it may be useful to keep a note of what symptoms you are experiencing.

Some specialist treating centres offer a Fabry App where you can record your symptoms. Ask your specialist doctor if this is available at your centre. You can find out more information on the MPS Society website.[9]

You may find it helpful to fill out a copy of the next page and take it to each clinic appointment.

APPOINTMENT DATE: ..... **Time:** .....

- What I want from this appointment:

- Questions I want to ask

SINCE MY LAST APPOINTMENT I HAVE FELT:

Symptom	Doesn't affect me	Rarely	A few times a week	Daily	Multiple times a day
Sharp burning pain in the hands or feet					
Stomach pain due to diarrhoea, constipation, or both					
Nausea or vomiting					
Headaches					
Ringing in the ears					
Small, raised dark spots on the body					
Chest pain					
Dizziness					
Excessive sweating					
Changes in the eyes					
Low mood, anxiety or depression					

Additional symptoms/concerns:

I AM SATISFIED WITH MY:

	Yes	No	Somewhat
Energy levels			
Sleep			
Activity levels			
Hearing			
Eyesight			
Overall quality of life			

Print this page as many times as you need it.

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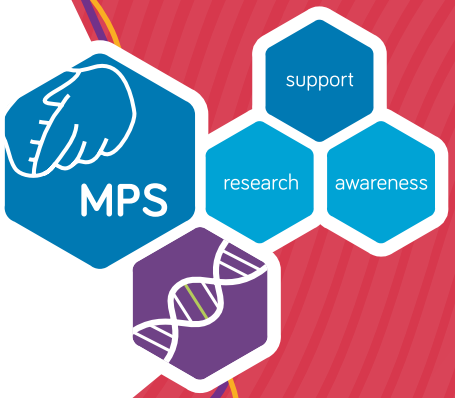
The MPS Society provides support to families affected by rare genetic conditions. We raise money and awareness of these conditions through fundraising, information, research and support.



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