

LIVING WITH FABRY DISEASE

PRACTICAL MINI-GUIDE 2

This leaflet is not intended to replace
medical advice or care.

Provided and funded by Takeda UK Ltd.



LIVING WITH FABRY DISEASE

- Doctors can now do a great deal to treat Fabry disease and improve patients' symptoms, quality of life, and outlook. However, patients can help to reduce or manage their own symptoms, and actively improve their overall health and well-being
- Fabry disease is a life-long genetic disorder in which the lack of a specific enzyme leads to the build-up of a fatty substance in certain cells and tissues, eventually causing them to malfunction and deteriorate. This results in a range of symptoms and complications, including pain, rashes, eye and hearing problems, and, more seriously, kidney disease and heart conditions
- Although therapies for Fabry disease target the underlying cause of the condition, choosing a healthy lifestyle and having regular medical check-ups will undoubtedly help your body to fight the disease and keep your heart and kidneys as fit as possible

This mini-guide contains practical advice on how to reduce the impact of Fabry disease. With a little help from the people around you – and a lot of help from yourself – there is no reason why you should not live a long and happy life

- Try not to let Fabry disease rule your life. Let the doctors take care of your illness, and you take care of you!

AVOIDING PAIN AND OTHER SYMPTOM TRIGGERS

Many people with Fabry disease report their pain as being the worst aspect of their condition, and it is often one of the most difficult symptoms to treat.

The pain frequently worsens with exercise, stress, alcohol consumption, fever and variations in temperature.

Many people with Fabry disease find that by avoiding these and other triggers, they can help to gain some control over their own symptoms.

Here we offer a few tips on how to avoid pain and other symptom triggers in Fabry disease.

To reduce the risk or severity of pain

- Avoid changes in temperature or humidity – layering clothing can help keep the body temperature steady
- Drink plenty of fluids after periods of activity
- Avoid excessive physical exertion
- Avoid stressful situations where possible
- Minimise your alcohol intake
- Try not to allow yourself to get over-tired – take frequent rests if you can
- Consider the use of cold compresses on painful areas of skin
- Keep away from sources of infection and visit your doctor regularly

To reduce the severity of skin symptoms

- Avoid prolonged exposure to direct sunlight

To reduce the incidence and severity of gastrointestinal symptoms

- Eat smaller meals more often – but do not be afraid to eat
- Keep a food diary and identify the foods that cause the most severe symptoms
- Make healthy food choices
- Reduce the amount of fat in your diet



“My hands and feet get unbearably painful, especially if they’re touched by hot or cold water. The pain prevents me from immersing myself and I shower at a specific temperature that keeps the trauma to my body at a minimum.”

George – A Fabry patient

Anecdotal quote for illustrative purposes. Patient name and picture are not real

LIVING A HEALTHY LIFESTYLE

We all know that choosing a healthy lifestyle can help protect us from serious illness and, possibly, increase our life expectancy. That's as true for people who don't have Fabry disease as it is for those who do. But what does 'living a healthy lifestyle' really mean?

1. Try not to smoke

Stopping smoking can be the single most effective thing you can do to reduce your risk of future illness.

There are five basic factors that most people consider important in order to stay as healthy as possible.

Some are more important for people with Fabry disease; others less so:

2. Do some regular physical activity

Regular physical activity (at least 30 minutes most days) is advisable for most individuals.

Unfortunately, people with Fabry disease often find it impossible to do so as exercise triggers or worsens painful symptoms, and causes over-heating and discomfort.

If you can find an activity that you enjoy and can tolerate, and that increases your heart rate without causing discomfort, that would be ideal.

3. Eat a healthy diet

People with Fabry disease are often fearful of food because eating can trigger abdominal symptoms. Nevertheless, eating a healthy diet is still important for you, so you should try to follow these guidelines, even if you have adapted the way you eat to suit your illness.

- Eat at least 5 portions of a variety of fruit and vegetables each day
- The bulk of most meals should be starch-based foods (such as cereals, wholegrain bread, potatoes, rice, pasta), plus fruit and vegetables
- Do not eat too much fatty food, such as fatty meats, cheese, full-cream milk, fried food, butter
- Include 2-3 portions of fish per week, at least one of which should be 'oily' (such as herring, mackerel, sardines or fresh tuna)
- If you eat meat, it is best to eat lean meat or poultry, such as chicken
- If you do fry food, choose a vegetable oil, such as sunflower or olive oil
- Try not to add salt to food, and limit foods that are salty. This is particularly important for people with Fabry as a low salt diet can help to maintain better kidney function

4. Try to achieve your ideal weight

Many people with Fabry disease are underweight, probably because they do not absorb nutrients from food as well as others, and because food can be a symptom trigger. Whether you are under or overweight, you should find out what the ideal weight for your height should be, and work towards achieving it. You may need the help of a dietician or doctor to do this.

5. Don't drink too much alcohol

A small amount of alcohol is usually fine, if it doesn't trigger your symptoms, but drinking too much can be harmful. Men and women should drink no more than 14 units per week (and no more than 3 units per day).

1 unit of alcohol = half a pint of normal strength beer, a small glass of wine or a small measure of spirits.

If you have Fabry disease you can help yourself stay healthy by:

- Having regular medical and eye check ups, and seeking help quickly if you feel unwell
- Keeping all your hospital appointments
- Understanding the impact that stress may have on you and avoiding stressful situations where possible
- You might consider learning some relaxation techniques to help you through times of stress

COPING WITH SCHOOL, COLLEGE OR WORK

The symptoms of Fabry disease typically appear during childhood or adolescence, although often people are not accurately diagnosed until their 20s or 30s.

Because of their severity and unpredictability, the symptoms of Fabry disease can sometimes interfere

with school or college life, and may cause some problems at work.

However, with a little forward planning and good communication with teachers or employers, it is perfectly possible for people with Fabry disease to do well at school and have good careers.

COPING WITH SCHOOL OR COLLEGE

- Children or young people with Fabry disease can feel different from their peers and may, as a consequence, withdraw unnecessarily from social or school activities
- Visible signs, such as rashes around the belly and legs, can make youngsters feel self-conscious and feel at risk of being singled out or bullied. A reduced ability to participate fully in sporting or other activities – because of the pain – can also set a child apart from their classmates
- By understanding the condition and the limitations it places on the child, schools and colleges should be able to ensure the child's comfort, while including them in most activities
- To help youngsters cope with their school life, and avoid feeling left out and lonely, parents or guardians should inform the school and explain to the teachers what happens to their child as a result of Fabry disease, and what they can and can't do
- A well-informed school or college should also be more understanding and supportive if the child needs to take medications while at school or have periods of absence for hospital visits

COPING WITH WORK



- Many people with Fabry disease have no problems with work and have successful careers, but that isn't true for everyone. Some people with Fabry disease will need more support than others at work because their symptoms make it more difficult to cope with work
- Certain activities and environments can produce discomfort and/or worsen symptoms for people with Fabry disease. In choosing a job, you should try and find one that you wish to do while recognising that some occupations might prove uncomfortable. Pain can be caused through activities that require a great deal of manual dexterity, exposure to rapid changes of temperature or humidity, that require physical exertion or cause stressful situations
- As tiredness is often a symptom of Fabry disease, periods of rest may need to be built into the normal working day
- A well-informed employer will probably be more accepting of any physical limitations or work absences due to sickness or treatment than an uninformed employer

BUILDING A GOOD SUPPORT NETWORK



- Everyone needs a good support network and to know that there are people around to share the good times, offer comfort during the bad times, and be a reliable back up when practical help is needed
- Having a long-term illness like Fabry disease, with its unpredictable, often disabling symptoms, can mean you need to push yourself a little harder than most – sometimes when you really don't feel like it – so make sure you have a strong team of people around you who understand your illness and what you might need from them. Friends, family and, of course, your medical team can make a really big difference to your experience of living with Fabry disease
- The MPS Society is an independent charity providing a range of information and individual support to Fabry patients and their families (see the contact details for the MPS Society at the end of this booklet)



"I am alive, with a new lease of life, and I feel very lucky. It is really important to be treated by a specialist unit where they understand the condition and how the symptoms manifest in different ways." David – A Fabry patient

Anecdotal quote for illustrative purposes. Patient name and picture are not real

THE MEDICAL AND HEALTHCARE TEAM

- Fabry disease is a complex illness that is usually managed by a variety of different healthcare professionals with different skills and expertise
- Because it is a rare condition, most people with Fabry disease are treated at specialist centres by doctors and nurses who are experts in treating lysosomal storage disorders like Fabry disease
- If you have been referred to a specialist centre, you will probably find that the centre oversees your ongoing care, bringing in other healthcare professionals when needed. You might see one or all of the following professionals while you are undergoing assessment or treatment for Fabry disease:



ENT, ear, nose and throat

YOUR FAMILY AND FRIENDS

- Family and friends are also vital members of your support network, but they need to understand Fabry disease and how it affects you if they are to offer the right help when you need it most. Learning that you have a genetic disorder will probably have been as much a shock to them as it was to you – especially members of your immediate family who will naturally worry about their own health, and whether they too have inherited the condition or passed it on to their children

It's important to remember – and to reassure your family and friends – that many people with Fabry disease live long, happy and fulfilling lives.

- Fabry disease is easy to diagnose with a simple blood test (if family members are worried they may have the condition), and effective treatments are now available. You are by far the best person to educate and inform your family and friends about your condition and how it affects you. However, if you need a little extra help, there is a Family and Friends guide available that is full of useful information for your support network. Alternatively, you could ask your doctors or nurses if they would be willing to talk to anyone in your family concerned about your, or their own, health



OTHER PEOPLE WITH FABRY DISEASE

Many people with Fabry disease gain great support from other patients.

Thanks to the internet, it is now possible for Fabry patients around the world to join together and share

- There are multiple forums on the internet and social media groups devoted to Fabry disease. These are independent websites unrelated to Takeda and Takeda has no control over their content
- Having so much information available on the internet and social media is great; however, not all of it is trustworthy or helpful. Use your critical thinking skills and talk to those around you to figure out what is reliable, helpful and good for you to use
- The MPS Society is an active support group for people with Fabry and their families, and provides lots of information and individual support and also connects patients and families with one another. (See the contact details for the MPS Society at the end of this booklet.)

OTHER MINI-GUIDES IN THIS SERIES INCLUDE:



What is Fabry disease?

An introductory information guide about Fabry disease. Topics covered include: what causes Fabry disease, how the disease is inherited, symptoms of the disease and how they can be managed.



Do you know someone with Fabry disease?

A practical guide to offer to your support network to aid their understanding of Fabry disease.



THE MPS SOCIETY

The MPS Society is a UK charity supporting individuals and families affected by mucopolysaccharide and lysosomal storage diseases, including Fabry Disease, throughout the UK. It offers a unique opportunity for people affected by Fabry disease to get in touch with other people in similar circumstances and it provides up-to-date information on management, treatment and living with Fabry disease.

For further information about the MPS Society please, contact:

MPS Society Helpline:
+44 (0) 345 389 9901

Email: mps@mpssociety.org.uk
Web: www.mpssociety.org.uk

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



Copyright© 2020
Takeda Pharmaceutical Company Limited

Date of preparation: June 2020
C-ANPROM/UKI//0308