foundation
Supporting your life with Fabry disease

# DO YOU KNOW SOMEONE WITH FABRY DISEASE?

## A GUIDE FOR FAMILY AND FRIENDS

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

Provided and funded by Takeda UK Ltd.

# THE ‘FAMILY AND FRIENDS’ TEAM

We all need a good support network and to know 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, means that your friend or relative may need more support than most. Friends, family and, of course, their medical team, can make a really big difference to how well they cope with having Fabry disease.

Learning that your friend or relative has been diagnosed with a genetic disorder will probably have been as much a shock to you as it was to them – especially if you are a member of the immediate family and are now worried about your own health, and whether you too have inherited the condition or passed it on to your children.

It’s important to remember – and to reassure all members of the ‘family and friends’ team – that many people with Fabry disease live long, happy and fulfilling lives. The condition is easy to diagnose with a simple blood test (if you are worried you may have the condition), and effective treatments are now available.

The best way for you to help your friend or relative is to learn as much as you can about Fabry disease, and try and understand how it affects them. And reading this booklet is certainly a good place to start! However, if you need a little extra help, you could look at the MPS Society mentioned at the end of this booklet. Alternatively, you could see if your friend or relative’s doctor or specialist nurse might be willing to talk to you if you are concerned about their, or your own, health.

# WHAT IS FABRY DISEASE?

**Fabry disease (sometimes known as Anderson–Fabry disease after the two scientists who discovered it) is a rare genetic disorder caused by the deficiency of an important enzyme called alpha-galactosidase A**

- Normally, the enzyme alpha-galactosidase A is present in most cells of the body and helps to break down fat-like substances called lipids
- People with Fabry disease have inherited a faulty gene, which leads to reduced levels of, or poorly functioning, alpha-galactosidase A. This causes a build up of a fatty substance called globotriaosylceramide (Gb3) in parts of the cells referred to as lysosomes
- Because Gb3 starts to accumulate in the lysosomes of cells, Fabry disease is often referred to as a `lysosomal storage disorder'
- Gb3 tends to build up most in kidney, heart and nerve cells, and in the walls of blood vessels. Eventually, the cells and tissues begin to deteriorate, and symptoms start to appear
- The tissues and organs most severely affected by Fabry disease are the skin, eyes, kidneys, heart and brain

# WHO GETS FABRY DISEASE?

- Fabry disease affects men and women of all ethnic backgrounds in all parts of the world. Symptoms of the illness can begin in childhood, although most appear during adolescence or early adulthood
- Although Fabry disease is one of the most common lysosomal storage disorders, it is classified as a rare disease, affecting an estimated 1 in 40,000–60,000 males and 1 in 20,000 females in the general population

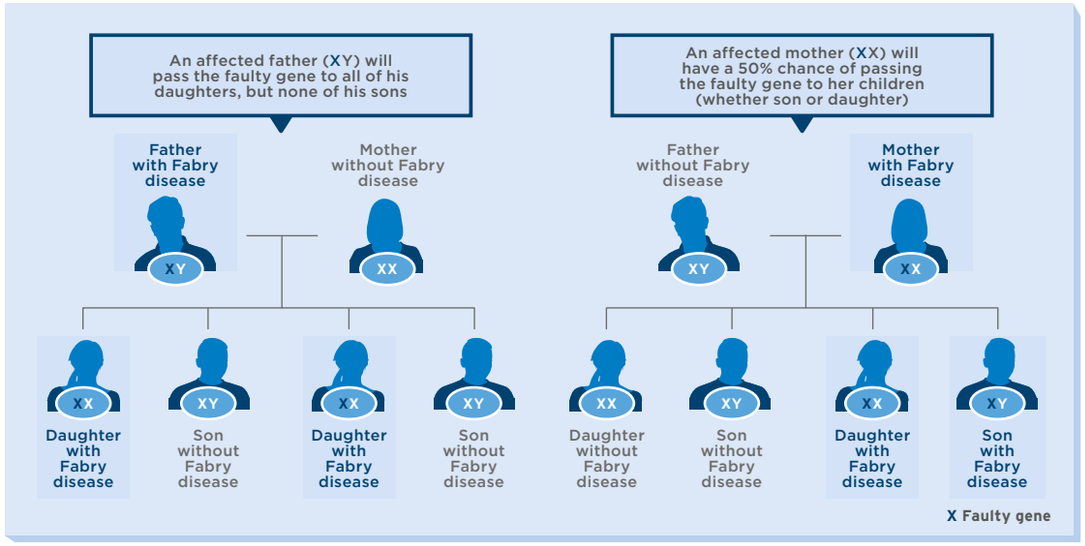


# MEN

- Men only have one X chromosome (and one Y chromosome), so if it contains the faulty Fabry gene, they will more than likely develop symptoms of Fabry disease
- Men pass on their Y chromosome to their son, so they cannot pass on the condition to their son. However, if they have a daughter, they will pass on their X chromosome and a copy of the faulty gene

# WOMEN

- Women have two X chromosomes, so if a woman has Fabry disease, one of these will have a copy of the faulty gene (unless both parents pass on X chromosomes containing copies of the faulty gene)
- Females with Fabry disease have a 50% chance of passing on the faulty gene to their children, regardless of whether the child is a boy or girl because they will pass on an X chromosome to their son or their daughter
- Females may have less severe disease manifestation than men but this is not always the case. The reason for this is that females have two X chromosomes, one of which will be active and one inactive. It is a matter of chance which chromosome is active in a particular cell. If the pattern of activation is skewed in favour of the X chromosome with the faulty gene, then a female is likely to have more severe symptoms




**For more information about how Fabry disease is inherited, please contact the MPS Society for their booklet on inheritance www.mpssociety.org.uk**

# WHAT ARE THE SYMPTOMS OF FABRY DISEASE?

- Fabry disease is associated with a wide range of symptoms that can vary from day-to-day and person-to-person. Some people with Fabry disease may have only mild symptoms for the whole of their lives, while others will suffer severe symptoms from an early age. Symptoms tend to appear at a younger age in boys than in girls
- Because of the lack of disease-specific symptoms, the diagnosis of Fabry disease is often delayed, with a mean delay from onset to diagnosis of 13.7 years in men and 16.3 years in women
- Pain is often the first symptom of Fabry disease that people notice, and is often in the form of a burning or tingling sensation in the hands and feet. However, diagnosis of what causes pain is difficult. Fabry disease is often initially misdiagnosed as neuropathic pain

**Early symptoms (sometimes appearing for the first time in childhood) include:**

- Pain and discomfort in the hands and feet, often brought on by hot weather or exercise
- Spotted, dark red rashes (called angiokeratomas) seen mostly between the belly button and knees
- A decreased ability to sweat (perspire), resulting in over-heating and decreased tolerance to heat
- Characteristic changes on the cornea of the eye, without the vision being affected
- Abdominal pain, diarrhoea, nausea and vomiting

*"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.



**As Fabry disease progresses with time, additional symptoms may appear, including:**

- Fatigue (often extreme and disabling)
- Abdominal cramping, frequent bowel movements shortly after eating, diarrhoea and nausea
- Headaches
- Hearing loss or tinnitus (ringing in the ears)
- Swelling of the ankles
- Chest pain or palpitations

**Potentially serious complications associated with Fabry disease include:**

- Kidney disease, leading to loss of protein in the urine (proteinuria) and possible kidney failure
- Heart conditions, including changes in the shape and function of the heart, irregular heartbeat, and leaky heart valves
- Disruption to normal blood flow in the brain. This can cause dizziness and, in some cases, strokes

- All of these symptoms result from the gradual build up of the fatty substance, Gb3, in the cells and tissues. If left untreated, vital organs eventually start to deteriorate, and severe, sometimes life-threatening, complications can arise
- The good news is that progression of the disease can be slowed with effective treatments

# WHAT IS THE OUTLOOK FOR PEOPLE WITH FABRY DISEASE?

- Fabry disease is a serious illness that can be painful, distressing and disabling. Many individuals with Fabry disease, if left untreated, find the pain and other symptoms intolerable, are unable to study or work effectively, and report feeling depressed, anxious, tired and frustrated
- The range and severity of day-to-day symptoms can be made worse by the fear of serious complications and the risk of major, potentially life-threatening events such as heart attacks and strokes
- Life expectancy for people with Fabry disease has, in the past, been estimated to be around 15-20 years shorter than for the general population if left untreated

Current treatments have improved life expectancy by about 15 years compared with untreated patients.

- Many experts now believe that earlier diagnosis and better treatment of Fabry disease hold the potential to change the natural course of the illness
- Put simply, the outlook for people with Fabry disease has never been brighter, thanks to the continuing work of thousands of scientists, doctors, and patients around the world

# AVAILABLE TREATMENTS FOR FABRY DISEASE

**There are two basic approaches to the management of Fabry disease:**

- Treatment to manage symptoms or the impact of the disease
- Treatments designed to replace or supplement the alpha-galactosidase A enzyme activity.
  - Enzyme replacement therapy (ERT), uses regular infusions to replace the missing enzyme
  - Chaperone therapy is an oral treatment to help restore the activity of the defective enzyme. Genetic testing will show whether a patient is expected to respond to this treatment option

*"It is vital to get help as early as possible, so that the condition may be stabilised to reduce the damage that is caused by the disease."*
Ted – A Fabry patient

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

# SELF-HELP FOR PEOPLE 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 themselves to reduce or manage their own symptoms, and actively improve their overall health and well-being
- Although therapeutic treatments can reduce progression of the disease, people with Fabry disease can also try to adopt a healthy lifestyle and have regular medical check ups in order to stay as fit as possible
- The next few pages of this booklet contain practical advice on how people with Fabry disease can help themselves to stay fit and well

**As a relative or friend of someone with Fabry disease, perhaps you could encourage them to follow some of these simple ideas?**

After all, with a little help from the people around them – and a lot of help from themselves – there is no reason why someone with Fabry disease should not live a long and happy life

# HOW PAIN AND SYMPTOM TRIGGERS CAN BE AVOIDED

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 symptoms. Here we offer a few tips on how people with Fabry disease can reduce the risk or severity of their symptoms.

**To help reduce the risk or severity of pain, patients can...**

- 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 alcohol intake
- Try not to allow themselves to get over-tired – rest frequently if they can
- Consider the use of cold compresses on painful areas of skin
- Keep away from sources of infection and visit their doctor regularly

**To help to reduce the severity of skin symptoms, patients can...**

- Avoid prolonged exposure to direct sunlight

**To help to reduce the incidence and severity of gastrointestinal symptoms, patients can...**

- Eat smaller meals more often – but 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 their diet

# THE IMPORTANCE OF A HEALTHY LIFESTYLE

We all know that choosing a healthy lifestyle can help protect us from serious illnesses 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?

**There are five basic factors that most people consider important in order to stay as healthy as possible. Some of these are more important for people with Fabry disease:**

1. Trying not to smoke
2. Ideally, trying to do some regular physical activity
3. Eating a healthy diet
4. Trying to achieve their ideal weight
5. Trying to avoid drinking too much alcohol

**People with Fabry disease can also help themselves stay healthy by:**

- Having regular medical and eye check ups
- Seeking help quickly if they start feeling unwell
- Keeping all their hospital appointments
- Understanding the impact stress may have on them and avoiding stressful situations where possible. As a friend or relative, perhaps you could help them to learn some relaxation techniques –after all, we could all benefit from learning how to cope with stress a little better!

*"Sports like football and swimming were out of the question because they made my limbs ache. The teachers thought I was faking it and gave little sympathy; meanwhile other kids thought I was a wimp because I wouldn't join in."* Sam – A Fabry patient

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



# COPING WITH SCHOOL, COLLEGE OR WORK

People with Fabry disease typically start to experience symptoms during childhood or adolescence although accurate diagnosis often won't occur until they reach their 20s or 30s. The severity and unpredictability of the symptoms of Fabry disease can sometimes interfere with normal life, at school, college or work.

By using a little forward planning and ensuring they communicate well 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

- The effects of Fabry disease on children or young people may lead them to withdraw unnecessarily from social or school activities because they may start to view themselves as different from their classmates
- 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
- To help youngsters cope with school life, parents or guardians should explain to teachers how Fabry disease affects their child, and what they can and can't do. By understanding the condition, and the adjustments which might be needed, schools and colleges should be able to ensure the child's comfort, while including them in most activities
- A well-informed school or college will 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, they should try and find one that they 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 feature 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

*"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

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# 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 your friend or relative has been referred to a specialist centre, you will probably find that the centre oversees their ongoing care, bringing in other healthcare professionals when needed. They might see one or all of the following professionals while they are undergoing assessment or treatment for Fabry disease:


ENT, ear, nose and throat

# MEETING 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 come together and share their stories and experiences.

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.

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.



# 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

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Date of preparation: June 2020
C-ANPROM/UKI//0309