



# Guide to Understanding Fabry Disease



Margaret & Family

## Contents

- What is Fabry Disease?
- What Causes Fabry?
- How Common is Fabry?
- Diagnosis of Fabry
- Overview of Symptoms
- How is Fabry Disease Inherited?
- Fabry in Males, Females and Children
- What Should I Do Now?
- How do I get tested for Fabry?
- Genetic Counselling
- Clinical Presentation of Fabry
- Development of Fabry Disease
  - Children
  - Adolescents
  - Adults
- Atypical Variants
- Living with Fabry
- Insights from Fabry Sufferers
- My Story by *D. Minett*
- Treatment for Fabry Disease
- Further Information
- About the MPS Society

## What is Fabry Disease?

Fabry Disease, also known as Anderson-Fabry Disease, is closely related to Mucopolysaccharidoses and is one of a number of disorders known as lysosomal storage diseases.

Whilst there is no cure for individuals affected by Fabry Disease this fact sheet explores the disease's presentation and clinical management.

This fact sheet is produced by the Society for Mucopolysaccharide Diseases (MPS Society) and draws upon the experiences of those affected by Fabry Disease including individuals, their families and doctors. It also makes reference to medical literature.

## What causes Fabry Disease?

In the course of normal life there is a continuous recycling process which consists of building new materials and breaking down old ones ready for disposal. This activity takes place in a special part of the body's cells called the lysosome. This process requires a series of biochemical tools called enzymes. Enzymes can only reach the lysosomes after a special signal has been attached to them.

Children and adults with Fabry Disease are missing, or are deficient in an enzyme called alpha-galactosidase A which is essential in breaking down certain waste products in the lysosomes of many different types of cell.

The main waste product is ceramide trihexoside (CTH), also known as globotriaosylceramide, Gb3 or G13.

When these waste products are not completely broken down they build up within the cells of the body causing progressive damage. Babies may show little sign of the disease but as more and more cells become damaged by an accumulation of these waste products, symptoms start to appear.

## How common is Fabry Disease?

The prevalence of Fabry Disease is estimated to range between 1:40,000 and 1:117,000 among males in Caucasian populations.\* The disease is seen across all ethnic and racial groups, but the population-specific incidence rates are unknown.

\*Branton, MH, Schiffmann, R, Sabris, SG et al. Natural History of Fabry renal disease: influence of alpha-galactosidase A activity and genetic mutations on clinical course. *Medicine (Baltimore)* 2002; 18:122. Ref 2: Meikle, PJ, Hopwood, JJ, Clague, AE, Carey, WF. Prevalence of lysosomal storage disorders. *JAMA* 1999; 281:249.

The prevalence previously stated may be underestimated as a number of males with organ-specific variants (e.g. cardiac variant) may not be diagnosed as having Fabry Disease although, for instance, their cardiomyopathy may be recognised and treated. It should also be noted that women can also be affected by Fabry Disease with the disease manifestation tending to occur at a later age of onset than in males, but this is not always the case.

## Diagnosis of Fabry Disease

Fabry Disease encompasses a wide spectrum of clinical symptoms which may or may not appear in all individuals with this disease. This together with the rarity of the disease often delays diagnosis. Many individuals may experience some of the symptoms that are outlined in this fact sheet before receiving an actual diagnosis of Fabry.

## Overview of Symptoms

The initial indications of Fabry Disease can appear in early childhood; however, not all patients will experience this at such a young age.

Symptoms Include:

- Burning sensations (or pain) in the hands and feet
- Headaches
- Vertigo
- Tinnitus (A ringing sound in the ears)
- Small raised dark red spots on your body (Called Angiokeratomas)
- Fever
- Sweating too little (A condition called hypohidrosis)
- Intolerance to heat
- Abdominal Pain
- Vomiting & Diarrhoea
- Impaired Hearing
- Alterations in the eye eventually leading to clouding that can occur in the cornea (Known as 'Opacity of the Cornea')

Studies indicate that patients experience pain in varying degrees that can cause depression, fatigue and feelings of social isolation. Over time, patients typically develop more serious symptoms that affect the kidneys, heart and brain. In many cases a diagnosis of Fabry Disease only occurs after another family member is found to have the disease. It is, therefore, important to let other members of your family know, particularly if any of them are pregnant, or planning to become pregnant. Your doctor can advise you where to obtain genetic counselling and give you information about pre-natal diagnosis of Fabry Disease. Both amniocentesis and chorionic villus sampling can be used to diagnose Fabry Disease in utero.

## How is Fabry Disease Inherited?

We all inherit genes from our parents. These genes are contained on 46 chromosomes arranged in 23 pairs. One of these pairs determines whether an individual is male or female. Females have two 'X' chromosomes and males have one 'X' chromosome and one 'Y' chromosome. The 'X' chromosome is inherited from the mother and the 'Y' chromosome is inherited from the father. In Fabry Disease, the defective gene is located on the 'X' chromosome. The disease, therefore,

follows an X-linked inheritance pattern.

On average, an affected female carrier will pass the disease on to 50% of her sons whilst 50% will remain healthy. 50% of her daughters will, on average, be carriers. An affected father will have sons who are all healthy, whilst all of his daughters will be affected.

## Fabry Disease in Males, Females and Children

Fabry Disease is an X-linked inherited disorder and is typically expressed in affected males. Females are symptomatic in a high proportion of cases and it is now clear that females may present a range of severe disease manifestation similar to that seen in males. Disease manifestation in females tends to occur at a later age than in males, but not always. Symptoms typically start in childhood but the diagnosis is often missed in the index case. As a result there is frequently a delay between the onset of the signs and symptoms of Fabry Disease and diagnosis.

## What Should I Do Now?

If you have been diagnosed with Fabry Disease you may want to consider discussing this with your relatives. Not only will this help your family understand about Fabry Disease, but it is also important because Fabry Disease is a genetically inherited condition.

If you or someone in your family has been diagnosed with Fabry, it is possible that other members of your family may also be at risk of having inherited the condition. If you suspect that this may be the case it is very important to contact your GP. You can take this fact sheet with you as it may be helpful for your doctor.

Alternatively, there are a number of Specialist Centres (known as National Commissioning Group or NCG Centres) where you can go to get tested and see a specialist in Fabry Disease. You will also have the opportunity to see a Genetic Counsellor. You can contact the MPS Society for a comprehensive list of NCG Centres.

It is vitally important that other family members are encouraged to consider undergoing testing themselves once a diagnosis of Fabry Disease has been received within the family. This will ensure that informed decisions can be made over treatment options and reproductive choices.



Laura & Juanita

## How do I get tested for Fabry Disease?

There is a genetic test that can be used to confirm whether or not a patient has Fabry. The doctor will check the level of the enzyme (Alpha-galactosidase A) as there are lower levels than normal in a Fabry sufferer.

Tests in the early stages of pregnancy are available and can determine whether the fetus is affected by Fabry Disease. DNA analysis determines whether or not a woman is a carrier by finding the specific change or mutation in the Fabry gene. When this is known, any female relatives can be reliably tested.

## Genetic Counselling

All parents of children with a lysosomal storage disease should consider asking for genetic counselling before having other children. The counsellor should be able to provide non-directive advice on the risk to close relatives, the pattern of inheritance, family planning, genetic screening and other issues. They will also be able to advise as to whether the wider family should be informed.

## Clinical Presentation of Fabry Disease

Fabry Disease encompasses a wide range of clinical symptoms which may not appear in all individuals with the disease. These symptoms usually worsen as the individual gets older. It should be noted that not all individuals with Fabry Disease will experience all of the symptoms outlined in this factsheet.

## Pain

Pain is one of the most distinctive symptoms of Fabry Disease and is often the symptom that sufferers first notice. It may even have gone undiagnosed in childhood. Fabry pain is caused by the accumulation of waste products in the nerve cells and it can be broadly divided into two distinct types, namely constant background pain and short term severe pain. Both types of pain are usually a result of certain trigger factors such as changes in temperature, episodes of stress, or physical activity.

Constant, chronic pain takes the form of a burning, tingling sensation (normally in the hands and feet), which results in constant discomfort. This is known as 'acroparasthesia'.

Short term, severe pain is often known as a 'Fabry Crisis'. Lasting for a variable period of time, from a few minutes to several days, a 'Fabry Crisis' is often described as an intense burn which starts at the extremities (the palms and the soles of the feet) and spreads throughout the rest of the body.

Fabry pain, in whichever form, can be debilitating and may affect everyday activities. Individuals affected by Fabry pain may choose to avoid certain activities and/or alter their employment to avoid worsening the pain. Such precautions may reduce the occurrence of pain and medication prescribed by a doctor may help to combat some of the discomfort. There is, however, no cure.

## Kidney Function

Due to the accumulation of waste products in kidney cells and in the wall of blood vessels supplying the kidney, kidney function may become impaired over time. Those affected by Fabry Disease may have symptoms of reduced kidney function by early adulthood (For example, too much protein in their urine).

Without enzyme replacement therapy (ERT) this will continue to worsen over the years and may become so severe that dialysis or kidney transplantation is required. (see the treatment section in the fact sheet). Studies demonstrate that kidney function can be affected early in life, even during childhood in males and females with Fabry Disease.

## Heart

Accumulation of waste products within the cells of the heart or the walls of the coronary arteries may cause individuals with Fabry Disease to develop heart problems. Initial symptoms may be an irregular or fast heartbeat, but over time, this can develop into more serious complications leading to an enlarged heart, chest pain (angina), increased risk of heart attack and heart failure.

Treatment for heart conditions associated with Fabry Disease may include medication and/or the fitting of a pacemaker. For more severe conditions heart bypass surgery and transplantation may lead to some improvement.

## Stroke

Individuals with Fabry Disease may develop a stroke. This may be a minor stroke followed by a full recovery (Transient Ischaemic Attacks or TIA's), but more severe strokes can occur. Medication to thin the blood may be prescribed by a doctor.

## The Bowel

Individuals with Fabry Disease may experience discomfort and pain after eating due to the accumulation of waste products within the cells of the intestine or the blood vessels and nerves supplying the intestine. Other possible symptoms include vomiting, nausea and diarrhoea.

Although these symptoms can be reduced by eating small meals regularly, adjusting the diet and seeking medication prescribed by a doctor, some individuals with Fabry Disease may experience weight loss and become quite thin.



Alan

## Skin

Small, dark red spots (known as angiokeratoma) often appear on the abdomen, groin, buttocks and thighs of individuals with Fabry Disease. They often appear during late childhood and could be one of the first signs of Fabry Disease.

Individuals with Fabry Disease also experience reduced sweating (hypohidrosis). Some individuals do not sweat at all (Anhidrosis). As a result, sufferers may experience fevers and find it difficult to exercise or cope with hot temperatures. Some patients may have increased sweating (Hyperhidrosis). This is more common in females.

## Eyes

Slight abnormalities are present in the eyes of individuals with Fabry Disease, but they do not affect sight.

## Nervous System

Individuals with Fabry Disease may suffer from headaches, vertigo and a ringing sound in the ears (Tinnitus).

## Development of Fabry Disease

There are differences between Fabry Disease in childhood, adolescence and adult life.

It should be noted that not all individuals with Fabry Disease will experience all of the symptoms outlined below and that some adults with the disease will experience some of the symptoms associated with the disease in childhood and vice versa.

## Children

Pain and skin problems (such as angiokeratoma), characteristic abnormalities of the eye, gastrointestinal symptoms (such as alternating bouts of diarrhoea and constipation), tummy pains and hearing problems are usually the only symptoms of Fabry Disease associated with childhood. Furthermore, the pain is often attributed to common growing pains. After diagnosis it is essential that children are given support in understanding their disease and any limitations it may impose, whilst still being encouraged to participate in activities and normal daily living with their peers.



Abby & Paige



Laura

## Adolescents

During adolescence, many of the skin problems experienced during childhood may worsen. This is often the time when bowel pain begins. The first stages of impaired functioning of the kidney and heart can also occur at this time.

## Adults

Kidney problems in males with Fabry Disease usually become much more severe during early adulthood. The heart becomes affected after the age of about 40 years and the risk of strokes increases. In females, end stage kidney disease is rare, despite a large number excreting a significant amount of protein in their urine, indicative of kidney problems.

## Atypical Variants

It has recently been reported that some patients with Fabry Disease present with features only involving a single organ system, specifically cardiac and renal. These arise as a result of mis-sense mutations when the patient has sufficient residual enzyme activity to prevent symptoms in childhood and early adult life.

However, the existence of atypical variants is still the subject of medical debate. Patients that have been diagnosed with an atypical variant may actually have changes in other organs.

Such patients may well have mutations associated with residual enzyme activity which could, in turn, be associated with a less severe presentation of the disease.



Margaret & Family

## Living with Fabry Disease

Fabry Disease is a debilitating condition associated with many problems and shortened life expectancy. With the development of Enzyme Replacement Therapy (ERT), however, many sufferers are able to live fulfilled lives. Alleviated symptoms often enable adults with Fabry Disease to participate in family life, sustain relationships and seek employment, whilst children affected by the disease can enjoy social and physical activities with their peers.

Many adults with Fabry Disease are aware that the condition runs in their family and have often prematurely experienced the loss of a parent through the disease. In a significant number of cases, Fabry Disease has gone undiagnosed in children and is simply dismissed as 'growing pains'. Upon diagnosis, sufferers are often relieved to find the cause of their unusual childhood symptoms of burning hands and feet, bowel problems, tiredness and the inability to sweat.

An individual's diagnosis of this hereditary condition may lead to diagnosis in several members of the same family and can raise issues of pre-natal diagnosis and genetic counselling for those considering having children.

Those individuals who receive treatment and whose symptoms have been alleviated can have a greatly enhanced quality of life. They feel able to participate in the general activities of daily life which, in turn, can have a positive effect on relationships and the family, without being so restricted by the debilitating symptoms and emotional burden of living with this disease.

## Insights from Fabry Sufferers

*"I didn't know that I was ill. I wouldn't admit that there was anything wrong with me."*

*"When I had the diagnosis I was distraught. It took me a long time to accept it. I think some people have to grow emotionally into dealing with it."*

*"When I received my diagnosis everything fell into place. It was like a jigsaw. All the pieces suddenly just all started to click together."*

*"When I was growing up my progress in all areas of life was always behind everyone else's. I went year after year having doctors tell me that there was nothing wrong with me. To finally have confirmation that I was ill was like a burden being lifted. At least I have a name for it now."*

*"I started out viewing my Fabry Diagnosis as a huge intrusion in my life. I couldn't spare the time to be ill, but I decided I was going to have to find a way to control the disease because I definitely wasn't going to allow the disease to control me."*

*"I can't plan holidays and activities far in advance in the same way as others can. No two days are ever really the same so I have learned that it is all about having to live each day as it comes."*

*"It may take me a lot longer than other people to achieve certain goals in life but once I have achieved what I set out to do the sense of accomplishment is overwhelming. Keeping my goals in mind helps me to get through some of the more difficult periods."*

*"Looking back, the only advice I would give to those who have a positive diagnosis would be to try and not panic. Do some research on the condition and don't listen to anyone but yourself."*

*"Everyone copes with things in their own unique way so don't allow anyone to become a negative influence in your life. Feed from the positives and try to ignore everyone else."*

*"In my mind it's all about education. The more you can learn from medical literature, specialist doctors and even fellow sufferers, the better equipped you will be when it comes to getting an idea of the best coping strategies to implement day to day."*

*"Dealing with Fabry Disease is a personal, ongoing battle. Each and every one of us has a personal journey to take. It's true of those suffering with Fabry and with life in general."*

*"You might have to try many different coping strategies to find the one that works for you but once you have established what works, chances are you can eventually overcome most things."*



Claire

## My Story, by Darrin Minett

*It had been an average sort of summer's day and I had just finished work. As I cycled home, I could feel my mobile vibrating in my pocket but as I was riding along a very congested busy road, I decided to ignore it until later.*

*On arriving home I checked my phone and realised my brother had rang, so I rang him back straight away.*

*When I got through he told me that he had something to tell me but that he wouldn't do it over the phone, so we arranged for him to pop round the following day for a chat.*

*When he turned up the next day, he instantly reminded me about the illness that he had suffered with back in February, earlier that year. I knew he had been diagnosed with Pneumonia at the time, but we'd never spoken about it.*

*He then proceeded to tell me that the doctor at the local hospital who had been dealing with him had noticed several symptoms that required further investigation. The doctor said that he believed that my brother had underlying health problems that may be connected to a rare disease. He was then sent to have tests.*

*He then told me he had been diagnosed with Fabry Disease and then proceeded to tell me about his own medical history. It was probably the first time in twenty years that he had told me anything personal as we are both very private about our lives.*

*None of what my brother was telling me had any resemblance to my own history other than the pains in the hands and feet that we both experienced when we were kids. He then told me that he had (probably) inherited it from our mum and that the chances that I had it were very high. I was advised to inform my doctor and to ask to be referred to a specialist doctor to have the same simple tests that my brother had undergone, just to be safe.*



Darrin

*Before my brother left he told me not to read about Fabry Disease on the internet, as the specialist had advised him against it.*

*As soon as my brother left, however, I ignored this advice. I found a medical page and read about some of the symptoms. Then things started to register as I started to recall my own medical history.*

*When I read about acroparasthesia, I finally knew I had found a possible answer to the pains in my hands and feet when I was younger. This often occurred when I did anything sporty, but it never stopped me. I played football daily, I ran for the school in the cross country team and everything I did as a child involved 'sports like' activities. I ignored the pains and became almost ignorant of them. I actually believed they were 'growing pains', as my doctor once told me they were those mythical ailments we suffer as children.*

*These pains often occurred just before the onset of damp weather. 'It'll rain in an hour' I used to say to people. Then in my late teens these pains almost disappeared and I no longer had any problems when doing physical activities. These pains, however, would always reappear whenever I had any illnesses. They seemed to make every illness follow the same pattern. It felt like my hands and feet were on fire and it would spread up my arms and legs and I would be in agony. This is when I established that I may have been having 'Fabry crises' for over twenty five years. The evidence was beginning to stack up. I haven't eaten or drunk any dairy products for over twenty years, as these caused upset stomachs and migraines in my late teens, so I removed them from my diet. The migraines stopped straight away and I've never had one since. I still had minor digestive problems which were getting worse, although I'd never admit it.*

*In my late twenties I had tests done to deal with this problem, but the doctors found nothing. Another problem arose during the tests when I had to have a barium drink so I could have my intestines and bowels X-rayed. This drink sat in my stomach for just under four hours. The doctor wasn't impressed; he couldn't understand why it sat there for that length of time and actually shouted at me asking 'what's wrong with you?'... "You tell me pal, that's why I'm here", I recall thinking.*

*The only other time I had been properly ill was back in 1998 when I contracted Chicken Pox. I caught it off my kids. I'd never had it as a child and while my kids were both back to full health in two to three days, I was very ill for just under five weeks. Since then, however, I've been in very good health.*

*Then in 2005 I was made redundant from my company, who were relocating to China. The job role that I took on after this was as a gas engineer based at a company on Merseyside. Sadly, doing work with vibrating tools had a massive effect on me. During the night I had very poor blood circulation in my fingers, hands and arms.*

*It got so bad that I couldn't lift myself up to try and let the blood flow back down my arms and eventually, due to sleep disruption and being unable to feel anything in my hands, I was medically finished.*

*I was diagnosed with Reynaud's Syndrome, which is extremely annoying, especially in my current job as a postman. My hands and feet react to the weather conditions, even when I wear waterproof, windproof, double insulated ski gloves. I have to put my hands into very warm water when I get in, to help get the blood flowing.*

*Although I was still very fit, (I attend a gym four days a week, as well as doing my job) I had noticed a slight deterioration in my energy levels, but I put that down to my diet and my age. On a couple of occasions I had palpitations, which did not go unnoticed. I know that most people have irregular heartbeats but these palpitations made me slow down. I stopped doing my job at one hundred miles per hour and took it steadier in the gym.*

*I had no need to rush about like I did, but I knew I had to keep an eye on it. Also, my digestive problems seemed to be getting worse, but like I say, I tolerated it. My own doctor thankfully, referred me to Manchester and the rest is history. I had no visible symptoms like my brother, but I do have minor heart problems, (i.e. LV Hypertrophy, Bradycardia), Tinnitus, and poor blood circulation in my hands and feet.*

*Since having my infusions at home, (by the wife, I must add), my digestive problems have almost ceased and there has been a welcome improvement in all the symptoms I have mentioned. My brain scan showed minor activity (yes, something is occurring inside, but not much, just the odd tumble-weed rolling by) but hopefully nothing too serious.*

*My sister has been tested all clear, which is great news. My daughter however, has the condition, but as yet she has no ailments. A search into the family history found that I could have ended up like my great grandma, who died aged forty. My grandmother also died aged forty and my uncle died at 54 from heart and kidney problems.*

*Although it was not diagnosed on their deaths, the problems they had indicates possible Fabry Disease. My mum as yet, has not been tested. At 65 I don't know whether it would be of any benefit to her. I'm not sure that she's interested about finding out. It's bad enough for her to think that she has been responsible for our illnesses, even though we don't see it that way.*

*The ERT has given me cause for optimism as my results show a reversal in some conditions, so hopefully this will continue. I have switched from Fabrazyme to Replagal, like most UK patients and I am still not sure which I like best.*

*My initial ERT with Fabrazyme gave me random side effects, which obviously diminished over time. The switch to Replagal was comfortable with only minor side effects. Time will tell. Luckily, Royal Mail have been fantastic about my illness, especially as it is covered by the Disability Discrimination Act.*

*Overall I consider myself very lucky, especially compared to some of the other Fabry patients I recently met at the German Morbus Fabry Patients Conference held in the beautiful city of Vienna. This condition affects everybody differently and I am not going to get complacent about my health. Every little shot of pain or palpitation is noted.*

*I'd like to wish everyone who has Fabry Disease all the best of luck and I'd like to thank the MPS Society for giving me opportunity to tell you all this.*

## **Treatment of Fabry Disease**

### **Enzyme Replacement Therapy (ERT)**

Enzyme Replacement Therapy (ERT) for Fabry Disease uses a recombinant form of the enzyme that is missing or malfunctioning in individuals with Fabry Disease. The enzyme is manufactured from cultures of cells which have been genetically engineered to express the human enzyme. This enzyme is then administered via repeated intravenous infusion to the patient, usually every two weeks.

The principle underlying this treatment is that the enzyme that is missing or not functional may be partially replaced by infusing this therapeutic form of the enzyme. The enzyme used in treatment contains a particular chemical address on its surface that allows it to be taken up by cells from the blood and transferred to the lysosome of the cell where it carries out its work in breaking down the storage material Gb3 (GL3). The aim of the treatment is to help relieve symptoms and to prevent progression of damage to important organs like the kidney and the heart.

Criteria have been devised for the initiation of therapy and are based on the responsiveness of various organs to treatment. In the UK, criteria are based around changes in the heart, brain and kidneys or pain and gastro-intestinal symptoms which affect quality of life.

Fabry Disease tends to have a different course in males than in females and the criteria for initiating therapy is different between the sexes. In males, treatment would be expected to commence as soon as the diagnosis is made which, when genetic screening is done, may be prior to symptoms and signs appearing.



Laura



Ian

In females, not everyone who carries a mutation in the  $\alpha$ -galactosidase gene will go on to develop symptoms of the disease so a positive genetic test on its own is not an indication for ERT. Instead, the decision to commence ERT in a woman is based on whether she has symptoms or evidence of significant tissue damage as a result of Fabry Disease. Distressing symptoms can start at an early age and treatment is available for children in co-ordination with a Paediatrician.

Studies have shown that ERT can greatly benefit individuals with Fabry Disease by reducing pain, stabilise or improve kidney function, stabilise or improve cardiac abnormalities and improve quality of life. It is hoped that by treating people with Fabry Disease as early as possible it will be feasible to prevent the clinical manifestations of the disease.

## Enzyme Replacement Therapy (Chaperone Treatment)

Some people with Fabry Disease make an abnormal  $\alpha$ -galactosidase enzyme which is unable to function properly in the cell. The aim of chaperone therapy, or enzyme enhancement therapy, is to find medicines which will stabilise these abnormal enzymes and allow them to function well enough to prevent lysosomal storage in cells.

Studies are ongoing in the UK into a new treatment for Fabry Disease which involves an oral therapy in tablet form. This contains a small molecule known as a pharmacological chaperone which binds the affected protein, stabilising it and allowing it to travel properly throughout the cell and function within the lysosome. Phase two studies to determine the safety of this drug have been completed. Phase three studies to determine how effective the treatment is are now underway in various international sites including two in the UK. If these clinical trials are successful, it is possible that this oral treatment may be another option for some people with Fabry Disease in the future.

If you would like more information on treatment options and clinical trials then please contact your Fabry specialist or the MPS Society.

Clinical guidelines for Fabry Disease can be found on the national commissioning website, [www.ncg.nhs.uk](http://www.ncg.nhs.uk).

## Further Information

The MPS Society produces a range of other publications that explain, in simple terms, the pattern of inheritance of Fabry Disease. These information resources also introduce some of the key issues that will affect individuals affected by Fabry Disease, their relatives and any children that may be born into the family. Please contact the MPS Society for more information or visit our website [www.mpsociety.co.uk](http://www.mpsociety.co.uk).

## About the MPS Society

The Society for Mucopolysaccharide Diseases (MPS Society) is a voluntary support group founded in 1982 which represents over 1200 children and adults suffering from MPS and related diseases including Fabry, their families, carers and professionals throughout the UK.

It is a registered charity entirely supported by voluntary donations and fundraising and is managed by the members themselves.

The Society produces a range of publications of which include: a quarterly MPS Magazine, a Fundraising Newsletter, Fact Sheets and various literature for children, adults, parents, siblings and individuals that are affected by MPS or a related disease.

Regional clinics, information days and conferences are arranged throughout the United Kingdom.

The MPS Society provides a range of services including: Advocacy Support to individuals suffering from MPS and related diseases, their families and carers; Ongoing Advocacy support for affected families following bereavement; telephone support including an out of hours helpline; a 'Befrienders Scheme' putting individuals suffering from MPS and their families in touch with each other for mutual support; Financial Assistance Scheme: Funds research and treatment into MPS and related diseases.

Membership to the MPS Society is open to all individual residents in the UK who meet the agreed criteria. Other individuals wishing to belong to the UK MPS Society can become 'Friends of MPS'.

The Society exists entirely on donations and fundraising from its members and the general public.

For further information about the work of the MPS Society and the service we provide please contact us.

Please note that this fact sheet is not intended to replace medical advice or care.

The Society has received unrestricted grants from a number of pharmaceutical companies with an interest in Lysosomal Storage Disorders for a range of projects.

Society for Mucopolysaccharide Diseases

MPS House, Repton Place, White Lion Road, Amersham, Buckinghamshire, HP7 9LP  
T: 0845 389 9901, F: 0845 389 9902, E: [mps@mpsociety.co.uk](mailto:mps@mpsociety.co.uk), [www.mpsociety.co.uk](http://www.mpsociety.co.uk)  
Registered Charity No. 287034, Charity Registered in Scotland SCO41012