WHAT ARE THE SYMPTOMS?

Fabry disease is a complex condition; each person can be affected by it in a different way and can therefore experience varying degrees and combinations of symptoms. The most common signs and symptoms are listed below.

**Pain and burning sensation in hands and feet**

**Small raised dark red spots (called angiokeratomas) on the skin**

**Impaired ability to sweat, which in turn causes frequent fevers and a low tolerance to heat or exercise**

**Stomach problems, such as pain, nausea, vomiting or diarrhoea**

**Hearing problems, such as ringing in the ear or hearing loss**

**Serious problems affecting the heart, kidney and nervous system can typically be seen in older patients as the disease advances**

**Kidney Disease**

**Small raised dark red spots (called angiokeratomas) on the skin**

**Changes in the eye, such as clouding of the cornea (which typically does not affect vision)**

**Depression and fatigue**

**Stomach problems, such as pain, nausea, vomiting or diarrhoea**

**Hearing problems, such as ringing in the ear or hearing loss**

**Serious problems affecting the heart, kidney and nervous system can typically be seen in older patients as the disease advances**

**Changes in the eye, such as clouding of the cornea (which typically does not affect vision)**

When patients present with signs and symptoms of serious or unexplained kidney conditions, it can be an indicator of more complex underlying causes, one of which can be Fabry disease.

This leaflet aims to answer some of the questions you may have about the causes, inheritance and treatment of Fabry disease, and offers recommendations on what to do if you have been diagnosed with the condition.
KIDNEY DISEASE IN PATIENTS WITH FABRY DISEASE

In the general population, serious kidney problems (such as kidney cysts, proteinuria or reduced renal function, which may eventually lead to dialysis) are more characteristic of an elderly population. In Fabry disease, however, more than 80% of patients suffer from some degree of kidney impairment; this typically becomes apparent in their mid-30s and can occur as early as childhood. Therefore, if unexplained kidney symptoms are present in a younger patient, and/or there is family history of early serious kidney conditions, then screening for Fabry disease is recommended.

WHAT IS THE TREATMENT FOR FABRY DISEASE?

If left untreated, Fabry disease can eventually cause life-threatening organ damage, such as end-stage kidney disease, heart failure or stroke. Therefore, it is vital that patients are tested and diagnosed as early as possible.

Your renal physician (nephrologist) will continue treatment for your kidney disease and its related symptoms; however, your Fabry disease specialist will be responsible for treating you to prevent further Fabry disease manifestations.

Nowadays, there is effective treatment available to help manage Fabry disease. Enzyme replacement therapy (ERT) is designed to replace the deficient enzyme. In most cases, ERT is combined with other treatments for the management of specific symptoms, which are tailored to the individual. Some patients who carry a specific Fabry mutation may be eligible for a treatment known as ‘chaperone therapy’, which supports delivery of the enzyme to the correct location in the cells.

Please ask your doctor to find out more about these treatments.

WHAT SHOULD I DO IF I HAVE BEEN DIAGNOSED WITH FABRY DISEASE?

Your renal physician and/or family doctor will be able to advise you on how best to manage your condition and refer you to a Fabry disease specialist, who will assess your condition and prescribe the appropriate treatment.

You may also be assigned a genetic counsellor, who will discuss the inherited nature of the disease and the associated issues with you.

HOW IS FABRY DISEASE DIAGNOSED?

Fabry disease can be very difficult to distinguish from more common conditions due to its wide range of symptoms, and patients can stay without a correct diagnosis for years. However, once it is recognised that a patient may have Fabry disease, the actual diagnosis can be performed accurately with a simple blood test.

Due to the hereditary nature of Fabry disease, a diagnosis not only has important health consequences for you, but may also have implications for your family. For each newly identified Fabry disease patient, several additional cases may be present in the extended family. It is therefore important, if you are diagnosed, that you seek advice from your doctor or genetic counsellor about family planning, genetic screening and other issues.

HOW DOES FABRY DISEASE RUN IN THE FAMILY?

Fabry disease is not contagious. It is a genetic condition that children can inherit from their parents.

Every cell in the human body contains chromosomes, which are thread-like structures carrying genetic information. Females have two X chromosomes in each cell and males have one X and one Y chromosome. The faulty gene causing Fabry disease is located on the X chromosome only.

Men carrying this gene are always affected by the disease, whereas women may not have symptoms, or their symptoms may be less severe or appear later in life.

If you are diagnosed with Fabry disease, your doctor or genetic counsellor will perform a family history evaluation (also called ‘pedigree analysis’) with you, to determine the risks of your family members having the condition.