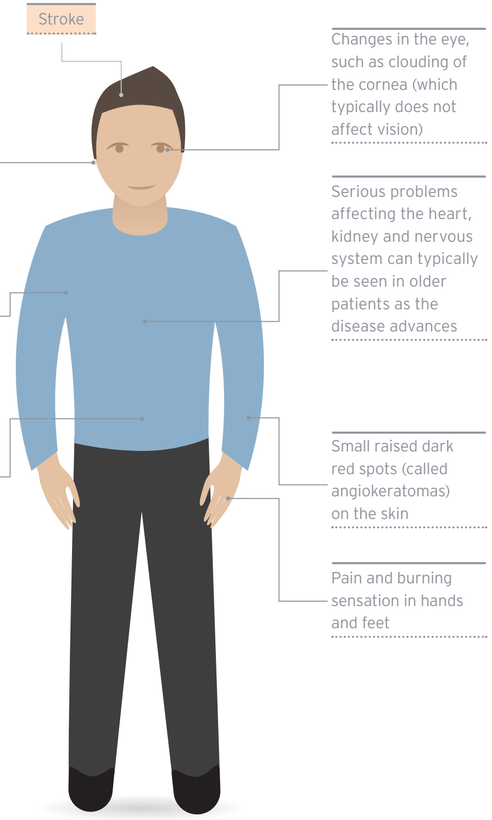
WHAT ARE THE SYMPTOMS?

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



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FABRY DISEASE
IN STROKE PATIENTS

INFORMATION LEAFLET

You have been given this leaflet because your doctor suspects that your stroke may be linked to Fabry disease, which is a rare, serious, but treatable condition.

A stroke is caused by a sudden shortage of blood supply to the brain. It typically occurs in the older population, but when it takes place in younger patients, it could be an indicator of more complex underlying causes, one of which is 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.

Prepared and funded by Shire.
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WHAT IS FABRY DISEASE?

Fabry disease (or Anderson-Fabry disease) is a rare, inherited disorder caused by a mutation in the gene that controls an essential enzyme in the body's cells. This enzyme metabolizes lipids, which are fat-like substances such as oils, waxes, and fatty acids.

The gene mutation causes either an absence in or reduced activity of the enzyme in patients. This results in a build-up of lipids in the cells of the organs, which eventually interferes with their normal functions. This leads to progressive damage to the body, resulting in a wide range of symptoms and complications. Stroke can occur either alone or in combination with other symptoms. Due to the risk of a recurrent stroke or other complications in patients with Fabry disease, early diagnosis and treatment are essential.

STROKE IN PATIENTS WITH FABRY DISEASE

Stroke causes vary significantly depending on the age of the patient; it is considered to be a disease of the older population. Therefore, special attention must be paid when stroke affects young people.

While stroke is common in young patients with Fabry disease, it can also occur in older patients with Fabry disease. It is often a first manifestation of Fabry disease and can result from a loss of blood supply to a critical part of the brain due to direct changes, or changes to the heart.

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.

WHAT IS THE TREATMENT FOR FABRY DISEASE?

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

Your stroke physician will continue your treatment for stroke and its related symptoms; however, your Fabry disease specialist will be responsible for treating you for the underlying cause of the stroke.

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 stroke 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.

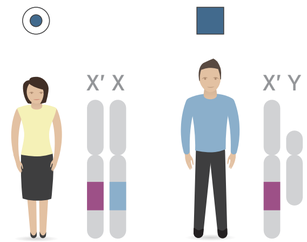
Due to the hereditary nature of Fabry disease, a diagnosis not only has important health consequences for you, but may also have implications for your family. For each newly identified Fabry disease patient, several additional cases may be present in the extended family. It is therefore important, if 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.



Female and male with the affected gene in X chromosome

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.