For patients

Fabry disease and stroke

Let's inspire a better future in Fabry

Takeda

A rare genetic condition called Fabry disease may be linked to stroke. This leaflet aims to answer some of the questions you may have about Fabry disease.

This leaflet does not replace your doctor's advice. Please speak to someone from your healthcare team if you have any concerns or want to know more.

What is Fabry disease?

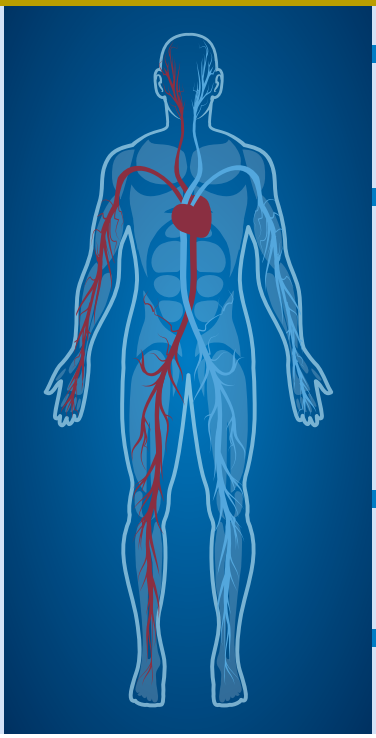
Fabry disease (also called Anderson-Fabry disease) is a rare inherited disorder affecting around 1 in 40,000 males and 1 in 20,000 females, although reported numbers vary by geographic region. In patients with Fabry disease, a change in a gene means that an enzyme called α-galactosidase A (α-Gal A) either is absent or does not work well. Because of this, the enzyme is unable to break down fatty substances called glycosphingolipids. These then build up in cells, which causes progressive damage to the body and a wide range of symptoms.

What are the symptoms?

Fabry disease is a complex condition and people can be affected in different ways; every person may experience different symptoms of varying severity.

Common signs and symptoms of Fabry disease (not everyone has all of these)

- **Hearing problems such as tinnitus (ringing in the ears) or hearing loss**
- **Impaired ability to sweat, which in turn causes frequent fevers and a low tolerance to heat or exercise**
- **Gastrointestinal or gut problems such as pain, nausea, vomiting or diarrhoea**
- **Depression and fatigue**



- **Changes in the eye, which usually do not affect vision, may be detected by a doctor**
- **More serious problems affecting the heart, kidney and brain (stroke and transient ischaemic attack, or 'mini-stroke') can typically be seen in adults as the disease advances**
- **Angiokeratomas (small, raised, dark red spots) on the skin**
- **Pain and burning sensation in hands and feet**

How may Fabry disease be linked to my stroke?

A stroke happens when the blood supply to part of the brain is cut off. Fabry disease can cause changes in blood vessels and/or the heart which result in a loss of blood supply to part of the brain.

Stroke is often a first symptom of Fabry disease. Stroke can happen in both young and older people with Fabry disease.

How can my doctor diagnose Fabry disease?

Fabry disease can be very difficult to distinguish from more common conditions owing to the wide range of symptoms, and some patients spend many years without a correct diagnosis.

Once your doctor suspects that you might have Fabry disease, a simple dried blood spot test can be used to screen for the disease, to measure the activity of the enzyme, and a genetic test to confirm the presence of a faulty gene.

What is the treatment for Fabry disease?

Fabry disease currently has no cure. However, there are treatments that can help with progression of the disease.

The two approved types of Fabry disease-specific treatment are described below.

Enzyme replacement therapy (ERT)	In people with Fabry disease the α-Gal A enzyme either is absent or does not function (or work) properly. In ERT, people with Fabry disease are provided with a working version of the enzyme.
Chaperone therapy (CT)	In Fabry disease the α-Gal A enzyme may be present but not working properly. CT can support the enzyme and help it to work better. However, not all patients can receive CT – only those with certain gene changes.

Complementary treatments that are not specific to Fabry disease are also available; these can help to manage the symptoms of Fabry disease. If you have a stroke, you may be given treatment to reduce the risk of it happening again (prophylaxis).

Speak to your doctor about the treatment options that may be appropriate for you and their possible effects.

Because of the hereditary nature of Fabry disease, a diagnosis not only has important health consequences for you, but also may have implications for your family. If you are diagnosed with Fabry disease, your doctor or healthcare professional will perform a family history evaluation (also called 'family tree analysis') with you, to determine the risk of your family members having the condition.

Where can people get information and advice?

For anyone concerned about the risk of Fabry disease, there are various sources of information and people they can speak to.

Healthcare professionals will be able to provide you with information about Fabry disease and how it is diagnosed and treated.

A **genetic counsellor** is a specially trained professional who works with people affected by a genetic disorder and their family members. Their goal is to provide these people with balanced information and support so that they can make informed choices about their own health and make life plans.

In addition, patient groups such as the Fabry International Network can provide information and support. Further details can be found at **http://www.fabrynetwork.org/**

www.fabrydisease.info is also useful, with experiences from people who have discussed Fabry disease with their family, plus further information about the condition.

The images used within this document are representative only and the models shown are not associated with Fabry disease in any way.

C-ANPROM/INT//4763 | Date of preparation: March 2019