Heart disease
Other symptoms too?

FABRY DISEASE
IN PATIENTS WITH UNEXPLAINED HEART CONDITIONS

You have been given this brochure because your heart condition may be linked to Fabry disease, which is a rare, potentially serious, genetic disorder.¹²

When patients present with signs and symptoms of serious or unexplained heart conditions, it can be an indicator of more complex underlying causes, one of which could be Fabry disease.³⁴

This brochure aims to answer some of the questions you may have about the causes, inheritance and management of Fabry disease.

www.fabrydisease.info
WHAT IS FABRY DISEASE?

Fabry disease (or Anderson-Fabry disease) is a rare inherited disorder caused by the lack of activity of an enzyme called alpha-galactosidase A, as the result of a gene change (mutation). This enzyme is responsible for breaking down a fatty substance, known as globotriaosylceramide (Gb₃), in the cells of the body. The build-up of Gb₃ deposits in the cells interferes with their normal functions, leading to progressive damage to the body and resulting in a wide range of symptoms and complications. Heart disease can arise as the result of Gb₃ deposits, in the presence or absence of other symptoms.

HOW IS FABRY DISEASE LINKED TO MY HEART CONDITION?

Heart problems are common in Fabry disease and may become progressively worse with age. As Gb₃ accumulates within the cells of the heart or the walls of the blood vessels that supply the heart, damage may occur. Heart problems due to Fabry disease may include heart enlargement; poorly functioning heart valves; irregular heartbeat; heart attack; or heart failure.

HOW CAN MY DOCTOR DIAGNOSE FABRY DISEASE?

Fabry disease can be very difficult to distinguish from more common conditions due to the wide range of symptoms, and some patients spend many years without a correct diagnosis. However, once your doctor suspects that you might have Fabry disease, a diagnosis can be made with a simple blood test.
WHAT ARE THE SYMPTOMS?

Fabry disease is a complex condition and 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 include:

- Pain and burning sensation in hands and feet
- Small raised dark red spots (called angiokeratomas) on your 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
- Depression and fatigue
- Changes in the eye such as clouding of the cornea
- Hearing problems such as ringing in the ear or hearing loss
- Heart Disease
  - More serious problems affecting the heart, kidney and nervous system can typically be seen in older patients as the disease advances
  - Small raised dark red spots (called angiokeratomas) on your skin
  - Pain and burning sensation in hands and feet
- Depression and fatigue
- Hearing problems such as ringing in the ear or hearing loss
- Stomach problems such as pain, nausea, vomiting or diarrhoea
- Depression and fatigue

HOW CAN I MANAGE FABRY DISEASE?

Your heart doctor (cardiologist) will continue your treatment for your heart problems and its related symptoms.

Fabry disease is not a curable condition, however, treatments are available to help manage the disease, if appropriate for you. Please consult your doctor or healthcare professional for further information.

*typically not affecting vision
**HOW DOES FABRY DISEASE RUN IN THE FAMILY?**

Fabry disease is not contagious, but it is a hereditary condition that children can genetically inherit from their parents, in a pattern illustrated below.

Every cell in the human body contains chromosomes, which are thread-like structures carrying genetic information. Females have two X chromosomes in each cell (an X from each parent), and males have one X and one Y chromosome.

The faulty gene causing Fabry disease is located on the X chromosome only. This means that 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.

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. If you are diagnosed with Fabry disease, your doctor or healthcare professional will perform a family history evaluation (also called ‘Pedigree analysis’) with you, to determine the risk of your family members having the condition.
References


