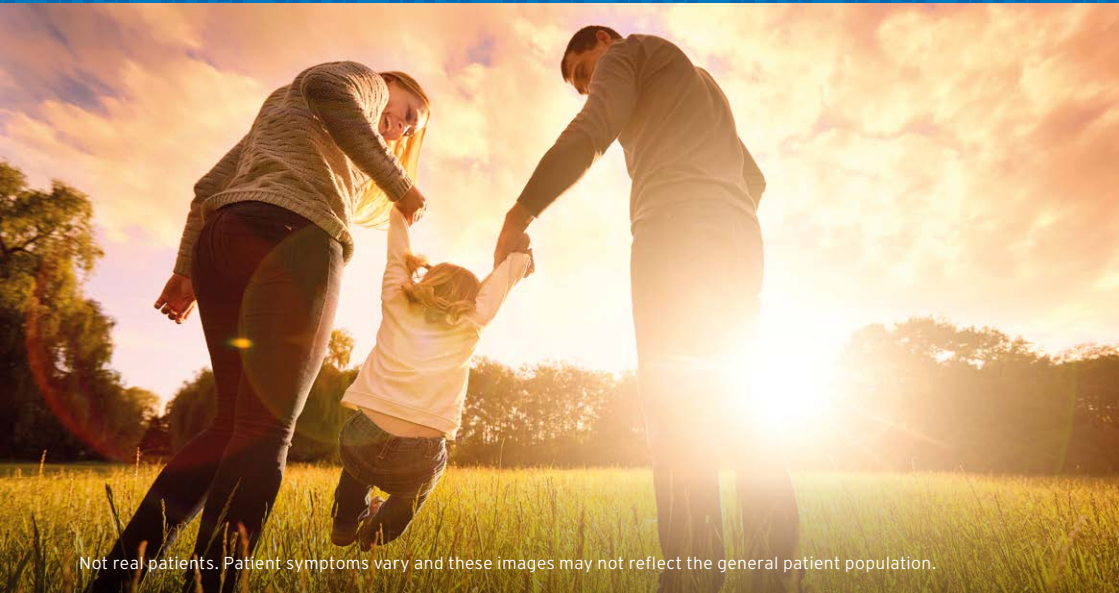


# Fabry disease: what it means for our family



Not real patients. Patient symptoms vary and these images may not reflect the general patient population.

*Let's inspire a better future in Fabry*



This leaflet has been written for people who have a family member who has been diagnosed with Fabry disease. It aims to answer some of the common questions they may have about Fabry disease, including determining their own risk of having the disease, diagnosis and treatment.

## What is Fabry disease?

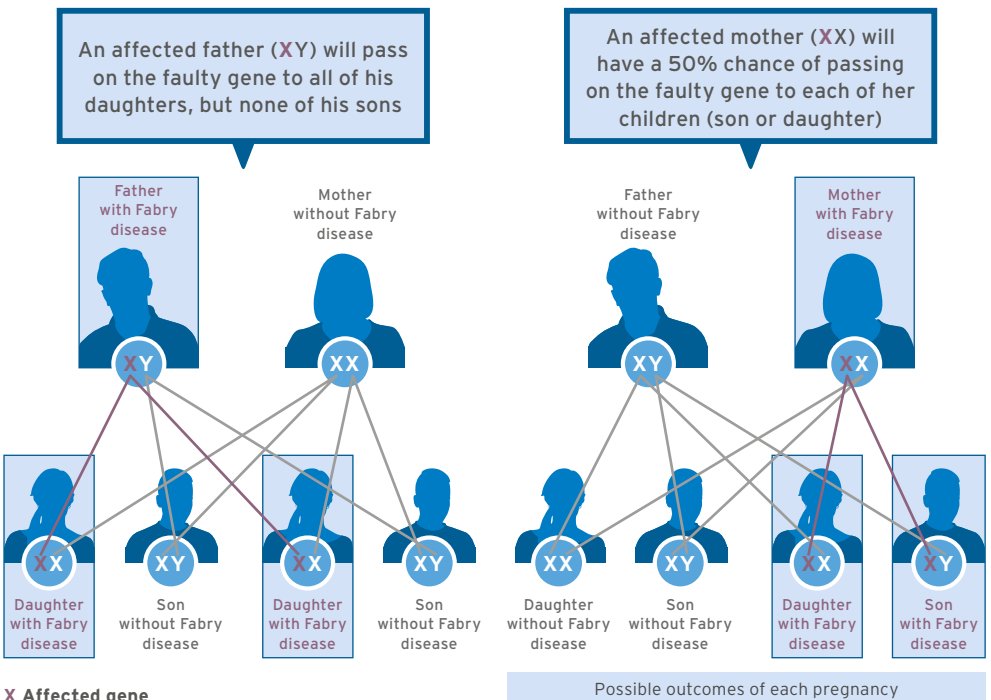
Fabry disease (also called Anderson-Fabry disease) is a rare inherited disorder affecting around 1 in 40,000 males, and females can also be affected.<sup>1-3</sup> However, reported numbers vary by geographic region.<sup>1,4-10</sup> In patients with Fabry disease, a genetic change means that an enzyme called  $\alpha$ -galactosidase A ( $\alpha$ -Gal A)

either is absent or does not work well.<sup>1-3</sup> Because of this, the enzyme is unable to break down fatty substances called glycosphingolipids.<sup>1-3</sup> These then build up in cells, which can cause damage to the body over time and a wide range of symptoms.<sup>1-3</sup>

## How do people inherit Fabry disease?

Fabry disease is not contagious, but it may be passed on in families through our genetic material (two X chromosomes [XX] for females, and an X and Y chromosome [XY] for males).<sup>2,11</sup>

The gene that causes Fabry disease is present on the X chromosome and is passed down from parents to their children, in what is known as X-linked inheritance, as illustrated in the diagram below.<sup>2,3,12</sup>



# What are the symptoms of Fabry disease?

Fabry disease is a complex condition and people can be affected in different ways; every person may experience different symptoms of varying severity.<sup>13</sup> In boys the first symptoms may appear at under 10 years of age, but the age of

symptom onset varies in girls and from person to person, even within the same family.<sup>13,14</sup> Some people do not have any symptoms until later in life, as adults.<sup>13</sup>

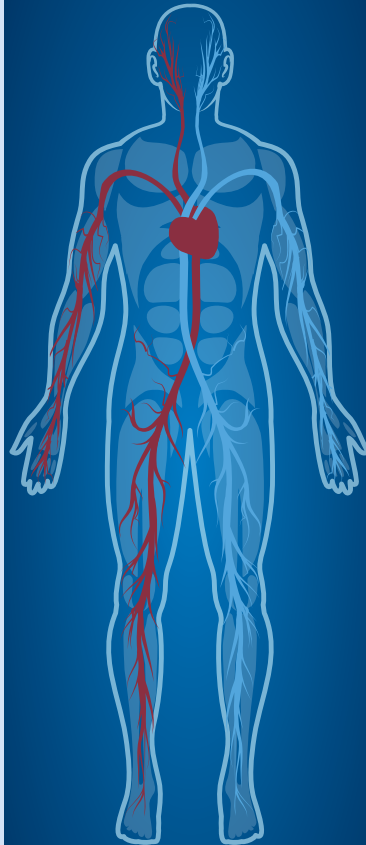
## Common signs and symptoms of Fabry disease (not everyone has all of these)<sup>13,15</sup>

**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**

# What is family tree analysis and how can it help?

If a person has been diagnosed with Fabry disease, other members of their family may be at risk of having the disease as well.<sup>13</sup> By looking at the person's family members and family history, a doctor and/or genetic counsellor can map out a family tree.<sup>13,16</sup> (See 'Where can people get information and advice?' on the back page.) This will help to identify family members who may be at risk.<sup>14,17</sup> An example family tree is shown in the diagram below.

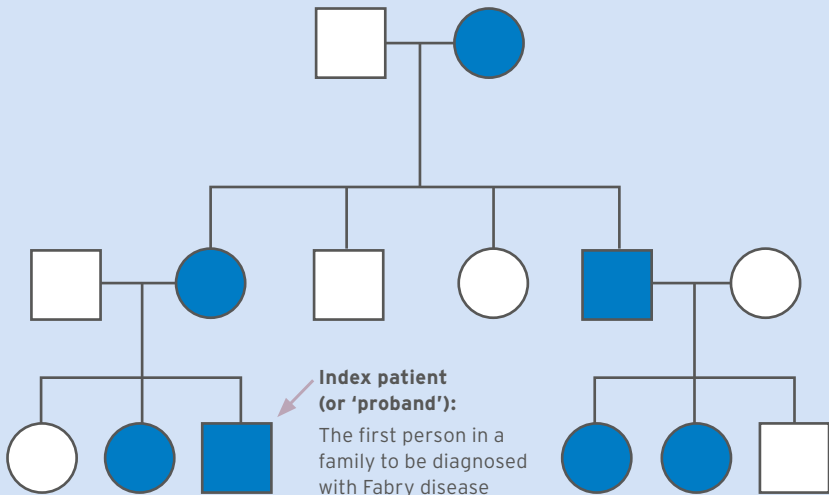
If a family member is found to be at risk, then it is important that they consider being tested for the disease, even if they have no symptoms, because symptoms may develop later in life.<sup>14,17</sup>

Genetic counsellors can provide information regarding testing, so that at-risk family members can decide what to do.<sup>16,18</sup>

Once a decision has been made to be tested, a simple dried blood spot test can be used to screen for Fabry disease.<sup>19,20</sup> If this is positive, then a blood test can be done to measure the activity of the enzyme, and a DNA test to confirm the faulty gene.<sup>21</sup>



## Example of a Fabry family tree analysis:



In a family tree analysis, squares represent males, circles females, and the shaded shapes are individuals who may be affected by Fabry disease.<sup>22</sup>

# What are the benefits of early testing and diagnosis?

Early diagnosis of Fabry disease can have a very positive effect on patients.<sup>23</sup> This is because there are treatments available that may help to slow the progression of the disease and improve quality of life.<sup>23</sup>

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

<b>Enzyme replacement therapy (ERT)</b>	In people with Fabry disease the $\alpha$ -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. <sup>24</sup>
<b>Chaperone therapy (CT)</b>	In Fabry disease the $\alpha$ -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 changes in the gene. <sup>25</sup>

Complementary treatments that are not specific to Fabry disease are also available; these are also important and can help to manage the symptoms of Fabry disease.<sup>13</sup>

A further benefit of being diagnosed is the ability to receive genetic counselling. A genetic counsellor will be able to:<sup>14</sup>

- Provide information about the disease and its inheritance
- Discuss prenatal testing and decision making
- Help identify support resources



# 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.<sup>18</sup> 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.<sup>18,26</sup>

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

For further information visit [www.Fabrydisease.info](http://www.Fabrydisease.info)

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

