Dear .....

As you may or may not know, I have been diagnosed with Fabry disease. Fabry disease is caused by genetic mutations that can be passed down the generations of a family. I've mapped out our family tree, and you may have a Fabry mutation, although this does not necessarily mean you have the disease.

Fabry affects both males and females. It can cause a range of symptoms and can affect most parts of the body. The type, onset and severity of symptoms can vary from person to person – even for those within the same family. However, Fabry is progressive, getting worse over time, potentially causing serious or life-threatening problems – although this may not be the case for everyone.

Fabry is caused by genetic mutations. Mutations are like spelling mistakes in the instructions that tell the cells in your body what to do. In people with Fabry, an enzyme that usually breaks down certain sugary-fatty substances in the cells of our body doesn't work properly because of a mutation. This allows the sugary-fatty substances to build up, causing the problems and symptoms of Fabry.

Fabry mutations happen to the GLA gene on the X chromosome. Females have two X chromosomes and give one to their children randomly: so if a mother of a child has Fabry there is a 50/50 chance for her son or daughter to inherit the mutation. Males have one X chromosome and one Y chromosome; the X chromosome is inherited by their daughters and the Y chromosome by their sons. This means a man with Fabry will pass the mutation on to all of his daughters but none of his sons.

If you wanted to, you could be tested to see if you have a Fabry mutation with a genetic test, usually on a cheek swab, blood or other tissue sample. If you have any concerns about being tested you can discuss these with your doctor or another member of your healthcare team. However getting tested could mean that you avoid a long journey to diagnosis – for some patients it can take an average of 15 years to identify Fabry. It often takes so long because Fabry is rare, and the symptoms vary a lot and are similar to those caused by more common conditions. This can also mean some people never receive a diagnosis. As Fabry may worsen over time, getting tested could mean that you get help in managing the disease sooner which could potentially lead to a healthier future. There are different treatment options available.

If you want to be tested or to learn more about Fabry you should go and speak to a healthcare professional. This could be my doctor or someone my doctor refers you to, or you could take this letter to your own doctor.

For more information about Fabry, including its symptoms and how it is passed down in families visit **www.fabryfamilytree.com.au**

This example letter was created by Amicus Therapeutics Ltd for people with Fabry disease who wish to contact their family members who may have a Fabry mutation. It can be modified as the individual with Fabry sees fit.

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