Family and friends guide to Fabry

A guide for family and friends of people living with Fabry disease

Takeda

beMi®

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This booklet provides general information and is not a replacement for medical advice. Any questions about medical conditions, including Fabry, should be discussed with a healthcare professional.

Supporting someone living with Fabry

- Fabry is a genetic condition that can have a big impact on everyday life for some people.

- Symptoms of Fabry can affect many parts of the body. They can be mild through to severe, and can change over time. As a result Fabry never looks the same in different people.

- Life with Fabry can sometimes be challenging. It can involve:
  - managing many different symptoms
  - attending many medical appointments with different doctors for check-ups and treatment
  - juggling all of this with day-to-day work and family commitments.

- While there is currently no cure for Fabry, treatments are available to help.

- Having a good network can make a difference. Support from family and friends may help reduce the stress and mental load of living with Fabry.

To find out more about Fabry, see the *What is Fabry?* booklet on the beMi website.

How you can help

- Learn as much about Fabry as possible.

- Take a look at other beMi resources like the following booklets:
  - What is Fabry?
  - Living with Fabry

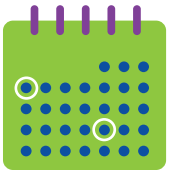
- Visit the websites of the support groups listed at the back of this booklet.

- Many people with Fabry are fully independent. Always ask if support is needed and what would be most helpful. Each person with Fabry will have different opinions on what might be useful for them. This can change depending on the symptoms they experience at any point in time, as well as other life commitments.

Here are some suggestions on how to help someone who might be in need.

Be a back-up when practical help is needed

- Offer to do the school drop off or pick up.
- Prepare a meal.
- Help create some time and space to rest if they need to.



Help with appointments

- Offer to be the designated driver or come along to medical appointments.
- Take notes during appointments, be a second set of ears or even record the appointment on your phone (with permission) so you can both remember what was said.
- Keep track of appointments in a diary or shared calendar.

Find Fabry support groups

- Finding other people with Fabry to talk to can be a great support for someone. Look for local or online support groups.

Listen/offer emotional support

- Just being there to hear someone or being able to offer emotional support can be a great help.

The health of carepartners is important, too! Care-partners can reach out to organisations for support, including:

- Fabry Australia
- Rare Voices Australia
- Carer Gateway

Who else might have Fabry?

Learning that a friend or relative has Fabry can come as a shock, especially for immediate family members. Family members may wonder whether they or other family members have also inherited the gene for Fabry.

How to find out

- The only way to know for sure is to get tested. Talk to a doctor about whether or not to get tested. Fabry is diagnosed from a blood sample.
- Family members of those diagnosed with Fabry may be referred to a genetic counsellor who can explain the risk of inheriting the Fabry gene and the likelihood of other family members also having the gene.
- An early diagnosis allows for discussions with a healthcare team about treatment options to manage symptoms and maintain a good quality of life.

Simple Science

- Fabry is a genetic condition that is inherited from a parent who has the *GLA* gene that causes Fabry.
- Fabry is caused by alterations to one specific gene (the *GLA* gene) on the X chromosome – one of the two chromosomes we inherit from our biological parents.

To find out more about how Fabry is inherited, see the **What is Fabry?** booklet.

Testing for Fabry

- Testing for Fabry involves taking a blood sample. The blood sample is checked for levels of an enzyme called alpha-gal A. This enzyme is needed to break down waste products inside cells and stop them building up. If these waste products build up in the cells, it can cause damage.

- Fabry changes the level of alpha-gal A that cells in the body can produce. Some people produce no alpha-gal A at all, and others may produce varying amounts of the enzyme. This means Fabry can look very different in different people.
  - Sometimes, females with Fabry have the same level of alpha-gal A as people who do not have Fabry. A genetic test is needed to look for the Fabry gene – an alteration to the *GLA* gene – in these people.
  - Prenatal testing can also be offered to females with Fabry who are expecting a baby.

- After a family member is diagnosed with Fabry, other relatives can be screened to see if they also have it.

Simple Science

Alpha-gal A's full name is alpha-galactosidase A.

Enzyme – things that help speed up chemical reactions.

More about genetic counselling

Genetic counsellors will usually ask questions about a person's family tree and talk about the potential risk of inheriting Fabry. These counsellors will also discuss the risk of other family members inheriting Fabry, too.

The counsellor may also talk about the pattern of inheritance, family planning, and genetic screening. They are also available to answer any other questions about Fabry within a person's family tree.

Doctors or Healthcare professionals are able to refer people to a genetic counselor or advise where to find a Fabry clinic. For more information about Fabry clinics in Australia visit Fabry Australia.

The future with Fabry

Fabry is a progressive condition and, while there is no cure now, treatment is available. Research continues to look for new treatments and ways to improve the quality of life for people living with Fabry.

With a diagnosis of Fabry, people can work with their healthcare team to find the best treatment to suit their needs and deal with any symptoms they may experience. For more information on living with Fabry, see the **Living with Fabry** booklet.

To find out more

To find out more about Fabry, visit the following independent Australian organisations.

Fabry Australia
www.fabry.com.au
admin@fabry.com.au

Rare Voices Australia
www.rarevoices.org.au
info@rarevoices.org.au
RARE Helpline:
www.rareportal.org.au/rare-helpline/

Other booklets and animations are also available on the beMi website at **bemi.health**

In the spirit of reconciliation, Takeda acknowledges the Traditional Custodians of Country throughout Australia, and their connections to the land, sea and community. We pay our respects to their Elders past, present and emerging, and extend that respect to all First Nations peoples.

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