Parents' guide to Gaucher

A guide for parents and care partners of a child living with type 1 Gaucher disease

Inside this booklet

This booklet provides general information about supporting children living with Gaucher. It is not intended to replace advice or care from a healthcare team. Readers are advised to speak with a healthcare professional for further advice and support.

Supporting a child living with Gaucher

- Gaucher (pronounced ‘go-shay’) is a genetic condition that may have an impact or affect everyday life for some children and their families.
- Symptoms of Gaucher can affect many parts of the body. They can be mild through to severe, and can change over time. Some people with Gaucher have no symptoms. As a result, Gaucher never looks the same in different people.
- Sometimes, life with Gaucher can be challenging. Having Gaucher can involve:
  - managing lots of different symptoms
  - attending many medical appointments with different doctors for check-ups and treatment
  - juggling all of this with day-to-day work, school and family commitments.
- While there is currently no cure for Gaucher, treatments are available to help.
- Supporting a child with Gaucher can be tough, but having a good support network can make a difference. Support from family and friends can reduce the stress of supporting a child with Gaucher.

This guide aims to provide helpful information and resources about Gaucher and what it may mean for a child living with Gaucher and their family.

To find out more about Gaucher, see the *What is Gaucher?* booklet at beMi.health.

How Gaucher might affect a child

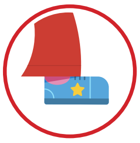
The support and treatment needs of a child diagnosed with Gaucher will depend on their symptoms and parts of the body affected.

Note: Some children may experience no symptoms at all.

Bruising easily. Avoid contact sports and consider gentler options such as swimming, dancing, table tennis or yoga. These may also help with tiredness and shortness of breath.

Fatigue and tiring easily. Try and adjust daily routines to allow more sleep time. Discuss what support schools can provide for school-aged children.

Pain. Some children with Gaucher experience bone pain, or pain from enlarged organs. Healthcare teams can suggest strategies or medicines to help.



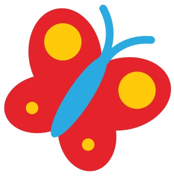
Mobility. Walking up and down stairs or walking for long distances may be challenging. Bone pain or bone fractures can impair mobility.

Changes in appetite and feeling full quickly. Eating a full meal may be difficult because enlarged organs can put pressure on the stomach. Smaller portions may help, and arranging a longer time to eat. The doctor may recommend a specific diet.

Some children with Gaucher may need to attend regular medical appointments to see different doctors and receive treatment.

Try activities such as reading, audiobooks, colouring in or portable games to keep children occupied in waiting rooms or when receiving treatment in hospital.

Support suggestions



Sometimes a diagnosis of Gaucher can be unsettling for the whole family. However there are things you can do to support a child and minimise the impact of the condition on family life.

Educate yourself – Try learning as much as possible about Gaucher. Take a look at the ***What is Gaucher?*** booklet on the beMi website and the ***Find out more*** section of this booklet to get started.

Keep others in the loop – Inform family, friends and other community members about Gaucher and what it means for someone living with the condition. Schools are often happy to help in whatever way they can, so it may help to discuss what support they can offer school-aged children.

Promote healthy food choices – Eating a varied, healthy diet is great for everyone. Try and have these healthly foods available at home. Healthcare teams may suggest a special diet or dietary supplements for bone and blood health.

Encourage activity – Regular exercise helps strengthen bones and muscles. Healthcare teams can advise which activities may be appropriate.

Establish a sleep routine – Some children may need more sleep than what is recommended for their age group. Encourage children to get enough sleep to help manage tiredness.

Talk to children about Gaucher. Where possible, be available to answer any questions about the condition.

Connect with the Gaucher community – Reach out to Gaucher organisations. The Gaucher community is a great place to find advice and support. See the ***Find out more*** section of this booklet to get started.

Emotional support

Having a chronic health condition can be tough ...

Sometimes people with Gaucher may feel anxious or sad about the challenges they are facing. They may be self-conscious about how they look. Teenagers may go through puberty later than their peers, which may make some feel different, isolated or self-conscious about how they look.

Some children may want to discuss how they feel. Make sure they know who they can speak to.

Healthcare teams can advise on mental health support that might be available. Some children may benefit from seeing a counsellor or a psychologist.

Having a child with a chronic health condition can also be tough ...

It is extremely important for parents and other care partners to look after their own wellbeing too!

At times, it is not uncommon to feel overwhelmed, exhausted, anxious or sad. Friends and family can be a helpful support even if it's just to listen.

Support for care partners is available from many Australian organisations. See the ***Find out more*** section of this booklet. Always speak to your doctor or a member of your child's healthcare team if you are feeling worried or sad.

Reach out for help

Having a child with a health condition such as Gaucher may have ups and downs and a lot of medical appointments. At times help may be needed.

Friends and family members may be available to lend a hand in times of need.

For example, they could:

- take children to their hospital appointments
- read children a story while they are receiving treatment
- cook a meal
- pick up other children from school or activities, or look after them during medical appointments.

There are many other different ways that help from others can make a big difference. Having a list of tasks can be handy for when people ask how they might be able to help.

Who else might have Gaucher?

Learning that a child has Gaucher may leave people wondering if other children or family members also have the condition, or if they are carriers of the Gaucher gene.

How to find out

- Healthcare professionals can discuss testing options for family members, and can arrange testing. Gaucher is diagnosed from a blood sample.
- Some doctors may suggest seeing a genetic counsellor to talk about the risk of other children or family members having Gaucher, or carrying one copy of the Gaucher gene.
- Being diagnosed as early as possible means earlier conversations with healthcare teams about management and treatment options. These treatments and management techniques may help to alleviate symptoms and keep focus on maintaining a good quality of life.

Simple science

- Gaucher is a genetic condition. It is inherited from parents who have the *GBA1* gene that causes Gaucher.
- Children with Gaucher receive one copy of the Gaucher gene from each parent.
- People with just one copy of the Gaucher gene are known as carriers.

To find out more about how Gaucher is inherited, see the ***What is Gaucher?*** booklet on the beMi website at bemi.health.

More about genetic counselling

This involves genetic counsellors asking about relatives to create a family tree and discussing the risk of family members having Gaucher.

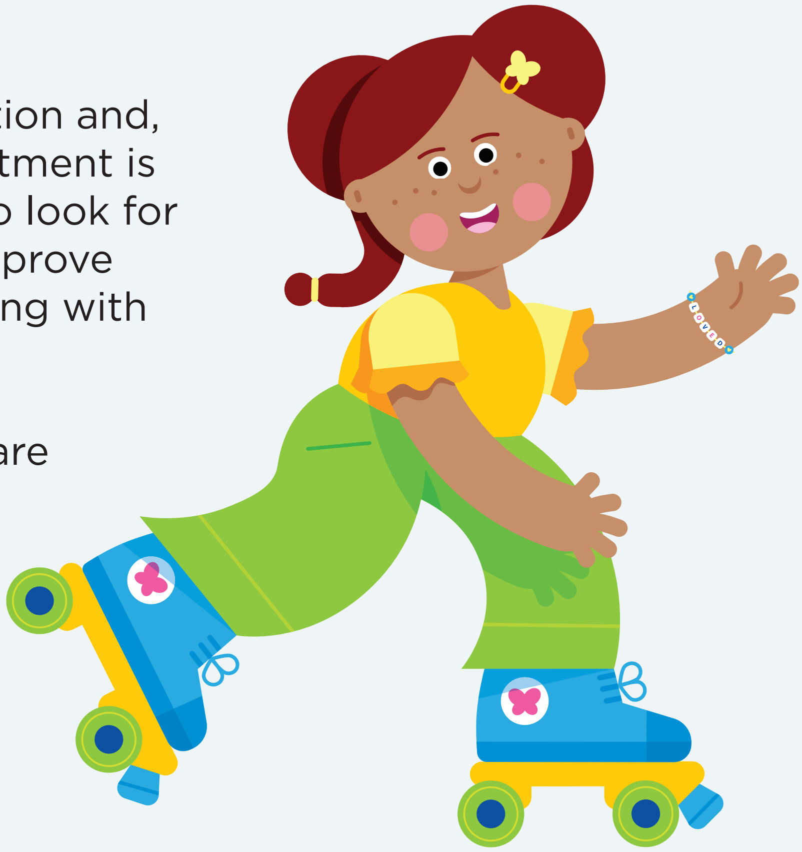
The counsellor can also discuss the pattern of inheritance, family planning, genetic screening and answer any questions about Gaucher within families.

Doctors can provide a referral to a genetic counsellor.

The future with Gaucher

Gaucher 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 Gaucher.

Working closely with a healthcare team is the best way to find the most appropriate treatment plan for each person with Gaucher depending on the symptoms they experience.



Find out more

To find out more about Gaucher or its treatment, speak with a healthcare professional.

The following independent Australian organisations have helpful resources:

- Gaucher Association of Australia & New Zealand gaucheranz.com.au
- Rare Voices Australia rarevoices.org.au

Other booklets, including the ***What is Gaucher booklet***, and animations are 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 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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