

A young boy with dark hair is smiling broadly while riding a carousel horse. He is wearing a light blue and white striped short-sleeved shirt. The background is blurred, showing warm lights from the carousel and other people. The right side of the image features a large, stylized purple graphic element that frames the text and logo.

PARENT'S GUIDE TO GAUCHER

 **Shire**

This booklet is available to the public for information only; it should not be used for diagnosing or treating a health problem or disease. It is not intended to substitute a consultation with a healthcare professional. Please consult your healthcare professional for further advice.

This booklet aims to answer any questions you may have about Gaucher disease and how it is likely to affect your child

Finding out that your child has Gaucher (pronounced “go-shay”) can be extremely worrying. If no one else in the family has it, diagnosis with Gaucher can come as a shock.

You will probably have lots of questions about how it will affect your child’s daily life (both at home and at school), what will happen to them in the future, and whether other family members may have the condition.

It is important to try and understand what your child is experiencing so that you can give them the best support and minimise the impact of their condition on family life.



ABOUT GAUCHER

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What is Gaucher?

Gaucher is a genetic lipid storage disorder, in which lipids (fatty substances) build up in certain cells in the body (called macrophages). It is caused by a missing or deficient enzyme called 'glucocerebrosidase', which normally breaks down these lipids. Accumulation of lipids in cells in the liver, spleen and bones can interfere with some of the normal processes of the body.¹

Gaucher belongs to a group of conditions called lysosomal storage disorders (LSDs).¹ There are 3 main types of Gaucher disease:

Type 1 (Non-neuronopathic) – the most common type² with variable symptoms and progression³

Type 2 (Acute neuronopathic) – an acute, severe form affecting the central nervous system in babies and infants³

Type 3 (Chronic neuronopathic) – a chronic form affecting the central nervous system, that progresses over time⁴

How common is it?

Gaucher is a rare genetic disorder affecting around 1 in 100,000 of the population.⁵ Most affected people (over 90%) have Type 1 Gaucher.² It affects males and females equally.⁶

Gaucher is more common within the Ashkenazi Jewish population, with approximately 1 in 1000 having Type 1 Gaucher.¹

What are the symptoms?

Gaucher is different for everyone, with symptoms varying considerably from person to person.^{3,7} Some children with Gaucher experience severe symptoms, while some only have mild symptoms and are not diagnosed until later in life.⁶

You may notice that your child gets tired easily and cannot do everything that they used to do. You may need to adjust their daily routine and allow them more time for particular tasks or activities.

Your child may also bruise more easily, so it is important for them to take extra care when playing and at school.

The most common clinical signs of Type 1 Gaucher are:^{3,4}

- Tiredness
- Bone pain
- Tendency to bruise
- Enlarged liver
- Enlarged spleen

In Type 3 Gaucher, the nervous system is also affected and additional signs may include:^{3,7}

- Abnormal eye movements
- Loss of muscle coordination
- Learning difficulties
- Seizures
- Dementia

If your child has been diagnosed with Type 2 Gaucher, speak to your healthcare professional for further information.

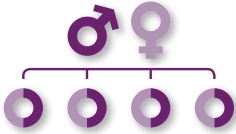
How is Gaucher passed on within families?

Finding out that your child has Gaucher may cause concern about how it will affect you and the rest of your family.

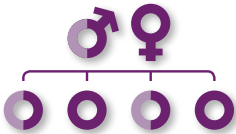
Gaucher is an inherited disorder that is said to be 'recessive'.⁶ This means that a child needs to inherit a copy of the faulty gene from both parents to have Gaucher. See the next page for a clearer picture of how Gaucher is passed on in families.

Your healthcare professional can help to explain how other family members may be affected and may recommend genetic testing. Prenatal screening is also available to check whether the unborn child is at risk of developing Gaucher.⁴

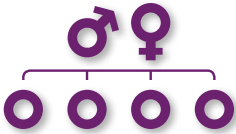
Here is a guide on how Gaucher is passed on within families:



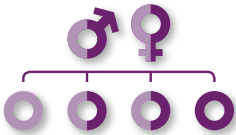
If one parent has Gaucher but the other does not, the child will be a 'carrier' of the condition but will not have Gaucher⁸



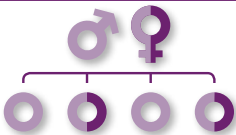
If one parent has Gaucher and the other is a carrier, there is a 1 in 2 chance that the child will also have Gaucher⁸



If both parents have Gaucher, their children will also have the condition⁸



If both parents are carriers but do not have Gaucher, there is a 1 in 4 chance that their child will have Gaucher⁸



If one parent is a carrier and the other is not, there is a 1 in 2 chance that their child will be a carrier, but they will not have Gaucher⁸



Parents

Child



No Gaucher



Gaucher carrier



Has Gaucher

TREATING GAUCHER

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What treatments are available?

There is currently no cure for Gaucher, but different therapies are available that can help to treat many of the major symptoms.⁹ Your child's healthcare professional will decide which treatment is suitable for them depending on their diagnosis and the severity of their condition. Their healthcare team will be able to provide information and advice on the treatments available.

Enzyme Replacement Therapy

Enzyme Replacement Therapy (ERT) replaces the missing or deficient enzyme in the affected cells, and can help to relieve symptoms.⁹ Long-term treatment in Type 1 Gaucher has been shown to reduce the size of the liver and spleen and improve red blood cell and platelet counts.⁹

In Type 3 Gaucher, there is evidence to show that an approved ERT improves haemoglobin and platelet levels as well as reduces enlarged liver and spleen, but there is no evidence that it improves neurological symptoms.³ However, there is some anecdotal evidence that some children appear to show improvements in unsteadiness (ataxia) and some forms of fits.¹⁰ It is currently being researched whether ERT can offer protective benefits in neurological disease.¹⁰ Patients with all forms of Gaucher may require supportive treatment for their disease at some time in their lives.

ERT is given by intravenous (IV) infusion, usually over a period of 1–2 hours every other week depending on individual patient requirements.⁹

Other treatments

Depending on individual symptoms, people with Gaucher may need treatment for bone complications and/or medication for pain.

Visiting the hospital

You will need to work closely with your child's care team to make important decisions about your child's treatment. It is important that your healthcare professional gets a clear picture of your child's symptoms to be able to provide the most effective treatment regimen.

Children can find it hard to communicate how they are feeling, particularly when trying to describe pain. Visual analogue scales may be useful to help them to communicate their symptoms. These scales often show a range of expressions on faces, from happy to sad.

It may be difficult for you to fit hospital appointments around family life and work. Patient associations are a good source of advice and support, as you can talk to other parents about their experiences.

Treatment at home

Most people receiving ERT in the UK will have the option to receive regular infusions at home with the help of a homecare company, if they are tolerating their infusions well. So, you may be able to arrange for a nurse to come to your home to administer your child's treatment when they are used to it. Speak to your healthcare professional for more information.

What can I expect from their treatment?

Everyone with Gaucher is different, and responses to treatment will vary from person to person.

Your healthcare professional will discuss their treatment plan with you in detail and help set individual aims for your child's therapy.

Depending on your child's initial symptoms, these aims may include:⁷

- Increasing haemoglobin count
- Increasing platelet count
- Reducing liver size
- Reducing spleen size
- Managing bone pain and preventing bone crises
- Improving quality of life



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CARING FOR A CHILD WITH GAUCHER

Looking after a child with Gaucher

This section has been developed to help parents support a child with Gaucher. Please discuss the specific needs of your child with your healthcare professional. Their needs will depend on their age and the severity of their symptoms.

What should I tell my child?

It can be difficult to explain Gaucher to your child, especially if they are still very young. Your child's care team and patient associations can advise you on the best way to approach it.

The 'Gaucher for children' section of this booklet provides some guidance on how to talk to your child about their condition.

Children with Gaucher need a lot of emotional support, as the condition can make them feel isolated or different. It is important that you ask them about how they are feeling and try to understand what they are going through. For example, they may be self-conscious about how they look with a swollen tummy, or worried about being different from other children at school. It is also important to provide emotional as well as practical relief if your child suffers an episode of pain.¹¹

By understanding your child's emotional needs, you can help them deal with the hurt that they may be experiencing.

Talk to your healthcare professional and the rest of the care team about receiving support for the whole family.

How will Gaucher affect their growth and development?

Some children with Gaucher may be shorter and smaller than other children their age.¹² The onset of puberty can also be delayed in teenagers with Gaucher, which may be difficult for them if they are not as developed as their friends.¹² However, they usually catch up later in life.¹²

How will it affect them in the future?

The symptoms and progression of Gaucher vary from person to person.^{3,7} Treatment can help reverse many of your child's symptoms and improve their quality of life.⁹

Talk to your healthcare professional if you have any concerns about your child's condition, as they will be able to discuss his/her individual symptoms in more detail.



What about my other children?

If you have other children, talk to your healthcare professional about genetic testing. Even if your other child/children do not have the same symptoms, the condition may also affect them in the future or they may be carriers. Further information on the inheritance of Gaucher can be found in the 'About Gaucher' section.



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GAUCHER AT SCHOOL

Making adjustments at school

It is important to talk to your child's school about their condition, as they may need to make some adjustments to your child's daily timetable. It may be a good idea to set up a meeting with teachers, healthcare professionals and other people involved in their care to discuss their needs.

Your child may find, at times, that they will not have enough energy to play with other children and may have difficulty concentrating in class. If tiredness is affecting your child at school, you may need to talk to their teachers about arranging times during the day for them to rest.

If you feel your child needs additional support at school, you will need to contact your local education authority. In some cases, they can allocate support staff who will work closely with your child and teachers to provide physical, emotional and educational support.

You can get more advice about additional support at school from patient associations, or by talking to other parents of children with Gaucher, perhaps via patient associations.

Gaucher and physical activity

It is important for your child to try and stay physically active since regular exercise can help to strengthen their bones and muscles.

Some children with Gaucher, however, will not be able to take part in many physical activities, as they bruise more easily and risk bleeding or damaging their spleen. They may feel left out, so you may want to find alternative activities for them to do. They may be able to take part in non-contact sports, such as swimming, cycling and dancing.¹¹

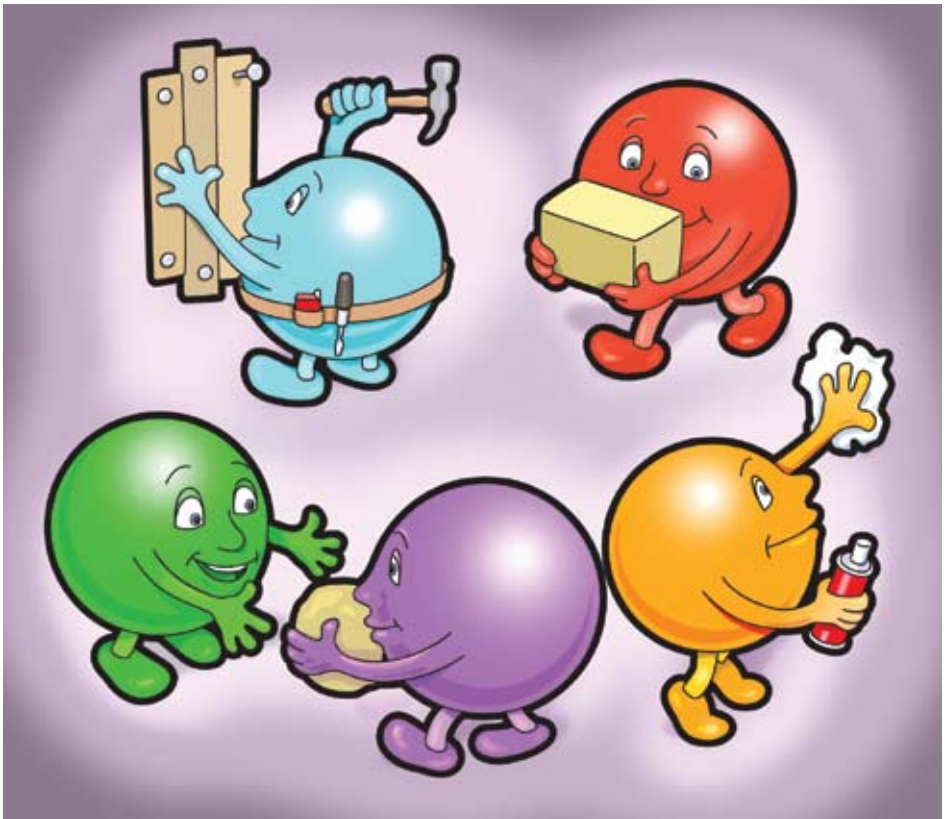
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GAUCHER FOR CHILDREN

How do I talk to my child about Gaucher?

This section provides an example of how you can talk to your child about their condition. It uses simple analogies to help them understand the things they are experiencing. You can either use it as a direct dialogue, or to guide your own discussions with your child. There are lots of accompanying pictures to help make the information more engaging and easier to understand.

Explaining what is going on inside their body:



Our bodies are like small factories. Inside we have lots of little molecules that all do very important jobs. That is what makes our bodies work properly.



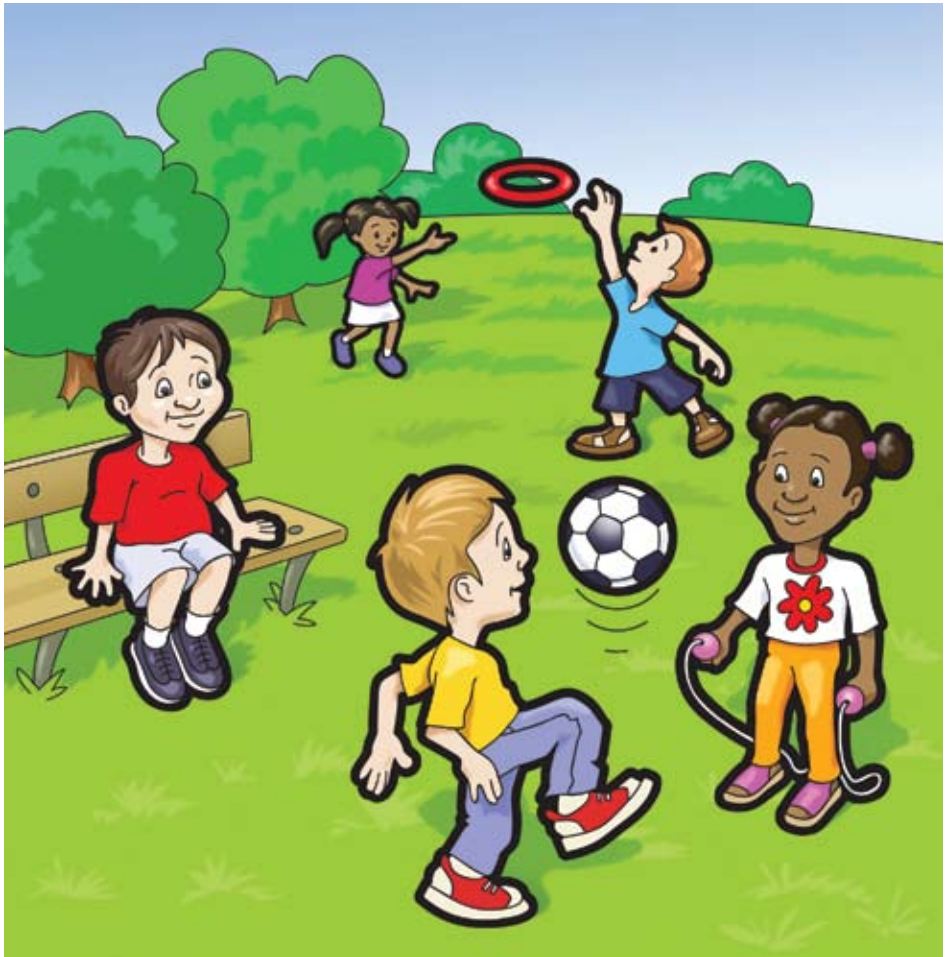
Inside your body, all of these molecules are working perfectly, apart from just one. This one little molecule isn't doing its job very well. It should be helping to clear out things that your body doesn't need – a bit like when you clean up your room and throw away things you don't want anymore. Because it is not doing its job, things start to pile up and get in the way.



There are special places in your body where these things might start to pile up – like in your bones, and in parts of your body called the ‘liver’ and the ‘spleen’.

You can see in this picture that your liver and the spleen are in your tummy. If things start to pile up inside them they can become bigger. So this can make your tummy poke out more.

Describing how this might affect their day-to-day life:



Just because this one little molecule isn't working properly, it doesn't mean that you are very different from anyone else. In fact, you can do most of the things that your friends can do. You might just feel really tired sometimes. For example, when you are out playing with your friends. If you do start to feel tired, just stop and take a little break. You can join back in again afterwards – with a bit more energy!

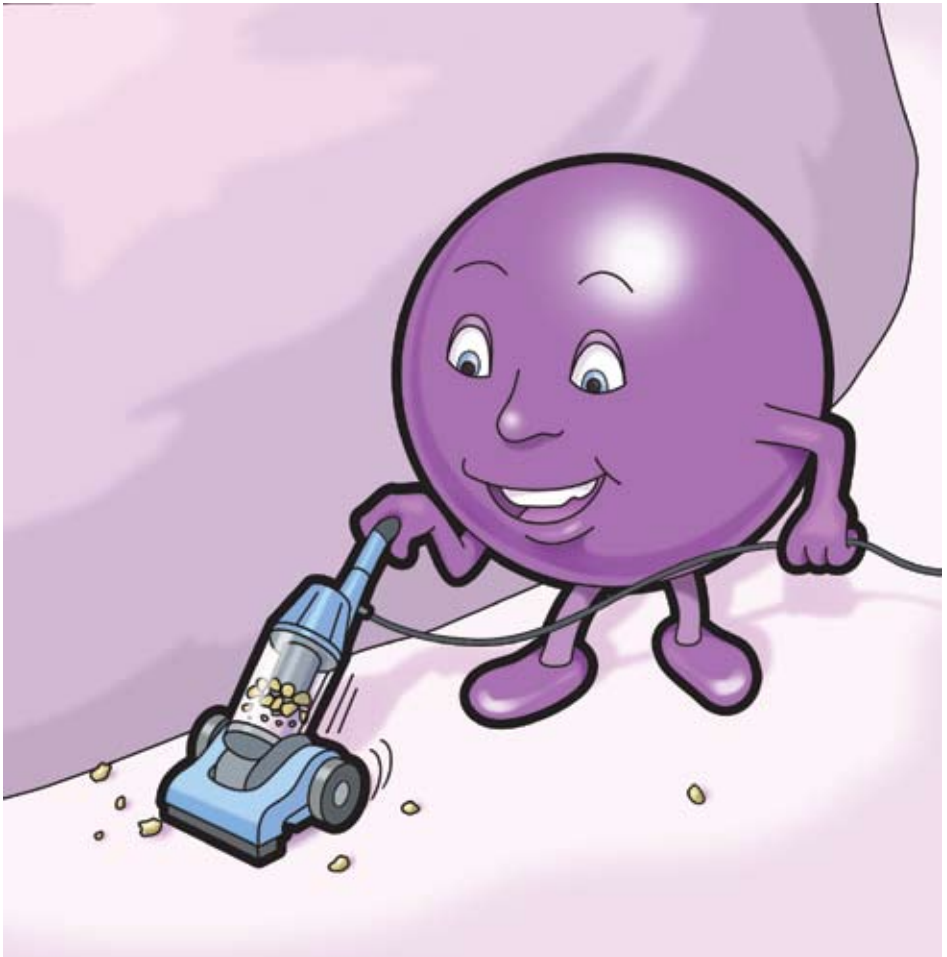


You might even feel tired at school. Sometimes it can be difficult to concentrate in class if you feel tired. It's ok to tell your teacher if this happens. And tell me if you have been feeling tired too. I can talk to your school so that they can make it easier for you to keep up with everyone.



There are a few sports at school that you might not be able to play. This is only because you can get hurt more easily than your friends. But, there are lots of fun sports you can enjoy - like swimming, cycling and dancing.

Talking about your child's medicine – how it works and what it will do:



We can't fix that molecule in your body that isn't working. But, we can give you a brand new one. That is what your medicine is – a brand new molecule that works perfectly well and can help get the job done! It will start to clear up all those things that have been piling up in your body. And it will make sure that they don't have the chance to pile up again!

This means that your liver and spleen will be cleared up of all that unwanted 'junk' and so they will become smaller again. And this will make your tummy go back to its normal size, so it won't poke out anymore.



Your medicine will help relieve the symptoms that are making you feel tired.

Explaining how your child receives their medicine, including visits to the hospital:



Your medicine, which gives you a brand new molecule, needs to be put inside your body. To start with, you will need to go to and see a special nurse at the hospital to receive your medicine.

She will put a really thin tube into your arm – this is for the new molecule to travel down to go into your body. Without it, the new molecule has no way of getting inside you, so it's really important. It only takes a moment to put it in, which might hurt a little bit. When she's put it in, the nurse will make sure it stays in place with some sticky tape – we don't want it coming out and your new molecule getting lost!

Because the molecule can take a little bit of time to travel down the tube, the nurse will give you a nice, comfortable chair to sit on and you can watch TV if there is one. You can even take your favourite book or toy with you if you want.



Your medicine is enough for about two weeks. So, to make sure you keep seeing the benefits, you will need to take some more medicine every two weeks.



ADDITIONAL RESOURCES

Gaucher dictionary

Anaemia

A low level of haemoglobin/red blood cells, leads to weakness and lack of energy as not enough oxygen is being circulated around the body.¹³

Gaucher

Gaucher (pronounced: go-shay) is a rare lysosomal storage disorder.¹ It is named after a French dermatologist Dr Phillipe Gaucher who first described the disease over 100 years ago.¹

Glucocerebroside

Glucocerebroside is a lipid (fatty substance), which builds up in certain cells in people with Gaucher.¹³

Glucocerebrosidase

The enzyme that breaks down glucocerebroside. In people with Gaucher, glucocerebrosidase is either missing or only produced in small amounts.¹³

Haemoglobin

Haemoglobin is the protein within the red blood cells that helps carry oxygen around your body, and is responsible for the red colour.¹³ Low haemoglobin levels (anaemia) cause tiredness as less oxygen is getting to where you need it.¹³

LSD (lysosomal storage disorder)

A disorder caused by a malfunction in a specific organelle in the body's cells called a lysosome. Gaucher is the most common LSD.¹⁴

Macrophages

A type of white blood cell that removes dead cells and helps to fight infection.¹³ In people with Gaucher, lipids accumulate within these cells.⁶

Neurological

Relating to the central nervous system. Type 2 and Type 3 Gaucher have a greater impact on the central nervous system, but neurological involvement has also been reported in Type 1 Gaucher.⁶

Platelets

Platelets help your blood to clot.¹³ Low levels of platelets (thrombocytopenia) in your blood make it easier for you to bruise and more difficult for you to stop bleeding.¹⁵

Recessive disorder

A disorder that requires two copies of the faulty form of a gene (one from each parent) to show a certain characteristic/symptoms.¹³

Spleen

The spleen is an organ that helps to break down old red blood cells and to fight infection.¹³

Useful contacts

UK and Ireland Patient Association

Gauchers Association
3 Bull Pitch, Dursley
Gloucestershire GL11 4NG
Tel/Fax: +44 (0)1453 549 231
www.gaucher.org.uk

European Gaucher Alliance

3 Bull Pitch
Dursley
Gloucestershire GL11 4NG
United Kingdom
Tel/Fax: +44 (0)1453 549 231
www.eurogaucher.org

EURORDIS

Patient-driven alliance for people living
with rare diseases in Europe
www.eurordis.org

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