

FINDING OUT
ABOUT 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 have about Gaucher disease and to provide practical information about how to minimise the impact that it may have on your life

Being diagnosed with Gaucher (pronounced “go-shay”) can be extremely distressing and confusing. You may have had lots of tests and appointments with doctors before finding out what is causing your symptoms.

If you have been diagnosed with Gaucher, your healthcare professional will discuss your treatment plan with you. Effective treatments are available to help minimise your symptoms, which may allow you to carry on with your normal daily activities.¹

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 your 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 your liver, spleen and bones can interfere with some of the normal processes of your body.²

Gaucher belongs to a group of conditions called lysosomal storage disorders (LSDs).² There are 3 main types of Gaucher disease:

Type 1 (Non-neuronopathic) – the most common type³ with variable symptoms and progression¹

Type 2 (Acute neuronopathic) – an acute, severe form affecting the central nervous system in babies and infants¹

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

How common is Gaucher?

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.³ 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.²

How will Gaucher affect me?

Gaucher is different for everyone, with symptoms varying considerably from person to person.^{1,7} Some people experience severe symptoms in childhood, while some people have no symptoms or only mild symptoms and are diagnosed later in life.⁶

You may find that you get tired easily and it may have become more difficult to do everything that you used to do. It may help to pace yourself, by changing your daily schedule and/or taking time in the day to rest.

You may also find that you bruise easily, so you might need to avoid or take extra care with some physical activities.

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

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

ASSESSING GAUCHER

What do all the tests mean?

To diagnose Gaucher properly, your healthcare professional will have carried out a number of tests – including checking the level of the glucocerebrosidase enzyme in your blood and a DNA test (genotyping) to check which mutation of the gene you have.⁹

A number of tests will be repeated at follow-up visits to check your progress and response to treatment (see Table 1). Your healthcare professional will carry out tests depending on your individual symptoms to provide you with the best possible care.

Some of the terms you hear or see on your assessment forms may not be familiar to you. An overview of some of the most common tests that are used can be found in Table 1 on the next page.



Common tests that are used to assess Gaucher symptoms¹⁰

Table 1

Blood test	Function
Haemoglobin / red blood cells	To assess anaemia. Low levels can cause tiredness and weakness
Platelets	Low levels (thrombocytopenia) can lead to bruising and bleeding
Protein 'biomarkers': • Chitotriosidase ⁷	Levels of these proteins increase in Gaucher. They can be used to assess severity and monitor response to treatment ⁷
White blood cells	Low levels (neutropenia) may increase the risk of infection
Iron	To exclude other causes of anaemia
Ferritin	To assess response to therapy
Scan	Function
MRI/CT scan	Scans can help to monitor bones and organs and to measure volume of liver and spleen
DEXA scan	Measures bone mineral density (BMD), an indicator of bone strength. DEXA provides a score relative to normal population (may not be as accurate in children)

Other tests

Lung tests

Although it is uncommon for people with Gaucher to develop respiratory symptoms,⁷ your healthcare professional may carry out lung function tests if you start to have any breathing difficulties

Eye movement test

Eye movement tests assess whether your condition has started to affect your central nervous system.⁷

Physical examination

Your healthcare professional will probably carry out a physical examination to assess the size of your liver and spleen. They may also measure your weight, muscle strength, bone tenderness and mobility.⁷

General health and wellbeing

Your healthcare professional will ask you questions about how active you are and how you are feeling.⁷ They may ask you to complete a quality of life questionnaire about your symptoms and their impact on your daily life.⁷

MANAGING GAUCHER

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What treatments are available?

Your doctor will talk to you about the treatment options available to you. There is currently no cure for Gaucher, but different therapies are available which can help to treat many of the major symptoms.¹¹ With treatment, people with Gaucher may lead full lives and may be able to carry out many of their normal daily activities.

Enzyme Replacement Therapy

Enzyme Replacement Therapy (ERT) replaces the missing or deficient enzyme in the affected cells, and can help to relieve symptoms.¹¹ Long-term treatment has been shown to reduce the size of the liver and spleen and improve red blood cell and platelet counts.¹¹

ERT will not treat the neurological symptoms of Gaucher.¹ 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.¹¹

Substrate Reduction Therapy

Substrate Reduction Therapy (SRT) reduces the production of lipids in your cells to help minimise symptoms.¹ It is currently recommended in patients who are unsuitable for ERT.¹²

Treatment for bone complications

Bone pain

If you experience chronic or acute episodes of pain, talk to your doctor about pain management. It may help to keep a pain diary to assess how you are feeling on a day-to-day basis. This can help you to explain to your doctor the impact it is having on your life.

Weakened bones

If your doctor has identified that you have weakened bones (low bone mineral density), you may be prescribed medication to help strengthen your bones.¹

Surgery

If bones or joints become badly damaged or fractured, surgery may be required. In some cases, hip or other joint replacements may be needed.



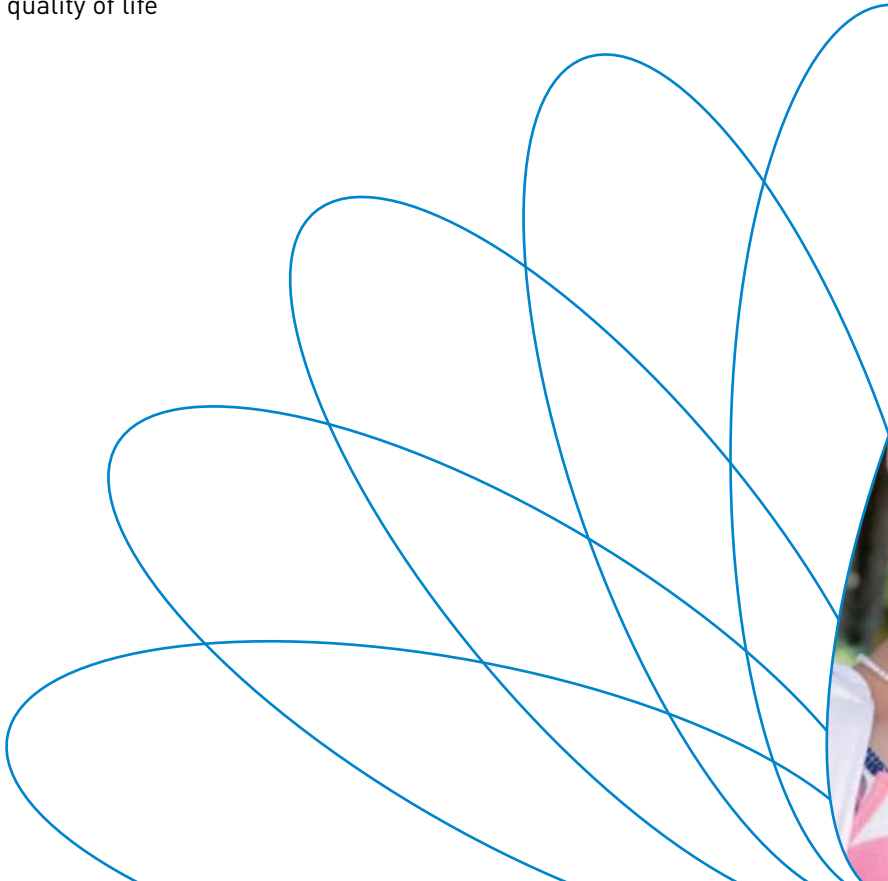
What can I expect from my treatment?

Everyone with Gaucher disease is different, and responses to treatment will vary from person to person. You may find your energy levels increase as your haemoglobin count increases. You may also notice that your liver and spleen reduce in size and become less swollen.

Your physician will discuss your treatment plan with you in detail and help set your personal therapeutic goals.

Depending on your initial symptoms, these goals 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



How often do I have to go to hospital?

Your healthcare professional will advise how often you will need to have assessments. Following your initial assessment, you are likely to have blood tests every 3 months, and a more detailed assessment with a specialist every 6–12 months. You will need to attend a national centre specialising in Gaucher.

If you have ERT, you will need to go to a specialist clinic or hospital to receive your infusion initially.

You may be able to arrange for a nurse to come to your home to administer your treatment when you are used to it.

If you need to travel a long way to see your specialist, you may be able to arrange to have some assessments at a hospital nearer to your home.

Contact your healthcare professional if you have any concerns about your treatment, or if you notice any changes in your symptoms.



Getting the best from your treatment

As Gaucher is a long-term condition, you will have a close partnership with your healthcare team. They will work with you to make the best possible decisions about your care.

- Try to be open and honest about your symptoms. Although it can be difficult to talk about how you are feeling, it is important that your healthcare professional understands how your symptoms are affecting you in order to manage your condition effectively
- If you do experience a change in symptoms, your healthcare professional may change your treatment regime if appropriate
- It may help to keep a diary of your symptoms or note down any questions you have, particularly if it is a long time until your next appointment
- Don't be embarrassed to ask questions if you don't understand anything or would like more information

What can I expect in the future?

Progression is variable and effective treatments are available that can help minimise many of the symptoms.

It is thought that people with Gaucher may have a risk of developing other conditions later in life, including Parkinson's disease (a progressive neurological disease) and multiple myeloma (cancer affecting white blood cells).^{2,8} However, the risk of these complications is still low.⁸

In some patients, bone disease may lead to permanent damage and fractures, which can cause physical disability and may require surgery.⁴

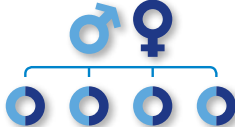
Talk to your healthcare professional if you have any concerns about your condition, as they will be able to discuss your individual symptoms in more detail.

LIVING WITH GAUCHER

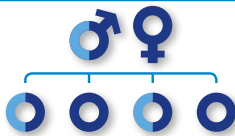
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Will my children also have Gaucher?

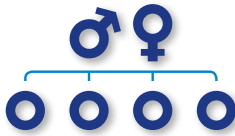
Gaucher is an inherited disorder that is said to be 'recessive'.⁶ This means that a child needs to inherit a copy of the faulty gene from both parents to display symptoms of the condition. It affects both males and females.



If your partner does not carry the faulty form of the Gaucher gene, then your child will be a 'carrier' of the condition but will not have Gaucher¹³



If your partner is a carrier, there is a 1 in 2 chance that your child will have Gaucher¹³



If your partner has Gaucher, your child will also have Gaucher¹³



Your healthcare professional can help to explain how other family members may be affected and may recommend genetic testing.

If you are pregnant, prenatal screening is also available to check whether the unborn child is at risk of developing Gaucher.⁴

Can I exercise and play sports?

It is important to try and stay physically active. Regular exercise can help to strengthen your bones and muscles. Your level of activity will depend on the nature of your symptoms:

- If you have an enlarged spleen, your healthcare professional may advise that you should not play contact sports. This is due to the risk of damaging your spleen. They may recommend other low-impact activities
- If you bruise easily, you should probably avoid contact sports
- If you have joint damage or weak bones, you may need to limit your physical activity in case you fracture your bones

Your healthcare professional/physiotherapist will be able to advise on what type of exercise is right for you.

Do I need to change my diet?

It is important for everyone to maintain a healthy balanced diet, whether they have Gaucher or not.

If your calcium and vitamin D levels are low, you may be advised to increase your calcium intake, and possibly take supplements, as these nutrients are important to help strengthen your bones. The main dietary sources of calcium are dairy products, some fish, spinach, broccoli, nuts and seeds.

If you have anaemia, your healthcare professional may advise that you take iron tablets or increase the iron content in your diet. Foods that are high in iron include spinach, wholegrain cereals, pulses such as lentils and kidney beans, and some dried fruits.

If you have an enlarged liver or spleen, you may find that you feel full more quickly. You may, therefore, need to consume food that is energy-rich.

Do I need to make other changes to my lifestyle?

Limiting your alcohol intake and not smoking can help to improve your overall health and maintain bone health.¹⁴ This is because excessive alcohol can affect the balance of calcium in your body and hormones involved in calcium absorption.¹⁴ Smoking may also lead to increased risk of osteoporosis and fractures.¹⁴

Can I go on holiday?

Talk to your healthcare professional if you are planning a holiday. If you are having ERT you may want to time your trip so that you don't miss an infusion. If you require treatment while you are away, patient associations in the country you are travelling to may be able to provide you with information and advice.

If you want to take a break from your treatment you should talk to your healthcare professional.

Will I still be able to study/work?

Many people receiving treatment for Gaucher are able to maintain normal lives at college, university and work. However, you may find you need to make adjustments to your daily routine and take time off regularly to attend medical appointments.

It is important that your educational institution or employer fully understands your condition and the impact of the symptoms you are experiencing, particularly as they have probably never heard of Gaucher. When you talk to them it may be helpful to show them information, which can help you to explain the condition in more detail.

Tiredness and lack of energy may prevent you from carrying out some tasks, particularly physical activities. You may even need to consider a career that is not as physically demanding. Career counsellors can help you to find suitable careers, taking into account your personal skills and interests.

At the back of this leaflet you will find details of patient organisations, which may be able to offer advice and information about your legal status and benefits available.



What if I am struggling with my studies/work?

If you are struggling at college/university or work, first talk to your healthcare professional to ensure that your medical treatment is optimised.

There are various adjustments that your institution could make to help make things easier for you.

College/University

You may need to talk to your tutors about managing your workload and meeting deadlines for assignments.

It may help to take regular breaks to rest if you feel tired during the day.

If you are receiving ERT, you may need to take time off and miss lessons. Talk to your tutors about catching up on anything you may miss. They may be able to make adjustments to your study schedule.

Work

You may need to talk to your employer about changing some of your responsibilities or working hours.

It may help to take regular breaks to rest if you feel tired during the working day.

If you are receiving ERT, you may need to take time off work for infusions. However, it may be possible to have infusions at home to minimise the impact on your working week. Speak to your healthcare professional about receiving therapy at home.

Talk to your employer as they may be able to make adjustments to your role so that you can stay at work, including reducing working hours and allowing paid leave to attend medical appointments. Patient associations may be able to advise you on your legal entitlements.

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.¹⁵

CT (computed tomography)

Also known as a CAT scan, helps to get a better picture of how your bones and organs are affected and can be used to measure liver and spleen volume.¹⁶

DEXA scan

Dual energy X-ray absorptiometry (DEXA) scans measure bone mineral density. DEXA scans are typically used to diagnose osteoporosis.¹⁰

Gaucher

Gaucher (pronounced: go-shay) is a rare lysosomal storage disorder.² It is named after a French dermatologist Dr Phillippe Gaucher who first described the disease over 100 years ago.²

Glucocerebroside

Glucocerebroside is a lipid (fatty substance), which builds up in certain cells in people with Gaucher.¹⁵

Glucocerebrosidase

The enzyme that breaks down glucocerebroside. In people with Gaucher, glucocerebrosidase is either missing or only produced in small amounts.¹⁵

Haemoglobin

Haemoglobin is the protein within the red blood cells that helps carry oxygen around your body, and is responsible for the red colour.¹⁵ Low haemoglobin levels (anaemia) cause tiredness as less oxygen is getting to where you need it.¹⁵

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.¹⁷

Macrophages

A type of white blood cell that removes dead cells and helps to fight infection.¹⁵ In people with Gaucher, lipids accumulate within these cells.⁶

MRI (magnetic resonance imaging)

Scan of body to help monitor bones and organs in Gaucher. Can be used to assess liver and spleen size and monitor the response to treatment.¹⁰

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.⁴

Neutropenia

Low level of neutrophils (white blood cells) in the blood, which can lead to an increased risk of infection.¹⁵

Parkinson's disease

A chronic, degenerative neurological disorder affecting movement. It is characterised by tremor and loss of co-ordination and balance.¹⁵

Platelets

Platelets help your blood to clot.¹⁵ Low levels of platelets (thrombocytopenia) in your blood make it easier for you to bruise and more difficult for you to stop bleeding.¹⁰

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.¹⁵

Spleen

The spleen is an organ that helps to break down old red blood cells and to fight infection.¹⁵

Splenectomy

An operation to remove the spleen.¹⁵

Thrombocytopenia

Low level of platelets in blood, leads to increased tendency for bruising and bleeding.^{10,15}

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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