SUPPORTING SOMEONE WITH 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 friend, partner or family member.

Finding out that someone close to you has Gaucher (pronounced "go-shay") can be extremely worrying, particularly if it is a family member. If no one else in the family has it, diagnosis with Gaucher can come as a bit of a shock.

You will probably have lots of questions about what will happen to them in the future, and whether other family members will develop the condition.

It is important to try and understand what they are going through, as they will probably require emotional support.

ABOUT GAUCHER

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).[2] There are 3 main types of Gaucher disease:

Type 1 (Non-neuronopathic) – the most common type[2] with variable symptoms and progression[3]

Type 2 (Acute neuronopathic) – an acute, severe form affecting the central nervous system in babies and infants[3]

Type 3 (Chronic neuronopathic) – a chronic form affecting the central nervous system, that progresses over time[4]

Who gets Gaucher?

Gaucher is a rare genetic disorder affecting around 1 in 100,000 of the population.[5] Most 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 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 notice that the person close to you gets tired easily and cannot do everything that they used to. They may need to change their daily schedule and pace themselves more.

If you have a child with Gaucher, they may bruise easily so will need to take extra care when playing and at school.

The most common clinical symptoms 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 symptoms may include:[3,7]

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

Can it be treated?

There is currently no cure for Gaucher, but different therapies are available which can help to treat many of the major symptoms.[9] Doctors and the rest of the healthcare team will be able to provide further information and advice on the treatments available. 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.[9] Long-term treatment has been shown to reduce the size of the liver and spleen and improve red blood cell and platelet counts.[9]

ERT will not treat the neurological symptoms of Gaucher.[3] 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]

Substrate Reduction Therapy

Substrate Reduction Therapy (SRT) reduces the production of lipids within the cells to help minimise symptoms.[3] It is currently recommended in patients who are unsuitable for ERT.[10]

Other treatments

Depending on individual symptoms people may need treatment for bone complications and/or medications for pain.

Medicines may be given to people with weakened bones, to help slow down loss of bone mass and encourage growth of new bone cells.[3] If bones or joints become badly damaged or fractured, surgery may be required.

People with chronic or acute episodes of pain can discuss pain management options with their healthcare professional.

Will it get worse?

Progression is variable and effective treatments are available which 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).[1,8] 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]

GAUCHER IN THE FAMILY

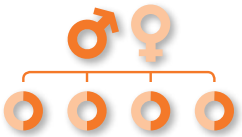
How is it passed on within families?

If you have found out that a family member has Gaucher, you may be concerned about how this will affect you and the rest of your family.

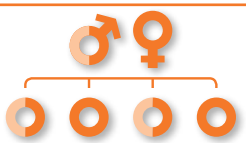
Gaucher is an inherited disorder that is said to be 'recessive'.[6] This means that a child needs to inherit a copy of the faulty gene from both parents to have Gaucher. See the next page for a clearer picture of how Gaucher is passed on in families

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]

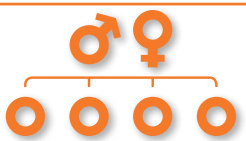
Here is a guide on how Gaucher is passed on within families:



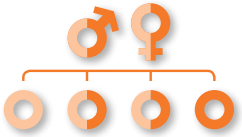
If one parent has Gaucher but the other does not, the child will be a 'carrier' of the condition but will not have Gaucher[11]



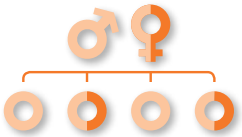
If one parent has Gaucher and the other is a carrier, there is a 1 in 2 chance that the child will also have Gaucher[11]



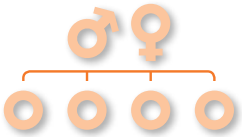
If both parents have Gaucher, their children will also have the condition[11]



If both parents are carriers but do not have Gaucher, there is a 1 in 4 chance that their child will have Gaucher[11]



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 Gaucher[11]



If neither parent is a carrier, Gaucher cannot be passed on to their children[11]


No Gaucher


Gaucher carrier


Has Gaucher

GAUCHER AT WORK

Gaucher at work

Many people receiving treatment for Gaucher are able to maintain normal working lives. However, symptoms can prevent them from carrying out some activities.

If one of your employees or colleagues has Gaucher, you may need to make some allowances for them and make adjustments to their daily routine:

- Tiredness is one of the key symptoms of Gaucher. It may help them to take regular breaks, to rest, if they are constantly feeling tired during the day
- They may lack the energy to carry out some tasks, particularly if the job is physically demanding
- If they are still struggling with work, they may need to reduce their working hours
- Look out for the signs, as they may find it difficult to speak up and ask for help
- If they are receiving ERT, they may need to take time off work regularly for infusions or other medical appointments

Your human resources department will be able to give you advice and provide you with more information on the legal aspects.

ADDITIONAL RESOURCES

Gaucher dictionary

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.[12]

Glucocerebrosidase
The enzyme that breaks down glucocerebroside. In people with Gaucher, glucocerebrosidase is either missing or only produced in small amounts.[12]

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.[13]

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.[4]

Parkinson's disease
A chronic, degenerative neurological disorder affecting movement. It is characterised by tremor and loss of co-ordination and balance.[12]

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.[12]

Spleen
The spleen is an organ that helps to break down old red blood cells and to fight infection.[12]

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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