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About Gaucher Disease

This booklet is for adults with Type 1 Gaucher Disease who have been prescribed Cerdelga.

The information in this booklet is not intended to replace the Patient Information Leaflet accompanying your medication and should act as further information. If you have any questions or concerns regarding your treatment, please consult your Patient Information Leaflet, or get in touch with your specialist healthcare team.

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Further information on Gaucher Disease is available on the Gauchers Association website: www.gaucher.org.uk

What is Gaucher Disease?

Gaucher Disease occurs when a person doesn't have enough of an enzyme called β-glucosidase, which breaks down a fatty substance called glucosylceramide (GL-1).

This means that GL-1 can gather in places like your spleen, liver, bone marrow and sometimes in your lungs (although this is rare), leading to symptoms of Gaucher Disease. It is therefore important to get the right treatment as soon as possible to help manage these symptoms properly.

There are three different types of Gaucher Disease; Type 1, Type 2 and Type 3. Each type is slightly different in terms of when you start to have symptoms and what symptoms you have. The booklet will focus on Type 1 Gaucher Disease.

Enzyme: A biological molecule that helps speed up chemical reactions in your body.

β-glucosidase : An enzyme in your body that breaks down GL-1.

Glucosylceramide (GL-1): A type of fat that is used as a building block to help make your cells.

Type 1 Gaucher Disease

You have been diagnosed with Type 1 Gaucher Disease, which is the most common form of Gaucher Disease. Anyone of any age can be affected.

Listed below are some of the signs and symptoms you may experience with Type 1 Gaucher Disease:

General symptoms:

- *Fatigue (tiredness)*
- *Lack of energy and stamina*
- *Delayed onset puberty*

Abdomen (tummy):

- *Enlarged liver and spleen*

Skeletal:

- *Bone pain*
- *Reduced bone density*
- *Widening of bones above the knee joint*
- *Spontaneous bone breaks (fractures)*
- *Spontaneous 'bone crisis', which can cause severe bone pain*
- *Bone necrosis (death of tissue)*

Blood:

- *Increased bleeding tendency, such as more nosebleeds and bruises*
- *Low levels of:*
  - *Blood platelets (thrombocytopenia)*
  - *Red blood cells (anaemia)*
  - *White blood cells (neutropenia)*

Digestive:

- *Loss of appetite*

Respiratory:

- *Shortness of breath/breathing difficulty*

How is Gaucher Disease inherited?

An individual inherits two copies of every gene - one from each parent. In Gaucher Disease, the gene that produces the enzyme β-glucosidase (that we talked about on page 2 of this booklet) does not work properly.

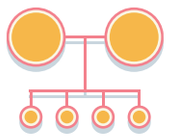
To develop Gaucher Disease you must have two copies of the faulty gene. A person who has one normal and one mutated gene is a carrier or Gaucher Disease. They do not have the condition but there is a 50% chance that they will pass on the faulty Gaucher gene to their children.

Genes: Are the inherited information passed on from parents to their children. They have a big role in what we look like and who we are, as they carry information like what colour hair you have and how tall you might be.

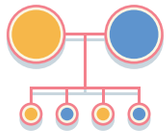
Interested in carrier testing for your family?
Discuss the possibility with your healthcare team.

The possibilities of passing on the mutated Gaucher gene:

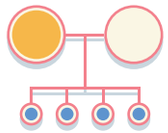
Legend — Gaucher Disease — A carrier — Not affected



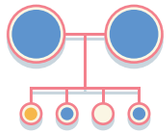
Both parents have Gaucher Disease. All their children will inherit two Gaucher genes – one from each parent. All children have Gaucher Disease.



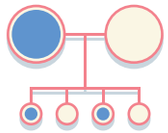
One parent has Gaucher Disease and the other parent is a carrier. Their children will have a 50% chance of having Gaucher Disease and a 50% chance of being a carrier.



One parent has Gaucher Disease and the other parent is not affected. All their children will inherit the Gaucher gene from the affected parent and become carriers, but none of the children will have Gaucher Disease.



Both parents are Gaucher carriers. Their children have a 50% chance of being a carrier, a 25% chance of having Gaucher Disease and 25% of being neither.



One parent is a Gaucher carrier (one mutated gene and one normal), and the other parent has two normal genes and is therefore not affected. Their children have a 50% chance of being a carrier but none of their children will have the disease.

Checking and monitoring Gaucher Disease

Your healthcare team will arrange regular tests/ investigations to assess your Gaucher Disease and to check that your treatment is working effectively for you. The more they understand how Gaucher affects you, the more they will be able to prevent/ treat problems that may occur. These tests commonly include blood tests, MRIs and X rays.

For more information about these tests, see the Gauchers Association website at: www.gaucher.org.uk. If you do not understand the tests your doctor has suggested for you, do ask them to explain it to you.

At each visit to your specialist centre the healthcare team will ask you lots of questions to see how you are. They may ask you to complete questionnaires about your health, undertake a physical examination (to feel your liver or spleen for example) or look at your eye movements.

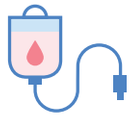
You may be invited to participate in relevant research to help improve understanding of Gaucher Disease.

Treating Type 1 Gaucher Disease

There is currently no cure for Gaucher Disease, but there are different therapies and treatments available which can help you to manage the symptoms.

With treatment, many people with Gaucher can lead full lives and are able to get on with many of their normal daily activities. You and your healthcare team will have discussed which treatment is right for you and made the decision that Cerdelga is suitable at this time. Below is a list of some other therapies available for Type 1 Gaucher Disease patients. If you have any questions about these or your treatment, please speak to your healthcare team.

For those that require treatment, the options are:



Enzyme replacement therapy (ERT)
Gaucher Disease occurs when a person doesn't have enough of an enzyme called β-glucosidase. Enzyme Replacement Therapy (ERT) looks to replace this enzyme. This is given by infusion directly into your vein.

Substrate reduction therapy (SRT)
This treatment reduces the amount of fatty substances created in the cell and therefore helps to reduce build up. This is taken as an oral capsule. SRTs are not suitable for everybody and your doctor will advise if they are right for you.

Treating Type 1 Gaucher Disease

Bisphosphonate
For patients with Gaucher Disease who have low bone density and experience fractures, the bisphosphonate group of drugs (e.g. oral alendronate or IV zoledranate) are often prescribed. These help combat symptoms of Type 1 Gaucher Disease like osteoporosis and bone disease.

Other
For those people who have had their spleen removed long-term antibiotic therapy and up-to-date vaccinations are essential.

Treatments for bone pain
If you experience bone pain, talk to your healthcare team about pain management - it may be helpful if you keep a pain diary. If it is necessary for you to undergo orthopaedic surgery, it is essential that this is planned in conjunction with your specialist centre. Refer to **Living well with Gaucher Disease** booklet for more information about Gaucher Disease diaries.

For up-to-date information on clinical trials currently being undertaken in patients with Gaucher Disease, see the Gauchers Association website at: www.gaucher.org.uk. If you have any concerns or questions regarding Gaucher Disease, please do not hesitate to talk to your healthcare team.

Notes

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You can refer to this booklet at any point in your treatment journey, for information on Gaucher Disease.

If you find you have more questions, there is a Patient Information Leaflet included in the packaging of your Cerdelga tablets. You can also speak to your doctor, nurse or pharmacist.

▼ This medicine is subject to additional monitoring. This will allow quick identification of new safety information. You can help by reporting any side effects that you may get. In the UK, see www.mhra.gov.uk/yellowcard for how to report side effects. In Ireland, see www.hpra.ie for how to report side effects.

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