Cerdelga® ▼
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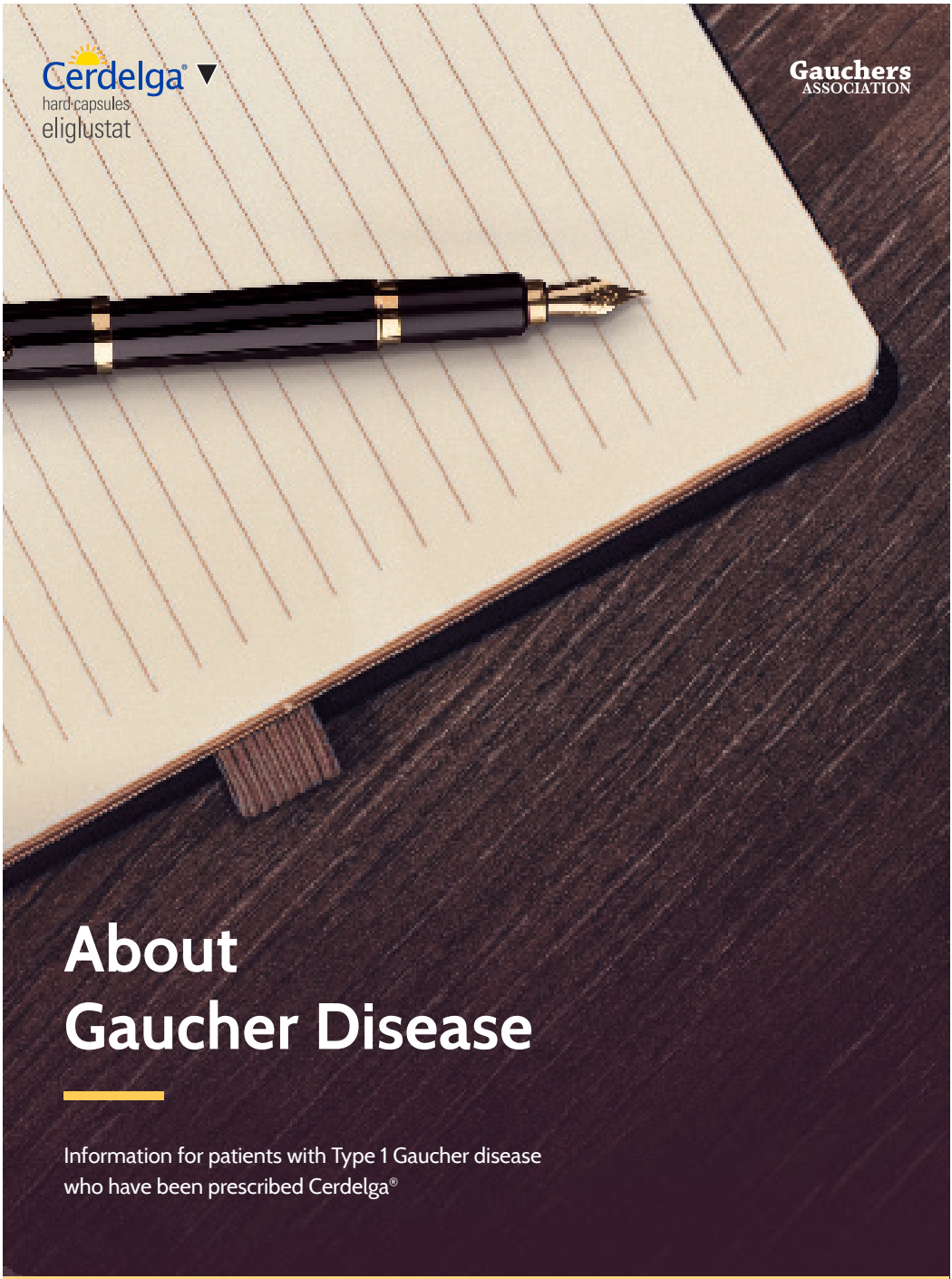
Gauchers ASSOCIATION



About Gaucher Disease

Information for patients with Type 1 Gaucher disease who have been prescribed Cerdelga®

This information has been produced in collaboration with The Gauchers Association

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SANOFI GENZYME

▼ This medicine is subject to additional monitoring. This will allow for quick identification of new safety information. You can help by reporting any side effects you may experience. If you get any side effects talk to your doctor, pharmacist or nurse. This includes any possible side effects not listed in the package leaflet. You can also report side effects directly via the Yellow Card Scheme at www.mhra.gov.uk/yellowcard.

What is Gaucher Disease?

What is Gaucher Disease?

Gaucher disease occurs when a person doesn't have enough of an enzyme called glucocerbrosidase, which breaks down a type of fat called glucocerbroside.

This means that this fat can gather in places like your spleen, liver, bone marrow and sometimes in the lungs (although this is rare). As a result of this fat gathering, people with Gaucher disease often experience symptoms such as bone pain, an enlarged liver or spleen, a protruding stomach, tiredness and an increased risk of fractures - so it's important to get the right treatment as soon as possible to help manage these symptoms properly.

For further information on neuronopathic Gaucher disease is available on the Gauchers Association website:

www.gaucher.org.uk

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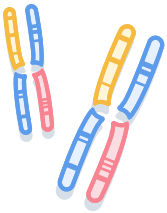
Type I
Gaucher Disease



Type 1 Gaucher Disease

You have been diagnosed with Type 1 Gaucher disease, which is the most common form of Gaucher disease and anyone of any age can be affected. Many people who have Type 1 Gaucher disease continue to lead normal lives with very few symptoms.

Signs and symptoms of Type 1 Gaucher disease:

General symptoms

- ***Fatigue/tiredness***
- ***Lack of energy and stamina***

Abdomen

- ***Enlarged liver and spleen***

Skeletal system

- ***Bone pain***
- ***Reduced bone density***
- ***Widening of bones*** *above the knee joint*
- ***Spontaneous fractures***
- ***Acute bone infarctions*** *-"bone crisis" e.g. severe pain*
- ***Bone necrosis*** *(death of tissue)*

Blood

- ***Increased bleeding tendency*** *such as nosebleeds and bruising*

  Low levels of:

  - ***Blood platelets*** *(thrombocytopenia)*
  - ***Red blood cells*** *(anaemia)*
  - ***White blood cells*** *(neutropenia)*

Digestive

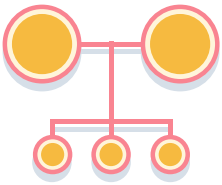
- *Loss of appetite*

Respiratory

- ***Shortness of breath***

Delayed onset of puberty

How is Gaucher Disease inherited?



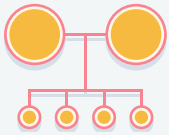
How is Gaucher Disease inherited?

An individual inherits two copies of every gene – one from each parent. In the case of Gaucher disease, the gene that produces glucocerebrosidase is faulty.

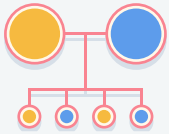
To develop Gaucher disease a person must have two copies of that faulty gene (often referred to as a 'mutation'). A person with one normal and one mutated gene is a "carrier" of Gaucher disease and will not have the condition, but there is a 50% chance that they will pass the "Gaucher gene" onto their children.

The possibilities of passing on the mutated Gaucher gene:

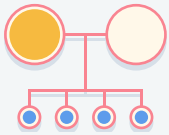
Legend Gaucher disease · A carrier · Not affected



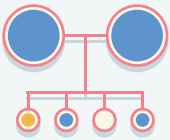
Both parents have Gaucher disease. All their children inherit two Gaucher genes – one from each parent, therefore all children have Gaucher disease.



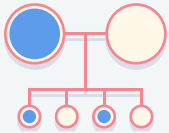
One parent has Gaucher disease and the other parent is a carrier. Their children will have a 50% chance of having Gaucher disease and a 50% chance of being a carrier.



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



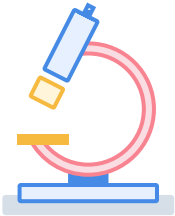
Both parents are Gaucher carriers. Their children have a 50% chance of being a carrier and a 25% chance of having Gaucher disease and 25% of being neither.



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.

Many of the mutations of the gene for Gaucher disease have been identified; and carrier testing is possible in affected families (if you are interested, discuss this with your specialist centre).

Checking and Monitoring Gaucher Disease



Checking and Monitoring Gaucher Disease

Your specialist team will arrange regular tests/investigations to assess your Gaucher disease and to check that your treatment is working effectively for you. The more your clinical team 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 clinical 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/spleen or assess how well your nerves are working for example) or look at your eye movements. You may be invited to participate in relevant research to help improve understanding of the disease.

Is there a cure?

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Is there a cure?

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

With treatment, many people with Gaucher can lead full lives and are able to carry out many of their normal daily activities. You and your doctor 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 treatments available for Gaucher Type 1 patients, but if you have any questions about these or your treatment, please speak to your doctor.

For those that do require treatment, the options are:

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

Substrate reduction therapy (SRT)
This treatment reduces the amount of fatty substances in your cells and therefore helps to reduce build up. This is administered by an oral capsule. These products are not suitable for everybody and your specialist doctor will advise if they are right for you.

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 to help combat 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.

Bone Pain

If you experience bone pain, talk to your doctor 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.

For up-to-date on clinical trials currently being undertaken in patients with Gauchers 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

Notes