SIGNS AND SYMPTOMS OF GAUCHER DISEASE

Gaucher disease is a rare inherited (genetic), enzyme deficiency disorder. Symptoms range from mild to severe and can appear at any time, from infancy to old age. They may include anaemia (low haemoglobin), tiredness (fatigue), easy bruising and a tendency to bleed. An enlarged spleen and liver with a protruding stomach may also occur as well as bone pain, loss of bone strength and density with an increased risk of fractures.

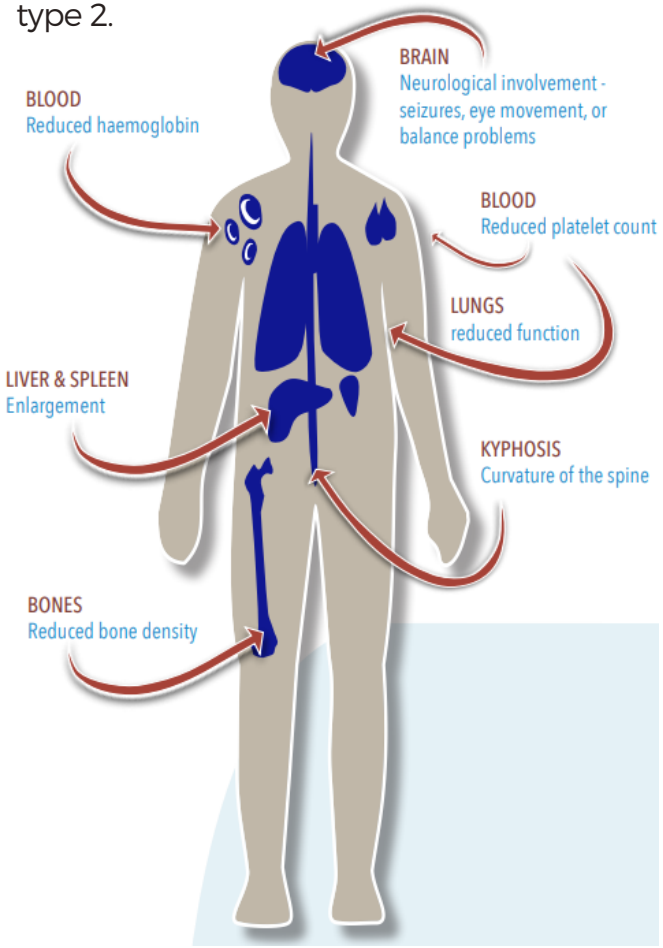
People with Gaucher disease lack sufficient activity of an enzyme called glucocerebrosidase. This enzyme helps the body to break down worn-out cells and as a result, a fatty substance called glucocerebroside accumulates usually in the spleen, liver, bone marrow, rarely in the lungs and in some types of Gaucher disease in the central nervous system.

Type 1 Gaucher disease is the most common form affecting 1 in 100,000 of the general population. Many people with type 1 Gaucher disease have no symptoms and lead normal lives.

Scan to download our full information booklet to learn more or visit bit.ly/gdinfor

Type 2 Gaucher disease is a very rare, rapidly progressive form of Gaucher disease which affects the central nervous system as well as the spleen, liver, lungs and bones.

Type 3 Gaucher disease, or chronic neuronopathic Gaucher disease, is intermediate between types 1 and 2. Patients have both visceral (lungs, liver and spleen) and neurological (brain) involvement. However, the neurological involvement is much less severe than in type 2.



INTERNATIONAL GAUCHER DAY
1 OCTOBER
International Gaucher Alliance

The power of knowledge in a timely diagnosis

Building a better future

GAUCHER DISEASE SIGNS AND SYMPTOMS

HOW IS GAUCHER DISEASE DIAGNOSED?

IGA INTERNATIONAL GAUCHER ALLIANCE
Driven by passion for patients

HOW IS GAUCHER DISEASE DIAGNOSED?

The process of diagnosing many diseases, and especially Gaucher disease, is not always straightforward.

Often, the patient initially visits their doctor for another problem such as the flu, for non-specific pain, or for a routine check-up. Although making a diagnosis of Gaucher disease is not difficult, some symptoms may resemble other diseases. The doctor may first perform other tests to eliminate more common disorders.

For example, in cases where patients have low platelet counts, doctors may first test for leukaemia. If a patient complains of joint pain, the doctor may first suspect arthritis. Sometimes specialists at a genetics unit, a haematologist or a metabolic physician, may be helpful in distinguishing the symptoms of Gaucher disease from other diseases with similar symptoms.

Gaucher disease can be diagnosed by a simple blood test – by measuring the amount of enzyme in your blood and checking for mutations in the glucocrebrosidase gene.

Gaucher disease might be suspected in a person who has an unexplained enlargement of the spleen, tendency toward bleeding, bone or joint pains or spontaneous fractures.

A PAEDIATRICIAN might make the diagnosis in a child complaining of abdominal discomfort or of frequent nosebleeds.

A HAEMATOLOGIST might make the diagnosis in a person with low blood or platelet counts.

AN ORTHOPAEDIC DOCTOR might diagnose Gaucher disease in the course of treating someone suffering with frequent unexplained fractures.

Gaucher disease would be particularly suspected in people with family members who are known to have the disease.

The International Gaucher Alliance is an umbrella organisation representing patient groups worldwide.

To see if there is a group in your country, and for more general information about Gaucher disease, visit our website at

www.gaucheralliance.org

IGA INTERNATIONAL GAUCHER ALLIANCE
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The IGA is a patient led international organisation that has become the 'go to' global voice for over 90% of the worldwide Gaucher community and has built its reputation through listening to and delivering outcomes that have impacted on patients and their carers' lives. If you need to find your local patient group, link to one of our 50+ member associations for local support, advice and news, visit gaucheralliance.org/member-directory.

OUR VISION:
A world where all Gaucher patients have access to the treatment and care they need and there is a possibility of a cure

OUR VALUES:
We are: patient focused, trustworthy, collaborative, visionary, passionate

OUR MISSION:

- to empower its members
- to advocate on behalf of Gaucher patients
- to ensure that the Gaucher research agenda is focused on patients' unmet needs
- to take collective action to address challenges Gaucher patients worldwide face in accessing early diagnosis and optimal treatment and care.