

# Understanding Gaucher Disease

A guide for patients, parents, relatives, and friends



Maria,  
living with  
Gaucher disease.

## What's Inside This Guide?

If you're newly diagnosed or if you've been living with Gaucher disease for a while, this guide provides valuable information for you, your family, and your friends.

-  **Overview of Gaucher disease**
-  **Different types of Gaucher disease**
-  **Possible symptoms**
-  **Getting tested and receiving a diagnosis**
-  **Early management**

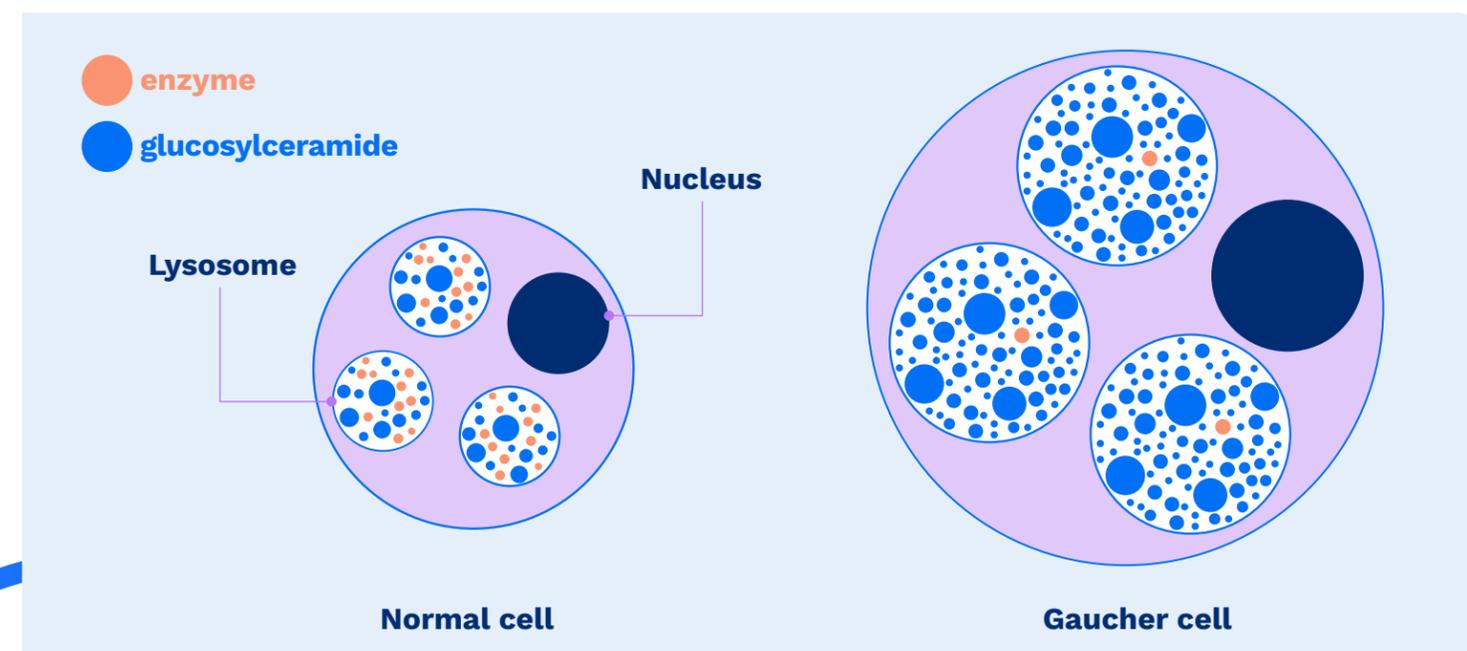
## What is Gaucher Disease?

**Gaucher (pronounced *go-shay*) disease is a rare, progressive, inherited condition that affects bone, liver, spleen, and blood health, and in some patients, brain health**

- A person inherits Gaucher disease through a gene change from both parents
- Gaucher disease gets worse over time, potentially damaging internal organs and bones
- However, finding and treating Gaucher disease early can help prevent this damage

### **Gaucher disease occurs when people don't have enough of a specific enzyme**

- The signs and symptoms of Gaucher disease are caused by accumulation of a naturally occurring substance called **GL-1 (glucosylceramide)** and its byproduct lyso-GL-1
- In healthy lysosomes (part of cells that contains digestive enzymes), an enzyme called **glucocerebrosidase** breaks down GL-1 into glucose and ceramide so it can be cleared from the cell
- However, people with Gaucher disease don't have enough of this enzyme, causing GL-1 and lyso-GL-1 to build up within lysosomes and their bodies



**If a family member has been diagnosed with Gaucher disease, it is important for relatives, especially siblings, to consider getting tested. Even if they do not have Gaucher disease, they may be a carrier for the genetic change that causes it.**

## What Are the Different Types of Gaucher Disease?

Different types of Gaucher disease vary in when symptoms start, how quickly they progress, and what parts of the body are affected

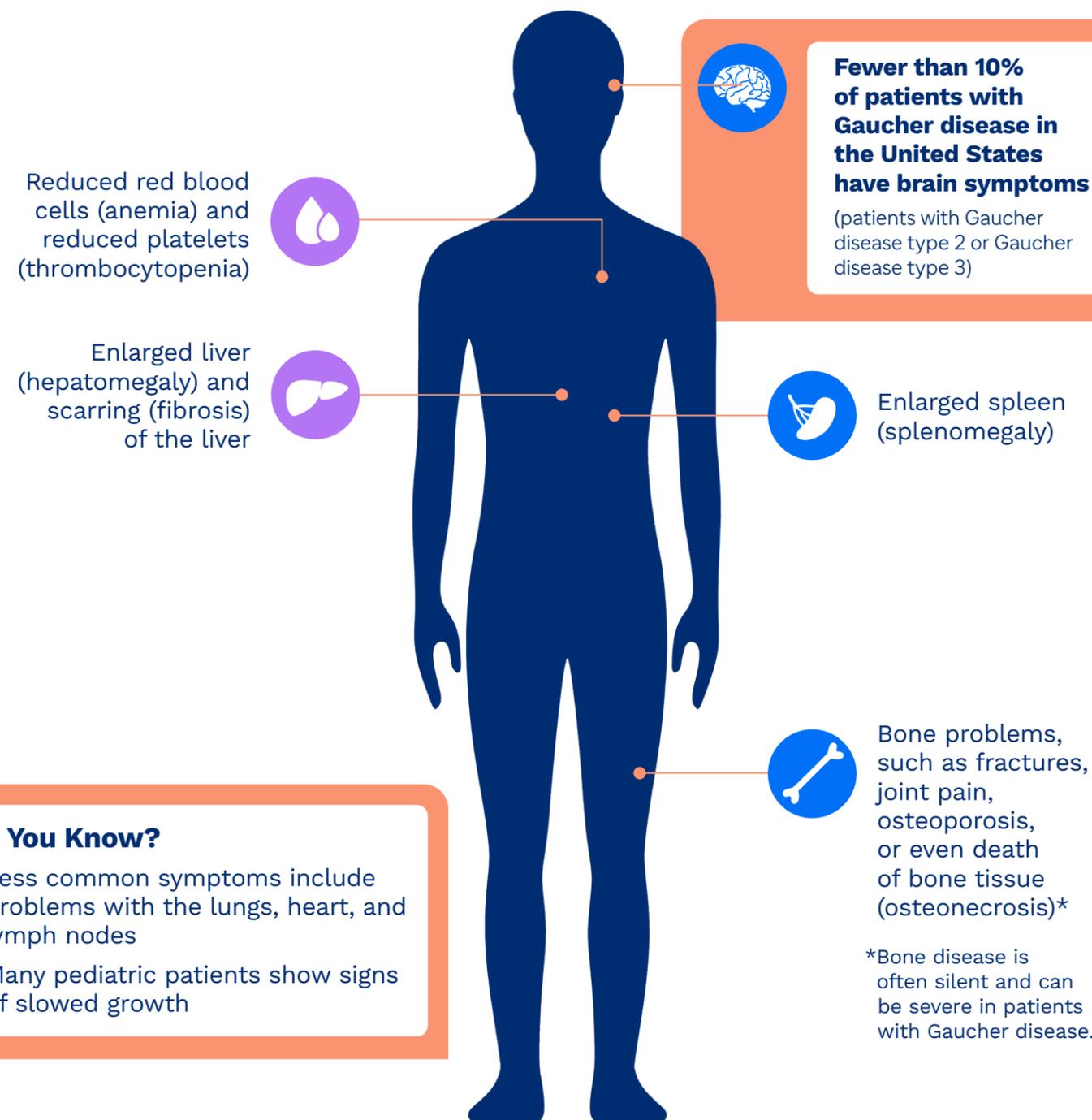
|                | Type 1   | Type 2  | Type 3  |
|----------------|--|---|---|
| Incidence      | ~1 in 40,000<br>~1 in 500 to 1000 in people with Ashkenazi Jewish heritage | ~1 in 100,000   | ~1 in 100,000   |
| Onset          | Childhood or adulthood   | Infancy (usually do not live past 2 years of age)                             | Typically in childhood  |
| Impact on body | Liver, spleen, blood, and bone symptoms                                    | Liver, spleen, blood, and rapidly progressing central nervous system symptoms | Liver, spleen, blood, bone, and central nervous system symptoms |

**There are no FDA-approved treatments for Gaucher disease type 2 or Gaucher disease type 3**

## What Are the Symptoms ?

**Gaucher disease symptoms are diverse, unpredictable, and variable**

- The signs and symptoms of Gaucher disease may be different from one person to another, even for people within the same family



### Did You Know?

- Less common symptoms include problems with the lungs, heart, and lymph nodes
- Many pediatric patients show signs of slowed growth

\*Bone disease is often silent and can be severe in patients with Gaucher disease.

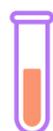
## Why is **Early Diagnosis** Important?



### **Patients can wait an average of 7 years for an accurate diagnosis of Gaucher disease**

Delayed diagnosis may be due to reasons such as lack of awareness about Gaucher disease, non-specific or mild symptoms, misdiagnosis of another condition, and how differently Gaucher disease can appear from patient to patient.

## How to get tested



Gaucher disease can be detected with a blood test.

- Enzyme testing is the best way to establish a diagnosis of Gaucher disease
- Genetic testing can confirm the diagnosis and may provide additional details about the disease

## Ongoing **Monitoring** of Symptoms

**Timely disease monitoring remains important, as damage caused by Gaucher disease may be irreversible**



Every patient with Gaucher disease should have a comprehensive evaluation of their blood, organs (including lungs), bone, and brain health when they are diagnosed and regularly to assess if the condition is getting worse or if a change in management is needed.

- Bone disease can be detected early with careful monitoring using MRI, X-ray, or other imaging techniques
- Regular neurological exams are important because brain-related symptoms may appear several years after other symptoms

**Talk with your healthcare team about what tests are right for you**

## Why is **Early Management** Important?



Managing Gaucher disease early can help patients potentially avoid blood transfusions and spleen removal, and may help reduce the discomfort caused by organ symptoms.



Early intervention can help avoid permanent organ damage, such as end-stage liver disease.



The risk of bone death is lower in patients who start managing Gaucher disease within 2 years of diagnosis.



**Alejandro,**  
living with  
Gaucher disease.

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