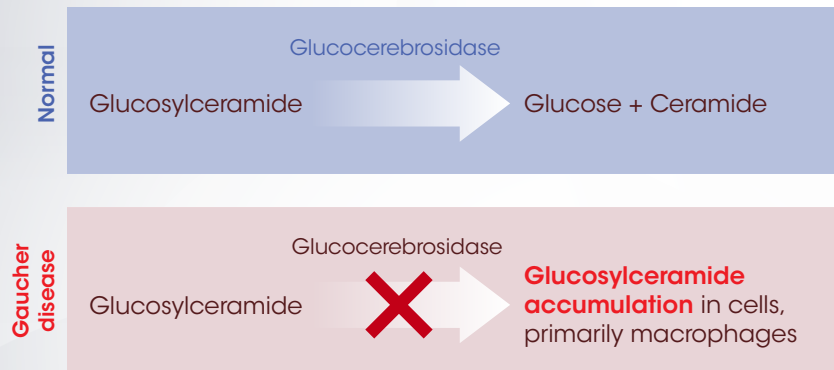


# ABOUT GAUCHER DISEASE

Gaucher disease is a lysosomal storage disorder caused by a deficiency of the enzyme glucocerebrosidase (acid  $\beta$ -glucosidase).<sup>1</sup>



The levels of glucosylceramide and other glycolipids may be **increased 20-100 times**, leading to clinical manifestations of Gaucher disease.<sup>2</sup>

## TYPES OF GAUCHER DISEASE<sup>1,3</sup>

### Type 1

Chronic  
Non-neuronopathic

**Most common**

Causes significant morbidity from fractures, bone pain, cytopenia secondary to hypersplenism and eventual death from bleeding or infection

### Type 2

Acute  
Neuronopathic

**Rare**

Most severe form, rapidly fatal by 2 years of age

### Type 3

Intermediate progression  
Variable central nervous system involvement, and severe visceral involvement

**Rare**

Some patients are fully functional and able to work, while others may have significant impairments

## GAUCHER DISEASE IS THE MOST COMMON LYSOSOMAL STORAGE DISEASE<sup>2</sup>

**Prevalence of Gaucher disease varies by type<sup>4</sup>:**

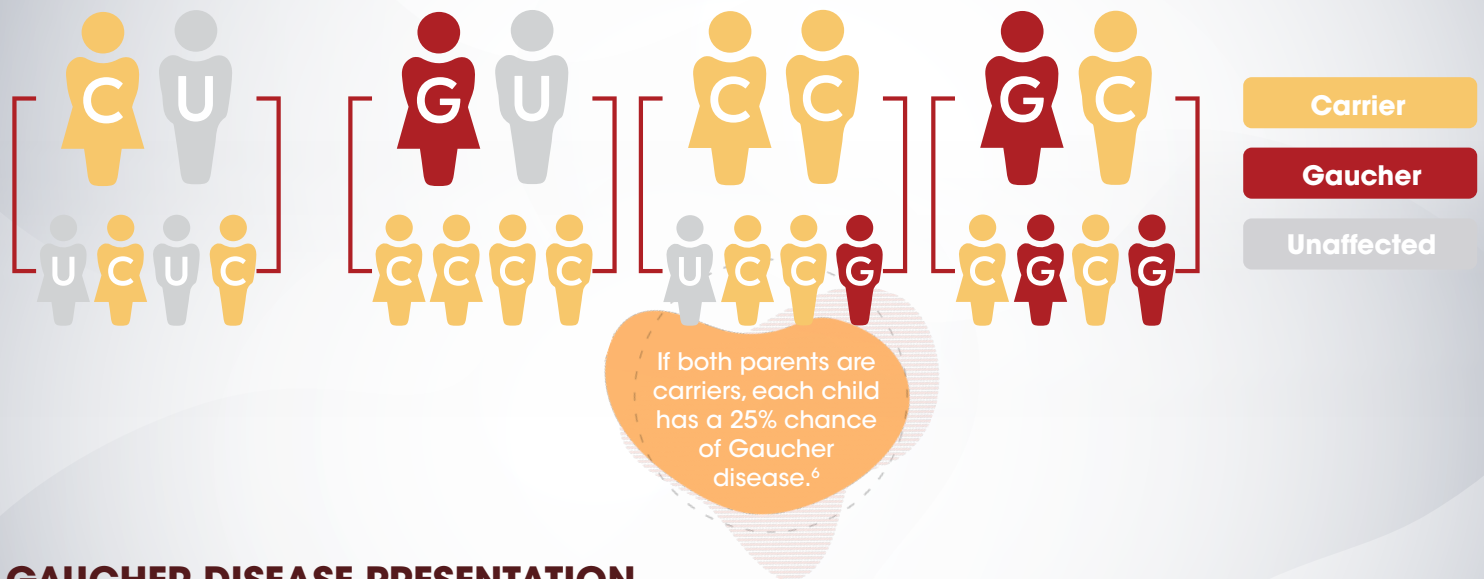
Type 1	Type 2	Type 3
1/20,000–1/40,000	1/100,000	1/100,000

Gaucher disease is far more common among people of Ashkenazi Jew descent (Eastern European)<sup>5</sup>:

**1:450**

# GAUCHER DISEASE IS GENETICALLY INHERITED

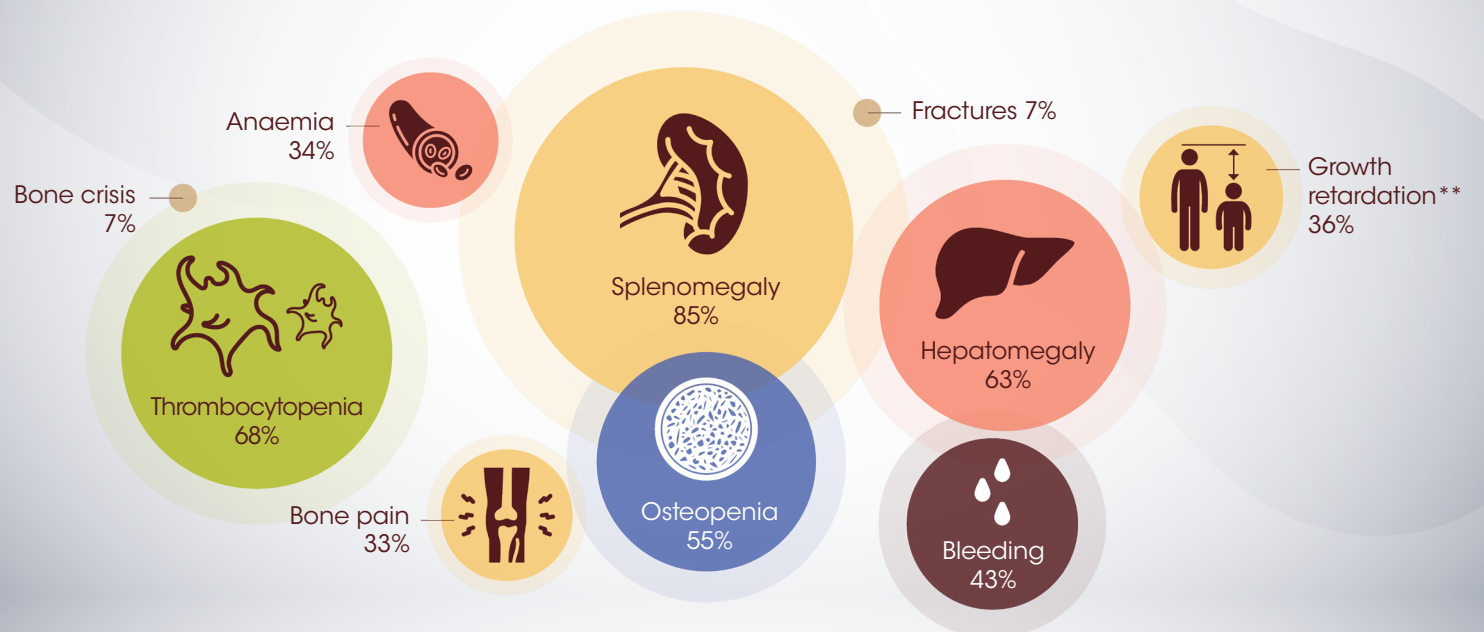
Gaucher disease is an **autosomal recessive disorder**, caused by mutations in the glucocerebrosidase gene located on chromosome 1q21.<sup>2</sup>



## GAUCHER DISEASE PRESENTATION

Gaucher disease has a variety of presenting features that may occur at any age, with varying severity.<sup>2</sup>

These features include (with percentage at presentation)<sup>\*2,7</sup>:



\*Percentage in the general patient population.  
 \*\*Applicable for children.

### For healthcare professionals only

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sanofi-aventis (Malaysia) Sdn Bhd (334110-P), Unit TB-18-1, Level 18, Tower B, Plaza 33, No. 1 Jalan Kemajuan, Seksyen 13, 46200 Petaling Jaya, Selangor Darul Ehsan, Malaysia.  
 Tel: 03 7651 0800, Fax: 03 7651 0805.

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