Information for Hematologist-Oncologists and Orthopedists

GAUCHER disease


Not an actual patient.

## GAUCHER DISEASE TYPE 1

# Identification and differential diagnosis

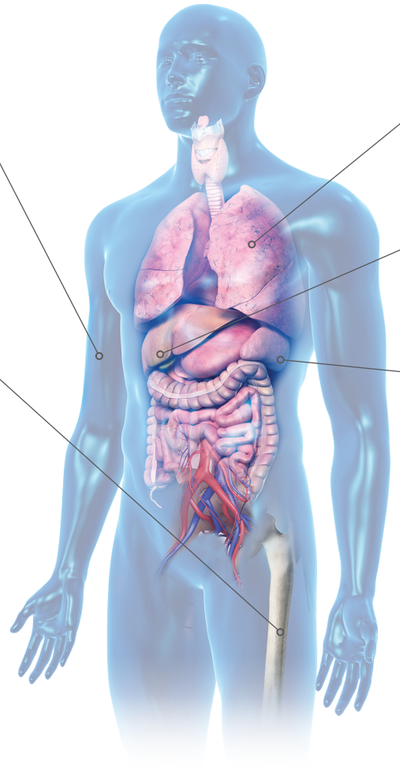
# Gaucher disease type 1: A rare, progressive disease that can go undiagnosed for up to 10 years[1]

**Gaucher disease** is caused by the accumulation of glucocerebroside (GL-1) in cells of monocyte/macrophage lineage.[1,2]

- Gaucher disease type 1 is pan-ethnic, with a frequency of ≈1:40,000 in the general population. However, in patients of Ashkenazi Jewish ancestry, the frequency is ≈1:850[3]

› GL-1 buildup can lead to progressive multiorgan dysfunction with lifelong consequences.[1]

## SYMPTOM PREVALENCE IN PATIENTS WITH GAUCHER DISEASE TYPE 1



### CYTOPENIAS[4]

Anemia: **64%**

Thrombocytopenia: **56%**

### BONE PATHOLOGIES[4]

Bone Pain: **63%**

Bone Crisis: **33%**

Erlenmeyer Flask Deformity: **46%**

Osteopenia: **42%**

Avascular Necrosis: **25%**

Fracture: **15%**

Joint Replacement: **8%**

### PULMONARY PATHOLOGIES[5]

**1% to 2%**

### HEPATOMEGALY[4]

**79%**

### SPLENOMEGALY[3]

**87%**

› Gaucher disease can also lead to a ***reduced quality of life, a shortened lifespan***, and an ***increased risk of cancer***.[1,3]

# Include Gaucher disease in your differential diagnosis

## GAUCHER DISEASE COMMONLY MIMICS THE SIGNS AND SYMPTOMS OF MANY HEMATOLOGICAL MALIGNANCIES[3,6-23]

| | Gaucher disease type 1 | ASMD* Type B | Acute lymphoblastic leukemia | Multiple myeloma | Non-Hodgkin lymphoma | Chronic myeloid leukemia | Hairy cell leukemia | Myelofibrosis |
|---|---|---|---|---|---|---|---|---|
| AGE OF ONSET (YEARS) | 0-80 | Infancy to adulthood | Usually children under 5/ adults >60 | Usually 65-70 | Usually ≈70 | Usually ≈50 | Usually ≈50 | Usually >50 |
| SPLENOMEGALY | ● | ● | ● | Less common | ● | ● | ● | ● |
| HEPATOMEGALY | ● | ● | ● | Less common | ● | ● | ● | ● |
| BRUISING/ BLEEDING | ● | ● | ● | ● | ● | ● | ● | ● |
| FATIGUE | ● | ● | ● | ● | ● | ● | ● | ● |
| BONE PAIN | ● | ● | ● | ● | ● | ● | | ● |
| GROWTH DELAY | ● | ● | N/A | N/A | No | N/A | N/A | N/A |
| GAUCHER CELLS ON BIOPSY | Occur in clusters | N/A | Sometimes pseudo-Gaucher cells | Sometimes pseudo-Gaucher cells | Sometimes pseudo-Gaucher cells | Sometimes pseudo-Gaucher cells | N/A | Sometimes pseudo-Gaucher cells |

*Acid sphingomyelinase deficiency (ASMD), historically known as Neimann-Pick disease types A, B, and A/B.

**Not all the signs and symptoms or potential differential diagnoses for Gaucher disease are included in this chart. Physicians should determine the appropriate differentials according to each patient's condition.**

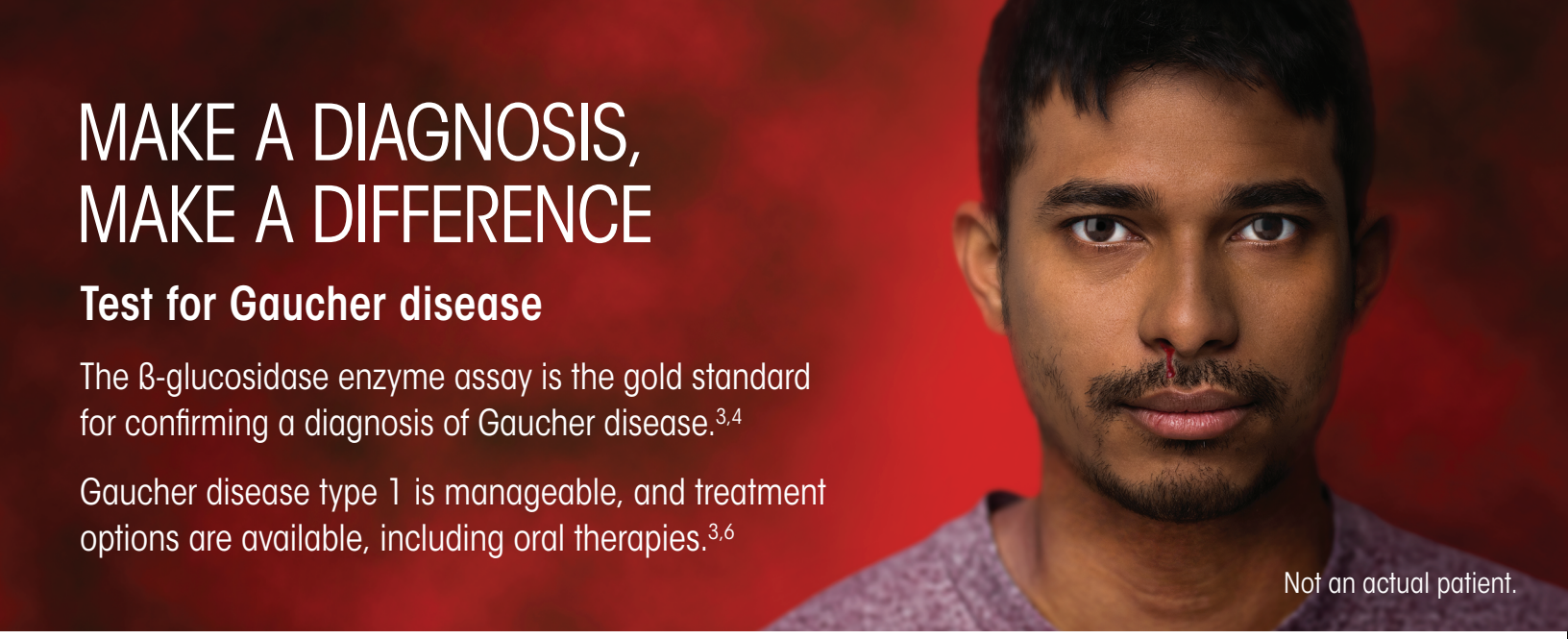
› If a patient presents with these symptoms, consider testing for Gaucher disease.

# Testing is simple

## TEST TO KNOW. IT COULD BE GAUCHER DISEASE TYPE 1.

Blood-based enzyme assay (acid beta-glucosidase) is the gold standard for definitive diagnosis of Gaucher disease type 1. Treatment options are available, including oral therapies.[3,4,6]

› **Learn more at GaucherCare.com/hcp**




## Learn more about diagnosing and testing for Gaucher disease at GaucherCare.com/hcp

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