Information for Gastroenterologists, Hepatologists, and Pulmonologists

GAUCHER disease

Not an actual patient.

GAUCHER DISEASE TYPE 1

Identification and differential diagnosis

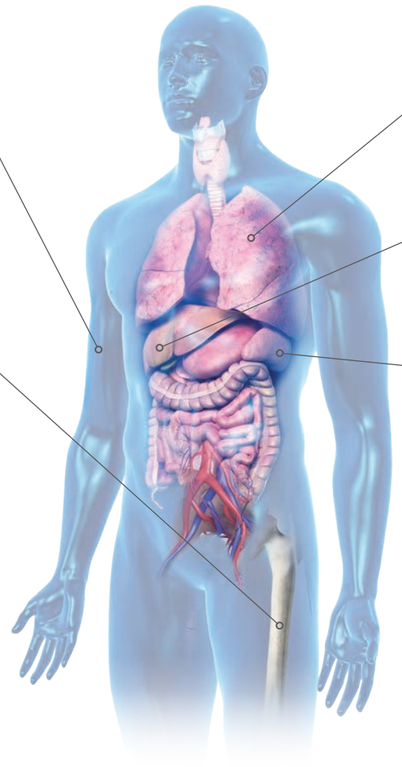
Gaucher disease type 1: A rare, progressive disease that can go undiagnosed up to 10 years[1]

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 FOR 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 OTHER CONDITIONS[3,6-14]

		GASTROENTEROLOGISTS		PULMONOLOGISTS		HEPATOLOGISTS		
	Gaucher disease type 1	Autoimmune lymphoproliferative syndrome	Inflammatory bowel disease (IBD)	Sarcoidosis	Interstitial lung disease	Liver fibrosis/ cirrhosis	Nonalcoholic fatty liver disease (NAFLD)	Hepatitis
HEPATOMEGALY/ SPLENOMEGALY	●	●		Less common		●	●	●
ANEMIA	●	●	●			●		
THROMBOCYTOPENIA	●	●				●		
FATIGUE	●	●	●	●	●	●	●	●
BONE PAIN	●		●					
PULMONARY INVOLVEMENT	●*			●	●	●		●

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.

*Pulmonary symptoms are uncommon for patients with Gaucher disease type 1.

› 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**

MAKE A DIAGNOSIS, MAKE A DIFFERENCE

Test for Gaucher disease

The ß-glucosidase enzyme assay is the gold standard for confirming a diagnosis of Gaucher disease.[3,4]

Gaucher disease type 1 is manageable and treatment options are available, including oral therapies.[3,6]

Not an actual patient.

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

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