Not an actual patient.

DON'T WAIT

HELP PREVENT PROGRESSION OF GAUCHER DISEASE TYPE 1

GAUCHER disease

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ABOUT GAUCHER DISEASE TYPE 1

Gaucher disease is the most common lysosomal storage disorder, caused by a deficiency in glucocerebrosidase enzyme activity, encoded by the *GBA* gene. Deficiency or absence of enzyme activity leads to accumulation of the enzyme substrate glucosylceramide (also known as glucocerebroside or GL-1) in cells of monocyte/macrophage lineage.

GL-1 buildup can lead to progressive, multiorgan dysfunction and lifelong consequences.[1-3]

Not an actual patient.

Timely management can help prevent disease progression

- Generally, pre-emptive therapy before irreversible complications occur can be more effective than a "watchful waiting" approach
- Even if your patient seems asymptomatic or their symptoms are mild, delaying or stopping treatment may lead to further GL-1 accumulation and disease progression[1,7]

Gaucher disease can progress with serious and irreversible complications, including[1,4]:

- Progressive visceral enlargement
- Bone pain, fractures, avascular necrosis, osteopenia, and osteoporosis
- Growth failure in children
- Hepatic, splenic, or marrow fibrosis
- Lung disease and pulmonary hypertension
- Bleeding and bleeding complications
- Anemia, fatigue, and pallor

The most common pathogenic variant among Ashkenazi Jews is N370S[5]*

~1 IN 850

While Gaucher disease is panethnic at ~1:40,000 in the general population, it is more common in Ashkenazi Jews at ~1:850.[5]

- *~90% of American Jews are Ashkenazi*[6]

- Patients may not notice disease manifestations themselves[5]
- These patients can experience adult-onset disease and a disproportionate burden of progressive skeletal disease, without major visceral or hematologic involvement[5]

*N370S is also known as c.1226A>G and p.Asp409Ser.

Gaucher disease can also lead to a **reduced quality of life**, a **shortened lifespan**, and an **increased risk of cancer**.[4]

Regardless of symptoms or severity, Gaucher disease is progressive, so it is important to **diagnose Gaucher disease type 1 as soon as possible and initiate timely treatment**.[7]

Gaucher disease type 1 can be effectively managed with appropriate treatment and continued monitoring.

TREATMENT OPTIONS ARE AVAILABLE, INCLUDING ORAL THERAPIES.[4,8]

Continued monitoring and testing are IMPORTANT to determine disease progression

Regular tests include[8,9]:

- **MRI or CT scan** to monitor splenomegaly, hepatomegaly, and changes in bone marrow
- **Blood tests** to measure hemoglobin, platelet counts, and biochemical evaluations of appropriate biomarkers
- **X-rays and DEXA scans** to check for fractures and osteopenia/osteoporosis

Physicians should determine necessary assessments and the actual frequency according to a patient's individualized therapeutic goals and routine follow-up.

CT=computed tomography; DEXA=dual-energy X-ray absorptiometry; MRI=magnetic resonance imaging.

Encourage **ALL** patients with Gaucher disease to follow their treatment and disease monitoring schedule.

See a recommended schedule of assessments at **GaucherCare.com**.

GAUCHER DISEASE TYPE 1 CAN BE EFFECTIVELY MANAGED.

DON'T WAIT. TREATMENT OPTIONS ARE AVAILABLE, INCLUDING ORAL THERAPIES.[4,8]

› Learn more at GaucherCare.com

RULE OUT MALIGNANCY[1]

PERFORM GAUCHER ENZYME TEST*

*Acid-ß-glucosidase enzyme activity assays.

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