

GAUCHER DISEASE

1 in 40,000 live births will have Gaucher Disease.

Gaucher disease is a genetic mutation and lysosomal storage disorder (LSD) that leads to the build of fatty materials in different areas of the body because of low levels of glucocerebrosidase (GCase) that is supposed to break down the lipids. Some prominent areas these collect are in bone marrow, the spleen, and the liver.

01 | Symptoms

The symptoms for this rare disease can vary depending on the individual. Some of the most common symptoms are:

- Anemia
- Fatigue
- Hepatomegaly — enlarged liver
- Splenomegaly — enlarged spleen

Some less common symptoms are:

- Abdominal pain
- Joint pain
- Delayed puberty/skeletal maturity
- Brain damage
- Seizures

02 | Diagnosis

There are several types of Gaucher disease, which are based on their severity as well as effects on the brain and spinal cord.

- **Type 1:** This is the most common, especially in western countries, with 95% of those with Gaucher disease having this type. This type generally doesn't affect the brain and spinal cord.
- **Type 2:** This is less common and the most severe due to how it affects the brain.
- **Type 3:** This is the least common in the U.S. and Europe, but is the most common type worldwide. Type 3's severity falls between Types 1 and 2.

03 | Treatment

Based on the different types of Gaucher disease, there are a few treatments available. A majority of treatments are available and work for curing Type 1. For example, enzyme replacement therapy (ERT) is affective for Type 1, but does not affect the neurological damage that results in Types 2 and 3. Unfortunately, this is the same case for most treatments for Gaucher disease with only the non-neurological symptoms being treatable.

For more information and supportive resources, please visit <https://www.gaucherdisease.org/>

Did You Know?

Those of Ashkenazic Jewish descent are more likely to be affected by this disease, happening in up to 1 in 450 births.

References

<https://rarediseases.info.nih.gov/diseases/8233/gaucher-disease>

<https://rarediseases.org/rare-diseases/gaucher-disease/>