Gaucher Disease
Treatment Guidelines Handout

What is Gaucher disease?

Gaucher disease is a rare genetic disorder caused by a deficiency of the enzyme glucocerebrosidase, leading to the accumulation of fatty substances primarily in the spleen, liver, and bone marrow. There are different types of Gaucher disease, with type 1 being the most common and generally less severe.

Symptoms of Gaucher disease

Symptoms can vary widely but may include:

- Enlarged liver and spleen
- Bone pain and skeletal abnormalities
- Fatigue and anemia
- Easy bruising and bleeding due to low platelet count
- Neurological symptoms resembling Parkinson's disease in rare cases

Treatments options

There is currently no cure for Gaucher disease, but there are several treatment options available to manage symptoms and slow the progression of the disease (National Gaucher Foundation, 2019; National Organization for Rare Disorders, 2020):

Enzyme replacement therapy (ERT)

ERT addresses low levels of the GCase enzyme by introducing a modified version of the natural human enzyme. This facilitates the breakdown of glucocerebroside, a fatty substance that accumulates in organs and bone marrow.

Patients typically receive ERT through intravenous (IV) infusion every two weeks, either at an infusion center or at home. The FDA has approved treatments for Gaucher Disease, which include:

- Cerezyme® (imiglucerase)
- VPRIV® (velaglucerase alfa)
- Elelyso® (taliglucerase alfa)

Substrate reduction therapy (SRT)

SRT operates differently from ERT. Rather than replenishing low levels of the GCase enzyme, SRT reduces the production of glucocerebroside in the body. This reduction eases the enzyme's workload by providing less glucocerebroside to break down. SRTs are administered orally.

Currently, there are two FDA-approved oral SRT medications for patients with Gaucher disease:

- Cerdelga® (eliglustat)
- Zavesca® (miglustat)

SRTs are limited to specific patients due to their distinct pharmacological properties. For example, SRTs are **not approved** for individuals under 18 or for women who are breastfeeding, pregnant, or attempting to conceive.

Treating the symptoms of Gaucher disease

Beyond ERT and SRT, which target enzyme deficiencies and the buildup of glucocerebroside, patients with Gaucher disease may require additional treatments to address symptoms and complications. These treatments can include:

- Blood transfusions to combat severe anemia and bleeding
- Prescription medications to alleviate bone pain and treat osteoporosis
- Orthopedic surgeries, such as joint replacements, to repair painful or damaged joints

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Genetic counselling

Genetic counseling is recommended for affected individuals and their families to help them understand the inheritance patterns, risks, and options available.

References

National Gaucher Foundation. (2019). *Gaucher disease treatment*. https://www.gaucherdisease.org/gaucher-diagnosis-treatment/treatment/

National Organization for Rare Disorders. (2020, March 3). *Gaucher disease*. https://rarediseases.org/rare-diseases/gaucher-disease/