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Insights on Gaucher Disease

Sanike Swapna*

Department of Biotechnology, Osmania University, Hyderabad, Telangana, India

Editorial

Gaucher disease is caused by an accumulation of certain fatty substances in certain organs, most notably your spleen and liver. This causes these organs to enlarge and has the potential to impair their function. Fatty substances can also accumulate in bone tissue, weakening it and increasing the risk of fracture. If your bone marrow is harmed, it can impair your blood's ability to clot. In people with Gaucher disease, an enzyme that breaks down these fatty substances does not function properly. Enzyme replacement therapy is frequently used in treatment. Gaucher disease is an inherited disorder that is most common in Jews of Eastern and Central European descent (Ashkenazi). Symptoms can manifest themselves at any age.

There are various types of Gaucher disease, and the signs and symptoms of the disease can vary greatly even within the same type. Type 1 is by far the most prevalent. Siblings, including identical twins, with the disease can have varying degrees of severity. Some people with Gaucher disease exhibit only minor or no symptoms. The following issues affect the majority of Gaucher disease patients to varying degrees: Complaints about the abdomen Because the liver and, in particular, the spleen can swell significantly, the abdomen can become painfully distended.

Anomalies of the skeleton. Gaucher disease can cause bone weakness, increasing the likelihood of painful fractures. It can also disrupt the blood supply to your bones, causing portions of the bone to die. Blood problems. Anemia (a decrease in healthy red blood cells) can cause extreme fatigue. Gaucher disease also affects the clotting cells, resulting in easy bruising and nosebleeds.Gaucher disease, which affects the brain more rarely, can cause abnormal eye movements, muscle rigidity, swallowing difficulties, and seizures. One rare subtype of Gaucher disease begins in infancy and usually kills the child before the age of two

Gaucher disease is passed down through an autosomal recessive inheritance pattern. For their child to inherit the condition, both parents must be carriers of a Gaucher changed (mutated) gene. The main tool used by doctors to diagnose gaucher disease is an enzyme test called a beta-glucosidase leukocyte (BGL) test. This is due to the fact that all Gaucher disease patients have low enzyme activity levels.A standard blood test can be used by your doctor to measure enzyme activity.

In the case of adults, the physician will draw a blood sample with a needle and send it for testing. Many doctors now use a less invasive heel stick on babies (a faster, smaller prick).Because all patients with Gaucher disease have low enzyme activity, a Beta-glucosidase Leukocyte (BGL) test will almost certainly reveal whether or not a person has the disease. Carriers may occasionally have borderline low enzyme levels, necessitating further genetic testing to determine whether the person has Gaucher disease.

A targeted mutation test will identify approximately 89 percent of carriers in carrier testing. Over 99 percent of gaucher disease carriers can be identified using gene sequencing. Gaucher disease cannot be diagnosed without a bone marrow test. However, some doctors use this test to diagnose Gaucher disease while trying to rule out other blood disorders. When a person has the disease, the test will typically reveal fat-laden Gaucher cells that have an abnormal appearance. In rare cases, the test fails to accurately identify gaucher cells, or those that are detected may be indicative of a blood disorder other than gaucher disease [1-5].

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***Address for Correspondence:** Sanike Swapna, Department of Biotechnology, Osmania University, Hyderabad, Telangana, India, E-mail: sanika.swapna5@gmail.com*

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