



COMMENTARY



Classification and Diagnosis of Gaucher Disease

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Description

A hereditary illness called Gaucher's Disease or Gaucher Disease (GD) causes glucocerebroside to build up in cells and some organs. The condition, which is brought on by a congenital lack of the enzyme glucocerebrosidase, which works on glucocerebroside, is characterised by bruising, exhaustion, anaemia, a low blood platelet count, and enlargement of the liver and spleen. Glucocerebroside increases when the enzyme is damaged, especially in macrophages and white blood cells. The spleen, liver, kidneys, lungs, brain, and bone marrow can all accumulate glucocerebroside.

Swollen lymph nodes and nearby joints, a distended abdomen, a brownish tint to the skin, anaemia, a low blood platelet count, and yellow fatty deposits on the white of the eye are just a few symptoms may appear. Other symptoms include enlarged spleen and liver, liver dysfunction, skeletal disorders or painful bone lesions, severe neurological complications, anaemia, low blood platelet count, and painful bone lesions. People who are extremely ill may also be more prone to infection. Enzyme replacement therapy may be used to treat some types of Gaucher's disease.

Diagnosis

Based on the overall clinical picture, Gaucher disease is hypothesised. Enzyme testing may be part of the initial laboratory examination. Therefore, less than 15% of mean normal activity is regarded as diagnostic. Genetic testing will frequently confirm decreased enzyme levels. There are numerous possible mutations, and to establish the diagnosis, the beta-glucosidase gene must occasionally be sequenced. When a known genetic risk factor is present, prenatal diagnostics is an option and is helpful.

A diagnosis may also be inferred from biochemical abnormalities, such as elevated levels of immunoglobulin, alkaline phosphatase, and angiotensin-converting enzyme, or from cell analyses that reveal macrophages

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with glycolipid-rich cytoplasm and "crinkled paper" cytoplasm.

Hexosaminidase, a tartrate-resistant acid phosphatase, and chitotriosidase, a human chitinase, are among the increased lysosomal enzymes. This latter enzyme has shown to be quite helpful for tracking Gaucher disease activity in response to treatment and may indicate the severity of the condition

Classification

There are three common clinical subgroups of Gaucher's Disease (GD). The fact that these classifications do not consider the whole range of observable symptoms has drawn considerable criticism. Additionally, compound heterozygous variants might exist, which significantly adds to the difficulty of forecasting the course of a disease.

The most widespread and mildest form of GD is type I. The liver, spleen, and bone are the main organs affected by the symptoms, which might start early in life or in age. Grossly enlarged spleen and an enlarged liver are both rather common; the spleen can rupture and lead to further difficulties. Bone disease and skeletal weakening may be severe. Anemia, thrombocytopenia, and leukopenia are brought on by bone marrow replacement and spleen enlargement. Although the lung and, in rare cases, kidney impairment, the brain and nervous system are not pathologically impacted. Due to low levels of red blood cells, patients in this group typically bleed easily and feel tired. Patients with type I diabetes may live long into adulthood, depending on the disease's severity and onset. Between patients, there can be a huge difference in the types and severity of symptoms.

GD type II usually manifests as within six months of birth and affects about one in 100,000 live births. A big liver and spleen, progressive brain damage, eye movement problems, spasticity, seizures, limb rigidity, and poor sucking and swallowing abilities are a few symptoms. By age two, affected children typically pass away. GD

type III, which affects one in 100,000 live births, can start at any point throughout childhood or even maturity. Compared to the acute or type II version, it is characterised by slowly developing, but milder neurologic symptoms. Seizures, poor coordination, skeletal abnormalities, eye movement difficulties, blood disorders like anaemia, and respiratory issues are among the major symptoms, along with an enlarged spleen and/or liver. Patients frequently survive into their early adolescent and adult years.