IJARSCT
ISSN (Online) 2581-9429
International Journal of Advanced Research in Science, Communication and Technology (IJARSCT)
International Open-Access, Double-Blind, Peer-Reviewed, Refereed, Multidisciplinary Online Journal
Impact Factor: 7.53
Volume 5, Issue 3, January 2025

To Review on Gaucher Disease

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Abstract: *A glucocerebrosidase deficiency causes Gaucher disease (GD), a hereditary metabolic mistake. As a result, the liver, spleen, bone, and bone marrow store an excessive amount of glucocerebroside. People develop anemia, a condition called a large liver, bone infarcts, aseptic death of bone, and osteoporosis. There are three forms of GD, with types 2 and 3 involving the nervous system. The natural history of the disease has changed dramatically with the introduction of enzyme replacement therapy and substrate reduction therapy, resulting in a notable decrease in morbidity, particularly for type 1 patients. This article covers a wide range of topics related to Gaucher illness, including its historical background and more recent treatments that are still in the research stage.*

A uncommon genetic illness that is autosomal recessive is Gaucher disease (GD, ORPHA355). The lysosomal enzyme glucocerebrosidase deficiency that causes an accumulation of glucosylceramide, the enzyme's substrate, in macrophages is the cause of this condition. Its incidence ranges from 1/40,000 to 1/60,000 births in the general population, and it rises to 1/800 in Ashkenazi Jews. The primary source of the disease's cytopenia, splenomegaly, a large liver, and bone lesions is thought to be the introduction of Gaucher cells into the liver, spleen, and bone marrow.

Keywords: glucocerebrosidase.

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DOI: 10.48175/568