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To Review on Gaucher Disease

Mansvi Kudwe, Yashwant Ingole, Rajlaxmi Deolekar
Students, Final Year, New Montfort Institute of Pharmacy, Ashti, Wardha, India

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.

I. INTRODUCTION

Glucocerebrosidase (GBA1) gene mutations are the cause of Gaucher disease, a hereditary autosomal recessive defect of metabolism that is pronounced as GO-SHEY[1]

The glucocerebroside lipids' beta-glucosidic bond is broken by the enzyme GBA1. A moving example of the complex interactions of biology, biochemistry, and clinical care is Gautier disease. This inherited metabolic illness is named for the French physician Philippe Charles Ernest Gaucher, who first reported it in 1882. It is characterised by the build-up of lipids, notably glucocerebroside, in different human tissues and organs. A wide range of ages and backgrounds can be affected by the mild to severe symptoms of Gaucher disease, which can present in a variety of clinical presentations.

Since inborn metabolic abnormalities typically (but not always) manifest during the neonatal stage of infancy, they are especially important in paediatrics. The five forms of Gaucher disease that are currently recognised are cardiovascular, perinatal fatal, type 1, type 2, and type 3. The most serious type, known as perinatal fatal form, might have difficulties that start before delivery or in the first few months of life.

Diagnosing any inborn defect of metabolism requires knowledge of its principal forms. The main causes of inborn errors of metabolism are (1) inadequate levels of certain enzymes that must break down amino acids or other metabolites, which can accumulate and become toxic if ignored, or (2) the absence of certain enzymes altogether. An inborn metabolic mistake known as "toxic accumulation" caused by glucocerebroside lipid buildup is Gaucher disease. Three main classifications exist for hazardous buildup inborn errors of metabolism: systemic toxicity, localised toxicity, or a mix of the two. A concentrated toxicity example would be Gaucher disease.

Gaucher's disease comes in three autosomal recessive forms. For an infant to be affected, both parents need to be carriers. Affected children have a one in four, or 25%, chance of contracting the illness with each pregnancy if both parents are carriers. Certain mutations have been connected to each kind. Approximately 80 reported mutations in the GBA gene can be divided into three primary categories.[2]

The entire clinical picture suggests the diagnosis of Gaucher disease. Enzyme testing may be part of the initial laboratory testing. As an outcome, below 15% of mean normal activity is regarded to be diagnostic.[3]

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Genetic testing is frequently used to confirm decreased enzyme levels. Since there are many distinct mutations, the beta-glucosidase gene may occasionally need to be sequenced in order to confirm the diagnosis. When a recognised biological risk factor is present, prenatal diagnostics is an option and can be helpful.

theme lysosomal enzymes are elevated, including tartrate-resistant acid phosphatase, hexosaminidase, and a human chitinase, chitotriosidase. This latter enzyme has proved to be very useful for monitoring Gaucher's disease activity in response to treatment, and may reflect the severity of the disease.[4]

Three common clinical categories of Gaucher's disease (GD) exist.[5]

Certain subtypes have drawn criticisms for failing to consider the entire range of observable symptoms, or phenotypes. Additionally, there are compound heterozygous variants, which significantly raise the difficulty of predicting the course of a diseases.[6,7]

Intracerenous recombinant glucocerebrosidase enzyme replacement therapy may improve additional signs, reduce skeletal deformities, and lower the size of the liver and spleen in individuals with type-I and most type-III.[8]

The rarity of the disease means dose-finding studies have been difficult to conduct, so controversy remains over the optimal dose and dosing frequency.[9]

Alglucerase, also known as Cerebase, was the first medication for Gaucher's disease. It was derived from human placental tissue and altered using enzymes.[10]

It was approved by the FDA in 1991[11] and has been withdrawn from the market.[12,13]

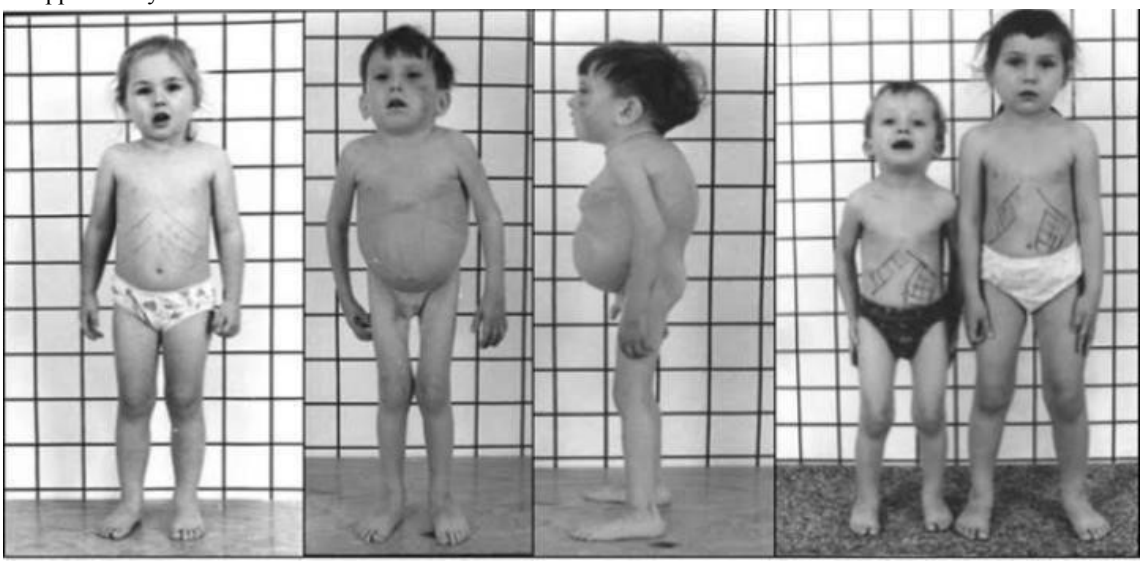

Fig.1

TYPES OF GAUCHER DISEASE

Type 1 Gaucher Disease (Non-Neuropathic):-

The most prevalent variant, known as the "non-neuropathic" type I (N370S homozygote), is 100 times more common in Ashkenazi Jews than in the overall population. The life expectancy is slightly reduced[14], and the median age upon diagnosis is 28 years of age .[14]

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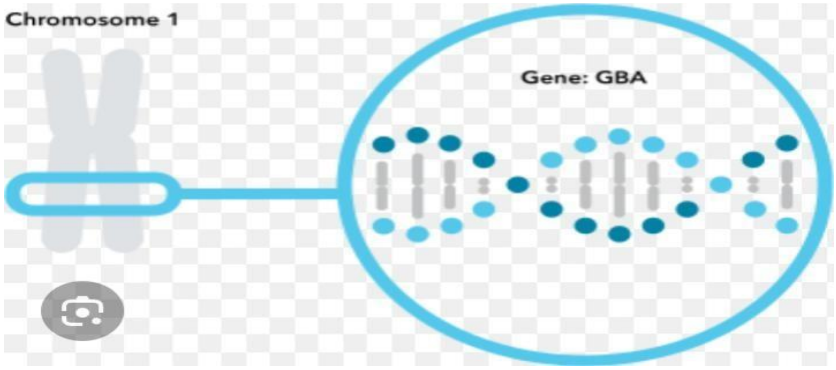

Fig. 2

Type 2 Gaucher Disease (Acute Neuropathic Form):-
In young children, Type II (one or two alleles L444P) is associated with neurological issues. Seldom is the enzyme sent into the lysosomes. The prognosis is bad; most pass away before turning three.

Type 3 Gaucher Disease (Chronic Neuropathic Form) :-
Patients from the Forgotten region of Sweden have type III (including one or two copies of L444P, potentially delayed by protective polymorphisms) in their blood. [15] While the disease strikes this group a little later, the majority of them pass away before turning thirty. The Gaucher-causing mutations may have infiltrated the Sephardic Jewish gene bank in the beginning of the Middle Ages (48–55 generations
ago). [16]

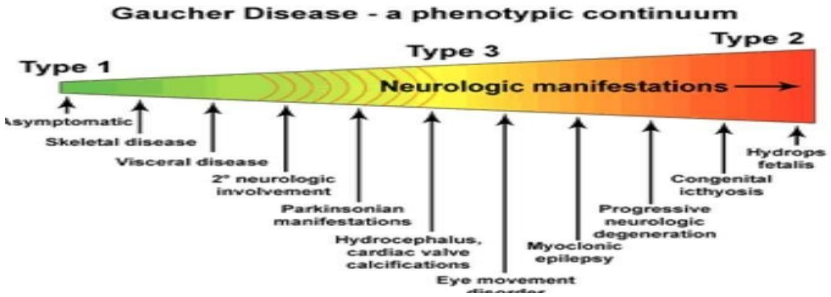

Fig. 3

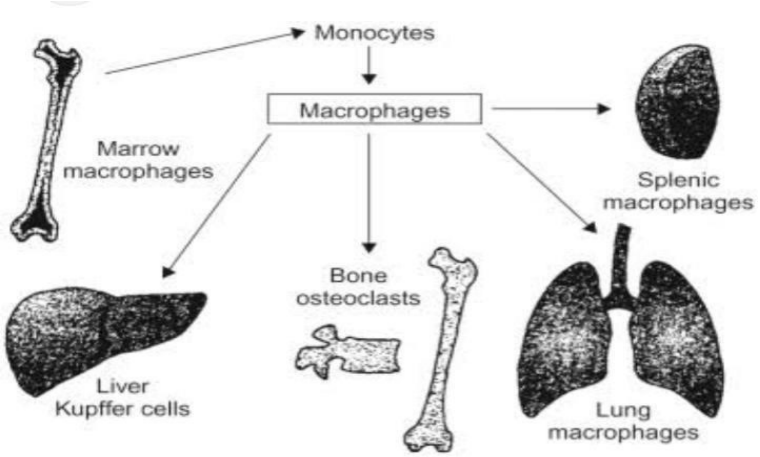

Fig. 4

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HISTORY AND PHYSICAL

Patients with Gaucher disease can present with several signs and symptoms, depending on the underlying type of disease. Commonly seen presenting signs and symptoms are as follows:

1.Painless hepatomegaly and splenomegaly
2. Hypersplenism and pancytopenia
3. Severe joint pains, most frequently affecting the hips and knees
4. olfaction and cognition (Type 1) Severe convulsions, hypertonia, intellectual disability, and apnea (Type 2)
5.Myoclonus, seizures, dementia, and ocular muscle apraxia (Type 3)
6.Parkinsonism
7.Osteoporosis
8.Yellowish-brown skin pigmentation

Gaucher disease ageing is dependent on detecting low GBA1 enzyme levels in peripheral blood leukocytes and confirming the existence of GBA1 gene mutation alleles. Gaucher disease can be diagnosed with merely a blood sample, however some people require needless invasive blood vessel or liver biopsies in order to receive the proper diagnosis. Such accidents can be prevented if physicians are aware of the symptoms and indicators of Gaucher disease. Furthermore, a lot of individuals with enlarged livers or spleens are informed they may have cancer before receiving a proper diagnosis.[17]

PATHOPHYSIOLOGY

It is possible to categorise the visceral, haematologic, bones, and metabolic components of Gaucher disease signs and symptoms. Hepatosplenomegaly, or a fatty liver and spleen, is one of the visceral components.

Leukopenia, anaemia, and thrombocytopenia are examples of haematological components. Low platelet counts are hypothesised to be caused by cerebroside buildup in the bone marrow, which reduces platelet production. Likewise, it is believed that cerebroside development in the spleen causes an increased rate of red blood cell lysis, which leads to anaemia, and increased clearance of white blood cells (WBC), which results in a low WBC count. Bleeding and infections may become more likely if blood cells are destroyed too quickly.

The skeletal components may consist of pathological bone fractures, Erlenmeyer flask deformity, low bone density relative to normal peak density, bone crises, and bone cell death (also known as avascular necrosis or osteonecrosis). The accumulation of glucocerebroside-loaded macrophages in the bone marrow is mostly responsible for these skeletal abnormalities. These macrophages impede blood flow and the intake of nutrients and oxygen, which can lead to low bone density, growth irregularities, excruciating pain, and bone cell necrosis.

Patients with Gaucher disease, both paediatric and adult, exhibit metabolic problems, particularly issues related to nutrition and glucose metabolism. Unusual body weight and associated metabolic disorders are prevalent problems. To maximise care management for impacted individuals, a heightened emphasis on identifying metabolic disturbances—particularly nutritional status abnormalities, the resistance to insulin, and cholesterol alterations—is highly advised. [18]

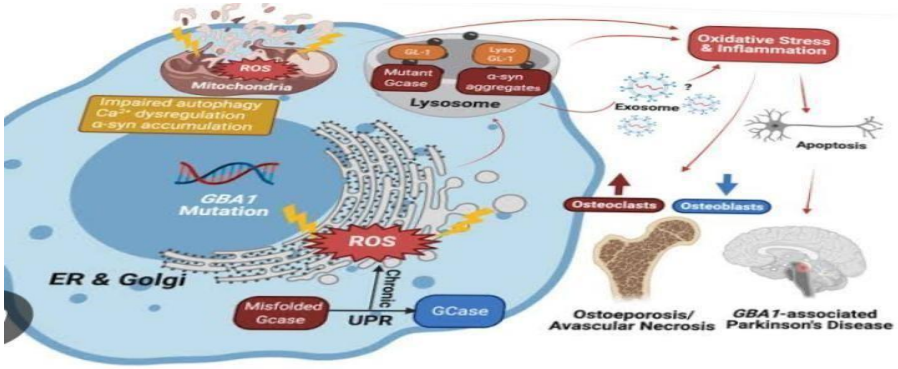

Fig. 5

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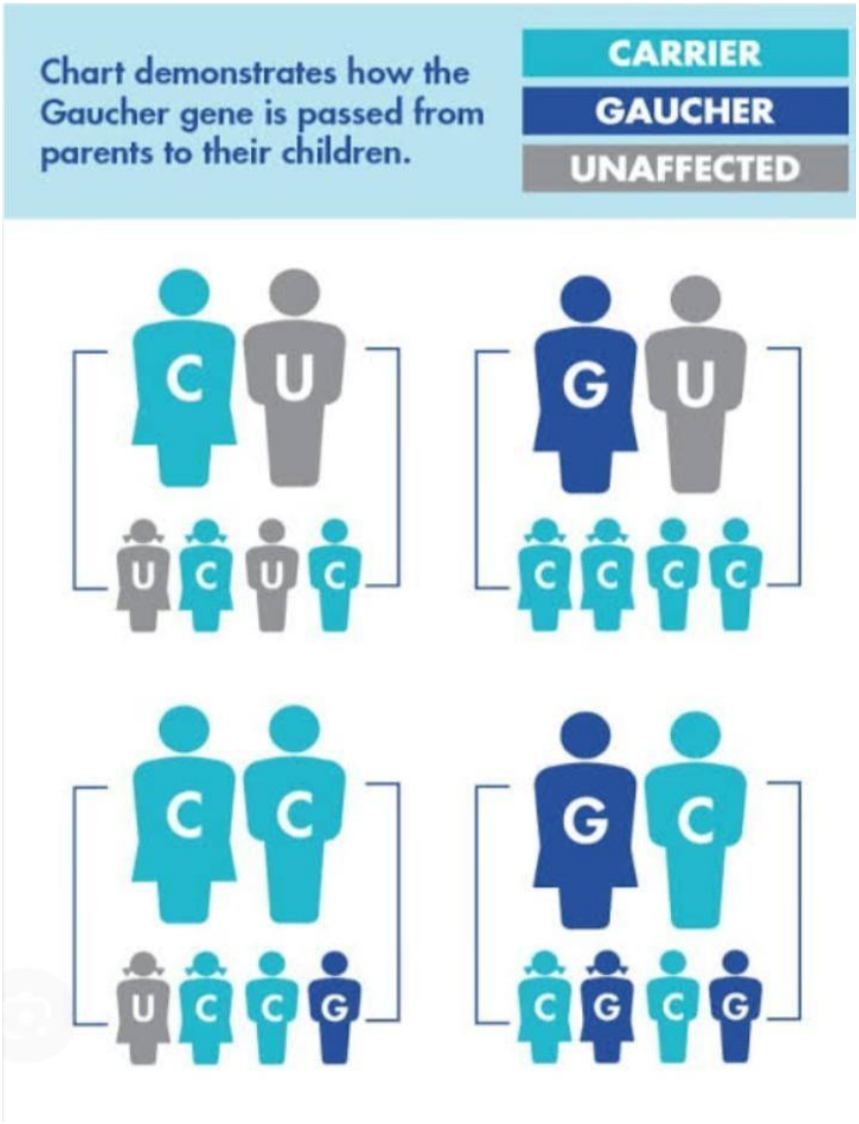
EPIDEMIOLOGY

In the Sephardic (Eastern European) Jewish humanity, Gaucher disease is the most prevalent autosomal recessive illness, with a prevalence of carriers of 6% as opposed to 0.7% to 0.8% in the non-Jewish population. The Ashkenazi community is also prone to Tay- Sachs illness (3.7% carrier frequency) and pulmonary fibrosis (4% carrier frequency). [19]



SYMPTOMS OF GAUCHER DISEASE

Hepatomegaly with splenomegaly without pain: the spleen can weigh 1,500–3,000 g (3.3–6.6 lb) instead of the typical 50–200 g (0.11–0.44 lb) in weight. May put pressure on the stomach, reducing the affected person's ability to eat. Splenic rupture is more likely when there is splenic hypertrophy, even when it is painless.

Anaemia, neutropenia, leukopenia, and neutropenia (with an increased risk of sickness and bleeding) are the results of hypersplenism and anaemia, the rapid and rapid destruction of blood cells.

Liver cirrhosis is not common.

Severe joint and bone pain can happen; it usually starts in the knees and hips.

certain kinds of Gaucher's disease cause neurological signs

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Type I: impaired olfaction and cognition.
Type II: serious convulsions, hypertonia, intellectual disability, and apnea.
Type III: muscle twitches known as myoclonus, convulsions, dementia, and ocular muscle apraxia
Parkinson's disease is recognized as being more common in Gaucher's disease patient's t and heir heterozygous carrier relatives.[20]

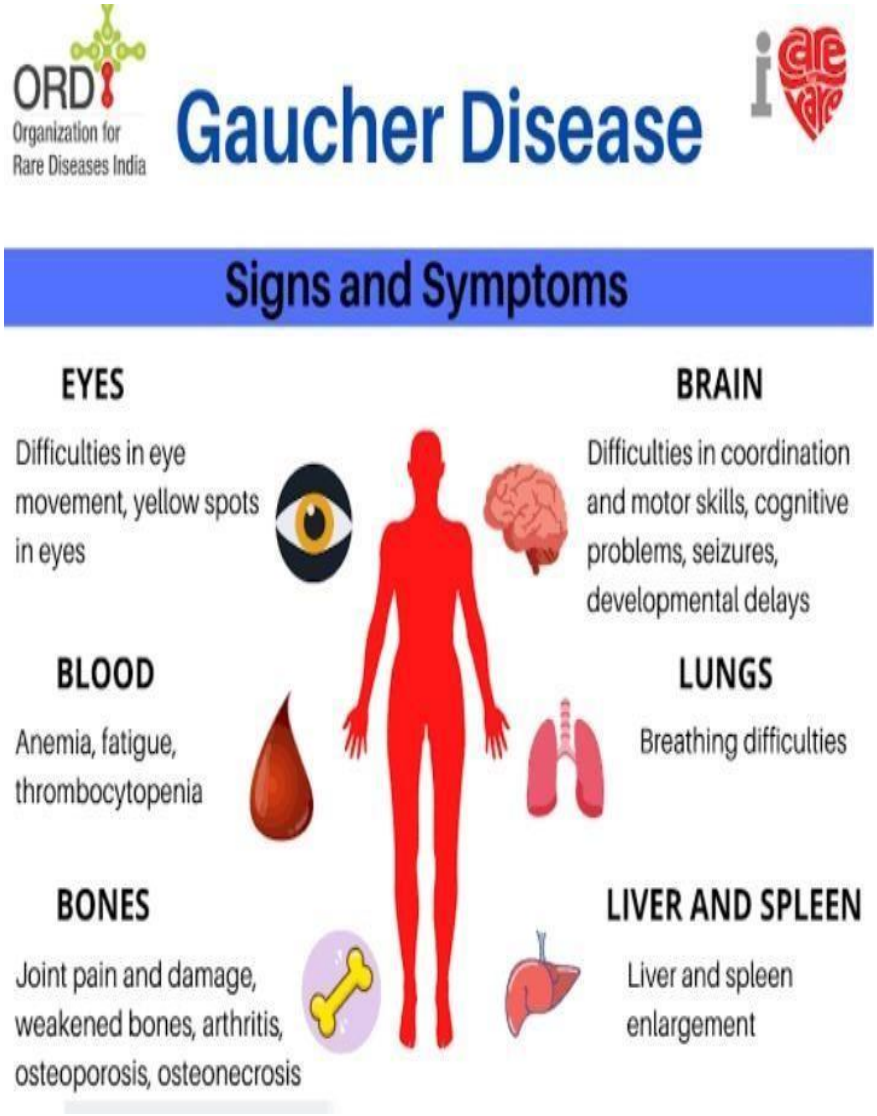

Fig. 7

CAUSES OF GAUCHER DISEASE

GBA gene mutations are the cause of Gaucher disease.
An accumulation of specific fatty molecules in your liver and spleen, among other organs.
A parent's aberrant GBA gene must be passed along to their child in order for Gaucher to occur.
Gaucher disease, which accumulates in specific bodily parts. 5. It has an impact on every organ in the body.

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Fig.7

DIAGNOSISS OF GAUCHER DISEASE

Multiple myeloma
Lewy body dementia
Niemann-Pick disease
Parkinson disease
Sphingomyelinase deficiency [21,22,23,24]

Data on the age of individuals at diagnosis or onset of symptoms was published in a number of research. [25]

In contrast, the mean age at diagnosis in an older US population that took part in a cross-section survey was 28.9 ± 21.2 years. [26]

Age at GD1 diagnosis was reported by genotype in one study: people who have genotypes N370S/?, N370S/N370S, N370S/L444P, and N370S/rare allele had a mean age at diagnosis of more than 10 years; patients with genotypes N370S/84GG, L444P/L444P, L444P/?, and N370S/IVS2+1, all of which are frequently linked to severe GD symptomatology, had an age at diagnosis of less than 10 years. [27]

TREATMENT OF GAUCHER DISEASE

There are two types of treatment for Gaucher disease: substrate reduction therapy and enzyme replacement therapy[28].

Enzyme replacement therapy will help with the "non-brain" signs and symptoms associated with type 3 Gaucher disease, e.g., enlarged organs and skeletal issues. Enzyme replacement therapy does not correct the underlying genetic defect and only relieves signs, symptoms, and ongoing damage caused by the accumulation of toxins. Moreover, it is possible to develop antibodies to the replacement enzyme[29].

In substrate reduction therapy, the goal is to reduce substrate levels such that toxic accumulation of the substrate's subsequent degradative product is diminished to a clinically less toxic level. In the case of Gaucher disease, the goal is to use substrate reduction therapies to inhibit the first committed step in glycosphingolipid biosynthesis. There are 2 FDA-approved substrate reduction therapy drugs to treat patients with Gaucher disease: eliglustat and miglustat. Eliglustat, a glucosylceramide synthase inhibitor, is indicated only for type 1 Gaucher disease and does not effectively cross the blood-brain barrier[30]

Hematopoietic stem cell transplantation is another option for treating Gaucher's disease. However, with the advent of enzyme and substrate replacement therapies, it has fallen out of favor, given its association with high morbidity and mortality risk. Currently, it is still a definitive treatment option for patients with type 3 Gaucher's disease[31,32,33,34,35,36].

Splenectomy is rarely used nowadays but can be useful in patients with uncontrolled and serious thrombocytopenia or with severe, uncontrolled abdominal pain[37]. Patients undergoing splenectomy should be counselled regarding the higher risk of infections and vaccinated against encapsulated organisms as per institutional policies.

Future advancements in managing Gaucher disease will be looking to gene editing and gene therapy[38].

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On the basis of pathogenic mechanisms, there are 2 specific ways to treat GD: (1) recovery of enzyme activity, such as enzyme replacement therapy; (2) reduction of accumulation of glucocerebrosidep in lysosome, such as substrate reduction therapy [39].

RISK FACTORS OF GAUCHER DISEASE

There is no lifestyle risk factors for the condition, but Gaucher disease more often affects people of Ashkenazi Jewish ancestry. About 1 in 14 Ashkenazi Jews is a carrier of the genetic mutation[40].

People of Eastern and Central European Jewish (Ashkenazi) ancestry are at higher risk of developing the most common variety of Gaucher disease.

II. CONCLUSION

A rare but preventable metabolic illness is Gaucher disease. A high degree of suspicion is necessary for an early diagnosis because the clinical symptoms of this disease can vary. Treatment early on will help avoid permanent repercussions.

Gaucher disease is the most prevalent lysosomal storage disorder, yet it is still uncommon. The majority of cases have a slow-onset phenotype, which accounts for the disease's delayed identification. In order to prevent potentially dangerous splenectomy, it is crucial to cover GD in the clinical process in cases of spleen cancer and/or thrombocytopenia. Notably, GD's pathogenesis can be better understood in light of the fact that GCase deficiency affects more than just the number of macrophages that become Gaucher cells. These revelations will pave the way for the creation of fresh treatment approaches. It is expected that medications capable of altering the neurological phenotype will eventually be created. It is expected that greater molecular research will ultimately lead to specific patient care.

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