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A Review on Lysosomal Storage Disease

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Abstract: *Lysosomal Storage Disorders (LSDs) comprise a group of at least 50 distinct genetic diseases, each one resulting from a deficiency of a particular lysosomal protein activity, or in a few cases from nonlysosomal activities that are involved in lysosomal biogenesis or protein maturation. All share a common biochemical characteristic in that they result in accumulation of normally degraded substrates within lysosomes. That is eventually leads to an irreversible cell damage, an ultimately multi-organ dysfunction. Th e substrates stored and site of storages vary, leading to a wide spectrum of clinical manifestations. Lysosomal storage disorders are a family of over seventy rare monogenic diseases that typically present in infancy or childhood and collectively affect 1:5000 live births Most of the causative genes encode lysosomal enzymes or proteins involved in lysosomal enzyme modification or transport, but they can also encode lysosomal membrane proteins.*

Keywords: Lysosomal Storage Disorders

I. INTRODUCTION

Lysosomal Storage Disorders (LSDs) comprise a group of at least 50 distinct genetic diseases, each one resulting from a deficiency of a particular lysosomal protein activity, or in a few cases from nonlysosomal activities that are involved in lysosomal biogenesis or protein maturation. All share a common biochemical characteristic in that they result in accumulation of normally degraded substrates within lysosomes. That is eventually leads to an irreversible cell damage, an ultimately multi-organ dysfunction. Th e substrates stored and site of storages vary, leading to a wide spectrum of clinical manifestations. Lysosomal storage disorders are a family of over seventy rare monogenic diseases that typically present in infancy or childhood and collectively affect 1:5000 live births Most of the causative genes encode lysosomal enzymes or proteins involved in lysosomal enzyme modification or transport, but they can also encode lysosomal membrane proteins. [1] Lysosomal storage diseases (LSDs) are a group of 50 rare, inherited, metabolic disorders characterized by deficiencies in normal lysosomal function and by intra lysosomal accumulation of undegraded substrates. Lysosomal storage diseases (LSDs) are a group of approximately 50 genetic disorders caused by mutations in genes encoding proteins necessary for lysosomal function and, in most cases, enzymes involved in the cellular degradation and traffic king of lipids and other macromolecules. Individual LSDs are rare but together they are a relatively common group of diseases with a combined prevalence of approximately 1:8000 live births.[2] LSDs were originally named and categorized by their clinical presentations or the name of the physician who first identified them, such as Batten, Gaucher, and Niemann–Pick diseases. Advances in the understanding of the causative mutated genes and affected cellular molecules in the lysosomes have now allowed for the classification of LSDs into 48 individual diseases, and some of them have been renamed according to the nature of the accumulated products in the lysosomes. The current naming of the LSDs as established by the Lysosomal Disease Network is shown in A common feature of LSDs is the accumulation of lipids, glycoproteins, and other macromolecules in lysosomes as a consequence of the functional deficiency of a specific lysosomal protein in the affected cells. The accumulation of these molecules normally causes an enlargement of the lysosomes and reduces the supply of macromolecules, or downstream products, for use in critical cellular functions, such as molecular biosynthesis and energy metabolism.[3] Although clinical manifestations and onset of disease vary significantly among the different LSDs, hepatomegaly and splenomegaly are two of the most common symptoms 6 and in more than half of LSDs, central nervous system (CNS) symptoms accompanied by neuronal degeneration occur. Lysosomal storage disorders (LSDs) are a group of rare genetic syndromes resulting from abnormal build-up of undegraded materials which accumulate in the lysosomes of different cells of the human body, as a result of total or partial functional loss of specific lysosomal enzymes or co-factors

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implicated in the degradation of those materials.[4] more specifically the upstream precursors accumulate, and LSD clinical presentation reflects the amount of residual enzymatic activity, ranging from infantile (little or no enzymatic function) to adolescent and adult disease (moderate or mild residual enzymatic function). In the mid1950s, the biochemist Christian de Duve identifiedand characterized the "lysosome" as a subcellular organelle responsible for digestion and recycling of different macromolecules, using cell fractionation techniques and biochemical analyses [5]: this was the scientific breakthrough that would lead to understand the physiological basis of LSDs. Pompe disease (or glycogen storage disease type 2, OMIM 232300) has been the first disease to be identified as an LSD in 1963, caused by the inherited deficiency of acid maltase (α-glucosidase), necessary to break down glycogen and convert it into glucose. [6] All LSDs may be fatal, and many display profound neurological impairment or multi-organ dysfunction that begin in childhood. Each disease be-longing to the group of LSDs encompasses several types that are named for the specific macromolecule that accumulates in each case, and the resulting disorders include Gaucher disease, Fabry disease, mucopoli saccharidoses, Niemann-Pick disease, and gangliosidoses, which are the syndromes analyzed in this review for their potential heterogeneous abnormalities in the immune system. With the exception of the X-linked recessive Fabry disease (and also mucopolysaccharidosis type II), LSDs share a common autosomal recessive inheritance pattern, and have an estimated overall incidence of 1 in 7.500–8.000 live births. [7] In all human cells the lysosome works as a highly complex regulatory and recycling district, and its discovery inaugurated a new era in cellular physiology, which was naturally followed by the identification of approximately 70 distinct LSDs, that collectively ac-count for 14% of all inherited metabolic diseases, resulting from mutations in specific genes encoding for lysosomal hydrolases.[8]

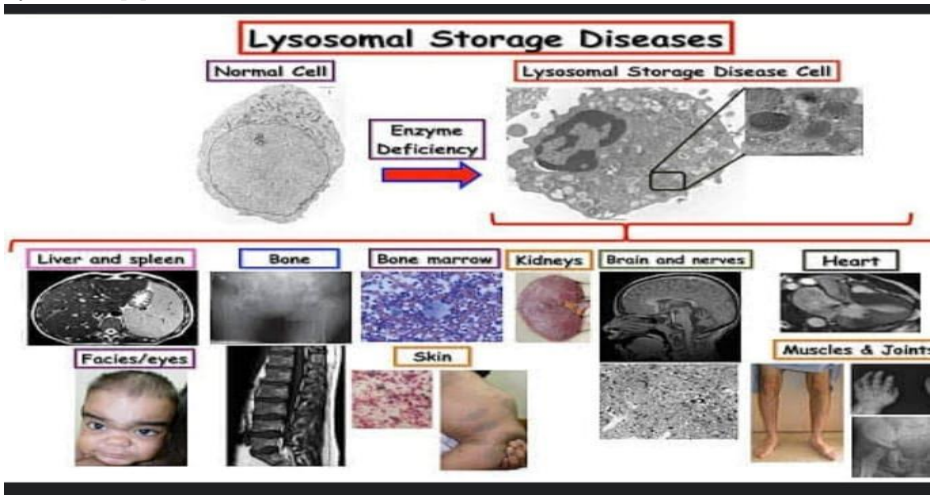

Fig. no.[1] :

Symptoms of lysosomal storage disease

- Pain, numbness, burning in the hands and feet
- Body aches
- Fever
- Tiredness
- Red or purple skin score
- Trouble sweating
- Swelling in the lower legs ankle and feet
- Diarrhea constipation

1. Burning sensation in the hands and feet could be a symptom of peripheral neuropathy, a condition that affects the peripheral nerves. Other symptoms include tingling, numbness, weakness, and pain, especially when the area is touched lightly.

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2. Body aches occur with many health conditions, including arthritis and the flu. If the pain lasts more than a few days, is severe, or occurs with other symptoms, you may need medical attention.
3. fever is a temporary increase in body temperature that's usually a sign that your body is fighting an infection or illness. Fevers are caused by the body's immune system resetting the body's thermostat to a higher temperature to kill bacteria and viruses.
4. Tiredness, also known as fatigue, is a feeling of exhaustion, weariness, or lack of energy that can impact your daily activities. It can be a normal response to physical activity, stress, boredom, or lack of sleep. However, it can also be a sign of an underlying medical condition or mental health issue.
5. Red or purple spots on the skin can have a number of causes, including: Petechiae These are pinpoint-sized spots that look like a rash and can be red,brown, or purple. They're caused by bleedingunder the skin and are often flat and don't fade when pressed.
6. Petechiae can be caused by a number of things, including a lack of vitamin C or K, or a reaction to certain medicines.
7. The medical term for the inability to sweat is anhidrosis. It can be caused by a number of things, including: Medications: Some medications, such as
8. antidepressants, anti-epilepsy drugs, and antipsychotics, can reduce or prevent sweating.
9. Swelling in the lower leg can have many causes, including: Fluid buildup This is often due to sitting or standing for too long, being overweight, or eating too much salty food. It can also be caused by certain medications, such as some blood pressure medicines, contraceptive pills, hormone therapy, antidepressants, or steroids.

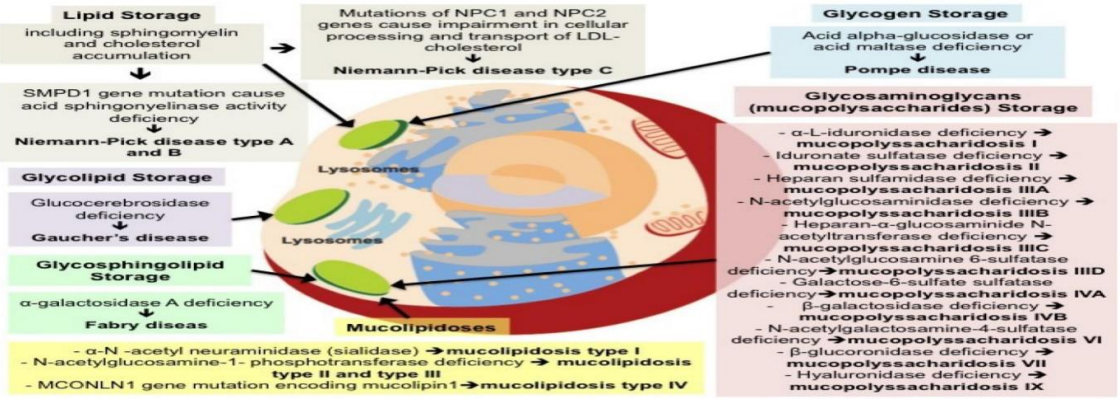

FIG NO.[2]

Causes of lysosomal storage disease

• The majority of lysosomal storage disease caused by mutations in the genes encoding a lysosomal enzyme. These monogenic disorders involve forty different acid hydrolases in the lysosomes.
• Genes with a specific chromosomal locus encode them. Many types of mutations can produce a defective enzyme.
• Inborn error of metabolism
• The fusion of transport vesicles budded from the trans golgi network with endosome.
• The lack of enzymes in the Lysosome. The enzymes present in the Lysosomes helps in digesting food particles, dead cells, old cells and engulfing disease-causing microorganisms including protozoa, fungi, bacteria also viruses. It is also involved in the ingestion of dead cells along with other invading microbes.
• Deficiency of proteins in our body also results in Lysosomal storage diseases as it plays a vital role in modification o f enzymes in Lysosome.
• Most of the Lysosomal storage diseases are inherited in an autosomal recessive manner.
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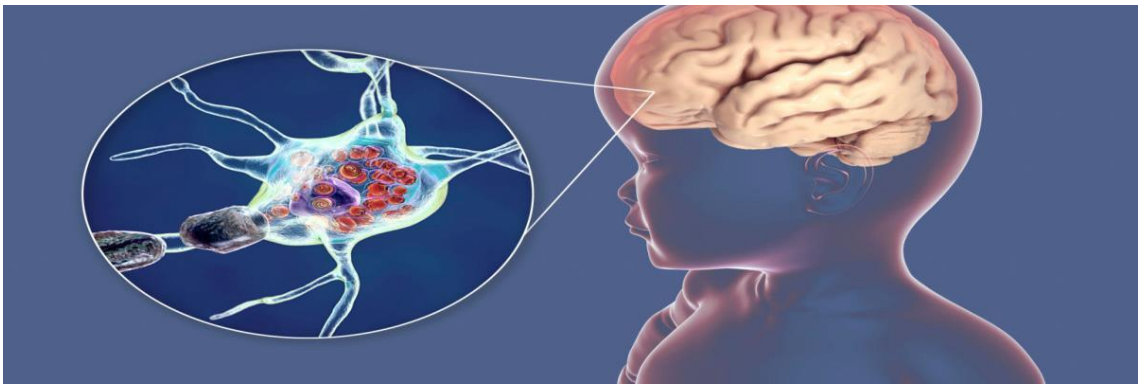

FIG NO[3]

Diagnosis of lysosomal storage disease

1. Clinical identification of MPS patients is not an easy task, and this is particularly true for patients with more attenuated forms of MPS. It was demonstrated that, for example, that the attenuated form of MPS I was recognized in less than 20% of cases.9 As a consequence, patients commonly experience significant diagnostic delays.

2. Because appropriate treatments for many of these conditions now exist, there is an imperative need to diagnose and recognize affected patients as soon as possible to stop the disease progression and prevent the occurrence of irreversible damages. A diagnostic algorithm for rheumatologic evaluation of joint and bone involvement has recently been proposed.[9]

3. The algorithm underlines the importance of considering MPS diagnosis if the development of stiffness or contractures of joints is not followed Clinical identification of MPS patients is not an easy task, and this is particularly true for patients with more attenuated forms of MPS. It was demonstrated that, for example, that the attenuated form of MPS I was recognized in less than20% of cases.[10]

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5. The algorithm underlines the importance of considering MPS diagnosis if the development of stiffness or contractures of joints is not followed of MPSs; it should be performed not only in patients but also their parents to use for genetic counseling and prenatal

6. diagnostics. Unfortunately, there is a lack of strict correlation between residual enzyme activity, genotype, and clinical phenotype, making the care of presymptomatic individuals. more complicated.[12]

7. Newborn screening also plays a role in reliable and prompt identification and classification of MPSs; however, technology requires some time before a routine application.[13]

8. Fig. 3. Knock-knee deformity in a 10-year-old patient with MPS IV (Morquiosyndrome). Finally, diseases that present musculoskeletal features similar to MPSs should be taken into account (eg, multiple sulfatase deficiency, ML, fucosidosis, amainnosidoses, and skeletal dysplasia of non-LSD origin). Because diagnosis is tricky and might be difficult and confusing because of there rare-ness of the diseases and their differential diagnosis, the authors recommend referring any suspicious patient to a specialized center.

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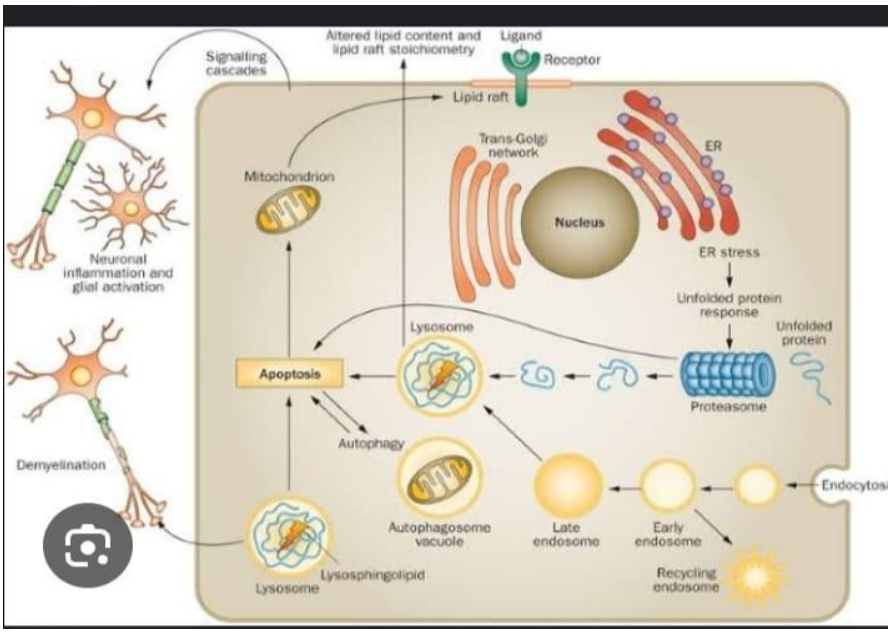

Fig no.[4]

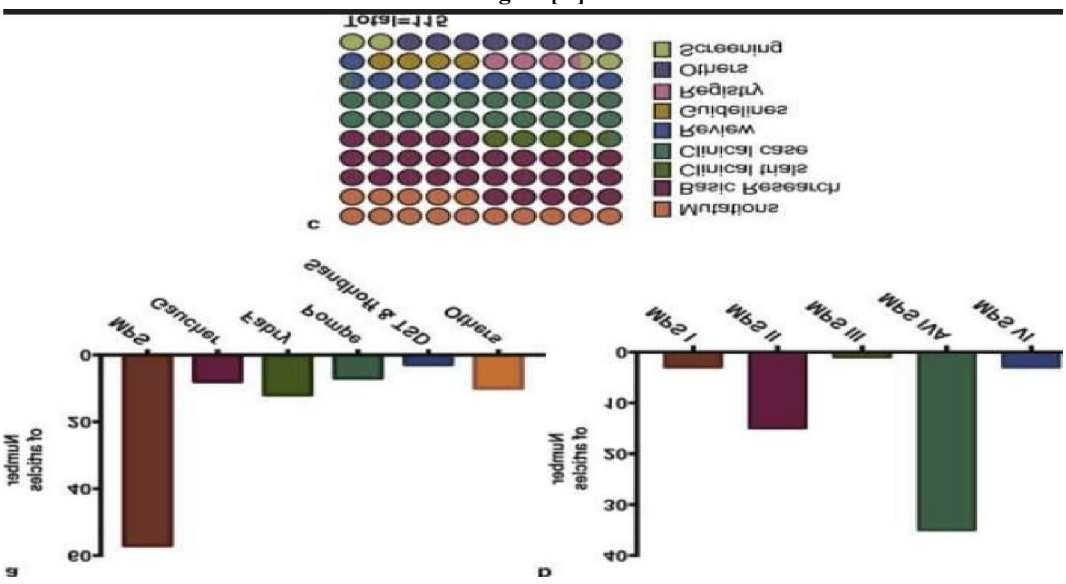

Fig no.[5]

Treatments of lysosomal storage disease

1. Because MPSs are progressive diseases characterized by complex pathophysiologic mechanisms and multisystemic deterioration, the disease may give rise to different symptoms and clinical manifestations, some of which are not easily reversible. For this reason, response to available therapies depends on the severity of the disease, phenotype, and degree of disease progression at treatment initiation.[14]

2. Timely diagnosis and early treatment can improve the outcome. Besides supportive therapy and pain management for MPSs I, II, and VI (in clinical trials, MPS IV A and VII), effective but unfortunately not curative treatments, such as ERT, and, particularly for MPS I, HSCT, are available.[15]

3. Enzyme Replacement Therapy The efficacy and safety of ERT with recombinant human enzyme is well accepted and confirmed by many clinical trials. Enzyme infusion, however, which is a life-long therapy, is administered

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intravenously once a week over approximately 4 hours and can be associated with immune reactions and thus could have a negative impact on dis-ease management.[16]

4. Nevertheless, the benefits include reduction of liver spleen size, improved endurance and shoulder mobility, improved respiration, and enhanced quality of life. So far, ERT efficacy mainly consists of reducing the burden of peripheral diseases; enzymes capable of effectively targeting the bones, especially the CNS, are still to be develop.[17]

5. There are ongoing clinical trials with intrathecal enzyme application in MPS I and MPS III A. ERT is available for MPSs I, II, and VI and in clinical trials in MPS IV A and MPS VII. Hematopoietic Stem Cell Transplantation When performed in patients under the age of 2 years and with a developmental quotient greater than or equal to 70%, HSCT is the gold standard treatment of MPS I patients. It was proved that HSCT may preserve cognitive function and extend life expectancy in severely affected MPS I pediatric patients59 and is under investigation.

Diagnostics in mucopolysaccharidoses

1. Step: screening test Urinary GAG excretion (pitfall: urinary GAG excretion can be in normal range in attenuated patients and in adults)

2. Step: gold standard Enzyme measurement in leukocytes, plasma,heparin blood, or fibroblasts

3. Step: genetic testing as an additional diagnostic test and to facilitate prenatal testing and is mandator in x-linked diseases to detect carriers Dried blood spot testing for MPSs I, II, and VI available Newborn screening under development Families at risk: prenatal testing (chorionic villi and amniocentesis) is available Supportive Therapy and Observation of Disease Progression The multisystemic course of MPSs requires a multidisciplinary team, at its best coordinated by an experienced metabolic physician. Patient disease progression should be observed by regular follow-up examinations at least annually. In addition to pain management, typical comorbidities often need surgical intervention, such as hernia repair, adenoidectomy or tympanostomy tube implantation, orthopedic surgeries (correction of skeletal changes), neurosurgical procedures (carpal tunnel release or craniocervical or thoracolumbar decompression), heart valve replacement, and corneal clouding. Because of the GAG accumulation in all tissues, anesthesia complications are much higher than in unaffected population; thus, every surgical intervention should be considered carefuly.[18,19] In particular, spinal cord compression at the craniocervical junction caused by thickening of the surrounding soft tissues and/or atlantoaxial instability should be recognized as a life threatening complication. Other specialists (eg, cardiologists; pulmologists; ophthalmologists; ear, nose, and throat physicians; physiotherapists; and psychologists) are essential for giving consideration to patients' needs and improving quality of life, including patients with types of MPSs for which no causal therapy is available.

OTHER LYSOSOMAL STORAGE DISEASES

Apart from MPSs, there are other LSDs that manifest with musculoskeletal symptoms. Because symptoms can mimic rheumatologic disorders, rheumatologists play a key role in diagnosis. Fabry Disease Fabry disease is an X linked inherited disorder due to a deficiency of the enzyme agalactosidase, resulting in accumulation of globotriaosyl ceramide and other sphingolipids within lysosomes of almost all cells and tissues, leading to a variability of progressive clinical signs and symptom

1. The oligosccharidoses are rare diseases. Although in most cases, the

diagnosis of LSD is made before rheumatologic symptoms evolve, many patients suffering from mental retardation seeking medical advice for a problem seemingly not related to their underlying condition may receive no accurate diagnosis. Furthermore, their prevalence is higher in certain populations, for instance, in the population of southern Italym (fucosidosis)98 and, as recently shown, in Cuba (fucosidosis and amannosidosi).[21,22] he autosomal recessive inherited diseases, fucosidosis (decreased activity of a-fucosidase) and a-mannosidosis (decreased activity of acid a-mannosidase), share some aspects of MPS, especially mild dysostosis multiplex, recurrent infections, and organomegaly, but progressive neurologic impairment predominate.[23] In the largest overview of patients with fucosidosis, 48% of all patients suffered from joint contractures and 66% from kyphoscoliosis; mental retardation was found in 95%.98 Patients with a-mannosidosis commonly exhibit, besides scoliosis and sternum deformity, genua valga, sometimes needing orthopedic correction100; in older patients, deforming and destructive arthropathy of large joints has been describe.[24]Bone marrow transplantation in the first decade of life seems to promise some benefit in a-

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mannosidosis100 and has been performed in fucosidoses.[25] Other oligsaccharidoses do not exhibitrheumatologic features. In particular, spinal cord compression at the craniocervical junction caused by thickening of the surrounding soft tissues and/or atlantoaxial instability should be recognized as a life-threatening complication. Other specialists (eg, cardiologists; pulmonologists; ophthalmologists; ear, nose, and throat physicians; physiotherapists; and psychologists) are essential for giving consideration to patients' needs and improving quality of life, including patients with types of MPSs for which no causal therapy is available. [26]

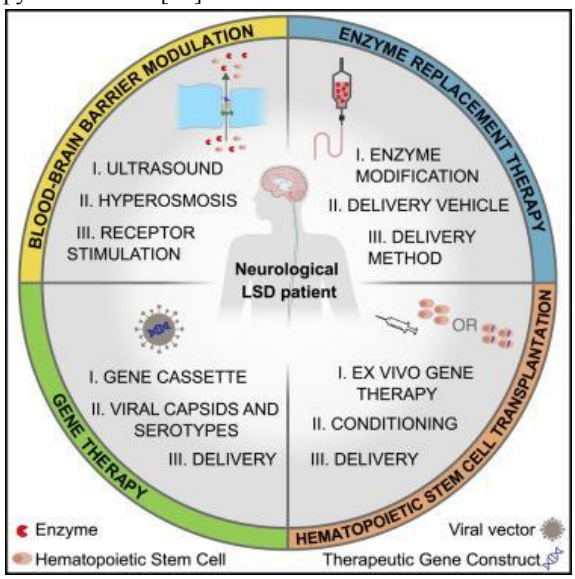

Fig no [6]

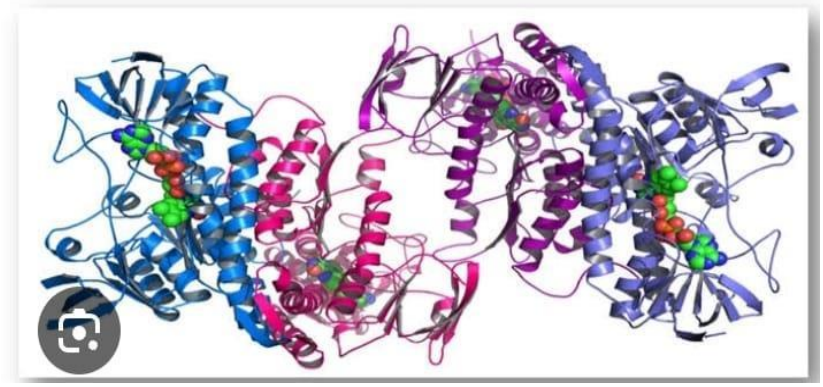

Fig no [7]

Pathophysiology of lysosomal storage disease

DNA methylation

DNA methylation is one of the most widely studied epigenetic modifications involving the transfer of a methyl group to the fifth position of a cytosine nucleotide. While DNA methylation has been appreciated since DNA was discovered as the genetic material in the past two decades, studies of DNA methylation have expanded because of its role in gene regulation. Methylation of Cytosine-phosphate-Guanine (CpG) dinucleotides, particularly in gene promoter regions and other regulatory elements, can directly block the binding of transcription factors [16,17], and can recruit methyl CpG binding domain (MBD) proteins, further attracting histone deacetylases and possibly other factors involved in altering chromatin architecture.[29] The family of DNA methyltransferases (DNMTs) catalyze the transfer of a methyl group from the donor S-adenosylmethionine (SAM) to cytosine residues. DNMT1 is involved in post-replicative maintenance of methylation by binding with proliferating cell nuclear antigen (PCNA). mDNMT3a and DNMT3b are responsible for

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de novo methylation and the spontaneously deaminate into thymidine Approximately 70% of all CpG dinucleotides in the genome are methylated. CpG-nucleotides in GC rich regions, known as CpG islands, are usually located within promoter regions, and have relatively no DNA methylation Exceptions occur during X-inactivation and genomic imprinting. DNA methylation is critical in marking the parental origin of alleles, and certain imprinted genes are influenced by epigenetic processes from parental germ cells, resulting in expression of only one of the parental alleles. However, additional processes, including transcription, histone modifications and higher order chromatin, may also be important in marking imprinted loci.CpG islands tend to occur near the 5 end of housekeeping genes, where they play a role in the regulation of transcription. In the human genome, 72% of promoters have been classified as being high in CpG content, while the remaining 28% have been classified as being high in CpG content, while the remaining 28% have low CpG content. Aberrant methylation of CpG islands has been implicated in diverse diseases including cancer, schizophrenia, congenital heart disease, amyotrophic lateral sclerosis, Alzheimer disease, and atherosclerosis. CpG methylation patterns are critical for proper development, and some have been conserved evolutionarily, such as differences in CpG methylation between neuronal and glial cells, conserved in mice and humans.[30]

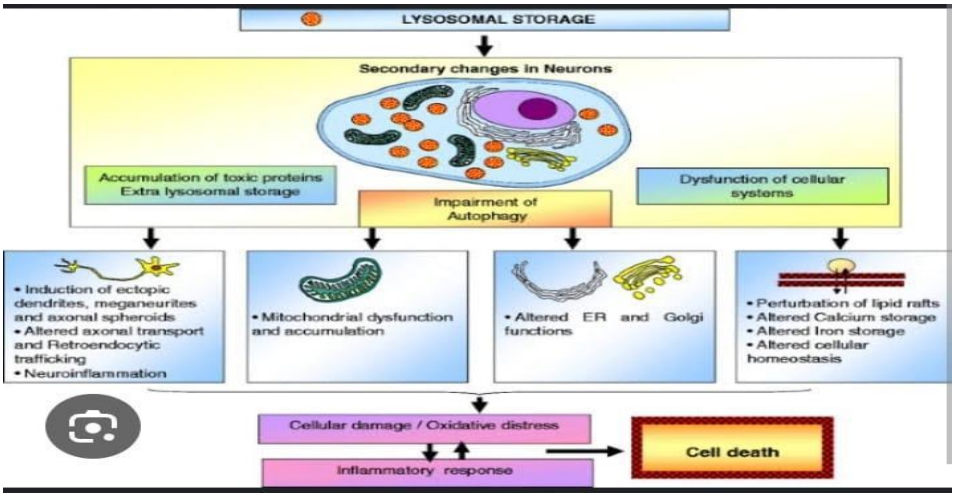

Fig no[8]

Histone modifications

Another class of epigenetic marks includes histone modifications, such as acetylation, phosphorylation, methylation, β-N-acetylglucosamine deamination, ADP ribosylation, ubiquitination, sumoylation, histone tail clipping, and histone proline isomerization.[31]

II. CONCLUSION

By these thesis of lysosomal storage disease we conclude that these disease are not treatable disease. Only we recover these disease highly predictive cellular and animal models are particularly critical of the clinical development of therapies for lysosomal storage disease. Because of the small and mostly juvenile patient population. Therapy of these disorder has made significant progress in the last tow decade. However significant challenges remain at the cellular level ex, precisely how lysosomal storage disease cause pathology. As ERT and HSG only have a very limited effect on brain, bone and cardiac and connective tissues. There is still an unmet clinical need for new drug that can acers these positive screening result will develop symptoms only in late adulthood or perhaps never in their live.

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