Pompe's Disease

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Abstract
Pompe disease is a rare multisystem disorder caused by pathogenic variations in the *GAA* gene containing the information for production and function of a protein called acid alpha-glucosidase (GAA). Because of the shortage of this protein (an enzyme) a complex sugar named 'glycogen' cannot be degraded to a simple sugar like glucose. This causes the glycogen to accumulate in all kinds of tissues, but primarily in skeletal muscle, smooth muscle and cardiac muscle, where it causes damage to tissue structure and function. Pompe disease is inherited as an autosomal recessive genetic trait.

Keywords: Pompe's disease, Autosomal recessive, Enzyme defect.

Introduction
Glycogen storage disease type II, also called Pompe's disease is an autosomal recessive metabolic disorder which damages muscle and nerve cells throughout the body. It is caused by an accumulation of glycogen in the lysosome due to deficiency of the lysosomal acid alpha-glucosidase enzyme. It is the only glycogen storage disease with a defect in lysosomal metabolism, and the first glycogen storage disease to be identified.

History
The disease is name daftera Dutch pathologist Joanne Scassianus Pompe, who identified it in 1932. Pompes described an accumulation of glycogen in muscle tissue in some cases of a previously unknown disorder [Nelson textbook.vol.1].

Epidemiology
The disease affects approximately 1 in 140,000 babies and 1 in 60,000 adults a year, it has been reported in almost all ethnic populations.

Causes
It has an autosomal recessive inheritance pattern. This means the defective gene is located on an autosome, and two copies of the gene-one from each parent are required to be born with the disorder. Although both parents carry one copy of the defective gene, they are usually not affected by the disorder. [kliegmanstamon.st.Gene.schor.]
The disease is caused by a mutation in a gene on the longarm of chromosome 17. [kliegmanStanton.StGene.schor].
The gene encodes a protein –acid alpha-glucosidase which is a lysosomal hydrolasedeficiency of this enzyme accumulation of structurally normal glycogen Excessive glycogen storageinterrupt normal functioninglead tocellular injury.

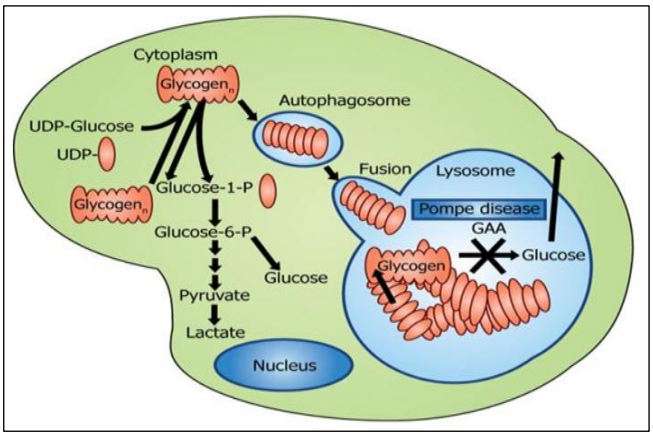

Fig. 1: Pathophysiology of Pompe's disease

Classification
GSDII is broadly divided into two types

1. **Infantile onset form:** Infantile-onset Pompe disease presents before the age of 12 months with cardiomyopathy and may be apparent in utero but more typically onset is at the median age of four months. Without treatment by enzyme replacement therapy [ERT], results in death by the age of two years
2. **Late onset form:** The late-onset type of Pompe disease may not become apparent until later in childhood, adolescence, or adulthood and is milder and rarely affects the heart.

Clinical Manifestations
1. Muscle weakness (floppy infants)
2. Diminished muscle tone (hypotonia)
3. Respiratory insufficiency
4. Hypertrophic cardiomyopathy, a condition characterized by abnormal thickening of the walls of the heart (mainly the left chamber and the wall between the left and right chamber) resulting in diminished cardiac function
5. Many infants have a large, protruding tongue and a moderate enlargement of the liver.

6. The legs often rest in a frog position
7. Feeding and swallowing problems
8. Delay in developmental milestones
9. Hearing loss
10. Failure to thrive

Diagnosis

1. Molecular genetic testing
2. sleep studies
3. breathing tests to measure lung capacity
4. Electromyography- measures muscle function
5. Muscle MRI's are also used to measure the degree of damage that has occurred to the muscles.
6. Electrocardiogram
7. Chest Xrays

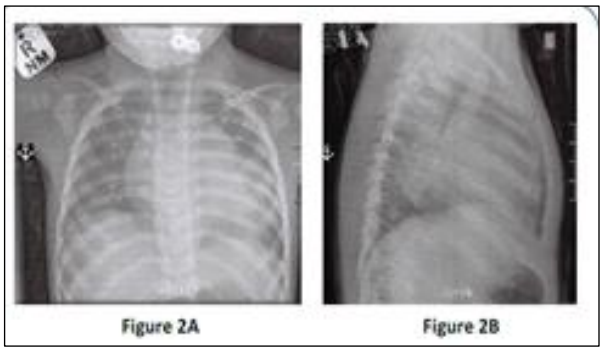

Fig. 2: Chest radiograph showed significant cardiomegaly. Seven month old male with history of pompe disease with significant cardiomegaly and a cardiothoracic ratio measuring 67% and multi-Chamber prominence.

Treatment

1. Cardiac and respiratory complications are treated symptomatically

SupportiveTherapies

Additional treatment of Pompe disease is symptomatic and supportive.

a. Respiratory support may be required
b. Physical therapy may be helpful to strengthen respiratory muscles and also to improve strength and physical ability
c. Some patients may need respiratory assistance through mechanical ventilation (i.e. bipap or volume ventilators) during the night and/or periods of the day

2. Occupational therapy, including the use of canes or walkers or wheelchair may be necessary.
3. Speech therapy can be beneficial to improve articulation and speech for some patients.
4. Alterations in diet may provide temporary improvement but will not alter the course of the disease.

PatientswithPompesdisease haveloss of muscle tissue and there is some evidence that a high-protein diet, in combination with appropriate exercise, can slow this loss. The recommended diet is approximately 25-30 percent proteins, 30-35 percent carbohydrates, and 35-40 percent fat. Proteins from meat, fish, egg, and dairy products are rich in an important amino acid calledAlanine. A feeding tube may be used to give enough nutrients.[Nelson 2015].

5. Genetic counselling can provide families with information regarding risk in future pregnancies.
6. Orthopedic devices including braces may be recommended for some patients. Surgery may be required for certain orthopedic symptoms such as contractures or spinal deformity.
7. Enzyme replacement therapy

Two medications replace the missing protein and help the body process glucose correctly.

a. Myozymes, for babies and children.
b. Lumizymes.

Myozyme is a recombinant form of the human enzyme acid alpha-glycosidase, and is also currently being used to replace the enzyme.

Myozymeadministration is by intravenous infusion. Myozyme treatment clearly prolongs ventilator-free survival and overall survival.

8. Early diagnosis and early treatment leads to much better outcomes.
9. Counselling –helps the family to come to terms to what is happening

Fig. 3: Supportive treatments in Pompe's Disease

Nursing Management

1. Follow strict hand washing and aggressive management of infections.
2. Routine immunizations, including pneumococcal vaccination, Influenza vaccination for patients
3. Careful use of over the counter medications

Prognosis

The prognosis for individuals with pompe disease varies according to the onset and severity of symptoms. Without treatment the disease is particularly lethal in infant and young children.[kleigmanstanton.St.Gene.Schor.vol-1]

Conclusion

Pompes disease is a rare condition seen in India and it requires co-ordinated care and optimal treatment strategies to increase survival and improve the quality of life for patients with this disease.

Conflict of Interest: None.

Reference

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