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Pompe Disease

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Abstract

Pompe disease[1] is a single disease continuum with variable rates of disease progression and different ages of onset. First symptoms can occur at any age from birth to late adulthood. Earlier onset compared to later onset is usually associated with more rapid progression and greater disease severity. At all ages, skeletal muscle weakness and wasting causing mobility problems, but also affecting respiratory function, characterizes the disease. The most severely affected infants usually present within the first 3 months after birth.

Keywords: *Pompe disease[1], wasting[2]*

Introduction

The human body can be seen as an assembly of interconnecting organs. Organs are composed of organ specific tissues, and tissues are composed of specialized cells like muscle cells, nerve cells, etc. Pompe disease belongs to a group of diseases known as the 'lysosomal storage disorders' (LSDs).

Meaning

Infantile-onset or 'classic infantile' refers to the originally described form of Pompe disease characterized by onset of symptoms shortly after birth, generalized muscle weakness, and cardiac hypertrophy, in association with glycogen storage in all organs. Terms like childhood, juvenile, and adult glycogenosis type II (Pompe disease, or acid maltase deficiency) were historically introduced as names for the less severe forms of Pompe disease characterized by delayed onset and usually slower progression. Adult-onset was alternatively called 'late-onset'.

Causes

Pompe disease is caused by pathogenic variations in the acid alpha-glucosidase (GAA) gene. Close to 500 different GAA gene variations have been identified in families with this disorder.

Symptoms

Respiratory

- Progressive respiratory muscle involvement
- Frequent respiratory infection
- Sleep- disordered breathing

Musculoskeletal

- Progressive muscle weakness
- Delayed motor milestones
- Profound hypotonia/floppy baby

Cardiac

- Cardiomegaly
- Progressive cardiomyopathy

Gastrointestinal

- Macroglossia
- Feeding Difficulties
- Failure to thrive
- Hepatomegaly

Diagnosis

A diagnosis of Pompe disease is based upon a thorough clinical evaluation, a detailed patient and family history, and a variety of tests. Prenatal diagnosis is possible when a pregnancy is believed to be at risk for Pompe disease.

Treatment[4]

The treatment of Pompe disease is disease-specific, symptomatic, and supportive. Treatment requires the coordinated efforts of a team of specialists with expertise in treating neuromuscular disorders. Pediatricians or internists, neurologists, orthopedists, cardiologists, dieticians, and other healthcare professionals may need to systematically and comprehensively plan an affect child's treatment. Genetic counseling is of utmost importance for affected individuals and their families.

Enzyme Replacement Therapy

Enzyme replacement therapy is an approved treatment for all patients with Pompe disease. It involves the intravenous administration of recombinant human

acid α-glucosidase. This treatment, manufactured by Genzyme, a Sanofi Corporation, is Lumizyme (marketed as Myozyme outside the United States), and was first approved by the U.S. Food and Drug Administration (FDA) in 2006. It has been approved for all patients with Pompe disease.

Supportive Therapies

Additional treatment of Pompe disease is symptomatic and supportive. Respiratory support may be required, as most patients have some degree of respiratory compromise and/or respiratory failure. Physical therapy may be helpful to strengthen respiratory muscles. Some patients may need respiratory assistance through mechanical ventilation (i.e. bipap or volume ventilators) during the night and/or periods of the day. In addition, it may be necessary for additional support during respiratory tract infections. Mechanical ventilation support can be through noninvasive or invasive techniques. The decision about the duration of respiratory support is best made by the family in careful consultation with the patient's physicians and other members of the healthcare team based upon the specifics of the patient.

Physiotherapy is recommended to improve strength and physical ability. Occupational therapy, including the use of canes or walkers, may be necessary. Eventually, some individuals may require the use of a wheelchair. Speech therapy can be beneficial to improve articulation and speech for some patients.

Orthopedic devices including braces may be recommended for some patients. Surgery may be required for certain orthopedic symptoms such as contractures or spinal deformity.

Ongoing Disease Progression[5]

Pompe disease is always progressive, meaning that its symptoms worsen over time. In general, the earlier in life the symptoms appear, the faster the rate of progression. Most infants affected by the disease experience very rapid progression, and they rarely survive past the age of 1 year.

When symptoms first appear later in life (children or adults), the rate of progression is generally slower than in infants, although there is great variability across different people. In addition, an abrupt and rapid decline can happen at any time, so careful monitoring of the disease's progression is very important. Whether the disease progresses fast or slowly, eventually movement and breathing difficulties worsen over time.

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