Living with Pompe Disease

There are up to 300 different mutations in the GAA gene that cause the symptoms of Pompe disease.

1. Introduction

Pompe disease is a rare genetic disorder. Mutations in the GAA gene reduce or eliminate an essential enzyme that the body uses to break down glycogen, causing a buildup of this complex sugar in the body's cells. This buildup often occurs in the heart and skeletal muscles, impairing their ability to function normally.

2. Types

There are two main types of Pompe disease:

+ **Infantile-onset** — A complete or near complete deficiency of GAA. Symptoms begin within the first year of life, usually around 4 months of age, and include an enlarged heart. This form of the disease progresses quickly, and without treatment, most babies will die within the first year or two from cardiac complications.
+ **Late-onset** — A partial deficiency of GAA. Symptoms may appear before the age of 1 but without an enlarged heart or later in a child's life during adolescence or into adulthood. This form is usually milder and progresses more slowly.

3. Symptoms

Symptoms differ between the two forms of this disease. Symptoms of the infantile-onset form include:

+ Poor muscle tone or floppy infant syndrome
+ Enlargement of the heart, liver and tongue
+ Trouble gaining weight and growing at the expected rate
+ Difficulty breathing and feeding issues
+ Respiratory infections

Late-onset symptoms may be milder and progress slower. Symptoms may include:

+ Progressive muscle weakness in the legs and trunk
+ Increased difficulty walking
+ Muscle pain over a large area
+ Falling often
+ Shortness of breath
+ Difficulty swallowing

4. Treatments

Enzyme replacement therapy is part of the treatment for both forms of Pompe disease. Other treatments may involve physical or occupational therapy, a feeding tube or mechanical ventilation, depending on the individual symptoms and their severity.

Did you know?

Pompe disease affects about 1 in every 40,000 people in the U.S.

For more information on Pompe disease and supportive resources, please visit rarediseases.org.

References:

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