Pompe Disease

Pompe disease is a rare genetic disorder. It is caused by a deficiency in the enzyme acid alpha-glucosidase (GAA). This is needed to break down glycogen. That is a stored form of sugar used for energy. The build-up of glycogen causes progressive muscle weakness throughout the body. It affects various body tissues, particularly in the heart, skeletal muscles, liver, and nervous system. Children have a 1 in 4 chance of inheriting the disease when both parents carry the abnormal gene. This disease is also known as infantile acid maltase deficiency and type 2 glycogen storage disease. It is estimated to occur in about 1 in 40,000 births.

Pompe disease has three forms. They are defined by age of onset and progression of symptoms.

- *Infantile*, or early onset. This is noticed shortly after birth. Symptoms include severe lack of muscle tone, weakness, and enlarged liver and heart. Mental function is not affected. Development appears normal for the first weeks or months. But it slowly declines as the disease progresses. Swallowing may become difficult. Also, the tongue may protrude and become enlarged. Most children die from respiratory or cardiac complications before 2 years of age.

- *Juvenile* onset. Symptoms appear in early to late childhood. They include:
  - Progressive weakness of respiratory muscles in the trunk, diaphragm, and lower limbs.
  - Exercise intolerance.

Intelligence is normal. Most patients do not live beyond the second or third decade of life.

- *Adult* onset. Symptoms also involve generalized muscle weakness and wasting of respiratory muscles in the trunk, lower limbs, and diaphragm. Many patients report:
  - Respiratory distress.
  - Headache at night or upon waking.
  - Decreased deep tendon reflexes.
  - Proximal muscle weakness, such as difficulty in climbing stairs.

Intellect is not affected. A small number of adult patients live without major symptoms or limitations

TREATMENT

Cardiac and respiratory complications are treated symptomatically. Physical and occupational therapy may be helpful for some patients. Changes in diet may provide temporary improvement. But they will not change the course of the disease. Genetic counseling can provide families with information regarding risk in future pregnancies.

The prognosis for individuals with this disease varies according to the onset and severity of symptoms. The disease is particularly lethal in infants and young children.

RESEARCH BEING DONE

The NINDS supports research on glycogen storage diseases. This includes Pompe disease. The goal is to find effective treatments or prevention strategies. Early studies used enzyme replacement for this disease. But they were unsuccessful. That led to a halt of such trials for some time. Recent trials in human and animal models have shown some success. A study of 3 infants with classic infantile Pompe disease, who were given twice-weekly injections of recombinant human GAA (a genetically engineered acid maltase protein), showed improvement in skeletal muscle function and lasting improvement in cardiac function. The babies have reached 2 years of age. But they were not expected to live beyond age 1 without treatment. Other research involves gene therapy for acid maltase deficiency in animal models.

Research is continually being done to learn more about this problem.

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