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Pompe Disease Fact Sheet

What is Pompe Disease?

Pompe disease is a rare (estimated at 1 in every 40,000 births), inherited and often fatal disorder that disables the heart and muscles. Pompe disease affects approximately 5,000 to 10,000 people worldwide. It occurs in babies, children, and adults who inherit a defective gene from each parent that causes a deficiency in one of the enzymes required to break down carbohydrates. The disease manifestations can vary greatly from one person to the next.

Early onset (or infantile Pompe disease) is the result of complete or near complete deficiency the enzyme. Symptoms begin in the first months of life, with feeding problems, poor weight gain, muscle weakness, floppiness, and head lag. Respiratory difficulties are often complicated by lung infections. The heart is grossly enlarged. Most babies with Pompe disease die from cardiac or respiratory complications before their first birthday without treatment.

Late onset (or juvenile/adult) Pompe disease is the result of a partial deficiency of the enzyme. The onset can be as early as the first decade of childhood or as late as the sixth decade of adulthood. The primary symptom is muscle weakness progressing to respiratory weakness and death from respiratory failure after several years.

A diagnosis of Pompe disease can be confirmed by screening for the common genetic mutations or measuring the level of GAA enzyme activity in a blood sample—a test that has 100 percent accuracy.

Is there any treatment?

In 2006, the FDA and EMEA approved Myozyme® (alglucosidase alfa), an enzyme replacement therapy (ERT) for the treatment of Pompe Disease. However, despite this approval, patients over the age of 18 are often unable to access treatment in the United States.

For More Information:

For more information about Pompe Disease please visit the following websites:

Acid Maltase Deficiency Association, Inc.
<http://www.amda-pompe.org>

International Pompe Association
<http://www.worldpompe.org>

Pompe Disease–The Real Story
<http://www.pompestory.blogspot.com/>