15 April 2016
Together We Are Strong
International Pompe Day

For Immediate Release

April 15, 2016 will be the Third Annual International Pompe Day. The International Pompe Association (IPA), with the support of the global Pompe community, launched International Pompe Day in 2014. The goal of International Pompe Day is to raise global awareness of Pompe disease, a rare neuromuscular condition that affects approximately 1 in 40,000 people around the world.

When we launched International Pompe Day, the Pompe Community selected "Together We Are Strong" as its motto. We believe that as a united community of patients, family members, friends, scientists, doctors, and industry partners we have accomplished, and can continue to accomplish, great things. Together we have seen a treatment for Pompe become a reality for most patients around the world, and working together we see new and better treatments on the horizon in the near future.

This year, as a global community we turn our attention and advocacy power to the importance of early diagnosis in Pompe disease. Unfortunately, many patients still go through a lengthy diagnostic odyssey before they are finally diagnosed. This must be changed to give Pompe patients around the world the best chance possible at preventing irreversible damage to their muscles.

Why is Early Diagnosis Important?
Pompe disease is a progressive, neuromuscular disease. While patients may not show symptoms for many years (sometimes even decades), the disease is present and active at birth. Multiple trials have shown that treatment with enzyme replacement therapy (ERT) can significantly alter the natural history of Pompe disease in patients. In short, it saves lives.

However, studies have also shown that a critical component of a successful outcome with ERT is the initiation of treatment before irreversible damage has occurred. The importance of early diagnosis is even more critical in Infantile-Onset patients. In fact, in "Very Early Treatment for Infantile-Onset Pompe Disease Contributes to Better Outcomes," the authors reported "we found that starting ERT even a few days earlier was associated with better biochemical responses and developmental outcomes" (http://amda-pompe.org/downloads/publications/Very Early Treatment for Infantile-Onset Pompe Disease Contributes to Better Outcomes.pdf)

The key to early initiation of treatment is early diagnosis. Without a diagnosis it is impossible to start treatment before irreversible damage has occurred. However, achieving an early diagnosis is complicated by the fact that symptoms of Pompe can occur at any age, and are often difficult to detect at first.

What are the Early Symptoms of Pompe disease?
The first (or early) symptoms of Pompe will often vary from person to person. It is important to remember that not all symptoms may be present, and that the severity and order of the symptoms may vary. Knowing and recognizing the early symptoms of Pompe disease is critical to achieving an early diagnosis.

In Infantile-Onset Pompe, the most common symptoms are related to poor muscle tone and heart involvement. Infants generally have poor muscle tone (i.e. are a "floppy baby"), fail to meet or maintain development milestones, have difficulty feeding (sucking, swallowing, or feeding in general), and have frequent chest infections. An enlarged heart is also seen in Infantile-Onset patients. These symptoms are typically seen in the first weeks, or months of life.

Late-Onset Pompe disease symptoms can present at any time from the age of 1 to 70 years old. In addition, the order the symptoms present can vary widely from patient to patient. If symptoms begin in childhood/young adulthood, the symptoms tend to worsen more quickly than if they present later in life. However, the most common early symptoms are similar regardless of when they present. These symptoms are: morning headaches/nausea; heavy breathing after mild/moderate exercise; difficulty breathing while lay flat; frequent chest infections that are difficult to overcome; failure to meet or maintain developmental milestones; poor performance in physical activities, difficulty gaining or maintaining a healthy weight; and difficulty with stairs, or getting off the floor.

A more detailed list and description of the early symptoms of Pompe disease can be found on the IPA website at http://worldpompe.org/index.php/pompe-disease/diagnosis#symptoms.

Once Pompe disease is suspected, there are a variety of quick and easy methods to diagnose (or rule out) Pompe disease. We have to get to the point where Pompe disease is suspected sooner, before irreversible muscular damage occurs.

How You Can Help

Pompe disease is so rare, most people have never heard of it. How can you recognize and diagnose a disease if you don't know it exists? But we can all help to change that! Let's show the world that "Together We Are Strong" and spread the word about the importance of Early Diagnosis for Pompe disease and how to recognize Pompe symptoms!

Flyers about the importance of early diagnosis have been created and translated into multiple languages for the patient community to use. Print some Flyers out and share and we will get the word out that Early Diagnosis is Key! The Flyers, and more information about Early Diagnosis of Pompe disease can be found at http://worldpompe.org/index.php/pompe-disease/diagnosis#flyers.

About Pompe Disease

Pompe disease, also termed glycogen storage disease type II or acid maltase deficiency, is an inherited lysosomal storage disorder with an estimated frequency of 1 in 40,000 births. The disease is characterized by a total or partial deficiency of the enzyme acid α-glucosidase. Deficiency of acid α-glucosidase leads to accumulation of lysosomal glycogen in virtually all cells of the body, but the effects are most notable in muscle. Pompe disease is a spectrum disease with classic infantile onset at the severe end of the spectrum and the late onset at the other end of the spectrum. Disease symptoms may include: heart involvement; breathing problems; difficulty walking; inability to walk or stand; inability to raise the arms; muscle pain; fatigue; and scoliosis. Pompe disease causes severe disability and can be fatal when not treated in time.

About the IPA

The International Pompe Association (IPA) is an international federation of Pompe disease patient's groups that seeks to coordinate activities and share experience and knowledge between different groups and individual patient advocates from around the world. The IPA was founded in the Netherlands in 1999 and is the first, and only, international Pompe organization and serves as a model for other disease groups. There are currently sixty (60) countries that are affiliated with (or contacts for) the IPA. For more information on the International Pompe Association, please go to www.worldpompe.org