



Together We Are Strong
International Pompe Day

For Immediate Release:
April 9, 2014

The IPA is excited to announce that the international Pompe community has chosen April 15, 2014 as the first annual International Pompe Day. The goal is to foster international awareness of Pompe Disease under the slogan "Together We Are Strong." As a united community of patients, family, friends, physicians, scientists, and industry we have an undeniable strength. It is as a united community (the Pompe Model) that we have made great strides in learning more about Pompe Disease and in working towards treatments and disease management. It is as a united community that we support each other when the daily struggles of living with Pompe become too much. Nothing that has been accomplished would have been possible without everyone working together. Together We Are Strong!

The International Pompe Association (IPA) is organizing a variety of awareness activities including an online Photo Gallery! Many other events are being planned around the world to mobilize the community and increase awareness. For more information please visit the IPA website www.worldpompe.org to see how YOU can participate!

Why April 15?

April 15th was chosen by the Pompe Community to honor Dr. Pompe.

Pompe disease was named after the Dutch pathologist Dr. J.C. Pompe who presented in 1932 the case of a 7-month old infant who had died of idiopathic hypertrophy of the heart. In addition to the cardiac problems, the infant had generalised muscle weakness. Dr. Pompe made the crucial observation that the baby's symptoms were associated with massive storage of glycogen in virtually all tissues. Dr. M. Putschar reported a similar case in the same year. In the listing of metabolic glycogen storage diseases, made by Dr. G.T. Cori in 1954, Pompe disease was ranked as number 'two' and named 'Glycogen Storage Disease type II' (GSD II).



Dr J C Pompe became active in the active resistance by finding hiding places for the Jews. When he was forced to become member of the Nazi Medical Association he refused to practice as a physician as he didn't want to be associated to any Nazi practice. He also kept an illegal transmitter in his laboratory to broadcast messages to the UK on behalf of the resistance. During a raid the Nazi's found the transmitter and Dr. Pompe was arrested. On 15th April 1945, at 8.30pm, in St,Pancras Netherlands, he was executed by a firing squad, together with 19 other Dutch men, as a reprisal to the resistance who killed two SS officers a day earlier

On May 5th,1945, the Netherlands were liberated by the Allied Forces.

What is Pompe Disease?

Pathology

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. This enzyme is needed to break down glycogen that is stored within the lysosome, a cytoplasmic organelle involved in cellular recycling and tissue remodeling. 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, an organ present in all areas of the body.

Presenting symptoms

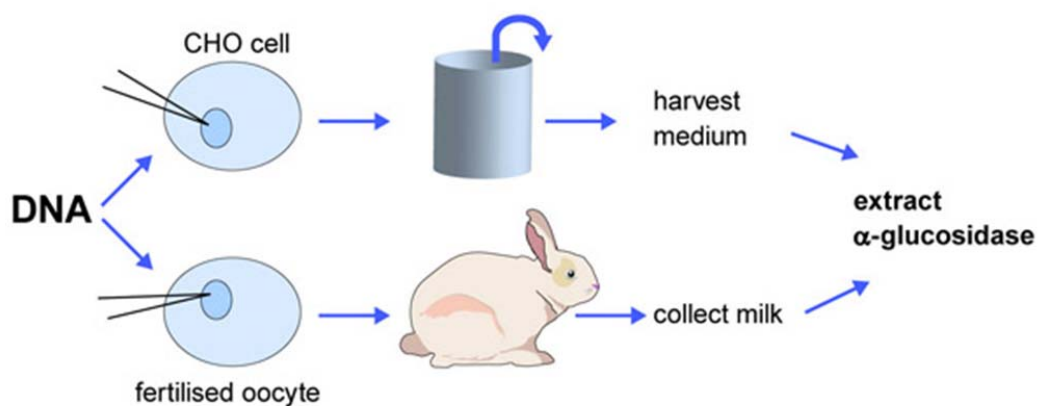
Pompe disease is a spectrum disease with classic infantile onset at the severe end of the spectrum and the late onset at the other side of the spectrum. Patients at the severe end of the spectrum typically present within the first several months of life. Affected infants have profound and progressive muscle weakness manifested in a 'floppy baby' appearance. Other key manifestations include respiratory insufficiency, enlarged heart, failure to reach developmental milestones, feeding difficulties and an enlarged liver

There also are 'milder' forms of Pompe disease presenting later in life from the age of 1 to the age of 99 with progressive weakness of mainly the skeletal muscles of the trunk and the lower limbs. In general one can say that the earlier symptoms appear in life the more severe the disease will develop. Patients often have a waddling gait and may have difficulty taking stairs or rising from a chair. Common respiratory symptoms include shortness of breath, sleep-disordered breathing, and progression to respiratory failure. Other symptoms may include scoliosis, scapular winging, muscle pain, fatigue and frequent falls. If Pompe patients are untreated they will need respiratory support, they become wheelchair dependent, will not be able to lift their arms and it can be fatal. When Pompe disease is suspected, the diagnosis can be confirmed by demonstrating the lack of acid α -glucosidase activity in cells or tissues of the patient. The acid α -glucosidase deficiency in Pompe disease is caused by pathogenic mutations in the gene (DNA). If the mutations are known, Pompe disease can also be diagnosed by DNA analysis.

Treatment

Pompe disease has long been an untreatable disorder, for which only supportive care was available. In March 2006 the first treatment for patients with Pompe disease, received marketing authorization in the European Union, followed in April 2006 by FDA approval in the United States. It is an 'enzyme replacement therapy' (ERT). The rationale for this therapy is to treat the disease by intravenous administration of the deficient enzyme.

The marketing approval of the first ERT for Pompe disease was the result of many years of research and development. During the development of the therapy, two methods using recombinant DNA technology have been explored: production of the enzyme in the milk of transgenic rabbits and production in Chinese Hamster Ovary cells (CHO-cells). The current product is recombinant human acid α -glucosidase derived from CHO-cells.



At the moment treatment is available in more than 40 countries. It gives hope and a new perspective to patients and their families who have to deal with this very rare disease. The IPA by starting this awareness campaign wants to pinpoint the effect working together can have. Together as a world wide community we have proven to be strong and effective. Together We Are Strong!

***Thank you to Erasmus University Medical Center for providing the pictures and the technical information on Pompe Disease.*

For more information please go to www.worldpompe.org