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European Commission
Health and Consumer Protection Directorate-General Rare Diseases consultation
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The AMDA is a national Pompe disease patient organization based in San Antonio, Texas, USA. Pompe disease is a very rare progressively fatal neuromuscular disease that affects an estimated 5,000-10,000 patients worldwide. Pompe disease is one of a family of 49 rare genetic disorders known as Lysosomal Storage Diseases or LSDs. Pompe disease is also known as Acid Maltase Deficiency or Glycogen Storage Disease type II.

Pompe disease is clinically heterogeneous in the age of onset, the extent of organ involvement, and the rate of progression. The early onset form of the disease is the most severe, progresses most rapidly, and is characterized by musculoskeletal, pulmonary, gastrointestinal, and cardiac symptoms that usually lead to death from cardio-respiratory failure between 1 and 2 years of age. The late onset form of the disease begins between childhood and adulthood and has a slower rate of progression that is characterized by musculoskeletal and pulmonary symptoms that usually lead to progressive weakness and respiratory insufficiency.

The National Organization for Rare Disorders (NORD) and rare-disease patient organizations across the United States are celebrating the 25th anniversary of the signing of the Orphan Drug Act during 2008. This groundbreaking legislation brought real hope to the more than 25 million Americans living with one of the nearly 7,000 diseases considered to be rare.

The U.S. Orphan Drug Act has touched the lives of all of our patients diagnosed with Pompe disease. This Orphan Drug Act encouraged pharmaceutical and biotechnology companies to take an interest in diseases that had previously been ignored. The research that resulted from this interest has led to a treatment for Pompe disease, and many other diseases. It has also created an awareness of the existence of these rare diseases.

The AMDA believes the first, and most important step in battling rare diseases is raising awareness. By their very nature, these rare diseases often live in the shadows; they are not known to the general public or even to the average doctor. People with Pompe disease usually spend years going to different doctors trying to find out what is wrong with

them. Most doctors dismiss the early symptoms of Pompe as being nothing to worry about. These doctors just do not know enough about Pompe to put the pieces together.

Improving the path to diagnosis and quality of care for patient with a rare disease can be initiated by developing a national or regional center. At these centers scientists and experts in rare diseases could improve the availability and accessibility of accurate clinical and genetic tests.

Our U.S. patients benefit from these types of institutions because they give them the opportunity to meet, individually with physicians, genetic counselors, dietitians and physical therapists who specialize in lysosomal and genetic diseases. This allows them to have their disease accurately diagnosed and properly treated. For instance, many of our patients do better if they initiate a special low carbohydrate diet and an exercise program while waiting for treatment. This change in lifestyle is made easier by being able to meet with a team of specialists well versed in how to care for a person with Pompe disease.

The AMDA would like to take this opportunity to work with European countries in order to strengthen the cooperation between the various programs. It is important to develop national health policies for rare diseases and to ensure that common policy guidelines in this field are shared throughout Europe. The AMDA has found that it is essential that knowledge and identification of rare diseases be improved. This allows patients to be diagnosed faster; provides them with a better quality of care; and encourages research that can lead to treatments being developed for rare diseases that otherwise receive little attention.

Sincerely,

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Director AMDA