



GETTING THE RIGHT CARE FOR POMPE DISEASE

It takes a team of healthcare providers to meet all the needs that may arise as Pompe disease progresses. Each member of your healthcare team has unique skills to offer. One doctor may serve as team leader and coordinate the care given by all healthcare providers. But because Pompe disease is so rare, you may see providers who have never treated anyone with this neuromuscular disorder. It will help to work closely with the medical team to get the treatment and services you or your child really need. Learning all you can about Pompe disease can help you take charge of your care. This handout will get you started.



Q: I know it's important to learn about Pompe disease, but where do I begin?

A: You can find good sources of information on the Internet. Much of it comes from groups that were started by patients or parents of children with Pompe disease. You can also learn a lot from groups that support research into other neuromuscular disorders, as well as from government agencies and major medical centers around the world. (The list of resources in Where to Learn More on the last page can help you get started).

Q: How is Pompe disease treated?

A: There is an effective enzyme replacement therapy called Myozyme that is commercially available in many countries worldwide. This treatment is intended to stop the progression of the disease and/or improve function.

Myozyme is marketed inside the United States (USA) as both Myozyme (160L) and Lumizyme (4000L) but both have the generic name of alglucosidase alfa. The name change from Myozyme to Lumizyme was based on the US Food and Drug Administration (FDA) determination that the Myozyme produced in the larger scale (4000L) possessed slightly different biochemical characteristics than the original Myozyme produced in the smaller scale (160L), and should therefore be classified as a different drug with a different name. In the US, Myozyme continues to be produced for

GETTING THE RIGHT CARE FOR POMPE DISEASE

the treatment of infantile / early onset Pompe disease. In all countries outside of the USA, alglucosidase alfa is called Myozyme. All Pompe patients in these countries receive Myozyme from the 4000L. The brochures will refer to both Myozyme and Lumizyme as just Myozyme. In the USA, Lumizyme is not available for children who were diagnosed with infantile Pompe disease regardless of age or to anyone under the age of 8 years.

Supportive therapies help people with Pompe disease keep up their strength and prevent health problems caused by muscle weakness. Supportive therapies are tailored to the specific needs of the patient. Respiratory therapy can provide the support needed for breathing. Physical therapy can help increase movement, lessen stiffness, strengthen muscle, and ease pain. Dietary therapy helps to maintain calories and prevent weight loss. Everyone with Pompe disease faces new challenges as their body mechanics change. Getting the right therapy from a knowledgeable medical team combined with the support and care of those around you can help you live each day to its fullest.

Q: Who are the key people on my healthcare team?

A: It takes a wide range of skills to meet the needs of someone with Pompe disease. In addition to your family doctor or primary care provider, the chart below lists possible members of your medical team. The team will need you to keep them informed if health changes or new symptoms occur. Be sure each team member knows the care you are getting from other team members. If you have a child with Pompe disease, you will need to keep track of your child's symptoms and reactions to treatment and assist with supportive therapies.

Primary Care Physician:	A physician who provides both the first contact for a person with an undiagnosed health concern as well as continuing care of varied medical conditions, not limited by cause, organ system, or diagnosis.
Pediatrician:	A doctor that specializes in the medical care of children.
Medical Geneticist:	The study of diseases and disorders that is at least partially genetic in origin.
Neurologist:	A specially trained physician who diagnoses and treats disorders in the nervous system. This includes diseases of the brain, spinal cord, nerves, and muscles.
Cardiologist:	A specialist in the structure and function and disorders of the heart.
Pulmonologist or Respiratory Specialist:	The branch of medicine that deals with dysfunction of the respiratory system. Treats the breathing problems caused by muscle weakness.
Gastroenterologist:	A physician who specializes in the diagnosis and treatment of disorders of the gastrointestinal tract. Treats problems related

GETTING THE RIGHT CARE FOR POMPE DISEASE

	to digestion and feeding difficulties.
Orthopedist:	A specialist in treating and correcting deformities of the skeletal system. An orthopedist treats scoliosis (curvature of the spine), contractures (muscle tightness), and other joint or bone problems related to muscle weakness.
Respiratory Therapist:	Work with the pulmonologist to treat patients who have difficulty breathing. They operate equipment such as respirators and ventilators.
Dietitian:	Concerned with the promotion of good health through proper diet and with the therapeutic use of diet in the treatment of disease.
Genetic Counselor:	Deals with individuals and families who have a genetic disease or who are at risk for such a disease. Genetic counseling is designed to provide patients and their families with information about their condition, risks for future pregnancies, and treatments to help them make informed decisions.
Psychologist:	A person trained and educated to perform psychological therapy, testing and research. Helps people cope with their fears and worries and deal with practical concerns.
Social Worker:	Draws on the social and behavioral sciences to meet the needs of clients including financial, family and resources.
Physical Therapist	Help individuals with disabling conditions by providing services that restore function, improve mobility, relieve pain, and prevent or limit permanent physical disabilities. They restore, maintain, and promote overall fitness and health.
Occupational Therapist:	Help to enable people to participate in the activities of everyday life. They achieve this outcome by enabling people to do things that will enhance their ability to participate or by modifying the environment to better support participation.
Nurse:	Assists people, sick or well, in the performance of the activities contributing to health or its recovery that they would perform unaided if they had the necessary strength, will or knowledge and to do this in such a way as to help the person gain independence as rapidly as possible.

Q: How do I find a medical center that has experience in dealing with Pompe disease?

A: There are only a handful of medical centers around the world that specialize in treating Pompe disease. But clinics and rehabilitation centers that treat similar neuromuscular disorders or rare genetic diseases may also provide expert care for people with Pompe disease. In the United States, the Muscular Dystrophy Association (MDA) has more than 235 hospital-affiliated clinics with nurses, physicians and therapists experienced in neuromuscular diseases. Pompe patients may access all of

GETTING THE RIGHT CARE FOR POMPE DISEASE

these services. The MDA is a not-for-profit organization whose goal is to fight neuromuscular diseases through a nationwide research effort, a program of patient medical services and extended education for professionals and families. Pompe disease is one of about 40 neuromuscular diseases included within the scope of the MDA's mission and services.

Pompe families will find of particular benefit a book published by MDA entitled "*Learning to Live with Neuromuscular Disease—a message for parents.*" This book addresses topics such as: taking on the challenge; facing your feelings; the importance of reaching out; strength in family relations; and, what your child needs. The book can be found on the MDA web site.

The MDA web site contains useful information. See www.mdaua.org or the MDA can be contacted by telephone at (800) 572-1717 (USA) or (520) 529-2000 (International).

In Europe and other continents, there are also many specialized centers for people with neuromuscular disorders. To locate medical centers in other countries, see *Where to Learn More* on the next page.

Q: How can the family stay involved?

A: Recognizing that the "family" is the constant in a patient's life, a plan of care should be developed by the family and treating physician/provider that centers on the needs identified by the family and health care providers working as a team. Families and patients should form a partnership with the professional team at all levels of care to allow for equal participation in establishing the goals of care. Multiple needs which may include separation from core family members due to hospitalization or therapy, transportation, lodging costs, loss of work days, specialized equipment including an adapted vehicle, household assistance, regular medical procedures, vocational retraining, and educational programming could be addressed. Information about therapies, treatments, and resources can be continuously shared with the family and/or patient in an appropriate and supportive manner that is sensitive to your family relationships. Timely referrals to appropriate community agencies, including a social worker or other case workers, and advocating for the needed community services are essential support that can be requested of the individual coordinating the care of the patient. The family and all providers should update the care plan and patient progress at regular intervals. This will insure comprehensive coordinated care that is family centered creating a partnership in care.

Q: Beyond medical advice, where can I turn for help with all the practical challenges I will face?

A: There is a vast network of resources and information, both public and private, that can ease the burdens of this disease and add greatly to the Pompe patient's quality of life. This network of resources can help review your finances, apply for government

GETTING THE RIGHT CARE FOR POMPE DISEASE

benefits and financial aid, find medical and social services, set up an education plan for your child, arrange for home care, learn your legal rights, and deal with employment issues and insurance problems. To find sources of support in your community, your country, or on the Internet, see *Where To Learn More* on the next page.

Parent to parent and patient to patient support has been reported by many to be one of the most valued resources provided. Another consideration is to identify support for siblings, grandparents and other family members.

Q: I feel like I know more about Pompe disease than some of the healthcare providers who are treating my child. What can I do about this?

A: Many caregivers and patients with rare disorders like Pompe disease feel that the only way to be sure of getting the right treatment is to learn all they can about the disease. Knowledge can be a real advantage when you are dealing with a large number of healthcare providers. You should keep track of what everyone is doing. This may help prevent mistakes or errors from occurring. There may be times when it is necessary to educate a pediatrician, or family physician, or emergency room doctor who has not had much experience with Pompe disease. Think of your knowledge as a skill you have to share with the healthcare team. And when you have concerns, do not hesitate to offer your opinion or question why something is being done. Remember that no one has a greater stake in your care or your child's care than you do.

Where to Learn More:

Some of the information for this brochure was obtained from the following resources:

- American College of Medical Genetics (ACMG) Practical Guideline: Pompe Disease Diagnosis and Management Guideline 2006. Vol. 8. No. 5. The ACMG guidelines were designed as an educational resource for physicians and other health care providers.
- The Genzyme Corporation's Pompe Community website offers the Pompe community comprehensive information on the disease, as well as resources and support to help manage the challenges it may bring www.pompe.com:
- Center for Lysosomal and Metabolic Diseases Erasmus MC University Medical Center www.erasmusmc.nl/

Information, Advice, and Support

- The International Pompe Association (IPA) is a global federation of Pompe disease patient groups. The IPA helps patients, family members, and healthcare providers from around the world share their experiences and knowledge across continents and cultures. To find the contact for your country, visit the IPA Web site at www.worldpompe.org

GETTING THE RIGHT CARE FOR POMPE DISEASE

- Acid Maltase Deficiency Association (AMDA): The AMDA was formed to assist in funding research and to promote public awareness of Acid Maltase Deficiency, also known as Pompe's Disease. Visit the website www.amda-pompe.org
- Understanding Pompe Disease is a free booklet that gives a good introduction to Pompe disease. It is available on the Pompe Community Web site at www.pompe.com.
- The Physicians Guide to Pompe Disease brochure can be ordered from the National Organization for Rare Disorders (NORD) at www.rarediseases.org/programs/pompe_brochure.html
- GSDNet: Join the GSDNet Listserv to exchange emails with people around the world living with Pompe disease. To subscribe to GSDNet by email:
"To" Line: Type the following onto the "To" Line: listserv@listserv.icors.org
"CC" Line: Leave the CC line blank
"Subject" Line: Leave the subject line blank.
Email Message: Type the following message into the body of the email:
Subscribe GSDNet <add your name>



Medical Centers Specializing in Neuromuscular Disorders:

To locate medical centers that specialize in treating neuromuscular disorders, contact the Neuromuscular disorders (NMD) association for your country.

- In the United States, contact the Muscular Dystrophy Association (MDA) at www.mdausa.org/clinics.
- In Europe, contact the European (EAMDA) at Web site www.eamda.net

GETTING THE RIGHT CARE FOR POMPE DISEASE

- In other continents, visit the World Alliance of Neuromuscular Disorder Associations (WANDA) at Web site www.wandaweb.org and click on Your Country.

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