



Adapting to Living with Pompe Disease

Everyone with Pompe disease must learn how to live with a serious illness that gets worse over time. Whether you are the parent of a child who needs full-time care, an adult with mild muscle weakness and breathing problems, or the partner of someone with Pompe disease, you will have to adapt to changes that can occur. This can be stressful and, at times, overwhelming. But taking steps to prepare for change can help you cope with the challenges you may face. The strategies suggest here will help you figure out what you need to do to live as well as you can.



Managing Pompe Disease

Once someone has received a conclusive diagnosis of Pompe disease, it's time to discuss the options for managing this complex disorder with a physician. While there is no cure for the disease, several types of care and treatment can help. Until recently, patient care focused entirely on symptom management, using a variety of supportive therapies to alleviate and compensate for the effects of the disease. Today, enzyme replacement therapy (ERT) is available which is intended to replace the deficient acid alpha-glucosidase (GAA) enzyme. Other potential treatments are in the research stage of development.

The muscle weakness caused by Pompe disease affects many parts of the body. It can result in difficulties with walking and moving around, breathing properly, even eating and performing basic day-to-day tasks.

Q: How will Pompe disease affect my daily life?

A: The more severe your symptoms, the more the disease may affect your daily life and your relationships with others. As muscles get weaker, it can get harder to stand, walk, bend, get up from a chair, climb stairs, drive a car, or move around. You may need to adjust your routine or find new ways to do things at home, school, or work. You may also need special equipment or help from other people to perform certain tasks. For most people diagnosed with Pompe disease and those people helping to care for them, the biggest challenge is trying to balance the increasing need for support with the desire to stay as independent as possible. One of the toughest decisions you may face is when to start using a wheelchair. When you reach a point where it takes all of your strength to walk a few steps, you may have little energy to keep up with social contacts and activities you enjoy. A wheelchair can help you to stay as active as you want to be.

Adapting to Living with Pompe Disease

Q: My child seems to be losing interest in doing a lot of activities he used to do. How can I help him stay active?

A: First try to figure out why your child is losing interest. If he lacks the strength to do a certain activity, see if it can be adapted to his needs. If he loves biking, for example, you might invest in a bicycle made for wheelchair users or people with lower body weakness. Or, suggest activities that any child with limited muscle strength might enjoy, such as swimming, computer games, cooking, gardening, or art projects. If your child seems lonely, visit an online support network (see *Where to Learn More* on the last page) to help find a person with Pompe disease who can serve as a role model for your child. You might also ask a young adult neighbor or a family friend to take him on outings once a week. Consider counseling with a mental health professional if these suggestions do not work or if he shows signs of becoming depressed. And do keep trying to help him be as independent as possible.

Q: What is Assistive Technology?

A: Assistive Technology is any device that helps a person with disabilities increase, improve or maintain the ability to perform a function. Assistive technologies can range from a simple spoon build-up to high tech computer devices. These devices foster independence, and can help people diagnosed with Pompe disease to live and fit better into the community.

Q: What equipment can help make life with Pompe disease easier?

A: Choosing the right equipment for your specific needs can make your life easier. It can also allow you to do more things for yourself. The chart below lists some basic activities of daily living and the kinds of equipment that may help you or your child to be more independent. Physiotherapists and occupational therapists can often give helpful advice. For other sources of help, see *Where to learn more* on the last page.

Q: Is there anything that can help make my life easier?

A: Yes, there is equipment that will help to make your life easier:

Bathing, Grooming and Toileting:

- Bath bench or shower chair
- Handheld shower head
- Lifter
- Raised toilet seat or adapted toilet
- Female toileting cushion
- An environmental control unit to turn a lamp on and off
- Bathtub grab bar

This equipment helps people with muscle weakness safely reach the sink, toilet, shower, and bathtub. It can also give users privacy and ease strain on their caregivers.

Sitting and Standing: This equipment eases pressure caused by sitting for long periods on a hard surface or in a wheelchair. It also helps people who have trouble getting out of a chair.

- Seat cushions
- Seating shells
- Stand-up chair
- Adapted seating

Adapting to Living with Pompe Disease

- Arm rests
- Adjustable tables

Sleeping:

- Hospital bed with foam mattress: Electric controls that move the bed up and down make it easier to get in and out of it. A foam mattress cushions the body to prevent bedsores.
- Foot splints

Moving Around:

- Canes
- Walkers
- wheelchairs
- Scooters
- Modified car

This equipment helps people get around by providing extra support for walking and easing pain from weak muscles. Electric wheelchairs, scooters, and cars allow users to be more independent.

Home/Work Site Modifications:

- Ramps
- Doorways to accommodate wheelchairs
- Wheelchair accessible kitchen and bathroom appliances
- Staircase lifts

Communicating and Being Independent: There are many devices and aids that enable communication skills for people who have speech difficulties or are non-verbal.

- A simple communication board with symbols and/or words that the person points to
- Text telephones
- Computers
- A speech output device
- Trackballs
- Intercom system
- Universal remote control

Principles for treatment of contractures (permanent shortening of a muscle or joint) and deformity in neuromuscular disorder are well established and should be followed for individuals with Pompe disease. Contractures and deformity should be prevented by counteracting deforming forces with the use of gentle forces over time including daily stretching, correction of positioning, use of splinting and orthotic intervention, provision of adequate support in all positions, especially sitting and supported standing as appropriate, and education of patients and families.

Adaptive equipment and orthotics can be essential in the control of contractures and deformity and can provide changes in position and pressure relief for maintenance of skin integrity in individuals who cannot shift their own weight or change positions independently. Orthotic intervention often includes the use of:

- Ankle foot orthoses (AFO's) to prevent plantar flexion (downward movement of the foot) contractures.

Adapting to Living with Pompe Disease

- Thigh binders to prevent iliotibial band contractures (iliotibial band is a thick band of connective tissue that runs along the outside of the thigh).
- Knee splints to prevent knee flexion contractures (knee won't fully straighten).
- Resting wrist/hand/finger splints combats mild to moderate hand and finger contractures.
- Seating systems in adapted strollers or wheelchairs are critical to prevent or minimize contractures and deformity, especially spinal deformity, and should include:
 - A solid seat and back
 - Hip guides
 - Lateral trunk supports
 - Knee adductors
 - Head support as needed
 - Custom made seating shells
- Supported standing is considered beneficial to prevent or minimize osteoporosis (loss of bone mass), and may include the use of:
 - Supine, prone, vertical and hydraulic standers
 - Power standing capabilities on motorized wheelchairs or other motorized standing devices.
 - Power tilt, recline and elevating leg rests on motorized wheelchairs can allow independent position change and weight shift for assistance in minimizing contracture and maintaining skin integrity.
- Orthopedic surgery may need to be considered in some cases (such as scoliosis management), in which progression occurs beyond that which is amenable to conservative management, although results of spinal surgery maybe suboptimal. Anesthesia precautions should be followed during orthopedic surgery.

These devices help people diagnosed with Pompe disease (and their caregivers) stay connected to the outside world. For example, they make it easier to turn on lights and equipment and to talk with someone who is at the front door from another room in the house.

Q: What else can I do to prepare for the future?

A: Planning for your changing needs will help you live as well as you can. For example, if you are caring for a young child with Pompe disease, you may need to set up a plan with the child's school to help him or her succeed there. If you are an adult with Pompe disease, you may need to ask your employer to make some changes in your job so that you can keep working. To learn your rights and benefits, reach out to government and social service agencies. As your physical abilities change, you may need to lean on other people for support. Accepting help — or asking for help when you need it — can make life much easier. Seeking the advice and support of other people diagnosed with Pompe disease can also help. They may be able to suggest creative ways to handle issues at home, school, or work. Keeping up with your interests and hobbies can also help you keep a positive attitude.

Adapting to Living with Pompe Disease

For more information on how to adapt your environment go to the IPA website www.worldpompe.org and click on the Patients Helping Patients section.

Where to Learn More

These groups can offer advice, information, and support to help you prepare for changing needs.

- 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
- Information on Adaptive Devices:
<http://www.pnwbooces.org/specialed/tam/adaptive.htm>
- 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.*
- Physical Therapy Management of Pompe Disease: May 2006. Vol 8. No.5
Laura E. Case, PT, DPT and Priya Kishnani, MD
- 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.
- Muscular Dystrophy Association: MDA's Publication - 101 Hints to "Help-with-Ease" for Patients with Neuromuscular Disease <http://www.mda.org/publications/101hints/>
- Abledata (www.abledata.com) is an excellent source of information about adaptive equipment and devices from around the world.

This publication is designed to provide general information in regard to the subject matter covered. It is distributed as a public service by the International Pompe Association, with the understanding that the International Pompe Association is not engaged in rendering medical or other professional services. Medicine is a constantly changing science. Human error and changes in practice make it impossible to certify the precise accuracy of such complex materials. Confirmation of this information from other sources, especially one's physician, is required.