



ADAPTING TO LIVING WITH POMPE DISEASE

Everyone with Pompe disease must learn how to live with a serious illness that can get worse over time—especially without treatment. 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 suggested here will help you figure out what you need to do to live as well as you can. Please keep in mind that Pompe disease affects patients differently as is discussed in the Pompe Connections Signs and Symptoms Brochure”. Therefore, the information below is meant as guidance only and does not mean that all patients will have same experiences.



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 as of 2022, several types of care and treatment can help slow and manage the disease. Until 2006, 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 clinical trials and 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: Pompe affects patients differently. However, 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

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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 walker or 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.

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 more comfortably and stay more active in their 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 tables 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 as well.

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

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

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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
Arm rests
Adjustable tables
Adapted seating

Sleeping:

<u>Hospital bed with Foam or Hybrid mattress:</u> Electric controls that move the bed up and down make it easier to get in and out of it. A Foam or Hybrid mattress cushions the body to prevent bedsores.
Foot splints

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

Canes
Wheelchairs and Scooters
Walkers
Modified car

Home/Work Modifications:

Ramps
Widen doorways to accommodate wheelchairs
Wheelchair accessible kitchen and bathroom appliances
Staircase Lifts and Lifts

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

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.

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

Q: I am suffering from contractures. Is there anything I can do?

A: Yes, there are a lot of approaches you can take to improve the contractures, and to lessen the pain that they cause.

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.
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.

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Seating systems in adapted strollers or wheelchairs are critical to prevent or minimize contractures and deformity, especially spinal deformity, and may 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 may be suboptimal. Anesthesia precautions should be followed during orthopedic surgery.

Q: My child seems to be losing interest in doing a lot of activities they once did . How can I help them stay active?

A: First try to figure out why your child is losing interest. If they lack the strength to do a certain activity, see if it can be adapted to his needs. If they love 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 board games, book clubs, computer games, cooking, gardening, or art projects. If your child seems lonely, visit an online support network 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. Do keep trying to help them be as independent as possible.

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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. It is important to focus on what you are able to do, instead of abilities you have lost or may lose in the future.

For more information on how to adapt your environment go to the IPA website www.worldpompe.org.

Where to Learn More Please see the More Information section

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