



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 suggested here will help you figure out what you will need to do to live as well as you can.



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 with Pompe disease and those 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 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 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.

Other names for Pompe disease

Acid alpha-glucosidase deficiency, acid maltase deficiency (AMD), glycogen storage disorder (GSD) type II, glycogenosis II, and lysosomal alpha-glucosidase deficiency. In different parts of the world, Pompe may be pronounced "pom-PAY," "POM-puh," or "pom-PEE."

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.

Equipment that may help make life easier

Bathing, Grooming, and Toileting

- Bath bench or shower chair
- Handheld shower
- Lifter
- Raised toilet seat or adapted toilet
- Female toileting cushion

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

- Cushions
- Seating shells
- Stand-up chair

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.

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.

Moving around

- Foldable cane
- Foot splints
- Wheelchair
- Scooter
- Adapted 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.

Communicating and being independent

- Computer
- Intercom system
- Universal remote control

These devices help people 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.

Equipment for breathing support is described in the *Breathing problems* handout. Depending on where you live, you may be able to get some help to pay for equipment from government sources, health insurance providers, social service agencies, or centers for independent living. Limited financial support may also be offered by patient groups or special funds for people with serious illness or disabilities.

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 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.

Where to learn more

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

- The **International Pompe Association (IPA)** Web site (www.worldpompe.org) has a list of worldwide sources of financial support for children in need. The Web site also lists contacts for IPA groups around the world. The contact for your country may be able to direct you to other funding sources or help you find a Pompe role model for your child
- You may also be able to find a Pompe role model through an online support community, such as GSDnet. This electronic mailing list can be accessed through the **Association for Glycogen Storage Disease (AGSD)** in the United States or through the **Association for Glycogen Storage Disease of the UK**. To subscribe, visit either site at www.agsdus.org or www.pompe.org.uk/agsdnet.html
- The AGSD's **Pompe's Group** in the United Kingdom runs a workshop on Pompe disease at the annual AGSD conference. The group also publishes the *Pompe's Bulletin*, a newsletter for patients, parents, and healthcare providers, as well as *Pompe's Update*, which profiles children and adults who are adapting to living with Pompe disease. To learn more, visit www.pompe.org.uk
- **Abledata** (www.abledata.com) is an excellent source of information about adaptive equipment and devices from around the world

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