



## Exercise and physical therapy

**O**ften the first sign of Pompe disease is weakness in the muscles used to move around. Babies may seem “floppy” or may not be able to sit up without support. Toddlers may not start to walk when other children do or may walk with a waddle or limp. Children and adults may have trouble walking, as well as rising from a chair, climbing stairs, or keeping their balance. Exercise and physical therapy can help keep muscles as strong as possible. These supportive therapies can also ease muscle aches and stiffness and help people learn new ways to perform tasks that are hard for them to do. This handout explains how Pompe disease affects the muscles and the types of exercise and physical therapy that may offer the most benefit.



**Q**

### How can Pompe disease affect muscles and the ability to walk?

**A**

The genetic defect that causes Pompe disease prevents the body from breaking down glycogen, a form of sugar that is stored in muscle cells. As a result, too much glycogen builds up in the cells. This weakens muscles throughout the body that support balance and mobility, or freedom of movement. Some parts of the body may be more affected than others. In infantile-onset Pompe disease, there is profound muscle weakness in the legs and hips, as well as in the trunk (the area between the neck and the pelvis to which the arms and legs are attached) and the muscles used for breathing. These babies

rarely have the strength to sit up without help. In late-onset Pompe disease, muscle weakness is seen in the lower part of the body (legs, hips, pelvis, and spine), the upper part of the body (neck, shoulders, and upper arms) and the muscles used for breathing. This is why children and adults with late-onset Pompe disease have trouble walking or walk with a waddle or limp. Increasing muscle weakness may lead to scoliosis (curvature of the spine), contractures (muscle tightness), lower back pain, muscle pain, fatigue, and breathing problems that further limit mobility. While many people with Pompe disease may at some point need to use a wheelchair, exercise and physical therapy can help preserve mobility for as long 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 are contractures?

A As muscles get weaker and are not used, they can become tight and stiff. Sometimes they get so tight that they cannot move. This may cause the muscles (or the joints and tendons that connect the bones) to freeze in one position, forming a **contracture**. If contractures develop in the parts of the body that support your weight, such as your legs, ankles, or knees, it can be hard to stand up straight, walk, or keep your balance. The best way to prevent contractures is to keep muscles stretched and moving.

### Q How can daily exercise help people with Pompe disease?

A Exercise has many benefits for people with Pompe disease. It can ease muscle aches and pains, lessen stiffness, and increase flexibility and mobility. It can also help you stay active longer, have more energy, and improve your physical and mental health. But doing the wrong kind of exercise or pushing yourself too hard may damage muscles and make you feel more tired. Be sure to work with your healthcare team to plan an exercise program geared to your abilities. For most people with Pompe disease, that means giving your muscles a workout but stopping before you reach the limit of what you can do. Some simple tests, called **exercise tolerance tests**, can help you figure out how much exercise you can do without straining your heart or your muscles. These tests measure your heart rate and how much oxygen you take in while on a treadmill or stationary bicycle. Your fitness plan should be supervised by your doctor or physical therapist and may need to be adjusted as muscle weakness increases.

### Q What type of exercise is best for people with Pompe disease?

A Exercise must be tailored to each person's needs and abilities. Many healthcare providers and people with Pompe disease have found that certain kinds of exercises offer better results than others and are less likely to cause muscle damage. These include:

- **Moderate aerobic exercises**, such as walking, swimming, biking, or using fitness machines such as a treadmill, stationary bicycle, or movement trainer for arms and legs
- **Gentle strengthening exercises**, such as Pilates — guided floor mat exercises that help improve muscle tone, flexibility, balance, and posture by gently stretching muscles against the weight of the body
- **Flexibility training**, such as exercises done in a swimming pool that help stretch stiff muscles and increase range of motion by using the water for support and resistance. A word of caution to ventilator users: Check with your doctor before trying this type of exercise because water pressure can make it more difficult to breathe

You may need to adjust your exercise routine as the disease advances and muscles get weaker or if you develop breathing problems, scoliosis, or contractures. Experts advise people with Pompe disease to avoid lifting heavy weights or doing intense aerobic exercise that can damage muscles by putting too much stress on them. To be sure you are exercising safely and getting the most benefit from your fitness plan, always follow the instructions of your healthcare team.

Q

### What else can I do to help keep my muscles strong?

A

Some healthcare providers feel that combining daily exercise with a diet that is high in protein and low in carbohydrates can help keep muscles strong for as long as possible. This combination may help replace some of the muscle protein that is lost when the muscles are damaged. A number of people with Pompe disease have even regained some of the function they had lost by following this type of program. But more studies are needed to see how much it would help people at different stages of the disease. The experts agree, however, that any type of diet and exercise program must be carefully supervised and adapted to each person's abilities and dietary needs.

Q

### How can physical therapy help someone with Pompe disease?

A

Physical therapy can help maintain flexibility and mobility, ease stiffness in joints, and prevent contractures. The physical therapist uses exercises, machines, and assistive devices (like wheelchairs, scooters, or night splints) to help people with Pompe disease learn new ways to move around and manage daily tasks. Treatment should be targeted to what each patient needs.

#### Different ways physical therapy can help

- Physical therapy can help a “floppy” baby develop better muscle tone and improve the infant's ability to breathe and swallow
- Range-of-motion and stretching exercises can strengthen joints and muscles to prevent contractures and help children and adults adapt their activities to their changing abilities
- Young children whose muscles are still growing may be helped by doing stretching exercises and wearing night splints (also called **ankle-foot orthoses**, or **AFOs**) — molded foot supports that keep the heel stretched out while the child sleeps
- The use of lightweight cuffs or sleeves that apply firm support to the thigh (also called **thigh binders**) can prevent muscle tightness that causes pain in the hips, knees, or buttocks

#### Learning to use assistive devices

This is a big part of the physical therapist's job. Teaching someone how to use a cane or walker to prevent falls might delay the need for a wheelchair. When a child or adult can no longer walk, learning how to get around with a scooter or wheelchair can greatly improve his or her quality of life. The important thing is to start physical therapy, as early as possible and to figure out in advance what kind of assistive devices or equipment you may need. For advice on how to find a physical therapist who understands the needs of people with neuromuscular disorders like Pompe disease, see *Where to learn more* on the next page.

## Where to learn more

Contact these groups for more advice and information about exercise and physical therapy for people with Pompe disease:

- The **International Pompe Association (IPA)** can direct you to Pompe disease patient groups around the world. To find the contact for your country, visit the IPA Web site at [www.worldpompe.org](http://www.worldpompe.org). The Web site also has links to current research articles on Pompe disease
- **Independent Living Centers (ILCs)** throughout the world offer information, skills training, and support services to people who are living with disabilities. For a worldwide directory of ILCs, visit <http://www.ilusa.com/links/ilcenters.htm>
- Your healthcare provider may be able to help you find a physical therapist who specializes in treating people with neuromuscular disorders. To locate a provider, search the online directory of the **World Confederation for Physical Therapy** at <http://www.wcpt.org/membership/index.php>

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