



Exercise and Physical Therapy

Often 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 arms and the trunk (the area between the neck and the pelvis to which the arms and legs are attached) and the muscles used for breathing (diaphragm, intercostals, abdominal, and accessory muscles). These babies rarely have the strength to sit up without help. In late-onset Pompe disease, muscle weakness is seen in the proximal muscles (nearest to the trunk) of the lower part of the body (legs, hips, pelvis, and spine), and of 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, a sideways (lateral) curving of the spine, contractures (an abnormal and usually permanent shortening of a muscle or tendon), 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

Exercise and Physical Therapy

wheelchair, exercise and physical therapy can help preserve mobility for as long as possible.

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 get stuck in one position, forming a **contracture**. If contractures develop in the parts of the body that support your weight, such as your legs, ankles, knees and /or hips, 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 Contractures and Deformity Be Prevented?

A: The principles for treatment of contractures and deformity in neuromuscular disorders are well established and should be followed for individuals with Pompe disease. Contracture 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
- Education of patients and families

Adaptive equipment and orthotics can be essential in the control of contracture 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 and splinting 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.
- Seating systems in adapted strollers or wheelchairs are critical to prevent or minimize contracture and deformity, especially spinal deformity, and should include:
 - A solid seat and back
 - Hip guides

Exercise and Physical Therapy

- Lateral trunk supports
- Knee adductors
- Head support as needed
- Custom made seating shells

Supported standing is considered beneficial and may include the use of:

- Supine, prone, vertical and hydraulic standers
- The use of 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 managing scoliosis. Anesthesia precautions should be followed during orthopedic surgery.

Q: Are people with Pompe disease at risk for developing Osteopenia?

A: Emerging reports of osteopenia, osteoporosis, and fracture, indicate that all affected children, wheelchair-bound and ventilator-dependent adults and patients with decreasing muscle strength should be evaluated for osteopenia.

Osteopenia is generally considered the first step along the road to osteoporosis. Both osteopenia and osteoporosis are varying degrees of bone loss, as measured by *bone mineral density*, a marker for how strong a bone is and the risk that it might break. The main way to determine bone density is to have a painless, noninvasive test called Dual-Energy X-Ray Absorptiometry (DEXA) that measures the mineral content of bone. Nutrition needs to be adequate especially in terms of intake of calcium and Vitamin D. In Pompe disease patients bone mass is strongly related to muscle strength. Thus if muscle strength decreases bone mass is expected to decrease as well. Potential interventions should focus on increasing muscle strength and might comprise exercise training programs or physical therapy programs. In more severely affected patients, the provision of weight bearing in physical therapy and in standing devices might be an option.

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

Exercise and Physical Therapy

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. 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.

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

A: In the past the general assumption was that people with a neuromuscular disease should perform gentle, submaximal (less than maximal effort), moderate exercises and avoid heavy resistance exercise. Concerns were raised that exercising too much could damage the muscles and accelerate deterioration. Recently, however, studies on the effects of exercise in patients with muscular dystrophies have shown improvements in physical fitness level. It appeared that exercise was well tolerated and safe. Preliminary data of an exercise tolerance study in patients receiving enzyme replacement therapy support that regular exercise is well tolerated and could improve muscle strength and endurance. Further studies are in progress. Exercise programs should be tailor made for the individual patient and take into consideration the specific needs, abilities, and disabilities of the participant.

- 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.
- Studies on exercise training in patients with Pompe disease are in progress. The studies will focus on a combination of endurance, strength and core stability training or endurance training alone.¹

Q: Why is physical therapy in Pompe patients important?

A: Physical exercises are important for several reasons. They may help to:

- Improve and preserve aerobic capacity of the muscles, which may make your muscles less easily fatigued
- Preserve muscle function (strength, coordination, and endurance)
- To prevent or reduce secondary problems such as contractures, overweight, pain and fatigue
- Enhance self-esteem, provide motivation, and protect individuals against stress.
- To stop secondary deconditioning. Secondary deconditioning is the situation people are getting into as they choose or were advised to adopt a lifestyle with no or irregular physical activity. This lifestyle reduces the circulatory and ventilator capacity of the body and decreases the aerobic capacity and the

¹ Center for Lysosomal and Metabolic Diseases Erasmus MC University Medical Center

Exercise and Physical Therapy

strength of the muscles.

Q: What is the difference between aerobic and anerobic exercises?

A: Aerobic exercise is physical exercise that intends to improve the oxidative capacity. Aerobic means “with oxygen”, and refers to the use of oxygen in the metabolic or energy-generating process of the human body. Examples of aerobic exercises are running, swimming and cycling. The opposite is anaerobic exercises: strength training and short-distance running are examples. The differences between the two types of exercises are: the duration and intensity of muscular contractions involved, and how energy is generated within the muscle.

During aerobic exercise, glycogen is catabolized with the help of oxygen to produce energy. Later on, when glycogen is absent, fat metabolism is initiated instead. Fat metabolism is a slow process, and is accompanied by a decline in performance. At this stage, the athlete will become fatigued. During anaerobic exercise glycogen is catabolized without the help of oxygen, which is a far less efficient process. During anaerobic exercise one will be fatigued more easily.

Benefits of regular aerobic exercise are:

- The muscles involved in respiration may become stronger and facilitate the flow of air in and out the lungs
- Condition of the heart muscle may improve as a consequence. The heart pump works more efficiently the heart rate during rest is lower. This is called aerobic conditioning
- The muscles of the body become stronger
- Your blood may circulate more efficiently. As a result the blood pressure may decrease.
- The total number of red blood cells may increase, which may facilitate transport of oxygen
- Improved mental health
- The risk for diabetes is reduced

Aerobic exercise may improve cardiovascular fitness. In addition, high-impact aerobic activities may stimulate bone growth, and reduce the risk of osteoporosis.

The major potential benefit from anaerobic exercise is that you built more muscle. Benefits of regular anaerobic exercise may be:

- Speeding up metabolism, even in rest. Muscles burn more calories per unit volume than any other tissue in the body.
- Strengthening the bones and reducing the risk of osteoporosis.
- Ease stiffness in the joints and protect them against potential injuries.
- Reducing the risk for diabetes.

Exercise and Physical Therapy

Q: When do I know that I trained too much?

A: One should always ask advice first. The scheme should be performed under the supervision of a physician or skilled physical therapist. It is important to monitor plasma CK-levels. In case of red urine, which may be hemoglobinuria or myoglobinuria training should be stopped immediately. The following guideline could be used: you did too much if the following occurs:

- Muscle pain and/or cramps after 48 hours after training
- Tachycardia (a very fast heart rate)
- Dizziness
- Red urine which means hemoglobinuria (the presence of free hemoglobin in the urine) and myoglobinuria (the presence of myoglobin in the urine, usually association with muscle destruction)¹

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

A: Some healthcare professionals feel that combining daily exercise with a diet high in protein and low in carbohydrates may help keep the muscles strong. 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 indicated that they regained some of the function they had lost by following this type of program. 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. Adequate calorie intake is mandatory for all patients. Too much calories should be avoided.

Q: How can physical therapy help someone with Pompe disease?

A: Physical therapy can help maintain flexibility and mobility, ease stiffness in joints, prevent contractures, and monitor exercise regimes. The physical therapist uses exercises, machines, and assistive devices (mentioned above) to help people with Pompe disease participate in gentle and beneficial strengthening and 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 is designed to:

- Optimize and preserve motor and physiological function as much as possible within the limits of the disease.
- Minimize the clinical impact of the disease process.
- Prevent or minimize secondary complications.
- Promote and maintain the maximum level of:
 - Function
 - Functional independence
 - Participation
- Optimize quality of life.

Exercise and Physical Therapy

- Maximize the benefits of enzyme replacement therapy or other treatments as they become available.

Q: How can physical therapy help someone with Pompe disease?

A: Physical therapy can help maintain flexibility and mobility, ease stiffness in joints, prevent contractures. Physical therapists help to monitor exercise training programs which may improve muscle strength and –function, endurance, balance, respiratory function and maintain the level of ambulation. The physical therapist uses exercises, machines, and assistive devices (mentioned above) to help people with Pompe disease participate in gentle and beneficial strengthening and learn new ways to move around and manage daily tasks. Treatment should be targeted to what each patient needs.

A part of the physical therapist’s job is to teach someone how to use assistive devices. Teaching someone how to use a cane or walker in order to prevent falls might delay the need for a wheelchair. When a child or adult finds it is getting progressively more difficult to walk, learning how to get around with a scooter or wheelchair can greatly improve his or her quality of life. 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.

Q: Are there recommendations for musculoskeletal / functional rehabilitation?

A: The following are recommendations for musculoskeletal / functional rehabilitation:

- Monitor cardio-respiratory status and response to position and activity with pulse oximetry during evaluation and treatment initially and with changes in status or activity.
- Screen for osteopenia/osteoporosis with DEXA and follow-up as needed.
- Assess musculoskeletal impairments, functional deficits, levels of disability, and societal participation at regular intervals and as needed, including radiographs as needed for monitoring of scoliosis, hip stability, and long bone integrity.

Enhance Muscle Function:

- Increase biomechanical advantage for movement:
 - Provide practice, movement, and gentle strengthening within limits of physiological stability.

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. The Web site also has links to current

Exercise and Physical Therapy

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>
- **World Confederation for Physical Therapy** Your healthcare provider may be able to help you find a physical therapist who specializes in treating people with neuromuscular disorders. To locate a professional, search the online directory of at <http://www.wcpt.org/membership/index.php>
- **Physical Therapy Management of Pompe Disease** May 2006. Vol 8. No.5
Laura E. Case, PT, DPT and Priya Kishnani, MD.
- **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** www.pompe.com: offers the Pompe community comprehensive information on the disease, as well as resources and support to help manage the challenges it may bring.
- Center for Lysosomal and Metabolic Diseases Erasmus MC University Medical Center www.erasmusmc.nl/

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