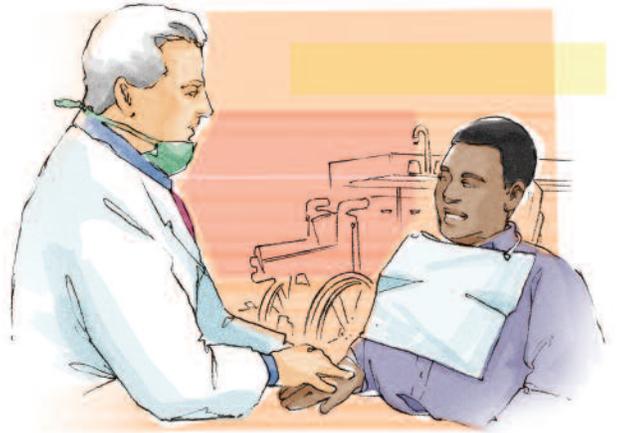




Common health concerns

Pompe disease affects everyone differently. This handout describes some of the medical issues you or your child may face and what to do about them.

To improve the quality of care for people with Pompe disease, researchers in the United States and Europe are creating treatment guidelines for healthcare providers to follow. These guidelines, called **standards of care**, will help make healthcare providers around the world more aware of how to manage the health concerns that may arise at each stage of Pompe disease.



Q How does Pompe disease affect the muscles? What can be done about it?

A Pompe disease weakens muscles throughout the body. Muscle weakness is most severe in the infantile-onset form of the disease. The heart muscle thickens and gets weaker. This can cause breathing problems and infections that may rapidly lead to heart failure and respiratory failure. Muscle weakness also causes a loss of muscle tone that makes infants look “floppy.”

In late-onset Pompe disease, weakness in the legs, arms, and muscles used for breathing can make it hard to move around or perform everyday tasks. Babies and toddlers may not roll over, sit up, crawl, stand, or walk when other children their age do. They may also lose some of the developmental gains they have made. Many children and adults with Pompe disease have trouble walking, keeping their balance, sitting comfortably, or standing up straight because the disease also weakens muscles in the back and pelvis.

Weakness of the muscles that run along the spine can lead to **contractures** (muscle tightness) or **scoliosis**

(a curvature of the spine that develops in growing children). Wearing a plastic brace may keep scoliosis from getting worse. Surgery should be considered if the curve becomes so severe that it interferes with breathing. In all cases, treatment must be tailored to the patient’s needs.

People who are more severely affected may have muscle weakness in the face, neck, throat, chest, and abdomen, making it hard to breathe, eat, swallow, and digest food. Loss of muscle strength in the abdomen may also cause the stomach to push forward and protrude, or stick out.

Supportive therapy can help manage the effects of increasing muscle weakness. This therapy may include dietary changes along with exercise and physical therapy to maintain strength and promote movement. It may also include assistive devices such as night splints that help prevent contractures. (For more information on these supportive therapies, see the handouts *Nutrition and dietary therapy*, *Breathing problems in Pompe disease*, and *Exercise and physical therapy*.)

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 kinds of breathing problems can occur with Pompe disease? And what can be done to make breathing easier?**

A Increasing weakness of the **diaphragm** (a flat muscle just below the lungs and heart) and other muscles used for breathing can make it hard to breathe deeply, especially when you are lying on your back. This may wake you up at night and make you more tired during the day. You may have morning headaches and difficulty concentrating. You may also have trouble coughing up mucus that collects in your lungs when you have a cold or respiratory infection. To help keep your lungs clear and prevent more serious problems like pneumonia and respiratory failure, follow these tips:

- Get regular checkups
- See your healthcare provider promptly if you have symptoms of infection such as runny or stuffy nose, fever, and earaches
- Make sure your healthcare provider is aware that any respiratory infection you have must be aggressively treated (with antibiotics, for example)
- Drink lots of fluids when you have a cold
- Have lung function tested periodically
- Get a flu shot every year and pneumonia vaccines

Respiratory therapy, the use of breathing exercises and devices such as ventilators, can help maintain breathing as muscles get weaker. (For more information on respiratory therapy, see the handout *Breathing problems in Pompe disease*.)

Q **What are the risks of having anesthetics during surgery? What can be done to lower the risk?**

A Anesthetics are drugs that are given to block pain during surgery. These drugs work by relaxing the heart muscle. They also reduce feeling in the nerves. Local anesthetics that numb a small area (for example, when you get stitches or have a tooth pulled) do not usually cause a problem for people with Pompe disease. But extra care must be taken with local anesthetics that numb a larger area (such as an epidural given to numb the spinal area) or with general anesthetics that put you to sleep during surgery. These drugs pose a risk for people with Pompe disease because they may have a greater effect on the heart muscle and the muscles used for breathing (which are already weakened in Pompe disease). Scoliosis (curvature of the spine) can also affect the way anesthesia works.

Meeting with the anesthesiologist (the healthcare provider who gives the anesthetics) before having surgery can help prevent problems. Your healthcare provider may find it helpful to review the scientific literature on anesthesia management in Pompe disease. Wearing a medical alert bracelet can help you get proper treatment in an emergency.

Q **What are the challenges of getting dental care when you have Pompe disease? What can I do about them?**

A Muscle weakness in different parts of the body can make it hard to take care of your teeth and get proper care at the dentist's office. An enlarged tongue or weak throat muscles can make it hard to chew or swallow. This can wear down the teeth and increase the risk for tooth decay and gum disease. It may also be hard to keep your mouth open when you are brushing, flossing, or having dental work done.

These tasks can be even more difficult if you do not have much strength in your arms. If your breathing muscles are weak, you may have trouble breathing when you lay back in the dentist's chair. And if you are in a wheelchair or use a ventilator, it can sometimes be a challenge to find a dentist who is willing to treat you (for advice on finding a dentist, see *Where to learn more* on page 4).

Keep in mind that many dentists have never heard of Pompe disease. You may therefore have expert knowledge to share with your dentist.

These steps can help you get the dental care you need:

- **Use an electric toothbrush, a mouth rinse that fights tooth decay, and special flossing aids to clean the teeth.** Or ask the dentist for toothbrushing tips and other oral care advice if help is needed with dental care or if you have a feeding tube
- **Try using a molded plastic or latex-free foam mouth prop to hold the mouth open.** Your dentist may be able to order it for you or suggest a supplier who carries it. The dentist can also show you how to use it
- **Make sure your dentist is aware of any special needs — and speak up if something causes pain or discomfort.** If you have trouble breathing when you lie on your back, tell the dentist you need to sit upright in the chair
- **Schedule several short appointments to complete your treatment if it is hard to sit in one position or keep your mouth open for long periods.** Explain that you may also need to take rest breaks during dental visits

Q

What can be done to relieve pain from muscle cramps or tension headaches?

A

Many people with Pompe disease complain of muscle cramps or aches. Pain in the head, neck, or shoulders may cause tension headaches. Unlike the morning headaches that are caused by nighttime breathing problems, tension headaches occur at different times of the day or night. Muscle weakness can put extra stress on your joints and ligaments, the tissue that connects the bones. As a result, pain in the lower back, hands, arms, legs, or feet that is not brought on by exercise, activity, or injury may occur. Colds, fever, infections, and emotional strain may also cause headaches or muscle pain. Pain medicines may bring some relief. Proper posture is very important, too. If you use a wheelchair, have the wheelchair adapted to your specific needs and always try to sit in a comfortable position.

You may also be helped by remedies that have worked for other people with neuromuscular disease. These include getting more rest or exercise, taking hot showers, and using natural healing methods such as massage, meditation, or acupuncture. Be sure to report pain to your healthcare provider and ask what else you can do to ease your discomfort.

Where to learn more

For more advice and information about the medical concerns discussed in this handout, these sources may be helpful:

- 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. Check the “Latest News” portion of the Web site for updates on the development of standards of care for Pompe disease
- To find a dentist who can meet the special needs of people with Pompe disease, contact large hospitals or rehabilitation centers, dental schools, or dental societies. The **International Association for Disability and Oral Health** maintains a list of members throughout the world who treat people with special needs. To view the list, visit the iADH Web site at www.iadh.org and click on “iADH Information”

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