Common health concerns

Pompe disease effects everyone differently. This brochure describes some of the Pompe Disease 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 have creating guidelines for health care providers to follow. These guidelines called standards of care, help make health care providers around the world more aware of how health concerns that may arise at each stage of Pompe disease. The health care standards will also help care providers separate the day today health issues that everyone experiences from the Pompe issues.

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 it 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, 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 back brace may keep scoliosis from getting worse. Surgery may also be considered if the curve becomes so severe that it interferes with breathing. In all cases treatment must be tailored to the patient needs. People who are more severely affected may have muscle weakness in the face, neck, throat, chest and abdomen making it hard to breath, eat, swallow, and digest food. Loss of muscle in the abdomen may cause the stomach to push forward and protrude or stick out. **Supportive therapy** can help to manage the effects

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

Q: What kinds of breathing problems can occur with Pompe disease? 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 laying 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 music 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 fellow failure, follow these tips

- Get regular check-ups
- See your healthcare provider promptly if you have symptoms of infection such as runny or stuffy nose, fever and earaches.
- Make sure your health care provider is aware that any respiratory infection 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, Pneumonia vaccination and Covid vaccinations.

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

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

A: Anaesthetics 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 anaesthetics that numb a small area (for example, when you get stitches or have a tooth extracted) do not usually cause a problem for people with Pompe disease. But extra care must be taken with local anaesthetics that numb a larger area (such as an epidural given to numb the spinal area) or with general anaesthetics 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 anaesthesia works.

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Meeting with the anaesthesiologist (the healthcare provider who gives the anaesthetics) before having surgery can help prevent problems. Your healthcare provider may find it helpful to review the scientific literature on anaesthesia management in Pompe disease. Wearing a medical alert bracelet or carrying a medical alert in your purse or wallet 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. Keep in mind that many dentists have never heard of Pompe disease. You may therefore have expert knowledge to share with your dentist.

These tips may be able to 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 moulded plastic or latex-free foam mouth prop to hold the mouth open.** Your dentist may have them or be able to order it for you or suggest a supplier who carries it.
  The dentist may 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

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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 Night-time 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.

Osteopenia and Osteoporosis

Q Why should patients with Pompe disease be screened for this?

A: Osteopenia is the thinning of bone mass. A decrease in bone mass is considered a serious risk factor for the development of osteoporosis. The diagnostic difference between osteopenia and osteoporosis is the measure of bone mineral density.

Osteoporosis, the "fragile bone" disease is characterized by a loss of bone mass caused by a deficiency in calcium, vitamin D, magnesium, and other vitamins and minerals. Many of the foods eaten contain these bone-building minerals.

Femur fracture and thoracic vertebral fractures have been identified in patients with infantile onset Pompe disease. Osteopenia has been seen in patients with Pompe disease as young as 4 months of age, which could be attributed to chronic immobilization and weakness, but osteopenia has also been identified in patients with Pompe disease with good motor strength and nutrition and needs to be further studied. The pathophysiologic mechanisms of osteopenia/osteoporosis in Pompe disease are not yet well understood so are managed generically.

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Factors that have the potential to contribute to osteopenia and osteoporosis have therapeutic implications. Nutrition needs to be adequate especially in terms of intake of calcium and Vitamin D and attention given to medications (e.g., long term use of certain diuretics which could cause hypercalciuria, long term steroid use). Potential interventions may include the provision of weight bearing in physical therapy and in standing devices. There is insufficient evidence to suggest pharmacologic therapy such as bisphosphonates as a preventive treatment in Pompe disease at the current time.

Emerging reports of osteopenia, osteoporosis, and fracture, suggest that screening patients with Pompe disease for osteopenia is indicated.

Bone mineral density (BMD) is the measurement of calcium levels in bones, which can estimate the risk of bone fractures. It is also used to determine if a patient has osteopenia or osteoporosis. Bone mineral density tests are non-invasive and painless procedures usually done on the hip, spine, wrist, finger, shin bone, or heel.
While osteopenia can be diagnosed using plain radiographs, the most common method for measuring BMD (and a way to definitively diagnose osteoporosis) is through Dual Energy X-ray Absorptiometry or DEXA. This scan uses low-energy x-rays that expose patients to much less radiation than standard x-rays and can assess calcium levels in bone. The results are measured as a "score" and are compared to those of healthy individuals.

What do the numbers mean? A patient's BMD is given a T-score, which is obtained by comparing it to an average score for a healthy 30-year-old of the same sex and race. The difference between the "normal young" score and the patient's score is referred to as a standard deviation (SD). T-scores can fall as low as -1 SD and still be considered healthy. Patients with T-scores between -1 SD and -2.5 SD are diagnosed with osteopenia and are considered at high risk for osteoporosis. Patients with T-scores lower than -2.5 SD are diagnosed with osteoporosis. For these patients, treatment may be necessary and may include the use of medications to help increase bone mass, as well as lifestyle changes such as diet and exercise