



## Breathing problems in Pompe disease

**M**any infants, children, and adults with Pompe disease have early signs of breathing problems that can become more serious as muscles get weaker. These problems may develop slowly or come on suddenly, depending on the stage or the severity of your disease. This handout describes the most common breathing problems and how they can be managed. It also explains what steps must be taken to maintain or restore lung function in an emergency or if you ever need surgery.



**Q**

### Why do people with Pompe disease develop breathing problems?

**A**

Pompe disease weakens the diaphragm (a sheet of muscle just below the lungs that separates the chest from the abdomen), as well as muscles in the rib cage. These muscles play a major role in breathing. When they get weaker, it can be hard to breathe deeply enough to fill your lungs with air and expel carbon dioxide (CO<sub>2</sub>). This causes CO<sub>2</sub> to build up in your blood, making you feel poorly and affecting your overall health. The medical term for this condition is **respiratory insufficiency**. Increasing

weakness of the breathing muscles also makes it hard to cough. If your cough is not strong enough to clear your airways of mucus, it can lead to lung infections and other health problems.

Breathing problems in Pompe disease can start while a person is still able to walk. In other neuromuscular disorders, breathing problems only appear when people can no longer walk. If your symptoms are not diagnosed until you are already in a wheelchair, it may prevent you from getting prompt treatment. That is why you must be alert to your symptoms and know what to do if they occur.

### 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 the signs of a breathing problem in a person with Pompe disease?

**A**

There are many signs and symptoms of respiratory insufficiency, the breathing problem caused by reduced air flow into the lungs. These include:

#### Impact on daily activities

- Having a hard time concentrating or staying alert
- Marked decline in performance (for example, a drop in grades at school or needing more time to complete tasks at work)
- Feeling short of breath when at rest or after physical effort or exercise
- Loss of appetite or desire to eat

#### Sleep problems

- Being very tired or sleepy during the day
- Having trouble falling asleep or staying asleep
- Frequent nightmares

#### Other problems

- Anxiety
- Depression
- Morning headaches
- Weak sneezing and coughing
- Using muscles in the neck and spine to help with breathing

Having one or more of these symptoms does not always mean you have a breathing problem. But becoming aware of symptoms that make it hard to function during the day or sleep soundly at night can help you take steps to manage them.

**Q**

### What health problems can occur if you have trouble breathing deeply?

**A**

Taking deep breaths helps keep lungs inflated. This powerful process enables the lungs to hold all the air needed for normal lung function. Over time, breathing too weakly reduces the amount of air the lungs hold, or the lung volume.

#### Hypoventilation during sleep

People with Pompe disease who have severely reduced lung volume (below 40% of normal) are likely to develop hypoventilation during sleep. This means you are not breathing deeply enough to maintain normal airflow into and out of the lungs. Some individuals with weakness of the throat muscles and the tongue may also develop obstructive sleep apnea, which occurs when the airway is blocked. This causes breathing to stop for short periods while sleeping. Hypoventilation and sleep apnea should be diagnosed and treated in a sleep lab with special experience in neuromuscular disorders.

#### Risk of infection

When you have a cold or bronchitis, a weak cough increases your risk for developing pneumonia (a lung infection).

#### Respiratory insufficiency

If you cannot breathe deeply enough to maintain normal amounts of oxygen in the blood and expel CO<sub>2</sub> from the lungs, you may develop respiratory insufficiency. If not treated properly, this can lead to respiratory failure, which means you are too weak to breathe on your own. Respiratory failure is a medical emergency. To restore breathing, a tube is inserted into the windpipe.

Q

### What can I do to prevent these health problems?

A

You can help prevent serious health problems from occurring by taking these steps:

- **Get regular testing to check lung function** when you are sitting up and lying down (testing in both positions helps detect weakness of the diaphragm)
- **Watch for other signs and symptoms of respiratory failure**, such as shortness of breath, rapid breathing, headaches, sleepiness, confusion, sweating, racing pulse, or cyanosis (when lips, skin, and nails turn a bluish color)
- **Seek prompt treatment to improve breathing**
- **Learn how to do assisted coughing**, and use a cough-assist machine if you have a weak cough
- **Get regular checkups, a flu shot every year, and a pneumonia vaccine** whenever you are due for one to reduce the risk for lung infections
- **Call your healthcare provider when you feel a lung infection may be starting** so that you can begin taking antibiotic medicine as soon as possible

Q

### What kinds of therapy may help people with breathing problems?

A

Mechanical ventilation helps people with Pompe disease manage breathing problems by sending air to the lungs. The use of machines (or ventilators) can greatly improve the quality of life for children and adults with respiratory insufficiency. At first, therapy may only be needed at night to help maintain normal airflow while sleeping. A portable machine is used to supply a steady stream of air through a mask that fits over the nose or over both the nose and mouth. Two types of ventilators are most often used by people with Pompe disease:

- **BiPAP** or (Bi-pap) ventilators can be preset to supply greater air pressure when you breathe in and less when you breathe out
- **Volume-cycle ventilators** allow you to adjust the volume of air instead of the pressure so that you can take deeper breaths when you need them

Another type of device, called **CPAP** (or C-pap), supplies air at a constant pressure level but does not give people with weak breathing muscles the kind of respiratory support they really need. CPAP should only be used by people with Pompe disease in rare cases to treat sleep apnea.

Therapy can also help stretch out the lungs and chest muscles and make it easier to cough. Methods include:

- **Air stacking**, which uses a ventilation device to help you increase the amount of air you can hold in your lungs by taking a series of deep breaths
- **Assisted coughing**, done manually or by a cough-assist machine, which helps you breathe out with enough force to cough up any mucus in the airways

If a person has acute respiratory failure or develops a respiratory infection that is too hard to treat, it may be necessary to make an incision (or cut) in the skin and insert a breathing tube into the windpipe to maintain ventilation. This surgical procedure is called a **tracheostomy**. Most people with Pompe disease should be able to get the respiratory support they need by using a combination of noninvasive therapies (therapies that use masks or a mouthpiece and do not require surgery). Sometimes, a breathing tube that is inserted during an illness can later be removed. It is vital for people with breathing problems to be under the care of a pulmonologist or respiratory therapist so that respiratory support can be started in time to avoid emergency situations.

**Q I am scheduled to have a surgical procedure, and there is a chance that I will need general anesthesia. What steps should I take to help prevent breathing problems or lung infections?**

**A** If a surgery requires general anesthesia, you should alert the medical team *ahead of time* that you will need extra support to help you breathe and cough when you come out of surgery. Make it clear what option you want. Some people with Pompe disease prefer to have noninvasive ventilation (such as a BiPAP ventilator and cough-assist machine). Others feel that invasive ventilation (such as a breathing tube) is more effective. Family members who will be helping you through the recovery period will need to learn how to use this equipment. A respiratory therapist can provide the necessary training. You will also want to inform the surgical team if you are already using a ventilator so that a plan can be worked out in advance to handle your anesthesia needs as smoothly as possible.

**Q What do I need to tell healthcare providers about using oxygen?**

**A** Many healthcare providers are unaware that it is not advisable to give oxygen to people with Pompe disease who are having trouble breathing. In fact, it can be dangerous. Remember that breathing problems in Pompe disease are caused by weak muscles not by lung disease. In fact, most Pompe patients have healthy lungs. Instead of helping you breathe more deeply, the rush of oxygen shuts down your natural urge to breathe. The level of CO<sub>2</sub> in your blood goes way up and puts you at risk for respiratory failure. This can be avoided by

giving you ventilation therapy that helps restore deep breathing. To protect yourself in an emergency, you may want to wear a medical-alert bracelet or carry written instructions from your pulmonologist or respiratory specialist. Oxygen may only be used in the hospital when you also suffer from a lung disease like pneumonia or to provide comfort in the late stages of Pompe disease.

### Where to learn more

Contact these groups for more advice and information about respiratory 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 information on respiratory support while traveling
- **INFORESP** is an information service that seeks to raise awareness of breathing problems related to neuromuscular disorders. It is available at <http://www.inforesp.org>
- The **International Ventilator Users Network** helps people adjust to living with ventilators and network with other users and their families. It is available online at [www.post-polio.org/ivun](http://www.post-polio.org/ivun)
- You may also find useful information on ventilation management at the Web site of Dr. John Bach: [www.doctorbach.com](http://www.doctorbach.com). Dr. Bach is an expert on respiratory therapy for people with neuromuscular diseases and a strong advocate of noninvasive mechanical ventilation

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