THE INSIDE STORY

BREATHING AND POMPE DISEASE
The primary function of the respiratory system is to take oxygen into the body and remove carbon dioxide. This exchange of gases in the lungs is vital to ensure the body functions well.

**The diaphragm plays a major role in the respiratory system**
The diaphragm, a dome-shaped muscle located just below the lungs, helps control breathing.

When you breathe in, the diaphragm contracts and moves downwards, allowing the lungs to fill up with oxygen. When you breathe out, the diaphragm relaxes and moves upwards, pushing out carbon dioxide from your lungs.

**A weakened diaphragm**
Breathing problems in Pompe are generally caused by the weakening of the diaphragm and other respiratory muscles. A weakened diaphragm makes it hard to breathe in deeply, so you may not get enough air into your lungs. This leads to respiratory insufficiency, when the lungs cannot take in enough oxygen or expel enough carbon dioxide to meet the needs of the body.

Pompe can weaken the muscles involved in breathing, including the diaphragm, leading to respiratory insufficiency. Therapy and support are available to help manage breathing problems and prevent complications from developing.
Breathing difficulties can lead to a number of problems, which can affect your daily life and result in sleep disturbances.

**Low lung volumes**
When it is hard to breathe deeply enough to fill your lungs with sufficient oxygen, you may find yourself lacking energy and running out of breath easily.

**Impaired cough**
Weak respiratory muscles can make it difficult to cough strongly enough to clear mucus out of your lungs. Secretions that remain can increase the risk of infections such as bronchitis or pneumonia.

**Speech difficulties**
If you have difficulties breathing, it can affect your ability to speak properly as you may find yourself having to pause between sentences to breathe. If you find yourself having problems talking, contact your doctor who will be able to refer you to a Speech and Language Therapist (SaLT).

**Carbon dioxide retention**
When too little carbon dioxide is removed from the lungs, it can build up in the bloodstream. This can cause restless sleep, nightmares, morning headaches, daytime sleepiness and general feelings of tiredness.

**Sleep disturbances**
People with Pompe may experience short pauses in breathing during sleep, a condition called sleep apnoea. These pauses occur repeatedly throughout the night, resulting in fragmented sleep. You may not realise what is disturbing your sleep, so a sleep study (where you are monitored while sleeping) may be required to observe if you are suffering from sleep apnoea.

The main types of sleep apnoea are obstructive sleep apnoea (when the throat muscles relax and block the airway) and central sleep apnoea (when the brain fails to send proper signals to the muscles involved in breathing). Symptoms typically include loud snoring, followed by choking or gasping noises. People with Pompe may experience either form of sleep apnoea.

Becoming aware of the symptoms associated with breathing problems caused by Pompe can help you take steps to manage them. Speak to a member of your healthcare team if you experience any of the above problems.
Respiratory therapy
You may be referred to a physiotherapist, who will advise you as to appropriate exercises which may improve your muscle strength and fitness levels. A physiotherapist could also recommend and demonstrate specific techniques to improve how you clear your lungs to minimise the risk of infection.

Cough assist
If you have a weak cough, manual or mechanical (via a cough assist machine) assistance is available to help stimulate a cough that can clear your airways of any secretions. Your doctor will be able to discuss this in greater detail with you.

Preventing infections
Being unable to clear your lungs effectively can make you more susceptible to infections. This means that a cold can easily develop into something more serious like pneumonia or bronchitis. Promptly treating any infections that occur can reduce the risk of this happening. Keeping up-to-date with all the recommended vaccinations can also help you stay healthy.

Weak swallowing muscles can also increase the risk of choking, when foods or liquids are accidentally inhaled into the lungs. If you find yourself having problems eating, consider choosing foods that are easier to swallow, and contact your doctor who will be able to refer you to a Speech and Language Therapist (SaLT).

Ventilators
Ventilators can greatly improve your quality of life, by supplying enough oxygen to your lungs to support your day-to-day activities and help you get a good night’s sleep.

If you experience breathing problems, your doctor may recommend the use of a non-invasive BiPAP ventilator (Bi-level Positive Airway Pressure). This can be used during the night to help with sleep apnoea, and can also help during the day if required. Air is delivered from the ventilator via a mask fitted over the nose and/or mouth. Alternatively, a nasal pillow (small, flexible cushioned discs inserted into or around the nose) may offer greater comfort and feel less obtrusive than a face mask.

Getting the right therapy and support can help ease your symptoms, improve your quality of life and prevent serious complications from occurring. A number of options are available – your healthcare team will be able to advise you on the most appropriate care for your condition.
Regular monitoring of your condition is important and your doctor will normally assess your respiratory function at each visit.

When you first display breathing problems, your doctor will do an assessment of your condition, measuring exercise tolerance, strength of respiratory muscles, effectiveness of cough and respiratory rate. He or she will also ask you about other symptoms you may be experiencing, such as sleep disturbances, shortness of breath and headaches.

As your condition progresses, your doctor may use one or more of the following tests to assess your condition:

- **Forced Vital Capacity (FVC) test**: determines your lung function
- **Peak Cough Flow (PCF)**: measures strength of your cough
- **Oxygen saturation level**: measures the amount of oxygen in your blood
- **Detailed sleep study**: to observe any sleep disturbances
- **Chest X-ray**: to determine the presence of any infections
Association for Glycogen Storage Disease UK (AGSD-UK)
www.agsd.org.uk
Telephone: 01489 877991
Useful source for information on treatment, research news, national Pompe events, links to who to contact for help and other support resources (eg GSDNet – electronic mailing list for affected families).

International Pompe Association (IPA)
www.worldpompe.org
Leaflets covering a range of different topics relating to Pompe (including breathing problems) are available for download in over 10 languages.