

## CAUTION USING OXYGEN IN POMPE DISEASE



*The Acid Maltase Deficiency Association (AMDA) provides this information to help clinicians, emergency personnel, and caregivers understand the unique respiratory considerations in individuals living with Pompe disease.*

Pompe disease weakens the [diaphragm](#) and other muscles needed for breathing, which can make it difficult to remove carbon dioxide (CO<sub>2</sub>) from the body. As CO<sub>2</sub> builds up, oxygen levels may fall ([hypoxemia](#)) and respiratory function can worsen if oxygen is given improperly.

This document summarizes the key clinical rationale and evidence for safe oxygen administration in Pompe disease and other neuromuscular disorders.

Providing supplemental oxygen alone, without [assisted ventilation](#), can raise oxygen levels but reduce the brain's drive to breathe, allowing CO<sub>2</sub> to accumulate further. This may lead to [respiratory acidosis](#) and respiratory failure if not recognized and treated promptly.

Because of this, oxygen should never be given without ventilatory support (such as [BiPAP](#)) unless specifically directed by the patient's metabolic or pulmonary specialist. If oxygen is required, it must be used alongside ventilatory support and with CO<sub>2</sub> monitoring ([capnography](#) or [arterial blood gas](#)) to ensure adequate ventilation.

In short: "Treat [hypoventilation](#), not just hypoxemia." Assisted ventilation should be prioritized before adding oxygen in Pompe or other neuromuscular diseases.

## References

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### Glossary of Terms:

**Arterial blood gas (ABG)** – A blood test that measures oxygen and carbon dioxide levels and shows how well the lungs and respiratory muscles are working.

**BiPAP (Bilevel Positive Airway Pressure)** – A form of [noninvasive ventilation](#) that gives two levels of air pressure: a higher pressure when breathing in and a lower pressure when breathing out. It helps clear carbon dioxide and support weak breathing muscles.

**Capnography** – A monitor that measures the amount of carbon dioxide (CO<sub>2</sub>) you breathe out, used to check how well you are ventilating.

**Diaphragm** – The main muscle used for breathing, located under the lungs. Pompe disease can weaken this muscle, making breathing more difficult.

**Hypoventilation** – Breathing that is too shallow or slow to remove enough carbon dioxide from the body.

**Hypoxemia** – Low levels of oxygen in the blood. It can happen when breathing is ineffective or airways are blocked.

**Noninvasive ventilation (NIV)** – Any type of breathing support delivered through a mask instead of a breathing tube. BiPAP is a common form of NIV.

**Respiratory acidosis** – A condition where CO<sub>2</sub> builds up in the blood, making it too acidic. It can occur if oxygen is given without proper ventilation.

**Ventilation / Ventilatory support** – The movement of air in and out of the lungs. When muscles are weak, machines such as BiPAP help support this process.