



EMERGENCY MEDICAL INFORMATION SHEET

ABOUT POMPE DISEASE

- Pompe disease is a rare, inherited metabolic disorder caused by a deficiency of the enzyme acid alpha-glucosidase (GAA).
- This enzyme deficiency leads to glycogen buildup in muscle tissue, resulting in progressive muscle weakness over time.

CAUTION WHEN USING OXYGEN

- Always assess respiratory muscle strength, even when oxygen saturation appears normal.
- People with Pompe disease may retain carbon dioxide (CO₂) despite normal pulse oximetry readings.
- Do **NOT** administer oxygen without continuous CO₂ monitoring (end-tidal CO₂ or blood gas).
 - *Normal end-tidal CO₂: 30–45 mmHg*
 - *>45 mmHg: Indicates CO₂ retention and hypoventilation*
 - Use non-invasive ventilation (BiPAP) to support breathing, assist oxygen delivery, and help remove CO₂.
- If respiratory distress is present:
 - Begin BiPAP immediately.
 - Avoid unnecessary intubation.
- If intubation is required:
 - Avoid succinylcholine (suxamethonium); use alternative muscle relaxants.
 - After anesthesia, closely monitor for hypoventilation and delayed recovery.

For more information check out the fact sheets on: [Caution Using Oxygen](#), [Suxamethonium \(Succinylcholine\)](#), and [CPAP vs BiPAP](#).

RESPIRATORY AND ANESTHESIA PRECAUTIONS

- Pompe disease affects the breathing muscles, so use extreme caution with anesthesia, oxygen, and muscle relaxants.
- Avoid suxamethonium (succinylcholine): This medication is a muscle relaxant sometimes used during anesthesia to temporarily paralyze muscles for intubation or surgery.
 - In patients with Pompe disease, it can cause serious complications such as hyperkalemia and rhabdomyolysis.
- In any emergency, contact the patient's metabolic or neuromuscular specialist immediately.

For more information check out the fact sheets on: [Caution Using Oxygen](#), [Anesthesia Precautions](#), and [Anesthesia Medical Safety List](#)

AVOID SEDATING MEDICATIONS:

- Avoid sedating medications (opioids, strong cough suppressants, benzodiazepines) unless approved by a Pompe specialist or pulmonologist.

For more information check out the fact sheet on: [Anesthesia Medical Safety List](#)

COUGH ASSIST (Mechanical Insufflator-Exsufflator):

- People with Pompe disease often cannot generate enough force to clear mucus or secretions, which can lead to infection or breathing difficulty.
- This device mimics a natural strong cough by inflating the lungs (*insufflation*) and then rapidly reversing airflow (*exsufflation*), simulating the forceful exhalation of a cough.

For more information check out the fact sheet on: [Cough Assist Machine](#)

INFECTION OR FEVER

- Treat infections promptly and aggressively.
- Even mild illnesses (such as colds, flu, bronchitis, or urinary tract infections) can rapidly worsen muscle and breathing weakness.
- Start antibiotics early if a bacterial infection is suspected.
 - Obtain cultures first if feasible, but do not delay antibiotics if:
 - Fever > 101 °F (38.3 °C)
 - Increased sputum production
 - New or worsening respiratory symptoms
 - Follow hospital or infectious-disease guidelines and avoid medications that depress breathing or weaken muscles.