



## ANESTHESIA PRECAUTIONS IN POMPE DISEASE

Patients diagnosed with Pompe disease (Glycogen Storage Disease Type II) have unique anesthesia risks due to respiratory muscle weakness, altered metabolism, and cardiac involvement. The following precautions are recommended for anesthesiologists.

### Respiratory Muscle Weakness

- Risk: Pompe disease affects the [diaphragm](#) and other respiratory muscles, leading to hypoventilation and possible CO<sub>2</sub> retention ([hypercapnia](#)).
- Caution: Even if oxygen saturation (SpO<sub>2</sub>) appears normal, patients may be retaining CO<sub>2</sub>.
- Recommendation:
  - Avoid giving supplemental oxygen without continuous CO<sub>2</sub> monitoring ([end-tidal CO<sub>2</sub>](#) or blood gas).
  - Support ventilation using noninvasive ventilation ([BiPAP](#)) as needed.
  - Plan for possible postoperative [ventilatory support](#), especially if baseline [FVC](#) <50% predicted or if sleep-disordered breathing is present.

### Avoidance of [Succinylcholine \(Suxamethonium\)](#)

- Reason: Risk of [rhabdomyolysis](#), [hyperkalemia](#), and [malignant-like hyperthermia](#) due to underlying myopathy.
- Alternative: Use [non-depolarizing neuromuscular blockers](#) in reduced doses and [titrate](#) carefully using a nerve stimulator.
- Monitor: Train-of-four response monitoring to guide reversal and recovery.

### Train-of-Four (TOF) Monitoring

- Definition: A neuromuscular monitoring technique that measures the degree of muscle relaxation produced by anesthesia drugs.
- How It Works: A nerve stimulator delivers four small electrical pulses (usually to the ulnar or facial nerve). Each pulse causes a muscle twitch, and the strength of these twitches shows how much blockade is present.
- Typical Interpretation:

TOF Response	Meaning
4 twitches, all equal	Minimal or no blockade
4 twitches, fading strength	Partial blockade
1–2 twitches present	Moderate blockade
0 twitches	Deep blockade

- Goal: Maintain adequate relaxation during surgery but avoid prolonged paralysis. A TOF ratio  $\geq 0.9$  indicates full recovery.
- In Pompe Disease: Use lower doses of non-depolarizing relaxants and monitor continuously with TOF. Weak baseline muscle tone makes accurate assessment and full reversal especially important.

## Cardiac Considerations

- Some Pompe patients (especially infantile or juvenile forms) may have [hypertrophic cardiomyopathy](#).
- Avoid: Agents that increase myocardial stress (e.g., halothane, high dose catecholamines).
- Use: ECG monitoring and consider echocardiogram review before anesthesia.

## Airway and Positioning

- [Macroglossia](#) or weak facial muscles can make [airway management](#) difficult.
- Plan for possible difficult intubation.
- Maintain neutral neck positioning; avoid excessive flexion that can compromise respiration.
- Avoid unnecessary intubation, use BiPAP if possible.

## Post-Anesthesia Recovery

- Extubation risk: Patients may have delayed emergence or reduced respiratory drive.
- Ensure full reversal of neuromuscular blockade.
- Monitor in a high-acuity or ICU setting postoperatively.
- Resume noninvasive ventilation as soon as possible.

## Anesthetic Drug Sensitivity

- Pompe patients can exhibit increased sensitivity to sedatives, opioids, and muscle relaxants due to low muscle mass or respiratory compromise.
- Use short-acting agents and titrate slowly.

## Multidisciplinary Coordination

- Pre-op evaluation should include:
  - Pulmonology (sleep study or CO<sub>2</sub> monitoring)
  - Cardiology (if any cardiac involvement)
  - Neuromuscular specialist
- Consider pre-op arterial blood gas or [capnography](#) baseline.

## Enzyme Replacement Therapy (ERT) Considerations

- If patient is on [ERT](#), note infusion schedules to avoid overlapping with surgery day to minimize infusion-related stress or allergic reactions.

## Temperature Control

- Maintain normothermia: Avoid hypothermia which can exacerbate muscle weakness and delay recovery.

## References

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### Glossary of Terms:

**Airway management** – The techniques used to keep the breathing passages open during anesthesia or emergency care.

**BiPAP (Bilevel Positive Airway Pressure)** – A noninvasive ventilation device that supports breathing by delivering two air pressures: one for inhalation and one for exhalation.

**Capnography** – A continuous monitoring method that measures carbon dioxide levels in exhaled air to assess ventilation.

**Cardiomyopathy** – A disease of the heart muscle that can make it harder for the heart to pump blood effectively; sometimes seen in Pompe disease.

**Depolarizing neuromuscular blocker** – A drug (like suxamethonium) that temporarily paralyzes muscles by activating and then blocking nerve signals.

**Diaphragm** – The main muscle involved in breathing, located below the lungs; weakness is common in Pompe disease.

**End-tidal CO<sub>2</sub> (ETCO<sub>2</sub>)** – The amount of carbon dioxide at the end of an exhaled breath; used to monitor ventilation during anesthesia.

**Enzyme Replacement Therapy (ERT)** – A treatment that provides the missing enzyme (acid alpha-glucosidase) to help reduce glycogen buildup in Pompe disease.

**FVC (Forced Vital Capacity)** – A breathing test that measures how much air a person can exhale after a deep breath, used to assess respiratory strength.

**Hypercapnia** – An excessive level of carbon dioxide in the blood due to inadequate ventilation.

**Hyperkalemia** – A condition where potassium levels in the blood are too high, which can disrupt heart rhythm and cause dangerous cardiac events.

**Hypertrophic cardiomyopathy** – Thickening of the heart muscle that can interfere with normal heart function.

**Macroglossia** – A condition where the tongue is abnormally large, which can make airway management more difficult.

**Malignant hyperthermia** – A rare, life-threatening reaction to certain anesthesia drugs that causes dangerously high body temperature and muscle rigidity.

**Neuromuscular blocker** – A medication used during anesthesia to relax or temporarily paralyze muscles to facilitate surgery or intubation.

**Non-depolarizing neuromuscular blocker** – A type of muscle relaxant that blocks nerve signals without activating the muscle first; preferred for Pompe disease patients.

**Rhabdomyolysis** – The breakdown of muscle tissue that releases proteins and electrolytes into the blood, which can damage the heart and kidneys.

**Suxamethonium (Succinylcholine)** – A short-acting depolarizing muscle relaxant; contraindicated in Pompe disease due to risk of hyperkalemia and muscle breakdown.

**Titrate** – To carefully adjust or measure the amount of a medication (like anesthesia or muscle relaxants) to achieve the desired effect without overdosing.

**TOF (Train-of-Four) monitoring** – A test used during anesthesia to measure how much muscle paralysis a neuromuscular blocker is causing.

**Ventilatory support** – Use of devices such as BiPAP or mechanical ventilation to help patients breathe when respiratory muscles are weak.