- Diagnosis confirmed by analysis of serum muscle enzymes, EMG findings, and muscle biopsy (most definitive test).
- A careful family history, medication list review, physical exam, blood test, and muscle biopsy are all crucial because they may help to exclude an alternative diagnosis, such as an inherited muscle disease or toxic myopathy.

Etiology

- An autoimmune etiology is suspected and hypothetically supported by an association with other autoimmune or connective tissue diseases and a response to immunotherapy.
- Drugs—especially D-penicillamine, statins, or zidovudine—may also trigger an inflammatory myopathy.
- Several viruses—including coxsackie, influenza, mumps, CMV, and Epstein-Barr virus—may also have an association.

Usual Treatment

- Treatment focuses on controlling inflammatory response through immunosuppression.
- Steroids, with prednisone as first-line agent.
- Immunosuppressive drugs, which include azathioprine, methotrexate, mycophenolate mofetil, rituximab, cyclosporine, tacrolimus, cyclophosphamide.
- IVIG.
- Physical therapy.

Assessment Points

System	Effect	Assessment by Hx	PE	Test
HEENT	Neck muscles weakness	Headache Head drop	Neck ROM Head lift	EMG
RESP	Inspiratory muscles weakness Interstitial lung disease Aspiration pneumonitis	Dyspnea Chronic cough Limited exercise tolerance	Dyspnea/tachypnea Wheezing Hypoxia	PFTs CXR, CT scan ABG Bronchoscopy
CV	Conduction abnormalities CHF	Chest pain Dyspnea Palpitations	Arrhythmia Edema Inspiratory crackles	ECG TTE Stress test
GI	Pharyngeal muscle weakness	Dysphagia	Regurgitation Aspiration	Endoscopy CXR
CNS	Systemic manifestations	Malaise Fever	Hyperthermia	
HEME	Raynaud phenomenon	Cold digits	Digit vasospasm	
DERM	Only seen with concomitant disease, dermatomyositis	Rash	Erythematous and raised papules on extensor surfaces Heliotrope rash	Muscle/skin biopsy
MS/RHEUM	Proximal muscle weakness Arthralgias or arthritis Calcinosis of subcutaneous tissue Coexisting rheumatologic disorder (scleroderma, SLE)	Myalgia Muscle tenderness Skin ulceration Joint swelling	Muscle weakness, atrophy Delayed reflexes	CPK, CK, ALT, AST, LDH Autoantibodies EMG Joint x-ray Muscle biopsy

Key References: Strauss KW, Gonzalez-Buritica H, Khamashta MA, et al.: Polymyositis and dermatomyositis: a clinical review, *Postgrad Med J* 65(765):437–443, 1989; Gunusen I, Karaman S, Nemli S, et al.: Anesthesia management for cesarean delivery in a pregnant woman with polymyositis: a case report and review of literature, *Cases J* 2:9107, 2009.

Perioperative Implications

Preoperative Preparation

- Assess cardiovascular and pulm status.
- Consider use of RA in order to limit GA and use of NMB; there are some case reports of successful and safe neuraxial techniques and limited reports on peripheral nerve blockade.
- Concomitant steroid therapy and necessity of stress doses should be considered.

Monitoring

- Arterial line if indicated (either owing to CHF or frequent blood draws for ABG)
- TOF peripheral nerve stimulation with NMB use (consider baseline stimulation before NMB given)
- Foley catheter for urine output assessment if pt has cardiac disease

Airway

- Consider rapid sequence intubation if pt has dysphagia.

Induction

- Avoid use of succinylcholine (may cause hyperkalemia).
- Volatile agents and succinylcholine may serve as a trigger malignant hyperthermia and should be avoided in pts with baseline elevated CPK levels.
- If not necessary, avoid nondepolarizing NMB due to increased sensitivity (vecuronium and pancuronium associated with prolonged neuromuscular paralysis).
- Consider use of remifentanil to aid with intubation/for hypokinesis.

Maintenance

- Volatile anesthetics may potentiate the effects of muscle relaxation.
- Consider total IV anesthetic technique.
- Antagonism to NMB may cause additional muscle weakness and/or cardiac dysrhythmias.
- Consider stress-dose steroids.
- Avoid overuse of narcotics.
- Keep pt euvolemic to avoid heart failure.

Extubation

- If NMB is used, confirm complete reversal using twitch monitor and confirm full strength of pt with head lift (if able to do so preop).
- Confirm that pt is completely awake and able to breathe independently of ventilator prior to extubation.
- Consider NIF test to assess adequacy of strength of ventilation.

Postoperative Period

- If possible, keep head of bed elevated to assist with pulm function and to avoid an aspiration event.
- Increased susceptibility to infection if on immunosuppression.

Anticipated Problems/Concerns

- May need ICU stay postop to wean off ventilator.
- Pain control management; avoid overuse of narcotics, which may lead to oversedation and/or apnea.
- May need continued dose of stress dose steroids through periop period.
- Volume shifts may complicate cardiac status.
- Consider swallow study before oral intake to avoid unanticipated dysphagia and an aspiration event.

Pompe Disease

Sheri Jones Oguh | Lee A. Fleisher

Risk

- Combined incidence (infantile vs. late-onset): 1:40,000.
- Infantile form has higher incidence in African-American and Chinese populations.
- Late-onset disease has a higher incidence in the Netherlands.

Perioperative Risks

- Respiratory insufficiency
- Aspiration pneumonia
- Pulm edema
- Myocardial ischemia

Worry About

- Respiratory insufficiency, which may require prolonged mechanical ventilation
- Myocardial ischemia
- Arrhythmias, sudden death

- GE reflux, aspiration pneumonia
- Difficult extubation and ventilator dependence

Overview

- Only glycogen storage disease that is also a lysosomal storage disease.
- Deficiency of lysosomal enzyme acid-α glucosidase.
- Lysosomal glycogen accumulates in several organ systems, most importantly cardiac, skeletal, and smooth muscle.
- Clinical features are mostly neuromuscular.
- Two major forms: Infantile versus late-onset.
- Infantile presents with cardiomyopathy, hypotonia, and muscle weakness ultimately leading to death secondary to cardiorespiratory failure within the first year of life.
- Cardiomyopathy usually not seen in late-onset variants, which can present at any age. Usually characterized by muscle weakness followed by muscles of respiration and diaphragm. Respiratory failure is usually cause of death or severe morbidity.
- Pts with late-onset Pompe disease usually present with slowly progressive myopathy, which can be mistaken for limb girdle muscular dystrophy.
- Measurement of GAA activity in skin fibroblasts is the current gold standard test to confirm diagnosis.

Etiology

- Disorder of acid α-glucosidase.
- Disease severity correlates inversely with residual acid α-glucosidase activity.
- Autosomal recessive inheritance pattern.

Usual Treatment

- Enzyme therapy can be helpful in infantile variants, improving cardiac and respiratory function
- Supportive therapy

Assessment Points

System	Effect	Assessment by Hx	PE	Test
HEENT	Weakness of muscles of inspiration and diaphragm	Open-mouth breathing, decreased gag reflex	Macroglossia	
RESP	Atelectasis (compression of bronchi by enlarged heart), Weakness of muscles of inspiration and diaphragm, decreased vital capacity, diaphragmatic dysfunction	Dyspnea Open-mouth breathing, protrusion of tongue (may mimic macroglossia) Frequent pulm infections, aspiration pneumonia Sleep apnea, morning headaches, somnolence Weak cough, use of accessory muscles	Decreased breath sounds	CXR Pulm function tests (e.g., FEV_1) Polysomnography ABG
CV	Biventricular hypertrophy, short PR interval (interference with specialized conducting tissues) Cardiomegaly LV dysfunction Cardiomyopathy with or without left ventricular outflow obstruction	Palpitations, CHF symptoms Exercise intolerance	Murmur, gallop, arrhythmias	ECG CXR ECHO 24-h Holter monitoring
GI	Weakness of oropharyngeal muscles	Difficulty chewing/swallowing, failure to thrive GE reflux	Underweight, macroglossia, decreased gag reflex Hepatomegaly	
CNS	Glycogen accumulation in skeletal, cardiac, and smooth muscle	Muscle weakness, hypotonia, developmental delay, gross motor delay, loss of early motor milestones		
HEME	Elevated CK, LDH, AST, ALT May reflect enzymes released from muscle			CK
MS	Glycogen accumulation	Myopathy Exercise intolerance Limb-girdle weakness Gait abnormalities		EMG, muscle biopsy

Key References: Kishnani PS, Steiner RD, Bali D, et al.: Pompe disease diagnosis and management guideline, *Genet Med* 8(5):267–288, 2006; McFarlane HJ, Soni N: Pompe's disease and anaesthesia, *Anaesthesia* 41(12):1219–1224, 1986.

Perioperative Implications

Preoperative Preparation

- Assess myocardial, respiratory, and volume status.
- Consider regional anesthetic techniques.

Monitoring

- Arterial line if indicated.
- Consider CVP or PA catheter if indicated.
- ECG with focus on ST segments; pulse oximetry.
- Capnography.

Airway

- Macroglossia
- Impaired gag reflex

Preinduction/Induction

- Avoid hypotension, as this can precipitate arrhythmias from ischemia due to a hypertrophied LV.
- Maintenance of normal coronary perfusion pressure is of utmost importance.
- Maintain a higher filling pressure for adequate preload and a normal to high systemic vascular resistance to ensure effective coronary perfusion.
- Consider ketamine, as it maintains SVR and contractility and is less likely to reduce preload.
- Etomidate is another consideration.
- Inhalation agents should be used cautiously and in pts with a lesser degree of cardiac hypertrophy.
- High induction doses of propofol should be avoided.
- Pts may be more sensitive to neuromuscular blockade.
- Malignant hyperthermia precautions should be followed.

Maintenance

- If neuromuscular blockade required, choose one that has the least amount of cardiac depressant ability and shortest duration.
- Beta blockers must be used carefully, as there is anecdotal evidence of sudden death in the pediatric population.
- Avoid drastic changes in volume status (hypovolemia vs. fluid overload).

Extubation

- Period with the greatest O_2 demands

Postoperative Period

- Impaired cough predisposes pt to atelectasis and aspiration pneumonia.
- Chronic CO_2 retention and hypoxemia.
- Sleep disordered breathing due to supine position and effect of sleep on respiratory control mechanism.
- Important to focus on pulm toilet, bronchodilators.
- Supplemental O_2 versus CPAP may be required to treat hypoxemia.

Anticipated Problems/Concerns

- Myocardial ischemia.
- Respiratory insufficiency.
- Decreased CO.
- Pulm infections should be treated aggressively.