Myotonia Dystrophica (Myotonic Dystrophy, Steinert Disease)

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Risk

- Incidence of approximately 1:8000.
- Incidence of the congenital form is higher, with an incidence of 1:100,000 compared with adult-onset form.

Perioperative Risks

- Operative/anesthetic and postop morbidity and mortality are increased and not proportional to severity of disease.
- High incidence of cardiopulmonary complications, including sudden death, cardiac failure, and cardiomyopathy.

Worry About

- Increasing frequency of symptoms
- Signs of respiratory or cardiac decompensation

Overview

- Degenerative disease of skeletal muscles. It consists of a triad of characteristic features, including frontal baldness, cataracts, and mental retardation.
- It can be variable in presentation. Some are asymptomatic, whereas more severe congenital manifestations include mental retardation and respiratory insufficiency.
- Typically the onset of symptoms in second and third decades of life with progressive muscular weakness and wasting, most common in the cranial and distal limb muscles (e.g., temporalis and masseter muscle atrophy, known as "hatchet face" and "limb muscles"). There may be diminished deep tendon reflexes and muscles of the vocal cord apparatus resulting in nasal speech. A proximal muscle variant has recently been recognized. Death frequently occurs in the fifth or sixth decade of life and is usually related to cardiopulmonary complications, including sudden death from conduction abnormalities, cardiomyopathy, and/or CHF.
- There is often persistent contracture after cessation of stimulation or voluntary contraction of the muscle. This inability of the skeletal muscle to relax is diagnostic. EMG is corroborative and pathognomonic and it shows continuous low-voltage activity with high-voltage, fibrillation-like potential bursts.
- Myotonic dystrophy is an intrinsic disorder of skeletal muscle linked to a myotonin-protein kinase gene on chromosome *19q13.2*. A defect in Na^+ and Cl^- channel function produces electrical instability of the muscle membrane and self-sustaining runs of depolarization. Abnormal metabolism of calcium may be seen.

Etiology

- Myotonic dystrophy is inherited via an autosomal dominant trait. It occurs from an abnormal expansion of the nucleotide CTG on chromosome 19, which codes for a serine-threonine protein kinase. Variable gene expressivity can be seen within the same family in which one member can have minimal affects and another be severely affected. Anticipation is seen with inheritance, and the longer the CTG repeat, the more severe the disease is.

Usual Treatment

- Quinine, procainamide, phenytoin, tocainide, mexiletine (depress Na^+ influx)

Assessment Points

System	Effect	Assessment by Hx	PE	Test
HEENT	Visual disturbance Speech/swallowing impaired		Presenile cataract, ptosis, strabismus, retinal pigmentation Generalized weakness of pharyngeal, mandibular (and thoracic) musculature Dysarthria, facial weakness Expressionless facies	Exam by ophthalmologist
CV	Dysrhythmias Cardiomyopathy	CHF uncommon but may occur with pregnancy	Delayed intraventricular conduction Heart block, hypotension Up to 20% with mitral valve prolapse, sudden death	ECG ECHO, Holter Cardiology consult
RESP	Restrictive lung disease Chronic aspiration Central hypoventilation	Weak cough Dyspnea Hx of pneumonia	Wasting of sternocleidomastoid muscles; respiratory muscle weakness Lungs intrinsically normal; decreased VC, decreased ERV, increased CO_2	PFTs ABG
GI	High aspiration potential Delayed esophageal and gastric emptying Gastric dilation/atony Increased incidence of cholelithiasis	Weak swallowing ability		
ENDO/IMMUNE	Testicular atrophy DM Decreased thyroid function Adrenal insufficiency Frontal balding Malignant hyperthermia		Thyroid nodules Decreased immunoglobulins	Blood/urine glucose tests Thyroid function tests
CNS	Mental retardation Associated with central sleep apnea and hypersomnolence Emotional abn		Myotonic handgrip (delayed, incomplete release), increased CK in serum Myotonia can be initiated or worsened by exercise or cold temp; decreased DTR	EMG CK
GYN	Pregnant pt is a challenge. Respiratory function threatened by decreased FRC and myotonic weakness, which may be exacerbated by pregnancy. Seems to be added risk for uterine hemorrhage at delivery due to uterine atony and retained placenta. C-section may be safer.			

Key References: Aldredge LM: Anaesthetic problems in myotonic dystrophy. A case report and review of the Aberdeen experience comprising 48 general anesthetics in a further 16 patients, *Br J Anaesth* 57:1119–1130, 1985; Mathieu J, Allard P, Gobeil G, et al.: Anesthestic and Sugrical complications in 219 cases of myotonic dystrophy, *Neurology* 49(6):1646–1650, 1997.

Perioperative Implications

Preoperative Preparation

- Ensuring NPO status (increased aspiration) and recent ECG.
- No preop analgesics or sedatives and caution with benzodiazepines.
- Warm ambient room air in OR may decrease incidence and severity of myotonia.
- Routine monitoring.

Airway

- Propensity for frequent jaw dislocation
- Potential inability to secure airway because of jaw muscle spasm

Preinduction/Induction

- Risk for aspiration of gastric contents.
- Induction: Gaseous; avoid slow metabolizing hypnotics; use lower doses on propofol.
- Relaxation: Avoid succinylcholine (link to malignant hyperthermia, severe extended contractures); use short-acting nondepolarizing agents at lower doses; recovery may be prolonged.
- May be hard to differentiate from onset of MH.

Maintenance

- Myotonia may be precipitated by drugs (e.g., propofol, succinylcholine, anticholinesterases, halothane, neuroleptics, liquid paraffin), physical factors (e.g., cold, shivering), surgical manipulation, or electrocautery.
- Avoid K^+-containing fluids.
- Regional or local anesthesia acceptable but will not block myotonic response.
- Regional ± TIVA may be preferable when suitable extubation.
- Beware of airway obstruction because of jaw muscle weakness.
- Delayed recovery from anesthetic common.

Extubation

- Beware of airway obstruction because of jaw muscle weakness.
- Delayed recovery from anesthetic common.
- Sugammadex has been described in literature with successful reversal of steroidal NMBs.

Adjuvants

- Increased sensitivity to ventilatory depressant effects of all premedicants, sedatives, and opioids.

- Reversal agents can theoretically precipitate skeletal muscle contraction by facilitating depolarization of NMJ, but adverse responses do not predictably occur.

Postoperative Period

- Increased sensitivity to respiratory depressant effects of opioids or sedatives, including epidural opioids
- Postop pain to be managed with NSAIDs, regional blocks, and acetaminophen if possible
- Pulm complications due to poor cough possible
- Cardiac and respiratory monitoring and early chest physiotherapy

Anticipated Problems/Concerns

- If myotonia develops intraop, neither GA nor RA nor NMBs will attenuate it. Local infiltration of involved muscles may help. Even asymptomatic pts may have some degree of cardiomyopathy. Beware of premature extubation, and consider postop ventilation.
- 57% of these pts have conduction defects, with one-third having primary block unresponsive to atropine. It is advisable to have antiarrhythmics and transthoracic pacing readily available as many anesthetic agents can increase vagal tone.
- For numerous reasons, it is advisable to avoid GA (e.g., myocardial depressants, conduction effects, link to malignant hyperthermia). Pts should have procedures done with RA if at all possible.

Myxoma

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Risk

- Although primary cardiac tumors are rare (<0.01%), myxoma is the most common type (50%).
- 75% develop in LA, with most attached to the interatrial septum.
- Rarely develop in ventricles.
- More common in females (70%).

Perioperative Risks

- May be friable and may embolize (30–40% of pts)
- LV- or RV-inflow obstruction with resultant hypotension
- May simulate pulm Htn and/or constrictive pericarditis physiology

Worry About

- Hypotension due to obstruction of ventricular inflow and/or incompetence of tricuspid (right) or mitral (left) valve, may be positional.
- Tumor flips on a stalk across valves, causing stenotic and/or incompetent symptoms.
- RV hypertrophy can occur because of longstanding left ventricular–inflow obstruction.
- There is the possibility of pulm or systemic embolization.

Overview

- Is a true neoplasm and distinct from a thrombus
- Usually polypoid, pedunculated with a 1–2 cm stalk, and round with smooth margins
- Typically grows very slowly before the patients becomes symptomatic (10–20 y)

Etiology

- Typically arises from the endocardium and rarely extends deeper.
- Polyhedral cells with small nuclei are separated by an afibrillar, eosinophilic myxomatous stroma that is predominantly a mucopolysaccharide.
- Although benign, this tumor rarely can undergo malignant degeneration.

Usual Treatment

- Surgical, usually curative
- Cardiopulmonary bypass required
- Median sternotomy, atriotomy with transseptal approach through fossa ovalis
- Resection including the root of the pedicle and the full thickness of the adjacent septum and then ASD closure

Assessment Points

System	Effect	Assessment by Hx	PE	Test
CV	Mitral or tricuspid stenosis or insufficiency syndromes	Edema and CHF	Atrial enlargement Systolic murmur (regurgitation) Diastolic murmur (stenosis)	ECHO, ECG, CXR CT, MRI
RESP	Pulmonary emboli (right)	DOE and cough	Rales, wheezing, and increased P_2	ECHO, CXR, ECG
GI		CHF	Hepatic enlargement	Hepatic enzymes (if symptoms of CHF)
RENAL	Emboli (left)			Urinalysis Cr clearance
CNS	Stroke (left)	CNS dysfunction	CNS dysfunction	ECHO
GENERAL	Constitutional symptoms	Fever and malaise	Weight loss	ESR, CRP, Hct (anemia)

Key References: Reynen K: Cardiac myxomas, *N Engl J Med* 333(24):1610–1617, 1995; Essandoh M, Andritsos M, Kilic A, Crestanello J: Anesthetic management of a patient with giant right atrial myxoma, *Semin Cardiothorac Vasc Anesth* 20(1):104–109, 2016.

Perioperative Implications

Preoperative Preparation

- Differential Dx: Mitral stenosis/insufficiency (left), tricuspid stenosis/insufficiency (right), constrictive pericarditis, pulm Htn, and subacute bacterial endocarditis.
- Mitral stenosis: Hemodynamic aim is to keep pt in normal sinus rhythm with adequate preload and high-normal afterload (see Mitral Stenosis).
- Mitral insufficiency (regurgitation): Hemodynamic aim is to keep HR normal or fast and to vasodilate.
- Hemodynamics can mimic any or all of the above, depending on load-dependent variables prevailing in the cardiac cycle at the time (e.g., preload, afterload, HR)

Monitoring

- Routine monitors otherwise needed for cardiopulmonary bypass (e.g., standard ASA monitors, temperature, ECG, coagulation, Foley).
- Intra-arterial catheters.
- Beware of central line with right-sided atrial myxoma (may cause dislodgment of friable debris as pulm emboli); TEE guidance (bicaval view) of guidewire placement may be helpful.
- TEE: Most sensitive way to guide hemodynamic management and assess the therapeutic approach.

Airway

- Routine

Preinduction/Induction

- Pt may develop hypotension if preload is decreased or HR is increased; best managed with a vasopressor (no or judicious use of inotropes).
- Insert a central venous catheter carefully and avoid a PA catheter in pt with a right-sided tumor.
- Intraop TTE is helpful/diagnostic (before induction) if there is concern about right inflow obstruction exacerbated by PPV and for monitoring of IV fluid administration.
- Avoid/treat atrial dysrhythmias.
- Have a surgical team present on induction in case of CV collapse.
- Initiate PPV carefully.

Maintenance

- May dislodge pieces during CPB venous cannulation; direct assessment of anatomy, physiology, and even placement of venous cannulas should be guided by TEE.
- If pedunculated, a tumor may obstruct inflow tract, and hemodynamics may present as low BP, low CO, or increased CVP (right)/increased PCWP (left).

Extubation

- Expect separation from CPB with minimal support and overall excellent recovery with primary myxomatous lesion.
- Extubation criteria should be based on myocardial protection techniques and post-CPB bleeding risk.
- Early extubation consideration is reasonable.

Postoperative Period

- Beware of residual ASD (as tumors typically originate in the atrial septum in the region of the fossa ovalis).
- Beware of conduction and rhythm disturbances (esp. in pediatric pts).
- Symptoms of pulm Htn usually regress quickly.

Anticipated Problems/Concerns

- Hypotension with inadequate preload when the lesion obstructs ventricular inflow