Muscular Dystrophy

Hereditary disease of painless degeneration and atrophy of skeletal muscles with preserved innervation, often accompanied by developmental delay.

ANESTHETIC CONSIDERATIONS:

- **Difficult airway** d/t poor mouth opening, hypertrophy of tongue & perioral muscles
- Increased **aspiration risk**
  - Decreased gut motility
  - Weak laryngeal reflexes
- Chronic respiratory insufficiency
  - Recurrent infections
  - **Restrictive lung pattern** (pulmonary HTN, cor pulmonale)
  - Postoperative respiratory failure
  - **Sleep apnea** may occur
- Cardiac **arrhythmias**, **cardiomyopathy** and **MR** / MVP
- Altered pharmacology
  - **SCh contraindicated d/t hyperkalemic arrest** from rhabdomyolysis
  - Volatiles: case reports of rhabdomyolysis / hyperkalemic arrest
  - NDMB: *may* have prolonged response
  - MH does not appear to be associated with DMD
- Potential for **increased blood loss during surgery**
- **Steroid use**

ANESTHETIC GOALS:

- Minimize risk of aspiration and failed intubation
- Minimize risk of cardiopulmonary insufficiency / failure
- Avoid drugs and situations leading to rhabdomyolysis / hyperkalemia (i.e. non-triggering anesthetic)

HISTORY

- In addition to usual anesthetic history – esp. interested in MH
- Functional enquiry often not helpful
  - If patient can stand, generally pulmonary function is OK
- Recent respiratory illnesses / hospitalizations / treatments, including steroid use
- Consider history of motor milestones in all young boys

PHYSICAL

- **GEN** – often obese – difficult IV start
- **HEENT** – macroglossia sometimes present
- **CVS** – opening snap (MVP), MR, CHF signs
- **CNS** – mental status exam

INVESTIGATIONS

- **Labs**
  - Depending on procedure, other labs – Hb, cross-match, etc.
- **Imaging**
  - CXR
  - EKG – tall R waves in V1, deep Q waves limb leads, short PR and sinus tachycardia
  - Echo
- **Special**
  - PFTs, esp. important if wheelchair-bound - at least ABG if unable to obtain PFTs

OPTIMIZATION

- Delay elective surgery if cardiopulmonary function not optimized
  - Consider canceling if function severely depressed
- Aspiration prophylaxis
- Cardiac, meds for arrhythmias, may need pacemaker, cardiology consult
- Optimization of respiratory status
- SBE prophylaxis as needed
- Steroid prophylaxis

ANESTHETIC OPTIONS

LOCAL

- Often muscle biopsy can be performed under LA with sedation

REGIONAL

- Consider regional technique as sole technique or as adjuvant
- Neuraxial techniques for lower limb or labor / c-section

GENERAL

- **TIVA** using propofol / opioids / NDMB considered safe for all MD
- Volatiles may cause rhabdomyolysis – thought to be due to myocyte fragility
- Consider short-acting or low dose muscle relaxants (or none)

ANESTHETIC SETUP

- **Drugs**
  - Standard
  - Have dantrolene available in case of MH (some case-reports but DMD not strictly considered a risk factor for MH - ++ controversy surrounding this)
- **Equipment**
  - Usual monitors
  - Foley catheter (urine dip for myoglobin)
  - Consider invasive hemodynamic monitoring for complex patients or cases
  - Consider PA catheter / TEE based on EF and surgical procedure
  - Nerve stimulator
  - Core temperature monitor
  - Prudent to document minute ventilation, core temperature in addition to usual vitals

MANAGEMENT OF ANESTHESIA

- **Induction**
  - Pre-oxygenate as FRC reduced, propofol / opioid +/- NDMB, consider remifentanil for intubation instead of NDMB
  - Succinylcholine contraindicated because of rhabdomyolysis, hyperkalemia
  - **Long gastric emptying times, possible full stomach therefore RSI (conflict with SCh)**
  - NDMB: usual dosing but *may* be slightly prolonged
  - **Non-triggering anesthetic if possible** (although doesn't guarantee against rhabdomyolysis, hyperkalemia)
  - Avoid cardio-depressants if cardiomyopathy present
- **Maintenance**
  - Consider **TIVA**
  - Titrate NDMB to effect
- **Emergence**
  - Ensure reversal of NDMB
  - Extubate in OR if concerned

DISPOSITION & MONITORING

- Potential for prolonged **ventilator dependence when VC < 30% predicted**
- Ensure adequate post-operative monitored environment for 24 – 48 hours
  - Delayed respiratory distress can occur up to 36 hrs postoperatively, cause unknown
- Monitor for cardiac events - telemetry

COMPLICATIONS

- If undiagnosed patient receives SCh or volatiles → hyperkalemia & cardiac arrest
  - WARNING – the hyperkalemic response to SCh (cardiac arrest) has been described in boys 4months old w/out clinical signs of Duchenne muscular dystrophy
- Inability to intubate / ventilate
- Cardiac arrhythmias (SVTs), complete heart block, cardiomyopathy decompensation (failure), cardiac arrest
- Hyperkalemia, rhabdomyolysis
- Aspiration
- Postoperative respiratory insufficiency / failure

PREGNANCY

- Pregnant women with muscular dystrophies have increased incidence of operative deliveries
- **Neuraxial techniques are preferred for L&D and C-section**
- Severe disease may result in airway abnormalities and spinal deformities which can complicate general or neuraxial anesthesia
- Severe kyphoscoliosis can prevent normal adaptive hyperventilation
- About 2.5% female carriers of Duchenne MD have symptoms, though usually milder forms
  - These patients are at risk of hypermetabolic syndrome similar to MH with Sux or volatile halogenated anesthetics (according to Chestnut 4th)
  - Therefore, non-triggering anesthetic should be used

PATHOPHYSIOLOGY

- Hereditary disease of painless degeneration and atrophy of skeletal muscles with preserved innervation
  - Often accompanied by developmental delay
- Frequency: Duchenne's muscular dystrophy > limb-girdle > fascioscapulohumeral > nemaline rod myopathy > oculopharyngeal dystrophy

DUCHENNE MD

- X-linked recessive
  - Deletion mutation of the dystrophin gene on Xp21
  - Female carriers may be affected (esp. cardiac)
- **Dystrophin** = protein in striated, smooth and cardiac muscle which stabilizes the cell membrane
  - Its **absence** results in muscle cell degeneration and death, replaced by fatty infiltrates
  - Steady deterioration in skeletal muscle strength with confinement to wheelchair by 8-11 years
  - Death usually by 15-25 d/t pneumonia and / or CHF
- Orthopedic complications
  - Fractures of extremities frequent
  - Progressive scoliosis
- Cardiopulmonary dysfunction

- **Chronic respiratory muscle weakness** with decreased ability to clear secretions
  - Recurrent pneumonias
  - **Kyphoscoliosis**: increased **restrictive** pulmonary pattern of disease – leading to pulmonary hypertension, cor pulmonale
  - Sleep apnea can also be contributing factor to **pulmonary HTN**
- **Cardiac abnormalities**
  - Characteristic changes in EKG:
    - Tall R waves in V1
    - Deep Q waves in limb leads
    - Short PR intervals
    - Sinus tachycardia
  - **Cardiomyopathy** – primarily dilated CM, biventricular dysfunction
    - Present in 1/3 by age 14
    - Present in 70% (symptomatic in 10%)
  - Cardiac **arrhythmias, heart block**
  - **MR, MVP d/t papillary muscle dysfunction** and decreased myocardial contractility

BECKER MD
- X-linked recessive
- Age of onset later than DMD
- **Milder** than DMD – ambulatory until 15 y.o.
- Mental retardation and contractures not as severe
- However, **cardiac involvement is usually more severe** (cardiomyopathy, arrhythmias, heart blocks)
- Usually survive beyond age 30

EMERY-DREIFUSS MD
- X-linked recessive
- Skeletal muscle **contractures** (humeroperoneal distribution) precede weakness
- **Cardiac** involvement may be life-threatening (CHF, bradycardia, etc.), **worse** than DMD
- Female carriers may experience cardiac impairment

LIMB-GIRDLE MUSCULAR DYSTROPHY
- Slowly progressive
- Relatively benign
- Onset 2-5th decade
- Shoulder or hip girdle muscle may be the only site involved

FACIOSCAPULOHUMERAL MD
- Slowly progressive wasting of facial, pectoral and should girdle muscles beginning in adolescence
- Eventually lower limbs are involved
- No cardiac involvement

NEMALINE ROD MD
- Autosomal dominant
- Slowly progressive or non-progressive symmetrical dystrophy of skeletal and smooth muscle
- Delayed motor development, hypotonia, loss of deep tendon reflexes
- Typical **dysmorphic** features
- Intelligence usually normal
- Micrognathia, dental malocclusion, high-arched palate → **potential difficult a/w**
- Bulbar palsy → aspiration risk
- Kyphoscoliosis and pectus excavatum
- Restrictive lung disease
- Dilated CM
- Response to Sux and NDMB unpredictable
- MH has *not* been reported

OCULOPHARYNGEAL DYSTROPHY
- Rare
- Progressive dysphagia and ptosis
- Aspiration risk

REFERENCES

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