

## PBLD – Table #17

### Anesthetic Implications of Duchenne Muscular Dystrophy

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#### Objectives:

1. Discuss the pathophysiology and natural progression of Duchenne Muscular Dystrophy (DMD)
2. Discuss the preoperative evaluation of a patient with DMD
3. Discuss the intraoperative and postoperative concerns for anesthesia in patients with DMD

**Case Presentation:** An 18-year-old male with DMD is scheduled for G-tube placement.

#### Questions:

1. What is muscular dystrophy? How does it compare to other muscle diseases (ie, myasthenic syndromes, myotonic syndromes, mitochondrial myopathies)? Discuss how action potentials are translated to muscle contraction and how that process is disturbed in each of these disorders.
2. How are muscular dystrophies classified? What other muscular dystrophies may be encountered by the anesthesiologist (ie, Becker or Emery-Dreifuss muscular dystrophy)?
3. Discuss the pathophysiology of DMD. How is DMD inherited? What protein is responsible? How does the lack of this protein affect muscle membrane stability? How does instability lead to the manifestations of DMD? What are the manifestations of DMD (ie, how and at what age does it present, which organ systems are involved in addition to musculoskeletal, what is the natural progression and life expectancy, what is the common cause of death)? How is a diagnosis of DMD made?
4. What medical and surgical management are commonly needed for DMD patients? For what procedures might they present to the OR?

**Case Presentation:** The patient is in preop holding and needs to be evaluated for readiness for his procedure.

#### Questions:

1. Should undiagnosed patients be screened for muscular dystrophies prior to anesthesia? How?
2. What kinds of studies might be helpful in evaluating these patients? Discuss appropriate cardiac and pulmonary evaluations.
3. What physical exam findings might affect your anesthetic plan? Discuss airway exam and implications of contractures on the anesthetic plan.
4. What history is important to elicit and document? Discuss the importance of documenting baseline neurologic and motor exams.

**Case Presentation:** The patient is 18-years-old. He is almost entirely bed-bound and refuses even wheelchair use. He has a significant cardiomyopathy with EF 23% and mild mitral regurgitation, but no history of arrhythmias. He has respiratory insufficiency with a severe restrictive defect (FVC 14% predicted) and requires nocturnal BiPAP support (settings 12/6 x10, FiO<sub>2</sub> 21%). His room air sats are in the mid-90s and he has mild CO<sub>2</sub> retention. His medications include carvedilol, spironolactone, furosemide, digoxin, ipratropium and albuterol. He has multiple contractures, including c-spine flexion, a limited mouth opening and an enlarged tongue. He has significant anxiety and panic attacks regarding

this procedure. He is currently at his baseline medical status, all his evaluations have been completed in the last 3-6months with follow-up clinic visits in the last month and he is appropriately NPO for surgery.

**Questions:**

1. Is any further work-up required at this time?
2. What are the risks for DMD patients undergoing anesthesia? Discuss rhabdomyolysis and hyperkalemia (specifically, what medications are implicated, timing, resuscitation, MH susceptibility), cardiac and respiratory failure, difficult airway, increased blood loss, abnormal response to neuromuscular blockade.
3. Given these risks, are additional monitors required for these patients? Discuss arterial and central venous lines. Should additional equipment and medications be available in the OR? Discuss difficult airway equipment, rapid transfusers, code drugs and defibrillator.
4. What surgical options are available for this procedure (open, laparoscopic, endoscopic)? How will each of these options affect the patient's cardiac and respiratory status? How will the surgical approach affect the anesthetic plan (general anesthesia vs MAC, controlled vs spontaneous ventilation)?
5. Should advanced directives be discussed with this patient as part of the evaluation? The patient wants a guarantee that he will not remain intubated post-operatively. What do you tell him?

**Case Presentation:** The decision was made to proceed with an EGD for PEG placement, as it was the least invasive option. Due to the patient's significant anxiety, a general anesthetic was necessary for the procedure. Controlled ventilation with intubation was chosen given his baseline respiratory insufficiency. He was difficult to intubate, requiring two attempts and a glidescope. Induction and maintenance were achieved with IV agents, including midazolam, etomidate, scopolamine, remifentanyl and ketamine. Surgical duration was less than 15minutes. The patient remained hemodynamically stable and was extubated at the end of the procedure.

**Questions:**

1. Discuss and debate the management of this case. Would other surgical or anesthetic options have been better?
2. What are the postoperative risks for this patient? Discuss respiratory failure and hyperkalemic arrest. What post-operative monitoring is required for this patient?
3. Where should this patient be sent post-operatively (ICU vs PACU)? Do all DMD patients require post-operative ICU admission? What factors should be considered in making this decision?

**Case Presentation:** The patient was sent to the ICU post-operatively and immediately started on BiPAP at his home settings. He restarted his home medications and pain was managed with tylenol. He was discharged on POD #1.

**Discussion:**

The muscular dystrophies are a group of genetic disorders that produce abnormalities of skeletal muscle resulting in variable and progressive muscle degeneration without primary structural abnormalities of the lower motor neuron. They are classified by distribution and severity of the weakness and may include abnormalities in other systems. Cardiac muscle is often involved and may be more severely affected than skeletal muscle. Duchenne and Becker muscular dystrophies (DMD and BMD) are caused by abnormal dystrophin, and are therefore called dystrophinopathies. DMD is the most common progressive muscular dystrophy with the most severe course. It is an x-linked dystrophin gene mutation that occurs in 1:3500 live male births. DMD patients experience painless skeletal muscle degeneration starting at 3-5 years old. They initially develop pelvic weakness and become non-ambulatory by 10-12 years old, then develop fixed skeletal deformities and upper extremity weakness in their teens. As the disease progresses, these patients develop a restrictive lung defect and a prone to recurrent aspiration, leading to respiratory failure,

and conduction abnormalities and dilated cardiomyopathy leading to cardiac failure. Patients with DMD usually die in their late teens-to-mid 20s from respiratory or cardiac failure. These patients may present to the operating room for a variety of procedures, including orthopedic procedures to maintain mobility and tracheostomy, ICD or G-tube placement.

The preoperative evaluation should focus on the patient's baseline cardiac, pulmonary and motor function, airway assessment (including both physical findings and aspiration risk) and evaluation for contractures that will affect IV access and positioning. The increased anesthetic risk for DMD patients has been documented in multiple case reports and series, though few large studies are available. DMD patients are at risk for rhabdomyolysis leading to hyperkalemic cardiac arrest. These cases have mostly occurred after anesthetics that include volatiles or succinylcholine, but can also occur after non-triggering anesthetics. The timing of onset is variable and can occur post-operatively from previously uneventful anesthetics and prolonged resuscitation is usually required. Dystrophic must in DMD patients has abnormal acetylcholine receptors, leading to hyperkalemia if succinylcholine is used. Volatile anesthetics cause further destabilization of an already unstable muscle membrane, leading to rhabdomyolysis. These risks are highest early in the course of the disease because there is still sufficient regenerating muscle mass to cause significant hyperkalemia, whereas later in the disease muscle mass is insufficient due to wasting. This rhabdomyolysis is not a manifestation of MH and these patients are not considered to be at increased risk of MH. Patients with DMD are also at increased risk of intraoperative cardiac and respiratory failure, especially late in the course of the disease when they have baseline cardiomyopathy and respiratory defects. DMD patients may have difficult airways due to contractures, tongue hypertrophy and aspiration risk. They also have abnormal responses to non-depolarizing neuromuscular blocking agents, including increased sensitivity, faster onset and prolonged duration. They are at risk for having increased blood loss during scoliosis surgeries due to abnormal blood vessels, osteopenic bone and impaired hemostatic function.

These anesthetic risks must be addressed in both the intraoperative and post-operative plan for anesthetic care. General anesthesia is contraindicated for non-essential procedures, especially in advanced stages of DMD. The intraoperative anesthetic plan must address needs for potential difficult airway management, cardiac resuscitation and volume resuscitation in the setting of large blood loss. The potential need for additional monitors and IV access must be addressed prior to beginning the procedure. The feasibility of alternative anesthetic plans, including sedation or regional anesthesia should be addressed. Post-operatively, the patient may require additional cardiac monitoring and respiratory support, and should be sent to a location capable of providing that level of care. The increased risk of anesthesia in these patients must be addressed preoperatively and advanced directive status should be determined prior to administering anesthesia.

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