A Duchenne Muscular Dystrophy Patient with Severe Dilated Cardiomyopathy and Respiratory Failure needing a PEG-tube

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Objectives:
- Discuss preoperative evaluation, preparation and anesthetic implications associated with muscular dystrophy
- Recognize the anesthetic implications associated with cardiomyopathies
- Describe appropriate consultation and perioperative care for a child with history of muscle dystrophy scheduled for a percutaneous gastrostomy tube (PEG tube) placement

Case history:
A 23 year old, 60 kg male with end-stage Duchenne muscular dystrophy (DMD) presented for an endoscopic percutaneous gastrostomy tube (PEG tube) placement. He has suffered from progressive swallowing difficulties leading to malnutrition prompting this procedure. He is wheelchair bound and totally immobile.

Questions:
What is Duchenne muscular dystrophy (DMD)? Are there other muscular dystrophies we need to worry about? What is the natural progress of DMD? What is the usual cause of death? What are the anesthetic implications of DMD? Does DMD carrier status in females matter? How should we continue?

Case history and physical examination (continued):
His DMD had progressed to severe muscular weakness with no ability to move, severe dilated cardiomyopathy (DCM) along with severe restrictive lung disorder. He is BiPAP dependant at night with a wheelchair affixed mouth piece ventilator for daytime support. His medications are included aspirin, carvedilol, furosemide and esomeprazole. He has previously undergone posterior spinal fusion for scoliosis correction. Physical examination revealed a thin male sitting in his wheelchair with no signs of distress. Vital signs were within normal limits including SpO2 97% on room air. Mouth opening and neck mobility were significantly limited (Mallampati score: III). Cardiac and lung examination were normal.
Questions:
What is the typical medical management strategy for DMD patients? Do we need further information about his medical treatment? He tolerated a posterior spinal fusion for his scoliosis in the past; can’t we go directly to the operating room for a PEG tube? Or do we need more information? Should we involve other health care members in taking these decisions? If so, who would we involve and what preoperative studies would you order?

Preoperative studies:
An EKG shows normal sinus rhythm, a left bundle branch block pattern, diffuse ST-T wave changes (unchanged from before) and a QTc of 499 ms. An earlier echocardiogram (ECHO) showed severe left ventricular (LV) function depression with a shortening fraction (SF) of 11%. The on-call junior cardiology fellow called you back, telling you that the patients ECHO windows were extremely poor and he was unable to estimate the LV function. A more senior person was later able to estimate his current LV function with a SF of 19% (improvement on current medications). Chest x-ray exam was clear. Pulmonary function studies showed severe pulmonary restriction (FVC and FEV1 both 0.57 L – only 11% of expected!) and severely depressed respiratory muscle strength (maximal expiratory and inspiratory pressures were 20 cmH₂O and 18 cmH₂O respectively), usually>130 cmH₂O and 100 cmH₂O respectively. Cough Peak flow was 100L per min.

Questions:
What risks and possible complication should be disclosed to the patient (and his parents)? Would you mention death as a possible complication? How do you present these complications? Do you discuss advance directives for the patient?

Dad was wondering if anesthesia is safe for his son and if you can give him a percentage of survival rates from anesthesia in children diagnosed with his son’s disease. Would you give specific number?

What is the cardiac involvement in DMD patients? What age is the onset of cardiac dysfunction? What does an EKG in DMD patients look like? Does he have myocardial ischemia? Does an EKG in DMD patients help us predict problems (normal vs. abnormal EKG)? A medical student that is with you asks whether that QTc interval is normal. Is this a concern?

Is an ECHO typically sufficient cardiac evaluation in DMD patients? The student asks what a shortening fraction mean. How that relates to this minor procedure we are planning on doing?

Do we have alternatives to poor ECHO images? If our ECHO showed adequate images with normal LV function are we safe to proceed? Would the same be true for a major operative procedure (posterior spine fusion)? Any other studies that might help us determine whether he would be safe for major surgery?
Is there anything else needed to optimize cardiac status prior to surgery (or is that necessary?).

What is the pulmonary involvement in DMD? How is his pulmonary status? Would you like other pulmonary studies? ABG? What is his pulmonary risk of anesthesia or sedation? Is there anything we can do preoperatively to optimize his respiratory status?

Case progression:
His pulmonologist, cardiologist and palliative care physician were consulted to discuss the procedure and the patient and his family were subsequently counseled regarding the high risks involved. The patient wished to be “Do-not-resuscitate” in the event of a cardiac arrest and to avoid a tracheostomy and continuous invasive mechanical ventilation.

Questions:
So what is your anesthetic plan? Local vs. sedation (MAC) vs. general anesthesia? Do we have alternative ways of taking care of this patient? What is your plan for IV access? Do we need any special monitoring (what would you use?) Are you happy with his airway? What airway (if any!) would you use and how would you obtain it? What is the basic pathophysiology of non-ischemic DCM and your anesthetic hemodynamic goals for this patient? What would you use for anesthesia/sedation? What would you use for analgesia? Would you use ondansetron to help with PONV and aspiration prophylaxis?

The ICU team called you and mentioned that there are no beds available but they can arrange a step down ICU bed but the patient would have to go through your PACU first? Would you agree?

Intraoperative care:
The patient had an uneventful induction of anesthesia and placement of an airway when he suddenly became hypotensive with a blood pressure of 52/31 with a heart rate of 78. How do you respond? What vasoactive medications would you consider using for this patient? Phenylephrine? Are there any other medications/treatments feasible at this time? How well do DMD patients respond to routine emergency vasopressors e.g. in the settings of major surgical procedures?

Your patient’s blood pressure recovers and you allow the surgeon and the gastroenterologist to proceed with the procedure. As they are securing the PEG tube you notice that your pulse oximetry “beep” disappears. You look at your monitors and see this ECG rhythm. How do you respond? The surgeon is asking whether you want to call for help.

You succeeded in treating the arrhythmias and your patient recovers to an acceptable hemodynamic status. You allow the surgeon to quickly secure his PEG tube and finish the
procedure. How do you complete your anesthetic? How do you manage your respiratory support? Are there ways of further supporting the respiratory system postoperatively?

**Postoperative care:**
Your patient is extubated later that day in the ICU. He was hemodynamically stable, tolerating his baseline BiPAP without any problems and comfortable. Do you discuss the intra-operative problems with the patient and his family? Do you call risk management?

**Discussion:**

Duchenne muscular dystrophy (DMD) is an X-lined disorder affecting 1 in 3500 males. A mutation on the Xp21 gene impacts the normal production of a cytoskeleton protein called dystrophin which is a vital structural part of skeletal and cardiac muscle. In the absence of this protein, the muscle cell becomes fragile and eventually succumbs to fatty infiltration and fibrosis.

Clinically, the disease is characterized by progressive severe muscle weakness. It initially presents around the age of 2-5 years with delayed motor development and gait impairment, eventually rendering the patient immobile and wheelchair dependent (around the age of 11-12 years). Further more, the progressive nature of the disorder results in severe restrictive pulmonary disease, dilated cardiomyopathy and arrhythmias, multiple contractures and scoliosis. Death typically occurs in the early twenties, although with current supportive measures, patients live longer. Modern management of DMD includes high doses of steroids to maintain muscle strength; non-invasive ventilation for respiratory failure; after-load reduction (ACE-I/ARB) and beta blockage for heart failure. Several novel therapies (including gene therapy) are in the pipelines for DMD patients.

Patients with DMD are considered to be at high risk for peri-operative complications. This necessitates careful preoperative multidisciplinary planning including counseling the family and discussing advance directives. Typical anesthetic risks may be related to hyperkalemic reactions from succinylcholine, malignant hyperthermia like reactions from volatile anesthetics or aspiration from bulbar insufficiency and GERD. DMD is also accompanied by heart failure attributable to dystrophic involvement of the myocardium with eventual development of heart failure, DCM and arrhythmias. One should have a low threshold for offering DMD patients (especially those who have advanced disease and/or are undergoing large procedures) extensive intra-operative monitoring (e.g. invasive arterial blood pressures, TEE, cardiac output monitoring etc.) and hemodynamic support. The peri-operative period places DMD patients at high risk for respiratory complications, especially those with advanced disease.

In summary, DMD have several anesthetic issues that need to be considered given their high risk nature. Although, DMD prognosis has classically been bleak, there have been strides in improving quality of life and life expectancy of these patients, thus making it ever more likely that they will come to our operating rooms.
References: