Chapter 14

Muscular Dystrophy

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A 16-year-old male with muscular dystrophy presents to the operating room for laparoscopic gastrostomy tube. He uses a wheelchair and undergoes physical therapy four times weekly.

The mother has brought his continuous positive airway pressure (CPAP) machine used nightly at 7 cm H_2O.

Current medications include: prednisone, albuterol, vitamin D, and calcium supplementation.

His baseline saturation is 96% on room air.

DIAGNOSIS

What Are the Important Diagnoses to Consider?

Patients undergoing muscle biopsy present with a wide range of symptoms and no definitive diagnosis. Shapiro et al. has described a classification system for these patients based on presumptive preoperative diagnosis: myopathy or muscular dystrophy (MD), neuropathy or neuronal degeneration, metabolic or mitochondrial myopathy, seizures, cardiomyopathy, dermatomyositis. Patients with suspected diagnosis categories of myopathy or muscular dystrophy as well as metabolic or mitochondrial myopathy often have diseases that will alter anesthetic management. Special consideration to management of patients in these subsets is warranted.

PREOPERATIVE EVALUATION

How Is the Diagnosis of Muscular Dystrophy Made?

Muscle biopsy is the gold standard for diagnosis of many muscular disorders. The muscle to be biopsied is chosen based on the distribution of muscle weakness on physical exam. If weakness is generalized, often the quadriceps, specifically vastus lateralis, is chosen.

What Historical and Clinical Exam Findings Are Consistent with Muscular Dystrophy?

Muscular dystrophies are quite a heterogeneous group of disorders; however, most children present under the age of 4 years for muscle biopsy. Children with more severe forms of muscular diseases will present earlier in life.

Muscular dystrophy is characterized by painless muscle degeneration and atrophy. Any exam findings of muscular tenderness make MD less likely. Assessment of muscle bulk may be of some utility as myotonias often present early in life with muscle hypertrophy while other muscular disorders present with muscle wasting and atrophy.

What Testing Is Prudent to Narrow the Differential Diagnosis and Optimize the Patient's Medical Condition Preoperatively?

Discussion with the referring physician and surgeon may be exceedingly helpful to ascertain likely diagnostic possibilities for each patient.

Muscular disorders may be suggested based on elevations of creatine phosphokinase (CK), creatine kinase of muscle (ckMB), blood urea nitrogen (BUN), aldolase, pyruvate, and lactic acid. More recently, genetic studies have become available for many disorders. For selected patients, hematologic studies and electromyography (EMG) can be suggestive of several diagnoses.

Medical optimization prior to biopsy includes determination of cardiopulmonary involvement. Tests such as chest roentgenogram, electrocardiogram, or echocardiogram may be indicated based on history, physical exam findings, and known comorbid associations with the presumptive diagnosis.

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Table 14.1 Muscular disorders associated with cardiac disease

Muscular disorder	Cardiac findings
Duchenne muscular dystrophy	ECG: sinus tachycardia with short PR interval, tall R waves in V1, and deep Q waves in limb leads Echo: Dilated cardiomyopathy, papillary muscle dysfunction leading to mitral regurgitation
Nemaline rod muscular dystrophy	Echo: dilated cardiomyopathy

Muscular disorders associated with cardiac disease are shown in Table 14.1.

Are Airway Abnormalities Associated with Neuromuscular Disease?

Structural airway abnormalities, while uncommon, do occur in muscular disease. Scoliosis with relative rigidity of the spinal column may pose positioning and airway management challenges. Chondrodysplastic myotonia (Schwartz–Jampel Syndrome) is associated with micrognathia. Functional airway abnormalities, however, are quite common and are discussed below.

Cough clearance may be impaired due to neuromuscular weakness. The five phases of cough include irritation, inspiratory phase, glottic closure, compressive phase, and expulsive phase. Each of these phases may be affected by neuromuscular disease. Diaphragmatic (or other inspiratory muscle weakness) may impair inspiratory phase. Alternatively, glottic closure is affected by bulbar involvement as seen in motor neuron disease such as Duchenne muscular dystrophy. Similar to inspiratory phase compromise, expiratory phase compromise is related to weakness of the expiratory muscles. While expiration is usually passive, during coughing, expiration is facilitated by the rectus abdominis, abdominal obliques, and internal intercostal muscles.

Mucociliary airway clearance may be impaired in patients with chronic aspiration. Chronic deposition of mucus in the tracheobronchial tree prohibits proper clearance of new secretions.

Obstructive sleep apnea occurs with disease progression due to pharyngeal muscle weakness. Sleep study results should be reviewed if available.

INTRAOPERATIVE MANAGEMENT

What Anesthetic Options Are Available for This Patient?

Local, regional, and general anesthetics have been used. In a one-year-old patient, local anesthesia is unlikely to be tolerated. Regional anesthetics may selectively block innervation to the muscle to be biopsied. Spinal, epidural (including caudal), femoral, fascial iliaca blocks may provide anesthesia to the vastus lateralis. These may be used alone or in combination with sedation.

General anesthesia with dexmedetomidine and ketamine has been studied by Kako et al., who concluded that sedation with ketamine 1 mg/kg IV with dexmedetomidine 0.5 mcg/kg IV bolus over 10 min plus continuous infusion at that rate was safe and effective with limited cardiovascular effects.

For Which Patients Should a Non-MH Triggering Anesthetic Probably Be Chosen?

Non-MH triggering anesthetics are chosen for those patients whose preoperative suspected diagnosis makes them susceptible to malignant hyperthermia (MH). These patients often present with a creatine kinase level greater than 2 times normal (for age). Modern anesthesia machines should be prepared according to manufacturer instructions and recommendations from the Malignant Hyperthermia Association of the United States (MHAUS). Additionally, activated charcoal filters could be considered for use. Muscular conditions which have definite or likely association with malignant hyperthermia include chondrodysplastic myotonia (Schwartz–Jampel Syndrome), King-Denborough syndrome, central core disease, multiminicore disease, and Brody myopathy.

Less clear recommendations can be made for those with diseases associated with adverse muscular or metabolic reaction to anesthesia (AMRA), or anesthesia-induced rhabdomyolysis (AIR). See below for discussion.

For Patients Receiving a Non-MH Triggering Anesthetic, How Should Temperature Be Measured, and Why?

Core temperature should be measured because temperature measurement location is significantly

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associated with death if an episode of malignant hyperthermia occurs. In a retrospective study of malignant hyperthermia patients, the risk of dying from the event was 13.8 times greater with no temperature monitoring than with core temperature monitoring. Similarly, monitoring skin temperature conferred a 9.7 fold increased risk of death once a malignant hyperthermia episode occurred.

Is Succinylcholine Safe to Use for Patients with Suspected Neuromuscular Disease?

Generally, succinylcholine should be avoided in patients with suspected neuromuscular disease. Certainly, succinylcholine is not safe for patients with suspected muscle disorders that are associated with malignant hyperthermia susceptibility. Additionally, those with Duchennes or Beckers muscular dystrophy reliably develop hyperkalemia following succinylcholine. Patients with hyperkalemic periodic paralysis will experience the usual potassium release following a dose of succinylcholine; however, the rise in plasma potassium may be significant and cause delayed hyperkalemic muscle weakness.

Will the Use of Nondepolarizing Muscle Relaxants Impact the Tissue Diagnosis of the Biopsy Specimen? Are They Safe to Use in This Patient Population?

Nondepolarizing neuromuscular blockers do not impact the diagnostic yield of the muscle biopsy. Some muscle disorders, however, place the patient at risk of extreme sensitivity to such agents and prolonged weakness may ensue.

Many primary or secondary muscular disorders, (myasthenia gravis), may display significant sensitivity to these agents. Additionally, patients with myotonias may experience sustained muscular contractions following reversal of neuromuscular blockade.

POSTOPERATIVE CONSIDERATIONS

How Can Adverse Muscular or Metabolic Reaction to Anesthesia (AMRA) Be Detected Postoperatively?

Adverse reactions include any unexplained hyperkalemia with ECG changes or cardiac arrest. Muscular manifestations include myoglobinuria, and a hypermetabolic state. Pre- and intraoperative level of suspicion for AMRA based on preoperative presumptive diagnosis may guide monitoring plans.

When Is Hospital Discharge Safe?

No definitive guidelines exist regarding management and discharge timing for these patients. If no adverse muscular or metabolic reaction to anesthesia is present and airway status returns to baseline, outpatient procedures are appropriate.

Parental counseling is advised for outpatients, as the patients must return to the hospital emergently if tachypnea, dark urine, muscle pains, or elevation in temperature should occur.

Special Considerations in Duchenne Muscular Dystrophy

Duchenne muscular dystrophy (DMD) occurs in approximately 1 in 5000 live (male) births. It results from a paucity of the protein dystrophin. Dystrophin is located adjacent to the sarcolemmal membrane in myocytes and connects the extracellular matrix with the intracellular contractile apparatus. Muscle biopsy demonstrates lack of dystrophin immunostaining. Genetic mutation analysis may also be diagnostic. Affected patients (males) develop muscular weakness by age two to three years. Nearly all patients are symptomatic by age five years.

Adult patients with advanced disease may continue to present to pediatric hospitals throughout their lives, although the Muscular Dystrophy Association (MDA) center offers help to transition pediatric patients into adult care.

What Preoperative Cardiac Considerations Are Necessary in Patients with Duchenne Muscular Dystrophy?

Sedentary lifestyle and inability to gather historical information on exercise tolerance warrants cardiac evaluation prior to elective anesthetics. Further, wheelchair user status puts little stress on the heart; consequently, even those with dilated cardiomyopathy may be asymptomatic. The incidence of cardiomyopathy is 25% by age six and 59% by age ten. By age 30, 90% have cardiac involvement.

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ECG changes listed in Table 14.1 are often present regardless of cardiomyopathy status (Thrush 2009). Additionally, in this study, patients' ages ranged from 3 to 27 years. Interpretation and interplay of QT and QTc varies over heart rate range and actual QT diverges from QTc with increasing heart rate. This suggests a possible small margin of safety for young children whose heart rates are faster at baseline. Dilated cardiomyopathy is common and treated with afterload reduction. Use of angiotensin converting enzyme inhibitors may predispose to post-induction hypotension. One fourth of these patients take chronic steroid therapy (even after full time wheelchair use), so adrenal suppression should be suspected (Wagner 2007).

Volatile anesthetic use. Boys with Duchenne or Becker MD are at risk for life-threatening hyperkalemia and rhabdomyolysis under general anesthesia. Although the reaction is not specifically MH, it can be equally lethal. Names for this type of reaction to inhaled anesthetics include Anesthesia Induced Rhabdomyolysis (AIR) and Adverse Muscular or Metabolic Reaction to Anesthesia (AMRA).

Previously, patients with DMD or Becker MD were often found to have positive caffeine-halothane contracture tests. For this reason, non-triggering anesthetics were recommended. Later, data from Europe suggested that dystrophin deficiency did not predispose to development of MH and that the caffeine-halothane contracture tests are unreliable in these patients. Presently, it is recognized that the genetic basis for either of DMD or Becker MD is x-linked while genetic predisposition to MH is autosomal (chromosome 19), making a genetic association between the two less likely.

In a recent retrospective chart review of 117 anesthetic exposures, the authors of the study concluded that although symptoms of leakage of muscle cell contents and resulting hyperkalemia or acidosis may be associated with volatile anesthetic exposure, severe intraoperative events also co-existed in each case. For example, one patient in the series developed metabolic acidosis but had also had 2.5 liter blood loss. As volatile anesthetic agents had not been implicated in these adverse events, the authors cannot definitively recommend against the use of volatile agents in children with Duchenne or Becker MD.

Suggested Reading

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