Lesson 236: PreAnesthetic Assessment of the Pregnant Patient With Myotonic Dystrophy

WRITTEN BY:
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Mount Sinai School of Medicine in New York City. She is the author of Curschmann-Steinert myotonia syndrome. It is a rare disease (prevalence, 1/8,000) that is seldom associated with pregnancy because affected patients often have hypogonadism. DM is a hereditary neuroendocrine, degenerative dystrophy with autosomal dominant transmission. When associated with pregnancy, the myotony of DM is often aggravated, leading to obstetric complications such as fetal loss, preterm delivery, hydrops, death in utero, difficulties in expulsion, hemorrhage during delivery, and anesthetic accidents.

Because DM affects smooth muscle in addition to cardiac and skeletal muscle, it is not unusual to find an increased incidence of uterine inertia, retained placenta, and postpartum hemorrhage. The key to prenatal and preanesthetic management is an awareness of the potential complications of pregnancy and delivery, along with a multidisciplinary approach to the disorder.2,3 Decisions regarding delivery and its timing must be collaborative between the obstetrician, neonatologist, and neurologist.

Classification
Myotonic dystrophy is the most common of the myotonic syndromes. Unlike the other myotonic disorders, DM is a multisystem disease associated with muscle wasting. Children who are symptomatic from infancy typically have a more severe form of the disease in adulthood than do those in whom symptoms appear later in childhood. Patients who have been symptomatic since infancy are often considered to have a clinically distinguishable disease referred to as congenital myotonic dystrophy.

Myotonia comprises several different types of disorders, of which DM is the most common. Less common types of myotonia include the following:

• myotonia congenita, which presents as impaired swallowing due to an inability of the oropharyngeal muscles to relax. There is no long-term weakness or cardiac involvement, and the patient's life span is normal.
• paramyotonia congenita, an extremely rare disorder presenting as stiffness and weakness after exposure to cold temperatures.

Pathogenesis and Molecular Pathology
The suspected pathogenesis of DM involves an increased muscle sodium conductance or an impaired calcium reuptake into the sarcoplasmic reticulum. The molecular defect in DM involves the amplification of a guanine–cytosine repeat in the DM-DMP1 gene.

PREANESTHETIC ASSESSMENT
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A COURSE OF STUDY FOR AMA/PRA CATEGORY 1 CREDIT
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cytosine-thymine (GCT) nucleotide triplet upstream from the gene for the DM kinase protein. This kinase normally limits the intracellular sodium current. Dysfunction of DM kinase results in a greater sodium current and altered muscle excitability. This is potentially the etiology of sudden—presumably tachyarrhythmic—death.

The defective gene of DM has been discovered on the long arm of chromosome 19 and is characterized by enlarged DNA fragments (GCT repeats) that are correlated with disease severity. The number of GCT trinucleotides on normal chromosomes ranges from 3 to 30; patients with DM show amplifications of >45 repeats. Meanwhile, molecular genetic testing of GCT expansions has become the most important diagnostic tool for confirming DM.8

Although DM is inherited in an autosomal dominant fashion, symptomatic infants are more likely to have inherited the disease from their mother. Each successive affected generation has an increased number of GCT triplets, and a more severe form of the disease.

**Signs and Symptoms**

Myotonic dystrophy is characterized by a progressive muscular dystrophy with muscle weakness and myotonia that can affect both mother and child.9 It manifests as persistent skeletal muscle contractions with voluntary or mechanical stimulation. Succinylcholine—and even neostigmine—may produce exaggerated contractures that make intubation and ventilation difficult. Once contractions occur, patients may be resistant to the effects of nondepolarizing muscle relaxants, regional anesthetics, and higher levels of general anesthetics. DM is a multisystem disorder involving several organs in addition to skeletal muscle. The following sections include descriptions of some of the more common manifestations of DM.

**Head, Neck, and Airway**

Premature frontal baldness and muscle wasting leading to hollowed cheeks and temporal fossae are common. Facial weakness causes an “expressionless” face. Extracocular muscle involvement is a consistent finding. Cataracts are not unusual. Neonates with congenital DM can have facial diplegia with a tent-shaped mouth.8

**Chest**

Bulbar weakness may lead to recurrent aspiration pneumonia. Involvement of the diaphragm and intercostal muscles may decrease the ability to cough and cause atelectasis and hypventilation. Patients may have central or obstructive sleep apnea. Chronic hypoxia and hypercarbia may be associated with cor pulmonale, hypercarbia, and diminished ventilatory responsiveness. Neonates with congenital DM may have profound weakness of respiratory muscles, requiring mechanical ventilation. Prolonged neonatal mechanical ventilation was previously thought to indicate a fatal outcome, but 2 recent cases of survivors have been reported.7

**Cardiovascular System**

Patients may present with cardiac symptoms. In 90% of patients, conduction abnormalities, secondary to degeneration of the conduction system, result in dysrhythmias and conduction blocks. First-degree heart block and intraventricular conduction delays are the most common findings. Patients may have left axis deviation and changes in ST-T waves. Sudden death has been associated with the development of third-degree atrioventricular block, but it can also occur in the presence of a functioning pacemaker, suggesting a ventricular tachydysrhythmia. Cardiac failure due to renal failure and fibrosis may occur. Impaired ventricular function is usually a late finding. Cardiomyopathy may be present, but cardiac failure is rare. The incidence of mitral valve prolapse is increased 20%. There is little correlation between the severity of the cardiac disease and the severity of the muscle disease.8

**Neuromuscular System**

Myotonia, the hallmark finding, refers to a delayed relaxation of contracted muscles. Examples include the inability to release a handshake (action myotonia) and sustained contraction on direct tapping or stimulation of a tendon reflex (percussion myotonia)—best elicited by tapping the thenar eminence or finger extensors. The facial, neck, and distal musculature is primarily involved, with preservation of limb-girdle strength until late in the disease. The myotonia of DM, unlike myotonia congenita, worsens with exercise. Muscle weakness and atrophy occur as the disease progresses. There may be mild mental retardation. Patients with congenital DM are hypotonic at birth. Muscle tone and strength then improve over the first years of life, but the disease inexorably progresses to the adult form during the first decade. In apparently unaffected siblings, myotonia can be detected by electromyography. An audible electromyogram sounds somewhat like the buzz from a propeller-driven dive-bomber.

**Orthopedic Problems**

Clubfoot deformity is common in young children with symmetric DM. Neonates with congenital DM may present with arthrogryposis.

**Gastrointestinal and Genitourinary Systems**

Dysphagia and reduced peristalsis are commonly seen in DM. Neonates with congenital DM may have such poor sucking and swallowing abilities that they require feeding through a nasogastric tube. Intestinal pseudo-obstruction and spontaneous pneumoperitoneum have been reported. Gadalna atrophy with infertility may occur.

**Other Features**

Myotonic dystrophy is associated with premature labor, uterine atony, and postpartum hemorrhage. Pregnancy may exacerbate muscle symptoms. Endocrine dysfunction (pancreatic, adrenal, thyroid, or gonadal insufficiency) should be suspected. Glucose metabolism is altered, secondary to hyperinsulinism that is often associated with real diabetes.1 The incidence of colloid goiters is increased in patients with DM. It is apparent that DM is a multisystem disorder involving several organs in addition to skeletal muscle. In the noncongenital form, DM presents at about the age of 18, but it may go unrecognized for a long time because of its insidious nature. Men with this disorder often have gonadal atrophy and are infertile, whereas affected women are able to become pregnant. When patients are pregnant, their myopathy and myotonia may become more severe, with a progressive worsening until delivery. Symptoms usually begin to abate during the puerperium. Complications of pregnancy and delivery may be the first symptoms of the disorder. In fact, a sudden deterioration in muscular strength is common at about 28 weeks of gestation and has been linked to an effect of increased levels of circulating progesterone on the cell membrane potential.10 The increased progesterone levels after the 22nd to 24th week of pregnancy affect the ratio of intracellular to extracellular potassium, the voltage-gated and calcium-activated potassium channels, and depolarization of the membrane potential. Furthermore, the modulation of potassium channels may be part of the mechanism that generates myotonia in patients with DM.11

Myotonic crisis is a severe form of deterioration. It can present as a symptom of painful dystonic postures of the hands and feet associated with autonomic dysfunction of serum creatinine and phosphokinase levels. Therapy with acetylcysteine may partially relieve the pain associated with dystonic postures.12 Previous studies have concluded that the number of obliterative complications—including fetal and neonatal loss, premature delivery, and abnormal labor—has increased in pregnant women with DM.13 There is a marked increase in polyhydramnios in these pregnancies, with affected fetuses. The high frequency of gastrointestinal symptomatology in patients with DM suggests a primary involvement of smooth muscle that is also likely to affect the genitourinary tract of female patients. Failure to progress in the first or second stage of labor, often in combination with fetal distress, has been documented in 7% of all pregnancies; most of these were associated with an affected fetus. Shore and MacLachlan14 have reported poor uterine contractions in 5 of every 8 pregnancies complicated by DM. Uncoordinated contraction of the myometrium in a nonpregnant woman was documented by the same authors. Other case reports have suggested an increased rate of abnormal placental evacuation causing massive hemorrhage and preterm labor in these patients.15

**Perioperative Considerations**

The goals of the perioperative management of a patient with DM may be divided into preoperative, intraoperative, and postoperative actions.

**Preoperative Management**

Preoperative goals are outlined below.

1. Assess the severity of the disease (including respiratory and cardiac involvement). Respiratory muscle involvement should always be suspected with muscle weakness and hypotonia. Pulmonary reserve may be assessed clinically by questions about dyspnea and activity level. Pulmonary function tests, including arterial blood gases, are indicated if significant dyspnea on exertion is present. Pulmonary function testing can show restrictive ventilatory defects with reductions in functional residual capacity, vital capacity, and total lung capacity. Findings may include mild arterial hypoxemia and diminished ventilatory responses to hypoxia and hypercarbia. Cardiac abnormalities may manifest as arrhythmias, mitral valve prolapse, or cardiomyopathy. A 12-lead electrocardiogram is helpful in excluding conduction abnormalities. A chest radiograph can evaluate inspiratory effort, the pulmonary parenchyma, and cardiac size; gastric distention secondary to smooth muscle or autonomic dysfunction may also be evident. Preoperative laboratory evaluations should include a metabolic cause of muscle weakness with measurement of serum sodium, potassium, magnesium, calcium, and phosphate concentrations. Similarly, the presence of thyrotoxicosis, adrenal, and ptilitary disorders should be excluded. A measurement of the plasma creatinine kinase may not be helpful, but very high levels (10 times normal) generally suggest a muscular dystrophy or polymyositis. Electromyography shows high-voltage, fibrillary bursts.

2. Take precautions to prevent aspiration. An increased risk for pulmonary aspiration is suggested by a history of dysphagia, perpiration, recurrent infections, or abdominal distention (due to gastrointestinal hypomotility, impaired cough, and pharyngeal muscle weakness).

3. Choose appropriate anesthetic therapy. Anxiolytics should be used with caution, if at all, because of the likelihood of respiratory depression. Severe bronchospasm may be dominated by sensitivity to preanesthetic drugs. Premedication must not have depressive characteristics. Patients with DM are sensitive to the respiratory depressant effects of sedatives (opioids, barbiturates, benzodiazepines). Talking to

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the patient to decrease anxiety will be helpful. The administra-
tion of premedications should be avoided, if possible.

**Intraoperative Management**

Seven intraoperative goals are described below.

1. Select an anesthetic that minimizes respiratory and car-
diac depression. Halothane, which depresses conduction, short-acted is to be avoided.

2. Consider the appropriate use of muscle relaxants. The depolarizing agent succinylcholine is contraindicated because it causes sustained contractions. Short-acting non-
depolarizing agents may be substituted, if necessary. Indeed, inducing muscle relaxation in a myotonic patient is one of the difficult problems facing the anesthesiologist. Anti-
cholinesterase drugs that are used to reverse nondepolariz-
ing muscle relaxants can precipitate myotonia; the use of short-acting muscle relaxants that do not require reversal has been suggested. Either atracurium or mivacurium, with-
out reversal, is an appropriate choice for relaxation. Tracheal intubation can often be performed after an I.V. induction fol-
lowed by anesthetic agents for volatile anesthetic, without the use of muscle relaxants.15

3. Select monitoring. The electrocardiogram should be monitored for conduction abnormalities—present in 90% of pa-
tients. Conduction abnormalities are usually first-degree heart block or intraventricular conduction delays. Patients may have episodes of ST-T changes that can be mistaken for ischemia. The development of third-degree heart block has been associated with asystole. Patients may be at risk for ventricular arrhythmias. Ventricular function usual-
ly remains normal until late in the disease. Patients with mitral valve prolapse and regurgitation require periopera-
tive antibiotics as prophylaxis. Hypothermia should be avoided because shivering can cause contrac-
tions. In addition to cold temperatures, many anesthetic and surgical manipulations can induce myotonic contrac-
tions. The patient’s body temperature should be main-
tained as close to normal as possible. Even pain from the I.V. injection of propofol may cause myotonic con-
tructions.16 It is also necessary to consider the muscu-
lar dystrophy a “predisposing” factor for malignant hyper-
thermia, although the exact relationship is unclear.

Interestingly, both disorders (DM and malignant hyperther-
mia) map to chromosome 19, albeit in different locations. Results of in vitro contracture testing for malignant hyper-
thermia may be positive, but it has been suggested that results of in vitro muscle testing may not be accurate in patients with neuromuscular diseases.17

4. Choose regional anesthesia techniques whenever possible. Epidural anesthesia is preferred but may still pre-
cipitate contractions because of associated shivering.18 Be-
bcause tracheal intubation may become necessary, the cervical mobility must be evaluated; this is sometimes lim-
ited by muscular retractions.

5. Recognize and treat myotonic contractions. Neither regional anesthesia nor muscle relaxants prevent or reverse myotonic contractions. Drugs that have been used to attenuate contractions include guaifenesin, procainamide, diphenhydramine, volatile anesthetics, and steroids. When all else fails, infiltration of the muscle with a local anesthetic has been recommended. Succinylcholine can induce sustained contractions of the chest wall muscles, making positive pressure ventilation difficult even in an intubated patient.19 Succinylcholine may cause tonic con-
tructions in infants with congenital DM by which muscle function is normal. Pre-
operatively, she received 2 units of packed red blood cells and was hydrated with I.V. crystalloid solutions. Fetal ultra-
sonography documented the period of gestation to be approx-
imately 33 to 34 weeks. The fetal echocardiogram showed 2 small ventricular septal defects, but otherwise normal cardiac anatomy and function. Magnesium sulfate was not given for preterm labor because of the presence of DM and the fact that the fetal membranes were ruptured. Group B streptococ-
cal antibiotic prophylaxis with ampicillin/azithromycin was given. Betamethasone was administered for fetal lung maturi-
ity. The uterine contractions were irregular. An epidural catheter was placed to relieve the pain of labor once cervical dilatation had reached 4 cm. An infusion of oxytocin was used to induce and maintain the uterine contractions but was later discontinued because of late decelerations in the fetal trac-
ings. The labor failed to progress, and the obstetrician opted to perform a Caesarean section. The epidural anesthesia was activated after a negative response to a test dose. No satisfac-
tory epidural level could be attained. The catheter was removed, and 1 shot of spinal analgesia (1 ml of bupiva-
caine 0.5%) was given. General anesthesia was maintained through the delivery. A baby boy (estimated weight, 30-31 weeks) was delivered who had a weight of 2.515 g, a length of 44.5 cm, and an Apgar score of 9 at 1 and 5 minutes, respectively. Total blood loss was 750 mL. The patient’s uterus was well contracted with the infusion of oxytocin after delivery. Postoperatively, her vital signs remained stable. No anesthetic complications developed. The electrocardiogram did not change from baseline postoperatively. The patient was placed in a monitored unit for 2 days of observation.

Reference


Lesson 236 continued from page 55
Lesson 236: Post-test

Select the single-letter response that most correctly answers the question or completes the sentence.

1. Which of the following statements is true regarding the genetics of myotonic dystrophy (DM)?
   a. DM is a rare disease (1/5,000) but is often associated with pregnancy.
   b. DM occurs via autosomal recessive transmission.
   c. In DM, there is amplification of the adenine–cytosine–thymine nucleotide triplet upstream from the gene for the protein DM kinase.
   d. Molecular genetic testing for guanine–cytosine–thymine expansions has become the most important diagnostic tool to confirm DM.
   
2. The clinical presentation of DM includes:
   a. progressive muscle stiffness and myoclonus
   b. persistent skeletal muscle contractions with voluntary or mechanical stimulation
   c. contractions relieved by muscle relaxants
   d. manifestations that involve only skeletal muscles

3. Which of the following statements is true regarding the cardiovascular presentation of DM?
   a. Conduction abnormalities occur in 40% of patients.
   b. Impaired ventricular function is usually an early finding.
   c. There is a 20% increased incidence of mitral valve prolapse.
   d. The severity of cardiac disease correlates with the severity of muscle disease.
   
4. Which of the following is a true statement describing characteristic neuromuscular manifestations of DM?
   a. Limb-girdle muscles are primarily involved early in the disease.
   b. The facial, neck, and distal musculature is involved late in the disease.
   c. Patients with congenital DM are spastic at birth.
   d. Myotonia worsens with exercise.

5. Which of the following is a true statement regarding the effects of pregnancy on DM?
   a. Myopathy may become more severe with pregnancy.
   b. Deterioration of muscle weakness has been linked to an effect of increased levels of circulating estrogen on the cell membrane potential.
   c. The increase in pregnancy hormones after the 22nd week of pregnancy affects the sodium chloride channels and repolarization of the membrane potential.
   d. Women with the disorder often have gonalad hypertrophy and multiple births.

6. Which of the following is a true statement about the preoperative goals of treating the patient with DM?
   a. Premedication should always be given to allay anxiety.
   b. Assess the severity of the disease by conducting computed tomography and magnetic resonance imaging of the head and chest, echocardiography, and a pulmonary function test.
   c. Take precautions to prevent aspiration.
   d. Give dantrolene as prophylaxis for malignant hyperthermia.

7. The least common findings during labor in pregnant patients with DM are:
   a. abnormal labor, neonatal loss, premature delivery
   b. failure to progress in the first or second stage of labor
   c. poor uterine contractions complicated by postpartum hemorrhage
   d. an abnormally enlarged placenta with increased calcifications

8. The use of which type of drug is contraindicated in patients with DM?
   a. a depolarizing muscle relaxant
   b. a nondepolarizing muscle relaxant
   c. an antimuscarinic agent (eg, atropine, glycopyrrolate)
   d. a volatile anesthetic

9. The best course for selecting an anesthetic regimen for labor and delivery in a pregnant patient with DM is to:
   a. choose a regional technique whenever possible
   b. avoid giving an epidural anesthetic because it may precipitate muscle contractions due to shivering
   c. add spinal morphine to all regional anesthetics
   d. control the airway with a general anesthetic

10. The treatment of patients with DM includes:
    a. physical therapy to prevent contracture
    b. quinidine, tocainide, mexiletine
    c. orthoses and corrective orthopedic surgery
    d. all of the above

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