Chapter 31

Duchenne Muscular Dystrophy

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Case Scenario

A 20-year-old male with Duchenne muscular dystrophy presents to the cardiac catheterization laboratory for automatic implantable cardioverter defibrillator insertion. He was recently evaluated for palpitations and found to have intermittent runs of nonsustained ventricular tachycardia. He follows up annually with his cardiologist. He had spine surgery at age 12 years for scoliosis, is no longer ambulatory, and uses a wheelchair. He is also followed by a pulmonologist. Pulmonary function tests 2 weeks earlier were consistent with severe restrictive lung disease with forced expiratory volume 10% predicted, forced vital capacity 8% predicted, and pCO_2 100 mm Hg. He was started on bilevel positive airway pressure respiratory support at night, which he tolerates. His current vital signs are heart rate 110 beats/minute, respiratory rate 20 breaths/minute, blood pressure 102/64, and SpO_2 93% on room air.

His last echocardiogram, performed 11 months ago, demonstrated the following:

- *Moderate-to-severe left ventricular dysfunction (ejection fraction 30%)*
- *Posterolateral akinesis*

Key Objectives

- Discuss the cardiopulmonary morbidity associated with Duchenne muscular dystrophy.
- Describe the preoperative assessment of patients with Duchenne muscular dystrophy.
- Understand important considerations for the anesthetic management of these patients.
- Evaluate various strategies for intra- and postoperative cardiac and respiratory support of patients with Duchenne muscular dystrophy.

Pathophysiology

What is Duchenne muscular dystrophy and what is the underlying pathophysiology?

Duchenne muscular dystrophy (DMD) is a progressive neuromuscular disorder that is inherited in an X-linked recessive pattern, therefore predominantly affecting the male offspring of maternal carriers. However, nearly 30% of cases are due to random mutation rather than hereditary transmission. The incidence is approximately 1:5000 to 1:3500 live births. Duchenne muscular dystrophy is caused by a mutation on Xp21 that results in the absence of dystrophin, a structural protein critical to the dystroglycan complex found in skeletal and cardiac muscle cells. This complex stabilizes the muscle cell membrane during contraction. Without the dystroglycan complex, the cellular membrane is fragile, leading to cell damage and eventual necrosis over time. The subsequent scarring with fatty infiltration and fibrosis results in progressive muscular weakness and dysfunction.

What is the typical timeline for presentation and progression of symptoms in patients with DMD?

Duchenne muscular dystrophy is characterized by progressive muscle weakness and damage that impacts both skeletal and cardiac muscle tissue. Symptoms typically manifest as clumsiness, weakness, or failure to meet gross motor milestones by 3–5 years of age. Loss of ability to ambulate usually occurs around age 12 years. Cardiac muscle damage and fibrosis lead to clinically evident cardiomyopathy in the mid-to-late teen years, though evidence of cardiac damage can be seen much earlier on cardiac magnetic resonance imaging (MRI) and electrocardiogram (ECG) evaluations. Chest wall and diaphragmatic weakness eventually result in respiratory insufficiency and the need for respiratory support in the late teens to early 20s. In the past, respiratory insufficiency and infections were the leading cause of death for patients with DMD, but now, with early noninvasive respiratory support, heart failure and arrhythmias are often life limiting.

Clinical Pearl

In the past, respiratory insufficiency and infections were the leading cause of death for patients with DMD, but now, with early noninvasive respiratory support, heart failure and arrhythmias are often life limiting.