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INTRODUCTION

Duchenne muscular dystrophy (DMD) is an X-linked recessive disorder, with a prevalence of 15.9 to 19.5 per 100,000 live male births,\(^1\) that is caused by absence or deficiency of functional dystrophin protein. Dystrophin stabilizes skeletal and cardiac muscle by connecting actin in muscle fibers to the extracellular matrix; in the absence of dystrophin, recurrent muscle fiber injury leads to chronic inflammation, replacement of muscle with fibrotic and fatty tissue, and associated weakness.\(^2\)

The progressive neuromuscular weakness is often first noted during the preschool years. Boys with DMD create accommodative patterns of movement, and independent ambulation has been prolonged with the widespread use of systemic steroids to minimize accumulation of chronic inflammation.\(^3,4\) In the current era, ambulation continues until median age 12 years,\(^5\) and adolescents typically begin noninvasive positive-pressure ventilation (bipap) instead of continuous positive airway pressure, and oxygen should never be used alone, as this can obscure and worsen hypoventilation.

The natural progression of DMD pathophysiology has changed with the introduction of therapies for downstream pathologic pathways and will continue to evolve with the development of therapies that target function and expression of dystrophin.

KEY WORDS

- Duchenne muscular dystrophy
- Polysomnogram
- Respiratory failure
- Neuromuscular disease
- Noninvasive ventilation
- Obstructive sleep apnea
- Insomnia hypoventilation

KEY POINTS

- Although sleep-disordered breathing is one of the best recognized disorders of sleep in boys and men with Duchenne muscular dystrophy (DMD), disorders of initiation and maintenance of sleep are more common and can significantly impact quality of life.
- Changes in mobility status predict need for increasing respiratory support.
- Nighttime hypoventilation precedes daytime symptoms and may be identified only on polysomnogram.
- Nighttime respiratory support should generally begin with bilevel noninvasive positive-pressure ventilation (bipap) instead of continuous positive airway pressure, and oxygen should never be used alone, as this can obscure and worsen hypoventilation.
- The natural progression of DMD pathophysiology has changed with the introduction of therapies for downstream pathologic pathways and will continue to evolve with the development of therapies that target function and expression of dystrophin.
restore function of partially functioning dystrophin protein. Although the trajectory has changed over time, individuals with DMD continue to have progressive sleep and respiratory disturbances that are best addressed in an anticipatory fashion, before accumulation of morbidity.

**SLEEP-RELATED BREATHING DISORDERS AND RESPIRATORY FAILURE**

Respiratory sufficiency, which is dependent on adequate ventilatory muscle strength, decreases as muscle weakness progresses. The decline in respiratory function and eventual failure typically parallels ambulation, with emergence of respiratory insufficiency when independent ambulation is lost. The diaphragm is the largest and most active muscle involved in breathing and thus the first to become significantly damaged and weakened in DMD.

Given the natural decrease in muscle tone and ventilatory drive during sleep, respiratory abnormalities in those with DMD tend to manifest first during sleep. Across the lifespan of DMD, various sleep-related breathing disorders (SRBD) have been described, including obstructive sleep apnea (OSA), central sleep apnea (CSA), and nocturnal hypoventilation (Fig. 1).

As respiratory complications are a major cause of morbidity and mortality in the individual with DMD, the respiratory management of the Duchenne patient, particularly the adult, should be anticipatory and preventive. Monitoring of respiratory muscle function, diurnal and nocturnal ventilation, cough, and swallowing function are foundational. Implementing a “package” of respiratory interventions at appropriate points is crucial for improvements in quality and length of life, and is discussed in detail as follows.

**Pathophysiology**

**Early ambulatory (0–10 years)**

With the emergence of widespread newborn screening and genetic testing, many children are now diagnosed with DMD in infancy. Infants with DMD have normal or near-normal respiratory physiology for age. Early ambulatory children with DMD should have typical trajectories of respiratory illnesses when compared with healthy peers and should not require additional diagnostic or therapeutic measures.

The most common SRBD during toddlerhood/early school years is OSA associated with adenotonsillar hypertrophy. OSA has a prevalence of 1% to 5% in the general population; specific rates of OSA in those with DMD during ambulatory years have not been studied. Manifestations of OSA in prepubertal childhood are different from those in older youth and adults. As opposed to overt sleepiness, younger children with OSA tend to have more difficulty with inattention and mood, manifestations of which can overlap with the comorbid attention-deficit hyperactivity disorder and/or autism present in DMD. Treatment of childhood OSA with adenotonsillectomy has been shown to help with behavior and quality of life. Although this has not been studied specifically in children with DMD, there is no indication that treating OSA in a child with DMD would not provide at least some of the same benefits.

**Late ambulatory (8–18 years)**

The use of chronic systemic steroids is commonly acknowledged as a risk factor for OSA with presumptive mechanism being the association with obesity, as well as increased fat deposits in the tongue and neck; this has not been well described.

Inhalation is an active process, whereas exhalation is passive. However, forced expiration maneuvers such as coughing are not passive, and are necessary to clear the airways of secretions or foreign matter. Although a strong, productive cough depends on muscles of expiration, it is just as dependent on first taking a large breath in: a cough with a low volume of air behind it will be less forceful. As the muscles of a youth with DMD slowly weaken, the diaphragm will maintain the ability to take small, “tidal” breaths much longer than the ability to take a large-volume inhale needed to create a cough.

Thus, decreasing inspiratory muscle strength first presents clinically with diminishing cough strength and decreased ability to clear airway secretions, before affecting ventilation. This becomes important in the setting of lower respiratory tract infections, with poor secretion clearance leading to atelectasis, VQ mismatch, hypoxemia, and increasing risk for secondary pneumonia and respiratory failure.

During times of health, late-ambulatory youth with DMD can maintain adequate minute ventilation when awake and upright, but ventilation can begin to be impaired during times of challenge (viral illnesses, sleep, supine positioning). Acute bone fractures can be associated with splinting of breathing, sedating pain medications, and changes to mobility status, which all increase the risk of hypoventilation.

**Early nonambulatory (12–20 years)**

Decreased mobility further limits airway clearance, again increasing risk of secondary pneumonia and respiratory failure with viral illnesses. Nonambulation is associated with development of scoliosis.
This typically begins as a thoracolumbar curve, and does not significantly affect respiratory status until later progression.

With progression of respiratory muscle weakness, the weakened diaphragm leaves the individual more dependent on muscles of the chest wall for respirations. This is first observed during rapid-eye movement (REM) sleep, when the diaphragm is unable to compensate for atonia of the chest wall, and hypoventilation develops. To compensate, individuals with DMD may show paradoxic breathing and tachypnea; this increased energy expenditure may prevent partial pressures of carbon dioxide \( \geq 50 \) mm Hg from being captured on capnography, as required for American Academy of Sleep Medicine diagnosis of hypoventilation, at the cost of disrupted REM sleep and failure to thrive. In this group, \( \text{PCO}_2 \geq 45 \) mm Hg can be considered abnormal.

Late nonambulatory
Patients with DMD have significant reductions in both lung and chest wall compliance. Some of this decrease in compliance may be due to atelectasis that occurs at low lung volumes. The inability to hyperinflate the lung via sigh breaths, due to muscle weakness, likely contributes to this progressive loss of compliance.

In addition to respiratory muscle weakness, progressive pharyngeal weakness and prolonged steroid exposure lead to increased rates of OSA. Dysphagia is increasingly common, and unlike in other disorders, occurs first with solid foods rather than liquids. Although clinically apparent choking can occur, it is often underrecognized and can lead to aspiration pneumonia.

Thoracolumbar scoliosis progresses to include compensatory thoracic curve with pelvic obliquity. This leads to further restrictive lung disease, hypoventilation, and risk for progressive respiratory failure with acute illness. Hypoventilation progresses to non-REM sleep and wake states.

**Assessments for Sleep-Related Breathing Disorders**

Due to the evolving respiratory pathophysiology through the lifespan with DMD, focus should vary with age:

**Physical examination and history**
- Early ambulatory: evaluate for tonsillar hypertrophy, mouth breathing, history of snoring, restless sleep, and mood or attention concerns
- Late ambulatory: history of snoring, restless sleep, mood or attention concerns, prolonged cough with respiratory illnesses, history of pneumonia
- Nonambulatory: monitor for changes in chest wall shape and compliance, symmetry of chest auscultation, reports of poor sleep and daytime sleepiness, signs and symptoms of dysphagia

**Pulmonary function testing**
Simple spirometry should be performed annually in ambulatory individuals. More extensive testing should be performed twice annually in nonambulatory individuals.

**Home pulse oximetry**
A pulse oximeter should be considered in an individual who has been prescribed cough augmentation to identify early, mild hypoxemia with illness or mucus plugging. A persistent value \( \text{SpO}_2 \) of less than 95% suggests that cough therapy and lung volume recruitment (LVR) should be instituted aggressively. Oximetry used at night is not a sensitive method for detecting the hypoventilation that is a major issue during sleep for individuals with respiratory muscle weakness.

**Polysomnography**
Polysomnogram (PSG) with continuous carbon dioxide monitoring by end-tidal or transcutaneous monitoring should be performed as follows:
- For any symptomatic individual with DMD (snoring, morning headaches, daytime sleepiness, observed apneic pauses), or asymptomatic individuals with significant weight gain or adenotonsillar hypertrophy
- Annually for nonambulatory individuals not already using nocturnal ventilation
- Preoperative PSG, if no recent study

For adult men with DMD using nocturnal ventilation, ongoing management of device settings during sleep are managed by recorded downloads from their nocturnal ventilatory devices as well as monitoring symptoms and diurnal carbon dioxide levels. A panel of clinical experts have suggested that yearly sleep studies should be performed in adults with DMD, although many adult sleep laboratories are not equipped to handle individuals with significant mobility issues, nocturnal nursing, and caregiver needs (discussed in Justin A. Fiala and John M. Coleman III’s article, “Tailoring the Sleep Laboratory for Chronic Respiratory Failure,” in this issue).
Respiratory status during acute illness
Patients should be instructed to contact their medical care team with symptoms of respiratory tract infection. In addition to history and physical examination, chest radiograph, oximetry, and assessment of Paco2 should be strongly considered. Patients and their families are often equipped to manage increased frequency of cough therapy and assisted ventilation; however, if adequate caregivers are unavailable at home or the trajectory of respiratory status is poor, admission to the hospital is appropriate for monitoring, augmented care, and family respite. Admission does not negate that family members often have better knowledge in assessment and treatment than some medical team members, as admissions overall for DMD are relatively uncommon. Use of oxygen without noninvasive ventilation (NIV) during hospitalization should be avoided given the risk of hypercarbia.

Respiratory Management During Childhood Immunizations
- All infants/children with DMD should receive standard pediatric immunizations.
- All individuals with DMD should receive annual inactivated influenza vaccination.
- All individuals with DMD should receive pneumococcal vaccination per Centers for Disease Control and Prevention guidelines.

Fig. 1. Progression of respiratory pathophysiology and care by stage of disease. (A) Sleep-related respiratory pathophysiology in patients with DMD by stage of disease. (B) Assessments and interventions for respiratory care of patients with DMD by stage of disease. MEP, maximum expiratory pressure; MIP, maximum inspiratory pressure; PCF, peak cough flow; petCO2, end-tidal partial pressure of CO2; ptcCO2, transcutaneous partial pressure of CO2; SpO2, blood oxygen saturation by pulse oximetry. See text for definitions of sleep study results. All specified threshold values of PCF, MEP, and MIP apply to older teenage and adult patients. Fatigue, dyspnea, morning or continuous headaches, frequent nocturnal awakenings or difficult arousal, hypersomnolence, difficulty concentrating, awakenings with dyspnea and tachycardia, or frequent nightmares. We strongly endorse the use of noninvasive methods of assisted ventilation instead of tracheostomy to optimize patient quality of life; indications for tracheostomy include patient preference, inability of patient to use noninvasive ventilation successfully, 3 failed extubation attempts during a critical illness despite optimum use of noninvasive ventilation and mechanically assisted coughing, or failure of noninvasive methods of cough assistance to prevent aspiration of secretions into the lungs due to weak bulbar muscles. (B) From Birnkrant, DJ, Bush KM, Bann CM, et al., Diagnosis and management of Duchenne muscular dystrophy, part 2: respiratory, cardiac, bone health, and orthopaedic management., The Lancet Neurology, 2018; volume 17: 347–361; Reprinted with permission from Elsevier.)
Control and Prevention (CDC)\(^\text{19}\) guidelines for children with underlying medical conditions. At the time of this writing, recommendations are as follows:
- PCV13 vaccine in a 4-part series before age 2 years
- PPSV23 with first dose administered at age \(\geq 2\) years (at least 8 weeks after final indicated dose of PCV13), second dose 5 years after the first
- See current guidelines for details

Adenotonsillectomy
Adenotonsillectomy should be considered in ambulatory children with OSA and in nonambulatory individuals with identified SRBD and adenotonsillar hypertrophy.

Cough augmentation
Diminishing percent-predicted forced vital capacity (FVC) or maximal inspiratory/expiratory pressures may correlate with decreasing efficacy of spontaneous airway clearance, as does prolonged cough or hypoxemia with viral illnesses. It benefits these patients to have equipment available at home before first severe respiratory exacerbation, so an anticipatory philosophy toward prescription of therapies is recommended. Cough augmentation is initiated when the patient’s peak cough expiratory flow falls below 270 LPM.\(^\text{20,21}\) There are 3 ways of increasing the cough peak flow: \(^\text{22}\)

- Mechanical insufflator-exsufflator (MI-E): this device, colloquially referred to as “cough assist,” is commonly prescribed in pediatric settings. It is an artificial cough generator used with a mask (or, later, tracheostomy adapter) that moves a volume of air into the subject’s lungs via positive pressure, and then creates airflow out of the lung, “sucking” out secretions in the process, via rapid switch to a negative pressure. This is most effective with patients who can cooperate to keep the glottis and upper airway open to allow airflow out of the lung.
- Manually assisted cough (MAC): a trained assistant applies a thrust to the upper abdomen or lower ribcage to increase transpulmonary pressure during exhalation, thus increasing flow.
- LVR or “breath stacking”: can be used either alone or in combination with MAC to increase flow. By increasing the volume of the lung before cough, there is a greater volume of air to exhale, as well as increased lung elastic recoil at greater lung volumes (see section on LVR in adult care).

Nighttime positive-pressure ventilation
Although the physiology of sleep-disordered breathing varies, it is generally recommended to begin nighttime respiratory support with bilevel pressure. Although continuous positive airway pressure (CPAP) may provide adequate initial support for OSA alone, CPAP may overpower respiratory muscle strength and worsen hypoventilation as weakness progresses. From a practical standpoint, initiation with bilevel support prevents the necessity of frequent follow-up polysomnograms as well as the need to upgrade the device. Oxygen should never be used in isolation to treat SRBD in DMD, as this can mask and worsen hypoventilation.\(^\text{23}\)

Although there are times that NIPPV is started in the setting of an acute illness while hospitalized, NIPPV is best initiated in the outpatient setting. Ideally, NIPPV will have been discussed as part of anticipatory guidance before imminent need. Allowing youth with DMD to see, feel, and try different styles of mask interfaces before PSG may provide a better first-time positive airway pressure (PAP) experience, which may affect long-term adherence to prescribed PAP therapy. Mask selection depends on age, craniofacial structure including obesity, ability to self-adjust mask, but mostly patient preference and comfort. Nasal masks including pillows are often preferred for decreased risk of aerophagia. Split-night or titration PSG can be used to help initiate and/or fine tune settings, although this is not absolutely necessary. Once PAP treatment is begun, follow-up within 1 to 2 months is important to identify barriers to use as well as adequacy of treatment, both clinically and often for insurance coverage. Subsequent follow-up frequency is related to adherence, tolerance, subjective benefit, and overall clinical trajectory. Remote downloads and telemedicine can be useful adjuncts or even substitutes to in-person follow-ups, particularly with the explosion of telemedicine platforms related to the Coronavirus Disease 2019 pandemic.

Transition from Pediatric to Adult Care
Medical care for pediatric patients has become much more sophisticated over the past several decades, resulting in increased survival of high-technology needs individuals into adulthood. Transition has been defined by one investigator as “the purposeful, planned movement of adolescents with chronic medical conditions from child-centered to adult-oriented health care.”\(^\text{24}\) Transition usually occurs between the ages 17 and 21, depending on the health care system.
Components of transition programs necessary for a successful transition in other complex diseases have been identified. These include the following:

- Disease-specific patient education including family self-management
- Creation of specific adolescent or joint pediatric/adult overlap clinics
- Creation of a transition-coordinator position with links to both programs

A list of important areas for consideration during transition specific to the young man with DMD is seen in Fig. 2. Poorly planned transition can result in a deterioration of health status, a loss of confidence in the adult health care system due to perceived ignorance of pertinent diseases by adult providers, and a limited ability of some patients to direct their own health care in the adult environment.

Respiratory Management During Adulthood

Nighttime positive-pressure ventilation

Nearly all individuals with DMD will be on nocturnal ventilatory support before transition to adult care.

Cough augmentation

By the time the individual has transitioned to the adult clinic, cough augmentation strategies will have been in place already, as discussed previously.

Lung volume recruitment

Hyperinflation of the lung via LVR may reverse progressive loss of lung and chest wall compliance in the individual with DMD, provide short-term and long-term improvements in lung compliance and vital capacity (VC), and can enhance cough peak flow for secretion clearance.

LVR should be started when the FVC falls below 60% predicted and be performed 2 to 3 times per day.

Fig. 2. Social and medical considerations during transition to adulthood for young men with DMD. (From Birnkrant DJ, Bushby KM, Bann CM, et al., Diagnosis and management of Duchenne muscular dystrophy, part 3: primary care, emergency management, psychosocial care, and transitions of care across the lifespan., The Lancet Neurology, 2018; volume 17: 445-55; with permission.)
LVR, or “breath stacking,” is an active assisted inhalation maneuver wherein the individual with DMD increases his lung volume above what he is able to achieve on his own by taking in extra positive-pressure breaths. This larger, assisted lung volume has been referred to as the maximum inflation capacity or lung inflation capacity. The maneuver can be performed with a manual resuscitator bag attached to a mouthpiece, with an MI-E device, or with a mouthpiece ventilator in a volume mode, by taking multiple breaths without exhaling. The maneuver itself is relatively easily learned by the individual with DMD and is often very effective unless significant oral weakness leading to leak of volume is present.

**Daytime noninvasive intermittent mechanical ventilation (“sip and puff”)**

Mouthpiece ventilation (MPV), also known as “sip and puff” ventilation, is a method using an open mouthpiece system that allows a breath to be delivered on demand when the patient “sips” from the mouthpiece interface. This requires a home ventilator that can deliver breaths in the volume mode. Modern ventilators can attach to the patient’s wheelchair, and the mouthpiece interface is held in place by a flexible mechanical arm that can be adjusted to keep the mouthpiece stable in the proper position.

Patients who exhibit the need for ventilation during the daytime are candidates for MPV. Symptoms and signs that suggest that need include (1) dyspnea during the daytime, (2) elevated daytime CO₂ levels despite adequate treatment of sleep-disordered breathing, and (3) inadequate ability to phonate because of lack of breath volume. In addition, MPV allows for frequent user-controlled breath-stacking maneuvers that can be very helpful in increasing patient independence.

**Tracheostomy/invasive mechanical ventilation**

The need for and timing of tracheostomy is a debated topic, but 24 hours per day noninvasive ventilation for individuals with DMD is entirely possible. At our institution, we strongly support the use of NIPPV for 24 hours per day, via mask ventilation at night and MPV during the day. This avoids the tracheostomy procedure, the increased secretions caused by a tracheostomy tube, and allows the patient a greater ability for natural speech and swallowing. Some have suggested that a patient requiring more than 16 hours per day of noninvasive ventilation should be considered for tracheostomy.

A recent consensus statement suggested the following criteria for considering tracheostomy:

- Three failed extubation attempts following intubation for a critical illness
- Patient preference
- Inability of the patient to use NIPPV successfully
- Presence of weak bulbar muscles causing failure of safe secretion management

**OTHER SLEEP DISORDERS**

In addition to direct benefits, treating these generalized sleep complaints is important in initiating and maintaining NIV in patients with DMD.

**Insomnia**

**Prevalence**

One cross-sectional study showed that insomnia is extremely common in boys and adolescents with DMD, with 30% of respondents screening positive for disorders of initiating or maintaining sleep.

**Etiology**

Boys and men with DMD have multiple risk factors for insomnia, including the following:

- Medications: commonly prescribed medications that can disrupt sleep in this population include systemic steroids and antidepressants.
- Psychological and socioeconomic stressors.
Sleep hygiene: Video games and online gaming are acknowledged by advocacy groups, such as Parent Project Muscular Dystrophy, as inclusive recreation that is popular for boys and men with DMD. While allowing for recreation and socializing, late-night and binged gaming sessions can contribute to poor sleep patterns if good sleep hygiene is not promoted.

Pain: Most boys with DMD and their caregivers report chronic pain. Pain is more common in nonambulatory individuals, possibly related to inability to independently shift body position requiring the caregiver to do so, or associated with muscle contractions and positioning device (AFOs).

SRBD: unrecognized or undertreated SRBD can result in sleep onset and sleep maintenance insomnia.

**Intervention**

As with the general population, a detailed history of sleep patterns and environment will often yield potential areas for intervention. Promoting good sleep hygiene is important regardless of the presence of an organic etiology for sleep disturbance; in particular, regular sleep-wake cycles and limits to nighttime screen usage are important to recommend. Ensuring that the sleep environment that is conducive to restfulness includes addressing chronic pain and nighttime caregiver availability.

Treating SRBD may promote more continuous and restful sleep. For medication-related sleep disturbances, consider alternatives to prescribed medications that can cause insomnia and recommend dosing systemic steroids in the morning.

For more challenging or multifactorial cases of insomnia, melatonin may help promote regular sleep cycles when combined with sleep hygiene practices; due to the potential to suppress respiratory drive, hypnotics/medications with sedative effects should be avoided when possible. Cognitive Behavioral Therapy for Insomnia can also be a useful therapy and may benefit those with DMD as well as their caregivers.

**Abnormal Autonomic Regulation**

Symptoms of excess sympathetic tone, including tachycardia, reduced heart rate variability, and hyperhidrosis, are commonly seen in individuals with DMD. In addition to physical discomfort that may interfere with sleep, abnormal autonomic tone may potentially contribute to psychological symptoms, such as depression.

**Restless Leg Syndrome**

Immobility, muscle contractures, and postural equipment (orthotics) can be associated with extremity pain and discomfort. In this setting, symptoms of restless leg syndrome (RLS) may be overlooked. An individual with underlying propensity for RLS may have symptoms exacerbated by other medications prescribed for neuromuscular pains or depression (tricyclic antidepressants, selective serotonin reuptake inhibitor, serotonin-norepinephrine reuptake inhibitors).

**Palliative/End-of-Life Care**

With the advent of noninvasive ventilation and advances in medications for the treatment of cardiomyopathy, individuals with DMD are currently living into their fourth and fifth decades. This is new territory for individuals with DMD and has been termed “unexpected adulthood.” As men with DMD age into their 40s and even 50s, the issue of losing parental or other family caregivers due to death arises. Therefore, advanced planning for both advanced care and end-of-life (EOL) issues is crucial. Unfortunately, it is difficult to determine when EOL is near in individuals with DMD.

One group has proposed the following indicators as indicating the approach of EOL in patients with neuromuscular diseases: a marked decline in pulmonary function, particularly FVC and peak cough flow, marked weight loss, recurrent infections (typically pulmonary or urinary), inability to heal wounds/pressure ulcers, swallowing problems, and cognitive decline. These predictors have not been studied in DMD and may be more appropriate for other diseases, such as amyotrophic lateral sclerosis. Ventilatory failure can generally be well controlled in patients with DMD, and therefore end-stage cardiomyopathy may be an important marker of EOL in DMD.

EOL issues and questions are best addressed through regular and open communication among the patient, their family/caregivers, and members of the health care team in advance of emergency situations. Patients and/or substitute decision-makers should be informed of the changes in respiratory function that can be expected over time, and the potential options available to address these changes. Rather than asking them to simply choose from a menu of options, a shared decision-making model is advocated when the right course is uncertain; health care providers should make recommendations after eliciting the values and preferences of decision-makers. Unfortunately, these discussions rarely happen. In a recent qualitative interview study of 15 young men with DMD...
in Britain, none of the men could recall any discussion about EOL with any clinician while on pediatric or adult services.\textsuperscript{50} Too often, health care providers and caregivers of patients with DMD are unprepared for EOL, preventing death from happening in the location and manner desired by the patient or the patient’s family. It may be helpful to engage a hospital-based or clinic-based palliative care service if providers are uncomfortable approaching these subjects with their patients.

Dyspnea is a common and distressing symptom at the EOL, and NIV can play a substantial role in palliation of dyspnea for patients with DMD and others with advanced ventilatory failure.\textsuperscript{51} Most individuals with DMD will already have been using NIV before this point in their disease trajectory and so it is natural, and very appropriate, to continue during EOL care with the purpose of relieving dyspnea as opposed to prolonging care. Clear communication with hospice or home nursing services is important to make sure that NIV is not stopped abruptly and that transition to using dyspnea-relieving medications is gradual and with input from the patient and their caregivers.

**SUMMARY**

The natural progression of DMD pathophysiology has changed with the introduction of therapies for downstream pathologic pathways and will continue to evolve with the development of therapies that target function and expression of dystrophin. However, the respiratory and sleep needs in this population are largely predictable over the lifespan, and are best handled in an anticipatory manner. The profile of SRBD changes through childhood and adolescence due to progressive respiratory pathology. Boys treated with systemic steroids develop increased rates of OSA, and progression of chest wall weakness leads to hypoventilation during sleep before daytime symptoms and may only be identified on polysomnogram. Nighttime respiratory support should generally begin with bilevel NIPPV instead of CPAP, and oxygen should never be used alone, as this can obscure and worsen hypoventilation. Although less commonly recognized than SRBD, boys and men with DMD are at increased risk for disorders of initiation and maintenance of sleep, caused by SRBD, anxiety/depression, autonomic dysregulation, and as a side effect from commonly prescribed medications.

**CLINICS CARE POINTS**

- Decline in respiratory function typically parallels decline in ambulation.
- Intermittent nighttime hypoventilation precedes daytime symptoms and may be identified only on polysomnogram.
- Oxygen should never be used alone for acute illness or sleep disordered breathing, as this can mask or worsen hypoventilation.
- Boys and men with DMD benefit from anticipatory guidance with regard to respiratory disturbances and support, transition from pediatric to adult care, and transition to end-of-life care.

**DISCLOSURE**

Dr E.W. MacKintosh has no relevant disclosures.

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