Respiratory Care of the Patient with Duchenne Muscular Dystrophy

ATS Consensus Statement

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TABLE OF CONTENTS

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BACKGROUND

Duchenne-type muscular dystrophy (DMD) is a disease characterized by progressive loss of muscle strength, eventually resulting in loss of ambulation, loss of respiratory muscle strength, and death from respiratory insufficiency. The majority of patients develop cardiomyopathy. DMD is an X-linked recessive trait that occurs almost exclusively in boys. The incidence of DMD is approximately 1:3,000 male births and is caused by mutation of the dystrophin gene. Clinical diagnosis is made after consideration of history, physical findings, and elevated serum creatine kinase level. Diagnosis is confirmed by finding an abnormality in the dystrophin gene by mutation analysis of blood leukocyte DNA. If DNA analysis is normal (as is the case in 1/3 of patients), diagnosis should be confirmed by finding absent or abnormal dystrophin using immunohistology or protein analysis of muscle tissue.

Although respiratory disease in DMD is its major cause of morbidity and mortality, there is inadequate awareness of its *treatable* nature. Recent advances in the respiratory care of the DMD patient have improved the outlook for these patients, and many caregivers have changed from a traditional non-interventional approach to a more aggressive, supportive approach. Despite the availability of new technologies to assist patients with DMD, many families do not receive sufficient information regarding their options in diagnosis and management of respiratory insufficiency.

PURPOSE

This statement is designed to educate the practitioner about new approaches and therapies available for the management of the respiratory complications of DMD. Many of the respiratory interventions reviewed in this statement can be adapted to the care of patients with other types of neuromuscular diseases.

METHODS

Formation of Consensus Committee

The consensus statement working group was formed May 2001 at the American Thoracic Society annual meeting. Members of the group represented experts in DMD respiratory care in institutions managing multiple patients with DMD, generally in conjunction with a Muscular Dystrophy Association–supported MD clinic. Although the majority of the members were pediatric pulmonologists, one member was a child neurologist, and one member was a nurse. American Thoracic Society sponsored consensus conferences on the respiratory care of the DMD patient were held on May 17, 2002 and May 19, 2003. Continued discussion in conference calls involving most members of the working group formed the basis for this document.

Methodologies for Synthesizing Expert Consensus

Critical review of pertinent literature was performed for the formation of this statement. Each consensus panel member was assigned a topic and reviewed all pertinent published literature using Medline, searching from 1966 through 2003, and including only human studies, which he or she then presented to the complete panel either at the initial two consensus conferences held at the American Thoracic Society annual meeting or in subsequent teleconferences. All members reviewed and approved the final manuscript. "Consensus" in this document refers to a unanimity of opinion of members of the group. Because the majority of the literature that addresses MD is limited by small patient numbers, this statement uses expert consensus in the majority of its recommendations. All recommendations made herein are therefore consensus guidelines.

Generous support from Parent Project MD and the Muscular Dystrophy Association is gratefully acknowledged.

Members of the ad hoc statement committee have disclosed any direct commercial associations (financial relationships or legal obligations) related to the preparation of this statement. This information is kept on file at the ATS headquarters.

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EVALUATION AND ANTICIPATORY GUIDANCE OF THE PATIENT WITH DMD

There are no prospective scientific data upon which to base recommendations regarding evaluation of patients with DMD. The committee has reviewed pertinent literature as referenced and have made all the following recommendations on routine evaluation based on expert consensus.

Routine Evaluation of Respiratory Function

DMD is associated with a gradual loss of muscle function over time. Loss of respiratory muscle strength, with ensuing ineffective cough and decreased ventilation, leads to pneumonia, atelectasis, and respiratory insufficiency in sleep and while awake (1). These complications are generally preventable with careful serial assessment of respiratory function. For patients with DMD, the optimal frequency of visits to the physician is not known. Respiratory evaluation of individuals with DMD includes obtaining a thorough history and physical examination, measurement of pulmonary function, and evaluation for sleep-disordered breathing (2).

Most patients with DMD do not realize when they have lost respiratory muscle strength to the point that they no longer have an effective cough until a respiratory viral infection leads to a prolonged cough or to pneumonia. Measurement of respiratory function and respiratory muscle strength allow the clinician to predict who will require assisted coughing and ventilation. Various levels of impairment of pulmonary function and gas exchange have been reported as associated with an increased risk of respiratory complications and death. One study reported a median survival of 3.1 years and 5-year survival of only 8% when the FVC fell below 1 L (3). Having an FVC less than 1 L remains the best negative predictor of survival in patients with DMD. An FEV_1 of 20% predicted or less has been associated with awake carbon dioxide retention (4). A poor 2- to 3-year survival has been seen in patients with awake levels of $PaCO_2$ on arterial puncture that were within normal limits (5, 6).

Recommendations

Multidisciplinary Care

- Patients with DMD should have regular visits and routine immunizations by a primary care physician as recommended for well children by the American Academy of Pediatrics.
- Patients should have access to specialists in pulmonology, neurology, cardiology, nutrition, physical medicine, orthopedic surgery, mental health, sleep medicine and social work.

Respiratory Care

- Patients should visit a physician specializing in pediatric respiratory care twice yearly after confinement to a wheelchair, fall in vital capacity below 80% predicted, and/or age 12 years.
- Children should have at least one visit with a physician specializing in pediatric respiratory care early in the course of the disease (between 4 and 6 years of age) and before confinement to a wheelchair to obtain baseline pulmonary function testing, for anticipatory medical guidance regarding the potential respiratory complications of the disease, and to assess the need for intensified therapy.
- Individuals who require mechanically assisted airway clearance therapy or mechanically assisted ventilation should see a pulmonologist every 3 to 6 months or as indicated for routine follow-up.
- All patients with DMD should undergo pulmonary and cardiac evaluations before surgeries.
- All patients with DMD should receive the pneumococcal vaccine and an annual influenza vaccination.

Routine Evaluation of Respiratory Function

- Objective evaluation at each clinic visit should include: oxyhemoglobin saturation by pulse oximetry, spirometric measurements of FVC, FEV_1, and maximal mid-expiratory flow rate (3), maximum inspiratory and expiratory pressures, and peak cough flow (8).
- Awake carbon dioxide tension should be evaluated at least annually in conjunction with spirometry. Where available, capnography is ideal for this purpose. Arterial blood gas analysis is not necessary for routine follow-up of patients with DMD. If capnography is not available, then a venous or capillary blood sample should be obtained to assess for the presence of alveolar hypoventilation.
- Additional measures of pulmonary function and gas exchange may be useful, including lung volumes, assisted cough peak flow, and maximum insufflation capacity.
- Carefully evaluate patients for evidence of other respiratory disorders, such as obstructive sleep apnea, oropharyngeal aspiration, gastroesophageal reflux, and asthma.
- Annual laboratory studies in patients requiring a wheelchair for ambulation should include a complete blood count, serum bicarbonate concentration, and a chest radiograph.

End of Life Directives

End of life directives are a critical part of the anticipatory care of individuals with DMD. There is evidence that health care professionals treating individuals with DMD underestimate the quality of life of ventilator-dependent people with DMD and that they may use their own perceptions of patient quality of life when deciding whether to discuss long-term ventilation with individuals with advanced DMD (9, 10). Quality of life judgments should be made with the informed participation of the patient and his or her family, and long-term mechanical ventilation should be offered for consideration even when the treating physician predicts that quality of life on long-term ventilation will be poor (11). As respiratory failure in DMD can occur either suddenly, in association with a respiratory tract infection, or gradually, education about ventilatory and palliative options should be provided before either of these scenarios occurs. The patient's and family's views on quality of life should be sought. The impact of long-term ventilation on the family and financial implications should be addressed and, when appropriate, the legal, religious, and cultural ramifications of these types of decisions should be discussed. These types of decisions are most difficult in the rare case of the young child with severe respiratory muscle weakness, who is too immature to participate in the discussion. End-of-life directives established by the patient, family, and health care team must be clearly documented and available for use in the case of an emergency.

Recommendations

- Physicians have a legal and ethical responsibility to disclose treatment options, including long term ventilation, to patients and/or their families. Physicians must avoid using their own perceptions of quality of life as the main factor in deciding whether to offer this type of information (10).
- End of life decision-making requires the provision of adequate information to the patient and family.
- Patients choosing to forgo long term ventilation must re-

ceive palliative care, in keeping with accepted standards (12).

Nutrition

Nutrition is a critical aspect of long-term management of patients with DMD. Regular involvement of a nutritionist with the care team can facilitate maintenance of ideal body weight, because both obesity (which can lead to obstructive sleep apnea) and malnutrition are detrimental to respiratory health. Although there are no data on nutrition and respiratory muscle strength in DMD, malnutrition has been associated with increased respiratory disease in other settings. It is therefore incumbent on clinicians to monitor and maintain ideal body weight in patients with DMD. Malnutrition and obesity appear to be equally common in young adults with DMD, each occurring in about 44% of individuals, and, in view of their deleterious effects on muscle and ventilatory function, should be avoided by careful dietary management (13). Reasons for malnutrition in late stages of DMD are primarily related to weakness and incoordination of the muscles of chewing and swallowing. Systemic steroid therapy also has nutritional implications, with potentially increased risks of osteoporosis and obesity, necessitating dietary manipulation (63). Nearly 1/3 of patients with DMD complain of choking while eating, and with disease progression, a heightened risk of aspiration may develop (14).

Recommendations

- Percentage ideal body weight and body mass index must be assessed regularly and counseling provided as necessary.
- A nutritionist should evaluate patients with DMD as part of their regular follow-up care.
- Swallowing should be assessed through the history, and clinically, by observing the patient's ability to manage various textures.
- Evaluation for dysfunctional swallowing should be performed if there is a history of choking or dysphagia.
- Video-fluoroscopy may be used to confirm the presence of aspiration, and to assist the clinician to prescribe safer swallowing techniques.
- When adequate nutrition cannot be safely accomplished with oral feedings, gastrostomy tube placement and enteral feedings under the guidance of a nutritionist is strongly recommended.

Sleep Evaluation in DMD

DMD is associated with sleep-disordered breathing and alveolar hypoventilation. Onset of respiratory insufficiency can be subtle. Symptoms of sleep hypoventilation include gradually increasing numbers of nocturnal awakenings, daytime sleepiness, morning headache, and, rarely, vomiting. Patients with DMD are also at risk for upper airway obstruction.

The timing of polysomnography to detect sleep hypoventilation has not been determined in patients with DMD. In one study, sleep hypoventilation correlated with an awake Pa_{CO_2} of $\geq$ 45 mm Hg and a base excess $\geq$ 4 mmol/L (4). Another study suggested that an unattended sleep study in the home could identify sleep-disordered breathing in patients with DMD, without polysomnography (15). Simple oximetry in the home can screen for sleep-related oxyhemoglobin desaturation.

Recommendations

- Review sleep quality and symptoms of sleep-disordered breathing at every patient encounter.
- Annual evaluation for sleep-disordered breathing should be performed in patients with DMD starting from the time they are wheelchair users and/or when clinically indicated.
- Where available, annual polysomnography with continuous CO_2 monitoring is ideal.
- In areas where full polysomnography is not readily available, overnight pulse oximetry with continuous CO_2 monitoring provides useful information about nighttime gas exchange, although sleep-disordered breathing not associated with desaturation or CO_2 retention will not be detected. A simple capillary blood gas upon arousal in the morning can demonstrate CO_2 retention, although not as sensitively as continuous capnography.

Cardiac Involvement

Cardiac involvement is universal in individuals with DMD. Cardiac disease is the second most common cause of death in persons with DMD, with 10–20% of individuals dying of cardiac failure (16). Dilated cardiomyopathy primarily involves the left ventricle, and can lead to dyspnea and other symptoms of congestive heart failure (17, 18). Conversely, right ventricular failure can result from respiratory failure and pulmonary hypertension. Individuals with DMD are also at risk for ventricular arrhythmias (19). Whereas some studies have suggested that the respiratory and peripheral muscle weakness tend to be inversely related to the risk of cardiac failure, other studies suggest that left heart and respiratory failure tend to occur in parallel (19–21). There are retrospective data suggesting that cardiac involvement is less frequent in children treated with deflazacort (22). Cardiac assessment and treatment of congestive heart failure in patients with DMD falls outside the scope of this document.

Recommendation

- All individuals with DMD require regular cardiac evaluation with annual electrocardiograms and echocardiograms, starting at least by school age.

MANAGEMENT

Airway Clearance

Effective airway clearance is critical for patients with DMD to prevent atelectasis and pneumonia. Ineffective airway clearance can hasten the onset of respiratory failure and death, whereas early intervention to improve airway clearance can prevent hospitalization and reduce the incidence of pneumonia (8). Assessment of cough effectiveness includes measurements of maximal inspiratory and expiratory pressures, peak cough expiratory flow, and either inspiratory or vital capacity. Cough peak flows correlate directly to the ability to clear secretions from the respiratory tract (23), and values below 160 L/min have been associated with ineffective airway clearance (24). Baseline peak cough expiratory flow rate measurements above 160 L/min, however, do not guarantee adequate airway clearance, because respiratory muscle function can deteriorate during respiratory infections (25). For this reason a peak cough expiratory flow rate of 270 has been used to identify patients who would benefit from assisted cough techniques (8). Another study found that the ability to generate adequate flow for effective coughing correlated with a maximum expiratory pressure (MEP) of 60 cm H_2O and above, and was absent at levels below 45 cm H_2O (26). Pulse oximetry has been used to screen for lower airway complications of respiratory tract infections and help caregivers know when to intensify airway clearance therapy (8). Various techniques have been developed to overcome ineffective cough in patients with neuromuscular weakness.

"Maximum insufflation capacity" is the maximum air volume that can be held with a closed glottis. It is influenced by strength of oropharyngeal and laryngeal musculature. A training program

in air stacking in patients with neuromuscular disease (including DMD) improves range of motion of the lung and chest wall and therefore maximum insufflation capacity (7). In theory this will aid in assisted coughing by increasing the volume of expelled air.

Manual Techniques. Manually assisted coughing involves inspiratory assistance followed by augmentation of the forced expiratory effort. An increase in inspiratory capacity can be achieved by the use of glossopharyngeal breathing (in essence forcing air into the lungs using one's mouth), air stacking (taking a series of tidal breaths without exhaling between them) (7), application of positive pressure with self-inflating bag and mask, intermittent positive pressure breathing device, or mechanical ventilator. Interfaces for inspiratory assistance include a facemask, mouthpiece, or direct attachment of the assisting device to a tracheostomy tube. Forced exhalation is augmented by pushing on the upper abdomen or chest wall in synchrony with the subject's own cough effort.

Mechanical Techniques. Mechanical insufflator-exsufflators simulate a cough by providing a positive pressure breath followed by a negative pressure exsufflation (27, 28). Comparison of peak cough expiratory flow rates by mechanical insufflation-exsufflation were shown to be superior to those generated either by breath stacking or manual cough assistance (29).

Use of mechanical insufflation-exsufflation was found to be particularly important in preventing hospitalization or need for tracheostomy in patients with DMD with peak cough expiratory flows around 160 L/min, especially when scoliosis prevented optimal use of manual assisted cough (8). The device has been shown to be well tolerated and effective in 42 pediatric patients with neuromuscular disease (15 with DMD) and ineffective cough (30). Reported complications include transient nausea, abdominal distention, bradycardia, and tachycardia (28). In patients with DMD with tracheostomies, mechanical insufflation-exsufflation offers a number of advantages over traditional suctioning, including clearance of secretions from peripheral airways, avoidance of mucosal trauma from direct tracheal suction, and improved patient comfort (31).

Mucus Mobilization Devices. Intrapulmonary percussive ventilation delivers bursts of high frequency, low amplitude oscillations superimposed on ramping continuous positive airway pressure. A recent case series including one patient with DMD reported the effectiveness of intrapulmonary percussive ventilation in resolving persistent pulmonary consolidations refractory to conventional therapies. (32) High frequency chest wall oscillation has been used in patients with neuromuscular weakness but there are no published data on which to base a recommendation. Any airway clearance device predicated upon normal cough is less likely to be effective in patients with DMD without concurrent use of assisted cough.

Bronchoscopy has been used in selected patients with DMD, generally in cases of persistent atelectasis, but has not been of proven benefit and therapy and should be considered only after all non-invasive airway clearance techniques have proven unsuccessful and a mucus plug is suspected.

Recommendations

- Patients with DMD should be taught strategies to improve airway clearance and how to employ those techniques early and aggressively.
- Use assisted cough technologies in patients whose clinical history suggests difficulty in airway clearance, or whose peak cough flow is less than 270 L/minute and/or whose maximal expiratory pressures are less than 60 cm H_2O.
- The committee strongly supports use of mechanical insufflation-exsufflation in patients with DMD and also recommends further studies of this modality.
- Home pulse oximetry is useful to monitor the effectiveness of airway clearance during respiratory illnesses and to identify patients with DMD needing hospitalization (8).

Respiratory Muscle Training

The rationale for respiratory muscle training in DMD is based on the assumption that improved muscle strength and endurance in patients affected with the condition may lead to improved preservation of lung function over time. However, the effects of respiratory muscle training in patients with DMD vary, with some studies reporting substantial improvements in muscle strength and endurance and others essentially demonstrating minimal or insignificant changes in respiratory muscle performance (33–43). In addition, the recently discovered protective mechanism of nitric oxide release in exercising muscle may be defective in children with DMD (44, 45). This could potentially lead to increased muscle damage during application of training protocols. Therefore, recommendations regarding respiratory muscle training cannot be fully endorsed and will have to await further studies.

Noninvasive Nocturnal Ventilation in DMD

Patients with DMD have increased risk for sleep-disordered breathing, including hypopnea, central and obstructive apnea, and hypoxemia. Treatment of these pulmonary complications with noninvasive ventilatory support may improve quality of life and reduce the high morbidity and early mortality associated with DMD (6, 46, 47).

Nocturnal nasal intermittent positive pressure ventilation with bilevel positive airway pressure generator or mechanical ventilator has been used successfully in the treatment of sleep-disordered breathing and nighttime hypoventilation in patients with DMD and other neuromuscular disorders (48–50). The level of positive pressure required to eliminate obstructive apneas or hypopneas and normalize ventilation and nighttime oxygen saturation must be determined in the sleep laboratory or with careful bedside monitoring and observation. Serial evaluation and adjustment of nasal intermittent positive pressure ventilation (NIPPV) is necessary, as the patient's requirements change with time (49). Nocturnal NIPPV in DMD has resulted in apparently improved survival (46, 51), improved quality of sleep, decreased daytime sleepiness, improved well-being and independence, improved daytime gas exchange, and a slower rate of decline in pulmonary function compared with nonventilated control subjects (6, 46, 47, 50, 52–54).

Complications of nasal intermittent positive pressure ventilation include eye irritation, conjunctivitis, skin ulceration, gastric distention, and emesis into a full face mask. Facial complications can be avoided by regular follow-up to assess mask fit. Nasal steroids or humidification of the delivered air can help relieve nasal obstruction. There has been a single case report of recurrent pneumothorax in a 26-year-old man with a non-Duchenne muscular dystrophy on nasal intermittent positive pressure ventilation who had subpleural blebs (55). In fragile patients, mask displacement can rapidly lead to severe hypoxemia and hypercapnia. Because most bilevel machines do not have built-in alarms, additional monitoring, such as pulse oximetry, is useful in this setting.

Other therapies. Nasal continuous positive airway pressure (CPAP) is likely to be limited utility in patients with DMD, and only in those with obstructive sleep apnea syndrome but with normal nocturnal ventilation. In cases of hypoxemia due solely or partially to hypoventilation, support with BiPAP or a volume ventilator should be considered. As hypoxemia in DMD is usu-

ally a manifestation of hypoventilation, treatment with oxygen without concurrent supplemental ventilatory support should be avoided. Negative pressure ventilators can lead to upper airway obstruction in patients with DMD, possibly due to the lack of synchrony between inspiration and vocal cord abduction (52, 56).

Recommendations

- Discussions regarding ventilatory support for each patient should involve the patient, caregivers, and medical team.
- Perform polysomnography with continuous CO_2 monitoring in patients with DMD to assess adequacy of home ventilatory support. In areas where polysomnography is not readily available, overnight pulse oximetry with continuous CO_2 monitoring can be used to monitor nighttime gas exchange. Where CO_2 monitoring is not available, overnight pulse oximetry can be used to detect nighttime oxyhemoglobin desaturation. Simple oximetry provides, at best, only indirect information on ventilation, and should be used to assess need for ventilatory support only when better alternatives are unavailable.
- Schedule periodic reassessment as appropriate to stage of disease. Follow-up visits should include monitoring for the development of daytime hypoventilation, which may necessitate around-the-clock ventilation.
- Use nasal intermittent positive pressure ventilation to treat sleep-related upper airway obstruction and chronic respiratory insufficiency in patients with DMD.
- Negative-pressure ventilators should be used with caution in patients with DMD due to the risk of precipitating upper airway obstruction and hypoxemia.
- Do not use oxygen to treat sleep-related hypoventilation without ventilatory assistance.

Daytime Noninvasive Ventilation

With time, patients with DMD progress to a state of constant hypoventilation, and require 24-hour support. Although such patients have traditionally received continuous ventilatory support by tracheostomy, ventilatory support can also be provided successfully using noninvasive methods.

The most commonly used noninvasive technique is mouthpiece intermittent positive pressure ventilation. This modality uses a commercially available or custom-made mouthpiece placed near the mouth using a flexible gooseneck attached to the wheelchair, and to a ventilator cycled using assist-control (51, 57, 58). The patient places the mouthpiece between the lips and inhales at regular intervals. This technique has been used successfully in patients with DMD with a mean FVC of 0.6 L (5% predicted) for greater than 8 years (47, 58–60). Mouthpiece ventilation is well tolerated and does not interfere with eating or speaking.

Other techniques for daytime noninvasive ventilation are also available. Glossopharyngeal breathing uses oral muscles to "gulp" small boluses of air into the lungs, with six or more gulps producing a V_T breath. This technique may allow short periods off mechanical ventilation, and is useful in the event of ventilator failure (47, 57). The intermittent abdominal pressure ventilator (or Pneumo-belt) uses an inflatable bladder placed over the abdomen, connected to a conventional portable ventilator. Inflation of the bladder, with the patient seated, creates a forced exhalation, and inhalation occurs through subsequent passive descent of the diaphragm and outward recoil of the ribcage. This method may not work in patients with scoliosis or obesity (60, 61). Negative-pressure ventilation using a chest cuirass can also be used for daytime ventilation, although current models are not portable (51, 58).

Recommendations

- Consider daytime ventilation when measured waking P_{CO_2} exceeds 50 mm Hg (*see* ROUTINE EVALUATION OF RESPIRATORY FUNCTION) or when hemoglobin saturation remains $< 92\%$ while awake.
- In centers with appropriate expertise, consider mouthpiece intermittent positive pressure ventilation or other forms of noninvasive daytime ventilation. Consider tracheostomy when contraindications or patient aversion to noninvasive ventilation are present.
- Patients receiving noninvasive ventilation should have regular (at least annual) noninvasive monitoring of gas exchange, including oxygen saturation and end-tidal P_{CO_2} levels.

Continuous Invasive Ventilation

Daytime and nighttime ventilation can be provided in individuals with DMD using a tracheostomy, when other device interfaces are poorly tolerated or the patient lacks sufficient oromotor and/or neck control to use a mouthpiece interface during the daytime. Advantages of a tracheostomy include a more secure ventilator–patient interface, the ability to provide higher ventilator pressures in patients with intrinsic lung disease or severe reductions in chest wall compliance (for example, secondary to scoliosis), and the ability to perform direct airway suctioning during respiratory infections. However, tracheostomies have many potential complications, including generating more secretions, impairing swallowing and increasing the risk of aspiration, and the bypassing of airway defenses, likely increasing the risk of infection (62). There is a risk of airway occlusion by a mucus plug (63). Traditionally, tracheostomies also impair oral communication. For many patients, communication may be restored using a relatively small tracheostomy tube allowing a "leak" around the airway, and a speaking valve (64). Loss of ventilator tidal volume with a leaky system can be compensated for by increasing the tidal volume (65). Many patients are concerned about the cosmetic and potential communication implications of tracheostomy, and this needs to be addressed with sensitivity during discussions about continuous ventilation (66).

Recommendations

- Tracheostomy should be considered when contraindications or patient aversion to noninvasive ventilation are present, or when noninvasive ventilation is not feasible due to severe bulbar weakness or dysfunction.
- Patient autonomy, to undertake or forgo ventilation by tracheostomy, must be respected, once appropriate education has been provided to the patient and family.
- Patients with a tracheostomy must receive adequate monitoring by pulse oximeter to detect mucus plugs (63).

Scoliosis in DMD

Nearly all patients with DMD develop scoliosis after losing independent ambulation (67–69), beginning in the second decade of life. Once scoliosis reaches 30 degrees, it progresses with age and growth (68, 70–72). Failure to repair scoliosis in DMD can result in increased hospitalization rates and poor quality of life.

Optimal timing for surgical intervention is while lung function is satisfactory and before cardiomyopathy becomes severe enough to risk arrhythmia under anesthesia. Surgery is usually scheduled once the Cobb angle measured on scoliosis films is between 30 and 50 degrees (68, 73, 74).

There are no absolute contraindications for surgery based on pulmonary function; some report good results even in patients whose FVC is 20% of predicted (75, 76). Best prognosis for

recovery seems to be FVC > 40% (77), although others use the absolute vital capacity of < 1,900 ml as an indicator of rapid progression of scoliosis and poor prognosis (78). A sleep study or nocturnal oximetry screen also helps with perioperative planning; if these tests are abnormal, patients can begin nocturnal noninvasive ventilation before surgery and extubate to noninvasive ventilation postoperatively. It is critical that the patient's cardiac, nutritional, and respiratory status be optimized before surgery. Postoperative pain management should be titrated to promote airway clearance and minimize respiratory suppression.

Recommendations

- Perform preoperative evaluation by pulmonologist and cardiologist at least 2 months before surgery, to allow for intervention.
- Assess for sleep hypoventilation preoperatively.
- Essential postoperative care includes aggressive airway clearance and respiratory support. Patients should be followed by a pulmonologist or physician specializing in respiratory care to optimize postoperative respiratory management and prevent complications.

Corticosteroids in the Management of DMD

Oral corticosteroids have been found to increase muscle mass and retard muscle deterioration in patients with DMD (79–81). Despite their potential benefit, their use is controversial and not uniformly recommended. In most studies, oral steroid therapy was initiated between 5 and 15 years of age, and at an average of approximately 8 years of age.

Prednisone is the most studied steroid in Duchenne muscular dystrophy (79, 82–89). Deflazacort, an oxazoline derivative of prednisone, has been shown to have similar benefits to prednisone, with possibly fewer side effects (90–94). Boys who receive deflazacort maintain ambulation longer and have significant sparing of pulmonary function (94).

Recommendations

- Future research is required to confirm and further define the potential pulmonary benefits of oral steroids.
- Decisions to start oral steroid therapy to help preserve lung function should be made in collaboration with the neuromuscular specialist and other members of the multidisciplinary care team and the family.

Patient Education in Duchenne Muscular Dystrophy

The goal of patient education is comanagement of care by the patient and family in collaboration with their health care providers. Educational strategies should be developmentally sensitive and appropriate for the current stage of disease (95, 96). Education should begin as soon as possible after diagnosis and continue as a key component of ongoing care (Table 1). The goals of patient/family education relating to the respiratory complications of DMD are to:

1. Understand the natural history of DMD.
2. Recognize early signs and symptoms of pulmonary complications.
3. Understand and make informed choices about treatment options for airway clearance and respiratory insufficiency. Discussion should include options for noninvasive ventilation as well as ventilation via tracheostomy. Risks, benefits, and quality of life issues for the different ventilatory support options should be reviewed.
4. Provide anticipatory guidance on the assessment and management of intercurrent respiratory illnesses.
5. Understand the role of the medical devices in use, and have sufficient skill to operate them effectively.
6. Understand and make informed decisions about end of life care.

Excellent resources for families include the pamphlet *Breathe Easy, Respiratory Care for Children with Muscular Dystrophy*; the video *Breathe Easy* (97); and internet web sites from the Muscular Dystrophy Association (http://www.mdausa.org) and the Parent Project Muscular Dystrophy (http://www.parentprojectmd.org).

Long-Term Care Issues

Several studies suggest that nocturnal or full-time mechanical ventilation increases survival among patients with DMD who are hypercapneic (46, 98–100). None of these studies, however, represents a controlled, prospective trial. Nevertheless, one large population study of all patients with DMD in Denmark showed a significant decrease in mortality rate and increase in 15- or 20-year survival in the era when mechanical ventilation was routinely offered compared with the period when mechanical ventilation was used only sporadically (98). In another large center where no patients were treated with home mechanical ventilation before 1991, survival since 1990 among patients with DMD who refused chronic mechanical ventilation was 19.29 years (95% CI 18.61, 19.97 years), compared with 25.3 years (95% CI 23.11, 26.58 years) for those patients who chose to use long-term mechanical ventilatory support (99). Using these studies, however, it is not possible to separate the salutary effects of mechanical ventilation from other improvements in the care of patients with DMD, such as use of aggressive airway techniques or the development of regional centers of excellence for the care of patients with neuromuscular disease (98).

Although the above reports support a role for mechanical ventilation in patients with established or impending respiratory failure, there are no data to support a preventive role for mechanical ventilation. In a multicenter, prospective, controlled trial, patients with DMD who were normocapneic with FVCs between 20 and 50% of predicted were randomized to receive either 6 or more hours of nocturnal noninvasive ventilation or no ventilatory support (101). Although 15 of the 35 patients receiving NIPPV did not adhere to the protocol, survival was significantly decreased in the group receiving "preventive" nasal ventilation. This caused the authors to conclude that NIPPV for preventive purposes should be avoided in patients with DMD, and to speculate that a false sense of security with less diligent monitoring was associated with the use of NIPPV and was responsible for the increased death rate among users in the study.

What impact long-term mechanical ventilation has on the quality of life of patients with DMD and their families is not straightforward. Several studies report either generally acceptable (102, 103) or improved quality of life (104, 105) among patients with DMD who chose to use long-term mechanical ventilation. Because mechanical ventilation does not prevent progression of the underlying disease, it was difficult to distinguish dissatisfaction related to disease progression and its impact on daily functioning from effects and family stress related to introduction of mechanical ventilation (102). It is clear, however, that physicians and other healthcare workers markedly underestimate the quality of life perceived by ventilator-dependent patients with DMD (104). Furthermore, those negative perceptions contribute to the failure of some physicians to offer mechanical ventilatory support as an option, or cause them to present the option in a negative light (10). Importantly, patients expressed value in being able to have meaningful discussions about mechanical ventilatory support repeatedly throughout the course of their

TABLE 1.

Stage	Decision	Knowledge/Skills
Normal respiratory function		Knowledge: • How the respiratory system works • Natural history of respiratory function in DMD • Preventive care: routine immunizations, annual influenza immunization, avoidance of secondhand smoke, avoidance of obesity, need for regular follow up • Discussion of airway clearance techniques Skills: • Performance of pulmonary function testing
Adequate Ventilation, Ineffective Cough		Knowledge: • Early and aggressive management of respiratory infections, respiratory insufficiency, and swallowing dysfunction • Understand the need for sleep and swallow studies • Introduce treatment options for long-term respiratory support Skills: • Assisted coughing techniques • Mucus mobilization techniques • Use of pulse oximetry
Adequate daytime ventilation, inadequate nighttime ventilation	Chooses ventilatory support	Knowledge: • Understand options for long term respiratory support • Avoidance of interface complications • Anticipatory guidance for management of intercurrent respiratory illnesses. • Discuss advanced directives Skills: • Use of assisted ventilation device(s) • Use of device interface • Tracheostomy care (if chosen).
	Chooses to have no ventilatory support	Knowledge: • Understand options for long term respiratory support • Provide end of life counseling • Offer consultation with palliative care specialists Skills: • Written advanced directives
Inadequate daytime and nighttime ventilation	Chooses ventilatory support	Knowledge: • Understand options for continuous ventilatory support • Anticipatory guidance for management of intercurrent respiratory illnesses. • Offer end of life counseling • Consider consultation with palliative care/hospice specialists. Skills: • Use of chosen ventilatory support • Tracheostomy care (if chosen) • Written advanced directives
	Chooses to have no ventilatory support	Knowledge: • Provide end of life counseling • Offer consultation with palliative care/hospice specialists Skills: • Written advanced directives

disease (103). Such opportunities, however, are frequently missed or not used effectively by the health care team (106).

Patients with DMD are surviving into adulthood as a result of improved respiratory care. This has placed families of these patients in the difficult situation of finding trained physicians who are comfortable taking over either primary care or specialty care of the technology-dependent patient.

Recommendations

- Discussions about mechanical ventilation should occur well before its need is apparent, should include alternate options, and should be repeated as the underlying disease progresses.
- Care of the technology-dependent young adult should be incorporated into the training programs of adult pulmonologists, adult neurologists, physiatrists, sleep medicine specialists, and internists.
- Pulmonologists, physiatrists, and internists with training and expertise in the care of the adult neuromuscularly weak patient should be identified in every community to aid in the transition to adult care.

End of Life Care

Care for someone in the terminal stages of a progressive chronic illness focuses on enhancement of quality of life for the patient and their family. An interdisciplinary approach is required, including primary and specialist physicians, hospice/palliative care specialists, social services, and spiritual care, family members, and others appropriate to the patient's cultural/religious background (107–110).

The goals of end of life care for patients with muscular dystrophy include:

1. Treating conditions (pain, dyspnea) that cause distress (palliative care).

2. Attending to the psychosocial and spiritual needs of the patient and their families.
3. Respecting the patient and family's choices concerning testing and treatment.

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