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A 2009 Perspective on the 2004 American Thoracic Society Statement, "Respiratory Care of the Patient With Duchenne Muscular Dystrophy"

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SUPPLEMENT ARTICLE

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ABSTRACT

This is a summary of the presentation "A 2009 Perspective on the 2004 American Thoracic Society Statement, 'Respiratory Care of the Patient With Duchenne Muscular Dystrophy,' " presented as part of the program on pulmonary management of pediatric patients with neuromuscular disorders at the 30th annual Carrell-Krusen Neuromuscular Symposium on February 20, 2008. *Pediatrics* 2009;123:S239–S241

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Abbreviations
ATS—American Thoracic Society
DMD—Duchenne muscular dystrophy

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THE 2004 AMERICAN Thoracic Society (ATS) publication of guidelines for respiratory care in Duchenne muscular dystrophy (DMD)[1] can be viewed as the result of a "perfect storm" of factors leading to its publication. New, noninvasive technologies had become available for respiratory management of the patient with DMD. Respiratory care protocols for prevention of pulmonary morbidity, as outlined by Bach et al,[2] had gained wide acceptance among respiratory specialists, whereas among nonpulmonary physicians, these therapies were not being prescribed. The major respiratory complications of DMD (inadequate cough and inadequate ventilation) had specific, noninvasive therapies. Yet, the major insurers (both private and Medicaid insurers) were refusing to pay for these therapies, citing a lack of efficacy data performed in the usual randomized, prospective manner. Mechanical insufflation-exsufflation, which had been shown to be quite efficacious in small physiologic and retrospective studies, was still deemed "experimental therapy" by most payers. In the 1990s, many clinicians charged with the care of patients with DMD viewed the disease as fatal and untreatable. Therapeutic nihilism (eg, the "take-them-home-and-love-them" approach) was common, and many families were not offered even moderately aggressive approaches toward maintenance of respiratory health.

The 1990s also saw the rise of the parental advocacy movement in parallel with the growth of the Internet, which instantly linked geographically distant parents. The parent advocacy organization Parent Project Muscular Dystrophy was founded in 1994.

The final factor was the rise of what has been referred to as the "guidelines movement." This refers to an increasing trend among medical specialties to create practice parameters and care documents based on careful review of the medical literature. "Evidence-based medicine" is an offshoot of this movement, with an emphasis on empiric data in medical decision-making.

As a result of the above-discussed factors, a number of the authors of the 2004 ATS statement found themselves frustrated by an inability to deliver appropriate care to their patients. The major impediment was, of course, insurance coverage. Meanwhile, parents around the country became aware of a newer standard of care for patients with DMD through the Internet and through involvement with parent groups such as Parent Project Muscular Dystrophy. This became the incentive to create a consensus statement: If the insurance companies were denying coverage and citing a lack of literature, we would have to create it ourselves in the form of an expert consensus panel.

The ATS panel was formed after an interest group was created on the Internet and by invitation based on publication and expertise. To not solely have pulmonologists, 1 neurologist and 1 nurse with expertise in the care of patients with DMD were also invited to round out the panel. The initial meeting took place in 2001 at the annual ATS meeting, with follow-up meetings in 2002 and 2003. Teleconference meetings were held to create a forum for discussion and to form consensus on all aspects of care. The final manuscript was reviewed by 6 peer reviewers and the entire board of directors of the ATS and published in August 2004.

One might presume that such a document, in which noninvasive respiratory management, mechanically assisted coughing, multidisciplinary care, and strong support of the family was included in all decisions, would have opened many doors for patients. In many areas, this is the case. For example, at present, most state Medicaid programs cover the mechanical insufflator-exsufflator. Yet, the majority of private payers still do not cover this device, even in the face of a published expert panel consensus report supporting its use.

Experience and training also dictate practice patterns. Because most clinicians involved in the care of patients with

DMD have not been trained in the noninvasive respiratory management of these patients, such therapies are not even offered in some communities. At present, the Muscular Dystrophy Association has not established minimal standards of care for their supported clinics (although they are clearly moving in this direction).

Worldwide, the same issues exist. The national health programs of Australia, for example, do not support mechanically assisted cough in DMD, and private funds are often used to get these devices into communities. Often, a single device must serve a single community despite the epidemic nature of respiratory infections. Many nations do not offer this technology to the patients, nor can families afford to obtain such care.

The 2004 ATS consensus statement on the respiratory care of the patient with DMD was designed to give both primary care doctors and specialists some guidance in an age in which there was a dichotomy of care philosophies. Depending on the training and experience of the clinicians involved, patients with DMD might be offered aggressive respiratory support (both invasive and noninvasive) or might be counseled to seek end-of-life care when there was evidence of prolongation of life by supportive therapies. "Therapeutic nihilism" had once been the norm for this patient population. Authors of the ATS consensus statement sought to improve care of individuals with DMD while allowing them to have a greater say in medical decision-making. The premise of the project was simple: Noninvasive options for respiratory management should be offered to patients, and invasive options should be offered when those noninvasive choices could not succeed for patient-related or system-of-care–related reasons. The basic principals of respiratory care of the patient with DMD (and for patients with all forms of weakness) are to support airway clearance and support breathing.

It should be pointed out that this document is neither a systematic review nor a practice guideline, both of which are based on evidence. The vast majority of the literature concerning the respiratory management of patients with DMD is not randomized or controlled or prospective. As such, the consensus committee (formed as a subcommittee of the pediatric section of the ATS) primarily used expert panel consensus as evidence of lacking for the majority of sections.

The following is a summary of the 2004 ATS consensus statement.

1. Surveillance: To avoid a crisis model of care (eg, meeting the patient in the ICU), the committee recommended early (ages 4–6) or presymptomatic consultation with the respiratory specialist and regular screening of respiratory function. The committee recommended regular (twice-annual) visits with a respiratory specialist once the patient is no longer ambulatory or once forced vital capacity is <80% predicted or after the age of 12 years. Once mechanical support of coughing or breathing is instituted, these visits are recommended to follow an every 3- to 6-month schedule.
2. Airway clearance: Airway clearance becomes impaired after the patient with DMD loses the ability to ambulate, generally in the second decade of life. The patient with neuromuscular disorders retains mucociliary clearance while losing cough clearance. As a result, therapies directed at improving mucociliary clearance (such as high-frequency chest wall compression) will not significantly aid these patients. Manually assisted cough is a technique supported by the committee. At the time of publication, the majority of the data published on mechanical insufflation-exsufflation were anecdotal and retrospective. Yet, the experience of the committee members with this new technology was compelling and supported the published experience. As a result, the committee strongly supported mechanical insufflation-exsufflation for those patients with evidence of impaired cough. Such evidence included but was not limited to diminished peak flow, peak cough flow, and maximal expiratory pressure measured in the pulmonary function laboratory or at the bedside.
3. Respiratory muscle training: Because published data on the value of respiratory muscle training are conflicting and there was concern that muscle training might, in fact, harm the patient (because of deficiency of nitric oxide), the committee did not support respiratory muscle training for patients with DMD.
4. Support of breathing in sleep: The second stage of respiratory impairment occurs after the loss of an effective cough. The ATS consensus committee recommended noninvasive options for supporting breathing in sleep and supported obtaining nocturnal polysomnograms (where available) when there is clinical evidence of respiratory insufficiency in sleep for diagnosis and management. Avoidance of supplemental oxygen to "treat" nocturnal hypoventilation is crucial, because supplemental oxygen can suppress respiratory drive and worsen the defect. The committee recommended regular screening to assess for complications of therapy (such as interface complications) and adequacy of support.
5. Support of breathing during the daytime: The final stage of respiratory impairment in DMD, inadequate ventilation during awake hours, occurs after the development of nocturnal hypoventilation. The committee advocated for noninvasive options for support (a volume ventilator with a mouthpiece interface) as first choice while acknowledging that invasive options (eg, tracheostomy and ventilator) may be preferable to certain patients and providers. Screening for hypoventilation at regular intervals was recommended, including measurement of oxyhemoglobin saturation and exhaled CO_2. A level of exhaled CO_2 at >50 mm Hg and/or hemoglobin saturation at <92% while awake was deemed to be an indication for daytime ventilatory support. Indications for invasive support cited in the consensus statement included patient preference and bulbar weakness.
6. Other issues in DMD: Scoliosis is a common problem in DMD, especially for those patients who do not

receive corticosteroids. The committee was careful to point out that there are no absolute contraindications to the scoliosis surgery based on pulmonary function and that it was important to optimize respiratory, cardiac, and nutritional health before the surgery. Polysomnography may identify those patients who are at risk for ventilatory failure in the postoperative period so that noninvasive ventilation can be used in recovery.

Corticosteroids are routinely used for DMD now, and the committee supported this.

End-of-life care is also an important aspect of caring for individuals with an ultimately fatal illness. Palliative care should be offered to those individuals who do not choose ventilatory support.

Patient and family education that is developmentally appropriate was advocated. Anticipatory guidance, with a goal of teaching the family how to manage intercurrent respiratory illness and anticipate the next phase of disease, was also stressed.

In summary, the ATS consensus statement on the respiratory care of patients with DMD has helped many patients receive improved care by offering clinicians guidance and helping medical directors of insurance companies make better decisions regarding use of technology to prevent morbidity and mortality. Despite this, much work remains to aid patients with DMD in the United States and worldwide. There remains a strong role for parental advocacy, especially in the realm of improving insurance coverage of life-saving technologies that prevent pulmonary morbidity. There remains a shortage of specialists trained in the respiratory management of DMD nationwide and worldwide. Although for many the situation has greatly improved since the 2004 consensus statement, much work remains to help those who remain in crisis.

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