Forty-Eight Years with Duchenne Muscular Dystrophy

ABSTRACT


Key Words: Duchenne Muscular Dystrophy, Neuromuscular Disease, Noninvasive Mechanical Ventilation, Insufflation-Exsufflation, CoughAssist, Survival

The Internet tells patients that, “The average life expectancy for patients afflicted with Duchenne Muscular Dystrophy (DMD) varies from late teens to early to mid-20s. There have been reports of a few DMD patients surviving to the age of 40 but this is extremely rare.”

The Muscular Dystrophy Association Web site tells us, “In all likelihood, your son will be a young man by the time his DMD becomes fatal. Eventually his respiratory system and his heart will be severely weakened by the disease. Then you’ll begin to face the fact that he doesn’t have much time left..... Some young men in the later stages of DMD welcome the end of their struggle, while others insist on every last medical intervention that might give them a few more days or hours of life..... One or both of you may experience a new wave of denial, believing that some divine miracle or new medical procedure will prevent the inevitable.”

On March 20, 2011, Jeff Gray (Fig. 1) died suddenly, without discomfort or any warning, while talking with his brother, 1 mo shy of his 48th birthday. Was it thanks to a “divine miracle” or a simple intervention that he has been using for the last 27 yrs to prevent respiratory failure? Although this intervention has been available to people with Duchenne muscular dystrophy (DMD) since at least 1969, it continues to be offered only to those coming to our center and to, perhaps, only two others in the United States.

Jeff had a typical clinical course for DMD and an Xp21 7 exon deletion that encodes for the missing dystrophin. In 1991, his left ventricular ejection fraction was normal, but by 1998, it was down to 25%. Without ever developing cardio-respiratory failure, Jeff gradually became continuously ventilator-dependent, using a 15-mm angled mouthpiece for daytime noninvasive mechanical ventilation (NIV) for 25 yrs (Fig. 1) and nasal or lip-seal ventilation for nocturnal support for 27 yrs. His vital capacity had been less than 200 ml since age 33 yrs and less than 100 ml since age 42 yrs (Fig. 2), but, despite a vital capacity of 0 ml on March 14, 2011, glottis function permitted him to air stack to 1530 ml and to perform glossopharyngeal breathing (GPB).

The ability to perform glossopharyngeal breathing permitted Jeff ventilator-free breathing ability until his maximum glossopharyngeal breathing capacity decreased to less than 400 ml after age 42 yrs (Fig. 2).
There have been no publications from United States centers on this full-support noninvasive approach other than ours, a 1979 paper by Alexander et al., and one by Josh Benditt in 2006; there was only one paper on the extubation of unweanable DMD patients to full-support NIV without resorting to tracheostomy. As a result, few DMD patients in the United States avoid acute respiratory failure, intubation, tracheostomy, and premature death. The question of why few physicians are learning to use these methods has already been considered. More puzzling is why patient-supported organizations dedicated to DMD are interested. Although there are about 230 Muscular Dystrophy Association clinics in the United States and other clinics associated with groups like the Parent Project for DMD, none of these offer mouthpiece NIV, mechanically assisted coughing, and extubation of “unweanable” DMD patients to avoid tracheostomy, thereby leaving these “young men [to] welcome the end of their struggle,” that is, to die prematurely or spend a lifetime with tubes in their necks.

Appreciating what it means to avoid respiratory failure and invasive tubes, Jeff developed a Web site in 1998 to spread this knowledge. This was the first Web site with a video showing mouthpiece NIV for ventilatory support. He also developed a DMD social network. Because of his personal experience with so many constipation regimens, Jeff contributed to the gastrointestinal management chapter of my last book. In February 2011, Jeff completed a book entitled, “I’m Not Dead Yet” to deliver his message, for prepublication editing. Living only 7 miles from my home, Jeff was the first patient visited by more than 100 physicians and therapists through the years, mostly from Europe and Canada, wanting to learn how to help their patients. He will be sorely missed by many.

Jeff was managed at home using a Medicaid waiver for up to $8,000 per month mainly for nursing services. Nursing home ventilator unit placement would have cost $25,000 to $30,000 per month. As an intelligent, self-directed individual, however, had Jeff been permitted to use personal attendant services rather than nursing, the cost could have been as low as the $2,400/mo that it is for one of our other DMD patients older than 40 yrs who is using continuous NIV support.

Jeff’s data are also in our in press publication on 101 continuously NIV-dependent DMD patients, of whom 39 were dependent for more than 10 yrs,

FIGURE 1 47.9-yr-old man with Duchenne muscular dystrophy using a 15-mm angled mouthpiece for daytime noninvasive ventilatory support in a regimen of continuous noninvasive support for 27 yrs.

FIGURE 2 The patient’s vital capacity, GPBmaxSBC, and MIC during a 20-yr period. GPBmaxSBC indicates glossopharyngeal breathing maximum insufflation capacity; MIC, maximum insufflation capacity.
In 1994, with a vital capacity of 100 ml, Jeff, intubated for the only time in his life for pneumonia and unable to satisfy any ventilator-weaning parameters or spontaneous breathing trials, was extubated back to NIV as have been more than 150 others in our center.8 Indeed, with 36 consecutive successful extubations of “unweanable” DMD patients, tracheostomy is not even a consideration in our center for these patients,3,8 nor in at least three other centers that routinely extubate “unweanable” DMD patients.8 This is fortunate, because tracheostomy is never preferred over noninvasive management when it is unnecessary, as it always appears to be, for people with DMD.15 Despite the overwhelming success of this approach, “unweanable” intubated DMD patients continue to be told that their only option for survival is tracheostomy, and physicians “caring” for these patients do not learn these methods.

This commentary was sent to the Muscular Dystrophy Association as a tribute to Jeff to be published in their patient magazine. Unfortunately, the editorial staff determined that, “The tone regarding NIV as the only way to manage ventilation in DMD is too prescriptive (and should only be) an individual voicing an opinion.” Therefore, permitting patients to avoid respiratory failure and the essentially complete avoidance of the need for tracheostomy tubes should be “toned down” so that patients do not solicit their physicians to learn noninvasive management. There are not multiple best ways to manage neuromuscular disease. There is only one “best” of anything. Is it not the time to open the prefrontal cortex to facts?16 How many more people must needlessly die prematurely or submit to respiratory failure, tracheostomy tubes, and lifetime institutionalization because of the failure of their physicians and organizations dedicated to help them to take the trouble to learn what can be done, spread the knowledge, and shift to more effective and humane treatment paradigms?

REFERENCES


3. Report to the 69th Congress of the Mexican Society of Respirologists and Thoracic Surgeons, 1st International Consensus on Diagnosis and Management of Respiratory Complications of Neuromuscular Disease, “Ventilación mecánica no invasiva, consensus,” April 6–9, 2010, Guadalajara, Jalisco, Mexico


7. Benditt J: Full-time noninvasive ventilation: Possible and desirable. Respir Care 2006;51:1005–12


