to gauge disabled patients' life satisfaction and potential for social and vocational productivity and refrain from letting their QOL judgments affect patient management decisions.

Conclusions

Children with SMA1 can be managed indefinitely by using CNVS and MIE. Few with SMA1 or milder neuromuscular disorders should require tracheostomies for long-term survival.

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COUNTERPOINT:

Is Noninvasive Ventilation Always the Most Appropriate Manner of Long-term Ventilation for Infants With Spinal Muscular Atrophy Type 1? No

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Among those physicians who offer ventilatory support to parents of children with SMA1, there seems to be a consensus that noninvasive ventilation (NIV) is the preferred method to provide that support.[1-3] An SMA standard of care document advocates for the use of NIV whenever possible in those infants whose families choose to provide ventilatory support.[4] Nevertheless, several groups advocate for the use of invasive ventilation via tracheostomy (IVTr) in a subset of their patients,[3,5,6] and many practitioners express a willingness to consider offering mechanical ventilation via tracheostomy to families.[1]

Infants with SMA1 who undergo tracheostomy placement may follow a different early course from

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those who do not undergo tracheostomy placement. Among the 42 (21.6%) infants with SMA1 in four Italian centers supported by IVTr, fewer displayed neurologic symptoms of SMA within the first 3 months of life than those who were supported with NIV, but onset of the first episode of respiratory decompensation occurred earlier (6.9 ± 4.3 months vs 12.6 ± 14.4 months; $P = .004$) in the IVTr group than in those supported noninvasively.[5] Similarly, in a French cohort, a greater percentage of infants with "intermediate type" SMA1 (presenting between 3 and 6 months of age) underwent tracheostomy placement compared with patients with the "true type" (presenting before 3 months of age).[7] Of the 20 infants with intermediate type SMA1 who underwent tracheostomy placement, one-half were initially supported with nocturnal NIV for 2.4 ± 2.6 months before switching to IVTr. Bach et al[8] reported their experience in caring for 92 children with SMA1, 27 of whom were treated with IVTr and 47 with NIV. No distinction was made between early and intermediate onset in that cohort. The group supported with IVTr experienced their first hospitalization at 6.1 ± 2.9 months, whereas those receiving NIV were first hospitalized at 8.6 ± 5.3 months; however, there was no statistical difference between the two groups in the age at first airway intubation for an acute respiratory illness. Following tracheostomy placement, the infants with IVTr were hospitalized significantly less frequently than the NIV group until 3 years of age but more frequently after age 5 years. Five of the patients underwent elective tracheostomy placement after having used NIV for 24 to 71 months; three patients required continuous NIV and experienced recurrent episodes of aspiration, and the other two experienced recurrent bradycardias associated with oxyhemoglobin desaturation. Two other children underwent tracheostomy placement, one because of congenital bronchomalacia and the other because of anoxic encephalopathy. According to their interim analysis, the investigators believed that the patients in the IVTr group were overall more severely affected than those treated with NIV.[9]

Although ventilatory support has been shown to improve survival in infants with SMA1,[10] how IVTr compared with NIV affects survival is less clear. Two studies confer a survival advantage to those patients treated with IVTr. Survival was significantly higher at 24 and 48 months in the group of children ventilated via tracheostomy compared with those ventilated noninvasively in a series from Italy.[5] In the French cohort, only six of 33 children with true SMA1 were still alive at the time of the publication, and all had undergone tracheostomy placement.[7] Five other children in that group who had undergone tracheostomy placement died between 19 months and 10.5 years of age (mean age, 5 years). Within the group of 35 children with intermediate type SMA1, three with tracheostomies died and six supported with NIV died. Causes of death in the patients with intermediate type SMA1 were not always determined but most often were related to secretion encumbrance and aspiration, regardless of whether a tracheostomy was present. In the series reported by Bach et al,[8] 18.5% of those infants supported by IVTr died, whereas 17% of those receiving NIV died. One of the infants receiving IVTr died 2 weeks after tracheostomy placement because he had experienced an anoxic event resulting from a mucus plug while he had been supported with NIV. Three of the infant deaths in the IVTr group were related to secretion encumbrance or atelectasis, whereas two of those treated with NIV died of airway obstruction from mucus plugs related to acute upper respiratory illnesses, three experienced profound bradycardia, and one developed intracranial hemorrhage while intubated for respiratory failure. Two others died of septic shock and a pulmonary embolism, respectively.

There are times that NIV can prove technically difficult. Nasal interfaces for infants are limited, and the paucity of different styles restricts practitioners' abilities to use interfaces with varying facial pressure points so as to avoid skin breakdown when continuous ventilation is required. Prolonged application of nasal masks ultimately can cause flattening of the midface and dental malocclusion with maxillary retrusion.[11,12] Although nasal prong systems are reportedly useful at supplying bilevel support,[13] the excessive leak that occurs when using such a system in an awake and weak infant often leads to poor patient-ventilator synchrony. In addition, trigger and cycle sensitivities of some bilevel generators may not be sensitive enough to detect the efforts of extremely weak infants.

As important as the limitations to ventilation are, difficulties with airway clearance in infants with SMA1 can prove life-threatening. It can be challenging to coordinate insufflation and exsufflation maneuvers with the breathing pattern of an infant who is both tachypneic and crying when an oronasal mask is placed on his or her face. Furthermore, the central airways of infants are more compliant than those of older children or adults[14]; exposure to exsufflation with negative

pressure can result in airway collapse. Bulbar dysfunction has been shown to cause significant pharyngeal collapse in adults with amyotrophic lateral sclerosis when undergoing MIE,[15] and the same bulbar dysfunction is present in some infants with SMA1. Thus, effective airway clearance can be difficult to achieve in some infants with SMA1, and even trivial upper respiratory illnesses can result in a need for airway intubation.[8,9,13]

A noninvasive approach to the management of an infant with SMA1 requires meticulous care, and parents and skilled caregivers must be comfortable in performing manual ventilation via a facemask as well as aggressive airway suctioning in the event of a mucus plug. Bach et al[13] noted that the approach would be unlikely to succeed if both parents had to work or if they were unable to learn the requisite skills. The need for multiple endotracheal intubations also carries risks. The effects of exposure of the developing human brain to multiple and often prolonged courses of sedation that are associated with recurrent intubation and mechanical ventilation episodes are not fully established, but there are concerns that such exposures are associated with subsequent neurocognitive deficits.[16,17]

In our center, a patient with SMA1 who requires ventilatory assistance and who lives at home will typically be approved by private or public insurance to receive 16 h per day of skilled nursing care, whether ventilation is by invasive or noninvasive means. Unlike ventilator-assisted adults who could receive much of their care at home from personal assistants,[18] no such reimbursable provision is available to young children. Instead, their skilled care is provided by registered nurses or licensed practical nurses. Because skilled nursing care represents the greatest cost of pediatric home ventilation care,[19] it is not clear that NIV results in significant cost savings over invasive ventilation in young children with neuromuscular diseases. Whether the presence of a tracheostomy results in more frequent hospitalizations in older children with SMA1 than those supported noninvasively[8] requires data from more than a single center.

Many practitioners question the ethics of offering long-term ventilatory support via tracheostomy to patients with SMA1, citing a concern that longevity, and not QOL, becomes the focus of care.[2,20-23] Such views are influenced by practitioners' training, experience and personal beliefs about pro-life interventions, and local culture and available resources.[1,2,4,23] It is critical that open lines of communication between parents and the care team be maintained, including frank discussions about the options for respiratory support. These discussions should cover benefits and repercussions of palliation, NIV, and tracheostomy placement. Simultaneously, unbiased assessments of the family's ability to provide care to the child and the community resources available to provide that care should be used to guide the discussion. Although such conversations are best conducted when the infant is well rather than during crises of care,[4,20] reassessment of decisions may be required based on the child's clinical course, including the need for recurrent airway intubation. It is reasonable to consider tracheostomy placement when parents of an infant with SMA1 have been informed about long-term care issues but are committed to providing ventilatory support, and the infant has required multiple airway intubations for acute respiratory illnesses or has experienced recurrent life-threatening episodes of airway obstruction from mucus plugs.

Tracheostomy placement in any infant carries tremendous repercussions regarding care, caregiver training, and resource utilization. Furthermore, IVTr in infants with SMA1 prevents neither progression of disease nor death. For the patient with SMA1, the role of IVTr is best summed up by the aphorism, "Never say never and always avoid always."

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Rebuttal From Dr Bach

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Dr Panitch's thoughtful commentary[1] deserves further consideration. The French and Italian users of NIV (ie, bilevel positive airway pressure [PAP]) who underwent tracheotomies included no CNVS users; thus, when needing more than sleep NIV or when intubated, they underwent tracheotomy. We routinely extubate "unweanable" patients with SMA1, including infants, to CNVS, thereby avoiding tracheotomies indefinitely provided that parents learn and administer CNVS and MIE.[2] Infants capable of 50-mL tidal volumes (intermediate type) invariably wean postextubation from CNVS to sleep NVS, for one-half of them until after 10 years of age,[3] and 80% develop functional speech. No patients with intermediate or severe SMA1 who undergo tracheotomy subsequently develop speech and few if any wean from continuous to sleep-only tracheostomy ventilatory support. Indeed, almost invariably in England and France, the infants with severe SMA1 who would require continuous support before 5 months of age die rather than receive CNVS or undergo tracheotomy. However, the medications and other treatments in the pipeline today[4] can only benefit the living.

Dr. Panitch noted that five of the patients in our series underwent tracheotomy after becoming CNVS dependent, but for three patients it was because they were out-of-state and not transferred to us for extubation.[1] He noted that in the Italian study, 4-year survival was greater for the tracheostomy ventilatory support users but, because they automatically placed tracheotomies for unweanable intubated infants (rather than extubate to CNVS and MIE), there were no CNVS users to compare them with; in the French study, only six of 33 patients with SMA1 survived until the time of publication, and even they were only a mean 5 years of age.[1] Currently, we have six CNVS users in our care who had no autonomous ability to breathe from as early as age 4 months who are now aged $>$ 20 years.

Dr Panitch[1] cited skin breakdown as a complication of CNVS but because all of the CNVS users in our series alternate interfaces, we have avoided this issue. He notes that although "nasal prong systems are reportedly useful at supplying bilevel support, the excessive leak that occurs…often leads to poor patient-ventilator synchrony." However, we stopped using bilevel PAP years ago because pressure preset NVS from ventilators is easier for weak infants to trigger for synchrony and to reverse paradoxing.[2]

When using MIE, airway collapse is transitory, which is why oxygen saturation levels return to baseline or greater several minutes after use. In addition, the reason MIE provides inadequate flows for bulbar amyotrophic lateral sclerosis but very effective flows for SMA1, especially after 3 years of age, is that upper motor

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