Respiratory Support in Spinal Muscular Atrophy Type I: A Survey of Physician Practices and Attitudes

M. Kathleen Moynihan Hardart, MD; Jeffrey P. Burns, MD, MPH; and Robert D. Truog, MD

ABSTRACT. Objective. To determine whether there is variability in the attitudes and practices of physicians regarding treatment of respiratory failure in children with spinal muscular atrophy type I (SMA type I) and, if so, whether this variation is associated with professional training.

Methods. This was a descriptive, cross-sectional survey mailed to a randomly selected subset of the Child Neurology Society, pediatric members of the Society of Critical Care Medicine and to the membership of the Pediatric Interest Section of the American Academy of Physical Medicine and Rehabilitation. A scenario of a child with SMA type I in respiratory distress was followed by questions that explored practices and attitudes regarding mechanical ventilation.

Results. Fifty-seven percent of intensivists (75 of 132), 39% physiatrists (61 of 155), and 34% of neurologists (61 of 155) responded. Specialists differed as to whether they offered and/or recommended respiratory support to patients with SMA type I. Intensivists were less likely to offer and recommend tracheostomy than physiatrists. Intensivists were also significantly less likely than physiatrists to agree with statements supporting the ethical necessity of noninvasive mechanical ventilation (NIMV) and intubation in the setting of an acute respiratory illness, and NIMV and tracheostomy in the setting of chronic respiratory failure. Although parallel differences were found between physiatrists and neurologists regarding their attitudes toward mechanical ventilation, no significant differences were detected between intensivists and neurologists. Finally, physicians who reported that a high percentage of their patients with SMA type I received “comfort care only” also tended to view mechanical ventilation, i.e., use of NIMV for chronic respiratory failure, use of intubation for an acute respiratory infection, and use of tracheostomy for chronic respiratory failure as an unreasonable intervention in most circumstances.

Conclusions. We found a wide variation in physician practice regarding the mechanical ventilation of patients with SMA type I. This study suggests a wide variation not only in what is recommended but also in what is actually offered to families of these children. Furthermore, the study suggests that physician training and attitudes affect recommendations regarding mechanical ventilation and ultimately family decision making.

ABBREVIATIONS. SMA, spinal muscular atrophy; NIMV, noninvasive mechanical ventilation.

Spinal muscular atrophy (SMA) type I, or Werdnig-Hoffman disease, is a devastating neuromuscular disease of childhood. Children with this disease typically have normal intelligence and remain alert and attentive to their environment, despite muscular weakness, and usually die from respiratory failure by age 2 years. Despite evidence from a small number of centers reporting success in prolonging life with mechanical ventilation, respiratory support for children with SMA type I is not widely accepted. Several important textbooks of pediatrics and pediatric neurology, for example, recommend against any mode of artificial ventilation for these patients. It is not clear whether these recommendations are based on the view that mechanical ventilation for these patients is not effective at sustaining life or that the quality of the life sustained is not of sufficient value. The purpose of this study was to describe the clinical practices of physicians regarding mechanical ventilation of patients with SMA type I. We also sought to explore the rationale underlying physician practices with the goal to understand better the basis for decision making. Our hypothesis was that wide variability exists in the care options presented and recommended to the families of children with SMA type I and that this variability may correlate with physician specialty, training, and attitudes.

METHODS

The study was designed as a cross-sectional mailed survey. We obtained membership lists of the Child Neurology Society and the Pediatric Section of the Society of Critical Care Medicine in September 1999. A cover letter that was signed by the principal investigators explaining the purpose of the study, along with an anonymous, self-administered questionnaire and a stamped return envelope, was mailed in November 1999 to a random selection of each membership list. In addition, all members of the Pediatric Interest Section of the American Academy of Physical Medicine and Rehabilitation were mailed a similar study packet. The questionnaire did not solicit any information that could link the responses to specific individuals. Nonresponders were tracked by the use of code numbers preprinted on return envelopes. A second questionnaire was mailed to nonresponders 3 months after the initial mailing. The study protocol and survey instrument were approved by the Children’s Hospital institutional review board.

The questionnaire (available on request) was developed with
### TABLE 1. Physician Attitudes Toward Mechanical Ventilation by Specialty*

<table>
<thead>
<tr>
<th>Statement</th>
<th>Response</th>
<th>Intensivists (N = 75)</th>
<th>Physiatrists (N = 61)</th>
<th>Neurologists (N = 51)</th>
<th>P Value</th>
</tr>
</thead>
<tbody>
<tr>
<td>SMA type I is a fatal condition and NIMV during a respiratory illness</td>
<td>Agree (%)</td>
<td>42</td>
<td>2</td>
<td>62</td>
<td>.006</td>
</tr>
<tr>
<td>just prolongs an inevitable death as a result of respiratory failure.</td>
<td>Neither agree nor disagree (%)</td>
<td>9</td>
<td>23</td>
<td>5</td>
<td>P vs N = .001</td>
</tr>
<tr>
<td>NIMV is a reasonable intervention for a child with SMA type I who has</td>
<td>Disagree (%)</td>
<td>49</td>
<td>55</td>
<td>33</td>
<td></td>
</tr>
<tr>
<td>respiratory failure in the setting of a respiratory illness.</td>
<td>Agree (%)</td>
<td>92</td>
<td>78</td>
<td>89</td>
<td>.04</td>
</tr>
<tr>
<td>NIMV is an ethically necessary intervention for a child with SMA type I</td>
<td>Neither agree nor disagree (%)</td>
<td>49</td>
<td>55</td>
<td>33</td>
<td></td>
</tr>
<tr>
<td>who has respiratory failure in the setting of a respiratory illness.</td>
<td>Disagree (%)</td>
<td>92</td>
<td>78</td>
<td>89</td>
<td>.04</td>
</tr>
<tr>
<td>NIMV is a reasonable intervention for a child with SMA type I who has</td>
<td>Agree (%)</td>
<td>67</td>
<td>47</td>
<td>60</td>
<td>.09</td>
</tr>
<tr>
<td>chronic respiratory failure.</td>
<td>Neither agree nor disagree (%)</td>
<td>63</td>
<td>78</td>
<td>58</td>
<td></td>
</tr>
<tr>
<td>NIMV is an ethically necessary intervention for a child with SMA type I</td>
<td>Disagree (%)</td>
<td>28</td>
<td>11</td>
<td>29</td>
<td></td>
</tr>
<tr>
<td>who has chronic respiratory failure.</td>
<td>Agree (%)</td>
<td>15</td>
<td>13</td>
<td>11</td>
<td>I vs P = .009</td>
</tr>
<tr>
<td>SMA type I is a fatal condition and intubation during a respiratory</td>
<td>Neither agree nor disagree (%)</td>
<td>10</td>
<td>25</td>
<td>20</td>
<td>I vs P &lt; .001</td>
</tr>
<tr>
<td>illness just prolongs an inevitable death as a result of respiratory</td>
<td>Disagree (%)</td>
<td>85</td>
<td>51</td>
<td>69</td>
<td></td>
</tr>
<tr>
<td>failure.</td>
<td>Agree (%)</td>
<td>51</td>
<td>41</td>
<td>61</td>
<td>.28</td>
</tr>
<tr>
<td>Intubation is a reasonable intervention for a child with SMA type I in</td>
<td>Neither agree nor disagree (%)</td>
<td>8</td>
<td>16</td>
<td>9</td>
<td></td>
</tr>
<tr>
<td>the setting of a respiratory infection.</td>
<td>Disagree (%)</td>
<td>41</td>
<td>43</td>
<td>30</td>
<td></td>
</tr>
<tr>
<td>Intubation is an ethically necessary intervention for a child with SMA</td>
<td>Agree (%)</td>
<td>78</td>
<td>78</td>
<td>63</td>
<td>.12</td>
</tr>
<tr>
<td>type I in the setting of a respiratory infection.</td>
<td>Neither agree nor disagree (%)</td>
<td>9</td>
<td>9</td>
<td>15</td>
<td></td>
</tr>
<tr>
<td>Redirection of care to comfort measures only is less likely once a</td>
<td>Disagree (%)</td>
<td>13</td>
<td>13</td>
<td>22</td>
<td></td>
</tr>
<tr>
<td>family has elected to intubate a child with SMA type I.</td>
<td>Agree (%)</td>
<td>7</td>
<td>15</td>
<td>13</td>
<td>.007</td>
</tr>
<tr>
<td>SMA type I is a fatal condition and tracheostomy just prolongs an</td>
<td>Neither agree nor disagree (%)</td>
<td>13</td>
<td>31</td>
<td>20</td>
<td>I vs P = .001</td>
</tr>
<tr>
<td>inevitable death as a result of respiratory failure.</td>
<td>Disagree (%)</td>
<td>80</td>
<td>54</td>
<td>67</td>
<td></td>
</tr>
<tr>
<td>Tracheostomy is a reasonable intervention for a child with SMA type I</td>
<td>Agree (%)</td>
<td>27</td>
<td>50</td>
<td>30</td>
<td>.009</td>
</tr>
<tr>
<td>and respiratory failure.</td>
<td>Neither agree nor disagree (%)</td>
<td>14</td>
<td>17</td>
<td>33</td>
<td>I vs P = .003</td>
</tr>
<tr>
<td>Tracheostomy is an ethically necessary intervention for a child with SMA</td>
<td>Disagree (%)</td>
<td>59</td>
<td>33</td>
<td>37</td>
<td></td>
</tr>
<tr>
<td>type I and respiratory failure.</td>
<td>Agree (%)</td>
<td>78</td>
<td>45</td>
<td>72</td>
<td>≤ .001</td>
</tr>
<tr>
<td>SMA type I is a fatal condition</td>
<td>Neither agree nor disagree (%)</td>
<td>8</td>
<td>22</td>
<td>15</td>
<td>I vs P &lt; .001</td>
</tr>
<tr>
<td>and tracheostomy just prolongs an inevitable death as a result of</td>
<td>Disagree (%)</td>
<td>14</td>
<td>33</td>
<td>13</td>
<td>P vs N = .006</td>
</tr>
<tr>
<td>respiratory failure.</td>
<td>Agree (%)</td>
<td>44</td>
<td>71</td>
<td>38</td>
<td>.002</td>
</tr>
<tr>
<td>Tracheostomy is a reasonable intervention for a child with SMA type I</td>
<td>Neither agree nor disagree (%)</td>
<td>10</td>
<td>10</td>
<td>13</td>
<td>I vs P = .002</td>
</tr>
<tr>
<td>and respiratory failure.</td>
<td>Disagree (%)</td>
<td>46</td>
<td>20</td>
<td>49</td>
<td>P vs N = .001</td>
</tr>
<tr>
<td>Tracheostomy is an ethically necessary intervention for a child with SMA</td>
<td>Agree (%)</td>
<td>1</td>
<td>12</td>
<td>4</td>
<td>&lt;.001</td>
</tr>
<tr>
<td>type I and respiratory failure.</td>
<td>Neither agree nor disagree (%)</td>
<td>5</td>
<td>25</td>
<td>7</td>
<td></td>
</tr>
<tr>
<td>Disagree (%)</td>
<td>94</td>
<td>63</td>
<td>89</td>
<td></td>
<td></td>
</tr>
</tbody>
</table>

* | I, intensivist; P, physiatrist; N, neurologist.  
* When Kruskal-Wallis $\chi^2 P < .05$, significant Wilcoxon pair-wise subgroup analyses are reported.

Special attention to clarity and the inclusion of the full range of response options by a local panel of experts in medical ethics, pediatric critical care, and survey research. The questionnaire was then pilot tested on 8 local physicians who practice pediatric neurology and pediatric critical care and who were not part of the final sample. The respondents of the pilot study were asked to rate each question on concept and clarity, and this feedback was used to make minor changes to the final instrument, resulting in improved content, construct, and face validity.

The questionnaire consisted of 16 questions divided into 3 parts and took approximately 8 minutes to complete. A scenario of a child who had SMA type I and was in respiratory distress from an acute upper respiratory illness was followed by questions that explored practices and attitudes regarding mechanical ventilation. As the scenario depicted the child’s clinical course over time, the question format allowed physicians to indicate their choice of intervention and attitudes in both acute and chronic respiratory failure. Responses to questions about a physician’s practice to offer and/or recommend different forms of mechanical ventilation were formatted as yes/no responses. Physician attitudes toward mechanical ventilation were reflected in 5-point Likert scale responses (strongly agree, agree, neither agree nor disagree, somewhat disagree, strongly disagree). Information regarding physician training, specialty, and experience with children with SMA type I was obtained by close-ended questions that asked physicians to indicate their answers from those provided. (For example, physicians were given the following options to answer the question, “What percentage of your SMA type I families have elected comfort care only?”: A) none B) 20% C) 50% D) 75% E) 100%

Statistical analysis was performed with version 10.0 of SPSS statistical software (SPSS, Inc, Chicago, IL). Yes/no questions exploring physician care practices were categorized with respect to the use of noninvasive mechanical ventilation (NIMV), endotracheal intubation, and tracheostomy. Physicians would 1) offer and recommend, 2) offer but not recommend, or 3) neither offer nor recommend the different forms of mechanical ventilation. The Likert scale data reflecting physician attitudes were categorized as agree, neither agree nor disagree, or disagree. Responses indicating specialty, training, and experience were recorded as nominal...
or continuous data. Pearson χ² analysis was used to detect associations between clinical practice pattern and specialty training. Kruskal-Wallis analysis was used to detect associations between 1) physician attitudes and specialty training and 2) physician attitudes and clinical practice pattern. When Pearson or Kruskal-Wallis χ² value reached .01 level of significance, subgroup Pearson or Wilcoxon χ² analysis between 2 specialties was performed, respectively, using 0.05 × 3 = 0.1667 level of significance. The Jonckheere-Terpstra test was used to detect association between physician attitudes and the respiratory support modes that their patients have elected. Surveys with incomplete responses account for the varying sample sizes in certain analyses. The maximum number of incomplete responses was 5.3.

RESULTS

Fifty-seven percent of pediatric intensivists (75 of 132), 39% of pediatric physiatrists (61 of 155), and 34% of pediatric neurologists (51 of 150) responded to the survey. Respondents reported the number of children with SMA type I for whom they have personally cared to be as follows: pediatric physiatrists—55% (0–5), 35% (5–15), 5% (15–30), 5% (>30); pediatric intensivists—25% (0–5), 59% (5–15), 12% (15–30), 5% (>30); pediatric neurologists—33% (0–5), 42% (5–15), 21% (10–15), 4% (>30). Overall, NIMV was offered and recommended by 70% (121 of 173), offered but not recommended by 22.5% (39 of 173), and neither offered nor recommended by 7.5% (13 of 173) of physicians. Intubation was offered and recommended by 39% (66 of 170), offered but not recommended by 49% (83 of 170), and neither offered nor recommended by 12% (21 of 170). Tracheostomy was offered and recommended by 27% (46 of 172), offered but not recommended by 49% (84 of 172), and neither offered nor recommended by 24% (42 of 172). Although no difference was appreciated between specialties with regard to their practices using NIMV or endotracheal intubation, there was a significant difference between specialties with regard to tracheostomy: 17% of intensivists (13 of 75), 42% of physiatrists (26 of 61), and 28% of neurologists (14 of 51) both offer and recommend tracheostomy; 58% of intensivists (43 of 75), 39% of physiatrists (24 of 61), and 44% of neurologists (23 of 51) offer but do not recommend tracheostomy; and 25% of intensivists (19 of 75), 19% of physiatrists (11 of 61), and 28% of neurologists (14 of 51) neither offer nor recommend tracheostomy (P = .04). On pair-wise analysis by specialty, intensivists were significantly less likely to offer and recommend tracheostomy than physiatrists (P = .009).

Associations between attitudes and specialty training are listed in Table 1. Subgroup analysis by specialty detected a consistently significant difference in attitudes toward all forms of mechanical ventilation between physiatrists and intensivists. Physiatrists were more likely than intensivists to agree with statements supporting the ethical necessity of NIMV (P = .009) and intubation (P = .001) in the setting of an acute respiratory illness, and NIMV (P < .001) and tracheostomy (P < .001) in the setting of chronic respiratory failure. Physiatrists were also more likely than intensivists to agree that tracheostomy is a reasonable intervention for children with SMA type I (P = .002) and to disagree that tracheostomy (P = .001) just prolongs inevitable death from respiratory failure. Finally, physiatrists were more likely than intensivists to disagree with the statement, “Redirection of care to comfort measures only is less likely once a family has elected to intubate a child” (P = .003). We also found significant differences between physiatrists and neurologists regarding their attitudes toward mechanical ventilation. Physiatrists were more likely than neurologists to disagree that NIMV (P = .001) and tracheostomy (P = .006) just prolong an inevitable death. In addition, physiatrists were more likely than neurologists to agree that tracheostomy is a reasonable (P = .001) and ethically necessary (P = .004) intervention in chronic respiratory failure. Of note, no significant differences in attitudes toward mechanical ventilation were detected between intensivists and neurologists.

We found significant associations between physician attitudes and their clinical practice. For example, 84% of respondents (65 of 77) who disagreed with the statement that “NIMV just prolongs inevitable death” were more likely to offer and recommend NIMV (P < .001). Seventy-six percent (117 of 154) of those who consider NIMV reasonable (P < .001) and 91% (42 of 46) of those who consider NIMV ethically necessary (P < .001) during an acute illness would offer and recommend NIMV. Eighty percent (90 of 113) of those who consider NIMV reasonable (P < .001) and 95% (20 of 21) of those who consider NIMV ethically necessary (P < .005) for chronic respiratory failure offer and recommend the intervention. Eighty-one percent (56 of 69) of those who consider extubation unlikely in SMA type I would not recommend intubation (P < .001). Ninety-six percent (120 of 125) of those who consider intubation reasonable (P < .001) and 94% (17 of 18) of those who consider intubation ethically necessary (P < .001) offer the intervention. Finally, 94% (79 of 84) of those who consider tracheostomy reasonable (P < .001) and 100% (11 of 11) of those who consider tracheostomy ethically necessary (P < .001) offer tracheostomy.

Physicians whose attitudes toward mechanical ventilation in SMA type I were favorable reported a smaller percentage of patients who elected comfort care only as demonstrated in Table 2. Not surprising then, those who considered NIMV for acute illnesses (P < .01), intubation (P < .001), and tracheostomy (P < .001) to be reasonable reported a higher percentage of patients who elected these respective modes of ventilation. In contrast, those who agreed that intubation (P < .001) and tracheostomy (P < .01) just prolong inevitable death and those who disagreed with the ethical necessity of intubation (P < .001) and tracheostomy (P < .001) reported a lower percentage of patients who elected the respective interventions.

DISCUSSION

During the past decade, there has been growing support for the practice of home mechanical ventilation for individuals with neuromuscular diseases such as Duchenne muscular dystrophy and amyotrophic lateral sclerosis.13–15 Conspicuously absent in this literature, however, is a comparable endorsement of mechanical ventilation for children with

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SMA type I. Our findings suggest that families of children with SMA type I are offered and recommended a wide array of options depending on where and from whom they receive their care. Although the majority of physicians in all 3 specialties surveyed would, in the setting of a respiratory illness, offer and recommend NIMV as well as offer endotracheal intubation, a significant number would not even present these interventions as options. We found the greatest practice variability in the utilization of tracheostomy and chronic mechanical ventilatory support for children with SMA type I. Nearly equal percentages of physicians would not offer as would both offer and recommend tracheostomy. It is interesting that intensivists were much less likely than physiatrists to offer tracheostomy as an option for patients.

Is the wide variation in practices based on ambiguity about the technical ability to provide long-term mechanical ventilation to these patients? Reports in the literature demonstrate that children with SMA type I can live for many years with long-term mechanical ventilatory support. For example, Japanese neurologists routinely treat children with SMA type I with long-term mechanical ventilation, citing the sanctity of life as an overriding principle guiding medical decision making in that country. In a national survey on the status of long-term ventilator-assisted children in Japan, Sakakihara et al found 55 children with SMA type I ranging from infancy to 14 years of age supported with mechanical ventilation. Although this survey does not provide details on the modes of mechanical ventilation used, it suggests that long-term support of respiratory failure in SMA type I is technically feasible. Similarly, in the United States, Bach et al reported their experience with 11 children with SMA type I over the course of 28 distinct episodes of respiratory distress. Those who could not be treated with NIMV and mechanically assisted cough devices were intubated and treated in accordance with a rigorous ventilator weaning protocol. All children were alive at follow-up 35 months after discharge. Two children did not require intubation or tracheostomy despite needing 24-hour assisted ventilation for >3 years, 2 children underwent tracheostomy and long-term mechanical ventilation, 6 children received only nighttime noninvasive support, and 1 child was lost to follow-up. In a separate report, Bach and Wang detailed the survival of some of their patients into their teenage years. These reports suggest that in addition to the traditional approach of providing comfort care only, long-term ventilation with either tracheostomy or noninvasive bi-level positive airway pressure has become a viable option for the support of chronic respiratory failure in children with SMA type I.

Divergent opinions about the practicality of prolonging life through mechanical respiratory support may reflect the variability of the natural history of SMA. The standard SMA classification system bases diagnosis of type I on age of onset of symptoms and predicts rapid neuromuscular deterioration. However, several studies have shown that prognosis based on this classification can be misleading, citing multiple examples of children who had onset of weakness before age 6 months and attained independent sitting skills and did not develop respiratory insufficiency for years. These children are ultimately reclassified as having SMA type II. In contrast, some children will experience a more aggressive and rapid deterioration complicated by severe restrictive kyphoscoliosis and repeated aspiration pneumonia. Because there are no clear genetic determinants that predict a more severe course, the classification remains clinical. Consequently, as the disease progression is different for each patient, measures that are successful in one child may not be effective in another and therapeutic interventions therefore must be tailored to each child’s individual needs. Variation in practice on the basis of the unique and differing needs of individual patients therefore might be both appropriate and necessary.

Our findings, however, suggest that the marked disparity in treatment recommendations for children with SMA type I is explained by variables other than heterogeneous disease progression. The greater tendency of physiatrists to offer and recommend tracheostomy when compared with intensivists suggests
that training background and or specialty significantly influence clinical practice. Physiatrists in general spend a greater percentage of their time caring for patients with chronic disability and perhaps have a better perspective on the possibility of long-term mechanical ventilation in these cases. In contrast, intensivists are more likely to see children when they are suffering in the context of an acute respiratory illness and may not appreciate the quality of their life in relative health. Similarly, neurologists who are generally involved in the diagnosis and perhaps not as often in the long-term care of the child with SMA type I may not be able to appreciate mechanical ventilation as anything more than a bridge to inevitable death. Variation in treatment recommendations may also reflect differences in physician attitudes. As physician attitudes were correlated not only with clinical practice but also with what families ultimately choose for their children, wide variations in attitudes likely translate into wide variations in care for children with this disease. As the care of children with SMA type I is not exclusively in the domain of 1 specialty, physicians who are involved with these children should be aware of the biases that they may bring to medical decision making. Moreover, physicians need to appreciate the powerful influence of their personal opinions on their clinical practice and families’ decision making.

Our study has several limitations. Although we did find significant differences among respondents relating to several of the study hypotheses, the overall response rate was low. Physicians who did respond to the survey may have had nonrepresentative attitudes and practices that biased our findings. In addition, responses to hypothetical scenarios may not correlate with actual practices and decision making.

The physician has a difficult and pivotal role in assisting family decision making regarding assisted ventilation of the patient with SMA type I. Although there is growing evidence that these children can have their lives significantly prolonged through the use of assisted ventilation, there is a real concern that they may have significant morbidity and suffer greatly from paralysis and respiratory compromise. We hope that the results of our study will heighten the awareness of those who care for children with SMA type I of the care options that are being offered to families and allow for better informed decision making. Future research efforts should aim to measure the quality of life among children who have SMA type I and receive long-term mechanical ventilation, to focus on discerning the feasibility of providing effective long-term ventilation to these children, and to identify those children who may benefit from such support.

REFERENCES

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