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Research Article

Spinal Muscular Atrophy Type 1 Quality of Life

ABSTRACT

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Objective: To compare healthcare professionals’ assessment of the quality of life of spinal muscular atrophy type 1 children with that of the care providers for the children.

Design: The care providers of all 53 surviving spinal muscular atrophy type 1 children managed in one neuromuscular disease clinic were sent Likert-scale surveys of six quality of life issues and ten polar-adjective pairs. The quality of life estimations were compared with those of 67 clinicians and with those of 30 parents considering their unaffected children.

Results: One hundred care providers from 46 out of the 53 families (87%) responded. Although the clinicians’ mean estimate of the children’s quality of life was $2.85 \pm 0.2/10$, the care providers’ estimate was $7.81 \pm 0.2/10$ ($P < 0.0001$). The care providers also found life with the children to be satisfying ($6.0 \pm 0.2/7$), interesting ($6.6 \pm 0.1/7$), friendly ($6.1 \pm 0.1/7$), enjoyable ($6.3 \pm 0.1/7$), worthwhile ($6.7 \pm 0.1/7$), full ($6.6 \pm 0.1/7$), hopeful ($5.9 \pm 0.2/7$), and rewarding ($6.4 \pm 0.1/7$), and they estimated the children to be happy ($8.5 \pm 0.2/10$) and their lives worth living ($9.6 \pm 0.1/10$). However, 69 of 104 felt that their lives were hard rather than easy, and 56 of 104 reported feeling tied down rather than free. Although the effort they felt for raising the child was high (8.3 ± 0.3 by comparison with 5 for an unaffected child), the burden they felt in doing so was not ($5.8 \pm 0.3/5$). When asked whether they would or would not recommend ventilator use, 31 clinicians (45.5%) indicated they would, 24 (36.4%) would not, and 12 (18.2%) chose not to respond to this question. Care provider responses did not differ significantly from the responses of the parents of unaffected children except for the easy/hard semantic differential (care providers, 3.80 ± 1.75 vs. controls, 5.27 ± 1.14 , $P < 0.001$).

Conclusions: Although there is a widespread perception that spinal muscular atrophy type 1 children have a poor quality of life, this perception is not shared by their care providers.

Key Words: Spinal Muscular Atrophy, Quality of Life, Neuromuscular Disease

The spinal muscular atrophies (SMAs) are inherited as autosomal recessive disorders of anterior horn cells with the genetic defect at chromosome 5q13. Gene deletions are detectable in 98% of patients. The prevalence is about 1 in 5000.¹ Severity is inversely proportional to the amount of survival motor neuron protein present in the anterior horn cells. It ranges from essentially total paralysis and need for ventilatory support from birth² to relatively mild muscle weakness presenting in the young adult.

The SMAs have been arbitrarily separated into four types based on clinical severity. SMA type 1 (SMA1) (Werdnig-Hoffmann disease) is defined by an infant who never attains the ability to sit.² However, occasionally, children with SMA who never attain the ability to sit can, nevertheless, roll and do not require enteral nutrition or develop acute respiratory failure before 2 yr of age. More typically, SMA1 children can never sit or roll, require enteral nutrition, and develop respiratory failure before age 2; they have paradoxical chest wall movement with inspiration, require at least ongoing nocturnal ventilatory assistance, and have only minimal finger and facial muscle movements. The most severe 10–15% of SMA1 patients require definitive continuous ventilatory support before 5 mo of age. For the purposes of this study, only typical and severe SMA1 patients will be considered. All these patients require nocturnal ventilator use but usually require continuous ventilatory support during the episodes of acute respiratory failure that are usually triggered by intercurrent upper respiratory tract infections.

Episodes of acute respiratory failure typically result in tracheotomy and long-term continuous ventilatory support or in death for these patients. In 2000, we reported an intensive care management protocol that permitted us to successfully ex-

tubate SMA1 children in ventilatory failure to noninvasive ventilatory support by using high span positive inspiratory pressure plus positive end-expiratory pressure (PIP+PEEP) delivered by BiPAP-ST devices (Respironics International, Murrysville, PA).³ Using nocturnal PIP+PEEP to prevent pectus excavatum, promote lung and chest wall growth, and provide nocturnal ventilatory assistance while avoiding oxygen supplementation and using mechanically assisted coughing were also important in the protocol. It was largely because of the child's familiarity with PIP+PEEP before developing respiratory failure that extubation to PIP+PEEP was so successful.

Members of pediatric sections of national medical societies, including 75 intensivists, 61 physiatrists, and 51 neurologists, responded to a survey regarding what their recommendations would be when faced with a SMA1 baby in respiratory distress. Noninvasive ventilation, an intervention that in itself could not sustain life, would be offered by 70% of the respondents, but it would be recommended by only 23% and neither offered nor recommended by 7%. Intubation would be offered and recommended by 38%, offered but not recommended by 48%, and neither offered nor recommended by 14%. Tracheotomy would be offered and recommended by 29%, offered but not recommended by 47%, and neither offered nor recommended by 24%.⁴ In another survey of 33 Japanese pediatricians, 80% considered quality of life (QOL) inadequate to justify survival, but about 50% said that they would begin ventilatory assistance for infant SMA patients. It was noted that strong familial endorsement, general pro-life beliefs, and secure medical funding might affect physicians' decisions in favor of providing life-sustaining treatments.⁵ Thus, when these children develop acute respi-

ratory failure during upper respiratory tract infections, more physicians recommend against rather than for intubation or tracheotomy, believing that QOL does not warrant taking life-sustaining measures. Even when the child in respiratory distress is intubated, after one or more failed extubations, reintubation is often refused and the patient dies.⁶ Other than for the subjects of our reports,^{3,7} patients satisfying the criteria for SMA1 noted above, and who do not undergo tracheotomy for continuous ventilatory support,⁸ die by 2 yr of age, have a median age of death of 7 mo, and have an 80% prevalence of death by 12 mo of age.²

Our center is currently managing 64 SMA1 patients. All of the parents of the 64 children reported to us having been informed by their physicians that their children would not live to age 2 without tracheostomy and that the burden, the effort involved in their care, and their QOL did not warrant this heroic measure. Eight patients died when parents or local medical staff decided not to provide vital respiratory interventions. Seventeen, with a mean age of 74 ± 57 mo, are managed by tracheostomy intermittent positive-pressure ventilation. One of the 17 died 3 mo after tracheotomy, and 15 of 16 are a verbal and require continuous ventilatory support. The oldest, 19 yr old, is a verbal and has been continuously ventilator dependent since 2 mo of age. Despite this, he graduated third in his high school class and is attending college. The remaining 39 children have been managed noninvasively using the intensive care protocol when ill,³ PIP+PEEP daily, and mechanically assisted coughing. Two died suddenly at 6 and 13 mo of age. The other 37 are now 41.8 ± 26.0 mo old. Fourteen children are over age 4 yr, eight are over the age of 5 yr, and one is 8 yr 3 mo old. Only five of the 37 chil-

TABLE 1*Estimation of quality of life (QOL) issues*

	Mothers	Fathers	Grandparents	Nurses	Deceased	Total
No.	44	30	8	20	4	104
Child's QOL? ^a	8.0 ± 0.2	7.3 ± 0.4	6.5 ± 1.2	8.4 ± 0.3	7.0 ± 0.6	7.8 ± 0.2
Your QOL? ^a	7.8 ± 0.3	7.4 ± 0.4	8.4 ± 0.8	8.7 ± 0.5	6.0 ± 0	7.9 ± 0.2
Effort to care for child? ^b	8.3 ± 0.3	7.8 ± 0.4	7.9 ± 1.0	9.2 ± 0.2	9.5 ± 0.3	8.3 ± 0.2
Burden of caring for child? ^b	5.1 ± 0.4	6.0 ± 0.5	3.9 ± 0.8	7.6 ± 0.5	8.0 ± 1.2	5.8 ± 0.3
How happy is the child? ^a	8.7 ± 0.2	8.3 ± 0.3	7.3 ± 1.4	9.0 ± 0.3	9.0 ± 0	8.5 ± 0.2
Child's life worth living? ^a	9.7 ± 0.2	9.6 ± 0.2	8.8 ± 0.8	9.6 ± 0.2	10.0 ± 0	9.6 ± 0.1

^aBy 0 to 10 Likert scale, with 0 = minimum and 10 = maximum.

^bBy 0 to 10, with 5 the average for unaffected children the same age.

dren have failed to develop the ability to speak, and only three require continuous PIP+PEEP. Even after age 10, SMA1 retain some facial and finger muscle function.

Respiratory management options now permit long-term survival for infants with SMA1. Their use, however, whether invasive or noninvasive, is not widespread. This is in large part because of the perceptions that prognosis and QOL are poor in this disorder. Because of these notions, we undertook a survey of the care providers of children with SMA1 to determine their estimates concerning the latter and compared

them with the estimates of health-care professionals.

PATIENTS AND METHODS

A study was made of all randomly encountered clinicians who came into contact with any one of the three authors in two hospital settings during an 8-hr day. They were recited the criteria for SMA1 (see "INTRODUCTION") and asked to fill out a form assessing the QOL of the typical 3-yr-old child with SMA1 using a Likert scale of 0–10 in which 0 is the minimum and 10 is the maximum. The healthcare professionals were also

asked their ages, whether the physician or parents should decide whether to provide ventilatory support for these children, and whether they would recommend it.

The parents and primary care providers of all 53 surviving children from the total of 64 SMA1 children managed by one author were sent surveys, and the parents of 30 small children consecutively solicited in a bowling alley were given surveys of the questions noted in Tables 1 and 2. The parents of two deceased children obtained the surveys on their own and also sent in four responses (from parents and grandparents). All data

TABLE 2*Semantic differential scales of life with the child*

SMA 1 Care Providers	Mothers	Fathers	Grandparents	Nurses	Total	
No. of respondents	45	31	8	20	104	<4
Unsatisfying–satisfying	5.8 ± 0.2	5.7 ± 0.3	5.9 ± 0.5	6.6 ± 0.4	6.0 ± 0.2	7
Boring–interesting	6.6 ± 0.2	6.7 ± 0.1	6.4 ± 0.4	6.8 ± 0.1	6.6 ± 0.1	0
Hard–easy	3.5 ± 0.3	3.7 ± 0.3	3.2 ± 0.6	4.8 ± 0.3	3.8 ± 0.2	53
Lonely–friendly	6.1 ± 0.2	6.2 ± 0.1	5.8 ± 0.5	6.3 ± 0.3	6.1 ± 0.1	9
Miserable–enjoyable	6.4 ± 0.1	6.1 ± 0.1	6.1 ± 0.4	6.7 ± 0.2	6.3 ± 0.1	2
Useless–worthwhile	6.8 ± 0.1	6.6 ± 0.1	6.7 ± 0.3	6.8 ± 0.1	6.7 ± 0.1	0
Empty–full	6.6 ± 0.1	6.6 ± 0.1	6.4 ± 0.4	6.7 ± 0.2	6.6 ± 0.1	0
Discouraging–hopeful	6.1 ± 0.2	5.6 ± 0.3	6.1 ± 0.4	6.5 ± 0.2	5.9 ± 0.2	10
Disappoint–rewarding	6.6 ± 0.1	6.1 ± 0.2	6.5 ± 0.4	6.8 ± 0.1	6.4 ± 0.1	1
Tied down–free	3.8 ± 0.3	3.9 ± 0.3	4.6 ± 0.3	4.9 ± 0.4	4.1 ± 0.2	40

SMA, spinal muscular atrophy.

The subjects (parents, grandparents, and nurses) were asked to indicate the extent that each heuristic dimension describes their lives with their affected children by indicating a number from 1 to 7, in which 1 and 7 reflect the extremes of the polar adjective pairs in a 7-point Likert-type scale (adapted from Campbell et al.¹⁵). The last column indicates the total number of responses <4. Data are given as mean ± SD.

TABLE 3*Significant differences in responses between care providers*

		Response Comparison			P Value	
QOL Issue						
Burden of caring	Fathers	5.11 ± 0.22	vs.	Mothers	6.02 ± 0.43	<0.05
QOL	Nurses	8.70 ± 0.50	vs.	Mothers	7.70 ± 0.30	<0.05
	Nurses	8.70 ± 0.50	vs.	Fathers	7.36 ± 0.40	<0.01
Child's QOL	Nurses	8.45 ± 0.33	vs.	Fathers	7.33 ± 0.38	<0.05
Child's happiness	Mothers	8.69 ± 0.21	vs.	Grandparents	7.25 ± 1.38	<0.05
Effort of caring	Nurses	9.25 ± 0.22	vs.	Mothers	8.27 ± 0.28	<0.05
	Nurses	9.25 ± 0.22	vs.	Fathers	7.77 ± 0.42	<0.01
Burden of caring	Nurses	7.60 ± 0.50	vs.	Mothers	5.10 ± 0.45	<0.01
	Nurses	7.60 ± 0.50	vs.	Fathers	6.03 ± 0.48	<0.01
Burden of caring	Grandparents	3.87 ± 0.81	vs.	Mothers	5.10 ± 0.45	<0.05
	Grandparents	3.87 ± 0.81	vs.	Fathers	6.03 ± 0.48	<0.01
Semantic differentials						
Hard–easy	Nurses	4.76 ± 0.30	vs.	Mothers	3.57 ± 0.27	<0.01
	Nurses	4.76 ± 0.30	vs.	Fathers	3.73 ± 0.33	<0.05
Tied down–free	Nurses	4.90 ± 0.40	vs.	Mothers	3.77 ± 0.26	<0.01

QOL, quality of life.

All data are mean ± SD.

are presented in mean ± standard deviation. Data were analyzed by one-way analysis of variance and, given the skewed distributions of the variables, the nonparametric Mann-Whitney *U* test to examine group differences. The Bonferroni correction factor for multiple comparisons, in this case 16, was used. This mandated a *P* value of <0.003 for statistical significance.

RESULTS

Out of a total of 64 SMA1 children managed in the clinic from 1996 to September 2001, the families of the 53 surviving children were sent surveys. We received 100 responses from 46 families (87% response rate) and four responses from the family of two deceased children. All parents of 30 unaffected children asked to fill out the questionnaire complied.

The responses of the children's healthcare providers to the six Likert-scale estimation of QOL issues and the ten polar-adjective pairs are noted in Tables 1 and 2. These 16 variables were compared across all four care provider groups (mothers,

fathers, grandparents, and nurses) via analysis of variance (F ratio = 24.60, *P* < 0.0001). There were no significant differences in the responses of the mothers, fathers, and other care providers other than for those noted in Table 3.

A *t* test was used to compare the differences in perception of QOL of SMA1 children between the children's care providers and the clinicians. The results showed a significant difference in this perception, with the 104 care providers estimating the children's QOL to be 7.8 ± 0.2, whereas the 67 clinicians estimated it to be 2.9 ± 0.2 (*t* = 16.007, *P* < 0.0001). Seven of 104 care providers and 66 of 67 clinicians estimated the children's QOL to be <5. When asked whether the physician or the parents should decide about the use of ventilatory support, six clinicians (9.1%) indicated the latter, 51 (77.3%) the former, five (6.8%) chose both, and five chose not to respond to that question. When asked whether they would recommend it, 31 clinicians (45.5%) indicated the former, 24

(36.4%) the latter, and 12 (18.2%) did not choose.

To determine whether the severity range of SMA1 might have affected care provider responses, the 29 care provider respondents for the 18 children too weak to speak or breathe without continuous ventilator use cited 7.7 ± 0.2/10 for their children's happiness, 9.6 ± 0.1/10 for the worthwhile nature of their lives, and 6.3 ± 0.4/7 for the satisfaction they derive from caring for the children. There were no significant differences in these mean values with those of the entire population of SMA1 care providers. The care provider responses were also compared with the responses of parents considering their 30 unaffected small children (Table 4).

DISCUSSION

This study demonstrated a significantly more positive estimate of the QOL of SMA1 children by their care providers than by clinicians in general. The care providers of these children noted that the children were very happy and that their lives were

TABLE 4*Response of 104 care providers vs. 30 parents of unaffected children*

	Care Providers	Controls	P Values
Age, mo	36.2 ± 35.3	28.17 ± 17.2	0.310
Quality of life (QOL) issues			
Child's QOL?	7.79 ± 1.91	8.67 ± 1.53	0.006
Your QOL?	7.89 ± 2.16	7.20 ± 2.57	0.016
Effort to care for child?	8.27 ± 1.99	7.21 ± 2.57	0.807
Burden of caring for child?	5.86 ± 2.84	6.00 ± 3.22	0.666
How happy is the child?	8.47 ± 1.80	8.60 ± 1.65	0.038
Semantic differentials			
Unsatisfying–satisfying	6.00 ± 1.40	5.97 ± 0.84	0.632
Boring–interesting	6.56 ± 0.91	6.53 ± 0.51	0.028
Hard–easy	3.80 ± 1.75	5.27 ± 1.14	0.000
Lonely–friendly	6.14 ± 1.37	6.20 ± 1.19	0.7444
Miserable–enjoyable	6.32 ± 0.98	6.33 ± 1.33	1.000
Useless–worthwhile	6.74 ± 0.54	6.53 ± 0.82	0.168
Empty–full	6.62 ± 0.75	6.33 ± 0.88	0.035
Discouraging–hopeful	5.91 ± 1.54	6.53 ± 0.63	0.173
Disappoint–rewarding	6.43 ± 0.96	6.53 ± 0.63	0.829
Tied down–free	3.99 ± 1.79	4.77 ± 0.90	0.032

very worthwhile. The effort needed for raising the children was not considered excessive. Even the nonrelative care providers (nurses) and the four responses from the families of the deceased children indicated that the children's lives had been happy and worthwhile. Likewise, the majority of the 104 care providers responded positively for all of the semantic differential responses (>4), except for the 53 who considered their lives hard. Indeed, other than for the easy/hard semantic differential, there were no other clearly significant differences ($P < 0.003$) between the responses of the care providers of the children with SMA 1 and the responses of parents considering their unaffected small children. It is possible that other differences might have become significant if a greater population of parents of unaffected children had been surveyed.

It is not surprising that mothers felt a significantly greater burden than fathers in caring for the chil-

dren nor is it surprising that the nurses felt that their own QOL was higher than the parents. The nurses also estimated greater effort and burden to raise the children and felt that their lives were significantly easier and less tied down than those of the parents. Thus, unlike non-care provider clinicians whose views were very negative, the nurses' views were very positive, but some were significantly less so than those of the parents.

This population of respondents represents a large sample of parents who have made the decision to follow a life-sustaining, noninvasive management protocol³ offered by the lead author of this study and requiring significant time and resources. In this sense, the sample may not reflect the general distribution of SMA families. Although there were 14 single mother family units, many of the single mothers had support from the child's father or grandparents. Families of other SMA1 children, particu-

larly single mother family units with no additional support, or families whose clinicians offer only a poor prognosis and fewer prevention or intervention strategies, are likely to feel more negative about the QOL of their children. In addition, SMA is a spectrum disorder. Although this study included some of the most severely affected children who could not speak or breathe without assistance, it is possible that our sample had fewer severely affected SMA1 children than average because some very severe children may die during the first few months of their lives before their parents can learn about these management strategies. Although not the case in this study, the families of the most severely affected SMA1 children may, in general, have reason to be less optimistic than families of less severely affected children.

These data demonstrate that the ongoing use of noninvasive ventilation was not considered an intolerable burden for the care providers. Indeed, studies of long-term ventilator users have revealed that ventilator use adds very little to the effort required for assisting severely disabled patients with their activities of daily living. Noninvasive approaches are also more convenient, safer, and less disruptive than invasive (tracheostomy) methods.^{9,10}

Although more clinicians in this survey favored rather than discouraged ventilator use, this is in contradistinction to the views of the majority of neurologists, both those cited in the INTRODUCTION of this article and those surveyed as directors of Muscular Dystrophy Association clinics, the majority of whom have discouraged the use of ventilators and 55% of whom cited poor QOL as the most frequent reason to do so for patients with neuromuscular diseases.^{11,12}

Purtilo¹³ summed up an article on ethical issues concerning the management of ventilator users by saying that misconceptions about the

undesirability of “going on a respirator’ have far-reaching negative effects for persons now happily being supported on a respirator, and mitigate the positive effects it could have for some types of chronically impaired persons whose QOL also could be enhanced by the use of a ventilator.” Freed¹⁴ stressed the importance of professionals not imposing their own concepts, values, and judgments onto the disabled person. Clinicians should be cognizant of their inability to gauge disabled patients’ QOL and refrain from letting inaccurate and unwarranted judgment of subjective issues associated with QOL in the general population affect patient management decisions.¹⁵

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