Assessing the Needs of the SMA Population: Survey Results of Health Care Providers and Families

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Abstract
To define issues of greatest importance to families and health care professionals (HCP) involved in caring for patients with spinal muscular atrophy (SMA), an electronic survey was distributed to patients, families, and HCPs to determine the most important issues facing patients with SMA. Seventy-seven patients/families (30 SMA type I, 36 SMA type II, 11 SMA type III) and 89 HCPs (17 pulmonologists; 10 physical, speech, and occupational therapists; 19 orthopedic surgeons, 12 neurologists; 13 nurses; 10 nutritionists; 8 others) completed the survey. Breathing issues, impact of diet, impact of disease on the family, spinal deformity, and surgical interventions were the five most frequently identified topics in order of importance to the patients/families. Important topics were similar for the HCP ranking those as follows: breathing issues, impact on family, spinal deformity, impact of diet, and impact of medications. This investigation reports the current most important perceived health care needs of patients with SMA. The perceived needs were similar between patients, families, and HCPs.

Keywords
SMA, FSMA, health care professional, needs, survey

Introduction
Spinal muscular atrophy (SMA) is an autosomal recessive genetic disorder affecting the motor neurons of the anterior horn cells due to the homozygous deletion of exon 7 of the survival motor neuron 1 (SMN1) gene and variable copy number of the modifying gene (SMN2). SMA is associated with progressive weakness and early mortality. Although SMA is a continuous clinical spectrum, children with SMA are classified into one of three types: type I has symptoms starting before 6 months of age, type II has onset between 7 and 18 months of age, and type III has onset of symptoms after 18 months of age (Munsat & Davies, 1992). Although the function and clinical course may vary, children with type I never sit and without intervention have a life expectancy <2 years; children with type II achieve sitting but do not walk and survive into the second decade. Children with type III ambulate and have a life expectancy into adulthood (Munsat & Davies, 1992). Pulmonary compromise is the primary cause of death in children with SMA type I or II (Chung, Wong, & Ip, 2004), due to restrictive lung disease (Iannaccone, 2007) and weak cough (Wang et al., 2007). In addition to the pulmonary complications, children with SMA suffer from gastrointestinal and orthopedic disorders (Granata, Merlina, Magni, Marini, & Stagni, 1989; Iannaccone, 2007; Rodillo, Marini, Heckmatt, & Dubowitz, 1989; Sporer & Smith, 2003; Wang et al., 2007). Although recommendations have been made regarding the care of patients with SMA, most information is based on expert opinion regarding a relatively small case series and has conflicting recommendations (Wang et al., 2007).

Although clinical advancements have been made in the care of children with SMA, many questions still remain. In nearly every aspect of care for these children (nutrition, pulmonary, rehabilitation medicine, and orthopedic), medical decisions are often based on the clinician’s or institution’s past experience or from small studies with low levels of evidence (Iannaccone, 2007; Wang et al., 2007). As a center that cares for a relatively large population of children with SMA, the investigators are actively developing a longitudinal multi-center, multi-disciplinary database in hopes to improve the care of these patients through evidence-based medicine.

To ensure that our future database was established to answer topics important to both clinicians and patient caregivers, a

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survey of both health care professionals (HCP) and patients was performed.

**Method**

A multi-disciplinary team experienced in the care of individuals with SMA developed a list of the nine important topics affecting this population. An electronic survey was constructed for patients and families, or HCPs to identify and rank five topics in order of importance. If topics of importance other than the nine listed were thought to be important, the survey allowed topics to be added via free text. The survey was distributed electronically to an informal SMA interest group, presented at the Families of SMA (FSMA) national meeting, and was distributed electronically to the FSMA list serve.

**Participants**

Although a rare disorder affecting <200,000 people in the United States (National Institutes of Health [NIH] Office of Rare Diseases Research), families of patients with this disorder are very motivated and organized to improve the care of their loved ones. An estimated 75% of patients with SMA belong to FSMA organization. After obtaining University of Wisconsin School of Medicine and Public Health (UWSMPH) Institutional Review Board (IRB) approval, the investigators approached the FSMA Medical Advisory Council to use the membership of FSMA as the basis of our sample population. Both HCP and patient/family belong to FSMA and the investigators would contact its members at their national annual meeting through handouts, on their website and social media page, and through an e-mail e-blast to their e-mail list serve. Additional surveys were sent to health care providers (N = 36) who had previously stated interest. Some of these health care providers may have received e-mails from both sources. While it is difficult to determine the absolute numbers of individuals who viewed the opportunity at the annual meeting or through the FSMA.org website/social media (an estimated 30,000 unique site visitors/month on the website and ~10,000 on social media), the e-mail was directly sent to 14,726 e-mail addresses through the list serve, of which 3,293 opened the e-mail. Roughly 13,000 of these e-mail addresses are patients and family members, whereas ~1,500 were HCPs.

**Survey**

Using a computer based survey program Qualtrics Survey Software (University of Wisconsin, Madison, Wisconsin), a multi-disciplinary team, including a pulmonologist, a physical therapist, and a pediatric orthopedic surgeon, experienced with the care of children with SMA, developed a list of nine important topics in the care of SMA. The survey was constructed for patients and families, or HCP could rank their five topics in order of importance. If topics other than the nine provided were thought to be important, the survey allowed these to be added via free text. The survey was presented to members attending the FSMA national meeting, distributed electronically to the FSMA list serve, and sent to other interested HCPs. The survey was left open for 50 days prior to data analysis. An example of the survey can be found in Figure 1.

**Statistical Analysis**

Data were automatically uploaded and stored within the University of Wisconsin Qualtrics Survey site. While it is difficult to know how many respondents filled out the survey as a result of the website, social media, annual meeting handouts, and direct HCP e-mail address, of the 14,726 e-mail addresses through the FSMA list serve, 3,293 opened the e-mail. (Roughly 13,000 of these list serve e-mail addresses are patients and family members, whereas ~1,500 were HCPs.) The total number of respondents starting and completing the survey was recorded. Two hundred twenty surveys were started (106 patient/families and 114 HCPs) and 156 completed the entire survey and were available for analysis. Using the number of opened e-mail list serve data and direct HCP addresses, an estimated response rate of 7% (220/3,329) was found for this survey, 5% (156/3,329) of whom filled out the survey to completion. As only fully completed surveys were analyzed, the completion rate was 71% (156/220). All free-text answers were reviewed by the multi-disciplinary study group to determine whether the free-text “additional” topics were new items to be considered or fell under the headings of one of the topic areas provided. Adjustments were then made to ensure that all free-text responses fitting under a provided topic were moved to that topic area and that the topic area was ranked appropriately. Descriptive statistical analysis of the responses was then performed. Topic importance was determined by ranking the frequency of being reported in the top five topics of concern for each person completing the survey.

**Results**

Seventy-seven patients/families (53 parents, 13 friend/relatives, and 11 affected individuals, 30 with SMA I, 36 with SMA II, and 11 with SMA III) and 89 HCPs (including 17 pulmonologists; 10 physical, speech, and occupational therapists; 19 orthopedic surgeons; 12 neurologists; 13 nurses; 10 nutritionists; and 8 others) completed the survey. Breathing issues, impact of diet and medications (tied), impact of disease on the family, spinal deformity, and surgical interventions were the five most important topics in order of importance to the patients/families. Important topics were similar for the HCP ranking those as follows: breathing issues, impact on family, spinal deformity, impact of diet, and impact of medications. Thus, topics of importance that
were similar between patients/families and HCPs were breathing issues, impact of diet, impact of disease on the family, spinal deformity, and impact of medications on disease as they were all rated as important to both groups.

Of the 156 completed surveys, only 18 additional free-text topics not included in the survey were added. Some of these “missed topics,” were similar among different respondents and could be grouped as “additional” important topics. For families, these included issues of fatigue, isolation, anxiety, contractures, disease progression rates, and cures. Additional HCP topics included rehab and therapy needs, learning and stimulation, independence issues, and counseling needs.

Discussion

SMA is a neurodegenerative disease with an incidence of 1/6,000 to 1/10,000 (Chung et al., 2004). Historically, children with SMA type I and type II died within the first two decades of life. With recent advances in pulmonary care and nutritional support, these children have an extended life span (Oskoui et al., 2007). With the increased life span, many questions arise as to how to best manage the medical needs of children with SMA (pulmonary, nutrition, rehabilitation medicine, orthopedic) and how to provide the patient and their families the best quality of life and resources needed. Scientifically answering any question at a single institution

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**Figure 1.** Example of the questionnaire filled out by participants.

*Note. SMA = spinal muscular atrophy; QOL = quality of life; GI = Gastrointestinal.*

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| 1) What type of SMA? (Families only) |
|-------------------------------------|
| a. Type I                          |
| b. Type II                         |
| c. Type III                        |

| 2) What is your Specialty/area practice? (Physicians only) |
|----------------------------------------------------------|
| a. Neurology                                            |
| b. Nursing                                              |
| c. Nutritionist                                         |
| d. Orthopedics                                          |
| e. Palliative Care                                      |
| f. Physiatry                                            |
| g. Physical Therapy, Occupational Therapy, and other    |
| h. Psychology/Mental Health Provider                    |
| i. Pulmonary                                            |
| j. Other                                                |

**Rank the following topics in order of importance (1 being most important)**

**Add additional topics if they are missing:**

A. Impact of SMA on family lives
   a. Missed work
   b. Number of clinic visits
   c. Costs of healthcare

B. Impact of Medications on pulmonary function, QOL, bone health
C. Impact of diet and dietary supplements on pulmonary function, QOL, bone health
D. Impact of smoke exposure on pulmonary function, bone health, life expectancy
E. Surgical interventions and QOL – orthopedic and GI
F. Bone Health – Weak Bone (Osteopenia), fractures, & medical treatments
G. Frequency of Pain & Pain Management
H. Breathing – respiratory treatments, medications, machines
I. Treatment of dislocated hips – risks, pain, and functional improvement
J. Treatment of spine deformity, e.g., bracing and/or surgery and their effects – pulmonary function, QOL, risks
K. Other 1 ___________________________________________
L. Other 2 ___________________________________________
M. Other 3 ___________________________________________
becomes difficult due to the relatively low incidence of the disorder and no evidence-based guidelines for management.

The purpose of this study was to provide investigators with baseline data from which to develop a multi-center database to begin answering some of the most important topics to both the patient/families and the HCPs who care for them. This may better help HCPs unfamiliar with SMA to better understand the needs of this complex population. In addition, the findings of this study identify areas to focus resources for further investigation by highlighting the topics most important to all parties (patient/families and HCPs) interested in improving the lives of these children. Through this study and the ultimate development of a multi-centered, multi-disciplinary database, the investigators look to fulfill the Institute of Medicine’s (IOM, 2009, p.5) call to identify ways to build an even stronger foundation of evidence-based medicine that effectively captures the promise of scientific discovery and technological innovation and enables doctors, nurses, and other health professionals to provide the right care for the right patient at the right time.

The similarity in responses of HCPs and patient/families demonstrates shared perceptions of SMA disease process. The free-text responses supplied important additional topics, which although not in the top five of order of importance, will be important to include in future studies. The additional free-text topics identified social and functional well-being issues of the child and his or her families. Data elements designed to assess and address these topics will be included in the multi-center database.

Limitations of this study should be pointed out. The survey provided to the families and HCPs may have overlooked issues. However, as free-text topics were allowed, this criticism is less warranted. Similarly, having HCP rank what items were most important may have been biased by the number of responses from a given specialty (i.e., pulmonary doctors may rank pulmonary issues as most important). However, as many specialists from many disciplines were involved in the survey and were allowed to rank the top five topics as opposed to the single “most” important topic, this bias is lessened. In addition, as the survey was left online for 50 days, the chance of “sharing” answers between respondents was possible. Due to this possible threat to internal validity, rank may not have been completely independent. Similarly, the low response rate to the survey may also be a valid criticism; however, the absolute number of respondents appears to be the largest to date dealing with this subject matter.

In conclusion, the survey results of the most important perceived medical needs of the SMA community are provided in this article. Topics important to patients/families and health care providers include breathing issues, impact of diet, impact of disease on the family, spinal deformity, and impact of medications on disease. Despite its limitations, this article provides insights into those areas of greatest concern for the SMA population.

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References
Battaglia, G., Princivalle, A., Forti, F., Lizier, C., & Zeviani, M. (1997). Expression of the SMN gene, the spinal muscular atrophy determining gene, in the mammalian central nervous system. Human Molecular Genetics, 6, 1961-1971.
Boda, B., Mas, C., Giudicelli, C., Nepote, V., Guimiot, F., Levacher, B., . . . Simonneau, M. (2004). Survival motor neuron SMN1 and SMN2 gene promoters: Identical sequences and differential expression in neurons and non-neuronal cells. European Journal of Human Genetics, 12, 729-737.
Chung, B. H., Wong, V. C., & Ip, P. (2004). Spinal muscular atrophy: Survival pattern and functional status. Pediatrics, 114, e548-553.
Granata, C., Merlini, L., Magni, E., Marini, M. L., & Stagni, S. B. (1989). Spinal muscular atrophy: Natural history and orthopaedic treatment of scoliosis. Spine, 14, 760-762.
Iannaccone, S. T. (2007). Modern management of spinal muscular atrophy. Journal of Child Neurology, 22, 974-978.
Institute of Medicine. (2009). Informing the future: Critical issues in health (5th ed.). Washington, DC: National Academy of Sciences.
Munsat, T. L., & Davies, K. E. (1992). International SMA consortium meeting. Neuromuscular Disorders, 2, 423-428.
Oskoui, M., Levy, G., Garland, C. J., Gray, J. M., O’Hagen, J., De Vivo, D. C., & Kaufmann, P. (2007). The changing natural history of spinal muscular atrophy type 1. Neurology, 69, 1931-1936. doi:10.1212/01.wnl.0000290830.40544.b9
Rodillo, E., Marini, M. L., Heckmatt, J. Z., & Dubowitz, V. (1989). Scoliosis in spinal muscular atrophy: Review of 63 cases. Journal of Child Neurology, 4, 118-123.
Sporer, S. M., & Smith, B. G. (2003). Hip dislocation in patients with spinal muscular atrophy. Journal of Pediatric Orthopedics, 23, 10-14.
Wang, C. H., Finkel, R. S., Bertini, E. S., Schroth, M., Simonds, A., Wong, B., . . . Trela, A. (2007). Consensus statement for standard of care in spinal muscular atrophy. Journal of Child Neurology, 22, 1027-1049. doi:10.1177/0883073807305788
Wirth, B., Brichta, L., Schrank, B., Lochmuller, H., Blick, S., Baasner, A., & Heller, R. (2006). Mildly affected patients with spinal muscular atrophy are partially protected by an increased SMN2 copy number. Human Genetics, 119, 422-428.
Zebala, L. P., Bridwell, K. H., Baldus, C., Richards, S. B., Dormans, J. P., Lenke, L. G., . . . Lovejoy, J. (2011). Minimum 5-year radiographic results of long scoliosis fusion in juvenile spinal muscular atrophy patients: Major curve progression after instrumented fusion. Journal of Pediatric Orthopedics, 31, 480-488.
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