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Original Article

Health-Related Quality of Life in Children and Adolescents With Spinal Muscular Atrophy in the Czech Republic

Helena Kocova MSc [a,*], Olga Dvorackova MSc [a], Petr Vondracek MD, PhD [b], Jana Haberlova MD, PhD [c]

[a] *Faculty of Health and Social Studies, University of South Bohemia, Ceske Budejovice, Czech Republic*
[b] *Medical Faculty, Masaryk University, Brno, Czech Republic*
[c] *Department of Child Neurology, 2nd School of Medicine of Charles University and Faculty Hospital, Praha 5, Czech Republic*

ABSTRACT

BACKGROUND: Spinal muscular atrophy is a rare hereditary neuromuscular disorder (with a prevalence of 1 per 30,000) that greatly debilitates patients and, in most cases, shortens their life expectancy. Although there is no causal therapy, improvements in symptomatic therapy have extended patients' life expectancy and increased their quality of life. Unfortunately, the advancements in care vary from country to country. To improve the care for children with spinal muscular atrophy in the Czech Republic, we created a survey to obtain the baseline information about their quality of life and compared the data with equivalent data from the United States. **METHODS:** We used the Pediatric Quality of Life Inventory 3.0 Neuromuscular Measurement Model, which is a health-related quality of life questionnaire specific to children with neuromuscular disorders. The survey was conducted on 35 children with genetically proven spinal muscular atrophy and their parents. **RESULTS:** Compared with the US data, the Czech data generally show a lower quality of life, mainly in the family resources part. The greatest score was achieved in the section about communication. Altogether, the parents' scores are lower than those of the children. **CONCLUSION:** In the Czech Republic, patients with spinal muscular atrophy and, especially their parents, have a significantly lower quality of life compared with US patients, mostly because of economic factors and a lack of social support. Our results reveal areas toward which improvement should be directed. The need for family support through social care as well as civic, patient, or organizational support is accentuated.

Keywords: health-related quality of life, neuromuscular disorders, spinal muscular atrophy, Czech Republic

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Introduction

Spinal muscular atrophy (SMA) is a severe neuromuscular disorder with autosomal-recessive inheritance that is clinically characterized by progressive, predominantly proximal muscle weakness and wasting.[1] Most patients are never able to walk independently and because of a weakness of intercostal muscles die prematurely from respiratory failure. With incidence of 1 in 6000 to 1 in 10,000 live births, it is the second most common fatal autosomal-recessive disorder after cystic fibrosis.[2]

The disease was first described clinically in the 1890s by Werdnig[3] and by Hoffmann.[4] The genetic defect was localized to 5q11.2-q13.3 a century later[5] in 1955 with the identification of the survival motor neuron (*SMN*) gene as the disease-causing gene.[6] Although the pathogenesis of the disease is still unknown, it is clear that there is a degeneration of alpha motor neurons in the spinal cord.[2]

SMA is clinically classified into four phenotypes on the basis of age of onset and motor function achieved.[7] Although there is some hope for possible future therapies in ongoing clinical trials,[8] there is no causal therapy to date. Nevertheless, the life expectancy and quality of life have increased greatly during the last two decades.[9,10] Despite the fact that the international standard of care has been published recently,[11] the standards of care and quality of life for patients with SMA in different countries still vary.[10]

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* Communications should be addressed to: Dr. Kocova; Kolping Family Smecno; Zizkova 1; Ceske Budejovice 37001, Czech Republic.
E-mail address: helena.kocova@dumrodin.cz

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So far, there are no data about the standard of care and quality of life of Czech patients with SMA. There are a few clinical data from the republic registry of patients with SMA that was established 2 years ago, which comprises only about 60 patients (less than one third of known cases). The goal of this study was to obtain baseline information about the health-related quality of life (HRQOL) in these patients as the first step to improve their health care.

Methods and Patients

Pediatric Quality of Life (PedsQL) 3.0 Neuromuscular Module

We used the PedsQL Inventory 3.0 Neuromuscular Module.[12] This scale is an HRQOL questionnaire specific to children ages 2-18 years with neuromuscular disorders that was recently validated for patients with SMA.[13] The PedsQL 3.0 Neuromuscular Module has 25 items and encompasses three scales: (1) About My Child's Neuromuscular Disease (17 items related to the disease process and associated symptomatology), (2) Communication (three items related to the ability to communicate with health care providers and others about his/her illness), and (3) About Our Family Resources (five items related to family financial and social support systems). The PedsQL Neuromuscular Module Scales comprise parallel child self-report and parent proxy-report formats for children ages 2-18 years. The scale is age-dependent and has four categories. The first category is for patients 2-4 years of age, and only the first part of the questionnaire on somatic functioning is filled out (the first 17 questions About My Neuromuscular Disease); other parts on Family and Communication are not evaluated because of the patients' young age. In the versions for children younger than 7 years of age, patients evaluate their quality of life on three-point Likert scale 0 = never, 1 = sometimes, and 2 = often with appropriate "smiley faces." Older respondents evaluate their quality of life by using a five-point Likert scale in response to the question "how big of a problem was the following for you in the past month," with 0 indicating never, 1 indicating almost never, 2 indicating sometimes, 3 indicating often, and 4 indicating almost always. Items are linearly transformed to a 0-100 scale (0 = 100, 1 = 75, 2 = 50, 3 = 25, and 4 = 0) so that greater scores indicate better HRQOL. The points in the form for the youngest children were transformed so that 0 equals 100, 1 equals 50, and 2 equals 0. Scale scores are computed as the sum of the items divided by the number of items that were answered. There are no upper or lower limit values that would determine how good or bad the quality of life is, but in general the greater the overall average value (arithmetic mean value), the greater quality of life.

SMA sample

The survey was conducted on children and adolescents with a genetically confirmed diagnosis of SMA who cooperated within the Early Care and the Support of Families with SMA project led at the civic association Kolping Family Smecno, as well as children and adolescents registered in the specialized centers office of pediatric neurologists—neuromuscular centers in Prague and Brno. In children younger than 8 years of age, the authors used interviews; otherwise, a self-administered questionnaire was used to collect the data. The questionnaires were distributed by the authors. Parental informed consent and child assent were obtained before enrollment. Parents and children completed the questionnaires separately.

The research group consisted of 35 children suffering from SMA type I (four children), type II (23 children), and type III (eight children) and one of their parents. The youngest child was 3 years of age and the oldest was 18 years. The first three age groups (2-4, 5-7, and 8-12 years) consisted of six, seven, and eight children, respectively. The largest age group represented was the category older than 12 years, with 14 children (40% of sample). With regard to gender, boys predominated over girls with 21 boys (60%) vs 14 girls (40%).

Statistical methods

Feasibility was determined from the percentage of missing values.[14] The internal consistency of the scale was determined by calculating Cronbach's coefficient alpha.[15] Scales with reliabilities of 0.70 or greater are recommended for comparing patient groups, whereas a reliability criterion of 0.90 is recommended for analyzing individual patient scores.[16]

Analysis of the main objective of the research—monitoring the degree of compliance of results of perception of quality of life in children with SMA and their parents—was (after being transferred to a 0-100 range) performed by calculating the arithmetic mean and S.D. Statistically significant differences between parents and children (with significance level $P < .05$) were identified with use of the nonparametric Mann-Whitney U test.[17] Data analysis was performed with the help of statistical program SPSS 16 (SPSS Inc., Chicago, IL).[18]

Results

The percentage of respondents with missing values was 8.6% in parents (3 of 35, all of them missing one item only) and 5.7% in children (2 of 35, both missing one item only). Table 1 summarizes the average values of parents and children's scores and internal consistency coefficients divided into the individual semantic parts of the questionnaire. All child self-report and parent proxy-report scales exceeded the minimum reliability standard of 0.70 required for group comparisons by Cronbach's coefficient. In the last column, there is a result of comparison between the present

TABLE 1.
Descriptive Statistics for the PedsQL 3.0 Neuromuscular Module Scores for Parent Proxy-Report and Child Self-Report and Comparison With Results of the US Study[12]

	Number of Items	n	α	Mean	S.D.	Difference CZ-United States
Parent						
Total score	25	35	0.90	52.08	16.37	−7.7
About My Child's Neuromuscular Disease	17	35	0.86	53.40	16.95	−5.4
Communication	3	35	0.83	62.38	29.94	−4.6
About Our Family Resources	5	35	0.82	41.39	21.64	−18.2
Child						
Total score	17/25	35	0.86	58.34	14.62	−9.2
About My Neuromuscular Disease	17	35	0.76	57.19	14.50	−8.8
Communication	3	22	0.74	71.97	20.34	1.1
About our family resources	5	22	0.85	56.82	22.65	−17.8

Abbreviations:
α = Cronbach's coefficient alpha
CZ = Czech Republic
n = number of parents/children answering the respective areas
PedsQL = Pediatric Quality of Life