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Original Article

Nutritional Status and Nutrient Intake Challenges in Children With Spinal Muscular Atrophy

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ABSTRACT

BACKGROUND: Nutrition is recognized as a core component of multidisciplinary care for patients with spinal muscular atrophy, but specific nutritional challenges in this population are not well described. We aimed to describe the nutritional status and nutrient intake in children with spinal muscular atrophy. **METHODS:** We performed a retrospective medical record review of prospectively collected data from children with spinal muscular atrophy followed at a multidisciplinary clinic at a tertiary referral center. We collected data including clinical parameters; anthropometrics, including weight, height, and body mass index (BMI); and 24-hour dietary intake records in all children followed in the clinic. Available data were found in records from the dietitian as part of a standard evaluation process, and additional clinical data were acquired from patient medical records. Subjects were classified based on spinal muscular atrophy type, and nutritional intake data were compared with dietary reference intakes for gender and age. *Z*-scores were calculated for weight for age (WAZ), height for age, and BMI (BMIZ) using the World Health Organization AnthroPlus software with appropriate World Health Organization reference growth standards. Subjects were classified as malnourished if their WAZ was <-2 or $>+2$. Anthropometric measurements were obtained at first visit and at a follow-up visit at an average of a 3-year interval between the clinic visits. A decline of more than 0.5 WAZ over this period was defined *a priori* as significant nutritional deterioration. **RESULTS:** We analyzed data from 60 subjects, 26 (43%) female, with median age 5.5 years (interquartile range 2 years to 12 years). The cohort consisted of children with spinal muscular atrophy type 1 (28 %), type 2 (45 %), and type 3 (27 %). At the first clinic visit, nine (15%) patients were malnourished. Thirteen (23%) subjects had a significant decline in WAZ from -0.35 (-1.31 to 0.58) to -1.04 (-2.15 to 0.02) at follow-up after approximately 3 years. A third of these subjects were already malnourished at first visit. A significant decline in BMIZ was noted in 47% of the cohort, and the prevalence of severe malnutrition (BMIZ < -3) increased from 2% to 17% after 3 years. In children receiving specialized enteral nutrition via a feeding tube, overfeeding was recorded in 29% and underfeeding was recorded in 35%. Suboptimal vitamin D intake was recorded in 35% of patients with enteral feeding device. **CONCLUSIONS:** Malnutrition was prevalent in children with spinal muscular atrophy, and nearly half the cohort demonstrated nutritional deterioration over time. Energy, protein, and vitamin D intakes were inadequate in a majority of the cohort. Underfeeding was highly prevalent, but overfeeding was also present in a third of the enterally fed cohort. Future studies describing optimal nutrient requirements and body composition variables in this group are required.

Keywords: spinal muscular atrophy, malnutrition, underfeeding, overfeeding, vitamin D

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Introduction

Spinal muscular atrophy (SMA) is a severe neuromuscular disease characterized by spinal motor neuron degeneration, resulting in progressive muscular atrophy and weakness.[1] With an estimated incidence between 1 in 6000 and 1 in 10,000 live births, it is the second most common fatal autosomal recessive disorder.[2] Although there have been significant improvements in other areas of clinical care, the delivery of optimal nutrition in patients with SMA remains challenging.[3] Children with SMA are at risk for both undernutrition and obesity, with significant risk of morbidity from both.[4] Significant lean body mass reduction and increased adiposity, secondary to disease process and the potential for unintended cumulative nutritional imbalance, may remain undetected despite seemingly normal weight gain in vulnerable children with chronic illnesses.[5] Hence, optimal nutritional needs, effective nutrient delivery, and reliable nutritional surveillance must be prioritized in children with SMA.

We aimed to examine the nutritional status, nutrient intake, challenges to optimal nutrition, and nutritional outcomes in a cohort of children with SMA. We hypothesized that malnutrition would be prevalent at baseline and worsen over time in this cohort. Furthermore, we expected energy and protein intake would be suboptimal and that there would be modifiable opportunities to improve nutrient delivery.

Materials and Methods

In a retrospective cohort study, we reviewed nutritional and clinical variables in children with SMA followed in a tertiary care, multidisciplinary clinic at Boston Children's Hospital. The study was approved by the Institutional Review Board at Boston Children's Hospital, and consecutive patients attending the clinic were included with no exclusions.

Clinical details, including demographics, disease characteristics, respiratory support, and hospitalizations were recorded, as well as gastrointestinal symptoms (i.e., vomiting, diarrhea, constipation) and difficulties with feeding via data collection from a full medical record chart review from the patient's initial visit to the most recent visit at the outpatient SMA clinic. Nutritional assessment including serial weight, height, and body mass index (BMI) were recorded at each clinic visit by a dedicated dietitian as part of a standard evaluation. In non-ambulatory patients, height was measured in a supine position using a flexible nonstretchable tape. *Z*-scores for weight for age (WAZ), height for age and BMI (BMIZ) were calculated using the World Health Organization AnthroPlus software and appropriate reference standards.[6] Growth was assessed by change in WAZ over time. A decline of more than 0.5 WAZ over an average period of 3 years on follow-up was *a priori* defined as significant change. Severe malnutrition (undernutrition) was defined as BMIZ < -3.

Dietary information was collected from a 24-hour dietary recall for oral feeding patients or based on the nutritional intake data for children with surgically implanted feeding devices. As part of routine clinical care the dietitian also provided recommendations when malnutrition issues were identified. Energy intake adequacy was determined based on the actual energy intake (AEI) as the percentage of estimated energy requirement (EER) by the Schofield equation[7] in subjects receiving nutrition via a specialized enteral feeding tube. Activity factors were not routinely calculated and were deemed to be unlikely to be significant among this cohort with muscular atrophy; hence basal metabolic rate was used as a measure of EER. Subjects were deemed to receive optimal energy intake if the ratio AEI:EER was between 90% and 110%, overfed if AEI:EER>110%, and underfed if AEI:EER<90%. For patients on oral intake, the dietitian classified these patients subjectively into three categories of underfed, adequately fed, and overfed, based on the dietary recall. Daily protein intake (g/kg) was compared with the age-based recommendations for daily intake values by the American Society for Parenteral and Enteral Nutrition.[8] Vitamin D and calcium intake was recorded where available and compared with daily-recommended intake (DRI) values.[9] Protein intake $< 90\%$ of the age-based recommendations from the American Society for Parenteral and Enteral Nutrition guidelines, and micronutrient intake <90% of the DRI value was deemed suboptimal.

Gastrointestinal symptoms, details of interval hospitalization, duration of mechanical ventilator support, and mortality before the three year follow-up were recorded from history and electronic medical records.

The primary outcomes for this study were (1) prevalence of malnutrition, defined as either underweight with WAZ < -2 or overweight with WAZ $> +2$, at baseline and at the average 3-year follow-up, based on World Health Organization Global Database on Child Growth and Malnutrition standards[10]; (2) percentage adequacy of energy and protein intake compared with recommended daily values; and (3) change in WAZ over time and the incidence of significant decline, defined as a WAZ decline >0.5 over 3 years.

Statistical analyses were performed using IBM/SPSS Statistics (version 21.0, IBM, Armonk, NY). Demographic and clinical data were tested for normality using the D'Agostino and Pearson normality test. Normally distributed data were presented as mean (S.D.) and non-normally distributed data as median (interquartile range [IQR]). Categorical data were presented as frequency (%). Statistical significance, where applicable, was set at $P < 0.05$. Paired *t* tests were performed to evaluate for significant changes between baseline and follow-up values of WAZ.

Results

We analyzed data from 60 subjects, 17 SMA type 1, 27 SMA type 2, and 16 SMA type 3. Descriptive data are presented in the Table.

The average number of hospitalizations since diagnosis was 2.0 (range 0-12) over the three year period, with an average length of hospital stay of 11 days (range 1-67). The overall mortality for the cohort was 20%. Twenty-two (37%) children were dependent on mechanical ventilator support at the time of this study, seven (12%) on tracheostomy and the remainder on noninvasive interface. SMA type 1

TABLE.
Clinical Characteristics of Cohort (N = 60)

Variable	N = 60
Age (in years), median (IQR)	5.5 (2-12)
Sex (M/F)	34/26
SMA type, n (%)	
1	17 (28.3%)
2	27 (45%)
3	16 (27%)
Ventilatory support	19 (31.7%)
Transtracheal ventilation	7 (37%)
Noninvasive ventilation	12 (63%)
Nutritional parameters	
WAZ (at first visit)	−0.35 (IQR −1.31 to 0.58)
BMIZ (at first visit)	0.015 (IQR −0.90 to 0.72)
WAZ (at follow-up visit)	−1.04 (IQR −6.81 to 2.55)
BMIZ (at follow-up visit)	−0.74 (IQR −8.36 to 2.55)
Route of feeding, n (%)	
Oral	43 (72%)
Gastrostomy	12 (20%)
Gastrojejunostomy	2 (3%)
Nasogastric tube	3 (5%)
Evidence of bulbar dysfunction	14 (23%)
Total mortality	11 (18%)
Mortality in patients with SMA type 1	11
Mortality in patients with SMA type 2	1