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Original Article

A Cross-sectional Study to Evaluate the Clinical Characteristics and Nutritional Status of Children with Spinal Muscular Atrophy

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ABSTRACT

Aim: This study aimed to evaluate the clinical characteristics, feeding features, and nutritional status of children diagnosed with spinal muscular atrophy (SMA) using multiple anthropometric indicators.

Materials and Methods: A cross-sectional observational study was conducted on 24 children (9 males, 15 females) with SMA, aged 5-192 months, in a tertiary care hospital in Türkiye. Clinical features, feeding methods, and anthropometric parameters including weight, height, body mass index, triceps skinfold thickness, and mid-upper arm circumference (MUAC) were recorded. Nutritional status was assessed using the Gomez and the Waterlow classifications, alongside MUAC z-scores based on Centers for Disease Control and Prevention growth charts.

Results: The cohort included 11 patients with SMA Type 1, 9 with Type 2, and 4 with Type 3. More than half (54.2%) were able to self-feed, while 33% were fed via percutaneous endoscopic gastrostomy. Feeding lasted longer than 15 minutes in 62.5% of cases. According to MUAC measurements, 58.3% of the patients were malnourished. Gomez classification identified 58.3% of the children as malnourished, and Waterlow classification indicated chronic malnutrition in 54.2%. A higher proportion of malnutrition was observed in those patients with SMA Type 1. MUAC measurements showed results comparable to traditional malnutrition classifications and emerged as a practical and reliable tool for nutritional assessment in SMA.

Conclusion: Malnutrition is highly prevalent among children with SMA, particularly in Type 1 patients. Comprehensive nutritional monitoring using MUAC alongside conventional anthropometric indices may improve the accuracy of malnutrition detection. A multidisciplinary approach, including early swallowing evaluations and timely nutritional interventions, is essential in order to optimize care and growth outcomes in this vulnerable population.

Keywords: Spinal muscular atrophy, malnutrition, upper arm circumference, anthropometric measurement

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Introduction

Spinal muscular atrophy (SMA) is a chronic neuromuscular disease characterized by spinal motor neuron degeneration (1). Its incidence is estimated as being between 1 in 6,000 and 1 in 10,000 live births (2).

SMA is currently divided into five subtypes: Type 0 (the most severe form with onset in the prenatal period and development of severe respiratory problems after birth), Type I (a severe form with onset before 6 months of age and the inability to sit unsupported), Type II (an intermediate form with onset before 18 months of age and the ability to sit unaided, but not to stand or walk), Type III (a mild form with onset after 18 months of age and the ability to stand and walk unaided), and Type IV (the mildest form with onset after 30 years of age) (3).

Although there have been significant improvements in the treatment of SMA, nutritional support is one of the main components in the guidelines for the standard of care in patients with SMA (4).

Common problems in SMA cases include feeding and swallowing problems such as weak sucking, limited opening of the mouth, and weak swallowing with dysfunctional airway protection (5). There is a considerable risk of morbidity from both undernutrition and obesity in children with SMA. Studies have shown that the prevalence of malnutrition is quite high, although it varies according to the type of SMA (6-8). Therefore, optimal nutritional needs, effective nutrient delivery, and reliable nutritional surveillance must be prioritized in those children with SMA.

Alongside a lack of a gold standard validated pediatric nutrition screening tool in routine clinical practice, nutritional assessment in SMA is further complicated by the challenges inherent in anthropometry in these children (8,9).

Thus, there is an ongoing effort to improve nutritional rehabilitation practice among children with SMA by using the most appropriate assessment tools in order to obtain a more accurate anthropometric profile and more realistic nutritional goals (10).

There is accumulating evidence that mid-upper arm circumference (MUAC) is the best case-detection method for pediatric malnutrition in terms of easiness, age independence, precision, accuracy, sensitivity, specificity and the lack of requirement for any special equipment, formal training or calculation (11,12).

This study aimed to assess the clinical characteristics, feeding features, and nutritional status of pediatric outpatients with SMA.

Materials and Methods

Participants

Patients had a confirmed diagnosis of SMA, were between one month and 18 years of age, and were enrolled in hospital-based clinical practices. This study was approved by Eskisehir Osmangazi University Non-Interventional Clinical Research Ethics Committee (approval no.: 28, date: 26.05.2023). Parents or legal representatives provided written, informed consent.

Study Design

This observational, cross-sectional study was conducted in a tertiary hospital in Türkiye.

Data Collection

In a single visit, information was gathered on the patients' anthropometric measurements, the results of their nutritional status assessment (using the Gomez and Waterlow classifications), date of birth, age at SMA diagnosis, etiology and their type of SMA. Information regarding disease-modifying therapy (nusinersen) was also recorded. All patients included in this study were receiving nusinersen treatment.

Nutritional Management

All of the patients received routine clinical care, including nutritional follow-up, as part of standard practice. Nutritional support and feeding methods [oral or percutaneous endoscopic gastrostomy (PEG)] were determined based on the clinical condition and needs of each patient. However, detailed data regarding specific nutritional interventions (such as caloric adjustments, dietary composition, or standardized dietitian-guided protocols) were not systematically recorded or analyzed within the scope of this study.

SMA Classification

SMA was clinically categorized into Types 1, 2, and 3 according to the established clinical criteria.

Anthropometrics

Anthropometric measurements included height (cm), body weight (kg), body mass index (BMI; kg/m^2), triceps skinfold thickness and MUAC along with estimations of mean z-scores and percentiles for weight-for-age (WFA), height-for-age (HFA), and weight-for-height (WFH) and these were then analyzed relative to the reference values established by Centers for Disease Control and Prevention (CDC) Growth Charts. MUAC z-scores were calculated using MUAC z-score reference standards. These standards were developed in order to assess pediatric malnutrition (13). Height measurements were obtained using standard clinical methods. In those patients with limited mobility or postural deformities, measurements were performed as accurately as possible under clinical conditions; however, alternative segmental measurements were not systematically applied. MUAC z-scores were calculated using the established reference data for pediatric populations based on age-appropriate standards. MUAC has been previously validated as a practical tool for assessing malnutrition in children, particularly in those with physical limitations.

Definition of Malnutrition

Malnutrition was defined according to the data of percentiles in the Gomez and Waterlow Classifications using CDC standard growth charts.

Gomez Classification

Based on WFA percentiles, the Gomez categorization scheme divides nutritional status into four categories: normal (≥90.00th percentile), first-degree/mild malnutrition (76th-90th percentiles), second-degree/moderate malnutrition (61st-75th percentiles), and third-degree/severe malnutrition (≤60th percentile) (14,15).

Waterlow Classification

The Waterlow categorization distinguishes between two types of malnutrition: "wasting," which is based on acute malnutrition and WFH, and "stunting," which is based on chronic malnutrition and height for age. Both the degree of stunting (the percentage of projected HFA) and wasting (the percentage of expected WFH) were calculated using the Waterlow Classification system. The severity of malnutrition cases was further classified using the WFH [overweight/obesity (≥110%), normal (≥90%), mild (80-89%), moderate (70-79%), severe (≤70%)] and HFA [normal (≥95%), mild (90-94%), moderate (85-89%), severe (≤85%)] parameters (14,15).

Statistical Analysis

The statistical analyses were performed using the Statistical Package for the Social Sciences SPSS version 11.0 (SPSS Inc., Chicago, IL, USA), which was available in our institutional system. Due to the relatively small sample size, particularly within the SMA subtype groups, inferential statistical analyses were not performed, and the results are presented descriptively.

Results

Demographic Parameters and Clinical Features of the Patients

This study was conducted with 24 (9 males, 15 females) children with SMA. The mean age of the patients was 90.67±50.72 months (5-192). Eleven children were diagnosed as SMA Type 1 thus representing the most severe form of SMA.

Hammersmith Functional Motor Scale (HFMS) data were available for 24 patients, whereas Children's Hospital of Philadelphia Infant Test of Neuromuscular Disorders (CHOP-INTEND) data were available for 15 patients. The mean HFMS and CHOP-INTEND scores were 36.18±17.08 and 37.85±21.21, respectively. The types of children with SMA are summarized in Table I.

More than half of the study group were able to self-feed (54.2%). Sixteen children had had a tracheostomy. These patients were under respiratory support as part of their clinical management; however, detailed data regarding the type and duration of ventilation were not analyzed in this study. The PEG was the route of feeding in 33.3% of cases (Table II). The distribution of PEG feeding varied across the SMA subtypes, with a higher frequency observed in the more severe forms; however, due to the limited sample size, subgroup comparisons were not performed. All of the patients included in this study were under nusinersen treatment at the time of evaluation.

Anthropometrics and Nutritional Status of the Study Population

The anthropometric measurements of the study group are presented in Table III.

The results of the MUAC measurements revealed that 14 (58.3%) children had malnutrition and one patient was overweight. The results of the MUAC measurements are presented in Table IV.

In general, the Gomez classification (WFA) revealed that malnutrition was evident in 58.3% of the children with SMA based on the CDC growth charts. According to the Waterlow (stunting) classification of HFA based on the CDC growth charts, 54.2% of the patients were chronically malnourished (Table V).

Table I. Clinical characteristics of study group

Type of SMA	SMA patients, n (%)
Type 1	11 (45.8)
Type 2	9 (37.5)
Type 3	4 (16.7)

SMA: Spinal muscular atrophy

Table II. Feeding features of children with SMA

Feeding-related characteristics	SMA patients, n (%)
Duration of feeding	
<15 min	9 (37.5)
15-30 min	13 (54.2)
>30 min	2 (8.3)
Self-feeding ability	
Self-fed	13 (54.2)
By caregiver	11 (45.8)
Feeding method	
Orally	16 (66.6)
PEG feeding	8 (33.3)

Feeding duration data were collected for all patients in the study cohort, regardless of feeding method.
PEG: Percutaneous endoscopic gastrostomy, SMA: Spinal muscular atrophy

Table III. The anthropometric measurements of patients

Anthropometric variables	All SMA Types	SMA Type 1	SMA Type 2	SMA Type 3
Mean Age				
Mean	90.67	47.18	117.78	149.25
Median	80.00	25.00	130.00	164.50
Std.	59.72	35.97	49.92	51.71
Min.	5	5	41.00	76
Max.	192	107	182.00	192
Height				
Mean	114.50	95.73	123.78	145.25
Median	114.00	85.00	117.00	149.50
Std.	30.08	22.56	25.42	26.61
Min.	64	64	85	109
Max.	173	133	162	173
Height Z Score				
Mean	-1.25	-0.55	-2.11	-1.08
Median	-1.20	-0.50	-2.05	-1.31
Std.	1.43	1.18	1.54	0.74
Min.	-4.33	-2.23	-4.28	-1.65
Max.	1.46	1.54	0.08	-0.07
Weight				
Mean	24.68	13.44	30.51	42.50
Median	19.50	12.00	22.00	47.50
Std.	16.69	6.34	16.36	17.33
Min.	5.0	5.0	10	18.0
Max.	57.0	26.0	52	57.0
Weight Z Score				
Mean	-1.73	-1.88	-1.55	-0.46
Median	-1.59	-1.43	-0.02	-0.26
Std.	2.35	1.46	3.29	0.62
Min.	-9.72	-5.29	-9.28	-1.37
Max.	1.19	-0.16	1.23	0.04

Table III. Continued

Anthropometric variables	All SMA Types	SMA Type 1	SMA Type 2	SMA Type 3
BMI				
Mean	16.51	14.07	18.64	19.16
Median	15.63	13.51	19.02	19.34
Std.	4.65	2.48	5.74	3.14
Min.	8.77	10.16	8.90	15.15
Max.	28.40	18.67	28.40	22.82
BMI Z Score				
Mean	-1.08	-1.69	-0.64	0.09
Median	0.23	-0.45	0.35	0.09
Std.	3.09	2.34	3.79	0.62
Min.	-10.65	-5.93	-10.18	-0.60
Max.	2.58	0.18	2.50	0.81
MUAC				
Mean	17.87	15.11	19.80	22.25
Median	18.00	15.00	22.00	24.00
Std.	4.49	2.05	5.64	4.19
Min.	10.00	13.0	10.00	16
Max.	25.0	18.0	25.00	25
TSFT				
Mean	11.04	10.82	11.33	11.00
Median	12	12.00	12.00	11.50
Std.	3.65	2.78	5.26	1.41
Min.	2	4	2	9
Max.	18	13	18	12

Std: Standard deviation, Min: Minimum, Max: Maximum, BMI: Body mass index, MUAC: Mid-upper arm circumference, TSFT: Triceps skinfold thickness

Table IV. The results of z-score of bands MUAC and other anthropometric measurements

Anthropometric measurements	+2 to +1	+1 to 0	0 to -1	-1 to -2	-2 to - 3	-3 to -4
MUAC Z Score						
All types	1	1	8	8	3	3
Type 1	0	0	5	2	3	1
Type 2	1	1	2	2	0	2
Type 3	0	0	1	3	0	0
Weight Z Score						
All types	1	4	7	5	4	3
Type 1	0	0	3	3	4	1
Type 2	1	3	2	1	0	2
Type 3	0	1	2	1	0	0
Height Z score						
All types	1	4	7	6	4	2
Type 1	1	3	3	3	1	0
Type 2	0	1	2	1	3	2
Type 3	0	0	2	2	0	0
BMI Z score						
All types	2	9	4	1	1	3
Type 1	0	3	1	0	1	2
Type 2	2	4	1	1	0	1
Type 3	0	2	2	0	0	0

Totals may not correspond to subgroup sizes in all categories because complete anthropometric data were not available for all patients.
MUAC: Mid-upper arm circumference, BMI: Body mass index

Table V. Assessment of nutritional status of the study group

	Nutritional status	Gomez Classification WFA CDC percentile	Waterlow Classification WFH CDC percentile	Waterlow Classification HFA CDC percentile	MUAC
SMA All Types		**n**	**n**	**n**	**n**
	Normal/Overweight	10	15	11	10
	Malnourished	14	4	13	14
	1st degree (n) **2nd degree (n)** **3rd degree (n)**	7 5 2	2 1 1	5 5 3	8 3 3
	Total	24	19	24	24
SMA Type 1	**Nutritional status**	**Gomez Classification WFA CDC percentile**	**Waterlow Classification WFH CDC percentile**	**Waterlow Classification HFA CDC percentile**	**MUAC**
		n	**n**	**n**	**n**
	Normal/Overweight	4	4	7	5
	Malnourished	7	2	4	6
	1st degree (n) **2nd degree (n)** **3rd degree (n)**	3 3 1	1 1 0	2 1 1	2 3 1
	Total	11	11	11	11
SMA Type 2	**Nutritional status**	**Gomez Classification WFA CDC percentile**	**Waterlow Classification WFH CDC percentile**	**Waterlow Classification HFA CDC percentile**	**MUAC**
		n	**n**	**n**	**n**
	Normal/Overweight	5	2	3	4
	Malnourished	4	7	6	5
	1st degree (n) **2nd degree (n)** **3rd degree (n)**	1 2 1	5 1 1	0 4 2	3 0 2
	Total	9	9	9	9
SMA Type 3	**Nutritional status**	**Gomez Classification WFA CDC percentile**	**Waterlow Classification WFH CDC percentile**	**Waterlow Classification HFA CDC percentile**	**MUAC**
		n	**n**	**n**	**n**
	Normal/Overweight	1	1	1	1
	Malnourished	3	3	3	3
	1st degree (n) **2nd degree (n)** **3rd degree (n)**	3 0 0	3 0 0	3 0 0	3 0 0
	Total	4	4	4	4

Waterlow classification based on WFH was calculated only for patients with available WFH measurements (n=19). Therefore, totals in this column do not correspond to the full cohort size (n=24).
MUAC: Mid-upper arm circumference, WFA: Weight-for-age, HFA: Height-for-age, WFH: Weight-for-height, CDC: Centers for disease control and prevention

Discussion

SMA affects multiple systems, despite it being primarily a chronic neurological condition (16). Therefore, the treatment of SMA must be planned in a multidisciplinary manner. Nutritional support is one of the main components of the guidelines for the standard of care in patients with SMA (4). This study presents a cross-sectional evaluation of the clinical features and nutritional status of children diagnosed with SMA, emphasizing the high prevalence of malnutrition and the factors influencing nutritional health in this population. According to the Gomez classification based on WFA, 58.3% of the patients in this study were malnourished. Similarly, the Waterlow classification using HFA indicated chronic malnutrition in 54.2% of the cohort.

The consistency across these different methods highlights this issue and underlines the importance and urgency of addressing malnutrition in children with SMA. A higher proportion of malnutrition was observed in those patients with SMA Type 1, the most severe phenotype in our study, suggesting a possible association between disease severity and nutritional status. MUAC z-score analysis also identified 58.3% of the children as being malnourished. De Amicis et al. (9) conducted a study with Type 1 and Type 2 cases which revealed that mean weight was significantly lower in SMA patients than in healthy controls, while supine length was more variable. They also present a set of disease-specific percentile curves of BW, SL (supine length), and BMI-for-age for girls and boys with SMA Type 1 and SMA Type 2. However, these specific curves had not yet been approved by ESPGAN as of the time of writing. During follow-up, SMA patients should be monitored in terms of their nutritional status. A retrospective study carried out with sixty cases reported that weight z-scores had decreased in 23% of patients. This ratio was 47% in terms of BMI (17).

Feeding and swallowing problems are one of the most important complications of SMA. Dysphagia in SMA Type 1 is described as disturbed and weak sucking, problems with handling oral secretions, weak swallowing with dysfunctional airway protection, and gastroesophageal reflux. Dysphagia in SMA Type 1 may also lead to other problems such as poor weight gain, discomfort and risk of aspiration pneumonia. Choking and sweating during feeding were reported as being 91% and 55% in SMA patients, respectively. It has been reported that 72% of patients had to interrupt feeding due to coughing attacks and sweating (18). As a result, the duration of feeding is prolonged. In our study, 62.5% of the participants reported feeding durations of longer than 15 minutes. PEG is an alternative for children with feeding difficulties. The present study revealed that only eight cases were feeding via PEG. This result was lower than has been reported in the literature (19). This difference may be influenced by several factors, including clinical decision-making processes, parental preferences, and the timing of PEG recommendations in routine practice. This point might be one of the reasons why the prevalence of malnutrition was high in present research (20).

Standard anthropometric measurements can be challenging in children with SMA due to spinal deformities (e.g., scoliosis), joint contractures, and altered body composition. In this context, MUAC stands out as a practical and reliable tool for nutritional assessment. In our study, MUAC showed comparable results with traditional malnutrition classification systems and offered significant advantages in terms of its usability, age independence, and minimal technical requirements. Prior studies have similarly highlighted MUAC as an effective screening tool for pediatric malnutrition, especially in populations with physical limitations (21,22). Additionally, the use of standard CDC growth charts may overestimate malnutrition in children with SMA, as reduced muscle mass and altered body composition may not necessarily reflect true caloric deficiencies.

In recent years, dramatic and promising developments have occurred in the treatment of SMA. Although some studies have reported improvements in dysphagia, it is not dramatic as motor functions (6,18). When considering nusinersen, which is the most commonly used drug, studies have suggested that this drug may have less effect on the brainstem than on other parts of the spinal cord (23-25).

Study Limitations

In the present study, all of the cases were being treated with nusinersen. However, detailed treatment-related variables (e.g., duration of therapy, dosing intervals) were not analyzed, which may be considered a limitation. This study had several limitations. The small sample size and single-center design may limit the generalizability of the findings. Moreover, the cross-sectional nature of this study precludes analysis of longitudinal changes in nutritional status or the effects of interventions over time. The lack of detailed data on nutritional interventions may be considered as another limitation of this study. In addition, the inclusion of different SMA types in a single analysis may also be considered as a limitation, as clinical severity and functional status vary significantly between SMA types, which may have influenced the nutritional findings and their interpretation. Nevertheless, the use of multiple anthropometric indicators and validated classification systems strengthens the credibility of our findings. In addition, the lack of inferential statistical analyses may be considered as a limitation of this study. Additionally, anthropometric measurements such as height may be affected by postural deformities, including scoliosis and joint contractures, which are common in SMA. Furthermore, standard MUAC reference values may not fully reflect body composition in children with SMA due to their reduced muscle mass. Additionally, potential differences in the nutritional statuses between PEG-fed and orally fed patients were not analyzed. An analysis of these differences in nutritional statuses in future studies may provide further insight into the issues affecting this population.

Conclusion

In conclusion, the management of nutrition in SMA should not be limited to dietitians. A multidisciplinary approach involving pediatric neurologists, gastroenterologists, rehabilitation specialists, and speech and swallowing therapists is essential. Early swallowing evaluations and a timely transition to enteral feeding in at-risk children can prevent further nutritional decline. Periodic reassessment using multiple anthropometric tools and functional indicators can help personalize interventions and improve outcomes. Further large-scale, prospective studies are warranted in order to evaluate the long-term progression of nutritional status in SMA patients and to assess the effectiveness of targeted nutritional interventions in improving both growth and functional outcomes.

Ethics

Ethics Committee Approval: This study was approved by Eskisehir Osmangazi University Non-Interventional Clinical Research Ethics Committee (approval no.: 28, date: 26.05.2023).

Informed Consent: Parents or legal representatives provided written, informed consent.

Footnotes

Authorship Contributions

Surgical and Medical Practices: K.B.Ç., A.D.Y.Ş., Concept: K.B.Ç., A.D.Y.Ş., Design: K.B.Ç., A.D.Y.Ş., Data Collection or Processing: A.D.Y.Ş., G.K.Y., Ö.U., C.Y., Analysis or Interpretation: E.A., Literature Search: K.B.Ç., Writing: K.B.Ç.

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