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Telenutrition Support in Rare Diseases May Have Positive Effects: Spinal Muscular Atrophy Type 2 Case Report

Nadir Hastalıklarda Telenütrisyon Desteği Olumlu Etkiler Oluşturabilir: Spinal Musküler Atrofi Tip 2 Olgu Sunumu

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ABSTRACT

In this case report, the 6-month telenutrition counseling process of a 13-year-old male patient diagnosed with spinal muscular atrophy type 2 was discussed. Severe malnutrition was observed in the patient, who used a wheelchair and was fed orally. As a result of examining all the findings, a 6-month telenutrition counseling process was started with a diet containing 50 kcal/kg of energy and 1.85 g/kg of protein per day. At the end of the process, the patient's body weight increased from 30.5 kg to 35 kg, the body mass index increased from 13.6 kg/m^2 to 15.6 kg/m^2, and the body mass index Z-score value for age increased from -3.04 to -1.75. The family expressed satisfaction with the counseling process. Patients diagnosed with spinal muscular atrophy require medical nutritional therapy counseling. Telenutrition counseling is considered beneficial in reaching patients and ensuring the treatment process.
Keywords: Rare diseases; spinal muscular atrophy; nutrition; telenutrition.

ÖZ

Bu olgu sunumunda, sipinal muskuler atrofi tip 2 tanısı olan 13 yaşında erkek hastanın 6 ay süren telenütrisyon danışmanlık süreci tartışılmıştır. Tekerlekli sandalye kullanan ve ağızdan beslenen hastada ağır malnütrisyon görülmüştür. Tüm bulguların incelenmesi sonucunda günlük 50 kkal/kg enerji ve 1.85 g/kg protein içeren diyet ile 6 aylık telenütrisyon danışmanlık süreci başlatılmıştır. Süreç sonunda hastanın vücut ağırlığı 30,5 kg'den 35 kg'ye, beden kütle indeksi 13,6 kg/m^2'den 15,6 kg/m^2'ye ve yaşa göre beden kütle indeksi Z-skoru değeri ise -3,04'den -1,75'e yükselmiştir. Aile danışmanlık sürecinden memnun kaldığını belirtmiştir. Sipinal muskuler atrofi tanılı hastalar tıbbi beslenme tedavisi danışmanlığına ihtiyaç duymaktadır. Hastalara ulaşmak ve tedavi sürecini sağlamak için telenütrisyon danışmanlığının faydalı olabileceği düşünülmektedir.
Anahtar kelimeler: Nadir hastalıklar; spinal muskuler atrofi; beslenme; telenütrisyon.

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INTRODUCTION

Spinal muscular atrophy (SMA) is a rare autosomal recessive neuromuscular disease characterized by progressive muscle atrophy, weakness, paralysis, and degeneration of the alpha motor neurons in the spinal cord. Among its various types, nerve, skeletal, and muscular system problems are prevalent, particularly in patients with type 1 and type 2 SMA (1). Most patients experience respiratory and feeding difficulties due to muscle atrophy, which affects chewing and swallowing functions. Consequently, expert dietitian counseling is crucial during the medical nutrition therapy process for patients with SMA (2). Telenutrition leverages electronic information and telecommunication technologies provided by licensed dietitians from

authorized institutions to manage all stages of medical nutrition therapy. This includes anamnesis, patient nutritional status determination, evaluation of anthropometric, laboratory, and physical findings, and the planning and monitoring of medical nutrition therapy (3,4). This case report aimed to elucidate the medical nutrition therapy process facilitated through a telenutrition system in a patient diagnosed with SMA type 2 in Türkiye.

CASE REPORT

A thirteen-year-old male patient sought telenutrition support via the website smailebeslen.com due to concerns about weight loss. The patient was diagnosed with SMA type 2 at the age of 11 months, has three copies of the survival motor neuron-2 gene, and regularly received Nusinersen treatment. A history of scoliosis surgery at the age of nine was noted, after which weight gain ceased. Currently, the patient uses a wheelchair and engages in physical therapy exercises twice a week. The patient, who has regular measurements taken at the family health center, was reported to have a body weight of 30.5 kg and a height of 150 cm. According to the World Health Organization (WHO) criteria, his body mass index (BMI) for age stands at 13.6 kg/m^2, with a BMI Z-score for age of -3.04. In terms of the biochemical parameters, his plasma creatinine value was noted as 0.09 mg/dL. The vitamin, mineral, and enzyme levels within his nutritional profile were within normal ranges.

The patient's family had previously implemented the Gut and Psychology Syndrome (GAPS) diet. The GAPS diet is an elimination diet based on the exclusion of all grains, pasteurized dairy products, starchy vegetables, and refined carbohydrates from the diet for an extended period. In its place, a dietary plan is followed, which includes homemade bone broths, vegetable juices, animal products, and fats. The family discontinued the implementation of the GAPS diet due to its adverse effects on the patient. At the first interview, it was reported that the patient ate three meals a day and had no chewing or swallowing problems. In general, it has been reported that there is no food selectivity when consuming all meals cooked at home. The patient experienced occasional constipation. When the three-day food record was examined, it was observed that he received an average of 1100 kcal (36 kcal/kg) of daily energy from nutrition. It has been calculated that 52.5% of the energy intake comes from carbohydrates, 14.5% from proteins, and 33% from lipids. It was determined that the vegetable intake is low and the individual consumes 8 grams of fiber daily. An initial diet containing 50 kcal/kg of energy and 1.85 g/kg of protein was planned to ensure weight gain. In the prepared 1500-kcal diet, 50% of the energy comes from carbohydrates, 15% from proteins, and 35% from lipids. The fibre content of the diet was 21 g. Daily fluid consumption is recommended to be at least 1500 ml. The patient was weighed before each interview.

The telenutrition counseling process consisted of monthly follow-ups conducted via the Zoom Pro (https://zoom.us/) program. The initial interview lasted for 60 minutes, during which a detailed anamnesis was conducted. Based on the patient's evaluation, a weight increase of 15% (5 kg) was targeted over a six-month period with diet monitoring. The patient and family were introduced to the 'Food Exchange List', explaining how their daily diet could be modified using this list. Written materials were provided to the patient, concerning the management of the diet process, daily meal plans, menu samples, a diet exchange list, and recommendations. Subsequent interviews were conducted for 30 minutes. Adherence to the diet during this process was assessed based on the information provided by the patient and the family. Recommendations were given to address difficulties encountered in adhering to the diet program. The patient's dietary follow-up process is summarized in Table 1.

The high educational level and motivation of the family had a significant impact on the adherence process to the diet. After six months of telenutrition counseling, the patient's severe malnutrition improved to a moderate level. He reported enhanced arm mobility during physical activities. The patient also expressed good compliance with the dietary regimen and expressed satisfaction with the monitoring process. The family requested continuation of dietary monitoring through telenutrition counseling.

Table 1. Telenutrition consultancy process

Interviews (Every month)	Body Mass (kg)	BMI (kg/m^2)	BMI for age Z-score	Interview details
1. Interview	30.5	13.6	-3.04	Detailed anamnesis was performed, and the anthropometric and biochemical parameters were evaluated. A targeted diet was developed, and the corresponding goals were established. Subsequently, nutritional education was provided to the patient.
2. Interview	31.5	14.0	-2.71	A question-and-answer session was conducted, and no problems were identified.
3. Interview	32.1	14.3	-2.54	A question-and-answer session was conducted, and constipation was observed.
4. Interview	32.3	14.4	-2.50	A question-and-answer session was conducted. An increase in body temperature was noted for several days following the administration of drugs for scoliosis treatment. This side effect disrupted the patient's adherence to the dietary regimen.
5. Interview	33.0	14.7	-2.30	A question-and-answer session was conducted, and no problems were identified.
6. Interview	33.9	15.1	-2.06	A question-and-answer session was conducted, and no problems were identified.
7. Interview	35.0	15.6	-1.75	A question-and-answer session was conducted, and no problems were identified. It was also noted that the Nusinersen treatment would be administered later in the month.

BMI: Body mass index

DISCUSSION

Nutrition is a very important issue for patients with SMA, and no dietary model has been developed based on scientific evidence. The medical nutrition process is managed by expert dietitians who prepare a personalized plan according to the patient's symptoms. Throughout this process, detailed anamnesis is taken from the patient, anthropometric measurements are conducted, biochemical and physiological parameters are assessed, and nutrition programs tailored to socio-cultural values are prepared (2). To plan a diet, it is necessary to first determine the daily energy required by the patient. Patients with SMA have a lower basal metabolic rate than their healthy peers because of muscle wasting or weakness. Simultaneously, movement restrictions reduce physical activity. Therefore, the daily energy requirements of children with SMA are lower than those of their healthy peers (5,6). Some clinics recommend 9-11 kcal/cm of energy intake for patients, according to their height (2). Energy calculation for height was not performed in this patient because of bone problems. When the patient's three-day food record was examined, he received an average of 37 kcal/kg of energy. International organizations recommend an average energy intake of 60 kcal/kg per day for healthy 13-year-old boys (7). To increase the weight of the patient, a diet program containing an average of 50 kcal/day (1500 kcal) of energy was determined in the first stage, taking into account the daily food consumption amount. Evidence of the percentage of energy from macronutrients in the diet of patients with SMA is limited. In general, the macronutrient distributions in SMA Type 2 patients were similar to those of their healthy peers. In addition, considering muscle loss, it is important to provide 1-2 g/kg of protein intake (2).

While planning the nutrition program for patients, the number and content of meals should be planned according to the patient's living conditions. In general, the energy requirement per body weight of children is higher than that of adults. In addition, their stomach capacity is lower than that of the adults. For this reason, the number of meals is increased to meet the high daily energy requirement. They are fed little and often. At the same time, it is necessary to provide a variety of food groups such as meat, milk, vegetables, fruits, and cereals at every meal, especially to avoid micronutrient deficiencies (8). The number of meals for the patient was planned according to living standards and was determined to be three main meals and two snacks, with high nutritional diversity provided at each meal. In addition, fibre and fluid consumption are increased to prevent constipation. In general, the recommendation for 14 g of dietary fibre per 1000 kcal has been adhered to (8).

Factors such as weight, height, BMI, blood parameters, and body tissue distribution are used in the follow-up of patients with severe malnutrition. Follow-up was performed according to the WHO growth curves. In these curves, it is recommended to follow up BMI values according to age in children over 10 years of age. (9). Height measurements and BMI values can be misinterpreted because of wheelchair use and scoliosis problems. Therefore, blood parameters and weight follow-up are prioritized during patient follow-up. The fact that telenutrition counseling is carried out remotely does not allow for physical examinations and measurements. The patient's anthropometric measurement data were regularly provided by the family in the same place and with the same measurement tool.

Telenutrition offers great opportunities for healthcare professionals, especially for patients with limited mobility (3). Although our patient lived in a different city, he regularly participated in his appointments. The appointment times were flexible and could be arranged. The patient had participated in their appointments using only the Internet and computers without experiencing transportation problems, so he saved time and money. It survived the stress factors of the hospital environment and transportation. During the COVID-19 process, similar positive results were seen in a study examining the medical nutrition treatment of phenylketonuria patients in Türkiye (10). Similar benefits have been reported in patients with obesity, cancer, spinal cord injury, kidney disease, and surgery (3).

The educational level of the family holds significant importance in the adherence process to medical nutritional therapy. This process is also valid for telenutrition counseling. It is necessary for the family to understand their education, actively utilize telecommunication systems, and possess the required educational level to maintain adherence to the diet (11). The high level of education of both parents in this case has increased the effectiveness of telenutrition counseling, facilitating adherence to the diet.

For patients who have difficulty reaching experts, such as those with rare diseases, telenutrition systems should be developed and dietitians should be able to actively use these systems (12,13). In addition, telenutrition systems have legal problems such as patient rights, ethical issues, health payments, or taxation. International organizations have supported the development of telenutrition systems by publishing various guidelines (3,14). Developed in Türkiye, smailebeslen.com is the first and only free telenutrition system and serves SMA patients.

Studies related to the nutrition of patients with spinal muscular atrophy have predominantly focused on tube feeding or identifying the nutrition-related challenges faced by these individuals. There is a lack of extensive research involving the detailed calculations required for planning the medical nutrition process for patients and monitoring adherence to the diet. Particularly, there is a scarcity of detailed studies concerning oral nutrition and the management of the nutritional process (15,16). It is believed that this case study, providing detailed insights into the management of oral nutrition in a spinal muscular atrophy patient, would contribute significantly to the literature in this area.

CONCLUSION

Nutritional problems are common in patients with SMA. It can be difficult for patients to access medical nutrition therapy services due to their mobility limitations. Telenutrition support has positive effects on patients' nutrition. Hence, the development of customized telenutrition systems for SMA patients is recommended. Training of dietitians to work within these systems and the involvement of expert dietitians in the counseling process are necessary.

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