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CASE REPORT

THE EFFECTS OF INDIVIDUAL EXERCISE PROGRAM ON SIBLINGS WITH SPINAL MUSCULAR ATROPHY TYPE 3: A CASE REPORT

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ABSTRACT

Spinal Muscular Atrophy (SMA) type 3 is an autosomal recessive disease that occurs as a result of the degeneration of the motor neurons of the spinal cord and causes severe functional impairment in patients. This case report aims to examine the effects of individual exercise programs on functional performance applied to two siblings with SMA type 3. Siblings were included in an individually planned exercise program for 60 minutes, 2 days a week for 8 weeks. Before and after the exercise program, the motor performance, trunk impairment, gait, balance and activities of daily living were evaluated with Motor Functional Measurement, Trunk Impairment Scale, GAITRite electronic walkway, Bertec Balance Check Screener^{TM} force platform system and Functional Impairment Measurement, respectively. The individually planned exercise program had positive effects on motor performance, trunk impairment, gait, balance and activities of daily living in siblings with SMA type 3. An individual exercise program that is planned by the functional status of the patient and includes different exercise types is extremely important to increase the functional independence of SMA type 3 patients during the rehabilitation process.

Keywords: SMA Type 3, siblings, individual, exercise.

INTRODUCTION

Spinal Muscular Atrophy (SMA) is an autosomal recessive disease that results from degeneration of motor neurons of the spinal cord (1). Progressive proximal muscle weakness, fatigue, scoliosis, respiratory complications, nutritional problems, and severe functional limitations as a result of degeneration of motor neurons in the spinal cord are the most common clinical findings in patients (2). Although SMA has a wide clinical variety, it is conventionally classified into four clinical types considering the age of onset and maximum motor function. An additional phenotype (type 0) is also included in the classification to describe severe forms of antenatal-onset SMA (3,4).

SMA type 3 (Kugelberg-Welander disease) is a comparatively mild subtype but shows wide clinical heterogeneity (5). When the literature is examined, it is seen that strengthening exercises and aerobic exercises are studied in patients with SMA type 3 (2, 5). Although there are studies in the literature showing the effects of exercise training on SMA type 3 patients, there are not many studies that measure the effects of an individually planned exercise program including functional exercises with objective methods in SMA type 3 patients. This case report

aims to examine the effects of individual exercise programs on functional performance applied to two siblings with SMA type 3.

CASE REPORT

Subjective history

The patients are 25-year-old female (Case-1) and 27-year-old male (Case-2) siblings with genetically confirmed SMA type 3. Siblings stated that their complaints about sit-to-stand, climbing up and down stairs started when they were about 18 years old. The siblings also stated that they had not received physical therapy and rehabilitation before. The siblings who preserved their walking functions applied to our clinic for an exercise program.

Verbal and written information was given to siblings and their written consents were obtained.

Table 1. Descriptives of patients

	Case-1	Case-2
Age (years)	25	27
Gender (F/M)	F	M
Weight (kg)	72	96
Height (cm)	160	177
BMI (kg/cm²)	28.12	30.8
Hammersmith Functional Motor Scale-Expanded Score (0-66)	62	43

Assessment of Motor Performance

The motor performances of the patients were evaluated with the Motor Function Measure (MFM) Scale and Hammersmith Functional Motor Scale-Expanded (EHFMS). The total score ranges from 0-96 in MFM and 0-66 in EHFMS. Higher scores indicate better motor performance (6, 7).

Assessment of Trunk Impairment

Trunk functions were evaluated with the Trunk Impairment Scale (TIS). TIS consists of static sitting balance, dynamic sitting balance and coordination subtitles. The total score ranges from 0 to 23. Higher scores indicate a better functional state of the trunk (8).

Gait Analysis

Gait parameters, consisting of step length, stride length, base of support, velocity, cadence and functional ambulation profile (FAP) were assessed using the GAITRite electronic walkway (CIR System INC. Clifton, NJ 07,012). The data were acquired with pressure active sensors from the system with 18,432 sensors, at 60–120 Hz (9).

Table 2. Clinical measurements

	Case-1		Case-2	
	Before	**After**	**Before**	**After**
Motor Function Measurement Scale (0-96)	93	94	65	72
Trunk Impairment Scale (0-23)	16	19	14	16
Functional Independence Measurement (0-126)	120	126	111	115

Assessment of Balance

Balance assessment was performed using the Bertec Balance Check Screener^{TM} force platform system (Bertec Co., Columbus, OH, USA) which measures three postural tests. These tests consist of normal stability eyes opened (NSEO), normal stability eyes closed (NSEC) and Limits of Stability (LoS) (10).

Assessment of Activities of Daily Living

Activities of daily living were evaluated with the Functional Independence Measurement. The scale assesses self-care, mobility, locomotion, communication, and social communication. The total score ranges from 0 to 126. High scores indicate increased independence in activities of daily living (11).

Intervention

The patients were included in an individual exercise program in 60-minute sessions, 2 days a week for 8 weeks. The exercise program consisted of functional strengthening of the trunk, back and extremity muscles by using bodyweight, trunk stabilization, bridge exercises with varying difficulty, rotations, functional reach of shoulder from different sides, balance and walking exercises and functional exercises. Balance and forward-sideways-backward walking exercises were applied in different soft ground properties. Functional exercises suitable for daily life activities such as squat, sit-to-stand, climbing and descending stairs were actively applied. The difficulties of the exercises were individually arranged by the functional capacities of the siblings.

Case-1

Case-1 had a better functional level than her sibling in terms of functional motor performance. As a result of the pre-treatment evaluations of case-1, trunk disorder and weakness of extremity muscles were detected. For this reason, in the rehabilitation goals of case-1, it was aimed primarily to preserve the current functional level and to improve it.

Case-2

Case-2 was more functionally affected than his sibling in terms of activities of daily living. He also complained of frequent falls. While planning the rehabilitation program of case-2, reduction of falls, preservation and improvement of functional capacity were taken into consideration. The demographic information and Hammersmith scores of siblings are shown in Table 1. The clinical measurements, gait and balance parameters are shown in Table 2, Table 3 and Table 4, respectively.

Table 3. Gait parameters

	Case-1		Case-2	
Gait Parameters	**Before**	**After**	**Before**	**After**
Step length (cm)	53.07	58.50	62.92	65.35
Stride length (cm)	106.22	117.33	126.02	130.08
Base of support (cm)	13.80	13.28	17.19	17.19
Velocity (cm/s)	83.96	101.06	96.8	105.8
Cadence (step/min)	95	104.06	92.3	97.2
FAP	93.3	97.3	87.3	96.5

FAP: Functional Ambulation Profile

Table 4. Balance parameters

		Case-1		Case-2	
Balance Parameters (cm)		**Before**	**After**	**Before**	**After**
NSEO	Anteroposterior	0.479	0.434	0.575	0.53
	Mediolateral	0.309	0.305	0.312	0.241
NSEC	Anteroposterior	0.596	0.561	0.584	0.524
	Mediolateral	0.209	0.362	0.38	0.347
LoS	Forward-Backward	13.541	15.79	15.928	16.41
	Left-Right	16.64	17.861	20.841	21.02

NSEO: Normal stability eyes opened, **NSEC:** Normal stability eyes

DISCUSSION

This case report examines the effects of individual exercises applied to siblings with SMA type 3 on functional performance, gait, balance and activities of daily living. In SMA type 3 patients, loss of strength can increase over the years, and patients may lose ambulation. Therefore, SMA patients need to make rehabilitation a part of their lives to slow down the loss of muscle strength as much as possible and to maintain ambulation.

Bartel et al. stated in their review study that the effects of combined strength and aerobic exercise training on SMA type 3 patients are uncertain due to the very low quality of evidence (5). This conclusion leads to think there are not many studies examining the effects of an individual exercise program designed according to the functional capacity of the patients on SMA type 3 in detail with objective methods.

327 et al. stated that 12-week training with the rgometer increases oxidative capacity in SMA type 3 patients, but causes fatigue (12). In our case report, the cases were included in a training program consisting of strengthening and functional exercises appropriate to their functional capacities. As a result of our case report, it is seen that the functional ability such as motor performance, trunk function, independence of activities of daily living, balance and walking performance of the patients improved. The positive results of our case report are thought to be due to exercises appropriate to the functional capacity of the patients. It is thought that uniform exercise will not be suitable for all SMA type 3 patients and may cause fatigue in patients. Exercise training, which combines different exercise methods suitable for the functional capacity and physical needs of the patients, is thought to have positive results in patients. On the other hand, Montes et al. examined the effects of aerobic and strengthening exercises on a group of children and adult SMA type 3 patients and presented the improvements in their patients' performance in their studies (13). As a result of this case report, it is thought that the improvement seen in patients may be due to the combination of different types of exercises consisting of aerobic and strengthening exercises.

CONCLUSION

Lifelong exercise training is extremely important in SMA type-3 patients to slow down the loss of muscle strength over the years and to maintain the independence of patients in their daily lives. Planning

these exercises according to the functional performance of the individual and including resting breaks without causing fatigue in the patients will increase the benefit from rehabilitation. In the future, it is thought that individual exercise training studies to be conducted with more SMA type 3 patients will reveal the effects of exercise training on the functional status of the patients more clearly.

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