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CASE REPORT

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Effects of Telerehabilitation in Niemann Pick Disease Type B: A Case Report

Aslı BAYRAM[1*], Hacer Sultan ÖZTÜRK [1], Ali Naim CEREN[1], İpek ALEMDAROĞLU-GÜRBÜZ[1]

Öznur YILMAZ[1], Numan BULUT[1]

[1] Hacettepe University, Faculty of Physical Therapy and Rehabilitation, Ankara, Türkiye

*Corresponding author e-mail: bayramasli.02@gmail.com

ABSTRACT

Purpose: The aim of this study was to investigate the effects of a 12-week telerehabilitation on muscle strength, range of motion, motor performance and pulmonary function in a patient with Niemann Pick Disease Type B (NPD-B).

Method: A 13-year-old boy diagnosed with NPD-B participated in the study and underwent a telerehabilitation program for 12 weeks, three sessions per week, each session lasting 60 minutes. The 6-minute walk test (6-MWT) and timed performance tests were used to determine motor function. The patient's range of motion (ROM) was measured using a goniometer. Muscle shortness was evaluated using a tape measure or a goniometer. Muscle strength was assessed using myometric measurement. Spirometry test was used to measure pulmonary function. Assessments were applied before and after the telerehabilitation program.

Results: A 61-meter increase was observed in the 6-MWT. The patient's right and left hip flexor muscle shortness improved by 7.0 and 9.0 degrees, respectively. Improvements were also observed in gastrocnemius muscle shortness (right 12, left 7 degrees). Knee extension and left ankle dorsiflexion reached normal ROM. The right shoulder, left shoulder, right hip, and left hip flexion muscle strength of the patient increased by 6.0, 6.0, 8.4, and 11.6 pounds, respectively. The patient's forced expiratory volume in one second/forced vital capacity ratio (FEV1/FVC) value was 87% before the telerehabilitation program and 89% after the telerehabilitation program. No side effects were reported during the telerehabilitation program.

Discussion: These findings suggest that 12-week telerehabilitation program can be used as a treatment method that contributes to the management of Niemann-Pick Disease Type B (NPD-B) in addition to medical treatments. However, controlled studies with a larger number of participants in this disease are needed.

Key Words: Telerehabilitation, Physiotherapy, Niemann Pick Disease Type B, Outcome Measurement.

INTRODUCTION

Niemann-Pick disease (NPD) is an autosomal recessive lysosomal lipid storage disorder caused by acid sphingomyelinase deficiency (ASMD) resulting from mutations in the SPMD1 gene (1). Sphingomyelin, a component of cell membranes and myelin sheaths, cannot be broken down as needed due to ASMD, resulting in lipid storage in various tissues. The primary organ systems affected by this enzyme deficiency are the spleen, liver, and lungs (1). While there is limited information available on the prevalence of NPD, which has three subtypes (A, B, and C), the birth rate is estimated to be 0.5 to 1 per 100,000 (2, 3). While types A and B are caused by ASMD, type C, despite sharing the same disease name, is a distinct genetic disorder characterized by central nervous system involvement due to impaired intracellular transport of cholesterol, a lipid molecule. Type A disease is characterized by progressive psychomotor and developmental delay, often leading to death by the age of 3-4 years (3). In contrast, type B is rarely associated with neurological findings but may present with hepatosplenomegaly, thrombocytopenia, dyslipidemia, and interstitial lung disease.

Skeletal involvement is common in patients with Niemann Pick Disease Type B (NPD-B). NPD-B can cause growth retardation, osteopenia, or osteoporosis in adolescent patients (4). Although lung involvement is observed in all three types of Niemann-Pick disease, it is most prevalent in type B (5). Respiratory symptoms are generally mild, presenting as recurrent coughing and moderate dyspnea with exertion (6). This condition may lead to reduced exercise tolerance in some patients (7). Secondary restrictive changes may occur in lung function due to significant hepatosplenomegaly (4).

There is no specific treatment for this disease. However, symptomatic treatments such as enzyme replacement therapy, substrate reduction therapy, gene therapy, bone marrow or stem cell transplantation are often preferred in the management of the disease. Treatment strategies for NPD focus on preventing symptoms and disease progression (8, 9). A case report of a female patient diagnosed with NPD-C, who was treated with Migalastat and applied an 8-week rehabilitation program, demonstrated an improvement in muscle strength and body balance, as well as a reduction in ankle joint limitation (10). Although the effectiveness of physiotherapy on osteoporosis and dyspnea is known (11, 12), there are no studies examining its effects on NPD-B. In this context, telerehabilitation, a current treatment approach, was selected to maintain a high rate of patient participation in treatment and minimize adverse situations that may arise in a clinical setting.

Therefore, we aimed to present findings on the effectiveness of the telerehabilitation program on various parameters in the patient with NPD-B, who was treated with a multidisciplinary approach.

METHODS

Case Presentation

A 13-year-old boy with a height of 1.44 m and a weight of 39 kg was included in this study. He was born with a normal weight and had a typical development. At the age of 7 months, he was taken to the hospital because of vomiting and sudden weight gain, and the disease was diagnosed after blood and genetic tests (SMPD1 gene c.416T>C homozygous mutation). There are individuals on his father's side with the same disease. He takes Enapril which is a blood pressure medication. No obvious problems were observed in the patient before receiving physical therapy at an external center. However, the patient was referred to physical therapy by the Department of Metabolism and Nutrition Diseases because of the observation of obvious contracture in the upper extremity. He has been undergoing physiotherapy at an external center for the past five months.

Assessments

Parents and children were informed about the content and duration of the study and consent was obtained. The demographic characteristics were recorded, and the following assessments were performed sequentially. The Modified Borg Scale was used to assess fatigue and adjust the duration, repetition, and resistance of the exercise. Breaks were taken during assessments to prevent fatigue. Assessments were conducted before and after telerehabilitation.

6-minute walk test (6-MWT) and timed performance tests were used to assess the motor function. The 6- MWT is a submaximal functional walking test that measures the maximum distance that individuals can walk in 6 minutes and reflects physical capacity (13). According to the application procedure, the patient was asked to walk as fast as possible in a 25-meter corridor for 6 minutes (14). Timed performance tests included the stand up from supine position, climb/descend four steps of stairs, and the ten-meter walk/run test. For the tests, the task was performed once after being explained, and the results were recorded in seconds.

Patient's range of motion (ROM) was measured using a goniometer (15). The results were recorded in degrees. The patient's muscle shortening in the trunk, lower, and upper extremities was measured using a tape measure or goniometer, and the results were recorded in degrees or centimeters. The measurement of muscle shortness was in accordance with the protocol of Otman et al (16).

Shoulder flexion and abduction, hip flexion, and knee extension muscle strength were measured using myometric measurement (Jtech Commander Muscle Dynamometer, USA). The muscle strength test was performed three times, and the highest score was recorded. The results were recorded in Newtons. Muscle strength was measured according to Sloan et al (17).

Pulmonary function tests were performed using a Pony FX spirometer (Cosmed, Rome, Italy). The tests were conducted in accordance with the guidelines of the American Thoracic Society (18). The test was performed in a comfortable sitting position. The mouthpiece and nose clip were worn to prevent leakage. He was first asked to breathe normally and to exhale maximally. When he reached full exhalation, he was told to breathe maximally and to blow quickly and hard (19). Repeated three times, and one minute rest was given between repetitions to prevent muscle fatigue. Best forced vital capacity (FVC), forced expiration volume in one second (FEV1), and forced expiratory volume in one second/forced vital capacity ratio (FEV1/FVC) values were recorded.

Table 1. The telerehabilitation program exercise

Modality	Exercise
Breathing Exercises	Active cycle of breathing techniques and diaphragmatic breathing.
Stretching	Straight leg raises for hamstrings and gastrocnemius, pulling knees to abdomen in supine position for lumbar extensors, stretching along the wall for pectoral muscles.
Posture and core exercise: Core stability refers to the ability of core musculature to stabilize the spine. It is required to increase stiffness of the trunk and hip in preparation for, and in response to, spinal loading, to prevent instability of the vertebral column and to facilitate return to equilibrium following perturbation (25).	Chin tuck, posterior pelvic tilt, bridge and clam exercise
Strengthening	Shoulder flexion-abduction-external rotation PNF pattern with TheraBand for strengthening upper extremity, performing the movement against the TheraBand for knee extension and hip flexion.
Functional exercise and cool down	Rhythmic hip abduction, arm crossing, walking on toe and heel, walking in place, side walking, jumping up and down. Cool down was done by stretching or breathing exercises

Telerehabilitation Program

The telerehabilitation program was conducted via online video calls using the WhatsApp application, as the patient was in a different city. For the patient to learn the exercises, the physiotherapist first demonstrated the exercise herself and explained the correct form. Any mistakes observed during the exercise were corrected as much as possible. Each session was conducted with the physiotherapist. The patient's environment was adapted to ensure that the program was performed in the most suitable conditions.

The patient underwent a 12-week telerehabilitation, three days a week, each session lasting 60 minutes. Each strengthening and respiratory exercise consisted of 8 repetitions in 3 sets, and each stretching exercise consisted of 10 repetitions lasting 30 seconds. Functional exercises started at 30 seconds in the first week. The duration was increased according to the patient's fatigue. When the patient's fatigue level reached 3-4 on the Modified Borg scale, which is rated from 0 to 10, the number of repetitions was increased. Strengthening exercises using a resistance band with a difficulty level of 4-6 on the Borg scale were added. There was a 3–5-minute rest period between exercises. Exercises were adjusted and progressed according to the patient's condition in terms of duration, number of repetitions, and resistance, ranging from 4 to 6 on the Modified Borg scale (20).

The telerehabilitation program is described in Table 1. Treatment effectiveness and side effects such as pain and fatigue were assessed from the parent and child after each session and at the end of telerehabilitation. The correlation between continuous variables was examined using Spearman's correlation coefficient test since the normality assumption did not seem satisfied. The correlation coefficient between 0–0.10, 0.10–0.39, 0.40–0.69, 0.70–0.89, and 0.90–1 was accepted as negligible, weak, moderate, strong, and very strong correlations, respectively (34). A p-value of <0.05 was statistically significant.

RESULTS

The patient completed the 12-week telerehabilitation program. An improvement of approximately 61 meters was observed in the 6-MWT. The results of the 6-MWT, timed performance tests (standing up from supine position, stair climbing and descending, 10-meter walk test, and Timed Up and Go), as well as upper and lower extremity muscle strength measurements, are presented in Table 2.

Following the intervention, knee extension and ankle dorsiflexion ranges reached normal limits. Overall improvements or stabilization were observed in joint mobility and muscle shortness across the upper and lower extremities. The detailed ROM and muscle shortness measurements are provided in Appendix 1 (Table 3).

Considering the results of the spirometry test, the FVC, FEV1, and FEV1/FVC values before rehabilitation were 72%, 66% and 87%, respectively. After rehabilitation, these values were 68%, 64% and 89% respectively.

Both the child and parent stated that there were no side effects from the treatment, that they wanted to continue the treatment, and that it was more effective than the treatment applied at the external center.

Table 2. The results related to 6-MWT, timed performance tests and muscle strength of a patient with Niemann Pick Disease Type B

	Pre-treatment		Post-treatment	
6-MWT (m)	460		521	
Standing up from supine position (s)	4.00		3.21	
Climbing 4 flights of stairs (s)	1.30		1.51	
4 steps down the stairs (s)	2.00		1.81	
10 meters walk (s)	8.81		8.77	
Time up and go (s)	7.49		5.29	
	Right	**Left**	**Right**	**Left**
Shoulder flexion (pound)	18.60	16.40	24.60	22.40
Shoulder abduction (pound)	18.40	14.80	17.10	15.90
Hip flexion (pound)	16.40	15.30	24.80	26.90
Knee extension (pound)	46.60	42.20	40.20	34.30

DISCUSSION

This study investigated the effects of a 12-week telerehabilitation program on the motor performance and pulmonary function of a patient with NPD-B. It was concluded that the telerehabilitation program may improve motor performance and stabilize muscle strength, range of motion and pulmonary function. The potential effects of telerehabilitation are supported by feedback from the child and their family.

It was shown that combined aerobic and resistance exercise is the most appropriate method for lipid management in a meta-analysis (21). In line with the literature, our combined telerehabilitation program achieved significant improvement. Considering that the minimal clinical important difference for the 6-MWT in neuromuscular diseases is 28.5 meters (13), an increase of approximately twice this value after 12 weeks of training shows the effectiveness of the telerehabilitation.

In a telerehabilitation study conducted on patients with hereditary neuromuscular disorders, improvement was observed in maximum inspiratory pressure values. However, other respiratory parameters (maximum expiratory pressure, FVC, peak cough flow), hand/pinch strength, and walking distance values remained stable (22). Contrary to the results of the study, there was an increase in walking distance and muscle strength in other muscle groups except for knee extension. The decrease in knee extension strength may be due to insufficient loading during sessions (23), and therefore no increase in strength was observed. Overall, our study demonstrated that the telerehabilitation program can effectively improve muscle strength and functional capacity.

A recent review emphasized the positive effects of ROM on mobility and the importance of preserving it (24). Considering the results of range of motion, stretching exercises have been shown to be effective in preventing the deterioration of ROM limitations or eliminating limitations even when applied through telerehabilitation.

In lung involvement problems most seen in type B, stem cell transplantation is considered (5). Active breathing techniques and diaphragmatic breathing exercises, which are part of telerehabilitation, stabilized the patient's lung function. The reason for not seeing improvement in pulmonary function is that the content of the telerehabilitation program is in the form of breathing exercises rather than breathing training. However, the maintenance of these parameters over 12 weeks has shown us that pulmonary physiotherapy is a treatment method that should be considered in the treatment of the disease, in addition to known treatment methods.

Limitations

The fact that our study is not a longitudinal follow-up study is a limitation, and being a single-center study is also a limitation. There is a need to examine the results of longitudinal studies in which the same patients are followed up in the future.

CONCLUSION

In conclusion, 12 weeks of telerehabilitation-based physical therapy can be effective treatment to improve the motor function and maintain the muscle strength, range of motion and pulmonary outcomes in NPD-B when proper application and optimal conditions are provided. This is supported by feedback from parents and children. This study suggests that a telerehabilitation program could be used alongside medical treatment methods to help manage the disease.

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Appendix

Table 3. The results related to ROM limitation degree and muscle shortness of the patient

	Pre-treatment		Post-treatment	
	Right	Left	Right	Left
Ankle dorsiflexion limitation (degree)	18	7	6	-
Knee extension limitation (degree)	5	4	-	-
Wrist extension limitation (degree)	55	22	14	18
Elbow extension limitation (degree)	7	3	12	7
Shoulder flexion limitation (degree)	21	20	20	23
Shoulder abduction limitation (degree)	32	32	28	38
Hip flexors (degree)	32	32	23	25
Hamstring muscles (degree)	42	35	39	38
Quadriceps (cm)	33	30	32	28
Gastrocnemius (degree)	18	7	6	-
Pectoral muscles (cm)	8	12	7.50	7
Lumbar extensor muscles (cm)	42		42	