

The Effect of Telerehabilitation Supported Occupational Therapy Intervention on Participation and Occupational Performance in Rare Diseases

Nadir Görülen Hastalıklarda Telerehabilitasyon Destekli Ergoterapi Müdahalesinin Katılım ve Okupasyonel Performans Üzerine Etkisi

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Abstract

The aim of this case study was to investigate the effects of occupational therapy and telerehabilitation supported occupational therapy intervention on participation and occupational performance in individuals with rare diseases. The study included 6 cases aged between 8-11 years with Cri du Chat syndrome, Rett syndrome and Tuberous Sclerosis. The cases were divided into two groups as occupational therapy intervention group (n=3) and telerehabilitation supported occupational therapy intervention group (n=3). Occupational therapy intervention was applied to the control group, and occupational therapy intervention with 40-minute telerehabilitation sessions was applied to the telerehabilitation group for 3 months. Before and after the intervention, occupational performances of the cases were measured with the Canadian Occupational Performance Measure (COPM), functional independence was measured with the Children's Functional Independence Measure (WeeFIM), and functional abilities and performances were measured with the Pediatric Evaluation of Disability Inventory (PEDI) and were applied to their caregivers. The study found that occupational therapy intervention supported by telerehabilitation increased the participation and occupational performance of individuals in the areas of self-care and communication. In the study, in the telerehabilitation group, supporting occupational therapy intervention with telerehabilitation made the intervention more effective and efficient compared to the control group.

Keywords: Occupational Performance, Occupational Therapy, Participation, Rare Diseases, Telerehabilitation.

Özet

Bu olgu çalışmasının amacı nadir hastalıklara sahip bireylerde ergoterapi ve telerehabilitasyon destekli ergoterapi müdahalesinin katılım ve okupasyonel performans üzerine etkisini incelemektir. Çalışmaya Cri du Chat sendromu, Rett sendromu ve Tübero-skleroz tanılı 8-11 yaş arası 6 olgu dahil edilmiştir. Olgular ergoterapi müdahale grubu (n=3) ve telerehabilitasyon destekli ergoterapi müdahale grubu (n=3) olacak şekilde iki gruba ayrılmıştır. Kontrol grubuna ergoterapi müdahalesi, telerehabilitasyon grubuna ise 3 ay boyunca 40 dakikalık telerehabilitasyon seanslarıyla ergoterapi müdahalesi uygulanmıştır. Olguların müdahale öncesi ve sonrasında aktivite performansları Kanada Aktivite Performans Ölçümü (KAPÖ) ile, fonksiyonel bağımsızlıkları Çocuklar İçin Fonksiyonel Bağımsızlık Ölçümü (WeeFIM) ile fonksiyonel yeteneklerini ve performansları ise Pediatrik Özürlülük Değerlendirme Envanteri (PÖDE) ile bakım verenlerine uygulanmıştır. Çalışmada telerehabilitasyon destekli ergoterapi müdahalesinin bireylerin öz bakım ve iletişim alanlarında katılım ve okupasyonel performanslarının artış gösterdiği bulgusuna ulaşılmıştır. Çalışmada kontrol grubuna oranla telerehabilitasyon grubunda, ergoterapi müdahalesinin telerehabilitasyonla desteklenmesi müdahalenin daha etkili ve verimli gerçekleşmesini sağlamıştır.

Anahtar Kelimeler: Ergoterapi, Katılım, Nadir Hastalıklar, Okupasyonel Performans, Telerehabilitasyon.

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1. Introduction

Rare diseases are diseases that affect a proportion of the human population. One type of rare disease may be less common in one population but more common in other populations. For this reason, definitions of rare diseases may vary according to the population in which the disease is seen (García-Pérez et al., 2021; Dawkins et al., 2018). Considering the prevalence of rare diseases, diseases that are less than 5 per 10,000 individuals are considered rare diseases (García-Pérez et al., 2021; Stolk et al., 2006). Rare diseases add 250-280 diseases each year and account for approximately 6% to 8% of the human population (Boycott et al., 2024; Melnikova, 2012). Rare diseases have mental and physical risk factors that put life at risk, and it is difficult to identify these risk factors because they are congenital or congenital (Boycott et al., 2017; Ramsey, 2011). Rare diseases usually occur in childhood and may cause individuals to end their lives early (Valdez et al., 2016; Rofail et al., 2013).

The diversity, complexity and multiplicity of rare diseases cause serious disparities in scientific knowledge, clinical expertise, availability of diagnostics and treatments, patient outcomes and quality of life (Monaco et al., 2022; Lochmüller et al., 2017). Rare diseases are generally known to affect more than one system and it is reported that 80% are genetic and 20% are caused by environmental or idiopathic causes (Köken et al., 2018). A genome-wide approach using exome or genome sequencing is used in rare diseases that cannot be diagnosed with a clear result (Stark and Scott, 2023; Marshall et al., 2020). Almost 95% of rare diseases have no FDA-approved drug treatment, putting pressure on matching patients with appropriate therapies. There is an urgent need to identify new treatment options for rare diseases (Tabor and Goldenberg, 2018; Foksinska et al., 2022). Currently, AI methods for treatment mostly belong to supervised learning, which uses labeled datasets to train algorithms that can accurately classify results (Tabor and Goldenberg, 2018; Chirmule et al., 2024)

The reason for this study is that the difficulties faced by individuals with rare diseases in the areas of participation and activity performance have not been adequately addressed and that specific intervention approaches for these individuals are limited. In this context, individuals with rare diseases such as Tuberous Sclerosis, Cri Du Chat and Rett Syndrome constituted the sample of the study and a case series was created by selecting two participants from each disease. The effects of telerehabilitation-supported occupational therapy interventions on the participation of these individuals in daily life and their activity performance were evaluated and the change processes were observed in each case. At the same time, this study aimed to raise awareness in Turkey about rare diseases.

2. Method

The study included 6 patients diagnosed with Tuberous Sclerosis, Cri Du Chat and Rett Syndrome. The subjects were divided into control and intervention groups in pairs among the three disease groups. The control group was formed with 6 people, two from each disease group. In the intervention group, one

person was selected from each disease group. The Consent Form prepared through occupational therapists was approved and signed by the participants that they would voluntarily participate in the study. At the same time, this study was supported by TUBITAK 2209-A University Students Research Projects Support Program with the number 1919B012334466. The study was conducted between January 2024 and March 2024 at Gülseren Özdemir Special Education School and Pediatric Occupational Therapy Laboratory at Istanbul Medipol University. Ethics Committee Approval was obtained from Istanbul Medipol University Non-Interventional Clinical Research Ethics Committee on 25.12.2023 with the number E-108400-772.02-7992.

Participants were evaluated before and after the intervention with scales whose validity and reliability have been proven in the literature. The Canadian Occupational Performance Measure was used to determine the occupational performance problems of the participants and to measure activity and participation limitations; the Functional Independence Measure for Children was used to assess the participants' self-care, sphincter control, movement, transfer and communication skills; and the Pediatric Evaluation of Disability Inventory was used to assess the functional abilities and performance of children with disabilities. Since the current health status of the participants included in the study was not sufficient to complete the COPM, the scale was completed through caregivers.

Following the assessments conducted with the participants, face-to-face sessions and telerehabilitation interviews were conducted. Participants in the telerehabilitation-supported occupational therapy intervention group received occupational therapy rare disease intervention twice a week, both face-to-face with 40-minute sessions (8 sessions) and via tablet for 4 weeks with 30-minute sessions (8 sessions). In our study, telerehabilitation sessions were conducted on Zoom and Microsoft Teams platforms under caregiver control. Participants in the control group received a structured occupational therapy intervention twice a week, 40-minute sessions (8 sessions) for 4 weeks. After the interviews and interventions were completed, the results were evaluated.

2.1. Case Report

The total number of subjects in the study (n=6) was equally distributed to the occupational therapy intervention group (n=3) and the telerehabilitation-supported occupational therapy intervention group (n=3). Six participants with Tuberous Sclerosis, Cri Du Chat and Rett Syndrome were included in the groups in pairs. The subjects in the intervention group were identified as Case 2, Case 4 and Case 6. The minimum age of the cases was 8 years and the maximum age was 11 years. Four of the cases were girls and two were boys. Regarding their educational status, 4 of the cases were in primary school and 2 were in secondary school. The clinical data of the participants are shown in Table 1.

Table 1. Data Table of Clinical Characteristics

Cases	Case 1	Case 2	Case 3	Case 4	Case 5	Case 6
Diagnosis	Cri Du Chat Syndrome	Cri Du Chat Syndrome	Rett Syndrome	Rett Syndrome	Tuberous sclerosis	Tuberous sclerosis
Treatment	Physical Therapy and Occupational Therapy	Speech and Language Therapy and Occupational Therapy	Physical Therapy and Occupational Therapy	Physical Therapy and Occupational Therapy	Speech and Language Therapy and Occupational Therapy	Speech and Language Therapy and Occupational Therapy
Duration of Illness	9	11	10	8	11	10
Concomitant Diseases	Speech and walking problems	Speech and walking problems	Caution Lack of	Autism	Attention and speech problems	Epilepsy

2.2. Intervention Plan

Occupational therapy-based intervention methods for rare diseases consisting of 40-minute sessions twice a week for 4 weeks were applied to all participants who met the inclusion criteria of the study. The content of this intervention included sensory integration, participation in daily life and auxiliary daily life activities, communication and interaction skills, group activities, planning and organizational skills, joint attention and coordination, balance and coordination skills, social and academic skills, fine motor, gross motor and oral motor skills, massages to ensure saliva control, family interviews, physical and environmental arrangements appropriate to the child's rare disease diagnosis. The telerehabilitation sessions of the study were customized for each rare disease group. The room where the telerehabilitation was to take place was prepared by the caregiver with the guidance of the occupational therapist. The occupational therapist made observations and evaluations via video conferencing. Then, during the sessions, caregivers participated in the session in order to provide their children with the help that the occupational therapist deemed appropriate. Care was taken to gradually reduce caregiver assistance in future sessions. The activities in the intervention plan were explained by the occupational therapist and visually supported with materials. Subsequently, under the guidance of the occupational therapist and accompanied by the caregiver, the child was encouraged to join the occupational therapist and reinforce the activity through repetitions. Throughout the session, motivational interviews, supporting social skills, strategies for participation in activities of daily living and gaining independence, activities to support motor skills, strategies for coping with problems accompanying the disease and ergonomic arrangements were included. After each session, the caregiver was informed about the session.

3. Results

As a result of the measurement of COPM before and after the intervention, an increase was observed for all cases. As a result of the measurement made after the intervention, it was found that the intervention was more effective in Case 2 diagnosed with Cri du Chat syndrome compared to Case 4 and Case 6 (Table 2).

Table 2. COPM Results of the Telerehabilitation-Supported Occupational Therapy Intervention Group

	Case 2 Before / After	Case 4 Before / After	Case 6 Before / After
COPM Importance Score	38 / 38	37 / 37	38 / 38
COPM Performance Score	17 / 32	15 / 33	17 / 27
COPM Satisfaction Score	12 / 29	10 / 23	13 / 28
COPM Measurement Result	2,4 / 5,8	2,0 / 4,6	2,6 / 5,6

COPM= Canadian Occupational Performance Measure.

Positive improvements were observed in all parameters of WeeFIM. The highest improvement was observed in parameter A) Self-Care, while the lowest improvement was observed in parameter B) Shincter Control. The intervention was found to be more effective in Case 2 with Cri du Chat syndrome compared to Case 4 and Case 6 (Table 3).

Table 3. WeeFIM Results of the Telerehabilitation-Supported Occupational Therapy Intervention Group

Parameters	Case 2 Before / After	Case 4 Before / After	Case 6 Before / After
A) Self Care	17 / 28	13 / 20	16 / 23
B) Sphincter Control	9 / 10	6 / 7	7 / 7
C) Mobility	18 / 21	9 / 12	14 / 18
D) Locomotion	11 / 13	10 / 12	10 / 13
E) Communication	6 / 10	5 / 8	7 / 13
F) Social Communication	9 / 14	8 / 13	10 / 14
Total Score	70 / 96	51 / 72	64 / 88

WeeFIM= Functional Independence Measurement for Children.

When we look at the PEDI in general, similar differences were seen in the parameters of Functional Skills Assessment and Child Caregiver Assistance Assessment, and positive developments were observed for all cases (Table 4).

Table 4. PEDI Results of the Telerehabilitation-Supported Occupational Therapy Intervention Group

Parameters	Case 2 Before / After	Case 4 Before / After	Case 6 Before / After
Functional Skill Assessment			
A) Self Care	33 / 59	24 / 38	34 / 56
B) Mobility	47 / 60	33 / 45	31 / 40
C) Social Function	42 / 62	27 / 38	43 / 57
Assessing the Assistance and Form of Assistance from Child Caregivers			
A) Self Care	15 / 31	10 / 15	13 / 22
B) Mobility	16 / 28	15 / 19	18 / 24
C) Social Function	12 / 26	8 / 15	10 / 17

PEDI= Pediatric Evaluation of Disability Inventory.

4. Discussion

The study was conducted to measure the effect of telerehabilitation-assisted occupational therapy intervention on participation and occupational performance compared to occupational therapy intervention in children diagnosed with rare diseases. It was observed that telerehabilitation-assisted occupational therapy interventions had more positive improvements on participation and occupational performance compared to structured occupational therapy interventions.

In the management of rare diseases, occupational therapists aim to improve individuals' coping skills and promote independence in daily activities (Demirkol and Abaoğlu, 2021; Kalil, 2020). Telerehabilitation is considered a promising approach to ensure continuity of therapeutic interventions and increase access to best practices, especially for populations with limited access to client services (Sarsak, 2020). In our study, telerehabilitation was implemented using appropriate digital platforms to increase the effectiveness of occupational therapy interventions in the intervention group. The occupational therapist focused on promoting independence in occupational therapy participation through remote sessions. Fabio et al. (2022) investigated the effects of telerehabilitation on motor and cognitive functions in individuals with Rett syndrome using Skype. Their findings showed significant improvements in attention and behavioral outcomes, assessed with the Vineland Adaptive Behavior Scales and Rett Rating Scales. While their study focused on motor and cognitive parameters, the present study additionally evaluated occupational performance and participation areas such as self-care and social skills, and similar positive outcomes were found (Fabio et al., 2022). In our study, in addition to the areas evaluated in this study, participation and occupational performance areas were also evaluated. Our study results found positive changes in self-care and social skills parameters. Poole et al. (2022) reported the successful use of telerehabilitation in a case study involving a 3-year-old child with Cri Du Chat syndrome, conducted via Microsoft Teams under caregiver supervision. Their study emphasized the importance of telerehabilitation in situations where home visits are restricted, such as during the COVID-19 pandemic (Poole et al., 2022). These findings support the conclusion that telerehabilitation is an effective method for delivering occupational therapy interventions in individuals with rare diseases. In our study, telerehabilitation was conducted via Zoom and Microsoft Teams by an occupational therapist and was combined with face-to-face sessions. This hybrid model contributed to increasing the effectiveness and efficiency of the intervention process by supporting continuity, flexibility, and caregiver involvement.

Lin et al. (2023) investigated the effects rehabilitation on the functional performance of 73 children with rare diseases in Eastern Taiwan. In this study, telerehabilitation was implemented for each child, and functional outcomes were evaluated using the WeeFIM-C questionnaire. The results revealed significant differences among the three study groups in terms of self-care, mobility, and cognitive functioning (Lin et al., 2023). Similarly, the our study employed the WeeFIM scale to assess functional independence in children with rare diseases. In addition to WeeFIM, the Pediatric Evaluation of Disability Inventory (PEDI) and the Canadian Occupational Performance Measure (COPM) were also utilized to provide a more comprehensive evaluation of occupational performance and participation (Lin et al., 2023). The WeeFIM scale was used to evaluate functional independence in rare disease groups in our study. In addition to

the WeeFIM scale, the Pediatric Evaluation of Disability Inventory and the Canadian Occupational Performance Measure were used. Erarslan et al. (2024) examined the effects of a structured occupational therapy program on a 9-year-old child diagnosed with Cri du Chat syndrome. Assessment tools such as the Pediatric Berg Balance Scale, WeeFIM, COPM, and PEDI were administered before and after the intervention. The findings indicated significant improvements across all measures post-intervention, suggesting an increase in the child's independence and participation in daily activities (Erarslan et al., 2024).

These studies are revealing the effectiveness of occupational therapy interventions in rare disease populations. Similar to the findings of the our study, positive outcomes have been reported in treatment programs designed and implemented by occupational therapists. In our study, an original intervention combining telerehabilitation and structured occupational therapy was applied to children with rare diseases, with the aim of further increasing the impact and effectiveness of occupational therapy. The analysis of pre- and post-intervention assessments revealed increases in all measured parameters. Notably, improvements in self-care and social skills contributed to enhanced participation and occupational performance. These findings highlight the potential benefits of integrating telerehabilitation into occupational therapy practices. Future studies are recommended to explore the targeted use of various telerehabilitation approaches and to include a broader range of rare disease groups to strengthen the evidence base and generalizability of outcomes. The limitation of our study is the restriction of sample size due to the low prevalence of rare diseases.

5. Conclusion

In our study, when the pre-intervention and post-intervention evaluation results were examined, an increase in the scores was detected in each parameter of the applied tests. It was observed that the positive change in the parameters evaluating self-care and social skills provided benefits in the participation and occupational performance areas examined in our study. In future studies, it is recommended that occupational therapists use different telerehabilitation applications in a targeted manner and include different rare disease groups in their interventions.

Authors Contributions

Topic selection: İE; Design: İE, FA, EÖ; Planning: İE, DT; Data collection and analysis: İE, FA, EÖ; Writing of the article: İE, FA, EÖ; Critical revision İE, DT.

Conflict of Interest

Our study has been approved by all authors and there is no conflict of interest with other authors, institutions or organizations.

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