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Olgu Raporu

Effects of Physiotherapy and Occupational Therapy in A Case with Winchester Syndrome

Winchester Sendromlu Bir Olguda Fizyoterapi ve Ergoterapinin Etkileri

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ABSTRACT

Purpose: The aim of this study was to investigate the effects of physiotherapy and occupational therapy in a case with Winchester syndrome. **Material and methods:** In this report, 13 years old boy with Winchester Syndrome and physical and occupational therapy approaches were presented. Kamakura's Hand Grips Evaluation Method, Canadian Occupational Performance Measure were used to assess hand ability and performance in daily living activities. Duration of writing sentences and hand functions were assessed by Jebsen Taylor Hand Function Test. Fine motor skills were assessed by using the fine motor skills' subtests of Bruininks- Oseretsky Motor Proficiency Test, activities of daily living by Functional Independence Measurement for children (Wee-FIM), and quality of life by Child Assessment of Health Questionnaire (CHAQ). The range of motions and posture analysis of the subject were also examined. Exercise and activity trainings that included physiotherapy and occupational therapy approaches were planned to increase physical functions and ability in motor skills and performed 3 days in a week during six months regularly. **Results:** The range of motions of joints, independency level in daily living activities and quality of life of the subject were improved obviously after trainings. **Conclusion:** It is concluded that physiotherapy and occupational therapy approaches may be one of the important supportive therapeutic options for the cases with Winchester Syndrome.

Keywords: Winchester syndrome; Physiotherapy; Occupational therapy; Neuromuscular diseases

ÖZET

Amaç: Çalışmanın amacı, Winchester Sendromu olan bir vakada fizyoterapi ve ergoterapinin etkilerini araştırmaktır. **Gereç ve Yöntem:** Bu raporda, Winchester Sendromu olan 13 yaşında bir erkek çocuğu ve uygulanan fizyoterapi ve ergoterapi yaklaşımları sunuldu. Kamakura's El Kavramaları Değerlendirme Yöntemi ve Kanada Aktivite Performans Ölçeği, el becerileri ve günlük yaşam aktivitelerindeki performansı değerlendirmek için kullanıldı. Cümleleri yazma süresi ve el fonksiyonları Jebsen Taylor El Fonksiyon Testi ile değerlendirildi. İnce motor beceriler Bruininks-Oseretsky Motor Beceri Testi'nin ince motor beceri alt testi ile, günlük yaşam aktiviteleri çocuklar için Fonksiyonel Bağımsızlık Ölçeği (WeeFIM) ile ve yaşam kalitesi Çocuk Sağlık Değerlendirme Anketi (CHAQ) ile değerlendirildi. Olgunun eklem hareket açıklığı ve postür analizi yapıldı. Fiziksel fonksiyonları ve motor becerileri artırmaya yönelik fizyoterapi ve ergoterapi yaklaşımlarını içeren egzersiz ve aktivite çalışmaları planlandı, 6 ay boyunca düzenli olarak haftada 3 gün uygulandı. **Sonuçlar:** Olgunun eklem hareket açıklığı, günlük yaşam aktivitelerindeki bağımsızlığı, yaşam kalitesi ve aktivite katılımının tedavi sonrasında arttığı bulundu. **Tartışma:** Winchester sendromlu vakalarda fizyoterapi ve ergoterapi yaklaşımları önemli bir tedavi seçeneği olabileceği sonucuna varıldı.

Anahtar Kelimeler: Winchester sendromu; Fizyoterapi; Ergoterap; Nöromusküler hastalıklar

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Winchester Syndrome is an autosomal recessive disorder, which is caused by an alteration in MMP2 gene (Rouzier et al., 2006; Zankl, Bonafe, Calcaterra, Di Rocco, & Superti-Furga, 2005). The syndrome was first described by Winchester et al. in 1969 in two sisters aged four and twelve years (Winchester, Grossman, Lim, & Danes, 1969). Between 1969 and 2001 only 12 cases of Winchester Syndrome were reported. The cases were dominantly females with female to male ratio of 3:1 (Ferguson, Mackay, & McNicol, 1970; Matthiesen, Pedersen, Helin, Jacobsen, & Nielsen, 2001; Winter, 1989). This syndrome may be diagnosed relying on clinical and radiological findings; however, the exact etiology is still unknown.

The syndrome usually presents itself in the first year of life with limitations in the movements of the large joints and symmetrical joint swelling. It is also characterized by generalized osteolysis, progressive painful arthropathy with joint stiffness, and contractures of distal phalanges, coarse face, short height, corneal opacities, and osteoporosis and skin changes. Growth restriction is probably due to degenerative changes in vertebras and long bones of the limbs. The osteolysis is most seen in the small bones of the hands and feet. Significant bone deformities like in the vertebral column may be seen due to the pathologic changes of the knee, elbow, hip, shoulder joints and joints of hands and feet (Ferguson, et al., 1970; Matthiesen, et al., 2001; Winchester, et al., 1969; Winter, 1989). In Winchester's original report, symptoms were expressed as painful enlargement of the wrist and proximal interphalangeal joints (Winchester, et al., 1969). Morphological changes may cause limited joint mobility and pain during active movements.

Other non-musculoskeletal symptoms include craniofacial abnormalities. Protruding through forehead, large nose, thickened lips, hypertrophy of gums and with the disease progression coarser face may occur (Winter, 1989). The skin may be thickened and may vary with personal characteristics (Hollister, Rimoin, Lachman, Westin, & Cohen, 1974). Systematic symptoms were also reported as ECG changes and hearth murmurs.

No previous study has been observed on the effects of physiotherapy and occupational therapy in Winchester syndrome in literature. In the light of these observations, it was hypothesized that both physical therapy and occupational therapy have effects on joint limitation of upper and lower extremities and ability of hand in this syndrome. The aim of the present study was to investigate if there are any positive effects of physiotherapy and occupational therapy in a case with Winchester syndrome.

Case Presentation

Our subject was born in 1998 and after birth he had an icterus and hospitalized for two months. Motor developmental milestones, especially head control, were delayed. Macrocephaly, cardiomegaly and hearth murmurs were also found just after birth. Denver test was abnormal. Genetic test results, including fibroblast analysis collagen type I, II and V were normal. Bone age was not matched with calendar age. Low bone density and increased trabeculation were observed. Increased BOS distance in both temporal lobes was identified in computerized tomography. The subject was diagnosed as Winchester Syndrome when he was 5 years old.

The subject walked until 5 years old and lost independent walking with a fracture after falling. Cast correction in the flexed position of the left knee was applied, but leg extension could not succeed again after the removal of the cast. Bilateral Knee Ankle Foot Orthosis (KAFO) was used to provide independent walking during one year, but walking ability was fully lost when the orthoses did not fit anymore.

He has a coarser face and limited mobility in all upper and lower extremity joints especially including hand and knee joints. Therefore, he has several difficulties in gross and fine motor skills.

We are consistent with the principles for human research established in the Declaration of Helsinki. The subject and his parent were asked to sign a written consent form, which has full information about assessments and therapy process. After signing the consent form, the subject, who had not walked for seven years and has several problems in daily living activities and hand skills, underwent physiotherapy and occupational therapy for six months regularly in 2009 after detailed assessments in our department by a physiotherapist and occupational therapist. The assessments were repeated after six-month treatments.

Table 1. Joint limitations of the subject

Limitations of joints (°)	BT		AT	
	L	R	L	R
Elbow flexion	45	92	45	85
Shoulder abduction	50	25	40	0
Shoulder flexion	60	25	40	25
Ankle plantar flexion	30	40	30	30
Knee extension	65	40	50	30
Hip extension	60	60	50	45

BT: Before Treartment; AT: After Treatment; L: Left; R: Right

Measures

Assessment methods and interventions in occupational therapy department were selected and planned according to the activities that he needs and requires and has to do considering the social roles (student, child, etc.) of the subject.

Kamakura's Hand Grips Evaluation Method was used for the assessment of hand grip(Exner, 2005). Duration of writing sentences and hand functions were assessed by Jebsen Taylor Hand Function subtests (Jebsen, Taylor, Trieschmann, Trotter, & Howard, 1969; Kasch, 1996). The fine motor skills' subtests of Bruininks-Oseretsky Motor Proficiency Test 9, including upper limb speed and dexterity, visual motor control and upper limb coordination were applied. These batteries were selected especially considering the student role of the subject. Independence in activities of daily living were assessed by Functional Independence Measurement (Msall et al., 1994) for children (Wee-FIM). Quality of life was evaluated with Child Assessment of Health Questionnaire (CHAQ), (Landgraft, 1996).

Canadian Occupational Performance Measure (COPM) which analyzes the activity performance of children was used. This measure consists of 3 parts including self-care, productivity and leisure. Each activity is composed of 3 problems. The participant scores the activities between 1 and 10, and then, she/he determines 5 activities that are most important for her/his self between these problems. These 5 activities are scored over 10 depending upon the performance and satisfaction. 1 is defined as the `worst score` and 10 is the 'best score` (Carswell et al., 2004; Cusick, Lannin, & Lowe, 2007; Law, 2005). By this way, the individuals determine their performance and satisfaction scores according to their own perspectives.

Because of the negative effects of the disease process on joints, using a manual goniometer assessed the range of motions of elbow, shoulder, wrist, ankle, knee and hip joints. Limitations of the joints were recorded and observational posture analysis in a sitting position was performed before and after treatment.

Trainings

The subject was taken to the physical and occupational therapy for 3 sessions in a week with 60 minutes (20 minutes physiotherapy, 40 minutes occupational therapy) during 6 months, totally 72 sessions.

Subject and his caregivers main goals were to gain independence in daily living activities and upper and lower extremity endurance. Therefore; subject who has limited mobility in wheelchair, begun physiotherapy with active-active assistive upper and lower extremity exercises with a stable bicycle called MotoMed by ReckMedizine (Arisoy et al., 2008) in a proper sitting position. Despite the fact that he is on wheelchair, lower extremity exercise on bicycle was preferred to protect the muscle strength, to stimulate the blood circulation and prevent the progression of joint limitations of lower extremities. The upper and lower extremity training was progressed according to the

toleration of the subject with forward and backward directions from 10-minute durations to 15-20 minutes in advancing resistance within 6 months (72 sessions). Stretching exercises for upper and lower extremities (10 repetitions with 10 seconds waiting in stretched position, two times in a day) were involved in the physiotherapy program and performed daily by caregiver of the child as introduced by the physiotherapist.

Occupational therapy was planned as client centered according to the baseline assessments. Subject and his caregiver wanted to develop the independence in the functions of upper extremity and develop the fine and gross motor skills of the hands such as handgrips, writing ability and speed. Therefore; the training progressed with the aim of strengthening of hand muscles, increasing visual-motor skills and upper limb coordination, improving hand dexterity, increasing writing speed and independence in the activities of daily living. ***Theraputty*** and ***rubber band*** were used for strengthening the hand muscles. Drawing picture, copying a figure and tracing away with using a paper and pen were used for visual motor control training. Throwing and holding a ball was performed to increase upper limb coordination. Using scissor and painting were used for the development of hand dexterity.

Table 2. Results of Hand Function Test

THE JEBSEN TAYLOR HAND FUNCTION TEST (second)	BT		BT	
	L	R	L	R
Writing Duration	3.30		2.21	
Turning page	6	5.25	3.51	2.91
Stacking objects	5	4.37	10.58	12.02
Feeding	9	16.50	6.97	13.41
Pick up objects	7.23	7.06	6.53	6.81

BT: Before Treartment; AT: After Treatment; L: Left; R: Right

Table 3. Results of motor proficiency and daily living activities

Tests	BT	AT
THE BRUININKS-OSERETSKY MOTOR PROFICIENCY TEST (point) (0-34)		
Upper limb coordination	2	2
Upper limb speed and dexterity	20	31
Visual motor	15	20
Wee-FIM (point) (18-126)		
Self care	15	19
Sphincter control	14	14
Mobility	3	3
Locomotion	2	2
Communication	14	14
Social cognition	21	21
Total	69	73

BT: Before Treartment; AT: After Treatment

To improve writing speed, initially, writing on a straight line was tried, and then more complex activities were requested (Fig. 1) after the development in speed within 6 month. All activities were timed in all sessions to increase endurance.

The daily living activities like buttoning up from with wide sized buttons to small sized ones and the types of grips from gross to fine grips which were needed by the subject were practiced. Writing and page turning activities were decided to be important for the subjects' student role. As the subject was a child, all the activities were conducted as the parts of several games. The subject also wanted to write better, and his caregiver wanted him to participate more actively in his self-care like dressing on and feeding. Considering the requires of the subject and his caregiver, occupational therapy program was advanced as practicing the dressing on and undressing his t-shirt and simulating the eating activity with proper sized spoons and crotches.

The physiotherapy and occupational therapy

assessments were repeated again after 6 months (72 sessions) interventions and compared to the first assessments. Limitations of joints before and after intervention are provided in Table 1.

The duration of the active bicycle training increased from 10 minutes to mean 18 minutes after training.

The right wrist of the patient was found rigid in the neutral position in the assessments before and after treatments. There was no active extension of the left wrist. It was found that the 10 degrees active flexion movement of the left wrist joint was increased to 20 degrees after treatment.

Light colored areas were observed in skin examination due to altered pigmentation of the skin.

Distal phalanges of hands and feet were hypoplasia.

The subject was observed as sitting on the sacrum with kyphosis and shoulder protraction was seen in posture analysis.

Finger contact was inadequate in the power grip both before and after training. However, all fingers, except the index and middle fingers, contact on an object were increased in hook grip. Palmar grip and lateral pinch didn't show any difference after training.

The performance of the subject in The Jebsen Hand Function subtests were increased (Table 2).

In The Bruininks-Oseretsky Motor Proficiency Test, there were improvements in the scores of visual- motor control and upper-limb speed and dexterity. After the training, only self-care score was found to have increase in Wee-FIM (Table 3).

There were significant differences between pre and post-training in CHAQ-PF28 scores of the subject in physical functioning, role/social limitation depending on emotional-behavioral and physical causes, bodly pain/discomfort, self esteem, parental impact emotional, parental impact time (Table 4).

The participant reported that he mostly experienced difficulties in productivity (doing homework), leisure and self-care (feeding and dressing). The children reported that his activity performance (from 2 to 6), satisfaction (from 3 to 9) and total scores (from 2 to 8) increased after the treatment.

Table 4. The results of CHAQ PF-28 scores before and after treatment

CHAQ PF-28 subgrups	BT	AT
GGH	60	60
PF	66,6	100
REB	60	100
RP	0	100
BP	20	100
BE	100	100
GBE	100	100
MH	100	100
SE	50	100
GH	27,5	27,5
PE	75	87,5
PT	66,6	100
FA	100	100
FC	100	100

BT: Before Treartment; AT: After Treatment; CHAQ PF-28: Child Assessment of Health Questionnaires- Parent Form-28; PF : Physical functioning; RP: role/ social-physical limitations; GH: general health perception; BP: bodily pain/discomfort; REB :role / social, emotional / behavioural limitations; SE: self-esteem; MH: mental health; BE: behavior; PE: emotional impact on parents; PT: impact on the parent's personal time; FA: limitations in family activities; FC: family cohesion; GBE: global behavior; GGH: global general health

Discussion

In rare disorders such as Winchester Syndrome, it is difficult to plan and perform a rehabilitation program both because the lacking of adequate amount of subjects with same diagnose does not allow to research on a rehabilitation procedure and its short and long term effects and because of the characteristics of the disease. But we also know that, the condition itself formulizes the short and long term goals of the treatment for

each individual (Cup et al., 2007; Dubowitz, 1995; Fowler, 2002). In the base of this point of view, we planned and performed a rehabilitation program both involving physiotherapy and occupational therapy interventions considering the disease characteristics, patient needs and requirements with client centered approach.

Increasing the abilities in daily living activities and function were the primary goals in the treatment for this subject. The secondary goals of the treatment were to prevent the possible further contractures, to provide the optimal sitting position and to help the active movements while protecting the normal range of motion.

According to the test results; all subtest scores were improved in Jebsen Hand Function Test after treatment. It was thought that page turning, writing, eating and self care activities which were all selected as to needs of the subject helped to development in the test results.

According to the Bruininks- Oseretsky Motor Ability Test results; Upper limb speed and dexterity and Visual motor control subtests results were increased, whereas Upper limb coordination subtest results remained the same after the treatment. The assessments of upper extremity coordination also include some grasping types. The lack of development in grasping ability during treatment was an expected result for our subject who had difficulties in accomplishing some daily activities. The muscle strengthening training for hands were thought to pave the way for improvement in grasping types in further months of treatment.

Improvements were obtained in Upper extremity speed and ability test and in the Visual motor control subtest parameters such as cutting figures with scissors and copying basic figures, meanwhile there was no development in copying complex figures. It was difficult for the subject and therapist to progress to more difficult hand ability activities because of the rigid finger joints of the subject.

There were significant differences between pre and post-treatment of the scores of Physical Functioning, Role/ Social limitation emotional/ behavioral, Role/ Social limitation-physical, Bodly Pain/discomfort, Self esteem, Parental impact emotional, Parental impact time. These alterations might be in a relation with improvements in the upper extremity functions that lead to more independency within the limits and more self-confidence. The significant difference shows the positive effects of training on the quality of life of the subject and his family.

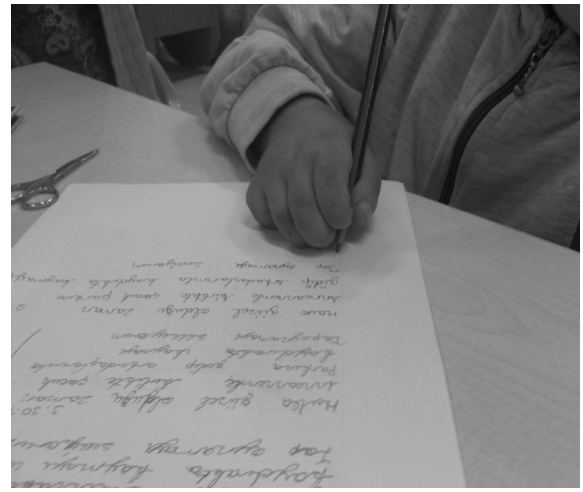

Fig 1. Training on writing activity to develop the writing skills

Self-care points were merely increased in the activities of daily living. Self care activities that were included in the treatment lead to minimal assistance in washing the lower extremities and trunk after treatment by the subject as indicated by the bathing item of the self-care subtest. Zip fastening was accomplished while dressing the lower trunk, but it was still difficult for the subject to fasten the small buttons which was an inconvenient activity because of the need for fine motor ability of the hand. The subject also started to help to clean himself in toilette which gave more courage to his caregiver, therapist and him to progress self care activities.

The increased COPm's scores also prove the increased occupational performance in the activities and satisfaction of child (Dunford, 2011; Petty, McArthur, & Treviranus, 2005; Sakzewski et al., 2011). We also concluded that this measure may be used to reflect the occupational performance of Winchester Syndrome like any other patients in childhood.

The decrease in joint limitation with the stretching exercises after six months was thought

to contribute to the success in upper extremity functions. The significant improvement in the duration of bicycle training showed the development in active participation to movement. This might contribute to more independency in daily living activities.

In conclusion; the originality of the case and the syndrome made us search any therapeutic option for Winchester Syndrome. There were not any study in literature for the supportive therapeutic interventions in this disorder. Furthermore; we designed a patient-centered active physical and occupational therapy program with the primary goals to increase the physical functioning, independency in daily living activities and quality of life while considering the needs and requirements of the caregiver and the subject. The therapy programs which were planned in the basis of these goals then directed and progressed to the needs of the patient with realistic goals. The training program showed increased success in upper extremity functions in this subject. These results indicate that physiotherapy and occupational therapy approaches may be one of the important supportive therapeutic options for the cases with Winchester Syndrome.

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