

The Effect of the Physiotherapy Program in Autosomal Recessive Spastic Ataxia of Charlevoix Saguenay (ARSACS): A Case Report

Neslihan KARABACAK*, Zübeyir SARI**

Abstract

Autosomal recessive spastic ataxia of Charlevoix Saguenay (ARSACS) is a rare neurodegenerative disease caused by a gene mutation. Cerebellar, pyramidal, and neuropathic symptoms have appeared in this disease. The patient participated in a two-week rehabilitation program for six weeks, which included strengthening, balance, proprioception exercises, and virtual reality games. Researchers performed an upper extremity coordination test, lower extremity endurance test, and balance test before- after treatment. Improvements in all measurements were observed and recorded.

Keywords: Ataxia, exercise, coordination, postural balance.

Charlevoix Saguenay Otozomal Resesif Spastik Ataksiside (ARSACS) Fizyoterapi Programının Etkinliği: Bir Olgu Sunumu

Öz

Charlevoix Saguenay otozomal resesif spastik ataksiside (ARSACS) bir gen mutasyonunun neden olduğu nadir görülen nörodejeneratif bir hastalıktır. Bu hastalıkta serebellar, piramidal ve nöropatik semptomlar ortaya çıkmaktadır. Hasta 6 hafta boyunca güçlendirme, denge, propriyosepsiyon egzersizleri ve sanal gerçeklik uygulamalarını içeren haftada iki seans olan bir rehabilitasyon programına katıldı. Araştırmacılar tedaviden önce ve sonra üst ekstremitte koordinasyon testi, alt ekstremitte dayanıklılık testi ve denge testi yaptılar. Tüm ölçümlerdeki gelişmeler gözlemlendi ve kaydedildi.

Anahtar Sözcükler: Ataksi, egzersiz, koordinasyon, postural denge.

Introduction

Ataxias can be studied as a group among neurodegenerative diseases. One of them is Autosomal recessive spastic ataxia (ARSACS) a rare species. It was first described in 1978 and observed in the Quebec / Canada region¹. A mutation in the SACS gene causes this ataxia type². In addition,

Olgu Sunumu (Case Report)

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* Lecturer, Nuh Naci Yazgan University, Vocational High School, Department of Therapy and Rehabilitation, Program of Physiotherapy, Kayseri, Türkiye; Specialist, Marmara University, Institute of Health Sciences, Orthopedic and Manipulative Physiotherapy and Rehabilitation, Istanbul, Türkiye. E-mail: neslikarabacak@gmail.com

ORCID <https://orcid.org/0000-0003-1177-7926>

** Prof. Dr., Marmara University, Faculty of Health Sciences, Department of Physiotherapy and Rehabilitation, Istanbul, Türkiye. E-mail: zsari@marmara.edu.tr ORCID <https://orcid.org/0000-0003-1643-5415>

“SACS” is involved in many protein domains. SACS has recently focused on its potential role in regulating mitochondrial physiology, encoding a protein of unknown function, saccin³.

Patients with ARSACS have neurodegenerative conditions such as slowly progressive cerebellar ataxia, spasticity, amyotrophy, involuntary eye movements (nystagmus), and dysarthria^{1, 4-7}. In addition, most of these patients show signs of early-onset cerebellar ataxia, lower limb spasticity, and peripheral neuropathy⁵. Dysarthria and dysphagia are the main features of ARSACS, but the symptom rate is less⁸. There are also cases reported in different regions^{3,6,7}.

Magnetic resonance imaging typically shows atrophy of the cerebellar vermis and linear hypointensities in the pons⁹. Patients 3-4. become wheelchair dependent income⁹. A case example was published in Turkey in 2018¹⁰.

Physiotherapy and rehabilitation is a conservative method of treatment used in patients with ataxia. This case report aims to investigate the effectiveness of the physiotherapy program in a patient with ARSACS. Thus, researchers are trying to contribute to the studies conducted on rare neurological diseases with our results.

Case Presentation

The patient is male and 32 years old. He did not have any complaints until the age of 22. After returning from the military, the patient consulted a doctor due to involuntary contractions, gait disturbance, and falling problems. In 2010, he was diagnosed with autosomal recessive spastic paraplegia type 11 due to the tests. After eight years, he received a diagnosis of ARSACS with a gene scan. It stated that no other person was diagnosed in the family with a similar disease. He can walk short distances with crutches. He has been using a wheelchair for the last year due to fear of falling and fatigue. The sessions were held two days a week for 45 minutes, and the patient participated in 12 sessions in this study. Researchers used classical rehabilitation programs and virtual reality systems. This program included strengthening, flexibility, balance, coordination, and proprioception exercises (Figure 1). Finally, researchers obtained patient approval regarding the publication of treatment images.

Figure 1. Examples of exercises in physiotherapy



In the first sessions, he was allowed to receive support from the assistive device to maintain balance. In the following sessions, the patient was able to continue the program without help. The patient reported a history of frequently falling before the rehabilitation program. However, his fear of falling decreased after starting the program. He did not fall during the program.

When starting the program, the patient needed unilateral support in a standing position and can now easily maintain his balance without physical assistance. In addition, he reported that at the end of the treatment, the complaints of contraction of the lower extremities decreased significantly, and the contractions in the back and abdomen continued.

The Pedalo Sensamove Balance and Proprioception Test was used to assess balance and proprioception before and after the treatment (Figure 2).

Figure 2. The Pedalo Sensamove Balance and Proprioception test



The balance test score increased from 83% to 90%. The proprioception test score increased from 75% to 81%. In addition, the number of standing up within 30 seconds was examined to assess lower limb endurance. While the number of standing up from sitting was four before the treatment, it increased to eight after (Table 1).

Table 1. The Pedalo Sensamove Balance and Proprioception Test and Lower Limb Endurance Test results before and after the treatment

	Before Treatment	After Treatment
Pedalo Sensamove Balance Test	83%	90%
Pedalo Sensamove Proprioception Test	75%	81%
Lower Limb Endurance	Four repeats	Eight repeats

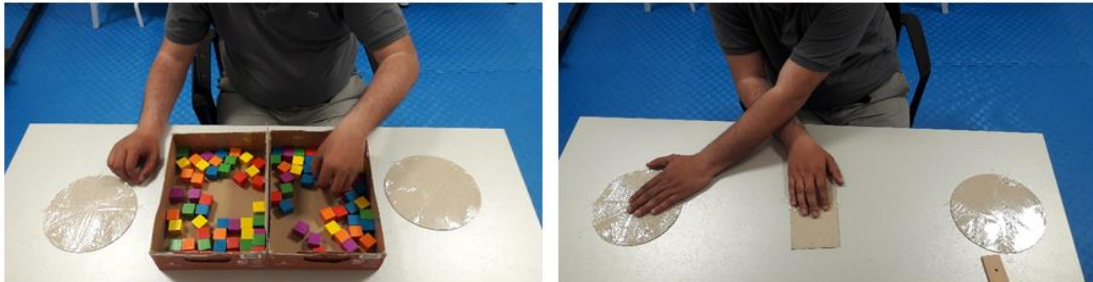
This study used The Box & Block Test, The Disc Test, The Ruler Holding Test, The Supination/Pronation Test, and The Finger-to-Nose Test to assess the upper limb. According to the pre-treatment results, there was an improvement in all measurements (Table 2).

Table 2. Upper extremity test results before and after the treatment

	Before Treatment	After Treatment
Box & Block Test		
Right	39 unit	58 unit
Left	39 unit	55 unit
Disc Test		
Right	37.90 sec	22.24 sec
Left	37.57 sec	23.82 sec
Ruler Holding Test (Dominant)		
1. trial	-	-
2. trial	-	26 cm
3. trial	37 cm	21 cm
Finger Nose Test		
Right	14.66 sec	10.49 sec
Left	17.42 sec	11.84 sec
Supination / Pronation test		
Right	07.22 sec	05.77 sec
Left	08.44 sec	07.08 sec

In the Box & Block Test was calculated the number of blocks thrown from one box to another in one minute. For the disk test, the patient was asked to touch the two disks 25 times in sequence, and this time was recorded (Figure 3). In the ruler holding drop test, when the patient caught the ruler drop from above, the distance on the ruler was displayed just above the thumb record, repeated three times. In addition, the duration of 10 repetitions of the movements for the supination/pronation test and the finger-nose test was recorded and evaluated.

Figure 3. The Box & Block Test and The Disc Test



Discussion

This study was designed to investigate the effectiveness of physiotherapy in an individual diagnosed with ARSACS. Although the severity of the disease generally increases with age, individual performance may differ by age group or stage¹¹. Rehabilitation studies on this subject are limited. An examination of the studies on ataxia rehabilitation revealed that the static and dynamic balance healed in the case study in which virtual reality rehabilitation had been performed in spinocerebellar ataxia¹². In this study, the balance score improved from 83% to 90%. Another study observed motor performance with video-based intensive coordination training in children with degenerative ataxia¹³. This study used the number of times to stand up from the chair to evaluate lower extremity endurance. The developments in the parameters examined are similar to the studies conducted on individuals with ataxia in the literature.

In Audet et al.¹⁴ study, a training program consisting of aerobic and strength exercises was given to 12 participants with ARSACS for eight weeks. It was stated that upper extremity coordination improved after treatment. The patient performed coordination exercises with upper extremity strengthening exercises in this study. The Box & Block Test, The Disc Test, The Supination/Pronation Test, The Finger Nose Test, and The Ruler Holding Test were used for upper extremity evaluation. Significant improvements were noted in all measurements. In a study investigating the effectiveness of 4-week exergaming in patients with spinocerebellar ataxia, the Nine Hole peg test was used. An improvement in this test was observed in the post-treatment period¹⁵. The Box and Block Test was used for upper extremity assessment, and the results improved. In a study investigating the effects of a rehabilitation program to increase balance and

mobility, an 8-week intervention program was applied¹⁶. Trunk control, balance, activities of daily living, mobility and degree of ataxia were evaluated. Improvements in results are promising.

Since this study is a case report, it limits the generalizability of the program applied to people with the stated condition. However, it may be possible to create physiotherapy and rehabilitation treatment protocols by comparing similar cases and case series. He signed the Voluntary/Consent Form.

Conclusion

An individualized combined physiotherapy program positively affected balance, coordination, and lower extremity endurance in patients with ARSACS. Examining the effectiveness of physiotherapy with larger sample groups will contribute to the literature for this disease group.

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