Black Sea Journal of Health Science
doi: 10.19127/bshealthscience.1275640

Open Access Journal
e-ISSN: 2619 – 9041

Case Report
Volume 6 - Issue 3: 494-497 / July 2023

EFFECT OF BALANCE EXERCISES ON BALANCE AND WALKING A PATIENT WITH ATAXIA TELANGIECTASIA

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Abstract: Ataxia Telangiectasia is a rare autosomal recessive disease characterized by progressive neurological dysfunction, immunodeficiency, telangiectasia, chromosomal problems, and sensitivity to radiation, infection, and susceptibility to cancer. A 5-year-old girl with Ataxia Telangiectasia applied to our rehabilitation center due to balance and coordination problems. Step length, double step length, step width and cadence were calculated in the patient and the distance parameters and the time of the walk were examined. The balance of the patient was also evaluated using the pediatric balance scale. It can be said that balance exercises performed in patients with AT provide increases in the patient's balance, coordination and normalize the gait pattern by reducing ataxia.

Keywords: Ataxia Telangiectasia, Balance, Walking, Pediatric balance scale

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Received: April 03, 2023
Accepted: April 26, 2023
Published: July 01, 2023

Cite as: Solgun Dağ S, Canbay Durmaz S, Ateşoğlu Karabaş S, Özbağ D. 2023. Effect of balance exercises on balance and walking a patient with ataxia telangiectasia. BSJ Health Sci, 6(3): 494-497.

1. Introduction

Ataxia Telangiectasia (AT) is a rare autosomal recessive disease characterized by progressive neurological dysfunction, immunodeficiency, telangiectasia, chromosomal problems, and sensitivity to radiation, infection, and susceptibility to cancer (Zaki-Dizaji et al., 2020). AT defective gene ATM (AT mutated); Located on chromosome 11q22-23 (Zaki-Dizaji et al., 2017). The ATM gene is the main gene that controls DNA repair. The mutated ATM gene causes the accumulation of damaged double-stranded DNA. As a result, neurodegeneration, progressive cerebellar atrophy and ataxia develop in the cerebellum (Biton et al., 2008). Over 400 mutations have been identified for AT. In 1941, Madame Louis Bar presented the first case in the literature. The first case was a 9-year-old boy with progressive cerebellar ataxia and bilateral oculocutaneous telangiectasias (Islam et al., 2010). The incidence of the disease is 1: 40.000-1: 100.000 (Zaki-Dizaji et al., 2017).

In patients with AT, the first symptom is seen as cerebellar ataxia at ages 1-5 (Rothblum-Oviatt et al., 2016). However, symptoms such as growth retardation, diabetes, infertility, absence or atrophy of the thymus, neuropathy, oculomotor apraxia, nystagmus, premature aging, dysarthria, dysphagia and mental retardation can be seen during the course of the disease. Serious sinopulmonary infections and cancers are frequently observed in patients with AT due to immunodeficiency (Teive et al., 2015). The first sign of the disease is walking difficulty, which usually occurs when the child learns to walk (Meyts et al., 2003).

Patients need a wheelchair in the 2nd decade of their lives due to progressive movement system disorders and can survive until the 3rd decade. However, patients living up to the 4th and 5th decades are included in the literature (Islam et al., 2010). Treatment in patients with AT is symptomatic and to protect the existing situation. Patients usually die due to problems in respiratory functions and malignancy (mostly lymphoma and leukemia) (Ghiasy et al., 2017).

AT is generally clinically investigated and tried to be defined. Balance exercises have not yet been done in individuals with AT. For this reason, we examined the effect of balance exercises performed with a patient with AT on the patient's balance, walking and step posture.

2. Case Report

A 5-year-old girl with AT applied to our rehabilitation center because of problems with balance and coordination, such as having difficulty standing in a static and dynamic position, tired quickly while playing games with her friends, having difficulty in games that require coordination and being unable to jump. Since our patient's older sister also has AT, our patient has been

under surveillance since she was born, but the diagnosis was made in November 2019 when the patient was 4 years old.

In the Denver II Developmental Screening Test; while his language and personal-social development were similar to his peers, it was observed that he had problems with balancing his fine and gross motor development.

In the anamnesis taken from the family, we learned that the mother and father had a consanguineous marriage, the 13-year-old sister of our patient also had AT and no other children were in the family. We noted that the first complaint of the family about the patient was weakness in the left arm and leg, the patient often had synopulmonary infections and limping in the left leg usually started 2 days before each synopulmonary infection.

In the physical examination; the patient had bilateral ocular telangiectasia, unsupported walking, bilateral gait ataxia, bilateral dysmetria, dysdiadokokinesia, and inability to climb stairs without support. The patient was conscious and speech was smooth. The patient was using only one daily dose of Coenzyme Q10 drug. The patient had not previously received physical therapy service anywhere.

We treated the patient in our rehabilitation center for 8 weeks. Our sessions consisted of 45 minutes sessions 3 times a week for 8 weeks. Balance and coordination exercises were practiced in the sessions. Balance board, svava and platform swing, exercise ball, climbing wall etc. auxiliary equipment such as (Figure 1 and 2). Before the treatment, the walking footprint of the patient was taken and a pediatric balance scale was applied. After the 8-week physical therapy session, the walking footprint and pediatric balance scale were repeated. Fabel Castell finger paint was used for the footprint (Figure 3 and 4). The distance parameters and the time of the walk were checked in the footprint taken.

Figure 1. Balance work on a svava.

Figure 2. Balance work on one leg.

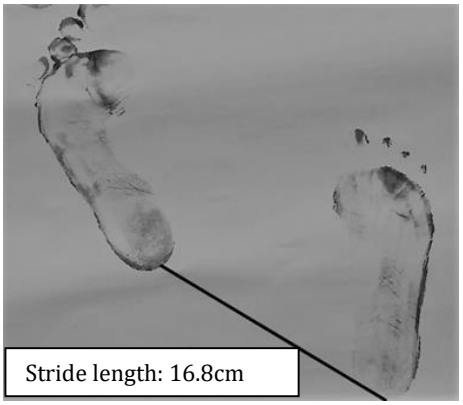

Figure 3. The value stride length, (pre-treatment).

Figure 4. The value stride length, (post treatment).

2.1. Stride Length

The distance between the point of one heel touching the ground and the point of the other heel touching the ground. This is called the right or left stride length. Step length in the patient was 16.8 cm before the treatment. At the end of the 2nd week, the step length decreased in

the horizontal plane and started to increase in the vertical plane and reached 20.6 cm at the end of the 8th week.

2.2. Double Stride Length

The distance between the point of a heel touching the ground and the point of the same heel touching the ground. Before the treatment, this distance was 25.5 cm. The double stride length started to increase at the end of the 2nd week and reached 32.2 cm at the end of the 8th week.

2.3. Step Width

The horizontal distance between the two heel midpoints. This distance is recorded as positive if they are far from each other and negative if they are crossed. The stride width was positive both before and after treatment. Before the treatment, the step width was 19 cm. At the end of the 2nd week, this distance started to decrease and 14,6 cm in the 8th week. It was.

2.4. Cadence

The number of steps in a certain period of time. Usually the number of steps per minute is calculated (Levine et al., 2012). Cadence was 95 steps before treatment. It started to decrease at the end of the 2nd week and reached 80 steps in the 8th week.

2.5. Pediatric Balance Scale

The Pediatric Balance Scale (PBS), a version of the Berg Balance Scale designed for children. To evaluate the functional balance of children in their daily life activities. The scale consists of 14 parts. Each section is scored between 0 and 4; the highest score that can be obtained from the scale is 56. The higher the score, the better the balance, the lower the better the balance, the worse (Tekin, 2016). In the pediatric scale, while the patient scored 38 points before treatment, it increased to 47 points after treatment. These elevations occurred in the parts of standing up from the sitting position, standing without support with the feet together, standing on one foot, turning 360 degrees, taking objects from the ground while standing and placing the foot on a step or stool alternately while standing without support.

3. Discussion

AT is a rare autosomal recessive neurodegenerative disease that usually starts in early childhood and progresses with progressive ataxia, sclera and skin telangiectasias, cutaneous findings, immunodeficiency (Gatti, 1995). In some of the cases, vibration and proprioceptive sense are also lost (Slight et al., 2013). Neurodegeneration in patients with AT appears as peripheral neuropathy, extrapyramidal disorders and cognitive disorders with cerebellar symptoms. In addition, brainstem nuclei and nerve pathways are affected (Hoche et al., 2012). Although cerebellar atrophy is observed in cranial MRI in patients with AT, growth retardation and small head circumference are another common finding (Nissenkorn et al., 2011). Our patient also had telangiectasia, muscle weakness, balance and gait problems.

The first sign of the disease is walking difficulty, which usually occurs when the child learns to walk. In addition, symptoms such as intentional tremor, oculomotor apraxia, and nystagmus may be observed during the course of the disease (Meyts et al., 2003). In our patient, the first to occur was muscle weakness in the left arm and leg.

Yaman M et al. (2005), a 14-year-old female AT patient applied to the clinic due to unstable gait, slurred speech, growth and developmental retardation. It was learned that the patient's complaints appeared after the age of 6, which had normal growth and development until the age of 6. When questioned, it was learned that he had a very frequent lung infection. The clinical findings of our patient are also compatible with this study.

Akarsu Ö.P et al. (2012), a 7-year-old male patient came to their clinic due to hyperemia in both eyes. The patient has had difficulty walking for 2 years. In family history, his older brother also had walking difficulties, bed dependency and hyperemia in the eyes, and it was learned that his parents were second generation relatives. Our patient had similar clinical findings and her parents were also relatives.

The studies carried out mostly focused on the clinical definition of AT. We could not find any study on balance and gait pattern of patients with AT in the literature. Therefore, in our study, we examined the effect of balance exercises performed with a patient with AT on the patient's balance, walking and step posture. We saw the positive reflections of the balance exercises we did with our case on the daily life of the patient. These positive reflections are the independence of descending and ascending stairs, decrease in ataxia in walking, increase in joint position sense, improvement in dysdiadokokinesia, and increase in balance and coordination. At the same time, we saw that muscle strength, hand-eye coordination and movement speed increased. We attribute these increases to increased afferent and efferent input related to balance, cerebro-cerebellar activity and neuroplasticity.

As a result, we can say that balance exercises performed in patients with AT provide increases in the patient's balance and coordination and normalize the gait pattern by reducing ataxia.

Author Contributions

The percentage of the author(s) contributions is present below. All authors reviewed and approved final version of the manuscript.

	S.S.D.	S.C.D.	S.A.K.	D.Ö.
C	30	30	20	10
D	25	25	25	25
S	50	50		
DCP	30	20	30	20
DAI	40	30	20	10
L	30	30	20	20
W	30	30	20	20
CR	20	20	30	30
SR	20	20	30	30
PM	40	20	10	30
FA	25	25	25	25

C=Concept, D= design, S= supervision, DCP= data collection and/or processing, DAI= data analysis and/or interpretation, L= literature search, W= writing, CR= critical review, SR= submission and revision, PM= project management, FA= funding acquisition

Conflict of Interest

The authors declared that there is no conflict of interest.

Ethical Approval/Informed Consent

For this study, necessary permission was obtained Health Sciences Non-interventional Ethics Committee of İnönü University (approval date: March 17, 2020, protocol code: 2020/411). The family was informed about the study and the Informed Consent Form was signed.

Acknowledgements

Thanks to all the individuals who participated in the study for their support.

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