

## *Physiotherapy and rehabilitation approach in ataxia telangiectasia syndrome: A case report*

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### ABSTRACT

**Objective:** Ataxia Telangiectasia (AT) also known as Louis-Bar syndrome, is a rare autosomal recessive genetic disease characterized by cerebellar ataxia, oculocutaneous telangiectasias, and immunodeficiency. This disease is characterized by clinical pictures such as balance, coordination, and gait disorder. This case report aims to examine the short-term effects of physiotherapy and rehabilitation practices consisting of balance and gait exercises on a pediatric patient with AT. **Case:** A 9-year-old girl diagnosed with AT presented with loss of balance. Sociodemographic data of the patient such as age, height, weight, sex, and BMI were recorded. The patient's balance was evaluated with The Pediatric Balance Scale (PBS), The Functional Reach Test (FRT) and The Timed Get Up and Go Test (TUG); the level of functional independence was evaluated with The Pediatric Functional Independence Scale (WeeFIM). The patient received 30 sessions of physiotherapy 4 times per week for 8 weeks, each session 45-60 minutes. Evaluations were made before and after 8 weeks of treatment. **Results:** The pre-treatment values were found to be PBS 22, FRT 16 cm, TUG 17.2 sec, and WeeFIM 62. Post-treatment values were calculated as PBS 22, FRT 18 cm, TUG 15.6 sec, and WeeFIM 62. **Conclusion:** The physiotherapy and rehabilitation approach in patients with AT will be effective in improving symptoms such as balance and coordination disorders.

**Keywords:** Ataxia telangiectasia, Louis-Bar Syndrome, physiotherapy, rehabilitation

### *Ataksi telenjipektazi sendromunda fizyoterapi ve rehabilitasyon uygulamaları: Bir olgu sunumu*

### ÖZET

**Amaç:** Louis-Bar sendromu olarak da bilinen Ataksi Telenjipektazi (AT), serebellar ataksi, okülokütanöz telenjipektaziler ve immün yetmezlikle karakterize olup nadir görülen otozomal resessif geçiş gösteren genetik bir hastalıktır. Bu hastalık denge, koordinasyon ve yürüme bozukluğu gibi klinik tablo ile karakterizedir. Bu olgu sunumunun amacı AT'li bir pediatrik hastada denge ve yürüme egzersizlerinden oluşan fizyoterapi ve rehabilitasyon uygulamalarının kısa dönem etkisini incelemektir. **Olgu:** AT tanısına sahip 9 yaşındaki kız çocuğu denge kaybı şikayeti ile çalışmaya alındı. Olgunun yaş, boy, kilo, cinsiyet ve VKİ gibi sosyodemografik bilgileri kaydedildi. Olgunun dengesi Pediatrik Denge Ölçeği (PDÖ), Fonksiyonel Uzanma Testi (FUT) ve Zamanlı Kalk ve Yürü Testi (ZKYT) ile; fonksiyonel bağımsızlık düzeyi Pediatrik Fonksiyonel Bağımsızlık Ölçeği (PFBÖ) ile değerlendirildi. Olguya 8 hafta boyunca haftada 4 gün 45-60 dakika süre ile 30 seans fizyoterapi uygulandı. Değerlendirmeler tedavi öncesi ve 8 haftalık tedavi sonunda yapıldı. **Bulgular:** Tedavi öncesi değerleri PDÖ 22, FUT 16 cm, ZKYT 17,2 sn, PFBÖ 62 olarak bulundu. Tedavi sonrası değerleri ise PDÖ 22, FUT 18 cm, ZKYT 15,6 sn, PFBÖ 62 olarak hesaplandı. **Sonuç:** AT'li olgularda fizyoterapi ve rehabilitasyon uygulamalarının olgulardaki denge ve koordinasyon bozukluğu gibi semptomların düzeltilmesinde etkili olacaktır.

**Anahtar Kelimeler:** Ataksi telenjipektazi, Louis-Bar Sendromu, fizyoterapi, rehabilitasyon

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## INTRODUCTION

Hereditary ataxia (HA) is a progressive neurodegenerative disorder caused by cerebral atrophy and characterized by motor incoordination.<sup>1</sup> HA is an extremely heterogeneous group of diseases. This disease is classified into 4 main groups according to the inheritance types: autosomal dominant, autosomal recessive, X-linked, and mitochondrial.<sup>2</sup>

Ataxia Telangiectasia (AT) is a rare autosomal recessive neurodegenerative disease characterized by cutaneous telangiectasia, cerebellar atrophy with progressive ataxia, high malignancy, and immunodeficiency.<sup>3</sup> The prevalence of AT is 1 in 40.000-100.000 live births.<sup>4</sup> Clinical findings such as gait, balance, coordination, and speech disorders are seen in AT. In addition, neurological findings such as tremor, dystonia, neuropathy, cardiomyopathy, and retinopathy may also be encountered.<sup>5,6</sup> The progression and severity of the disease vary within the cases with AT. The symptoms in the cases are chronically progressive and there is no standard treatment method. Symptoms lead to a decrease in the quality of life of patients and an inactive lifestyle.<sup>7,8</sup> Studies are showing that physiotherapy and rehabilitation practices improve these symptoms.<sup>9,10</sup>

Upon reviewing the literature, it is noteworthy that there is a lack of research on the impact of rehabilitation and physiotherapy on individuals with AT. The effect of an 8-week physiotherapy and rehabilitation program on functional balance and functional independence in a patient with AT was investigated in this case study.

## CASE REPORT

A 9-year-old girl was admitted to the Physical Medicine and Rehabilitation polyclinic of a university hospital in Anatolia with the complaint of a balance disorder. According to the anamnesis taken from the mother of the case, she was born from a 31-year-old mother in the 39th week of pregnancy with a weight of 4080 grams. She was born with a normal spontaneous birth without intervention in the hospital. In the prenatal follow-ups, there was no eclampsia or bleeding, and no anomaly was detected in the ultrasonographic examinations. There was no history of anoxia at birth. There was no history of sepsis, meningitis, or trauma in the postnatal period. The parents of cases have third-degree relative (first-degree cousin) marriage. She has a 20-year-old sister diagnosed with ataxia telangiectasia and a healthy 14-year-old sister. Her eldest brother was also diagnosed with ataxia telangiectasia and died due to lymphoma when he was 22 years old. No impairment was found

in the cognitive and fine motor skills of the case. The gross motor developmental stages of the case were questioned. The case held her head up at 1 month old; sat unsupported at 6 months old; walked at 13 months old. The patient's family noticed a disability in walking at the age of two. Our case was investigated by pediatric neurology and she was diagnosed with Ataxia Telangiectasia. In the examination dated January 2023, the case had visual impairment and urinary incontinence due to telangiectasias in the eye. She had an ataxic gait pattern. No kyphosis, scoliosis, chest deformity, hip, knee, or foot deformity was detected. A full range of motion was measured in bilateral knee, hip, ankle, and bilateral upper extremity joint. No spasticity was found in the lower extremity and upper extremity muscles of the patient whose Thomas test was bilaterally negative (Stage 0 according to the Modified Ashworth Scale). The patient was included in the rehabilitation program for ataxic gait and balance disorder.

## Physiotherapy and Rehabilitation Program

An evaluation table was created according to the clinical findings of the case. Evaluations were made before and after 8 weeks of treatment. Sociodemographic data of the patient such as age, height, weight, sex, and BMI were recorded. Functional independence was evaluated with the Functional Independence Measure for Children (WeeFIM); the balance was evaluated with the Pediatric Balance Scale (PBS), the Functional Reach Test (FRT) and the Timed Get Up and Go Test (TUG).

After the evaluation of the case, a purposeful physiotherapy and rehabilitation program was designed. The functional status and active participation in the case were taken into consideration in creating the program. It was ensured that the case participated regularly in the program. This program was applied in 30 sessions, 4 times per week for 8 weeks, each session 45-60 minutes. The physiotherapy and rehabilitation program applied to the case included active, active assisted, and passive lower and upper extremity range of motion (ROM) exercises; stabilization exercises including sit-ups, cross body sit-ups, glute bridge, opposite arm and leg raise, plank (Fig.1); balance exercises including, Frenkel exercises in the supine position, pilates and balance board exercises, weight shifting to the right, left, front and back in sitting and standing positions (Figure 2); walking exercises including forward, backward, side and cross walking, various walking speeds, walking over obstacles, taking steps, stopping and turning during walking; stair climbing exercise; fine motor activities including buttoning, zipping/unzipping, lacing (Figure 3).



a-) Sit-Up Exercise



b-) Opposite Arm and Leg  
Raise Exercise



c-) Glute Bridge Exercise

**Figure 1. Stabilization Exercises**



a-) Pilates Ball Exercise

**Figure 2. Balance Exercises**



a-) Buttoning, Zipping/Unzipping,  
Lacing Activities

**Figure 3. Fine Motor Activities**

**Table 1. WeeFIM Sub-dimensions and total values of the case pre-treatment and post-treatment**

	Pre-Treatment	Post-Treatment	Change (%)	MCID
Self-Care	13	13	-	-
Sphincter Control	18	18	-	-
Transfer	8	8	-	-
Locomotion	8	8	-	-
Motor Score Total	47	47	-	-
Communication	7	7	-	-
Social Cognitive	8	8	-	-
Cognitive Score Total	15	15	-	-
Total Score	62	62	-	-

WeeFIM: Functional independence measure for children, MCID: Minimal Clinically Important Difference

**Table 2. PBS, FRT and TUG values of the case pre-treatment and post-treatment**

	Pre-Treatment	Post-Treatment	Change (%)	MCID
Pediatric Balance Scale	22	22	-	3.66-5.83
Functional Reach Test (cm)	16	18	+2 (12.5)	-
Timed Get Up and Go Test (sec)	17.2	15.6	-1,6 (9.3)	10%

PBS: Pediatric Balance Scale; FRT: Functional Reach Test; TUG: Timed Get Up and Go Test, MCID: Minimal Clinically Important Difference

WeeFIM sub-dimensions and total values of the case pre-treatment and post-treatment are shown in Table 1. No changes were observed in these values. PBS, FRT, and TUG values of the case pre-treatment and post-treatment are given in Table 2. 12.5% increase in FRT and 9.3% decrease in TUG were found.

## DISCUSSION

We present the clinical symptoms of a 9-year-old girl diagnosed with ataxia-telangiectasia (AT) and assess the impact of an 8-week physical therapy and rehabilitation program, implemented four days per week. Following treatment, significant improvements were observed in TUG and FRT test results. At the initial evaluation, the patient presented with extremity weakness, loss of balance, dysmetria, and limited independent ambulation. Following 8 weeks of physical therapy, she achieved the ability to walk extended distances without assistance and participate in school games.

Physiotherapy typically focuses on one or multiple domains, frequently utilizing a blend of them: balance, gait, coordination, strength, endurance, and posture.<sup>11</sup> Physical therapy exercises such as traditional techniques, treadmill training, biofeedback therapy, computer-assisted training, relaxation, and supervised sports are among the available intervention approaches.<sup>12</sup> According to Félix et al.<sup>13</sup>, training the inspiratory muscles has a positive effect on the pulmonary function in AT. Also, Rondon-Melo et al.<sup>14</sup> stated that oral rehabilitation is needed early in AT. One of the first studies in this area, Unes et al.<sup>15</sup> found that 3 months of training consisting of balance and strength exercises and Wii Fit Balance-based video games resulted in significant improvements in AT. Other than these studies, there were no studies that demonstrated the benefit of a physiotherapy program. In our case, there were positive improvements in FRT and TUG after 8-weeks. An increase in FRT score by 12.5% was noteworthy. However, the observed 9.3% reduction in TUG was not significant compared to the reported maximum standard error of measurement of 10% for the TUG in children with disabilities.<sup>16</sup>

A critical component, participation is seen as the result of the interaction of bodily processes, physical characteristics, activities, and elements of the environment. Both children with and without disabilities benefit from participation in daily activities, which also has a significant impact on their health and well-being.<sup>15,17</sup> In a present case report, no major changes were observed in the Functional Independence Measure (WeeFIM). Before starting the physical therapy program, our subject had no significant cognitive problems and was an excellent communicator.

In conclusion, positive improvements in both balance and mobility were achieved in a child with AT after an 8-week physical therapy and rehabilitation program. The most important change was the increase in dynamic balance.

## Ethical Aspect of Research

Before starting the study, the aim of the study, the evaluation methods, and the physiotherapy program were explained in detail to the patient and her parents. The written informed consent form was obtained from the parent of the case.

### Author contributions

*Study idea/design:* AT, FT, ÖFÖ, BÇK

*Data collection:* ÖFÖ

*Data analysis and interpretation:* AT, İC

*Literature review:* AT, ÖFÖ

*Critical review:* İC, FT, BÇK

*Final approval and responsibility:* FT, AT

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