

Frederic Ataksili Hastalarda Robot Destekli Yürüme Eğitiminin Yürüme Ve Denge Üzerine Etkisi: Olgu Sunumu

The Effect Of Robot-Assisted Gait Training On Walking And Balance In Patients With Friedreich's Ataxia: Case Report

Hamit YILMAZ¹

ÖZET

Friedreich ataksisi (FA) nadir görülen, hereditör otozomal resesif kalıtmı, progresif nörodejeneratif bir hastalık olup toplumda yaklaşık olarak görülme sıklığı 1/50.000'dir.

Hastalık da genellikle ilerleyici ataksi, kas güçsüzlüğü ve alt ekstremitelerde refleks kaybı gibi nöropatolojik semptomlar ve bunlara bağlı yürüme ve denge kaybı görülür.

Günümüzde FA'nın gelişimini engellemeye yönelik bazı medikal ilaçlar kullanılmaktaysa da tam anlamıyla geliştirilmiş bir tedavi seçeneği yoktur. Fizik tedavi, hastalarda ortaya çıkan semptomik etkilerin baskılanması ve böylece kişinin daha bağımsız kalmasına yönelik tek tedavi seçeneğidir.

6 yıl önce FA tanısı konulmuş 15 yaşında ki erkek hasta olgusunu sunmakta ki amacımız geleneksel fizik tedavi yöntemlerine ek olarak uygulanan robot yardımcı yürüme eğitiminin (RDYE) hastanın fonksiyonelliğine katkısını araştırmaktır.

Anahtar Sözcükler: Frederic Ataksi, robot destekli yürüme eğitimi, denge

ABSTRACT

Friedreich ataxia (FA) is a rare, hereditary autosomal recessive inheritance, progressive neurodegenerative disease, and its incidence in the community is approximately 1 / 50,000.

The disease usually has neuropathological symptoms such as progressive ataxia, muscle weakness, and loss of reflex in the lower extremity, and associated loss of gait and balance.

Although some medical drugs are used to prevent the development of FA today, there is no fully developed treatment option. Physical therapy is the only treatment option for the suppression of symptomatic effects that occur in patients and thus to make the person more independent.

Our patient was a 15-year-old male patient diagnosed with FA 6 years ago. The aim of this study is to investigate the contribution of robot-assisted walking training (RDI), which is applied in addition to traditional physical therapy methods, to the functionality of the patient.

Key words: Friedreich's Ataxia, robot-assisted gait training, walking, balance

¹ Öğretim Görevlisi, Recep Tayyip Erdoğan Üniversitesi Sağlık Meslek Yüksek Okulu Fizyoterapi PR. Rize, Türkiye, hamit.yilmaz@erdogan.edu.tr, ORCID ID: 0000-0002-8324-1891



INTRODUCTION

Friedreich's ataxia (FA) is a rare, hereditary, autosomal recessively-inherited, progressive neurodegenerative disease, and its prevalence in the community is approximately 1/50.000 (1).

Problems, which arise from the recurrence of Guanidin-Adenine-Adenine (GAA) formed in the X25 gene's chromosome 9q13, are responsible for the pathogenesis of FA. Because this gene encodes the frataxin protein, and the decrease in this amount of protein causes iron accumulation in mitochondria and deterioration in oxidative phosphorylation, which leads to an increase in free radicals (12). Therefore, antioxidants were preferred in the treatment of the disease (2). The initial symptoms of FA appear before the age of 25. The disease also includes neuropathological symptoms, such as progressive ataxia, muscle weakness and loss of reflex in the lower extremity, as well as the loss of the ability to walk, and loss of balance. Pathological reflexes and spasticity occur as the corticospinal tract is affected. Patients with hypertrophic cardiomyopathy, scoliosis, and foot deformities are also frequently encountered (3).

In clinical examinations, the sensory conduction velocity in EMNG either slows down or cannot be taken at all. Motor conduction velocities are either normal or slightly reduced. No response can be taken in the sensory evoked potential (SEP) examination. In approximately 1/3 of the patients, visual evoked potential (VEP) and brain stem auditory evoked potential (BAEP) are abnormal (4).

Although some medical drugs are used to prevent the development of FA today, there is no fully developed treatment option. Physical therapy is the only treatment option for the suppression of the symptomatic effects that occur in patients, and thus to make the person more independent.

Our aim in this case report is to investigate the contribution of robot-assisted gait training (RAGT) to the patient's functionality, in addition to traditional physical therapy methods.

CASE REPORT

Our 15-year-old male patient, who was diagnosed with FA 6 years ago, applied to our PTR clinic with complaints of gait disorder and loss of balance. We learned that the complaints of gait disorder, which started at the age of 9, were later followed by hand tremors, unsteadiness in walking, and stumbling while walking at older ages. Three years ago, it was reported that he received 30 sessions of rehabilitation but did not follow the home program. He had not undergone any surgical interventions. It was reported that the patient used idebenone, which aims to reduce free radical production, as well as deferiprone (5mg/kg/day) to reduce the iron accumulation in mitochondria, following his FA diagnosis.

In the background, it was learned that he was delivered through the cesarean section as a full-term at the hospital and

that there was no problem in the postnatal period. It was reported that the normal developmental stages of the patient had a proper course in line with his age and that the symptoms related to FA started to appear at the age of 9 for the first time. There was no history of illness in the family.

The patient, who had an independent ataxic gait during the physical examination, did not use any orthoses, and no DTRs could be obtained from the lower extremity. A Babinski flexor was detected. Romberg's finding was positive. Passive ROM values of the lower extremity were within the normal range.

According to the Medical Research Council's (MRC) scale, hip flexion strength was 4, knee flexion strength was 3, and foot dorsi-plantar flexion strength was 3. During the evaluations carried out based on Bilateral Modified Ashworth Scale (MAS), the lower extremity spasticity in the right hip was found as 3 in the knee and 2 in the ankle, and as 3 in the knee and 2 in the ankle in the left hip. The ankle clonus was grade 2 on both sides as per the Tardieu Scale.

The patient was included in a rehabilitation program consisting of 45 sessions of 60 minutes on a daily basis. Balance exercises, strengthening exercises and stretching exercises for the related muscles were performed in each session during the entire rehabilitation to treat the patient's primary complaint, which is gait disorder. Robot-assisted gait training (RAGT) was added to these traditional FTR practices. RAGT was applied for 5 days a week in 20 sessions of 40 minutes.

The gait robot-controlled all lower extremity joint movements, allowing the patient to walk on a treadmill in the normal walking cycle. During the exercise, the bodyweight support provided by the robot was gradually decreased, and the patient walked on the treadmill by his own weight.

Clinical and functional evaluations were made before and after the treatment. Muscle strength was measured using MR, MAS for spasticity, as well as 6-minute walking test (6MWT), 10-meter walking test, timed up and go (TUG), and Berg balance scale (BBS) within the functional ambulation category among these evaluations.

While lower extremity bilateral muscle strength did not change for hip and ankle, knee muscle strength increased from 3 to 4. 6MWT increased from 340m to 372m. The 10-meter walk test decreased from 25 sec to 19 sec. TUG decreased from 15 to 11. The BBS score increased from 52 to 59.

The evaluation in the functional ambulation category was the same as the one before the treatment.

DISCUSSION

Our patient showed a functional improvement compared to the combined rehabilitation (traditional FTR + RAGT) program that we implemented prior to the treatment. The patient could now walk safer and faster.



RAGT can contribute to FTR in parameters, such as improving the gait and balance, reducing spasticity, and increasing muscle strength.

This combined rehabilitation approach can contribute to treatment in gait problems associated with various neurological disorders. In a study conducted in patients who had a stroke, it was reported that robotic rehabilitation increased the gait speed and endurance but did not make any contributions in independent gait. Moreover, the robotic rehabilitation practice was reported to be ineffective when applied in less than 3 sessions in a week. Therefore, we planned the rehabilitation program as 5 sessions per week (5).

RAGT can make contributions to the treatment by causing an increase in the neuroplasty in the spinal and supraspinal control pathways of the gait. In this context, robotic treatment can be used to treat the symptoms of gait disorders that occur in chronic spinal cord injuries and after a stroke. It can also increase muscle strength, gait ability, and balance parameters (6, 7).

In a study comparing traditional FTR applications and robot-assisted gait training in ataxic patients, the GATR was reported to be as effective as traditional FTR applications (8).

In another study investigating the role of GATR and tDCS in FA rehabilitation, it was reported that there was a significant increase in motor functions and gait pattern according to the Ataxia assessment scale (SARA) in patients, who underwent tDCS and GATR simultaneously (9).

Nielsen et al. reported that RAGT is effective in ataxia-induced walking problems that occur after a brain injury (10).

Strud et al. investigated the efficacy of the Lokomat in multiple sclerosis (MS)-related gait and balance disorder and reported an increase in endurance, along with an improvement in spatiotemporal gait pattern (11).

Although different information is available in the literature about RAGT, it was reported that it could be applied in walking problems induced by many neurological diseases, but the effectiveness of the Lokomat therapy and its contribution to treatment compared to other treatment methods are not fully clear at the moment.

More specific studies with large populations are needed to demonstrate the effectiveness of RAGT in different disorders, such as FA.

Following the combined treatment (RAGT + traditional FTR) applied to our patient, who was the subject of this study, progress was achieved in terms of function evaluations. In this context, RAGT can make contributions

to the improvement of gait and balance, and to the FTR applications for spasticity reduction and muscle strength increase.

REFERENCES

1. Pandolfo M. Friedreich ataxia: the clinical picture. *J Neurol* 2009; 1: 3-8
2. Morgan GE, Michail SM, Murray JM, Larson CP. *Klinik Anesteziyoloji*, 3. baskı, 2004; 9: 178-198.
3. Spacey SD, Szczygielski BI, Young SP et al. Malaysian siblings with Friedreich ataxia and chorea: a novel deletion in the frataxin gene. *Can J Neurol Sci* 2004; 31(3):383-386. PMID:15376485
4. Klockgether T. Ataxias. In Goetz C, *Textbook of Clinical Neurology*. 3rd ed, New York: Saunders, 2007; Chapter 35,765-780.
5. Mehrholz J, Kugler J, Pohl M. Locomotor training for walking after spinal cord injury. *Cochrane Database Syst Rev* 2012;14;11.
6. Mehrholz J, Pohl M, Elsner B. Treadmill training and body weight support for walking after stroke. *Cochrane Database Syst Rev* 2014;23;1.
7. Mehrholz J, Elsner B, Werner C, Kugler J, Pohl M. Electromechanical-assisted training for walking after stroke. *Stroke* 2013;44(10):127-8.
8. Belas dos Santos, M., Barros de Oliveira, C., dos Santos, A., Garabello Pires, C., Dylewski, V., & Arida, R. M. (2018). A comparative study of conventional physiotherapy versus robot-assisted gait training associated to physiotherapy in individuals with ataxia after stroke. *Behavioural neurology*, 2018.
9. Portaro, S., Russo, M., Bramanti, A., Leo, A., Billeri, L., Manuli, A., & Calabrò, R. S. (2019). The role of robotic gait training and tDCS in Friedrich ataxia rehabilitation: A case report. *Medicine*, 98(8).
10. Nielsen, M., Brincks, J., & Nielsen, J. (2009). Effect of robotic gait training on ataxia in patients with acquired brain damage: P1794. *European Journal of Neurology*, 16.
11. Straudi, S., Benedetti, M. G., Venturini, E., Manca, M., Foti, C., & Basaglia, N. (2013). Does robot-assisted gait training ameliorate gait abnormalities in multiple sclerosis? A pilot randomized-control trial. *NeuroRehabilitation*, 33(4), 555-563.
12. Rötig, A., de Lonlay, P., Chretien, D., Foury, F., Koenig, M., Sidi, D., & Rustin, P. (1997). Aconitase and mitochondrial iron-sulphur protein deficiency in Friedreich ataxia. *Nature genetics*, 17(2), 215-217.