

Physiotherapy and Rehabilitation in Fahr's Disease: A Case Report

Deniz TUNCER*, Betül ÇINAR**, Haşmet Ayhan HANAĞASI***

Abstract

Fahr's disease is a rare neurodegenerative disorder characterized by symmetrical and bilateral calcification of the basal ganglia. The purpose of this case report is to evaluate the effects of physiotherapy and rehabilitation program on a patient with Fahr's disease. Biodex Balance System®, Functional Reach Test, Six Minute Walk Test, Short Form 36, Beck Depression Inventory, Barthel Index, and bilateral ankle goniometric measurements were assessed prior to and after completion of the 12-week physiotherapy program. The improvement was observed in static and dynamic balance scores, functional capacity, health-related quality of life, performance in activities of daily living, and depressive symptoms. Physiotherapy may be recommended in patients with Fahr's disease to increase the quality of life by maximizing functional capability and preventing or minimising secondary complications. The long-term effects of physiotherapy should be investigated in more depth in the future.

Keywords: Fahr's disease, neurodegenerative disorders, physiotherapy, rare diseases.

Fahr's Hastalığında Fizyoterapi ve Rehabilitasyon: Olgu Sunumu

Öz

Fahr's hastalığı, bazal ganglionların simetrik ve bilateral kalsifikasyonu ile karakterize, nadir görülen nörodejeneratif bir hastalıktır. Bu olgu sunumunun amacı, Fahr's hastalığı olan bir hastada fizyoterapi programının etkilerini değerlendirmektir. Biodex Balance System®, Fonksiyonel Uzanma Testi, Altı Dakika Yürüme Testi, Kısa Form 36, Beck Depresyon Envanteri, Barthel İndeksi ve bilateral ayak bileği gonyometrik ölçümleri 12 haftalık fizyoterapi programı başlamadan önce ve tamamlandıktan sonra değerlendirildi. Statik ve dinamik denge skorlarında, fonksiyonel kapasitede, sağlıkla ilgili yaşam kalitesinde, günlük yaşam aktiviteleri performansında ve depresif belirtilerde iyileşme gözlemlendi. Fahr's hastalığı tanımlı bireylerde fonksiyonel kapasiteyi en üst düzeye çıkararak yaşam kalitesini artırmak ve ikincil komplikasyonları önlemek veya en aza indirmek için fizyoterapi önerilebilir. İleride yapılacak çalışmalar ile bu hastalarda fizyoterapinin uzun vadeli etkileri daha derinlemesine araştırılmalıdır.

Anahtar Sözcükler: Fahr's hastalığı, nörodejeneratif bozukluklar, fizyoterapi, nadir hastalıklar.

Olgu Sunumu (Case Report)

Geliş / Received: 11.10.2022 & **Kabul / Accepted:** 12.04.2023

DOI: <https://doi.org/10.38079/igusabder.1183826>

* Ph.D., PT (Corresponding Author), Bezmialem Vakıf University, Faculty of Health Sciences, Division of Physiotherapy and Rehabilitation, İstanbul, Türkiye, E-mail: dtuncer@bezmialem.edu.tr [ORCID https://orcid.org/0000-0003-4975-827X](https://orcid.org/0000-0003-4975-827X)

** MSc., PT, Bezmialem Vakıf University, Faculty of Health Sciences, Division of Physiotherapy and Rehabilitation, İstanbul, Türkiye. E-mail: bcinar@bezmialem.edu.tr [ORCID https://orcid.org/0000-0002-0691-0780](https://orcid.org/0000-0002-0691-0780)

*** Prof. Dr., MD, İstanbul University, Faculty of Medicine, Department of Neurology, İstanbul, Türkiye. E-mail: hasmet.hanagasi@istanbul.edu.tr [ORCID https://orcid.org/0000-0002-4624-4428](https://orcid.org/0000-0002-4624-4428)

Introduction

Fahr's disease is a rare inherited or sporadic neurological condition described by German neurologist Karl Theodor Fahr in 1930¹ characterized by abnormal bilateral calcium deposition in the basal ganglia and white matter². Its prevalence is estimated to be <0.5%³. The prognosis of a patient with Fahr's disease is variable and unpredictable⁴. Movement disorders appear as a spectrum of symptoms including fatigability, clumsiness, unsteady gait, dysarthria, slow or slurred speech, dysphagia, involuntary movements, and muscle cramping in Fahr's disease^{5,6}.

Currently, there is no specific treatment that can cure Fahr's disease or limit its progression, only symptomatic therapies are available⁷. Stretching and strengthening the muscles, soft tissue release for spasticity, balance exercises for improving postural stability, relaxation techniques for anxiety, progressive activities from static to the dynamic surface, wide to narrow base, or lower to higher cognition, and gait exercises for gait dysfunction can be used as physiotherapy approaches to promote, maintain, and restore physical, psychological, and social well-being^{8,9}. The present study is aimed to investigate the effects of physiotherapy in a patient with Fahr's disease. To help further clarify the physiotherapeutic treatment approach to subjects with Fahr's disease, this study evaluated the response to physiotherapy in a patient diagnosed with Fahr's disease.

Case Report

This case report refers to a 62-year-old woman diagnosed with Fahr's disease. Before being referred to department, the patient participated in no exercises or functional training. The exercises included the following categories: balance and coordination exercises, stretching and strengthening of lower extremity muscles and mobilization techniques to increase the normal range of motion of the ankle, strengthening abdominal and back muscles, and posture exercises. Circuit training was also performed as dual-task training for functional activities. The patient underwent supervised physiotherapy for 12 weeks, 2 times a week with 2–3 sets of 8–10 repetitions with the intensity level suitable to ensure correct movement patterns.

The blinded assessor obtained the outcomes at baseline and 12 weeks with the postural stability test of Biodex Balance System® (BBS)¹⁰, Functional Reach Test (FRT)¹¹, Six Minute Walk Test (6MWT)¹², Short Form 36 (SF-36)¹³, Beck Depression Inventory (BDI)¹⁴, and Barthel Index (BI)¹⁵. Active and passive ankle dorsiflexion range of motion (ROM) were measured with a universal goniometer and the average value was recorded for both sides. Static postural stability was measured with the postural stability parameter of BBS, which evaluates the person's ability to maintain the center of balance on a stable platform and records deviations from the center. A high score indicates poor postural stability. Dynamic postural stability was measured with FRT. The test was performed three times, and the average of the last two was recorded. Longer reaching distances indicate better dynamic stability. Functional capacity was measured with 6MWT. The distance covered over a time of six minutes is used as the outcome by which to compare changes

in performance capacity. SF-36, BDI, and BI were also used for evaluating the quality of health, depressive symptoms, and performance in activities of daily living, respectively. Higher scores mean better performance for SF-36 and BI although lower scores indicate less depressive symptoms for BDI. Informed consent was obtained and the rights of the subject were protected.

According to the average value of active and passive ankle dorsiflexion ROM were increased from a neutral degree (absence of dorsiflexion) to 10° and from 10° to 15°, respectively. Overall, anterior/posterior, and medial/lateral stability index and functional reach distance were better after treatment. The six-minute walk distance was increased 85 meter with the treatment program (Table 1).

Table 1. Results of static and dynamic postural stability and functional capacity scores before and after physiotherapy.

	Before physiotherapy	After physiotherapy
Range of Motion (ROM) °		
Ankle dorsiflexion ROM (active)	N	10
Ankle dorsiflexion ROM (passive)	10	15
Biodex Balance System®		
Postural Stability Test (SI)		
Overall SI	1.4±0.61	0.7±0.46
Anterior/posterior SI	0.7±0.62	0.6±0.47
Medial/lateral SI	1.2±0.49	0.2±0.20
Functional Reach Test (cm)	11	17
6 MWD (m)	260	345

N: Neutral, SI: Stability index, MWD: Minute walk test. Data are presented as mean ± standard deviation

While the SF-36 and BI scores increased, the BDI score decreased following the physiotherapy program (Table 2).

Table 2: Results of quality of life, performance in activities of daily living scores, and depressive symptoms before and after physiotherapy.

	Before physiotherapy	After physiotherapy
Short Form 36 (total score)	325.5	372.3
Barthel Index	80	85
Beck Depression Inventory	20	8

Discussion

These results showed an improvement in static and dynamic balance, functional capacity, quality of life, performance in activities of daily living, and depressive symptoms in the patient diagnosed with Fahr's disease.

Although patients with Fahr's disease display different clinical features, the most common findings in these patients are movement disorders similar to Parkinsonism and hyperkinetic movement disorders. In terms of prevalence, these findings are followed by cognitive impairment and hypokinetic movement disorders due to cerebellar involvement. The clinical features are less frequently accompanied by gait disturbances and psychiatric and emotional changes. In this case, there were problems in maintaining postural stability and balance, increased depressive symptoms, and functional inadequacies. Compared with the scores before the physiotherapy program, the ones following intervention turned out indicative of clinical improvement in static and dynamic balance and functional capacity. After 4 weeks, the patient walked without using her cane and was able to make transfers independently and climb stairs under supervision. Functional reach test is associated with an increased risk of fall and frailty in elderly people who are unable to reach more than 15 cm¹¹. In this patient, the functional reach distance, which was measured as 11 cm before the physiotherapy, was measured as 17 cm after the physiotherapy. Fahr's disease is a chronic and progressive neurological disease and performance in activities of daily living may include difficulties for many patients. There are also neuropsychiatric features of this disease¹⁶. The results of this study reveal an improvement in performance in activities of daily living measured by BI and a reduction of depression symptoms measured by the BDI after the rehabilitation program. The severity of depression increased from moderate to minimal. Before the rehabilitation program, the patient did not have heel strikes because of limited ROM in ankle dorsiflexion while walking and needed a unilateral cane to maintain her balance. At the end of the program, she was able to perform the heel strike on the stance phase without the need for an ambulation aid and increased the 6-MWD by 85 meters. It should be underlined that different degrees of severity and symptoms can be seen in a patient with Fahr's disease or it can remain asymptomatic throughout life⁷, therefore, specific exercise training approaches targeting specific disorders in these patients may be necessary. Clarifying the benefits of each type of exercise included in the program will help developing a specific exercise prescription for these patients.

There is no certain cure for Fahr's disease yet, various treatment strategies are used to relieve symptoms and prevent remissions^{4,7}. So it is unclear if a long-term physiotherapy program will affect physical function in these individuals. Since not encountered any physiotherapeutic approach in the published literature related to these patients, created a 12 weeks exercise program although it was not specific to Fahr's disease including balance and coordination exercises, stretching and strengthening of lower extremity muscles, and mobilization techniques to increase normal range of motion, strengthening abdominal and back muscles, and posture exercises.

Conclusion

Fahr's disease is a rare disease with impaired functional performance and an increased level of disability and has no cure. Hence a multidisciplinary rehabilitation team approach may be needed to improve functional skills and quality of life. Based on the results of this case report believed that physiotherapy may play a pivotal role in the long-term management and follow-up of Fahr's disease due to its incurable and natural progression.

Ethical Approval

No personal details or identifying information are included in this article. Informed consent was obtained and signed by the patient prior to the study procedure and preparation of the case report for the purpose of publication. Ethical approval was not needed.

REFERENCES

1. Fahr T. Idiopathische verkalkung der hirngefasse. *Zentralbl Allg Pathol.* 1930;50:129-133.
2. Manyam B, Walters A, Keller I, Ghobrial M. Parkinsonism associated with autosomal dominant bilateral striopallidodentate calcinosis. *Parkinsonism & Related Disorders.* 2001;7(4):289-295. doi:10.1016/s1353-8020(00)00036-5.
3. Kechaou I, Boukhris I. Hypoparathyroidism and early onset dementia: Fahr syndrome should be suspected. *The Pan African Medical Journal.* 2018;30:82-82. doi:10.11604/pamj.2018.30.82.15587.
4. Saleem S, Aslam HM, Anwar M, et al. Fahr's syndrome: Literature review of current evidence. *Orphanet Journal of Rare Diseases.* 2013;8(1):1-9. doi:10.1186/1750-1172-8-156.
5. Manyam BV. What is and what is not 'Fahr's disease'. *Parkinsonism & Related Disorders.* 2005;11(2):73-80. doi:10.1016/j.parkreldis.2004.12.001.
6. Asokan AG, D'souza S, Jeganathan J, Pai S. Fahr's syndrome-an interesting case presentation. *Journal of Clinical and Diagnostic Research: JCDR.* 2013;7(3):532. doi:10.7860/JCDR/2013/4946.2814.
7. Pistacchi M, Gioulis M, Sanson F, Marsala SZ. Fahr's syndrome and clinical correlation: A case series and literature review. *Folia Neuropathologica.* 2016;54(3):282-294. doi:10.5114/fn.2016.62538.
8. di Biase L, Munhoz RP. Deep brain stimulation for the treatment of hyperkinetic movement disorders. *Expert Review of Neurotherapeutics.* 2016;16(9):1067-1078.
9. Amisha F, Munakomi S. Fahr syndrome. In StatPearls; Stat Pearls Publishing: Treasure Island, FL, USA. 2023 Jan. PMID: 32809692. Accessed March 11, 2023. <https://www.ncbi.nlm.nih.gov/books/NBK560857/>.

10. Cachupe WJ, Shifflett B, Kahanov L, Wughalter EH. Reliability of biodex balance system measures. *Measurement in Physical Education and Exercise Science*. 2001;5(2):97-108.
11. Duncan PW, Weiner DK, Chandler J, Studenski S. Functional reach: A new clinical measure of balance. *Journal of Gerontology*. 1990;45(6):M192-M197. doi:10.1093/geronj/45.6.m192.
12. ATS statement: Guidelines for the six-minute walk test. *Am J Respir Crit Care Med*. 2002;166:111-7. doi: 10.1164/ajrccm.166.1.at1102.
13. Ware JE. *SF-36 Health Survey: Manual And Interpretation Guide*. New England Medical Center, *Health Institute*. 1993.
14. Beck AT, Ward C, Mendelson M, Mock J, Erbaugh J. Beck depression inventory (BDI). *Arch Gen Psychiatry*. 1961;4(6):561-571. doi:10.1001/archpsyc.1961.01710120031004.
15. Collin C, Wade D, Davies S, Horne V. The Barthel ADL Index: A reliability study. *International Disability Studies*. 1988;10(2):61-63. doi:10.3109/09638288809164103.
16. Weisman DC, Yaari R, Hansen LA, Thal LJ. Density of the brain, decline of the mind: An atypical case of Fahr disease. *Archives of Neurology*. 2007;64(5):756-757. doi:10.1001/archneur.64.5.756.