DOI: 10.4274/npa.y5580

Fahr's Disease: A Case Report

Fahr Hastalığı: Olgu Sunumu

Orhan DOĞAN, Güngör MEYDAN, Murat SEMİZ, Osman YILDIRIM, Gözde YONTAR
Cumhuriyet Üniversitesi Tıp Fakültesi, Psikiyatri Anabilim Dalı, Sivas, Türkiye

ABSTRACT

Fahr's disease is a rare neuropsychiatric illness characterized by bilateral calcifications of the basal ganglia and white matter. Although its etiology is not yet fully understood, infectious, metabolic and genetic factors have been demonstrated in some cases. Movement disorders, together with dementia and psychiatric symptoms, may be observed as the clinical manifestations of Fahr's disease. In this case report, a 53-year-old male patient who had complaints of skepticism and nervousness was reported. His physical examination revealed signs of dementia. The serum ionized calcium, phosphorus and parathyroid hormone levels were within normal limits. A computed tomography scan of the brain demonstrated bilateral calcifications. *(Archives of Neuropsychiatry 2011; 48: 82-4)*

Key words: Dementia, Fahr's disease, psychosis

ÖZET

Fahr hastalığı bazal ganglionların ve beyaz cevherin bilateral kalsifikasyonu ile karakterize, nadir görülen bir nöropsikiyatrik hastalıktır. Etiyolojisi tam olarak aydınlatılamamış olup bazı vakalarda infeksiyöz, metabolik ve kalıtsal nedenler gösterilmiştir. Fahr hastalığının kliniğinde hareket bozukluklarının yanı sıra, demans ve psikiyatrik belirtiler görülebilir. Bu yazıda şüphecilik ve sinirlilik yakınması bulunan, muayenesinde demansiyel bulguları olan, serum iyonize kalsiyum düzeyi, fosfor ve paratiroid hormon düzeyleri normal olan, bilgisayarlı beyin tomografisinde bilateral kalsifikasyonları olan 53 yaşında erkek hasta sunulmuştur. *(Nöropsikiyatri Arşivi 2011; 48: 82-4)*

Anahtar kelimeler: Demans, Fahr hastalığı, psikoz

Introduction

Fahr's disease (FD) was described in 1930 by Fahr as idiopathic calcifications of the cerebral blood vessels identified in an adult patient who presented with progressive neurological symptoms. FD is a rarely observed syndrome characterized by bilateral, symmetric intracranial calcifications and neuropsychiatric disorders (1). Metabolic diseases and infectious diseases may play role in its etiology (2). The relationship of FD with abnormal phenotypes and abnormal genes has not been clearly described and attention has been drawn to chromosome 14 (3,4).

The diagnosis of FD is based on the evaluation of three main features, namely bilateral idiopathic non-atherosclerotic calcification of the basal ganglia, psychiatric symptoms, and choreoathetotic and extrapyramidal movement disorders (5). Computed tomography (CT) is more effective than magnetic resonance imaging (MRI) in the diagnosis of FD; it demonstrates calcified areas in the cerebellum and basal ganglia. On histological evaluation of FD, calcifications are observed on the vessel walls and perivascular spaces of arterioles, capillaries, and veins (6). The movement disorders and psychiatric symptoms in these patients are associated with calcification of the basal ganglia (7).

Case

A 53-year-old, male, married patient, who was high school graduate and civil servant, has complained of forgetfulness, skepticism, nervousness, chuckling, and irrelevant behaviors over the past 5-6 years. He was on the verge of divorcing his wife due to the skepticism. He could not remember names, forgot things to do and places of objects, he was disoriented to time and place, and talked meaninglessly from time-to-time. The patient and his family had no history of psychiatric disease. The mental state examination demonstrated that he could no longer

Address for Correspondence/Yazışma Adresi: Dr. Orhan Doğan, Cumhuriyet Üniversitesi Tıp Fakültesi, Psikiyatri Anabilim Dalı, Sivas, Türkiye
Gsm: +90 532 342 02 02 E-mail: ordogan@gmail.com **Received/Geliş tarihi:** 15.03.2010 **Accepted/Kabul tarihi:** 25.06.2010

maintain proper self-care, talked meaninglessly from time-to-time, had emotional blunting, and slowed thought process, including paranoid delusions. He showed no perceptual deficit. Chuckling to himself was very obvious in his extroverted behavior. The patient scored 36 points on the Brief Psychiatric Rating Scale (BPRS). His standardized Mini-Mental State Examination (MMSE) score was 18. On MMSE, there was a decrease in orientation, attention, calculation, and recall; therefore, the executive functions were impaired. The patient was diagnosed with a subdural hematoma following a head trauma 4 months ago. His physical examination was normal. The neurological examination demonstrated that the patient was conscious, disoriented to time, place, and person, and uncooperative. The cranial nerve, motor and sensory examinations were normal, the deep tendon reflexes were normoactive, and there was no movement disorder. The ionized calcium, phosphorus, parathyroid hormone (PTH), thyroid hormones, serum glucose and lipid levels, as well as vitamin B12 and folic acid levels, were within normal limits; liver and kidney function tests showed no abnormalities. The patient was negative for hepatitis markers, anti-HIV marker, and C-reactive protein (CRP). His brain CT scan demonstrated symmetric band-like calcifications in the periventricular region, bilateral millimeter-sized nodular calcifications localized in globus pallidus, and a chronic subdural hematoma was observed in the right parietal region (Figure 1, 2, and 3). His electroencephalogram was normal.

The patient was diagnosed with FD based on evaluation of his medical history, neurologic and psychiatric examinations, and brain CT scan. Treatment with quetiapine (300 mg/day) was initiated. The symptoms of chuckling and self-talking were partially resolved; however, the quetiapine dose was increased to 600 mg/day due to persisting paranoid thoughts and nervousness. By the 3rd week of treatment, marked improvement was observed in the patient's complaints, such as chuckling to himself, self-talking, nervousness, and skepticism. Prior to discharge, the BPRS score decreased to 11 points, but dementia remained.

Discussion

FD is characterized by widespread calcification of the basal ganglion. The causes of calcification of the basal ganglion have been classified as idiopathic, FD, post-inflammatory, congenital and post-anoxic/toxic. FD is the most commonly encountered cause of basal ganglion calcification (8) and may also develop in association with various metabolic disorders (9); in our case, there was no metabolic disorder. Ionized calcium level decreases in cases of hypoparathyroidism and pseudohypoparathyroidism, therefore, clinical signs of tetany and convulsions may be seen (10). In congenital diseases, such as tuberosclerosis, Down syndrome, some abnormalities may be identified in other organ systems together with calcification of the basal ganglion (11).

Boller et al. (12) reported calcification of the basal ganglion that demonstrated autosomal dominant inheritance in some cases. No genetic analysis for possible genes in this case, however, the family history did not suggest any possible genetic inheritance.

The brain CT scan can be used in the diagnosis of FD since it is easy to identify hyperdense areas (7). The brain CT scan of our patient showed bilateral band-like calcifications in the periventricular regions and millimetric nodular calcifications in the globus pallidus. Various metabolic and infectious factors may cause visualization of calcifications in the cranial imaging procedure; in particular, identification of bilateral symmetric calcifications in the basal ganglia may suggest FD. Maghraoui et al. (14) reported that FD may accompany psychiatric disorders, dementia, seizures, extrapyramidal symptoms, and various neurologic conditions, and suggested that brain CT scan is beneficial in the diagnosis of FD.

Psychiatric disorders and extrapyramidal symptoms may be associated with calcification of the basal ganglia (2). Extrapyramidal symptoms were not observed in our patient, however, we suggested that paranoid delusions may be related with calcification of the basal ganglion.

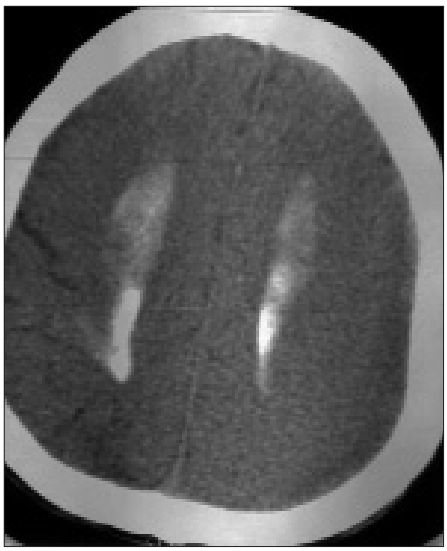

Figure 1. CT scan of our case

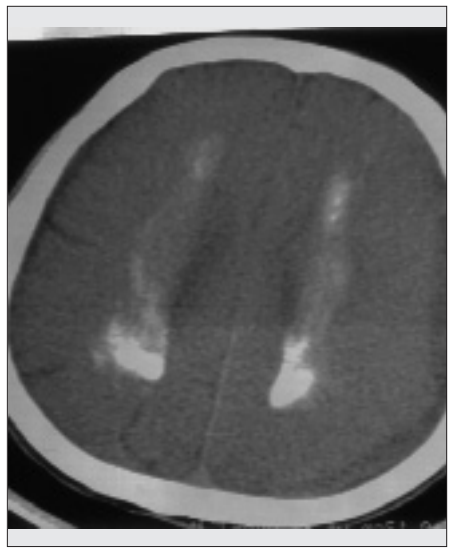

Figure 2. CT scan of our case

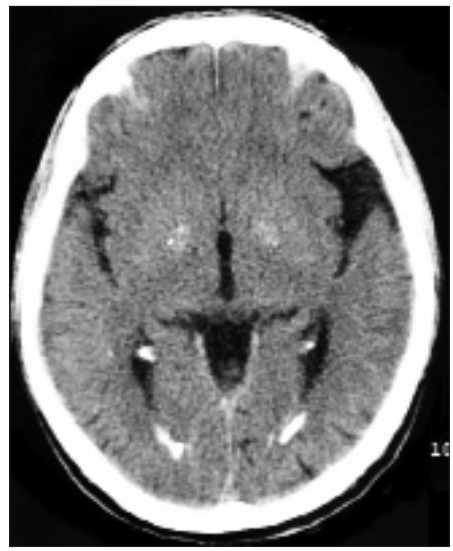

Figure 3. CT scan at basal ganglia level

The patients with calcification of the basal ganglion may be more sensitive to the extrapyramidal side effects of antipsychotics (15); thus, quetiapine was administered for treatment. The patient's psychotic symptoms improved and no extrapyramidal side effects were observed.

In conclusion, patients with adult-onset psychotic symptoms and/or movement disorders and/or complaints of dementia should be subjected to investigation of an organic cause. FD should be considered following the identification of the signs and symptoms of neuropsychological disease as well as calcifications observed on brain imaging.

References

1. Acou M, Vanslembrouck J, Deblaere K et al. JBR-BTR. 2008; 91:19. [Abstract]
2. Boller F, Boller M, Gilbert J. Familial idiopathic cerebral calcification. J Neurol Neurosurg Psychiat 1977; 40:280-5. [Abstract] / [PDF]
3. Chiu HF, Lam LC, Shum PP et al. Idiopathic calcification of the basal ganglia. Postgrad Med J 1993;69:68-70. [Full Text] / [PDF]
4. Cummings JL, Gosenfield LF, Houlihan JP et al. Neuropsychiatric disturbances associated with idiopathic calcification of the basal ganglia. Biol Psychiatry 1983; 18:591-601. [Abstract]
5. Gulsun M, Baykiz AF, Kabatas S et al. Fahr syndrome. Three cases presenting with psychiatric signs. Eur J Gen Med 2006; 3:35-40.
6. Kotan D, Aygul R. Familial Fahr disease in a Turkish family. South Med J 2009; 102:85-6. [Abstract]
7. Maghraoui A, Birouk N, Zaim A et al. Fahr syndrome and dysparathyroidism. 3 cases. Presse Med 1995; 24:1301-4. [Abstract]
8. Malik R, Panday VK, Naik D. Fahr's disease. A rare neurodegenerative disorder. Int J Radiol Image 2004; 14:383-4. [PDF]
9. Moskowitz MA, Winickoff RN, Heinz ER. Familial calcification of the basal ganglions: a metabolic and genetic study. N Engl J Med 1971; 285:72-7. [Abstract]
10. Aslantekin N, Çelik Y, Yeni N ve ark. Fahr hastalığı olan iki hasta: Vaka bildirimi. Yeni Symposium 1999; 37:60-3.
11. Sobrido MJ, Hopfer S, Geschwind DH. Familial idiopathic basal ganglia calcification. Pagon RA, Bird TC, Dolan CR, Stephens K, editörler. Gene Reviews [Internet] içinde. Seattle (WA): University of Washington, [updated 2007 Sep 20].
12. Narayan SK, Sivaprasad P, Sahoo RN et al. Teaching video NeuroImage: Chvostek sign with Fahr syndrome in a patient with hypoparathyroidism. Neurology 2008; 71:e79. [Full Text] / [PDF]
13. Oliveira JR, Spiteri E, Sobrido MJ et al. Genetic heterogeneity in familial idiopathic basal ganglia calcification (Fahr disease). Neurology 2004; 63:2165-7. [Abstract] / [Full Text] / [PDF]
14. Ozkur A, Sirikci A, Bayram M. Fahr disease: BT bulgulari. Journal of Turkish Radiology 2001; 7:142-3.
15. Stip E, Black N, Ekoé JM et al. Fahr's disease and Asperger's syndrome in a patient with primary hypoparathyroidism. J Neurol Neurosurg Psychiatry 2000; 68:115-6. [Full Text] / [PDF]