Submitted: 12 May, 2022 Accepted: 28 June, 2022 Published: 08 July, 2023 DOI:10.22514/sv.2023.066

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CASE REPORT

A case of Fahr disease with transient slurred speech and abnormal movement of the left arm

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Abstract

Fahr disease is a rare neurological condition characterized by abnormal basal ganglia calcification. Some patients with Fahr disease have been reported to present to the emergency department (ED) with stroke-like symptoms. Here, we present a case of Fahr disease with transient focal neurological signs. A 63-year-old man came to the ED with complaints of slurred speech and abnormal movement of his left arm. His symptoms lasted only 5 minutes before he recovered. He had no focal neurological deficits when he arrived at the ED, had no previous medical history and had never taken any medication. A physical examination revealed that the patient was healthy, and the initial results of laboratory tests were all within normal range. A brain computed tomography (CT) without contrast enhancement revealed symmetric calcification of the basal ganglia, thalamus and dentate nucleus of the cerebellum. Magnetic resonance imaging of the brain and neck CT angiography revealed no evidence of acute cerebral infarction, intracranial large-vessel occlusion, or stenosis. Based on his clinical presentation and examination results, the patient was diagnosed with Fahr disease in the ED. He was recommended to be admitted for differential diagnosis of seizures and additional tests, such as electroencephalography, but opted for discharge against medical advice. Emergency physicians should consider the possibility of Fahr disease if patients have focal neurological signs with characteristic symmetric calcification on brain CT. A detailed history and examination should be performed to detect other secondary causes associated with Fahr syndrome.

Keywords

Fahr disease; Basal ganglia calcification; Computed tomography

1. Introduction

Fahr disease is a rare, genetically dominant inherited neurological condition characterized by abnormal idiopathic calcification of the basal ganglia. The most commonly involved regions in brain imaging are the basal ganglia, namely the globus pallidus, caudate nucleus, and putamen [1], but the disease can also occur in the thalamus, cerebral cortex, cerebellum, and hippocampus. 'Fahr disease' refers to a primary case that occurs without a specific cause, while the term 'Fahr syndrome' has been suggested when a secondary, potentially treatable cause is found, for instance, an endocrinological disorder, mitochondrial myopathies, a dermatological disorder, and an infectious disease [2, 3].

Fahr disease/syndrome has been linked to various clinical symptoms, including movement disorders such as rigidity, hypokinesia, tremor, choreoathetosis, and ataxia, as well as behavioral disorders such as dementia and mood disorders [3, 4]. Further, some patients present at the emergency department (ED) complaining of stroke-like symptoms or focal neurological signs such as headache, syncope, seizure, and hemiparesis [5, 6].

In this study, we present a case of Fahr disease with acute onset of slurred speech and abnormal movement of the left arm.

2. Case report

A previously healthy 63-year-old man presented at the ED with a sudden onset of slurred speech and abnormal left arm movement. He had experienced slurred speech at his table 40 minutes before presenting at the ED and had an increased tone of the left arm with twitching and involuntary rhythmic movement. His symptoms lasted for only 5 minutes before he recovered. He had no aura, automatism, or seizure-like symptoms, such as eyeball deviation, lip cyanosis, tongue bite, or an 'epileptic cry'. On arrival at the ED, he complained of general weakness but was otherwise asymptomatic. His blood pressure was 98/66 mmHg, heart rate was 93 beats/min, and body temperature was 36.0 °C. He had no medical history of hypertension, diabetes mellitus, or hyperlipidemia and had never taken any medication. No abnormalities were found on physical examination. Neurological examination revealed

normal motor power of the extremity with no lateralizing signs and discernible dysarthria. Initial laboratory results showed a white blood cell count of 8240/μL with 42.1% neutrophils, hemoglobin level of 14.0 g/dL, platelet count of 266,000/μL, sodium level of 143.0 mmol/L, potassium level of 3.8 mmol/L, lactic acid level of 2.2 mmol/L, myoglobin level of 33 ng/mL, creatine phosphokinase level of 99.0 U/L, ammonia level of 29 μmol/L, a glucose level of 84.0 mg/dL, and an ionized calcium concentration of 1.22 mmol/L (reference range 1.12–1.32 mmol/L). Lipid profiles, such as cholesterol and triglyceride levels, were not tested. Electrocardiography showed sinus rhythm without ST-segment changes. Brain computed tomography (CT) without contrast enhancement revealed symmetric calcification of the basal ganglia, thalamus and dentate nucleus of the cerebellum. Acute intracranial hemorrhage and territorial infarction were not observed (Fig. 1). Magnetic resonance imaging (MRI) of the brain showed a T2-weighted image with low signal intensity in both basal ganglia. A low signal intensity in diffusion-weighted imaging and an apparent diffusion coefficient map was observed but without restricted diffusion, suggesting acute cerebral infarction (Fig. 2). Neck CT angiography revealed no evidence of stenosis or occlusion of the carotid artery, vertebral artery, or large intracranial vessels. The patient was initially diagnosed with Fahr disease in the ED because his brain imaging findings and focal neurologic signs were consistent with Fahr disease. He was recommended to be admitted for differential diagnosis of seizures and perform additional evaluations, such as electroencephalography, but he was discharged against medical advice and without medication due to complete recovery of his neurological symptoms.

3. Discussion

Fahr syndrome/disease is a neurological disorder in which there are abnormal calcium deposits in areas of the brain that control motor activity, causing neuropsychiatric symptoms. The term 'Fahr syndrome' is used when basal ganglia calcification is associated with a secondary cause, such as parathyroid dysfunction, particularly hypoparathyroidism, which is the most common etiology [3]. Various potential causes of intracranial calcification are associated with Fahr syndrome, such as birth anoxia, tuberous sclerosis, astrocytoma, cysticercosis, toxoplasmosis, human immunodeficiency virus infection, and so on [7]. After ruling out any other causes of neuropsychiatric symptoms with intracranial calcification, the patient was diagnosed with Fahr disease. In this case report, the patient was healthy before the incident and had never had developmental disorders, seizures, infections, or poisoning. Laboratory results, including blood ionized calcium levels, were all within normal limits, indicating no metabolic disease or other systemic disorders. Brain CT and MRI revealed bilateral and symmetric calcification in the basal ganglia, thalamus, and cerebellum, consistent with Fahr disease radiological findings and without findings suggestive of acute stroke.

Several case reports have been published in which patients with Fahr disease/syndrome presented to the ED with speech abnormalities or dysarthria as their primary complaint. However, in these cases, the symptoms usually lasted several weeks or occurred as a recurrent history; unlike in our case [8, 9]. Although the pathophysiology of Fahr disease remains unclear, it has been reported that decreased blood flow to the calcified regions due to calcium deposits begins in the third decade and is usually associated with clinical signs [10]. Sava *et al.* [11] demonstrated the presence of calcium deposits in various forms and degrees within the walls of capillaries and small and medium arteries on histopathological examination. Uygur *et al.* [12] confirmed significantly decreased regional blood flow in the intracerebral calcified regions using cerebral perfusion single-photon emission computerized tomography of the brain with technetium-99m hexamethylpropyleneamine oxime, and was reported to lead to transient focal ischemic changes [13]. These symptoms typically appear gradually and progressively later during the progression of the disease. However, in this case, the patient's symptoms appeared suddenly with temporary neurological signs. The diagnostic criteria for Fahr's syndrome are as follows: (1) bilateral calcification of the basal ganglia on neuroimaging; (2) progressive neurologic dysfunction; (3) onset between the ages of fourth or fifth decades; (4) absence of biochemical abnormalities and somatic states suggestive of metabolic or mitochondrial disease; (5) absence of infectious, toxic or traumatic causes; and (6) a family history consistent with autosomal dominant inheritance [14, 15]. If a patient has a negative or unknown family history, all other criteria must be fulfilled. In the presented case, the patient had typical brain CT findings that met nearly all diagnostic criteria, except for the sudden onset of symptoms. Although acute-onset transient symptoms are uncommon in Fahr disease, a transient ischemic attack (TIA) associated with Fahr disease has been reported, with symptoms improving within 24 h of presenting at the ED for dysarthria [16]. A correlation between calcification and decreased cerebral blood flow has been demonstrated in up to one-third of patients with Fahr syndrome, and in these patients, circulatory disturbances have been reported to be present in the form of TIA or stroke [14]. Therefore, patients with typical brain CT findings and clinical features can be considered to have Fahr disease, even if the onset of symptoms does not occur acutely. A detailed family history should also be taken, and examinations should be performed to determine potential secondary causes.

Non-contrast brain CT scan is the preferred modality for diagnosing Fahr disease because it can detect and localize the extent of intracranial calcification better than MRI. Symmetric calcification of the basal ganglia in brain CT scans is a common incidental finding in the elderly, occurring in 0.3 to 0.5% of the population. However, unlike Fahr disease, the calcification is generally confined to the globus pallidus and is not associated with clinical findings [3]. Brain MRI has been reported to underestimate most calcific foci and some undetectable foci because the MR signal for calcification can vary in conventional sequences, as the composition of calcium deposits can include calcium and other minerals such as zinc, magnesium, iron, and so on [17, 18].

There is no specific treatment for Fahr disease that can limit the progression of brain calcification, and treatment is usually symptomatic. The prognosis of patients with Fahr syndrome is variable and difficult to predict. A previous study reported no correlation between clinical severity or prognosis and the extent of brain calcification [19].

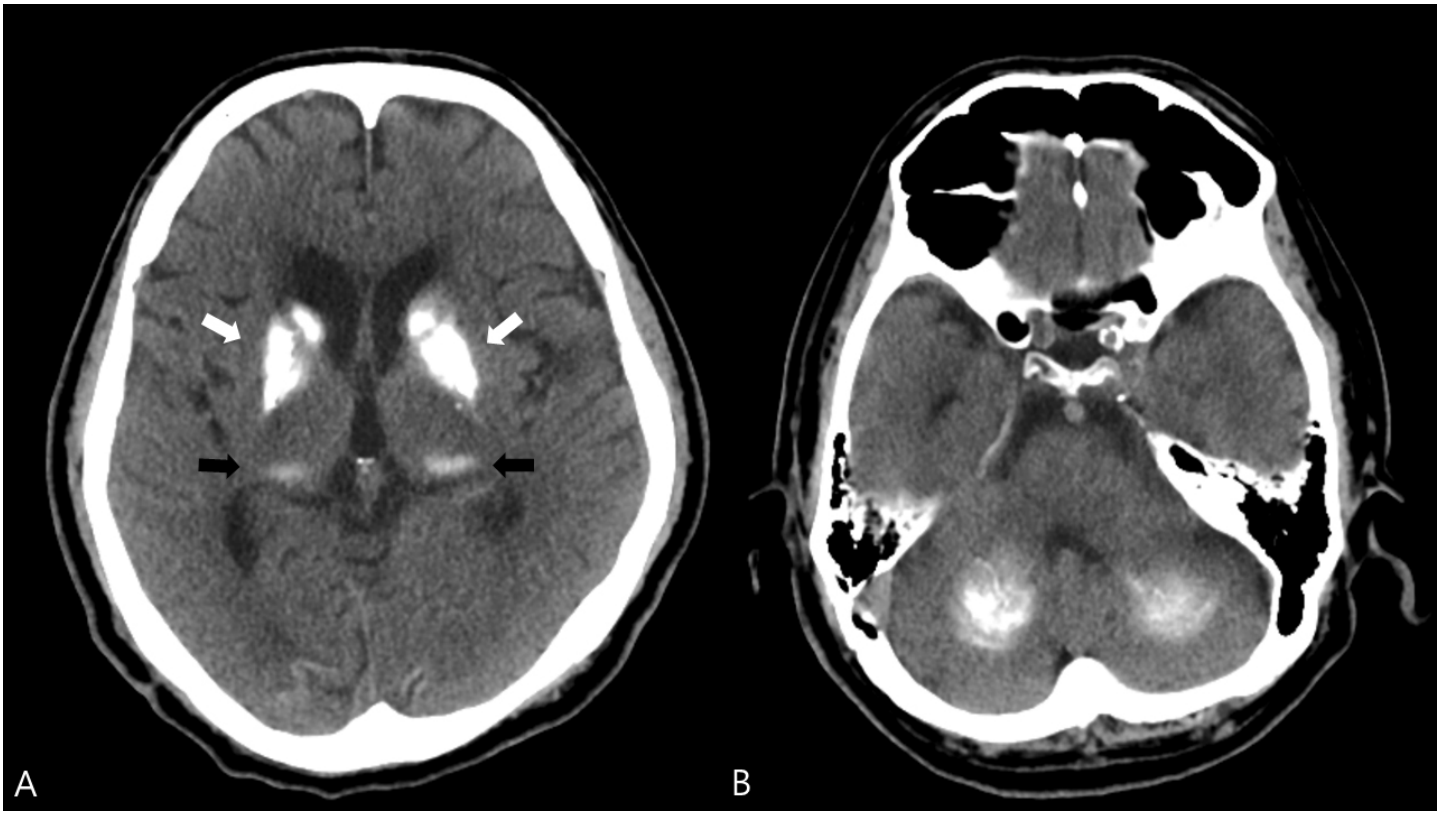

FIGURE 1. Brain CT scans showing symmetric calcifications. (A) Basal ganglia (white arrow) and thalamus (black arrow), and (B) dentate nucleus of the cerebellum.

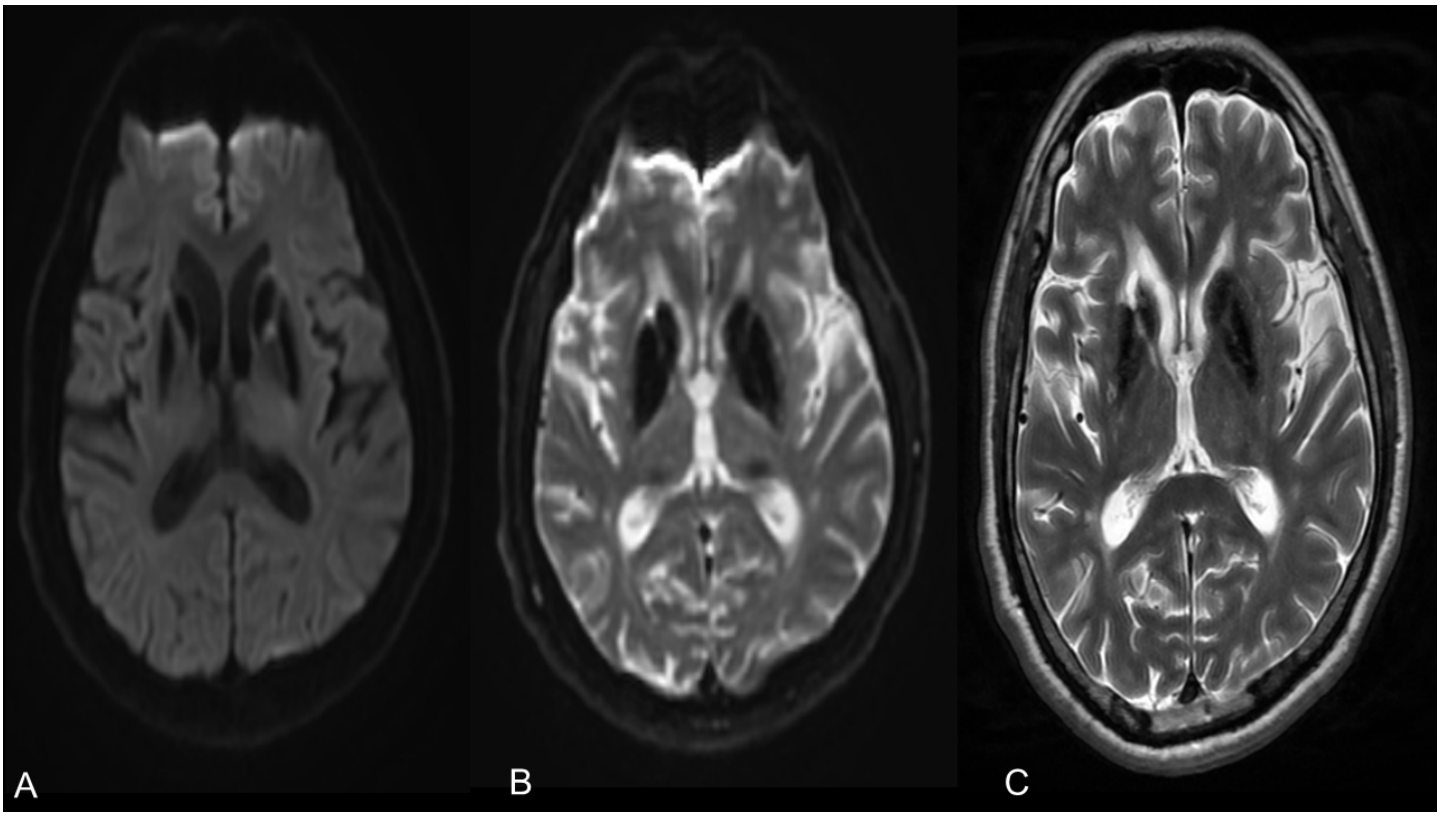

FIGURE 2. Brain MRI showing symmetric calcifications. (A) Diffusion-weighted image, and (B) apparent diffusion coefficient map with low signal intensity in both basal ganglia, but no restricted diffusion. (C) T2-weighted image with low signal intensity in both basal ganglia.

4. Conclusions

Despite the rarity of Fahr disease, our experience suggests that emergency physicians should consider Fahr disease if patients have focal neurological findings with characteristic symmetric calcifications in the affected area on brain CT. A detailed examination of other secondary causes associated with intracranial calcification should also be performed for the differential diagnosis of Fahr syndrome.

AVAILABILITY OF DATA AND MATERIALS

Not applicable.

AUTHOR CONTRIBUTIONS

JYA—examined and diagnosed the patient. HJ and JYA—drafted the manuscript. JWC, SM, and HWR—reviewed and revised the manuscript and supervised the study. All authors have read and approved the final manuscript.

ETHICS APPROVAL AND CONSENT TO PARTICIPATE

Informed consent was obtained to have the history published in the medical literature. The manuscript does not include any information indicating the identity of the patient. This study was reviewed and approved by the institutional review board of Kyungpook National University Hospital (202206005).

ACKNOWLEDGMENT

Not applicable.

FUNDING

This research received no external funding.

CONFLICT OF INTEREST

The authors declare no conflict of interest.

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How to cite this article: Haewon Jung, Jae Yun Ahn, Jae Wan Cho, Sungbae Moon, Hyun Wook Ryoo. A case of Fahr disease with transient slurred speech and abnormal movement of the left arm. Signa Vitae. 2023; 19(4): 213-216. doi: 10.22514/sv.2023.066.