Radiology Case Reports 17 (2022) 3139–3141

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Case Report

Fahr's disease with an atypical onset of epileptic seizure ☆,☆☆

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ARTICLE INFO

Article history:
Received 8 May 2022
Revised 30 May 2022
Accepted 1 June 2022

Keywords:
Fahr's disease
Brain calcinosis
Basal ganglia
Seizures

ABSTRACT

Fahr's disease is a rare neurodegenerative disorder first described by Karl Theodor in 1930, defined by abnormal calcified deposits in the basal ganglia and cerebral cortex. Fahr's disease commonly affects young to middle-aged adults with various clinical presentations, including endocrinologic, dermatologic, and neurologic problems, with extrapyramidal symptoms being the most common manifestation. In this case report, we present a case of an epileptic seizure as the first manifestation of Fahr's disease in a 45-year-old male.

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Introduction

Fahr's disease, also known as idiopathic basal ganglia calcification, bilateral strio-pallido-dentate calcinosis [1], and primary familial brain calcification [2], is a rare neurodegenerative disorder identified by abnormal calcium deposits. Calcium deposits are formed of calcium carbonate and calcium phosphate, commonly deposited in the basal ganglia, thalamus, hippocampus, cerebral cortex, cerebellar subcortical white matter, and dentate nucleus [2]. As per studies, there is no specific etiological agent for Fahr's disease. However, it was reported to have associations with several conditions like endocrine disorders, mitochondrial myopathies, dermatological abnormalities, and infectious diseases [3]. There are various presenting symptoms of Fahr's disease, with the most common being the extrapyramidal symptoms, including movement disorders, dystonia, athetosis, and dysarthria [2,3]. Seizures are seldom found as a presenting symptom, which makes this case rare [4]. In this report, we present a case of a 45-year-old male with epileptic seizure as the first manifestation of Fahr's disease.

Case report

A 45-year-old man was brought to the emergency department by his wife after witnessing an episode of abnormal

☆ Funding: None.
☆☆ Competing Interests: All authors declare no conflict of interest.
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https://doi.org/10.1016/j.radcr.2022.06.003
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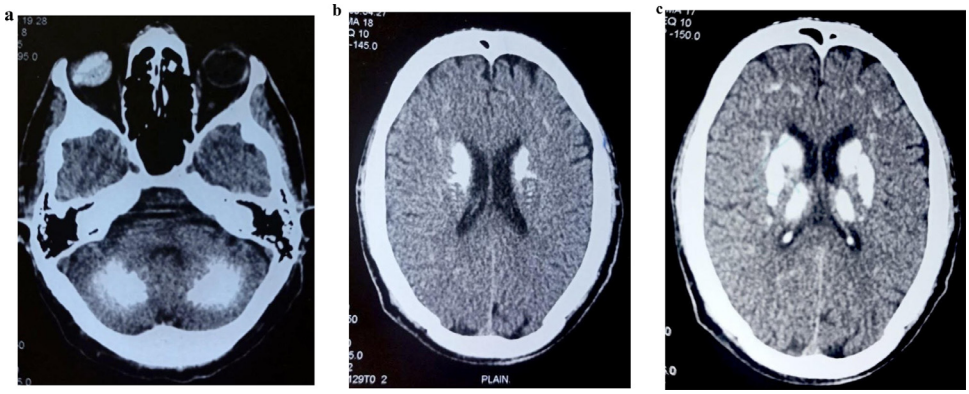

Fig. 1 – (A) Axial noncontrast computerized tomographic (CT) image depicts calcification in the right globe (right globe with a white haze). (B) Nonenhanced computerized tomographic (CT) scan of the brain demonstrating bilateral symmetrical calcifications in the basal ganglia. (C) Axial noncontrast computerized tomographic (CT) scan of the brain shows extensive bilateral symmetrical calcification in the head of caudate nuclei, putamen, globus pallidus, thalami, cerebellar dentate nuclei, centrum semiovale, and subcortical white matter.

whole-body movements. His wife reported that the patient was watching television and suddenly experienced blurred vision and loss of consciousness with sudden jerky movements of his upper and lower limbs. It lasted for 2 minutes, and after that, the patient was tired and sleepy. She also mentioned that the patient had a mild fever for the last 3 days and witnessed a similar episode of abnormal body movements 3 years back which was not evaluated. There was no history of headache, nausea, vomiting, weakness, slurred speech, or neck stiffness. Also, there was no relevant past medical, surgical, or family history. The patient was a nonsmoker, and he occasionally drinks alcohol.

On general examination, the patient appeared tired and confused. He was febrile with a temperature of 38.4°C, blood pressure of 120/80 mmHg, respiratory rate of 16 breaths per minute, heart rate of 80 beats per minute, and oxygen saturation of 97% in room air. He was alert and conscious, but not oriented to time and place. No dysarthria was noted. Neurological examination of the upper and lower limbs showed normal muscle tone and power. The reflexes were normal. There was normal gait and coordination, with no signs of meningeal irritation. Examination of other systems was unremarkable.

The initial laboratory investigations revealed no abnormalities. As there was a rise in the number of cases of dengue fever in that region, the NS1 antigen rapid test was requested, with the result being nonreactive. Subsequent investigations included a lumbar puncture that revealed normal biochemistry and cytology. A computed tomography (CT) scan and magnetic resonance imaging (MRI) of the head was performed and they revealed bilateral symmetrical calcification of the head of caudate nuclei, putamen, globus pallidus, thalami, cerebellar dentate nuclei, centrum semiovale, and subcortical white matter (Figs. 1 and 2). There were no ischemic or hemorrhagic changes in the brain imaging. All secondary causes of calcification were ruled out. The results gathered were suggestive of Fahr's disease. He was discharged on oral carbamazepine therapy and was followed up for 3 months postdischarge with no further reported seizure episodes.

Discussion

Fahr's disease is a neurological condition that occurs as familial or sporadic, inherited mainly in an autosomal dominant pattern [5], and rarely as an autosomal recessive pattern. Several studies have shown that it affects the genetic loci SLC20A2, PDGFB, and PDGFB, and a locus at 14q (IBGC1) is commonly involved [3,6]. A second locus has been identified on chromosome 8 and a third on chromosome 2 as well [3]. The term Fahr's disease refers to cases of idiopathic basal ganglia calcification in the literature [7]. There is no clear evidence for the occurrence of calcification in patients with Fahr's disease. As per studies, the potential causes of calcification are metastatic deposition, secondary to local disruption of the blood-brain barrier, or disorder of neuronal calcium metabolism [7].

In the literature, Fahr's disease has got various neuropsychiatric manifestations, including memory disturbance, hallucination, delusions, personality change, depression, motor, and phonic tics, stereotyped behaviors, and extrapyramidal signs, such as Parkinsonism and paroxysmal nonkinesigenic dyskinesia [7,8]. As such, recognition of Fahr's disease is significant to be able to distinguish it from other movement disorders. Seizures are one of the rarest symptoms that can reveal a Fahr's disease. In the literature, Aldawsari et al. have reported a case of Fahr's disease with a rare presentation of generalized tonic-clonic seizure as the primary manifestation in a 35-year-old female [8]. On further evaluation, the CT brain demonstrated bilateral symmetrical calcification of the basal ganglia with no other intracranial pathology confirming the diagnosis of Fahr's disease [8]. Mookerjee et al. have documented a case of Fahr's disease in an 18-year-old girl with a history of hypocalcemic seizure disorder. She was diagnosed with a hypocalcemic seizure disorder at 14 years of age, which was refractory to treatment with phenytoin, phenobarbital, and valproate with recurring episodes of seizures [9]. Later, on detailed evaluation, CT brain revealed bilateral

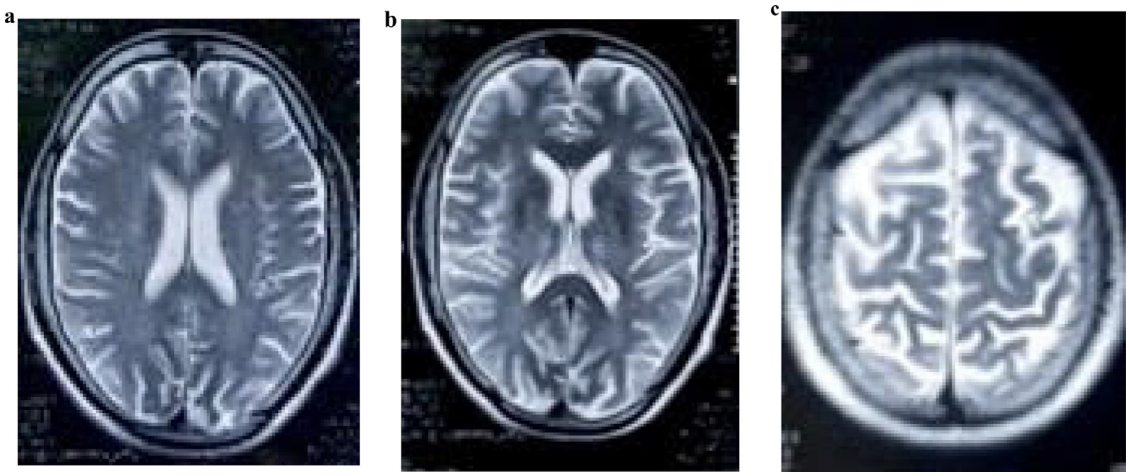

Fig. 2 – (A) Axial T2 magnetic resonance imaging (MRI) scan of the brain reveals altered signal intensity in the bilateral basal ganglia, appearing hypointense in the basal ganglia region. (B) Axial T2 magnetic resonance imaging (MRI) scan of the brain reveals altered signal intensity in the bilateral basal ganglia appearing hypointense in the external capsule and hyperintense in thalami. (C) Axial T2 magnetic resonance imaging (MRI) scan of the brain reveals hypointensity in the subcortical white matter.

symmetrical calcification of the caudate nucleus, lenticular nucleus (putamen+globus pallidus), thalamus, and the dentate nucleus of the cerebellum, validating the diagnosis as striatopallidodentate calcinosis (or Fahr's disease) secondary to hypocalcemia due to pseudohypoparathyroidism type Ib [9].

Diagnostic approaches to Fahr's disease include non-contrast computed tomography (CT) scan, magnetic resonance imaging (MRI), specific blood tests to exclude other potential causes of calcifications, and molecular genetic testing [1,4]. Evidence suggests a CT scan as the most preferred method of localizing and assessing the extent of cerebral calcification [3]. Pharmacological treatment is the usual mode of management in this condition with antiepileptic, antiparkinsonian, antipsychotic, and mood stabilizer drugs to address the presenting symptoms [1,7,8]. Overall, the treatment strategy shall be centered on the symptomatic treatment of patients and their quality of life. Regardless of its rarity, Fahr's disease should be included in the differential diagnosis of adult patients with first-time seizures, especially in the background of neuropsychiatric disorders. As it can mimic other movement disorders, proper recognition must be given to address the problem, especially in patients with other neurological conditions that can complicate the identification of the disease.

Patient consent

The case information in this manuscript has been provided with informed consent from the patient presented.

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