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Fahr syndrome presenting with epileptic seizure: A rare case report

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Abstract

Fahr's syndrome is a rare neurodegenerative disorder characterized by abnormal symmetrical calcifications of the basal ganglia, thalami, subcortical hemispheric white matter and deep cerebellar nuclei. The most common clinical presentations are movement disorders, pyramidal signs, parkinsonism, gait disorders, cerebellar anormalities, psychotic symptoms and cognitive disorders. This article discusses rare presentation of Fahr's syndrome with epileptic seizure.

Keywords: Fahr syndrome; Epileptic seizure; Bilateral basal ganglia calcifications; Fahr disease; Hypocalcemia.

Abbreviations: CT: Computed Tomography; EEG: Electroencephalograph.

Introduction

Fahr syndrome was first described by Karl Theodor Fahr in 1930 as a rare familial autosomal dominant disease with idiopathic basal ganglia calcification [1]. Fahr's syndrome is a neuropsychiatric syndrome characterized by symmetrical and bilateral intracerebral calcifications located in the basal ganglia, thalami, subcortical hemispheric white matte and deep cerebellar nuclei and usually associated with a phosphorus and calcium metabolism disorder. It may present clinically with movement disorder, cognitive decline, cerebellar signs and behavioral disorders [2]. Although etiology is not certain; it may be associated with neurological disorders and metabolic disorders A certain degree of calcification of basal ganglia can be considered physiological with aging, over 50 years, and it could be an incidental finding in 15-20% of asymptomatic patients undergoing computed tomography (CT) scan [3,4].

Case report

A 28-year-old male patient was admitted to our emergency department with a generalized tonic-clonic seizure that lasted for 3 minutes and passed spontaneously. After the patient had generalized seizures 3 more times without regaining consciousness, it was considered as status epilepticus and intravenous loading of 3000 mg/day levetiracetam was performed. After his antiepileptic loading, the seizure did not recur, and he gradually regained consciousness. Neurological examination was normal. Non-contrast brain tomography (CT) imaging revealed calcifications in both basal ganglia, more prominent in bilateral caudate nuclei and lentiform nuclei (Figure 1). Electroencephalography (EEG) examination was normal. Laboratory investigations revealed a hypocalcaemia level of 8.2 mg/dl (normal:8.4-10.5 mg/dl) and serum phosphorus level of 3.53 mg/dl (normal:2.6-4.5 mg/dl), although a serum parathyroid level was not evaluated. The serum electrolytes levels and thyroid function tests were normal. 25 Hydroxy-Vitamin D3 level 5.09 µg/L (N: 10-30 µg) /L) was measured. The patient was started on oral vitamin D and calcium therapy. The patient, who did not have seizures again in the follow-ups, was discharged with levetiracetam, oral calcium and vitamin D treatments.

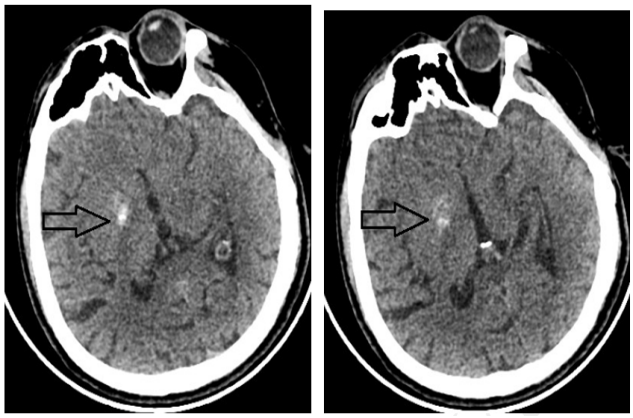

Figure 1: Calcifications in both basal ganglia, more prominent in bilateral caudate nuclei and lentiform nuclei (black arrow).

Discussion

Fahr syndrome is a condition with bilateral symmetrical calcifications in the basal ganglia as a result of calcium metabolism disorder, which has been shown to occur due to many different causes in the literature, and various causes have been reported, including hypoparathyroidism or pseudohypoparathyroidism, as well as genetic, developmental, metabolic, infectious, sporadic and other conditions [5].

A variety of neurological signs and symptoms are associated with Fahr's syndrome. Seizures and confusion may occur. Tetany is present, but it is difficult to distinguish from occasional myoclonus of epileptic disorder. Also, spasticity, gait disorder, speech impairment, dementia, parkinsonism, chorea, tremors, dystonia, myoclonus and coma are present. Neurological manifestations may be confined to one side of the body despite the presence of bilateral calcifications. Neuropsychiatric symptoms range from mild difficulty with concentration and memory to changes in personality or behaviors to psychosis and dementia [6].

Calcium/phosphorus abnormalities, idiopathic or secondary hypoparathyroidism, infections (brucellosis, Acquired Immune Deficiency Syndrome, toxoplasmosis, rubella, cytomegalovirus, herpes simplex virus), toxic exposure (lead, carbon monoxide), vasculitis, pseudohypoparathyroidism, Cockayne syndrome I and II, Aicardi-Goutieres syndrome, mitochondrial diseases, Coat's syndrome and neuroferritinopathy can cause bilateral basal ganglia calcification [4]. Histopathological studies showed that calcium is the major element present and it accounts for the radiological appearance of the disease, together with the involvement of several minerals like iron, magnesium, aluminum, and zinc [7].

Regarding lesions' distribution pattern and qualitative aspects, calcifications are typically bilateral and symmetrical, most frequently located in the basal ganglia, but also in dentate nuclei, thalamus, brain stem, centrum semiovale, and subcortical white matter [8]. Pathological lesions can be easily distinguished from age-related physiological calcifications of basal ganglia for their features: the latter are often small and faint, symmetrical and confined into the globus pallidus, whereas the first are more diffuse and extensive, involving also the putamen and the dentate nucleus, and usually show a coarse conglomerate pattern [9].

Conclusion

Basal ganglia calcification is a rare neurodegenerative disorder occurring as primary/idiopathic disease or as secondary manifestation of a condition, leading to an abnormal deposition of calcium in peculiar brain structures. Clinically, it can be difficult to diagnose because of its wide neuropsychiatric spectrum. Diagnosis may be more difficult since epileptic seizure presentation is not common. When the diagnosis is suspected together with clinical and neuroimaging, blood calcium, phosphorus, manganese, parathormone and thyroid function tests should be studied. Vitamin D levels should be measured. In case of doubt, blood and urine tests can be sent to rule out toxicity. Cerebrospinal fluid can be studied to rule out an infectious or autoimmune cause. There is no specific treatment for Fahr Syndrome. Treatment is determined by the underlying cause. Symptomatic treatment is most helpful. The aim is to replace the deficient electrolytes or to eliminate the underlying cause [10].

Declarations

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Declaration of competing interest: The authors declare that they have no known competing financial or personal relationships that could have appeared to influence to work reported in this paper.

Consent: As per international standard or university standard, patient's written consent has been collected and preserved by the author(s).

Ethical approval: As per international standard or university standard written ethical approval has been collected and preserved by the author(s).

Competing interests: Authors have declared that no competing interests exist.

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