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Case Report

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Fahr's Syndrome

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ABSTRACT

Fahr's syndrome is a rare neurodegenerative disorder. Bilateral basal ganglia and dentate nuclei of the cerebellum are involved in this disorder and deposition of calcium is the hallmark of this syndrome. It has been recognized as a sporadic or inherited disease with variable presentations. In this article, we report a 40-year-old man with incidentally discovered brain calcification as the sole manifestation of Fahr's syndrome. A 40-year-old male without any comorbidities was presented with brain calcification that was found incidentally. Brain imaging revealed symmetric calcifications in bilateral basal ganglia, internal capsules, and cerebral white matter. This pattern of calcification is highly suspicious of Fahr's syndrome. Other pathologic processes that could lead to intracranial calcification were excluded. We present a young patient with sporadic and asymptomatic Fahr's syndrome after ruling out abnormalities of known calcium metabolism and developmental defects.

Introduction

Calcification of the basal ganglia has many causes. It is an incidental finding in up to 1% of all CT brain scans. Basal ganglia calcifications can also be seen in infectious, metabolic, and genetic disorders affecting this brain region [1].

Fahr's disease (familial idiopathic calcification of the basal ganglia) is a rare (prevalence less than 1 in 1000000 people) neurodegenerative disease characterized by symmetrical bilateral calcifications in the basal

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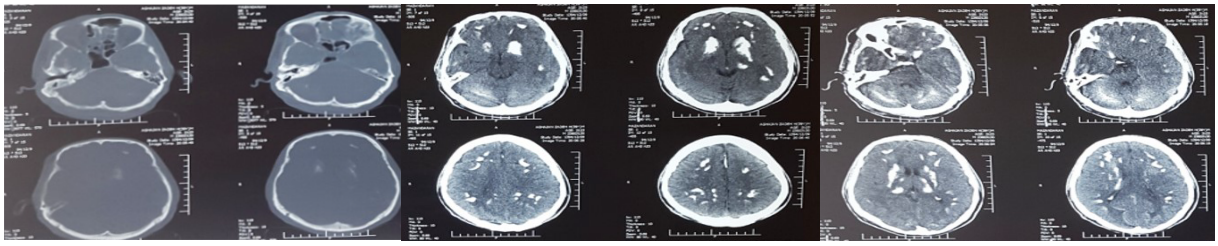

Figure 1. Sparse subcortical calcification of brain

ganglia and some other brain structures such as dentate nucleus, thalamus, cerebral cortex, subcortical white matter, and hippocampus [2, 3]. It has been recognized as a sporadic or inherited disease with identified loci in chromosomes 2, 8 and 14q and autosomal dominant type of inheritance [3-5]. In this article, we report a 40-year-old man with incidentally discovered brain calcifications as the sole manifestation of Fahr's syndrome.

Case Presentation

A 40-year-old man was first looked up a surgeon for excision of a subcutaneous mass. His mass was in right temporal region of head and seems to be lipoma which was approved by pathology after total excision. However, in preoperative evaluation, multiple calcifications of brain were observed as an incidental finding (Figure 1). Then, he was referred to Neurology Clinic of Rohani Hospital in Babol, Iran, in 2016 for further evaluation, definite diagnosis, and proper treatment. There were no history of symptoms or remarkable comorbidities. There was not any developmental impairment in his previous childhood history. No evidence of similar problem was reported in his family.

He reported no high risk behaviors and no history of travels abroad. It should be noted that he was not addicted without history of smoking. In examination, scar of mentioned excision was evident in right temporal region. All other parts of the examination were within normal limits. Precise neurological examination was done and no abnormality was found. According to previous imaging findings and to rule out differential diagnosis (Table 1), further paraclinical evaluations were done.

The results of following tests were within reference values: full blood count, differential blood count, erythrocyte sedimentation rate, blood glucose, cholesterol, total protein, albumin, urea, creatinine, total and direct bilirubin, electrolytes, troponin, cerebrospinal fluid test, LFTs. Serology tests for HIV, toxoplasmosis, CMV and syphilis were all reported as negative. Markers of autoimmunity were also negative. His N-TACT PTH II result was 35 pg/mL (NL Range 11.7-61.1). X-ray of the lungs showed no active pulmonary disease. For evaluation of intra-cranial vascular lesion, MRI (Magnetic Resonance Imaging) (FLAIR, with contrast) and MRV (Magnetic Resonance Venography) and MRA (Magnetic Resonance Angiography) were taken (Figures 2).

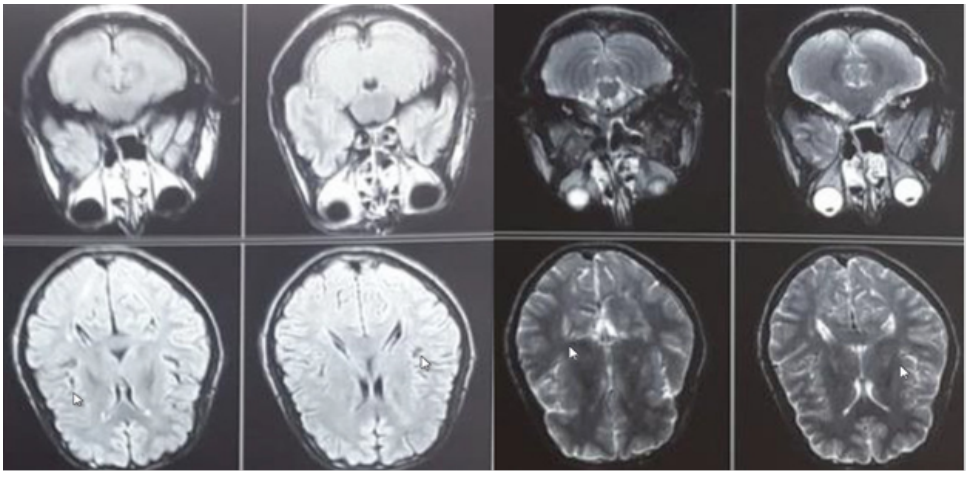

Figure 2. Foci of signals in centrum semiovale, both internal capsules and lentiform nucleus (MRI)

Table 1. Differential diagnosis of brain calcification [3]

Causes		Structures Involved
Intra-axial calcification	Structures involved	Basal ganglia Cerebellum
	Causes: Neoplasms	Astrocytomas Oligodendrogliomas Metastatic tumors Medulloblastomas Other primary brain tumors
	Endocrine/metabolic	Diabetes mellitus Hypoparathyroidism Pseudohypoparathyroidism Hyperparathyroidism
	Infectious	Congenital childhood infectious, particularly TORCH Tuberculosis Parasitic infections such as neurocysticercosis and cerebral hydatid cyst disease
	Congenital	Sturge-Weber syndrome Tuberous sclerosis Lipomas Neurofibromatosis
	Vascular	Dystrophic calcification in chronic infarction Aneurysms Angiomatous malformations Arteriovenous malformations Chronic vasculitis
	Idiopathic genetic	Familial idiopathic basal ganglia calcification
Extra-axial calcification	Structures involved	Flax Cerebri The pineal gland Dura and arachnoid Tentorium cerebelli Superior sagittal sinus Petroclinoid and interclinoid ligaments Choroid plexus Habenula Arachnoid granulation
	Causes	Meningiomas Dural osteomas Calcifying tumors Exaggerated physiological calcifications

According to varied features of Fahr's syndrome, diagnosis in this patient was established by obtaining MRI scan of the head and ruling out abnormalities of known calcium metabolism and developmental defects.

Discussion

Fahr's syndrome or idiopathic basal ganglia calcification is a rare neurologic disorder with variable clinical presentations and distinctive neuroradiological features. Fahr's syndrome manifests as autosomal dominant, familial and sporadic forms [6]. According to a registry of Fahr's syndrome, symptomatic individuals account for 67% of the total cases [6, 7]. Of the symptomatic cases, the incidence among men was higher compared to women and movement disorder was the most common manifestation [8]. The common sites of calcifications in Fahr's syndrome are globus pallidus, putamen, caudate nucleus, internal capsule, dentate nucleus, thalamus, and cerebral white matter [9].

Differential diagnosis of pathologic basal ganglia calcification are presented in detail in Table 1. Detailed history taking and laboratory investigations are helpful in determining the etiology of pathologic intracranial calcification. As for our patient, he had been well without developmental anomaly, infection signs or toxin exposure. He had no systemic disease, metabolic disorder or hypoxia history. His thorough laboratory studies excluded the presence of other pathologic processes. Brain CT, supplemented by MRI study exhibited symmetric calcifications in the bilateral basal ganglia, thalami, bilateral cerebral subcortical white matter cerebellar dentate nuclei and deep cerebellar white matter, which were all consistent with Fahr's syndrome.

The patient denied previous familial illness. Therefore, we considered him a sporadic case of Fahr's syndrome. The most frequent initial symptoms are associated with extrapyramidal system disorders [2], including Parkinson syndrome [3], choreoathetosis [10] and dystonia [11]. Other symptoms include coordination impairment [2, 3], dysarthria [2, 3], psychiatric disorders (depression, anxiety, visual, auditory hallucinations, delusions, mania, personality and behavior problems, schizophreniform psychoses, delirium) [2-4, 9, 12, 13] and cognitive impairment as a part of subcortical dementia (impaired verbal, visual-spatial memory, planning, attention, concentration, visual constructive abilities) [14, 15].

Epileptic seizures (complex partial seizures) [3, 16], stroke-like incidents [3], vertigo [3], headache [3], paresis [3], orthostatic hypotension [2, 3] have been rarely described. There is no specific treatment for Fahr's syndrome to limit the progression of brain calcification. Treatment is usually symptomatic. In some preliminary studies, disodium etidronate, a bisphosphonate, exhibits functional benefits and symptomatic improvement without reduction in the amount of calcifications [17, 18]. His MRV/MRA findings including narrowing of anterior communicating artery (MRA) and narrowing of right transverse and sigmoid sinus (MRV) also were attributed to congenital hypoplasia which was a normal variability.

Ethical Considerations

Compliance with ethical guidelines

All ethical principles were considered in this article. The participant was informed about the purpose of the research and its implementation stages.

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Conflict of interest

The authors declare that there is no conflict of interest regarding the publication of this paper. All authors contributed in the management of the case.

The authors contributions is as follows: Tahereh Hejazian collected case's data. Reza Mohseni reviewed the notes and images and wrote the manuscript. Payam Saadat reviewed the literature and did the final edit. All authors read and approved the final manuscript.

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