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Case Report

A case report of Fahr's disease presenting with cerebellar ataxia

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Abstract

Fahr's disease is a rare condition characterized by the symmetrical calcification of basal ganglia and dentate nuclei of the cerebellum calcification. It is usually manifested by involuntary movements and psychiatric symptoms. Ataxia has been reported as an uncommon manifestation of Fahr's disease. Herein we introduce a 41-year-old man presenting with progressive gait disturbance who was finally diagnosed with Fahr's disease.

Introduction

Idiopathic basal ganglia calcification or Fahr's syndrome is a rare neurological disease characterized with calcification in the basal ganglia and cerebellum. For the first time, it was reported by Karl Theodor Fahr in 1930.[1] The term, Fahr's disease, is applied to primary familial brain calcification, while the term, Fahr's syndrome, is used when it is a result of various secondary causes.[2] Its prevalence is less than 1/1 000 000 and the symptoms usually begin between the ages 40 and 60.[3-5] About 40% of patients with Fahr's disease manifest initially with psychotic features. Other symptoms include cognitive impairment, seizure, extrapyramidal, and cerebellar symptoms.[3,6] Here, we report a 41 years old man with progressive gait disturbance and symmetrical calcification involving bilateral dentate nuclei.

Case Report

A 41-year-old man was presented to the neurology clinic with the chief complaint of spasms in the legs and fatigue during past three years, but had not bothered enough to seek any medical consultation. In the past year, he had been experiencing the similar symptoms in the upper limbs and became unable to coordinate the movements of his hands. More recently, he gradually developed a dull sensation in legs, unsteady gait, slurred speech, and bladder control loss. The recent events had profoundly affected his quality of life, to the extent of becoming dependent on external help for daily chores. His past and family medical history was unremarkable. He had no history of using tobacco, alcohol or any other illegal substances. There was no history of neuropsychiatric problems. He was a high school graduate, working as a salesman prior to the current presentation. He had neither sustained head trauma nor exposed to any toxic chemical. His physical examination was significant for overt spasticity of four limbs with mild diminished muscle force, hyperactive deep tendon reflexes, ataxic gate, positive Romberg test, and dysarthria. He was conscious with normal mental status examination. Cranial nerves were normal. No tremor, rigidity and abnormal movements were detected.

Routine blood tests including renal and liver function tests were all within normal ranges. Laboratory investigations consisting of serum calcium, magnesium, phosphate, parathyroid hormone (PTH), calcitonin, 25-hydroxy vitamin D3, iron, and urinary calcium excretion were also normal. Results of serologic investigations for cytomegalovirus, toxoplasmosis and brucellosis were negative. Brain magnetic resonance imaging (MRI) study revealed the calcification of bilateral basal ganglia, thalamus, and cerebellar hemispheres which were the indicative of Fahr's disease (Figures 1A and 1B). The patient declined genetic testing due to the financial constraints. Unfortunately, in the absence of any curative medication, our patient was solely started on symptomatic treatment with baclofen.

Discussion

Fahr's disease is a rare neurodegenerative syndrome and very little known regarding to its etiopathogenesis. The strio-pallido-dentate calcinosis is referred to as "Fahr's syndrome" if it occurs secondary to an infectious agent (e.g., *Toxoplasma*, *Brucella*, CMV) or a pre-existing endocrinopathy (i.e., impaired parathyroid

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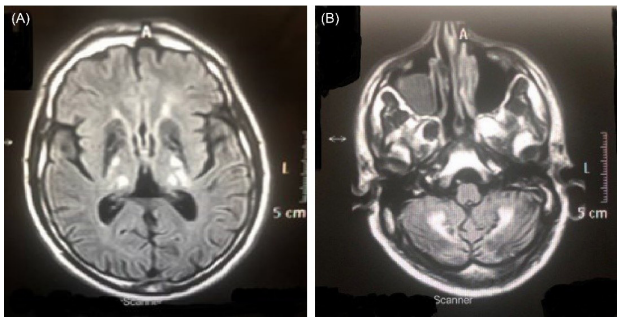

Figure 1. MRI showing bilateral symmetrical calcification in the basal ganglia (A), cerebellum (B)

function), mitochondrial myopathy, iron accumulatory disorder, and a long list of exceptionally rare congenital syndromes.[5,7,8] If the calcification is not explained by any associated condition, it will be termed as "idiopathic" calcification of basal ganglia. Among several mutations of genes arguably contributing to the Fahr's pathogenesis, IBGC1 (idiopathic bilateral basal ganglia calcification1) gene locus on chromosome 14q is the best known for its linkage to disease.[9] It is generally inherited in an autosomal dominant or recessive manner. Although without proper genetic evaluation, a recessive transmission can by no means be excluded, given the absence of any affected family members our patient is speculated to be a sporadic case. In Fahr's syndrome, since the brain calcification may involve various brain areas; it can manifest with various neuropsychological signs. It usually presents with movement disorder such as Parkinsonian symptoms. Chorea and athetosis are among the other common symptoms, while cognitive dysfunction, seizure and cerebellar symptoms are less common findings.[10] The exact pathogenesis of Fahr's disease is unclear. In the pathologic studies, the deposits of calcium and other minerals were observed in extracellular space as well as in the walls of capillaries. Electron microscopic studies, however, showed calcium deposits in the cytoplasmic processes of glial cells.[11,12]

Depositions of calcium, magnesium, iron and mucopolysaccharides are found in the microvasculature of affected areas, the most common of them being globus pallidus.[6] While calcifications can be detected with plain radiography, CT scan study is utilized to determine their exact localization and extent. Likewise, calcinosis of basal ganglia is viewed as high signal areas in T1 MRI images.

In the absence of any curative treatment, the aim of treatment is symptomatic therapy. In patients with Parkinson's symptoms, levodopa has poor results. For psychiatric symptoms, antipsychotic drugs are prescribed.[13]

Conclusion

We reported a case of Fahr's disease with calcification of the dentate nucleus and basal ganglia in a patient with gait disturbance as the main presenting symptom. Although rare, Fahr's disease should be suspected if there is a symmetrical calcification in basal ganglia and cerebellum. Depending on the site of involvement, it can present with various neurologic symptoms, especially movement disorders.

Authors' Contribution

Conceptualization: Mahnaz Talebi, Mohammad Yazdchi.
Formal Analysis: Zahra Jalali.
Investigation: Nasrin Forghani, Ramin Khanalizadeh.
Methodology: Nasrin Forghani, Ramin Khanalizadeh.
Writing – original draft: Mahnaz Talebi.
Writing – review & editing: Mahnaz Talebi.

Competing Interests

The authors have no conflicts of interest to declare.

Ethical Approval

This study was performed in line with the principles of the Declaration of Helsinki. The patient has provided an informed consent to publish this case report.

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