Neurological Case Reports

Case Report
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Unique Neuropsychiatric Presentation of a 43-year-old with Fahr's Disease

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Abstract

Introduction: Fahr's disease is a rare neurodegenerative disorder characterized by idiopathic calcifications within the basal ganglia and cerebral cortex. Although traditionally associated with movement disorders, the psychiatric manifestations of Fahr's disease are not well-documented. This case report presents a unique instance of a patient with predominant psychiatric symptoms prior to the onset of motor abnormalities.

Case Presentation: We report the case of a 43-year-old female with no significant medical history, who presented initially with delusions and hallucinations, leading to her admission under the Baker Act in 2019. Imaging at that time revealed basal ganglia calcifications, which were dismissed due to the lack of significant clinical symptoms. Four years later, she presented again with exacerbated psychiatric symptoms and new-onset motor abnormalities. Repeat imaging showed progression of the calcifications, including new involvement of the cerebral cortex. Despite treatment with antipsychotics and therapy, her symptoms only partially improved, highlighting the progressive nature of the disease.

Discussion: This case underscores the importance of considering Fahr's disease in the differential diagnosis of psychiatric symptoms, particularly when imaging reveals brain calcifications. The case is notable as it represents a rare documentation of concomitant psychiatric and motor symptoms in Fahr's disease, emphasizing the need for awareness and further research into its pathophysiology and treatment.

Introduction

Fahr's disease, sometimes known as "Fahr's syndrome, named after Karl Theodor Fahr, otherwise known as Bilateral Striopallidodentate Calcinosis (BSPDC), is a rare neurodegenerative disorder in which idiopathic calcifications are found deposited in the basal ganglia and cerebral cortex [1]. These deposits can also be found in the thalamus, hippocampus, cerebellar subcortical white matter, and dentate nucleus. The depositions are typically made of calcium carbonate and calcium phosphate [1]. It has been documented that some patients suffer from familial autosomal dominant mutations in the genes SLC20A2, PDGFRB, PDGFB, and XPR [1,2]. These depositions are different from physiological depositions that can be identified in individuals later in life (typically sixties to seventies) as part of the aging process. While the origin of these deposits cannot always be determined, every patient should be considered individually as many different neurological disorders can present with similar symptoms. Patients with Fahr's disease usually present in their forties to fifties with neurological or psychiatric symptomology and diagnosis is confirmed with brain MRI. Ruling out other neurodegenerative conditions is necessary to make the diagnosis. Current literature on Fahr disease primarily presents patients with purely neurological presentations including movement changes such as Parkinsonism, chorea, and tremors. Other features include ataxia, headache, seizure, and cognitive impairment [3]. Psychiatric symptoms are thought to be present in these patients but rarely identified. Psychiatric symptoms could include psychosis, neurocognitive symptoms, anxiety, and depression [4]. While several researchers have theorized on the psychiatric presentations of Fahr's disease there have been few reports in the clinical literature and even fewer with a purely psychiatric presentation. There are no clear clinical guidelines on how to treat Fahr's syndrome, thus successful management of the symptoms of Fahr's should be reported to identify treatment options for further study. We present a case of a 43-year-old female with no significant past medical history who presents with delusions, auditory and visual hallucinations, and motor symptoms with confirmed bilateral basal ganglia calcifications on MRI.

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She was admitted to our psychiatric hospital and over a few weeks treated with antipsychotic medication and group psychotherapy.

Case Report

The patient is a 43-year-old woman who first presented to our health system 4 years prior to the current admission. During this initial presentation, she was brought into the emergency department after being found in one of the school libraries saying that she was "trying to call the government" and that she was not safe. She was preoccupied regarding an unknown man harassing her and that no one believed her. She was transferred to the psychiatric hospital where she was admitted. She initially refused medication, but frequently noted that she was "listening to the lord" and would carry her bible around the unit. An array of blood work including BMP, CBC, LFTs, salicylate levels, acetaminophen levels, ethanol levels, UDS, urine culture, pregnancy test, folate, B12, cholesterol, ceruloplasmin, and serum copper, and thyroid tests were all within normal ranges. At that time a CT and MRI were completed showing "prominent mineralization in the basal ganglia". While it was noted that this was abnormal for her age, at that time it was not investigated further as her only symptom at this point was delusions. The patient was placed on olanzapine, which was discontinued after 2.5 weeks due to noncompliance and lack of efficacy. After 33 days, she was deemed safe to discharge as she was not a danger to herself or others with these delusions.

One year later, the patient was admitted to the hospital after a bicycle accident. Nothing was noted in her chart at that time that discusses her mental state and at that point she was not on any medications. Post discharge, she had two in-office and one telemedicine appointment with orthopedics during which she was able to contribute fully to her care and no concerns were raised with her capacity.

Three years later, leading up to the current admission, the patient was again found in a school library. Library staff called the police as she appeared to be reacting to internal stimuli and would not leave the library with their prompting. When police arrived, the library staff said this was a recurring situation that had happened multiple times within the past few months, which has required police assistance on multiple occasions. The patient was noted to be tense, hyperverbal, and had poor insight into her condition. The patient was found to be disorganized in her thought process and behaviour. Additionally, she had persecutory and grandiose delusions. The police brought her to our psychiatric hospital. On admission, the patient argued with staff that the university has been using traffic noises to harass students for four years. Additionally, she was laughing inappropriately and frequently seen jerking and swatting at the air. Collateral information was obtained from a contact that the patient provided, who reported that the patient started calling him and texting him excessively after not hearing from her in four years. The contact had blocked her number and the patient started to email him 20-50 times daily. The emails were illogical and demonstrated paranoia with phrases such as "we are in danger, come to my place".

Upon initial assessment the patient exhibited persecutory delusions that put her at risk of self-neglect. She also displayed uncoordinated, bilateral jerking movements. These included jerking of her torso with twitching of her accessory muscles, sudden motions of her head to one side or the other, and sudden flails of her arms to the side. These presumably involuntary motions were not distractible. A baseline CBC, CMP, B12, vitamin D, folate, TSH, urinalysis, and EKG were found to be within normal ranges. Further workup including calcium, ceruloplasmin, lead level, lyme IgM, PTH, syphilis screen, NMDA receptor antibody, and ANA ruled out other conditions such as Wilson's disease, Anti-NMDA encephalitis, and lupus. A CT head was done and found bilateral basal ganglia calcifications similar to the finding in 2019 with newer calcifications noted in the cerebral cortex. Neurology consulted and described her movements as hyperkinetic and that the findings on her CT were not consistent with her age. They discussed the possibility of the movements being tics, however with the patient unable to give a clear history and the inability to reach her family, this couldn't be a formal diagnosis. The patient was administered diphenhydramine, haloperidol, lorazepam, and olanzapine. While these calmed her agitation and her delusions some, she continued to have limited insight into the reason for admission. Additionally, her movements neither improved or worsened. She tolerated the medication well over the course of two and a half weeks and ultimately was given haloperidol decanoate 50mg long acting injectable prior to discharge. By the day of discharge, her movements improved moderately with decreased jerking of her torso, diminished movements of her head from one side to another and decreased arm flailing but never completely resolved. The patient started to attend group therapy sessions without prompting and interacted with the other patients and staff appropriately. She was discharged with the plan to return to her family in China.

Discussion

There is limited research currently in the literature on the various possible psychiatric presentations of Fahr disease. It is thought that the more calcifications found on imaging the more prominent the psychiatric presentation can be in these patients [2]. One case report detailed a predominantly psychiatric presentation of a patient with Fahr's disease. In this case the patient experienced auditory hallucinations and delusions [4]. These symptoms were responsive to antipsychotic therapy. Another case report detailed a similar case in which the patient presented with auditory hallucinations [5]. However, this patient did exhibit some motor involvement in which she would have jerking of her upper extremities intermittently, which were not severe and not the true target of the treatment provided.

Ultimately, both case reports of patients with psychiatric symptoms were diagnosed with brain MRI and an otherwise normal workup. It is critical in these patients to rule out other etiolgies for these patient's symptoms as different disease processes can have the same presentation. Our patient had a more "classic" presentation with movement symptoms in addition to her psychosis. Further characterization of patient's presentations with Fahr disease and what treatment they are responsive to is critical for treatment of future patients.

While the etiology of Fahrs disease is not specific to any condition, it is critical to screen for endocrine disorders, mitochondrial myopathies, dermatological abnormalities, and infectious disease as there is associations with these conditions [1]. A case series and literature review completed in 2016 found that Fahr's disease is probably secondary to the dysfunction of cortico-basal connections and their interhemispheric relations6. This research did not find a significant correlation between calcifications and neurological symptoms, however did find that there is a larger amount of calcification in symptomatic patients compared to asymptomatic patients [6]. Fahr's disease is often associated with a disorder of

calcium and phosphate metabolism, particularly hypoparathyroidism [6]. Our patient had PTH and calcium levels within normal limits.

While treatment of the underlying cause for cases caused by hypoparathyroidism or mitochondrial encephalopathy does lead to improvement in symptoms, there are no treatments at the moment that inhibit the progression of calcification in the basal ganglia [6]. Studies show that improvement in psychosis with psychiatric treatment is variable, and at times completely ineffective [6]. In our case, the patient's psychotic and cognitive symptoms improved mildly throughout course of hospitalization. Interestingly, her neurological symptoms also decreased in frequency and intensity. This leads us to believe that some of her movements may have been voluntary, while others were neurological.

Conclusion

Overall, the pathophysiology behind calcium deposition in Fahr's Disease is complex, possibly multifactorial, and is not well understood. The disease often manifests with both psychiatric and neurological symptoms; however, one can precede the other. Treatment of the underlying condition, if there is one, can improve neuropsychiatric symptoms as demonstrated above. Treatment with antipsychotics has variable results in these cases. Further research is needed to better understand the pathological process and, in turn, treatment options.

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