Psychosis possibly associated with Fahr's disease: A case report from the Himalayan country

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Abstract

Fahr's disease (FD) is a progressive and degenerative familial autosomal dominant, rare neurological disorder, characterized by bilateral calcifications in the brain. FD may have neurological manifestations corresponding to the area of the brain involved. Significant proportions have neuro-psychiatric symptoms and psychosis is one. There is no specific treatment for FD but the neuropsychiatric manifestations can be managed. We present a case of a 30-year-old male with a 12-year history of psychosis, an episode of mania with psychotic symptoms, and an unsteady gait for 3 years. Baseline investigations were normal and MRI of the brain showed bilateral calcification involving significant brain areas suggestive of FD. He was managed with antipsychotics and antiepileptics. This case highlights the need for neuroimaging in cases of psychosis with neurological symptoms and puts forward the possibility of FD causing psychosis in this case.

Keywords
Fahr's disease, psychosis, basal ganglia calcification.

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INTRODUCTION

FD, also known as bilateral strio-pallido-dentate calcinosis, is a rare neurological disorder characterized by abnormal deposits of calcium in certain areas of the brain, particularly the basal ganglia and dentate nucleus. Calcifications are hypothesized to be due to lipid deposition and demyelination. These calcifications can be asymptomatic mostly but may cause a range of symptoms, including movement disorders, cognitive impairment, and psychiatric symptoms . [1-4] The usual age of onset is between 40 to 60 years, with the presence of genetic autosomal dominant or recessive trait.[5] The prevalence of FD is relatively low, 0.7-1.2% of routine cerebral computed tomography (CT)-scan making it a challenging diagnosis for clinicians.[6]

The exact mechanism by which FD induces psychosis is not fully understood. However, it is believed that the extensive calcification of the basal ganglia, may disrupt the normal functioning of the brain and lead to neuropsychiatric symptoms, including psychosis. In FD, psychotic presentation includes auditory hallucinations, complex visual hallucinations, paranoid delusions or fugue states with ideas of reference or influence and catatonia. [6]

In this case report, we present a detailed account of a patient diagnosed and treated, as a case of psychosis for 12 years, with an incidental diagnosis of FD. We aim to shed light on the clinical characteristics and challenges of diagnosis and management strategies associated with this rare neurological condition with neuro-psychiatric manifestation. This case report could have important implications for both clinical practice and future research, as there is a paucity of knowledge on FD due to its rarity, complexity in the treatment of FD, and the relationship between FD and psychosis.

CASE PRESENTATION

Mr. D, a 30-year-old divorced male, from western Nepal with a low socio-economic status, was educated up to high school with no known medical illness a well-adjusted premorbid personality and without a family history of psychiatric illness. The total duration of the illness was 12 years, with insidious onset, and continuous course with periodic exacerbations. The symptoms started with sleep disturbance and unprovoked anger, followed by self-muttering, delusion of reference and persecution. He was taken to a district hospital and prescribed olanzapine. There was partial improvement but he was frequently non-compliant and had exacerbations in-between. Five years back he had symptoms of talkativeness, over-activity, irritability,

delusion of grandiosity, delusion of persecution and decreased need for sleep, for a month. He was taken to a local hospital and treated with tablet olanzapine 20 mg, tablet trifluoperazine 10 mg and tablet sodium valproate 1000 mg. The symptoms gradually improved over the next month. For the last 3 years, there was a gradual onset instability of gait, he would walk slowly and frequently stand still to maintain the balance. There were no tremors, spasticity or sudden fall. He visited a tertiary hospital 2 years ago due to exacerbation of psychotic symptoms and was admitted for 2 weeks. On detail evaluation, he was poorly groomed with a downward gaze, slowness of movement and unsteady gait but no tripping. Vitals were within normal limits, and nervous system examination revealed intact cranial nerves, muscle bulk, tone and power were normal, deep tendon reflex was brisk in bilateral knee, plantar bilateral down-going, sensory function was intact, and cerebellar or meningeal signs were absent. On mental status examination, the speech was slow, affect irritable, delusion of persecution and elementary auditory hallucinations with no insight of illness.

The Brief Psychiatric Rating Scale (BPRS) score was 46 and 20 on admission and discharge respectively. Blood investigations including complete blood picture, renal function test, liver function test, thyroid function test, blood sugar, parathyroid level, serum calcium, phosphorus and albumin were within the normal levels. MRI brain showed symmetrical calcifications along bilateral basal ganglia, thalamii, midbrain, pons, fronto-parietal cortices and cerebellum (Figure: 1). He was diagnosed as a case of Fahr's disease with psychosis (Organic psychosis). Neurological consultation was done and advised physiotherapy for 2 weeks and yearly follow-up. We psycho-educated the caretakers and the patient about the illness and also advocated about the need for medication adherence. Genetic testing and radio-imaging of parents and siblings were unavailable, to look for inheritance. He has been maintaining well with olanzapine and sodium valproate for the last 2 years and is helping in household chores, better in gait, communication, and self-care and regular in follow-up.

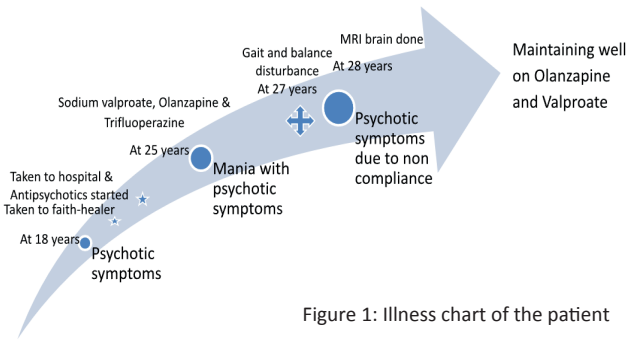

Figure 1: Illness chart of the patient

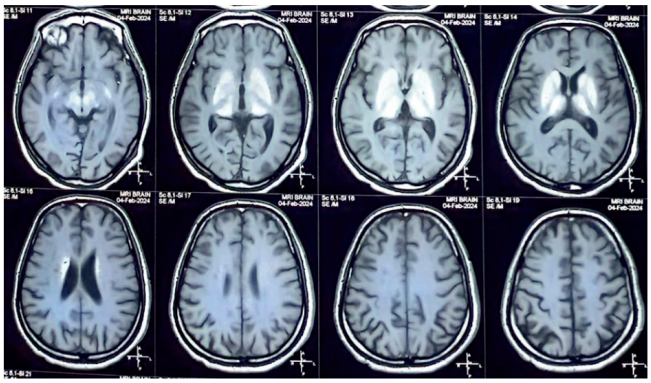

Figure: 2 – MRI brain of Mr D showing symmetrical calcifications along bilateral basal ganglia, thalamii, midbrain, pons and cerebellum

DISCUSSION

Mostly FD is asymptomatic, if symptomatic, it presents with neurological and neuropsychiatric symptoms. Early onset (mean age 30.7 years) presents with psychiatric symptoms with minimal neurological symptoms, as in this case. The late onset FD (mean age 49.4 years) presents with neurological and neuro-cognitive symptoms [7] Psychotic symptoms in FD include symptoms such as auditory and visual hallucinations, perceptual distortions, delusions, ideas of reference or influence, catatonia and fugue state. The neurological presentations are parkinsonism, dystonia, dysarthria, tremors, chorea, seizures, headache, vertigo, gait disturbance, myoclonus, etc. [8].A study (n=62) showed about 50% of patients with a diagnosis of FD initially presented with neurological symptoms and 40% with neuropsychiatric symptoms, but approximately 1% had psychotic symptoms [9]. This shows the rarity of FD and psychosis in FD.

Mohapatra et al. reported a case of a 55-year-old male, with no significant family history, who presented with psychotic symptoms (self-muttering, delusion of persecution, auditory hallucination of commenting type) and neurologic symptoms (tremor). Similar to our case, with lab parameters being within normal limits, and bilateral calcification on neuroimaging, a diagnosis of FD with psychosis was considered [6].

Similar to the response to treatment with mood stabilizers and antipsychotics in our case, Faye et al. reported a case of psychosis due to FD and a response to behavioral disturbances with risperidone and oxcarbazepine [10]. The disease can present with diverse symptoms, often mimicking other neurological and psychiatric disorders, and leading to potential misdiagnoses or delays in appropriate treatment. As a result, it is essential to raise awareness among medical

practitioners about FD and the unique psychiatric manifestations of FD, such as psychosis, to facilitate timely and accurate diagnosis and management. There is no standard treatment guideline for FD yet, nor any cure. Psychotic features can be managed symptomatically with antipsychotics. The diagnostic and treatment guidelines, if available, could have cleared the dilemma of diagnosis and treatment.

We could not confirm the causal relation of FD and psychosis, and could not see the family clustering due to the unavailability of genetic testing. We attempted to put forward a rare condition, FD with psychosis and the management that suited the patient. To conclude, our case highlights the importance of the appropriate use of neuroimaging in the diagnosis of psychiatric illness, where the age of onset is atypical and psychosis presents with neurological manifestations. Knowing the cause of psychosis helps us to target our therapy and to psycho-educate the patient and the caretakers. A comprehensive assessment would guide us to proceed with the best management strategy.

CONCLUSION

Psychotic symptoms in all cases need exclusion of organicity. This highlights the need for neuroimaging in most cases of psychosis. The presentation with motor, behavioral and psychotic symptoms in an incidental FD in this case neither proves association nor causation.

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