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Case Report

Fahr's Disease: An Incidental Finding in a Case Presenting with Psychosis

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Abstract

Fahr's syndrome refers to a rare syndrome characterized by symmetrical and bilateral intracranial calcification. We present a 24-year-old male with Fahr disease, presenting with psychosis and recurrent seizures of generalized tonic clonic type, but lacking evidence of a metabolic disorder. His neurological examination was normal. MRI Brain of the patient revealed symmetrical large areas and foci of calcification in bilateral basal ganglia, thalami, cerebellar parenchyma and subcortical regions of bilateral cerebral hemispheres. When screening other family members, we detected Fahr syndrome in his elder brother with hypocalcemia. Fahr disease may present with psychosis, have pronounced positive brain imaging findings. Magnetic Resonance Imaging can also be effective screening tool for adult relatives (German J Psychiatry 2010; 13 (2): 86-90).

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Introduction

Fahr's disease (FD) is a rare, degenerative, neurological condition characterized by idiopathic calcification of the basal ganglia. This condition has been known since the middle 1800s. The clinical manifestations of Fahr's disease vary. One definition proposed by Trautner et al., 1988 requires bilateral calcifications with neuropsychiatric and extrapyramidal disorders with normal calcium and phosphorus metabolism. Bealle et al., 1989 gave another definition which had seizures, rigidity, and dementia with characteristic calcification of the basal ganglia.

Flint and Goldstein, 1992 opined that radiologists may view basal ganglia calcification (BGC) as an incidental finding so clinical findings associated with Fahr's disease are important. According to Rasmussen et al. (1991) before age 50 incidental discovery of BGC merits diagnostic investigation. The course of Fahr's disease is progressive as reported by Nishiyama et al., 1991. In adult-onset FD, calcium deposition generally begins in the third decade of life, with neurological deterioration two decades later as reported by Manyam et al., 1992, but BGC can also occur in pediatric populations.

The basal ganglia and dentate nucleus are the most common site of involvement and most cases present with extra pyramidal symptoms. This disease is mostly associated with a phosphocalcic metabolism disorder, especially to hypoparathyroidism.

Defective iron transport and free radical production may damage tissue, initiating calcification. (Beall et al, 1989)In adult-onset FD, calcium deposition generally begins in the third decade of life, with neurological deterioration two decades later, (Manyam et al, 1992) Reduced blood flow to calcified regions correlates with clinical signs. (Uygur et al, 1995) Symptoms develop when the deposits accumulate, including progressive deterioration of mental function, loss of previous motor development, spastic paralysis, and athetosis. In addition, optic atrophy may occur.

About 40% of patients with basal ganglia calcification present initially with psychiatric features (Konig, 1989). Cognitive, psychotic, and mood disorders are common. Symptomatic features may change over time. More extensive calcification and subarachnoid space dilatation are known to correlate with the presence of psychiatric manifestations (Konig et al, 1989).

Figure 1: MRI Image of the Patient

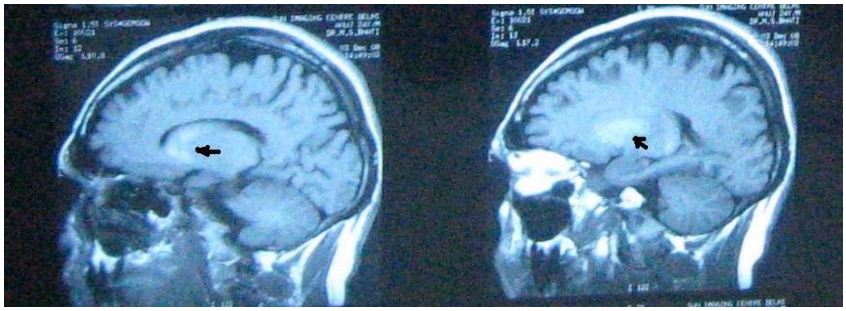
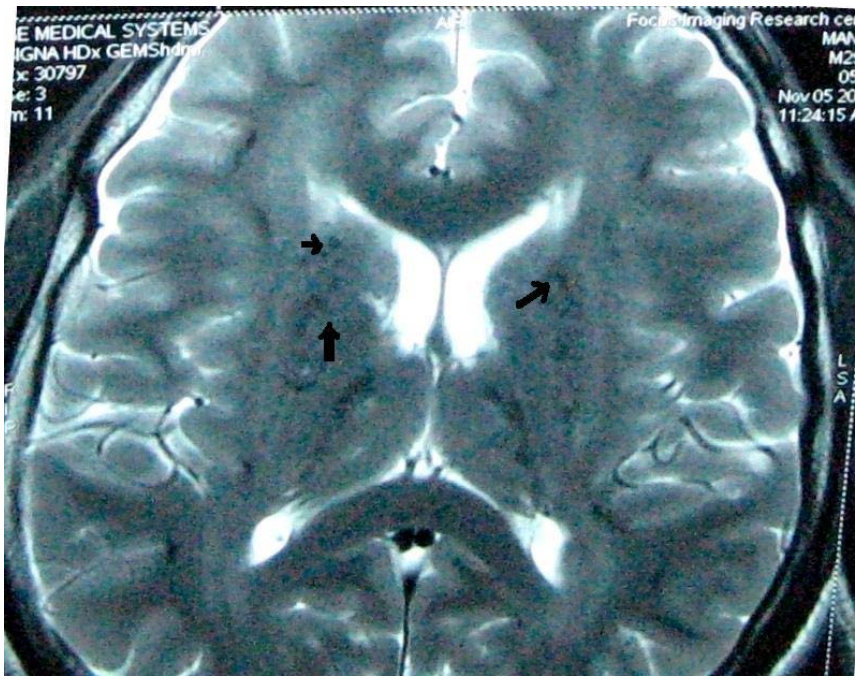


Figure 2: MRI Image of the Patient's Elder Brother



Case report

A 24 year old Hindu male, presented to outpatient clinic of tertiary care centre (Guru Tegh Bahadur Hospital & University College of Sciences, Delhi) with recent onset complaints of aggressive behavior and use of abusive language with friends and family. Relatives also revealed behaviours as smiling to himself, talking to himself, talking to ghosts and being afraid of other people. Recently his sleep pattern had also become quite irregular, sleeping for only three hours a day. On further evaluation, the patient also revealed history suggestive of obsessive behaviors (repeatedly cleaning utensils) four years back and history of seizure disorder since last ten years. Patient had generalized tonic clonic seizures with loss of consciousness. Apparently his scholaristic performance was good till sixth grade but subsequently there was gradual deterioration and he had failed his tenth class examinations. His neurological examination was normal. His mental status examination revealed delusion of persecution, auditory hallucinations as two or more voices discussing among themselves and commanding the patient. These findings were suggestive of psychosis, resembling schizophrenia and patient had difficulty in describing these experiences.

He had three more siblings, an elder brother and two sisters. His elder brother (29 years old) also had similar complaints of recurrent seizures attacks since age of seven years and also had poor academic performance. He had a complaint of knock knee since the age of twelve years. His social behavior was otherwise normal to others. His other two sisters had no neuro-psychiatric complaints. There was no history of similar complaints in any other relatives.

MRI Brain of the patient revealed symmetrical large areas and foci of calcification in bilateral basal ganglia, thalami, cerebellar parenchyma and subcortical regions of bilateral cerebral hemispheres which were suggestive of Fahr's disease in the patient (Figure 1) MRI findings of elder brother also showed symmetrical calcifications in bilateral gangliothalamic complexes, both cerebral and cerebellar hemispheres including dentate nuclei with confluent white matter hyper intensity in both centrum semiovale region, findings suggestive of most probably Fahr's disease in elder brother (Figure 2).

Blood investigations were within normal range for the patient though elder brother had low serum calcium levels of 6mg/dl (normal range of 8mg/dl-11mg/dl) and high serum phosphate levels of 7.7mg/dl (normal range of 3.0mg/dl-5.0mg/dl) for elder brother.

The patient was initially prescribed paliperidone (6 mg/day), haloperidol decanoate (50 mg) and sodium valproate (1500mg/day) and clonazepam (1 mg) for control of agitation. The psychotic symptoms responded poorly to the initial treatment and as subsequently the diagnosis was confirmed amisulpride 200 mg/day was added. The patient showed marked improvement over period of three to four weeks, with psychotic symptoms decreasing and seizures were controlled. Later family members could engage the patient in farming activities. Six months after the initiation of treatment, patient is functioning well as a farmer in village with support of his brothers in the same profession.

Discussion

Fahr's disease may present in familial form with history suggestive of psychosis and generalised tonic clonic epilepsy with intellectual decline. There was no evidence of hypocalcaemia and hyperphosphatemia in the patient, though the elder brother had such evidence and was suggested calcium supplements by endocrinologist.

The patient lacked extrapyramidal symptoms or a metabolic disorder and had normal neurological examination. Similar cases have been reported by Kotan et al. (2009). With such a clinical findings, the presentation of patient in early twenties with recent onset of first episode psychosis with schizophreniform symptomatology can lead to misdiagnosis of schizophrenia or acute transient psychotic disorder (ATPD), especially in Asian country like India where ATPD is far more commoner than schizophrenia.

The clinical expression of Fahr's disease can vary greatly. Symptom scan include features of psychiatric disorders, epileptic seizures and dementia (Modrego et al 2005). But other presentations have also been noted, like Simone et al. (2008) reported case of Fahr's disease with syncope and pseudohypoparathyroidism.

About 40% of patients are known to present initially with psychiatric features like this case, cognitive, psychotic, and mood disorders are common Konig et al, 1989). Paranoid and psychotic features often present between the ages of 20 and 40 in FD (Cummings, 1985). Two patterns of psychotic presentation in FD are known, including early onset (mean age 30.7 years) with minimal movement disorder and late onset (mean age 49.4 years) attended by dementia and movement disorder (Cummings et. al., 1983. This patient had presented at 24 years with psychosis and no extra pyramidal involvement. Symptoms included auditory hallucinations, perceptual distortions and paranoid delusions which have been associated with FD (Rosenberg et al 1991). As in our case schizophreniform psychoses have been reported to present in familial FD (Francis et al, 1984).

On initial brief evaluation in walk in clinic, the patient was initially prescribed paliperidone and haloperidol depot by senior registrar, as there was possibility of poor compliance. As mentioned earlier, the presentation of patient in early twenties with recent onset of first episode psychosis with schizophreniform symptomatology in India raises possibility of diagnosis of ATPD which is far more commonly seen in outpatient clinics than schizophrenia. But as psychosis in setting of FD is known to respond variably to treatment and is sometimes unresponsive (Cummings et. al., 1983). This patient also responded poorly to initial treatment and as diagnosis was confirmed, amisulpiride was added as these patients are known to have high propensity of developing extrapyramidal symptoms due to basal ganglia involvement (Francis, 1979. Subsequently the psychotic symptoms responded well to treatment over three to four weeks with improvement of occupational functioning. Six months later patient is on maintainance treatment, has no psychotic features or seizures and is functioning well in less challenging occupation of farming in village with good social support.

The correction of phospho-calcium metabolism disorder led to clinical improvement in brother, as also noted in other cases (Abe et al., 1996). This case study emphasizes that cases presenting with schizophreniform symptomatology, even in countries with known high incidence of ATPD, must be thoroughly investigated, family history be given due importance and possibility of disorders presenting with protean clinical manifestations as Fahr's disease be considered.

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