

Case Report

A case report of Fahr's Syndrome presenting with psychiatric manifestations

Ashwini CS¹, Chandrashekar TR², Sumathi Arikere³, Keerthana M⁴

¹Postgraduate student, Department of Psychiatry, Belagavi Institute of Medical Sciences, Belagavi, Karnataka.

²Professor and Head, Department of Psychiatry, Belagavi Institute of Medical Sciences, Belagavi, Karnataka.

³Assistant Professor, Department of Psychiatry, Belagavi Institute of Medical Sciences, Belagavi, Karnataka.

⁴Senior Resident, Department of Psychiatry, Belagavi Institute of Medical Sciences, Belagavi, Karnataka.

Corresponding author: Ashwini CS

Email – csashwini47@gmail.com

ABSTRACT

Fahr's disease is a rare neurodegenerative syndrome, which is also known as Familial Idiopathic Basal Ganglia Calcification, where there is presence of idiopathic bilateral and symmetrical calcifications in globus pallidus, putamen, caudate nucleus, dentate nucleus, thalamus and in cerebral cortex and cerebellar areas. A variety of psychiatric symptoms have been reported in patients with Fahr's disease like attention, memory issues and personality changes. Depressive features and psychosis have also been reported. We report here a case of Fahr's disease that also manifested with psychiatric symptoms.

Keywords: Fahr's disease, basal ganglia, calcification, psychiatric manifestations.

(Paper received – 26th January 2023, Peer review completed – 18th February 2023)

(Accepted – 25th February 2023)

INTRODUCTION

Fahr's disease is a rare neurodegenerative syndrome, which is also known as Familial Idiopathic Basal Ganglia Calcification, where there is presence of idiopathic bilateral and symmetrical calcifications in globus pallidus, putamen, caudate nucleus, dentate nucleus, thalamus and in cerebral cortex and cerebellar areas [1-2]. In 1930, German Neurologist named Fahr reported a case of 81-year-old patient with fever, immobility, dementia symptoms and autopsy of brain revealed calcification in the striatum [2].

Cases of idiopathic basal ganglia classification are known as Fahr's disease, whereas Fahr's syndrome refers to those cases which are basal ganglia classification secondary to endocrinological causes [1,3]. Neurological presentation commonly seen are chorea, parkinsonism, dystonia, tremor, gait disturbance, dysarthria, seizures, headache etc. Psychiatric manifestations include psychotic features, mood symptoms, personality changes, anxiety, dementia, apathy, and amnesia etc [3]. Treatment options are symptomatic management and treatment for the cause of calcification, if any. Prognosis of the illness is variable and difficult to predict [4].

CASE REPORT

25 year old male, who was premorbidly well adjusted, presented with decreased sleep, irritability, aggressiveness against family members without provocation, suspiciousness that neighbours were doing black magic against him, were talking about him, because of which he would confront them and fight with them, suspiciousness on mother that she was trying to poison him because of which he had decreased food

intake, talking to self but the content of the talk was not clear to the relatives, would get aggressive when asked about the same. He had the following symptoms for a duration of 2 months. He was also found to be withdrawn to self, with decreased interaction with family members with poor hygiene. He denied of hearing any voices, seeing of images not seen by others. No significant past or family history of any psychiatric illness.

He is a case of neonatal hypothyroidism since birth and was on medication. At 12 years of age, he underwent thyroidectomy and 6 months post-surgery, patient developed seizures, generalised tonic clonic type, which were recurrent. CT and MRI were found to be normal and was then started on antiepileptics but had poor compliance to treatment.

On physical examination, thyroidectomy scar was present. Chvostek sign – negative

On Mental Status Examination, patient was found irritable, had relevant speech, delusions of reference and persecution. Was provisionally diagnosed as Organic delusional (schizophrenia like) disorder.

All relevant investigations were requested.

The laboratory investigations were conducted, and results were as follows –

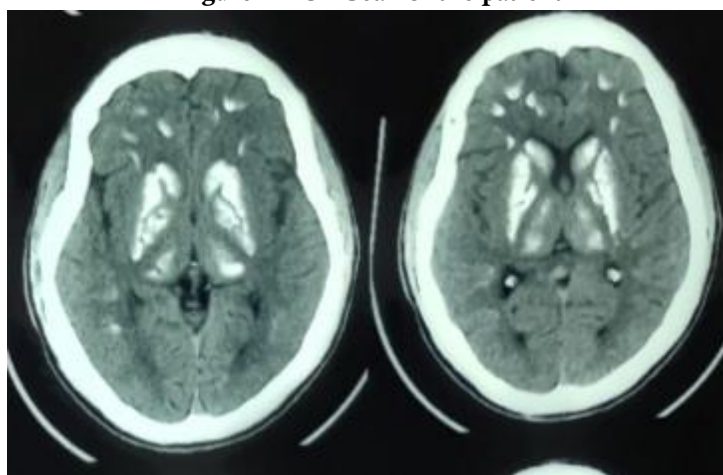
INVESTIGATION	VALUES
CBC/ Serum Electrolytes/ RFT LFT	WNL
ALP	121.6IU/L
Calcium	6.8mg/dL
Phosphorus	8.69mg/dL
PTH	8pg/ml
T3	0.7pmol/L
T4	6.9pmol/L
TSH	58.0mIU

CBC – Complete Blood Count, PTH – Parathormone, RFT – Renal Function Test, T3 -Triiodothyronine, LFT - liver function test, T4- thyroxine, ALP – Alkaline Phosphatase, TSH – thyroid stimulating hormone

The computed tomography scan showed areas of dense calcification in bilateral subcortical, fronto-parieto-occipital temporal white matter, bilateral basal ganglia, bilateral thalami, bilateral dentate nucleus, and bilateral cerebellar lobes (Figure 1).

Patient was diagnosed as case of Fahr's syndrome with psychotic features. Was started on atypical antipsychotics- Tab Olanzapine 10mg and dose was increased to 20mg/day. Medical opinion was obtained for the treatment of comorbid thyroid and calcium abnormalities. Patient was followed up after 1 month, reported of 25% improvement in symptoms. But on subsequent visit, he developed signs of Extrapryramidal symptoms and was then treated for the same.

Figure 1 – CT Scan of the patient



DISCUSSION

It is a rare disorder with prevalence is $<1/1,000,000$. It usually manifests in 3rd decade to 5th decade [5]. May occur in childhood or later in life³. Fahr's disease is idiopathic/genetic whereas Fahr's syndrome occurs secondary to endocrinological cause like hypothyroidism or hypoparathyroidism [1]. Fahr's syndrome is associated with various psychiatric illnesses like anorexia nervosa, dementia, mania, psychosis, depression, OCD, cognitive decline etc [6].

They may have mild difficulty with memory and concentration, and personality changes [5]. Neurological impairment was less seen in patients with extensive calcification (35.8%) and patients with limited calcification were found to have more neurological impairment (34.5), but higher rates of psychiatric disorders (50%) were seen in patients with extensive calcification than limited ones (34.5%) [5]. It has been seen that more extensive calcification and dilatation of subarachnoid space were correlated with psychiatric symptoms, but distribution of calcification couldn't be correlated [7]. Neuropsychological deficits like disturbed selective attention and cognitive flexibility, deficits in declarative memory, verbal perseverations maybe present. There may be deficits in functional circuits, involving basal ganglia and the frontal, parietal, and temporal lobes.

Neurological symptoms like spasticity, gait disorder, speech impairment, dementia, parkinsonism, chorea, tremors, dystonia, myoclonus, and coma are present. In patients who are hypothyroid, with hypocalcaemia, they may also have loss of consciousness and seizures [5]. Cognitive impairment and Positive symptoms, hallucinations, and paranoia maybe due to the destruction of connection between basal ganglia and cortex (cortico-subcortical disconnection), involving the front striatal and limbic circuits which may lead to fronto-temporal atrophy, cortical neurofibrillary aggregate formation and neuronal loss in cortex and basal ganglia [8-9]. 2 patterns of presentation were observed - early onset and Late onset type. Early onset type had psychotic symptoms with minimal movement related disorders and mean age of presentation was 30.9yrs. Late onset type had mean age of 49.4yrs with predominant dementia and movement related disorders [3-4]. Fahr's disease presenting with disturbances in calcium metabolism may have idiopathic hypoparathyroidism, hyperparathyroidism, pseudo-hypoparathyroidism, and postoperative hypoparathyroidism. More mental deterioration was found in patients with parathyroid hormone deficiency secondary to thyroidectomy [2].

Treatment of psychotic symptoms in Fahr's syndrome involves starting the patient with antipsychotics. Patients with Fahr's syndrome were found to be more susceptible to develop neuroleptic malignant syndrome when treated with antipsychotic drugs. So, it's safer to start atypical antipsychotics or those with less extrapyramidal side-effects as there is chances of developing extra-pyramidal symptoms due to the disease process [3].

In this case, the patient presented with early onset type with younger age of onset, no movement disorders despite extensive involvement and more of psychiatry symptoms and extrapyramidal symptoms secondary to antipsychotic drug use and Fahr's syndrome may have occurred secondary to hypoparathyroidism.

CONCLUSION

Fahr's syndrome is a rare neurological disorder which is associated with many other diseases, with genetic basis and can present with psychiatric symptoms. So further research is required to identify the exact mechanism, there may also be a need for finding newer treatment modalities, need for genetic counselling so as to minimize the loss of function [3-5]. Diagnosis of Fahr's syndrome must be considered when there is atypical presentation like when psychosis presents with motor abnormalities or when psychosis presents at a later age [3-10]. Psychiatrists must always give importance to the neurological or medical causative factors which may play role in psychiatric cases and relevant investigations like neuroimaging and endocrinological hormone assays needs to be done to rule out the same [11].

REFERENCES

1. Buono VL, Corallo F, Costa A, Bramanti P, Marino S. Quantitative MR markers and psychiatric symptoms in a patient with Fahr disease. *Am J Case Reports* 2015;16:382.

2. Mufaddel AA, Al-Hassani GA. Familial idiopathic basal ganglia calcification (Fahr's disease). *Neurosci J* 2014;19(3):171-7.
3. Mohapatra S, Satapathy A. A case of schizophrenia like psychosis due to Fahr's disease. *Indian J Psychol Med* 2016;38:155-6.
4. Faye AD, Gawande S, Tadke R, Kirpekar VC, Bhawe SH. A case of psychosis due to Fahr's syndrome and response to behavioural disturbances with risperidone and oxcarbazepine. *Indian J Psychiatry* 2014;56(2):188-90.
5. Saleem S, Aslam HM, Anwar M, Anwar S, Saleem M, Saleem A, Rehmani MA. Fahr's syndrome: literature review of current evidence. *Orphanet J Rare Diseases* 2013;8(1):1-9.
6. Naqvi S, Arshad S, Hanif R, Elfert KA, Elfert KA. Fahr's syndrome misdiagnosed as schizophrenia: a case report. *Cureus* 2017;9(3).
7. Ghormode D, Maheshwari U, Kate N, Grover S. Fahr's disease and psychiatric syndromes: A case series. *Industr Psychiatry J* 2011;20(2):136-8.
8. Savino E, Soavi C, Capatti E, Borrelli M, Vigna GB, Passaro A, Zuliani G. Bilateral stratio-pallido-dentate calcinosis (Fahr's disease): report of seven cases and revision of literature. *BMC Neurol* 2016;16(1):165-8.
9. Hurşitoğlu O, Tuman TC. Fahr's Syndrome Misdiagnosed as Delusional Disorder: A Case Report. *Arch Neuropsychiatry* 2020;57:254-6.
10. Carbone GM, Rocca F. Neuropsychiatric manifestations of Fahr's disease, diagnostic and therapeutic challenge: a case report and a literature review. *Clin Neuropsychiatr* 2019;19(2):121-31.
11. Kumar P, Singh R, Shah K. Psychiatric Manifestations in Fahr's Syndrome: A Case Report. *Cureus* 2020;12(10):e10770.

Acknowledgements – Nil
 Conflict of Interest – Nil
 Funding – Nil