Case Report

Fahr's Disease Presenting with Neuropsychiatric Symptoms: A Case Report

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ABSTRACT

Introduction: Fahr's disease is a rare neurological disorder characterized by idiopathic bilateral calcifications in the basal ganglia and other regions of the brain. This case highlights the diagnostic approach and management of a patient presenting with progressive neuropsychiatric symptoms.

Case Presentation: A 72-year-old male presented with tremors, memory impairment, and disorientation. Neurological examination revealed bradykinesia, reduced arm swing, and stooped posture. Computed Tomography (CT) of brain revealed extensive bilateral symmetrical calcifications in the basal ganglia. Secondary causes were excluded through normal biochemical investigations. The patient was treated with memantine, amantadine, ropinirole, and vitamin D supplementation.

Results: Over six weeks, cognitive function and sleep improved, with reduction in tremors and rigidity.

Conclusion: Fahr's disease should be suspected in elderly patients presenting with progressive neuropsychiatric symptoms and characteristic imaging findings. Early recognition and symptomatic management improve quality of life.

KEYWORDS: Fahr's disease, Basal ganglia calcification, Neuropsychiatric symptoms, Parkinsonism, Case Report.

INTRODUCTION

Fahr's disease, also known as primary familial brain calcification, is an uncommon neurological condition that involves idiopathic bilateral and symmetrical calcifications of brain structures such as the basal ganglia, cerebellar dentate nuclei, thalami, and subcortical white matter. Unlike Fahr's syndrome, which results from underlying metabolic or endocrine disturbances, Fahr's disease typically has no identifiable secondary cause and is often linked to genetic mutations in genes such as *SLC20A2, PDGFB, PDGFRB, MYORG, XPR1,* and *JAM2*.[1-3] The clinical presentation varies widely, ranging from movement disorders to cognitive and psychiatric symptoms.

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CASE PRESENTATION

A 72-year-old man presented to the outpatient department of a tertiary care hospital with progressive tremors in both hands, memory impairment, disturbed sleep, and frequent forgetfulness over the past six years. His family denied any history of seizures, mood disturbances, or motor difficulties. There was no history of heat intolerance, frequent bowel movements, excessive sweating, dizziness, syncope, headaches, or unintentional weight loss. The patient was hypertensive, compliant with his medication, and had no history of smoking.

Physical examination revealed a well-nourished, average-built male, who was conscious but disoriented to time and place. The Mini-Mental State Examination (MMSE) score was 18/30, indicating moderate cognitive decline. His speech was slow and slurred, facial expression was reduced with diminished blinking, and he displayed bradykinesia, a stooped posture, and reduced arm swing. There were no focal neurological deficits, cranial nerve involvement, or primitive reflexes.

Laboratory Findings: To exclude secondary causes, the following investigations were performed which showed normal complete blood counts and his renal and liver function tests were within normal limits. His serum vitamin D levels were slightly below normal, but the serum of calcium, magnesium, phosphorus, alkaline phosphatase and parathyroid hormone (intact) levels were normal. His thyroid function tests were normal and his Cerebrospinal Fluid (CSF) analysis was also normal. His tests for syphilis and Human Immunodeficiency Virus (HIV) were also negative. Tables-I and II.

Neuroimaging: A non-contrast computed tomography (CT) scan of the brain showed extensive bilateral symmetrical calcifications involving the basal ganglia and thalami (as shown in Fig.1), consistent with Fahr's disease.

Family History: The patient's daughter exhibited severe psychiatric symptoms, including mood changes, hallucinations, and irritability, while his son had symptoms suggestive of depression.

Treatment And Follow Up: He was started on a combination of memantine (5mg OD), amantadine

Table-I: Baseline Lab Investigations.

Complete Blood Count		
	Patient's Results	*Normal ranges*
Haemoglobin		12.0-16.0 g/dl
White cell count	7.8*10^9/L	4.0-10.0*10^9/L
Neutrophils	74.7%	40-75%
Lymphocytes	16.6%	20-45%
Platelets	174 *10^9/L	150-400* 10^9/L
Urea	15 mg/dL	7-20 mg/dL
Creatinine	1.0 mg/dL	0.6-1.3 mg/dL
	Patient's Result	*Normal ranges*
Serum Vitamin D	18.7 ng/mL	30-100 ng/mL
Serum Calcium	9.4 mg/dL	8.5-10.5 mg/dL
Serum Magnesium	2.36 mg/dL	1.7-2.2 mg/dL
Serum Phosphorus	3.55 mg/dL	2.5-4.5 mg/dL
Alkaline Phosphatase	50 IU/L	44-147 IU/L
Parathyroid Hormone (PTH) Intact	53.7 pg/mL	16-87 pg/mL
TSH	0.6 mIU/L	0.4-4.0 mIU/L
Syphilis Test	Negative	
HIV Test	Negative	

Table-II: CSF Analysis.

CSF Analysis		
	Patient's Results	*Normal ranges*
Appearance	Clear	Clear
Pressure	15 cm H_2O	< 20 cm H_2O
Glucose	55 mg/dL	50-75 mg/dL
Protein	25 mg/dL	20-40 mg/dL
WCC	0 /mm^3	0-5/mm^3

sulphate (100mg OD), Sinemet (25/250mg BD), ropinirole (0.25mg OD), and supplemented with vitamin D and calcium. At the six-week follow-up, the patient showed noticeable improvement, with better memory retention, reduced tremors, and improved sleep quality.

DISCUSSION

Fahr's disease is defined by abnormal intracranial calcifications in the absence of metabolic or endocrinological disturbances. Genetic mutations—particularly in *SLC20A2*, which encodes a phosphate transporter—are the most commonly implicated[3,4] Mutations in *PDGFB, PDGFRB, MYORG, JAM2,* and *XPR1* have also been identified as contributors to primary familial brain calcification. The disease may present with diverse neurological and psychiatric symptoms, including cognitive decline, parkinsonism, psychosis, and mood disturbances.

Our patient's presentation, along with the CT findings and normal metabolic profile, aligns with the classical presentation of Fahr's disease. The observation of psychiatric manifestations in the patient's children raises the possibility of familial brain calcification, confirms the need for genetic consideration.[5]

CT imaging plays a pivotal role in confirming the diagnosis. There is currently no disease-modifying therapy for primary familial brain calcification; therefore, management is primarily symptomatic, targeting movement disorders, cognitive impairment, and psychiatric manifestations to improve patient quality of life.[1]

Genetic testing is becoming increasingly valuable for confirming diagnoses and understanding phenotypic

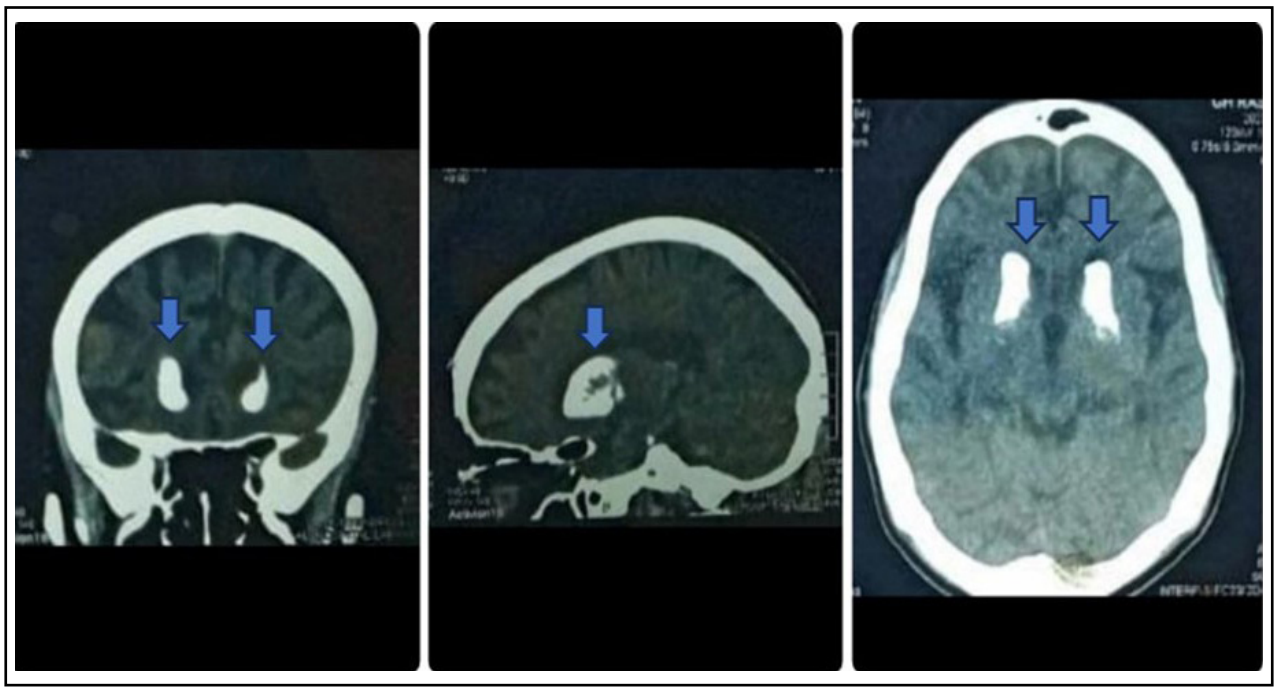

Fig.1: CT brain plain shows calcifications in basal ganglia that are bilaterally symmetrical as shown by blue arrows.

variability, especially with newly identified mutations.

Although neuroimaging was recommended for the family members to assess possible familial involvement, they declined further evaluation and due to cost constrains and unavailability of the genetic testing in Pakistan we could not do genetic confirmation.

In our patient, positive clinical features, laboratory and radiological findings with positive family history points strongly toward the diagnosis of Fahr's disease.

CONCLUSION

Fahr's disease should be considered in patients presenting with unexplained neuropsychiatric symptoms and characteristic calcifications on neuroimaging. The presence of psychiatric symptoms in family members raises the possibility of a hereditary form of the disorder. Early diagnosis and comprehensive management, including neurological, psychiatric, and radiological expertise, are essential for optimizing patient outcomes. Genetic counselling and family screening may also provide valuable insights for early detection and support in affected families.

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Authors Contribution:

SS: Conception of the study, literature review, study design, data interpretation, manuscript drafting, and preparation of the initial manuscript.
ULR: Critical revision of the manuscript for important intellectual content, data interpretation, and correspondence with the journal.
RJ: Study supervision, review of the manuscript, and final approval of the version to be published.
UAA Study supervision, contribution to study design, manuscript review, and final approval of the manuscript.
All authors reviewed and approved the final manuscript and agree to be accountable for all aspects of the work.