**OLGU SUNUMU** / *CASE REPORT*

# Fahr`s Syndrome Accompanied by Psychotic Symptoms and Memory Impairment

## *Psikotik Belirtiler ve Bellek Bozukluğu ile Birlikte Seyreden Fahr Sendromu*

**Beyazit Garip[1], Begum Aritan[2]**

*[1]Department of Psychiatry, Gulhane Training and Research Hospital, Ankara; [2]Department of Psychiatry, Moodist Psychiatric Hospital, Istanbul, Türkiye*

**ABSTRACT**

*This case report highlights the importance of considering Fahr's syndrome in the differential diagnosis of neuropsychiatric disorders and underscores the necessity of a comprehensive metabolic evaluation in affected patients. Fahr's syndrome, also known as Primary Familial Brain Calcification (PFBC), is a rare disorder characterized by bilateral, symmetric intracranial calcifications, neurodegenerative changes, and disturbances in calcium-phosphorus metabolism. A patient presenting with psychotic symptoms and cognitive impairment is described, with an unusual finding of elevated urinary copper levels despite normal blood calcium levels. The case demonstrated typical features of Fahr's syndrome, including bilateral intracranial calcifications and cognitive dysfunction, along with potential disturbances in copper metabolism. This finding highlights the complexity of metabolic abnormalities associated with the condition and the importance of a thorough diagnostic approach. The report emphasizes the need to consider Fahr's syndrome in patients with neuropsychiatric symptoms, particularly those with cognitive impairment and psychotic features. The presence of elevated urinary copper levels in the absence of blood calcium abnormalities suggests the necessity of a comprehensive metabolic assessment. Early recognition and appropriate diagnostic work-up are crucial for guiding treatment and management, as Fahr's syndrome is often overlooked in the presence of more common neuropsychiatric disorders.*

**Key words:** *Fahr syndrome; primary familial brain calcification; neurodegeneration; calcium; psychosis; cognitive impairment*

**ÖZET**

*Bu olgu sunumu, Fahr sendromunun nöropsikiyatrik bozuklukların ayırıcı tanısında dikkate alınmasının önemini vurgulamakta ve etkilenen hastalarda kapsamlı bir metabolik değerlendirme gerekliliğinin altını çizmektedir. Fahr sendromu (güncel literatürde Primary Familial Brain Calcification – PFBC olarak da adlandırılır), bilateral, simetrik intrakraniyal kalsifikasyonlar, nörodejeneratif değişiklikler ve kalsiyum-fosfor metabolizmasındaki bozukluklarla karakterize nadir bir hastalıktır. Psikotik semptomlar ve bilişsel bozukluklar gösteren bir hasta sunulmuş olup, bu vakada kan kalsiyum seviyeleri normal olmasına rağmen idrar bakır seviyelerinin yüksek olması dikkat çekici bir bulgudur. Olgu, Fahr sendromunun tipik özelliklerini, yani bilateral intrakraniyal kalsifikasyonlar ve bilişsel işlev bozukluğunu sergilemiş ve bakır metabolizmasındaki olası bozuklukları da ortaya koymuştur. Bu bulgu, hastalığa eşlik eden metabolik anormalliklerin karmaşıklığını ve kapsamlı bir tanısal yaklaşımın önemini vurgulamaktadır. Sunulan olgu, bilişsel bozukluk ve psikotik belirtiler gösteren hastalarda Fahr sendromunun ayırıcı tanıda göz önünde bulundurulması gerektiğini ortaya koymaktadır. Kan kalsiyum seviyelerinde herhangi bir anormallik olmaksızın idrar bakır seviyelerinin yüksek olması, kapsamlı bir metabolik değerlendirmenin gerekliliğini düşündürmektedir. Fahr sendromu, yaygın nöropsikiyatrik hastalıkların varlığında sıklıkla gözden kaçabileceğinden, erken tanı ve uygun tanısal değerlendirme, tedavi ve yönetimin yönlendirilmesi açısından büyük önem taşımaktadır.*

**Anahtar kelimeler:** *Fahr sendromu; nörodejenerasyon; kalsiyum; psikoz; bilişsel bozukluk*

***İletişim/Contact:*** *Beyazit Garip; Department of Psychiatry, Gulhane Training and Research Hospital, Ankara, Türkiye* • ***Tel:*** *0530 973 11 05* • ***E-mail:*** *beyazitgarip@gmail.com* • ***Geliş/Received:*** *12.03.2025* • ***Kabul/Accepted:*** *12.05.2025*

***ORCID:*** *Beyazıt Garip:0000-0001-8897-0949* • *Begüm Arıtan: 0000-0002-8625-0232*

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## Introduction

Fahr's syndrome is a rare neurodegenerative disorder characterized by bilateral, symmetric intracranial calcifications, particularly in the basal ganglia, thalamus, cerebellum, and subcortical white matter. It was first described in 1930, but its exact pathophysiology remains poorly understood. Fahr's syndrome is associated with several etiologies, including primary genetic mutations (particularly in the MYORG gene), metabolic disturbances (especially calcium-phosphorus imbalances), infections, and endocrine dysfunctions such as hypoparathyroidism. Clinical presentation varies greatly, ranging from asymptomatic calcifications detected incidentally to progressive neurological and psychiatric manifestations. Symptoms may include movement disorders, seizures, psychosis, cognitive decline, and mood disturbances. Neuropsychiatric symptoms can sometimes precede motor abnormalities, leading to frequent misdiagnoses[1–3].

The clinical manifestations of Fahr's syndrome are highly variable, with neuropsychiatric symptoms being among the most prominent. Patients often present with movement disorders, cognitive decline, and psychiatric disturbances, including psychosis, mood disorders, and personality changes. Psychotic symptoms such as hallucinations and delusions have been reported in cases where basal ganglia calcifications disrupt key neurotransmitter pathways, particularly those involving dopamine. Cognitive impairment is also common, with affected individuals exhibiting deficits in executive function, memory, and processing speed. These features can lead to misdiagnosis as schizophrenia or other primary psychiatric disorders[4,5].

Neuroimaging, particularly computed tomography (CT), is crucial in diagnosing Fahr's syndrome, as it reveals the characteristic bilateral calcifications. However, a thorough metabolic and genetic workup is necessary to differentiate idiopathic Fahr's syndrome from secondary causes linked to metabolic disturbances[5].

This case report describes a patient with Fahr's syndrome presenting with psychotic symptoms and cognitive impairment, adding to the literature on its neuropsychiatric manifestations and highlighting the importance of comprehensive diagnostic evaluations.

## Case Report

A 55-year-old female presented with delusions of reference and persecution, a depressive mood, and memory impairment, which had developed over 1.5 months. The patient had no previous psychiatric history or significant medical conditions. She had a university degree and had worked as a teacher until retirement. There was no family history of psychiatric disorders or neurodegenerative diseases.

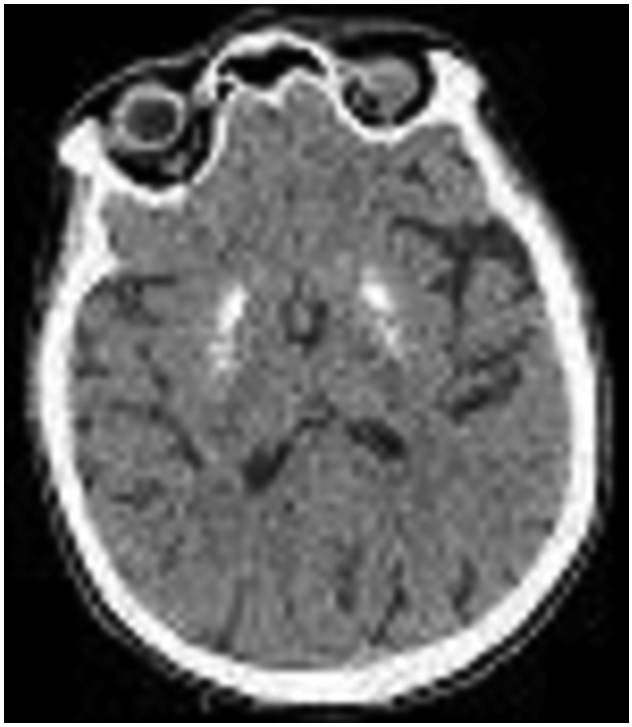

***Figure 1.*** *Tomography of basal ganglia calcification.*

Her symptoms included fear of being harmed, poisoned, and followed, along with disorganized behavior, speech difficulties, anger management issues, and sleep disturbances. Given the acute onset of symptoms and their severity, an organic etiology was suspected. A neurology consultation was requested to rule out underlying structural or metabolic causes of her psychiatric symptoms. The neurological examination revealed mild bradykinesia and subtle gait abnormalities, warranting further neuroimaging.

Brain CT revealed extensive bilateral lentiform nucleus calcifications (Fig. 1), which were highly suggestive of Fahr's syndrome. Laboratory evaluations showed normal blood calcium, phosphate, and parathyroid hormone (PTH) levels, ruling out common metabolic causes of basal ganglia calcifications. However, a 24-hour urine analysis revealed elevated copper levels (113 μg, normal range: 2–60 μg), while blood copper levels remained within the normal range. Additional metabolic and genetic screenings were unremarkable.

Psychiatric evaluation was performed according to the Structured Clinical Interview for DSM-5 Disorders (SCID-5-TR), and a diagnosis of psychotic disorders was made based on DSM-5 TR (Diagnostic and Statistical Manual of Mental Disorders, 5th Edition, Text Revision) criteria. The Positive and Negative Syndrome Scale (PANSS) and neurocognitive assessments were administered. Due to her psychotic symptoms, olanzapine was prescribed at 10 mg/day, leading to significant symptom improvement after four weeks.

## Neuropsychiatric Assessments

*Pre-treatment PANSS Scores:*

- PANSS Positive: 36
- PANSS Negative: 16
- PANSS General: 37
- PANSS Total: 89

*Post-treatment PANSS Scores (week 4):*

- PANSS Positive: 18
- PANSS Negative: 12
- PANSS General: 24
- PANSS Total: 54

The patient demonstrated a significant reduction in positive and general symptoms following olanzapine treatment, with mild improvements in negative symptoms.

### *Neurocognitive Assessment*

The neuropsychological tests indicated deficits in multiple cognitive domains:

**Montreal Cognitive Assessment (MOCA):** The patient showed impairments in visual and spatial functions, attention, language, abstract thinking, and delayed recall, suggesting cognitive decline.

**Audio-Visual Number Sequence Test:** The score of 14 indicated difficulties in attention and short-term memory.

**Benton Visual Retention Test:** A total of 17 errors were observed, indicating significant visual memory impairment. Errors included distortions, misplacements, rotations, and perseverations, distributed across both visual fields.

**Stroop Test:** Prolonged response times in sections 3, 4, and 5 suggested deficits in attention control, response inhibition, and cognitive processing speed.

These findings highlight the presence of cognitive dysfunction in Fahr's syndrome, particularly in executive functioning, memory, and information processing.

## Discussion

Fahr's syndrome is primarily associated with movement disorders; however, psychiatric symptoms, including psychosis and cognitive impairment, are increasingly recognized as significant components of the disease. This case contributes to the literature by emphasizing the importance of considering Fahr's syndrome in patients presenting with new-onset psychotic symptoms and cognitive dysfunction, even in the absence of metabolic abnormalities such as calcium disturbances.

To enhance diagnostic clarity, especially in cases presenting with overlapping neuropsychiatric symptoms, the use of proposed diagnostic criteria for Fahr's syndrome may be beneficial. According to Saleem et al.[1], these criteria include a) bilateral basal ganglia calcifications, b) progressive neurological dysfunction, c) absence of biochemical abnormalities or infectious/metabolic causes, and d) a positive family history or idiopathic presentation. In the presented case, bilateral calcifications and cognitive dysfunction were evident, while the metabolic workup was largely unremarkable except for elevated urinary copper. However, the lack of family history and the incomplete exclusion of differential diagnoses, such as Wilson's disease, represent limitations in applying these criteria comprehensively.

Several case reports in recent years have highlighted the diverse psychiatric manifestations of Fahr syndrome, including cases mimicking late-onset schizophrenia, catatonia, and treatment-resistant psychosis[6]. The current case adds to this growing body of literature by demonstrating a rare presentation of psychotic symptoms accompanied by elevated urinary copper levels. Wazir et al. (2023) and Haider et al. (2025) have emphasized the diagnostic complexity when psychiatric symptoms precede calcification-related motor signs, underscoring the importance of early imaging[7,8]. Notably, Sieffien et al. (2025) also reported psychosis as a primary presentation, emphasizing the need for neuroimaging in atypical psychotic disorders[9].

In contrast to the aforementioned cases, the present case uniquely highlights an atypical copper metabolism

profile without calcium-phosphorus imbalance. This metabolic anomaly, although not diagnostic of Wilson's disease, raises important questions regarding trace metal metabolism in PFBC patients. Although the patient demonstrated marked improvement after four weeks of 10 mg/day olanzapine, long-term follow-up data is limited. Follow-up evaluations revealed no significant extrapyramidal side effects or metabolic disturbances (e.g., weight gain, hyperlipidemia, or glucose dysregulation).

One of the unique aspects of this case is the presence of elevated urinary copper levels, which have not been widely reported in association with Fahr's syndrome. The role of altered metal metabolism in the pathogenesis of the disorder remains unclear. Still, this finding suggests that further research is warranted to investigate potential metabolic contributors beyond calcium and phosphorus disturbances[10].

This case report highlights the need for increased awareness among psychiatrists regarding Fahr's syndrome as a differential diagnosis for psychotic disorders. Misdiagnosis as schizophrenia or primary psychotic disorder can lead to inappropriate treatment and delayed recognition of the underlying neurodegenerative condition. Neuroimaging, particularly CT scans, should be considered in cases where atypical psychiatric symptoms emerge in middle-aged or elderly individuals with no prior psychiatric history[7].

## Conclusion

Fahr's syndrome should be considered in the differential diagnoses for patients with unexplained neuropsychiatric symptoms. The presence of elevated urinary copper levels in this case suggests the need for further research into metabolic disruptions associated with Fahr's syndrome. Early recognition and appropriate psychiatric and neurological management are crucial for improving patient outcomes. Increased awareness of the psychiatric presentations of Fahr's syndrome will aid in timely diagnosis and intervention, ultimately improving patient prognosis.

Although the patient's elevated urinary copper levels raise suspicion for Wilson's disease, further diagnostic steps such as serum ceruloplasmin measurement or ophthalmologic evaluation for Kayser-Fleischer rings were not conducted. As copper accumulation can manifest neuropsychiatric symptoms similar to Fahr's syndrome, this represents a limitation in the differential diagnostic process. Future cases should consider a complete Wilson's disease workup when copper metabolism abnormalities are observed, even in the absence of typical hepatic or ocular findings.

Unlike most reported cases, this patient presented with psychosis as a first and predominant symptom, without motor symptoms or calcium-phosphorus abnormalities. The unusual presence of elevated urinary copper adds a novel biochemical finding not commonly reported in Fahr syndrome literature, raising the possibility of non-traditional metabolic contributors.

This case has several limitations. First, the short duration of follow-up limits conclusions regarding long-term psychiatric and neurological outcomes. Second, although elevated urinary copper was observed, comprehensive testing for Wilson's disease (such as serum ceruloplasmin levels or ophthalmological evaluation) was not performed. Third, genetic testing for known PFBC mutations was not available, precluding a definitive familial classification. Future studies should investigate the role of copper and other trace elements in the pathophysiology of PFBC. In addition, longitudinal case series with genetic and metabolic profiling could enhance our understanding of phenotypic variability and treatment response in Fahr syndrome.

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### *Conflict of Interest Statement*

There are no conflicts of interest.

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## References


1. Saleem S, Aslam HM, Anwar M, Anwar S, Saleem M, Saleem A, et al. Fahr's syndrome: literature review of current evidence. Orphanet J Rare Dis. 2013;8:156.
2. Manyam BV. What is and what is not "Fahr's disease". Parkinsonism Relat Disord. 2005;11(2):73–80.
3. Hoque MA, Siddiqui MR, Arafat Y, Khan SU, Rahman KM, Mondol BA, et al. Fahr's disease: a very rare cause of epilepsy. Mymensingh Med J. 2010;19(1):127–9.
4. De Pieri M, Poglia G, Bartolomei J. Case report:10 years follow-up of psychosis due to Fahr's disease complicated by a left temporal stroke. Front Psychiatry. 2023;14:1283241.
5. Supit VD, Kurniawan D, Fatimah E. Fahr syndrome and neurological manifestations in hypoparathyroidism patients. Radiol Case Rep. 2024;19(4):1248–53.
6. Okumuş B, Hocaoğlu C. Fahr syndrome followed for many years with the diagnosis of schizophrenia: a case report. Psychiatry Behav Sci. 2019;9(1–2):45–9.
7. Wazir MH, Ali Y, Mufti AZ, Ahmad A, Ahmad H. Fahr's syndrome: a rare case presentation. Cureus. 2023;15(10):e47812.
8. Haider A, Danish A, Khan A, Rehman AU, Latif MH, Basit SA, et al. Fahr's syndrome with hypoparathyroidism in a patient with schizophrenia and iron deficiency anemia: a case report. Clin Case Rep. 2025;13(1):e70093.
9. Sieffien W, Sorial S, Ure R, Golts M. Psychosis in Fahr syndrome: a case report. J ECT. 2025;41(1):e1–3.
10. Jaworski K, Styczyńska M, Mandecka M, Walecki J, Kosior DA. Fahr syndrome - an important piece of a puzzle in the differential diagnosis of many diseases. Pol J Radiol. 2017;82:490–3.