



## CASE REPORT

# Fahr Syndrome Followed for Many Years with the Diagnosis of Schizophrenia: A Case Report

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### ABSTRACT

Fahr syndrome (FS) is a rare disorder where bilateral, almost symmetric, calcium and other mineral deposits occur in basal ganglia, cerebellar dentate nucleus and white matter. The clinical pattern is variable and may be associated with neuropsychiatric symptoms, seizures, cerebellar or extrapyramidal dysfunction and dementia. In this study, it was aimed to present FS diagnosed with schizophrenia for many years with misdiagnosis in the context of literature information. FS can sometimes be detected without symptoms. The brain computed tomography (BCT) is the best useful diagnosis method.

**Keywords:** Diagnosis, fahr syndrome, schizophrenia

## INTRODUCTION

Fahr syndrome (FS) is a rare condition which may be characterized by abnormal calcification in the bilateral basal ganglion, thalamus, cerebral cortex, and cerebellar dentate nuclei (1). There are two different definitions of Fahr's disease (FD) and Fahr syndrome (FS). FD is usually autosomal dominant (2). However, there are also studies reporting sporadic and autosomal recessive cases (3,4). The underlying causes of FD is accumulation of minerals such as calcium, iron, zinc, aluminum, magnesium, mucopolysaccharides and glycoproteins due to metabolic disorders such as parathyroid disorders or local circulatory disorders such as regional ischemia and inflammation (5). The term FS is used for idiopathic cases (5). FS was first described by Karl Theodore Fahr in 1930 as an idiopathic basal ganglion calcification (6,7). For the diagnosis of FS three basic features are needed (8). These are bilateral

nonatherosclerotic idiopathic calcification of the basal ganglia, psychiatric symptoms and choreoathetoid or extrapyramidal movement disorders (8). In one study, it was stated that there were two subtypes, including early onset and movement disorders accompanied by psychotic symptoms and late onset with dementia (9). In adults' FD, calcium accumulation usually begins in the third decade of life and neurological deterioration begins 20 years later (10). Approximately %40 of patients with basal ganglion calcification may initially have psychiatric features (10). For this reason, many patients are followed up for a long time with different diagnoses and FD or FS diagnoses may be delayed (11). FS include different neuropsychiatric symptoms and variable clinical manifestations in different patients (12). This situation leads to difficulties in differential diagnosis (12). A standard treatment for FS is not yet known. Regular follow-up exams and treatments for the symptoms that occur due to calcification are mainly administered. It is difficult to predict the prognosis. There is no reliable relationship between age, neurological and psychiatric symptoms of calcium accumulation in the brain. Here, we present a case report of a female patient who presented to our psychiatry outpatient clinic with thoughts

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of aggression, suicidal ideation, and delusions of persecution and diagnosed with FS.

## CASE PRESENTATION

Patient is a 49 years old woman, single, never married, college graduate, was born in Rize and still lives in the same district with her family. The patient was brought to the emergency department of our hospital by her relatives because of the delusions of persecution, aggressive behavior, inability to stay in place and suicide attempt. During the interview with her relatives, the patient had been treated with sertraline 100 mg/day, clomipramine 75 mg/day, amisulpride 400 mg/day and haloperidol 10 mg/day for the last two months and she was discharged hospital one day before her admission to the hospital. After the discharge, the patient was nervous, irritable, and claimed that she was treated badly and a bachelor girl like her was given these medications on purpose. She was stopped by her family when she was about to take too many of her medication at one night.

Twenty years ago, patient's relatives said that she graduated from university in 8 years. Although she worked hard, she could not succeed. After graduating from university, she could not find a job for a while and then she started to work as an officer in a bank. The patient received warnings from his supervisors because of her difficulties in making the calculations during the training period, naming the items, and her increasing forgetfulness. She associated this situation with intensive work stress. In the same period, the patient could not stay alone at home, had a constant state of fear, crying episodes and thoughts of being followed. Because of this situation she had been hospitalized. She did not go to her follow-up visits after discharge and did not use her medication. One year later, she had similar complaints and she was hospitalized for the second time with the diagnosis of schizophrenia. She was discharged from the hospital with the medication of thioridazine, zuclopenthixol, fluvoxamine, and biperiden. At the time, she said that she had simple clumsiness while she was doing works that she was able to do easily before. The patient used the treatment regularly for 10 years. She

thought drugs were responsible of her symptoms such as forgetfulness of doing things and difficulties in naming things. Because of this, patient started reducing her medication without consulting with her doctor. Later, due to complaints of suspiciousness, irritability, crankiness, and insomnia the patient was hospitalized again. After discharge, the patient started to work as an officer in a disability office and because of her forgetfulness, she only did simple calculations and paperwork. When she was doing her job, she constantly worried about forgetting the place where she put the papers. For the last one year, she said that she complained about sleep disturbances, crankiness, and emotional fluctuations during the day accompanied by fear, cries and laughs that she cannot explain why, difficulty using her right and left hands harmoniously and started to stutter. Patient presented to the psychiatry outpatient clinic with these complaints and started on paliperidone palmitate treatment but her complaints did not improve and then she was hospitalized for two months at a local mental health institution. She claimed that she was discharged from the hospital without fully recovering and was brought to our emergency service by her relatives against patient's will. Patient was evaluated with psychiatric consultation in the emergency department of our hospital and then admitted to our inpatient unit for further examination and treatment.

**Personal and Family History:** The patient was born without any complication by normal vaginal birth in hospital accompanied by health personal. The patient did not have any disease in infancy. Hygiene education, walking and talking was on time. When she was in elementary school, her teacher noticed that she was talented in handicraft; that's why her teacher guided her family about her talent. Although she worked so hard, due to forgetfulness she could not succeed in high school and university education. She never married because of her illness. The patient had 5 siblings, her siblings were healthy. Her father's cousin was diagnosed with schizophrenia. Family history did not reveal additional psychiatric and neurological disorders.

**Physical Examination:** Neurological examination revealed short-term memory abnormalities and nominal

aphasia but other systemic examinations was normal. Due to pathological calcifications seen in imaging methods; internal medicine, neurology and neurosurgery departments evaluated the patient with additional methods but no reason was found.

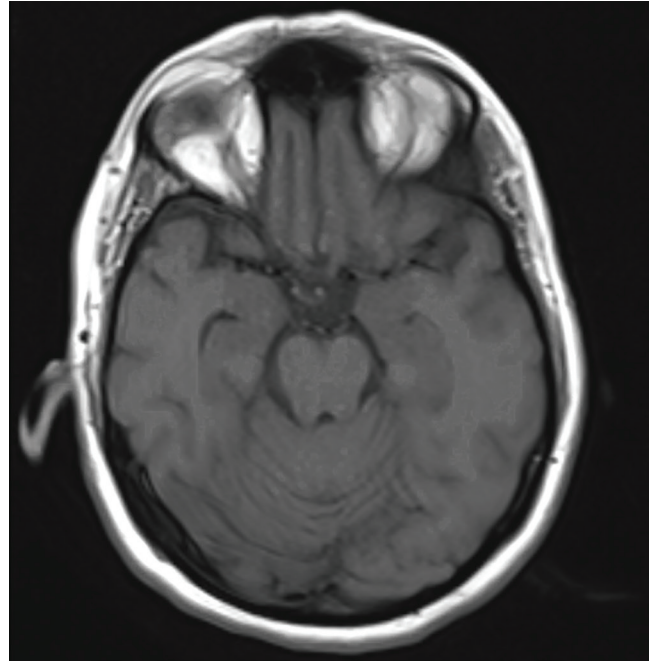
Size: 160 cm, Weight: 62 kg, Body Mass Index (BMI): 24.2 kg/m<sup>2</sup>

**Laboratory Findings:** Complete blood count, biochemical findings of the hormones were normal (thyroid function tests, parathormone, ceruloplasmin, vasculitis markers, vitamin B12, ferritin, and folic acid levels were in the normal range). The 24-hour urinary copper level was normal. Symmetrical calcifications were observed in bilateral basal ganglia in her cranial computerized tomography (CT). Her magnetic resonance imaging (MRI) of the brain showed no pathological findings (Figures 1 and 2). Electroencephalography (EEG) was evaluated as unremarkable with no epileptic discharges.

**Mental State Examination:** The patient's self-care was decreased and she walked with short steps. During the interview, her eye contact was intermittent and she was reluctant to interview. She looked appropriate for her



**Figure 1:** Cranial CT scan of the patient shows symmetrical calcifications in bilateral basal ganglia.



**Figure 2:** Magnetic resonance imaging (MRI) of the brain at basal ganglia level.

age but she acted intimidating. The content of the conversation was poor. She was describing the objects that she could not name in a roundabout way. Her answers to the questions were not purposeful and the reaction time of the patient was slightly longer. The patient was talking with spammers and stutters. Her mood was anxious, affect was anxious, irritable, and her thought content was limited. Her thought content consisted of thoughts of getting harmed and not being able to make sense of her current situation. Her consciousness was open, oriented and cooperative. No pathological finding was detected in the perception. Memory and attention examination revealed short-term memory abnormalities, nominal aphasia, and instantaneous increase of attention against external stimuli. Abstract thought, reality assessment and judgment were impaired, partial psychomotor agitation was accompanying the status.

**Clinical Course:** Following the first psychiatric evaluation in the emergency department, the patient was admitted to our clinic with the diagnosis of schizophrenia according to the DSM-5. In the clinical observations of the patient, it was observed that there were forgetfulness, speech disorders, difficulties in balance, mood changes during the day, persecution against service workers,

referential delusions and inharmoniousness of arms' movement while concurrent adjustment. Her treatment included intra-muscular haloperidol 15 mg/day, biperiden 2 mg/day, chlorpromazine 100 mg/day, due to her lack of oral compliance and aggressive behavior. She fell down from her chair due to lack of coordination in her arms. She could not name some objects. On the 14th day of the treatment, the patient hit her head on the couch in service. Therefore, another CT of the head was performed. Bilateral symmetrical calcifications were observed in the basal ganglia level. No pathological finding was found in the MRI. The current treatment of the patient was changed to clozapine 25 mg/day. In the first month of treatment, clozapine dose was given as 200 mg/day. No hematological side effects were observed. There was a significant decrease in her psychotic symptoms. Positive and Negative Symptoms Assessment Scale-PANSS score was: 65/142 and the Scale for Assessment of Positive Symptoms Scale score was 21/52. As the symptoms of fluctuations in mood persisted during the day, 25 mg/day of lamotrigine was added to the treatment and the drug dose was gradually increased. She was consulted by internal medicine, neurology, and neurosurgery clinics. The requested examinations were performed. She was re-evaluated with the results and included in the outpatient clinic controls without any additional recommendations. The computerized tomography (CT) of the head which was performed on her brother also showed a calcific lesion that is 6.2x4 mm in diameter but it could not be differentiated from the internal tabula at the right vertex level. On the 45th day of hospitalization, she was discharged with clozapine 300 mg/day and lamotrigine 100 mg/day medication.

## DISCUSSION

A female patient who was followed up with a diagnosis of schizophrenia in an outpatient clinic for many years and diagnosed with FS after hospitalization was included in this report. FS includes both neurological and psychiatric symptoms (13,14). Neurological symptoms include Parkinson-like movement disorder, vertigo, epilepsy,

syncope, cerebellar ataxia and dementia (15). Psychiatric symptoms include cognitive, psychotic and mood disorders, especially at the onset of the syndrome (12). Paranoid and psychotic features are usually observed between the ages of 20 and 40 years (14). In our case, it is noteworthy that the delusions of persecution started at the age of 26 years, and in the following period, increased short memory abnormalities and movement disorders were added to the clinical course. Similar to the CT findings of the patient, the presence of calcified lesion of 6.2x4 mm size in diameter which cannot be differentiated from the internal tabula in the frontal area at the right vertex level in her brother's CT may be referring to the genetic transition of FD. MRI, which is one of the commonly used imaging modalities for differential diagnosis, is sensitive to the detection of brain abnormalities; however, it is difficult to define calcifications by routine MRI (16,17). Therefore, CT of head screening is considered to be critical to detect and localize the intracranial calcifications (18,19). In some cases, the data that obtained as a result of neurological examination may be less significant, not persistent, and may not indicate certain localization. These are called soft neurological signs (SNS) (20). Our patient has a vague appearance, articulation difficulties, dysarthria and asymmetry in co-movement which can be attributed to soft neurological symptoms. Recent studies suggest that soft neurological symptoms may be associated with certain brain regions or even brain connections (21,22). In other words, early diagnosis of cases with FS or FD is difficult for clinicians. Since psychiatric and neurological symptoms in each patient may vary from time to time, this causes misdiagnosis of cases with different diagnoses. The patient was diagnosed with schizophrenia and was treated with antipsychotic medication for a long time. She was treated with sertraline, clomipramine and amisulpride for a period of 2 months before hospitalization. This can be explained by the possibility that the relevant treatment team may have diagnosed the patient with the diagnosis of obsessive-compulsive disorder in the differential diagnosis of current psychiatric symptoms during the follow-up of the patient. However, the patient's charts and hospital registry information could not be reached. The symptoms of obsessive-compulsive disorder were not observed during



the follow-up period. In addition to diagnostic difficulties related to FS, information about treatment is limited. Presently, current treatment approaches are for the control of symptoms that cause calcification. In the studies conducted on this subject, it was stated that the response to antipsychotics was variable, patients should be carefully monitored for the development of neuroleptic malign syndrome and especially atypical antipsychotics should be preferred in the treatment (11,14). The preference of clozapine treatment in our case is consistent with these results.

In sum, overlapping clinical manifestations of FS and schizophrenia may lead to diagnostic difficulties. Similar features were present in our case. Clinicians should consider FS as a differential diagnosis in assessment of the psychosis accompanied with neurological symptoms. Our patient has the potential to contribute to the literature in this aspect. This study emphasizes the importance of neuroimaging in patients with atypical psychotic symptoms. MRI is sensitive in detection of brain abnormalities; however, it is difficult to define calcifications by routine MRI. Therefore, it is important to use cranial CT scan screening to detect and localize the size of intracranial calcifications. Further research should focus on the pharmacological treatment of FS. Clinicians are advised to be careful about the presence of FS in patients with atypical neuropsychiatric symptoms.

**Ethics Committee Approval:** The case report is approved by the ethics committee.

**Patient Informed Consent:** Written informed consent was obtained from the patient for the publication of the case report and the accompanying images.

**Conflict of Interest:** Authors declared no conflicts of interest.

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