

Tara Khoeini¹ and Mohammad Javad Sinaeefar¹

¹Iran University of Medical Sciences

September 03, 2024

Addressing Behavioral and Psychiatric Symptoms in Dystonia: A Case Report of Fahr Disease

Tara Khoeini¹, Mohammad javad Sinaeefar^{1*}

¹: Department of Neurology, Firoozgar Hospital, School of Medicine, Iran University of Medical Sciences, Tehran, Iran

* Corresponding author: Mohammad javad Sinaeefar E-mail: Mohammad.javad.snf95@gmail.com Tel: (+98)9118215779, Fax: (+98)21-82141795 Present Address: Department of Neurology, Firoozgar Hospital, Beh Afarin St, Karim Khan Zand Blvd, Tehran, Iran.

Ethical Approval

The researchers were committed and adhered to the principles of the Helsinki Convention and the Ethics Committee of the Iran University of Medical Sciences in all stages.

not-yet-known not-yet-known

not-yet-known

unknown

Consent statement After providing the necessary explanations, written informed consent was obtained from the patient regarding the submission of their clinical condition to medical journals. Additionally, the patient has been assured that their name and personal details will be kept confidential by the authors.

Key Clinical Message:

Abnormal calcium deposits in areas of the brain that control movement, including basal ganglia and cerebellum are the hallmark of Fahr's syndrome. This report highlights the importance of clinicians being vigilant regarding behavioral and neuropsychiatric symptoms when evaluating cases of dystonia. These importing clues indicate underlying inherited and neurodegenerative disorders.

Keywords: fahr's syndrome, fahr's disease, basal ganglia calcification, movement disorders

Introduction:

Fahr's disease, named after German neurologist Karl Theodor Fahr, was first reported in 1930. This rare neurological condition is characterized by abnormal calcium deposits in the basal ganglia, thalamus, dentate nuclei, and subcortical white matter. It is commonly inherited in about 60% of diagnoses in an autosomal dominant manner (Amisha and Munakomi 2023).

The symptoms of Fahr disease, also known as bilateral striatopallidodentate calcinosis, dysarthria seizure, involuntary movements, headaches, dementia, and vision impairment (Rissardo, Caprara et al. 2019Bhandari, Ghimire et al. 2023,). This syndrome can present differently in each patient, and there is no clear correlation between the brain lesion and the clinical symptoms (Amisha and Munakomi 2023).

javascript:void(0)**Case History/examination:**

A 29-year-old right-handed, single, and unemployed woman was referred to Firoozgar Hospital, Tehran, Iran, due to behavioral changes and abnormal posturing in her hands in January 2024. Symptoms began five years ago when the patient slowly developed behavioral changes in the form of aggression and withdrawal behaviors and mood disorders, as well as abnormal movements and postures in the left hand and dysarthria. Subsequently, three years later, the patient also experienced a gait disorder that has been worsening progressively over this period. There was no history of seizure, memory loss, or visuospatial disorder, as well as no symptoms of hallucinations, delirium, or catatonia.

In her personal history, she had normal motor milestones, although her primary education was restricted because of difficulties in learning, leading her to discontinue schooling at the age of 11. In the family history, the patient's parents were first cousins. There were no similar symptoms in other family members. Psychological examination showed a disorganized and slightly disheveled appearance with psychomotor slowness, she has good visual and verbal communication. Consciousness, orientation to time, place, and persons, and also the state of memory, were normal.

In the subsequent examinations, she had a Mini-Mental State Examination (MMSE) score of 22/30. In the neurological examination, she had slurred speech, dystonic posture in her left hand, mild bradykinesia and a masked-face and also dystonic posture in the left foot, which was exaggerated while standing and walking. Kayser-Fleischer (KF) ring was not detected. Eye movements and cerebellar examinations were normal. (Video 1)

Methods (Differential diagnosis, investigations and treatment):

In the brain CT scan, multiple bilateral calcifications were evident in the BG, White Matter, and cerebellar hemispheres, and possible diagnoses of hypoparathyroidism, familial calcification, and Fahr syndrome were proposed for the patient. (Figure 1)

The primary lab tests were all reported within the normal range. (Table 1)

According to the history and the findings of the paraclinical evaluations and lab tests, Fahr syndrome was suggested for the patient. Amantadine 100 mg 2 tablets per day, Levodopa/carbidopa 100/25 twice a day, and sertraline 50 mg daily were started.

Conclusion and Results (Outcome and follow-up):

After three months of follow-up, there was a slight improvement in parkinsonism and dystonic postures. This case report illustrates that recognizing psychiatric and cognitive symptoms, as well as parkinsonism and dystonia, coupled with distinctive imaging results, was critical in establishing the underlying illness. This report, along with findings of imaging and symptom presentation, can be helpful to increase understanding of this disease because our current knowledge of Fahr syndrome is limited and the set of symptoms in these patients is diverse including psychiatric symptoms, behavioral and cognitive changes, and abnormal movements.

Discussion:

It is essential to emphasize that distinguishing between Fahr's disease and Fahr's syndrome is crucial, in which there are specific causes for the calcification of the basal ganglia, such as hypoparathyroidism. Senile calcification of the basal ganglia without clinical symptoms with no underlying cause is an incidental finding that is seen in some people over 60 years old. These are also important differentials Perugula and Lippmann 2016(). When psychiatric symptoms and abnormal movements coincide with idiopathic calcification of the brain in a patient, Fahr syndrome is suggested, especially if it is accompanied by neurological symptoms, cognitive deficits, and some degree of mental retardation (Rücker, Halder et al. 2019).

The gradual onset of symptoms in this patient characterized by behavioral changes and a lower-than-normal IQ, has been reported in several studies Naqvi, Arshad et al. 2017Palu, Moraes et al. 2021)(. Psychiatric manifesting in the patient's third decade of life, followed by movement disorders, is consistent with the early-onset variant of Fahr's disease (Aghemo, Salmanzadeh et al. 2023).

Despite the co-occurrence of behavioral changes and neurological symptoms, along with neuroimaging findings that suggest Fahr syndrome, it has been reported that there is no clear connection between calcifications and neurological symptoms Pistacchi, Gioulis et al. 2016(). The treatment goals involve providing symptomatic support. The response to levodopa in those with Parkinsonian features is reportedly poor. Atypical antipsychotics are the preferred choice for psychiatric symptoms due to the coexistence of the extrapyramidal syndrome in this group of patients (Asokan, D'souza et al. 2013).

Data Availability Statement:

The data that support the findings of this study are available from the corresponding author upon reasonable request.

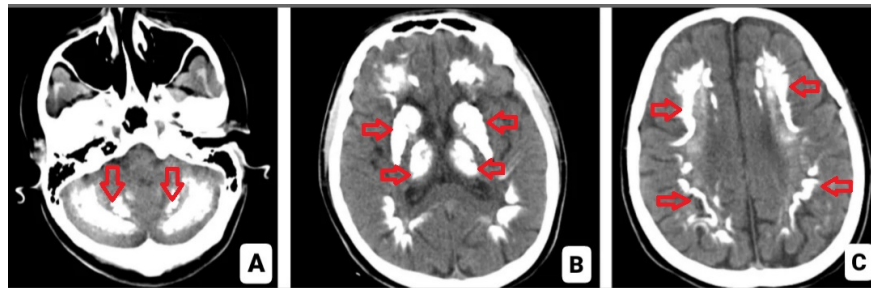
Author Contributions:

Tara Khoeini: Conceptualization; methodology; validation; writing-original draft; writing-review and editing.

Mohammad javad Sinaeefar: Conceptualization; data curation; investigation; writing-original draft; writing-review and editing.

Funding Information:

The authors received no financial support for the research or authorship of this article.



Lab Test	Serum Level	Normal Rnge
Sodium (Na)	135	135-145 mEq/L
Potassium (K)	3.7	3.5-5.5 mEq/L
Calcium (Ca)	8.4	8.5-10.5 mEq/L
Phosphorus (P)	3.7	3.4-4.5 mg/dl
intact parathyroid hormone (iPTH)	27	10-55 pg/mL
Ceruloplasmin	26	20-35 mg/dl

not-yet-known not-yet-known

not-yet-known

unknown

Table 1. The primary lab tests were all reported within the normal range.

Hosted file

Figure 1.docx available at <https://authorea.com/users/799345/articles/1222109-not-yet-known-not-yet-known-not-yet-known-unknown-addressing-behavioral-and-psychiatric-symptoms-in-dystonia-a-case-report-of-fahr-disease>

Hosted file

Table 1.docx available at <https://authorea.com/users/799345/articles/1222109-not-yet-known-not-yet-known-not-yet-known-unknown-addressing-behavioral-and-psychiatric-symptoms-in-dystonia-a-case-report-of-fahr-disease>