Case Report

Fahr Disease Presenting with Progressive Generalized Rigidity Symptomp: A Case Report in a 58 Year-Old Female

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ABSTRACT

Fahr disease, also known as Primary Familial Brain Calcification (PFBC), is a rare neurodegenerative disorder characterized by bilateral intracranial calcifications, most commonly affecting the basal ganglia. Its clinical manifestations are heterogeneous, often leading to diagnostic challenges. We report a case of a 58-year-old female presenting with progressive generalized rigidity, dysphagia, dysarthria, and emotional instability over the course of one year. Physical examination revealed preserved motor strength and sensation, with hyporeflexia on deep tendon reflex assessment. Laboratory findings were largely within normal limits, except for mild hypokalemia and slight elevation of liver transaminases. Non contrast CT Scan of the head demonstrated bilateral, extensive and symmetrical calcifications involving the basal ganglia, thalamus, dentate nuclei, cerebellum, and bilateral occipital cortex, consistent with Fahr disease. No metabolic, infectious, or secondary causes of intracranial calcification were identified, supporting a diagnosis of idiopathic basal ganglia calcification. Management was supportive, including nasogastric tube placement, electrolyte correction, benzodiazepines, physiotherapy, and speech therapy. This case emphasizes the importance of recognizing characteristic radiological findings in patients with progressive neurological and neuropsychiatric symptoms to facilitate early diagnosis and appropriate management.

Keywords : Fahr disease, basal ganglia calcification, PFBC, neurodegenerative disorder
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ABSTRAK

Penyakit Fahr, yang juga dikenal sebagai Primary Familial Brain Calcification (PFBC), merupakan kelainan neurodegeneratif langka yang ditandai dengan kalsifikasi intrakranial bilateral, yang paling sering memengaruhi ganglia basalis. Manifestasi

klinisnya bersifat heterogen, sehingga sering kali menimbulkan tantangan diagnostik. Kami melaporkan kasus seorang wanita berusia 58 tahun yang datang dengan rigiditas umum yang progresif, disfagia, disartria, dan ketidakstabilan emosional selama satu tahun terakhir. Pemeriksaan fisik menunjukkan kekuatan motorik dan sensorik yang masih baik, dengan hiporefleksia pada penilaian refleks tendon dalam. Hasil laboratorium sebagian besar berada dalam batas normal, kecuali hipokalemia ringan dan sedikit peningkatan transaminase hati. Pemindaian computed tomography (CT) kepala tanpa kontras menunjukkan kalsifikasi yang luas, bilateral, dan simetris yang melibatkan ganglia basalis, talamus, nukleus dentatus, serebelum, dan korteks oksipital bilateral, yang konsisten dengan penyakit Fahr. Tidak ditemukan adanya penyebab metabolik, infeksi, maupun penyebab sekunder lainnya dari kalsifikasi intrakranial, sehingga mendukung diagnosis kalsifikasi ganglia basalis idiopatik. Penatalaksanaan yang diberikan bersifat suportif, meliputi pemasangan pipa nasogastrik, koreksi elektrolit, benzodiazepine, fisioterapi, dan terapi wicara. Kasus ini menekankan pentingnya mengenali temuan radiologis yang khas pada pasien dengan gejala neurologis dan neuropsikiatri yang progresif guna memfasilitasi diagnosis dini dan penatalaksanaan yang tepat.

Kata kunci **:** *Penyakit Fahr, kalsifikasi ganglia basalis, PFBC, gangguan neurodegeneratif.*
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INTRODUCTION

Fahr disease is a rare neurodegenerative disorder characterized by bilateral calcification of the basal ganglia. The condition is commonly associated with genetic factors and is often inherited in a familial pattern. It was first described by the German neurologist Theodor Fahr in 1930. Over time, some researchers have considered the term *"Fahr disease"* to be imprecise in reflecting the anatomical and genetic characteristics of the condition. Consequently, a more accurate term, Primary Familial Brain Calcification (PFBC), was introduced in 2013 and has been widely adopted in genetic-based research over the past decade. Other earlier terms include *Idiopathic Basal Ganglia Calcification (IBGC)* and *Bilateral Striopallidodentate Calcinosis* (Batla and Tai, 2015; Carecchio, Mainardi and Bonato, 2023; Magrinelli *et al.*, 2025) Nevertheless, the term *Fahr disease* remains commonly used in the literature. (Balck, Klein and Westenberger, 2004) It is important to distinguish between Fahr disease and Fahr syndrome. Fahr disease refers to idiopathic intracranial calcification without an identifiable underlying cause. In contrast, Fahr syndrome is associated with disturbances in calcium metabolism. It is commonly observed in patients with conditions such as hypoparathyroidism, chronic hypocalcemia, vitamin D deficiency, and chronic kidney disease. These metabolic abnormalities lead to secondary intracranial calcifications. (Batla and Tai, 2015)

The global prevalence of Fahr disease is estimated to be less than 1 per 1,000 individuals.(Carecchio, Mainardi and Bonato, 2023). The condition most frequently presents between the ages of 40 and 60 years, although earlier onset has also been reported. The disease course is typically progressive and tends to worsen with increasing age.(Wazir *et al.*, 2023) The clinical manifestations of Fahr disease are highly heterogeneous, making diagnosis particularly challenging.(Batla and Tai, 2015) Non-contrast CT Scan remains the gold standard imaging modality for detecting intracranial calcifications due to its superior sensitivity compared to magnetic resonance imaging (MRI). (Balck, Klein and Westenberger, 2004; Batla and Tai, 2015)

CASE REPORT

A 58-year-old woman was admitted to Dr. Soegiri Regional Hospital with a chief complaint of generalized body rigidity, predominantly affecting the neck region. The patient also reported difficulty swallowing (dysphagia) and difficulty opening her mouth. The rigidity had been present for approximately one year and had progressively worsened over time. There was no history of penetrating injury or trauma. A history of seizures was unknown. There was no known family history of similar illness.

According to her family, the patient had been previously healthy and active during her younger years, with the onset of symptoms occurring later in life. Since the onset of

illness, she had developed emotional lability and increased irritability. The patient also complained of headache and difficulty speaking.

On physical examination, the patient was afebrile with a blood pressure of 125/80 mmHg, heart rate of 90 beats per minute, and oxygen saturation of 98% on room air. Her Glasgow Coma Scale (GCS) score was E4V5M6. Motor strength and sensory examination were within normal limits. Deep tendon reflexes were hyporeflexic, and the Babinski reflex was negative. Further neurological examination was limited due to the patient's condition and poor cooperativeness.

Laboratory investigations revealed normal serum sodium (143 mEq/L; reference range 136–145 mEq/L) and normal chloride levels (101 mEq/L; reference range 98–106 mEq/L). Serum potassium was slightly decreased at 3.3 mEq/L (reference range 3.5–5.0 mEq/L). Liver function tests showed mild elevation of transaminases, with aspartate aminotransferase (AST) of 59 U/L (reference <37 U/L) and alanine aminotransferase (ALT) of 55 U/L (reference <39 U/L). Renal function and complete blood count were within normal limits. Electroencephalography (EEG) was not performed.

A non-contrast computed tomography (CT) scan of the head demonstrated extensive bilateral and symmetrical calcifications involving the corona radiata, basal ganglia, dentate nuclei, globus pallidus, putamen, thalamus, cerebellum, and bilateral occipital cortices, suggestive of Fahr disease. There was no evidence of midline shift, osteolytic or osteoblastic lesions, ischemia, or hemorrhage.

The patient was managed with nasogastric tube (NGT) insertion, potassium correction, intravenous diazepam, physiotherapy, and speech therapy.

DISCUSSION

Intracranial calcifications are frequently detected incidentally on neuroimaging in both pediatric and adult populations. Based on etiology, intracranial calcifications can be classified into three major categories: physiological (age-related) calcification, genetic pathological calcification, and secondary (non-genetic) pathological calcification. The distinction between physiological and pathological calcifications is primarily based on the onset of symptoms, anatomical distribution, and radiological characteristics. (Magrinelli *et al.*, 2025)

Physiological intracranial calcifications are generally asymptomatic and may be present even in childhood, with increasing prevalence with age. On CT scan, they are typically located in the choroid plexus, falx cerebri, pineal gland and occasionally the basal ganglia, with patterns that may appear punctate, coarse, or curvilinear. (Batla and Tai, 2015)

In contrast, secondary pathological calcifications (Fahr syndrome) are associated with identifiable underlying conditions such as TORCH infections, parathyroid disorders, toxic exposures, or chemotherapy. These conditions demonstrate distinct radiological

patterns and clinical contexts that differentiate them from physiological calcifications and Fahr disease (Batla & Tai, 2015). In the present case, the onset of symptoms occurred at the age of 58 years, and no secondary causes were identified, suggesting a primary (idiopathic) pathological calcification, although confirmation relies on imaging findings. (Batla and Tai, 2015) In the present case, the onset of symptoms occurred at the age of 58 years, and no secondary causes were identified, suggesting a primary (idiopathic) pathological calcification, although confirmation relies on imaging findings.

One of the primary genetic causes of intracranial calcification is Fahr disease, also known as *Idiopathic Basal Ganglia Calcification*. This condition is most commonly inherited in an autosomal dominant pattern, although autosomal recessive cases have also been reported. Neuroimaging, particularly CT scan, typically reveals bilateral calcification of the basal ganglia, especially the globus pallidus, often extending to other regions. (Carecchio, Mainardi and Bonato, 2023) From a pathophysiological perspective, Fahr disease is associated with genetic mutations that lead to abnormal calcium deposition in brain tissue, appearing as hyperdense areas on CT imaging. The most commonly affected regions include the globus pallidus, thalamus, and dentate nucleus, although calcifications may also be observed in the internal capsule, cerebral cortex, subcortical white matter, and brainstem.(Chen *et al.*, 2023; Magrinelli *et al.*, 2025) In this patient, the CT findings were consistent with the typical pattern of Fahr disease.

The clinical manifestations in this case were heterogeneous, including both non-motor and motor symptomps. Motor symptoms included progressive generalized rigidity and speech impairment, while non-motor symptoms included headache and emotional changes. This combination may be explained by calcifications involving the basal ganglia and dentate nucleus, which disrupt neurotransmitter pathways and extrapyramidal circuits. (Mufaddel, 2014)

These findings are consistent with the broad clinical spectrum of Fahr disease, which ranges from asymptomatic cases to complex neurological presentations. Motor symptoms may include parkinsonism (bradykinesia, rigidity, tremor, postural instability), dystonia, ataxia, seizures, and chorea, while non-motor manifestations may include cognitive decline, memory impairment, executive dysfunction, headache, and psychiatric symptoms such as depression, anxiety, psychosis, and personality changes.(Balck, Klein and Westenberger, 2004; Mufaddel, 2014)

Physical examination findings are often unremarkable, with preserved motor strength and sensory function. Laboratory investigations are primarily useful to exclude differential diagnoses, particularly metabolic causes such as abnormalities in serum calcium, phosphate, alkaline phosphatase, and thyroid hormones (Balck, Klein and Westenberger, 2004). In this case, neurological examination was largely normal except for hyporeflexia, which may be attributed to age-related changes or basal ganglia

dysfunction.(Mufaddel, 2014; Taams *et al.*, 2023) Laboratory findings showed mild elevation of liver transaminases, which are likely incidental and not directly related to the pathogenesis of Fahr disease. Other laboratory parameters were within normal limits.

Due to the wide variability in clinical manifestations and the evolving understanding of the disease, diagnosis can be challenging. However, Fahr disease can be diagnosed based on characteristic bilateral intracranial calcifications on CT scan in the absence of identifiable secondary causes.(Chen *et al.*, 2023; Magrinelli *et al.*, 2025)

Management of Fahr disease remains largely symptomatic and supportive, as no definitive therapy has been established to halt or reverse calcification. Treatment options include levodopa for parkinsonian symptoms, trihexyphenidyl or botulinum toxin for dystonia, antiepileptic drugs for seizures, antidepressants for mood disorders, and cautious use of antipsychotics for psychiatric symptoms. (Peters *et al.*, 2020; Magrinelli *et al.*, 2025)

Emerging therapies have been proposed, including bisphosphonates such as alendronate and etidronate, which may inhibit calcium deposition by affecting osteoclastic activity. Although promising, current evidence remains limited and requires further investigation. (Peters *et al.*, 2020; Magrinelli *et al.*, 2025)

Regular follow-up with a neurologist is strongly recommended to monitor disease progression. Physiotherapy, occupational therapy, and speech therapy play crucial roles in maintaining functional independence and quality of life. In advanced cases, nutritional status should be closely monitored due to the risk of malnutrition. Patient education and family support are also essential components of comprehensive care. (Peters *et al.*, 2020; Magrinelli *et al.*, 2025)

CONCLUSION

Fahr disease is a rare cause of pathological intracranial calcification that can present with highly variable and progressive clinical manifestations. This case highlights the importance of considering Fahr disease in elderly patients presenting with chronic rigidity, bulbar symptoms, and behavioral changes, particularly when symptoms are progressive and not accompanied by identifiable metabolic abnormalities.

Non-contrast head computed tomography (CT) plays a crucial role in establishing the diagnosis through the identification of characteristic bilateral basal ganglia calcifications. Early recognition of the clinical and radiological features of Fahr disease is essential to prevent diagnostic delay and to facilitate appropriate supportive management planning.

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