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Earliest Manifestation of Idiopathic Hypoparathyroidism Presenting as Fahr Syndrome with Chronic Hypocalcaemia: A Rare Case Report from Saudi Arabia

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ABSTRACT

Fahr's syndrome is a rare neurological condition characterized by symmetrical intracranial calcifications. We report a case of 43-year-old Saudi female with a known history of epilepsy who presented with fever, recurrent tonicclonic seizures, and a productive cough. Initial investigations revealed severe hypocalcemia, hypomagnesemia, and hypokalemia, alongside low parathyroid hormone levels. Imaging studies, including CT confirmed symmetrical calcifications in the basal ganglia, thalami, dentate nuclei, and corona radiata, consistent with Fahr's syndrome. The patient was diagnosed with Fahr's syndrome secondary to idiopathic primary hypoparathyroidism. There is no specific treatment is currently available, therefore, the patient treated with intravenous calcium gluconate, magnesium, potassium replacements, vitamin D supplementation, and antibiotics for aspiration pneumonia. Neuropsychiatric symptoms and seizures improved following electrolyte normalization, and she was discharged on maintenance therapy. This case highlights the importance of recognizing systemic manifestations of electrolyte imbalances, particularly in rare disorders like Fahr's syndrome, to enable prompt diagnosis and appropriate management.

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Introduction

Fahr's syndrome, also known as idiopathic basal ganglia calcification, is a rare neurological disorder characterized by symmetrical calcification in the basal ganglia and other brain regions, including the cerebellum, thalamus, and cortex. This condition, initially identified by Karl Theodor Fahr in 1930, frequently manifests with a spectrum of neuropsychiatric symptoms, such as movement disorders, cognitive impairment, and seizures [1,2].

It is important to distinguish Fahr's syndrome from Fahr's disease; while Fahr's disease refers to a primary, genetic form with no underlying metabolic abnormalities, Fahr's syndrome is typically secondary to conditions like hypoparathyroidism and is associated with disruptions in calcium and phosphate metabolism [2,3].

The most common metabolic cause of Fahr's syndrome is hypoparathyroidism, which leads to chronic hypocalcemia and calcifications in brain regions, emphasizing the necessity for comprehensive metabolic and endocrine examination in patients presenting with basal ganglia calcifications [2,4].

Clinically, Fahr syndrome can present with a wide range of symptoms, including neurological and neuropsychiatric manifestations such as movement disorders (e.g., Parkinsonism, chorea, tremors), cognitive dysfunction, and psychiatric symptoms [5]. Diagnosis relies on neuroimaging, particularly CT scans, which reveal the characteristic bilateral calcifications, while treatment focuses on managing the underlying metabolic abnormalities to alleviate symptoms and potentially limit progression [6,7].

Case Report

A 43-year-old Saudi female presented with a history of fever and recurrent tonicclonic seizures. She reported a productive cough and dyspnoea that started a week prior. Additionally, she described preictal symptoms, including headaches and hand tetany, which progressed to tonicclonic seizures. There was no associated weakness or numbness of the extremities. She had a known history of epilepsy, controlled on anti-epileptic medications: carbamazepine, valproic acid, and olanzapine.

On examination in the emergency department, the patient was conscious, alert, and oriented, with stable vital signs. Neurological examination was normal, while chest examination revealed bilateral basal crackles. Chvostek's and Trousseau's signs were negative. Initial laboratory findings revealed severe hypocalcemia (3.8 mg/dL), hypomagnesemia (1.44 mg/dL), hypokalemia (2.4 mmol/L), albumin (4 g/dL; normal), parathyroid hormone (PTH) (0.47 ng/mL; low), thyroid stimulating hormone (TSH) (6 µIU/mL; mildly elevated), free T3 (4.2 pmol/L), and free T4 (13.73 pmol/L).

An electrocardiogram showed a prolonged QT interval. A chest X-ray revealed lower zone opacities, and high-resolution computed tomography (HRCT) demonstrated atelectasis bands, bilateral

posterior pleural thickening, and a small area of consolidation, consistent with aspiration pneumonia. A head CT scan revealed bilateral symmetrical dense calcifications in the dentate nuclei, basal ganglia, thalami, and corona radiata, suggestive of Fahr's syndrome (Figures 2 and 3).

The patient was diagnosed with Fahr's syndrome secondary to idiopathic primary hypoparathyroidism. She was admitted to the intensive care unit for management of severe hypocalcemia and aspiration pneumonia. Intravenous calcium gluconate was immediately started and continued until normalization of calcium levels and resolution of the QT interval prolongation, followed by oral calcium and alfacalcidol supplementation. Intravenous magnesium and potassium replacements were administered to correct hypomagnesemia and hypokalemia. Additionally, she started on antibiotics for aspiration pneumonia and completed a five day course. Carbamazepine and valproic acid were prescribed to prevent further seizures.

During hospitalization, the patient's symptoms improved as serum calcium levels normalized. She remained stable without further seizures and was discharged on maintenance therapy with calcium carbonate, alfacalcidol, vitamin D, and antiepileptic medications.

This case illustrates the rare presentation of Fahr's syndrome due to idiopathic primary hypoparathyroidism, highlighting the importance of recognizing systemic manifestations of electrolyte imbalances and initiating prompt treatment.

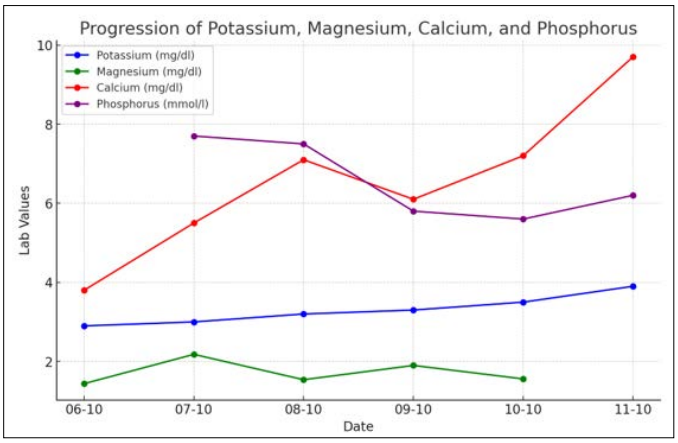

Figure 1: Graph Shows the Progression of Key Laboratory Values (Calcium, Potassium, Magnesium, and Phosphorus) during Hospitalization.

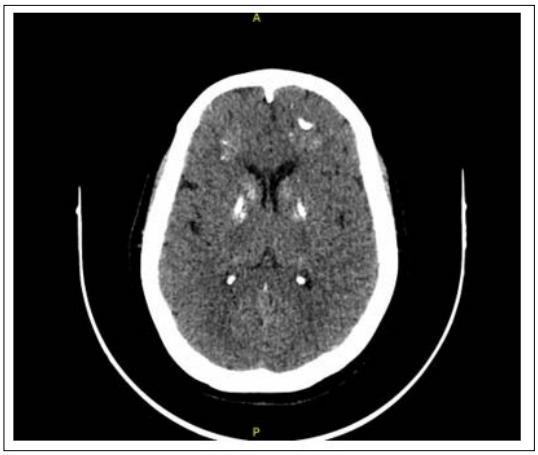

Figure 2: CT Brain Showing Calcification in Basel Ganglia and Thalmi

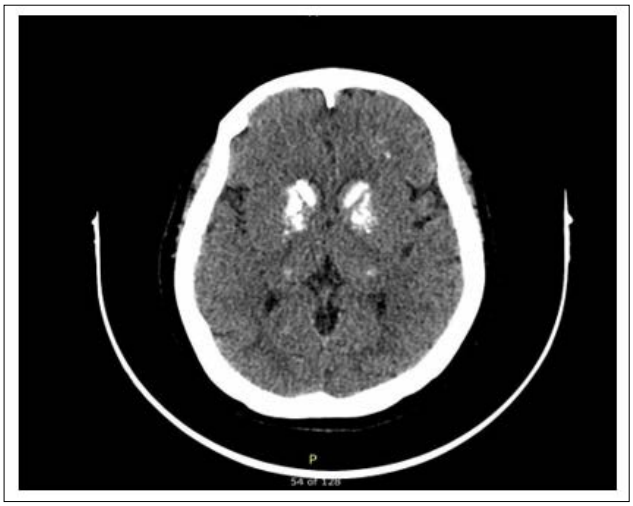

Figure 3: CT Brain Sowed in Calcification Basel Ganglia

Discussion

Fahr's syndrome, also known as idiopathic basal ganglia calcification, is a rare neurological disorder characterized by symmetrical calcifications in specific brain regions, such as the basal ganglia, thalami, cerebellum, and cortex. First described by Karl Theodor Fahr in 1930, it presents a spectrum of neuropsychiatric symptoms including movement disorders, cognitive deficits, and seizures [1,2]. The exact pathophysiology remains unclear but is thought to involve disrupted calcium metabolism and impaired clearance mechanisms in the brain [1,2].

While Fahr's disease denotes the primary genetic form, Fahr's syndrome is typically secondary to metabolic or endocrine abnormalities, with hypoparathyroidism being the most common cause [4]. Regarding the prevalence of Fahr syndrome in Saudi Arabia, the medical literature does not address this issue. Fahr syndrome is a rare disorder, therefore thorough epidemiological data for Saudi Arabia or the gulf regions may not be readily available. With an incidence of less than 1 in a million, its rarity underscores the need for heightened clinical awareness. It is crucial to remember that the prevalence of rare conditions varies greatly across populations and can be impacted by genetic, environmental, and healthcare access variables [8].

Comparison of Fahr's Syndrome Case Reports in Saudi Arabia. The uploaded case report describes a 43-year-old Saudi female who developed Fahr's syndrome due to idiopathic primary hypoparathyroidism and presented with seizures, electrolyte abnormalities, and aspiration pneumonia. In example, Khan et al. (2020) documented a 34-year-old Ethiopian female with Fahr's syndrome caused by hypoparathyroidism, which manifested largely as seizures, tetany, and altered consciousness [7]. Both cases show the common underlying cause of Fahr's syndrome in hypoparathyroidism, which results in hypocalcaemia and consequent brain calcifications. Neuroimaging in all studies revealed symmetrical calcifications in the basal ganglia and thalami, which are characteristic of the condition. Both instances were treated with intravenous calcium gluconate and vitamin D supplements to correct hypocalcaemia, as well as anti-epileptic medications to manage seizures.

Differences in Imaging

While both cases included calcifications in the basal ganglia and thalami, ours included dentate nuclei and corona radiata. This could imply a more advanced or severe presentation, possibly

due to a delay in diagnosis or progression [8]. Additionally, the involvement of the dentate nuclei may correlate with specific symptoms, such as cerebellar dysfunction, which should be monitored [8].

Differences in Management

In our case, management comprised antibiotics for aspiration pneumonia as well as electrolyte replacement (magnesium and potassium), indicating a more complex clinical picture. In contrast, the Khan et al. [7] case cantered on calcium and vitamin D supplements, as well as seizure management. The inclusion of antibiotics highlights the importance of addressing complications arising from delayed diagnosis.

Fahr Syndrome and Cognitive Impairment

Our patient exhibited some elements of cognitive impairment. Cognitive symptoms in Fahr syndrome are associated with widespread brain calcifications, particularly in regions involved in executive function and memory. Psychiatric symptoms associated with Fahr Syndrome are diverse and can include cognitive deficits, mood disorders, and psychotic symptoms [9]. Cognitive impairments often manifest as memory disturbances, executive dysfunction, and attentional deficits [10,11]. Mood disorders such as anxiety and depression are common and can be resistant to treatment [10]. Psychotic symptoms, although less frequent, can be severe and include delusions, hallucinations, and disorganized behaviour [10,12,13]. Management of these symptoms can significantly affect the patient's quality of life. In our case, further neuropsychiatric evaluation would provide insights into the progression of cognitive impairments and their management [14].

Conclusion

Our case is the first recorded occurrence of Fahr's syndrome in a Saudi patient, especially because of idiopathic primary hypoparathyroidism. This example underlines the necessity of diagnosing systemic electrolyte abnormalities, as well as the vital requirement for early detection and complete treatment. By reporting this distinct presentation, we want to raise awareness of Fahr's syndrome among Saudi practitioners and contribute to the increasing worldwide literature on this uncommon neurological condition.

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