



Diagnosis of Fahr syndrome in a patient with cerebral palsy and hypothyroidism: a case report

Diagnóstico da síndrome de Fahr em um paciente com paralisia cerebral e hipotireoidismo: um relato de caso

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ABSTRACT

We present the case of a 51-year-old male with cerebral palsy and hypothyroidism, admitted due to recurrent upper limb spasms and a generalized tonic-clonic seizure. Initial evaluation revealed symptomatic hypocalcemia and extensive intracranial calcifications, leading to a diagnosis of Fahr syndrome. Clinical management included calcium supplementation, resulting in progressive improvement and hemodynamic stability. This case highlights the importance of considering Fahr syndrome in patients with unexplained seizures and metabolic disturbances.

Keywords: Fahr syndrome; Hypocalcemia; Intracranial calcifications; Calcium disorder; Hypothyroidism.

RESUMO

Apresentamos o caso de um homem de 51 anos com paralisia cerebral e hipotireoidismo, admitido devido a espasmos recorrentes nos membros superiores e uma crise tônico-clônica generalizada. A avaliação inicial revelou hipocalcemia sintomática e extensas calcificações intracranianas, levando ao diagnóstico de síndrome de Fahr. O manejo clínico incluiu suplementação de cálcio, resultando em melhora progressiva e estabilidade hemodinâmica. Este caso destaca a importância de considerar a síndrome de Fahr em pacientes com crises convulsivas inexplicáveis e distúrbios metabólicos.

Descritores: Síndrome de Fahr; Hipocalcemia; Calcificações intracranianas; Distúrbios do cálcio; Hipotireoidismo.

INTRODUCTION

Fahr syndrome is a rare neurodegenerative disorder characterized by abnormal calcifications in various brain regions, particularly the basal ganglia, cerebellum, and cerebral cortex⁽¹⁾. The syndrome presents with a range of neurological and psychiatric symptoms, including movement disorders, cognitive impairment, and seizures. Fahr syndrome is often associated with metabolic disturbances, such as hypocalcemia, hypoparathyroidism, and other electrolyte imbalances⁽²⁾. The pathogenesis involves the deposition of calcium

and other minerals in the brain, leading to progressive neurological dysfunction⁽¹⁾.

- The diagnostic criteria for Fahr syndrome include⁽³⁾:
- **Bilateral Calcifications:** Detected in the basal ganglia, cerebellum, and other brain regions through neuroimaging (CT).
 - **Progressive Neurological Dysfunction:** Manifesting as movement disorders, cognitive decline, neuropsychiatric symptoms, and seizures.
 - **Exclusion of Other Causes:** No other biochemical abnormalities, metabolic disorders, or infectious diseases explaining the calcifications.

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- **Family History:** In some cases, a family history suggesting autosomal dominant inheritance.

OBJECTIVE

To report a case of Fahr syndrome presenting with symptomatic hypocalcemia and recurrent seizures in a 51-year-old male with a history of cerebral palsy, emphasizing the diagnostic challenges and clinical management strategies.

CASE PRESENTATION

A 51-year-old male was admitted on February 21, 2024, after experiencing recurrent spasms in the upper limbs and a generalized tonic-clonic seizure. These episodes occurred following a recent fall. There was no associated history of head trauma.

The patient had a known diagnosis of cerebral palsy and a history of three childhood seizures. He was on Levothyroxine and Phenobarbital for hypothyroidism and seizure control, respectively. Additionally, he had undergone an adenoidectomy.

IMAGING TEST

Multislice computed tomography (CT) of the skull performed on February 21, 2024, revealed:

- Extensive calcifications in the cerebellar hemispheres and basal ganglia.
- Punctate calcifications in cortical sulci.

RELEVANT LABORATORY TESTS

- Ionized calcium: 0.56mmol/L (February 21, 2024) → 0.96mmol/L (February 25, 2024).
- Parathyroid hormone (PTH): 9.3pg/mL (February 25, 2024).
- Others: Potassium 3.1mmol/L, Lactate 5.4mmol/L, Creatine kinase (CK) 1684U/L.

CLINICAL PROGRESSION

Following admission, the patient was treated with calcium supplementation and monitored to prevent further seizures. No additional anticonvulsants were required as there were no recurrent symptomatic episodes. The patient showed progressive clinical improvement and maintained hemodynamic stability.

DISCUSSION

Fahr syndrome is often diagnosed through imaging studies that reveal characteristic intracranial



Figure 1. A and B: Calcifications in the cerebellar hemispheres.

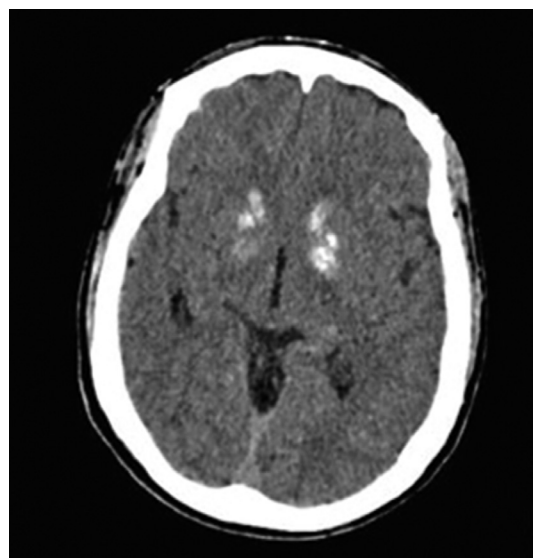


Figure 2. Symmetrical bilateral calcifications in the basal ganglia.

calcifications⁽⁴⁾. In this case, the CT scan findings, coupled with the patient's clinical presentation and laboratory results, supported the diagnosis of Fahr syndrome. The patient's hypocalcemia and low PTH levels suggest a possible underlying hypoparathyroidism, a known association with Fahr syndrome^(5,6).

In this patient, the combination of cerebral palsy, hypothyroidism, and recurrent seizures posed a diagnostic challenge. However, the presence of extensive intracranial calcifications was a key indicator for Fahr syndrome. The correlation between hypoparathyroidism and Fahr syndrome highlights the importance of thorough metabolic evaluation in patients with unexplained neurological symptoms^(5,1).

The management of Fahr syndrome primarily focuses on addressing the underlying metabolic disturbances. In this case, calcium supplementation effectively managed the hypocalcemia, leading to clinical improvement. Continuous monitoring and long-term management strategies are essential to prevent recurrent symptoms and complications⁽¹⁾.

Fahr syndrome can present with a variety of neurological and psychiatric symptoms, which can complicate the diagnosis. Literature reports varied presentations of Fahr syndrome, ranging from asymptomatic individuals to those with severe neurological impairment. The progression and severity of symptoms are often linked to the extent of calcification and underlying metabolic disturbances⁽⁴⁾. Early diagnosis and intervention can significantly improve patient outcomes, as seen in this case, where timely calcium supplementation led to clinical stability.

A multidisciplinary approach involving neurologists, endocrinologists, and radiologists is essential in managing complex cases of Fahr syndrome. Comprehensive management plans that address both the neurological and metabolic aspects of the disease are crucial for improving patient quality of life⁽³⁾.

In addition to calcium supplementation, addressing other potential metabolic imbalances is crucial. In this

case, the patient's potassium and lactate levels were also monitored and managed to prevent additional complications. The importance of regular follow-up and monitoring cannot be overstated, as Fahr syndrome patients are at risk of recurrent symptoms and potential progression of neurological deficits⁽³⁾.

Further research is needed to better understand the pathophysiology of Fahr syndrome and its varied clinical presentations. Genetic studies have shown familial cases with autosomal dominant inheritance patterns, indicating a possible genetic component to the disease⁽⁷⁾. Understanding these genetic factors could lead to improved diagnostic and therapeutic approaches in the future.

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