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# Fahr Syndrome Associated with Pseudohypoparathyroidism in an Adolescent: A Rare Case with Diagnostic and Therapeutic Challenges

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**Abstract** | **Case Report**

Fahr syndrome (FS) is a rare neurological disorder characterized by bilateral and symmetrical intracerebral calcifications, mainly affecting the basal ganglia, thalamus, and cerebellar nuclei. It is frequently associated with metabolic disturbances, particularly parathyroid dysfunctions such as pseudohypoparathyroidism (PHP). PHP is a rare inherited condition marked by resistance to parathyroid hormone, resulting in hypocalcemia and hyperphosphatemia despite normal or elevated PTH levels. This article reports the case of a 15-year-old adolescent presenting with abnormal movements and confusion, found to have severe hypocalcemia and extensive brain calcifications on imaging. The diagnosis of Fahr syndrome secondary to PHP was established. The patient was treated with intravenous then oral calcium and active vitamin D, leading to clinical and biochemical improvement. This case highlights the diagnostic and therapeutic challenges posed by the rare association of FS and PHP, underlining the importance of early recognition, multidisciplinary management, and regular follow-up to prevent irreversible neurological complications. The article also reviews the pathophysiology, clinical features, and management strategies for FS and PHP, emphasizing the need for awareness of metabolic causes in patients with unexplained neurological symptoms and intracranial calcifications.

**Keywords:** Fahr Syndrome, Pseudohypoparathyroidism, Intracerebral Calcifications, Hypocalcemia, Adolescent, Metabolic Disorders, Neurological Symptoms.



## INTRODUCTION

Fahr syndrome (FS) is a rare clinical entity defined by bilateral and symmetrical intracerebral calcifications, mainly located in the basal ganglia, thalamus and serrated nuclei. First described by Theodor Fahr in 1930, it is often associated with metabolic disorders, notably parathyroid dysfunction such as hypoparathyroidism or pseudohypoparathyroidism (PHP) [1, 2].

PHP is a group of rare inherited metabolic disorders characterized by resistance of target tissues to parathyroid hormone (PTH), resulting in hypocalcemia and hyperphosphatemia despite normal or high levels of PTH [3]. PHP is classified into several subtypes according to the presence or absence of specific clinical features (such as skeletal abnormalities or multiple hormone resistances) [4].

The clinical manifestations of FH are varied, ranging from seizures to neuropsychiatric disorders and motor abnormalities [5]. The association between FH and PHP is exceptional, but represents a significant diagnostic challenge, as it can delay appropriate management. Chronic hypocalcemia, common in PHP, promotes the formation of cerebral calcifications, explaining this association [6]. We report here on a clinical case illustrating this association, followed by a discussion of the diagnostic and therapeutic implications.

## CASE REPORT

A 15-year-old patient, with no significant medical history, presented three months prior to hospitalization with abnormal movements. The condition progressed, and one week before admission, fasciculations and a confusion syndrome appeared, prompting the family to seek emergency care. Upon admission, severe hypocalcemia at 32 mg/L was identified.

**Initial Workup:**

**Complete Blood Count (CBC):**

White Blood Cells (WBC): 4890/mm³ Neutrophils: 3940/mm³ Lymphocytes: 480/mm³

Hemoglobin (Hb): 10.5 g/dL

**Phosphocalcic Panel:**

Parathyroid Hormone (PTH): 147 pg/mL (normal: 10–65)

Phosphatemia: 52 mg/L (elevated)

Corrected Calcium: 32 mg/L (low)

Magnesium and Vitamin D levels not measured

24-hour Urinary Calcium: 33.65 mg/24h

**Electrolytes:**

Sodium: 132.47 mmol/L Potassium: 3.43 mmol/L

Bicarbonates: 18 mmol/L

**Renal Function:**

Creatinine: 6.12 mg/L

Glomerular Filtration Rate (GFR): 189.41 mL/min/1.73 m²

**Liver Function Tests:**

AST: 53 IU/L ALT: 34 IU/L GGT: 12 IU/L

**Urinalysis (ECBU):** sterile

**Lumbar Puncture:** sterile

**Brain Imaging:**

A CT scan revealed deep, bilateral, and symmetrical calcifications located in both supra- and infratentorial regions, consistent with Fahr's syndrome.

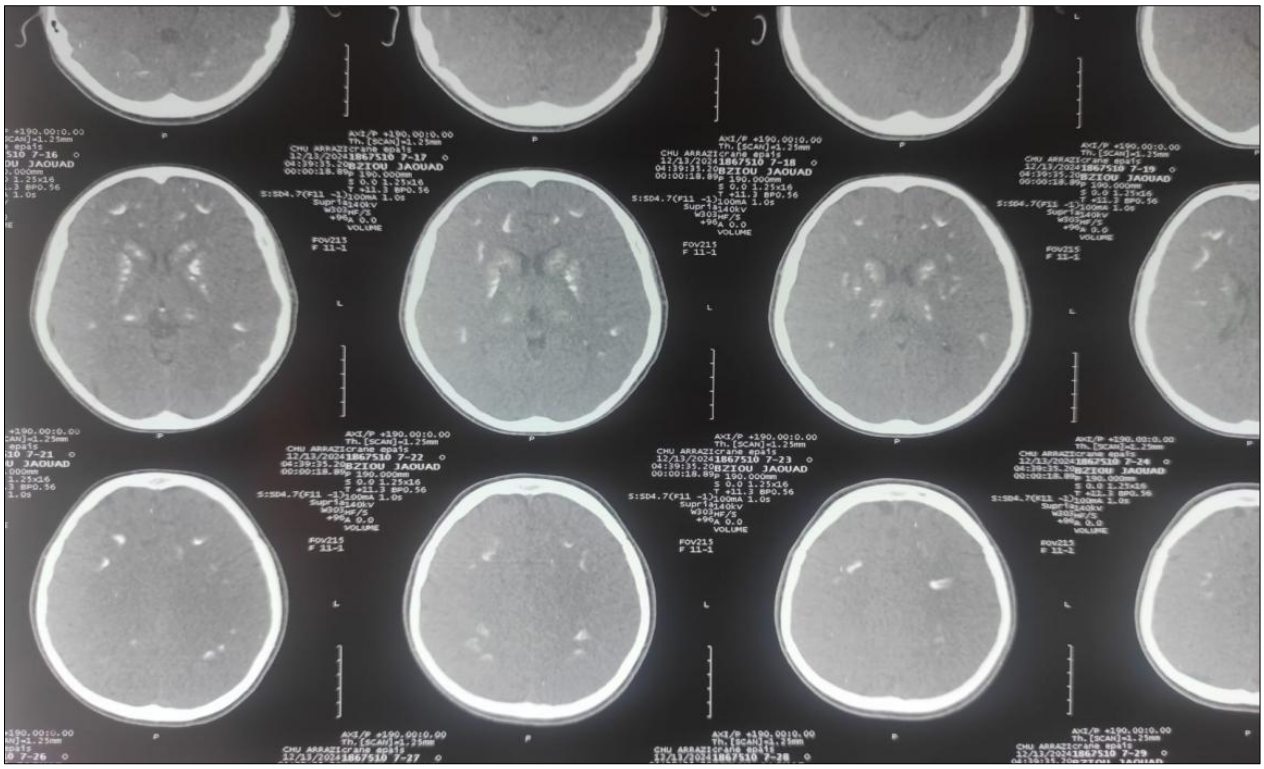

**Figure 1: Computed tomography scan of the head showing deep, bilateral, and symmetrical calcifications located in both supra- and infratentorial regions**

**Management and Outcome**

The patient initially received intravenous correction of hypocalcemia. Following stabilization of the clinical condition and improvement in laboratory parameters, oral therapy was initiated, including:

- Calcium: 2 g/day
- Alfa-calcidiol: 1 µg/day

The clinical and biochemical evolution was favorable, with improvement in neurological symptoms and stabilization of calcium levels. The therapeutic objective is to maintain calcium levels at the lower limit of normal, requiring regular follow-up to adjust treatment and prevent recurrence.

## DISCUSSION

Fahr syndrome (FS) is a rare pathology whose etiology is dominated by disorders of phosphocalcium metabolism, in particular dysparathyroidism [7]. Bilateral and symmetrical cerebral calcifications, characteristic of FS, are the result of chronic hypocalcemia favoring calcium deposits in the small vessels and cerebral parenchyma. Once formed, these calcifications are irreversible and can lead to a variety of neurological complications, such as seizures, motor disorders and cognitive impairment [8].

PHP, as an underlying cause of FH, is a rare genetic disorder. PHP subtype 1A, linked to a mutation

in the GNAS gene, often manifests with clinical features such as a lunar facies, short stature and brachymetacarpia. This subtype is also associated with multiple hormone resistances, notably to TSH [9]. In contrast, subtype 1B, linked to an epigenetic anomaly, is limited to isolated PTH resistance with no phenotypic abnormalities [10]. Subtype 2, less well characterized, presents resistance to PTH without identified genetic abnormalities [11].

In reported cases, the diagnosis of PHP was delayed due to the initial presentation as convulsive seizures, often attributed to idiopathic epilepsy. This diagnostic delay underlines the importance of looking for metabolic abnormalities in patients presenting with unexplained intracranial calcifications or atypical neurological symptoms [12].

Treatment of PHP is based on correction of hypocalcemia by supplementation with calcium and active vitamin D (1-alpha-hydroxyvitamin D or calcitriol). Early management can prevent the progression of cerebral calcifications and improve neurological symptoms [13]. However, calcifications that have already formed are irreversible, underscoring the importance of rapid diagnosis and rigorous follow-up. In addition, the multiple hormone resistances observed in certain subtypes of PHP call for multidisciplinary management involving endocrinologists, neurologists and radiologists [14].

Delayed diagnosis of FS-associated PHP is a major clinical challenge. Seizures, often attributed to idiopathic epilepsy, mask underlying metabolic abnormalities. A systematic diagnostic approach, including biological assays (calcium, phosphate, PTH) and brain imaging, is needed to identify these complex cases [15]. Recognition of specific phenotypic signs, such as those seen in PHP 1A subtypes, can also guide the diagnosis.

Although treatment can improve neurological and metabolic symptoms, long-term prognosis depends on early diagnosis and management. Irreversible cerebral calcifications can lead to persistent neurological sequelae. Regular follow-up is therefore essential to adjust treatment and monitor the development of complications [16].

Advances in our understanding of the genetic and epigenetic mechanisms of PHP open up prospects for earlier diagnosis and targeted treatments. In addition, further research into the mechanisms of cerebral calcification could lead to the development of preventive strategies [17].

## CONCLUSION

The association between FH and PHP is rare but clinically significant. These cases underline the importance of early diagnosis and multidisciplinary management involving neurologists, endocrinologists and radiologists. In the presence of intracranial calcifications or unexplained neurological symptoms, it is essential to look for metabolic abnormalities, particularly hypocalcemia and hyperphosphatemia. Appropriate management can improve prognosis and limit long-term complications.

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