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Case Report

Fahr Disease: A Case Report in Radio-Pediatric Field

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Abstract

Fahr syndrome is a rare neurodegenerative disorder characterized by abnormal bilateral calcifications in the basal ganglia and other brain regions, including the cerebellum and cerebral cortex. It may occur secondary to metabolic abnormalities, particularly disorders of calcium-phosphorus metabolism such as hypoparathyroidism, or as a primary familial condition. Clinical manifestations are heterogeneous and may include movement disorders, cognitive impairment, psychiatric symptoms, seizures, and speech disturbances. Brain computed tomography is the imaging modality of choice, typically revealing symmetrical intracranial calcifications. Management is mainly symptomatic and directed toward treating the underlying metabolic cause when identified. Early diagnosis is important to improve clinical outcomes and prevent disease progression.

Keywords: *Fahr Syndrome; Idiopathic Basal Ganglia Calcification; Intracranial Calcifications; Neuropsychiatric Manifestations*

Introduction

Fahr syndrome, also known as Fahr's disease, is a rare neurodegenerative disorder characterized by bilateral and abnormal intracranial calcifications involving the basal ganglia and other cerebral structures [1]. These calcified deposits, mainly composed of calcium phosphate and calcium carbonate, typically affect the basal ganglia, thalamus, hippocampus, cerebral cortex, cerebellar subcortical white matter, and dentate nucleus [1].

Fahr syndrome, first described by Karl Theodor Fahr in 1930, is radiologically characterized by bilateral and symmetrical non-arteriosclerotic calcifications involving the striatum, globus pallidus, and dentate nuclei [2].

Fahr syndrome is a rare neurological entity characterized by bilateral and symmetrical intracerebral calcifications involving the basal ganglia, thalamus, dentate nucleus, and centrum semi-ovale, occurring in the absence of metabolic abnormalities, particularly hypoparathyroidism [3].

Computed tomography is the imaging modality of choice for detecting and evaluating the extent of cerebral calcifications, which predominantly involve the lenticular nucleus, particularly the globus pallidus [3].

In the literature, the terms Fahr disease and Fahr syndrome are often used interchangeably; however, Fahr disease generally refers to primary idiopathic basal ganglia calcifications, whereas Fahr syndrome designates secondary forms associated with an identifiable underlying cause [4].

The clinical manifestations are highly variable, and many patients remain asymptomatic for long periods. When symptomatic, the condition may progress to include parkinsonism, various movement disorders such as chorea or dystonia, as well as headache, seizures or epilepsy, psychiatric disturbances including psychosis and depression, and progressive cognitive decline [5,6].

Fahr disease is a progressive disorder without curative treatment, and management is mainly symptomatic and supportive. Clinical severity is generally associated with the extent of intracranial calcifications [7].

Case Presentation

We report the case of a pediatric patient referred to the Department of Pediatric Radiology for evaluation of progressive neurological symptoms. The patient was a 9 year old child presenting with a history of recurrent generalized seizures, mild psychomotor delay, and recent onset of behavioral changes and decreased school performance. No history of head trauma, infection, or toxin exposure was reported.

Neurological examination revealed mild cognitive impairment without focal neurological deficits. There were no clinical signs suggestive of movement disorders such as parkinsonism, dystonia, or chorea at the time of presentation.

Laboratory investigations were performed to exclude secondary causes of intracranial calcifications. Serum calcium, phosphate, magnesium, and alkaline phosphatase levels were within normal limits. Importantly, parathyroid hormone (PTH) levels were normal, ruling out hypoparathyroidism and other calcium-phosphate metabolism disorders. Thyroid function tests and renal function were also unremarkable. Infectious and toxicological work-up was negative.

Brain computed tomography (CT) revealed bilateral, symmetrical, and extensive calcifications involving the basal ganglia (predominantly the globus pallidus), thalami, and dentate nuclei, with no associated mass effect or edema (See figure 1-4).

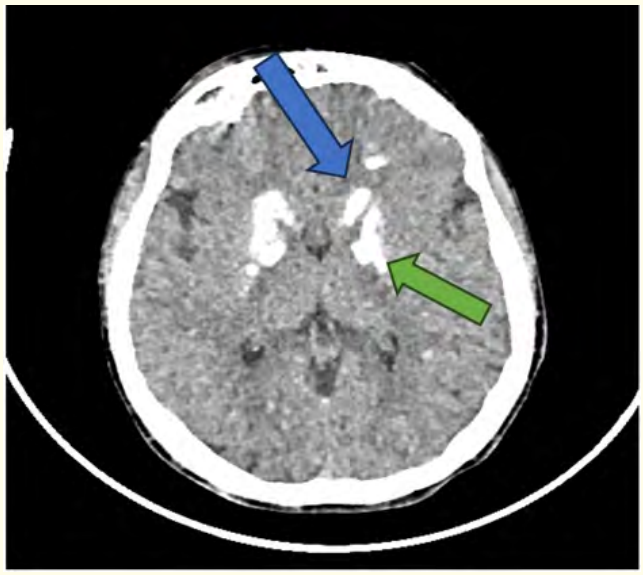

Figure 1: *Axial non enhanced head Ct scan displaying bilateral, symmetrical calcification within caudate nucleus (Blue arrow) and lenticular nucleus (Green arrow).*

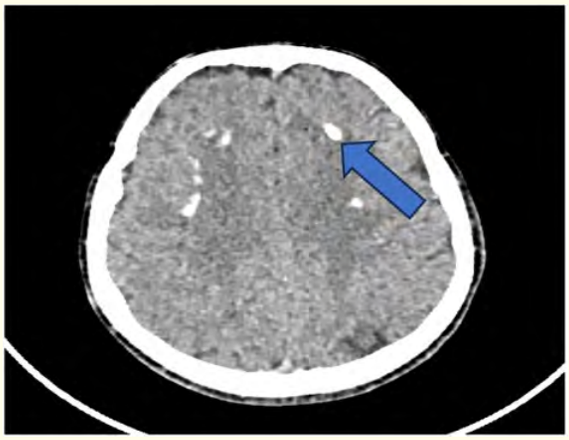

Figure 2: *Axial head Ct scan C-, displaying bilateral and symmetrical calcification in the deep white matter of centrum semi-oval (Blue arrow).*

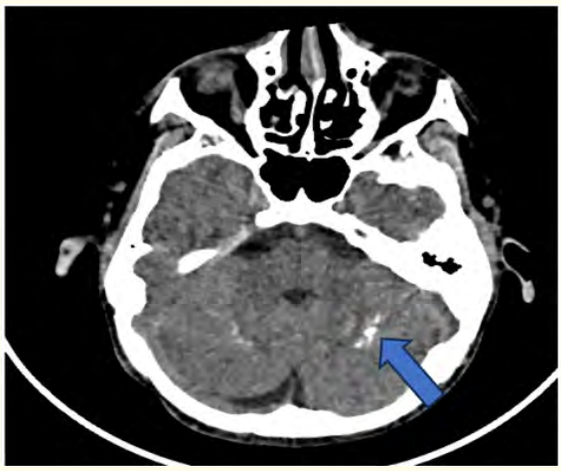

Figure 3: *Axial non enhanced Ct scan displaying bilateral and symmetrical calcification in the deep white matter of the cerebellar hemisphere, Consistent with involvement of the dentate nuclei (Blue arrow).*

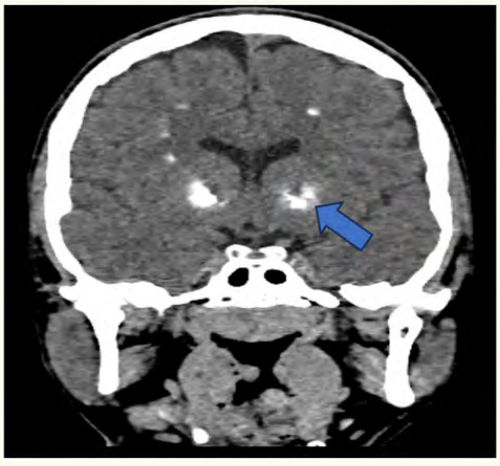

Figure 4: *Coronal non enhanced Ct scan displaying bilateral calcification within the lenticular nucleus, and deep white matter of centrum semi-oval (Blue arrow).*

Given the radiological findings and the exclusion of metabolic, infectious, and toxic causes, the diagnosis of Fahr disease was retained.

Discussion

Fahr syndrome is a rare neurodegenerative condition characterized by bilateral and symmetrical intracranial calcifications, primarily involving the basal ganglia, dentate nucleus, and cerebral cortex. The increasing availability and widespread use of advanced neuroimaging techniques have significantly contributed to higher detection rates of these calcific changes [7]. It was initially described in 1930 by the German neurologist Karl Theodor Fahr [8].

Fahr disease most commonly presents in middle-aged adults, typically in the fourth to fifth decades of life, following an asymptomatic early period. It is a progressive neurodegenerative disorder whose true prevalence remains uncertain due to underdiagnosis and limited screening of first-degree relatives [4]. In contrast to this usual epidemiological profile, our case involves an atypical presentation in a 9-year-old child, highlighting the rare pediatric occurrence of the disease.

Fahr disease is a rare neurodegenerative disorder characterized by progressive bilateral and symmetrical intracerebral calcifications, mainly involving the basal ganglia, dentate nuclei, thalami, and cerebral cortex. These calcifications are thought to result from disturbances in calcium-phosphate metabolism and impaired blood-brain barrier integrity, leading to abnormal mineral deposition within perivascular spaces and neuronal tissue. Over time, this process disrupts neuronal circuits involved in motor control, cognition, and behavior [9].

Clinically, the disease is typically described in adults, where it manifests with a broad spectrum of neurological and psychiatric symptoms, including movement disorders (parkinsonism, chorea, dystonia), cognitive decline, seizures, and behavioral or psychiatric disturbances, symptom severity often correlates with the extent and distribution of intracranial calcifications [9].

In contrast, our case demonstrates an exceptionally early presentation in a 9-year-old child, which is highly uncommon for this condition. The patient presented mainly with seizures and early neurodevelopmental/cognitive impairment, without the full spectrum of movement disorders usually described in adult cases. This pediatric presentation emphasizes that, although classically considered an adult-onset disorder, Fahr disease can rarely manifest early in life, with a clinical picture dominated by epileptic and cognitive symptoms rather than extrapyramidal signs.

This pathology is a rare neurological condition with few cases reported in the literature, particularly in the pediatric population. Its underlying pathophysiology and etiologies are not yet fully elucidated, and no specific curative treatment is currently established [10].

In children, Fahr syndrome may present with severe encephalopathy associated with growth delay, microcephaly, and optic atrophy; however, these features were absent in our 9-year-old patient [11].

The diagnosis of Fahr syndrome is established on a combination of strict clinical, radiological, and exclusion criteria. First, neuroimaging must demonstrate bilateral and symmetrical calcifications of the basal ganglia, which may extend to other regions such as the cerebellum or cerebral cortex, second, patients typically exhibit progressive neurological impairment, including movement disorders (e.g. parkinsonism, dystonia, chorea) and/or neuropsychiatric symptoms, with onset ranging from childhood to adulthood, third, laboratory evaluation must confirm the absence of biochemical abnormalities, particularly normal calcium-phosphate metabolism and no evidence of mitochondrial or other systemic metabolic disorders. Fourth, the diagnosis requires the exclusion of secondary causes, including infectious, toxic, or traumatic etiologies that could explain intracranial calcifications, finally, a positive family history with autosomal dominant inheritance supports a primary genetic form of the disease, reinforcing the diagnosis [1].

The differential diagnosis of Fahr disease includes several conditions associated with basal ganglia calcifications that must be excluded, such as endocrine disorders (hypoparathyroidism, pseudohypoparathyroidism), mitochondrial diseases, congenital syndromes (Cockayne, Aicardi-Goutières), and congenital infections (CMV, rubella, toxoplasmosis, herpes) [1].

Computed tomography (CT) is considered the gold standard imaging modality for diagnosing Fahr syndrome, as it clearly demonstrates intracerebral calcifications. It typically reveals multiple bilateral and symmetrical hyperdensities involving the brain parenchyma, predominantly affecting the basal ganglia, thalami, periventricular regions, and cerebellar nuclei [12].

In our 9-year-old patient, CT imaging similarly showed bilateral symmetrical calcifications, mainly involving the basal ganglia, confirming the typical radiological pattern of the disease. However, the distribution in our case was more localized, without significant extension to the cerebellar nuclei or widespread cortical involvement, highlighting a relatively limited but characteristic pediatric presentation.

Consequently, computed tomography (CT) remains the imaging modality of choice for diagnosing Fahr syndrome, as it is more sensitive than MRI in detecting intracranial calcifications. Electroencephalography has limited diagnostic value due to its non-specific findings. Therefore, the diagnosis ultimately relies on a combination of careful neurological assessment and CT imaging as the key investigative tools [11].

Management of Fahr disease remains purely symptomatic, as no curative therapy is currently available to halt or reverse disease progression. Treatment is therefore tailored to clinical manifestations, including the use of antiparkinsonian agents, antidepressants, and antipsychotic medications according to the neurological and psychiatric symptoms presented by the patient [13].

Although disease progression continues over time, therapeutic strategies aim to improve quality of life and functional outcomes. Emerging insights into the genetic and pathophysiological mechanisms may open new therapeutic avenues in the future, and bisphosphonate therapy has been suggested as a potential option in ongoing research [13].

Conclusion

Fahr disease is a rare neurodegenerative disorder that is exceptionally uncommon in the pediatric population, where it may present with nonspecific neurological manifestations such as seizures and cognitive or behavioral impairment. The diagnosis in children is particularly challenging due to its rarity, atypical clinical presentation, and the need for extensive exclusion of secondary causes of intracranial calcifications. In our experience within the pediatric radiology department, CT imaging played a pivotal role in establishing the diagnosis by revealing characteristic bilateral symmetrical basal ganglia calcifications.

This case highlights several diagnostic challenges, including low clinical suspicion in children, the broad differential diagnosis of intracranial calcifications, and the necessity of comprehensive metabolic, infectious, and genetic work-up to exclude secondary causes. Early recognition remains essential to avoid unnecessary investigations and to ensure appropriate symptomatic management. Overall, pediatric cases of Fahr disease underscore the importance of correlating clinical, biological, and radiological findings, while also emphasizing the need for further research to better understand its early-onset forms and underlying mechanisms.

Bibliography

1. Saleem S., *et al*. “Fahr’s syndrome: literature review of current evidence”. *Orphanet Journal of Rare Diseases* 8.1 (2013): 156.

2. Sbai H., *et al*. “Syndrome de Fahr découvert à la suite d’une méningite à pneumocoque”. *La Revue de Médecine Interne* 29.5 (2008): 412-414.

3. Gligorievski A. “CT Diagnosis of Fahr disease, a case report”. *Clinical Case Reports and Reviews* 4.2 (2018): 1-3.

4. Amisha F and Munakomi S. “Fahr Syndrome”. StatPearls Publishing (2020).

5. Perugula ML and Lippmann S. "Fahr's disease or Fahr's syndrome?". *Innovations in Clinical Neuroscience* 13.7-8 (2016): 45-46.

6. Batla A., *et al*. "Deconstructing Fahr's disease/syndrome of brain calcification in the era of new genes". *Parkinsonism and Related Disorders* 37 (2017): 1-10.

7. Balck A., *et al*. "Genotype-phenotype relations in primary familial brain calcification: systematic MDS Gene review". *Movement Disorders* 36.11 (2021): 2468-2480.

8. Fahr TV. "Verkalkung der Hirngefasse". *Zentralblatt für Allgemeine Pathologie und Pathologische Anatomie* 50 (1930): 129.

9. Rafai MA., *et al*. "Le syndrome de Fahr: aspects cliniques, radiologiques et étiologiques". *Feuillets de Radiologie* 54.1 (2014): 2-8.

10. Kassal G., *et al*. "Unveiling Fahr's syndrome in a child: A case linked to congenital hypoparathyroidism". *Cureus* 17.5 (2025): e84001.

11. Jaworski K., *et al*. "Fahr syndrome-an important piece of a puzzle in the differential diagnosis of many diseases". *Polish Journal of Radiology* 82 (2017): 490-493.

12. Ahad MA., *et al*. "Fahr's syndrome". *Bangladesh Medical Journal Khulna* 45.1-2 (2012): 33-35.

13. Peters ME., *et al*. "Mechanisms of calcification in Fahr disease and exposure of potential therapeutic targets". *Neurology: Clinical Practice* 10.5 (2020): 449-457.

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