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Incidental Detection of Familial Fahr's Disease Following a Traffic Accident - A Case Series

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Abstract

Objective: To present a case series of asymptomatic patients in whom Fahr's disease was incidentally identified during routine CT imaging after a car accident.

Methods: We report three asymptomatic individuals a 28-year-old mother and her two children (aged 8 and 5) who presented to the emergency department following a car accident. All underwent non-contrast cranial CT as part of standard trauma protocol. Neurological examinations were non-specific, with no clinical signs of neurological dysfunction.

Results: CT imaging revealed symmetrical bilateral calcifications in the basal ganglia in all three patients. In the mother, additional calcifications were present in the caudate nucleus. No evidence of acute traumatic brain injury was found. There was no known family history of neurological disease. Based on the characteristic calcification pattern and familial clustering, a presumptive diagnosis of familial Fahr's disease was made. Further evaluation, including calcium-phosphorus metabolism testing and genetic screening, was recommended.

Conclusion: These cases demonstrate that routine trauma imaging can incidentally reveal undiagnosed familial Fahr's disease in asymptomatic individuals. Recognition of the typical radiological features allows for early diagnosis, clinical follow-up, and consideration of genetic counseling for affected individuals and their families.

Keywords: Fahr disease; Computed tomography; incidental findings

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Introduction

Primary Familial Brain Calcification (PFBC), historically known as Fahr's disease, is a rare neurodegenerative condition characterized by bilateral and symmetrical intracranial calcifications, most prominently affecting the basal ganglia, thalami, dentate nuclei, and subcortical white matter. The disorder arises from abnormalities in genes regulating phosphate transport and vascular integrity most commonly SLC20A2, PDGFB, PDGFRB, or XPR1-resulting in pathological mineral deposition within neuronal and vascular structures [1]. The pathophysiology reflects a complex interplay between genetic predisposition and disturbances in local calcium-phosphate homeostasis, although the exact mechanisms remain incompletely understood.

PFBC exhibits a wide clinical spectrum. While some individuals develop movement disorders, neuropsychiatric manifestations, cognitive impairment, or seizures, a substantial proportion remain entirely asymptomatic throughout life. Radiologic abnormalities often precede clinical symptoms by decades, and incidental findings on neuroimaging are increasingly common due to the widespread use of Computed Tomography (CT) in emergency and trauma settings [1-4]. Importantly, the burden of calcification does not reliably correlate with clinical severity, complicating efforts to predict symptom onset or progression.

Diagnosis relies heavily on non-contrast CT, which remains the most sensitive modality for detecting intracranial calcifications. The characteristic distribution and symmetry of hyperdense foci guide clinicians toward a diagnosis of PFBC; however, secondary causes of calcification including metabolic disorders, endocrine abnormalities, infectious etiologies, and toxic exposures must be excluded to avoid misclassification [1-4]. Once secondary etiologies are ruled out, a family history of similar findings or symptoms strengthens the suspicion of an inherited form, although de novo mutations may also occur.

The growing availability of advanced imaging has led to increased recognition of familial clusters identified incidentally, particularly during evaluations unrelated to neurological disease. In such

cases, radiologists play a central role in initiating appropriate follow-up, recommending metabolic workup, and guiding genetic counseling. In this context, we present a case series of three asymptomatic family members a young mother and her two children in whom PFBC was incidentally detected during routine post-trauma CT imaging. This cluster highlights the importance of recognizing the classic radiologic pattern of PFBC, even in patients without neurological symptoms, and underscores the value of early identification for long-term management and family evaluation.

Case Presentation

Three asymptomatic individuals a 28-year-old mother and her two children (aged 8 and 6) presented to the emergency department following a traffic accident. All underwent non-contrast brain CT as part of the trauma protocol. Neurological examination in all cases was nonspecific. CT findings demonstrated symmetrical bilateral calcifications in the basal ganglia across all patients, with additional calcifications in the caudate nucleus in the mother. No acute traumatic brain injury, intracranial hemorrhage, mass effect, midline shift, or skull fracture was detected (Figures 1-8).

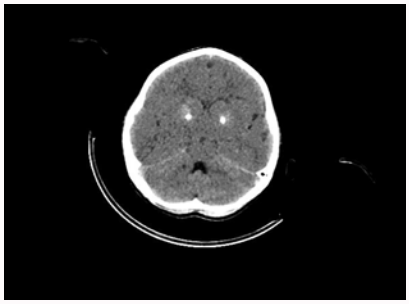

Figure 1: Axial CT of Patient 1 showing bilateral globus pallidus calcifications.

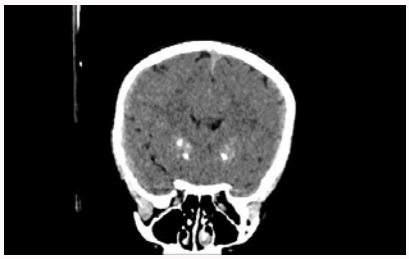

Figure 2: Coronal CT of Patient 1 demonstrating symmetric basal ganglia calcifications.

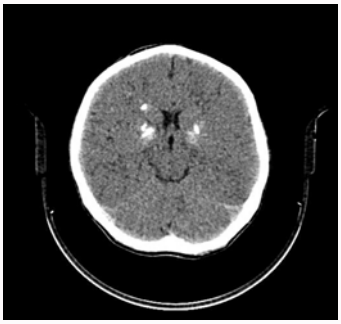

Figure 3: Axial CT of Patient 2 showing bilateral globus pallidus calcifications.

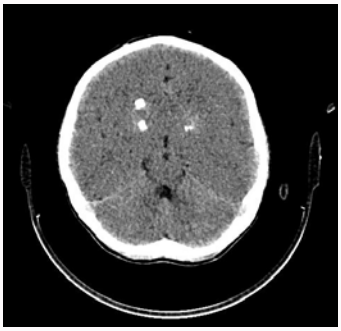

Figure 4: Additional axial CT of Patient 2 showing deep gray nuclei calcifications.

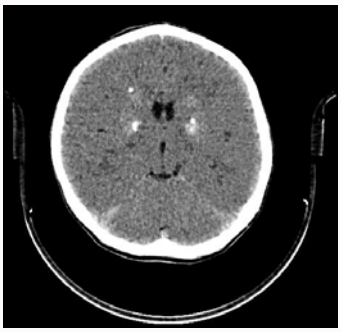

Figure 5: Further axial CT slice of patient 2.

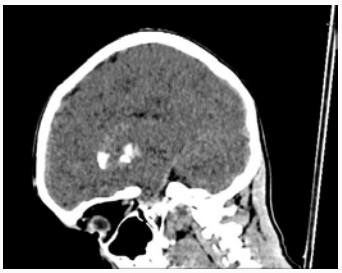

Figure 6: Sagittal CT of Patient 2 demonstrating calcifications.

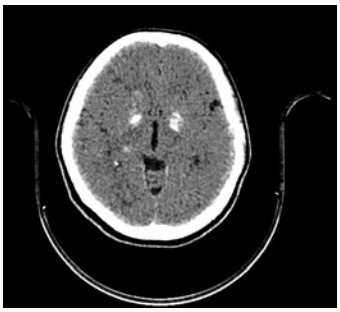

Figure 7: Axial CT of Patient 3 showing bilateral basal ganglia calcifications.

Discussion

PFBC is an uncommon but increasingly recognized condition, largely due to the widespread use of neuroimaging in emergency departments and trauma evaluations. The disorder is classically defined by bilateral, symmetric calcifications of the basal ganglia, often extending to additional deep gray and cerebellar structures.

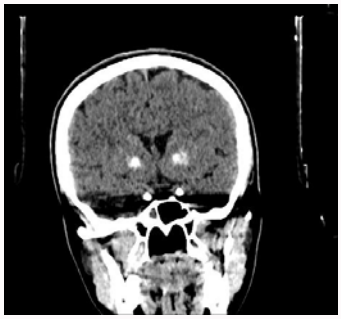

Figure 8: Coronal CT of Patient 3 showing calcifications.

These findings arise from genetic alterations affecting phosphate transport or platelet-derived growth factor signaling, leading to progressive mineral deposition within microvascular and neuronal tissues [1,2]. Although PFBC has traditionally been associated with neurological or psychiatric manifestations, recent studies indicate that a significant proportion of affected individuals remain asymptomatic, with incidental detection becoming more common across diverse clinical settings [1,2].

In the present case series, all three patients—despite their young ages—demonstrated radiologic features characteristic of PFBC, yet none exhibited neurological deficits. This aligns with existing literature highlighting that radiologic calcification frequently precedes the onset of symptoms and may remain clinically silent for years. Several reports have documented asymptomatic individuals identified during unrelated imaging, including head trauma evaluations or preoperative assessments, reinforcing the importance of radiologist familiarity with PFBC's imaging hallmarks [4]. The familial clustering observed in our series further supports an inherited etiology, most likely autosomal dominant, although definitive genetic testing would be required for molecular confirmation.

CT remains the gold standard for detecting intracranial calcifications due to its excellent sensitivity to calcium. In PFBC, CT typically demonstrates sharply defined hyperdensities in the globus pallidus, putamen, and caudate nuclei. MRI may provide complementary information regarding associated neurodegenerative changes but is comparatively less effective in visualizing calcifications. The distribution and symmetry of the findings in our patients were highly characteristic and helped exclude alternative etiologies such as metabolic disorders, prior infections, or toxic exposures. Nevertheless, a comprehensive metabolic evaluation including serum calcium, phosphate, magnesium, and parathyroid hormone levels—is recommended to rule out secondary causes before confirming PFBC [4].

Although our patients were asymptomatic, recognition of this condition carries important implications. First, early diagnosis enables appropriate counseling regarding potential future symptoms, which may include motor disturbances, psychiatric manifestations, or seizures. Second, identification of radiologic PFBC in multiple family members supports referral for genetic testing, aiding in prognostication and family planning [5-7]. Third, awareness of the diagnosis can guide clinical decision-making in the event of future neurological symptoms, preventing unnecessary investigations and providing a clear diagnostic framework.

Our findings underscore the crucial role of radiologists in identifying PFBC, particularly when calcifications are discovered incidentally. Familiarity with the characteristic imaging pattern enables timely recognition, even in the absence of symptoms. As illustrated by these cases, routine trauma imaging can uncover significant underlying conditions with long-term clinical implications. Early recognition encourages appropriate follow-up, facilitates genetic evaluation, and ensures that patients and families receive accurate counseling regarding prognosis and potential symptom development.

Conclusion

Routine trauma CT imaging can incidentally reveal undiagnosed familial Fahr's disease in asymptomatic individuals. Radiologists should remain aware of characteristic imaging features suggestive of this condition, enabling timely clinical evaluation and genetic counseling.

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