# Electrolyte abnormalities and seizure like activity unmasking an underlying rare genetic disorder

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## INTRODUCTION

Fahr's syndrome, or idiopathic basal ganglia calcification, is a rare neurological disorder that runs in families as an autosomal dominant trait.

It is associated with a variety of other diseases, especially Parkinson's disease, but the etiology remains a mystery.

On imaging, it presents as abnormal calcium deposits found primarily in the basal ganglia and cerebral cortex, both of which are areas responsible for controlling movement.

## CASE PRESENTATION

A 73-year old woman with a past medical history of hypertension and seizures presented to the emergency department due to altered mental status.

The patient was indulged in a conversation with her daughter when all of a sudden, she became silent. Within a few seconds she became very stiff with arms extended, her head turned towards the left, and her eyes rolled towards the back of her head.

After unsuccessful re-orientation, EMS was called and upon awakening she became lethargic, confused, disoriented, and nonverbal. Her only further complaint was that she felt numbness to her lower and upper extremities.

On further questioning it was revealed that about 20 years ago, the patient was advised to take medication for hypocalcemia, but was non-compliant.

## IMAGING & RESULTS

Initial lab findings revealed critically low calcium and parathyroid hormone levels, and normal vitamin D levels.

Computed Tomography (CT) head/brain revealed extensive intracranial calcification and mineralization (Figure 1). Subsequently, Magnetic Resonance Imaging of head/brain was completed which revealed abnormal signals in the bilateral basal ganglia, thalami, and cerebellum corresponding to calcifications seen on CT scan (Figure 2). EEG was also done given her underlying seizure.

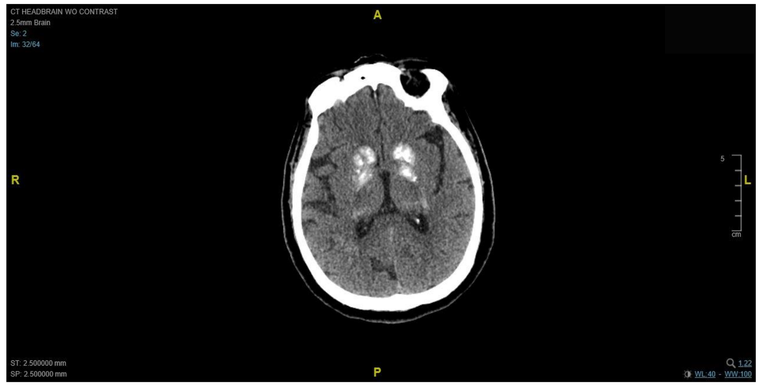


Figure 1: CT Head/Brain without contrast

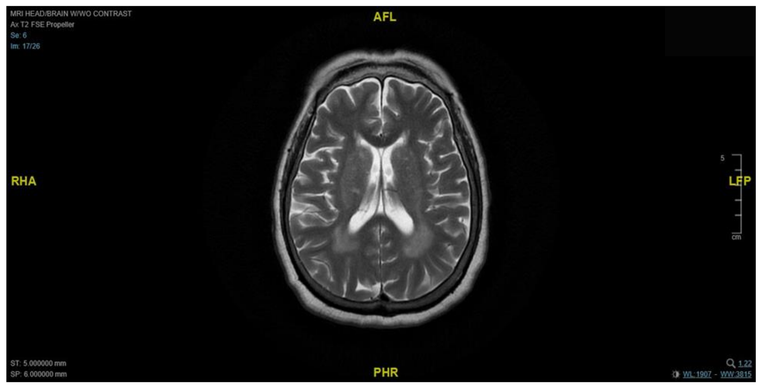


Figure 2: MRI Head/Brain w/wo contrast

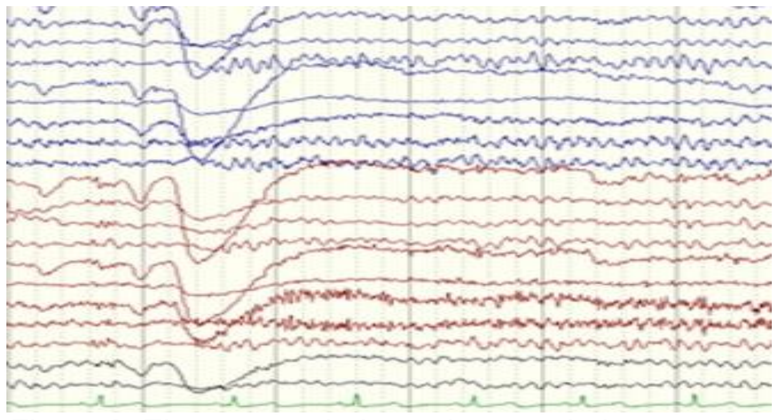

Figure 3: Electroencephalography revealed some bifrontal fast activity which constitutes a characteristic electrophysiological pattern in focal seizures of human epilepsy.

## HOSPITAL COURSE

Based on the workup, it was suggested that these findings were concerning for Fahr's syndrome, which was secondary to the underlying hypoparathyroidism.

When the electrolyte abnormalities were corrected and the patient returned to baseline, she was determined stable for discharge.

At discharge, the patient was continued on calcium and Vitamin D supplements, and Levetiracetam 500mg twice daily for seizure prophylaxis. She was also advised to see an endocrinologist for further management of her hypoparathyroidism.

## DISCUSSION

This case illustrates that a rare disorder like Fahr's syndrome has a similar presentation to various other neurological diseases, but no etiological agent has yet been established.

The presence of certain clinical characteristics that may cause the diagnosis to be confused with other illnesses is required for diagnosis.

To reduce the loss of functionality associated with the condition, new therapeutic approaches must be identified and implemented.

Successful therapeutic interventions can only be established upon identification of the underlying etiology.

Additionally, it is critical to stress the importance of genetic counseling for known at-risk parents before conception.

## REFERENCES & DISCLOSURES

No disclosures

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*This research was supported (in whole or in part) by HCA Healthcare and/or an HCA Healthcare affiliated entity. The views expressed in this publication represent those of the author(s) and do not necessarily represent the official views of HCA Healthcare or any of its affiliated entities.*