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Secondary Fahr's Syndrome in Patient with Chronic Renal Disease

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ABSTRACT

Background: Fahr's syndrome is an uncommon neurodegenerative condition marked by bilateral cerebral cortex and basal ganglia calcifications. It is believed that anomalies in calcium-phosphate balance are the cause of these deposits, which are composed of calcium and phosphorus.

Objective: This study aimed to investigate the best way for diagnosis of Fahr's syndrome and the association between Fahr's syndrome and disordered calcium-phosphorous metabolism.

Subject and methods: Female patient 37-year-old with chronic kidney disease had chest infection and septic shock complicated by acute renal failure requiring hemodialysis. The patient was admitted at El Ahrar teaching hospital, for hemodialysis and correction of infection.

Result: Neuropsychiatric symptoms, cognitive decline, extrapyramidal symptoms, and partial or generalized seizures are among the clinical manifestations. In this case study, we examined a middle-aged woman who had a history of chronic kidney illness and was discovered to have bilateral, symmetrical calcifications in her cerebral cortex and basal ganglia. Her lab tests revealed disordered calcium-phosphorus balance and hyperparathyroidism. Unlike Fahr's syndrome, Fahr's disease doesn't have impaired phosphocalcic metabolism.

Conclusion: Our study showed the association between Fahr's syndrome and disordered calcium-phosphorous metabolism as in hyperparathyroidism secondary to chronic renal disease. Although, there were many different clinical characteristics, the diagnosis is initially made primarily on the presence of bilateral symmetrical calcifications in the gray matter and impairment of phosphocalcic metabolism.

Keywords: Fahr's syndrome, Calcium, Calcifications, Basal ganglia.

INTRODUCTION

Fahr's syndrome, a cause of bilateral basal ganglia and cerebral calcifications believed to be caused by a number of endocrine disorders, genetic disorders, infections, or toxins. This can be revealed on computed tomography (CT) or magnetic resonance imaging (MRI) **(1).** The most common is calcium and phosphorus homeostasis. Hypoparathyroidism and these bilateral calcifications are linked in numerous case reports. Hyperparathyroidism is less frequently linked to this association (2). It is believed that these metabolic disorders, which result in an improper calcium/phosphorous ratio, cause colloids precipitation and calcified deposits formation in cerebral vessels. These bilateral calcifications are frequently asymptomatic and are discovered on brain imaging.

Neuropsychiatric signs such as cognitive impairment, intellectual incapacity, extrapyramidal abnormalities, psychiatric problems, worsening of motor function, stroke-like episodes, spastic paralysis, and infrequently seizures can identify Fahr's syndrome **(1)**. In contrast to Fahr's disease, an autosomal dominant condition brought on by BGC1 gene mutations, Fahr's syndrome causes phosphocalcic metabolism impairment which is absent in Fahr's disease **(2)**.

CASE PRESENTATION

Female patient 37-year-old with chronic kidney disease had chest infection and septic shock complicated by acute renal failure requiring hemodialysis. The patient was admitted at El Ahrar Teaching Hospital, for hemodialysis and correction of infection. Few days after, the patient had disturbed conscious levels, fainting with history of muscle weakness, some intellectual disabilities and behavioral disorders. Neurological consultation asked for CT and MRI brain.

CT imaging of the brain didn't reveal signs of acute infarction. On the other hand, it displayed symmetrical bilateral extensive calcifications affecting the thalami and bilateral basal ganglia (Figure 1). In the bilateral cerebral white matter, linear calcifications were observed. Fahr's disease or syndrome was supported by these results.

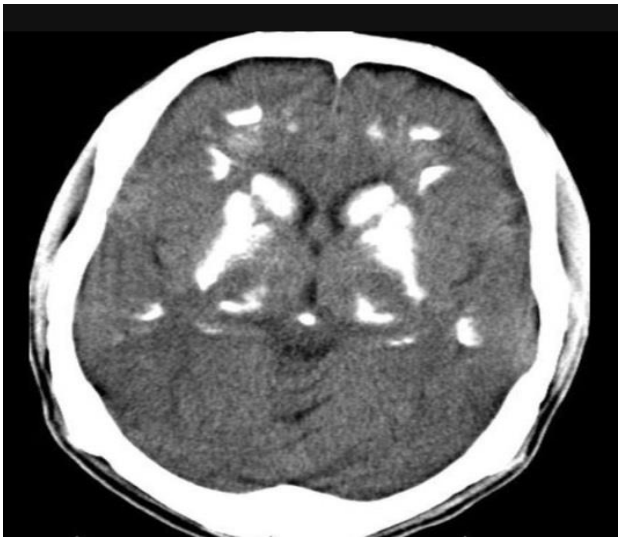

Figure (1): CT brain without contrast showing bilateral, symmetrical basal ganglia, thalami and cerebral white matter calcifications.

MRI of the brain showed basal ganglia calcifications of signal loss at SWI (Figure 2) and abnormal diffuse peri-ventricular signal that was corresponding to calcifications shown at CT.

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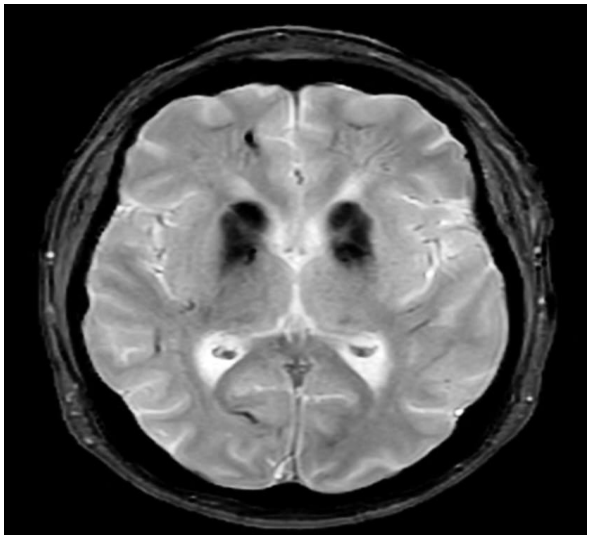

Figure (2): SWI sequence of MRI brain showed bilateral basal ganglia calcifications with signal loss.

The patient had a history of hypertension since nearly 10 years, which mostly was the cause of chronic renal disease, renal transplantation was done 11 years ago. Recently the patient was admitted as she had sepsis and anuria, which detects acute renal failure. Lab investigations revealed high parathyroid hormone (PTH) with elevated phosphorous level and low ionized calcium level indicative of secondary hyperparathyroidism, which is most likely brought on by long-term renal failure.

Bilateral calcifications have no discernible underlying etiology. There were no significant neurological or psychological abnormalities in the patient that would point to Fahr's syndrome. Fahr's disease did not run in her family either.

DISCUSSION

Fahr's syndrome typically manifests in middle age or adolescence and affects both sexes in the same proportion. Fahr's syndrome is frequently initially identified when bilateral intracerebral calcifications in the gray matter are seen on CT imaging. However, there are significant differences in the age at symptoms' onset and clinical presentations [(3)]**.** In younger individuals, basal ganglia calcifications are more problematic, even if they are a common idiopathic finding in older patients. Additionally, those accidental calcifications typically show no symptoms [(4)].

Generally speaking, the disease is clinically challenging to identify due to its wide and varied clinical presentation. Numerous clinical characteristics, including neurological symptoms, mobility difficulties, and neuropsychiatric symptoms, can be linked to Fahr's syndrome. Intellectual incapacity, mental debility, loss of consciousness, aberrant mobility, behavioral abnormalities, and occasionally delirious episodes are some of these symptoms [(5-7)]. When compared to the severity of anatomical and radiological abnormalities, neurological symptoms are frequently subtle. Some of these symptoms were seen in our patient, with her brain imaging findings and history, Fahr's syndrome was highly suggested.

These calcifications' pathophysiology and symptoms are believed to be associated with endocrine problems, including hypoparathyroidism or pseudohypoparathyroidism. It has also been linked to hyperparathyroidism, according to some reports [(7, 8)]. In our case, the gray matter calcifications is due to abnormal calcium/phosphate ratios caused by chronic renal failure. Likely extended time of disease, helps these calcifications develop insidiously till became symptomatic. Other illnesses, such as certain endocrine disorders, systemic illnesses, and brain infections, can also result in intracerebral calcifications, but they are usually not symmetrical, bilateral, or ideally located in the central gray nuclei [(3)].

Fahr's syndrome has an unpredictable prognosis. Research has not demonstrated a relationship between the degree of calcification and the severity of the illness [(8)]. This condition does not currently have a proven cure. The majority of patients receive symptomatic care [(9)].

CONCLUSION

Our study showed the association between Fahr's syndrome and disordered calcium-phosphorous metabolism as in hyperparathyroidism secondary to chronic renal disease. Although, there were many different clinical characteristics, the diagnosis was initially made primarily on the presence of bilateral symmetrical calcifications in the gray matter and impairment of phosphocalcic metabolism. Fahr's syndrome, which is secondary, is of earlier onset than Fahr's disease which is primary.

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