Case Report

Fahr's Disease a Rare calcific Neurodegenerative Disorder: A case report

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Abstract

Fahr's disease, also known as Fahr's syndrome is a rare neurodegenerative disorder mentioned by abnormal deposition of calcium in the bilateral basal ganglia and other regions of the brain. We report a case presented to us with detailed description diagnosed as Fahr's disease, the clinical presentation, diagnostic challenges, and treatment options.

Introduction

This patient, a 40 year female presented to the Neurology department with a history of cognitive decline, seizure episodes over the past eighteen months. There is no family history of any neurological disorders. Physical examination revealed bradykinesia, rigidity, and postural instability. The patient had a score of 23/30 on the Mini-Mental State Examination (MMSE), indicating mild cognitive impairment.

Diagnostic Assessment:

To investigate the etiology of the symptoms, a comprehensive diagnostic workup was initiated. Blood parameters and serology test were normal along with cardiac echocardiography which was normal. We did EEG which was reported as borderline abnormal. Serum ceruloplasmin level was normal and urine was assessed for color change on exposure to sunlight was normal in view of porphyria. Neuroimaging studies, including brain CT scan(CT number 4535), revealed extensive bilateral calcification within the basal ganglia, thalamus, cerebellum, and subcortical white matter. These findings were consistent with Fahr's disease.

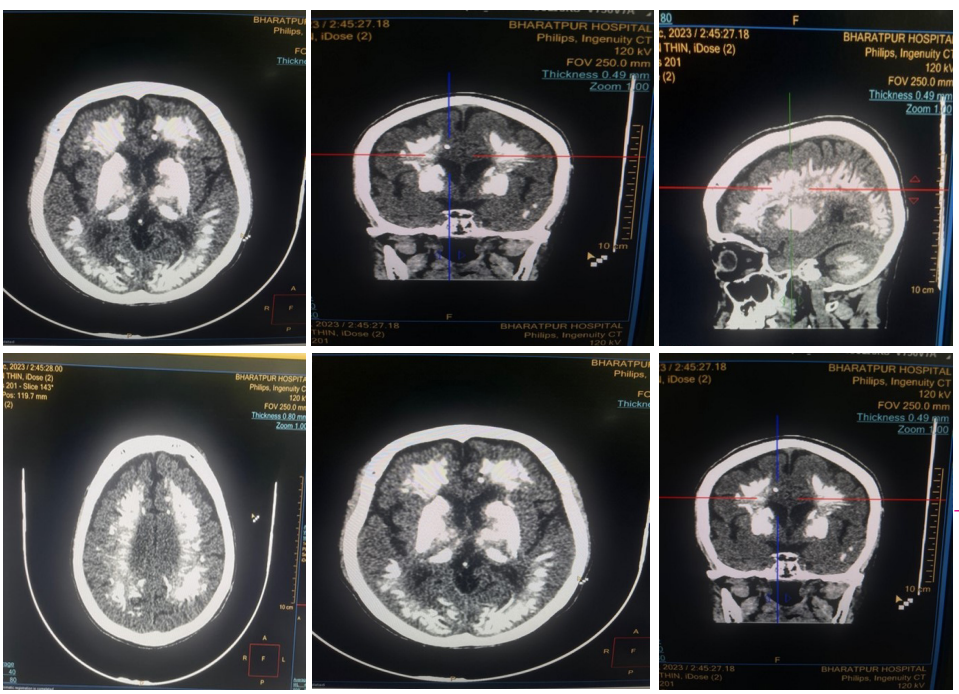

Fig: Fahr's disease (Striopallidodentate calcinosis). Axial non-contrast CT depicts bilateral symmetric calcifications in cerebellar foliae. Also note calcification in both dentate nuclei. There is extensive bilateral calcification in head of caudate nuclei, putamina and lateral globus pallidi with relative sparing of medial globus pallidi and thalami. There is calcification at grey–white matter junction as well. Linear calcification is seen to extend perpendicularly from the caudate nuclei.

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Discussion:

Fahr's disease is an autosomal dominant inherited disorder characterised by basal ganglia and extraganglionic calcification, extrapyramidal symptoms and psychosis[1]. Usual age of presentation is 40–60 years with no gender predilection(1) .Psychosis is proportionate to degree of calcification and cerebral atrophy. Calcification typically occurs in the lateral part of the globus pallidus, dentate nuclei and caudate nuclei(1-2). Normal calcium, phosphate and parathormone levels in Fahr's disease aid in diagnosis(1,2). Other differentials include senescent calcification (punctate, less extensive, occurring in the medial globus pallidus), HIV encephalopathy (cerebral atrophy, low CD4 counts), Cockayne syndrome (dwarfism, microcephaly, mental retardation, cerebral atrophy), and as sequelae to intrathecal chemotherapy and radiotherapy to the skull[2].

Differential diagnosis was challenging in this case, as Fahr's disease shares clinical features with other neurodegenerative disorders, such as Parkinson's disease, Huntington's disease, and Wilson's disease. Ocular examination was normal no evidence of KF rings. Serum ceruloplasmin levels was normal (30 mg/dl).

Non-contrast CT is the most sensitive modality and there is lack of enhancement in regions of calcification (2). Prognosis is poor since the disease is progressive and there is no specific treatment. Antipsychotics may be used; but patients with Fahr's disease are more susceptible to adverse effects such as malignant neuroleptic syndrome.[3]

There is currently no specific treatment for Fahr's disease. Management mainly focuses on symptomatic relief and supportive care. In this case, the patient was prescribed levodopa/carbidopa and leveracitam for seizure, and for the management of parkinsonian symptoms. Physical and occupational therapy were also recommended to help maintain mobility and independence.

Conclusion

Fahr's disease is a rare neurodegenerative disorder characterized by diffuse calcium deposition in the brain. Early recognition and diagnosis of Fahr's disease are crucial to manage effectively and alleviate symptoms. Further research is needed for identifying and mapping the treatment .

We reported this case with an aim to increase awareness among healthcare professionals about Fahr's disease, its clinical manifestations, diagnostic challenges, and available treatment options.

Conflict of interests None.

Patient consent Obtained.

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