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FAHR'S DISEASE-A CASE REPORT

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ABSTRACT We report a case of Fahr's Disease in a 10-year-old girl presenting with generalized seizures, dysarthria and altered mental status. CT imaging showed bilateral, multiple, symmetrical calcifications in the basal ganglia, thalamus and cerebellum. Other causes for intracranial calcifications were ruled out. Most of the literature available on Fahr's disease is primarily based on signs and symptoms presenting well into adulthood or old age. This condition causing idiopathic calcification of the Striopallidodentate area has rarely been reported in the pediatric age group. The following case study highlights the uncommon occurrence of Fahr's disease in a child and the distinction between this condition and Fahr's syndrome- these conditions share the same name but there is a divergence in the definition and diagnostic criteria which has been discussed in this study.

KEYWORDS :

INTRODUCTION

Fahr's Disease, also known as Idiopathic Striopallidodentate Calcinosis (1) is a rare neurodegenerative disorder characterized by abnormal calcium deposits in the basal ganglia and surrounding parenchyma associated with cell loss primarily involving bilateral basal ganglia and dentate nuclei of the cerebellum with no other systemic or metabolic condition causing the intracranial mineralization. Usually this condition presents with psychiatric features and movement disorders (3), which has been reported in several studies. We report a case of Fahr's Disease presenting with seizures (epilepsy?) in a 10-year-old female child.

Case report

On 2nd December 2021 a 10-year-old female child, a known case of seizure disorder on epileptic drugs since the age of two years presented to the hospital with generalized tonic clonic seizure and altered mental state. The child also had dysarthria, linguistic regression. There was no focal neurological deficit. No significant family history was elicited. She was the first-born child with home delivery. Peripartum period was uneventful. On clinical examination the weight and height were below the 5th percentile. General examination did not yield any significant findings. Systemic examination including neurological examination was normal. The developmental milestones were abnormal for age. An IQ test revealed a score of 70 (assessment tool used?). the investigations revealed normal hematological parameters, Ionized serum Ca^{++}- 3.0 mg/dl, Total Ca^{++}- 9.6 mg/dl, serum phosphorus- 4.5 mg/dl and serum PTH- 30 pg/dl. Thyroid function tests were normal. The serum anti nuclear antibody workup for intrauterine infections and ELISA for HIV were negative. Mantoux test was negative. Urine for metachromatic granules and aminoacidurias were negative. ABG was done and found to be normal. Chest X ray and USG abdomen revealed no abnormalities. EEG showed features suggestive of generalized epilepsy. CT scan of the brain revealed symmetric, bilateral, extensive calcification in the basal ganglia, thalamus, dentate nucleus, subcortical white matter of the cerebral and cerebellar areas. Diagnostic criteria for this disease have been formulated after modifications from previous evidence and it consist of bilateral calcification of basal ganglia, progressive neurologic dysfunction, absence of biochemical abnormalities, absence of an infectious, traumatic or toxic cause and a significant family history, all of which was done to arrive at the final diagnosis(1)(6). The child's seizures were controlled by carbamazepine and she was put on regular outpatient follow up.

DISCUSSION

Fahr's disease was first noticed by German neurologist Karl Theodor Fahr. **Fahr's disease can be autosominal dominant inheritance or sporadic in nature. Recent genetic studies identified mutations in SLC20A2, a gene located in the IBGC3 region that leads to a loss of function of type III sodium-dependent phosphate transporter 2 (PiT2) as a major cause for inherited Fahr's disease**. It has a prevalence of <1/1,000,000. Genetic heterogeneity and an anticipatory effect have been observed. There is no prenatal or genetic test available for genetic counselling. (5) Some studies postulate that the condition maybe secondary to intrauterine infections (4). Patients suffering from this condition most commonly present with parkinsonism, paresis, dystonia, speech impairment, psychiatric conditions like depression, psychosis, and dementia (1). Other uncommon presentations include stroke like events and epileptic syncope (8). The child reported in this study neither had any neurological abnormalities nor psychiatric manifestations. It is postulated that calcifications in Fahr's disease may be attributed to a metastatic deposition, secondary to local disruption of blood–brain barrier (BBB), or disorder of neuronal calcium phosphorus metabolism (7).

Fahr's disease needs to be differentiated from Fahr's syndrome in which the basal ganglia calcification is secondary to either a systemic or metabolic disorder, the most common of which is hypoparathy roidism. The differentiation between Fahr's disease and Fahr's syndrome is specially highlighted when brain CT exhibits diffuse, symmetric calcifications in bilateral basal ganglia, thalami, cerebellar dentate nuclei and cerebral white matter (11). Other conditions which could lead to intracranial calcifications include- hyperparathyroidism, pseudohypoparathyroidism, neurocysticercosis, toxoplasmosis, tuberculosis, HIV infection, astrocytoma, hypervitaminosis, calcified infarct, radiotherapy, mitochondrial encephalopathies, SLE, tuberous sclerosis and leukodystrophies. Bilateral almost symmetric calcification involving striatum, pallidum with or without deposits in dentate nucleus, thalamus and white matter is reported from asymptomatic individuals to a variety of neurological conditions including autosomal dominant inheritance to pseudo-pseudohypopa rathyroidism. (1)(2) Some studies have shown presence of elements (zinc, aluminum, copper) along with cerebral calcifications. CT scan is the best modality of investigation in the diagnosis of Fahr's disease. The minimum age at which a negative CT scan can exclude the disease is not established yet.(5). MRI is not very useful in this particular condition as demonstrated in some studies- as the signal maybe variable. On T1 weighted images, low signal is due to low proton density of calcium and presence of other minerals in higher concentrations. They might also present with hyperintense signal in the presence of proteins and mucopolysaccharides binding the mineral ions. If the disease is in the intermediary stage, even in that case it might go undetected on the MRI (9). The prognosis is variable and difficult to predict, but in a study serial CT scan performed in an adult patient revealed a progressive increase in cerebral atrophy (5). There is no cure for this condition. The treatment should be focused on symptomatic relief for the patient with improved quality of life for booth the patient and the caretaker.

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