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CASE REPORT

A Rare Case of Pediatric Fahr's Disease with Myoclonus as Initial Presentation

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ABSTRACT

Background: Fahr's disease is a rare neurological disorder characterized by abnormal calcified deposits in the basal ganglia. Only a small number of pediatric onset cases have been reported in the literature to date, and the majority of the Fahr's disease literature is based on case reports.

Case report: We are reporting a rare case of an 8-year-old child with Fahr's disease presenting with myoclonus and dysarthria. Bilateral calcifications in the subcortical white matter and basal ganglia were visible on his CT brain. Clinically, he is treated symptomatically and is responding well to clonazepam.

Conclusion: We want to highlight that Fahr's disease can have a wider spectrum of age of onset and also to point out that it should be considered a differential in children presenting with movement disorder with intracerebral calcifications. Our aim is to expand the current knowledge available on clinic-radiological features of this disease entity and encourage further research in genetic and treatment aspects by increasing awareness regarding Fahr's disease.

KEYWORDS

• Fahr's disease • Myoclonus • Basal ganglia calcification

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INTRODUCTION

Fahr's disease, or idiopathic basal ganglia calcification, is a rare neurological disorder characterized by abnormal calcified deposits in the basal ganglia. Progressive calcium deposition in the central nervous system, particularly in the brain regions that regulate movement, including the hippocampus, cerebral cortex, cerebellum, thalamus, dentate nucleus, subcortical white matter, and basal ganglia, is a hallmark of the condition.[1] The calcium deposits are observed in the white matter's capillaries and arteries. It is an inherited or sporadic neurological disorder with a prevalence of <1/1 000 000.[2] Most of the literature available on Fahr's disease is based on case reports with few studies on the prevalence of the disease. It typically manifests in the fourth to sixth decade.

Fahr's syndrome, which is typically a subsequent illness to underlying conditions, must be differentiated from Fahr's disease, an idiopathic condition of intracerebral calcification. Clinically, it manifests as speech impairment, parkinsonism, paresis, and dystonia, among other movement problems. In addition, they might have neuropsychiatric symptoms, seizures, or stroke-like episodes.[3] The diagnostic criteria for Fahr's disease include bilateral calcification of the basal ganglia visualized on neuroimaging, especially CT scans, and progressive neurological dysfunction, which includes movement disorder and/or neuropsychiatric manifestations.[4] We are reporting a rare case of an 8-year-old child with Fahr's disease presenting with myoclonus.

CASE REPORT

We are presenting the case of an 8-year-old boy who presented with a complaint of myoclonus. The myoclonus began approximately six months ago with a subacute onset and gradually progressive in nature. He exhibits shock-like jerks primarily affecting his upper limbs, occurring about 4 to 5 times a day intermittently. The frequency of these jerks has been gradually increasing over the past few months. Additionally, he has started to experience speech difficulties over the last month, with his dysarthria worsening progressively.

His birth history was not contributory. His development was age-appropriate. He is the firstborn child, and there is no history of consanguinity. Additionally, there is no significant family history.

On examination, he was vitally stable and exhibited intermittent myoclonic jerks in his upper limbs. There were no other significant findings during the general examination. Upon assessing the central nervous system, he displayed dysarthric speech. His higher cognitive functions and both motor and sensory examinations were unremarkable. The remainder of the systemic examination was also normal.

Hematological analysis revealed a white blood cell count of 10,800 cells/mm^3 (5000–11000 Cu/mm), a hemoglobin concentration of 11.4 gm/dl (12–14 gm/dl), and a platelet count of 248,000 Cu/mm (1.5–4,00,000 Cu/mm). The C-reactive protein level was measured at 2 mg/dl (< 6 mg/dl). Values for serum electrolytes, liver and renal function tests, along with an international normalized ratio of 1.13 (control 1.1), prothrombin time of 13 seconds (control 11.6 seconds), and activated partial thromboplastin time of 26 seconds (control 26.6 seconds), were all within normal limits. Serum biochemistry showed an ionized calcium level of 4.4 mg/dl and a total calcium level of 9.9 mg/dl (9–11 mg/dl), a serum phosphorus level of 4.3 mg/dl (2.5–4.5 mg/dl), and a serum parathyroid hormone level of 34 pg/dl (normal range: 9–65 pg/dl). Thyroid function tests, including T3, T4, and TSH, were all normal. The serum antinuclear antibody, TORCH profile, was normal, and anti nuclear antibody was negative and vitamin D level was within normal limit.

A CT scan of the brain showed calcification in bilateral basal ganglia and along deep sulci involving the cortex and subcortical white matter along bilateral anterior frontal lobes and parieto-occipital lobes.

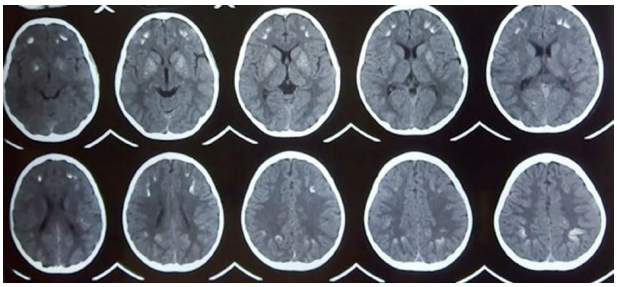


Figure 1: CT Brain showing calcification seen in bilateral basal ganglia and subcortical white matter

Clinically, the child was started on clonazepam, and his myoclonus significantly improved. But the dysarthria was persistent.

DISCUSSION

Fahr's disease is a rare neurodegenerative disorder characterized by progressive calcium deposition in various parts of the central nervous system, like the basal ganglia, thalamus, dentate nucleus, cerebral cortex, cerebellum, subcortical white matter, and hippocampus. Fahr's disease was first noted by German neurologist Karl Theodor Fahr in 1930.[2] The mode of inheritance is either autosomal dominant/recessive or sporadic.

Most of the literature available on Fahr's disease is based on case reports with few studies on the prevalence of the disease. Although the actual prevalence of Fahr's disease is unknown, the publications that are now available maintain that it is less than 1/1,000,000. It presents usually between the 4th and 6th decades. But our case is of an 8-year-old boy with Fahr's disease. Only a handful of pediatric cases have been described till now and none previously reported had myoclonus as initial presentation.

The clinical presentation in our case was with myoclonus and dysarthria. The previously reported clinical features include Parkinsonian symptoms, chorea, and athetosis.[5,6] Additionally, cognitive impairment, seizures, speech difficulties, cerebellar symptoms, and stroke-like events are frequently paired with mental health illnesses such as dementia, mood disorders, and psychosis. There have previously been three pediatric cases documented: a 5-year-old who presented with seizures and developmental delay[7], a 14-year-old boy[8], and a 15-year-old who experienced recurrent seizures.[9]

The proposed underlying pathogenesis of disease at the molecular level is that calcification generally develops within the blood vessel wall and in the perivascular space, which progresses further to involve surrounding neuron cells. Defective iron transport and the generation of free radicals cause tissue damage, which triggers the start of calcification. It occurs secondarily around a nidus composed of mucopolysaccharide and related substances.[10]

Progressive mineralization leads to compression of the underlying blood vessel lumen, thus initiating a cycle of impaired blood flow, neural tissue injury, and mineral deposition. Basal ganglia concretions are recognized as basophilic globules tracking the vessels of arteries, veins and capillaries. Dysmyelination and gradual calcification of the brain are caused by aberrant calcium and phosphate deposition.[11]

Fahr's disease is an idiopathic condition without any underlying secondary cause for calcification, and it needs to be diffentiated from Fahr's syndrome in which a secondary underlying etiology is present. The causes for Fahr's syndrome include TORCH infection, neurocysticercosis, hypoparathyroidism, pseudohypoparathyroidism, pseudo pseudohypoparathyroidism, hypothyroidism, neurofibromatosis, tuberculosis, tuberous sclerosis, cerebral hemorrhage, and vascular disease. Lead poisoning, cancer, radiation therapy, hypervitaminosis D, and systemic lupus are additional possible causes to take into account.[(12)(13)]

In our case, a diagnosis of Fahr's disease was made having met the criteria for diagnosis, which included the presence of symmetrical intracranial calcification in the CT scan of the basal ganglia, normal calcium/phosphate levels, absence of other metabolic disorders underlying diseases that could cause derangement in calcium/phosphate metabolism, and normal levels of thyroid and parathyroid hormones and alkaline phosphatase.

CT scan is the imaging modality of choice. Our case showed calcification seen in bilateral basal ganglia and subcortical white matter. CT shows dense calcifications within the basal ganglia, subcortical white matter of the posterior parietal lobes, and the dentate nuclei of the cerebellum.[14] MRI is not as specific as a CT scan. It shows that the signal is variable. On T1-weighted images, low signal is due to the low proton density of calcium and other mineral ions present in higher concentration. However, they might present hyperintense signals due to proteins and mucopolysaccharides binding the mineral ions. The calcification might also be undetected on MRI when they are in an intermediary stage.[15]

There is no specific treatment for Fahr's disease; only symptomatic management is available according to the presenting complaints. Our patient is responding well to clonazepam and is clinically better.

CONCLUSION

Pediatric onset Fahr's disease is considerably rarer than the more frequently encountered Fahr's syndrome, which typically manifests after the age of 40. We aim to emphasize that Fahr's disease can present across a broader age range and should be considered when evaluating a child with movement disorders accompanied by intracerebral calcifications. Our objective is to enhance the existing understanding of the clinical and radiological characteristics of this condition and to promote further investigation into its genetic and treatment aspects by raising awareness about Fahr's disease.

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No Ethical issues involved

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