

A Rare Case report of Fahr's Disease with typical clinical presentation

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Abstract: Fahr's disease is a rare, degenerative neurological condition which is characterized by idiopathic calcification of the basal ganglia and dentate nuclei. Neuropsychiatric, extra pyramidal & cerebellar symptoms, convulsive seizures, parkinsonian features, dementia and speech disorders may accompany the clinical picture. There are also cases without neurological signs, which have been reported. This disease usually appears between the age of 40-60 years. We discuss one such case which came to us with typical neuro psychiatric presentations.

Case Report: A 40 year old male (Fig 1) presented to our hospital with history of head injury following fall. On admission patient revealed h/o visual hallucinations followed by fall from bike. Patient was a known alcoholic for the past 20 years and a hypertensive for the last 7 years. Patient's family history is unremarkable but for the fact that his younger brother had committed suicide before 7 years. No proper medical records of his brother was available. The patient had been performing his day to day activities without any major shortcomings. Off late the patients' relatives have started observing subtle neuro psychiatric symptoms.

Clinical Examination : He was moderately built and nourished without any skeletal abnormalities or neurocutaneous markers. On neurological examination, the patient revealed extra pyramidal and cerebellar symptoms. He was conscious and oriented to the time and place, with a normal memory and intelligence. His speech was slow and dysarthric. He had a mask like, expressionless face with a reduced blink rate. Bradykinesia was noted, with a reduced arm swing and a stooped posture. He did not have any motor weakness or cranial nerve involvement. Psychiatric examination came to a provisional diagnosis of delirium.

His biochemical examination, cardiac evaluation, toxic screen, metabolic screen, RFT, LFT blood counts, ECG were all within normal limits

Patient was subjected to radiological investigations. Routine chest xray and abdominal USG were normal. Plain CT brain was done with thin cut reformatted images analysed (Fig 2). Plain CT brain revealed dense coarse calcific densities in clumps in bilateral thalami, globus pallidus, caudate nuclei, and cerebellar dentate nuclei in fairly symmetric fashion.

Keywords: Fahr's syndrome, Bilateral intracranial calcification, Extrapyramidal, Parkinson's disease, intracerebral calcinosis & dysarthria.

I. Introduction

Fahr's disease is a rare, degenerative neurological condition which is characterized by the idiopathic calcification of the basal ganglia and dentate nuclei. This disease was described for the first time by a German neurologist, Karl Theodor Fahr in 1930. Neuropsychiatric, extra pyramidal and cerebellar symptoms, convulsive seizures, Parkinsonian features, dementia and speech disorders may accompany the clinical picture. This disease usually appears between the age of 40-60 years . It is rare in children, who may present with choreoathetotic movements. Within the basal ganglia, the globus pallidus is the most frequent site of the calcification but deposits may be present in the putamen, the caudate nucleus, internal capsule, dentate nucleus, thalamus, cerebellum and cerebral white matter. Histologically, these deposits contain proteins and polysaccharides and are found in the media layer of the small vessels. Although the pathogenesis is not known, it may be secondary to impaired BBB or to a neuronal calcium phosphoric metabolism disorder [1].

II. Discussion

Bilateral Striopallido Dentate Calcinosis (BSPDC)/ idiopathic Basal ganglia calcification, is a neurodegenerative disorder which is popularly known as Fahr's Syndrome. A bilateral, symmetrical, intra cranial calcification characterizes Fahr's disease with a predilection for the basal ganglia and the dentate nuclei. Because of the symmetrical involvement of these nuclei, the descriptive terminology, BSPDC, has been put forth. The minimum age at which a negative CT scan can suggest the exclusion of the disease has not been established as yet. To clarify whether the disease is sporadic or familial, doing the imaging scan of the parents and other kindred is more reliable than their clinical screening. This is a very rare disease of unknown prevalence. The typical age at the onset of the symptoms is 40-60 years. This is among the few inherited neurological conditions that lead to progressive dystonia Parkinsonism and neuropsychiatric manifestations. The

most common presentations as per the Fahr's Disease Registry are movement disorders, which account for about 55% of the cases. Among these, Parkinsonism was seen in 57% cases, chorea was seen in 19% cases, tremor was seen in 8% cases, dystonia was seen in 8% cases, athetosis was seen in 5% cases and orofacial dyskinesia was seen in 3% cases[2]. The other neurological manifestations include a cognitive impairment, cerebellar signs, speech disorders, pyramidal signs, psychiatric features, gait disorders and sensory changes. The clinical diagnosis of Fahr's disease is based on the combination of clinical features, brain imaging and on exclusion of other causes of the intracranial calcification. The imaging findings of the symmetric and extensive calcification are usually typical, as was seen in our case. The disorders of calcium metabolism may occur in association with the intracerebral calcification. Hypoparathyroidism, pseudohypoparathyroidism and hyperparathyroidism may be associated with the intracerebral calcification. Other causes of intracranial calcification include infectious diseases like Toxoplasmosis and Syphilis and inflammatory illnesses like SLE. But Fahr's disease represents a heterogeneous group of disorders that are not associated with any known disorder of the calcium metabolism [3]. Genetic studies have shown an autosomal dominant inheritance in the familial cases. A genetic heterogeneity and an anticipatory effect also have been observed. One multigenerational family with a linkage to the IBGC1 of chromosome 14 has been identified, but the causal gene is still unknown[4]. Computed tomography scan remains the most effective screening tool. No prenatal or genetic tests are available for genetic counseling.

Treatment

There is no cure for Fahr's syndrome, nor is there a standard course of treatment. Treatment targets symptomatic support. The response to Levodopa in those with Parkinson's features is reportedly poor. Antipsychotics may be indicated in those with psychotic symptoms and behavioural problems, and anticonvulsants for the control of seizures as was done in our case [5]. The National Institute of Neurological Disorders and Stroke (NINDS) supports and conducts research on neurogenetic disorders such as Fahr's disease. The goals of this research are to locate and understand the actions of the genes which are involved in this disorder. Finding these genes could lead to an effective method of treating and preventing Fahr's syndrome. Our patient was referred to neuropsychiatric evaluation and is under follow up after receiving medical management for his symptoms. We have asked the patient to come for regular follow up in our department atleast once a year.

III. Conclusion

Fahr's syndrome is an idiopathic neurodegenerative disorder. With proper knowledge of its clinical manifestations and more vigilant approach, a diagnosis can be made in time. Radiologists' role is paramount in not only early detection of this disease but also correlating clinically and alert the neurophysician. More research is required to locate and understand the action of the genes involved in this disorder. Finding these genes could lead to effective ways to treat and prevent Fahr's syndrome.

References

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Fig 1/Tab1.



Profile picture of the patient with masked identity.

Fig 2/ Tab2.

Axial ,sagittal and coronal sections of plain CT Brain of the patient showing symmetrical dense coarse calcific densities in basal ganglia, thalami and cerebellar dentate nuclei. Calcifications ranged from 230 – 660 HU in their min and max density.

