

A case of Fahr's disease presenting as epilepsy

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Abstract

Fahr's disease refers to sporadic or familial idiopathic basal ganglia calcification that is associated with many neurological, psychiatric and cognitive abnormalities. We report a case of Fahr's disease in a 25-year old man who presented to us with generalized tonic clonic seizure and behavioral abnormalities. Clinical examination did not show any abnormal physical findings. The CT scan of brain showed multiple, symmetric and extensive calcifications in the basal ganglia, thalamus, and dentate nucleus, subcortical white matter of cerebral and cerebellar areas. Presently the patient is under follow up with good seizure control with phenytoin and sodium valproate.

Keywords: Basal ganglia, Calcification, Fahr's disease, Seizure.

Introduction

Fahr's Disease (FD) is a rare degenerative neurological disorder, first noted by German neurologist Karl Theodor Fahr in 1930. It is characterized by intracranial calcification. The most common sites of calcification are basal ganglia and dentate nuclei of the cerebellum. Hence, one of the other names of the disease is Bilateral Striopallidodentate Calcification (BSPDC). People with FD frequently present with movement disorders such as rigidity, hypokinesia, tremor, choreoathetosis, and ataxia and with frontal subcortical and cortical patterns of behavioral disturbances such as psychosis, mood disorders, and dysexecutive neuropsychological syndrome.¹ Other neurological features are seizures or stroke-like events.² Sometimes patients may be asymptomatic. Higher incidence reported among males and typical age of onset is third to fifth decade^{3,4} although it has been described in younger age groups. It is often familial and this form may be transmitted as an autosomal recessive trait or may have autosomal dominant inheritance. Geschwind et al. in a genetic study, described a dominant autosomal inheritance of the hereditary form of FD and suggested that the disease is caused by mutations in genes located on the long arm of chromosome 14 (14q48).⁵ In other instances, the condition appears to be sporadic. There is no cure for FD, nor is there a standard course of treatment. Treatment is symptomatic. Most cases present with extrapyramidal symptoms. But here we describe a case of Fahr's disease, who presented with generalized tonic clonic seizures and behavioral abnormalities. Computed tomography scan of brain showed calcification of bilateral basal ganglia, cerebellum and cerebral subcortical white matter.

Case Report

25 years old man Mr. Mohan, presented to our neurology op with C / O Seizure - 7-8 years duration.

Semiology was generalized tonic clonic seizure (GTCS). Patient was on tab. phenytoin 300 mg/day and was taking drug irregularly. For the past 3-4 years frequency and severity of the seizures were increasing. Now along with phenytoin, tab. sodium valproate 600 mg/day had been started. Patient was an alcoholic, consumes alcohol twice to thrice/week. H/o substance abuse (cannabis and hash) was present. He was working as a mason and attending job irregularly. Also history revealed his behavioral disturbances in the form of not going along with parents and family members and quarrel with them frequently with aggressiveness. This sort of behaviour disturbance present for the past three to four years. H/ O poor scholastic performance was present. Studied upto 4th standard and discontinued. Born out of non-consanguineous marriage. No family h/o similar seizure or behaviour disorder. His younger brother was apparently normal. Patient was born by normal delivery with no features of birth asphyxia. No history suggestive of maternal illness or complications to his mother. Motor and mental milestones were normal. On examination patient was conscious, oriented. Mild cognitive impairment was present. Cooperative fairly while examination. Bp: 116/80 mmhg. PR: 86 / min. Neurological examination was normal. No extrapyramidal or cerebellar signs. Laboratory investigation revealed a normal hemogram, a normocytic normochromic picture on peripheral smear study. His urine analysis, LFT, RFT, blood sugar and thyroid function testing were all normal. Serum Calcium: 10.2 (8.5 - 10.5 mg/dl) and Phosphorus: 4 (2.5 - 4.5 mg/dl). Serum parathormone levels were normal. Chest X Ray was non contributory. HIV testing was negative. EEG: generalised 5-6 Hz slow waves present bilaterally. No epileptiform discharges seen. CT scan brain showed bilateral globus pallidus, putamen, caudate nucleus, thalamus calcification and cerebellar calcification and cerebral subcortical white matter

calcification (Fig. 1-4). With these clinical features and investigation reports diagnosis of Fahr's disease was

made out and patient was treated symptomatically.

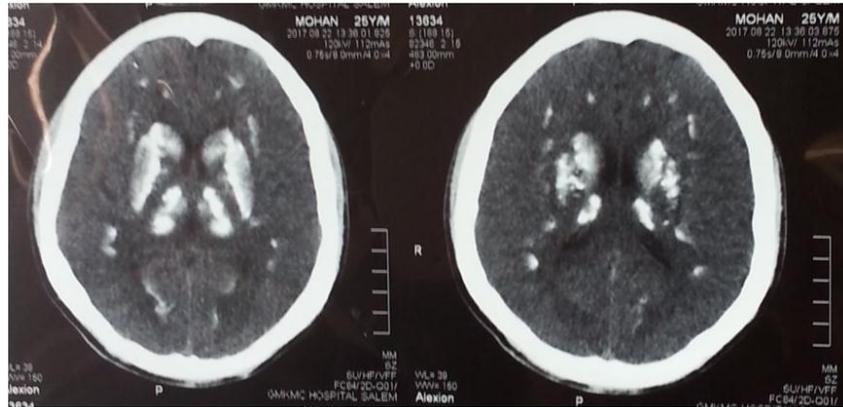


Fig. 1:



Fig. 2:



Fig. 3:

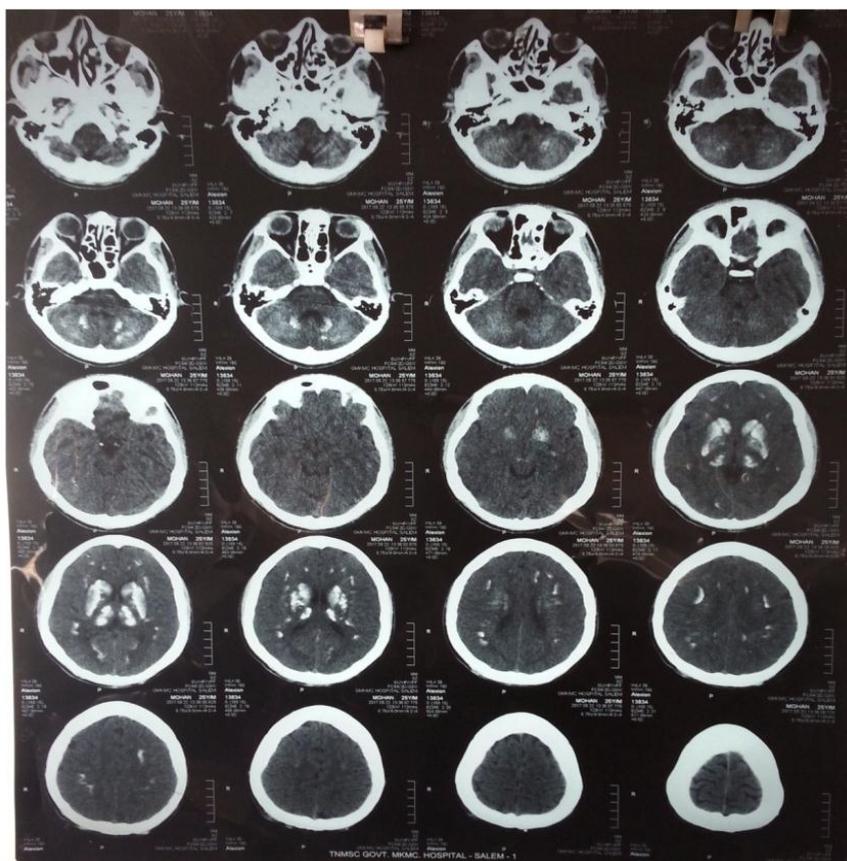


Fig. 4:

Discussion

FD is a rare condition with specific neuroradiological features but numerous clinical manifestations. In “Fahr’s Disease Registry,” the common manifestation was movement disorders (55%), (in particular parkinsonism (57%), while the hyperkinetic movement disorders accounted for the rest); cognitive impairment^{6,7} was the second most common manifestation followed by cerebellar impairment and speech disorder. Manyam BV et al. 2001 analyzed presentations of sixty one symptomatic cases and found that movement disorders were the most common presentation, which accounted for 55% of clinical presentations (Among this, parkinsonism 57%, chorea 19%, tremor 8%, dystonia 8%, athetosis 5% and orofacial dyskinesia 3%).⁴ Rarely epilepsy is a presenting symptom.⁸ The exact pathological process initiating the calcifying changes is not known, it may reflect slowly progressive metabolic or inflammatory processes in the brain, which subsequently calcifies and is probably responsible for the neurologic deficit observed. Also it has been suggested that tissue damage by free radicals or by abnormal iron transport may trigger calcification. Availability of brain CT-scan has increased the number of case reports of intracranial calcification, and brain CT- scan is considered more sensitive than magnetic resonance imaging for finding calcified deposits.⁹ It should be emphasized that there

are other conditions that can also produce intracranial calcifications. These conditions include endocrinopathies (hypoparathyroidism, hyperparathyroidism), systemic diseases (systemic sclerosis, systemic lupus erythematosus), infections (toxoplasmosis, neurocysticercosis, CMV, neurobrucellosis, HIV), calcified brain tumors, and various diseases such as tuberous sclerosis, mitochondrial encephalopathy, myotonic muscle dystrophy post-anoxic disorders, idiopathic hemochromatosis, heavy metal and carbon monoxide intoxication. These diseases may cause bilateral and non-symmetric cerebral calcifications mainly located in the basal ganglia and cerebellum. More over Fahr’s disease should be distinguished from incidentally found basal ganglia calcification in elderly population. The diagnosis of Fahr’s disease is complex and requires both clinical and radiological evidence and exclusion of other causes of intracranial calcification. The criteria include bilateral calcification of the basal ganglia with neuropsychiatric and/or extrapyramidal features associated with normal calcium and phosphate metabolism.¹⁰ In our patient the neuroimaging findings of symmetric and extensive calcification laid the basis of diagnosis. Our case fulfilled the diagnostic criteria – having presented with seizure disorder and behavioural disturbance associated with CT scan brain showing bilateral globus pallidus, putamen, caudate nucleus, thalamus calcification and cerebellar calcification and

cerebral subcortical white matter calcification.

Conclusion

In conclusion, while rare, Fahr's disease should be considered as a differential diagnosis for seizures, movement disorders or cognitive impairment in a tropical setting. This case is presented for the early age of onset of Fahr's Disease and its presentation as epilepsy with no evidence of a movement disorder, though movement disorder is the typical presentation.

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