Fahr’s Disease with Seizure Presentation

Rangaswamy¹, V Ranjith², L Vikas¹, R Santosh³

Abstract

Fahr’s disease or Fahr’s syndrome is a rare neurodegenerative disorder characterized by abnormal symmetrical calcifications of the basal ganglia, thalami, sub-cortical hemispheric white matter and deep cerebellar nuclei. It can be idiopathic or associated with an endocrinopathy, frequently with parathyroid disorder.

Clinical spectrum of the disease is wide ranging from neurological features like seizure, syncope, stroke like events, extra-pyramidal symptoms often combined with frontal sub-cortical pattern of behavioural dysfunction and psychiatric symptoms such as psychosis, mood disorder and dementia. Diagnosis is based on the clinical features, progressive neurological dysfunction, absence of biochemical abnormality and characteristic radiological imaging.

Introduction

We describe a case of idiopathic Fahr’s disease. Fahr’s Disease or Fahr’s syndrome is a rare neurodegenerative disorder...
characterized by abnormal symmetrical calcifications of the basal ganglia, thalami, sub-cortical hemispheric white matter and deep cerebellar nuclei.

Case Report

A 50 year old male patient presented with history of involuntary movements lasting for about 15 minutes, followed by disorientation and loss of consciousness. He was a chronic smoker with no history of hypertension or diabetes mellitus in the past. Patient’s attendant gave history of behavioural changes and reduced mentation since past 6 months with impaired cognitive function. He was shown to a local doctor in their village but condition remained the same after treatment. There is no history of similar complaints in the family members.

On examination vital parameters were normal. Mini mental score was only 11 out of 30. Orientation to time place and person, recent and remote memory were impaired, immediate memory was intact. Emotional lability was present. Routine blood investigations were normal.

X-ray skull showed intra-cerebral calcification (Figure 1).

CT scan brain plain showed dense calcifications in bilateral dentate nuclei, basal ganglia and corona radiata. Features were representing Fahr’s disease (Figures 2 - 4). Electroencephalogram revealed generalised epileptiform discharges diagnostic of tonic-clonic seizures with secondary generalization (Figures 5).

With strong clinical and radiological features of Fahr’s disease further investigations were done to find out the likely cause. To rule out the endocrinial involvement, serum calcium, serum phosphorous, alkaline phosphate levels, serum calcitonin and serum parathormone level assays were done. All the reports were within normal limits. Thus indicating an idiopathic basis of disease.

Discussion

Idiopathic basal ganglia calcification also known as Fahr’s disease/ Fahr’s syndrome is a rare inherited neurological disorder with abnormal calcium deposits in the brain. It was first noted by German neurologist Karl Theodor Fahr’s in 1930.1

Like our case; Benke et al study also showed that the patients experienced marked personali ty changes with apa thy compulsive features and anxiety.

Benke et al in another study showed that higher mental function remained unimpaired contrary to our case study where higher mental functions were impaired with Mini mental score examination was only 11 out of 30.2

Letterio Morgante et al in his study showed that Fahr’s disease had generalized tonic clonic seizures and CT Brain showing symmetrical dense calcification similar to our case.3,4 Whereas it also showed the cause to be hyperparathyroidism with hypocalcemia, hyperphosphatemia, high serum parathyroid hormone levels in our case all the parameters were within normal range.

The present case report shows that Fahr’s disease may remain asymptomatic in most cases with only certain neurological and psychiatric presentation.5,6 In our case the clinical features started 6 months prior to the admission itself. Strong suspicion is necessary for the diagnosis of the condition. He was evaluated at our centre based on the history and characteristic calcifications in the brain, which helped us to diagnose the disease.

Reference