Fahr’s Syndrome: a Rare Case of Idiopathic Basal Ganglia Calcification

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Abstract

Fahr’s syndrome (or Fahr’s disease) involves progressive calcific deposition in the wall of blood vessels of the basal ganglia and dentate nuclei of the cerebellum in young and middle-age persons. It is occasionally associated with mental retardation and extrapyramidal symptoms. Clinically it may present with an array of movement disorders, dementia and other behavioural disturbances. We report a case of 52-year-old male with progressive decline of motor and mental function and increasing frequency of seizures. Brain CT showed symmetrical calcification in basal ganglia, thalami, cerebellar dentate nuclei, centrum semiovale, and corona radiata. The clinical and radiological features in our case point towards this uncommon disorder.

Key words: Extrapyramidal, Parkinson’s disease, intracerebral calcinosis, dysarthria.

Introduction

Fahr’s syndrome also known as idiopathic basal ganglion calcification (IBGC), cerebrovascular ferrocalcinosis, Fahr’s intracerebral calcinosis, Fahr’s disease, or Morbus Fahr is a rare neurological disorder of unknown aetiology, characterised by neuropsychiatric abnormalities, Parkinsonian or choreoathetotic-type movement disturbances, and extensive symmetrical calcification of basal ganglia and dentate nuclei in the cerebellum. It was first noted by the German neurologist Karl Theodore Fahr in 1930. The disease is often familial with autosomal dominant inheritance, but a few cases have been reported to have autosomal recessive inheritance and even sporadic cases have been reported in the literature.

Case report

A 52-year-old male, who was a chronic smoker and alcoholic, presented with a history of progressive deterioration of mental and motor functions for the last five years. There was history of generalised tonic-clonic seizures with increased frequency since the past few months. The patient was emotionally labile with a history of sudden crying, aggressive behaviour, and frequent mood changes. He tried to strangulate his wife once. His memory was declining and sleep was getting affected. There was no family history of mental illness, dementia, or major physical illness.

On examination, the patient was normotensive but had abnormal mental functions. He had dysarthria with cognitive impairment. The patient was conscious, but had inconsistency regarding orientation to time, place, and person. Sometimes he did not obey verbal commands and lay still with a mask-like expressionless face. Ataxia and rigidity of all four limbs was present.

Fig. 1: CT scan showing bilateral calcification in the dentate nuclei.
prevalence, characterised by symmetrical intracranial calcification with a predilection for the basal ganglia and dentate nuclei. As the basal ganglia and dentate nuclei are always involved in a symmetrical pattern, the term Bilateral Striopallidodentate Calcinosi (BSPDC) was suggested. Typical age of presentation is in middle-age between the 4th - 6th decades of life (as seen in our case), although an early onset type may also occur. Symptoms of this disorder may include deterioration of motor function, dementia, seizures, headache, dysarthria, spasticity, visual impairments, and athetosis. Fahr’s syndrome can also include symptoms of Parkinson’s disease such as tremors, muscle rigidity, mask-like facial appearance, shuffling gait, and a “pin-rolling” motion of the fingers.

Our case presented with cognitive impairment, speech disorder, extrapyramidal features like rigidity and mask-like face, and psychotic features.

Discussion

Fahr’s syndrome is a very rare disease with unknown prevalence, characterised by symmetrical intracranial calcification with a predilection for the basal ganglia and dentate nuclei. As the basal ganglia and dentate nuclei are always involved in a symmetrical pattern, the term Bilateral Striopallidodentate Calcinosi (BSPDC) was suggested. Typical age of presentation is in middle-age between the 4th - 6th decades of life (as seen in our case), although an early onset type may also occur. Symptoms of this disorder may include deterioration of motor function, dementia, seizures, headache, dysarthria, spasticity, visual impairments, and athetosis. Fahr’s syndrome can also include symptoms of Parkinson’s disease such as tremors, muscle rigidity, mask-like facial appearance, shuffling gait, and a “pin-rolling” motion of the fingers.

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Benke et al studied the brain metabolism in a person with Fahr’s disease who presented with predominant frontal lobe syndrome and dementia. There was a massive...
reduction in the glucose metabolism in both the basal ganglia and frontal lobes which correlated with the clinical picture of disinhibition and personality change. The involvement of frontal-subcortical circuits provides a hypothetical framework for the interpretation of cognitive and psychotic problems in Fahr’s disease.

Making a clinical diagnosis of Fahr’s syndrome relies on the combination of clinical features, brain imaging, and exclusion of other causes of intracranial calcification. Symmetrical and extensive calcification of basal ganglia, dentate nuclei, and centrum semiovale are typical and conspicuous of Fahr’s syndrome.

Normal serum levels of calcium, phosphorus, alkaline phosphatase, and parathormone can help in differentiating it from the endocrine disorders like hyperparathyroidism, pseudo-hypoparathyroidism, and pseudo-pseudo-hypoparathyroidism as happened in our case. Other rare causes of scattered basal ganglia and dentate nuclei calcification are tuberous sclerosis, toxoplasmosis, syphilis, and inflammatory illness such as systemic lupus erythematosus and all these must be ruled-out in suspected cases.

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.

**Conclusion**

Though Fahr’s syndrome is a rare idiopathic neurodegenerative disorder, yet, with proper knowledge of its clinical manifestations and more vigilant approach, a diagnosis can be made in time. 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**