CASE REPORT

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Fahr's Disease: A Rare Neuro-degenerative Disorder

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Abstract

Fahr's disease is a rare, autosomal dominant/sporadic inherited neurological disorder characterised by abnormal deposits of calcium in areas of Basal Ganglia, Thalamus, Hippocampus, Cerebral cortex, Cerebellum, and Dentate Nucleus. Clinical manifestations of this disease incorporate a wide variety of symptoms, ranging from neurological symptoms involving extrapyramidal system to neuropsychiatric abnormalities of memory and concentration to movement disorders including Parkinsonism, chorea, and tremors. These symptoms generally occur later in the course of the disease. Age of onset is typically in the 40s or 50s, although it can occur at any time during life. We are presenting a case of Fahr's disease in a 56-year-old female who presented with chronic headache, focal seizures, Parkinsonism and psychiatric symptoms.

Key words: Fahr's disease, Basal Ganglia Calcification, Parkinsonism, Neuro-degeneration.

Introduction

Fahr's disease also called as Primary Familial Basal ganglia Calcification (PFBC) or Familial Idiopathic Basal Ganglia Calcification (FIBGC) is a rare autosomal dominantly inherited or sporadic neurological disorder characterised by abnormal deposition of calcium in various parts of brain such as Basal Ganglia, Thalamus, Hippocampus, Cerebral cortex, Cerebellum, and Dentate Nucleus[1]. It has a prevalence of <1/1,000,000 population[2]. It was first described in 1930 by the German neuropathologist Karl Theodor Fahr[3]. Symptoms of Fahr's disease vary and may range from extrapyramidal symptoms (spastisity, dysarthria, gait disturbances, tremors, athetosis), neuro-psychiatric symptoms, headache and seizures to cerebellar symptoms and dementia[4]. Very few cases have been reported worldwide and literature on this disease is limited. Hence, we are reporting a case of Fahr's disease which was admitted in our institution.

Case report

A 56-year-old lady was admitted in the medical ward of RNT Medical College and MBGH, Udaipur, Rajasthan with history of headache and slowness of movements for the last six months which was gradually progressive in nature. On further evaluation she gave history that 15 years ago she had abnormal tonic movements of all four limbs which persisted for 3 - 5 minutes without loss of consciousness, urinary or a fecal incontinence or tongue bite. These episodes were one to two times in a month along with behavioural changes. She consulted a psychiatrist for this illness and she was prescribed medications in the form of carbamazepine 200 mg three times a day, clobazam 10 mg once a day and clonazepam 0. 5 mg once a day. Her symptoms were well controlled and she was on regular follow-up with the psychiatrist for the last 14 years. No neuro-imaging was done at the time of her diagnosis and during follow-up.

This time the patient was admitted with complaints of headache and slowness of movements . She was examined clinically and we found to have rigidity in all four limbs with normal deep tendon reflexes (DTR), bilaterally extensor plantar reflex, short stepping gait with loss of associated movements. Primordial reflexes in the form of glabellar tap, palmomental reflex and snuffle reflex were found positive. Hence, we made the provisional diagnosis of an extra-pyramidal syndrome possibly Parkinsons disease along with pyramidal tract involvement (Parkinson plus syndrome).

She underwent all routine investigations which included complete blood count, ESR, renal and liver function tests, urine routine, chest X-ray and electrocardiogram which were found to be normal. She also underwent brain imaging in the form of CT scan and it revealed extensive bilateral calcification of basal ganglia, cerebral hemispheres, thalamus, and cerebellum (Fig. 1). Hence, we suspected a diagnosis of Primary Familial Basal Ganglia Calcification (PFBC). To confirm the diagnosis of Fahr's disease, this patient was extensively evaluated to rule-out secondary causes of bilateral basal ganglia calcification like infection, trauma, metabolic disorders, etc.

She then underwent specific investigation like serum VDRL, serum for TORCH infection, ANA, LE cell, CRP, serum calcium,

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phosphorus, alkaline phosphatase, serum parathyroid hormone estimation and all these were found to be normal. She was investigated further and MRI brain was done showing symmetrical areas of calcification of bilateral basal ganglia, corona radiata, cerebellar folia and thalami appearing hypointense on T2W and FLAIR images and hyperintense on T1W and showing blooming on SW images

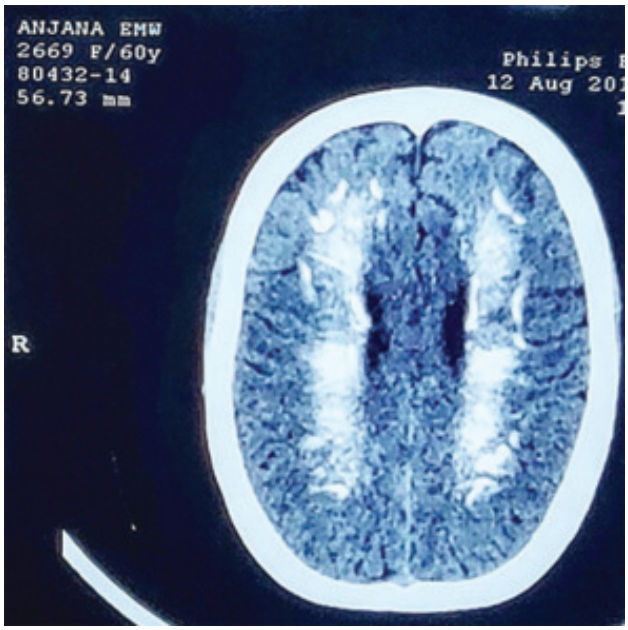

Fig. 1: *CT imaging of brain of the patient showing extensive calcification in bilateral basal Ganglia.*

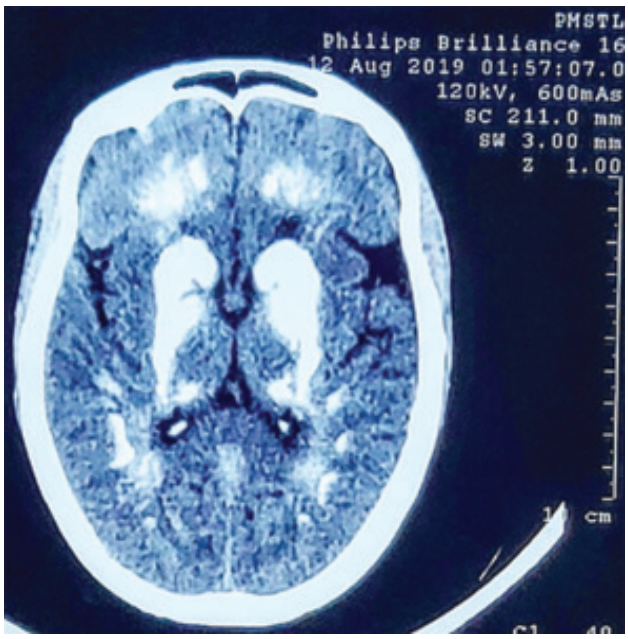

Fig. 2: *CT imaging brain of the son of the patient showing extensive calcification in bilateral basal Ganglia.*

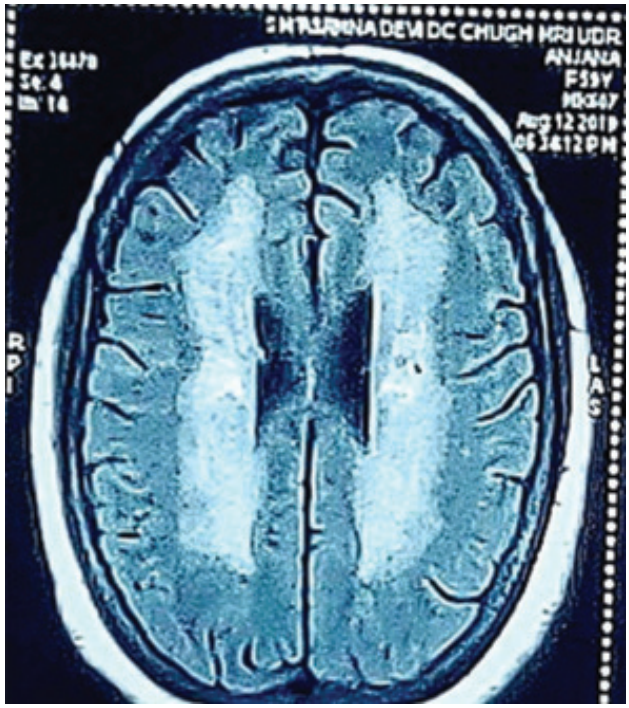

Fig. 3: *MRI imaging of the brain of the patient showing extensive calcification in bilateral basal Ganglia.*

suggestive of Fahr's disease (Fig. 3).

The patient's parents were no longer alive. She had two offsprings, a son of age 35 years and a daughter of age 30 years. Both of them were healthy and they did not have any significant history suggestive of Fahr's disease. They were screened by CT scan of brain. The CT brain of the son showed extensive calcification in the bilateral basal ganglia (Fig. 2) while the CT brain of daughter was found to be normal. The patient was asked to go for chromosomal studies but her relatives refused for the same.

Discussion

Fahr's disease is a rare autosomal dominant/sporadic neurodegenerative disease which occurs commonly in the fourth and fifth decade of life, but rarely cases have been reported in childhood[5]. Very few cases have been reported worldwide and not much is known regarding the aetiopathogenesis of this disease. In Fahr's disease, there is extensive calcification of the brain tissues and to label as primary calcification we should rule-out secondary causes of basal ganglia calcification like infections (syphilis, TORCH complex), trauma, calcium metabolism abnormalities, toxins, hyperparathyroidism, and pseudohyperparathyroidism.

Moskowitz *et al* 1971[6], Eliet *et al* 1989[7] and Manyam *et al* 2005[8] described the diagnostic criteria of Fahr's disease:-

- Bilateral calcification of basal ganglia visualised on neuro-imaging.
- Progressive neurological dysfunction which generally includes movement disorders and/or neuro-psychiatric manifestations. Age of onset is typically in the fourth and fifth decade of life although this dysfunction may present in childhood.
- Absence of biochemical abnormality and somatic features suggestive of mitochondrial or metabolic disease.
- Absence of an infection, toxins, or trauma as a cause.
- Genetic study or family history suggestive of autosomal dominant inheritance of the disease.

For the treatment of this disease, not is available. The only available options may be supportive in the form of anti-epileptic drugs, anti-psychotic drugs, and anti-Parkinsons drugs. However, patients are poorly responsive to these drugs.

Conclusion

Fahr's disease is a rare neuro-degenerative disease which may present in the fourth and fifth decade of life. It may clinically manifest with variable neurological, psychiatric symptoms, or both. Our patient was diagnosed as a psychiatric disorder and was on treatment for the same for the past 15 years without undergoing any neuro-imaging. Hence, it is advisable that before labelling a diagnosis of a psychiatric illness, neuro-imaging is very important to rule-out all primary neurological disorders.

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