Case Report

A rare case report – FAHR disease

Neha Gupta[1*], Poonam Gupta[2], Jai Chowdhary[3], A P Verma[4]

Mahatma Gandhi Medical College and Hospital, Sitapura, Jaipur, Rajasthan, INDIA.
Email: drguptaneha234@gmail.com

Abstract

Fahr's disease or Fahr's syndrome is a rare, neurological disorder characterized by abnormal calcified deposits in basal ganglia and cerebral cortex. Calcified deposits are made up of calcium carbonate and calcium phosphate, and are commonly located in the Basal Ganglia, Thalamus, Hippocampus, Cerebral cortex, Cerebellar Subcortical white matter and Dentate Nucleus. Molecular genetics of this disease haven't been studied extensively; hence evidence at the molecular and genetic level is limited. Fahr's disease commonly affects young to middle aged adults. Etiology of this syndrome does not identify a specific agent but associations with a number of conditions have been noted; most common of which are endocrine disorders, mitochondrial myopathies, dermatological abnormalities and infectious diseases. Clinical manifestations of this disease incorporate a wide variety of symptoms, ranging from neurological symptoms of extrapyramidal system to neuropsychiatric abnormalities of memory and concentration to movement disorders including Parkinsonism, chorea and tremors amongst others. Diagnostic criteria for this disease has been formulated after modifications from previous evidence and can be stated briefly, it consist of bilateral calcification of basal ganglia, progressive neurologic dysfunction, absence of biochemical abnormalities, absence of an infectious, traumatic or toxic cause and a significant family history. Imaging modalities for the diagnosis include CT, MRI, and plain radiography of skull. Other investigations include blood and urine testing for hematologic and biochemical indices. Disease is as yet incurable but management and treatment strategies mainly focus on symptomatic relief and eradication of causative factors; however certain evidence is present to suggest that early diagnosis and treatment can reverse the calcification process leading to complete recovery of mental functions.

Keywords: Fahr's syndrome, Hippocampus, myopathies, Parkinsonism, Imaging.

Address for Correspondence
Dr. Neha Gupta, Mahatma Gandhi Medical College and Hospital, Sitapura, Jaipur, Rajasthan, INDIA.
Email: drguptaneha234@gmail.com
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INTRODUCTION

Basal ganglia calcification is also known as Fahr's disease or Fahr's syndrome. It is a rare inherited or sporadic neurological disorder with a prevalence of <1/1,000,000[1-3]. It was first described by German neurologist Karl Theodor Fahr in 1930[4]. It is characterized by abnormal deposition of calcium in areas of the brain that control movements including basal ganglia, thalamus, dentatenucleus, cerebral cortex, cerebellum, subcortical white matter, and hippocampus[5]. Most cases present with extra pyramidal symptoms initially. Additionally, they may present with cerebellar dysfunction, speech difficulty, dementia and neuropsychiatric symptoms[6].

CASE REPORT

A 62 yr old female patient came to neurological department of Mahatma Gandhi hospital with c/o Neurological disturbance like unable to walk properly, recent memory loss and No h/o fever. Lab and imaging studies, histologic findings reveals standard laboratory tests values were in the normal range.

MR FINDINGS

MR Brain was done by GE optima 1.5 tesla MR machine. MR scan revealed foci of b/l symmetrical calcifaction in basal ganglia involving caudate nucleus, putamen, and globus pallidus appearing hypointense on GRE images, iso to slightly hypo on T2W sequences and hyperintense on T1W sequence. Multiple foci of b/l symmetrical T1W hyperintensity are seen in medial aspect of b/l cerebellum (dentate nuclei), thalami and periventricular and deep white matter of b/l cerebral hemispheres and centrum semiovale.

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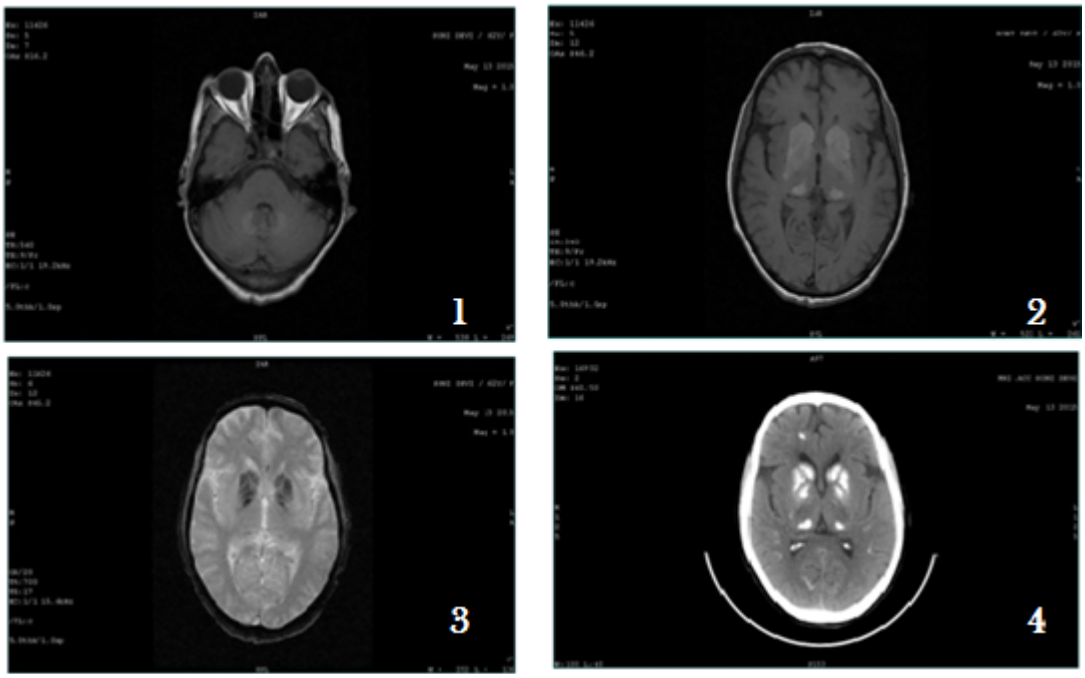

Figure 1: MR T1 W AXIAL SCAN OF BRAIN hyperintensities in bilateral dentate nuclei
Figure 2: And bilateral basalganglion
Figure 3: Shows GRE MR axial scan of brain fig4Axial CT scan brain showed hyperdensities
Figure 4: Shows calcification in bilateral basal ganglion in bilateral basal ganglion

CT FINDINGS

CT scan brain was performed by GE optima 128 slice machine and revealed B/L symmetrical calcification seen in basal ganglion, b/l cerebellum, thalami, and periventricular and deep white matter of b/l cerebral hemisphere and centrum semiovale.

DISCUSSION

Diagnostic criteria of Fahr's syndrome has been modified and derived from Moskowitz et al. 1971, Ellie et al. 1989, Manyam 2005[3,7,8] and it can be stated as follows:

- Bilateral calcification of the basal ganglia visualizedon neuroimaging. Other brain regions may also beobserved.
- Progressive neurologic dysfunction, which generally includes a movement disorder and/orneuropsychiatric manifestations. Age of onset is typically in the fourth or fifth decade, although this dysfunction may also present in childhood.*
- Absence of biochemical abnormalities and somaticfeatures suggestive of a mitochondrial or metabolic disease or other systemic disorder.
- Absence of an infectious, toxic, or traumatic cause.
- Family history consistent with autosomal dominant inheritance.

Fahr's disease is most commonly transmitted as an Autosomal Dominant trait; but it may also be passed on as an autosomal recessive trait or it may occur sporadically. A Locus at 14q (IBGC1) has been suggested to be involved commonly. A second locus has been identified on chromosome 8 and a third on chromosome 2.Fahr's syndrome typically affects individuals in the 3rd and 4th decades of their lives[2,8]. A variety of neurological signs and symptoms are associated with Fahr's syndrome. In adults loss of consciousness and seizures have been reported with hypothyroid hypocalcaemia. Tetany is present, but it is difficult to distinguish from occasional myoclonus of epileptic disorder. Also, spasticity, gait disorder, speech impairment, dementia, parkinsonism, chorea, tremors, dystonia, myoclonus and coma are present. Papilledema of intracranial hypertension, Pleocytosis of CSF, paroxysmal Choreothetosis and paroxysma Movement disorders in Fahr syndrome unveil as a spectrum of symptoms including clumsiness, fatigability, unsteady gait, slow or slurred speech, dysarthria dysphagia, involuntary movements and muscle cramping. Neurological evaluation reveals that parkinsonism is present in 57%, chorea 19%, tremors 8%, dystonia 8%, athetosis 5% and orofacial dyskinesia in 3% of patients[2]. The plain skull radiograph has been shown to be the imaging modality of diagnostic value. Calcifications appear as clusters of punctate densities symmetrically distributed above Sella Turica and lateral

to the midline, while subcortical and cerebellar calcifications appear wavy. Computed tomography is preferable method of localizing and assessing the extent of Cerebral Calcification. Most frequently affected area is the lenticular nucleus, especially the internal globus pallidus while Cerebellar gyri, brain stem, centrum semiovale, and subcortical white matter may also affected. Calcifications in the putamen, thalami, caudate, and dentate nuclei are also common. Occasionally, calcium deposits begin or predominate in regions outside the basal ganglia. Calcification seems to be progressive and gradual. MRI (magnetic resonance imaging) shows calcified areas in basal ganglia give a low intensity signal on a T2 image and low or high intensity signals on a T1weighted plane[9]. The cerebellar lesions are found to be more heterogeneous. There may be a chance of high intensity signals in both T1 and T2 images due to reactive gliosis or degenerating tissue within calcified areas[10].

CONCLUSION

Idiopathic basal ganglia calcification or Fahr's syndrome is a rare neurological disorder that is passed on in families as an autosomal dominant trait. This disorder is associated with a variety of other diseases but no specific etiologic agent has been identified yet. Diagnosis requires the presence of certain clinical criteria that may confuse the diagnosis with other conditions. New treatment modalities. Need to be discovered and employed to minimize loss of functionality associated with the disease. Of even more importance is to emphasize on the significance of genetic counseling of known at risk parents before conception.

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