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FAHR'S SYNDROME : A CASE REPORT

General Medicine

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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. Patients often present extrapyramidal or neuropsychological symptoms. Seizure is one of the rarest manifestations that can reveal Fahr disease. Here we present a 34 year old male who presented with convulsions of Generalized tonic clonic type . His CT Brain plain was suggestive of extensive calcification of bilateral basal ganglia and cerebellar tonsils.

KEYWORDS

INTRODUCTION

Basal ganglia calcification is also known as Fahr's disease Or Fahr's syndrome. It was first described by German neurologist Karl Theodor Fahr in 1930.

It is characterized by abnormal Deposition of calcium in areas of the brain that control Movements such as basal ganglia, thalamus, dentate Nucleus, cerebral cortex, cerebellum, subcortical white Matter, and hippocampus.

Case Report

34-year-old male was brought by relatives in unconscious state. As per informant he had a convulsion episode of GTCS variety which lasted for around 2-3 minutes and had self-resolved 1 hour back. It was associated with frothing from mouth, spontaneous urination in clothes. However, there was no tongue bite present. Immediately he was loaded with injection levetiracetam 1gm. After 30 minutes he was moving his limbs in response to pain. Patient GCS was 5/13. Patient was intubated and was kept on ventilator support.

He had similar episode of convulsion 3 months back which was his first convulsion episode. However no formal radiological scan was done then. Following first episode of convulsion he developed behavioural abnormalities. He was refer to psychiatrist and started on Tablet phenobarbitone 60mg once day. He was compliant to medication till 3 days back, but he missed his tablets giving rise to present episode.

In significant family history first degree sister had convulsion episode at the age of 8 years for which she took medication for 3 years but stopped medication by self. She had no further episodes of convulsions. Patient GCS was 5/13 but patient was hemodynamically stable.

CT Brain(Plain) Images :-

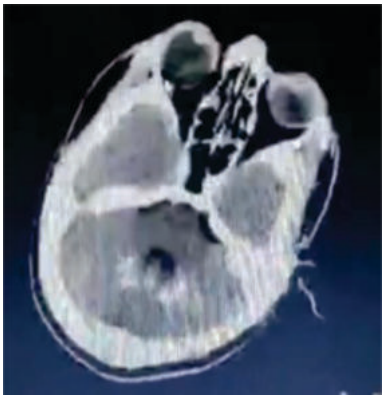

Figure A- Transverse Section Showing Cerebellar Tonsil Calcification

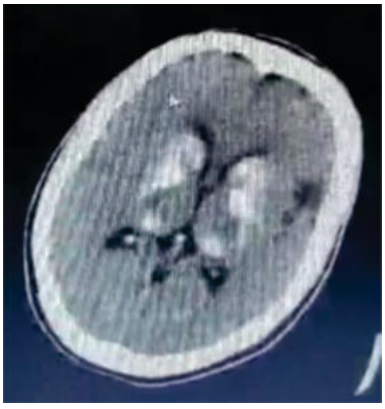

Figure B- Transverse Section Showing bilateral Basal Ganglia Calcification

Extensive calcification involving bilateral gangliocapsular region, corona radiata, centrum semiovale and cerebellar tonsils were strongly in favour of Fahr syndrome.

Serum calcium level 5. 5mg/dl. Serum Parathyroid hormone level could not be done due to resource limitation.

He was continued on anti-epileptics and intravenous calcium replacement was done. However, he developed ventilator associated pneumonia on day 6. Antibiotics were stepped up, but patient did not respond. He went into septic shock to which he succumbed.

DISCUSSION

Pathophysiological mechanism in Fahr syndrome involve multiple hypothesis The causes of brain calcifications, including parathyroid dysfunction and imbalances of calcium metabolism in general(1). It is not entirely clear whether abnormal calcium deposition in the brain is caused by a local destruction of the blood brain barrier or an intraneuronal calcium metabolic imbalance(2).

Regarding the etiopathogenic mechanism, the epileptic seizures may be caused by hypocalcemia due to primary hypoparathyroidism (explained by an increase in neuronal excitability induced by the drop inextracellular calcium concentration); however, another hypothesis that has gained prominence is , the dysfunction of the cortico-basal connections and their interhemispheric relationships (3)(4)

At present there are no specific guidelines regarding the efficiency of the anti-epileptic treatment in resolution of seizures in patients with hypocalcemia and cerebral calcifications secondary to hypocalcemia(4)

A trial exploring the efficiency of anti-epileptic therapy performed on a limited number of patients with hypoparathyroidism and epileptic seizures as the first symptom showed no significant difference in terms of seizure-free interval between the treatment and no-treatment groups. The treatment made no difference even in the subset of patients with subcortical calcifications. What is significant to note is that seizure suppression is co-related with calcium level normalization(4).

CONCLUSION

1. Parathyroid hormone is a conclusive parameter to diagnose hypoparathyroidism but in view of resource limited setting it was not possible.
2. Very low values of calcium <5. 5mg/dl is a pointer to underlying hypoparathyroidism.
3. The responsive to intravenous calcium was both therapeutic in terms of absence of further seizure and laboratory parameters.
4. The seizure in Fahr s disease showed better response in terms of efficacy to intravenous calcium over anti-epileptic drugs and oral calcium due to its faster onset of action and early normalization of serum calcium.

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