JMSCR Vol||05||Issue||11||Page 29946-29948||November 2017

www.jmscr.igmpublication.org
Impact Factor 5.84
Index Copernicus Value: 71.58
ISSN (e)-2347-176x ISSN (p) 2455-0450
crossref DOI: https://dx.doi.org/10.18535/jmscr/v5i11.23

IGM Publication
Journal Of Medical Science And Clinical Research
An Official Publication Of IGM Publication

Fahr's disease with Epilepsy: A Case Report

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Abstract
Fahr's disease, first described by Karl Theodor Fahr in 1930, refers to sporadic or familial idiopathic basal ganglia calcification that is associated with many neurological and psychiatric abnormalities, but may also be secondary to other diseases. Most cases present with extrapyramidal symptoms. But here we describe a case of Fahr's disease, who presented with extrapyramidal symptoms with generalized tonic clonic seizures. Computed tomography scan of brain showed bilateral calcification of cerebral hemispheres, basal ganglia and in parietal regions, due to abnormal calcium and phosphate metabolism.

Introduction

Fahr's disease or Fahr's syndrome is a rare inherited or sporadic neurological disorder with a prevalence of <1/1,000,000 and characterized by abnormal deposition of calcium in areas of the brain that control movements including basal ganglia, thalamus, dentate nucleus, cerebral cortex, cerebellum, subcortical white matter, and hippocampus.[1,2] It was first described by German neurologist Karl Theodor Fahr in 1930.[3] Most cases present with extra pyramidal symptoms initially. Additionally, they may present with cerebellar dysfunction, speech difficulty, dementia and neuropsychiatric symptoms. We describe a case of Fahr's disease, who presented with extrapyramidal symptoms with generalized tonic clonic seizures and behavioral abnormalities.

Case Report

A 17-year-old male presented with 2 weeks history of sudden involuntary movement involving both side of his body and multiple episodes of generalized tonic clonic seizures (GTCS). It was characterized by involuntary, unsustained, irregular choreoathetoid. The movement disappeared with sleep. He had no precipitating factors for GTCS and had mild dysphagia and slurring of speech. He had no cognitive or psychiatric symptoms. No history suggestive of cerebrovascular disease or similar symptoms of movement in the past was recorded. There was also no family history of movement disorder. His past medical history was not significant. Neurological examination revealed his normal neurocognitve assessment but had ill

sustained choreoathetoid movements involving all the body, other aspects of the neurological examination were normal. Admission blood pressure was 130/90 mmHg, and the abdominal and chest examinations were unremarkable. A diagnosis of epileptic encephalopathy involving the basal ganglia was made initially. The result of fasting plasma glucose was 96 mg/dl, while that of serum calcium was 9.5 mg/dL (8.8-10.2), albumin 3.8 g/dL, phosphate 5 mg/dL (2.5-4.9) and parathyroid hormone 31.5 pg/mL (15-65 pg/mL) were all normal. Brain CT-scan revealed symmetrical non enhancing calcifications in both cerebellar hemispheres, both basal ganglia and in both parietal regions (figure 1).

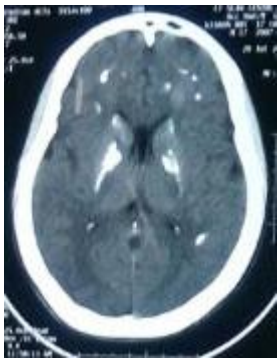

Figure 1.Bilateral basal ganglia calcification

A diagnosis of Fahr's disease with GTCS and choreo- athetoid movement disorder was made. He was placed on tabs haloperidol 5 mg daily and sodium valproate .His seizures were controlled. The chorea athetoid movement responded remarkably and the patient was free of movement disorder by the tenth day on admission and was discharged from hospital after 14 days of hospitalization. Prognosis was explained to parents. He is currently been followed-up in the medical out-patient unit.

Discussion

Fahr's disease (FD) is characterized by sporadic or familiar idiopathic calcification of the basal ganglia, dentate nuclei of the cerebellum, and centrum semiovale.[1] People with FD frequently present with movement disorders such as rigidity, hypokinesia, tremor, choreoathetosis, and ataxia and with frontal subcortical and cortical patterns of behavioral disturbances such as psychosis, mood disorders, and seizures or stroke-like events.[4] 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.[5] Availability of brain CT-scan has increased the number of case reports of intracranial calcification, and brain CT-scan is considered more sensitive than magnetic resonance imaging for finding calcified deposits in Fahr syndrome. Clinical and pathological criteria have been set forth for defining Fahr syndrome.[7] The clinical criteria require bilateral calcification of the basal ganglia with neuropsychiatric and/or extrapyramidal disorders associated with normal calcium and phosphate metabolism; other criteria stipulate seizures, rigidity and dementia with characteristic calcification of the basal ganglia. Our case fulfilled the diagnostic criteria, having presented with movement disorder associated with bilateral basal ganglia calcification. Parathyroid hormone abnormality appears to be the most common definable etiology for calcification of bilateral subcortical nuclei and white matter, but the mechanism of selective deposition of calcium and other trace element in the presence of normal serum parathyroid hormone, serum and cerebrospinal fluid (CSF) calcium and phosphate is not yet known.[5] The most common manifestation of FD is movement disorders (55%), of which accounts for over half of all movement disorders while hyperkinetic movement disorders (chorea, tremor, dystonia, athetosis and orofacial dyskinesia) account for the rest. Our index case had choreo atheoid movements. Cognitive impairment is the second most common manifestation, followed by cerebellar impairments and speech disorders. Overlap of neurologic manifestation such as hypokinetic movement disorder associated with cognitive impairment and cerebellar signs are often seen.[5] The course of FD is progressive as calcium deposition generally begins in the third

decade of life, with neurological deterioration two decades later.[8] FD may also occur in young adolescents and young adults, as in our case. Although calcification can involve other structures, globus pallidus is by far the most common site, with lateral pallidus been more affected than the medial. [9] The exact pathological process initiating the calcifying changes is not known, it may reflect slowly progressive metabolic or inflammatory processes in the brain, which subsequently calcifies and is probably responsible for the neurologic deficit observed.

Several approaches based on diverse biological theories and small scale clinical experiences have been proposed for management of FD. Pharmacological treatment should be used to improve anxiety, depression, and obsessive compulsive disorder and to alleviate dystonia. Seizures and movement disorders in Fahr's syndrome which are related to the parathyroid disorder can be resolved with the correction of phosphate and calcium levels for e.g. treatment with alpha hydroxy vitamin D3 and corticosteroids reversed neurological deficits.[4] Clonazepam and atypical antipsychotic also offer a distinct advantage in treating patients with Fahr's syndrome.[5]

Fahr's disease could manifest with hyperkinetic movement disorder including choreiform movements and seizure disorders. It may alsooccur in adolescents and young adults. High index of suspicion can help in early diagnosis and management of Fahr disease especially associated with hypoparathyroidism.

Conflict of Interest: None
Funding: None

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