FAHR’S DISEASE - A RARE CAUSE FOR SEIZURES - A CASE REPORT

Manohari R    Ramkumar S
GOVERNMENT MOHAN KUMARAMANGALAM MEDICAL COLLEGE, SALEM

Abstract:

German neurologist, Karl Theodor Fahr was the first to throw light on this disease entity in 1930. The disease has classically been described in individuals aged between 40-60 years. However, it may rarely be seen in children in whom it may cause choreathetoid movements.(1,2) These patients may develop mental and motor disability, tetany, paraesthesia, cataract, epileptic syncope and intracranial calcification.(3) Aetiology is not clear. A familial transmission has been reported in literature. In addition, Fahr’s disease may not be accompanied by any neurological signs in a small subset of patients.

Case report:

A 14 year girl with a history of a seizure disorder since 6 years of age presented with breakthrough seizures. She was poorly compliant with the anti epileptic medications she was started on during childhood. There was family history of seizures in the younger sister as well as the mother. No other family members had similar ailments. Patient did not give any history of bone pains, behavioural disturbances, abdominal pain, visual disturbances or tetanic spells. There was no history of birth asphyxia, prolonged labor or delayed attainment of milestones. She did not give any history of previous radiation exposure.

Her scholastic performance at school was good. The general physical examination of the patient was unremarkable.
Cardiovascular and respiratory system examination was unremarkable. Abdominal examination revealed no organomegaly. Direct ophthalmoscopic examination of the fundus showed no anomalies. CNS evaluation revealed no deficits clinically.

Laboratory investigation revealed a normal hemogram, a normocytic normochromic picture on peripheral smear study. Her urine analysis, LFT, RFT, blood sugar and thyroid function testing were all normal. Serum Na+ 139 (135-145 mEq/L), K+ 4.1 (3.5-5 mEq/L), Ca++ 9.2 (9-11mEq/L) and PO4--- 4 (upto 4.5mEq/L). Serum parathormone levels were normal. CSF Analysis was as follows: sugar 65 mg%, protein 54 mg% with normal cytology. An ultrasonogram of the neck revealed no thyroid or parathyroid anomalies and abdominal ultrasound was also normal. Chest X Ray was also not contributory. HIV testing was also negative.

Plain films of CT Brain showed bilateral basal ganglia and bilateral dentate nucleus of cerebellum calcification. An EEG revealed bilateral frontal intermittent slowing in an otherwise normal background activity. No spikes or sharp wave patterns were noted. The younger sister showed similar distribution of intracranial calcification on CT scanning. The patient was started on anti-epileptic drugs. The patient has been compliant to treatment and has subsequently remained seizure free over a 4 month period.

An Initiative of The Tamil Nadu Dr M.G.R. Medical University
Discussion

Fahr’s disease, synonymous with bilateral striopallidodentate calcinosis (BSPDC) is a rare syndrome characterized by symmetrical calcification over the basal ganglion and dentate nucleus.\(^4\) The basal ganglia have been described to be the most common site of involvement.

Various presentations of Fahr’s disease have been described ranging from an extrapyramidal movement disorder to behavioural disturbances or other neurological manifestations that may include a cognitive impairment, cerebellar signs, speech disorders, pyramidal signs, gait disorders and sensory changes.\(^5\) The clinical diagnosis of Fahr’s disease is based on the combination of clinical features, brain imaging and on an exclusion of other causes of the intracranial calcification.\(^6\) This is a rare disease with the usual age of presentation being 40-60 years. However our patient presented at a younger age.

Epilepsy can occur in Fahr’s disease.\(^7\) This was the presenting feature in our patient. Cases presenting as complex partial seizures with secondary generalization have been described in literature.\(^8\) However our patient presented with a generalised tonic clonic form of seizures.

Both sporadic and familial forms of the disease have been described, with an autosomal recessive as well as dominant pattern of inheritance.\(^9,10\) This patient had a positive family history in the sister who also had similar pattern of intracranial calcification evident on imaging. Therefore, this could be a familial form of Fahr’s disease. Genetic heterogeneity and an anticipatory effect have been observed. A multigenerational family with linkage to the IBGC1 of chromosome 14 had been identified; however the causal gene remains still unknown. Genetic studies on other families have not been able to replicate this result.\(^11\) No prenatal or genetic test is available for genetic counseling at present.

CT scanning has continued to remain the most effective screening tool for adult relatives. However, false negative results may still occur. The minimum age at which a negative CT scan can exclude the disease has not been established yet.\(^4\)

A clinical diagnosis of Fahr’s disease relies on a combination of clinical features, brain imaging, and exclusion of other causes of intracranial calcification. Symmetric and extensive calcification is the usual typical and conspicuous imaging finding. The basal ganglia, dentate nuclei of the cerebellum, and centrum semiovale are the commonly involved sites. Other causes of intracranial calcification such as parathyroid disorders, vascular lesions, and infectious diseases like toxoplasmosis have to be ruled out in a patient with suspected Fahr’s Disease.
Literature has described patients presenting with movement disorders as well as patients presenting with Parkinson like features resistant to conventional therapy with similar imaging findings. Our patient had presented at an early stage. This patient may require further follow up and evaluation for possible development of movement disorder and behavioural disorders. This case is presented for the early presentation of Fahr’s Disease and its presentation as seizures with no evidence of a movement disorder.

References:


