International Journal of Medical and Health Research
ISSN: 2454-9142
www.medicalsciencejournal.com
Volume 4; Issue 5; May 2018; Page No. 26-27

A case report of Fahr's disease in a young male: Rare presentation of a rare disease

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Abstract
Fahr's disease is rare clinical entity characterized by symmetrical and bilateral intracranial calcification. It is usually reported in late adult and old age. We report a rare case of a 25 year old male presenting with seizures and cerebellar signs, with calcifications in bilateral basal ganglia and cerebellar hemispheres on computed tomography of the head, no biochemical or endocrinological abnormalities and no family history. His seizures responded well to antiepileptic but cerebellar features did not improve.

Keywords: Fahr's disease, bilateral intracranial calcification, young male, seizures, cerebellar features

Introduction

Fahr's disease is a rare neurodegenerative disease characterized by sporadic or familial idiopathic basal ganglia calcification that is associated with many neurological, psychiatric and cognitive abnormalities [1]. It should be differentiated from Fahr's syndrome that refers to calcification secondary to some underlying metabolic, infective or other cause [2].

Case report

We present a case of a 25 year old male presenting with history of seizures since 2 years and recurrent falls since few days. Patient was already on single antiepileptic drug but seizures were not controlled. There was no family history of similar or any other neurological disorder. His general physical examination was normal. On neurological examination, cerebellar signs including gait instability, dysdiadokinesia and dysmetria were present. Mental status examination was unremarkable. Laboratory investigations including complete blood count, renal, liver and thyroid functions, serum calcium, phosphate, magnesium, parathyroid hormone, vitamin D were within normal limits (Table 1).

Table 1

Investigations	Patient's Values	Reference Range
Hemoglobin	13 gm%	12-17gm%
Total Leucocyte Count	6800/ µl	4000-10000/µl
Random blood sugar	120 mg/dl	70-140 mg/dl
Blood Urea	19 mg/dl	12-20 mg/dl
Serum Creatinine	0.8 mg/dl	0.7-1.3 mg/dl
Serum TSH	2.58 µIU/ml	0.22-4.4 µIU/ml
Free T3	300 pg/ml	260-480 pg/ml
Free T4	1 ng/dl	0.7-1.53 ng/dl
Aspartate transaminase	22U/l	12-38 U/l
Alanine transaminase	34 U/l	7-41U/l
Serum Calcium	10 mg/dl	8.5-10.5 mg/dl
Serum Magnesium	2.1 mg/dl	1.6-2.6 mg/dl
Serum Phosphorus	3.2 mg/dl	2.5-4.5 mg/dl
Serum intact PTH	5.2 pmol/l	1.3-6.8 pmol/l
Vitamin-D levels	35 ng/ml	30-80 ng/ml

Workup for infectious, autoimmune and inflammatory conditions revealed no abnormalities. EEG was suggestive of epilepsy. Non contrast computed tomography of the brain (Figure 1) revealed symmetric and extensive calcification in the basal ganglia, thalamus, dentate nucleus, subcortical white matter of cerebellar hemispheres.

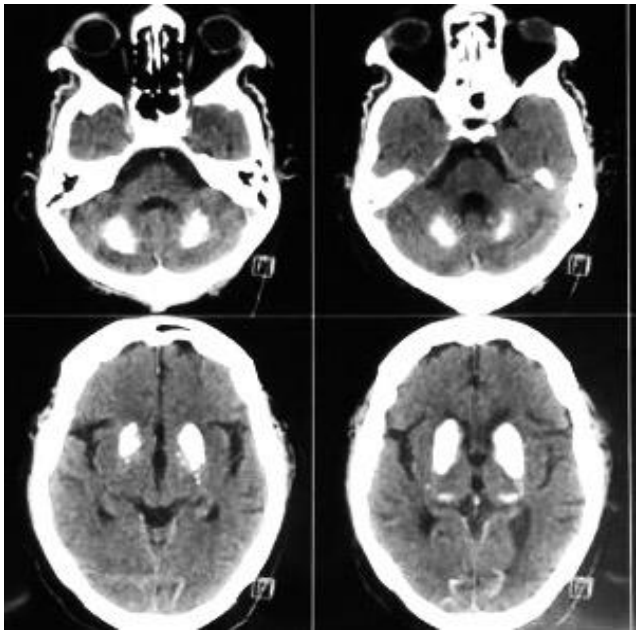

Fig 1: CT showing extensive calcification in basal ganglia, thalamus, dentate nucleus, subcortical white matter of cerebellar hemispheres

Patient's seizures responded well to two antiepileptic drugs however cerebellar features did not improve.

Discussion

Fahr's disease was first described by German neurologist Karl Theodor Fahr in 1930 [3], although Delacour in 1850 [4] and Bamberger in 1855 [4] had reported similar cases. Exact incidence of the disease is unknown. Symptomatic onset for Fahr's disease tends to be between age 40 and 60 [6]. Most of the cases are familial however sporadic forms have also been documented [7]. About 60% of cases of Fahr's diseases have been linked to mutations found in four genes. Two (SLC20A2, XPR1) have been linked to phosphate metabolism, while the other two (PDGFB, PDGFRB) are associated with blood–brain barrier integrity and pericyte maintenance [8]. Patients with Fahr's disease can present with parkinsonism, speech disorder, cognitive impairment, psychosis, paresis and cerebellar features. Our patient was a 25 year old male presenting with seizures and cerebellar features, with no family history.

Fahr's disease should be differentiated from Fahr's syndrome.[2] Diagnosis of Fahr's disease is made from clinical features, neuroimaging and by excluding other causes of intracranial calcification. Calcification can be found in the globus pallidus, putamen, caudate nucleus, thalamus, cerebellum, corona radiata, and subcortical white matter [9]. In our case diagnosis was confirmed by neuroimaging that showed extensive intracranial calcification in bilateral basal ganglia, thalamus, dentate nucleus and subcortical white matter of cerebellar hemispheres. We excluded all the common secondary causes of intracranial calcification like hypoparathyroidism, hypervitaminosis D, tuberculosis, neurocysticercosis, brucellosis, HIV, history suggestive of connective tissue disease or radiotherapy etc. Fahr's disease should also be distinguished from incidentally found basal ganglia calcification in elderly population.

There is no specific treatment for Fahr's disease. Various pharmacological therapies have been tried including levodopa, lithium, trihexiphenidyl, antiepileptic medication for seizures, antichoreic medications such as sulpiride or tetrabenazine, antipsychotics for psychiatric features and botulinum toxin for focal symptoms [10]. Recently bisphosphonates have been used in Fahr's disease but need further trials [11]. Our patient responded well to two antiepileptics and other supportive measures, however his cerebellar features did not improve.

Conclusion

Fahr's disease is a rare neurodegenerative disease with varied presentations. It is difficult to diagnose clinically. CT head and biochemical investigations helps in establishing the diagnosis of this rare disease.

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