Review Article

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A Case Report of Fahr's Syndrome Presenting with Seizures

Rahul Deep G[1], Yashas Ullas L[2], Sahana N Gowda[3], N Rachegowda[4]

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Abstract

Introduction: Fahr's syndrome is a rare, degenerative neurological idiopathic condition characterized by basal ganglia calcification. It often appears between 40 - 60 years of age and can occasionally be seen in children.

Case Report: In our case, it was a 40 year old female who presented with seizures and lab investigation showed-hypocalcemia & hypothyroidism. On Computed Tomography (CT) study, bilateral symmetrical calcifications in centrum semiovale, corona radiata, subcortical white matter of fronto parietal regions, caudate & lentiform nuclei and cerebellum were noted.

Conclusion: Patients presenting with seizures and laboratory investigations showing hypocalcemia and abnormal parathyroid hormone levels, detailed neurological clinical examination and Computed tomography should be performed to rule out any abnormal intracranial calcification.

Keywords: Fahrs syndrome; Seizures; Basal ganglia calcifiaction; Neurological disease.

Key Messages: Provide appropriate messages of about 35-50 words to be printed in centre box.

INTRODUCTION

Fahr's disease or Fahr's syndrome is a rare, degenerative neurological idiopathic condition characterized by basal ganglia calcification. Description of this disease was first attempted by Karl Theodor Fahr, a German neurologist in 1930.[1] Convulsive seizures, neuropsychiatric, extra-pyramidal & cerebellar symptoms, dementia, speech disorders or Parkinsonian features may be the clinical presentation.

Author's Affiliation: [1]Assistant Professor, [2,3] Postgraduate, [4]Professor, Department of Radiodiagnosis, Sri Devaraj URS Institute of Medical Sciences, Kolar 563101, Karnataka, India.

Corresponding Author: Yashas Ullas L, Postgraduate, Department of Radiodiagnosis, Sri Devaraj URS Institute of Medical Sciences, Kolar 563101, Karnataka, India.

E-mail: dryashasullas@gmail.com

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This disease often presents between 40-60 years of age.[2,3] It can be occasionally seen in children, presenting with choreoathetotic movements.[2,4] Globus pallidus is the most common site of calcification in basal ganglia but may be seen in putamen, caudate nucleus, internal capsule, dentate nucleus, thalamus, cerebellum and cerebral white matter.

Histologically, proteins and polysaccharides are present in these deposits which are seen in the perivascular space and in media layer of the small vessels. The pathogenesis may be secondary to impairment of the blood brain barrier or to a neuronal calcium phosphoric metabolism disorder; however, exact pathogenesis is unknown.[3,5]

Case Report A 40 year old female patient presented with two episodes of seizures and post-ictal headache with generalized weakness. Patient is a known case of hypothyroidism since 3 years with no other co-morbidities. Patient is on thyroxin supplement. Lab investigations also showed

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hypocalcaemia and hypoparathyroidism. Other lab investigations showed normal levels of complete blood count, sedimentation rate, fasting blood glucose and blood electrolytes.

Imaging Findings

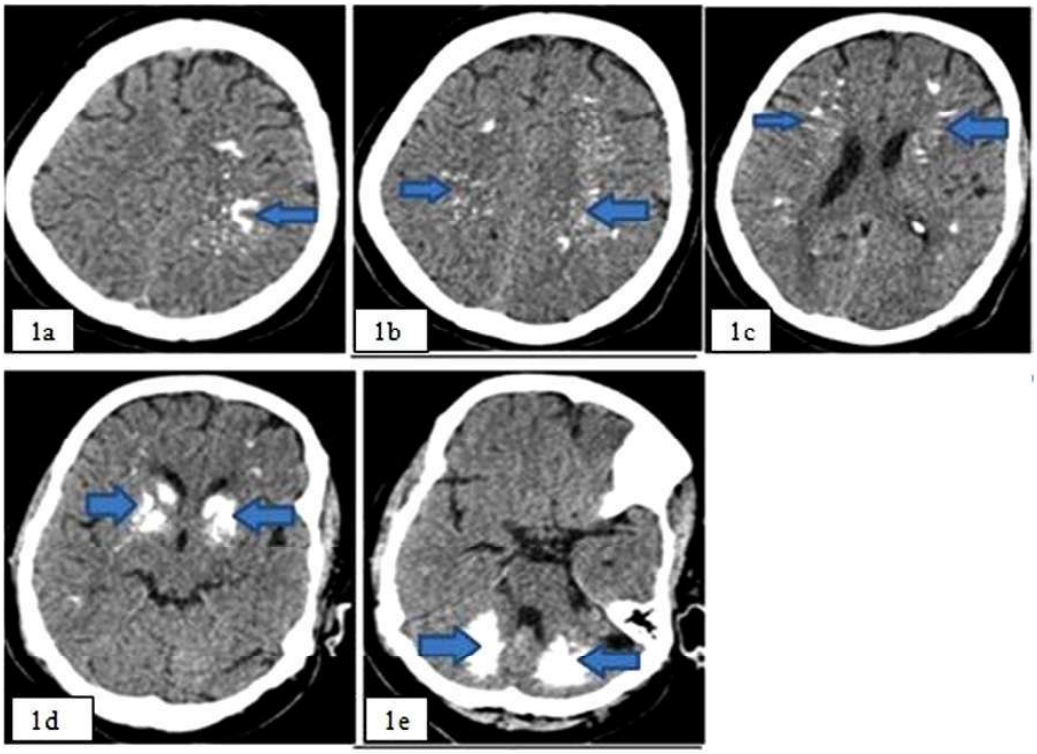

Fig. 1: CT scan axial section brain window shows bilateral symmetrical calcifications noted in centrum semiovale (Fig. 1a), corona radiate (Fig. 1b), subcortical white matter of fronto-parietal regions (Fig. 1c), caudate nuclei (Fig. 1d), lentiform nucleus (Fig. 1e) and cerebellum (Fig. 1f). Rest of the brain parenchyma appears unremarkable.

DISCUSSION

Idiopathic basal ganglia calcification known as bilateral Striopallido Dentate Calcinosis (BSPDC) or as Fahr's Syndrome, is a rare neurodegenerative disorder with unknown prevalence. Fahr's disease is characterized by bilateral, symmetrical, intra cranial calcification with a predilection for dentate nuclei and basal ganglia. The descriptive terminology, BSPDC is because of symmetrical involvement of these nuclei.[6] This disease commonly presents at 40-60 years of age. This condition is a inherited neurological condition which may lead to progressive dystonia, neuropsychiatric manifestations and Parkinsonism. According to Fahr's Disease Registry, the most common clinical presentation accounting for about 55% of cases are movement disorders, among which: 57% cases presented as Parkinsonism, 19% cases of cases presented with chorea, 8% cases presented with tremors, 8% cases presented with dystonia, 5% cases presented with athetosis and 3% cases presented with orofacial dyskinesia. Cognitive impairment, cerebellar signs, speech disorders, pyramidal signs, psychiatric features, gait disorders and sensory changes are the other neurological manifestations of this disease.[7] The combination of clinical features, brain imaging and on an exclusion of other causes of intracranial calcification forms the basis for the clinical diagnosis of Fahr's disease.[8]

The typical imaging findings of Fahr's disease i.e, the symmetric and extensive parenchymal calcification of brain was seen in our case. Fahr's disease represents a heterogeneous group of disorders. Intracerebral calcification can be associated with disorders of calcium metabolism, hypoparathyroidism, pseudohypoparathyroidism and hyperparathyroidism. Infectious diseases like syphilis & toxoplasmosis and inflammatory illnesses like SLE can also be the causes of

intracranial calcification.[9]

In familial cases, genetic studies have demonstrated an autosomal dominant inheritance. The causal gene is still unknown; however, one multigenerational family with a linkage to the IBGC1 of chromosome 14 has been identified.[10]

Benke et al., studied brain metabolism in a patient with Fahr's disease who presented with a predominant frontal lobe syndrome and dementia, by using brain positron emission tomography with flurodeoxyglucose. Massive reduction of the glucose metabolism in bilateral frontal lobes and basal ganglia was noted, which correlated with the clinical picture of disinhibition and a personality change.[11]

The most effective screening tool is computed tomography scan. No genetic or prenatal tests are available for genetic counseling. No minimum age been established yet to suggest the exclusion of the disease with a negative CT scan. Doing imaging scan of the parents and other kindred is more reliable than their clinical screening to suggest the exclusion of the disease.[8]

The treatment for this disease is symptomatic support. For the psychiatric symptoms, atypical antipsychotics are preferred because of coexistence of the extra pyramidal syndrome in these groups of patients.

CONCLUSION

Patients presenting with seizures and laboratory investigations showing hypocalcemia and abnormal parathyroid hormone levels, detailed neurological clinical examination and Computed tomography should be performed to rule out any abnormal intracranial calcification.

Conflict of Interest: Nil

REFERENCES

1. Asokan A. Fahr's Syndrome-An Interesting Case Presentation. Journal of clinical and diagnostic research. 2013.
2. Murat Gülsün , Ali FuatBaykız, SerdarKabata, Hasan Belli. Fahr's Syndrome-Three cases presenting with psychiatric signs. Eur J Gen Med. 2006;3:35-40.
3. Kotan D, Aygul R. Familial Fahr's disease in a Turkish family. South Med J. 2009;102:85-6.
4. Billard C, Dulac O, Bouloche J, Echenne B, Lebon P, Motte J. Encephalopathy with calcifications of the basal ganglia in children. A reappraisal of Fahr's syndrome with respect to 14 new cases. Neuropediatrics. 1989;20:12-9.
5. Malik R, Pandya VK, Naik D. Fahr's disease: A rare neurodegenerative disorder. Indian J Radiol Imaging. 2004;14:383-4.
6. Manyam BV. Bilateral strio-pallido-dentate calcinosis: a proposed classification of genetic and secondary causes. Mov Disord. 1990;5:94.
7. Manyam BV, Walters AS, Narla KR. Bilateral striopallido dentate calcinosis: clinical characteristics of patients seen in a registry. Mov Disord. 2001;16:258-64.
8. Lam JS, Fong SY, Yiu GC, Wing YK. Fahr's disease: a differential diagnosis of frontal lobe syndrome. Hong Kong Med J. 2007;13:75-7.
9. Manyam BV. What is and what is not 'Fahr's disease'. Parkinsonism Relat Disord. 2005;11:73-80.
10. Geschwind DH, Loginov M, Stern JM. Identification of a locus on chromosome 14q .for idiopathic basal ganglia calcification (Fahr's disease). Am J Hum Genet. 1999; 65:764-2.
11. Benke T, Karner E, Seppi K, Delazer M, Marksteiner J, Donnemiller E. Subacute dementia and imaging correlates in a case of Fahr's disease. J Neurol Neurosurg Psychiatry. 2004;75:1163-5.