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A Rare Fahr's disease Diagnosed in Two siblings: A case report

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Abstract

Fahr disease is a rare neurological disorder mostly presented with movement disorder, extrapyramidal symptoms and chronic cognitive dysfunction. They seldomly presented with seizures.

We report cases of Fahr disease in two siblings, aged 17 and 15 years, who presented with seizures.

This case highlights that Fahr disease may present early with atypical features and should be considered in young patients with unexplained seizures.

Introduction

Fahr disease is a rare inherited neurological disorder marked by bilateral basal ganglia calcifications, unlike Fahr syndrome, which results from an identifiable underlying cause. (1, 2).

Fahr disease is usually presents at middle age group (third to fifth decade) and rarely present in the young. There is a unique report of this condition from a three years old child with implicated with infancy onset (9). Most of these patients with fahr disease present with movement disorders, extrapyramidal symptoms and chronic cognitive deterioration. Seizure and epilepsy are ones of the rarest manifestation of this illness (3, 4).

Recently a 60 years old man from Ethiopia was reported with diagnoses of fahr disease after presented with convulsive (5). A couple of Fahrs disease cases reported from Africa includes a 45 yrs old patient presented with seizure from morocco (6), a 66 yrs old man with recurrent episodes of change in mentation and a 9 years old female with seizure from Nigeria (7, 8). Fahr disease in two siblings is also documented from Angola, one aged 46 and the other aged 42, both with psychiatric symptoms as the initial presentation and from china in a 31 yrs old man and his sister 29 yrs old after they presented with progressive movement problems (10, 24). When it comes to documented diagnoses of fahr disease in siblings at age of 17 and 15 years, we believe this is the first report from East Africa.

Case presentations

Case one (01)

Clinical history

Here we report a 17 years old female adolescent from Somaliland came to our hospital with complaint of an episodes of abnormal body movement for total of 10 years duration. The episodes are characterized by abnormal versive head shifting to one side, tonic extension of extremities followed by flexion and extension movement of upper extremities lasting for two minutes. In association she had up rolling of eyes, drooling of saliva and non-responsive. These episodes are subsequent to sleep and fatigue for 30 minutes. The symptoms started when she was eight years old and occurred not more than

once per month. Recently in the past 02 months, the episodes then become much frequent about two to three times per week. The last episode was 02 days before presentation. She had previous history of unspecified eye surgery and appendectomy. She has seven sisters and three brothers. One of her younger sister has similar illness (case two).

Timeline -summarized in Table 03 for both cases

Physical findings

Physical examination shows conscious, normal orientation to time, place and person. She had no sign of extremity weakness. Cerebellar examination reveals difficulty to perform tandem walk.

Diagnostic Investigations

We did routine laboratory tests for seizure and did readily available brain CT scan with contrast followed by further evaluation. The laboratory results are unremarkable (Table 01). Brain CT scan shows hyper dense lesions distributed symmetrically over head of caudate nucleus, putamen and dentate nucleus area of cerebellum (Fig. 01). Routine 30 minute electroencephalogram (EEG) did not detect definitive epileptiform discharge.

Table 01

laboratory summary of case one (01) ALT: alanine transaminase, AST: aspartate transaminase, ALP: alkaline phosphatase, TSH: thyroid stimulating hormone, HbSAg: hepatitis B surface antigen HIV: human immunodeficiency virus.

(A) **(B) (C)**

Figure 01: Brain CT scan of case one (01) showing multiple brain calcifications. Figure A: shows Bilateral caudate and putamen calcifications with left frontal subcortical lesion. Figure B: shows symmetrical calcifications over dentate nucleus area of cerebellum and figure C: Post contrast CT scan do not demonstrate enhancement of calcifications.

Case Two (02)

Clinical History

Here we report the second case, who is a 15 years old female (younger sister of case one) adolescent from Somaliland came to our hospital with similar complaint of abnormal body movement episodes but relatively infrequent and less severe for total of 06 years duration. The episodes are characterized by tonic extension of extremities and flexion and extension movement of upper extremities lasting for a minute. In association she had up rolling of eyes drooling of saliva and unresponsive. These episodes are followed by post ictal confusion for 15 minutes. The symptoms started when she was 9 years old and once to twice per month.

Physical examination

Physical examination shows conscious, normal orientation to time, place and person. She had no sign of focal neurologic deficit.

Diagnostic investigations

We did routine laboratory tests for seizure and we did readily available brain CT scan. The

laboratory resulted are unremarkable (Table 02). Brain CT scan shows hyper dense lesions distributed symmetrically over caudate nucleus, putamen & dentate nucleus areas (Fig. 02). Routine 30minute electroencephalogram (EEG) did not detect definitive epileptiform discharge. One of their elder brother had normal non contrast brain CT scan.

Table 02

laboratory summary of case two (02) ALT: alanine transaminase, AST: aspartate transaminase, ALP: alkaline phosphatase, TSH: thyroid stimulating hormone, HbSAg: hepatitis B surface antigen HIV: human immunodeficiency virus. PTH: Parathyroid hormone

(A) **(B) (C)**

(D) (E)

Figure 02: Brain CT scan of case two (02) showing multiple brain calcifications. Figure A: shows bilateral caudate, thalamus, putamen and extensive subcortical white matter calcifications. Figure B shows symmetrical calcifications over centrum semiovale. Figure C shows bilateral dentate nucleus involvement. Figure D: sagittal brain CT showing subcortical, basal ganglia and cerebellum together. Figure E: Post contrast CT scan did not demonstrate enhancement.

Diagnosis and Management

We compile the clinical, laboratory, neuroimaging and EEG finding to look for possible alternative diagnosis. As seizure diagnosis mainly relay on our clinical semiology of those episodes of abnormal body movement, we made diagnosis of Epilepsy and looking for possible association of multiple basal ganglia calcifications with seizure. Therefore, given similar onset of illness, presentation, typical imaging finding, presence of family history and absence of alternative diagnosis we conclude that this is fahr disease with possibility of familial type. We initiate both patients on oral sodium valproate 200mg twice daily and escalated to 400mg po BID a week later.

Outcome and Follow Up

On subsequent follow up after one month we have seen these patients and case one had two episodes of seizure with an interval of a week between episodes (the frequency of seizure is reduced) while case two did not experience any seizure in a month period. We opted for short appointment and follow up, however due to far distance from their residency we unable to follow regularly for prolonged period of time

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Table 03
Timeline of key clinical events of both cases from initial presentation to diagnosis and treatment of Fahrs disease

key clinical events of case 1		**key clinical events of case 1**	
2months before diagnosis (for 10 years)	Intermittent abnormal body movement characterized by flexion and extension of all extremities once or twice per month with post ictal sleepness for 30minutes	Before diagnosis (for 6 years) Infrequent episodes of abnormal body movement characterized by flexion and extension of all extremities with post ictal sleepness for 15minutes	
During preceding two months before diagnosis	Frequent episodes of similar type of abnormal body movement		
One month after diagnosis	two episodes of seizure with an interval of a week between episodes only	One month after diagnosis	No abnormal body movement episodes

Discussion

Fahr disease is most commonly known to be transmitted in an autosomal dominant pattern, sometimes inherited as an autosomal recessive or found sporadically Therefore, the term Idiopathic basal ganglia calcification (IBGC) is less preferred because some genes have been implicated in this condition (11). The alternative term bilateral striopallidodentate calcinosis is also used in literatures. Most importantly this rare neurodegenerative condition is noted by accumulation of calcium deposits; most easily visualized on computed tomography (CT) scan, in different brain regions. These calcium containing deposits most commonly seen in basal ganglia but other brain regions can also involved (like in our cases) (12). The familial type of IBGC, now called "primary familial brain calcification" (PFBC) have several causative genes mutations. These includes four dominant inherited genes (SLC20A2, PDGFRB, PDGFB, and XPR1) and three recessive inherited genes (MYORG, JAM2, and CMPK2). However, around half of patients with PFBC do not demonstrate these genes which implies more PFBC-associated pathogenic variants are yet to be identified (13, 14). Clinical manifestations of fahr disease are variably include one or more features of Parkinsonism, chorea, dystonia, cognitive impairment, or ataxia (15). Other less common presentations include neurobehavioral or psychiatric manifestations (like depression, anxiety and psychosis), ischemic stroke and seizures (16, 17, 18).

Presentation with epileptic seizures is rare. The relationship between seizure and fahr disease is complex. The possible explanation mentioned by scholars are abnormal calcium- phosphorus metabolism contributes to neuronal excitability and secondary to a probable dysfunction of connections between cortico-basal networking and their relationship between the two hemispheres of the brain (19, 20).

Unfortunately, there is no complete recovery for this condition. The treatment is commonly symptomatic and is based on administration of anti-seizure medications, drugs for Parkinsonism and antipsychotic medications based on the clinical scenario and comorbidities (19, 21). Ant seizure medication that has been used in case reports includes oral phenytoin, carbamazepine and sodium valproate (5, 16, and 22). Many literatures mentioned that seizures associated with fahr disease are usually respond well to ant seizure medications (23).

Strengths of the Report

The two cases reported have several strengths, including the comprehensive clinical evaluation of patients presenting with seizures, particularly in the unusual context of seizures occurring in siblings. The cases also involved careful assessment of potential underlying and confounding causes, which helped support the diagnostic consideration of Fahr’s disease. Furthermore, reporting these cases has enhanced the authors’ awareness of this rare condition and may contribute to increased recognition and awareness among healthcare professionals and the wider health community.

Limitations of case report

The major limitations of this report include the inability to confirm the familial nature of Fahr's disease through genetic analysis. We were also unable to trace and clinically evaluate the other siblings, limiting assessment of the extent of familial involvement. Video-EEG was not available, which limited detailed electroclinical characterization of the seizures. In addition, long-term follow-up was not feasible, restricting assessment of the patients' subsequent clinical course and neurological outcomes.

Conclusion

Fahr disease is a rare neurodegenerative disease nevertheless physicians has to consider as possible differential diagnosis in seizure and epilepsy evaluation. The diagnosis has to be considered after careful evaluation of clinical, brain CT scan and EEG findings. Brain MRI usually done in chronic epilepsy; here we highlighted the importance of Brain CT scan for calcification detection.

Patient perspectives

Case 1

Before diagnosis, I had recurrent abnormal body movement for few minutes ; however ; I have only two episodes only since past 1month.

Case 2

Before Diagnosis ,I had intermittent abnormal body movement; however, totally subsided now.

Declarations

Ethics approval and consent to participate:

Written informed consent was obtained from parents of both the patients for publication of this case report and any accompanying images. A copy of the written consent is available for review by the Editor-in-Chief of this journal.

Consent for publication:

The patients provided written informed consent for the publication of this case report.

Competing interests:

The authors declare that they have no competing interests.

CRediT authorship contribution statement

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Author Contribution

Yaarosan Zawde Kibrat: Writing – review & editing, Writing – original draft, Visualization, Validation, Supervision, Resources, Project administration, Methodology, Investigation, Formal analysis, Data curation, Conceptualization.Wubsew Shimelis Guadie: Writing – review & editing, Validation, Resources, Investigation, patient managementNuredin Ahmed Hassen: Writing – review & editing,Validation, Resources, Methodology, Investigation, Data curation. Sisay Gizaw Gabremichael: review and editing, validation, MethodologyMalegna Temesgen Garuma: Writing – review & editing, Validation, Resources, Methodology, Investigation, Data curation. Wondimu Bekele Nega: Writing – review & editing, Visualization, Validation, Methodology, Investigation.

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Availability of data and materials:

All data generated or analyzed during this study are included in this published article.

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Figures

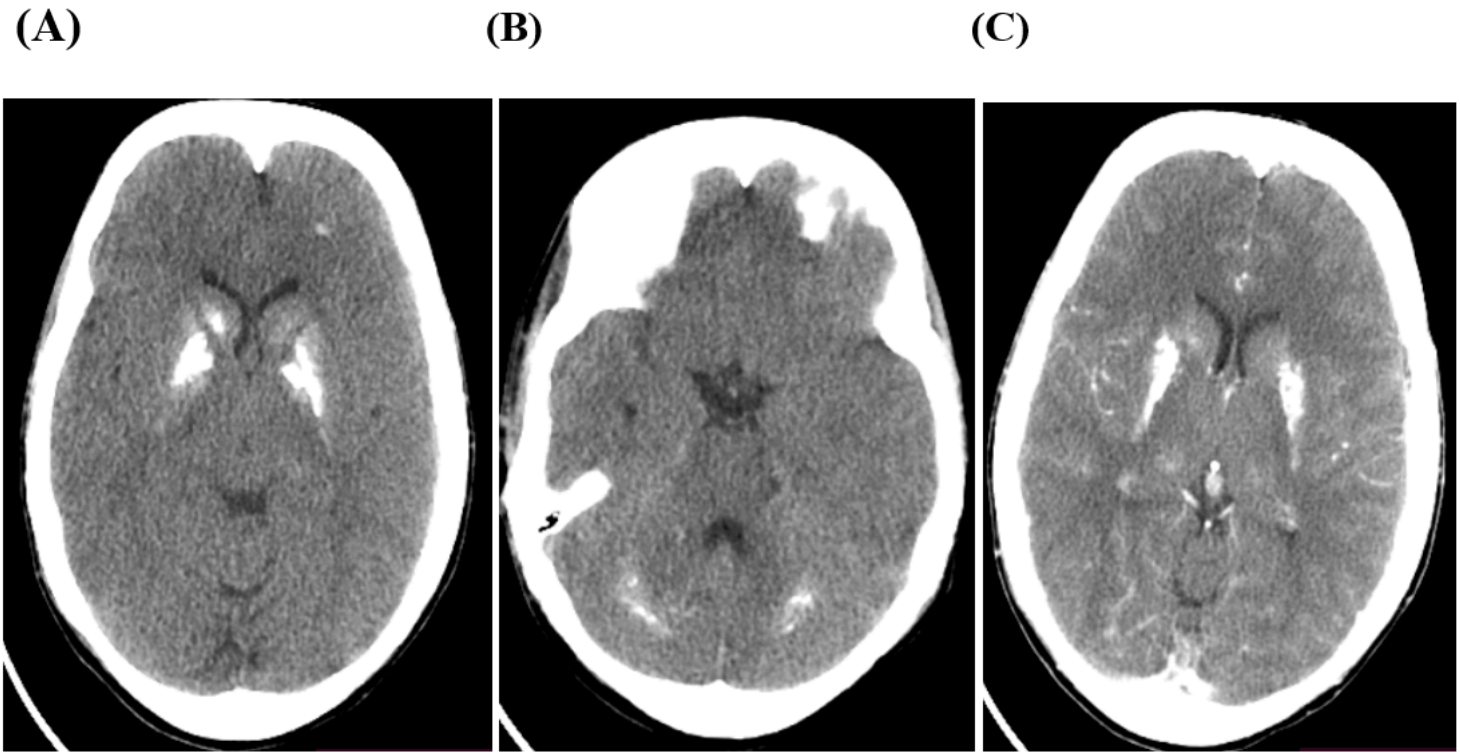


Figure 1

Brain CT scan of case one (01) showing multiple brain calcifications. Figure A: shows Bilateral caudate and putamen calcifications with left frontal subcortical lesion. Figure B: shows symmetrical calcifications over dentate nucleus area of cerebellum and figure C: Post contrast CT scan do not demonstrate enhancement of calcifications.

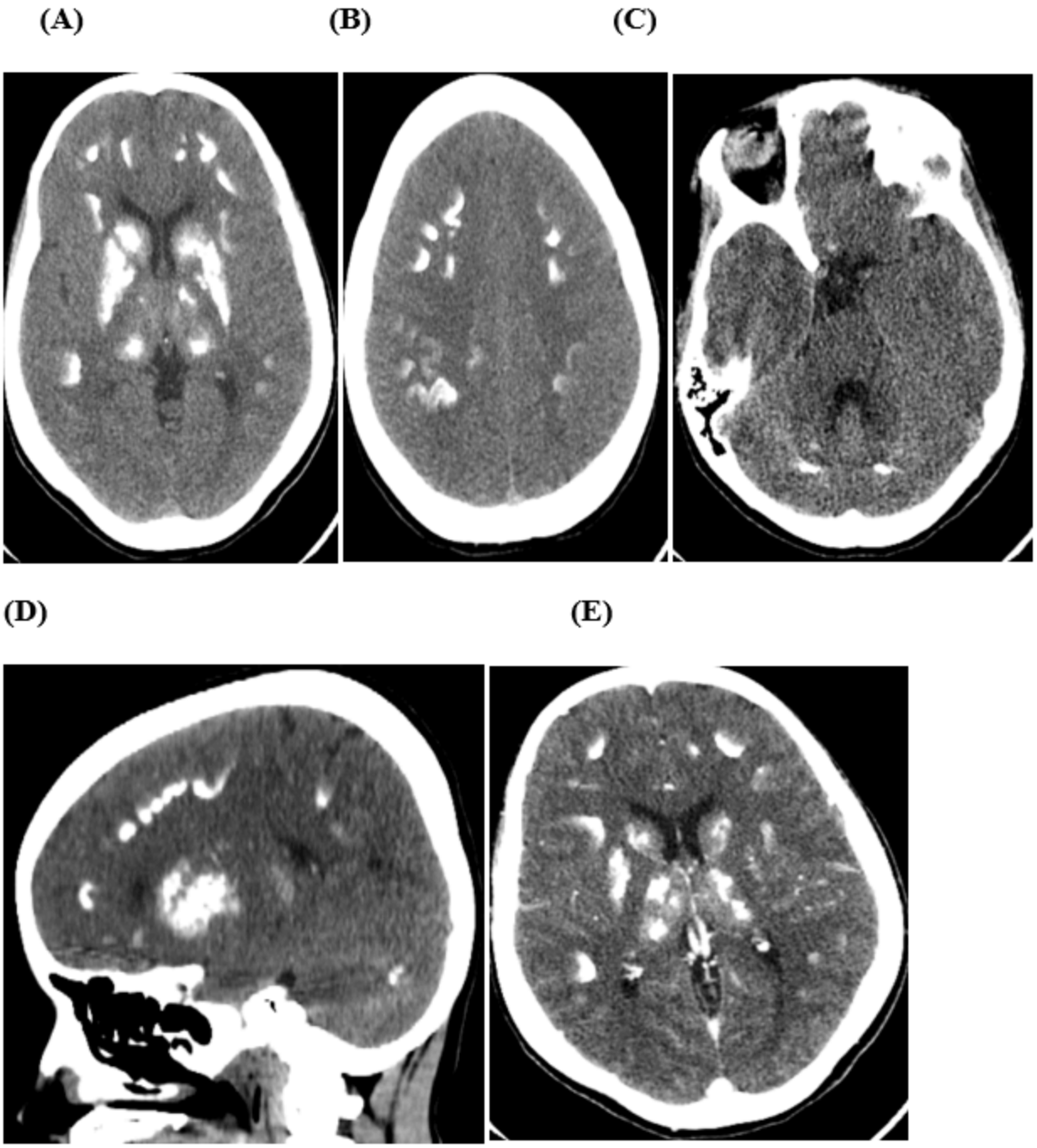


Figure 2

Brain CT scan of case two (02) showing multiple brain calcifications. Figure A: shows bilateral caudate, thalamus, putamen and extensive subcortical white matter calcifications. Figure B shows symmetrical calcifications over centrum semiovale. Figure C shows bilateral dentate nucleus involvement. Figure D: sagittal brain CT showing subcortical, basal ganglia and cerebellum together. Figure E: Post contrast CT scan did not demonstrate enhancement.