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Fahr's Disease Presenting with Seizures and Behavioral Disturbance in a 14-Year-Old Adolescent

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Abstract:

Fahr's disease is a rare neurodegenerative disorder characterized by abnormal brain calcifications, typically presented in middle age. This case report describes an unusual presentation in a 14-year-old boy, highlighting the importance of considering Fahr's disease in pediatric patients. The patient had a 5-year history of seizures, cognitive decline, and behavioral disturbances, along with obesity, short stature, low intelligence, and a clumsy gait. Imaging revealed extensive symmetrical calcifications in the basal ganglia, dentate nucleus, and cerebral white matter, confirming Fahr's disease. The condition can present diverse neurological and psychiatric symptoms, including seizures, movement disorders, cognitive impairment, and mood disturbances. Diagnosis requires excluding secondary causes of brain calcification and is supported by characteristic radiological findings and the absence of biochemical abnormalities. This case underscores the need for early recognition and comprehensive evaluation to optimize management and improve the patient's quality of life, even in the absence of family history.

Keywords: neurodegenerative disorder; dementia; rare psychiatric disorder; vessel calcifications

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Introduction:

Fahr's disease, also known as bilateral symmetrical striopallidodentate calcinosis, is characterized histopathologically by finer cerebral vessel calcifications[1]. The dentate nuclei, basal ganglia, and cerebellar and cerebral white matter are the most affected by these calcifications. Symptoms typically appear between the fourth and sixth decades of life, but cases in children have been reported[2].

Case presentation:

A 14-year-old male adolescent presented to us at the mental health institute in Dhaka with a history of repeated generalized tonic-clonic seizures, deteriorating intelligence, and behavioral changes over the previous 5 years. The boy had no significant medical illnesses in the past. There was no abnormality in the family history, personal history, or drug history. However, the disease forced him to drop out of school after the third grade. Although the boy's mental state examination revealed low intelligence (Weschler Abbreviated Scale for Intelligence), his premorbid temperament indicated he was friendly, social, and amiable. On clinical examination, he was obese and short in stature. There were no other notable findings on the general examination. A clumsy gait was identified during a neurological examination but no other systemic examinations revealed any abnormalities. The laboratory tests showed a normal hemogram. The liver and kidney function tests were both normal. Thyroid hormone levels were normal, but parathyroid hormone levels were elevated at 345 pg/mL (normal range is 15–68 pg/mL). Serum calcium and electrolytes were found to be normal. The EEG revealed characteristics suggestive of generalized epilepsy. A CT scan of the brain revealed symmetrical and multiple irregularly shaped calcifications of the fronto-parietal regions—along the ventricular lining, and in the dentate nucleus consistent with Fahr's disease. To control the child's epilepsy, sodium valproate and carbamazepine were prescribed which was later altered with sodium valproate and levetiracetam. Risperidone was prescribed to help with the behavioral changes. Physical

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rehabilitation was recommended, and despite a negative family history, parents were counseled about the risk of the disease if they conceive again.

Table-I

Clinical features of bilateral basal ganglia calcification

Psychiatric features:
• Cognitive deterioration: dementia, delirium, confusion
• Psychotic symptoms such as hallucinations, delusions
• Catatonia
• Mood disorders: depression, manic symptoms
• Anxiety, panic attacks, and obsessive behaviors
• Irritability
• Aggression
• Personality disorder and personality changes
Somatic symptoms:
• Parkinsonism and movement disorders
• Seizures
• Headache
• Vertigo
• Paresis
• Stroke
• Syncope
• Tremor
• Ataxia
• Dysarthria
• Orthostatic hypotension
Radiologic findings:
• Bilateral symmetrical calcifications of basal ganglia and dentate nucleus
• Other sites of calcifications: thalamus, centrum semi-ovale, cerebellum, and cerebral white matter

Discussion:

Fahr's disease, characterized by basal ganglia calcifications, typically has a slow onset and presents in middle-aged patients with neurological and psychiatric symptoms, though it may remain asymptomatic[3]. Psychiatric symptoms, including cognitive deterioration, mood disorders, and psychotic features, are the initial presentation in approximately 40% of cases, with depressive symptoms being the most common across all age groups[4,5]. Movement disorders are the most frequent manifestation, found in 55% of symptomatic patients, with parkinsonism (57%), chorea (19%), tremor (8%), dystonia (8%), athetosis (5%), and orofacial dyskinesia (3%) being reported[6]. Symptomatic patients exhibit significantly more calcification than asymptomatic patients[6]. Childhood seizures are also noted as a neurological symptom[7]. Genetic mutations in six genes (SLC20A2, PDGFRB, PDGFB, XPR1, MYORG, JAM2) are linked to primary familial brain calcification (PFBC)[8–13]. Additionally, bilateral symmetric calcifications can result from calcium metabolism abnormalities, systemic diseases (e.g., lupus), toxins (e.g., carbon monoxide), encephalitis, radiation, anoxia, Kearns-Sayre syndrome, and Cockayne's syndrome[14–16].

Fahr's disease diagnosis requires evidence of bilateral basal ganglia calcification, progressive neurological or neuropsychiatric manifestations, symptom onset typically in the fourth or fifth decade (though earlier onset is possible), absence of biochemical abnormalities or features suggesting mitochondrial, metabolic, or systemic disorders, and no association with infection, trauma, or toxins; a family history consistent with autosomal dominant inheritance can also support diagnosis[3, 5]. CT scan imaging is more sensitive than MRI in detecting basal ganglia calcifications[17].

Our patient fits Fahr's disease diagnostic criteria. The patient appeared with repeated seizures, deteriorating intelligence, and escalating behavioral abnormalities with a negative family history. A clumsy stride was seen during the neurological evaluation, but no biochemical abnormalities were found to support a secondary link. In addition, the results of the CT scan revealed many symmetrical calcifications of irregular shapes in the fronto-parietal areas, along the ventricular lining, and in the dentate nucleus.

The treatment of Fahr's disease focuses on symptom management and improving quality of life, as there is no cure[18]. Addressing the underlying illness process may slightly improve neuropsychiatric symptoms, though the prognosis remains uncertain and independent of calcification severity[19]. Antipsychotics like haloperidol or lithium carbonate can benefit patients with psychotic symptoms[20].

Author contribution

Dr. Tumpa Indrani Ghose conceived and designed the study, conducted research, provided research materials,

and collected and organized data. Dr. Sayedul Ashraf Kushal analyzed and interpreted the data. Dr. Mohidul Islam, Zunayed al Azdi, and Saiful Islam Saif wrote the initial and final draft of the article and Yahia Md. Amin provided logistical support. Zunayed Al Azdi and Saiful Islam Saif supervised and guided the first author to complete the research. All authors have critically reviewed and approved the final draft and are responsible for the content and similarity index of the manuscript.

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Ethical Approval Statement

There is no ethical issue.

Conflict of interest statement

As stated in the conflict of interest letter, we have no conflicts of interest.

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