Case Report

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A Case Report of Rare Syndrome: Fahr's Syndrome

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Abstract

Fahr's syndrome is a rare neurological disorder characterized by abnormal deposits of calcium in the brain, particularly in the basal ganglia. This can lead to various neurological symptoms such as movement disorders, cognitive impairment, and psychiatric manifestations. The exact cause of Fahr's syndrome is not well understood, and treatment typically focuses on managing symptoms. The study comprehensively examines the clinical presentation, ranging from cognitive impairments to motor disturbances, and investigates potential etiological factors, including genetic predispositions. Existing literature on diagnostic approaches, management strategies, and the challenges encountered in treating Fahr's syndrome is critically analyzed.

Keywords: Fahr's Syndrome; Movement disorders; Cognitive impairment; Psychiatric manifestations; Neurological; Abnormal.

INTRODUCTION

Fahr's syndrome, also known as bilateral striopallidodentatecalcinosis, is a rare neurological disorder characterized by abnormal deposits of calcium in the brain, particularly within the basal ganglia, thalamus, and dentate nucleus.

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These calcifications can lead to neurological dysfunction and various symptoms. Common symptoms of Fahr's syndrome include:

- Dementia
- Headache
- Deterioration of motor function
- Dysarthria (poorly articulated speech)
- Spasticity (stiffness of the limbs) and spastic paralysis
- Eye impairments
- Athetosis (involuntary, writhing movements)
- Dystonia (disordered muscle tone)
- Chorea (involuntary, rapid, jerky movements)
- Seizures
- Mask-like facial appearances
- Shuffling gait
- "Pill-rolling" motion of the fingers

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DIAGNOSTIC CRITERIA:

- Bilateral calcification of the basal ganglia can be seen upon neuroimaging. Other brain regions are also observed.
- Progressive neurologic dysfunction, which generally includes movement disorders and/or neuropsychiatric manifestations.
- Absence of biochemical abnormalities & somatic features suggestive of a mitochondrial or metabolic disease.
- Absence of an infectious or traumatic cause.
- Inheritance family history of autosomal dominant.

CASE REPORT

A 78-year-old male patient presented to the Emergency department with complaints of abnormal staring gaze since a week, associated with multiple episodes of vomiting. Patient was disoriented and confused, also unable to perform day to day activities.

Patient is a known case of CAD with ICMP, LV Dysfunction, CKD.

On clinical examination, patient was disoriented to his surrounding and to people and confused. On primary assessment, the vitals of the patient were:

Respiratory Rate: 18/min

SpO2: 89% on room air

SpO2: 98% on 3L O2

Pulse Rate: 78bpm

Blood Pressure: 110/70 mmhg

Peripheral Pulses: Palpable

Temperature: 98.5F

Random Blood Sugar: 123 mg/dL

EKG: Normal Sinus Rhythm

Pupils: Bilateral Eye Normal in size and Normally Reactive

The provisional diagnosis was to rule out any acute bleed or infarct or lesion in brain and seizures, to prevent seizures episode Inj. levipril 1gm was administered to the patient. Normal findings were received in Arterial blood gas along with initial blood investigations were sent, NCCT HEAD suggestive of calcification of the corpus striatum, periventricular white matter, sub cortical white matter in occipital lobes and deep portion of cerebellum. Neurological advised was taken after initial treatment and investigations. Advised admission for further evaluation

DISCUSSION

Fahr's syndrome is a rare neurological disorder characterized by abnormal calcium deposits in the brain, primarily in the basal ganglia. This condition leads to a diverse range of symptoms, including movement disorders, cognitive impairment, and behavioral changes. While the exact causes are not fully understood, genetic factors, metabolic dysfunction, and secondary causes are implicated. Diagnosis involves clinical evaluation and neuroimaging, with treatment focusing on symptom management. The rarity of the syndrome and its varied presentation pose diagnostic challenges, and ongoing research aims to unravel its complexities for improved understanding and potential therapeutic advancements.

MANAGEMENT

The management of Fahr's syndrome is primarily symptomatic, aiming to alleviate specific neurological and cognitive symptoms associated with abnormal brain calcifications. Treatment often involves a multidisciplinary approach, including neurologists, physical therapists, and other specialists. Medications may be prescribed to address movement disorders, cognitive impairment, or psychiatric symptoms on a case-by-case basis. Physical therapy can be beneficial in managing motor symptoms and enhancing overall mobility. As there is currently no cure for Fahr's syndrome, the focus remains on improving the quality of life for affected individuals through targeted symptom management and supportive care. Regular monitoring and adjustments in treatment strategies may be necessary to address the evolving nature of the condition.

CHALLENGES AND COMPLICATIONS

The treatment of Fahr's syndrome poses significant challenges due to the complexity of its symptoms and the absence of a definitive cure. Managing the diverse neurological manifestations, including movement disorders and cognitive impairment, requires a tailored approach for everyone. Complications arise from the progressive nature of the disorder, necessitating ongoing adjustments to treatment plans. Additionally, the rarity of Fahr's syndrome

contributes to limited research on standardized therapeutic interventions. Psychosocial challenges for both patients and caregivers further compound the difficulties in providing comprehensive care.

CONCLUSION

Treating Fahr's syndrome is inherently challenging due to the absence of a cure and the progressive nature of the disorder. Current approaches focus on symptom management through medications, physical therapy, and supportive care. However, the lack of standardized treatment protocols, coupled with the diverse and evolving symptoms, complicates therapeutic interventions. Ongoing research is crucial for a deeper understanding of Fahr's syndrome, with the hope that future advancements will offer more targeted and effective strategies. The multidisciplinary nature of care and the need for continuous adaptation underscore the complexities involved in addressing this rare neurological condition.

REFERENCES

1. NCBI - https://www.ninds.nih.gov/health-information/disorders/fahrs-syndrome#:~:text=-Fahr's%20syndrome%20is%20a%20rare,Seizures
2. American Journal of Case Report - https://amjcaserep.com/abstract/index/idArt/913382
3. BJM Journals - https://casereports.bmj.com/content/2013/bcr-2013-201556