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Case Report

A Rare Case of Fahr Disease in a Child: Case Report

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Abstract

Fahr's disease is an uncommon neurodegenerative condition especially in the pediatric group, identified by calcium accumulation in the Basal ganglia and Dentate nuclei of the Cerebellum. Patients may exhibit various symptoms, including seizures, extrapyramidal symptoms, and intellectual disability.

Here, we present a case of a 10-year-old boy who had Fahr disease revealed by recurring seizures.

Keywords: *Calcification; Fahr; Basal Ganglia*

Introduction

Fahr's disease, also referred to as Fahr's syndrome or idiopathic basal ganglia calcification, is a neurological disorder that is inherited or occurs spontaneously. It was initially identified and described by Karl Theodor Fahr, a German neurologist, in 1930 [1]. The condition is primarily characterized by the abnormal accumulation of calcium in various regions of the brain responsible for controlling movement, including the basal ganglia, thalamus, dentate nucleus, cerebral cortex, cerebellum, subcortical white matter, and hippocampus [2].

Case Report

We present a case of a 10-year-old child born to non-consanguineous parents, with no history of neonatal distress, infection, or toxic exposure, who presented with recurrent epileptic seizures. Physical examination revealed normal blood pressure and blood sugar levels, while the neurological examination showed no abnormalities and there was no personal or family history of seizures or epilepsy. A CT scan showed bilateral, symmetrical calcifications in the basal ganglia, thalami, dentate nucleus, centrum semiovale, and subcortical white matter (Figure 1). No ischemic or hemorrhagic lesions were detected. Blood tests did not indicate any abnormal metabolism, confirming the diagnosis of Fahr's disease. The patient was administered antiepileptic treatment, and no further seizures were reported during the follow-up period.

Discussion

Fahr's disease, also referred to as primary familial brain calcification, is a rare neurodegenerative disorder characterized by the accumulation of calcium deposits composed of calcium phosphate and calcium carbonate. These calcifications are typically found in various

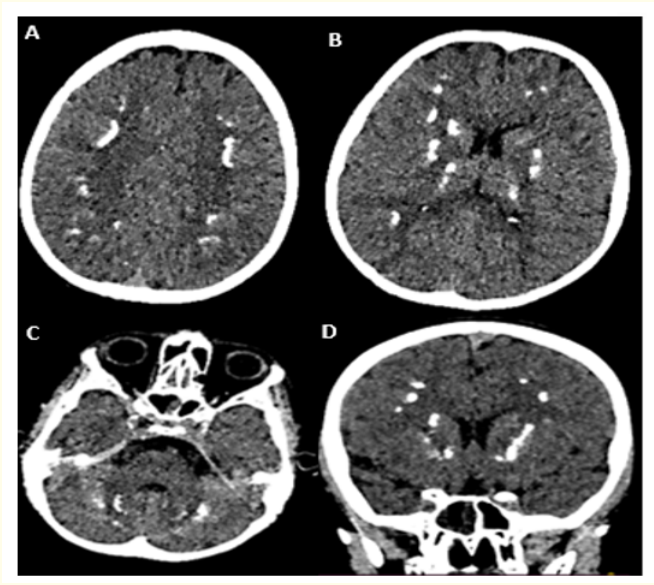

Figure 1: *Axial sections (A, B and C) and coronal reconstruction (D) of a brain CT scan showing bilateral and symmetrical calcifications of the basal ganglia, thalami, white matter and semi-oval center.*

regions of the brain, including the basal ganglia, thalami, dentate nucleus, hippocampus, cerebral cortex, and sub-cortical white matter [3]. Among these, the basal ganglia are the most commonly affected [4]. This condition is primarily associated with a genetic mutation located on chromosome locus 14q48 (IBCG1), often exhibiting dominant transmission [5]. However, sporadic cases of the disease have also been reported [5]. The condition was initially reported by Karl Theodor Fahr in 1930 [1].

Fahr's disease typically manifests in adult patients during their thirties or forties [5]. However, it is rare to observe this condition in children, and when it does occur, it is often associated with choreoathetoid movements [4]. According to the Fahr disease registry, approximately 67% of patients experience symptoms, with a higher incidence in males [5].

It presents with a diverse range of symptoms, including abnormal movements, parkinsonism, dementia, seizures, and cerebellar dysfunction. The diagnosis is typically made based on clinical evaluation, in conjunction with imaging exams that reveal calcification of the basal ganglia, along with hypocalcemia. Unfortunately, there is no known cure for Fahr's disease, and it follows a progressive and irreversible course. The prognosis is generally poor, as the central nervous system is affected, and the disease eventually leads to a fatal outcome [6].

Imaging plays a crucial role in the diagnosis of Fahr's disease. Non-contrast brain scans are particularly useful as they can reveal bilateral and symmetrical calcifications in key areas such as the subcortical white matter, basal ganglia, thalami, hippocampus, and dentate nucleus. Additionally, SWI MRI (susceptibility-weighted imaging) is known to be more sensitive in detecting early changes associated with Fahr's disease [7].

In the case of intracranial calcifications, it is important to consider and rule out other potential causes such as CMV infection, neurocysticercosis, toxoplasmosis, neurotuberculosis, HIV infection, astrocytoma, calcified infarct, hyperparathyroidism, vitamin D hypervita-

minosis, and tuberous sclerosis. However, in our patient's case, further investigations and medical history evaluation excluded infectious and metabolic diseases. Therefore, the diagnosis of Fahr's disease was confirmed.

Regrettably, there is currently no known cure for Fahr's disease [8]. Treatment mainly focuses on managing the symptoms and is primarily symptomatic in nature. It typically involves the prescription of medications such as antiepileptic drugs, antiparkinsonian drugs, and antipsychotic drugs, depending on the specific clinical manifestations of the disease.

Conclusion

Fahr's disease is a rare neurodegenerative disorder that particularly affects children. It is characterized by the presence of calcium deposits in the basal ganglia and white matter on brain scans. Diagnosis is based on clinical, imaging and biological findings.

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Conflict of Interest

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