Case Report

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Rare case in Somalia: Fahr's syndrome

Aisha M. Adam, MD[a,*], Omar A. Sheikh, MD[c], Miski A. Roble, MD[a], Mehmet Z. Yaşar, MD[d], Shukri S. Mohamed, MD[b], Mohamed M. Kassim, MD[a]

Introduction and importance: Fahr's syndrome is primarily familial, autosomal dominant, and genetically diverse. Basal ganglia calcification that is bilaterally symmetrical is a hallmark of this illness. Although the specific origins of this illness are unknown, it may be brought on by problems with calcium metabolism, infections, toxins, hereditary factors, hypoparathyroidism, and pseudohypoparathyroidism. The prevalence of this syndrome is less than 0.5%.

Case presentation: An 11-year-old female comes to the Emergency Department with her parents complaining of high-grade fever and convulsions for 1 week. Convulsion, which is a generalized tonic–clonic seizure, duration was ~5 min and associated with urinary incontinence and biting tongue. On examination, the patient was confused and irritable. Vital signs were normal; there is weakness in the right arm and right leg, associated with irregular movement. There was alternation in her level of consciousness, slurring of speech, and psychiatric symptoms. Another aspect of the neurological examination and systems was normal, and there was no meningeal irritation.

Clinical discussion: The pathogenesis of Fahr's syndrome is not completely known. The calcification is caused by flaws in the transport of radioactive particles and tissue damage caused by free radicals. Bilateral calcification found on a computed tomography (CT) scan of the brain, autosomal dominant inheritance, the absence of any infection, drugs, or toxins, the absence of mitochondrial dysfunction, and the presence of progressive neurological dysfunction is the clinical criteria for diagnosing Fahr's syndrome.

Conclusion: Basal ganglia calcification that is bilaterally symmetrical is a hallmark of Fahr's syndrome. CT scans are the gold standard for conclusively diagnosing Fahr's syndrome.

Keywords: autosomal dominant, basal ganglia calcification, Fahr's syndrome

Introduction

Karl Theodor Fahr, a scientist, initially identified Fahr's syndrome as an uncommon condition in 1930. Basal ganglia calcification that is bilaterally symmetrical is a hallmark of this illness[1]. Fahr's syndrome is primarily familial, autosomal dominant, and genetically diverse[2]. Although the specific origins of this illness are unknown, it may be brought on by problems with calcium metabolism, infections, toxins, hereditary factors, hypoparathyroidism, and pseudohypoparathyroidism. The prevalence of this syndrome is less than 0.5%[3]. Both neurological and behavioral signs are present clinically; vertigo, epilepsy, syncope, cerebellar ataxia, dementia, and movement disorders similar to Parkinson's disease are examples of neurological symptoms.

Unbalanced walking, easy tiredness, speech impairment, involuntary movements, swallowing difficulty, muscle cramps, dementia, personality changes, and neuropsychiatric disorders are some of the clinical signs and symptoms of Fahr's syndrome[4].

HIGHLIGHTS

- Fahr's syndrome, also known as idiopathic basal ganglia calcification, is a rare neurological condition that runs in families as an autosomal dominant feature.
- Fahr's syndrome is primarily familial, autosomal dominant, and genetically diverse. Although the specific origins of this illness are unknown, it may be brought on by problems with calcium metabolism, infections, toxins, hereditary factors, hypoparathyroidism, and pseudohypoparathyroidism.
- Fahr's syndrome, which gets worse over time, has no cure, and the calcification process cannot be stopped or reversed.

Case presentation

Patient background and clinical presentation: An 11-year-old female came to the emergency department and complained of high-grade fever and convulsion for 1 week. Convulsion, which is a generalized tonic–clonic seizure, duration was ~5 min and associated with urinary incontinence and biting tongue. On

[a]Department of Pediatrics, [b]Department of Pediatric Surgery, Mogadishu Somali Turkey Recep Tayyip Erdoğan Training and Research Hospital, [c]Department of Basic Medical Science, Faculty of Medicine, Somali National University, Mogadishu, Somalia and [d]Department of Pediatrics, Izmir Çiğli Training and Research Hospital, Çiğli/Izmir, Turkey

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*Corresponding author. Address: Mogadishu Somali Turkey Recep Tayyip Erdoğan Training and Research Hospital, Digfer Street, Hodan, Mogadishu, Somalia. Tel.: +252 615 651742. E-mail: cayuniadan@gmail.com (A.M. Adam).

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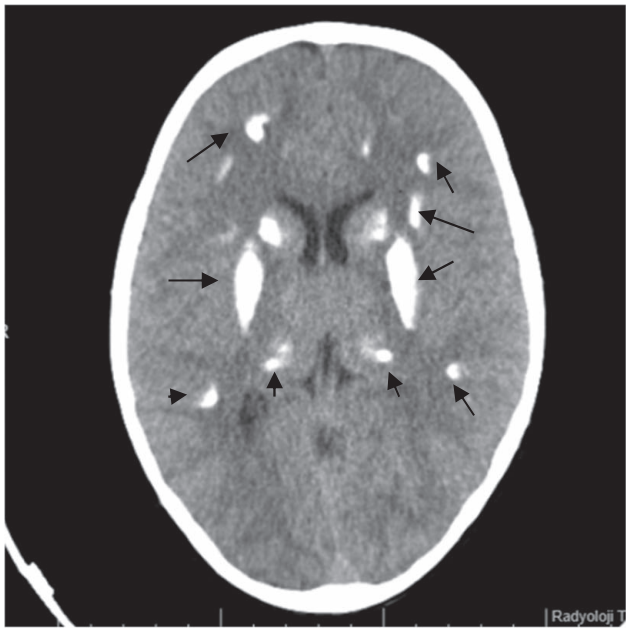

Figure 1. Bilateral and symmetric calcifications both involve subcortical frontoparietal white matter.

examination, the vital signs were normal, but the patient was confused and irritable. There is weakness in the right arm and right leg associated with irregular movement. There was an alternation of her level of consciousness and slurring of speech, but there was no meningeal irritation. Other systemic examinations were normal, and she has a past history of epilepsy for five years and no family history. *Laboratory finding:* WBC: $23 \times 1000/mm^3$, serum calcium: 5.2 mg/dl, magnesium: 1.7 mg/dl, parathyroid hormone (PTH): 2.3 mg/dl, phosphorus (P): 2.7 mg/dl, C-reactive protein (CRP) was 15 mg/l. *Brain computed tomography (CT) scan:* Revealed bilateral and symmetric calcifications of both subcortical frontoparietal white matter, basal ganglia, thalamus, and cerebellum (Fig. 1, black arrows). *MRI finding:* Hyperintense signal changes were observed on T1 and flair images at the level of the basal ganglia, at the level of the thalamus and in the cerebellum adjacent to the fourth ventricle. The work has been reported in line with the SCARE criteria[5].

Discussion

Fahr's syndrome's pathogenesis is not completely known. The beginning of calcification is caused by flaws in the transport of radioactive particles and tissue damage brought on by free radicals[4]. The same illness has been described using up to 35 different descriptive words[6]. The frequency of physiological intracranial calcification, with a 1.3–1.5% detection rate in studies using neuroimaging. Bilateral calcification found on a CT scan of the brain, autosomal dominant inheritance, the absence of any infection, drugs, or toxins, the absence of mitochondrial dysfunction, and the presence of progressive neurological dysfunction are the clinical criteria for diagnosing Fahr's syndrome[1]. Pathologies are divided into four categories: Adult-Onset Neurodegenerative Growth Anomalies, Infectious Disease, Genetic Syndromes, and Endocrinological Disorders[7].

Pathologies are categorized into four groups: Adult-Onset Infectious Illness, Genetic Syndromes, Endocrinological Diseases, and Neurodegenerative Growth Anomalies. There is frequent overlap between neurological symptoms, such as hypokinetic movement disorder associated with cognitive impairment and cerebellar signs[8].

Fahr's illness in contrast to atherosclerosis, the mineral deposits are found in the small capillaries and vessels of white matter[9]. Endothelial and stromal vascular cells, as well as the interstitium, are included in the calcification. However, the primary cause of the deposition of calcium and other minerals is local circulatory disturbances like regional schemia[10].

There is a strong genetic connection between IBGC (idiopathic basal ganglia calcification) and most cases of autosomal dominant inheritance in families. Although other genes have produced comparable manifestations, the most often linked genetic alterations are SLC20A2 on chromosome 8p11.2 and PDGFRB (platelet-derived growth factor receptor beta) on chromosome 5q32[11].

The most prevalent theory about the cause of basal ganglia calcification is that it results from endocrine issues, notably problems with parathyroid hormone balance. Increased calcinosis can result from hypoparathyroidism, and it is thought that calcium deposits first build up in the vessel wall before moving on to the neuron. This basal ganglia deposition results in decreased local blood flow, resulting in more neuron damage and hence higher levels of calcium deposition[7].

To identify the source of basal ganglia calcifications, further research must be done on the many intracranial calcification etiologies, such as vascular, metabolic, congenital, and viral. For instance, it is important to take into account the TORCH infections (toxoplasmosis, other, rubella, cytomegalovirus, and herpes simplex virus), as they might result in cerebral calcifications. Calcium homeostasis issues resulting from metabolic disturbances are the most frequent cause of basal ganglia calcification[12].

Fahr's syndrome, which gets worse over time, has no cure, and the calcification process cannot be stopped or reversed. Therapy is focused on reducing symptoms. Doctors concentrate on reducing its various mental and physical effects wherever possible. There have been reports of chelators that contain antioxidants and calcium antagonists, however these findings have not been verified. In the uncommon case when a patient exhibits seizure activity, antiepileptic, it is appropriate to combine drug use with drugs like levetiracetam. When treating a young female patient who is ready to have children, it is crucial to use various teratogenic drugs, such as carbamazepine and valproic acid[13].

Our patient has movement disorder, neuropsychiatric symptoms, and Fahr's disease presented with generalized tonic–clonic seizure.

Cranial CT scan showed bilateral and symmetric calcifications in both subcortical frontoparietal white matter, basal ganglia, thalamus, and cerebellum. The CT scan findings were interpreted in favor of Fahr's syndrome. A further blood chemistry metabolic panel test showed hypocalcemia and hypoparathyroidism.

Conclusion

Basal ganglia calcification that is bilaterally symmetrical is a hallmark of Fahr's syndrome. CT scans are the gold standard for conclusively diagnosing Fahr syndrome.

Ethical approval

Based on the regulations of the review board of the Mogadishu Somali Turkish Training and Research Hospital, institutional review board approval is not required for case reports.

Consent

Written informed consent was obtained from the patient's parent for publication and any accompanying images. A copy of the written consent is available for review by the Editor-in-Chief of this journal on request.

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We declare that we have no funding source.

Author contribution

A.M.A.: idea of the research, writing of the manuscript, final revision of the data, and intellectual content related to pediatrics; M.A.R.: idea of the research, writing of the manuscript, final revision of the data, and intellectual content related to pediatrics; M.Z.Y.: review of data and intellectual content related to pediatrics; S.S.M.: idea of the research, writing of the manuscript, and final revision of the data; M.M.K.: review of data and intellectual content related to pediatrics; O.A.S.: writing of the manuscript and review of data.

Conflicts of interest disclosure

No conflicts of interest in this work.

Research registration unique identifying number (UIN)

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Guarantor

As corresponding author, I confirm that the manuscript has been read and approved by all named authors.

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