Case Report

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Fahr's syndrome with hypoparathyroidism, thrombocytopenia, and seizure: a rare case report

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Introduction and Importance: Fahr's syndrome, also known as bilateral striatopallidodentate calcinosis is a rare neurological disorder that is characterized by abnormal deposition of calcium in the basal ganglia, cerebellar dentate nuclei, and cerebral cortical white matter. The authors report an extremely uncommon case of Fahr's syndrome with thrombocytopenia, hypoparathyroidism, and seizure.

Case Presentation: A 32-year-old male was brought with a repeated history of twitching of hands, tingling sensation, and uncontrolled seizure despite medications.

Clinical Discussion: Computed tomography findings showed bilateral basal ganglia, cerebellar dentate nuclei, and subcortical cerebral white matter calcifications. Laboratory studies revealed reduced levels of calcium, parathyroid hormone, and thrombocyte count. Based on these investigations Fahr's syndrome probably due to hypoparathyroidism with thrombocytopenia was diagnosed. The patient was initially treated with intravenous calcium gluconate and platelet transfusion followed by oral calcium supplementation.

Conclusion: Fahr's syndrome due to hypoparathyroidism should be suspected in any patient with neurological symptoms and hypocalcemia. Seizures in the patient of Fahr's syndrome with thrombocytopenia could be very detrimental due to the risk of intracranial hemorrhage. Hence, treatment should be started as early as possible.

Keywords: case report, Fahr's syndrome, thrombocytopenia, hypoparathyroidism, hypocalcemia

Introduction

Fahr's syndrome is a rare inherited neurodegenerative disorder that is characterized by calcium deposition in the basal ganglia, cerebellar nuclei, and cerebral white matter. It was first reported by German neurologist, Fahr in an 81-year-old male with symptoms of immobility, fever, and dementia with striatal calcification in the autopsy[1]. The exact etiology of calcium deposition in Fahr's syndrome is not well known. Disruption of the blood-brain barrier may be one of the causes of metastatic calcification or may be due to defective calcium metabolism in the brain[2].

The clinical presentation of Fahr's syndrome may vary depending upon the extent of calcification experiencing a wide range of motor and cognitive dysfunction, seizure, behavioral changes, and somatic changes like headache, vertigo, paresis, syncope, tremor, and ataxia[2].

HIGHLIGHTS

- Fahr's Syndrome is a neurodegenerative disorder associated with calcification of the basal ganglia and cerebellar nuclei.
- The diagnosis of Fahr's syndrome is based on the location of calcification and clinical features.
- Fahr's syndrome may be rarely associated with hypoparathyroidism and thrombocytopenia, which should be corrected early.

In our case, we report a case with hypoparathyroidism, thrombocytopenia, and seizure, which is a very unusual presentation of Fahr's syndrome.

Case report

A 32-year-old male presented to the medicine outpatient department for evaluation of repeated episodes of twitching of hands with a tingling sensation. He had multiple episodes of tonic-clonic type of seizure which was anteceded by visual aura. He was taking antiepileptic medication; however, no satisfactory control of seizure was noted. There was no history of memory loss, heat or cold intolerance, postictal confusion, or bowel or bladder incontinence. He denied any history of trauma or similar family history. He was having diffuse myalgia with a feverish sensation and a pulsatile headache in the occipital region. On physical examination, the respiratory rate was 18/min, blood pressure of 120/86 mmHg, pulse was 82/min, and the temperature of 100°F. He was lethargic but well oriented to time, place, and person. Deep tendon reflexes were normal. Nuchal rigidity was absent. Thyroid

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Table 1

Biochemical laboratory workup

Test	Result	Normal values	Test	Result	Normal values
Total leukocyte count	4800	4000–11 000/cumm	Urea(serum)	26.2	15–45 mg/dl
Neutrophil	68	40–70%	Creatinine(serum)	0.6	0.4–14 mg/dl
Lymphocyte	29	20–45%	Sodium (serum)	135	135–145 mmol/L
Hemoglobin	15 g/dl	13-18 g/dl	Potassium(serum)	5	3.5–5.5 mmol/L
MCV	89	80–100fl	Blood sugar(serum)	125	70–140 mg/dl
MCH	30	26–34 pg	Vitamin B12	222.4	200–1100 pg/ml
RBC count	4.96	4.5–5.5 million/cubic mm	Free thyroxine (T4)	0.7	0.5–1.4 ng/dl
Platelet count	0.8	$1.5–4.5 \times 10^5$/cubic mm	Free triiodothyronine (T3)	2.7	1.8–4.2 pg/ml
Serum calcium	5.5	8.5–11.5 mg/dl	Thyroid stimulating hormone (TSH)	3.1	0.35–5.1 mIU/ml
Intact parathyroid hormone (iPTH)	12	15.5–65.5	Urine pus cells	4–8/hpf	
Antinuclear antibody	0.5	0–40 AU/ml	Urine RBC	1–2/hpf	

Reduced level of thrombocyte, parathyroid hormone, and calcium is seen.

examination was normal. The mental status examination was normal. No significant systemic examination findings. Laboratory workup showed hypocalcemia, hypoparathyroidism, and thrombocytopenia. We observed that Vitamin B12, Vitamin D, and thyroid hormone levels were within normal limits (Table 1). Serological tests for dengue, scrub typhus, and typhoid were all negative for both IgG and IgM. Noncontrast computed tomography of the head showed symmetrical calcifications in the bilateral basal ganglia, dentate nuclei of the cerebellum, and subcortical white matter of the bilateral frontoparietal and left occipital lobe (Fig. 1). Urine screening for calcium and heavy metals was negative. Ultrasonography of the thyroid and parathyroid glands was normal. A chest radiograph was done, which revealed normal findings. Urine analysis showed few pus cells and red blood cells. Based on the above, Fahr's syndrome with hypoparathyroidism was diagnosed and calcium gluconate 10% by intravenous route was given for an acute episode followed by oral calcium gluconate 600 mg thrice a day. On follow up examination, level of calcium and platelet was improved with calcium(8 mg/dl) and platelet of (1.2×10^5/cubic mm) showing good adherence to medication. No obvious adverse event noted.

Discussion

Fahr's syndrome is a rare neurological disorder, predominately occurring in the third to fourth decade with calcification of the basal ganglia, cerebellar cortex, and cortical white matter, affecting normal body movement[3]. Fahr's disease refers to the idiopathic cause while Fahr's syndrome is secondary to the underlying cause. Fahr's syndrome shows dominant, recessive, and sporadic traits, although the dominant type is more common. It is associated with various endocrinopathies including hypothyroidism and hypoparathyroidism. Hypoparathyroidism with hypocalcemia alters bone homeostasis and hampers iron transport, generates free radicals, and leads to extraosseous calcium deposition[4]. Increased levels of copper, zinc, iron, and magnesium in the Cerebrospinal fluid of patients with Fahr's syndrome were noted. However, the calcium level in cerebrospinal fluid was found to be normal[5]. Disruption of the frontostriatal circuits at the basal ganglia level causing a reduction of glucose metabolism is another postulated etiopathology of Fahr's syndrome[6]. Single-photon emission computed tomography study of the brain showed markedly reduced perfusion in the cerebral cortices and

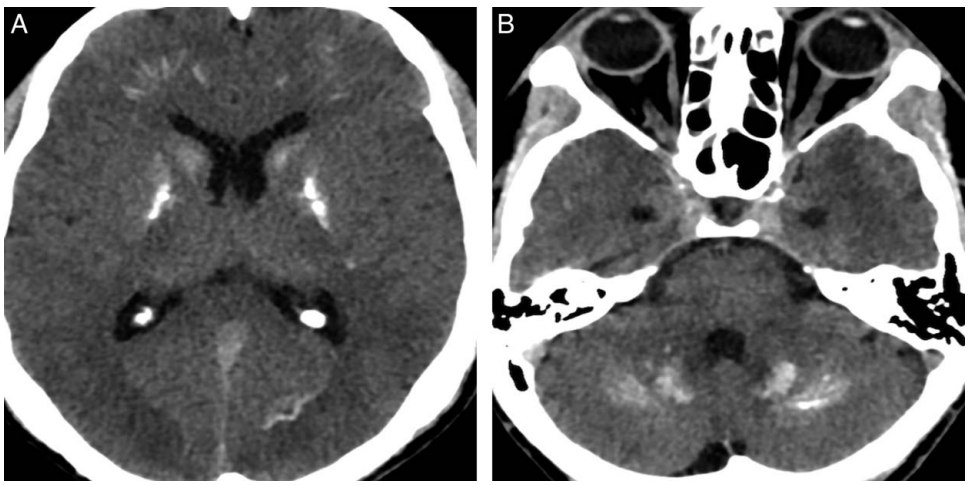

Figure 1. (A) Noncontrast axial computed tomography image at the basal ganglia level shows the calcification in the bilateral basal ganglia, the subcortical white matter of the bilateral frontoparietal, and the left occipital lobe. (B) Noncontrast axial computed tomography image at the level of cerebellum shows symmetric calcification of bilateral dentate nuclei of cerebellum.

basal ganglia[7]. Extraosseous calcifications in the brain and bone marrow may cause thrombocytopenia similar to our patients.

Clinically, Fahr's syndrome may be asymptomatic or present with tetany, convulsions, intellectual disabilities, choreoathetotic type body movements, and somatic symptoms like headache, vertigo, paresis, stroke, syncope, ataxia, and tremor[2]. Our patient had tetanic twitching, tingling, seizure, and a headache. Although mood disorder is the most common psychiatric symptom, other features like anxiety, personality disorder, hallucination, and dementia may also be present[8]. None of those psychiatric symptoms were present in our case.

The differential diagnosis of Fahr's syndrome is vague. The location of calcification and clinical features are of utmost importance in making the diagnosis. The most common differentials include Parkinson's disease, Huntington's disease, Progressive supranuclear palsy, and calcifying brain tumors like low-grade astrocytoma, oligodendroglioma, meningioma, and vascular malformations[9]. Biochemical evaluation of metabolic, inflammatory, and infectious disorders; heavy metal concentration, cerebrospinal fluid evaluation, hormonal assay including thyroid, parathyroid hormone, and calcitonin is needed to identify the possible etiology[2].

There is no established treatment modality to limit the progression of calcification in the basal ganglia. Treatment in Fahr's syndrome is directed towards the treatment of the underlying etiology and associated neuropsychiatric symptoms. Early identification and treatment of hypoparathyroidism can help to prevent calcification and neurophysiological disorder[10].

In our case, twitching and tingling sensations were reduced after starting calcium therapy. The level of platelet is rising. The patient is also treated with antibiotics for the urinary tract infection. Antiepileptic medication is continued and the patient's condition is well controlled.

This case report highlights an uncommon manifestation of Fahr's syndrome with thrombocytopenia and hypoparathyroidism, providing insights into the diagnostic workup and treatment strategies. The lack of long-term follow-up and single case study limits generalizability.

Conclusion

In a young patient with neurological symptoms and hypocalcemia, detailed endocrine evaluation and brain imaging should be performed to rule out possible etiology. Although very rare, thrombocytopenia in Fahr's syndrome should be identified early as there is a high chance of intracranial bleeding in a patient with recurrent seizure episodes. Appropriate seizure management and close observation are needed.

Ethical approval

This case report did not require review by the ethical committee.

Consent

Written informed consent was obtained from the patient for publication of this case report and the accompanying images. A copy of the written consent is available for review by the Editor-in-chief of this journal on request.

Author contribution

S.K.: conceptualization, as mentor and reviewer for this case report and for data interpretation; S.B.: contributed in performing literature review and editing; A.G.: contributed in writing the paper and reviewer for this case; P.G.: contributed in writing the paper. All authors have read and approved the manuscript.

Conflicts of interest disclosure

All the authors declare that they have no competing interest.

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