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Case Report

Fahr's syndrome associated with primary hypoparathyroidism: insights from a case report in Western Odisha

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ABSTRACT

Fahr's syndrome is characterized by having cortical, subcortical, deep grey nuclei calcification. It is an extremely rare clinic-radiological entity. Hypoparathyroidism may contribute to the pathogenesis, as a secondary cause. Calcium and phosphorus imbalance is main culprit for the disease process. Patients can present with varied neurologic and endocrinal disorder related to hypoparathyroidism. Diagnosis needs high index of suspicion or may be prompted from hematologic or endocrinal dysfunction. Treatment protocol based on managing underlying cause, as most of the causes for Fahr's syndrome are secondary in nature, as in our case. Majorities of case reports are of adult origin that caters to Fahr's disease which is quite different from Fahr's syndrome. Pediatric case reports data lagging in India, hence we tried to have a insight to the syndrome through a case of a young girl child, which may guide other pediatricians to have a knowledge about the syndrome with appropriate diagnosis and treatment options.

Keywords: Fahr's syndrome, Basal ganglia, Calcification, Calcium phosphorus metabolism, Neurologic disorder

INTRODUCTION

Fahr's disease is a primary hereditary neurodegenerative disorder, whereas Fahr's syndrome refers to secondary forms associated with various underlying metabolic or endocrine abnormalities.[1] Fahr's syndrome is an uncommon, progressive condition characterized by abnormal bilateral and symmetrical calcification of the deep grey matter of the brain, particularly involving the basal ganglia, cerebral cortex, and cerebellum.[2] The disorder was first described by the German neurologist Karl Theodor Fahr in 1930.[2,3]

Fahr's disease is most often inherited in an autosomal dominant manner, although sporadic cases have also been reported, and the causative gene has been mapped to chromosome 14q48.[2] Clinically, affected individuals may present with a wide range of neuropsychiatric manifestations, including cognitive impairment, behavioral changes, and mood disturbances.[4] Hypoparathyroidism is a rare endocrine disorder that leads to hypocalcemia, resulting from impaired calcium mobilization from bone and reduced renal calcium reabsorption, and the accompanying hyperphosphatemia causes precipitation of calcium-phosphate salts in soft tissues.[5]

Although various organs may be affected, intracranial calcification, particularly in the basal ganglia, is chiefly responsible for the neurological manifestations seen in Fahr's syndrome.[6] While Fahr's syndrome is rare, hypoparathyroidism remains one of its most frequent secondary associations.[7]

In our setup pediatric cases are exceedingly uncommon. Here, we report a pediatric case of Fahr's syndrome secondary to hypoparathyroidism due to dyshormonogenesis, presenting with recurrent seizures. This case underscores the importance of identifying metabolic and endocrine causes of intracranial calcification and highlights the rare but significant association between hypoparathyroidism and Fahr's syndrome.

CASE REPORT

A 12-year-old girl from Sonepur district, Odisha, who was fully immunized and neurodevelopmentally normal, born to non-consanguineous parents, presented to the pediatric emergency department with multiple episodes of generalized tonic-clonic seizures (GTCS). Each episode lasted approximately three to five minutes and was followed by a post-ictal phase with transient loss of consciousness, with full recovery between episodes. There was no history of fever, cough, cold, vomiting, headache, slurring of speech, blurring of vision, fall, trauma, animal bite, or recent surgery. The child had experienced her first seizure episode at seven years of age, also of GTCS type. She was treated at a local hospital with sodium valproate at a basic dose, without undergoing further evaluation or investigations. Due to poor compliance, she experienced multiple breakthrough seizures of similar nature over the following years. Her perinatal period was uneventful, and she had achieved age-appropriate developmental milestones across all domains, with a normal developmental quotient. Her mother had a history of three first-trimester spontaneous abortions, but there was no family history of seizures or neurological disorders. The family belonged to the lower-middle socioeconomic class according to the Modified Kuppuswamy scale. A three-generation pedigree chart (Figure 1) was constructed, which did not reveal any significant genetic association.

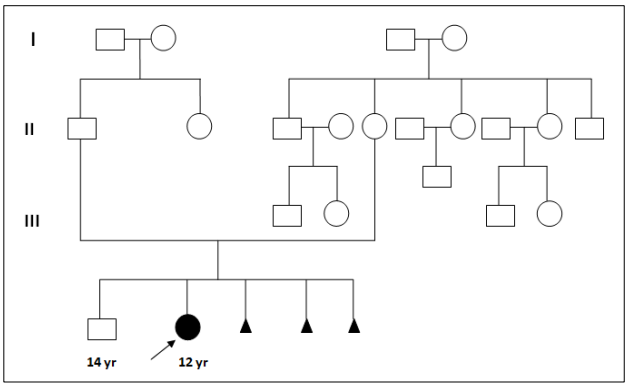

Figure 1: Three-generation family pedigree analysis of the child.

On admission to the emergency department, the patient's vital signs were within normal limits. The general physical examination was unremarkable, and anthropometric measurements were appropriate for age-weight: 31 kg (10th-25th centile) and height: 146 cm (25th-50th centile). A head-to-toe examination revealed no abnormalities. Higher mental functions were intact, with no evidence of focal neurological deficits, cranial nerve involvement, or speech abnormalities. There were no involuntary movements or gait disturbances. Motor system examination revealed bilateral extensor plantar responses (positive Babinski sign) and brisk deep tendon reflexes, with no clonus. Sensory and cerebellar system examinations were within normal limits, and there were no signs of meningeal irritation. The skull and spine appeared normal. Respiratory examination revealed normal breath sounds, and cardiovascular examination showed both heart sounds audible without any murmurs. Abdominal examination did not reveal hepatosplenomegaly or any other organomegaly. Routine hematological investigations demonstrated features of iron deficiency anemia. A neurological evaluation was undertaken. Electroencephalography (EEG) showed generalized epileptiform discharges, and non-contrast computed tomography (NCCT) of the brain revealed symmetrical bilateral basal ganglia and frontal cortical hyperdensities, consistent with intracranial calcifications (Figure 2).

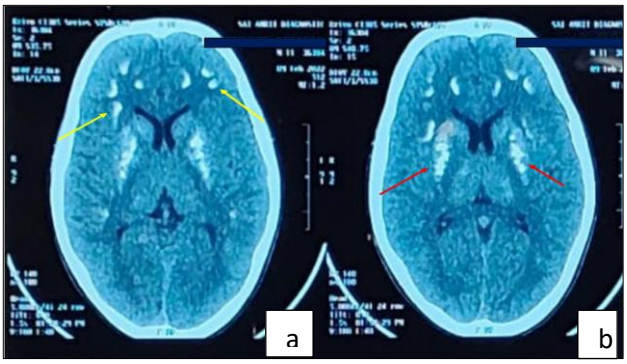

Figure 2 (a and b): NCCT brain showing hyperdense lesions in bilateral basal ganglia (red arrows) and frontal cortex (yellow arrows).

Further neurological evaluation was undertaken to assess for underlying causes, including metabolic and mitochondrial disorders. As magnetic resonance imaging (MRI) was readily available in our setup, the patient underwent detailed imaging studies. Susceptibility-weighted imaging (SWI) revealed patchy blooming foci involving the bilateral basal ganglia and frontal cortex, suggestive of calcifications (Figure 3). Subsequently, magnetic resonance angiography (MRA) of the cerebral arteries was performed, which revealed a normal study with preserved flow in all major vessels and their branches (Figure 4). These imaging findings, in conjunction with the patient's clinical and laboratory data, supported the presence of bilateral basal ganglia and cortical calcifications consistent with Fahr's syndrome.

Despite the mother's history of three first-trimester spontaneous abortions, the patient's siblings were neurodevelopmentally normal, and the patient herself had normal development. Moreover, the onset of symptoms was delayed, suggesting a secondary rather than primary cause. Given the presence of symmetrical intracranial calcifications in an older child, Fahr's syndrome was favoured over primary Fahr's disease. Further evaluation for secondary causes revealed normal renal function, liver function, thyroid profile, and ultrasonography of the neck and kidney-ureter-bladder (KUB). Autoimmune markers, including antinuclear antibody (ANA), were negative. Electrocardiogram (ECG) and arterial blood gas analysis

were within normal limits. Laboratory investigations revealed hypocalcemia (total serum calcium 5.9 mg/dl), hyperphosphatemia (serum phosphorus 12.1 mg/dl), normal magnesium (2.1 mg/dl), and insufficient vitamin D3 (15 ng/ml). The combination of low calcium and high phosphorus prompted evaluation of parathyroid function. While thyroid function was normal, parathormone (PTH) level was markedly low (<4 pg/ml). Based on these findings and the absence of secondary causes for hypoparathyroidism, a diagnosis of Fahr's syndrome secondary to primary hypoparathyroidism was made.

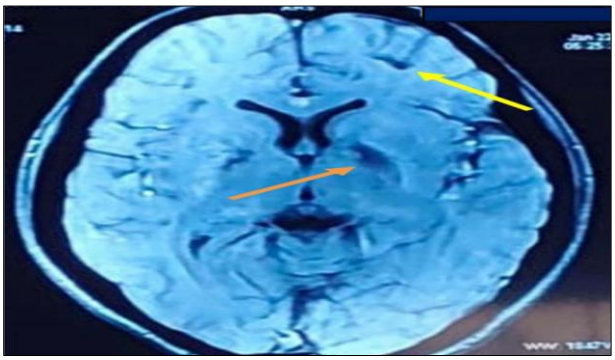

Figure 3: Susceptibility-weighted MRI of the brain showing patchy blooming foci in the bilateral basal ganglia (orange arrow) and frontal cortex (yellow arrow).

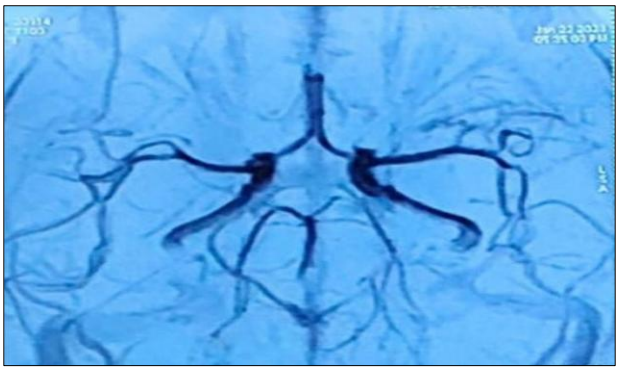

Figure 4: Magnetic resonance angiography (MRA) of the cerebral arteries showing normal blood flow in all major vessels and their branches.

During hospitalization, the patient continued on sodium valproate (60 mg/kg/day) and was started on levetiracetam (30 mg/kg/day) as an add-on therapy. She remained asymptomatic throughout her hospital stay, with no seizure activity documented. Supplemental oral calcium was administered at a dose of 1000 mg/day, and parathormone replacement therapy was initiated subcutaneously at 25 mcg/day. The patient and her parents were counseled regarding the condition, including proper storage and administration of the hormone. On subsequent follow-ups, the patient remained symptom-free, with good compliance to both antiepileptic and hormone therapy and no recurrence of seizures.

DISCUSSION

Fahr's syndrome is a clinico-radiological condition characterized by bilateral and symmetrical calcifications in the brain parenchyma and basal ganglia.[8] It is a rare entity, with an estimated prevalence of less than 1 in 1,000,000 children.[5,6] Fahr's disease tends to follow a progressive course, making it potentially lethal, but it has a variable prognosis and unpredictable outcome, which is not related to the position or extent of calcification.[9,10]

The high metabolic rate and rich cerebral circulation may explain why the basal ganglia are particularly prone to calcification. Calcium and phosphorus can also be easily exchanged from cerebrospinal fluid (CSF) due to the periventricular location of these structures.[11,12] Hypoparathyroidism can lead to hypocalcemia, which causes derangements in calcium and phosphorus metabolism and affects other micronutrients such as iron, and this imbalance increases oxidative stress through free radical injury, ultimately resulting in abnormal calcium deposition in tissues.[13] Although neck surgeries are the most common cause of hypoparathyroidism, our patient had no history of surgical intervention, and other causes of hypoparathyroidism were also investigated but found negative.[7]

Fahr's syndrome may be asymptomatic or may present with various neurological or psychiatric manifestations like twitching, seizures, poor scholastic performance, psychiatric manifestations, behavioral abnormalities, abnormal body movements (related to basal ganglia involvement), headaches, focal neurological deficits, stroke, syncope, gait abnormalities, and other movement disorders.[14] In our case, the patient presented solely with seizures, without any other neurological or psychiatric symptoms. Classic features of hypocalcemia, such as carpopedal spasms, Trousseau's sign, and Chvostek's sign, were also absent.

Imaging remains the cornerstone for diagnosis, out of which non-contrast computed tomography (NCCT) is more reliable than magnetic resonance imaging (MRI) for detecting calcifications.[15] In our patient, NCCT demonstrated bilateral symmetrical calcium deposition in the basal ganglia and frontal lobes, which was diagnostic of Fahr's syndrome. The differential diagnoses for Fahr's syndrome are extensive and include autoimmune disorders such as systemic lupus erythematosus (SLE), metabolic disorders like mitochondrial disease, intrauterine infections, intracranial hemorrhage, subarachnoid hemorrhage, human immunodeficiency virus infection (HIV), neurocysticercosis, and arteriovenous malformations.[16]

There is no specific treatment available for Fahr's syndrome, so the goal of management is to treat symptoms and address underlying or secondary causes.[6] In our case, the patient was managed with anti-seizure medication, calcium supplementation, and hormone replacement

therapy for hypoparathyroidism, which resulted in a good quality of life and symptom-free intervals.

A review of the literature revealed that most reported cases involve adults, with only a few describing pediatric associations. We report this case in an adolescent girl, highlighting an uncommon presentation of Fahr's syndrome associated with primary hypoparathyroidism. This case underscores the importance of considering this rare entity in the differential diagnosis of seizures in children and adolescents. It may also serve as a foundation for further diagnostic and research strategies. However, this report has certain limitations. Being a single case, the findings cannot be generalized to all patients with Fahr's syndrome. Exome sequencing could not be performed due to limitations in availability and affordability. Additionally, long-term follow-up of the patient was not possible, which further restricts the scope of the study.

CONCLUSION

Fahr's syndrome is an extremely rare clinical entity as far as the pediatric age group is concerned. Moreover, primary hypothyroidism as a surrogate cause for the syndrome is unique, making the clinician more troubled and enthusiastic at the same time for the evaluation of such patients. The combination of both conditions may present with chronic hypocalcemia, seizure episodes, neuropsychiatric manifestations, movement disorders, etc. Like our case, it was very difficult to suspect the syndrome in the background of endocrinal dysfunction, as the presentation was only seizure episodes, and this was unevaluated for a long duration. In this type of presentation, brain imaging is of utmost importance.

Calcium and phosphorus metabolism markers are also handy in finding the cause of the syndrome, if you are lucky enough to get a cerebral calcification in brain imaging. Because directed therapy is helpful but still challenging. Despite proper treatment, some may land in serious residual neurological issues and speech and language dysfunction due to the involvement of crucial brain parts. However, due to its non-progressive nature, early diagnosis and treatment has better results than Fahr's disease, which is a progressive one.

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REFERENCES

1. Supit VD, Kurniawan D, Fatimah E. Fahr's syndrome and neurological manifestations in hypoparathyroidism patients. Radiol Case Rep. 2024;19:1248-53.
2. Kundu GK, Bhadra S, Ahmed S: A child with Fahr's disease. BAOJ Pediat. 2017;3:036.
3. Saleem S, Aslam HM, Anwar M, Anwar S, Saleem M, Saleem A, et al. Fahr's syndrome: literature review of current evidence. Orphanet J Rare Dis. 2013;8:156.
4. König P: Psychopathological alterations in cases of symmetrical basal ganglia sclerosis. Biol Psychiatry. 1989;25(4):459-68.
5. Berrabeh S, Messaoudi N, Elmehraoui O, Assarrar I, Karabila I, Jamal A, et al. Hypoparathyroidism and Fahr's syndrome: a case series. Cureus. 2023;15:e40502.
6. Arruda AC, Guerra AC, Pessoa CH, Marquezine GF, Delfino VD. Hypoparathyroidism and Fahr's syndrome: case series. J Bras Nefrol. 2022;44:592-6.
7. Zhou YY, Yang Y, Qiu HM. Hypoparathyroidism with Fahr's syndrome: a case report and review of the literature. World J Clin Cases. 2019;7:3662-70.
8. Manyam BV, Walters AS, Keller IA, Ghobrial M. Parkinsonism associated with autosomal dominant bilateral striopallidodentate calcinosis. Parkinsonism Relat Disord. 2001;7:289-95.
9. Nishiyama K, Honda E, Mizuno T. A case of idiopathic, symmetrical non-arterioscelerotic, intracerebral calcification (Fahr's disease) associated with M protenemia, followed by multiple myeloma. Rinsho Shinkeigaku. 1991;31:781-4.
10. Rastogi R, Singh AK, Rastogi UC, Mohan C, Rastogi V. Fahr's syndrome: a rare clinico-radiologic entity. Med J Armed Forces India. 2011;67:159-61.
11. Goswami R, Sharma R, Sreenivas V, Gupta N, Ganapathy A, Das S. Prevalence and progression of basal ganglia calcification and its pathogenic mechanism in patients with idiopathic hypoparathyroidism. Clin Endocrinol. 2012;77:200-6.
12. Zavatta G, Clarke BL. Basal ganglia calcification in hypoparathyroidism and pseudohypoparathyroidism: local and systemic metabolic mechanisms. J Endocrinol Invest. 2021;44(2):245-53.
13. Cummings JL, Gosenfeld LF, Houlihan JP, McCaffrey T. Neuropsychiatric disturbances associated with idiopathic basal ganglia calcification. Biol Psychiatry. 1983;18:591-601.
14. Mufaddel AA, Al-Hassani GA. Familial idiopathic basal ganglia calcification (Fahr's disease). Neurosciences (Riyadh). 2014;19:171-7.
15. Manyam BV, Bhatt MH, Moore WD, Devleschoward AB, Anderson DR, Calne DB. Bilateral striopallidodentate calcinosis: cerebrospinal fluid, imaging and electrophysiological studies. Ann Neurol. 1992;31:379-84.
16. Saade C, Najem E, Asmar K, Salman R, Achkar BE, Naffaa L. Intracranial calcifications on CT: an updated review. J Radiol Case Rep. 2019;13:1-18.

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