ISSN Print: 2664-7591
ISSN Online: 2664-7605
Impact Factor: RJIF 5.2
IJAN 2023; 5(2): 117-119
www.pharmaceuticaljournal.in
Received: 05-09-2023
Accepted: 09-10-2023

Reshma P Varghese
Doctor, Department, of Pharmacy, Annai JKK Sampoorani Ammal College of Pharmacy, Komarapalayam, Tamil Nadu, India

Dr. Jincy Jesus
Department of Clinical Pharmacology, MGM Muthoot Hospital, Kozhenchery, Kerala, India

Dr. Alsa Mariyam Thomas
Department of Clinical Pharmacology, MGM Muthoot Hospital, Kozhenchery, Kerala, India

Dr. Jophine John
Department of Clinical Pharmacology, MGM Muthoot Hospital, Kozhenchery, Kerala, India

Corresponding Author:
Reshma P Varghese
Doctor, Department, of Pharmacy, Annai JKK Sampoorani Ammal College of Pharmacy, Komarapalayam, Tamil Nadu, India

Case report on Fahr's disease

Reshma P Varghese, Dr. Jincy Jesus, Dr. Alsa Mariyam Thomas and Dr. Jophine John

DOI: https://doi.org/10.33545/26647591.2023.v5.i2b.70

Abstract
Background: Fahr's disease is a rare neurological disorder characterized by abnormal deposition of calcium in brain. Etiology of Fahr's disease does not identify but most common of which are endocrine disorders, mitochondrial myopathy, dermatological disorders and infectious diseases. Management strategies and treatment of Fahr's disease mainly focus on symptomatic relief.
Case Presentation: A 49 year old female patient was admitted with complaints of vertiginous sensation followed by fall and altered level of consciousness. She had a medical history of thyroid carcinoma followed by s/p of 26 years back thyroidectomy. The laboratory investigations revealed elevated phosphorus and decreased calcium and Parathyroid hormone level. The CT scan revealed symmetrical hyperdense areas of calcification –that suggestive of Fahr's disease.
Conclusion: Fahr's disease is an uncommon neurodegenerative disorder characterized by idiopathic bilateral deposits of calcium in the striopallidodentate area, basal ganglia, thalamus, cerebral cortex, cerebellum and hippocampus.

Keywords: Fahr's disease, thyroidectomy, hypoparathyroidism, hyperphosphatemia

Introduction
Fahr's disease (FD) is a rare genetically dominant neurodegenerative disorder characterized by idiopathic bilateral deposits of calcium in the striopallidodentate area, basal ganglia, thalamus, cerebral cortex, cerebellum and hippocampus. Fahr's disease is a rare condition with a prevalence of 1/1,000,000, and the total number of cases reported so far is less than 200. Clinical manifestations are mainly depending upon the area of the brain affected. Fahr's disease has various clinical presentations such as - motor symptoms including tremors, ataxia, rigidity, and aphasia to psychiatric manifestations such as hallucinations, delusions, and cognitive impairment deterioration of motor function, dementia, seizures, headache, dysarthria, spasticty, eye-impairments and athetosis. The most common metabolic cause of Fahr's disease is hypoparathyroidism. Treatment is mainly symptomatic. Therapy of Fahr's disease is usually symptomatic and adjusted based on the etiology. The prognosis is varying, unpredictable, and depends on the extension of calcification. The use of carbamazepine, benzodiazepines and barbiturates in patients with Fahr's disease can lead to increased gait dysfunction.
Hypoparathyroidism (HPT) is an endocrine disease, which can be idiopathic or secondary. Idiopathic form is characterized by deficient secretion of parathyroid hormone (PTH) without a definitive cause. Parathyroid hormone controls the levels of calcium and phosphorus.

Case Report
A 49 year old female patient was admitted with complaints of vertiginous sensation followed by fall and altered level of consciousness (LOC) lasting around 15-20 minutes. Her LOC was considered probably due to acute symptomatic seizures secondary to hypocalcaemia. She had a medical history of thyroid carcinoma followed by s/p of 26 years back thyroidectomy. She also had a past medication history of Tab.Thyronorm100 mcg daily before food. She also had hypoparathyroidism associated hyperphosphatemia, i.e., hypoparathyroidism, acromegaly, and thyrotoxicosis which enhances the renal phosphate reabsorption resulting in hyperphosphatemia.

Computed tomography (CT) of brain shows symmetrical hyperdense areas of calcification are seen and evolving a bilateral centrum semiovale, corona radiata, gangliocapsular regions, fronto-parietal lobes and bilateral cerebellar hemispheres (dentate nuclei) –that suggestive of Fahr's disease. Rest of the brain parenchyma, basal ganglia, thalami and posterior fossa structures are normal. There is no evidence of intracranial bleed /infract / SOL (space-occupying lesions of the brain), no intra axial collection and no midline shift is seen, the cortical sulci, basal cisterns and ventricular system are normal, skull bones appear normal, no evidence of any fracture noted and incidentally noted a bilateral ethmoid sinusitis.

On physical examination, patient was conscious, oriented and mild gait ataxia (+). On vital sign examination, pulse rate was 66 beats per min, BP was 110/70 mmHg, respiratory rate was 21 beats per min, temperature was 98ºF and SPO2 was 98%. An improper calcium and phosphorus metabolism likely contributed to the severe cerebral calcification. Consequently, linked laboratory investigations were conducted and found elevated phosphorus level: 5 mg/dl (Normal value: 2.5-4.5mg/dl), decreased calcium level: 4.3 mg/dl (Normal value:8.4-10.4mg/dl), elevated urea level: 39mg/dl (Normal value:15-36mg/dl), low Parathyroid hormone (PTH) level: 0.5pg/ml (Normal level:15-68.3 pg/ml), decreased T3(Total T3) level: 0.51ng/dl (Normal value:0.58-1.59 ng/dl), elevated TSH (Third generation) level: 15.1µlU/ml (Normal value:0.34-4.94µlU/ml), decreased PCV: 35% (Normal value:36-41%), decreased Haemoglobin level:11.8gm% (Normal value:12-15gm%), elevated Neutrophils level: 78% (Normal value: 40-70%), elevated ESR level: 52mm/hr (Normal value: 5-20 mm/hr).

Table 1: Shows the Constituents, Detected vale and Normal value

Constituents	Detected vale	Normal value
Phosphorus	5 mg/dl ↑	2.5-4.5mg/dl
Calcium	4.3 mg/dl ↓	8.4-10.4mg/dl
Urea	39mg/dl ↑	15-36mg/dl
Parathyroid hormone	0.5pg/ml↓	15-68.3 pg/ml
T3(Total T3)	0.51ng/dl ↓	0.58-1.59 ng/dl
TSH(Third generation)	15.1µlU/ml ↑	0.34-4.94µlU/ml
PCV	35% ↓	36-41%
Haemoglobin	11.8gm% ↓	12-15gm%
ESR	52mm/hr ↑	5-20 mm/hr
Neutrophils	78% ↑	40-70%

The presence of secondary hypoparathyroidism with hypocalcemia as the etiology of fahr's disease was positively identified. The patient underwent therapy using Inj. Calcium gluconate 10 ml thrice daily, Tab. Shelcal C T (Calcium 500 mg + Vitamin D3 0.25mcg) thrice daily, Tab. Thyronorm100 mcg daily before food, Tab. Encorate Chrono (Sodium valproate + Valproic acid) 200 mg twice in a day, Tab. Stugeron Plus (Cinnarizine 20 mg + Dimenhydrine 40 mg), Cap. Pantop D once in a day and Syp. Arachitol Nano (Vitamin D3) 60000 units daily once weekly. Her condition was improved symptomatically and she was discharged in a stable state.

Discussion

Fahr's disease is a neurodegenerative condition characterized by neuropsychiatric and neurological manifestations. It is rare and occurs in people of 40-60 years of age. Even within the same family, various patients may experience varied clinical manifestations, ranging from asymptomatic patients to more severe courses. There are several medical conditions associated with Fahr's disease. The most common one is endocrine disorder, which is hypoparathyroidism. The gold standard method for an accurate diagnosis is a brain Computed Tomography (CT) scan, which makes it simple to distinguish calcified areas from spontaneous hyperdense lesions. Other laboratory examinations may be performed as well, such as serum calcium, phosphate, magnesium, ALP, Calcitonin, and PTH. These examinations are usually directed to rule out the underlying etiology of this syndrome. The calcification may become aggravated as the patients grow older and thus the symptoms may also become worse. Primary Fahr's disease does not have a specific treatment. Correction of metabolic abnormalities, such as low calcium or phosphorus levels, can, however, produce some improvement, as in our case, and can halt progression. According to the injured brain area, other symptoms are typically treated symptomatically. Treatment approaches concentrate on symptom management and avoiding consequences. The following are potential therapies:

- Anticonvulsant medications to control seizures
- Antidepressants or other medications to treat psychiatric symptoms such as anxiety, depression, or obsessive-compulsive disorder (OCD)
- Medication to treat bladder-control problems
- Continual observation of symptoms related to mental and neurological health

Hypoparathyroidism (HPT) is a clinical syndrome caused by insufficient secretion and or influence of Parathyroid hormone (PTH). Hypocalcemia, hyperphosphatemia, enhanced neuromuscular excitability, and heterotopic calcification of soft tissues are its clinical features. A wide range of clinical symptoms are possible when HPT is present. Numerous factors can lead to hypoparathyroidism. The majority (About 75%) of hypoparathyroidism cases are caused by neck surgery. Early HPT treatment can stop the development of calcifications and neurophysiological issues. Calcium and vitamin D supplements are used as part of the conventional HPT treatment.

Conclusion

Fahr's disease is an uncommon neurodegenerative disorder characterized by idiopathic bilateral deposits of calcium in the striopallidodentate area, basal ganglia, thalamus, cerebral cortex, cerebellum and hippocampus. Patients generally present with neuropsychiatric and neurological manifestations. Fahr's disease should be suspected if the patient has hypoparathyroidism, worsening neurological symptoms, and symmetrical and abnormal basal ganglia calcifications on imaging. Computed tomography has been shown to be the most accurate diagnostic tool. The calcifications of the basal ganglia, despite being a suggestive finding of an evolved disease, may be a finding of enormous value since they can immediately guide the diagnosis and allow rapid treatment of hypocalcemia and hypoparathyroidism.

References

1. Chen JQ. Fahr's syndrome, intracranial calcification and secondary hypoparathyroidism. QJM [Internet].

[Cited 2023 Jun 7]. 2022;115(7):481-2. Available from: https://academic.oup.com/qjmed/article/115/7/481/6550063

2. Soares FB, Amorim FF, Santana AR, de Moura EB, Margalho SB, Amorim APP, *et al*. Fahr's Syndrome due to Hypoparathyroidism Following Thyroidectomy. J Med Cases [Internet]. [Cited 2023 Jun 7]. 2013;4(6):380-4. Available from: https://www.journalmc.org/index.php/JMC/article/view/1357/687
3. Zhou YY, Yang Y, Qiu HM. Hypoparathyroidism with Fahr's syndrome: A case report and review of the literature. World J Clin Cases [Internet]. [Cited 2023 Jun 7]. 2019;7(21):3662-70. Available from: http://dx.doi.org/10.12998/wjcc.v7.i21.3662
4. Rashid Khan Z, Waheed W, Mabood J, Ali A, Burki G. A unique presentation of Fahr's syndrome secondary to hypoparathyroidism. Cureus [Internet]. [Cited 2023 Jun 7]. 2021;13(6):e16063. Available from: http://dx.doi.org/10.7759/cureus.16063
5. Dos Santos VM, Da Mata AMDF, Ribeiro KRA, Calvo ICDS. Fahr's syndrome and secondary hypoparathyroidism. Rom J Intern Med [Internet]. 2016;54(1):63-5. Available from: http://dx.doi.org/10.1515/rjim-2016-0007
6. Manohari R, Ramkumar S. Fahr's Disease - A rare cause for seizures - a case report. University Journal of Medicine and Medical Specialities [Internet]. [Cited 2023 Jun 7]; c2015, 1(1). Available from: http://ejournal-tnmgrmu.ac.in/index.php/medicine/article/view/11
7. Shah SY, Hadi FA, Idrees M, Levin-Carrion Y, Pande H, Oorloff MD, *et al*. Fahr's syndrome secondary to primary hypoparathyroidism presenting with seizures and the role of steroid therapy. Clin Med Insights Case Rep [Internet]. 2023;16:117954762311781. Available from: http://dx.doi.org/10.1177/11795476231178119
8. Sinha R, Sodhi K, John BM, Singh D. Fahr's Disease: A Case Report. J Nepal Paediatr Soc [Internet]. [Cited 2023 Jun 7]. 1970;30(1):44-5. Available from: https://www.nepjol.info/index.php/JNPS/article/view/2459
9. Mendes T, Fontes J, Sousa B, Silva J. Síndrome de Fahr - A propósito de un caso. Galicia Clin [Internet]. 2022;83(3):44. Available from: https://galiciaclinica.info/PDF/66/2592.pdf
10. Rashid Khan Z, Waheed W, Mabood J, Ali A, Burki G. A unique presentation of Fahr's syndrome secondary to hypoparathyroidism. Cureus [Internet]. [Cited 2023 Jun 7]. 2021;13(6):e16063. Available from: https://pubmed.ncbi.nlm.nih.gov/34345548/