

Fahr's Syndrome with Hypoparathyroidism Presenting with Acute Ischemic Stroke

Krishnendu Choudhury*, Biswajit Paul, Koushik Ray, and Shibendu Ghosh

Department of Medicine and Neurology, Jagannath Gupta Institute of Medical Science, Budge Budge, Kolkata

Abstract

Fahr's syndrome comprises of calcification in basal ganglia, thalamus and cerebellar dentate nucleus with or without neurodegenerative or psychiatric manifestations. It may be idiopathic or secondary to hypoparathyroidism. It can be genetically transmitted to families in an autosomal dominant manner, or can be sporadic. Common neurological manifestations of Fahr's syndrome are movement disorders including Parkinsonism, chorea, dystonia or spasticity. Dystonia and paratonia also manifest as abnormalities of muscle tone, but arise more due to the network dysfunction between the basal ganglia and the Cerebello-thalamo-cortical connections. Ischemic cerebrovascular disease may be associated, and incidence of ischemic or hemorrhagic stroke in Fahr's syndrome has been rarely reported.

We report here an elderly woman who had presented with acute ischemic stroke and found to have Fahr's syndrome in brain imaging. She was also detected to have primary hypoparathyroidism with decreased serum vitamin D3 and parathormone deficiency.

Keywords: Fahr's syndrome • Hypoparathyroidism • Acute Ischemic Stroke

Introduction

Idiopathic basal ganglia calcification, also known as Fahr's disease, is a rare, genetically transmitted autosomal dominant, neurological disorder characterized by abnormal deposits of calcium in areas of the brain that control movement. It can also occur independently as dentato-thalamo-lenticular calcification as a sporadic disorder in elderly people. Manifestations such as movement disorders like chorea, dystonia or spasticity, paratonic rigidity are present in minority of patients and most of the patients are asymptomatic. Some of them are reported to have associated hypoparathyroidism with decreased vitamin D and parathormone level in serum.

CT scan and MRI brain can reveal calcifications primarily in the basal ganglia, lenticular nucleus and dentate nuclei of cerebellum and in other areas such as the thalamus and cerebral cortex.

There is no reliable correlation between age, extent of calcium deposits in the brain, and development of neurological features.

Dystonia and paratonia also manifest as abnormalities of muscle tone, but arise more due to the network dysfunction between the basal ganglia and the thalamo-cerebello-cortical connections.

Ischemic cerebrovascular disease may be associated, and incidence of ischemic or hemorrhagic stroke in Fahr's syndrome has been rarely reported. We report here an elderly woman who had presented with acute ischemic stroke and found to have Fahr's syndrome in brain imaging. She was also detected to have primary hypoparathyroidism with decreased serum vitamin D₃ and parathormone deficiency.

Case Report

A 65 year old female admitted with sudden onset slurring of speech and

weakness of left side of face and left half of her body. There was no fever, diarrhea, convulsion or loss of consciousness. She did not have any history of chronic hypertension, heart disease, kidney disease or diabetes mellitus. There was no history of cognitive impairment or abnormal movement in her or any family members. There was no history of neck radiation or any thyroid disease or operation of neck or thyroid gland.

On examination she was conscious, orientated, her pulse, temperature and respiration were within normal range. Blood pressure was 170/100 mm Hg. Her mental status did not show any significant impairment; Speech was dysarthric with normal comprehension and speech output. Cranial

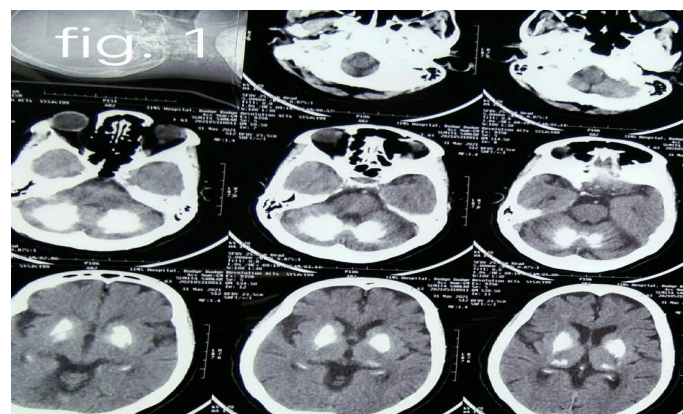


Figure 1. Calcification in the putamen, globus pallidus, thalamus.

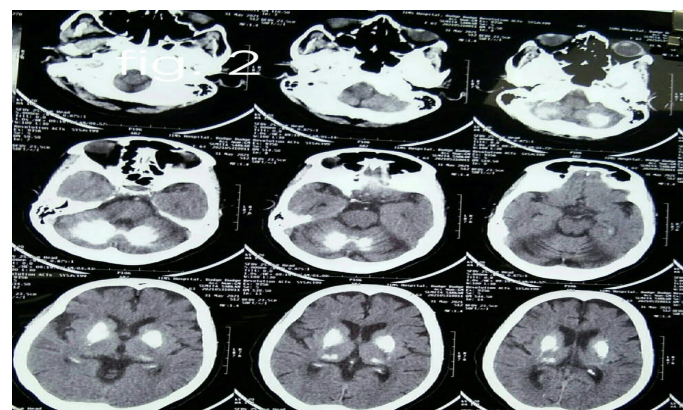


Figure 2. Dentate nucleus of cerebellum bilaterally.

***Address for Correspondence:** Choudhury K, Department of Medicine and Neurology, Assistant professor and Consultant Neurologist, Jagannath Gupta Institute of Medical Science, Budge Budge, Kolkata, E-mail: kushal.ch2009@gmail.com

Copyright: © 2021 Choudhury K, et al. This is an open-access article distributed under the terms of the Creative Commons Attribution License, which permits unrestricted use, distribution, and reproduction in any medium, provided the original author and source are credited.

Received: 13 August, 2021; **Accepted:** 27 August, 2021; **Published:** 04 September, 2021

nerves examination showed visual acuity 6/18 with immature cataract in both eyes and left facial palsy (lower face) upper motor neuron type; Motor system examination revealed decreased power (grade 1/5) in left upper and lower limbs with hypertonia, hyperreflexia and Babinski sign. Her right upper and lower limbs power was slightly reduced with paratonic rigidity in right upper limb. There was sensory extinction in her left side. Cerebellar signs were absent on right side. There was no carotid bruit. Investigations: Routine blood examinations CBC ESR CRP were normal. CBG and RBS were normal, PPBS and FBS, HbA1C were normal. CT scan did not show any hypodensity. There was calcification in the putamen, globus pallidus, thalamus and dentate nucleus of cerebellum bilaterally (Figures 1 and 2). MRI brain revealed diffusion restricted lesion (Figure 3) in right parietal, frontal and external capsule regions (Middle Cerebral Artery territory infarcts) with corresponding ADC dark mapping (Figure 4). There was Gradient Echo blooming in bilateral thalamus, lenticular nucleus and dentate nucleus of cerebellum indicating calcification (Figures 5 and 6).

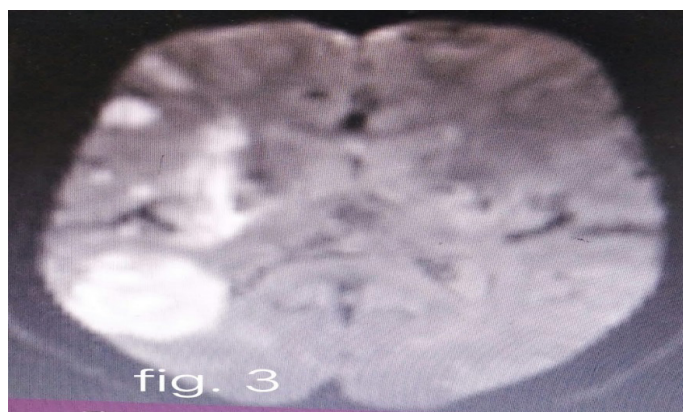


Figure 3. MRI brain revealed diffusion restricted lesion.



Figure 4. Frontal and external capsule regions (Middle Cerebral Artery territory infarcts) with corresponding ADC dark mapping.

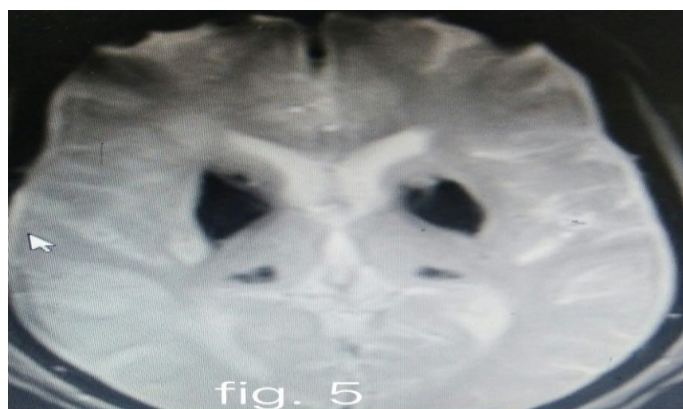


Figure 5. There was Gradient Echo blooming in bilateral thalamus, lenticular nucleus.

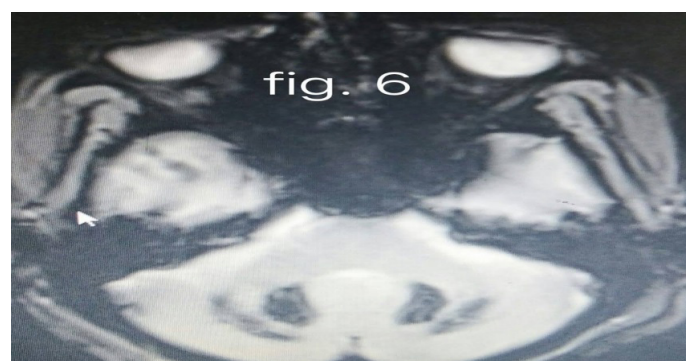


Figure 6. Dentate nucleus of cerebellum indicating calcification.

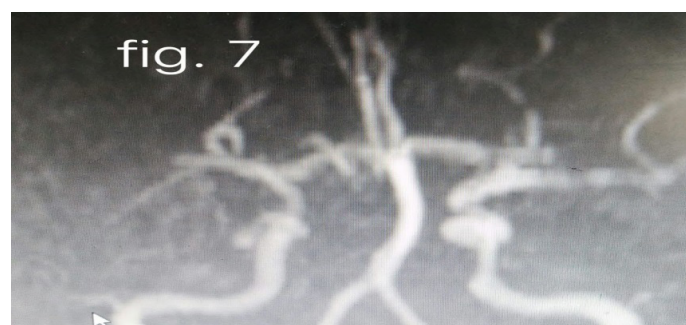


Figure 7. MR angio showed occlusion in distal right MCA with decreased flow along M2, M3 branches.



Figure 8. MR angio showed occlusion in distal right MCA with decreased flow along M2, M3 branches.

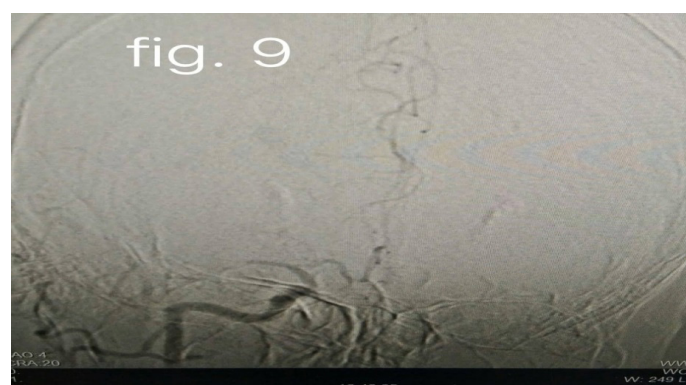


Figure 9. DSA showed distal MCA occlusion with collaterals from ACA-PCA.

MR angio showed occlusion in distal right MCA with decreased flow along M2, M3 branches (Figures 7 and 8). DSA showed distal MCA occlusion with collaterals from ACA-PCA, and calcified plaque in right ICA origin with 50% luminal narrowing (Figures 9 and 10). Her serum calcium level was low-normal (8.6 mg/dl) with normal phosphate (4.2 mg/dl) and marginally low ionised calcium (1.1 mol/l). Alkaline phosphatase was normal. Her plasma iPTH (5.98 pg/ml) and vitamin D (6.09 ng/ml) levels were very low. Thyroid function tests were within normal range. ECG, echocardiograms were normal.

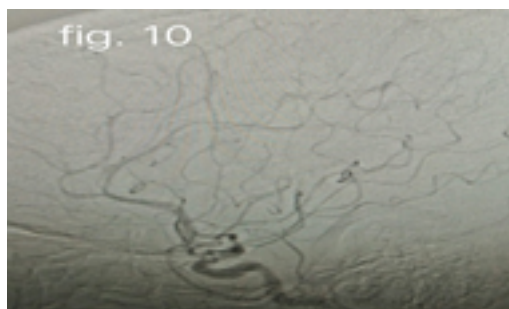


Figure 10. Calcified plaque in right ICA origin with 50% luminal narrowing.

Final diagnosis, Dentato-thalamo-lenticular calcification (Fahr's syndrome) with ischemic cerebral infarct in right MCA territory with newly detected hypertension and primary hypoparathyroidism.

Discussion

This elderly woman had a recent acute ischemic stroke with left hemiparesis and without any identifiable history of neurological illness. She also had hypertension and hyperlipidemia which were not detected or treated earlier. No other risk factors of stroke were present. CT scan of brain showed hyperdense calcifications in putamen, globus pallidus, thalamus and cerebellar dentate nucleus and white matter consistent with Fahr's syndrome. MRI brain revealed acute ischemic infarct in right parietal, external capsular, and frontal lobe regions consistent with right MCA occlusion.

In our patient, there was no overt manifestation of hypothyroidism but she had significantly reduced serum parathormone (iPTH) and vitamin D₃ consistent with primary hypoparathyroidism and treatment was started with aspirin, atorvastatin, vitamin D₃, calcium and calcitriol. Physiotherapy was initiated within 24 hrs and continued daily.

Movement disorder like dystonia and paratonia with Parkinsonism mimics are sometimes associated with cerebellar thalamic lenticular network dysfunction, and may be presenting symptoms of Fahr's syndrome [1,2]. Fahr's syndrome is often associated with other diseases like parathyroid dysfunction, usually hypoparathyroidism.

In our patient, there was no symptomatic movement disorder, but she had paratonic rigidity in her non-paretic upper limb which may be partially related to the basal ganglia degeneration. There has been one case report demonstrating acute ischemic stroke in Fahr's disease with positive MRI

findings of lacunar infarct in right thalamus [3]. Another case of lacunar infarct in left posterior limb of internal capsule has been reported [4]. Only one case of a young child with hypoparathyroidism has been reported having large artery thrombotic stroke [5].

Our patient was an elderly woman with Fahr's syndrome and hypoparathyroidism who had a large artery athero-thrombotic stroke, and probably first of its kind reported in literature.

Researchers have observed a distinct association between reduced circulating 25(OH)D and risk of ischemic stroke in hypertensives [6].

Conclusion

The risk factor association between ischemic stroke and Fahr's syndrome has yet to be established, but cannot be overlooked especially in patients with hypoparathyroidism and hypovitaminosis D, considering its potentially treatable nature, with control of calcium and phosphate homeostasis. It is recommended that clinicians first consider the possibility of hypoparathyroidism when looking for the cause of basal ganglia calcification.

References

1. Amir A, Ghanem AA. "Familial Idiopathic Basal Ganglia Calcification (Fahr's disease)". *Neurosciences (Riyadh)* 19 (2014): 171-7.
2. Sanjay BK, Bhansali A, Upreti V, Subbiah S, et al. "Spectrum of Neurological Manifestations of Idiopathic Hypoparathyroidism and Pseudohypoparathyroidism". *Neurol India* 59 (2011): 586-9.
3. Rocha E, Rouanet C, Reges DS, V, Gagliardi V, et al. "Fahr's Syndrome and Acute Ischemic Stroke: A Case Report". *Stroke and Neurocritical Care Division, Department of Neurology and Neurosurgery, Federal University of Sao Paulo-UNIFESP, Sao Paulo, Brazil*.
4. Chih Y, Lo CP, Wu MC. "Ischemic Stroke in a Young Patient With Fahr's Disease: A Case Report". *BMC Neurol* 16 (2016): 33.
5. Dhawan RS, Jondhale SN, Sahu JK, Vyas S, et al. "Large Artery Stroke in a Child With Hypoparathyroidism". *J Child Neurol* 30 (2015): 1084-5.
6. Christopher R, Majumdar V, Nagaraja D. "Vitamin D and Cerebrovascular Disease". *Ann Indian Acad Neurol* 24 (2021): 130-1.

How to cite this article: Choudhury K, Paul B, Roy K, Ghosh S. "Fahr's Syndrome with Hypoparathyroidism Presenting with Acute Ischemic Stroke". *J Clin Neurol Neurosurg* 4 (2021) : 126