Fahr's disease presenting with thalamic hemorrhage: a case report

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Case Report

Keywords: Fahr's disease, thalamic hemorrhage

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Abstract

Background: Fahr's disease is a rare neurological disorder which is characterized by diffuse intracranial calcification documented in bilateral basal ganglia and dentate nuclei of the cerebellum. In recent years, some acute presentations of Fahr's disease have been reported. Here we describe a Fahr's disease patient presenting with thalamic hemorrhage, which has not been reported so far.

Case presentation: A 51-year-old man doesn't have any prior premonitory symptoms and significant family history of neurologic disorder, he presented to our emergency department because of acute left limb numbness. Both brain CT and MRI study exhibited symmetric calcifications in bilateral basal ganglia and bilateral cerebellar dentate nuclei, which was consistent with Fahr's disease. In addition, the blood investigation provided no criteria to secondary intracranial calcification. After proper clinical medical treatment and sustained physical rehabilitation, the patient's symptoms relieved and he was discharged from hospital.

Conclusion: We describe a case of sporadic Fahr's disease presenting with thalamic hemorrhage. It is unknown whether the calcification in Fahr's disease negatively affects intracranial vessels predisposing acute presentation. Therefore, the association between idiopathic basal ganglia calcification and acute cerebrovascular disease is worth special attention and needs further studies.

Keywords: Fahr's disease, thalamic hemorrhage.

Background

Fahr's disease is a rare neurological disorder which was first described by Karl Theodor Fahr in 1930. [1] It is characterized by diffuse intracranial calcification documented in bilateral basal ganglia and dentate nuclei of the cerebellum, the Computed Tomography (CT) scan is the most accurate diagnostic test. Clinical symptoms don't present in most people at first, when the patients present cognitive or movement disorders, they can hardly be cured, but symptomatic treatment may be beneficial in some extent. Fahr's disease patients usually present with slow progressive cognitive impairment, psychiatric disturbance, or behavioral dysfunction.[2] However, some acute deterioration of Fahr's disease has gained more attention in recent years, including subarachnoid hemorrhage, aneurysmal subarachnoid hemorrhage and epileptic syncope. Here we describe a patient with acute sensory disability and both thalamic hemorrhage and idiopathic basal ganglia calcification was demonstrated in CT scanning.

Case Presentation

A 51-year-old man presented to our emergency department because of acute left limb numbness for 12 hours when he was working. He didn't present with any premonitory symptoms, cognitive and movement disorder prior to this event. No significant family history was provided, and he denied any significant previous medical history, or any neurological problem presented with transient consciousness or headache. At emergency room, his initial vital signs were: body temperature 37.0°C, pulse 82 beats/min,

respirations 19 breaths/min, blood pressure 160/80 mmHg. The general physical examination was unremarkable, the neurologic examination revealed nothing but pain and warm sense dysfunction of left limb, no cognitive disorder, no facial palsy, no dysarthria, manual muscle power grade 5, no meningeal irritation, no cerebellar ataxia etc. The laboratory examinations at emergency room were normal including blood cell count and serum electrolytes. Brain CT revealed (**Fig. 1**): 1. Right thalamus showed acute hemorrhage; 2. Extensive and symmetric calcifications involving the bilateral basal ganglia, bilateral cerebellar dentate nuclei, thalami, centrum semiovale; 3. Diffuse lacunar infarct in bilateral basal ganglia and centrum semiovale. Then the patient was admitted to neurologic ward with the tentative diagnosis of hemorrhagic stroke.

More investigations were made at hospitalization. The level of serum thyroxine, thyroid stimulating hormone(1.15 μ IU/L), inflammation indices, coagulation profiles, D-dimer level(0.26mg/L), hormone and enzyme-linked immunosorbant assay for human immunodeficiency virus (HIV), renal function indicator, liver function indicator, serum calcium (2.11 mmol/L), serum phosphorus (0.99 mmol/L), alkaline phosphatase(66 U/L), tumor markers were all within normal limits. Brain magnetic resonance imaging (MRI), magnetic resonance angiography (MRA) and diffusion-weighted imaging(DWI) showed (**Fig. 2**):1. Acute right thalamus hemorrhage involving the right thalamus. 2. Hyperintense signal showed in T1 weighted images and hypointense signal showed in T2 weighted images involving the bilateral basal ganglia, bilateral cerebellar dentate nuclei, thalami, centrum semiovale, which is coincident with CT scans. 3. No vascular malformation was found in MRA.

Triclotin and cerebroprotein hydrolysate and tranexamic acid were regularly administered for 8 days, and the sensory disturbance of left limb was improved. Because of the cost, the patient was discharged according to his requirement. Before leaving, his doctor taught him how to make physical rehabilitation and come back to hospital for body check in a week. 13 days later, the patient was followed up with brain CT scanning in neurosurgery clinic (**Fig. 3**).

Discussion And Conclusions

Fahr's disease, or idiopathic basal ganglia calcification is a rare inherited or sporadic neurological disorder that is characterized by abnormal intracranial calcification without biochemical abnormalities and significant family history. [3]The areas of abnormal calcium deposition has been described in bilateral basal ganglia, dentate nuclei of the cerebellum, caudate nucleus, the globus pallidus, thalamus, putamen, the white matter of the cerebral cortex, as well as major intracranial vessels etc.[4] Some patients may remain asymptomatic all their life. Affected patients present typical clinical express mainly in their fourth to sixth decade. They exhibit distinctive neuroradiological features and variable clinical presentations.[4] Most of population display slow progressive impairment. However, various acute presentations have been described in recent years, including epilepsy,[5] syncope[6], aneurysmal hemorrhage[7, 8], ischemic stroke[9], Subarachnoid hemorrhage. Fahr's disease presenting with thalamic hemorrhage has not been reported to date.

Differential diagnosis to rule out endocrine disease is essential. [10]As for this patient, The laboratory studies did not provide criteria of idiopathic hypoparathyroidism, secondary hypoparathyroidism, hyperparathyroidism, post-thyroidectomy, HIV infection...etc.[10] And the brain MRA didn't indicate vascular abnormality, aneurysm or vascular malformation. Brain CT and MRI study both exhibited symmetric calcifications in the bilateral basal ganglia, bilateral cerebellar dentate nuclei, which is consistent with Fahr's disease. The patient denied similar familial illness, systematic disease, metabolic disorder or hypoxia history. Therefore, we considered him as a sporadic case of Fahr's disease. As reported by Hosam Al-Jehani, we hypothesis that the extensive calcification in Fahr's disease negatively affects intracranial vessels predisposing these individuals to such an acute presentation. Therefore, the association between idiopathic basal ganglia calcification and acute cerebrovascular disease is worth special attention and the exact mechanism needs further investigation.

Abbreviations

CT: Computed Tomography

HIV: human immunodeficiency virus

MRI: magnetic resonance imaging

MRA: magnetic resonance angiography

DWI: diffusion-weighted imaging

Declarations

Ethics approval and consent to participate

All procedures performed in studies involving human participants were in accordance with the ethical standards of the ethics committee of Thaizhou Municipal Hospital of Zhejiang Province and with the 1964 Helsinki declaration and its later amendments or comparable ethical standards.

Consent for publication

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

Availability of data and material

The data supporting our findings has been presented within the manuscript.

Competing interests

The authors declare that they have no conflict of interest.

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Author's contributions

GTW examined the patient and drafted the manuscript. ZNY and JC evaluated the neuroimaging findings and gave the important clinical opinions. GHM participated in the design of the case report and helped to draft the manuscript. All authors read and approved the final manuscript.

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Figures

Figure 1

First brain CT scanning - (1) Extensive and symmetric calcifications can be observed in a. bilateral cerebellar dentate nuclei, b. Bilateral basal ganglia and thalami, c. centrum semiovale. (2) Acute hemorrhage involving the right thalamus (arrowhead in b).

Figure 2

MRI, MRA and DWI - a. T1 weighted images showed hyperintense signal in bilateral basal ganglia (arrowed), thalami (arrowed) and cerebellar dentate nuclei; b. T2 weighted images showed hypointense signal in bilateral basal ganglia (arrowed), thalami (arrowed) and cerebellar dentate nuclei; c. No vascular malformation or hemangioma was found.

Figure 3

Reexamining brain CT scanning - liquefactive necrosis in a cerebral hemorrhage showed in right thalamus, extensive and symmetric calcifications is same with before.

Supplementary Files

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