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A Case of Seizure Revealing Fahr's Syndrome with Primary Hypoparathyroidism

Authors' Contribution:
Study Design A
Data Collection B
Statistical Analysis C
Data Interpretation D
Manuscript Preparation E
Literature Search F
Funds Collection G

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Conflict of interest: None declared

Patient: Male, 52
Final Diagnosis: Fahr's syndrome
Symptoms: Seizure
Medication: —
Clinical Procedure: —
Specialty: Endocrinology and Metabolic

Objective: Rare disease

Background: Idiopathic basal ganglia calcification, also known as Fahr's disease or Fahr's syndrome, is a rare neurological disorder characterized by abnormal calcified deposits in the basal ganglia. Here, we report a case of Fahr's syndrome with calcification of the basal ganglia due to hypoparathyroidism in a patient with seizures.

Case Report: A 52-year-old male patient visited our clinic with seizures. Brain computed tomography (CT) showed bilateral symmetrical calcifications in cerebellar white matter, the corpus striatum, the posterior thalami, and the centrum semiovale of both cerebral hemispheres. He had symptoms of hypocalcemia and low parathyroid hormone levels. The patient was diagnosed with Fahr's syndrome due to primary hypoparathyroidism. He underwent calcium supplementation and calcifediol treatment. His symptoms improved, and he was discharged from the hospital.

Conclusions: In patients with hypocalcemia accompanied by parathyroid dysfunction, neurological examination and CT should be performed to confirm abnormal intracranial calcification.

MeSH Keywords: Basal Ganglia Diseases • Hypothyroidism • Seizures

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Background

Fahr's syndrome, also known as Fahr's disease or idiopathic basal ganglia calcification, is a rare neurological disorder characterized by abnormal calcified deposits in the basal ganglia. Fahr's syndrome was first reported in 1930 by Karl Theodor Fahr [1]. Diagnostic criteria of Fahr's syndrome are as follows: bilateral calcification of the basal ganglia on neuroimaging, progressive neurologic dysfunction, absence of biochemical abnormalities, and family history consistent with autosomal dominant inheritance [2,3]. The term Fahr's disease is used when primary familial brain calcification is present, and the term Fahr's syndrome is used for secondary causes [4]. It is an inherited or sporadic neurological disorder with a prevalence of <1/1 000 000 [2]. Most of the literature on Fahr's disease is based on case reports with few studies on the prevalence of the disease. Previously, in Korea, cases have been reported of epileptic seizure in pseudohypoparathyroidism [5] and recurrent epileptic seizure in idiopathic hypoparathyroidism [6]. Here, we report a case of Fahr's syndrome with calcification of the basal ganglia caused by hypoparathyroidism in a patient with seizures.

Case Report

A 52-year-old male patient visited our clinic with seizures. Suddenly, while drinking alcohol, he fell to the side, his face became pale, and he showed signs of a generalized tonic-clonic type seizure during which his arms and legs were slightly tilted for about 10 minutes. There was no aura or focal deficit. He did not have a history of seizures, head trauma, central nervous system infection, stroke, hypertension, diabetes, thyroid disease, or autoimmune disease. He did not have a family history of epilepsy; furthermore, his brothers and children were healthy. He had been drinking alcohol for 28 days out of 30 days, but did not smoke. His vital signs were as follows: blood pressure was 110/70 mm Hg; pulse rate was 74 beat per minutes; respiratory rate was 20 breaths per minutes; and body temperature was 36.3°C. Neurological examination revealed clear consciousness, no cognitive impairment, and no cranial nerve abnormalities. The laboratory test results were as follows: hemoglobin (Hb) 13.2 g/dL, white blood cell count (WBC) 7.70×10^9/L; platelet 300×10^9/L, glucose 111 mg/dL, prothrombin time (PT) 12.3 sec; activated partial thromboplastin time (aPTT) 29.2 sec; aspartate aminotransferase (ASL) 22 U/L; alanine aminotransferase (ALT) 14 U/L; total bilirubin 0.3 mg/dL; protein 7.5 g/dL; albumin 4.1 g/dL; creatinine 0.85 mg/dL; potassium 3.5 mmol/L; calcium 4.8 mg/dL; phosphorus 4.8 mg/dL; magnesium 2.1 mg/dL (1.9–3.1 mg/dL); ethanol 168.8 mg/dL; intact parathormone (PTH) 3.5 pg/dL (15–65 pg/dL); 25-hydroxy (OH) total vitamin D (D2+D3) 25.1 ng/mL (4.8–52.8 ng/mL); 25-OH vitamin D3 29.12 ng/mL (8.0–51.9 ng/mL); anti-nuclear antibody negative; P-anti neutrophil cytoplasmic antibody negative; C-anti neutrophil cytoplasmic antibody negative; spot urine creatinine 69.38 mg/dL; and spot urine calcium 12.0 mg/dL. There were no unusual findings from the chest x-rays upon admission. Thyroid sonography did not reveal any abnormal findings in the thyroid or its periphery. However, brain computed tomography (CT) showed bilateral symmetrical calcifications in cerebellar white matter, the corpus striatum, the posterior thalami and the centrum semiovale of both cerebral hemispheres. Results from 32 channel digital electroencephalography (EEG) with electrocardiography (ECG) monitoring was performed, but revealed that there were no definite interictal epileptiform discharges or focal slowings (Figure 1). The patient's reduced parathyroid hormone levels suggested hypocalcemia due to hypoparathyroidism. Primary hypoparathyroidism was diagnosed because there were no indications of secondary causes such as a history of congenital anomalies, thyroid surgery, history of neck radiation, history of autoimmune disease, or hypomagnesemia. In response to the seizures, hypocalcemia correction was performed. The patient received 2 g of calcium gluconate, administered, followed by 10 g of calcium gluconate for 24 hours intravenously. Additionally, oral medication started. 1000 mg calcium carbonate, $CaCO_3$, was administered 3 times a day, while 20 mcg calcifediol and 12.5 mg hydrochlorothiazide were administered once daily. Topiramate 50 mg for seizure was started twice a day. At the time of discharge, education was given to reduce alcohol consumption. The patient was diagnosed with Fahr's syndrome due to primary hypoparathyroidism based on the CT and laboratory findings. The patient was discharged from the hospital on the 17th day after admission and is undergoing outpatient follow-up without symptoms such as stiffness or seizures.

Discussion

Fahr's syndrome is suspected to be a calcification of the basal ganglia of the brain based on brain imaging studies of patients with neurological or psychiatric symptoms such as seizure, dyskinesia, dementia, and depression. Related diseases include hyperparathyroidism, hypoparathyroidism, hypoparathyroidism, hypervitaminosis D, tuberculosis, cytomegalovirus infection, toxoplasmosis, and astrocytoma [7,8].

Brain calcification in Fahr's syndrome is widespread at various sites; therefore, there is a high frequency of neurological symptoms. In Fahr's syndrome, many areas of the brain can exhibit calcification including the cerebellum, thalamus, frontal lobe, temporal lobe, and the basal ganglia. If brain calcifications are detected in imaging studies, neoplastic, infectious, congenital (eg tuberous sclerosis), endocrine/metabolic causes, and idiopathic causes should be differentiated. Because of the variety of causes, such as infectious diseases and endocrine ailments, Fahr's syndrome patients may have various neurological signs

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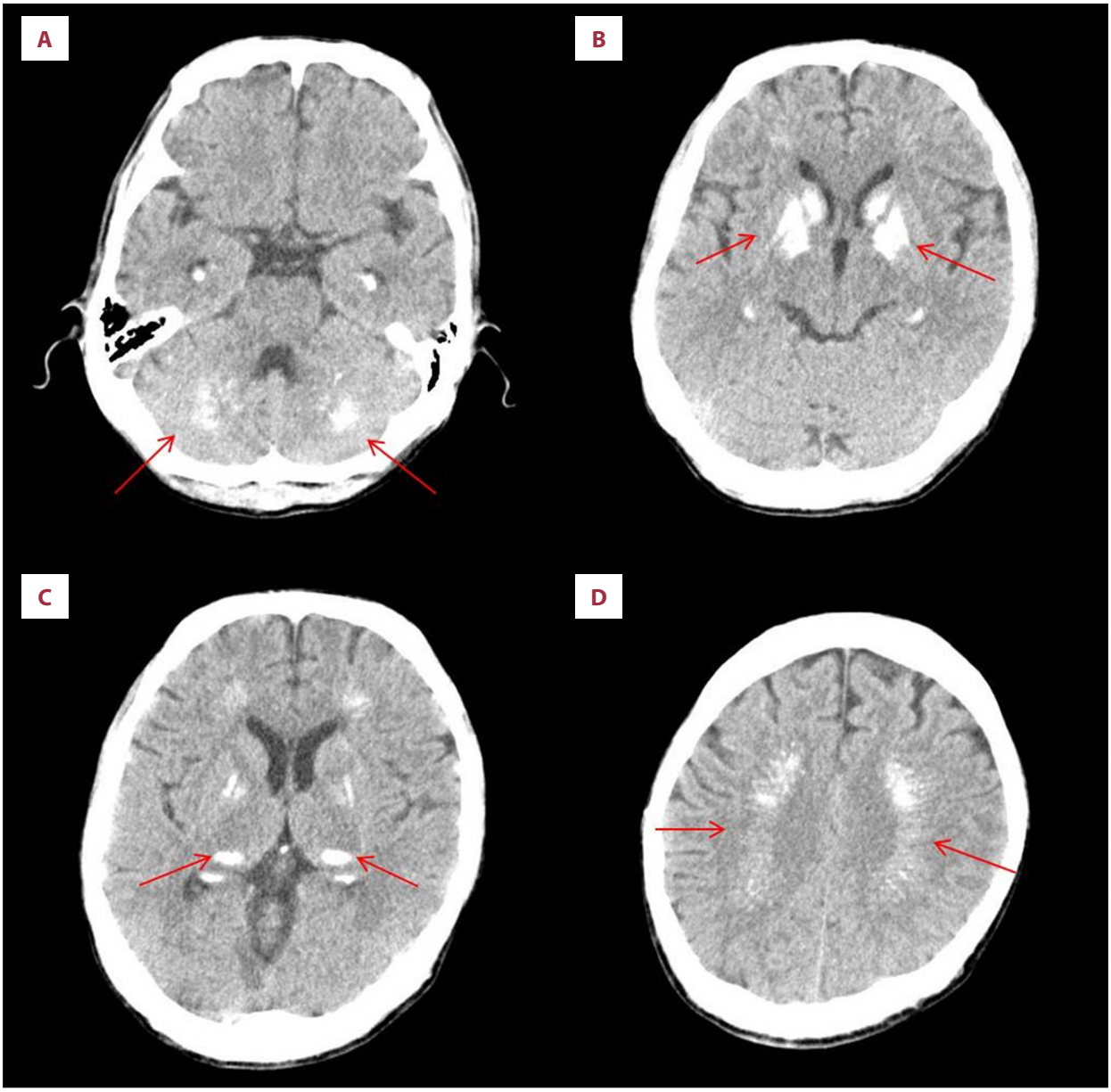

Figure 1. Brain computed tomography (CT) showed bilateral symmetrical calcification in cerebellar white matter, the corpus striatum, the posterior thalami, and the centrum semiovale of both cerebral hemispheres: (**A**) cerebellum white matter; (**B**) corpus striatum; (**C**) posterior thalami; (**D**) centrum semiovale of both cerebral hemispheres.

or symptoms such as seizures, dystonia, myoclonus, and Parkinson's disease [9]. For example, Hempel et al. analyzed positron emission tomography (PET) images of patients with Fahr's disease and found that glucose uptake was reduced in the temporal lobe, parietal lobe, and the basal ganglia [10]; these functional changes in the brain cause a variety of mental neurological symptoms. Additionally, Lopez-Vilegas et al. reported that 18 patients with Fahr's syndrome had different neurological symptoms according to calcified areas of the basal ganglia [11].

Although there are many specific hypotheses, the causes of abnormal calcification in the brain are related to parathyroid dysfunction or other causes of calcium metabolism overall [12]. In the pathologic examination of Fahr's syndrome, calcium deposits were present in extracellular or extravascular space, especially around the capillaries. However, it is not clear whether abnormal calcium deposition in the brain is caused by the local destruction of the blood brain barrier or by calcium metabolic disorder of neurons [13].

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Conclusions

We report a case of Fahr's syndrome with calcification of the basal ganglia due to hypoparathyroidism in a patient with seizures as the main symptom. Although rare, Fahr's syndrome should be suspected if there is symmetrical and abnormal calcification of both basal ganglia, visible via brain CT, of patients with seizures or loss of consciousness. Moreover, thyroid and parathyroid function should be tested along with the calcium concentration in the blood; calcium concentration correction should be started early if hypocalcemia is present. We believe that confirming brain imaging in hypoparathyroidism patients with all neurological symptoms will help to improve the prognosis.

Conflicts of interest

None.

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