

A rare case of Fahr's disease presenting as epileptic seizure in the emergency department

在急症室呈現癲癇發作的一個法爾病的罕有個案

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Fahr's disease (striato-pallido-dentate calcifications) is a rare neurodegenerative disorder characterised by bihemispherical and symmetrical calcium deposits in certain areas of the brain, particularly in the basal ganglia. We report an unusual case of Fahr's disease in a 36-year-old man who presented with generalised seizure in our emergency department. Basing on clinical, radiological, and endocrinological aspects, the patient was diagnosed as Fahr's disease associated with hypoparathyroidism. Administration of parenteral calcium and calcitriol supplementation were started in the emergency department. The clinical outcome was favourable after the treatment. The case illustrates that Fahr's disease, though rarely seen, has to be considered in a patient with convulsive state associated with calcifications of the basal ganglia. Systematically searching for dysparathyroidism in such patients is extremely important. (*Hong Kong j.emerg.med.* 2010;17:262-264)

法爾病（紋狀體·蒼白球·齒狀核鈣化）是罕見的神經退化病症，特徵為在腦部一些地區，特別是基底神經節，有兩半球對稱的鈣沉積。本文報告一個不尋常的法爾病個案，一名36歲男子因全身性抽搐而到急症室求診。基於臨床、放射學及內分泌學各方面，病人被診斷為與甲狀旁腺機能減退有關的法爾病。在急症室開始注射鈣及補充骨化三醇。治療後的臨床結果滿意。這個案顯示有關基底神經節鈣化而在抽搐狀態下的病人，要考慮罕見的法爾病。在這些病人有系統地搜查甲狀旁腺機能障礙是極重要的。

Keywords: Basal ganglia disease, calcinosis, epilepsy, hypoparathyroidism, X-ray computed tomography

關鍵詞：基底神經節疾病、鈣質沉著、癲癇、甲狀旁腺機能減退、X光電腦掃描

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Introduction

Bilateral basal ganglionic calcification, also known as Fahr's disease (FD) or Fahr's syndrome is a rare entity characterised by symmetrical and bilateral calcifications over the basal ganglia, thalami, cerebellar dentate nucleus or white matter of the cerebral hemispheres.¹ Clinical manifestations of Fahr's syndrome varies greatly. Common clinical features include movement disorders such as parkinsonism, speech disorders, psychiatric disorders, epileptic seizure, dementia, cerebellar or extra-pyramidal dysfunction. Some cases with FD may present without neurological abnormalities.^{2,3} Cranial computed tomography (CT) is employed for diagnosing FD.³ The origin and

pathophysiology of this condition are unknown and the outcomes of treatment are often unsatisfactory.⁴ It may be sporadic or familial as well as secondary to anoxia, irradiation, systemic disorders, toxins, and disorders of calcium metabolism.^{5,6} The main cause is dysparathyroidism particularly hypoparathyroidism, whether idiopathic or post-surgery.⁷ We report a very rare case of FD due to idiopathic hypoparathyroidism in a young man diagnosed by clinical and radiological evidences.

Case report

A 36-year-old male presented with sudden onset of generalised tonic-clonic seizure to our emergency department (ED) in April 2009. Oropharyngeal airway was inserted to maintain the unconscious patient's airway. Midazolam was given as the first-line antiepileptic drug, and the seizure was responsive to the treatment. After the patient regained consciousness, a neurological examination was performed. Abnormal neurological signs including dysarthria, hemiparesis, paresthesia and generalised neuromuscular irritability such as muscle cramps and tetany were found. Latent tetanic convulsions could also be displayed through the elicitation of Chvostek's sign and Trousseau's sign. There were convulsive episodes diagnosed as epilepsy in his past medical history. Laboratory studies including serum calcium 4.6 mg/dl (normal 8.4-10.6 mg/dl), phosphate 7.5 mg/dl (normal 2.3-4.7 mg/dl) and parathormone level 1.2 pg/ml (normal 15-65 pg/ml) demonstrated idiopathic hypoparathyroidism. Additional diagnostic laboratory tests including thyroid hormones and vitamins were within normal range. Sonographic examination revealed normal thyroid gland and parathyroid glands. Cranial CT scan showed extensive symmetrical intracerebral calcifications in both basal ganglia and internal capsules (Figure 1). In addition, bilateral and symmetrical calcifications were also detected over the lateral periventricular areas (Figure 2). A diagnosis of Fahr's disease associated with idiopathic hypoparathyroidism was made on the basis of the clinical, laboratory and radiological findings. Parenteral calcium, calcitriol supplementation and intravenous fluid support were given in the ED. The

clinical outcome was favourable after the therapy. The patient's abnormal neurological signs detected upon admission to the ED and serum calcium and phosphate levels improved gradually over the next 12 hours. During the observation in the ED, no other abnormalities were noted on physical examination.

Discussion

Physiological intracranial calcification occurs in about 1.3-1.5% of cases. It is asymptomatic and is detected incidentally by neuroimaging. Pathological basal ganglia calcification is due to various causes such as

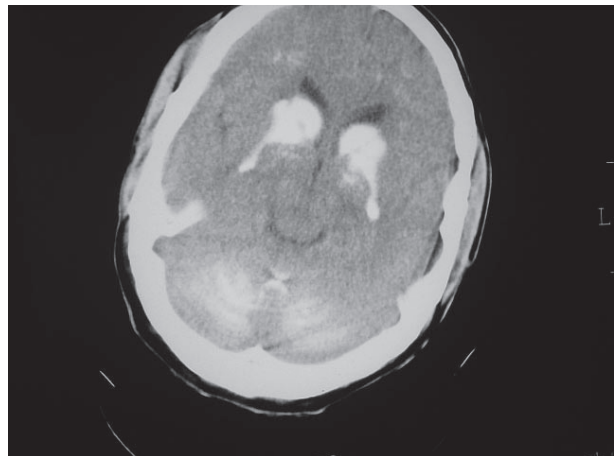


Figure 1. Cranial CT axial section, showing bilateral calcifications in the basal ganglia and internal capsules.

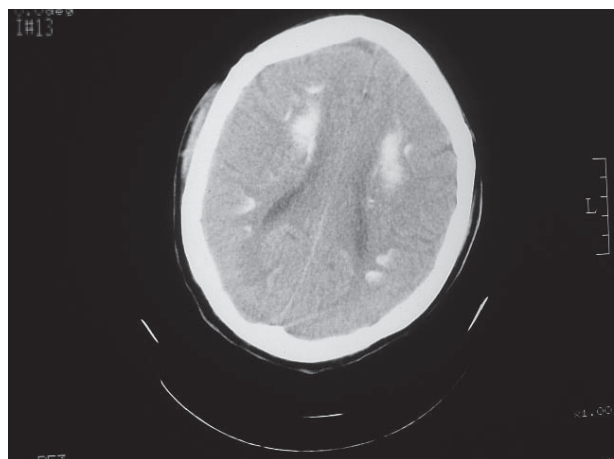


Figure 2. Bilateral calcifications in lateral periventricular areas.

metabolic disorders, infectious or genetic diseases.⁵ Metabolic derangements, especially parathyroid disorders related to Fahr's disease, are the most common causes of pathological basal ganglia calcifications.^{6,7}

The clinical course of the disease has a degenerative component which is often slowly progressive, lasting for many years. The mineral deposition may lead to cell loss in the cerebral cortex, basal ganglia, dentate nucleus or subthalamus.⁸ Clinical diagnosis is facilitated by the presence of bilateral and symmetrical calcifications in the basal ganglia in cranial CT. Detection of intracranial calcifications in CT scan is more sensitive comparing with skull X-ray or MRI.^{4,8} It was reported in the literature that in Fahr's disease, there were symmetrical and extensive calcifications in the white matter of the cerebral or cerebellar hemispheres and basal ganglia.^{4,8-10} As in the present case, the cranial CT findings were typical and consistent with those of FD. Clinical presentations may be quite variable. Neurological and psychiatric symptoms, if present at all, are highly variable and include progressive mental deterioration, convulsive seizures, parkinsonism, difficulty in speaking, ataxia, psychosis or affective disorders.^{1,9} In the present case, there were no psychiatric symptoms. There were neurological signs such as dysarthria or those of hypoparathyroidism such as latent tetany, seizure and hemiparesis. In suspected cases of FD, other causes of intracranial calcification should be considered such as cerebrovascular lesions, infectious diseases like toxoplasmosis, syphilis and inflammatory illnesses such as systemic lupus erythematosus.^{3,10} There is no satisfactory cure for FD or standard course of treatment. Treatment addresses symptoms on an individual basis.^{5,6} Correction of calcium and phosphate levels may lead to clinical improvement, in particular, disappearance of epileptic seizures and abnormal movements.⁷ As in the present case, after treatment with parenteral calcium and calcitriol supplementation, there were clinical and laboratory improvements.

Conclusion

The present case emphasizes that the rarely seen Fahr's syndrome should be considered in patients presenting with convulsion. FD is associated with various metabolic disorders especially with the parathyroid. It should be considered in patients with bilateral and symmetrical calcifications on cranial CT that cannot be explained by other causes of intracranial calcifications. Clinical features of neurological symptoms, laboratory findings of hypoparathyroidism and radiological features of intracranial calcifications form the basis of diagnosis of FD.

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