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Case Report

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Fahr's Syndrome: A Report of Two Cases

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ABSTRACT

Fahr's syndrome is a rare clinical entity that presents mainly with extrapyramidal signs and accompanied with metabolic, biochemical, neuroradiological and neuro-psychiatric situations at the same time. Bilateral intracranial calcifications are usually encountered as an incidental radiological finding. In this study, we discuss two cases of Fahr's syndrome in the light of the literature; one of them idiopathic and the other one occurring secondary to hypoparathyroidism. Clinically, one of the cases presented with seizures and the other one with imbalance of the coordination system. Cranial computerized scans of both patients revealed intracranial diffuse bilateral calcifications in thalamus, basal ganglia and cerebellum. Fahr's syndrome, although encountered rarely, should also be taken into account in the differential diagnosis of cases with abnormal intracranial calcifications along with other familial, congenital and metabolic diseases and syndromes.*©2007, Fırat Üniversitesi, Tıp Fakültesi*

Key words: *Fahr's syndrome, hypoparathyroidism, epilepsy, bilateral intracranial calcifications*

ÖZET

Fahr's Sendromu: İki Olgunun Raporları

Fahr sendromu esas olarak ekstrapiramidal bulguların ve aynı zaman dametabolik,biyokimyasal, nöroradyolijik ve nöropsikiyatrik durumların birlikte prezente oldugu nadir bir klınik antitedir. Genellikle bilateralin trakraniyal kalsifikasyonlar insidental radyolijik bulgudur. Bu calışmada, biz iki fahr sendromlu olguyu literatur ışıgında tartışdık.Olgulardan birisi idiyopatik ve digeri hipoparatiroidizme sekonder oluşan olgu idi. Klinik olarak olgulardan biri nöbet ile ve digeri denge bozuklugu ile prezente oldu. Heriki hastanin kranial bilgisayarlı tomografisinde talamus, bazal ganglionlar ve serebellumda intrakranyaldiffuz bilateral kalsifikasyonlar mevcut idi. Fahr sendromu nadir karsılaşılan bir antite olmasına ragmen, anormal intrakraniyal kalsifikasyonlu vakaların ayırıcı tanısında diger familyal, konjenital ve metabolik hastaliklar ve sendromlar ile birlikte göz önünde bulundurulmalıdır. *©2007, Fırat Üniversitesi, Tıp Fakültesi*

Anahtar kelimeler: *Fahr's sendromu, hipoparatiroidizm, epilepsi, bilateral intrakranial kalsifikasyonlar.*

Fahr's syndrome is characterized clinically by seizures, extrapyramidal and neuro-psychiatric signs as a result of bilateral diffuse calcifications of the basal ganglia, dentate nucleus and white matter (1-4). Intracranial calcifications are encountered accidentally in 0.3-1.2% of routine radiological examinations (5,6). Bilateral basal ganglion calcifications can be related with various pathological conditions including disorders of calcium-phosphate metabolism, carbon monoxide or lead intoxication, birth anoxia, therapeutical radiation, methotrexate and long term anticonvulsant therapy and some inflammatory and infectious diseases. Endocrine disorders such as hypoparathyroidism are also important conditions in the etiology of the basal ganglia calcification so as idiopathic Fahr's disease (6-9) (Table 1).

Bilateral striopallidodentate calcinosis (also known as Fahr's disease) is a rare neurodegenerative disease, characterized by bilateral symmetrical deposition of calcium and other minerals in basal ganglia, thalamus, dentate nucleus and centrum semiovale. Fahr's syndrome is a general term including not only Fahr's disease but also other conditions presenting with secondary basal ganglia calcifications (2,8).

In this study, we present two cases diagnosed as Fahr's syndrome with their clinical signs and implicating the treatment alternatives under the light of the literature.

CASE REPORT

Case 1

A 60-year-old male, presented with the complaint of seizure disorder lasting since 2001. According to his medical history, his complaints started with contractions in his hands when he was 17 years old. He mentioned about speech disorder after two years and inclusion of seizures 3 years ago. His seizures were resistant to antiepileptic medical treatment reaching the frequency of 2-3 seizures in a day recently. Irritability was prominent since the beginning of his sickness, getting worse also with the addition of forgetfulness in the recent years.

Patient's physical examination revealed urinary incontinence. There was disorientation to place and time and dysphonia whereas his consciousness was full. Neurological examination showed less visual acuity in the left eye as a result of lenticular opacity, increased hypertonicity and impaired cerebellar tests.

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Laboratory investigations showed normal levels of whole blood count, sedimentation rate, fasting blood glucose, blood electrolytes, thyroid hormones and ferritin. Blood calcium (4.5mg/dl), ionized calcium and parathyroid hormone levels (5.00pg/ml) were lower and phosphorus (6.5mg/dl) higher than normal levels. Neither EEG nor ECG or echocardiogram showed any abnormality. Computerized tomography of the brain demonstrated diffuse bilateral parenchymal calcifications most prominent in the central portions of the cerebellar hemispheres, head of the nucleus caudatus, anterior of the lenticular nucleus, anterior genus of the capsula interna and corona radiata (prominent in the frontal region) (Figure 1).

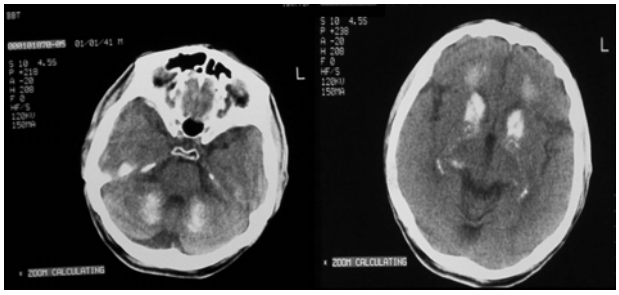

Figure 1. Bilateral cerebellar calcifications most prominent in dentate nucleus.

Table 1: Etiological factors of basal ganglia calcifications

Idiopathic hypoparathyroidism
Secondary hypoparathyroidism
Pseudo-hypoparathyroidism
Pseudo Pseudo-hypoparathyroidism
Hyper-hypoparathyroidism
Hypothyroidism
Perinatal asphyxia
Carbonmonoxide intoxication
Lead intoxication
Fahr's syndrome
Familial idiopathic symmetrical basal ganglia calcification
Hastings-James syndrome
Cockayne's syndrome
Lipoid proteinozis
Tubero sclerosis
Parkinsonism
Vascular diseases
Cerebral hemorrhage
Radiation therapy
Methotrexate treatment
Cytomegaly inclusion disease
Ancephalitis
Toxoplasmosis
Cysticercosis
Sturge Weber syndrome
Neurofibromatosis

The patient was treated with intravenous calcium infusion followed by parenteral calcium supplementation and alfacalcidiol medication. Neurological deficits of the patient regressed in 2 days along with urinary incontinence. He experienced no seizures during his follow-up period also with amelioration of his urinary incontinence, speech disorder and contractures of his hands. Mini-Mental State exam score of 21/30 was obtained after his orientation improved.

CASE 2

A 50-year-old male referred to our clinics with a history of recently getting worse speech disorder, ongoing for 10 years. According to his past medical history he had no seizure disorder. Physical examination was normal whereas neurological exam revealed dysarthria, bilateral mild dysdiadochokinesis and dysmetria. He experienced no spasticity.

Laboratory investigations including whole blood count, urine analysis, sedimentation rate, fasting blood glucose, blood electrolytes, thyroid hormones, blood serum iron, ferritin, total iron binding capacity, showed no abnormality. Blood calcium 8.19 mg/dl (8.2-10.2 mg/dl) and parathyroid hormone levels 36.306 pg/ml (15-65 pg/ml) were in normal limits. The patient had normal ECG with no abnormality on his EEG examination. Computerized tomography of the brain demonstrated hyperdense bilateral calcifications in basal ganglia, thalamus and orbitofrontal cortex (Figure 2).

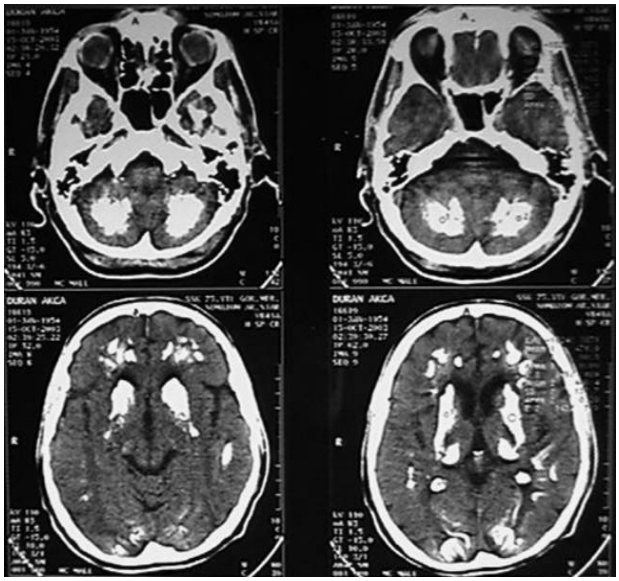

Figure 2. Cranial CT scan revealing bilateral basal ganglia calcifications.

DISCUSSION

Bilateral symmetrical calcifications of the basal ganglia and cerebellar nuclei are denominated as Fahr's syndrome following the publication of Fahr in 1930. Thereafter, the classical triads of this sydrome consisting of Bilateral Striopallidodentate Calcinosis (BPSDC), Hypoparathyroidism neurological manifestations are defined. (10-12).

Basal ganglia calcifications can originate from more than 30 medical conditions including some infectious, metabolic and genetic conditions (Table 1). Although most of these conditions are systemic diseases, the reason of the focal accumulation of calcium in basal ganglia is not known well currently (13-16).

About half of the patients with basal ganglia calcifications exhibit neurological manifestations and deficits. The most common neurological signs are headache, vertigo, movement disorders, syncope and seizures. Other specific neurological deficits consist of paresis, spasticity, gait disturbance, speech disorders, coma, dementia, Parkinsonism, chorea, tremor, dystonia, myoclonia and orthostatic hypotension (13,14).

Manyam et al. reported movement disorders in 56% and seizures in 22% of cases in a review of 213 patients with this disorder (13). Seizure disorder was prominent in our first case whereas coordination system deficits in the second. Our first patient referred to our clinics with medically intractable seizure disorder. Furthermore his seizures increased in number despite the constitution of the anticonvulsant therapy. There is no evidence that anticonvulsant drugs may give rise to calcifications but its coincidence with intracranial calcifications is well known (3,11,15).

Forty percent of patients with basal ganglia calcifications present with psychiatric symptoms at the beginning of the disease. Among these; cognitive and psychotic disorders are most prominent (7,9,14). Our first case also manifested severe irritability lasting for a long time with the addition of amnesia at the latter terms of his disease. Therefore his mini mental status examination revealed a low score (21/30).

According to the laboratory findings, first of our cases was thought to be secondary to hypoparathyroidism and the second as idiopathic Fahr's syndrome. Calcifications, originating from primary hypoparathyroidism are more diffuse than other situations resulting from various etiologies such as post-thyroidectomy hypoparathyroidism. Our first case that we thought about primary hypoparathyroidism as the causative etiological factor also showed diffuse parenchyma calcifications in his CT scan of the brain.

Early treatment of hypoparathyroidism with supplementation of Calcium and vitamin D can prevent calcifications and probable neurophysiological disorders. Our case also demonstrated major benefits with supplementation of calcium and alpha-calcidiol although he was in the late stage of the disease. He exhibited no seizure during his follow-up period and his laboratory findings were in normal limits. We did not institute any treatment for our idiopathic Fahr's syndrome case and just kept him under clinical follow-up.

As a conclusion, we presented 2 cases of intracranial calcifications resulting from hypoparathyroidism and idiopathic, in this study. We want to emphasize that hypoparathyroidism cases which show great clinical improvement from medical treatment must be kept in mind as a secondary causative factor of intracranial calcifications and must be investigated with advanced laboratory tests.

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