North Clin Istanbul 2016;3(1):71-4
doi: 10.14744/nci.2015.47966

CASE REPORT NEUROLOGY

Fahr's syndrome presenting with epileptic seizure: Two case reports

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ABSTRACT

Fahr's syndrome is a neuropsychiatric syndrome characterized by symmetrical and bilateral intracerebral calcifications located in the basal ganglia and usually associated with a phosphorus and calcium metabolism disorder. Clinical manifestations of Fahr's syndrome vary; it may start at different ages and have a variety of presentations. This article discusses rare presentation of Fahr's syndrome with epileptic seizure. These cases are important because they appear to be the first cases in the literature of Fahr's syndrome presenting with generalized tonic clonic seizure.

Keywords: Epilepsy; Fahr's syndrome; intracerebral calcification.

Extensive cerebral calcification may occur idiopathically as Fahr's syndrome, or may arise from secondary metabolic disorders, such as hypoparathyroidism. Fahr's syndrome is a neuropsychiatric syndrome characterized by symmetrical and bilateral intracerebral calcifications located in the basal ganglia and usually associated with a phosphorus and calcium metabolism disorder. Fahr's syndrome or Striato-pallido-dentate calcification (SPDC) is a well-defined entity with familial or sporadic presentation and approximately two-thirds of patients are symptomatic. It may clinically present with an array of movement disorders, dementia, epileptic seizures, various degrees of neuropsychological impairment and behavioral disturbances [1].

The present report is of rare presentation of epileptic seizure in 2 patients with Fahr's syndrome.

CASE REPORT

Case 1– A 52-year-old male was examined in the clinic on first generalized tonic clonic seizure. His neurological examination was normal. He was hospitalized and evaluated for differential diagnosis of epileptic seizure. Blood tests were normal, except decreased levels of parathormone (6.46 pg/mL [Range: 15-65 pg/mL] and calcium (5.7 mg/dL [Range: 8.6-10.2 mg/dL]). Medical history included subtotal thyroidectomy 20 years prior. He was not taking any medication, and there was no

Received: July 26, 2014 *Accepted:* June 18, 2015 *Online:* May 01, 2016

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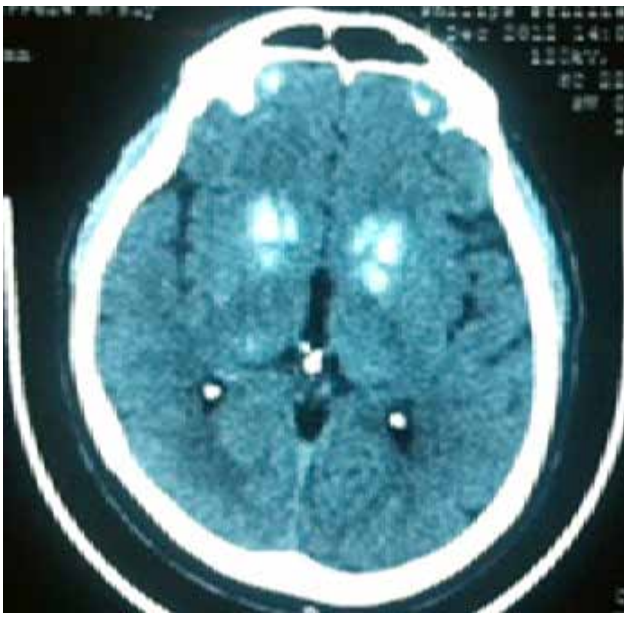

Figure 1. Brain computerized tomography: Extensive, bilateral calcification of basal ganglia.

family history of dementia, movement disorder or other neurological illness. His brain computerized tomography (CT) scans demonstrated extensive, bilateral calcification of basal ganglia (Figure 1). His electroencephalographic (EEG) examination was normal. It was his first and last seizure. Clinical evaluation was acute symptomatic seizure. No antiepileptic treatment was recommended and underlying situation was medicated by endocrinologists.

Case 2– A 56-year-old male who had his first epileptic seizure, a generalized tonic clonic motor seizure, was referred to the hospital. Blood tests were in normal range, except decreased vitamin D level (12 ug/L [Range: 20-120 ug/L]). There was no patient medical history or family history of neurological illness. His CT scans demonstrated bilateral calcification of basal ganglia and cerebellum (Figure 2). His EEG examination showed left frontal epileptiform activity. Though it was patient's first seizure, sodium valproat was prescribed for 6 months in addition to vitamin D replacement treatment because of the EEG abnormality and no further seizure was seen after treatment.

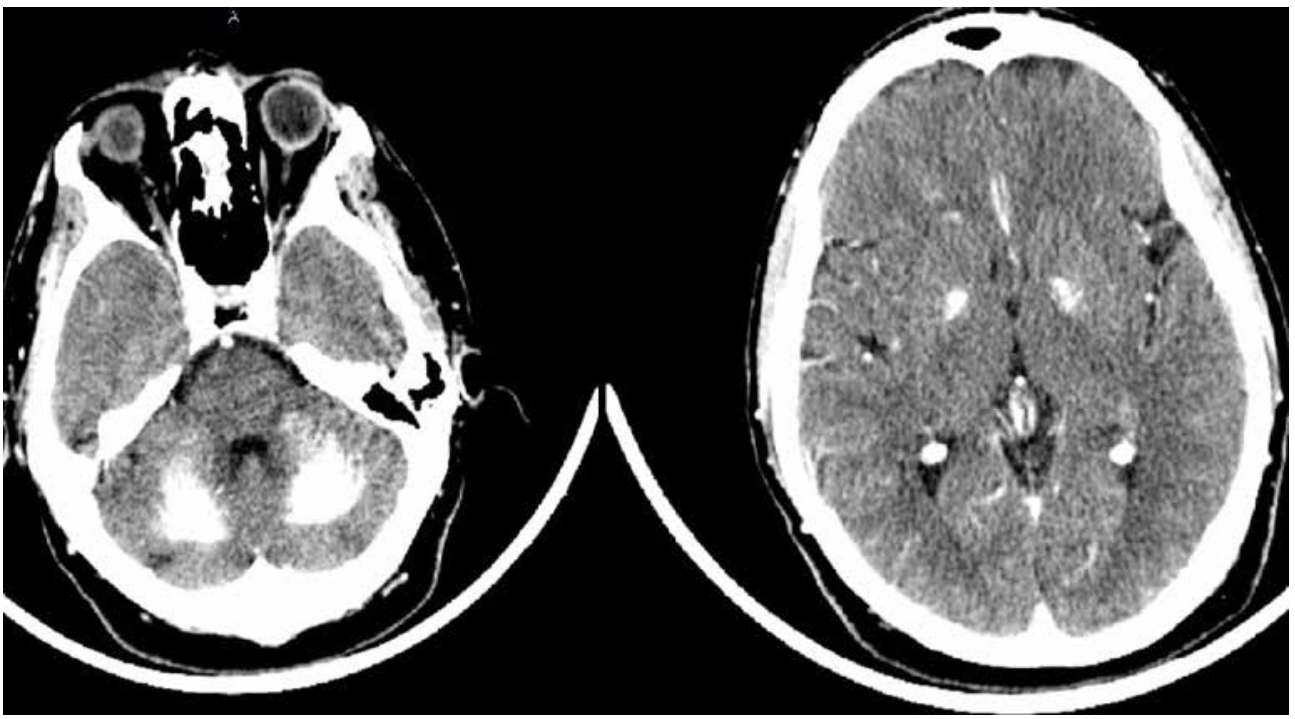

Figure 2. Brain computerized tomography: Bilateral calcification of basal ganglia and cerebellum.

DISCUSSION

In the literature, varying manifestations of Fahr's syndrome are described as memory disturbance, hallucination, delusions, personality change, and depression [2]; motor and phonic tics, stereotyped behaviors [3]; and extrapyramidal signs, such as Parkinsonism and paroxysmal nonkinesigenic dyskinesia [4]. Hoque et al. [5] described a case of Fahr's disease that presented with complex partial seizure and behavioral abnormalities. Several families with basal ganglia calcification, representing a heterogeneous group of disorders with variable inheritance, have been described [6].

Fahr's syndrome is typically inherited. In Case 1, the cause of the intracranial calcification is abnormal calcium metabolism due to iatrogenic hypoparathyroidism. This case is a typical example of secondary Fahr's syndrome. However, no phosphorus or calcium metabolism disorder, with the exception of decreased vitamin D level, was found in Case 2. Since just decreased vitamin D level could not be the cause of intracerebral calcification, Case 2 was diagnosed as primary Fahr's syndrome. It was notable that no family history of neurological disease or psychiatric, demential or extrapiramidal signs were found in neurological examination of Case 2. It is possible that others with Fahr's syndrome in this patient's family are asymptomatic.

The cause of seizure in Case 1 seems to be hypocalcaemia due to iatrogenic hypoparathyroidism. In addition, another theory of pathogenesis in these patients may be a dysfunction of cortico-basal connections and their interhemispheric relationship. In Case 2, no pathological cause was found for acute symptomatic seizure.

The term "Fahr's disease" has been used to describe a characteristic pathological pattern of nonarteriosclerotic vascular calcification of the striopallidodentate system bilaterally, with variable deposition of ferro-calcareous concretions in cortical sulci, thalamus, cerebral white matter, and cerebellum [7]. It is a misnomer, as Fahr's original case was a rare example of hypoparathyroidism associated with calcification in the media of larger vessels in the cerebral white matter, without basal ganglia calcification. Basal ganglia calcification may be categorized as idiopathic versus symptomatic (especially of parathyroid insufficiency), or sporadic versus familial. Prevalence of certain common neurological disorders, including dementia, stroke, and epilepsy, is similar in patients with incidentally discovered basal ganglia calcification and in age-matched controls [8]. However, the relative prevalence of extrapyramidal disorders is contentious. Forty-two patients with incidentally discovered radiological basal ganglia calcification in the series of Harrington et al. [9], and 33 cases reported by Vles et al. [10], had no clinical evidence of basal ganglia disorder. Murphy [11] found basal ganglia calcification in 53 of 7081 consecutive CT scans. In patients over 50 years of age, it was associated with clinical signs of basal ganglia dysfunction (Parkinsonism) in only 3 patients. In a series of 42 cases of basal ganglia calcification revealed on CT (performed for "various reasons"), Puvanendran et al. [12] found a single patient with Parkinsonism (right-sided tremor and rigidity) associated with dementia. Most of reported families with basal ganglia calcification do not display clinical evidence of basal ganglia disease. The clinical feature of epileptic seizure in present cases was not typical for basal ganglia calcifications, whereas radiological appearances were those of Fahr's syndrome.

In conclusion, Fahr's syndrome clinical manifestations can vary. It may begin at different ages and have a variety of presentations. The present cases are important because it would appear that there is no case in the literature of Fahr's disease presenting with generalized tonic clonic seizure. The seizures in such patients may be due to calcium metabolism abnormities and/or dysfunction of cortico-basal connections and their interhemispheric relationship.

Conflict of Interest: No conflict of interest was declared by the authors.

Financial Disclosure: The authors declared that this study has received no financial support.

Authorship contributions: Data collection and/or processing - N.O.; Interpretation - E.D., C.E.; Literature search - N.O.; Writing - N.O.; Critical review - C.E.

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