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CASE REPORT

Syncope as a presentation of Fahr's disease

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ABSTRACT

Fahr's Disease is characterized by neurologic, psychiatric, and cognitive disorders of unknown etiology, and is also characterized by symmetric calcifications in the basal ganglia and cerebellar dentate nucleus.The most common symptom of the disease is considered to be movement disorders, both Parkinsonism and hyperkinetic movement disorders (chorea, tremor, dystonia, athetosis, oro-facial dyskinesia). The second most common mani-festation of BSPDC is cognitive impairment followed by cerebellar impairment and speech disorder. Psychiatric features (dementia, psychosis, affective disturbances), gait disorders, and sensory changes are also reported. A 32-year-old male patient presented to Emergency department due to syncope. Coincidentally, hypocalcemia and widespread calcifications in brain computerized tomography indicating Fahr's Disease were determined. Patients with Fahr's disease generally presents to Emergency departments with epileptic seizures. Syncope as a presentation of the disease is extremely rare. In patients with hypocalcemia and calcifications in brain computerized tomography, Fahr's Disease must be suspected.

Key words: magnetic resonance imaging, computerized tomography, hypocalcemia, calcification, emergency department, Fahr, syncope

INTRODUCTION

Fahr's disease (FD), also called bilateral striatopallidodentate calcinosis (BSPDC), was first defined by German neurologist Karl Theodor Fahr in 1930. It is characterized by neurologic, psychiatric, and cognitive disorders of unknown etiology, and is also characterized by symmetric calcifications in the basal ganglia and cerebellar dentate nucleus, in individuals aged 5- 65 years.[1–3]

Both familial and sporadic cases have been previously reported in the literature. A locus on chromosome 14q has been identified as associated with the disease.[4] The disease has a wide range of clinical presentations, predominantly with neuropsychiatric features and movement disorders. Psychiatric features reported in the literature include: cognitive impairment, depression, hallucinations, delusions, manic symptoms, anxiety, schizophrenia-like psychosis, and personality change. Other clinical features include: Parkinsonism, ataxia, headache, seizures, vertigo, stroke-like events, orthostatic hypotension, tremor, dysarthria, and paresis.[5] Correlated with the literature, our case had a history of epilepsy.

The calcifications of FD may be detected by CT in the dentate nuclei of the cerebellum, basal ganglia, and white matter of the cerebral hemispheres.[6]

In this report, we present you a 32-year-old male patient who subsequently diagnosed as FD presented to Emergency department (ED) with syncope.

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CASE REPORT

A 32-year-old male patient was referred to our ED due to head trauma after syncope. The witnesses stated that he experienced loss of consciousness for 10 seconds after fainting. On his medical history, it was reported that he suffered hypoclacemia due to pseudohypoparathyrodism for 5 years and epilepsy for 10 years. His relatives stated that the latter was not a epileptic seizure but more likely to look like a fainting. On initial examination, the patient was conscious and his Glasgow coma scale was 15. His vitals were normal. He had a mild swelling on the temporal region of his head. Blood tests (complete blood count and routine biochemistry) were obtained and the patient has undergone computerized tomography (CT) scanning. His blood test revealed hypocalcemia (Ca: 4,59 mg/dl). Other results were found to be normal. On his CT scanning; on the level basal ganglia, particularly lentiform nucleus, caudate nucleus and thalamus, hyperdense lesions correlated with calcifications were observed. Also an MR imaging was performed and in T1-weighted images hyperintense lesions in the same localizations were observed (figure 1 and 2).

In the ED, patient was administered Calcium gluco-

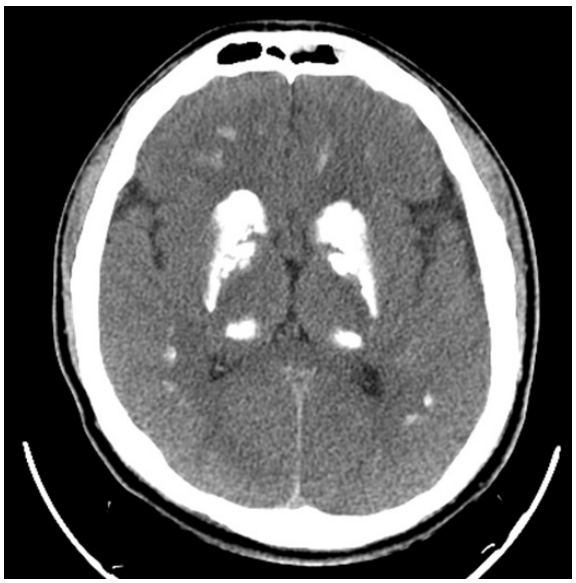

Figure 1, Brain compurterized tomography image of the patient. Bilateral calcifications in the basal ganglia.

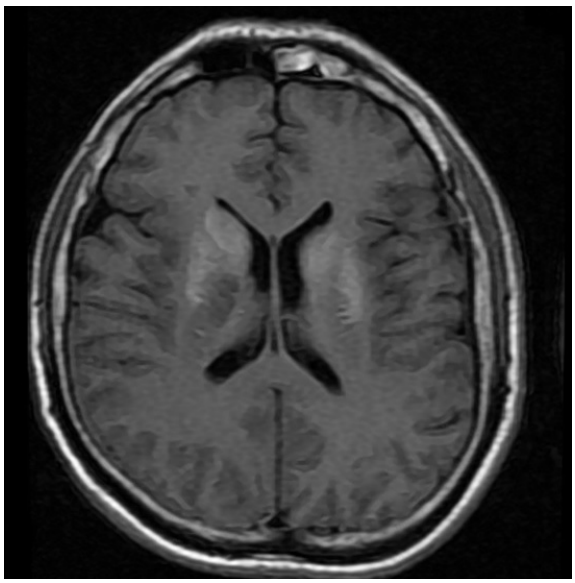

Figure 2, T1-weighted magnetic resonance image of the patient. Hyperintense densities correlated with calcification are visible.

nate in 5%dextrose and consulted with neurology and endcrinology. Later, patient was hospitalized in the endocrinology service with FD diagnosis. Written consent was obtained from one of patient's relatives.

DISCUSSION

When Fahr's disease was first identified by a German neurologist, Karl Theodor Fahr, in 1930, it was reported as a disease characterized by dementia and hypothyroidism, immobility without paralysis, and the calcifications of the basal ganglia.[1] Later in 1986, crtieria of the disease was defined in details by Lowenthal. According to Laowenthal, calcifications should have a characteristic distribution or inquire at least globus pallidus, with or without cerebellar calcification, the calcifications should be obvious on the computed tomography, the calcifications should be large enough to be detected at macroscopic examination.[7]

Lately, it was reported that calcifications in FD might also occur in other brain regions such as dentate nucleus, thalamus, and cerebral cortex. In genetical studies, both familial and non-familial cases of FD have been reported, predominantly with autosomal-dominant fashion.[5]

Clinical symptoms of FD are found in the literature as case reports, because the disease is very rare. The most common symptom of FD is considered to be movement disorders, both Parkinsonism and hyperkinetic movement disorders (chorea, tremor, dystonia, athetosis, oro-facial dyskinesia). The second most common manifestation of FD is cognitive impairment followed by cerebellar impairment and speech disorder. Psychiatric findings (dementia, psychosis, affective disturbances), gait disorders, and sensory changes are also reported. Neurological disorders such as extrapyramidal symptoms, pyramidal symptoms, epileptic seizures, choreoathetosis, and glossopharyngeal neuralgia may also be associated with FD. Although rare, there are reports in the literature that some patients may present with syncope.[3] In our case, unique symptom for ED admission was syncope. And after imaging and blood tests, the disease was diagnosed coincidentally.

Parathyroid diseases are one of the most common causes of FD. One of the commonest causes is pseudohypoparathyroidism which is due to resistance to the action of parathyroid hormone (PTH) causing hypocalcaemia and a high level of PTH.[8]

Also, our laboratory findings revealed hypocalcemia, which supported the diagnosis of FD.

Diagnosis of the disease is based on finding pathognomonic basal ganglia calcifications in computed tomography or magnetic resonance imaging.[9] In our case the diagnosis was based mainly on CT images and an MR imaging was performed for confirmation.

CONCLUSION

Emergency physicians must be aware of FD in patients with a history of neurological and/or psychiatric diseases who has hypocalcemia and widespread calcifications in brain CT or MR imaging.

CONFLICT OF INTEREST

None.

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