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An unexpected diagnosis in early pediatric age group: Fahr syndrome manifested with epileptic seizure

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ABSTRACT

Intracerebral calcifications are one of the essential parameters in the diagnosis of some neurological diseases. Fahr group is a rare pathologic entity presenting with basal ganglion calcifications. Symmetrical basal ganglion calcifications and an aetiological reason (metabolic, hormonal, or infectious) constitute the main picture of Fahr syndrome. This syndrome usually begins at the third and fourth decade and is often with accompanying neuropsychiatric symptoms. We presented the case of a 7 years old child who was admitted to the hospital with an atypical tonic-clonic epileptic seizure. Bilateral basal ganglion calcifications were detected at the level of globus pallidus. Clinical and laboratory findings were compatible with secondary hyperparathyroidism.

Keywords: Fahr syndrome, hyperparathyroidism, basal ganglion calcification

INTRODUCTION

The symmetrical calcifications of basal ganglia were first time described by Bamberger in 1855. In 1930, Fahr explained the association between progressive neurological symptoms and idiopathic calcifications. Nowadays, the primary form is defined as Fahr's disease, whereas the seconder type is defined as Fahr syndrome. Fahr syndrome occurs secondary to calcium metabolism disorders, endocrinopathies, infections and some storage diseases. The findings start at the third and fourth decades. Fahr syndrome is rarely seen in the early pediatric age group (1,2). We presented the case of a 7 years old child with Fahr syndrome accompanied by clinical and radiological findings.

CASE PRESENTATION

A 7 years old boy presented to our hospital emergency department with a history of seizure. Family of the patient reported that the seizure was started with a scream. Respectively, muscle rigidity, twitching, shortness of breath were added. The anamnesis was compatible with the tonic-clonic epileptic seizure.

In the history of the patient; the patient was the product of normal pregnancy. The baby was born at 39 weeks with a weight of 3000 grams. Family history was normal.

In the clinic exam, there were no neurological deficit or additional finding. However, there was evidence of infection. Tonsillar hyperaemia, bilaterally rhonchus and rales resulting from acute tonsillitis and pneumonia were observed. It was thought that the infection might have triggered the epileptic attack. The cardiac examination and the rest of the physical examination were normal.

Under these circumstances, Cranial MRI and CT were requested. Cranial CT showed symmetrical calcifications in the bilateral globus pallidus. Other basal ganglions were normal (Figure 1 a,b,c). Laterally ventricles were symmetrical. Parenchymal anomaly and mass were not observed. Cerebellum and brain stem were normal. The MRI was requested for advanced radiological assessment. The high signal areas caused by mineral deposition were

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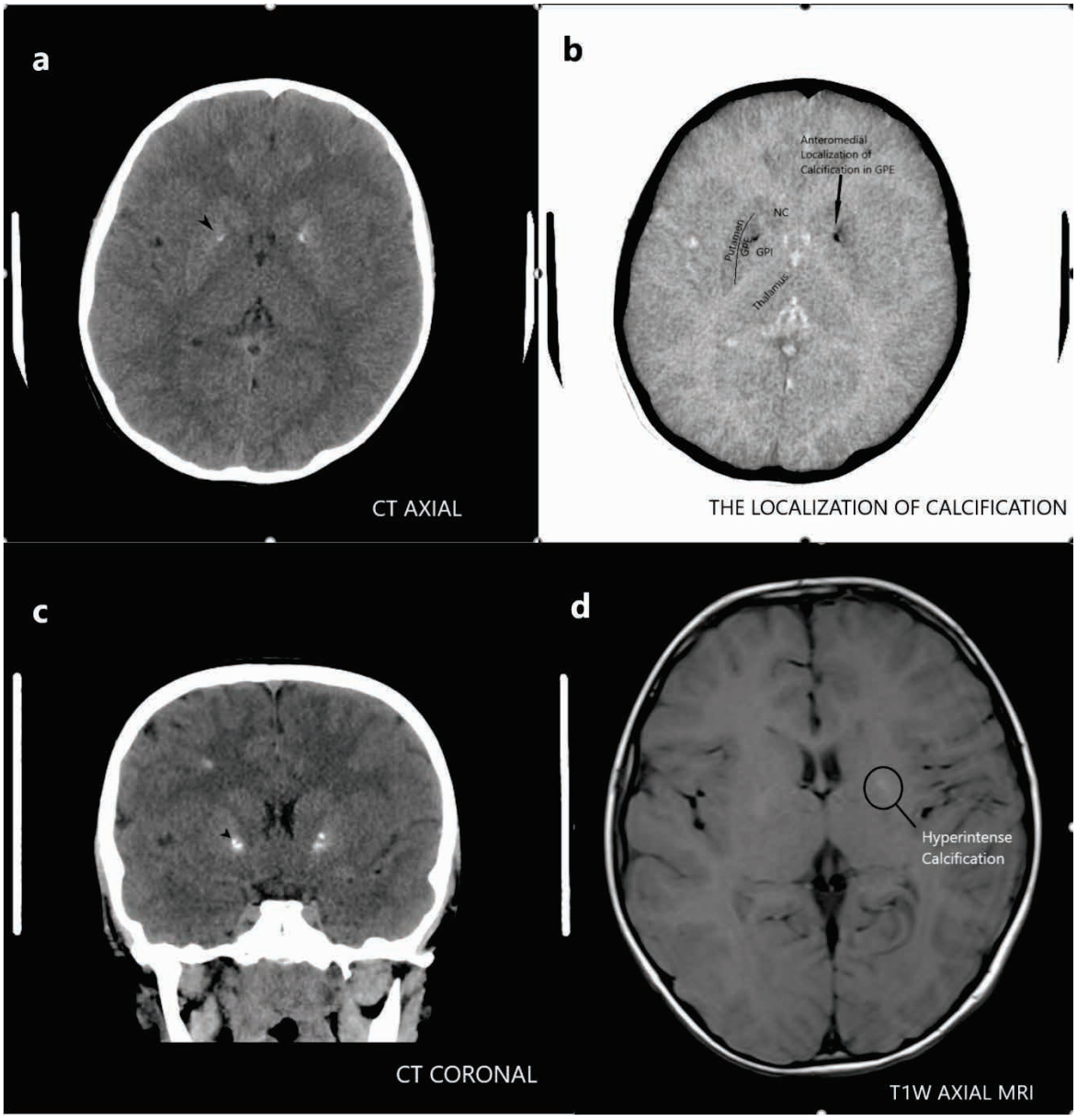

FIGURE 1 a,b,c,d. a. Basal ganglion calcification (arrowhead) in axial CT b. The demonstration of anteromedial localization of calcification in globus pallidus externus (GPE) other abbreviations:
NC: Nucleus caudatus GPI: Globus pallidus internus c. Basal ganglion calcification (arrowhead) in coronal view d. Hyperintense basal ganglion calcifications in axial T1 weighted MRI

seen on T1-weighted images on MRI. No additional findings in conventional MRI and cranial MR angiography (Figure 1d). The patient was diagnosed as Fahr syndrome on the base of clinical, laboratory, and radiological findings.

Basal ganglia calcifications and absence a history of the family directed us to further laboratory tests. Therefore, metabolic and hormonal tests were requested. Laboratory tests showed a hypocalcaemia of 5.19 mg/dl (normal range: 8.6 to 10.3 mg/dl), a low phosphate level of 2.84 mg/dl (3.4 to 4.5 mg/dl) and an elevated level of parathyroid hormone (PTH) of 278 pg/ml (10-55 pg/ml), and low vitamin D of 24 ng/ml (30 and 50 ng/ml). The hormone profile was compatible with secondary hyperparathyroidism (Table 1).

TABLE 1. Hormone and electrolyte profiles in primary, secondary and tertiary hyperparathyroidism

HPT	Calcium	Phosphate	Vitamin D	PTH
Primary	↑	↑ or N	↑	↑ or N
Secondary	↓ or N	↑ or ↓	↓	↑
Tertiary	↑	↑	↓	↑

HPT – hyperparathyroidism; PTH – parathormone

Other blood electrolytes; complete blood count; liver, kidney, and thyroid function tests were normal. There was no evidence of parathyroid adeno-

ma in the ultrasound. The patient's relative refused advanced evaluation in our hospital.

DISCUSSION

Fahr disease is a neuropsychiatric disorder associated with calcium deposits in bilateral basal ganglions (1,2). The primary type is called familial cerebral ferro calcinosis, primary familial brain calcification, or Fahr disease. It is an autosomal dominant disease, and the age of onset is between 40 and 60 years old. In the secondary type, metabolic, infectious or other underlying factors should be considered. In the second type called Fahr syndrome, metabolic, infectious or other underlying factors should be considered. The age of onset is generally earlier, between 30 and 40 years old (2). Fahr syndrome is two times more common in male to female (3). Our patient was a boy. Fahr syndrome related to hypocalcaemia is not common in pediatric ages (4).

Hypocalcaemia is characterized by neuromuscular symptoms like tetany, paraesthesia, seizures, calcium accumulation in soft tissues (cataract and basal ganglion calcification, etc.) (5). It may present with an organic brain syndrome. Movement disorder is the most common symptom. Manyam et al. reported that 67 of 99 Fahr patients were symptomatic while 32 were asymptomatic (6). 55% of symptomatic cases have also been diagnosed with movement disorders. 57% of the patients with movement disorder had parkinsonism, 19% Korea, 8% tremor, 8% dystonia, and 5% athetosis. Other neurological findings include cognitive impairment, speech disorders, gait disturbance, psychiatric changes, emotional lability, pain, cerebellar and pyramidal symptoms (6).

Fahr syndrome is usually associated with an inhibition of mental development in the early paediatric age group. Mental retardation or behavioural disorder was not detected in our patient. Cognitive abilities were normal. Atypically, the first clinical presentation was an epileptic tonic-clonic seizure.

Disorders of calcium and phosphate metabolism are mainly responsible for intracranial calcification. Hypoparathyroidism is the most common metabolic disorder associated with Fahr syndrome, whereas hyperparathyroidism is rare (7). In our case, the results of the analysis were in favour of secondary hyperparathyroidism.

Radiologically, calcifications are easily recognized with CT. MRI is sensitive to developmental anomalies and neurological pathologies, such as intracranial masses, dystrophy, and demyelinating disease. However, it is not a useful imaging method for the detection of calcification. Although the signal is variable in T1 and T2-weighted images, calcified regions are more visible on T1-weighted images. Calcium deposits may contain proteins that bind to polysaccharides during the development of the pathological process. This protein effect can explain the high signal on T1-weighted images (8). In our case, basal ganglion calcifications were observed on CT. T1 signal was high on MRI.

In rare cases, basal ganglion calcifications may be confused with haemorrhagia. Three points are important here to separate the two entities. The first is the symmetry. Basal ganglion calcifications are seen symmetrically. The second is the anatomical localization. Deposition in the basal ganglia affects the globus pallidum part of the lentiform nucleus at an early stage of Fahr disease and syndrome, as in our case. The third is the HU value. HU value of bleeding generally does not exceed +100, while HU calcifications are not below this value (9).

We searched in PubMed with the following keywords, 'Fahr syndrome' OR 'Fahr' OR 'basal ganglion calcification', and found eleven papers published on this topic. One of these papers was the review article, and nine of them were the case reports. The range of the ages was altered from 41 to 68 years. In a case report, Fahr syndrome was secondary to pseudo pseudohypoparathyroidism. The others were regarding hypoparathyroidism. Two cases were manifested with neuropsychiatric findings. One patient is present with status epilepticus, similar to our article, but the patient is older in the age group (10). The only case with Fahr syndrome in the early paediatric age group published during this year was a case due to spherocytosis (11). Our case was unique with age group, clinical presentation and presence of secondary hyperparathyroidism.

CONCLUSIONS

There are few remarkable points of our case report. The first is that Fahr syndrome is seen in the early pediatric period. The second is the presence of hyperparathyroidism. Third one; it is an undiagnosed and untreated case presented with an epileptic seizure. Overall, Fahr syndrome is a rare disease that can be suspected radiologically. Our case was one of the first Fahr syndromes resulting from hyperparathyroidism in his age group.

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