Fahr's Syndrome – A Case Report

Síndrome de Fahr: un caso clínico

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ABSTRACT

Fahr's syndrome is an uncommon neurodegenerative disorder, characterized by bilateral and extensive deposition of calcium in the basal ganglia.
We present the case of a 66-year-old female presented to the Emergency Department with (a sudden and) intense holocranial headache accompanied by nausea. She also reported paresthesias of the hands and feet with several years of evolution, mnesic changes and periods of uninhibited behavior. Head CT revealed extensive and bilateral calcifications of the basal ganglia, cerebellum and frontal region and the analytical findings showed a severe hypocalcemia. She begun replacement with intravenous and oral calcium. An improvement of signs and symptoms were observed during the hospitalization.
Although rare, Fahr's syndrome must be kept in mind and appropriate treatment should be aplied in order to stop it's progression and improve the clinic symptoms and signs.

Keywords: Fahr's syndrome; basal ganglia; hypoparathyroidism; hypocalcemia; neuropsychiatric.

INTRODUCTION

Fahr's syndrome is a rare and chronic disease characterized by bilateral and extensive deposition of calcium in the basal ganglia, thalamus, cerebral cortex, cerebellum and hippocampus[1,2,3]. The most common presenting signs and symptoms are neuropsychiatric manifestations like seizures, cognitive impairment and can also be associated with parestesias due to hypocalcemia. Fahr's syndrome usually appear in the fourth or fifth decade of life[2,3].
The most common metabolic cause of Fahr's syndrome is hypoparathyroidism[4,5].
Hypoparathyroidism is an endocrine disease, that can be idiopathic or secondary. Idiopathic form is characterized by deficient secretion of parathyroid hormone (PTH) without a definitive cause.

CASE REPORT

We report a case of a 66-year-old woman with history of hypertension and dyslipidemia and with no significant family history, presented to the Emergency Department (ED) with (a sudden and) intense holocranial headache accompanied by nausea. She also reported paresthesias of the hands and feet with several years of evolution, mnesic changes and periods of uninhibited behavior. In the ED, the patient had a generalized tonic-clonic seizure, that ceased after the administration of 5 mg of diazepam. She was afebrile and euglycemic. No nuchal rigidity or focal neurologic signs were observed.
Head CT revealed extensive and bilateral calcifications of the basal ganglia, cerebellum and frontal region (Figure 1). Analytical findings showed a severe hypocalcemia of 5.7 mg/dL (N: 8.6 –

Figure 1

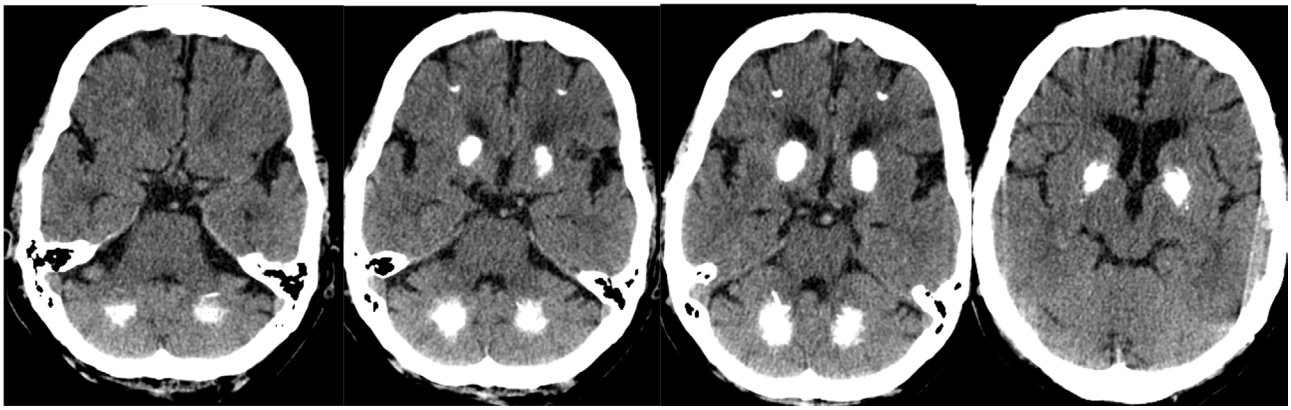

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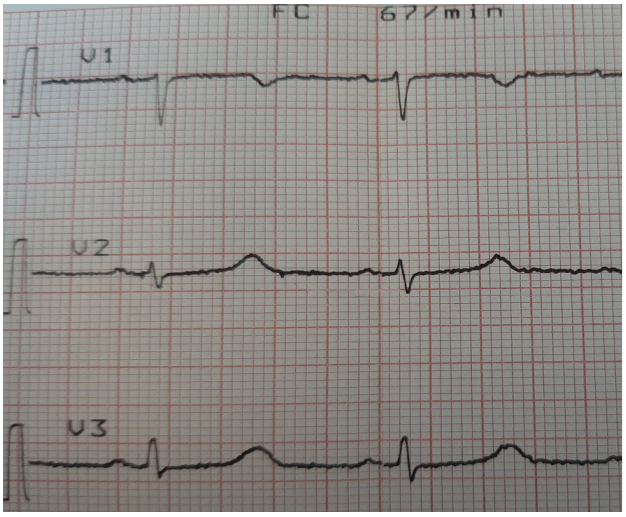
Figure 2



10.3 mg/dL) with an ionized calcium level of 0.60 mmol/L (N: 1.13 – 1.31 mmol/L). Other analytical findings showed a PTH of 11.08 pg/mL (N: 15-65 pg/mL), phosphorus of 6.4 mg/dL (N: 2,5 – 4,9 mg/dL), magnesium of 1,26 mg/dL (N: 1,6 – 2.6 mg/dL) and a vitamin D of 30 ng/mL (N: 6,2 – 53,2 ng/mL). An electrocardiogram was performed and revealed a prolongation of the QT interval (Figure 2).

The electroencephalogram confirmed the diagnosis of epilepsy. Thyroid and parathyroid ultrasonography were normal and parathyroid antibodies were also negative.

Based in all these findings the diagnosis of Fahr's syndrome due idiopathic primary hypoparathyroidism was made.

The patient began immediate replacement with intravenous calcium gluconate, until there was normalization of the electrocardiographic changes, followed by oral supplemental calcium and calcitriol. She also started replacement for the hypomagnesemia with intravenous magnesium until the levels were normalized.

Levetiracetam 250 mg twice daily was prescribed to prevent any seizures.

During hospitalization, the symptoms of the patient improved as the levels of calcium were normalized. She remained stable, without any more seizures.

The patient was discharged and referred to an Internal Medicine and Endocrinology consult.

DISCUSSION

Fahr's syndrome is due to calcification of the basal ganglia and other regions of the brain, that are bilateral and symmetric[1,2,4,6]. The most common reported metabolic disorder are hypoparathiroidism and pseudo-parathiroidism.

Hypoparathyroidism is an endocrine disorder that can be iatrogenic, due the surgical removal or radiotherapy, or idiopathic. The idiopathic form is uncommon and of unknown etiology[4,6]. It refers to the deficient PTH secretion, with the presentation of levels of PTH and calcium that are low. In pseudo-parathiroidism PTH levels will be high with hypocalcemia.

There is no clear explanation for the mechanism that cause these calcifications. It is suggested that the increased calcium-phosphorus complex plays an important role 6, by deposition in the vessel wall and eventually extending to the neuron.

The most common clinical manifestations of this syndrome are neuropsychiatric like seizures, headache, cognitive decline and parkinsonism[1,2].

The alterations found in the electrocardiogram are a well-known impact of the low levels of the calcium.

To the date there's is no specific treatment for Fahr's syndrome[2,4,6]. The management of the clinical manifestations is limited to supportive care, using anxiolytics, anticonvulsant and a hemodynamic balance. If the Fahr' syndrome is due to the hypoparathyroidism, like in our case, the symptoms usually improve with the normalization of the calcium and phosphorus levels.

The patient was treated with IV and oral calcium and anti-convulsant, but no other medications were needed once that the neuropsyquiatric symptoms resolved after the normalization of the plasmatic calcium level. She maintained treatment with oral calcium and calcitriol to prevent symptomatic hypocalcemia.

CONCLUSION

Fahr's syndrome, although rare, should incorporate the list of differential diagnoses of neuropsychiatric disorders and epileptic seizures. The calcifications of the basal ganglia, despite being a suggestive finding of an evolved disease, may be a finding of enormous value since they can immediately guide the diagnosis and allow rapid treatment of hypocalcemia and hypoparathyroidism.

CONFLICTS OF INTEREST

The authors declare that there is no potential conflict of interest relevant to this article.

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AUTHORS' CONTRIBUTION

Costa, Raquel and Mendes, Tiago wrote de paper; Fontes, Joana, Sousa, Barbara and Silva, Joana reviewed the paper.

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