INTRODUCTION

Fahr’s syndrome is a rare condition characterized by calcifications of basal ganglia and cerebral cortex, with possible relation to metabolic disorders. We present a case of a patient diagnosed with Fahr’s syndrome after experiencing episodes of involuntary movements of the limbs, cognitive impairment, hallucinations, and abducens nerve palsy for months.

Fahr’s syndrome is a rare neurological disorder characterized by abnormal calcified deposits in basal ganglia and cerebral cortex, commonly affects young-to-middle-aged adults and clinical manifestations incorporate a wide variety of symptoms, ranging from neurological symptoms of extrapyramidal system to neuropsychiatric abnormalities of memory and concentration to movement disorders, that preferentially occur in patients with parathyroid disorders, especially hypoparathyroidism.

The examination of choice for diagnostic confirmation is computed tomography (CT) that enables localizing and assessing the extent of cerebral calcifications.

The prognosis is variable and hard to predict, since there is no reliable correlation between calcium deposits extension in the brain and neurological deficit. It is a progressive and degenerative disease and its evolution depends on the identification and treatment of the underlying pathology.

CASE REPORT/CASE PRESENTATION

An 52-year-old woman was admitted at the Emergency Department with sudden onset of generalized tonic-clonic seizure, treated first with intravenous diazepam and then started enteral levetiracetam, with good response.

Regarding her past medical history, the patient had diabetes mellitus, diagnosed around 18 years ago, with bad glycemia control which complicated with diabetic retinopathy (stage 3), essential hypertension, dyslipidemia, obesity, and heavy alcohol consumption (60 g/d) during the last five years. Tobacco use or illicit drugs consumption was denied.

According to her family, during these past 18 months, the patient had experienced multiple self-limited episodes of involuntary movements of the limbs, fast and progressive cognitive impairment, and audio-visual hallucinations with psychomotor agitation, for which she was evaluated by the family physician, who interpreted it as a case of psychiatric...
illness underlying alcohol abuse and medicated the patient accordingly. Her family history of neurodegenerative or movement disorders was negative.

Her physical examination revealed poor general condition, pallor, hydrated, afebrile, anicteric, and acyanotic. Her blood pressure was 117/69 mm Hg, and her heart rate was 65 beats per min. After the patient regained consciousness, a neurological examination was performed and abnormal neurological signs including discreet dysarthria, sixth nerve palsy of the right eye (which had been reported already five months ago during an observation by the Ophthalmologist) and generalized neuromuscular irritability such as muscle cramps and tetany were found. Chvostek's sign and Trousseau's sign were positive.

Laboratory studies including serum calcium 4.9 mg/dL (normal range 8.4-10.6 mg/dL), phosphate 5.8 mg/dL (normal range 2.3-4.7 mg/dL) and parathormone level 6 pg/mL (normal range 15-65 pg/mL) were compatible with idiopathic hypoparathyroidism. Thyroid function was normal, thyroid stimulating hormone (TSH) 2.39 mg/dL (normal range 0.358-3.74), free thyroxine level (T4) 1.33 mg/dL (normal range 0.76-1.46 mg/dL), TSH receptor antibodies and thyroglobulin antibodies were at normal range, but anti-thyroid peroxidase (anti-TPO) antibodies were increased, 195.9 (normal range <34).

The remaining results as hemogram, sedimentation rate, iron, ferritin, magnesium, potassium, rheumatoid factor, antinuclear antibodies, thyroid function, ceruloplasmin, VDRL, HIV, hepatitis C and B virus, serum copper, CPK, lactate, hepatic and renal function tests, vitamin B12, vitamin D, folic acid, pituitary hormones, and protein electrophoresis were unremarkable. Eletrocardiogram and eletroencephalogram showed no alterations. Cranial CT scan showed extensive symmetrical intracerebral calcifications in both basal ganglia and radiated crown (shown in Figure 1). Thyroid ultrasound was normal.

Initially, the patient received treatment with intravenous calcium gluconate and then continued to take oral calcium carbonate and calcitriol, that maintained after hospital discharge. She became seizure free, nevertheless there was no cognitive improvement.

3 | DISCUSSION/CONCLUSION

Fahr's syndrome is a rare neurological condition, which prevalence is unknown, and preferably affects middle-aged people. It is reported an incidence of basal ganglia calcifications ranging from 0.24% to 2% in radiological studies, and most of the literature on Fahr's disease is based on case reports.1,2

It differs from Fahr's disease in terms of etiology, prognosis, and treatment because Fahr's syndrome is associated with secondary causes that may be potentially treated, and the first is a genetic condition, dividing into hereditary or sporadic cases.2 The most typical association corresponds to endocrine diseases, where calcium and phosphorus metabolism disorders predispose the precipitation of calcified deposits, such as parathyroid dysfunction (mostly hypoparathyroidism) that is the most common cause of Fahr's syndrome.3,4

Despite of a significant number of patients remaining asymptomatic (approximately 20%) until accidental diagnosis through neuroimaging, the clinical manifestation may be a varied spectrum of neuropsychiatric symptoms including movement disorders, seizures, speech disorders, spasticity, psychosis, cognitive impairment, and syncope.4,5

The preferred method for localizing and assessing the extent of cerebral calcification is the cerebral CT scan, although magnetic resonance imaging may have diagnostic value in some cases. Often the most affected areas are lenticular nucleus, putamen, thalamus, caudate, and dentate nuclei. Cerebellum, cerebral cortex, hippocampus, and subcortical white matter may also be affected. Calcification areas are easily identified as hyperdense lesions on CT, which are typically bilateral and symmetrical and show signs of having progressive and gradual establishment.6-8

Multiple clinical conditions are associated with intracranial calcifications, including infectious diseases (acquired immune deficiency syndrome, toxoplasmosis, cytomegalovirus, herpes simplex, brucellosis), genetic conditions (Fahr's disease, pseudohypothyroidism), vascular, toxic exposure (lead poisoning, hypervitaminosis D), autoimmune disease (systemic lupus), and endocrine disorders (calcium/phosphorus abnormalities).9,10 Hypoparathyroidism is a common cause of basal ganglia calcification, and its etiology can be
idiopathic (uncommon condition characterized by the absence or atrophy of the parathyroid glands) or secondary (usually a complication of thyroid surgery).5,11,12

As regards the present case, the patient had no history of neck surgery, neither was there evidence of an infectious disease. It were not found any vitamin deficiencies or signs of malnutrition. No neoplasms were noticed. Liver and kidney function, as well as hormone assays, had no alterations. Nevertheless, our patient indeed had diabetes mellitus with age onset in her early thirties, which initially had good control of hyperglycemia with oral antidiabetic agents, but within months needed to initiate treatment with insulin, suggesting that it can be a latent autoimmune diabetes in adult (LADA), and still, the fact that anti-TPO antibodies are increased that may indicate an early stage of a possible autoimmune thyroiditis, what could lead us to think we are facing an autoimmune polyglandular syndrome.13,14 Due to various limitations, genetic factors could not be tested. Consequently, the etiology of hypoparathyroidism remains undetermined.

There is no specific treatment, besides correction of possible metabolic disturbances (calcium, vitamin D, or 1, 25-dihydroxyvitamin D supplementation) and supportive care for neurological and psychiatric symptoms, in order to improve the presenting symptoms and prognosis. Subcutaneous synthetic PTH could be used in refractory HPT.15

To conclude, all patients with long-term recurrent tetany, epileptic-like and/or psychiatric symptoms, along with chronic cognitive impairment, clinicians should associate with the possibility of Fahr’s syndrome and have the patients undergo a neuroimaging test in order to make early diagnosis and try to identify potentially treatable secondary causes. Although hypoparathyroidism is one of the most frequent causes of calcifications on the basal ganglia, it is recommended to determine serum calcium, phosphorus, and PTH levels, since its correction can remarkably improve symptoms and be a prognosis modifier.

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CONFLICT OF INTEREST

The authors declare no conflicts of interest.

AUTHOR CONTRIBUTION

GP and AF: wrote the manuscript. LC, MG: responsible for scientific input and revision. JC: senior author and was involved in scientific input and revision of the final manuscript.

ETHICAL APPROVAL

Written informed consent was obtained from the patient for publication of this case report and accompanying images.

DATA AVAILABILITY STATEMENT

No data are associated with this article.

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