Síndrome de Fahr: A Demência como Apresentação para o Diagnóstico Imagiológico

Fahr's Syndrome: Dementia as a Feature for an Imaging Diagnosis

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Palavras-chave: Calcinose/diagnóstico por imagem; Demência/diagnóstico por imagem; Doenças Neurodegenerativas/diagnóstico por imagem.

Keywords: *Calcinosis/diagnostic imaging; Dementia/diagnostic imaging; Neurodegenerative Diseases/diagnostic imaging.*

A 79-year-old female presented to the emergency department (ED) with sudden loss of consciousness. The patient had a five-year medical history of progressive memory loss, with follow-up in a Neurology outpatient clinic with an initial Mini-Mental State Examination (MMSE) of 17/30 and neuropsychological tests with evidence of evolution of cognitive impairment from mild to severe in one year. Vitamin B12 was within normal range, Treponemal testing was negative, and the patient was not taking anticholinergic medication. The patient was started on donepezil 10 mg, with no therapeutic effect according to her career, and did not attend to the follow-up assessments and brain computed tomography (CT) scan. Medical history of hypothyroidism was noted, secondary to subtotal thyroidectomy, on levothyroxine replacement. The patient had no family history of neurological and endocrinological disorders.

On examination in the ED the patient presented Glasgow coma scale 6 (O2 M2 V2), with four limb hypotonia and bilateral extensor plantar response. During examination the patient presented a seizure, reversed with diazepam, and started levetiracetam. Blood tests showed evidence of hyperphosphatemia, decreased PTH, normal calcium levels (on oral supplementation) and normal thyroid function. Brain CT scan was performed, with evidence of calcified lesions in the region of the globus pallidus and dentate nuclei of the cerebellum (Fig. 1). Lumbar puncture was performed with no concerns. Heavy metals screening test was not conducted.

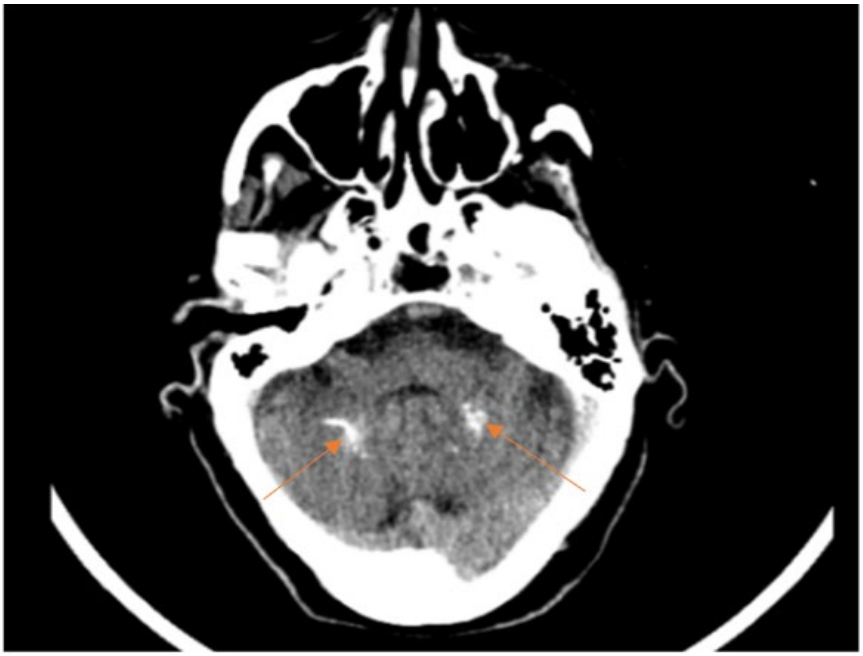

Figure 1: Image from brain CT scan, with calcified lesions in the dentate nuclei of the cerebellum.

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https://doi.org/10.60591/crspmi.119

Diagnosis of Fahr's syndrome was achieved with typical brain CT scan findings and the evidence of phosphocalcium disorder. During admission the patient developed a nosocomial pneumonia, and deceased before it was possible to perform a parathyroid scan.

Fahr's syndrome is a rare neurological disorder characterized by abnormal calcified deposits in basal ganglia and cerebral cortex, associated with phosphocalcium metabolism dysfunction.[1] Diagnosis consists of bilateral calcification of basal ganglia, progressive neurologic dysfunction, and absence of other causes for these features. This condition is as yet incurable, but management and treatment strategies mainly focus on symptomatic relief and eradication of causative factors.[2] Differential diagnosis must be made with infectious diseases (such as toxoplasmosis, cytomegalovirus and neurocysticercosis), vascular atherosclerosis and intracerebral hemorrhage, Fabry disease and neoplasms. ■

Declaração de Contribuição

AMV – Preparação do manuscrito
SR – Preparação da Imagem
TP, TSS – Revisão do manuscrito

Contributorship Statement

AMV - Preparation of the manuscript
SR - Image Preparation
TP, TSS - Revision of the manuscript

Responsabilidades Éticas

Conflitos de Interesse: Os autores declaram a inexistência de conflitos de interesse na realização do presente trabalho.
Fontes de Financiamento: Não existiram fontes externas de financiamento para a realização deste artigo.
Confidencialidade dos Dados: Os autores declaram ter seguido os protocolos da sua instituição acerca da publicação dos dados de doentes.
Consentimento: Consentimento do doente para publicação obtido.
Proveniência e Revisão por Pares: Não comissionado; revisão externa por pares.

Ethical Disclosures

Conflicts of interest: The authors have no conflicts of interest to declare.
Financing Support: This work has not received any contribution, grant or scholarship
Confidentiality of Data: The authors declare that they have followed the protocols of their work center on the publication of data from patients.
Patient Consent: Consent for publication was obtained.

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Recebido / Received: 2022/07/21
Aceite / Accepted: 2022/09/29
Publicado online / Published online: 2023/11/29

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