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Case report

Medical monitoring of patient with cavernous hemangioma of the retina and intracranial involvement

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ABSTRACT

Purpose: To describe a case report of Cavernous Hemangioma of the Retina (CHR) and highlight the importance of investigating intracranial system when retinal vascular alterations are present.
Observations: Patient of 26 years old, with right eye fundus alteration since 7 years old, no visual complaint. In the exam, there were sacular lesions with hematic content in the macula, configurating liquid level, associated with whitish lesion suggestive of fibrosis, compatible with cavernous hemangioma; best visual accuity of 20/20 both eyes. Comparing the current retinography with the previous one, there was no significant lesion progression of the exam. By neuroimaging investigation, the Magnetic Ressonance Imaging showed multiple cavernomas. As his sister had been diagnosed with cerebral cavernous malformation 3 years before, due to termic hiposensitivity on left leg investigation, familial multiple cavernous malformation syndrome was defined.
Conclusion: Imaging investigation of the Central Nervous System should be considered when CHR is observed. Clinical monitoring is necessary, interventional treatment is selected only for particular cases.

1. Introduction

Cavernous Hemangioma of the Retina (CHR) is a rare form of vascular hamartoma of the retina. In a review paper of 96 cases it was observed that 14% of the patients had intracranial alterations. The great majority of the patients were assymptomatic, but from those who had focal neurologic deficit, 80% had Central Nervous System (CNS) involvement, such as cavernous malformations.[1]

Cavernous malformations are characterized by the presence of cavernomas, and bleeding is its main factor for growth. It presents autosomal dominant inheritance and affects 0,4-0,8% of the population. From these, about 40% are assymptomatic.[2]

Five percent of patients with Familial Cerebral Cavernomas (FCC) have retinal cavernomas. These lesions are clinically asymptomatic. They can be associated with one of the 3 cerebral cavernous malformation genes (KRIT1, MGC4607, and PDCD10).[3]

The main goal of this paper is to document a case report of CHR, highlighting the importance of investigating intracranial system when retinal vascular alterations are presented.

2. Case report

Patient, male, 26 years old, physician, with no visual complaints, went to the office to undergo opthalmologic exam due to right eye (OD) fundus alteration since childhood. At 8 years old, had best corrected visual accuity (BCVA) of 20/20 partial and 20/20. Retinography, red free and angiofluoresceinography (AGF) were performed at the same period, and are illustrated in Figs. 1 and 2. Right eye presents macular sacular hematic lesions associated with fibrosis. Left eye with normal fundus. Past medical history: migraine with aura and episode of *meningitis retention syndrome* (meningitis with accute urinary retention)[4] at 22 years old, with discrete alterations in the CNS Magnetic Ressonance Imaging (MRI), but with no diagnostic conclusion.

In the current exam, presented BCVA of 20/20 in both eyes, reflexes, extrinsic ocular motility and slit lamp exam with no alterations, fundus OD (Fig. 3) shows macular sacular lesions with hematic content, configurating liquid level, associated with whitish lesion suggestive of fibrosis, compatible with cavernous hemangioma, fundus OS within normal limits. By comparing retinographies from 1999 to 2017, it is observed that there was unremarkable progression. Facing this scenary, it was requested a CNS MRI, which was described with multiple cavernomas (Fig. 4). As family history, his sister was diagnosed with multiple cavernomatosis 3 years ago, through imaging (Fig. 5) at 26 years old, after investigation of termic sensibility alteration on left leg. Presented cavernous hemangioma from previous foot nevus anatomopathological examination (Fig. 6). No ophthalmologic alterations.

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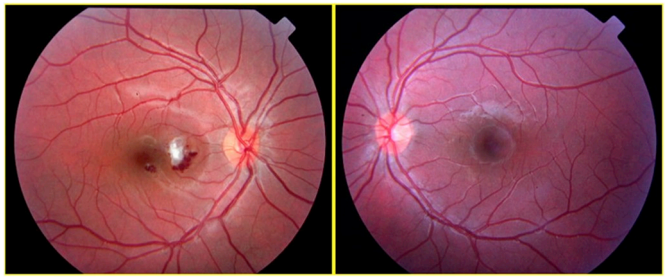

Fig. 1. Retinography (1999). Right eye: macular sacular hematic lesions associated with fibrosis. Left eye: normal fundus.

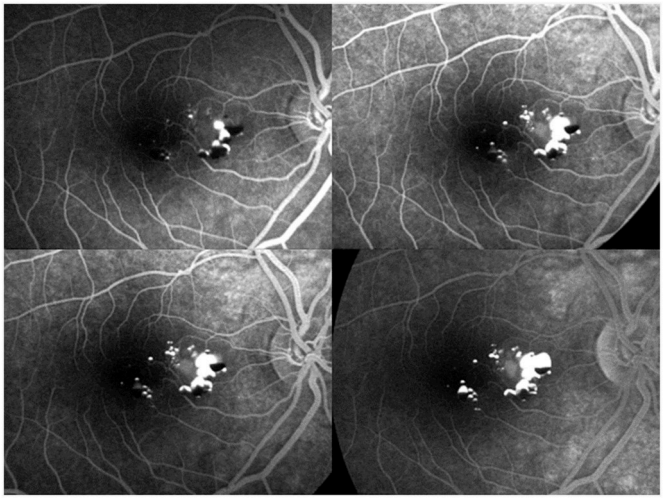

Fig. 2. Fluorescein angiography late phase (1999). Macular sacular lesions with hematic content, configurating liquid level.

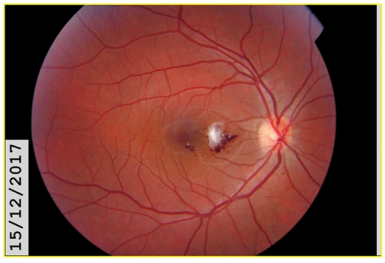

Fig. 3. Retinography (2017). Right eye: macular sacular hematic lesions associated with fibrosis.

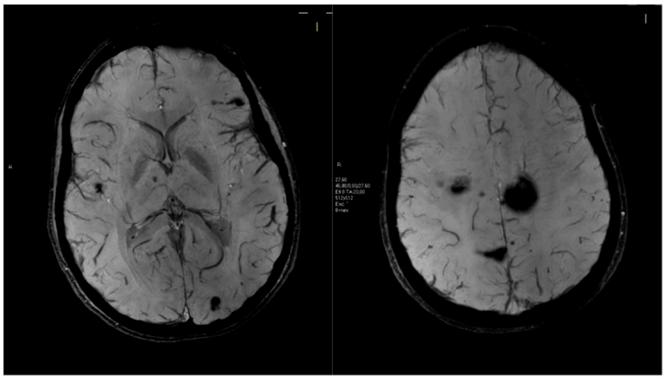

Fig. 5. Multiple cavernomas showing foci of marked hypointense signal on axial T2* weighted images.

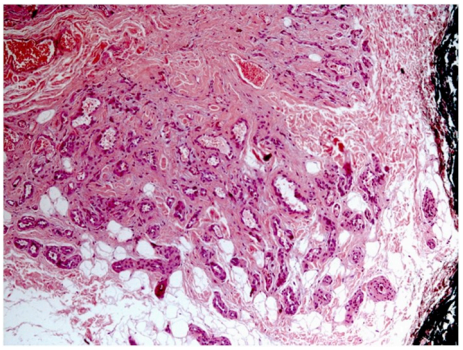

Fig. 6. Cutaneous hystologic exam, hematoxyline eosine, 100× objective. Benign proliferation of endothelial cells, forming blood vessels full of erythrocytes.

Therefore, it was confirmed the diagnosis of Familial Multiple Cavernous Malformation Syndrome (FMCMS).

3. Discussion

CHR is a rare disease, which was described in the present case report associated with FMC, with no ophthalmological alterations from the first exam to the current one. From the first fundus exam during childhood, it could have been considered an imaging investigation of the CNS and a precocious diagnosis, as suggested in a case report of a family which a woman had multiple cerebral cavernous hemangiomas and a choroidal hemangioma, and her father, sister, daughter and son

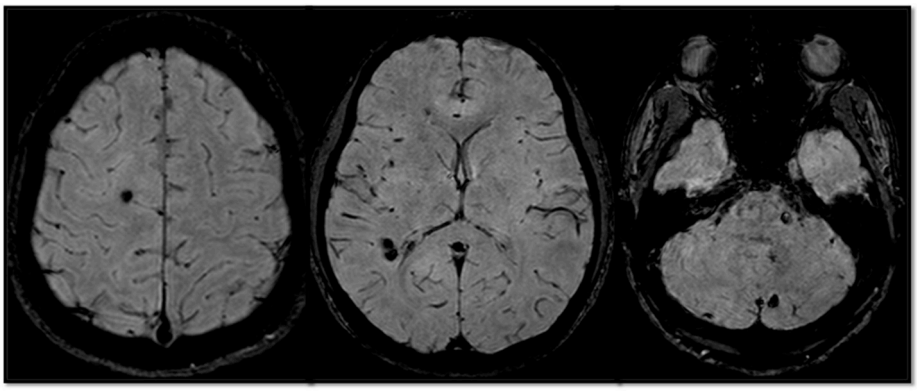

Fig. 4. Multiple cavernomas showing at least five foci of marked hypointense signal on axial T2* weighted images.

manifested some findings of this neuro-oculo-cutaneous syndrome.[5]

Clinical monitoring is necessary, MRI is the technique of choice for follow-up of theses lesions. It is advised an MRI of the spinal cord at the time of diagnosis to serve as a baseline and a control MRI of the brain every one to two years. MRI is also indicated in individuals with new neurologic symptoms. Both symptomatic and asymptomatic siblings should undergo an MRI of the brain to either determine presence, size, and location of the lesions and do a screening MRI, as there may be an increased risk of hemorrhage, spontaneous or due to the use of certain medications.[6]

No clinical treatment is available. Surgical removal of a cerebral cavernous malformation may be justified to prevent a life-threatening hemorrhage.[7]

This long term follow-up case report supports the importance of cerebral vascular investigation when there is suggestion of cavernous hemangioma of the retina, especially in familial cases.

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