Short communication

Cilioretinal artery occlusion in hemochromatosis

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A R T I C L E   I N F O

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A B S T R A C T

Clinical case: We report a case of a 31-year-old woman with a sudden visual loss due to a cilioretinal artery occlusion. The physical examination showed hepatomegaly. Serum iron and ferritin and transferrin saturation were unusually high. The Doppler scan of carotid arteries showed no relevant signs of atheromatous disease. Dilated cardiomyopathy was revealed in the B-scan with subendocardial calcium deposits. Genetic tests were positive for hemochromatosis.

Discussion: Subendocardial calcification due to hemochromatosis could be the embolic source in our patient. This embolic ocular disease is the first presentation of hemochromatosis in this patient.

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Oclusión de arteria ciliorretiniana en la hemocromatosis

R E S U M E N

Caso clínico: Se presenta el caso de una mujer de 31 años con pérdida brusca de visión de un ojo debido a una oclusión de arteria cilioretiniana. La exploración presentaba hepatoen- megaly y en la analítica los niveles séricos de hierro, saturación de transferrina y ferritina estaban elevados. Los perfiles de autoinmunidad y de hipercoagulabilidad fueron normales. El estudio doppler-ultrasónico de los troncos supraaórticos fue anodino, pero la ecografía cardíaca evidenció una miocardiopatía con calcificación subendocárdica. El estudio genético para la hemocromatosis fue positivo.

Discusión: La calcificación subendocárdica secundaria a hemocromatosis puede ser la causa de la oclusión embólica de la arteria cilioretiniana. El cuadro embólico ocular fue la forma de presentación de la hemocromatosis en nuestra paciente.

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Introduction

Hemochromatosis (HC) is an iron metabolism alteration of genetic origin with recessive autosomic inheritance. It courses with pathological iron deposits in various organs. Ophthalmological involvement is rare, with the literature describing iron deposits in various ocular tissue as well as association to dry eye.\(^1\)

The obstruction of the cilioretinal artery (OCLR A) is infrequent. In patients over 60 years of age it is due to vascular alterations and carotid arteriosclerosis. In younger patients it is usually associated to embolic events.

The case of a young patient is described presenting OCLR A due to calcium embolism as the first symptom of hemochromatosis.

Clinic case

Female, 31, with acute loss of visual acuity (VA) in the right eye, with several hours evolution. Personal history includes diabetes mellitus with 20 years evolution and good metabolic control without signs of diabetic retinopathy.

Ophthalmological exploration revealed VA in right eye of 0.7 and in left eye of 1. Absence of right afferent pupil defect,

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Fig. 1 – Fundoscopic image of occluded cilioretinal artery, showing whitish ischemic area in the cilioretinal artery area.

Fig. 2 – Arrow: embolism view at the level of the cilioretinal artery.
normal anterior pole and intraocular pressure. Right eye ocular fundus presented ischemic whitish well-defined area in the papillomacular bundle, around a cilioretinal artery, showing juxtapapillary whitish embolism, probably made up of calcium (Figs. 1 and 2).

The diagnosis was cilioretinal artery obstruction.

At the systemic level, 4 cm hepatomegalia was observed without alteration of liver function.

In order to find the embolic locus, a self immunity and hyper-coagulability study was performed, as well as carotid echography, all with normal results. Cardiac echography and RNM evidenced a non-specific subendocardiac calcification in the left ventricle, with general involvement of the right ventricle and restrictive myocardiopathy pattern (Fig. 3).

Due to the diabetes, hepatomegalia and cardiac alteration of the patient, iron metabolism study was carried out finding: sideremia 142 μg/dl, ferritin 313 μg/l and transferrin saturation 48%.

The diagnosis was presumed HC, confirmed with a genetic study which revealed C282Y/H63D heterozygosis in the HFE gene of chromosome 6.

Treatment was established with monthly phlebotomy, oral anticoagulation and nutritional measures. After 1 month, angiography revealed vascular recanalization (Fig. 4). VA in the right eye improved up to 0.9.

At month 36, the patient has not exhibited new diminished VA episodes, with favorable evolution of the cardiac alteration and hepatomegalia reduction.

Discussion

The cilioretinal arteries are branches of the posterior ciliary arteries which are present in only 15% of individuals. The conclusion of these arteries is rare and accounts for 5% of retinal artery occlusions. The incidence is of 1/10,000.

OCLRA can present in central retinal vein occlusion, associated to ischemic or isolated optic neuropathy, with the latter form (exhibited by the present patient) being more frequent and having the best prognosis.

The diagnosis is usually funduscopy with the visualization in this case of the ischemic retinal area in the territory of the cilioretinal artery visualization of the embolism. In the acute phase, angiography is not essential unless clear retinal ischemia signs or diminished flow cannot be identified.

In what concerns etiology, the occlusion of the central retinal artery usually appears in patients over 60 with carotid artery disease, whereas OCLRA expresses in young patients in relation to embolism events.

In the present case (a young patient) exhibiting retinal artery obstruction, the thromboembolic origin must be the subject of a screening including hyper-coagulability, self immunity and carotid and cardiac echo Doppler study.

The patient presented herein exhibited restrictive myocardiopathy with blood stasis and subendocardiac and ring calcification which was considered to be the origin of the embolism.

The authors have not found any case of retinal vascular occlusion associated to HC in the references.

Hemochromatosis is a recessive autosomic genetic disease caused by gene HFE mutations in chromosome 6. The most frequent is homozygosis for C282Y followed by the mutation (H63D) which gives rise to the composite heterozygote (C282Y/H63D) that appears in 4–7% of cases (including the present patient) who exhibited a subdued form of the disease.

The mutation of said protein produces an accumulation of iron in the body which causes cellular damage due to oxidation caused by free radicals, mostly compromising the liver, myocardium and endocrine glands.

The clinical expressions of the present patient due to HC were diabetes mellitus, hepatomegalia and restrictive myocardiopathy which secondarily involved the formation of thrombi which produced OCLRA (episodes of which led to the study that produced the hemochromatosis diagnosis).

The differential diagnosis should discard other deposits and diseases such as amyloidosis, diseases due to glycogen deposits, sarcoidosis, scleroderma, lupus, neoplastic infiltration, carcinoid syndrome, etc.

The HC diagnosis is suspected due to iron metabolism alterations (plasma iron above 150 μg/dl, ferritin above 200 ng/ml and transferrin saturation above 45%). In the present patient, said values did not immediately raise attention due to being a heterozygote form.

The diagnosis was confirmed detecting the genetic mutations.

Hepatic biopsy is not essential unless ferritin is above 1000 ng/ml, in which case it becomes necessary to discard hepatic fibrosis.

The evolution of this form of HC is generally positive with good response to treatment with periodic phlebotomy is as in the case of the present patient, without the occurrence of new thromboembolic events as well as improvement of cardiac function and hepatomegalia.
Conflict of interests

No conflict of interests has been declared by the authors.

REFERENCES


Fig. 4 – Angiofluorescingraph at month 1: vascular recanalization.