

Atypical Presentation of Antiphospholipid Syndrome: A Case Report

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Key Words

Antiphospholipid syndrome · Subhyaloid hemorrhage · Intraretinal hemorrhage

Abstract

We report an atypical presentation of Antiphospholipid syndrome (APS) with concomitant subhyaloid hemorrhage, engorged and tortuous retinal veins, intraretinal hemorrhages, and cotton wool spots in a 38-year-old female. Medical treatment was preferred to any invasive treatment. The subhyaloid hemorrhage resolved spontaneously and the patient recovered a visual acuity of 20/20 in her right eye 3 months after the initial episode. A prompt diagnosis of this condition is fundamental to consider a systemic treatment to avoid any further thrombosis.

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Introduction

Antiphospholipid syndrome (APS) is an autoimmune disease characterized by the presence of antiphospholipid antibodies, venous and/or arterial thrombosis, and repetitive fetal loss [1, 2]. The laboratory criteria to diagnose APS are high serum levels of lupus anticoagulant as well as anticardiolipin and β -2 glycoprotein-I antibodies [3]. APS can occur with other systemic diseases such as systemic lupus erythematosus or as a single entity. Deep vein thrombosis represents the most frequent APS complication, as it occurs in 29–55% of patients [4]; however, acute or indolent chronic ischemic presentations can involve any organ including the lung, skin, brain, liver, kidneys, adrenal glands, heart, and eyes [2]. Ocular involvement can occur in 8–88% of patients [5–7] and it can be the first sign at presentation. It is therefore important to make a quick diagnosis in order to consider

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anticoagulant treatment to prevent further thrombosis, as the risk of this complication is up to 29% in untreated APS patients [8]. Visual symptoms at presentation vary from a reduction of visual acuity and amaurosis fugax to transient scotoma and visual field defect [2], all of which can be unilateral or bilateral. It is important to underline that, when amaurosis fugax involves both eyes simultaneously, it is often a manifestation of central nervous system ischemia. Ocular discomfort, conjunctival hyperemia, and pain have often been reported in the literature as common symptoms of APS patients [2].

Case Presentation

A 38-year-old female patient presented with reduced vision in the right eye, which had occurred the same day. She also reported dyspnea for the past 30 days after minor physical exercise. The patient had a negative ocular and medical history and no previous miscarriage as well as no relevant family history for ophthalmic pathologies. She stated that the reduced visual acuity appeared suddenly after mild physical exercise. Visual acuity was finger count in the right eye (1-meter distance) and 20/20 in the left eye.

Fundoscopy examination revealed a subhyaloid hemorrhage, engorged and tortuous retinal veins, intraretinal hemorrhages, and cotton wool spots (fig. 1). Fundus fluorescein angiography demonstrated a delayed inferior hemiretinal venous filling (fig. 2). OCT scans confirmed the subhyaloid localization of the hemorrhage (fig. 3). As choroidal ischemia has been previously reported with APS [9], indocyanine green angiography was performed, which did not show any significant modification of choroidal perfusion. Prothrombotic conditions secondary to other factors such as sepsis, homocystinemia, and genetic defects of coagulation factors (thrombin mutations, factor V Leiden, antithrombin deficiency, etc.) were ruled out.

Laboratory investigations showed that the erythrocyte sedimentation rate, IgG anticardiolipin, and β -2 glycoprotein-I antibody were above normal limits. Thromboplastin inhibition test was strongly positive for lupus anticoagulant, while antinuclear antibody and antinative DNA antibodies were negative. Serum total complement (C3, C4), proteins S and C, and antithrombin III were within normal limits. Other studies were normal or negative, including complete blood count, platelet count, renal, liver, and thyroid functions, hemoglobin A1c, ferritin, transferrin, and LDL cholesterol. Finally, the patient was found to have iron deficiency anemia.

Discussion

The patient met the criteria for APS; however, according to the American College of Rheumatology classification criteria, a concomitant diagnosis of systemic lupus erythematosus could not be established. She started treatment with hydroxychloroquine 200 mg/daily and ticlopidine 250 mg/daily. Considering the central location of the hemorrhage, we deferred any invasive treatment and opted for watchful waiting. The subhyaloid hemorrhage resolved spontaneously and the patient recovered a visual acuity of 20/20 in her right eye 3 months after the initial episode (fig. 4).

This is the first reported case of simultaneous subhyaloid hemorrhage and branch retinal vein subocclusion as presentations of APS. A prompt diagnosis of this condition is fundamental to consider a systemic treatment to avoid any further thrombosis. This is often difficult, as ocular manifestations of APS can be variable. Assessing the patient's thrombotic

risk by investigating the presence of high blood pressure and hypercholesterolemia, the use of tobacco and oral contraceptive agents as well as the history of previous of thrombosis, fetal loss, and acute ischemic events is mandatory in the process of deciding whether to start systemic treatment.

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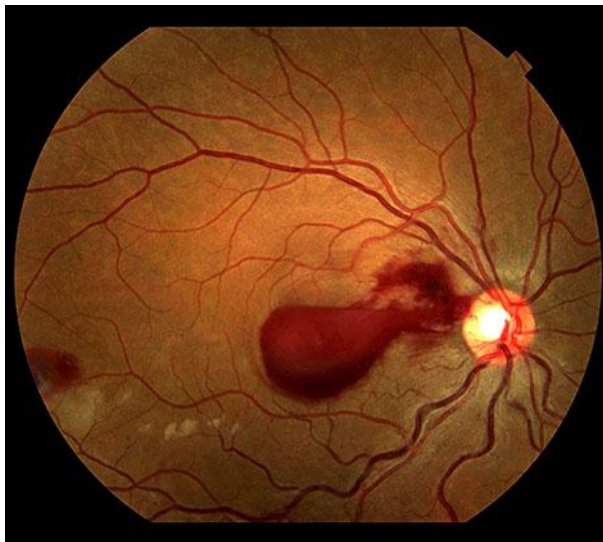


Fig. 1. Fundoscopic photography at presentation.

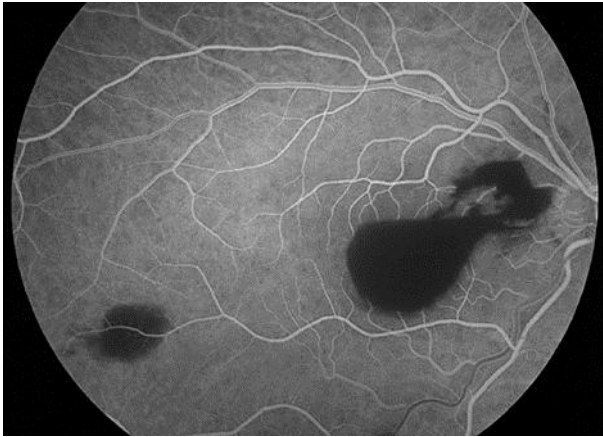


Fig. 2. Fundus fluorescein angiography demonstrates a delayed inferior hemiretinal venous filling.

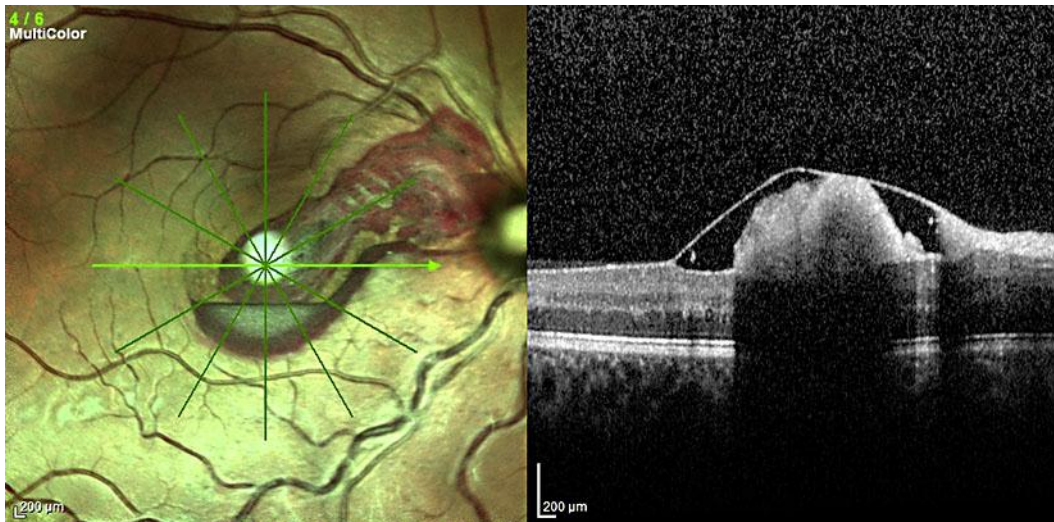


Fig. 3. OCT scans confirm the subhyaloid localization of the hemorrhage.



Fig. 4. Fundoscopic photography 3 months after the initial episode.