International Clinical and Medical Case Reports
2024 Volume 3, Issue 1
DOI: 10.59657/2837-5998.brs.24.041

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When Pseudoxanthoma Elasticum Breaks Through the Striae: A Case Report

S.Sebbata, T.Abdellaoui, H.Brarou, S.Laaouina, S.Sadiki, B.Oubaaz, A.Fiqhi, Y.Mouzari, A.Oubaaz

***Corresponding author: Soundouss Sebbata,**

Ophthalmology Department, Military Hospital of Rabat, Morocco.

Abstract

Angioid streaks (AS) represent Bruch's membrane dehiscence, it is a rare condition often part of a general pathology, essentially pseudoxanthoma elasticum (PXE). We report the case of a 55-year-old woman whose condition led to a fundus abnormality following low-kinetic trauma eventually resulting in the pathology's underlying identification. She exhibited reduced visual acuity in her right eye, and upon fundus examination, we observed a subretinal haemorrhage affecting the posterior pole (explaining the visual impairment) and the peripapillary region, alongside the identification of angioid streaks encircling the optic disc. A proper clinical examination alongside a dermatologist was conducted and the diagnosis of Pseudoxanthoma elasticum was established. This condition remains the most common systemic disease associated with angioid streaks. The incidence of AS in PXE ranges from 59% to 87% depending on whether the diagnosis is made clinically or by skin biopsy. Choroidal neovascularization represents a significant concern in this pathology, emphasizing the crucial need for vigilant monitoring to identify this complication. Additionally, it is imperative to actively investigate extraocular complications, particularly those associated with the cardiovascular manifestations of the disease.

Keywords: pseudoxanthoma elasticum; angioid streaks; contusion

Introduction

Angioid streaks (AS) represent Bruch's membrane dehiscence's. They are confined to the peripapillary region or extended to the posterior pole and are frequently bilaterally located. This rare condition is often part of a general pathology, essentially pseudoxanthoma elasticum (PXE), which may remain silent or be revealed by one of its complications. We present a case in which a low-kinetic trauma resulted in a fundus abnormality, leading to the identification of the underlying pathology.

Case report

A 55-year-old woman presented to the ophthalmologic emergency department with a sudden loss of vision in the right eye (OD) following a contusive trauma. According to the patient's account, she was struck by another woman's backhand during a gym session. She had no history of underlying disease or previous ocular surgery. Examination of the right eye showed a best-corrected visual acuity at counting fingers from 10 centimetres. On biomicroscope; no redness nor conjunctival haemorrhage was noted; the anterior chamber was calm, the pupillary reflex was present, the intraocular pressure was 13 mmHg and the lens was intact. Fundus examination showed a subretinal haemorrhage involving the posterior pole and the peripapillary area. We came across radiating lines arranged in a wheel spoke and extending from the optic disc revealed to be AS (figure 1-a). On the left eye, the visual acuity was 10/10 and the slit lamp examination of the anterior segment was unremarkable. The fundus examination found the same AS lesions as in the right eye (figure1-b).

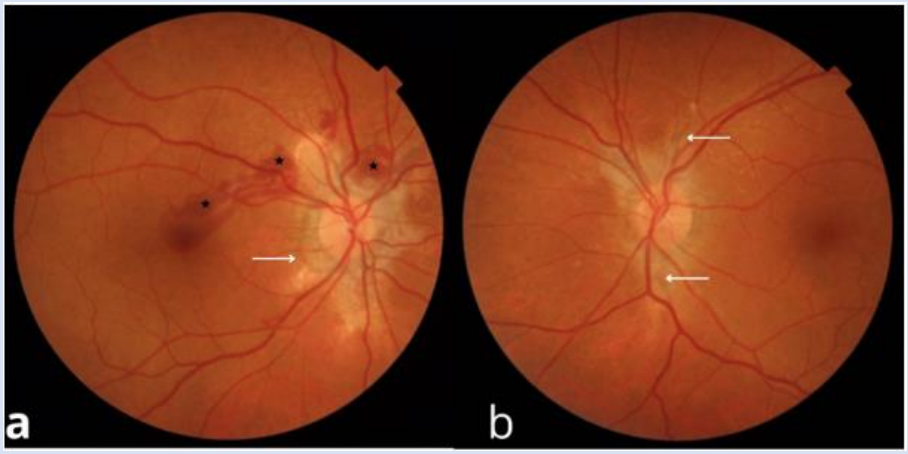

Figure 1: 1a- photofundus of the right eye demonstrating hemorragies (asterix) longing the angioid streaks lines (white arrows) 1b- photofundus of the left eye demonstrating angioid streaks lines

On autofluorescence, the stria appears as an irregular black line, with a variable hyper autofluorescence border. The red-free fundus photo enhances the striae (figure 2).

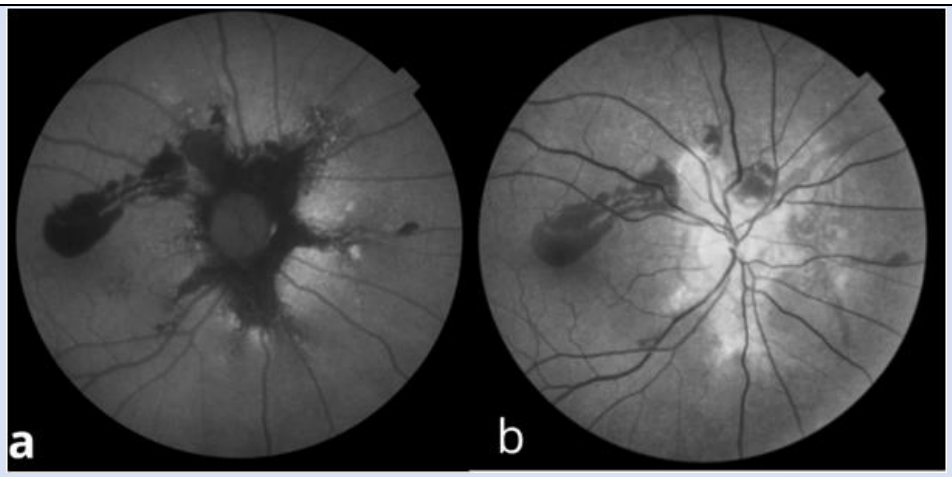

Figure 2: 2a- autofluorescence image of the right eye showing areas of decreased autofluorescence corresponding to alteration of the retinal pigment epithelium; haemorrhages/ 2b- Redfree fundus photo enhancing the angioid streaks

Optical coherence tomography (OCT) showed in the right eye a disorganization of the outer structures of the macular zone, corresponding to the hemorrhage seen on the fundus, and appeared to be longing throughout the streaks. No neovascularisation was noted (figure 3).

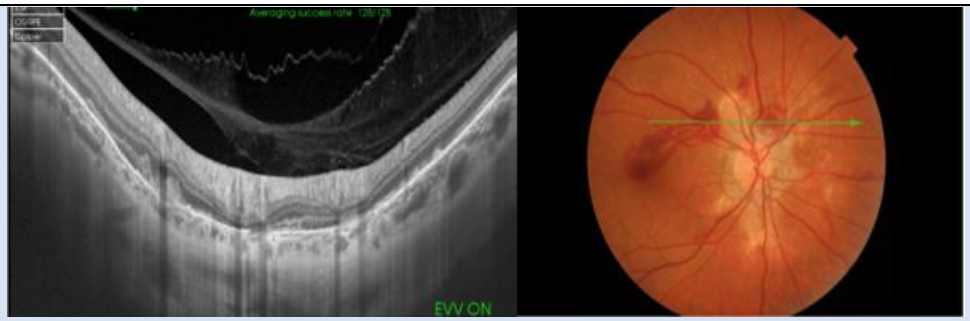

Figure 3: Macular OCT of the right eye showing a disorganisation of the outer retinal structures and accumulation of the blood within the retinal layers.

Upon conducting a comprehensive patient examination, distinct yellow papules were observed in the cervical region (figure 4).

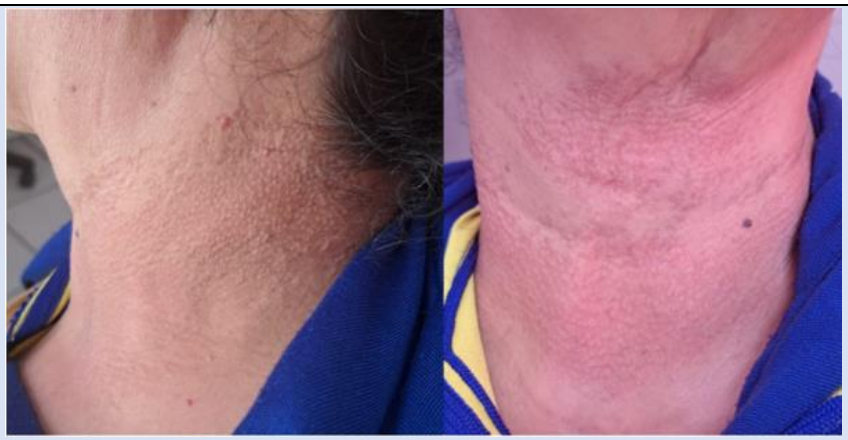

Figure 4: Images of papules observed in the cervical region.

The patient noted their presence since early childhood, and they have consistently remained unchanged over time without any interventions or treatments. Given the ocular and skin manifestations, it was deemed prudent to refer the patient for a more in-depth assessment at the dermatology department. Considering all the clinical features, the diagnosis of Pseudoxanthoma elasticum was established. The subretinal haemorrhage in the right eye was partially absorbed three weeks after the injury. The fundus examination showed a white well-defined line involving the macula corresponding to a Bruch's membrane rupture. In instances of Bruch's membrane damage, a strategy of therapeutic abstention was employed. Close monitoring for the potential development of neovascularization was undertaken. The progression was positive, marked by the resolution of the retinal haemorrhage. Subsequent follow-ups, conducted six months post-trauma, revealed no signs of neovascularization, but remaining of fibrosis in the macular region set by OCT.

Discussion

Angioid streaks (AS) were first defined by Doyne in 1889 as "irregular jagged lines extending from the peripapillary region to the retinal periphery". Initially thought to be of a vascular origin, it was only later revealed to be a dysfunction of Bruck's membrane [1] . AS are calcified, brittle Bruch's membrane dehiscences that often have ragged borders. Their colour varies between red, grey, mixed, or pigmented independently from the underlying systemic disease. They can be confined only to the peripapillary region or extended to the posterior pole and are frequently bilaterally located. Mostly surrounding the optic head, they appear to extend outward toward the retinal periphery. This specific localization is attributed to the mechanical force applied by the extraocular muscles to a posterior pole that is both fragile and less flexible [2]. It has been hypothesized that this alteration of the Bruch membrane may also lead to localized atrophy of the choriocapillaris and the Retinal pigment epithelium [3]. If angioid streaks are not located at the fovea, patients primarily stay asymptomatic, and the condition is frequently discovered accidentally [1]. Similar to our situation, it was discovered in certain instances as a result of blunt trauma [4]. PXE remains the most common systemic disease associated with AS that may be indicative of the condition [5]. The incidence of AS in PXE is ranging from 59% to 87% depending on whether the diagnosis is made clinically or by skin biopsy [1]. It is a rare, recessive, genetic multisystem disease that mostly affects the elastic tissues of the eyes, skin, and cardiovascular system. It results from a defect in a reputed transport protein encoded by ABCC6 gene [6]. The diagnosis is established in individuals with characteristic skin lesions and at least one characteristic retinal finding such as; Angioid streaks, peau d'orange, drusen of the optic nerve head, and comet tail lesions [7]. Skin lesions include the presence of yellow-white papules located on flexion zones on the neck, armpits, groin and popliteal fossa with a "chicken skin" appearance on the neck, axillae, and/or antecubital fosse [7,8]. The diagnosis can also be confirmed by the presence of calcified dystrophic elastic fibres on skin biopsy. Some of the cardiovascular manifestations of PXE are arterial hypertension, cardiomyopathy, atherosclerosis, calcification of peripheral arteries and mitral valve prolapse. All that could lead to cardiac failure [9]. Choroidal neovascularisation (CNV) is counted as a natural evolution of the disease. Even though it initially has little effect on visual function by itself and

largely seems to be stable, CNV affects 42%–86% of patients with AS. It continues to be the most morbid complication of this condition [10]. Due to exudation, bleeding, and subsequent subretinal fibrosis and atrophy, CNV may cause significant and rapid vision impairment. The visual prognosis is dismal without treatment and frequently results in legal blindness [2]. Even with minimal trauma, individuals with AS may experience ruptures in Bruch's membrane, resulting in subsequent visual impairment, as observed in our patient. It is imperative to systematically screen for CNV, as its existence could exacerbate the already incurred damage to Bruch's membrane and implies a different management approach [8].

Conclusion

Patients with angioid striae need to be vigilant, as even minor trauma can be fatal and compromise their vision. Medical surveillance is also crucial to identify potential complications associated with these striae, extending beyond instances of trauma.

Conflict of interest

None; This manuscript has not been presented in a conference. There is no commercial interest that could cause or be perceived to be a conflict of interest.

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Cite this article: Soundouss S, T.Abdellaoui, H.Brarou, S.Laaouina, S.Sadiki, et al. (2024). When Pseudoxanthoma Elasticum Breaks Through the Striae: A Case Report. *International Clinical and Medical Case Reports,* BioRes Scientia Publishers. 3(1):1-4. DOI: 10.59657/2837-5998.brs.24.041

Article History: **Received:** March 16, 2024 | **Accepted:** May 10, 2024 | **Published:** July 26, 2024