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Pseudoxanthoma Elasticum Revealed by Angioid Streaks Complicated with Choroidal Neovascularization (Case Report)

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Abstract **Case Report**

Pseudoxanthoma elasticum (PXE) is a systemic disorder often revealed by angioid streaks (AS), whose main vision-threatening complication is choroidal neovascularization (CNV). We report the case of a 42-year-old patient presenting with decreased visual acuity and metamorphopsia in the right eye. Fundus examination showed bilateral AS with a "peau d'orange" pattern, and OCT confirmed CNV with subretinal fibrosis. Despite intravitreal anti-VEGF therapy leading to partial anatomical improvement, visual function remained limited. This case highlights the importance of early detection and prompt treatment of PXE-related CNV to preserve vision.

Keywords: Pseudoxanthoma Elasticum, Angioid Streaks, Choroidal Neovascularization, Intravitreal Anti-VEGF Therapy.

INTRODUCTION

Angioid streaks (AS) are disruptions of Bruch's membrane resulting from pathological calcification of the elastic layer, which predispose to the development of choroidal neovascularization (CNV) within the subretinal space [1, 2]. CNV represents the most severe complication of AS, typically arising in young adults during the third or fourth decade of life, and is associated with a poor visual prognosis, particularly when the macula is involved [3–6]. Although AS may occur sporadically, they are frequently linked to systemic conditions, most notably pseudoxanthoma elasticum (PXE), reported in 53–65% of cases [7, 8]. Other associations include Paget's disease of bone, sickle-cell disease, and Ehlers-Danlos syndrome [9–11]. We present the case of a patient with AS complicated by CNV, which led to the diagnosis of PXE, emphasizing the role of ophthalmologic assessment both in the early recognition of vision-threatening complications and in the detection of underlying systemic disorders.

CASE REPORT

A 42-year-old patient, with no significant systemic medical history, presented with an incidental finding of decreased visual acuity in the right eye, accompanied by metamorphopsia.

On initial ophthalmologic examination, best-corrected visual acuity (BCVA) was 1/10 with P4 in the right eye and 10/10 with P2 in the left eye. The anterior segment examination was unremarkable, and intraocular pressure was measured at 17 mmHg in both eyes.

Fundus examination revealed bilateral angioid streaks associated with a characteristic "peau d'orange" appearance in the temporal retina. In the right eye, an intraretinal hemorrhage was observed in association with perimacular exudates overlying a fibrovascular membrane (Figure 1). Fluorescein angiography (FA) confirmed the diagnosis of angioid streaks complicated by choroidal neovascularization (CNV) in the right eye (Figure 1).

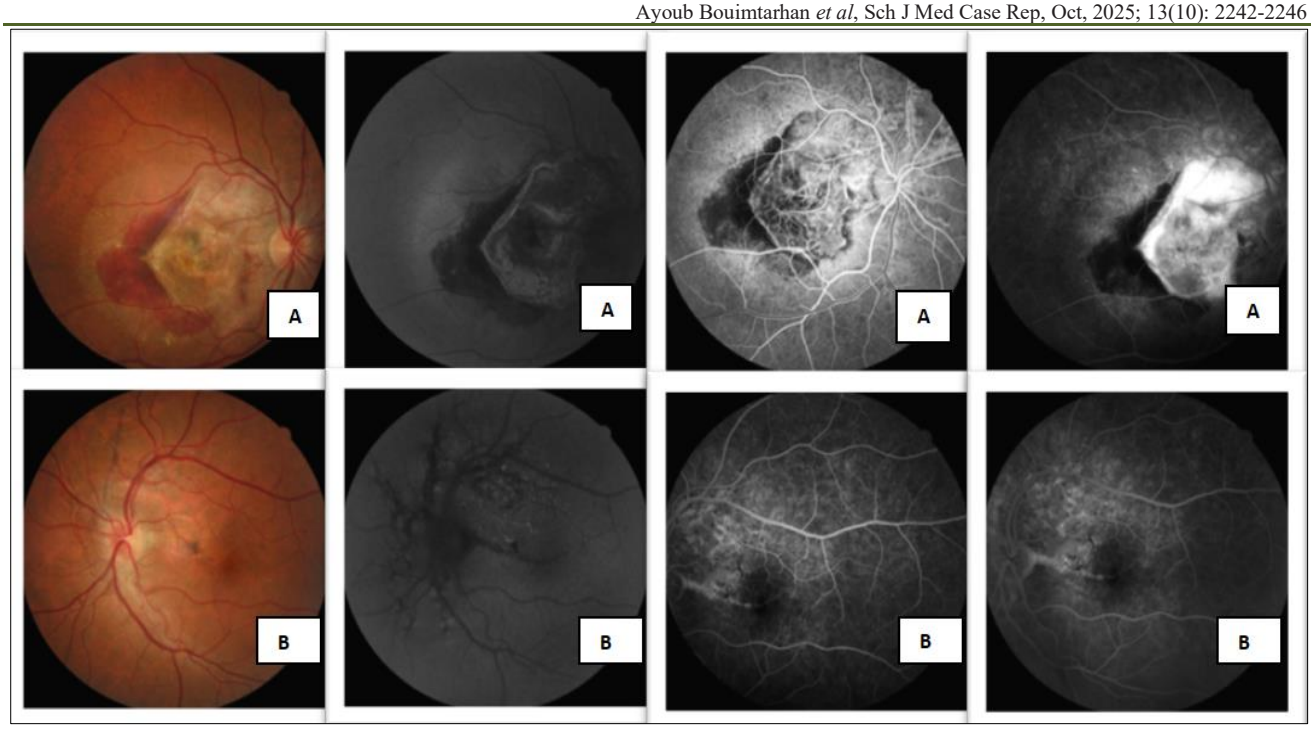

Figure 1
(A) Intraretinal hemorrhage associated with perimacular exudates, overlaid by a fibrovascular membrane demonstrating late-phase hyperfluorescence on fluorescein angiography
(B) Angioid streaks associated with a characteristic "peau d'orange" appearance in the temporal retina, without evidence of fluorescein leakage.

Spectral-domain optical coherence tomography (SD-OCT) of the right eye demonstrated a disrupted foveal profile with loss of the normal foveal depression, extensive macular remodeling with cystic spaces, subretinal fibrosis, and pigment epithelial detachment (PED). Central foveal thickness was measured at 391 µm (Figure 2).

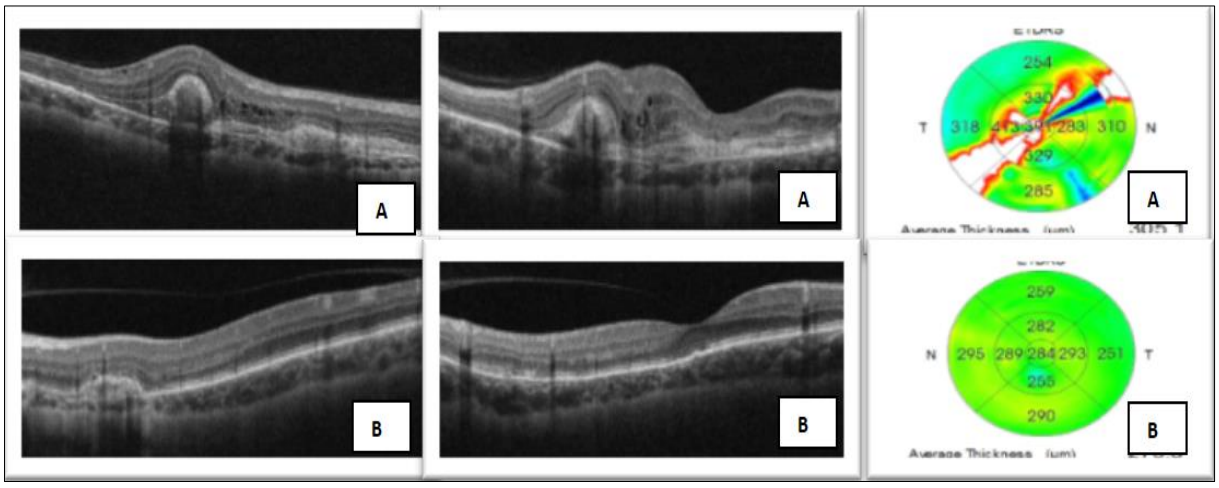

Figure 2
(A) Loss of the foveal depression with marked macular remodeling, associated with small intraretinal cystic spaces, elevation of the retinal pigment epithelium, and subretinal fibrosis.
(B) Retinal pigment epithelium detachment without evidence of exudation, with preservation of the foveal depression.

In the left eye, SD-OCT revealed a pigment epithelial detachment without exudation, while the foveal contour was preserved (Figure 2).

As part of the etiological work-up, dermatologic examination revealed multiple grouped papules and micropapules arranged in plaques on the lateral aspects of the neck and in the axillary folds. A skin

biopsy confirmed the diagnosis of pseudoxanthoma elasticum (PXE) (Figure 3).

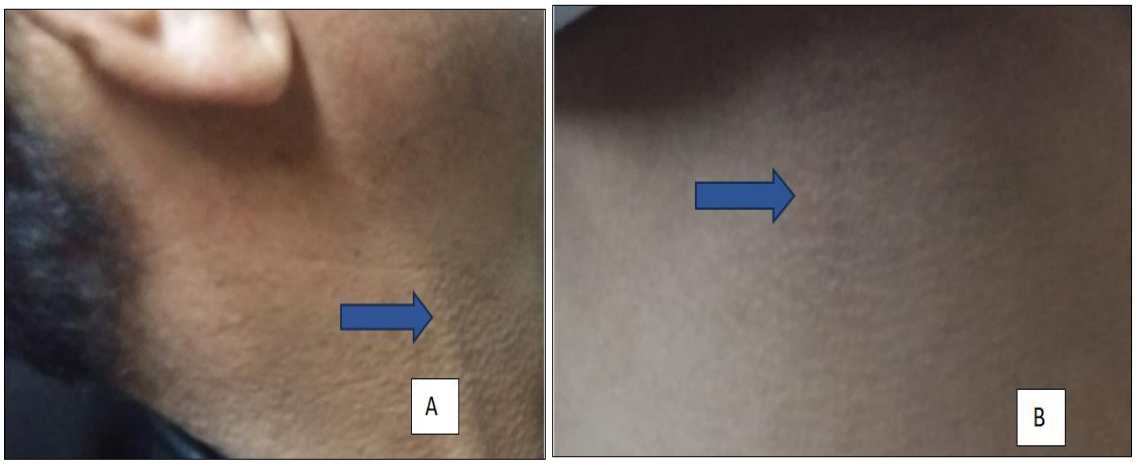

Figure 3: Clusters of micropapules arranged in plaque-like formations (blue arrow), located on the lateral aspect of the neck (A) and in the axillary fold (B) of the patient.

Complementary cardiovascular assessment, including ECG Holter monitoring and echocardiography, was within normal limits and did not reveal any cardiovascular involvement.

The patient underwent an intravitreal injection of anti-VEGF in the right eye. Follow-up optical coherence tomography (OCT), performed one month later (figure 4), demonstrated a slight anatomical improvement, with a reduction in foveal thickness to 273 µm compared to 391 µm at baseline. However, this structural improvement contrasted with a stagnation of visual function, as visual acuity remained limited to 1/10, P4.

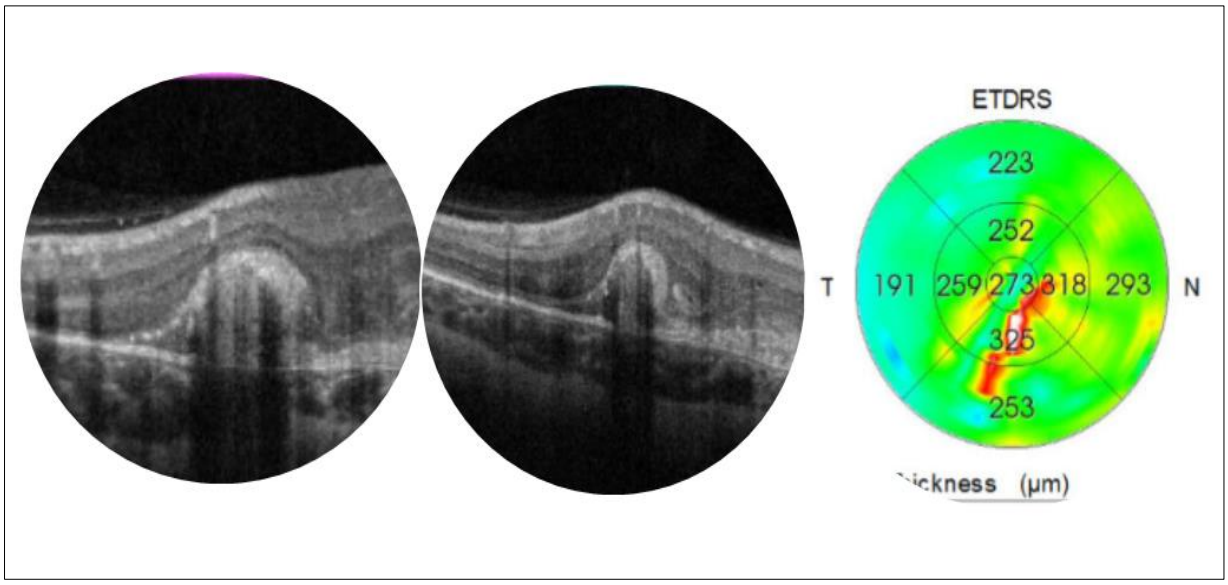

Figure 4: OCT image obtained one month after intravitreal anti-VEGF injection.

DISCUSSION

Angioid streaks (AS) are usually asymptomatic unless complicated by foveal involvement, and are clinically identified as dark, radially oriented lines at the posterior pole [12]. Their strong association with pseudoxanthoma elasticum (PXE) is well established, being present in about 85% of cases, where ophthalmic findings often precede the systemic diagnosis [13]. PXE is an autosomal recessive disorder linked to mutations in the **ABCC6** gene, with a prevalence estimated between 1/25,000 and 1/100,000, characterized by calcification of elastic fibers affecting skin, eyes, and cardiovascular tissues [14–16].

Fundus findings often include the “peau d’orange” pattern, while cutaneous lesions manifest as yellowish papules on flexural areas [17, 18]. Cardiovascular involvement may occur, underscoring the need for systematic evaluation [19]. The most serious

ocular complication is choroidal neovascularization (CNV), reported in 72–86% of patients, and responsible for significant visual loss [20].

Therapeutic strategies such as laser photocoagulation or photodynamic therapy have shown inconsistent results [21]. The advent of intravitreal anti-VEGF therapy has dramatically improved management, with studies confirming short- and medium-term efficacy of ranibizumab in reducing CNV activity and improving vision [22, 23]. Nevertheless, recurrences remain frequent, and long-term outcomes are uncertain, requiring individualized follow-up and treatment protocols [24, 25].

CONCLUSION

Pseudoxanthoma elasticum (PXE) is a systemic disorder in which angioid streaks (AS) often guide diagnosis. The prognosis depends largely on ocular and cardiovascular involvement. Choroidal neovascularization (CNV) is the most vision-threatening complication, typically affecting young adults and leading to severe visual impairment if untreated. Intravitreal anti-VEGF injections remain the only effective therapy, though their benefit is temporary and requires close follow-up. Preventive measures, including patient education on early symptoms and avoidance of trauma, are essential to improve outcomes.

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