Scholars Journal of Medical Case Reports
Abbreviated Key Title: Sch J Med Case Rep
ISSN 2347-9507 (Print) | ISSN 2347-6559 (Online)
Journal homepage: https://saspublishers.com

OPEN ACCESS

Ophthalmology

Ophthalmologic Complications of Pseudoxanthoma Elasticum

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DOI: https://doi.org/10.36347/sjmcr.2026.v14i05.107 | **Received:** 07.04.2026 | **Accepted:** 25.05.2026 | **Published:** 30.05.2026

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Abstract | **Case Report**

Pseudoxanthoma elasticum [PXE] is a rare multisystem genetic disorder characterized by progressive calcification of elastic fibers affecting primarily the skin, cardiovascular system, and eyes. Ophthalmologic involvement, mainly represented by angioid streaks and choroidal neovascularization [CNV], is a major cause of visual impairment in young patients. We report the case of a 54-year-old woman with clinically and histologically confirmed pseudoxanthoma elasticum who presented with progressive bilateral visual loss. Fundus examination revealed bilateral angioid streaks associated with a "peau d'orange" appearance and fibrotic macular changes. Multimodal imaging including fluorescein angiography, optical coherence tomography [OCT], and OCT angiography [OCT-A] demonstrated inactive cicatricial choroidal neovascularization complicating angioid streaks. Management consisted of close monitoring and intravitreal anti-VEGF injections administered on a pro re nata [PRN] basis. Ophthalmologic complications of PXE mainly result from calcification and fragility of Bruch's membrane. Anti-VEGF therapy is currently considered the standard treatment for PXE-associated CNV, allowing anatomical and functional stabilization in most cases. However, long-term evolution remains marked by the risk of neovascular recurrence and progressive macular atrophy. Novel therapeutic approaches targeting calcification pathways and underlying genetic abnormalities are currently under investigation. Ophthalmologic complications of PXE represent an important cause of visual loss. Regular follow-up based on multimodal imaging and early management of choroidal neovascularization are essential to improve visual prognosis. Targeted therapies and gene therapy represent promising future perspectives.

Keywords: pseudoxanthoma elasticum; angioid streaks; choroidal neovascularization; anti-VEGF; Bruch's membrane; OCT-A.

INTRODUCTION

Pseudoxanthoma elasticum [PXE] is a rare hereditary multisystem disorder inherited in an autosomal recessive manner and mainly associated with mutations in the *ABCC6* gene located on chromosome 16p13.1. [1] This genetic abnormality leads to progressive ectopic calcification of elastic fibers, primarily affecting the skin, cardiovascular system, and ocular tissues. [2]

Ophthalmologic involvement is a major manifestation of PXE and constitutes an important cause of visual disability in young individuals. [3] The pathophysiology mainly relies on progressive alteration of Bruch's membrane, resulting in its calcification and fragmentation, thereby promoting the development of angioid streaks and subsequent choroidal neovascularization [CNV]. [4]

The progression of macular complications determines the visual prognosis of the disease. The aim of this work is to present the main ophthalmologic complications of PXE, their diagnostic modalities, currently available treatments, and future therapeutic perspectives.

CASE REPORT

We report the case of a 54-year-old woman followed for six years for clinically and histologically confirmed pseudoxanthoma elasticum, referred for progressive worsening of bilateral visual acuity.

The patient complained of progressive central visual loss associated with metamorphopsia evolving over several months, without ocular pain or inflammatory symptoms. Past medical history was notable for cervical cutaneous lesions suggestive of PXE and well-controlled arterial hypertension under medical treatment.

Ophthalmologic examination revealed a best-corrected visual acuity of 2/10 in the right eye and 3/10 in the left eye. Anterior segment examination showed bilateral early cortico-nuclear cataract without other abnormalities. Intraocular pressure was 15 mmHg in both eyes.

Fundus examination demonstrated bilateral angioid streaks radiating from the optic disc associated with a "peau d'orange" appearance of the posterior pole. Bilateral macular changes with subretinal foveal fibrosis and diffuse pigmentary alterations were also observed.

Fluorescein angiography revealed hyperfluorescent window defects corresponding to angioid streaks associated with heterogeneous late macular hyperfluorescence consistent with old cicatricial choroidal neovascularization without clear exudative activity. [Figure 1]

Macular OCT showed severe disruption of the foveal architecture with hyperreflective subretinal fibrosis, serous retinal detachment, and diffuse irregularities of the retinal pigment epithelium. [Figure 2] OCT-A demonstrated an inactive choroidal neovascular network embedded within subretinal fibrosis associated with perifoveal choriocapillaris flow impairment. [Figure 3]

The diagnosis of bilateral macular complications of pseudoxanthoma elasticum complicated by cicatricial choroidal neovascularization was established. Close follow-up associated with a PRN intravitreal anti-VEGF injection regimen was initiated with regular macular OCT monitoring. [Figure 4]

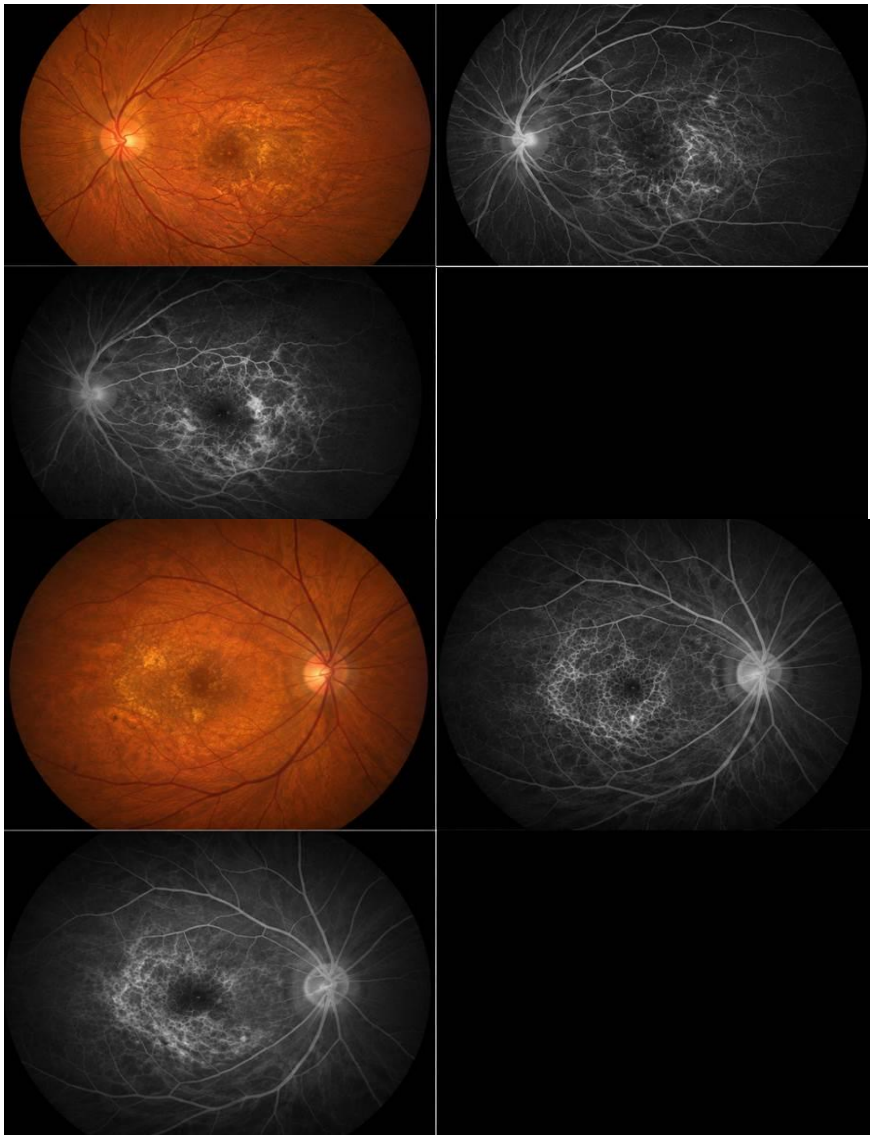

Figure 1: Fluorescein angiography demonstrating hyperfluorescent angioid streaks exhibiting a window defect, associated with heterogeneous late macular hyperfluorescence.

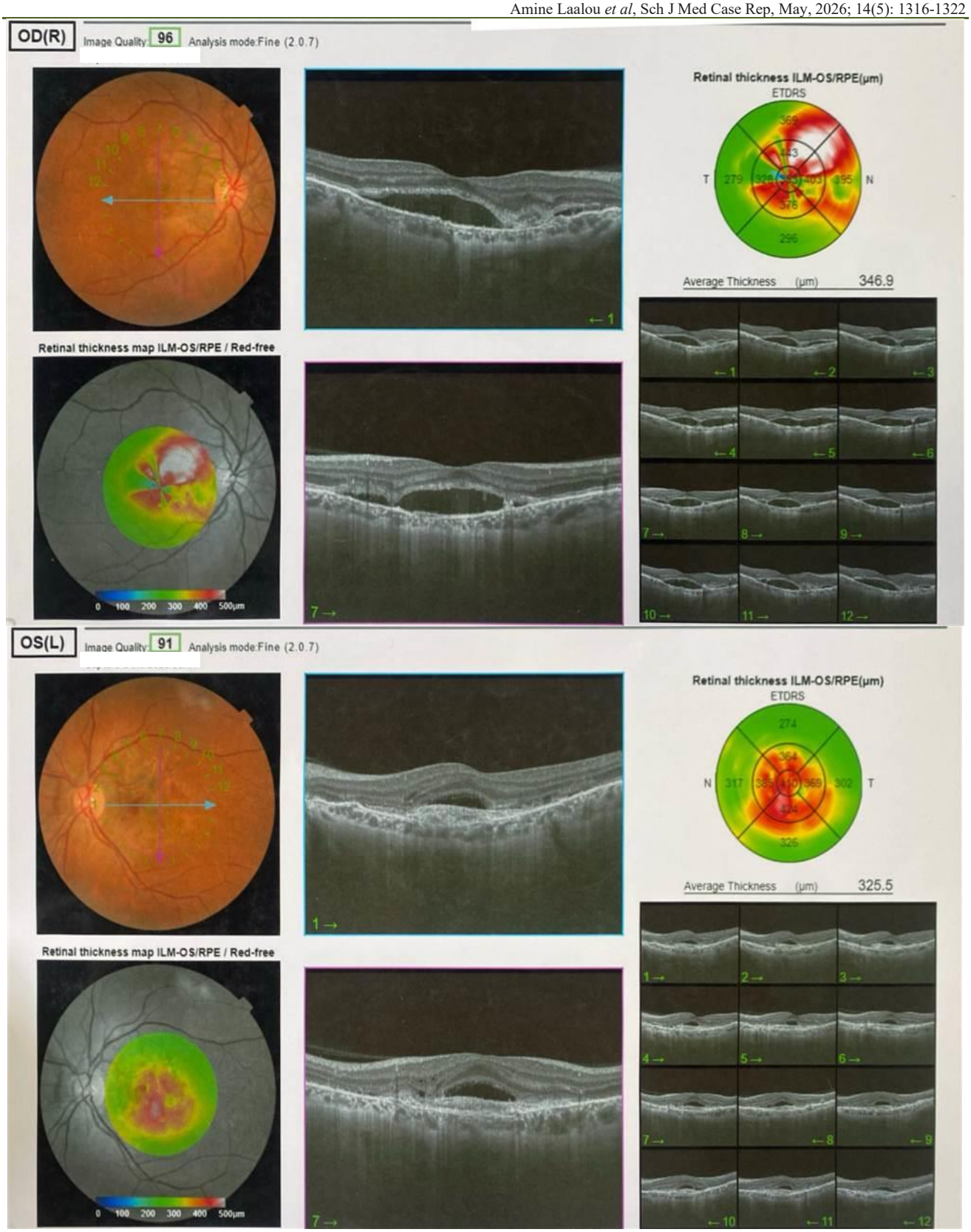

Figure 2: Macular OCT showing severe disruption of foveolar architecture, with hyperreflective subretinal fibrosis, serous retinal detachment, and diffuse irregularities of the retinal pigment epithelium.

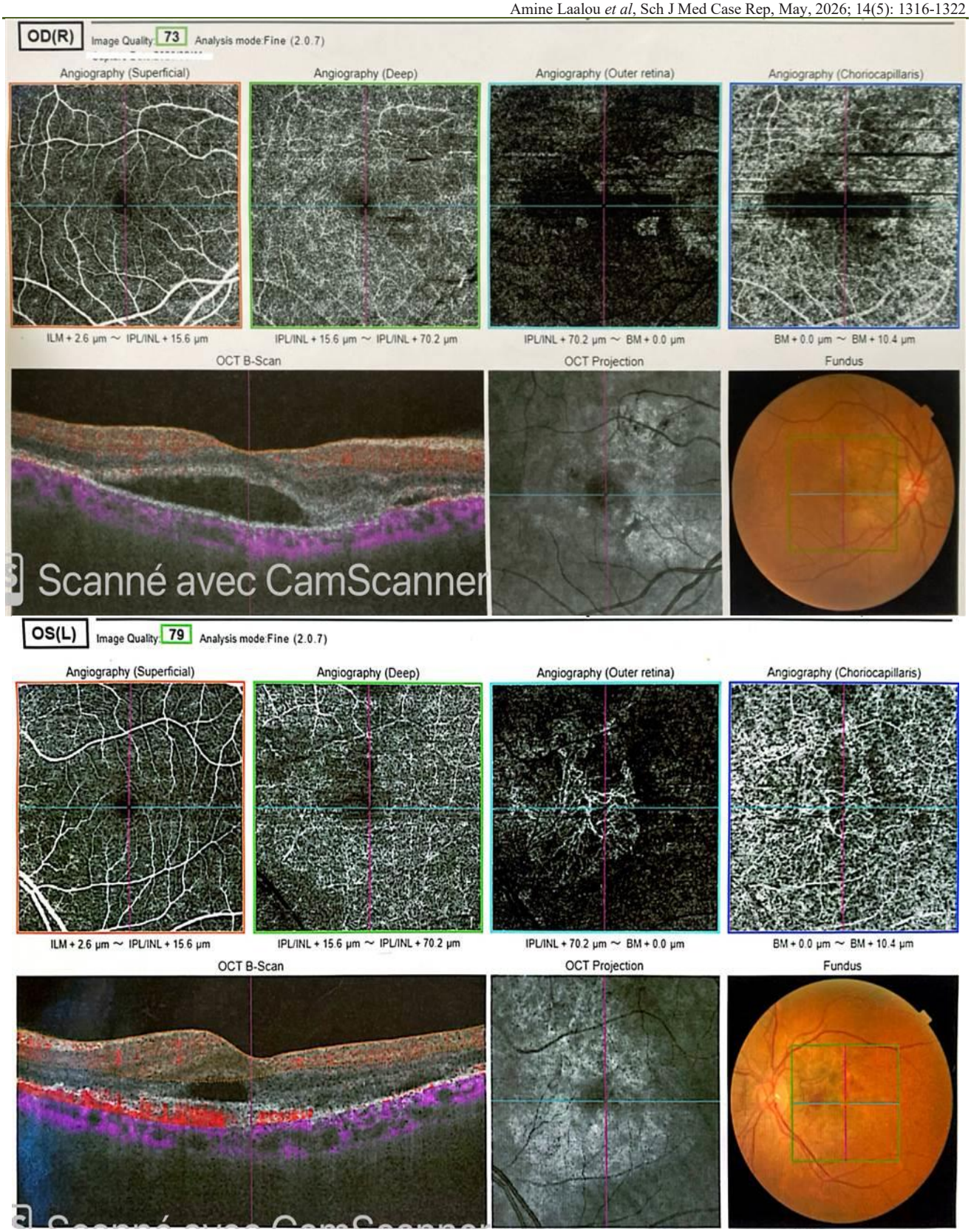

Figure 3: OCT-A revealing a choroidal neovascular network associated with rarefaction of perifoveolar choriocapillary flow.

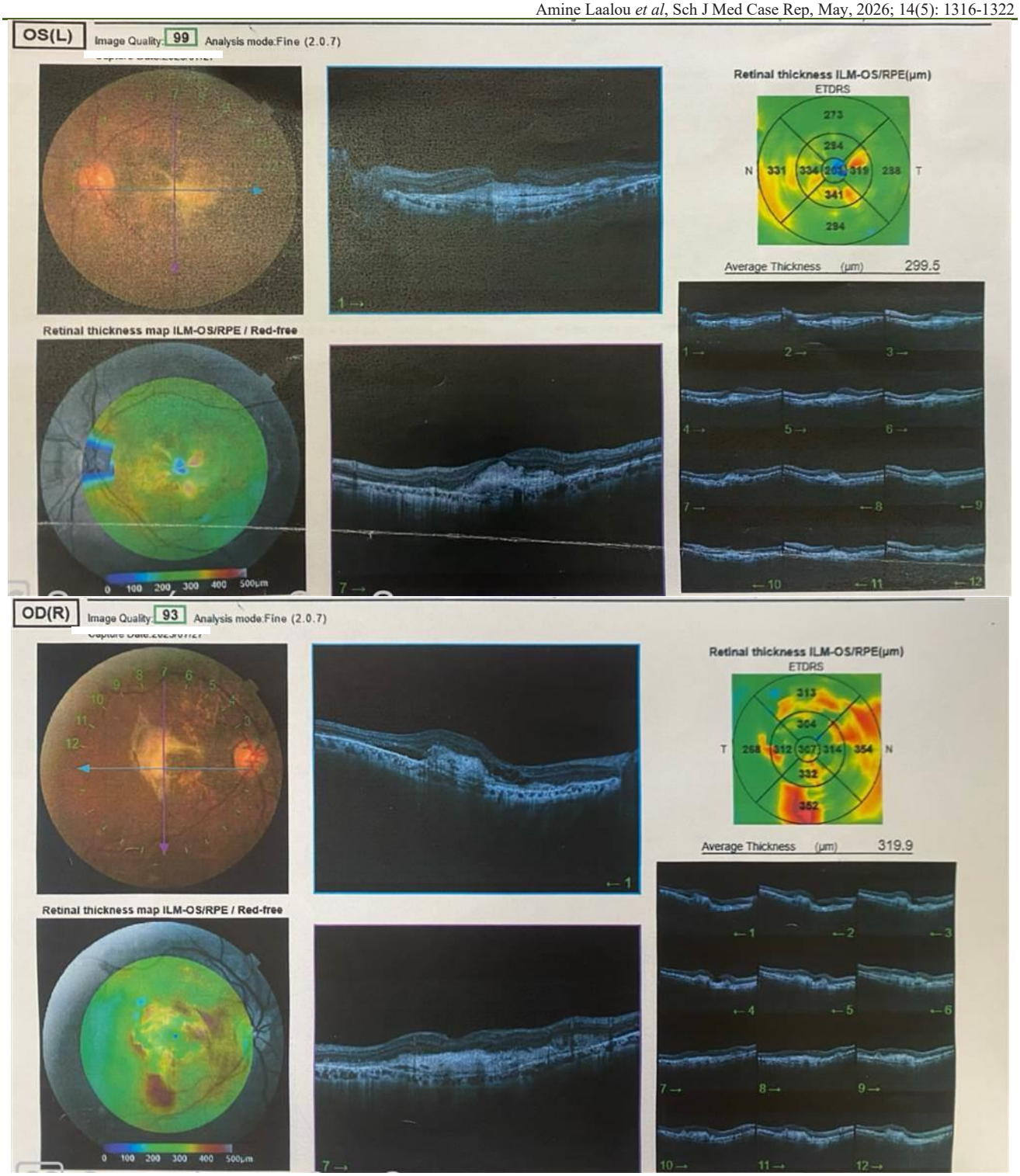

Figure 4: Macular OCT revealing resolution of the serous retinal detachment associated with subretinal fibrosis.

DISCUSSION

Ophthalmologic manifestations of PXE generally occur after cutaneous lesions but may occasionally reveal the disease. [5] Early fundus abnormalities are dominated by the "peau d'orange" appearance, corresponding to diffuse alterations of Bruch's membrane. [6]

Angioid streaks constitute the most characteristic ophthalmologic manifestation. They appear as brownish or grayish lines radiating from the optic disc. [7] Histologically, they correspond to breaks in Bruch's membrane weakened by calcification processes. [8]

The major vision-threatening complication remains choroidal neovascularization, secondary to the

proliferation of neovessels through ruptures in Bruch's membrane. This complication may lead to subretinal hemorrhage, macular edema, and cicatricial fibrosis responsible for irreversible visual acuity loss. [9]

Diagnosis currently relies on multimodal imaging. OCT enables the assessment of retinal structural changes and neovascular membrane activity, while fluorescein angiography and OCT-A provide detailed vascular characterization of lesions. [10,11]

Therapeutic management has dramatically evolved with the advent of anti-VEGF therapy. Before its introduction, treatment mainly relied on laser photocoagulation and photodynamic therapy, whose functional outcomes remained limited. [12]

Currently, intravitreal injections of ranibizumab, bevacizumab, or aflibercept represent the standard treatment for PXE-associated CNV. [13,14] PRN ["pro re nata"] and treat-and-extend regimens are the most commonly used protocols. [15] These treatments generally allow anatomical and functional stabilization, although the risk of neovascular recurrence and progressive macular atrophy persists over the long term. [16,17]

Current research aims to target the disease earlier at the pathophysiological level. The deficiency of inorganic pyrophosphate observed in PXE has led to the development of anti-calcification strategies such as bisphosphonates and pyrophosphate supplementation. [18]

Furthermore, gene therapy approaches targeting the *ABCC6* gene represent promising perspectives for future etiological management. [19] Other experimental strategies are also investigating modulation of inflammatory and angiogenic mechanisms involved in the progressive degradation of Bruch's membrane. [20]

Conclusion

Ophthalmologic complications of pseudoxanthoma elasticum represent a major cause of visual disability due to the high risk of choroidal neovascularization and macular fibrosis. Multimodal imaging allows early diagnosis and precise monitoring of lesion progression. Anti-VEGF therapy has considerably improved the visual prognosis of patients with PXE-associated CNV. Nevertheless, the absence of curative treatment justifies ongoing research focusing on anti-calcification therapies and gene therapy. Regular ophthalmologic follow-up remains essential to ensure early management of complications.

Declarations

Conflicts of Interest

The authors declare no conflicts of interest.

Authors' Contributions

All authors contributed to patient management, manuscript drafting, and revision. All authors approved the final version of the manuscript.

Patient Consent

Informed consent was obtained from the patient for publication of this case report and the associated images.

Funding: No funding was received for this work.

References

1. Uitto J, Bercovitch L, Terry SF, Terry PF. Pseudoxanthoma elasticum: progress in research toward treatment. Am J Med Genet C Semin Med Genet. 2010;148C [1]:56-63.
2. Neldner KH. Pseudoxanthoma elasticum. Clin Dermatol. 1988 ;6[1] :1-159.
3. Finger RP, Charbel Issa P, Ladewig M, Götting C, Szliska C, Scholl HP, *et al.,* Pseudoxanthoma elasticum: genetics, clinical manifestations and therapeutic approaches. Surv Ophthalmol. 2009;54[2]:272-285.
4. Gliem M, Zaeytijd JD, Finger RP, Holz FG, Leroy BP, Charbel Issa P. An update on the ocular phenotype in patients with pseudoxanthoma elasticum. Front Genet. 2013; 4:14.
5. Naouri M, Boisseau C, Bonicel P, Daudon P. Manifestations ophtalmologiques du pseudoxanthome élastique. J Fr Ophtalmol. 2011 ;34[9] :652-658.
6. Charbel Issa P, Finger RP, Götting C, Hendig D, Holz FG, Scholl HP. Centrifugal fundus abnormalities in pseudoxanthoma elasticum. Ophthalmology. 2010;117[7]:1406-1414.
7. Georgalas I, Papaconstantinou D, Koutsandrea C, Kalantzis G, Ladas I. Angioid streaks, clinical course, complications, and current therapeutic management. Ther Clin Risk Manag. 2009; 5:81-89.
8. Lebwohl M, Schwartz E, Lemlich G, Lovelace O, Shaikh-Bahai F, Fleischmajer R. Abnormal mineralization in pseudoxanthoma elasticum. Dermatologica. 1993;186[1]:33-37.
9. Finger RP, Charbel Issa P, Schmitz-Valckenberg S, Holz FG, Scholl HP. Long-term effectiveness of intravitreal bevacizumab for choroidal neovascularization secondary to angioid streaks in pseudoxanthoma elasticum. Retina. 2011;31[7]:1268-1278.
10. Gliem M, Fimmers R, Müller PL, Brinkmann CK, Finger RP, Holz FG, *et al.,* Choroidal neovascularization in pseudoxanthoma elasticum. Retina. 2020 ;40[11] :2138-2148.
11. Spaide RF. Enhanced depth imaging optical coherence tomography of pseudoxanthoma elasticum. Retina. 2012;32[2]:240-245.
12. Pece A, Avanza P, Galli L, Brancato R. Photodynamic therapy for choroidal

neovascularization associated with angioid streaks. Retina. 2005;25[7]:825-831.
13. Zweifel SA, Imamura Y, Freund KB. Treat and extend regimen with anti-VEGF agents in retinal diseases. Retina. 2011 ;31[3] :433-445.
14. Wiegand TW, Rogers AH, McCabe F, Reichel E, Duker JS. Intravitreal bevacizumab therapy for choroidal neovascularization associated with angioid streaks. Retina. 2009 ;29[1] :47-51.
15. Schaal S, Kaplan HJ, Tezel TH. Is there any role for photodynamic therapy in the anti-VEGF era? Ophthalmic Surg Lasers Imaging. 2008 ;39[6] :460-468.
16. Gliem M, Müller PL, Birtel J, Hendig D, Holz FG, Charbel Issa P. Quantitative fundus autofluorescence in pseudoxanthoma elasticum. Invest Ophthalmol Vis Sci. 2017 ;58[8] :3394-3399.
17. Finger RP, Charbel Issa P, Ladewig MS, Götting C, Szliska C, Holz FG, *et al.,* Pseudoxanthoma elasticum : genetics, clinical manifestations and therapeutic approaches. Surv Ophthalmol. 2009 ;54[2] :272-285.
18. Li Q, van de Wetering K, Uitto J. Pseudoxanthoma elasticum as a paradigm of heritable ectopic mineralization disorders. Curr Opin Pharmacol. 2019; 46:45-52.
19. Van Gils M, Willaert A, De Vilder E, Coucke PJ, Vanakker OM. Generation and analysis of a humanized mouse model for pseudoxanthoma elasticum. Matrix Biol. 2019; 80:51-63.
20. Hendig D, Schulz V, Eichgrün J, Szliska C, Götting C. New insights into pseudoxanthoma elasticum. Exp Dermatol. 2006 ;15[11] :851-860.