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Case Presentation

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Angioid Streaks and Pseudoxanthoma Elasticum with Severe Ocular Manifestations: A Case Report

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Abstract

Pseudoxanthoma elasticum (PXE) is a rare hereditary disorder characterized by progressive calcification and fragmentation of elastic fibers affecting multiple systems, including the skin, eyes, and cardiovascular system. We present the case of a 51-year-old Brazilian woman with angioid streaks and severe ocular manifestations. This case highlights the complexity of diagnosis and management strategies to prevent vision loss.

Keywords: Pseudoxanthoma elasticum; Angioid streaks; Choroidal neovascularization; OCT; Anti-VEGF; Ocular manifestations

Introduction

Pseudoxanthoma Elasticum (PXE) is a genetic condition affecting elastic tissue in various body parts, including the skin, eyes, and cardiovascular system. Angioid streaks are a common ocular finding in PXE and can lead to significant visual complications. This case report describes a patient with PXE and severe

ocular manifestations, emphasizing the importance of early diagnosis and appropriate management.

Case Presentation

A 51-year-old Brazilian woman with a history of hypothyroidism, vitiligo, dyslipidemia, and hypertension presented with vision loss. Her grandmother has a history of blindness, and her mother is undergoing anti-VEGF treatment. Clinical Findings: Best-corrected visual acuity was 20/200 in the right eye and light perception in the left eye. Intraocular pressure was 12 mmHg in the right eye and 10 mmHg in the left eye. Dermatological examination revealed typical PXE findings on the neck. Diagnostic Assessment: Optical coherence tomography (OCT) showed a discontinuity of Bruch's Membrane, thickening and irregularity of the RPE-Bruch's membrane complex and presence of a neovascular membrane. The right eye OCT, showed intraretinal cysts and subretinal fibrosis. The left eye exhibited fewer cysts. Fundus examination revealed bilateral angioid streaks. Therapeutic Intervention: Intravitreal injections of anti-VEGF were indicated for the right eye to manage choroidal neovascularization (CNV) and stabilize the ocular condition. The patient was advised to undergo regular ophthalmic follow-ups to monitor disease progression and adjust treatment as necessary. Follow-Up and Outcomes: Following the initiation of anti-VEGF therapy, the patient showed stabilization of visual acuity in the right eye. Further examinations will be necessary to assess the complete therapeutic response, including possible changes in the intraretinal cysts observed on OCT. No new CNV lesions were observed. The patient continues with regular follow-up for further treatment adjustments as needed **(Figure 1 and 2).**

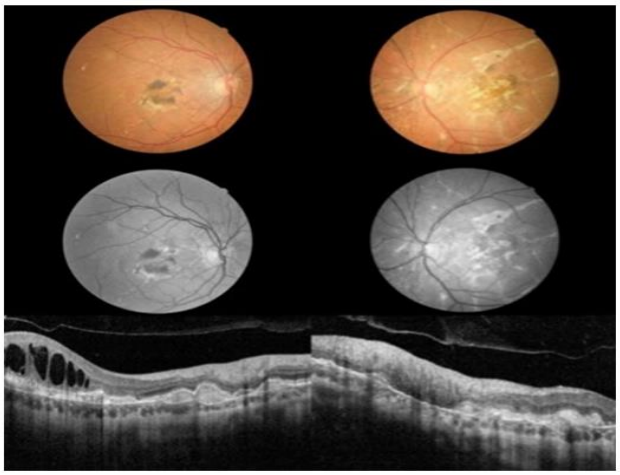

Figure 1: OCT: RE Intraretinal cysts and subretinal fibrosis. LE: Few cysts. Discontinuity of Bruch's Membrane, thickening and irregularity of the RPE-Bruch's membrane complex and presence of a neovascular membrane. Fundoscopy revealed bilateral angioid streaks.

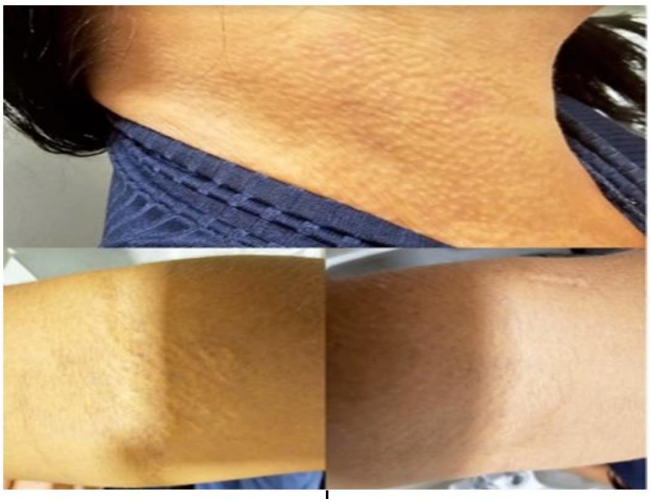

Figure 2: Pseudoxanthoma elasticum affecting the neck and forearm.

Discussion

PXE is a complex condition with significant ocular implications. Angioid streaks, resulting from breaks in Bruch's membrane, can lead to CNV and subsequent severe vision loss. Early recognition and timely intervention with anti-VEGF therapy are crucial for preserving vision. A multidisciplinary approach involving dermatologists, cardiologists, and geneticists is essential for comprehensive care **[1-5]**.

Conclusion

This case underscores the importance of vigilance in the ophthalmic evaluation of patients with PXE. Early detection and appropriate management of ocular manifestations are crucial for improving visual prognosis.

Ethical Statement

Informed consent has been provided by the patient for publication of this case report.

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