Angioid Streaks with Peau D'Orange in a Patient with Pseudoxanthoma Elasticum (PXE)

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Introduction

Angioid streaks are breaks in Bruch's membrane that appear as reddish-brown, jagged lines radiating outward from the peripapillary region.[1] Often bilateral, these streaks may increase in size over time.[1]

While optic disc drusen are commonly associated with angioid streaks, other fundoscopic findings may include atrophy, subretinal crystalline deposits, or peau d'orange.[1,2]

Patients with angioid streaks are usually asymptomatic but have a greater risk of developing choroidal neovascularization (CNV).[1,2] They are also more susceptible to choroidal rupture following ocular trauma.[2]

Case Presentation

A 29-year-old female with no history of any systemic diseases presented to the clinic for retinal evaluation of angioid streaks OU. She had no visual complaints and denied flashes, floaters, shadow, curtain, or veil. Medications include escitalopram 5 mg daily, and oral contraceptive. The patient denied smoking and has no pertinent family history. Review of systems was positive for hyperextensible joints.

• Visual Acuity	IOP
• OD: dsc 20/30, PH: 20/20-1	12
• OS: dsc 20/30-1, PH: 20/25	14

- External and anterior segment exams revealed no abnormalities OU.
- Posterior segment exam revealed RPE changes, angioid streaks, and peau d'orange appearance OU. Negative for CNV OU.

Imaging

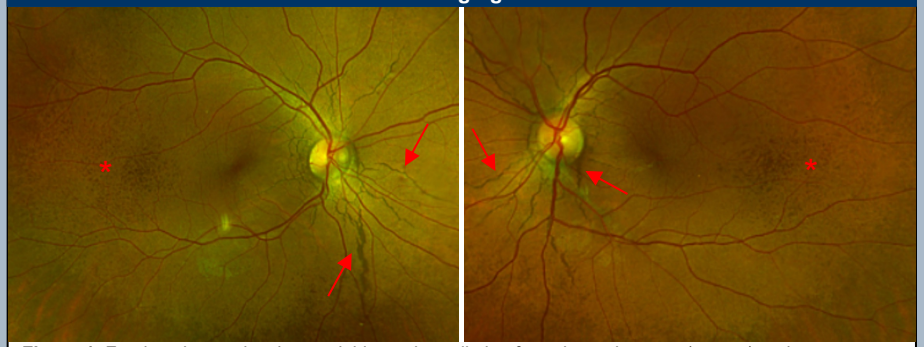

Figure 1. Fundus photos showing angioid streaks radiating from the optic nerve (arrows) and peau d'orange appearance of the retina temporally OU (asterisk).

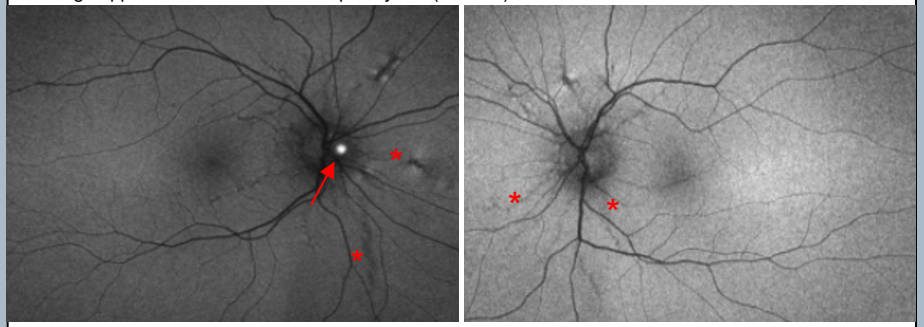

Figure 2. Fundus autofluorescence showing hyperautofluorescent optic disc drusen OD (arrow) and hypoautofluorescent angioid streaks OU (asterisks).

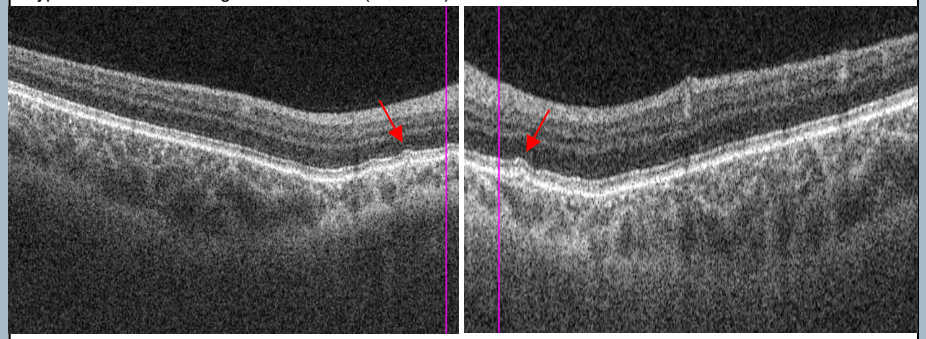

Figure 3. OCT showing breaks in Bruch's membrane (arrow) and no evidence of macular edema or subretinal fluid OU. Foveal Thickness: OD 266 μm, OS 273 μm.

Discussion

Although angioid streaks may be idiopathic, the most common systemic association is pseudoxanthoma elasticum (PXE) – an autosomal recessive disorder caused by mutations in the ABCC6 gene and characterized by mineralization and fragmentation of elastic fibers in connective tissue.[1-3] Other common systemic associations include Ehlers-Danlos syndrome, Paget's disease, and sickle cell anemia.[1-3]

The presence of angioid streaks with peau d'orange – yellow mottling of the retinal pigment epithelium (RPE) – are pathognomonic for PXE.[3,4] While not exclusive to PXE, the presence of disc drusen supported our suspicion of PXE.[3,4] As such, genetic testing was recommended. The results revealed two variants in the ABCC6 gene. Taken together, the diagnosis of PXE was established.

Additionally, observation was recommended because the patient was asymptomatic and negative for CNV. She was also advised to wear eye protection due to the increased risk for choroidal rupture and to follow up with her PCP to monitor for PXE-associated systemic complications.

Conclusion

The presence of angioid streaks should prompt one to perform a thorough medical workup and consider the possibility of underlying systemic disease.

Patients with angioid streaks should be monitored regularly for complications including CNV.

References

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