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Multimodal imaging in Angioid streaks with Pseudoxanthoma elasticum : a case report

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ABSTRACT

Angioid streaks refer to dehiscence in the Bruch's membrane- retinal pigment epithelium complex. It appear as dark to brownish streaks radiating from optic nerve on funduscopy. It is a rare occurrence and pseudoxanthoma elasticum is the most common systemic association (59%-87%). Angioid streaks lead to avoidable blindness if associated with choroidal neovascular membrane. We report a case of biopsy proven Pseudoxanthoma elasticum with angioid streaks in a young lady. It is not reported in our country till now.

Keywords: Angioid streaks; Autofluorescence; Choroidal neovascular membrane; Pseudoxanthoma elasticum.

INTRODUCTION

Angioid streaks (AS) is an associated fundus finding in a patient with pseudoxanthoma elasticum (PXE).It represent a break in the thickened and calcified Bruch's membrane in retina. Clinically it appear as dark red to brown bands of irregular lines which radiate from optic nerve head. It is almost always bilateral. Angioid streaks are usually asymptomatic unless associated with complications like choroidal neovascular membrane[1,2].

Its incidence in PXE is 59 % if PXE is diagnosed clinically. If PXE is diagnosed by skin biopsy then incidence of angioid streaks is increased by 87%[1]. We report a biopsy proven PXE with associated angioid streaks. Hence a simple fundus evaluation is needed to diagnose it and alert both physician and ophthalmologist for further interventions.

CASE REPORT

A 29 year old female presented at National Eye Centre (NEC) for fundus evaluation as she was diagnosed as Pseudoxanthoma Elasticum(PXE). Her past history revealed that she was under care of dermatologist for her skin issues. Initially she was diagnosed as post acne scarring. The patient was treated with 0.05 % tretinion cream and 10 % acetic acid cream locally. A fractional carbondioxide laser was also performed (2 episodes) after one month of first visit. Since there was no improvement,a punch biopsy was done on 26/10/2021 from neck region. The sample was sent for histopathological examination and report was compatible with PXE. At our NEC,the patient underwent thorough eye examination.The lid and adnexa were normal.Her best corrected visual acuity was 6/6 in both eyes.The intraocular pressure was 12 mm of Hg in both eyes. The anterior segment examination were unremarkable. Both eyes were dilated with 1% tropicamide for fundus evaluation.

On systemic examination, there was multiple pigmented papules in the neck. Rest examination were unremarkable (Figure 1).

On fundus examination, media was clear,normal optic disc with cup disc ratio of 0.3:1 and normal vessels in both eyes. The fovea was healthy and retina was attached. The patient underwent battery of investigations to confirm AS and its associated complications. Colored fundus photograph revealed dark to brown chorioretinal streaks radiating from optic disc extending to the posterior pole.The streaks were asymmetric in distribution in both eyes.It is more prominent in left eye than right eye (Figure 2).

The angioid streaks were more prominently seen in autofluorescence as hypoautofluorescent fissures (Figure 3).

Figure 1. Multiple pigmented papular lesions on the neck (black arrows)

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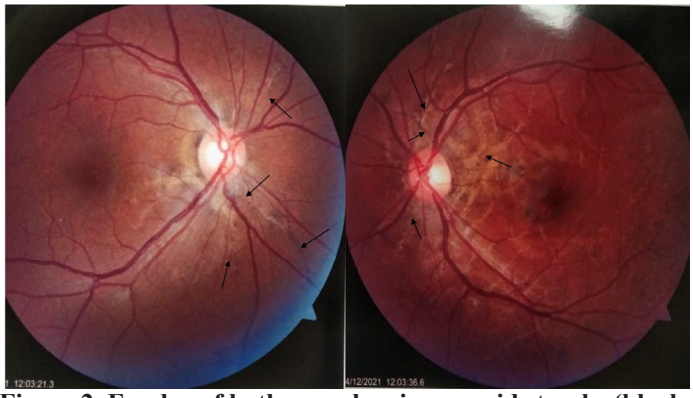

Figure 2. Fundus of both eyes showing angoid streaks (black arrows)

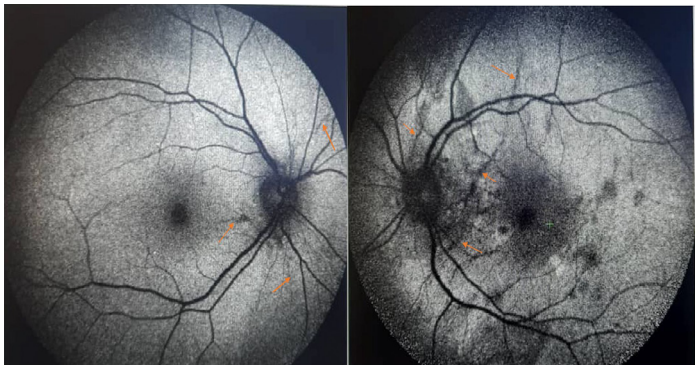

Figure 3. Autofluorescence of both eyes showing hypoautofluorescence (orange arrows)

However optical coherence tomography (OCT) showed no evidence of choroidal neovascular membrane (CNVM) in both eyes. This was further supported by optical coherence tomography angiography (OCTA) as there was no evidence of hyperfluorescent vascular networks in different slabs (Figure 4).

Hence a patient was diagnosed as both eyes AS without CNVM. Since the patient doesnot have any treatable retinal lesion, she was asked to follow up 3 monthly and advised to avoid any trauma to the eye.

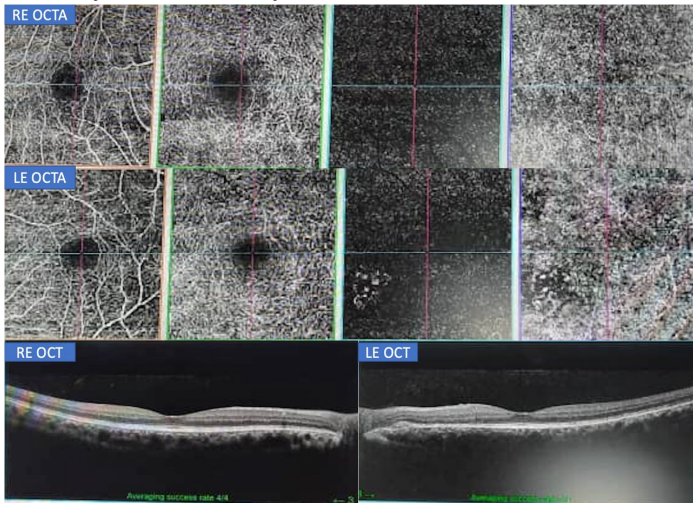

Figure 4. OCTA and OCT showing no evidence of CNVM

DISCUSSION

Angioid streaks associated with pseudoxanthoma elasticum is a rare clinical disease. It is crucial for patients diagnosed with pseudoxanthoma elasticum to undergo ocular examination as angioid streaks can lead to visual impairment. Angioid streaks alone does not cause visual impairment unless associated with complications like choroidal neovascular membrane, traumatic macular hemorrhage and macular degeneration[1-3]. It is an incidental finding unless there is involvement of macula. If macula is affected then patient presents with decreased vision, scotoma and metamorphopsia[1].

AS is associated with CNVM in 70% - 86% of cases 1 .CNVM leads to exudation and macular hemorrhage beneath the neurosensory retina leading to photoreceptor damage[4]. Later it will lead to severe visual impairment due to pigment epithelial detachment, subretinal fibrosis and atrophy.

Routine funduscopy in a patient with pseudoxanthoma elasticum, paget's disease, sickle cell anaemia, Ehlers- Danlos syndrome and marfan syndrome is mandatory to look for AS. Similarly, if AS is an incidental finding during funduscopy then we have to refer to physician for evaluation for aforementioned systemic conditions.

We report a case of pseudoxanthoma elasticum (PXE) associated with angioid streaks (AS) in a young female patient. It was incidental finding when she was undergoing ocular screening for skin lesion. She was diagnosed as PXE by dermatologist. It was proven by skin biopsy. The diagnosis is further confirmed by presence of AS in retina.

Multimodal imaging plays an adjunctive role in the diagnosis of AS and its complications. Coloured Fundus photograph is essential for documentation and monitoring of progress of disease. It also helps to identify as better as small lesion can be missed during indirect ophthalmoscopy. Autofluorescence is an important imaging modality as subtle lesion appear more prominent than in coloured fundus photograph. AS appear as hypoautoflourescent Figure 5. OCT and OCTA helps us to identify and confirm CNVM. In OCT, there will be subretinal hyperreflective material suggestive of type 2 CNVM 6 . In OCTA, CNVM is seen as hyperreflective vascular network at the level of outer retina and choriocapillaris layer 7. However our patient did not have any evidence of CNVM. Hence no treatment was initiated.

Anti-vascular endothelial growth factors (Anti-VEGFs) are available and they have been proven to be effective in the treatment of CNVM associated with AS. In our country Bevacizumab is the only Anti-VEGF available and it has promising result as with other Anti-VEGFs[8-10].

In our case, patient did not have any treatable lesion right now. Hence she was asked to follow up every 3 monthly or as and when deemed necessary. During follow up we have to monitor for visual acuity as patient can have decreased vision and or metamorphopsia due to development of CNVM. However there are several risk factors for development of CNVM. These

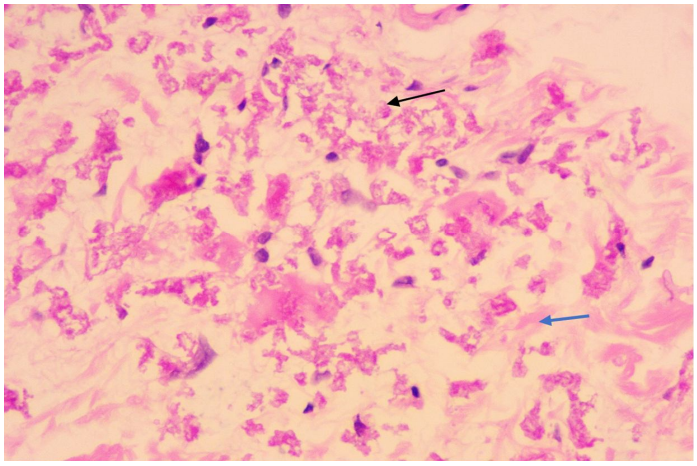

Figure 5. Histopathological report H&E section: X400
Microscopic description: *Sections show a skin with unremarkable epidermis. Dermis show affected elastic fibers appearing as widely dispersed granular material amidst normal collagen fibers with a bizarre appearance reminiscent of a bishops crook. Abnormal fibers are bright pink with an altered architecture, as they have lost their normal interlacing pattern (black arrow).Normal collagen fibres (blue arrow)*

are elderly population, width, length and location of angioid streaks. More is the width and length of AS higher the risk for CNVM. Similarly AS located within one disc diameter from the optic disc has higher risk of CNVM[1,11]. Therefore follow up needs to be customized depending on risk factors .

CONCLUSIONS

Angioid streaks is a blinding condition if associated with choroidal neovascular membrane. A patient of pseudoxanthoma elasticum needs ocular screening for angioid streaks or vice-versa. Angioid streaks can be seen in systemic conditions like pseudoxanthoma elasticum, thalassemia, sickle cell anaemia and Ehlers-Danlos syndrome. Skin biopsy is gold standard for diagnosis of pseudoxanthoma elasticum. If facility for biopsy is not available then cutaneous manifestation (yellowish papules in the nape , side of neck and in flexural areas followed by plaque formation and wrinkling of skin) will help in the diagnosis of pseudoxanthoma elasticum. Close follow up is needed as there is high incidence of development of CNVM. Therefore timely cross consultation between ophthalmologist and physician is essential to prevent needless blindness. In addition these patients should avoid blunt trauma to eyes as even trivial trauma can lead to traumatic retinal pigment epithelium -Bruch's membrane complex rupture.

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