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AN UNFORTUNATE STREAK – CHOROIDAL NEOVASCULARIZATION REVEALING A GENETIC DISEASE

Ophthalmology

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ABSTRACT

Purpose: To emphasize the importance of a thorough fundoscopic and physical examination in a young patient with a new choroidal neovascularization (CNV). **Observations:** We report a young patient with a new CNV in her right eye, developed upon an angioid streak. Fundoscopic examination revealed peau d'orange and angioid streaks, a combination that is pathognomonic of a relatively rare genetic disease – Pseudoxanthoma Elasticum (PXE), which has important dermatologic and cardiac manifestations. **Conclusions:** A new CNV in a young non-myopic patient should raise suspicion of a specific underlying etiology. Identifying the pathognomonic retinal features of PXE allowed prompt diagnosis, appropriate treatment and further systemic workup.

KEYWORDS

Pseudoxanthoma Elasticum (PXE), Angioid Streaks, Choroidal Neovascularization (CNV)

INTRODUCTION

Pseudoxanthoma Elasticum (PXE) is a rare heritable multi-system disorder, characterized by aberrant mineralization and fragmentation of elastic fibers, involving primarily the skin, eyes and cardiovascular system. The ocular manifestations of PXE involve the retina. Peau d'orange (pigmented spots seen as yellowish mottling) represents a progressive calcification of Bruch's membrane which is composed primarily of elastic fibers. Angioid streaks (narrow irregular lines radiating from the optic disc towards the periphery), represent breaks of the calcified and thickened Bruch's membrane, and may be associated with choroidal neovascularization (CNV) and compromised vision. Timely diagnosis of PXE is important in order to achieve improved visual outcomes and anticipate cardiovascular complications.

Case Report

A healthy 45-year-old non-myopic Arab woman was referred to the hospital's retina clinic for assessment of decreased visual acuity and suspected retinal hemorrhage. Funduscopic examination showed macular subretinal fluid and hemorrhage in her right eye, as well as choroidal neovascularization (CNV) which was confirmed by optic coherence tomography (OCT) and angiography (OCT-A). A more thorough fundoscopic examination revealed angioid streaks - irregular and jagged brownish-grey lines radiating from a concentric peripapillary ring into the periphery, as well as Peau d'orange - pigment small dark spots resulting in a mottled aspect prominently of the temporal retinal mid-periphery (Figure 1). The CNV developed upon an angioid streak.

The patient's hair and neck were hidden by the hijab, traditional head covering worn by Muslim women. When gently persuaded to remove it, her neck examination was immediately diagnostic of pseudoxanthoma elasticum, revealing a lax, wrinkled and redundant skin in the neck, consisting of large plaques presenting in a reticular pattern. The characteristic papules produce a 'peau d'orange' appearance (Figure 2). The diagnosis of Pseudoxanthoma Elasticum (PXE) was established. The cardiovascular system was not involved. The patient was treated with three monthly intravitreal anti-vascular endothelial growth factor (anti-VEGF) injections to the right eye. A fundoscopic examination of her siblings revealed the same clinical picture in one of them, and an annual follow up was recommended. Our patient's follow-up 4 months later, showed a complete resolution of the macular CNV in her right eye. Only signs of PXE were seen in both eyes (Figure 3).

DISCUSSION

A new CNV in a young non-myopic patient should raise suspicion of an underlying etiology. A thorough fundoscopic and physical examination may reveal signs of relatively rare disorders, such as in our case. Some disorders have potential systemic complications, hence the importance of prompt diagnosis and further workup. CNV of the macular region is a frequent complication in patients with PXE, usually occurring in association with angioid streaks. Most cases respond well to a single round of three monthly intravitreal anti-VEGF injections, with complete resolution of CNV and significant improvement in visual acuity.

Pseudoxanthoma elasticum (PXE) is a rare autosomal recessive disorder now known to be caused by mutations in the ABCC6 gene, important in adenosine triphosphate (ATP) transport and thus, inorganic pyrophosphate generation - a potent anti-mineralization factor. Due to its reduced levels, mineralization evolves in elastin-rich tissues – especially the skin, eyes, and arterial blood vessels (1). Slow elastic fiber calcification results in late-onset presentation which is highly variable. Skin (pseudoxanthomatous papules on the neck and flexural skin creases causing lax, redundant appearance) and eye involvement (as in our patient) are most characteristic. However, diminished peripheral pulses; renovascular hypertension, premature coronary artery and cerebrovascular disease may also become prominent, due to calcification of dystrophic elastic fibers in the wall of medium- and small-sized arteries. Histology, genetic testing, and identification of an affected first-degree relative confirm the diagnosis. Macular neovascularization, her prominent presenting feature, may have myriad causes (2). However, the well-known dictum “you see what you know, you find what you look for” proved right again. The appearance of the neck, a 'once seen – never forgotten' picture (Fig. 2) firmly established the diagnosis, reminding us again that looking at the whole patient, even parts hidden by the hijab, remains an essential part of diagnostic success (3).

Legends to the figures

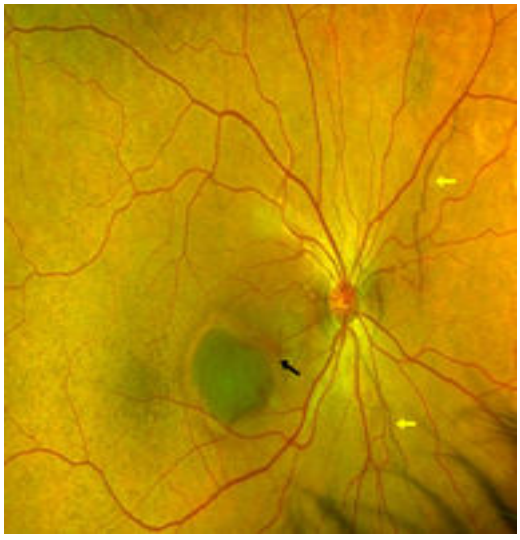

Fig. 1. Optos wide-field fundus examination of the right eye showing macular neovascularization (black arrow), and numerous narrow irregular angioid streaks radiating towards the periphery (arrows). Pigmented spots of peau d'orange can also be seen.

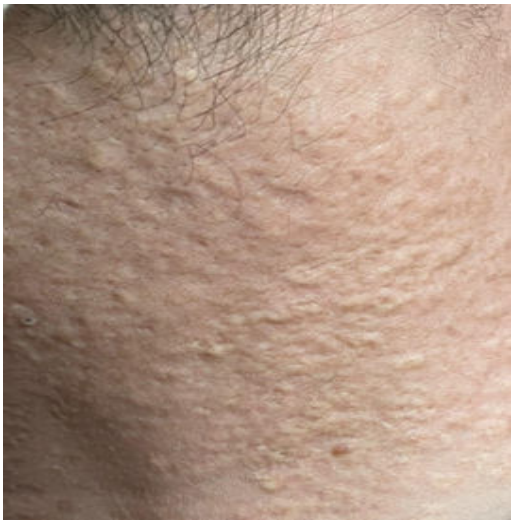

Figure 2: Color photography of the patient's neck showing lax, wrinkled and redundant skin. The pseudoxanthomatous papules on the patient's neck (also common on flexural skin creases) cause the lax, redundant appearance characteristic of pseudoxanthoma elasticum

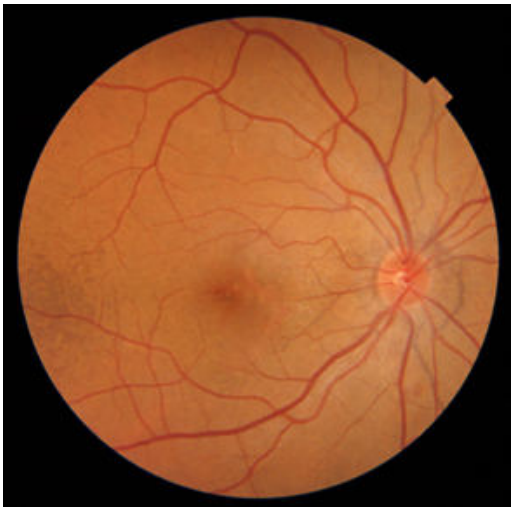

Figure 3: Color fundus photography showing bilateral angioid streaks and peau d'orange.

Brief Summary:

Choroidal neovascularization discovered in a young non-myopic patient revealed a relatively rare genetic disease – Pseudoxanthoma Elasticum. Identifying its ocular retinal features allowed prompt diagnosis, appropriate treatment and further systemic workup.

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