

Granulomatous Anterior Uveitis & Pseudoxanthoma Elasticum: What Association?

Dr Houda Ahammou¹, Dr Fatima Belarbi², Dr Nabil Albab¹, Pr Said Amal², Pr Ibtissam Hajji¹, Pr Nadia Akhdari², Pr Ouafa Hocar², Pr Abdeljalil Moutaouakil¹

¹Ophthalmologue, UHC Mohammed VI, Marrakesh, Morocco

²Dermatologue, UHC Mohammed VI, Marrakesh, Morocco

***Corresponding author**

Dr Houda Ahammou

Email: houdaahammou@gmail.com

Abstract: Pseudoxanthoma elasticum (PXE) is a hereditary disease characterized by skin, ocular, and cardiovascular lesions. The main ocular feature of PXE is the presence of angioid streaks in the retina. Methods: We report the case of a patient with PXE. The diagnosis of PXE was made by the presence of cutaneous lesions typically. Results: For our patient, the ophthalmological manifestations of PXE were the angioid streaks and “peau d’orange” without choroidal neovascularization. The particularity of our case was the onset of Granulomatous anterior uveitis. Conclusions: Is this an unusual manifestation of PXE or just a coincidence? Only a systematic and regular ophthalmological examination of new PXE cases can answer this question.

Keywords: Pseudoxanthoma elasticum, Granulomatous uveitis, physiopathology

INTRODUCTION:

Pseudoxanthoma elasticum (PXE) is an autosomal recessive hereditary genetic disorder linked to a mutation in the ABCC6 gene that encodes a carrier protein present in the liver and kidney and responsible for impairment of the elastic connective tissue [1]. It mainly affects the skin, eyes, arteries and heart. The skin manifestations of PXE are the most specific and evocative signs. The diagnosis is almost always retained in front of the cutaneous lesion, rarely in case of an ophthalmological injury or during a vascular complication [1, 2]. The ocular lesion, observed in the case of PXE, is due to rupture of the Bruch membrane leading to the appearance of angioid streaks and complications (choroidal hemorrhage, retinal neovascularization, etc.). These abnormalities are not pathognomonic of PXE. They are observed in 90% of cases [3].

The aim of our work is to report a particular case of a girl followed up for PXE, who presents retinal lesions, characteristic of PXE associated with anterior granulomatous uveitis. An association between pseudoxanthoma elasticum and uveitis has never been described. Is this a coincidence? Or an unusual PXE manifestation?

CASE REPORT:

We report a case of a 26-year-old patient, followed in dermatology for a pseudoxanthoma

elasticum retained in front of the histological data and the specific cutaneous lesion, characterized by the presence of asymptomatic confluent yellowish papules with smooth surface, firm consistency, seated at the level of the large folds (FIG. 1a, 1b). The cardiovascular examination with electrocardiogram and cardiac ultrasound were not abnormal, but ophthalmologic examination showed angioid streaks with an orange-peel “peau d’orange” appearance of the fundus of the eye at the level (Figure 2) without choroidal neovascularization (Figure 3) and visual acuity at 10/10 ODG. A month later, the patient presented for visual fog in the left eye. Slit-lamp examination showed pigmented retro-descemetica (PRD) precipitates, central sheep fat (Figure 4) with a minimal Tyndall in the anterior chamber and a visual acuity reduced to 6/10 in the left eye. Examination of the right eye did not demonstrate signs of uveitis. Moreover, the patient showed no extraocular clinical signs other than the cutaneous signs of PXE. In the case of anterior uveitis, non-synthetic non-hypertensive unilateral granulomatous without heterochromia, we performed a clinical and paraclinical etiological assessment, which was normal, with a sarcoidosis, tuberculosis, Lyme serology, syphilitic serology, herpes. After eliminating an infectious cause, we put the patient under local corticosteroid therapy. After a one-year follow-up, the evolution was marked by the disappearance of Tyndall, persistence of (PRD) and improvement of visual acuity.



Fig-1: Picture, of asymptomatic confluent yellowish papules, smooth surface, firm consistency, sitting at neck (a) and underarms (b)

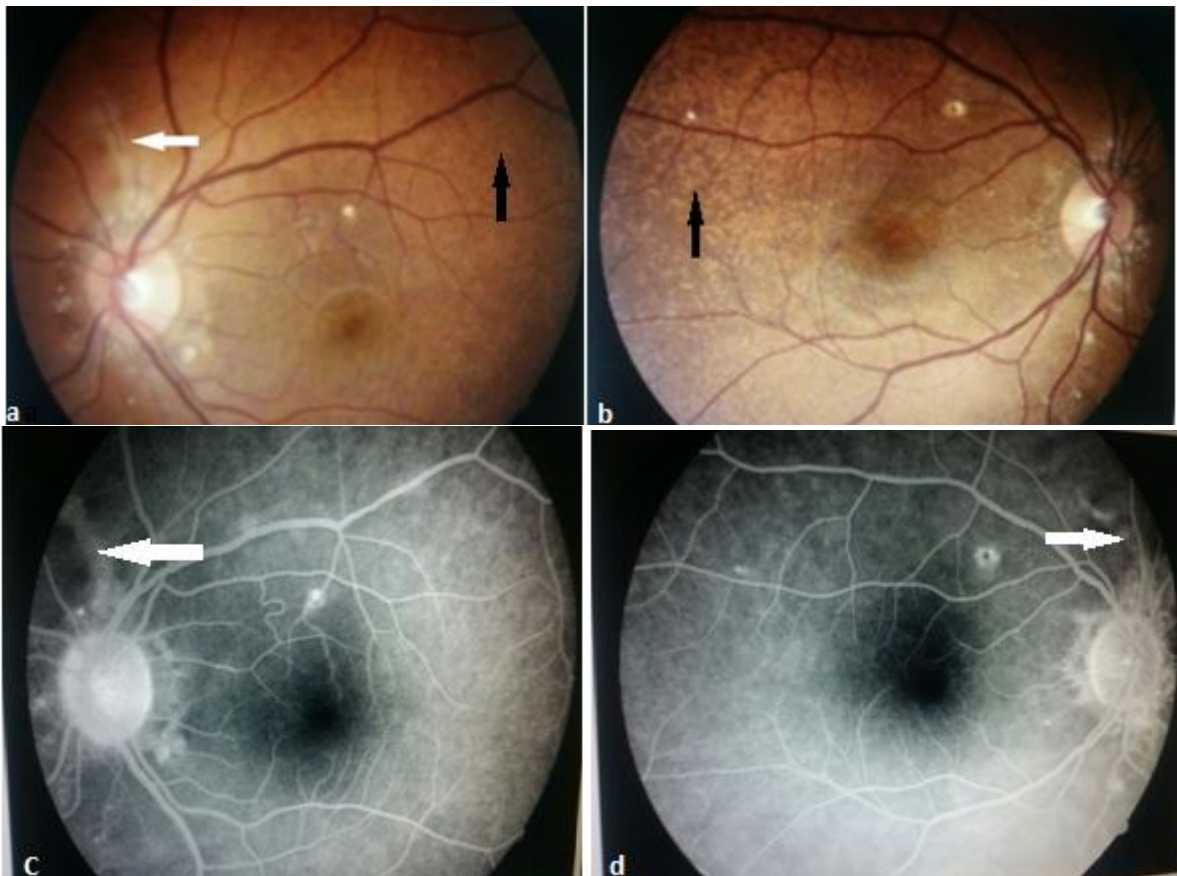


Fig-2: Color photographs (a, b), in fluorescein angiography (c, d) (black arrows: orange peel, white arrows: angioid streak).

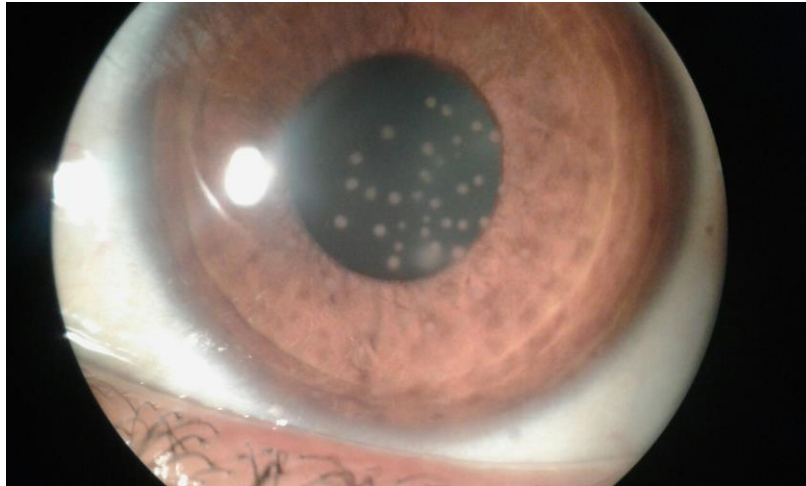


Fig-3: Picture, of central retrodescentic precipitates, pigmented sheep fat.

DISCUSSION

Pseudoxanthoma elasticum (PXE) or systematized elastorrhexis is a rare inherited disorder of the elastic connective tissue due to a mutation in the ABCC6 (ATP-binding-cassette) gene of the chromosome 16p13.1 with considerable phenotypic variability [4]. The pathophysiology of PXE is particular. It combines fragmentation, mineralization and degeneration of the elastic tissue, collagen changes and abnormal amounts of proteoglycans [1, 4].

The ocular lesion in this disease is due to an attack of the elastic fibers contained in Bruch's blade, resulting in the appearance of strictly retinal lesions. The characteristic ocular lesions correspond to angioid streaks. They are present in 80% of PXE cases [3,5,6]. Although not pathognomonic, their presence is of great diagnostic value for PXE. The lesion that first appears is an "orange peel" appearance in the fundus [6]. Calcification in the connective tissue may lead to retinal hemorrhage, neovascularization and loss of central vision [3, 6]. For our patient, we retained the diagnosis of PXE following the presence of angioid streaks with an orange peel aspect and specific cutaneous lesion.

Our case is particular, due to the unusual combination of pseudoxanthomaelasticum (PXE) and anterior granulomatous uveitis. There is currently no similar case in the literature. The pathophysiology of uveitis (inflammatory origin) is totally different from that of PXE. In our case, could granulomatous uveitis be explained by inflammatory mechanisms secondary to ocular lesion in PXE? Or is it merely a coincidence and a fortuitous association of two pathologies of different physiopathological mechanisms, but which has never been reported in the literature?

CONCLUSION

The particularity of our observation lies in the rarity of this pathology and especially in this unusual association with anterior granulomatous uveitis. The pathophysiology of ocular lesion in PXE is different

from that of granulomatous uveitis. Is this a new manifestation of the PXE or of a fortuitous association?

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