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Complementary angiographic and autofluorescence findings in pseudoxanthoma elasticum

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Abstract

Pseudoxanthoma elasticum (PXE) is a systemic disease with characteristic findings on fundus examination. The fundus findings may be difficult to detect with ophthalmoscopy. A case report is described as follows. A PXE patient had subtle retinal findings on fundoscopy that were more prominently seen using a combination of both fundus autofluorescence (FAF) imaging and indocyanine green (ICG) angiography. The fundus features visualized using each of these two modalities appeared different from each other. FAF imaging and ICG angiography may be able to more prominently detect pathology at the level of the retinal pigment epithelium and Bruch's membrane, respectively. The use of these imaging modalities together may be complementary and useful in the evaluation of patients with PXE.

Keywords

Angiography; Autofluorescence; Fluorescein; Indocyanine green; *Pseudoxanthoma elasticum*

Introduction

Pseudoxanthoma elasticum (PXE) is a systemic disorder affecting connective tissue. A constellation of retinal findings can be seen, including angioid streaks, peau d'orange, and

macular pigment mottling. Occasionally, the ocular features of PXE can be subtle and not easily identifiable on clinical examination. Indocyanine green angiography (ICG) and fundus autofluorescence (FAF) imaging have both been utilized separately to reveal the retinal features of PXE. However, the use of both modalities together has not previously been described in the setting of PXE.

Case report

A 62-year-old male patient with PXE was seen for routine ophthalmic examination. Best-corrected visual acuity was 20/25 in the right eye and 20/32 in the left eye. Anterior segment examination and intraocular pressures were within normal limits. Fundus examination revealed retinal pigment epithelial (RPE) mottling in the macular and peripapillary regions along with mild peripapillary atrophy (Fig. 1a, b). The late phase of the fluorescein angiogram (FA) showed changes (window defects, blocking) consistent with the pigment mottling (Fig. 1c, d). Angioid streaks and peau d'orange changes were not evident on fundoscopy or FA.

FAF images were obtained with a confocal laser scanning ophthalmoscope (Heidelberg Retina Angiograph, HRA2) using 480-nm excitation and 500-nm barrier filters, and a conventional fundus camera with 580-nm excitation and 700-nm barrier filters. FAF imaging showed distinctive areas of autofluorescent changes that were correlated with areas of pigment mottling (Fig. 2a–d). FAF imaging showed the areas of macular RPE mottling more extensively than were seen on clinical exam and FA. FAF imaging also demonstrated angioid streaks better than clinical exam. ICG angiography in the late phase showed extensive angioid streaks, along with peau d'orange in the temporal macula (Fig. 2e, f). Visualization of angioid streaks and peau d'orange was superior on ICG angiography as compared to FA and FAF imaging.

Discussion

Ocular manifestations of PXE consist of changes in the fundus that include angioid streaks and peau d'orange, which are hypothesized to originate from degeneration of Brush's membrane [1,2]. However, these changes are not always clearly visualized on fundoscopy or FA. It has been demonstrated that ICG angiography outlines angioid streaks as well as peau d'orange better than FA in the majority of cases [3]. Consistent with this report, we found ICG angiography to reveal angioid streaks and peau d'orange better than FA in our patient.

FAF imaging relies on light emission from lipo-fuscin in RPE cells and is thought to reflect the metabolic activity of the RPE [4]. FAF imaging can demonstrate altered metabolic activity within the RPE that may not be seen on fundoscopy or FA. FAF imaging in patients with PXE has been shown to demonstrate more widespread areas of RPE disturbance, particularly angioid streaks, than what is detectable by fundoscopy or FA [5].

FAF imaging and ICG angiography findings have not previously been compared together in patients with PXE. Our case, with FAF highlighting RPE pigment mottling and with ICG angiography delineating the peau d'orange and angioid streaks, suggests that ICG angiography may be more specific for structural changes at the level of Bruch's membrane, while FAF imaging allows for visualization of metabolic changes at the level of the RPE. Since fundus changes in PXE may reflect different disease patterns at the level of the RPE and Bruch's membrane, combined imaging with both ICG angiography and FAF imaging may allow for better visualization of retinal disease in patients with PXE.

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Fig. 1.
Fundus photography (**a, b**) and late phase fluorescein angiography (**c, d**) of the right and left eye

Fig. 2. Fundus autofluorescence imaging obtained using confocal laser scanning ophthalmoscopy (**a, b**) and conventional fundus photography (**c, d**). Images from late phase indocyanine green angiography are also shown (**e, f**)