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Non-vascular vision loss in pseudoxanthoma elasticum

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Abstract

Pseudoxanthoma elasticum patients with angioid streaks are well-known to have acute vision loss due to choroidal bleeding. However, chronic vision loss due to macular atrophy is less well characterized. We describe a patient with sub-acute vision loss in one eye due to loss of macular retinal pigment epithelium function. Autofluorescence and pattern electroretinogram were useful adjuncts to help diagnose the source of her vision loss.

Keywords

Pseudoxanthoma elasticum; Autofluorescence; Pattern electroretinogram; Maculopathy; Angioid streaks

Introduction

Pseudoxanthoma elasticum (PXE) [OMIM #264800] is an autosomal recessive systemic connective tissue disorder involving elastic fiber calcification and fragmentation with major clinical manifestations occurring in the ocular, cutaneous, and cardiovascular systems [1]. It is due to mutations in the *ABCC6* gene and its prevalence is estimated to be as high as one in 70,000 [2,3]. Choroidal neovascularization (CNV) with subsequent leakage and bleeding is one of the most common causes of acute vision loss in PXE patients with angioid streaks.

Case

A 65-year-old woman with skin biopsy-proven PXE complained of gradual vision loss for 2 months in her left eye with metamorphopsia. Visual acuity was 20/20 in the right eye and 20/40 in the left eye. Fundus examination showed tapering angioid streaks radiating from the optic nerve in both eyes (Fig. 1). Absence of choroidal neovascularization was confirmed by fluorescein.

Autofluorescence revealed extensive retinal pigment epithelium (RPE) atrophy with surrounding hyperfluorescence in both eyes and an island of preserved macula in her right eye (Fig. 1c, d). International Society for Clinical Electrophysiology of Vision (ISCEV) standardized full-field electroretinogram (ERG) was normal in each eye. Pattern electroretinogram (PERG) P50 and N95 components were normal in the right eye; the left eye had a significantly decreased P50 with a normal N95/P50 ratio (Fig. 2).

Comment

This is a patient with an established diagnosis of PXE and striking funduscopy and autofluorescence. In a patient with hyperfluorescent specks and RPE atrophy without a clear diagnosis, it is important to distinguish Stargardt disease from PXE macular atrophy [4]. Autofluorescence is beneficial to visualize peripapillary sparing of RPE cells, a unique and previously well-described feature of Stargardt patients [5,6].

In patients with angioid streaks, choroidal neovascularization and traumatic choroidal rupture are among the most well studied reasons for visual loss. However, central vision loss may also occur from progressive central atrophy of the RPE. This chronic form of vision loss in PXE is poorly characterized [2] and it has not been emphasized in the literature.

We hypothesize that our patient experienced subacute vision loss in her left eye due to a “snuff-out” phenomenon of her macular RPE. Her right eye remains 20/20 due to an island of preserved RPE, as detected by autofluorescence. The disparity in visual function between her two eyes is corroborated by her PERG which shows decreased P50 amplitude in her left eye only. The normal N95/P50 is inconsistent with vision loss secondary to optic neuropathy.

In conclusion, this case illustrates sub-acute vision loss in PXE due to RPE atrophy which was diagnosed with the help of autofluorescence. It serves to emphasize that the combination of autofluorescence imaging [7] and functional testing with PERG play an important role in elucidating the etiology of vision loss and monitoring in individuals with macular disease.

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Fig. 1.

(a, b) Color fundus photographs of the right and left maculae, showing central atrophy with tapering angioid streaks. (c, d) Fundus autofluorescence was obtained by illuminating the fundus with argon laser light (488 nm) and viewing the resultant fluorescence through a band pass filter with a short wavelength cut off at 495 nm. Right eye shows loss of RPE adjacent to the optic nerve and in the macula with an island of functioning cells. Left eye also has loss of RPE adjacent to the optic nerve with more extensive loss of macula cells. Note the hyperfluorescent specks surrounding these areas of atrophy

Fig. 2. PERGs recorded using Dawson, Trick, and Litzkow electrodes according to International Society for Clinical Electrophysiology of Vision (ISCEV) guidelines. **(a)** PERG right eye. P50 = 2 microvolts (normal ≥ 2 mv); N95/P50 = 2 (normal > 1). **(b)** PERG left eye. P50 = 0.8 (normal ≥ 2); N95/P50 = 2.5 (normal > 1)