



PURPOSE

To report a case of a patient diagnosed with Pseudoxanthoma elasticum (PXE).

METHODOLOGY

Descriptive Observational Study.

RESULTS

An otherwise healthy 31-year-old man presented with decreased visual acuity (VA) three months after trauma to the left eye (LE). Best-corrected VA was 20/20 in the right eye and counting fingers at 2 meters in the LE. Biomicroscopy of the anterior chamber and tonometry were normal. Fundus examination (Image 1) revealed linear, brown-grayish, irregular lesions arranged radially from the optic nerve in both eyes, suggestive of of angioid streaks (AS); associated with choroidal rupture in the LE.

On physical examination, yellowish papules were observed as plaques in the cervical region (Image 2). Fluorescein angiography confirmed AS in both eyes, and pre-retinal hemorrhage in the LE. Optical coherence tomography did not find choroidal neovascularization.

In December 2023, skin biopsy findings confirmed the diagnosis of PXE. The patient maintains ophthalmological, cardiological and dermatological follow-up.

DISCUSSION

PXE is a rare multisystem disorder causing the progressive degeneration and calcification of elastic fibers, with manifestation in dermatology, ophthalmology and cardiovascular systems that may lead to early death.

The appearance of typical dermatological lesions is an early sign of PXE and could assist in preventing ophthalmological and cardiovascular complications.

Reduced VA can occur if there is macular involvement by AS or when complications arise, such as choroidal neovascularization and hemorrhages. In ocular trauma, choroidal rupture can occur leading to acute symptoms.

Although the ophthalmological treatment can be proven challenging, better impacts can be achieved in cardiovascular aspects. Therefore, early screening conducted by a cardiologist is crucial, for these complications most affect the prognosis and life expectancy of such patients.



Image 1: Binocular indirect ophthalmoscopy showing AS (arrow)



Image 2: Dermatological lesions on the neck

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