Development of angioid streaks in association with pseudoxanthoma elasticum: report of 5 cases

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**PURPOSE**

- Angioid streaks are visible irregular crack-like dehiscences in an abnormal and calcified Bruch’s membrane that appear as dark or reddish streaks originating from the optic disc, almost always bilaterally.
- Angioid streaks have been commonly associated with systemic conditions such as pseudoxanthoma elasticum (PXE), Paget’s disease, Marfan syndrome, Ehlers-Danlos syndrome and blood dyscrasias, but also appear in patients without any systemic disease.
- Patients are generally asymptomatic, unless the lesions extend towards the fovea or develop complications such as traumatic Bruch’s membrane rupture or macular choroidal neovascularization (CNV).
- PXE is by far the most common association of angioid streaks. PXE is a rare hereditary disorder of connective tissue in which there is progressive calcification, fragmentation and degeneration of elastic fibers in the skin, eye and cardiovascular system. Ocular involvement is present in 85% of the cases, usually after 20 years of age.
- The authors present the cases of 5 patients with angioid streaks secondary to PXE.

**RESULTS**

**CASE 1:**
- Women, 51 years old. Diagnosis of PXE. Without relevant family history.
- Observed in our Department (4 years ago) for screening of diabetic retinopathy.
- Best corrected visual acuity (BCVA) of 10/10 OD and 10/10 OS. Normal biomicroscopy.
- Fundoscopy and Angiography: bilateral angioid streaks.

**CASE 2:**
- Women, 37 years. Without relevant family history.
- Went to our Emergency Department with complaints of bilateral visual acuity decrease.
- BCVA 10/10 OD and 10/10 OS. Normal biomicroscopy.
- Fundoscopy and angiography showed presence of bilateral angioid streaks, without foveal involvement.
- Presence of skin lesions in the neck compatible with the diagnosis of PXE. Submitted to skin biopsy with histological findings compatible with the diagnosis of PXE.
- Genetic analysis of the gene ABC6 showed the mutation c.3421C>T (p.Arg1141*) and the variation c.887G>A (p.Trp296*) in heterozygosity, confirming PXE.

**CASE 3:**
- Women, 44 years. Without relevant family history.
- Diagnosis of PXE 20 years ago.
- Complaints of significant visual acuity decrease in OS.
- Ophthalmologic exam:
  - BCVA 10/10 OD and 1/20 OS.
  - Biomicroscopy: normal
  - Fundoscopy: bilateral angioid streaks, with foveal involvement and maculopathy in the left eye.
  - Angiography: Subfoveal CNV OS.
- Underwent 4 sessions of photodynamic therapy (PDT) with BCVA improvement to 1/10 OS.

**CASE 4:**
- Women, 35 years. Without relevant family history.
- At 15 years of age refers ocular pain, visual acuity decrease and headaches. BCVA of 10/10 bilaterally. At fundoscopy showed drusen with 2 disc diameters, perivascular, white and refringent.
- Diagnosis of PXE at the age of 25 years:
  - Angiography: bilateral angioid streaks, without foveal involvement.
  - BCVA of 3/10 OD and 1/10 OS, normal biomicroscopy, fundoscopy and angiography showed angioid streaks with foveal involvement.
  - BVA 10/10 OD and 9/10 OS until recently.

**CASE 5:**
- Women, 50 years. Diagnosis of PXE. Without relevant family history.
- At first observation in our Department (8 years ago), presented with BCVA of 3/10 OD and 1/10 OS, normal biomicroscopy, fundoscopy and angiography showed angioid streaks with foveal involvement.
- Later developed CNV and was treated with PDT with poor outcome: BCVA of 1/10 OD and counting fingers OS.

**CONCLUSIONS**

Angioid streaks can be associated with multiple systemic diseases and ophthalmologists must be aware of this condition and provide rapid treatment in case of ophthalmological complications. We also have to guide patients to a diagnostic survey when none of the associated systemic disease is known.