Visual loss associated with angioid streaks in sickle thalassemia

Haematologica 2005; 90:(1)e4-e5

Angioid streaks (AS) represent ruptures of the elastic lamina of Bruch's membrane and are typically seen in the fundus of patients with pseudoxanthoma elasticum (PXE), a rare inherited elastic tissue defect. The development of AS in hemoglobinopathies is not a novel observation, as it has been reported in sickle-cell disease since the late 1950s.[1] Despite however the frequent presence of AS in these patients (22% in sickle-cell anemia, 20% in ,-thalassemia and 10% in sickle thalassemia), only recently it became clear that AS are a part of an underlying diffuse elastic tissue defect, which characterizes inherited hemoglobinopathies.[2] This syndrome, altough acquired, is clinically and histopathological identical to hereditary PXE.[3,4] Typical subclinical pathology findings are present since childhood, while the clinical manifestations are age-related and their prevalence reaches 85% in hemoglobinopathy patients older than 30 years.[2,5]

We present here the case of a 45-year-old female patient with sickle thalassemia, who presented with severe unilateral visual acuity impairment due to AS. The patient had experienced only mild and rare sickling crises over the past decade and had not received regular transfusions or hydroxyurea therapy; however, she had suffered bilateral necrosis of femoral heads. Approximately 10 years before, she was diagnosed as having typical cutaneous PXE lesions at the dorsal aspect of the cervical area and the axillae; the diagnosis was confirmed by biopsies of the affected skin areas. At that time, screening for AS was negative. On her current admission, the ophthalmologic examination revealed a visual acuity of 1/10 in the right eye, not accepting any correction. Left eye had a 10/10 visual acuity with correction. Fundoscopy of the right eye showed AS with associated pigmentary clumping (Figure); the parafoveal area was characterized by retinal edema and hard exudates and the perifoveal area was scarred due to previous choroidal neovascular membrane development. Fundoscopy of the left eye revealed AS, without any associated complications. The fluorescein angiography in the right eye (Figure) showed hyperfluorescence over the AS, as well as late fluorescence caused by staining of the fibrous tissue. Screening of patients' relatives for PXE stigmata was negative.

To the best of our knowledge, this is the first report of severe visual acuity impairment related to AS in a patient with a sickling syndrome. Angioid streaks remain usually asymptomatic, but the potential development of subretinal neovascularisation with macular involvement may result in hemorrhage and scarring with visual acuity impairment. Although it has been postulated that AS in the context of hemoglobinopathy-related PXE bear generally a more indolent course compared to those observed in inherited PXE, visual loss due to AS has already been described in two patients with thalassemia major[6,7] and one with thalassemia intermedia.[8] Moreover, subretinal neovascularization due to AS has been reported in a patient with sickle-cell anemia, without however visual acuity impairement[9]

In the present case, AS were combined with skin lesions typical of PXE. Nevertheless, ocular involvement may exist even in the absence of cutaneous stigmata. Actually, in a series of 58 sickle-thalassemia patients, only half of AS-positive cases also had clinically evident skin lesions.[10] Therefore, awareness of this entity by the attending physicians and repetitive ophthalmologic examinations seem indispensable for the early detection of AS and the prevention of the associated complications in patients with hemoglobinopathies. In this context, the use of the Amsler grid as a rather simple method to detect choroidal neovascularization may prove quite useful to detect an evovling ocular condition.

Figure. Upper panel: Fundoscopy of the posterior pole of the right eye of the patient, showing large angioid streaks radiating outwards from the peripapillary area with associated pigmentary clumping. The parafoveal area is characterized by retinal edema with hard exudates arranged in ring (circinate). The perifoveal area is scarred due to previous choroidal neovascular membrane development. **Lower panel:** Fluorescein angiography of the right eye showing hyperfluorescence over the AS, as well as late fluorescence caused by staining of the fibrous tissue.

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Key Words: sickle thalassemia, angioid streaks, visual acuity, pseudoxanthoma elasticum

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