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Sir,

Retinopathy in a patient with Fanconi anemia and vitamin B12 deficiency

Fanconi anemia (FA) is rarely associated with vitamin B12 deficiency. We first report a patient of FA with vitamin B12 deficiency, who developed retinal pigment epithelial changes in two different sites of the retina, including bilateral maculopathy and significant pigmentary changes nasal to the optic disc.

Case report

The patient is a 13-year-old boy, a known case of FA with vitamin B12 deficiency, with a bilateral foot deformity since birth. He was well until 3-years-old when generalized progressive hyperpigmentation of the skin developed, and spread from the face to the trunk. Muscle wasting occurred subsequently. He presented to the Department of Ophthalmology of National Taiwan University Hospital on 9 February 1988 with a 1-year history of progressive blurring of vision in both eyes.

Ophthalmologic examination of the patient disclosed a visual acuity of 20/200 in the right eye and 20/400 in the left eye. Ophthalmoscopic examination revealed a yellowish-white atrophic lesion in the macula of both eyes. There was also an area of significant pigmentary change nasal to the optic disc in both eyes (Figures 1a and b). Examination of color vision with the Farnsworth–Munsell 100-Hue test revealed a total error score of 112 in the right eye and 204 in the left eye, with no discernible axis for each eye. Visual fields performed on the Goldmann perimetry revealed central scotoma in both eyes. Electroretinography revealed abnormal amplitudes consistent with a cone–rod dystrophy in both eyes, and electrooculography showed a light-peak/dark-trough ratio of 1.1 in the right eye and 1.5 in the left eye.

Comment

Some retinopathy has been reported to be present in FA, including retinal hemorrhage, microaneurysm, peripheral ischemic retinopathy, and retinal neovascularization.[1,2] However, retinal degeneration has not been reported in patients with FA.

Few retinopathy has been reported in patients with vitamin B12 deficiency, including maculopathy and retinal degeneration.[3,4] The coexistence of bilateral maculopathy and significant pigment changes nasal to the optic disc was first reported in a patient with inherited transcobalamin II deficiency.[5] This is the first case in which such a similar retinopathy was noticed in a patient of FA with vitamin B12 deficiency.

Conflict of interest

The authors declare no conflict of interest.

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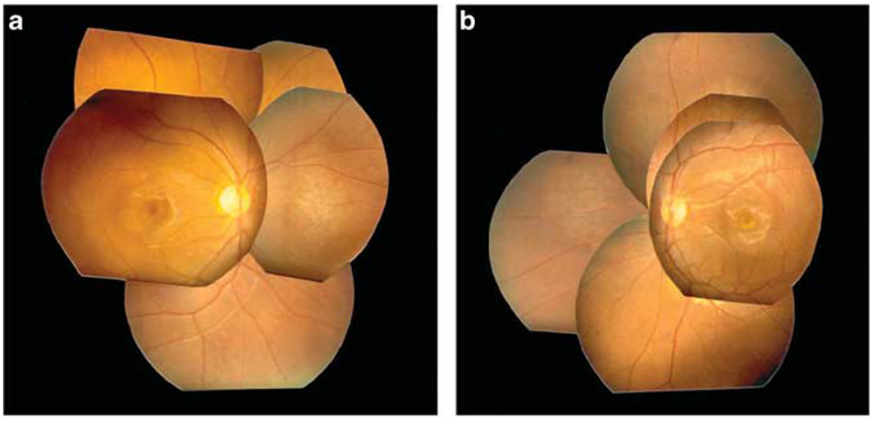

Figure 1 Ophthalmoscopic examination of both eyes. An atrophic macular lesion and significant pigmentary changes nasal to the optic disc are noted in the (a) right eye and (b) left eye.