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Sir,

Cavernous haemangioma of retina and skin

Cavernous haemangioma of the retina is a rare hamartoma first described by Weskamp and Cotlier in 1934[1] in association with similar lesions in the brain and skin. Gass[2] proposed that it should be a distinct component of a neuro-oculocutaneous phacomatosis, which was further supported by Goldberg *et al.*[3] and Pancurak *et al.*[4] who stressed the autosomal dominant inheritance pattern. Autosomal dominant cavernous haemangiomas affecting the skin have been mapped to chromosome 9p[5] and are felt to be distinct from those that affect the central nervous system, which map to chromosome 7q,[6] but this is not universal.[7] The familial form of cavernous haemangioma appears to be most common among Hispanics but has been reported among most ethnic groups. We describe a family with autosomal dominant retinal and cutaneous cavernous haemangiomas.

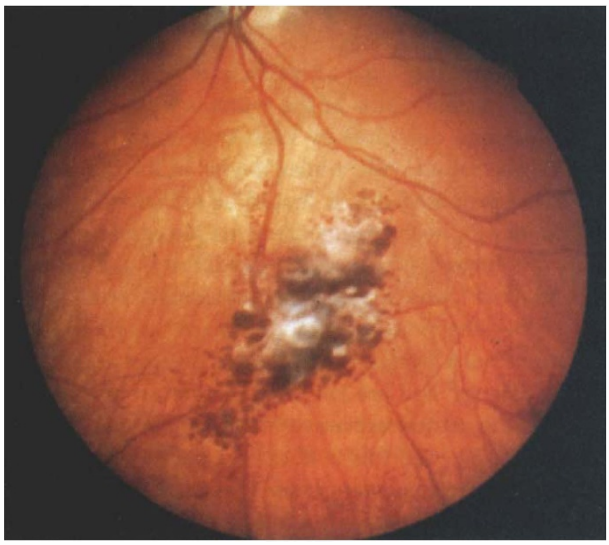
Case report

A 16-year-old Caucasian male was referred by an optician who noted a 'vascular bunch of grapes' in the left eye just below the disc. The patient was asymptomatic and had a Snellen visual acuity of 6/5 in either eye. Fundoscopy revealed clumps of pre-retinal aneurysms with fluid levels and a varying degree of grey fibrosis over the surface, no leakage and normal surrounding vasculature (Fig. 1). A histologically confirmed cavernous haemangioma had been removed from his left forearm at age 9 years. Magnetic resonance imaging did not reveal any brain involvement. His father had angiomas of both facial cheeks and paternal grandfather an angioma of the brow. They were otherwise asymptomatic and the father did not have any ocular involvement. It was not possible to examine his grandfather's eyes.

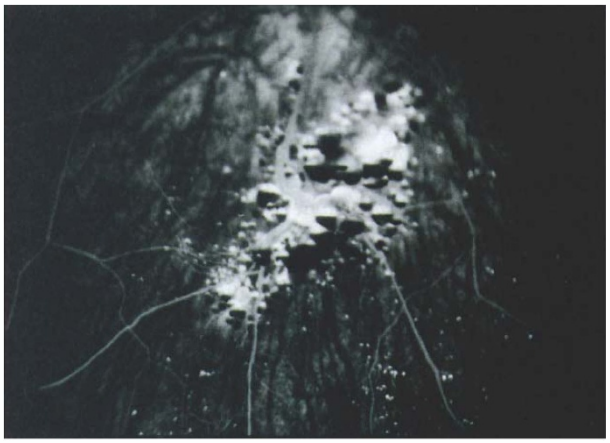
Discussion

The clinical appearance of the retinal cavernous haemangioma in our patient is classical. No treatment is necessary though rarely they may increase in size and bleed, so serial fundus photography is recommended.[8] The lesion may be differentiated from retinal telangiectasias due to the absence of clinical or angiographic evidence of exudation. The lesion of angiomatosis retinae has a dilated tortuous feeder vessel and there is progressive enlargement and exudation.

The association of retinal and cutaneous cavernous haemangioma with cavernous haemangioma of the central nervous system is important as the patient may have a potentially life-threatening lesion. It is recommended that the individual and family members be questioned regarding central nervous system symptoms such as headache, seizures, parasthaesias and convulsions and, if reported, magnetic resonance imaging undertaken and referral made to a



(a)



(b)

Fig. 1. *(a) Retinal cavernous haemangioma with clumps of pre-retinal aneurysms, blood levels, no leakage and normal surrounding vasculature. (b) Fluoroscein angiogram showing classical fluid levels.*

neurosurgeon[8] for consideration of excision or proton beam therapy. As some central nervous system cavernous haemangiomas are clinically silent and may later bleed, some authors recommend neuroimaging of all and surgical removal if the lesion is accessible surgically,[9] but this standpoint is not universally accepted as the lesion may be more benign than previously thought.[10,11] Rates of haemorrhage may vary considerably depending on the patient's age, sex, presentation and lesion location within the central nervous system.[12,13] *De novo* formation of cavernous haemangiomas has been documented,[13,14] which may explain the higher bleeding rates reported in prospective compared with retrospective studies of these lesions.

More information is needed to better define the prevalence and natural history of sporadic and familial cerebral cavernous haemangiomas so that more specific recommendations can be made regarding clinical management, especially of the asymptomatic patient.

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