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Sir,

An unusual presentation of a case of blue rubber bleb naevus syndrome

Blue rubber bleb naevus syndrome (BRBNS) is a rare cutaneo-visceral haemangiomatosis. Bean coined the term BRBNS in 1958,[1] though its association with visceral involvement was reported as early as 1860.[2] The term is derived from the vascular compressible blood sacs that appear blue under the skin; blood may be expressed from these vascular malformations to form a bleb.[3]

It usually presents sporadically as a new mutation, but may be inherited as an autosomal dominant trait.[4] The histological descriptions of these lesions vary from venous and cavernous haemangiomas to arteriovenous malformations and venous aneurysms.[3] A number of ocular manifestations have been described where the lesions involve the orbit, conjunctiva, iris and retina.[3,5] The ocular lesions are associated with cutaneous and visceral lesions that are potential sources of life-threatening haemorrhages. It is imperative that the syndrome is recognised when it presents to the ophthalmologist, and investigated appropriately to avoid the complications associated with the condition.

We describe a patient with BRBNS who presented to the ophthalmologist with bleeding from the conjunctival sac.

Case report

A 26-year-old Caucasian woman presented to the eye casualty with a history of spontaneous bleeding from the left eye associated with periorbital bruising. Episodes of pain and a sensation of fullness around the left eye for many years preceded this. Horizontal diplopia, which resolved spontaneously, occurred when she was 10 years old. At the age of 5 years she had a severe gastrointestinal haemorrhage which necessitated a blood transfusion. A few years later she had episodes of epistaxis, which was treated conservatively. There was no history of trauma, or family history of a bleeding diathesis.

On examination she had an ecchymotic lower lid with haemangiomatous lesions on her left upper lid, caruncle, lower forniceal and bulbar conjunctiva (Fig. 1). There was evidence of fresh blood in the lower fornices. The rest of the ocular examination was normal. Bluish vascular malformations were present on the face and in the buccal mucosa (Fig. 2). A contrast-enhanced CT scan of the brain and orbits was normal. A clinical diagnosis of BRBNS was made. The haemangiomatous lesion on

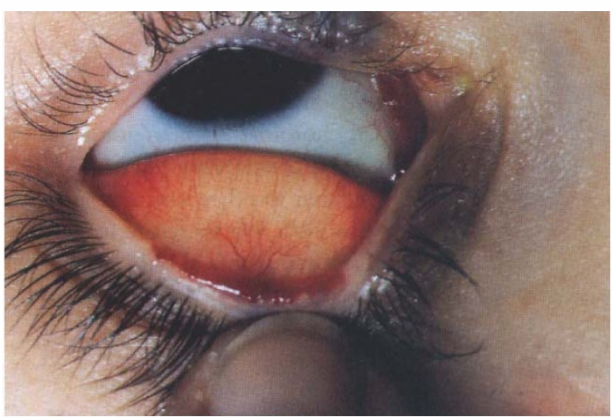

Fig. 1. *The ecchymotic lower lid with haemangiomatous lesion on the left upper lid, caruncle and lower forniceal and bulbar conjunctiva.*

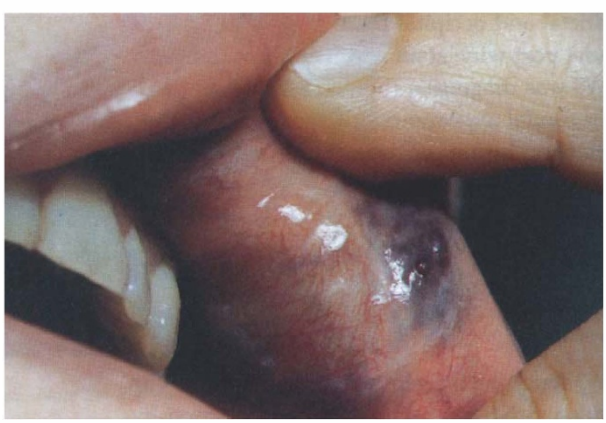

Fig. 2. *Bluish vascular malformation in the buccal mucosa.*

the bulbar conjunctiva was treated with cryotherapy, following which there have been no further episodes of bleeding from the eye over the last 2 years.

Comment

The patient presented had episodes of nasal and gastrointestinal bleeding as a child; she then presented in the third decade with bleeding from the eye. A diagnosis of BRBNS was established clinically, with the history of episodes of blood loss and multiple cavernous-type haemangiomas involving the skin of the face, the trunk and mucosal lesions affecting the eye, mouth and nose.

The episodes of pain and a sensation of fullness, together with the episode of horizontal diplopia, might represent intermittent haemorrhages into the closed tissue spaces of the orbit with spontaneous reabsorption. However, a contrast-enhanced CT scan failed to demonstrate any orbital lesions, in addition to the fact there was no proptosis. This is the first case as far as we are aware of BRBNS presenting with bleeding from the palpebral aperture. As there were no further gastrointestinal haemorrhages since the age of 5 years, endoscopy was not contemplated by the gastroenterologists; however, this does not rule out the presence of asymptomatic vascular malformation in the gut.

Though previous studies[3,5] have reported vascular malformations in the orbit,[6] iris and retina as a part of BRBNS, our case did not demonstrate these features. Three types of vascular lesions have been described: (1) large cavernous haemangiomas affecting vital structures, (2) the classical compressible blood sacs and (3) irregular blue-black spots within the skin. Other viscera such as the thyroid gland, the kidney and the heart may also be affected.[7] Though the central nervous system is only rarely involved,[8] hemispheric and cervical nerve involvement has been described.[9] Some histopathological studies have identified the vascular lesion in BRBNS as erectile cavernous tissue.[10] This might explain bleeding in our case.

Maffucci syndrome,[11] Klippel-Trenauny syndrome[11] and multiple glomus tumour[12] should be considered in the differential diagnosis of BRBNS.

In summary, an ophthalmologist should entertain a high index of suspicion of BRBNS when confronted with bleeding from cavernous conjunctival haemangiomas. This syndrome is associated with cutaneous and gastrointestinal vascular malformations and hence the onus is on the surgeon to make the appropriate referrals and order appropriate investigations to avoid potential life-threatening haemorrhages. As far as we are aware, this is the first case of bleeding from the conjunctival sac in BRBNS.

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Sir,

Mantle cell lymphoma presenting as a choroidal mass: part of the spectrum of uveal lymphoid infiltration

Although intraocular involvement with large cell high-grade B cell lymphoma is rare but well recognised, small B cell intraocular lymphoid infiltration is not well recognised and is extremely rare. We describe the clinical, radiological and pathological features of a mantle cell lymphoma presenting as a choroidal mass.

Case report

A generally fit 57-year-old man presented to the Ophthalmology Department at Leicester Royal Infirmary with a 2 month history of aching eyes and a 3 day history of gradual decreased visual acuity in the left eye. His left visual acuity was 6/24, with mild inflammatory activity in the anterior chamber and mild ocular injection. There was an area of choroidal infiltration and elevation at the posterior pole (Fig. 1a) but minimal vitreous cellular