Sir,

Circumscribed choroidal haemangioma in a patient with Sturge Weber syndrome

Sturge Weber syndrome (SWS) is a rare systemic disorder characterised by vascular malformations affecting the central nervous system (CNS), skin and, in addition to other organs, the eye. The clinical sequelae of CNS involvement include epilepsy, mental retardation and hemiparesis. The eye is variably affected, with vascular malformations of the conjunctiva, episclera and choroid. It is well known that the typical choroidal lesion observed is the diffuse choroidal haemangioma, such that it is used in the diagnosis of SWS by some authors.[1] We describe a case of SWS with unilateral visual loss secondary to circumscribed choroidal haemangioma with serous retinal detachment.

Case report

A 19-year-old woman attended the Sheffield Ophthalmic Oncology Unit in June 1998 with decreased left visual acuity lasting 1 year. Although her 'left vision had always been weaker', this had recently deteriorated, preceded by a left superior hemifield scotoma. Past medical history was of cosmetic laser treatment only for a congenital facial port wine stain that had remained constant in distribution since birth. She denied epilepsy and had performed well in further education. Review of systems was otherwise unremarkable. Family history was normal.

Physical examination showed a typical naevus flammeus with moderate left supraorbital rim hypertrophy. Systemic examination including full CNS examination was normal. Left visual acuity was counting fingers with a left afferent pupillary defect. No vascular malformations were observed in the anterior

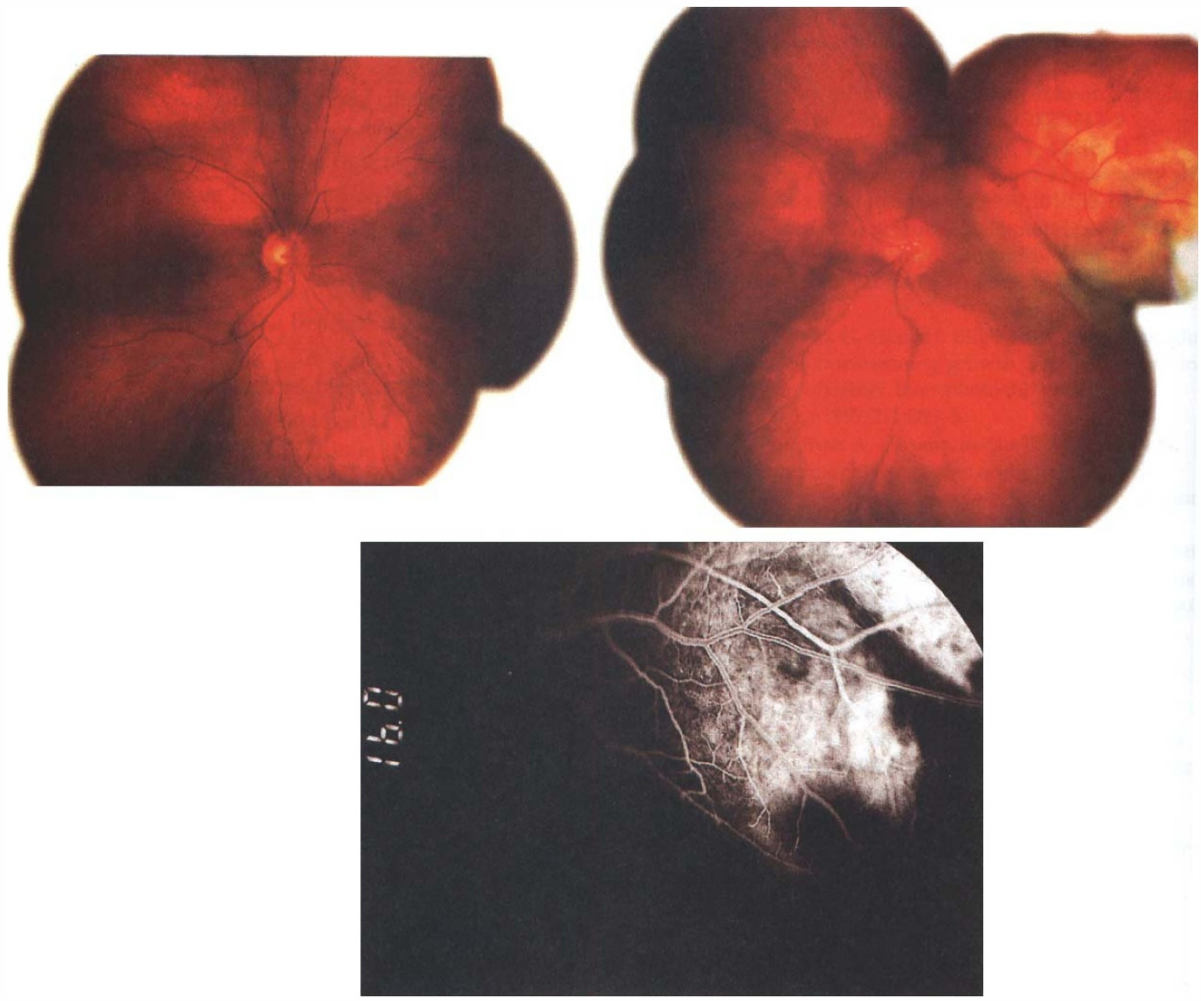

Fig. 1. *Above: Colour digital photomontages of the right and left fundi. Note the dome-shaped reddish-orange circumscribed lesion in the left superotemporal quadrant. Below: Early arteriovenous phase fluorescein angiogram showing the mottled appearance of the choroidal vasculature within the lesion.*

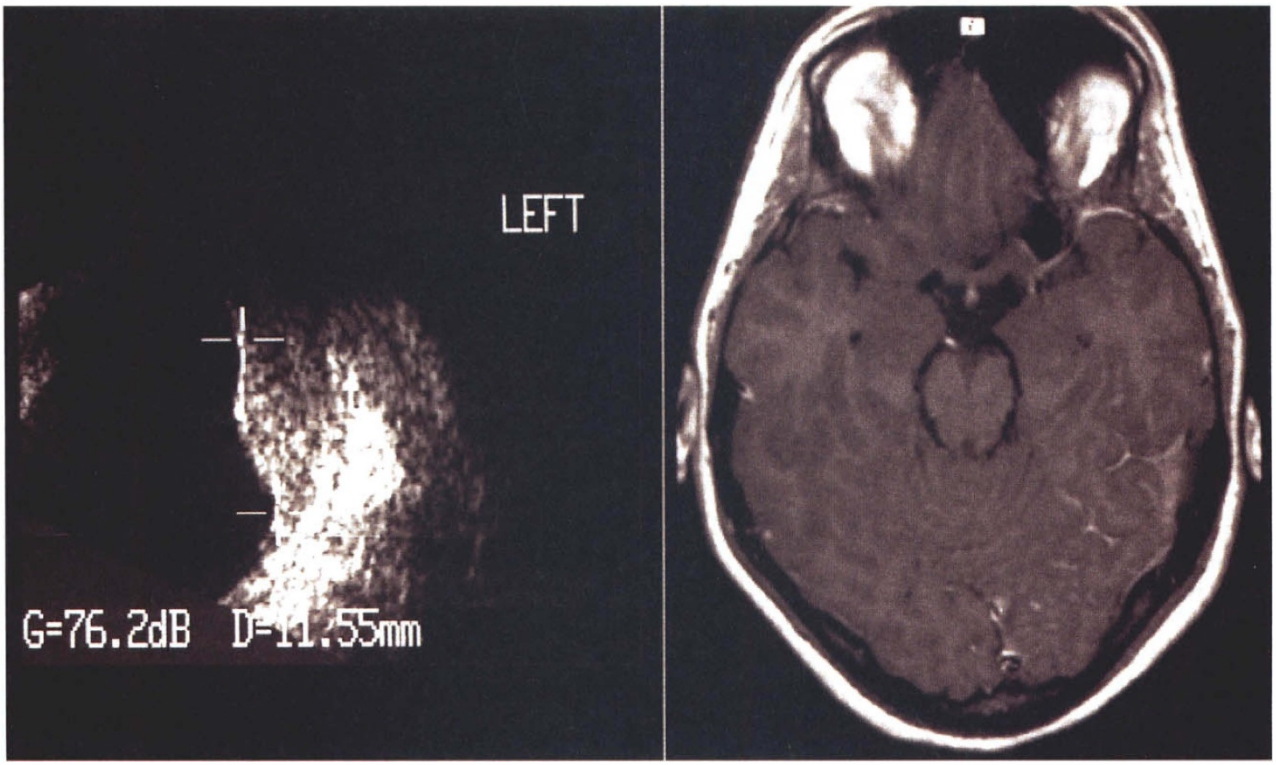

Fig. 2. *Left: B-scan ultrasound showing the circumscribed lesion with regular internal structure; there is no choroidal excavation. Right: Post-gadolinium axial MRI scan showing enhancement of the parietal and occipital leptomeningeal regions.*

segment. Applanation tonometry and gonioscopy were normal. Left fundoscopy revealed a dome-shaped reddish-orange amelanotic circumscribed lesion affecting the temporal aspect of the macula with an exudative retinal detachment extending inferiorly. Areas of pigmentation surrounded the base of the lesion. The optic disc and peripheral fundus were normal (Fig. 1, above).

Fluorescein angiography showed early filling of the lesion during the choroidal phase giving a mottled appearance initially, with later filling of the choroidal vessels within the lesion (Fig. 1, below). Ultrasonography confirmed the dome-shaped choroidal lesion (11.97 mm 11.58 mm 2.97 mm), with regular internal structure, no choroidal excavation and a slightly thickened surrounding choroid with regular internal structure. A-scan ultrasonography demonstrated high internal reflectivity (Fig. 2, left). MRI of the head and orbits showed leptomeningeal high signals affecting the left parietal and occipital brain (Fig. 2, right). In view of the facial port wine stain and MRI appearance, complete SWS was diagnosed (based on François classification[2]). The fundal appearance, ultrasonography and fluorescein angiography are consistent with circumscribed choroidal haemangioma. Due to its location and resultant poor visual acuity, the patient opted for periodic observation. At 6 months there was no change.

Comment

SWS (encephalotrigeminal angiomatosis) is an oculoneurodermal syndrome characterised by haemangiomas typically affecting the trigeminal distribution of the face (naevus flammeus), the choroid and meninges, and glaucoma. The condition is thought not to be hereditary in nature, although families with SWS have been reported. The leptomeningeal angiomas observed are typically on the temporal or occipital meninges on the ipsilateral side of the naevus flammeus. Calcification within the leptomeninigeal angiomas gives classical convoluted areas of high signal on neuroimaging and appear early in childhood. Epilepsy is common, but learning difficulty is uncommon. Various vascular malformations affecting the pancreas, lung, pituitary and gastrointestinal tract have been described but are rare in comparison with neurocutaneous lesions.

The putative mechanism of SWS is a developmental defect in the tissues arising from the prosencephalic and mesencephalic neural crests and/or a defect in neural crest cell migration. The variability of expression of this defect is thought to be responsible for the large variation in phenotypic expression seen between different patients with SWS.[3–5]

Although the classical fundal lesion of SWS is the diffuse choroidal haemangioma (tomato catsup fundus), we feel the circumscribed choroidal haemangioma as seen in our case report could conceivably have developed in a similar manner to other vascular malformations observed in SWS. We therefore feel that circumscribed choroidal haemangioma should be considered to be a rare association of SWS.

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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

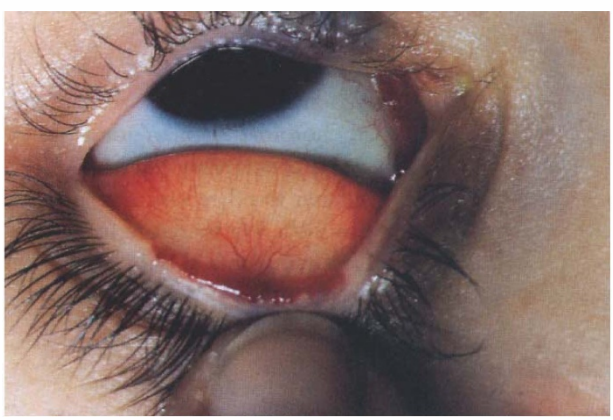

Fig. 1. *The ecchymotic lower lid with haemangiomatous lesion on the left upper lid, caruncle and lower forniceal and bulbar conjunctiva.*

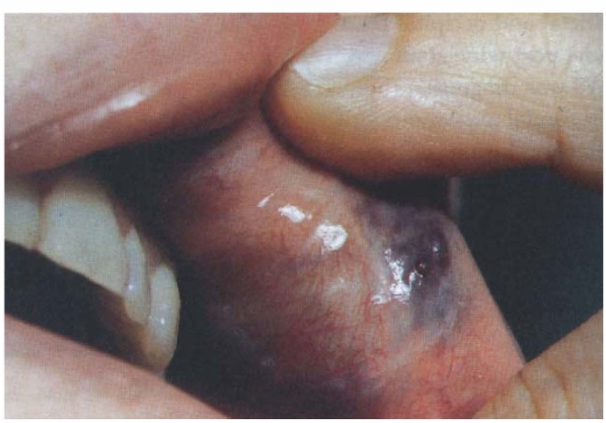

Fig. 2. *Bluish vascular malformation in the buccal mucosa.*