CASE REPORT

Blue rubber bleb naevus syndrome associated with cortical blindness

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SUMMARY

An 83-year-old woman was admitted for investigation of sudden loss of vision. Extensive plum-purple papules and nodules involved her skin and lips but not mucosae. A cutaneous biopsy demonstrated irregular vascular cavernous channels in the dermis and subcutis; deeper vessels displayed smooth muscle. Blue rubber bleb naevus syndrome was diagnosed. This patient had no gastrointestinal symptoms, no family history of blue rubber bleb naevus syndrome, and only developed cutaneous signs after 60 years of age. Magnetic resonance imaging of the brain found multiple cerebral and cerebellar cavernomas with two larger foci of haemorrhage in both occipital lobes, with the largest in the right occipital lobe being associated with calcification. These most likely represent bleeding in relation to cavernomas believed to be the cause of decreased vision, predominantly in the left eye.

Key words: cerebellar cavernoma, cerebral cavernoma, elderly, venous malformation, visual loss.

INTRODUCTION

Sudden-onset visual loss is a medical emergency with multiple possible causes. Blue rubber bleb naevus syndrome is a rare cause of vascular anomalies, which are primarily found in the skin but may affect other organs, particularly the gastrointestinal tract and rarely the brain, including in this case the visual cortex. This report demonstrates the importance of linking previously undiagnosed blue rubber

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bleb naevus syndrome with rapid visual loss, in the absence of other visceral symptoms.

CASE REPORT

An 83-year-old woman presented with rapid overnight deterioration in vision in both eyes. She had been able to watch television and read the newspaper the previous evening, but not the next day. She described no jaw claudication, scalp tenderness, fevers, sweats, weight loss, flashes, floaters or curtains.

Intraocular pressure, extraocular movements and visual fields were normal. The fundus was normal with no swelling or pallor. The retina displayed no red desaturations. Visual acuity of the right eye was 6/60 and that of the left eye was able to count fingers (with glasses). This represented a significant deterioration from a year previously (visual acuity of the right eye was 6/12 and that of the left eye was 6/36 (with glasses)). Coincidentally, a large number of long-standing, asymptomatic widely distributed, compressible, rubbery plum-purple papules and nodules were noted involving the face including her lips, trunk and limbs (Fig. 1), but sparing the mucous membranes. The lesions varied in size from 1 mm to 1 cm in diameter, with the majority of lesions being 2 mm in diameter.

This skin disorder had previously been misdiagnosed as Campbell de Morgan spots. The patient was adamant that the skin lesions were asymptomatic, had only appeared after the age of 60 years, had never bled, had never regressed, and had gradually become more numerous and extensive. There was no family history of BRBNS. The patient had not suffered from anaemia, had received neither iron supplementation nor transfusions, and had not had a colonoscopy. Past medical history included deep vein thrombosis and cardiac failure, for which current medications included perindopril, digoxin and warfarin.

Having excluded glaucoma, retinal artery embolism, giant cell arteritis, retinal detachment and other pathology of the cornea, lens, globe, retina and fundus, the cause of

Abbreviations:

BRBNS	blue rubber bleb naevus syndrome
GIT	gastrointestinal tract



Figure 1 (a) Extensive plum-purple papules and nodules of blue rubber bleb naevus syndrome (BRBNS) on the face, including involvement of the lips. (b) Extensive plum-purple papules and nodules of BRBNS on the trunk. (c) Extensive plum purple papules and nodules of BRBNS on the upper limb.

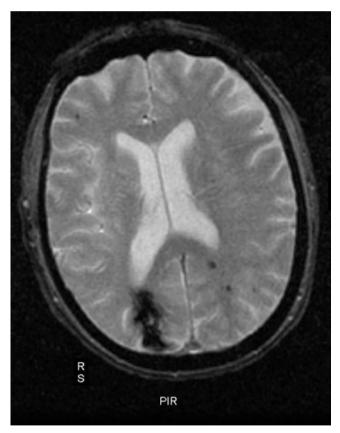


Figure 2 Transverse horizontal section computed tomography scan of the brain showing the right occipital area of haemorrhage.

visual loss was investigated further using computed tomography (Fig. 2) and magnetic resonance imaging (Fig. 3), which were performed 4 weeks after presentation. Multiple cerebral and cerebellar cavernomas with two larger foci of

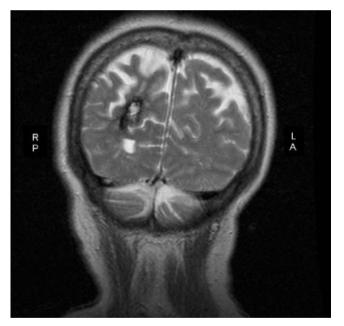


Figure 5 Coronal magnetic resonance imaging scan showing the right occipital area of haemorrhage.

old haemorrhage in both occipital lobes, with the largest being associated with calcification in the right occipital lobe, were found. A ruptured arteriovenous malformation was believed less likely, due to lack of significant vascular dilation. No optical chiasmal abnormality was depicted. Blood investigations taken on admission included a normal C-reactive protein, electrolytes, urea, creatinine, liver function tests and full blood examination. The international ratio was 1.7 (patient on warfarin). Results of a skin biopsy taken at another institution were retrieved, where histopathology reported a 'punch biopsy of a cavernous

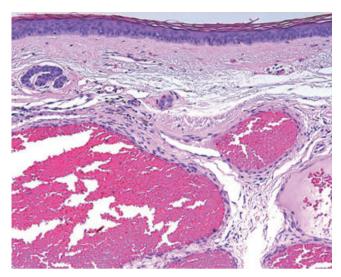


Figure 4 Skin biopsy from papule on the forearm in blue rubber bleb naevus syndrome shows irregular dilated and congested vascular space lined by a thin layer of regular flattened endothelium in the papillary dermis (H&E).

haemangioma' (Fig. 4). On review, the sections showed a true venous malformation.

DISCUSSION

Blue rubber bleb naevus syndrome is a rare condition, consisting of bluish-purple venous malformations of the skin and viscera, predominantly the GIT and mucosal surfaces, but also involving multiple other organs. Our patient's clinical presentation was atypical inasmuch that her cutaneous lesions presented after her 60th birthday, whereas the most common cutaneous lesions of BRBNS are present at birth or shortly after. Other cases of late presentation have been described, but are noted to be unusual.1 Our patient's cutaneous lesions were typical in size and number, but lack of mucosal involvement is remarkable. Typically, the cutaneous lesions increase in size and number with age. Once developed, the vascular malformations of cutaneous BRBNS display no spontaneous regression, bleeding, or malignant change; however, lesions of internal organs do spontaneously bleed. Gastrointestinal tract lesions may cause anaemia and sometimes intussusception.² Our patient had no anaemia and has no history of GIT symptoms, and as a consequence has neither been investigated for faecal occult blood nor undergone colonoscopy. The lack of GIT involvement is notable in our case.

Haemorrhage of central nervous system lesions can cause ataxia, dementia, nystagmus and facial palsy,⁵ or even slowly progressive ataxia and brain stem signs.⁴ Lesions have been described in adrenal glands, thyroid, spleen, ocular orbit and muscle.² A number of musculoskeletal problems have occurred in affected individuals, including gigantism of an arm requiring amputation in infancy. Consumption coagulopathy has been reported.⁵ The severity and clinical course of the cutaneous lesions in no way predicts that of the internal ones. Most cases are

sporadic, as in our patient; however, autosomal dominant inheritance has been described in a number of families. ¹ Cases have been described in all races. ²

This patient had originally been biopsied to exclude Kaposi's sarcoma or metastatic melanoma; both easily distinguished on histology. Our patient's histopathology report of cavernous haemangioma reflects the common yet inappropriate labelling of all benign vascular lesions as haemangiomas, whereas the vascular lesions of BRBNS are true venous malformations. Blue rubber bleb naevus syndrome may also occasionally be confused with eccrine angiomatous harmartomas, glomangiomas, Maffucci's syndrome and angiokeratoma corporis diffusum. By comparison, Campbell de Morgan spots tend to be bright cherryred papules rather than the blue-purple of BRBNS, and histologically show dilated and interconnected segments of venous capillaries and postcapillary venules in the dermal papillae. In hereditary haemorrhagic telangiectasias. which may also involve skin and the GIT, lesions are typically smaller (2–5 mm) and punctiform. 6 Subungual lesions, nail bed involvement and spider and punctuate telangiecatasias will also help distinguish hereditary haemorrhagic telangiectasias from BRBNS. Histologically, hereditary haemorrhagic telangiectasias display dilated thin-walled vessels lined by a single layer of endothelium. Ultrastructurally these are venules or telangiectasias, whereas in BRBNS the cutaneous lesions are venous malformations composed of irregular cavernous channels in the deep dermis and subcutis, and contain smooth muscle in the vessel walls. Treatment of the BRBNS largely comprises symptomatic treatment of its complications. If anaemia cannot be controlled using iron supplements or intermittent transfusion, resection of heavily involved lengths of bowel may be indicated. Successful carbon dioxide laser treatment of the skin lesions has been reported.⁷ This patient did not request any treatment for her lesions. Warfarin was ceased after intracranial haemorrhage; though it is difficult to estimate what role warfarin may have played in causing the original bleed.

This patient was notable because of the relatively late onset in life and lack of GIT symptoms. In this case, visual loss appears to be the only systemic feature attributable to her BRBNS.

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