Blue rubber bleb naevus syndrome: A rare cause of gastrointestinal bleeding in an African child

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ABSTRACT

Blue rubber bleb naevus syndrome (BRBNS) is characterised by vascular malformations of the skin and gastrointestinal tract. We present the rare case of BRBNS in an African child. She presented with large-volume gastrointestinal bleeding and was managed by on-table colonoscopic identification and surgical excision, of all her enteric, vascular malformations.

Key words: Blue rubber bleb naevus syndrome, gastrointestinal bleeding, on-table colonoscopy, surgery, vascular malformation

DOI: 10.4103/0189-6725.70430

INTRODUCTION

This case is of a 9-year-old girl with Blue rubber bleb naevus syndrome (BRBNS), who demonstrated the classic cutaneous and oral, venous malformations. It took three presentations to hospital with significant gastrointestinal bleeding before the diagnosis was made. Undoubtedly, the delay in establishing the diagnosis reflects the rarity of this syndrome, particularly in the Black population.

CASE REPORT

A 9-year-old, Black girl from Kwa-Zulu Natal, presented with a 5-day history of lower abdominal pain and multiple episodes of fresh, red, rectal bleeding. Inquiry into past medical history revealed two previous episodes of rectal bleeding over the proceeding 3 years; the worst resulted in an anaemia of 1.6 g/dl. These episodes had settled spontaneously and investigation at another institution failed to identify a cause. Three years earlier she had undergone surgery to excise a benign, soft tissue, vascular lesion from her right leg. Her family history was that both parents had died from HIV-related pathology and her remaining five elder half-siblings (with shared mother) were healthy.

On examination she was haemodynamically stable and clinically anaemic. Two smooth, submucosal, vascular swellings were noted in her mouth; one distorting the contour of her lower lip and the second seen in a sublingual position [Figure 1].

Abdominal examination confirmed the presence of fresh blood in her rectum. A 10-cm scar from her previous surgery was noted on her right leg and a 2-cm compressible, subcutaneous vascular lesion on her proximal calf.

On admission the patient's haemoglobin was 6.4 g/dl, mean cell volume was 73 fl and platelets 233 $\times$ 10^3/μl with hypochromia and anisocytosis noted on film. Colonoscopy demonstrated two submucosal, vascular lesions of about 1 cm diameter in the transverse colon and the proximal rectum. The latter was ulcerated with fresh clot adherent. No abnormalities were detected on oesophago gastroduodenoscopy.

Following blood transfusion and bowel preparation, a laparotomy and combined on-table colonoscopy was

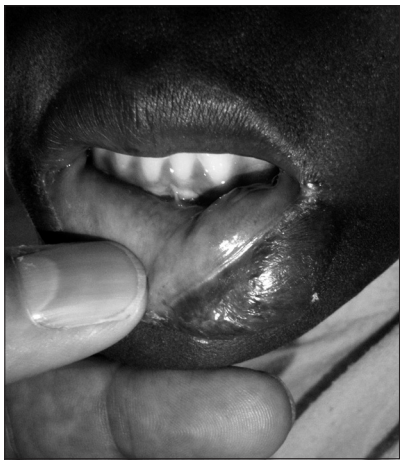


Figure 1: Venous malformation of child's lower lip

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performed and a further two colonic and six small bowel lesions were identified. Both colonic lesions were in the right colon and measured about 0.7 cm in diameter. The six small bowel lesions were sporadically distributed throughout the ileum (four) and jejunum (two). They ranged from 0.5 to 1 cm in diameter. The symptomatic, proximal rectal lesion, seen at initial endoscopy, was located on the antemesenteric border of the recto-sigmoid junction. All the lesions were excised through transverse enterotomies [Figure 2]. Histology reported multiple submucosal cavernous haemangiomas.

The child made an uneventful recovery and has remained bleed-free with a stable haemoglobin at 9 months. Follow-up outpatient Computed Tomography and Magnetic Resonance Imaging of her head revealed four incidental, intracranial, venous malformations and referral to a quaternary centre for follow up with a neuroradiologist was made.

DISCUSSION

The association between cutaneous and intestinal vascular lesions was first reported by Gascoyne in the 1860s. In 1958 William Bennet Bean publicised BRBNS with his description of the cutaneous lesions having "the feel and look of cutaneous nipples".[1-2] Over 150 cases of BRBNS are reported in today's literature, the majority of which affect Japanese or White patients; however, at least three black patients with BRBNS are reported (two African-American patients and one South African child also from KwaZulu Natal).[3,4]

BRBNS affects both sexes and is usually sporadic, although autosomal dominant inheritance has been reported.[4] Bean described three types of lesions in BRBNS, which are often noted at birth or during the neonatal period, although they can present later. The first type is the compressible, red-blue, nipple-like lesions. The second type is blue-black non-blanching macular lesions and the third type is subcutaneous, vascular, soft tissue lesions as seen in our case.[2,4] It is these soft tissue malformations that are thought to signify the involvement of the gastrointestinal tract.

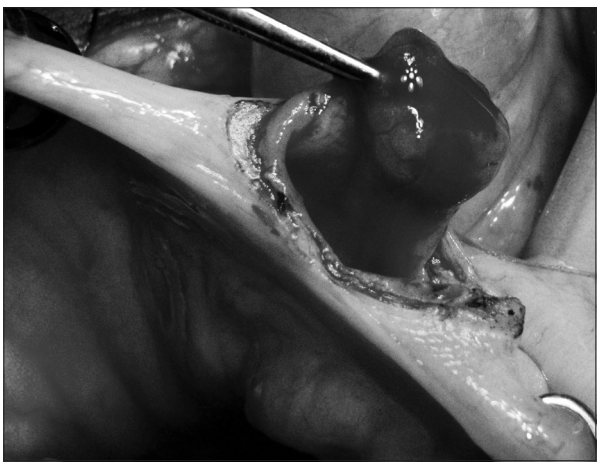


Figure 2: Excision of colonic submucosal cavernous haemangioma

The skin lesions of BRBNS are usually asymptomatic unlike the gastrointestinal lesions which classically produce anaemia secondary to occult blood loss. Abdominal pain, infarction, volvulus, haematemesis and rectal prolapse or bleeding are also recognised presentations.[3-5]

The literature describes conservative management, with iron supplementation and blood transfusion, as the initial approach to manage the gastrointestinal bleeding. Successful, non-surgical control of symptomatic lesions using anti-angiogenic treatments and endoscopic snare polypectomy and argon plasma coagulation are reported. Persistent bleeding from residual untreated small bowel disease and recurrent disease has lead the reliability and consistency of results using this approach to be questioned.[3-6] As BRBNS most commonly affects small bowel, some surgeons argue for early surgical intervention.[5]

Fishman *et al*.[5] reported a series of 10 BRBNS patients presenting with symptomatic gastrointestinal lesions which were treated with operative excision. At the time of operation these patients had received a median of 28 units of blood transfusion (mean 53, range 1–>200) during previous conservative management. Follow-up ranged from 2.9 to 10.3 years, at which only one patient had required further transfusion requirement and two others were noted to have residual or recurrent disease.[5] All three of these patients had sub-optimal excisions at their initial laparotomy. One was not assessed with on-table enteroscopy; the second, with extensive disease, only completed the first-stage of a planned two-stage procedure, and the third patient, also with extensive disease, opted for a compromised single-stage procedure. This relied on a greater use of band and suture-ligation treatment over more radical surgical excision techniques to make the procedure feasible.[5]

Our child was from a rural location in Kwa-Zulu Natal where access to medical facilities equipped to deal with significant, acute bleeding may take several hours by road travel. In addition, the availability of blood transfusion in our region is not guaranteed as

there remains a chronic shortage of blood transfusions resulting from HIV-related reductions to the donor pool.[7] Furthermore, equipment to perform therapeutic endoscopy was not available in the public-funded locality at that time.

Guided by Fishman *et al.*'s[5] data and these specific local considerations, we elected for early operative intervention to manage our patient. Fishman *et al.*'s[5] experiences demonstrate the value of on-table enteroscopy; use of this approach in our case allowed the identification of additional lesions.[5] Although not currently available at our centre, pre-operative, video-capsule endoscopy to define a patient's small bowel involvement, has avoided the need for on-table enteroscopy in such cases, with external inspection of the bowel alone identifying all known lesions.[8] We chose to excise the 10 enteric vascular malformations found in our case through transverse enterotomies as data suggest that this gives the best chance of avoiding recurrence.[5] Consideration to operative length and maximal bowel preservation may favour alternative techniques such as segmental bowel resection, wedge-resection, suture ligation and extramural occlusion with purse-string suturing in patients with more extensive disease.[5,9]

Whilst the coexistence of cutaneous and gastrointestinal venous malformations are the hallmark of BRBNS, venous malformations in additional sites, such as the oral cavity, central nervous system, endocrine glands, musculoskeletal system, kidney, bladder, liver, spleen, heart and lungs, are also recognised.[3,4] The extra-cutaneous and gastrointestinal lesions may present with haemoptysis, haematuria and skeletal deformities or be asymptomatic, as with our child's intracranial lesions.

Developmental venous anomalies and developmental arteriovenous fistulas of the brain are associated with BRBNS. They present with the signs and symptoms of increased intracranial pressure, thrombosis and associated cerebral infarction. Management with endovascular embolisation has been successfully reported.[10]

CONCLUSION

Our management of a case of BRBNS in Kwa-Zulu Natal was modified to accommodate our local geographical and resource constraints opting for early operative intervention of the gastrointestinal lesions over more conservative approaches. It also demonstrated how the use of on-table colonoscopy can aid identification of additional gastrointestinal lesions. Furthermore, awareness of the associated extra-cutaneous lesions has allowed early diagnosis and follow up of intracranial involvement.

In summary, a greater awareness of BRBNS will allow earlier diagnosis, focused endoscopic investigation and timely surgical intervention with appropriate follow up in future cases.

ACKNOWLEDGEMENT

We would like to acknowledge Edmund JN Jack (MA, MB BS) who worked in the Department of Paediatrics at Ngwelezane Hospital and helped and supported the writing of this case report.

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Source of Support: Nil, **Conflict of Interest:** None declare.