

Case Report

Blue rubber bleb naevus syndrome: a rare presentation of vascular disorder

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Received: 11 June 2022

Revised: 01 July 2022

Accepted: 07 July 2022

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ABSTRACT

Blue rubber bleb naevus syndrome (BRBNS) is a rare syndrome of AV malformation which mainly involves skin, soft tissue, gastrointestinal tract and other parts of the body. Its occurrence is usually sporadic, although cases of autosomal dominant inheritance is reported. At presentation symptoms depend on the organs involved. Patients may present with acute or chronic gastrointestinal bleed and it usually seen in children and rarely in adults. We herein reported a rare case of BRBNS in 17 years old male patient who suffered from Malena for four years with past history of haemangioma excision on back. Patient was on regular blood transfusion for six months. Endoscopy revealed multiple AV malformation in gastrointestinal tract. Patient underwent laparotomy with resection of involved segments of ileum with primary end to end anastomosis. Post-operative recovery was uneventful. So here we can conclude that the definitive management of BRBNS affecting the gastrointestinal tract is excision. This article was an original case review of management of BRBNS. The clinical presentation, required investigations and management of the same.

Keywords: BRBNS, Venous malformation, Gastrointestinal bleeding

INTRODUCTION

BRBNS is a rare vascular anomaly syndrome usually consisting of multifocal venous malformations.¹ It is congenital but presentation is in childhood or early adulthood. It affects skin and internal organs of body particularly gastrointestinal tract (GI). This association of “haemangiomas” of the skin and GI tract was first reported in 1860, and fully characterized by William Bean in 1958, giving rise to the eponym “Bean syndrome”.² GI lesions of BRBNS are more clinically relevant than the skin and soft tissue lesions.³ BRBNS presents with multiple, bluish vascular swellings particularly in skin and gastrointestinal tract but may involve other viscera like liver, spleen, lungs etc.⁴ Small intestine is most commonly affected by BRBNS. Bleeding from GI tract can cause mild to moderate anaemia. These patients require iron supplementation and multiple blood transfusions. Most cases are sporadic,

some researchers believe that cases are inherited as autosomal dominant.⁵⁻⁸ The molecular mechanisms underlying this disease are yet to be fully elucidated. It has been identified that normal endothelial cells of adult vessels do not show C kit expression, whereas at least partial C kit positivity has been reported in angiosarcomas.⁹ In addition, it has been demonstrated that pharmacological inhibition of the C kit signalling pathway in cavernous haemangiomas by selective kinase inhibitors may offer options in the treatment of BRBNS patients.¹⁰

CASE REPORT

The 17 years male Hindu patient from low socio-economic class had black coloured stool for 4 years. He had melena and anaemia. Multiple blood transfusion was given for 6 months and later it increased to once every 15 days. He had no abdominal pain, vomiting, constipation,

fever, hematemesis, haemoptysis and haematuria. Patient was not taking anti-platelet or anti-coagulant therapy. He had history of excision of haemangioma on right scapular lesion at the age of 2 years and recurrence developed at same site after 8 years and re-excision done. On general examination pallor was present, per-abdominal examination revealed no abnormality, digital rectal examination and proctoscopy was normal.



Figure 1: Intra-operative finding of 6 lesions in terminal ileum.

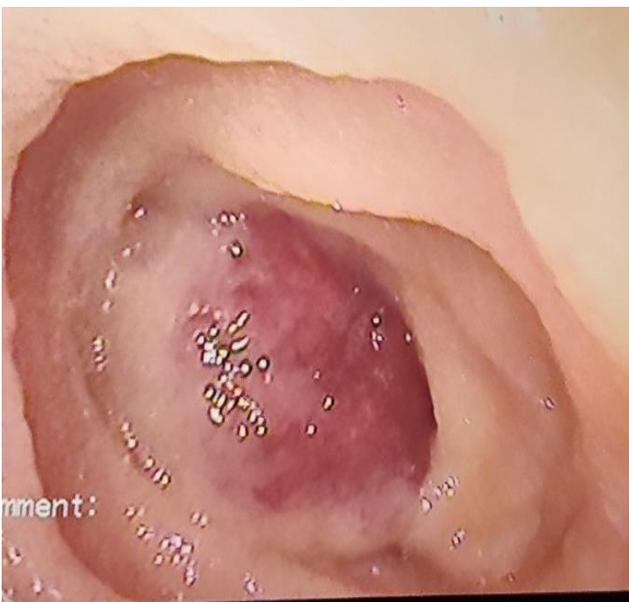


Figure 2: Lesion seen during enteroscopy.

On investigations complete hemogram, stool routine/microscopy, X-ray abdomen and ultrasonography abdomen pelvis were within normal limits except haemoglobin 5 gm/dl, haematocrit 19% and stool for occult blood positive. Upper GI scopy, capsule

endoscopy and colonoscopy was suggestive of 1 elevated, bullous, hyperpigmented lesion in lower oesophagus p/o venous malformation, 3 lesions in stomach, 5-6 lesions in mid-terminal ileum and two in caecum. Two lesions in caecum were fulgurated endoscopically.

Patient had persistent complain of melena and operative intervention was decided. On exploratory laparotomy and enteroscopy, 6 bluish coloured vascular lesions found in middle and terminal portions of ileum. Enteroscopy was done proximal to the most proximal lesion at junction of jejunum-ileum. Wedge resection/resection of involved segments of ileum and primary end to end anastomosis done at 6 sites.

Histopathology suggested haemangioma of ileum.

Post-operative recovery was uneventful. Patient was discharged on post-operative day 8. On follow-up patient had no complain of blood in stool or black stool, stool routine/microscopy did not reveal occult blood and there was no requirement of blood transfusion.



Figure 3: Lesions seen in resected specimen.

DISCUSSION

BRBNS is a rare specialized vascular malformation. They are small, circumscribed and multifocal. It is most commonly present on skin, GI tract, soft tissues, but may occur in any tissue.⁷ Cutaneous lesions are small measuring less than 1-2 cm and blue to purple in colour. BRBNS lesions in GI tract present with bleeding or act as lead point for small bowel intussusception. GI bleeding is chronic continual requiring multiple blood transfusions. Antiangiogenic agents and endoscopic methods have been tried which are unable to permanently control blood loss. So, for patients with bleeding from BRBNS, GI lesions should undergo complete gastro-intestinal tract

exploration and removal of all lesions. After removal of lesions bleeding is less likely and recurrence is not found.

CONCLUSION

The definitive management of BRBNS affecting the gastrointestinal tract is excision.

Funding: No funding sources

Conflict of interest: None declared

Ethical approval: Not required

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Cite this article as: Dalal PR, Parikh JV, Patel AM, Suratwala PT. Blue rubber bleb naevus syndrome: a rare presentation of vascular disorder. Int Surg J 2022;9:1486-8.