

*Case Report*

# A rare cause of recurrent gastrointestinal bleed: Blue rubber bleb nevus syndrome

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Congenital venous malformations (VMs) are rare cause of gastrointestinal (GI) bleed in children. Blue rubber bleb nevus syndrome characterized by VMs in GI tract and skin affect at early age in life. Diagnosis is based on typical skin lesion and history of recurrent GI bleed. In this article, we presented a similar case of young girl with typical skin lesion and recurrent GI bleed requiring multiple blood transfusions presenting our department. She was subjected to surgical treatment and endoscopy therapy due to failure of conservative therapy. This case report depicts importance of typical clinical features of rare diseases.

**Keywords:** Venous malformations, Blue rubber bleb nevus syndrome, Melena, Anemia

**INTRODUCTION**

Blue rubber bleb nevus syndrome (BRBNS) or bean syndrome is a rare congenital disorder, characterized by numerous, diffuse venous malformations (VMs) affecting cutaneous, and internal organs commonly gastrointestinal (GI) tract. BRBNS patients present with chronic iron deficiency and GI bleeding.<sup>[1]</sup> It usually present at any age from birth to adulthood with no sex prediction.<sup>[2]</sup> Skin lesions usually do not require treatment while treatment of GI lesion depends on extent of lesions and severity of bleeding. Most of them are treated with iron therapy and blood transfusion. In cases of recurrent or symptomatic GI bleeding, endoscopic and/or surgical treatment has been proposed. We present a case of young girl who presented with recurrent GI bleed from the VMs in GI tract for which she was subjected to surgical resection and further planned for endoscopic therapy.

**CASE REPORT**

Sixteen years old female patient presented to our emergency room with history of black color stools, pallor, and bluish nodules on her soles. She had history of operated Meningocele in childhood. She had asymptomatic bluish rubbery nodule on her sole and lower limb since childhood which has increased in numbers with increase in age. She never had melena in her school age but felt fatigue during activities like playing games. At age of 11 she was evaluated for fatigue her hemoglobin was 8 g% and anemia profile was

suggestive of iron deficiency anemia. She was treated with oral iron therapy. Her symptoms recur 3 years back and her hemoglobin was found to be 6 g%. For that she underwent upper esophagogastroduodenoscopy which showed duodenal angioma. Patient hemoglobin improved with blood transfusion and iron therapy. Patient were not on follow-up and now from the past 3 months she noticed black stools and was referred to our hospital. On examination we found multiple bluish non-tender nodules on her soles [Figure 1a] and lower limbs. Nodules size around 1–2 cm, compressible with refilling. Laboratory findings showed hemoglobin 5 g/dl, total count 5000 cells/mm<sup>3</sup>, and platelets 1.8 lac/mm<sup>3</sup>. Upper GI endoscopy showed polypoidal growth at D1-D2 junction. Colonoscopy showed large bluish vascular malformation in descending and sigmoid colon measuring around 1–2 cm, another small vascular bleb seen in rectum [Figure 1b]. To evaluate small intestinal vascular malformations, we performed capsule endoscopy. It showed multiple vascular malformations in distal small intestine [Figure 1c]. Based on multiple VMs in skin and gastrointestinal tract diagnosis of BRBNS was made. After studying the literature relatives were explained regarding the combine options of limited small bowel resection and submucosal resection of large bowel lesion. The decision for extent of small bowel resection was based on intraoperative enteroscopy. During surgery 50 cm of proximal ileum was resected and primary anastomosis was done. Post-surgical resection no further bleeding episode

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noticed and her hemoglobin remained stable. She is now planned for submucosal resection of colonic lesion if anemia or GI bleed recurrence occurs.

## DISCUSSION

BRBNS is named after William Bennett Bean in 1958, although it has been described later in 1860 by Gascoyen.<sup>[3]</sup> Most of cases are sporadic in nature but studies have described its genetic predisposition as autosomal dominant trait in some families. Recent genetics studies have described locus on chromosome nine causing VM.

BRBNS can present at any age, oldest case reported was at 82 years. Skin lesions rarely bleed and remain asymptomatic throughout the life. GI tract lesion may appear from mouth to anus commonly present in small intestine and colon.<sup>[4]</sup> Patient usually presents with chronic iron deficiency anemia or GI bleed. These VMs can be found in many other organ of body including brain, muscle, spine, and other visceral organs.<sup>[5]</sup> Diagnosis is made on clinical grounds with typical skin and GI lesions. In present case, patient has multiple skin and GI lesions.

Treatment for skin lesions required only when symptomatic, include surgical excision or laser ablations. GI lesion treatment depends on size, numbers, and length of bowel involved and evolutions of lesions. There are no clinical guidelines or definite treatment of GI lesions as we have only case reports in literature. Management options include

iron replacement therapy, endoscopic therapy, surgical interventions, and pharmacotherapy. Agents such as corticosteroids, thalidomide, interferon- $\alpha$ ,  $\beta$ -blocker, and sirolimus have been attempted based on anti-angiogenesis effect seen in infantile hemangioma but no durable beneficial effects of any drug seen in case reports.<sup>[6]</sup> Sirolimus has been reported in several case reports to be effective in reducing VMs related to BRBNS.<sup>[7]</sup> Endoscopic interventions such as argon plasma coagulation, band ligation, sclerotherapy, and endoscopic submucosal dissection are the useful in small and non-transmural VMs.<sup>[8]</sup> Patient with recurrent bleeding or if lesions are in small bowel, transmural, and widely distributed in intestine surgical treatment including combination of wedge resection, polypectomy, and segmental bowel resection should be considered.<sup>[9]</sup> However, recurrence of lesion is seen in follow-up case studies; hence, close follow-up of such patients is required. In our case also patient had recurrent GI bleed with transfusion dependent anemia. As lesions were in distal small intestine and distal colon, she was subjected for surgical resection of small bowel lesions and further planned for endoscopic submucosal resection of colonic lesion.

## CONCLUSION

We present a rare case of BRBNS presented with iron deficiency anemia and melena. BRBNS is rare venous malformation, with unclear pathogenesis, typical skin and other organs lesions and without any standardized treatment protocol. Combination of endoscopic and surgical treatment seems to be effective in medically refractive cases. Long follow up of every diagnosed patient is required for case base treatment and to see for recurrence.

## Declaration of patient consent

The authors certify that they have obtained all appropriate patient consent forms.

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Nil.

## Conflicts of interest

There are no conflicts of interest.

## REFERENCES

1. Liu Q, Chen YP, Li YM. Blue rubber bleb nevus syndrome: A report of one case associated with recurrent epistaxis. *Chin Med J (Engl)* 2007;120:731-3.
2. Deng ZH, Xu CD, Chen SN. Diagnosis and treatment of blue rubber bleb nevus syndrome in children. *World J Pediatr* 2008;4:70-3.
3. Dobru D, Seuceha N, Dorin M, Careianu V. Blue rubber bleb



**Figure 1:** (a) Multiple bluish non tender nodules on her soles, (b) large bluish vascular malformation in sigmoid colon, (c) capsule endoscopy depicting large vascular malformation in distal small intestine.

- nevus syndrome: Case report and literature review. *Roman J Gastroenterol* 2004;13:237-40.
4. Boente MD, Cordisco MR, Frontini MD, Asial RA. Blue rubber bleb nevus (Bean syndrome): Evolution of four cases and clinical response to pharmacologic agents. *Pediatr Dermatol* 1999;16:222-7.
  5. Beck PL, Aspinall AI, Kilvert VM, Dort J. Blue rubber bleb nevus syndrome. *Gastroint Endosc* 2002;56:598-600.
  6. Yuksekkaya H, Ozbek O, Keser M, Toy H. Blue rubber bleb nevus syndrome: Successful treatment with sirolimus. *Pediatrics* 2012;129:e1080-4.
  7. Ogu UO, Abusin G, Abu-Arja RF, Staber JM. Successful management of blue rubber bleb nevus syndrome (BRBNS) with sirolimus. *Case Rep Pediatr* 2018;2018:7654278.
  8. Malafaia MC, Heissat S, Jacob P, Gall CL, Ruiz M, Saurin JC, *et al.* Blue rubber bleb nevus syndrome: Endoscopic treatment with sclerotherapy during double-balloon enteroscopy in a 9-year-old boy. *Endoscopy* 2019;51:E98-100.
  9. Fishman SJ, Smithers CJ, Folkman J, Lund DP, Burrows PE, Mulliken JB, *et al.* Blue rubber bleb nevus syndrome: Surgical eradication of gastrointestinal bleeding. *Ann Surg* 2005;241:523-8.

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