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# Unusual presentation of Blue Rubber Bleb Nevus syndrome with multiorgan involvement and successful mTOR inhibitor therapy

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## Abstract

Blue Rubber Bleb Nevus Syndrome (BRBNS) is a rare vascular anomaly characterised by venous malformations involving the skin and internal organs, particularly the gastrointestinal tract. We present the case of a 36-year-old female with extensive mucocutaneous and visceral lesions, recurrent anemia, hematuria, and a history of recurrent abortions. Diagnosis was established through imaging and endoscopic evaluation. She was successfully managed with Everolimus, a mechanistic target of rapamycin (mTOR) inhibitor, leading to clinical stabilization and elimination of transfusion needs. This case underscores the therapeutic potential of mTOR inhibitors in complex BRBNS cases with multiorgan involvement.

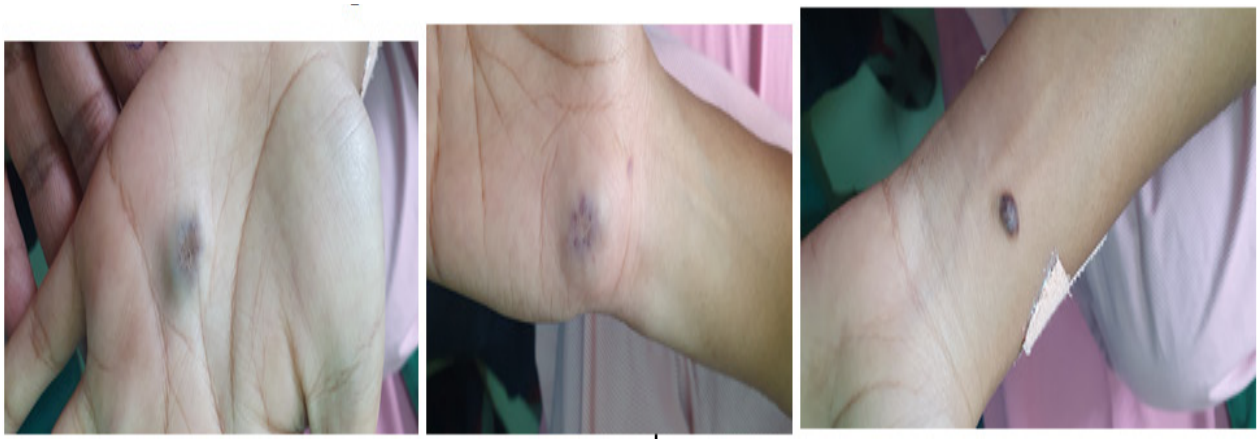
***Keywords:*** Blue Rubber Bleb Nevus syndrome (BRBNS); Everolimus; mTOR inhibitor; Venous malformations; Case report.

## Introduction

Blue Rubber Bleb Nevus Syndrome (BRBNS), or Bean syndrome, is an extremely rare condition with fewer than 250 documented cases globally. It is characterized by multiple, sporadically distributed venous malformations involving the skin, gastrointestinal tract, and, occasionally, other visceral organs. While most patients present with anemia due to occult GI bleeding, systemic manifestations may include menorrhagia, hematuria, hemoptysis, or neurological symptoms. Here, we report a complex case of BRBNS in a middle-aged Indian woman with multisystem involvement and an excellent therapeutic response to Everolimus.

## Case presentation

A 36-year-old woman presented with gross hematuria and generalized fatigue for 3–5 days. Her medical history was notable for severe iron-deficiency anemia and seven prior transfusions over the last two years. Obstetric history revealed recurrent first-trimester miscarriages despite two successful deliveries. Physical examination revealed severe pallor and 16 bluish-violaceous, compressible cutaneous lesions (0.5–4 cm) on her palms, soles, limbs, abdomen, back, and oral mucosa. Laboratory workup confirmed microcytic hypochromic anemia (Hb 4.1 g/dL), positive fecal occult blood, and hematuria. Initial management included blood transfusions and IV iron. Imaging and endoscopy revealed venous malformations in the GI tract, liver, adrenal glands, urinary bladder, skeletal muscle, and brainstem. Cystoscopy identified active bladder lesions, which were treated with sclerotherapy. Upper and lower GI endoscopy revealed venous blebs in the esophagus, stomach, and colon. Based on these findings, a diagnosis of BRBNS was made. Given her history of severe anemia and multisystem involvement, Everolimus 0.5 mg twice daily was initiated. Over six months of follow-up, her hemoglobin stabilized between 9.5-11 g/dL without need for further transfusions or hospitalizations.



**Figures 1-3:** Cutaneous lesions over palm, back, limbs.

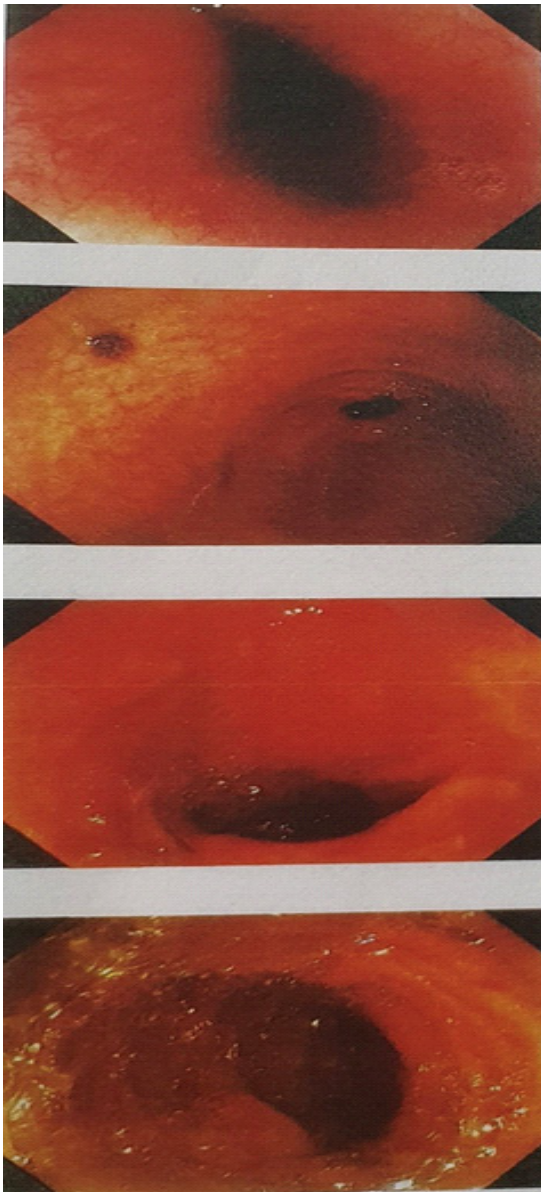


**Figure 4:** Upper GI endoscopy showing gastric venous blebs.

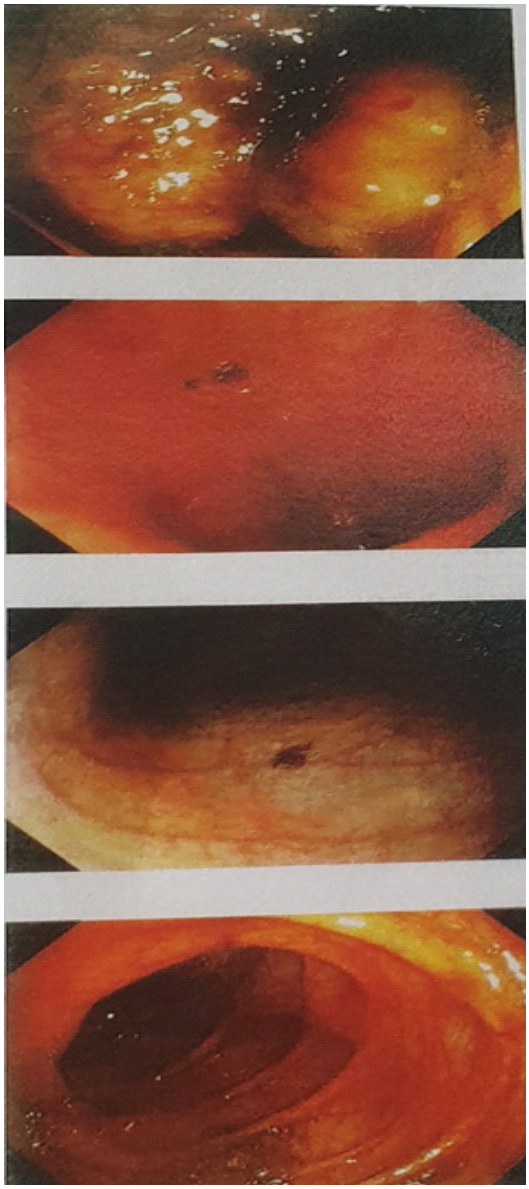


**Figure 5:** Colonoscopic image showing colonic venous blebs.

## Discussion

BRBNS presents a diagnostic and therapeutic challenge due to its variable presentation and multisystem involvement. GI bleeding remains the most common complication, requiring thorough endoscopic and radiological evaluation. While conservative measures including transfusions and iron supplementation are the mainstay, interventional procedures (e.g., sclerotherapy) and systemic therapies like mTOR inhibitors are increasingly utilized. Everolimus, an mTOR pathway inhibitor, suppresses angiogenesis and has demonstrated benefit in controlling lesion growth and hemorrhage. In our patient, Everolimus resulted in marked clinical improvement, reinforcing its role as a viable non-invasive alternative in complex BRBNS.

## Conclusion

This case highlights the importance of a high index of suspicion, especially in patients with unexplained anemia and characteristic skin lesions. Everolimus therapy was effective in stabilizing hemoglobin levels and reducing transfusion dependence. Timely diagnosis and individualized management can significantly improve patient outcomes in BRBNS.

## Declarations

**Patient consent:** Informed written consent was obtained from the patient.

**Conflict of interest:** None declared.

**Funding:** None.

## References

1. Jin XL, et al. Blue rubber bleb nevus syndrome: a case report and literature review. World J Gastroenterol. 2014; 20(45): 17254-9.
2. Aravindan U, et al. Surgery for Blue Rubber Bleb Nevus Syndrome—a Case Report. Indian J Surg. 2018; 80(3): 272-274.
3. Kaur T, Singh S. Blue rubber bleb nevus syndrome: a case report. Indian J Dermatol. 2014; 59(1): 98-9.
4. Baigrie D, et al. Blue Rubber Bleb Nevus Syndrome. StatPearls [Internet]. 2024 Jan.
5. Dobru D, et al. Blue Rubber Bleb Nevus Syndrome: Case Report and. Rom J Gastroenterol. 2004; 13(3): 237-240.
6. Agnese M, et al. Blue rubber bleb nevus syndrome. Acta Pædiatrica. 2010; 99: 632–635.
7. Yuksekkaya H, et al. Blue rubber bleb nevus syndrome: successful treatment with sirolimus. Pediatrics. 2012; 129(4): e1080-e1084.