Blue rubber bleb nevus syndrome (BRBNS) is a congenital disorder with characteristic venous anomalies that can present with varying degree of blood loss. The most clinically significant symptoms in adults include gastrointestinal (GI) bleeding and iron deficiency anemia. Severe complications can include intestinal torsion, intussusception, and even perforation, with each leading to significant morbidity and mortality. This report serves to give a brief understanding of this rare disease along with current diagnostic and therapeutic options.

Keywords: bleb; nevus; blue rubber bleb; gastrointestinal; bleeding; anemia

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lue rubber bleb nevus syndrome (BRBNS) is a rare disorder, first described by Gascoyen in 1860, but was later made famous by William Bennett Bean in 1958 and called ‘bean syndrome’. It causes venous malformations of the skin and other organs. This congenital disorder can manifest at any age and has no sex predilection (1). Patients with BRBNS may present with chronic anemia with or without cutaneous manifestations and a history of blood transfusions. The morbidity and mortality of this disease is related to the location of the malformations: primarily, the gastrointestinal tract (GIT) and uncommonly, the skin. A retrospective analysis of 120 BRBNS cases reported the GIT is involved in over 75% of cases and about 7% without any skin manifestations (2). The treatment approach to intestinal BRBNS has ranged from conservative to interventional treatment, depending on the severity and nature of the lesions.

Case

A 76-year-old male with a past medical history of coronary artery disease, status post coronary artery bypass graft and chronic anemia presented to the emergency department after he developed episodes of chest pain and pre-syncpe. He denied any non-steroidal drug or alcohol use.

On presentation, he was normotensive with a sinus tachycardia. His hemoglobin was 6.1 mg/dL, and the platelet count was 202.0 mg/dL. Initial fecal occult blood test was negative. He received multiple blood transfusions for his symptomatic anemia with a goal hemoglobin of greater than 7.0 mg/dL. There was a transient increase in his hemoglobin. A computerized tomography (CT) scan of the abdomen and pelvis without contrast did not reveal any bleeding source.

The following day the nurse witnessed him having a significant amount of hematochezia. Subsequent upper and lower endoscopy was done that revealed two 5 mm blue venous blebs in the esophagus and multiple patches of blue venous blebs throughout the colon (Figs. 1 and 2). Given the absence of active bleeding, no endoscopic intervention was undertaken. Through the course of his hospitalization, the patient received a total of five units of blood. He was managed medically with oral iron and periodic monitoring of his hemoglobin.

One year after initial diagnosis, the patient is doing well on oral iron supplementation, and his hemoglobin is being closely monitored by his primary care physician.

Discussion

Given our patient’s history of his smoldering anemia and characteristic endoscopic findings, he was suspected to have BRBNS.

BRBNS can be an indolent syndrome that presents with acute gastrointestinal (GI) bleeding, the small bowel being the most commonly affected. The etiology remains
poorly understood. The diagnosis is based on clinical history, clinical exam, and objective findings. Unlike infantile hemangiomas, these nevi can first appear in adulthood making an early diagnosis challenging (3). Histologically, it is composed of dilated venous tissue with a thin layer of endothelial cells (4).

Given the variable organs affected and variable presentation, there is no standard diagnostic test. For small bowel evaluation, it is best to use CT enterography, MRI, double-balloon enteroscopy, or capsule endoscopy (5–7). Since venous malformations are not typically transmural lesions, non-invasive imaging often underestimates the extent and quantity of GI lesions (8). Surgical exploration would have higher yield for extra-luminal lesions but should not be used as a common diagnostic modality. Tagged RBC nuclear scans have been shown to outline the extent of disease throughout the body and identify active bleeding; however, the scans can fail to identify multiple lesions (9). The diagnostic yield of tagged RBC nuclear scan can be improved when used with MRI findings. Endoscopy remains a common and readily used method for evaluating the colon. In this patient, CT imaging did not yield any intraluminal small or large bowel vascular abnormalities.

Currently, there has been no consensus on a treatment of choice. There is a high recurrence rate with GI bleeding. Intestinal nevi increase in quantity with age, increasing the risk of recurrence. As such, conservative treatment with blood transfusions and iron supplementation are done for minor, intermittent intestinal bleeding. In the setting of significant and persistent bleeding or complications that include intestinal torsion, intussusception, or intestinal perforation, interventional methods are often needed. These include endoscopic sclerotherapy, band ligation, or surgical resection (10, 11). Resection may be a preferred option in adults with numerous lesions, with local disease.

Endoscopic interventions, such as band ligation, argon plasma coagulation, laser photocoagulation, polypectomy with electrocauterization, and sclerosis for other GI lesions, have been utilized in numerous case reports with variable outcomes (12–14). These reports indicate thermal damage in the small bowel, which can result in perforation.

A randomized trial comparing interventional with conservative therapy with intestinal BRBNS in adult populations would help unify a treatment approach and regimen but remains a challenge given the rarity of this disease. A literature review of adults with intestinal BRBNS remains supportive of a conservative approach with escalation of care when clinically indicated. In cases of profound bleeding or being refractory to conservative measures, it may be reasonable to proceed with endoscopic intervention or surgical resection.

**Conclusions**

In adults without cutaneous manifestations, BRBNS should not be excluded as a possible differential diagnosis in setting of chronic and even acute symptomatic anemia. Several diagnostic methods such as imaging and endoscopy may be needed to make the diagnosis. In treating intestinal BRBNS, a non-invasive approach is preferred with escalation of care if refractory to conservative measures. In cases of profound bleeding, it may be reasonable to proceed with endoscopic intervention or surgical resection. A randomized trial comparing interventional with conservative approach would help unify a treatment approach and regimen but remains a challenge given the rarity of this disease.
therapy with intestinal BRBNS in adult populations would help unify a treatment approach and regimen, but remains a challenge given the rarity of this disease. By increasing awareness, incidence is expected to increase with the hope that this will increase data regarding management in an effort to identify the optimal treatment for the most common manifestations of this rare syndrome.

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