International Journal of Medicine and Medical Sciences Vol. 1(2), pp. 035-037, 27 December, 2011 © International Scholars Journals (http://internationalscholarsjournals.org)

Case Study

Blue Rubber Bleb Nevus Syndrome: An Unusual Cause of Abdominal Pain

Raj Majithia MD, Michael Gold MD FAGA

Division of Gastroenterology, Washington Hospital Center 110 Irving St., N.W. Suite 3A3 Washington, DC 20010 Phone: (202) 877-9074
Michael Gold MD FAGA Division of Gastroenterology, Washington Hospital Center 110 Irving St., N.W. Suite 3A3 Washington, DC 20010 Phone: (202) 877-9074
Corresponding Author's Name: raj.majithia@gmail.com

Received October 27, 2011; Accepted December 23, 2011

We present a case of a 22 year old female who presented to the hospital with abdominal pain and iron deficiency anemia. She had an esophagogastroduodenoscopy (EGD) that revealed blue rubber bled nevus syndrome (BRBNS), a rare condition characterized by multiple relapsing and remitting cutaneous venous malformations in association with visceral lesions most commonly affecting the GI tract. In 1860, Gascoyen first described an association between cavernous hemangiomas of the skin and similar lesions in the GI tract. In 1958, Bean further described these lesions and coined the term blue rubber bleb nevus syndrome. Chronic iron deficiency anemia is the most common complication. The patient's abdominal pain and anemia were due to the intraluminal thrombus from a previously bleeding bleb. She recovered from the hospitalization and has been undergoing repeated transfusions to correct her anemia.

Keywords: Blue rubber bleb nevus syndrome, abdominal pain, gastrointestinal bleeding, EGD, Bean syndrome, cavernous hemangioma.

INTRODUCTION

Blue rubber bleb nevus syndrome (BRBNS) is a rare gastrointestinal disease chatracterized by the association between cavernous hemangiomas of the skin and similar lesions in the GI tract.[1]These lesions have a tendency to bleed and can cause chronic iron deficiency anemia or even require significant transfusions.[2] We present an unusual case of abdominal pain associated with iron deficiency anemia. The patient underwent an esophagogastroduodenoscopy (EGD) and was endoscopically noted to have vascular lesions consistent with BRBNS. The case report will discuss the presentation, diagnosis and management of BRBNS.

Case Study

A 22-year-old African American female presented to the Emergency Department with abdominal pain and nausea for the past 24 hours. She denied all other symptoms including weight loss, fevers, chills, recent travel, chest pain, jaundice. She did complain of intermittent melena for the past 3 months. Her physical examination revealed a significantly tender right upper quadrant without a palpable liver or spleen. Murphy's sign was not present. She had no physical exam findings to suggest cirrhosis. On her skin exam, there were multiple small hemangiomas found on her anterior thighs and in her axilla. Laboratory evaluation was significant for a hemoglobin of 7.1 mg/dL (nml 12.5-16.5 mg/dL) and a ferritin of 6 (nml 10-148 ng/mL).

An EGD was performed to rule out gastric pathology as the cause of her melena and abdominal pain. On her EGD she was found to have a medium-sized, blue tinged submucosal mass found 30 cm from the

Figure 1. Submucosal blue rubber nevus of the lower esophagus

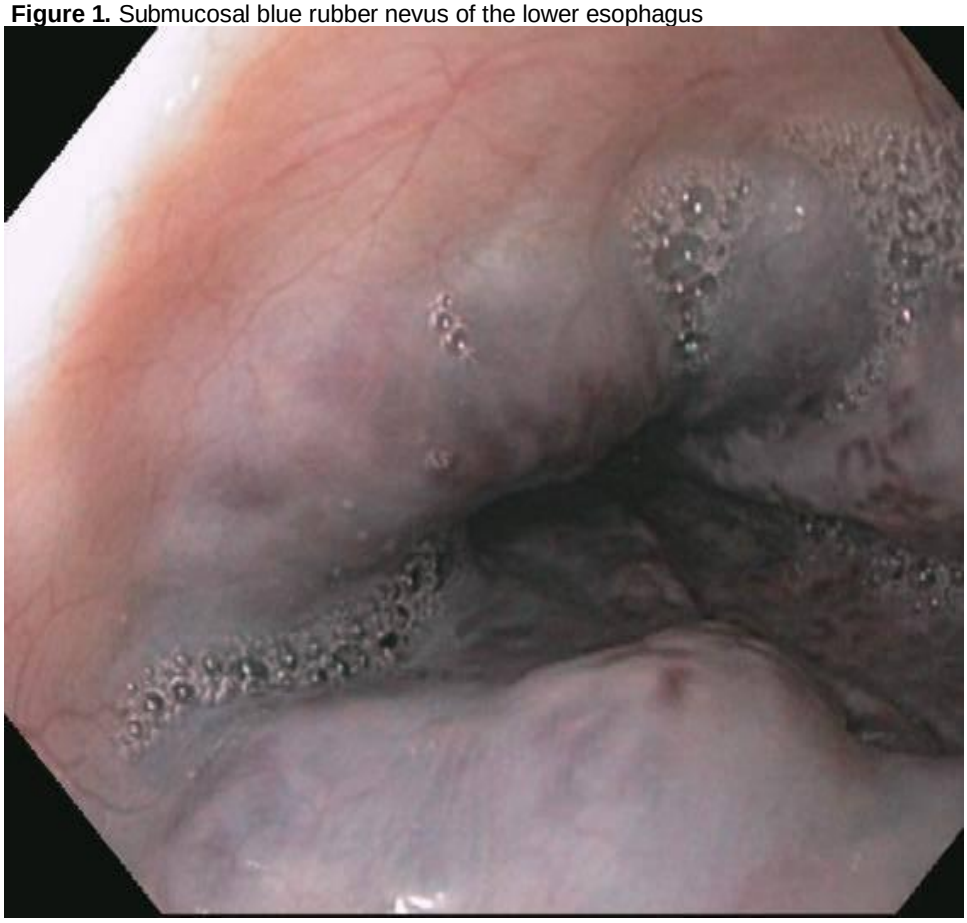


Figure 2. Blue Rubber Nevus of the duodenum

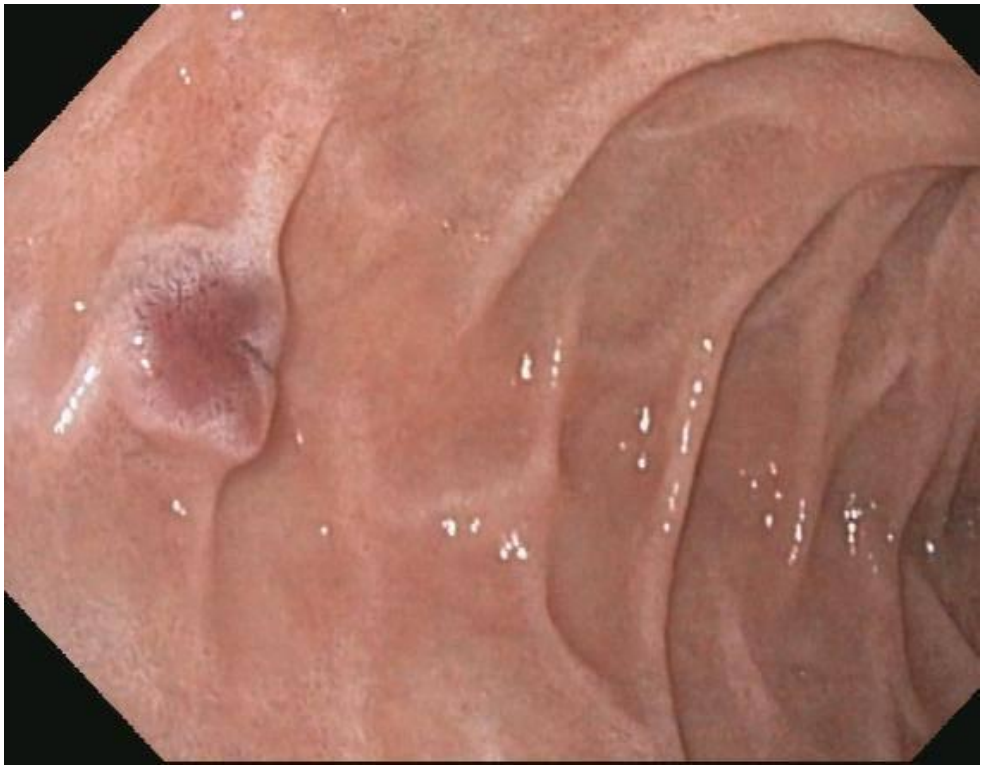
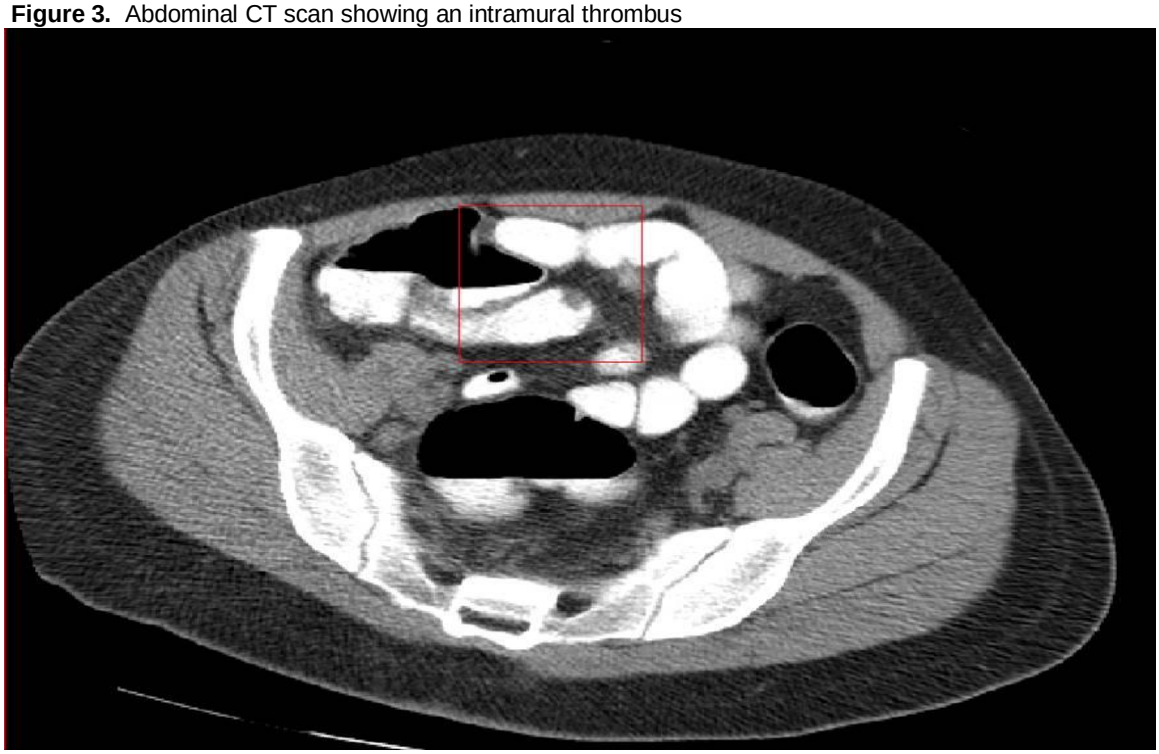


Figure 3. Abdominal CT scan showing an intramural thrombus



incisors. The mass was partially obstructing consistent with a submucosal blue rubber nevus (Refer to Figure 1). Additionally, there were multiple small to medium-sized submucosal masses found in the duodenum including the duodenal bulb (Refer to Figure 2). The findings on endoscopic and dermatologic examination were consistent with a diagnosis of BRBNS. Computed Tomography (CT) of the abdomen revealed a possible intramural filling defect which was consistent with thrombi from previously bleeding nevi. The patient's abdominal pain was diagnosed as being due to an intramural bleed from a blue rubber nevus which resolved without further intervention. The patient was discharged to follow up for hemoglobin checks and iron infusions.

DISCUSSION

In 1860, Gascoyen first described an association between cavernous hemangiomas of the skin and similar lesions in the GI tract. In 1958, Bean further described these lesions and coined the term blue rubber bleb nevus syndrome. He described lesions that looked and felt like rubber nipples. BRBNS is thought to inherited in an autosomal dominant fashion, however, sporadic cases have been reported. The disease has mostly been reported in Whites and Japanese patients and to a much lesser extent, African Americans. Cutaneous lesions are most often noted at birth, but have been shown to present in adulthood as well.[3]

Blue Rubber Bleb Nevus Syndrome is characterized by multiple relapsing and remitting cutaneous venous malformations in association with visceral lesions most commonly affecting the GI tract. It is a rare disorder with approximately 150-200 cases reported in the literature. There have been some reports of associations with medulloblastoma, renal cell carcinoma, squamous cell cancer of the lung, however, the causality has not been established. Chronic iron deficiency anemia is the most common complication with occasional severe gastrointestinal bleeding.[4] Interestingly, our patient presented with abdominal pain and prior to this had maintained iron stores to avoid the transfusion requirement up to this admission.

Treatment for BRBNS is conservative whenever possible. The cutaneous manifestations of the disease can be monitored without treatment as treatment is generally reserved for cosmetic purposes. Observation and iron supplements are adequate in

most cases where the patient is asymptomatic or only has mild symptoms.[5]

Due to the rarity of cases, there has been no published standardized method of management to our knowledge. Based on a literature review of case reports, patients have been successfully treated in the short term with octreotide, argon plasma coagulation, and interferon beta. All of these case reports followed the patient for short periods after treatment with the longest duration lasting 4 weeks. If gastrointestinal ligation. Preliminary results show that from those who underwent aggressive intervention, 11 of 13 patients had gone without transfusion for 5 years. This approach is debated however, due to the ability in a majority of patient to avoid massive transfusions, as well as the morbidity of such invasive procedures.

We present an uncommon presentation of BRBNS as the cause of right upper quadrant abdominal pain.

Despite the rarity of the disease, we feel that this case highlights the importance of considering it in the

differential of an otherwise health patient who presents with iron deficiency anemia and abdominal pain.

malformations are confined to one segment of the intestinal tract, resection may provide longlasting resolution of bleeding, however the data to support this approach is limited.[6]

Fishman and colleagues reported an aggressive endoscopic and surgical approach.[7] Gastrointestinal venous malformations were identified endoscopically and then treated with wedge resection, polypectomy, suture ligation, segmental bowel resection, and band

REFERENCES

Ertem D, Acar Y, Kotiloglu E, (2001). Blue rubber bleb nevus syndrome. Pediatrics. Feb;107(2):418-20

Fishman SJ, Smithers CJ, Folkman J, Lund DP, Burrows PE, Mulliken JB (2005). Blue rubber bleb nevus syndrome: surgical eradication of gastrointestinal bleeding. Ann Surg. 241(3):523-8.

Nahm WK, Moise S, Eichenfield LF (2004). Venous malformations in blue rubber bleb nevus syndrome: Variable onset of presentation. J. Am' Acad Dermatol;50:S101-6

Oranje, AP. Blue Rubber Bleb Nevus Syndrome. Pediatric Dermatol. 1986. 3: 304–310.

Rodrigues D (2000). Blue Rubber Bleb Nevus Syndrome. Rev. Hosp. Clín. Fac. Med. S. Paulo. 55 (1):29-34.

Wong CH, Tan YM, Chow WC, (2003). Blue Rubber Bleb Nevus Syndrome: A Clinical Spectrum with Correlation Between Cutaneous and Gastrointestinal Manifestations. J Gastroenterol. Hepatol.18(8):1000-2

Wong W, Cheung C, Lau K (2001). A rare cause of gastrointestinal bleeding: Blue rubber bleb nevus syndrome. Annals of the College of Surgeons of Hong Kong; 5(1):25-8.