Rectal Blue Nevus: Distinguishing Features of a Rare Entity

Neena Mohan, MD¹, Christina Tofani, MD², Peter McCue, MD³, and Daniel Quirk, MD, MBA²

¹Division of Internal Medicine, Department of Medicine, Thomas Jefferson University Hospital, Philadelphia, PA
²Division of Gastroenterology and Hepatology, Department of Medicine, Thomas Jefferson University Hospital, Philadelphia, PA
³Department of Pathology, Thomas Jefferson University Hospital, Philadelphia, PA

ABSTRACT

A 26-year-old African American man with a history of depression and tuberculosis presented to the gastroenterology department after several months of rectal pain with bowel movements. Colonoscopy revealed hyperpigmentation in the distal rectum and internal hemorrhoids, which resulted in a diagnosis of blue nevi. This is only the third known description of a blue nevus involving the gastrointestinal mucosa.

INTRODUCTION

A blue nevus is a pigmented skin lesion affecting the dermal melanocytes. While its occurrence on skin is well described in the literature, involvement of the mucosal surfaces is less commonly described. To our knowledge, there were only two known descriptions of a blue nevus involving the rectum prior to this case.¹ ²

CASE REPORT

A 26-year-old African American man with a history of depression and tuberculosis presented to the gastroenterology department after several months of rectal pain with bowel movements. He reported that his pain occurred about twice a week and was on the right side of his rectum; the pain resolved after a bowel movement. He usually had one bowel movement per day, with varied consistency. Rectal pain was exacerbated by passage of hard or more voluminous stool. He noted slightly increased frequency in bowel movements over the past few months and noticed some dark red blood in his stool; these symptoms started around the same time as the pain. The patient denied abdominal pain and had no change in appetite. He had not had any recent weight loss and denied dysphagia, odynophagia, rashes, fevers, and rigors. There was no family history of colon cancer or inflammatory bowel disease.

On exam, there were normal bowel sounds and a soft, non-tender, non-distended abdomen without hepatomegaly or splenomegaly. Rectal exam revealed a small external hemorrhoid. No fissures or masses were noted. His stool was guaiac-negative with positive control. The patient was scheduled for a colonoscopy for further evaluation of pain and dark blood in stool, and he was started on zinc sulfate suppositories and fiber supplements for management of hemorrhoids and constipation.

Colonoscopy revealed hyperpigmentation in the distal rectum and internal hemorrhoids (Figure 1). Two grayish soft tissue biopsy fragments, 0.4 and 0.2 cm in greatest dimension, showed positive staining of HMB45, S100, and Melan-A, pathological features of rectal blue nevi (Figure 2).

DISCUSSION

The origin of blue nevi cells is unclear. The most commonly held theory is that they originate from migrating neural crest cells. Melanocytes that comprise the blue nevus contain varying amounts of melanin, which gives the
characteristic blue color. The differential diagnosis of blue nevi includes pigmented scars, postinflammatory hyperpigmentation, drug-induced pigmentation, and regressing nevi. Differentiation of blue nevi from melanoma is very important. Desmoplastic melanoma can closely imitate blue nevi, especially its sclerosing variant. Presence of tumor-associated lymphocytic infiltrate and perineural invasion are strongly suspicious for melanoma. Also in contrast to blue nevi, most desmoplastic melanomas are HMB45-negative. S100 is a sensitive marker of melanocytic differentiation in benign melanocytic lesions, primary cutaneous malignant melanoma, metastatic melanoma, and desmoplastic melanoma. Melan-A is a supportive marker to S100 for melanocytic lesions and has greater sensitivity for malignant melanoma than HMB45.

There are two conventional histological variants of the blue nevus, which are the common blue nevus and the cellular blue nevus. The common blue nevus predominantly affects young females and is most often found on the extremities, hips, and scalp. Cellular blue nevus usually affects individuals before their fifth decade and is most commonly found on the sacrum. The cellular blue nevus tends to be larger than the common blue nevus and can grow up to 2 cm in size.

In general, blue nevi behave in a benign manner, and regional lymph node involvement is rare. Very rare cases of persistent recurrence of blue nevi have been reported, often after incomplete excision. “Malignant blue nevus” is a term that was first coined to describe blue nevus-like lesions that had resulted in metastasis and, in some cases, patient death. It has been used by various authors to denote entities including malignant changes arising in cellular blue nevus, melanoma with features resembling cellular blue nevus but apparently arising de novo, or melanoma with an admixed residual benign cellular blue nevus component. Murali et al. note that it is more appropriate to term such lesions as “blue nevus-like melanoma (BNLM) or atypical blue nevus-like lesions of uncertain malignant potential,” as the term nevus denotes a benign lesion. Most of these cases are associated with cellular blue nevus or with cellular blue nevus in combination with other nevus types. BNLM is rare and occurs most often in older individuals (often >45 y) and are usually large (3–13 cm). Many of the reported cases exhibit local invasion or widespread metastasis, with the scalp being the most common site, followed by the face, buttocks, and chest. A comprehensive analysis of 23 patients with BNLM found that clinical outcome was similar to that of conventional melanoma when matched for tumor thickness and other important prognostic factors.

Blue rubber bleb nevus syndrome (BRBNS) is a rare disease of multiple venous malformations and hemangiomas in the skin and visceral organs, often involving the cutaneous and gastrointestinal (GI) systems. The most common symptoms are GI bleeding and secondary iron deficiency anemia. An analysis of 120 cases found that cutaneous angiomas were observed in 93% of cases and GI hemangiomas were seen in
76% of cases. Histologic exam of BRBNS lesions reveals cavernous venous dilations, with a thin wall of smooth muscle cells lined by endothelial cells. While lesions are often present from birth or early childhood, cases of adult-onset BRBNS have been reported. The treatment of BRBNS depends on the extent of intestinal involvement and the severity of disease, ranging from conservative management with blood transfusions and iron supplements to surgical resection. In our patient’s case, the rectal pain and mild bleeding were likely secondary to hemorrhoids. Only one area of hyperpigmentation was noted, and there was no other evidence of BRBNS.

Our case is only the third reported case of rectal blue nevus and was found in a young male patient. Due to the rarity of its occurrence, the clinical course of the rectal blue nevus is mostly unknown. While it is a benign lesion, there are concerns about its differentiation from or transformation to a malignant lesion. Therefore it is recommended to completely remove rectal blue nevi during a colonoscopy or monitor closely if not removed. Pathology should be evaluated to confirm the absence of features such as cellular atypia, pleomorphism, and increased mitoses.

DISCLOSURES
Author contributions: N. Mohan drafted the manuscript, C. Tofani and D. Quirk reviewed the manuscript, and P. McCue performed the pathology review. C. Tofani is the article guarantor.

Financial disclosure: None to report.

Informed consent was obtained for this case report.

Received April 22, 2016; Accepted July 7, 2016

REFERENCES
1. Makker J, Sakam S, Arety P, et al. Rectal blue nevus: Case report of a rare entity and literature review. Pathol Res and Pract. 2015;211(8):625-7.
2. Schreiber ZJ, Pal TR, Hwang SJ. Blue nevus of the colorectal mucosa. Ann Diagn Pathol. 2011;15(2):128-30.
3. Phadke PA, Zembowicz A. Blue nevi and related tumors. Clin Lab Med. 2011;31(2):345-58.
4. Orchard GE. Comparison of immunohistochemical labelling of melanocyte differentiation antibodies Melan-A, tyrosinase and HMB45 with NRIC3 and S100 protein in the evaluation of benign naevi and malignant melanoma. Histoch J. 2000;32(8):475-81.
5. Zembowicz A, Phadke PA. Blue nevi and variants: An update. Arch Pathol Lab Med. 2011;135(3):327-36.
6. Gonzalez-Campora R, Galera-Davidson H, Vazquez-Ramirez FJ, et al. Blue nevus: Classical types and new related entities. A differential diagnostic review. Pathol Res Pract. 1994;190(5):627-35.
7. Murali R, McCarthy SW, Scolyer RA. Blue nevi and related lesions: A review highlighting atypical and newly described variants, distinguishing features and diagnostic pitfalls. Adv Anat Pathol. 2009;16:365-82.
8. Jin LX, Wang ZH, Xiao XB, et al. Blue rubber bleb nevus syndrome: A case report and literature review. World J Gastroenterol. 2014;20(49):17254-9.