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CASE REPORT

Gastroenterology

Mucosal prolapse syndrome mimicking Peutz–Jeghers syndrome in a pediatric patient

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Abstract

Mucosal prolapse syndrome (MPS) is a rare group of benign conditions characterized by a set of unifying histologic findings thought to be the result of repeated mucosal shearing and submucosal vascular congestion caused by straining. This set of conditions is often misdiagnosed as other polyposis syndromes, inflammatory bowel disease, or malignancy due to its clinical presentation, appearance, and rarity. We report a case of a 15-year-old male who presented with painless rectal bleeding. He was found to have four rectal polyps thought to be due to Peutz–Jeghers syndrome. A repeat colonoscopy with biopsies a year later revealed a diagnosis of MPS. Our case highlights the morphologic similarity between hamartomatous polyp and mucosal prolapse histology. Since MPS is a rare diagnosis even among the adult population, it has not been well described in pediatrics. This syndrome should be on the differential diagnosis for pediatric rectal polyps to prevent unnecessary invasive testing and a delay in treatment.

KEYWORDS

constipation, polyposis, solitary rectal ulcer

1 | INTRODUCTION

Mucosal prolapse syndrome (MPS) is a rare group of conditions including solitary rectal ulcer syndrome (SRUS), gastric antral vascular ectasia, inflammatory cloacogenic polyps, inflammatory cap polyps, and prolapsing mucosal polyps.[1,2] With a reported incidence of 1 in 100,000 in the adult population, reports of MPS in the pediatric population are sparse. Though this is a benign entity thought to be the result of repeated straining to cause mucosal shearing, submucosal vascular ischemia, and venous congestion, it is often misdiagnosed as inflammatory bowel disease, polyposis syndrome or malignancy due to its rarity, endoscopic and histologic appearance.[3,4] Patients most commonly present with blood per rectum, which can sometimes be accompanied by mucous. A careful history will often elicit reports of straining, constipation, or small, frequent bowel movements.[5] The endoscopic appearance can be variable, ranging from mucosal erythema to single or multiple polypoid lesions and rectal ulcers.[5] The diagnosis can be made when a history of frequent straining is observed in the setting of appropriate histologic characteristics such as crypt distortion, proliferation of fibromuscular cells in the lamina propria, inflammatory cell infiltration of the lamina propria, thickened and distorted muscularis mucosae, dilated capillaries, and a villous appearance of the epithelial surface. Histologically, mucosal prolapse polyps can share the smooth muscle arborization pattern that is characteristic of colonic Peutz–Jeghers polyps.[6] Here, we present a case of a teenager with hematochezia, which highlights the diagnostic challenge that MPS presents due to its shared clinical, endoscopic, and histologic features with other pathologic conditions.

2 | CASE REPORT

A 15-year-old previously healthy male presented for evaluation of 1.5 years of epigastric pain, prolonged straining during defecation, and hematochezia (visualized blood streaks on the toilet paper). Family history was notable for benign colonic polyps in mother and maternal grandmother. He had a history of constipation although complained of multiple loose stools a day at the time of the first visit. His first colonoscopy showed four pedunculated polyps in the rectum, one of which had ulceration (Figure 1A,B). Microscopic examination of these polyps revealed elongated crypts with villiform changes and smooth muscle in the stroma between the lobules of overgrown glandular elements (Figure 2A,B). Though he had no history of hyperpigmented macules and other exam findings suggestive of Peutz–Jeghers syndrome (PJS), histologic findings were thought to be consistent with Peutz–Jeghers Polyps so the diagnosis of PJS was made based on histology. He underwent genetic testing for polyposis syndromes, which was negative. He continued to have intermittent blood in the stool, epigastric abdominal pain, and worsening constipation. Repeat colonoscopy 1 year later showed erythematous and edematous rectal mucosal folds with friability without discrete polyps (Figure 1C). Histologic evaluation showed crypt distortion, an inflammatory infiltrate of the lamina propria, and smooth muscle cells in the lamina propria on rectal mucosal biopsy (Figure 2C–E). Review of the previously removed polyps indicated that their histology was more compatible with inflammatory cloacogenic polyps, especially in light of negative genetic testing for PJS. He was diagnosed with MPS and treated with polyethylene glycol 3350 and a fiber supplement. Due to persistent constipation, treatment was changed to linaclotide which resulted in the resolution of abdominal pain, hematochezia, constipation, and straining complaints at 1.5 years from the change in diagnosis.

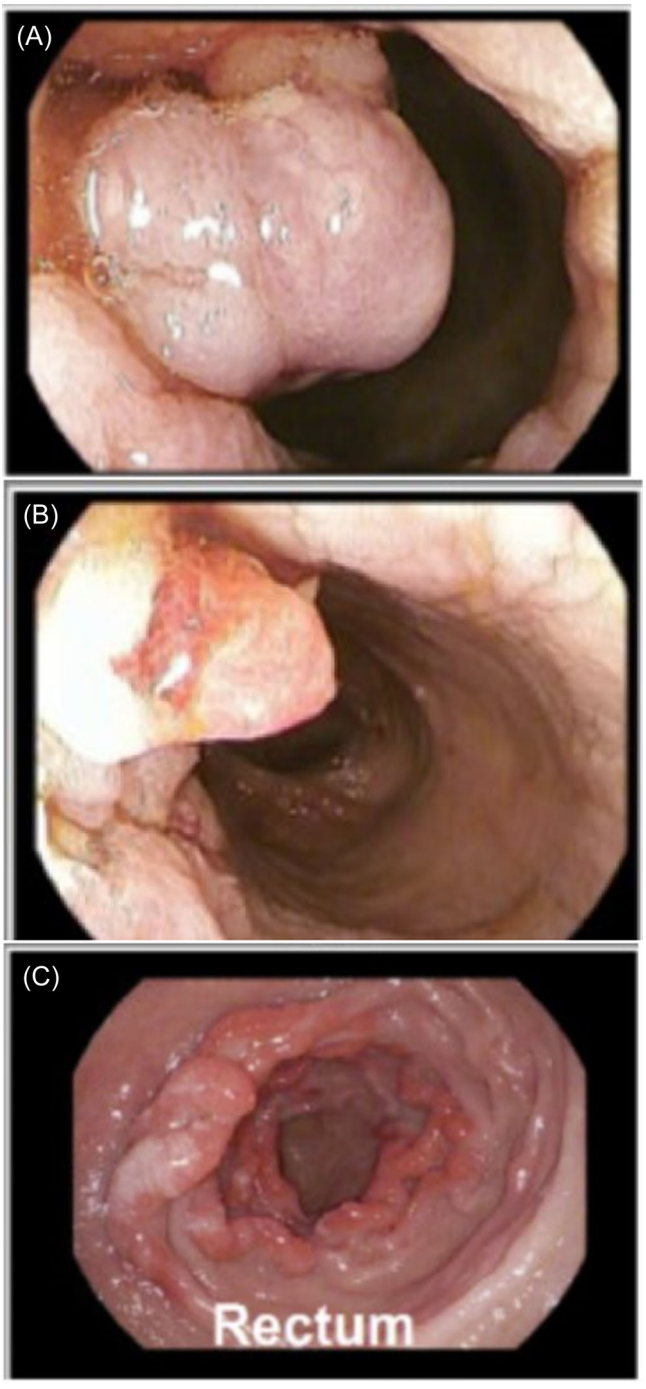

FIGURE 1 Endoscopic findings. First endoscopy, (A) rectal pedunculated polyp, (B) rectal polyp with ulceration. Second endoscopy, (C) edematous and erythematous rectal mucosal fold without polyp.

3 | DISCUSSION

Our patient exhibits many features compatible with MPS such as history of constipation, frequent small stools, and rectal polyp with ulceration. However, many of his symptoms also overlap with more aggressive gastrointestinal diseases. Inflammatory bowel disease, colorectal malignancy, and polyposis syndromes are all on the differential diagnoses for hematochezia, abdominal pain, and abnormal bowel habits. Due to MPS's ability to mimic polyposis syndromes and other inflammatory and malignant conditions on histology, this case highlights the diagnostic difficulties that this condition presents and underscores the similarities and differences between MPS and PJS in a pediatric patient.[7]

PJS is an autosomal dominant condition caused by mutation of the *STK11/LKB1* gene. Even though mucocutaneous pigmentation and a positive family history can provide a clue to this diagnosis, a minority of patients with PJS may not exhibit either. Due to the high risk of gastrointestinal and extra-intestinal malignancies, it is crucial to make the correct diagnosis for

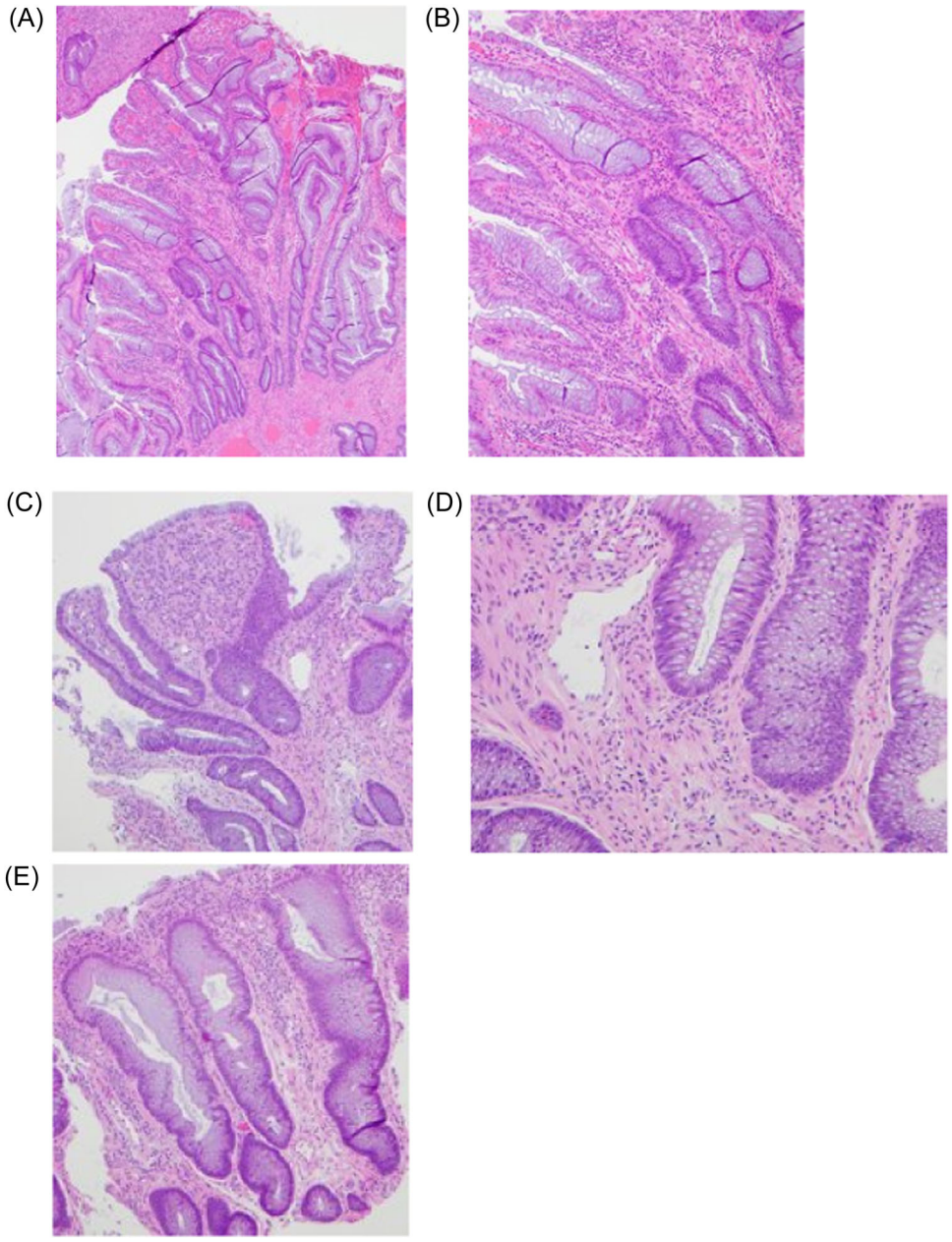

FIGURE 2 Histologic findings on hematoxylin and eosin (H&E) stain. (A, B) Polyp excised at first endoscopy. (A) H&E stained image depicts villiform surface with vague arborization, and (B) prominent smooth muscle infiltrating villous stroma. (C–E) Lesion excised at second endoscopy. (C) focal villiform change of surface, (D) dilated capillaries, and (E) fibromuscular proliferation.

future surveillance and genetic counseling. Often, the diagnosis is made following endoscopic evaluation based on histopathology by the presence of two or more Peutz–Jeghers-type polyps.

Historically, the histologic hallmark of Peutz–Jeghers polyps has been the smooth muscle arborization pattern within the lamina propria. However, recent studies have suggested that smooth muscle arborization is not the predominant pattern in colonic Peutz–Jeghers polyps, especially in those measuring less than 1 cm in diameter. Rather, the predominant pattern appears to be desmin-positive smooth muscle fibers surrounding lobules of colonic crypts.[6] On the other hand, smooth muscle arborization pattern may be found in colonic polyps such as hyperplastic polyps, tubular adenoma, and MPS polyps while the lobular pattern of smooth muscle is notably absent.[6] Diamond-shaped crypts and/or mucosal elastin have been shown to be present in all cases of MPS polyps and in some cases of large adenomas.[8] MPS polyps tend to show capillary ectasia, surface villiform changes, and increased fibrous tissue in lamina propria, which are present in the polyps excised at the second endoscopy of our patient. Table 1 highlights some known histologic characteristics of Peutz–Jeghers polyps versus MPS

TABLE 1 Histological differences between Peutz–Jeghers and mucosal prolapse syndrome polyps.

Histological findings		
Intestinal location	**Peutz–Jeghers**	**Mucosal prolapse syndrome**
Small intestinal polyps	Smooth muscle arborization within lamina propria	Not applicable
Colonic polyps/ lesions	Smooth muscle arborization is frequent and prominent	Fibromuscular cell fascicles in vertical orientation between crypts. Smooth muscle arborization can be seen, but uncommon.
	Lobular arrangement of crypts surrounded by smooth muscle fibers is dominant pattern, especially in polyps <1 cm	Crypts not organized in lobular pattern
	Usually no significant inflammatory cells in lamina propria	Inflammatory cell infiltration of the lamina propria is common
	Crypt distortion and villous appearance of epithelial surface may be present	Crypt distortion, thickened and distorted muscularis mucosae, dilated capillaries, and a villous appearance of the epithelial surface

TABLE 2 Differences in clinical presentation between Peutz–Jeghers and mucosal prolapse syndrome.

	Peutz–Jeghers	**Mucosal prolapse syndrome**
Presenting symptom		
Obstruction	43%	Not available
Abdominal pain	23%	49%
Rectal bleeding	14%	82%–94%
Constipation	Not available	23%–27%
Diarrhea	Not available	22%–56%
Most commonly involved segment	Small intestine	Rectum

polyps. Future studies on these findings in patients with PJS will be helpful in providing additional histologic features to further differentiate the two conditions.

Despite clinical and endoscopic similarities between MPS and PJS, there are subtle differences that can give clues to the diagnosis (Table 2).[5,9,10]

Though historically a rare condition in the pediatric population, the increasing number of large case series in pediatric MPS in recent years suggests that this condition is encountered more frequently.[5,11] A high index of suspicion and attention to clinical, endoscopic, and histologic findings, are needed to limit costly and invasive testing and misdiagnosis.

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CONFLICT OF INTEREST STATEMENT

The authors declare no conflict of interest.

ETHICS STATEMENT

Informed patient consent was obtained for publication of the case details. The patient is over 18 years of age at the time of this submission and can legally provide consent.

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