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Rare Isolated Duodenal Hamartomatous Polyp in an Elderly Patient

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Data Collection B
Statistical Analysis C
Data Interpretation D
Manuscript Preparation E
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Patient: Female, 73-year-old
Final Diagnosis: Hamartoma • iron deficiency anemia
Symptoms: Fatigue
Clinical Procedure: Endoscopy
Specialty: Gastroenterology and Hepatology

Objective: Rare disease

Background: Hamartomatous polyps represent rare sporadic lesions, characterized by fibrous stroma, vascular infiltration, and dilation of mucous glands. The lesions present in a bimodal fashion in adults as well as children from 1 to 7 years old, and are often diagnosed during endoscopic procedures. Specifically, solitary Peutz-Jeghers represents a type of hamartoma that has similar histologic features to typical Peutz-Jeghers syndrome. Hamartomatous polyps represent disorganized tissue growth and can bear relationships with genetic syndromes classified as hamartomatous polyposis syndromes. A number of these syndromes, such as Peutz-Jeghers and Cowden syndrome, can demonstrate an increased risk of malignancy. A variety of symptoms, or no symptoms at all, can accompany these polyps, such as abdominal discomfort, bowel obstruction, gastrointestinal bleeding, or intussusception in severe cases. Histologically, these polyps appear similar to Peutz-Jeghers syndrome growths; however, they lack extraintestinal manifestations. Given fairly low risk of development into malignancy, patients have a good prognosis if presenting with a solitary hamartomatous polyp. There is limited data regarding screening guidelines for this patient population.

Case Report: Here, we present a rare case of a 73-year-old woman who had a history of anemia and status post endoscopic evaluation and was diagnosed with a benign hamartomatous polyp (juvenile-like), histologically consistent with tubulovillous adenoma.

Conclusions: Differentiating sporadic polyps from syndromic polyps is important, as sporadic polyps have a benign course, while those associated with a syndrome have an increased lifetime malignancy risk.

Keywords: Colorectal Neoplasms • Intestinal Polyps • Hamartoma • Duodenal Neoplasms • Gastrointestinal Hemorrhage

Full-text PDF: https://www.amjcaserep.com/abstract/index/idArt/938929

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Background

Hamartomatous polyps represent tissue growth of a particular organ that is disorganized in nature and mostly benign; however, malignant transformation can occur. Hamartomatous polyps are more common in the pediatric population, with a prevalence of 2%, compared to an incidence of 0.15% in the adult population [1,2]. The prevalence of hamartomatous polyps at index colonoscopy is approximately 0.073% [3]. When visualized via endoscopy, hamartomatous polyps can be sessile or pedunculated, which may obscure the clinical picture when differentiating these polyps from adenomatous polyps [4]. Sporadic polyps can arise anywhere along the gastrointestinal tract, including the gastric mucosa, and therefore, it is essential to remove such polyps to further histologically evaluate an accurate diagnosis [4]. Differential diagnosis of these polyps includes primary duodenal carcinoid tumors, which can present in the first or second part of the duodenum or the ampulla of Vader, with most masses encompassing less than 2 cm in size [5].

Case Report

A 73-year-old woman with a past medical history of recently diagnosed essential thrombocythemia was referred to Gastroenterology Clinic for low hemoglobin levels. Small bowel enteroscopy revealed a single 20-mm semi-pedunculated sessile polyp, which was sent for pathological analysis; there was no bleeding in the proximal duodenal bulb (**Figure 1**). Patchy mildly erythematous mucosa without bleeding was also found in the gastric antrum. Concomitant colonoscopy revealed a small sessile polyp at the base of the cecum (**Figure 2**), a diminutive polyp in the transverse colon, many small and large left-sided diverticula, and small internal hemorrhoids. Endoscopy showed gastric angioectasia. A polypectomy was performed for each noted polyp. Histopathological examination of the duodenal polyp revealed a benign hamartomatous polyp (juvenile-like), which was negative for adenomatous dysplasia. The patient's cecal polyp was histologically consistent with tubulovillous adenoma (**Figure 3**). Genetic mutations were not evaluated in this patient.

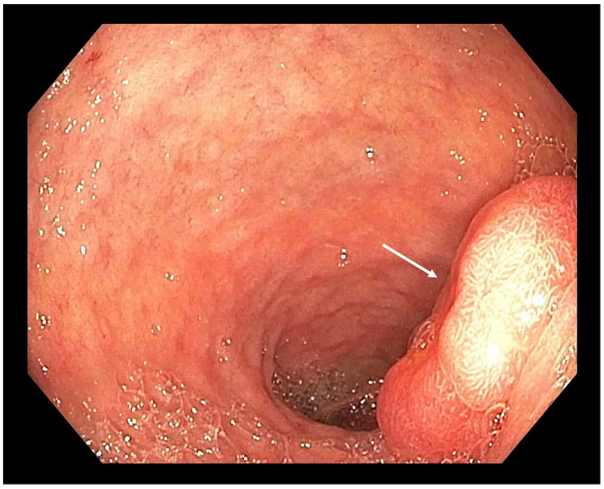

Figure 1. A single 20-mm semi-pedunculated sessile duodenal bulb polyp (white arrows) visualized from small bowel enteroscopy.

Discussion

A hamartomatous polyp is a disorganized growth of normal tissue that is native to the site of origin. These polyps typically occur in conjunction with genetic syndromes known as hamartomatous polyposis syndromes [1]. Hamartomatous polyposis syndromes include but are not limited to Peutz-Jeghers syndrome, Cowden syndrome, juvenile polyposis syndrome, and hereditary mixed polyposis syndrome [1]. Each of these syndromes has its own epidemiological factors as well as manifestations. Both Peutz-Jeghers and Cowden syndrome have an increased risk of malignancy; for example, Peutz-Jeghers syndrome is associated with gastric cancer, while endometrial and thyroid cancers are commonly seen in Cowden syndrome [6]. Although malignant transformation is rare, there is an increased risk of colon cancer in all patients with hamartomatous polyps that are associated with hamartomatous polyposis syndrome [7]. Compared with neoplastic or hyperplastic polyps, hamartomatous polyps are rare [1]. It is more common for hamartomatous polyps to be associated with one of the hamartomatous polyposis syndromes as opposed to presenting as a solitary lesion as seen with sporadic hamartomatous polyps [1]. There are 2 subtypes of sporadic hamartomatous polyps: Peutz-Jeghers type solitary polyps and juvenile solitary polyps [2].

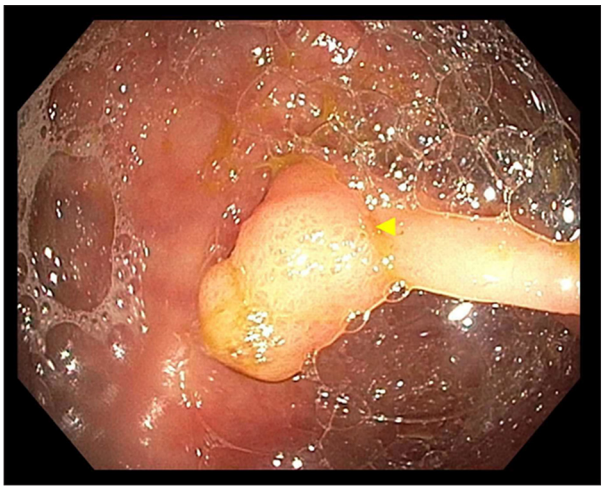

Figure 2. Small sessile polyp (yellow arrow) at the best of the cecum visualized on colonoscopy.

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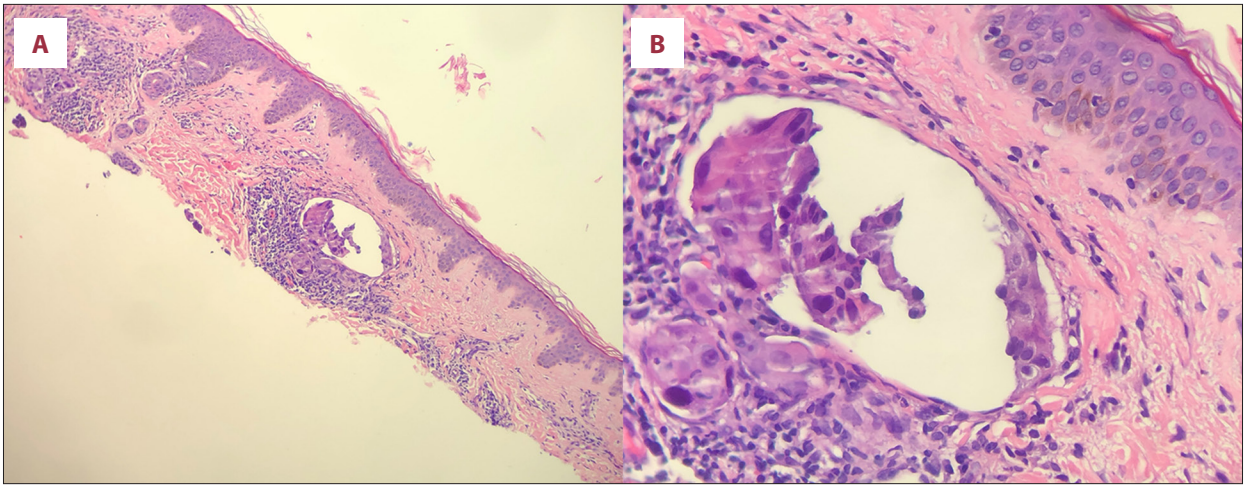

Figure 3. Duodenal bulb polyp, histologically consistent with benign hamartomatous polyp (juvenile-like), negative for adenomatous features or dysplasia (**A** at low magnification; **B** at high magnification).

Patients with hamartomatous polyps can present with a variety of symptoms, ranging from abdominal pain to gastrointestinal bleeding, and alternatively, patients can be entirely asymptomatic [2]. The presentation of hamartomatous polyps is similar to that of other colonic polyp subtypes, and hematochezia is a common symptom secondary to gastrointestinal bleeding, considering these polyps tend to be highly vascularized [8]. Patients commonly experience a change in bowel habits, whether that be an increase in diarrhea or constipation, and can experience abdominal pain [9]. Although it is not common, patients can present with bowel obstruction, considering large polyps can act as a lead point for intussusception [8]. Our patient presented with anemia secondary to gastrointestinal bleeding, which is a common presentation for many patients who have intestinal polyps, whether they are hamartomas in nature or not. For patients who present with anemia secondary to a possible gastrointestinal bleed, a diagnostic colonoscopy is advised. Typically, a hamartomatous polyp is visualized as a cherry red, smooth, pedunculated lesion [8]. It can be difficult to differentiate hamartomatous from other adenomatous subtypes of colonic polyps on macroscopy, and therefore, polypectomy and biopsy with the histological examination are often done to rule out malignancy and identify the subtype [8].

On histological examination, these polyps mostly consist of connective tissue composed of smooth muscle, the lamina propria, and inflammatory infiltrates that are covered by a hypertrophic epithelium [8]. Histologically, hamartomatous polyps are nearly identical to those seen in Peutz-Jeghers syndrome, but as opposed to patients with Peutz-Jeghers syndrome, these patients typically do not possess other extraintestinal manifestations [10]. These polyps can demonstrate epithelial overgrowth, with an arborizing core of smooth muscle on histologic examination [11]. Patients with solitary Peutz-Jeghers polyps typically do not have a genetic mutation of STK11/LKB-1 [11]. According to the American Gastrointestinal Association, genetic testing is indicated with the use of a multigene panel test if the patient has 2 or more lifetime hamartomatous polyps, a family history of hamartomatous polyps, or cancer associated with hamartomatous polyposis syndrome in a first- or second-degree relative [12]. Considering our patient did not meet any of these criteria, genetic testing was not indicated and therefore not performed. The American Gastrointestinal Association also recommends polypectomy in symptomatic polyps or those ≥10 mm to prevent intussusception or other complications, such as bleeding [12]. Considering the polyp found in our patient was 20 mm and symptomatic, a polypectomy was performed. Unlike for many of the hereditary polyposis syndromes, there is currently no clear literature stating screening guidelines for solitary hamartomas. In terms of outcomes, solitary hamartomatous polyps have a promising prognosis; one retrospective study found no increase in the relative risk of dying or developing colorectal cancer [13].

Conclusions

Solitary hamartomatous polyps are typically benign and are most commonly discovered in the pediatric population [1,2]. Symptoms are relatively nonspecific and variable in severity, ranging from abdominal discomfort to overt gastrointestinal bleeding [2]. Polyp resection for histologic evaluation is crucial in differentiating these growths from one another. Solitary hamartomatous polyps appear similar to those of Peutz-Jeghers syndrome, presenting with smooth muscle, lamina propria, and inflammatory infiltrates coated by an epithelium that is hypertrophic [8]. In general, solitary hamartomas have been found to have a good prognosis; however, there is limited evidence toward screening guidelines for such polyps [13]. Additional research is warranted regarding this rare manifestation to develop standardized screening and treatment for these patients.

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