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Case Report

Gastric Heterotopia in the Rectum? A Rare Finding with Unclear Malignant risk

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Abstract

Introduction: Gastric heterotopia (GHT) is the presence of normal gastric mucosa in extra-gastric regions in the gastrointestinal tract (GIT). The most common locations are the esophagus, duodenum, and distal ileum (Meckel's diverticulum), but rarely it can also occur in the rectum producing symptoms and long-term risk.

Case Description: We describe a case of a 27 year old, healthy woman, with a long-standing history of loose stools, up to 12 bowel movements per day, and cramping in the abdomen. After diagnostic workup was negative, the presumptive diagnosis was irritable bowel syndrome (IBS) - diarrhea subtype. Her colonoscopy had normal biopsies of the cecum and transverse colon; however, the rectum contained a well demarcated erythematous region of approximately 1 inch diameter, adjacent to a prominent fold and resembling a flat polyp. Histopathology of the folds identified benign gastric mucosa without *H. pylori* and benign large intestinal mucosa. In addition to IBS, the diagnosis of rectal GHT was also made. The patient's IBS was treated with diet and lifestyle. Repeat sigmoidoscopy 2 years later confirmed stable GHT in rectum. She remains stable 8 years after diagnosis with no IBS symptoms, and no clinical concerns about her GHT.

Discussion: GHT is believed to be caused by pluripotent stem cells abnormally differentiating into gastric mucosa distal to the foregut. Common symptoms of GHT include abdominal pain and gastrointestinal bleeding but many cases are asymptomatic. A literature review demonstrates higher prevalence in males with a younger median age at diagnosis. Complications such a ulceration, fistula or perforation are more common in the pediatric population, while risk of malignant transformation is higher in adults. Treatment is with H2-receptor antagonists or proton-pump inhibitors for bleeding, or complete excision for refractory symptoms. There are a few cases of neoplastic changes reported in the literature but further observation is needed to appreciate the risk and benefit of preemptive excision by surgery or endoscopic submucosal dissection.

Keywords: Gastric Heterotopia; Rectum; Heterotopic Gastric Mucosa; Case Report

Abbreviations

GIT: Gastrointestinal Tract; GHT: Gastric Heterotopia; IBS: Irritable Bowel Syndrome

Introduction

Heterotopic mucosa is the presence of normal tissue displaced in foreign regions with demarcation from surrounding mucosa. A common form in the gastrointestinal tract (GIT) is gastric heterotopia (GHT) [1]. GHT can occur in all areas of the GIT and has even been found in the scrotum and spinal cord [1]. GHT is most commonly found in the esophagus, duodenum, and terminal ileum (Meckel's diverticulum), with prevalence in the foregut and midgut ranging from 0.1% to 10% [2]. In the rectum and anus, GHT is rare and since the first reported case in 1939 by Ewell and Jackson [3], 83 cases have been reported (including this one) with an age range of 0 to 70 years (median age 23 years) [1,4-9]. GHT can be congenital or acquired during epithelial repair and has potential for malignant transformation [9]. Herein we will review a recent case and summarize the literature on GHT in the rectum and colon.

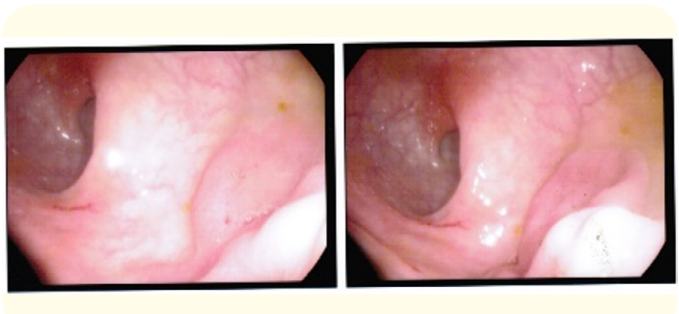
Case Report

A 27-year-old, healthy woman, with a history of cholecystectomy 1.5 years prior, and a 0.5 pack/day year history of smoking, presented with a chief complaint of loose stools and bloating. Her diarrhea started approximately 1 year prior, ranging from 4 to 12 loose stools per day, typically minutes after eating. The loose watery stools were not bloody, oily or mucousy, with no nocturnal stooling. Her symptoms worsened with meals, and improved with fasting, with dairy as a potential food trigger.

Her review of systems was benign, with no fever, night sweats, or weight loss. No extraintestinal manifestations or symptoms of Inflammatory Bowel Disease such as inflamed joints, oral ulcers, skin manifestations or eye inflammation. Her dietary history includes 2 to 3 caffeinated beverages per day, and no artificial sweeteners. Her examination was unremarkable, abdomen was soft, non-tender, and normal rectal examination.

Her initial diagnostic workup was negative; including lactose-breath test, celiac screen, microbiology cultures, and endocrinology work up. Subsequently, endoscopic examination was undertaken with colonoscopy showing no microscopic colitis; leading to a presumptive diagnosis of irritable bowel syndrome (IBS) - diarrhea subtype. However, the rectum contained a well demarcated, erythematous region of approximately 1-inch diameter, adjacent to a prominent fold resembling a flat polyp. Pathologic biopsy of the folds identified benign gastric mucosa and benign large intestinal mucosa. The diagnosis of rectal GHT was made based on the presence of gastric oxyntic mucosa with chief, parietal, and foveolar cells. *H. pylori* was not seen on histology.

The patient's IBS was treated with diet and lifestyle intervention. Loperamide was used to reduce stool frequency. The patient also responded to a lactose free diet with resolution of her long-standing diarrhea. Repeat sigmoidoscopy 2 years later confirmed stable rectal GHT. She remains stable 8 years after diagnosis with no IBS symptoms, and no clinical concerns related to her GHT.



Discussion

A literature review demonstrates higher prevalence of rectal specific GHT in males, approximately 64% of cases, with a median age of 12 years among males [1,4-6]. Pediatric populations have significantly more anorectal symptoms and complications than adult populations; including ulcers, fistula, and bowel perforation [4].

Working theories of the mechanism of GHT distal to the foregut include pluripotent stem cells abnormally differentiating into gastric mucosa either randomly or in response to damaged epithelium [9].

Common symptoms of anal and rectal GHT in order of descending frequency include painless rectal bleeding, perianal ulcer, anal pain, abdominal pain, and melena [1]; other symptoms include change in bowel habits and bloating but many cases are asymptomatic [4]. Life threatening bleeding was seen in 2 cases [4] and complications occurred in 23 cases (29%) [1,4-9]. GHT is diagnosed on histology of tissue and can be missed when abnormal cells are thought to be due to biopsy contamination (floaters) during endoscopic procedures [9]. A retrospective study found 2.9% of slides contained extraneous tissue [10].

Data on treatment are available for 65 cases with 45 cases consisting of excision (69%), 14 (22%) cases treated with H2 receptor antagonists, proton pump inhibitor, bismuth subsalicylate, and antibiotics for *H. pylori*, and 6 (9%) cases reported observational follow up [1,4-8]. H2-receptor antagonists or proton-pump inhibitors were primarily used to treat bleeding, whereas complete excision was used to treat refractory symptoms [9].

Only one case to date of anorectal GHT was found to be malignant [11]. However, several cases of GHT in other regions of the GIT, including the esophagus, pancreas, and small bowel, have been reported as malignant [11-14]. Moreover, it is possible that the number of cases of malignant GHT is underestimated due to poor differentiation of tumor in later stages overshadowing the initial GHT [1]. Thus, further observation is needed to appreciate the risk and benefit of pre-emptive excision by surgery or endoscopic submucosal dissection.

Conclusion

GHT is rarely diagnosed in the anorectal region and can mimic and be misdiagnosed as IBS or dyspepsia [2]. Also, histologic examination of tissue can be misdiagnosed as contamination [9]. Thus,

it is important to consider GHT as a potential diagnosis when er countering abnormal folds, polyps or irregular mucosa in the GI' Medical treatment may be considered, but the preferred and mos effective treatment is excision [4]. The rate of malignancy is lo for anorectal GHT, but follow up colonoscopy should be considere every 2 - 3 years, as GHT has been found to develop to malignanc in other regions of the GIT and may be underdiagnosed in the anc rectal region [1,4].

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