case report

Gastric heterotopia of rectum in a child: a mimicker of solitary rectal ulcer syndrome

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Bleeding per rectum is an uncommon presentation in pediatric patients. Heterotopic gastric mucosa in the rectum is a rare cause of rectal bleeding. Here, we report a 3-year-old child with a bleeding rectal ulcer that was initially diagnosed and managed as a solitary rectal ulcer syndrome. After 1 month, the patient persisted to have intermittent rectal bleed and severe anal pain. Repeat colonoscopy showed the worsening of the rectal ulcer in size. Pediatric surgeon excised the ulcer, and histopathological examination revealed a gastric fundic-type mucosa consistent with the diagnosis of gastric heterotopia of the rectum. Over the following 18 months, our patient had experienced no rectal bleeding and remained entirely asymptomatic. In conclusion, heterotopic gastric mucosa of the rectum should be considered in the differential diagnosis of a bleeding rectal ulcer.

Bleeding per rectum is an uncommon symptom in pediatric patients. Heterotopic gastric mucosa in the rectum is a rare cause of rectal bleeding. Heterotopic gastric mucosa has been identified throughout the gastrointestinal tract including the nasopharynx, tongue, esophagus, small intestine, gallbladder, biliary tract, colon, and rectum.[1] Typically, ectopic gastric mucosa is asymptomatic. The rarity of symptomatic rectal gastric mucosa, decreased awareness of pediatricians and gastroenterologists about the possibility of gastric heterotopia of rectum in the differential diagnosis of rectal bleeding, and non-specificity of clinical presentation lead to significant delay in diagnosis and late initiation of appropriate management. Here, we report a 3-year-old child with bleeding rectal ulcer due to rectal gastric heterotopia that was misdiagnosed as solitary rectal ulcer syndrome, with extensive review of pediatric medical published reports. We aim to increase the awareness of pediatricians and gastroenterologists about the possibility of heterotopic gastric mucosa in the rectum as a cause of rectal bleeding to make early diagnosis and initiate early management.

CASE

A 3-year-old male presented to the emergency department at King Fahad Medical City with a history of episodic rectal bleeding and anal pain for 8 months. The bleeding was described as bright red blood streaked on the stool and blood in the toilet bowl. He had occasional abdominal pain not associated with defecation. The anal pain was not related to episodes of hematochezia. There was no family history of any bleeding disorders, recurrent epistaxis, gastrointestinal polyps, or cancer. He had been investigated in 2 other local hospitals due to same complaint, and 2 colonoscopies had been done during the illness period but were reported to be normal.

On physical examination, the height was 93 cm (75th centile) and the weight was 13 kg (75th centile); the abdomen was soft and lax without organomegaly. On anal examination, he had no anal fissures or skin tags. Other systemic examination was unremarkable. Laboratory investigations showed normal complete blood count and coagulation profile. Stool examination for ova and parasites and culture were negative.

On colonoscopy, an ulcerative lesion of 2×2 cm was observed, which was 0.5 centimeter proximal to the dentate line of anal canal (**Figure 1**). Biopsies obtained from mucosa around the ulcer showed no specific histopathological changes. The child was managed as solitary rectal ulcer with rectal sucralfate and cortifoam enema. After 1 month, the patient persisted to have intermittent rectal bleed and severe anal pain. Repeated sigmoidoscopy showed worsening of the ulcer in size

and depth (**Figure 2a**). Worsening of the ulcer despite traditional therapy of solitary rectal ulcer raised suspicion of different pathology and led to further diagnostic workup to exclude infectious and malignant etiologies. Biopsies from the edge of the ulcer and the rectal mucosa around the ulcer were subjected to Zeel-Nelson stain, mycobacterium tuberculosis DNA polymerase chain reaction test, lymphoma stain, and fungal stain, but all turned out to be negative. Magnetic resonance imaging of pelvis and abdomen was normal. Pediatric surgeon was consulted who performed cauterization of the ulcer. On follow up 1 month later, the child was asymptomatic and sigmoidoscopy revealed a healed ulcer (**Figure 2b**).

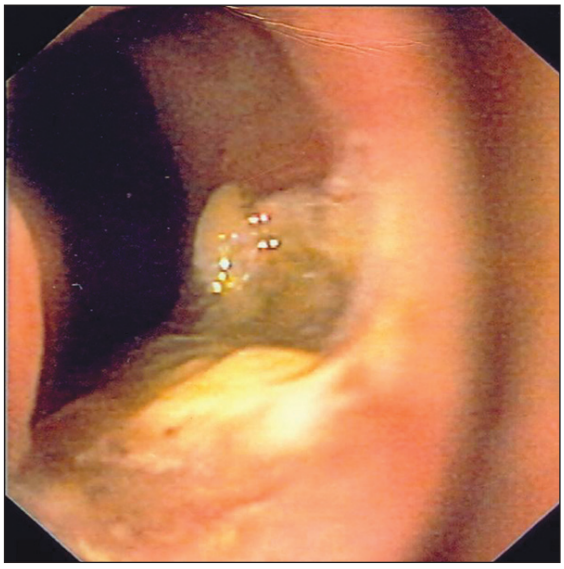

Figure 1. An ulcerative lesion of 2×2 cm observed on colonoscopy.

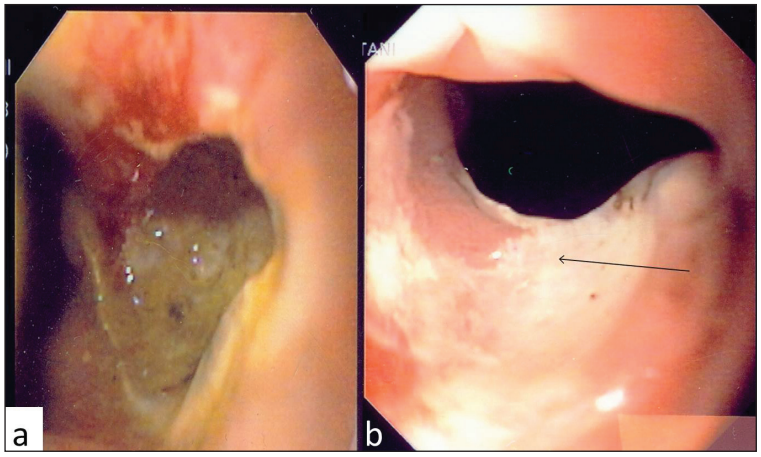

Figure 2. Sigmoidoscopy (a) showing worsening of the ulcer in size and (b) revealing a healed ulcer.

One year later, the patient presented with the same complaint of rectal bleeding and anal pain. Sigmoidoscopy showed recurrence of a small ulcer 1 X 1 cm at a different site than the first ulcer, located just above the dentate line of the anal canal (**Figure 3**). Pediatric surgeon excised the ulcer, and histopathological examination revealed a gastric fundic-type mucosa (**Figure 4**) consistent with the diagnosis of gastric heterotopia of the rectum. The staining of the excised specimen with Geimsa stain was negative for Helicobacter pylori organism. Meckel's scan did not show any residual gastric mucosa in the rectum or in other parts of gastrointestinal tract. Over the following 18 months, our patient did not experience any recurrence of rectal bleeding and he remained entirely asymptomatic.

DISCUSSION

The most accepted hypothesis for heterotopia of gastric mucosa is an error of differentiation.1 Pluripotent endoderm stem cells have the capability of differentiating into all types of gastrointestinal epithelium. This theory is supported by immunocytochemical studies demonstrating that the metabolic and functional activity of gastric heterotopic mucosa, regarding the production of acid and mucin, is identical to that of the normal stomach.[2]

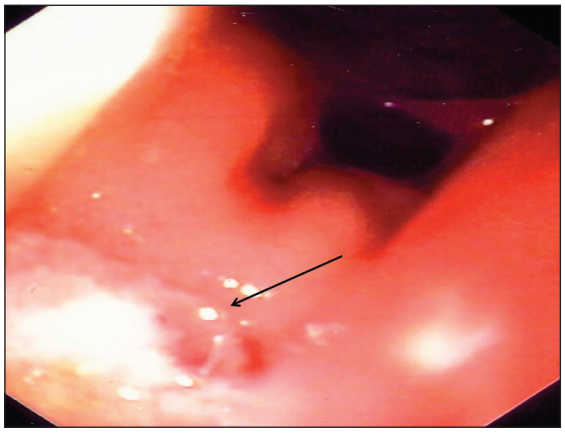

Figure 3. Sigmoidoscopy showing recurrence of a small ulcer 1×1 cm at a different site than the first ulcer.

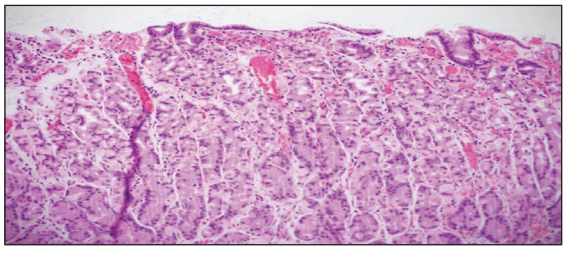

Figure 4. Histopathological examination revealing a gastric fundic-type mucosa.

Table 1. Summary of reported pediatric cases of gastric heterotopia in rectum.

Reference	Age/Sex	Presenting symptoms	Duration	Site of lesion	Lesion
Shawartzenberg and Whitington[15]	6 mo/M	Intermittent rectal bleeding and colic	4 mo	2 cm from anus	Diverticulum
Sugarman et al[16]	13 mo/F	Rectal bleeding and ulcers	1 d	6 cm from anus	Polyp
Parkash et al[17]	18 mo/F	Perianal ulceration, pruritus ani, and anocutaneous fistula	15 mo	Left posterior	Diverticulum
Murray et al[18]	2 y/F	Rectal bleeding	16 mo	Throughout colon	Polypoid, ulcer
Wiersma et al[19]	2 y /M	Rectal bleeding	NA	1 cm from the dentate line	Ulcer
Cheli et al[20]	2 y/M	Rectal bleeding	1 y	4 cm from anal verge	Mucosal lesion
Marines et al[21]	3 y/M	Rectal bleeding and abdominal pain	5 mo	5 cm from anus	Polyp
Garmendia et al[22]	4 y/M	Rectal bleeding , weight loss, and loose stools	NA	5 cm from anorectal junction	Mass
Kalani et al[5]	4 y/M	Bloody diarrhea, ulcers, and rectovesical fistula	1 yr	2 cm above pectinate line	Mucosal fold
Stockman et al[23]	4 y/F	Rectal bleeding	2 y	At 5 cm from anorectal junction	Diverticulum
Nigro et al[24]	5 y/M	Rectal bleeding	1 mo	6 cm from anal verge	Polyp
Sauer C et al[13]	5 y/F	Painless rectal bleeding	2.5 y	5 cm from anus	Polypoid lesion
Ewell and Jackson[25]	6 y/M	Rectal bleeding and ulcer	1 wk	5 cm above anus	Polyp
Thompson et al[26]	6 y/M	Rectal bleeding	NA	Anus	Diverticulum
Wolff M[27]	7 y/M	Rectal bleeding	NA	8 cm from pectinate line	Polyp
Kestemberg et al[7]	9 y/ M	Rectal bleeding	NA	Rectum	Polyp
Kumar et al[28]	10 y/M	Rectal bleeding and proctalgia	2 yr	9 cm from anal verge	Diverticulum
Lord and Tribe[29]	11 y/M	Rectal bleeding	6 mo	1 cm above anus	Mucosal flap
Carlei et al[2]	13 y/ M	Rectal bleeding and tenesmus		2-3 cm above dentate line	Ulcer
Antonietta et al[30]	13 y/M	Rectal bleeding and tenesmus	1 d	2-3 cm above pectinate line	Ulcer
Picard et al[31]	14 y/M	Rectal bleeding and ulcers	12 y	3 cm above dentate line	Ulcer
Jordan et al[32]	16 y/M	Rectal bleeding, pain, and ulcer	4 y	4 cm above dentate line	Polyp
Edouard et al[33]	17 y/M	Rectal bleeding, rectal syndrome, and ulcer	1 d	3 cm from anus	Polyp
Kokil et al[34]	12 y/M	Rectal bleeding	10 y	3 cm from anus	Polyp
Present case	3 y/M	Rectal bleeding anal pain	8 mo	0.5 cm proximal to dentate line	Rectal ulcer

Cm: Centimeter, F: female, M: male, mo: month, NA: not available, Wk: week, y: year.

We searched the English-language published reports in Pubmed, Embase, and Medline (1966-2012) for pediatric cases of ectopic gastric mucosa using the following search words: gastric heterotopia, rectal gastric mucosa, rectal bleeding, and child. To the best of our knowledge, 24 pediatric cases of gastric heterotopia in the rectum have been reported so far. The review of published reports (**Table 1**) revealed that males were more commonly affected (M:F, 19:5), with an average age at presentation of about 7 years (range, 6 months to 17 years). The most common presentation of rectal gastric heterotopia was painless rectal bleeding (95.6%); other less common presentations included perianal ulceration (4.3%), anal pain (17%), abdominal pain (8.6%), and diarrhea (8%). Symptoms were present from 1 day to 12 years prior to diagnosis. Heterotopic gastric tissue has most commonly been identified in association with a polyp (n=11), followed by diverticula (n=5), ulcer (n=5), and in reddish-appearing mucosal plaque, folds, or flaps (n=4). The majority of the lesions were located more than 5 cm above from the anal verge. However, lesions less than 2 cm above the anal verge and more than 9 cm do occur.

The natural history of gastric heterotopia is unknown. Serious complications because of heterotopic gastric mucosa included major gastrointestinal bleeding, bowel perforation, intussusception, and rectovesical fistula.[3-6] *H pylori* organisms have been noted in rectal gastric heterotopic mucosa; the eradication of the organism resulted in the resolution of chronic active gastritis in the heterotopic mucosa.[7] This finding supports the possibility that *H pylori* organisms might pass along the gastrointestinal tract in a viable form to colonize ectopic gastric tissue in the rectum and contribute to the ulceration and bleeding seen in these cases. It is unclear whether and how often heterotopic gastric mucosa progresses to malignancy. Although there have been no case reports that specifically describe malignant transformation of heterotopic gastric mucosa of the rectum, there have been 6 cases of gastric heterotopia of the esophagus that presented as adenocarcinoma[8,9] and 1 report described a relationship between heterotopic gastric mucosa in the colon and a premalignant tubule-villous adenoma.[10] All of these cases were in older adults, and the duration of gastric heterotopia in the esophagus was unknown.

The definitive diagnosis of gastric heterotopia requires histopathological demonstration of a gastric mucosa outside the stomach. Technetium scanning can be used as adjunctive aid in the localization of gastric heterotopias,[11,12] but direct visualization and biopsy are needed to confirm the diagnosis. In many of the cases cited in **Table 1**, the symptoms were present for many years, and patients underwent extensive work-up prior to diagnosis including laparotomy in 1 case. Endoscopist may miss the lesion if it is too close to the anal verge, which emphasizes on the importance to carefully inspect the rectal segment just above the anus during colonoscopy for rectal bleeding. Sampling error, when biopsies are obtained from an inappropriate site, is another reason for delay in the diagnosis of gastric heterotopia of the rectum.

Gastric heterotopia of the rectum, presenting with a bleeding ulcer, can be mistaken with solitary rectal ulcer. These two different rectal pathologies should be differentiated because the therapy is different, and indeed the use of steroid enema to treat solitary rectal ulcer can worsen the outcome of ulcer secondary to gastric heterotopia, as in our case. Constipation and rectal prolapse usually accompany solitary rectal ulcer, while anal pain occurs with rectal gastric heterotopia. The definitive diagnosis of both entities mandates histopathological confirmation. The characteristic histopathological findings of solitary rectal ulcer constitute elongation with fibrosis and extension of fibers from the muscularis mucosa into the lamina propria, while the histopathological diagnosis of gastric heterotopia necessitates the demonstration of gastric mucosa.

In a number of case reports, patients with heterotopic gastric mucosa were treated with H^2 receptor blockers or proton pump inhibitors.[11,12] These therapies may ameliorate or eliminate symptoms, but they do not cause involution of the mucosal abnormalities; when the medication is discontinued, bleeding quickly recurs.[13] Given concerns for possible malignant transformation over the long term, the resection of the lesion should be performed surgically or endoscopically.[14] No recurrences have been reported.

In conclusion, heterotopic gastric mucosa of rectum should be considered in the differential diagnosis of a bleeding rectal ulcer to prompt early diagnosis and surgical resection of the ectopic gastric mucosa.

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