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Case Report

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A rare case of heterotopic gastric mucosa of the rectum with complete intestinal metaplasia highlighting the utility of molecular microsatellite testing in diagnostic corroboration

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Abstract

Heterotopic gastric mucosa (HGM) is common in the esophagus but exceedingly rare in other sites. Only 73 cases of HGM of the rectum have been described in the literature. Here, we present a 32-year-old patient who underwent colonoscopy for persistent abdominal pain, wherein a lesion ultimately described as rectal HGM was discovered. Biopsy tissue was exclusively composed of gastric mucosa, raising concern for preanalytical error by sample mix-up. Microsatellite testing against a colonic polyp excised during the colonoscopy was employed to confirm the diagnosis and exclude sample mix-up. Subsequent resection of the HGM corroborated the diagnosis, and focal complete intestinal metaplasia was found at its epicenter. This report highlights the utility of molecular microsatellite testing in confirming the diagnosis of ectopic tissues.

Introduction

Heterotopic gastric mucosa (HGM) is the presence of gastric mucosa outside of the stomach, most commonly found in the esophagus [1]. Only 73 cases of HGM of the anus or rectum have been reported in the literature [2,3]. Resection of the lesion is generally recommended as treatment for rectal HGM [2,4]. While malignant transformation has not been reported in rectal HGM, it has been reported in other sites of HGM [5,6]. The absence of reported malignant transformation in rectal HGM may be due to its rare nature and not necessarily due to pathophysiological differences in transformation between HGM sites. Resection of rectal HGM is recommended due to the potential for malignant transformation via the intestinal metaplasia pathway, similar to that seen in gastric *Helicobacter pylori* infection. Microsatellite testing is a molecular technique which "fingerprints" samples by identifying alleles of short tandem repeat sequences, allowing identification of the patient of origin when compared to distinct tissue samples [7]. Here we employ microsatellite testing to corroborate a diagnosis of HGM by excluding the remote possibility of tissue contamination in surgical pathology.

Case summary

A 32-year-old female with past medical history of smoking, obesity, gallstone pancreatitis, and polycystic ovarian syndrome presented to clinic with right upper quadrant abdominal pain associated with hunger and fatty food consumption. Ultimately, a colonoscopy was performed, wherein a 12 mm sessile polyploid lesion with focal villous-appearing mucosa was discovered in the distal rectum (Figure 1). The distal border of the lesion was adjacent to the dentate line. The lesion was biopsied in addition to resection of a 2 mm polyp that was discovered in the sigmoid colon.

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Oxyntic gastric mucosa lining the lamina propria was identified throughout the entirety of the rectal biopsy specimen, with notable absence of any colonic mucosa. Due to the lack of colonic mucosa seen in the biopsy specimen, preanalytical error by sample mix-up was suspected. Molecular microsatellite testing between the biopsy specimen and the concomitantly resected 2 mm polyp was performed. Testing for 16 short tandem repeats (STR) was performed in both the resected polyp and biopsy specimen (Table 1). Alleles for each STR were found to be identical between the two specimens, supporting that the two specimens originated from the same patient and confirming the diagnosis of HGM.

The rectal HGM lesion was resected five months later. Histopathologic examination of the resection revealed gastric mucosa with an abrupt transition to colonic mucosa, as well as a focal point of complete intestinal metaplasia within the central HGM (Figure 2). This finding correlates with the focal villous changes observed grossly at colonoscopy (Figure 1C, circle). After thorough examination, no dysplasia was identified.

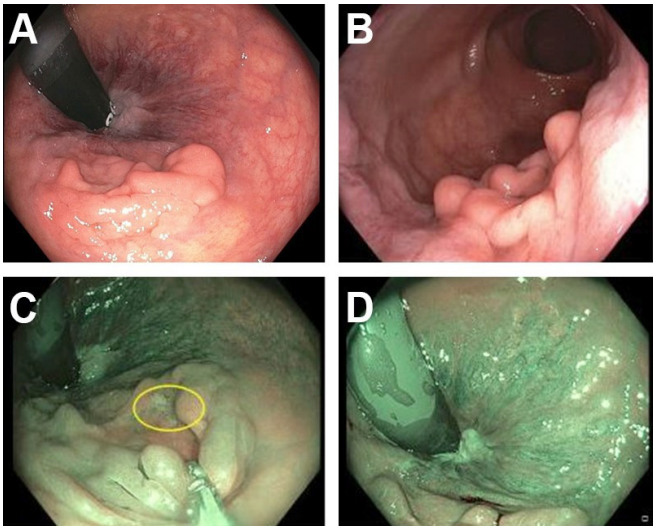

Figure 1: Colonoscopy findings, rectum. **(A)** Sessile polyploid lesion, seen in retroflexion. **(B)** Lesion, seen in forward view. **(C)** Lesion, seen in retroflexion. Villous mucosa circled. **(D)** Relation of dentate line to lesion.

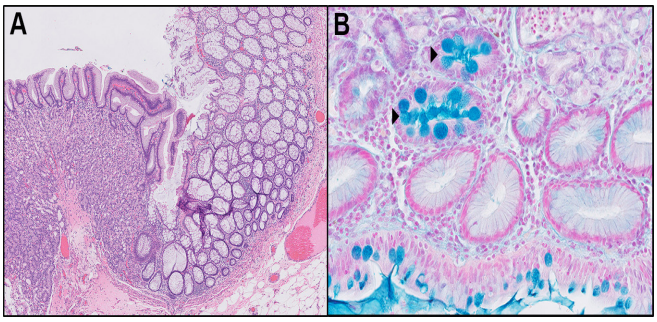

Figure 2: Resection histopathology. **(A)** Rectal HGM resection specimen with benign oxyntic mucosa abutting colonic crypts (H&E; 20X magnification). **(B)** Rectal resection specimen with focal complete intestinal metaplasia with goblet cells (arrowhead) arising within a region of heterotopic gastric mucosa (Alcian blue, 200X magnification). No dysplasia was identified.

Table 1: Microsatellite testing of resected lesions. Microsatellite testing of 16 short tandem repeats between the rectal HGM lesion and the concomitantly resected sigmoid polyp. Test reveals identical alleles between the two samples, ruling out preanalytical error (specimen mix up) and confirming the diagnosis of HGM.

STR Marker	HGM Lesion alleles	Sigmoid Polyp alleles
vWA	16	16
TPOX	8	8
TH01	9.3	9.3
FGA	20, 21	20, 21
D8S1179	11, 15	11, 15
D7S820	7, 12	7, 12
D5S818	11, 12	11, 12
D3S1358	16, 17	16, 17
D2S1338	18, 21	18, 21
D21S11	28, 29	28, 29
D19S433	12, 15	12, 15
D18S51	12, 15	12, 15
D16S539	12, 13	12, 13
D13S317	10, 12	10, 12
CSF1PO	10, 12	10, 12
AMEL	X	X

STR: short tandem repeat; HGM: heterotopic gastric mucosa.

Discussion

HGM is an exceedingly rare finding outside of the esophagus, with only 73 cases of rectal HGM reported in the literature. While HGM is discovered incidentally in 19% of patients, it most commonly presents with hematochezia, but is also reported to present with upper abdominal pain as a non-specific symptom [2]. The patient presented here is another case of incidentally discovered HGM of the rectum.

Histopathological examination revealed the gastric mucosa was oxyntic in nature, which is seen in 83% of reported cases of HGM of the rectum [2,8]. Additionally, we report the discovery of focal complete intestinal metaplasia within the rectal HGM. Complete intestinal metaplasia (CIM) within rectal HGM is a rare finding, with only two other cases reported in the literature [2,9,10]. CIM in rectal HGM is reported to be concerning for development of dysplasia within the HGM, as seen in the metaplasia-adenoma-carcinoma sequence of antral gastric *Helicobacter pylori* infection [11,12]. In fact, four cases of infection of HGM by *Helicobacter pylori* have been previously reported [2]. Acid production by HGM has been confirmed by pH probe, and similar to Meckel's diverticulum the lesion can cause irritation and ulceration of the affected colon. Treatment with proton pump inhibitor (PPI) or H2-receptor antagonist medications have been shown to lead to healing and resolution of ulcers associated with HGM [2].

Short tandem repeats (STRs), or microsatellites, are regions of the genome consisting of short (<10 bp) repeating DNA se-

quences [13]. Mutations in these microsatellites are more common than in other areas of the genome, particularly in the number of repeats [14]. As a result, sequencing of STRs can be used as a molecular "fingerprint" of patient identity in tissue specimens. This is commonly employed in forensic pathology and paternity testing [15,16]. STR testing is a powerful molecular technique capable of confirming or excluding relationships between two specimens or individuals. Cases such as this rectal HGM are highly suspicious for preanalytical error (sample mix-up) due to their rare nature and unexpected histological features.

Conclusion

Preanalytical errors in tissue diagnosis portend devastating clinical and medicolegal consequences and warrant rigorous investigation. We establish here that when multiple specimens are available, molecular microsatellite testing is a reliable method that can be used to confirm rare ectopic diagnoses (such as rectal HGM) which may otherwise dismissed.

References

1. Chong VH. Clinical significance of heterotopic gastric mucosal patch of the proximal 106 esophagus. World J Gastroenterol. 2013; 19: 331-338.
2. Giacomini F, Gotoda T, Elisei W, Rigato P, Montagnese F, Saito Y, et al. Heterotopic gastric mucosa in the anus and rectum: First case report of endoscopic submucosal dissection and systematic review. Gastroenterol Rep (Oxf). 2016; 4: 196-205.
3. Cheung M, Elfanagely Y, Skole K. 1761 Heterotopic Pancreatic, Sebaceous, and Gastric Tissue: Report of Case and Review of the Literature. American Journal of Gastroenterology. 2019; 114: S988.
4. Chen WG, Zhu HT, Yang M, Xu GQ, Chen LH, Chen HT. Large heterotopic gastric mucosa and a concomitant diverticulum in the rectum: Clinical experience and endoscopic management. World J Gastroenterol. 2018; 24: 3462-3468.
5. Srinivasan R, Loewenstine H, Mayle JE. Sessile polypoid gastric heterotopia of rectum: a report of 2 cases and review of the literature. Arch Pathol Lab Med. 1999; 123: 222-224.
6. Xeropotamos N, Skopelitou AS, Batsis C, Kappas AM. Heterotopic gastric mucosa together with intestinal metaplasia and moderate dysplasia in the gall bladder: report of two clinically unusual cases with literature review. Gut. 2001; 48: 719-723.
7. Schlotterer C. Evolutionary dynamics of microsatellite DNA. Chromosoma. 2000; 109: 365-371.
8. Mannan AASR, Vieth M, Khararjian A, Khandakar B, Lam-Himlin D, Heydt D, et al. The outlet patch: gastric heterotopia of the colorectum and anus. Histopathology. 2018; 73: 220-229.
9. Seitz G, Schuder G. Gastric mucosal heterotopia of the rectum. Pathologist. 1987; 8: 52-55.
10. Kim HS, Kim JH, Yoon SO. Heterotopic gastric mucosa with focal intestinal metaplasia and squamous epithelium in the rectum. Digestive Endoscopy. 2012; 24: 46-48.
11. Shao L, Li P, Ye J, Chen J, Han Y, Cai J, et al. Risk of gastric cancer among patients with gastric intestinal metaplasia. Int J Cancer. 2018; 143: 1671-1677.
12. Correa P, Piazuelo MB, Camargo MC. The future of gastric cancer prevention. Gastric Cancer. 2004; 7: 9-16.
13. Gymrek M. A genomic view of short tandem repeats. Curr Opin Genet Dev. 2017; 44: 9-16.
14. Sun JX, Helgason A, Masson G, Ebenesersdóttir S, Li H, Mallick S, et al. A direct characterization of human mutation based on microsatellites. Nature Genetics. 2012; 44: 1161-1165.
15. S. B. S. Silva D, Sawitzki FR, Scheible MKR, Bailey SF, S. Alho C, et al. Paternity testing using massively parallel sequencing and the PowerSeqTM AUTO/Y system for short tandem repeat sequencing. Electrophoresis. 2018; 39: 2669-2673.
16. Kayser M. Forensic use of Y-chromosome DNA: a general overview. Hum Genet. 2017; 136: 621-635.