Philippine Journal of Gastroenterology

Familial adenomatous polyposis with rectal adenocarcinoma presenting as hematochezia: A case report

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Abstract

Familial adenomatous polyposis (FAP) is a hereditary syndrome characterized by the development of hundreds to thousands of adenomatous polyps in the gastrointestinal tract. If untreated, it carries a nearly 100% lifetime risk of colorectal cancer. This is a case report of a 43-year-old Filipino woman with a one-year history of hematochezia initially misdiagnosed as Grade IV internal hemorrhoids. Computed tomography revealed a rectal mass, and colonoscopy showed an ulceroproliferative rectal mass with hundreds of polyps carpeted throughout the colon. Genetic testing identified a pathogenic *APC* mutation (c.3927_3931del), confirming *de novo* FAP despite the absence of family history. The patient underwent exploratory laparotomy, total proctocolectomy, total mesorectal excision, and end ileostomy. This case illustrates how FAP may masquerade as common benign anorectal disease, leading to significant diagnostic delay. It further highlights the central role of genetic testing in confirming *de novo* cases and the impact of healthcare access barriers in resource-limited settings. Early recognition and timely surgical management are crucial to reducing cancer mortality in FAP.

Keywords: *Familial adenomatous polyposis, Adenomatous polyposis coli, Rectal adenocarcinoma, Hematochezia, Case report*

Introduction

Familial adenomatous polyposis (FAP) is an inherited syndrome associated with colorectal cancer, marked by the presence of numerous adenomatous polyps in the gastrointestinal mucosa. This condition carries an almost 100% lifetime risk of developing colorectal cancer and occurs with an incidence of approximately 1 in 7,000 to 30,000 births.[1] If left untreated, nearly all individuals with FAP will develop colorectal cancer, with the average age of diagnosis being 39 years. About 40% of those diagnosed with colorectal cancer have synchronous malignancies, and more than 80% of the tumors are located on the left side of the colon. Early identification of FAP, particularly through genetic testing, enables timely surveillance and preventive interventions, which can markedly reduce the risk of cancer.[2,3,4]

Case Presentation

The patient was a 43-year-old Filipino woman with a history of diabetes mellitus and hypertension who presented with a one-year history of intermittent hematochezia and weight loss. The unintentional weight loss was not quantified by the patient. At presentation, the patient was fully ambulatory and independent in activities of daily living, corresponding to an ECOG performance status of 0–1. At this time, the patient did not present with diarrhea, no vomiting, nor abdominal pain. The patient has no family history of colon, gastric, endometrial, or any cancer. Initially, she was diagnosed with grade 4 internal hemorrhoids and treated symptomatically.

The patient's financial constraints limited her access to appropriate diagnostic procedures. Digital rectal examination revealed a circumferential, friable mass in the distal rectum, with no active bleeding at the time of examination. Initial evaluation was performed by a primary care physician, who attributed symptoms to hemorrhoidal disease. Due to persistent hematochezia, the patient was subsequently referred to a gastroenterologist approximately one year after symptom onset, leading to definitive imaging, colonoscopy, and diagnosis. Prior to referral, the patient had no previous colonoscopy or cross-sectional abdominal imaging performed. She was initially managed symptomatically for presumed grade IV internal hemorrhoids with conservative medical therapy. A subsequent whole abdominal CT scan revealed a semi- circumferential mass in the mid to low rectum, measuring approximately 64 mm (Figure 1). Contrast-enhanced CT of the whole abdomen demonstrated a semi-circumferential mass in the mid to low rectum measuring approximately 6.4 cm in length, causing luminal narrowing with minimal mesorectal fat stranding. Several prominent mesorectal lymph nodes were identified, the largest measuring 9 mm in short-axis, as well as an enlarged left iliac chain lymph node measuring 20 × 26 mm, raising concern for nodal metastasis. No hepatic lesions, ascites, bowel obstruction, or compression or invasion of adjacent organs were identified. The pancreas, spleen, kidneys, adrenal glands, and urinary bladder were unremarkable. No peritoneal or retroperitoneal metastases were seen. The patient was then referred for a colonoscopy (Figure 2) which revealed hundreds to thousands of sessile and pedunculated polyps diffusely involving the colon, ranging from 2–10 mm in size, with surface patterns consistent with adenomatous morphology (NICE type 2; JNET type 2A). A circumferential, friable rectal mass measuring approximately 5–6 cm in length, located 2 cm from the dentate line, was noted, partially obstructing the lumen. The terminal ileum was intubated and appeared normal, with no polyps identified. Esophagogastroduodenoscopy was not performed at the time of diagnosis.

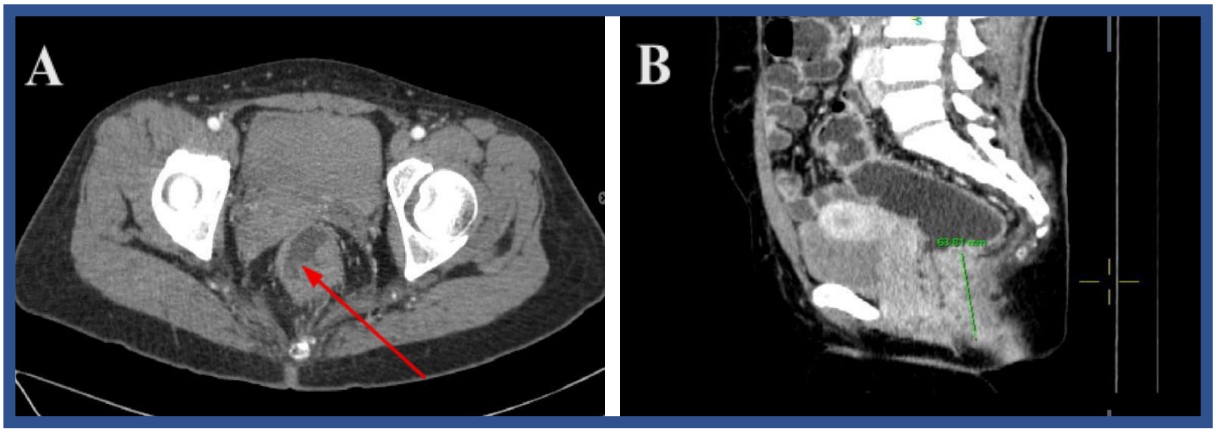

Figure 1. Contrast-enhanced computed tomography (CT) of the abdomen and pelvis in (A) axial and (B) sagittal views. The arrows indicate a heterogeneously enhancing mass in the rectum involving the posterior and lateral walls. (With axial however without any coronal views)

Colonoscopy and Histopathology Findings

Biopsies from colonoscopy confirmed tubular and tubulovillous adenoma in the colon where in Figure 3, microsections show a polypoid colonic mucosa covered with low-grade dysplastic epithelium comprised of hyperchromatic, elongated nuclei arranged in a pseudostratified manner. On the other hand, rectal mass was diagnosed as adenocarcinoma as shown on Figure 4 showing many well to moderately differentiated dysplastic colonic glands with cribriforming pattern and some fusing with each other, invading into the stroma.

The patient subsequently underwent exploratory laparotomy, total proctocolectomy, total mesorectal excision, with end ileostomy (Figure 5). An ileal pouch–anal anastomosis (IPAA) was not considered due to the presence of a low-lying rectal adenocarcinoma with sphincter involvement (T3b disease), threatened circumferential resection margin, and the need for oncologic clearance. Given these factors, total proctocolectomy with end ileostomy was deemed the most appropriate oncologic and functional option.

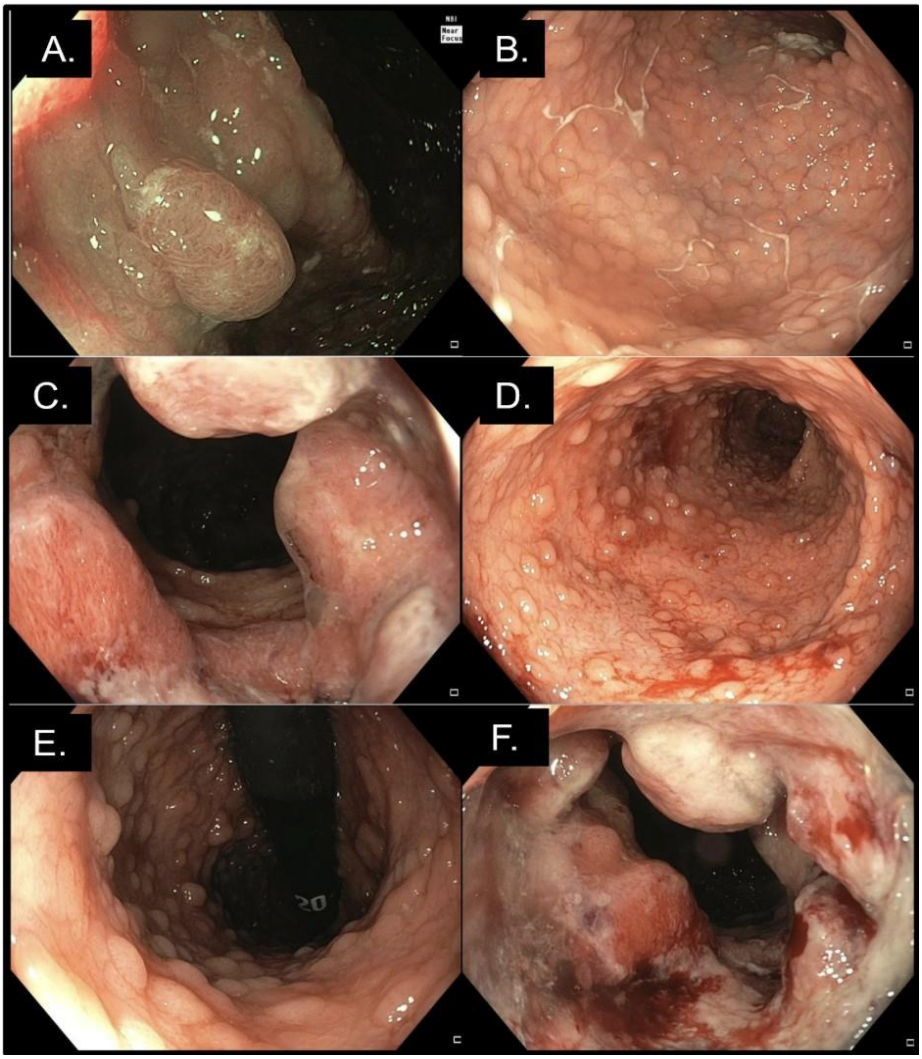

Figure 2. Colonoscopy showing multiple polyps on the colonic (A) Sigmoid (B) Descending (C) Rectum (D) Transverse colon (E) Retroflexed view of the rectal vault and rectal mucosa (F) Forward view of rectum

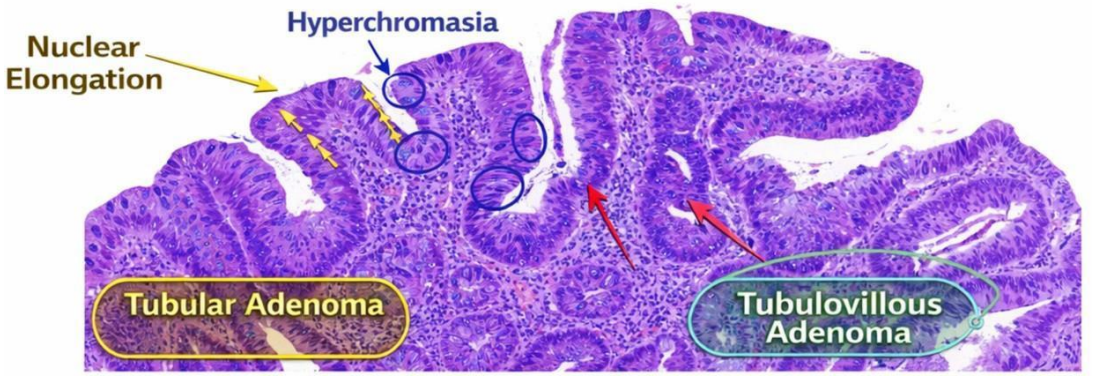

Figure 3. Low-grade dysplastic adenomatous epithelium (tubular and tubulovillous adenomas)

Figure 3 shows high-power H&E photomicrograph demonstrating low-grade dysplasia in adenomatous colonic mucosa. Yellow arrows highlight nuclear elongation with preserved basal polarity. Blue-circled nuclei emphasize hyperchromasia and pseudostratification confined to the lower half of the epithelium. The yellow-outlined region represents a tubular adenoma with predominantly tubular gland architecture, while the green-outlined region demonstrates a tubulovillous adenoma showing villiform projections with dysplastic lining epithelium. No stromal invasion is identified.

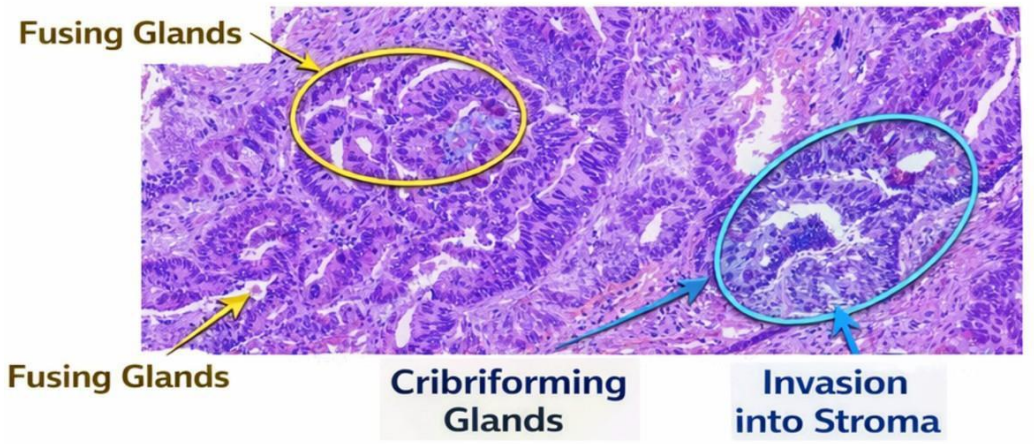

Figure 4. Rectal adenocarcinoma

Figure 4 shows a high-power H&E photomicrograph of rectal adenocarcinoma. Yellow-circled areas indicate gland fusion with loss of normal intervening lamina propria. The central labeled region demonstrates cribriform glandular architecture characterized by back-to-back malignant glands with punched-out lumina. The blue-circled area highlights invasion into desmoplastic stroma, confirming infiltrative growth. Architectural complexity and stromal infiltration are consistent with well to moderately differentiated invasive rectal adenocarcinoma.

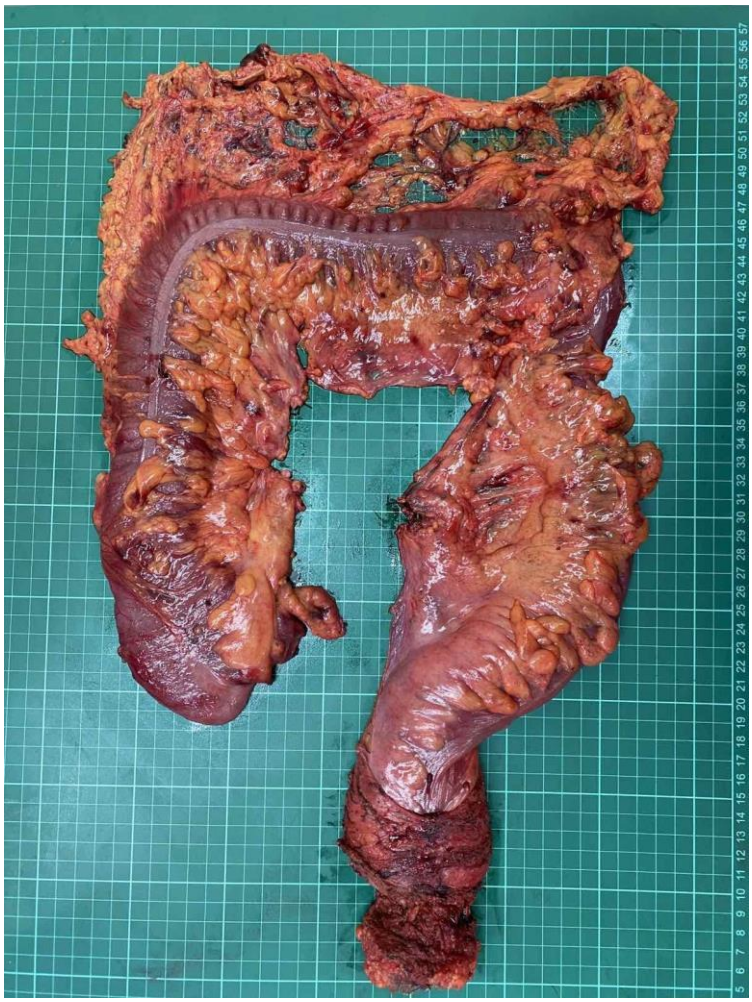

Figure 5. Gross specimen from exploratory laparotomy, total proctocolectomy, total mesorectal excision, with end ileostomy performed on the patient

Discussion

Familial adenomatous polyposis (FAP) is a well-established risk factor for colorectal cancer (CRC), with nearly all untreated patients progressing to malignancy.[1, 2] While the majority of individuals with FAP have an inherited family history, approximately 11–25% of cases arise from *de novo* germline mutations in the *APC* gene, with emerging Asian data suggesting under-recognition due to limited access to genetic testing.[6]

Patient diagnosis was particularly challenging due to the absence of a known family history. Notably, the specific variant identified in this case, c.3927_3931del (p.Glu1309fs), was recognized as a "hotspot" for *de novo* events, accounting for 30–45% of such cases in various cohorts.[7] Mutations at codon 1309 were clinically significant as they were frequently associated with a "profuse" polyposis phenotype (often >1,000 polyps) and an earlier onset of CRC compared to mutations located elsewhere on the *APC* gene.[7, 8]

Diagnostic delay remains a critical challenge in the management of CRC and hereditary polyposis syndromes in low- and middle-income countries (LMICs). Limited access to endoscopic services, high out-of-pocket healthcare costs, and delayed specialist referral frequently result in advanced-stage disease at presentation. In such settings, alarm symptoms, such as rectal bleeding, are often initially attributed to benign anorectal conditions, particularly hemorrhoids, leading to missed opportunities for early diagnostic evaluation. Misattribution of hematochezia to hemorrhoidal disease is especially problematic in middle-aged patients, who may fall outside routine CRC screening programs and are therefore perceived as lower risk. Several studies have demonstrated that rectal bleeding in patients over 40 years of age warrants prompt endoscopic evaluation, as reliance on presumptive hemorrhoidal diagnoses has been associated with delayed detection and poorer oncologic outcomes.[2, 9]

In the present case, the patient experienced a delay of nearly one year from symptom onset to definitive diagnosis, largely due to initial conservative management for presumed hemorrhoids and financial constraints limiting access to colonoscopy. This pattern mirrored findings from local and regional case reports from Southeast Asia, which described delayed diagnoses of FAP and early-onset CRC attributed to healthcare access barriers and underutilization of genetic testing.

Published Filipino case reports on polyposis syndromes remain limited. This case adds to the growing body of regional evidence emphasizing the need for earlier endoscopic evaluation and heightened suspicion for hereditary syndromes even in the absence of family history.

Study	Year	Age / Sex	Presentation	Key Work-up	Management
Current Case	2026	43 / F	Hematochezia (misdiagnosed as hemorrhoids)	Colonoscopy, CT, Genetic Testing (APC mutation)	Total proctocolectomy, end ileostomy
JRRE Case Series (rid=72)	2015	Siblings (2M, 1F)	2 siblings asymptomatic (screening); 1 with hematochezia	Family genogram, Colonoscopy, Genetic Testing	Total colectomy and proctocolectomy
EJMACES Case (2017)	2017	25 / F	Abdominal wall mass (Unusual/Attenuated FAP)	CT, Incision Biopsy, Colonoscopy	Restorative proctocolectomy (RPC) with IPAA
Reyes et. al (HERDIN)	1999	31/F	Rectal Bleeding	Colonoscopy	Family tracing

Table 1. Comparison of locally published cases of Familial Adenomatous Polyposis (FAP)

Current international guidelines, including those from the National Comprehensive Cancer Network (NCCN) and the Japan Society for Cancer of the Colon and Rectum (JSCCR), emphasize the necessity of early and rigorous surveillance.[9,10,11] For classic FAP, annual screening should ideally begin at age 10–15 years.[10,11] Following surgical intervention, such as the total proctocolectomy performed in this patient, life-long surveillance remains mandatory. The residual rectal cuff or ileal pouch should be monitored endoscopically every 1 to 3 years, depending on the polyp burden.[10]

Furthermore, given that the APC mutation is systemic, extraintestinal surveillance—specifically annual thyroid ultrasounds and upper GI endoscopy (every 6 months to 4 years based on Spigelman staging)—is critical to managing the risk of duodenal and periampullary adenocarcinoma.[2,10] In this case, the patient's rectal adenocarcinoma might have been avoidable with timely surveillance and prophylactic surgery. Given the high penetrance of FAP, first-degree relatives should also be offered genetic testing and screening to ensure early detection.[9,12] Improved access to genetic counseling and testing in resource-limited settings remain a vital goal for reducing the morbidity associated with hereditary colorectal cancer in the Philippines.

Conclusion

This case underscores three key clinical lessons: first, persistent rectal bleeding in middle-aged patients warrants early endoscopic evaluation and should not be routinely attributed to hemorrhoidal disease; second, the presence of early-onset colorectal cancer with a diffuse adenomatous polyp burden should prompt evaluation for familial adenomatous polyposis, even in the absence of a family history; and third, delayed access to diagnostic procedures and genetic testing in resource-limited settings can lead to advanced disease at presentation. Early recognition, timely genetic testing, and appropriate surgical management remain critical to reducing morbidity and mortality in patients with FAP.

Funding: The authors have not declared a specific grant for this research from any funding agency in the public, commercial or not-for-profit sectors.

Competing interests: None declared.

Previous presentation: Presented as poster presentation at Asian Pacific Digestive Week (APDW) in Bali, Indonesia, November 23, 2024.

Patient consent for publication Obtained.

Provenance and peer review Not commissioned; externally peer reviewed IRB permission not yet secured.

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