Familial Adenomatous Polyposis: A Case Report

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Summary

Familial adenomatous polyposis (FAP) is a rare disease syndrome characterized by mutations in the Adenomatous Polyposis Coli gene. Affected people have a markedly increased lifetime risk of colorectal cancer. The patient was a 17-year-old female with a 4-year history of intermittent abdominal pain, rectal bleeding, and anal masses. A proctocolectomy with perineal resection and a permanent ileostomy were performed. The histological findings were consistent with that of FAP. FAP can present in young patients with rectal bleeding and anal masses.

Keywords: Familial adenomatous polyposis, Anal mass, Rectal bleeding, Proctocolectomy

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Introduction

Familial adenomatous polyposis (FAP) is a rare autosomal dominant condition characterized by mutation on Adenomatous Polyposis Coli (APC) gene. The classic presentation is the growth of hundreds to thousands of polyps throughout the colon. Eventually, this syndrome leads to a nearly 100% lifetime risk of colorectal cancer (CRC), which usually manifests by the age of 40 (1). The prevalence of FAP is 1 in 8000 and equally affects both genders. It makes up 1% of all colorectal malignancies (2). High-risk individuals for FAP include first-degree relatives, individuals with >10 cumulative colorectal adenomas, or colorectal adenomas in combination with extracolonic features (3). Most polyps start growing after adolescence and are usually diagnosed in the third decade of life (3). Symptomatic patients may present with tenesmus, altered bowel habits, abdominal pain, rectal bleeding, and occasionally signs of bowel obstruction. Polyposis is confirmed by colonoscopy (4). FAP may present with a number of extracolonic manifestations, including a 10% lifetime risk of developing periampullary and duodenal cancers. Approximately 90% of cases have associated gastric polyps, with few having the potential to develop into malignancy. Fifteen percent have a history of desmoid tumors. Other pathologies include papillary thyroid cancers, congenital hypertrophy of the retinal pigment epithelium, epidermal cysts, osteomas, and hepatoblastoma based on published data (3). The National Comprehensive Cancer Network has customized surveillance guidelines for each of these manifestations, with some starting as early as 20 years (3, 4). Early detection and appropriate treatment planning are predicted by adequate screening programs, which are not optimal in sub-Saharan Africa due to

limited healthcare infrastructures (5). The cornerstone of treatment continues to be the surgical resection of colon and rectal tissue at risk of malignancy. To do this, there are several resection and reconstruction options available (5).

FAP being rare, there is not much documentation on the condition in Africa aside from sporadic case reports (1,4). Although most of those reports describe certain similarities with our case, they, however, have patients presenting at a later age. Given the rarity of this condition, its early presentation, and the fact that our screening programs are not the most effective as colonoscopies are still expensive and not readily available for all our population, we wanted to draw attention to the need of having a high suspicion index of FAP in the differential diagnosis of rectal bleeding and expose some of the challenges faced in the management.

Case Presentation

We present a 17-year-old female whose history dated 4 years prior to presentation, with gradual onset of diffuse abdominal pain and per rectal bleeding, along with palpable anal masses. There was associated history of intermittent vomiting. The anal masses were excised 1 year following the onset of symptoms, on suspicion of hemorrhoids, no biopsy was done, but the masses recurred. She then reported the onset of jaundice and generalized body swelling and was admitted for liver injury. There was marked improvement following admission, but the cause was not found. The abdominal pains and per rectal bleeding worsened over the last year of symptoms. She also noticed the onset of easy fatigability and weight loss. She does not report any relatives with a history of colon malignancy and has not had her menarche. On admission, she was clinically sick looking, with diffuse abdominal tenderness but no organomegaly. A rectal examination found multiple masses in rectum and anal canal, with a bloodstained examining finger. Her hemoglobin level was at 9 g/dl microcytic hypochromic, and her albumin was low at 20. Electrolytes, kidney and liver function tests were within normal ranges. A colonoscopy done revealed numerous polyps (more than 100) of various sizes in the entire colon, including rectum and anal canal (Figure 1) concluding on FAP. The terminal ileum was spared of polyps.

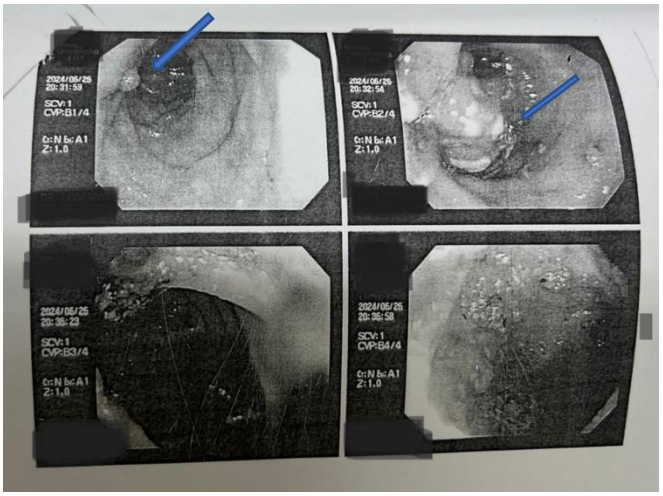

Figure 1. Image showing the colonoscopy result of the patient. Arrows point to various polyps of different sizes and shapes.

An abdominal computed tomography scan showed distal colon wall thickening, mesenteric nodes, and diffuse hepatic steatosis, with a normal-sized uterus and no adnexal lesions. The worsening abdominal pains and rectal bleeding and the investigation findings prompted the decision to optimized the patient in view of surgical intervention, where a total proctocolectomy and perineal resection was performed with a permanent end ileostomy fashioned. This procedure was selected because of the wide dispersion of polyps up to the anal canal and the possibility of cancer.

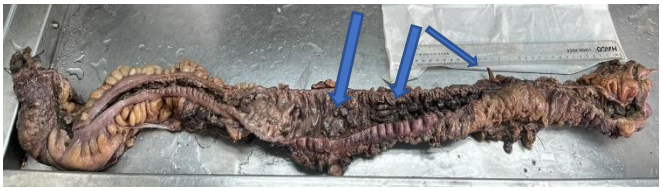

Figure 2. Gross specimen of the entire colon and rectum. Arrows point to polyps.

Pathological assessment of the sample consisted of 2 cm of terminal ileum, 9 cm appendix, colon (ascending, transverse, descending) measuring 79 cm, and a rectum of 5 cm in length (Figure2). All containing multiple pedunculated polyps of range 5–20 mm in diameter, with the nearest polyp to the proximal margin at 35 mm

and nearest to the distal margin at 15mm. sections (Figure 3) showed adenoma with glands of variable sizes lined by benign epithelium with focal pseudo-stratification and low-grade dysplasia with necrosis seen within some of the glands but no evidence of malignancy. Mesenteric nodes were reactive nodes with no malignant cells. Genetic analysis was not done.

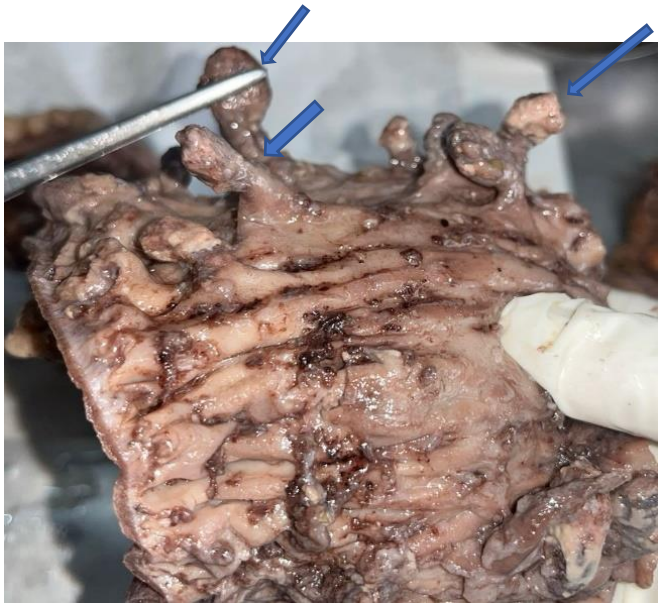

Figure 3. Cross-section of dissected colon segment showing multiple polyps of varying length, shape, and form. These could be found in the entire colon until the anal canal. Arrows point to polyps.

The patient developed a surgical site infection post–operatively that was managed with wound care. She was counseled on lifelong stoma use and the need to have a tailored screening scheduled for surveillance, such as upper gastrointestinal endoscopy, hepatic, thyroid, and ophthalmologic evaluations. Written informed consent was obtained from the patient for the publication of this case report.

Discussion

The diagnosis of FAP is uncommon in Africa. The majority of publications on the subject are isolated case studies. The fact that our patient was only 17 years old made this report necessary in order to raise awareness. Just like in other reports (1, 4), the patients were in their early 20s. However, Pasquer et al. reported that the mean age at presentation was 29.5 years in their four-decade systematic review (6). The patient had been experiencing symptoms for 4 years before a proper diagnosis was made. This was a lengthy period of time that could be explained by the doctors' low level of suspicion, financial limitations, and the lack of easily accessible colonoscopies in low-income areas. This presentation is similar to the report from a case in Uganda (4). A third of patients with FAP have no relatives with a history of polyposis or malignancy; hence, it is not unusual that our patient's siblings are healthy while reporting no relatives with colon cancer. However, her family members are unwilling to perform colonoscopies for screening, drawing attention to a problem with cultural issues and diagnostic anxiety in African communities. (7). In sub-Saharan Africa, the urgency of developing efficient screening policies was highlighted in all the other case reports. This is mostly because their patients usually presented after developing CRC, and the outcome at that stage is poor owing to the difficulties in management in our facilities (1, 4).

When fewer than five rectal adenomas are discovered during the preoperative workup, rectal preservation is typically advised. Total proctocolectomy is still the gold standard when more than 20 adenomas are discovered. Total proctocolectomy plus perineal resection was the least chosen surgical procedure (2.4%) according to Pasquer and colleagues' systematic evaluation (6). Total colectomy plus ileorectal anastomosis was chosen in the majority of cases (67%), and proctocolectomy with ileoanal anastomosis in 32% of instances. The average cancer-free survival for both groups was 46.5 years (6). A more conservative approach is made possible by technological advancements in endoscopy, which enable the resection of any remaining adenomas in the rectal stump. This was made possible by advancements in cold snare resection, though close observation will still be required to quickly act in case of recurrence (3). With appropriate psychosocial counseling and education, the patient's concerns about her quality of life with a permanent ileostomy were addressed, and the benefits of this treatment over alternative methods were discussed. Patients with anastomosis have a comparatively greater quality of life

even if they are at risk of leaks, fecal incontinence, recurrence, and will need continuous surveillance (8).

Gross pathology in our case showed multiple pedunculated polyps involving the entire colon and rectum with evidence of low-grade dysplasia. This is similar to documented reports on histopathologic analysis of colon in patients with FAP (9). Histologically, the polyps demonstrate irregular-shaped glands, which are dysplastic, enlarged, pseudostratified with smaller to absent goblet vacuoles, akin to our observation. Prominent nucleoli and increased mitosis may be seen at higher magnification. These histological changes are typically seen in dysplasia of glandular epithelium (4). Our patient showed no signs of cancer, despite the fact that 25% of colectomy procedures for FAP are associated with carcinoma (6). Genetic research could have helped our patient identify the exact form of FAP syndrome she had and receive specialized treatment, but it was not done. Patients from low-income nations encounter these difficulties since most patients cannot pay the high cost of molecular investigations and only a small number of private sector clinical laboratories assist physicians in the molecular diagnosis of cancer (1, 5).

Although FAP includes several extracolonic symptoms, our patient's history of symptomatic liver illness was ambiguous, and additional testing did not identify a chronic problem. There are no explicit guidelines on surveillance for liver disorders linked to FAP, and liver pathologies other than hepatoblastomas in very young children are rarely reported (9). In addition, she recalls never having had a period at the age of 17, which is unusual for patients with FAP. Other indicators of failure of secondary sexual development were also observed. Instead of being a related manifestation of FAP, this could simply be a transient clinical state caused by several stressors to the body, such as a chronic sickness (10). She is still at risk for a number of other cancers linked to FAP, the majority of which manifests later in life. Therefore, careful observation and adequate surveillance are still required, even if the chance of developing CRC later in life is virtually zero. A number of these potential extracolonic diseases have been extensively recorded in the literature, along with their respective surveillance protocols (2, 11).

The main limitations in this case were the fact that the patient had some investigations done in different facilities as images and reports were not of best quality, the poor documentation of patient's medical history concerning her admission for jaundice and the absent biopsy for the excised anal mass.

Conclusion

FAP is a rare condition; however, it should be suspected in young patients with rectal bleeding and anal masses. Because it can present very early, physicians ought to have a high index of suspicion. The polyps in affected people can involve the entire colon, including the anal canal. Challenges regarding the management of our patients varied, including the difficulty of proper screening and diagnosing, and the availability of genetic analysis.

Author contributions

FAB led in conceptualization, visualization and writing of the original draft. All authors equally contributed to supervision and reviewing & editing of the original draft.

References

1. Kindie EA, Beyera TD, et al. Familial adenomatous polyposis: a case report. J Med Case Rep. 2024; 18: 415.
2. Menon G, Carr S, Kasi A. Familial adenomatous polyposis. StatPearls Publishing. 2024.
3. Sammour T, Hayes IP, et al. Familial colorectal cancer syndromes: an overview of clinical management. Expert Rev Gastroenterol Hepatol. 2015; 9: 757-64.
4. Wismayer R. Familial Adenomatous Polyposis Coli in East Africa: a case report and review of the literature. JAMMR. 2020;32 : 74-80.
5. Khan SZ, Lengyel CG. Challenges in the management of colorectal cancer in low- and middle-income countries. Cancer Treat Res Commun. 2023; 35: 100705.
6. Pasquer A, Benech N, et al. Prophylactic colectomy and rectal preservation in FAP: systematic endoscopic follow-up and adenoma destruction changes natural history of polyposis. Endosc Int Open. 2021; 09: E1014-22.
7. Lee R, Holmes D. Barriers and recommendations for colorectal cancer screening in Africa. Glob Health Action. 2023; 16: 2181920.

8. Monahan KJ, Bradshaw N, et al. Guidelines for the management of hereditary colorectal cancer from the British Society of Gastroenterology (BSG)/Association of Coloproctology of Great Britain and Ireland (ACPGBI)/United Kingdom Cancer Genetics Group (UKCGG). Gut. 2020; 69: 411-44.
9. Kanth P, Grimmrtt J, Champine M, et al. Hereditary colorectal polyposis and cancer syndromes: a primer on diagnosis and management. Am J Gastroenterol. 2017; 112: 1509-25.
10. Tang C, Zafar Gondal A, Damian M. Delayed puberty. StatPearls Publishing. 2024.
11. Dinarvand P, Davaro EP, et al. Familial adenomatous polyposis syndrome: an update and review of extraintestinal manifestations. Arch Pathol Lab Med. 2019; 143: 1382-98.