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Case Report

Familial Adenomatous Polyposis In A Nigerian Adolescent: A Case Report Of A Rare Hereditary Colonic Disorder.

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Abstract

Familial Adenomatous Polyposis (FAP) is a rare inherited disorder characterized by early onset of numerous adenomatous polyps in the colon with near certainty of colorectal cancer if untreated. Though it is quite uncommon in the African population, increasing diagnostic access is bringing more cases to clinical attention. We present a case of a 16-year-old Nigerian girl who presented with history of recurrent diarrhea, abdominal discomfort, hematochezia and weight loss. She was diagnosed of FAP following colonoscopy and histological confirmation. She was counseled on the treatment options and the possible sequelae of colorectal malignancy. The significance of this case report lies in the rarity of FAP among Nigerians and the high index of suspicion needed to prompt colonoscopy evaluation.

Keywords: Familial Adenomatous Polyposis, Colorectal Polyps, Nigeria, Adolescent.

INTRODUCTION

Familial Adenomatous Polyposis (FAP) is an autosomal dominant disorder caused by mutations in the Adenomatous Polyposis Coli (APC) gene located on chromosome 5q21.[1] It predisposes affected individuals to the development of numerous colorectal adenomatous polyps – often in hundreds to thousands – and ultimately colorectal cancer, usually by the fourth decade of life if untreated.[1] The condition may also have extracolonic manifestations of adenomatous polyps in other parts of the digestive tract including the duodenum, stomach, and even gallbladder.[2]

Though widely reported in developed nations, FAP is rarely diagnosed in sub-Saharan Africa presumably due to limited access to advanced gastrointestinal diagnostics like colonoscopy and genetic testing.[3] Early detection remains key in preventing development of colorectal cancers.

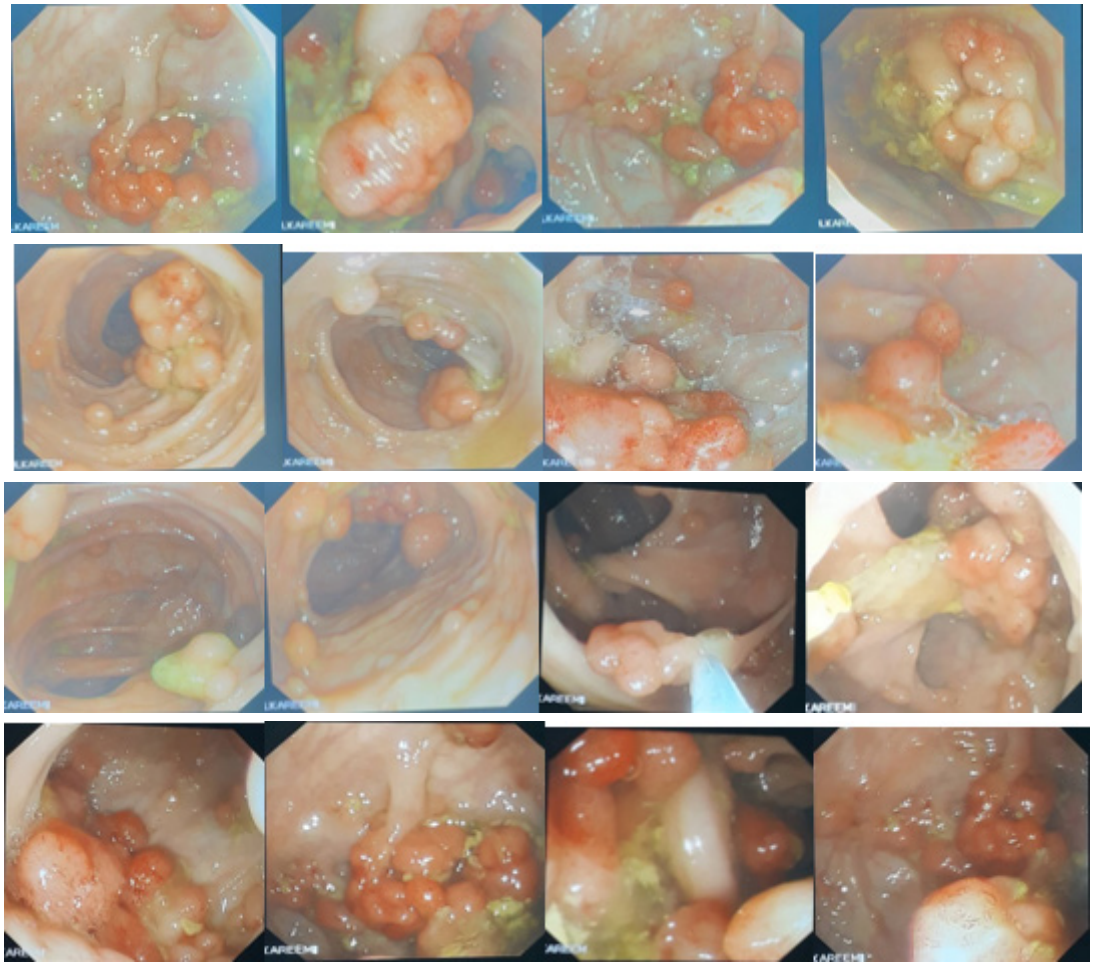
CASE PRESENTATION

We present a 16-year-old female who presented to our clinic with a two-year history of recurrent passage of loose stools, rectal bleeding, abdominal discomfort and weight loss. Her symptoms had progressively worsened over the preceding three months necessitating her presentation for thorough evaluation. She had a family history of recurrent rectal bleeding in her maternal grandmother, but no similar history in her parents or siblings. However, her grandmother and parent had never had a colonoscopy done. Our index case had an upper gastrointestinal endoscopy done in another health facility a few months prior to her presentation to our clinic. This showed a lone duodenal polyp (<1 cm) in the second part of the duodenum, but normal gastric and oesophageal mucosa. Histology of duodenal polyp was reported as an "adenomatous polyp with low-grade dysplasia". Colonoscopy could not be done at that time due to faulty colonoscope and this necessitated her presentation to our clinic. Significant examination findings at presentation included mild pallor and mild vague abdominal tenderness. Laboratory investigations included full blood count, erythrocyte sedimentation rate (ESR), carcinoembryonic antigen (CEA), screenings for hepatitis B surface antigen (HbsAg) and Hepatitis C virus antibodies (anti-HCV) as well as HIV. Her results were as follows: PCV of 33%, with decreased MCV, MCH, and MCHC. Total white blood cell count was 3.87 x10^9/l, and CEA was normal (1.56 ng/l). Screening for HIV and viral hepatitis B and

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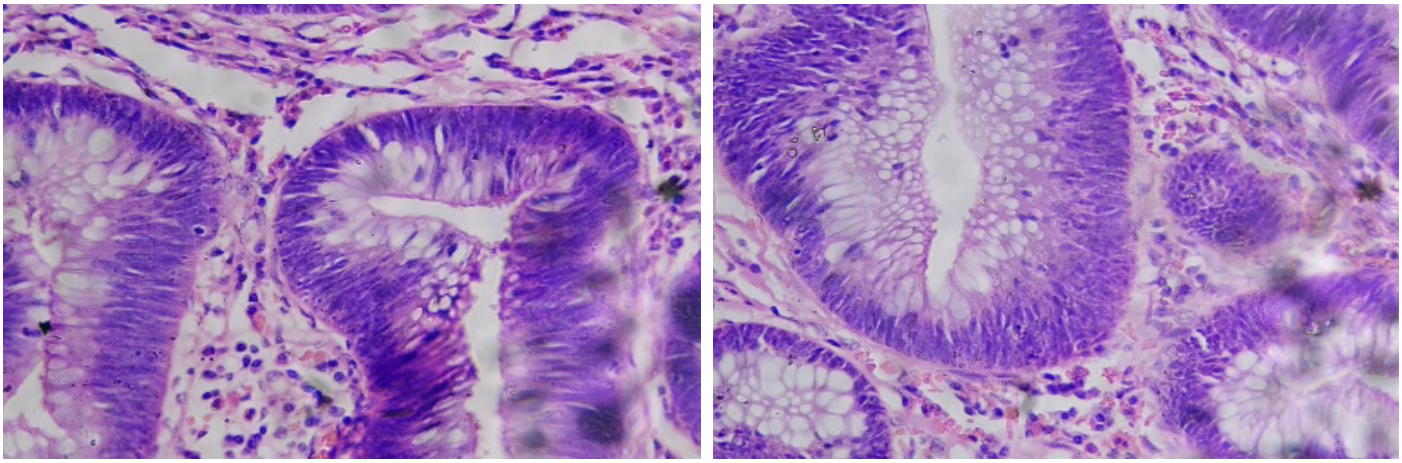
C were negative. Her abdominal ultrasound scan did not detect any abnormality. A full-length colonoscopy was performed, revealing innumerable (hundreds) sessile and pedunculated polyps distributed throughout the colon. The polyps in the rectum were sessile and smaller (<0.7cm) while larger polyps were seen extending from the sigmoid to the ascending colon and caecum (**figure 1**). Some small polyps in the rectum were excised with biopsy forceps and few large polyps in different parts of the colon were excised with hot polypectomy snare and sent to the histopathologist for histologic assessment.

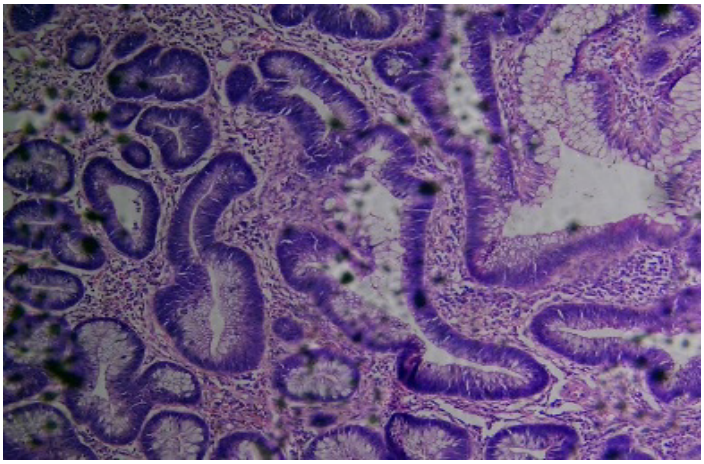
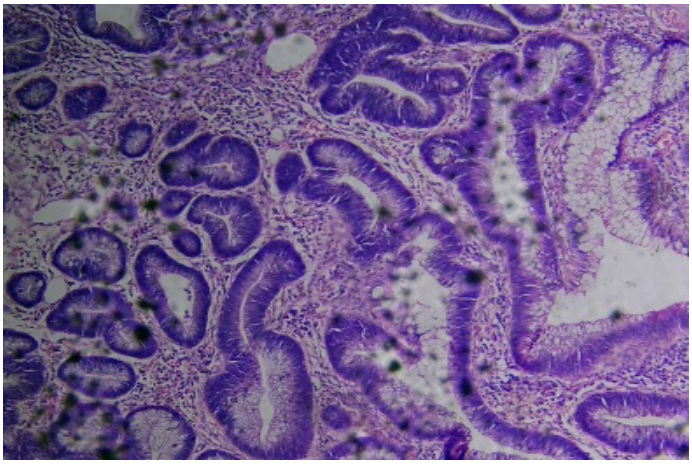
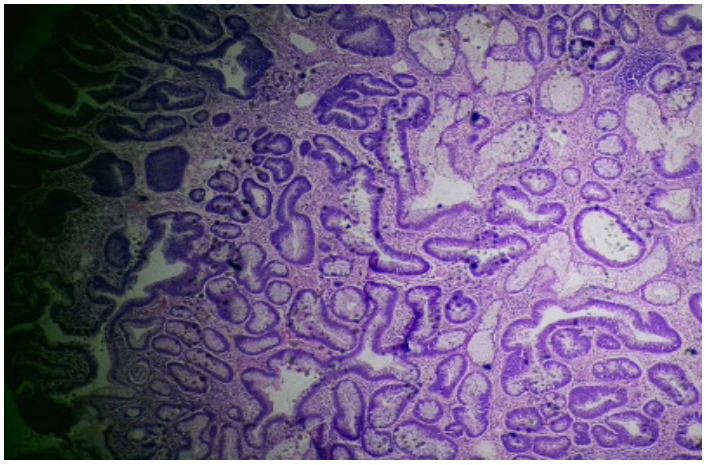
Figure 1. Colonoscopy images of Z.B showing numerous polyps of varying sizes and morphology.



Histopathology report of the polyps showed "overlying cuboidal epithelial cells with foci of moderate dysplasia. The lamina propria show fairly rounded-shaped spaced glands with dysplastic epithelial lining and reduced apical mucin production. Seen elsewhere were unremarkable glands and mixed inflammatory cells." These features were consistent with "Tubular Adenoma with Moderate Dysplasia". See **figure 2**.

Figure 2. Photomicrograph of our patient's histology showing features of tubular adenomas with moderate dysplasia x100.



Based on the presentation as well as the colonoscopy features and histology report, a diagnosis of FAP was made. She and her parents were counseled on doing genetic testing for FAP to confirm the diagnosis however, facility for this was lacking in our region. Her parents were also counseled on the need for them to have colonoscopy done in order to screen them for possible FAP. Our index case was counseled along with her parents on the implication of her diagnosis and treatment options, both adjunctive and surgical, particularly colectomy in order to prevent the likely sequelae of colorectal cancer later in life. Her management by a multi-disciplinary team comprising her Paediatrician, Gastroenterologist and a Colorectal Surgeon was discussed and a follow up was scheduled.

DISCUSSION

Familial Adenomatous Polyposis (FAP) is a rare autosomal dominant inherited disorder resulting from mutations in the Adenomatous Polyposis Coli (APC) gene located on chromosome 5q21.[1] This gene acts as a tumor suppressor gene and plays an improtant role in regulating cell growth through the Wnt signaling pathway.[4] The APC gene's tumor suppressor function is lost in FAP due to a germline mutation, leading to abnormal accumulation of the protein beta-catenin in the colonic epithelial cells.[5] This excess beta-catenin activates the Wnt signaling pathway, promoting uncontrolled cellular proliferation and ultimately forming hundreds to thousands of adenomatous polyps in the colon and rectum.[5] Polyps usually begin to form in teenage years and over time, polyp burden increases. The risk of colorectal cancer approaches 100% by age 40 in untreated individuals.[6]

FAP is the second most common inherited polyposis syndrome. The incidence rate is estimated to be about 1 in 5000 and 1 in 18,000 and affecting men and women equally.[6] It accounts for approximately 1% of colorectal cancer cases.[7] Approximately About 20-30% of FAP cases arise from a new (de novo) mutation in the APC gene and lack a documented family history of FAP.[6] It is quite a very rare disorder in in Nigeria. While specific prevalence rates are not readily available, studies and reports indicate a significant paucity of reported cases, with only a few documented instances over decades, suggesting a much lower occurrence compared to developed countries.[8-10]

FAP may be asymptomatic in some individuals with low polyp burden and may be picked during imaging or screening colonoscopy. Some may present with abdominal symptoms such as abdominal pains, change in bowel habit, change in stool caliber, hematochezia and weight loss. These were some of the symptoms our index case presented with. Complications such as intestinal obstruction may be a presenting feature in some individuals with FAP.[9] There may be a family history of FAP one of the parents, though no family history may be obtained in cases with de novo mutations. While neither parent of our index case had history to suggest FAP, her maternal

grandmother had history of recurrent rectal bleeds which had not been investigated by colonoscopy. The presence of multiple (usually hundreds to thousands) polyps across the entire colon and rectum on colonoscopy is the hallmark of diagnosis. Histological examination of the biopsied polyps confirmed their adenomatous nature, clinching the diagnosis of FAP. While genetic testing is recommended to confirm the APC gene mutation and facilitate family screening, it is often unavailable or unaffordable in resource-limited settings like Nigeria.

Management of FAP requires a multidisciplinary approach. Colonoscopy surveillance should begin in the early teens for at-risk individuals. Once the polyp burden becomes significant, prophylactic colectomy is indicated to prevent colonic cancers. Recommended treatment is surgical and options include a total proctocolectomy with ileal pouch-anal anastomosis or subtotal colectomy with ileo-rectal anastomosis. The latter is recommended if rectal polyp burden is minimal. Generally, colectomy is indicated if there is presence of multiple polyps > 10 mm, high grade dysplastic polyps or a rapid increase in the number of polyps.[11] Timing of colectomy should however, be a shared decision with the patient, taking into account socio-cultural, psychological and career factors.

Adjunct therapies include Non-Steroidal Anti-Inflammatory Drugs or Cyclooxygenase-2 (COX-2) inhibitors, which may reduce polyp burden but do not eliminate the risk of colonic cancer.[12] Treatment with Sulindac has been reported to significant reduce polyp count and diameter compared with treatment with placebo and in post-colectomy patients it was found to significantly reduce rectal polyp number grade of dysplasia.[12] Celecoxib has shown similar effect on reducing polyp burden.[12] Genetic counseling and testing of first-degree relatives are essential for early detection and prophylactic care. This case illustrates the importance of colonoscopy in the early diagnosis of FAP in adolescents with persistent gastrointestinal symptoms particularly hematochezia with or without a typical family history. Paucity of diagnostic colonoscopy and genetic testing in Nigeria and other low-middle income countries may pose a challenge to diagnosis of FAP. Prompt diagnosis, counseling and a proactive multidisciplinary approach to management are crucial in preventing progression to colorectal carcinoma.

CONCLUSION

This case highlights the need for heightened clinical suspicion for FAP in young patients presenting with gastrointestinal bleeding, chronic diarrhea, and weight loss. Improved access to colonoscopy and laboratory evaluation in Nigeria and other low/middle-income countries will enhance early diagnosis and timely intervention, reducing the burden of preventable colorectal malignancy.

DECLARATIONS

Conflict Of Interest: The authors have no conflicts of interest to declare.

Authors Contribution: All the authors were involved in the management of the patient and contributed to the writing of the manuscript. All the authors gave approval for the manuscript to be published.

Ethical Consideration: The patient and her parent gave oral consent to publish this case report.

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