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*Corresponding author

*Nguyen Ngoc Huan. MD, Digestive Surgery Department, Cho Ray Hospital, Viet Nam.

Familial Adenomatous Polyposis: A Case Report

Nguyen Ngoc Huan*, Ly Pham Quoc Hau. MD,

Department of Digestive Surgery, Cho Ray Hospital, Viet Nam

Abstract

Colon polyps, which are atypical growths on the colonic lining, exhibit significant variability in size, morphology, and risk for malignancy. Although numerous polyps are nonmalignant, several types possess the capacity to progress into colorectal cancer over time. When numerous colon polyps are identified, particularly in high-risk individuals or those with familial adenomatous polyposis (FAP), a predominant genetic syndrome marked by aberrant cell proliferation, its most prevalent manifestation is the formation of extensive colorectal adenomas, ranging from hundreds to thousands. Surgical intervention may be required to excise these tumors and reduce the risk of cancer progression. We present a clinical case of a 31-year-old male admitted to the Digestive Surgery Department at Cho Ray Hospital with gastrointestinal complaints: constipation, persistent abdominal discomfort, and intermittent rectal hemorrhage. The colonoscopy findings revealed many polyps in the colon. Histological analysis verified the presence of a tubular adenoma with low-grade dysplasia. The patient underwent laparoscopic total proctocolectomy for the treatment of colon polyps, accompanied with ileoanal anastomosis and ileostomy. The patient was discharged home seven days after surgery, without complications. Surgical surgery for numerous colon polyps or familial adenomatous polyposis can diminish the risk of colorectal cancer. The objective is to efficiently excise polyps and avert their progression to malignancy, utilizing minimally invasive methods such as endoscopic polypectomy, endoscopic colectomy, or total colectomy.

Introduction

Familial adenomatous polyposis (FAP) is an inherited disorder characterized by many adenomatous polyps in the gastrointestinal mucosa, leading to an almost 100% lifetime risk of colorectal cancer (CRC) and an incidence rate of 1 in 7,000 to 1 in 30,000 live births [1,2]. Both males and females have an identical probability of developing the disorder, regardless of the parents' gender, with an average onset happening after the age of 15 [3]. Before this age, the disease is recorded as asymptomatic and lacking any indications [4].

Familial adenomatous polyposis (FAP) frequently results from a germline pathogenic mutation in the adenomatous polyposis coli (APC) tumor suppressor gene situated on chromosome 5 [5]. This mutation mostly results in the formation of over 100 colorectal adenomatous polyps [6]. Most patients usually do not have symptoms, but they may present with diarrhea, rectal bleeding, abdominal pain, nausea, and obstruction, typically occurring in the age group of 20 to 30 years [7].

The risk of developing colorectal cancer is approximately 87% when the patient reaches 45 years of age. Patients who are not treated will have a risk of dying from colon cancer before the age of 40. The disease is often detected in the symptomatic stage due to blood in the stool. The best treatment method is a total colectomy with ileorectal anastomosis. In the case of malignant changes in the rectum, a total colectomy and a complete rectal resection with an ileostomy are necessary.

Case presentation

The patient is a 31-year-old male, admitted to the gastrointestinal surgery

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department with digestive issues: constipation, persistent abdominal pain, and occasional rectal bleeding. Upon examination, the patient showed no special signs. Blood tests show that the patient has mild anemia: RBC: 3.3 T/L; HGB: 105g/L; HCT: 30%; WBC: 7.3 G/L; CEA 4 ng/ml.

The colonoscopy results show multiple polyps in the colon (Figure 1)

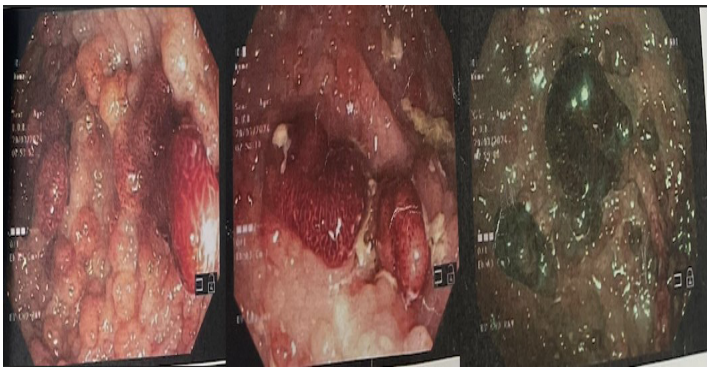

Figure 1: *Colonoscopy with presence of multiple polyps*

Histological examination confirmed tubular adenoma with low-grade dysplasia. Abdominal and pelvic CT normal. After discussing the treatment method with the patient and their family, the patient underwent laparoscopic total colectomy with multiple polyps, ileoanal anastomosis, and the ileal stoma was brought to the skin to protect the anastomosis. After the surgery, the colon was opened and examined, revealing numerous polyps covering the entire colonic mucosa (Figure 2)

The surgery duration was 262 minutes, with a blood loss of 125ml, and the time to pass gas was 3 days. The patient was given sugar water on the first day after surgery and solid food on the second day after surgery. After 7 days in the hospital, with no complications, the patient was discharged home. The patient was re-examined after 1 month with stable clinical symptoms. Three months after the first surgery, the patient was hospitalized to close the ileostomy. The patient was discharged from the hospital 5 days after the surgery. Regular follow-up every 3 months and no abnormal signs within 1 year.

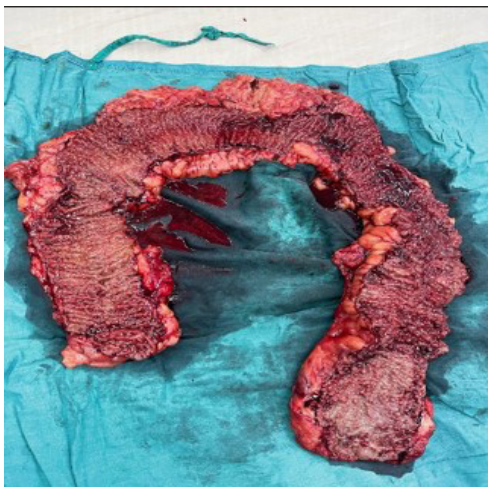

Figure 2: *The colon after resection has many polyps on the mucosal surface.*

Discussion

Familial adenomatous polyposis (FAP) is an autosomal dominant disorder with high penetrance, affecting both sexes equally. Approximately 20–30% of cases may arise from de novo mutations, although the majority of affected individuals have a familial history of FAP syndrome [8]. The patient's father experienced a similar ailment; however, there have been no signs of the condition in the other siblings thus far. CT scans and colonoscopy are essential for diagnosing and assessing rectal bleeding, particularly in young patients with high-risk factors such anemia and a substantial family history [9]. The diagnosis in our case was conducted using abdominal CT and colonoscopy to assess rectal bleeding, anemia, and a familial history of relatives with many colorectal polyps. The development of many adenomatous polyps in the colon and rectum, beginning at birth and almost usually progressing to colorectal cancer by age 40, is a characteristic hallmark of FAP [6]. The diagnosis in our case was conclusively determined after examining the resected colon, which disclosed more than 1000 polyps devoid of any malignant changes in the adenomas. The molecular characterisation of APC is crucial for confirming the diagnosis of FAP [6]. Accurate diagnosis of FAP requires comprehensive sampling of polyps for pathological analysis and germline mutation assessment, alongside detailed evaluations of clinical manifestations, endoscopic images, and genetic tests, rather than relying solely on the presence or absence of adenomatous polyps [10]. Annual screening for classic familial adenomatous polyposis (FAP) with flexible sigmoidoscopy or colonoscopy often begins at ages 10 to 12. Consequently, among individuals with familial adenomatous polyposis (FAP), screening colonoscopy coupled with prompt intervention for detected lesions has resulted in a 55% decrease in the colorectal cancer incidence, marking the initial indicator of enhanced survival rates for patients [11]. The patient's family members are uninformed of the ailment and the possible cause of the father's death.

Surgery is essential in the care after a diagnosis of FAP, with typical surgical options comprising total colectomy (accompanied by a Brooke ileostomy or ileal pouch-anal anastomosis) and subtotal colectomy with ileo-rectal anastomosis [12]. Indications for colectomy encompass symptomatic polyps, advanced adenomas including colorectal cancer, severe or progressive polyps, a polyposis load unmanageable by endoscopy, or situations where surveillance is unfeasible [13]. In young patients with a prolonged life expectancy and no advancing dysplasia or malignancy, preventative surgery involving ileorectal

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anastomosis (IRA) facilitates rectal preservation and should be contemplated to improve quality of life without compromising prognosis [14]. Endoscopic complete proctocolectomy with ileorectal anastomosis is a safe and practical prophylactic procedure for young patients, characterized by a low risk of complications and favorable outcomes [15]. The patient underwent total rectal resection through laparoscopy with ileal pouch-anal anastomosis due to rectal polyps, accompanied by loop ileostomy.

The risk of upper gastrointestinal cancer and desmoid tumors in patients with familial adenomatous polyposis persists post-colectomy, resulting in postoperative morbidity or diminished lifespan [16]. Regular endoscopic or colonoscopy evaluations of the anastomosis site, pouch, and residual rectum are advised post-surgery every 6 to 12 months [17]. Annual monitoring is crucial in the context of residual rectum [18]. Our patients undergo constant monitoring and supervision. Furthermore, persons with first-degree relatives diagnosed with familial adenomatous polyposis (FAP), those exhibiting over 10 to 20 intestinal polyps, and/or patients with colonic adenomas linked to extraintestinal symptoms of FAP are at elevated risk for the condition and should consider genetic testing [13]. None of the family members in our case have yet taken genetic testing and are hesitant to pursue it due to the family's financial circumstances.

Conclusion

Effective management of patients with FAP is crucial to guarantee proper screening and suitability for these intricate cases. Timely colonoscopic screening is crucial for establishing the optimal schedule for surgical resection. While medicinal interventions can offer substantial assistance, the primary approach for treating FAP is colectomy, with or without proctectomy. Patients with FAP necessitate comprehensive screening protocols and diligent clinical monitoring for extraintestinal symptoms. Through surveillance and surgical excision, individuals with FAP can markedly diminish the risk of colorectal carcinoma and other associated neoplasms. Laparoscopic surgery for the treatment of multiple colon polyps appears feasible and safe, with a low complication rate. When the techniques and instruments for endoscopic colon surgery are perfected, this procedure could become an attractive option for managing patients with multiple colon polyps.

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