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Case Presentation

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Familial Adenomatous Polyposis: A Generational Disease Managed by Generations of Surgeons with an Interesting Indication for Intersphincteric Proctectomy

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Abstract

Background: Classic Familial Adenomatous Polyposis (FAP) is an inherited disorder in which hundreds to thousands of adenomatous polyps develop in the gastrointestinal (GI) tract, predominantly in the colon and rectum. Most patients with classic FAP initially present with nonspecific GI symptoms such as diarrhea, abdominal discomfort, or rectal bleeding. The American Society of Colon and Rectal Surgeons recommends that polyposis syndromes be considered in patients with greater than 20 lifetime adenomas, a personal history of desmoid tumor or other extracolonic manifestations of FAP, or in individuals with known family history of FAP or other subtypes of FAP. Screening individuals with a family history of Classic Familial Adenomatous Polyposis is essential to survival, as colorectal cancer is inevitable with a diagnosis of FAP.

Case Presentation: We present multiple examples of cases of classic FAP across several generations of one family including an uncommon indication for intersphincteric proctectomy, all of whom are being treated by their original surgeon Dr. Charles Lago along with his son, Dr. Nicholas Lago.

Conclusion: The mainstay treatment of FAP is colectomy with or without proctectomy. Therefore, an intersphincteric proctectomy should be considered a safe and suitable option for patients with retained rectum and non-oncologic indications for resection.

Keywords: Intersphincteric proctectomy; Familial adenomatous polyposis; Restorative proctocolectomy; J-pouch; General surgery residency

Introduction

Classic Familial Adenomatous Polyposis (FAP) is an inherited disorder in which hundreds to thousands of adenomatous polyps develop in the gastrointestinal (GI) tract, predominantly in the colon and rectum. The adenomatous polyposis coli (APC) gene located on chromosome 5q21-q22 is responsible for classic FAP [1]. The APC gene is a tumor suppressor that functions to promote apoptosis in colonic epithelial cells [1]. Classic FAP with an APC gene mutation follows an autosomal dominant inheritance pattern, resulting in multiple generations of the disease; other subtypes of FAP follow different inheritance patterns and can involve different gene mutations. More than 1,000 different mutations of the APC gene have been discovered to be associated with FAP. The location of the mutation within the APC gene is associated with the severity of colonic polyposis, degree of cancer risk, age of cancer onset, survival, and the presence and frequency of extracolonic manifestations [2,3]. Identification of a heterozygous germline

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pathogenic variant in the APC gene leads to uncontrolled cell growth, resulting in polyps of the large intestine and rectum, and sometimes in the upper GI tract and small intestines [4]. If not screened, detected, and removed early enough, the polyps have a nearly 100% chance of becoming cancerous [5].

Case Presentation

We present multiple examples of cases of classic FAP across several generations of one family including an uncommon indication for intersphincteric proctectomy, all of whom are being treated by two generations of surgeons.

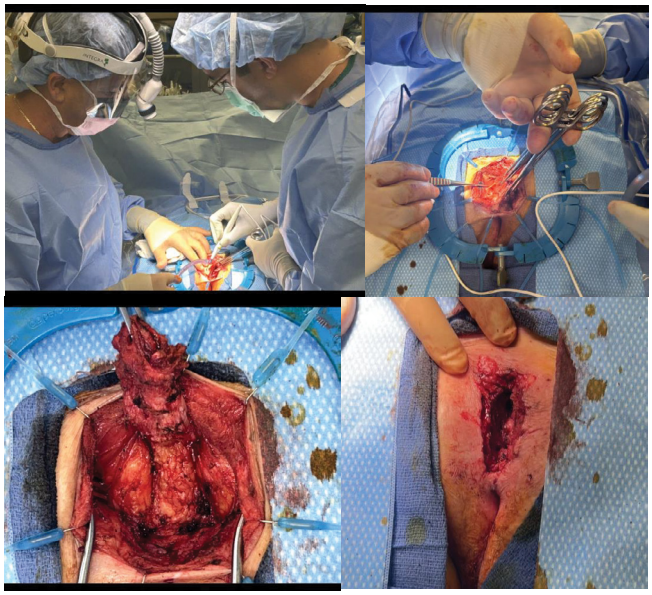
A 51-year-old female presented in 1997 complaining of bright red blood per rectum and underwent a colonoscopy which revealed hundreds of polyps throughout the entirety of her colon, however, sparing the rectum. She was diagnosed with FAP and underwent a subtotal colectomy with ileostomy. Although the appropriate surgical option is a restorative proctocolectomy with J-pouch ileoanal anastomosis, the patient adamantly refused the removal of her rectum and agreed to an annual proctoscopy. Since her rectum was spared of polyps, it was agreed to proceed with subtotal colectomy with ileorectal anastomosis. However, due to technical issues, anastomosis was not performed. She recovered well post-operatively and continued with her screenings. When offered reversal of her ileostomy, she refused another operation stating she was comfortable with her ostomy. The second generation: She had two daughters who were subsequently diagnosed with FAP in their 20s, but who had significantly different clinical outcomes. One agreed to genetic testing and colonoscopy in her early twenties when her mother was first diagnosed. She was diagnosed with the disease and underwent a prophylactic restorative proctocolectomy with J-pouch ileoanal anastomosis. She is currently alive and well with two children.

The other daughter refused to get screening colonoscopies and genetic testing due to personal reasons and emotional trauma from observing her close family undergo invasive procedures and hospital admissions. One evening at the age of 38, she arrived at the emergency department complaining of significant abdominal pain and was found to have a right-sided colo-colonic intussusception resulting in a large bowel obstruction. She was taken for an emergent right hemicolectomy. Final pathology revealed stage three adenocarcinoma for which she underwent adjuvant chemotherapy. She then underwent a complete restorative proctocolectomy with J-pouch ileoanal anastomosis. Unfortunately, she was found to have a recurrence of the adenocarcinoma with metastatic disease and passed away roughly two years after initially presenting with her large bowel obstruction. The third generation: Each of these daughters had two sons, both of whom began screening at the age of 10. One son from each of these two daughters was diagnosed with FAP and underwent prophylactic restorative proctocolectomies with J-pouch ileoanal anastomosis. They are presently alive and healthy in their 30s.

One colorectal surgeon performed the operations and managed all the above-mentioned patients. Recently the first diagnosed patient in this family presented with persistent bleeding from her retained rectum. She was diagnosed with atrial fibrillation and began long-term oral anticoagulation. Although she had suffered from diversion proctitis for many years that was adequately controlled with topical non-steroidal anti-inflammatory medications, she was now chronically anemic requiring transfusions. First generation physician: Her original surgeon along with his son (second generation physician), decided to perform an intersphincteric proctectomy (Images 1-3). Distance of retained rectum was approximately 15cm. She had an uncomplicated postoperative course and is currently healthy, living with her daughter and grandchildren. Final pathology revealed proctitis with no evidence of polyps or neoplasia.

Clinical Presentation and Complications

Most patients with classic FAP initially present with nonspecific GI symptoms such as diarrhea, abdominal discomfort, or rectal bleeding [1]. The diagnosis is confirmed with the visualization of more than 100 adenomatous polyps on colonoscopy. The polyps usually begin growing during the teenage years and increase in number with age. Classic FAP can result in polyps in the duodenum, fundus of the stomach, and ampulla, where the bile and pancreatic ducts enter the small intestine in addition to colorectal polyps and cancer



Images 1-4: Two generations of surgeons depicted performing an intersphincteric proctectomy. Dissection began into the intersphincteric plane between the external and internal sphincter. Entire remaining rectum (15cm) was removed and the levator plate was reapproximated. The skin was then reapproximated with stapples (not depicted)

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[5]. Although very rare, polyps in the thyroid gland, central nervous system, adrenal glands, and liver may also develop and lead to cancer [5]. A patient with classic FAP may also develop desmoids which are abnormal growths arising from connective tissue. A physical exam may reveal congenital hypertrophy of the retinal epithelium, abnormalities in dentition, epidermoid cysts on the face, extremities, and scalp in young children, and fibromas on the extremities, back, and trunk, aside from GI signs and symptoms [1]. Classic FAP may also result in osteomas and anemia [5].

Screening and Treatment

According to the American Society of Colon and Rectal Surgeons, polyposis syndromes should be considered in patients with greater than 20 lifetime adenomas, a personal history of desmoid tumor or other extracolonic manifestations of FAP, or in individuals with known family history of FAP or other subtypes of FAP [6]. If FAP is suspected, genetic counseling and testing are recommended. Identifying a pathogenic or likely pathogenic variant, such as the APC mutation, via multigene panel testing allows for screening of asymptomatic, at-risk family members [7]. Prenatal testing and preimplantation genetic testing are also possible if a pathogenic variant has been identified in an affected family member [8].

Patients with a family history of FAP should undergo a flexible sigmoidoscopy or colonoscopy every 1-2 years starting at age 10, just as the grandsons in the case study had done.4 Annual colonoscopies should be performed once polyps are discovered until the patient undergoes a colectomy. Once colorectal polyps are found, or by the age of 25, regular upper endoscopies should also be scheduled to monitor for duodenal or ampullary polyps [9]. Ultrasounds of the thyroid should be considered due to the increased risk of thyroid cancer. Computed tomography or magnetic resonance imaging scans may also be recommended in a patient with a personal or family history of desmoid tumors [9].

Customarily, patients diagnosed with FAP should consider a form of colectomy with or without proctectomy by late adolescence or early adulthood to reduce their otherwise inevitable cancer prognosis [7]. Typical surgical options include total colectomy with ileorectal anastomosis, total proctocolectomy with ileostomy, and restorative proctocolectomy with or without mucosectomy and ileal-pouch anal anastomosis [10]. Since the rectal mucosa is still at high risk of developing adenocarcinoma, careful patient selection for rectal sparing intervention is crucial and may be considered in those with a low polyp burden within the rectum, lack of advanced rectal neoplasia, and no evidence of colorectal cancer at the time of resection [1]. In those with a retained rectum, annual surveillance is essential. Conversion to total proctocolectomy is near zero with proper surveillance but can increase to as high as 35% in patients with poor follow-up [1]. However, if patients with retained rectum present with a non-oncologic indication for resection, such as in the aforementioned case, an intersphincteric proctectomy is an effective and safe surgical option with minimal damage to the pelvic floor and nerves, leaving a smaller perineal wound compared to conventional proctectomy [11]. Observational data support potential functional benefits of sparing the rectum, including decreased bowel frequency, decreased incontinence, decreased risk of urinary and sexual dysfunction, higher chance of a single-stage operation, and avoidance of an ileostomy. Annual surveillance of the rectum is required [6].

Conclusion

Screening individuals with a family history of Classic FAP is essential to survival, as colorectal cancer is inevitable with a diagnosis of FAP. The mainstay treatment is colectomy with or without proctectomy. However, as evident in the case study, an intersphincteric proctectomy should also be considered a safe and suitable option for patients with retained rectum and non-oncologic indications for resection [12].

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