Adenocarcinoma After Ileoanal Anastomosis for Familial Adenomatous Polyposis: Review of Risk Factors and Current Surveillance Apropos of a Case

Fábio Guilherme Campos, M.D., Angelita Habr-Gama, M.D., Desidério Roberto Kiss, M.D., Edésio Vieira da Silva, M.D., Viviane Rawet, M.D., Antônio Rocco Imperiale, M.D., Rodrigo Perez, M.D., José Hyppólito da Silva, M.D., Afonso Henrique S. Sousa, Jr., M.D., Joaquim Gama-Rodrigues, M.D.

Restorative proctocolectomy has become the most common surgical option for familial adenomatous polyposis (FAP) patients, based on the premise that it provides good functional results and reduces colorectal cancer risk. But several adenomas may develop in the pouch mucosa over the years, and even cancer at the anastomosis or in the pouch mucosa has been reported rarely. This article aims to describe a case of pouch cancer after restorative proctocolectomy for FAP, reviewing the possible causes of this unfortunate outcome. A 40-year-old man started presenting with fecal blood loss 12 years after restorative proctocolectomy with mucosectomy and hand-sewn anastomosis for FAP. Proctologic examination revealed an elevated mass 3 cm from the anal margin, which biopsy determined to be a mucinous adenocarcinoma. The patient underwent pouch excision and terminal ileostomy. Histologic analysis showed a 2.2 cm mucinous adenocarcinoma between the ileal and anal mucosa (T2N0Mx) and multiple tubular microadenomas in the ileal pouch. The present case and the data presented here suggest that restorative proctocolectomy is not a "cancer-free" alternative to ileorectal anastomosis, because it does not remove the risk of metachronous intestinal neoplasia. Although the long-term risk of malignancy is not known, lifelong follow-up seems to be necessary after restorative proctocolectomy. Current recommendations for pouch surveillance are presented. (J GASTROINTEST SURG 2005;9:695–702)

KEY WORDS: Adenocarcinoma, ileal-pouch anal anastomosis, restorative proctocolectomy, familial adenomatous polyposis, cancer

INTRODUCTION

Familial adenomatous polyposis (FAP) is an autosomic hereditary disease characterized by the presence of numerous colorectal adenomatous polyps. It is associated with germinative or acquired mutations in the APC gene that predispose to cell proliferation and development of benign and malignant extracolonic manifestations in many organs.[1]

The disease may account for almost 1% of colorectal cancer (CRC) cases. The malignant evolution of colorectal polyps in the third to fourth decades of life is now practically an established, extensively documented fact, although the syndrome may present a variable biological and clinical behavior.[2] Thus, early detection, prophylactic colectomy, and family surveillance are the main steps in managing FAP patients. Furthermore, recognition and appropriate treatment of the associated extracolonic manifestations is essential to reduce disease morbidity.[3]

Surgical options include proctocolectomy and ileostomy, total abdominal colectomy with ileorectal anastomosis (IRA), and restorative proctocolectomy with an ileal pouch-anal anastomosis (RPC). Nowadays, permanent ileostomy is performed only in patients with advanced low rectal cancer or fecal incontinence.

In each patient, the surgical procedure should be selected on the basis of parameters such as age, site/number of the polyps, location of the mutation, and patient willingness to undergo regular check-ups.

From the Colorectal Surgery Division (F.G.C., A.H.-G., D.R.K., V.R., J.H.d.S., A.H.S.S.) and Department of Gastroenterology (E.V.d.S., A.R.I., R.P., J.G.-R.), Hospital das Clínicas—University of São Paulo School of Medicine, São Paulo, Brazil.
Reprint requests: Fábio Guilherme Campos, Alameda Jaú, 1477, Apt 111A, 01420-002, São Paulo (SP), Brazil. e-mail: fgmcampos@terra.com.br

1091-255X/05/$—see front matter
doi:10.1016/j.gassur.2004.10.017

In this context, IRA and RPC are surgical procedures that yield different results in terms of functional capability and oncologic radicality. When selecting the primary surgery, one must remember that although IRA exhibits good surgical and functional outcomes,[4] it has been associated with an elevated risk of metachronous rectal cancer after IRA, with rates varying from 12% to 43%.[5,6]

Since its introduction to clinical practice, RPC has been progressively modified in an attempt to improve functionality and reduce complication rates while providing control of the mucosal disease.[7] Despite some controversies, many technical advances in pouch surgery have allowed it to become the gold standard for the elective treatment of ulcerative colitis (UC) and FAP patients.[8] In the latter, this technique aims to reduce CRC risk and maintain acceptable anal function,[9] although desmoid tumors and duodenal and ileal adenomas may still develop.[10]

RPC was initially thought to abolish the risk of colorectal adenoma development in FAP patients, making surveillance of the lower gastrointestinal tract no longer necessary. But several papers have documented the appearance of pouch adenomas after RPC, usually after an interval of several years.[11] The potential for adenomatous polyp formation in the terminal ileum has been estimated to manifest in 9% to 20% of FAP patients, even 25 years after the colectomy.[12]

Furthermore, rectal mucosa may be left behind after the stapled technique (with conservation of the anal transitional zone) or after the standard Park's procedure (because of incomplete mucosectomy), exposing the patient to the risk of polyp development and subsequent malignancy.

During the last decade, the description of some pouch cancer cases definitely confirmed that RPC is not a "cancer-free" alternative to IRA.[13–21] Subsequently, as the long-term risk of the development of malignancy after RPC has been evaluated, suggestions for surveillance have been recently raised in the literature.

The present paper describes a rare case of pouch cancer after RPC for FAP, reviews the cases published in the English literature, and discusses the potential carcinogenic mechanisms that may be involved in this outcome.

CASE REPORT

A 40-year-old man with rectal bleeding and a familial history of FAP (mother, sister, and brother) was admitted for surgical treatment in December 1985. Colonoscopy showed multiple colorectal adenomatous polyps and a tumor in the upper rectum. Routine preoperative staging with CT showed no evidence of metastasis.

As a result, he underwent a restorative proctocolectomy with mucosectomy, construction of an ileal J pouch, and hand-sewn pouch–anal anastomosis. Technical steps were performed following oncologic principles, and rectal dissection was carried out down to the pelvic floor (up to the levator plane). With the aid of two Gelpi retractors to expose the distal rectum and anal canal, adrenaline solution was instilled into the submucosa in four quadrants. Circumferential mucosal dissection begun at the dentate line, progressing cranially toward the dissected rectum above.

Pouch–anal anastomosis was made with separated 4-0 Vicryl stitches, being temporarily defunctioned with a loop ileostomy. Histologic examination of the surgical specimen confirmed many tubular adenomas distributed through the colon and a 3-cm well-differentiated rectal adenocarcinoma situated 9 cm from the distal margin (Dukes A, Astler-Coller B1, T2N0M0). The 51 resected lymph nodes had no metastatic spread. Ileostomy closure was carried out 3 months later without operative complications. Genetic tests were not performed on this patient or his family.

He was clinically followed for 18 months, when he moved to another city and did not return to follow-up. Twelve years after surgical treatment (December 1997), he started to experience fecal blood loss. In March 1998, digital examination showed a right lateral elevated mass over a firm basis, located 3 cm from the anal margin and extending cranially to the ileal pouch.

At that time, endoscopic evaluation of the ileal pouch revealed some small polyps, which biopsy showed to be tubulovillous adenomas with moderate atypia (Fig. 1). A prior biopsy of the elevated mass disclosed a tubulovillous adenoma with severe atypia, and in a subsequent attempt under anesthesia the biopsy displayed a mucinous adenocarcinoma invading the muscular layer (Fig. 2). Abdominal CT scan and carcinoembryonic antigen levels (1.3 ng/ml) were normal.

With the diagnosis of a pouch cancer, the patient underwent pouch excision and definitive terminal ileostomy. Histologic analysis showed a 2.2-cm mucinous adenocarcinoma between the ileal and anal mucosa. Tumoral invasion extended to the muscular layer, and there was distal invasion of the anal canal through the submucosa (Dukes A, Astler-Coller B1, T2N0Mx) (Fig. 3). Resection margins were free of neoplasia, and three resected lymph nodes showed no tumor invasion. The ileal pouch mucosa presented multiple tubular microadenomas with moderate atypia.