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## Cancer in an unexpected site post pouch surgery for familial adenomatous polyposis (FAP)

Omar A. Alwahbi<sup>a,\*</sup>, Alaa S. Abduljabbar<sup>a</sup>, Lucman A. Anwer<sup>b</sup><sup>a</sup> King Faisal Specialist Hospital & Research Center, Riyadh, Saudi Arabia<sup>b</sup> Mayo Clinic, Rochester, MN, United States

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## ABSTRACT

**INTRODUCTION:** Familial Adenomatous Polyposis (FAP) is a hereditary condition characterized by multiple colorectal adenomatous polyps. FAP is the most common adenomatous polyposis syndrome. Restorative proctocolectomy is the most commonly performed surgical procedure performed for patients suffering from FAP with different options for anastomosis, namely ileorectal anastomosis (IRA) or ileal pouch anal anastomosis (IPAA). The occurrence of adenomas is a common finding during follow up and surveillance post surgery for these patients. Although there are a few cases of carcinoma that were namely at the anal transitional zone (ATZ), there are only a few cases of ileal pouch related adenocarcinoma reported. This work has been reported in line with the SCARE criteria (Agha et al., 2016) [1].

**PRESENTATION OF CASE:** We report a case of a 34-year-old man diagnosed with FAP who underwent proctocolectomy with IPAA, and subsequently referred to our center, who, despite appropriate measures and surveillance, developed adenocarcinoma in the ileal pouch.

**DISCUSSION:** Restorative proctocolectomy for Familial Adenomatous Polyposis (FAP) is the mainstay of treatment. There are different surgical options, each with its own set of advantages and disadvantages. The most favored option is proctocolectomy with ileal pouch anal anastomosis (IPAA) due to because it involves resection of the rectum. Despite these interventions, adenomas and/or carcinomas have been reported on follow up post surgery.

**CONCLUSION:** Although the risk of developing adenomas or carcinomas in the ileal pouch post proctocolectomy with IPAA is low it should not be neglected as cancer occurrence or recurrence is unpredictable even with appropriate measures.

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## 1. Background

Familial Adenomatous Polyposis (FAP) is a hereditary condition characterized by multiple (usually over 100) colorectal adenomatous polyps. Familial Adenomatous Polyposis is the most common adenomatous polyposis syndrome. Occurs approximately 1 in 10,000 to 1 in 30,000. It accounts for 1% of all colorectal cancers in the United States (US) [2]. FAP is caused by germline mutations in the tumor suppressor gene Adenomatous Polyposis Coli (APC) and follows an Autosomal Dominant (AD) pattern of inheritance. About 25% of FAP cases arise from de novo APC mutations meaning that these patients do not have a family history of FAP [3–5].

Restorative proctocolectomy is the most commonly performed surgical procedure performed for patients suffering from Familial Adenomatous Polyposis (FAP) with different types of anastomoses

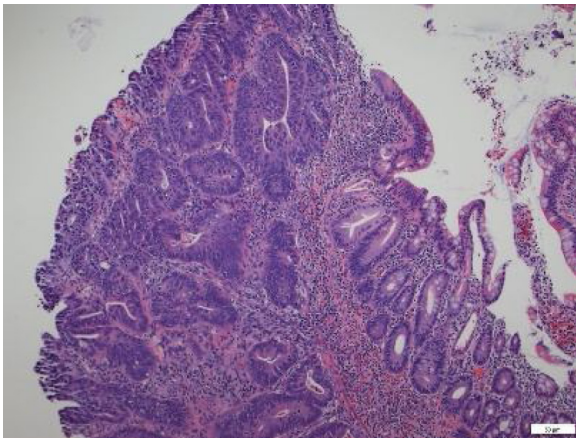
used such as ileorectal anastomosis (IRA) or ileal pouch anal anastomosis (IPAA) [11–13]. Cancer has been reported to reappear despite surgery, namely in the anal transitional zone (ATZ). Ileal-pouch related adenocarcinoma is still a rarity, there are only 22 cases of FAP-related adenocarcinoma reported [14,15]. However, as we all know, cancer is unpredictable and can occur or recur anywhere. We report this case of a 34-year-old man with FAP who underwent proctocolectomy with IPAA at King Faisal Specialist Hospital in Riyadh who, despite surveillance, had a new development in his condition in a short period of time.

## 2. Case presentation

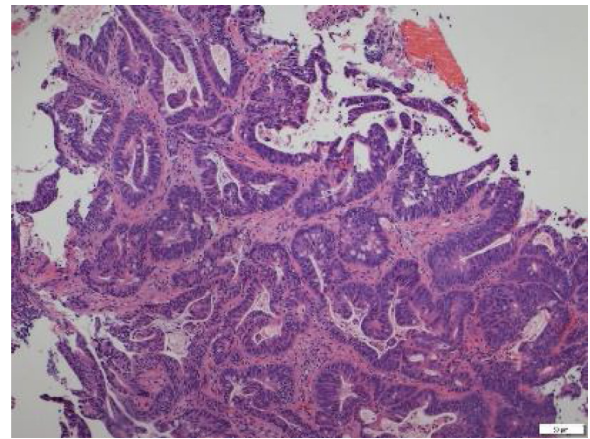
A middle aged male patient known case of Familial Adenomatous Polyposis (FAP) was referred to our center for follow up and genetic counseling. He underwent proctocolectomy with ileal pouch anal anastomosis 15 months prior to referral and closure of ileostomy 5 months prior. Upon his first visit on the April 2012 he was complaining of intermittent mild abdominal pain. His bowel motions were 8–10 motions per day with semisolid stools. No

\* Corresponding author.

E-mail addresses: [o.a.w.7@hotmail.com](mailto:o.a.w.7@hotmail.com) (O.A. Alwahbi),[aabduljabbar@kfshrc.edu.sa](mailto:aabduljabbar@kfshrc.edu.sa) (A.S. Abduljabbar), [Lucman.Anwer@hotmail.com](mailto:Lucman.Anwer@hotmail.com) (L.A. Anwer).



**Fig. 1.** H&E stain of colonic biopsy shows multiple solid features of malignancy including an increase in nuclear:cytoplasmic ratio, glandular crowding, increased mitotic counts in all epithelial layers and a cribriform pattern.



**Fig. 2.** H&E stain of colonic biopsy showing multiple solid features of malignancy including increased nuclear size, glandular crowding, irregular nuclear membrane with an infiltrating pattern along with increased mitotic counts.

rectal bleeding and no vomiting. On examination, he had a mid-line abdominal surgical scar and a stoma scar at the left lower abdomen. No hernias or palpable masses were found. His family history was negative. The digital rectal exam (DRE) revealed a palpable mass and a polyp felt posteriorly, no hemorrhoids. He was subjected to anoscopy but it wasn't seen through because the patient was not tolerating the procedure. He was scheduled for a chest, abdomen and pelvis computed tomography (CT), an esophagogastroduodenoscopy, (EGD) and a sigmoidoscopy and a series of blood tests were ordered including Carcinoembryonic Antigen (CEA) which was 1.1. CT showed several suspicious lesions in the pelvic area; MRI was recommended. The EGD showed a small hiatus hernia, grade A Gastroesophageal Reflux Disease (GERD) and diffuse mild gastritis. Sigmoidoscopy revealed a remnant of 10 cm in size of rectal stump filled with a carpet of polyps, the ileorectal anastomosis was normal at 10 cm from the anal verge and the ileal pouch looked normal with no evidence of pouchitis. On his visit to the Colorectal Clinic on May 2012 he had no complaints, he was sent for genetic counseling where a pedigree was done and showed that none of his six siblings had any Gastrointestinal (GI) problems and that none of his uncles had any complaints. His family was invited in for genetic testing of APC.

Sigmoidoscopy done on December 2012 showed an ulcerating mass proximal to the anastomosis with numerous polyps at the rectal stump with an element of regression compared to the previous exam. He was diagnosed with an ileal pouch tumor. Multiple biopsies were taken (Figs. 1 and 2) which showed adenocarcinoma of the ileal pouch. CT showed infiltration to the right seminal vesicle, pelvic wall and lymph nodes with mesenteric metastasis.

On January 2013 he was seen in the Oncology clinic. A Positron Emission Tomography (PET) scan was ordered which showed low-grade FDG-avid focal nodal lesions in the peritoneum and mesentery indicative of carcinomatosis. A repeat CEA was 8.3 and K-RAS was requested. He was discussed in the tumor board and the plan was to start chemotherapy and if there was an adequate response he was planned to undergo Hyperthermic Intraperitoneal Chemotherapy (HIPEC) but the Oncologist decided to start chemotherapy and then stage his case instead. He was started on Xelox and Avastin chemotherapy for 5 cycles given every 3 weeks starting late January. A CT done in April 2013 showed a 30% decrease in size of the rectal mass with no change in mesenteric nodules. He still had two more chemotherapy cycles left and his CEA is now 1.6.

He completed his chemotherapy cycle on March 2013 and has been scheduled for follow up appointments but was lost to follow up.

### 3. Discussion

Restorative proctocolectomy for Familial Adenomatous Polyposis (FAP) is the mainstay of treatment [6]. The main surgical options for anastomoses include proctocolectomy with ileo-rectal anastomosis (IRA), and proctocolectomy with ileal pouch anal anastomosis (IPAA). The choice of either is not without risks or benefits; with IRA on one hand the major advantage is the preservation of the rectum and better functional outcome and quality of life. On the other hand, the drawback is that it requires much more aggressive surveillance as the cumulative risk of cancer can reach up to 25%. Therefore, rectal involvement of the disease must be ruled out. However, with IPAA there is a smaller chance of developing cancer, the draw-back to this option is a drop in quality and functional outcome compared to the former [11–13]. Traditionally, IPAA included a mucosectomy to eliminate the risk of malignancy. Surgeons have also opted to preserve the anal transitional zone (ATZ) due to its simplicity and better functional outcome.

Despite these interventions, adenomas and/or carcinomas have been reported post surgery [7]. More commonly in the ATZ or with remnant mucosal tissue from the rectum. Adenomas and even carcinomas of the pouch have also been reported. Tajika et al. stated that there had only been 21 reported cases of ileal pouch carcinoma in the literature to date [15]. Also, that the incidence of adenomas in the ileal pouch ranged from 6.7% up to 73.9%. Several reports also support the claim that patients who have undergone prophylactic proctocolectomy remain at risk of developing adenomas and carcinomas in the pouch and that the risk increased with time (i.e. the age of the pouch). The risk appears to be 7–16% after 5 years, 35–42% after 10 years, and around 75% after 15 years. The median age of diagnosis of pouch carcinoma was 10 years post pouch construction [7–10].

It is thought that the pathogenesis of pouch adenomas/carcinomas does not only involve the effects of APC mutation due to primary disease and does not seem to follow the usual adenoma-carcinoma sequence. The mucosa is also subjected to luminal factors due to fecal stasis, which might lead to colonic metaplasia. Several authors have implicated this observation as a precursor for the development of adenomas [14,15].

We conclude from this case report and subsequent discussion that although the risk of developing adenomas or carcinomas in the ileal pouch with proctocolectomy with IPAA is low, it is not negligible [8]. Despite regular follow up and surveillance, malignant transformation can occur. As we've seen from this case, recurrence is often unpredictable and can occur anywhere.

**Conflicts of interest**

None.

**Funding**

None.

**Ethical approval**

Cleared by institution Research Center; ORA clearance (attached).

**Consent**

The head of the medical team/hospital or legal team has taken responsibility that exhaustive attempts have been made to contact the family and that the paper has been sufficiently anonymised not to cause harm to the patient or their family.

**Authors contribution**

Alwahbi, Omar: Writing and editing, data retrieval.  
Abuljabbar, Alaa: Supervisor, reviewed case and paper.  
Anwer, Lucman: Selected case, writing.

**Guarantors**

Alwahbi, Omar.  
Abduljabbar, Alaa.

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