Familial Adenomatous Polyposis (FAP)—A Case Study and Review of Literature

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Abstract
Familial adenomatous polyposis (FAP) is a syndrome characteristically having numerous (hundreds to thousands) polyps in the epithelium of the large intestines with an autosomal dominant inheritance caused by germ line mutations in adenomatous polyposis coli (APC) gene in chromosome 5q21. Most FAP patients have a family history of colorectal polyps and cancer but 25-30% of them are "de novo", without any clinical or genetic evidence of FAP in family members. Prophylactic proctocolectomy is required in almost all patients since all affected patients inevitably develop cancer. We report a case of a 24-year-old girl who presented with recurrent bleeding per rectum, which on evaluation as found to have multiple colorectal polyps and underwent a prophylactic proctocolectomy with end continent ileostomy. In conclusion, patients with FAP may present with vague abdominal complaints and without any family history, hence need to be carefully evaluated. Good patient compliance is of prime importance in deciding the treatment and surveillance modality subsequently determining the prognosis of patients with FAP.

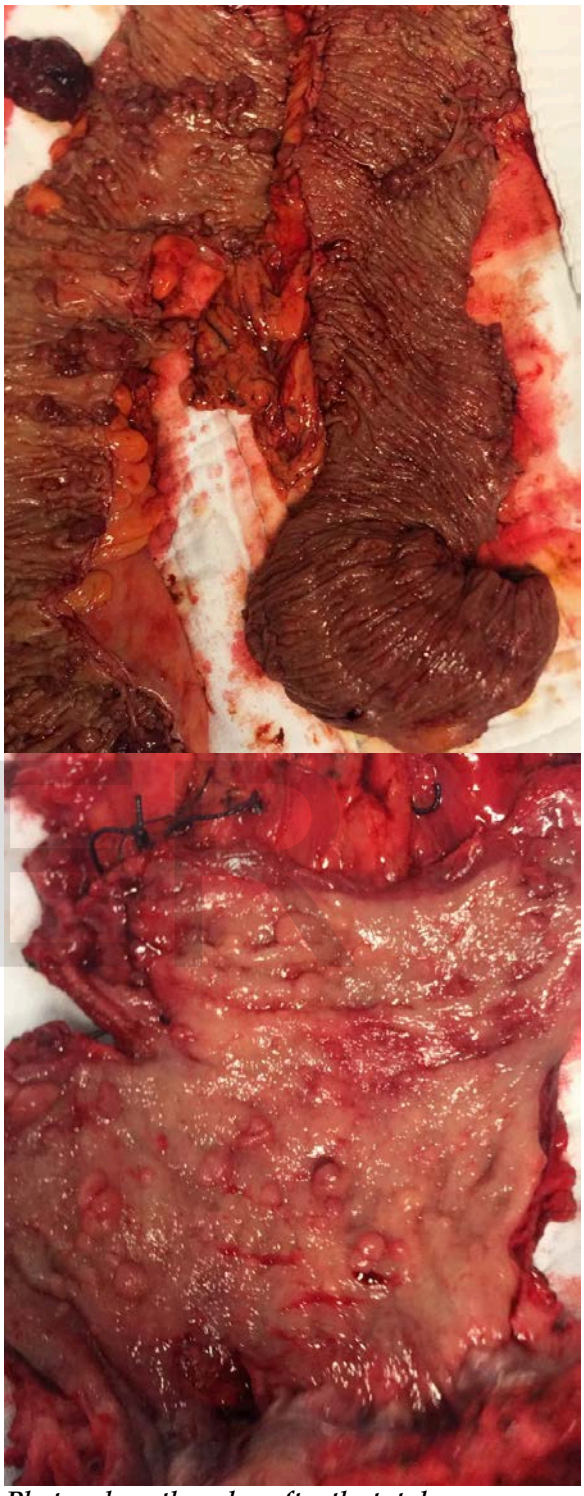
Introduction: FAP is the result of a tumor suppressor mutation of the APC gene (Adenomatous Polyposis Coli, 5q21–q22), which causes multiple (classically over 100) colorectal adenomatous polyps. By 45 years of age, untreated Classic FAP will cause colorectal cancer in appraoximately 100% of cases. While most patients are asymptomatic, themost common clinical presentation of colon cancer and FAP is rectal bleeding, which occurs in 58% and 37% of patients, respectively. The initial clinical presentation in patients with FAP under 20 years old is hematochezia with diarrhea, occurring in 28% and 13% of them, respectively. The reason is that most cases are asymptomatic until the polyps are large enough to cause GI bleeding and anemia.It is recommended to manage FAP with initial genetic screening,followed by yearly colonoscopy from 10 to 40 years old. Once FAP is diagnosed, colectomy or total proctocolectomy is recommended.

Case Presantation: A 24 year old female patient had reported complaints of recurrent bleeding per rectum since a few months. Stool mixed with blood and sometimes only fresh blood was passed. The patient also suffered from general weakness and weight loss. She did not complain of anorexia, vomiting, and abdominal distension; or did not show any signs of jaundice or any lumps. She underwent colonoscopy by which hundreeds sessile and pedunculated polyps were seen in the entire colon up to the cecum. Multiple biopsies were taken and report confirmed the diagnosis of adenomatous polyposis coli. Familial type was considered. Histopathological reports of biopsies revealed villous tubular adenoma with moderate dysplasia. The ultrasound of abdomen was normal and upper gastrointestinal endoscopy results showed helicobacter pylori for which she was treated with anitibiotics for two weeks.
Patient was planned for prophylactic total anoproctocolectomy. Patients consent was obtained for surgical management and permanent ileostomy was opted by the patient and she was subsequently operated and recovered well.

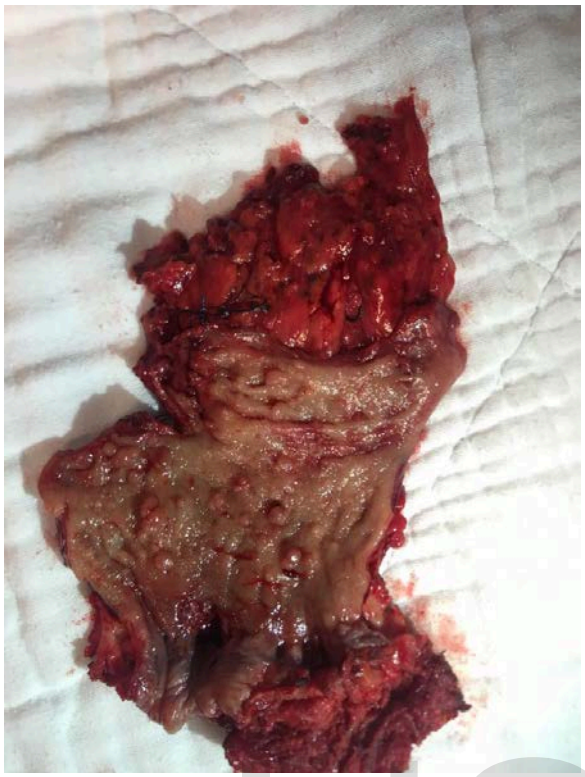
Discussion: FAP is a syndrome characteristically having numerous (hundreds to thousands) polyps in the epithelium of the large intestines with an autosomal dominant inheritance caused by germ line mutations in APC gene in chromosome 5q21. Its prevalence being 3–10/100,000, affecting both sexes equally, with development of symptoms by the late teens and in the twenties. Most FAP

patients have a family history of colorectal polyps and cancer but 25-30% of them are "de novo", without any clinical or genetic evidence of FAP in family members. Patient's family history was insignificant. Majority of the patients develop polyps in the childhood, which gradually increase in size and number throughout the colon until adolescence. Nearly fifty percent of patients develop adenomas by the age of 15 years and 95% by the age of 35 years. Malignancy begins to develop approximately 10 years after the appearance of polyps. Other common clinical features in patients with FAP include multiple gastric fundic gland polyps, duodenal, periampullar or ampullar adenomas, while extra intestinal features are desmoid tumours, congenital hypertrophy of the retinal pigment epithelium (CHRPE), epidermoid cysts, osteomas and thyroid cancer.

Prophylactic surgery is recommended before the age of 25 years. The main surgical options for patients with FAP are (1) total proctocolectomy with terminal ileostomy; (2) subtotal colectomy with ileorectal anastomosis, and (3) restorative proctocolectomy with the formation of an ileal reservoir and ileoanal anastomosis. The decision to remove the rectum depend upon the number of rectal polyps and the family history. For few polyps in the rectum, total colectomy with ileorectal anastomosis is recommended. In rectal involvement, a proctocolectomy with ileal pouch-anal anastomosis is the treatment of choice. If the anus is involved too the golden choice of treatment is proctocolectomy with terminal ileostomy. Prophylactic surgery improves the outcome of patients with FAP significantly and it is recommended in late teens or early twenties. The risk of developing rectal adenomas and carcinomas after ileorectal anastomosis is approximately 13 to 59%, after 25 years mandating a lifelong rectal surveillance.



Photos show the colon after the total proctocolectomy



Conclusion: Patients with FAP may present with vague abdominal complaints and without any family history, hence need to be carefully evaluated. Good patient compliance is of prime importance in deciding the treatment and surveillance modality subsequently determining the prognosis of patients with FAP.

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