Case Report

Development of rectal cancer in a young patient with a retained rectum after total colectomy for Familial Adenomatous Polyposis.

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Introduction

Familial Adenomatous polyposis (FAP) is an inherited disorder and it represents the most common gastro intestinal Polyposis syndrome. The mutation of germ line in the APC gene is the main cause for FAP. The FAP is an inherited syndrome characterized by multiple (more than hundred) polyps in the colorectum with high risk of colorectal carcinoma and existence of extra colonic manifestations. Cancer prevention and maintaining a good quality of life are the main goals of management. The recommended surgical modalities are total proctocolectomy and ileoanal pouch or ileorectal anastomosis for AFAP.

Case Presentation

A 28 yr old married, otherwise healthy women presented with a history of altered bowel habit for 10 months duration. She gave a history of passing stools 10 to 15 times daily, and it was mixed with altered blood with bad odor. Her appetite was good but she had lost about 10kg of weight during this period. She denied history of fever, abdominal pain or any other systemic symptoms. Her father underwent an abdominal surgery and died 20 years back and no records available for further evaluation.

Her abdominal examination and digital rectal examination were found to be normal. No any peripheral stigmata of any chronic illness were present and her BMI was 17. Her haemotological investigations revealed an Hb of 7.4mg/dl, ESR – 74mm/hr. CEA was 1.49ng/ml. More than 100 polyps seen in colonoscopy and the biopsy showed two tubular adenomas and one villous adenoma. Ultrasound scan of abdomen and pelvis revealed a significant wall thickening of colon with multiple mesenteric lymphadenopathy. Double contrast barium enema revealed multiple polyps in ascending and transverse colon. The diagnosis was made as Adenomatous polyposis coli.

She initially underwent total colectomy (Stage I procedure) and ileostomy. The biopsy revealed a large tubulo – villous adenoma with high grade dysplasia and numourous (>100) adenomatous polyps with low grade dysplasia throughout the colon including the distal resection margin. Three months later she was admitted for ileal pouch creation. The procedure was converted to abdomino perineal resection since multiple polyps involving upto anus noted intra operatively. This biopsy revealed a well differentiated adenocarcinoma (Duke C1, pT3N1bMx)

Discussion

The most common adenomatous polyposis syndrome is FAP. Its an autosomal dominant inherited disorder characterized by more than 100 – 1000 of polyps throughout the colon . FAP most commonly affect the left side colon (4) . All patients with FAP develop colorectal cancer by the age of 35 – 40 yrs if untreated (5). Rectal carcinoma developed in 59 % of FAP patients as in this patient (4,6). Familial adenomatous ployposis (FAP) is a rare condition that occurs 1 in 6850 to 1 in 30000 new born and also responsible for approximately 1% of colorectal carcinoma in the world (5,6). The mutation of the APC gene is one of the earliest event leading to polyp formation and subsequently lead to malignant formation(1,3,5). This is known as adenoma carcinoma sequence. There are more than six hundred mutations have been discovered in APC gene in these patients (5).

The APC gene mutation has a high penetrance rate and in untreated or undetected patients the natural history of the disease is as follows (2,4,5).

- Age of appearance of adenomas is 20 – 23 yrs
- Age of onset of symptoms 30 – 35 yrs
- Age of diagnosis of adenomas 30 – 37 yrs
- Age of diagnosis of carcinomas 30 -40 yrs

Among those who were diagnosed with symptoms of FAP, 65% already had a colorectal malignant lesion (4,5). Our patient unusually had a malignant lesion in the rectum at the age of 28.

Usually, by the late teens or early twenties, due to the increasing number of adenomas, prophylactic cancer preventive surgery is advocated(2). Surgical options include subtotal colectomy with ileorectal anastomosis, total proctocolectomy with ileostomy, and proctocolectomy with or without mucosectomy and ileal pouch anal anastomosis. Given the substantial risk of rectal cancer developing after colectomy and ileorectal anastomosis, most experts advise total proctocolectomy for the typical FAP patient with multiple rectal adenomas(2).

The absolute indications for immediate colorectal surgery in FAP include documented or suspected cancer with significant symptoms(3). Considering this patient's age and investigation findings, she underwent total colectomy and planned for ileal pouch creation later. However, during the second surgery she was found to have multiple polyps involving rectum and the surgery was converted to abdomino perineal resection.

The individuals with family history of FAP or subtypes, FAP – type with extra colonic manifestations, colonoscopy and all detection of more than 10 cumulative colorectal adenomas or patients with colorectal carcinomas should undergo for genetic assessment(1,3). Genetic testing of these patients include APC and MUTYH gene mutation analysis (1,3).. The surgery option is indicated in all proved patients with FAP (1).

Conclusion

Cancer prevention and maintaining a good quality of life are the main goals of management in FAP. Early detection, genetic counseling and decision making after providing adequate information to the patient improve the prognostic index after surgery in patients with FAP. In the management of patients with FAP, it should be born in mind that there is a possibility of malignant transformation even in younger patients and development of rectal cancer in patients with a retained rectum after total colectomy.

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