

Familial adenomatous polyposis coli: Report of a rare entity

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ABSTRACT

We report a sporadic case of a familial adenomatous polyposis coli (FAP) in a 25-year-old male who came with no family history of the same. FAP is the most common inherited adenomatous polyposis syndrome. Because of its rarity, the incidence in a developing country like ours is unknown. It is characterized by more than 100 adenomatous polyps and if left untreated carries a 100% risk of progression to colorectal cancer by the fourth decade. The early recognition and characterization of the polyposis syndrome is vital since early intervention and surgery will help in prevention against the development of invasive colorectal cancer.

Key words: Adenomatous polyp, colorectal cancer, familial, screening

INTRODUCTION

Colorectal carcinoma (CRC) is the second most common cancer in the industrialized world.^[1] About 1% of all cases of CRCs are caused by inherited gastrointestinal polyposis (IGP) syndromes. IGP syndromes include adenomatous polyposis syndromes and hamartomatous polyposis syndromes. Classic familial adenomatous polyposis (FAP) with its variants like Gardner's syndrome, Turcot's syndrome, attenuated adenomatous polyposis coli, and MUTYH-associated polyposis (MAP) account for the majority of the adenomatous polyposis syndromes.^[2] FAP is the most common inherited adenomatous polyposis syndrome. In the United States, the incidence is about 1:13,000 births.^[3] Because of its rarity, its incidence in a developing country like ours is unknown. FAP is characterized by more than 100 adenomatous polyps and if left untreated, carries a nearly 100% risk of progression to CRC by the age of 35-40 years along with increased risk of other malignancies.^[3]

CASE REPORT

A 25-year-old male presented with complaints of intermittent bleeding per rectum over a period of 4-5 months. Patient gave a history of similar episodes in the past and had taken oral medications for the same from a local general physician. Colonoscopic examination showed numerous (more than 100) sessile and pedunculated polyps present over the entire colonic mucosa. Biopsy taken from one of the polyp revealed features of adenomatous polyp. Further investigations of the family members revealed none with polyposis syndrome. Total proctocolectomy with ileoanal anastomosis was performed. We received two segments of intestine. The longer segment of colon and rectum measured 60 cm in length and a short segment of caecum measured 20 cm in length. On examining the mucosal aspect of both the segments received, revealed about 163 sessile as well as pedunculated polyps ranging from 2 mm to 3 cm in size [Figure 1a and b]. Microscopic examination of 15 polyps which included five pedunculated and ten sessile polyps was carried out. Three of the pedunculated polyps showed glandular epithelium arranged in a tubulovillous pattern lined by stratified columnar epithelium with moderate nuclear atypia overlying a thin fibrovascular core, characteristic of a tubulovillous adenoma [Figure 2]. Two of the pedunculated polyps additionally displayed focal area showing glands arranged in a back to back pattern and lined by columnar cells with moderate to severe nuclear atypia indicating a transformation into *in situ* colorectal carcinoma [Figure 3]. The remaining ten sessile polyps showed features of villous adenomas.

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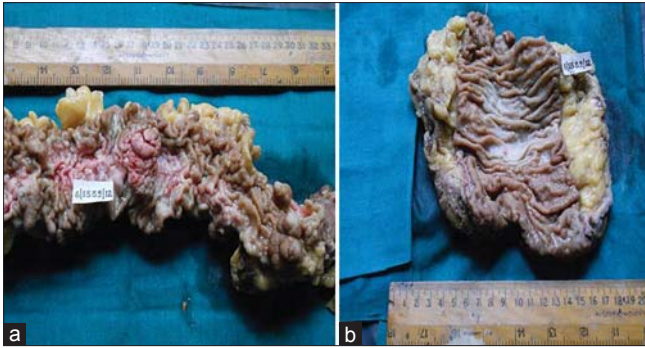


Figure 1: (a and b) Gross photograph showing entire mucosa of colon and rectum along with caecum studded with 163 sessile as well as pedunculated polyps ranging from 2 mm to 3 cm in size

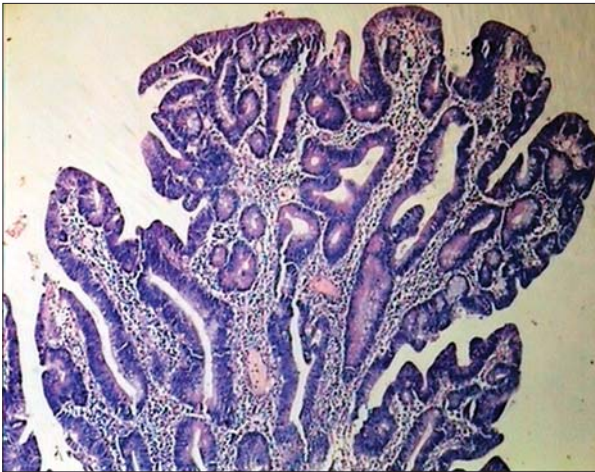


Figure 2: Photomicrograph showing adenomatous polyp (H and E, $\times 40$)

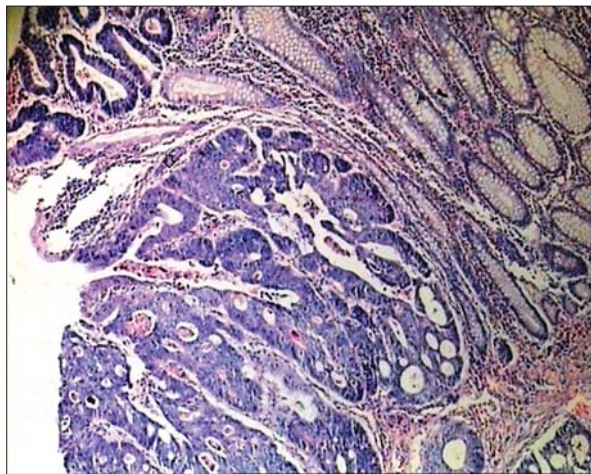


Figure 3: Photomicrograph showing adenomatous polyp with carcinoma *in situ* (arrow head). (H and E, $\times 40$)

Taking into consideration that the total number of adenomas amounted to more than hundred, a diagnosis of familial adenomatous polyposis with some polyps showing *in situ* colorectal carcinoma was given with an advice for screening of the first degree family members.

DISCUSSION

The diagnostic criteria for FAP are (1) 100 or more colorectal adenomatous polyps, or (2) germ line mutation in APC gene, or (3) any colorectal adenomas under age of 30 in a patient with a family history of FAP or (4) intra-abdominal desmoid fibromatosis, osteoma of the mandible, or multiple epidermoid cysts in a patient with a family history of FAP.^[4] The extra intestinal manifestations of FAP include: Cutaneous lesions (fibromas, lipomas, sebaceous and epidermoid cysts, and nasopharyngeal angiofibromas), osteomas, congenital hypertrophy of the retinal pigment epithelium (CHRPE), soft tissue tumor (desmoid tumors) in the mesentery, abdominal wall or areas of scars, pancreatic mucinous adenocarcinoma, hepatoblastoma, brain tumors like glioblastoma multiforme or medulloblastoma, and thyroid cancer.^[5]

FAP is inherited as an autosomal dominant disease with 80-100% penetrance. The FAP gene is located in the 5q21-q22 region and the gene product is termed as APC for adenomatous polyposis coli and is a multifaceted regulator of colonic epithelial homeostasis.^[6,7] The patient in our case presented with more than 100 adenomatous polyps with no family history of FAP. Nearly 30% cases of FAP have no family history of FAP, which is speculated to be caused by a new germ-line mutation. These patients are treated with any of the following three main surgical options available: Total proctocolectomy with permanent ileostomy; subtotal colectomy with ileorectal anastomosis; and proctocolectomy with ileoanal anastomosis.^[8]

The patient suffering from FAP including its first degree relatives carries a high life time risk of gastrointestinal and extraintestinal malignancy. Early recognition and correct characterization of the polyposis syndrome is vital since effective methods of surveillance in the form of endoscopic screening and treatment modalities are available.^[9] We report this case because of its rarity in our region and also to highlight the importance of screening of close family members for early diagnosis and prevention of invasive colorectal cancer.

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