Case Report

TOTAL PROCTOCOLECTOMY WITH ILEOANAL ENDORECTAL PULLTHROUGH IN FAMILIAL ADENOMATOUS POLYPOSIS

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Abstract

Familial adenomatous polyposis (FAP) is an uncommon condition for rectal bleeding, presents in older children usually after the 1st decade of life. The polyps are adenomatous in nature and are potentially malignant. A 30 year old married female presented with complaints of increased frequency of loose stool, lower abdominal pain, rectal bleeding, anaemia and gradual weight loss for last 5 years. Double contrast Ba enema and colonoscopic biopsy confirmed the diagnosis. She underwent total proctocolectomy with ileoanal endorectal pull through without pouch and stoma in 'CMH' Dhaka. On subsequent follow-ups she gained weight and is free of symptoms.

Introduction

Familial adenomatous polyposis (FAP) is an autosomal dominant form of intestinal polyposis and colorectal cancer caused by germ line mutations in the adenomatous polyposis coli (APC) gene[1]. Colorectal cancer is a multifactorial disease and the aetiology is complex, It involves dietary and other environmental factors and in between 15 to 30% of cases, inherited genetic factors are significant[2]. Carriers of the mutated APC gene develop hundreds to thousands of adenomatous polyps in the colon and rectum; stomach duodenum and small intestine can also be affected [3,4]. Since the abnormal gene that causes FAP is present in all the body's cells, other organs may also develop growths[5]. The other organs that form tumours include skin, bones, eye (congenital hypertrophy of the retinal pigment epithelium CHRPE), thyroid, abdomen (desmoid tumours), brain and adrenal gland[6,7]. Treatment of FAP depends on the genotype. Most individuals with the APC mutant will develop colon cancer by the age of 40. Therefore prophylactic surgery is generally recommended before the age of 25, if there are severely dysplastic polyps, or if multiple polyps larger than 1 cm are present[6].

Case Report

A 30 year old married female was admitted in CMH Dhaka on 19 Jun 2010 with complaints of increased frequency of loose stool, lower abdominal pain, rectal bleeding, anaemia and gradual weight loss for last 5 years. There was no definite history of similar complaints in parents or grand parents but her elder sister had similar complaints and which later on was diagnosed to be a case of FAP. Her sister also developed Ca breast prior to the diagnosis of FAP. She had two kids born by normal vaginal delivery. She was treated locally and in peripheral CMH with repeated doses of different anti diarrhoeal drugs but she had little if any relief.

She was anxious, emaciated with ill health. Her weight was 40 kg only. Thorough evaluation of the patient was done. Digital rectal examination revealed multiple polyps. Double contrast Ba-enema was suggestive of polyposis coli. Colonoscopic biopsy confirmed the diagnosis of FAP. Her CEA (carcino embryonic antigen), upper 'GI' endoscopy was normal. Different options of treatment were available and their sequelae were discussed with the patient and her husband.

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Finally considering her age, sex, symptoms total proctocolectomy with ileoanal endorectal pull through was planned. This operation was never done earlier in CMH Dhaka.

By supplementary diet and medications her health condition got improved. After proper gut preparation total proctocolectomy with ileoanal endorectal pull through without pouch and stoma was done on 25 Jul 2010. In the initial few days she had 16-20 motions per day requiring enteral and parenteral supplementary support. Gradually stool got thickened and the number of motion reduced. She was discharged on 22 Aug 2010. She was reviewed after 3 months and at that time she had 5-6 motions per day. By 6 months number of motion reduced to 3-4 motions per day. Now she is coming for follow up in every 6 months.

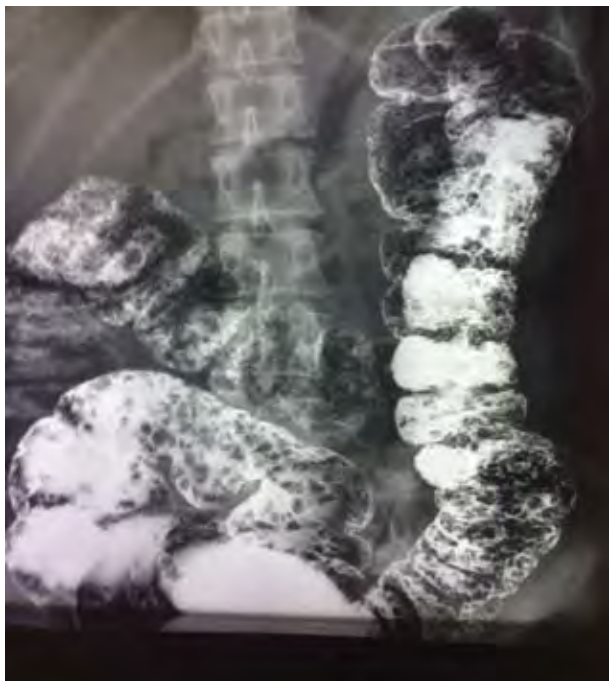

Fig-1: Double contrast Ba enema showing numerous polyps

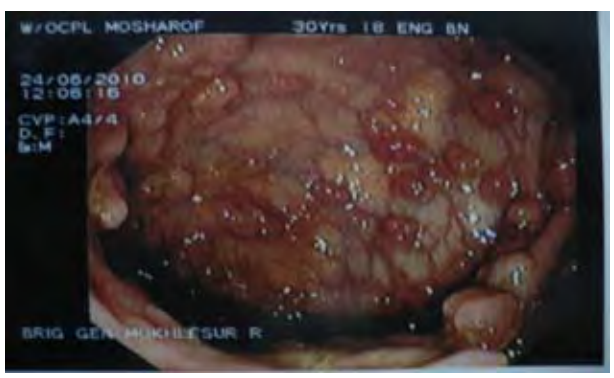

Fig-2: Colonoscopic view of polyps

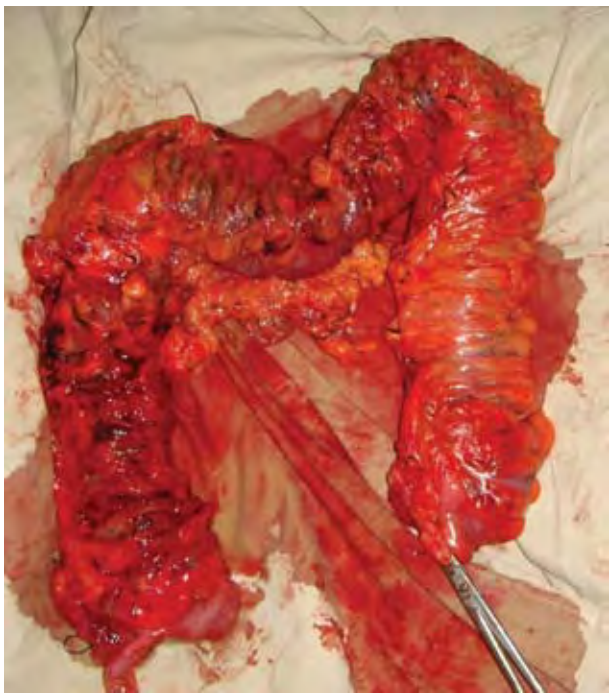

Fig-3: Resected specimen showing whole colon

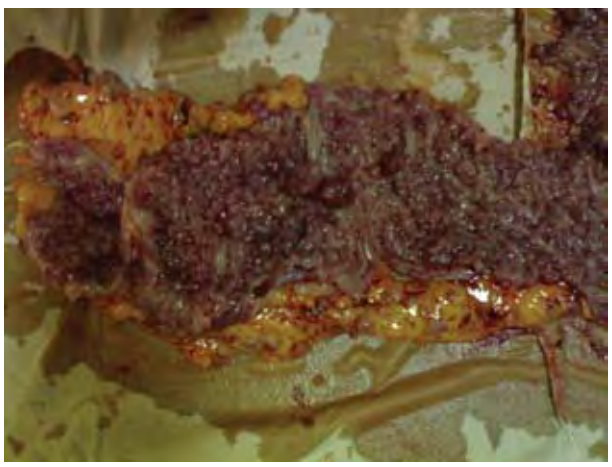

Fig-4: Resected specimen showing numerous polyps

Discussion

FAP is defined as the presence of 100 or more visible adenomatous polyps in the large intestine and rectum [8,9]. Two types of FAP seem to exist: the sparse, which is characterized by hundreds of polyps and the profuse type, which is characterized by thousands of polyps. The majority (60-70%) of patients with FAP have inherited the gene from one of their parents. Therefore, consecutive generations may have FAP.In about 30% of FAP patients the abnormal gene was produced at the time of conception. Males and females are equally affected[10]. Once the risk is appreciated, screening for the disease must take place[11].

All members of the family should be examined at the age of 10-12 years, repeated every 1-2 years. If no polyps are detected, yearly colonoscopy should be continued until approximate age of 35 years[5]. If diagnosis is made during adolescence, operation is deferred usually to the age of 17 or 18yrs. Both Gardner's syndrome (familial colonic polyps, osteoma, fibroma and sebaceous cysts) and FAP are due to inherited mutant APC gene. Attenuate FAP is a variant of FAP where there are fewer polyps in the colon[5].

The incidence of FAP ranges from 1 in 6000 to 1 in 12000 births. The median age for development of polyp is 16 years, for bowel symptoms 29 years and for colorectal cancer 40 years[12]. Colonoscopy is considered as the diagnostic test of choice as it can provide not only a quantification of polyps through out the colon but also for a histological diagnosis. Ba-enema and virtual colonoscopy can suggest the diagnosis of FAP[6]. Genetic testing provides the ultimate diagnosis in 95% of cases. The most common of which is Direct DNA sequencing. Other methods which can be used as second-line tests are monoallelic mutation analysis (MAMA), conversion analysis and multiplex ligation-dependent probe amplification (MLPA)[7].

Because of the diffuse nature of the polyposis and the inevitability of colorectal cancer surgical therapy is ultimately required[13]. Surgical options are total proctocolectomy with a permanent ileostomy, total colectomy with ileorectal anastomosis, total proctocolectomy with ileoanal anastomosis with pouch, total colectomy with mucosal proctectomy and ileoanal endorectal pull through [5,6,12,14]. The choice of treatment depends on many factors and should be individualized. The most important determinants are rectal polyp burden, presence of cancer or severe dysplasia, age, symptoms, continence, genotype and patient compliance[7].

While surgery is most common for treating the colon polyps, it does not cure the disease. Polyps continue to form in the pouch, rectum or small intestine. A variety of medications have been developed to cause regression (shrink of existing polyps) or prevention of recurrent polyps. While these medications help control the colorectal polyps, they should only be used in conjunction with ongoing checkups and will not replace the usual care of patients with FAP[5,6]. Patients with FAP or family members at risk of FAP should have regular checkups of the colon, rectum or pouch and the upper gastrointestinal tract. Upper gastrointestinal polyps occur in up to 80 percent patients with FAP[5,6]. Carcinoma in the duodenum and periampullary area is the most common cause of death after colorectal cancer in FAP patients [8,12]. Duodenal surveillance should begin before age 30 and the best yield will be obtained using a side viewing endoscope[7]. Removal of polyps in the duodenum can be done by endoscopic snaring or by open duodenotomy[8]. If polyps are found in the remaining rectum or pouch they are burnt out before turning cancerous[5].

In our case, after detail discussion with the patient and her husband and considering all parameters of the patient we performed proctocolectomy with ileal endorectal pull through. Only the elder sister of the patient had complaints suggesting FAP. Later she was diagnosed to have FAP and total proctocolectomy with permanent ileostomy was done in another hospital.

Conclusion

The diagnosis of FAP should be considered while dealing with patients of chronic dysentery. High level of suspicion is required to diagnose a patient of FAP especially when family history is negative. Early diagnosis, timely treatment and lifelong follow up are required to prevent inevitable malignancy in colon and elsewhere. The patient and the whole family have to be kept under surveillance as per screening protocol.

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