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Case report

A RARE CASE OF FAMILIAL ADENOMATOUS POLYPOSIS

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ABSTRACT

FAP is an autosomal dominant disease characterized by numerous polyps, numbering from hundreds to thousands, in intestine. It is a very rare condition with incidence ranging from 1 in 6000 to 1 in 12000 births. In absence of surgical intervention, their malignant transformation is inevitable. We report this case because of its rarity.

Keywords: Familial adenomatous polyposis, Colonoscopy, Adenomatous polyposis coli gene, Ileal pouch-anal anastomosis

INTRODUCTION

Colorectal carcinoma is one of the leading cause of deaths due to malignancy globally. FAP is a type of inherited colorectal carcinoma which is autosomal dominant and characteried by numerous polyps in the epithelium of intestine. These polyps are benign in beginning and their malignant transformation is inevitable around the age of 34-43 years, if no surgical intervention is done.[1,2]

CASE REPORT

A 24 years old male presented with complaints of tenesmus and passing watery stools since 2 yrs and passing blood with stools since 4 months. There was no significant family history .His vitals were normal and on per-rectal examination, multiple polyps were palpable around 2cms from anal verge. On proctoscopy there was mucoid discharge with multiple polyps all around. His routine blood investigations were normal and USG abdomen also did not reveal any abnormality. On Colonoscopy there were multiple polyps in rectum, 1-4cms in size, which were both pedunculated and sessile. There were more than 100 polyps throughout the colon till caecum (Fig.1). Their density decreased proximally. The biopsy report was suggestive of adenomatous polyps. A diagnosis of FAP was kept and decision to perform exploratory laparotomy with total proctocolectomy with ileal pouch – anal anastmosis was planned**.** Surgery was performed by midline incision, specimen was dissected from terminal ileum up to anal canal, around 2cms from anal verge .Colon was studded with polyps throughout its length (figure 2,3). Ileal “J pouch” was created (figure 3) and anastmosed with anal canal using circular staplers. A diverting loop ileostomy was done

before closure. The histopathologic findings were consistent with multiple adenomatous polyps with mild to moderate dysplasia (figure4). Patient was regularly followed up and 6 months later ileostomy closure was planned. A distal loopogram was done which was within normal limits. Anal continence was checked and ileostomy closure was done. The immediate family members of patient could not be screened as they lived in a different city. Patient has been followed up to date and no complications have been detected so far.

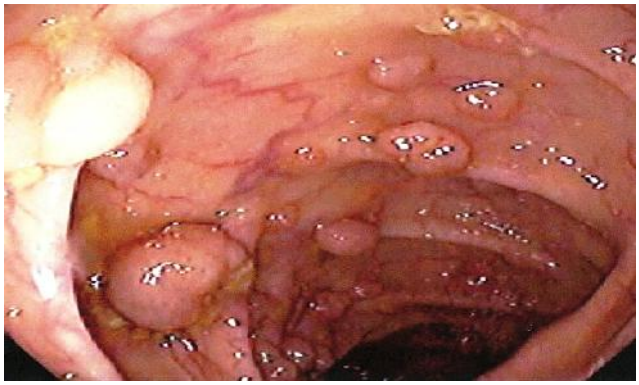

Fig 1: Colonoscopic image showing numerous polyps throughout colon

Fig 2: Photograph of colon after resection

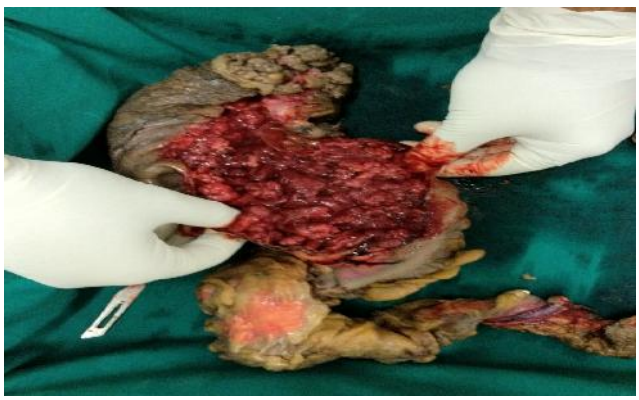

Fig 3: The opened section of specimen showing numerous sessile and pedunculated polyps

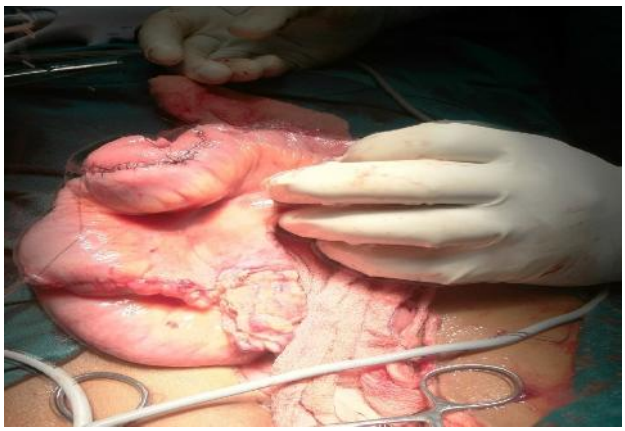

Fig 4: Creating ileal "j-pouch"

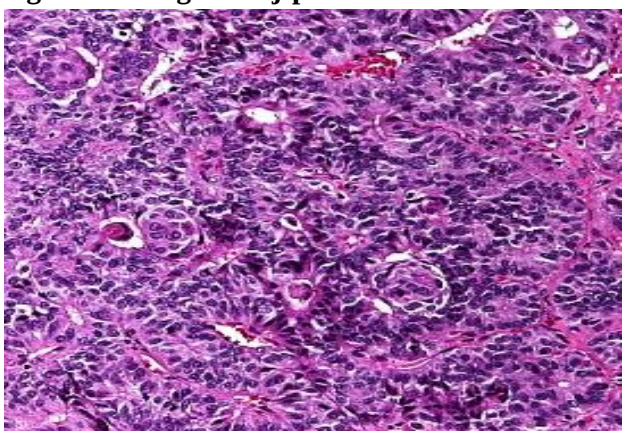

Fig 5: Histological appearance of specimen (magnification x200)

DISCUSSION

FAP is one of the familial causes for colorectal carcinoma. Sklifasowski published the first verified case of FAP in 1881 in Russia. Over the years, with discovery of APC gene FAP has become a separate entity.[3] FAP results from mutation in the APC (adenomatous polyposis gene) gene. *APC* is a tumor suppressor gene located on the long arm of chromosome 5 in band q21.[4]

Symptoms may not present until the adenomas are large and numerous so as to cause intestinal bleeding. Patient may complain of change in bowel habits, constipation/diarrhea, abdominal pain or weight loss.[5]

Extraintestinal manifestations may include Osteomas, which can be identified as occult radio-opaque jaw lesions, Congenital hypertrophy of the retinal pigment epithelium and cutaneous lesions such as fibromas, lipomas,

sebaceous cysts, epidermoid cysts and nasopharyngeal angiofibromas.[3] Association of gastro-intestinal polyps with other extra intestinal manifestations is known as "Gardner's syndrome". [6]

Surveillance examination in patients with family history or suspected cases, is recommended beginning at age 10-12 years .Sigmoidoscopy is recommended every year. Once colon polyps are found, or by age 20-25 years, colonoscopy should be done. Multiple colonic polyps, usually more than 100 are diagnostic for FAP. Genetic testing is useful in confirming a diagnosis of FAP in those cases where there is some doubt about the diagnosis. [7]

The aim of the surgical treatment of FAP is to remove the polyps before the transformation to malignancy occurs. The surgical options are –

1. Colectomy and ileo-rectal anastomosis(IRA)
2. Proctocolectomy with ileal pouch-anal anastomosis (IPAA)
3. Proctocolectomy and ileostomy

Choice of surgery depends upon extent of rectal involvement. IRA is a simple operation with quick recovery, low complication rates and minimal lifestyle interference; however the risk of cancer in rectum mandates yearly surveillance. IPAA minimizes this risk but adenomas develop in the ileal pouch so surveillance is still necessary. A hand sewn IPAA has more complications and poorer function than a stapled IPAA. Laparoscopic surgery can be attempted in cases which are detected early and with less extensive intestinal involvement.[8]

Medical management of FAP is also under research. Sulindac, a non-steroidal anti-inflammatory drug, has been shown in studies to achieve prolonged remission of polyps. However it is still under investigation and not a routine treatment replacing surgery. Aptosyn, a derivative of sulindac and Celecoxib, a COX-2 specific antagonist have also been tried but are not as effective as Sulindac.[9]

Genetic counseling should be offered to family members of a diagnosed patient.

REFERENCES

1. Boia ES, Mejdi R. Familial adenomatous polyposis (FAP); What must be known and what should be done-case report. Pediatr J 2008;11:46-9.
2. Srinivasamurthy M, Geethamala K, Kumar BD, Sudhrao M . Familial adenomatous polyposis coli and adenocarcinoma of the colon: A silent synchronous presentation. Arch Int Surg 2012;2:101-4.
3. Steffen Bulow, Terri Berk, Kay Neale. The history of Familial Adenomatous Polyposis. Familial Cancer 2006;5:213-220.
4. Groden J, Thliveris A, Samowitz W, Carlson M, Gelbert L, Albertsen H, Joslyn G, Stevens J, Spirio L, Robertson M: Identification and characterization of the familial adenomatous polyposis coli gene. Cell 1991, 66**:**589-600.
5. Elizabeth Half**,** Dani Bercovich**,** Paul Rozen; Review of Familial adenomatous polyposis. Orphanet Journal of Rare Diseases**.** 2009, 4:22
6. Polymnia Galiatsatos, William D. Foulkes; Familial Adenomatous Polyposis, American Journal of Gastroenterology. 2006;101:385–98
7. Familial Adenomatous Polyposis (FAP) 2002, 2008 The University of Texas MD Anderson Cancer Center 10/01/08
8. Ambroze WL, Dozois RR, Pemberton JH. Familial adenomatous polyposis: results following ileal pouch-anal anastomosis and ileorectostomy. Dis Colon Rectum.1992;35:12-15
9. Giardello FM, Hamilton SR, Krush AJ. Treatment of colonic and rectal adenomas with sulindac in familial adenomatous polyposis. N Engl J Med. 1993;328:1313-16.