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Familial Adenomatous Polyposis- Presenting with Partial Bowel Obstruction

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Abstract: *Familial adenomatous polyposis ia an autosomal dominant disorder that is responsible for 0.5% of all colorectal cancer. Patients will develop hundreds to thousands of colonic polyp in their second decades of life. The life time risk of developing colorectal cancer is 100% at the median age of 39 years. Extra intestinal manifestations are rare and mostly are benign. Family history of colorectal cancer is common in FAP. Screening of FAP family members should begin with age of 10-12 years. Management option includes total proctocolectomy with end ileostomy, total colectomy with ileorectal anastamosis, and total proctocolectomy and ileal pouch anal anastamosis.Here is a report of rare case of familial adenomatous polyposis presented with intermittent partial bowel obstruction and managed with total proctocolectomy with end ileostomy and reviewed the literature and discussed tumorogenesis , screening recommendation and management options*.
Keywords*: Familial adenomatous polyposis , colorectal cancer*

I. Introduction

Familial adenomatous polyposis is responsible for less than 1% of all colorectal cancer incidence .Hallmark of familial adenomatous polyposis includes hundreds to thousands of colonic polyps that develop in patients with 20 to 30 years of lifes, and the risk of developing CRC approaches 100% in their fifties . Extracolonic manifestations of FAP includes congenital hypertrophy of the retinal pigment epithelium, mandibular osteomas, supernumerary teeth, mandibular osteomas , adrenal cortical adenomas ,epidermal cysts, adrenal cortical adenomas, desmoid tumors and malignant conditions includes thyroid tumors, gastric and small intestinal polyps with a risk of duodenal or ampullary adenocarcinoma at 5 to 10%, and brain tumors.Brain tumors will present in two types—glioblastoma multiforme or medulloblastoma—and the association of brain tumors and colonic polyposis is called Turcot syndrome.In Turcot syndrome, colonic polyps are fewer and larger than in classical FAP. An attenuated form of FAP harbors up to 100 colonic polyps and will lead on to colorectal cancer in patients in their 50s or 60s[2]

II. Case Report

A 46 year old male was presented to surgical oncology department , Government Royapettah Hospital, Chennai with a complaints of intermittent lower abdominal pain and distension of abdomen for 15 days duration , relieved by conservative management . History of loose stool was present on and off. No history of bladder or chest symptoms. No history suggestive of neurologic involvement. Patient was an alcoholic and smoker .
Family history suggestive of colonic cancer was present . Both father and brother died of colonic cancer at the age of 50 years.

On clinical examination, performance status 1, no palpable supraclavicular or inguinal lymph nodes. General examination revealed multiple osteoma in frontal bone. Ophthalmologic examination found congenital hypertrophy of retinal pigment epithelium. Abdomen was not distended. There was no ascitis. A mass of 5*5 cm was palpable in umbilical region and was firm and mobile . On rectal examination , friable polypoidal growth was palpable 4 cm from anal verge . multiple rectal polyps of varying size were palpable . Contrast CT scan of abdomen and pelvis revealed multiple polyps in rectum and entire colon. Liver was normal. Features of intestinal obstruction was present. CECT chest showed no pulmonary metastasis. Tumor marker CEA was-5.49 ng/ml. Colonoscopy showed multiple polyps in rectum, sigmoid colon and descending colon and colonoscope was not passed beyond because of polyps. Biopsy showed adenomatous polyp. Patient was prepared for surgery after explaining the procedure of total procto colectomy and permanent end ileostomy .

Under general anesthesia, in lithotomy position,midline laparotomy was done. There was a palpable growth in rectosigmoid region and was adherent to caecum , and multiple polyps were palpable in entire colon. Small bowel , liver and retroperitoneum were normal on palpation.

Entire colon was mobilised by incising white line of toldt .Testicular vessels and ureters were identified and preserved. Right colic, middle colic, left colic, sigmoidal vessels were ligated and divided. Multiple

lymphnodes in large bowel mesentry was removed along with greater omentum. Total mesorectal excision done . Perineal dissection was completed. Distal ileum was brought outside as end ileostomy in right iliac fossa.

Cut section of specimen showed multiple polyps starting from rectum through caecum with relative sparing of transverse colon. There was an Ulcero infiltrative lesion at recto sigmoid region with transmural involvement present .

Post operative period was uneventful. Ileostomy was functioning well. No electrolyte disturbance occurred post operatively.

Post operative histopathology- pT3N2b. 22 out of 55 lymph node showed metastatic tumor deposits. Patient was treated with adjuvant chemotherapy -folfox4 regimen.

His son 18 years old was asymptomatic and screening colonoscopy showed normal colorectal mucosa.

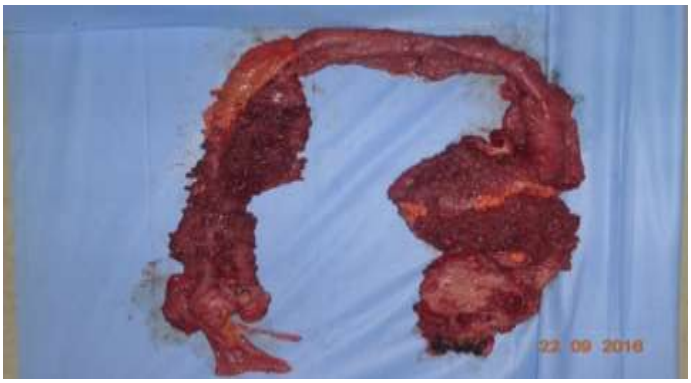

Fig.1 shows total proctocolectomy with multiple polyps in colon and rectum.

Review of literature :

Familial Adenomatous Polyposis : Of all of the colonic polyposis conditions, FAP is the most common and best characterized. FAP is caused by germ-line mutations in the adenomatous polyposis coli (*APC*) gene and is estimated to occur in about 1 in 10,000 individuals. Classic presentation of FAP includes hundreds to thousands of adenomatous polyps occur by the age of 20 to 40 years.[1] The attenuated or less severe colonic phenotype associated with AFAP may mimic sporadic colon polyps and cancer, or other known syndromes, such as MAP. This creates diagnostic difficulties when evaluating an individual with moderate adenomatous polyposis. Other conditions linked to germ-line *APC* mutations include Gardner syndrome (with association of colonic polyposis and osteomas, epidermoid cysts, fibromas, and/or desmoid tumors) and Turcot syndrome (with association of colonic polyposis and medulloblastomas).[2] However, it is now believed that the features associated with Gardner syndrome and Turcot syndrome are the result of variable expressivity of *APC* mutations as opposed to being distinct clinical entities.

Screening Recommendation For Familial Adenomatous Polyposis:

Flexible sigmoidoscope should be used for screening at the age of 10-12 years. Optical colonoscopy is currently the most sensitive method for screening. Flexible sigmoidoscopy does not require conscious sedation and hemodynamic monitoring, and will typically allow visualization of the rectum, sigmoid colon, and descending colon to the splenic flexure. Barium enema allows visualization of the entire colon.

CT and magnetic resonance colonography, are receiving increased attention in clinical studies. CT colonography may also offer improvements in preoperative staging as one study found this technique to be highly predictive of T3-4 tumors.

Colonoscopy annually after age 10, gastroduodenoscopy after age 25, and treatment with nonsteroidal anti-inflammatory drugs reduces CRC risk. A prophylactic colectomy is highly recommended, with continued vigilance over the rectal stump and other at-risk tissues.

III. Management

Without treatment, CRC is inevitable in FAP. However, with early screening and polypectomies, in addition to prophylactic colectomies after polyps become too difficult to manage endoscopically, most CRCs can be prevented in AFAP and FAP. In FAP, annual colonoscopies or flexible sigmoidoscopies are recommended starting around the age of 10 years. In AFAP, screening begins in the late teenage years, and colonoscopies, rather than sigmoidoscopies, are necessary because of proximally located polyps.[4]. After polyps become too numerous (usually >20 to 30 polyps) to manage endoscopically or when adenomas with advanced histology are identified, a prophylactic colectomy is advised. A proctocolectomy with an ileal pouch anal anastomosis is the standard surgery in FAP, whereas a total colectomy with ileorectal anastomosis is often the

preferred approach with AFAP or in FAP cases with limited rectal involvement.[4] Continued screening of the remaining rectum or ileal pouch is still necessary.[4]

Recently, it has been shown that duodenal cancer detected through surveillance improves survival compared with individuals presenting because of symptoms.[5] NCCN currently recommends the consideration of an esophagogastroduodenoscopy (EGD) with a side-viewing examination beginning around the age of 25 years for duodenal cancer surveillance.[4]

IV. Surgical management

The three current surgical options for patients with FAP are total proctocolectomy (TPC) with permanent ileostomy, total colectomy with ileorectal anastomosis (IRA), and proctocolectomy with ileal pouch-anal anastomosis (IPAA).

IPAA can be a double-stapled, end-of-pouch-to-anus anastomosis, which may leave behind approximately 1 cm of anal transition zone.

Selection of the optimal procedure for an individual patient is based on several factors, including characteristics of the FAP syndrome within the patient and family, differences in likely postoperative functional outcome, preoperative anal sphincter status, and patient preference.[8]

TPC with permanent ileostomy is used in patients with invasive cancer involving the sphincters or levator complex, or patients for whom an IPAA is not technically feasible nor likely to lead to good function such as massive obesity or weak anal sphincters. However, TPC is occasionally chosen as a primary procedure by patients who perceive that their lifestyle would be compromised by the frequent bowel movements.

The key in deciding between an IPAA and an IRA is based primarily on the risk of rectal cancer development if the rectum is left in situ.

The risk of secondary rectal excision, due to uncontrollable rectal polyposis or rectal cancer, may be estimated by identifying the specific location of the causative *APC* mutation. At the current time, the choice between an IRA and an IPAA should be based primarily on clinical (rather than genetic) grounds[9]

Another important consideration in choosing between IPAA and IRA is postoperative bowel function and quality of life. Long-term follow-up demonstrates a comparable quality of life following IPAA for FAP relative to the patient's preoperative baseline.An IRA should be considered in specific circumstances, such as when there is mild rectal polyposis (as in AFAP), or a young patient with rectal sparing who is not interested in undergoing the multiple procedures that accompany an IPAA and diverting loop ileostomy, or a young woman interested in having children and trying to avoid the decreased fecundity associated with an IPAA procedure[10] The use of minimally invasive techniques such as laparoscopy may reduce the risk of infertility associated with IPAA.A diverting loop ileostomy should be performed in all IPAA procedures.

Endoscopic surveillance of the rectal segment at 6- to 12-month intervals after the index surgery is recommended. However, repeated fulguration and polypectomy over many years can lead to difficulty with subsequent polypectomy, reduced rectal compliance, and difficulty identifying flat cancers in the background of scar tissue. The development of severe dysplasia and/or villous adenomas not amenable to endoscopic removal is indication for proctectomy.

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