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An Unusual Case of Fap in Jharkhand-A Case Report

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ABSTRACT: Familial adenomatous polyposis (FAP) is an autosomal dominant condition characterized by diffuse intestinal polyposis, specific gene mutation, and predisposition for developing colon cancer. Left untreated, patients with FAP will develop colorectal carcinoma during early adulthood. Hence, early detection and surgical intervention are of the utmost importance. The most life-threatening extracolonic manifestations of FAP are periampullary carcinoma and desmoid tumors. The upper gastrointestinal tract should be monitored endoscopically at the time of diagnosis and assessed regularly thereafter. Further research is indicated in the development of effective screening and treatment for this condition.
Keywords:polyposis,colectomy,ileostomy,periampullary carcinoma ,desmoid tumour

I. INTRODUCTION :

Familial adenomatous polyposis (FAP) is an autosomal dominant inherited cancer-predisposition syndrome that is causally linked to the APC gene located on chromosome 5q21. It is defined a condition in which the large intestine sometimes small intestine contains multiple adenomatous polyps (typically more than 100).[1] The reported incidence of the disease ranges from 1 in 5,000 to 1 in 17,000 live births annually.. Disease penetrance is nearly 100% by 40 yr of age.[4]. The natural history of FAP includes the development of adenomatous polyps in the late teens to early twenties. Symptoms typically develop by the third decade of life. The most common symptom is bloody rectal discharge; however, complaints of abdominal pain, tenesmus, diarrhoea, and obstruction have been noted.[5] Often, the disease may be asymptomatic in the absence of malignant transformation of a colonic polyp. Left untreated, death usually occurs at a mean age of 42 yr,[5] which is approximately 20 yr earlier than the mean age of death from sporadic colorectal carcinoma.

II.CASE REPORT:

A 20 years old Hindu male was admitted to outpatient department with c/o bleeding per rectum and something coming out per rectum during defecation since 2 months. There was no history of weight loss or loss of appetite ,no comorbid condition with a similar complaints of bleeding per rectum is present among family members, However none have taken any proper medical advice and no diagnosis has been made earlier. Since his initial diagnosis, the patient has had intermittent episodes of hematochezia, shedding of polyps in the stool, and failure to thrive. He has had chronicanemia, requiring occasional transfusion over the last 20 yrs. On physical exam, the findings werenormal . Positive physical findings were limited to the rectal exam, with multiple small polypoidal growth of variable size ,smooth surface, friable in nature, irregular starting 4 cm from anal verge,proximal limit of extension couldn't be reached. Finger stained with blood on withdrawl.Laboratory examinations demonstrated hemoglobin of 4 gm/dL and hematocrit of 30.4%. The platelet count was normal. The patient underwent total proctocolectomy with ileoanal J-pouch anastomosis ,protective loop ileostomy was done. Colonoscopy demonstrated multiple polyps supportive of the diagnosis of FAP(fig 1).Macroscopic pathological examination of the colon demonstrated numerous polyps that ranged in size from 0.5-cm(sessile polyps) to 3.5-cm(pedunculated polyps) (Fig. 2). Microscopically, the polyps displayed severe glandular epithelial atypism (Fig. 3,4),suggestive of FAP.

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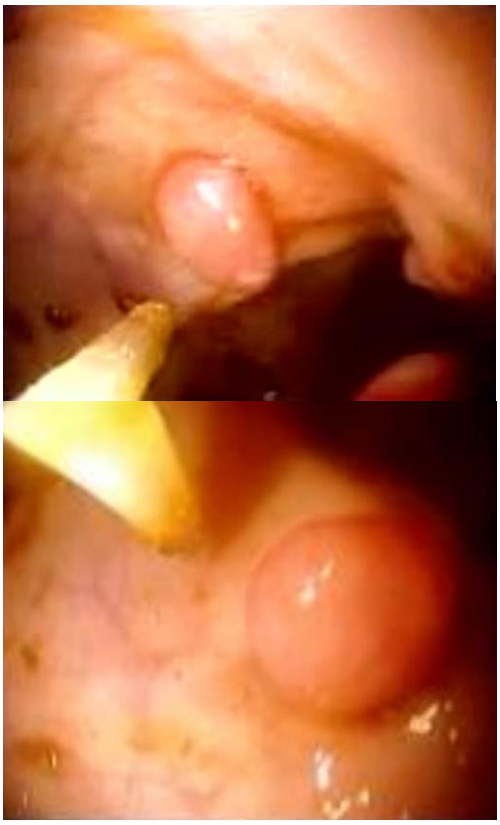

fig1

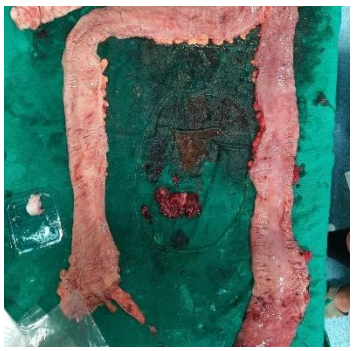

Fig 2

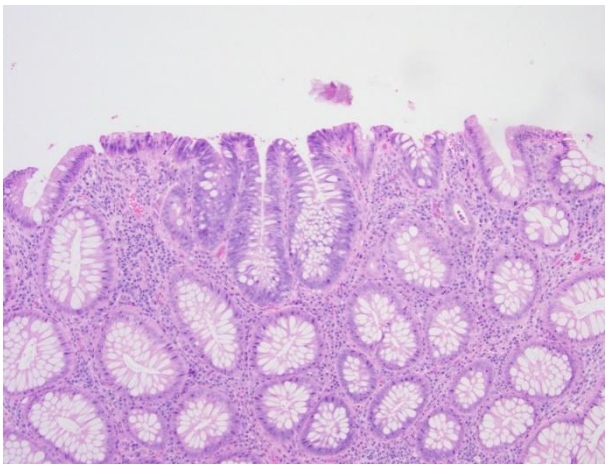

Fig 3

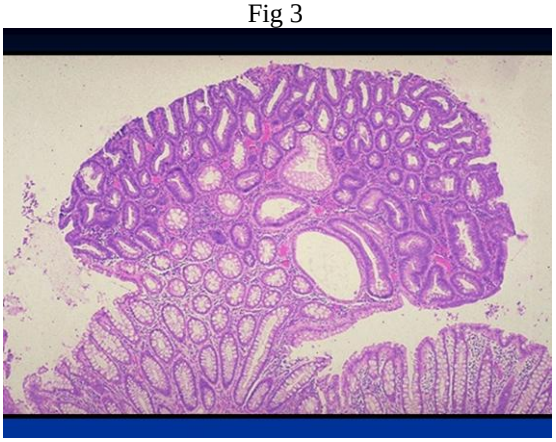

Fig 4

III.DISCUSSION :

The initial clinical description of multiple polyps of the large bowel is attributed to Menzelio who published his data in 1721[6]. The familial nature of multiple colonic polyposis was not recognized until nearly 100 years later when Cripps reported his findings in 1882.[5] The genetic nature of FAP was further elicited in 1925 by Lockhart-Mummery, who suggested the presence of an inherited predisposition that contributed to the development of adenomatous polyps with the potential for malignant change[7]. Identification of the specific genetic abnormality was discovered accidently in 1986 by Herrera and colleagues [8] when they found a deletion in the long arm of chromosome 5 of a patient with multiple colon and rectal polyps. The possibility that the APC gene was localized to 5q was confirmed by Bodmer and associates[9] and Leppert and coworkers[10]. These authors further recognized that theAPC gene is a tumour suppressor gene with mutations in the region of 5q21 that lead to hyperproliferative mucosa throughout the gastrointestinal tract and diffuse polyposis. Clinical presentation of patients with FAP typically include rectal bleeding or

diarrhea, the combination of which usually indicates the development of full-blown polyposis. Anemia resulting from blood loss may be present, as is evidenced in our patient. The presence of diarrhoea, bleeding, and abdominal cramping is an ominous finding, indicating that these patients may have developed an invasive malignancy[10]. Rarely,our patient developed symptoms at 20 years of age . Specific mutations of the APC gene correlate with the degree of colonic polyposis. Although unconfirmed by genetic studies, our patient likely has a mutation proximal to codon 1249 most likely including codon 1309 leading to a more aggressive disease course. Mutations proximal to codon 1249 are associated with sparse polyposis (less than 1000) mutations between codon 1250 and 1330 lead to a profuse phenotype (less than 5000) and mutations distal to codon 1465 again lead to sparse polyposis[11]. Furthermore, a mutation at codon 1309 has beenlinked to a more aggressive disease course with earlier onset of GI Symptoms.[12] Further diagnostic studies of patients suspected of having FAP include contrast barium enema, proctosigmoidoscopy, or colonoscopy. Diagnosis is then confirmed by the histologic findings of adenomatous polyps with a diffuse distribution throughout the large intestines.Prophylactic colectomy is required for patients with FAP to avoid the development of invasive colon cancer. Operative management includes removal of all large bowel mucosa that is at risk for malignant transformation while preserving near-normal bowel function and avoiding an ileostomy whenever possible. Options of therapies are: (a) total proctocolectomy with a Brooke ileostomy; (b) subtotal colectomy with ileorectal anastomosis; and (c) restorative proctocolectomy, including the formation of an ileal reservoir and an ileoanal anastomosis. Factors that may account for the outcome differences are age of the patient at initial operation, length of the retained rectal segment, quality and frequency of sigmoidoscopic follow-up, and cancer in the resected colon[13]. Restorative proctocolectomy with pouch formation and ileoanal anastomosis is an ideal surgical procedure for disease eradication in patients with FAP. The advantages include complete removal of all the large bowel mucosa, thereby eliminating colonic manifestations of the disease and risk of malignancy. Additionally, adequate bowel function is preserved and a permanent ileostomy is avoided. Bowel movements are generally of acceptable caliber and frequency with very few episodes of urgency, especially at night. Visualization of the sacral plexus with complete preservation was performed in our patient using meticulous sharp dissection. This procedure was further indicated in our patient due to the extensive disease involvement of his rectum. The complications of this procedure include, pouchitis, pelvic sepsis, anastomotic breakdown, and bowel obstruction. Performing a two-stage procedure by creating a temporary diverting loop ileostomy allows time for the anastomosis to heal minimizing anastomotic complications. Additionally, pouchitis is quite rare when restorative proctocolectomy is performed for FAP[14]. Overall, the ileoanal procedure appears to have particularly satisfactory outcome and is the procedure of choice for most patients.

IV.CONCLUSION:

Patients with FAP is very rare in Jharkhand and can be presented mostly with bleeding per rectum with or without any family history ,hence need to be carefully evaluated and all symptomatic patients need surgical treatment ,procedure of choice for most of the patients is total proctocolectomy with ileoanal j pouch anastomoses and protective loop ileostomy .

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