Abstract:
Familial adenomatous polyposis (FAP) coli is an autosomal dominant syndrome characterised by germline mutations of adenomatous polyposis coli (APC) gene. A 35 years old male presented with diarrhoea on and off, pain abdomen and weight loss since one year. Upper gastroendoscopy did not reveal any focal lesion. Colonoscopy showed numerous polyps throughout the colon and an ulcerative growth in the rectum. Patient underwent distal proctocolectomy with ileostomy. Excised specimen showed adenomatous polyposis coli with one focus of invasive carcinoma in the rectum. As mentioned in the literature familial adenomatous polyposis coli (FAP) can transform to invasive adenocarcinoma which was true in our case. We present this case because of its rarity.

Key words: Adenomatous polyps; Adenocarcinoma; Colonoscopy; Familial adenomatous polyposis coli; Sessile polyps

Introduction
Familial adenomatous polyposis (FAP) is an autosomal dominant inherited cancer-predisposition syndrome that is causally linked to the adenomatous polyposis coli (APC) gene located on chromosome5q21. Familial adenomatous polyposis is defined as an inheritable condition in which the large intestine contains multiple adenomatous polyps (typically more than 100) [1]. The incidence of the disease ranges from 1 in 5,000 to 1 in 17,000 live births annually [2]. Spontaneous germline mutations are common with resultant cases accounting for 20%-30% of patients with FAP [3]. Disease penetrance is nearly 100% by 40 yr of age.
Adenomatous polyps may also develop more proximally in the gastrointestinal (GI) tract, most notably in the stomach and second part of the duodenum. 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 bleeding per rectum however, complaints of abdominal pain, tenesmus, diarrhea, and obstruction have been noted [4]. These polyps are benign to start with and if no proper surgical intervention done malignant transformation is inevitable around the mean age range of 34 to 43 [5].

**Case report**

A 35 years patient presented with complaints of diarrhoea on and off, pain abdomen and weight loss since one year. Patient was ill nourished with pallor. Abdominal examination revealed vague tenderness in lower abdomen, no palpable mass or hepatosplenomegaly. There was no bleeding or mass on per rectal digital examination. Laboratory examination revealed microcytic hypochromic anaemia (haemoglobin 7gm/dl), liver and kidney function tests were within normal limits. Serological tests for human immunodeficiency virus, hepatitis B surface antigen were non-reactive. Upper gastrointestinal tract endoscopy showed no focal mucosal lesions. Colonoscopy detected numerous sessile polyps involving the entire colon extending from rectum to caecum, hence provisional diagnosis of FAP was considered. Detailed family history revealed no such illness in near kins. Total proctocolectomy with ileostomy was done.

Microscopic examination of the polyps revealed adenomatous polyps with mild to moderate dysplasia in some of them. Ulcerated area showed features of invasive well differentiated adenocarcinoma infiltrating up to serosa .Ileal surgical margins and anal verge were free of tumor infiltration, appendix was nil remarkable. Five out of fifteen lymph nodes (5/15) showed metastatic deposits of adenocarcinoma. A final diagnosis of FAP with synchronous invasive adenocarcinoma
colon with metastasis in the lymphnode, T3N1MX (American joint committee on cancer) was made.

Figure 4: Adenocarcinoma of colon – Malignant glands infiltrating Muscularis Propria (H & E, X40)

Figure 5: Lymph Node- Metastatic Adenocarcinomatous deposits (H & E, X40)

Discussion

The initial clinical description of multiple polyps of the large bowel is attributed to Menzelio who published his data in 1720 [6]. The familial nature of multiple colonic polyposis was not recognized until nearly 100 years later when Cripps reported his findings in 1882. The genetic nature of FAP was further elicited in 1925 by Lockhart Mummery, who suggested the presence of an inherited predisposition that attributed to the development of adenomatous polyps with the potential for malignant change. It was not until a formalized data base was developed by Cuthbert Dukes who organized the screening of relatives of patients with polyposis during the 1930’s and created the first register at St. Mark’s hospital in London, that the natural history of FAP was elicited in several generations. Majority of colorectal carcinomas are sporadic accounting for 75% followed by inherited genetic mutations in 25% among inherited colorectal carcinomas FAP and Hereditary Non Polyposis Colorectal Carcinomas (HNPCC) account for 5%. FAP is an autosomal dominant disease that results from mutations in adenomatous polposis coli gene located on chromosome 5q21. One third of all cases of FAP have no family history of FAP and these cases are thought to be caused by new germ line mutation [7], as is evidenced in our case.

The incidence of FAP ranges from 1 in 6000 to 1 in 12000 births with both sexes being equally affecting. By the age of 35 years 95% of individuals with FAP have more than 100 adenomatous polyps. Without colectomy colon cancer is inevitable in 100% by the mean age range of 34 – 43 years as is noted in our case. In 80% of patients with FAP there is family history of polyps and or colorectal cancer, while 20% recurrence is due to new germline mutations without prior family history as noted in our case [8,9].

Clinically presentation of patients with FAP typically include rectal bleeding or diarrhoea, the combination of which usually indicates the development of full blown polyposis. Anaemia resulting from blood loss maybe present. Most patients are asymptomatic until colorectal cancer develops. Making the diagnosis of FAP before the development of colorectal cancer is important for the patient as well as his family members who may be affected. Colonoscopy and sigmoidoscopy plays an important role in diagnosing the condition. ultrasound of the abdomen, liver function tests and computerized tomography scan aids in ruling out liver metastasis and other extraintestinal manifestations, diagnosis is then confirmed by the histopathologic findings of adenomatous polyps with a diffuse distribution throughout the large intestine, genetic testing provides ultimate diagnosis in 95% of cases but same was not carried out in our case due to financial constraints counselling for the family members regarding the hereditary nature of illness was explained. Most common surgical intervention for FAP is proctocolectomy with ileoanal anastamosis .If FAP is associated with adeno
carcinoma the treatment is surgical resection with chemotherapy and radiotherapy depending upon the stage of the disease.

**Conclusion**

Familial adenomatous polyposis colon is an autosomal dominant most commonly caused by a mutation in the APC gene at chromosome 5q21. It is characterised by early onset of numerous colonic adenomas with an inevitable progression to colorectal carcinoma if not detected early, so endoscopic surveillance, establishment of polyposis and surgical resection of FAP colon as early as possible can decrease the incidence and mortality from colorectal carcinomas.

**References**


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