Familial adenomatous polyposis (FAP), an autosomal dominant condition which is characterized by diffuse intestinal polyposis, specific gene mutation, and predisposes colon cancer. This inherited condition primarily affects the gastro-intestinal tract. This causes hundreds of polyps inside the colon and rectum. Symptoms typically appear in teenagers or young adults. Hereditary polyposis of the colorectum and familial polyposis are the other names given.

**ABSTRACT**

Familial adenomatous polyposis (FAP), an autosomal dominant condition which is characterized by diffuse intestinal polyposis, specific gene mutation, and predisposes colon cancer. This inherited condition primarily affects the gastro-intestinal tract. This causes hundreds of polyps inside the colon and rectum. Symptoms typically appear in teenagers or young adults. Hereditary polyposis of the colorectum and familial polyposis are the other names given.

**KEY WORDS**: Familial adenomatous polyposis (FAP), Inheritance, Colorectal cancer, APC mutations, Colonoscopy, Intestinal obstruction.

**Introduction**

FAP is a rare entity, inherited as autosomal dominant disease with 80-100% penetrance. The reported incidence of familial adenomatous polyposis varies from 1 in 7,000 to 1 in 22,000 individuals and it was estimated the prevalence of 1 in 5000 to 7500 Colorectal cancer which is the third leading cancer and the second leading cause of cancer deaths in the world. Approximately 5% of colorectal cancers have shown inherited syndromes of familial adenomatous polyposis or hereditary non-polyposis colon cancer. It has a high malignant potential of more than 95%. The average age at which an individual develops colon cancer in classic familial adenomatous polyposis is 39 years. The average age of colorectal cancer onset for attenuated familial adenomatous polyposis is 55 years. The offspring of affected individuals have a risk of inheriting FAP. However 20% of patients with FAP are new mutations without a family history. A patient affected with FAP was accidentally diagnosed and studied and the case study is presented here.

**Case Report**

48 years old lady presented in the casualty with...
complaints of abdominal pain since 15 days with 3 episodes of vomiting and loose stools since 1 day. Patient had a past history of few episodes of vomiting and loose stools 2 months back for which she was treated conservatively. On examination patient was conscious, dehydrated, moderately built & nourished with stable vitals. P/A – distended, diffuse tenderness, no VIP, no palpable mass, no organomegally, no ascitis and BS exaggerated. P/R – normal and empty. Plain X-Ray abdomen showed multiple air-fluid levels. USG showed features suggestive of acute intestinal obstruction. No liver metastasis was seen.

A provisional diagnosis of acute intestinal obstruction was made. After initial resuscitation under GA exploratory laprotomy was done and a growth was seen in sigmoid colon with no nodes in the mesocolon and after sigmoidectomy with a marginal clearance of 5 cms, numerous polyps were seen in proximal colon and in resected sigmoid, so temporary descending colostomy was done. On further investigation by colonoscopy through descending colon and anus revealed normal rectum and anal canal. Colonoscopy through colostomy and contrast CT showed multiple polyps throughout the colon. Biopsy of the resected specimen revealed well differentiated adenocarcinoma of sigmoid colon with resected margins free of malignancy and adenocarcinoma infiltrating the muscular layer sparring the serosa. Pericolic nodes were not metastatic. Her UGI scopy showed normal features. Her CEA values were normal. A diagnosis of FAP turning malignant at sigmoid level and presenting as acute intestinal obstruction was made and patient was taken up for total colectomy followed by ileo-rectal anastomosis. Postoperative period was uneventful. Biopsy of the total colectomy specimen showed familial adenomatosis polyposis. None of the polyps showed dysplasia or malignant transformation. There were no extra colonic manifestations like gastric or duodenal polyps, osteomas and soft tissue tumors. Post-operative UGI scopy showed normal features. CEA values were in normal limits. Patient was followed with chemotheraphy. Colonoscopy of her elder brother showed normal features. Colonoscopy for patient's 19 year old son revealed multiple polyps and biopsy revealed lymphoid hyperplasia. UGI scopy showed normal features. CEA values were normal. Patient is followed for 2 yr still date with regular colonoscopic examination of rectum showing normal features.
Discussion

The occurrence of colorectal cancer in more than one family member may be seen. It implies that the potential for developing colorectal cancer has been passed from one generation to the next. Relatives of a person with colorectal cancer may be more likely to develop it.

The initial clinical description of multiple polyps of the large bowel is attributed to Menzelio who published his data in 1721[1]. The familial nature of multiple colonic polyposis was not recognized until nearly 100 years later when Cripps reported his findings in 1882[2]. In 1925, Lockhart-Mummery suggested the presence of an inherited predisposition that contributes to the development of adenomatous polyps with the potential for malignant change[3]. Cuthbert Dukes developed a formalised database and organized the screening of relatives of patients with polyposis during the 1930s. He takes the credit for creating the first register at St.Mark's Hospital in London, about the natural history of FAP which was elicited in several generations.

In 1950, Gardner and Stephens described a syndrome comprising extra colonic manifestations of polyposis, including benign tumors of bone and soft tissue tumors such as lipomas, fibromas and sebaceous cysts (Gardner's syndrome). In 1986 by Herrera and colleagues found a deletion in the long arm of chromosome 5 of a patient with multiple colon and rectal polyps and identified the specific genetic abnormality. The APC gene was localized to 5q21 which was confirmed by Bodmer and associates, and Leppert and coworkers[4]. According to them Mutations proximal to codon 1249 are associated with sparse polyposis (<1,000 polyps), mutations between codon 1250 and 1330 lead to a profuse phenotype (<5,000 polyps), and mutations distal to codon 1465 again lead to sparse polyposis.

FAP is divided into two types - classic and attenuated based on the number of polyps and the age of onset.

A number of APC mutations have been described in a series of FAP patients. The clinical features of FAP appear to be generally associated with the location of the mutation in the APC gene and the type of mutation. Researchers have found that dense carpeting of colonic polyps is seen in APC mutations particularly that occur in between codons 169 and 1393.

Conclusion

First-degree relatives need rigorous screening and will need to have colonoscopy once every year from the age of 10. Most of these who are going to get polyps will have them at 20 yrs, and they require prophylactic colectomy following biopsy. If there are no polyps by 20 yrs, continue with 5 yearly examination until 50 yrs.

Screening should be carried out on children of affected parents, members of extended family who are at risk, and patients with suspected hereditary colon cancer.
Intraoperative Images

References: