Familial Adenomatous Polyposis Coli: A Case Report.

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Case Report: A 25-year-old man was admitted with a 6-month history of rectal bleeding, anal discharge and alteration in bowel habits. He gave a family history of a great-grandmother who died of unknown causes but had a colostomy, and a grandmother who died of Carcinoma of the oesophagus. Both relatives were on maternal side. On examination he had a low rectal mass, which was obliterating the anal orifice. Colonoscopy was not possible but rectal biopsy revealed a well-differentiated carcinoma of the rectum. Abdomino- perineal resection was done. During surgery, he was found to have multiple polyps. Colonoscopy through the colostomy and biopsy confirmed the presence of multiple benign polyps involving the entire colon up to the caecum. After counselling, the patient had total colectomy and an ileostomy done. Subsequent screening of family members revealed a 28-year-old sister who had occasional painless rectal bleeding. The 28-year-old sister of the patient above had colonoscopy done and was found to have adenomatous polyps involving most of the colon up to the caecum. A near- total colectomy and ileorectal anastomosis was done and cautery of the rectal polyps. She is being followed up with 6-monthly proctoscopy and is doing well.

Introduction

The incidence Familial Adenomatous Polyposis (FAP) in developed countries is 1:10,000 – 1:30,000 births. It is more common in males than females with an age of onset of 15 years. Before this age, the disease is reported to be asymptomatic and with no macroscopically visible polyps[1,2,3]. Some of these cases show numerous microscopic polyps and the smaller polyps tend to be asymptomatic[4], so it could be that younger patients have the microscopic form of the disease, which is asymptomatic.

There are very few symptomatic cases in the first decade of life. FAP is an autonomic dominant syndrome occurring due to a mutation in the APC gene on chromosome 5 locus q21. It is a pre-cancerous with 100% risk of malignancy. Extra colonic site involvement is found in 40 – 90% of patients, where polyps have been seen in the antrum, duodenum, periampullary region and the ileum[2,5]. Mandibular osteomas have been seen in some cases.

The diagnosis of FAP is made by the presence of 100 or more colonic polyps in a partially expressed phenotype and 1000 to 5000 polyps in a fully expressed phenotype. The disease may occur following spontaneous mutation in 20%-30%of patients, who therefore show a negative family history. A family history of FAP and colonic cancer is found in the rest of the patients[6,7]. All affected family members exhibit malignant change by the age of 35 years that is within 20 years of FAP diagnosis.

Colorectal cancer is the third leading cancer and the second leading cause of cancer deaths in the United States. Approximately 5 % of colorectal cancers have clearly defined inherited syndromes of familial adenomatous polyposis or hereditary non-polyposis colon cancer. The presence of a first-degree relative with colon cancer or adenomatous polyps increases the risk of other family members by two to three fold and is even higher if the cancer occurs in the relative before the age of 50 years. Early diagnosis and timely intervention is invaluable bearing in mind that 7% of untreated patients get cancer by age 21 and 90% by age 45 years[2,7].

Diagnosis is mainly by colonoscopy and presence of 100 or more polyps that are later confirmed adenomas is clear indication of the diagnosis[3]. Double contrast barium enema is also very useful but lacks the ability to confirm that the polyps are adenomas. First-degree relatives need rigorous screening and will need to have colonoscopy once every year. 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.The main stay of treatment of colorectal cancer is surgery; total colectomy is done with a permanent illeostomy or an illeal porch and illeal anal anastomosis[3,7].

Discussion

FAP has been a very rare diagnosis in Africa; the above family is the first documented one in Uganda and probably in East Africa. FAP diagnosis presents challenges in the African setting; colonoscopy is not readily available in many African centres so probably many cases will go undiagnosed. Barium enemas may be more available but as earlier discussed, the asymptomatic cases that are so important to catch early, exhibit very tiny polyps that are very difficult to pick on routine double contrast barium enema. Colonospy has further advantage, that when biopsies are taken, even the microscopic polyps may not be missed[3].

The grandmother who had a colostomy probably also had FAP related carcinoma colon, even the cancer oesophagus patient probably had extra colonic FAP. This family is a wake up call to Ugandan and indeed African Surgeons to realize that FAP should always be considered a possibility in many of the colon conditions that we handle.

The second case was treated by a near total colectomy and illeal rectal anastomosis. This is acceptable considering that she had very few rectal polyps[1], however one needs to regularly do proctoscopies, on a keen follow up program.

Total colectomy and illealanal anastomosis with or without anal pouch would sound more acceptable, but in the absence of stapling devices it is a very difficult procedure to do. This is yet another challenge, these devices are very costly and largely unavailable in our setting but preserving the anal sphincter is very important in these very young patients.

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