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A Rare Case of Familial Adenomatous Polyposis

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ABSTRACT

Familial adenomatous polyposis (FAP) is rare one.1 of colorectal cancer associated with FAP. It is a precancerous condition and is having 100 malignant potential. FAP is present in younger age group, so proper screening and prophylactic total colectomy is required. FAP is an autosomal dominant disease that results from mutation in the adenomatous polyposis coli (APC) gene located on chromosome5q21-22. One third of all cases of FAP have nil family history of FAP, and these cases are thought to be caused by a new germ-line mutation. The diagnosis of FAP is based on the detection of more than 100 adenomatous Colon polyps. We had a case of FAP in a 37-year-old man with history of abdominal pain and bleeding per rectum for 6 months.

Keyword: Familial Adenomatous Polyposis, Colonoscopy, Sigmoidoscopy and Colorectal Cancer.

CASE REPORT

A 37 years old man was presented to a primary care physician with recurrent hematochezia and constipation. He was diagnosed as hemorrhoids and was treated with laxatives (oral bisacodyl, 100 mg/day) and a sitzbath at bedtime. Approximately 6 months after the patient came to our hospital with complaints of abdominal pain, persistent hematochezia with loss of weight and fatigue.

Family History and Physical Examination

The patient had no family history of colorectal cancer or polyps. He has a male child of age 10 years and he has no specific complaints. Physical examination showed pallor. The blood pressure was 113/72 mm Hg, and pulse rate 85 bpm. The patient was afebrile. No pigmentation, osteoma, lipoma, or cyst was noted. Ophthalmologic examination was normal. The patient's abdomen was tender in the left upper quadrant, extending to the epigastric region. No mass lesions were palpated, and no ascites or rebound tenderness was demonstrated. P/R- no hemorrhoids, no mass palpable.

Laboratory and Radiographic Studies

Alanine aminotransferase level - 21 U/L (normal, 10 to 40 U/L) and Serum alkaline phosphatase level - 52 U/L (normal, 30 to 85 U/L)

Colonoscopy

Multiple polyps (more than 150) was present in the entire colon and ulceroproliferative lesion was seen in splenic flexure. No polyps were seen in rectum, biopsy was taken from splenic flexure. Biopsy from splenic flexure growth showed tubular adenoma and adenocarcinoma. Showed ulceroproliferative mass in splenic flexure with 150 to 250 polyps throughout the colon.

Discussion

Familial adenomatous polyposis (FAP) is a genetic condition where affected individuals will develop hundreds to thousands of polyps (abnormal, mushroom-like growths) throughout their gastrointestinal (GI) tract beginning at a young age (usually as a teenager or young adult). These polyps are usually found in the large intestine (colon and rectum), but they can develop in the stomach and small intestine as well. The polyps that form in the large intestine are known as adenomas. Adenomas are considered to be precancerous. In addition, individuals with FAP may also develop other features outside of the gastrointestinal tract. Attenuated FAP (AFAP) is a milder form of FAP. People with AFAP also develop precancerous polyps throughout the GI tract; however, the polyps in the colon tend to be fewer in number, usually less than 100. Since people with FAP or AFAP develop

many precancerous polyps in the colon and rectum, they have an increased chance that one or more of the polyps may develop into cancer of the large intestine (also known as colon cancer, colorectal cancer, or rectal cancer).

Pathogenesis

FAP is caused by an inherited gene change (mutation) in the Adenomatous Polyposis Coli (APC) gene. If the APC gene carries the correct biologic information, it helps protect the colon from developing polyps and cancer. If a mutation occurs in the *APC* gene, precancerous polyps will develop because the APC gene is no longer functioning properly and the colon is not protected. Most of the time, FAP is passed on to a child from the parent who has the condition. If the APC gene mutation is passed on to a child, he or she will inherit FAP. It is important to remember that parents do not have control over which genes are passed on to their children, and that passing on the APC gene mutation occurs by chance. In about one-third (33%) of all cases, people develop FAP even though their parents do not have FAP. When this occurs, it is due to a new gene alteration, or mutation, and it occurs by chance.

Diagnosis

There are several tests that can be used to diagnose FAP. These tests include genetic testing, flexible sigmoidoscopy and colonoscopy. Individuals with a family history of FAP should begin annual colon evaluation at 10-12 years of age. People with a family history of AFAP should begin colon evaluation by 18 years of age, or earlier based on family history. The following options should be discussed with a physician.

Genetic testing can be used to help diagnose FAP. With genetic testing, a small blood sample is taken from the individual with FAP and is sent to a special laboratory that studies the APC gene. About 80% of the time a change (also called an alteration or mutation) in the APC gene that leads to FAP is found. Genetic testing is useful in confirming a diagnosis of FAP in those rare cases where there is some doubt about the diagnosis. Once an individual is confirmed to have an APC gene mutation, genetic testing can help identify whether or not family members have FAP. Family members who do not have the APC gene mutation, did not inherit FAP. Therefore, they do not need to undergo screening procedures recommended for individuals with FAP. Genetic testing is recommended beginning at 10-12 years of age in families with FAP, which is also the age when screening for FAP should start. In families with AFAP, genetic testing is recommended beginning at 18 years of age, or earlier based on family history, which is also the age when screening for AFAP should start. Most people find it helpful to meet with a genetic counselor to talk about genetic testing. A **flexible sigmoidoscopy** is an examination of the rectum and the lower colon through a sigmoidoscope. The sigmoidoscope is a small flexible tube with a light on one end, which allows the

doctor to examine the inner lining of the lower part of the colon and rectum after inserting it through the anus. Prior to the exam, a sedative may be given to help the patient relax. During the exam, the doctor may take a small amount of tissue (biopsy) from the polyps for examination under a microscope.

A **colonoscopy** is a test in which the doctor looks at the inner lining of the large intestine (colon and rectum). This is done using an instrument called a colonoscope. A colonoscope is similar to the flexible sigmoidoscope, but longer. Prior to the colonoscopy, a sedative will be given to help the patient relax. Most people sleep through this procedure and feel little or no discomfort during this test. During the colonoscopy, the doctor may take a biopsy from the polyps for examination under a microscope. This is the recommended method of screening for families with AFAP. Both the flexible sigmoidoscopy and colonoscopy require that the colon be cleaned out ahead of time. The patient cleans out the colon by drinking a large volume of liquid laxative. The liquid laxative causes a temporary, overnight diarrhea.

Importance of Early Diagnosis

Early diagnosis of FAP is important for early detection and prevention of cancer. Cancer in FAP develops when cells in a polyp begin to grow out of control. People who are diagnosed with FAP are at risk for developing colon cancer. When multiple precancerous polyps are detected, it is treated by removing the colon before cancer occurs (Nagase et al., 1992; Rhodes and Bradburn, 1992; Jass et al., 1994; Yagi et al., 1998).

Surveillance Colon Polyps and Colorectal Cancer

A surveillance examination at regular intervals is critical. Beginning at age 10-12 years a flexible sigmoidoscopy is recommended every year for individuals with FAP. Once colon polyps are found, or by age 20-25 years, colonoscopy with chromoendoscopy (dye spray) should be completed every year. Individuals with AFAP are recommended to undergo a colonoscopy beginning at age 18 years, or earlier based on family history, and repeated every year. If present, precancerous polyps are removed during the colonoscopy **before** they develop into cancer, unless they are too numerous or too large to remove. **Upper Intestinal Polyps and Cancer** Surveillance of the upper GI tract is also very important for people with FAP or AFAP. Upper endoscopy exam (also called an EGD) should be performed every 1-3 years for monitoring of fundic gland (stomach) polyps and duodenal (first part of the small intestine) adenomas. A sideviewing scope is also recommended for viewing of the ampulla of Vater, which is a common location for polyps in the duodenum. Most individuals are recommended to start having EGD at age 20-25 yrs or just prior to the colorectal surgery.

Colectomy With Ileorectostomy (Ileorectal Anastomosis)

In this procedure, the colon is removed, but all or most of the rectum is left in place. The small intestine is attached to the upper portion of the rectum. The advantage of this procedure is that it is the least complicated operation. Most patients maintain very good bowel function, though antidiarrheal medications are sometimes needed. This procedure is typically recommended when there are very few polyps in the rectum.

Restorative Proctocolectomy (Ileal Pouch Anal Anastomosis)

This operation involves removing the entire colon and most of the rectum. A new rectum, or reservoir for stool, called a pouch, is made out of the lower end of the small intestine (ileum). The pouch is joined to the anus so bowel movements can flow in the normal way. A temporary ileostomy, or a stoma where the waste empties into a bag through the abdominal wall, is usually needed to help heal this delicate connection. The temporary ileostomy is then removed during a second less involved surgery about 8-10 weeks after the first surgery. This surgery is typically recommended when there are many polyps in the rectum.

Total Proctocolectomy With Ileostomy

This operation involves removing the entire colon and rectum. The end of the small bowel (ileum) is brought to the surface of the abdomen, where it is permanently stitched into place. This is called an end ileostomy. Because the rectum is removed, it is not possible to control bowel functions in the normal way. Liquid stool will come out of the ileostomy into a bag that is securely attached onto the skin of the abdomen. People go on to live normal lives after this type of surgery. An ileostomy should not be viewed as a handicap. Fortunately, few people need to have this kind of operation today.

Conclusion

FAP is an autosomal dominant disorder. 1% of colorectal cancer associated with FAP. It is a precancerous condition and associated with 100% malignant potential. Proper screening and early diagnosis with prophylactic total colectomy needed.

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Study significance:

The diagnosis of FAP is based on the detection of more than 100 adenomatous Colon polyps. We had a case of FAP in a 37-year-old man with history of abdominal pain and bleeding per rectum for 6 months.

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