Familial Adenomatous Polyposis: Introduction

The Hereditary Colorectal Cancer Website has been sponsored by the Robert Rauschenberg Foundation

Familial adenomatous polyposis (FAP) is an inherited condition that primarily affects the gastrointestinal tract. This disorder leads to hundreds or thousands of polyps inside the colon and rectum (less often in the stomach and small intestine). Symptoms typically appear in teenagers or young adults. Other names for this condition include hereditary polyposis of the colorectum, familial polyposis, and Gardner’s syndrome.

What is Colorectal Cancer?
Colorectal cancer is also called colon cancer or rectal cancer. It refers to any cancer in the colon from the beginning (cecum) to the end (rectum). Colorectal cancer occurs when cells that line the colon (large bowel or intestine) or the rectum become abnormal and grow out of control. Polyps are usually benign growths that protrude from a mucus membrane. They can form in the colon and rectum. These polyps are adenomatous polyps and may eventually progress into cancer if left untreated.

What is Familial Colorectal Cancer?
The occurrence of colorectal cancer in more than one family member may be due to chance alone. However, it could also mean the potential for developing colorectal cancer has been passed from one generation to the next but the exact gene has not been identified. Relatives of a person with colorectal cancer may be more likely to develop it.
It is estimated that 15–50% of colorectal cancers are familial. A single gene, a combination of genes, or a combination of genetic and environmental factors can contribute to familial colorectal cancer. Typically these families have one or two members with a history of colorectal cancer or pre-cancerous polyps.

What is Hereditary Colorectal Cancer?
A family has hereditary colorectal cancer when the exact gene that causes the disease is known. Several genes that cause hereditary colorectal cancer have been identified. There may be other genes that have not yet been discovered.

If more than one person in a family has colorectal cancer, it could mean that the potential for developing this form of cancer has been passed from one generation to the next. In these families, relatives of people with colorectal cancer may be much more likely to develop it.

Inherited colorectal cancers are associated with a genetic mutation in a cancer susceptibility gene. Everyone inherits one susceptibility gene from each parent. If a mutation in one copy of a cancer susceptibility gene is passed from the parent to child, the child is predisposed to develop cancer.

The genetic causes of two hereditary colorectal cancer syndromes, familial adenomatous polyposis (FAP) and hereditary nonpolyposis colorectal cancer (HNPCC) have been identified.

What is FAP (Familial Adenomatous Polyposis)?
Familial adenomatous polyposis is an inherited condition caused by a mutation on chromosome 5. FAP is characterized by hundreds to thousands of polyps in the gastrointestinal tract (primarily the colon and rectum). These polyps are at increased risk for malignancy. Polyps begin to appear during teen years and often become cancerous by age 40.

Polyps (abnormal mushroom-like growths) may vary in size from less than one tenth of an inch to 1-2 inches. They may be large enough to block part of the intestine. If patients with FAP are left untreated, one or more of these polyps may develop into cancer. FAP may also be associated with benign tumors of the skin, soft connective tissue, and bones. It is caused by a mutation in a gene called APC. In some people FAP may be inherited, whereas in others it is not (sporadic).
What is Attenuated FAP?
Some families are affected with attenuated FAP (AFAP). This is a mild (or attenuated) form of FAP in which affected individuals develop fewer polyps at a later age than those with typical FAP. Although people with AFAP tend to develop colon cancer at a later age than individuals with typical FAP, they still have a near 100% lifetime risk of colon cancer. If AFAP is suspected within a family, it is important that family members be screened with colonoscopy rather than flexible sigmoidoscopy because polyps are not evenly distributed throughout the colon. Because the number of polyps and age of onset can vary greatly from one family member to another in a family with AFAP, screening should begin at age 11.

How is FAP inherited?
People with FAP have a 50% chance of passing the condition to each of their children. The condition can be passed on even if the patient has had his or her own colon removed. Children who do not inherit the condition from their parent cannot pass it to their own children.

About one third of people with FAP do not have an affected parent. An individual who is the first in the family to have the condition has a new or spontaneous mutation. Individuals with new mutations can pass FAP to their children.

Figure 7. FAP family pedigree

(1) George has FAP, his wife Susan is unaffected. They have 3 children, George, Jr., Stephen, and Carol. All were at 50% risk of developing FAP. George, Jr., and Carol are affected.

(2) George, Jr. and his wife Connie have 2 children. Both children had a 50% chance of inheriting FAP. Their daughter Alice is affected.

(3) Stephen and his wife Gloria have 2 children. Because Stephen is unaffected, there was no risk to his children of developing FAP.

(4) Carol and Bill have 3 children. Each child had a 50% chance of inheriting FAP and two are affected; son Billy is not.

Genetic counseling is available and recommended for individuals with FAP and for their family members. Genetic counselors will explain the inheritance pattern of FAP, discuss which family members are at risk for developing the condition, and provide necessary information regarding genetic testing. Counseling services are available through genetic and oncology departments in many hospitals. To make an appointment with a member of the Johns Hopkins professional medical team or to speak with someone at the Colorectal Cancer Risk Assessment Service, please call (410) 614-LIFE (5433). Also, it is recommended that FAP families contact registries for access to resources and for help with identification of family members at risk.

Symptoms
Many patients with colorectal cancer experience no symptoms in the early stage of their disease. In fact, many people have no symptoms until the disease is quite advanced. Therefore, routine colorectal screening and an appreciation of risk factors are extremely important.

Since many of the symptoms of colorectal cancer are also symptoms of a variety of other colon diseases, it is important to see your physician so that the necessary tests can be run and a diagnosis made.

The following is a list of symptoms that may occur:
- Blood in the stool
- Diarrhea that is not the result of diet or illness
- A long period of constipation
- Crampy pain in the abdomen
- Change in bowel habits
- Persistent decrease in the size or caliber of stool
- Frequent feeling of distention in the abdomen or bowel region (gas pain, bloating, fullness, with or without cramping)
Weight loss with no known reason
Vomiting and continual lack of energy

In addition to polyps, abnormalities in other areas of the body may give early clues to the presence of FAP. These abnormalities may include bumps or lumps on the bones of the legs, arms, skull, and jaw; cysts of the skin; teeth that do not erupt when they should; and freckle-like spots on the inside lining of the eyes.

It should be emphasized that there is no safety in simply waiting for symptoms to develop. It is vital that parents and guardians make every effort to have examinations of their children starting at age 11 even if they do not have symptoms.

Resources

American Cancer Society
The American Cancer Society
National Headquarters
1599 Clifton Road, N.E.
Atlanta, Georgia 30329
800-ACS-2345
http://www.cancer.org/

The ACS can offer assistance if cancer should occur. Check the telephone directory for your local chapter.

State Vocational Rehabilitation Service
This service offers training for another vocation if one should be physically unable to return to the same kind of work performed before the surgery. Check your telephone directory for your state services.

Generation to Generation
This newsletter is for people concerned about hereditary colon cancer and polyposis. It is free to all individuals with these conditions and their families and features articles on new research, the genetics of cancer, treatment, and quality of life, among many other topics. The editors welcome suggestions, questions, and personal stories from readers. This publication may be obtained by contacting Editor, Generation to Generation, Dept. of Behavioral Science, Box 243, M.D. Anderson Cancer Center, 1515 Holcombe Blvd., Houston, TX 77030.

Polyposis and Hereditary Colorectal Cancer Registries
Registries may be contacted for the names of experts in the management of FAP. Registries can also help to identify relatives at risk for the disorder. Further information concerning Polyposis Registries may be obtained by clicking here or contacting:

Coordinator, Hereditary Colorectal Cancer Registry
The Johns Hopkins Hospital
550 North Broadway, Suite 108
Baltimore, MD 21250-2011
Phone: 1-888-77-COLON
Fax: 410-614-9544
E-mail: hccregistry@jhmi.edu

Cancer Information Service (CIS)
Toll Free: 1-800-4-CANCER
The Cancer Information Service is a national toll-free telephone inquiry system that provides information about cancer and cancer-related resources to the general public, patients and their families, as well as health professionals. Most CIS offices are associated with Comprehensive Cancer Centers or community hospitals. CIS offices do not diagnose or recommend treatment for individuals. They provide support, understanding, and rapid access to the latest information, as well as referral to local services and resources. Printed materials may supplement telephone information. All calls are kept confidential, and individuals do not need to give their names.

The National Society of Genetic Counselors, Inc. (NSGC)
The National Society of Genetic Counselors, Inc. (NSGC)
233 Canterbury Drive
Wallingford, PA 19086-6617
610-872-7608
www.nsgc.org

The National Society of Genetic Counselors is the professional membership association for the genetic counseling profession. NSGC has developed a resource link to assist consumers in locating genetic counseling services.
The colon and rectum are part of the digestive tract. The digestive tract is a hollow tube that begins at the mouth and ends at the anus. It has several parts including the esophagus, stomach, small intestine, colon, and rectum (the colon and rectum make up the large intestine).

The intestine is about 28 feet long. The last 5–6 feet of the intestine is called the colon or large intestine. This structure has six major divisions: cecum, ascending colon, transverse colon, descending colon, sigmoid colon, and rectum. The last 5 or 6 inches of the large intestine is the rectum.

The purpose of the digestive system is to remove nutrients (minerals, vitamins, carbohydrates, proteins, fats, and water) from the foods we eat and to store the waste. After food is digested, solid wastes move through the colon and rectum to the anus, where they are passed out of the body.
Familial Adenomatous Polyposis: Causes

Overview
The exact causes of colorectal cancer are not known. However, studies have shown that genetics, diet, and lifestyle may affect the risk of developing colorectal cancer.

Family History
Family history is one of the most significant risk factors for colorectal cancer. People who have cancer, colorectal cancer, non-cancerous colon polyps, or inflammatory bowel disease are at increased risk. People who have been treated previously for colorectal cancer are at risk for recurrence. Women with uterine, ovarian, and breast cancer are also at increased risk.

Genetics
People with FAP, HNPCC, and APC I1307K have a 50% chance of passing the condition to each of their children.

Diet and Lifestyle
Cancer of the colon or rectum (colorectal cancer) is one of the most common cancers in the United States. It occurs in both men and women, usually over the age of 50. The exact causes of colorectal cancer are not known. However, studies have shown that changes in diet and lifestyle can decrease your risk of developing colorectal cancer.

The American Cancer Society recommendations for a healthy diet are consistent with the USDA Food Guide Pyramid. Although no diet can guarantee full protection against any disease, the Food Pyramid offers the best nutritional information currently available to help reduce the risk of cancer.

Dietary Recommendations
Fruits and vegetables
The American Cancer Society recommends eating at least five servings of fruits and vegetables daily. Diets rich in raw, green, dark yellow, and cruciferous (cabbage family) vegetables are best.

Fiber
Fiber forms bulk that keeps things moving through the colon, preventing constipation. It fills you up, aids digestion, and lowers cholesterol and blood pressure. A high-fiber diet includes vegetables, whole grains, and beans. High-fiber (starchy) vegetables include potatoes, lima beans, green peas, winter squash, corn, yams, and sweet potatoes. Whole grains are brown rice, barley, millet, and quinoa. Beans are lentils, chickpeas, great northern/navy/lima beans, and split and black-eyed peas (prepared without meat or grease).

Meats
The American Cancer Society recommends no more than 6 ounces of cooked lean meat, fish, and poultry per day. Avoid red, processed, cured or fried meat with heavily browned surfaces.

Fats
Limit your intake of high-fat foods. Avoid foods high in saturated/animal fats. Reduce your total amount of dietary fat to less than 30% of your total daily calories. Use low-fat cooking methods such as steaming, broiling, and baking.

Calcium
Be sure you are getting enough calcium in your diet. Good sources of calcium include milk, yogurt, broccoli, and grains. Take supplements if needed.

Folate
A daily multivitamin containing folic acid or folate may lower colorectal cancer risk.

Lifestyle Recommendations
Physical activity
Increased physical activity is a way people can decrease their risk for colorectal cancer. Small amounts of exercise on a regular basis can be helpful. The American Cancer Society recommends at least 30 minutes of physical activity three times a week.

Maintain a healthy weight/low body mass
Decreasing obesity can be achieved by maintaining a healthy weight through a healthy eating plan using the USDA Food Pyramid and exercise.
**Avoid smoking**
There is an increased risk of developing precancerous colorectal polyps in people with a long smoking history.

**Avoid excessive alcohol**
There is an increased risk of developing colorectal cancer in people with a history of excessive alcohol intake, regardless of the type of drink.

**Other Recommendations**

*NSAIDs (Nonsteroidal anti-inflammatory drugs)*
Studies suggest that taking NSAIDs (such as aspirin and ibuprofen) reduces the risk of colorectal cancer. These medications work by reducing polyp formation. Not everyone should take NSAIDs. Talk to your doctor about whether this is right for you and, if so, how much should you take.

*Estrogen replacement therapy*
Studies show that ERT (estrogen replacement therapy) may reduce the risk of colorectal cancer by one half in postmenopausal women. The decision to take estrogen should be based on discussions of the benefits and risks with your doctor.
Familial Adenomatous Polyposis: Diagnosis

Genetic/DNA Testing

Genetic testing is available for Familial Adenomatous Polyposis (FAP), Hereditary Nonpolyposis Colorectal Cancer (HNPCC), and APC I1307K.

The altered gene that causes FAP in most patients is now known.

A blood test can locate the change in the APC gene, which is causing the disease in about 80% of families with FAP. In these families, children can learn if they have inherited the changed APC gene from their parent with FAP. If a child does have the gene mutation, he or she will eventually develop FAP. The mutation in the APC gene cannot be detected in about 20% of families with FAP. Children in these families must continue to have regular colon screening according to the guidelines for families in which a gene mutation could not be found.

Figure 10. Chromosome 5 for FAP.

The test requires a small blood sample. The gene test results will influence the future management of individuals who are at risk for FAP. For example, if the test shows that a person does not have the APC gene mutation known in the family, he or she can avoid many unnecessary colon examinations. If the test shows that a person does have the gene mutation, then the physician will need to be alerted to schedule colon examinations at least yearly and to plan for future treatment when polyps develop.

Is insurance discrimination a risk for those seeking genetic testing?

With any gene test, there is a risk of insurance (life or health) or employment discrimination. At this time, federal laws offer some protection against the discrimination against people who already have medical insurance through a group health plan. Some states have other laws to protect people, but the laws on insurance and employment discrimination vary from state-to-state. The Americans with Disabilities Act may protect people with a positive gene test result from discrimination in the workplace. These issues will be carefully explained during the genetic counseling session.

Exam guidelines for people at risk for FAP

Follow the guidelines listed below if:

Table 1. You have never had genetic testing for FAP, or a family member with FAP had DNA testing and a mutation could not be found:

<table>
<thead>
<tr>
<th>Age</th>
<th>Examination</th>
</tr>
</thead>
<tbody>
<tr>
<td>Birth - 7 years</td>
<td>Physical exam and/or abdominal ultrasound for liver enlargement and serum alpha-fetoprotein test every year to check for hepatoblastoma.</td>
</tr>
<tr>
<td>11 - 24 years</td>
<td>Flexible sigmoidoscopy* every year</td>
</tr>
<tr>
<td>25 - 34 years</td>
<td>Flexible sigmoidoscopy* every other year</td>
</tr>
<tr>
<td>35 - 50 years</td>
<td>Flexible sigmoidoscopy* every 3 years</td>
</tr>
<tr>
<td>Over 50 years</td>
<td>Flexible sigmoidoscopy* every 5-5 years</td>
</tr>
</tbody>
</table>

*If polyps are found, surgery will be recommended.

Table 2. If you had DNA testing and you do have a gene mutation causing FAP:

<table>
<thead>
<tr>
<th>Age</th>
<th>Examination</th>
</tr>
</thead>
<tbody>
<tr>
<td>Birth - 7 years</td>
<td>Physical exam and/or abdominal ultrasound for liver enlargement and serum alpha-fetoprotein test every year to check for hepatoblastoma.</td>
</tr>
<tr>
<td>Beginning at age 11</td>
<td>Flexible sigmoidoscopy* every year</td>
</tr>
</tbody>
</table>

*If polyps are found, surgery will be recommended.

Table 3. You have had DNA testing and you do not have the gene mutation causing FAP that was found in your affected family member:
Table 3.

<table>
<thead>
<tr>
<th>Age</th>
<th>Examination</th>
</tr>
</thead>
<tbody>
<tr>
<td>25 - 35 years</td>
<td>To be certain that the DNA test was correct, a flexible sigmoidoscopy* should be performed at least once between the ages of 25 and 35.</td>
</tr>
</tbody>
</table>

*Colonoscopy recommended in place of flexible sigmoidoscopy for families with Attenuated FAP (AFAP).

Screening Guidelines for Families with a History of Colorectal Cancer or Polyps

The following cancer screening guidelines are for people who have a family member with colon or rectal (colorectal) cancer or adenomatous (precancerous) polyps. When one family member gets colon or rectal cancer, his or her close relatives (parents, sisters, brothers, and children) have a higher risk of developing colon or rectal cancer than the general population.

These guidelines were developed to help patients and physicians. They are for people who may be at risk for developing colon or rectal cancer.

**Group 1**

People who do not have a family member with colorectal cancer or adenomatous polyps.

or

People who have a family member with colorectal cancer or adenomatous polyps but the affected family member is not their parent, sister, brother, or child (not a first-degree relative).

**Group 2**

People who have one parent, sister, brother, or child who had colon or rectal cancer after age 50 or adenomatous polyps after age 60.

**Group 3**

People who have one parent, sister, brother, or child who had colon or rectal cancer before age 50 or an adenomatous polyp before age 60. OR People who have two close relatives (parent, sister, brother, child) who had colon or rectal cancer after age 50 and/or an adenomatous polyp at any age. Individuals should consider genetic counseling and testing for genes associated with hereditary colon and rectal cancer.

**Group 4**

People who have a stronger family history than described in Group 3 should discuss the Group 4 guidelines with their doctor. For example, they would fit in Group 4 if their mother, her father, and her sister have colon or rectal cancer and at least one of them was under age 50 when they were diagnosed. Individuals should consider genetic counseling and testing for genes associated with hereditary colon and rectal cancer.

**Colon/Rectum**

**Group 1**

Yearly test for blood in stool, start at age 50. Flexible sigmoidoscopy with digital rectal exam every 5 years, start age 50 (1; see note at the bottom of the page).

**Group 2**

See note (4) at the bottom of the page for Group 2.

**Group 3**

Yearly test for blood in stool, beginning at age of first colonoscopy. Colonoscopy with digital rectal exam every 3–5 years, beginning either at age 35–40 or 5–10 years earlier than the earliest age at which colon or rectal cancer occurred in your family, whichever is younger (5).

**Group 4**

Yearly test for blood in stool, beginning at age of first colonoscopy. Colonoscopy with digital rectal exam every 1–3 years, beginning either at age 25 or 5–10 years earlier than the earliest age at which colon or rectal cancer occurred in your family, whichever is younger (6).

**Female organs**

**Group 1**

Yearly pelvic exam with Pap test starting at age 18, younger if sexually active (2). At menopause, endometrial biopsy if recommended by your doctor (3).

**Group 2**

See note for Group 2 (4).

**Group 3**

Yearly pelvic exam with Pap test starting at age 18, younger if sexually active (2). Endometrial biopsy and/or transvaginal ultrasound if recommended by your doctor (3).

**Group 4**

Yearly pelvic exam with Pap test starting at age 18, younger if sexually active (2). Endometrial biopsy and/or transvaginal ultrasound beginning age 25–35 (3).

**Breast**

**Group 1**

Breast self-exam every month. Exam by doctor and mammography (breast x-ray) at intervals suggested by your doctor.

**Group 2**

See note for Group 2 (4).

**Group 3**

Breast self-exam every month. Exam by doctor and mammography (breast x-ray) at intervals suggested by your doctor.

**Group 4**

Breast self-exam every month. Exam by doctor and mammography (breast x-ray) at intervals suggested by your doctor.

(1) Other alternative exams include double-contrast barium enema every 5–10 years or colonoscopy every 10 years (both with digital rectal exam). Discuss these options with your doctor.

(2) After three normal exams, Pap test may be done less frequently. Discuss with your doctor.

(3) A family history of endometrial and/or colon cancer may increase your risk of endometrial cancer.

(4) Note for Group 2: Because doctors are uncertain about the best guidelines for patients in this group, we recommend that you discuss the guidelines with your doctor.

(5) These are broad guidelines. Discuss them with your doctor to decide what age and testing interval is right for you. Flexible sigmoidoscopy and air-contrast barium enema exam may be substituted for colonoscopy but are not preferred.

(6) Your doctor should decide what age and testing interval are right for you.
Physical Exam and History
Doctors should keep a detailed history on each patient including personal and family medical histories. The physician will perform a physical examination and may order laboratory tests. Other diagnostic tests described below may be performed.

Digital Rectal Exam
This is a painless examination of the rectal area. The doctor inserts a gloved, lubricated finger into the rectum to gently feel for any abnormalities.

Fecal Occult Blood Test
The fecal occult blood test is performed to identify hidden (occult) blood in the stool. The patient is given three small cards and asked to provide samples from three consecutive bowel movements. A small amount of stool is placed on a special test strip on each card. This stool may be tested in the doctor’s office or sent to a laboratory to see if there is any occult blood in the specimen.

Figure 11. Fecal occult blood test card

Endoscopic Diagnosis

Flexible Sigmoidoscopy
The flexible sigmoidoscopy is an examination of the rectum and the lower colon. It is performed with a lighted, flexible, hollow tube, which is about the thickness of a finger.

Figure 12. A, Position of the flexible sigmoidoscope in the colon; B, tip of the sigmoidoscope; C, endoscopic image.

Before having a sigmoidoscopy the colon must be clear of stool so that the doctor has good visibility. The patient must undergo a preparation that may include a liquid diet, enema, and laxatives to clear stool from the colon. The technique for bowel preparation may differ by health facility.

The sigmoidoscope is inserted into the anus through the rectum and into the large intestine. Using the sigmoidoscope, the doctor can see whether polyps or cancer are present. At the time of sigmoidoscopy, a biopsy forceps may be inserted through a channel of the scope to remove a small sample of tissue for microscopic examination. Sometimes it is necessary for the doctor to introduce air into the sigmoid colon to improve visibility. Most patients feel a little cramping or discomfort when having a flexible sigmoidoscopy.
**Colonoscopy**

A colonoscopy is an examination of the rectum and the entire colon. It is performed with a lighted, flexible, hollow tube, which is slightly larger in diameter than an enema tube. Colonoscopy permits the doctor to see much farther into the bowel than sigmoidoscopy.

The colonoscope allows the doctor to see whether polyps or cancer are present.

A biopsy forceps may be inserted through a channel in the colonoscope to remove a small sample of tissue for microscopic examination. Sometimes it is necessary for the doctor to introduce air into the colon to improve visibility. Before having a colonoscopy the colon must be clear of stool so that the doctor has good visibility.

The technique for bowel preparation may differ by health facility.

Moderate sedation is administered before a person undergoes colonoscopy. Many people sleep through the whole procedure and feel little or no discomfort. Occasionally, the insertion of air during the procedure may cause the same kind of discomfort as gas pain.

**Barium Enema**

A barium enema is a radiological examination of the rectum and the entire colon. Before a barium enema, the patient may have to undergo a preparation that includes a liquid diet, enema, and laxatives to clear stool from the colon. This preparation may differ from exam to exam and from one doctor to another. Before having this exam, a barium preparation (a contrast material) is administered through a rectal tube. This contrast material outlines the colon. The test allows the colon to be visualized when the x-ray picture is taken. If polyps or cancer are present, they can usually be seen on the x-ray.
The barium enema feels similar to an ordinary enema, causing a feeling of fullness. This test should not be performed on pregnant women because of the risk of x-rays (radiation) to the fetus.
Familial Adenomatous Polyposis: Therapy

Surgical Therapy
If polyps are found at examination, the doctor will recommend colorectal surgery. Removing the colon after polyps begin to develop is the only way to prevent the development of colorectal cancer. The goal of surgery is to completely remove the cancer, alleviating symptoms and offering the best chance of cure. Preoperative evaluations to rule out metastatic disease may include colonoscopy, barium enema, endoscopic ultrasound (EUS), CAT scan, chest x-ray, liver, and CEA (for baseline levels). Proctocolectomy and colectomy, demonstrated below, show the difference between having both colon and rectum removed and having just colon removed, while the rectum remains intact.

Several different operations are currently available for treatment. The four most commonly performed operations are:

1. Total proctocolectomy with Brooke ileostomy (with pouch)
2. Total proctocolectomy with Koch pouch
3. Colectomy with ileorectostomy
4. Restorative proctocolectomy (ileoanal pouch procedure)

Total Proctocolectomy with Brooke Ileostomy
This procedure involves complete removal of the entire colon and rectum. The end of the small intestine is brought out as an ileostomy. Although the restorative proctocolectomy is the preferred procedure in most cases, total proctocolectomy with Brooke ileostomy is generally performed in situations when invasive cancers are present in the rectum, when the anal sphincter is not functioning correctly, or in elderly patients in whom strength of the sphincter muscles is diminished.

An ileostomy is an opening on the abdomen through which stool leaves the body. An ileostomy can be temporary or permanent. In most cases it is necessary to wear an appliance called an ileostomy bag to collect body wastes. An ileostomy should not be considered a handicap, although it is an inconvenience. With proper care, there should be no odor or uncleanliness. Thousands of people of every age and of both sexes have had ileostomy surgery. After surgery, people can be just as busy, successful, and involved in daily routines as before surgery. In fact, they may be more active because of improved health.

Total Proctocolectomy with Koch Pouch
This operation involves complete removal of the colon and rectum with the creation of a continent ileostomy. It is similar to that of the Brooke ileostomy, but here a pouch is created under the abdominal wall with a continent. This operation is rarely done today, but can be considered in selected cases in which transanal reconstruction is not a good option.

Colectomy with Ileopectomy (ileopectomy Anastomosis)
In this procedure, the colon is removed but all or most of the rectum is preserved. The small intestine is attached to the upper portion of the rectum. The advantage of
the ileorectostomy procedure is that it is a less complicated, one-stage operation, yet still preserves fecal continence and maintains tolerable bowel function. The main concern with this procedure is that the rectum is left in place, despite its potential propensity for the development of polyps and cancer. Colectomy with ileorectal attachment is generally performed in patients who have few or no polyps in the rectum. In this case, often the remaining rectum requires frequent surveillance and removal of premalignant polyps.

Restorative Proctocolectomy (Ileoanal Pouch Procedure)
The restorative proctocolectomy involves removal of the entire colon and most of or the entire rectum. The end of the small intestine (ileum) is attached to the very distal rectum with the creation of an ileal pouch. With this operation, either a small portion of rectal mucosa can be left intact or the remainder of the rectal lining can be stripped. In this way, continence can be maintained and yet all or nearly all of the at-risk large intestinal lining can be removed. The ileal pouch provides a reservoir for fecal storage. Typically, the operation is performed in two stages. In the first stage a temporary ileostomy is created. After a period of time the ileostomy is removed to direct the intestinal stream during the healing of the ileoanal pouch. Alternatively some medical centers favor a single-stage procedure where no temporary ileostomy is used.
All operations involve removal of all or most of the colon. After a complete discussion of these operations, the patient and surgeon together can decide which one is best.

In some cases, after colon removal, a person may have an ileostomy. An ileostomy is an opening on the abdomen through which stool leaves the body. An ileostomy can be temporary or permanent. In most cases it is necessary to wear an appliance called an ileostomy bag to collect body wastes. An ileostomy should not be considered a handicap, although it is an inconvenience. With proper care, there should be no odor or uncleanness. Thousands of people of every age and of both sexes have had ileostomy surgery. After surgery, people can be just as busy, successful, and involved in daily routines as before surgery. In fact, they may be more active because of improved health.

**Colorectal Cancer Staging**

Colorectal cancers are staged (or classified) at the time of surgery. Staging describes the extent that the tumor has gone into or through the bowel wall and determines whether or not the cancer has spread to lymph nodes or other organs. This is done by microscopic exam of the tumor cells removed during surgery and by radiological exam of the area around the colon using CAT scan or endoscopic ultrasound (EUS).

There are two different methods of describing a colorectal cancer stage.

Both systems are described below, and are compared with each other in the table at the end of this section.

**Dukes System**

The modified Dukes system separates colorectal cancers into four groups—A, B, C, and D.

- *Dukes A*—includes tumors that are found only in the inner wall of the colon or rectum.
- *Dukes B*—includes tumors that have penetrated the muscle layer of the bowel wall or have gone through the bowel.
- *Dukes C*—includes tumors that have spread to lymph nodes in the same region.
- *Dukes D*—includes tumors that have spread to distant sites, such as the liver.

**TNM System**

The TNM system separates colorectal cancer into five stages—Stages 0–IV.

This system is recommended by the American Joint Commission on Cancer. In this classification system three different characteristics of the tumor are evaluated and assigned a value.
Table 4.

<table>
<thead>
<tr>
<th>T</th>
<th>Description</th>
</tr>
</thead>
<tbody>
<tr>
<td>T0</td>
<td>No evidence of primary tumor</td>
</tr>
<tr>
<td>Tis</td>
<td>Carcinoma in situ: intraepithelial invasion of lamina propria</td>
</tr>
<tr>
<td>T1</td>
<td>Tumor invades submucosa</td>
</tr>
<tr>
<td>T2</td>
<td>Tumor invades muscularis propria</td>
</tr>
<tr>
<td>T3</td>
<td>Tumor invades through muscularis propria into subserosa or into nonperitonealized pericolic or perirectal tissues</td>
</tr>
<tr>
<td>T4</td>
<td>Tumor directly invades other organs or structures and/or perforates visceral peritoneum</td>
</tr>
</tbody>
</table>

N:

<table>
<thead>
<tr>
<th>N</th>
<th>Description</th>
</tr>
</thead>
<tbody>
<tr>
<td>N0</td>
<td>No regional lymph node metastasis</td>
</tr>
<tr>
<td>N1</td>
<td>Metastasis in 1–3 regional lymph nodes</td>
</tr>
<tr>
<td>N2</td>
<td>Metastasis in 4 or more regional lymph nodes</td>
</tr>
<tr>
<td>N3</td>
<td>Distant metastasis cannot be assessed</td>
</tr>
</tbody>
</table>

M:

<table>
<thead>
<tr>
<th>M</th>
<th>Description</th>
</tr>
</thead>
<tbody>
<tr>
<td>M0</td>
<td>No distant metastasis</td>
</tr>
<tr>
<td>M1</td>
<td>Distant metastasis</td>
</tr>
</tbody>
</table>

T is used to describe the size and extent of invasion of the main tumor. Levels 1 through 4 describe the depth of tumor penetration through the bowel wall. Tis means that the tumor is “in situ” (has not gone into the bowel wall).

N is used to describe whether lymph nodes have any cancer cells and the number of lymph nodes involved. N0 means no nodes are involved, N1 when one to three nodes are involved and N2 when more than three lymph nodes are positive.

M refers to metastasis or cancer spread to other parts of the body. For example, M0 means that there is no evidence of distant metastatic disease, M1 means there is cancer spread to another site.

Table 5. Comparison of the Staging Systems

The prognosis for colorectal cancer patients depends on the extent of disease and the adequacy of the surgical procedure. Patients have a worse prognosis if the cancer has spread to lymph nodes or distant organs, has invaded blood and lymphatic vessels, or is poorly differentiated.

Follow-up Care After Surgery
1. Complete physical exam every year
2. Stool blood testing every year
3. Upper endoscopy at least every 4 years
4. a. Flexible sigmoidoscopy every 6 months for patients who still have their rectum; b. Flexible sigmoidoscopy every 6 months to 1 year for patients with ileoanal pouches
5. Follow American Cancer Society guidelines for cancer surveillance.

Dietary Considerations after Surgery
The following are suggestions for people who have a total colectomy with an ileostomy: Chew foods well, and eat slowly, Drink 6–8 glasses of liquids every day.

In the first 6 weeks after surgery avoid fibrous and stringy foods that could cause a food blockage:

- Popcorn
- Nuts
- Corn
- Mushrooms
- Celery
- Skins of fruit and vegetables
- Fresh pineapple
- Raw carrots
- Chinese vegetables
- Wild rice
- Stalks of broccoli
- Cabbage
- Coconut
- Dried fruits (raisins, etc.)
- Seeds

After 6 weeks these foods may be eaten in moderation as long as you chew well and drink fluid with this food.

When the ileostomy is closed, stool is passing via the ileal pouch and the person is ready for solid food. At this time patients should:

- Follow a low-residue diet.
- Avoid fatty foods, spicy foods, and caffeinated beverages.
- Chew food well.
- Eat 6 small meals rather than 3 regular meals (moderate amount).
- Do not skip meals, this will not stop stool output. In fact, you will probably have increased gas and more liquid, irritating the output.
- Drink 6–8 glasses of fluids per day.

Foods that may increase pouch output:

- Raw fruits and vegetables
- Leafy green vegetables
- Spicy foods
- Beer
- Chocolate
- Wine
- Caffeinated beverages
- High-fat foods
- Foods high in simple sugars (honey, candy, jellies, sweetened beverages)

Foods that may decrease output:

- Bananas
- Applesauce
- Creamy peanut butter
- Rice
- Tapioca pudding

Foods that may contribute to anal irritation:

- Spicy foods
- Foods with seeds
- Nuts
- Popcorn
- Dried fruits
- Chinese vegetables
- Raw fruits and vegetables
- Corn, coconut

Foods that may increase gas:

- Milk and milk products
- Carbonated drinks
- Onions
- Beer, beans, and the cabbage family (broccoli, Brussels sprouts, cauliflower)
- Cucumbers
- Melons
- Nuts
- Eggs

After 6 weeks, you may experiment with foods, one at a time. There are individual variations. If a particular food causes a problem, wait a few weeks and try it again.

The pouch or reservoir adapts over time. The pouch enlarges and irritability decreases. The number of stools per day decreases and they become pastier.

After a year most people find they can eat most foods. Some foods may continue to cause diarrhea, gas, and anal irritation.

Lifestyle After Surgery
Sexual function is not impaired after surgery or ileostomy. It is important that both partners understand the surgery, by talking with the surgeon or the family physician. There is usually no need for change in established sex practices or in one's capacity to enjoy sexual intercourse. It is also possible to have successful pregnancies.
However, a woman who plans to become pregnant should consult her physician before becoming pregnant. Physicians usually recommend that a woman wait about a year after a colorectal operation before becoming pregnant. This delay gives plenty of time for abdominal scars to heal soundly and for the woman’s health to return to normal. An ileostomy should not harm the baby or endanger the mother during childbirth.

Overview
In addition to the risk for colorectal cancer, other complications of this hereditary condition may occur. For example, pre-cancerous polyps may develop in other parts of the gastrointestinal system, such as the stomach and small intestine. Although most polyps that develop in the stomach and small intestine are benign, cancer may develop in them. Young children affected with FAP have a small increased risk for hepatoblastoma, a liver tumor. Tumors may also occur in the thyroid gland, adrenal gland, bile ducts, and pancreas. A type of tumor known as a desmoid can also occur in the abdomen. Thus, physicians recommend that patients continue with life-long follow-up examination for cancer prevention.