

The Johns Hopkins Guide for
Patients and Families:

Familial Adenomatous Polyposis

The Johns Hopkins Guide for
Patients and Families:

Familial Adenomatous Polyposis

**THE JOHNS HOPKINS GUIDE
FOR PATIENTS AND FAMILIES:
FAMILIAL ADENOMATOUS POLYPOSIS**

TABLE OF CONTENTS

Introduction	1
What are Polyps	2
What is Familial Adenomatous Polyposis (FAP)?	2
What is Attenuated FAP (AFAP)?.....	2
What is the Gastrointestinal Tract?	3
How is FAP Inherited?	4
DNA Test for FAP	4
Why is Early Diagnosis Important?	6
Exam Guidelines for People At Risk	6
What are the Symptoms of FAP?.....	7
Other Tumors Associated with FAP.....	7
How is FAP Diagnosed?.....	8
What is the Treatment?	9
Sexual Function and Childbirth After Surgery	9
Guidelines for Follow Up Care for People with FAP	10
Support Groups for Individuals and Families	11
Resources	12
Publications	14
Glossary	15
Appendix	18

INTRODUCTION

This booklet is written for individuals with familial adenomatous polyposis (FAP) and their families. The information provided is intended to add to, and is not a substitute for, discussions with doctors, genetic counselors, nurses, and other members of the health care team. We encourage you to read the entire booklet in the order in which it is written since each section is built on information in preceding sections. We want to emphasize the need for regularly scheduled, thorough medical examinations for persons who already have FAP. More importantly, relatives at risk for this condition need to have regular examinations beginning at an early age. Names of support groups and additional publications concerning this condition are found at the end of the booklet.

The information included in this booklet can also be found on the Johns Hopkins Hereditary Colorectal Cancer website, www.hopkins-coloncancer.org. Please visit that site to find the most current information on FAP.

WHAT ARE POLYPS?

Polyps are abnormal, mushroom-like growths. When found in the gastrointestinal tract, they occur most commonly inside the colon (large intestine, large bowel). Less often they develop in the stomach and small intestine (small bowel). Polyps vary in size from less than one-tenth of an inch to 1-2 inches. They may be so large as to block part of the intestine. In some people polyps may be inherited, while in others they are not inherited.

WHAT IS FAMILIAL ADENOMATOUS POLYPOSIS (FAP)?

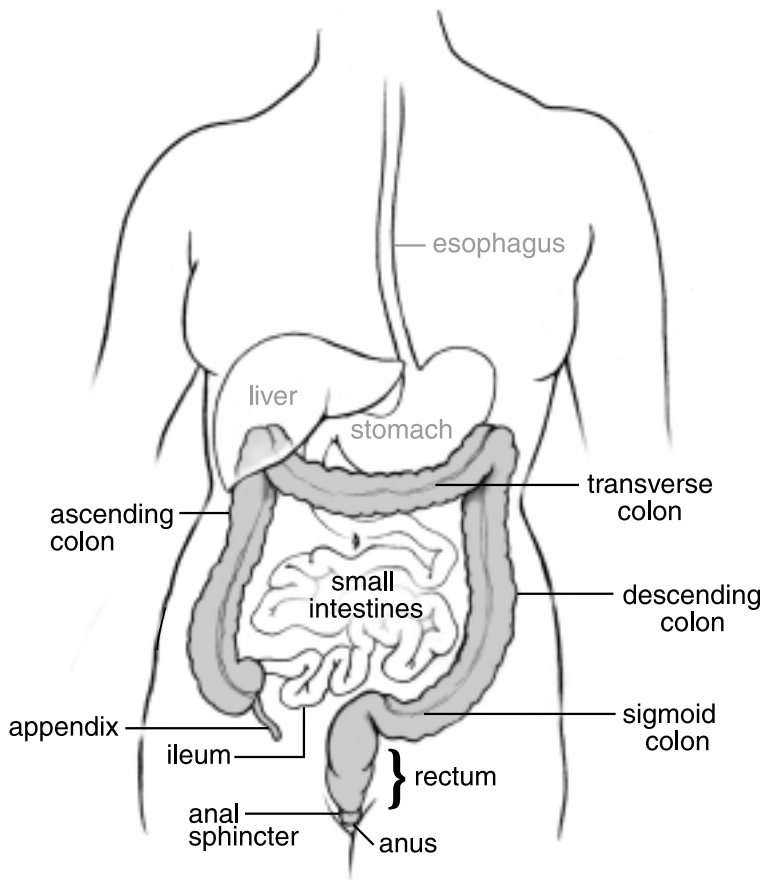
FAP is a condition in which the tendency to develop large numbers of a certain type of polyp is inherited. These polyps are called adenomatous polyps or adenomas. This hereditary (genetic) disease mainly affects the gastrointestinal tract. Other names for this condition are hereditary polyposis of the colon, familial polyposis, and Gardner syndrome. Individuals with this condition typically develop hundreds to thousands of polyps throughout the colon at a young age, usually as a teenager or young adult. The major concern in this condition is that the adenomatous polyps will become cancerous.

WHAT IS ATTENUATED FAP (AFAP)?

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, people with AFAP 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 since polyps are not evenly distributed through the colon in AFAP. 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.

WHAT IS THE GASTROINTESTINAL TRACT?

The gastrointestinal digestive tract is a hollow tube which begins at the mouth and ends at the anus. It has several parts including the esophagus, stomach, small intestine and colon (large intestine). Its total length is about 28 feet. The last 5-6 feet of the intestine is called the colon (large intestine, large bowel). The last 5 or 6 inches of the colon is the rectum. After food is digested, solid wastes move through the colon and rectum to the anus, where they are passed out of the body.



HOW IS FAP INHERITED?

FAP is an autosomal dominant condition. This means that 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 parent has had his or her own colon removed. Children who are not affected with the condition cannot pass it to their own children.

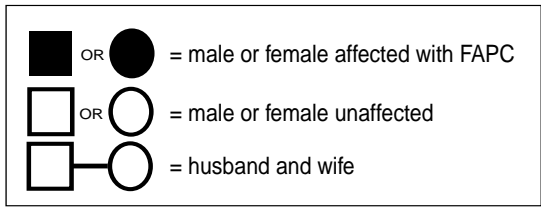
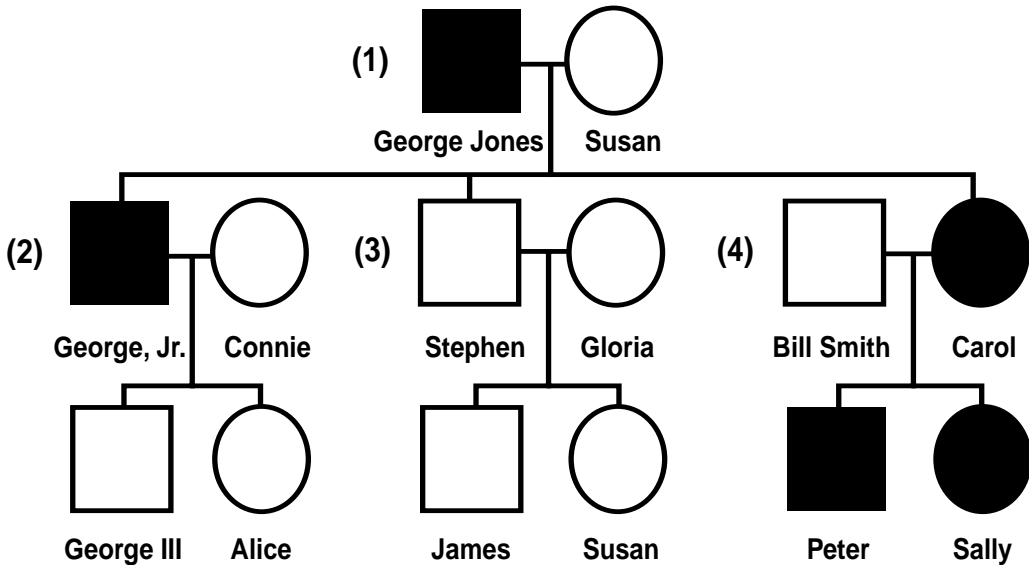
About one-third of people with FAP do not have an affected parent. These individuals, who are the first in the family to have the condition, have a new mutation of the gene. They can pass FAP to their children.

Genetic counseling is available, and recommended, for families affected with FAP. Genetic counselors are trained to explain hereditary conditions and the advantages and disadvantages of genetic testing. Counseling services are available through genetics and oncology departments in many hospitals. Also it is recommended that families with FAP contact local registries (see RESOURCES) for access to resources and for help with identification of family members at risk.

DNA TEST FOR FAMILIAL ADENOMATOUS POLYPOSIS (FAP)

FAP is caused by a mutation (change) in the Adenomatous Polyposis Coli (APC) gene. 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 the 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 on page 6.

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.



- (1) George Jones has FAP. His wife, Susan, is unaffected. They have three 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 two children. Although both children had a 50% chance of inheriting FAP, neither did.
- (3) Stephen and his wife, Gloria, have two children. Since Stephen is unaffected, there was no risk of FAP to his children.
- (4) Carol and Bill Smith have two children. Both had a 50% chance of inheriting FAP and both are affected.

WHY IS EARLY DIAGNOSIS IMPORTANT?

Early diagnosis is important for early detection and prevention of cancer. Regular screening should start as a young teenager, because when FAP is detected early, treatment will nearly always be effective. Furthermore, early and frequent examination might reassure a person without polyps that he or she does not have the disorder even though relatives do.

EXAM GUIDELINES FOR PEOPLE AT RISK

Follow the guidelines listed below if:

- You have *never had* gene testing for FAP or if a family member with FAP had DNA testing and a mutation *could not be found*.

Age	Examination
Birth - 7 yrs —————	Physical exam and/or abdominal ultrasound for liver enlargement and serum alpha-fetoprotein test every year to check for hepatoblastoma.
11-24 yrs —————	Flexible sigmoidoscopy* every year
25-34 yrs —————	Flexible sigmoidoscopy* every other year
35-50 yrs —————	Flexible sigmoidoscopy* every 3 years
over 50 yrs —————	Flexible sigmoidoscopy* every 3-5 years <i>If polyps are found, surgery will be recommended</i>

- You have had DNA testing and you *do have* a gene mutation causing FAP.

Age	Examination
Birth - 7 yrs —————	Physical exam and/or abdominal ultrasound for liver enlargement and serum alpha-fetoprotein test every year to check for hepatoblastoma.
Starting at age 11 —	Flexible sigmoidoscopy* every year <i>If polyps are found, surgery will be recommended</i>

- You have had DNA testing and you *do not have* the gene mutation causing FAP that was found in your affected family member

Age	Examination
25-30 yrs —————	To be certain that the DNA test was correct, a flexible sigmoidoscopy* should be performed at least once between the ages of 25 and 30.

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

WHAT ARE THE SYMPTOMS OF FAP?

Most patients develop polyps without symptoms. However, the following symptoms may occur:

1. Bright red blood in the stool
2. Periods of diarrhea and/or constipation not explained by diet or flu
3. Crampy pain in the stomach region
4. Pencil thin stools
5. Frequent feeling of distention (or bloating) in the abdominal or bowel region
6. Weight loss - persistent and unexplained
7. Unusual and continuing lack of energy

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

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.

OTHER TUMORS ASSOCIATED WITH FAP

In addition to the risk for colon cancer, other complications of this hereditary condition may occur. For example, precancerous 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 can occur in them. Tumors may also occur in the thyroid gland, adrenal gland, bile ducts and pancreas, while desmoid tumors can occur in the abdomen. Young children (birth to age 7) have a small increased risk for hepatoblastoma, a liver tumor. Because of the risk of these polyps and tumors, physicians recommend that patients continue with lifelong follow up examinations for cancer prevention.

HOW IS FAP DIAGNOSED?

Persons at risk for FAP initially need yearly examinations of the colon. Several tests are available to tell whether polyps are present.

1. *Flexible sigmoidoscopy* is the primary test used to examine the inside of the lower part of the colon. It is done with a lighted, flexible, hollow tube called a sigmoidoscope which is inserted into the anus. With the sigmoidoscope, the doctor can determine whether polyps or cancer are present. At the time of sigmoidoscopy, a biopsy forceps may be inserted through the inside of the sigmoidoscope to remove a bit of tissue for examination under the microscope. Most patients feel little or no discomfort during this examination.
2. *Colonoscopy* is an examination by means of a flexible, lighted tube, slightly larger in diameter than an enema tube. It permits the doctor to see much farther into the bowel than with the sigmoidoscope. Tissue may be removed from any part of the colon for microscopic study during this procedure. Before a person undergoes a colonoscopy a sedative is given; many persons sleep through the whole procedure and feel little or no discomfort. During this procedure it is sometimes necessary for the doctor to insert some air into the colon. Occasionally, air will cause the same kind of discomfort as a gas pain.
3. *Barium enema* is a test in which a white liquid called barium is inserted as an enema into the colon. This test allows the colon to be outlined when an x-ray picture is taken. If polyps are present they can be seen on the x-ray. The barium enema feels much like an ordinary enema, causing a feeling of fullness. This test should not be performed on pregnant women because of the risk of x-rays to the fetus.

For all these tests of the colon, the patient must undergo a preparation before examination. The preparation, which includes a liquid diet and laxatives, clears stool from the colon so that all areas of the colon can be inspected. Exact instructions will be provided by the doctor before the examination.

WHAT IS THE TREATMENT?

If polyps are found at examination the doctor will recommend colon surgery. Removing the colon which is full of polyps is the only way to prevent the development of colon cancer.

Several different operations are currently available for treatment of FAP. The three most commonly performed operations are: 1) total proctocolectomy with ileostomy, 2) colectomy with ileorectostomy (ileorectal anastomosis) and 3) restorative proctocolectomy (ileoanal pull-through, pouch procedure). All three 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. Illustrations of these operations can be found at our website, www.hopkins-coloncancer.org.

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 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.

SEXUAL FUNCTION AND CHILDBIRTH 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. 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 colon 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.

GUIDELINES FOR FOLLOW UP CARE FOR PEOPLE WITH FAP

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 a patient whose rectum has not been removed.
b. Flexible sigmoidoscopy every 6 months to 1 year for a patient with an ileoanal pouch
5. Follow American Cancer Society guidelines for cancer surveillance
See Appendix, page 18

In patients who have a portion of their colon or rectum remaining after surgery, adenomas can occur. An aspirin-like medicine called sulindac (trade name Clinoril) can be used to treat these polyps. *Treatment with Sulindac does not replace the need for surgery.*

SUPPORT GROUPS FOR INDIVIDUALS AND FAMILIES

1. *IMPACC*

(Intestinal Multiple Polyposis And Colorectal Cancer)

Mrs. Ann Fagan, Administrator

P.O. Box 11

Conyngham, Pennsylvania 18219

570-788-1818 or 788-3712

Fax: 570-788-4046

Email: impacc@epix.net

A support group for families with FAP and/or hereditary colon cancer.

2. *UNITED OSTOMY ASSOCIATION, INC.*

National Headquarters

19772 Macarthur Blvd., Suite 200

Irvine, California 92612

800-826-0826

The United Ostomy Association is a national organization of individuals with ostomies who work together for the benefit of all. More than 500 chapters are made up of people with ostomies whose goal is to provide mutual aid, moral support, and education to those who have had colostomy or ileostomy surgery. Individuals who have undergone this surgery may want to take the training course offered by the Ostomy Association so that they might help others who have had a similar operation. Check the telephone directory for your local chapter.

RESOURCES

1. *THE AMERICAN CANCER SOCIETY*

National Headquarters
1599 Clifton Road, N.E.
Atlanta, Georgia 30329
800-ACS-2345

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

2. *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 prior to surgery. Check your telephone directory for your state services.

3. *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.

4. *POLYPOSIS AND HEREDITARY COLON 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 contacting:

Coordinator, Hereditary Colorectal Cancer Registry
The Johns Hopkins Hospital
550 North Broadway, Suite 108
Baltimore, MD 21205-2011
Phone 888-77-COLON (772-6566), 410-955-3875, Fax 410-614-9544,
Email hccregistry@jhmi.edu.

5. *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. Telephone information may be supplemented by printed materials. All calls are kept confidential, and individuals do not need to give their name.

6. *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.

7. *INTERNET RESOURCES*

www.hopkins-coloncancer.org

The official site of the Johns Hopkins Hereditary Colorectal Cancer Registry. This site contains information on topics of interest to both patients and professionals and includes a special *Kids/Teens FAP Site*.

www.cancer.org

The American Cancer Society web site.

PUBLICATIONS

Contact the suppliers of the publications listed below for prices and/or mailing charges before ordering.

- *Ileal Pouch Pull-Through*, Lee Elton Smith, M.D. Washington Hospital Center, 110 Irving St. N.W., Suite 3B-31, Washington, D.C. 20010
- *Managing Your Ileostomy and Managing Your Colostomy* (English), *Managing Your Ostomy* (Spanish), Hollister, Inc., 2000 Hollister Drive, Libertyville, Illinois 60048.
- *The Ostomy Book*, B.D. Mullen and K.A. McGuinn. Bruce Medical Supply, 411 Waverly Oaks Road, Waltham, Massachusetts 02254.
- *Understanding DNA Testing: A Basic Guide for Families*, National Center for Education in Maternal and Child Health, 38th and R St., N.W., Washington, D.C. 20057.

The following publications, and many others, may be obtained from the United Ostomy Association, Inc., 19772 Macarthur Blvd., Suite 200, Irvine, California 92612. 800-826-0826. Send for their publication brochure for a complete list.

- *Ostomy Quarterly*.
- *Ileostomy, A Guide*, L. Gross.

GLOSSARY

Adenoma – A polyp or area of abnormal tissue that may be precancerous.

Alpha-fetoprotein – A protein produced by the liver that can be measured by the blood. Used to detect hepatoblastoma.

Anus – Outlet of the rectum.

APC – The abbreviated name of the gene (*Adenomatous Polyposis Coli*) that, when abnormal, causes FAP. It is located on chromosome 5 (at position 5q21).

At risk – A person at risk has the possibility of developing the condition which is present in his/her family.

Attenuated – Mild or less severe.

Barium enema – A test in which a chalky liquid which is resistant to x-rays is inserted into the large intestine, making it visible on x-ray film and permitting the doctor to see any defects, obstructions or masses.

Biopsy – Removal of a small bit of tissue for examination under the microscope.

CAT scan (Computerized Axial Tomography) – a form of x-ray that shows the size and shape of body organs layer by layer.

Chromosome – The carrier of the genetic material of a cell (genes). The normal number of chromosomes in a human cell is 46 (23 pairs).

CHRPE (Congenital *Hypertrophy of the Retinal Pigment Epithelium*) See **POFL**.

Clinoril – See Sulindac.

Colectomy – The surgical removal of the colon (large intestine).

Colon (Large intestine, large bowel) – The last portion of the intestine. It comprises the cecum, ascending colon, transverse colon, descending colon and sigmoid colon, and is 5-6 feet in length.

Colonoscopy – A test in which a flexible tube, about 5 feet in length, is used to examine the inside of the entire colon. The tube has a light source, a magnifying eyepiece, and an open channel through which air can be passed and a biopsy taken.

Desmoid – A benign connective tissue (fibrous) growth that may occur in the abdomen or abdominal wall. It may enlarge and may cause pressure on or encircle the bowel, stomach and other organs.

DNA (*DeoxyriboNucleic Acid*) – The molecule that contains the code for the genetic blueprint. It is found in the nucleus of cells.

Duodenum – The first part of the small intestine. It is 12-15 inches in length.

Epidermoid (epidermal) cyst – A benign cyst generally found on the face, scalp, arms, legs, and back. It is common in adults but is unusual before puberty. It is a signal for investigation of possible intestinal polyps.

Esophagogastroduodenoscopy (EGD, Upper Endoscopy) – A test in which a flexible tube is used to examine the interior of the upper GI tract (esophagus, stomach, and duodenum). The tube has a light source, a magnifying eyepiece, and an open channel through which a biopsy can be taken.

ET – Enterostomal Therapist; a specialist, often a nurse, who assists individuals who wear an external abdominal appliance to collect body waste.

Extracolonic manifestations – Lesions or abnormal growths occurring outside the large intestine in people with FAP.

FAP (Familial Adenomatous Polyposis) – An inherited disorder of the gastrointestinal tract in which there are 100 or more precancerous polyps.

Flexible sigmoidoscopy – A test in which a flexible tube about 2 1/2 feet in length is used to examine the rectum and lower part of the large bowel. The tube has a light source, a magnifying eyepiece, and an open channel through which air can be passed and a biopsy taken.

Gardner syndrome – A hereditary condition characterized by intestinal polyposis, soft-tissue tumors of the skin, jaw lesions, and other abnormalities.

GI (gastrointestinal) tract – The digestive system, consists of the esophagus, stomach, small intestine (22-25 feet in length), and large intestine (5-6 feet in length).

Gene – The basic unit of heredity, each one occupying a certain place on a chromosome.

Hemoccult test – A test using specially treated cardboard slides to check for hidden blood in the stool.

Hepatoblastoma – A tumor of the liver that occurs in infancy and early childhood.

Hereditary – genetically transmitted from parent to children.

Ileoanal pull-through (pelvic pouch procedure, ileoanal anastomosis procedure) – An operation removing the colon and the lining of the rectum, leaving the underlying anal muscles, or sphincters. The last part of the small intestine is joined to the anus and an internal pelvic pouch is created.

Ileorectal anastomosis – An operation that removes the colon and joins the last part of the small intestine (ileum) to the rectum.

Ileostomy (proctocolectomy) – An operation that removes the colon, rectum, and anus. An opening is then made from the ileum through the abdominal wall.

Ileum – The last part of the small intestine, 12-15 feet long.

Jejunum – The middle part of the small intestine, 8-10 feet long.

Karyotype – A picture of the chromosomes.

Marker – A physical abnormality that may indicate the presence of, or may predict the future occurrence of a specific disorder in an individual.

Metastasis – Spread of cancer by the lymphatics or bloodstream to other sites in the body.

Mutation – A change in a gene which may result in a specific disorder.

NSAID – Non-steroidal anti-inflammatory drugs (NSAID's) are a class of drugs which includes aspirin, ibuprofen and sulindac. They reduce pain and inflammation among other effects.

Odontoma – A group of small, unerupted teeth.

Osteoma – A bony enlargement most commonly found in the skull, jaws and limbs.

Ostomate – A person with an ileostomy (or colostomy).

Pedigree – family tree; genealogy.

POFL – (*Pigmented Ocular Fundus Lesion*). Freckle-like spots on the retina of the eye that can be seen by a special eye examination. They are harmless. If present in sufficient numbers they indicate that a person has FAP. Their absence in an individual does not indicate freedom from the disease. Also called **CHRPE**.

Propositus/Proposita – (Proband; Index case). The first individual to be identified in a family that has a specific hereditary disorder.

Stoma – Artificially created opening in the abdomen.

Sulindac (trade name Clinoril) – An anti-inflammatory aspirin-like drug.

Syndrome – A collection of abnormal physical characteristics occurring in an individual (Example: Gardner syndrome with epidermoid cysts, osteomas, and sclerotic jaw lesions, in addition to polyposis).

APPENDIX

The American Cancer Society recommends the following guidelines for early detection of cancer in people without symptoms:

Age 20-40

Cancer-related checkup every 3 years

Should include the procedures listed below plus health counseling (such as tips on quitting cigarettes) and examinations for cancers of the thyroid, testes, prostate, oral region, ovaries, skin and lymph nodes. Some people are at higher risk for certain cancers and may need to have tests more frequently.

Breast

- Exam by doctor every 3 years
- Self-exam every month

Higher risk for breast cancer: Personal or family history of breast cancer, never had children, first child after 30.

Female Reproductive System

- Pelvic exam every 1-3 years with PAP test. Includes women age 18 and over.
- Pap test – after 3 initial negative tests 1 year apart, test should be done at the discretion of the physician, includes women who are, or who have been, sexually active, or have reached age 18.

Higher risk for cervical cancer: Early age at first intercourse, multiple sex partners.

Age 40 & over *Cancer-related checkup every year*

Should include the procedures listed below plus health counseling (such as tips on quitting cigarettes) and examinations for cancers of the thyroid, testes, prostate, oral region, ovaries, skin and lymph nodes. Some people are at higher risk for certain cancers and may need to have tests more frequently.

- Breast
- Exam by doctor every year
 - Self-exam every month
 - Breast x-ray every year

Higher risk for breast cancer: Personal or family history of breast cancer, never had children, first child after 30.

- Female
Reproductive
System
- Pelvic exam every year
 - Pap test – after 3 initial negative tests 1 year apart test should be done at the discretion of the physician
 - Endometrial tissue sample at menopause if at risk

Higher risk for cervical cancer: Early age at first intercourse, multiple sex partners.

Higher risk for endometrial cancer: Infertility, obesity, failure of ovulation, abnormal uterine bleeding, estrogen therapy.

- Colon/
Rectum
- For family members *not* at risk for FAP. All exams should begin at age 50.
- Fecal occult blood test every year
 - Flexible sigmoidoscopy and digital rectal exam every 5 years, or Colonoscopy and digital rectal exam every 10 years, or Double-contrast barium enema and digital rectal exam every 5-10 years

Higher risk for colorectal cancer: Personal or family history of colon or rectal cancer, personal or family history of polyps in the colon or rectum, personal history of chronic inflammatory bowel disease.

- Prostate
- Prostate-specific antigen (PSA) every year beginning age 50
 - Digital rectal exam every year beginning age 50

Higher risk for prostate cancer: Strong family history of prostate cancer, African-American heritage.

The cancer-related checkup guidelines are not rules and only apply to people without symptoms

*If you have any of the **Seven Warning Signals** listed below,
see your doctor or go to your clinic without delay*

CANCER'S SEVEN WARNING SIGNALS

1. Change in bowel or bladder habits
2. A sore that does not heal
3. Unusual bleeding or discharge
4. Thickening or lump in breast or elsewhere
5. Indigestion or difficulty in swallowing
6. Obvious change in wart or mole
7. Nagging cough or hoarseness

ACKNOWLEDGMENTS

This booklet was prepared by the following members of the Johns Hopkins Hereditary Colorectal Cancer Program in 1988, revised 1994 and 2000: Judith Bacon, B.S., Susan Viles Booker, B.A., Jill D. Brensinger, M.S., C.G.C., Paul Celano, M.D., Donna Cox, M.Ed., Francis M. Giardiello, M.D., Stanley R. Hamilton, M.D., Linda Hyland, R.N., Karen A. Johnson, M.S., C.G.C., Anne J. Krush, M.S., L. Stefan Levin, D.D.S., M.S.D., Gloria M. Petersen, Ph.D., Kathy Potter, R.N., Katherine Romans, M.S., Elias I. Traboulsi, M.D., and John Yardley, M.D.

We would like to thank Linda M. Welch for secretarial assistance, Theresa Berk, M.S. for assistance in compiling the glossary, and Joanna Strayer Amberger, B.A. for technical assistance on the pedigree illustration, page 5., and Corinne Sandone, M.A. for the illustration on page 3.

We would also like to thank the Clayton Fund, The Johns Hopkins Oncology Center's Community Programs and the Cancer Research Foundation of America for their support in the development of this booklet.

Supported by the Mid-Atlantic Cancer Genetics Network.

To order copies of this booklet, please contact the Coordinator, Hereditary Colorectal Cancer Registry, The Johns Hopkins Hospital, 550 North Broadway, Suite 108, Baltimore, MD 21205. 888-77-COLON (772-6566), hccregistry@jhmi.edu.