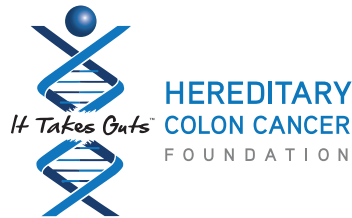


A Patient's Guide to FAP



It Takes Ours

HEREDITARY
COLON CANCER
FOUNDATION



Dear Reader,

“A Patient’s Guide to FAP” was created by the Hereditary Colon Cancer Foundation and was authored by Travis H. Bray, PhD (Hereditary Colon Cancer Foundation), Randall W. Burt, MD (Huntsman Cancer Institute) and Douglas L. Riegert-Johnson, MD (Mayo Clinic). The content is based on the National Comprehensive Cancer Network Clinical Practice Guidelines in Oncology “Genetic/Familial High-Risk Assessment: Colorectal” (2015).

This guide is intended to provide information for those affected by hereditary colon cancer syndromes and should not replace discussions or advice from your medical provider. We suggest you read this Guide in the order in which it is written, as each section builds upon information in previous sections. Medical terms in blue are explained in the glossary.

Be well and stay strong!

Travis H. Bray, PhD
Previvor, Founder, and Executive Director
Hereditary Colon Cancer Foundation

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Table of Contents

What is FAP?	4	Genetic Testing	12
Introduction	4	Decide if Genetic Testing is Right for You and Your Family	12
What is a Colon? What is the Rectum?	5	Genetic Testing Results	13
Variations of FAP	5	What are the Risks and Limitations of Genetic Testing?	13
Symptoms of Colon and Rectal Cancer	6	Genetic Counseling	14
Cancer Risks Outside of the Colon	7	What Happens During a Consultation with a Genetic Counselor?	14
Desmoid Tumors	7	Communicating Your Diagnosis with Your Family	14
Small Intestine	8	Is There a Cure?	15
Pancreatic Cancer	8	How Should I Manage FAP?	15
Thyroid Cancer	8	What's the Plan?	15
Hepatoblastoma	8	Where Should I Go?	15
Medulloblastoma	8	How Often Should I be Checked?	15
Stomach Cancers	9	If You Are Unsure If You Have FAP	15
Noncancerous Growths Outside the Intestine	9	If You Are Unsure If You Have AFAP	15
Dental Effect	9	If You Have FAP	16
Osteomas	9	Surgeries to Prevent Colorectal Cancer	18
CHRPEs	10	Chemoprevention Drugs for FAP	22
Adrenal Glands	10	What is the Average Life Expectancy?	22
Skin Effects	10	Glossary	23
How Did I Get This?	11		
How is FAP Diagnosed?	11		
Endoscopy	11		

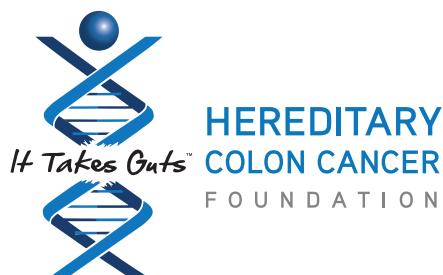


If you or your loved one has just been diagnosed with FAP:

Don't panic...

There are about 30,000 people in the United States with FAP. They go about their lives, attend school, work, play, and have families. Knowing you have FAP is the most important step! Once someone knows they have FAP, steps can be taken to prevent cancer and manage problems as they come up.

The Hereditary Colon Cancer Foundation is here to help you.



What is FAP?

Introduction

What does “FAP” stand for?

“F” is for **Familial** and means it occurs in a family.

“A” is **Adenomatous** which is another word for precancerous.

“P” is **Polyposis** and describes a large number of **polyps** in the **colon**.

Polyps are usually the first sign of FAP. **Polyps** are growths that look like small bumps of tissue. As they grow larger, **polyps** look like cherries on a stalk and can turn into cancer.

Early diagnosis of FAP can dramatically decrease the chances of developing cancer. *Without treatment*, a person with FAP can have a 100% risk of colorectal cancer. *With treatment*, the risk of colorectal cancer can be reduced to 0%!

FAP is not very common. Only about 1 in 10,000 people have FAP. Less than 1% of all **colorectal** cancer cases come from FAP. FAP is usually diagnosed between the ages of 10 and 19 through **genetic testing** or **colonoscopy**. FAP can be diagnosed during a

colonoscopy based on the presence of over 100 **polyps**. Fewer **polyps** are required for a diagnosis in young people or in a relative of someone known to have FAP. The presence of **polyps** is especially significant if the individual has a family history of FAP. Attenuated FAP can also be diagnosed during a **colonoscopy**, but usually with only 10 to 100 **polyps**.

Once someone is diagnosed with FAP, anyone in their family is at risk for having the condition. If a parent has FAP, each child has a 50% risk, or 1 in 2 chance of having FAP. Although most people with FAP inherit it from a parent, FAP does not have to be inherited. In fact, 30% of people are the first in their family to have it.

There are other cancers people with FAP need to think about as well. The risks for these cancers are not nearly as high as the risk for **colorectal** cancer. These include **thyroid** cancer (3%), small intestine cancer (5%) and liver cancer (1%).

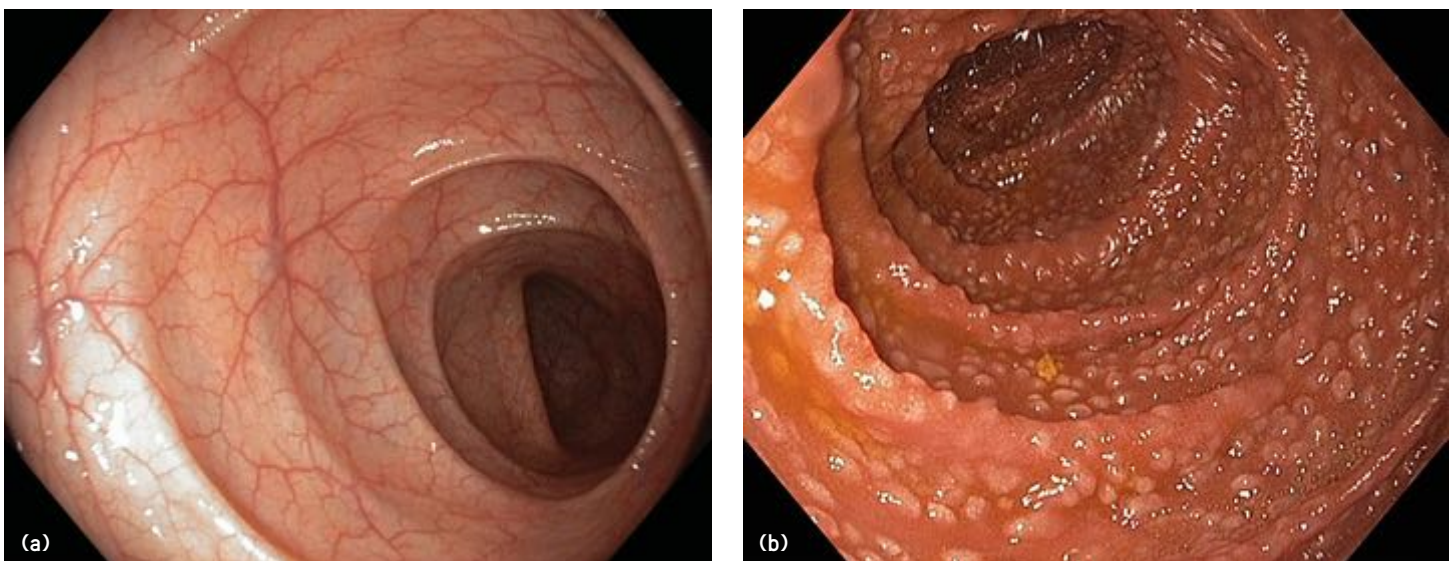


Figure 1: (a) A normal, healthy looking colon compared to (b) a colon showing severe polyposis. Photos courtesy of 2015 Gastrolab Image Library and Endoscopic Archives.

What is FAP?

What is a Colon? What is a Rectum?

The **colon** and the **rectum** are the last parts of the intestine before the **anus**. The **colon** is about 4-5 feet long in an adult and empties into the **rectum**. The **rectum** is about 5 inches long, and connects to the anus.

Many anatomy terms mean the same thing. The **colon**, large intestine, and bowel all refer to the same body part. When people say “**colon**,” they are often referring to the **colon** AND the **rectum**. When doctors and other providers say “**colon**,” they are referring to the **colon** ONLY, and NOT the **rectum**.

In this guide, we will use “colon” for **colon**, “rectum” for **rectum**, and “**colorectal**” if we are talking about both.

Variations of FAP

There are different types of FAP. When these conditions were originally described, it was not clear they were all related to FAP. They are now all known to be FAP.

Attenuated FAP (AFAP): **Attenuated** is a medical term that means ‘weakened in effect.’ So **Attenuated** FAP is a milder form of FAP. Patients with AFAP typically have 10 to 100 **colon polyps**.

“Classical” FAP: Patients with FAP have more than 100 **polyps**.

Gardner’s syndrome: In addition to **colon polyps**, patients with Gardner’s Syndrome also have features outside of the **colon**.

We avoid the term Gardner’s Syndrome because it is confusing for some doctors and people with FAP. Instead of Gardner’s Syndrome, we recommend using FAP.



Figure 2: Lower Gastrointestinal Anatomy.

“Classical” FAP (FAP)

- Diagnosed when there are over 100 **polyps** found in the **colon** and/or **rectum**.
- **Polyps** usually begin to grow between the ages of 10 and 19.
- As classical FAP progresses, hundreds to thousands of **polyps** will grow in the **colon**.
- 100% risk of **colorectal** cancer if left untreated.

Attenuated FAP (AFAP)

- Polyps start to appear at a later age, usually in the 20s or 30s.
- The risk of **colorectal** cancer is increased, but occurs later than in classical FAP. This risk may reach near 70% by age 80.
- Usually develop between 10 and 100 **polyps**.
- Other issues, such as an increased risk of **thyroid** and **duodenal cancer**, are similar to those of classical FAP.

What is FAP?

Symptoms of Colorectal Cancer

Without treatment, people with FAP have nearly a 100% chance of developing **colorectal** cancer. Do not wait for these to occur if you or a family member has FAP. **Colon** cancer can be avoided through **colonoscopies** and **genetic testing**. Screening should start before symptoms ever occur.

To be safe, one should know the symptoms of **colon** cancer. These include:

Signs of Colon Cancer

1. A change in your bowel movements, including diarrhea or constipation, that lasts more than a few days
2. Bleeding during bowel movements
3. Dark stool or blood in your stool
4. Abdominal (gut) discomfort, such as cramps, gas, or pain, that lasts more than a few days
5. A feeling that, when you need to go, you can't or you can't seem to empty it all
6. Stools that are more narrow than usual
7. Weakness or feeling very tired
8. Unexplained weight loss

Many people with **colorectal** cancer do not notice symptoms in the early stages of the disease. When symptoms do appear, they can be different depending on the cancer's size and location. This is why it is critical to follow the screening recommendations in this guide.

Information regarding cancer treatment options can be found at www.cancer.org.

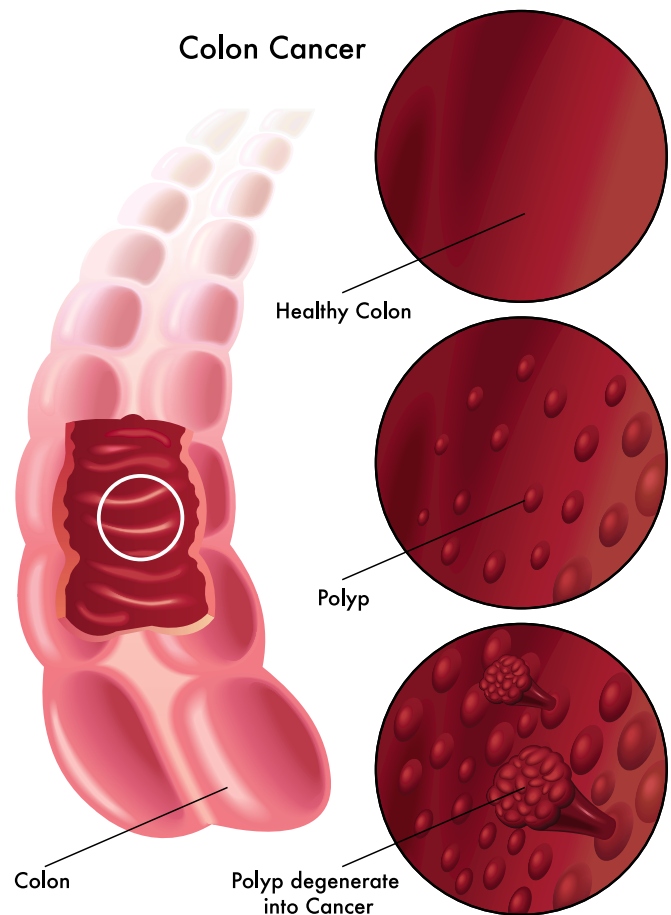


Figure 3: In a person with FAP, a healthy colon grows polyps that will eventually turn into cancer.

What is FAP?

Cancer Risks Outside of the Colon

Individuals with FAP have an increased risk of certain cancers outside of the large intestine. These include **duodenal** cancer, **hepatoblastomas**, **thyroid** cancer, and **desmoid tumors**.

Cancer Location	Chances of happening in a lifetime
Colon and rectum cancer	Almost 100% if not prevented by treatment
Desmoid tumors (this is a benign tumor but may cause problems)	10% to 20%
Small intestine (mostly in the first section called the duodenum)	4% to 12%
Pancreatic	2%
Thyroid	2% to 6%
Hepatoblastoma (a type of liver cancer)	1 to 2%
Medulloblastoma (a type of brain cancer)	Less than 1%
Stomach	About 1%
Bile duct cancer	Very small, but increased
Adrenal gland cancer	Very small, but increased

Desmoid Tumors

Some people with FAP get one or more **desmoid tumors**. These are benign tumors that form from excess scar tissue. **Desmoids** do not spread to the liver and other organs like **colorectal** cancer sometimes does. Regardless, they can cause major health problems.

Because **desmoids** can become deadly by pressing on organs, it is important to find them as early as possible. Their location is often in the abdominal area (the middle of the body) although they can occur in any area of the body. The most common health problem a **desmoid** causes is blockage of the **intestines**. Symptoms vary based on location, but the most common symptom is pain.

A person is at high risk for **desmoids** if someone else in the family has had them. The results of a **genetic test** may also suggest an increased risk for **desmoids**. **Desmoids** usually grow within five years of having colon surgery.

There are many treatments available for **desmoids**. The most common are antiinflammatory, antihormone, and **chemotherapy** pills taken by mouth. Some patients undergo **chemotherapy** by IV; some need surgery.

For more information and resources, visit the Desmoid Tumor Research Foundation at www.dtrf.org.



What is FAP?

Small Intestine

In patients with FAP, occasional checking of the small **intestine** is very important. This is especially true in the upper portion of the small **intestine** (the “**duodenum**”). The severity of **polyp** growth in the **duodenum** is determined by the number, size, and shape of the **polyps**, and how much they look like cancer under the microscope. This affects the treatment and frequency of screening.

It is very important that doctors who perform **endoscopies** on the **duodenum** also look for “**periampulla cancer**.” Be sure to ask your doctor to look for **polyps** on or near the “**ampulla**.” This is the where the **bile** and digestive fluids enter the **duodenum**.

While over 90% of patients with FAP will have **polyps** in their **duodenum**, only about 35% will have severe **polyp** growth. Severe **polyp** growth requires treatment.

If a **polyp** near the **ampulla** cannot be removed, your doctor may suggest a “Whipple” surgery. This surgery involves removing the gall bladder, part of the **duodenum**, part of the pancreas, and part of the stomach. Any patient considering a Whipple procedure should seek a second opinion from a FAP expert. Doctors who specialize in FAP may be able to remove a **polyp** by **endoscopy**.

Pancreatic Cancer

The rate of pancreatic cancer in FAP patients is not well-known, but is very low. No additional medical exams are necessary to screen for this tumor other than an annual physical exam.

Thyroid Cancer

The lifetime risk of **thyroid** cancer in FAP is 2–6%. The most common time for **thyroid** cancer to develop is around 30 years of age. Physical examination by the doctor (**palpation**) of the **thyroid** is recommended every year starting in the late teenage years. A **thyroid ultrasound** may be also be considered.

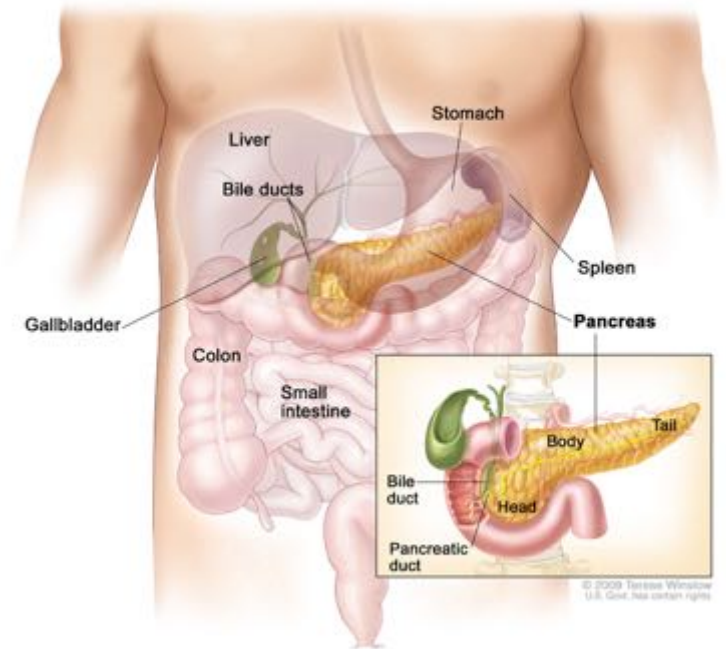


Figure 4: Anatomy of the Upper Small Intestine, Gall Bladder, Liver, and Pancreas.

Hepatoblastoma

A **Hepatoblastoma** is a type of liver cancer that occurs almost exclusively in children with FAP under 5 years old. It is very rare and only occurs in about 1.5% of FAP patients. The danger of this cancer means that active screening every 3 to 6 months until age 5 may be considered. Screening may be by liver exam (**palpation**), **ultrasound**, and/or a blood test (**AFP**).

Medulloblastoma

This is a type of brain cancer that occurs in FAP, but only very rarely. There is no screening recommended for this cancer unless someone in the family already has it. Individuals with FAP should report new headaches that do not go away.

What is FAP?

Stomach Cancers

Fundic gland polyps (FGPs) of the stomach occur in the majority of FAP patients. There are often too many to count, but it is uncommon for FGPs to cause symptoms or cancer. FGPs often worry gastroenterologists and **pathologists** who are not experts in **hereditary colon** cancer and/or gastric cancer.

An upper **endoscopy** exam is enough to check for gastric cancers such as these. It is recommended that doctors look carefully for gastric polyps that appear strange in shape or texture, or are large. These characteristics might suggest a cancer-causing polyp. It is also recommended that polyps in the very bottom of the stomach, just before its connection to the small **intestines** (an area called the “antrum”), should be removed if possible. These polyps are less common but may be cancer-causing.

From time to time, we hear about a patient being referred for removal of the stomach (**gastrectomy**). A **gastrectomy** is a very serious surgery that can reduce quality of life. Any patient considering a **gastrectomy** should first seek the opinion of an expert. Only doctors with experience treating FAP, stomach **polyps**, and/or stomach cancer should make the recommendation of a **gastrectomy**.



Noncancerous Growths Outside of the Colon

Dental Effect

Some individuals may have extra teeth or teeth that do not completely grow in.

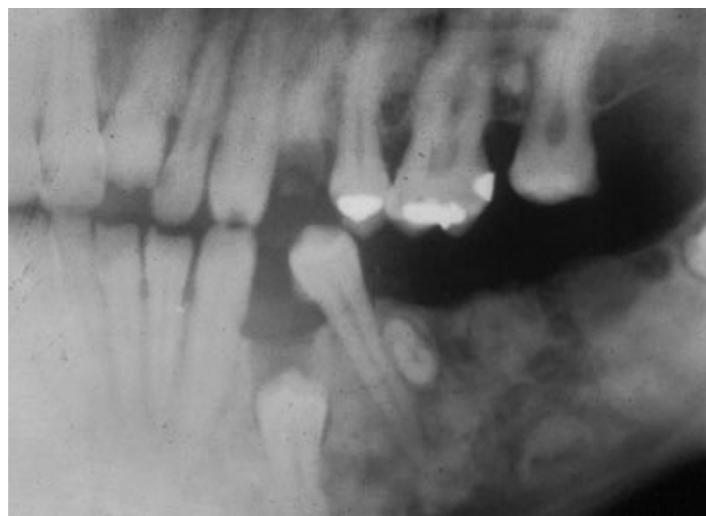


Figure 5: An X-ray of a tooth that never completely grew in due to FAP. Photo courtesy of Douglas Riegert-Johnson, MD.

Osteomas

Osteomas are bone growths that feel like hard bumps. They usually grow on the jaw or skull but may grow on any bone of the body. They are only removed if they cause problems. Otherwise, they are usually left alone.



Figure 6: Osteoma of the Jaw. Photo courtesy of Gundewar S, et al., *Indian J Plast Surg* (2013).

What is FAP?

CHRPE

A **CHRPE** (pronounced 'chirpee') is a **benign** spot in the back of eye that is sometimes related to FAP. **CHRPEs** can be seen during a doctor's examination. **CHRPEs** do not affect a person's vision, will not become cancer, and do not require treatment.

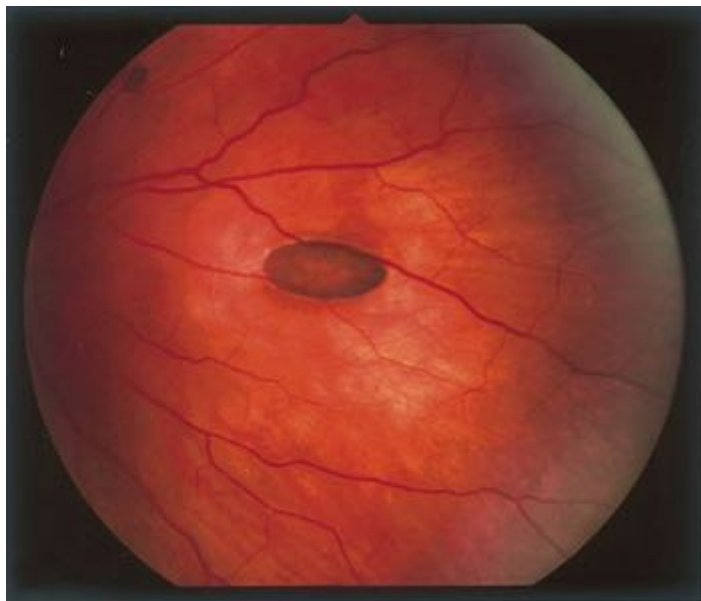


Figure 7: The view of a CHRPE as seen during an eye exam. Photo courtesy of Douglas Riegert-Johnson, MD.

Adrenal Glands

FAP patients may also develop growths on the **adrenal glands**. These growths are not cancer and do not usually need medical attention.

Skin Effects

Nearly half of FAP patients will have non-cancerous growths in the skin. These can be any of the following:

- Fat-filled sacs under the skin called sebaceous cysts or "lipomas." These are enclosed and will 'roll around' under the skin.
- Liquid-filled sacs under the skin called "epidermal cysts." These may release whitish liquid when squeezed.
- Stringy, more solid growths above the skin are called "fibromas."

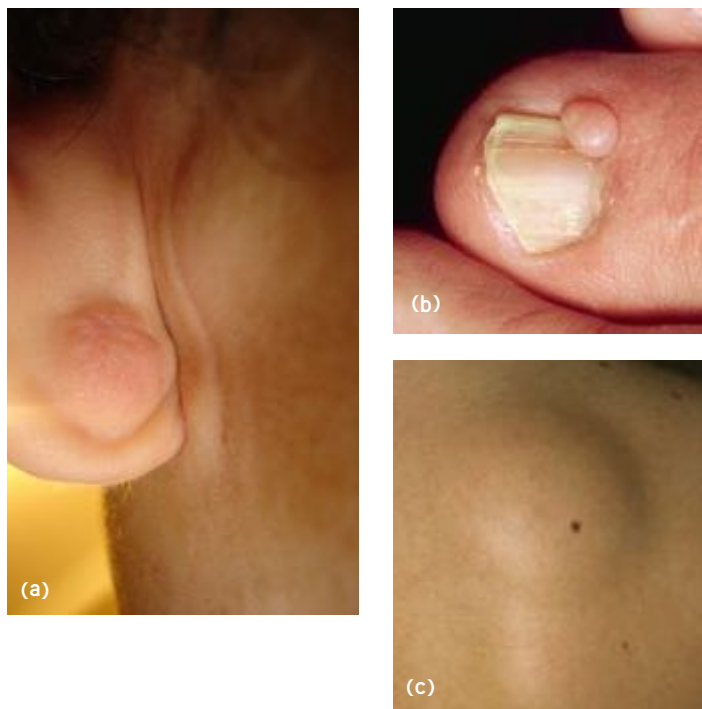


Figure 8: An image of (a) an epidermal cyst, (b) a fibroma, and (c) a lipoma. Photos courtesy of Douglas Riegert-Johnson, MD.

How Did I Get This?

FAP is caused by a change in a specific **gene** of our body. **Genes** are like a set of instructions. They provide information for cells to divide, proteins to be produced, and our body to grow and heal itself.

When a gene changes from its normal form, this is called a **mutation**. People born with a **mutation** in their APC (Adenomatous Polyposis Coli) gene have FAP. Most people who are born with FAP got it from either their mother or father. In fact, a person with FAP has a 50% chance of passing it on to each of their offspring.

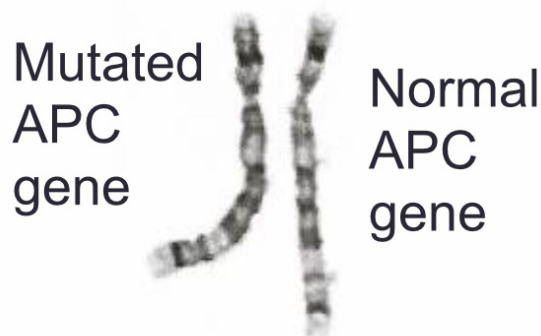


Figure 9: An image of a normal APC gene compared to a mutated APC gene that will cause FAP. Photo courtesy of Douglas Riegert-Johnson, MD.

Although most people with FAP inherited it, FAP does not have to be inherited. About 30% of individuals with FAP are the first people in their family to have it. These people are referred to as *de novo* (meaning “from the beginning”). For them, the gene **mutation** happened at the time of conception.

How is FAP Diagnosed?

Endoscopy

Polyyps are usually the first sign of FAP. FAP can be diagnosed during a **colonoscopy** based on the presence of over 100 **polyyps**. Fewer **polyyps** are required for a diagnosis in young people or in a relative of someone known to have FAP. The presence of **polyyps** is especially significant if the individual has a family history of FAP.

AFAP can also be diagnosed during a **colonoscopy** but usually with only 10–100 polyyps.

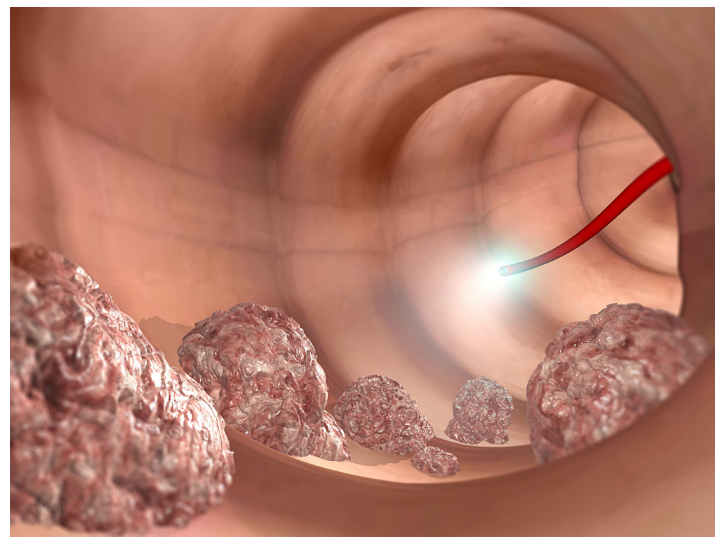


Figure 10: Illustration of a colonoscopy looking for polyyps.

How is FAP Diagnosed?

Genetic Testing

The goal of genetic testing is to avoid cancer. If there is a chance you have FAP, **genetic testing** may confirm it and help you learn more about your risk of cancers. Family members may avoid unnecessary screening if **genetic testing** confirms that they do not have FAP.

Using a **genetic test** to confirm you have FAP helps you know what to expect. It can help you and your doctors make better decisions about your health care. It can guide you and your health care team in creating an individualized prevention, screening, and treatment plan.

The DNA needed for a genetic test is most easily obtained from blood, a mouth rinse or an inner cheek swab. The blood or mouth swab is sent to a laboratory where scientists analyze your DNA to see if you have FAP. Depending on the laboratory, this process can take 2-12 weeks. The **genetic test** results are sent in writing to your doctor or **genetic counselor**, who will contact you to discuss the results.

There are many different **genetic tests**. We recommend you work with a **genetic counselor** to identify the best **genetic test** for you.

Decide if Genetic Testing is Right for You and Your Family

Genetic testing is a personal choice. Before you have a **genetic test**, you should have the opportunity to understand the testing procedure, benefits and limitations, and possible consequences of the results. The process of learning about the test and giving your permission is called 'informed consent.' **Genetic testing** is often done as part of a genetic consultation. **Genetic counselors** can answer your questions as well as help you prepare for the possible results.

Children with FAP can begin developing **polyps** by their early to mid-teens. So, **genetic testing** is typically offered around the age of 10 and up.

If you decide to proceed with **genetic testing**, the following people can order a **genetic test**:

- Gastroenterologists (GI)
- Medical geneticists
- Nurse practitioners
- Oncologists
- Primary care doctors
- Surgeons



How is FAP Diagnosed?

What are the risks and limitations of genetic testing?

Before you have a **genetic test**, a knowledgeable healthcare provider should explain the risks and limitations of genetic testing. Most of the risks associated with **genetic testing** involve emotional, social, and/or financial consequences of the test results. The risks include:

- **Emotional Risks:** Undergoing **genetic testing** may significantly alter someone’s self-identity. Emotional risks include feelings of isolation, anger, depression, anxiety, or guilt. Emotional risks are not limited to those who discover they have FAP. Other family members who discover they do not have the condition may also be affected.
- **Family Dynamics:** **Genetic testing** may create tension within a family. The results can reveal information about other family members.
- **Genetic Discrimination:** Fear of genetic discrimination is a common concern. The Genetic Information Nondiscrimination Act (GINA) is a federal law that protects people from genetic

discrimination. Genetic information includes **genetic test** results and family history information. GINA has two parts:

Title I says that health insurance providers:

- Cannot use the fact that you have FAP to determine eligibility or to set rates.
- Cannot require you or a family member to take a **genetic test**.

Title II prohibits genetic discrimination in employment and says:

- Genetic information may not be used for hiring, firing, promoting, determining pay or assigning jobs.
- Employers cannot require you to take a **genetic test**.

There are important limitations to this law. GINA does not apply to:

- Small businesses (fewer than 15 employees),
- The Federal Employees Health Benefits Program,
- The U.S. military / TRICARE,
- The Veterans Health Administration,
- The Indian Health Service,
- Other types of insurance including life, disability, or long-term care insurance.

Negative Result (-)	→ If the gene mutation has already been identified in the family.	→ The person does not have FAP. People who do not have FAP can avoid unnecessary screenings like colonoscopies .
	→ If the gene mutation has NOT already been identified in the family.	→ The presence of FAP could not be confirmed at this time. Routine screening should continue as advised by doctor. As research and technologies improve, scientists are sometimes able to identify mutations that were not previously found using older technologies.
Uncertain Result (*)	An uncertain result occurs because the genetic test reported a “variant of uncertain significance” (VUS). A VUS is a mutation that may or may not cause a problem. As more research is done, scientists may determine if this genetic change is harmful or not. A VUS does not confirm or deny FAP. A VUS should NOT be used to make medical decisions.	
Positive Result (+)	FAP was identified. This clarifies your cancer risks, helps your doctor manage those risks, and allows for testing of family members. Once a family member has a positive result, other family members can be tested for FAP. This can help make sure family members with FAP can get screening and/or surgeries in time to prevent cancers.	

How is FAP Diagnosed?

Genetic counseling

Genetic counselors can help diagnose FAP. They are experts at explaining it in a way that people without a background in medicine can easily understand. These are some common questions that are addressed in a **genetic counseling** appointment:

“FAP runs in my family. What does this mean for me?”

“I’ve had cancer. What does that mean for my children? What does this mean about future children?”

“My partner and I are planning a pregnancy. What types of testing are available to us?”

“Could I pass FAP onto my children?”

“Would genetic testing be useful for me or my family?”

“What can I do to manage my own care?”

“How do I talk to my relatives or my doctors about FAP?”

What Happens During a Consultation with a Genetic Counselor?

Genetic counseling can be provided to an individual, couple, or family. Appointments are usually in person and can take place in a clinic or doctor’s office. During your appointment, the **genetic counselor** may:

- Review your personal and family medical history
- Identify possible cancer risks and discuss strategies to prevent cancer
- Review **genetic testing** options
- Provide FAP-related information and resources
- Discuss family planning

You may speak with a **genetic counselor** once or work with your counselor over time. Meeting with a genetic counselor does not always mean that you need to proceed with **genetic testing** or that **genetic testing** will be recommended.

Communicating Your Diagnosis with Family

Finding out that you have FAP can be life saving information for your family members. If your relatives are aware that FAP runs in the family, they have the opportunity to see if they also have it. This allows them to take steps to prevent cancer. However, family dynamics and the complexity of genetic information can make it hard to communicate. Below are three excellent resources that can help you prepare for these conversations:

- **www.kintalk.org** is a service of the University of California
- **www.nsgc.org** is the website of the National Society of Genetic Counselors

hccf.support/TipsforChildren, created by Dana Farber Cancer Institute, provides excellent tips for explaining hereditary cancer to children. There is no cure for FAP. The good news, though, is that FAP is an ‘actionable’ disease. This means it can be managed when found and treated early. Someone with FAP can expect a normal, or nearly normal, lifespan IF they receive care from physicians with experience treating FAP.

Is There a Cure?

An expert care team can put together a care plan just for you. This can lead to a high quality of life. So while there may not be a cure, there is reason to have hope. Remember, in some cases, cancer risk can be reduced from 100% to 0%!

How Should I Manage FAP?

What's the Plan?

A doctor who specializes in **hereditary colon** cancers should make a care plan for FAP patients. When possible, this should be done at a high risk cancer clinic. The care plan should start as soon as FAP is diagnosed or suspected.

The care plan usually starts with **colonoscopies** to look for **polyps**. When **polyps** start growing too fast to be removed during a **colonoscopy**, removal of the **colon** (**colectomy**) is advised. Surgical decisions depend on a number of factors. It is important to seek advice from a surgeon trained in FAP.

A care plan does not stop once the **colon** has been removed. Exams of the small **intestine**, pouch, **rectum**, **thyroid**, and other areas outside of the **colon** are very important throughout an FAP patient's life. It is through these checkups that cancer and other problems can be prevented.

Where Should I Go?

High risk cancer clinics find those who are likely to get cancer and help reduce their risk. This includes people diagnosed with FAP, have a family history of FAP, or have a strong family history of cancer. Someone is considered high risk if they have a family member diagnosed with cancer under the age of 50 or two or more relatives diagnosed with any cancer on the same side of the family.

High risk cancer clinics usually include one or more gastroenterologists, **genetic counselors**, **oncologists**, and surgeons. An **endocrinologist**, gynecologist, and pediatrician (child expert) may also be part of their team. Some of these clinics have a registry of other patients with FAP. There are many benefits of joining a clinical registry, including the opportunity to participate in trials of new treatments.

How Often Should I be Checked?

For a person who might have FAP, the screening and care plan depends on:

- Whether the family's FAP **mutation** is known through genetic testing, and
- Whether or not a person has had a **colectomy**.

If You Are Unsure If You Have FAP

Those born into a family with FAP are at risk of having FAP. **Polyps** often develop in the teen years. Yearly **colonoscopies** will usually detect FAP early and help prevent cancer.

Colonoscopies should start at age 10 to 12 years and continue yearly. Once **polyps** are found, **colonoscopies** should continue until **colectomy** is performed. Depending on the severity of the **polyp** growth, **colonoscopies** may be more frequent.

Because most people with FAP begin to grow **polyps** before the age of 25, if **polyps** are not found before this age, **colonoscopies** may be reduced to:

- Every 2 years until age 34, then
- Every 3 years until age 44, and then
- Every 3 to 5 years after that.

If You Are Unsure If You Have AFAP

Those born into a family with AFAP are at risk of having AFAP. Symptoms of AFAP may appear at a later age than in those with classical FAP. Polyps may range from 10 to 100. **Colonoscopies** should begin in the late teens and be repeated every 2-3 years, or more frequently if advised by your doctor. Once **polyps** are found, **colonoscopies** should continue until **colectomy** is performed.

How Should I Manage FAP?

If You Have FAP/AFAP

Screening options may change over time as new technologies grow and more is learned about FAP. It is important to talk with your doctor about the most up-to-date screening tests. Below are the guidelines as of 2015.

Body Part	Frequency
Colon & Rectum (Before Colectomy)	• Colonoscopy every one to two years, starting at: - FAP: age 10 to 12 - AFAP: late teens • Colonoscopy every year once polyps are found until colectomy • When polyps can no longer be managed by removal during a colonoscopy, surgery is advised. Your doctor will consider colectomy when: - there are more than 20 polyps; - a polyp is greater than, or equal to, 1 cm across; or - the threat is high that polyps will soon turn into cancer
Colon & Rectum (After Colectomy)	After colectomy, regular endoscopies are needed to check for polyps: • IRA: Rectum examined every 6 to 12 months. • J-pouch or ileostomy examined every 1 to 3 years (Every 6 months if polyps are found that are at high-risk of growing into cancer.)
Important Considerations for the Colon and Rectum: During an endoscopy, a physician may use treatments such as polypectomy to prevent or delay the need for surgery. Chemoprevention may also be an option.	
Desmoid Tumors	Abdominal palpation (manual examination) during yearly physical exams is advised to check for desmoid tumors. Patients with a family history of desmoids, or who have a type of FAP that increases the risk for desmoids, should have a CT scan or MRI of the abdomen: • Immediately if there are symptoms of a desmoid • Every 1 to 3 years after colectomy and, if no desmoids are found, every 5 to 10 years
Important Considerations for Desmoids: Most desmoid tumors show up within five years of colectomy. Because desmoid tumors can cause significant problems, it is vital to detect them early. For more information and resources, visit the Desmoid Tumor Research Foundation (www.dtrf.org).	

How Should I Manage FAP?

Body Part	Frequency
Small Intestine (duodenum)	Upper endoscopy should begin at age 25 to 30 <i>OR</i> when colorectal polyps are found, whichever occurs first: How often an upper endoscopy is needed depends on: - the number of polyps and - how quickly they may turn into cancer. This will range from every 4 years if no polyps are found to 3 to 6 months for severe polyp growth.
Important Considerations for the Small Intestine: Checking the small intestine is very important, especially in the first part of the small intestine (duodenum). Doctors perform an endoscopy to look for polyps. They monitor the number of polyps, and their size and shape, to see if they may turn into cancer or are cancerous. When getting an endoscopy of the duodenum, it is very important to look for polyps or cancer on or near the ampulla. A “side-viewing” scope will be necessary for this exam. You may need to speak with a surgeon and/or be examined by an expert every 3 to 6 months if you have: - a large number of polyps, - polyps that may quickly turn into cancer (usually judged by appearance, size and appearance under the microscope), and/or - aggressive cancer. During an endoscopy, a physician may use treatments such as a polypectomy to prevent or delay the need for surgery. Chemoprevention may also be an option.	
Thyroid	- Yearly palpation is recommended every year starting in the late teenage years. Ultrasounds may be considered if any possible signs of cancer are seen. - Exams may be less often after age 30 if no cancer is found.
Important Considerations for the Thyroid: The lifetime risk of thyroid cancer is about 2–6%. The most common time for thyroid cancer to develop is around 30 years of age.	

How Should I Manage FAP?

Body Part	Frequency
Liver	Palpation, ultrasound, and/or AFP blood tests every 3 to 6 months during the first five years of life may be considered.
Important Considerations for the Liver: The risk of liver cancer is very small (1.5%) and only occurs in infants under 5 years old. But because it is dangerous if it does occur, it is important to discuss these recommendations with your doctor.	
Medulloblastoma (Brain Cancer)	No screening other than an annual physical exam is called for.
Pancreas	No screening other than an annual physical exam is called for.
Stomach	An upper endoscopy can be used to check for stomach cancers and to look for FGPs that might turn into cancer. Screen at same time as the small intestine.
Important Considerations for the Stomach: Fundic gland polyps (FGPs) grow in the stomach of most FAP and AFAP patients. It is rare for FGPs to cause cancer.	

Surgeries to Prevent Colorectal Cancer

In choosing which surgery is best, the two most important things doctors will consider are:

- The number of polyps in the rectum, and
- If colon or rectal cancer is present.

The colectomy or proctocolectomy is often performed once polyps begin to grow but can be delayed depending on:

- family genetics and how FAP has behaved in other family members
- number of polyps and how they look
- experience and preferences of the doctor
- the individual's current situation (pregnancy, scar tissue, tumors, etc)

Once polyps begin to grow, an annual colonoscopy is essential until a colectomy is performed.

Several surgical options are available. Each of these will be discussed in detail on the following pages.



How Should I Manage FAP?

Surgeries to Prevent Colorectal Cancer

1. J-Pouch (Total Proctocolectomy with Ileal Pouch Anal Anastomosis or TPC/IPAA)

The most common surgery is a **colectomy** and creation of a **J-pouch**. The patient's large **intestine** (**colon**) and **rectum** (all or a part of) are removed. A small pouch is then created, using the lower section of the small **intestines** (the **ileum**). This pouch replaces the **rectum**. The bottom of the **J-pouch** is connected to the anal canal and the pouch looks like the letter 'J'.

Patients who get this procedure sometimes spend a short period of time (about three months) with a temporary **ileostomy**. This allows waste to leave the body through a stoma (or opening) into a bag while the surgical area is healing.

Additional information about living with a **J-pouch** can be found at www.j-pouch.org.

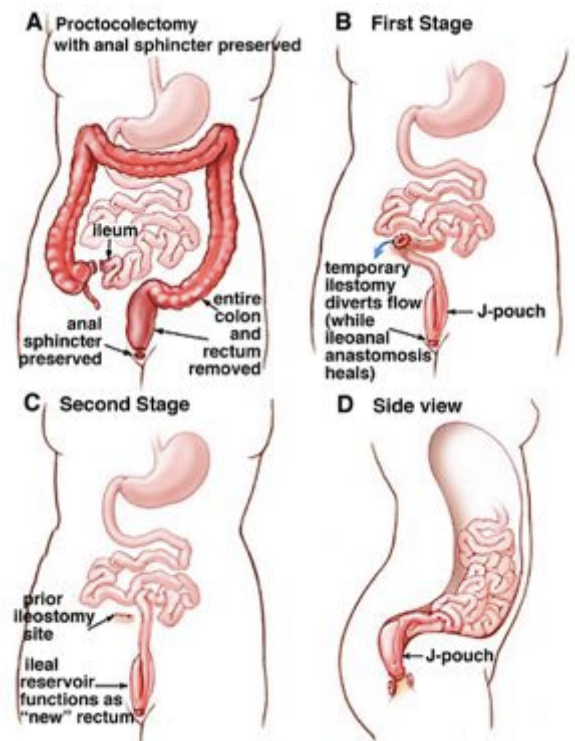


Figure 11: Illustration of how a J-Pouch is created. Image courtesy of Johns Hopkins Health Library.

A J-pouch is an option for:	A J-pouch is not ideal for:
• Patients with FAP • Patients with AFAP with polyps that cover the surface of the rectum • Patients with FAP/AFAP who have curable colorectal cancer • Patients with an IRA (discussed below) and now have polyps that cannot be treated during endoscopy	• Patients with cancer that cannot be cured • Patients with a desmoid tumor that may prevent surgery • Patients who have a condition that would block the setup of a J-pouch
What are the advantages to having a J-pouch? • The risk of rectal cancer is extremely small. • A permanent ileostomy is not needed. The patient does not have to wear a bag outside the body.	
What are concerns about having a J-pouch? • It is a long and complex operation, so there are more risks. • A temporary ileostomy is usually needed. • There is a small risk of bladder and sexual problems after the rectum is removed. • Bowel function will vary. Soon after surgery is complete, 8-10 bowel movements per day is common. As the the body becomes accustomed to the J-pouch, bowel movements usually go down to 5-6 per day or even fewer. Diet will affect the frequency and consistency of bowel movements.	

How Should I Manage FAP?

Surgeries to Prevent Colorectal Cancer

2. IRA (Total Abdominal Colectomy with Ileorectal Anastomosis)

The first step to make an IRA is a **colectomy**, with the **rectum** left intact. The last part of the small **intestine** (the ileum section) and **rectum** are then joined together. Where they meet is called an anastomosis (space where two parts meet).

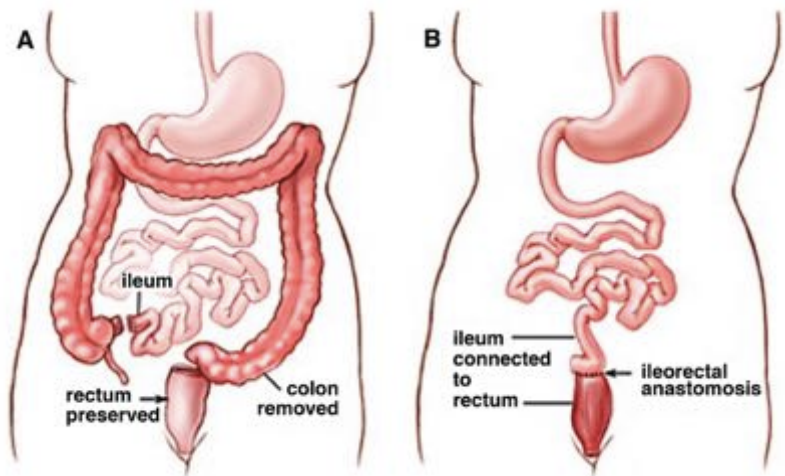


Figure 13: Illustration of how an IRA is created. Image courtesy of Johns Hopkins Health Library.

An IRA is an option for:	An IRA is not ideal for:
• Patients whose rectal polyps can be managed during endoscopies • Patients who can keep appointments for follow-up exams • For most AFAP patients, this option is best.	• Patients with rectal cancer that can be cured • Patients with a lot of polyp growth in the colon and/or rectum
What are the advantages to having an IRA? • It is a fairly clear-cut surgery. • It can be done in one step. • It does not require the patient to have a short-term ileostomy (with bag). • It has a low risk of bladder or sexual function problems. • Bowel function will vary. Soon after surgery is complete, 4-6 bowel movements per day is common. As the body heals, bowel movements usually go down to 3-4 per day. Diet will affect the frequency and consistency of bowel movements.	
What are concerns about an IRA surgery? • It is possible that the rectum might need to be removed later if polyps start growing fast. • There is a higher risk for rectal cancer (compared to the J-pouch), especially if regular rectal examinations are not done.	

How Should I Manage FAP?

Surgeries to Prevent Colorectal Cancer

3. Ileostomy (Total Proctocolectomy with Permanent End Ileostomy or TPC/EI)

The first step to make an **ileostomy** is a **proctocolectomy**. The patient's **colon** and **rectum** are removed. Next, a hole is made in the **abdomen** (**stoma**) and a small loop of the small **intestines** is pulled through (**ileostomy**). The body's waste is collected in an '**ostomy**' bag that covers the **stoma**.

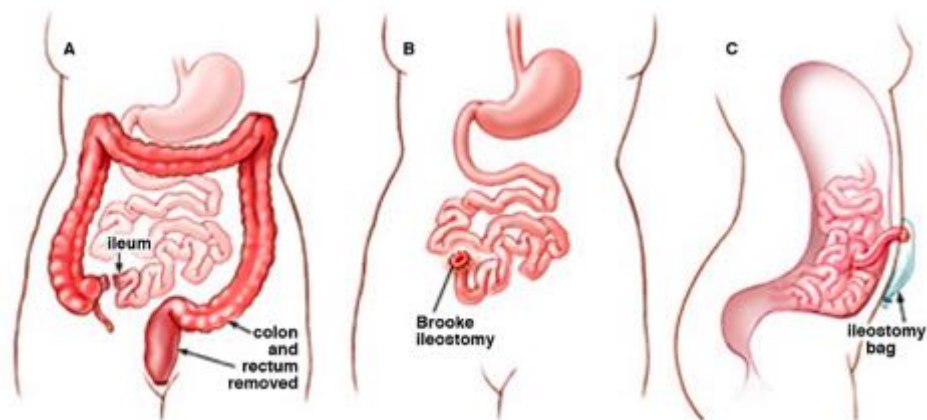


Figure 13: Illustration of how an Ileostomy is created. Image ourtesy of Johns Hopkins Health Library.

Ileostomy is an option for:	Ileostomy is not ideal for:
- Patients with early rectal cancer - Patients who cannot have a J-pouch due to a desmoid tumor - Patients with a J-pouch that is no longer functioning properly - Patients who have a condition that prevents making a J-pouch	With the option of a J-pouch or IRA, an ileostomy is rarely needed. An ileostomy may be necessary for the issues listed to the left.
What are the good things about having an Ileostomy? - It is a fairly clear-cut surgery. - It can be done in one step. - It removes all risk for colorectal cancer. - Only occasional examinations of the small bowel are needed through the stoma to be sure polyps are not growing there.	
What are concerns about having an Ileostomy? - There is more chance of bladder or sexual problems. Ostomy supplies will need to be cleaned and changed regularly.	

How Should I Manage FAP?

Chemoprevention Drugs for FAP and AFAP

Unfortunately, there are no drugs that permanently stop the growth of **polyps** in those with FAP or AFAP.

However, there are drugs that doctors may prescribe to slow the growth of **polyps** and delay the need for **colectomy**. The most common is Sulindac. EPA (Eicosapentaenoic Acid) fish oil is sometimes prescribed to reduce the number and size of **polyps** in the **rectum**.

NSAIDs (nonsteroidal anti-inflammatory drugs) are not good enough to prevent a **colectomy**, but may delay it. However, there are several clinical trials happening today that may have useful results. NSAIDs are most commonly used to suppress **polyps** in a **rectum** after surgery if many **polyps** occur. This makes it easier for doctors to manage rectal **polyps**.

What is the Average Life Expectancy?

What is the Average Life Expectancy?

Having Familial Adenomatous Polyposis increases your risk for a variety of cancers, most notably **colorectal** cancer. It is recommended that individuals with FAP work with experts to develop a comprehensive care plan. Doing so will greatly reduce the risk of cancers related to FAP. Although FAP can become life-threatening, proper screening and treatment greatly reduces cancer risk.

If left untreated, the average age of **colorectal** cancer is 39 years old for FAP and 55 for AFAP. According to the Cleveland Clinic website, "With prompt treatment, FAP patients lead normal, healthy lives. Untreated, FAP will lead to colon cancer."

Dr. Randall Burt, MD, gastroenterology emeritus professor and clinical investigator at the Huntsman Cancer Institute, adds "With the proper screening and treatment, a person with FAP can live well into their 70's or 80's."



Glossary

Abdomen— the part of the body containing the digestive organs; the belly

Abdominal Colectomy— removal of only the large intestine, not the rectum or small intestines

Adenomatous Polyp— a tumor that, in people with FAP, can become cancerous and spread if not removed; these grow out of the lining of the colon and rectum, as well as a small part in the top of the small-intestine

Adrenal Gland—one of a pair of glands, located above the kidneys, which produces steroidal hormones, epinephrine, and norepinephrine

AFP (Alpha-Fetoprotein)—a protein used to predict the possible presence of liver cancer, such as a hepatoblastoma

Ampulla (of Vater)—in the small intestine, the ampulla is the area where the small intestine is connected to the pancreas, liver, and gall bladder

Anastomosis— a connection between the small intestine and J-pouch or rectum

Anus—the opening at the end of the large intestine where waste leaves the body

Attenuated—weakened in effect

Benign—a tumor that is not at risk of spreading throughout the body

Bile—liquid produced by the liver that aids in absorption and digestion, especially of fats

Bile Duct— large duct that transports bile from the liver to the duodenum

Biopsy— procedure where the doctor takes a sample of tissue from the body in order to examine it more closely

Chemoprevention— use of a medication to prevent disease or infection

Chemotherapy— the use of medication to treat diseases such as cancer

CHRPE— “Congenital Hypertrophy of the Retinal Pigmentation Epithelial”— a non-cancerous spot found on the back of the eye; CHRPE’s are a possible symptom of FAP

Colectomy— removal of all or part of the large intestine

Colon— large intestine

Colonoscopy— visual exam of the inside of the colon with a flexible, lighted tube inserted through the rectum

Colorectal— relating to the colon and rectum

Colorectal Cancer— cancer of the colon and/or rectum

CAT Scan (CT Scan or Computed Tomography Scan)— combines many X-ray pictures of an object from different angles so doctors can see inside an object without cutting

Desmoid tumors—tumors that can grow in almost any part of the body; desmoid tumors can be very aggressive and dangerous; visit the Desmoid Tumor Research Foundation (www.dtrf.org) for more information and resources

Duodenum— the first part of the small intestine

Duodenal cancer— cancer in the duodenum

Glossary

Endocrinologist— a doctor who specializes in the glands and hormones of the body; an endocrinologist can find and treat illnesses in organs like the thyroid

Endoscope— literally means “to look inside”; this tube-shaped tool allows doctors to look inside parts of the body; for people with FAP, an endoscope normally has a light, camera, and small tools to remove polyps and other tissue

Endoscopy— using an endoscope to look inside parts of the body, such as the stomach, small intestine, or large intestine; Learn more about what to expect when having an endoscopy at www.cancer.org.

Familial— traits that are passed from one family member (mother or father) to their child

Fundic glands— glands in the top section of the stomach that produce certain acids that help with digestion

Fundic polyp— a polyp that forms in the top part of the stomach. People with FAP do NOT have a higher risk of stomach cancer due to fundic polyps than anyone else

Gastrectomy— removal of the stomach, sometimes INCORRECTLY recommended due to the presence of fundic polyps

Genetic testing— used to look for possible genetic diseases, or mutated genes that mean an increased risk of developing a genetic disorder such as FAP

Genetic counseling— when patients or relatives at risk for an inherited disorder are advised about the disorder, the chance of getting or spreading it, and their options for dealing with it

Hepatoblastoma— a type of liver cancer in infants and children

Hereditary— illnesses or characteristics passed from parents to children through their genes

Ileal— related to the ileum

Ileostomy— An opening in the ileum that is pulled through an opening in the abdominal wall that allows waste to leave the body

Ileum— the bottom section of the small intestines; it is used to create the j-pouch, connected directly to the rectum in an IRA, or pulled through the stoma in an ileostomy

Intestine— the intestines are a long (nearly 20 feet long!), continuous tube running from the stomach to the anus; the intestines absorb most of our nutrients and water; the intestine consist of the small intestine, large intestine (colon), and rectum

IRA— Total Abdominal Colectomy with Ileorectal Anastomosis (TAC/IRA)

J-pouch— an internal reservoir (storage area) surgically made to temporarily store stool waste when the large intestine has been removed from a patient

Medulloblastoma— a type of brain cancer in young children that effects the back, bottom part of the brain

MRI— a very common tool that uses magnets and radio waves to look for and keep an eye on certain diseases

Mutation— when a gene changes from its normal form

Oncologist— doctor who specializes in finding and treating cancer

Ostomy— A surgical opening in the abdominal wall that allows waste from the bowel or urinary tract to leave the body

Glossary

Palpation— a way to look for disease by pressing on an area of the body; used to feel an object in, or on, the body and determine its size, shape, firmness, or location

Pathologist— a doctor who is an expert at identifying diseases, usually by observing or testing tissue from our body

Periampulla— in the small intestine, this is the area surrounding the ampulla

Periampulla cancer— cancer that forms in the area around the ampulla; for people with FAP, this area is examined by a gastroenterologist and requires a “side-viewing scope”

Polyp— a growth from a mucous surface that can become a cancer tumor

Polyposis— the growth of very large numbers of polyps

Proctocolectomy— the complete removal of the colon and rectum

Prophylactic— a treatment or medication designed to prevent the effects of a disease; for people with FAP, a “prophylactic colectomy” means removing all, or part of, the colon and/or rectum

Polypectomy— removal of a polyp

Rectal— relating to the rectum

Rectum— the final part of the large intestine

Sigmoid— the last part of the colon before the rectum

Sigmoidoscopy— an endoscopy that looks at the rectum and the sigmoid

Stoma— Opening from inside the abdomen to the outside of the body

Thyroid— butterfly-shaped organ in the neck that controls how the body uses energy, makes proteins, and controls how sensitive the body is to certain hormones

Ultrasound— a test where a machine sends sound waves to see what is going on inside the body; an ultrasound can see the size, shape, and consistency of tissues and organs