Familial Adenomatous Polyposis (FAP)

Familial adenomatous polyposis (FAP) is a genetic condition that affects the gastrointestinal (GI) tract. FAP causes many abnormal growths (polyps) to develop in the GI tract (See Figure 1).

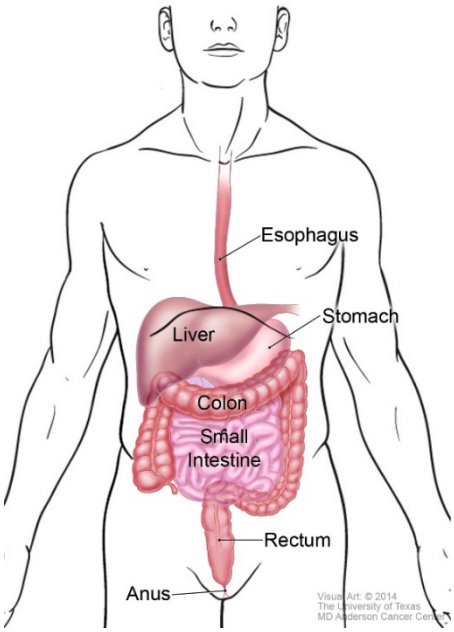


Figure 1
Organs in the Gastrointestinal (GI) Tract

Polyps are common, even in people without FAP. People with FAP often develop a higher number of polyps and tend to get them at a younger age, such as in their teen or young adult years. People with FAP can develop hundreds to thousands of polyps.

Most often, FAP causes polyps in the large intestine (colon and rectum). The most common type of polyps that people with FAP develop in the large intestine are called adenomas. Adenomas are pre-cancerous growths that increase the risk of cancer in the large intestine. Polyps may also develop in the stomach and small intestine.

FAP is caused by changes (mutations) in the APC gene. A mutation causes the gene to stop working normally. The APC gene normally works to protect the colon from polyps and cancer. The APC gene mutation that causes FAP makes it so that the body does not protect the colon and causes polyps to develop.

Attenuated FAP (AFAP) is a milder form of FAP. It is also caused by APC gene mutations. People with AFAP develop pre-cancerous polyps in the GI tract. People with AFAP develop less polyps than people with FAP do. Most people with AFAP develop less than 100 polyps in their lifetime.

Cancer Risks

People with FAP and AFAP have an increased risk of developing certain cancers. Cancer develops when cells in a polyp begin to grow out of control. You and your doctor can manage your cancer risk with early diagnosis of FAP or AFAP, regular screenings for polyps and monitoring polyps.

Large Intestine

Most often, polyps are found in the large intestine. Cancer of the large intestine is called colorectal cancer. People with FAP and AFAP are at a high risk of developing colorectal cancer.

Small Intestine
People with FAP and AFAP can develop adenomas in the small intestine, most often in the duodenum, which is the first part of the small intestine. Sometimes the adenomas occur on an opening in the duodenum (called the ampulla of Vater). These adenomas have a small chance (about 1 to 10 in 100) to develop into cancer.

Stomach
Most people with FAP and many people with AFAP develop polyps in the stomach, called fundic gland polyps. The chance of a fundic gland polyp developing into stomach cancer is low.

Other Cancers
People with FAP and AFAP are also at risk of certain other cancers. These include thyroid cancer, brain cancer (called medulloblastoma), pancreatic cancer, and liver cancer in children. These cancers are rare. Most people with FAP do not develop them.

Signs and Symptoms

Signs and symptoms of polyps are rare when they first develop. As polyps grow, multiply, and become cancerous over time, signs and symptoms may occur. These include:
- Bright red blood in the stool
- Thin stools
- Diarrhea or constipation that cannot be explained by diet or illness
- Abdominal pain, cramping or bloating
- Continued weight loss
- Continued lack of energy
- Anemia

People with FAP may have other symptoms. These include:
- Lumps or bumps on the skull and jaw (osteomas)
- Cysts on the skin (epidermoid cysts)
- Dental changes, such as extra teeth
- Non-cancerous tumors most often found in the abdomen (desmoid tumors)
- Freckle-like spots on the inside of the eye, called congenital hypertrophy of the retinal pigmented epithelium (CHRPE)

In the past, people with FAP who had these other symptoms were considered to have Gardner's syndrome. Research has shown that Gardner's syndrome and FAP are the same condition.

Diagnosis

Early diagnosis of FAP and AFAP can help plan for additional cancer screenings and preventive care. This can help to detect cancer at an earlier stage. There is no cure for FAP or AFAP, but you can lower your cancer risks by following cancer screening and prevention guidelines. Personal and family history, exams, and genetic testing may be used to diagnose FAP or AFAP.

Genetic Testing

Genetic testing is a blood or saliva test that looks for mutations in the APC gene. If there is a chance you have FAP, genetic testing may confirm it and help you make informed choices about your health care.

Genetic tests are not perfect. Even if the test does not find an APC mutation you may still have FAP based on the number of polyps you have. Speak with your doctor or a genetic counselor for more information.

Polyps can occur at a young age. If a parent has FAP or AFAP, children should see a doctor right away to be checked for FAP. **Seek testing for children as early as possible**. **Do not** wait for children to develop symptoms.

- In families with FAP, it is recommended to start genetic testing and screening for FAP no later than age 15.
- In families with AFAP, it is recommended to start genetic testing and screening for AFAP in the late teens or earlier, based on family history.

Flexible Sigmoidoscopy

This is an exam of the rectum and the lower colon through a sigmoidoscope, a small flexible tube with a light on one end. The sigmoidoscope allows your doctor to examine the inner lining of the lower part of the colon and rectum. Your doctor may take a biopsy (a small sample of tissue) from polyps for further testing.

Colonoscopy

This is an exam of the large intestine through a colonoscope. The colonoscope is similar to the flexible sigmoidoscope, but the tube is longer. The exam allows your doctor to look at the inner lining of the large intestine, which is made up of the colon and rectum. Your doctor may take a biopsy from the polyps for further testing.

You will receive a sedative medicine to help you relax during the flexible sigmoidoscopy and colonoscopy exams. Most people sleep through the procedures and feel little or no discomfort.

Family Members

FAP and AFAP are genetic conditions, so family members may have FAP as well. The parents of a person with FAP may or may not have FAP.

- If a parent has FAP or AFAP, each child has a 1 in 2 chance of having FAP or AFAP.
- FAP and AFAP do not skip generations.
- Both males and females are equally affected.

Sometimes people develop FAP even though their parents do not have FAP. This is due to a new APC gene mutation happening by chance. About 1 in 4 people with FAP are the first people in their family to have it.

Family members of people with FAP or AFAP may benefit from genetic testing. Genetic testing can help find out if other family members have FAP or AFAP. This can help doctors decide which family members should consider additional cancer screenings and preventative care.

Screening and Prevention

Early diagnosis of FAP and AFAP is important for early detection and prevention of cancer. Because cancer is a major health risk related to FAP and AFAP, it is important to have a management plan. Talk with your doctor about management options. These may include taking medicine, regular screening exams for polyps, and surgery.

Screening

Screening exams can help prevent cancer by finding and removing polyps as early as possible. A regular schedule for screening exams may include the exams described below.

Colon Polyps and Colorectal Cancer

- Colonoscopy (preferred) or flexible sigmoidoscopy beginning at age 10 to 15 is recommended yearly for people with FAP.
- People with AFAP should consider having a yearly colonoscopy beginning in the late teens or earlier, based on family history.
- If present, precancerous polyps are removed during the colonoscopy **before** they develop into cancer. Your doctor may recommend surgery if they are too numerous or too large to remove.

Upper Intestinal Polyps and Cancer

- Screening of the upper GI tract is important for people with FAP and AFAP.
- Upper endoscopy exam (also called an EGD) should be performed at least every 3 to 5 years to monitor for fundic gland (stomach) polyps and duodenal (first part of the small intestine) adenomas.
- A side-viewing scope is also recommended for viewing the ampulla of Vater, which is a common location for polyps in the duodenum.
- Doctors often recommended having an EGD at age 20 to 25 or just before colorectal surgery. Recommendations may differ based on family history. Your doctor may recommend an EGD more frequently based on duodenoscopic findings.

Other Screening Tests

- Have a complete physical exam every year to check for symptoms of FAP outside of the large intestine.
- Baseline thyroid ultrasound beginning in late teenage years. If normal, repeat every 2-5 years.
- Some families may also consider liver cancer screening for young children, from birth to age 5. Screening includes a physical exam every year or an abdominal ultrasound exam and a blood test for a blood marker called alpha-fetoprotein (AFP).
- An abdominal ultrasound or computerized tomography (CT) scan is recommended before colon surgery to check for desmoid tumors

Colon Surgery

Cancer develops when cells in a polyp begin to grow out of control. When multiple precancerous polyps are found, it is treated by removing the colon before cancer occurs.

When people with FAP develop too many precancerous colon polyps to remove, colon surgery is recommended to help prevent cancer. When doctors recommend surgery, it means that removing the colon is the only way to prevent colon cancer. Without surgery, all patients with FAP will develop colorectal cancer.

The timing of surgery can vary among family members who have FAP or AFAP. You and your doctor will determine when you need to have surgery and the best type of surgery for you. The type of surgery depends on various factors including, but not limited to:

- The number of rectal polyps
- Whether or not you have colorectal cancer
- Your age
- You history of desmoids tumors
- Your preferences

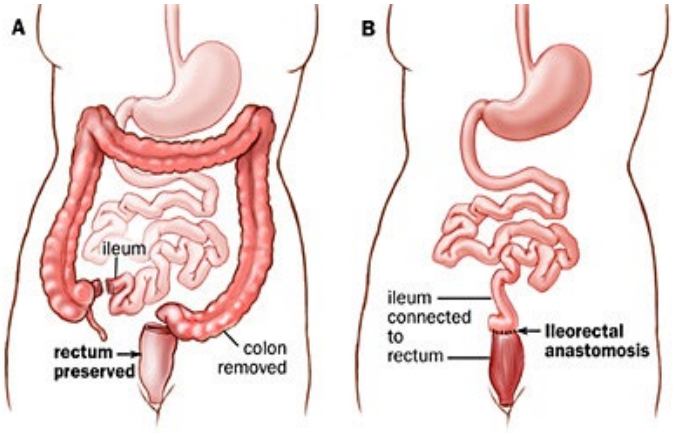
The 3 most common types of colon surgery are described below. All surgeries involve removing all or most of the colon.

Colectomy with Ileorectostomy (Ileorectal Anastomosis)

- The colon is removed, but all or most of the rectum is left in place. The small intestine is attached to the upper part of the rectum (See Figure 2).

- This procedure is recommended when there are very few polyps in the rectum.

- This is the least complicated colon surgery. Most patients maintain very good bowel function. Sometimes patients need anti-diarrheal medicines.



Figures 2 and 3
Reproduced with permission from
Johns Hopkins University.

Restorative Proctocolectomy
(Ileal Pouch Anal Anastomosis)

- The colon and most of the rectum are removed (See Figure 3).

- This surgery is recommended when there are many polyps in the rectum.

- A new rectum, or reservoir for stool, called a pouch, is made out of the lower end of the small intestine (ileum). The pouch is joined to the anus so bowel movements can flow in the normal way.

- A temporary ileostomy is needed – This involves having stool empty into a bag through the abdominal wall. This allows the pouch to heal. The ileostomy is used for about 8 to 10 weeks. It is removed through a second, less complex surgery. After the ileostomy is removed, many patients will have bowel movements like before surgery.

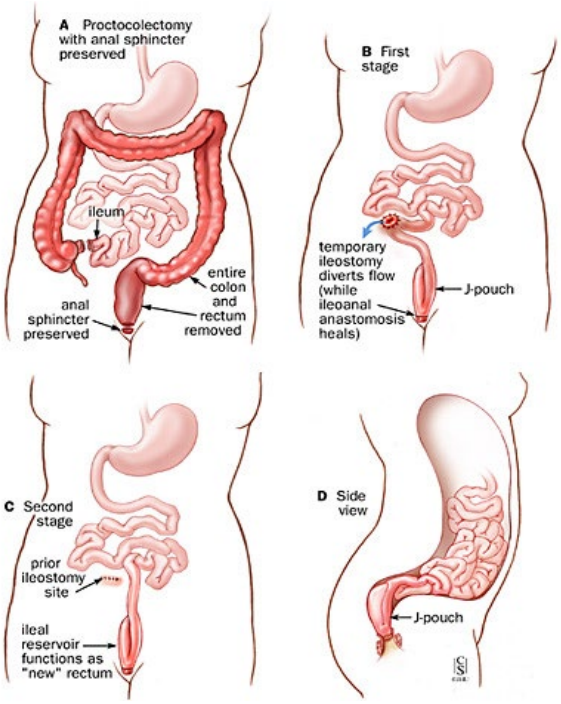


Figure 3

Total Proctocolectomy with Ileostomy

- The entire colon and rectum are removed. (See Figure 4).

- Few people need to have this kind of surgery.

- An end ileostomy is needed –The end of the small bowel (ileum) is brought to the surface of the abdomen, where it is permanently stitched into place.

- The rectum is removed, so it is not possible to control bowel functions in the normal way. Liquid stool will come out of the ileostomy into a bag that is securely attached onto the skin of the abdomen.

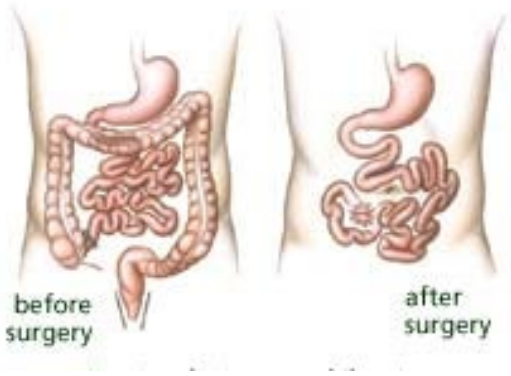


Figure 4
Reproduced with permission.
©2008, Cleveland Clinic

After Surgery

After surgery, polyps can occur in the remaining portion of the rectum. To treat these polyps, patients will have regular endoscopic exams of the rectum or the ileal pouch.

Prescription medicines may help reduce the risk of polyps returning. These medicines include nonsteroidal anti-inflammatory drugs (NSAIDs) such as ibuprofen, sulindac (Clinoril®) or celecoxib (Celebrex®). Treatment with these medicines will not replace endoscopy or surgery if it is needed.

The information below shows the recommended exams for patients after surgery.

	Patients with Rectum in Place	**Patients with Ileoanal Pouch**	**Patients with Ileostomy**
Complete Physical Exam	Every year	Every year	Every year
Upper Endoscopy (EGD) with Side-Viewing Scope	Every 1 to 5 years	Every 1 to 5 years	Every 1 to 5 years
Flexible Sigmoidoscopy	• Every 6 to 12 months for the first few years • Every 1 to 2 years, if the rectum remains free of polyps	Every 1 to 2 years	N/A
Ileostomy Surveillance	N/A	N/A	Every 1 to 3 years

Clinical Trials

Researchers are working to find medicines that prevent and reduce polyps. Research aims to find ways to treat FAP in the future with medicine instead of surgery.

Clinical trials are research studies that involve patients. They test new ways to treat or prevent diseases. MD Anderson has hosted many clinical trials for FAP. Ask your doctor or genetic counselor for information about clinical trials you may qualify for.

Resources

The University of Texas MD Anderson Cancer Center
Clinical Cancer Genetics Program
www.MDAnderson.org/Research/Departments-Labs-Institutes/Programs-Centers/Clinical-Cancer-Genetics/Educational-Resources.html
Learn more about FAP and general information on cancer genetics.

Desmoid Tumor Research Foundation
http://www.DTRF.org/
The Desmoid Tumor Research Foundation seeks to advance the science related to desmoid tumors.

Collaborative Group of the Americas on Inherited Colorectal Cancer (CGA)
http://www.CGAIGC.com/
The CGA focuses on families with rare forms of colorectal cancer, including FAP.