

Familial adenomatous polyposis (FAP) is an inherited condition that gives you a high risk for polyps and colon cancer.

Familial Adenomatous Polyposis

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Features of Familial Adenomatous Polyposis (FAP)

Polyps and Cancer: The main feature of FAP is the presence of many polyps in the colon and other parts of the gastrointestinal (GI) tract. Polyps are pre-cancerous growths that range in size from small to large. People with FAP may have a few dozen to several thousand colon polyps. Colon polyps often start forming in children as young as 10 years of age (or even younger sometimes). If polyps are left untreated, there is more than a 95% chance to develop colon or rectal cancer by age 40. Polyps and cancer may also develop in other areas of the GI tract, such as the small intestine and the stomach. Some families have a milder type of FAP, called “attenuated FAP” or AFAP, in which there are fewer polyps and colon cancer develops at a later age.

Benign tumors: A person with FAP can also develop benign tumors that sometimes cause medical problems. Examples of these non-cancerous growths include desmoid tumors (usually found in the abdomen) and osteomas (bony tumors in the skull and jaw).

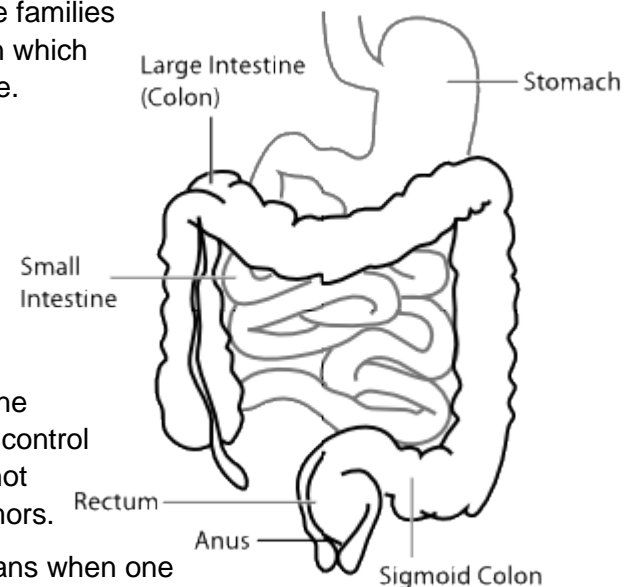
Genetics of FAP

FAP and AFAP are caused by a mutation (genetic change) in the adenomatous polyposis coli (APC) gene. The APC gene helps control how often a cell divides to make new cells. When this gene is not working normally, it leads to the growth of polyps and other tumors.

FAP and AFAP runs in families in a dominant pattern. This means when one parent has a mutation in the APC gene, each child has a 50% chance to inherit the mutation. The chance of inheriting an APC mutation is the same whether the parent with the mutation is the mother or father.

Cancers seen in FAP:

- Colon cancer
- Small intestine cancer
- Stomach cancer
- Pancreatic cancer
- Thyroid cancer (papillary)



You might have an APC mutation if you answer “yes” to any of these statements:

- I have had many colon polyps (10 or more)
- I had colon or rectal cancer before 50 years old
- I have had a desmoid tumor or an osteoma
- I have a parent or sibling with many polyps
- I have a relative with a mutation in the APC gene

Genetic Counseling

A genetic counseling visit can help determine if your cancer history might be due to FAP or AFAP. A genetic counselor reviews your medical records, your health history, and your family history. Genetic counselors are trained in genetics and counseling. You may also meet with a medical geneticist. Medical geneticists are doctors who specialize in genetic conditions. These genetic specialists can help guide you through decisions about genetic testing, when indicated. If you have testing, they will help you understand the results and arrange any follow-up services that are needed.

Genetic Testing for FAP or AFAP

Genetic testing is usually done on a blood sample from a person who has had colon cancer or many polyps. The test looks for mutations in the APC gene. Testing may include other cancer risk genes at the same time.

There are three possible results you can get from genetic testing:

- **NEGATIVE** - A negative result means that no mutation was found in the APC gene.
 - When a blood relative has an APC mutation, a negative result means you did not inherit the mutation and are not at high risk for cancer.
 - When you are the first person in the family to have testing, a negative result does not give you a complete answer about your cancer risk. Genetic testing does not always find a mutation in families with FAP or AFAP. If the history of polyps or cancer is suspicious, there may still be an inherited risk for cancer, even with a negative result.
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- **VARIANT of UNKNOWN SIGNIFICANCE (VUS)** - A VUS result means a genetic change was found in the APC gene, but we do not know if it causes a risk for polyps and cancer. Many variants are harmless DNA changes. A VUS result will not be used to make decisions about your medical care.

Cancer Management in FAP

If you have not had cancer, but have a family history of FAP or test positive for an APC mutation, your doctor will develop a cancer management plan that could include cancer screening at an early age or surgery to lower the risk of cancer. Research is also being done to find medications that might prevent new cancers or polyps from forming.

Cancer screening

Screening for cancer can help your doctor find and remove polyps as they form and help with decisions about surgical treatments.

- Colon cancer screening: People with FAP usually start colon cancer screening by age 10 to 15. Screening may include a sigmoidoscopy or a colonoscopy. A sigmoidoscopy allows your doctor to see inside the lower portion of the colon and look for areas of abnormal growth (polyps) that may lead to cancer. A colonoscopy looks at the entire colon (large intestine).
- Stomach and small intestine cancer screening: Screening of the upper gastrointestinal (GI) tract usually starts by age 25 using a procedure called esophagogastroduodenoscopy (EGD). This test uses a small camera on a flexible tube, that is put down the throat. The camera can be guided to the first part of the small intestine.

Risk-lowering surgery

Removal of the colon (colectomy) is usually advised once polyps start to form. However, the surgery may be delayed depending on the number and size of the polyps. For individuals with AFAP, regular colon cancer screening is sometimes able to manage the polyps without needing surgery.

Reference: NCCN Clinical Practice Guidelines, version 2.2019