

Familial adenomatous polyposis (FAP) syndrome is an inherited condition that gives you a higher risk for pre-cancerous polyps and colon cancer

Familial Adenomatous Polyposis

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Features of Familial Adenomatous Polyposis (FAP)

The main feature of FAP is the presence of multiple polyps in the gastrointestinal (GI) tract, most often in the colon. These polyps are pre-cancerous growths that can range in size from small to large. Men and women with FAP can have fewer than one hundred to several thousand polyps. Colon polyps can start forming as early as 10 years of age (or earlier in some cases). If polyps are left untreated, there is more than a 95% chance to develop colon or rectal cancer by 40 years of age. FAP also causes a higher chance for polyps and cancer in other areas of the GI tract, such as the small intestine and the stomach.

Individuals with FAP can also develop non-cancerous growths that sometimes cause medical problems, such as desmoid tumors (benign tumors, usually in the abdomen) and osteomas (bony growths in the skull and jaw). Some families have a milder type of FAP, called *attenuated FAP* or *AFAP*, in which there are fewer polyps and colon cancer typically develops at a later age.

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 an overgrowth of cells and the formation of polyps and other tumors.

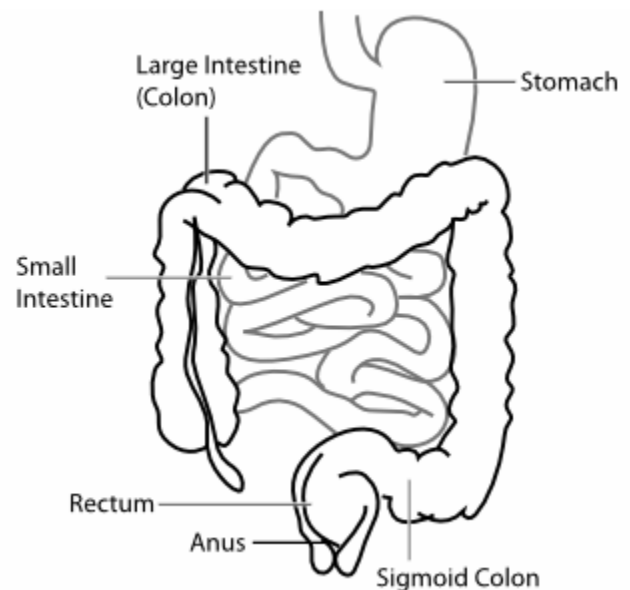
FAP and AFAP run in families in an autosomal dominant pattern. This means that if one parent carries a mutation in the APC gene, each child has a 50% chance to inherit the mutation and have a high risk to develop polyps and cancer. The chance of inheriting an APC mutation is the same whether the parent with the mutation is the mother or father.

Genetic Counseling

As part of a genetics cancer visit, 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. Your genetic providers can help determine if your history might be due to an APC mutation and guide you through decisions about genetic testing, if indicated. If you have testing, they will help you understand the result and arrange any follow-up services that are needed.

Cancers seen in FAP:

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



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The information is not intended to diagnose health problems or to take the place of professional medical care. If you have persistent health problems or if you have further questions, please consult your health care provider.



You may have a higher chance of carrying an APC mutation if you answer "yes" to one or more of the following statements:

- I have had many colon polyps (10 or more)
- I have 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 multiple polyps
- I have a relative with a mutation in the APC gene

Genetic Testing For FAP or AFAP

Genetic testing looks for mutations in the APC gene. Testing is usually done on a blood sample from you or a family member who has multiple polyps or colon cancer.

Results of genetic testing may show:

- **A positive genetic test.** A positive result means that an APC mutation was identified that increases your chance for polyps and cancer. It does NOT mean that you will definitely get cancer, but it does mean your lifetime risk of developing cancer is higher than average.
- **A negative genetic test.** A negative result means that no mutation was found in the APC gene. When a mutation has already been identified in another family member, a negative test provides reassurance that your cancer risk is not higher than average. However, when you are the first person in the family to have testing, a negative test may not be reassuring. Genetic testing is not always able to find a mutation in families with FAP or AFAP. If the history of polyps or cancer is suspicious, there may still be a hereditary risk for cancer, even if your test result is negative.
- **A gene variation of unknown significance.** Genetic testing sometimes finds genetic mutations that are not able to be interpreted. Some mutations do not affect the way a gene works at all, while other mutations completely stop a gene from working. Sometimes when a mutation is found, it is not clear whether or not the gene can still work. In these cases, we do not know whether or not the identified mutation will cause any increase in the risk for cancer to develop.

Cancer Management in FAP

If you have not had cancer, but have a family history of FAP or have tested positive for an APC mutation, your doctor will develop a cancer management plan that includes starting cancer screening at an early age or having risk-reducing surgery.

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 exam 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, which is inserted down the throat. The camera can be guided to the first part of the small intestine.

Surgical options

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 2.2011