

What you need to know

APC Gene

What does it mean to have an APC pathogenic variant?

Having an APC pathogenic variant, also known as a mutation, means a person has a version of the APC gene that may cause the following cancer predisposition conditions:

- Familial Adenomatous Polyposis (FAP) syndrome (also known as Classic FAP)
- Attenuated Familial Adenomatous Polyposis (AFAP) syndrome

What is my risk for cancer if I have an APC pathogenic variant?

If you have an APC pathogenic variant, you have a greater risk of developing gastrointestinal polyps (pre-cancerous growths also known as adenomas), as well as certain types of cancer.

What is the difference between FAP and AFAP syndrome?

Classic FAP:

- All individuals with FAP are at an increased risk to develop many precancerous polyps (100s-1000s of polyps).
- Children with FAP are at risk for developing intestinal polyps and certain cancers.
- There is a nearly 100% chance of developing colon cancer by age 50 without regular screening and/or surgery.
 - This is based on individuals who did not have regular screening and/or other treatments such as
 preventative surgery. There are data suggesting that people with an APC pathogenic variant can
 significantly decrease their risk of developing cancer by careful medical and surgical follow-up.
- Individuals with FAP may also develop cancer of the small bowel, thyroid, liver, central nervous system (brain), bile ducts, and stomach.
- Non-cancerous features of FAP may include: polyps of the stomach, polyps of the small intestine, bony
 growths (typically on the jaw or skull), dental problems, unusual pigment in the eye (CHRPE), and soft
 tissue tumors.

Attenuated FAP (AFAP):

- The features of AFAP are similar to those seen in classic FAP; however, they are typically milder and begin at a later age.
- Individuals with AFAP usually have between 10 and 100 precancerous colon polyps.
- Without intervention, there is a 70% lifetime chance of colon cancer, which is usually diagnosed after the age of 50.

If I have an APC pathogenic variant, what is the chance my family members will have it?

There is a 50% chance that a person with a pathogenic variant will pass it on to each of their children. In most cases, siblings of a person with a pathogenic variant each have a 50% chance to have the pathogenic variant. Additionally, other family members are at risk to have the pathogenic variant.



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