APC gene: What You Need to Know

What does it mean to test positive for an APC gene mutation?
Mutations in the APC gene 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 gene mutation?
If you have an APC gene mutation, 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 (100-1000s).
- Children with FAP are at risk for developing intestinal polyps and cancer.
- There is a nearly 100% chance of developing colon cancer by age 50 without regular screening and/or surgery*. The average age of diagnosis is around 40 years old.
- Individuals with FAP may also have cancer of the small bowel, thyroid, liver, pancreas, adrenal glands, 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 very similar to those seen in classic FAP; however, they are typically milder and begin at a later age. Individuals usually have between 10 and 100 precancerous colon polyps and a 70% lifetime chance of colon cancer, which is usually diagnosed between the ages of 50-55 years*. Although other features of classic FAP may be present, the eye pigmentation (CHRPE) and soft tissue tumors are rare.

*It is important to note that these risks are based on individuals who did not have regular screening and/or other treatments such as preventative surgery. There are data that suggest that people with an APC mutation can significantly decrease their risk of developing cancer by careful medical and surgical follow-up.

What is the chance that my family members will have an APC mutation if I test positive?
There is a 50% chance that a person with a mutation will pass it on to each of his/her children. In most cases, brothers and sisters of a person with a mutation have a 50% chance to have the mutation. Additionally, other family members are at risk to have the mutation.