Quest Advanced® Oncology

Hereditary Cancer Genetic Test Results

This report is intended to facilitate a discussion between providers and their patients.

INFORMATION FOR INDIVIDUALS WITH A PATHOGENIC OR LIKELY PATHOGENIC VARIANT IN THE *APC* GENE

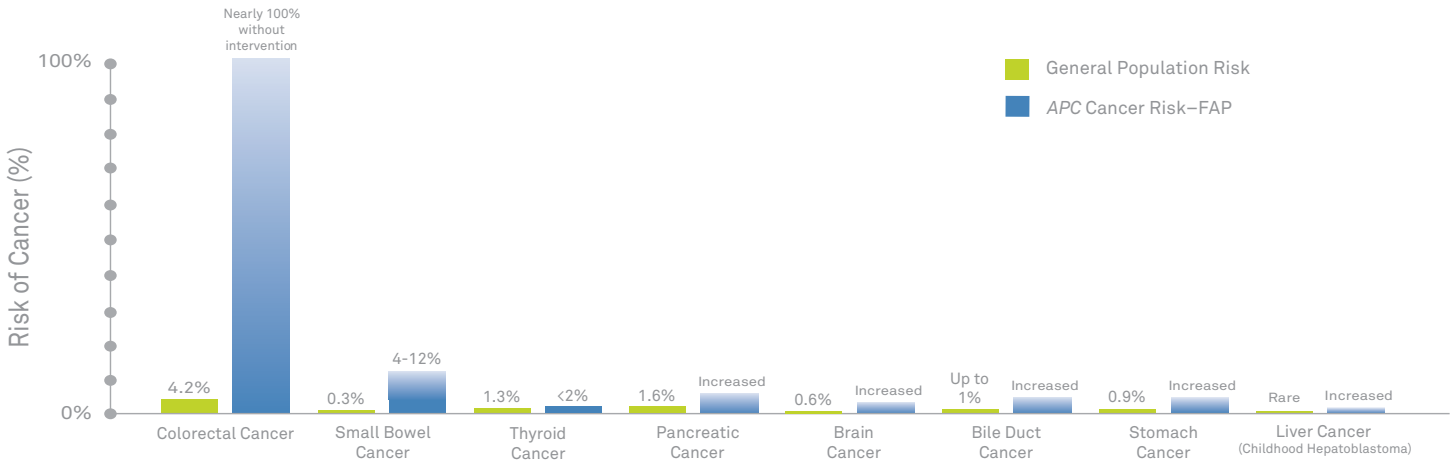
What this result means

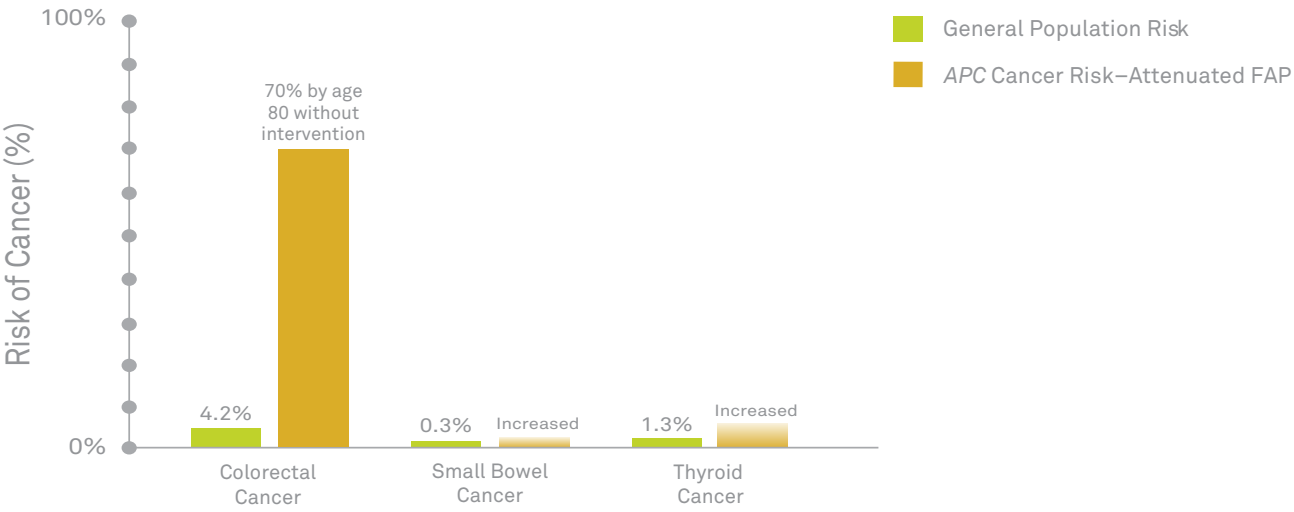
Individuals who have a pathogenic or likely pathogenic variant (sometimes called a mutation) in the *APC* gene have familial adenomatous polyposis (FAP). Individuals with FAP have a higher-than-average chance to develop adenomatous colorectal polyps as well as the following cancers: colorectal, small bowel, thyroid, pancreatic, brain, bile duct, stomach, and hepatoblastoma. Non-cancerous findings may include fundic gland and duodenal polyps, desmoid tumors, osteomas, soft tissue growths, dental anomalies, and congenital hypertrophy of the retinal pigment epithelium (CHRPE). Some individuals with pathogenic variants in *APC* have attenuated FAP (AFAP), with fewer polyps and a later-onset of colorectal polyps and colorectal cancer. Individuals with AFAP may also have a higher-than-average cancer to develop small bowel and thyroid cancer.

Cancer risk

These graphs below compares general population cancer risks to the potential cancer risks associated with pathogenic variants in the *APC* gene. Individual cancer risks may be higher or lower depending on the specific variant identified in addition to each individual's gender, age, medical history, and family history.
Not everyone with a pathogenic or likely pathogenic variant will develop cancer.

Information about cancer risks related to pathogenic variants in *APC* may change over time, so it is important for the ordering healthcare provider, genetic counselor, and patient to keep in contact regarding this result.





*Data on file.

Options for managing cancer risk

There are options for cancer prevention and early detection. The following are general guidelines for individuals who have an *APC* pathogenic variant. These guidelines evolving and are not specific to any one individual. Each individual's gender, age, medical history, family history, quality of life goals, reproductive desires, general health status, and other medical information should be taken into account when developing a medical management plan.

	Considerations for cancer prevention/early detection	Age to begin	Frequency
Colon Cancer	Colonoscopy	10-15 years	FAP: Annual; If adenomas are found, consider surgical evaluation and counseling AFAP: 1-2 years; Surgical evaluation and counseling if appropriate
	Proctocolectomy or colectomy	Individualized based on polyp burden and clinical presentation	—
	Post-colectomy surveillance	Following Surgery	Based on surgery performed and polyp burden
	Post-colectomy chemoprevention	Individualized	Individualized
Duodenal or Periampullary Cancer	Upper endoscopy	20-25 years; Earlier if colectomy prior to age 20 or based on family history of duodenal polyp burden or cancer	Every 4 years; If polyps are found, adjust frequency and surgical consideration based on polyp burden

	Considerations for cancer prevention/early detection	Age to begin	Frequency
Gastric Cancer	Monitor for high risk endoscopic findings. Consider center of expertise if high risk features are present or lesions cannot be removed endoscopically.	—	—
Thyroid Cancer	Thyroid ultrasound	Late teenage years	Every 2-5 years unless abnormal findings or family history of thyroid cancer
CNS	Patient should be educated on the signs and symptoms of neurologic cancer and should report abnormal symptoms to their providers		
Intra-abdominal desmoids	Abdominal palpation	—	Annual
	Abdominal imaging (MRI or CT), if family history of desmoids or if abdominal symptoms are present	—	Annual
Small bowel polyps and cancer	Consider including small bowel imaging for desmoids *limited evidence	—	—
Hepatoblastoma	Consider liver palpation, abdominal ultrasound and AFP	0-5 years	Every 3-6 months
Pancreatic Cancer	Limited evidence, individualized based on family history		
Other Management	Refer to multidisciplinary team of specialists familiar with FAP for evaluation and management Individualize management based on specific variant, clinical presentation and personal considerations		

Source: National Comprehensive Cancer Network. NCCN Clinical Practice Guidelines in Oncology. Genetic/Familial High-Risk Assessment: Colorectal. V1.2021. **www.NCCN.org**

What this result means for family members

Family members may have the same *APC* variant that was identified in this individual. Parents, brothers, sisters, and children may each have a 50% chance of having the same variant. Other blood relatives also have an increased risk for the variant. It is important to share these test results with family members to allow each of them to decide if they want to be tested. Some family members may only need testing for this one *APC* variant, while other relatives may need a more comprehensive test with multiple genes. A genetic counselor or other healthcare provider can help determine the most appropriate testing options.

Reproductive information

Individuals interested in family planning should speak to their doctor and/or genetic counselor to discuss reproductive options. This may include discussion of prenatal diagnosis or pre-implantation genetic testing.

Risk assessment and counseling: an important first step

A genetic counselor or other qualified healthcare professional can help explain test results and what they mean for a patient and family members. A team of specialized Quest genetic counselors or clinical geneticists are available to speak with healthcare providers about test results by calling 1.866.GENE.INFO. Patients can access a directory of independent genetic counselors at **FindAGeneticCounselor.com**.

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Creating a plan: a checklist for patients

- ☐ Get a copy of your genetic test results.
- ☐ Talk with your healthcare provider about what this result means and the things you can do to manage your risk.
- ☐ Ask your healthcare provider if additional genetic testing may benefit you.
- ☐ Share your test results with your family members and give them a copy. Their healthcare provider will need this information in order to provide them with the most accurate risk assessment.
- ☐ Talk with your healthcare provider regularly so that you know about any important changes in genetic testing and cancer screening options. Be sure to let him/her know of any changes in your family history, including family members' genetic test results.
- ☐ Consider talking to a genetic counselor about your results.

Research opportunities

Prospective Registry of MultiPlex Testing (PROMPT) PromptStudy.info

GenomeConnect: The ClinGen Patient Portal GenomeConnect.org

Additional resources

Hereditary Colon Cancer Takes Guts
hcctakesguts.org

Colorectal Cancer Alliance
ccalliance.org

National Colorectal Cancer Roundtable
nccrt.org

Quest Hereditary Cancer Testing Solutions
QuestHereditaryCancer.com

Genetic Information Nondiscrimination Act (GINA) GINAhelp.org

National Society of Genetic Counselors
FindAGeneticCounselor.com

This information is not a substitute for medical advice, diagnosis, or treatment. The diagnosis or treatment of any disease or condition may be based on personal history, family history, symptoms, a physical examination, laboratory test results, and other information considered important by a healthcare provider. Always talk with a healthcare provider about the meaning of genetic test results and before stopping, starting or changing any medication or treatment.

The classification and interpretation of the variant(s) identified reflect the current state of Quest Diagnostics' understanding at the time of this report. Variant classification and interpretation are subject to professional judgment, and may change for a variety of reasons, including but not limited to, updates in classification guidelines and availability of additional scientific and clinical information. This test result should be used in conjunction with the healthcare provider's clinical evaluation. Inquiry regarding potential changes to the classification of the variant is strongly recommended prior to making any clinical decision. For questions regarding variant classification updates, please call Quest Diagnostics at 1.866.GENE.INFO (1.866.436.3463) to speak to a genetic counselor or laboratory director, or visit **QuestDiagnostics.com/VariantIQ**.