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Clinician Management Resource for *APC* (Familial Adenomatous Polyposis)

This overview of clinical management guidelines is based on this patient's positive test result for a pathogenic or likely pathogenic *APC* variant. Unless otherwise stated, medical management guidelines used here are limited to those issued by the National Comprehensive Cancer Network® (NCCN®)[1] in the U.S. Please consult the referenced guideline for complete details and further information.

Clinical correlation with the patient's past medical history, treatments, surgeries and family history may lead to changes in clinical management decisions; therefore, other management recommendations may be considered. Genetic testing results and medical society guidelines help inform medical management decisions but do not constitute formal recommendations. Discussions of medical management decisions and individualized treatment plans should be made in consultation between each patient and his or her healthcare provider, and may change over time.

SCREENING/SURGICAL CONSIDERATIONS*,[1]	AGE TO START	FREQUENCY
Colorectal Cancer		
Asymptomatic patients: High quality colonoscopy	10-15 years old†	Every 12 months
Affected patients: Colectomy or proctocolectomy**	Individualized by polyp burden	N/A
Additional surveillance post-colectomy is recommended and varies by surgery type performed. *Please consult complete NCCN Clinical Practice Guidelines in Oncology (NCCN Guidelines®) for additional details.*	Post-surgery	Varies by surgery type
Consider chemoprevention to facilitate management of the remaining rectum or pouch post-surgery in select patients with progressive polyp burden (eg, based on size, number, and pathology).	Post-surgery	N/A
Duodenal Or Periampullary Cancer^		
Upper endoscopy including complete visualization of the ampulla of Vater *Refer to the complete NCCN Guidelines® for further details regarding type and frequency of surveillance.[1]*	20-25 years old or earlier if the patient has a family history of aggressive duodenal adenoma burden or cancer	Individualized
Gastric Cancer^		
Refer to the complete NCCN Guidelines for further details regarding the management of fundic gland and non-fundic gland polyps, if applicable.[1]	Individualized	Individualized
Thyroid Cancer^		
Thyroid ultrasound	Late teenage years	If normal, consider repeating every 2-5 years. If abnormal, refer to a thyroid specialist††
Central Nervous System Cancer^		
There is currently no support for routine surveillance imaging. However, patients should be educated regarding signs and symptoms of neurologic cancer and the importance of prompt reporting of abnormal symptoms to their physicians.	Individualized	Individualized
Intra-Abdominal Desmoids^		
Suggestive abdominal symptoms should prompt immediate abdominal imaging. Patients should be educated regarding signs and symptoms of intra-abdominal desmoids and the importance of prompt reporting of abdominal symptoms to their physicians.	Individualized	Individualized

SCREENING/SURGICAL CONSIDERATIONS*,[1]	AGE TO START	FREQUENCY
Small Bowel Polyps And Cancer^		
Consider small bowel visualization via capsule endoscopy or CT/MRI enterography, especially if duodenal polyposis is advanced.^^	Individualized	Individualized
Hepatoblastoma^		
May consider liver palpation, abdominal ultrasound, measurement of alpha-fetoprotein (AFP)^^.	0-5 years old	Every 3-6 months

* Some individuals have a milder phenotype of FAP, known as attenuated FAP (AFAP). Recommendations for individuals with AFAP may vary from the guidelines outlined here, including differences in the age, frequency, and type of management recommended. It is recommended that patients be managed by physicians or centers with expertise in FAP and that management be individualized to account for genotype, phenotype, and personal considerations. Additionally, these risks and management recommendations do not apply to individuals with the p.I1307K or promoter IB (GAPPS) mutations in the *APC* gene. Please consult the complete NCCN Guidelines (as referenced below) for management details.

** Timing of proctocolectomy in patients <18 y of age is not established since colon cancer is rare before age 18. In patients <18 y without severe polyposis and without family history of early cancer or severe genotype, the timing of proctocolectomy can be individualized. An annual colonoscopy is recommended if surgery is delayed.

† Earlier initiation of screening can be considered based on family history.

†† Shorter intervals may be considered for individuals with a family history of thyroid cancer.

^ Other than colorectal cancer, screening recommendations are expert opinion rather than evidence-based.

^^ Limited data

1. Referenced with permission from the NCCN Clinical Practice Guidelines in Oncology (NCCN Guidelines®) for Genetic/Familial High-Risk Assessment: Colorectal, Endometrial, and Gastric. v3.2024. © National Comprehensive Cancer Network, Inc. 2024. All rights reserved. Accessed October 31, 2024. To view the most recent and complete version of the guideline, go online to NCCN.org. NCCN makes no warranties of any kind whatsoever regarding their content, use or application and disclaims any responsibility for their application or use in any way.

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Understanding Your Positive *APC* Genetic Test Result

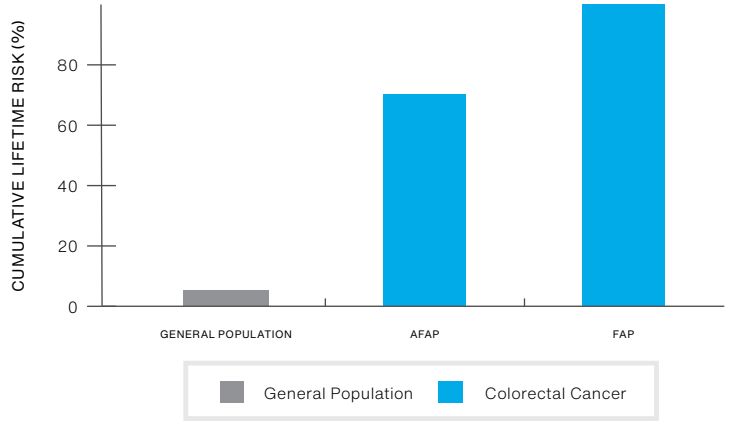
INFORMATION FOR PATIENTS WITH A PATHOGENIC OR LIKELY PATHOGENIC VARIANT

5 Things To Know^

1	Result	Your testing shows that you have a pathogenic or likely pathogenic variant in the *APC* gene.
2	FAP or AFAP	People with a pathogenic or likely pathogenic *APC* variant have familial adenomatous polyposis (FAP) or attenuated FAP (AFAP).
3	Cancer risks and other medical concerns	You have an increased chance to develop multiple gastrointestinal polyps, colorectal cancer, and possibly cancers of the thyroid, stomach, pancreas, liver, and central nervous system.
4	What you can do	Risk management decisions are very personal. There are options to detect cancer early or lower the risk to develop cancer. It is important to discuss these options with your healthcare provider and decide on a plan that works for you.
5	Family	Family members may also be at risk – they can be tested for the pathogenic or likely pathogenic *APC* variant that was found in you. It is recommended that you share this information with your family members so they can learn more and discuss with their healthcare providers.

^ These risks and management recommendations do not apply to individuals with the p.I1307K moderate risk mutation or promoter IB (GAPPS) mutations in the *APC* gene. See the NCCN Genetic/Familial High-Risk Assessment: Colorectal guidelines for information on these conditions.

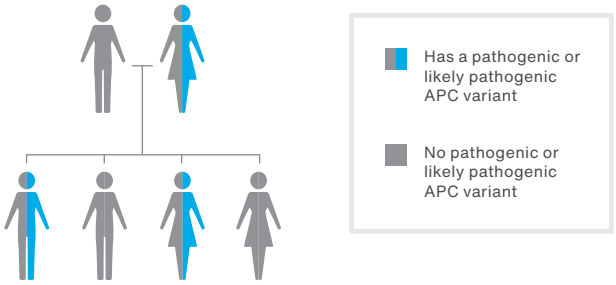
APC Lifetime Colorectal Cancer Risks*



* Because risk estimates vary in different studies, only approximate risks are given. Cancer risks will differ based on individual and family history.

APC in the Family

There is a 50/50 random chance to pass on the pathogenic or likely pathogenic *APC* variant to each of your children.



RESOURCES

- American Cancer Society cancer.org
- National Society of Genetic Counselors nsgc.org
- Canadian Society of Genetic Counsellors cagc-accg.ca

Please discuss this information with your healthcare provider. The cancer genetics field is continuously evolving, so updates related to your *APC* result, medical recommendations, and/or potential treatments may be available over time. This information is not meant to replace a discussion with a healthcare provider, and should not be considered or interpreted as medical advice.

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