



# Adenomatous Polyposis Syndromes (FAP/AFAP and MAP)

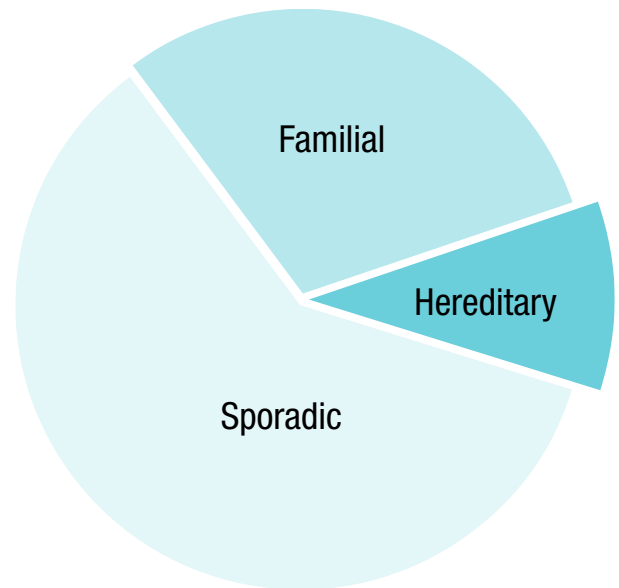
**A Patient's Guide**  
to risk assessment

# Hereditary Cancer Testing: Is it Right for You?

This workbook is designed to help you decide if hereditary cancer testing is right for you. Testing should be completed with a trained healthcare provider.

## Introduction

Most cancer occurs by chance. This is often called “sporadic cancer.” In some families we see more cancer than we would expect by chance alone. Determining which of these families have cancer related to an inherited gene mutation is important, as the cancer risks in hereditary cancer families are much higher than the general population.



**Sporadic Cancer** - Cancer which occurs by chance. People with sporadic cancer typically do not have relatives with the same type of cancer.

**Familial Cancer** - Cancer likely caused by a combination of genetic and environmental risk factors. People with familial cancer may have one or more relatives with the same type of cancer; however, there does not appear to be a specific pattern of inheritance (eg, the cancer risk is not clearly passed from parent to child).

**Hereditary Cancer** - Cancer occurs when an altered gene (gene change) is passed down in the family from parent to child. People with hereditary cancer are more likely to have relatives with the same type or a related type of cancer. They may develop more than one cancer and their cancer often occurs at an earlier than average age.

## Personal and Family History\*

Check all that apply:

- Ten or more cumulative colorectal adenomatous polyp(s)  
Number: \_\_\_\_\_ Age: \_\_\_\_\_
- A previously identified *MYH* or *APC* mutation in the family

Adenomatous polyposis syndromes are inherited conditions that are associated with the development of multiple polyps in the colon and rectum. Mutations in the *APC* gene cause **familial adenomatous polyposis (FAP)** or **attenuated FAP (AFAP)**. Patients who have a mutation in the *APC* gene can have tens to hundreds of colon polyps (adenomas), a greatly increased risk of colorectal cancer, and an increased risk for other associated cancers. An *APC* mutation can be inherited from either your mother or father.

***MYH*-associated polyposis (MAP)** is a hereditary condition that causes an increased risk for colorectal cancer and colorectal polyps. MAP is caused by mutations in the *MYH* gene. Mutations in the *MYH* gene are inherited in an autosomal recessive pattern, meaning individuals with MAP have mutations in both of their *MYH* genes (one from each parent). Individuals with MAP often do not have a family history of colon cancer or colon polyps in family members (although siblings may be affected).

\*Assessment criteria based on medical society guidelines. For these individual medical society guidelines go to [www.MyriadPro.com](http://www.MyriadPro.com)

## Cancer Risks for People Who Have an *APC* or *MYH* Gene Mutation(s)

	Gene Mutation Carrier Risk	General Population Risk <sup>1</sup>
Colorectal cancer in FAP	approximately 100%	2%
Colorectal cancer in AFAP	80%-100%	2%
Duodenal cancer	5%-12%	NA <sup>2</sup>

*APC* gene mutation carriers have a slightly elevated risk over the general population of developing cancers of the pancreas, thyroid, stomach, and brain. Liver cancer risk in children is also increased.

Because of the numerous colorectal adenomas that occur in MAP, the colorectal cancer risk is known to be significantly increased. Additionally, it is possible that risks of other cancers, such as small bowel, may be increased as well.

1. Risk by age 70  
2. Not available

# Managing Risk for APC Mutation Carriers\*

## INCREASED SURVEILLANCE

SITE	PROCEDURE	AGE TO BEGIN	REPEAT TEST
Colon-FAP	Sigmoidoscopy or colonoscopy	10-15 years	Annually
Colon-AFAP	Colonoscopy	Late teens or early 20s (depending on age of polyp development in the family)	1-3 years
Colon-After colon surgery	Endoscopy of remaining rectum, ileal pouch, or ileostomy	After colon surgery	6 months to 3 years (depending on polyp number and type of surgery)
Duodenum and stomach	Baseline upper endoscopy (including side-viewing examination)	20-25 years	1-4 years
Thyroid	Physical exam and consideration of ultrasound	Late teens	Annually

Screening for other FAP/AFAP-related cancers (brain, pancreatic, hepatoblastoma, etc.) may be considered. Please speak to your healthcare provider.

\*For reference and supporting data on risk factors and medical management visit [www.MyriadPro.com/references](http://www.MyriadPro.com/references)

## SURGICAL MANAGEMENT

- FAP—Preventive removal of the colon and rectum is recommended. The timing of surgery is based on the number/size of polyps.
- AFAP—Preventive removal of the colon and the rectum may be recommended depending on the number of polyps.

## CHEMOPREVENTION

- Medications may be used to reduce the number of polyps in any rectum tissue that remains after colon surgery.

## Managing Your MAP Cancer Risks\*

Options for reducing cancer risk are available whether or not you have already had a diagnosis of cancer and/or polyps (adenomas). It is recommended that you be managed according to these guidelines, depending on the number of colorectal polyps (adenomas) in you and your family members. Discuss these options with the appropriate medical professionals to determine how you will manage your cancer risks.

### INCREASED SURVEILLANCE

SITE	PROCEDURE	AGE TO BEGIN	REPEAT
Colon—Small polyp (adenoma) burden, manageable by colonoscopy	Colonoscopy	25-30 years	1-2 years
Colon—Large polyp (adenoma) burden	Counseling regarding surgical options	Varies based upon polyp (adenoma) burden	N/A
Colon—After colon surgery	Endoscopy of any remaining colon and rectum	After colon surgery	1-2 years
Duodenum and stomach <sup>†</sup>	Upper endoscopy and side viewing duodenoscopy	30-35 years	3-5 years

<sup>†</sup>Patients who have small bowel polyps (adenomas) should follow FAP small bowel screening guidelines.

### SURGICAL MANAGEMENT

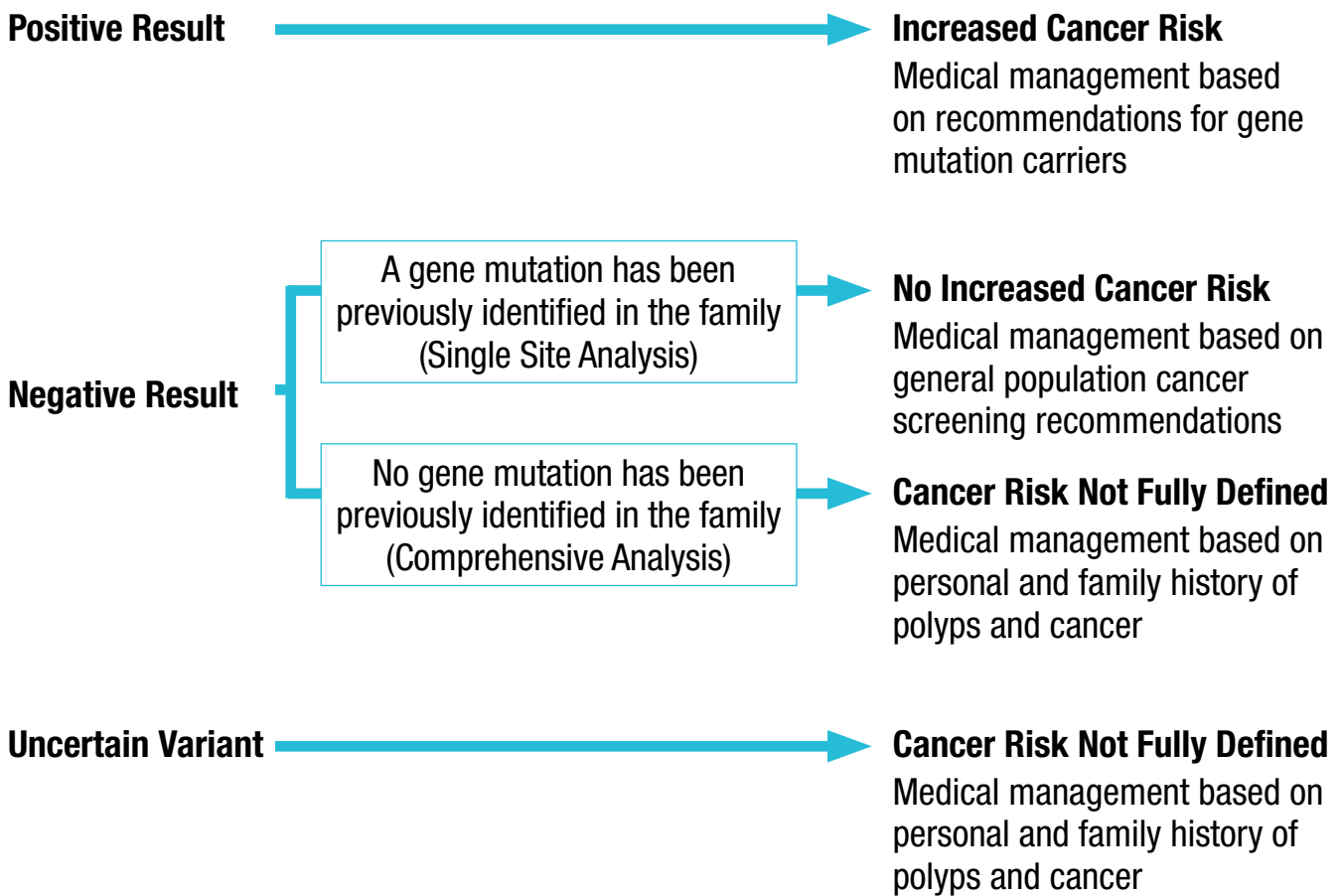
- Preventive removal of the colon and rectum may be recommended depending on the number of adenomas (polyps).

\*For references and supporting data on risk factors and medical management, visit [www.MyriadPro.com/references](http://www.MyriadPro.com/references)

## Testing Options

- COLARIS *AP*<sup>PLUS</sup>® (AFAP, FAP, and MAP):**  
Sequence and large rearrangement analysis of the *APC* and *MYH* genes.
- Single Gene Testing:**  
Sequence and large rearrangement analysis of one of the *APC* or *MYH* gene.
- Single Site Testing:**  
Mutation specific analysis for individuals with a known AFAP, FAP, or MAP mutation in the family.

## Possible Test Results

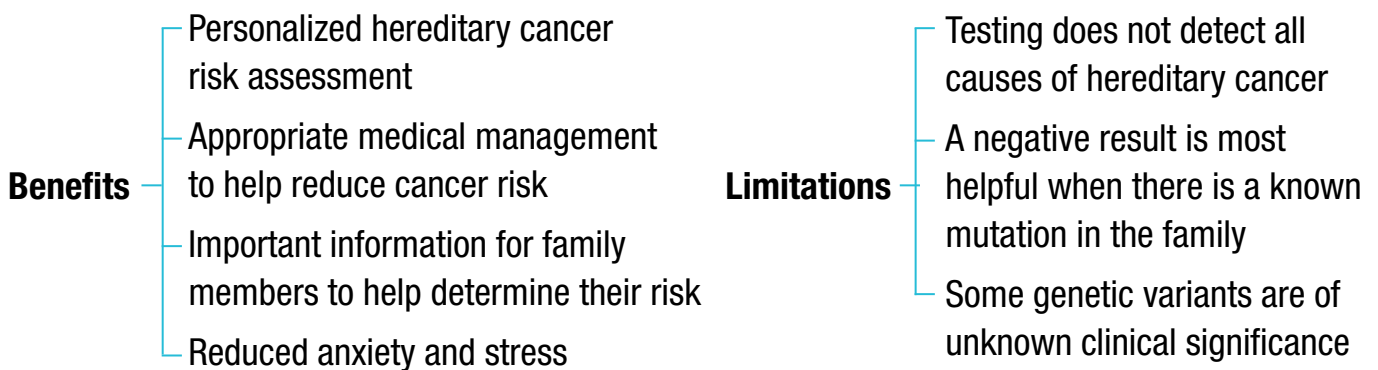


Patients who test positive for one *MYH* gene mutation do not have *MYH*-associated polyposis (MAP), but may have a small increased risk for colon cancer.

## It's a Family Affair

- *APC* and *MYH* gene mutations are passed on in a family
  - If you have gene mutations in either of these genes, your parents, your children, and your brothers and sisters may also have the same gene mutation
  - Other relatives such as aunts, uncles and cousins may also be at risk to carry the same gene mutation(s)
- Testing is the only way to identify gene mutation carriers
- It is important to share genetic test results with family members
- Individuals may have different viewpoints and reactions to genetic testing

## Benefits and Limitations



## Health Care Coverage

Insurance coverage for genetic testing of at-risk patients is excellent, with the majority of patients covered for testing. The average patient payment for an integrated COLARIS *AP* test, including patient who pay nothing, is less than \$100.\*

For information regarding Myriad's Patient Financial Assistance Program visit [www.MyriadPro.com](http://www.MyriadPro.com) or contact Customer Service at 800-4-MYRIAD (800-469-7423).

\*Test prices may be confirmed by calling Myriad Customer Service at 800-469-7423. Unmet deductibles are always the responsibility of the patient.

## Privacy

Federal laws (HIPAA and GINA) and laws in most states prohibit discrimination regarding employment eligibility, health benefits, or insurance premiums based solely on genetic information. Additionally, it is Myriad's policy that test results are disclosed only to the ordering healthcare professional or designee, unless the patient consents otherwise.

## Next Steps

Pursue testing

Schedule follow-up appointment for results disclosure

Date: \_\_\_\_\_ Time: \_\_\_\_\_

Decline testing-Medical management based on personal and family history of cancer

Undecided

Who to contact with questions: \_\_\_\_\_

## Additional Resources

### Colorectal Cancer Network

A support network for individuals and families touched by colon cancer that promotes awareness, screening, and early detection programs as well as legislative actions.

[www.colorectal-cancer.net](http://www.colorectal-cancer.net)

301-879-1500

### Colon Cancer Alliance

The Colon Cancer Alliance (CCA) is a national patient advocacy organization dedicated to ending the suffering caused by colorectal cancer.

[www.ccalliance.org](http://www.ccalliance.org)

877-422-2030 (toll free)

### Myriad Genetic Laboratories, Inc.

[www.MySupport360.com](http://www.MySupport360.com)

800-4-MYRIAD (800-469-7423)

E-mail: [helpmed@myriad.com](mailto:helpmed@myriad.com)

[www.HereditaryCancerQuiz.com](http://www.HereditaryCancerQuiz.com)



[MySupport360.com](http://MySupport360.com)



MYRIAD®

Myriad Genetic Laboratories, Inc.  
320 Wakara Way  
Salt Lake City, UT 84108

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