Understanding Your Genetic Test Result

Positive for a Deleterious Mutation or Suspected Deleterious

COLARIS *AP*®

This workbook is designed to help you understand the results of your genetic test and is best reviewed with your healthcare provider. Please verify that your test result matches the following information by looking at the patient copy of your test result or contacting the healthcare provider who ordered your test. If your test result does not match, please disregard this brochure, and contact your healthcare provider.

Overview of FAP and AFAP

Mutations in *APC* cause one of two adenomatous polyposis syndromes: familial adenomatous polyposis (FAP) or attenuated familial adenomatous polyposis (AFAP). These syndromes are associated with the development of varying numbers of adenomas (pre-cancerous polyps) in the colon and rectum.

	Number of Colorectal Adenomas (Polyps)
Familial Adenomatous Polyposis (FAP)	Hundreds to thousands
Attenuated FAP (AFAP)	Between 10 and 99 over a lifetime

FAP and AFAP also lead to an increased risk of colon/rectal (colorectal) cancer and, in some cases, other types of cancer.

Your Genetic Test Result (check the appropriate boxes below)

THE GENETIC TEST YOU RECEIVED

❑ **COLARIS *AP*® PLUS:**
Full sequence and large rearrangement analysis of the *APC* and *MYH* genes.

❑ **Gene-Specific COLARIS *AP*—*APC* Analysis:**
Full sequence and large rearrangement analysis of the *APC* gene.

❑ **Single Site COLARIS *AP*:**
Mutation-specific analysis for individuals with a known *APC* gene mutation in the family.

YOUR TEST RESULT

❑ Positive for a Deleterious Mutation
❑ Genetic Variant, Suspected Deleterious

Overview of Your Test Result

- You have a mutation or alteration in the *APC* gene.
- You have an adenomatous polyposis syndrome (either FAP or AFAP, depending on the number of colorectal adenomas).
- FAP and AFAP increase the risk of colorectal adenomas/cancer, and in some cases, other types of cancer.
- The risk of developing these cancers is less than 100%. Not everyone with FAP or AFAP will develop cancer.

Your Cancer Risks

Having FAP or AFAP increases the risk of certain cancers.

Colorectal cancer risks by age 70	Mutation Carrier	General Population
FAP	~100	2%
AFAP	80 - 100%	2%

Other cancer risks by age 70*	Mutation Carrier	General Population
Duodenal cancer	4 - 12%	<1%
Thyroid cancer	~2%	<1%
Pancreatic cancer	~2%	<1%
Hepatoblastoma (liver cancer in children)	1.6%	<1%
Brain cancer	<1%	<0.6%
Stomach cancer	0.6%	<1%

* Less information is available about the risks of these cancers than about colorectal cancer.

Notes/Questions

Managing Your Risks*

Options for reducing cancer risk are available whether or not you have already had a diagnosis of cancer and/or adenomas. The following are medical management guidelines for individuals with FAP and AFAP. 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 – 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 number of polyps on previous exam)
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

*For references and supporting data on risk factors and medical management, visit www.MyriadPro.com/references

Screening for other related cancers (brain, pancreatic, hepatoblastoma, etc.) may be considered. Please speak to your healthcare provider about this option.

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 rectum may be recommended depending on the number of polyps.

CHEMOPREVENTION

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

It's a Family Affair

APC mutations are passed on in a family. Now that a mutation has been identified in you:

- Your close blood relatives (parents, children, brothers, and sisters) have a 50% chance of having the same mutation.
- More distant relatives (cousins, uncles, and aunts) also have a chance of having the mutation that runs in your family.
- Generally, the mutation is only going to be found on the side of your family (father's or mother's) that has the history of cancer and/or adenomas.

NOTE: If you have no family history of adenomas (polyps) and/or cancer, it is possible the *APC* mutation is new in you. Your parents can be tested for your mutation to confirm that this is the case. In this situation, your children have a 50% chance of having the same *APC* mutation and there is a small chance that your brothers and sisters might also have the mutation. However, your more distant relatives are not at-risk.

Your relatives can be offered Single Site COLARIS *AP*® to determine whether or not they have the same mutation.

- Relatives interested in genetic testing will need to know your specific mutation. It is best to provide your relatives with a copy of your test result which you can obtain from your healthcare provider.
- If your relative is:
  - **Positive** for the mutation, he/she has the increased cancer risks associated with FAP or AFAP and can benefit from appropriate medical management.
  - **Negative** for the mutation, he/she has an average risk of cancer and can follow general population screening guidelines.

Your healthcare provider can assist in determining which of your relatives should consider genetic testing.

Myriad has resources available to help you with your genetic test result.

- **Contact Myriad's Medical Services Department at 800-469-7423 for:**
  - Answers to questions about your test result.
  - Information about genetic testing for your relatives.
- **Or, visit Myriad's website for:**
  - A sample letter that can be sent to relatives who may need genetic testing can be found at www.MyriadTests.com/letterCOAP1Positive.
  - A healthcare provider who can offer genetic testing to relatives in any state can be found at www.MyriadTests.com/findadoctor.

If you need a copy of your genetic test result, please contact the healthcare provider who ordered your test.

Notes/Questions

Next Steps

Please work with your healthcare provider to determine the most appropriate next steps for you.

- ❑ Obtain a copy of your test result.
- ❑ Schedule consultations with appropriate healthcare providers (list below).

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- ❑ Create a plan for medical management.

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- ❑ Consider sharing your genetic test result with your relatives.
- ❑ Re-contact your healthcare provider on a regular basis for new information.

Notes/Questions

Additional Resources

Colon Cancer Alliance

A national patient advocacy organization dedicated to ending the suffering caused by colorectal cancer.
www.ccalliance.org
877-422-2030

Colorectal Cancer Coalition

A national organization whose mission is to eliminate suffering and death due to colorectal cancer.
www.c-three.org
202-244-2906

Myriad Genetic Laboratories, Inc.

www.myriadtests.com
800-4-MYRIAD (800-469-7423)
E-mail: helpmed@myriad.com

MySupport360.com

Myriad Genetic Laboratories, Inc.
320 Wakara Way
Salt Lake City, UT 84108
800-469-7423

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