Understanding Your Genetic Test Result

Positive for Two Copies of an *MYH* Mutation

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 *MYH*-Associated Polyposis

Mutations in BOTH copies of the *MYH* gene cause an adenomatous polyposis syndrome called *MYH*-associated polyposis (MAP). MAP is associated with the development of multiple adenomas (pre-cancerous polyps) in the colon and rectum, most often more than 10 adenomas over a lifetime. This syndrome also leads to an increased risk of colon/rectal (colorectal) cancer and possibly other types of cancer.

Your Genetic Test Result (check the appropriate boxes below)

THE GENETIC TEST(S) YOU RECEIVED

- ❑ Gene-Specific – *MYH* Analysis*:
  Full sequence and large rearrangement analysis of the *MYH* gene.

- ❑ Single Site *MYH* Analysis:
  Mutation-specific analysis for individuals with known *MYH* gene mutations in the family.

YOUR TEST RESULT

Positive for Two Copies of an *MYH* Mutation

**MYH* analysis may be done alone or as part of COLARIS®PLUS or COLARIS *AP*®PLUS testing.

Overview of Your Test Result

- You have mutations or alterations in both copies of your *MYH* genes.
- You have an adenomatous polyposis syndrome called *MYH*-associated polyposis (MAP).
- MAP causes people to have an increased risk of colorectal cancer, adenomas (polyps) and, in some cases, other cancers.
- The lifetime risk of developing colorectal cancer is up to 80%. Not everyone with MAP will develop cancer.

Your Cancer Risks

MYH-associated polyposis (MAP) is a syndrome that was discovered fairly recently. MAP causes an increased risk for developing colon adenomas (polyps). 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 stomach and duodenal, may be increased as well. More detailed information about cancer risks in MAP will likely be available in the future. Contact your healthcare provider on a regular basis for up-to-date information on MAP.

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 (polyps). It is recommended that you be managed according to these guidelines, depending on the number of colorectal adenomas (polyps) 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—No previous polyp (adenoma) history, 1st colonoscopy is negative	Colonoscopy	25-30 years	2-3 years
Colon—Small adenoma burden, manageable by colonoscopy	Colonoscopy	25-30 years	1-2 years
Colon—Large adenoma burden	Counseling regarding surgical options	Varies based upon adenoma burden	N/A
Colon—After colon surgery	Endoscopy of any remaining colon and rectum	After colon surgery	1-2 years
Duodenum and stomach†	Upper endoscopy and side viewing duodenoscopy	Varies	3-5 years

† Patients who have duodenal adenomas should follow FAP duodenal 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

Notes/Questions

It's a Family Affair

MYH mutations are passed on in a family. Now that these two mutations have been identified in you:

- Each of your parents is likely a "carrier" of one of your *MYH* mutations. Your parents can be tested to confirm that this is the case. The risk of colorectal cancer and adenomas in *MYH* mutation carriers is unclear but it appears that any increase in risk is small and is associated with a later age of onset (usually after age 50).
- Your biological children will all be *MYH* mutation carriers. In the rare case that your reproductive partner carries an *MYH* mutation, your children may inherit two *MYH* mutations and have MAP. Your partner can be tested to determine if he/she also carries an *MYH* mutation.
- Your brothers and sisters have a 25% chance of having the same two *MYH* mutations identified in you and a 50% chance of being *MYH* mutation carriers.
- More distant relatives (cousins, uncles, and aunts) also may carry one of the *MYH* mutations identified in you.

Your relatives can be offered Single Site *MYH* analysis to determine whether or not they have the same mutation.

- Relatives interested in genetic testing will need to know your specific mutations. 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 **two** mutations, he/she has the increased cancer risks associated with MAP and can benefit from appropriate medical management.
  - **Positive** for **one** mutation, he/she is an *MYH* mutation carrier, and may have a small increase in risk for colon cancer and adenomas (polyps).
  - **Negative** for both mutations, he/she does not have the increased cancer risks associated with MAP or *MYH* carriers

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.MyriadPro.com/ColarisFamilyLetters.
  - A healthcare provider who can offer genetic testing to relatives in any state can be found at https://www.MySupport360.com/find-provider/

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

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.

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