# Understanding Your Genetic Test Result

## Positive for a Deleterious Mutation or Suspected Deleterious

COLARIS®

**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.**

## Your Genetic Test Result (check the appropriate boxes below)

### THE GENETIC TEST(S) YOU RECEIVED

❑ **COLARIS®PLUS:**
Sequence and large rearrangement analysis of multiple genes responsible for Lynch syndrome and *MYH*-associated polyposis (MAP). See your COLARIS results report for details.*

❑ **Gene-Specific Lynch Syndrome Testing:**
Analysis of one of the genes responsible for Lynch syndrome (HNPCC). See your COLARIS results report for details.

❑ **Single Site Lynch Syndrome Testing:**
Mutation-specific analysis for individuals with a known Lynch syndrome mutation in the family.

### YOUR TEST RESULT

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

*Other combinations of genes can be tested at your healthcare provider's request.

## Overview of Your Test Result

Mutations in the gene(s) analyzed by this test are known to cause one of two hereditary cancer syndromes: Lynch syndrome – also known as hereditary nonpolyposis colorectal cancer (HNPCC) – a syndrome that involves various cancers, primarily colon/rectal (colorectal) and endometrial (uterine) or *MYH*-associated polyposis (MAP).

- You have a mutation (or alteration) in a gene that causes Lynch syndrome.
- You have Lynch syndrome.
- Lynch syndrome increases the risk of various cancers, primarily colorectal and endometrial (uterine).
- The risk of developing these cancers is less than 100%. Not everyone with Lynch syndrome will develop cancer.

## Your Cancer Risks

Having a mutation in a Lynch syndrome gene increases the risk of certain cancers. If you have already had a cancer diagnosis, you have an increased risk of developing another cancer.

| If you have **NOT** yet had cancer | Mutation Carrier | General Population |
|---|---|---|
| Colorectal cancer by age 70 | Up to 82% | 2% |
| Endometrial (uterine) cancer by age 70 | Up to 71% | 1.5% |
| Stomach cancer by age 70 | Up to 13% | <1% |
| Ovarian cancer by age 70 | Up to 12% | <1% |

| If you **HAVE** had colorectal cancer | Mutation Carrier | General Population |
|---|---|---|
| Second cancer within 10 years | 30% | 3.5% |
| Second cancer within 15 years | 50% | 5% |

Lynch syndrome mutation carriers have a slightly elevated risk over the general population of developing cancers of the kidney/urinary tract, brain, biliary tract, small bowel, pancreas, and sebaceous adenomas.

## Notes/Questions

# Managing Your Risks*

Options for reducing cancer risk are available whether or not you have already had a cancer diagnosis. The following are medical management options for individuals with Lynch syndrome (HNPCC). 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 | Colonoscopy | 20-25 years<br>(or 2-5 years prior to the earliest colorectal cancer if it is diagnosed under age 25) | 1-2 years |
| Endometrium (Uterus)/Ovaries | Gynecologic exam<br>Transvaginal ultrasound<br>Endometrial aspiration (tissue sample)<br>CA-125 | 25-35 years | 1-2 years |

Screening for other Lynch syndrome-related cancers (stomach, kidney/urinary tract, biliary tract, brain, small bowel, pancreatic) may be considered based on the presence of that cancer in a family member. Please speak to your healthcare provider about this option.

## SURGICAL MANAGEMENT

- Colectomy (removal of the colon) reduces the risk of colon cancer. The rectum is usually left in place. This is an option for:
  - patients diagnosed with colorectal cancer or advanced adenomas (polyps).
  - unaffected individuals with Lynch syndrome who are unwilling or unable to undergo regular screening colonoscopies.
- Preventive removal of the endometrium (uterus) and/or ovaries reduces the risk of endometrial and ovarian cancer. This is an option for:
  - females with Lynch syndrome who have completed childbearing.
  - females with Lynch syndrome at the time of other intra-abdominal surgery (for example: surgery for colorectal cancer).

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

## It's a Family Affair

Lynch syndrome mutations can be 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.

Your relatives can be offered Single Site COLARIS® 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 Lynch syndrome and can benefit from appropriate medical management.
  - **Negative** for the mutation, he/she has an average risk of cancer and can usually 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 additional 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.

## 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).

- ❑ Create a plan for medical management.

- ❑ Consider sharing your genetic test result with your relatives (identify your specific mutation so relatives can have Single Site testing).
- ❑ Re-contact your healthcare provider on a regular basis for new information about Lynch syndrome.

## 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

## Gynecologic Cancers Foundation

This group aims to ensure public awareness of gynecologic cancer prevention, early diagnosis, and proper treatment, as well as supports research and training related to gynecologic cancers.
www.thegcf.org
312-578-1429

## Lynch Syndrome International

The primary mission of Lynch Syndrome International (LSI) is to serve global communities by focusing on providing support for individuals afflicted with Lynch syndrome, creating public awareness of the syndrome, educating members of the general public and health care professionals and providing support for Lynch syndrome research endeavors.
www.lynchcancers.org
707-689-5089

## Myriad Genetic Laboratories, Inc.

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THIS INFORMATION IS PROVIDED TO HELP ANSWER SOME OF YOUR QUESTIONS WITH RESPECT TO CANCER RISKS, HEREDITARY CANCER RISKS, AND PRE-DISPOSITIONAL CANCER TESTING. IT IS GENERAL IN NATURE AND IS NOT INTENDED TO PROVIDE A DEFINITIVE ANALYSIS OF YOUR SPECIFIC RISK FACTORS FOR CANCER OR YOUR HEREDITARY CANCER RISKS. YOU SHOULD NOT RELY ON THE INFORMATION PROVIDED HEREIN; BUT RATHER, YOU SHOULD CONSULT WITH YOUR DOCTOR OR A QUALIFIED HEALTHCARE PROFESSIONAL TO REVIEW THIS INFORMATION ALONG WITH YOUR INDIVIDUAL HEALTH CONDITIONS AND RISK FACTORS.



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