Lynch Syndrome
PATIENT GUIDE

A Guide To Genetic Testing For
Hereditary Colorectal And Uterine Cancer

Ambry Genetics®

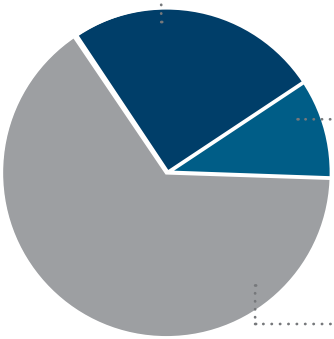
Understanding The Basics

People with Lynch syndrome may have an increased risk for cancers including:

- colorectal
- uterine
- ovarian
- other

Lynch syndrome is the most
common cause
of hereditary colorectal
and uterine cancer

CANCER FALLS INTO 1 OF 3 CATEGORIES



FAMILIAL CANCER

A pattern of the same or related types of cancer within a family that may be due to genes and/or other shared factors, such as environment and lifestyle

HEREDITARY CANCER

A pattern of the same or related types of cancer within a family due to inherited gene changes (called mutations), that can be passed from parent to child

SPORADIC CANCER

Cancers that occur by chance in one or two family members that are not due to an inherited gene mutation. Approximately 80% of all cancers are sporadic.

An estimated

1/279 - 1/440

Americans has Lynch syndrome

Lynch syndrome is caused by mutations in these genes:

MLH1, MSH2, MSH6, PMS2, and *EPCAM*

ABOUT HEREDITARY CANCER

Many people have a family history of cancer, but only **5-10% of cancer is hereditary.**

People who have these gene mutations **are born with them** – they do not develop over time.

Learning if you have an **inherited mutation** can help you know more about your cancer risks.

People with a higher chance of developing cancer may need screening, like colonoscopies, that start at **younger ages, and occur more often.**

Should You Have Genetic Testing?

IF YOU ANSWER "YES" TO ANY OF THE QUESTIONS BELOW,

genetic testing for Lynch syndrome may be something for you and/or your family members to consider.

1. Have you/your family members* been diagnosed with colorectal or uterine cancer at <50 years old?

2. Have you/your family members* been diagnosed with more than one Lynch syndrome cancer, such as colorectal and uterine cancer?

3. Have you been diagnosed with uterine and/or ovarian cancer with a family history of gastrointestinal cancers, like colorectal cancer?

4. Have multiple people on the same side of your family had colorectal, uterine, and/or other Lynch syndrome cancers?

5. Have any of your family members* been found to have Lynch syndrome?

Your healthcare provider may identify other reasons why you could consider genetic testing.

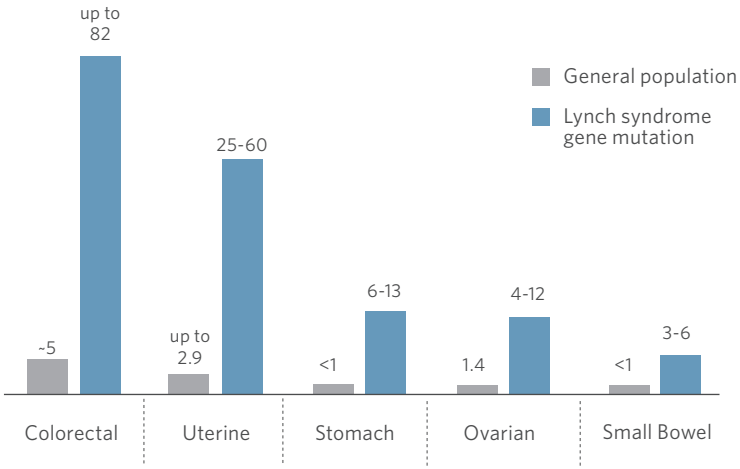
**"Family members" refers to blood relatives, such as brothers/sisters/parents/grandparents/aunts/uncles/cousins*

Understanding Disease Better Through Quality Testing

YOUR GENES CARRY A STORY THAT IS UNIQUE TO YOU AND MAKES YOU WHO YOU ARE. GENETIC TESTING CAN HELP YOU BETTER UNDERSTAND YOUR RISKS FOR CANCER.

Lynch syndrome is associated with an increased risk for multiple cancers, including colorectal, uterine, and others. Genetic testing can help determine if you and your family members may be at an increased risk for these cancers, so that you know how to best manage your health. The graph below highlights some of the cancer risks for a person with Lynch syndrome compared to someone in the general population who does not have this condition.

Lynch Syndrome Lifetime Cancer Risks (%)*



**Recent publications suggest lower risks for MSH6 and PMS2 mutation carriers*

VISIT OUR WEBSITE

See more information on which genes are included in Lynch syndrome testing: ambrygen.com/patient/cancer

How Genetic Testing Can Impact You and Your Family

FOR YOU:

Your test results may help your healthcare provider fine-tune your cancer screening plan, including the type, timing (age) of your initial screening, and its frequency.

Examples of cancer screening include mammogram, breast MRI, colonoscopy, prostate exam, dermatology (skin) exam, or other screenings indicated for your specific cancer risk.

Based on your results, your healthcare provider may review possible cancer prevention options with you, such as preventive, or prophylactic, surgery that can reduce the risk for certain cancers.

Examples include prophylactic mastectomy (removing one or both breasts before a cancer occurs) or prophylactic oophorectomy (removing the ovaries and Fallopian tubes before a cancer occurs)

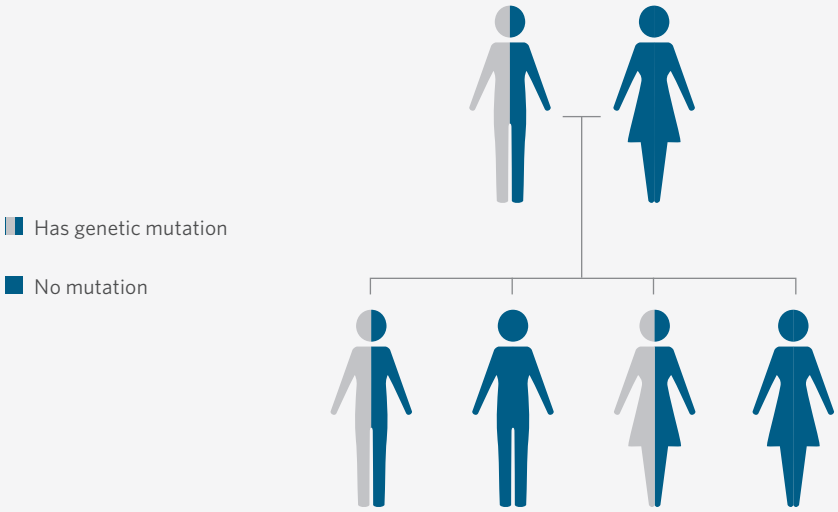
Your doctor can also identify and discuss other personalized medical management options that might be appropriate based on your genetic test results.

AMBRY GENETICS OFFERS NO-COST TESTING TO CLOSE RELATIVES

This is available for the specific genetic mutation identified in the first family member tested at Ambry within 90 days of the original report date.

FOR YOUR FAMILY MEMBERS:

If you test positive for a genetic mutation, your close family members (like your parents, brothers, sisters and children) have a 50/50 random chance of also having the same mutation.



- Men and women have the same chance of inheriting a mutation, but their chance of developing cancer may be different.

Possible Genetic Test Results

+ POSITIVE

A mutation was found in at least one of your genes tested

Detection of a cancer-related gene mutation is not a guarantee that you will develop cancer, but a warning that you are at increased risk compared to others.

Based on your results, genetic testing for certain family members may be recommended.

− NEGATIVE

No genetic mutations were found in any of your genes tested

While your genetic test results were negative, personal and family history may also be a strong indicator of cancer risk(s) and may inform your medical management.

Talk to your healthcare provider to find out if genetic testing should be considered for your family members.

? VARIANT OF UNKNOWN SIGNIFICANCE (VUS)

At least one genetic change was found, but it is unclear if this change causes an increased risk for cancer

While your genetic test results were inconclusive and do not change your medical management, personal and family history may also be a strong indicator of cancer risk(s) and may inform your care.

Talk to your healthcare provider to find out if genetic testing should be considered for your family members.

It is possible to have a combination of positive and VUS results, since multiple genes are tested.

Resources For You

Ambry Patient Education Website

ambrygen.com/patient

Colon Cancer Alliance

ccalliance.org

American Cancer Society

cancer.org

Genetic Information Nondiscrimination Act

ginahelp.org

American Society of Clinical Oncology

cancer.net

National Cancer Institute

cancer.gov

Cancercare

cancercare.org

FIND A GENETIC COUNSELOR

National Society of Genetic Counselors

nsgc.org

Canadian Association of Genetic Counsellors

cagc-accg.ca

Frequently Asked Questions

1 HOW IS GENETIC TESTING PERFORMED AND HOW LONG DOES IT TAKE?

Genetic testing requires a blood or saliva sample, which is collected using a special kit that is shipped overnight to Ambry Genetics by your healthcare provider. The testing, which looks for mutations that cause an increased risk for cancer, takes less than three weeks to complete, and results are sent to your healthcare provider.

2 WHAT WILL HAPPEN WHEN MY RESULTS ARE READY?

Your healthcare provider will receive your results; they will not be sent directly to you. Every healthcare provider may have a different method and time frame for reviewing your results with you, so it is important to discuss this process with them when your test is performed. Your healthcare provider will discuss recommended next steps based on your test results.

3 WILL MY GENETIC TEST RESULTS AFFECT MY INSURANCE COVERAGE?

In the U.S., the Genetic Information Nondiscrimination Act (2008) prohibits discrimination by health insurance companies and employers, based on genetic information. Depending on where you live in the world, you may have different (or fewer) laws in this area. Visit ginahelp.org to learn more.

4 HOW WILL MY TEST RESULTS BE PROTECTED?

We are required by law to maintain the confidentiality of your protected health information in accordance with the Health Insurance Portability and Accountability Act (HIPAA). Visit HHS.gov to learn more.

5 SHOULD I TELL MY FAMILY MEMBERS ABOUT MY GENETIC TEST RESULTS?

It is important to share your results with your family members, because they may provide additional information about their own cancer risks and management options. If you feel unsure about how to approach the subject, your healthcare provider may be able to offer some advice.

6 WILL GENETIC TESTING BE COVERED BY MY INSURANCE?

Many insurance plans cover genetic testing, and Ambry Genetics is contracted with the majority of U.S. health plans. Your out-of-pocket cost may vary based on your individual plan. A team of dedicated specialists is available to help you get access to the genetic testing you need, and provide further details about our payment options. Please call or email our Billing department at +1.949.900.5795 or billing@ambrygen.com with any questions. Visit ambrygen.com/patientbilling for more information.

7 WHAT IS AN EXPLANATION OF BENEFITS (EOB)?

Your insurance company sends you an EOB to explain any services paid on your behalf. You can contact us directly to speak with a Billing specialist with any questions or concerns about Ambry Genetics genetic testing that appears on your EOB. It is important to remember that insurance companies can take several weeks or even a couple of months to process claims.

STILL HAVE QUESTIONS?

Talk to your healthcare provider or visit our website: ambrygen.com

Elevating the Standard of Care™

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