

Lynch Syndrome (HNPCC)

A Patient's Guide

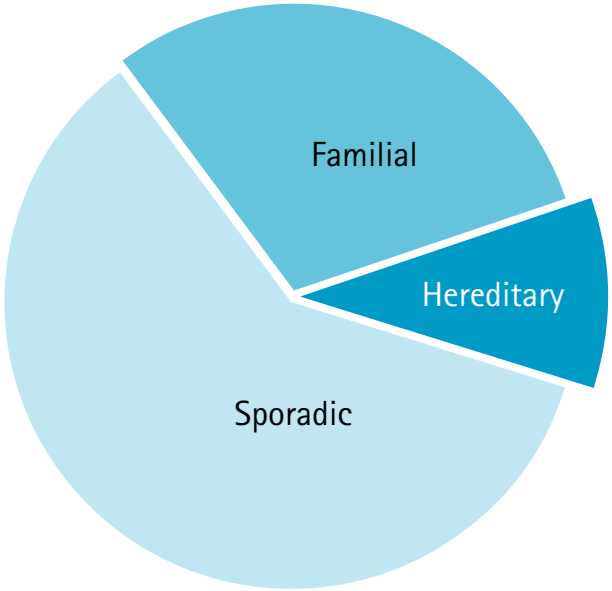
to risk assessment

Hereditary Cancer Testing: Is it Right for You?

This workbook is designed to help you decide if hereditary cancer testing is right for you and 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 (broken) gene 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:

- ☐ Colorectal cancer before age 50
- ☐ Endometrial cancer before age 50
- ☐ 2 or more Lynch syndrome cancers† at any age
- ☐ A previously identified Lynch syndrome mutation in the family

Lynch syndrome, also known as hereditary nonpolyposis colorectal cancer (HNPCC), is an inherited condition that causes an increased risk for early onset colorectal cancer (often before age 50) as well as other related cancers.† The majority of Lynch syndrome is due to a mutation in the *MLH1*, *MSH2*, *MSH6*, *PMS2*, or *EPCAM* (also known as *TACSTD1*) genes. These mutations can be inherited from either your mother or father.

*Assessment criteria based on medical society guidelines. For individual society guidelines go to www.myriadtests.com_patientguidelines

†ovarian, stomach, gastric, ureter/renal pelvis, biliary tract, small bowel, pancreas, brain, sebaceous adenomas

Cancer Risks for Lynch Syndrome Mutation Carriers

	Mutation Carrier Risk	General Population Risk
Colorectal	up to 82%	2%
Endometrial (uterine)	up to 71%	1.5%
Stomach	up to 13%	<1%
Ovarian	up to 12%	1%
Second cancer within 10 years	30%	3%
Second cancer within 15 years	50%	5%

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

Managing Lynch Syndrome Risk*

INCREASED SURVEILLANCE

Site	Procedure	Age to Begin	Repeat Test
Colon	Colonoscopy	20-25 years (or 2-5 years prior to the earliest colorectal cancer if it is diagnoses under age 25) 40 years	1-2 years Annually
Endometrium (Uterus)/ Ovaries	Gynecologic exam Transvaginal ultrasound Endometrial tissue sample 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.

SURGICAL MANAGEMENT

- Removal of the colon is often recommended in patients who develop colon cancer. The rectum is usually left in place.
- Preventive removal of the endometrium (uterus) and/or ovaries reduces the risk of endometrial and ovarian cancer and may be an option when childbearing is complete.
- Unaffected mutation carriers not willing or unable to undergo screening colonoscopies may consider removal of the colon.

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

Notes:

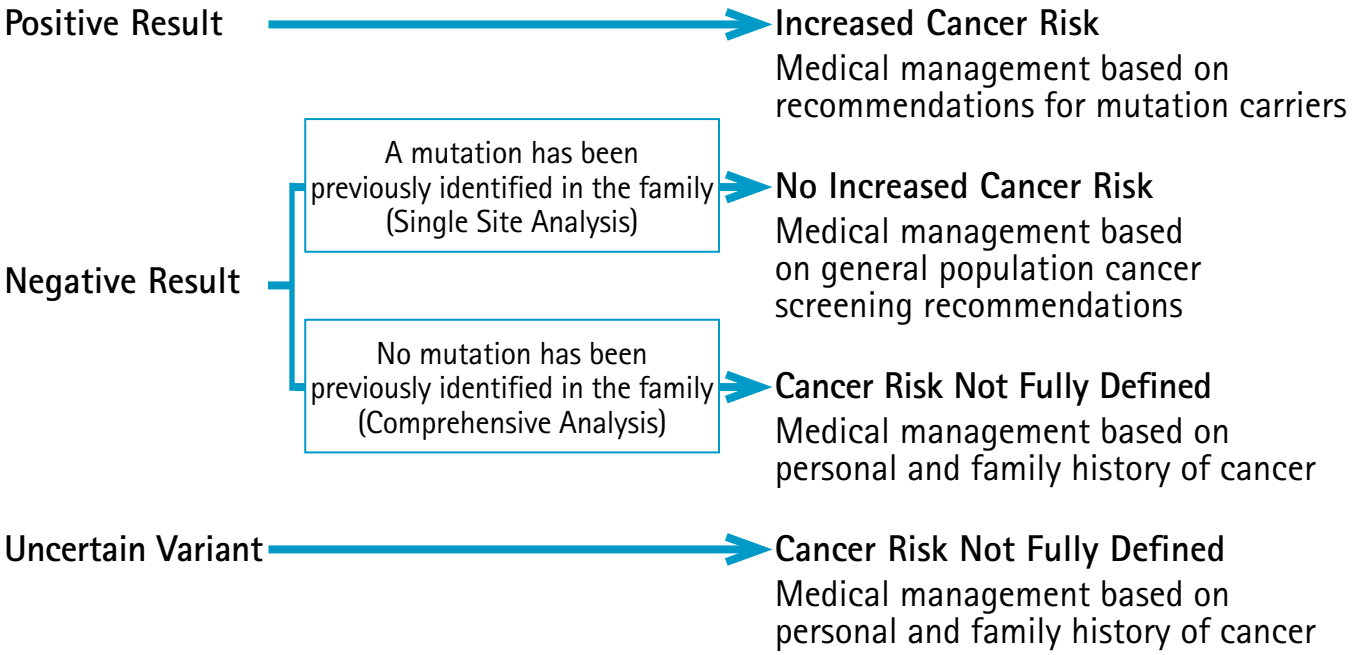
Testing Options

There are two types of tests to look for Lynch syndrome mutations:

❑ **COLARIS®:**
Sequence and large rearrangement analysis of multiple genes responsible for Lynch syndrome.

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

Possible Test Results



It's a Family Affair

- Lynch syndrome-related mutations can be passed on in a family.
  - If you have a mutation in one of these genes, your parents, your children, and your brothers and sisters have a 50% chance that they have the same mutation.
  - Other relatives may be at risk to carry the same mutation.
- Testing is the only way to accurately identify mutation carriers.
- It is important to share test results with family members.
- Individuals may differ in their viewpoints and reactions to genetic testing.

Benefits and Limitations of Testing

Benefits

- Personalized risk assessment
- Appropriate medical management to help reduce cancer risk
- Important information for family members
- Reduced anxiety and stress

Limitations

- Testing does not detect all causes of hereditary cancer
- A negative result is most helpful when there is a known mutation in the family
- Some variants are of unknown clinical significance

Notes:

Health Care Coverage

- Insurance coverage for genetic testing of at-risk patients is excellent, with the majority of patients covered for testing. Although each case is unique, the average patient pays coinsurance of less than $100.*

Privacy

- Federal laws (HIPAA and GINA) and laws in most states prohibit discrimination regarding employment eligibility, benefits, or premiums based soley 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: _____

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

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
301-879-1500

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

Myriad Genetic Laboratories, Inc.

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

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
877-422-2030

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

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COPET/03-11