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Lynch Syndrome

Lynch syndrome, also called **hereditary non-polyposis colorectal cancer (HNPCC)**, is the most commonly inherited cancer syndrome that increases a person's risk for colon cancer.

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What is Lynch syndrome?

People with Lynch syndrome are at high risk of developing colorectal cancer. These cancers are more likely to develop at earlier ages, often before the age of 50. People with Lynch syndrome are also at increased risk for other cancers, including:

- [Endometrial](#)¹ (cancer in the lining of the uterus)
- [Ovarian](#)²
- [Stomach](#)³
- [Small intestine](#)⁴
- [Pancreatic](#)⁵
- [Kidney](#)⁶
- [Brain tumors](#)⁷
- [Skin](#)⁸

What causes Lynch syndrome?

Lynch syndrome is caused by an inherited mutation in any of several genes, including:

- *MLH1*
- *MSH2*
- *MSH6*
- *PMS2*
- *EPCAM*

These are known as **mismatch repair (MMR)** genes (except for *EPCAM*, which affects the function of the *MSH2* MMR gene). The proteins encoded by MMR genes help repair damaged DNA when cells replicate themselves (DNA is the substance in our cells that makes up our genes.) However, when MMR genes become mutated, the MMR proteins are not able to work properly and are no longer able to repair damaged DNA. This can lead eventually to the development of cancer cells.

How is Lynch syndrome diagnosed?

Lynch syndrome is diagnosed when a person suspected of having it has genetic testing to look for an inherited mutation in one of MMR genes (see above).

Doctors and genetics professionals can determine if you are likely to have Lynch syndrome, based on your personal and family cancer history, using certain criteria known as the **Amsterdam criteria** and the revised **Bethesda guidelines**. These criteria are discussed in detail in [Genetic Testing, Screening, and Prevention for People with a Strong Family History of Colorectal Cancer](#)¹³. If you meet criteria that suggest that you might have Lynch syndrome, genetic counseling and testing can be done to look for mutations in the MMR genes.

For people who are diagnosed with certain types of cancer (usually colon, rectal, gastric, and endometrial cancer), the cancer cells can be tested for microsatellite instability (MSI). If a cancer cell is found to have MSI, that means that its DNA appears to be unstable. This suggests that the person's inherited MMR gene(s) may not be working properly. In this case, genetic counseling and testing can be done to look for mutations in the MMR genes to diagnose Lynch syndrome. Not all people who have cancer cells with MSI, have Lynch syndrome.

To learn about genetic counseling and testing, including how it's done, see [Understanding Genetic Testing for Cancer Risk](#)¹⁴.

Why is it important to know if you have Lynch syndrome?

People with Lynch syndrome have a high risk of developing several types of cancer during their lifetime. Some people with might even get more than one type of cancer.

There are reasons why knowing if you have Lynch syndrome might be important.

It might let you take steps to lower your cancer risk or find it early

- Ask your doctor if you should start getting cancer screenings at an early age, if you should be screened more often than normal, and if you should get certain kinds of screening tests. For example, a person known to carry a gene mutation linked to Lynch syndrome may be advised to start [screening for colorectal cancer](#)¹⁵ when they are younger (such as during their early 20s) or take other steps to try to [lower their risk of colorectal cancer](#)¹⁶.
- Women with Lynch syndrome may be advised to start [screening for endometrial cancer](#)¹⁷ or take other steps to try to lower their risk of this cancer.
- Ask your doctor if you should consider preventive surgery to lower your risk of getting certain cancers.
- Ask your doctor if there are other things you can do to lower your cancer risk, such as staying at a [healthy weight](#)¹⁸, [being active](#)¹⁹, and avoiding or limiting [alcohol](#)²⁰.

It might affect your family members

If you have a gene mutation linked to Lynch syndrome, some of your blood-related family members might have it, too. Talk to your close relatives (parents, siblings, and children) about getting tested for Lynch syndrome. They have a 50/50 chance of having the same mutation you have. If they prefer to not get tested, they might want to start screening for certain cancers early or take other precautions to lower their risk of cancer.

Hyperlinks

1. www.cancer.org/cancer/types/endometrial-cancer.html
2. www.cancer.org/cancer/types/ovarian-cancer.html
3. www.cancer.org/cancer/types/stomach-cancer.html
4. www.cancer.org/cancer/types/small-intestine-cancer.html
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19. www.cancer.org/cancer/risk-prevention/diet-physical-activity/get-active.html
20. www.cancer.org/cancer/risk-prevention/diet-physical-activity/alcohol-use-and-cancer.html

References

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Yurgelun MB, Hampel H. Recent advances in Lynch syndrome: diagnosis, treatment, and cancer prevention. *Am Soc Clin Oncol Educ Book*. 2018;38:101-109. doi: 10.1200/EDBK_208341.

National Comprehensive Cancer Network (NCCN) *Genetic/Familial High-Risk Assessment: Colorectal* V2.2023. © National Comprehensive Cancer Network, Inc. Accessed on May 17, 2024 at nccn.org.

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