



## Fact Sheet

# Lynch Syndrome

Brought to you by the *National Society of Genetic Counselors, Cancer Special Interest Group*

## What you should know about Lynch syndrome

Lynch syndrome (formerly called Hereditary Non-Polyposis Colon Cancer syndrome or HNPCC) is a genetic condition characterized by early onset colorectal cancer and an elevated risk of other cancers (see below). Individuals with Lynch syndrome have an increased risk to develop cancers at young ages and multiple primary cancers throughout their lifetime.

## Cancer Risks and Features Associated with Lynch syndrome

Type of Cancer	General Population Risk	Lynch syndrome-associated risk ( <i>MLH1</i> )	Lynch syndrome-associated risk ( <i>MSH2</i> )	Lynch syndrome-associated risk ( <i>MSH6</i> )	Lynch syndrome-associated risk ( <i>PMS2</i> )
Colon	4.5%	46-49%	43-52%	15-44%	12-20%
Endometrial	2.7%	43-57%	21-57%	17-46%	0-15%
Ovarian	1.3%	5-20%	10-38%	1-11%	Risk not well established
Gastric/Stomach	<1%	5-7%	0.2-16%	0-5%	
Small bowel	<1%	0.4-11%	1-10%	0-3%	
Bladder, Kidney, and Urinary tract	<1%	0.2-5%	2-18%	0.7-7%	
Prostate	11.6%	0-17%	30-32%	0-5%	
Brain/CNS, biliary tract, pancreas, sebaceous gland tumors, other	<1%	2- 6%	Risk not well established	Risk not well established	

## Genetics and Inheritance of Lynch syndrome

Genes are our body's instructions. They provide our body with information about how to grow and develop. When there is a mutation in a gene, it can cause the gene to no longer function correctly. Each person has two copies of every gene. One copy is inherited from their mother and the other copy is inherited from their father.

Lynch syndrome is due to a mutation in one of five genes: *MLH1*, *MSH2*, *EPCAM*, *MSH6* or *PMS2*. Cancer risks associated with Lynch syndrome are inherited in an autosomal dominant manner. This means that children, siblings and parents of individuals with Lynch syndrome have a 50% (1 in 2) chance of having the mutation as well. Lynch syndrome can be passed down through the family by both men and women.

If two individuals with a mutation in the same Lynch syndrome associated gene have a child together, there is a 25% (1 in 4) chance that the child will inherit both mutations and have a genetic condition called Constitutional Mismatch Repair Deficiency (CMMRD), which increases the risk for childhood colon cancer, lymphoma, brain tumors, and café au lait spots.

To find a genetic counselor near you, go to [www.nsgc.org](http://www.nsgc.org)



## Managing Cancer Risks

The National Comprehensive Cancer Network (NCCN) provides regularly updated recommendations and guidelines for management of individuals with Lynch syndrome. Interventions *may* include:

- Screening for colon and other gastrointestinal cancers at younger ages and more frequently than the general population
- For women, symptom awareness and screening for endometrial (uterine) cancer
- For women, consideration of hysterectomy with bilateral salpingo-oophorectomy (removal of the uterus, ovaries, and fallopian tubes) once childbearing is complete
- Consideration of additional screening guided by family history of cancer

## When to Consider Evaluation for Lynch syndrome

Features in a personal/family history that may suggest Lynch syndrome:

- Colon cancer or endometrial (uterine) cancer diagnosed before age 50
- Two separate Lynch syndrome-associated cancers in the same person\*
- A family history of Lynch syndrome-associated cancers in 3 or more family members\*
- Abnormal tumor screening results of MSI and/or IHC (tests performed on tumor samples)

\*Lynch syndrome-associated cancers include colon, endometrial, gastric, ovarian, pancreatic urinary tract, hepatobiliary tract, small intestine, brain, pancreatic and sebaceous gland tumors.

## Genetic Counseling

In many families, the cancer history may be due to a combination of genetic and environmental factors. In addition, other genetic conditions (i.e. other gene mutations) may appear clinically similar to Lynch syndrome. For this reason, a detailed review of the family history by a genetics professional is important before pursuing genetic testing. A genetic counselor can help determine which, if any, genetic tests may be helpful for a family and review the benefits, risks and limitations of genetic testing. Genetic testing is usually performed through a blood or saliva sample.

Genetic test results can be complicated and are most useful when interpreted by a genetics professional in the context of an individual's personal and family history. To locate a genetic counselor near you, please visit [www.nsgc.org](http://www.nsgc.org) and click on the 'Find a Genetic Counselor' link.

## Genetic Discrimination

The Genetic Information Nondiscrimination Act (GINA) was signed into federal law in 2008. GINA prohibits health insurers and most employers from discriminating against individuals based on genetic information (including the results of genetic tests and family history information). According to GINA, health insurance companies cannot consider genetic information to be a preexisting condition, nor can they use it to make decisions regarding coverage or rates. GINA also makes it illegal for most employers to use genetic information in making decisions about hiring, firing, promotion, or terms of employment. It is important to note that GINA does not offer protections for life insurance, disability insurance, or long-term care insurance. More information about GINA can be found by contacting a local genetic counselor or by visiting [www.ginahelp.org](http://www.ginahelp.org).

## Resources

- CCARE Lynch Syndrome – [www.fightlynch.org](http://www.fightlynch.org)
- Hereditary Colorectal Cancer Foundation - [www.hcctakesguts.org](http://www.hcctakesguts.org)

## References

- Provenzale D, et al. NCCN Clinical Practice Guidelines in Oncology® Genetic/Familial High-Risk Assessment: Colorectal. V 3.2019.
- Kohlmann W, Gruber SB. Lynch Syndrome. 2018 April 12. In: Pagon RA, et al., editors. GeneReviews® [Internet]. Seattle (WA): University of Washington, Seattle; 1993-2020.

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