



Lynch syndrome

Lynch syndrome, often called hereditary nonpolyposis colorectal cancer (HNPCC), is an inherited disorder that increases the risk of many types of cancer, particularly cancers of the colon (large intestine) and rectum, which are collectively referred to as colorectal cancer. People with Lynch syndrome also have an increased risk of cancers of the stomach, small intestine, liver, gallbladder ducts, upper urinary tract, brain, and skin. Additionally, women with this disorder have a high risk of cancer of the ovaries and lining of the uterus (the endometrium). People with Lynch syndrome may occasionally have noncancerous (benign) growths (polyps) in the colon, called colon polyps. In individuals with this disorder, colon polyps occur earlier but not in greater numbers than they do in the general population.

Frequency

In the United States, about 140,000 new cases of colorectal cancer are diagnosed each year. Approximately 3 to 5 percent of these cancers are caused by Lynch syndrome.

Causes

Variations in the *MLH1*, *MSH2*, *MSH6*, *PMS2*, or *EPCAM* gene increase the risk of developing Lynch syndrome.

The *MLH1*, *MSH2*, *MSH6*, and *PMS2* genes are involved in the repair of errors that occur when DNA is copied in preparation for cell division (a process called DNA replication). Mutations in any of these genes prevent the proper repair of DNA replication errors. As the abnormal cells continue to divide, the accumulated errors can lead to uncontrolled cell growth and possibly cancer.

Mutations in the *EPCAM* gene also lead to impaired DNA repair, although the gene is not itself involved in this process. The *EPCAM* gene lies next to the *MSH2* gene on chromosome 2; certain *EPCAM* gene mutations cause the *MSH2* gene to be turned off (inactivated), interrupting DNA repair and leading to accumulated DNA errors.

Although mutations in these genes predispose individuals to cancer, not all people who carry these mutations develop cancerous tumors.

Inheritance Pattern

Lynch syndrome cancer risk is inherited in an autosomal dominant pattern, which means one inherited copy of the altered gene in each cell is sufficient to increase cancer risk. It is important to note that people inherit an increased risk of cancer, not the disease itself. Not all people who inherit mutations in these genes will develop cancer.

Other Names for This Condition

- cancer family syndrome
- familial nonpolyposis colon cancer
- hereditary nonpolyposis colorectal cancer
- hereditary nonpolyposis colorectal neoplasms
- HNPCC

Diagnosis & Management

Genetic Testing Information

- What is genetic testing?
[/primer/testing/genetictesting](#)
- Genetic Testing Registry: Hereditary nonpolyposis colorectal cancer type 3
<https://www.ncbi.nlm.nih.gov/gtr/conditions/C1838344/>
- Genetic Testing Registry: Hereditary nonpolyposis colorectal cancer type 4
<https://www.ncbi.nlm.nih.gov/gtr/conditions/C1838333/>
- Genetic Testing Registry: Hereditary nonpolyposis colorectal cancer type 5
<https://www.ncbi.nlm.nih.gov/gtr/conditions/C1833477/>
- Genetic Testing Registry: Hereditary nonpolyposis colorectal cancer type 8
<https://www.ncbi.nlm.nih.gov/gtr/conditions/C2750471/>
- Genetic Testing Registry: Lynch syndrome
<https://www.ncbi.nlm.nih.gov/gtr/conditions/C1333990/>
- Genetic Testing Registry: Lynch syndrome I
<https://www.ncbi.nlm.nih.gov/gtr/conditions/C2936783/>
- Genetic Testing Registry: Lynch syndrome II
<https://www.ncbi.nlm.nih.gov/gtr/conditions/C1333991/>

Research Studies from ClinicalTrials.gov

- ClinicalTrials.gov
<https://clinicaltrials.gov/ct2/results?cond=%22hereditary+nonpolyposis+colorectal+cancer%22+OR+%22colorectal+neoplasms%2C+hereditary+nonpolyposis%22>

Other Diagnosis and Management Resources

- GeneReview: Lynch Syndrome
<https://www.ncbi.nlm.nih.gov/books/NBK1211>
- Genomics Education Programme (UK)
<https://www.genomicseducation.hee.nhs.uk/resources/genetic-conditions-factsheets/item/81-lynch-syndrome/>

- MedlinePlus Encyclopedia: Colon Cancer
<https://medlineplus.gov/ency/article/000262.htm>
- National Cancer Institute: Genetic Testing for Hereditary Cancer Syndromes
<https://www.cancer.gov/about-cancer/causes-prevention/genetics/genetic-testing-fact-sheet>

Additional Information & Resources

Health Information from MedlinePlus

- Encyclopedia: Colon Cancer
<https://medlineplus.gov/ency/article/000262.htm>
- Health Topic: Cancer--Living with Cancer
<https://medlineplus.gov/cancerlivingwithcancer.html>
- Health Topic: Colorectal Cancer
<https://medlineplus.gov/colorectalcancer.html>

Genetic and Rare Diseases Information Center

- Lynch syndrome
<https://rarediseases.info.nih.gov/diseases/9905/lynch-syndrome>

Additional NIH Resources

- National Cancer Institute: Colorectal Cancer
<https://www.cancer.gov/types/colorectal>
- National Cancer Institute: Genetics of Colorectal Cancer
<https://www.cancer.gov/types/colorectal/hp/colorectal-genetics-pdq>
- National Human Genome Research Institute: Learning About Colon Cancer
<https://www.genome.gov/Genetic-Disorders/Colon-Cancer>

Educational Resources

- American Cancer Society: Colon and Rectum Cancer
<https://www.cancer.org/cancer/colon-rectal-cancer.html>
- CDC: Colorectal (Colon) Cancer
<https://www.cdc.gov/cancer/colorectal/>
- Dr. Terrilea Burnett, University of Hawaii Cancer Center: Lynch Syndrome: An Explanation for Families
<https://scholarspace.manoa.hawaii.edu/bitstream/handle/10125/30728/Lynchreviewarticleforfamiliesrev.pdf>
- Genetic Science Learning Center, University of Utah
<https://learn.genetics.utah.edu/content/disorders/multifactorial/>

- MalaCards: lynch syndrome
https://www.malacards.org/card/lynch_syndrome
- Orphanet: Lynch syndrome
https://www.orpha.net/consor/cgi-bin/OC_Exp.php?Lng=EN&Expert=144
- Stanford Cancer Center
<https://stanfordhealthcare.org/medical-conditions/cancer/lynch-syndrome.html>
- Your Genome from Wellcome Genome Campus: What is Hereditary Non-Polyposis Colorectal Cancer?
<https://www.yourgenome.org/facts/what-is-hereditary-non-polyposis-colorectal-cancer>

Patient Support and Advocacy Resources

- Colon Cancer Alliance
<https://www.ccalliance.org/>
- Colon Cancer Alliance for Research and Education for Lynch Syndrome
<https://fightlynch.org/>
- Colorectal Cancer Coalition
<https://fightcolorectalcancer.org>
- Lynch Syndrome International
<https://lynchcancers.com/>

Clinical Information from GeneReviews

- Lynch Syndrome
<https://www.ncbi.nlm.nih.gov/books/NBK1211>

Scientific Articles on PubMed

- PubMed
<https://www.ncbi.nlm.nih.gov/pubmed?term=%28lynch+syndrome%5BTIAB%5D%29+AND+english%5BIa%5D+AND+human%5Bmh%5D+AND+%22last+360+days%22%5Bdp%5D>

Catalog of Genes and Diseases from OMIM

- COLORECTAL CANCER
<http://omim.org/entry/114500>
- LYNCH SYNDROME I
<http://omim.org/entry/120435>

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<https://ghr.nlm.nih.gov/condition/lynch-syndrome>

Reviewed: May 2013

Published: June 11, 2019

Lister Hill National Center for Biomedical Communications
U.S. National Library of Medicine
National Institutes of Health
Department of Health & Human Services